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Healthcare Satisfaction and Access to Genetic Counseling for Individuals with Disorders of Sex Development and Individuals Who Identify as Transgender

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Healthcare Satisfaction and Access to Genetic Counseling for Individuals with Disorders of Sex Development and Individuals Who Identify as Transgender

THESIS

submitted in partial satisfaction of the requirements
for the degree of

MASTER OF SCIENCE

in Genetic Counseling

by

Rebecca Louise Freeman

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Professor Maureen Bocian, MD, MS, Chair
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ABSTRACT OF THE THESIS

Healthcare Satisfaction and Access to Genetic Counseling for Individuals with Disorders of Sex Development and Individuals Who Identify as Transgender

By

Rebecca Louise Freeman

Master of Science in Genetic Counseling

University of California, Irvine, 2016

Professor Maureen Bocian, MD, MS, Chair

Individuals who identify as transgender and individuals who have been diagnosed with a disorder of sex development (DSD) likely have different challenges and experiences with the medical world. Both individuals with DSD as well as transgender individuals can be perceived as "gender non-conforming," although their non-conformity would be attributed to differing etiologies. This study was designed to collect feedback from both of these groups in order to clarify similarities and differences with respect to satisfaction with healthcare, access to Medical Genetics and Genetic Counseling, diagnosis disclosure, and perceived stigma. By forming a better understanding of these two groups, we can begin to improve medical care and how they are perceived by healthcare professionals. With this aim, an anonymous survey was distributed online and responses from 104 eligible individuals analyzed.

Using the patient satisfaction questionnaire (PSQ-18), our study population reported lower general healthcare satisfaction than other previously-studied groups. Fewer than one-third of all participants with DSD reported having seen a medical geneticist or genetic counselor, even though a Medical Genetics evaluation is indicated in most, if not all, cases of DSD. Forty percent of all survey respondents reported that a healthcare practitioner had been unwilling or unable to treat them due to their gender or medical condition (eg: DSD). The results of this study indicate the need for further broad educational and sensitivity efforts in healthcare organizations. Additionally, further improvement efforts are needed in order to increase access to Medical Genetics and Genetic Counseling for individuals with DSD.

I. INTRODUCTION

1.1 Defining transgender and disorders of sex development (DSD)

This thesis aims to describe healthcare satisfaction, access to Medical Genetics/Genetic Counseling, diagnosis disclosure, and perceived stigma in individuals who identify as transgender and individuals who have been diagnosed with a disorder of sex development (DSD). While both of these groups have their own distinct experiences, stories and challenges, for the purpose of this research both groups are considered gender non-conforming. In modern Western society, gender is presented as an “either/or” binary consisting of male and female identities, but both DSD and transgender individuals can be perceived as falling outside these norms.

Before one can begin to describe variation from gender norms, it would be best to establish a baseline understanding of gender and to explain how gender functions “normally.” While gender-specific behaviors appear to have a biological basis, gender is also understood as a process and ultimately as a social accomplishment.[1] According to the “Social Cognitive Theory of Gender Development and Differentiation,” gender is a primarily social process that begins in infancy and continues to develop across an individual’s lifetime.[2] This development is strongly influenced by social pressures, and variance from typical gender patterns is often met with negative social backlash, also known as “gender policing”.[3] It should be noted that there are many hypotheses concerning gender development, including essentialist, biologically-oriented perspectives that assert that gender is produced primarily from the reproductive roles of the sexes.[4] These “essentialist” theories are currently out-of-favor, but they retain some amount of influence in gender studies as a modern field. However, for the purposes of this thesis, gender and sex will be discussed through the lens of the social cognitive theory, and gender will often be described as an expression or “performativity,” using language established by leading gender theorist, Judith Butler.[5]

Although the two are related, sex and gender are most often described as separate traits; sex is distinguished by chromosomal, hormonal and physiological makeup, while gender is most often conveyed through appearance, behavior and other social performances.[1,5,6] It is not possible to see a person walking down the street and know his or her chromosomal, hormonal or physiological makeup (assuming this person is fully clothed), but we can immediately assess that person’s gender based on agreed-upon social clues.[7] In other words, in this hypothetical scenario of observing an individual walking down the street, we would be assessing for gender as opposed to sex. We often *infer* sex based on gender, but these two traits do not always align as we would expect.

This is the case with people who identify as transgender or who have a DSD. The term DSD was embraced by the medical community after the publication in 2006 of the “Consensus statement on management of intersex disorders” (hereafter referred to as the Chicago Consensus Statement), in which DSD is defined as an umbrella term for inborn medical conditions or “congenital conditions in which development of chromosomal, gonadal, or anatomical sex is atypical” (the biological basis of these conditions will be discussed in more detail below).[8] Before the Chicago Consensus Statement, a variety of non-standardized and often gender-based terminology was used to describe DSDs, which often offended patients and confused healthcare practitioners. These historical terms (hermaphrodite, pseudohermaphrodite, etc.) are still frequently seen in older literature, but the majority are now generally considered pejorative. The term “intersex,” however, is often considered acceptable and is widely used by DSD activists and individuals who are uncomfortable with the medicalization of DSD.[9] The term “intersex” is now used more as an expression of identity as opposed to a medical term, and many individuals with DSD do not consider themselves intersex.[10,]

The incidence of DSD/intersex is heavily debated, with some sources quoting an incidence as high as 1/100.[11] However, the Chicago Consensus Statement quotes an incidence of 1/4,500, so that frequency will be used for the purposes of this paper. To clarify, the 1/4,500 incidence refers to individuals born with external and/or internal genital anomalies that fall into the classification of ambiguous genitalia, which contradicts the binary model of sex.[8] This likely excludes individuals who develop features of DSD later in life (such as in late-onset adrenal hyperplasia) or individuals with diagnoses that are usually not considered part of the DSD spectrum (such as Turner syndrome).[12] The term "ambiguous genitalia" is often used to describe the genital phenotype of individuals with DSD, and it should be noted that this term represents a broad spectrum of phenotypes and is somewhat subjective.[13] Many of these cases are believed to have an etiology that is either partially or fully genetic, but discrete genetic conditions are identified in a minority of cases; some sources report that fewer than 20% of DSD patients receive a molecular diagnosis.[14] This discrepancy has been attributed to a variety of factors, including lack of access to quality molecular laboratories as well as a limited awareness of the value of a molecular genetic diagnosis for care and management of the patient.[15]

Individuals who identify as transgender differ from individuals with DSD in that in most cases they do not have an inborn medical condition. Transgender is an umbrella term for an individual whose gender identity differs from what he or she was assigned at birth.[16] An estimated 0.3% of population the United States identifies as transgender. Similar to DSD, there are many derogatory terms for transgender individuals, including some that were more acceptable historically. "Transsexual," for instance, is largely falling out of favor because it has implications about an individual’s surgical status that are now considered insensitive (see Table 1 for a more complete list of preferred and derogatory terms pertaining to individuals who are transgender and/or who have DSD).[17]

It should be clarified that it is possible to be both transgender and be diagnosed with a DSD, although typically an individual with a DSD does not identify as transgender, and transgender individuals are usually not diagnosed with DSD. However, individuals with DSD and transgender people can be seen as falling outside the typical dichotomy of male and female as it pertains to gender and sex. Therefore, for the purposes of this thesis, both groups are considered “gender non-conforming” in that they do not necessarily fit into completely separated male and female categories but instead may have some overlapping characteristics.

Term	**Definition**	**Possible Acceptable Synonyms**
Agender (adj)	Identifying as non-binary or gender neutral	Genderqueer, gender non-binary
Asexual (adj)	A complete lack of sexual desires	
Bisexual (adj)	Sexually attracted to members of both sexes	Bi
Cisgender (adj)	Having the same gender identity as was assigned at birth	
Cross-dress (v) or Cross-dresser (n)	To wear clothing, makeup and/or accessories usually reserved for the opposite gender	Drag king, drag queen
Disorder of sex development (n)	Congenital conditions in which development of sex is atypical	Intersex, DSD
FtM (adj), MtF (adj)	Acronyms to denote "Female to Male" or "Male to Female" transgender status	Trans man, trans woman, trans, transgender
Gender (n)	A primarily social expression of male or female (or other) identity	
Genderqueer (adj)	Identifying as outside the typical gender binary of male and female	Gender neutral, genderqueer, agender
Heterosexual (adj)	Sexually attracted to members of the opposite sex	Straight
Homosexual (adj)	Sexually attracted to members of the same sex	Gay, lesbian
Intersex (adj)	The state of being born with ambiguous genitalia or other DSD	DSD
Sex (n)	A primarily biological category that determines if a human is male or female based on hormones, physiology, chromosomes, etc	
Transgender (adj)	Having a different gender identity than was assigned at birth	Trans, trans man, trans woman, trans person
Transsexual (adj)	Another term for transgender but implies that the individual has had sex reassignment surgery(ies)	Transgender, trans, trans man, trans woman, trans person
Examples of derogatory terms to avoid: Tranny, she-male, he/she, transvestite, hermaphrodite, pseudohermaphrodite		

***Table 1**: **Preferred vocabulary** Terms and definitions relating to individuals who identify as transgender and individuals who have been diagnosed with DSD and/or identify as intersex.* [1,8,17]

1.2 Gender variance and social inequality

Before discussing gender social power dynamics, it is important to acknowledge intersectionality within any culture and to consider other factors contributing to social power, namely economic status, race, education, sexual orientation, age, etc. While much of the following discussion does not explicitly consider these other aspects, their influence on stigma and social maneuverability should not be understated. While gender is widely considered the most socially salient characteristic in modern Western society,[18-20] this, of course, does not invalidate the contributions of other social status markers.

Historically, so-called "feminine" attributes have been considered less desirable than "masculine" attributes. Indeed, in some cultures more feminine behaviors have been deemed abnormal because those cultures were founded on a masculine norm.[21] In more modern Western societies, there are still strong cultural preferences for masculinity, which is especially reflected in the often-violent reactions to masculine individuals performing feminine behaviors. For example, as a group, gay men receive more hostility than lesbian women, and male-to-female (MtF) transgender individuals experience more violence than female-to-male (FtM) transgender individuals.[20,22] Hostile reactions occur because these individuals are performing a larger social transgression by choosing to surrender their masculinity for femininity: gay men are rejecting heterosexuality (a hallmark of Westernized masculinity), and the MtF transgender individuals are becoming women. When viewed in this perspective, lesbians and FtM transgender individuals can even be considered as socially elevating themselves by performing masculinity. As another consequence of the strong preference for masculinity, boys must act under stricter gender roles than girls do;[23] it is much more socially acceptable for a little girl to be a "tomboy" than it is for a little boy to prefer feminine toys.[24-26] The emphatic need to be strong and masculine has a very real health consequence for men, who often delay seeking medical care for injury or illness by attempting to “man up” until care is absolutely necessary. As a result, men are more likely than women to die from treatable or preventable illness.[27]

The above points are very important to consider when designing research involving gender and healthcare. In transgender populations, researchers must consider FtM and MtF populations as distinct entities because the two groups can experience differing levels of violence and stigma due to the patriarchal nature of the societies within which they function. It is interesting to note that, to this author’s knowledge, all studies of DSD health except one[28] do not compare respondents’ data based on social sex. This is a major failing in the current literature, and re-analysis may reveal that certain populations have different needs than others. For example, similar to results in non-DSD populations, further research may reveal that male-identified DSD patients choose to postpone seeking medical care more often than female-identified DSD patients. Since gendered behaviors are largely socialized, we tend to expect that most male-identified DSD patients would follow typical male patterns of behavior and that most female-identified DSD patients would follow typical female patterns of behavior; furthermore, we would expect that there would be differences between those patterns similar to male-identified and female-identified individuals without DSD. While DSD groups are often analyzed as a whole, it is imperative to continue to look for differences within these groups as well. The gender of a patient should not be ignored just because that patient has a DSD. Gender remains the most socially salient characteristic in Western culture and has an impact on all aspects of an individual’s life.[18,19]

Since gender is such an important aspect of human identity, individuals with ambiguous genitalia have bodies that are, in the words of Judith Butler, “culturally unintelligible.”[29] This means that their bodies fall outside the scope of what our culture allows us to imagine, since our society constructs gender and biological sex as a binary: male or female. Due largely to this social contradiction, the medical treatment of individuals with DSD has a controversial and evolving history and has been influenced strongly by stigma.

One of the first publications on the care and management of DSD patients came from Dr. Morris at Yale in 1953, entitled "The syndrome of testicular feminization in male pseudohermaphrodites."[30] His recommendations include non-disclosure of the karyotype to adult patients, based on concerns prevalent at that time about the possible psychological ramifications and awareness of high levels of psychiatric morbidities in intersex conditions. He was concerned especially about the greatly increased medical care and attention towards DSD individuals and wanted to protect his patients from associated "psychiatric causality and suicide." This was accepted as ethical in the 1950's and conformed to the ideal of "therapeutic privilege," which allowed doctors to withhold medical information if they believed their patients could be psychologically harmed by a disclosure. This practice is now considered to be unacceptably paternalistic, but in the 1950's and 1960's, this form of nondisclosure was believed to be in keeping with the ethical concept of beneficence. Obviously, therapeutic privilege is no longer held in favor due to the rise in popularity of informed consent and patient autonomy.[31] However, nondisclosure of diagnosis continued to be debated and practiced at least until the 1990's.[32]

1.3 Stigma and its relevancy to transgender individuals and individuals with DSD

The word "stigma" originates with the Latin *stigma*, meaning a mark or brand, and the Greek *stizein*, meaning to tattoo.[33] The Latin *stigma* was used to describe brands or tattoos on an individual that were meant to distinguish him/her as socially tainted in some way—as a slave, criminal, or an otherwise undesirable person. Today, “stigma” is not used to describe physical evidence of discredit but rather to indicate social and behavioral evidences of deviance from social expectation.[34] While in the past, stigmata were physical marks, modern day stigmata can be behaviors or other non-physical traits as long as they present an observable discrepancy between expected social performance and actual social performance. A stigma is a characteristic(s) that prevents an individual from assimilating to current stereotypes and that causes him or her to be exposed to status loss and discrimination.[35] Not all stigmata are perceived with equal distaste, but every stigma confers a certain level of difficulty or even danger to its owner. A stigmatized individual would then be said to have stigma ascribed to him or her; in other words, this individual would have one or more of his or her characteristics identified as discreditable based on culturally-agreed upon norms. The study of stigma was popularized in modern academia largely through the work of Erving Goffman in his seminal 1963 paper, Stigma: Notes on the Management of Spoiled Identity. Especially since the 1970’s and 1980’s, a large amount of investigation has been dedicated to the phenomena of stigma in general, and the stigmatization of gender non-conforming individuals in particular has evolved into a well-studied area.

Upon reviewing current research conducted into the lives of transgender individuals, it immediately becomes apparent that transgender individuals often are surveyed in conjunction with individuals who identify as gay, bisexual or lesbian, in large part due to the collective identity fostered by the LGBT (lesbian, gay, bisexual, transgender) community (the LGBT community is now expanding to be widely known as LGBTQIA, which includes lesbian, gay, bisexual, transgender, queer and/or questioning, intersex and asexual individuals). However, while well-intentioned, this clustering is often inappropriate for a variety of reasons.

Firstly, gender identity is separate from sexual orientation, even though these two identities are often related. While it is widely known that not all homosexual or bisexual individuals identify as transgender (indeed, only a small minority do), it is also entirely possible for an individual to identify as both transgender and heterosexual. That is, gender identity indicates who a person is, while sexual orientation indicates who a person is attracted to.[36]

Secondly, statistically there are far more homosexual and bisexual individuals than there are transgender individuals. The most comprehensive estimation of the LGBT population reported in 2011 that approximately 3.5% of adults in the United States identify as gay, lesbian or bisexual, while only 0.3% identify as transgender.[37] In other words, while there are likely over 8 million gay, lesbian and bisexual adults, there are 700,000 transgender individuals in the United States. Thus, in any study where gay, lesbian and bisexual adults are compared to transgender adults, we would expect roughly 10 times as many LGB responses as we would receive transgender responses. This sampling bias could be harmful to any results produced, especially when all individuals are grouped together to compare against the general population, because the struggles common to all LGBT people or even just to the LGB portion would statistically outweigh those of transgender individuals. This is particularly concerning since it has been observed that transgender individuals experience disproportionately higher levels of discrimination, anxiety and violence due to stigma than individuals in either the general population or other sexual minority populations.[22,38-40] Thus, the following is a summary of the current literature that focuses solely on the stigma faced by transgender individuals, either compared with LGB groups or with typical populations. Studies that lump transgender experiences with LGB have been excluded.

Experience of stigma among transgender populations can be assessed using an adaptation of the Minority Stress Model,[41,42] which has been successfully implemented to demonstrate that "minority stressors," or the stigma associated with gender non-conforming, correlate with clinical depression and anxiety in transgender individuals.[43] While social support, often from other transgender peers or peers who identify as bisexual, gay or lesbian, may help to reduce the psychological impact, the negative effects from stigma remain.[38,44] A major negative effect of stigma is discrimination, defined as the process through which social, cultural, political and economic powers are applied to devalue stigmatized individuals.[45] Stigma, and its resulting discrimination, has been identified as a major cause for health gaps between general populations and minority groups.[46,47]

While the stigma ascribed by transgender status has been relatively well studied, there is minimal literature measuring stigma in DSD patients. Although DSD is frequently referred to as a threat to healthy psychological development, conclusive overall data are limited, and documentation of stigma in DSD individuals is largely confined to first-person accounts or case studies.

1.4 Transgender community satisfaction with and access to healthcare

As noted above, many studies have surveyed attitudes of health care providers toward the LGBT community as opposed to transgender groups in particular.[48] While few studies specifically target healthcare satisfaction for transgender individuals, the behavior patterns of these individuals are likely indicative of an overall lack of perceived personal safety and certainly of a lack of satisfaction with the healthcare industry.[49]

In 2010, the National Center for Transgender Equality and the National Gay and Lesbian Task force released the "National Transgender Discrimination Survey Report on Health and Health Care" (NTDS), which surveyed over 7,000 individuals who self-identified as transgender or as gender non-conforming.[49] The NTDS was a hugely successful national undertaking and was the most extensive survey of transgender discrimination to date.

Several conclusions from this study shine light on difficulties in satisfaction with, and access to, medical care for transgendered people. A majority of those surveyed had been able to access and utilize transition-related medical care, yet 33% of responding patients reported postponing preventative medical care due to discrimination, and 28% report postponing care even while sick or injured.[49] This delay is largely attributed to both a lack of financial access to medical care and issues surrounding discrimination by healthcare professionals. Research in general populations has shown that postponement of care is often due to a perceived limitation to access to care or a belief that a physician will not act in the patient's best interest.[50,51]

Of the transgender individuals who responded to the NTDS report, 19% indicated that they had been refused medical treatment based on their transgender/gender non-conforming status and 50% reported that it was necessary for them to teach their healthcare providers how to provide proper medical care for them.[49] More recent studies continue to demonstrate a lack of physician knowledge and preparedness to care for transgender patients.[52] Factors such as age and income also affected respondents' likelihood of experiencing discrimination in healthcare, with low-income and non-Asian/non-Caucasian transgender individuals being more likely to experience discrimination than Caucasian and/or higher-income transgender individuals. The impact of these socioeconomic factors has also been studied in non-transgender groups, with similar results.[53,54]

Having access to medical insurance gives transgender individuals more ability to afford routine medical care than those without. When measured as a single group, transgender individuals were less likely to have health insurance and were more likely to be unemployed than the general population and the LGB population.[55-57] Additionally, transgender individuals were less likely to have a primary care physician and access to general medical care.[56] These are troubling statistics when one considers that transgender people are at an increased risk for depression, suicide, and HIV infection.[58-60] These statistics would likely lead most people to expect transgender individuals to seek healthcare services more often than the general population, particularly if they are interested in pursuing medical transition, and yet they are the group that more often postpones and even avoids care.

Satisfaction with transgender healthcare is additionally impacted by the perception of counseling "gate-keepers."[61] In accordance with the World Professional Association for Transgender Health (WPATH) Standards of Care, version 7, extensive counseling is recommended and may even be required during the medical transition process.[62] This recommendation or requirement for counseling may incite resentment in some transgender individuals, just as some individuals resent the medicalization of transgenderism and the implication that it is pathologic.[63-65] The inclusion of gender dysphoria in the Diagnostic and Statistical Manual of Mental Disorders (DSM) continues to be an area of intense debate as critics weigh the potential benefits of increased likelihood of health insurance coverage and physician recognition against risk of categorizing transgender identities as pathological.[66]

It must be noted, however, that not all research in transgender health has demonstrated reduced healthcare satisfaction. While the NTDS report is the largest study to date in transgender healthcare, other data have been published. In a survey targeting transgender satisfaction with healthcare that drew on five consecutive surveys taken at a university-based sexual health clinic, transgender patients reported similar or higher satisfaction with their healthcare than the general population.[61] While the underlying cause of this high satisfaction was not specifically assessed, past improvement efforts were briefly acknowledged. It is, perhaps, not surprising that a medical clinic in a university setting (widely stereotyped as a more liberal and accepting atmosphere) would receive high patient satisfaction. The authors of the study noted similar limitations as with all healthcare satisfaction studies—namely, that these surveys are likely to result in high reported satisfaction.[67] This tendency has led many to question the utility and the meaning of data obtained from healthcare satisfaction surveys, although for many these surveys represent a practical way of eliciting feedback from patients about their own healthcare.[68,69] While data have shown that improvements can be made in transgender healthcare, it is encouraging to see positive statistics and feedback from the community.

1.5 DSD community satisfaction with and access to healthcare

While substantial literature has been published on the satisfaction of individuals with DSD in their healthcare, a majority of this work has emphasized satisfaction with sexual function, psychosexual outcome, and outcomes of corrective genital/gonadal surgeries as opposed to satisfaction with the overall healthcare experience.[70] Since this type of surgery is both medically complex as well as socially controversial, it is not surprising that there would be an emphasis on these procedures in the medical literature. Likewise, sexually-charged aspects of medical outcomes of DSD patients, such as gender identity and sexual orientation, are attention-grabbing and frankly intriguing, and they will likely continue to be areas of interest in research about individuals with DSD. It is interesting to note that much less emphasis has been placed on surgical management of individuals who identify as transgender, although the surgeries can be similar in some instances.

Women with congenital adrenal hyperplasia (CAH; a group of genetic disorders, each characterized by a deficiency of one of the enzymes needed to make the hormone cortisol, that can cause virilization in affected females) are most often the focus of long-term DSD studies, in large part due to the relatively high frequency of the disorder (1/10,000 – 1/15,000 in the US and Europe).[71] Even research in this group has only recently begun to focus on more holistic topics, such as quality of life (QoL) and health-related quality of life (HRQoL) measures. Critics of current DSD literature assert that poor psychosocial and psychosexual outcomes in DSD patients may be due more to personal struggles in society and medicine than to surgical outcomes and hormonal exposures (both of which have received far more academic attention).[72]

Certainly, the importance of the psychosocial aspects of healthcare and more routine procedures should not be underestimated in the care of affected patients in general. As previously acknowledged, it would be remiss not to mention that some researchers question the utility of measuring patient satisfaction, especially since evidence suggests that patient satisfaction in healthcare may depend more on outside societal factors than on the healthcare itself.[73] Nonetheless, patient satisfaction with care remains a well-studied area because these surveys are some of the only devices that allow patients to express concerns or contentment.[74]

Although general healthcare satisfaction is not often surveyed in DSD populations, a recent publication by Thyen, et al details the responses of 110 adults with DSDs in four medical centers in Austria, Germany and Switzerland. Respondents were surveyed regarding their satisfaction with care, health-related quality of life (HRQoL), psychological symptoms and gender identity.[75] More than half of these participants scored below the established cut-off, indicating a low perceived quality of care. Not surprisingly, individuals with more common DSDs reported higher healthcare satisfaction than those with rare disorders; these results mirror previous studies that found that patients with rare diseases in general have lower levels of healthcare satisfaction than the typical population.[76] It should be emphasized that individuals who reported difficulty accessing care also reported significantly lower satisfaction, whereas access to psychological counseling and appropriate follow-up appear to be correlated with higher patient satisfaction and well-being.[75,77]

Thyen, et al also determined that understanding information about an individual's diagnosis was an important predictor of satisfaction with care, and additional research revealed a similar result for parents of children with DSD.[78] Thyen, et al found that at the time of disclosure of diagnosis, patients who understood the information fully or even partially reported significantly higher satisfaction than patients who did not understand their diagnosis. Notably, the circumstances and the type of health care provider who gave the diagnosis did not seem to affect patient satisfaction; the main determinant seemed to be patients' comprehension rather than the process itself.

To return to the topic of genital reassignment surgery, Thyen, et al found that positive surgical results in terms of appearance and sexual functioning were correlated with higher satisfaction. There is a long history of mismanagement of care of the DSD population, particularly concerning surgical management, as discussed in more detail at the beginning of this section. This is important to reiterate here, because the care and management of individuals with DSDs has changed considerably over time. This variation will inevitably impact patients' satisfaction with healthcare as well as their relationships with the healthcare system as a whole;

for example, a 60-year-old woman with virilizing CAH will likely have drastically different opinions of healthcare providers than an 18-year-old woman with the same diagnosis.

In a significant forward step in the modern management of DSD, the 2006 Chicago Consensus Statement not only established medical terminology for those affected with DSDs but also developed clinical management guidelines, directly quoted below:[8]

- Gender assignment must be avoided before expert evaluation in newborns;
- Evaluation and long term management must be carried out at a center with an experienced multidisciplinary team;
- All individuals should receive a gender assignment;
- Open communication with patients and families is essential, and participation in decision making is encouraged;
- Patient and family concerns should be respected and addressed in strict confidence.

Based on previous research, the Chicago Consensus Statement emphasized the need for a multidisciplinary healthcare team, including members of pediatric specialties in the following areas: endocrinology, urology, psychology and/or psychiatry, gynecology, genetics, neonatology, social work, nursing and medical ethics.[79] This team is encouraged to educate other health care staff as well as family members in the care of each newborn with DSD.

1.6 A brief review of existing literature on patients with DSD and transgender patients regarding Medical Genetics and Genetic Counseling

According to the National Society of Genetic Counselors (NSGC), Genetic Counseling is “the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease."[80] A Medical Geneticist is an individual with a medical doctorate (MD) that specializes in the diagnosis and treatment of genetic conditions. Genetic Counselors and Medical Geneticists both practice within the broader field of Medical Genetics which is defined as the study and treatment of diseases that are etiologically, partially or entirely, genetic.[81]

While many DSDs have a genetic etiology, only a minority of individuals with DSD receive a molecular diagnosis (some estimate as low as 20%).[14] Medical and social management of DSD is an evolving practice, although some patients continue to feel dissatisfied with healthcare.[9,82] Medical Genetics services and Genetic Counseling are recommended components of the medical management of patients with DSD,[8,83] but there has not been much research to evaluate access to genetics services within the DSD community. Medical Genetics technology is evolving rapidly, and the introduction of broad multi-gene panels and whole exome sequencing (WES) will likely increase the diagnostic yield in DSD populations. While this should benefit individuals with DSD,[15] it cannot be realized without adequate access to genetics services.

Although there have been excellent publications on psychological counseling for transgender individuals,[84] to the author's knowledge, this is the first study surveying the experience of transgender patients in the area of Medical Genetics and Genetic Counseling. There have been several studies into the role of Genetic Counseling in the LBGT community,[85-87] but none of them actually surveyed anyone who identified as transgender. As discussed above, transgender individuals are often closely associated by others with homosexual and bisexual individuals due to the umbrella term LGBT and its resulting community. However, this association could be misrepresentative and possibly silencing.

There is no known genetic etiology for transgender identities. Be that as it may, a transgender individual could be referred to Medical Genetics/Genetic Counseling for multiple reasons, including those that commonly arise in the general population (e.g., family history of cancer, artificial reproductive technology, etc). Centers for assisted reproduction often counsel individuals who are interested in banking their gametes before undergoing hormonal therapies and/or surgeries during their gender transitions. These individuals should also be speaking with genetic counselors during these processes, although there is no current research into their experiences/access to genetics services.

