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An Assessment of the Use and Impact of Genetic Testing Among Deaf Adults and Parents of Deaf Children

THESIS

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by

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DEDICATION

To

My family, thank you for continuing to believe in me.

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ABSTRACT OF THE THESIS

An Assessment of the Use and Impact of Genetic Testing Among Deaf Adults and Parents of Deaf Children

by
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Master of Science in Genetic Counseling
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Hearing loss is a common human sensory disorder affecting millions globally, and many cases are due to a genetic etiology. Genetic testing for hearing loss is evolving as new genetic tests and genes related to hearing loss emerge; however, there has been limited research on how testing for hearing loss is impacting individuals. The purpose of this study was to assess how genetic testing for hearing loss influences decision making regarding medical management, language development, education, family planning and spouse selection by deaf adults and parents of deaf children as well as the impact on their emotional well-being. An anonymous online survey was created to assess experiences with genetic counseling and genetic testing for hearing loss. Study participants were recruited through online national organizations and social media advocacy groups, and 84 responses were analyzed. Hearing parents of deaf children were more likely to make post-genetic testing decisions regarding their children's medical care and language development than deaf parents did for their children or themselves. Family planning decisions were made less frequently, but there were still 31% of deaf adults and 36% of parents of deaf children whose family planning was influenced by their test results. Individuals with, and parents of children with syndromic deafness were significantly more likely to make medical and family planning decisions after genetic testing than were participants or parents of children without a syndrome. Genetic testing revealed a diagnosis more frequently in deaf children than deaf adults, and deaf children also received genetic counseling more often than deaf adults.

Respondents were more likely to have an overall positive affect than a negative or neutral affect post-genetic testing on the Positive and Negative Affect Schedule (PANAS). The results of this study illustrate the diversity of impact that genetic testing for hearing loss has on individuals and families and highlights the need for more exploration into the utilization of genetic test results as more genes and genetic tests for hearing loss emerge.

I. INTRODUCTION

1.1 Background and Significance of the Research

1.1.1 Defining Hearing Loss

Hearing loss is the most common human sensory disorder, affecting over 5% of the global population, or 466 million people, according to the World Health Organization (World Health Organization 2020). There are many different ways to define hearing loss from an audiologic standpoint that can be based on the age of onset, type, laterality, stability and degree of hearing loss. It is important first to delineate the differences between audiologic definitions for an individual who is hard-of-hearing versus an individual who is deaf. An individual who is hard-of-hearing is someone who has mild to severe hearing loss but will usually have some residual hearing (Shearer *et al.* 2017). An individual who is deaf has little to no residual hearing, regardless of their cultural affiliation to the Deaf community (Alford *et al.* 2014, HLAA 2020). In the Deaf community, persons who are deaf do not typically use oral language, whereas individuals who are hard-of-hearing usually have some oral language (Shearer *et al.* 2017). These definitions pertain to the auditory phenotype and do not include other factors related to an individual's identity, including cultural affiliation or preferred method of communication or language.

There are many different factors involved in the classification of hearing loss. One of the primary considerations is the age at which the hearing loss began. Hearing loss that is present at birth is termed congenital, while hearing loss that presents after the newborn period but before or after the typical age of language development is called prelingual or postlingual, respectively. It is also defined based on the type, which focuses on the physical location causing the hearing loss

within the pathway of sound. Types of hearing loss include conductive, sensorineural, mixed and central auditory dysfunction. Conductive hearing loss occurs when there is a mechanical disorder caused by an abnormality in the outer or middle ear structures, such as the external auditory canal or the ossicles. Sensorineural hearing loss occurs if there is damage to the inner ear structures, such as the auditory nerve or the cochlea, which is the spiral-shaped structure that converts mechanical vibrations into nerve impulses. Mixed hearing loss is a combination of conductive and sensorineural hearing loss. The final type of hearing loss is central auditory dysfunction resulting from damage to the eighth cranial nerve, the auditory brain stem or the cerebral cortex (Shearer *et al.* 2017). Hearing loss may be unilateral (occurring only on one side) or bilateral (occurring on both sides). It can have variation in stability and be defined as progressive (becomes more severe over time) or nonprogressive (remains stable over time), or it can fluctuate. Finally, we define hearing loss according to the degree of loss on a decibel (dB) scale, which ranges anywhere from slight loss (16-25 decibels) to profound loss (91 decibels or greater), with other definitions in between. The threshold for each frequency starts at 0 dB and is compared to the level at which the average young adult is able to hear a particular tone 50% of the time (Shearer *et al.* 2017; Kochhar *et al.* 2007). Hearing is considered normal if an individual's thresholds are within 15 dB of normal thresholds (Shearer *et al.* 2017).

In the United States, approximately 2 to 3 of every 1,000 children are born with a detectable level of hearing loss (CDC 1999-2007). The majority (~90%) of infants born with hearing loss are born to hearing parents (Mitchell & Karchmer 2004). According to the Survey of Income and Program Participation (SIPP), which regularly collects data identifying persons with hearing loss, approximately 10 million Americans identify as hard-of-hearing, and nearly one

million are considered functionally deaf (Mitchell 2006). Overall, the prevalence of hearing loss increases in the older age brackets, with age being the strongest predictor of hearing loss among adults aged 20-69 (Hoffman *et al.* 2016).

Although this research study focuses on the potential genetic factors involved in hearing loss, the etiology can include genetic, environmental or acquired factors. Due to the wide range of causes, identifying an etiology for hearing loss can be a complex and extensive process. There are a variety of genetic causes including variants in a single gene or in multiple genes as well as chromosome abnormalities (detailed below). Acquired causes can be markedly variable and can lead to conductive, sensorineural or mixed hearing loss. Conductive hearing loss may be due to fluid accumulation in the middle ear, foreign objects inserted into the ear canal, impacted earwax in the external auditory canal, allergies, a ruptured tympanic membrane, auditory canal atresia or stenosis, or abnormalities of the ossicles. Sensorineural hearing loss can be caused by exposure to excessive and repetitive noise damage, side effects of ototoxic medications, combined effects of aging (also called presbycusis), head trauma, auditory tumors or impact due to an explosion. Both conductive and sensorineural hearing loss may be a result of prenatal or postnatal infections, such as toxoplasmosis, rubella, cytomegalovirus, herpes and bacterial meningitis (Kochhar *et al.* 2007; Shearer *et al.* 2017). The significance of understanding the genetic causes of hearing loss continues to increase as the rates of acquired hearing loss decrease due to advances in both medical treatment and prevention (Kochhar *et al.* 2007).

1.1.2 Deaf Identity

When discussing individuals with hearing loss, it is crucial not only to detail the audiologic and clinical identifiers but also to discuss the diversity and fluidity of deaf identity. The term deaf (lowercase "d") is used in this thesis to describe an individual who is deaf or hard-of-hearing—referring to the audiological condition of being unable to hear—and does not presume the cultural affiliation or preferred method of communication of that person. The term Deaf (capital "D") refers to an individual who identifies as part of the Deaf community – individuals who share a common language (typically American Sign Language in the United States) and culture (Padden & Humphries 1988). A person who identifies as hard-of-hearing may have hearing loss ranging from mild to moderate and often has some residual hearing, but the term may also be used to describe someone who is deaf but does not identify culturally with the Deaf community. Identities are personal to every individual and may be influenced by a variety of experiences, including their level of hearing, hearing status of their parents, educational background, age of onset of hearing loss, method of communication, and cultural identity (National Association of the Deaf 2020). Although terms such as "hearing impaired" have been used to define an individual with hearing loss, it is important to recognize the negative connotations that can be associated with such terminology and that this term is no longer accepted by most in the Deaf community. Furthermore, terms such as "deaf and dumb" or "deaf-mute" are considered to be offensive, are highly inaccurate, and should never be used to describe a person who is deaf.

1.1.3 History of Discrimination

Through various advances in medical genetics, many individuals and families have been provided with explanations for their deafness. Although these genetic discoveries are perceived by some as exciting pieces of information meant to empower individuals, there is also some skepticism and caution within the Deaf community surrounding the genetics field due to a history of broader societal prejudice against deaf persons. To fully understand the concern and antipathy that some deaf individuals have toward medical genetics, we must address the history of discrimination that has occurred over the past 150 years. Unfortunately, there are many examples of intolerance and bias against deaf and hard-of-hearing people. By the end of the American Civil War, there was a push for "oralism" in schools in order to eliminate the use of sign language and reinforce the use of speech, lipreading, and written English (Baynton 1996). Alexander Graham Bell was a major proponent of oralism and even published a paper entitled, "Upon the Formation of a Deaf Variety of the Human Race" that detailed his concern that socialization and marriage among deaf people would lead to the creation of a "deaf race" (Bell 1883; Arnos 2002). These were some of the ideologies that fueled the eugenics movement, which sought to improve the human species by "breeding out" undesirable characteristics, disabilities and disease. In America, this movement led to marriage prohibition by both federal and state governments as well as forced sterilization and abortion of anyone deemed unfit to procreate, which included individuals with intellectual disabilities, mental illnesses, "undesirable" traits such as deafness, certain ethnic backgrounds such as Native Americans, and those in poverty (Arnos 2002; Norrgard 2008; Rivard 2014). The eugenics movement was not confined only to the United States, and during World War II, the concept of a superior race led to the mass killings and forced sterilizations of individuals viewed as inferior, including Jewish

people and those with mental or physical disabilities, including deaf individuals (Padden & Humphries 1988; Biesold 1999; Arnos 2002). Due to the atrocities committed against deaf individuals in the past, many still hold valid concerns that genetics research and testing could devalue deaf people or that it could be used to try to "cure" deafness (Martinez *et al.* 2003; Middleton *et al.* 1998). Despite the fact that there have been policies and movements in the United States that discriminated against deaf individuals, there also have been several federal civil rights laws starting in the 1970's that have played a part in creating equal opportunities and rights for individuals with hearing loss (Civil Rights Laws, 2020); some of these include the Individuals with Disabilities Education Act (IDEA), the Rehabilitation Act of 1973, and the Disabilities Act of 1990 (Civil Rights Laws, 2020). It is valuable for us to understand the history of discrimination against deaf individuals and the Deaf community in order to appreciate any wariness that they may have about medical genetics and the medical community as a whole.

1.1.4 Genetics of Hearing Loss

Hearing loss is an etiologically heterogenous disorder, which presents certain challenges in identifying a genetic diagnosis for many deaf and hard-of-hearing individuals. There are currently more than 400 known genetic syndromes that include hearing loss as a feature and over 120 genes associated with non-syndromic hearing loss (NSHL) (Shearer *et al.* 2017; Hereditary Hearing Loss Homepage 2020). There can be different types of inheritance patterns depending on the genetic etiology, including autosomal recessive, autosomal dominant, X-linked, and mitochondrial. Identifying a genetic etiology for hearing loss in an individual or a family can have direct effects on clinical management, prognosis, and the chance of recurrence. However, as is the case for many genetic conditions, individuals who pursue genetic testing do not always

receive a genetic diagnosis for their hearing loss, either because their hearing loss does not have a genetic cause or because of limitations in the testing technology used to identify the specific genetic cause.

We can separate genetic hearing loss into two classifications: syndromic and non-syndromic. Non-syndromic hearing loss occurs when there are no other associated features in any other organs or body systems, including malformations of the outer ear. Approximately 70% of hearing loss with a known genetic etiology is considered to be non-syndromic (Shearer *et al.* 2017). Of disorders with prelingual non-syndromic hearing loss, the majority (80%) are autosomal recessive, 20% are autosomal dominant, and 1%-1.5% are X-linked, mitochondrial, or due to other forms of inheritance, whereas most disorders with postlingual non-syndromic hearing loss are autosomal dominant (Shearer *et al.* 2017; Smith *et al.* 2005; Alford *et al.* 2014). Roughly half of cases of severe-to-profound prelingual non-syndromic hearing loss are due to autosomal recessive pathogenic variants in the *GJB2* gene located on chromosome 13q12 (Smith & Jones 2016). The *GJB2* gene encodes for the gap junction beta 2 protein and is commonly referred to as Connexin 26 (Snoeckx *et al.* 2005). Non-syndromic hearing loss may be referred to by the specific gene involved or by the genetic locus, which refers to the physical location of a gene or DNA sequence along the chromosome. For example, the *GJB2* and *GJB6* genes encoding Connexin 26 and Connexin 30, respectively, are located in the DFNB1 locus (Kelsell *et al.* 2017). In this system of nomenclature, the first three letters (DFN) refer to deafness, the fourth letter denotes the mode of inheritance, with DFNA indicating autosomal dominant, DFNB indicating autosomal recessive, and DFNX indicating X-linked inheritance, and the number indicates the order of discovery of the locus; genetic loci are often defined before a particular

causative gene within the locus is identified (Shearer *et al.* 2017). This nomenclature came about as a result of the order of discovery of non-syndromic hearing loss loci that were then grouped according to pattern of inheritance (Vona *et al.* 2015). Although the majority of prelingual non-syndromic hearing loss is autosomal recessive, there have been more than 25 genes associated with autosomal dominant non-syndromic hearing loss, and this mode of inheritance is more common in individuals with postlingual deafness (Shearer *et al.* 2017). For cases of mitochondrial inheritance—which are maternally inherited, occur due to variants in the mitochondrial DNA, and do not have DFN reference numbers—there is typically moderate-to-profound hearing loss, primarily due to pathogenic variants in either the *MT-RNR1* or the *MT-TS1* gene, and severe-to-profound deafness due to pathogenic variants in *MT-CO1* (Shearer *et al.* 2017). Although this is the least common mode of inheritance for hereditary hearing loss, it is worth noting due to the association with pathogenic variants in *MT-RNR1* and *MT-CO1* with aminoglycoside ototoxicity (Shearer *et al.* 2017). Aminoglycosides are a group of antibiotics that can produce free radicals that lead to damage of the inner ear and can result in permanent hearing loss, especially in individuals with one of these pathogenic variants in mitochondrial DNA (Selimoglu 2007). Since these genes are located within the mitochondrial genome, they are subject to the effects of heteroplasmy, which is defined as the presence of more than one type of mitochondrial DNA within a single cell (Shearer *et al.* 2017). Therefore, different individuals with a pathogenic variant in the same mitochondrial gene can have different degrees of severity and ages of onset, even within the same family.

Syndromic hearing loss accounts for approximately 30% of cases of prelingual genetic deafness and is defined as having additional clinical features present, such as malformations of

the external ear and/or other organs or organ systems (Shearer *et al.* 2017). Similar to non-syndromic types of hearing loss, syndromic forms are categorized according to mode of inheritance, including autosomal recessive, autosomal dominant and X-linked (Allen & Goldman 2020). Examples of more common syndromes associated with autosomal recessive inheritance include Usher syndrome, which also involves a progressive loss of vision, Pendred syndrome, which also involves thyroid dysfunction, and Jervell and Lange-Nielsen syndrome, which includes cardiac arrythmia and may lead to sudden death. Usher syndrome is the most common type of autosomal recessive syndromic hearing loss and affects over 50% of all the deaf-blind individuals in the United States (Allen & Goldman 2020).

Examples of more common autosomal dominant syndromic forms of hearing loss include Waardenburg syndrome, which also involves pigmentation abnormalities of the hair, eyes, and skin, Treacher-Collins syndrome, which affects the development of the bones and tissues in the face, Stickler syndrome, which includes orofacial, ocular and skeletal features, and Branchio-Oto-Renal syndrome, which also includes abnormalities of the external and middle ears, the kidneys, and second branchial arch development (Allen & Goldman 2020; Koffler *et al.* 2015). There are also syndromes associated with hearing loss that have X-linked inheritance or even multiple forms of inheritance; Alport syndrome is the most common example and can be inherited in autosomal recessive, autosomal dominant, or X-linked fashion. Individuals with Alport syndrome have progressive sensorineural hearing loss, progressive renal insufficiency that can lead to end-stage renal disease if untreated, and ophthalmologic findings (Kochhar *et al.* 2007). Given the challenges that can arise in distinguishing among syndromic and non-syndromic forms of hearing loss due to overlapping or variable clinical features, individuals may

be referred to a medical geneticist to be evaluated if an etiology is not already known or for ongoing management of a known syndrome.

1.1.5 Management of Hearing Loss

Due to the Early Hearing Detection and Intervention (EHDI) process for newborns, management and treatment of hearing loss often begins shortly after birth or in early childhood, which maximizes communication, language development and literacy in children who are deaf or hard-of-hearing (Joint Committee on Infant Hearing 2007, EHDI 2019)[1]. Once a cause for hearing loss is identified, there are different ways in which the hearing loss and/or associated clinical features are managed. For individuals with non-syndromic hearing loss, the primary focus is the development of communication and language, particularly in infants and young children. Even if a specific genetic etiology is unknown, once hearing loss is identified, it is important to create a plan for effective communication, which can vary from family to family depending on their values, cultural affiliation, access to resources, and the degree of hearing loss. Some families will consider using assistive hearing devices, such as hearing aids or cochlear implants (Shearer *et al.* 2017). Hearing aids are amplification devices that magnify sound vibrations entering the ear and can improve speech comprehension, particularly for persons with damage to sensory cells in the inner ear (Hearing Aids, 2018). Cochlear implants consist of electronic devices surgically implanted under the skin behind the ear and electrodes attached to the cochlea that bypass the damaged portion of the ear and directly stimulate the cochlear nerve by converting sound waves to electrical impulses that are transmitted to the implant. This type of

[1] Early Hearing Detection and Intervention (EHDI), first authorized by Congress in 2000, refers to the practice of screening every newborn for hearing loss prior to hospital discharge. Infants not passing the screening receive diagnostic evaluation before three months of age and, when necessary, are enrolled in early intervention programs by six months of age. All 50 states and the District of Columbia have Early Hearing Detection and Intervention (EHDI) laws or voluntary compliance programs that screen hearing.

assistive hearing device provides individuals with severe to profound hearing loss the ability to hear sound but does not restore typical hearing. Which type of assistive hearing device to use is a very personal choice and often requires a great deal of discussion and consideration before making a final decision. This is particularly challenging for parents who have to make decisions for their children when they are too young to be involved in the decision-making process themselves and who may later disagree with the decision that was made. Hearing assistive devices are utilized for all types of hearing loss, including acquired, non-syndromic and syndromic.

In individuals with syndromic hearing loss, there will still be a plan to address the hearing loss, but it often will differ from non-syndromic hearing loss because these individuals may also require clinical management of their associated features. It is important to note that some individuals initially thought to have non-syndromic hearing loss may actually be found later to have syndromic hearing loss, which can delay medical management of the non-audiologic features. This can occur for genetic syndromes in which the other features have later ages of onset or are progressive, such as vision loss in Usher syndrome. For syndromic hearing loss, there will likely be recommendations for additional clinical follow-up, such as referrals to appropriate medical specialists, alterations to or initiation of a clinical management plan, and testing and/or treatment for any associated features of that particular syndrome. A variety of health professionals may be involved in the care of an individual with hearing loss, including but not limited to an otolaryngologist, audiologist, speech and language specialist, medical geneticist, genetic counselor, early intervention specialist, and other medical specialists, depending on the presence of additional clinical features. Management of hearing loss for some

families may not include any assistive hearing devices and instead may focus on developing communication through other methods, such as lipreading or signed English or American sign language (ASL). Although many of the basic signs are shared between signed English and ASL, they are different languages with their own grammar, syntax and vocabularies. Signed English is the translation of English through ASL signs, modified ASL signs, and fingerspelling utilizing English grammar rules (Stephenson & Wolkow 2020), while ASL is its own unique language created for deaf people that uses signs to represent concepts and phrases rather than being a direct English translation (Kelly & Benedict 2020). It is important to note that communication management will be different for each family depending on their own experiences and values surrounding hearing loss.

