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The Impact of Living with Klinefelter Syndrome: A Qualitative Exploration of Adolescents and Adults

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Abstract

Klinefelter syndrome (XXY) is a common yet significantly underdiagnosed condition with considerable medical, psychological and social implications. Many health care providers lack familiarity with XXY, resulting in medical management challenges and a limited understanding of the personal impact of the condition. Genetic counselors benefit from understanding the challenges adolescents and men with XXY face to effectively address their medical and psychosocial needs. The purpose of this study was to understand the impact of living with XXY as an adolescent or an adult. Individuals aged 14 to 75 years with self-reported XXY were recruited from online support networks to complete a web-based survey that included open-ended questions. Open-ended responses were coded and analyzed thematically (n = 169 to 210 for each open-ended question). Over half of respondents to the open-ended questions reported challenges in finding health care providers who are knowledgeable about XXY, with many describing an extensive diagnostic odyssey and relief when receiving a diagnosis. Individuals sought support coping with the challenges they face and acknowledgement of the positive aspects of XXY. Recommendations are made for how genetic counseling can enhance quality of life for individuals living with XXY.

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Conflict of Interest Amy Turriff, Ellen Macnamara, Howard Levy, and Barbara Biesecker declare that they have no conflict of interest.

Comment This manuscript is submitted solely to this journal, has not been published elsewhere, and is not currently under consideration for publication elsewhere.

Human Studies and Informed Consent All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all participants for being included in the study.

Animal Studies No animal studies were carried out by the authors for this article.

Keywords

Klinefelter syndrome; 47,XXY; Sex chromosome aneuploidy; Psychosocial impact

Introduction

Klinefelter syndrome (XXY) is a genetic condition that affects approximately 1 in 650 males, although the majority of individuals are undiagnosed as a result of considerable phenotypic variability and lack of familiarity and recognition among health care providers (Bojesen et al. 2003). With noninvasive prenatal testing, it is expected that diagnosis rates will increase and genetic counselors will provide counseling for XXY more frequently. It is imperative that genetic counselors and other health care providers understand the impact of living with XXY to provide adequate counseling and to help meet the medical and psychosocial needs of those living with the condition and their families.

Two major findings have emerged from several studies that have examined the counseling that follows a prenatal diagnosis of XXY. First, there is considerable variation in what health care providers know and communicate to parents receiving a prenatal diagnosis of XXY and also differences in their perceptions about the quality of life of individuals with XXY (Abramsky et al. 2001; Hall et al. 2001; Bourke et al. 2014). Second, the outcomes of pregnancies diagnosed with XXY appear to be influenced by the specialty of the clinician providing the counseling (Marteau et al. 2002). Those parents receiving post-diagnosis counseling from a genetics professional are more likely to receive positive information about the condition (Bourke et al. 2014; Hall et al. 2001) and are also more likely to continue their pregnancy (relative risk: 2.42) (Marteau et al. 2002).

Inconsistencies in the information provided to families and in health care providers' perceptions of what it means to live with XXY may be, in part, the result of significant inconsistencies present in the XXY literature. Some of the earliest publications described individuals with XXY as having increased rates of intellectual disability, psychosexual confusion, and criminality (Eriksson 1972; Kvale and Fishman 1965; Nielsen 1970). These findings likely reflect significant sampling bias, as research subjects were ascertained from prisons and mental health facilities. While a series of prospective longitudinal studies of children with XXY provided new insight into the cognitive, behavioral, and psychological implications of XXY (Bender et al. 1995; Evans and Hamerton 1990; Ratcliffe and Paul 1986), recent studies have found that some health care providers are still providing inaccurate, out-of-date information when counseling families (Bourke et al. 2014; Close et al. 2016). New research over the past decade has led to improvements in our understanding of the neuropsychological phenotype of XXY, the psychosocial impact of the condition, and advances in fertility treatments for XXY (Herlihy et al. 2011; Mehta et al. 2013; Ross et al. 2012; van Rijn et al. 2012). It is of critical importance for providers to convey updated, accurate information when counseling individuals and their families about XXY.

