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Autism Spectrum Disorders: Parents, Scientists and the Interpretations of Genetic  
Knowledge

by

Jennifer S. Singh

DISSERTATION

Submitted in partial satisfaction of the requirements for the degree of

DOCTOR OF PHILOSOPHY

in

Sociology

in the

GRADUATE DIVISION

of the

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by  
Jennifer S. Singh

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## **ABSTRACT**

This dissertation is a sociological study of social and scientific representations of autism genetics. Over the last decade, there has been an increase in the awareness and prevalence of autism spectrum disorders (ASD) and efforts to identify the causes and etiology of this disorder have been unprecedented, particularly in genetics research. To address the production, representations and implications of genetic knowledge of autism, this dissertation maps out, identifies and ultimately compares the various genetic interpretations in four different autism spectrum disorder (ASD) sites, including: health social movements concerned with autism and autism genetics ASD; scientists of various disciplines who study autism genetics; parents of children diagnosed with an ASD who participate in genetics research; and individuals experiencing ASD. Based on over fifty interviews with scientists, parents, and individuals with autism, and the incorporation of grounded theory methods, this dissertation literally “follows the DNA” in order to trace the heterogeneous processes of many institutions, people, theories, materials and practices involved in the production and representation of genetic knowledge.

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## CHAPTER 1: INTRODUCTION, THEORETICAL CONSIDERATIONS, AND RESEARCH METHODS

### Glimpse Genetic Interpretations

*Genetics defines what the core of the person is. If there is a gene for Asperger's, I think that gene has an important role in what makes them a person. I don't think it should be taken out or modified or anything in that way.*

*Young adult diagnosed with Asperger syndrome*

*People are improperly addressing [it] by thinking of whether it is a question of genetics or not. I don't see why it would really make a practical difference. In terms of what actually happens if it is genetic or something else, you know, some people have it, some people don't. Some people are in between. You deal with them based on who they are, not how they got to be that way.*

*Older adult and parent of a child diagnosed with Asperger syndrome who self identifies with autism spectrum disorders*

*Genetics is just sort of how your body is built and how all of the billions of little pieces of your body were put together and a map for how you're going to grow and a map of where you are now to where you're going to go for your body physically and mentally. In terms of autism, their map is just different. How they are going to learn, the way that their body is built, how they're going to take the information is just different. But everyone is different.*

*Mother of a 5-year-old boy diagnosed with Pervasive Developmental Disorder, Not Otherwise Specified (PDD-NOS) whose family participated in a genetic research study*

*My biggest motivation for participating in a genetic research study was just the idea of being part of something that could ultimately help us better understand this disorder, for us and for everybody else. You know, this is a great mystery... I mean we're getting little signs but we still don't know. And obviously it's got to be something environmental. I mean if you're crossing, you know racial lines, wealth lines, cultures, countries, it's all over the world, there's something that we're all doing or that's happening that is affecting our children....And it's huge. And I don't think that people get how huge it is. You think about 1 in 150. One in 94 boys. That's incredible. So that's the main reason, is just the idea that we could actually do something that might help better understand where this is--why this is happening and--and ultimately help us better understand, what I think right now for most parents is how to treat it. What will and will not work. I think, unfortunately, we don't have a model. You have doctors, pediatricians who don't know anything about autism.*

*Mother of an 11-year-old boy diagnosed with autism whose family participated in a genetic research study*

*I think in the end, at the end of the day...genetic factors will probably account for most of autism. So the working model that we have now is that there are multiple genetic variants involved in autism...There are examples where mutations in a single gene will be sufficient to cause autism as a--as an entity.... And then there are going to be a whole bunch of other genes where if you get, say, a mutation in one gene, may cause the cognitive impairment component of autism and then other mutations may increase likelihood for speech and language problems or repetitive behavior or whatever it is. So it's culminations of these things coming together [that] can lead to autism. Some cases there might be an environmental component to it, too. Or an imprinting component.....So, you know, I'm sticking with genetics right now. But it's going to be in the majority cases complex combinations of genes that are contributing to it, and there seems to be a lot of genes involved.*

*Molecular geneticists and member of the Autism Genome Project*

*There might be many rare causes of autism and many of these could be cytogenetic deletions or duplications. So we've sort of coming from the, a lot of rare disorders that cause a very similar phenotype but maybe many, many different genes. And I mean with autism alone there is the whole spectrum. So even when you say autism you know you fall within Asperger's or PDDNOS or outright autism. And then you get down to autism with mental retardation and seizures and autism with dysmorphic features. So there's a--I think there's a huge spectrum in people sometimes probably generalize it too much. That sort of defines what the autism spectrum disorders are.... Using the genetic basis of their autism to define the syndrome instead of just calling them all autism is probably the way you could find what pathways are involved, what drugs might interact better.*

*Cytogeneticist and co-Investigator of the Autism Genetic Resource Exchange*

*I think scientists struggle with the complexity [of autism]. I think that everybody struggles. I think the parents struggle. I think that's part of why there is a lot of possible confusion or discussion or disagreement, because you have so many different phenotypes that are under a particular umbrella. Yeah, so I think you're talking about all neuropsychiatric disorders where, you know, the phenotyping is difficult. You want to include people but now you're including people that have basically heterogeneous causes of the disease that make it difficult for geneticists or others to try to understand the basis of it.*

*Geneticist who utilizes the AGRE database*



I begin this dissertation with glimpses of different perspectives, interpretations, approaches, and meanings of autism genetics to offer a view of the many and varied social worlds involved in the production and circulation of genetic knowledge. These quotes show the processes of transformation from subjective experiences of individuals diagnosed or self-identified with autism spectrum disorders (ASD) to the emotional knowledge of parents who participate in genetics research. The interpretations of scientists through the use of molecular genetic technologies further transform genetic knowledge into DNA sequences that explain neurological pathways, targets for treatment and intervention, and seek to associate each symptom of ASD with particular genetic codes or extra or eliminated DNA segments. These interpretations are currently at the cutting edge of the production of knowledge around autism genetics.

This dissertation is a sociological study of social and scientific representations of autism genetics. Over the last decade, there has been dramatic increase in the awareness and prevalence of autism spectrum disorders (ASD). In 2010, the Center for Disease Control and Prevention estimated that 1 in 110 children were diagnosed with an autism spectrum disorder (CDC, 2009), which is over 20 times higher than prevalence estimates in the 1970's. Public and private investments to identify the causes and etiologies of this disorder have been extraordinary, particularly in genetics research (Singh, Hallmayer, & Illes, 2007; Singh, Illes, Lazeroni, & Hallmayer, 2009). The development of autism specific genetic databases, international research consortia, and public and private funding, including the most recent \$60 million offered through the American Recovery

and Reinvestment Act of 2009,<sup>1</sup> has catapulted autism to a new frontier of genetic knowledge production.

Despite growing social and scientific investments in autism genetics, the production, representations and implications of genetic knowledge of autism are not well understood. For example, little is known regarding how individuals and families with autism influence and interpret genetic knowledge. Nor is there a good understanding of how scientists utilize genetic information and translate meaning into the etiology of autism. The representations of autism genetics through technoscientific research could have a profound influence on how autism is ‘imagined’, defined, and treated in the future.

To investigate and address these issues, the primary objectives of this dissertation are to map out, identify and ultimately compare the various processes and implications of:

- a) Health social movements of parent advocacy groups who promote genetics research on autism
- b) The construction of autism by scientist based on emerging genetic technologies
- c) The motivations, hopes and realities of parents who have child diagnosed with ASD and participate in genetics research
- d) The influence of genetic knowledge on the social identities of adults diagnosed or self-identified on the autism spectrum

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<sup>1</sup> American Recovery and Reinvestment Act of 2009 (ARRA). Retrieved November 15, 2009, from <http://grants.nih.gov/recovery/>.

Within this framework, many institutions, people, theories, materials and practices are identified, tracing the heterogeneous processes of producing, representing, and using genetic knowledge.

Thus, from a science, technology and medicine perspective, this dissertation literally “follows the DNA” and its’ many transformations starting from representations produced by families and individuals with autism. In the case of ASD, this includes participation in genetics research through donation of biological research materials and family medical histories. Many scientists utilize genetic material and family pedigree information to identify genes involved in ASD and the molecular mechanisms underlying this disorder. The filtration of scientific knowledge production to families and individuals with ASD and to other scientists’ conducting genetics research on autism creates flows of knowledge that are continually changing and transforming. As this dissertation will demonstrate, the flows of materials and knowledge also seriously engage an especially active health social movement on autism spectrum disorders.

### **Background on Autism**

He seems to be self-satisfied. He has no apparent affection when petted. He does not observe the fact that anyone comes or goes, and never seems glad to see father or mother or any playmate. He seems almost to draw into his shell and live within himself.

Leo Kanner, 1943, p.218

The understanding of autism has changed considerably since Leo Kanner first formally documented autism in 1943, where he described “early infantile autism” as a somewhat psychotic or psychiatric state (Kanner, 1943). One year later, Hans Asperger described a group of boys as having an “autistic psychopathy” form of personality

disorder, which was very similar to Kanner's descriptions, but framed as a personality trait rather than a psychotic state (Wing, 1981). Psychoanalytic thought at the time, however, pinpointed autism as a form of psychosis akin to childhood schizophrenia and a developmental anomaly ascribed exclusively to maternal emotional determinants, (i.e., the refrigerator mother theory) (Nadesan, 2005; Silverman, 2004). This ideology was promoted by Freudian child psychiatrist, Bruno Bettelheim, who attributed the cause of autism to mothering styles in his book, *The Empty Fortress: Infantile Autism and the Birth of the Self* (Bettelheim, 1967). Over time, parent advocates such as Bernard Rimland and others challenged this psychogenic theory by alternatively framing autism as a medical condition based on apparent neurological features (Rimland, 1964). Currently, autism is described as an increasingly prevalent neurological disorder with a strong genetic basis (Rutter, 2005).

Autism spectrum disorders (ASD) are diagnosed with reference to a triad of symptoms, including: communication and language impairments, social interaction deficits, and the presence of stereotyped and repetitive behaviors (APA, 2000). Prevalence estimates of ASD have increased substantially from 4 cases/10,000 in the 1970's to 27.5 cases/10,000 in 1987, and 60 cases/10,000 in 2001 (Fombonne, 2003). In 2010, the Center for Disease Control and Prevention estimated that 1 in 110 children were diagnosed with ASD (CDC, 2009), making the diagnosis of autism higher than AIDS, diabetes, and pediatric cancer combined (Autism-Speaks, 2008b).

Currently, there are no specific medical treatments for autism. However, a variety of medications and/or special diets exist that target specific symptoms of autism in children. One successful treatment has been early intervention through intense therapy,

education and behavior modification, which highlights the importance of early identification. However, this treatment approach has generated its share of skepticism among parents and professionals. Although autism was once considered to have little if any genetic in its etiology, the last few decades of family and twin studies have provided support for a strong genetic basis. Autism is now considered the most strongly genetically influenced of all multifactorial child neuropsychiatric disorders (Rutter, 2005).

Unlike single gene disorders such as cystic fibrosis and Huntington's disease where mutations in single genes can cause disease pathology, autism is a complex disorder that most likely involves many genes interacting with each other *and* with multiple environmental exposures. Furthermore, unlike diseases such as Huntington's and breast cancer, autism is diagnosed in early childhood and is accompanied by neurological challenges affecting social and cognitive behavior. Hence, the voices and representations of children with autism are often manifested through their parent advocates.

### **Social Science Research on Autism**

The social science research on autism to date has been quite impressive and diverse, ranging in different areas of study such as: the diagnosis (Grinker, 2007); autism genetics (Bumiller, 2009; Miller, Hayeems, & Bytautas, 2010; Rabeharisoa & Bourret, 2009; Silverman, 2008a; Silverman & Herbert, 2003), adults on the autism spectrum (Bagatell, 2007; Bumiller, 2008; Chamak, Bonniau, Jauney, & Cohen, 2008; Hacking, 2009; Hurlbutt, 2002; O'Neil, 2008; Orsini, 2009); stigma of parents of a child with ASD (Farrugia, 2009; Gray, 1993, 1994, 1997, 2002); social movements (Chamak, 2008), and

issues around childhood vaccinations (Kaufman, 2010). This work demonstrates the broad range of issues surrounding autism and the increased level of focus by social scientists.

Chloe Silverman addresses many of these issues in her historical and social scientific investigation on autism (Silverman, 2004, 2008a, 2008b). Her dissertation work offers a compelling historical ethnography of the role of affect in the production of citizenship through biomedical techniques and knowledge (Silverman, 2004). Using autism as a lens, she examines how affect influences different knowledge producing practices, including research, treatment, parent groups, and educational systems. Throughout her dissertation, Silverman examines the practices and discourses produced throughout the history of autism in the United States to unravel the politics of scientific research and public health, as well as the social contexts of biomedical fact production (Silverman, 2004, p. 12). Her research deals mainly with the ways in which knowledges about autism have reflected social and institutional changes and the contributions of social action to the active construction of autism (Silverman, 2004, p. 54).

Silverman also considers the construction of autism as a “genetic” disorder and the emerging collaborations between scientists and parents to create repositories for autism genetics research (e.g., Cure Autism Now and the Autism Genetic Resource Exchange). She analyzes the politics of scientific collaboration and materials sharing in a community with a diverse set of stakeholders and focuses on how genetic information is constructed as a valuable resource and utilized by citizens and stakeholders (Silverman, 2004, p. 282). Her main consideration is the extent to which parent groups “buy into” the established economies of contemporary biomedical research and the actions they take to

change academic medicine, political funding decisions, and implement the incorporation of parent knowledge into corporate research (Silverman, 2004, p. 283). In her analysis, Silverman focuses on two “contemporary parent” groups that have embraced a definition of autism as a genetic disorder (Cure Autism Now (CAN) and the National Alliance for Autism Research (NAAR)) and the specific genetic research agendas initiated by these groups --- the Autism Genetic Resource Exchange (AGRE) and the Autism Genetic Consortium (AGC), respectively.

More recently, Silverman (2008a) considered the biosociality of autism by exploring the politics and economies based on biological knowledge and social practices that work to construct and stabilize autism spectrum disorder. Here, she identifies two very different discourses of kinship based on autistic behaviors listed in the diagnosis criteria. The first course of discourse is based on likeness across individuals with autism (i.e., autistic biosociality) who view the desire for a “cure” as unethical in the sense that it denies “autistic humanity” (Silverman, 2008a, p. 47). Priorities set forth by these groups are devoted to diagnosis issues, as well as specific questions of rights, employment, treatment and services. A second discourse of kinship is based on familial tendencies, which Silverman demonstrates by discussing the work of the parent advocacy group, Cure Autism Now, (CAN) and the development of the Autism Genetic Resource Exchange (AGRE), as well as the National Alliance for Autism Research (NAAR) and the development of Autism Genome Project (AGP). Here, genetic information operates as a resource for parent advocacy organizations, and becomes “the means to repair broken families” (Silverman, 2008a, p. 43). Thus, the ideals composed of these two different biosocial groups contradict one another as one group --- those with autism ---

accepts “neurological diversity” while the other group --- composed largely of parents --- develops programs based on the genetic causation model seeking to eradicate “neurological disability” (Silverman, 2008a, p. 50).

Other important social scientific research on autism that informs this dissertation is the work that focuses on adults on the autism spectrum. For example Michael Orsini (2009) draws on the notion of “biological citizenship” to reflect on the important challenges raised by autistic citizens wanting to speak for themselves and represent autism based on lived experiences (Orsini, 2009). He argues that autistics are “using the Internet or other fora to counter what they see as avalanche of advocacy in the name of, but not for, autistic children” (Orsini, 2009, p. 183). Bridgett Chamak (2008) describes how the Autism Network International (ANI), which is considered the first and largest autistic organization run by autistics, has made a political issue of autism by redefining it as a different way of being and not a disease. Chamak argues that the labeling of autism has evolved from “a stigma to a liberation”, describing the action of autistic persons as “the latest generation of the disability movements” (Chamak, 2008, p. 90).

Several scholars have also investigated the implications of clinical and research genetics of autism (F. A. Miller, et al., 2010; Rabeharisoa & Bourret, 2009). For example, Rabeharisoa and Bourret (2009) examine the clinical work of autism genetics compared to cancer genetics. They argue that genetic mutations of autism reinforce the complexity of pathological categories by expanding and recomposing them rather than reifying and simplifying pathological situations (p. 699). Furthermore, they argue that the work in the clinic does not reflect genetic reductionism, nor does it entail a straightforward return to the previous clinical tradition. Rather, clinical practices that develop in the field of



medical genetics are producing “a new clinic corresponding to the genomic turn characterized by a syndromic and multi-factorial approach to pathologies.” (p. 709)

In an analysis of genetics research, Miller, Hayeems, & Bytautas (2010) conducted a study on the disclosure of genetics research findings to families with ASD. They revealed that parents wanted genetic research results to help them understand ‘why’ their child had autism and for some families, this information also reduced self-blame or brought peace of mind (F. A. Miller, et al., 2010). This brings up ethical issues of reporting genetic information back to participants in research studies (i.e., duty to disclose), which according to Miller and colleagues (2010) requires specific disclosure standards for different disease context. For autism, these authors found that researchers and parents set a standard of reportability that reflected the kind of meaning autism genetics research results might yield, such as explaining the cause. However, evidentiary standards within specific research disciplines (i.e., research, clinical, or statistics), as well as fundamental theories about how autism is “genetic” influenced whether or not results were deemed “true” (F. A. Miller, et al., 2010). Thus, consensus disclosure standards are unlikely to work because they do not take into consideration appropriate evidentiary standards and the status of “real time epistemological debates regarding the nature and cause of a given disorder” (F.A. Miller, et al., 2010, p.5).

The work of these scholars is relevant to my own work on genetics and autism. In my own research, I will attempt to address some lacunae. Specifically, no studies to date specifically address how genetic knowledge of autism influences the identity of individual’s diagnosed or self-identified with autism spectrum disorders or their opinions of this type of research. A unique aspect of my research is that it consists of adults who

had a diagnosis later in life, as well as those with an early childhood diagnosis. This sample is also representative of individuals on the spectrum who are not currently engaged in activism for autistic individuals. Rather, they represent a snapshot of experiences of the everyday lives of autism.

My research will also investigate the motivations, hopes and realities of parents of a child diagnosed with ASD who participate in genetics research, specifically those who donate biological materials (e.g., blood) to an autism specific research database (i.e., Simons Simplex Collection). The representations of genetics in this context may hold different meanings compared to parent advocates that promote biomedical research. This dissertation also analyzes autism genetics from the perspective of scientists involved in autism genetic research. No research to date has considered this perspective and the influence it is having on the classification and construction of ASD. My research also provides an update on the AGRE and AGP genetic research initiatives, the scientific implications (i.e., successes, limitations, challenges), and the future directions based on interviews with scientists involved in the AGP and/or who utilize the AGRE database for their own research. It will also consider some of the benefits and challenges of parent initiated genetic research programs (AGRE) versus scientist initiated genetic research programs (AGP). Finally, this dissertation considers the broader health social movements within CAN, NAAR and Autism Speaks that extend beyond research activities. The interactions between these multiple human actors and non-human actors and the boundaries they cross in the production of genetic knowledge, will offer new insights into the social scientific perspectives of autism spectrum disorder.

## **Autism Genetic Collections and Collaborations**

To situate this dissertation, I will offer a social history of the development and uses of the Autism Genetic Resource Exchange (AGRE) and the Autism Genome Project (AGP), both funded by parent advocacy groups to advance research on autism genetics. AGRE is an autism specific genetic database, initially developed by the parent advocacy group Cure Autism Now (CAN) to promote collaborative work on shared genetic samples. The AGP was initially conceived to serve as an international data-sharing collective through the collaboration of the parent advocacy group the National Alliance for Autism Research and the National Institutes of Health. Both AGRE and AGP are now part of the advocacy group Autism Speaks, serving as a collection of autism-specific databanks available to researchers transnationally and a collaboration of genetic research collectives and scientists (Silverman, 2004, 2008a).

The AGRE database was designed under the assumption that autism is a genetic disorder inherited from one generation to the next. Hence, the majority of samples consist of multiplex families (families who have two or more children diagnosed with an ASD). A more recent autism advocacy group, The Simons Foundation, has funded the development of a different genetic database called the Simons Simplex Collection (SSC). The development and use of this database operates under the assumption that “autism genes” occur spontaneously (i.e., *de-novo*) and that rare mutations occur only in the child with autism and do not exist in the parents or unaffected siblings. Thus, this collection only recruits simplex (families with one child diagnosed with autism).

## **Sites of Linkages**

The AGRE, AGP, and SSC each and all serve as sites of linkages between families and the scientists who conduct autism genetics research. For example, the availability of blood that can be transformed into computerized genetic information consisting of millions of nucleotide sequences provides scientists with the information needed to conduct large-scale genomewide association studies using microarray technologies. Likewise, the personal and “embodied” experiences of having a child with autism are captured to some degree in the phenotypic observations and parental interviews used for most genetic analysis. However the motivations for participating in genetics research on the part of families versus the motivations of scientists to conduct genetics research may be very different and, at times, contradictory. In this regard, I am concerned about the motivations of science and ethical implications of participating in genetics research for families who are seeking answers and solutions regarding how best to help their children.

The health social movements of autism that generated the AGRE database also demonstrate how familial donation of genetic material for research can influence the priorities and practices of biomedical research and create cooperation between families and researchers (Rabeharisoa, 2006; Terry & Boyd, 2001; Terry, Terry, Rauen, Uitto, & Bercovitch, 2007; A. Wexler, 1996; N. Wexler, 1992). For example, in the cases of PXE International (pseudoxanthoma elasticum) and the French Muscular Dystrophy Organization (AFM), the funding and establishment of genetic databanks by advocacy groups allowed these groups to privately hold, support, oversee and maintain their genetic databanks, as well as to determine certain direction of genetics research (Terry & Boyd,

2001; Rabeharisoa, 2006; Terry et al., 2007). As I demonstrate in this dissertation, the successes of autism parent advocacy groups in the governance of biomedical research is in accordance with their collectively shared goals – to understand the genetic underpinnings of ASD and ultimately reduce its occurrence.

For scientists, genetic representations of autism are viewed through more technical lenses that incorporate laboratory technologies such as genome-wide association studies, candidate gene studies, micro-array analysis (i.e., gene chips) and/or chromosomal mapping. These technologies help to identify the specific locations on the chromosome of potential disease alleles. Scientists' reliance on databases such as the AGRE and the SSC are apparent, as are the need to combine efforts with other scientists to generate samples sizes adequate to yield statistical genetic research results (i.e., AGP). The technologies that scientists use are also driving the direction of genetics research and the interpretations that are represented in scientific literatures. For example, the Autism Speaks website currently offers over 160 scientific papers citing the AGRE database from 2002-2010.<sup>2</sup> Likewise, the initial results of the AGP have been cited within the scientific literature over 300 times.

Genetics research is also increasingly present at national and international autism scientific meetings, which also features a wide range of research such as neurology, immunology, cognition, language and development, sensory processing, epidemiology, diagnosis, and treatment. In 2009, two keynote addresses discussed autism genetics at the International Meeting for Autism Research. One was titled, "Fulfilling the Promise of

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<sup>2</sup> Autism Speaks: AGRE collects 10,000<sup>th</sup> DNA sample. Retrieved April 21, 2010 from website: [http://www.autismspeaks.org/science/science\\_news/agre\\_milestone\\_10000.php](http://www.autismspeaks.org/science/science_news/agre_milestone_10000.php).

Molecular Medicine in Autism” and the second “Copy Number Variations (CNVs) in Autism: What do they mean?” There were also three different sessions focused on genetics and an invited educational symposium that discussed the molecular genetics of autism. Thus, the use of genetic material supplied by families for scientific research takes on new meanings in the laboratory, within scientific literatures and disciplines.

For individuals with ASD and their families, genetic information takes on different meanings usually far removed from the complex genetic interpretations produced in a laboratory. It may be represented through autism traits that “run in the family” (Richards & Ponder, 1996) or what Margaret Lock and colleagues refer to as “blended inheritance” (2006, p.282). Here, individuals and families view the blending of entities or traits from both parents as passed on in clusters from one generation to the next (Lock, Freeman, Sharples, & Lloyd, 2006). To add to these representations of inheritance of disease within families, this dissertation seeks to unravel the multiple subjectivities experienced by those living with ASD and their interpretations of genetics in the context of their daily lives and interactions with their families.

Within each of these sites, layers of transformation and processes of producing genetic knowledge of autism generate agreements, disagreements, contestations, and negotiations. These occur between and within families and individuals with ASD, within and between autism advocacy groups, among the various scientists who conduct autism genetics research and within and across their disciplines. As the flows of knowledge about autism increase, it can be anticipated that these complex relations will further elaborate. The goal of this dissertation is to map out these complex relationships to better

understand the multiple meanings of autism spectrum disorders within the production and circulation of genetics knowledge in the U.S. today.

### **Theoretical Considerations**

This dissertation theoretically engages contemporary social theory and social studies of science, technology and medicine in particular. Since the scope of my research ranges from collectivizing movements of action to individual experiences of living with autism, a range of social scientific scholarship has influenced this dissertation. I am particularly indebted to social scientific analysis that has focused on the implications of genetic technologies (Duster, 2003; Fujimura, 1996; Gibbon & Novas, 2008; L. E. Kay, 2000; Kerr, 2004; Lindee, 2005; Parthasarathy, 2007; Rabinow, 1999; Rajan, 2006; Rapp, 2000; Reardon, 2005; Shostak, 2003; Silverman, 2004; Taussig, 2009; Tutton & Corrigan, 2004; Wailoo, 1997). These ethnographies and edited volumes provided me invaluable resources. I gained much insight on health social movements from the political and historical analyses of AIDS and breast cancer activism (Epstein, 1996; Klawiter, 2008). The political, social, and cultural analysis of these health social movements, as well as those focused on genetic diseases more specifically (Heath, 1998; Kerr, Cunningham-Burley, & Amos, 1998; Novas, 2006; Rabeharisoa, 2006; Stockdale, 1999; Stockdale & Terry, 2002; Terry, et al., 2007; A. Wexler, 1996), served as important guideposts. Finally, my research has been influenced by previous social scientific work on autism (Grinker, 2007; Nadesan, 2005; Silverman, 2004). These three very recent and important ethnographies have served as a foundation for my project and offered

comparative lenses on the processes explored in this dissertation. Highlighted next are the central theoretical themes of this dissertation.

### ***Symbolic Interactionism and Social Worlds/Arenas Theory***

To address the meanings, understandings, and interpretations of autism genetics among the various actors represented, this dissertation draws upon the theoretical perspective of symbolic interactionism. According to Herbert Blumer (1969), there are three simple premises on which symbolic interactionism is based: human beings act towards things on the basis of meanings that the things have for them; meaning of things are derived from, or arises out of, the processes of social interaction between people; and the use of meanings by actors occurs through a process of interpretation. Symbolic interactionism is a theory through which social life consists of a complex fabric, woven of countless interactions through which life takes on shapes and meanings that change with time and circumstances. A symbolic interactionist (SI) perspective allows for the stimulus and exploration of many research questions surrounding the production of knowledge of autism genetics. It also provides a guide to understanding the multiple interpretations that are represented in this dissertation by focusing on the “role of the situated and negotiated order” (Clarke, 2005, p.5).

Throughout the dissertation, I draw on several insights from SI perspectives that have been applied to a wide variety of questions and projects in the study of science technology and medicine. These include: “disciplining” the tools, whereby tools shape disciplines and how disciplines shape tools to accomplish rightness (Clarke & Fujimura, 1992; Shostak, 2005). Here, tools are used as metaphors through which to better



understand scientific practice. Elements of the situation, such as laboratories, technicians, theories, genetic models, research materials (e.g., DNA and family medical histories), instruments (e.g., diagnostic tools), technologies (e.g., microarray), skills and techniques, funding organizations (e.g., NIH), advocacy groups (e.g., CAN, NAAR, Autism Speaks, Simons Foundation), audiences and consumers are all mutually articulated with the “tools”, “jobs” in the production of the “rightness” of the science (Clarke and Fujimura 1992, p.5).

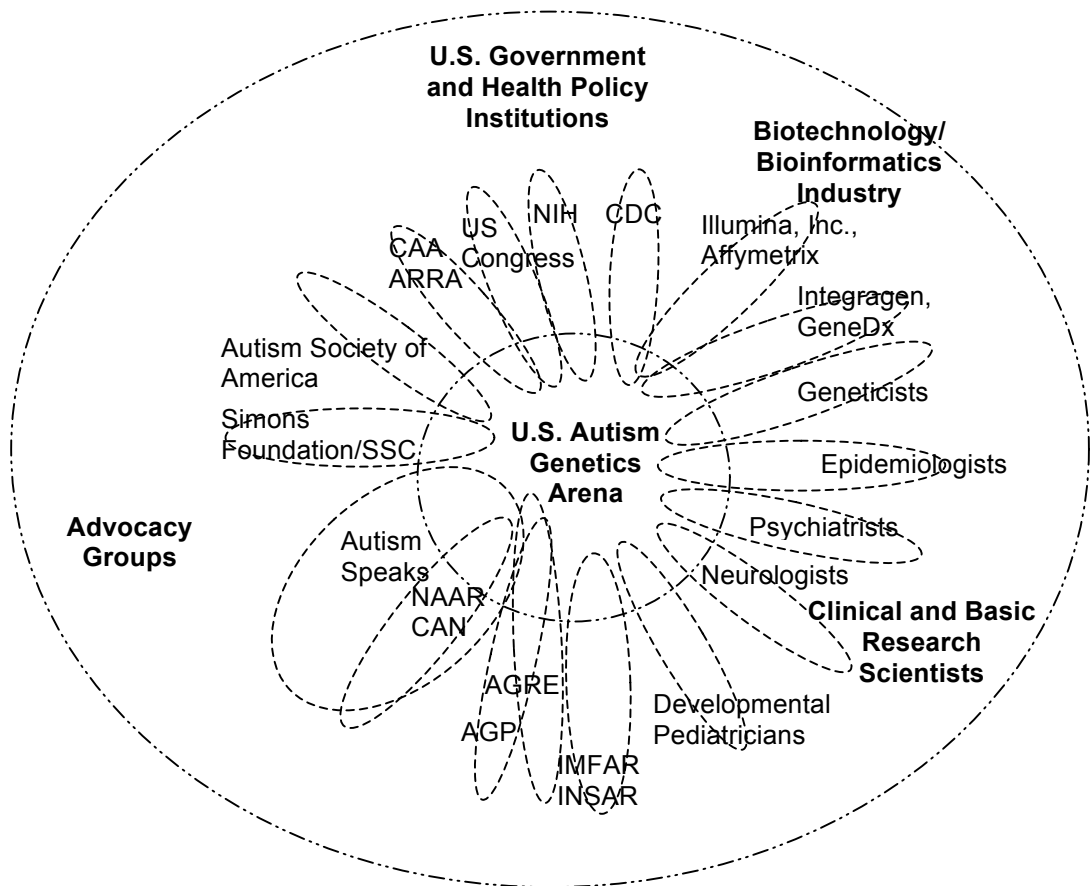
The complexity of scientific work and the multiple cascading commitments to molecular genetic research (i.e., scientific bandwagons) (Fujimura, 1996) are processes that I attend to throughout this dissertation. A scientific bandwagon exists when large numbers of people, laboratories, and organizations commit their resources to one approach to a problem because *others* are doing so *and* the technologies, skill sets, funding, and other infrastructural elements are accessible and usable (Fujimura, 2006, p. 225-227). Similarly, Sara Shostak’s analysis of the emergence of toxicogenomics demonstrates how material conditions, work practices, scientific institutions and relations of knowledge facilitated the molecularization of toxicology (Shostak, 2005). Shostak (2005) also contends that the molecularization of toxicology’s technologies, languages, practices and markets lead to the success of toxicogenomics. Following from this work, I argue that for autism genetics, commitments of parent advocacy groups, public and private investments, academic institutions, governmental organizations, emerging molecular and genetic technologies, and computer informatics drove the scientific bandwagon now so actively seeking genetic understandings of autism.

Given the range of human (i.e., parent advocacy group, scientific disciplines, and family and individuals experiencing autism), and non-human actors (AGRE, SSC) involved in the production of genetic knowledge, I started my theoretical analysis by developing a social worlds/arenas map to clarify the assumptions under-girding this project and the types of social practices considered in developing the plan of research (Clarke, 2005; Strauss, 1978). Social worlds/arenas maps lie at the meso level of social action “not at an aggregate level of individuals, but where individuals become social beings again and again through their actions of commitment and social worlds and their participation in those worlds’ activities” (Clarke, 2005, p.110). The purpose of this map is to portray the production and circulation of different social worlds, arenas, regimes of practice, social formations and discourses contributing to the production of genetic knowledge of autism.

Figure 2.1 maps the different social worlds in the U.S. autism genetics arena. More abstractly, these social worlds represent various levels of organization of actors and collectivities, including: advocacy groups instrumental in the creation of autism genetic databases, research consortia and increased awareness and funding for ASD; clinical and basic science researchers involved in genetics research on autism, ranging from geneticists, neuroscientists, to epidemiologists; segments of U.S. government agencies and health policy organizations, especially the funding support of the National Institutes of Health (NIH) and the passage of the Combating Autism Act.

The map also depicts the International Meeting for Autism Research, held annually to highlight current research on autism. This is a shifting group of basic and clinical researchers from many parts of the world. Because it meets regularly, includes

many of the same people over time, and in some senses is itself a meaningful actor, I consider it a social world in itself. Furthermore, as Garrety describes in her comparison of actor-networks and social worlds approaches on the controversies over cholesterol, dietary fat, and heart disease, such a staged intersection can impact the future of all social worlds involved in that arena and beyond (Garrety, 1997, 1998).



**Figure 2.1. Social Worlds/Arenas Map of the U.S. Autism Genetics Arena**

Acronym Legend: AGP – Autism Genome Project; AGRE – Autism Genetic Resource Exchange (genetic database); ARRA – American Recovery and Reinvestment Act; ASD – Autism Spectrum Disorders; CAA – Combating Autism Act; CAN – Cure Autism Now; CDC – Center for Disease Control and Prevention; IMFAR – International Meeting for Autism Research; INSAR – International Society for Autism Research; NAAR – National Alliance for Autism Research; NIH – National Institutes of Health; SSC – Simons Simplex Collection.

This map also emphasizes the social world of biotechnology and bioinformatics industries. These include industries that develop and sell genetic technologies used by scientists to conduct research. For example, companies such as Illumina, Affymetix, and others, provide genomic analysis tools and reagents, such as microarray technologies and computer software to translate the data. The biotechnology industry also includes new companies that are selling autism specific genetic testing panels on the Internet based on a physician recommendation. These include companies such as IntegraGen and GeneX, which provide genome-wide microarray analysis and DNA sequence analysis for individuals with autism. Although I do not specifically address these social worlds, their intersections with other social worlds are highlighted throughout the analysis.

As collectivities of human actors, social worlds may also be organized distinctively around tools and technologies (Strauss, 1978). Here, these include: the Autism Genetic Resource Exchange (AGRE), the largest autism genetic database in the world; the Autism Genome Project, consisting of both human collectives of autism genetic researchers transnationally and a collection of autism genetic databases generated internationally; and the Simons Simplex Collection (SSC), a genetic database that emphasizes a new genetics research paradigm of *de-novo* copy number variants. Social worlds also come together around genetic technologies that transform the DNA into “scientific facts” through many different processes such as genome wide association studies that utilize micro array technologies, candidate gene studies, and chromosomal mapping. The results of genetics research are also reported in scientific meetings and literature. Thus, all of these social worlds and their associated tools and technologies are potentially salient in the knowledge production of autism genetics.

In the AGRE and SSC, parents and children with ASD and their unaffected siblings are represented not through their physical body but through their genotypes and phenotypic characteristics that are banked in genomic and computer databases. These are referred to as “implicated actors”, which are not physically present in a given social world but “solely discursively constructed; they are conceived, represented, and perhaps targeted by the work of those others” (Clarke, 2005, p.46). In many regards, the AGRE and the SSC represent a boundary between human and non-human actors, where the physical representation of the ASD family is literally lost in translation of producing genetic knowledge. Another “implicated actor” that is silenced or only discursively present are individuals living with ASD. In the U.S. autism genetics arena, children and adults are often silenced. They are represented largely through parent advocates and in many regards have been reconstituted by the emerging disciplines of autism genetics. I attend to this silence by including interviews with adults diagnosed or self-identified with ASD.

The social worlds/arenas maps allow me to follow the patterns of collective commitment and salient social worlds that are involved in autism genetics research over time. There may be social worlds missing from this map revealed through the research process; however, they were not directly involved in the production of genetic knowledge (e.g., public educational systems). It is also worth noting that some of the same individual human actors participate in multiple social worlds in the autism genetics arena. In fact, this fluidity of participation is something I attend to analytically in terms of understanding relations among the different social worlds. This type of analysis will

allow me to determine which segments or subworlds come together in the production of genetic knowledge of ASD.

### ***Medicalization, Biomedicalization and Geneticization***

Theories of medicalization, biomedicalization, and geneticization also serve as frameworks and guideposts in theoretically analyzing the complex processes of producing and representing genetic knowledge of autism throughout this dissertation. In the 1970's, there was a growing dialogue among scholars about the increasing role of medicine in society, including Eliot Freidson's work on the medical professional claim to the jurisdiction over the label of illness (Freidson, 1970), as well as the work of Irving Zola, who described medicine as becoming a major institution of social control through the extension of medical jurisdiction, authority, and practice into many aspects of people's lives (Zola, 1972). This dialogue promulgated the concept of medicalization, which, since its formulation, has provided a plethora of social scientific debate around its definition, processes, and impacts it has vis-à-vis social control. In general terms, medicalization is a process that takes many different aspects of life, which may or may not be broadly construed as problematic, and defines and treats them as medical problems.

The literal meaning of medicalization "to make medical" is somewhat limiting according to sociologist Peter Conrad, who argues that medicalization has broader and more complex meanings (Conrad, 2000a). Specifically, Conrad contends that there are three distinct levels of medicalization: the conceptual, the institution, and the interactional (Conrad, 2000a). At the conceptual level, problems are defined based on a

medical vocabulary; on the institutional level, medical approaches to treating a particular problem are adopted by organizations; and at the interactional level, the physician defines a problem as medical and/or treats a “social” problem with medical treatment (Conrad, 2000a, p.106). At each of these levels physician’s roles become increasingly apparent. Conrad argues this broadens the definitional process of medicalization, since at the conceptual and institutional level, more actors are involved other than physicians in the medicalization process.

A shift from medicalization to biomedicalization occurred around 1985 due to “technoscientific” innovations that further expanded medicalization from medical control over external nature (i.e., the world around us) to transformations occurring from the inside out (i.e., capacities to change the biological processes of human and nonhuman life forms) (Clarke, Shim, Mamo, Fosket, & Fishman, 2003). Clarke and colleagues (2003) contend that the historical shift is due to technoscientific innovations occurring at the micro-level of change such as new personal identities, the meso-level change in terms of new organizational infrastructures and social forms, and the macro-level of change such as the transnational corporatization of biomedicine. These scholars define biomedicalization as “the increasingly complex, multi-sited, multidirectional processes of medicalization and reconstituted through the new social forms of highly technoscientific biomedicine.” (Clarke et al., 2003, p.161)

The major processes of biomedicalization are discernible through five interactive processes described by Clarke and colleagues (2003), including: political economics, which emphasizes the corporatization and privatization of research, products and services made possible by technoscience innovations; the focus on health itself and elaboration of

risk and surveillance biomedicines, as opposed to the medicalization focus on illness, disability, and disease; the technoscientization of biomedicine, which includes three overlapping areas: a) computerization and data banking; b) molecularization and geneticization of biomedicine and drug design; and c) medical technology design, development and distribution; transformations of biomedical knowledge production, information management, distribution, and consumption; and the transformation of bodies and the production of new individual and collective technoscientific identities (Clarke, et al., 2003).

We can view geneticization as a small part of biomedicalization, indicating that it too is a result of the manifestation (through technoscientific elaboration) of medicalization. As with the medicalization processes described above, these interactive processes are useful in grasping the emerging technoscientific processes and their implications described throughout this dissertation. As Clarke and colleagues (2009) insightfully point out, “both concepts of medicalization and biomedicalization are vital to understanding the increasing and widening impacts of genetics” (p.23).

The concept of “geneticization” emerged in the 1990’s to capture the ever growing tendency to distinguish people from one another on the basis of genetics and to define most disorders, behaviors, and even psychological variations as wholly or in part genetic in origin (Lippman, 1992, 1998). Lippman argues that geneticization obscures issues of equity and justice with respect to health and blinds these issues from the public mind (Lippman, 1998). The belief that differences among people can best be understood as primarily genetic in origin also indirectly reinforces issues of racism, social inequalities, and discrimination of various kinds (Lippman, 1992, 1998). Lippman’s



definition of geneticization emphasizes the conceptual and the processes involved in aspects of defining difference based on genetics. Thus, geneticization brings determinism to the level of our genetic makeup, which can be described as finite, unchanging and predictive of certain qualities and traits, including disease.<sup>3</sup>

Over some years, there has been considerable critique of geneticization as too categorical and too simplifying (e.g., (Hedgecoe, 1998, 2001; Rabeharisoa & Bourret, 2009; ten Have, 2001; Weiner & Martin, 2008). Thus, alternatives to this concept have been generated. For example, Hedgecoe (2001) is interested in a process of geneticization that can be used for research that reveals something about the ethical implications of genetic technologies. He developed the term “enlightened geneticization” to highlight the sophisticated way in which narratives of diseases such as schizophrenia prioritize genetic explanations, “appear” to accept environmental causation, and carefully avoid genetic ‘hype’ (Hedgecoe, 2001, p. 903). In chapter three, I draw on Hedgecoe’s concept of “enlightened geneticization” to highlight how some scientists are privileging genetic explanations for autism through various mechanisms, such as the minimalization of environmental factors contributing to autism, the consistent acknowledgement that autism is highly heritable and thus has a genetic etiology, and how, in many regards, the failure

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<sup>3</sup> Similarly, Dorothy Nelkin and Susan Lindee (2000) developed the concept of “genetic essentialism”, which they argue “reduces the self to a molecular entity, equating human beings, in all their social, historical, and moral complexity, with their genes.” (Nelkin and Lindee, 2000, p. 407). They describe the gene as a symbol, a metaphor, or a convenient way to define personhood, identity, and relationships through socially meaningful ways (Nelkin & Lindee, 2000). The concept of genetic exceptionalism refers to the idea that genetic information is qualitatively different than other forms of medical information (e.g., blood pressure, cholesterol levels, mammography results), and thus potentially more harmful with respect to discrimination, stigma and privacy concerns (Murray, 1998).

to identify genes that have major influences in causing autism are driving new genetic research models, methodologies, technologies, and demand for larger sample sizes.

Empirical work investigating the geneticization of disease reveals a complex picture that is often dependent on the particular disease and the particular study at hand. The research challenges facile use of the geneticization concept, demonstrating the density of disease construction when social, cultural and political factors are considered (Cox & Starzomski, 2004; Hall, 2005; J. Latimer, et al., 2006; Shostak, Conrad, & Horwitz, 2008; Weiner & Martin, 2008). For example, Edward Hall demonstrates that rather than a “straightforward geneticization of heart disease” there is a “contested, complex and uncertain understanding of heart disease as genetic” (Hall, 2005, p. 2673). He reveals the contested networks between key social actors (i.e., laboratory directors and geneticists) and how the meaning of geneticization becomes translated into so many different forms such that no ‘true’ or real much less final genetic answer can be identified (Hall, 2005, p. 2680). Cox and Starzomski (2004) consider the geneticization of polycystic kidney disease (PCK) through interviews with patients, family members and health care providers. They found that although PKD is one of the most common life-threatening single gene disorders, geneticization of PKD was mitigated and in some cases actively resisted. Factors that mitigated geneticization included focusing on prevention and clinical management of disease, the absence of disease-specific support groups, and provider uncertainty (i.e., Nephrology Healthcare) regarding genetic and/or hereditary aspects of PKD.

The different processes of medicalization and biomedicalization are engaged at various levels of development and understanding. Thus, the concept of geneticization

would benefit from the analytics of medicalization and biomedicalization. For example, Conrad's analysis of geneticization assumes that geneticization is an aspect of the medicalization process, which cannot take on full meaning without being 'medicalized' (Conrad, 2000b). This suggests that geneticization cannot occur independently of medicalization and that what is experienced is a form of medicalized genetics. Clarke and colleagues (2003) indicate that geneticization is a product of a larger biomedicalization process, which is described as a manifestation of medicalization through the use of technoscientific innovations (Clarke, et al., 2003). In this understanding, geneticization is not confined to the medicalization process but is rather a component of biomedicalization itself. In each case, geneticization does not exist independently, but within a larger fabric of "networked complexities" consisting of biological materials, technologies, scientists, families, advocacy groups and many others. The salience of technoscience continues to expand in this domain. The studies highlighted also provide useful examples of the complexity of disease construction when social, cultural and political factors are considered. Many of the same key social actors are considered in this dissertation including individuals with ASD, parents of ASD children, and scientists conducting autism genetics research. My own analysis also includes the work of parent advocacy groups and adults on the autism spectrum in this matrix of social interactions in the negotiations of autism as a genetic disorder.

### ***Biopower, Biopolitics, and the Technologies of the Self***

Power is situated and exercised at the level of life, the species, the race and the large-scale phenomena of population

Michel Foucault 1984, p.260

This dissertation also considers the biopolitics (Foucault, 1978; Rabinow & Rose, 2006) of the molecularization (Rose 2007; Shostak 2004) of autism spectrum disorders. Grounded in historical and genealogical analysis, Foucault describes biopower as the historical disciplining of two poles of development around which the organization of power over life is deployed, namely, the disciplines of the individual/organism bodies and the regulations of populations (Foucault, 1984). The first pole of biopower is centered on the “anatomy-politics of the human body” which is disciplining and optimizing “systems of efficient and economic controls” (Foucault, 1984, p.261). The second pole of biopower focuses on species body where the body is “imbued with the mechanisms of life and serving as the basis of the biological process: propagation, births and mortality, the level of health, life expectancy and longevity” (Foucault, 1984, p.262). This pole is centered on regulatory controls, which Foucault denotes as a “bio-politics of population.” (Foucault, 1984, p.262). Foucault (1984) argued that these two poles – the disciplines of the body and the regulations of populations – were conjoined within a series of “great technologies of power”.

Among the major types of technologies articulated by Foucault (1997), the technologies of the self have generated much discourse within science and technology studies. Foucault focuses on technologies of domination and the self, describing the technologies of the self, as that which permits “individuals to effect by their own means,

or with help of others, a certain number of operations on their own bodies and souls, thoughts, conduct, and way of being, so as to transform themselves in order to attain a certain state of happiness, purity, wisdom, perfection, or immortality” (Foucault, 1997, p.225). Technologies of power, on the other hand, objectivize the subject by determining the conduct of individuals and submitting them to certain ends or domination, which Foucault refers to as “governmentality” (Foucault, 1997). This is a theoretical concept that aims to reveal the general mechanisms of governance (Foucault, 1991). Foucault describes governmentality as a “complex form of power” created by an ensemble of “institutions, procedures, analyses and reflections” and whose target is the population (Foucault, 1991, p.102).

Drawing on Foucault’s concept of biopower, Rabinow and Rose (2006) define biopower in terms of “biopolitics”, which they regard as “the specific strategies and contestations over problematizations of collective human vitality, morbidity, and mortality, over forms of knowledge, regimes of authority, and practices of intervention that are desirable, legitimate and efficacious” (p. 3). Biopower today, Rabinow and Rose and Rose (2006) argue, must have “one or more truth discourses about the ‘vital character of living human beings, and an array of authorities considered competent to speak that truth’; “strategies for intervention upon collective existence in the name of life and health...may also be specified in terms of emergent biosocial collectivities”; and “modes of subjectification, in which individuals can be brought to work on themselves, under certain forms of authority, in relation to truth discourses by means of practices of the self, in the name of individual or collective life or health” (p.3-4). Biopower in this regard can be used as an analytic tool to study empirical changes at each of these three axes.

With regard to new forms of knowledge linked to genomics Rabinow and Rose (2006) argue a “modified bio-political rationality in relation to health is taking shape, in which knowledge, power, and subjectivity are entering into new configurations” (p. 29). This involves a variety of forces such as the investments of national governments, pharmaceutical and biotech companies in molecular and genetic technologies, as well as groups that invest hope, money, political capital, and biological tissue in the search for genetic treatments. Thus, biopolitics today, involves more than politicians, or the kinds of professions that were invented in the 19<sup>th</sup> and 20<sup>th</sup> centuries to make liberal freedom possible. They depend on meticulous work in the laboratory, in the creation of new phenomena, massive computing power, marketing powers, regulatory strategies, drug licensing bodies, bioethics commissions, and profits and shareholder value (Rose, 2007, p.28).

Carlos Novas and Nikolas Rose (2000, 2003) also expand on Foucault’s concepts of governmentality and technologies of self by considering the implication of the rise of new molecular genetics in the context of ways in which we are governed and the ways in which we govern ourselves. They describe a key event in this process as the creation of the person who is “genetically at risk”, which not only induces new and active relations to oneself and one’s future but also generates new forms of “genetic responsibility” (Novas & Rose, 2000). Rose also describes selfhood as intrinsically somatic, “where ethical practices increasingly take the body as a key site for work on the self.” (Rose, 2001, p.18). It is here, Rose argues, where biopolitics merges with a concept he describes as “‘ethopolitics’: the politics of life itself and how it should be lived.” (Rose, 2001, p.18 and 2007) Ethopolitics concerns itself with self-judgment and self-improvement and

where life itself is the object of adjudication (Rose, 2007). Our very personhood, Rose argues, is being defined “in terms of our contemporary understandings of the possibilities and limits of our corporeality.” (Rose, 2001, p.20, 2007) At the same time, our somatic individuality is subject “to choice, prudence and responsibility, to experimentation, to contestation”, what Rose describes as a “vital politics” (Rose, 2001, p.20, 2007).

Rose’s concept of ethopolitics is useful in my analysis of parents of children with who participate in genetics research. Although the parents in this study are not concerned necessarily with “self-techniques” to improve *their* health, they are concerned with techniques that they can “judge and act upon” to make their children “better than they are” (Rose, 2007). In chapter four, I argue that the responsibility and novel forms of authority parents take upon themselves to help their children and families in the future is an emergent form of biological ethopolitics.

This literature generated from theoretical writings of Foucault on biopower, biopolitics and technologies of the self has offered theoretical signposts for this dissertation. In an era where science is getting closer to identifying all forms of genomic variation through whole genome sequencing, the “molecularization” of disease will become magnified to the point of single base pairs existing throughout the human genome that can be “identified, isolated, manipulated, mobilized, recombined, in new tactics of intervention” (Rose, 2007, p. 6). In chapter three, I argue that the increased surveillance due to new genetic knowledges and monitoring of new genetic variants in populations of people with ASD constitute a form of biopower. Furthermore, the emerging technological advances of microarray analysis in clinical genetics to diagnose autism have lead to a level of social control, especially vis-à-vis parents of children with

ASD. Parents are advised by clinical geneticists to constantly check back and actively monitor their child's genetic mutation status and are continuously subject to new possibilities of risk and surveillance due to the "discoveries" of interconnected molecular pathways that converge on common biological pathways (e.g., autism and cancer).

### ***Health Social Movements, Biosociality, and Genetic Citizenship***

This dissertation also takes into consideration the theoretical frameworks of health social movements (Brown & Zavestoski, 2004; Brown, et al., 2004; Rabeharisoa, 2003) and draws on the theoretical concepts of biological and genetic citizenship (Heath, Rapp, & Taussig, 2004; Novas & Rose, 2000; Rose, 2007; Rose & Novas, 2003). Health social movements (HSMs) have been described as "collective challenges to medical policy, public health policy and politics, belief systems, research and practice, which include an array of formal and informal organizations, supporters, networks of cooperation and media" (Brown & Zavestoski, 2004, p. 679). In chapter two, I identify the processes and strategies that parent advocates of autism engage in their efforts to unravel the biomedical basis of autism spectrum disorders, as well as the broader agendas of parent advocacy groups Cure Autism Now (CAN), National Alliance for Autism Research (NAAR), now merged into Autism Speaks.

Among the theoretical models of HSMs proposed by Brown and Zavestoski (2004) is the "embodied health movement", which recognizes how illness experiences challenge science on etiology, diagnosis and/or prevention. An expansion of the "embodied health movement" also highlights the collaboration of activists with scientists and health professionals in pursuing treatment, prevention, research and expanded



funding (Brown, et al., 2004). In chapter two I discuss how the health social movements advancing biological understanding of autism did not emerge from the individual experiences of people with autism. Rather, the movement emerged from the emotional experiences of parents of children diagnosed with autism and their motivation to change the direction of autism research towards enhanced funding, awareness, and acceleration of biomedical research to prevent, treat and cure autism. At the time, autism research was poorly funded in both the public and private sectors and only a handful of investigators were seriously focusing on the disorder

The focus of parent advocates on the production of genetic knowledge also took action by developing strong collaboration efforts described within the embodied health movement. In chapter two, I describe the development of the AGRE database and the AGP, efforts that required collaboration among parents, clinical and basic researchers, and government agencies. I argue that these genomic “tools” strongly support basic and clinical researchers and serve as two major genetic knowledge producing enterprises.

The “partnership model” proposed by Rabeharisoa (2003) is also used in this dissertation to help articulate the collaboration efforts and shifts in the balance of power among parents and scientists. In the “partnership model”, Rabeharisoa characterizes the patient organization as the master of its research policy and patients as specialists in their own right (Rabeharisoa, 2003). The first characteristic demonstrates a reversal of traditional power relations between patient organizations’ board of governors and its scientific council, where the former is in total control of its research policy and the latter is an advisory body whose opinions are subject to approval by the board. The second characteristic places patients and their families as specialist partners in the production of

knowledge and in the care and treatment of their disease (Rabeharisoa, 2003). In chapter two, I demonstrate how the reversal of power for CAN and NAAR was apparent since these organizations redirected autism research to consider the biological origins of autism. I argue, however, that differential stakeholder power emerged in these two genetic research initiatives. For CAN/AGRE, the power was clearly group initiated and remains within the parent organization itself. In contrast, the AGP comprises a mixture of power relations among the scientists themselves and with the parent organization. I also discuss how these fundamental differences in power reflect the different trajectories and outcomes of these projects.

Social science scholars have also described actions like those of CAN and NAAR as active forms of citizenship (Heath, et al., 2004; Novas & Rose, 2000; Rose, 2007; Rose & Novas, 2003). For example, Rose and Novas (2000, 2003) describe this active participation in the production of genetic knowledge in their concept of “biological citizenship”. Based on their study of Huntington’s disease, they describe biological citizenship as generating new active consumer-like citizens, who govern themselves through self education and self-management of disease (Novas & Rose, 2000; Rose & Novas, 2003). As Rose points out, biological citizenship is both individualizing and collectivizing. Individualizing is to the extent that “individuals shape their relations with themselves in terms of a knowledge of their somatic individuality...where somatic individuals must also know and manage the implications of one’s own genome.” (Rose, 2007, p.134)

The collectivizing movements of biological citizenship are organized around specific biomedical classifications and often involve specialized scientific and medical

knowledge of the condition at issue (Rose, 2007). Rose articulates the collective biosocial citizenship by drawing on Paul Rabinow's (1992) concept of biosociality, which characterizes these forms of collectivization as organized around the commonality of shared somatic or genetic status (Rose, 2007). Rabinow theorizes that arising out of the new truths generated by the new genetics will be the formation of new social groups based on individual identities and practices, where conceivably groups will be formed around specific genetic mutations (Rabinow, 1992). Rabinow and Rose describe this as "strategies for intervention upon collective existence in the name of life and health" (Rabinow & Rose, 2006, p.197), which are now being specified in terms of emergent biosocial collectivities based on specific diseases, as in the emerging forms of genetic or biological citizenship (Rabinow & Rose, 2006). In chapter two I describe how the belief among parent advocates that autism had a heritable component collectively activated them to promote research on the genetic etiology of autism hoping that a "cure" might emerge. Furthermore, in chapter three, I utilize these theoretical concepts to describe how collectivities were formed based on specific copy number variant (CNV) mutations.

Within the broader concept of biological citizenship, which encompasses citizenship projects linking conceptions of citizens to beliefs about the biology of human, lies the concept of genetic citizenship (Rose, 2007). Genetic citizenship is described as a complex and multi-sited network of associations that link lay health activist, clinicians, scientist, politicians, and corporate interests in the collective formation of the public sphere (Heath, Rapp, and Taussig, 2004, p.154). It also represents a diverse array of nonhuman actors, such as genes and molecules implicated in particular diseases and the technologies used to study them. By forging these alliances, Heath and colleagues (2004)

argue that genetic advocacy groups are “making citizenship claims on behalf of their genetically vulnerable offspring” (p.155). I utilize the concept of genetic citizenship to show how parent advocates of CAN and NAAR and now Autism Speaks established networks of associations among clinical and basic researchers, policy makers, governmental agencies, and families of children with autism. The non-human actors, such as the AGRE, the collection of DNA and phenotypic samples combined through the AGP, and various emerging technologies such as microarrays, are also closely linked to these networks of associations.

