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Parents' perceptions of diagnostic genetic testing for children with inherited retinal disease in China

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Abstract

Background: In this study, we aim to investigate the awareness of, attitudes toward, and experiences with diagnostic genetic testing among parents of children suspected of having inherited retinal disease (IRDs) in China.

Methods: Semistructured, face-to-face, and in-depth interviews were carried out with parents of children with suspected IRDs in this qualitative study. Inductive content analysis was used for data processing.

Results: Forty-six parents participated in our interviews, and 47.8% of them supported genetic testing for following four main reasons: to help in making informed reproductive health decisions, to prepare for novel potential treatment, to identify the underlying causes of IRDs, and to satisfy curiosity about the heredity of IRDs. Among them, 19.6% were opposed to the testing for four main reasons, namely lack of therapeutic benefit, difficulty in affording the testing cost, doubt in the accuracy of clinical diagnosis, and the presence of concerns about the limitations of genetic testing. 47.8% of the parents expressed concerns that the genetic findings might lead to potential psychological stress.

Conclusion: In this study, we showed that nearly half of the parents supported genetic testing mainly for family planning, and a fifth of the parents were opposed to the testing mainly for lack of therapeutic benefit. Moreover, half of the parents expressed concern that a positive genetic result may create potential psychological burden to the parents and children.

KEYWORDS

genetic testing, inherited retinal disease, parents, pediatrics, qualitative interviews

1 | INTRODUCTION

Inherited retinal diseases (IRDs) are clinically and genetically a heterogeneous group of disorders that are important causes of childhood visual impairment (Sundaram, Moore, Ali, & Bainbridge, 2012). So far, more than 100 types of IRDs are known, including familiar exudative vitreoretinopathy (FEVR), retinitis pigmentosa (RP), Stargardt disease, and some complicated retinopathies with variable ocular involvements. IRDs are actually common. RP occurs with a frequency of 1:4000 (Pawlyk et al., 2016). The incidence rates of Stargardt disease vary from 1 in 10,000 to <1 in a million

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for rare clinical subtypes (Kalatzis, Hamel, & MacDonald, 2013; Lu, Liu, & Adelman, 2017; Mockel et al., 2011). At least a third of blindness and severe visual impairment in children is diagnosed as IRDs, and the number is increasing (Miyadera, Acland, & Aguirre, 2012). Until October 2018, 329 pathogenic genes associated with IRDs have been identified (https://sph.uth.edu/RetNet/disease.htm).

Genetic testing for IRDs has led to a considerable increase in the number of new genes or identified mutations and has now become a major tool in daily clinical practice. In the past decades, in addition to the four classical genes, namely *LRP5* (low-density lipoprotein receptor-related protein 5, OMIM *603506), *FZD4* (frizzled, drosophila, homolog of, 4, OMIM *604579), *NDP* (NDP gene, OMIM *300658), and *TSPAN12* (tetraspanin 12, OMIM *613138) in the Wnt signaling pathway, the genes *KIF11* (kinesin family member 11, OMIM *148760), *ZNF408* (zinc finger protein 408), and *CTNNB1* (catenin, beta-1, OMIM *116806) have recently been identified to be associated with FEVR (Hull et al., 2019; Poulter et al., 2012; Salvo et al., 2015; Tang et al., 2017). Genetic testing also facilitates the development of gene therapy for those once considered being untreatable IRDs (Jacobson et al., 2012; Sundaram et al., 2012), and human *RPE* 65 (ribulose 5-phosphate 3-epimerase 65, OMIM *180480) gene therapy for Leber's congenital amaurosis has been approved by the United States Food and Drug Administration (Pierce & Bennett, 2015). It is helpful for detecting other mutation-positive but asymptomatic family members and for making family planning decisions (Douma et al., 2010; Evers-Kiebooms & van den Berghe, 1979). However, the increasing availability of genetic tests for the general public has also raised ethical, social, and political concerns, such as testing confidentiality and disclosure of genetic information to family members (Billings et al., 1992; Botkin et al., 2015).

