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"There are hills and valleys": Experiences of parenting a son with X-linked retinoschisis

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Introduction

X-linked retinoschisis (XLRS) is an inherited retinal disorder that typically presents as reduced visual acuity in boys. Although a broad spectrum of clinical phenotypes has been reported across individuals with XLRS, longitudinal studies have demonstrated that, in the absence of significant complications, most individuals with XLRS have stable vision over several years' time.[1-4]

Receiving news of a medical diagnosis in a child often poses significant challenges for families. Parents must cope with a profound sense of loss, helplessness, unfairness, isolation, fear, and uncertainty.[5-9] Although some families struggle to adapt to life with a chronic medical condition, prior research across a range of conditions has found effective coping, benefit-finding, successful adaptation, and resiliency in most families.[5,10-12] Healthcare

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models that address the psychosocial needs of the entire family have been associated with improved physical and emotional outcomes for children with chronic medical conditions.[12-14]

The purpose of this study was to explore the experiences of parents of sons with XLRS. Providers benefit from understanding the impact of XLRS on families to guide how to effectively address their medical, psychological and social needs.

Methods

Parents of sons with XLRS who were evaluated at the National Eye Institute (NEI), National Institutes of Health (NIH) (www.clinicaltrials.gov: NCT00055029), between December 2017 and January 2019 were invited to participate in this mixed methods study on the day of their son's appointment. Parents of sons who were previously evaluated at the NEI and who contacted the study investigator (AT) during this time period were also invited to participate. Parents were eligible if they had a biological son of any age with XLRS who was enrolled in NCT00055029. Recruitment occurred until the investigators determined that no new concepts were emerging from the interviews, and data saturation was reached. This study was approved by the NIH Combined Neuroscience Institutional Review Board and conducted in accordance with the Declaration of Helsinki. Informed consent was provided by all participants.

Two investigators (CDA and RN) who had no prior interactions with the study participants or their sons conducted semi-structured interviews either in person at the NIH or by phone. The interview guide was informed by prior studies in the literature investigating the experiences of parenting a child with a disability[7,10] and the impact of living as a carrier of a genetic condition.[15,16] Interviews explored the experience of XLRS in the family, and the perceived impact of XLRS on participants' sons and their parenting experiences. Interviews were audio-recorded and transcribed verbatim.

We included an anonymous self-administered survey to assess character traits that may influence parents' experiences in response to the diagnosis of XLRS, based on prior studies investigating predictors of effective coping and adaptation.[5,17] Optimism was assessed using the Revised Life Orientation Test (LOT-R),[18] a 10-item measure that includes 6 scored items and 4 filler items. Respondents were asked to indicate the extent to which they agreed with statements such as "In uncertain times, I usually expect the best" on a scale from 0 (strongly disagree) to 4 (strongly agree). A total score was obtained by omitting the filler items, reverse scoring items with a negative valence, and summing the scores. Higher scores indicate a generally hopeful orientation towards life. Cronbach alpha was 0.95.

Anxiety was measured using the trait form of the State-Trait Anxiety Inventory (STAI).[19] Trait anxiety refers to a more general, long-standing quality, as opposed to a temporary condition or state. Participants rated 20 statements on a scale from 1 (almost never) to 4 (almost always). Scores were summed, and higher scores indicate greater anxiety. Cronbach alpha was 0.93.

Personality traits were measured using the Big Five Inventory (BFI),[20] a 44-item measure of the five dimensions of personality: extraversion, agreeableness, conscientiousness, neuroticism, and openness to experience. Participants rated each item on a scale from 1 (disagree strongly) to 5 (agree strongly). Overall scores were calculated by reverse-scoring certain items and then averaging the items representing each dimension. Cronbach alpha for each dimension ranged from 0.73-0.90.

Participants also reported demographic information, including their age, their son's age, race/ethnicity, and level of formal education.

Data Analysis

Transcripts of interviews were checked for accuracy and imported into QSR International's NVivo 11 qualitative analysis software. A preliminary codebook was developed based on a priori topics, and two investigators (AT and RN) applied these codes to one mother and one father interview, adding to the codebook as additional topics emerged. Transcripts were coded by one investigator (RN) concurrently with data collection, and the codebook was further expanded as needed. All transcripts were recoded using the final codebook. A second coder (AT) coded half of the transcripts, and minor discrepancies in coding were discussed between investigators until agreement was reached. Thematic analysis was used to interpret coded findings. Detailed notes taken throughout the process and coded data were reviewed repeatedly to identify potential themes.[21]

Quantitative data were analyzed using IBM's Statistical Package for Social Sciences (SPSS) Version 25.