### ***STS and the Construction of Genetic Disease***

Over the last fifteen years, there has been an expansion in the production of scientific knowledge and technologies from human genetics research. This research has been highlighted through knowledge producing events such as the sequencing of the human genome and the cloning of various genes that “cause” diseases. Science and technology studies (STS) focused on genetic science and technologies have produced thoughtful analyses and critiques of the production of genetic knowledge and how assemblages of human and non-human actors produce new individual technoscientific identities, shape society, and politics. As Charis Thompson points out, some STS scholars investigate scientific and technical knowledge, while others focus more on ontology, technology, or science as practice (Thompson, 2005).

Compared to social science research on the Human Genome Project or the social implications of genetic testing, STS studies have paid less attention to the social construction of “genetic” diseases and the development of genetic disease categories.

How genes and mutations are identified, the development of disease etiologies, the implementation of genetic tests, and the integration of genetics research into clinical practice are all areas of research that would benefit from critical STS analysis. Those scholars who have embarked in this area of study demonstrate the political, social, cultural and etiological complexity of diseases marked as genetic. Furthermore, as Conrad and others demonstrate, the use of genetics to define human conditions moves beyond diseases that have clear biological components to address behaviors, traits, or conditions society has defined as deviant (Conrad, 2000b).

The social construction of cystic fibrosis (CF) has been a site of intense STS research since the cloning of the cystic fibrosis gene and the identification of alleles thought to be involved in the etiology of CF (Hedgecoe, 2003; Kerr, 2000, 2005; Miller, Ahern, Ogilvie, Giacomini, & Schwartz, 2005; Miller, Begbie, Giacomini, Ahern, & Harvey, 2006). This work serves as a useful framework for analyzing the social construction of ASD. For example, Anne Kerr has explored the definition and diagnosis of CF from 1948-2005 to identify the ways in which different techniques, ideas, and relationships have shaped conceptions of CF as a genetic disease, among other terms (i.e., classic CF, heterogeneous CF) (Kerr, 2005). She demonstrates that although the discovery of the CF gene and associated mutations did not raise the possibility of a new definition of classic CF, the terms of CF had to be renegotiated in two ways. The first was the incorporation of genetic evidence for CF into the diagnostic process, where genetic mutations became one part of the evidence for CF (Kerr, 2005, p. 888). Second, additional features became prominent in the definition of classical CF due to genotyping such as male infertility. As Kerr carefully points out, “CF is a dynamic entity, whose

various meanings and interpretations are shaped by a range of social and material actors, including professionals, patients, technologies and bodies” (Kerr, 2005, p. 890). Thus, recent genetic approaches to defining, diagnosing and screening CF are not radically different from past practices, do not clarify or confound the nature of disease, and remains open to multiple interpretations (Kerr, 2005, p. 892).

Similarly, Miller and colleagues contends that genes for CF, as well as other disease categories (e.g., Huntington’s disease and tuberous sclerosis) do not always and uniformly define the parameters of the disease. Other factors are involved, such as technical capacity, professional identity, and institutional organization, all of which must contend with etiologic knowledge in the production of disease categories and classification systems (Miller, et al., 2005; Miller, et al., 2006)

Adam Hedgecoe also investigates how genetic explanations play a role in the reclassification of Cystic Fibrosis (Hedgecoe, 2003). Like Kerr, he investigates the production of medical knowledge to understand how the disease category of cystic fibrosis has been re-constructed along genetic lines to incorporate related, but separate, conditions such as male infertility (Hedgecoe, 2003, p. 51). Hedgecoe delivers this analysis by focusing on how classification systems are represented in medical/scientific texts. He draws on the work of Bowker and Star (1999) and their argument that classifications are shaped by contingent and social concerns, while also recognizing their material aspects. In his analysis, Hedgecoe identifies several “discursive mechanics of knowledge production” to support his thesis, such as the interchangeable use of CF gene with the CFTR (Cystic Fibrosis Transmembrane Conductance Regulator) gene, and how in the graphic form of the classification system used in reviews and presentations it

appears as if *all* cases of male infertility are associated with the CFTR mutations (Hedgecoe, 2003, p. 57). He concludes by stating that disease classifications are socially constructed and how “the test for the CFTR gene did not remove social decisions from the classification system, it highlighted them” (Hedgecoe, 2003, p. 64).

In sum this work focuses on the production of knowledge and insists on the constructed nature of scientific truth. These authors consider the agency of both human and non-human actors in the production of “objective” scientific facts. Kerr’s investigation of different techniques, ideas and relationships that shape the conception of CF as a genetic disease will be a particularly useful for this dissertation. I will draw on her work in identifying the knowledge making processes of defining autism as a genetic disease especially with regard to the use of genetic mutations (or copy number variants) in the diagnosis of autism, and whether significant symptoms of autism become prominent in the diagnosis based on genetic knowledge. Hedgecoe’s (2003) concept of “discursive mechanics” will also serve as a guide in analyzing how genes for autism are described with reference to symptoms of ASD and how visual representations of the physical mutation are used in scientific presentations and publications.

Throughout the dissertation, I also draw on various conceptual theories developed within science and technology studies (STS). These ideas and writings have been useful throughout my research process. For example, I employ Merton’s (1973) scientific norms of communism, universalism, disinterestedness, and organized skepticism to describe how scientists altered their scientific practices to reflect the rules associated with access to AGRE or participation in AGP. The development of public databases like the AGRE and collaborative efforts like the AGP required scientists to pull their samples and share

unpublished data with the scientific community, which is counter to the usual scientific competitiveness. At various places throughout the dissertation I also rely on theoretical concepts drawn from “science in action” such as “obligatory passage points” (Latour, 1987). For example, the AGRE database has been transformed into a working tool that scientists use to generate knowledge. In a sense, it has created an “obligatory passage point”, where in the start, scientists were skeptical of parents and their ability to create a quality database (Latour, 1987).<sup>4</sup> Now many scientists described it as “indispensable” rendering the “passage” of using the AGRE an obligation in order to conduct autism genetics research.

To conceptualize the boundaries of social worlds that come together in the production of genetic knowledge on autism, I also draw on work that theorizes, boundary objects (Star & Greisemer, 1989) and local contingencies, uncertainties, differences, and processes that are often “lost in translation” during the production of genetic knowledge (Fujimura, 2005; Fujimura & Fortun, 1996). Following from Fujimura, translation in this sense “can distort, transform, delete, and add.” (Fujimura, 2005, p. 220). Inspired by these theoretical concepts, I elucidate what I refer to as the “social and scientific transcriptions and translations of DNA”. By reversing and distorting the scientific dogma of “RNA > DNA > protein > organ > systems > organisms”, I attempt to sociologically unpack the intermediary processes of transcription and translation to reveal social and scientific processes described throughout this dissertation.

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<sup>4</sup> Chloe Silverman also makes this point in her research on CAN and the AGRE database (Silverman, 2004, 2008a).



## **Research Methods, Data Sources, Analysis**

### ***Methods***

This dissertation is a multi-sited ethnographic investigation and interview-based study of four sites associated with knowledge production and understanding of autism genetics: individuals experiencing ASD; parents of children diagnosed with ASD who have participated in a genetics research study; scientists of various disciplines who study autism genetics; and the health social movement around autism and autism genetics. It draws on data collected from 2005 – 2010.<sup>5</sup> Multi-sited ethnography is appropriate for the scope of this dissertation research because it places no clear boundaries to the unit or object of analysis, focusing instead on connections and associations rather than a particular place (Marcus, 1995, 1998; Rapp, 1999). This allows for more complex objects of study and “unexpected trajectories in tracing a cultural formation across and within multiple sites of activity” (Marcus, 1995, p.3). Thus, this approach enabled me to follow the trajectories of the technologies, concepts, people, institutions, research funding, and bordering social worlds that have been involved in the production, representation and implications of autism genetics.

This approach also allowed me to move beyond personal experiences, which in my case were the experiences of working with recombinant DNA technology. I was able

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<sup>5</sup> From November 2005 through October 2007, I conducted interviews with adults on the autism spectrum as part of my qualitative training at UCSF. This particular aspect of my research was supported by the Center for Integration of Research on Genetics and Ethics at the Stanford Center for Biomedical Ethics. From October 2007 – February 2009, I conducted background research, wrote my dissertation proposal and applied for grant support for my research. I also published two articles on the historical milestones of autism research and trends in autism funding research (Singh, et al., 2007; Singh, et al., 2009). In February 2009, my dissertation proposal was approved and the UCSF Committee on Human Subjects approved my research protocol as # H6577-32483-01. From April 2009 – October 2009, I conducted my primary data collection.

to utilize my genetic expertise to translate scientific literature of autism genetics and be actively present in scientific meetings. At times, I was a parent empathizing with other parents who only wanted the best for their children. Other times I was a concerned parent about the neurotoxins in our environment (e.g., DAN! Conference). Many times, I was an observer and a curious spectator at national parent advocacy group meetings, support group meetings and fundraising events. In the beginning, I was a student wanting to learn about autism from people experiencing it everyday. By expanding my empirical research into many social worlds, I was better able to identify where some of the “force fields” of autism genetics meet and resonate (Rapp, 1999, p.119).

### ***Interviews***

The primary mode of data collection for this dissertation was in-depth qualitative interviews. A total of 58 interviews were conducted from November 2005 – October 2009. The first set of interviews (n=18) was conducted during my qualitative course training at UCSF.<sup>5</sup> This consisted of a convenience sample based on referrals from professionals who work with individuals on the autism spectrum, including a child psychiatrist, a psychoanalyst, and a counselor of transitioning teens with neurocognitive disabilities. The inclusion criteria were: a diagnosis or self-identification with high functioning autism or Asperger syndrome; 18 years or older; and the capacity to consent to a research study, which was determined by the referring specialist. A total of 18 participants were interviewed, including: four males and one female who self identified as having ASD, ages 40-50 years old, all of whom had a child with an ASD diagnosis; 10 males with a childhood diagnosis of ASD, age 18 – 25 years old; and three females with a

diagnosis of ASD, age 18 – 33 years. Although all the participants experienced ASD in their lives, multiple perspectives were apparent, including: living with or without a diagnosis of ASD; the age at diagnosis; two age cohorts (i.e., age 17-30 to > 50 years old); having a child with autism; and gender. All the participants were Caucasian.

Another element of this sample that influences the results are the interviews I conducted in multiple generations for three families. Here, I interviewed the parent who self identifies with ASD and their child diagnosed with an ASD. All interviews were held separately, audio taped, transcribed, and coded. Face-to-face interviews, either at the participant's home, school, or work office, were conducted for all but one participant who was interviewed over the telephone. The participants were asked to describe ASD in their own words, where or from whom they gained understanding of ASD, their opinion about the diagnosis of ASD, and their awareness, understanding and opinions of research on the genetics of ASD. The second major means of data collection was through participant observation that played a central part in the interpretation and analysis of the interviews. During the interviews, particular attention was paid to non-verbal cues and social interaction processes that were taking place or in several cases, that were not taking place. These observations were documented through field notes systematically written after each interview.

A second set of interviews was with scientists (n=19) who were either members of the Autism Genome Project (AGP) and/or who utilized the Autism Genetic Resource Exchange (AGRE) database. Potential participants were identified through publicly available information on AGP members and through literature reviews of autism genetics research based on the keyword "Autism Genetic Resource Exchange." These scientists

were all working in university-based scientific research. The majority of scientists interviewed were in the field of genetics (n=11), followed by psychology (n=5), neurobiology (n=2) and epidemiology (n=1). The years of experience within their discipline ranged from 9-40 years (average 21 years) and the number of years specific to autism research ranged from 4-28 years (average 11 years).

The scientists were asked to describe current and future goals of their research, their contributions to the field, and their impression of the current state of autism genetics research. Scientists who participated in the AGP were asked specific questions about the history and current status of the AGP. Scientists who utilize the AGRE were asked to comment on how they specifically utilized this resource. The majority of interviews were conducted over the phone (n=16) due to the geographic distribution of scientists. All interviews were audio taped, transcribed, and coded.

I also interviewed coordinators of the AGRE and AGP (n=4) who were themselves scientists but were representatives of the parent advocacy groups (i.e., CAN and NAAR, which is now Autism Speaks). Another interview was also conducted with a study coordinator of the Simons Simplex Collection (SSC), a new project collecting genetic and phenotypic data from families with one child diagnosed with autism. These interviews were less formal than those with scientists and were conducted to gather background information about these different projects. Field notes were coded for these interviews and analyzed as primary data.

A third set of interviews was with parents whose families participated in the Simons Simplex Collection (SSC) (n=16). Respondents were identified through a flyer sent from the coordinator of the SSC. The majority of interviews were conducted face-to-

face at either the parent's home (n=10) or another location (n=4), and 2 interviews were conducted over the telephone. The parents were asked questions about their experiences with autism and their motivations for participating in a genetic research study. There were also asked to describe their understanding of genetics based on their experiences with autism and how they would utilize genetic information if it were available. All interviews were audio taped, transcribed, and coded.

### ***Participant Observation***

Participant observation was the second major source of primary data for this analysis. I conducted participant observation at a variety of scientific conferences, meetings and symposia. These included the following:

- a) The International Meeting for Autism Research: May 3-5, 2007 (Seattle, WA); May 15-17, 2008 (London, England); and May 7-9, 2009 (Chicago, IL)
- b) Autism Society for America Conference 2008 and 2009 (Suwannee, GA)
- c) Defeat Autism Now! Conference, April 2009 (Atlanta, GA)
- d) Stanford Autism Symposium, May 2006 (Palo Alto, CA)

I also attended a variety of public symposia and lectures, including a lecture given by Dr. Temple Grandin, an adult with autism who has written extensively about her experiences living with autism, as well as Stephen Shore, who is also diagnosed with autism and has written about the importance of self-advocacy for adults on the spectrum. I attended grand rounds and seminars at Emory University given through the Department of Human Genetics focused on autism genetics. Finally, I attended numerous parent support group meetings offered through the Marcus Institute, in Atlanta, GA, for families with children

on the autism spectrum. My field notes from all of these forums have been coded and analyzed as primary data for this dissertation.

### ***Scientific Literature Review and Document Analysis***

As supplementary material to this analysis, literature generated from scientific peer reviewed papers and other media, such as parent advocacy newsletters, pamphlets, and brochures were also part of the data collection. First, I read and analyzed scientific reviews on autism genetics that were identified by searching PubMed with the keyword/phrase “autism” and “genetic” from 2008-2009. This helped me to establish the current state of autism genetics research and the key players in producing knowledge of autism genetics. I also read and analyzed scientific peer-reviewed papers that utilized the AGRE database. I limited this analysis to the scientists I interviewed to help me understand the type of research they were conducting and how they utilized the AGRE.

All media sources pertaining to autism genetics, as well as health social movements of autism more broadly, were also reviewed for this analysis. These sources were publically available from CAN, NAAR, and Autism Speaks, including: weekly newsletters published on-line from 2005-2008; the Autism Speaks UTube website; recruiting materials (as well as newsletters) used by advocacy groups for donation of biological research materials for the AGRE and SSC; and publicly available resources on the AGRE, AGP and SSC (e.g., reports, briefs, fact sheets, etc. produced by NAAR, CAN, Autism Speaks, and Simons Foundation).

Collectively, these sources helped me to identify the people, places, technologies, and practices involved in the knowledge production of autism genetics. They also helped

to distinguish the relationships parent advocates of CAN, NAAR and Autism Speaks have with scientists and government officials. In many regards, they constituted an historical map tracing the social structures, relations, tools, and processes that have been part of the emergence of autism genetics. Thus, these data analyses helped to identify the negotiated social relationships, the production of identities and subjectivities, and the production of power/knowledge, ideologies, and social control produced within and around autism genetics (Clarke, 2005; Jaworski & Coupland, 1999).

### *Analysis*

This research project utilized and incorporated the theory/methods package of symbolic interactionism and grounded theory methods (Strauss, 1987; Clarke, 2005). Symbolic interactionism centers on situated interpretive actors and the negotiated nature of the social order (Clarke, 2005; Strauss, 1987). Grounded theory methods consist of systematic abductive (Reichertz, 2007) guidelines for gathering, synthesizing, analyzing, and conceptualizing qualitative data to construct theory. Given my central question – what people and processes are involved in the production and representations of genetic knowledge around autism spectrum disorders (ASD) – grounded theory is especially advantageous for the development of more refined ideas about lay and scientific understandings and implications of the genetics of ASD.

Using grounded theory grounded in interactionism, the analysis of this dissertation started with open coding---unrestricted coding of the field notes based on observations, interview transcripts, and all other textual data to help produce codes and then categories that capture the data more conceptually (Strauss, 1987). Due to the large

volume of data generated, I incorporated the processes of “constant comparative method” (Glaser & Strauss, 1967), “focused coding” (Charmaz, 2006), and “selective coding” (Strauss, 1987) to help synthesize and explain larger segments of data. This coding took into consideration the relevance of the research questions and was referenced by a given category for the following conditions: interaction among the actors, strategies and tactics, and consequences (Strauss, 1987). The conceptual categories I developed explicate events, incidents, actions, and/or social processes in the data (Strauss & Corbin, 1990). A key element of the analysis was writing detailed memos on all conceptual categories to elaborate the processes, assumptions, interpretations and actions covered by the codes and categories (Lempert, 2007). Thus, theoretical memoing allowed exploration of ideas, expansion of the processes they identify, and served as a tool for linking analytic interpretation with empirical reality.

Grounded theory methods are particularly appropriate for this project because they offer a flexible yet systematic set of strategies for collecting and analyzing qualitative data. Collection and analysis of data are simultaneous. This flexibility was invaluable due to the various representations of genetic knowledge investigated. The flexibility of theoretical sampling in grounded theory methodology also help me to shape and alter my data collection strategies to pursue the most interesting and relevant material gathered for the analysis. Since the theoretical categories generated through grounded theory are essentially “grounded” in the data, it is well suited for the emergent nature of the research conducted in this dissertation. To prevent oversimplification of the data, the results of this research represent not only difference(s) but complications, inconsistencies, and incoherencies in the data (Law & Mol, 2002). Furthermore, this approach



analytically allowed the possibilities for multiple major processes, as well as contradictory processes, involved in the production and representation of knowledge on autism genetics to be investigated.

The analysis of this dissertation was facilitated by HyperRESEARCH qualitative data analysis software. All the interviews, field notes, and theoretical memos were entered into HyperRESEARCH. Codes, categories, and theoretical memos were generated separately for each of the four sites, allowing subsequent comparative work (Clarke, 2005).

### **Chapter Outline**

Chapter two provides a social history of parent advocacy groups on autism that prioritize biomedical research. It highlights parent groups that have made research on the genetics of autism a priority, and the specific genetic research agendas that have been initiated through their efforts. Specifically, I discuss the establishment and implications of two scientific genetic enterprises: the Autism Genome Project (AGP), which is the largest international consortium of scientists studying autism genetics, and the Autism Genetic Resource Exchange (AGRE), the largest autism specific gene bank in the world. Drawing on theoretical analyses of health social movements discussed above, this chapter also identifies processes that parent advocates of autism are engaging in through their efforts to unravel biomedical understanding of autism spectrum disorders.

Chapter three describes historical transformations in autism genetics research, the research challenges in ASD that are prompting new genetically constructed meanings of autism, and insights into new knowledge producing technologies shifting the genetic

disease paradigm from inherited single gene causing mutations to rare genetic variants that are spontaneously acquired. Based on interviews with scientists conducting autism genetics research and a review of scientific literature on autism genetics, this chapter specifically highlights three ways in which autism is being redefined based on genetic knowledge: the identification of copy number variants, genetic reclassification of autism phenotypes, and the convergence of common biological pathways. In closing, this chapter analyzes how scientists are imagining the future of autism through their research and constructing autism as a genetic disorder.

Chapter four is based on interviews with parents who have participated in the Simons Simplex Collection, a privately funded genetics research study that is generating a genetic database of DNA and clinical phenotypes of families with one child diagnosed with an ASD. The first part briefly outlines the Simons Simplex Collection, followed by an analysis of the social processes that motivated parents to participate in genetics research. This chapter concludes with a discussion of ethical implications that arise in these narratives by highlighting and addressing specific social dimensions entangled within the fabric of participating in genetics research.

Chapter five literally “follows the DNA,” to highlight the ways in which family information (i.e., blood and family characteristics) are transformed and processed into genetic knowledge through the different yet overlapping spaces of families and individuals with autism, parent advocacy groups, and scientists. By drawing on the work that theorizes boundary objects (Star & Greisemer, 1989) and local contingencies, uncertainties, differences, and processes that are “lost in translation” during the production of genetic knowledge (Fujimura, 2005; Fujimura & Fortun, 1996), this

chapter focuses on the boundaries of social worlds that come together and how families are “lost in translation” in the production of genetic knowledge on autism.

Chapter six explores the notions of genetic identity within the context of autism spectrum disorders. The intent of this chapter is to better understand how adults diagnosed or self-identified on the autism spectrum view the genetics of autism and how their understandings are reflected in their everyday lives. The emphasis here is to explore the different forms of subjectivity that exist among adults either diagnosed or self-identified with high functioning autism or Asperger syndrome. Furthermore, this chapter questions the boundaries placed on medical diagnosis and what symptoms constitute a disorder when people identify certain traits and characteristics to be associated with autism that are representative of the family itself and not necessarily part of the ASD classification under the DSM-IVR diagnostic criteria.

Chapter seven, the final chapter, critically engages the social implications of genetic technologies and genetic knowledge generated through various processes of representing autism. It provides a summary of each chapter that analytically engages a full range of human and non-human actors involved in the production of autism genetic knowledge. I discuss the theoretical implications of this research by discussing its contributions to social studies of science and technology, health social movements, and the interactive and overlapping processes of biomedicalization. I also highlight the substantive implications for the sociology of genetics and the sociology of autism. In closing, I discuss the implications of this research for future sociological research on autism and autism genetics.

## **CHAPTER 2: COLLABORATIONS, CONSORTIA, AND COLLECTIONS FUELING AUTISM GENETICS RESEARCH**

The work of parent advocacy groups has dramatically changed the direction of autism awareness and research over the last fifteen years. This health social movement has generated new forms of collaboration between parent activists and scientists as both pursuers of treatment, prevention, research and expanded funding, as well as active participants in the research enterprise (Silverman, 2004). The efforts of parent advocacy groups such as Cure Autism Now (CAN) and the National Alliance for Autism Research (NAAR), which have now merged into Autism Speaks, demonstrate collective mobilization around disease, especially through their engagement of research activities. As indicated in the scientific overview of Autism Speaks that is promoted on their website, these activities include: promoting cross-disciplinary cooperation, funding research, organizing research summit meetings, and establishing standards for data collection and management to benefit the scientific community.<sup>6</sup> Like many patient (Barbot, 2006; Epstein, 1996; Klawiter, 1999, 2004; Kolker, 2004) and parent (Stockdale & Terry, 2002; Terry & Boyd, 2001) advocacy groups before them, parent advocates of autism are emerging as new partners in the production of scientific knowledge.

The purpose of this chapter is to provide a social history of parent advocacy on autism that focused on biomedical research. It will highlight parent groups that have made research on the genetics of autism a priority and the specific genetic research agendas they have initiated. It will specifically discuss the establishment and implications

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<sup>6</sup> Autism Speaks: Science overview. Retrieved February 17, 2010, from <http://www.autismspeaks.org/science/overview.php>.

of two genetics research enterprises: the Autism Genome Project (AGP), which the largest international consortium of scientists studying autism genetics, and the Autism Genetic Resource Exchange (AGRE). AGRE is now the largest autism-specific gene bank in the world. Drawing on theoretical analyses of health social movements (Brown & Zavestoski, 2004; Brown, et al., 2004; Rabeharisoa, 2003) and theoretical concepts of biological and genetic citizenship, (Heath, et al., 2004; Novas & Rose, 2000; Rose, 2007; Rose & Novas, 2003) this chapter will also identify the processes and strategies that parent advocates of autism engage in their efforts to unravel the biomedical basis of autism spectrum disorders.

Chloe Silverman has also described the social history of AGRE and AGP, characterizing the advocacy groups CAN and NAAR as “reform movements directed toward ‘public-interest science’” (Silverman, 2004, p.308). She argues that their use of commercial and economic tactics go beyond funding autism research, and now involve governance of biological materials, investment strategies, and the production of commodities (Silverman, 2004). Silverman demonstrates how genetic information is constructed as a valuable resource and utilized by citizens and stakeholders, arguing that different “types of strategies, ways of mobilizing resources and knowledge, and alternative conceptions of disease processes and treatments advocated by these groups...are indicative of ways of thinking about autism made possible by broad cultural change.” (Silverman, 2004, p. 40).

The current study provides an update on these genetic research initiatives and future directions. This chapter also provides an analysis of the scientific implications (i.e., successes, limitations, challenges) of the AGRE and AGP based on interviews with

scientists involved in the AGP and/or who utilize the AGRE database for their own research. Furthermore, this chapter analyzes some of the benefits and challenges of parent initiated genetic research programs (AGRE) versus scientist initiated genetic research programs (AGP). Moreover, this chapter will consider the broader health social movements within these advocacy groups that extend beyond research activities.

### **“Cultures of Action”**

Before describing the social history and activities of parent advocacy groups that embrace autism as a genetic disorder, it is important to note that there are major debates about autism causality, which in turn have dramatically affected the health social movements around autism. Three broad movements, which are not mutually exclusive, or exhaustive, include the Autism Research Institute (ARI)/Defeat Autism Now! (DAN!) movement, the anti-vaccine movements, and the Neurodiversity movements. The ARI/DAN! is a specific advocacy group that views autism can be treated effectively through intensive behavior modification and a variety of individualized biomedical treatments (Silverman, 2004).<sup>7</sup> ARI was established by Dr. Bernard Rimland, a parent of a child diagnosed with autism who challenged and changed the long-held belief that autism was an emotional disorder caused by poor mothering (Rimland, 1964). In 1994, Rimland and others developed DAN!, a project that pulled together the different threads of autism symptoms—biochemical, immunologic, and gastroenterological—into a unified whole that would point the way to new biomedical treatments.<sup>6</sup> DAN! brings together hundreds

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<sup>7</sup> Autism Research Institute and Defeat Autism Now!: Who we are, and what we do. Retrieved April 27, 2010, from [http://www.autism.com/ari/editorials/ed\\_aridan.htm](http://www.autism.com/ari/editorials/ed_aridan.htm).

of experts in biannual think tanks and holds conferences throughout the year to spread the word that “recovery” from autism is possible. ARI/DAN! contests the genetic and neurological causes of autism and propose, instead for specific biomedical therapeutics that target environmental insults.

The anti-vaccine movements stem from the idea that either vaccines additives or the vaccines themselves are the cause of or trigger in autism. The health social movements that have been generated from vaccine concerns are very diverse and range from parents advocating for the removal of mercury based preservatives used in vaccines and flu shots (e.g., Moms Against Mercury and SafeMinds), to parents who advocate for delaying vaccines or considering an alternative vaccination schedule (e.g., Generation Rescue). Other parent-initiated groups such as The National Vaccine Information Center are advocating for vaccine safety and informed consent protections in the mass vaccination system. As demonstrated in a recent PBS special on *Frontline: The Vaccine War* (Palfreman, 2010), certain pockets of US communities (e.g., Ashland, Oregon) are choosing not to vaccinate their children. Likewise, a recent “Hope for Autism” conference I attended had medical professionals and scientists publically advocating for parents not to vaccinate their children.<sup>8</sup>

The neurodiversity movement is a new wave of activists comprised of adults with Asperger syndrome or high functioning autism who want to celebrate atypical brain function as a positive identity and not a disability. This group of activists is also diverse

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<sup>8</sup> “Hope for Autism” is a training conference for chiropractors who are interested in learning about BioNutritional Care™, Neurological Therapies, Biomedical Interventions and Integrative Medicine. BioNutritional Care™ utilizes diagnostic tools and methods to determine the underlying physiological causes of symptoms particular to a disease or condition and focus on non-invasive treatments and modalities (Hope for Autism: Bionutritional Care training conference. Retrieved April 27, 2010, from <http://www.hopeforautism.us/>).

and varies in their social and political activities. For example, the Autism Network International is considered the first and largest organization run by autistics and have made a political issue of autism by redefining it as a different way of being and not a disease (Chamak, 2008). Chloe Silverman also demonstrates how autism self-advocacy groups view the desire for a cure as unethical in the sense that it denies “autistic humanity” (Silverman, 2008b). Nancy Bagatell also describes the emergence of the “Aspie” community, where autism is seen as a neurological “difference”, not as an illness or disability (Bagatell, 2007). Many social scientists are beginning to address these emerging social movements of adults on the autism spectrum (Bumiller, 2008; Orsini, 2009), which are and will continue to be important issues given the current proposed changes in the DSM-V to eliminate the Asperger diagnosis. Details of this issue are addressed in chapter six.

In many regards, these diverse social movements reflect Maren Klawiter’s concept of “cultures of action” (Klawiter, 1999). In her research on the social movements of breast cancer, Klawiter demonstrates how three different social movement events sought to raise awareness and reshape the social terrain by creating a different culture of action (Klawiter, 1999, p.121). Although this chapter focuses on health social movements around the production of genetic knowledge, the diverse movements described above must be considered within the broader context of autism health social movements. Many of these movements intersect with the enactments, embodiments, and articulations described throughout this dissertation, as we shall see next.



## **The National Alliance for Autism Research (NAAR)**

When Eric and I established NAAR in the basement of our home, all we knew was that there was a pitiful amount of autism research being conducted and not a single nonprofit organization in this great nation pushing this agenda – Karen London (NAARRATIVE, 2001)

In the months following their son's diagnosis of autism in 1989, Eric and Karen London noticed that there was a severe lack of information available about autism. When they tried to donate money for autism research, they were shocked to learn that there was not one non-profit organization dedicated to biomedical research focusing on the disorder. Five years later, autism research was still poorly funded in both the public and private sectors and only a handful of investigators were seriously focusing on the disorder. Thus, in 1994 Eric and Karen London founded the National Alliance for Autism Research (NAAR) in the basement of their suburban New Jersey home. NAAR was the first national non-profit organization dedicated to funding and promoting biomedical research and treatment of autism spectrum disorders. The organization's name represented the goals of achieving a nationwide alliance of families, autism organizations, researchers and concerned others united in and supportive of a common purpose (NAARRATIVE, 1997). NAAR's primary mission was to stimulate biomedical research into the causes, prevention and ultimately cure for autism spectrum disorders.

Principal components of NAAR's agenda were to accelerate the science by providing direct funding for specific research projects, facilitating communication among researchers, and recruiting new scientific talent into the field (NAARRATIVE, 1997). Among NAAR's many accomplishments was the establishment of the Autism Tissue Program, which they established in partnership with the Autism Society of

America Foundation (ASA). This is a centralized source of brain tissue and associated clinical data from deceased individuals with autism and their families. This tissue is made available to qualified scientists worldwide who want to understand how and why the brain is different in individuals with autism. NAAR also funded over 250 projects, fellowships and collaborative programs worldwide from 1997-2005, contributing an estimated \$30 million towards autism research, more than any other non-governmental organization during this time.<sup>9</sup> Today, the research initially funded by NAAR has been leveraged into more than \$64.5 million over time in autism research awards funded by the National Institutes of Health (NIH) and other governmental sources.<sup>10</sup>

On February 13, 2006, NAAR merged its efforts with Autism Speaks based on their joint commitment to: accelerate and fund biomedical research into the causes, prevention, treatments and cure for autism spectrum disorders; increase awareness; and advocate for the needs of affected families.<sup>9</sup> Autism Speaks was co-founded by Suzanne and Bob Wright<sup>11</sup> after learning their grandson had been diagnosed with autism. The guiding principals of Autism Speaks have been to raise public awareness about autism and its devastating effects on individuals, families, and society, and to raise funds to support effective biomedical research on autism.

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<sup>9</sup> Autism Speaks: Research we have funded. Retrieved February 15, 2010, from [http://www.autismspeaks.org/science/research/grants/research\\_we\\_have\\_funded.php](http://www.autismspeaks.org/science/research/grants/research_we_have_funded.php).

<sup>10</sup> Autism-Speaks: Autism Speaks and The National Alliance for Autism Research complete merger. Retrieved February 15, 2010, from [http://www.autismspeaks.org/press/autism\\_speaks\\_naar\\_merger.php](http://www.autismspeaks.org/press/autism_speaks_naar_merger.php).

<sup>11</sup> Bob Wright is the vice chairman of General Electric and former CEO of NBC Universal, Inc., a media and entertainment company. The Wright's have received numerous awards for their work with Autism Speaks. In 2008 they were recognized as among the top 100 most influential people in the world (Brokaw, 2009). Since its inception in 2005, Autism Speaks has committed over \$131 million in autism research and development of innovative resources for families through 2014.

After the merger, Eric London joined the executive and scientific advisory committees of Autism Speaks. However, in 2009 he resigned from his position and affiliation with Autism Speaks due to differences in the organizations' direction, prioritization of the science program, and decision-making processes. London's resignation letter indicated that the pivotal issue compelling his decision was the position of Autism Speaks concerning vaccinations and their investment and advocacy for research on the rare cases of "biologically plausible" vaccine involvement in autism causation(ASF, 2009). The issue of childhood vaccinations, most notably the measles, mumps and rubella (MMR) vaccine, has caused much controversy regarding the cause of autism. As noted earlier, there are strong movements by parents and scientists against the use of the MMR vaccine based on the assumption that the vaccine causes autism.<sup>12</sup>

Since his resignation, the Londons have become affiliated with a new organization, the Autism Science Foundation (ASF), which was co-founded by Karen London and Alison Tepper Singer.<sup>13</sup> The ASF mission is to support autism research by providing funding and other assistance to scientists and organizations conducting, facilitating, publicizing and disseminating autism research. The mission statement

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<sup>12</sup> The controversy originated with a 1998 study conducted in the UK in which data from 12 children indicated a possible temporal association between the MMR vaccine and autism (Wakefield, 1998). Public concerns prompted a flow of research funds to test the claims of the Wakefield study, which disproved the hypothesis of a link between autism and the MMR vaccine. Despite this evidence, many parents continue to be skeptical of the safety of the vaccine. For more discussion on this pressing issue see Silverman (2004), Hobsen-West (2007), Senier (2008), and Kaufman (2010).

<sup>13</sup> Allison Tepper Singer was formerly Executive Vice President of Autism Speaks, who resigned due to disagreement on whether to support research into a possible link between vaccines and the onset of autism. Singer believes research in this area has already proved there is no link and that future research needs to support other kinds of science that will yield new and useful information.

specifically states that further investment in the link between vaccines and autism is not warranted at this time.<sup>14</sup> Eric London serves on the Scientific Advisory Board of ASF. His departure from Autism Speaks and the London's affiliation with ASF vividly demonstrates their main intention in establishing NAAR: to fund and promote evidence based biomedical research and treatment for autism spectrum disorders.

### **Consortium of Consortia**

The best opportunities for determining the causes of autism, developing more accurate diagnostic techniques, specific medical treatments and cures will occur more rapidly when researchers and organizations work together and share their collective resources. (NAARRATIVE, Winter 2005)

One of the original goals of NAAR was to facilitate communication among researchers. Thus, it is no surprise that they funded efforts to bring scientist together to inspire research collaborations and promote biomedical research. For autism genetics, this started in 2000 through a three-day retreat including about 40 scientists affiliated with seven of the major autism genetics research groups around the world.<sup>15</sup> The retreat was initiated and organized by the scientists themselves based on the realization that collaboration would be advantageous if not essential to identify one or more “autism genes”. At the time, there was no available government funding for such an effort, thus

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<sup>14</sup> Autism Science Foundation: Our mission. Retrieved February 15, 2010, from <http://www.autismsciencefoundation.org/aboutasf.html>.

<sup>15</sup> The scientists included senior investigators and team members from the Collaborative Linkage Study of Autism (Tufts, Vanderbilt, University of Iowa and University of North Carolina); Stanford University; Mt. Sinai's Seaver Center; Duke University/University of South Carolina; University of Missouri; McMaster University, McGill University and the University of Toronto (the “Canadians”); and the Paris Autism Research International Sib Pair Study (PARIS).

NAAR and the Nancy Lurie Marks (NLM) Family Foundation<sup>16</sup> co-funded the annual meetings until 2003.

Parent representatives from NAAR and the NLM Family Foundation also attended these yearly workshops. The scientists involved in these initial meetings acknowledged the importance of their early support and encouragement, especially given the political and competitive nature of collaborative scientific endeavors. As one scientist reflected, *“They would occasionally remind us why **we** were there in the first place, why **they** were there in the first place and get everyone back on track.”*<sup>17</sup>

This group of scientist became known as the Autism Genetics Cooperative (AGC) and was originally organized by Dr. Susan Folstein,<sup>18</sup> a geneticist from Tufts University. It was called a cooperative because everybody had an equal say, there was no specific leader, and decision-making was completely democratic. The short-term goals of the AGC were to combine all the genetic linkage data together from these different research groups to conduct a larger comprehensive analysis. They predicted that larger sample sizes would help resolve the problems they were having of replicating research results. The scientists met at Calloway Gardens near Atlanta, Georgia. In order to participate,

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<sup>16</sup> The Nancy Lurie Marks (NLM) Foundation donated over \$1.5 million to NAAR during their early efforts of starting the organization (Autism Speaks: NAAR news archive NLM family foundation helps kick off \$7.5 million matching gift campaign for autism research. Retrieved February 15, 2010, from

[http://www.autismspeaks.org/inthenews/naar\\_archive/nlm\\_family\\_foundation.php](http://www.autismspeaks.org/inthenews/naar_archive/nlm_family_foundation.php)). The NLM Foundation is committed to understanding autism from a scientific perspective, increasing opportunities and services available to the autism community and educating the public about autism. The NLM Foundation also donated \$1 million to the Autism Genome Project (Nancy Lurie Marks Family Foundation: About the NLM. Retrieved May 18, 2010, from [http://www.nlmfoundation.org/about\\_nlm.aspx](http://www.nlmfoundation.org/about_nlm.aspx)).

<sup>17</sup> Scientist Interview #8 - Geneticist

<sup>18</sup> Prior to the first workshop in 2000, it took Dr. Folstein two years just to get the scientists organized to work together through a few meetings and various conference calls (personal communication). Dr. Folstein is also one of the first scientists to conduct twin studies on autism (Folstein & Rutter, 1977).

they had to agree to two rules: they must present unpublished data; and the presented data could not leave the conference and had to remain confidential until investigators published their results. The workshops essentially provided a forum for the researchers to meet and discuss unpublished data and share intellectual resources. These face-to-face meetings were essential in the early days of collaboration to establish trust among the researchers who, prior to the AGC, did not know each other very well and/or viewed each other as competitors. In 2002, The National Institute of Neurological Disorders and Stroke (NINDS) began supporting the AGC through a five-year grant to develop an infrastructure to enhance collaboration. In addition to annual meetings, this infrastructure included a Virtual Private Network (VPN), and an accessible database that included phenotype and genotype data from all participating groups (Folstein, 2002).

In the wake of this success, NAAR wanted to expand the AGC to include more research institutes that were collecting multiplex families (families with two or more children diagnosed with autism). The Director of Research and Programs at NAAR, Andy Shih, saw their role as an “*honest broker*” in the process of building a larger autism genetics consortium. He explained to me that since NAAR was a parent advocacy group, they were able to ask the scientist for anything they wanted and “*people listened*”. In 2003, the National Institutes of Health committed its support to the project and later that year, NAAR and the NIH officially announced the development of the Autism Genome Project at the Autism Summit Conference in Washington, D.C.<sup>19</sup>

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<sup>19</sup> Autism Speaks: NAAR Autism Genome Project Frequently asked questions. Retrieved 2010, from [http://www.autismspeaks.org/docs/Autism\\_FAQ\\_revised\\_NAAR.pdf](http://www.autismspeaks.org/docs/Autism_FAQ_revised_NAAR.pdf).

Through a joint commitment of \$4.5 million, NAAR and four institutes at the NIH<sup>20</sup> co-funded the Autism Genome Project (AGP), whose goal was to locate the genes associated with autism spectrum disorders (ASD). The AGP “was designed to enable doctors to biologically diagnose autism and enable researchers to develop universal medical treatments and a cure”.<sup>21</sup> This private/public partnership was described as a “consortium of consortia” in press releases because it was composed of four main research teams including: the Autism Genetics Cooperative (AGC); the International Molecular Genetic Study of Autism Consortium (IMGSAC); the Collaborative Program of Excellence (CPEA); and the Autism Genetic Resource Exchange (AGRE).<sup>22</sup> Collectively, these teams included over 120 genetic researchers from over 50 academic and research institutions throughout the United States, Canada, the United Kingdom, France, Sweden, Denmark and Germany. It marked the largest collaboration ever to focus on the genetics of autism and the largest sample set ever assembled. The sample consisted of approximately 6,000 samples of DNA from 1,500 multiplex families.

The collaboration consisted of two inter-related phases funded separately. The first phase was to conduct a genome-wide scan,<sup>23</sup> utilizing the Affymetrix 10K

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<sup>20</sup> These include the National Institute of Mental Health (NIMH), National Institute of Child Health and Human Development (NICHD), National Institute of Neurological Disorders & Stroke (NINDS), and National Institute of Deafness and Other Communication Disorders (NIDCD) (Autism Speaks: NAAR Autism Genome Project Frequently asked questions. Retrieved 2010, from [http://www.autismspeaks.org/docs/Autism\\_FAQ\\_revised\\_NAAR.pdf](http://www.autismspeaks.org/docs/Autism_FAQ_revised_NAAR.pdf)).

<sup>21</sup> Autism Speaks: Autism Genome Project. Retrieved February 17, 2010, from [http://www.autismspeaks.org/science/research/initiatives/autism\\_genome\\_project.php](http://www.autismspeaks.org/science/research/initiatives/autism_genome_project.php).

<sup>22</sup> For a complete list of all the institutions involved in each of these research teams or consortia, see Hu Lince, et al., 2005.

<sup>23</sup> A genome scan is a genetic analysis designed to identify intervals in the human genome that show the highest priority for further investigation. Intervals are like “genomic neighborhoods” that appear to be most likely places where the genes believed to be involved with autism exists.

microarray,<sup>24</sup> and a second scan using standard microsatellite technology.<sup>25,26</sup> Once regions of significant linkage are identified, the second phase of the AGP was to focus on identifying the exact nucleotide changes that predispose to autism and reside within the linkage intervals derived from phase 1 (Hu-Lince et al, 2005). The first phase of the AGP was co-funded by NAAR and the NIH and the results were published in 2007 (The Autism-Genome-Project-Consortium, 2007).

It was anticipated by NAAR that the second phase would be funded through a public/private Request for Application (RFA) announced by the NIH in 2005 to identify autism susceptibility genes (DHHS, 2004). NAAR was an integral part of developing the partnerships in this RFA, which included three public and three private sector funding partners.<sup>27</sup> Collectively these groups committed more than \$21 million to fund research to

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<sup>24</sup> Affymetrix is a pioneering company that creates tools for genomic analysis. In the late 1980's a team of scientists lead by Stephen P.A. Fodor, Ph.D., combined semiconductor manufacturing techniques with advances in combinatorial chemistry to build vast amounts of biological data on a small glass chip. This chip, the GeneChip® brand microarray, a tool that allows scientists to identify mutations, or changes, in a person's DNA at finer detail than was previously possible. It has become the industry-standard tool that researchers use to analyze genetic information (Affymetrix: About Affymetrix. Retrieved February 2, 2010, from [http://www.affymetrix.com/estore/about\\_affymetrix/index.affx?category=34003&categoryIdClicked=34003&rootCategoryId=34003&navMode=34003&aId=aboutNav#1\\_3](http://www.affymetrix.com/estore/about_affymetrix/index.affx?category=34003&categoryIdClicked=34003&rootCategoryId=34003&navMode=34003&aId=aboutNav#1_3))

<sup>25</sup> Microsatellite markers are polymorphic stretches of DNA that consist of tandem repeats of a simple sequence of nucleotides, which vary in length throughout the genome. These markers are spaced at intervals of ~10cM across the genome and have been the traditional method used in genome-wide linkage scans (Hu-Lince, Craig, Huentelman, & Stephan, 2005).

<sup>26</sup> NAAR funded the DNA micro-array scan, which was conducted by a non-profit research group, Translational Genomics Research Institute in Phoenix. The microsatellite scan was funded by the NIH, which was conducted by the Center for Inherited Disease Research (NAARRATIVE, 2005). All the genotype analysis was conducted by the AGP data-coordinating center located at the Battelle Center for Mathematical Medicine (formerly known as the Center for Quantitative and Computational Biology) in Ohio.

<sup>27</sup> The public sector funders included: the National Institutes of Health (\$7.0 million), the Irish Health Research Board (\$6.1 million), and the Canadian Institutes of Health Research (\$1.25 million). The private sector funders included: the National Alliance for Autism Research (\$6.5 million), Cure Autism Now (\$500, 000), and the Southwest Autism Research and Resource Center (\$100,000) (NAARRATIVE, 2005).



identify autism susceptibility genes. However, under the NIH review process, the funding partners were told a week before hand that the review process was going to be closed and none of the funding partners were allowed to participate in the application review except for the NIH. This was extremely disappointing for NAAR, since they were committing \$6.5 million to this five-year grant. To their dismay, the AGP was not competitive in the final review and did not get funded for the second phase of the project.

Ultimately, the second phase of the AGP was funded by a \$14.5 million investment over three years from an unique combination of international, public and private partners including: Autism Speaks, the UK Medical Research Council (MRC), the Health Research Board of Ireland (HRB), Genome Canada and partners, Canadian Institutes for Health Research (CIHR), Southwest Autism Research and Resource Center (SARRC), and the Hilibrand Foundation. Phase two progress of the AGP was reported at the 2009 International Meeting for Autism Research. Reports described the expansion of the collective data to ~3000 families, including approximately 1,500 trio families (two typical parents and one child diagnosed with autism). Their conclusion was that more samples were needed to find a common allele that contributes to autism spectrum disorders.

### ***Implications of the AGP***

The scientific implications of the AGP reside largely in its ability to effectively collaborate internationally with multiple groups of scientists, making it the largest collaboration in the world studying the genetics of autism. By pooling samples, they were able to conduct the largest genetic linkage analysis that had ever been studied. According

to AGP scientists, the results of the first phase also gave insight into the underlying genomic architecture, showing that there were multiple genetic factors involved with autism (The-Autism-Genome-Project-Consortium, 2007). Furthermore, the AGP had a large enough dataset to identify rare *de novo* copy number variants (CNVs) through the use of new DNA microarray technologies.<sup>28</sup> This legitimized CNVs as an emergent set of genetic elements scientists believe are involved in the etiology of autism spectrum disorder. The AGP scientists were also optimistic about the AGP samples becoming publically available in future and the potential for secondary analysis in a subset of the families due to the large sample size.

The social implications of the AGP are the new modes of being a scientist and approaches to genetics research. This approach is based on collaboration, sharing, openness, and trust. In this context, science is a field of exchange rather than a one-way production.<sup>29</sup> This approach is also favored by funding agencies, which further stipulates the need for scientists to collaborate. Thus, the AGP brought scientist studying other diseases into the world of autism genetics due to the availability of funding and scientific enthusiasm for autism genetics. This created what Joan Fujimura describes as a “scientific bandwagon” (Fujimura, 1996).<sup>30</sup> Having this consortium helped scientists talk to each other and think about the science in different ways due to the range of ideas and opinions about the best way to approach genetic understandings of autism. Furthermore,

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<sup>28</sup> Chapter three discusses the CNV hypothesis in further detail, as well as the implications of DNA microarray technologies.

<sup>29</sup> I thank Janet Shim for pointing out this important social implication of the AGP.

<sup>30</sup> A scientific bandwagon exists when large numbers of people, laboratories, and organizations commit their resources to one approach to a problem because *others* are doing so and the technologies, skill sets, funding, and other infrastructural elements are accessible and usable (see: Fujimura, 1996: 225-227).

as one scientist stated, “*people who now have substantial time dedicated to the AGP instead of other things...get individual labs and individual clinical investigators to do things differently.*”<sup>31</sup> Thus, the AGP has helped to foster continuing research in individual labs that may not have focused on autism otherwise.

### ***Challenges of the AGP***

Despite relative optimism regarding the success of the AGP, one of the biggest challenges for the AGP was the quality of the genotype and phenotype data collected. First, the logistics of actually pooling the data into a central databank proved to be very time consuming and labor intensive. Obtaining samples was difficult because of issues of consent. Some families did not originally agree to participate in the AGP, and thus had to be re-contacted and re-consented. This was a challenge because some of these samples had been collected over many years with different consent forms and from different geographical locations. These logistical challenges also prevented the AGP collection to be publically available.

Once the samples were collected, the second major hurdle for the AGP was the process of “*cleaning up the data*”. Since the samples were derived from multiple groups based on completely different research projects, there was high variability in what was actually collected for each sample. For example, many samples did not utilize both the Autism Diagnostic Interview-Revised (ADI-R) and the Autism Diagnostic Observation Scale (ADOS) to diagnose autism. Furthermore, over 20 different IQ tests were used in

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<sup>31</sup> Scientist Interview #15 - Developmental psychologist

the various collections that make up the AGP, resulting in disparate quality of data across the sites. Even if researchers were using the same assessment tools, there was still variability across the different sites, causing inconsistencies in the data collected.

The genotypic data were of variable quality as well, since different sites used different platforms to genotype the data, some of which would not be considered adequate marker density for research conducted today. Thus, coordinating consistent phenotyping and genotyping among all the groups and combining samples of variable quality into something that could be used for research was a very labor intensive and time consuming process for the AGP consortium. In the end, some AGP scientist have argued that the data in the AGP was “*never as good as it needed to be*”,<sup>32</sup> which in turn has caused much speculation of the results of phase 1 and 2 of the AGP.

Another major challenge for the AGP was overcoming the scientific differences that were represented in the group. According to several involved scientists, there were legitimate differences in how to approach scientific experiments and each group had their own objectives, needs and motivations. A clear example was the disagreement in phase one over which technology to use for the initial linkage scan: the older more established method of microsatellites markers or the newer and less developed approach using single nucleotide polymorphism (SNP) markers.<sup>33</sup> After many heated discussions and meetings among the AGP consortium, the NIH convened a panel of experts who recommended the AGP use the microsatellite technology. In the end, however both technologies were used

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<sup>32</sup> Scientist Interview #15 – Developmental psychologist

<sup>33</sup> For a detailed analysis of SNP vs. microsatellite technologies see Hu-Lince et al. (2005, especially pgs. 240-242).

for the analysis. The NIH funded the microsatellite technology and the newer microarray SNP technology was funded by NAAR. This is just one of many scientific differences that the AGP had to negotiate and resolve. Some would argue such differences manifest distrust among the scientists within the group, especially in the beginning.

There were several implications of approaching science in an innovatively collaborative manner, including the speed at which things got done, and producing what several AGP scientists refer to as “*lowest common denominator science*”. First, the speed at which things have been done has been slow for the AGP. The initial data pooling and data cleaning took longer than expected, and several scientists have been disappointed with the slow pace. The scientific differences, especially regarding how to analyze and interpret the data, were also rate-limiting factors. One scientist described the pace of the AGP as like “*navigating a bathtub down a set of rapids. It's very slow. It's very slow. It doesn't have the agility of a smaller group to make rapid decisions and to move forward quickly*”.<sup>34</sup>

“*Lowest common denominator science*” is described as “*boilerplate*” and “*standard*” science that everyone can agree on and is the “*blandest, most uninteresting, least productive thing to be doing*”. In many regards, the AGP, like many large consortiums, produces knowledge that has been compromised by everyone in the group. Socially, this makes sense in order to get things done in a large group of people. Scientifically, however, it may not be the best approach for complex diseases like autism. Critics of consortia science or “*megascience*” contend that it stymies real innovation and prevents researchers from making the type of serendipitous findings that have

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<sup>34</sup> Scientist Interview #14 – Child psychologist

historically been huge turning points in science (Salisbury, 2010). As a result, it can be argued that big consortia like the AGP are not producing meaningful scientific results and in many regards overshadow smaller and more interesting studies.

To date, the AGP has published two papers, one which reported the results of phase 1 (The-Autism-Genome-Project-Consortium, 2007) and another that reported the results of a linkage analysis of quantitative and categorical subphenotypes (Liu, Paterson, Szatmari, & The-Autism-Genome-Project-Consortium, 2008). Some of the AGP scientists regard this work as contributing to a better understanding of the heterogeneity of autism genetics and moving the field in the right direction. Others regard the progress of the AGP as contributing very little and not shedding fundamental light on the genetics of autism. Thus, conflicts continue to exist within the AGP with regard to the relative importance of the work they have produced thus far.

### ***Future of the AGP***

The future of the AGP is speculative and will depend largely on whether funding is available. There is a belief among the AGP scientists that funders, especially in the United States, are less enthusiastic about the future of AGP than they were in the first two phases. The major emphasis for phase three of the AGP will be to focus on translating genetic knowledge produced by the AGP into clinical applications. Their plan is to screen children at birth based on the genetic information that has been identified in the first two phases of the AGP. It is anticipated that this screening, most likely utilizing microarray-based technologies, will help to identify children at higher risk of having autism and those who need behavioral and neural training intervention. However, the future of this

research depends on many contingencies: the state of the economy, whether AGP gets refunded, and whether different groups decide to continue their participation in the AGP. More importantly, as I discuss in chapter three, the clinical implications of genetic knowledge of autism will also rely on the ability of scientists to “generate” meaning of new genetic mutations (i.e., copy number variants) that are being generated with current sophisticated technologies.

### **Cure Autism Now (CAN)**

Cure Autism Now (CAN) is an organization of parents, clinicians, and scientists committed to accelerating research to prevent, treat and cure autism – for individuals and families today and for future generations. The organization is one of the leading private funders of biomedical research in autism, providing more than \$31 million for research grants, education, outreach and scientific resources, including the establishment and ongoing support of the Autism Genetic Resource Exchange (AGRE) (CAN, 2005b).

As the name indicates, the parent advocacy group Cure Autism Now (CAN) was on a mission not only to raise awareness and increase funding for autism research, but also to find a cure. CAN was established in 1995 by Jon Shestack and Portia Iversen, two Hollywood professionals who were given the advice by doctors to “get on with your lives” after their son was diagnosed with autism (Iversen, 2006, p.31). After being told there was nothing they could do for their son, they assessed the state of autism research and funding, and quickly realized that limited research was being done. As they saw it, there had never been a more optimal time in the history of science and medicine to set out to discover the causes and treatments of autism (Iversen, 2006). Thus, in 1995 they

started the research foundation, Cure Autism Now (CAN), a name that met much resistance by professionals and parents for providing “false hope” (Iversen, 2006, p.31).

Despite resistance, they effectively lobbied Washington to increase new federal dollars for autism research, became experts in the science of autism, and recruited and funded researchers from related fields to conduct autism research. From 1997 – 2007, CAN funded over 200 “field-building” research grants, including pilot projects, young investigator, treatment, and innovative technology awards. These efforts were born out of the necessity to stimulate novel research and entice investigators to join the fight to understand autism. In 2007, CAN merged with Autism Speaks, making it the largest autism science and advocacy organization. Jon Shestack, co-founder of CAN, joined and currently remains on the board of directors and executive committee of the board for Autism Speaks.

### **The Autism Genetic Resource Exchange (AGRE)**

A major contribution of CAN was the establishment of the Autism Genetic Resource Exchange (AGRE).<sup>35</sup> The development of AGRE through families literally “becoming the data” emerged through a question CAN posed to a group of scientists regarding the single most important thing they could do to speed progress in autism research. The advice they received was to establish an open-access gene bank for autism research that consisted of DNA and high-quality clinical data of multiplex families (families with two or more children diagnosed with autism). At the time, the limited

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<sup>35</sup> Chloe Silverman (2004, 2008) also provides detailed historical accounts of CAN and the Autism Genetic Resource Exchange (AGRE). She describes CAN/AGRE as parental networks that chose to “create a material resource in the form of a genetic repository” (Silverman, 2008:44).



collections of blood samples from multiplex families and the lack of sharing or pooling of samples among researchers resulted in sample sizes that were too small to conduct meaningful genetics research.