There has been much more research exploring the experiences of individuals with DSD in Medical Genetics and in healthcare in general then there has been about transgender individuals. By far the most well-studied aspect of healthcare for individuals with DSD is genital surgical management and satisfaction with surgical outcome,[70,88,89] although more recently, data have been published about general healthcare satisfaction and quality of life (QoL) for individuals with DSD.[28,75,90] It is not surprising then, that there are few, if any, specific studies assessing the experiences of individuals with DSD in Genetic Counseling,[91] although most Medical Genetics publications pertaining to DSD emphasize the importance of Genetic Counseling and acknowledge its role within the multidisciplinary model recommended by the Chicago Consensus Statement. .[8,15,91,92]

1.7 Aims of this research

The purpose of this study is to identify potential barriers to healthcare for both transgender individuals and individuals with DSD and to determine if there are measureable differences between these groups. Consider a health care practitioner treating two patients—one diagnosed with type 1 diabetes as a child and one diagnosed with type 2 diabetes later in life due, at least in part, to a self-inflicted poor diet. Is it possible that these two patients would be treated differently based on their differing etiologies – namely, whether the diabetes was due to life habits or was the result of an autoimmune disorder? While individuals with DSD have unavoidable physical conditions, those who identify as transgender may be perceived as having choice in their gender non-conformity. I hope to determine whether these differing etiologies of gender non-conformity result in differences in healthcare satisfaction, access to genetics services, and methods of disclosure for diagnoses relating to the patient's sex or gender. While individuals who have a DSD and who also identify as transgender are uncommon, the survey is designed to identify such individuals in order to minimize potentially confounding situations. Ideally, this study will serve as a starting point for additional research into gender non-conforming individuals (of any etiology) and their experiences with genetic counselors. Genetic counselors serve a unique role as medical educators and could potentially help to reduce stigma as well as better serve the medical needs of these populations.

There are several hypotheses driving this research:

1. Individuals with DSD will report higher healthcare satisfaction than individuals who are transgender.
2. Male individuals with DSD and MtF transgender individuals will report lower satisfaction with healthcare than female individuals with DSD and FtM transgender individuals.
3. Respondents with DSD will report that they have received a medical diagnosis relating to their sex or gender more often than transgender groups.

4. Transgender individuals will report greater difficulty accessing Medical Genetics services, possibly attributed to lack of indication for referral.
5. Transgender individuals will report greater stigma.

II. METHODS

This study was reviewed and classified as exempt research by the Institutional Review Board (IRB) of the University of California at Irvine (HS# 2015-2528).

2.1 Recruitment

Participants were recruited to partake in an anonymous online survey hosted through SurveyMonkey, a web-based survey development software. The url "genderhealthcaresurvey.com" was used to redirect to the survey hosted on the SurveyMonkey site. None of the researchers, including the lead researcher and the faculty sponsor, had direct contact with any of the participants, and no participant identifying information was collected. All recruitment was done through social media and through the emailing lists of participating transgender or DSD support groups in the United States. A link to the survey was posted on the lead researcher's Facebook and Twitter accounts as well as on Facebook groups specific to transgender and/or DSD populations, such as "Trans*," "The Science of Sex and Gender," and "AIS DSD Support Group." Some support groups, such as AIS-DSD, San Diego Transgender Alliance, Gaylesta, and Santa Rosa LGBTQ, sent the survey link to their email lists. Electronic flyers were created, posted on social media, and sent to these support groups as well.

2.2 Participation

The participant population consisted of individuals with DSD or who identified as transgender and who lived in the United States, were at least 18 years of age, and chose to participate. Participants were able to self-identify within these categories. Any individual who did not select transgender or DSD identifiers or who was under 18 years of age was automatically disqualified from the survey. The online survey required Internet access in order to participate. Additionally, since the survey was only available in English, participants were required to read, understand and respond in the English language. All other social, economic and population characteristics were accepted, and there was no discrimination based on race, religion, employment or education.

Participants were not asked to disclose any identifying information, and their IP addresses were not recorded in order to ensure anonymity. While few foreseeable risks were associated with the study information sheet, the possibility for emotional disturbance was acknowledged, and national, toll-free hotlines (the Trevor Lifeline and the GLBT hotline) were provided in case any participants desired a resource to process these feelings. All data collected for this research study was stored in a password-protected computer in order to keep everything secure and confidential.

Participants were offered the chance to provide email addresses that would be used for a raffle for one $20 Amazon gift card. Participants were informed that research participation was not required to be eligible for the drawing, winning a prize was not guaranteed, and the chance of winning was approximately 1 in 200. Participants were informed that the raffle was optional, and they were not required to provide an email unless they wanted to participate in the raffle. These emails were collected using a separate, single-question survey hosted on SurveyMonkey that was not connected with any responses from the research survey. The emails were stored in a separate, secure database and were destroyed immediately after the raffle.

2.3 Obtaining informed consent

Informed consent was obtained through the use of an IRB-approved study information sheet (SIS) that was displayed to participants as the first page of the online survey. Clicking “next” indicated consent as a research participant. The SIS included information concerning the purpose of the research study, possible risks and benefits, and eligibility requirements. The SIS additionally informed participants that they could exit the survey at any time and that if there were any questions or concerns, the lead researcher or faculty sponsor could be contacted. Raffle information was included in the SIS, and participants were informed that they would have the option of providing an email to be entered into a drawing for one $20 Amazon gift card. Additionally, the SIS described the potential benefit to the DSD and/or transgender communities in that research results could help clarify potential areas for improving healthcare satisfaction or access to Medical Genetics and Genetic Counseling for either or both populations.

2.4 Survey construction

The survey consisted of three portions: 25 multiple-choice questions, 3 free-response questions, and 18 previously-validated, Likert scale questions from the Patient Satisfaction Questionnaire Short-Form (PSQ-18).[93] The multiple-choice and free-response questions were created by the research team for the purposes of this study. The data collected from the previously-validated PSQ-18 related to satisfaction with healthcare, while the multiple-choice questions solicited information including personal demographics, diagnosis disclosure methods, access to genetics services, and perceived stigma. The three free-response questions were included to elicit suggestions for healthcare improvement and to obtain more detailed responses concerning potential stigmatizing experiences. The survey used skip-logic technology, which allowed participants to answer follow-up questions if indicated and to skip to the next section if not.

A DSD support group asked for slight modification to the survey language after distribution had commenced (15 responses from transgender individuals had already been collected). Terminology in the survey was modified in order to reflect more sensitive language for participants—specifically, “disorder of sex development” was changed to “difference of sex development,” and in one question, “diagnosis” was changed to “medical condition.” In the study information sheet, SIS, risks were changed from “no perceivable risk” to include a risk for emotional disturbance. Two national hotlines were included as a resource for any negative feelings generated through survey participation. An additional question was also added to inquire about gender expression (see question 12, Appendix A). In addition, the options of “Both” and “Neither” were added to all gender identity categories. Since these modifications were considered minor, the original 15 responses were included in the final data analysis.

2.5 Statistical methods

Participants were assigned to either “transgender” or “DSD” groups based on their response to the initial identifier question. While a majority (90%) selected either “transgender” or “DSD,” some participants chose to specify their identities further utilizing the “other” category with free-response. These participants were manually sorted into “transgender” or “DSD” categories based on their responses.

Data analysis was conducted using the statistical software, Statistical Package for Social Sciences (SPSS). Continuous variables were compared between subgroups of interest using two group t-tests. Categorical variables were compared between subgroups using a Pearson chi-square test or Fisher Exact test. A p-value of less than 0.05 was used to indicate statistical significance. All tables and figures were produced using Microsoft Excel 2016. When necessary, due to low response numbers, categorical answers were combined for the purposes of statistical analysis as detailed in the results section.

III. RESULTS

3.1 Demographics analysis

In total, 141 participants began the survey. Of these, 106 were eligible to complete the survey based on age and self-selected identifier (DSD, transgender or "other"). Two additional participants were excluded from data analysis due to the selection of both DSD and transgender identifiers, so the study population consisted of 104 individuals (Table 2A). Forty-seven participants (45%) identified as DSD, while 55 (53%) identified as transgender (102/104 total). Twenty-six of the total 104 respondents selected "other" (Table 2B) in order to clarify or specify an identity, but most (24/26, 92%) selected either DSD or transgender in addition to "other." The remaining two participants were assigned to the "transgender group" or the "DSD group" based on their responses ("intersex" and "non-binary"). Forty percent of all respondents were between the ages of 18-30 years. Transgender individuals were generally younger than those with DSD and were more likely to select 18-25 years (Table 1, $p<0.001$). The majority of respondents were non-Hispanic white/Caucasian (83%). Other selected ethnic groups included Latino, Hispanic or Spanish (8%), "Other" (6%), Asian (2%) and Native American or Alaskan Native (2%). Due to small sample sizes, non-Caucasian ethnic groups were combined in order to compare ethnicity. For the same reason, age categories were combined into the following categories: 18-25 years, 26-30 years, 31-40 years, 41-50 years, 51-60 years, and over 60 years. When analyzing medical insurance, Medicare and Medicaid were combined, and "I don't know" and "Other" were combined. Ethnicity—Caucasian v non-Caucasian—did not differ significantly between subgroups defined by gender identity ($p=0.713$) or transgender vs DSD identifier ($p=0.873$). Ninety percent (93/103) of all participants completed at least some college; no significant differences in education were measured between DSD and transgender groups.

Demographics	**Total**		**DSD Group**		**Transgender**		**p-value**
	n	**%**	**n**	**%**	**n**	**%**	
Self-Identifier**							N/A
DSD	47	45%	42	88%	0	0%	
Transgender	55	53%	0	0%	50	89%	
Other	26	25%	9	19%	12	21%	
Total	*104****		*48****		*56****		
Age							<0.001*
18-25 years	**23**	**22%**	**2**	**4%**	**21**	**38%**	
26-30 years	**19**	**18%**	**5**	**10%**	**14**	**25%**	
31-40 years	**19**	**18%**	**9**	**19%**	**10**	**18%**	
31-35 years	10	10%	4	8%	6	11%	
36-40 years	9	9%	5	10%	4	7%	
41-50 years	**12**	**12%**	**8**	**17%**	**4**	**7%**	
41-45 years	5	5%	5	10%	0	0%	
46-50 years	7	7%	3	6%	4	7%	
51-60 years	**19**	**18%**	**15**	**31%**	**4**	**7%**	
51-55 years	5	5%	3	6%	2	4%	
56-60 years	14	13%	12	25%	2	4%	
Older than 60 years old	**12**	**12%**	**9**	**19%**	**3**	**5%**	
61-65 years	5	5%	4	8%	1	2%	
66-70 years	2	2%	2	4%	0	0%	
71-75 years	4	4%	3	6%	3	5%	
Older than 75 years old	1	1%	0	0%	0	0%	
Total	*104*		*48*		*56*		
Ethnicity							0.837*
Non-Hispanic	**86**	**83%**	**40**	**83%**	**46**	**82%**	
Non-Caucasian	**18**	**17%**	**8**	**17%**	**10**	**18%**	
African American	0	0%	0	0%	0	0%	
Latino, Hispanic or Spanish	8	8%	1	2%	7	13%	
Asian	2	2%	1	2%	1	2%	
Native Hawaiian or Pacific	0	0%	0	0%	0	0%	
Middle Eastern	0	0%	0	0%	0	0%	
Native American or Alaskan	2	2%	2	4%	0	0%	
Other	6	6%	4	8%	2	4%	
Total	*104*		*48*		*56*		
Highest level of education							0.237
High School	10	10%	4	9%	6	11%	
Associate's Degree or some	27	26%	9	19%	18	32%	
Bachelor's Degree	34	33%	15	32%	19	34%	
Post-baccalaureate education	32	31%	19	40%	13	23%	
Total	*103*		*47*		*56*		

Type of medical insurance							0.603*
PPO	**42**	**41%**	**21**	**44%**	**21**	**38%**	
HMO	**20**	**19%**	**12**	**25%**	**8**	**15%**	
Private provider	**12**	**12%**	**5**	**10%**	**7**	**13%**	
Medicare/Medicaid	**11**	**11%**	**4**	**8%**	**7**	**13%**	
Medicare	6	6%	3	6%	3	5%	
Medicaid	5	5%	1	2%	4	7%	
I don't have medical	**4**	**4%**	**1**	**2%**	**3**	**5%**	
Other/I don't know	**14**	**13%**	**5**	**5%**	**9**	**16%**	
I don't know	5	5%	1	2%	4	7%	
Other	9	9%	4	8%	5	9%	
Total	*103*		*47*		*56*		
Religion							<0.001*
Atheist/Agnostic	**43**	**42%**	**15**	**32%**	**28**	**51%**	
Christian/Catholic	**31**	**30%**	**23**	**49%**	**8**	**15%**	
Catholic	11	11%	9	19%	2	4%	
Christian	20	20%	14	30%	6	11%	
Other	**28**	**27%**	**9**	**19%**	**19**	**35%**	
Buddhist	9	9%	4	9%	5	9%	
Jewish	2	2%	1	2%	1	2%	
Other	17	17%	4	9%	13	24%	
Total	*102*		*47*		*55*		
Income							0.002*
Less than $25,000 per year	**26**	**25%**	**4**	**9%**	**22**	**40%**	
$25,000-$50,000	**25**	**25%**	**10**	**21%**	**15**	**27%**	
$50,001-$75,000	**15**	**15%**	**9**	**19%**	**6**	**11%**	
$75,001-$100,000	**17**	**17%**	**12**	**26%**	**5**	**9%**	
$100,001-$150,000	**8**	**8%**	**6**	**13%**	**2**	**4%**	
More than $150,000 per year	**11**	**11%**	**6**	**13%**	**5**	**9%**	
$150,001-$200,000	7	7%	3	6%	4	7%	
More than $200,000 per year	4	4%	3	6%	1	2%	
Total	*102*		*47*		*55*		
Relationship status							0.002*
Single	**48**	**46%**	**21**	**44%**	**27**	**48%**	
Married	**29**	**28%**	**21**	**44%**	**8**	**14%**	
Domestic partnership	**22**	**21%**	**5**	**10%**	**17**	**30%**	
Other	5	5%	1	2%	4	7%	
Total	*104*		*48*		*56*		

***Table 2A**: **Self-reported demographics of all respondents**, stratified by DSD and transgender groups. Bolded entries signify combined categories used for statistical analysis, and their original components are indented below them. *Indicated p-values were calculated using the combined categories indicated in bold. **DSD or transgender identifier was assigned to each participant based on their selection of DSD, transgender and/or "other" responses. ***Participants were able to select more than one response for self-identifier, so all individual* *responses summed will be greater than total respondents. For all other demographic categories, only one response was allowed. Degrees of freedom, and Pearson chi-square value are as follows: Ethnicity df=1, Pearson chi-square value=0.026 Education df=3, Pearson chi-square value=4.242, Medical Insurance df=5, Pearson chi-square value=3.635*

Free-response clarifications for "other" selections, selected in combination with DSD or transgender or independently
"Intersex"
"Non binary"
"AMAB, identify & live as female"*
"Trans woman"
"FTM Transsexual"
*"CAIS**"*
"Trans, ftm, trans man, femme ftm, male"
"Transmasculine Genderqueer"
"gender non-conforming"

***Table 2B**: A selection of "other (please specify)" responses that were manually entered as a response to the survey statement "Mark all the following terms that apply to you." *AMAB is an acronym for "assigned male at birth" **CAIS is an acronym for "complete androgen insensitivity syndrome"*

Religious affiliation differed significantly (p<0.001, Table 2A) between DSD and transgender respondents, as displayed in Figure 1. For the purposes of statistical analysis, categories were combined and compared as "Atheist/Agnostic," "Christian/Catholic," and "Other." Transgender individuals were more likely to select "Atheist/Agnostic" (28/55; p=0.05) compared to individuals with DSD, whereas individuals with DSD were far more likely to select "Catholic" or "Christian" (24/47; p=0.002) than transgender participants.

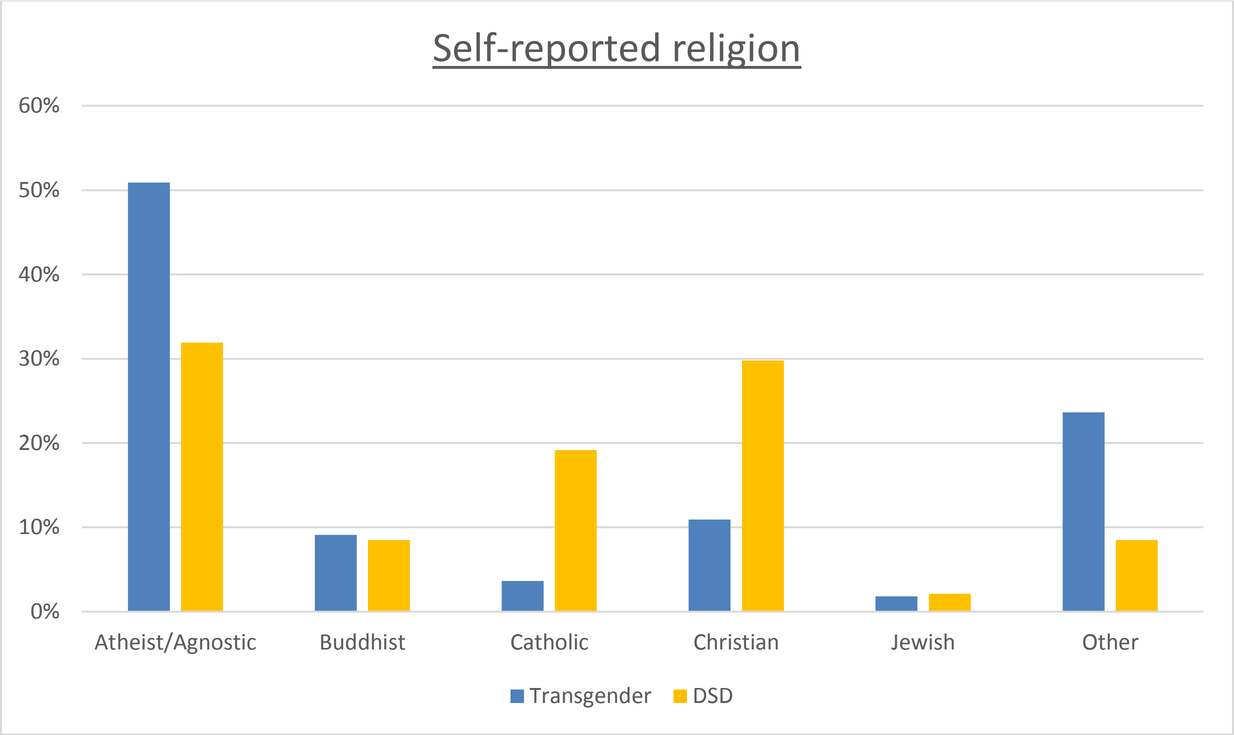

Figure 1: Self-reported religion stratified by transgender or DSD population. *Y-axis represents percentage of participants. See frequencies in Table 2A. N=102 Pearson chi-square value=15.707, df=2, p<0.001.*

While respondents who identify as transgender appeared to report slightly higher education, these differences were not statistically significant (p=0.233, Table 2A). However, transgender respondents generally reported lower incomes (Table 2A) and were significantly more likely to report income less than $25,000 per year (22/55 transgender respondents; p<0.001).

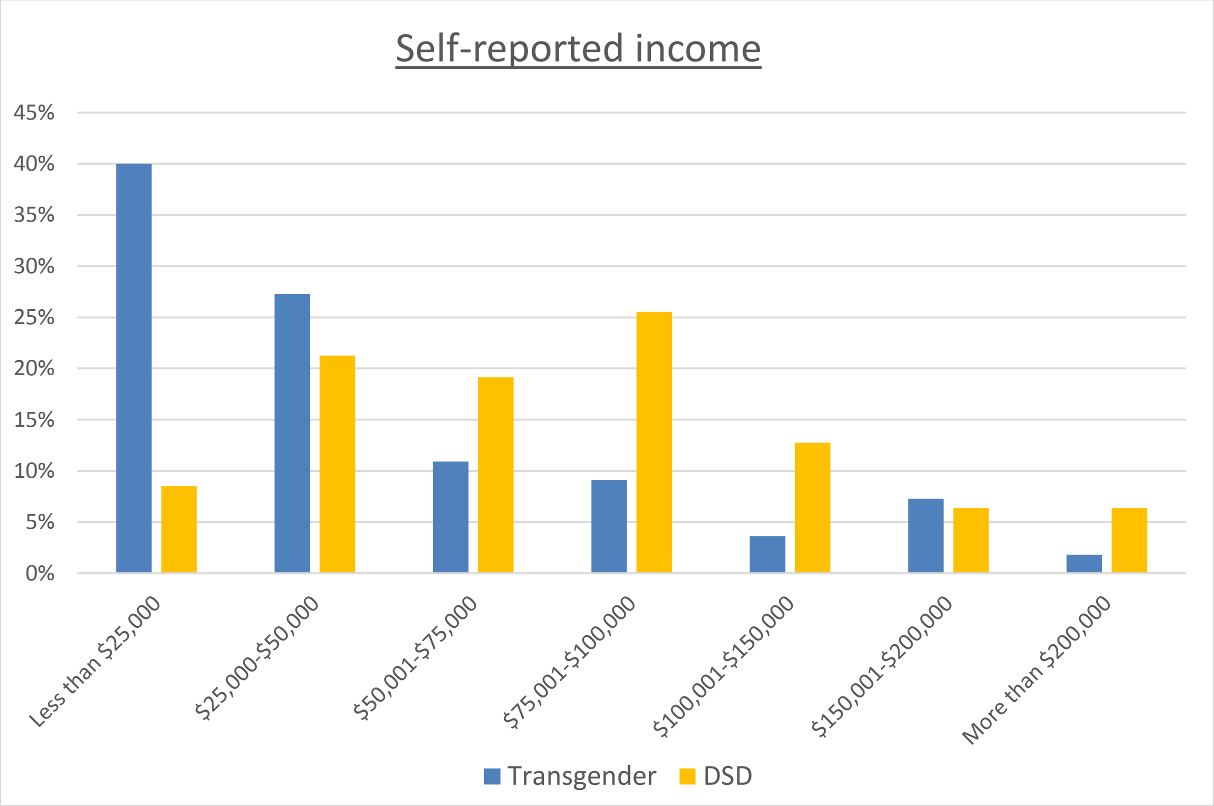

***Figure 2: Self-reported income stratified by DSD or transgender identity**. Y-axis represents percentage of participants. See frequencies and uncombined data in Table 2A. N=102. Pearson chi-square value= 18.976, df=5, p=0.002*

Relationship status differed significantly between transgender participants and those with DSD (Table 2A, p=0.002). However, transgender individuals were more likely to select “domestic partnership, long-term relationship, or other committed romantic partnership” (17/52; p=0.19), while individuals with DSD were more likely to select “married” (21/47; p=0.003). “Other” responses were specified with free-response text in two of the five responses: once as “widowed,” and once as “divorced.” All five “other” responses were excluded in chi-square analysis due to low number of responses, and three out of five respondents did not clarify what “other” signified.

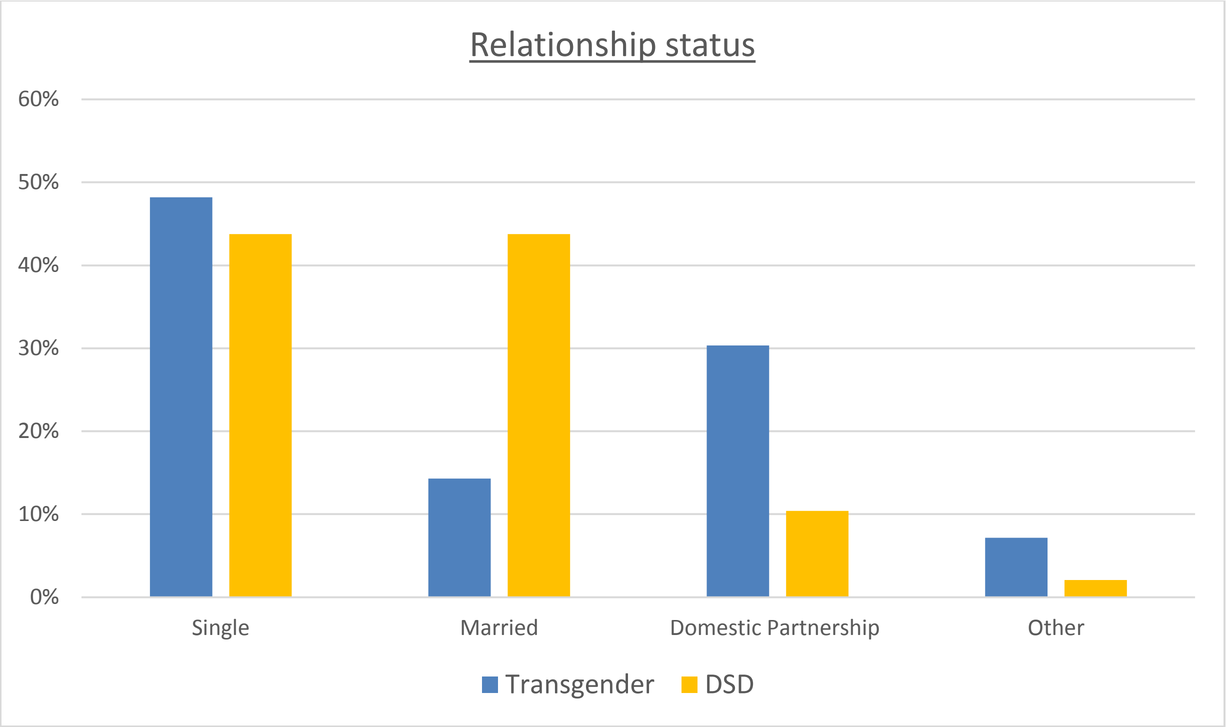

Figure 3: *Relationship status stratified by DSD or transgender identifier. Y-axis represents percentage of participants. See frequencies in Table 2A. N=104. Pearson chi-square value=12.903, df=2, p=0.002*

3.2 Gender Identities

A majority of respondents were assigned female at birth (61%), were raised as female (58%) and currently identify (53%) and "live" as female (64%) (Table 3A). Gender identity is defined as an internal process that is not necessarily reflected as behavior. We used the phrase "gender living as" to describe gender expression, defined by the American Pyschological Association (APA) as the process in which "a person acts to communicate gender."[94] Gender identity was used to assess for gender differences in all analyses unless otherwise indicated. For the purposes of statistical analysis, "Both," "Neither," and "Other" gender identity categories were combined for all categories in which gender was reported, except for "Gender raised as," in which only "Male" and "Female" categories were used; all "other responses" were not included in analysis due to low numbers of responses (total n=6). Some participants chose to clarify "other" selections further, included in Table 3B.

Gender Categories	**Total**		**Transgender**		**DSD Group**		**p-value**
	n	**%**	**n**	**%**	**n**	**%**	
Gender assigned at birth							
Female	**63**	**61%**	**27**	**48%**	**36**	**75%**	0.002
Male	**39**	**38%**	**29**	**52%**	**10**	**21%**	
Both	2	2%	0	0%	2	4%	
Neither	0	0%	0	0%	0	0%	
Other	0	0%	0	0%	0	0%	
Total	*104*		*56*		*48*		
Gender raised as							
Female	**60**	**58%**	**26**	**46%**	**34**	**71%**	0.007
Male	**38**	**37%**	**27**	**48%**	**11**	**23%**	
Other	**6**	**6%**	**3**	**5%**	**3**	**6%**	
Both	1	1%	1	2%	0	0%	
Neither	2	2%	1	2%	1	2%	
Other	3	3%	1	2%	2	4%	
Total	*104*		*56*		*48*		
Current gender identity							
Female	**55**	**53%**	**22**	**39%**	**33**	**69%**	0.01
Male	**23**	**22%**	**15**	**27%**	**8**	**17%**	
Other	**26**	**25%**	**19**	**34%**	**7**	**15%**	
Both	9	9%	5	9%	4	8%	
Neither	5	5%	4	7%	1	2%	
Other	12	12%	10	18%	2	4%	
Total	*104*		*56*		*48*		
Gender living as							
Female	**53**	**64%**	**17**	**49%**	**36**	**75%**	0.012
Male	**15**	**18%**	**8**	**23%**	**9**	**19%**	
Other	**13**	**16%**	**10**	**29%**	**3**	**6%**	
Both	6	7%	3	9%	3	6%	
Neither	4	5%	4	11%	0	0%	
Other	3	4%	3	9%	0	0%	
Total	*83**		*35**		*48*		

***Table 3A**: **Gender category frequencies**. Frequencies for all participants' genders in each category, stratified by transgender and DSD identifiers. Bolded entries signify combined categories used for statistical analysis, and their original components are indented below them. All p-values were calculated using these combines categories. *Fewer transgender respondents were able to reply to this question, since it was added after the survey began circulating. Degrees of freedom and Pearson chi-square value are as follows: Gender assigned at birth* *df=1, Pearson chi-square value=11.998, Gender raised as df=1, Pearson chi-square value= 7.198, Current gender identity df=2, Pearson chi-square value=9.309, Gender living as df=2, Pearson chi-square value=8.820*

All free-response clarifications for "other" gender selections
"Transmasculine"
"I consider myself an "Intersex Female"
"both, neither, genderfluid, pangender, nonbinary"
"Genderqueer"
"Transmasculine genderqueer"
"Non-binary, ilyagender; I also accept maverique and aporagender and aliagender"
genderqueer or gender-nonconforming"
"non-binary"
"Fluidflux (This means that my gender can both change and fluctuate in intensity.)"
"Like I said I'm ployamorous but probably more of a gender fluid type"
"genderqueer/transgender, more male than female"
"both, neither, agender, fluid, masc, femme"

***Table 3B**: All free-response explanations clarifying the selection of "other" gender identities*

Gender assigned at birth and gender identity differed significantly between DSD and transgender groups (Table 2A, p=0.002). More transgender individuals were assigned male at birth (29/56, 52%) compared with individuals with DSD (10/48, 21%), and more individuals with DSD were assigned female at birth (36/48, 75%)(p=0.002). "Gender assigned at birth" and "gender raised as" received very similar response frequencies, with eight (three transgender participants and five participants with DSD) out of the total 104 respondents making variant selections between the responses. More participants with DSD identified as female (33/55, 60%) compared with transgender individuals (22/55, 40%) (p=0.01), while transgender participants were more likely to select "both," "neither," or "other" as their current gender identity (19/56, 34%)(p=0.04). Fewer transgender respondents were able to select a response for current gender expression (35/56, 63%), since this question was added after the survey began circulating (further described in Methods); nevertheless, similar trends were seen. Transgender participants selected “both,” “neither,” or “other” genders more often (10/35, 29%)(p=0.014), while participants with DSD selected “female” more frequently (36/48, 75%)(p=0.025).