Since the onset of most of the IRDs is during childhood, visual acuity loss usually begins in their first two decades. Parents play an important role in opting for genetic testing. Many factors determine parents' decision-making, including, but not limited to, religion, disease severity, testing procedures, psychological conditions, and previous experience with the disease (Ahmed et al., 2008; Hurford, Hawkins, Hudgins, & Taylor, 2013; Ngim, Lai, Ibrahim, & Ratnasingam, 2013; Wertz, Janes, Rosenfield, & Erbe, 1992). Although research in the gene therapy of IRDs is already in clinical trials and the results are encouraging (Pierce & Bennett, 2015), research studies on its social and psychological impacts are lagging behind. Currently, most of the research studies were conducted in Western countries. Little is known about how much Chinese parents understand about the genetic information or the potential benefits and risks of genetic testing. Owing to the culture gap, there will be some novel findings in Eastern populations. Drawing on the analysis of the results of these in-depth interviews, we aim to evaluate the attitudes of parents of children with suspected IRDs in China toward genetic testing.

2 | MATERIALS AND METHODS

2.1 | Ethical compliance

This study was approved by the Institutional Review Board of Zhongshan Ophthalmic Center. All procedures performed in this study that involved human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. Written informed consent was obtained from parents prior to interviews.

2.2 | Study population

The qualitative study was conducted from August to December 2018 at Zhongshan Ophthalmic Center, Sun Yat-sen University. For capturing a wide range of perspectives, we recruited consecutive participants from the clinic of fundus diseases. The final sample consisted of 46 parents from 46 families, whose children were firstly diagnosed with suspected IRDs. To avoid the possible impact of positive results of genetic testing on the parents' responses, all the interviews were completed before genetic testing was prescribed.

2.3 | Data collection

On the basis of a literature review, we developed a semistructured interview guide to evaluate parents' opinions of eye-related genetic testing for their children. Once the children were suspected of having IRDs, the parents were first introduced to the up-to-date genetics/genomics information (e.g., types of genetic testing, the availability of direct-to-consumer genetic testing, and the potential availability of genetic testing for detecting genomic diseases and traits by the X. Ding), and then they were invited to a face-to-face, audio-taped, in-depth interview. Data on the sociodemographic characteristics of the participants and their families were collected, and parental attitudes toward genetic testing were assessed with open-ended questions by a single investigator (Y. Zhang) who had received training in qualitative research. All the interviewees were then asked three questions, "Question 1: how did you obtain the information about genetic testing?," and "Question 2: will you (not) allow your children to undergo genetic testing for IRDs?" These two questions were followed by an open-ended one: "Question 3 (in a hypothetical scenario): what effects will a positive genetic test result have on your family life?" With the participants' permission, all the interviews, which ranged from 20 to 40 min, were recorded digitally.

2.4 | Qualitative data analyses

NVivo 11.0 software (QSR International Pty, Doncaster, Australia) was used to transcribe the digital recording,

facilitate the coding process, and compare recurring themes across the transcription data. The inductive content analysis approach was adopted to analyze the participants' responses to open-ended questions (Graneheim, Lindgren, & Lundman, 2017). Initially, two investigators (S. Huang and L. Sun) independently reviewed the transcription data and developed a thematic framework. Then, the thematic framework was applied to the entire set of open-ended responses and refined to include new themes as they emerged. Although the codings of themes and subthemes between these two investigators were similar overall, two other investigators (S. Zhao and Y. Zhong) reviewed the transcriptions and checked the coded themes. Intercoder discussion was carried out when there were differences in coding assignments. Then, the coding structure was revised throughout the analysis and agreement about the themes for all responses was established. Finally, the first author (Y. Zhang) translated the quotes into the English, which were checked by a translator.

3 | RESULTS

3.1 | Sample characteristics

Of the 46 participating parents, 73.9% were mothers who had at least one child with a suspected IRD. The average age was 33.5 ± 5.3 years old (range 23–43, median 34). Of the children, 69.6% were boys and 30.4% were girls. The median duration of the disease was 23.0 months. Parents' educational levels included middle school or less (n = 17; 37.0%), high school diploma (n = 15; 32.6%), and college graduate or higher (n = 14; 30.4%). Annual household incomes (CNY) varied from <100K (n = 14; 30.4%), 100–200K (n = 18; 39.1%), 200–300K (n = 10; 21.7%), and ≥300K (n = 4; 8.7.%). Only eight of the participants (17.4%) had a family history of eye disease, and 13 (28.3%) intended to have more children. None of the parents had previously undergone genetic testing for IRDs. The sociodemographic characteristics of the interviewees are shown in Table 1.