The establishment of the AGRE in 1997, however, met resistance and skepticism from the scientific community, mainly objecting that the AGRE data would be freely shared. The competitive and “paranoid” nature of scientific research at the time discouraged researchers to share data and results prior publication. Samples were collected for the benefit of the researcher, not to benefit science or the families of children with autism. In contrast, AGRE intended to open its data to any qualified researcher who promised to share raw data from their analysis. This was a different than the AGP, which intended to only combine the samples for an agreed upon analysis among the members of the AGP only. To date, the samples pooled from various research groups contributing to the AGP does not allow for other investigators outside of the AGP to access the pooled samples. AGRE, which is part of the AGP pooled sample, is the exception. According to AGP members, this was a point of conflict between CAN/AGRE and the AGP. CAN wanted AGRE samples to be publically available regardless of whether the scientists were part of the AGP. It was explained to me that since the majority of samples in the AGP were from different studies, different IRB issues applied. This made the different samples pooled for the AGP amenable to more restrictions in terms of distribution. Thus, only the AGP as a group can use the pooled samples collectively, and no data can be published independently if it is based on the pooled samples. In the end, 426 families from AGRE were part of the AGP sample, which

remained an open source of data and required the submittal of raw data prior to publication.

Scientists also doubted whether a parent organization could carry out the AGRE data collection with the scientific rigor that was necessary to conduct quality genetics research. Initially the quality and completeness of the data were problematic because the parent group thought they could simply mail out blood kits to families who said they had a child with autism. As one scientist commented, *“it took a number of years for them to generate the data at a level of quality, both the phenotypic and the genotypic data, that scientists felt reasonably comfortable with.”*<sup>36</sup> Thus, in the early stages there was a lot of pressure by scientists to have standardized diagnostic measures used in the AGRE collection. This prompted CAN to hire top specialists in autism diagnostics<sup>37</sup> to train their staff in evaluating the families. Along the way, many standard scientific protocols were put in place based on the advice of scientists, such as the use of legitimate diagnostic instruments, the use of proper informed consent documents, and the grant review processes.

Despite initial challenges, AGRE enrolled 100 families the first year and operated independently until 2002. Then, the National Institute of Mental Health (NIMH) granted \$6 million in additional funding to expand the AGRE program to 800 families (D.H. Geschwind, 2002). By partnering with NIMH, AGRE's collection is now available to all NIMH-funded researchers at no cost and to other qualified researchers for a fee through

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<sup>36</sup> Scientist Interview #7 – Psychiatric geneticist

<sup>37</sup> The standard diagnostic tools used at the time were the Autism Diagnostic Interview – Revised (ADI-R) and the Autism Diagnostic Observation Scale (ADOS). Currently the AGRE families are assessed using up to ten different diagnostic tools as well as race and ethnicity, language data, head circumference and handedness.

the NIMH Genetics Initiative.<sup>38</sup> In 2007, AGRE partnered with several academic institutions to establish the Center for Phenomic and Genomic Studies through a five-year, \$8 million grant from the National Institutes of Health (Lajonchere, 2007). The NIH funds will double the number of families and expand the data beyond genetic and clinical profiles to include what the researchers call phenomics: the systematic study of the outward physical and behavioral marks of autism. Phenomics is not a concept unique to autism, rather a transdiscipline dedicated to the systematic study of phenotypes on a genome-wide scale by integrating basic, clinical and information sciences (Freimer & Sabatti, 2003).<sup>39</sup> AGRE's expansion will focus on recruiting an ethnically diverse group of families, since Caucasians have been over-represented in genetic studies to the point that diagnostic tools are unreliable for minorities (Marziali, 2007). It will also expand the type of data collected in a subset of families to include additional biological measures, such as structural brain imaging, DNA microarrays and immunological assays.

Currently, AGRE is the world's largest private repository of clinical and genetic information on over 1250 families affected with an ASD.<sup>40</sup> AGRE's biomaterials repository, which includes DNA, plasma, serum, and cell lines, has now grown to exceed 10,000 samples, offering researchers an unprecedented resource for their scientific studies. The community of AGRE-approved researchers has grown to include 240

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<sup>38</sup> The goal of this initiative is to establish a national resource of clinical data and biomaterials collected from individuals with autism, schizophrenia, bipolar disorder, depression, obsessive-compulsive disorder, and other mental disorders (National Institutes of Mental Health: Center for Collaborative Genetic Studies on Mental Disorders. Retrieved February 18, 2010, from [http://zork.wustl.edu/nimh/home/d\\_autism.html](http://zork.wustl.edu/nimh/home/d_autism.html)). The autism data in the NIMH Genetics Initiative comes from many different sources in addition to AGRE.

<sup>39</sup> Phenomics approaches require collecting phenotypic information, in any given individual, at a series of different levels of resolution (molecules, cells, tissues and whole organisms) and then determining how these features can profitably be studied together (Freimer & Sabatti, 2003).

<sup>40</sup> AGRE website accessed May 30, 2010 from: <http://www.agre.org/>

scientists worldwide and over the last decade, the resource has generated over 160 research papers, 52 papers in the last 2 years alone.<sup>41</sup> Although AGRE began as a grass roots project initiated and managed by parents, the current collection is “*firmly in the hands of the top autism researchers in the country*”.<sup>42</sup> Jon Shestack remains as the only parent among the fourteen members of the AGRE steering committee, which provides oversight and direction for this program. Other current steering committee members are researchers representing fields highly relevant to autism research such as: neurodevelopmental biology, neurology, neuroimaging, learning and cognition, immunology, genetics, biochemistry, and pharmacology.<sup>43</sup> Although the AGRE collection is now under the auspices of Autism Speaks (CAN merged with Autism Speaks in 2007), the operations of AGRE still remain in Los Angeles, as opposed to New York, where Autism Speaks is located.

### ***Implications of AGRE***

AGRE has established a partnership between families and researchers that is changing the landscape of autism genetics by leaps and bounds. Without the availability of biomaterials and clinical information from thousands of participating families, the field would not be where it is today - Clara Lajonchere, VP of Clinical Programs and Managing Director of AGRE<sup>44</sup>

Without doubt, scientists agree that the development and use of the AGRE

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<sup>41</sup> Autism Speaks: AGRE collects 10,000<sup>th</sup> DNA sample. Retrieved April 21, 2010, from [http://www.autismspeaks.org/science/science\\_news/agre\\_milestone\\_10000.php](http://www.autismspeaks.org/science/science_news/agre_milestone_10000.php).

<sup>42</sup> Scientist Interview #18 – AGRE Researcher Liaison

<sup>43</sup> Autism Speaks: Family Services 100 Day Kit. Retrieved February 18, 2010, from [http://www.autismspeaks.org/community/family\\_services/100\\_day\\_kit.php](http://www.autismspeaks.org/community/family_services/100_day_kit.php).

<sup>44</sup> Autism-Speaks (2009). Autism Speaks' Autism Genetic Resource Exchange (AGRE) continues to support genetic research and findings Retrieved December 18, 2009, from [http://www.autismspeaks.org/press/agre\\_supports\\_genetic\\_research\\_and\\_findings.php](http://www.autismspeaks.org/press/agre_supports_genetic_research_and_findings.php)

collection has dramatically accelerated autism genetics research. In 2009, there were 24 publications, including the identification of variations on a region of chromosome 5 (5p14.1), recognized as one of the top 10 medical breakthroughs by *Time Magazine* (Park, 2009) and was among the top 10 Autism Research Achievements of 2009.<sup>45</sup> This particular study utilized over 2500 samples, almost half of which were from AGRE, to identify genes associated with risk for ASD. These genes code for proteins called cadherins, which help neurons find the correct place in the brain and make connections with other neurons during early brain development.

The public availability of AGRE has also opened up the possibility of conducting autism research to smaller and broader research groups. For smaller groups, merely collecting samples could take years and would be impossible for many researchers not affiliated with a clinical setting or having the resources and time available to recruit families. This has been especially useful for small labs and young researchers just starting out who want to test a hypothesis very quickly. The availability of AGRE also opened up autism research to a broader group of scientists who would not necessarily study autism if samples and funding were not available.

We hear echos of classic scientists arguments in the data. For example, some scientists assert that the availability of the AGRE has allowed the best ideas to come forward because different researchers, not only geneticists, are utilizing the publically available data. Thus, many scientists studying autism genetics feel that the AGRE has

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<sup>45</sup> Autism-Speaks: Top 10 autism research achievements of 2009. Retrieved January 18, 2010, from [http://www.autismspeaks.org/science/science\\_news/top\\_ten\\_autism\\_research\\_events\\_2009\\_gwas.php](http://www.autismspeaks.org/science/science_news/top_ten_autism_research_events_2009_gwas.php).

been vital for the progress of autism research. They also noted consequences of good access to needed materials. As one scientist pointed out “*having samples available could now allow me to focus on just doing the science*”.<sup>46</sup> Technology matters too. Focusing on “*just doing the science*” is further enabled by the ability to transfer the phenotype and genotype data electronically back and forth through the Internet System for Assessing Autistic Children (ISAAC), a web-based data management system that allows researchers to enter, manage, and share clinical data among researchers in the community.<sup>47</sup>

Perhaps the most important scientific impact of AGRE has been what is asserted as a paradigm shift in collaboration and data sharing policies in the genetics of psychiatric and neurological diseases and disorders. AGRE forced researchers to collaborate whether they wanted to or not and marked the beginning of multiple collaborations between scientists studying autism. It also influenced broader data sharing policies at the national level, which is evident in the development of the NIMH Genetics Initiative and the National Database for Autism Research (NDAR).<sup>48</sup>

Robert Merton’s (1973) communitarian expectations of science are also transpired by the development of AGRE. By taking the responsibility of data collection away from

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<sup>46</sup> Scientist Interview #9 – Molecular Geneticist

<sup>47</sup> This system was designed and developed by Paul Law, a parent of a child with autism who partnered with CAN to develop ISAAC. He has an MD and MPH from John’s Hopkins and specializes in health informatics and pediatrics. Over 400 researchers have access to ISAAC and the system has served as the prototype for larger scale research databases such as the National Database for Autism Research.

<sup>48</sup> NDAR is a secure bioinformatics platform for scientific collaboration around autism spectrum disorders to facilitate data sharing and collaboration. It is a research portal that links data, supporting documentation, publications, and grants information relevant to autism research (National Database for Autism Research. Retrieved January 19, 2010, from <http://ndar.nih.gov/ndarpublicweb/aboutNDAR.go>). It was described to me as “*a big ocean that all the NIMH autism research data is supposed to flow into*” (Scientist Interview #18 – AGRE research liaison).

individual researchers or labs and providing it at a reasonable cost to the entire scientific community, the data itself became a public commodity. This engendered a sense of obligation for researchers in turn to provide raw data (i.e., genotypes) back to the scientific community on any research that utilizes the AGRE database. Thus, the AGRE enabled scientific knowledge to be accessible to all members of the scientific community to use (communism) (Merton, 1973). Chloe Silverman also argues that parent advocates used Merton's scientific norms "to subvert established practices in genetics research and in funding, pointing to sets of practices that were institution outcomes, rather than established elements of practice in genetics research." (Silverman, 2004, p.309) The scientists I interviewed acknowledged their altered scientific practices when they utilized the AGRE database. For example, one scientist described his actions in the following statement:

[w]e completed data collection and we released it long before even we analyzed it, and clearly long before our papers were published. Most people don't do that...By the way, if *I* go out and collect families, *I* may have behaved differently.<sup>49</sup>

Here, the motivation to share unpublished genotype data is driven by the public availability of the AGRE samples. Furthermore, if the researcher collected the samples him/herself, sharing samples and data results would not be a likely practice.

Altering the scientific practice of researchers also required a dramatic shift in governance over the gene banks. In this case, the governance of AGRE was in the control of the parent advocacy group since they owned the biomaterials (i.e., DNA, cell lines, plasma and blood serum) and clinical data (e.g., diagnostic assessments, family histories, and medical evaluations). This shift in governance is evident in the AGRE researcher

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<sup>49</sup> Scientist Interview #1 – Human geneticist

agreement form that must be submitted in order to receive AGRE biomaterials and clinical data. The agreement clearly prohibits the use of any AGRE materials for commercialization purposes without authorization of AGRE, and requires all ‘Researcher Generated Data’ to be supplied to AGRE, including all descriptive genetic analysis data regarding the genotyped genetic markers identified. Furthermore, AGRE has sole discretion as to whether to approve or disapprove researcher applications, which includes applications from academic, clinical and private institutions.

However, despite the AGRE researcher agreement, the communitarian expectations have been betrayed within the private sector. For example, CAN (now Autism Speaks) has sold and distributed AGRE samples to a handful of private companies, including: Lippomix, deCode Genetics, Perlegen, and IntegraGen, who according to the AGRE research liaison, are primarily interested in creating diagnostic tools for autism. Currently, IntegraGen is offering an autism specific “panel of risk biomarkers”, which is used to “identify individuals with a brother or sister already diagnosed as autistic as having a risk of contracting autism” (IntegraGen, 2010). Despite the AGRE researcher distribution agreement signed by IntegraGen, they have filed a patent based on the AGRE data without the permission of AGRE.<sup>50</sup>

AGRE also set out to maintain strong relationships with families who participated in the collection. Unlike any other gene bank developed, AGRE families were able to participate from the comfort of their home since the blood draw, clinical assessment, and diagnostic evaluations were all conducted in the family’s home. For AGRE, families are considered essential partners for the success of the program and have been described as

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<sup>50</sup> Scientist Interview #18 - AGRE Researcher Liaison



the “real heroes” and “the heart and soul” of the program (CAN, 2005a). This detailed and comprehensive relationship with the families has allowed CAN and now Autism Speaks to go back to AGRE families and get additional information that researchers request for future studies. As many researchers agree, this has been extremely useful since there are many gaps and holes in the clinical data and new results generate new questions that often require additional information from the families. A new addition to AGRE in this regard has been the development of an Online System for Clinical Research (OSCR), a tool designed to accelerate the pace of research and keep families involved in the research process. One component of this system consists of a series of online questionnaires that families can fill out and whose responses can be shared with scientists quickly. Thus, the relationship between families of AGRE and the scientists has been fostered through the grass roots efforts of placing families first during the development of AGRE and continues through efforts like OSCR. Sustaining research donor involvement over time is a rare genre of scientific work.

### *Limitations of AGRE*

Although the researchers in this study recognized the value and importance of the AGRE database, they also acknowledged some of the scientific limitations of AGRE, including the quality of the data itself and the bias that exists in the sample. The incomplete phenotypic data available for each family is an issue that reflects the history of AGRE, the development of new diagnostic tools, and the evolution of new theories involved in the etiology of autism. Since families have been collected for over a period of 12 years, some families have not provided data for newer diagnostic tools, such as the

Social Responsive Scale, or phenotypic information, such as head circumference. Furthermore, co-morbidities such as epilepsy or gastro intestinal issues are not collected reliably or systematically, limiting the number of samples researchers can use in their studies.

To illustrate the limitations of the phenotypic data, one scientist went through the AGRE data and tried to isolate a “*squeaky clean set*” of families who have at least two children that met strict ADI and ADOS criteria. Families were also excluded if the children with autism had certain medical conditions such as dysmorphology or if the mothers took certain medication during pregnancy. Since there is so much missing data they ended up with only sixty families, which is typically not enough to conduct a useful genetic study. Thus, despite efforts on the part of AGRE to go back to families and collect more data, several of the researchers agreed that there were many “*gaps and holes*” in the data, “*many more than anybody wants to admit*”.<sup>51</sup> To a large degree, the “*messy data*” is also a result of the heterogeneity of autism spectrum disorders, an issue addressed in detail in chapter three.

The AGRE samples are biased as well. First, unlike typical collections generated through recruitment at a clinical setting, the AGRE does most of their recruiting on-line. Thus, families are actively seeking participation and self-selecting to participate in the AGRE database. Such “*passive recruiting*” or “*opportunistic ascertainment*” has resulted in a sample collection that consists largely of Caucasians (85%) and of families with a higher socioeconomic status. As Heath and colleagues (2004) have pointed out, the

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<sup>51</sup> Scientist Interview #8 – Human Geneticist

widening of the “digital divide” expands of technoscientific literacy among many, which further increases the inclusion and isolation of those without access (pg.156). One scientist I interviewed reflected that his experience in recruiting Latino families for his own study (separate from AGRE) presented many barriers such as fear of having a child with a disability and immigration status, as well as the feasibility of traveling somewhere to participate in a research study. Furthermore, the diagnostic instruments used for autism are limited to English-speaking families and only a few phenotype measures are reliable and valid in other languages.

These biases in the sample can have major scientific implications, especially since the AGRE database is a major resource that scientists use to conduct their research. As one researcher pointed out, “*whenever you start with a group of families, if that's what everybody is using and if the families turn out for whatever reasons to be atypical then it'll lead us down a path that might not be helpful*”.<sup>52</sup> It is evident that the AGRE samples are not representative of the United States population, a major criticism of biomedical research with regard to the limited participation of racial and ethnic minority populations. In an effort to address these very serious limitations of the AGRE, current efforts are underway to expand the ethnic diversity of the AGRE population by targeting recruitment efforts in African American communities and bilingual families. However, a new study showed that even after traditional barriers to research participation were addressed in an African American community, 67% of reachable families were disqualified from participation because of family structure alone (i.e., no siblings or no full siblings, only

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<sup>52</sup> Scientist Interview #1 – Human Geneticist

one parent or one parent available) (Hilton, et al., 2010). These findings challenge researchers to reconsider how to conduct genetics research within the changing and diverse family structures of the twenty first century.

### ***Future of AGRE***

The future goals of AGRE are to continue to recruit more diverse families and to make the data more accessible. Vital to its success is the continued development of more public/private partnerships and collaborations with researchers that can offer some kind of support for doing additional data collection. The AGRE model is also being implemented in other countries to help establish the prevalence of autism internationally and possibly the genetic risk, especially in other countries with more homogeneous populations. The ultimate goal is to establish a comprehensive biological understanding of autism. Most importantly, however, according to the managing director of the AGRE program, *“we have a responsibility to accommodate the emerging needs of science and if that requires us to change our model, then that's what we have to do.... So we really have to kind of meet the needs of science.”*

Thus, the AGRE collection is very much a science driven enterprise even though a parent advocacy group originally initiated it and governs it. In sum, the AGRE was developed based on what scientists needed at the time in order to speed up autism genetics research and it will change depending on the future needs of science. This is also evident in a new autism genetic collection, the Simons Simplex Collection (SSC), which is recruiting 3000 families with only one child diagnosed with autism. Like the AGRE collection, the SSC is funded by a private foundation and was developed based on the

current needs of science, which, I argue in chapter three, are driven largely by emerging microarray technologies. Details of the SSC collection will be discussed in chapter four, along with the experiences of families who have participated in the SSC study.

### **Scientist vs. Parent-Initiated Research Agendas**

A major difference between the establishment of the AGP and the AGRE lies in their origins. At the onset, the AGP was initiated by scientists but supported by parent advocacy groups. In contrast, the AGRE was a parent driven initiative initially established by approaching scientists to understand what was needed in order to progress autism genetics research. Although both required partnerships and collaborations between the parent advocacy group and the scientists, their processes and outcomes were very different.

The processes underlying the AGP relied first on the ability of scientists to trust one another. The initial AGC meeting funded by NAAR and the NLM Foundation was framed internationally and explicitly allowed scientific relationships and trust to be established. Thus, NAAR was able to break down some of the competitive boundaries of science by funding initiatives that brought scientists together to form collaborations. The funding of the AGC and the AGP by the NIH was pivotal to the success of these projects and marked the beginning of future private and public partnerships that have been essential elements in the advancement of autism research. A collaboration of this size attracted new talent and expanded the knowledge base contributing to autism genetics.

However, depending on whom you ask, the results of the AGP are mixed. To date, there have only been two major publications from the AGP, and the experiments

conducted cannot be replicated since it is the largest sample used for genetics research, and constitutes many of the largest samples collected throughout the world, including the AGRE. This calls into question the balance that needs to be made between having everything in one collection versus having several different collections with very distinct populations so that findings can be generalized from one collection to another. The quality of the data is also under considerable scrutiny, which many argue will affect the results. Furthermore, the slow pace of large consortiums and the idea of “*lowest common denominator science*”, in many regards, go against the initial goals of NAAR.

Optimist regarding the AGP view the results of phase one as essential in understanding the heterogeneity of autism genetics and the identification of copy number variants (CNV) as an emergent set of genetic knowledges. Many scientists in the field of autism genetics, as well as other disease specific genetics research agendas, are now pursuing the CNV hypothesis. Thus, the AGP has been influential on two fronts. First, they established the largest scientific consortium dedicated to a specific disease. As a result, other large consortiums have followed suit, such as the International Schizophrenia Consortium. Second, the results of the AGP have been influential to genetic understandings of autism and have influenced the future direction of autism genetics research. For example, the AGP publication of phase 1 (Szatmari, et al., 2007) has been cited over 300 times within the scientific literature.

The AGRE database was a parent driven initiative that was derived based on the advice of scientists as to what could be done to advance autism genetics research. Unlike the AGP project, there was much doubt and skepticism on the part of the scientists as to whether a parent advocacy groups could produce a “scientific” worthy database. The

parents of CAN, in many regards, relied on the expertise of the scientists to determine how best to establish a database by enlisting their help along the way with scientific protocols such as informed consent processes and data collection techniques. Without doubt, the AGRE collection was developed to benefit science, but it was based on a strong partnership between families and scientists. Like the AGP, a major criticism of this project is the quality of the samples, both regarding phenotypic measurements and the Caucasian bias within the sample. However, unlike the AGP, the AGRE is a single collection, and they have a strong relationship with the families who participated. Thus, they are able to return to families to gather additional data as needed, and continue to work towards growing the collection and increasing its racial and ethnic diversity.

The amount of scientific knowledge generated by the AGRE collection is undeniable. There have been over 160 peer-reviewed publications that utilized the AGRE database, not all of which are strictly dedicated to autism genetics. Thus, the database has been transformed into a working tool that scientists use to generate knowledge. It has created what Latour (1987) refers to an “obligatory passage point”, where in the start, scientists were skeptical of parents and their ability to create a quality database (Latour, 1987). Now, it has been described as “indispensable” for many scientists, rendering the “passage” by use of the AGRE an obligation in order to conduct autism genetics research. Chloe Silverman also makes this point in her research on CAN and the AGRE (Silverman, 2008a).

The AGRE has allowed scientists to “*focus on just doing science*” and opened up the field of autism research to broader scientific interests, smaller labs, and new investigators. Most importantly, the AGRE collection shifted the collaboration and data

sharing policies, which has influenced national data sharing initiatives. The idea of a publically available database open to any qualified researcher went against the normative workings of scientific discovery at the time. However, once the AGRE was up and running, many scientists lined up to receive AGRE samples with the stipulation that they would provide AGRE will all the raw data they generated. Any publication that utilized AGRE samples also has to acknowledge the AGRE families in the acknowledgement section.

The AGRE has also launched an international effort to help establish prevalence rates of ASDs in other countries and possibly genetic risk assessments in countries with more homogeneous populations. However, Bridget Chamak details the historical role of the French autism parents' associations and shows that despite their adoption of American models of classification and intervention,<sup>53</sup> there was much resistance by French professionals who still widely used psychoanalysis and were not supportive of behavioral or educational methods (Chamak, 2008). Thus, the parents' association failed to modify autism intervention on a large scale in France. Similar challenges may emerge in the application of the AGRE model internationally, which will have to address cultural, social and professional boundaries in the diagnosis, intervention, and research on autism.

These efforts highlight the collaboration of parent advocates with scientists in pursuing genetics research and the expansion of funding to support new and innovative

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<sup>53</sup> Chamak describes three different generations of French parent associations, with the third (i.e., Autisme France) being the most active. They defined autism as a genetic disorder involving an atypical development of the nervous system, implying that bad parenting was not the cause autism (Chamak, 2008).



research. In both the AGRE and the AGP, there is an overriding push towards understanding autism as a genetic condition. Furthermore, it is evident that the AGRE has been a major site of knowledge production since the collection has been the source of data for hundreds of studies. Likewise, the AGP has been an integral part of the initiation of the scientific response to consider the role of copy number variants and the genetic heterogeneity autism. Thus, the AGRE and the AGP strongly support basic and clinical researchers and serve as two major genetic knowledge producing enterprises.

### **Health Social Movements**

Social movements based on health and disease-based collectivities are proliferating throughout Western countries, including the United States, United Kingdom and France (Epstein, 2008). There has been wide range of social scientific studies on the politics of patient group activism vis-à-vis diseases such as AIDS (Barbot, 2006; Epstein, 1996), breast cancer (Klawiter, 1999, 2004, 2008; Kolker, 2004; McCormick, Brown, & Zavestoski, 2003; Thomson, 2009), Alzheimer's disease (Baird, et al., 2006), muscular dystrophy (Rabeharisoa, 2006; Rabeharisoa & Callon, 2002), cystic fibrosis (Stockdale, 1999) and pseudoxanthoma elasticum (PXE) (Heath, et al., 2004; Novas, 2006). Given this wide range of scholarship, there are many different interpretations of what constitutes a health social movement depending on the politics surrounding the disease, its severity and stigma, treatments available, age of onset, clinical implications, and the state of knowledge of the disease. Furthermore, there are various practices of representation by which spokespersons come to stand for a group, which in many cases are not necessarily the patients.

Health social movements (HSMs) have been described as “collective challenges to medical policy, public health policy and politics, belief systems, research and practice, which include an array of formal and informal organizations, supporters, networks of cooperation and media” (Brown & Zavestoski, 2004, p. 679). HSMs by this definition challenge political power, professional authority, and personal and collective identity. Autism Health Social Movements that exists in the actions of NAAR and CAN, and now Autism Speaks, are reflective of health social movements with respect to creating public awareness campaigns, fundraising events, enhancing family services, influencing policy, and establishing governmental relations and partnerships with scientists. Some of the more unique aspects of the autism health social movements compared to health social movements of the past have been within the research agendas of these organizations. These include: promoting cross-disciplinary cooperation among scientists, which is evident in the establishment of the AGP; organizing research initiatives such as the AGRE and AGP; and establishing standards for data collection, which emerged as a necessary goal during the process of generating and pooling large samples (e.g., AGRE and AGP). Thus, their ability to collectively challenge scientists to advance autism research *and* practice through these efforts stand out as exceptional aspects of this movement.

### ***Autism Health Social Movements***

Between NAAR and CAN, and now Autism Speaks, there are a plethora of examples that justifies the broader autism movement initiated by these parent advocacy groups to be framed within a health social movement.

*Awareness Campaigns.* Awareness campaigns have taken on many forms within these autism parent advocacy organizations. For example NAAR and CAN partnered with several other national autism organizations and the Center for Disease Control and Prevention (CDC) in launching *Learn the Signs, Act Early* campaign, designed to help healthcare providers and parents identify signs of developmental disorders earlier, including autism, hearing loss, and cerebral palsy.<sup>54</sup> In 2006, Autism Speaks launched a public service announcement campaign through the Ad Council in order to communicate the high prevalence of ASDs and encourage families to learn about early signs of autism. As of March 2010, the Ad Council has received over \$210 million in donated media.<sup>55</sup> There have also been countless appearances of parent advocates on shows like “The View”, “The Today Show”, “Good Morning America”, “The Oprah Winfrey Show”, and many others. One of the most substantial awareness efforts was the creation of the annual United Nations World Autism Awareness Day, which started on April 2, 2008.<sup>56</sup> It is a global effort to heighten awareness of autism, one of three official disease-specific United Nations Days.

*Family Services.* Enhancing a range of family services has also been a priority for these advocacy groups. NAAR and CAN offered newsletters to keep families informed of the organizations activities. NAAR launched the *NAARRATIVE*, which provided

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<sup>54</sup> Autism Speaks: NAAR news archive NAAR partners on new CDC awareness campaign. Retrieved February 15, 2010, from [http://www.autismspeaks.org/inthenews/naar\\_archive/naar\\_cdc\\_awareness\\_campaign.php](http://www.autismspeaks.org/inthenews/naar_archive/naar_cdc_awareness_campaign.php).

<sup>55</sup> Autism-Speaks: Autism Speaks to celebrate third annual world autism awareness day with partners around the world. Retrieved April 22, 2010, from [http://www.autismspeaks.org/press/world\\_autism\\_awareness\\_day\\_2010.php](http://www.autismspeaks.org/press/world_autism_awareness_day_2010.php).

<sup>56</sup> Autism-Speaks: Autism Speaks applauds the state of Qatar for the creation of Annual United Nations World Autism Awareness Day. Retrieved January 23, 2010, from [http://www.autismspeaks.org/press/qatar\\_un\\_world\\_autism\\_day.php](http://www.autismspeaks.org/press/qatar_un_world_autism_day.php).

information regarding autism biomedical research, NAAR's mission and philosophy, and what parents could do to participate and strengthen their cause (NAARRATIVE, 1997). CAN provided AGRE families with a yearly newsletter that provided AGRE updates, introductions to staff members, fundraising progress, scientific achievements and a section that highlighted stories of families who participated in AGRE. More recently, Autism Speaks developed an autism video glossary to help parents and teachers learn early signs of autism and a 100 Day Kit created specifically for newly diagnosed families to make the best possible use of the 100 days following the diagnosis of autism.<sup>57</sup> In 2007, Autism Speaks also launched the Family Services Community Grants, which provide funding to organizations involved in building services for individuals with autism and expanding the capacity to effectively serve the autism community. Since 2007, they have funded 71 grants totaling close to \$1.4 million.<sup>58</sup>

*Fundraising.* Fundraising events have been a mainstay for these parent advocacy groups as well. In addition to the annual Walk Now for Autism,<sup>59</sup> which is a grassroots fundraising effort powered by volunteers and families of children with autism, these advocacy groups have initiated very creative ways of raising money and have used their Hollywood and political connections to draw a lot of awareness and fund raising opportunities for autism research and services. For example, there have been fundraising

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<sup>57</sup> Autism Speaks: Family Services 100 Day Kit. Retrieved February 18, 2010, from [http://www.autismspeaks.org/community/family\\_services/100\\_day\\_kit.php](http://www.autismspeaks.org/community/family_services/100_day_kit.php).

<sup>58</sup> Autism Speaks: Family Services Community Grants. Retrieved February 18, 2010, from [http://www.autismspeaks.org/community/family\\_services/community\\_grants.php](http://www.autismspeaks.org/community/family_services/community_grants.php).

<sup>59</sup> CAN sponsored the WALK NOW annual fundraiser and NAAR sponsored the Walk F.A.R. for NAAR annual fundraisers. Since their merge with Autism Speaks, the annual walk has been renamed to Walk Now for Autism. Each year these walks attract tens of thousands of people who raise millions for biomedical research on autism.

benefits showcasing famous comedians and/or performers, celebrity sports challenges, as well as collaborations with corporate sponsors such as Toys R Us, TJ Maxx, Kellogg, NASCAR, Chevrolet, Bank of America, and Barnes and Noble.

*Establishing partnerships and policy.* The success of CAN and NAAR, and now Autism Speaks, also lies in their ability to establish strong relationships with the government in order to secure federal legislation that advances the government's response to autism. For example, these groups were pivotal in initiating and pushing for the 2006 passage of the Combating Autism Act, the first-ever autism-specific legislation that authorized nearly \$1 billion for autism biomedical and environmental research, surveillance, awareness and early identification ("Combating autism act of 2006," 2006). The Act represents years of dedicated effort by parents and families, bringing legislative action to confront the increasing prevalence of autism. The history of such legislation began with Cure Autism Now's grassroots leadership of the Advancement in Pediatric Autism Research Act, which later became Title 1 of the Children's Health Act of 2000, which NAAR also helped draft and promote<sup>60</sup>. Jon Shestack, co-founder of Cure Autism Now, regarded this legislation as, "a federal declaration of war on the epidemic of autism". He contends that it created "a congressionally mandated roadmap for a federal assault on autism, including requirements for strategic planning, budget transparency,

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<sup>60</sup> The Children's Health Act authorizes the Secretary of Health and Human Services to conduct certain activities relevant to autism and pervasive developmental disorders, including: expansion, intensification, and coordination of activities of the National Institutes of Health (NIH) with respect to research on autism; developmental disabilities surveillance and research programs; information and education; establishment of an Interagency Autism Coordinating Committee; and an annual report provided to Congress. For more information see (DHHS, 2003).

Congressional oversight, and a substantial role for parents of children with autism in the federal decision-making process”.<sup>61</sup>

The passage of the CAA is among many legislative agendas that have been initiated by Autism Speaks, CAN and NAAR.<sup>62</sup> Over the last several years Autism Speaks has also been an advocate for insurance reform to require insurers to cover evidence-based, medically necessary autism treatments and therapies, such as behavioral health therapies like Applied Behavior Analysis (ABA). To date, fifteen states have passed autism insurance reform and eighteen states have endorsed autism insurance reform bills.<sup>63</sup> In 2007, Autism Speaks launched the “Autism Votes” website, which is a grassroots advocacy program, that coordinates activist efforts. It allows parents and other activist to stay instantly informed of state and federal initiatives dedicated to autism, and resources on how to contact state legislators.<sup>64</sup> More recently, the Autism Treatment Acceleration Act of 2009 (ATAA) was introduced to Congress and features provisions for federal reform of autism insurance coverage.<sup>65</sup> To make sure this bill is given priority, Autism Speaks has launched an Ad Council campaign urging Congressional support for this bill to end autism insurance discrimination.

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<sup>61</sup> Autism-Speaks: Cure Autism Now and Autism Speaks applaud approval of the Combating Autism Act by United States House of Representatives. Retrieved February 3, 2010, from [http://www.autismspeaks.org/inthenews/can\\_archive/caa\\_approval\\_house.php](http://www.autismspeaks.org/inthenews/can_archive/caa_approval_house.php).

<sup>62</sup> For a list of current federal initiatives specific to autism see Autism Votes website: [http://www.autismvotes.org/site/c.frKNI3PCImE/b.3909865/k.F405/Federal\\_Initiatives.htm](http://www.autismvotes.org/site/c.frKNI3PCImE/b.3909865/k.F405/Federal_Initiatives.htm)

<sup>63</sup> For a list of states that have passed and endorsed autism insurance reforms bills see Autism Votes website:

[http://www.autismvotes.org/site/c.frKNI3PCImE/b.3909861/k.B9DF/State\\_Initiatives.htm](http://www.autismvotes.org/site/c.frKNI3PCImE/b.3909861/k.B9DF/State_Initiatives.htm)

<sup>64</sup> Autism Votes home page. Retrieved February 18, 2010, from <http://www.autismvotes.org/site/c.frKNI3PCImE/b.3909853/k.BE44/Home.htm>

<sup>65</sup> Autism Votes: Autism Treatment Acceleration Act of 2009 - federal autism insurance reform (S. 819, H.R. 2413). Retrieved February 18, 2010, from <http://www.autismvotes.org/site/c.frKNI3PCImE/b.4784269/k.C0E5/ATAA.htm>.

### *Emotional Health Movements*

It was like a force was driving me, something I couldn't resist even if I tried. All parents have it -- basically you will do anything to save your child - Portia Iversen<sup>66</sup>

Among the theoretical models of HSMs proposed by Brown and Zavestoski (2004) is the “embodied health movement”, which recognizes how illness experiences challenge science on etiology, diagnosis and/or prevention. An expansion of the “embodied health movement” also highlights the collaboration of activists with scientists and health professionals in pursuing treatment, prevention, research and expanded funding (Brown, et al., 2004). For example, Deborah Heath describes what I would consider to be an embodied health movement in the context of genetic knowledge in her study on Marfan syndrome, a heritable connective tissue condition (Heath, 1998). She shows how the emphasis on lived experience intersects with the need for phenotypic markers to identify Marfan’s patients, support groups that serve as a source of medical information, and the push towards understanding Marfan syndrome as a genetic condition. Heath argues that these practices by Marfan activists “materially constitute key factors of biomedical knowledge production, supporting the work of basic and clinical researchers” (Heath, 1998, p. 83).

Yet the work of CAN and NAAR are not quite captured in the framework of an “embodied health movement” mainly because the movement for advancing the biological understanding of autism did not emerge from the individual experiences of people with autism. Rather, the movement emerged from emotional experiences of parents who have a child diagnosed with autism and their motivation to change the direction of autism

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<sup>66</sup> Quotation taken from the article “Shattering the shell: Autism breakthrough” (Friedman, 2007).

research. As indicated by the quote above, this motivation was driven by their desire to help their children in any way possible. The lived experiences are not necessarily those of the autistic individuals, but rather, the emotional experiences of parents of a child diagnosed with autism. Thus, personal connection families have to autism, although different than what may be expected from people experiencing autism, performs the same function (Steuernagel & Barnett, 2007). Silverman (2004) makes this point precisely in her research on autism. A major theme of her work is the role of affect, especially love, in “constituting knowledge, in establishing subjects of knowledge, and in establishing and stabilizing epistemological communities” (p. 51). Silverman uses the term “affect” in reference to “passions”, “the neural structure of emotions”, and “the confusing and enabling properties of love” that are embraced by scientists, parents, and practitioners “working” on autism (Silverman, 2004, p. 4).

The embodiment of individuals with ASD, at least among the health social movements of CAN, NAAR and Autism Speaks, have largely been through the promotion of materials that contain images of children on the autism spectrum. For example, current brochures created by Autism Speaks are decorated with images of young children (mostly boys) whose eyes are generally shifted away from the camera. These images are a reflection of the social deficits used to diagnose autism and the fact that boys are more likely (4:1) to get a diagnosis compared to girls. The isolation and focused interests often associated with autism is also represented in a different Autism Speaks brochure handed out at the 2006 Georgia Autism Walk, which depicts a child sitting in the corner alone and another child fixated on a toy train. A more recent Autism Speaks brochure, handed out at the 2009 Autism Society for America meeting in



Georgia, has images of parents with their children to perhaps demonstrate the affectionate nature of many individuals on the autism spectrum. This “public mediation of intimate difference” (Heath et al., 2004, p.157) rarely depicts images of autistic adults, who are typically not part of social and scientific discourses surrounding biomedical research.

Unlike the case of Marfan’s, the lived experiences are also not driving the phenotypic markers that define autism in the Diagnostic and Statistical Manual of Mental Disorders (DSM) (APA, 2000). The symptoms that distinguish autism spectrum disorders are based on behavioral characteristics and on specific criteria determined by the DSM. Typically, diagnostic instruments for autism rely on two main sources of information: descriptions of caregivers of the course of development (e.g., ADI-R), and current behavior and communication patterns and information from direct observation of behavior (e.g., ADOS).<sup>67</sup> Thus, the phenotypic characteristics that define autism spectrum disorder are based on external observations and parental experiences, which Majia Nadesan argues must be understood in relation to a matrix of professional and parental practices, and institutions that have enabled the identification and interpretation of autism (Nadesan, 2005). This matrix of practices and institutions has clearly left out the “lived” experience of those on the autism spectrum.

In many regards, the production of autism genetic knowledge lies at the intersection of multiple phenotypic characteristics defined by the DSM and genetic markers identified through various genetic technologies. This is especially apparent in

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<sup>67</sup> Although the ADOS and ADI-R are among the most used diagnostic instruments for ASD, there are many other instruments used as part of the diagnosis such as: the Aberrant Behavioral Checklist, the Child Behavior Checklist, the Developmental Coordination Disorder Questionnaire, the Peabody Picture Vocabulary Test, the Raven’s Standard Progressive Matrices, the Social Communication Questionnaire, and the Social Responsiveness Scale.

research that focuses on the “genetic reclassification” of disease based on autism sub-phenotypes, which is discussed in chapter three. Thus, what constitutes key factors of genetic knowledge in autism research are the “observed” ASD symptoms of either professionals who diagnose autism or parents who participate in genetics research studies, as well as the genotype information generated through various genetic technologies. However, it must be noted that the “observed” symptoms documented and used as phenotypic data are predetermined and essentially follow the DSM diagnostic criteria (APA, 2000).

Similar to embodied health movements, however, were the parents’ drive and motivation to collaborate with scientists and health professionals in pursuing treatment, prevention, research and expanded funding. For example, NAAR, CAN and now Autism Speaks have provided unprecedented amounts of funding for several areas of autism research, mainly in brain and behavioral mechanisms, genetics, diagnosis, and treatment (Singh, et al., 2009). This helped to open up the field to a lot of researchers then outside of the field of autism and provided opportunities for young investigators to begin their careers in autism research. It also leveraged additional investments by the NIH and other governmental agencies.

### ***Advancing the Research and Science of Autism***

The ability of CAN and NAAR to promote genetics research through the funding and development of the AGRE and the AGP demonstrates the “force” autism parent advocacy groups enabled in order to move the science of autism genetics forward. These initiatives are exemplars of the four specific agendas promoted by these groups,

including: initiating cross-disciplinary cooperation; funding investigator initiated research; organizing research initiatives; and establishing standards for data collection and management to benefit the scientific community.<sup>68</sup> However, more examples exist, that move beyond genetics research and into a broader and quite unique autism research initiatives.

*Organize and promote research.* A prime example of CAN and NAAR's ability to organize and promote autism research was their joint collaboration with the UC Davis MIND Institute<sup>69</sup> to launch the first interdisciplinary autism research conference in 2001. The International Meeting for Autism Research (IMFAR) represented the first time that an interdisciplinary conference was held for researchers involved with all aspects of autism research. The goals of the annual conference were to accelerate global research efforts and recruit prominent researchers in related fields who were not involved in autism research. Until this meeting, investigators interested in autism research attended one or more meetings that were geared to their area of expertise. The idea for a scientific meeting specific to autism research was conceived by Portia Iversen, co-founder of Cure Autism Now, who realized after attending an annual Society for Neuroscience meeting that the few autism researchers presenting at the meeting were not aware of each other and she had to personally introduce them herself. Thus, the IMFAR conference offers

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<sup>68</sup> Autism-Speaks: Science Overview. Retrieved February 18, 2010, from <http://www.autismspeaks.org/science/overview.php>

<sup>69</sup> The UC Davis MIND Institute is a collaborative international research center committed to improving the awareness, understanding, prevention, care and cure of neurodevelopmental disorders. It was established in 1998 by six families, five whom have sons with autism, who had the vision to gather experts from every discipline related to the brain working together under one roof and working toward a common goal of curing neurodevelopmental disorders, starting with autism (UC Davis M.I.N.D. Institute. Retrieved February 5, 2010, from <http://www.ucdmc.ucdavis.edu/mindinstitute/aboutus/index.html>).

ASD researchers from around the world a focused opportunity to share the rapidly moving scientific investigation of ASD. It has clearly been a site for establishing collaborations among scientists and knowledge production and has grown into the largest international meeting dedicated to autism research. Each year the meeting gets larger, and the 2009 meeting, held in Chicago, IL, drew over 1,500 researchers, delegates, autism specialists, and students from around the world.

The scientists I interviewed repeatedly acknowledged the work of parents advocacy groups like CAN and NAAR and their tireless efforts to promote autism research and funding. They described parents of autism as “*smart, resourceful, and creative*”, “*organized and aggressive*”, and “*very visible and available*” compared to other childhood disease advocacy groups. In their opinion, the parents were *the* driving force in establishing and forming the field of autism research. In fact, one scientist who utilizes the AGRE samples for his own research nominated Portia Iversen, the co-founder CAN, for a McArthur Genius Award. “*If you've ever met her,*” he stated, “*she is a genius with no scientific training. She has as much scientific information and understanding of science as anybody who I've met. She's amazing. But she also had this remarkable perspective on how to advance the scientific field without being a scientist.*”<sup>70</sup>

*Initiate Research Collaborations.* Another common theme that the scientist recognized in the work of CAN and NAAR was the ability of parents to put a lot of pressure on researchers to collaborate. This is evident in the establishment of the Autism Genome Project, as well as other collaborative endeavors such as the Autism Treatment Network (ATN), the Interactive Autism Network (IAN), and the Autism Clinical Trials

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<sup>70</sup> Scientist Interview #13 - Neuroscientist

Network (ACTN)<sup>71</sup>. These networks bring together hospitals and physicians (ATN), families of autism and researchers (IAN), and treatment centers throughout the United States (ACTN). As one scientist remarked, “*I think the word community really was infused by the advocates. It's all about community and I think that advocates of autism really infused that noun into the formula.*”<sup>72</sup> Thus, their pressure on scientists to work together as a community as opposed to individually made a huge difference in the direction autism research, and not just in genetics research. These collaborative efforts also established multidisciplinary approaches involving, for example, basic scientists, geneticists, and clinical investigators working together to think about how to best serve families and push research forward at the same time.

### **A Reversal of Power**

The traditional model for advocate-supported research foundations goes something like this: Raise money, turn it over to a higher power, and don't ask questions. This was not a model I could devote my life to. Portia Iverson in *Strange Son* (2006, p.32)

We must have our collective voices heard by relevant governmental agencies, Congress and the scientific community and be viewed by them as partners engaged in a common mission – Karen London (NAARATIVE, 1997, p. 3)

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<sup>71</sup> The Autism Treatment Network (ATN) is the nation's first network of hospitals and physicians dedicated to developing a model of comprehensive medical care for children and adolescents with autism. The Interactive Autism Network (IAN) is an innovative online project bringing together tens of thousands of people nationwide affected by autism spectrum disorders (ASD) and hundreds of researchers in a search for answers. The Autism Clinical Trials Network (ACTN) is a collaboration of treatment and research centers dedicated to accelerating clinical trials of investigational treatments for autism and to increasing the number of biological treatments available to families and clinicians.

<sup>72</sup> Scientist Interview #13 - Neuroscientist

It is apparent in the vision provided by Portia Iversen and the ideals the Londons generated in developing NAAR that their efforts would be based on partnerships with scientists, personal ownership, and governance of the direction of autism research. The “partnership model” proposed by Rabeharisoa (2003), reflects the work of CAN and NAAR nicely especially with regard to the shift in the balance of power. In the “partnership model”, Rabeharisoa, characterizes the patient organization as the master of its research policy and patients as specialists in their own right (Rabeharisoa, 2003). The first characteristic demonstrates a reversal of traditional power relations between the patient organizations’ board of governors and its scientific council, where the former is in total control of its research policy and the latter is an advisory body whose opinions are subject to approval by the board. The second characteristic places patients and their families as specialist partners in the production of knowledge and in the care and treatment of their disease (Rabeharisoa, 2003).

By examining similarities and differences between AGRE and AGP, I distinguish the relative importance (and power) of different stakeholders in the production of genetic knowledge. Furthermore, and perhaps more importantly, the development of AGRE and the AGP are two examples of how parent advocacy groups, rather than the patients themselves, are promoting and contributing to genetic knowledge of autism. In essence, these parent advocacy groups are representing a somewhat “voiceless” community of individuals or implicated actors comprised mainly of children with autism.

For both CAN and NAAR, the reversal of power was apparent given that these organizations took ownership of the direction of autism research to consider the biological origins of autism. Chloe Silverman also asserts that the power differential

between advocacy groups and government agencies was essential for CAN and NAAR to succeed in altering the format of autism genetics research and creating an alternative culture for cooperative research on autism genetics (Silverman, 2004). However, the specifics of these power differentials are different due to the distinct natures of the AGP and AGRE projects. Since CAN developed the AGRE database from the ground up, they own and govern the database themselves. The shift in data collection and sharing policies was a result of CAN's insistence on making the AGRE database openly available to any qualified researcher. For the AGP, not one group owns the AGP collection. Instead, each collaborator who contributed samples has jurisdiction over the samples they collected (i.e., they can conduct research and publish at will) but cannot utilize the AGP collection for studies outside the AGP. Nor does the AGP distribute samples; it only pooled different samples in order to conduct a larger genetic study. Although there are plans to make the AGP collection publically available, it still remains in the control of the AGP scientists. However, in the event that results from Phase I or II lead to the identification of a genetic variant or mutation that contributes to autism, the AGP Memorandum of Agreement states that NAAR (now Autism Speaks) would take the lead on any issue of intellectual property rights (Szatmari, 2005). Thus, differential stakeholder power emerged in these two genetic research initiatives. For CAN/AGRE, the power was clearly group initiated and remains within the parent organization itself. In contrast, the AGP comprises a mixture of power relations among the scientists themselves and with the parent organization.

In both CAN and NAAR, parents of children with autism were on the Board of Directors and Trustees. Currently, Autism Speaks has twelve members on the Board of

Directors who have a family member with autism. Several staff members are also parents of children with autism, many of whom transformed their talents towards the needs of the organization.<sup>73</sup> For example, Peter Bell joined Cure Autism Now Foundation in 2004 as Executive Director and CEO after a successful 12-year marketing career at McNeil Consumer & Specialty Pharmaceuticals. He led the foundation's funding total to \$39 million, enhanced the foundation's research, education and outreach initiatives, and expanded the foundation's treatment portfolio.<sup>74</sup> Thus, the work of these parent organizations went beyond raising and spending money. They became a new breed of parent advocate, one who fueled their board and staff with parents and families of autism that could sit at the table with scientists, health professionals, and government officials and set research agendas.

CAN also recruited a scientific advisory board (SAB) composed of prestigious researchers and clinicians representing a myriad of disciplines relevant to autism research. Their primary responsibility was to review the grants submitted for scientific merit. However, in 2000 the power of the SAB was augmented by Portia Iverson and Dr. David Baskin who put a different mechanism in place after experiencing the frustration of watching seemingly beneficial proposals passed over because of concerns such as diagnostic procedures, statistical power or the lack of experience of the investigator. They formed the Scientific Review Council, which played a unique role in setting the direction of the CAN's scientific research and leveraging research dollars. The Council was

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<sup>73</sup> For example, the Executive Vice President of Program and Services, the Director of Family Services, and the Executive Vice President of Awareness Events of Autism Speaks all have children on the autism spectrum.

<sup>74</sup> Autism Speaks: Autism Speaks leadership. Retrieved February 19, 2010 from <http://www.autismspeaks.org/leadership.php#peter>.



comprised of parents or other family members of people with autism who are also researchers or physicians, whose personal dedication and relevant expertise helped the SAB prioritize Cure Autism Now's research goals, objectives, and initiatives. The Council brought to the process the ability to determine whether the science funded was relevant, and whether it represented a balanced pool of research projects.<sup>75</sup> Thus, although scientific partnerships were essential in progressing the direction of autism research, power still remained within the auspice of the parent organization.

These parents also formed collaborations with scientists in order to speed up the pace of autism research. For example, CAN recruited Dan Geschwind, a scientist from UCLA, early on when they were developing the AGRE database. He was encouraged by Jon Shestack to write a grant to start developing genetic studies in autism.<sup>76</sup> Once Geschwind received NIH funding, he was granted a supplement to support the AGRE recruitment efforts (D. H. Geschwind, 2003). AGRE continues to rely on multiple collaborations with scientists to improve their data collection efforts.

NAAR established a different kind of collaboration between families of autism and scientist through what they called “Parents as Partners in Research”. Here the goal was to connect families of individuals with autism with investigators conducting clinical autism research studies. In their view, it was critical that families participate in research studies funded by the NIH. A similar approach is now underway under the auspice of Autism Speaks, called the Interactive Autism Network (IAN). NAAR also took much pride in their Scientific Advisory Board (SAB), describing it as “one of the finest

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<sup>75</sup> Autism Speaks: CAN news archive Cure Autism Now scientific advisory board meetings held in Santa Monica, California. Retrieved February 15, 2010, from [http://www.autismspeaks.org/science/science\\_news/board\\_meetings\\_santa\\_monica.php](http://www.autismspeaks.org/science/science_news/board_meetings_santa_monica.php).

<sup>76</sup> Scientist Interview #18 - AGRE Researcher Liaison

Scientific Advisory Boards in the country” and “the jewel of this organization” (NAARATIVE, 1997, p.3). The SAB determined which scientific proposals were funded and brought scientific legitimacy to the organization. Thus, for NAAR, the power among the stakeholders remains distributed across the scientists and the parent organization. To the degree that the scientists are regarded as ones “jewels” of the organization, greater power was within the scientists’ court. CAN on the other hand, in the case of AGRE, places the “heart and soul” of the program in the families and places much discretion regarding future research in integrating governing councils and policies that place them in control of the future direction of research.

Echoing the second criterion of Rabeharisoa’s “partnership model”, parents involved in CAN also became specialists in their own right in order to better understand the potentials and limitations of science. Portia Iversen writes in her book about how she got a tutor in basic science and molecular biology, and read countless scientific articles to “piece together a picture of the state of autism research” (Iverson, 2006, p.36). It was during this research that she came across a small group of genetic studies done in the 1970’s and 80’s that showed an increased risk of autism in identical twins. Hence, the motivation to establish the AGRE database was born. Both Portia Iversen and Jon Shestack appear as authors on the paper discussing the AGRE project (D.H. Geschwind, et al., 2001) and were critical stakeholders in the success of this project. This is parallel in the case of pseudoxanthoma elasticum (PXE)<sup>77</sup> and the work of parent advocate Sharon Terry, who was directly involved in the discovery of the PXE gene and appeared as one of the scientific authors for the discovery and patent of the PXE gene (E. Marshall, 2004).

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<sup>77</sup> PXE is a rare genetic disorder that can result in skin lesions, blindness and even early death through hardening of the arteries or gastrointestinal bleeding (Terry & Boyd, 2001).

Like CAN, the PXE International created research consortia and patient registries, initiated several clinical studies and in 1996, established the PXE International Blood and Tissue Bank, which was privately held, supported and maintained by PXE International (Heath, et al., 2004; Novas, 2006; Stockdale & Terry, 2002; Terry & Boyd, 2001).

In contrast with Rabeharisoa's "partnership model", however, the autism genetics movement is not based in the lived experience of the patients, as discussed previously. Instead, parents were the laypersons that developed scientific expertise to better communicate with scientists and negotiate with government officials. However, NAAR was co-founded by two psychiatrists, a corporate lawyer and a professor of chemistry at Princeton University. In many regards, NAAR followed a similar model to that used in the initiation and discovery of the Huntington disease (HD) gene. Founded by a family whose mother suffered and died of Huntington's disease,<sup>78</sup> the Hereditary Disease Foundation initiated HD research through interdisciplinary workshops, collaborative efforts, and promoted a very high degree of cooperation between families and investigators; one daughter herself changed careers to become a scientist in the field (A. Wexler, 1996).<sup>79</sup> The leaders of the Hereditary Disease Foundation were both situated in the academic and scientific world, and transformed their specialties towards the biomedical approach to HD. Similarly, Eric London, the co-founder of NAAR, is a psychiatrist by training who was able to use his scientific training and experiences as a

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<sup>78</sup> Huntington's disease (HD) is a movement disorder that causes uncontrollable jerking and writhing movements of all parts of the body.

<sup>79</sup> The Huntington's Disease Foundation also organized clinical workshops centered on DNA banking and in 1983, established the world's first DNA bank of families with HD (A. Wexler, 1996; N. Wexler, 1992).

parent to successfully establish the first advocacy organization dedicated to autism research (London, 1997).

### **Infusing Community into Science**

The autism health social movements demonstrate the successes of parent advocacy groups in influencing priorities and practices of biomedical research and the active role they have played in science through fund raising, lobbying, participating in research priorities, donating specimens, and organizing scientist to conduct autism research. NAAR and CAN, and now Autism Speaks have become significant authorities in the engagement of health and well-being of individuals with autisms, direct contributors to the production of biomedical knowledge, and specifically, in the case of AGRE and AGP, initiators in the production of genetic knowledge. However, unlike PXE and Huntington's disease advocacy groups, whose efforts have resulted in the identification of "a gene" that causes disease, the autism genetics movement is in many ways reshaping the way in which the "genetics" for any complex disease is being constructed. Rather than a single gene causing autism, genetic contributions to ASDs are much more complex, and likely involve multiple genetic interactions with the environment. If anything, the push towards genetic understandings of autisms through efforts like the AGRE and the AGP have made the biomedical understanding of autisms more complicated and the stakes much higher given the personal and financial investments of families, scientists, and governments over the last sixteen years.

For the AGRE project, the stakes seem much higher for the families, given their donation of blood and family health information. The investments made by families have

clearly influenced the priorities and practices of biomedical research and have created a perhaps unprecedented degree of cooperation between families and researchers. Furthermore, as already demonstrated in the case of PXE International and the French Muscular Dystrophy Organization (AFM), the funding and establishment of the AGRE database allowed CAN (and now Autism Speaks) to privately hold, support, oversee and maintain the AGRE database, as well as determine certain direction of genetics research (Heath, et al., 2004; Novas, 2006; Rabeharisoa, 2006; Stockdale & Terry, 2002; Terry & Boyd, 2001).

For the AGP, the stakes seem much higher for the scientists involved in the consortium, since the family collections are from different research studies throughout the world, including the AGRE database. Furthermore, the AGP was an investigator-initiated project whose establishment was based on the failure of individual labs to identify genetic mechanisms involved in autism. The success of the AGP does not ride on the families who participated but rather on the scientists involved in the project.

There are unique aspects of the autism health social movements described in this chapter that are not necessarily articulated in either the “embodied health movement” (Brown, et al., 2004) or the “partnership model” (Rabeharisoa, 2003). First, the lived experiences of individuals with ASD are not central to advancing the biomedical understanding of autism. Rather, it is the emotional experiences of parents and families of ASD that are challenging science on etiology, diagnosis and/or prevention of disease. This type of knowledge is similar to what Susan Lindee describes as “emotional knowledge” (Lindee, 2005). In her research on familial dysautonomia (FD), Lindee describes families of FD as a “social and medical conglomerate” that sought out, or as

she describes it, “collected” scientific experts. Their emotional knowledge, she argues, “does not just provide comfort and pain. It also produces scientific papers and gene maps” (Lindee, 2005, p.179). Similarly, the emotional knowledge of parents and families of ASD and their strong desire to help their children were the “embodied” experiences driving biomedical research on autism. These experiences have produced genetic databases and research consortiums that are enabling the production of genetic knowledge.