3.3 Medical Genetics and Genetic Counseling

Access to Medical Genetics and Genetic Counseling was assessed through two questions, both of which directly asked participants if they had ever had an appointment with a genetic counselor or geneticist. “Don’t know” responses were not included in the final data analysis due to low response numbers (n=4 for having seen a medical geneticist and n=3 for having seen a genetic counselor) and the inappropriateness of combining uncertainty with firm positive or negative answers.

Pearson chi-square analysis showed no significance (Table 4, p=0.063) between the number of DSD individuals who had seen a genetic counselor (8/47, 17%) compared with the number of transgender individuals who had seen a genetic counselor (3/53, 6%). However, DSD individuals were more likely to have seen a medical geneticist than transgender individuals (p=0.02). Fourteen of 47 (30%) DSD individuals had seen a medical geneticist compared to 6 of 53 (11%) transgender individuals.

Provider	Total		Transgender Group		DSD Group		p-value
	n	%	n	%	n	%	
Genetic Counselor							0.063
Yes	11	11%	3	6%	8	17%	
No	86	86%	49	92%	37	79%	
I don’t know	3	3%	1	2%	2	4%	
Total	100		53		47		
Medical Geneticist							0.02
Yes	20	20%	6	11%	14	30%	
No	66	66%	45	85%	31	66%	
I don't know	4	4%	2	4%	2	4%	
Total	100		53		47		
Either Genetic Counselor or Medical Geneticist							0.031
Yes	21	21%	7	13%	14	30%	
No	76	76%	45	13%	31	66%	
Don't know	3	3%	1	13%	2	4%	
Total	100		53		47		

Table 4: Access to Medical Genetics services*. Reported frequencies for all participants’ reported access of Medical Genetics services via appointments with genetic counselors, medical geneticists or either. “I don’t know” answers were excluded in all data analyses.*

3.4 Diagnosis disclosure

Eighty-five participants (82%) reported that they had received a diagnosis related to their gender. Fewer participants who identified as transgender (42/56) reported a diagnosis pertaining to their gender as compared to the participants who identified as DSD (43/47; p=0.028). Figure 4 displays the percentages of groups that selected each diagnosis, ascertained through a follow-up question asking participants to clarify their specific diagnosis. Eighty-four of the eighty-five participants responded with their specific diagnoses, the most common of which include “Gender dysphoria or other medical term for transgender” (39/84) and “androgen insensitivity syndrome” (25/84) (Table 5A). While 39/41 transgender respondents selected “gender dysphoria” as a diagnosis, one respondent selected “Klinefelter syndrome,” and another selected “Turner syndrome.” No respondents with DSD selected “gender dysphoria” as a diagnosis. The remaining reported forms of DSD collectively comprised 24% of the diagnoses and are further detailed in Table 5A. Note that many diagnoses are not mutually exclusive (example: “46,XY Female or 46,XY DSD” and “Androgen Insensitivity Syndrome”); although participants were only able to select one diagnosis. 6 participants selected “other” diagnosis, and all chose to clarify this selection in a free-response text answer (Table 5B). Pearson chi-square analysis was performed using the following three categories: “gender dysphoria,” “androgen insensitivity syndrome,” and “other DSD.”

Diagnosis	**Transgender group**	**DSD group**	**Total**
	n	**n**	**n**
Androgen Insensitivity Syndrome (AIS)	**0**	**25**	**25**
Other DSD	**2**	**18**	**20**
5-alpha reductase deficiency	0	1	1
46,XY Female or 46,XY DSD	0	2	2
46,XX Male or 46,XX DSD	0	1	1
Clitoromegaly	0	2	2
Gonadal Dysgenesis	0	1	1
Hypospadias	0	2	2
Klinefelter syndrome	1	2	3
Turner syndrome	1	1	2
Other	0	6	6
"Gender Dysphoria" or other medical term for transgender	**39**	**0**	**39**
Total	*41*	*43*	*84*

***Table 5A**: **All reported diagnoses**, stratified by transgender and DSD groups. Bolded entries signify combined categories used for statistical analysis, and their original components are indented below them. Pearson chi-square=72.936, df=2, p<0.001*

Free-response clarifications for “other” diagnosis selections
"46, XX/XY mosaic chimerism"
"Mosaic Turner Syndrome"
"Bilateral ovotestes, 46xx"
"XY/XO"
"17beta Hydroxysteroid deficiency syndrome"
"46XX/46XY chimerism/mosaicism, ovotestes, hypospasias[sic]"

Table 5B: *All free-response explanations clarifying the selection of “other” diagnoses*

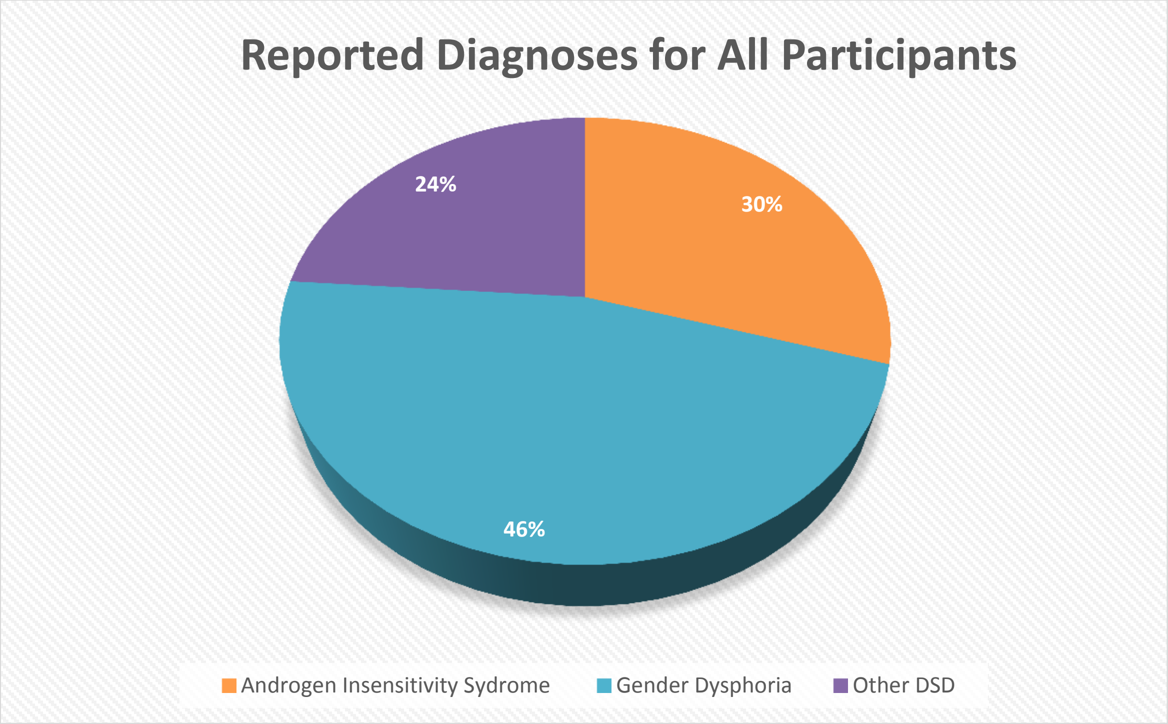

Figure 4**: **Categorized diagnoses for all participants. *A pie chart reflecting the frequencies of self-reported diagnosis of the 84 respondents who reported a diagnosis categorized by into three groups: Androgen Insensitivity Syndrome, Gender Dysphoria and Other DSD. Of note, two of the “other DSD” diagnoses were reported by transgender individuals. All frequencies are listed in Table 5A.*

A majority of participants who identified as transgender and reported receiving a diagnosis indicated that they themselves were the first to be informed of their diagnosis (37/41, 90%), while diagnoses for individuals with DSD were most often received by the parents of those individuals (28/43, 65%. $p<0.001$). Individuals with DSD were more likely to be diagnosed before five years of age ($p<0.001$) than were individuals who identify as transgender ($p<0.001$, Table 6). There was no significant difference between DSD and transgender groups with respect to when participants personally learned of their diagnoses ($p=0.292$).

Disclosing Diagnosis	**Total**		**Transgender Group**		**DSD Group**		p-value
	n	%	n	%	n	%	
Age at which participant was diagnosed							
Younger than 5	**20**	**24%**	**1**	**2%**	**19**	**44%**	<0.001*
5-15 years	**9**	**11%**	**2**	**5%**	**7**	**16%**	
5-10 years	5	**6%**	1	**2%**	4	**9%**	
11-15 years	4	**5%**	1	**2%**	3	**7%**	
16-19 years	**18**	**21%**	**7**	**17%**	**11**	**26%**	
Older than 20 years	**37**	**44%**	**6**	**15%**	**6**	**14%**	
Total	*84*		*41*		*43*		
Who was told about your diagnosis first?							
Me	**49**	**58%**	**37**	**90%**	**12**	**28%**	<0.001
Someone else	**35**	**42%**	**4**	**10%**	**31**	**72%**	
My parents or guardian	30	36%	2	5%	28	65%	
Other	5	6%	2	5%	3	7%	
Total	*84*		*41*		*43*		
Age at which participant learned of diagnosis							
Age 15 or younger	**25**	**30%**	**10**	**24%**	**15**	**35%**	0.292*
Younger than 5	3	**4%**	1	**2%**	2	**5%**	
5-10 years	7	**8%**	2	**5%**	5	**12%**	
11-15 years	15	**18%**	7	**17%**	8	**19%**	
16-19 years	**15**	**18%**	**9**	**22%**	**6**	**14%**	
Older than 20 years	**44**	**52%**	**22**	**54%**	**22**	**51%**	
Total	*84*		*41*		*43*		

***Table 6**: **Ages for diagnosis and disclosure, stratified by transgender and DSD identifiers**. Ages at which participants received a diagnosis relating to their sex or gender, and ages when participants actually learned of this diagnosis. Bolded entries signify combined categories used for statistical analysis, and their original components are indented below them. All p-values were calculated using these combined categories. Pearson chi-square value and degrees of freedom are as follows: Age diagnosis was made, Pearson chi-square value=35.564, df=3, Who told you about your diagnosis first, Pearson chi-square value=31.039, df=1, Age at which diagnosis was learned, Pearson chi-square value=2.464, df=2.*

DSD and transgender groups also differed with respect to who gave them their diagnoses (Table 7, p=0.003); many transgender individuals (20/41, 49%) reported “figuring out” their diagnoses themselves. When asked who gave them their diagnosis, the two most common responses from individuals with DSD were "a non-genetics medical specialist" (10/43, 23%), or "I figured it out myself" (8/43, 19%) (Figure 5). In contrast, the most common responses from transgender individuals were "I figured it out myself" (20/41, 51%) and "my primary care provider" (8/41, 20%). These responses were compared using Fisher exact test due to low response numbers for several categories such as "Medical geneticist." "My parent(s) or guardian(s)" and "Another family member" were combined, while "Other" and "I don't know/don't remember" were excluded from analysis.

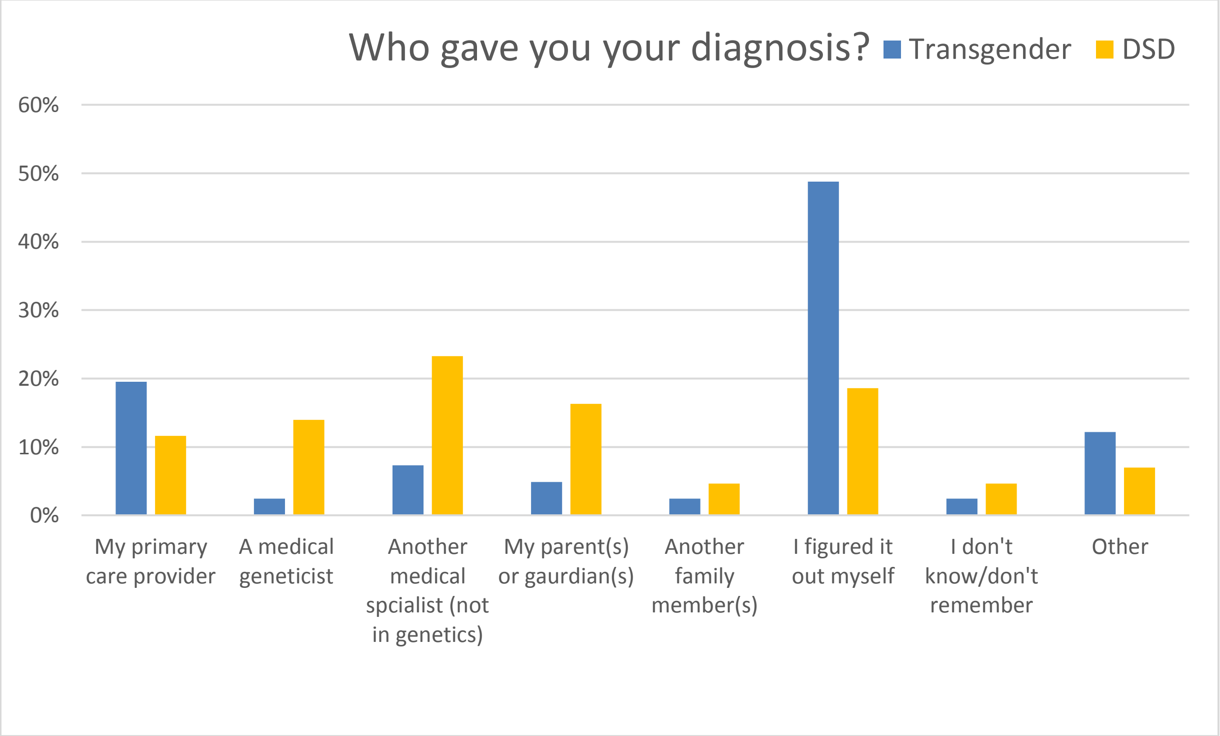

***Figure 5**: Bar graph for who disclosed diagnosis, stratified by DSD and transgender identities. See frequencies below in Table 7.*

Who gave you your diagnosis?	**Total**		**Transgender Group**		**DSD Group**		p-value
	n	%	n	%	n	%	
My primary care provider	**13**	**15%**	**8**	**20%**	**5**	**12%**	0.003
A medical geneticist	**7**	**8%**	**1**	**2%**	**6**	**14%**	
Another medical specialist (not in genetics)	**13**	**15%**	**3**	**7%**	**10**	**23%**	
A family member	**12**	**14%**	**3**	**7%**	**9**	**21%**	
My parent or guardian	9	11%	2	5%	7	16%	
Another family member	3	4%	1	2%	2	5%	
I figured it out myself	**28**	**33%**	**20**	**49%**	**8**	**19%**	
I don't know/don't remember	3	4%	1	2%	2	5%	
Other	8	10%	5	12%	3	7%	
Total	*84*		*41*		*43*		

Table 7: Who disclosed diagnosis, stratified by DSD and transgender identifiers. *Fisher exact test was performed using the five categories indicated above in bold, including "family member" which combined the two variables indented below. "Other" and "I don't know/don't remember were excluded from analysis.*

Twenty-six respondents (31%) reported that someone was with them at the time they were informed of their diagnosis, most frequently a parent (54%). How the diagnosis was revealed with respect to the individuals present is detailed in Table 8. Six of the 26 respondents reported that they were asked for their privacy preferences before diagnosis disclosure.

Disclosure methodology	Total		Transgender Group		DSD Group	
	n	%	n	%	n	%
Who accompanied to receive diagnosis						
Parent	14	54%	1	17%	13	65%
Other family member	2	8%	0	0%	2	10%
Romantic Partner	5	19%	4	67%	1	5%
Friend	1	4%	1	17%	0	0%
Other	4	15%	0	0%	4	20%
Total	26		6		20	
How diagnosis was disclosed						
To me privately	10	38%	2	33%	8	40%
To me and to the person/people accompanying me, at the same time	10	38%	3	50%	7	35%
To me privately first, and then to the person/people accompanying me	1	4%	0	0%	1	5%
To the person/people accompanying me first, privately, and then to me	5	19%	1	17%	4	20%
Total	26		6		20	

***Table 8**: **Methodology of diagnosis disclosure when participant was accompanied.** Total response frequencies, and stratified by DSD and transgender groups.*

Of the individuals who were accompanied when they were told about their diagnosis, 20/26 were asked about their privacy preferences first (17/20 individuals with DSD and 3/6 transgender individuals)(Table 8).

3.5 Healthcare satisfaction

The PSQ-18 is a previously-validated survey with 18 individual questions designed to contribute to 7 “subscales” or themes including general satisfaction, technical quality, interpersonal manner, communication, financial aspects, time spent with doctor, and accessibility and convenience. The PSQ-18 consists of Likert-scale questions, with 1=Strongly Agree through 5=Strongly Disagree. Answers from each of the 18 questions are used to calculate a mean score for that individual question or “item,” and then these items are categorized together within their respective subscales. The means are then averaged together to create a subscale mean, which can be used to draw conclusions about the different assessed areas of healthcare satisfaction.

When responses from the PSQ-18 were compared based on gender, age, ethnicity, income and transgender v DSD self-identifier, only one significant difference was observed. “I think my doctor’s office has everything needed to provide complete care” received significantly lower agreement from respondents who identify as Transgender (mean=2.32) compared with the DSD group (mean=2.80) (p=0.046). Scoring from the PSQ-18 allows for the creation of subscales in order to more concisely assess for healthcare satisfaction. No clear differences were seen between groups with respect to subscales; total means as well as means of participants who identify as transgender or DSD are displayed in Table 9 (see all analyzed responses in Appendix F).

PSQ-18 Subscale	**Total Subscale Mean Score**	**DSD Group Subscale Mean Score**	**Transgender Group Subscale Mean score**
General Satisfaction	2.73	2.75	2.72
Communication	2.45	2.48	2.42
Technical Quality	2.38	2.43	2.33
Interpersonal Manner	1.84	1.91	1.78
Financial Aspects	2.51	2.35	2.65
Time Spent with Doctor	2.41	2.38	2.44
Accessibility and Convenience	2.50	2.42	2.58

***Table 9**: **Healthcare satisfaction scores for all participants.** The means of subscale for all participants (total mean) and for DSD and Transgender groups. Higher numbers indicated higher satisfaction; the highest possible score is 5. See all individual item mean values and p-values in Appendix F. With one exception detailed in text above, no p-values were significant.*

3.6 Perceived Stigma measurements

Perceived stigma was assessed through two lines of questioning in the survey. Participants were asked, “Have you ever felt that a health professional (doctor, nurse, social worker, etc.) was unable or unwilling to treat you due to your diagnosis or due to your gender?” and participants were asked to describe potential denial of care through a series of paired questions regarding hormonal therapy requisition and possible denial (see survey in Appendix A, questions 25, 27-28).

When asked if they felt that a healthcare practitioner had been unwilling or unable to treat them due to their diagnosis or due to their gender, forty participants (40%) indicated that they had. Fifty-two participants (52%) did not report that this had ever happened to them, while eight (8%) reported uncertainty (Figure 6A, 6B). There were no statistically significant differences in response between DSD and transgender participants, nor between genders, education levels, or income levels.

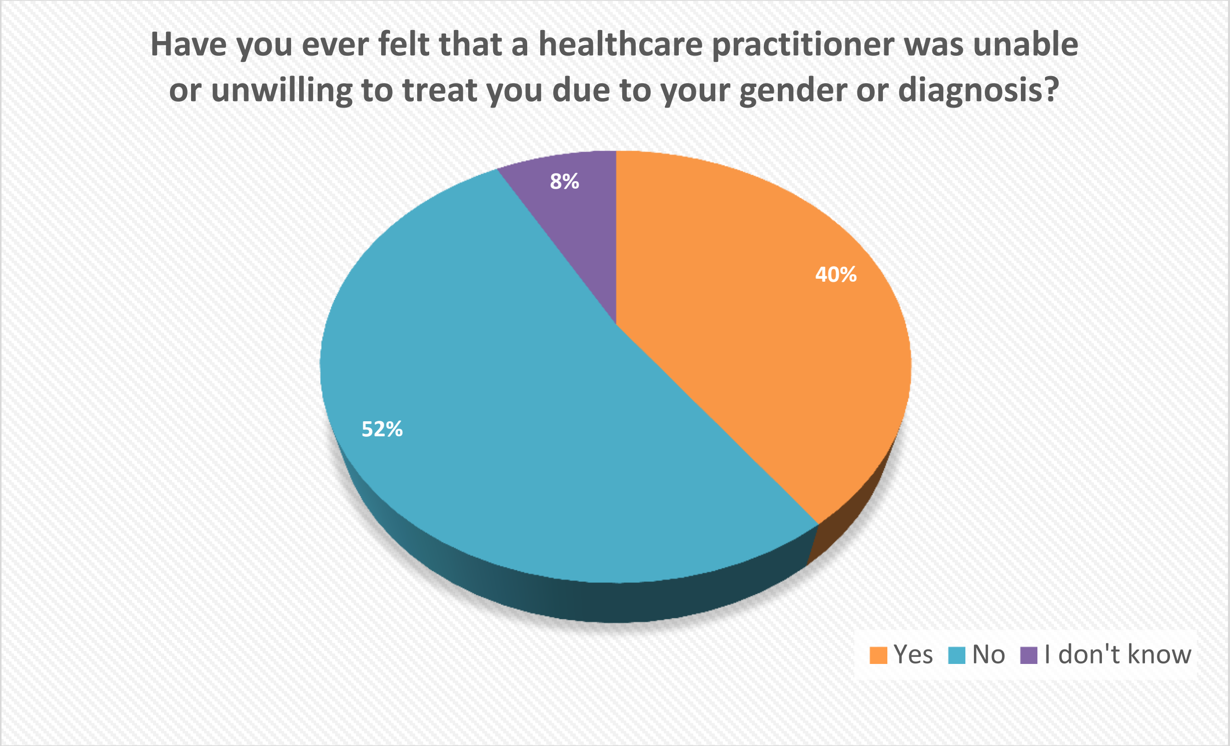

***Figure 6A**: Total responses to "Have you ever felt that a healthcare practitioner was unable or unwilling to treat you due to your gender or diagnosis?"* *N=100.*

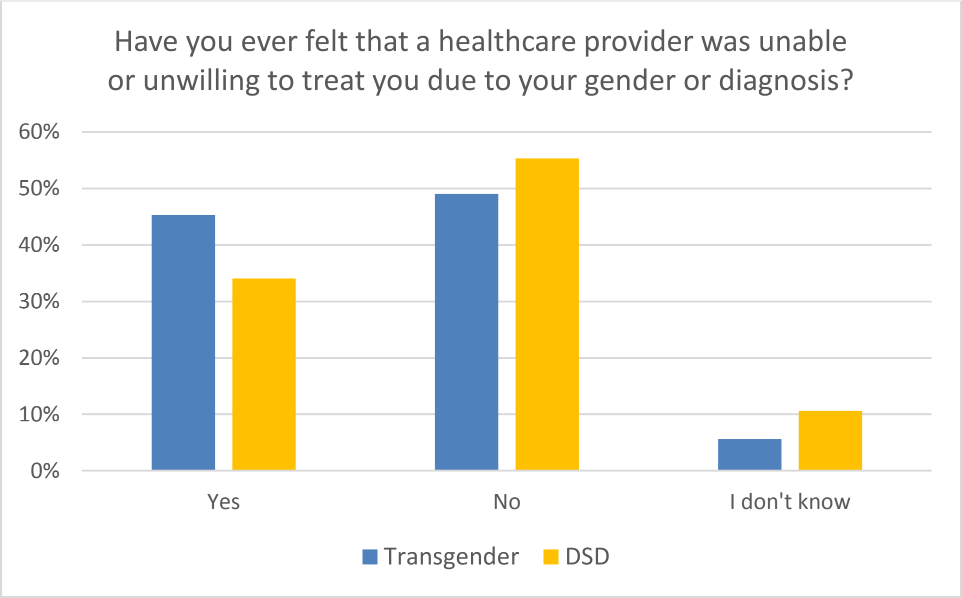

Figure 6B: *Responses to "Have you ever felt that a healthcare practitioner was unable or unwilling to treat you due to your gender or diagnosis?" separated by DSD and transgender groups. Pearson chi-square value=1.746, df=2, p=0.418.*

Of the 102 participants who responded to the question, "Have you ever requested hormonal therapy related to a disorder of sex development or to being transgender?" 77 (75%) responded "Yes." Of these, 76/77 participants responded to a follow-up question inquiring if this request was denied. Eleven of the 76 (14%) reported having their request for hormonal therapy denied. Respondents cited numerous reasons for this denial, with the majority (6/11, 55%) choosing to select the "other" option and to write in specific information concerning this experience (Table 10).

Free-response clarifications to "other" responses describing rationale for denial of hormonal therapy
“Doctor didn't know how to go about it or where to even start with finding out.”
“Doctor did not want to give me testosterone because I had been castrated as a child without my consent and raised as a girl.”
“Strict and arbitrary requirements before they'd be willing to prescribe”
“The UCSB student health doctors did not feel comfortable continuing my prescriptions”
“My doctor did not agree morally with prescribing hormones for me, doctor did not agree with my requested HRT, doctor insisted I see a therapist and must pick a binary gender before prescribing hormones”
“The doctor didn't agree with me trying testosterone”
“Primary doctor uncomfortable with dosage of testosterone that keeps me in a normal, healthy male range. Referring me to an endocrinologist. Scheduling pending months now, due to reluctance of local endocrinologists, because of DSD.”

Table 10: *All free response answers clarifying “other” responses when survey participants were asked to explain why hormonal therapy was denied to them.*

3.7 Free-Response Answers

Three free-response questions were included at the end of the survey in order to collect personal stories from survey participants, to collect further details concerning denials of medical care, and to solicit suggestions relating to diagnosis disclosure and overall healthcare for individuals with differences of sex or gender, such as DSD or transgender status. Twenty-three participants (22.1%) answered all three questions, and 33 (32%) answered at least one. No trends were seen among participants who chose to respond with respect to gender assigned at birth, current gender identity, DSD or transgender identifiers, income, or education. Selected responses are shown in Table 11A-C, and all answers are tabulated in Appendix G. General themes that emerged from the free-response questions included a desire for honesty and respect for gender identity, greater education and sensitivity training for healthcare providers, and increased use of preferred names and preferred pronouns (such as “he/him,” “she/her” or “they/their”) on all healthcare paperwork.