The purpose of the open-ended questions in this study was to understand the personal impact of living with XXY as an adolescent or an adult and to understand what individuals with XXY want their health care providers to know about their experiences.

Methods

Participants

This study's overall methods have been described in detail in our previous publications reporting prevalence and correlates of depressive symptoms and correlates of adaptation (Turriff et al. 2011, 2015). Briefly, individuals 14 years and older with self-reported XXY were recruited through national and international support networks via website postings, email listservs, newsletters, and online message boards. In addition, participants were recruited in person at a sex chromosome aneuploidy conference in Los Angeles, California, and at a XXY regional support group meeting in New York City.

Procedures

Prospective participants were invited to complete a self-administered web-based survey that contained quantitative measures and open-ended questions. Use of open-ended questions reduces the bias that we as professionals may project when asking close-ended questions and avoids missing opportunities to capture experiences we do not know enough to ask about. Compensation for participation was not provided. This study was approved by the National Human Genome Research Institute's Institutional Review Board (09-HG-N142).

Survey Instrument

The survey instrument included several quantitative measures that are detailed elsewhere (Turriff et al. 2011) and questions about demographics, features of XXY, and timing of diagnosis. This paper focuses on the responses to four open-ended questions. Study participants responded to two open-ended questions that captured the most challenging aspects of their diagnosis: "What is the worst part about having XXY when thinking about your life now?" and "What has been the worst part over your whole life?" These two questions were included in an attempt to qualitatively capture the impact of the diagnosis over time. Participants also responded to two questions asking, "In what ways has your XXY had a positive impact on your life?" and "What would you like your health care providers (your doctors, nurses, counselors) to know about having XXY?"

Data Analysis

Responses were entered into NVivo 10 (QSR International 2013) qualitative analysis software and conventional content analysis was conducted (Hsieh and Shannon 2005). Codes originated directly from the responses and were revised as needed. One coder applied the codes and a second coder recoded half of the responses using the same code-book. The coders discussed discrepant codes until a mutually agreed upon code was achieved. Inter-coder consistency was greater than 90%. Themes were identified and frequencies calculated. Differences in response patterns between adolescents and adults, as well as according to timing of diagnosis were analyzed.

Results

A total of 310 adolescents and adults with XXY completed the survey. Of those who reported their age, 41 participants were categorized as adolescents 14 to 24-years-old, and

199 were categorized as adults 25 years and older. Participant characteristics are summarized in Table 1. Open-ended responses for each question included in the thematic analysis ranged from 169 to 210 due to missing data. Responses were excluded from the thematic analysis (n = 6–21) if they were not written in English or if the participant did not respond to the question asked.

Though limited by sample size, there were not striking differences between adolescent and adult responses or according to timing of diagnosis. The nine individuals who were diagnosed prenatally had responses that overlapped with those individuals diagnosed in childhood, adolescence or adulthood.

Challenges of XXY

A total of 169 responses were included in the thematic analysis of the worst part of XXY when thinking about life now, and 170 responses were included in the thematic analysis of the worst part of XXY when thinking about your entire life. Responses to the two questions were similar overall, though there were a few important differences. Infertility and the psychological impact of XXY were the two greatest challenges faced by study participants. When thinking about life now, 31% of respondents indicated infertility was their greatest challenge and 27% mentioned psychological challenges. When thinking about their entire life, 27% of respondents mentioned infertility and 31% mentioned psychological challenges as a top concern. Adolescents and adults alike described feelings of sadness and loss when the prospect of biological fatherhood was threatened. Several participants elaborated that as upsetting as it was to learn that they may be infertile, the loss of control and loss of choice was particularly difficult to manage. A number of men indicated that their infertility had a significant impact on their relationships; some felt as though they had let down their partners and their families. Psychological challenges, including depression, anxiety, low self-esteem, and mood instability, were another top concern among participants. Some of the adolescents and adults described these challenges as endless, exhausting, and at times hopeless.