TABLE 1 Sociodemographic characteristics of interviewees (n = 46)

	Value
Age (years, mean ± *SD*, rang)	33.5 ± 5.3, 23–43
Gender (No. [%])	
Male	12 (26.1)
Female	34 (73.9)
Married (No. [%])	46 (100.0)
Education levels (No. [%])	
Middle school or less	17 (37.0)
High school diploma	15 (32.6)
College graduate or higher	14 (30.4)
Annual household incomes (No. [%])	
<100K	14 (30.4)
100–200K	18 (39.2)
200–300K	10 (21.7)
≥300K	4 (8.7)
Family history (No. [%])	
Yes	8 (17.4)
No	36 (78.3)
Not sure	2 (4.3)
Whether to reproduce or not (No. [%])	
Yes	13 (28.3)
No	32 (69.5)
Not sure	1 (2.2)

3.2 | Interview findings

3.2.1 | Question 1: How did you obtain information about genetic testing?

For question 1, 27 (58.7%) of the 46 parents reported that they had never heard of genetic testing before visiting an ophthalmologist. The remaining 12 (26.1%) had heard from doctors only, but they did not clearly understand genetic testing, and only seven (15.2%) had acquired additional information from the Internet, but none had obtained information from scientific publications.

3.2.2 | Question 2: Will you (not) allow your children to undergo genetic testing for IRDs? And why?

For question 2, parents were asked to express their attitudes toward diagnostic genetic testing for IRDs, and 22 (47.8%) expressed willingness to undergo genetic testing. Nine parents (19.6%) expressed unwillingness, whereas 15 (32.6%) could not decide. Thirty-three parents (71.7%) had an annual household income of 100K or less, and all of them were unwilling to take the test. Seventeen parents (37.0%) had an education level of middle school or less, and 13 (76.5%) showed unfavorable attitudes toward the testing. No parents with a college degree or higher was against the genetic testing. Twenty-three parents (63.9%) who had no family history expressed opposition or conditional support for the test, and only two parents with a family history had unfavorable attitudes. Seven parents (53.8%) who intended to make family planning decisions had a positive attitude, and 14 (30.4%) who did not want to have offspring supported the genetic testing unconditionally. The attitudes of interviewees toward diagnostic genetic testing are listed in Table 2.

TABLE 2 Parents' attitudes to childhood diagnostic genetic testing for IRDs ($n = 46$)

	No. (%)	Unconditional support (*n*)	Conditional support (*n*)	Opposing testing (*n*)
Education levels				
Middle school or less	17 (37.0)	4	7	6
High school diploma	15 (32.6)	8	4	3
College graduate or higher	14 (30.4)	9	5	0
Annual household incomes				
<100K	14 (30.4)	4	7	3
100–200K	18 (39.2)	10	4	4
200–300K	10 (21.7)	3	5	2
≥300K	4 (8.7)	4	0	0
Family history				
Yes	8 (17.4)	6	0	2
No	36 (78.3)	13	16	7
Not sure	2 (4.3)	2	0	0
Whether to reproduce or not				
Yes	13 (28.3)	7	2	4
No	32 (69.5)	14	13	5
Not sure	1 (2.2)	0	1	0

Abbreviation: IRDs, inherited retinal diseases.

TABLE 3 Reasons for parents' perceptions to childhood diagnostic genetic testing for IRDs ($n = 46$)

	No. (%)
Reasons for unconditional support of diagnostic genetic testing for IRDs	22 (47.8)
Helping to make informed reproductive health decisions	16 (34.8)
Preparing for novel potential treatment	15 (32.6)
Identifying the underlying causes	10 (21.7)
Satisfying curiosity about the heredity of the disease	8 (17.4)
Reasons for opposing diagnostic genetic testing for IRDs	9 (19.6)
Lack of therapeutic benefit	7 (15.2)
Difficulty in affording the testing cost	5 (10.9)
Questioning the accuracy of clinical diagnosis of IRDs	4 (8.7)
Raising concerns about the limitations of genetic testing	3 (6.5)
Reasons for conditional supporting diagnostic genetic testing for IRDs	15 (32.6)
If the doctors highly recommended it	8 (17.4)
If the result would be helpful to the treatment	7 (15.2)
If the testing was affordable	5 (10.9)

Abbreviation: IRDs, inherited retinal diseases.