Results

This study included 19 parents (11 mothers and 8 fathers) from 13 families. In 6 families, both parents participated, whereas in 7 families only one parent was interviewed. A total of 14 mothers and 9 fathers were invited to participate, for an overall response rate of 83%. Logistical barriers to participation was the reason some parents were unable to participate. Interviews lasted an average of 59 (range 34-86) minutes for mothers and 45 (range 30-63) minutes for fathers.

Sixteen participants (9 mothers and 7 fathers) were white and non-Hispanic, and 3 participants (2 mothers and 1 father) were Hispanic. The mean age of the mothers was 49.1 (range 41-56) years, and the mean age of the fathers was 46.25 (range 41-54) years. All but 3 parents had college or post-graduate degrees. Most families had one son with XLRS, but in 4 families there were two sons with XLRS. The mean age of the sons with XLRS was 16.1 (range 8-25) years.

All but two mother participants completed the survey. The means, standard deviations, ranges, and internal reliability values for each scale are presented in Table 1. Participants reported high levels of dispositional optimism and low levels of trait anxiety. Participants rated themselves highest on the agreeableness, conscientiousness, and openness scales, and lowest on the neuroticism scale. The mean scores for all of the scales were consistent with

published normative data,[22-24] suggesting that there is nothing atypical about our study population that might influence their interview responses.

Experience of Diagnosis

Parents described the experience of learning their son's diagnosis as shocking, devastating, overwhelming, sad, hopeless, isolating, and scary. "It's very hard to explain. It's almost like they tell you…it feels like your kid's life is over. That's the only way to explain it" (017 [participant number], Mother, 9 [age of son(s)]). Although 7/13 families had a family history of vision loss, none of the families knew that their relatives had XLRS, or that it was hereditary, until their son's diagnosis. As such, learning their son's diagnosis was as unexpected as in families with no family history of vision loss.

Although in all families the diagnosis had occurred years earlier, many parents were emotional when recounting their experience and indicated that they remembered that day like it was yesterday. As one mother said, "I still remember [that] Friday morning at 9AM… [My son] is now almost 17, and he was a baby. I still remember sitting in that doctor's office, and I remember everything about that appointment. Life was never, ever the same after that" (016, 16&18).

Twelve parents described less than desirable health care interactions with providers who lacked familiarity with XLRS. Many parents felt that they did not receive quality information, a balanced perspective, or adequate support. Several parents recalled being told that their sons would go blind, would never be able to drive a car, would struggle in school, and were given contact information for the school for the blind. "[Being told] there was no cure and no treatment was devastating. [The doctor] was not really able to explain to us what to do or to refer us to someone who knew about it. That was hard. I ended up taking like two days off of work…I just researched and researched until I found doctors who had experience with XLRS…and until I could figure out a path forward" (003, Father, 14).

When asked what would have been most helpful at the time of diagnosis, parents indicated that they wanted quality information and resources, to be connected with other families, guidance as to what to expect and prepare for, and hope. "I know that for me knowing that there was hope would have made all the difference. It would have made all the difference in the world. I would have had something to hold onto. And we had nothing" (016, Mother, 16&18).

Maternal Guilt

Seven of the mothers reported feeling guilty about the X-linked inheritance of the condition. In all but 2/7 mothers, their guilt lessened with time. "There was this instant guilt. Like, oh my god, I didn't know…I gave this to him and I'm the reason that this kid isn't going to be able to do what he loves in the future" (010, Mother, 8).

One mother with persistent guilt felt like she was blamed by her husband. However, none of the other mothers felt blamed by their partners or other people. Many of the fathers struggled to understand their wives' feelings of guilt. "It was really interesting. [My wife] had this sense of completely illogical and irrational guilt. Really weird…She just said she felt guilty

about it, which is kind of silly. It's like feeling guilty for being blonde. But she just felt bad about it. I just, I never quite could get it…Anyway, maybe it's a mother thing" (006, Father, 17).