In reporting current challenges, participants described concerns about their appearance and testosterone treatment challenges. About 20% of respondents indicated that differences in appearance due to XXY, such as small testes, gynecomastia, increased height, or lack of muscle mass was the worst part about living with XXY. Many reported being the target of bullying as a result of differences in their appearance. Challenges with testosterone, reported by 9% of respondents, included the associated cost, difficulty in finding the right form and right level, and lack of consensus among health care providers about how it should be administered to achieve desired outcomes while minimizing adverse side effects.

When reflecting on living with their condition over the course of their life, individuals commented more frequently on their learning disabilities (23%). Adolescent participants reported challenges disclosing their diagnosis to friends and romantic partners. Adult participants recalled challenges they encountered when being diagnosed, including a late or missed diagnosis, negative interactions with health care providers, and the limited information made available after the diagnosis. Participants of both age groups commented on the social impact of their condition. Many reported difficulty making friends, feeling socially isolated, and being the victims of bullying. Others commented on feeling

misunderstood by others, including health care providers. Results of the thematic analysis are summarized in Table 2.

Positive Impact of XXY

A total of 210 individuals commented on the ways in which XXY has had a positive impact on their lives. Of those, 41 adults and 7 adolescents indicated that there was nothing positive about having XXY. These responses were excluded from the thematic analysis of positive outcomes. Major themes that emerged included increased empathy (20%), new life perspective or worldview (19%), and relief from learning what affects them (19%). Adult participants commented that being diagnosed validated the symptoms that they knew they were experiencing but that may have been disregarded by their physicians. Adolescent participants commented that having a diagnosis allowed them to accept themselves and to understand their differences. both the adolescents and adults mentioned that having a diagnosis also helped them to understand past experiences, provided them with something to cope with and adapt to, and allowed them to realize that they were not alone. Participants reported strengthening of relationships (16%), becoming a stronger person (8%), and having a sense of meaning in life (10%) as a result of their XXY. Many participants (15%) perceived they have many positive characteristics that they considered to be XXY traits. These included that they were kind, honest, and gentle individuals. While infertility and one's appearance were often mentioned as negative aspects of XXY, a subset of participants (7%) commented that not needing to use contraception and their appearance (6%), particularly their height and youthful appearance, were positive aspects of living with XXY. These results are summarized in Table 3.

Suggestions for Health Care Providers

Table 4 summarizes the recommendations study participants (n = 199) had for health care providers. The major theme that emerged from this open-ended question was their desire to find a health care provider who knows more about XXY than they do. Over 50% of respondents wanted their health care providers to know what XXY is, to know how to diagnosis it, how to treat it, and to know the side effects of the treatment being offered.

In addition to understanding the medical aspects of XXY, 36% of participants wanted their health care providers to better understand the psychosocial implications of living with XXY and to offer support and/or to refer them to the appropriate providers. This was the top response among adolescent participants.

Sixteen percent of all participants considered their health care providers to have poor bedside manner, particularly when addressing sensitive topics, and many perceived health care providers to hold negative assumptions about XXY. Several adolescents commented that they felt as though they had to prove their doctors wrong by achieving more than what they were led to believe they could achieve.

Discussion

Responses to these four open-ended questions provide descriptions and insight into the personal impact of living with XXY, as relayed by the adolescents and men living with this

condition. To date, this has been an under-addressed topic in the literature, but is one that is critical to providing quality genetic counseling to individuals and their families when a diagnosis is made.

A prenatal diagnosis of XXY is often considered incidental and some providers even dread it, as XXY is generally viewed as having mild clinical consequences, especially relative to the other trisomies routinely tested in the prenatal period. In many instances, families are unaware of the possibility of a sex chromosome aneuploidy diagnosis until they are faced with a decision about a pregnancy that is no longer “normal,” but often associated with a mild phenotype (Lalatta and Tint 2013; Petrucelli et al. 1998). A postnatal diagnosis of XXY is often delayed, if made at all, leaving families searching for answers to their son’s physical and neurodevelopmental concerns for years (Visootsak et al. 2013).