The reasons for and illustrative quotations on parents' perceptions of genetic testing for childhood IRDs are shown in Table 3. The details are described as follows.

3.3 | Reasons for unconditional support of diagnostic genetic testing for IRDs

3.3.1 | Helping to make informed reproductive health decisions

Thirty-three parents (72.8%) who were in favor of undergoing genetic testing indicated that it would be useful for avoiding giving birth to another mutation-positive child. One participant expressed:

> "In regard to our family, I would allow my children to take the test. We believe that the genetic testing would help us achieve the birth of a healthy child, and we can also know how to modify our child's diet." (Pt 16)

3.3.2 | Preparing for novel potential treatment

Fifteen parents (32.6%) expressed interest in testing their children for any potential therapy to obtain better eyesight. One parents indicated:

> "Although we were told that this disease currently is untreatable, we would like to have information and be prepared for new treatments in the future. A positive result in the genetic test could help us to look for new or alternative health care treatments." (Pt 5)

3.3.3 | Identifying the underlying causes

More than a fifth of the parents ($n = 10$; 21.7%) agreed to test their children and desired to know the underlying etiology. One mother stated:

> "My husband and I had a conversation yesterday. We should try our best to find the real reason and we would like to know who else in our family is at risk..." (Pt 12)

3.3.4 | Satisfying curiosity of the heredity of the disease

Eight parents (17.4%) indicated that they have no family history. Therefore, they were curious and wanted to verify whether there were any hereditary factors that cause the disease. One participant expressed:

> "All of my family members had no similar disease. Thus, I would like to know whether a genetic mutation was the underlying reason for my son's disease." (Pt 13)

3.4 | Reasons for opposing diagnostic genetic testing for IRDs

Nine parents (19.6%) expressed that they would not allow their children to take the testing. The main reasons are listed below.

3.4.1 | Lack of therapeutic benefit

Eight parents (16.4%) were opposed to the genetic testing. These participants declared that they did not perceive any value from the testing. They believed that, since the disease is untreatable, the testing was unnecessary. For instance, one participant indicated:

> "The test will not be helpful regarding his further treatment. My husband said that our son had been diagnosed with an inherited eye disease, which had been confirmed by several doctors and that no treatment is available." (Pt 22)

3.4.2 | Difficulty in affording the testing cost

In this study, 33 parents (77%) were of low socioeconomic status, with an annual household income of 100K or less. All of them were unwilling to take the test. Among them, five parents (10.9%) considered that the testing was too expensive, which affected their decision-making. For example, a mother stated:

> "The testing is too expensive, and I don't think I can afford it now. I prefer to do the testing in the future, if it becomes cheaper." (Pt 7)

3.4.3 | Questioning the accuracy of clinical diagnosis of IRDs

Considering that they had no family history, four parents from four families (8.7%) strongly doubted the accuracy of the clinical diagnosis. They did not think that the eye disease of their children is inherited. One mother doubted:

> "It is impossible. There is totally no family history of eye disease in our entire family. You see, my father-in-law and mother-in law, and my husband and I are completely normal…There should be something wrong…OK, I would like to see another doctor." (Pt 22)

3.4.4 | Raising concerns about the limitation of genetic testing

Three parents (6.5%) complained that the sampling procedure was invasive and that they had to wait for six weeks before obtaining the genetic results. One parents stated:

> "My baby is too young, considering the obvious discomfort during the blood sampling. Furthermore, I hope that I can get the result in two days." (Pt 3)

3.5 | Reasons for conditional supporting diagnostic genetic testing for IRDs

Fifteen parents (32.6%) expressed that they would be willing to receive genetic testing if (a) the doctors highly recommended it ($n = 8$; 17.4%), (b) the results would be helpful to

TABLE 4 Parents' perceptions to the influences of positive genetic testing result ($n = 46$)

	No. (%)
Being beneficial to family life	6 (13.1)
Potential negative effects for entire families	22 (47.8)
Hesitating to have another child	10 (21.7)
Worrying about future vision loss	7 (15.2)
Causing family conflicts	7 (15.2)
Potential discrimination for children	4 (8.7)
Have no effects on the family life	15 (32.6)
Having not considered this issue yet	3 (6.5)

the treatment ($n = 7$; 15.2%), or (c) the testing was affordable ($n = 5$; 10.9%).