1.2 Purpose of Study and Specific Aims

1.2.1 Growing Interest in Genetics and Genetic Testing

Attitudes of deaf and hard-of-hearing individuals toward the medical genetics field and genetic testing have fluctuated over time. Genetic testing for hearing loss raises both ethical and social concerns, especially among those who consider deafness to be a cultural trait rather than a medical one (Martinez *et al.* 2003). A relatively early study with respect to testing that asked 87 deaf individuals in the United Kingdom their attitudes toward genetics reported that 55% thought that genetic testing would do more harm than good, and 46% believed that its potential use would devalue deaf people (Middleton *et al.* 1998). Particularly for those who do not perceive deafness as a medical condition or disability, the practice of genetic testing and genetic counseling may feel like a threat to Deaf culture and its set of beliefs. However, later studies have shown a growing interest in genetic testing and the possible benefits of receiving a genetic diagnosis for hearing loss (Dagan *et al.* 2002, Burton *et al.* 2006; Withrow *et al.* 2009).

According to a study conducted in 2003, 62% of deaf or hard-of-hearing individuals would allow genetic testing of their own newborn (Martinez *et al.* 2003). There has been some research analyzing the ethnic differences in how parents perceive genetic testing. In a study published in 2008 that enrolled parents of deaf and hard-of-hearing children and children referred for additional hearing screening, Asian and Hispanic parents were more likely than Caucasian parents to view family planning, medical care and helping the family as important reasons for pursuing genetic testing (Palmer *et al.* 2008). All parents from this study viewed genetic testing as beneficial for learning the cause of their child's deafness (Palmer *et al.* 2008). As the interest in genetic testing increases along with the ongoing discovery of novel genes associated with hearing loss, providers will have to be prepared to discuss the complexities of testing with their patients and to aid in informed decision making.

1.2.2 Motivations for Pursuing Genetic Testing

Genetic information can be empowering for patients and their families, including those who are deaf or hard-of-hearing. The pursuit of a genetic etiology for hearing loss can be motivated by an abundance of factors that are influenced by the experiences, beliefs and circumstances of each person. For some, a genetic diagnosis comes as a relief to finally have an explanation for their child's or their own deafness. The information itself can benefit their emotional well-being by reducing anxiety and fear of the unknown while empowering them with knowledge of the cause of their hearing loss (Burton *et al.* 2006). For some, the knowledge of a genetic cause can alleviate guilt associated with hearing loss diagnosed in a child, particularly for those who may have incorrectly believed that something they did during pregnancy caused their child's deafness (Withrow *et al.* 2008). It can provide helpful information in terms of what to expect, particularly if it is progressive or if it is related to a syndrome with other clinical features.

Some individuals or couples may be motivated to use the information to prepare for the future or to know the chance for deafness to occur in their family members. They may use the diagnosis to prepare for having a deaf child, to know the chance that they could have deaf children, or to inform relatives that they also may have an increased chance to have a deaf child (Withrow *et al.* 2008). For instance, if a genetic variant that is determined to be the cause of their hearing loss is identified in a parent and is inherited in an autosomal dominant fashion, then there would be a 50% chance of each of their children inheriting the same deafness-associated variant. Additionally, genetic services, such as genetic counseling, provide the opportunity to discuss empiric risks, even if a specific etiology cannot be identified. For example, a hearing couple with one child with non-syndromic deafness and no family history of hearing loss would have an ~18% empiric probability that a future child would also be deaf (Shearer *et al.* 2017). It is also important to parents that they feel prepared not only to have a deaf child but also to have a plan for their child's educational and language development (Withrow *et al.* 2008; Burton *et al.* 2006).

In cases of syndromic hearing loss, there are additional benefits to having a genetic diagnosis. Using the example of Usher syndrome, which results in progressive blindness in addition to hearing loss, knowing that these individuals will have vision loss over time could allow them to prepare for it. They may choose to engage in learning other communication methods, such as Braille or tactile signing, and to work on establishing a strong support network. Other syndromes may require closer medical monitoring, additional testing, or referrals to other specialists. Another example is Jervell and Lange-Nielsen syndrome, which is known to have associated cardiac arrhythmias and a risk for sudden death in those affected, so it would be

important to have ongoing cardiac monitoring. Thus, parents and families may be even more motivated to pursue a genetic diagnosis if there could be other associated features that should be identified and managed as early as possible.

While considering the possible motivations for pursuing genetic testing, it is important to recognize that there are many people who feel very differently and would not agree with them. Although many are interested to know why they or their family members are deaf, for some there is little to no interest in utilizing this information to make decisions about future children or a partner (Boudreault *et al.* 2010). Some studies have shown that cultural affiliation tends to be a strong factor in determining people's motivations for genetic testing (Boudreault *et al.* 2010). Therefore, every individual and situation should be approached without assumptions surrounding their feelings toward genetics research and genetic testing.

1.2.3 Limited Research on the Impact of Genetic Testing in Deaf Individuals

Although there has been research regarding the motivations and attitudes of culturally and nonculturally deaf and hard-of-hearing individuals toward genetic testing, there is a paucity of research examining the effects after testing has been done. Some research has assessed comprehension of genetic test results and their impact on deaf identify in deaf adults (Palmer *et al.* 2014). Another study analyzed attitudes and beliefs of culturally hearing parents—those who identify as being part of "mainstream hearing" culture— of deaf and hard-of-hearing infants (ages 0-3) after genetic counseling (Palmer *et al.* 2009). According to this study, parents felt that diagnostic genetic test results should be accompanied by genetic counseling to promote comprehension of the results. It is important to note that this prior research focused specifically

on *GJB2* (Connexin 26) and *GJB6* (Connexin 30) genetic testing. To date, there has not been a study published that analyzes the impact of genetic testing on decision-making for deaf adults and parents of deaf children.

Research conducted in 2009 examined the perceived impact that genetic testing may have on parents of children with hearing loss and deaf adults at Gallaudet University, which is a private university established specifically to educate those who are deaf and hard-of-hearing (Withrow *et al.* 2009). This study asked participants if they felt genetic testing would affect their lives or their children's lives in a variety of ways, including learning the etiology, understanding recurrence, knowing about related medical conditions or treatment, or affecting their choice of spouse or future children (Withrow *et al.* 2009). The largest perceived potential impact of genetic testing, according to 72.1% of parents of deaf children and 65% of deaf adults, was learning the cause of their hearing loss (Withrow *et al.* 2009). This study highlights the need for those who have already had genetic testing to determine whether or not these perceived impacts remain true for these groups. Another study by Palmer *et al* in 2013 surveyed deaf adults' perception of their personal control, anxiety and depression following genetic testing for *GJB2* and *GJB6* (Palmer *et al.* 2013). This study concluded that genetic testing in deaf adults does impact their psychological well-being for the three emotional measures mentioned and that receiving a genetic diagnosis may enhance self-knowledge (Palmer *et al.* 2013). The data surrounding the impacts of genetic testing and genetic counseling on deaf and hard-of-hearing persons remains limited, and additional research surrounding the effects on individuals and families is needed.

1.2.4 Significance of Research

To fully appreciate the significance of the present study, it is important to recognize the history of the Deaf community, the different causes of hearing loss, and the process that each family goes through regarding the identification, evaluation and management of hearing loss. Some studies have even suggested that Early Hearing Detection and Intervention (EHDI) newborn hearing screening be paired with molecular diagnosis for newborns who are identified as having hearing loss, which would vastly expand the impact of genetic testing on parents of deaf and hard-of-hearing children (Pandya 2016). One of the guidelines noted in the 2007 position statement by the Joint Committee on Infant Hearing states that infants with confirmed hearing loss should be offered a medical genetics consultation (American Academy of Pediatrics 2007). Additional research has found that many parents prefer a medical genetics evaluation, and that testing should be available and take place immediately or within a few months after an audiologic diagnosis (Parker *et al.* 2000; Withrow *et al.* 2008). As clinical genetics services continue to expand, understanding the effects of these services and genetic testing on deaf patients and families will be increasingly important.

One of the primary roles of genetic counselors is to translate complex genetic information to patients and families in a way that is effective, clear and culturally sensitive. Due to the complex and diverse beliefs and values of individuals who are deaf and hard-of-hearing, genetic counselors are particularly well suited for these situations. Genetic counselors are well-equipped to evaluate a family history of hearing loss, understand complex risk figures and the value of a correct diagnosis, help families make informed decisions, and offer information regarding support services (Arnos *et al.* 1991). Studies have found that deaf individuals and parents of deaf

children believe that a genetic counselor or a medical geneticist would be the most appropriate professional(s) to provide these services and to discuss genetic testing results (Withrow *et al.* 2009). If genetic counselors become more involved in these cases, it will be critical for them to understand the effects their services may have on families, not only through knowledge of research on the impacts of genetic testing but also through cultural sensitivity workshops to fully appreciate the complexity involved (Enns *et al.* 2010).

1.2.5 Purpose of Study and Study Specific Aims

The aim of this research is to assess the effects of genetic testing for hearing loss on deaf individuals and parents of deaf children. This study will use the term "deaf" to be inclusive of both deaf and hard-of-hearing individuals. Identifying the kinds of potential impact that this type of genetic testing can have on persons who are deaf or hard-of-hearing is important with respect to how providers may choose to educate and counsel these families. In order to serve deaf and hard-of-hearing patients better, we must appreciate the influence and consequences that the results of genetic testing can have on them. The broader goal of this study is to see how genetic test results are utilized by those who had genetic testing for hearing loss. The sub-categories of this broader goal include assessing the impacts of genetic testing and genetic counseling on each individual's or parent's decision-making surrounding their medical care, family planning, partner selection and language or educational development. This study also assesses the effects of genetic testing for deafness on emotional well-being after results were received. A secondary aim of the study is to determine whether deaf and hard-of-hearing adults experienced different effects after genetic testing than parents of deaf and hard-of-hearing children. Ultimately, this study seeks to gain insight into the experiences of individuals and families who have had genetic

testing for hearing loss in hopes that these results can provide guidance for genetic counselors and medical geneticists. The more we understand about the experiences of deaf and hard-of-hearing families, the more we as providers can continue to improve them.

II. METHODS

2.1 IRB Approval

The University of California, Irvine Institutional Review Board (IRB) Human Research Protections Exempt Self-Determination Tool permits that this research is exempt from IRB review under categories 2i and 2ii. This distinction requires that all study participant information is obtained in a manner in which the identity of the human subjects cannot be readily ascertained and that any disclosure of the human subjects' responses outside this research would not reasonably place the subjects at risk (Appendix A).

2.2 Study Design

2.2.1 Anonymous Online Survey

An anonymous online survey was created using the Research Electronic Data Capture REDCap® software, available through the University of California, Irvine. A unique survey link at https://is.gd/deafgenetictesting was created and publicly distributed so that responses could not be linked to the identity of the participant. The survey did not include any questions that could link participants' identities to their survey responses, and no HIPAA protected identifying information or contact information was requested in the survey in order to maintain participant privacy. Participation in this study was entirely voluntary, and responses were categorized by a research identification number based on the date and time of the response. All study information and questions were in English, so basic literacy in written English was required to participate.

This survey included 53 total questions consisting of multiple choice, short response and free response (Appendix B). With the use of skip logic technology, the total number of questions that appeared for each participant was dependent on how they answered the questions at the start of the survey. For example, if participants answered that they did not have children, they would not be asked any questions regarding having a deaf child. Of the 53 total questions, the majority were multiple choice, with a few questions asking the participants to type in a number (i.e., age, number of children) and a free response question at the very end. The questions pertaining to the emotional well-being of participants post-genetic testing were on a Likert scale ranging from "very slight to not at all" to "extremely." The survey was estimated to take each participant approximately 10-15 minutes to complete, and there were no monetary incentives for participating in this study. Participants were unable to skip certain questions that asked important demographic information, such as hearing status, but could skip questions pertaining to their emotional well-being and a free response question asking about their overall experiences if they did not wish to answer. All study questions were developed and adapted by the research team, except for the questions asking about the emotional well-being of the study participants after they received theirs or their child's genetic test results. In order to assess participants' emotional well-being after having genetic testing, these questions utilized the Positive and Negative Affect Schedule (PANAS), a validated survey tool (Watson *et al.* 1988). The PANAS tool was created to provide a consistent scale by which to measure positive and negative affect after an event. All responses were recorded in REDCap®, including incomplete or partial survey responses.

Informed consent for this survey was detailed on the first page of the online survey (Appendix B). Participants provided informed consent by reading this initial page and then

continuing to the next page of the survey. This information page also contained detailed information regarding the purpose of the study, risks and benefits, eligibility criteria, how information would be collected and stored, and contact information for the lead researcher. Participants were able to exit the survey at any time and could choose to skip non-demographic questions they did not want to answer.

2.2.2 Study Population and Recruitment

The target population for this study was deaf adults, with or without deaf children, and hearing or non-hearing parents of children who are deaf and who have had genetic testing for hearing loss. Participants were not eligible for the study if they met any of the following exclusion criteria: under 18 years of age, not living within the United States, not deaf or hard-of-hearing themselves and do not have a child who is deaf. Participants were not excluded based on the degree or severity of hearing loss or type of genetic testing performed.

Participants were recruited through advocacy groups, online organizations and social media pages. National and local organizations were contacted by email with a description of the research, study aims, and a survey link with a request to distribute the study survey to their member listservs. Several organizations agreed to post a short description of the study along with the survey link on their research homepage to increase study exposure. In addition to recruitment through larger organizations, the study link was posted on social media pages and advocacy group pages along with information about the study and the lead researcher's contact information for questions. The survey was open from October 23rd, 2020, through March 31st, 2021.

2.3 Data Analysis: Survey

Survey data was exported from REDCap® into a password protected file in Microsoft Excel and then was subsequently analyzed using IBM Statistical Package for the Social Sciences (SPSS) software version 26 (IMB SPSS Statistics for Mac, Armonk, N.Y., USA). Respondents' characteristics were summarized using total counts (N) and percentages for all categorical variables. Chi-square analysis and Fisher's exact test (used only when expected cell counts were <5) were used to test for statistical significance between groups; a difference was considered statistically significant if $p < 0.05$.

The Positive and Negative Affect Schedule tool was used in this study to analyze the emotional well-being of participants after receiving their genetic test results. This tool requires respondents to self-report their measure of affect for ten positive and ten negative emotions (Watson et al., 1988). The total score for each participant was calculated by combining the sum of the positive items and subtracting the sum of the negative items. A score greater than zero indicates a more positive affect after receiving genetic test results, while a score less than zero indicates a more negative affect.

III. RESULTS

3.1 General Survey Demographics

A total of 95 participants began the survey, 11 of whom did not respond to any survey questions and were excluded from the data analysis, leaving a study population of 84. Table 1 provides a summary of the general demographic characteristics of the participants, including their age, gender, ethnicity, hearing status and community affiliation. The majority of the respondents were between 30 and 49 years (73%) with a median age from 30 to 39 years, female (89%), Caucasian (87%) and identified as hearing (64%). Respondents who did not identify as female included eight males and one non-binary person. Individuals who did not identify as Caucasian included one who identified as African American or Black, four who identified as Asian, three who identified as Hispanic and four who selected "other" in this category. Within the total sample population, 19% identified as being hard-of-hearing, and 17% identified as being deaf. Regarding community affiliation, many participants identified as part of the Deaf community (69%), but only 13% identified as being a part of the hearing community. Participants could choose to identify with both the Deaf and hearing communities in the survey, and these choices were not mutually exclusive. Demographic categories and answer choices were created to aid data analysis; however, these categories and single response limitations may not be inclusive of all groups, particularly for the gender and ethnicity questions. Respondents were required to select an answer to all general demographic characteristic questions (Table 1) to continue the survey.

Table 1. General Demographic Characteristics of Respondents

Characteristic	N=84	%
Age (categories)		
20-29	10	12
30-39	35	42
40-49	26	31
50-59	2	2
60-69	6	7
70-79	4	5
No Response	1	1
Gender		
Female	75	89
Male	8	10
Non-Binary	1	1
Ethnicity		
Black or African American	1	1
Asian	4	4
Caucasian	73	87
Hispanic	3	4
Native American	0	0
Native Hawaiian or Other Pacific Islander	0	0
Other	3	4
Respondent Hearing Status		
Hearing	54	64
Hard-of-Hearing	16	19
d/Deaf	14	17
Community Affiliation		
Deaf Community		
Yes	58	69
No	26	31
Hearing Community		
Yes	11	13
No	73	87

Summaries of the general demographic information from 84 completed survey responses. Percentages rounded to the nearest single digit.

3.2 Deaf Adults

After responding to the general demographic questions, subgroups were created within the total data sample to analyze for differences in the utilization of genetic test results and emotional affect for those tested for hearing loss. The survey also assessed respondents' experiences with genetic counseling with questions regarding explanation of testing, explanation of results and willingness to have genetic counseling. Branching logic within the survey created two main groups: adults with hearing loss who had genetic testing, and parents of children with hearing loss who had genetic testing. Individuals who identified their hearing status as either deaf or hard-of-hearing were asked a specific set of questions pertaining to their own genetic testing, decision-making and emotional well-being. From this point on, the term "deaf" will be used as an inclusive term for individuals who identified as both deaf and hard-of-hearing. The branching logic was designed so that respondents could be a deaf adult and also a parent of a deaf child. The eight participants who fell into the latter group were prompted to answer questions pertaining to both their personal experiences with genetic testing and genetic counseling and their experience with their child's testing for hearing loss. Table 2a details the specific characteristics and response choices for deaf adults. There were 30 participants in this category, and fewer than half responded to questions asking about their decision-making after genetic testing. Those who skipped the questions regarding decision-making for medical management, education, language development, family planning and spouse selection were still able to complete the remaining survey questions, and their responses were used in the data analysis (Table 2a).

All 30 respondents who identified as deaf adults answered the questions pertaining to general demographics, their own hearing loss, assistive hearing devices and whether or not they had children with hearing loss. Two-thirds of the respondents reported wearing a hearing assistive device, either a hearing aid (53%) or a cochlear implant (13%). Adults with hearing loss who reported having deaf relatives (59%) were able to select multiple relatives but could not specify the number of relatives within that category (i.e., there was no difference between having one deaf sibling and three deaf siblings). Of those who had deaf relatives, three responded that they had a deaf spouse or romantic partner, four had one or two deaf parents, three had one or more deaf siblings, two had one or more deaf uncles or aunts, three had one or more deaf grandparents and three had one or more deaf cousins. Among the 16 who had deaf relatives, four reported having a known genetic syndrome associated with their deafness. Half of the deaf adult respondents stated that they use both oral language and sign language to communicate, which is comparable to the percent (52%) of parents of deaf children who use bilingual communication with their child. Bilingual communication was common among participants, and this percentage of ~50% held steady across other categories, including those with syndromic and non-syndromic hearing loss.

The response rate to questions regarding utilization of genetic test results among deaf adults was poor. Only four responded to questions concerning medical decisions, eight to questions concerning language and education, 13 to questions concerning family planning, and two to questions concerning spouse and partner selection.

Table 2a. Characteristics of Adults Who Are deaf

Characteristic	N	%
Total Participants	N=30	
Assistive Hearing Device		
Cochlear Implant	4	13
Hearing Aid	16	54
Both	0	0
None	10	33
Have deaf Children	N=30	
Yes	8	27
No	8	27
Do not have children	14	46
Have deaf Relatives	N=27	
Yes	16	59
No	11	41
Communication Method	N=26	
Oral Language	9	35
Sign Language	4	15
Both	13	50
Syndromic Deafness	N=26	
Yes (please specify)[a]	10	39
No	16	61
Ordering Provider	N=13	
Primary Care Provider	2	15
Pediatrician	0	0
ENT	1	8
Genetics Doctor	3	23
Genetic Counselor	1	8
Audiologist	0	0
Other Healthcare Provider[a]	6	46
Direct-to-Consumer	0	0
Genetic Counseling Provided	N=13	
Yes	4	31
No	5	38
Unsure	4	31
Medical Decisions	N=4	
Hearing Device Placed	0	0
Additional Testing and/or Imaging Ordered	2	50
Referral to Specialist Placed	2	50
Other	0	0
Language Decisions for deaf Adults with Children	N=7	
Primary Focus on Sign Language	1	14
Primary Focus on Oral Language	0	0
Both Sign and Oral Language	2	29
None of the Above	4	57

Education Decisions for deaf Adults with Children	N=6	
Oral Instruction without Sign Language Interpreter	2	33
Oral Instruction with Sign Language Interpreter	0	0
Primarily Sign Language Instruction	2	33
Other[a]	1	17
No Children of School Age	1	17
Did Genetic Testing Affect Family Planning	N=13	
Yes	4	31
No	9	69
Family Planning Decisions (Responded Yes)	N=4	
Decided to have more children	1	25
Decided to not have more children	1	25
Decided to have prenatal genetic testing	0	0
Decided to have partner carrier testing	0	0
Other[a]	1	25
None	1	25
Spouse/Partner Selection	N=2	
Chose a spouse/partner who is deaf	0	0
Chose a spouse/partner who is hearing	1	50
Partner received genetic testing	0	0
Other[a]	1	50

Characteristics and decisions made after receiving genetic test results of all respondents who identified as either deaf or hard-of-hearing adults. Percentages rounded to the nearest single digit. Participants could choose to skip questions, so totals of respondents to each question are noted above their respective groups. [a]See table 2b for other full description of "other" responses.