Life Post-Diagnosis

Following the initial crisis of their son's diagnosis, parents described a process of accepting their son's condition, coping with feelings of helplessness and an inability to protect their son from hardship, and devising a "new normal." "You persevere. You go on. There are hills and valleys" (007, Mother, 14). Parents often normalized their experience by reminding themselves that "everyone has something," and tried to stay positive and help their sons feel like they can do anything despite their diagnosis. As one mother said, "[XLRS] doesn't define them. It's a small part of who they are" (010, 8).

Most parents reported that life with XLRS was not as bad as they feared at the time of diagnosis. "Parents who are just learning about their son's diagnosis are all scared to death that their son will be blind by the time he is 8…but then your child gets older and you reflect on the fact that this hasn't really slowed them down. They're successful. Maybe they can't drive, but no one says that this condition is the most horrible thing that could have happened to us. Your kid learns to deal with it, and you do too" (012, Mother, 13&23).

Initially, almost all parents were concerned about vision loss or blindness, but parents' concerns often shifted as they learned more about the natural history of XLRS and/or their son demonstrated stable vision over several years. Parents expressed concerns about helping their sons cope with disappointment, fostering self-esteem, and preventing XLRS from being an excuse to help their sons reach their full potential. All parents said that the hardest part of XLRS was the unknown. "You never want to see your kids hurt or disappointed, even though that's part of life. Whether it's because of [XLRS] or something else, that's a normal part of life. And so [the hardest part] is not knowing what the future holds and what emotions and fears he may have as time goes on" (010, Mother, 8yo). "I think as a parent my greatest concern is helping him understand that even with this disability there are so many great things he can do…he doesn't understand that right now" (003, Father, 14).

Although almost all parents recognized that their sons were doing well with XLRS, several parents indicated that they thought about their son's condition every day, and many described a state of constant worry. The fear of a retinal detachment was an ongoing concern for almost all parents.

Balancing Act—Parents described the challenge of balancing their fear of a retinal detachment and their desire to protect their son's vision with wanting to promote normalcy, allowing their sons to participate in activities they enjoy, and giving their sons the freedom they deserve to experience life. "They are both active boys…It's a constant worry that they could fall off a horse or get hit in the head with a baseball…and that could cause permanent damage to [their] eyes. But then at the same time, neither one of them wants to give up what they're doing" (014, Father, 14&18).

Ambiguities about what activities are considered safe for individuals with XLRS often added to parents' stress and could be a source of conflict among family members. Several parents, especially mothers, felt as though they were overprotective or the "bad cop" parent because they restricted their son's activities.

Sports—Eighteen parents discussed sports without being prompted. Sports were viewed as positive activities that offered valuable life lessons, making the decision to limit sports participation especially difficult for parents. "It's very difficult to say no to something that you believe is good for him" (014, Father, 14&18).

Parents described varying levels of restrictions and had different perspectives on which sports were acceptable for people with XLRS. As a father of a 13-year-old son stated, "You weigh all the possible consequences and then make the best decision you can" (013). Many parents felt as though there were consequences regardless, whether it was the risk of a retinal detachment or the risk of making their son feel overprotected and further limited by his condition. "The doctors didn't think [water polo] was safe, but they have to do something. They can't just…They can't play baseball, they can't play soccer, football. [I] had to give in a little bit and just pray that practice goes well." (008, Mother, 14&18).

Parents perceived social implications to restricting sports both in terms of peer relationships and father-son bonding. One mother reflected on the impact this has had on her husband, stating "[Sports] is the one thing that's the closest tie for them. The most passionate item in the world. And as a daddy and a boy, there's that pride. I think that's really challenging [for my husband]" (010, 8). Related to this were perceived societal expectations and gender norms. As one father stated, "The ideal boy as portrayed to us as parents is not the norm" (002, 12).