If you have ever felt that a health professional (doctor, nurse, social worker, etc.) was unable or unwilling to treat you due to your diagnosis because of your medical condition or gender identity, please describe that occurrence(s):
"When I was still assigned female in spite of a rather male external genital appearance, I had a hard time getting care that involved anything that had to do with me not conforming with my assigned sex/gender. I never had the surgery that doctors recommended so I had no surgical SRS to make me conform with female. I wanted to get surgery for my hypospadias and until I found a surgeon who also performed surgery on FTM transgender people, I was never even considered for the surgery."
"'Lack of medical necessity' has been used throughout my transition to deny me service, such as facial feminization surgery, electrolysis, top surgery, and progesterone."
"A Veterans Administration Primary Caregiver refused outright to take me a patient."
"I went to the ER with a problem. As soon as they found out I was intersex they stopped everything and sent me home with no medical treatment of any kind. I almost died."
"At an LGBT health clinic a doctor claimed I was lying about being intersex and said my body was "impossible" then encouraged me to let them examine my genitals."
"I had one dr attempt to coercively perform a genital exam that had nothing to do with what i was there for because they were curious about my surgical results"
"A nurse misgendered me immediately after a gender affirmation surgery; when I corrected her, she said she'd already called me that and I hadn't corrected her then, and that she hadn't had time to check my chart, and she then complained audibly in the hall to another nurse about how "rude" I was."

Table 11A*: *Personal experiences with denial of care due to sex or gender differences, *selected responses to an optional, free-response question at the end of the survey.*

Do you have any ideas or suggestions to improve healthcare for people who have sex or gender differences?
"Have more education on these subjects and not just be a chapter they skim over in class."
"My current doctor is great, everyone should practice like her. She takes time to listen, answer questions, she has talked with me about any concerns about sex or hormones and treats you like you are the only patients she needs to worry about for that time."
"Be our advocate as well as our Doctor."
"Ask gender and pronouns immediately. Change forms to allow spots for identified gender. Allow to use a preferred name at the pharmacy."
"Be honest, listen to us and take our concerns seriously."
"Health care providers need more education about DSD/intersex, transgender, transsexual. They need to learn about "thinking outside the box" in order to treat those of us with genetic and other differences."
"A friend who was an ultrasound instructor used to say, "Treat the patient not the picture." Consider individual needs for every patient."
"No infant cosmetic surgery unless the condition is absolutely life threatening. Doctors listening to patients first especially if that patient is an adult."
"Have safe zone training and education on Trans issues. Don't make access to care harder for non-binary people. Believe patients when they reveal information about themselves"

***Table 11B**: **Suggestions to improve healthcare for individuals with sex or gender differences**, selected responses to an optional, free-response question at the end of the survey.*

Do you have any ideas or suggestions to improve the process of revealing diagnoses that involve a patient’s gender?
"Be honest and open, as a teenager I would have preferred to hear this first without anyone present. I am certain a teenager has the ability to make important decisions regarding their health. The teenager should aslo be asked if it is appropiate to talk to parents, care givers. This is done when it comes to birth control and reproductive issues and I believe a diagnosis of DSD certainly is in that group."
"the person who is getting the diagnosis should definitely be involved at the initial outset. Yes, with more direct discussion to the patient most of all."
"The important thing is that the affected individual be given the chance to make informed choices with the support of social workers or other supportive person who is not a member of the family. The family should be made aware that there are many happy people with DSD's and that genital surgery is only appropriate when the affected individual has made a fully informed decision."
"The doctor should not in any way participate in subterfuge. My endocrinologist knew that my parents were lying to me and kept the information she gave me at a minimum to collude."
"Discuss with person diagnosed first. If the patient is a minor child, this should take into consideration ethical legal consideration prior to treatment."
"Doctors should use appropriate language that's not stigmatizing but they should be honest"
"Reveal in a way that is honest yet appropriate. Include family and provide family and patient counseling."
"Ask the patient first and make sure they understand what's going on and that they have access to counseling and other support"
"Should always talk to teen first! It can be a safety issue. And because being ordered can be a safety issue, it should never be disclosed without taking with the patient."

Table 11C: Suggestions to improve diagnosis disclosure for individuals with sex or gender differences, *selected responses to an optional, free-response question at the end of the survey.*

IV. DISCUSSION

4.1 Reviewing rationale for research and initial hypotheses

People with DSD and people who identify as transgender have stories, challenges and perspectives that are unique to these populations. Individuals in both groups can be perceived as "gender non-conforming," although their non-conformity would be attributed to different etiologies. While most people, healthcare practitioners included, would recognize DSDs as having an innate biological basis, transgender identities are believed to have a more psychological basis. Thus, transgender individuals could be perceived by others as having choice in their gender non-conformity. However, most, if not all, transgender individuals would emphatically disagree with the notion of choice in their gender identity.

While DSD and "intersex" are often used interchangeably, there are individuals with DSD who identify as solely male or female and do not identify as intersex at all.[10] Intersex has recently begun to be closely associated with transgender identities as well as with the gay and lesbian community. While the acronym "LGBT" is still very prominent, the more inclusive "LGBTQIA" is increasing in popularity and includes queer/questioning (Q), intersex (I) and asexual (A) designations.[95] It is worth emphasizing again that not all individuals with DSD consider themselves "intersex" and that intersex is considered to be more of an identity, while DSD is a diagnosis or a description of variance of anatomical, gonadal or chromosomal sex.[96] Thus, while DSD has traditionally been used to describe medical conditions, the term has recently become closely associated with transgender identities and incorporated into the overarching gay community.

This research was born out of a desire to explore the opinions and experiences of transgender individuals and individuals with DSD about their healthcare. More specifically, the goal of the research was to determine if differences emerged between the groups, especially in access to Medical Genetics and Genetic Counseling, diagnosis disclosure, healthcare satisfaction, and perceived stigma. There were five specific hypotheses driving this research:

1. Individuals with DSD will report higher healthcare satisfaction than individuals who are transgender.
2. Male individuals with DSD and MtF transgender individuals will report lower satisfaction with healthcare than female individuals with DSD and FtM transgender individuals.
3. Respondents with DSD will report that they have been given a medical diagnosis relating to their sex or gender more often than transgender groups.
4. Transgender individuals will report greater difficulty accessing Medical Genetics services, possibly attributed to lack of indication for referral.
5. Transgender individuals will report greater stigma.

We assessed for differences within our participant population using many other categories beyond DSD and transgender groups, such as different genders (e.g., male and female) and income brackets. However, few measureable differences were observed. We were limited by a small sample size, which may have obscured differences between groups. For all areas in which no significant difference was measured, it is very possible that differences could have been revealed if more individuals had participated in the survey.

4.2 Differences between genders

We initially hypothesized that male individuals with DSD and transgender individuals who were assigned male at birth would report greater stigma and lower healthcare satisfaction than female individuals with DSD and transgender individuals who were assigned female at birth. Interestingly, no measurable differences were seen between any of the responses based on either gender assigned at birth or current gender identity. Genders are often compared in research oriented towards experiences of transgender individuals, and previous studies have shown that FtM transgender individuals experience greater violence and stigma than MtF individuals.[22] To the authors' knowledge, only one study has compared male and female gender in individuals with DSD. In that study male-identified individuals reported higher quality of life (QoL) and better psychological status than female-identified individuals.[28] While it is likely that the different cultural aspects of Sao Paolo, Brazil (where the study was performed) influenced these results, the measured difference between genders should not be ignored and was influential in our decision to measure differences between genders in this study.

There are several possible explanations for the lack of measurable differences between genders in our study. First, a larger percentage of our population (61%) was assigned female at birth, so it is possible that there were small differences between the two groups but the population that was assigned male at birth was too small to measure that difference. Secondly, due to our small sample size, transgender and DSD groups had to be combined for gender comparisons. This limited our ability to compare male-identified transgender individuals to male-identified individuals with DSD, for example, which may have confounded some differences.

4.3 Access to Medical Genetics and Genetic Counseling

We had originally hypothesized that transgender individuals would report accessing Medical Genetics services less frequently than individuals with DSD due to the lack of a medical indication for referral. Individuals with DSD did report accessing a genetic counselor more frequently than transgender individuals (17% compared with 6%), but this difference was not measured as statistically significant (p=0.063). However, due to the mathematical limitations arising from our small cohort, it is likely that if the cohort had been larger, significance would have been achieved. For the individuals that had been seen in Medical Genetics and/or Genetic Counseling, we also did not ask for their referral indication. Thus, it is possible that an individual with DSD was seen in Genetic Counseling due to a family history of cancer, for example, as opposed to their DSD.

Even though individuals with DSD were more likely to have seen a genetic counselor, fewer than 20% had done so. Substantially more individuals with DSD reported having seen a medical geneticist (31%), but still a majority had not. Importantly, the survey did not specify that the interaction with Genetics had to have been related to their diagnosis. Since Medical Genetics and Genetic Counseling are indicated in many, if not all, cases of DSD[97], it is surprising that fewer than 20% of individuals with DSD reported having had Genetic Counseling. It is possible that survey respondents did not remember past appointments in Medical Genetics and reported inaccurately. Additionally, many participants with DSD were born in the 1950’s and 1960’s when Medical Genetics services were non-existent and they may not have been offered a referral once they were older and the field was established. It is also possible that other medical specialties that manage care of DSD, such as Endocrinology and Urology, do not offer referrals to Medical Genetics, either due to lack of availability of genetics services, lack of awareness of genetics services or to a belief that such a referral is not indicated. An additional possibility is that individuals with DSD are not receiving referrals to genetics because they are often infertile, leading to the misperception that there is no risk of recurrence and thus no need for Genetic

Counseling. Of course, beyond discussing recurrence risk, there are many other benefits of a Medical Genetics consultation, such as education, health management, and risk assessment for other family members. This concerning lack of referrals for individuals with DSD could be mitigated through further communication and educational efforts to spread awareness of the potential benefits of Medical Genetics and Genetic Counseling services.

4.4 Reported diagnoses and diagnosis disclosure

Most respondents reported that they had been given a diagnosis pertaining to their sex or gender, although individuals with DSD more often reported receiving a diagnosis. In theory, nearly every person responding to this survey would report a diagnosis; DSD is an umbrella diagnosis itself, and transgender individuals would most often have received a diagnosis of gender dysphoria. However, it is possible to identify as transgender and not receive a gender dysphoria diagnosis, either due to nondisclosure to one's doctor or due to the phrasing of requirements in the DSM to receive a diagnosis, namely, the requirement of distress. Due to the possibility of identifying as transgender without an actual diagnosis pertaining to sex or gender, we had originally hypothesized that participants with DSD would report a diagnosis more often, which was confirmed.

Interestingly, two individuals who initially identified themselves as transgender reported diagnoses other than gender dysphoria, namely, Turner syndrome and Klinefelter syndrome. It is probably not a coincidence that these two syndromes are sometimes not included in the umbrella term "DSD" —indeed, the DSD incidence of 1/4,500 quoted by the Chicago Consensus Statement implicitly excludes them. The incidence of Klinefelter syndrome is 1/500, and the incidence of Turner syndrome is 1/2,500, both of which independently outnumber the quoted

1/4500 incidence.[98,99] It is possible that the two transgender participants with these diagnoses have never been informed of the broader DSD category, or, alternatively, that they themselves choose not to identify as having a DSD.

Within our study population, the most common DSD diagnosis by a large margin was Androgen Insensitivity Syndrome (AIS). While AIS is one of the most common DSDs, it was represented far more often that would be expected; this is likely explained by the DSD group that circulated the survey, namely, AIS-DSD. This group is welcoming to all individuals with DSDs but emphasizes support for those with AIS—hence the group name. It is possible that our study outcomes would have changed if all DSDs has been proportionally represented, especially considering that AIS does not typically present with significant associated health concerns (such as salt wasting in CAH). In other words, it is possible that the experiences of individuals with AIS are not representative of the larger DSD community and that the abundance of participants with AIS influenced our results.

While individuals with DSD were *diagnosed* at significantly younger ages than transgender individuals, the two groups did not differ in the ages at which they *learned of* their diagnoses. Fifty-four percent of transgender participants and 51% of individuals with DSD learned of their diagnoses after age 20. Numerous survey respondents with DSD mentioned in free-response answers that while they were diagnosed as children, their diagnoses were kept secret from them by their families and physicians. This type of secrecy was, in fact, standard of care in the 1950's and 1960's, and 50% of our participants with DSD were born in 1965 or earlier.[30,32] It is very likely that when these individuals were born, their parents were cautioned to keep their diagnoses from them, if their parents were even informed themselves. It is possible that this cohort effect is responsible for numerous other differences between DSD and transgender groups in this study.

A majority of diagnoses were disclosed either with other people present at the appointment or to other people without the individual him/herself present Half of the transgender patients and the vast majority of patients with DSD were not asked about their privacy preferences before disclosure of diagnosis. In the case of those with DSD, this can be attributed, at least in part, to parents initially learning of the diagnosis of their infant children, but other scenarios can be imagined. This lack of privacy consideration is concerning, given the implications of sex and gender differences and the possible dangers in disclosing such differences in the presence of others. There was an open-ended question at the end of the survey soliciting advice to improve the process of disclosing a diagnosis to a patient when that diagnosis involved the patient's sex or gender. Notably, there were answers endorsing the disclosure of diagnosis to the entire family, as well as responses that suggested asking privacy preference before such a disclosure. Other themes that emerged included provision of additional resources (especially counseling and social work) at the time of disclosure and honest discussions with the patient and family. Importantly, a genetic counselor is ideally suited to act as such a support person at the time of diagnosis disclosure because they are trained in both the medical aspects and the psychosocial aspects of genetic disorders.

4.5 Healthcare satisfaction

It is of particular importance to medical providers to understand the healthcare experiences of these two population subgroups so that steps can be taken to better meet their needs. The previously-validated PSQ-18 is a useful tool to assess for healthcare satisfaction, in large part due to the generation of "subscores" or numerical representations for satisfaction in the following areas: general satisfaction, technical quality, interpersonal manner, communication, financial aspects, time spent with doctor, and accessibility and convenience. Among the data generated by our participants' responses, the subscore means did not differ significantly between DSD and transgender groups, and thus all participants were considered together. When compared to other populations that have been surveyed using the PSQ-18, the total subscore means collected here were universally lower. In other words, healthcare satisfaction as measured by the PSQ-18 was lower for transgender individuals as well as for individuals with DSD when compared to other groups using this same measurement. For example, patients with chronic disease (such as epilepsy and melanoma)[100,101] and typical female outpatient populations[102] all reported higher healthcare satisfaction than our cohort.

While we had originally hypothesized that individuals with DSD would report higher healthcare satisfaction than transgender individuals, there were no substantial differences between the DSD and transgender groups. While this is possibly a result of low statistical power, the close similarity of the scores makes this less likely. It is possible that the lack of difference between the two groups may have been influenced by the DSD support group that helped to distribute the survey, AIS-DSD. Based on personal communications and the expressed opinions of other professionals specializing in DSD, this group is known for having more radical views and distrust towards the medical community, so individuals affiliated with AIS-DSD may have lower healthcare satisfaction. In other words, it is possible that the views of this group may not constitute an accurate representation of healthcare satisfaction for all patients with DSD. It would likely be best for future studies in this subject to recruit several additional support groups with potentially different views concerning healthcare to assess if reported satisfaction increases, decreases or remains the same.

At the end of the survey, there was an open-ended question inviting participants to share suggestions for overall healthcare improvement for individuals with sex or gender differences (such as transgender or DSD status). Suggestions varied, although several themes emerged. Honesty and greater education for physicians were suggested often, similarly to diagnosis disclosure responses. In addition, many participants suggested that physicians should be sensitive and keep an open mind towards gender and sex variance. One individual wrote, “Drop the binary, if you have Intersex/DSD you are living proof that sex is not binary it is on a spectrum.” Many individuals requested that healthcare offices allow patients to write in preferred names and preferred pronouns (such as he/his, she/hers, and they/theirs) as well as gender identity in order to affirm and respect gender non-conforming individuals.

4.6 Stigma and denial of healthcare

Since stigma itself is difficult to measure, denial of healthcare was assessed as a measurable effect of presumed stigma. We had originally hypothesized that transgender participants would more frequently report denial of care, although no measurable differences were seen between the transgender and DSD groups. In total, 40% of all survey respondents reported that they believe a healthcare practitioner had been unable or unwilling to treat them due to their diagnosis or their gender. In an open-ended question inviting participants to share their stories, denial of care was attributed most often to ignorance or perceived physician discomfort with DSDs or transgender patients. Respondents also chose to report what they felt was improper care, as opposed to denial of care, such as physicians performing or asking to perform what respondents considered unnecessary or extended genital exams. Of course, a genital exam can be a required component to a physician evaluation, and without further detail it is not possible to know if these exams were truly unwarranted or extended; however, regardless of its technical validity, these complaints represents a miscommunication between patient and provider.

In the "National Transgender Discrimination Survey Report on Health and Healthcare," of the over 7,000 participants, 19% reported having been refused care "due to their transgender or gender non-conforming status."[49] In the same survey, half of all respondents reported that they had to educate their providers in how to provide transgender care. We phrased our question so that it asked if a healthcare practitioner had ever been "unwilling or unable to treat you due to your gender or due to your diagnosis," which would likely encompass refusal of care as well as the need to educate providers. With that acknowledgement, 40% of participants responded "yes," which appears to be similar to the responses in the National Transgender Discrimination Survey, although the comparison is not perfect.

4.7 Secondary findings

Over 90% of participants, both transgender and with DSD, reported that they had completed at least some college education. However, while both groups had very similar levels of education, transgender participants reported significantly lower incomes, with 40% reporting a household income of less than $25,000 per year. This is likely explained in part by the younger ages of the transgender respondents, but previous research has shown that transgender people are disproportionately more likely to live in poverty despite higher overall education levels.[103]

Previous studies have shown reduced healthcare satisfaction among non-Caucasian gender non-conforming individuals, especially among Hispanic and African American individuals.[49] While there are no significant differences across any of our examined variables when Caucasian and non-Caucasian groups are compared, this may be due to the small sample size of non-Caucasian respondents (n=18). It is possible and even probable that non-Caucasian individuals experience higher levels of stigma and reduced healthcare satisfaction that were not able to be detected here.

Nearly half of the individuals with DSD identified as Christian or Catholic, while a majority of transgender individuals selected Atheist or Agnostic. This observed trend of transgender respondents reporting less religious affiliation may be due to the rejection of transgender identities among some sects of conservative Christianity. Moreover, this could represent a form of social support available to individuals with DSD that is not available to transgender individuals. Relationship status, which can also be perceived as a form of social support, differed between transgender and DSD groups as well. Similar percentages of each group reported being single, but transgender individuals were more likely to report being in a "domestic partnership or other long-term, committed relationship," while individuals with DSD more frequently reported being "married." While this could also be due to the younger ages of transgender respondents, the trend could possibly be influenced by the reduced religious affiliation of transgender respondents that could perhaps dissuade them from marriage.

4.8 Limitations of this study

As discussed previously, a substantial limitation in this study was the low statistical power resulting from small overall sample size. While a variety of small support groups for transgender individuals participated in the survey, only one support group for individuals with DSD chose to participate. This was a large, national group (AIS-DSD), whose participation was critical for the success of this study, but it would have been preferable to have had other groups participate as well to provide a larger sample size and perhaps a more balanced perspective. Unfortunately, all other contacted DSD support groups declined to participate.

AIS-DSD reviewed the SIS and the survey and provided feedback after the survey had already begun circulating among transgender groups (15 responses had been collected). Their feedback was minor but appropriate and led to the overall improvement of the survey. Additionally, AIS-DSD's participation in the survey was contingent on the implementation of this feedback. Their changes were incorporated, but the initial 15 participants saw a slightly different survey that was missing one question compared to the final form. Since these modifications were considered to be minor and only one question was added, the initial 15 responses were included in the final data analysis. Due to the nature of the question, it is not believed that the absence of this question would have influenced answers to any of the others.

The modality of survey distribution (electronically via SurveyMonkey) may have selected against some participants who did not have email or internet access. Additionally, since all participants were recruited through support groups, it is possible the collected opinions were biased. It may be that individuals in support groups would report different experiences than individuals who are not in support groups. In other words, the collected responses and experiences in this study may not be representative of larger DSD and transgender communities. Lastly, since the survey was only available in English, individuals who speak other languages would not have been able to participate.

4.9 Future studies

This study could benefit from repetition with a significantly larger sample size to allow further statistical power and greater insight and suggestions through the free-response questions.

There has been little research published about Genetic Counseling for DSD, and, to our knowledge, there is no published research on Genetic Counseling for transgender patients. A recent LGBT curriculum pilot study produced by Genetic Counseling graduate students at Sarah Lawrence College[86] shows great promise, and it would be interesting to see a similar curriculum pilot study specifically for gender non-conforming patients.

Additionally, research is needed to assess the efficacy and benefits of Genetic Counseling in the care and management of individuals with DSD. These studies could then provide evidence of the advantages to patients of being referred for Medical Genetics services.

Since this study suggests that broad educational initiatives are indicated for healthcare practitioners, it may be helpful to measure healthcare satisfaction of gender non-conforming individuals before and after education is implemented. This could assess for any remaining areas of dissatisfaction and could guide future educational initiatives.

4.10 Conclusions

This study provides insight into the experiences of individuals with DSD and transgender individuals with respect to access to Medical Genetics and Genetic Counseling services, preferences regarding diagnosis disclosure, overall healthcare satisfaction, and perceived stigma. The study consisted of responses from 104 eligible participants, including 56 transgender individuals and 48 individuals with DSD. The original intent was to explore differences between these groups, to identify areas of dissatisfaction, and to solicit suggestions for improvement.

Surprisingly, there were no substantial differences between the DSD and transgender groups with respect to healthcare satisfaction. However, one of the most noteworthy conclusions from this study was the frequency of denial of care and the repeated suggestion to educate healthcare providers on how to treat gender non-conforming patients. Nearly half of all survey respondents reported that a healthcare practitioner had been unable or unwilling to treat them due to their diagnosis or their gender, which was attributed to ignorance or perceived physician discomfort with DSDs or transgender patients. Since all areas of healthcare satisfaction measured lower through the PSQ-18 compared with populations with chronic disease, broad educational efforts are indicated. This could help ease healthcare practitioner discomfort both with DSD and transgender identities and could improve knowledge of any beneficial treatments.

Several themes emerged with respect to the open-ended questions at the end of the survey. Participants suggested that physicians should be better educated and more honest and encouraged them to be sensitive and open-minded toward gender and sex variance.

It is particularly concerning to see that only 20% of individuals with DSD reported having seen a genetic counselor. Since Genetic Counseling is indicated in most, if not all, cases of DSD, it is clear that future efforts should be made to educate healthcare providers about the potential benefits of medical genetics assessment and genetic counseling for affected individuals and their families.

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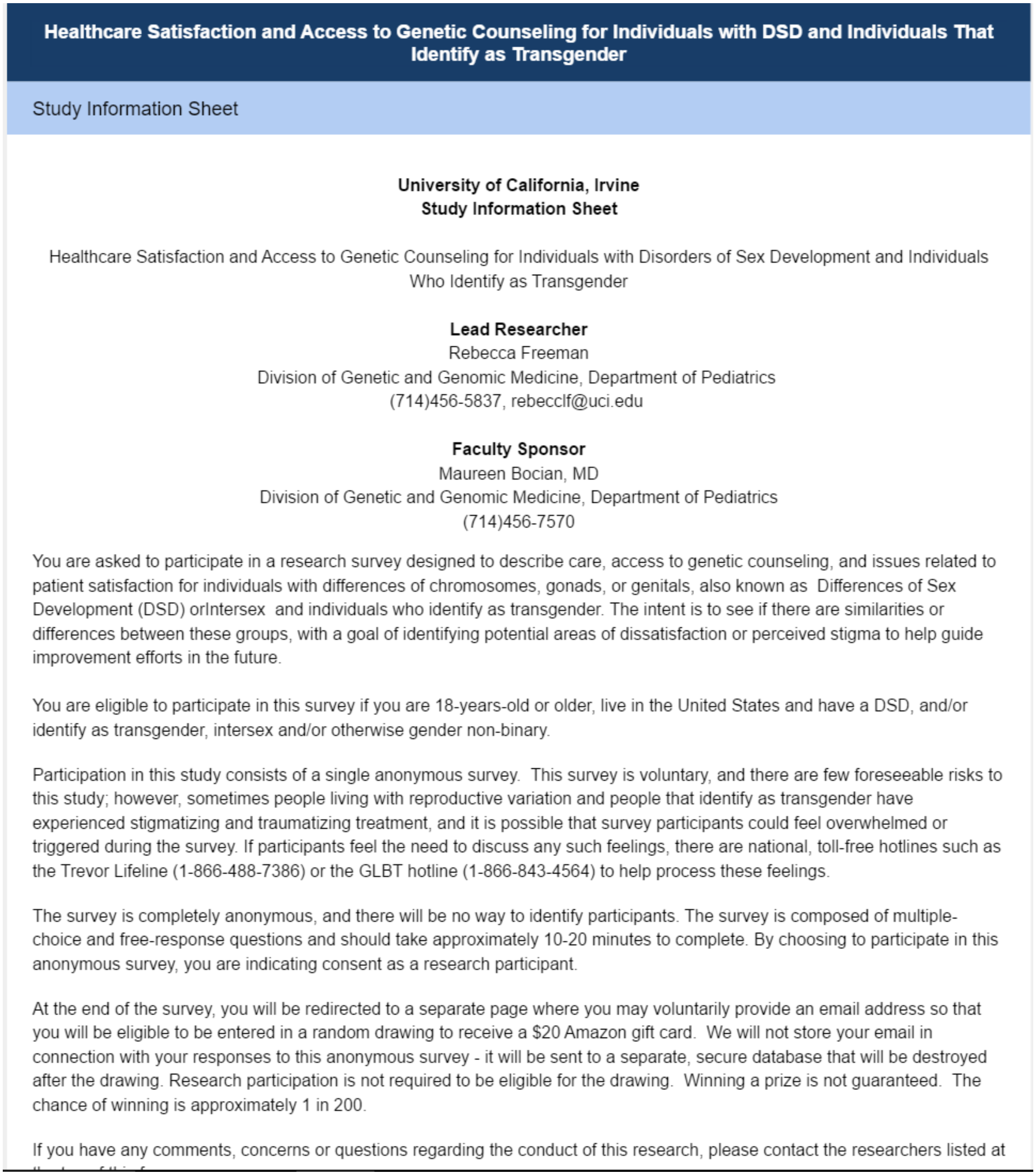
APPENDIX A: Survey



Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

Study Information Sheet

University of California, Irvine
Study Information Sheet

Healthcare Satisfaction and Access to Genetic Counseling for Individuals with Disorders of Sex Development and Individuals Who Identify as Transgender

Lead Researcher
Rebecca Freeman
Division of Genetic and Genomic Medicine, Department of Pediatrics
(714)456-5837, rebecclf@uci.edu

Faculty Sponsor
Maureen Bocian, MD
Division of Genetic and Genomic Medicine, Department of Pediatrics
(714)456-7570

You are asked to participate in a research survey designed to describe care, access to genetic counseling, and issues related to patient satisfaction for individuals with differences of chromosomes, gonads, or genitals, also known as Differences of Sex Development (DSD) orIntersex and individuals who identify as transgender. The intent is to see if there are similarities or differences between these groups, with a goal of identifying potential areas of dissatisfaction or perceived stigma to help guide improvement efforts in the future.

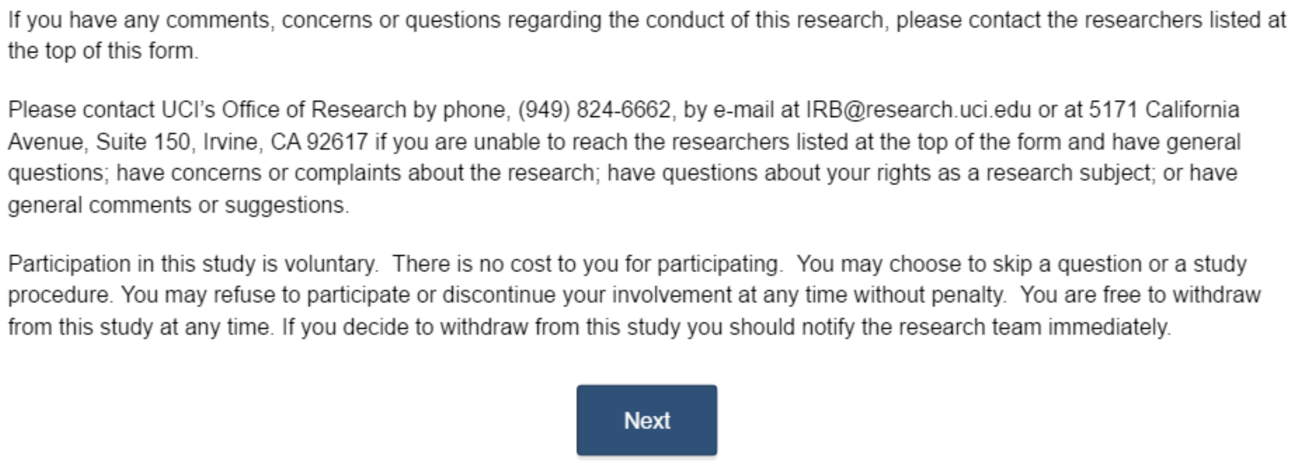
You are eligible to participate in this survey if you are 18-years-old or older, live in the United States and have a DSD, and/or identify as transgender, intersex and/or otherwise gender non-binary.