Several studies, including this one, have found that whether a diagnosis is made in the prenatal or postnatal period, families are unsettled by how unfamiliar health care providers are with the condition and its implications (Bourke et al. 2014; Close et al. 2016). A recent study by Close et al. found that as parents try to understand the impact of their sons’ condition, their questions about the favorable versus unfavorable characteristics of the condition largely go unanswered (Close et al. 2016). The descriptions of what it means to live with XXY provided by the adolescents and men in this study can be used to inform discussions about the full range of experiences, both positive and negative, of living with XXY.

Infertility and psychological morbidity were the greatest sources of concern and challenge among participants, regardless of age, followed by learning disabilities and differences in appearance. The inability to have biological children was experienced as profound loss for participants of all ages in this study. Many recounted the moment they learned of their infertility and their reactions of devastation and loss of control. These findings are supported by the broader infertility literature that has found male infertility to be associated with distress, stigmatization, loss of control, low self-esteem, guilt and anxiety (Cousineau and Domar 2007; Kedem et al. 1990). Studies of parents of boys with XXY have revealed that parents also struggle with their son’s infertility. A study of parents of 14 boys with XXY found that parents experienced vicarious grief and anxiety about their son’s infertility (Borelli et al. 1984). Moreover, they expressed concern about what this meant for their son’s gender identity, sexual orientation and function (Borelli et al. 1984), which was a concern also raised in a study of parents of 16 boys with XXY (Bourke et al. 2014). The assumption that infertility implies a difference in gender identity, sexual orientation or one’s ability to function sexually is unwarranted and critical for counselors to address.

While advances in fertility treatment offer hope of biological fatherhood to boys and men with XXY, it is important for health care providers to acknowledge the emotional impact of infertility and to facilitate effective coping, not only for individuals with XXY, but also for their families. Parents are critical in helping their children adapt to potential infertility by providing information in gradual steps appropriate to their son’s age and maturity level and by promoting open discussion and sharing of feelings. However, many parents are uncertain as to how to approach these conversations and may have questions or misunderstandings of

the implications of infertility, as found in the studies above (Borelli et al. 1984; Bourke et al. 2014). A recent study by Dennis and colleagues found that while there were many ways parents prepared themselves for conversations with their children, common concerns included how to present information in an age-appropriate manner, how to discuss infertility, and concerns on how disclosure may impact their child's self-esteem (Dennis et al. 2015). Genetic counselors could be integral in providing guidance to families on how to approach conversations, including how to discuss infertility, with their sons.

Depression and anxiety were another top concern raised by participants of all ages in this study, which poses a significant quality of life issue. Health care providers are becoming increasingly aware of the psychiatric and psychosocial challenges faced by boys and men with XXY (Bruining et al. 2009; Close et al. 2015; Geschwind and Dykens 2004; Herlihy et al. 2011; Ross et al. 2012; Tartaglia et al. 2010). Screening boys and men for depression, anxiety, and other mental health issues can facilitate early diagnosis and treatment, and should be a routine part of caring for individuals with XXY.

The struggle with learning disabilities was mentioned by adult participants often in the context of a late or missed diagnosis. Many participants reported going through school without a diagnosis, leaving them wondering why they struggled with learning and contributing to feelings of inferiority, low self-esteem and depression. Adolescent participants more often reported learning disabilities as a current challenge. Receptive-expressive language disorders posed social challenges for some, making it difficult to engage in conversations and to keep up with peers.