A minority of parents did not report a significant impact of sports restrictions on their son, either because they guided their son towards sports that they considered acceptable or because they guided their son to different activities altogether. "I don't have the kid that plays baseball, football, the sports kid. I don't. He's not my child. It doesn't affect me, [although] it could make me a little bit sad sometimes as far as him not getting that experience." (006, Father, 17)

Perceived Impact on Son—Parents' perceptions of how XLRS impacts their sons varied from no impact at all to impacting every aspect of their son's life. Most parents thought that the hardest part for their sons was feeling different during a life stage when fitting in and belonging to a desired peer group was critical. "It's embarrassing I think for them…to have to acknowledge that they can't see something…at their age, when it's important to fit in and be like everyone else, having to highlight themselves in that manner is difficult" (016, Mother, 16&18).The desire to fit in often resulted in a reluctance to have accommodations in school, which was a source of stress for parents.

Fifteen parents brought up driving without being prompted. Driving was viewed as a developmental milestone and a rite-of-passage, especially for young men. Eight of the nine sons who were of driving age were able to get either a restricted or unrestricted driver's

license. “When they’re 16, that’s what you do. Everybody goes and gets their driver’s license. I want him to be able to be like everyone else” (015, Mother, 14&18). “Not being able to drive at night is a big deal for boys…because of dating and jobs and just being a teenager where you go and hang out with friends.” (012, Mother, 13&23).

Several parents felt like the experience of trying to get a driver’s license was the first time their sons considered themselves to be limited because of their XLRS. “Trying to get a license was what brought it to a head [for my son] that ‘oh my gosh, I really do have something wrong with me’” (019, Mother, 25). “I would say that [trying to get a driver’s license] was probably the first time ever in my children’s life that they felt disabled” (016, Mother, 16&18).

Furthermore, whether or not their son was able to get a license framed the experience and perceived severity of XLRS. “He was borderline on being able to get a driver’s license when he turned 16. And his being able to get one was a big deal…Had he not gotten a driver’s license, our experience would be a lot different” (006, Father, 17).

Benefit-Finding and Positive Outlook

Parents identified numerous lessons learned and ways in which they believed they had grown from their experience, including increased gratitude, renewed life values, increased empathy, family cohesion, and strengthened faith. Many moms felt proud of their advocacy, and many parents felt good about their participation in vision research. “I take every day as a blessing when my kids can see, and they can go on with their normal life. To me, that’s a blessing. I don’t care how bad of a work day it was, or traffic, money [problems]. My kids have vision” (008, Mother, 16&18). “Some things cut you, and they’re deep, but then you gain from it. I’ve tried to be a better mother because of this [experience]” (019, Mother, 25).

Fifteen of the parents were hopeful about a potential treatment or technological advances, like self-driving cars. “I really believe that one day there will be a cure for this” (007, Mother, 14).

Discussion

This study contributes to the understanding of the impact of parenting a son with XLRS and identifies opportunities to enhance the care provided to families with XLRS.

The diagnosis of XLRS was a transformative event for many parents. What providers communicated at the time of diagnosis had wide-ranging effects on parents and framed the way that they began to understand XLRS and its potential impact.[25] Consistent with prior studies of individuals with genetic conditions,[26-28] many parents expressed dissatisfaction with the way the diagnosis was delivered and felt as though they were left to navigate the implications of XLRS and to cope with feelings of helplessness and loss of control with little support or guidance from their providers. Ophthalmologists, nurses, and genetic counselors are well-positioned to help parents anticipate practical and emotional demands associated with XLRS so that families may prepare for potential challenges. Although there is no current treatment for XLRS, families should be encouraged to identify aspects of XLRS

that are within their control, such as participation in XLRS research studies, advocating for school accommodations, and directing their son to hobbies and interests that will not be impacted by reduced vision. Referrals to genetic counselors and ophthalmic genetics clinics experienced in caring for individuals with XLRS might also help to address the needs of patients with XLRS and their families.

This study adds to the findings of several prior studies that have reported that guilt and self-blame are prominent among mothers of children with X-linked conditions.[16,29,30] Providers might help families anticipate this reaction, which both normalizes the feeling and provides an opportunity for parents to discuss their reactions. Connecting parents with other families with XLRS may also help mitigate and normalize feelings of guilt. Two of the mothers in this study had persistent guilt, which has been associated with depression, anxiety, and poorer adjustment.[31] Mental health referrals should be provided if clinical depression or anxiety is suspected.