Perhaps a better way of theoretically conceptualizing this phenomenon is to refer to what Heath and colleagues describe as “genetic citizenship” (Heath, Rapp, and Taussig, 2004). Their concept of genetic citizenship describes a complex and multi-sited network of associations that link lay health activist, clinicians, scientist, politicians, and corporate interests in the collective formation of the public sphere. It also represents a diverse array of nonhuman actors, such as genes and molecules implicated in particular diseases and the technologies used to study them (Heath, et al., 2004, p.154). By forging these alliances, these authors argue that genetic advocacy groups are “making citizenship claims on behalf of their genetically vulnerable offspring” (Heath, et al., 2004, p.155).

The parent advocates of CAN and NAAR and now Autism Speaks clearly established networks of associations among clinical and basic researchers, policy makers, governmental agencies, and families of children with autism. The non-human actors, such as the AGRE, the collection of DNA and phenotypic samples combined through the AGP, and various emerging technologies such as microarrays, are also closely linked to these networks of associations. These human actors and non-human actors represent the

dispersed power relations and cultural-technical alliances that characterize the geneticization of contemporary life science and social life (Heath, et al., 2004).

The concern with technoscientific development and application by patient activists is also described as an “emergent concerned group” (Callon & Rabeharisoa, 2008). In their study of the French Association of individuals suffering from muscular dystrophies, Callon and Rabeharisoa demonstrate how patients were able to construct their individual and collective identities based on the association’s intense engagement in scientific and technological research activities, an engagement, which they argue “enabled them to change their ontological status” (Callon and Rabeharisoa, 2008, p. 231). These active forms of citizenship engaged in scientific and technological research activities reflect the “ontological status” of parent advocates in CAN and NAAR. The diagnosis of their child was not met with passivity, but rather activism based on subjective and emotional knowledge in order to direct research towards a cure.<sup>80</sup>

Another feature of the parent advocates described in this chapter is their ability to transform their talents to fit the needs of promoting biomedical research on autism. This is evident of the founders of CAN, Jon Shestack and Portia Iverson, who are quoted as saying “[p]eople told us in the beginning you can't hurry science. Well, you can. You

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<sup>80</sup> Anne Kerr also considers some of the tensions around the “new genetic citizenship” that are articulated in professional discourses and practices in the clinic, as well as in wider policy making networks (Kerr, 2003). Kerr contends, “models of genetic citizenship involve several processes of mediation between the obligations, immunities, and entitlements of professionals, clients and publics where the production, application and regulation of genetic information are concerned” (Kerr, 2003:48). These inter-linked processes that privilege professional entitlements and patient or public obligations show parallels of past models of genetic citizenship, namely the lead role of professionals in determining policies about genetic research and services and patient/public responsibility for eliminating hereditary disease. Kerr argues that this runs counter to the conclusions of other studies that tend to highlight novel gene technologies and their role in transforming patients’ experiences of health and their bodies (Kerr, 2003).

really can. You can treat it like a low-budget movie and make it go fast. And that's what we've done" (Bazell, 2005). These parents transformed their skills of being Hollywood professionals into “producers” of genetic knowledge. Similarly, Eric London was able to use his scientific training as a psychiatrist and tapped into his medical resources to establish the first advocacy organization dedicated to autism research. These transformations are also evident within the workings of the advocacy organizations that recruited parents of children with ASD who have expertise in medicine, law, science, marketing, and health informatics.

The emphasis placed on families in the collection of biomedical data and participation in autism research is also an emerging activity for the autism health social movements. In the case of AGRE, the family centered approach through the in-home collection of data and the AGRE newsletters, created a strong relationship and commitment between AGRE and the families who are part of the collection. This relationship has enabled researchers to request additional information from families if needed. It also created additional biovalue to the existing collection (Novas, 2006). The approach taken by NAAR of encouraging families to participate in NIH research initiatives through newsletters, was also a prelude to the current efforts by Autism Speaks to bring together people nationwide affected by ASD with autism researchers through the Interactive Autism Network (IAN). This project is capitalizing on the ability to gather information electronically and provides another example of the promotion of new technologies by parent advocacy groups. As Heath and colleagues also point out, “the internet has provided novel possibilities for translocal engagements and intimacies, and



for sharing of both biomedical knowledge and life experiences among advocates, scientists, and clinicians.” (Heath, et al., 2004, p.155)

The ability of both CAN and NAAR to leverage their pilot projects into millions of dollars over time in autism research awards by the National Institutes of Health (NIH) and other governmental funding sources also created a sustainable research endeavor on autism. It also established a level of legitimacy of their research goals within the broader scientific community. This success relied on their ability to establish strong relationships with the government in order to secure federal legislation that advances the government’s responses to autism. The ability to enact public policy such as the Children’s Health Act of 2000 and the Combating Autism Act are direct results of action taken by autism parent advocacy groups. The convergence between the needs of families with ASD and congressional commitments represents another aspect of “genetic citizenship” (Heath, et al., 2004).

Finally, the shift in scientific practices of data sharing policies initiated through AGRE and collaborations among a diverse set of scientists in efforts like AGP have been transformative and influential in the current conduct of genome science. The AGRE data sharing policies have likely influenced the development of national research databases, such as the NIMH Genetics Initiative and the National Database for Autism Research, which both initiate and promote data sharing and collaboration among scientists. The ability of the AGP to effectively collaborate internationally with multiple groups of scientists has influenced the development of additional autism

consortium, such as the Boston Autism Consortium,<sup>81</sup> as well as other psychiatric disease-based research consortiums (e.g., schizophrenia). The future of AGRE and AGP will also rely on the ability to developed collaborative efforts with new investigators both within and outside of autism-based research. These collaborations will likely extend beyond the academic and non-profit world to medical venture capital firms who are currently entering the autism field.<sup>82</sup>

Within the broader health social movements of autism discussed in this chapter, there are also new and innovative ways CAN, NAAR and now Autism Speaks are using cultural resonant and viable frames in discursive activities to redefine autism from a rare childhood disease to a major public health issue (Kolker, 2004). First, well-known people who have children with autism have entered the public sphere, such as actress Holly Robinson-Peete who is on the Autism Speaks board of directors and a public spokesperson for autism. Singer Toni Braxton is also featured on a public service announcement for autism awareness that emphasizes the prevalence of autism. These famous parents are essentially ambassadors for Autism Speaks, who raise awareness through their celebrity status and personal experiences of having a child with autism.

The innovative ways Autism Speaks has created autism awareness through the Ad Council campaigns, Utube videos, and public appearances in major media venues have allowed them to claim autism as an epidemic. Every discursive form of awareness

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<sup>81</sup> Autism Consortium. Retrieved April 23, 2010, from <http://www.autismconsortium.org/about-us/about-us.html>.

<sup>82</sup> Bob Wright emphasized this point throughout many his appearances on World Autism Day, April 2, 2010. He stated that translational research has captured the interest of venture capital and medical venture capital firms in the last four to five months and predicts progress in this area within the year (CNBC: Squawk on the street: World Autism Awareness Day. Podcast retrieved from <http://classic.cnbc.com/id/15840232?video=1457305587&play=1>).

emphasizes the increased prevalence of autism, which is currently at 1 in 110 children diagnosed with autism. For example, an Autism Speaks brochure handed out at a 2009 Autism Society for America Meeting states, “Autism is the nation’s fastest-growing serious developmental disorder. It has reached *epidemic* proportions. In 1993, 1 in 10,000 child was diagnosed with autism. Today that number is 1 in 150. Every 20 minutes another child is diagnosed.” (Autism-Speaks, 2008b) (my emphasis). Under this prevalence estimate (1 in 150), 67 children are diagnosed per day, amounting to “more children...diagnosed with autism this year than with AIDS, diabetes and pediatric cancer combined.” (Autism-Speaks, 2008b). These discursive messages are continually repeated through the awareness campaigns of Autism Speaks bringing the autism “epidemic” to the public sphere. Furthermore, the awareness of autism is now reaching international scale with the establishment of the United Nations World Autism Awareness Day on April 2.

Autism Speaks also utilizes social networking technologies such as Facebook, Twitter, Ning, YouTube, and has an official Autism Speaks Blog, which allows for “technosocial mediation of intimate differences” made routinely available to the public through electronic means (Heath et al., 2004, p.157). This new generation of technological mediations keeps autism at the forefront of social networking. It has also been a space for different autism “cultures of action” to articulate each other’s positions, disagreements, and disputes. Other projects, such as Autism Votes, electronically alerts advocates to participate in political pressure to enact legislation that favors research, health and educational support for autism. These new forms of social networking or what Rose (2007) refers to as “informational biocitizenship” or “digital citizenship” (pg.135),

will undoubtedly influence the future of health social movements and should be a focused area social scientific inquiry in the future.

In sum, these parent organizations have redefined how autism research is initiated, funded and changed through policies regarding the future of autism research and practice both in the United States and countries abroad. Their push towards a genetic understanding of autism through the development of collaborations, consortia and collections has fueled autism research towards a mainstream research agenda that investigates autism as a genetic and neurological disease. However, the question - what is causing the increase in prevalence of autism spectrum disorders - remains unanswered. In the next chapter, I describe some of the underlying findings of genetics research to date and how the AGRE and AGP have contributed to genetic knowledge of autism. It will also take a closer look at the interpretations of autism genetics from the perspective of the scientists involved in the AGP and/or who utilize the AGRE database.

## CHAPTER 3: GENETIC RECLASSIFICATION OF AUTISM SPECTRUM DISORDERS

Over the last decade, there has been a major increase in the awareness and prevalence of autism spectrum disorders (ASD). Efforts to identify the causes and etiologies of this disorder have been unprecedented, particularly in genetics research (Singh et al., 2007, 2009). Because genetics research has been of interest to both public and private stakeholders, an unprecedented amount of resources has become available to conduct autism genetics research. The institutionalization of autism genetics research has emerged through the development of autism specific genetic databases, such as the Autism Genetic Resource Exchange (AGRE), and of international genetic research collaborations such as the Autism Genome Project (AGP)(discussed in detail in chapter two, see also Silverman, 2004, 2008). Furthermore, public and private funding for autism genetics research, including the most recent \$60 million offered through the American Recovery and Reinvestment Act of 2009,<sup>83</sup> has catapulted autism to a new frontier of genetic knowledge production.<sup>84</sup>

Despite growing social and scientific investments in autism genetics, the production, representations and implications of genetic knowledge of autism are not well understood. The representations of autism genetics through technoscientific research

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<sup>83</sup> American Recovery and Reinvestment Act of 2009 (ARRA). Retrieved November 15, 2009, from <http://grants.nih.gov/recovery/>.

<sup>84</sup> Chloe Silverman's research on autism carefully articulates the "economies of government research funding, status, prestige, and markers of research promise and success" to show why genetics research is so heavily funded and how different parties contribute to and participate in this process (Silverman, 2004, p. 292).

could have a profound influence on how autism is ‘imagined’, defined, and treated in the future. These representations are often viewed through a technical lens that incorporates laboratory technologies such as whole genome microarray analysis and/or DNA sequencing. These technologies help to identify the specific chromosomal locations of potential ‘disease alleles’ and in essence create new meanings of the blood, behavioral characteristics and medical information supplied by families, vital to the production of genetic knowledge.

To analyze the production of genetic knowledge within the social world of autism genetics, this chapter describes the historical transformations in autism genetics research, the research challenges in ASD that are prompting new genetically constructed meanings of autism, and new knowledge producing technologies that are shifting the genetic disease paradigm from inherited single gene causing mutations to rare genetic variants that are spontaneously acquired. Specifically, this chapter highlights three ways in which autism is being redefined based on genetic knowledge: identification of copy number variants; genetic reclassification of autism phenotypes; and convergence of common biological pathways. The social implications of these changes in scientific knowledge are then elucidated. In closing, this chapter analyzes how scientists are imagining the future of autism through their research and reconstructing autism as a genetic disorder.

### ***Situating Autism Genetics***

To situate the field of autism genetics, a brief review of the political support for autism research is warranted because it establishes the public investment in autism

research in general and genetics research more specifically.<sup>85</sup> As described in previous reports (Singh, et al., 2007; Singh, et al., 2009), there was a dramatic increase in autism research funding starting in 1997, coinciding with the establishment of Collaborative Programs of Excellence in Autism (CPEAs) by the National Institutes of Health (NIH) (National Institute of Health (NIH), 2006). From 1997 to 2007, this U.S. research initiative invested \$105 million towards eight CPEAs to “conduct research to learn about the possible causes of autism, including genetic, immunological, and environmental factors”.<sup>86</sup> Nine CPEAs were established during this time and six of these centers had a genetic focus that concentrated mainly in the search for “genes and functional domains within genes that are likely sites of disease-related mutations”.<sup>87</sup>

The Children’s Health Act of 2000 also mandated the expansion of autism research activities through the establishment of the Studies to Advance Autism Research and Treatment (STAART) Centers, which was a \$65 million investment to conduct basic and clinical research.<sup>86</sup> The Children’s Act also required the establishment of the Interagency Autism Coordinating Committee (IACC) to coordinate all efforts within the U.S. Department of Health and Human Services (DHHS) concerning autism research (DHHS, 2003). Also, transpiring from this work was the development of the National Database for Autism Research, a national resource that collects, stores, and distributes

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<sup>85</sup> Chloe Silverman also points out in her research on autism the significance of governmental activities and how they affect the “activities of participants via regulation, review of products, safety legislation, but also as sources of funding for research and as the site at which public health programs are designed and implicated” (Silverman, 2004, p. 280).

<sup>86</sup> Collaborative Programs of Excellence in Autism (CPEAs)/Studies to Advance Autism Research and Treatment (STAART) Centers. Retrieved November 14, 2009, from [http://www.nichd.nih.gov/research/supported/cpea\\_staart.cfm](http://www.nichd.nih.gov/research/supported/cpea_staart.cfm).

<sup>87</sup> The Collaborative Programs of Excellence in Autism (CPEAs). Retrieved November 14, 2009, from <http://www.nichd.nih.gov/autism/research/cpea.cfm>.

blood samples, cell lines and genetic materials very broadly across the scientific community.<sup>88</sup> These events were significant given the constraints on the NIH budget (Mervis, 2007) and, as described in chapter two, were influenced by the strong political momentum of autism advocacy groups to promote research and awareness of autism spectrum disorders.

The current public agenda for autism research continues to support these efforts largely through the passage of the Combating Autism Act of 2006 ("Combating autism act of 2006," 2006). Under this new law, approximately \$950 million will be allocated to autism over five years, doubling expenditures on existing programs. As a result of this act, the CPEAs and STAART programs consolidated under the Autism Centers for Excellence (ACE) to establish six ACE centers (single site) and five ACE networks (multiple sites) throughout the United States. Although these grants support a broad range of autism research, two of the main goals are to identify rare genetic variants and mutations, and to make associations between autism-related genes and physical traits.<sup>89</sup>The Combating Autism Act of 2006 also re-authorized the IACC to develop and implement a strategic plan for autism research and a budget to fund this plan (DHHS, 2009c). This is particularly important since the IACC strategic plan was used as a guide for the recent grants funded through the American Recovery and Rehabilitation Act (ARRA) of 2009. Autism was the only disease specifically earmarked by the ARRA, which had committed approximately \$30 million towards autism genetics research to

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<sup>88</sup> National Database for Autism Research. Retrieved November 14 2009, from <http://ndar.nih.gov/ndarpublicweb/home.go>.

<sup>89</sup> NIH News: Newly awarded autism centers of excellence to further autism research. Retrieved November 14, 2009, from <http://www.nih.gov/news/health/apr2008/nimh-01.htm>.



conduct full sequencing of target genes and whole-genome sequencing for a few individuals with ASD.<sup>90</sup>

Given this backdrop of public support for autism research and its success in generating support, it is not surprising that many scientists jumped on the autism research “bandwagon” (Fujimura, 1996). In addition, private sources from parent advocacy groups such as Autism Speaks have committed up to \$89 million towards autism research since 1997.<sup>91</sup> The annual budget for NIH research dollars supporting autism research has increased fivefold since 1997, from \$22 to \$118 million in 2008.<sup>92</sup> A review of private and public funded research from 1997 to 2006 shows a steady increase in the number of autism research projects, with genetics research second only to research on brain and behavioral mechanisms (Singh, et al., 2009). The funding of autism genetics research is also reflected in the scientific literature, where genetics research articles are among the most published peer-reviewed research topic in autism (Matson & LoVullo, 2009). Chloe Silverman also highlights that “disorders become genetic through the production of texts and papers which certify them as such, as well as research programs which serve to certify the validity of this framing” (Silverman, 2004, p. 291).

The heightened exposure towards identifying the genetic underpinnings of autism has also resulted in a languished focus of identifying the multiplicity of causes and consequences of autism. For those who see autism as an environmentally mediated illness, the genetic approach to autism has in a sense “betrayed autistic children” by

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<sup>90</sup> Simons Foundation Autism Research Initiative (SFARI): NIH director has big plans for autism research. Retrieved October 28, 2009, from <https://sfari.org/news-and-commentary/all>.

<sup>91</sup> Autism Speaks: What we fund and how we fund it. Retrieved November 19, 2009, from <http://www.autismspeaks.org/science/research/grants/index.php>.

<sup>92</sup> NIH: Estimates of funding for various research, condition, and disease categories (RCDC). Retrieved November 14, 2009, 2009, from <http://report.nih.gov/rcdc/categories/Default.aspx>.

assuming autism in terms of unchangeable and genetically determined (Herbert & Silverman, 2003). These authors argue that advances in biomedical treatments for autism will only occur by moving beyond a gene-brain paradigm and by allocating financial investments to physiological and toxicological autism research (Herbert & Silverman, 2003). To achieve this, Martha Herbert, who a pediatric neurologist and brain development researcher at Massachusetts general hospital, argues for research that embraces a broader systems-organism biology perspective and an expansion from a strongly genetic disease paradigm to a genetically influenced and gene-environmental interaction perspective (Herbert, 2005).

Margaret Lock also challenges the assumption of genetic determinism in her research on late on-set Alzheimer's disease (Lock, 2005). Like Herbert, Lock argues for a critical form of epigenetics (i.e., research that focuses on gene-environmental interaction) known as "developmental systems theory", which gives priority to dynamic interactions among many variables with numerous possible outcomes (Lock, 2005, p. S52)<sup>93</sup>. However, Lock takes her notion of environment a step further, to include social influences such as the effects of human relationships over a life span. While these approaches to investigating disease etiologies are not new, they have attracted far less attention (and funding) than research activities on the genetics of autism. These critiques and others (Duster, 2003; Lippman, 1992; Shostak, 2003) call attention to the implications of focusing on the genetic contribution to disease to the point that it obfuscates research directed toward the social and environmental causes of illness.

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<sup>93</sup> For an historical description of developmental systems theory, see (Fujimura, 2005).

## Implications of Funding Autism Research

The landscape of public and private investment in autism research and the continued commitment to set research priorities for autism at the national level are key components fueling the scientific drive in autism research. For scientist conducting autism genetics research, particularly those who utilize the AGRE database and/or are involved in the Autism Genome Project (details provided in chapter two), funding for autism research has impacted the field immensely by providing opportunities for new scientists and broader experts in the field to focus on autism research. For example, several of the participants who utilized the AGRE database would not be in this field if funding and the AGRE samples were not available. The availability of resources such as the AGRE opens up the field to smaller labs and new researchers entering the field. Funding also enables researchers outside of autism to re-shift their focus. As one scientist states, *“some of the best researchers not in autism but some of the best neural scientists and functional biologists and geneticists and such.... come to the table simply by virtue of money.”*<sup>94</sup>

However, as with any scientific endeavor, there are unintended consequences, which, for better or worse, can affect the direction of research. As one autism researcher lamented, who has been in this field prior to the 1997 boost in funding:

Be careful what you wish for... I mean it was great when there started to be more money for research but it has these, I think unintended consequences. When there's too much money it's also not always a great thing.<sup>95</sup>

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<sup>94</sup> Scientist Interview #1 – Human geneticist

<sup>95</sup> Scientist Interview #19 – Statistical geneticist

What this particular scientist was referring to, and what other autism genetic scientists agree on, is how the political nature of the funding for autism has directed the type of genetics research that is being conducted. Funding priorities generally focus on large consortia instead of smaller individual labs, and the use of newer and faster technologies such as microarrays and genome wide associations studies. Thus, certain types of research are being prioritized without careful consideration of whether this is the best method for researching the genetics of autism. Furthermore, one scientist worries that the uniqueness of basic science is being lost due to government and public demands to translate and apply genetic knowledge. He states, “*We scientists are playing into it. We are saying yes, if you give us money we can find something for autism. And I'm actually not quite so sure that we can promise that.*”<sup>96</sup> This scientist was reflecting on the push by funders for scientists to “translate” genetics research findings into clinical practice, when in reality they are conducting basic science, which is far from any clinical application. Despite this scientific reality, scientists are making promises that they are unlikely able to keep. As discussed below, the complexity of autism spectrum disorders has also inhibited broad clinical applications. However the genetic components of autism are still being sought based on newer technologies and the push for larger and more characterized samples.

### ***Autism Genetics Past***

We must, then, assume that these children have come into the world with innate inability to form the usual, biologically provided affective contact with people, just as other children come into the world with innate physical or intellectual handicaps. (Kanner, 1943, p. 250)

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<sup>96</sup> Scientist Interview #1 – Human geneticist

Leo Kanner's first descriptions of eleven children with "inborn autistic disturbances of affective contact" are strikingly similar to the diagnostic criteria used for autism today, including the "inability to relate themselves in the ordinary way to people and situations", "anxiously obsessive desire for the maintenance of sameness", "monotonously repetitious", "the children's relation to people is altogether different" (Kanner, 1943). Kanner's first descriptions also implied a biological origin. However psychoanalytic thought was at the time increasingly popular in North America and Europe, pinpointing autism as a form of psychosis akin to childhood schizophrenia and a developmental anomaly ascribed exclusively to maternal emotional determinants, (i.e., the refrigerator mother theory) (Nadesan, 2005; Silverman, 2004). The cause of autism being attributed to mothering styles was promoted by Freudian child psychologist, Bruno Bettelheim, in his book *The Empty Fortress: Infantile Autism and the Birth of the Self* (Bettelheim, 1967). Thus, the refrigerator mother theory and subsequent forms of treatment (e.g., psychoanalysis) were the prevailing ideas about the origins and course of autism in both professional and popular social discourses well into the late 1970's (Nadesan, 2005; Silverman, 2004). This shift started in 1964 by Bernard Rimland, a psychologist and parent of a child with autism, through the publication of his book *Infantile Autism: The Syndrome and Its Implications for a Neural Theory of Behavior*. He was among the first to challenge the psychogenic theory of autism and played a central role through scientific work and activism to frame autism as a medical condition based on apparent neurological features of autism (Rimland, 1964).

In the 1970's, some of the first studies on the etiology of autism occurred that demonstrated autism was associated with neurological disease in children with congenital

rubella (Chess, 1971, 1977), as well as other medical conditions (Coleman, 1976). In 1977, the first systematic twin study was conducted that demonstrated autism to be highly heritable (Folstein & Rutter, 1977). It also indicated that it probably extended beyond the traditional diagnosis of autism to include a broader range of social and communicative deficits in individuals of normal intelligence (Folstein & Rutter, 1977). In the 1980's autism was further associated with a variety of chromosomal disorders and rare genetic syndromes such as Fragile X (Gillberg & Wahlstrom, 1985; Wahlstrom, Gillberg, & Gustavson, 1986). These studies were the first strands of evidence that genetic factors played an important part in autism.

Thus, the current understanding that autism has a genetic etiology has a rather short history, starting only in the 1980's. This shift was in conjunction to the official classification of "infantile autism" as a subclass of "pervasive developmental disorders" in the Diagnostic and Statistical Manual of Mental Health Disorders 3<sup>rd</sup> edition (APA, 1980). This was the first recognition that autism was a separate but related disorder to schizophrenia and was a new diagnosis that served as a pathway into the history of the medical knowledge and treatment of autism (Brown, 2000). Like depression and other mental health disorders, the biomedical categorization of this condition has been a "critical juncture" in the process of constructing autism as a genetic disease (Shostak, et al., 2008). The institutional stabilization of phenotypes and the constant re-framing and expansion of ASD through various versions of the DSM has been an essential element in the production of genetic knowledge. The heterogeneity of the ASD phenotype and the potential genes underlying this wide spectrum of symptoms, as well as the technologies that identify them have constituted a compelling target for scientific research, as we shall

see next.

### **In Search of the “Autism Gene”**

At the time when scientists were contemplating the nature of autism genetics, there was a consensus early on that the genetics of autism would be relatively straightforward due to the high concordance rates in identical (or monozygotic) twins that ranged from 50 – 90% compared to fraternal (or dizygotic) twins that ranged from 0-10% (Bailey, et al., 1995; Folstein & Rutter, 1977; Steffenburg, et al., 1989). The consolidation of these genetic findings translated into a heritability<sup>97</sup> of autism of about 90%. Scientists argued that this made autism the most “strongly genetically influenced of all multifactorial child psychiatric disorders” (Rutter, 1995, p. 177). Chloe Silverman (2004) also points out how the successes of common diseases such as Phenylketonuria (PKU) and sickle cell trait served as “emblems for the possibility of clear gene identification and clean-cut medical targets” (Silverman, 2004, p. 302). Furthermore, the identification and cloning of the Rett’s syndrome gene, MECP2, was among the most significant of these emblems in autism research since Rett’s syndrome is classified in the DSM as one of the pervasive developmental disorders, linking it to autism (Silverman, 2004, p. 302).

Based on the strong “genetic liability”<sup>98</sup> of autism more than any other psychiatric disease, there was a belief among many scientists that one or maybe two major genes were involved in the majority of autism cases. In fact, several of the scientists

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<sup>97</sup> Heritability is the proportion of phenotypic variation that is attributable to inherited genetic factors (in contrast to environmental ones). Because heritability is a proportion, its numerical value will range from 0.0 (genes do not contribute at all to phenotypic individual differences) to 1.0 (genes are the only reason for individual differences).

<sup>98</sup> Genetic liability a term used throughout the field of genetics research and refers to the degree genetics is responsible for human disease or other conditions.

interviewed for this study entered the field thinking that with the technologies available at the time and the strong heritability of autism, “the gene for autism” would be identified quickly and would likely lead to a better understanding of the etiology autism. However, what has transpired over the years is a very different story. As one scientist describes it:

It's become abundantly clear..... When you add our research into everybody else's research and look at it as a whole that the simple answer to autism genetics doesn't exist and you know there was a lot of feeling early on that there would be just a few, you know, one or two or a few major genes. Clearly not the case.<sup>99</sup>

Thus, the initial optimism for finding the “few major genes” for autism was short lived and new scientific and technical approaches ensued as a result.

### **Autism Genetics Present**

The medical and scientific literatures often assume autism is some *thing or things*, some essential biogenetic condition(s), which will ultimately be unequivocally identified and known as a spatially centered genetic, neurological, or chemical abnormality through the efforts of scientists toiling in their laboratories” (Nadesan, 2005, pg.2).

The efforts of scientists “toiling in their laboratories” over the past thirty years have scientifically constructed autism to be a heterogeneous disorder that most likely involves many genes interacting with each other and with multiple environmental exposures, most of which have yet to be identified. According to a recent review on autism genetics, “genetic linkage studies have failed to identify a single chromosomal region of strong effect” (Abrahams & Geschwind, 2008). In other words, after almost 30 years of research using traditional genetic research technologies, in this case over a dozen

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<sup>99</sup> Scientist Interview #8 – Human geneticist



linkage studies,<sup>100</sup> there is not one region in the entire genome that provides evidence of a major gene involved in autism. Instead, these studies reveal numerous “suggestive” linkage peaks but with relatively little congruence across them and limited compelling evidence for replication (Losh, Sullivan, Trembath, & Piven, 2008).

Instead of a major gene causing autism, the current data suggests that a small percentage of ASDs are caused by rare genetic mutations (e.g., Neurologin 4, PTEN, and Contactin Associated Protein-Like, other genetic syndromes (e.g., Fragile X and tuber sclerosis), and *de novo* (non-inherited) copy number variations (e.g., 16p.11.2, 7q, and 22q13), which together account for about 10-20% of ASD cases (Abrahams & Geschwind, 2008). However, some clinical experts consider this estimate high and the causes of the remaining 80-90% of autism cases are still unknown. The current state of autism genetics, according to the scientists themselves, is reflected in recent report on genome-wide linkage and association scans for autism. The authors stated, “*although autism is a highly heritable neurodevelopmental disorder, attempts to identify specific susceptibility genes have thus far met with limited success*” (Weiss et al., 2009, p. 802). This sentiment was lamented by many of the scientists I interviewed who felt their research contributions to the understanding of autism genetics was “*very limited*”, “*minimal*” and provided “*no final conclusions*”.

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<sup>100</sup> A whole genome linkage study is a statistical evaluation of genetic variation throughout the genome that is used to identify polymorphic loci (multiple gene regions) that segregate with a phenotype of interest.

## **The Heterogeneity of Autism**

The obvious question to ask would be why have there not been any major advances in the field of autism genetics despite the private and public financial support of research in this area and clear motivation on the part of scientists? For scientists, one answer resides in the predominant theme of heterogeneity used to describe autism and the many challenges it places on autism genetics research. Heterogeneity exists at *both* the phenotypic and genotypic levels and it is considered a major issue in scientific papers that discuss autism (Bill & Geschwind, 2009; Hus, Pickles, Cook, Risi, & Lord, 2007; Sutcliffe, 2008; Szatmari, 1999). At the center of the heterogeneity of autism is the variability that exists in the core components of autism which, according to the DSM-IV, include: impairments in reciprocal social interaction, impairments in verbal and non verbal communication, and a pattern of repetitive, stereotypical behaviors, activities, or interests (APA, 1994).

In addition to the presence or absence of anyone of these core symptoms of ASD, there is also a considerable range of severity in cognitive functioning, verbal abilities and social skills, each of which has its own developmental trajectory and outcome (Szatmari, 1999). Furthermore, there are many co-morbidities that occur in autism that are not part of the ASD diagnosis, such as sensory abnormalities, gross motor delays, sleep disturbances, gastrointestinal disturbances, attention deficit and hyper activity disorder and epilepsy (Abrahams & Geschwind, 2008). Scientists refer to this as “phenotypic heterogeneity”, which in essence means diverse forms of an observable characteristic or trait.

As one might imagine, this degree of heterogeneity has made the investigation of autism genetics challenging because as any genetic epidemiologists will attest, a major prerequisite for a successful genetic analysis is having an accurate definition of the phenotype (e.g., Szatmari, 1999). The genetic mechanisms believed to be involved in ASD thus far are turning out to be quite complex in the absence of the identification of a major gene for autism. Scientists are referring to this as “genetic heterogeneity” meaning there is the possibility that two or more independent genetic mechanisms might lead to the disorder (Szatmari, 1999). The current NIH director, Francis Collins, summed up the genetic heterogeneity of autism in a statement at the 2009 Society for Neuroscience meeting, “Autism at the DNA level is not one disease. It may be a hundred or a thousand different diseases all of which have in common this affect on the brain”.<sup>101</sup>

The lack of identification of major genes involved in autism, and the diversity of the results of genetics research to date, namely the identification of many different chromosomal loci and genetic alleles, most of which have not been replicated, has resulted in much frustration in the autism genetics research community. Scientists describe this lack of correspondence between the ASD phenotypes and the underlying genetics as a result of “*messy phenotypic data*”, and the idea that ASD is “*essentially a dozen different disorders each of which has a genetic risk*”. One geneticist believes that scientists could end up identifying 100 or more different kinds of autism, each with a different genetic basis and each accounting for just a small percentage of the total (Wrobel, 2009). Thus, the genetic approaches to autism have been hindered by the heterogeneity of the autism phenotypes. This has lead scientists to assert that there will be

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<sup>101</sup> Simons Foundation Autism Research Initiative (SFARI): NIH director has big plans for autism research. Retrieved October 28, 2009, from <https://sfari.org/news-and-commentary/all>.

no “*quick and easy answers*” and “*that there’s going to be many answers because of the variability of the kids*”.

Despite this phenotypic heterogeneity and the challenges it poses for research, scientists are viewing the complexity of autism genetics as an opportunity to identify new genetic knowledge and develop new analytical and genomic tools and technologies in the pursuit of this knowledge (Gupta & State, 2007). Thus, the complexity of autism genetics has in many ways driven the direction of genetic study designs and the way scientists are thinking about this problem. As one scientist described it, “*working on a complex system or condition like autism, push you toward, thinking complex. Push you toward thinking out of ordinary rules of even science or genetics*”.<sup>102</sup> Despite the limited progress made in autism genetics thus far, the knowledge gained through the process of “genetic dissection” or the incremental contributions made to the biology of autism based on genetic research findings thus far, have made the pursuit of the genes for autism worthwhile for those working in this field.

### **Genomewide Association Studies (GWAS)**

To fully understand the predominant technological approaches utilized by scientists who study autism genetics and how the technologies themselves are in many ways driving the direction of research, a brief introduction of genomewide association studies (GWAS) and associated technologies is necessary. After the initial sequencing of the human genome in 2001 (Lander, et al., 2001; Venter, et al., 2001), the HapMap project was initiated to characterize patterns of common genetic variation in populations

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<sup>102</sup> Scientist Interview #20 – Molecular geneticist

around the world.<sup>103</sup> A major goal was to provide a catalogue of common variants that could be used to perform genome-wide association studies (GWAS), a test of the association between markers, called single-nucleotide polymorphisms (SNPs), across the genome and human disease.<sup>104</sup> GWAS are based on the hypothesis that common diseases, such as type 2 diabetes or breast cancer, are to a large extent caused by common genetic variants (frequency of 5% or greater in the general population). Thus, if a genetic variant increases the risk of the disease in question, it will be more common in people with common diseases than in controls. Companies like Illumina<sup>105</sup> and Affymetrix<sup>106</sup> generated the chip-based technology (i.e., microarrays) needed to cost-effectively genotype hundreds of thousands of markers from a patient's DNA sample. To date, more than 100 genomewide association studies have been conducted on numerous diseases based on information from the HapMap project using microarrays, often incorporating patient samples and research institutes from around the world (Goldstein, 2009; Hardy & Singleton, 2009). The development of this technology has been described as “the common thread in an extremely productive synergistic relationship between advances in biological understanding, computational methodology and the technological development in the arrays themselves” (LaFramboise, 2009, p. 4182). Other social science projects have examined the use of this technology in other lines of scientific work (Hedgecoe &

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<sup>103</sup> International HapMap Project: About the HapMap. Retrieved November 27, 2009, from <http://hapmap.ncbi.nlm.nih.gov/thehapmap.html.en>.

<sup>104</sup> For a thorough review of GWAS studies with regard to psychiatric diseases see: (Psychiatric-GWAS-Consortium-Coordinating-Committee, 2009)

<sup>105</sup> Illumina: BeadArray technology. Retrieved December 1, 2009, from [http://www.illumina.com/technology/beadarray\\_technology.ilmn](http://www.illumina.com/technology/beadarray_technology.ilmn).

<sup>106</sup> Affymetrix: GeneChip® mapping 10K 2.0 array. Retrieved December 1, 2009, from [http://www.affymetrix.com/products\\_services/arrays/specific/10k2.affx](http://www.affymetrix.com/products_services/arrays/specific/10k2.affx).

Martin, 2003; Shostak, 2005). For example, Sara Shostak demonstrated how microarray technology, the development of its application, and subsequent transformations contributed to the emergence of toxicogenomics (Shostak, 2005).

Despite the efforts and huge investments to uncover the common genetic variants involved in common diseases, the findings of GWAS have explained only a very small proportion of the underlying genetic contributions for most studied diseases (Kraft & Hunter, 2009). For autism, only a few variants have been identified as possible candidate genes in linkage studies (Abrahams & Geschwind, 2008) and the largest genomewide association studies conducted to date indicate that the locus-specific effects of common genetic variations are very small (Wang, et al., 2009; Weiss, Arking, The-Gene-Discovery-Project-of-Johns-Hopkins, & The-Autism-Consortium, 2009). For scientists, these findings suggest that autism is a heterogeneous and multigenic disorder and that there is a considerable amount of genetic variation that remains to be identified. These findings have also engendered a shift towards the investigation of rare *de novo* (spontaneous or not-inherited) copy number variants (CNVs). Parallel failures of explanatory power have also characterized research in other psychiatric disorders, such as schizophrenia (O'Donovan, et al., 2008; Sullivan, et al., 2008), bipolar disorder (Ferreira, et al., 2008; Sklar, et al., 2008) and major depressive disorder (Bosker, et al., 2010; Shi, et al., 2010).

## Copy Number Variation

Technological innovations have opened the door to a fundamental aspect of human genomic variation that was previously unrecognized and have opened a new window into the genetic basis of disease. Methods for detecting CNVs genome-wide have the power to identify risk factors for disease directly. (Sebat, 2007)(p. 297)

Copy number variants (CNVs) are one form of structural variation in the genome that consists of a gain or loss in a chromosomal region greater than 1 kilobase (kb) in size. Such micro deletions or duplications occur in abundance in the general population and appear widespread throughout the genome (Sebat, 2004). Due to the lack of success in linkage and association studies, as well as the development of high resolution DNA microarrays, the investigation of CNVs have become a central focus of research on autism genetics (Sebat, et al., 2007). Microarrays<sup>107</sup> are high-resolution platforms originally designed to genotype markers (i.e., SNPs) in genomewide association studies. However the increased ability for microarray technologies to scan over 500,000 markers throughout the genome has uncovered a high degree of structural variation within the genome, which researchers have regarded as important contributors to human genetic variation.

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<sup>107</sup>“Microarray” is a general term used to describe the various types of DNA array-based technologies such as array-based comparative genomic hybridization (aCGH), and arrays or bead chips that genotype single-nucleotide polymorphisms (SNP arrays). This technology is based on the biochemical principle that nucleotide bases bind to their complementary partners – specifically, A binds to T and C binds to G. Array protocols generally consist of hybridization of fragmented single-stranded DNA to arrays containing hundreds of thousands of unique nucleotide probe sequences. Each probe is designed to bind to a target DNA subsequence. Specialized equipment can produce a measure of the signal intensity associated with each probe and its target after hybridization. The underlying principle is that the signal intensity depends upon the amount of target DNA in the sample, as well as the affinity between target and probe (LaFramboise, 2009). For specific details and schematics of array-based comparative genomic hybridization see (Wain, Armour, & Tobin, 2009). Sara Shostak (2005) also provides a detailed history of the development and use of microarray technology in her sociological analysis of the emergence of toxicogenomics.

Scientists, and parent advocacy groups alike, are describing CNVs as “a new window on human genetic variation” (Abrahams and Geschwind, 2008, p. 347) and “a new theory of autism risk that stands to influence how future autism genetic research is conceptualized”.<sup>108</sup> Over the last several years high-resolution microarray platforms have identified a handful of *de novo* and inherited CNVs that are believed to be important causes of ASDs, either as rare variants that strongly modulate risk or as potentially new syndromes linked to the ASDs (Abrahams & Geschwind, 2008). Thus, the ability to quantitatively assess genetic changes at a resolution of a few hundred base pairs have made it possible for scientists to “discover” new syndromes based on the deletion or duplication of genomic segments of 500 kb to 2Mb in size. This new classification of genetic syndromes is expected to grow in the future now that current microarray platforms can scan up to one million segments of DNA distributed throughout the genome.<sup>109,110</sup> The impact and influence of CNV discoveries on autism research is already being acknowledged in the scientific community. For example, in 2009, the Interagency Autism Coordinating Committee (IACC) acknowledged four different CNV discoveries among the most important work in biomedical research (DHHS, 2010).

Although CNVs were being examined in other diseases prior to autism, several scientists regard autism at the forefront of CNV discoveries. Thus, CNVs are being exploited in other mental health disorders such as schizophrenia, bipolar disorder and

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<sup>108</sup> Autism-Speaks: Top 10 autism research events of 2007. Retrieved December 2, 2009, from [http://www.autismspeaks.org/science/science\\_news/top\\_ten\\_autism\\_research\\_events\\_2007\\_cnv.php](http://www.autismspeaks.org/science/science_news/top_ten_autism_research_events_2007_cnv.php).

<sup>109</sup> Affymetrix: Genome-wide human SNP array 6.0. Retrieved December 4, 2009, from [http://www.affymetrix.com/products\\_services/arrays/specific/genome\\_wide\\_snp6/genome\\_wide\\_snp\\_6.affx](http://www.affymetrix.com/products_services/arrays/specific/genome_wide_snp6/genome_wide_snp_6.affx).

<sup>110</sup> Illumina: Human1M-Duo DNA Analysis BeadChip Kits. Retrieved December 5, 2009, from [http://www.illumina.com/products/human1m\\_duo\\_dna\\_analysis\\_beadchip\\_kits.ilmn](http://www.illumina.com/products/human1m_duo_dna_analysis_beadchip_kits.ilmn).



attention deficit and hyperactivity disorder. Many scientists working in this field highlighted the importance of microarray technologies and their influence on the direction of science. As one scientist noted, “*the technology, as it always does, drives the science now...So we have to formulate the right questions for the technology but the technology will give us new insights into these questions*”.<sup>111</sup> As will be discussed later, scientists refer to GWAS technology as a “reverse genomic” approach since no assumptions are being made about which genes or genomic regions might be important in causing or contributing to disease. In this hypothesis free approach, gene discovery is based on data generated by newer and faster technologies that determine where the important genetic variants are located. What we can see here, then, is how “jobs” are made “right” for the “tools” available to research. Theory is not driving science (Clarke & Fujimura, 1992; Shostak, 2005).

#### ***A New Genetic Classification: 16p11.2 Deletion***

One specific CNV that has received wide spread attention in the scientific and clinical genetics community is the recurrent *de novo* (spontaneous or non-inherited) deletion on chromosomal region 16p11.2, identified using high resolution microarrays in three separate populations (Kumar, et al., 2008; C. R. Marshall, et al., 2008; Weiss, et al., 2008). This deletion spans an estimated 25 genes across 600 kb. However many of the genes and their functions are unknown. Autism Speaks acknowledged the identification of the 16p11.2 deletion as among the top ten research events of 2008 and regarded this CNV and others like it to have important implications for autism diagnosis and treatment

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<sup>111</sup> Scientist Interview #17 – Molecular geneticist

despite not knowing which genes in this region are contributing to autism and how they interact with the environment.<sup>112</sup>

To return to the theme of “technology driving the science”, one researcher described the 16p11.2 finding as “*accidental*” and a “*by product*” of the raw data produced by the microarray analysis, which is based on fluorescence intensity.<sup>113</sup> She described the study as originally designed to detect common and inherited genetic variation that may contribute to the risk of autism. However, since the fluorescence is proportionate to how many gene copies are present (i.e., genetic load), they were able to use the same data to scan the genome for micro deletions and duplications. Hence, the technology literally enabled the production of genetic knowledge. This was in essence a by-product or “*side-effect*” of the original research design. Since the 2008 publications, 16p11.2 has been considered a “hot spot” for susceptibility genes that are likely involved in autism and research efforts have shifted towards identifying and understanding the genes spanning the deleted 600 kb region.

However, the interpretations of the *de novo* 16p11.2 deletion is not strait forward since some of the deletions identified are inherited, a small percentage of deletions have been found in control samples, and some family members with this deletion have no ASD symptoms (Rosenfeld, et al., 2009; Shinawi, et al., 2009; Weiss, et al., 2008). The strict interpretation of these findings, as one scientist pointed out, is that this CNV and others like it have nothing to do with autism and that there is something else involved.<sup>114</sup>

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<sup>112</sup> Autism-Speaks: Top 10 autism research events of 2008. Retrieved December 2, 2009, from [http://www.autismspeaks.org/science/science\\_news/top\\_ten\\_autism\\_research\\_events\\_2008\\_16p11\\_cnv.php](http://www.autismspeaks.org/science/science_news/top_ten_autism_research_events_2008_16p11_cnv.php)

<sup>113</sup> Scientist Interview #11 – Human geneticist

<sup>114</sup> Scientist Interview #6 - Neurobiologist

Furthermore, two recent studies have shown that the 16p11.2 deletion is observed in approximately one of every 200 – 250 samples submitted for clinical microarray testing and is more likely to be seen in people with speech and language delays, intellectual disability, various behavioral problems, and macrocephaly; yet these specific symptoms *alone* do not constitute an autism diagnosis (Rosenfeld, et al., 2009; Shinawi, et al., 2009). To put it simply, these results demonstrate that there is *no* simple relation between the 16p11.2 deletion and ASD. To further complicate the story, recent data suggests that individuals who carry a large and rare deletion on 16p11.2 are likely to have developmental delays, be obese, or both (Bochukova, et al., 2010; Walters, et al., 2010).

Thus, these results further expand the clinical manifestations associated with this deletion, challenging scientists to reconsider specific autism phenotypes that may be associated with the 16p11.2 deletion, as well as other genetic and environmental factors that may be involved in causing full-blown autism. It also further blurs the boundaries of the normal and abnormal, diagnostic certainty and uncertainty, and the genotypic and phenotypic heterogeneity associated with ASD. Despite these unexpected findings, scientists believe the association between the CNVs and autism to be real and an important area of research to be pursued in the future. In fact, one project involving thirteen university-affiliated research clinics throughout the United States, the Simons Simplex Collection, is developing a phenotypic and genotypic database derived of families with only one child diagnosed with ASD to investigate the *de novo* CNV hypothesis.<sup>115</sup> Furthermore, with regard to clinical genetics, the 16p11.2 deletion is being

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<sup>115</sup> Simons Foundation Autism Research Initiative (SFARI): Simons Simplex Collection. Retrieved December 8, 2009, from <https://sfari.org/simons-simplex-collection>.

screened in some labs as a first tier analysis using microarray technologies, and labeled “*causative*” of the ASD diagnosis if the deletion is found.<sup>116</sup>

### ***Questionable Approach to Science***

Yet, some autism genetics researchers are skeptical of the *de novo* CNV hypothesis that has spawned from genome-wide association studies, calling it a “*hypothesis free*” or “*reverse genomic*” approach to science. That is, instead of starting with a well-characterized phenotype that segregates in families or a candidate gene that has biological relevance, the starting points are various genetic loci that have been identified through the advances in microarray and sequencing technologies. One scientist described this process as “*bootstrapping our way back to the phenotype*”,<sup>117</sup> where researchers start with a genotype and work backwards to find a clinical description. Here, scientific decisions are not being based on what makes sense biologically or whether it is grounded in empirical evidence, but rather, the degree to which “unbiased canvassing of the genome” can identify areas in the genome that contribute to the risk of autism (Losh, et al. 2009, p. 3). Furthermore, funding agencies have preferably funded research that utilizes SNP technologies for whole genome scans, not because they make sense based on epidemiology, but because the technologies are on the “cutting edge” of genetic technology (Fujimura, 1996). This has led to the predominant genetic approach that allows scientist to proceed agnostically without any *a priori* expectations as to the specific genetic information they may find. Nikolas Rose (2010) describes the GWAS

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<sup>116</sup> Scientist Interview #4 – Human cytogeneticist

<sup>117</sup> Scientist Interview #19 – Statistical geneticist

approach as “economies of scale” that have “come to the rescue and enabled the emergence of a new way of thinking, a new way of hope, a new business model” (p.71). Rose argues that this way of thinking has been central to the new buoyancy of the market in genomics and is the kind of research geared toward personal genomics (i.e., 23andme, deCodeme, and Navigenetics) (Rose, 2010).

The complexity of autism in many ways has also enabled the “buy into” microarray technologies and the CNV hypothesis because traditional genetic research approaches have failed. This is reflected by one scientist who states, “*I think people are more willing to go this --the CNV-- route because we haven't found, you know, there's no holy grail of genetics. There are a few really promising things for subsets of kids with autism and there hasn't been a single variant that's been identified for all of autism.*”<sup>118</sup> Furthermore, the lack of replication in genetic research findings based on GWAS, despite increased sample size, has produced results, which one scientist bravely admits, “*nobody really believes in*”.<sup>119</sup> This sentiment was also revealed at the 2009 International Meeting for Autism Research in a presentation describing phase 2 of the Autism Genome Project, which referred to the best evidence of association in a sample of 1500 probands as “*not that compelling*” (Sutcliffe, 2009).

These criticisms are not specific to autism genetics research but in the field of genetics research as a whole. A recent set of commentaries in *The New England Journal of Medicine* discussed the successes and failures of genome-wide association studies (GWAS) and whether to continue with these studies in light of the overall limited returns

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<sup>118</sup> Scientist Interview #6 – Neurobiologist

<sup>119</sup> Scientist Interview #19 – Statistical geneticist

(Goldstein, 2009; Hirschhorn, 2009; Kraft & Hunter, 2009). The authors grapple with the fact that after over 100 GWAS conducted to date, the findings collectively explain only a very small portion of the underlying genetic contribution to most studied diseases. Like autism, there has been a shift in the broader genetics research community in considering rare variants, either single site or structural, that would have a large effect size. Scientists argue that this would account for the missing heritability that has not panned out in the research on common variation.

Despite the challenges in the genetics research on autism, the accomplishments made thus far are viewed very optimistically in the field of autism genetics (Abrahams & Geschwind, 2008). As one scientist stated,

Five years ago we didn't have any of these clues so it's really amazing that we've got some explanations now and we've got genes that identify pathways in the brain that may be involved. So I think the momentum in the field is very, very exciting. People are really excited.<sup>120</sup>

In the flows of this excitement about the science of autism, genetics research initiatives continue to be supported both publicly through the National Institutes of Health and other public organizations, as well as through the private sector, namely large autism advocacy organizations such as Autism Speaks and the Simons Foundation. It is estimated that the NIH will contribute over 100 million dollars a year for the next five years towards autism research.

Thus, the field of autism genetics research moves forward in a space of ambiguity and uncertainty within the current understanding of genetic knowledge of autism. Scientists seem somewhat conflicted in their efforts where, on the one hand, they

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<sup>120</sup> Scientist Interview #17 – Molecular geneticist

recognize that the scientific results to date have been limited. Yet, on the other hand, they are motivated to continue their research in this area due to the serious financial investments and biomedical resources made available by both private and public funders, as well as sheer excitement of the progress made in the field. Furthermore, scientists view the lack of identifying the “autism gene” and the phenotypic and genetic heterogeneity of ASD, as motivating factors to continue down this scientific inquiry. Even the most skeptical scientists I interviewed for this study, who were critical of the GWAS approach, were still swimming within the “*belly of the beast*” (Field Notes, April 2009). In other words, for this scientist, despite being in conflict with the predominate approach to autism research (i.e., GWAS and CNVs) she is still pursuing research grants that focus on these types of studies. This is reflective of the work of Sara Shostak (2005) who highlights the imperative of scientific disciplines to participate in the “discursive practices of molecular biology, genetics, and/or genomics” in order to fully participate in the knowledge production in the contemporary life sciences (pg. 394).

The ambiguities and uncertainties of genetic knowledge has created what Joanna Latimer and colleagues (2006) describe as “more moments of interpretation” (pg. 602) and “an imperative space that helps legitimate the need for more technoscience, and consequently, more clinical judgment with which to fix the genetic future” (pg. 599). Although the current study does not investigate the genetic clinic per se, the scientists I interviewed continually “deferred” autism to a “genetic ground” despite limited success in identifying genetic mutations that contribute to the etiology of autism. Genetic technological progressions of microarray technology have clearly created a space of expanded interpretation of ASD based on new types of genetic mutations, namely the

CNVs. The majority of scientists were also anticipating (Adams, Murphy, & Clarke, 2009) the future success of microarray technologies to scan over a million markers throughout the genome, which they believed would further delineate genetic complexities of autism.

However, this was viewed as an intermediary step before the feasibility of sequencing entire genomes of a specific “diseased” population became a reality (i.e., next generation sequencing). The first human genomes in 2001 cost over \$3 billion each to decode (Lander, et al., 2001; Venter, et al., 2001). Today, the wholesale cost of whole genome sequencing is down to about \$4,500 and falling (Drmanac, et al., 2010). The scientists I interviewed regarded this as the future direction of autism genetics, as well as the need to generate larger sample sizes in order to identify rare genetic mutations with a large effect size. These goals have been further legitimized by the private and public support to conduct this type of research, as well as the development of new biomedical resources that support the *de novo* CNV hypothesis (i.e., Simons Simplex Collection discussed in chapter four). The process of “abduction” articulated by Adams and colleagues (2009) demonstrates the anticipation these scientists have in future technologies. As such, “abduction moves reasoning temporally from data gathered about the past to simulations or probabilistic anticipations of the future that in turn demand action in the present” (Adams, Mamo and Clarke, 2009, p. 255). The actions taken by scientists and parent advocacy groups to generate larger samples and set up autism specific genetic databases to test the anticipated futures echo the processes of abduction.

The speed of new genetic technologies is also moving exponentially and requires new forms of knowledge to translate the “terabytes” of information that are being



generated through this technology, namely computer bioinformatics systems (i.e., computational techniques). One scientist described the changes in genetic technology as going from the “*stone age to the space age*”.<sup>121</sup> Essentially this refers to the increased volume of genetic data that is now generated with newer genetic technologies. It has grown over the last ten years from a few hundred genotypes a week to a few hundred million genotypes in one day. This has created somewhat of a bottleneck in the data analysis and has required laboratories to “*retool their laboratories*” with computer technologies that can process, analyze and store these large volumes of data. As a consequence, the professional scientific domains of autism genetics research have expanded to include computer scientists, epidemiologists, and statisticians, in addition to geneticists, neurologists, psychiatrists, psychologists and clinical scientists. This is by no means an exhaustive list but one that is continually expanding as new forms of knowledge and technology take hold and create new “moments of genetic interpretation” of ASD.

### **Genetic Reclassification of Autism Phenotypes**

Amid the many uncertainties that surround genetics research on autism, namely the lack of understanding genetic or environmental causes of 80-90% of ASD cases, parallel approaches have been taken by scientists to advance the understanding of genetic factors underlying ASDs. As described in a recent review on autism, "the emerging notion of ASD as ‘the autisms’, a collection of dozen or perhaps hundreds of etiologic forms that converge on common behavioral and cognitive phenotypes, is largely a result

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<sup>121</sup> Scientist Interview #20 – Molecular geneticist

of advances in autism genetics" (Geschwind, 2009, p. 372). Thus, another major approach taken in current genetics research on autism has been the reclassification of ASD according to specific phenotypes in order to reduce the heterogeneity. This has resulted in new sub-groupings of ASD based on phenotypes such as language development, or other neurobehavioral features such as behavioral inflexibility (D.H. Geschwind, 2009).

Some researchers are also sub-grouping ASD based on co-morbid conditions that are not part of the diagnosis such as gastro-intestinal issues, immune dysfunction, seizures, and sleeping disorders to try and find “threads of homogeneity”. According to scientists, these approaches underscore how diagnostic categories used in clinical practice (e.g., autistic disorder, Asperger syndrome, PDDNOS) might not properly represent genetic risk. And in the case of co-morbid manifestations, the limits of a diagnosis based on behavioral characteristics alone. As one scientist describes it, *“the phenotypes that we have--autism, Asperger syndrome, PDDNOS--are not genetically informative. They're too far downstream from the initial genetic mechanism”*.<sup>122</sup> Thus, autism genetic researchers are devising ways in which to manipulate the phenotypic data to gain statistically significant results. In the process, however, they are creating new classifications of autism based strictly on genetic interpretations of disease. This is what anthropologist Rayna Rapp describes as phenotypes dissolving into gentyypes, where the “world of genomics has produced a set of highly materialist procedures that elegantly reduce transmission of continuity and change to the computational alphabet of life” (Rapp, 2003, p. 141). However, this “computational alphabet of life” is currently only

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<sup>122</sup> Scientist Interview #14 – Child psychologist

beneficial for the scientific enterprise and not for families or individuals with ASD as will be evident in Chapter 4 and Chapter 6.

Genetic reclassification of ASD traits has also expanded to include unaffected family members, such as parents and siblings of a child diagnosed with ASD. This is based on the model that key aspects of ASDs might be at one end of the continuum of “normal” behavior and cognition (Abrahams & Geschwind, 2008). In this approach, called qualitative trait loci (QTL) mapping, autism endophenotypes (i.e., measurable traits that are both heritable and related to a specific aspect of a condition under investigation) are studied as opposed to whether an individual has an ASD diagnosis. These refined quantitative endpoints such as speech and language delay, age at first word and aspects of social behavior have all identified several genetic regions linked to ASDs (Abrahams & Geschwind, 2008). Thus, what is transpiring in autism genetics research today, or at least what is presented by autism scientists, is the reclassification of ASD at the genetic level based on identification and development of sub-phenotypes represented in ASD. This logic runs parallel to current approaches to pharmacogenomics, where pharmaceutical companies “operationalize human genetic variation by matching patients to the most appropriate pharmaceutical intervention” (Lakhoff, 2008, p. 753). In this particular application, gene-based diagnostic tests are used as coding mechanisms to distinguish heterogeneous groups of subjects (Lakhoff, 2008).