Differences in appearance were another source of social challenges. Participants described feeling self-conscious about various physical features, including small testes, gynecomastia, reduced muscle mass, tall stature, and decreased body hair. Several reported being the victims of bullying as a result of their physical appearance. Herihly et al. found poor body image and reduced self-esteem in a cohort of 87 men with XXY (Herlihy et al. 2011). Close et al. found that a higher number of XXY-related physical characteristics was associated with decreased quality of life and self-concept in a group of 43 boys with XXY (Close et al. 2015). These findings are supported by research that has revealed that perceived body image dissatisfaction is positively correlated with anxiety and depression and negatively correlated with self-esteem (Kostanski and Gullone 1998). Furthermore, teasing or bullying is also associated with anxiety, depression, suicidality, reduced self-esteem and low self-confidence (Pham and Adesman 2015). While bullying often occurs in childhood or adolescence, it has been shown to have long-lasting effects that persist into adulthood (Copeland et al. 2013; Takizawa et al. 2014). It is important for parents and health care providers to be vigilant in monitoring boys and men with XXY for body image issues, social challenges and bullying. Being proactive about these conversations and offering anticipatory guidance as to how to manage teasing or bullying if it should occur is warranted. Again, parents are critical in promoting self-esteem and body image in their children by promoting communication, setting a good example, reframing inaccurate beliefs, and creating a safe and loving home environment. Health care providers can help parents feel well-supported and equipped for managing these issues and should assist in identifying individuals who may benefit from additional counseling intervention.

Study participants were able to offer several positive aspects of living with XXY. Participants most often reported that as a result of living with XXY they were more empathic and had new life perspective and meaning. Some reported that they had become stronger people and had stronger relationships. New meaning, restored life values, and personal growth are all indicators of successful adaptation (Taylor 1983). Health care providers should work to identify and acknowledge individual's strengths and resilience to help promote self-esteem and self-efficacy in managing current or future challenges.

Notably, almost 25% (48/210) of respondents indicated that there was nothing positive about living with XXY. Although most people manage the stress of a genetic condition, some could benefit from help offered by their health care providers. In eliciting a person's narrative, genetic counselors can help individuals find meaning in their experience, identify personal strengths and identify effective coping strategies to ultimately facilitate adaptation.

Another positive aspect mentioned by participants was the relief of being diagnosed. Significant phenotypic variability, nonspecific or sometimes subtle symptoms and lack of recognition among health care providers has contributed to significant underdiagnosis of XXY. Studies in the United Kingdom and Denmark have suggested that 64–75% of individuals with XXY are not diagnosed in their lifetime (Abramsky and Chapple 1997; Bojesen et al. 2003). Of those diagnosed postnatally, approximately 25% are diagnosed in childhood or adolescence; the remainder are diagnosed in adulthood due to hypogonadism or infertility (Abramsky and Chapple 1997). Moreover, a recent study of parents of 89 sons with XXY found that the average time between initial parental concern and diagnosis of XXY was 2 to 5 years (Visootsak et al. 2013). The diagnostic odyssey can be very distressing and finally obtaining a diagnosis can be extremely validating for both patients and their families. It helps individuals and families to understand their experience, connect with others, access resources, and cope and adapt to living with a genetic condition. The adolescent participants, particularly those diagnosed prenatally or in early childhood, did not experience the distress of the diagnostic odyssey, but still mentioned considerable relief in having a diagnosis, as it allowed them to understand their differences and accept themselves.

Suggestions for health care providers provided by the adolescents and men in this study support and extend findings that have emerged from previous studies. The suggestion most frequently mentioned by the adolescents and men was to know more about XXY. Lack of awareness and knowledge not only contributes to late and missed diagnosis, but also presents significant medical management challenges. Participants in this study reported feeling unsupported and uninformed. They received a diagnosis but had no idea where to go from there. Similarly, parents have expressed feeling lost, unsupported and uninformed in making medical and educational decisions for their sons (Close et al. 2016). Without information and support from health care providers, individuals and their families are left to navigate and manage information on their own, which can be overwhelming, confusing and distressing. Participants wanted more support from their providers or wanted to be referred to other providers.