Participation in this study consists of a single anonymous survey. This survey is voluntary, and there are few foreseeable risks to this study; however, sometimes people living with reproductive variation and people that identify as transgender have experienced stigmatizing and traumatizing treatment, and it is possible that survey participants could feel overwhelmed or triggered during the survey. If participants feel the need to discuss any such feelings, there are national, toll-free hotlines such as the Trevor Lifeline (1-866-488-7386) or the GLBT hotline (1-866-843-4564) to help process these feelings.

The survey is completely anonymous, and there will be no way to identify participants. The survey is composed of multiple-choice and free-response questions and should take approximately 10-20 minutes to complete. By choosing to participate in this anonymous survey, you are indicating consent as a research participant.

At the end of the survey, you will be redirected to a separate page where you may voluntarily provide an email address so that you will be eligible to be entered in a random drawing to receive a $20 Amazon gift card. We will not store your email in connection with your responses to this anonymous survey - it will be sent to a separate, secure database that will be destroyed after the drawing. Research participation is not required to be eligible for the drawing. Winning a prize is not guaranteed. The chance of winning is approximately 1 in 200.

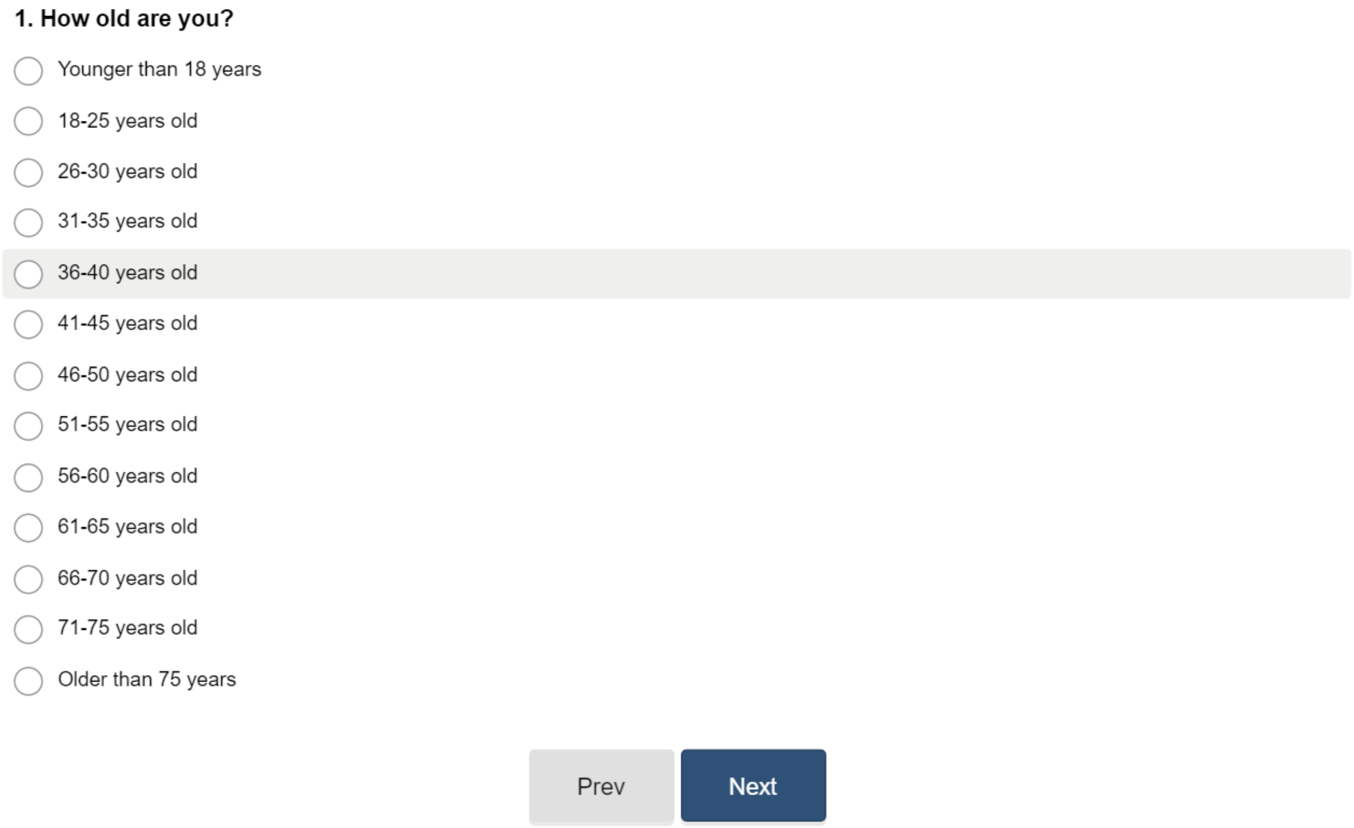
If you have any comments, concerns or questions regarding the conduct of this research, please contact the researchers listed at



If you have any comments, concerns or questions regarding the conduct of this research, please contact the researchers listed at the top of this form.

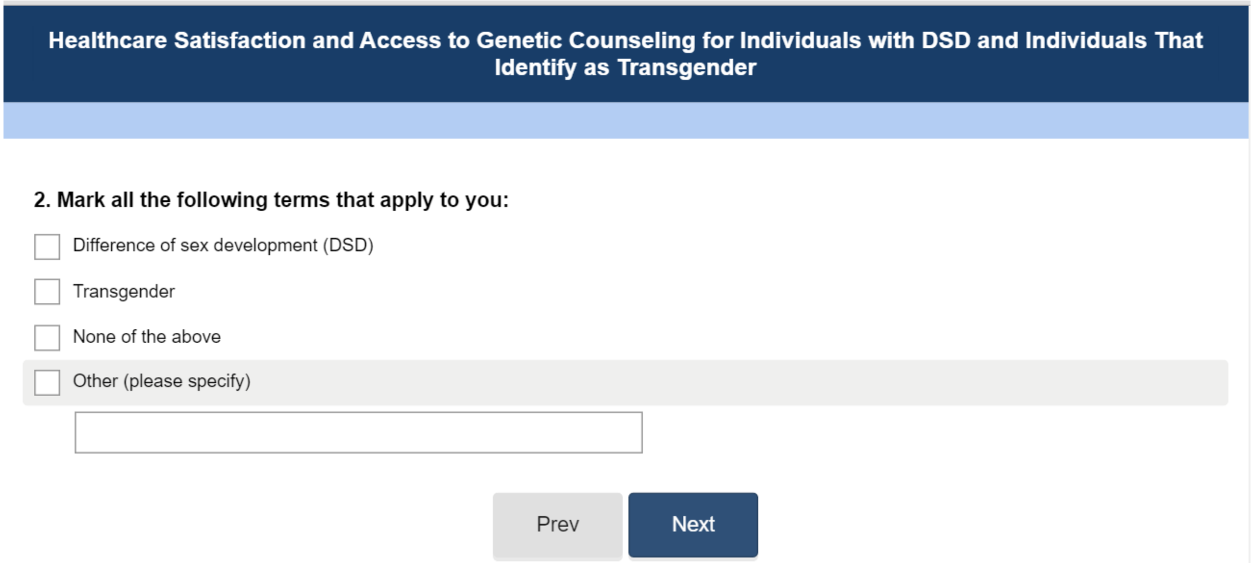
Please contact UCI's Office of Research by phone, (949) 824-6662, by e-mail at IRB@research.uci.edu or at 5171 California Avenue, Suite 150, Irvine, CA 92617 if you are unable to reach the researchers listed at the top of the form and have general questions; have concerns or complaints about the research; have questions about your rights as a research subject; or have general comments or suggestions.

Participation in this study is voluntary. There is no cost to you for participating. You may choose to skip a question or a study procedure. You may refuse to participate or discontinue your involvement at any time without penalty. You are free to withdraw from this study at any time. If you decide to withdraw from this study you should notify the research team immediately.



1. How old are you?

○ Younger than 18 years
○ 18-25 years old
○ 26-30 years old
○ 31-35 years old
○ 36-40 years old
○ 41-45 years old
○ 46-50 years old
○ 51-55 years old
○ 56-60 years old
○ 61-65 years old
○ 66-70 years old
○ 71-75 years old
○ Older than 75 years



Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

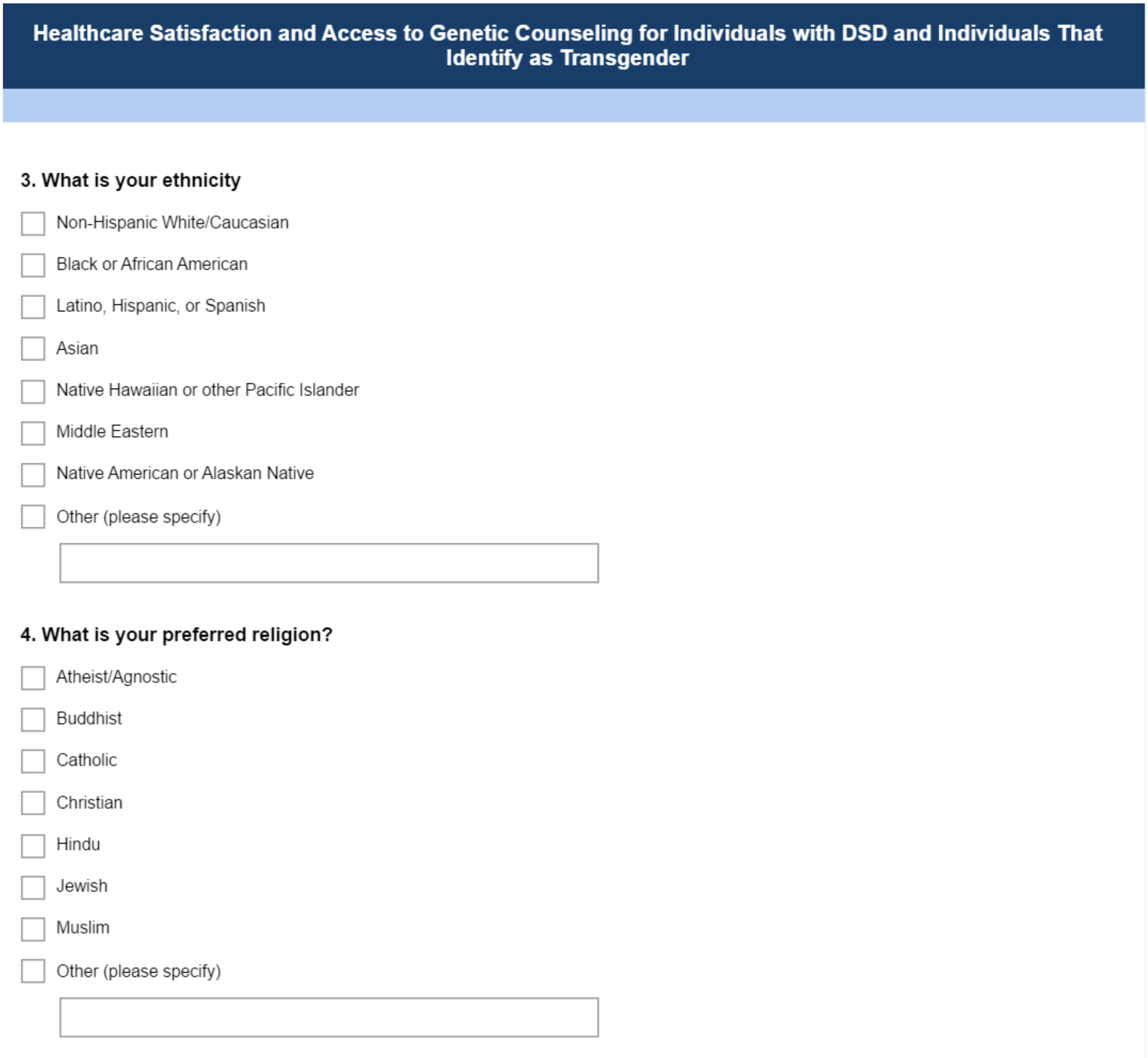
2. Mark all the following terms that apply to you:

☐ Difference of sex development (DSD)

☐ Transgender

☐ None of the above

☐ Other (please specify)



Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

3. What is your ethnicity

☐ Non-Hispanic White/Caucasian

☐ Black or African American

☐ Latino, Hispanic, or Spanish

☐ Asian

☐ Native Hawaiian or other Pacific Islander

☐ Middle Eastern

☐ Native American or Alaskan Native

☐ Other (please specify)

4. What is your preferred religion?

☐ Atheist/Agnostic

☐ Buddhist

☐ Catholic

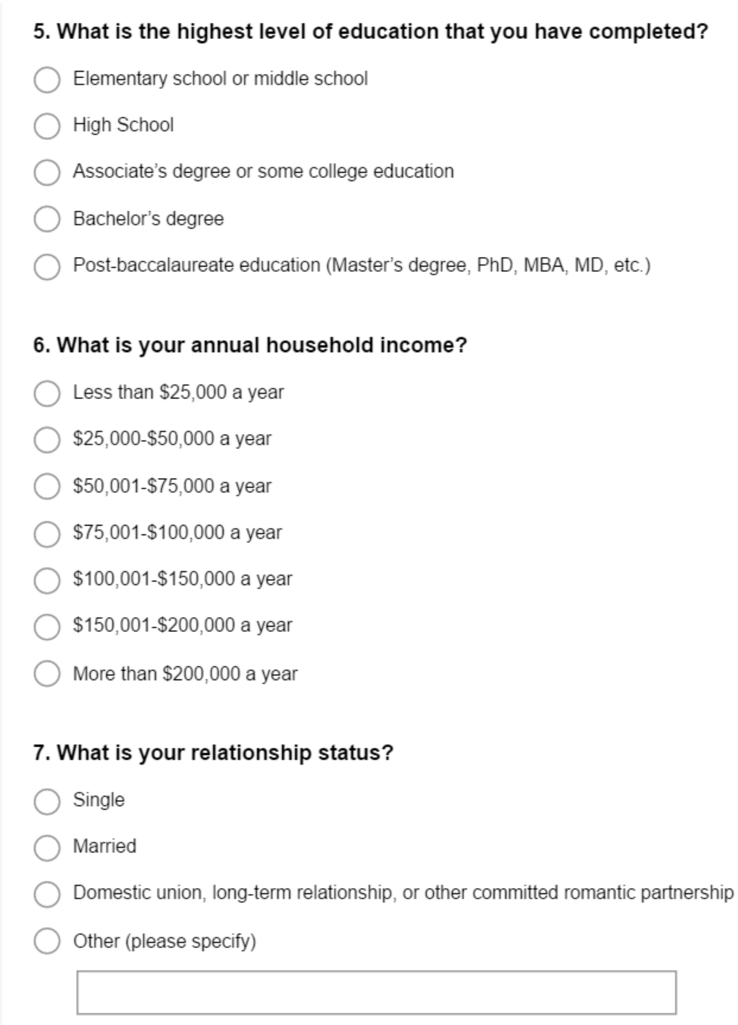
☐ Christian

☐ Hindu

☐ Jewish

☐ Muslim

☐ Other (please specify)



5. What is the highest level of education that you have completed?

○ Elementary school or middle school

○ High School

○ Associate’s degree or some college education

○ Bachelor’s degree

○ Post-baccalaureate education (Master’s degree, PhD, MBA, MD, etc.)

6. What is your annual household income?

○ Less than $25,000 a year

○ $25,000-$50,000 a year

○ $50,001-$75,000 a year

○ $75,001-$100,000 a year

○ $100,001-$150,000 a year

○ $150,001-$200,000 a year

○ More than $200,000 a year

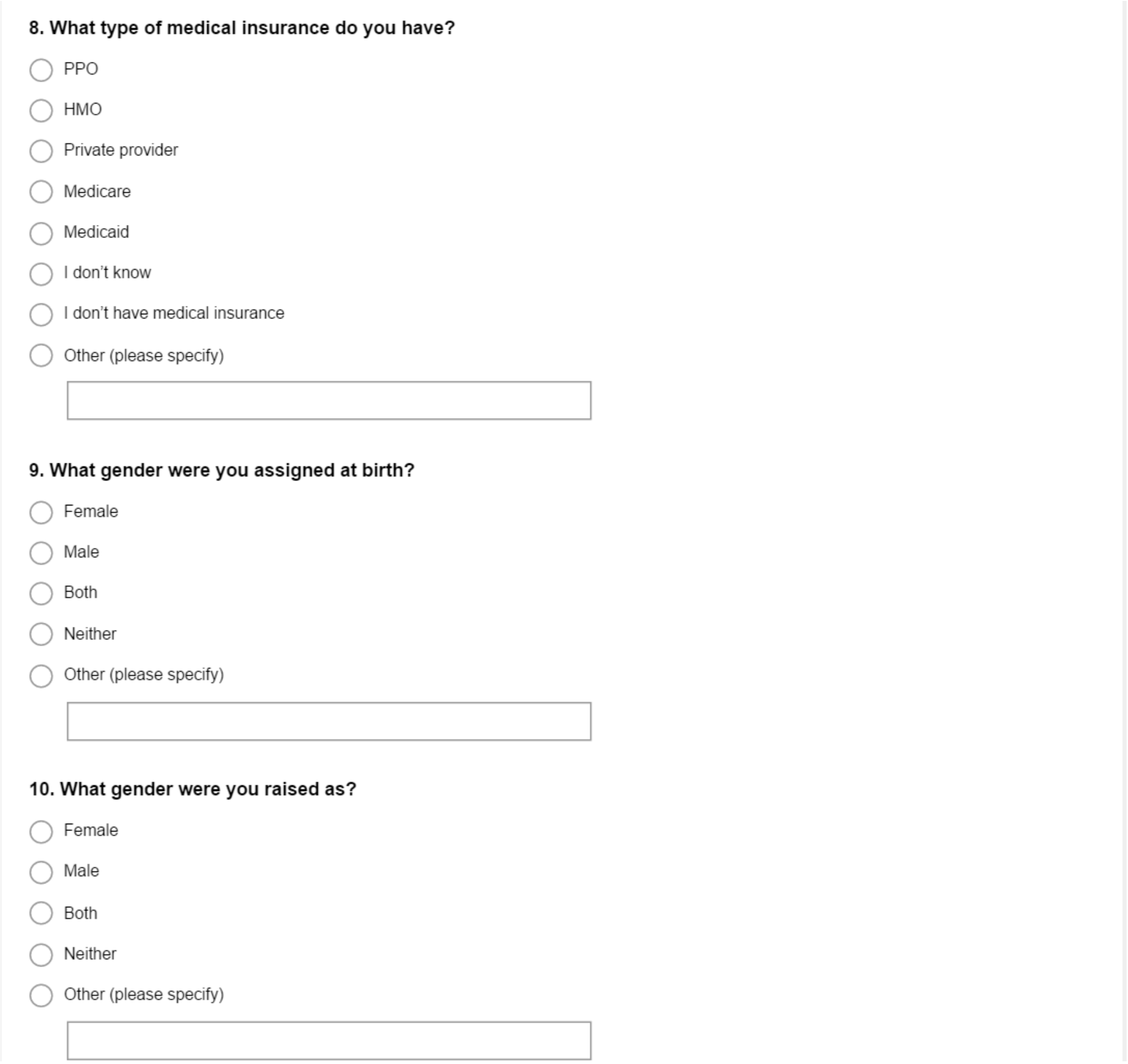
7. What is your relationship status?

○ Single

○ Married

○ Domestic union, long-term relationship, or other committed romantic partnership

○ Other (please specify)



8. What type of medical insurance do you have?

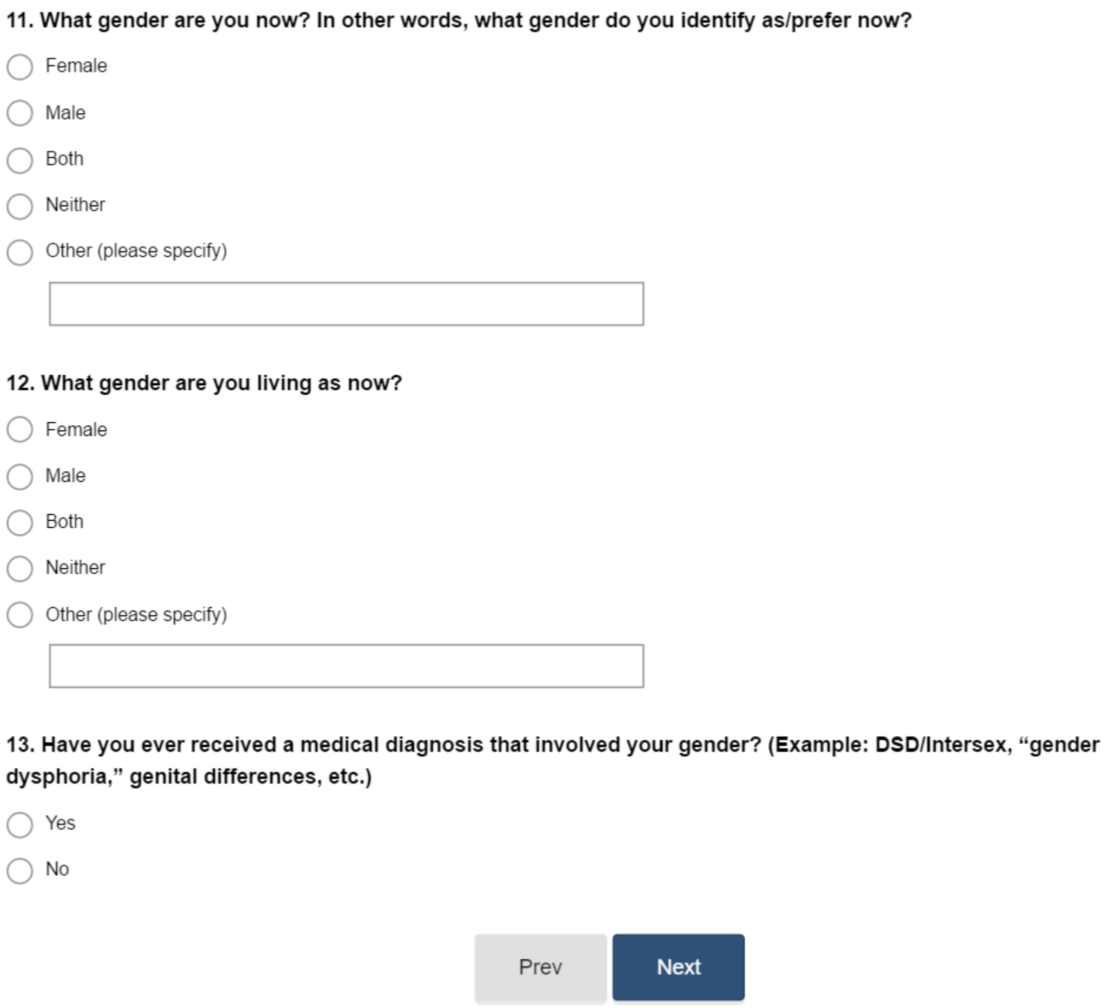
○ PPO
○ HMO
○ Private provider
○ Medicare
○ Medicaid
○ I don't know
○ I don't have medical insurance
○ Other (please specify)

9. What gender were you assigned at birth?

○ Female
○ Male
○ Both
○ Neither
○ Other (please specify)

10. What gender were you raised as?

○ Female
○ Male
○ Both
○ Neither
○ Other (please specify)



11. What gender are you now? In other words, what gender do you identify as/prefer now?

○ Female

○ Male

○ Both

○ Neither

○ Other (please specify)

12. What gender are you living as now?

○ Female

○ Male

○ Both

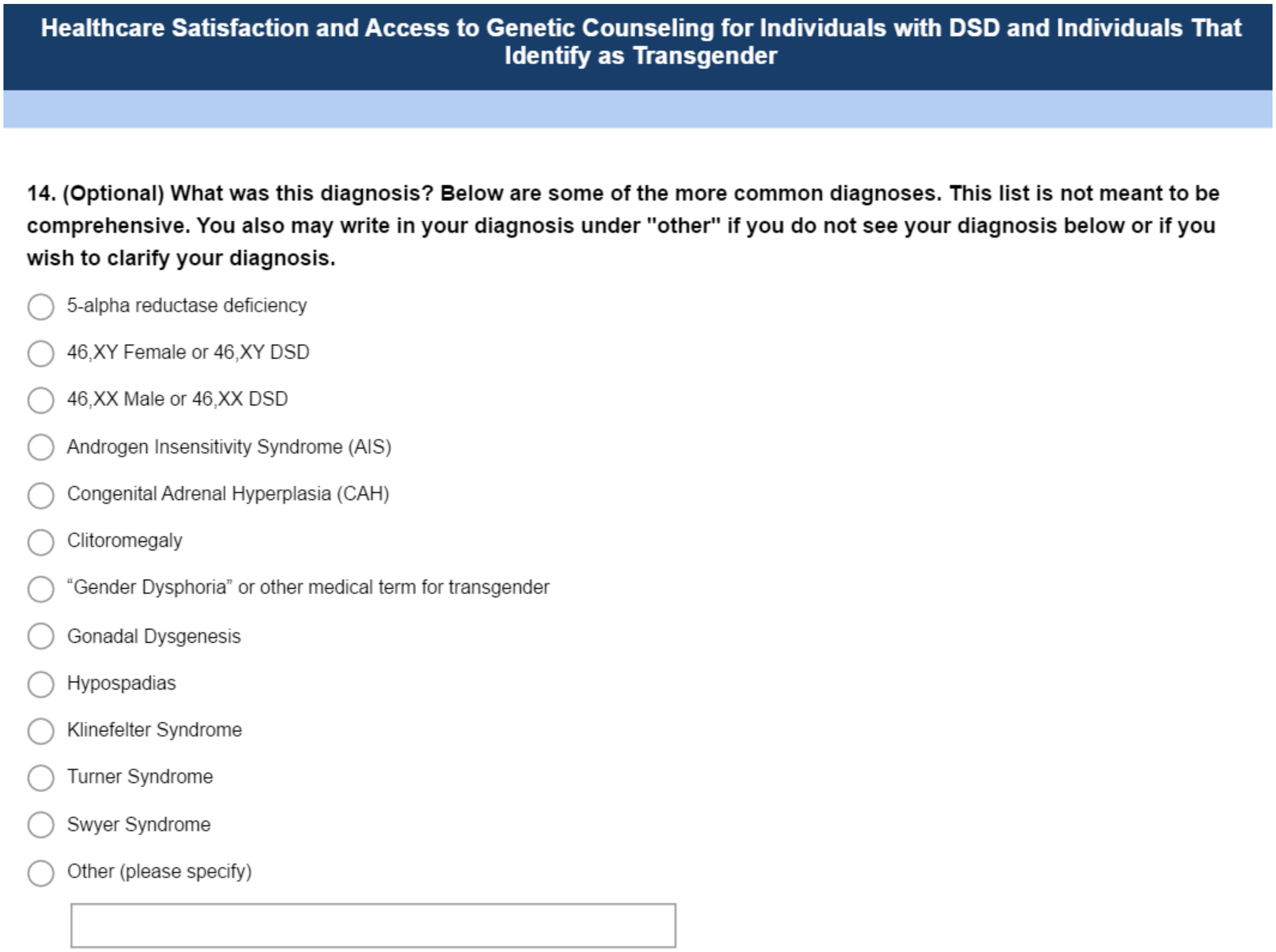
○ Neither

○ Other (please specify)

13. Have you ever received a medical diagnosis that involved your gender? (Example: DSD/Intersex, "gender dysphoria," genital differences, etc.)

○ Yes

○ No



Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

14. (Optional) What was this diagnosis? Below are some of the more common diagnoses. This list is not meant to be comprehensive. You also may write in your diagnosis under "other" if you do not see your diagnosis below or if you wish to clarify your diagnosis.