3.5.1 | Question 3 (in a hypothetical scenario): What effects will a positive genetic testing result have on your family life?

For question 3, all the parents were further asked about the effects of the genetic testing. Six parents (13.0%) expressed that the information would be beneficial to their family life if a mutation is identified. However, 22 parents (47.8%) indicated that a positive result would have negative effects on their families, either on the parents or on the children. Among them, 18 parents (39.1%) stated that the result might increase anxiety and stress for parents; another four parents (8.7%) reported that the result would affect the psychological health of their children. Moreover, 15 parents (32.6%) said their family lives would not be affected by a positive result, and three parents (6.5%) responded that they had never considered this issue. The parents' perceptions toward the effects of the genetic testing result are described in Table 4.

3.6 | Being beneficial to family life

Six parents (13.0%) expressed that the genetic testing information would have a positive effect on their family. One mother indicated:

> "I think the genetic testing will be beneficial for our entire family. If I knew the test result, I would pay more attention to the latest progress in gene therapy and make sure that my child could get timely treatments. Additionally, I wish to understand the child's behaviors and take care of my child better in my daily life." (Pt 1)

3.7 | Potential negative effects for entire families

3.7.1 | Hesitating to have another child

Ten out of the 46 parents (21.7%) revealed that a positive result would increase parents' distress. One participant reported that she hesitated to have another child. She expressed her opinion:

> "My husband and I have been thinking about having our second baby for a long while. But now, if the genetic result is positive, we will have a big concern about this. As you know, a baby with poor visual acuity will bring great burden to himself, to the whole family, and even to society." (Pt 14)

3.7.2 | Worrying about future vision loss

Since most diseases associated with a genetic mutation are progressive and untreatable so far, seven parents (15.2%) worried that, if the disease is confirmed to have a genetic origin, the vision of their children would become worse in the future, which might make them lose the ability to take care of themselves in the future. One mother affirmed:

> "I am extremely worried that my child will become totally blind one day in the future. I think that is more terrible than suffering from cancer." (Pt 1)

3.7.3 | Causing family conflicts

Seven parents (15.2%) emphasized that the result would affect their family life, including their marital relationship and in-law relationships. One mother said:

> "I come from Zhanjiang, a city in the south of Guangdong province, where the traditional culture stresses that it is better to give birth to lots of children. Moreover, my husband and I would like to have more children. Thus, it might start family conflicts if the gene is from me. I would be blamed by my mother-in-law and my husband. For example, my mother-in-law will complain about why I have such a disease, and my husband may divorce me." (Pt 9)

3.7.4 | Potential discrimination for children

Four parents (8.7%) demonstrated that children might be hurt from potential discrimination because of the genetic abnormalities. One mother reported:

> "My child might have to face the issues of bullying and discrimination owing to a genetic disease, which would lead to the loss of self-confidence and study motivation." (Pt 9)

3.8 | Having no effects on the family life

Fifteen parents (32.6%) expressed that genetic testing was similar to other types of examination and had no effects on a person's life. One mother said:

> "It is just a testing, no matter what the result is. It would be better if there were no disease. I don't think there will be much influence on my family." (Pt 20)

3.9 | Having not considered this issue yet

Three parents reported that they had not yet considered this issue. One mother responded:

> "I had never thought about this issue. As for the future, it is hard for me to foresee it." (Pt 26)

4 | DISCUSSION

Similar qualitative studies have explored parents' attitudes toward childhood genetic testing for cancer (Alderfer et al., 2015; Bradbury et al., 2010; Godino, Jackson, Turchetti, Hennessy, & Skirton, 2018), autism spectrum disorders (Chen et al., 2015; Chen, Xu, Huang, & Dhar, 2013), and deafness (Mackley et al., 2018) in Western countries. This is a qualitative investigation that has explored parents' in-depth opinions toward genetic testing for IRDs in a Chinese population. This qualitative method can lead to learning about the impact of a genetic disorder on families by listening to their collective stories, analyzing their responses, and summarizing their experiences (Bernhardt, 2008).