Charles Rosenberg highlights the importance of technologies in our ability to create and modify disease entities (Rosenburg, 2002). Rosenberg argues, “these conditions become emotional and clinical realities, occupying a position somewhere between warning signal and pathology” (Rosenburg, 2002, p. 254). As exemplified in

the case of autism genetics, the technological advancements of genetics have enabled new categories of autism to emerge and have expanded the boundaries of ASD to include co-morbid symptoms outside of the diagnostic category. This scientific approach also implicates parents of children with ASD (as well as their unaffected siblings) who may have specific traits that are at one end of the continuum of “normal” behavior and cognition. The identification of these traits by parents is evident, which I demonstrate in chapter six. In this case, non-diagnosed individuals become associated with ASD based on non-normative phenotypes that are believed to be genetically associated to ASD. Thus, the extension of intermediary phenotypes that occurs in the process of scientific inquiry of autism genetics (e.g., endophenotypes or co-morbid conditions) expands the biomedicalization of ASD, which can have a profound influence on the stigmatization of and identification with ASD (Clarke, Shim, Shostak, & Nelson, 2009).

However, this finding should not be interpreted as a form of genetic determinism, especially given the depth of complexity in the etiology of ASD. For example, Rabeharisoa and Bourret show how genetics reinforces the complexity of pathological categories (Rabeharisoa & Bourret, 2009). These authors demonstrate how genetic mutations “expand and recompose” pathological situations rather than reifying and simplifying disease entities (Rabeharisoa and Bourret, 2009, p. 699). Although their level of analysis was framed within the medical clinic, this interpretation can also be applied to the current analysis of autism genetics. Here, scientists are collecting and comparing multiple and heterogeneous data to identify genetic mutations relevant to ASD, whose “status is ambiguous and whose effects are uncertain” (Rabeharisoa and Bourret, 2002, p. 691).

Although genetic reclassifications of ASD are based on the need to obtain “threads of homogeneity”, the scientific results are instead highlighting the etiologic complexity of ASD. Evelyn Keller also points out how advances in molecular biology have given us a new appreciation of the enormous gap between genetic information and biological meaning (Keller, 2000). Thus, it is unknown how this reclassification will affect the future diagnostic entities of ASD. Research on the processes of diagnostic classification of cystic fibrosis shows that even when a single gene is identified and implicated in the etiology of disease, multiple interpretations ensue (Hedgecoe, 2003; Kerr, 2005). Furthermore, Fiona Miller and her colleagues (2006) demonstrate that new molecular genetic knowledge has influenced the classification of Rett syndrome in complex ways by allowing the phenotype of MECP2 mutations to be broadened beyond Rett syndrome, however mutations at this locus alone do not define Rett syndrome. Thus, this specific genetic locus has not been used to generate a new nosology and is only partially linked to the disease identity (Miller, et al., 2006). Given the current uncertainties and ambiguities surrounding the genetics of autism, the various interpretations and meanings of genetic knowledge will undoubtedly be shaped by a range of social and material actors, including professionals, patient and parent advocates, technologies, institutional organizations, and bodies, all of which must contend with etiologic knowledge in the production of disease categories and classification systems (Kerr, 2005; Miller, et al., 2005; Miller, et al., 2006).

## **Convergence of Common Genetic Pathways**

According to autism genetic scientists, one of the more intriguing findings has been the overlap in autism susceptibility candidate genes with other neurodevelopmental disorders such as intellectual disability, epilepsy, or psychiatric conditions (Bill & Geschwind, 2009). These findings have in large part been due to the ability of computational technologies to conduct pathway analysis, which scientists could never have conceived of doing ten years ago. For example, the same genomic region (CNTNAP2) that is associated with some of the language deficits observed in the ASDs are also observed in the developmental disorder specific language impairment (Vernes, et al., 2008) Furthermore, a recent study showed that disruptions in the front of this gene could lead to a more severe disorder, like full-blown autism or severe expressive language delay. While mutations toward the back end of CNTANP2 may not lead to the disorder or may cause Tourettes syndrome, a condition characterized by the presence of multiple physical and vocal tics (Poot, et al., 2009).

Some of the copy number variants found in ASD are also shared in schizophrenia, attention deficit hyperactivity disorder and bipolar disorder. For example, the CNV duplication on 16p11.2 has been associated with schizophrenia in two large cohorts (McCarthy, et al., 2009). McCarthy and colleagues also performed a meta-analysis and showed this duplication to be associated with bipolar disorder and autism. Thus, according to some scientists the conceptualization of what a disorder is must change towards thinking of “*families of disorders that share similar etiologic mechanisms.*”<sup>123</sup> This is particularly challenging especially in cases like schizophrenia where historically

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<sup>123</sup> Scientist Interview #14 – Child psychologist

people fought to have autism separated from childhood schizophrenia. To have to revert back to the idea that disorders with different manifestations and treatments, to some extent share a common etiology, can have major clinical, social and scientific implications. It raises questions of the extent these distinct disorders share similar phenotypes and could challenge the differentiation between autism and schizophrenia that has historically been firmly based on clinical symptomatology and diagnostic criteria (Volkmar & Cohen, 1991).

However, the convergences on common genetic pathways do not end with neurodevelopmental or psychiatric disorders. Molecular pathways of ASD are also becoming apparent in well-known cancer genetic pathways, which further complicates the notion of disease etiologies and genetic risk (Butler, et al., 2005; Herman, et al., 2007). In this case the same genes and gene pathways (e.g., PTEN) can be involved in two very different disorders or diseases. For example, specific alleles in the APOE gene (APOE $\epsilon$ 4) increases the risk for late onset Alzheimer's disease and the development of serious illness associated with lipid metabolism and heart disease (Lock, 2005). Lock argues that in order to fully conceptualize the genome, scientists "must pay attention to feedback loops and networks of interaction, privilege a synchrony of events over linear trajectories, and take seriously the idea that social and macro-environmental context can influence the regulation of genes" (Lock, 2005, p. S60). This has been described as a developmental-systems approach or theory, which incorporates more complex models of biology using development and ecology as their primary examples (Fujimura, 2005).

The ambiguous and uncertain status of genetic mutations and their interconnectedness to other disease pathways also transpires a level of uncertainty in the

clinic. For example, Rabeharisoa, and Bourret (2009) demonstrate how the status of mutations (i.e., CNVs) in clinical psychiatric genetics were “not presented as *the* molecular cause of the patient’s problems, but as an element in a complex chain of mechanisms and pathological events which make it possible to describe the association of singular disorders between the psychiatric, the physical, and the organic” (Rabeharisoa and Bourret, 2009, p. 704 emphasis in original). Rabeharisoa and Bourret argue that distinct chromosomal abnormalities (i.e., CNVs) challenged clinicians, and researchers alike, to explore nosographical domains, which served as “‘binding objects’, either between pathological categories formerly assumed to be unconnected to one another, or between pathological entities that lay on the border between psychiatry and organic medicine” (Rabeharisoa and Bourret, 2009, p. 707). Thus, as Rose proposes, “in the light of genomic knowledge, what is required is not a binary judgment of normality and pathology, but a constant modulation of the relationship between biology and forms of life” (Rose, 2010, p. 74).

### **A Glimpse of Imagined Futures for ASD**

Given the complexity of autism genetics and the unknown causes of 80-90% of ASD cases, the future of ASD remains within the imaginary of scientist working in this field. Fujimura (2003) argues that scientist themselves are imagining the future through their research, “participating not merely in the practice of science but also in redesigning culture and society” (Fujimura, 2003, p. 191). She views genome scientists as writing *a* book of life that differs in form, content, and interpretation within different historical periods and locations. Following from these ideas, I argue that current scientific

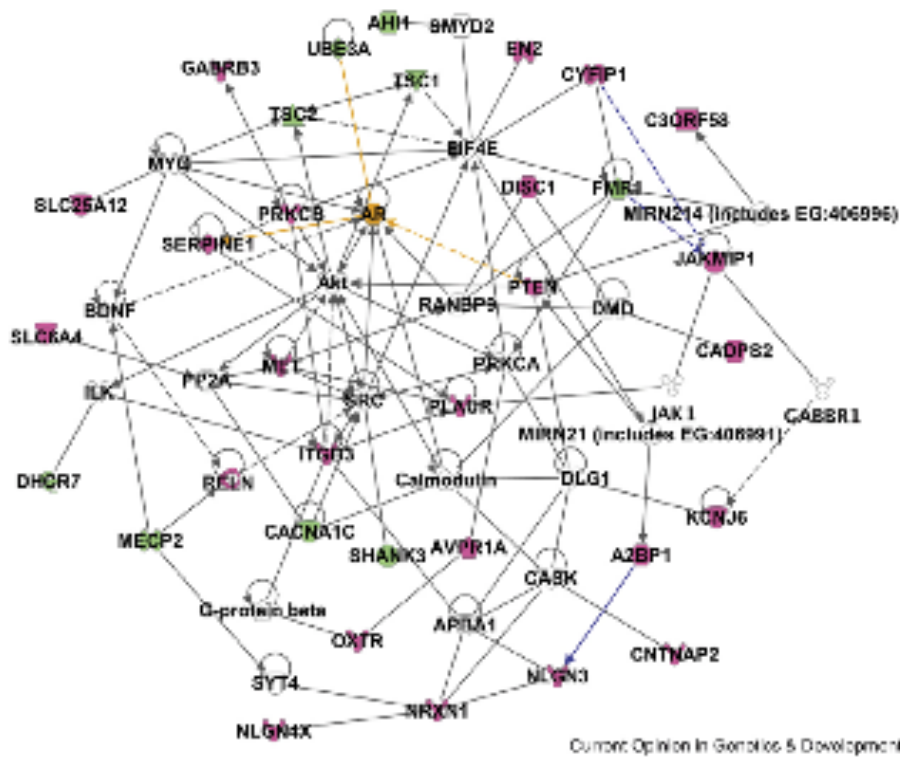


interpretations of autism genetics are very much driven by the technological advances in microarray technologies and the ability to scan the genome at higher resolutions. The ability to identify copy number variants has generated a new class of genetic mutations and chromosomal disorders associated with autism. Furthermore, genetic reclassification of ASD phenotypes and the convergence of disorders at the molecular level call into question the ASD diagnosis and current classification boundaries. These are of course, classic occurrences in the history of disease (Bowker & Star, 1999).

This phenomenon is also submerged within the larger national agenda to identify common variants responsible for complex diseases. This, in turn is politically driven by the increased funding and awareness of parent advocacy groups and public institutions (Silverman, 2004). Historically, the *de novo* CNV hypothesis has emerged within the last few years and is located primarily within and through academic-based genetic laboratories and research clinics throughout the United States, the U.K. and Canada.

Although the future of autism genetics is uncertain, predictions are being made based on the large volumes of genomic data that have been generated over the last five to ten years and new bioinformatics systems software for mapping regulatory networks. Sophisticated computer algorithms have begun to imagine what these complex pathways may look like if one were to “genetically dissect” the interconnected genetic mechanisms involved in autism. See Figure 3.1. This image depicts how ASD is beginning to emerge at the genomic level, as a series of interconnected molecular pathways. The authors describe this image as a “social network for autism susceptibility candidate genes” (Bill & Geschwind, 2009, p. 273). Why this image is referred as a social network by the authors is questionable but it implies interaction and connectivity among genetic

pathways that are potentially involved in ASD. Thus, the gene is becoming a verb, where the gene shifts from objects to interactive processes (Rapp, 2003). However, the interactions must be viewed as temporal and “spatially specific events”, meaning, that although the interactions are possible, they may not occur in the right tissue or at the right time to affect the pathology of ASD (Bill & Geschwind, 2009).



**Figure 3.1.** A social network for autism susceptibility candidate genes (Bill & Geschwind, 2009)

I show this image to illustrate future directions of autism genetics research and other complex diseases that are not caused by mutations in a single gene. It represents the genetic heterogeneity I discussed earlier and the results of the magnitude of genetic information that are now being generated through whole genome microarray and

sequencing technologies. Thus, the ability to “reinvent” the future of autism has relied heavily, on one hand, current technological advances. On the other hand, imagined futures are fueled by the current social and political awareness of autism that has arisen largely over the past 15 years due to parent advocacy groups pushing for organized research funding, clinical research networks, and new avenues for autism research (Silverman & Brosco, 2007). This level of human interaction is clearly absent from this image.

This image also envisions future possibilities around which scientific practices and communities are organized (Fujimura, 2003). For example, the future of autism genetics research heavily relies on the ability to identify specific subtypes of ASD based on genetic information to help inform clinical practice, treatments, and trajectory. Scientists imagine that the future will allow us to predict or diagnose whether a child will have specific ASD phenotype characteristics based “functional variants” in a genetic pathway such as this. Communities are also being organized based on genetic information, a concept Paul Rabinow refers to as biosociality (Rabinow, 1992). For example, a story in the New York Times documents the meeting of two families, each with a child harboring a 16p11.2 deletion (Harmon, 2007). Although the families are not related, their children share the bond of 16p11.2 deletion and similar features such as the flat bridge of their noses, the thin lips, the fold near the corner of their eyes (Ballif, et al., 2007). They are also among the emerging numbers of children given a specific diagnosis based on the new microarray technologies that can detect these smaller genomic deletions. Thus, in essence new communities are being organized based on similar genetic and phenotypic characteristics.

For treatment, the genetic pathways depicted in this image may provide a roadmap for scientists to begin to think about how pharmaceuticals can modulate these pathways to alter some of the effects of autism earlier. Treatment may also be altered based on targeted behavioral therapies that are more likely to help someone with a specific genetic profile. Although these are some of the projections made by scientists for the future of autism genetics, most would agree that clinical applications based on current autism genetic knowledges are really far off. Furthermore, the complexities of research findings are only a prelude to how complex any genetic testing and interpretation of the results will be for ASD in a clinical setting. For example, the levels of uncertainty, ambiguity, flexibility and resistance of genetic information in a clinical context have been demonstrated in various diseases (Bharadwaj, 2002; Cox & Starzomski, 2004; Kerr, 2000, 2005; J. Latimer, et al., 2006; Miller, et al., 2005; Miller, et al., 2006; Rabeharisoa & Bourret, 2009; Turney & Turner, 2000; Weiner & Martin, 2008). These examples illustrate how genetic information is only one part of a very complex story in defining, diagnosing, screening, and treating disease.

Scientifically, this complex molecular pathway represents the research agenda at the national level to “unravel the genetic architecture of ASD” through “deep sequencing” and related phenotypes and the “identification and/or functional characterization of genetic variants that have a large effect on the ASD phenotypes” (DHHS, 2009a). The concept of “genetic architecture” has been a metaphor for genetics research well over a decade and consists of all of the genetic and environmental factors

that contribute to the trait, as well as their magnitude and their interactions.<sup>124</sup> The interaction among genes based on this diagram has clearly been established, but what remains silent or missing, are the numerous potential environmental components that also interact or change the directions of these pathways. This is significant given the beliefs of parents, clinicians, and scientists alike, that environmental factors may be primary to the cause of autism. It also ignores the prospective cohort studies currently underway to assess environmental exposures (Lappe, In Prep). Thus, the image segments genetic mechanisms and ignores environmental causality precisely when their interactions are being tested.

This model represents a systems biology approach, which attempts to “provide rules and principles to organize these [genetic] bits of information into systems that help to explain the function and dysfunction of organisms” (Fujimura, 2005, p. 219). Fujimura (2005) argues that these models are being used to manipulate systems to produce different natures and new biologies. Furthermore, they incorporate “theoretical assumptions and principles, including researchers’ assumptions of minds, bodies, and nature” (Fujimura, 2005, p. 216). Such a model also represents the narrow biological terms of the biomedical model, whereby “disease is reconfigured *only* as an alteration in biological structure or functioning” (Kleinman, 1988). Rayna Rapp describes such models as “open-ended and nearly infinite interactions at the level of nucleotides”, which she argues is “a theory of far greater material complexity [that] paradoxically utilizes extreme methodological elementalism” (Rapp, 2003, p. 140). At the very extreme

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<sup>124</sup> National Institutes of General Medical Sciences (NIGMS): The genetic architecture of complex traits workshop. Retrieved December 19, 2009, from [http://www.nigms.nih.gov/News/Reports/genetic\\_arch.htm#at](http://www.nigms.nih.gov/News/Reports/genetic_arch.htm#at).

reductionism of technologies of information, images such as these do not represent ASD genes per se, but rather computational sequences arranged and compared bioinformatically (Rapp, 2003).

This image also signifies the construction and expansion of a range of bordering conditions and disease entities (Hedgecoe, 2001) such as Fragile X, Angelman syndrome or tuberous sclerosis, which contribute to the genetic risk for autism. Furthermore, it represents the expansion and blurring of diagnosis categories due to converging genetic pathways that are emerging out of new genomic technologies. Although no single gene has been implicated to be the cause of autism, a new genetic model has transpired, namely copy number variations (CNVs). CNVs add to the existing multiple entities of mutations that can be involved in the genetics of autism, which can range in the size of deletion or duplication, and whether or not it is rare, common, spontaneous, or inherited forms of autism genetic risk. Drawing from the work of sociologist Ann Kerr (2000) the flexibility (or ambivalence) of what constitutes an autism gene, a symptom or subtype of ASD, or a significant results appears to be a necessary condition of knowledge making (Kerr, 2000). This is reflective in the many uncertainties surrounding the new knowledges of CNVs and their place in the etiology of complex diseases. The hypothesis free approach to current genetics research also requires that scientist step into scientific inquiries with limited certainty of the outcomes. This can be interpreted as a form of “radical uncertainty” in science laboratories (Roth, 2009) Wolff-Michael Roth shows how scientists interpret scientific results “after the fact”, based on the data they produce (Roth, 2009, p. 315). He highlights the uncertainties of objects, actions, and technological

means that surround scientific work, whose dialectical relationship is necessary to stabilize one another (Roth, 2009).

Notwithstanding the many uncertainties of the molecular pathways involved in autism genetics and the limited ability to translate this into practical clinical applications in the near future, genomic technologies continue to move forward. Microarray technologies have led to the ‘discovery’ of several CNVs that are now being recommended as part of clinical genetic testing despite their unknown function and lack of availability of targeted treatment (Lintas & Persico, 2009). In 2008, the American College of Medical Genetics stated that due to the advances in microarray technologies, array-based genomic hybridization will likely be the first tier of testing in the future for clinical genetic diagnostic evaluations of autism spectrum disorders (Schaefer, Mendelsohn, & Professional-Practice-and-Guidelines-Committee, 2008). Furthermore, in 2010, the Division of Genetics and Department of Laboratory Medicine, Children's Hospital Boston and Harvard Medical School published a consensus statement recommending chromosomal microarray testing as a first-tier clinical diagnostic for individuals with autism (D. T. Miller, et al., 2010).<sup>125</sup> In other words, future clinical testing for autism will recommend starting with microarray genetic testing, a protocol already in place for several clinical genetic testing laboratories in the United States.<sup>126</sup> A clinical genetic laboratory director refers this to the “genotype first” model of diagnosis

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<sup>125</sup> The same sentiment is expressed in the new consensus report from the International Standard Cytogenomic Array Consortium (ISCA), a group of researchers from clinical genetics laboratories. After reviewing 33 studies, the group found that chromosomal microarray analysis finds a genetic cause of autism in up to 20 percent of all individuals tested, significantly higher than the 5 percent yield of karyotyping (ISCA, 2010).

<sup>126</sup> Emory Genetics Laboratory: Autism panel test details. Retrieved December 10, 2009, from [http://genetics.emory.edu/egl/test.php?test\\_id=93](http://genetics.emory.edu/egl/test.php?test_id=93).

for children with unexplained developmental abnormalities (Ledbetter, 2008). Thus, if the tool is there give it a job to make it the “right” tool (Clarke & Fujimura, 1992).

Clinical management is already beginning to change based on microarray analysis in the clinical genetics laboratory. As one clinical geneticist described to me:

There was one case that we had where [the CNV] took out P53 which is a known tumor suppressor gene. So now you have a child with developmental disability but also needs to be monitored for cancer.... you wouldn't have been able to do that without a targeted analysis and knowing that that gene is in that region, but now you have that information immediately, so for clinical purposes it's been really, I think, eye opening to see how many times we've changed clinical management.<sup>127</sup>

This clinical geneticist goes on to say how people need to continuously check back on their genotype status since the technology is always changing and genetic information is thus constantly being updated. Thus, the ability to detect more and more genomic variation that is presumed to be associated with disease, the more surveillance and clinical “intervention” people are likely to be subjected to. In this case, people with developmental disabilities need to be monitored for cancer, throughout their life, which in many cases will also involve the disciplined monitoring by parents or primary care givers.

The increased surveillance due to new genetic knowledges and monitoring of new genetic variants in populations of people with ASD is reminiscent of Foucault’s theory of biopower and the historical disciplining of two poles of development around which the organization of power over life is deployed, namely, the disciplining of the individual/organism body and regulation of populations (Foucault, 1984). Current technological applications of human genetics involve conceptualizing disease at the

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<sup>127</sup> Scientist Interview #4 – Human cytogeneticist



molecular level as a means of producing and representing new forms of human disease and categorization. As science moves closer to the ability to identify all forms of genomic variation, which will be the case when whole genome sequencing becomes economically available, the “molecularization” of disease will become magnified to the point of single base pairs existing throughout the human genome that can be “identified, isolated, manipulated, mobilized, recombined, in new tactics of intervention” (Rose, 2007, p. 6). Nikolas Rose sees this as engendering many new forms of somatic expertise by recognizing not only the limits but also the possibilities of our corporeality, where the somatic individual is subject to vital politics and new possibilities for action (Rose, 2001, 2007).

Charles Rosenburg also reminds us that technological diagnostic tools, such as genetic testing, “operationalize and embody disease entities”, which “become more plausible, more sharply defined and more frequently the framework and rationale for predetermined therapeutic interventions” (Rosenburg, 2002, p. 249). Drawing from his work and many others, Annemarie Jutel (2009) describes how “the disease potential is a salient means of social control and is amplified by the ever-expanding technical access to new screening tools” (pg. 291). Jutel also highlights how medicine is temporally situated and how diagnosis is determined based on the technology and values available at a specific point in time (Jutel, 2009). Thus, I argue that the emerging technological advances of microarray analysis in clinical genetics to diagnose autism and the constant “moments of interpretation” of genetic information, has lead to a level of social control, especially of parents in the case of ASD. Parents are advised to constantly check back on their child’s genetic mutation status. The interconnected pathways that converge on

common biological pathways, also opens up possibilities of the risk of future disease and surveillance mechanisms.

### **Constructing Autism as a Genetic Disorder**

In conclusion, I want to further highlight how scientists are privileging genetic explanations for autism by drawing on Adam Hedgecoe's concept of "enlightened geneticization" (Hedgecoe, 2001). He describes this as the discourse surrounding the genetics of a disease that is "constructed to prioritize genetic explanations, and subtly undermine non-genetic factors, while at the same time accepting that they have a role in its etiology" (Hedgecoe, 2001, p. 875). The scientists in this study undoubtedly acknowledge that there are environmental factors involved in the etiology of autism. However, they contend that the primary cause they would find at the end of the day would largely be due to genetics. There was agreement among these scientists that the concept of environment was so large it could essentially be anything other than genetic. A central argument against research on environmental causes of autism was that measurements were hard to do and very unreliable, creating what they believed to be even greater research challenges than what they faced with genetics. A recent review regarded that "[a] more thorough understanding of the genetic factors, which compose a significantly larger proportion of ASD risk than environmental factors, will facilitate identification of environmental contributions by suggesting mechanisms and providing more etiological subtypes in which to examine gene-environment interaction." (Geschwind, 2009, p. 370) Thus, genetics research according to this view is necessary in order to enable the identification of environmental factors contributing to autism.

Scientists also favored a genetic etiology for autism based on the fact that ASD is typically early onset as opposed to late onset allowing for less time for environmental exposures to cause the disorder. This of course ignores the consequences of prenatal exposure.

The construction of autism as a genetic disease is also present through the consistent acknowledgement that autism is highly heritable based on twin studies as well as their relationship to other chromosomal and rare genetic syndromes. The positive emphasis and progress made on the 10 - 20% of cases of ASD that are attributable to genetic causes also reinforce a genetic etiology of autism, despite the large portion—80-90%-- of ASD that have unknown causes. The ability to identify only one percent of ASD cases through clinical microarray testing was considered a success and justification for continuing research on CNVs and clinical application of this technology. Downstream, but soon, the health economics of such screening will be widely debated. In fact, at least five private companies are already offering high-resolution microarray analysis, such as GeneDx's "a la carte" genetic testing for individuals with autism<sup>128</sup> or CombiMatrix Diagnostics' "AT Scan" for ASD.<sup>129</sup> These microarray analysis genetic test, as well as others (IntegraGen,<sup>130</sup> Population Diagnostics,<sup>131</sup> Signature Genomics<sup>132</sup>) require a physician referral and can range in cost from \$1,200 - \$2,900 depending on the

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<sup>128</sup> GeneDx: Where rare is common: Autism/autism spectrum disorders. Retrieved April 15, 2010, from <http://www.genedx.com/site/autismdx>.

<sup>129</sup> CombiMatrix-Diagnostics: Autism test. Retrieved May 29, 2010, from <http://www.cmdiagnostics.com/testmenu2.htm#ATS>.

<sup>130</sup> IntegraGen: Autism IntegraGen's product. Retrieved May 16, 2010, from <http://www.integragen.fr/33-integragens-product.htm>

<sup>131</sup> Population Diagnostics Inc.: Technology overview. Retrieved May 29, 2010, from <http://www.populationdiagnostics.com/technology.htm>.

<sup>132</sup> Signature Genomics: Autism loci list. Retrieved May 29, 2010, from <http://www.signaturegenomics.com/documents/SGL0016.pdf>.

test. Furthermore, Holistic Health International is selling a comprehensive methylation panel with methylation pathway analysis to “optimize supplementation” that will address “genes, environmental toxins, and infections [that] all contribute to autism”.<sup>133</sup> Thus, the commercialization of genetic tests for ASD is evident. As Kaushik Rajan points out in his ethnography of genomic research and drug developmental marketplaces, “one can understand emergent biotechnologies such as genomics only by simultaneously analyzing the market frameworks within which they emerge” (Rajan, 2006, p. 33).

The limitations of genetics research are also driving factors for future genetics research. First, the shift towards the *de novo* CNV hypothesis is partly a result of the failure to find a major gene for autism and partly a result of the advanced technologies’ capacities to identify smaller chromosomal duplications and deletions. Second, the challenges of genetics research due to the heterogeneity of ASD symptoms have moved the genetics research agenda towards new genetic reclassifications based on ASD subtype. Third, the lack of reproducibility of most genetic studies to date has shifted priorities to increase sample size. This has involved a new era of genetic consortiums among researchers around the world and the establishment of publicly available autism specific genotype and phenotype databases (discussed in Chapter two and Chapter four). One particular research group is also recruiting autism families over the Internet to try and establish even larger genetic networks of families who can be used for genetics research.<sup>134</sup>

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<sup>133</sup> Holistic Health International: Comprehensive methylation panel with methylation pathway analysis Retrieved May 25, 2010, from <http://www.holisticheal.com/health-tests/nutrigenomic-testing/comprehensive-methylation-panel-with-methylation-pathway-analysis.html>

<sup>134</sup> Center for Autism Research & Treatment: The Interactive Autism Network. Retrieved December 10, 2009, from <http://www.semel.ucla.edu/autism/research/project/interactive-autism->

These efforts are explicitly justified through the belief that a gene for autism does exist and blatantly ignores both the latest environmental research initiatives (Lappe, In Prep) and the counter movements that believe some of autism is environmentally caused. This assertion is most vociferously expressed by parents who believe childhood vaccinations are at the root of autism (Kaufman, 2010; Senier, 2008; Silverman, 2004). The genetic scientists I interviewed regarded research in this area as an “*unscientific approach*” with “*no scientific basis*”. This claim is disputed by most scientists and leading scientific organizations, including the National Academy of Science’s Institute of Medicine that found no association between autism and childhood vaccines based on a series of large epidemiological studies (IOM, 2004). This conflict of autism etiologies has resulted in tensions between advocates pushing for more research linking childhood vaccinations and autism and scientists who believe continued research efforts in this area are not in the best interest of people with ASD.

As the refinement of the “molecular gaze” seeps down to individual nucleic acid pairs, the interpretations of this genetic knowledge will continue to take on new meanings and representation both within and outside of the scientific regime (Rose, 2007, p. 108). Thus, at the center of this analysis is the technology itself and the movement towards what I would describe as the “molecular-sopic” understanding of illness and disease. Microarray analysis can scan over 500,000 markers in the human genome and has essentially replaced the human eye previously used in cytogenetics, a branch of genetics that traditionally used a microscope to identify gross chromosomal abnormalities. The ability of genetic technologies to refine the DNA analysis to smaller and smaller

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network-ian.

segments of each individual chromosome has constructed new ways of interpreting and understanding illness and disease within the scientific domain. The proliferation of the molecular vision of life is not a new idea (Chadarevian & Harmke, 1998; L.E. Kay, 1993; Novas & Rose, 2000; Rose, 2007) and lives not only within laboratory and clinical science but also among a matrix of institutions, private organizations, and scientific domains (Bharadwaj, 2002; Hedgecoe & Martin, 2003; J. Latimer, et al., 2006; Rabeharisoa & Bourret, 2009; Rabinow, 1992; Shostak, 2005; Vailly, 2006, 2008). The knowledge production of autism genetics requires many different sites and stakeholders in addition to the scientists who create knowledge through sophisticated genetic technologies. These include, for example, families who provide biological and medical information for research, parent advocacy groups who demand and support genetics research, governmental institutions and private organizations that fund autism research, and academic and clinical institutions that conduct the science.

Although the scientists interviewed for this study framed autism as a genetic disorder, the interpretations of autism genetics exist among many uncertainties, ambiguities, and unknowns. Furthermore, as I have tried to articulate in this chapter and throughout this dissertation, the production of genetic knowledge dwells within a larger arena of many social worlds colliding at social, scientific and political levels. The next chapter will consider the construction of autism genetics at the family level based on interviews with parents of children diagnosed with autism who have also participated in a genetics research study. The fusion of familial and scientific social worlds is apparent given the necessity of blood, family medical histories, and behavioral and cognitive assessments needed to conduct autism genetics research. However, interpretations of

autism genetics are quite different at the family level because it takes into consideration the social and moral context of living everyday with ASD. As I will discuss in the chapter that follows, families' motivations to participate in genetics research and their hopes in the future of autism genetics intersected in complicated ways with their need for an accurate diagnosis, appropriate services and treatment, and the limited resources available to parents throughout the process.

## CHAPTER 4: MOTIVATIONS, HOPES, AND REALITIES OF PARTICIPAING IN GENETICS RESEARCH

### Welcome to Holland

I am often asked to describe the experience of raising a child with a disability - to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this.....When you're going to have a baby, it's like planning a fabulous vacation trip - to Italy. You buy a bunch of guide books and make your wonderful plans. The Coliseum. The Michelangelo David. The gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting. After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands. The stewardess comes in and says, "Welcome to Holland." "Holland?!?" you say. "What do you mean Holland?? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy." But there's been a change in the flight plan. They've landed in Holland and there you must stay. The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place. So you must go out and buy new guide books. And you must learn a whole new language. And you will meet a whole new group of people you would never have met. It's just a different place. It's slower-paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around... and you begin to notice that Holland has windmills...and Holland has tulips. Holland even has Rembrandts. But everyone you know is busy coming and going from Italy... and they're all bragging about what a wonderful time they had there. And for the rest of your life, you will say "Yes, that's where I was supposed to go. That's what I had planned." And the pain of that will never, ever, ever, ever go away... because the loss of that dream is a very very significant loss. But... if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things ... about Holland.

c1987 by Emily Perl Kingsley



This story captures the essence of parental hopes, losses, and life reorientations in the world of autism spectrum disorders. It was brought to my attention by several parents who I interviewed, who reread it often to remind themselves of the many possibilities surrounding autism spectrum disorders (ASD). For the particular parents I interviewed, this poem also represents how parents of children who are on the “higher end of the spectrum” are pushing for the acceptance of their child’s differences, a way to proceed in life that allows their child to be happy each and every day. These parents described their children as “*unique*”, “*one of a kind*”, “*creative*”, and “*caring*”. Their children also displayed atypical characteristics that generally do not fall into the negative stereotypes of autism, such as having a sense of humor and being very smart. This story also signifies the major adjustments parents must make who by have a child with autism, and the overwhelming confusion, frustration, and anxiety they face when their child is first diagnosed. Often, parents have to gather their own resources, determine what is best for their child, and act as a coordinator between schools, pediatricians, therapists, and many others. Finally, this story demonstrates that life with autism is a process for parents, one that leads them in many directions within their own family, their community, the educational and medical systems, and beyond.

Within this context, parents must make decisions about how best to help their child and become involved. As was evident in chapter two, such involvements can take an extremely powerful form through the work of large parent advocacy groups such as Cure Autism Now and the National Alliance for Autism Research. However, the parents in this study are being advocates somewhat differently—through participation in a genetics research study, a process where they donate blood and agree to extensive

interviews and observations that pertain to their life with autism spectrum disorders. As a sociologist, I was interested in what motivated parents to participate in a genetic research study and what they were hoping the research would find: What concerns did parents have of participating and what was their overall experience? Although these were the central research questions at the outset of the study, what I heard were complicated stories about interactions between the need for an accurate diagnosis, appropriate services and treatment, and the limited resources available to parents throughout the process. These different interactions intersect with the motivations for participating in research and the hopes of what it will find, which calls into question the research ethics of enlisting parents in a genetics research study who are already in many ways “desperate for answers”

The purpose of this chapter is to portray and unravel these processes. I also discuss the many ways families come to participate in genetics research on a disorder that is on the rise and continually changing--- contracting and expanding in definition. I first briefly outline the genetic study these parents participated in, the Simons Simplex Collection, followed by an analysis of the social processes that motivated them to participate and their hopes of genetics research. This chapter then turns to a discussion of the concerns parents had about participating in a genetics research study, their understanding of the study, their knowledge in genetics in particular, and their opinions regarding the future of autism research. In conclusion, I discuss some of the ethical implications that arose through these interviews by highlighting and addressing the specific social dimensions that are entangled within the fabric of participating in genetics research.

## Simons Simplex Collection

This initiative is different. It is focused on families with just one child with autism, called simplex families, which will provide insight into the most common and unexplained form of autism. This comes at an exciting time in history, in which breakthroughs in gene mapping, advancement of high-tech tools and the latest brain research present a unique opportunity for progress.<sup>135</sup>

The parents interviewed (N=15) for this chapter all participated in the Simons Simplex Collection (SSC), a study to establish a permanent repository of genetic samples from 3000 families, each of which has one child affected with an Autism Spectrum Disorder (ASD).<sup>136</sup> The collection consists of blood samples drawn from the biological parents, one child with an ASD and one unaffected sibling, as well as detailed phenotypic information about the affected child using standardized diagnostic instruments.<sup>136</sup> There are thirteen clinical collection sites throughout the United States and Canada<sup>137</sup> and the samples are stored in a central repository, which scientists may request for use in their own experiments. The SSC is still in the process of collecting data (estimated completion in Spring 2011) and is being funded exclusively by the Simon's Foundation. This is a New York-based philanthropic organization that has committed millions of dollars to autism research and intends to spend \$100 million dollars more in what is rapidly becoming the largest private investment in the field.

The Simons Foundation was established by Jim and Marilyn Simons, parents of a young adult daughter who displays symptoms of Asperger syndrome, a milder disorder

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<sup>135</sup> Simons Foundation Autism Research Initiative (SFARI): Simons Simplex Collection. Retrieved March 17, 2010, from <https://sfari.org/simons-simplex-collection>.

<sup>136</sup> For a list of instruments used for the SSC study see <https://sfari.org/ssc-instruments>.

<sup>137</sup> For a list of participating SSC clinical sites see <https://sfari.org/simons-simplex-collection>. These sites are under the guidance of the University of Michigan Autism and Communications Disorder Center.

that bears similarities to autism. Jim Simons is a trained mathematician and is president and founder of Renaissance Technologies Corporation, one of the world's most successful hedge funds that utilize complex mathematical models to analyze and execute trades. His philanthropy initially focused on donation of tens of millions of dollars to math and science endeavors worldwide, including Stony Brook University and the Mathematical Sciences Research Institute (MSRI).

The Simons Foundation started their philanthropy for autism in June 2003. They hosted an autism workshop at New York's Plaza Hotel to bring together a group of renowned academic figures that represented top minds in the fields of epidemiology, neuroscience, psychiatry, and autism research. It was a daylong event dedicated to discussion of how to break new ground in research into the causes of autism, the accurate genomic mapping of autism, and the biochemical mechanisms that occur in people with autism.<sup>138</sup> The take away message according to Mr. Simons was that scientists had only one solid lead, which was based on studies on identical twins – that genes play a key role in autism (Regalado, 2005). Then, in 2003, the Simons Foundation Autism Research Initiative (SFARI) was started to combine the foundations interests in scientific research and learning disabilities. The mission of the SFARI was to improve diagnosis, treatment, and prevention of autism and related developmental disorders, and to provide tools that scientists can use to enhance their understanding of autism.

Based on the recommendation of leading autism researchers, the SFARI initiated the Simons Simplex Collection (SSC), which has been described as the “Cadillac resource” for conducting autism genetics research. Geneticists wanted a resource

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<sup>138</sup> Simons Foundation Autism Research Initiative (SFARI): SFARI history. Retrieved March 17, 2010, from <https://sfari.org/sfari-history>.

different from the Autism Genetic Resource Exchange (AGRE), one that consisted of families with only one child diagnosed with an ASD (i.e., simplex families) and one that could easily re-contact families through the recruiting clinic. The idea to establish a genetic database of only simplex families was based on preliminary results that *de novo* copy number variants were present at a higher rate in kids with autism than in unaffected children. But this was only significant in simplex families, not multiplex families (families with two or more children on the spectrum). Thus, a collection of simplex families was designed to discover new mutations that occurred spontaneously in the parental germ line (Simons-Foundation, 2008). Currently, more than 35 research projects are using SSC genetic data and phenotypic information. Two independent groups, one led by Dr. Michael Wigler at Cold Spring Harbor Laboratory and another led by Dr. Matthew State at Yale University, are conducting genome wide scans of the collection.<sup>139</sup>

The majority of families who participate in the SSC are recruited through parent support groups, parent-to-parent referrals, and conferences, as well as through referrals from physicians, teachers, clinicians, and school psychologists. Participation generally consists of two visits. The first requires an extensive parent interview and evaluation of the child with ASD, and the second consists of the blood draw from each family member. Once the families are pre-screened and qualify for the study, they schedule a time to come to the clinic to participate. The parents must sign three documents before they participate: an informed consent document to participate in the study; the Health

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<sup>139</sup> Simons Foundation Autism Research Initiative (SFARI): Database allows easy navigation of simplex collection Retrieved January 18, 2010, from [https://sfari.org/sfari-announcements/-/asset\\_publisher/p14J/content/database-allows-easy-navigation-of-simplex-collection?redirect=%2Fsfari-announcements](https://sfari.org/sfari-announcements/-/asset_publisher/p14J/content/database-allows-easy-navigation-of-simplex-collection?redirect=%2Fsfari-announcements).

Insurance Portability and Accountability Act (HIPPA) authorization form; and a blood and cell line banking consent form.

### **Motivations for Participating**

The motivation to participate in a genetics research study for the parents interviewed in this study centered around three themes: to get a free diagnostic evaluation, the desire to help their child, and the willingness to help in this area in any way possible. Yet, these processes are not as straightforward as they may seem, since each is tied to different situations and experiences prior to participating in the study. Eleven participants were motivated to sign up for the study based on the free diagnostic evaluation; however, not all placed it among the first reason they participated. And, the altruistic commitments of families who participated for the sake of helping in any way (n=10) were also tied to their obligations as a parent to help their own child, as well as future generations of families with ASD.

### ***A Diagnostic Evaluation***

A huge incentive for families to participate is the free psychological evaluation that is provided as part of the study. This diagnostic evaluation is a significant incentive since parents often have to wait over a year to see a developmental pediatrician or neurologist who can accurately diagnose an ASD. Furthermore, the cost of a clinical diagnostic evaluation is well over \$2000, which many parents have to pay out of pocket since it is not covered by insurance. The study provides all participants with a written research report that includes information about the child's diagnosis, cognition and

adaptive behavior, and recommendations for intervention. The parents are encouraged to use this evaluation to help qualify for educational services. The families are not given any genetic testing results, nor will they ever receive any in the future. Rather, they are referred for additional genetic testing if warranted. This last condition was a point of confusion for several of the families I interviewed.

Five sets of parents who were interviewed in this study participated in the SSC to get a definitive diagnosis for their child. All these parents were new to the world of ASD and saw this study as an opportunity to get answers to questions they were desperately seeking. These mainly revolved around what exactly was going on with their child and what could they possibly do to help. Parents indicated that it was extremely stressful to be so worried about their child and not know whether something was truly wrong. One parent whose son was never formally diagnosed stated:

That's what we wanted first and foremost was somebody to say, okay, look, he's autistic. And then tell us what level he's capable of operating at...and you know, evaluate him and kind of help us figure out what to do to get him, you know, the services that he needed early.<sup>140</sup>

These parents wanted to know with certainty whether their child was on the autism spectrum and assumed that a proper diagnosis would allow them to seek the most appropriate care for their child. One mom who suspected her son had Asperger syndrome wanted to find out for certain if this was what her son had. She stated:

For two years there in school I was wanting to know why is he doing this? Why won't he listen when we're telling him you have to take turns? I mean, it was every year--every year it was the same ritual.<sup>141</sup>

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<sup>140</sup> Parent Interview #8

<sup>141</sup> Parent Interview #4

Thus, the motivation to participate based on the free diagnostic evaluation was evident for parents who were longing for answers. This was especially strong for those who had no definitive diagnosis (or a diagnosis that seemed inappropriate such as attention deficit hyperactivity disorder) and who did not have the support from their pediatricians, family members and/or the educational system. These feelings of frustration and of not being believed are reminiscent of research on medically unexplained symptoms (Dumit, 2006; Nettleton, 2006; Peters, Stanley, Rose, & Salmon, 1998; Ring, Dowrick, Humphris, Davis, & Salmon, 2005; Werner & Malterud, 2003; Zevestoski, et al., 2004). For example, Nettleton (2006) argues that the uncertainty of non-diagnosis and the questioning by others of the legitimacy of complaints create significant doubt, distress, and chaos. Thus, it is not surprising that parents in this study expected the diagnosis not only to provide answers and but also directions they could follow to help their children. This was a reoccurring theme throughout the accounts of parent's motivations to participate, their hopes for autism science, as well as the directions they want scientists to pursue in future.

A second group of parents (N=7) whose children already had a diagnosis were also motivated to participate in the SSC because of the free diagnostic evaluation. However, these parents were not seeking a specific diagnosis but an update on the progress their child had made since their first diagnostic evaluation. These parents were mainly interested in using the diagnostic evaluation to add to the existing diagnostic assessment(s) and to fine-tune the educational services they already had in place. For example, one parent whose son was fourteen wanted to have the diagnostic evaluation in place before he started high school, and saw this component of the study as a "perk".



Other parents whose children were younger wanted to see if the diagnostic evaluation would qualify their child for additional services that they were not yet receiving. One parent was using the evaluation to compare it with other clinical evaluations she had received in the past. She stated:

I feel like I'm kind of cross referencing and maybe making sure that I can do the best I can for him, but also that if there are other issues or if somebody got it wrong along the way, somebody's going to tell me.<sup>142</sup>

Thus, the free diagnostic evaluation motivated parents to participate. This motivation was tied to the previous diagnosis history of the parent's child and the desire to help their child obtain appropriate educational services.

### ***“Helping my Child”***

It is unquestionable that the parents who participated in the SSC did so in some way to help their children. However, some parents viewed the study as a way to alleviate the guilt and obligation they had to help their child in any way possible. For example, one parent felt that participating in the study was the easiest way to be involved and stated, *“It helps alleviate some of the guilt that you feel as a parent that you are never doing enough to help your kid, so at least in some small way I am trying to help.”*<sup>143</sup> Another mother also viewed it as an obligation to participate since she knows what it is like to be a parent of a child on the autism spectrum. She stated:

As a parent, you know, that's our job, really, is too--you know, we have a child who's affected by this. And we have people around us who are

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<sup>142</sup> Parent Interview #16

<sup>143</sup> Parent Interview #12

affected by this. And we want to do whatever we can to help ourselves and others who are affected.<sup>144</sup>

This parental sense of obligation was combined with the desire to seek ways to help their child such as, “*helping him help himself*”, “*getting him to some kind of normality in life*”, “*helping him be more functional socially*” and “*helping him organize his head*”. These ways of helping their child were in no way linked to a desire for a cure or any expectations of a cure. Rather, they constituted a broad range of requests to very specific challenges experienced by their children. Thus, for some parents, the motivation to participate was driven by their need to help their child in ways that addressed the realities of living day to day with a child on the autism spectrum.

### ***Altruism and the Benefit of Future Families***

Parents were also motivated to participate based on altruism and the willingness to help out in any way possible regardless of whether their own family would directly benefit from the study. It clearly states in the informed consent documents that there will be no direct benefits to families who participate in the SSC, other than the free diagnostic evaluation. This understanding was clearly evident to the participants I interviewed, indicating the altruistic nature of their participation through the donation of blood and personal family history. This altruism was directly related to the idea that the research would benefit families like theirs in the future. For example, one mom who had a ten-year-old son diagnosed with Asperger syndrome described it in the following way:

For us the reason why we participated in the genetics studies are for people in the future so that, if there was a way for us to have known at

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<sup>144</sup> Parent Interview #13

birth that this could happen, then we would have been prepared...There's so many families who don't have any idea what to expect, um, that if in any way this helps inform other parents early in the process then, we're all for it.<sup>145</sup>

A few parents emphasized that it would be “*years and years*” before anybody figured out something genetic, and it was unlikely that it would benefit their family directly. Thus, their motivation to participate was driven by the desire to be part of a bigger solution or, as one mom described it, “*a bigger village*” or “*one link in a continuing spectrum*” that is required to move the understanding of autism forward.

This sense of altruism, of “*willing to do what ever it takes*” to benefit other families with autism, was also tied to the emotional experiences parents brought to the table of learning their child had autism, living day to day with the challenges they faced, and doing all the work it takes to create the best quality of life they can offer their child.

As one mom described it:

It's all relative. If you've experienced this kind of situation with these kinds of difficulties, I think that you want to do whatever you can to make that situation better for someone else, or for their children or their grandchildren or whatever.<sup>146</sup>

Another group of parents also viewed their participation as imperative to advancing autism awareness and research. One mom, whose son was unable to verbally communicate, felt that not enough studies were being conducted on autism and people were unaware of studies like the SSC that were recruiting families to participate. Thus, she took it upon herself to participate and promote the SSC study in her local autism advocacy group. Another mom, who had a son diagnosed with Asperger syndrome,

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<sup>145</sup> Parent Interview #12

<sup>146</sup> Parent Interview #2

viewed her participation as “*taking a chance*” on research. She felt that the way things became “*the norm*” was through experimentation. She vividly described families who participate in research as “*brave souls*” because there was no guarantee of directly benefiting from participation.<sup>147</sup>

Clearly the motivations to participate were multi-dimensional for the families interviewed in this study. They ranged from personal benefit of obtaining a diagnostic evaluation to more altruistic notions of helping other families in the future regardless of immediate benefit for their own family. Thus, the social, cultural and moral contexts within which individuals participate in the donation to genetic databases imply a wider concept of engagement and many levels of participation (Busby, 2004; Haines & Whong-Barr, 2004; Tutton, 2007). For example, Haines and Wong-Barr (2004) describe the notion of participation and decision making around the aspect of genetic databases as a “highly varied social process, with multiple meanings” (p.57). Based on interviews with people who participated in a community genetic database, these authors analyzed several “styles of participation”: the “active participant” who is willing to help in any way; the “cost/benefit participant” who balances the cost to themselves to the greater collective good; the “passive participant” who can see no reason not to participate; and the “reluctant participant” who now regrets having participated in the study (Haines & Whong-Barr, 2004, p.70). Many of these styles of participation were apparent in the current case study except for the “reluctant participant”. No parent I interviewed regretted his or her participation in the study. The current study also highlights what I call the “desperate participant” who participates based on their desperate need for answers and

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<sup>147</sup> Parent Interview #6

their desire and commitment to help their children. As will become evident, these styles of participation also linked up to the parent's hopes for science to unravel the mysteries of ASD.

### **Hopes of Science**

The current state of autism research still lacks knowledge of what causes the disorder, how to diagnose it earlier, and the best treatments and therapies to use given the range of options available to parents. These problems are exacerbated by the heterogeneity of symptoms<sup>148</sup> that exists in the growing population of children with this disorder. Thus, it is not surprising that the hope parents place on research in general, and the SSC more specifically, was centered on finding what causes this disorder, how it can be better diagnosed, and how it can be “*fixed*”, “*cured*” and/or “*prevented*” in the future. Although the SSC specifically states in the informed consent document that “we are working to understand how genes might contribute to the common behavioral, social and communication symptoms of ASD”, the parents hoped the study would contribute to the understanding of ASD in a much broader sense. In fact, only three parents mentioned that they were hoping the study would find a “*genetic link*”. This is illustrated in the following discussions of the range of causes, treatments, and cures parents were hoping the SSC would reveal.

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<sup>148</sup> See Chapter three for a thorough discussion of the heterogeneity of ASD.

### *Hoping to Find the Cause*

At the onset of the interview, I asked parents what they believe caused ASD in their child. Their responses ranged from pregnancy complications, to environmental exposures such as vaccines or other toxins, to genetics, or the combination of environmental exposures and genetic susceptibilities. Only one parent felt confident that vaccinations were the cause of her son's problems, mainly because he had a severe adverse reaction after his 15-month shots. Six parents felt vaccines were not the cause, and the rest felt that it was a possibility.<sup>149</sup> Other environmental exposures that parents thought may have contributed to their child's ASD ranged from drugs given to the baby during pregnancy, at birth, or while nursing; a high fever at five months of age; a result of in-vitro fertilization; anesthesia given to a child during surgery; and lead paint. Other than the one parent who believed it was the vaccines that caused her son's ASD, all the other parents were uncertain of the causes and were only guessing based on what they had thought might have caused it after they learned their child had an ASD.

The hope that the SSC would determine the cause of ASD was the predominant theme among parents interviewed in this study (N=7). Interestingly, the hope to unravel a cause or causes had different implications for parents, and was not necessarily tied to the hope for a cure. One mom felt that just knowing the cause would help her and her family, even though there is no cure for ASD. One clear implication of finding the cause could be the alleviation of guilt that parents experience if they found out once and for all what

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<sup>149</sup> See Kaufman (2010) for a fascinating ethnography that traces parent anxiety about the connections between autism and vaccines. She explores the ways in which parents think about potential risks of vaccines and make decisions about immunizing their children.

caused their child's ASD. For example, the mom who believes her son got ASD from his childhood vaccinations described her hopes for finding a cause in the following way:

It would bring closure, sort of, to it because as a mother, I mean, I still, in the back of my mind, I'm still like what could I have done. What--what did I allow happen or should I have not gave him the shots. You know you can always go back, hindsight. You know, and it's just trying to reconcile it in your head.<sup>150</sup>

Most of the parents continuously tried to pinpoint one particular incident that may have caused their child's ASD. Thus, finding the cause would help bring closure to the constant worrisome questions of causation, responsibility, and what one parent referred to as being "*in the dark*" as to what happened.

Finding a cause was also viewed as part of the process of legitimization and acceptance of ASD. One mom felt that people were skeptical of accepting ASD as something real precisely because of its unknown etiology. She felt that finding a cause would legitimize ASD as a real disorder and not something that "*doctors are making up*". She went on to say that her family members tell her "doctors just want to diagnose your son with something" and they question whether she should be subjecting her son to different therapies.<sup>151</sup> Thus, there is a strong interplay between identifying the causes and legitimacy of ASD. Without knowing the causes, parents are at risk of being denied social recognition of the challenges experienced by their child. This pattern also reflects the research conducted by Miller and colleagues (2010) on the disclosure of genetics research finding to families with ASD. These authors contend that the dominant expectation of parents from genetics research was to understand 'why' their child had

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<sup>150</sup> Parent Interview #14

<sup>151</sup> Parent Interview #9

autism. In addition to providing important information about reproductive risks, especially for siblings and extended family members, for some families this information also reduced self-blame and brought peace of mind (F. A. Miller, et al., 2010).

Understanding the root cause can also help parents accept their child's diagnosis. For example, one mother, whose fourteen-year-old son just got a diagnosis of autism from the SSC research evaluation, was having a hard time accepting that he did not have a milder form of ASD like Asperger syndrome or PDD-NOS. She stated, *"it's like if you know what's causing something, if you know what causes cancer, abnormal cells and all this. So it's almost like if you know what causes it, you can accept it more."*<sup>152</sup> Thus, understanding the cause can bring legitimization and acceptance for parents and for those associated with families of ASD. As David Gray points out, the process of parenting a child diagnosed with ASD requires constant negotiation of the different ways of knowing disability (Gray, 1997). One of these is medical knowledge, which David Farrugia argues allows parents of children diagnosed with ASD to avoid "felt stigma" (Farrugia, 2009).

### ***The Diagnostic Odyssey***

The hope for better diagnostics to identify ASD earlier was tied to hopes for better treatments and therapies since these processes go hand in hand. Parents were hoping for a diagnostic test that could be used when children were very young. An earlier diagnosis would allow parents to start therapies sooner, which many professionals believe is key to helping many of these children. This hope is a reflection of the realities of current diagnostic timelines. For example, one mom felt that children were diagnosed *"too late"*

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<sup>152</sup> Parent Interview #5



making it “*even that much harder to get above and beyond the symptoms of autism.*”<sup>153</sup>

A recent report by the Interactive Autism Network (IAN) indicated that on average children were diagnosed with autism at 3.2 years old, PDD-NOS at 3.7 years old, and Asperger syndrome at 7.2 years old. (IAN, 2010). Thus, the less severe children, much like the children whose parents were interviewed in this study, are likely to receive a later diagnosis and also start therapy much later.<sup>154</sup> One father hoped that by having a diagnosis in place earlier, people would be able to qualify for services sooner through health insurance. This particular family was unable to get timely services for their son because it was too expensive and insurance would not cover it.<sup>155</sup>

The experience of the diagnosis process per se also caused much frustration and anxiety for the parents I interviewed. This has been described by professionals in the field of clinical autism genetics as the “diagnostic odyssey”. This concept encompasses the processes families go through to understand the problems their child is experiencing in order to get them the appropriate help they need. Based on my interviews, this process has many levels of diagnostic uncertainty such as getting an incorrect diagnosis and the repercussions of improper medical treatment. Parents also experienced uncertainties associated with arbitrary labels given to their children such as “*not quite autism*”, “*pervasive developmental disorder autistic like*”, or “*mild autism*”. These were labels parents could not identify with or attach any meaning to. Within the diagnostic odyssey, parents were also overwhelmed after the initial diagnosis, which many parents described as being given a diagnosis without any direction as to how to proceed in helping their

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<sup>153</sup> Parent Interview #7

<sup>154</sup> This disparity is even greater in children of different racial and ethnic backgrounds. See (Mandell, et al., 2009)

<sup>155</sup> Parent Interview #11

child. Most parents had to do their own research, felt there were limited resources they could draw upon, and were responsible for locating, assessing, and coordinating everyone involved in their child's care.

The parents also described to me how they utilized the Internet to help navigate the possibilities following an ASD diagnosis. These ranged from trying to translate the particular diagnosis their child received (such as PDD-NOS or Asperger syndrome) to deciphering the best treatment options they could feasibly try in order to help their child. This information did not typically come from the clinician providing the diagnosis and was usually acquired outside of the institutional domains of medicine.<sup>156</sup> Parents had to negotiate between the “science and fiction” disseminated in the media and used the Internet as a “bridge out” of the new uncertainties that accompanied a diagnosis.

Thus, the “diagnostic odyssey” was a journey of many uncertainties for parents,<sup>157</sup> which further supports their hope that autism genetics research will help untangle this complicated labyrinth they must navigate to help their children and ultimately provide better understanding of something that is very much part of the fabric of their lives. The diagnosis enables what Annemarie Jutel (2009) describes as “discovery of pathology, the

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<sup>156</sup> Nettleton (2004) describes this diffusion of information as “e-scaped” medicine, which are networks of contemporary “info-scapes” that can be “accessed, assessed and reappropriated” (p.676). She argues that the spaces, sites and locations of the production of medical knowledge are no longer exclusive to the medical academy and the formal medical text and are now more diffuse and are invariably mediated by means of digital technologies (i.e., biomedicalized) (Nettleton, 2004).