○ 5-alpha reductase deficiency

○ 46,XY Female or 46,XY DSD

○ 46,XX Male or 46,XX DSD

○ Androgen Insensitivity Syndrome (AIS)

○ Congenital Adrenal Hyperplasia (CAH)

○ Clitoromegaly

○ “Gender Dysphoria” or other medical term for transgender

○ Gonadal Dysgenesis

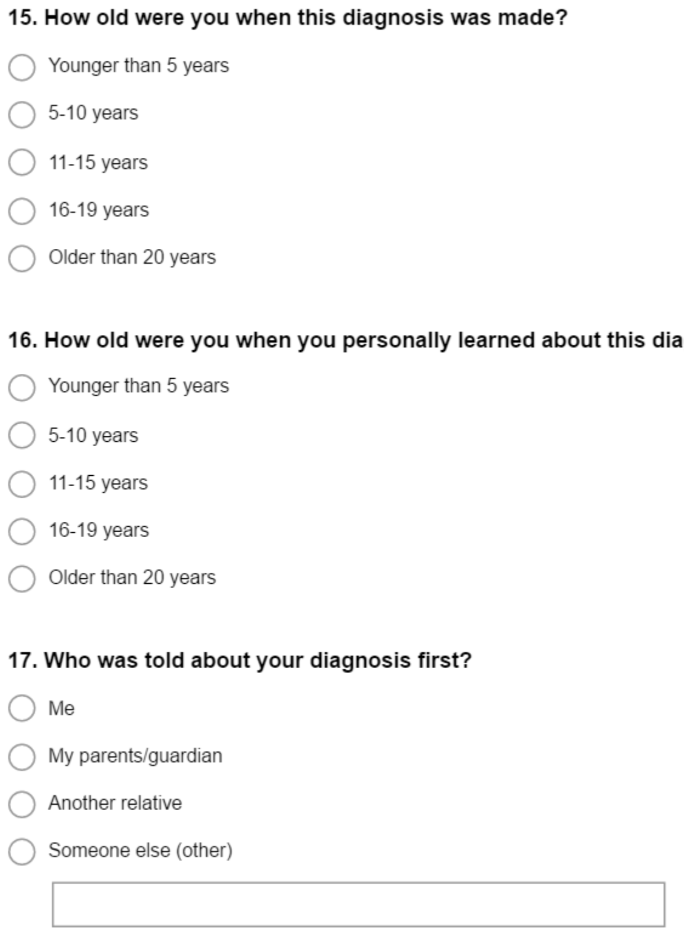
○ Hypospadias

○ Klinefelter Syndrome

○ Turner Syndrome

○ Swyer Syndrome

○ Other (please specify)



15. How old were you when this diagnosis was made?

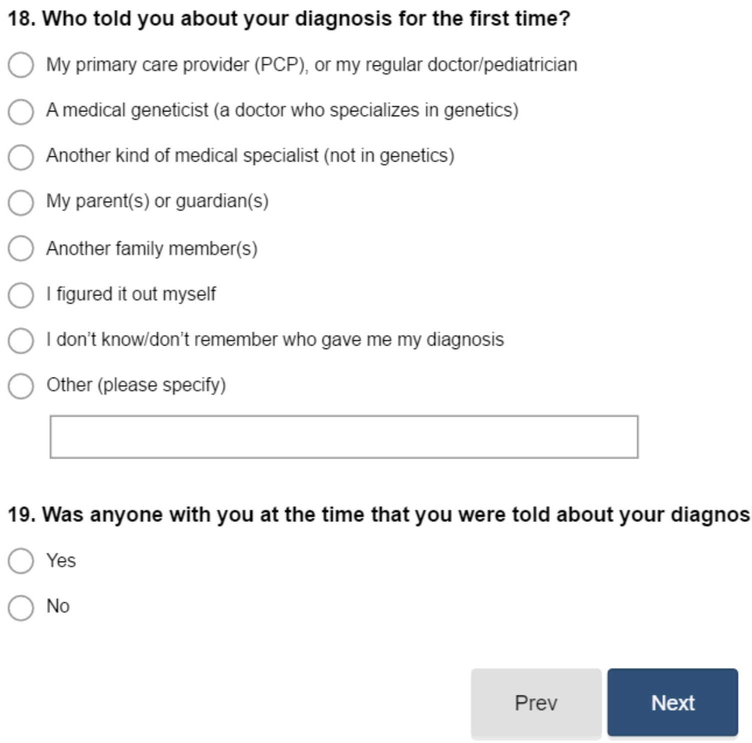
- ○ Younger than 5 years
- ○ 5-10 years
- ○ 11-15 years
- ○ 16-19 years
- ○ Older than 20 years

16. How old were you when you personally learned about this diagnosis?

- ○ Younger than 5 years
- ○ 5-10 years
- ○ 11-15 years
- ○ 16-19 years
- ○ Older than 20 years

17. Who was told about your diagnosis first?

- ○ Me
- ○ My parents/guardian
- ○ Another relative
- ○ Someone else (other)

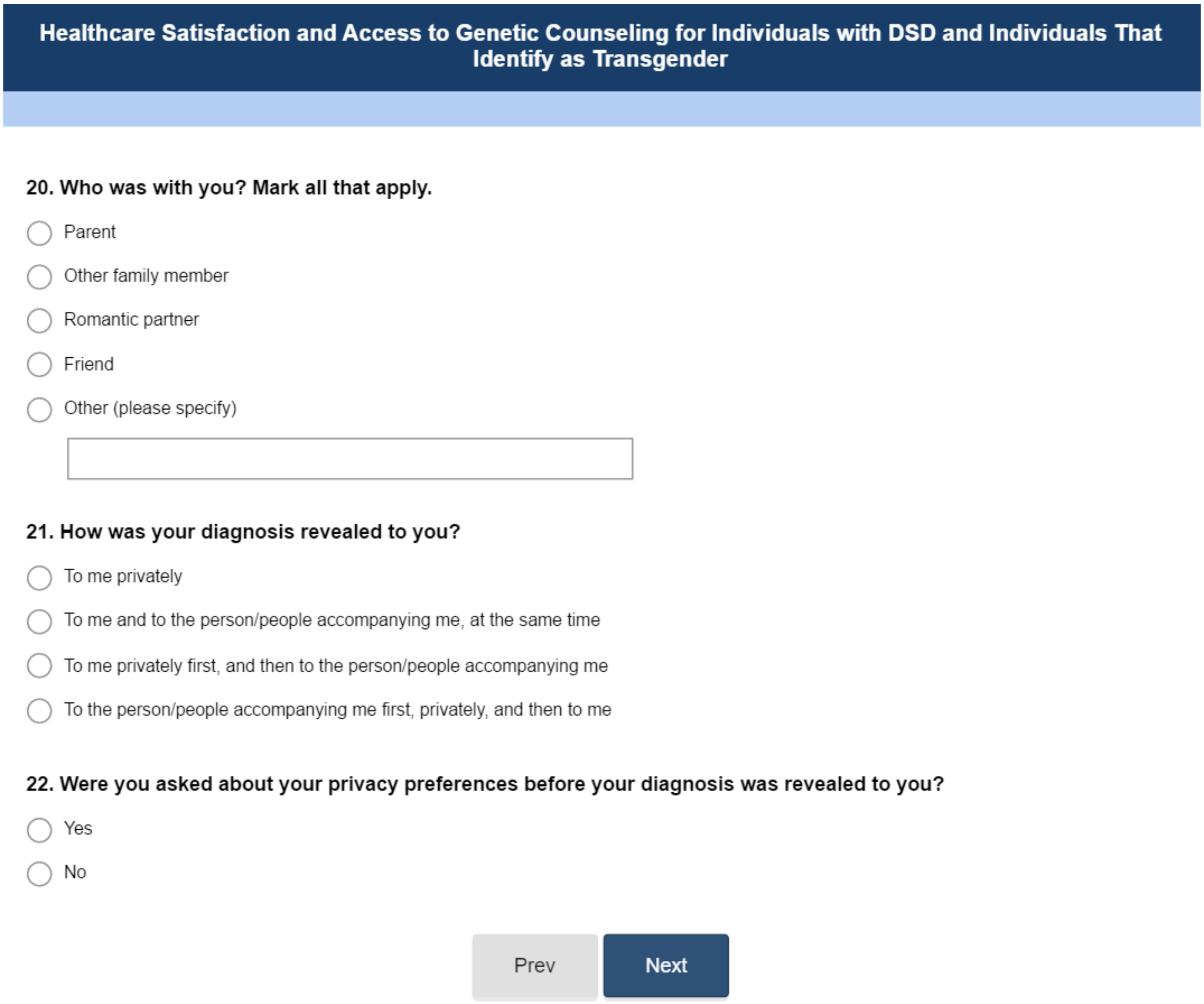


18. Who told you about your diagnosis for the first time?

- ○ My primary care provider (PCP), or my regular doctor/pediatrician
- ○ A medical geneticist (a doctor who specializes in genetics)
- ○ Another kind of medical specialist (not in genetics)
- ○ My parent(s) or guardian(s)
- ○ Another family member(s)
- ○ I figured it out myself
- ○ I don't know/don't remember who gave me my diagnosis
- ○ Other (please specify)

19. Was anyone with you at the time that you were told about your diagnosis?

- ○ Yes
- ○ No



Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

20. Who was with you? Mark all that apply.

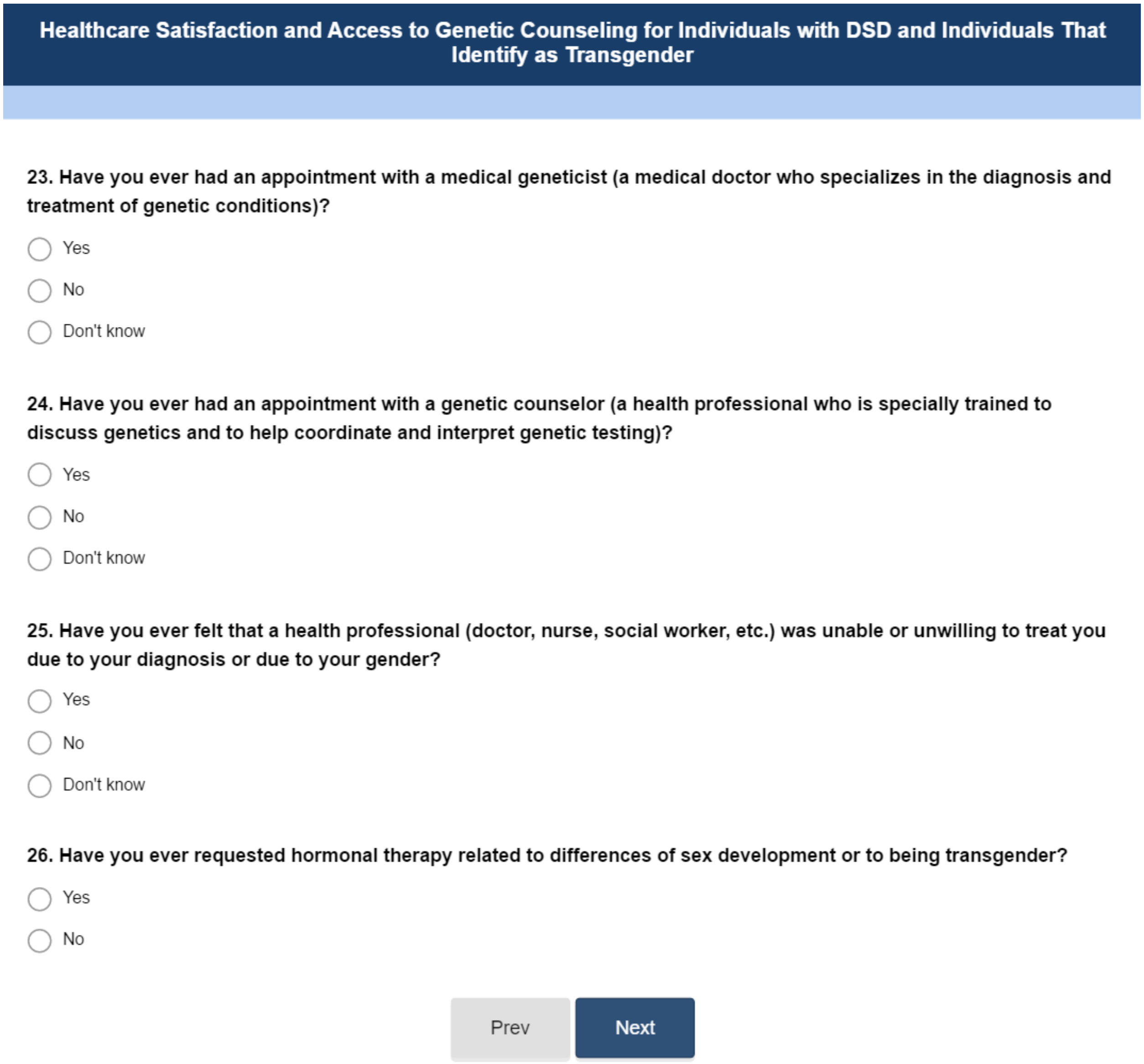
○ Parent
○ Other family member
○ Romantic partner
○ Friend
○ Other (please specify)

21. How was your diagnosis revealed to you?

○ To me privately
○ To me and to the person/people accompanying me, at the same time
○ To me privately first, and then to the person/people accompanying me
○ To the person/people accompanying me first, privately, and then to me

22. Were you asked about your privacy preferences before your diagnosis was revealed to you?

○ Yes
○ No



Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

23. Have you ever had an appointment with a medical geneticist (a medical doctor who specializes in the diagnosis and treatment of genetic conditions)?

○ Yes
○ No
○ Don't know

24. Have you ever had an appointment with a genetic counselor (a health professional who is specially trained to discuss genetics and to help coordinate and interpret genetic testing)?

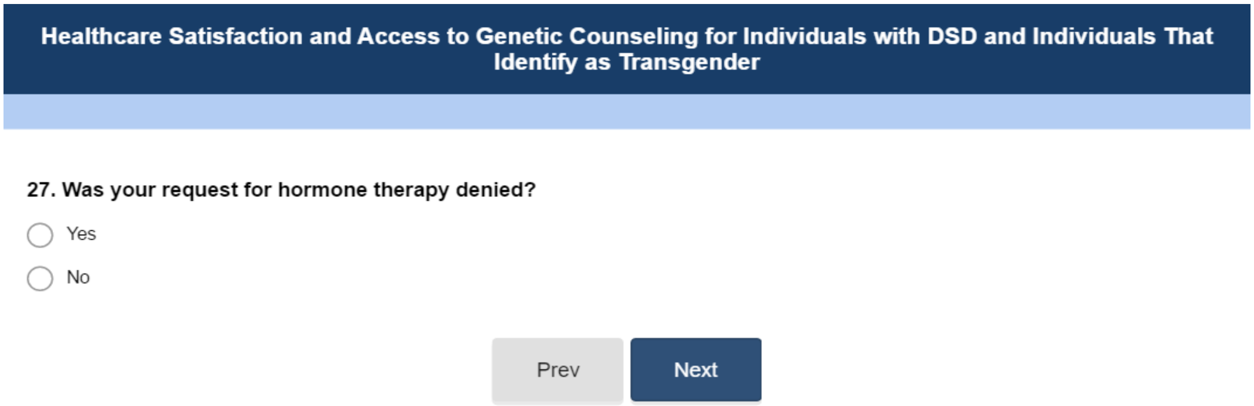
○ Yes
○ No
○ Don't know

25. Have you ever felt that a health professional (doctor, nurse, social worker, etc.) was unable or unwilling to treat you due to your diagnosis or due to your gender?

○ Yes
○ No
○ Don't know

26. Have you ever requested hormonal therapy related to differences of sex development or to being transgender?

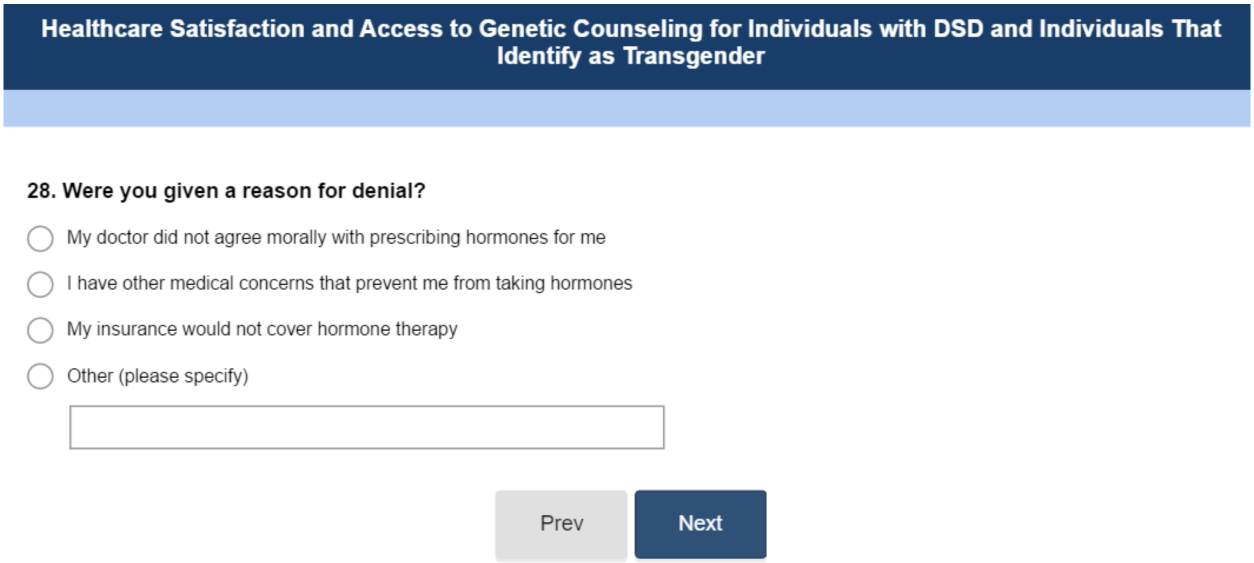
○ Yes
○ No



Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

27. Was your request for hormone therapy denied?

○ Yes
○ No



Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

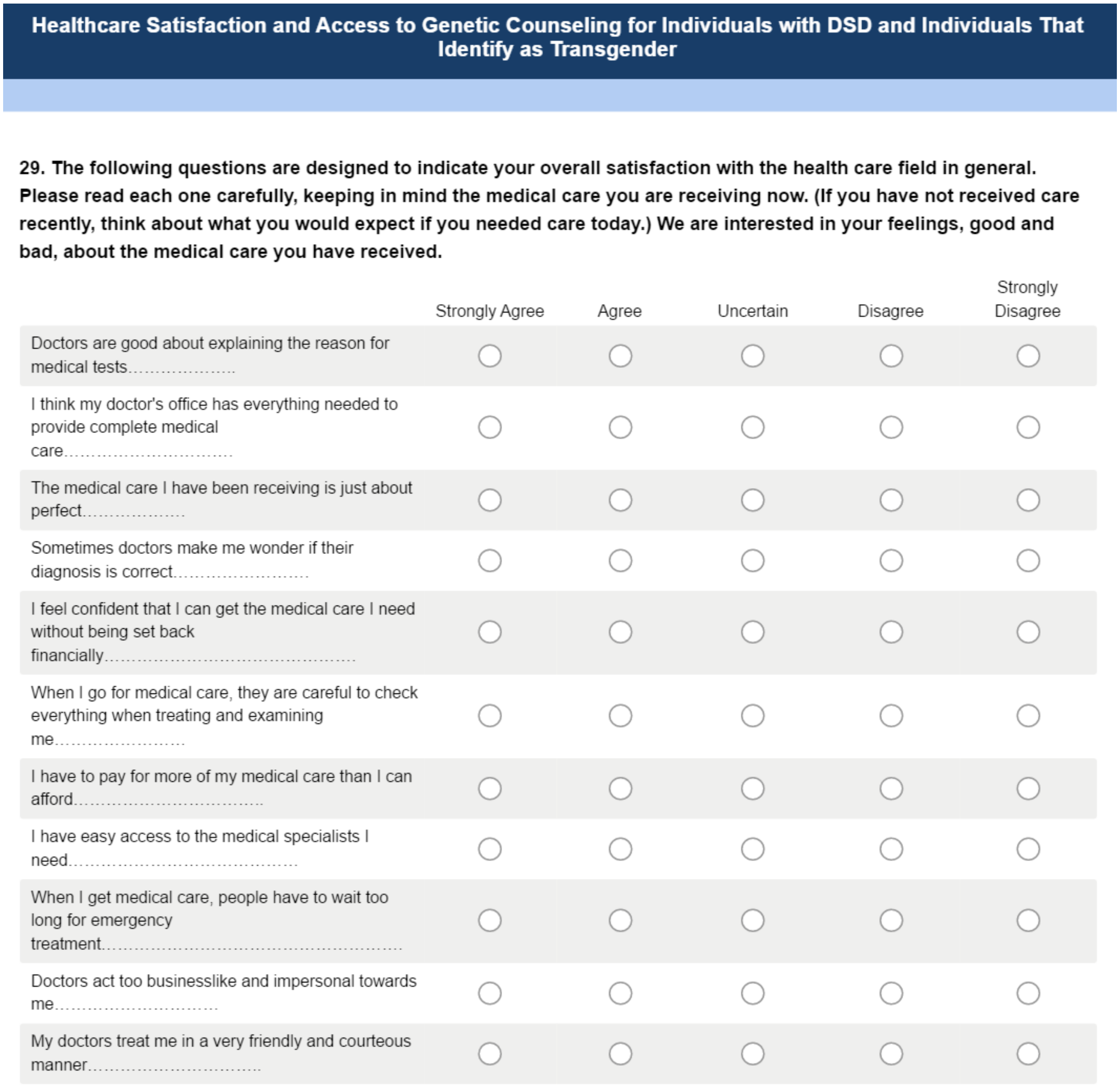
28. Were you given a reason for denial?

○ My doctor did not agree morally with prescribing hormones for me

○ I have other medical concerns that prevent me from taking hormones

○ My insurance would not cover hormone therapy

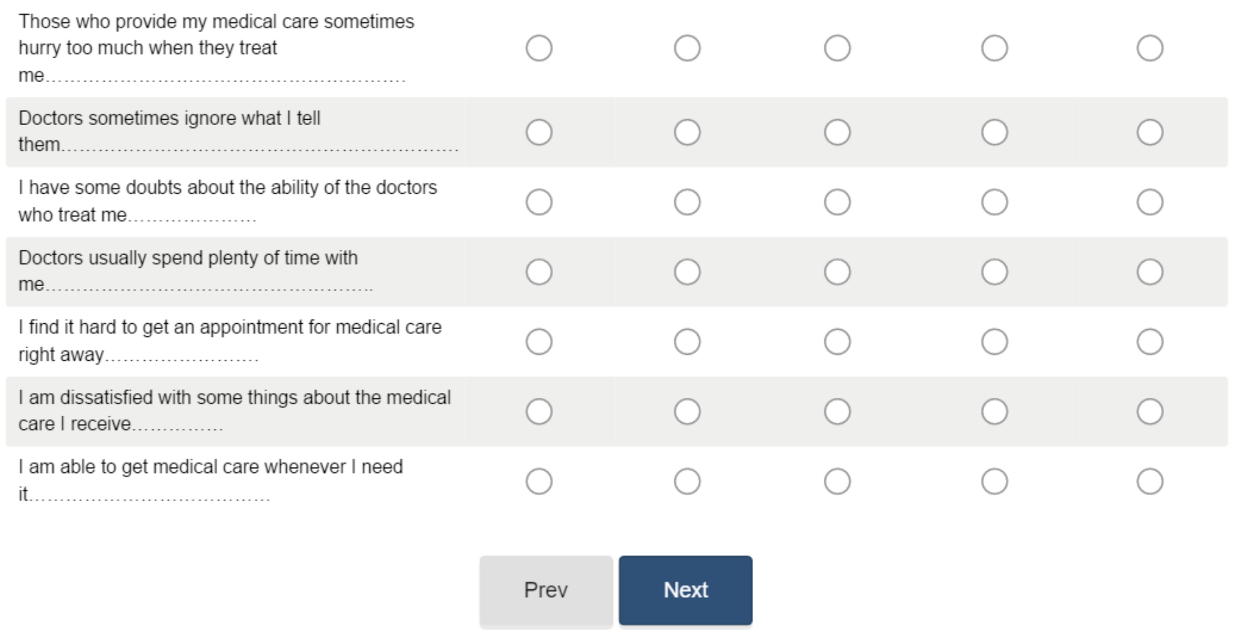
○ Other (please specify)



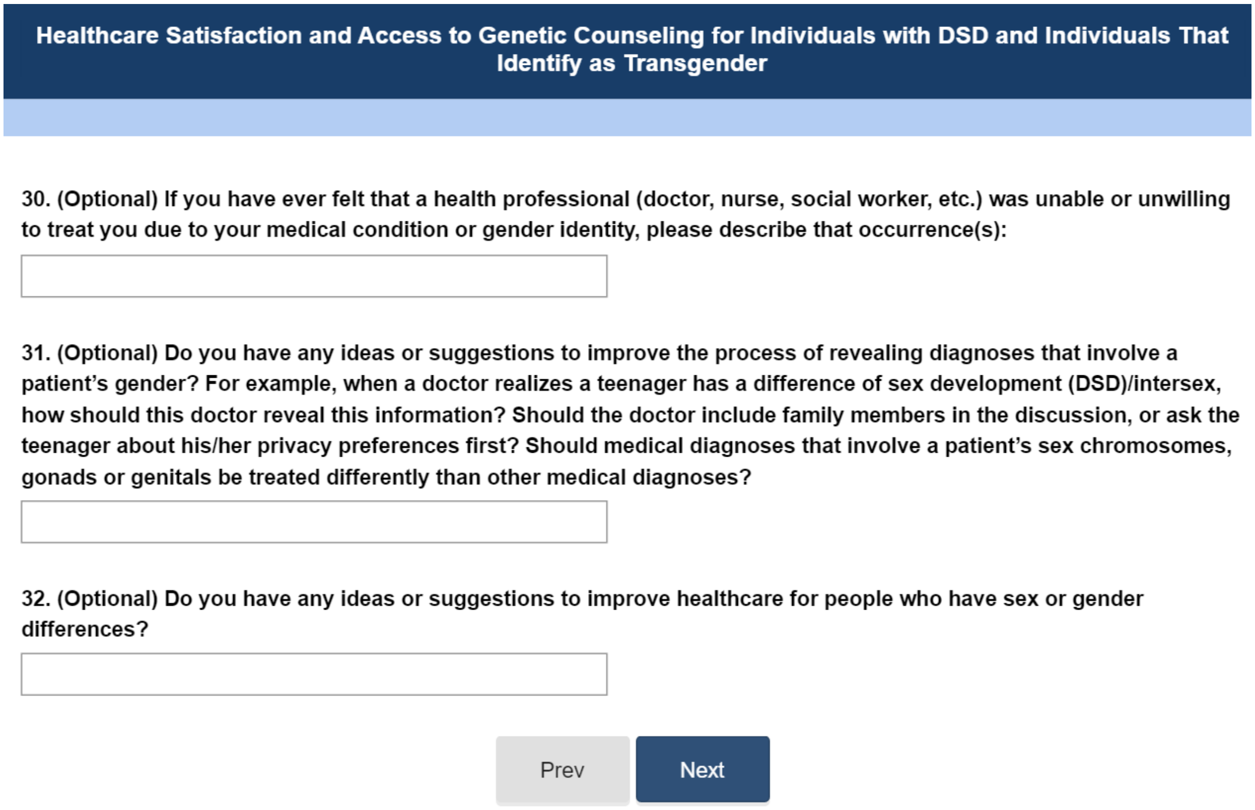
Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

29. The following questions are designed to indicate your overall satisfaction with the health care field in general. Please read each one carefully, keeping in mind the medical care you are receiving now. (If you have not received care recently, think about what you would expect if you needed care today.) We are interested in your feelings, good and bad, about the medical care you have received.