Table 2b. List of Other Responses for Adults Who Are deaf

Other Health Provider
Ophthalmologist
OBGYN
Other Education Decisions
"When child was identified as Deaf, we chose a school for the Deaf, as we are both Deaf parents."
Other Family Planning Decisions
"It probably affected my hesitancy about having children, but it didn't affect it a great deal."
Other Spouse/Partner Decisions
"It will probably affect how I ascertain if a partner would be caring or patient enough to be a good one throughout my vision loss.

Detailed responses to the deaf adult participants who selected "other" for each of the above categories, including the health provider who ordered the genetic testing, education decision made post-genetic testing, family planning decision made post-genetic testing, and spouse/partner decision. Participants used this section to clarify an existing answer choice or to add one that was not listed. Responses were not altered or edited.

3.3 Parents of Deaf Children

The response rate for parents who have deaf children was double that of deaf adults, which included eight deaf parents and 54 hearing parents. A total of 62 (74% of total) respondents who identified as parents whose children had genetic testing associated with their hearing loss completed the survey (Table 3a). They were asked specific questions via RedCap® branching logic about their children's characteristics (i.e., assistive hearing device, method of communication, age of diagnosis, etc.) and the decisions made for their children. This could also include adult children for whom the parent is answering the survey; however, the survey did not gather data regarding the current age of their child. Participants in this category included both hearing and deaf parents. All questions were asked of all participants, with the exception of language and education decisions, which only applied to hearing and deaf parents of deaf children.

Of those who reported having deaf relatives (29%), three reported having a spouse or romantic partner who is deaf, two had a deaf parent, three had at least one deaf sibling, five had at least one deaf uncle or aunt, one had a deaf grandparent and six reported having at least one deaf cousin. Participants were allowed to select more than one relative with hearing loss, and there was some overlap for the eight participants who are deaf themselves and have a deaf child.

Parents reported that their children used assistive hearing devices at higher frequencies than deaf adults, 90% and 66%, respectively. Of those who use assistive hearing devices, 29% have a cochlear implant, 57% wear a hearing aid (unilateral or bilateral not specified), and 5% use both. The rates of syndromic deafness ($p=0.222$) among deaf adults compared with parents

of deaf children was not significant. However, the frequency of specific communication methods used among these two groups was statistically significant ($p<0.001$). Comparisons were conducted between individuals with syndromic and non-syndromic hearing loss (Figure 4 and Table 5).

Parents with a deaf child were significantly more likely to select "other" in their responses to clarify their child's syndrome, associated features, or ordering provider and to include additional information pertaining to the questions regarding decision-making after genetic testing (Table 3b). Of those who indicated that their child has or is suspected to have a genetic syndrome, participants were able to list their child's specific symptoms or features. Features listed included microtia, hemifacial microsomia, heterochromia ("different colored eyes"), premature graying hair, vision loss and infertility.

Table 3a. Characteristics of Children Who Are deaf

Characteristic	N	%
Assistive Hearing Device	N=62	
Cochlear Implant	18	29
Hearing Aid	35	56
Both	3	5
None	6	10
Have deaf Relatives	N=62	
Yes	18	29
No	44	71
Communication Method	N=61	
Oral Language	23	38
Sign Language	6	10
Both	32	52
Syndromic Deafness	N=61	
Yes (please specify syndrome)[a]	22	36
No	39	64
Ordering Provider	N=54	
Primary Care Provider	2	3
Pediatrician	3	6
ENT	14	26
Genetics Doctor	21	39
Genetic Counselor	3	6
Audiologist	3	6
Other Healthcare Provider[a]	8	14
Direct-to-Consumer	0	0
Genetic Counseling Provided	N=54	
Yes	34	63
No	16	30
Unsure	4	7
Medical Decisions	N=41	
Hearing Device Placed	14	34
Additional Testing and/or Imaging Ordered	15	37
Referral to Specialist Placed	8	19
Other[a]	4	10
Language Decisions	N=49	
Primary Focus on Sign Language	3	6
Primary Focus on Oral Language	9	18
Both Sign and Oral Language	13	27
Other[a]	4	8
None of the above	20	41
Education Decisions	N=49	
Oral Instruction without Sign Language Interpreter	5	10
Oral Instruction with Sign Language Interpreter	14	29
Primarily Sign Language Instruction	5	10

Other[a]	7	14
No Children at School Age	18	37
Family Planning	N=49	
Decided to have more children	1	7
Decided to not have more children	9	65
Decided to have prenatal genetic testing	0	0
Decided to have partner carrier testing	2	14
Other[a]	2	14
Spouse/Partner Selection	N=0	
Chose a spouse/partner who is deaf	0	0
Chose a spouse/partner who is hearing	0	0
Partner received genetic testing	0	0
Other	0	0

Respondents did not answer all questions, so there are some characteristic totals that do not equate to the sum of the responses in that group. Percentages rounded to the nearest single digit. [a]See table 3b for other full description of "other" responses.

Table 3b. List of Other Responses for Children Who Are deaf

Specific Genetic Syndrome
- "Schwartz Jamel [sic] Syndrome Type 2"
- "Long QT syndrome"
- "Waardenburg Syndrome"
- "Usher syndrome 2a"
- "Usher Syndrome"
- "Usher syndrome (combined hearing, vision loss)"
- "Possible Waardenburg"
- "1q21.1 microdeletion, ADHD, developmental delays, FTT, GERD."
- "22q11.21"
- "Chromosome 13q deletion (midline defects – neurological deformities, vision issues, cardiac issues, kidney issues, genitourinary issues, hand deformities, supernumerary nipples, webbed neck, deafness."
- "Chromosome abnormality."
- "Progressive vision loss, Ushers 2a"
- "Partial trisomy 13q"
- "Genetic testing confirmed my child has Usher Syndrome."
- "Hemifacial microsomia (microtia atresia)."
- "Her loss is mixed (both conductive and sensorineural), and her doctors believe that the conductive element might be related to her Binder's syndrome (congenital malformation of the skull)."

Other Health Provider
- Cardiologist
- Gynecologist/OBGYN
- Neurologist
- Ophthalmologist
- Psychologist

Other Medical Decisions

"Genetic testing indicated a predisposition to worsening hearing loss with aminoglycoside antibiotics. I notify all medical providers and list it as an allergy on forms for school, etc."

"Helped us decide if we wanted to have another child."

"Knowing the cause of deafness was nonsyndromic we could focus on language support via ASL."

"Other children hearing tests."

Other Language Decisions

"Beginning to learn ASL, but mostly oral language."

"Learning Braille."

"Learning sign."

"We will learn tactile sign."

Other Education Decisions

"Bilingual oral asl school."

"Deaf, ASL-fluent nanny, 30 hours/weekly."

"Homeschool."

"No change in decisions. Speech therapy support continued."

"Total communication class."

"We didn't make a decision based on the results. For her sake, we wanted to know what caused her hearing loss. We took guidance from her ENT and Audiologist re: schools."

"We had already settled on a course of education before test results were returned."

"When child was identified as Deaf, we chose a school for the Deaf, as we are both Deaf parents."

Other Family Planning Decisions

"Knowing we both carried a gene for deafness, we were able to communicate this to other carriers in our families."

"We still had more children, but along with other factors, decided after 4 if we ended up wanting another we'd foster."

Detailed responses to the participants who selected "other" for each of the above categories including the specific genetic syndrome identified, the health provider who ordered the genetic testing, medical decision made post-genetic testing, language decision made post-genetic testing, education decision made post-genetic testing, and family planning decision made post-genetic testing. Participants used this section to clarify an existing answer choice or to add one that was not listed. Responses were not altered or edited so some are technically incorrect.

3.4 Genetic Counseling

Part of the motivation for this study was to understand the experiences that both deaf individuals and their families have had with genetic counseling. Participants were asked if they were ever provided genetic counseling for genetic testing for hearing loss and then were asked if they would want genetic counseling if it were offered to them. As shown in Figure 1a, many respondents did not have genetic counseling at any point, but most would have preferred genetic counseling if it were available to them.

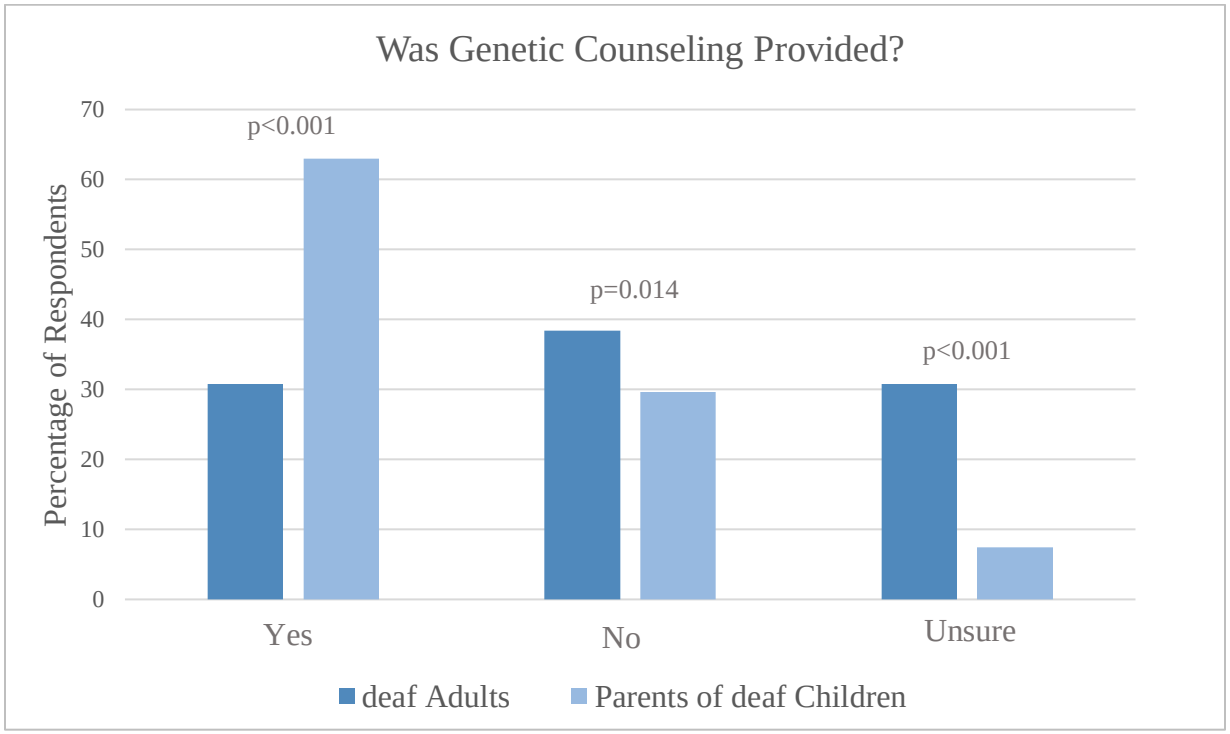

Figure 1a. Parents with deaf children received genetic counseling more frequently than deaf adults for genetic testing for hearing loss. P values were calculated using chi-square analysis unless the variable had an expected count of less than five, then Fisher's exact test was used, which is indicated with an asterisk*. Percentages were rounded to the nearest single digit.

Parents of deaf children received genetic counseling more frequently (63%) than deaf adults (31%), which was a significant difference between these two groups ($p<0.001$). Adults with hearing loss reported more often that they either did not receive genetic counseling (38%)

or were unsure if they had ever received it (31%). Among the remaining parents of deaf children, 30% did not receive genetic counseling and 7% were unsure if they had ever received counseling. The following definition of genetic counseling was provided in the question itself: Genetic counseling is the process of advising individuals and families who are affected by or at risk of genetic conditions to help them better understand the medical, psychological and familial implications.

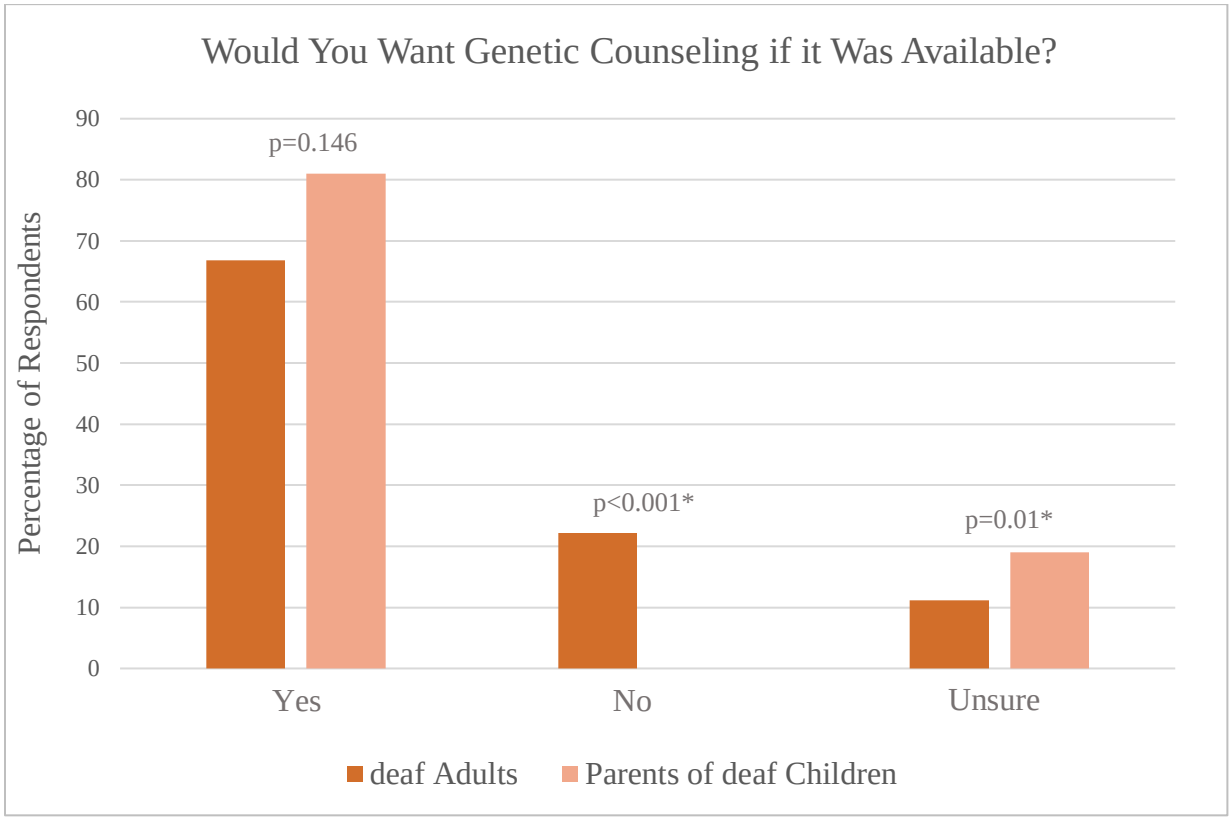

Figure 1b. Both deaf adults and parents of deaf children would want genetic counseling if it were offered for genetic testing for hearing loss. However, some deaf adults (22%) would still not want genetic counseling if it were available to them. P values were calculated using chi-square analysis unless the variable had an expected count of less than five, then Fisher's exact test was used, which is indicated with an asterisk*. Percentages were rounded to the nearest single digit.

The majority of both deaf adults (67%) and parents with deaf children (77%) would want genetic counseling if it were offered (Figure 1b). There were no respondents among the parents of deaf children group who reported that they would not want genetic counseling if it were

available to them; this was identified as a significant difference from the deaf adults ($p<0.001$). The remainder of the parents with a deaf child (24%) were unsure if they would want genetic counseling. The remaining deaf adults were either unsure if they would want genetic counseling (11%) or would not want it at all (22%).

Of those respondents who received genetic counseling, deaf adults (50%) and parents of deaf children (58%) reported more frequently that they received genetic counseling both prior to and after having genetic testing performed (Figure 1c). The remainder of deaf adults and parents of deaf children received genetic counseling either only before testing, 25% and 15%, or only after testing, 25% and 24%, respectively. A small percentage (3%) of parents were unsure of when genetic counseling was provided. The timing of when genetic counseling was provided was not significantly different between parents of deaf children and deaf adults. There were no statistically significant differences between when genetic counseling was provided between deaf adults and parents of deaf children.

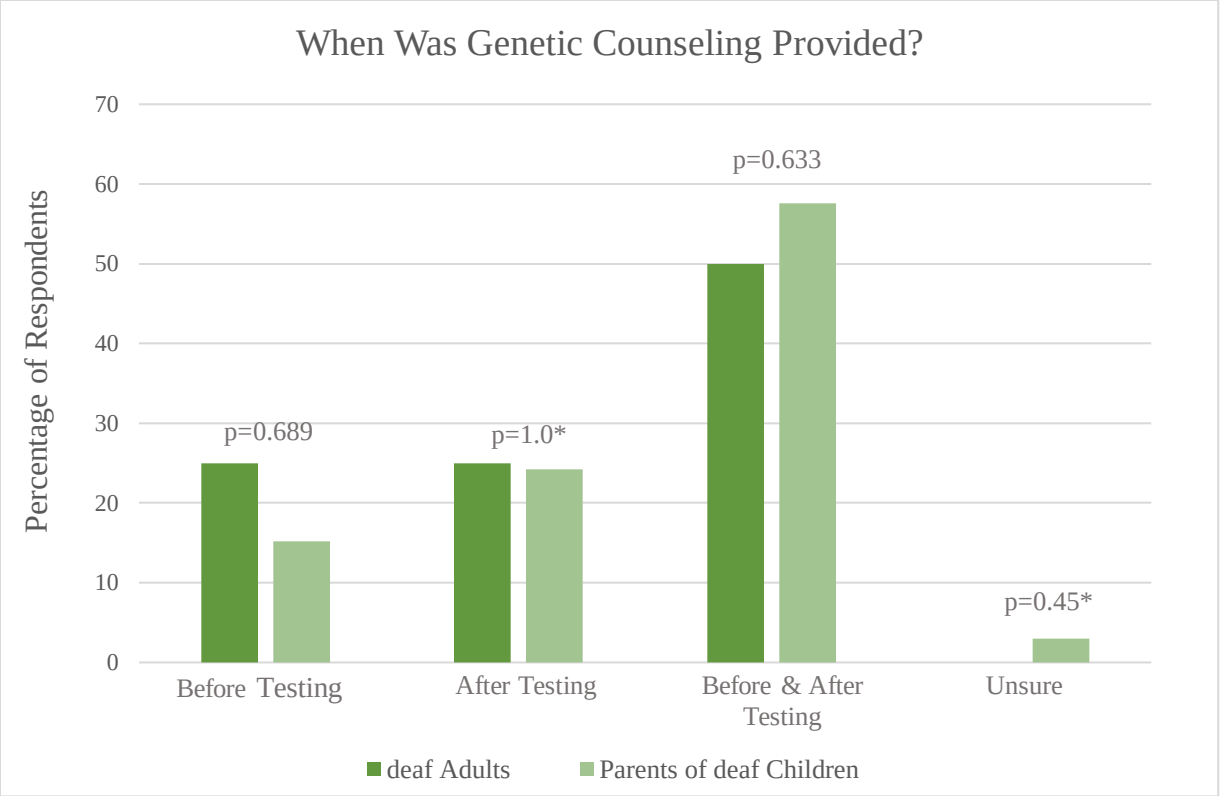

Figure 1c. Participants who received genetic testing for hearing loss received genetic counseling (GC) both prior to testing and after testing. P values were calculated using chi-square analysis unless the variable had an expected count of less than five, then Fisher's exact test was used, which is indicated with an asterisk*. Percentages were rounded to the nearest single digit.