Our study has several important findings. First, although the last decade has seen the extensive development of eye-related genetic testing, approximately half of our study participants had never heard about genetic testing, which was in line with a previous study about genetic testing for autism (Chen et al., 2013). Seven participants (15.2%) without a family history considered that this testing would not be useful for their children. Moreover, 46.2% did not perceive any value to family planning, and the percentage was higher than in other studies on prenatal genetic testing (Chen et al., 2015). In our study, 64.3% of college graduates and all the parents with annual household incomes ≥300K had a favorable attitude toward genetic testing, whereas 23.5% of the parents who graduated from middle school or less and 28.5% of the parents with annual household incomes of <100K supported the testing unconditionally. Our findings revealed that parents with low educational levels and annual household incomes tended to have relatively low levels of knowledge and awareness of genetic testing. Most published studies on genetic testing for children had focused on highly educated parents in high-income countries (Alderfer et al., 2015; Chen et al., 2015; Lim et al., 2017; Mackley et al., 2018), and we approached this important topic in a very different environment.

Second, 47.8% of the participants were in favor of eye-related genetic testing. Facilitating better arrangements for family planning, preparing for novel potential treatment, identifying the underlying causes and satisfying curiosity about the heredity of the disease were the main supporting reasons. However, approximately a fifth of the parents were opposed to genetic testing for their child and expressed their concerns regarding its cost, method, and effectiveness (Bradbury et al., 2010; Lowe, Corben, Duncan, Yoon, & Delatycki, 2015; Taber, Aspinwall, Kohlmann, Dow, & Leachman, 2010). Some of their concerns, such as genetic discrimination and potential psychological burden on parents and children, were in line with other similar studies (Bradbury et al., 2010; Lowe et al., 2015). These results confirmed that the behavior of Chinese parents might be shaped by the traditional Chinese culture, in which people with a genetic mutation are sometimes stigmatized (Chen et al., 2015; Chen, Zhao, Zhou, & Xu, 2012; Yang et al., 2013).

We found that various reasons contributed to the different attitudes toward genetic testing in our study. One was that few participants obtained sufficient information about eye-related genetic testing from their doctors. The other was that parents did not take the initiative and lacked effective means to acquire such knowledge. Given those factors affecting parents' decision, general knowledge of genetics and genetic testing needs to be publicized, and education on genetics for healthcare providers is needed to offer better genetic services.

Our study has several limitations. First, this study was conducted with only 46 parents, and the study setting was a regional referral hospital where parents were eager to seek treatments for their children; thus, the potential generalizability of the findings may be limited. In addition, previous studies have suggested that sociodemographic characteristics might affect individuals' perception of genetic testing (Chen et al., 2012). Different groups of ethnicity, income,

and educational level lead to various awareness levels and opinions. Therefore, a multicenter study should be conducted to compile the testing experiences of a large number of parents. Furthermore, a quantitative analysis that includes sociodemographic aspects, combined with a qualitative method, should also be carried out to comprehensively explore the factors that affect parents' attitudes toward genetic testing.

Overall, this study provided insight into the hopes and fears of Chinese parents on genetic testing for pediatric IRD parents and showed the main reasons for their support of or opposition to genetic testing. Half of the parents support IRD genetic testing, whereas a fifth of them oppose it. The main reasons for supporting are to help make informed reproductive health decisions and to prepare for novel potential treatment. However, the main reasons for opposing are the current lack of therapeutic benefit and difficulty in affording the testing cost. Furthermore, the genetic testing could produce multifaceted negative effects, including emotional stress in the whole family, fostering interpersonal and intrafamily hostilities, as well as potential genetic discrimination. We believe that the current study deepened our understanding about the attitudes of parents with IRD children, and thus is pivotal in offering better counseling services.

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CONFLICT OF INTEREST

None declared.