In addition to receiving quality information about testosterone treatment and its effects, fertility preservation and other medical aspects of the condition, adolescents and men also

wanted their health care providers to understand the personal impact of the condition. This included acknowledging both their strengths and positive attributes, in addition to the challenges they face. Participants reported feeling as though their condition was mild enough that the impact of living with the condition, both positive and negative, was largely ignored by providers. The personal experiences described in this study should offer insight to health care providers, highlight topics to explore in clinical encounters and help to enhance the patient-provider relationship.

Other recommendations included suggestions for improving communication in health care interactions. The insensitive nature in which participants were told about their infertility was a frequent example among the adolescents and men in this study. Participants wanted their providers to acknowledge the emotional impact of infertility in addition to providing medical information. A subset of respondents also perceived their providers to hold negative, inaccurate assumptions about what it means to have XXY. Some felt as though their providers viewed them as stupid, lazy or less masculine. While this may represent their perception and not the views of their providers, it does reflect the need for more effective and compassionate patient-provider communication and relationships.

While there was a limited number of adolescent participants relative to adult participants, there were not striking differences in response patterns between adolescents and adults or according to timing of diagnosis. This is somewhat unexpected, as we would have assumed that those diagnosed in the last fifteen years may have received different information and intervention than those diagnosed many years ago. Timing of diagnosis was not a significant predictor in our quantitative analyses (Turriff et al. 2011, 2015). Age, however, was negatively correlated with adaptation, which could suggest that older participants had experienced more challenges throughout their lives or were currently facing challenges to their adaptation process compared to younger participants (Turriff et al. 2015).

Although XXY is a common genetic condition, there are few providers who are truly expert in the condition. The establishment of multidisciplinary clinics for XXY will significantly improve the quality of care and guidance offered to individuals and their families and will improve the patient’s and their family’s experience with the health care system (Tartaglia et al. 2015).

Practice Implications

This study offers personal insight into the impact of living with XXY as an adolescent or an adult. In the case of a prenatal diagnosis, families may benefit from imagining what life could be like for their son and how this may impact their family. Parents could be encouraged to think about how they might react to specific challenges while being offered anticipatory guidance and support as to how others have managed similar challenges.

In the case of a postnatal diagnosis, this information could help individuals and parents better understand their experiences and to offer anticipatory guidance. Having additional insight into the personal impact of living with XXY could also help health care providers to more readily identify opportunities for intervention to promote quality of life and adaptation. For instance, issues with body image, self-esteem or bullying may be more salient among

adolescents and young adults, while the heartbreak of infertility may become more salient as people enter the age group in which people generally start families. Periodic screening for depression, anxiety and other mental health conditions should be a routine part of health care for all individuals with XXY.

Research Recommendations

Additional research that captures the conversations that occur during genetic counseling sessions or other health care interactions could help to highlight opportunities to further enhance counseling and care of individuals with XXY and their families. Research including multiple stakeholders (individuals with XXY, parents, families, health care providers, support group leaders) could provide additional guidance as to what information or descriptions of XXY should be included in genetic counseling and other health care interactions. Finally, additional research following those individuals diagnosed more recently is warranted, as the increase in prenatal diagnoses and the establishment of multidisciplinary specialty clinics likely will impact the experiences of individuals with XXY and their families.

Study Limitations

A number of limitations should be considered. Although the sample size is large, this study's findings are not generalizable to the experience of all adolescents and men with XXY. Those who did not participate in this study could have had sufficiently different responses to the four open-ended questions included in this study. Recruiting through advocacy groups could bias the results given that there may be differences between those who choose to join support groups and those who do not. Respondents were primarily Caucasian, which also limits the generalizability of the results. The limited number of adolescent responses relative to adult responses did not allow for direct comparisons to be made between the groups. Finally, this study did not include an open-ended question encouraging participants to identify aspects of their lives not affected by their XXY, which would have offered additional insight and helped to inform genetic counseling of individuals and their families.

