

The Expert in the Room: Parental Advocacy for Children with Sex Chromosome Aneuploidies

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ABSTRACT: *Objective:* Owing to fragmentation in the medical system, many parents of children with disabilities report taking on a care coordinator and advocate role. The parental advocacy and care coordination requirements are further amplified in this population because of a lack of awareness about sex chromosome aneuploidies (SCAs) in medical and social services settings, as well as the complex needs of affected children. This burden disproportionately affects mothers and low-resource families as a result of gendered ideas of parenthood and social stratification in resource access. The aim of this study is to understand the unique parental burdens of SCAs and family support needs. *Methods:* We conducted 43 interviews with individuals with SCAs and/or their parents, and qualitatively coded and analyzed the transcripts for themes relating to parent advocacy, medical services, social and educational services, and coping. *Results:* Our findings indicate that parents must repeatedly advocate for their concerns about their child to be taken seriously before diagnosis and continue to advocate for services and interventions throughout childhood and adolescence. Parents also report the need to educate health care professionals about their child's medical condition. A majority of the parent participants were women, and single mothers reported high levels of emotional burden. *Conclusion:* Parents of children with SCAs shoulder additional roles of medical advocate and care coordinator. This causes excessive burden on families but also disadvantages families in which parents are unable to act as an advocate for their child.

(*J Dev Behav Pediatr* 00:1–7, 2020) **Index terms:** sex chromosome aneuploidy, advocacy, social services, medical services, gender.

Taken together, sex chromosome aneuploidies (SCAs) are relatively common chromosomal aneuploidies and are represented in approximately 1 in 400 individuals.[¶] SCAs include a variety of karyotypes. Monosomies and trisomies, including Turner syndrome (45, X), trisomy X (47, XXX), Klinefelter syndrome (47, XXY), and Jacobs syndrome (47, XYY), are the most common. However, live births with tetrasomies such as 48, XXYY and 48, XXXY also occur. Although phenotypically different, SCAs share some neurocognitive, social, physical, endocrine, and infertility symptoms.^{2–6} In general, individuals with tetrasomies experience more severe symptoms than those with trisomies and monosomies.[¶]

Sex chromosome aneuploidies are increasingly identified prenatally through genetic testing; however, most individuals are diagnosed during childhood or adoles-

cence, often as a secondary finding to other exploratory developmental tests.^{1,6,8–10} There is large variability in the management of these conditions after diagnosis; management is dependent on phenotype variability, age at diagnosis, and provider specialty and training on SCAs. Effective management strategies range from physical therapy, occupational therapy, hormone supplements, speech therapy, and/or behavioral health interventions. In addition, some individuals with SCAs benefit from educational and social services such as individualized education program designation and expanded Medicaid coverage.^{1,5,8,9,11,12} There are relatively few interdisciplinary clinics that specialize in SCA management in the United States. Most families experience fragmented care among a variety of specialists, health systems, and agencies; the responsibility of managing and coordinating care is often left to individuals and parents.

In-depth exploration of the parent experience as advocates is currently limited in the literature among parents of children with SCAs. As part of a larger family of studies with individuals and families of children with SCAs, we engaged with parents of individuals with SCAs to understand how they navigate various systems in seeking diagnosis and management services. We found that medical and social service discontinuity led many parents to feel that they were required to adopt a strong advocacy stance for their child. Understanding the kind of services for which parents advocate, the gaps in coverage they experience, and the psychosocial impact

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on children and families is necessary to meet the needs of the families and assure better outcomes for patients. This qualitative study investigates how parents of children and adolescents with SCAs advocate for medical and educational services as well as the impact of ongoing advocacy on parents.

METHODS

This study was reviewed and approved by the Mayo Clinic Institutional Review Board. We partnered with the nonprofit patient advocacy organizations the Association for X and Y Chromosome Variations Foundation and the XXYY Project. Study design was informed by principles of community engagement, including recruitment, development of study materials, and discussion of findings after completion of data analysis for informal feedback. Community partners recruited their members through mailing lists, web postings, and social media. Turner syndrome (45, XO) was not included on the recommendation of the advocacy organizations, who felt that the phenotype was significantly different from other sex chromosome aneuploidies (SCAs). Turner syndrome is more thoroughly characterized in the literature than other SCAs,¹³ and the patient experience literature suggests a diversion in lived experiences.^{14,15} Participants completed Health Insurance Portability and Accountability Act authorization before participation.