The possibility of a retinal detachment, though an infrequent occurrence among individuals with XLRS,[2] added an element of unpredictability to a condition that parents otherwise perceived to be relatively stable. Decisions to limit sports participation were especially difficult for parents since it seemed counter to the natural tendencies of many sons, and participation in sports was perceived as an important part of childhood. Parents struggled with ambiguities over what activities were considered safe and described a process of constantly considering what was necessary to protect their son from potential vision loss versus what was necessary to promote normalcy, often compromising one for the benefit of the other. Prior studies of children with congenital heart conditions have also reported that the inability to play sports is experienced as a loss, highly distressing, and socially isolating.[32] While there are no clear guidelines on sports participation for individuals with XLRS, parents may benefit from their providers playing a more active role in helping them make decisions about activity restrictions.

Consistent with previous studies across a range of conditions,[33-36] this study found that the challenges of adolescence and the issue of driving were significant stressors for families. The need for adolescents to feel accepted by their peers, to establish independence, and to dream about the future can be complicated by XLRS and associated feelings of unpredictability or uncertainty. Parents play a critical role in helping their children cope effectively[33,37] and helping them recognize who they are as people despite a medical diagnosis.[38] Further research with adolescents with XLRS would help to illuminate ways in which parents and health care providers can offer support during this developmentally challenging time.

This study adds to a growing body of research on adaptation[5] and resilience[39] showing that a majority of families are able to successfully manage and overcome significant life challenges, to identify positive changes that result from the experience, and to become stronger and more resourceful in the process. Prior research has found that the ability to find benefit in stressful life experiences is associated with positive physical and psychological outcomes.[40] Clinicians can encourage parents to reflect on their personal growth and resilience to further promote self-efficacy.

This study has several limitations. The study population was largely white, non-Hispanic, and highly educated. All parents had sons who were participating in an observational research study at the NIH. Those families that are resourceful in finding a research study and enrolling their son may have more personal resources to cope and adapt over time. Thus, the results of this study are not generalizable to all parents of sons with XLRS.

Efforts to improve the experiences of families with XLRS are implied by our findings. Parents seek a useful and compassionate diagnosis disclosure. Good communication of quality information is critical so that parents feel equipped with the knowledge and confidence they need to manage life with XLRS. Anticipatory guidance that is in line with what is known about the natural history of XLRS is highly valued. Parents can be helped to inventory their coping resources to assess whether there are more positive or effective strategies they could be using. Further, parents desire access to other families with XLRS for both practical guidance and emotional support. Referrals to genetic counselors and ophthalmic genetics clinics should be considered. Additional research with a more diverse population is needed to replicate or refute this study's findings.

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References

1. Roesch MT, Ewing CC, Gibson AE, Weber BH. The natural history of X-linked retinoschisis. Can J Ophthalmol 1998;33(3):149–158. [PubMed: 9606571]
2. Cukras CA, Huryn LA, Jeffrey BG, Turriff A, Sieving PA. Analysis of anatomic and functional measures in X-linked retinoschisis. Invest Ophthalmol Vis Sci 2018;59(7):2841–2847. [PubMed: 30025115]
3. Kjellstrom S, Vijayasarathy C, Ponjavic V, Sieving PA, Andreasson S. Long-term 12 year follow-up of X-linked congenital retinoschisis. Ophthalmic Genet 2010;31(3):114–125. [PubMed: 20569020]
4. Wood EH, Lertjirachai I, Ghiam BK, et al. The natural history of congenital X-linked retinoschisis and conversion between phenotypes over time. Ophthalmol Retina 2019;3(1):77–82. [PubMed: 30935660]
5. Biesecker BB, Erby L. Adaptation to living with a genetic condition or risk: a mini-review. Clin Genet 2008;74(5):401–407. [PubMed: 18823383]
6. Hodgkinson R, Lester H. Stresses and coping strategies of mothers living with a child with cystic fibrosis: implications for nursing professionals. J Adv Nurs 2002;39(4):377–383. [PubMed: 12139650]
7. Whittingham K, Wee D, Sanders MR, Boyd R. Sorrow, coping and resiliency: parents of children with cerebral palsy share their experiences. Disabil Rehabil 2013;35(17):1447–1452. [PubMed: 23167453]
8. Baker BL, McIntyre LL, Blacher J, Crnic K, Edelbrock C, Low C. Pre-school children with and without developmental delay: behaviour problems and parenting stress over time. J Intellect Disabil Res 2003;47(4-5):217–230. [PubMed: 12787154]