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REFERENCES

Ahmed, S., Hewison, J., Green, J. M., Cuckle, H. S., Hirst, J., & Thornton, J. G. (2008). Decisions about testing and termination of pregnancy for different fetal conditions: A qualitative study of European White and Pakistani mothers of affected children. *Journal of Genetic Counseling*, *17*(6), 560–572. https://doi.org/10.1007/s10897-008-9176-x

Alderfer, M. A., Zelley, K., Lindell, R. B., Novokmet, A., Mai, P. L., Garber, J. E., … Nichols, K. E. (2015). Parent decision-making around the genetic testing of children for germline TP53 mutations. *Cancer*, *121*(2), 286–293. https://doi.org/10.1002/cncr.29027

Bernhardt, B. A. (2008). The role of qualitative research in medical genetics: Listening to the voices of our patients. *American Journal of Medical Genetics Part A*, *146A*(24), 3132–3135. https://doi.org/10.1002/ajmg.a.32587

Billings, P. R., Kohn, M. A., de Cuevas, M., Beckwith, J., Alper, J. S., & Natowicz, M. R. (1992). Discrimination as a consequence of genetic testing. *American Journal of Human Genetics*, *50*(3), 476–482.

Botkin, J. R., Belmont, J. W., Berg, J. S., Berkman, B. E., Bombard, Y., Holm, I. A., … McInerney, J. D. (2015). Points to consider: Ethical, legal, and psychosocial implications of genetic testing in children and adolescents. *American Journal of Human Genetics*, *97*(1), 6–21. https://doi.org/10.1016/j.ajhg.2015.05.022

Bradbury, A. R., Patrick-Miller, L., Egleston, B., Sands, C. B., Li, T., Schmidheiser, H., … Daugherty, C. K. (2010). Parent opinions regarding the genetic testing of minors for BRCA1/2. *Journal of Clinical Oncology*, *28*(21), 3498–3505. https://doi.org/10.1200/jco.2009.27.2971

Chen, L. S., Xu, L., Dhar, S. U., Li, M., Talwar, D., & Jung, E. (2015). Autism spectrum disorders: A qualitative study of attitudes toward prenatal genetic testing and termination decisions of affected pregnancies. *Clinical Genetics*, *88*(2), 122–128. https://doi.org/10.1111/cge.12504

Chen, L. S., Xu, L., Huang, T. Y., & Dhar, S. U. (2013). Autism genetic testing: A qualitative study of awareness, attitudes, and experiences among parents of children with autism spectrum disorders. *Genetics in Medicine*, *15*(4), 274–281. https://doi.org/10.1038/gim.2012.145

Chen, L. S., Zhao, M., Zhou, Q., & Xu, L. (2012). Chinese Americans' views of prenatal genetic testing in the genomic era: A qualitative study. *Clinical Genetics*, *82*(1), 22–27. https://doi.org/10.1111/j.1399-0004.2012.01871.x

Douma, K. F., Aaronson, N. K., Vasen, H. F., Verhoef, S., Gundy, C. M., & Bleiker, E. M. (2010). Attitudes toward genetic testing in childhood and reproductive decision-making for familial adenomatous polyposis. *European Journal of Human Genetics*, *18*(2), 186–193. https://doi.org/10.1038/ejhg.2009.151

Evers-Kiebooms, G., & van den Berghe, H. (1979). Impact of genetic counseling: A review of published follow-up studies. *Clinical Genetics*, *15*(6), 465–474. https://doi.org/10.1111/j.1399-0004.1979.tb00827.x

Godino, L., Jackson, L., Turchetti, D., Hennessy, C., & Skirton, H. (2018). Decision making and experiences of young adults undergoing presymptomatic genetic testing for familial cancer: A longitudinal grounded theory study. *European Journal of Human Genetics*, *26*(1), 44–53. https://doi.org/10.1038/s41431-017-0030-1

Graneheim, U. H., Lindgren, B. M., & Lundman, B. (2017). Methodological challenges in qualitative content analysis: A discussion paper. *Nurse Education Today*, *56*, 29–34. https://doi.org/10.1016/j.nedt.2017.06.002

Hull, S., Arno, G., Ostergaard, P., Pontikos, N., Robson, A. G., Webster, A. R., … Michaelides, M. (2019). Clinical and molecular characterization of familial exudative vitreoretinopathy associated with microcephaly. *American Journal of Ophthalmology*, *207*, 87–98. https://doi.org/10.1016/j.ajo.2019.05.001