Interviews were conducted using a semistructured interview guide (available in supplementary materials of Jaramillo et al. 2019, <http://links.lww.com/JDBP/A275>) featuring questions about their child's diagnostic process, medical care, social and educational services, and relationships with family and health care providers.^{16,17} The interview guide was developed by a multidisciplinary team of bioethicists, clinicians specializing in SCA, and SCA advocates and lightly edited throughout the study to provide clarification for some themes based on participant feedback.

AUS Two researchers (C.J. and J.E.) conducted a total of 32 interviews by phone between October of 2016 and August of 2017. Two interviews were parent-parent dyads, and 4 interviews were parent-child dyads. Analysis of the affected individual interviews was conducted separately. The remaining 26 interviews were individual parents. Interviews lasted an average of 35 minutes (range 21–61 minutes), and recruitment persisted until thematic saturation—defined as the point at which no new themes were encountered in additional interview—was achieved. Interviews were recorded, anonymized, and transcribed verbatim.

Data were qualitatively coded using the software package NVivo Version 11. A team of researchers with expertise in bioethics, qualitative methods, and disability ethics used a grounded theory approach to create an inductive codebook capturing the iterative themes that surfaced in the interviews.¹⁸ Two researchers (C.J. and C.N.) coded 20% of the interviews to consensus to vali-

date the codebook. The remaining transcripts were divided between the same researchers with frequent and recurrent discussion to ensure coding consistency and resolve discrepancies. After the content was inductively coded, parent responses were analyzed for themes related to parent advocacy. Given the research question, responses of individuals with SCAs were excluded for analysis.

RESULTS

Demographics

As shown in Table 1, we interviewed a total of 34 parents who consisted of 29 (85%) mothers and 5 (15%) fathers. Participants in our data set represented individuals or parents of individuals with trisomy X (47, XXX), Klinefelter syndrome (47, XXY), Jacobs syndrome (47, XYY), and the tetrasomies 48, XXYY, 48, XXXY, and 48, XXXX. Participants were geographically dispersed including 13 different states in the United States (n = 26), the United Kingdom (n = 1), Australia (n = 1), and Canada (n = 3). Because there are relatively few clinics that specialize in sex chromosome aneuploidy (SCA), this geographic diversity helps to ensure that multiple families were not reporting experiences with the same health system.

Advocacy During the Diagnostic Odyssey

Parents of children with SCAs who received a postnatal diagnosis frequently recalled that they knew there was something developmentally different about their child and described their efforts to attain a diagnosis in response to their concerns. Several parents stated that they had to continuously bring their child's symptoms to the attention of their medical care providers to initiate the diagnostic process.

Our family doctor didn't really bring anything up, so it was kind of our own research that directed us toward the chromosome issues...I went to our family doctor and requested that we get a Fragile X Karyotype test, and she wasn't really on board with it, but I told her we were gonna have it done either through the health system, or we would pay for it privately just because it is learning delays and speech delays. Everything is so slow and so far behind that we had to look into doing something. (Father of 8-year-old, 48, XXYY; Pediatric Diagnosis: 7 years old)

However, when families did bring their concerns to their physicians, they often felt that providers were dismissive. Despite parental insistence of symptom severity and persistence, parents reported that providers frequently suggested that developmental delays and other symptoms were something the child would grow out of.

Probably around the 6-month mark is when we noticed, "Gee, he's certainly not making those milestones. He's not rolling over." He wouldn't lift his head up. When you tried to lift him he wouldn't assist with lifting his head...

Table 1. Participant Demographics

Parent participants	
Total participants	N = 34
Total interviews	N = 32
Mothers	N = 29 (85%)
Fathers	N = 5 (15%)
Children with sex chromosome aneuploidy demographics	
XXX (trisomy X) diagnosis	N = 11 (34%)
XXY (Klinefelter syndrome) diagnosis	N = 7 (22%)
YY (Jacobs syndrome) diagnosis	N = 7 (22%)
XXYY diagnosis	N = 6 (19%)
XXX/XXXX mosaicism	N = 1 (3%)
Mean age of the child at the time of interview (range)	9.5 yr (2 mo–24 yr)
Mean age of the child at the time of diagnosis (range)	8.3 yr (6 mo–17 yr)
Participants with prenatal diagnosis	12 (32%)
Participants with pediatric diagnosis	25 (68%)
Geographic information ^a	
United States: states include CA, FL, ID, KY, MA, MD, MI, MN, NE, NJ, NY, OR, and SD	N = 26
Canada	N = 3
United Kingdom	N = 1
Australia	N = 1

^aLocation data are missing from 3 participants.