9. Baumbusch J, Mayer S, Sloan-Yip I. Alone in a crowd? Parents of children with rare diseases' experiences of navigating the healthcare system. J Genet Couns 2019;28(1):80–90.
10. Lee Y-J, Park HJ, Recchia SL. Embracing each other and growing together: redefining the meaning of caregiving a child with disabilities. J Child Fam Stud 2015;24(12):3662–3675.
11. Green SE. "We're tired, not sad": Benefits and burdens of mothering a child with a disability. Soc Sci Med 2007;64(1):150–163. [PubMed: 17014946]
12. Weihs K, Fisher L, Baird M. Families, health, and behavior: A section of the commissioned report by the Committee on Health and Behavior: Research, Practice, and Policy Division of Neuroscience and Behavioral Health and Division of Health Promotion and Disease Prevention Institute of Medicine, National Academy of Sciences. Families, Systems, & Health. 2002;20(1):7–46.
13. Emerson L-M, Bögels S. A systemic approach to pediatric chronic health conditions: why we need to address parental stress. J Child Fam Stud 2017;26(9):2347–2348.
14. Drotar D Relating parent and family functioning to the psychological adjustment of children with chronic health conditions: What have we learned? What do we need to know?. J Pediatr Psychol 1997;22(2):149–165. [PubMed: 9114640]
15. Lewis C, Skirton H, Jones R. Can we make assumptions about the psychosocial impact of living as a carrier, based on studies assessing the effects of carrier testing? J Genet Couns 2011;20(1):80–97. [PubMed: 20878544]
16. James CA, Hadley DW, Holtzman NA, Winkelstein JA. How does the mode of inheritance of a genetic condition influence families? A study of guilt, blame, stigma, and understanding of inheritance and reproductive risks in families with X-linked and autosomal recessive diseases. Genet Med 2006;8(4):234–242. [PubMed: 16617244]
17. Barskova T, Oesterreich R. Post-traumatic growth in people living with a serious medical condition and its relations to physical and mental health: A systematic review. Disabil Rehabil 2009;31(21):1709–1733. [PubMed: 19350430]
18. Scheier MF, Carver CS, Bridges MW. Distinguishing optimism from neuroticism (and trait anxiety, self-mastery, and self-esteem): A reevaluation of the Life Orientation Test. J Pers Soc Psychol 1994;67(6):1063–1078. [PubMed: 7815302]
19. Spielberger CD, Gorsuch RL, Lushene R, Vagg PR, Jacobs GA. Manual for the State-Trait Anxiety Inventory. Palo Alto: Consulting Psychologists Press, 1983.
20. John OP, Srivastava S. The Big-Five trait taxonomy: History, measurement, and theoretical perspectives. In: John OP, ed. Handbook of personality: Theory and research. Vol 2. New York: Guilford Press, 1999:102–138.
21. Braun V, Clarke V. Using thematic analysis in psychology. Qual Res Psychol 2006;3(2):77–101.
22. Glaesmer H, Rief W, Martin A, et al. Psychometric properties and population-based norms of the Life Orientation Test Revised (LOT-R). Br J Health Psychol. 2012;17(2):432–445. [PubMed: 22106985]
23. Kendall PC, Sheldrick RC. Normative data for normative comparisons. J Consult Clin Psychol 2000;68(5):767–773. [PubMed: 11068962]
24. Srivastava S, John OP, Gosling SD, Potter J. Development of personality in early and middle adulthood: Set like plaster or persistent change? J Pers Soc Psychol 2003;84(5):1041–1053. [PubMed: 12757147]
25. Glare P, Fridman I, Ashton-James CE. Choose your words wisely: the impact of message framing on patients' responses to treatment advice. Int Rev Neurobiol 2018;139:159–190. [PubMed: 30146046]
26. Sutton EJ, Young J, McInerney-Leo A, Bondy CA, Gollust SE, Biesecker BB. Truth-telling and Turner syndrome: the importance of diagnostic disclosure. J Pediatr 2006;148(1):102–107. [PubMed: 16423607]
27. Turriff A, Macnamara E, Levy HP, Biesecker B. The impact of living with Klinefelter syndrome: a qualitative exploration of adolescents and adults. J Genet Couns 2017;26(4):728–737. [PubMed: 27832510]