After receiving their genetic testing results, 62% of deaf adults and 88% of parents of deaf children said that the genetic test results were clearly explained to them (Figure 1d). Nearly one-third (31%) of deaf adults and 8% of parents of deaf children did not feel that the genetic testing was clearly explained to them, and this difference between the two groups was significant (p=0.001). A small percentage of both deaf adults (8%) and parents of deaf children (4%) were unsure if the testing had been clearly explained to them.

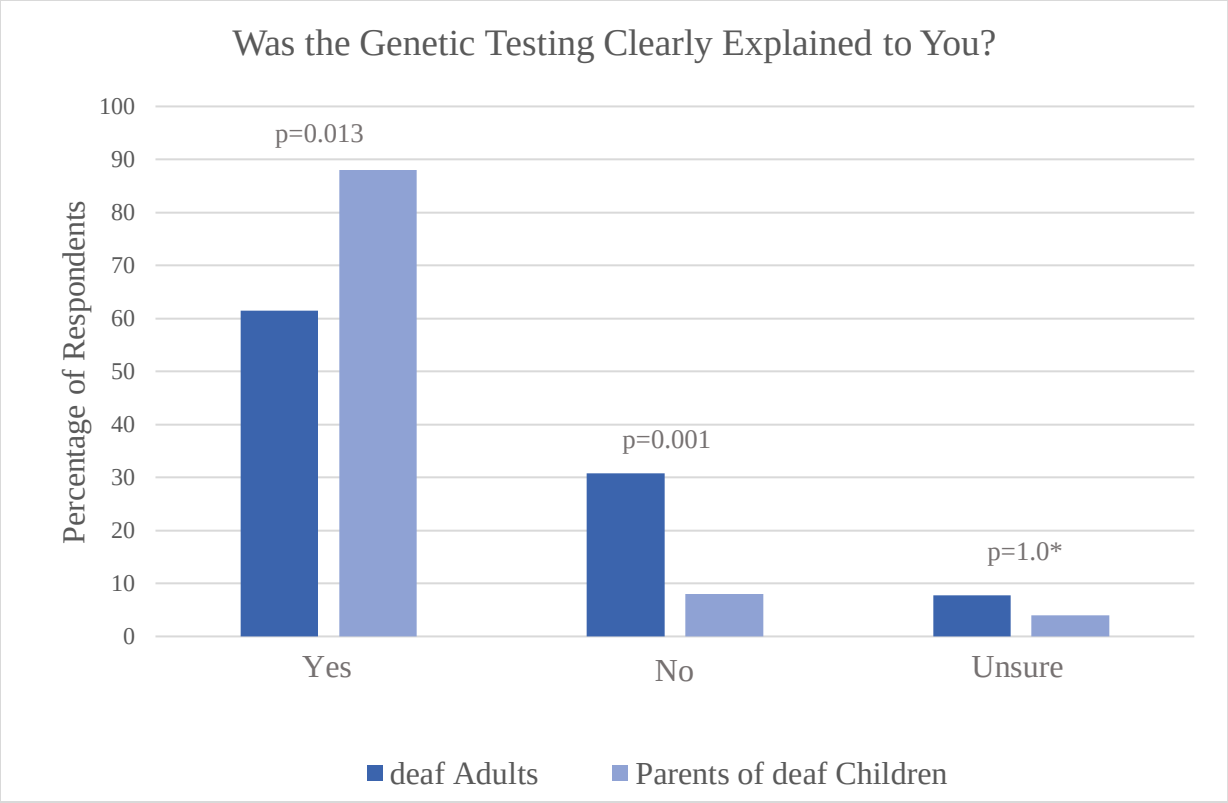

Figure 1d. Both parents of deaf children and deaf adults reported that the genetic testing was clearly explained to them regardless of when they received genetic counseling. However, 31% of deaf adults (dark blue) believed that the testing was not clearly explained. P values were calculated using chi-square analysis unless the variable had an expected count of less than five, then Fisher's exact test was used, which is indicated with an asterisk*. Percentages were rounded to the nearest single digit.

Respondents were also asked if the results provided a genetic etiology for their or their child's hearing loss (Figure 1e). Only 13 deaf adults responded to this question, among whom five (38%) said their results explained the genetic cause of their hearing loss, four (31%) said the testing did not explain the cause, three (23%) said the results were inconclusive, and one (8%) was not sure. For both deaf and hearing parents, 32 (58%) reported that the genetic test results did explain the cause of their child's hearing loss, 11 (20%) indicated that the testing did not find the cause, 11 (20%) said that the results were inconclusive, and one (2%) was not sure of the results. Parents of deaf children were more likely than deaf adults to report that genetic testing did provide a genetic explanation for hearing loss, and this difference was significant between the two groups (p=0.04).

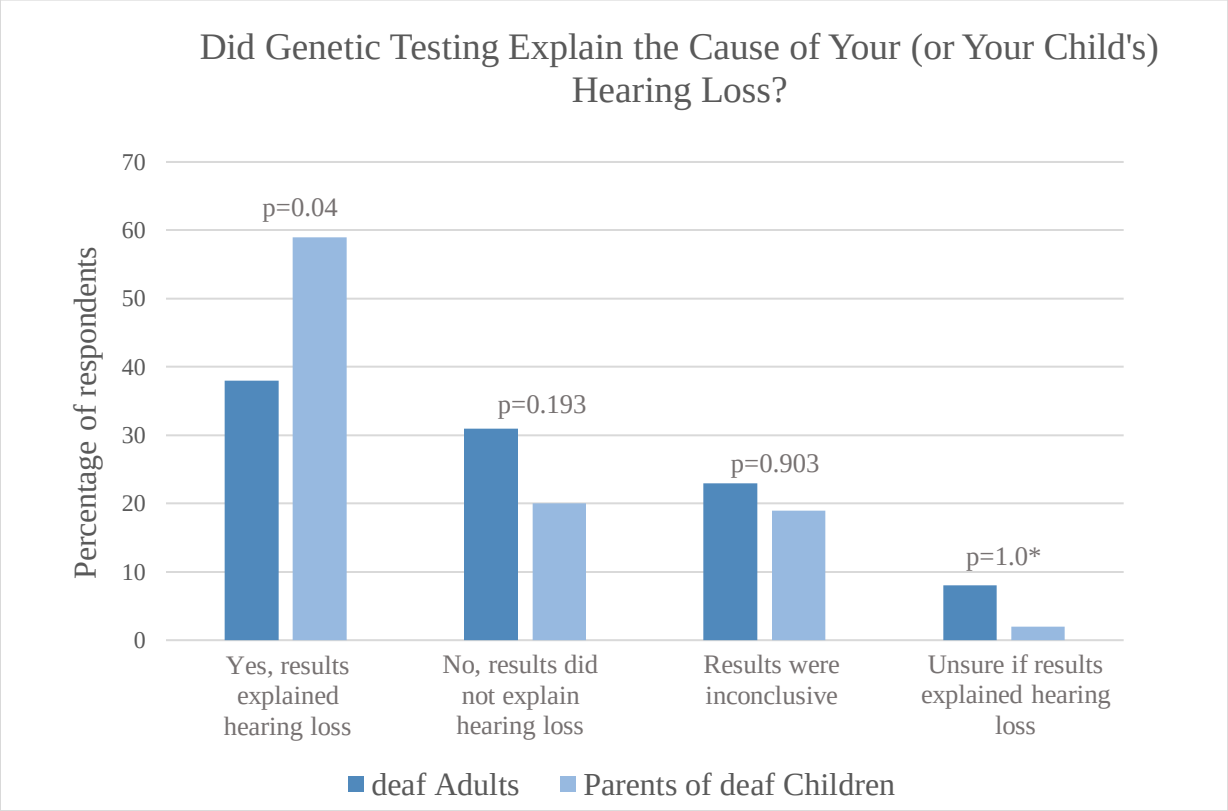

Figure 1e. Parents with deaf children (59%) reported that genetic testing explained the genetic cause of hearing loss than it did for deaf adults (38%). P values were calculated using chi-square analysis unless the variable had an expected count of less than five, then Fisher's exact test was used, which is indicated with an asterisk*. Percentages were rounded to the nearest single digit.

Genetic counseling was most frequently provided by genetic counselors for both deaf adults and parents of deaf children (Tables 2a and 3a). Parents of deaf children reported that genetic counselors provided genetic counseling more frequently (76%) than other healthcare providers. The remaining genetic counseling for this group was provided by a medical geneticist (15%), an ENT specialist (3%), or another type of provider (6%), noted in Table 3a as "other." Two respondents noted that a cardiologist and a reproductive endocrinologist, respectively, provided their genetic counseling. Only three individuals in the deaf adult subgroup answered this question, and all stated that a genetic counselor provided the counseling.

3.5 Utilization of Genetic Testing Results

One of the primary study aims was to assess how respondents used their or their child's genetic test results to make decisions surrounding medical management, language development, education, family planning and spouse selection. All participants were asked how their genetic test results influenced their decision-making (Table 4), and they were provided with additional space to clarify or add any additional information regarding their response.

Table 4. Impact of Genetic Testing on Decision Making for deaf Adults and Parents of deaf Children

	N	P-Value
Impacted medical decisions	N=66	
Yes		
deaf Adults	4	
Parents of deaf Children	32	0.011
No		
deaf Adults	10	
Parents of deaf Children	20	
Impacted language decisions	N=57	
Yes		
deaf Adults	0	
Parents of deaf Children	29	<0.001
No		
deaf Adults	8	
Parents of deaf Children	20	
Impacted education decisions	N=64	
Yes		
deaf Adults	6	
Parents of deaf Children	31	0.148
No		
deaf Adults	2	
Parents of deaf Children	25	
Impacted family planning decisions	N=62	
Yes		
deaf Adults	4	
Parents of deaf Children	14	0.258
No		
deaf Adults	9	
Parents of deaf Children	35	

Language decisions were only assessed for respondents with children (i.e., deaf adults who have children but not deaf adults without children). The deaf adult population was asked questions about how they made language development and education decisions for their children; there were only eight total respondents in that group, compared to 56 responses in the group of hearing parents. P values were calculated using chi-square analysis unless the variable had an expected count of less than five, then Fisher's exact test was used, which is indicated with an asterisk*. Percentages were rounded to the nearest single digit.

Participants could choose to disregard questions pertaining to decision-making, which resulted in a 68%-79% overall response rate to those questions. The response rate for questions

about whether or not spouse or romantic partner selection was influenced by testing was very low—2% of deaf adults and no parents of deaf children answered this question. Therefore, this question was left out of the analysis in Table 4. Overall, both groups reported that they made decisions based on their genetic test results for hearing loss. Within the deaf adult group, there were only eight deaf adults who also had deaf children, so there are only eight responses for the language development and education decision-making questions.

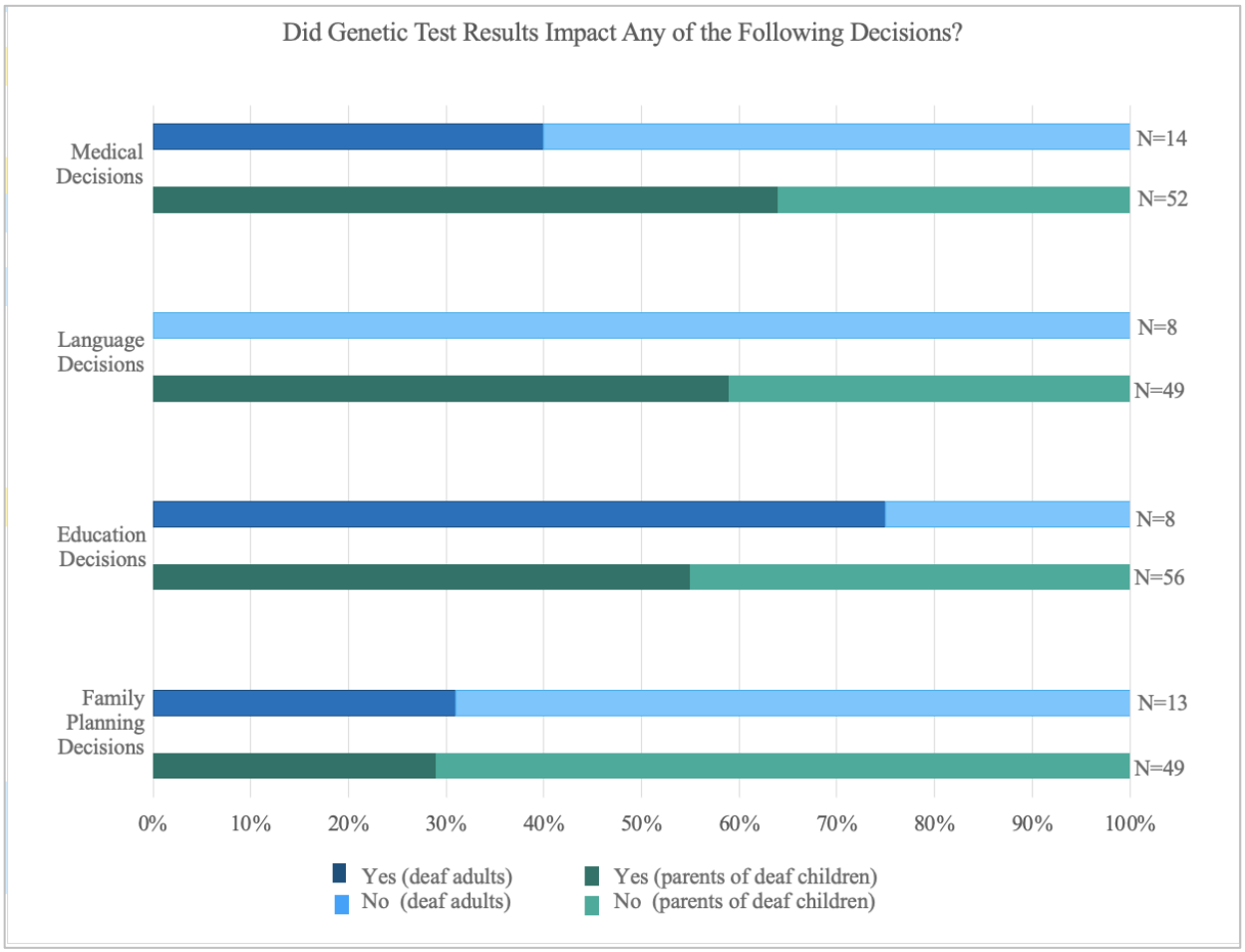

Figure 2. Medical decisions and language development decisions for parents of deaf children were influenced by genetic test results as compared to deaf adults. Comparison of the categorical utilization of genetic test results between deaf adults and parents of deaf children for decisions regarding medical management, language development, education and family planning. Respondents who identified as a deaf adult who received genetic testing are shown if they made a specific decision due to their results (dark blue) or did not (light blue). Respondents who are the parent of a deaf child are shown if they made a specific decision based on their child's results (dark green) or did not (light green). The number of responses for each category is shown to the right of the figure.

There was a 100% response rate to the question asking about changes in medical management post-genetic testing among those who identified as a parent of a deaf child. However, only 14 deaf adults (47%) responded to this question. Although many parents with deaf children made a specific medical decision after receiving their child's genetic test results, the single most common response was that there were no changes to their child's medical management (32%). The number of deaf adults who responded to this question was small, with only 14 total responses, and only four participants (29%) changed their own medical management after receiving their results. All four deaf adults who either had additional testing performed or had a referral to another provider identified as having a genetic syndrome.

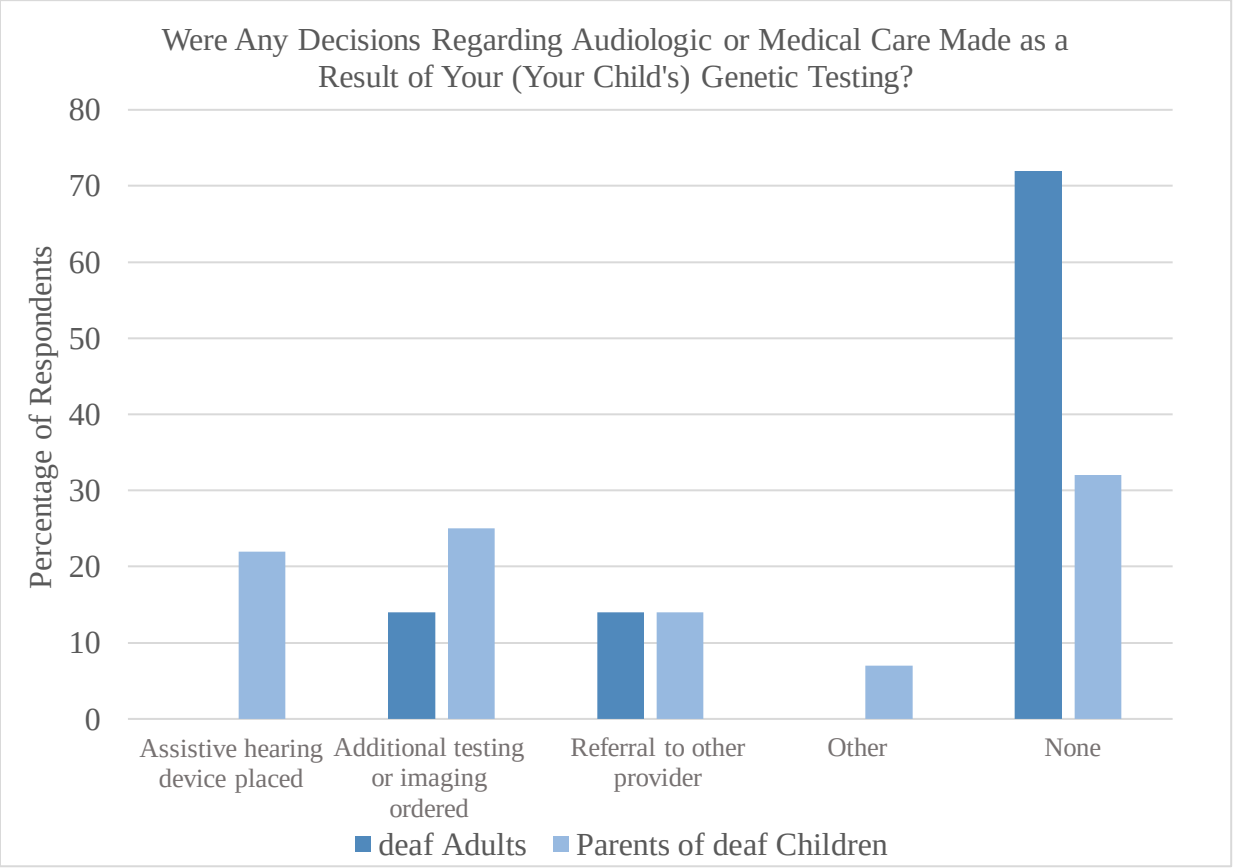

Figure 3a. Parents of deaf children made medical decisions (i.e., assistive hearing device placement, additional testing, referral or other) post-genetic testing for hearing loss. Respondents who selected "other" in this category were able to write in a clarification to their response or add an additional response (Table 2b and 3b). P values were calculated using chi-square analysis unless the variable had an expected count of less than five, then Fisher's exact test was used. Percentages were rounded to the nearest single digit (Table 4).