We had talked to the pediatrician about it. He brushed it off and was like, “No, it should be fine. Wait until 9 months.” (Mother of 9-year-old, 48, XXYY; Pediatric Diagnosis: 5 years old)

Parents also expressed frustration over the lengthy process of testing and the multiple medical appointments with different physicians that their child endured. Many families reported repeated testing with different specialists, including therapists, orthopedists, neurologists, urologists, and geneticists.

I knew, since she was very little, that something was different. We had her in speech therapy for years. She has low muscle tone. I had taken her to see a neurologist and a developmental pediatrician and fought for a year to get speech therapy. They never—no one ever put it together. When she was, I think, 3 or 4, and we saw the neurologist, the neurologist had offered genetic testing but didn't think that we would necessarily find anything, because she had delays in only 2 areas at the time. Said, “I don't think it would change treatment.” Then [child] started having this behavior at school that I thought it

was more of a medical thing where she just would stay so sick all day... I had—we did a blood panel. She was a little low on iron, so didn't really have—know what was happening... Then, when [neurologist] found out that [child] was also having receptive language issues and all these physical complaints, she re-brought up the idea of the genetic testing. That's how we ended up getting the diagnosis, because she went back. (Mother of 7-year-old, 47, XXX; Pediatric Diagnosis: 6 years old)

Parents as Medical “Experts”

Even after receiving a diagnosis, parents reported that advocacy continued to be a large factor in navigating the health care space. Parents often report being “the experts in the room” at medical appointments, a role that many parents reluctantly accepted.

I have learned that in any given room, I am usually the trisomy X expert wherever I am. Most parents of kids with X chromosome aneuploidies, they very often are, sad but true. (Mother of 10-year-old, 47, XXX; Pediatric Diagnosis: 18 weeks)

Many parents felt that their role in medical and social service appointments was not only as the parent/caregiver of the child but also as a necessary educator of providers. They expressed concerns that providers would be unable to adequately care for their child if parents could not explain their child's condition in detail.

That's why I walked into this adult endocrinologist appointment, and I was giving him information. I was trying to be very respectful, but I have so many questions that he couldn't answer. I just think it kind of opened my eyes to the fact that—and he told me. He goes, “You are giving me more information than I know.” I think that's sort of reality, right? (Mother of 18-year-old, 47, XXY; Pediatric Diagnosis: 17 years old)

Although some parents found the need to constantly educate others about their child's condition taxing, others saw an opportunity to educate providers as an extension of their advocacy. They conceptualized it as a way to help both their child and the SCA community.

Every time I walk in, I say, “Hey, this is what my child has.” They all go, “Huh! I don't know what that is.” It's not very reassuring. Every time there's a med student or a resident, or whatever it may be, and they say, “Can they come in?” I say, “Yes. You may. Because you need to know what this is. This is my child.” He's probably the only one in [state] right now that they're aware of, that has it. I want everybody else to be aware in case their child ends up with it. (Mother of 8-year-old, 48, XXYY; Pediatric Diagnosis: 5 years old)

Advocating for Services

Beyond medical care, parents recounted the need to advocate for education and occupational services for

their child to address learning and motor delays. Families often felt that they had to battle school districts and social institutions to attain necessary services for their child.

In New York, they reevaluate every 6 months, so in New York, [child] kept getting kicked out 'cuz she'd catch up and then they'd kick her out, and then she'd fall behind again and I'd have to get her restarted. So, I finally got a letter from [child's doctor] that said, "Stop doing this to this poor child. You haven't cured her trisomy X. She's still gonna' have trouble. Just leave her in services," and then they did. (Mother of 10-year-old, 47, XXX; Pediatric Diagnosis: 18 weeks)

Parents advocated and received services predominantly within the school system. Parents made efforts to establish relationships with their child's educators and met with them frequently to ensure that their child's educational needs were met.

Hours per week, I mean, it's minimal because, where we are right now, he's been at the same school since August. It's more along the lines, when something changes. When he goes into a new school year, then, we have to sit down and talk to the teacher. Make sure his team is aware what's going on. They all have the updated information. That's where it takes more time than it does when he's been at the same place for 6 months. (Mother of 5-year-old, 48, XXYY; Pediatric Diagnosis: 3 years old)

Parents also described the ongoing need to coordinate multiple service providers, such as schools, governmental assistance programs, and therapeutic services such as cognitive behavioral therapy and speech and learning. This required parents to work with multiple entities to ensure that their child was accessing necessary services.