28. Zurynski Y, Deverell M, Dalkeith T, et al. Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. Orphanet J Rare Dis 2017;12(1):68. [PubMed: 28399928]
29. Williams JK, Schutte DL. Benefits and burdens of genetic carrier identification. West J Nurs Res 1997;19(1):71–81. [PubMed: 9030039]
30. von der Lippe C, Frich JC, Harris A, Solbraekke KN. "It was a lot tougher than I thought It would be". A qualitative study on the changing nature of being a hemophilia carrier J Genet Couns. 2017;26(6):1324–1332. [PubMed: 28547664]
31. Hawkins L, Centifanti LCM, Holman N, Taylor P. Parental adjustment following pediatric burn injury: the role of guilt, shame, and self-compassion. J Pediatr Psychol 2019;44(2):229–237. [PubMed: 30357372]
32. Chong LSH, Fitzgerald DA, Craig JC, et al. Children's experiences of congenital heart disease: a systematic review of qualitative studies. Eur J Pediatr 2018; 177(3):319–336. [PubMed: 29327140]
33. Taylor RM, Gibson F, Franck LS. The experience of living with a chronic illness during adolescence: a critical review of the literature. J Clin Nurs 2008; 17(23):3083–3091. [PubMed: 19012778]
34. Sander JW. Ultimate success in epilepsy--the patient's perspective. Eur J Neurol 2005;12 Suppl 4:3–11.
35. Liddle J, Fleming J, McKenna K, Turpin M, Whitelaw P, Allen S. Adjustment to loss of the driving role following traumatic brain injury: A qualitative exploration with key stakeholders. Aust Occup Ther J 2012;59(1):79–88. [PubMed: 22272886]
36. Kef S, Dekovic M. The role of parental and peer support in adolescents well-being: a comparison of adolescents with and without a visual impairment. J Adolesc 2004;27(4):453–466. [PubMed: 15288754]
37. Admi H Growing up with a chronic health condition: a model of an ordinary lifestyle. Qual Health Res 1996;6(2):163–183.
38. Rolland JS. Families, Illness, and Disability: An Integrative Treatment Model. New York: BasicBooks, 1994.
39. Walsh F Family resilience: a framework for clinical practice. Fam Process 2003;42(1):1–18. [PubMed: 12698595]
40. Bower JE, Moskowitz JT, Epel E. Is benefit finding good for your health?:Pathways linking positive life changes after stress and physical health outcomes. Curr Dir Psychol Sci 2009;18(6):337–341.

Table 1.

Means, Standard Deviations, Ranges, and Reliability Values for Quantitative Measures

	Mothers (n=9)	**Fathers (n=8)**	**Total Sample (n=17)**
Dispositional Optimism (Cronbach alpha 0.95)			
Mean (SD)	16.89 (6.90)	19.50 (1.93)	18.12 (5.22)
Range	3-24	17-23	3-24
Trait Anxiety (Cronbach alpha 0.93)			
Mean (SD)	34.56 (11.01)	35.10 (7.53)	34.53 (9.57)
Range	26-57	26-48	22-57
Extraversion (Cronbach alpha 0.90)			
Mean (SD)	4.04 (0.76)	3.69 (0.79)	3.88 (0.77)
Range	2.63-4.88	2.38-4.50	2.38-4.88
Agreeableness (Cronbach alpha 0.86)			
Mean (SD)	4.17 (0.71)	4.01 (0.67)	4.10 (0.67)
Range	2.89-5	2.89-4.67	2.89-5
Conscientiousness (Cronbach alpha 0.76)			
Mean (SD)	4.23 (0.55)	4.21 (0.51)	4.22 (0.51)
Range	3.44-4.89	3.11-4.67	3.11-4.89
Neuroticism (Cronbach alpha 0.90)			
Mean (SD)	2.32 (1.04)	2.48 (0.81)	2.40 (0.91)
Range	1.25-4	1.25-3.50	1.25-4
Openness (Cronbach alpha 0.73)			
Mean (SD)	3.84 (0.51)	3.51 (0.42)	3.69 (0.48)
Range	3.30-4.90	2.90-4.20	2.90-4.90