<sup>157</sup> The diagnostic uncertainties discussed here are also related to the range of symptoms associated with a diagnosis of an ASD under the current diagnostic criteria in the DSM-IV (revised), as well as symptoms that are not part of the criteria, which parents believe are part of “the autism”. Furthermore, there are many levels of severity, which change over the course of a child's life that further complicate the diagnostic odyssey. These issues combined with parental denial, the possible “invisibility” of the disorder to people outside the family, and the levels of stigma attached to different diagnostic labels are topics beyond the scope of this dissertation, but are areas of investigation that will be considered in the future.

treatment, or correction of the biological abnormality”, which in the case of ASD, enables parents to identify a path they can follow to help their children in the future (p. 288). However, resolving the ASD diagnostic odyssey through a “definitive” diagnosis was not the end for parents, but the beginning of another complex journey of investigating, contemplating, and negotiating the future of their children’s lives. Furthermore, as Charles Rosenberg (2002) argues, the disease category also provides a framework for “assimilating the incoherence and arbitrariness of human experience to the larger system of institutions, relationships, and meanings in which we all exist as social beings” (p. 257).

### ***Hoping for a Cure***

Hopes for the SSC to find a cure were less represented in this sample (N=3), and one parent referred to a cure as a way of “*fixing it*” in the future. For example, one mother, whose son was non-verbal and had a hard time communicating, did not hesitate to say, “*if there was a way to cure him we would use it. It would make him more like him, [the way] he's supposed to be.*”<sup>158</sup> Another parent described his hope for a cure as a way for his son “*to be back to normal*”.<sup>159</sup> These parents were holding on to visions of how they thought their children should be and deeply desired for a cure.

One parent did not use the word “cure”, but referred to her hopes of research as a way of “*fixing*” or preventing future children of having ASD. She envisioned this “*fix*” as:

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<sup>158</sup> Parent Interview #3

<sup>159</sup> Parent Interview #5

Something genetically that we can do that the mother could take or the father could take that would change that chromosome or that genetic part of it and mesh it and make it better. Maybe not cure it but make it better to where it's not sometimes so severe.<sup>160</sup>

This parent sought a way to alter or alleviate severe symptoms in a future child. However, as I describe in chapter six, parents of children with ASD who themselves identify as being on the autism spectrum felt that if we proceed down the path of altering “*autistic genes*”, we may lose the very things that make individuals who they are.

These interviews underscore how involvement in a genetic study is intertwined with hope and expectations that it will address major issues facing ASD families. Such expectations are beyond the reach of any current research study on autism, especially given the complexity of genetics and the heterogeneities of ASD that scientist must confront.<sup>161</sup> Although the families who participated in the SSC study are not part of a single parent advocacy organization per se, their actions can be viewed within the “political economy of hope”. This concept was developed by Carlos Novas to describe how families contribute to the transformation of blood, tissue or DNA into resources that generate biovalue such as information or techniques that can be used to enhance human health (Novas, 2006). Many parents saw their participation in the SSC study as a form of activism and believed their participation would help move the understanding of autism forward. Much like the parents in the organization Novas profiles in his analysis of the political economy of hope, PXE International, these parents are placing significant hope in the possibilities of biomedical research to identify causes, develop refined diagnoses, and therapies or cures that will someday benefit future families. However, the

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<sup>160</sup> Parent Interview #7

<sup>161</sup> See Chapter 3 for an analysis of the current state of genetics research, which describes in detail the complexity and heterogeneity of ASD.

“economies of knowledge” fall short for this group of parents as will become evident in the following sections.<sup>162</sup>

### **Genetic Study Concerns**

Starting with the sequencing of the human genome, ethical considerations of genetics research have been a mainstay of research and scholarship in many academic fields such as law, philosophy, public health, medicine, and many others. In the development of genetic databanks, discussions of ethics have generally centered on issues of autonomy (i.e., respect for persons) such as confidentiality, privacy, and concerns around informed consent (Beskow, et al., 2001; Chadwick & Berg, 2001; Knoppers & Chadwick, 2005). Despite the range of ethical problems that have been brought to the forefront of genetics research and the development of genetic databanks, the participants in this study did not have any concerns about participating in genetics research. Only one mom stated that her husband was concerned about having his information in a databank, but she assured him that his privacy would not be compromised.<sup>163</sup> One other parent was concerned but participated despite her worries because she wanted to do anything that would help her son. She described how her uneasy feeling about participating was overcome by *“this thing that is taking these children...it’s so big”*.<sup>164</sup> Another parent felt that if this study was going to help their child, then they were behind it *“100 percent”*,

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<sup>162</sup> Novas argues that the PXE International’s participation in the “economies of knowledge” extends beyond the augmentation of human health and the generation of economic wealth towards the elaboration of new standards relating to how biomedical research should be conducted and how its therapeutic and economic benefits should be distributed (Novas, 2006:297).

<sup>163</sup> Parent Interview #5

<sup>164</sup> Parent Interview #4

despite it being a genetic research study.<sup>165</sup> Thus, the commitment of these parents to help their children was strong enough to override any concerns about participating in genetic research.

The rest of the participants I interviewed expressed no concerns about participating in the SSC study. This is captured in the following quote when I asked a parent whether they were concerned about participating in genetics research:

No. Not at all.... it did not deter us in any way, make us nervous or anything like that....When we read all the information we were okay with it. From day one we were willing to do whatever it takes.<sup>166</sup>

These parents had no concerns about donating blood and family information to the SSC study. Several parents referred to the fact that the study did not physically harm them in any way (i.e., they did not take drugs or endure experimental treatment), and thus posed no threat to their wellbeing. Their experience with the blood draw was also described as relatively smooth. However more surprising and for some a bit intrusive, were the pictures and measurements they took of their heads, ears, hands and feet. Joanna Latimer (2007) refers to such measurements as “the ‘gestalt’ of seeing a syndrome in the face and body of a child.” (p.100). The accumulation of such evidence, Latimer argues, is “accumulated for the possibility that the troubles the child presents with are ‘syndromic’ and that they have a genetic as opposed to some other base.” (Latimer, 2007, p.100) Furthermore, syndromes are made “visible” by “establishing patterns of signs and symptoms across bodily systems as well as across family members (the phenotype)” (Latimer, 2007, p. 105).

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<sup>165</sup> Parent Interview #10

<sup>166</sup> Parent Interview #9

Privacy was also not a huge concern for several parents. In fact the sharing of information in their view was essential for autism awareness and research to progress. For example, one mom was not worried about researchers having information about her family, she stated:

I would share this with anybody who'd listen in hopes that, you know, they would learn from us whatever we're saying and whatever patterns there are they can decode them and say this is what it is. So I really don't worry about that.<sup>167</sup>

Another parent said:

No. I mean, I don't, I can't even think of a concern to have honestly. I mean, I'm not even very concerned about privacy and stuff like that, so, there wasn't really anything that bothered me about it.<sup>168</sup>

Another mom was very open about her son having autism and was not worried if his information was “*leaked*” because, as far as she was concerned, autism was part of their family and it was not something they were hiding.<sup>169</sup>

This lack of concern of about participating in a genetics research study was also tied to the trust parents had in science and in clinical institutions that conduct genetics research. For example, one mom felt “*it was a good thing to do*” because she trusted the Institute conducting the study and felt confident that the research was safe because “*They're looking out for our kids*”.<sup>170</sup> All but two of the parents interviewed were referred to the SSC study by someone they trusted such as an educational professional, pediatrician, or medical institute where their child was receiving services. Thus, it could

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<sup>167</sup> Parent Interview #6

<sup>168</sup> Parent Interview #12

<sup>169</sup> Parent Interview #11

<sup>170</sup> Parent Interview #13

be argued that the trust the families had in the study was a reflection of the trust they had in the people who suggested they participate and in the institutions conducting the study. Furthermore, the fact that so many clinical institutions around the US were spending money and time on this study also indicated for one parent that there must be some scientific basis for it, which gave the whole field credibility.<sup>171</sup>

### **Knowledge about the SSC and Genetics of Human Disease**

The lack of knowledge about the SSC study goals and objectives among the parents could also be related to the lack of concern expressed by them. Only two parents were able to describe the SSC study in terms of the development of a genetic database that would be used by autism researchers throughout the world. Five parents knew the research was related to understanding the genetics of autism by describing the SSC study in terms of identifying a “*genetic link*”, “*genetic cause*”, “*genetic marker*”, “*genetic predisposition*” or a “*clue from our genetic makeup*”. Other parents described the study as a way to find the cause of ASD, how to diagnose it at a specific age, or as a study to properly diagnose children with ASD. One parent recalled that the study was collecting data but was uncertain of the final destination. Two parents wished the study offered genetic tests results back to those who donated, and were surprised when the diagnostic evaluation only had recommendations for future genetic testing rather than the specific results of the genetic tests the SSC study conducted. These findings are surprising given the extensive explanations provided to the parents in the informed consent documents, which clearly indicate the purposes of the research and that results of the genetic tests

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<sup>171</sup> Parent Interview #2



conducted on the DNA sample provided by families would not be revealed to families or to their health care providers. All the families were consented in person prior to participating in the study by the study coordinator,<sup>172</sup> who estimated that the informed consent process took anywhere from 15-45 minutes. According to a SSC study coordinator, only a few families decided not to participate after reviewing the informed consent documents. This was mainly due to ethical concerns of what families might do with genetic information (i.e., abortion) and the risk of having their information in a genetic database. As mentioned before, the parents must sign three documents before they participate: an informed consent document to participate in the study; the Health Insurance Portability and Accountability Act (HIPPA) authorization form; and a blood and cell line banking consent form.

The lack of depth of parents' understanding of the SSC study goals may also be related to the general lack of knowledge the interviewees had of how genetics contributes to human disease. While, all but one parent was able to give some explanation of how genetics contributes to human disease, their descriptions were limited and were often followed by the parents' comment that they "*knew very little*". A few parents provided an explanation in terms of the genetic mechanisms involved in contributing to disease such as "*errors in copying*" or "*abnormalities in the chromosomes*". One parent described the genetic mechanisms the following way:

I think of it just like a computer, you know, or a copy machine. The more copies you make, the more likely there are to be errors and it comes down

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<sup>172</sup> According to the SSC study coordinator, the informed consent documents are not sent ahead of time since the questions could more appropriately be answered in person rather than before hand or over the telephone. There was a concern that if the families received the informed consent documents ahead of time, they would be scared off and not participate in the SSC (Interview with SSC study coordinator).

to just transferring the data. And then the bottom line is sometimes things get a little messed up, and eventually it might develop into something problematic or it may not.<sup>173</sup>

Although their descriptions of how genetics contributes to human disease were vague, ten parents described this in terms of heritability from one generation to the next. For example, one parent described it as starting with one person that gets genetically “*filtered down*”. Three parents described genetics and human disease as something that gets passed down “*from generation to generation*” and one parent described it as “*the genetics of the family [that] affects what genetics the child has*”. Four parents described the genetics of human disease based on their personal family history of disease, such as deafness, sickle cell disease, and cancer. For these interviewees, the descriptions were very specific to their experience, which they generally described as a genetic mutation they inherited from their parents. These descriptions clearly indicate a basic understanding of genetics and human disease to be something that is inherited from one generation to the next.

However, this basic understanding became somewhat complicated when parents learned by participating in the SSC study that the genetic mutation(s) their child might have (assuming, of course, that their child’s ASD is caused by a genetic mutation) could be *de novo* or spontaneous, and not necessarily something that the parents passed onto their child. On the one hand, this hypothesis made sense to the parents in this study since they only had one child with an ASD and the rest of their children were “neurotypical”. Only one family I interviewed thought they had a family history of undiagnosed ASD in the paternal line of their family. The rest of these parents experienced ASD for the first

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<sup>173</sup> Parent Interview #16

time through the diagnosis of their child and never suspected “autism”.<sup>174</sup> However, on the other hand, their notions of what constituted a “genetic disease” were in conflict with the SSC study’s general hypothesis – that for simplex families, ASD is caused by spontaneous mutations in the parental germ line. According to the project manager of one of the SSC study sites, parents were surprised to learn that genetic mutations were not necessarily inherited and could occur spontaneously. She commented that this was a huge issue parents grappled with compared to the few concerns they had about genetic privacy and the potential use of the genetic database in future. Thus, the expansion of parents’ genetic understanding of disease, from inherited single gene mutations to include copy number variants that are spontaneously acquired, engender a new level of ambiguity in their understanding of genetic contributions to disease and heritability of complex disorders like autism.

### **What if there was a Genetic Test?**

The parents in this study were asked to comment on what they would do if there were a genetic test that could offer information about their risks of having a child with autism. The most frequent response was that this information would benefit their family’s future generations, since there was not much they could do with that information today. Parents felt that it would be useful to know if their child’s ASD would be passed down

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<sup>174</sup> Families who had a family history of ASD were excluded from the SSC study, thus this finding is not surprising. A developmental pediatrician also interviewed the parents to determine whether parents displayed any “autism phenotypes”. If a parent appeared to be on the “autism spectrum” they were excluded from the study. Similar to Joanna Latimer’s (2007) work on dysmorphology, some of the things about the child that appear unusual and abnormal, such as particular faces, sizes, and shapes, may gain their significance as signs when they are compared with the features of other family members.

genetically if they had children in the future, whether it would skip a generation, or be worse with every generation. One mom responded by saying, *“if they truly say it could be genetic and things are passed on in carriers and identifying those, it sure could affect our children's children and so forth. That would be information that would be nice to have.”*<sup>175</sup> Parents also felt that genetic information would have an impact on their ‘typical’ children. One mom was concerned about her older son (who does not have an ASD) and whether he would want to have children if they knew it was something that could be passed on genetically. Although her son wants to have children of his own in the future, he is skeptical based on his experiences of how hard it is to have a brother with ASD.<sup>176</sup>

The second most common response was that genetic information about the risk of ASD, whether provided prenatally or when the child was born, would help parents prepare for a child with a disability. One mom was hoping genetic information would provide a *“definitive test”* to determine if a child has an ASD. She felt that knowing that autism was a possibility through genetic information would help parents be more abreast of the signs and issues that may arise by *“keeping their eyes open”*.<sup>177</sup> Several parents stated that their pediatrician did not take their worries seriously and wished that there were some definitive way that could have identified ASD earlier. This would allow parents to take action sooner, such as starting therapies that are known to only benefit a child at an early age. This was especially true for parents of older children who had a delayed or incorrect diagnosis due to the lack of awareness of ASD in the early 90’s

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<sup>175</sup> Parent Interview #10

<sup>176</sup> Parent Interview #14

<sup>177</sup> Parent Interview #13

among medical and educational specialists. However, two parents expressed their concerns that genetic information would only help those who could afford long term therapies and would further increase the disparities that exist among those who can and can not afford diagnosis and treatment.

### **Progress of Science and What Families Need in the Future**

The parents interviewed in this study generally felt that the progress of research was somewhat limited and very slow since they became part of the ASD community. Four parents commented on the lack of progress in finding the causes of ASD. A few parents mentioned that there was limited progress made in testing and developing different treatments and finding a link to genetics. One mom described this lack of progress as a result of research “*going in a lot of directions*”, which have not identified a common causal factor. However, one mother, who was among the most scientifically savvy parents I interviewed, saw progress in the fact that we were no longer institutionalizing children and/or blaming the mother as the cause of ASD.

Parents did agree on the progress made regarding the awareness of ASD. Six parents acknowledged this progress in various ways. One family felt there was more information about ASD than there was a few years ago, especially about diagnosis and awareness of Asperger syndrome. Another mom felt that there was more awareness of ASD through the media, a result of Autism Awareness Month.

I asked the parents what they felt should be the priorities for autism research and they responded with four main issues: identifying the causes; increasing awareness; identifying and testing new treatments; and helping adults with ASD. Among parents

who wished future research to address the causes of autism (N=9), four parents specifically mentioned that environmental causes needed to be considered and three parents wanted to see continued research on genetics. Parents described these as “*internal or external causes*” and referred to “*external*” causes as “*the medicine, procedures [done during pregnancy and delivery], chemical aspects of what’s being put into the foods*”, “*vaccinations*”, or “*pregnancy and birth conditions*”. These particular “*external*” exposures were directly related to the experiences of the parents and what they thought might be the cause of their own child’s ASD.

Nine parents also felt that the future of ASD research should focus on increasing the awareness of ASD in the general public, and especially among professionals who interact with children such as pediatricians and teachers. Parents wanted to see more research focused on educating schools and pediatricians about ASD and how to identify it sooner. Over half the parents mentioned that they were given the diagnosis without any directions to resources on how to proceed, such as identifying the therapies that work best, how to work with the school system, and how to navigate and coordinate all the different therapists, doctors, special educational teachers, and insurance companies. These constituted “*all the pieces that can benefit the child*”. Thus, early identification of ASD through increased awareness of professionals who interact with children also needs to be accompanied by access to “*trusted*” resources parents can use to move forward and help their child.

Parents also wanted to see more awareness efforts in training educators about how to help autistic children. These parents felt that some educators did not know how to work with a child with an ASD. One mother stated:

Hopefully that after it's all said and done and they've done the research, hopefully, a lot of people would have a better perspective on autism. That don't have the problems that I've encountered where an educator don't know how to deal with a child. That it can help teach educators. And help them to do their job better.<sup>178</sup>

This mom, like many other parents, struggled with the educational system and was frustrated with the lack of attention given to the needs of her child. Most parents talked about how they had to advocate for their child in school and tried desperately to get their child into mainstream classes so they could model their typical peers. Parents whose children were diagnosed with higher functioning labels such as Asperger syndrome or pervasive developmental disorder – not otherwise specified (PDD-NOS), also wanted research to focus on the best ways to train educators of the broad spectrum of ASD and how to best address the needs of children who are higher functioning. These parents felt that their children were often overlooked because they were verbal and highly intelligent, but lacked key social and coping skills of everyday interactions and activities. As one mom commented about how teachers can help their children in the classroom, *“if they're having issues with social interaction be there. Be able to have either somebody there to see the signs and get them back before it goes too far.”*<sup>179</sup>

Identifying and testing new treatments were also among the highest priorities parents had for the future of autism research (N=8). Much like the need for increasing awareness of ASD in pediatricians and educators, the desire for future research on treatments was related to the need for resources that provide information on the best therapies available based on the specific needs of a child. Since autism has such a huge

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<sup>178</sup> Parent Interview #9

<sup>179</sup> Parent Interview #7

spectrum, parents felt that treatment options also needed to be varied and tailored. However, there was no useful way to navigate all the different treatment options available, to determine which ones to trust, and to ascertain how long it should take for certain treatments to be effective. One parent expressed these concerns in the following way:

It would be great if the scientific community could really start evaluating some of these treatment options that are out there and weeding the ones out that aren't really effective. Or even if you can say this is effective for kids who have this. But it's not effective for kids who have that.<sup>180</sup>

Thus, parents wanted scientists to focus their research on the effectiveness of different treatment options available that do not have any scientific research supporting their effectiveness such as the gluten and casein free diets, water therapy, or music therapy.<sup>181</sup> This would save parents a lot of time, energy, money and frustration in navigating many of these therapeutic possibilities. Finally, parents specifically mentioned that they wanted future research to consider cheaper therapy options besides Applied Behavioral Analysis (ABA), which is generally only effective in younger children and is so expensive that most parents cannot afford long-term treatment.<sup>182</sup>

The future of children diagnosed with ASD was also among the most important research foci parents wanted to see in the future (N=9). One mom described it in the following way, “*the United States or everywhere this happens has got to realize that*

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<sup>180</sup> Parent Interview #13

<sup>181</sup> For a thorough list of different autism treatments ranging from applied behavioral analysis (ABA) to biomedical and dietary treatments see Kyle's Treehouse at <http://www.kylestreehouse.org/treatments.aspx>.

<sup>182</sup> The cost of applied behavioral analysis (ABA) can range from \$5,000 to \$50,000 a year depending on how it is implemented (i.e., through specialized schools, using therapists in training or college students in home, or a trained ABA therapist) and the number of hours per week. ABA usually recommends 30-40 hours a week.



*these kids are growing up, they've got to function, and they've got to have a capacity in the social sector and acceptance.*"<sup>183</sup> Like many parents, this mom was worried about the future of her child who was entering his teenage years and was still having trouble with everyday social interactions. These parents recognize that ASD is a life long disorder and does not stop when they are 18 years old, the age when services for children diagnosed with ASD end. "*This is our future*", one mom stated who was worried about the future of her son, "*and if our future can't concentrate very well and can't function very well, then it's going to be a huge burden on the ones that are 'normal'*".<sup>184</sup> In this case, this parent was worried about the unaffected siblings who would be responsible for not only taking care of their elderly parents but also a sibling who has ASD. These parents wanted to see future research focus on how these children could be enabled to function in college, obtain a job, and live an independent life.

These research requests represent priorities most beneficial to families with ASD. They also echo the Strategic Plan developed by the Interagency Autism Coordinating Committee (IACC). The IACC claims they took the needs of people with ASD and their families, as well as other consumers of these efforts, into account in developing specific research priorities for autism (DHHS, 2009b). IACC research priorities similar to the findings of this study include: identifying the causes of ASD; determining what treatments will help; details on how to obtain services; and the future of adults with ASD.<sup>185</sup> The research priorities posed by the parents I interviewed were also tied to their

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<sup>183</sup> Parent Interview #14

<sup>184</sup> Parent Interview #6

<sup>185</sup> At the 2010 International Meeting for Autism Research, the director of the National Institute of Mental Health, Tom Insel, stated that public comments on the 2009 IACC Strategic Plan emphasized more research on environmental factors, nonverbal individuals with ASD, adults with

motivations to participate in and hopes for autism research, indicating that these processes are aligned in many ways. These research priorities also highlight that although these parents participated in a genetics research study, they did not hold this area of research as a sole priority for the future.

### **“We are Not a Science Project”**

For many parents, the SSC study was their first encounter with scientists, and thus the nature of their experiences may reflect their overall impression of how they think scientists view parent involvement in research studies. Overall, most of the parents interviewed had good experiences in participating in the SSC study. Five parents specifically felt that the study was genuinely interested in the day-to-day experiences and concerns of the family. These parents felt the scientists viewed their contributions as an *“important piece of the puzzle”* and represented families by taking detailed histories of all the major events that occurred in their child’s life. As one mom explained, *“I felt that they really wanted to get to know who we were, how we lived, our concerns and how we go day to day”*.<sup>186</sup> These parents felt that they were an important part of the process and the needs of their families were taken very seriously throughout the study participation.

Not all the parents, however, felt that their participation in the SSC study was appreciated. One father, whose son was diagnosed with an ASD, felt that the SSC study did not really *“see the parents”*. He compared his participation to that of a processing

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ASD and delivery of health services tailored for people with ASD. Thus, the 2010 updated plan will have twice the number of research objectives than the 2009 plan to address these issues (Autism Speaks: From molecules to medicine: Thoughts on the 2010 International Meeting for Autism Research (IMFAR) from the chief science officer. Retrieved May 29, 2010, from [http://www.autismspeaks.org/science/science\\_news/geri\\_dawson\\_imfar\\_2010\\_recap.php](http://www.autismspeaks.org/science/science_news/geri_dawson_imfar_2010_recap.php)).

<sup>186</sup> Parent Interview #7

plant where the study coordinators “*came in, took the blood and chatted with us for a second.*” This family was particularly disappointed with the lack of information they got back from the study and was hoping for more than just an evaluation of their son.<sup>187</sup> In fact, five parents wished that the results of the study would become available to the parents once anything (no matter how minor) was found. They saw this as one way to increase the awareness of ASD research and “*only fair*” given the sacrifices and contributions families make to participate in research studies.

In addition to sharing research results, parents also hoped that scientists in general would embrace family experiences as an important piece of data. Seven parents really hoped that the scientists appreciated their participation because as they saw it, they knew their children better than anybody else and their contributions regarding every day life experiences were valuable components of any research study. One mom felt that families who participate in research should be placed on a pedestal because, as she explained it, “*if it wasn't for us they couldn't do their job*”.<sup>188</sup> This statement is true given the nature of the SSC study and other collections like the AGRE, which is to build a database of DNA and clinical information based entirely on families who have children diagnosed with ASD.

These parents also wanted scientists to consider them to be more than just a sample number or a science project when they are conducting and representing their research. One parent offered the following advice to scientists:

Keep in mind that this is not a science project to the parent... because they have a child who is afflicted or affected by this and they want to fix

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<sup>187</sup> Parent Interview #5

<sup>188</sup> Parent Interview #9

things either for themselves or try to help other people. And it is very dear to their heart.<sup>189</sup>

This parent also emphasized that her family's participation in research was important and meaningful to them and only hoped that it would be equally important to the scientists utilizing their data. Thus, these parents want to remind scientists that they are more than just a sample number but real families "*dealing and living it.... [and] trying to get their kid through the day*".<sup>190</sup>

### **Ethical Implications of Participating in Genetics Research**

#### ***"Desperate for Answers"***

The social and moral contexts within which parents of children diagnosed with an ASD come to participate in genetics research imply a wider concept of engagement and a level of participation that moves beyond the conventional mode of bioethics formulations. Traditionally, bioethics has been concerned with securing individuals' informed consent and ensuring that confidentiality and privacy are respected. However, as the data suggest, confidentiality and privacy concerns about participating in genetics research and donating blood and medical information to a genetic database were not major concerns for parents interviewed in this study. Their daily life concerns about having a child with ASD and desire for answers trumped any worries they had about participating. Furthermore, their moral duty as parents to participate in anyway to help their child or families in the future outweighed any doubts they may have had.

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<sup>189</sup> Parent Interview #13

<sup>190</sup> Parent Interview #14

Parents' lack of concern about participating in a genetics research study also brings up important issues of knowledge and trust. As Sharon Kaufman (2010) points out in her analysis of parent anxiety over the connection between autism and vaccines, the role of trust in systems of knowledge are "fundamental to grounding in the empirical world, and...remains background to everyday judgment and action most of the time." (p.24) The dynamics of knowledge and trust in participating in genetics research and their embeddedness within the context of work and family are issues brought forth in the current study, as well as others (Busby, 2004; Tutton, 2007). For example, Helen Busby argues that contrary to the emphasis in bioethics of the importance of conveying greater levels of information as a way of facilitating trust (i.e., the careful implementation of informed consent), substantial trust often arises instead from the perceptions of and relationships with the institutions conducting the research (Busby, 2004). This is clearly the case for the parents I interviewed who participated in the SSC study. They had a clear level of trust in the academic institute conducting the research, as well as in the professionals (i.e., doctors, educators, and therapists) who referred them to the study. The academic institute also collaborated with the local autism center for the recruitment and collection of participants for the SSC study, which many parents already knew through the services they offer children and families with ASD. Thus, the limited of knowledge that many of the participants had about specific aspects of the study itself were to some degree linked to their choice to place trust in the academic institution conducting the research. This level of trust is imperative for studies like the SSC study to succeed, which really calls into question the implications of having limited knowledge and expertise in relation to research participation (Busby, 2004).

However, when trust is less a matter of choice and more a matter of hope and resignation, which is often the case for the parents who participated in the study for the free diagnostic evaluation and professional recommendations about how they could help their child, the ethical and social implications become somewhat more complicated. These parents did not think twice about being part of a national genetic database. They were more concerned about getting a proper diagnosis, normally costing up to \$2000, and could take up to a year just to get an appointment for an evaluation. This motivation clearly blurs the boundaries of bioethics and is a strong example where the research situation could be construed as coercive to a certain degree. Typically, payment for participation is generally supposed to be quite minor such as a gift certificate, or compensation for travel and lodging. These parents saw the SSC study as an opportunity to find out “once and for all”, whether their child was on the autism spectrum and were hoping that the evaluation would open a passage into ways to better help their children. As Sarah Cunningham-Burley also points out, such vulnerability may lead to expectations of research far beyond realistic longer-term outcomes (Cunningham-Burley, 2006). This also appears to be the case in the current study, where expectations for the SSC study were beyond the goals of the study for several of the parents. Not surprisingly, these parents were also less likely to truly understand the goals of the study to the point where one parent thought the goals of the research were to properly diagnose children with ASD.

This case study also shows that there are many “styles of participation” embraced by these parents, including the “active participant”, and the “cost/benefit participant” (Haimes & Whong-Barr, 2004). Based on this data, there are also the “desperate

participants”, those who participate out of longing and desperation for answers. All these styles of participation include the emotional knowledge or expertise the parents bring to the table regarding raising a child on the autism spectrum. The emotional need for answers were also evident as parents attempt to navigate the diagnostic odyssey of ASD, confront the lack of resources available once given a diagnosis, and face their new position as coordinators of and advocates for their children’s therapies and educational services. Thus, emotional knowledge and need were deeply connected to parents’ motivations to participate and the hopes they had for the outcomes of research on the genetics of autism.

The limited concerns parents had about participating in a genetics research study were also tied to the political economy of hope mentioned earlier (Novas, 2006), however their “economies of knowledge” are less formulated than those of larger advocacy groups such as Cure Autism Now. As discussed in chapter two, these parents became experts in the science of autism and initiated the development of the Autism Genetic Resource Exchange to advance the biomedical understanding of autism.<sup>191</sup> Although the parents interviewed in this study had limited expertise about the science of autism generally and genetics more specifically, like the parents of CAN, they were able to draw on their emotional knowledge (Lindee, 2005) of having a child with ASD. This emotional knowledge is deeply connected to their motivations to participate, the hope they have for the future of their children, as well as the outcomes of research on the genetics of autism. Furthermore, the parent’s expectations for genetics research to find a cause, improve

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<sup>191</sup> See Chapter 2 for a complete description and analysis of the AGRE project.

diagnoses, or “fix” their child could also underline the ways in which parents come to grips with the limitations of their knowledge and expertise.

Another ethical consideration was the lengthy time it took for the SSC study to return the research evaluations back to the parents who participated. This was among the biggest complaint parents had about the study in general, especially for parents relying on the research evaluation to aid in access to educational services. Some parents had been waiting for their results for over six months. Once they finally got the results back, however, the terminology or “*foreign language*” used in the research evaluation confused several of the parents. They were also disappointed with the lack of recommendations on how to specifically help their child. For example, one family was not sure about the scores given to their son, whether they were “*good or bad*,” and wished the study provided a follow up visit with the family to answer questions about the evaluation.

This raises ethical issues of follow up in research. Specifically, whether it should be part of the study protocol to verbally follow up with participants when they receive a specific diagnosis, especially for those receiving a diagnosis for the first time. Furthermore, the research evaluations I read only listed resources to national organizations such as Autism Speaks, the Center for Disease Control and Prevention, and the National Institute of Mental Health. Local agencies that could be more beneficial to the parents were not included. Although the report supplied examples of how to address difficulties such as articulation and pragmatic skills or explicit instruction and intervention in social skills, parents thought the report fell short regarding where and how to obtain these types of services.



### ***Implications of the Long-Term Use of Genetic Databases***

A unique aspect of the SSC study is that it focuses on a specific disorder (ASD) and involves the collection of data from children who are affected as well as one of their unaffected siblings. The long-term use of their data for unspecified research purposes in the future raises issues that generate bioethical debates about whether children are able to consent to genetics research. Social scientists have begun to question who has a voice in generating discourses about genetic databases and who remains or is forced to remain silent? (Haimes & Whong-Barr, 2004; Williamson, Goodenough, Kent, & Ashcroft, 2004). The children who participate in the SSC study range from ages 4 – 17, their voices are likely to remain silent for quite some time. Haimes and Whong-Barr (2004) describe children in this context as “passive participants” whose consent to participate must be revisited when they grow up into teenagehood and adulthood. Williamson and colleagues (2004) argue that children’s views are important for ensuring that their own concerns and interest are addressed both when they are children and when they become adults. They found that children currently underestimate the amount of control that they have with regard to their participation in non-therapeutic research and question the ‘right’ of parents to consent to the long-term use of their children’s genetic information (Williamson et al., 2004, p.157).

Drawing on situational analysis (Clarke, 2005; Clarke & Montini, 1993), children who participate in data collection efforts like the SSC study can be referred to as “implicated actors” because although they are physically present in a given social world, they are generally silenced by those in power. In genetic databases such as the SSC and the Autism Genetic Resource Exchange (AGRE), children with ASD, as well as their

parents and unaffected siblings are represented through an empirical mode of inquiry (e.g., blood sample, extensive interviews and observations). However, the children (both affected and unaffected siblings) hold less power since the decision to participate is based solely on the parents' willingness to be part of the study. Thus, children are implicated actors in the autism genetics arena. They are physically present during the data collection but are not actively involved in the negotiations to participate. Although the future consequences of parents' actions to participate in long-term genetics research were not issues brought up in the interview (nor did I inquire about them), these issues will be an important area of research for social scientists in the future concerned about the ethics of children participating in genetics research and the long-term use of biological materials.

Because samples can be immortalized and stored indefinitely, genetic databases also present a particular challenge to informed consent requirements. It is also often impossible to anticipate the types of studies that will utilize the samples in the future given the constant changes in genetic knowledge and technologies. This raises another ethical concern of how often families should be burdened with follow up studies if scientists 'discover' a genetic mutation in only a select number of families. This issue was brought to my attention by a study coordinator from another ASD genetic database (i.e., AGRE). It raises questions as to how often families should be re-contacted if their DNA is deemed valuable to the scientific enterprise and to what degree families should learn about the uniqueness of their DNA? What counts as a "causal" genetic mutation is also debated among scientists, which poses ethical problems of reporting genetic information back to participants that may or may not be the cause of their child's ASD.

The ethical dilemma here lies in scientists not yet fully understanding new genetic anomalies such as copy number variants (CNVs).<sup>192</sup>

### ***Returning Results from Genetics Research***

The reporting of genetics research results was also a point of confusion for the parents interviewed in this study. The SSC informed consent documents clearly states:

In regards to the genetic testing that will be conducted on the DNA sample you provide, the research team does *not* expect that the results of these tests would be medically helpful for you to know. Therefore, as a general policy, the team does not reveal the results of these tests to you or your doctor or other health-care providers. (my emphasis)

However, the SSC study informed consent document also states in the following paragraph that in the unlikely chance that genetic information identified in the study would be medically helpful for the family, the parents had to make a choice on how the research team should handle genetic testing results. Parents could either receive the results, under the stipulation that this would be in the form of a referral to experts in medical genetics and not covered by the research team, or they could choose not to receive any such information. Thus, parents did not receive any formal genetic information, only a referral. Several parents in this study were confused about this and were genuinely surprised that they did not receive any genetic information in return for their participation.

This raises ethical issues of reporting genetic or other information to participants in research studies (i.e., duty to disclose). According to Miller and colleagues (2010), specific disclosure standards are appropriate for different disease contexts. For autism,

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<sup>192</sup> For a complete review and analysis of CNVs see Chapter 3.

these authors found that researchers and parents set a standard of reportability that reflected the kind of meaning autism genetics research results might yield, such as explaining the cause. However, evidentiary standards within specific research disciplines (i.e., research, clinical, or statistics), as well as fundamental theories about how autism is “genetic” (or not) influenced whether or not results were deemed “true” (F. A. Miller, et al., 2010). Thus, consensus disclosure standards are unlikely to work well here because they do not take into consideration appropriate evidentiary standards and the status of “real time epistemological debates regarding the nature and cause of a given disorder” (Miller, et al., 2010, p.5).

### ***“The Ethos of Hope”***

When you are making decisions about your child’s life, health, and future based on obviously bad information, you are definitely taking risks. The risks from inaction could be as devastating as taking the wrong action. These are not the kind of risks any parent should ever be faced with (Cody, 2006)(p.797)

The range of ethical implications brought forth in the narratives of parents who participate in genetics research highlight many contextual issues not often considered in current bioethical frameworks. To further articulate the broader social terrain within which parents are embedded as they come to participate in genetics research, I draw on Nikolas Rose’s concept of the “ethos of hope” (Rose, 2007, p.27). In the processes of knowledge production regarding ASD, this concept links together many different actors: parents of children with ASD, individuals with ASD, and parent advocacy groups hoping to find the cause, improve therapy, or a cure; scientists studying autism genetics hoping for a breakthrough to advance their careers; developmental pediatricians, neurologists,

psychologists, occupational therapists, speech therapists, and special educational teachers treating their patients; governments obliged to act for the common good as well as generate economic activity through industrial and commercial developments; and pharmaceutical and biotechnology companies potentially generating profits from genetic research enterprises.

This economy of hope, according to Rose (2007), is one dimension of a wider shift that he describes as “ethopolitics”, a concept that “attempts to shape the conduct of human beings by acting upon their sentiments, beliefs, and values” (Rose, 2007, p.27). Although the parents in this study are not concerned necessarily with “self-techniques” to improve *their* health, they are concerned with techniques that they can “judge and act upon” to make their children “better than they are” (Rose, 2007). The responsibility parents take upon themselves to help their children and families in the future also speaks to the notion of biological ethopolitics and the novel forms of authority parents assume to help their children.

These economies of hope, however, must be examined vis-à-vis the prevalent views among researchers, institutions, and sponsors in regard to the allocation of intellectual property rights and associated profits that can result from genetics research (Beskow, et al., 2001; Merz, Magnus, Cho, & Caplan, 2002). The SSC study consent form clearly states:

.. blood removed from you for this study may be valuable for scientific, research, or teaching purposes, or for the development of new medical products. For example, the analysis of your blood samples may contribute to the creation of new diagnostic tests, new medicines, or other uses that may be commercially valuable to the sponsor. Neither you nor your child will receive any financial benefits and may not receive any health-related benefits from such developments.

Thus, the interests of the donors, in this case the Simons Foundation, and institutions and individuals (e.g., universities and researchers) do not consider affected families among their primary interests. Although the Simons Foundation is technically a private advocacy group, the study is being run like a for-profit business venture, which, according to one SSC study coordinator, has been problematic for the study investigators. Although none of the participants in this study mentioned the concern or implications of future commercialization of the research generated from their blood and medical information, these parents were participating with the understanding that future families would benefit from this research. As Merz and colleagues (2002) and others (Haddow, Laurie, Cunningham-Burley, & Hunter, 2006; Hayden, 2007) have pointed out, this altruistic notion of participants needs to be re-examined to consider the value added to the research enterprise by parents and their families willing to participate in ways that would recognize and reward their contributions.

In conclusion, this analysis demonstrates the ways in which parents of children come to participate in genetics research. Many uncertainties, forms of knowledge, and emotional experiences flow from the decision and experience of donating blood and medical information to a national disease-specific genetic database. Unlike those parents involved in the early days of the AGRE project, these parents are being collectively drawn together by the research study itself, rather than through the grassroots efforts of collective mobilization to “speed up science”. Thus, the information they receive and the potential benefits that may result from the research are under the control of sponsors funding the research. These interviews also highlight ethical implications that extend

beyond traditional bioethics and engage in a new dialogue of bioethics that takes into consideration the social and moral situations of families living everyday with ASD and who want only the best for their child.

## **CHAPTER 5: SOCIAL AND SCIENTIFIC ‘TRANSCRIPTIONS’ AND ‘TRANSLATIONS’ OF FAMILY DNA**

This dissertation considers various social worlds involved in the production of autism genetic knowledge. However, as highlighted in the introduction and elsewhere, these social worlds do not interact only among themselves but also with other social worlds. Thus, this chapter will attempt to “follow the DNA” to highlight the ways in which family information (i.e., blood and family characteristics) are transformed and processed into genetic knowledge through the different yet overlapping spaces of families and individuals with autism, parent advocacy groups, and scientists. Many processes of transformation take place starting from the emotional knowledge of parents who participate in genetics research, to the transformations of DNA sequences into genetic knowledge that explain molecular and neurological pathways of ASD. These transformations represent the ongoing array of possibilities, negotiations, heterogeneities, contradictions, situatedness, fragmentations, partialities, and positionalities involved in such knowledge production (Clarke, 2005).

The focus of this chapter is to conceptualize the boundaries of social worlds that come together in the production of genetic knowledge on autism. I draw on work that theorizes boundary objects (Star & Griesemer, 1989) and local contingencies, uncertainties, differences, and processes that are often “lost in translation” during the production of genetic knowledge (Fujimura, 2005; Fujimura & Fortun, 1996). Following Joan Fujimura, translation in this sense “can distort, transform, delete, and add.” (Fujimura, 2005:220). Inspired by these theoretical concepts, I would like to elucidate



what I refer to as the “social and scientific transcriptions and translations of family DNA”. By reversing and distorting the scientific dogma of “RNA > DNA > protein > organ > systems > organism”, I will sociologically unpack the intermediary processes of transcription/translation to reveal social and scientific processes described throughout this dissertation. In this analysis I draw from several sites of linkage analyzed in this study, especially data derived from families who participate in genetics research by donating to DNA repositories (i.e., AGRE and SSC), and scientists who are members of the AGP and/or utilize the AGRE database.

The explication of these processes is particularly important because it allows me to elucidate the transformations of family experiences and their material blood, to the creation of pedigrees, the generation of immortalized cell lines, the creation of bioinformatic and genetic databases, the production of DNA, the identification of genotypes, the “discovery” of nucleotide alterations, and finally, the scientific interpretations of microarray fluorescence intensities. By “following the DNA”, I can analyze the boundaries in which these transformations take place and the people, institutions, tools, and technologies needed to enable such transformations. I also show how the movement of biological materials and clinical information from families, to central repositories, to laboratories around the world, and back again are fluid and multi-directional processes.

In this chapter I identify four sites of transcription/translation processes:

- a) The transformation of parent interviews and child observations into a quantifiable and standardized database (transcription)

- b) The creation of family pedigrees, which is based strictly on biological relationships (transcription)
- c) The process of immortalizing family blood cells (translation)
- d) The process of scientific inquiry using various genetic technologies (translation)

I argue that within the scientific domain, the processes of transcription/translation may distort, transform and in some cases delete families. Furthermore, scientists use the family's biological relationships in order to support their research results, however, family relations are literally deleted from the discourse once individual chromosomal characteristics are analyzed. In essence, scientists who utilize the AGRE or SSC for their research are re-constructing genotypic and phenotypic characteristics of the family data to generate scientific meanings of ASD.

### **Autism Everyday**

I start this analysis by first considering the families who participated in genetic research by donating biological materials (i.e., blood) and phenotype information (i.e., clinical information and family characteristic traits), either through the AGRE or the SSC. As I described in chapter four, families who participated in the Simons Simplex Collection (SSC) were not the type of parent advocates who promoted and supported the biological understanding of autism described in chapter two (e.g., CAN and NAAR). Rather, these families, who also similar to families of CAN/AGRE in this regard, participated based on their desire to help their children and families in the future. As they saw it, participation in the SSC was a means to an end and being a part of research was a

very important process. For some parents, the desire to participate in genetics research was driven by their need for answers and a proper diagnosis that would entitle their children to educational services.

Many of these families invited me to their homes to conduct the interviews. Here, I experienced first hand a glimpse of the everyday lives of families whose children are diagnosed with ASD. If I did not get to meet the entire family personally, most parents brought out pictures of their children to help give me perspectives on the individuals our conversation was centered around. I could and did easily relate to these families. They were present, alive, living, breathing, working, and trying to make each day the best they could for their children.

Throughout the interviews, these families described to me the daily challenges (and blessings) of having children diagnosed on the autism spectrum. Many parents described their lives as “*alternative*”, “*a daily struggle*” or a series of “*accommodations and rearrangements*”. Although I did not interview the families of AGRE, many of the themes brought up by these SSC families were apparent in the AGRE families’ stories highlighted in the AGRE newsletters. For example, one parent stated:

We’ve managed to develop our own lifestyle. Short visits work best, going places early, getting tag- a-longs so we could take family bike rides. Using Thomas the Tank Engine to get [our son] to do his homework. Centering vacations around swimming. Using special park programs and other resources to give the boys opportunities and us respite. Setting up schedules. Sometimes we realize that it’s easier to stay home.<sup>193</sup>

Parents regarded their knowledge of living with autism everyday as an invaluable contribution to science. As one mom stated, “*you get more information from the family*

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<sup>193</sup> From AGRE Newsletter (2004): Parents of a two boys diagnosed with autism who participated in the AGRE.

*than you do from observing the child. You need everything but there's so much information.*” Thus, the lay knowledge based on the lived experiences of families came from an emotional perspective and not what one mom termed a “*clean slate of objectivity.*” In chapters two and four, I discuss how emotional knowledge of parents with a child or children with ASD drove them to want to speed up the pace of biological research on autism (e.g., CAN/AGRE) and served as a motivating factor to participate in genetics research.

### **“Turning Stories into Numbers”**

One of the prevailing themes that emerged from this data is the incredible variability of symptoms of children (and adults) who are on the autism spectrum. Significantly, the heterogeneity of ASD had quite different implications for the various ASD sites investigated. For example, parents who participated in the SSC made comments about the broad spectrum of ASD by stating: “*There’s no one category that they fit in.*” “*If you know [only] one kid you don’t know autism.*” “*There's no one way to be autistic.*” And “*The spectrum is so broad and there are so many little intricacies throughout the spectrum that you're never going to find two kids that are exactly alike.*” As discussed in chapter four, this variability has contributed substantially to the diagnostic odyssey parent’s experience. Adults on the autism spectrum struggled with the issue of heterogeneity because they felt that often once people knew they had autism, stereotypes emerged, such as being non-verbal and unintelligent. Thus, as I discussed in chapter six, adults diagnosed or self-identified with ASD wished for future scientific investigations that focused on the vast differences among people with autism and how

different manifestations of the same illness might be incorporated into specific diagnoses and targeted treatment.

For scientists, both the phenotypic and genotypic heterogeneity of ASD were challenging issues for genetics research. This is now being addressed scientifically through genetic reclassification of ASD phenotypes, as well as the standardization of diagnostic tools. I learned through this investigation that initially non-psychiatrists such as neurologists and geneticists were the professionals requesting standardized diagnostic tools for ASD (i.e., ADI-R, a diagnostic interview of caregivers and the ADOS, a diagnostic observation conducted by a trained professional). These investigators were not qualified to diagnose ASD, and thus needed standardized diagnostic tools in order to secure funding from the NIH. The use of these standardizing tools was also needed when large collaborations such as the AGP were starting up in order to be able to combine samples for larger genetic analysis. Further, the development of AGRE and the pressure by scientists for this parent-driven initiative to be scientifically credible, pushed AGRE to use the ADI-R and ADOS for the samples they collected. Finally, the CPEA centers established by the NIH in 1997, had to agree on standardized diagnostic tools for research, and settled on the ADI-R and the ADOS. Thus, it was the research communities, as opposed to clinical communities, that were responsible for the use and promotion of these particular standardized diagnostic instruments for autism. The dominance of research needs is also becoming apparent in the Diagnostic Statistical Manual for Mental Health Disorders version five (DSM-V), currently being revised to improve its utility for scientific research. Although these tools were largely driven by the

needs of science, their use impacts many actors involved, including: individuals with ASD, parents, clinicians, scientists, and health and educational policy makers.

Thus, the heterogeneity of ASD brings up issues of classification of ASD and standardizations of research samples used for ASD research. Bowker and Star (1999) carefully distinguish these definitions by taking a Pragmatic approach. They define a “classification system” as “a set of boxes into which things can be put to then do some kind of work” (p. 10). For ASD, the classification systems used are the DSM-IVR and the ICD-10 (International Statistical Classification of Diseases and Related Health Problems; Tenth Revision). These classification systems are predominately used to define ASD. Bowker and Star (1999) point out that not all classifications become standardized, however every standard imposes a classifications system. Thus, standards have several idealized dimensions including: agreed upon rules for the production of objects; ability to span more than one community of practice; allow things to work together over distance and heterogeneous metrics; often enforced by legal bodies; and the use of the best standard follows no natural law (Bowker and Star, 1999, p.14). In the case of ASD, standardized diagnostic tools, (e.g., ADI-R and ADOS) were needed for the scientific production of knowledge. These tools allowed for a consistent set of symptoms (i.e., phenotypes) to be measured and counted in the process of scientific inquiries of ASD. The NIH also enforced the use of these tools to allow data to be pooled together across a geographic and disciplinary range of investigators.

The majority of parents interviewed saw their child’s limited social skills as a major “symptom” of ASD (n=13). Second were problems with language, and speech (n=8) and sensory issues (n=8). Third were rigid and literal behaviors (n=7) and intensely

focused interests on particular things or subjects (n=7). The major concerns of adults on the autism spectrum, who I interviewed, revolved around communication and social interaction. They experienced high levels of anxiety and, for some, major depression or obsessive behavior (Singh, 2006). This range of symptoms represents some of the observations “captured” in the data collection process for both AGRE and SSC, where some symptoms are regarded more important than others. For example, the “delay or total lack of spoken language, not compensated by gesture” or “encompassing preoccupations” are two measures taken in the Autism Diagnostic Interview – Revised (ADI-R) (Lord, Rutter, & LeCouteur, 1994). However, symptoms such as sensory issues or depression are not systematically collected despite being among the top issues parents as well as adults on the autism spectrum experienced and noted.

As described in chapter four, the “observed” experience is based on parent interviews and professional observations of children suspected to be on the autism spectrum. However, those observations are organized and defined based on standardized diagnostic tools like ADI-R and the ADOS. Thus, the first level of transcription/translation of parent experiences is immediately lost based on the limitations of the diagnostic tools used to collect research data. However, as Star and Griesemer (1989: 407) point out in their analysis of boundary objects, standardization “makes information compatible and allows for a longer 'reach' across divergent worlds.” (Star and Griesemer, These authors argue that standardization of data collection and techniques provide information for future generations or for researchers at a distance. Therefore, although the creation of new scientific knowledge requires lay knowledge of “non-scientific” participants (i.e., families with ASD), this knowledge is “translated” or re-

interpreted by scientists to fit their own goals. The standardization of diagnostic tools used for both the AGRE and the SSC thus eliminate much of the lived experiences of parents (and ASD adults), as well as symptoms not officially recognized by the DSM – IV. The standardization of ASD diagnostic tools reflects what Andrew Lakhoff (2008) describes as “turning stories into numbers – to translate subjective experience into something collectively measurable.” (p. 749) Thus, the key technology for assembling populations for ASD research has been the ADOS and the ADI-R, which attempts to produce stable illness collectives defined by measurable symptoms. The everyday stories of families and individuals with ASD are lost in the process.

The quantifiable measures are collected and stored in computerized databases. The AGRE and the SSC have both generated their own computer network systems to allow easy access by scientists to phenotype and genotype information, as well as re-distribute raw genotype data back into the system. These databases are constantly updated, and serve as sites of continual movement of information. For example, the AGRE developed the network system of ISAAC – the Internet System for Assessing Autistic Children. Several of its key features highlight the fluid and interactive nature of the database, such as being able to: collect and manage data from different sites; share data with funders, review boards or other scientists; and import data to any other compatible data application or analysis tool.<sup>194</sup> These databases also serve as sites where information related to the phenotype data and biological materials are de-identified (i.e., recorded in such a manner that subjects cannot be identified, directly or through identifiers linked to the subjects). Thus, in many regards, the development of these

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<sup>194</sup> Internet system for accessing autistic children (ISAAC). Retrieved June 2, 2010, from <http://www.autismtools.org/index.cfm>.



databases bring together many different social worlds and serve as a second level of transcription/translation, where families are given de-identification numbers and compiled based on quantifiable phenotypic and genotypic characteristics.

### **A Family Portrait**

To guide readers through the processes of social and scientific transcriptions and translations of DNA, I provide visual representations of how families, their blood, and clinical information are transformed in throughout this process. See Figure 6.1. The first set of images is of families represented on the Autism Speaks and Simons Foundation websites, used to recruit families for the AGRE and SSC, respectively.<sup>195,196</sup> One such pamphlet at the 2009 IMFAR conference emphasized “finding a cure for autism” and “how families can *help* scientists” by participating in AGRE (Autism-Speaks, 2008a) (my emphasis). Recruiting materials for the Simons Simplex Collection (SSC) also highlight family pictures, emphasizing “strength in numbers” and how families connected by autism share a common bond – they all want to know the causes, treatments, and what the future holds for their children. Both sets of recruiting materials emphasize how participation in the study would *help* their families; including free diagnostic evaluations or access to staff that are experts in autism.

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<sup>195</sup> Autism Genetic Resource Exchange: Family contact form. Retrieved June 1, 2010, from <https://www.autismtools.org/familyagre/register/>.

<sup>196</sup> Simons Foundation Autism Research Initiative (SFARI) Simplex Collection: Get involved. Retrieved June 1, 2010, from <https://sfari.org/ssc-get-involved>.



**Figure 6.1.** Visual representations of families with ASD that participate in AGRE and SSC

These pictures are symbolic of the centrality of families to the production of genetic knowledge. The AGRE newsletter also provided vignettes of families who participated in the research to further reiterate their commitment to families throughout the collection/post collection process. Although the SSC initially promised in one of their earlier brochures to “offer ongoing support as well as access to the latest research results”, a major complaint I received from parents who participated in the SSC concerned the lack of follow-up. In chapter four, I discuss many of the issues parents had about the “results” of the SSC such as, the time it took to get the written diagnostic evaluation, and the difficulty of interpreting the results. There was also confusion among

several parents about whether genetic testing was conducted and whether they were going to get genetic information back.

### Production of Family Pedigrees

Once families donate their blood, and scientists document clinical information on their child with ASD, the data is transformed in several additional ways. First a family pedigree (family tree) is created to establish a visual representation of family relationships and a medical history of autism. See Figure 6.2.

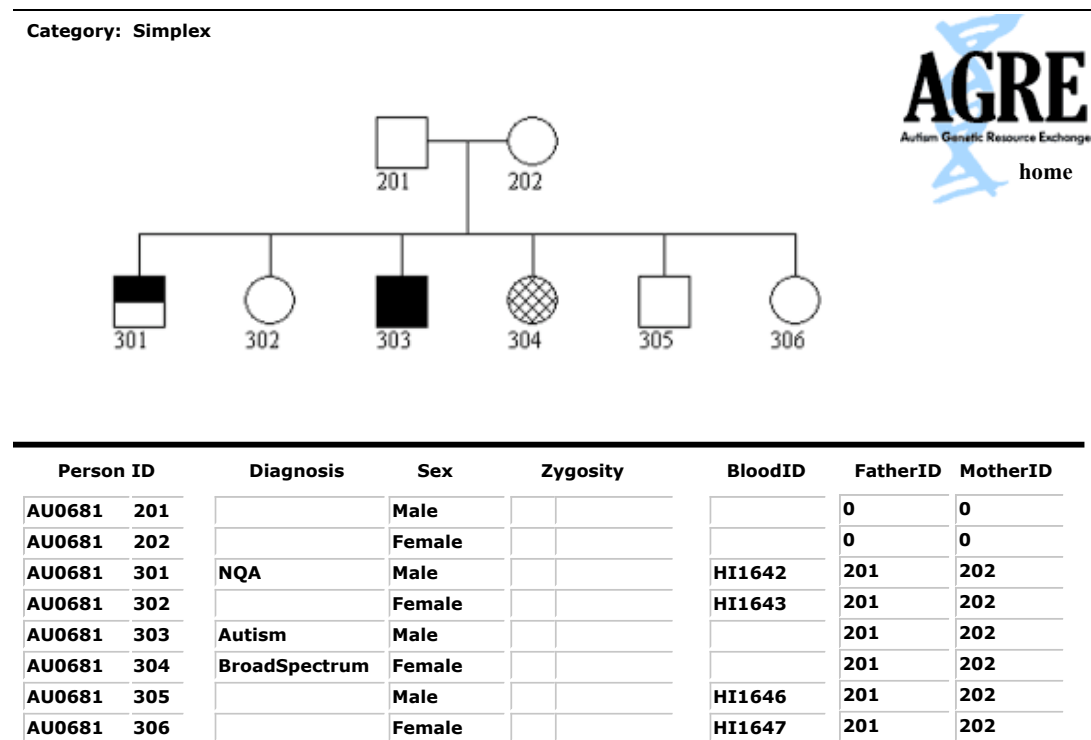


Figure 6.2. Pedigree of a simplex family in the AGRE collection

This figure represents two generations of a “simplex” family currently in the AGRE database.<sup>197</sup> As you can see, each member of the family is given a “Person ID”, “Blood ID”, and for the children a “Father ID” and “Mother ID”. Females are represented as circles, and males are represented as squares. It is a prerequisite for both AGRE and SSC that the parents are the biological parents and that none of the children are adopted. This is a prerequisite for both AGRE and SSC. Thus, medical pedigrees generated from these projects are limited to family associations based strictly on genetic relationships, thereby formalizing genetic bounds and excluding social relationships. Yoshio Nukaga (2002, p.40) describes medical pedigrees as “visual tools used to translate family problems into visual inscriptions in order to understand genetic nature of a given disease.” (p. 40) He argues that the introduction of new technologies in combination with medical pedigrees has made the development of the new genetics possible. Furthermore, the uncertainty and complexity of early forms of medical pedigrees lead to the standardization of pedigree styles that are used as a form of scientific evidence. Thus, as Atkinson, Parsons and Featherstone (2001)<sup>198</sup> point out, “the format and conventions of a pedigree are so well known that it can readily take on the appearance of a neutral or transparent medium.” (p. 10)

I use this particular family pedigree because it shows how the “autism spectrum” is transformed into information scientists can utilize in their research. One scientist explained to me that, without the phenotype information including the pedigrees, the

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<sup>197</sup> AGRE pedigrees are publically available through the 2010 AGRE pedigree catalog. See AGRE catalog at <http://www.agre.org/agrecatalog/agrecatalog.cfm?do=cat>.