Since he's so little, they're allowing me to bring the ABA therapist in. Of course, they have their policies where they have to do all their checkups, before they allow anybody in the school because we have a therapist come in to shadow him and to helping kids with the school. Yeah. They've been very helpful. They learn from the therapist too. (Mother of 3-year-old, 47, XYY; Prenatal Diagnosis)

Parental Burden

Parents described the unique emotional toll of being the default expert and advocate for their child, including varying degrees of stress and frustration. Parents report that these feelings are the result of repeatedly explaining their child's diagnosis to each medical, educational, and social professional and the constant need to fight for services or understanding.

Oh my gosh. It's so challenging. Sometimes, I just get tired and I cry. It really helps me to cry because I just let

it out. Sometimes, as stressful as I am, it's a lot to keep up with every day. It's a like a new thing. We're just living day-by-day with my husband. It's very hard. (Mother of 3-year-old, 47, XYY; Prenatal Diagnosis)

Many parents expressed feeling overwhelmed by the complexity of raising a child with an SCA, especially if they were a single parent or performed the bulk of childcare. They reported high cognitive burden from the large amounts of energy required for coordinating services and care for their child. Notably, the primary caretakers were often the mother.

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Well, as a mom and being divorced, and not having an ex-husband who was supportive in the whole thing, it was on my shoulders. I just wanted what was best for my son. As a parent that's all you want. Right? (Mother of 18-year-old, 47, XYY; Pediatric Diagnosis: at birth)

While this caretaking/advocate role weighed on parents, they also reported finding a sense of community and support from other parents in the SCA community. Access to this community was often facilitated through either online support groups or conferences. This had a practical benefit of parents being able to learn what has worked well for others in a similar position.

What was helpful was finding the website, the conference, and we ended up meeting a group of people who were extremely supportive. It was just a fantastic group of people. Also, they had a lot of talks by doctors and researchers, who were dealing with trisomy X. I think that was the most comforting and educational experience, for us. (Mother of 10-month-old, 47, XXX; Prenatal Diagnosis)

Parents often felt a deep relational and empathetic connection with other parents of children with SCAs. This relational aspect of social support frequently served as a sense of strength and belonging for parents who otherwise felt isolated by their child's complex situation.

DISCUSSION

Parents in our cohort overwhelmingly expressed that they actively fought for resources and services for their child. Without this effort, they felt, their child would not have access to these resources at all. The parents-as-advocates model is a familiar one to the developmental and intellectual disabilities and rare diseases communities.^{18,19} Parents of children with rare diseases report feeling the need to fill the roles of parent, primary care provider, medical expert, and care coordinator. Particularly for children with rarer conditions, this can require breaking new ground with providers and service custodians, leading high levels of stress and feelings of burn-out.¹⁹ The creation and maintenance of the parent-as-advocate role is unlikely to be a result of actions or behaviors of any set of individuals in the medical and social services fields but a result of the fragmentation of the US

health and social services systems, with its significant variation between payors, state agencies, and public services. Similar to participants in this study, parents of children with many forms of medical or intellectual conditions report difficulty in accessing and using services, being denied insurance coverage for required services, and repeated inability to identify a central source of care.²⁰

Participants often reported filling the role of medical expert when interacting with medical providers. This was true across sex chromosome aneuploidies (SCAs) but was especially pronounced in the rarer tetrasomies. Shifting the role of health and medical knowledge to the parent is potentially problematic for several reasons. First, it places additional burden on parents, who often feel that they are responsible for constantly researching, maintaining medical records, and chronicalling the specifics of their child's phenotype. Second, there are practical medical concerns in relying on parents to explain their child's medical condition and treatment to providers. Most participants, despite their extensive and dedicated knowledge gathering, had a layman's understanding of medical terminology and ability to access and reinterpret medical texts and the peer-reviewed literature. Third, parents are not impartial observers of their child's medical, social, and intellectual condition. Clinical and behavioral information presented to providers is inevitably filtered through parents' own experiences and understanding of the "default state" from which their child may be differing. Parents may ascribe traits to an SCA that are more likely the result of normal childhood experiences or of a secondary diagnosis; research has shown that parents of children with a genetic test result, even an uncertain one, tend to overprescribe normal variations in childhood metrics to the genetic condition in question.²¹ When providers rely on parents to supply expert knowledge, they not only place additional burden on the parent but potentially accept an uncontested clinical understanding of the child.