Conclusions

Genetic counseling can be enhanced for individuals with XXY and their families by providing accurate, up-to-date information, acknowledging the psychosocial impact of living with XXY, preparing individuals and families for potential challenges, and facilitating effective coping. Genetic counselors may play an integral role as advocates to raise awareness about XXY among other health care providers.

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Table 1

Demographic characteristics of study population

Characteristic	*n* (%)
Age (years)	
Mean (SD)	40.7 (14)
Range	14–75
Timing of Diagnosis	
Prenatal	9 (4.1)
As a child, before 14 years	22 (10.0)
As an adolescent, 14–24 years	79 (35.7)
As an adult, 25 years and older	111 (50.2)
Race	
Caucasian	224 (92.2)
Black/African American	2 (0.8)
Asian	5 (2.1)
American Indian/Alaska Native	2 (0.8)
More than one race indicated	10 (4.1)
Ethnicity	
Not Hispanic or Latino	237 (97.1)
Hispanic or Latino	7 (2.9)
Education Level	
Elementary/Junior High	10 (4.1)
High school/GED	54 (22.2)
Technical school	33 (13.6)
Some college	55 (22.6)
Completed college	58 (23.9)
Post-graduate	33 (13.6)
Marital Status	
Single	97 (39.8)
In a partnered relationship	28 (11.5)
Married	93 (38.1)
Divorced or separated	24 (9.8)
Widowed	2 (0.8)
Children	
No	188 (76.4)
Yes	58 (23.6)
Relationship to Children	
Adopted	27 (41.5)
Step-children	12 (18.5)
Donor sperm	17 (26.2)
Biological	9 (13.8)

Table 2

Challenges of XXY

Challenge	Now *n* (%)	Whole Life *n* (%)	Quote(s)
Infertility	53 (31)	46 (27)	"The worst moment was when the doctor explained the infertility aspect. My thoughts and dreams ended at a full stop. I have become a fatalist." (33, 14[a]) "For me, the worst part has got to be the sterility. I always thought I would naturally have a family eventually and raise kids. But when I found out I was sterile (and tested for certain), I became further depressed for quite a long time—thinking there was no way I'd ever find a woman who would want me with such a genetic "defect." It's also not so much that I can't father children, it's more that I had no choice in the matter." (Age not reported) "The worst part was finding out at such a young age that I could never be what my mother had wanted me to be—that I could never have children." (Age not reported)
Psychological Impact	46 (27)	53 (31)	"Dealing with depression is like a roller coaster—endless." (28, 16) "I have suffered from anxiety and depression since I was about 10." (59, 57)
Appearance/Body Image	35 (21)	8 (5)	"Feminized body proportions, breast development, and small testes made adolescence and early adulthood (especially activities such as PE class, tennis, etc.) socially challenging. I still have to compensate." (54, 45)
Learning Disabilities	27 (16)	39 (23)	"The worst part has been dealing with activities that require auditory processing, such as trying to process listening exercises in foreign language class, listening to comedians, or even responding to open-ended questions such as the popular greeting 'how are you?'" (19, prenatal)
Social Challenges	22 (13)	21 (12)	"I was teased by other children until I graduated from high school. I had difficulty making friends because I was shy, had a poor self-image, and didn't fully understand the people around me." (45, 35) "XXY has had a profound effect on me socially. I am still very shy and only have a few close friends." (46, 45)
Testosterone Treatment Challenges	16 (9)	4 (2)	"Getting testosterone levels to work for me. Different doctors have different opinions on what's the right amount for my desired outcome." (56, 21)
Co-morbidities	13 (8)	8 (5)	"The health problems—like osteoporosis." (31, 15)
Romantic Relationship Challenges	13 (8)	9 (5)	"The worst part is not having a long term relationship and feeling unloved." (42, 30)
Misunderstood by Others	10 (6)	10 (6)	"I've felt very misunderstood for most of my life and still do." (32, 27)
Challenges with Health Care Providers	8 (5)	24 (14)	"Lack of knowledge by medical professionals and being treated differently based on outdated and inaccurate information." (34, 14) "The failure of doctors to explain or listen. I've had 30 years of 'it's not my field, you know more than I do.'" (67, 36)
Late Diagnosis	9 (5)	15 (9)	"Finding out at the age of 28, even though all of the symptoms have always been there." (28, 28) "Had I known about it much sooner, I could've gotten the help that I needed." (46, 45)
Employment Problems	4 (2)	9 (5)	"I don't have a career and I've been struggling to even remain employed - which largely comes down to my learning difficulties. Now, I suspect, too, that there might be other things going on as well - I can bet that there were some interpersonal "signals" that I completely don't see or ignore or whatever the heck goes on - but how can I fix what I can't observe?" (44, 35)
Disclosure Challenges	2 (1)	10 (6)	"Keeping it a secret. That shame was instilled early on." (34, 14) "Explaining to a new partner anytime I meet someone what I have and that I will never be able to have children." (22, 13)