	Strongly Agree	Agree	Uncertain	Disagree	Strongly Disagree
Doctors are good about explaining the reason for medical tests……………….	○	○	○	○	○
I think my doctor's office has everything needed to provide complete medical care………………………….	○	○	○	○	○
The medical care I have been receiving is just about perfect……………….	○	○	○	○	○
Sometimes doctors make me wonder if their diagnosis is correct…………………….	○	○	○	○	○
I feel confident that I can get the medical care I need without being set back financially………………………………………	○	○	○	○	○
When I go for medical care, they are careful to check everything when treating and examining me……………………	○	○	○	○	○
I have to pay for more of my medical care than I can afford…………………………….	○	○	○	○	○
I have easy access to the medical specialists I need……………………………………	○	○	○	○	○
When I get medical care, people have to wait too long for emergency treatment……………………………………………	○	○	○	○	○
Doctors act too businesslike and impersonal towards me…………………………	○	○	○	○	○
My doctors treat me in a very friendly and courteous manner………………………….	○	○	○	○	○



Those who provide my medical care sometimes hurry too much when they treat me..	○	○	○	○	○
Doctors sometimes ignore what I tell them..	○	○	○	○	○
I have some doubts about the ability of the doctors who treat me....................	○	○	○	○	○
Doctors usually spend plenty of time with me...	○	○	○	○	○
I find it hard to get an appointment for medical care right away........................	○	○	○	○	○
I am dissatisfied with some things about the medical care I receive..............	○	○	○	○	○
I am able to get medical care whenever I need it.....................................	○	○	○	○	○



Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

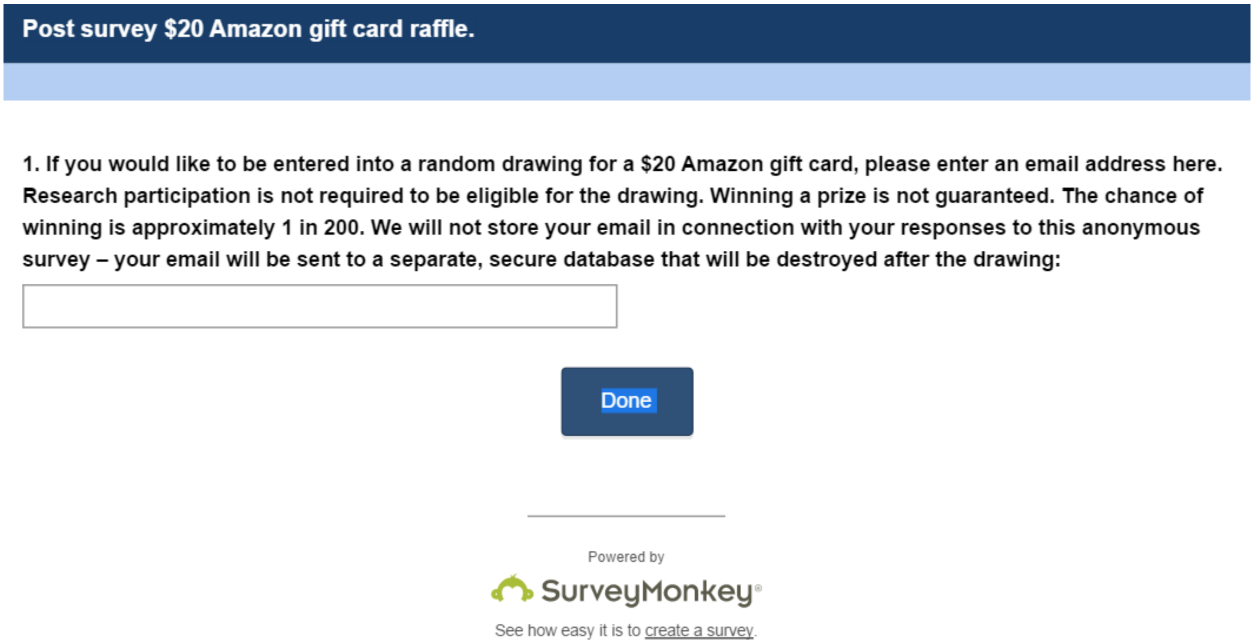
30. (Optional) If you have ever felt that a health professional (doctor, nurse, social worker, etc.) was unable or unwilling to treat you due to your medical condition or gender identity, please describe that occurrence(s):

31. (Optional) Do you have any ideas or suggestions to improve the process of revealing diagnoses that involve a patient's gender? For example, when a doctor realizes a teenager has a difference of sex development (DSD)/intersex, how should this doctor reveal this information? Should the doctor include family members in the discussion, or ask the teenager about his/her privacy preferences first? Should medical diagnoses that involve a patient's sex chromosomes, gonads or genitals be treated differently than other medical diagnoses?

32. (Optional) Do you have any ideas or suggestions to improve healthcare for people who have sex or gender differences?

Healthcare Satisfaction and Access to Genetic Counseling for Individuals with DSD and Individuals That Identify as Transgender

Thank you for completing the survey! Please click here if you would like to enter into a raffle for a $20 Amazon gift card.



Post survey $20 Amazon gift card raffle.

1. If you would like to be entered into a random drawing for a $20 Amazon gift card, please enter an email address here. Research participation is not required to be eligible for the drawing. Winning a prize is not guaranteed. The chance of winning is approximately 1 in 200. We will not store your email in connection with your responses to this anonymous survey – your email will be sent to a separate, secure database that will be destroyed after the drawing:

APPENDIX B: Request for Survey Distribution Blurbs

DSD Support group request:

Hello,

My name is Rebecca Freeman, and I am a graduate student at the University of California, Irvine, in the Genetic Counseling Master's program. I am currently working on my thesis project, which aims to assess the experiences of individuals with Disorders of Sex Development (DSDs) in healthcare, specifically with respect to disclosure of diagnoses, access to Medical Genetics services, and satisfaction with healthcare in general. My intent is to compare the experiences of people with DSDs to people who identify as transgender to see if there are similarities or differences between these groups with the long-term goal of improving healthcare for both.

While both DSD and transgender populations can be classified as falling outside of the typical gender binary, to my knowledge, no research has been done to compare/contrast these groups with respect to their experiences in the healthcare world. I believe my research will help clarify these perspectives, with the ultimate goal of helping to direct future improvement efforts in medicine— particularly in Medical Genetics.

We must educate health professionals about the unique challenges and needs of the DSD community and about how these needs should be distinguished from those of other groups with similarly socially-charged struggles. We have to work together to obtain the best possible healthcare for these potentially vulnerable individuals. With that in mind, I would greatly appreciate it if you would send the attached flyer and internet link to my anonymous survey to the members of your support group. The survey takes about 10-20 minutes to complete, and at the end of the survey there is an option to be entered into an anonymous drawing for a $20 Amazon gift card. Research participation is not required to be eligible for the drawing. Winning a prize is not guaranteed. The chance of winning is approximately 1 in 200.

The link to the survey is: www.genderhealthcaresurvey.com

Thank you for your time and consideration.

Sincerely,

Rebecca Freeman
Lead Study Researcher, Genetic Counseling Intern
Division of Genetic and Genomic Medicine
Department of Pediatrics
University of California, Irvine School of Medicine
Email: rebecclf@uci.edu
Phone: 714-456-5837

Under the supervision of Dr. Maureen Bocian, MD
Division of Genetic and Genomic Medicine
Department of Pediatrics
University of California, Irvine School of Medicine
714-456-7570

Transgender support group request:

Hello,

My name is Rebecca Freeman, and I am a graduate student at the University of California, Irvine, in the Genetic Counseling Master's program. I am currently working on my thesis project, which aims to assess the experiences of individuals that identify as transgender in healthcare, specifically with respect to disclosure of diagnoses, access to Medical Genetics services, and satisfaction with healthcare in general. My intent is to compare the experiences of people who identify as transgender to people who have been diagnosed with a disorder of sex development (DSD), to see if there are similarities or differences between these groups with the long-term goal of improving healthcare for both.

While both DSD and transgender populations can be classified as falling outside of the typical gender binary, to my knowledge, no research has been done to compare/contrast these groups with respect to their experiences in the healthcare world. I believe my research will help clarify these perspectives, with the ultimate goal of helping to direct future improvement efforts in medicine- particularly in Medical Genetics.

We must educate health professionals about the unique challenges and needs of the transgender community and about how these needs should be distinguished from those of other groups with similarly socially-charged struggles. We have to work together to obtain the best possible healthcare for these potentially vulnerable individuals. With that in mind, I would greatly appreciate it if you would send the attached flyer and internet link to my anonymous survey to the members of your support group. The survey takes about 10-20 minutes to complete, and at the end of the survey there is an option to be entered into an anonymous drawing for a $20 Amazon gift card. Research participation is not required to be eligible for the drawing. Winning a prize is not guaranteed. The chance of winning is approximately 1 in 200.

The link to the survey is: www.genderhealthcaresurvey.com

Thank you for your time and consideration.

Sincerely,

Rebecca Freeman
Lead Study Researcher, Genetic Counseling Intern
Division of Genetic and Genomic Medicine
Department of Pediatrics
University of California, Irvine School of Medicine
Email: rebecclf@uci.edu
Phone: 714-456-5837

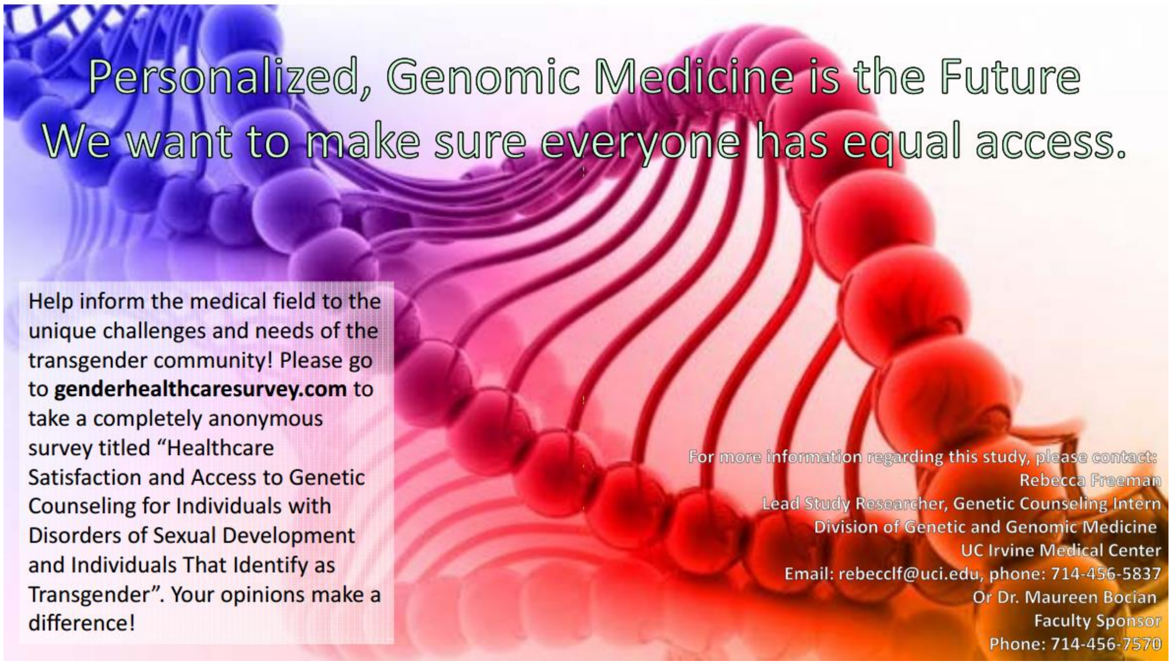
Under the supervision of Dr. Maureen Bocian, MD
Division of Genetic and Genomic Medicine
Department of Pediatrics
University of California, Irvine School of Medicine
714-456-7570

Facebook status:
Hi everyone! Please take a moment and help me out with my thesis project by taking this anonymous survey. We're hoping to hear from people who identify as transgender or who have been diagnosed with a disorder of sex development (DSD). Thank you, and please share for visibility ☺ To ensure proper handling and a quick response for any questions or concerns, please use the listed official contact information and not social media.
www.genderhealthcaresurvey.com

Twitter statuses:
First tweet:
DSD? Transgender? Please take a moment and help with my thesis project by taking this anonymous survey!
www.genderhealthcaresurvey.com #transhealth
Follow-up tweet:
To ensure proper handling for any questions or concerns, please use the listed official contact information and not social media :)

First tweet:
DSD? Transgender? Please take a moment and help with my thesis project by taking this anonymous survey!
www.genderhealthcaresurvey.com #intersex
Follow-up tweet:
To ensure proper handling for any questions or concerns, please use the listed official contact information and not social media :)

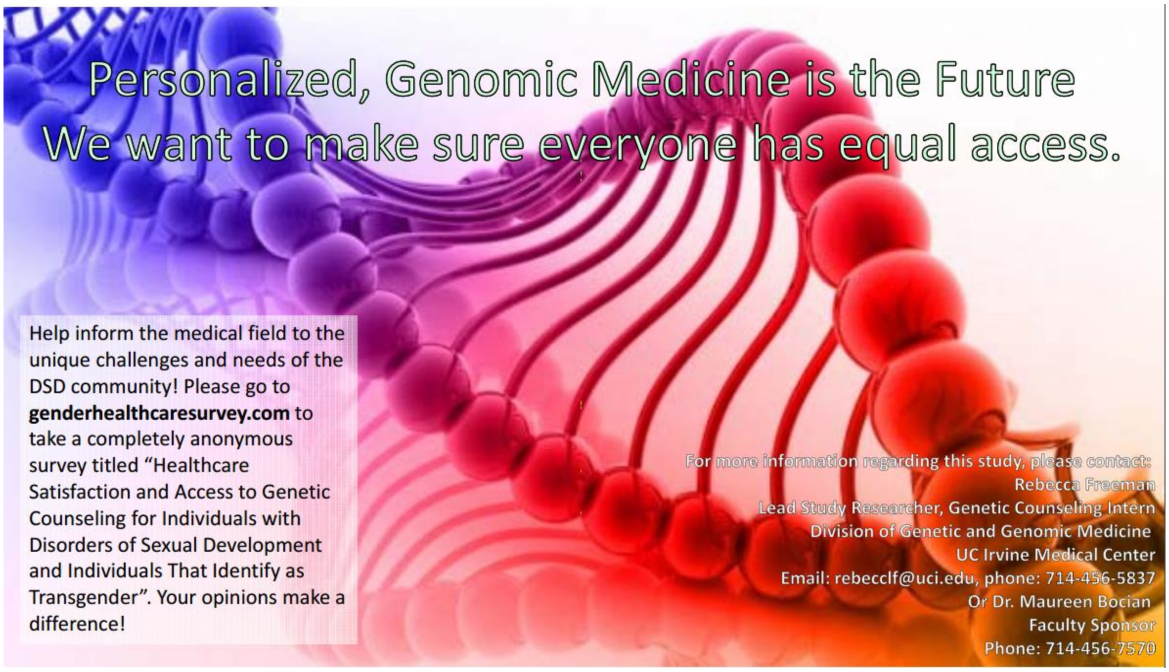
APPENDIX C: Flyers



Personalized, Genomic Medicine is the Future
We want to make sure everyone has equal access.

Help inform the medical field to the unique challenges and needs of the transgender community! Please go to **genderhealthcaresurvey.com** to take a completely anonymous survey titled “Healthcare Satisfaction and Access to Genetic Counseling for Individuals with Disorders of Sexual Development and Individuals That Identify as Transgender”. Your opinions make a difference!

For more information regarding this study, please contact:
Rebecca Freeman
Lead Study Researcher, Genetic Counseling Intern
Division of Genetic and Genomic Medicine
UC Irvine Medical Center
Email: rebecclf@uci.edu, phone: 714-456-5837
Or Dr. Maureen Bocian
Faculty Sponsor
Phone: 714-456-7570



Personalized, Genomic Medicine is the Future
We want to make sure everyone has equal access.

Help inform the medical field to the unique challenges and needs of the DSD community! Please go to **genderhealthcaresurvey.com** to take a completely anonymous survey titled “Healthcare Satisfaction and Access to Genetic Counseling for Individuals with Disorders of Sexual Development and Individuals That Identify as Transgender”. Your opinions make a difference!

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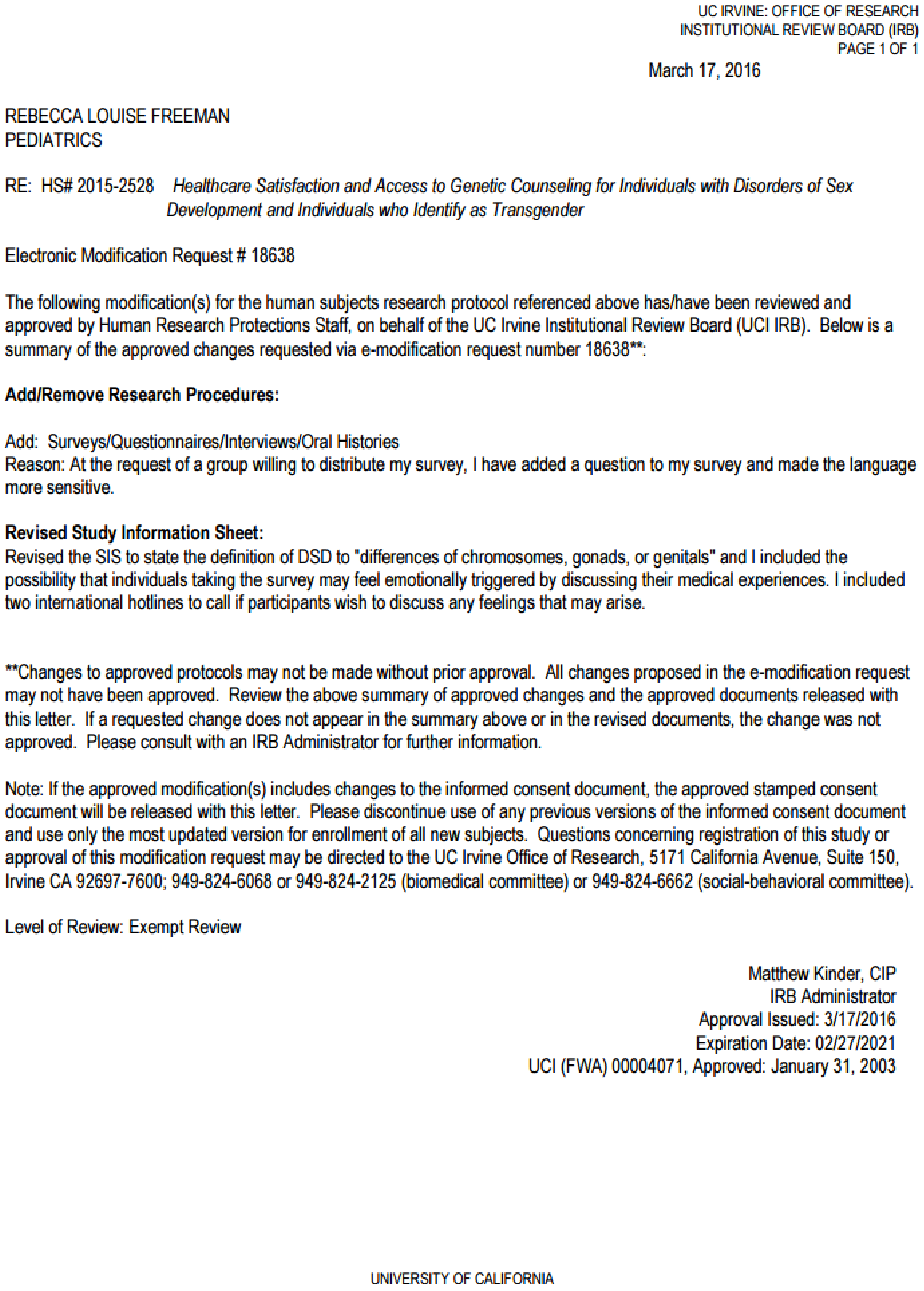
Division of Genetic and Genomic Medicine
UC Irvine Medical Center
Email: rebecclf@uci.edu, phone: 714-456-5837
Or Dr. Maureen Bocian
Faculty Sponsor
Phone: 714-456-7570

APPENDIX D: IRB e-mod approval letter



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March 17, 2016

REBECCA LOUISE FREEMAN
PEDIATRICS

RE: HS# 2015-2528 *Healthcare Satisfaction and Access to Genetic Counseling for Individuals with Disorders of Sex Development and Individuals who Identify as Transgender*

Electronic Modification Request # 18638

The following modification(s) for the human subjects research protocol referenced above has/have been reviewed and approved by Human Research Protections Staff, on behalf of the UC Irvine Institutional Review Board (UCI IRB). Below is a summary of the approved changes requested via e-modification request number 18638**:

Add/Remove Research Procedures:

Add: Surveys/Questionnaires/Interviews/Oral Histories
Reason: At the request of a group willing to distribute my survey, I have added a question to my survey and made the language more sensitive.

Revised Study Information Sheet:
Revised the SIS to state the definition of DSD to "differences of chromosomes, gonads, or genitals" and I included the possibility that individuals taking the survey may feel emotionally triggered by discussing their medical experiences. I included two international hotlines to call if participants wish to discuss any feelings that may arise.

**Changes to approved protocols may not be made without prior approval. All changes proposed in the e-modification request may not have been approved. Review the above summary of approved changes and the approved documents released with this letter. If a requested change does not appear in the summary above or in the revised documents, the change was not approved. Please consult with an IRB Administrator for further information.

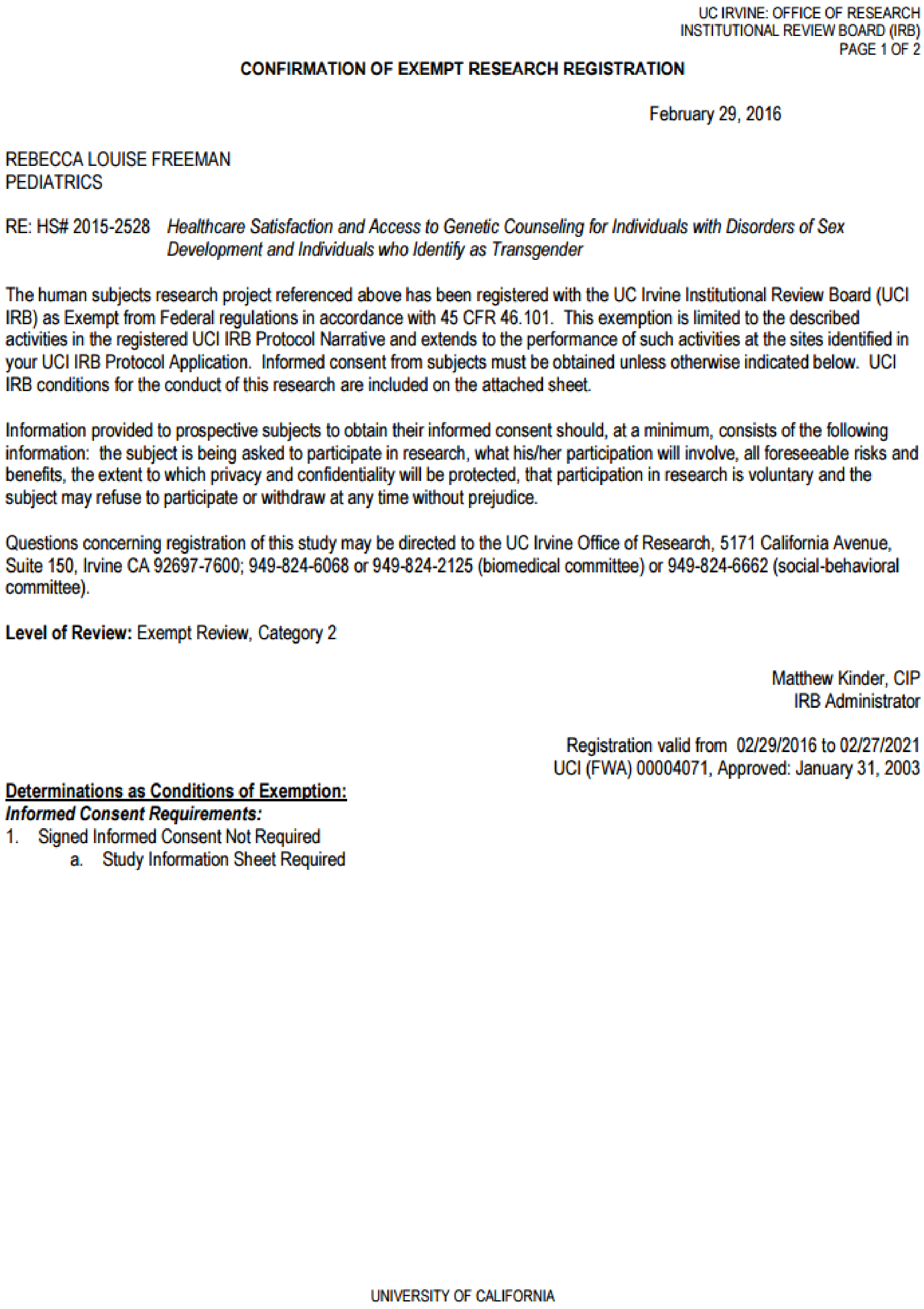
Note: If the approved modification(s) includes changes to the informed consent document, the approved stamped consent document will be released with this letter. Please discontinue use of any previous versions of the informed consent document and use only the most updated version for enrollment of all new subjects. Questions concerning registration of this study or approval of this modification request may be directed to the UC Irvine Office of Research, 5171 California Avenue, Suite 150, Irvine CA 92697-7600; 949-824-6068 or 949-824-2125 (biomedical committee) or 949-824-6662 (social-behavioral committee).

Level of Review: Exempt Review

Matthew Kinder, CIP
IRB Administrator
Approval Issued: 3/17/2016
Expiration Date: 02/27/2021
UCI (FWA) 00004071, Approved: January 31, 2003

UNIVERSITY OF CALIFORNIA

APPENDIX E: IRB Confirmation of Exempt Research Registration



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CONFIRMATION OF EXEMPT RESEARCH REGISTRATION

February 29, 2016

REBECCA LOUISE FREEMAN
PEDIATRICS

RE: HS# 2015-2528 *Healthcare Satisfaction and Access to Genetic Counseling for Individuals with Disorders of Sex Development and Individuals who Identify as Transgender*

The human subjects research project referenced above has been registered with the UC Irvine Institutional Review Board (UCI IRB) as Exempt from Federal regulations in accordance with 45 CFR 46.101. This exemption is limited to the described activities in the registered UCI IRB Protocol Narrative and extends to the performance of such activities at the sites identified in your UCI IRB Protocol Application. Informed consent from subjects must be obtained unless otherwise indicated below. UCI IRB conditions for the conduct of this research are included on the attached sheet.

Information provided to prospective subjects to obtain their informed consent should, at a minimum, consists of the following information: the subject is being asked to participate in research, what his/her participation will involve, all foreseeable risks and benefits, the extent to which privacy and confidentiality will be protected, that participation in research is voluntary and the subject may refuse to participate or withdraw at any time without prejudice.

Questions concerning registration of this study may be directed to the UC Irvine Office of Research, 5171 California Avenue, Suite 150, Irvine CA 92697-7600; 949-824-6068 or 949-824-2125 (biomedical committee) or 949-824-6662 (social-behavioral committee).