With all language development choices combined, 61% of hearing parents based their approach to their child's language development on their genetic test results. 39% of hearing parents of deaf children did not change anything about their plans for their child's language and communication development. Of that group, 6% decided to focus on sign language, 18% focused only on oral language, 75% incorporated both sign and oral language, and 8% selected "other" (Table 3b). Of the eight participants who identify as a deaf adult and who also have a deaf child, seven responded to this question (Figure 3b).

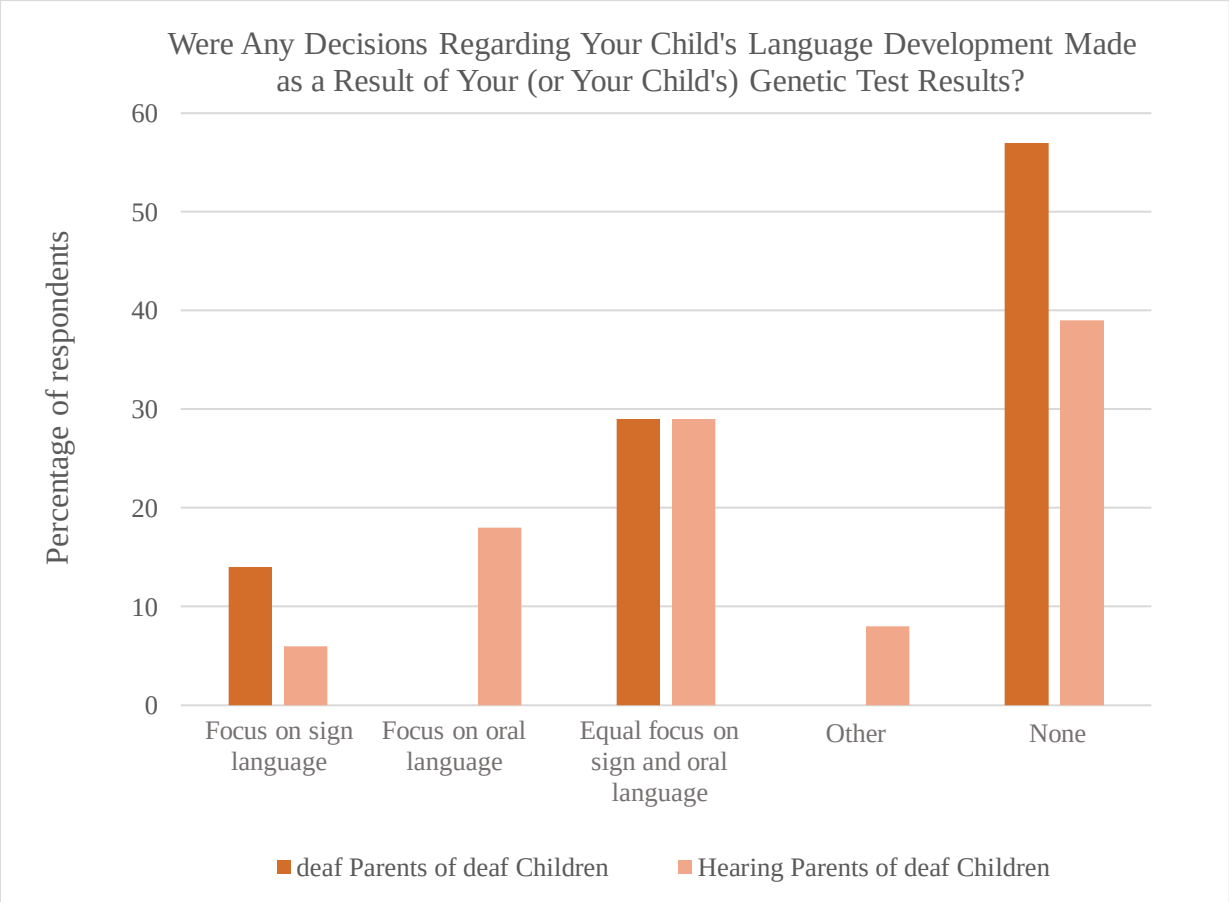

Figure 3b. Hearing parents of deaf children altered their child's language and communication less often than deaf parents based on their genetic test results. Of the hearing parents who made decisions regarding their child's language development post-genetic testing, more chose to use a bilingual approach, including a focus on both sign language and oral language. There was a small sample size of deaf parents who had a deaf child for this question. Respondents who selected "other" in this category were able to write in a clarification to their response or add an additional response (Table 3b). P values were calculated using chi-square analysis unless the variable had an expected count of less than five, then Fisher's exact test was used. Percentages were rounded to the nearest single digit (Table 4).

Respondents could only answer questions pertaining to changes to their child's education if they indicated that they had a deaf child. There were only eight deaf adults who also had a deaf child who were eligible to respond to this question. Of the seven deaf adults who answered this question, two indicated they chose a school with only oral instruction for their child, three chose a school with a focus on sign language, one chose "other" but specified that a school for the Deaf was chosen, and the final participant's child was not of school age. Educational decisions made by hearing parents were divided, with 10% choosing an oral-only curriculum, 28% preferring a bilingual approach, 10% choosing a sign language-only program, and 16% selecting a different educational approach not listed in the survey (Table 2b and 3b).

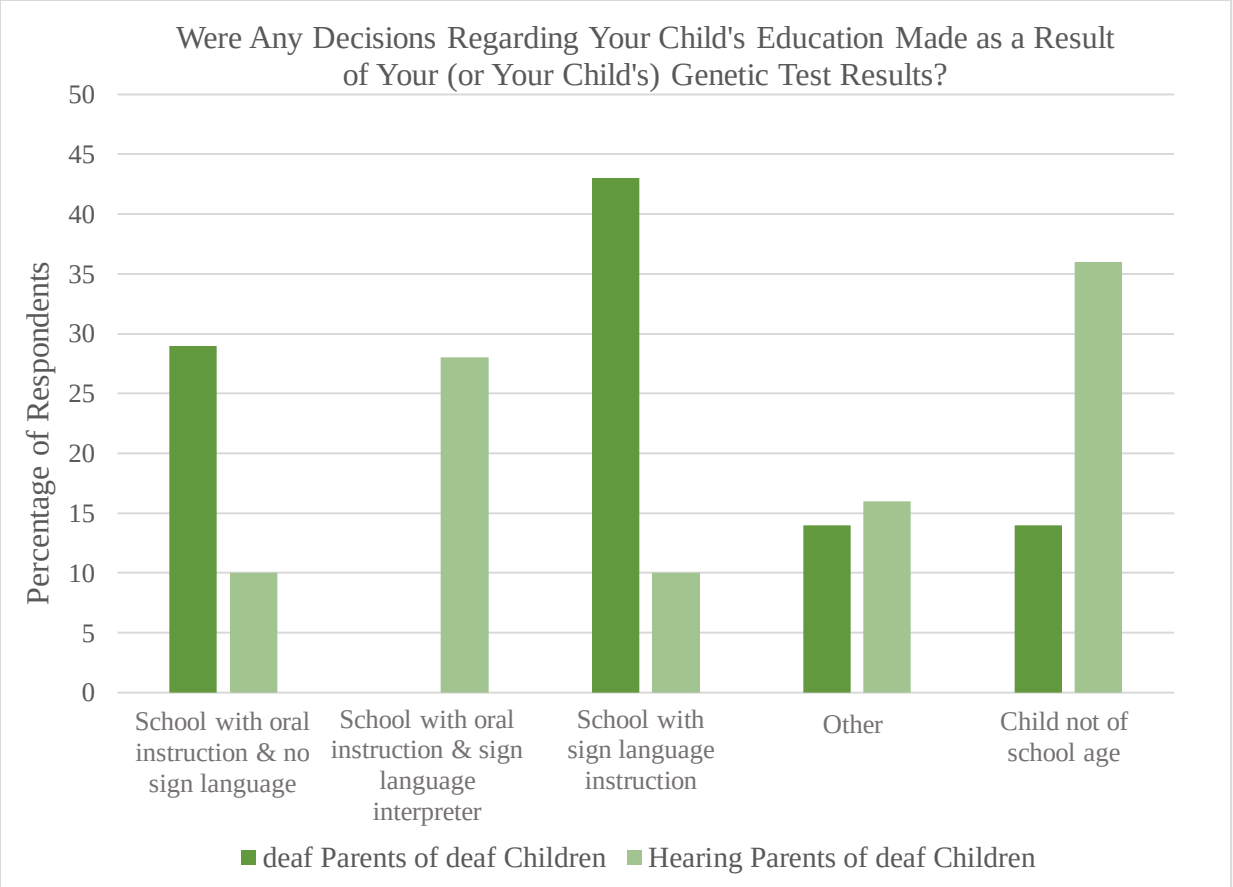

Figure 3c. Hearing parents of deaf children prefer a school that provides both oral instruction and a sign language curriculum or interpreter. Hearing parents who have deaf children of school age (44%) preferred a bilingual approach that includes both oral language and sign instruction. Respondents who selected "other" in this category were able to write in a clarification of their response or add an additional response (Table 2b and 3b). Not all children were of school age, which respondents were able to select as an option. P values were calculated using chi-square analysis unless the variable had an expected count of less than five, then Fisher's exact test was used. Percentages were rounded to the nearest single digit (Table 4).

The vast majority of participants did not change their family planning based on their or their child's genetic test results for hearing loss. Total response rate for parents of deaf children was 81%, and response rate of deaf adults was 43%. Of those who responded, 77% of deaf adults and 72% of parents with deaf children did not make any family planning decisions post-genetic testing results. There were only three deaf adults who made any family planning decisions, one decided to have more children, one decided not to have more children and one stated that it made them more hesitant to have children. The majority of parents of deaf children who did make

family planning decisions post-genetic testing decided not to have more children, but this was still only 18% of the total responses for that group.

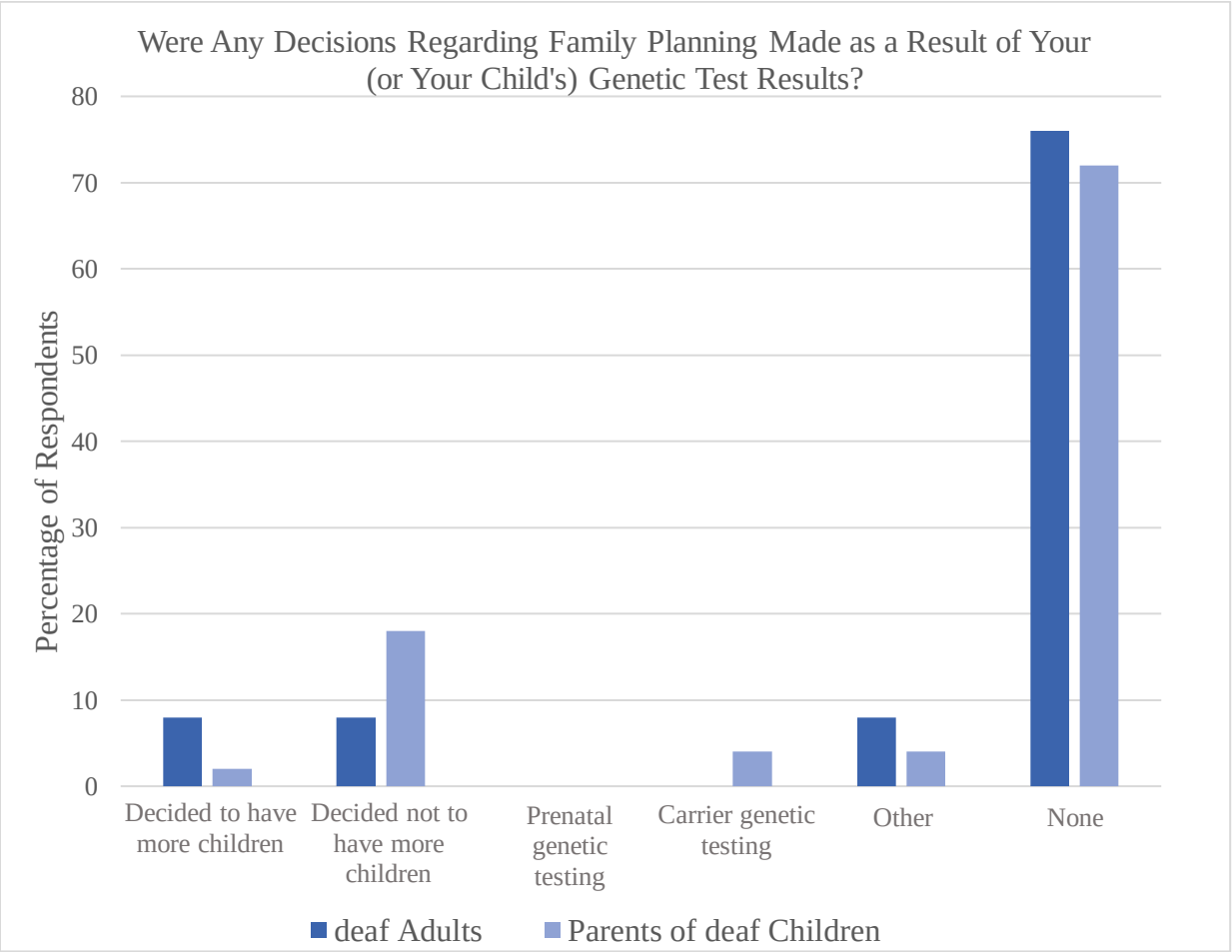

Figure 3d. Most participants did not make family planning decisions after receiving genetic test results. The majority of respondents did not make any changes or decisions regarding their family planning, with 72% and 76% of deaf adults and parents of deaf children selecting "none," respectively. P values were calculated using chi-square analysis unless the variable had an expected count of less than five, then Fisher's exact test was used. Percentages were rounded to the nearest single digit (Table 4).

3.6 Individuals with Syndromic and Non-syndromic Hearing Loss

A secondary study aim was to assess if there were any differences in the utilization and impact of genetic test results between individuals with syndromic and non-syndromic hearing loss. Participants were asked if their hearing loss was associated with additional symptoms or clinical features and if they had been diagnosed with a genetic syndrome. The survey provided additional space to allow participants to write in the specific features or genetic syndrome if known. Comparison of the responses from those with syndromic and non-syndromic hearing loss are shown below (Table 5).

Table 5. Impact of Genetic Testing on Decision Making When Considering Syndromic and Non-syndromic Deafness

	N	P-Value
Impacted medical decisions	N=63	
Yes		
Syndromic	16	
Non-syndromic	19	0.027
No		
Syndromic	8	
Non-syndromic	20	
Impacted language decisions	N=52	
Yes		
Syndromic	13	
Non-syndromic	18	0.061
No		
Syndromic	7	
Non-syndromic	14	
Impacted education decisions	N=52	
Yes		
Syndromic	16	
Non-syndromic	19	0.108
No		
Syndromic	6	
Non-syndromic	11	
Impacted family planning decisions	N=55	
Yes		
Syndromic	9	
Non-syndromic	8	0.001
No		
Syndromic	13	
Non-syndromic	25	

Respondents were grouped into syndromic and non-syndromic groups that included both deaf adults and parents of deaf children. Total sample size of each subgroup is noted above each category in the table. Specific genetic syndromes are listed (Table 3b). Table 4 does not include the spouse/partner selection category due to low number of responses. Chi-square analysis was used to calculate p-values.

There was a 73% response rate to questions pertaining to whether their or their child's hearing loss was syndromic. For some responses, there was overlap if the respondent was a deaf adult with a syndrome who also had a deaf child. These responses were counted separately since the participants were asked additional questions for both themselves and their child. Overall, 22

respondents reported being diagnosed with a genetic syndrome, and 39 reported having non-syndromic hearing loss.

Individuals diagnosed with a genetic syndrome associated with their hearing loss usually have additional clinical features that require further clinical management. This aligns with our data indicating that those with a genetic syndrome were more likely to make medical decisions based on their genetic test results than those with non-syndromic hearing loss (Figure 4). Within the syndromic group, 73% reported that they made specific medical decisions after receiving their or their child's genetic test results, whereas 48% of individuals with non-syndromic deafness made medical decisions post-genetic testing, a statistically significant difference ($p=0.027$).

Nearly two-thirds (65%) of individuals with syndromic deafness and 56% of individuals with non-syndromic deafness made decisions regarding their child's language development based on their genetic test result ($p=0.061$). Responses regarding their deaf child's education were higher, with 73% of individuals with syndromic deafness and 63% of individuals with non-syndromic deafness having made decisions regarding their child's communication development in school ($p=0.108$).

Overall, individuals with syndromic and non-syndromic hearing loss were least likely to make family planning decisions after genetic testing (Figure 4). However, nearly 40% of those with a genetic syndrome and 24% of those with non-syndromic hearing loss did change their family planning goals based on genetic testing. Respondents with syndromic hearing loss were

more likely overall than those with non-syndromic hearing loss to have made medical decisions (p=0.027) and family planning decisions (p=0.001) based on their or their child's genetic test results.

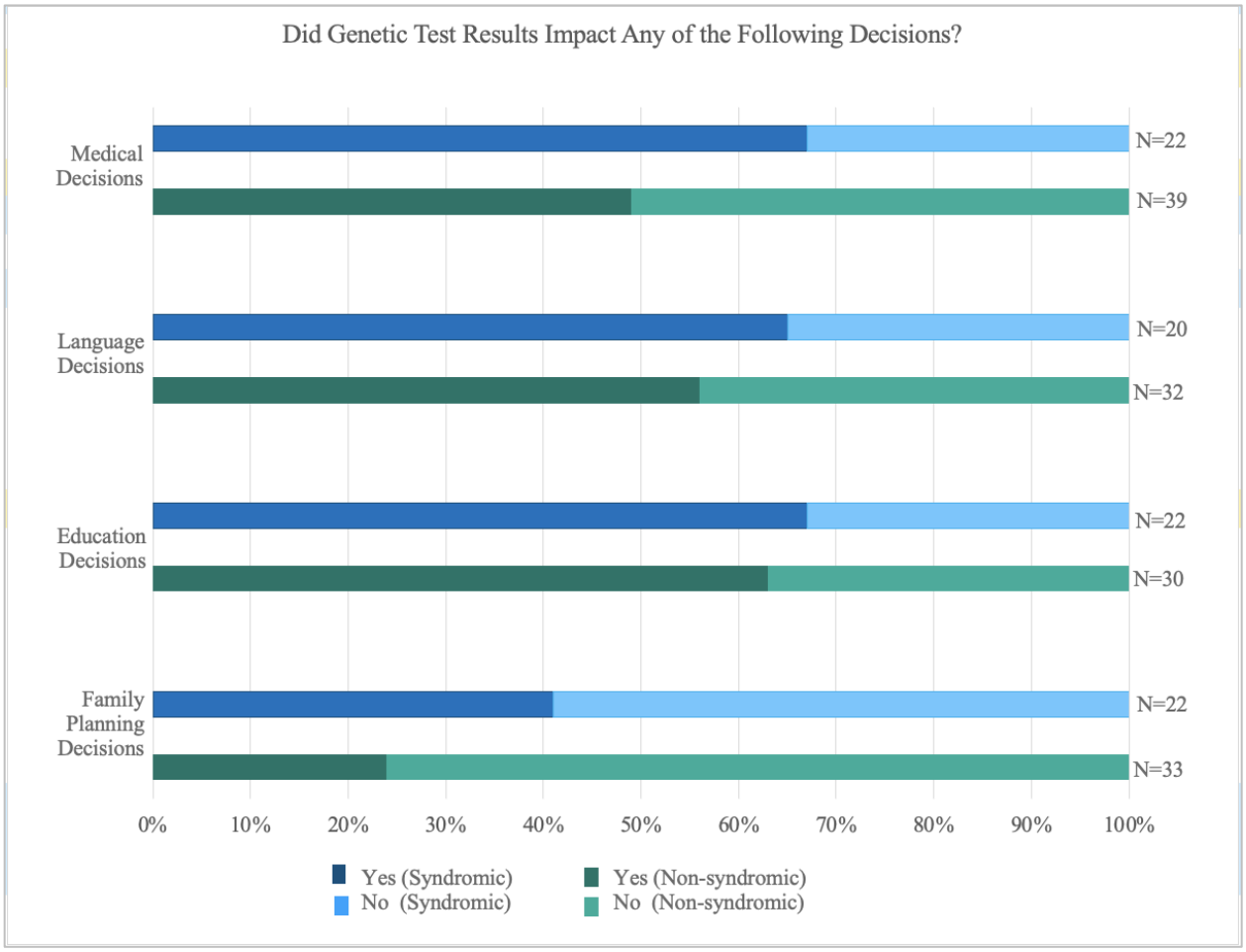

Figure 4. Respondents with a genetic syndrome associated with their hearing loss made more decisions overall based on their genetic test results than those with non-syndromic hearing loss. Individuals with syndromic deafness who made a medical, language, education or family planning decision after receiving their genetic test results are shown in dark blue, while those who did not are shown in light blue. Non-syndromic individuals are shown in dark green for those who made a decision in that category and in light green for those who did not. Percentages were rounded to the nearest single digit.