[a](Age, Age at Diagnosis)

Table 3

Positive impact of XXY

Positive Aspect	***n* (%)**	**Quote(s)**
Increased Empathy	32 (20)	"The positive effects for me would be a higher sensitivity to others' feelings and greater sense of empathy. It has given me a deeper insight on the pain others feel when they are pointed out as being different." (Age not reported)
New Perspective or Worldview	31 (19)	"It has allowed me to see the world differently. It has allowed me to assist others and to change the focus of my life since my diagnosis occurred." (39, 26)
Relief of Diagnosis	31 (19)	"It was a relief knowing there actually was a definable cause for the questions, feelings, and issues I was going through…that others shared the same problems, that I was not alone." (44, 33) "Finding out that I have it helped me to accept that I am different. It's helped me to stop battling so hard from within." (56, 53)
Strengthened Relationships	26 (16)	"I've had to realize that life isn't all about having kids, so I've focused on my other relationships. It's made me a better uncle and a better friend." (30, 19)
XXY Traits (kind, honest)	25 (15)	"I am patient, kind, and always willing to help." (53, 46)
Gave Life Meaning	17 (10)	"I feel like God put me here to help others with this condition." (59, 24)
Made Me a Stronger Person	13 (8)	"This experience made me a very strong person." (58, 38)
Impact on Sex Life	12 (7)	"It's crazy, I guess, but because I can't get a woman pregnant, it means I no longer need a condom, so I think sexual experiences are better." (39, 28)
Appearance (Height, Youthful Appearance)	10 (6)	"My height. I'm 6′2″." (29, 20)

Table 4

Suggestions for health care providers

Suggestion	*n* (%)	Quote(s)
Know More about XXY	112 (56)	"90% of health care providers do not have a solid understanding of XXY and I always have to tell them about it. I find that extremely embarrassing." (35, 23) "We're all different, and since we AREN'T XY, constantly comparing us to normal males isn't particularly helpful. I want to know what the standard is for XXY's not XY's - I want help to be the best XXY I can be, not instead being constantly compared to a "gold standard" of a "normal male". Aside from such a comparison being patronising, it doesn't help me - they're never going to change the chromosomes in all my cells, so it's an impossible standard." (44, 35)
Understand What It Means to Live with XXY	72 (36)	"I would like for them to acknowledge that this is something that causes challenges and that it shouldn't be ignored or brushed under the rug." (32, 27)
Improve Communication/Bedside Manner	32 (16)	"They should know that not everything they read on the internet is true when it comes to people who have XXY. Also, when telling someone they can't have kids at 15, be a little more caring and act like you really care what the other person may be feeling." (36, 15)
Support and/or Referrals Needed	18 (9)	"There is much more to us than hormone replacement. Some of us need help with a lot of stuff like learning disabilities, life skills, psychological help." (28, 14) "They need to take more of an interest and try to head off problems for those of us…to be advocates for us." (59, 49)
Diagnose Earlier	13 (7)	"Diagnose it earlier. I was 25 and that was too late to help me." (Not reported, 25)