There is also a known relationship between successful advocacy and socioeconomic status. Many of the families interviewed reported that the additional care and services they required meant paying for services out of their own pocket or not working to care for the child. This financial burden is similar to the phenomena reported by parents of children with disabilities generally^{22,23}; being unable to afford care due to financial or coverage restrictions can serve as a major barrier to a child receiving the resources they need to flourish. Furthermore, parental factors such as educational attainment and income level affect health and care coordination behaviors for the entire family. In other instances of children with disability, parental income and education level negatively correlated with feeling empowered to seek treatment and viewing their child's disability as a mystery.²⁴ Thus, not only does the socioeconomic situation of the family predispose them to face financial barriers to optimum services and interventions but may disempower them

from seeking such services in the first place. In particular, negotiating the complex system of social service applications, Medicaid requirements, and school system bureaucracy requires time, language competence, and the ability to be present and interact with individuals of multiple genders—a requirement that places undue burden particularly on women from certain religious backgrounds. A fragmented system that nonetheless requires parents to step into a role as advocate and expert to ensure their child receives necessary care and services results in children from socioeconomically disadvantaged families being less likely to receive the interventions they need to develop successfully.

Another important disparity is the effect of continuous advocacy on the parents themselves. Many participants related explicit frustration with individuals or institutions they saw as working against them. Some parents linked this frustration to feelings of burden and burnout, and most stated that they felt this way at some point during the process of raising their child with an SCA. In situations in which additional pressures from lack of income, immigrant status, or inconsistent contact with the medical system provide secondary pressures, this psychological impact may be compounded. Nevertheless, participants generally connected an advocacy role as a necessary and indistinguishable aspect of caring for their child. Several parents reported feelings of satisfaction at hard-won progress they made in accessing services or recognition and the beneficial impact on their child's development.

Interestingly, a significant majority of parents in our cohort who self-described as the primary care provider and advocate identified as women. This is reflective of larger social and cultural narratives of femininity and motherhood²⁵ and suggests that, in many families, the additional burden placed on families by the need to advocate for the child with an SCA is often born by mothers. Previous research has shown that mothers of children with a disability often experience significantly higher amounts of financial stress and mental illness related to the burdens of parenting a child with such complex needs.²⁶ Given that most of the participating parents were women, including a subset who reported as single mothers, gender is an important consideration when understanding these results. The women who identified as single mothers also reported the highest amounts of burden, with additional struggles in being able to transport their child to all necessary appointments and services alone.

One of the primary sources of relief from these additional burdens was through socializing and sharing with other parents. These connections were most often formed through SCA advocacy organizations, such as Association for X and Y Chromosome Variations, either through online forums or at national conferences. Parents frequently reported that they felt this community of individuals with shared experiences was able to uniquely support them emotionally and practically. This is similar

to work exploring the social experience of parent advocates more generally in that the parents in our study also reported discussing challenges such as isolation, stigma, logistics, and parenting.²⁷ This avenue of support and resiliency appears essential for parents in maintaining their personal health and continuing to advocate for their child.

A known limitation of qualitative methods using a finite cohort is that the findings cannot be generalized to all individuals and families with SCAs. In addition, verbal phone interviews did not allow for the analysis of non-verbal communication. Although parents in different geographic areas described similar experiences, experiences of advocacy may be different depending on insurance status and variations in health systems. Socioeconomic data were also not collected other than what was directly discussed by participants. Future research directions should include a better understanding of the effectiveness of advocacy methods at achieving care and service goals, socioeconomic stratification in access to services and support, and whether there may be systemic and practice interventions that could shift burden off of parents.

CONCLUSIONS

Parents feel a strong imperative to advocate for their child with a sex chromosome aneuploidy (SCA) in the medical and health services systems. Because parents of children with disabilities and complex medical needs play such a crucial role in determining which services their child receives, understanding parental advocacy and its consequences is critical to understanding how SCAs are actually managed.^{18,19,27} In many instances, this advocacy is a problematic indictment of the fragmented nature of the US health care system, requiring parents, and most often mothers, to assume the challenging and burdensome role of care coordinator. This also suggests that families in which parents do not have the resources to advocate may be receiving less robust care and services. Disparities in access and outcomes may be improved by clinicians and other care providers through better care coordination and understanding of SCAs across medical, educational, and social services settings in addition to the benefit of relieving parents of the burdens associated with the advocacy of a child with an SCA.

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