3.7 Ages of Diagnosis and Genetic Testing

Regardless of the type of hearing loss, most individuals (both adults and children) were diagnosed with hearing loss before one year of age (Figures 5a and 5b). Ages were broken into four separate brackets: birth to 1 year old, 2 to 10 years old, 11 to 18 years old and over 18 years

old. All groups, including deaf adults, parents of deaf children, those with syndromic hearing loss and those with non-syndromic hearing loss, reported having been diagnosed with hearing loss most frequently under 1 year of age, with the second highest age bracket for diagnosis between 2 to 10 years old. The only groups that reported being diagnosed as an adult (older than 18 years of age) were the deaf adults and individuals with non-syndromic deafness.

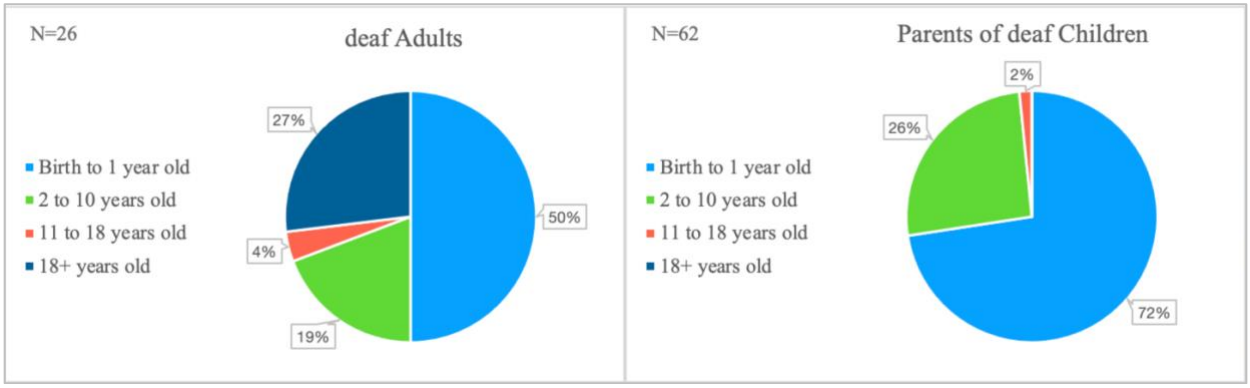

Figure 5a. Birth to one year of age is the most reported age that deaf adults and deaf children are diagnosed with hearing loss. Adulthood was the second most common time for hearing loss to be diagnosed in the population of adults who are deaf (27%). Deaf children had the second highest age bracket for hearing loss diagnosis in childhood between 2 and 10 years old (26%). Percentages were rounded to the nearest single digit.

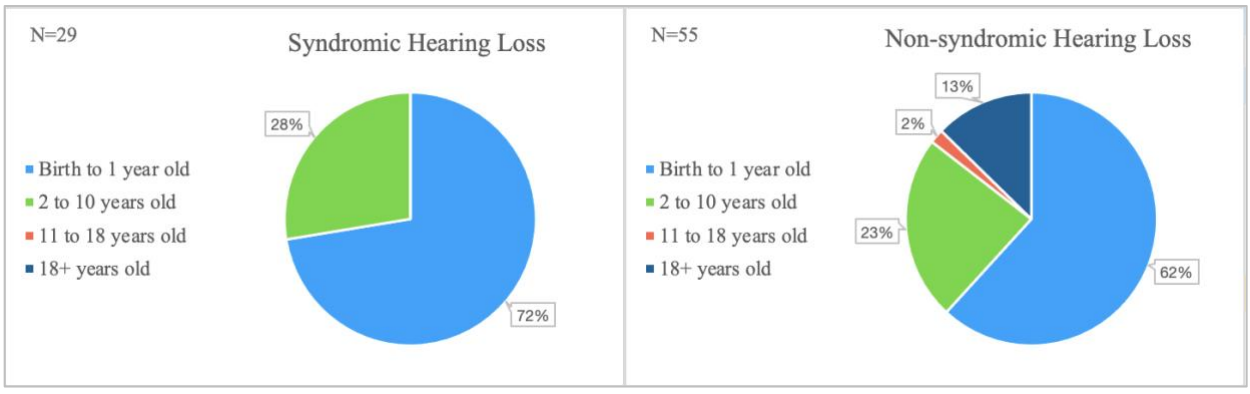

Figure 5b. Birth to one year of age is the most reported age that both syndromic and non-syndromic deafness was diagnosed. Individuals with both syndromic and non-syndromic hearing loss had the second highest age bracket for hearing loss diagnosis in childhood between 2 and 10 years old. Percentages were rounded to the nearest single digit.

Deaf adults were most likely to be diagnosed with hearing loss at an older age, with 27% being diagnosed in adulthood. Parents of deaf children and those with syndromic hearing loss both reported receiving a hearing loss diagnosis prior to 1 year of age.

Individuals with syndromic hearing loss were more likely to be diagnosed with hearing loss earlier than those with non-syndromic hearing loss, with 72% of those with syndromic deafness being diagnosed before 1 year of age, and 62% of those with non-syndromic deafness. ($p=0.378$). None of the participants who had syndromic hearing loss themselves or a child with syndromic hearing loss reported being diagnosed after 11 years old, whereas those with non-syndromic deafness were diagnosed with hearing loss between 11 and 18 years (2%) and in adulthood after 18 years of age (13%).

When individuals actually had their genetic testing was similarly variable among the groups compared to the ages of their hearing loss diagnosis (Figures 6a and 6b). The majority of deaf adults (77%) reported that they had genetic testing performed after 18 years of age. The opposite is true for the remaining groups, which shows that 92% of parents with deaf children, 64% of those with syndromic hearing loss and 85% of those with non-syndromic hearing loss had genetic testing in childhood (0 to 9 years old).

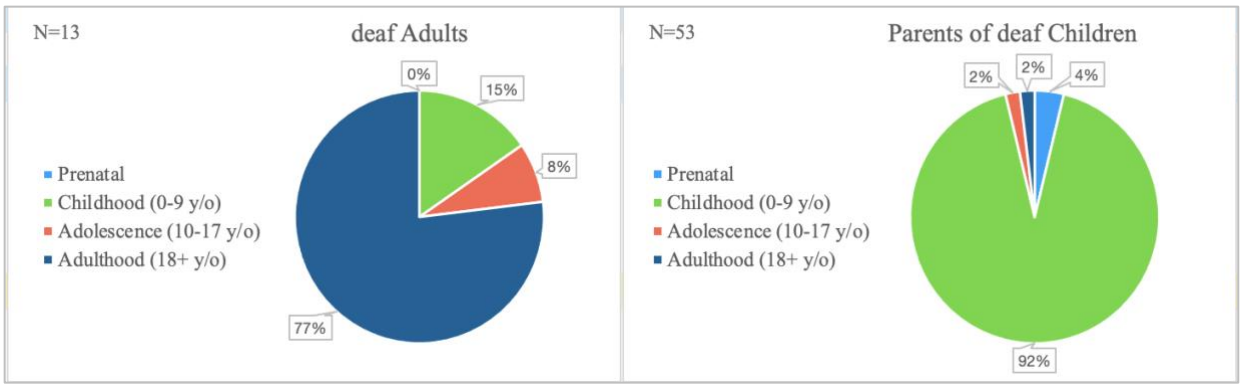

Figure 6a. Adults who are deaf received genetic testing in adulthood, while deaf children received genetic testing in childhood. No deaf adults reported receiving genetic testing during the prenatal period. Only 8% of deaf children child received genetic testing outside of the 0-9 years age bracket. Percentages were rounded to the nearest single digit.

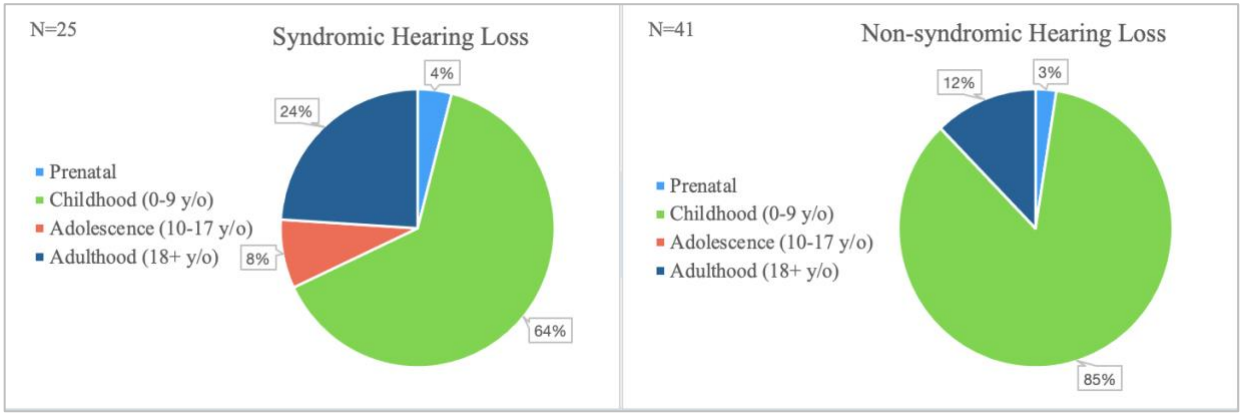

Figure 6b. The most common age bracket for both individuals with syndromic hearing loss and those with non-syndromic hearing loss to receive genetic testing for hearing loss is in childhood, followed by adulthood. Percentages were rounded to the nearest single digit.

3.8 Emotional Affect Post-Genetic Testing

Another study aim was to assess what type of emotional effect(s) genetic testing had on respondents. It should be noted that some participants received genetic testing for themselves or their child many years ago, and the Positive and Negative Affect Scale (PANAS) does not take time into account when calculating the total affect score. Total sum affect was measured using PANAS, which is a validated tool used to measure the emotional affect of an individual after a potentially triggering or emotional event. It relies on 20 self-reported measures of both positive and negative feelings (Watson et al. 1988). Sum totals of parents of deaf children are shown in Figure 7a, and sum totals of deaf adults are shown in Figure 7b. Total outcome scores above zero are considered a positive, and scores below zero are considered negative.

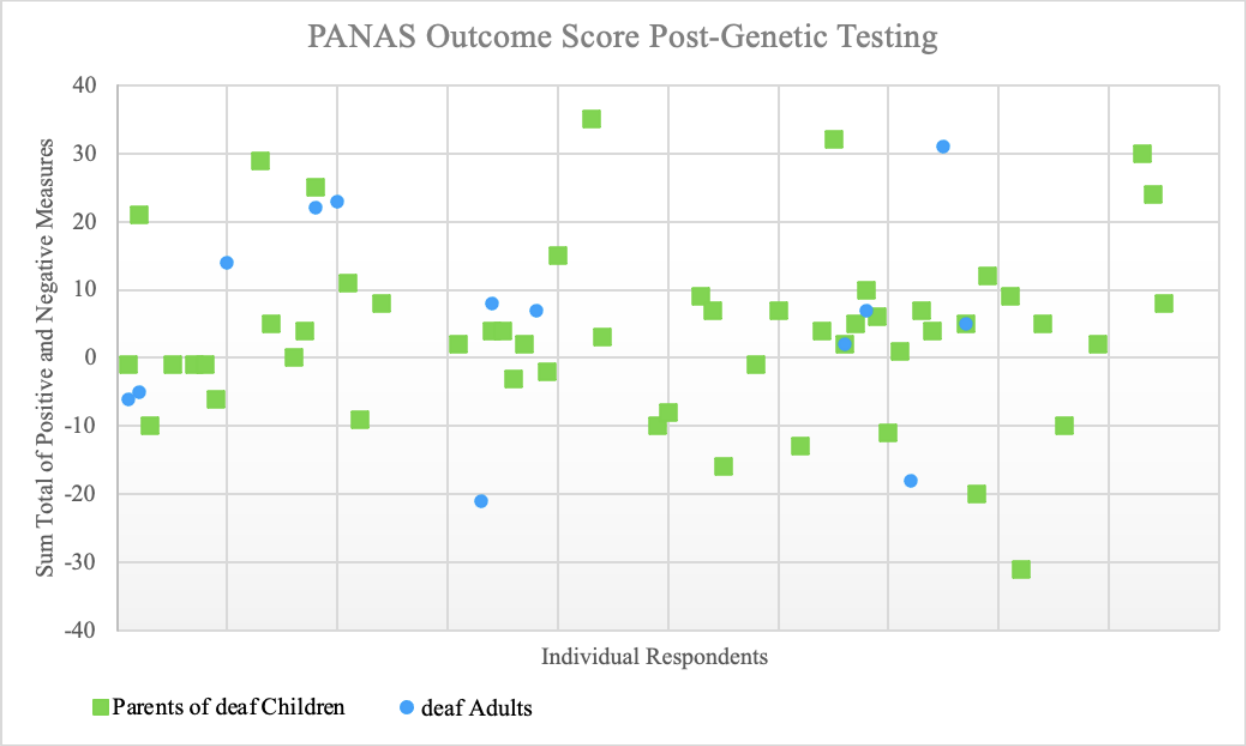

Figure 7. Emotional affect scores of parents of deaf children and deaf adults. An overall positive affect (64%) and (69%) was reported in parents of deaf children and deaf adults after receiving genetic test results. PANAS total score was calculated for each respondent (see methods). The x-axis denotes the individual respondents in order of survey submission. Parents who are deaf and also have a deaf child were able to respond the PANAS questions for themselves and their children separately. Some deaf parents who also have a deaf child had scores that overlapped, while others had a different outcome score for themselves than for their child. Total response rate to all PANAS scale questions was 86%. The remainder of respondents either had a sum negative affect (34%) or neutral affect (2%) on their emotional well-being after receiving genetic test results. Respondents were not required to answer any PANAS measurement questions. Responses that had a positive affect sum are greater than zero. Responses that had a negative affect sum are less than zero.

There was limited sample size in the deaf adult population, with a response rate of 43% of the total participants in this group who completed the survey. The remainder of respondents (31%) reported a sum negative affect on their emotional well-being after receiving their genetic test results. Parents of deaf children had an 85% response rate to the PANAS scale, while deaf adults had a 43% response rate. Both groups reported an overall positive affect for 64% of parents of deaf children and 69% of deaf adults, which also aligns with many of the positive comments written by participants at the end of the survey. Although some felt that genetic testing had a negative impact on their emotional well-being, the comments at the end of the survey

largely align with the finding of the PANAS scale. One participant stated, “the genetic testing and counseling helped put a lot of concerns to rest and also shed a lot of light regarding Deafness in our family history,” while another participant wrote, “it was a largely positive and helpful experience…but I also felt overwhelmed and at times, numb, throughout the process.”

IV. DISCUSSION

This study aimed to define the utilization of genetic test results for hearing loss by both deaf adults as well as parents of deaf children. Although research has previously been done on the motivations for pursuing genetic testing for hearing loss, the ways in which results are used by these individuals had not yet been assessed. The primary goal was to increase understanding of the impact that genetic testing has on deaf families so we as genetics professionals can approach testing and counseling in an effective, respectful and culturally competent manner. Understanding the perspectives of those with hearing loss is a crucial part of the genetic counseling process, and this study is a small step toward amplifying those experiences.

4.1 Utilization of Genetic Test Results

There is marked variability in the ways in which individuals and families use genetic test results for hearing loss, which is understandable considering the different genetic conditions associated with hearing loss, attitudes toward genetic testing, cultural beliefs and values. Overall, this study found that both deaf and hearing parents with deaf children made post-genetic testing decisions regarding medical management ($p<0.001$) and language development ($p=0.011$) for their children more frequently than deaf adults made for themselves.

More than two-thirds (68%) of parents of deaf children, or 32 respondents, made a decision regarding their child's medical management based on their genetic test results. This is more than twice that of the deaf adult group who made medical decisions (28%), which only consisted of four participants. The difference between the frequencies at which both groups made

medical decisions was found to be statistically significant and suggested that parents were more likely to change their child's medical management after having genetic testing than deaf adults were for themselves. We also must consider that there are individuals with syndromic and non-syndromic hearing loss within both the deaf adult group and the parents of deaf children group. There are similar percentages of people with syndromic and non-syndromic deafness in both groups; 38% of deaf adults and 36% of parents of deaf children reported that their children's hearing loss was related to a genetic syndrome (Tables 2a and 3a). There could be several reasons that parents of deaf children were more likely to make medical decisions than deaf adults, one of which may be the increased availability of medical technology now compared with how long-ago deaf adults received testing. For deaf adults who may have had testing many years ago, there would likely be a difference in the number of genes tested and, therefore, potential differences in the impact of testing. This result could also be reflective of the age of the individual at the time that testing was done. For example, adults having genetic testing likely had already made decisions, or had decisions made for them, about using hearing-assistive devices and, therefore, receiving genetic test results would not necessarily have changed that. This interpretation certainly fits with our data, since no deaf adults indicated that they made the decision to use an assistive hearing device post genetic testing. It is important to consider whether parents of deaf children would have made the same decisions if they had genetic testing later or not at all. Many deaf children had genetic testing early in childhood, which means that testing may have been done before any medical decisions regarding placement of assistive hearing devices would have been made. More exploration as to the reason for this difference in medical decision-making would be valuable in terms of understanding why parents tend to make these types of decisions based on genetic test results.

Although the data suggested an association between deaf parents and hearing parents with the decisions regarding their children's language development, there were only eight deaf parents who responded to this question compared with 49 hearing parents, so no assumptions of an association can be made because there is not sufficient statistical power. However, 61% of hearing parents and 52% of parents overall noted that their child's language development was influenced by their genetic test results. The preferred approach among this group was the bilingual approach, which combines both oral language and sign language. This result does not come as much of a surprise, since at the beginning of the survey participants were asked the method of communication they use, which aligns with the results from this question. Parents were asked about the method of communication they use with their deaf child, so we would expect this to be similar to their choices for language development.

The number of deaf parents who responded to questions regarding education decisions for their children was small (n=8) so there was not much power to assess for statistical differences between hearing and non-hearing parents, but it appeared that hearing parents of deaf children were more likely than deaf parents to make decisions regarding the specific school or communication method they chose based on their genetic test results. The most popular choice overall was a school that used primarily oral language but had a sign language interpreter available. This seems to fit with our other results that showed a preference for bilingual language development. The difference between deaf parents and hearing parents in this category was not statistically significant, but the sample size was too small to allow for any conclusions to be made.

It was very clear that participants did not feel that genetic testing influenced their choices in terms of spouse or romantic partner selection, such as choosing a partner who is deaf or a partner who is hearing. One participant was more concerned about having a partner who would have the emotional capacity to help them through their progressive vision loss, stating, "It will probably affect how I ascertain if a partner would be caring or patient enough to be a good one throughout my vision loss." This question had the lowest response rate of any question in the survey, which could be due to multiple factors. One possible explanation could be that they may have already chosen a partner or spouse before they had testing, while it may also suggest that individuals who are deaf or have a deaf child are not using genetic test results to make decisions about choosing a partner.