<sup>198</sup> Atkinson et al. (2001), explore the practical reasoning that informs the professional construction of family and kinship on the part of the clinical geneticists and genetic scientists. They argue “the ‘natural’ facts of biological relations and the ‘social’ facts of kinship are brought together through the everyday work and talk of such specialists.” (p. 21).

genotype data is meaningless. This statement reflects Nukaga's assertion that the combination of standardized pedigrees allows clinicians and researchers to go a step further in their investigation of hereditary disease (Nukaga, 2002, p. 47).<sup>199</sup> Furthermore, it reinforces the assertion by Atkinson and colleagues (2001) that "DNA samples are drawn into the idiom of family and kinship [where] they are rendered 'social' as well as 'natural' entities as they are infused with the discourse of family and kinship." (p. 21) This information is a critical tool for producing genetic knowledge. In fact the Simons Foundation refers to the Simons Simplex Collection as a tool used by researchers.

This pedigree also represents the three different "affected status categories" provided in the AGRE database: "autism", "not quite autism", and "broad spectrum". The "affected status" categories are based on the ADI-R domain scores, created to facilitate analysis by researchers who may not be comfortable interpreting the ADI-R data to formulate their own diagnoses.<sup>200</sup> In this family there are three children who are given an "affected status". Going from left to right, the first male (#301) is given the label "Not Quite Autism" (NQA), indicated by the square that is half black and white. The shading

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<sup>199</sup> Several social scientists have investigated the importance of medical pedigrees as investigative tools. For examples, see (Armstrong, et al., 1998; Atkinson, Parsons, & Featherstone, 2001; Nukaga, 2002; Richards, 1996).

<sup>200</sup> Autism is identified using the well-validated ADI-R scoring algorithm (Lord, et al., 1994). NQA (Not Quite Autism) represents individuals who are no more than one point away from meeting autism criteria on any or all of the 3 "content" domains (i.e., social, communication, and/or behavior), and meet criteria on the "age of onset" domain; or, individuals who meet criteria on all 3 "content" domains, but do not meet criteria on the "age of onset" domain. Broad Spectrum defines individuals who show patterns of impairment along the spectrum of pervasive developmental disorders. This is a broad diagnostic category that encompasses individuals ranging from mildly- to severely-impaired. This category potentially includes such pervasive developmental disorders as PDD-NOS and Asperger syndrome, which are used in many genome scans; however, this classification is not based on any validated algorithms. For more information about the "affected status" categories see AGRE website at <http://agre.org/agrecatalog/algorithm.cfm>.

of black in this case represents characteristics of autism, but since half of the box is white, this child does not “quite” meet all the diagnostic criteria for a label of “autism”. The second male (#303) is the one family member who really meets the criteria for a diagnosis of autism. This family member is represented with a solid black box to indicate the diagnosis of autism using the ADI-R diagnostic tool. The third child, (#304) is a female labeled as “Broad Spectrum”, displayed as crossed lines within the circle. The crossed lines indicate that this child displays features of autism but is likely to be “high functioning” and could possibly be classified under the Asperger’s label or Pervasive Developmental Disorder, Not Otherwise Specified (PDD-NOS). Although the family members assigned “affected status” classifications of “NQA” and “Broad Spectrum” do not meet the diagnostic criteria for “autism”, they are given “affected status” labels to help researchers easily identify family members on the spectrum who, according to AGRE, “show potential value for inclusion in genetic or behavioral studies of autism.”<sup>201</sup> Thus, in many regards, the production of the pedigree expands the diagnostic boundaries to help scientists identify heritable genes involved in autism, while at the same time restricts the constitution of families because the pedigree is based on genetic relationships alone.

Although the pedigrees in the AGRE and SSC are not representative of detailed medical pedigrees typically taken by genetic counselors or medical geneticists,<sup>202</sup> they do

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<sup>201</sup> NQA and Broad Spectrum are not DSM-IV diagnoses, but rather, diagnostic classifications that would require further clinical analysis in order to establish a diagnosis based on DSM-IV criteria. See AGRE website at <http://agre.org/agrecatalog/algorithm.cfm>.

<sup>202</sup> For example, medical pedigrees obtained from genetic counselors or medical geneticists usually include at least three generations. Many symbols are also missing from this example that are typically used in creating a medical pedigree. Furthermore, a major situating element of the pedigree is an arrow pointed to the “proband”, which indicates who has attended the genetic

represent a heterogeneous system of boundary objects as described by Star and Griesemer (1989). Simultaneously, these pedigrees and their associated phenotypic data represent a system of *repositories*, which are indexed in standardized form. Researchers from different scientific disciplines can use this data for their own purposes as long as their work falls within the bounds of legitimate autism research. The pedigrees represent an *ideal type* because the pedigree and phenotypic information is divorced from any local contingencies of the social and cultural aspects of the family. This allows it to be adaptable to various scientific questions. The scientists I interviewed were approaching research on the genetics of autism from many different disciplines (i.e., genetics, neurology, psychology, epidemiology) and applied different genetic approaches (i.e., genome-wide association studies, candidate gene approach, sub-classification of phenotypes).

The pedigree and associated phenotypic data also represents *standardized forms*, which serve as objects that can be used to communicate across a diverse set of scientific expertise. For example, among the scientists I interviewed who utilized the AGRE database, several were not directly using the data for genetics research. Rather, they were “combing the phenotypic data” to pick out patterns of characteristics or traits that could be subdivided for use in epidemiological studies. In this way, the pedigree is reconfigured by the discipline utilizing it. The knowledge, skills and expertise of different scientific disciplines influence the utilization of the pedigree (Atkinson, et al., 2001) but also

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clinic and is the primary source of information about the family. Typically, this is an affected member of the family, but in the case of autism, it is usually an unaffected parent. Thus, the pedigrees used for the AGRE and SSC are somewhat limited compared to medical pedigrees that are used for linkage analysis. See Richards (1996) for examples of extended pedigrees used in clinical genetics (p. 256-257).

reconstitute it. Hence, the pedigree serves as a classic boundary object of shared discourse among various scientific disciplines (Star & Griesemer, 1989).

Thus, the AGRE database and SSC are types of standardized indexed boundary objects that move fluidly to researchers throughout the world and meet the different needs of different researchers through their means of adaptation. This mutability directly challenges what Bruno Latour (1987) refers to “immutable or combinable mobiles”. The researchers who utilize the database can combine the data any way they see fit, thereby further altering, sub-dividing, and eliminating families at will. Thus, as Star and Griesemer (1989) point out, the “local uncertainties are deleted” in the creation of mutable boundary objects (p. 411). In the case of autism, the uncertainties and complexities of distinguishing familial, social and diagnostic boundaries are lost in the transcription/translation process of generating databases composed of pedigrees, DNA, and clinical information.

### **Immortalization of Families**

The social, living and biological representations of families with ASD takes on another level of transformation through the immortalization of blood donated by families to the AGRE and SSC. Once the family’s blood is drawn, it is shipped to Rutgers University Cell and DNA Repository (RUCDR), the largest university based repository in the world, located on the Busch Campus of Rutgers University, Piscataway, New Jersey. RUCDR, established in 1998, develops, stores, and distributes cell and DNA repositories



through grants and contracts with the government and private agencies.<sup>203</sup> RUCDR currently has over 150,000 cell lines in their repository, including 8,504 cell lines for the AGRE database and 5,370 for the SSC. This is the site where blood is physically transformed into immortalized lymphoblastoid cell lines (LCLs), then grown to large-scale production for DNA/RNA extraction or cryopreservation. The cost for scientists to order samples from RUCDR are \$1,650 per 96-well plate of DNA (96 samples), individual cell lines are up to \$75, and 200 µl (microliter) vial of plasma is \$16.<sup>204</sup> The majority of scientists I interviewed ordered DNA received in 96-well plates along with a corresponding sample number for each well that represented one individual in the AGRE database.

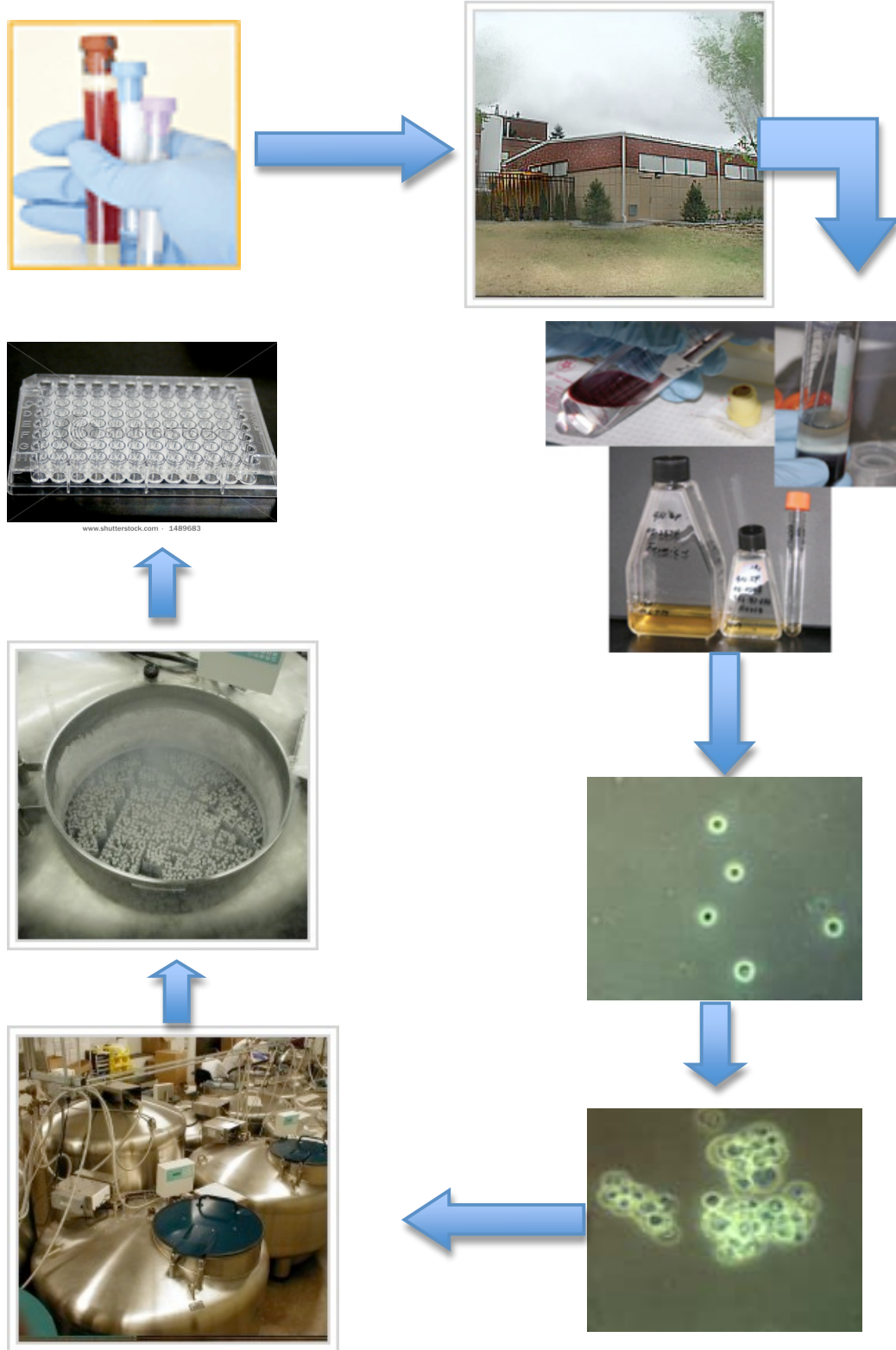
Figure 6.3 is a visual representation of the process of immortalizing cell lines derived from blood donated by families. It demonstrates the movement and transformation of materials that occurs during the process of large scale DNA production. As the images imply, after the blood is shipped to RUCDR, lymphoblastoid cell lines (LCLs) are generated by Epstein Barr Virus (EBV) transformation of isolated lymphocytes. Essentially this involves separating the blood into different components and then infecting it with a foreign body (EBV), which immortalizes the cells. This process takes about six to eight weeks to generate large amounts of cells that can be indefinitely grown as a continuing source of genetic material. This image demonstrates the growth of single infected cells into large-scale production of cell cultures that are then frozen in liquid nitrogen tanks. These cell lines can then be transformed into DNA, which further

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<sup>203</sup> Rutgers Cell and DNA Repository. Retrieved May 30, 2010, from <http://www.rucdr.org/index.html>

<sup>204</sup> Simons Foundation Autism Research Initiative (SFARI): Rutgers University cell & DNA repository biospecimen prices Retrieved May 30, 2010, from <https://sfari.org/biospecimen-prices>.

eliminates all the intracellular components, leaving only the DNA. The 96 well plates is the physical representation of the families the scientists receive when they are ready to conduct their research. Each well represents one member of the family available in the AGRE database. Thus, the process of immortalizing human blood requires many human actors (i.e., families, phlebotomists, Fed-Ex couriers, laboratory directors, laboratory technicians) and many non-human actors (i.e., blood containers, shipping materials, Epstein Barr Virus, fetal bovine serum, centrifuges, bioreactors, liquid nitrogen tanks, and all the necessary biotechnological tools and technologies needed to process the blood such as pipettes, chemicals, computers, and protocols). The end result is a 96-well plate of micro grams of DNA available throughout the world to conduct autism genetics research. Laboratory scientists rarely meet families and individuals who participate in either the SSC or the AGRE. As Atkinson and colleagues point out, the scientists' "discourse is informed by the specimens and samples that are available for testing and by the pedigree of biological relatedness. The discourse is constructed primarily in the domain of biomedicine." (p. 21)



**Figure 6.3.** Immortalization of family blood into lymphoblastoid cell lines

## **Fluorescence and Digitization of Families**

In many regards, the transformations of DNA within each laboratory that utilizes AGRE and SSC samples are points of alternative expansion from previous steps. Here, the social, cultural, and biological representations of families with ASD are reduced to what appear to be invisible specks of DNA on a 96-well plate, and a database that provides de-identified pedigrees and information regarding the family's affected status, diagnostic evaluations, and medical histories. In the genetic laboratory, new tools and technologies are brought to bear in order to create new meanings associated with the pedigree, DNA, and phenotypic characteristics. In essence, scientists who utilize these samples for their research are re-constructing these different elements of family data to generate scientific meanings of ASD. As I argue and illustrate in chapter three, the large volumes of genomic data that have been generated through whole genome microarray analysis, coupled with new bioinformatic systems software, are beginning to “imagine” complex and interconnected networks of genetic mechanisms involved in autism. Edward Hall (2005) refers to this phase as “digitization”, where extracted biological material is transformed into digital data (p. 2670).

The translations produced in the laboratory take on many different forms such as peer-reviewed scientific reports, media releases, and professional conference presentations. Visual representations of different scientific transformations of the DNA and associated pedigrees and phenotypic information are provided in Figures 6.4 and 6.5. These images are from a highly publicized study that utilized the AGRE database to

identify the copy number variants (CNVs): 16p11.2 deletions and duplications (Weiss, et al., 2008).<sup>205</sup>

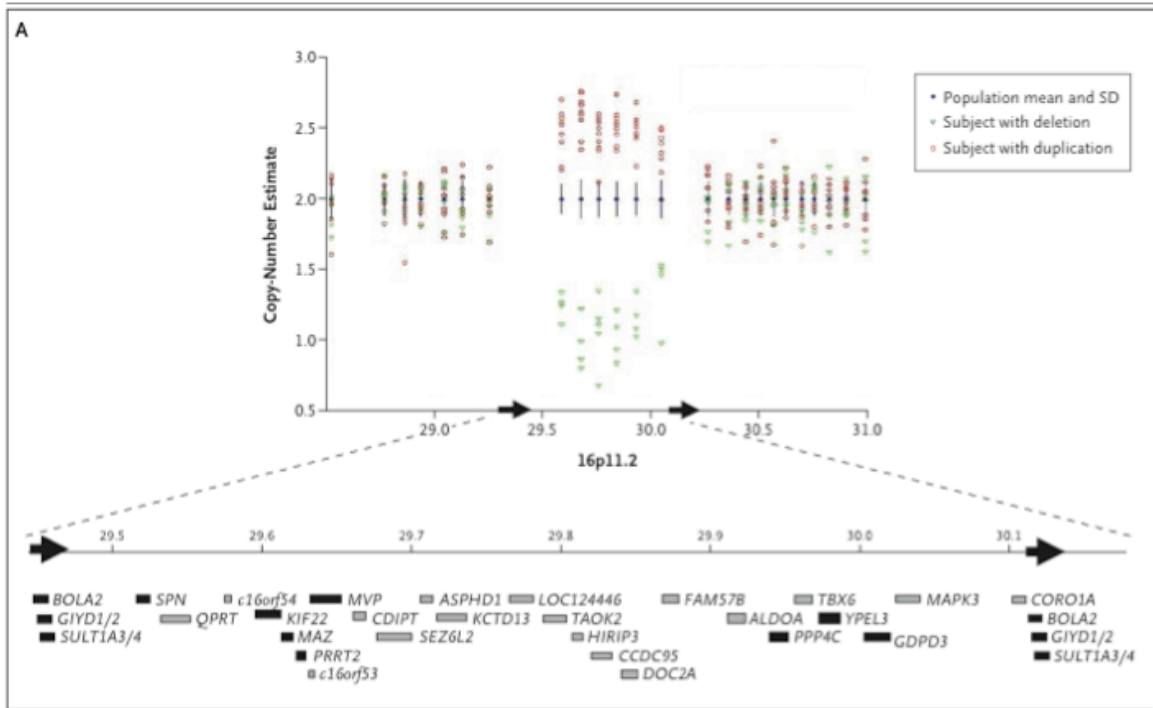
The authors describe figure 6.4 as “normalized intensity data” generated using a program called Birdseye<sup>206</sup> across a 2 megabase (Mb) region of chromosome 16. The deletions are denoted in green triangles (lower scatter) and represent “five children (four boys and one girl) with autism in four independent families” (Weiss, et al., 2008, p. 669). The duplications are denoted by red open circles (upper scatter) and represent “three AGRE families”. They were “transmitted from [one] parent to two of two affected offspring (male and female), as well as to one unaffected daughter and from another parent to four of four affected sons.” (Weiss, et al., 2008, p. 670) Below the intensity data are annotated genes in the 16p11.2 region, with grey denoting genes expressed in the brain and black denoting unknown or little gene expression in the brain.

An alternative representation that the authors used to confirm these findings is shown in Figure 6.5, an analysis using Fluorescence in Situ Hybridization (FISH). This figure shows FISH for a control population in panel A, with arrows pointing to “normal” fluorescence on chromosome 16. Panel B represents a sample with a 16p11.2 deletion where a fluorescent signal is seen on one chromosome 16 (arrow) and absent in on the other chromosome 16 (arrowhead). Panels C and D represent a sample with a 16p11.2 duplication where fluorescent signals appear to be stronger on one chromosome 16 (arrows) than on the other chromosome 16 (arrowhead).

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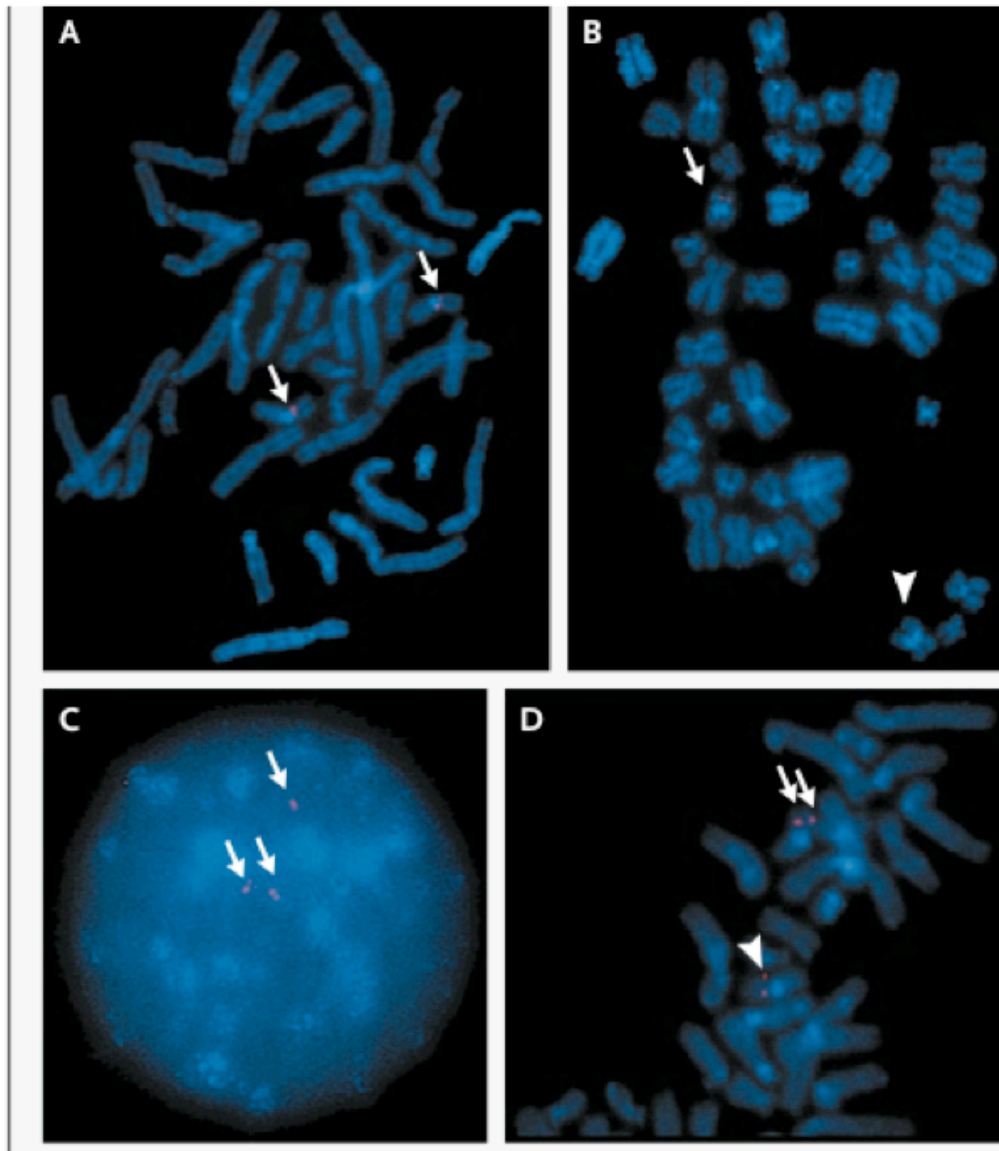
<sup>205</sup> See chapter three for a discussion of the 16p11.2 CNV deletion.

<sup>206</sup> Birdseye is computer software used to find regions of variable copy number in a sample. The hidden state is the true copy number of the individual’s genome; the observed states are the normalized intensity measurements of each probe on the array (Korn, et al., 2008).



**Figure 6.4.** Scientific representations of children with autism

The language used by the authors to describe these two figures further demonstrates the reconfiguration and disappearance of the family through the process of technoscientific inquiries. In the first figure, families are described on the basis of their biological relationship to one another to prove the *de novo* hypothesis for the 16p11.2 deletion. Here the authors state, “five children (four boys and one girl) with autism in four independent families carried the *de novo* deletions; we observed no deletions in the parents” (Weiss et al., 2008, p. 669). This statement indicates that the deletion was not inherited. Similarly, for the 16p11.2 duplications, the biological relationships in families are used to argue in favor of the inheritance of the duplication, which the authors describe as “transmitted from a parent to...affected offspring” (Weiss, et al., 2008, p. 670).



**Figure 6.5.** Scientific representation of chromosomal deletion and duplication of 16p11.2

Thus, the biological structure of the family is needed to make an argument for the results of the study, even though on the surface it appears contradictory. The family however disappears all together in the description of Figure 6.5. In this case, the fluorescent chromosomes are representative of “a sample” with a deletion or duplication of

chromosome 16p11.2. Hence, the family is no longer present in the description of the data, losing all forms of narrative presence.

### **Families Lost in Translation**

I started this chapter with a description of the everyday lives of families with ASD who participated in either the SSC or the AGRE database. Having the opportunity to interview these families in their homes, away from any medical or educational setting, highlighted for me the struggles, joys, and everyday lives families experienced. These families described to me the major issues their children faced such as trouble with social interaction, problems with language and speech, sensory issues, rigid and literal behaviors, and intensely focused interests on particular things or subjects. Many of these symptoms are part of the standardized diagnostic tools used for the AGRE and SSC, and thus were translated into quantifiable traits that could be categorized and counted during the process of collecting data for these projects. I demonstrate how the ADI-R and ADOS came to be diagnostic standards through the demands of the research community, rather than the clinical community. Thus, the first form of transcription/translation is the transformation of parent interviews and child observations into a quantifiable and standardized database that scientists can easily access to conduct their research. However, this process occurs within a larger social arena of autism genetics where the social worlds of funding agencies (i.e., NIH), large consortia (i.e., AGP) and family genotype and phenotype collections (i.e., AGRE) come together.

A second form of transcription/translation occurred during the creation of the family pedigree, which was based strictly on biological relationships. Thus, medical



pedigrees generated for AGRE or SSC are limited to family associations based strictly on genetic relationships, thereby formalizing genetic bounds and excluding social relationships. Thus, social relationships are excluded from scientific concerns, while at the same time diagnostic boundaries maybe expanded by labeling family members as “Not Quite Autism” or “Broad Spectrum”. I argue that the standardization of diagnostic tools used for both the AGRE and the SSC are eliminating much of the lived experiences of parents (and ASD adults), as well as symptoms not officially recognized by the DSM – IV. Thus, the AGRE and SSC are standardized indexed boundary objects (Star & Greisemer, 1989) that move fluidly to researchers throughout the world and convey information reconfigured by their users for their own purposes. Here, the uncertainties and complexities of distinguishing familial, social and diagnostic boundaries are lost in transcription/translation.

The process of immortalizing family blood cells is a third site of transcription/translation. Here the blood of families is physically transformed into cell lines that can make DNA indefinitely for future genetics research. These cell lines are either cryo-preserved for future use or reduced to isolated DNA. This process also highlights the social worlds that come together such as RUDCR, the laboratory director, technicians, funding agencies, and even Fed-Ex Couriers. It also sheds light into the many different non-human actors involved in developing genetic databases, such as the Epstein Barr Virus, bioreactors, and liquid nitrogen tanks. The end result is a bank of immortalized lymphoblastoid cells, which can be converted into DNA and shipped to researchers around the world in a transparent 96 well plate.

A fourth site of transcription/translation occurs within the scientific domain, during the process of scientific inquiry using various genetic technologies such as fluorescent intensities generated with microarrays or FISH analysis. Here scientists utilize family samples for their research and in essence reconstructing different elements of family data to generate scientific meanings of ASD. I argue that scientists use the family's biological relationships in order to support their research results, but family relations are literally deleted from the discourse once individual chromosomal characteristics are analyzed. In essence, scientists who utilize the AGRE or SSC for their research are re-constructing genotypic and phenotypic characteristics of the family data to generate scientific meanings of ASD.

Thus, by literally "following the DNA", I demonstrate how the social, cultural and biological representations of families get completely distorted, transformed, deleted, and re-constructed through the process of scientification. The components of each family member are moved from one site to another to enable these transformations to take place (i.e., phenotypic database, RUCDR, individual laboratory), thereby fragmenting the family into components that science deems essential in the pursuit of genetic knowledge. However, these family components (i.e., pedigree, phenotypic information, DNA) are reconstructed in the laboratory to generate new genetic knowledge of autism spectrum disorders. Throughout this process many people, places, and institutions come together, who are associated with non-human objects such as the databases networks (i.e., ISAAC), family pedigrees, diagnostic tools, immortalized lymphoblastoid cell lines, DNA, and all the tools and technologies associated with genetic knowledge production (e.g., sequencing, microarray, FISH, pipettes, etc.).

## CHAPTER 6: A GLIMPSE OF AUTISM PERSPECTIVES

The ability of genetic information to define current notions of individual and group identity has expanded within our society, as more and more genetic information continues to be identified and used for diagnosis and prediction of diseases. The sequence of the human genome and its recent applications in medicine has created potential sites where identities might be transformed based on information literally from inside bodies. As one scholar notes, genetic information has the qualities of being “inborn, natural, and unalterable”, which challenges other claims to authentic identity and group membership traditionally based on family histories, written documentation, cultural practices and inner convictions (Brodwin, 2002). Social scientists have begun to investigate various discourses on the influences of genetic knowledge, including analyses of how it shapes disease classification and individual and collective identity practices (Hedgecoe, 2003; Kerr, 2000, 2005; Novas & Rose, 2000; Rabinow & Rose, 2006; Rose, 2007; Taussig, Rapp, & Heath, 2003); how individuals view themselves in relation to others (Elliott, 2002; Finkler, Skrzynia, & Evans, 2003; Lock, 2008; Lock, et al., 2006); and how it mobilizes disease advocates’ involvement in the production of genetic knowledge (Novas, 2006; Rabeharisoa, 2006; Stockdale & Terry, 2002; A. Wexler, 1996). Within these discourses is an underlying, polarized and reified debate between nature and nurture - whether genetic information, social and/or “natural” environments, or a combination thereof co-constitutes humans.

Very limited research has focused on the perceptions and experiences of those who are on the autism spectrum. Rarely are examples found in the literature that help

scientist understand the heterogeneity within ASD and the life experiences that *may* contribute to how people with ASD come to understand this disorder. Furthermore, studies of autism perspectives have largely been focused on children and not necessarily adults. ASD adult experiences of the social world may be very different. The practices involved in constructing ASD as either a childhood disorder, a psychological disorder, a neurological disorder, a genetic disorder, or a combination there of are at issue.

The purpose of this chapter is to explore notions of genetic identity regarding autism spectrum disorders (ASDs). ASDs are complex conditions that are currently defined and medicalized as mental health disorders with neurologic and genetic bases. The intent of this analysis is to better understand how adults diagnosed or self-identified on the autism spectrum view the genetics of autism, and how their understandings are reflected in their everyday lives. I seek to map out the transformations of identities engendered through “lay knowledges” of the genetics of autism. Based on analysis of 18 interviews with adults on the autism spectrum, this chapter explores different forms of subjectivity that exist among adults either diagnosed or self-identified with high functioning autism or Asperger syndrome. I consider how these individuals negotiate scientific notions that autism is a genetic disorder, as well as their awareness and attitude about autism genetics research and the implications of genetic testing.

### **Insights into the World Autism**

I use the term “way of being” rather than “disorder” because I wonder whether the autism spectrum should be considered as “another order” of being as opposed to a disordered, deviant way of existing (Shore, 2003, p.v).

Although there has been a modest amount of qualitative research on autism, it has generally focused on the experiences of parents and their children. Over the course of the last 25 years, the available perspectives on living with autism have been autobiographical accounts by adults on the autism spectrum voicing their opinions, experiences and, often, resistance to biomedical research on autism (Grandin, 1986, 1995; Lawson, 1998; Shore, 2003; Willey, 1999; Williams, 1992, 1994). These narratives demonstrate that individuals with autism are capable of communication and able to share insights into their way of thinking. These works contradict claims that individuals with autism have limited narrative capabilities (Bruner & Feldman, 1993; Capps & Sigman, 1996). For example, Temple Grandin shares her personal journey with autism, providing a glimpse of how she constructs the world and lives her life in unimaginably different ways. In her book *Thinking in Pictures*, she provides a personal account of what it was like for her as a child, describing her issues with sensory overload of smell, sound and touch, her spontaneous epileptic tantrums, her endless rocking, and disconnection with others (Grandin, 1995). Wendy Lawson (1998) and Liane Holliday Willey (1999) offer accounts of isolation, and often depression, as individuals not properly diagnosed. Both women were not diagnosed until they were adults. Like many adults on the spectrum, Willey did not discover that she was on the autism spectrum until her daughter was diagnosed as having Asperger syndrome (Willey, 1999).

Studies of ASD adults have found that many autistic adults are proud to have autism and do not desire to be “cured” (Chamak, 2008; Orsini, 2009; Silverman, 2008a). Brigitte Chamak (2008) describes how the Autism Network International (ANI), considered the first and largest autistic organization run by autistics, has made a political

issue of autism by redefining it as a different way of being and not a disease. The aims of the ANI are not to cure or treat autism, which are both promoted by some parent advocacy groups, but rather to organize conferences to enhance autism self-advocacy, to promote a new awareness and reduce stigmatization of autism through public visibility of their actions, to meet other individuals on the autism spectrum, and to educate potential allies within the non-autistic community (Chamak, 2008).

Michael Orsini draws on the notion of “biological citizenship” to reflect on the important challenges raised by autistic citizens wanting to speak for themselves and represent autism based on lived experiences (Orsini, 2009). He argues that autistics are “using the Internet or other fora to counter what they see as avalanche of advocacy in the name of, but not for, autistic children” (Orsini, 2009, p.1983). He discusses the contradictions of biological citizenship where, on the one hand, it can be empowering and affirming to share “neurological distinctiveness” and to build networks of support (Orsini, 2009, p.184). On the other hand, Orsini draws on the work of Majia Nadesan (2005, p. 208) who argues that by suggesting biological differences of individuals with autism, whether genetic or neurological, “is both divisive and affirmative in its representation of autistic difference.” This contradiction lies in the efforts by advocates/activists to embrace and reclaim the autistic label while at the same time “advocating for understanding of and special care for the more troubling autistic deficits or symptoms.” (Nadesan, 2004, p. 209)

Chloe Silverman also considers the contradictions of biosociality by identifying two very different discourses of kinship in the world of autism research based on autistic behaviors (Silverman, 2008). One set of discourses is based on familial tendencies and

affinities built on assumptions of genetic association and physiological likeness (Silverman, 2008, p. 39). In this case, parent advocacy groups have relied on the existence of autistic traits within families to establish genetic research programs, as “the means to repair broken families” (Silverman, 2008, p.43). A second set of discourses is based on likeness across groups of people with autism. In this case, the “autistic biosociality” of self-advocacy groups views the desire for a “cure” as unethical in the sense that it denies “autistic humanity” (Silverman, 2008, p.47). Priorities set forth by these groups are more devoted to diagnosis issues, as well as specific questions of rights, employment, treatment and services. The ideals of these two different biosocial groups contradict one another, as one group accepts “neurological diversity” while the other develops programs based on a genetic causation model to eradicate “neurological disability” (Silverman, 2008, p.50).

Silverman’s work (2008) is the only study to date that specifically addresses how genetic knowledge of autism influences the identity of individuals on the autism spectrum and their opinions of this type of research. What is unique about the sample in current study reported here, is that it includes interviews with four families where both the parent who self-identifies with having autism and the diagnosed child (in this case a young adult) were interviewed, thus offering a multigenerational perspective. This sample also represents individuals on the spectrum who are not currently engaged in activism for autistic individuals. Rather, they represent snapshots of experiences of the everyday lives of autism. The influence of genetic knowledge on the identity of autism and how it is described within everyday lives may represent a very different form of subjectivity than described by Chamak (2008), Orsini (2009), and Silverman (2008).

## **On Reflexivity**

This study utilized and incorporated the theory/methods package of symbolic interactionism and grounded theory methods (Clarke, 2005; Strauss, 1987).<sup>207</sup> As with many methodologies, reflexivity on the part of the researcher is an important element in the process of interpreting qualitative data. I have been told from conversations with specialists who work with autistic individuals that being able to truly understand the “essence” of what it is like to live in the world of autism is impossible due to their “different kind of mind.” As such, this posed challenges in my ability to truly be reflexive and reflective during the analysis. The following interpretations are based on the interviews and observations of one researcher and have been reviewed by several of the participants for accuracy of interpretation.

## **Shadows of Autism**

Awareness of research on the genetic etiology of autism spectrum disorders (ASD) was largely non-existent in this sample. What little participants knew about autism genetics was gathered from random articles in popular magazines and “Google” searches on the Internet. Regardless of their knowledge about of autism genetics research, the participants overwhelmingly favored the idea that ASD had a genetic component. Some viewed ASD as exclusively based on genetics, while others considered environmental causes to also be involved.

Their understandings that autism had a genetic component were largely based on personal experiences within the family. These experiences were based on descriptions of

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<sup>207</sup> See chapter one for a detailed description of this theory/methods package.



relatedness to other family members through the use of certain traits associated with ASD, and were not based on genetic information per se. Instead, these “shadows of autism” were represented through similar characteristics of the participants which they recognized in other family members as well, such as problems with social skills, language delays, sleeping problems, sensory issues, as well as aptitude for skills such as computer programming or the ability to decipher complex systems. To the participants, these characteristics essentially represented heritable traits associated with ASD shared among family members. For example, one participant described her family as follows:

I think it's genetic because I know my Dad has it and I can see it in my two older half brothers from my Dad's previous marriage.... And then there is my Dad's siblings.<sup>208</sup>

This participant has a twin sister also diagnosed with Asperger syndrome and goes on to say how her uncle, along with everyone on her Dad's side of the family, is awkward in social situations. She also described how her aunt lives alone and away from the entire family, never makes eye contact, and has a lack of awareness of how she appears to others.

Multi-generational descriptions such as these appeared throughout most of the interviews, where family members were described as “*being on the spectrum*”, “*aspie-like*”, “*Asperger esque*”, and “*non-neurotypical*”. All but one of the participants mentioned that ASD traits, characteristics, or the diagnosis itself existed in one or more of their immediate family members (e.g. children, siblings, parents). Not everyone used the language of “genetics” but rather implied an inherited nature of ASD by referring to

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<sup>208</sup> ASD Adult Interview #13 - Childhood diagnosis of Asperger syndrome

family members and specific traits. For example, one participant talked about how there was a “*legacy of late talkers*” among the men in his family, including his father, himself and his son, whom he considered to all be “*socially backwards.*” Similarly, one participant described his sleeping disorder as an Asperger’s symptom that ran in the paternal line of his family. Another participant identified similar traits in his family members that were characteristic of ASD, such as lack of sociability, depression, and the inability to ask for help when help was needed.

One participant viewed Asperger’s as part of his entire family, where his mother and father both demonstrated certain sensibilities of ASD. He described his family in the following way:

We always said that if you put my mother together with my father and made them the same person, that person might have Asperger’s disorder... they both have different and opposing symptoms and are perfectly fine with the other.<sup>209</sup> (Participant 10, childhood diagnosis of Asperger syndrome)

This participant goes on to say that Asperger syndrome is part of his family, even if he was the only one who had an official diagnosis. He stated, “*our family is the way it is and if Asperger’s disorder wasn’t in the dictionary then it wouldn’t be in our family.*”

The point made here is particularly poignant with regard to the medicalization of disease (Conrad, 1992, 2000b), and how diseases are defined, categorized, and then taken up as a form of identity within families. For this particular family, the medicalization of Asperger syndrome, which officially became part of the ASD diagnosis in 1994 (APA, 1994), was viewed positively. For example, the father of this participant was particularly

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<sup>209</sup> ASD Adult Interview #16 - Childhood diagnosis of Asperger syndrome

proud of his capabilities of deciphering complex systems and viewed his autistic characteristics as what made him valued as a computer engineer. Another family referred to themselves as “*Aspies*”, which represents people with high functioning autism or Asperger syndrome that view autism as a ‘neurological difference’, not an illness or disability (Bagatell, 2007). I highlight these particular examples to demonstrate that not all of the ASD characteristics described by these participants were framed in the context of a deficit model, which some autism advocates argue is how the DSM-IVR currently frames ASD (Carley, 2008). The meaning of autism beyond the disorder in these cases demonstrates how the characteristics conceptualized as pathology in the context of the diagnosis can be completely adaptive in everyday life (O'Neil, 2008).

### **Self-Identification with ASD**

Interpretations of the genetics of autism also influenced ASD identity formation in parents in this study who had a child diagnosed with an ASD. These parents reconstituted their identities and self-diagnosed as on the autism spectrum based on their experiences and perceptions of their autistic children. Five participants had self-identified with ASD through their child’s diagnosis and none had previously considered ASD as part of their identity. It is not surprising that this age cohort (> 40 years old) did not have a diagnosis of autism or Asperger’s since the diagnosis of autistic disorder was not instituted until 1980 (APA, 1980), and Asperger’s disorder was not diagnosed until 1994 (APA, 1994). For some of the participants, the process of self-identification with ASD began when their children were first getting diagnosed with autism and their identification with the unique qualities they shared with their children. For example, one

participant remembers reading through the diagnostic criteria, acknowledging, “*well, this one is me, this one is me, this one is not me*”.<sup>210</sup> This participant specifically recognized his lack of executive functioning skills (i.e., ability to prioritize, set goals, plan and organize) and auditory sensory issues. These issues were also apparent in his two sons, one who had a diagnosis of Asperger syndrome.

Another participant recognized similarities with his son who has a diagnosis of Asperger’s, especially as his son got older. He states:

I think for me it was understanding Asperger’s through what my son was going through that help me match my own traits... As my son has gotten more to being an adult, at least chronologically, it becomes more apparent.... The comparison becomes easier.<sup>211</sup>

When this father started learning about his son’s social challenges, he recognized within himself many Asperger syndrome features he experienced when he was younger and continued to struggle with as an adult. As his son aged, he recognized similarities in his addictive qualities, bouts of depression, inappropriate behavior, and struggles to stay within the bounds of whatever group he interacts with. He sees his son struggling with many of these same challenges, and it was through this recognition that he self-identified with ASD.

Self-identification with ASD was also an evolving and life long process. For example, one participant’s experience with his son’s autism over the last 15 years triggered an evolving relationship with himself in terms of his identity. This participant believed all his life he was “*odd*” or “*different*” and described himself as “*socially*

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<sup>210</sup> ASD Adult Interview #4 – Parent of a child diagnosed with Asperger syndrome, self-identified ASD.

<sup>211</sup> ASD Adult Interview #1 – Parent of a child diagnosed with Asperger syndrome, self-identified with ASD

*awkward*". When his son was first showing signs of being a "*late talker*" like himself and his father, he expected his son to talk any day and assumed his son was part of his family legacy of late talkers. At the time his son was diagnosed with autism at about age three, he immediately viewed the differences in his son as not like himself but "*abnormal*". He stated:

I would say that the time that happened [the diagnosis] I really considered him to be no longer on the same journey that I was and it became his problem. I didn't see any connection between my oddities and him.<sup>212</sup>

He immediately divorced his "*difference*" from his sons and considered him to be on a different journey in life. From the time of his son's diagnosis until a just a few years ago (about 12 years), he did not see any connection between his oddities and his son's. This participant's father was also "*socially backwards*" until about 30 years old and thus he rationalized that he himself would also "*break out and be more socially accepted*". Only in the last couple of years has he actually recognized commonalities with his son. With the help of a family therapist who specializes in Asperger syndrome, he now realizes that he has many similarities to his son, and recognizes that the communication challenges he has experienced throughout his life are most likely because of ASD.

As demonstrated by this narrative, this participant's self-identity in relation to ASD has been an evolving processes and one with which continues to struggle. The label of ASD holds some appeal, mainly because he would like to fit into a community and feel a sense of belonging. Although he realizes there is no "*hard red line*" distinguishing people as ASD from "*normal*", he is interested in exploring how he fits in with the diagnosis mainly for the novelty of how to view himself with respect to the rest of the

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<sup>212</sup> ASD Adult Interview #5 – Parent of a child diagnosed with autism, self-identified with ASD

world. In this regard, the diagnosis of ASD may confer a collective identity for people not diagnosed by removing them from the isolation of their differences and providing them with new potential networks of support (Jutel, 2009).

### **Potential Impacts of Genetic Testing for ASD**

All the participants were asked for their opinions about genetic testing for ASD. Although there is no such approved test as yet, this hypothetical question did not seem surprising to any of the participants. My intention in asking this question was focused more on the use of a genetic test per se to diagnose ASD. However, the responses reflected participants' opinions about how a genetic test if taken today would affect their lives. Not surprisingly, some of the participants did not think a genetic test would change anything. As a father stated, who has a child diagnosed with Asperger syndrome and self-identifies with ASD:

Having a genetic test just to say that you have it is useless to me because you still have to deal with it. Knowing doesn't give you any advantage over not knowing as far as I'm concerned.... The action that is important is what you do with who you are and whether you give it a name or not is not important.<sup>213</sup>

Interestingly, the sentiments of this participant were a central theme for participants both with and without a diagnosis. For many, the availability of a genetic test would not change who they were and genetic information "*all by itself*" did not seem to be of particular personal significance.

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<sup>213</sup> ASD Adult Interview #1 – Parent of a child diagnosed with Asperger syndrome, self-identified with ASD

Several of the participants were undecided about how they viewed the use of a genetic test to diagnose ASD. One participant diagnosed with ASD in her thirties was curious, but worried that a genetic test would prove that she did not have Asperger syndrome. As she stated in her own words, *“I have this fear of being found out all the time, like someone’s going to find out I don’t really have it and I’m just making it up. It kind of scares me.”*<sup>214</sup> She worried that a genetic test would mean that her problems were *“pathological”* rather than *“a character trait that was more acceptable and that could be fixed.”* At the time of the interview, this participant was newly diagnosed with Asperger syndrome and was relinquishing much of the guilt and shame she had about the way she had socially interacted in the past and the difficulties it caused her. She described her life as constantly being misunderstood and how, since the diagnosis, she has made tremendous progress in communicating with her mother. She worried that a genetic test would take away her new ASD identity, which was helping her to forgive herself for the problems she experienced in the past.

Another participant responded that it was hard to conceive of a genetic test working when the current definition of ASD is so loosely defined. He described the current diagnosis of ASD as a *“grab bag”* of a number of different things that seem to have a sub-set of similarities. He feels that in the case of ASD, there is no *“hard red line”* that can be used to say for sure whether someone had ASD or some variation of it. Thus, a genetic test would only increase the number of *“symptoms”* associated with ASD, resulting in a higher number of ASD diagnoses. The youngest participant, who had a diagnosis of high functioning autism, demonstrated an indifferent attitude about genetic

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<sup>214</sup> ASD Adult Interview #8 – Diagnosed with Asperger syndrome at age 32.

testing, viewing his situation as “*not that serious*” and acknowledging that “*if it’s there, it’s there, if it’s offered, it’s offered. I may or may not take it.*” He did not see any value in the diagnosis, or a genetic test for that matter, since it would not change the person he was.<sup>215</sup>

For two of the fathers who self-identified with ASD and had children diagnosed on the autistic spectrum, a genetic test was viewed positively in that it could potentially help their children early on or could help alleviate specific symptoms. For example, one father felt that if they had known sooner what they now know about their son, they could have started some of the interventions earlier, such as dietary changes and behavioral therapy.<sup>216</sup> Another father described the development of a genetic test as “*worth it*” if there was a targeted medication that could alleviate exactly the symptoms of Asperger’s they wanted to erase. However, this participant also viewed medication as merely “*masking*” what you really are, which he stated is “*determined by what your genetics allows you to be.*”<sup>217</sup>

A younger participant with a diagnosis of Asperger syndrome viewed genetics as what “*defines the core of a person.*” He felt that if a gene for Asperger’s was identified it would have an important role in explaining what makes him a person. However, he did not think genes for Asperger’s should be taken out or modified in any way.<sup>218</sup> The potential for genetic technology to ‘modify’ or ‘alter’ who we are also came up in another

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<sup>215</sup> ASD Adult Interview #2 - Childhood diagnosis of autism

<sup>216</sup> ASD Adult Interview #5 – Parent of a child diagnosed with autism, self-identified with ASD

<sup>217</sup> ASD Adult Interview #1 – Parent of a child diagnosed with Asperger syndrome, self-identified with ASD

<sup>218</sup> ASD Adult Interview #9 - Childhood diagnosis of Asperger syndrome



interview. This participant was reflecting on his son who is diagnosed with ASD. He stated:

If there was a genetic cure right now for my son I don't know that I can even allow him to take it .....But if he took it he wouldn't be him. I don't think it may be worthwhile because the things that I gain may not be balanced by the things that I lose.<sup>219</sup>

This description of “*loosing*” a person if there were a genetic cure for autism raises important ethical questions about the intent of genetics research on autism. Is it to merely learn about the etiology of disease? Causal pathways? For diagnosis and treatment? Or is it intended to establish a “cure” for autism? One participant described his condition as “*not a core flaw*” and although he may have “*a genetic inclination for certain things*” he did not see the need for genetic therapy if it existed. In his opinion, “*unless there is an extremely good reason to do so...most people with ASD don't need genetic therapy.*”<sup>220</sup>

The majority of participants in this study valued their strengths, which they felt were very much part of their autism. Thus, the idea of “*erasing*” autism through genetic technologies was not a desired option.

### **Advice to Researchers**

A common theme in many interviews was that current research should focus on the everyday struggles of people with ASD instead of genetics research or other scientific pursuits in ASD. One participant stated that instead of identifying genetic deficiencies that can be “*switched off*” to remove the symptoms of ASD, “*the research should be more focused on how we live with who we are and what we do.*”<sup>206</sup> Similarly, another

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<sup>219</sup> ASD Adult Interview #1 – Parent of a child diagnosed with Asperger syndrome, self-identified with ASD

<sup>220</sup> ASD Adult Interview #9 - Childhood diagnosis of Asperger syndrome

participant expressed the need for research on the everyday lives of people with ASD. He stated:

People are improperly addressing by thinking of whether it is a question of genetics or not. I don't see why it would really make a practical difference. In terms of what actually happens if it is genetic or something else, you know, some people have it, some people don't. Some people are in between. *You deal with them based on who they are not how they got to be that way.*<sup>221</sup>

Such perspectives encourage researchers to focus less on the genetic or biological causes of autism and more on how to work with people as they are right now. This father hopes that researchers will focus on how to connect their specific capabilities and lives with the rest of the “neurotypical” world. One participant was worried that genetics research could lead to a way of making it possible for fewer people like himself to exist and suggested that research should focus more on how people can be independent, contributing members of society.

Another predominant theme regarding advice participants wanted to share with scientists conducting autism research concerned the issue of difference. Many of the participants emphasized that with ASD came variability and that no two people with ASD were alike. As such, research should look at how people on the autism spectrum are similar but different. As one father described:

Maybe look at how we are all differently the same or the same only different. I think you'll find a large amount of variation in a lot of people. It's like we've all decided to specialize in different things but the way in which we decide to specialize in them may be of similar mechanisms.<sup>208</sup>

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<sup>221</sup> ASD Adult Interview #4 – Parent of a child diagnosed with Asperger syndrome, self-identified with ASD

One participant wanted to remind researchers to be opened minded and to not expect everybody to exactly fit diagnostic criteria. She points out that there are many different manifestations of the same illness. To address this variability of manifestations, one participant's advice was for researchers to consider creating a refined taxonomy or sub-grouping of diagnostic criteria that translated into specific treatments. At the same time, one parent wanted to see less "*esoteric*" terms used to describe autism (e.g., pervasive developmental disorder, non-verbal learning disorder) and more simplified ways of describing it without making it "*too simplistic*" or "*so labeled*" that people no longer know what researchers are talking about.

### **Subjectivities of ASD**

This chapter reveals that the interpretations of autism genetics by adults on the autism spectrum are grounded in their everyday life experiences that reflect heritable traits associated with autism and, for some, the self-identification process of being on the autism spectrum. The first form of subjectivity is associated with similar characteristics and traits the participants recognized in family members, based on everyday life experiences. Not everyone used the language of "genetics" but rather implied an inherited nature of autism by referring to different generations of family members and their specific traits associated with autism. Such traits or characteristics included problems with social skills, language delays, sleeping problems, depression, as well as aptitude for skills such as computer programming or the ability to decipher complex systems. All served as heritable traits associated with ASD shared among family members and formed a link between certain family characteristics and autism. Although

many of these specific qualities or traits are not officially part of the diagnosis of autism based on the most recent *Diagnostic and Statistical Manual of Psychiatric Disorder 4<sup>th</sup> Edition Revised (DSM-IVR)*(APA, 2000), they were recognized by these adults on the autism spectrum to be very much part of “the autism” in their family.

These findings parallel other research on risk perceptions of complex diseases in families, as based on the disease experiences within the family regardless of knowledge of genetic risk information (Richards, 1996; Richards & Ponders, 1996; Lock et al., 2006). Richards and Ponder demonstrate how knowledge of “disease running in the family” can exist independent of medical (or genetic) definitions and that susceptibility to illness can be associated with similar personality and physical dimensions (Richards, 1996; Richards & Ponder, 1996). They argue that lay knowledge of inheritance is grounded in concepts of kinship and is sustained by everyday social activities and relationships. This may make them particularly resistant to change. Thus, lay accounts of inheritance diverge from Mendelian explanations and rely instead on the relations and resemblances to affected relatives in the perception of disease risk (Richards & Ponder, 1996). This is also similar to Lock and colleague’s expansion of the concept “blended inheritance”, an idea prevalent among many people about inheriting a mixing or blending of entities or traits from both parents, assumed to be passed on from generation to generation in clusters are related to risk of disease (Lock et al., 2006, p.282). Based on studies of late onset Alzheimer’s disease (LOAD), Lock and colleagues show that when diseases such as LOAD occur in a family, there is a consistent tendency to identify a family member who in some way resembles the affected person as the individual most likely to be at risk for developing the disorder, despite the knowledge of genetic

information (Lock et al., 2006). Anne Kerr and colleagues (1998) refer to this as “technical knowledge”, which is among the different types of knowledge (or lay expertise) that people have about the new genetics.<sup>222</sup> Under technical knowledge, these authors found that participants generally centered around notions of heredity through the identification of physical characteristics and occurrence of disease (Kerr, et al., 1998).

What differs in this study compared to concepts of “disease running in the family” or “blended inheritance” is that the traits in question are related to the characteristics of autism itself (i.e., social awkwardness, being a late talker, depression) and not just physical characteristics associated with the diseased individual, such as looks or body shape. A discourse on risk was also not a central theme in these interviews, since the onset of autism is typically before the age of three.

A second form of autism subjectivity was based on the self-identification process of being on the autism spectrum among parents of a child diagnosed with autism. Here, the interpretations of autism genetics manifested based on life long experiences of feeling “odd” and having a child diagnosed on the autism spectrum. For several of these participants, self-identification with ASD had been a life long process of reflecting on past experiences and future possibilities based on shared experiences and challenges they have in common with their children. This cohort of parents who self-identify as being on the autism spectrum represents a generation of individuals who grew up with labels of being “odd”, “weird” or “quirky” during a time when the diagnosis of “autistic disorder” or “Asperger syndrome” were not available.

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<sup>222</sup> The other types of knowledge include: methodological, institutional, and cultural (Kerr, et al., 1998).

The ongoing and changing processes of identifying with autism in this older cohort was not without denial, confusion or questioning of how autism is part of their identity both in the past and the future. Most in this older cohort did not feel a diagnosis or a genetic test would help them in any way because a diagnosis would not change who they were. One father was willing to get a diagnosis so that he would feel like he had a community he could call his own. The ongoing reframing of past experiences and behaviors and coming to terms with what it means to be autistic is reminiscent of autobiographies of adults who did not grow up with a diagnosis of ASD (Lawson, 1998; Willey, 1999). For example, Liane Willey eloquently describes her past as follows:

Remembering can teach me who I am and guide me toward who I will be. Remembering can set me free...I would never turn back in search of regrets or mistakes or misdirected thoughts. I simply use my past as a catalyst for conscious thought and for self-appreciation (Willey, 1999, p.17)

Similarly, the older adults in this study placed past experiences and challenges into perspective once they came to terms with their autistic identity. Some were hopeful and optimistic like Willey, viewing their autistic identity positively in the sense that it helped them explain problems they have had throughout their lives and gave them something that they could use to move forward in their lives. Others struggled with the past because it brought into perspective just how much they were “*missing out*” on throughout their lives.

The links individuals made with their past to the present and the process of self-identifying with ASD is also similar to Armstrong and colleagues (1998) notion of “revealed identity”. These authors argue that, unlike identities conferred by the diagnosis of many chronic medical conditions, a genetic identity is presented as “an old one that is

now revealed” (Armstrong et al., 1998, p.1653). By considering the past self and future self of patients at risk for genetic diseases, these authors show how illness became a ‘family’ matter, where genetic identity is revealed and established within a web of genetic connectedness to their past and future family relationships (Armstrong, Michie, & Marteau, 1998). Although no formal genetic information associated with autism was part of the self-identification process of ASD in this study, the notion of interconnectedness to past, present and future generations based on what they believed to be heritable traits of autism, influenced families to view themselves in the context of ASD.