Level of Review: Exempt Review, Category 2

Matthew Kinder, CIP
IRB Administrator

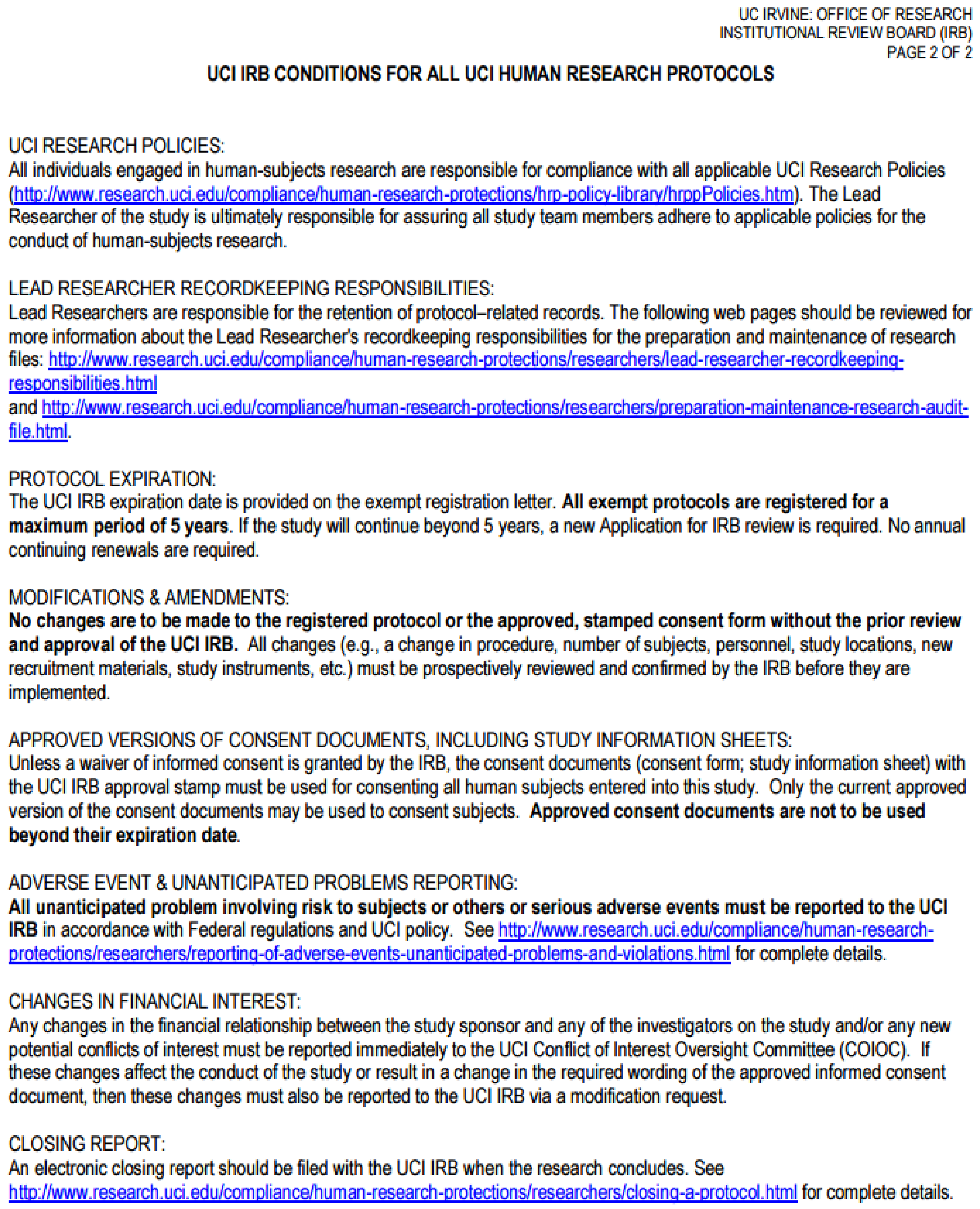
Registration valid from 02/29/2016 to 02/27/2021
UCI (FWA) 00004071, Approved: January 31, 2003

Determinations as Conditions of Exemption:

Informed Consent Requirements:

1. Signed Informed Consent Not Required
    a. Study Information Sheet Required

UNIVERSITY OF CALIFORNIA



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UCI IRB CONDITIONS FOR ALL UCI HUMAN RESEARCH PROTOCOLS

UCI RESEARCH POLICIES:
All individuals engaged in human-subjects research are responsible for compliance with all applicable UCI Research Policies (http://www.research.uci.edu/compliance/human-research-protections/hrp-policy-library/hrppPolicies.htm). The Lead Researcher of the study is ultimately responsible for assuring all study team members adhere to applicable policies for the conduct of human-subjects research.

LEAD RESEARCHER RECORDKEEPING RESPONSIBILITIES:
Lead Researchers are responsible for the retention of protocol–related records. The following web pages should be reviewed for more information about the Lead Researcher's recordkeeping responsibilities for the preparation and maintenance of research files: http://www.research.uci.edu/compliance/human-research-protections/researchers/lead-researcher-recordkeeping-responsibilities.html
and http://www.research.uci.edu/compliance/human-research-protections/researchers/preparation-maintenance-research-audit-file.html.

PROTOCOL EXPIRATION:
The UCI IRB expiration date is provided on the exempt registration letter. **All exempt protocols are registered for a maximum period of 5 years**. If the study will continue beyond 5 years, a new Application for IRB review is required. No annual continuing renewals are required.

MODIFICATIONS & AMENDMENTS:
No changes are to be made to the registered protocol or the approved, stamped consent form without the prior review and approval of the UCI IRB. All changes (e.g., a change in procedure, number of subjects, personnel, study locations, new recruitment materials, study instruments, etc.) must be prospectively reviewed and confirmed by the IRB before they are implemented.

APPROVED VERSIONS OF CONSENT DOCUMENTS, INCLUDING STUDY INFORMATION SHEETS:
Unless a waiver of informed consent is granted by the IRB, the consent documents (consent form; study information sheet) with the UCI IRB approval stamp must be used for consenting all human subjects entered into this study. Only the current approved version of the consent documents may be used to consent subjects. **Approved consent documents are not to be used beyond their expiration date**.

ADVERSE EVENT & UNANTICIPATED PROBLEMS REPORTING:
All unanticipated problem involving risk to subjects or others or serious adverse events must be reported to the UCI IRB in accordance with Federal regulations and UCI policy. See http://www.research.uci.edu/compliance/human-research-protections/researchers/reporting-of-adverse-events-unanticipated-problems-and-violations.html for complete details.

CHANGES IN FINANCIAL INTEREST:
Any changes in the financial relationship between the study sponsor and any of the investigators on the study and/or any new potential conflicts of interest must be reported immediately to the UCI Conflict of Interest Oversight Committee (COIOC). If these changes affect the conduct of the study or result in a change in the required wording of the approved informed consent document, then these changes must also be reported to the UCI IRB via a modification request.

CLOSING REPORT:
An electronic closing report should be filed with the UCI IRB when the research concludes. See http://www.research.uci.edu/compliance/human-research-protections/researchers/closing-a-protocol.html for complete details.

APPENDIX F: All response means to the PSQ-18, stratified by transgender and DSD groups

Subscale and Item	DSD and Transgender Mean	SD, SE	DSD Mean	SD, SE2	Transgender Mean	SD, SE3	p-value
General Satisfaction							
The medical care I've been receiving is just about perfect.	2.97	1.195, 0.128	2.95	1.244, 0.194	2.98	1.164, 0.172	0.917
I am dissatisfied with some things about the medical care I receive.	2.51	1.235, 0.131	2.45	1.273, 0.196	2.55	1.212, 0.177	0.703
Communication							
Doctors are good about explaining the reason for medical tests	2.62	1.16, 0.123	2.79	1.22, 0.188	2.47	1.1, 0.161	0.2
Doctors sometimes ignore what I tell them	2.73	1.26, 0.134	2.83	1.248, 0.193	2.64	1.293, 0.189	0.471
Technical Quality							
I think my doctor's office has everything needed to provide complete care	2.55	1.14, 0.122	2.80	1.002, 0.146	2.32	1.002, 0.146	0.046
Sometimes doctors make me wonder if their diagnosis is incorrect	2.97	1.2, 0.127	2.93	1.332, 0.206	3.00	1.083, 0.158	0.781
When I go for medical care, they are careful to check everything when treating and examining	2.92	1.17	2.86	1.241, 0.191	2.98	1.113, 0.162	0.627

me							
I have some doubts about the ability of the doctors who treat me	3.00	1.3, 0.140	3.00	1.361, 0.210	3.00	1.261, 0.188	1
Interpersonal Manner							
Doctors act too buisnesslike and impersonal toward me	3.48	1.08, 0.115	3.38	1.147, 0.177	3.57	1.147, 0.177	0.405
My doctors treat me in a very friendly and couteous manner	2.16	0.85, 0.09	2.19	0.943, 0.146	2.13	0.769, 0.112	0.739
Financial Aspects							
I feel confident that I can get the medical care I need without being set back financially	3.25	1.39, 0.148	3.20	1.382, 0.216	3.30	1.413, 0.206	0.732
I have to pay for more of my medical care than I can afford	3.24	1.37, 0.145	3.50	1.33, 0.205	3.00	1.367, 0.199	0.085
Time Spent with Doctor							
Those who provide my medical care sometimes hurry too much when they treat me	2.92	1.17, 0.124	3.00	1.126, 0.174	2.85	1.215, 0.177	0.552
Doctors usually spend plenty of time with me	2.74	1.07, 0.114	2.76	1.055, 0.163	2.72	1.097, 0.160	0.867
Accessibility and Convenience							

I have easy access to the medical specialists I need	3.01	1.23, 0.132	3.05	1.322, 0.206	2.98	1.164, 0.172	0.792
Where I get medical care, people have to wait too long for emergency treatment	2.91	1.09, 0.116	2.98	1.050, 0.166	2.85	1.122, 0.164	0.598
I find it hard to get an appointment for medical care right away	2.87	1.25, 0.132	3.00	1.230, 0.184	2.72	1.259, 0.184	0.337
I am able to get medical care whenever I need it	2.76	1.18, 0.125	2.60	1.211, 0.187	2.91	1.139, 0.166	0.203

APPENDIX G: All free-response answers*

*Answers are copied here exactly as the participant wrote them. *All typographical errors and misspellings have been left as-is*.

All responses to "If you have ever felt that a health professional (doctor, nurse, social worker, etc.) was unable or unwilling to treat you because of your medical condition or gender identity, please describe that occurrence(s):"

“No”

“When I was still assigned female in spite of a rather male external genital appearance, I had a hard time getting care that involved anything that had to do with me not conforming with my assigned sex/gender. I never had the surgery that doctors recommmended so I had no surgical SRS to make me conform with female. I wanted to get surgery for my hypospadias and until I found a surgeon who also performed surgery on FTM transgender people, I was never even considered for the surgery.”

“I once had a pediatric endocrinologist that always wanted to talk to my mom without me, but my mom would tell her "what ever you have to say you can say it to both of us"

“they lied / withheld truth /unwilling to deal with it”

“Mostly my experience with health care providers have been good and he bad has been more about not listening to my gender or assuming i am a female because of how i look”

"Lack of medical necessity" has been used throughout my transition to deny me service, such as facial feminization surgery, electrolysis, top surgery, and progesterone. Lots of health care professionals don't have any idea where to even start. I think it should be required that any new professional ask a new patient what their gender and pronouns are right off the bat. For every patient.”

“Doctor was uncomfortable explaining why he was prescribing estrogen when I did not know diagnosis.”

“Nobody knows the proper hormone therapy for turners syndrome. Every doctor I have been to has prescribed birth control. This is too low a dose for ovarian failure.”

“A Veterans Administration Primary Caregiver refused outright to take me a a patient.”

“Did not understand what was necessary to provide appropriate care. Asked me to find a doctor she could consult with, or seek care from a more knowledgeable care provider”

“Primary care clinic unwilling to prescribe ongoing testosterone dosage at level needed for me to have a normal, health testosterone level as male, because dosage is higher than "typical". I've been referred to 2 different endocrinologists. One was not local, so insurance denied coverage and I couldn't afford to go. Local endocrinologist unresponsive so far for scheduling. So far, local medical providers uncomfortable with my DSD and my testosterone prescription.”

"1) Company I work for had Kaiser and for the first few years could not use it unless it was not gender specific condition. When I did have a gender related issue, had to go before a panel of doctors before being allowed to make an appointment. 2) I have a blockage between the Uterus and the outside, plus stuck eggs. The Ultra-sound technician found the problem, no follicles on the Fallopian Tube. The Radiologist did three quick swipes of the area and then went looking for Urethra blockages. Why? Because I am XXY47 (normally Klienfelter) with both sets of female and male reproductive organs. This happens over and over because doctors assume Klinefelter and set out to prove it and discredit any information contrary to that. 3) To have a proper menstrual cycle (currently menses through the rectum) I would need surgery to disconnect the Uterus from the Prostrate and a Vagina created. The six Surgeons I talked to so far say they will remove all female reproductive organs and then do a Vagina. 4) Required to follow Benjamin Standards when I am not Transsexual."

"The problem is they have black and white solutions for a medical condition that results in only grays actually working."

"I went to the ER with a problem. As soon as they found out I was intersex they stoped everything and sent me home with no medical treatment of any kind. I almost died."

"I was misdiagnosed as a teenager, and denied the truth about my condition. My medical records were sent to a new Dr. Who shared them with a therapist. Both conspired to keep the truth from me. I diagnosed myself as a 50 year old, and got genetic confirmation."

"I was seeing a surgeon and they said they did not have the knowledge or expertise in treating my condition"

"emergency room. test came back as Gardenella and I waa incormed thar o ly women can have this infection"

"A gynecologist made innapropriate comments about my genitals during an exam. At an LGBT health clinic a doctor claimed I was lying about being intersex and said my body was "impossible" then encouraged me to let them examine my genitals. The same doctor refused to prescribe me hormones until I saw a therapist to pick a binary gender expression. A different doctor at the same LGBT health clinic insisted that I permit her to do genital exams and allow her to do a prostate exam to see if I had a prostate (even though I am under 30 with no symptoms of prostate issues, and 1% PSA level in bloodwork),"

"No I have never experienced this".

"I had one dr attempt to coercively perform a genital exam that had nothing to do with what i was there for because the were curious about my surgical results
When I first came out to my primary doctor and asked her if she'd be willing to prescribe HRT, she was unwilling to do so, but only because I was her first trans patient and she said she wouldn't have known where to start. She did, however, get me a referral to an endocrinologist who was willing to do so, and whom I still see a few times a year."

"Not respecting use of preferred name and pronouns, referring to a gender alignment procedure as though it were a procedure that people of my gender assigned at birth go through involuntary and concluding it would therefore cause depression, telling me I don't come across as a grown man but rather as androgynous and a 13 year

old boy"

"a doctor specializing in trans issues didn't like my nonbinary identity and concluded from the fact that I didn't want trans surgeries that I had internalized misogyny. this, she said, explained my testicle pain, without any tests or physical examination. I left in tears and eventually found another doctor when the pain intensified. she identified it as an infection, prescribed antibiotics, and fixed the pain. I'd been afraid to see another doctor after the first bad experience. I'm lucky it wasn't life threatening or my doctor's hostility to my gender could have killed me"

"This concerns a psychiatrist, not a physician, but one doctor I saw as a teen when seeking the letters I needed to start hormone therapy refused to provide a letter, suggesting instead a course of "treatment" to cure me of my gender identity. He had a religious objection to trans people."

"When I first arrived at UCSB from San Francisco, they had no doctors knowledgeable about transgender patients, so they refused to refill my existing prescriptions"

"A nurse misgendered me immediately after a gender affirmation surgery; when I corrected her, she said she'd already called me that and I hadn't corrected her then, and that she hadn't had time to check my chart, and she then complained audibly in the hall to another nurse about how "rude" I was. When I reported this, I was told to be patient with people who were learning and who weren't "progressive like us," implying whiteness. This is not the only time my gender has been completely disrespected and I have been gaslit by medical professionals."

"I came into the office, about 30 years ago, still fluid in my gender (very uncommon back then) and the doctor asked me basic questions about my sexuality. When I told him I didn't like to refer to a specific gender for me all the time (since it's all a mental construct and has nothing to do with my physical body) he sat there with a confused look. He asked if he could put down male. I told him that would be offensive and I might start crying. He told me to man up, which triggered me very hard and I started crying uncontrollably and yelling. He sent me out and said that he'd never serve a gender fluid (I quote) "class A weirdo" again"

"No, because I have only recently been out about my gender with doctors due to fear of this happening."

All responses to: "Do you have any ideas or suggestions to improve the process of revealing diagnoses that involve a patient's gender? For example, when a doctor realizes a teenager has a difference of sex development (DSD)/intersex, how should this doctor reveal this information? Should the doctor include family members in the discussion, or ask the teenager about his/her privacy preferences first? Should medical diagnoses that involve a patient's sex chromosomes, gonads or genitals be treated differently than other medical diagnoses?"

"Be honest and open, as a teenager I would have preferred to hear this first without anyone present. I am certain a teenager has the ability to make important decisions regarding their health. The teenager should aslo be asked if it is appropiate to talk to parents, care givers. This is done when it comes to birth control and reproductive issues and I believe a diagnosis of DSD certainly is in that group."

"When a resident talked to me about having XY chromosomes, they assumed I knew what it was supposed to mean and didn't ask me if I had any questions. They needed to break it down for me, ask me if I had any questions, and also offer me continued support with a counselor for someone to talk to (if needed). Since none of that happened, I spent several years knowing I was different, but not knowing fully why until I saw medical papers with my diagnosis on it and googled it. I was shocked!"

"team approach / no lying / truth at all costs / access to care / cost not a factor for patient"

"Should be treated differently! Psych help should be available on the spot & for regular follow-up"

"The teen should be asked about his/her privacy preferences first, then the parents should be included, but the person who is getting the diagnosis should definitely be involved at the initial outset. Yes, with more direct discussion to the patient most of all."

"I do not feel qualified to answer this question."

"I think that teenagers are capable of making their own decisions around what and how they want to reveal that info"

"An intersex diagnosis is not shameful nor is it an emergency. The important thing is that the affected individual be given the chance to make informed choices with the support of social workers or other supportive person who is not a member of the family. The family should be made aware that there are many happy people with DSD's and that genital surgery is only appropriate when the affected individual has made a fully informed decision."

"The doctor should not in any way participate in subterfuge. My endocrinologist knew that my parents were lying to me and kept the information she gave me at a minimum to collude.

I think that this is a whole family matter - esp. when others in the family have the syndrome and the ability to pass it along to their children."

"Nobody told me I had an intersex condition. I discovered the intersex aspect to my condition several months ago. I would have liked to read about it on my own."

"Much of the system seems designed to make DSDs go away rather than help understand and live with them. My body appears to have Mild androgen Insentivity, but getting a confirmation of whether or not that is true, or even to get a Dr to look at the evidence has been nearly impossible. I've been left on my own to deal with it alone. From what I can tell, it is "not of clinical interest if infertility is not an issue" so they simply do not want to deal with it. Simple peace of mind does not seem to be a priority, just treatable symptoms."

"A teen should certainly be asked about their privacy preferences first. I would not have wanted parents in the room during such a moment as a teen."

"Doctor should ask the teenager if he/she wants to know, before telling parents. Sex diagnoses should be handled with sensitivity, but given equal priority with other medical diagnoses."

"Discuss with person diagnosed first. If the patient is a minor child, this should take into consideration ethical legal consideration prior to treatment."

"Teenager should be told privately first. DSD diagnosis should be handled with more sensitivity and encouragement of autonomy for the person whose body is involved--no surgeries that are coerced, etc."

"Parent/guardian should be involved if under 18"

"For me, it is a miracle just to see a doctor prior to Obama Care, now it is all a scam. I see doctors and all they want to do is bill the insurance companies with the most expensive tests: Mammogram; Colonscopy; CT Scan; MRI with Contrast. How does that solve my hormonal problems?"

"They need to be told about their diagnosis by a entire group of doctors, that are on thier team. I never had one. They need to be told long before they ever have surgery."

"There is no such thing as a diagnosis of any sort that involves a person's gender. You are confusing sex (biology) and gender. I'm surprised you don't know the difference. My best advice is to not assume any diagnosis that involves one's biology is going to automatically involve that person's gender."

"Doctors should use appropriate language that's not stigmatizing but they should be honest"

"Ask the teen first. Have a counselor there. Be completely open about the diagnosis. Provide age-appropriate information. Medically, they should be treated the same, but realizing that there are social issues that need to be considered."

"As with any medical condition the doctor should be honest and explain the condition, and all possible treatment options."

"It can be traumatic; someone loved and compassionate should be with the client. Shouldn't be treated radically different from other non-fatal diagnoses.
Reveal in a way that is honest yet appropriate. Include family and provide family and patient counseling."

"When it is obvious at birth that a child has ambiguous genitalia, there should be NO "normalizing" surgery until the child is of age to give consent. Also, if it is known at birth that a child as a DSD they should be told information about their DSD as soon as they can comprehend, there should be not secrets and no shame at all."

"I believe the doctor should include the parents if the child is younger than 18. Doctor should be as gender neutral as possible in explaining diagnosis no statements like "you're actually a guy" or "genetically you are male" you can explain a DSD in gender neutral ways.
I would tell the parents and child together. My mom knew already and was in tears and denial when I walked into the room."

"Teens should be given information about their body first and discuss with the doctor how this information should be presented to parents. Support groups and additional resources should be provided with diagnoses of sex chromosomes, gonads, and genitals."

"I believe age is a large factor. The closer to adulthood they are, I believe they should be asked about privacy preferences. One thing I think should never be done is hide the condition, just if they're younger reveal along with the parents and explain it in a way they can understand without it being routed through parents who might be confused as well or hide it out of shame."

"Should definitely ask for privacy preferences first. Should be sensitive to the patients sense of self and identity regardless of medical diagnosis, as in not asserting that patients identity is or should be a certain way, not assuming patients emotional response, and making sure that they are respecting preferences including name and gender pronouns for example from the very start of the relationship with the patient."

"talk to the vulnerable young person first! any diagnosis that carries such a huge risk of hostility from caretakers should be privately discussed with the person first. this must be done empathetically, so it doesn't contribute to a sense of shame"

"I don't know, when I was a teenager, I was trying desperately not to be transgender"

"Tell that teenager first. Do not let parents pressure their child into medical treatment, certainly not before the person can form an opinion first. Don't diagnose without consent. Diagnoses can make accessing future care more difficult, or less difficult, and it all depends on what the person wants. Stop nonconsensual diagnostic records."

"This should definitely be kept between the patient and the doctor, unless the patient wants to involve their families."

"Ask the patient first and make sure they understand what's going on and that they have access to counseling and other support"

(Optional) Do you have any ideas or suggestions to improve healthcare for people who have sex or gender differences?

"Have more education on these subjects and not just be a chapter they skim over in class. "

"1. Drop the binary, if you have Intersex/DSD you are living proof that sex is not binary it is on a spectrum, hence binary barriers such as testosterone for female identified patients post gonadectomy with a diagnosis such as AIS should not be an issue. 2. Only you know your gender or sexual orientation nobody can tell you what this is, this needs to be respected by providers. 3. More evidence, there is a pathetic lack of evidence when it comes to the treatment of patients with DSD regarding their true needs. There has to be more research done for the community rather than on the community. 4. End childhood surgeries that conform to a certain sex/gender for the sake of cosmesis. You don't know if you are causing irreparable damage, 5. Involve advocacy groups and patient groups in DSD-teams, don't vilify advocates as fanatics or gender dysphoric individuals who just oppose medical interventions., 6. Educate medical providers on DSD, involve advocates in the process, a patient panel can be a very profound experience, it is more than just anecdortal as some want us to believe, 7. OB/Gyn providers educate your pregnant patients on the possibility of Intersex/DSD, we are as common as natural redheads. If expecting parents are prepared there will be much less panic or confusion once the child is born. I am sure there is more but I think I brought some important points across."

"My current doctor is great, everyone should practice like her. She takes time to listen, answer questions, she has talked with me about any concerns about sex or hormones and treats you like you are the only patients she needs to worry about for that time."

"don't lie"

"No corrective surgery. Psyc therapy for parents to help them cope, accept & be open with involved kid."

"I had one doctor ask for sex and gender on the initial forms, thsy way, she would know what to identify me as. More doctors should do this."

"We should not be treated in any way other than like a person with any other situation."

"Listen to what we say, don't follow the most conservative treatment plan when that comes at a personal cost to the patient, and stay up to date on what is considered acceptable and necessary in terms of treatment. Be our advocate as well as our Doctor."

"Ask gender and pronouns immediately. Change forms to allow spots for identified gender. Allow to use a preferred name at the pharmacy"

"They are not huge deal. People are different, gender identity can be complex. More and more young people are getting along well with creative gender identities and expressions. What we need is an open mind and an accepting society, not surgery to fix us."

"Honesty. Children deserve to know the truth about their bodies. Saying they won't understand is a a way to protect the parent/doctor, not the child."

"More education of providers in outlying areas, including techs (mammogram; bone density scan, etc.)"

"People have given me pregnancy tests when I felt ill from hormones. I can't get pregnant, and it was tactless. I understand that spontaneous pregnancies are possible, but it should not be the firth thing that comes to mind. Medication adjustments should be considered first. I would like to take one at home on my own if a doctor feels it is necessary."

"Be honest, listen to us and take our concerns seriously."

"Provide mandatory Trans/Intersex/gender training and workshops to providers."

"Education. All care providers should be required while in training to attend a classes(with grading associated) on physical and emotional care, professionalism, and cultural sensitivity. All currently practicing care providers should be required to take classes or seminars in appropriate care and cultural sensitivity for trans patients. And very importantly, all support staff should receive appropriate sensitivity training"

"Health care providers need more education about DSD/intersex, transgender, transsexual. They need to learn about "thinking outside the box" in order to treat those of us with genetic and other differences."

"Insurance needs to bloody pay for laser hair removal and electrolysis! I have NO money with which to do so. :("

“Yes, I hoped there were guidelines for us on the website as we age to be able to inform our physicians about what is needed.”

“Need to stop using the John Money Model. It is better to not have any information than wrong information.”

“Need to halt unnecessary sexual surgeries on children”

“Don't treat us like leppers & remember we are all different.”

“Don's assume sex or biology is the same as gender. Respect people's own assessment and identity of their sex or gender.”

“Doctors should be well educated on differences of sexual development so they can provide the best care for people that have DS doctors should be well educated on differences of sexual development so they can provide the best healthcare for people that have DSD.”

“A friend who was an ultrasound instructor used to say, "Treat the patient not the picture." Consider individual needs for every patient.”

“Don't just accept the body you have but prosper in it. Exercise, eat healthy, take good care of yourself and make the best of it.”

“Once diagnosed, it's vital to find a non-judgemental medical practitioner.”

“Don't be afraid to advocate for yourself. It makes you whole.”

“specialized teams that treat DSD, including psychiatrists/psychologists and therapists.”

“No infant cosmetic surgery unless the condition is absolutely life threatening. Doctors listening to patients first especially if that patient is an adult.”

“More education on these topics for Drs”

“More information programs in health science schools and public forums”

“Educate doctors, focus on education from patients instead of doctor to doctor education. Education from intersex organizations and intersex bodied researchers should come first. Acknowledge damage done in the past and currently happening to intersex people. Accept criticism and create a patient centered model of care for intersex people (specefically one that does not view us as disordered) similar to the WPATH standards of care for transgender people. WPATH's inclusion of intersex people (they use DSD language and concepts) is unnacceptable.”

“I have no ideas because I've never been treated differently as far as I know.”

*“I've noticed that many newly self-identified transgender people think they *must* see an endocrinologist for HRT, and are always surprised to find out that any MD/DO can write for hormones and that many (most?) doctors providing HRT and transition care are GP or Internal Med. Better education about options would help.”*

“Require all insurance, public or private or mixed, to treat any and all transition-related expenses as medically necessary, including voice training, hair removal, breast augmentation, and so on, in accordance with AMA recommendations.”

"Providers who specifically treat trans patients can be so respectful and make a person feel empowered and in control of their health, but many providers treating broadly can be a nightmare like psychiatrists, social workers, health insurance agents, dentists, and their supporting staff. I wish that these providers took the time to learn some Trans 101 and bedside manner, basic things like respecting the use of preferred name and pronouns for a start because they have a measure of authority and power and they need to use that responsibly to help people."

"use gender neutral language by default (affirming people's gender when they ask, but first doing no harm). insurance needs to cover stuff that people need early in transition, like hair removal for AMABs. not covering this leads to unhealthy depression and often to suffering violent hostility from others, which are obvious health hazards! it's not 'just cosmetic' when it drastically affects your life expectancy."

"Training and systems are necessary so that front desk staff and medical assistants use the right name when speaking to patients and calling them to exam rooms."

"Have safe zone training and education on Trans issues. Don't make access to care harder for non-binary people. Believe patients when they reveal information about themselves"

"More doctors familiar with it over a wide geographical area would be the best start. You shouldn't be afraid to leave San Francisco."

"Put pronouns in charts ON TOP. Put preferred names on all paperwork, and keep legal names segregated into the computer instead of printed on ID bracelets. Quit putting M or F on medical bracelets/records, ESPECIALLY when a patient is trans, non-binary, or intersex. Require all medical and reception staff to receive competency trainings, plural, on not assuming gender from appearance, on checking pronouns, on never ever misgendering someone especially while vulnerable recovering in a hospital. Stop gendering required "orientation" materials such as information on hysterectomies, especially when you KNOW the patient is not cisgender and non-intersex."

"I feel as if there should be more doctors available that specialize in hrt that are spread out in all different cities."

"Listen to your patients, don't assume people's genders"

"expand or change the M/F option at the top of forms - right away this is something that makes non-binary people uncomfortable. It also doesn't explain why a provider would need to know and may not give a provider all the information they actually need for good treatment/billing. It also can contribute towards discriminatory medical coding"

"Doctors should ask for pronouns, use preferred names only (I am called my legal name, which outs me). All personnel should get training on handling trans issues. My obgyn was great recently. I clearly stated I was trans when making the appointment, and the doctor and his assistant were very transfriendly, making no assumptions about my feelings about my body parts and how I felt about menopause."