Study participants reported an overall positive effect on their emotional well-being after receiving their genetic test results. There was no significant difference in emotional outcome between deaf adults and parents of deaf children. This means that there was still about one-third of participants who had genetic testing that resulted in either a negative impact or no impact on their well-being. This could be due to a multitude of reasons, and it is difficult to speculate based on these results alone. However, many participants provided additional context in their free responses and described their feelings after having received their or their child's results. Several participants mentioned positive effects of their genetic test results on their emotional well-being. One individual said, "The genetic testing and counseling helped put a lot of concerns to rest and also shed a lot of light regarding Deafness in our family history." Another stated that the results bonded her family, "Knowing there was a genetic reason for my children's deafness AND knowing that it was non-syndromic was such a relief for me. It gave me closure, and actually

helped me feel more connected to their deafness and bonded as a family since we now know both my husband and I passed the same genetic mutation down to them." However, there were also written responses describing stress, worry and anxiety after having genetic testing. One participant who was negatively affected by genetic testing stated, "I was very overwhelmed with all the questions from my genetic counselor, I got very upset and cried and I felt pushed through interview without acknowledgement or a chance to catch my breath." These comments illustrate how the emotional responses to the process of genetic counseling for hearing loss are diverse and the approach to them should be individualized. We also want to consider which participants chose to respond to the PANAS questions in the first place. Individuals who had a strong reaction after receiving their results may have been more likely to participate in this survey and thus to have introduced some bias into these results. The results highlight the need for increased psychosocial counseling for individuals and families receiving genetic testing for hearing loss.

4.2 Syndromic and Non-syndromic Hearing Loss

A secondary aim of this study was to assess whether any differences exist between individuals with syndromic and non-syndromic hearing loss with respect to how their genetic test results impact their decisions and emotional well-being. The overall frequency of syndromic hearing loss for deaf adults (38%) and parents of deaf children (36%) were similar, but parents were more likely (73%) to specify the name of the genetic syndrome or describe the features. None of the adults who had syndromic hearing loss chose to specify the name of the syndrome or describe the associated features. The difference in response to describing the syndrome was significant ($p<0.001$). It is unclear exactly what caused this discrepancy, but the data potentially could have been skewed by the low sample population.

The participants who had a genetic syndrome were more likely to make test result-based decisions regarding medical management, language development, education and family planning than the participants with non-syndromic hearing loss. However, the difference was only significant for two categories, medical management ($p=0.027$) and family planning decisions ($p=0.001$).

Sixty seven percent of those with syndromic deafness made medical management decisions after receiving genetic test results, whereas only 45% of the participants with non-syndromic deafness made medical decisions after testing. Hearing parents of deaf children whose child was found to have a genetic syndrome were also more likely to have made medical decisions based on genetic test results than adults with a syndromic etiology. Nearly 78% of the parents with a child who had syndromic deafness made changes to their child's medical management after receiving their results. This is not surprising, considering that those with a genetic syndrome will usually have additional clinical features that require management and medical follow-up. However, it is remarkable that nearly half of those with non-syndromic hearing loss also made medical decisions based on their or their children's genetic test results. Even though respondents with syndromic deafness made result-based medical decisions more frequently, participants overall were influenced by their genetic test results to alter their or their child's medical management.

In addition to the impact that genetic test results had on medical management decisions for those who reported syndromic deafness in themselves or their children, participants' decisions regarding language development and education were also heavily influenced. Both

deaf and hearing parents of deaf children reported genetic test results impacting their decisions regarding how to handle their child's language development (65%) and education (73%). One participant wrote in the free response section that their child's diagnosis of Usher syndrome not only influenced the decisions for the child's educational support but also for their future career path, "Our youngest (13 year old) received the diagnosis of Usher syndrome type 1b in September 2020. This completely changes the path he will take for his career. It also requires more supports in the classroom. We have the opportunity to provide this for him while he is young to give him the most benefit. His siblings have also received testing and have USH 1b. We're scrambling to get them services. It would have been better to know while they were younger."

For some participants, eliminating the possibility of a syndromic cause for hearing loss is their primary concern when pursuing genetic testing. This is apparent in some of the responses written at the end of the survey. One individual said, "Genetic testing had several functions for our family. It quelled any concerns about syndromic issues possibly related to his deafness." Another participant wrote, "Our primary concern with testing was initially determining if our son had syndromic deafness and whether we needed to make any other medical interventions on his behalf. When his deafness was revealed to be non-syndromic, we moved on to other issues." These responses may provide an explanation as to why the results show less impact on decisions overall post-genetic testing, and especially for medical management, than for those with non-syndromic deafness.

4.3 Incomplete Responses

The only questions that required an answer choice in order to continue with the survey were the initial demographic questions on the first page of the survey. Although participants were not explicitly told they could skip questions, many participants "completed" the survey without answering all questions. There were no assumptions made in the data analysis if respondents did not complete an answer. For example, for questions in which there was the option to select "no" or "none of the above," the participant had to select that response, and a skipped question did not default to a "no" or "none of the above" response.

Some questions in the survey were more likely to be skipped. In particular, the questions pertaining to decisions regarding spouse or romantic partner selection only received two responses from the entire sample population of 84 individuals. The absence of responses to certain questions, such as spouse selection, may be an indicator that participants did not feel these questions applied to them or that those types of decisions were not related to their genetic test results (Table 2a, Table 3a). Many of the respondents were parents and may have already chosen a partner or spouse, in which case this question would be irrelevant since genetic testing would have taken place later.

There was a deficit in responses from deaf adults for the questions pertaining to decision-making overall. This could be due to multiple factors, such as whether the time to make these decisions had passed prior to having genetic testing or decisions were made for them by their parents that they are unaware of. Once again, if we had converted absent responses into a default "no" or "none of the above" response, this could have affected the significance of some of the

data analysis when comparing the adults to the parents of deaf children. However, without concrete responses, no assumptions could be made, and only the responses of those who answered these questions were used in the data analysis.

4.4 Genetic Counseling

In an ideal world, all individuals would have access to genetic counseling so they could make well-informed decisions for themselves and their families. However, as shown in Figure 1a, many respondents never received genetic counseling, either before or after they or their child had genetic testing. Nearly one-third of the parents of deaf children reported that they were unsure if they had received genetic counseling at any point in the genetic testing process. One interpretation of this uncertainty could stem from a general lack of understanding of what genetic counseling entails. A general definition of genetic counseling was provided in the question, but it is possible there was still confusion surrounding the question. We could also consider who was ordering the genetic testing and if participants believed that provider did, in fact, provide genetic counseling. Some of the ordering provider specialties that were reported included a gynecologist, a psychologist and an ophthalmologist, who likely did not provide genetic counseling since genetics is not their primary specialty. One participant wrote about their experience in the free response section and indicated that the doctor may have had some uncertainty in the genetic testing process: “We were told there was a mutation on a gene that might be linked to hearing loss but they are unsure if that is the cause. I feel like it's a lot of work and even the Drs are unsure when you do get results.” There are several possible explanations for this individual’s statement. One interpretation could be that they received a variant of uncertain significance (VUS) result, and that is why they noted that they were “unsure” about the mutation identified.

Another interpretation may be that the provider did not have a full understanding of the results themself and thus could not explain the results effectively to this individual. If this was the case, then this provider's lack of genetics knowledge not only could have impacted the patient's understanding of the results but also could have impacted their care. As genetic testing for hearing loss evolves and becomes more widespread, it may be appropriate to consider ways that we can educate non-genetics providers about genetic testing and results disclosure. These discussions with providers outside of the genetics specialty are worth exploring to determine what kind of information they provide to patients when they order genetic testing and whether additional provider education may be warranted.

Parents of deaf children were far more likely to report that they had received genetic counseling than were deaf adults. This is an interesting finding and could be associated with how long ago the deaf adults were diagnosed with hearing loss and had genetic testing. There is a more streamlined process now for the diagnosis of hearing loss for newborns; however, the process of obtaining genetic testing for hearing loss is still evolving. One explanation for parents of deaf children receiving genetic counseling more often than deaf adults could be a lack of availability or perhaps of awareness of genetic counseling at the time that genetic testing was performed if the two groups had testing at entirely different times (such as years ago for the deaf adults). Nearly a third of deaf adults noted that the genetic testing was not clearly explained to them. Some of deaf adults noted that they had genetic testing in childhood, so they themselves may not be aware of the discussion of genetic testing or may have not fully understood the information. This study did not account for this type of circumstance, but it should be considered when evaluating the results.

The overwhelming majority of participants who had not previously received genetic counseling indicated that they would want genetic counseling if it were offered to them. This suggests an increased benefit of genetic counselor involvement in situations where a patient or their child may have genetic testing for hearing loss. This is an important finding from this study and reinforces the argument for increased genetic counselor involvement in both adult and pediatric settings when genetic testing is being contemplated for a personal or family history of hearing loss. This will become even more relevant as hearing loss panels and carrier screening for genes associated with both syndromic and non-syndromic hearing loss become increasingly available.

4.5 Study Limitations

This study had several limitations surrounding the sample population, recruitment and study design. A major limitation is the small sample size. There were only 84 total responses used in data analysis from the survey, which does not provide enough statistical power to make strong associations between variables. This also put constraints on the different groups that could have been analyzed in the study since some groups were too small to compare. In addition to the lack of statistical significance, the small study sample was likely not to be representative of the broader deaf population. The respondents were primarily female, Caucasian and under 49 years old, which does not reflect the diversity of deaf individuals and their families.

Another limitation of this study was that many questions, apart from the initial demographic questions, did not require a response in order to continue taking the survey. This resulted in many individuals only responding to certain questions or sections of the survey.

Although we used partially completed surveys in the data analysis, it would have provided stronger associations if all participants had responded to all survey questions. It also included pre-written answer choices, which may not accurately or completely describe each participant's circumstances. Although there was an "other" option for most questions, some might have benefitted from also having a "not sure" or "I don't know" option to illustrate their true experience.

The recruitment method used for this study was solely online and primarily targeted toward those who are involved in social media support groups and communities. This limited the exposure of the survey to certain groups and was more likely to include those who are more involved in social media communities. Another limitation was possible self-selection bias and whether or not those who choose to take part in surveys have differing perspectives than those who do not. Participants needed access to the internet to take the survey, since it was not available in hard-copy format. Furthermore, the survey was only available in written English and was not provided in American sign language, Spanish or any other languages, so those whose primary language is not English or who are not proficient in written English would not have been able to take the survey. This survey did not ask whether or not a sign language interpreter was involved when genetic testing and the results were discussed. This would be an important factor in understanding the comprehension of genetic test results.

The timeline of when individuals or their children had genetic testing performed likely varied among participants. This study asked questions regarding decisions and feelings after genetic testing, which for some individuals may have been within the last 6 months but for others

may have been many years ago. The ability to recall exactly what decisions were made as a result of genetic testing or what the emotional impact was may have changed over time. A way to control for this would be to ask participants these questions closer to the time they received the genetic test results to decrease any recall bias.

4.6 Future Directions

This is an important area of research, and future studies should be considered to advance our understanding of the implications that genetic testing has for deaf families. A follow-up to this study could include an analysis of the decisions made by deaf individuals who have not had genetic testing. Such a study could incorporate both those who chose to have genetic testing and those who did not, ideally after receiving genetic counseling. By creating a control group of those without genetic testing, comparisons could be made to those who did have testing to see if genetic testing truly impacts how deaf individuals approach medical management, language development, education decisions and family planning. This would be particularly interesting to compare for those with non-syndromic hearing loss, in which there are no other features that would potentially require further medical evaluation. Additional exploration within this area would increase our understanding of whether these groups find genetic testing valuable and under what circumstances.

Additional studies should also include long-term follow-up of these individuals, especially for those tested in childhood, to see if any issues of autonomy arise for them. These types of long-term studies could also include examining health outcomes, language development and emotional well-being. Knowing whether or not adults would make similar or different

decisions for themselves than their parents made for them could provide guidance as to how genetic testing is discussed with parents of deaf children.

Lastly, this study lacked diversity in the sample population, and more extensive studies are needed to examine the generalizability of the results. This should include a larger sample population in order to capture the true outlook of individuals with hearing loss. To make future studies more accessible to the larger hearing loss community, they should be available in multiple languages, including American sign language, Spanish, and Mexican sign language. Other methods for recruitment should also be explored, such as recruiting directly through audiology clinics, ENT offices, and other centers for hearing loss. Alternative recruitment methods would address the issue of accessibility for those who wish to participate but are not involved in social media communities or do not have adequate internet access.

4.7 Conclusions

As evidenced by both the data and by the open-ended responses that participants shared at the end of the survey, the way that genetic testing is valued by deaf individuals is not the same for everyone. Although some individuals and families found genetic testing to be very important in their decision-making, others did not and would not consider genetic testing for hearing loss alone. This means that the ways in which we approach discussions surrounding genetic testing for hearing loss must be tailored not only to an individual's specific medical and family history but also to their personal and cultural beliefs. This study highlights the importance of access not only to genetic testing but also to genetic counseling services so that families understand all the potential implications of the results.

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APPENDIX A: IRB Exempt Self-Determination

UCI Office of Research

THE EXEMPT SELF DETERMINATION PROCESS

October 2019

To Whom it May Concern:

Exempt confirmation may be made by various mechanisms at UCI. Please note the following:

All undergraduate exempt research is submitted for exempt review and confirmation through the Undergraduate Research Opportunities Program (UROP).

The Exempt Self-Determination Tool may be used for self- determining certain types of exempt research at UCI. Exceptions do apply. Please refer to UCI HRPP Policy # 12 for the current exceptions. As part of the UROP and Exempt Self-Determination Tool process, UCI IRB review is not required and will not be provided. For studies that are submitted to the IRB where the Exempt Self-Determination Tool may be used instead, the study will be returned to the researcher to self-exempt. For exempt studies that require UCI IRB review, Lead Researchers must submit an application and supporting documents to the UCI IRB for review.

As part of using the Exempt Self-Determination Tool, Lead Researchers and Faculty Sponsors (as applicable) provide their assurance that they will follow relevant Human Research Protection Program (HRPP) policies and procedures, among other criteria. For a copy of the assurance, please review the following page.

If there are any questions regarding the exempt process at UCI, please contact HRPP Staff.

-The UCI HRPP

AS PART OF THE EXEMPT SELF DETERMINATION PROCESS AT UCI, THE LEAD RESEARCHER AND FACULTY SPONSOR (AS APPLICABLE) ASSURES THE FOLLOWING:

1. The information provided in this application is accurate to the best of my knowledge.

2. All named individuals on this project have read the procedures outlined in the protocol, are aware of and have reviewed relevant HRPP Policies and Procedures and understand their role on the study.

3. All named individuals on this project have completed the required electronic educational research tutorials and have been made aware of the "Common Rule" (45 CFR Part 46) and acknowledge the importance of the Belmont Principles - Respect for Persons, Beneficence and Justice in conducting research involving human participants. Also UCI has signed the Federalwide Assurance (FWA) that is available for review on the Human Research Protections (HRP) website.

4. Minor changes to the research that do not increase risk to participants, or significantly alter the study aims or procedures, such as the addition or removal of students researchers, do not require additional self-confirmation of exemption or approval from the IRB. Major changes that increase risk or constitute substantive revisions to the research including procedural changes will require a new self-confirmation of exemption or approval from the IRB.

5. When conducting research off-site or collaborating with an investigator at another institution (e.g., another UC, CHOC, CSUF, or a local school district), Lead Researchers must comply with the requirements and policies of the site, including securing Confirmation of Exempt Status from the IRB.

6. The Exempt Self-Determination, consent documents including recruitment materials and data collection materials will be maintained by the Lead Researcher or Faculty Sponsor for 10 years beyond the completion of the research.

7. This research study is subject to routine monitoring by the Human Research Protections (HRP) unit of the Office of Research. Through the Education Quality and Improvement Program (EQUIP) program, HRP staff conduct periodic quality improvement monitoring and educational outreach.

APPENDIX B: Thesis Survey

Assessment of the Impact of Genetic Testing and Genetic Counseling Among Deaf Adults and Parents of Deaf Children

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· You are being asked to participate in a research study being conducted as part of a Master's Thesis at the University of California, Irvine. Participation in this online study is voluntary.

- You must be at least 18 years old to participate in the study.
- You must be currently living in the United States.
- You must be diagnosed with hearing loss/deafness OR have a child diagnosed with hearing loss/deafness OR both

· If you choose to participate in the study, you will be asked to complete an anonymous survey. The questions in the survey will ask about your experience with genetic testing for hearing loss/deafness for you and/or your child. These questions will assess what decisions were made as a result of genetic testing. This survey also addresses any experiences with genetic counseling and if the genetic testing was explained to you.

· You may choose to discontinue the survey at any time. The survey is anonymous, and your responses are anonymous.

· The survey is expected to take about 10-15 minutes.

· There are no direct benefits from participation in the study. However, the goal of this study is to investigate the impact of genetic testing for hearing loss/deafness on individuals and families. It will assess whether or not genetic testing influences decisions regarding medical care, family planning and emotional well-being. We hope this study will provide insight into how genetic testing is utilized and how we may best serve deaf individuals and their families.

· This study was determined to meet UCI IRB exemption criteria.

· All research data collected will be stored securely and confidentially within a secure server through the UCI Department of Pediatrics, Division of Genetic and Genomic Medicine.