### **Diagnostic Boundaries**

The familial ASD traits described by the participants were a combination of first, symptoms used for an official diagnosis of autism, which are based on language impairments, social interaction deficits, and the presence of stereotyped and repetitive behaviors (APA, 2000). Second, were characteristics such as depression, sleeping problems, sensory issues, and positive aspects of ASD, which are not part of the ASD diagnosis. This calls to question the boundaries placed on medical diagnosis and what symptoms constitute a disorder when, in cases like this, people identify certain traits and characteristics to be associated with autism that are representative of the family itself and not necessarily part of the ASD classification under the DSM-IVR diagnostic criteria (APA, 2000). Bridgett Chamak and colleagues also highlight how personal experiences of adults on the autism spectrum and the core symptoms they recognize as autism, such as unusual perceptions and information processing, are not part of current diagnostic

criteria (Chamak, et al., 2008). Thus, scientific and medical knowledge of autism does not represent the range of symptoms experienced by people on the autism spectrum.

This is reflective of Bowker and Star's (1989) concept of "torque", which they describe as "misalignment of a patient's life expectation, the uncertainties of the disease and of the treatment, and the negotiations laden with other sorts of interactional burdens...A twisting of time lines that pull at each other, and bend or twist both patient biography and the process of metrication" (p.27). Individuals with ASD are caught in the transitions between symptoms outside the boundaries of standardized classification systems such as the DSM, which Bowker and Star argue is "itself a broken and moving target." (p.191) This creates uncertainties as to what to call it, how to treat it, and the social entitlements of individuals in this state. Miller and colleagues (2005) refer to this as "nosological torques" in the cases of cystic fibrosis, tuberous sclerosis, and muscular dystrophy. These authors highlight conflicts between a clinical way of defining disease, one which demands manifest evidence of dysfunction in the organism, and more esoteric and occult measures of disease existence.

This can certainly affect the possibilities for future research on the genetics of autism, which, as Shostak and colleagues (2008) have demonstrated in the case of clinical depression, rely on prior medicalization and standardization of the phenotype. Phenotypes outside of the diagnostic frame may not be legitimate starting points for researchers because, despite recognition that particular traits are apparent in certain families with autism, the institutional and structural processes in health-care delivery, research and development, and advocacy are likely to be based on the medical definitions of autism (Shostak, et al., 2008). This is evident in chapter three, where I discuss the



current state of autism genetics research and how the phenotypes used for the diagnosis are being genetically reclassified and divided into subgroups to enable statistical genetic results. The autism specific genetic databases (i.e., AGRE and Simons Simplex Collection), systematically collect phenotype data from standardized instruments used for clinical diagnosis of ASD, thus the research that results from the utilization of these resources represents pre-determined symptoms that exists in current diagnostic criteria. Furthermore, qualification for services relies on the diagnosis of autism based on amendments made in 1990 to the Individuals with Disabilities Education Act (IDEA).<sup>223</sup> By adding “autism” to the list of conditions recognized by law as a disability, IDEA expanded the class of individuals who could be protected by antidiscrimination law. Moreover, as demonstrated in chapter two, the AGRE and the AGP are genetic initiatives that were generated and supported by parent advocacy groups that support the biomedical model of autism. The trajectories and progress of all these examples rely on the medicalized definition of ASD, which has expanded over time and taken little account of the experiences of people with autism.

The historical expansion and contractions of the diagnosis of autism and other autism spectrum disorders such as Asperger syndrome and pervasive developmental disorder not otherwise specified (PDD-NOS) has undoubtedly influenced the identities of people diagnosed with an ASD. But it has also influenced older adults without an ‘official’ diagnosis, as demonstrated in this study. Since 1980 when autism first became a

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<sup>223</sup> The IDEA mandates “free and appropriate public education for all students with disabilities in the least restrictive and most integrated environment possible” (Public Law 94-142, 1975). As a result of adding “autism” as a disability protected by law, numerous state and local educational agencies had to alter their policies and practices of educational programming for young children with autism. For a thorough analysis of the impact of the legal system on educational programming for young children with ASD see (Mandlawitz, 2002).

diagnostic entity, the term itself has changed as well as the criteria of diagnosis. What began as a diagnostic label of “infantile autism” with a set of specific and narrow criteria ranging from the age of onset before age 30 months to gross impairments in communication and language (APA, 1980) is now described as “autistic disorder” with a broader set of criteria based on a range of behavioral criteria (APA, 2000).

In 1994, the DSM added Asperger’s disorder among one of the pervasive developmental disorders under which autistic disorder is classified. Since the addition of the Asperger’s label, a community of “Aspies” has emerged, many of who want their strengths to be acknowledged and wish not to be “cured” (Bagatell, 2007; Chamak, 2008; Silverman, 2008a). In 2010, the American Psychiatric Association proposed a draft of the DSM-V, which eliminates the diagnosis of “Asperger’s disorder” to be replaced as a certain level of severity among the “Autism Spectrum Disorder” (APA, 2010). These changes have ignited severe scrutiny among the “Aspie” community and have major implications for the identities of people diagnosed or self identify with Asperger syndrome.<sup>224</sup> Although the interviews conducted for this study occurred before the publication of the DSM-V draft, the majority of participants were either diagnosed or self-identified with Asperger syndrome. They also clearly distinguished themselves from autism or autistic disorder and felt that this label misrepresented who they were – verbal, smart and “high functioning” adults. Thus, the changes in the DSM-V to dissolve Asperger’s under the label of “autism spectrum disorder” may be somewhat problematic for the participants interviewed in this study.

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<sup>224</sup> There has been extensive debate about the elimination of the Asperger’s diagnosis in the DSM-V draft among all the stakeholders involved, including people with ASD, parents, clinicians, scientists, and policy makers. This issue is beyond the scope of this chapter and dissertation but will be addressed in the future. The debut for the DSM-V is set for May 2013.

## **Conclusion**

The unique nature of this study sample offers important messages about the potential use of genetic testing for the diagnosis of autism in the future. Advice from the participants of this study to researchers studying the genetics of autism reflects some of the issues raised by autistic advocacy groups analyzed by Silverman (2008) and Orsini (2009). These included focusing on the everyday struggles of autism and less on the biological or genetic causes. They wished to see research that focused on how people with autism can become independent contributing members of society and how society can better work with people on the autism spectrum as they are right now. They also hoped that future research would consider the vast differences of people with autism and how different manifestations of the same illness might be incorporated into specific diagnoses and targeted treatment. These perspectives lend insight as to how researchers and people can create space, acceptance, and understanding towards people with autism with the aim of helping them live more functional and happy lives.

## **CHAPTER 7: CONCLUSIONS, THEORETICAL IMPLICATIONS AND FUTURE RESEARCH**

### **Social and Scientific Representations of Autism Genetics**

This dissertation has provided a broad view of social and scientific representations of autism genetics. It engages the social implications of genetic technologies and the production of genetic knowledge by analyzing autism genetic interpretations generated by four different sites: (a) health social movements concerned with advancing the understanding of autism genetics; (b) scientists of various disciplines who study autism genetics; (c) parents with ASD children who participate in genetics research; and (d) individuals experiencing ASD. The analysis of these social worlds together highlights the many institutions, people, theories, materials and practices involved in the heterogeneous processes of producing and representing genetic knowledge.

I began this dissertation by analyzing the health social movements of autism and autism genetics. In chapter two, I discussed how parent advocacy groups (i.e., the National Alliance for Autism Research (NAAR) and Cure Autism Now (CAN), and now Autism Speaks) have become significant authorities in the engagement of health and well-being of individuals with autisms, direct contributors to the production of biomedical knowledge, and specifically, in the case of the Autism Genetic Resource Exchange (AGRE) and the Autism Genome Project (AGP), initiators in the production of genetic knowledge. I highlighted the active role parents have played in science through fund raising, lobbying, participating in research prioritization, donating specimens, and organizing scientists to conduct autism research. I argue that the shift in scientific norms

of data sharing policies initiated through AGRE and collaborations among a diverse set of scientists in efforts like AGP have been transformative and influential in the current conduct of genome science. Their push towards a genetic understanding of autism through the development of collaborations, consortia and collections has fueled autism research towards a mainstream research agenda that investigates autism as a genetic and neurological disease.

Within the broader health social movements of autism discussed in chapter two, I also discussed a plethora activities conducted by these parent advocacy groups, ranging from public awareness campaigns, to family services, fundraising events, to government relations. I also highlighted the new and innovative ways CAN, NAAR, and now Autism Speaks are redefining autism from a rare childhood disease to a major public health issue through their use of well-known people who have children with autism in the public sphere; the creation of the Ad Council campaigns, Utube videos, and public appearances in major media venues that claim autism as an epidemic; and the use of the new generation of technological mediations that keeps autism at the forefront of social networking. I argue that collectively these actions justify viewing autism activism as a health social movement comparable to HIV/AIDS (Barbot, 2006; Epstein, 1996) and breast cancer movements (Klawiter, 1999, 2004; Kolker, 2004).

In chapter three, I analyzed the production of genetic knowledge within the social world of autism genetics. I described the historical transformations in autism genetics research, the research challenges in ASD that are prompting new genetically constructed meanings of autism, and new knowledge producing technologies that are shifting the genetic disease paradigm from inherited single gene causing mutations to rare genetic

variants that are spontaneously acquired. I argued that autism is currently being redefined based on genetic knowledge in three ways: the identification of copy number variants; genetic reclassification of autism phenotypes; and the convergence of common biological pathways. Throughout this chapter, I contend that current microarray technologies that can scan the entire genome at higher and higher resolutions are driving the science of autism genetics reorganizing this domain of research. This parallels with Shostak's (2005) findings that these technologies were key in the development of toxicogenomics. Thus, the ability of genetic technologies to refine the DNA analysis to smaller and smaller segments of each individual chromosome has constructed new ways of interpreting and understanding illness and disease. I demonstrate that despite the limited successes of autism genetic research to date, private and public support continues to focus on providing funding and biological resources to conduct autism genetics research.

In chapter four, I shifted focus to the social worlds of families with children diagnosed on the autism spectrum. Here, I analyzed how parents of children with ASD came to participate in genetics research. I demonstrated that there are many uncertainties, forms of knowledge, and emotional experiences embedded in the decision of donating blood and medical information to a national disease specific genetic database (i.e., Simons Simplex Collection (SSC)). For these parents, the motivation to participate in a genetics research study centered around three themes: to get a free diagnostic evaluation; the desire to help their child; and the willingness to help further autism science in any way possible. However, I argue that these processes are tied to different situations and experiences that occurred prior to participation in the study, such as the need for a diagnostic evaluation for educational services. Parents also hoped the SSC study would

find the causes of this disorder, how it can be identified sooner, and how it can be “fixed”, “cured” or “prevented” in the future.

Despite the range of ethical possibilities that have been brought to the forefront of genetics research and the development of genetic databanks, the participants in this study did not have any major concerns about participating in a genetic research study. I contend that this lack of concern can be attributed to the altruism of parents wanting to help their children in any ways possible for them, as well as the trust these parents have in science and clinical institutions that conduct genetics research. The data further suggested that the limited knowledge these parents had of how genetics contributes to human disease in general, and the goals of the SSC study in particular, may also be related to the lack of concern expressed by the parents. Thus, this chapter highlights ethical implications that extend beyond traditional bioethics and engage in a new dialogue of bioethics that takes into consideration the social and moral situations of families living with ASD and who want the best for their child.

Chapter five of this dissertation is an attempt to literally “follow the DNA” in order to highlight the ways in which family information (i.e., blood and family characteristics) is transformed and processed into genetic knowledge through the different yet overlapping spaces of families and individuals with autism, parent advocacy groups, and scientists. Drawing from previous chapters and collected visual and technoscientific representations of families of autism, this chapter sociologically unpacks the transformations of family experiences and of their material blood produced through key site processes. These include the process of creating a pedigree, the generation of immortalized cell lines, the production of DNA, the creation of bioinformatic and genetic

databases, the “transcriptions” of genotypes, the “discovery” of nucleotide alterations, and finally, scientific “translations” of microarray fluorescence intensities. By “following the DNA” in this way, I analyze the boundaries within which these transformations take place, and the people, institutions, tools, and technologies that enable such transformations. I also show how the movement of biological materials and clinical information from families to central repositories, to laboratories around the world, and back again are fluid and multi-directional processes.

The social world of adults living with autism is the center of analysis of chapter six. Here, I argued that the interpretations of autism genetics in adults on the autism spectrum are grounded in everyday life experiences that reflect heritable traits associated with autism and, for some, the self-identification process of being on the autism spectrum. The first form of autism subjectivity is associated with similar characteristics and traits the participants recognized in family members, such as social abilities, language delays, sleeping problems, depression, as well as aptitude for learning skills such as computer programming or the ability to decipher complex systems. These were all heritable traits associated with ASD notably shared among family members. It was these traits that formed a genetic link between certain family characteristics and autism.

The second form of subjectivity was based on the self-identification process of being on the autism spectrum that occurred in parents of a child diagnosed with autism subsequent to that diagnosis. Here, interpretations of autism genetics are based on life long experiences of feeling “odd” and then having a child diagnosed on the autism spectrum. Interestingly, no formal genetic information associated with autism was part of the self-identification process of ASD in this study. Instead, a sense of interconnectedness



to past, present and future generations based on what they believed to be heritable traits of autism influenced families to view themselves as situated vis-a-vis ASD. This chapter also calls into question the boundaries placed on medical diagnosis and what symptoms constitute a disorder when, in the case of autism, people identify certain traits and characteristics as associated with autism that are representative of the family itself while not necessarily part of the ASD classification under the current DSM-IVR diagnostic criteria.

Together, these chapters demonstrate that when the social worlds of these actors come together there is both agreement and contestation as to what constitutes the meaning of autism spectrum disorders, the causes, the treatments, and the outcomes. The creation of new scientific knowledge is dependent on intersections of these different social worlds that require communication, and in some cases governance, for the production of new genetic knowledge of autism. The non-human actors (e.g., AGRE, SSC, microarray technologies, CNV deletion/duplication) have different meanings in these different worlds. Human actors must somehow reconcile these varied meanings if they wish to participate.

As I demonstrate throughout this dissertation, the impacts of autism genetic knowledge are constantly changing, ambiguous, and different interpretations are offered based on whom you ask. For parent advocates promoting the genetic understanding of autism (i.e., CAN, NAAR and Autism Speaks), genetic knowledge holds the keys to earlier diagnosis, future targeted treatments, and possibly a “cure”. For scientists conducting autism genetic research, the heterogeneity of ASD is impacting the direction of scientific inquiry towards newer genomic technologies and larger sample sizes. For

clinical geneticists, genetic reclassification is being implemented in the genetics clinic and being offered as a first tiered diagnostic analysis for ASD. For families who participate in genetics research studies, proper diagnosis and guidance on how they could best help their child were core priorities, with or without a genetic diagnosis. For individuals on the autism spectrum, genetic information was rather abstract and was instead framed as traits and characteristics that “run in the family”. Thus, not only do multiple social worlds contribute to the production of knowledge, but multiple, and at times conflicting, interpretations of genetic knowledge abound.

### **Theoretical Implications**

This dissertation draws heavily on science and technology studies and offers insight and expansion of the theories of biomedicalization and geneticization. Science and Technology Studies (STS) is a dynamic interdisciplinary field that investigates the contents, processes and outcomes of science and technology. It starts from the assumption that science and technology are social activities, where the sources and interpretations of sciences, technologies, and knowledge are complex and various. In general, the field of STS investigates how scientific knowledge is constructed through work, instruments, institutions, and conventions of practice to produce and legitimize it. Over the last fifteen years, there has been an expansion in the production of scientific knowledge and technologies from human genetics research. This research has been highlighted through knowledge producing events such as the sequencing of the human genome and the cloning of various genes that “cause” diseases.

Compared to social science research on the Human Genome Project or the social implications of genetic testing, STS studies have paid less attention to the social construction of “genetic” diseases and the development of genetic disease categories. Thus, this dissertation investigates how genes and mutations are identified and their influence on disease etiologies. It also considers the processes of genetics research and the implications of translating genetic technologies into clinical practice. Like other scholars who have embarked in this area of study, I demonstrate the political, social, cultural and etiological complexities of constructing autism as a genetic disorder.

***STS: The Production of Scientific Knowledge***

This dissertation theoretically offers insight into three major sensibilities of STS (Thompson, 2005), including: the production of scientific knowledge; the ontology of science; and the politics between science and technology. First, this dissertation adds to the STS scholarship focused on the production of scientific knowledge. Using social worlds/arenas theory, I identify the connections between knowledge production of autism genetics and how this influences classification and evaluation of individuals (and their families) with ASD. For example, in chapter three, I argue that current scientific interpretations of autism genetics are very much driven by technological advances in microarray technologies and the ability to scan the genome at higher resolutions. The ability to identify copy number variants has generated a new class of genetic mutations and chromosomal disorders associated with autism. Furthermore, genetic reclassification of ASD phenotypes and the convergence of disorders at the molecular level call into question the ASD diagnosis and current classification boundaries. Thus, the new genetic

knowledge producing technologies (i.e., microarrays) are redefining autism based on genetic information.

I also draw on Merton's scientific norms of communism, universalism, disinterestedness, and organized skepticism to describe how scientists altered their scientific practices to reflect the rules associated with access to AGRE or participation in AGP. The development of public databases like the AGRE and collaborative efforts like the AGP required scientists to pull their samples and share unpublished data with the scientific community, which is counter to the usual scientific competitiveness. Altering the scientific practices of researchers also required a dramatic shift in governance over the gene banks. In this case, the governance of AGRE was in the control of the parent advocacy group since they owned the biomaterials (i.e., DNA, cell lines, plasma and blood serum) and clinical data (e.g., diagnostic assessments, family histories, and medical evaluations). Thus, by coordinating and controlling the collection and distribution of AGRE samples to laboratories throughout the world, CAN influenced the conduct of biomedical research and the production of knowledge.

### ***STS: The Ontology of Science***

The ontology of science considers the nature of being, existence, or reality in general and its basic categories and their relations, particularly on the connections between science, technology and the world (Thompson, 2005). In many of the chapters presented in this dissertation, I am interested in the process of "science in the making" (Latour, 1987). Although I do not specifically follow scientists in their laboratories, I do follow transformations of DNA in the process of producing and representing "scientific

facts” (Latour & Woolgar, 1979). In chapter five, I provide a visual process of these transformations of nature by identifying the boundaries in which these transformations take place and the people, institutions, tools, and technologies needed to enable such transformations. Here, I argue that the pedigrees and phenotypic information used for AGRE and SSC represent heterogeneous systems of boundary objects described by Star and Griesemer (1989). Simultaneously, these pedigrees and their associated phenotypic data represent a system of *repositories*, *ideal types*, and *standardized forms*. I argue that these systems allowed for pedigrees and their associated phenotypic data of the AGRE and SSC to be used by various disciplines, for a diverse set of questions, and as objects that could be used to communicate across a diverse set of scientific expertise.

In chapter two, I also draw upon Bruno Latour’s concept of “obligatory passage points”, where I argue that the AGRE database has been transformed into a working tool that scientists use to generate knowledge (Latour, 1987). Silverman (2008a) also make this point in her research on CAN and the AGRE database. I argue that although scientists were skeptical of parents and their ability to create a quality database, it is now “indispensable” for many scientists, rendering the “passage” by use of the AGRE an obligation in order to conduct autism genetics research. The AGRE allowed scientists to “*focus on just doing science*” and has opened up the field of autism research to broader scientific interests, smaller labs, and new investigators. Chloe Silverman also makes this point in her research on CAN and the AGRE database (Silverman, 2008a). Thus, non-human actors, such as the AGRE and associated genetic technologies, such as microarrays, have agency. They “interact with human actors, swap properties, and are together the condition of each other’s identities.” (Thompson, 2005:47).

### ***STS: The Politics between Science and Technology***

This dissertation also engages STS theories in the politics between science and technology, where scientific truth is the object of study to identify the interactions between nature, politics and identity (Thompson, 2005: 34). Specifically, this dissertation is concerned with lay participation in the production of scientific knowledge. Chapter two is devoted entirely to the influence of autism parent advocacy groups on the production of genetic knowledge of autism. I show how the collective mobilization of CAN and NAAR, now Autism Speaks, initiated genetics research through their development and support of the AGRE database and the AGP. I argue that the emotional knowledge of parents and families of ASD and their strong desire to help their children were the “embodied” experiences that drove this biomedical research on autism. These experiences have produced genetic databases and research consortia that have enabled the production of genetic knowledge. Furthermore, in chapter four, I argue that although parents whose families participated in the SSC were less scientifically savvy than parents who initially started CAN and NAAR, these parents also drew on their emotional expertise of having a child with ASD. This emotional knowledge was deeply connected to their motivations to participate in genetics research and the hope they have for the future of their children.

### ***Biomedicalization***

This dissertation also contributes to the understanding of interactive and overlapping processes of the theoretical framework of biomedicalization (Clarke, et al., 2009; Clarke, et al., 2003). It specifically addresses: the focus on health itself and

elaboration of risk and surveillance biomedicines; the technoscientization of biomedicine; transformations of biomedical knowledge; and the transformation of bodies and the production of new individual and collective technoscientific identities.

*Risk Factors and Self-Surveillance.* The elaboration of risk and self-surveillance mechanisms at individual, group and population levels is brought forth in this dissertation in several chapters. For example, in chapter three, I analyze how scientists are constructing autism as a genetic disorder and provide an example of how the new microarray technologies and the associated scientific “facts” that are produced (i.e., CNVs) are being integrated into clinical genetics practice. Here, I make the case that the technological abilities to detect more and more genomic variation presumed to be associated with disease will result in more surveillance and clinical “intervention”. Further, the convergences of molecular pathways of different diseases, such as neurodevelopmental disabilities and cancers, open up possibilities for future risks of disease that require additional disease and surveillance mechanisms. In this particular example, people with developmental disabilities need to be monitored for cancer throughout their life, which in many cases will also involve the disciplined monitoring and surveillance by parents or primary care givers. This echoes the work of Foucault (1984) and his theory of biopower, where power is situated and exercised at the level of life itself. It represents both the disciplining of the human body and the regulatory controls of the “species body” (i.e., the biopolitics of populations).

In chapter four, I also analyzed the social world of parents of children with ASD who participated in a genetic research study (i.e., the SSC). Here, I expand on Nikolas

Rose's concept of "ethopolitics", which is a concept that "attempts to shape the conduct of human beings by acting upon their sentiments, beliefs, and values" (Rose, 2007:27). Although the parents in this study are not concerned necessarily with "self-techniques" to improve *their* own health, they are concerned with techniques that they can "judge and act upon" to make their children "better than they are" (Rose, 2007). Thus, I argue, that the responsibility parents take upon themselves to help their children and families in the future (in this case by participating in a genetic research study) also speaks to the notion of biological ethopolitics and novel forms of authority parents take upon themselves to help their children that may, in fact, be experienced as obligation.

Throughout this dissertation, I also elaborate on the standardization of diagnostic tools used for autism spectrum disorders. In chapter three, I argue that the institutional stabilization of phenotypes and the constant re-framing and expansion of ASD through various versions of the Diagnostic Statistical Manual of Mental Health Disorders (DSM) has been an essential element in the production of genetic knowledge. Moreover, emerging microarray technologies are becoming first tiered diagnostic tools for autism spectrum disorders in clinical genetics, creating a new technological risk assessments based on genetic information.

*The Technoscientization of Biomedicine.* Technological innovations in biomedicine is a predominate theme throughout this dissertation. I demonstrate that the technoscientization of biomedicine has seeped into the research process itself through the development of genetic databases that require extensive computational capabilities in order process and store large volumes of genotypic and phenotypic data generated in the research process. In chapter two, I highlight the development of the Autism Genetic



Resource Exchange (AGRE), which requires extensive computational capabilities in order to transfer the phenotype and genotype data electronically back and forth. This is done through the Internet System for Assessing Autistic Children (ISAAC), a web-based data management system that allows researchers to enter, manage, and share clinical data among other researchers in the community. This system has also served as a prototype for the National Database for Autism Research (NDAR), a bioinformatics platform for scientific collaboration around autism spectrum disorders to facilitate data sharing and collaboration within the National Institutes of Health (NIH). The NDAR is a research portal that links data, supporting documentation, publications, and grants information relevant to autism research.<sup>225</sup> These meso-level scientific infrastructures create greater private-public linkages and new portals into the manipulation and analysis of human genes.

In this dissertation, I also argue that autism spectrum disorders are being genetically reclassified based on the discovery of copy number variants, the reclassification of ASD phenotypes, and the convergence of common molecular pathways of different diseases. These developments in autism genetics research are new ways of articulating molecularization and geneticization. In chapter three, I provide an example of the types of interconnected molecular pathways that are being “imagined” based on the large volumes of genomic data and new bioinformatic systems software for mapping regulatory networks. I argue that the ability to “genetically dissect” the interconnected genetic mechanisms involved in autism are creating “scientific imaginary” pathways that could be used for diagnosis, treatment, and future genetics research on autism.

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<sup>225</sup> NIH: National Database for Autism Research. Retrieved January 19, 2010, from <http://ndar.nih.gov/ndarpublicweb/aboutNDAR.go>.

The molecularization and geneticization of autism spectrum disorders also rely heavily on the mutual construction between biological understanding of disease, computational methodologies, and the technological development of microarrays. This synergistic relationship has been essential in the development and manifestation of genomewide association studies in current genetics research. Thus, I contend that the speed of new genetic technologies is moving exponentially and requires new forms of knowledge to translate the “terabytes” of information that are being generated through this technology, namely computer bioinformatics systems (i.e., computational techniques). As a consequence, the professional scientific domains of autism genetics research have expanded to include computer scientists, epidemiologists, and statisticians, in addition to geneticists, neurologists, psychiatrists, psychologists and clinical scientists.

*Transformations of Biomedical Knowledge.* This dissertation also identifies new ways biomedical knowledge production is transformed, especially through the heterogeneity of production, distribution, and access to biomedical knowledges (Clarke, et al., 2003). For example, in chapter two, I describe how the parent advocacy group, NAAR, recruited families through their newsletter, which provided information regarding autism biomedical research and what parents could do to participate in research and strengthen their cause. Here the goal was to connect families of individuals with autism with investigators conducting clinical autism research studies. In their view, it was critical that families participate in research studies funded by the National Institutes of Health. The Internet has also served as a major recruiting tool for the AGRE database and newer technological innovations such as the IAN network.

Autism parent advocacy groups have also been tremendous resources for parents by creating and supplying different resources and tools through the Internet. These include tools such as an autism video glossary to help parents and teachers learn early signs of autism and a 100 Day Kit created specifically for newly diagnosed families to make the best possible use of the 100 days following the diagnosis of autism. The goal of this kit was to provide families with a greater sense of hope, with resources, and information that will help the first couple of months after the diagnosis easier. More recently, Internet resources created by parent advocacy groups have taken on a more political role, which enables parents and other activist to stay instantly informed of state and federal initiatives dedicated to autism, and resources on how to contact state legislators.<sup>226</sup>

The promotion of autism awareness by parent advocacy groups has also taken on many different and creative forms. For examples, in chapter two, I describe how the parent advocacy group, Autism Speaks has created autism awareness through the Ad Council campaigns and public appearances in major media venues. They also utilize social networking technologies such as Facebook, Twitter, Ning, UTube, and created an official Autism Speaks Blog, which allows for “technosocial mediation” (Heath et al., 2004, p.157) among parents and other people associated with autism. Thus, autism awareness is made routinely available to the public through electronic means. This new generation of technological mediations keeps autism at the forefront of social networking and will undoubtedly be an emerging area of research for social scientists in the future.

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<sup>226</sup> Autism Votes home page. Retrieved February 18, 2010, from <http://www.autismvotes.org/site/c.frKNI3PCImE/b.3909853/k.BE44/Home.htm>.

Parents who have a child recently diagnosed on the autism spectrum also use the Internet to help navigate the inertia that commonly follows an ASD diagnosis. For example, the parents I interviewed consistently sought resources from the Internet to translate the particular diagnosis their child received, such as PDD-NOS or Asperger's disorder. They also utilized the Internet to help decipher the best treatment options available to help their child. This information did not typically come from the clinician providing the diagnosis and was usually acquired outside of the institutional domains of medicine. This was largely due to the limited expertise clinicians had on autism. Thus, I argue that parents had to negotiate between the "science and fiction" disseminated in the media and used the Internet as a "bridge out of the uncertainties" that accompanied a diagnosis.

The collection and dissemination of genotypic and phenotypic information of the Autism Genetic Resource Exchange (AGRE) is perhaps the most compelling example of how autism parent advocates challenged the professional monopoly over the production of medical knowledge. In chapter two, I analyzed how parent advocates of Cure Autism Now developed an open-access gene bank for autism research that consisted of DNA and high-quality clinical data of multiplex families. This was in response to advice they received from scientists regarding the single most important thing they could do to speed progress in autism research. As a result, the AGRE is the world's largest private repository of clinical and genetic information on families affected with an ASD. The development and use of the AGRE collection has dramatically accelerated autism genetics research, opened up the possibility of conducting autism research to smaller and broader research groups, and created a paradigm shift in collaboration and data sharing

policies. Furthermore, the governance of AGRE is in the control of the parent advocacy group (now Autism Speaks) since they exclusively own all the biomaterials (i.e., DNA, cell lines, plasma and blood serum) and the clinical data (e.g., diagnostic assessments, family histories, and medical evaluations) in the AGRE database.

Another parent advocacy group, the National Alliance for Autism Research (NAAR), also challenged the exclusive production of knowledge by funding of the Autism Genome Project (AGP). This project facilitated collaboration and communication among autism genetic scientists around the world and marked the largest collaboration ever to focus on the genetics of autism. By pooling their samples, the AGP was able to conduct the largest genetic linkage analysis that had ever been studied. I argue in chapter two that the AGP was an integral part of the initiation of the scientific response to consider the role of copy number variants and the genetic heterogeneity autism.

Thus, the transformations of information and the production and distribution of new knowledge about autisms took many forms, especially via the Internet and largely through the promotion of parent advocacy groups. Not all are beneficial. For example, despite limited knowledge of the genetics of autism, marketing of autism-specific genetic tests are emerging via the web. Private companies such as IntegraGen and GeneDx are now selling autism specific genome-wide microarray analysis and DNA sequence analysis directly to the public (through a physician's recommendation). Holistic Health International (HHI) is also selling a comprehensive methylation panel with methylation pathway analysis to "optimize supplementation" that will address "genes, environmental

toxins, and infections [that] all contribute to autism”.<sup>227</sup> Thus, in the absence of any regulation, genetic testing for autism is available directly to the public and the proliferation of tests of varied value and accuracy that specifically target autism will continue to rise.

*Transformation of Bodies and Identities.* The transformation of bodies and the production of new individual and collective technoscientific identities marks the final process of biomedicalization (Clarke, et al., 2003). The ability to obtain enhanced knowledge about individualized susceptibilities and potential pathologies has undeniably expanded through new genomic technologies. For example, in chapter three, I argue that the ability to quantitatively assess genetic changes at a resolution of a few hundred base pairs have made it possible for scientists to “discover” new syndromes based on the deletion or duplication of genomic segments of 500 kb to 2Mb in size. Thus, current technological applications of human genetics, such as microarray technologies, involve conceptualizing disease at the molecular level and serve as a means of producing and representing new forms of human disease and categorization. This new classification of genetic syndromes is expected to grow in the future now that current microarray platforms can scan up to one million segments of DNA distributed throughout the genome.

A form of “stratified biomedicalization” (Clarke et al., 2003:182) is also exemplified through genetic reclassification of ASD phenotypes. In chapter three, I contend that autism genetic researchers are devising ways in which to manipulate the

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<sup>227</sup> Holistic Health International: Comprehensive methylation panel with methylation pathway analysis. Retrieved May 25, 2010, from <http://www.holisticheal.com/health-tests/nutrigenomic-testing/comprehensive-methylation-panel-with-methylation-pathway-analysis.html>.

phenotypic data to gain statistically significant results. In the process, however, they are creating new classifications of autism based strictly on genetic interpretations of disease. Furthermore, genetic reclassification of ASD traits has also expanded to include unaffected family members, such as parents and siblings of a child diagnosed with ASD. Thus, what is transpiring in autism genetics research today is the reclassification of ASD at the genetic level based on identification and development of sub-phenotypes represented in ASD. This scientific approach has enabled new categories of people with autism to emerge, expanding the boundaries of ASD to include symptoms here to fore outside of the diagnostic category, and implicates parents of children with ASD (as well as their unaffected siblings) who may have specific traits that are at one end of the continuum of “normal” behavior and cognition.

Identities constructed through technoscientific means are also emerging through the production of genetic knowledge of autism. For example, in chapter three, I discuss in detail the emergence of new ASD genetic classifications based entirely on genetic technologies such as the copy number variant (CNV) 16p11.2 deletion. Prior to the use of high-resolution microarray technologies, micro deletions of this size were unknowable. Now they are among the most highly sought genetic mutations by scientific research communities studying autism genetics. Furthermore, with regard to clinical genetics, the 16p11.2 deletion is being screened in some labs as a first tier analysis using microarray technologies, and labeled “causative” of the ASD diagnosis if found. Hence, new biosocial collectivities (Rabinow, 1992) have emerged consisting of children given a specific diagnosis based on the new microarray technologies that can detect the 16p11.2 deletion and other copy number variants.

In chapter six, I also demonstrate how technoscientific identities are negotiated, accepted, or rejected. This chapter reveals that the interpretations of autism genetics by adults deemed to be the autism spectrum are grounded in everyday life experiences rather than genetic information alone. Autism subjectivities based on genetics reflected traits associated with autism that “run in the family”, and for older adults whose children were diagnosed with an ASD, the self-identification process of being on the autism spectrum subsequent to their child’s diagnosis. For these participants, current scientific efforts to identify the genetic mechanisms for autism had not yet influenced their identity. What had influenced identity in this particular set of individuals were everyday life experiences of living with autism and the identification of traits and characteristics in themselves and in other family members that were believed to be genetically associated to ASD.

### ***Health Social Movements***

This dissertation also adds to the growing body of literature that focuses on health social movements. In chapter two, I identify how parent advocacy groups influenced priorities and practices of biomedical research and the active role they have played in scientific knowledge production through fund raising, lobbying, participating in research priorities, donating specimens, and organizing scientist to conduct autism research. I contend that NAAR and CAN, and now Autism Speaks have become significant authorities in the engagement of health and well-being of individuals with autisms, direct contributors to the production of biomedical knowledge, and specifically in the case of AGRE and AGP, initiators of the production of genetic knowledge.



There are unique dimensions of the autism health social movements that are not necessarily articulated in either the “embodied health movement” (Brown, et al., 2004) or the “partnership model” (Rabeharisoa, 2003). First, the lived experiences of individuals with ASD are not central to advancing the biomedical understanding of autism. Rather, it was the emotional experiences of parents and families of children diagnosed with ASD who were challenging science on etiology, diagnosis and/or prevention of disease. I argue that the emotional knowledge of parents and families of ASD and their strong desire to help their children were the “embodied” experiences driving the biomedical research on autism. As noted, these experiences helped produce genetic databases and research consortiums that enabled the production of genetic knowledge.

Another unique feature of the parent advocates described in this dissertation was their ability to transform their talents to fit the needs of promoting biomedical research on autism. For example, the founders of CAN, Jon Shestack and Portia Iverson, transformed their skills of being Hollywood professionals into “producers” of genetic knowledge. These transformations were also evident within the workings of the advocacy organizations that recruited parents of children with ASD who have expertise in medicine, law, science, marketing, and health informatics.

The emphasis placed on families in the collection of biomedical data and participation in autism research was also crucial for autism health social movements. In the case of AGRE, the family centered approach through in-home collection of data and AGRE newsletters created a strong relationship and commitment between AGRE and the families who are part of the collection. Another emerging action of autism health social movements was the ability of both CAN and NAAR to leverage their pilot projects into

millions of dollars over time in autism research awards by the National Institutes of Health (NIH) and other governmental funding sources. This created a sustainable research endeavor on autism and established a level of legitimacy of their research goals within the broader scientific community that enabled and sustains commitment to such research.

Finally, the shift in scientific norms of data sharing policies initiated through AGRE and collaborations among a diverse set of scientists in efforts like AGP have been transformative and influential in the current conduct of genome science. The AGRE data sharing policies have likely influenced the development of national research databases, such as the NIMH Genetics Initiative and the National Database for Autism Research, which both initiate and promote data sharing and collaboration among scientists. The ability of the AGP to effectively collaborate internationally with multiple groups of scientists has influenced the development of additional autism consortium, such as the Boston Autism Consortium<sup>228</sup> and the Autism Sequencing Consortia (Packer, 2010), as well as other psychiatric disease-based research consortia (e.g., schizophrenia).

Similar to “embodied health movements” framed by Brown and colleagues (2003) and the “partnership model” proposed by Rabeharisoa (2003), as well as other health social movements described in the past (Epstein, 1996, 2008), parent advocates for autism described in this study were highly motivated to collaborate with scientists and health professionals in pursuing treatment, prevention, research and funding. The initiation and establishment of the AGP and AGRE highlights the collaboration of parent

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<sup>228</sup> Autism Consortium. Retrieved April 23, 2010, from <http://www.autismconsortium.org/about-us/about-us.html>.

advocates with scientists in pursuing genetics research and the expansion of funding to support new and innovative research. These parents also became specialists in their own right in order to better understand the potentials and limitations of science. Furthermore, the “partnership model” proposed by Rabeharisoa (2003) also reflects the work of CAN and NAAR effectively, especially with regard to the shift in the balance of power. For both CAN and NAAR, the reversal of power was apparent given that these organizations took ownership of the direction of autism research to consider the biological origins of autism. However, I argue that differential stakeholder power emerged in these two genetic research initiatives. For CAN/AGRE, the power was clearly group initiated and remains within the parent organization itself. In contrast, the AGP comprised a mixture of power relations among the scientists themselves and with the parent organization.

## **Substantive Implications**

### ***Sociology of Genetics***

*Geneticization.* This dissertation also adds to the growing literature on the sociology of genetics. It offers a critique to the concept of “geneticization” that emerged in the 1990’s to capture the ever growing tendency to distinguish people from one another on the basis of genetics and to define most disorders, behaviors, and psychological variations as wholly or in part genetic in origin (Lippman, 1992, 1998). In the case of autism spectrum disorders, I challenge the geneticization thesis by considering the social, cultural and political factors involved in the production of genetic knowledge. I demonstrate that geneticization does not exist independently, but within a larger fabric of “networked complexities” consisting of biological materials, technologies, scientists,

families, advocacy groups and many others. Furthermore, the meanings, interpretations, and representations of autism genetics take on many forms within each of the social worlds examined in this study, which future challenges assertions of one “true” meaning of ASD.

However, in chapter three, I demonstrate how genetic determinism lives subtly in the discourses of scientists conducting autism genetics research. Following from Adam Hedgecoe’s concept of “enlightened geneticization” (2001), I argue that on one hand scientists are privileging genetic explanations for autism through various mechanisms, such as minimalizing environmental factors contributing to autism and the consistent acknowledgement that autism is highly heritable, and thus has a genetic etiology. Further, scientific failures to identify genes that have a major effect in causing autism are in fact driving new genetic research models, methodologies, technologies, as well as the drive for larger sample sizes. Contrary to this discourse, I demonstrate how genetic results are highlighting and reinforcing the etiologic complexity of ASD, as well as the boundaries of diagnostic categories.

Despite the ambiguous and uncertain nature of current knowledge of autism genetics, research in this area continues to move forward. Microarray technologies are very significant here in terms of organizing new lines of work that essentially represent a scientific “bandwagon” (Fujimura, 1996). These technologies have led to the ‘discovery’ of several CNVs that are now being recommended as part of clinical genetic testing despite their unknown function and lack of availability of targeted treatment. Thus, I argue that the emerging technological advances of microarray analysis in clinical genetics to diagnose autism expand on Foucault’s concept of biopower. Here, autism is

conceptualized at the molecular level, which serves as a means of producing and representing new forms of human disease and categorization. This in turn has led to new levels of social control, especially by parents and caregivers of children with ASD.

*Ethics of Genetics Research and Testing.* This dissertation also offers insight into the genetic research process from the perspective of families experiencing ASD. Genetics research of the past would typically derive information from the individual in the family experiencing disease (i.e., proband). However, in the current study, participation is at the discretion of parents, and the information supplied is based on parent and professional observations and blood donations. Thus, in many regards, a new type of genetic research study is ensuing via childhood developmental disorders like autism. As I discuss in chapter four, the social and moral contexts within which parents of children diagnosed with an ASD come to participate in genetics research imply a wider concept of engagement and another level of participation that moves beyond the conventional mode of bioethics formulations. For example, confidentiality and privacy concerns of participating in genetics research and donating blood and medical information to a genetic database were not major concerns for parents interviewed in this study. This was largely due to their overriding goal of helping their children in any way possible. For some, there was also the desperation of receiving a proper diagnosis, normally costing parents up to \$2000 and taking up to a year to get an appointment for an evaluation.

Although the majority of parents did not have a firm understanding or genuine concerns of the SSC study goals, their trust in the academic institution and associated autism center conducting the study enabled parents to “feel good” about participating

regardless of this knowledge. Thus, the limitations of knowledge that many of the participants had about the specific aspects of the study itself were to some degree allied with a choice to place trust in the academic institution conducting the research. This level of trust is imperative for studies like the SSC to succeed and really calls to question the implications of having limited knowledge and expertise in relation to research participation

Another layer of ethical consideration regarding families participating in genetics research was the time it took for the SSC study to return the research evaluations to the parents. This was the biggest complaint parent's had about the study in general, especially for those parents who were relying on the research evaluation to aid them in obtaining special educational services. This raises issues of follow up in research: Should it be part of the study protocol to verbally follow up with participants when they are seeking a specific diagnosis, especially for participants potentially receiving a complex diagnosis like autism for the first time?

The fact that these families are donating blood and medical information to a genetic database also informs sociological research on genetic databases. For example, since samples can be immortalized and stored indefinitely, genetic databases present a particular challenge to informed consent requirements. It is also often impossible to anticipate the types of studies that will utilize the samples in the future, given the constant changes in genetic knowledge and technologies. This raises another ethical concern of how often families should be burdened with follow up studies if scientists 'discover' a genetic mutation in only a select number of families.

This dissertation also explored the influence and use of genetic testing for autism in families of children with autism, as well as adults diagnosed or self-identified with ASD. Parents of children with ASD whose families participated in a genetics research study overwhelmingly felt that genetic information would benefit their family's future generations, since there was not much they could do with that information today. Parents also felt that genetic information on the risk of ASD, whether provided prenatally or when the child was born, would help parents prepare for a child with a disability. However, two parents expressed concerns that genetic information would only help those who could afford long term therapies and would further increase the disparities that exists among those who can and can not afford diagnosis and treatment.

For some adults on the autism spectrum, genetic information alone would not change anything. Several participants felt that even if a genetic test existed for autism, the results would not be important since their issues of autism would still have to be addressed. One participant was worried that a genetic test would disprove her diagnosis and that her challenges with communication, social interaction and extreme anxiety would be deemed "*pathological*" rather than "*a character trait*". However, two fathers with children diagnosed with autism and who self-identified with ASD viewed genetic testing positively in the sense that a test early on would help with how best to target treatment for a child with autism.

### ***Sociology of Autism***

This work also contributes to the growing sociological analyses on autism spectrum disorders by considering multiple sites of knowledge production on autism genetics. It

offers insights into current work that focuses on adults on the autism spectrum, especially, older adults who do not have an official diagnosis. Furthermore, it represents individuals on the spectrum who are not currently engaged in activism for autistic individuals. Rather, they represent a snapshot of experiences of the everyday lives of autism. The focus on families and their participation in genetics research by donating blood and clinical information is also a very unique contribution to the field of autism. Between AGRE and SSC, there will be over 5,000 families who participate in these efforts. This study offers a snapshot of why parents participate and their hopes and expectations for genetics research. The perspective of scientists who participated in the AGP and/or utilize the AGRE is a unique example of how scientists view large research collaborations, consortia, and collections. The futures of these projects are moving targets, but the influence of their existence will undoubtedly be recognized for years to come as more disease specific consortia and genetic databases begin to emerge. By focusing on the broader health social movements of autism, this research also begins to introduce the impact of new and innovative ways of increasing the awareness of disease. Finally, the focus on the science of autism genetics sheds light into the complexities, negotiations, contradictions, and uncertainties of autism genetic knowledge.

In sum, this dissertation offers a wide range of theoretical and substantive insight into the interpretations and representations of autism genetics. By considering “science in action”, the production of scientific facts, the process of genetics research, and subjectivities of autism in adults experiencing autism, I analyzed the impacts of genetic technologies based on multiple levels of understanding. Any of these autism sites can serve as a basis for further investigation, discussed next.



## **Implications for Future Research**

Autism spectrum disorders are not alone in their etiological complexity and the scientific drive to determine the genetic causes. Thus, the results of this study may reflect other sociological analyses of complex diseases that embrace a genetic etiology and/or “cure” through genetics research and technologies. However, the enormity of autism spectrum disorders from all fronts – science, advocacy, causes, education, treatment, and policy – reveals the partialities of this dissertation. Thus, the implications of this research warrant future investigations in many areas concerning autism and genetics. Furthermore, the pace of new genetic technologies and scientific “discoveries” will continue to evolve. This will undoubtedly generate new questions for social scientific inquiry.

First, drawing from the biomedicalization process of political economic shifts (Clarke, et al., 2003) and theories of biocapital (Rajan, 2006), future work must consider the political economic emergence of pharmaceutical compounds that are targeting specific symptoms of autism, as well as autism genetic testing panels that are available directly through the Internet. For example, despite limited knowledge of the genetics of autism, marketing of autism-specific genetic tests are emerging via the web. Private companies such as IntegraGen and GeneDx are now selling autism specific genome-wide microarray analysis and DNA sequence analysis directly to the public. Pharmaceutical giants, Novartis and Pfizer, have also recently announced commitments to develop treatments for autism spectrum disorders (Harris, 2010). As autism continues to be at the forefront of public awareness and strongly supported through both private and public

entities, the issues of patent commodities, industry-academic collaborations, and industry-sponsored research will be of increasing importance.

Second, the impacts of using microarray technologies in the clinic to screen emerging CNVs believed to be involved in autism call for further investigation. The ways in which these types of mutations are conveyed in the clinic and how they influence disease identity constructions at the individual and family levels will undoubtedly be an important area of social scientific investigation in the future (Latimer, 2007; J. Latimer, et al., 2006; Rabeharisoa & Bourret, 2009). Furthermore, the extent to which reclassifications of ASD based on genetic information impacts the diagnosis, treatment, and technoscientific identities will be of import in the future. Especially as whole genome sequencing becomes feasibly available, it will undoubtedly enable future ASD classifications to elaborate.

Third, there is an emergence of neurogenetics research, which combines the use of neuroimaging techniques with genetic technologies (i.e., imaging genetics) to identify specific regions of the brain and the associated genes involved in autism spectrum disorders. According to scientists at the 2010 International Meeting for Autism Research (IMFAR), one way imaging genetics can move the field forward is to use brain imaging to identify more similar patient groups in which the genetic study can be performed. The goal of this emerging research is to identify the effects of candidate genes in the brain and potentially impact drug development (Scott-VanZeeland, 2010). Thus, the reclassification of ASD will be further implicated by emerging technologies, this time by functional characteristics of the brain, which will undoubtedly be matched to specific aspects of human behavior. The disciplinary emergence of neurogenetics as well as the clinical

implications of such technologies in classification of ASDs will be important areas of social scientific research in the future.

Since this study interviewed only families who participated in the Simons Simplex Collection (families with only one child diagnosed with an ASD), an important comparison should also be made with families of the AGRE database, who have more than one child diagnosed with an ASD. This comparison would shed light on the influence and understandings of the inheritance of ASD among families by addressing how parents and scientist view the multiplex vs. simplex models of genetic disease. It would also highlight social and cultural issues of participating in genetics research in the clinic versus the family's home. Further, it would further elucidate some of the ethical complexities of participating in a genetic database.

The cultural imaginaries of autism generated through different media will also be important areas of research in the future. Based on interviews conducted in this study, the media has served as a double-edged sword. Scientists acknowledged that the media often oversimplify messages of genetic research findings since the complexity of ASD is hard to capture. Scientists also felt there was a lack of balanced coverage about science that is being done and what people are claiming based on no science, such as coverage on the vaccine wars. Parents, on the other hand, felt the media did not represent the broad spectrum of ASD and only emphasized severe cases and negative aspects of autism. Thus, a content analysis of representations of ASD in the media and public discourses would be an additional social world to the broader project of understanding autism, especially autism genetics.

Perhaps one of the most pressing areas of future sociological research are the impacts of the proposed changes in the DSM-V to eliminate the diagnosis of “Asperger’s disorder” to be replaced with a certain level of severity among the “Autism Spectrum Disorders”. These changes have ignited severe scrutiny among the “Aspie” community and have major implications for the identities of people diagnosed or self-diagnosed with Asperger’s disorder. The proposed changes in the DSM-V are also a reflection of scientific utility rather than what might benefit individuals on the autism spectrum and their families. Thus, future sociological research within the context of the absence, emergence and future disappearance of the Asperger’s diagnostic label is warranted.

Related to this is the diagnosis of ASD itself and the implications for the sociology of diagnosis. In chapter four, I highlight the diagnostic uncertainties faced by parents, which is partly a result of the range of symptoms associated with a diagnosis of an ASD under the current diagnostic criteria (DSM-IVR). It is also related to the broad range of symptoms that are not part of the criteria, which parents believe are part of “the autism”. Furthermore, there are many levels of severity, which change over the course of a child’s life that further complicate the diagnostic odyssey. These issues combined with parental denial, the “invisibility” of the disorder to people outside of the family, and the levels of stigma attached to different diagnostic labels are areas of investigation that should be considered in the future.

Another area of future research based on the current study would be to consider the counter movements in the autism community against the biological or genetic understandings of ASD. It would be important to analyze perspectives of adults on the autism spectrum concerning biomedical research on autism in general, and more

specifically, the genetics research on autism. These alternative viewpoints compared to the current study would offer additional insights into the social world of autism spectrum disorders.

Finally, the parents and the scientists interviewed in this study reminded me of the huge disparities in diagnosis, treatment, and access to educational services. The access to genomic technologies and associated treatments, as well as representation in genetic research databases like the AGRE and SSC, are likely to reinforce these disparities. Future sociological research needs to address this major issue, as well as investigate the how genetic knowledge is conceived in families of different racial and ethnic backgrounds.

## APPENDIX A: INDIVIDUALS WITH ASD INTERVIEW GUIDE

The interviews will be conducted in English and in a location agreed upon by the interviewee. Each interview will take approximately 60 minutes. Prior to the interview, Ms. Singh will ask participants to complete a consent form and verbally reiterate her permission to audio record and write notes pertaining to the conversation. It will be made clear in both the informed consent document, as well as just prior to asking the questions that the participant can refuse to answer any questions and may stop the interview at anytime. If there is a question you do not want to answer say “PASS”. To begin the interview, Ms. Singh will introduce herself and will emphasize that she is a student researcher interested in learning the perspective of participants on how they view the topic of Autism (or Asperger syndrome). The following questions will be asked:

1. As a student researcher I am interested in learning about the opinions and understandings of Autism (or Asperger syndrome) from different perspectives. Since you know more about this topic than I do, can you tell me in your own words, what you know about autism (or Asperger syndrome) and how would you describe it? If you were to explain to somebody that doesn't know anything about autism? How would you define/describe it?
2. When you think about what you know about autism (or Asperger syndrome), from what people or what places do your ideas come from? Do they come from your parents? Doctors? Teachers? Friends? TV?
  - a. Follow-up: Which of these has been the most influential (or important) to you in understanding Autism (or Asperger syndrome)?
  - b. Follow-up: What do you think (agree or disagree) about what your (parent's, doctors, teachers, etc. think?)
3. The next few questions are about the diagnosis of Autism (AD). What are your first memories of being told that you had autism? How old were you when you knew for sure you had autism? What was your experience with the diagnosis and assessment process – if you remember?
  - a. Follow-up: At the time of your first memory of being told you had autism, what were your thoughts? Did it help answer any questions you had about yourself? Angry? Sad? Was it significant/non-significant?
  - b. Follow-up: What do you think about your diagnosis of Autism now?
  - c. Follow-up: Do you agree or disagree with the diagnosis?
4. How or when do you tell others that you are diagnosed with autism/AD?
  - a. Follow-up: Do you think the diagnosis of Autism/AD is a “label” good or bad?
  - b. Does the diagnosis of autism feel like a label for you?
5. Do you think people with HFA or AD should be treated differently whether it be at school or work or in the public?

- a. Based on your experience, do you think having a diagnosis of autism has made people treat you differently? Can you describe a situation when you, yourself remember being treated differently?
6. Some people think autism (or Asperger syndrome) is a genetic disorder, while others think it is caused by environmental exposures such as vaccines, others think it is another type of person. What do you think about these different views? How would you describe it (based on these views)?
7. Scientists are spending a lot of time trying to find genes that are involved with the symptoms and causes of autism (or Asperger syndrome). What do you know about this type of research?
  - a. Follow-up: What is your opinion about this type of research?
8. Are you aware of all the public information such as TV, newspaper, or books that talk about Autism (or Asperger syndrome) and if so, what do you think about all of it? Is it helpful for you? Completely misrepresented?
  - a. Follow-up: What would you like to see or read?
9. Is there anything else I have not asked you that you think is important for me to know as a new researcher in this area?
  - a. Follow-up: Do you have any advise that you would like to tell other researchers in this area?

## **APPENDIX B: PARENT INTERVIEW GUIDE**

This interview will be open-ended, allowing participants to articulate their own perspectives and experiences. The questions will serve to guide the interview and other questions may be asked in response to the participant's statements.

1. To start, please tell about your experiences of autism within your family.
  - a. Based on your experiences how would you describe autism spectrum disorder?
  - b. What do you think are the major factors that are causing autism?
2. As a parent with a child with autism, tell me about your involvement in raising awareness of autism spectrum disorder and/or participating in research activities such as fund raising or donation of biological materials?
  - a. How did you get involved in these activities and how long have you been involved?
  - b. Based on your experience, do you feel that families and advocates of autism have changed the direction of autism research in general and the research on the genetics of autism more specifically?
3. In your opinion, what should be some of the short and long term goals of autism research?
  - a. Do you feel there has been progress made in autism research in general and in autism genetics research more specifically?
  - b. What areas of research do feel would be most important for families with autism?
4. The next few questions are about the Autism Genetic Resource Exchange and the Autism Genome Project.
  - a. Are you aware of these two projects and if so how would you describe them?
  - b. Have you been involved in the AGRE and/or AGP and if so how?
  - c. In your opinion, has the availability of these resources changed the focus of autism research?
5. What other genetics research projects are you aware of?
6. What has been your experience with scientists conducting autism genetics research?
7. Based on your experiences with ASD, how would you describe the genetics of autism?
  - a. Did this understanding change after the diagnosis or treatment of autism in your family?
  - b. What do you believe will be the major impacts of genetics research on families with autism? (e.g., diagnosis, treatment, experiences with ASD)
8. What is your opinion of how the results of research on the genetics of autism are portrayed in the media? (i.e., TV, newspapers)



- a. Do you feel the media portrays families with autism accurately?
- 9. How would you like to see the future of autism research evolve?
- 10. How would you like to see the future of autism genetics research evolve?
- 11. Is there anything else that you think I should know or that you would like to tell me?

## APPENDIX C: SCIENTIST INTERVIEW GUIDE

This interview will be open-ended, allowing participants to articulate their own perspectives and experiences. The questions will serve to guide the interview and other questions may be asked in response to the participant's statements.

1. What is the scientific discipline of your research?
  - a. How long have you been in this area of research?
2. How and why did you get involved in autism research?
3. Do you see yourself as an activist in this arena or an advocate for autism research?
4. Now I would like to ask you specifically about autism genetics research.
  - a. How would you describe the research you do on the genetics of autism? What are the short and long term goals of your research?
  - b. How long have you been doing autism genetic research?
  - c. What percentage of your time is dedicated to research on the genetics of autism? How has this changed since you started doing research on autism?
  - d. In your opinion, how has your research contributed to the understanding of the genetics of autism?
5. What specific technologies and tools do you utilize in order to conduct your research?
6. The next few questions are about the Autism Genetic Resource Exchange and the Autism Genome Project.
  - a. In what ways have you been involved in the AGRE and AGP?
  - b. In your opinion, has the availability of these resources changed the focus of autism research?
  - c. How have these resources specifically affected your own research on the genetics of autism?
7. In your experience, how has the involvement of families and advocates of autism changed the direction of autism research in general and the research on the genetics of autism more specifically?
8. What is your opinion on how the results of research on the genetics of autism are portrayed in the media? (i.e., TV, newspapers)
  - a. If applicable, do you feel that your research has been represented accurately in the media?
9. How do you see the future of autism genetics evolving in general and in your own research more specifically?

10. Is there anything else that you think would be important for me to know or that you would like to tell me?

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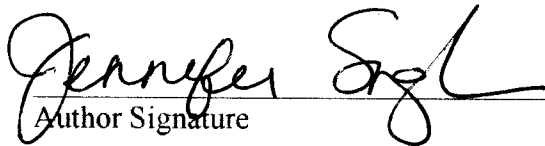
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