The following questions will ask about your demographics.	
Are you currently living in the United States?	◯ Yes ◯ No
How old are you?	_____
What is your gender?	◯ Female ◯ Male ◯ Non-binary
What is your ethnicity?	◯ Black or African-American ◯ Asian ◯ Caucasian ◯ Hispanic ◯ Native American ◯ Native Hawaiian or Other Pacific Islander ◯ Other
Which of the following best describes you?	◯ Hearing (An individual who is hearing is able to perceive sounds without hearing assistance devices, i.e. hearing aids/cochlear implants, and does not have any known or significant loss of hearing.) ◯ Hard-of-Hearing (An individual who is hard-of-hearing has some usable hearing and may have mild to severe hearing loss.) ◯ deaf (An individual who is deaf is defined as having severe to profound hearing loss.)
Do you use a form of hearing assistance device?	◯ Cochlear Implant ◯ Hearing Aid ◯ Both ◯ None
Do you consider yourself part of the Deaf community?	◯ Yes ◯ No
Do you consider yourself part of the Hearing community?	◯ Yes ◯ No

The following questions will ask about any children or relatives with hearing loss/deafness.	
Do you have children?	◯ Yes ◯ No
How many children do you have?	_____
Do you have one or more children who were born deaf/hard-of-hearing or developed hearing loss early in childhood (under 8 years old)?	◯ Yes ◯ No
How many of your children are deaf or hard-of-hearing?	_____
How many of your children are hearing?	_____
Does your child use any form of hearing assistance device?	◯ Cochlear Implant ◯ Hearing Aid ◯ Both ◯ My child does not use a hearing assistance device
Do you or your partner/spouse have any other family members who were born deaf or developed hearing loss early in childhood?	◯ Yes ◯ No
Please select which family members were born deaf or developed hearing loss early in childhood. (Select all that apply)	☐ Partner/Spouse ☐ Parent ☐ Brother/Sister ☐ Uncle/Aunt ☐ Grandparent ☐ Cousin

The following questions will ask about you and/or your child's hearing loss/deafness.	
What forms of communication do you use?	◯ Oral language only ◯ Sign language only ◯ Both oral and sign language
What forms of communication do you use with your child?	◯ Oral language only ◯ Sign language only ◯ Both oral and sign language
How old were you when you were diagnosed with hearing loss/deafness?	◯ Birth - 1 year ◯ 2 - 10 years ◯ 11 - 18 years ◯ 18+ years
How old was your child when they were diagnosed with deafness?	◯ Birth - 1 year ◯ 2 - 10 years ◯ 11 - 18 years ◯ 18+ years
Is your deafness associated with any other symptoms (i.e. vision loss, cleft palate, etc.) or part of a syndrome?	◯ Yes ◯ No
Is your child's deafness associated with any other symptoms (i.e. vision loss, cleft palate, etc.) or part of a syndrome?	◯ Yes ◯ No
Please specify the symptoms associated with your deafness.	_____
Please specify the symptoms associated with your child's deafness.	_____

The following questions will ask about any experience with deaf genetic testing or genetic counseling.	
Have you ever had genetic testing to find out if there is a genetic explanation for why you are deaf?	◯ Yes ◯ No
Has your child ever had genetic testing to find out if there is a genetic explanation for why he/she/they is deaf?	◯ Yes ◯ No
Who ordered the genetic testing?	◯ Primary Care Doctor ◯ Pediatrician ◯ ENT Doctor ◯ Genetics Doctor ◯ Genetic Counselor ◯ Audiologist ◯ Other Health Provider ◯ I ordered genetic testing on the internet
Please specify what type of health provider ordered the genetic testing.	_____
Please specify the name of the company you ordered genetic testing from.	_____
How old were you when you had genetic testing?	◯ Prenatal (before birth) ◯ Childhood (0-9 years) ◯ Adolescence (10-17 years) ◯ Adulthood (18+ years)
How old was your child when he or she had genetic testing?	◯ Prenatal (before birth) ◯ Childhood (0-9 years) ◯ Adolescence (10-17 years) ◯ Adulthood (18+ years)
What were the results of the genetic testing?	◯ Test result explained the cause of deafness ◯ Test result did not explain the cause of deafness ◯ Test result was inconclusive/unclear as to whether or not it explained the cause of deafness ◯ I'm not sure what the results were
Were you offered genetic counseling before or after having genetic testing? (Genetic counseling is the process of advising individuals and families who are affected by or at risk of genetic conditions to help them better understand the medical, psychological and familial implications.)	◯ Yes ◯ No ◯ I'm not sure
Would you have been interested in genetic counseling if it had been offered?	◯ Yes ◯ No ◯ I'm not sure
Did you have genetic counseling before or after genetic testing?	◯ Before testing ◯ After testing to explain the result ◯ Both before and after testing ◯ I did not have genetic counseling

Who provided the genetic counseling?	◯ Primary Care Doctor ◯ Pediatrician ◯ ENT Doctor ◯ Genetics Doctor ◯ Genetic Counselor ◯ Audiologist ◯ Other Health Provider
Please specify who provided the genetic counseling.	_____
Do you feel that the genetic testing was clearly explained to you before doing the testing?	◯ Yes ◯ No ◯ Unsure

How did you feel after receiving your genetic test results?					
	Very Slightly or Not at all	A Little	Moderately	Quite a Bit	Extremely
Interested	○	○	○	○	○
Distressed	○	○	○	○	○
Excited	○	○	○	○	○
Upset	○	○	○	○	○
Strong	○	○	○	○	○
Guilty	○	○	○	○	○
Scared	○	○	○	○	○
Hostile	○	○	○	○	○
Enthusiastic	○	○	○	○	○
Proud	○	○	○	○	○
Irritable	○	○	○	○	○
Alert	○	○	○	○	○
Ashamed	○	○	○	○	○
Inspired	○	○	○	○	○
Nervous	○	○	○	○	○
Determined	○	○	○	○	○
Attentive	○	○	○	○	○
Jittery	○	○	○	○	○
Active	○	○	○	○	○
Afraid	○	○	○	○	○

How did you feel after receiving your child's genetic test results?					
	Very Slightly or Not at all	A Little	Moderately	Quite a Bit	Extremely
Interested	○	○	○	○	○
Distressed	○	○	○	○	○
Excited	○	○	○	○	○
Upset	○	○	○	○	○
Strong	○	○	○	○	○
Guilty	○	○	○	○	○
Scared	○	○	○	○	○
Hostile	○	○	○	○	○
Enthusiastic	○	○	○	○	○
Proud	○	○	○	○	○
Irritable	○	○	○	○	○
Alert	○	○	○	○	○
Ashamed	○	○	○	○	○
Inspired	○	○	○	○	○
Nervous	○	○	○	○	○
Determined	○	○	○	○	○
Attentive	○	○	○	○	○
Jittery	○	○	○	○	○
Active	○	○	○	○	○
Afraid	○	○	○	○	○

The following questions will ask you about any medical decisions made as a result of genetic testing.	
Did you make any decisions regarding your audiologic or medical care as a result of your genetic test results?	☐ Assistive hearing device was placed (i.e. Cochlear implant, hearing aid) ☐ Additional lab testing or imaging was ordered (i.e. blood test, CT or MRI scan, thyroid testing, heart examination, etc.) ☐ Referral was made to see another doctor or health provider ☐ Other ☐ None of the above
Please specify what other medical decisions were made as a result of your deaf genetic test results.	_____
Did you make any decisions regarding your child's audiologic or medical care as a result of your child's genetic test results?	☐ Assistive hearing device was placed (i.e. Cochlear implant, hearing aid) ☐ Additional lab testing or imaging was ordered (i.e. blood test, CT or MRI scan, thyroid testing, heart examination, etc.) ☐ Referral was made to see another doctor or health provider ☐ Other ☐ None of the above
Please specify what other medical decisions were made as a result of your child's deaf genetic test results.	_____

The following questions will ask about any language or educational decisions made as a result of your child's genetic testing.	
Did you make any decisions regarding your child's language development as a result of your genetic test results?	◯ Focus on sign language (e.g. American Sign Language) ◯ Focus on oral language (e.g. English [reading; writing; and/or oral]) ◯ Focus on both sign and oral language (e.g. American Sign Language and English [reading; writing; and/or oral]) ◯ Other ◯ None of the above
Did you make any decisions regarding your child's language development as a result of your child's genetic test results?	◯ Sign language focus (e.g. American Sign Language) ◯ Oral language focus (e.g. English [reading; writing; and/or oral]) ◯ Bilingual language focus (e.g. American Sign Language and English [reading; writing; and/or oral]) ◯ Other ◯ None of the above
Please specify what other decisions were made regarding your child's language development.	_____
Did you make any decisions regarding your child's education as a result of your genetic test results?	◯ School with mainly oral instruction without an interpreter or support services ◯ School with mainly oral instruction with an interpreter or support services ◯ School with mainly sign instruction ◯ Other ◯ My child is not yet school age
Did you make any decisions regarding your child's education as a result of your child's genetic test results?	◯ School with mainly oral instruction without an interpreter or support services ◯ School with mainly oral instruction with an interpreter or support services ◯ School with mainly sign instruction ◯ Other ◯ My child is not yet school age
Please specify what other decisions were made regarding your child's education.	_____

The following questions will ask you about any decisions made regarding family planning or partner/spouse selection as a result of genetic testing.	
Did your genetic test results influence your decisions regarding family planning? Family planning refers to how a person or couple thinks about and plans the number of children they want to have and how long they want to wait between pregnancies.	◯ Yes ◯ No
Did your child's genetic test results influence your decisions regarding family planning? Family planning refers to how a person or couple thinks about and plans the number of children they want to have and how long they want to wait between pregnancies.	◯ Yes ◯ No
Please select which of the following decisions regarding family planning were influenced as a result of your genetic test results.	◯ I/we decided to have (more) children ◯ I/we decided not to have any (more) children ◯ I/we decided to have prenatal genetic diagnosis ◯ I/we decided to have genetic carrier testing ◯ Other ◯ None of these
Please select which of the following decisions regarding family planning were influenced as a result of your child's genetic test results.	◯ I/we decided to have (more) children ◯ I/we decided not to have any (more) children ◯ I/we decided to have prenatal genetic diagnosis ◯ I/we decided to have genetic carrier testing ◯ Other ◯ None of these
Please specify what other decisions were made regarding family planning.	_____
Did your genetic test results influence your decisions regarding whom you choose to be your spouse or partner?	◯ Yes ◯ No
Please select which of the following decisions regarding partner/mate selection were influenced as a result of your genetic test results.	◯ Choosing a partner who is also deaf or hard of hearing ◯ Choosing a partner who is hearing ◯ Having my partner get deaf genetic testing ◯ Other
Please specify what decisions were made regarding partner/mate selection.	_____

The following questions will ask about your experience with genetic testing and genetic counseling.	
Thinking about your experience with genetic counseling, would you recommend someone in a similar situation receive genetic counseling to learn more about how hearing loss/deafness can be inherited in some families and to talk about what this means to them?	◯ Yes ◯ No ◯ Unsure
Thinking about your experience with genetic testing, would you recommend someone in a similar situation have genetic testing for deafness?	◯ Yes ◯ No ◯ Unsure
Please add any additional comments about your experience with genetic testing for deafness and/or genetic counseling.	_____

APPENDIX C: Request for Survey Distribution

To Whom It May Concern,

My name is Alaina Heinen, and I am a second-year genetic counseling student at the University of California, Irvine. I am currently working on a thesis surrounding the experiences of adults and parents of children who are deaf/hard-of-hearing that have had genetic testing. I have created an anonymous online survey for participants to complete that will ask questions about their experiences, how they felt about the testing and what decisions were made as a result. Due to COVID-19 restrictions, I am recruiting participants through online organizations, advocacy groups and community social media pages. I am reaching out to see if my survey (either the link below or the attached handout) could be posted on your organizations' webpage or distributed to your listserv to increase exposure to potential participants.

Survey Link: https://is.gd/deafgenetictesting

Thank you very much for your time, I look forward to hearing from you!

APPENDIX D: Recruitment Flyer

GENETIC TESTING FOR DEAFNESS:

A Study on the Impact of Genetic Testing for Deaf Adults and Parents of Deaf Children

SURVEY LINK: **https://is.gd/deafgenetictesting**

Have you or your child had genetic testing for deafness?
If so, we want to hear from you!

This study is exploring the impact that genetic testing for deafness has on deaf individuals and parents of deaf children. It will ask about your experiences with genetic testing for you and/or your child. The information gathered from this study will provide healthcare professionals, such as genetic counselors, deeper insight into how individuals and families are impacted by genetic testing and how they utilize their results.

Please visit the link above to participate in a short confidential survey! To be eligible for the survey you must be 18 years old, live in the United States and:

> Are an adult who is deaf or hard-of-hearing
OR
> Are the parent of a child who is deaf or hard-of-hearing

Please feel free to contact me with any questions.

CONTACT INFORMATION
Attn: Alaina Heinen
Phone: (714) 456-5837
Fax: (714) 456-5330
Email: aheinen@hs.uci.edu

APPENDIX E: Full List of Open-Ended Responses

“We were told to get testing to help determine if CI was a better plan for single sided deaf child, due to inconclusive results they refused a CI as unnecessary even as we begged for it.”
“This was very important to us in our journey. We continue to see a geneticist every year to make sure his diagnosis has not changed.”
“While we did not get a definitive answer as to my child's unilateral hearing loss, it was informative to know she didn’t have any syndromes and to also know what she is a carrier.”
“In my opinion it was done to give my child a diagnosis, when result came back unresolved he was not longer an candidate is what it felt like to me. He has slips through the parish since no true diagnosis was determined. It seemed be easier to blame issues on diagnosis then none not having fun.”
“Genetic testing was for other purposes, but did rule out deafness.”
“My son is now 21 years old and native in ASL. 21 years ago testing was very expensive at the time, so we chose to just accept the deafness. After spending years in the Community and learning the differences of Deafness, my ENT agrees that I may show signs of Wardenburg Syndrome {WS} (the worst of the Syndrome is cochlea deafness once the gene mutates so often) IF it is WS then his children will have a 50/50 chance of being Deaf. I know this is an anonymous survey - I'm willing to talk if you have more questions – [NUMBER REDACTED].”
“I think it's crazy that there is a variety of companies who test for different things, and our geneticist had to use probabilities in order to determine which to choose since we were having our insurance cover the cost. I think knowing that up front was eye-opening and helpful. This isn't quite the exact science / binary decision I expected. Also - I was warned up front that genetic testing would possibly explain the cause, which would be good for her to know when she's older. But if nothing came back, we were reminded that our focus should still be on what to do about her hearing loss. I think learning about the hearing loss in the first place is devastating. Expecting that it might be due to CMV, which might then impact her thyroid later in life is very concerning. If genetics were a cause in our case, I would probably have very different feelings and would want counseling.”
“Prior issues with our children prompted us to have genetic testing to see if hearing loss was part of a syndrome. We had or son [NAME REDACTED] pass away at 8 months old with a very vague explanation and both of our other children have a mildly dilated aortic root.”
“The Results explained that my deafness was genetics. I have several family members who are deaf too. This is one reasons why I took the genetic test to see if I could have deaf children or not. The results said I have "second generation skips".”
“My daughter was a high risk pregnancy due to intrauterine growth restriction. She was born slightly premature at 36 weeks weighing only 4 lbs 5 oz. other than her hearing and growth we have no that diagnosis or answers as to the cause of her hearing loss.”
“Genetic testing had several functions for our family. It quelled any concerns about syndromic issues possibly related to his deafness, and eliminated any concerns about environmental things that may have other adverse health effects. It also allowed us to inform our other child and other family members about the trait they may carry so that they can be prepared to have a deaf child in the future.”

“We were told there was a mutation on a gene that might be linked to hearing loss but they are unsure if that is the cause. I feel like it's a lot of work and even the Drs are unsure when you do get results.”
“I am currently pregnant with my 2nd child and my gynecologist recomended the gentetic test for my deaf son to see what the chances are of the 2nd child being deaf and he mentioned that my time was running out for termination. It made me very upset and I put in a complaint to the hospital.”
“I only got testing because of my vision. I personally would never have been tested for my hearing it not for the vision issues. I personally don't believe that hearing loss should a) warrant genetic testing or b) a decision not to have children. All my decisions are made because of my vision, not the hearing loss.”
“I am not sure if I would necessarily recommend someone get genetic testing for deafness unless they were concerned there was some underlying issue. I don't really think deafness in and of itself is something that needs genetic testing to ascertain. It would depend on the situation. But I am a huge proponent of genetic testing in general. Financial ability notwithstanding, I would probably encourage anyone that was curious to do it.”
“Knowing there was a genetic reason for my children's deafness, AND knowing that it was non-syndromic was such a relief for me. It gave me closure, and actually helped me feel more connected to their deafness and bonded as a family since we now know both my husband and I passed the same genetic mutation down to them. I find genetics fascinating, and hope to learn more as my children get older and become interested.”
“While it never mattered to me if my child was Deaf or not, I was glad to know it was a possibility so that I could be prepared (learning ASL, ensuring we had appropriate local resources, etc.)”
“I had genetic testing because of my retinitis pigmentosa.”
“SPARK labs gave the Usher 2a result, heterozygous. An earlier 23andMe test, with raw data run through Promethease, gave more info: Usher 1d, 2a, GJB2. No counseling, what I know is basic genetics taken in college.”
“Our primary concern with testing was initially determining if our son had syndromic deafness and whether we needed to make any other medical interventions on his behalf. When his deafness was revealed to be non-syndromic, we moved on to other issues. Secondarily, it helped us discover the trait's heritability, and inform other members of our family that they may carry the trait. Unexpectedly, I felt a small sense of relief that there were no environmental factors in our son's deafness that we could have avoided. Ultimately, given that we had Deaf relatives and ancestors, (great grandparents on my side) the overall picture of deafness in our family became clear, and knowing that our grandchildren or niece / nephews could also be deaf was a good thing to know and possibly prepare for in the future.”
“Appreciated the genetic counselors empathy and compassion when working with us. We didn't feel like a "number" or a "case." We were given initial results over the phone which I'm partially thankful for, so I was in my own home to process information vs in the office setting. (Others may feel differently) we later went in for further explanation, etc.”
“We participated in a genetic deafness study at Virginia Commonwealth University.”
“Historically it has been very difficult to access genetic testing due to high costs and lack of knowledge about genetic testing from providers offering care to these children; especially those that do not present with other indications of other syndromic causes. My kids are now teenagers and aren't ever seeing health care providers that would ever feel comfortable

ordering genetic testing i.e.; audiology, ENT. I think that it would be helpful to educate those providers on testing and encourage their patients/families to have testing done and provide resources on the best labs that offer testing. You can then set up a triage plan for them to utilizing genetic counseling if needed for follow-up. If children are older and they're not seeing a specialist that would openly discuss genetic testing, they wouldn't have access to testing unless a parent openly advocated and pushed for testing. I work in genetics for a commercial lab, and can attest that most providers from pediatricians, audiology, ENT's all look like deer in the headlights when you mention ordering genetic testing for kids. My girls were both part of the connexion 26 genetic research project at the University of UT about 19 years ago; both tested negative. I'd like to see Genetics Divisions recommunicate new offerings to families as availability of testing becomes more accessible. I imagine the triage of care and recommendations for genetic services are quite different for babies and young children receiving a diagnosis now, vs. what they have been in the past."
"Because our genetic testing didn't show it was a passed on, just a random birth defect and no known way to be sure if it was hemifacial microsomnia or just microtia atresia it didn't affect our choices we made once scans showed no other abnormal things linked with hemifacial microsomnia were present."
"My daughter had genetic testing in 2004 which did not show any genetic cause for her hearing loss. She was tested again in 2018 at the recommendation of a genetic counselor that we visited because her younger sister has a mutation in one TNXB gene causing symptoms similiar to Ehlers Danlos. We were there to see if she had hEDS like her sister. The genetic counselor recommended that we test her TNXB gene and do a complete genetic hearing panel. The diagnosis of USH2a was a complete surprise. She does not have hEDS. Two fully biological sisters, two completely different, rare genetic conditions."
"It was a largely positive and helpful experience, but because it was one component of a series of medical assessments, I felt overwhelmed and at times, numb, throughout the process."
"After having 2 children with hearing loss, we would really like to know more. It's good to know the cause. I fortunately we were never given gentetic testing results, as the blood work came back with a false positive for an infection and the ENT referred us out and we haven't heard back from the ENT who had the genetic testing done."
"My son's genetic testing was done more so for some other medical issues that are unrelated to him being deaf."
"Not interested in doing any more testing at this point. MRI showed nothing to be concerned about, all other developments seems normal."
"The genetic testing and counseling helped put a lot of concerns to rest and also shed a lot of light regarding Deafness in our family history."
"I had genetic testing when I was around 29 years old at Gallaudet and the dr believed that I had the features of Branchio-Oto-Renal Syndrome but could not "prove it" based on limited testing available. He recommended that I see a genetic counselor before having children. Fast forward twelve years later, I finally went to see a genetic dr. I haven't had any children but I had hoped to and I am 42 now so i feel like my biological clock is ticking. I went for genetic testing again at Tufts in Boston but they could not find any outcome. Tests were negative. I was told that 50% of the time they dont know what the genetic causes of deafness are even with the advances in science over the last ten years and my case falls into that category. So its disappointing to still not have answers but I may still take the risk of having children even tho

there's a 50% possible chance of passing it on to my children. At this point, there is just no way for me to know."
"We participated in a study at Virginia Commonwealth University"
"I was very overwhelmed with all the questions from my genetic counselor, I got very upset and cried and I felt pushed through interview without acknowledgement or a chance to catch my breath."
"Our youngest (13 year old) received the diagnosis of Usher syndrome type 1b in September 2020. This completely changes the path he will take for his career. It also requires more supports in the classroom. We have the opportunity to provide this for him while he is young to give him the most benefit. His siblings have also received testing and have USH 1b. We're scrambling to get them services. It would have been better to know while they were younger."
"Due to expense, we did not seek out genetic testing. What I had was requested through my opthalmologist since I knew I had retinitis pigmentosa."
"I think there is excellent reason for genetic testing and find it frustrating that it's difficult to get insurance coverage. My example is meaningful as I would not have known to take extra caution with certain medications which could ultimately lead to deafness for myself and for all my children, both the HOH and the hearing children. I know that if the mitochondrial gene that is suspected to be the cause of the loss is in fact the cause, then my 4 boys actually cannot pass that gene on to their children. So many other reasons it's important too (e.g. Usher syndrome, potential kidney issues, family planning, career planning for the individual, language modality choices if progression of the HL is likely, etc)."
"We had a genealogist who tried to see if there is a hereditary condition for the deaf. My four distant cousins and I are deaf and third cousins."