Hurford, E., Hawkins, A., Hudgins, L., & Taylor, J. (2013). The decision to continue a pregnancy affected by Down syndrome: Timing of decision and satisfaction with receiving a prenatal diagnosis. *Journal*

of Genetic Counseling, *22*(5), 587–593. https://doi.org/10.1007/s10897-013-9590-6

Jacobson, S. G., Cideciyan, A. V., Ratnakaram, R., Heon, E., Schwartz, S. B., Roman, A. J., … Hauswirth, W. W. (2012). Gene therapy for leber congenital amaurosis caused by RPE65 mutations: Safety and efficacy in 15 children and adults followed up to 3 years. *Archives of Ophthalmology*, *130*(1), 9–24. https://doi.org/10.1001/archophthalmol.2011.298

Kalatzis, V., Hamel, C. P., & MacDonald, I. M. (2013). Choroideremia: Towards a therapy. *American Journal of Ophthalmology*, *156*(3), 433–437.e433. https://doi.org/10.1016/j.ajo.2013.05.009

Lim, Q., McGill, B. C., Quinn, V. F., Tucker, K. M., Mizrahi, D., Patenaude, A. F., … Wakefield, C. E. (2017). Parents' attitudes toward genetic testing of children for health conditions: A systematic review. *Clinical Genetics*, *92*(6), 569–578. https://doi.org/10.1111/cge.12989

Lowe, G. C., Corben, L. A., Duncan, R. E., Yoon, G., & Delatycki, M. B. (2015). "Both Sides of the Wheelchair": The views of individuals with, and parents of individuals with friedreich ataxia regarding pre-symptomatic testing of minors. *Journal of Genetic Counseling*, *24*(5), 732–743. https://doi.org/10.1007/s10897-014-9801-9

Lu, L. J., Liu, J., & Adelman, R. A. (2017). Novel therapeutics for Stargardt disease. *Graefes Archive for Clinical and Experimental Ophthalmology*, *255*(6), 1057–1062. https://doi.org/10.1007/s00417-017-3619-8

Mackley, M. P., Blair, E., Parker, M., Taylor, J. C., Watkins, H., & Ormondroyd, E. (2018). Views of rare disease participants in a UK whole-genome sequencing study towards secondary findings: A qualitative study. *European Journal of Human Genetics*. *26*(5), 652–659. https://doi.org/10.1038/s41431-018-0106-6

Miyadera, K., Acland, G. M., & Aguirre, G. D. (2012). Genetic and phenotypic variations of inherited retinal diseases in dogs: The power of within- and across-breed studies. *Mammalian Genome*, *23*(1–2), 40–61. https://doi.org/10.1007/s00335-011-9361-3

Mockel, A., Perdomo, Y., Stutzmann, F., Letsch, J., Marion, V., & Dollfus, H. (2011). Retinal dystrophy in Bardet-Biedl syndrome and related syndromic ciliopathies. *Progress in Retinal and Eye Research*, *30*(4), 258–274. https://doi.org/10.1016/j.preteyeres.2011.03.001

Ngim, C. F., Lai, N. M., Ibrahim, H., & Ratnasingam, V. (2013). Attitudes towards prenatal diagnosis and abortion in a multi-ethnic country: A survey among parents of children with thalassaemia major in Malaysia. *Journal of Community Genetics*, *4*(2), 215–221. https://doi.org/10.1007/s12687-012-0133-x

Pawlyk, B. S., Bulgakov, O. V., Sun, X., Adamian, M., Shu, X., Smith, A. J., … Li, T. (2016). Photoreceptor rescue by an abbreviated human RPGR gene in a murine model of X-linked retinitis pigmentosa. *Gene Therapy*, *23*(2), 196–204. https://doi.org/10.1038/gt.2015.93

Pierce, E. A., & Bennett, J. (2015). The status of RPE65 gene therapy trials: Safety and efficacy. *Cold Spring Harbor Perspectives in Medicine*, *5*(9), a017285. https://doi.org/10.1101/cshperspect.a017285

Poulter, J. A., Davidson, A. E., Ali, M., Gilmour, D. F., Parry, D. A., Mintz-Hittner, H. A., … Toomes, C. (2012). Recessive mutations in TSPAN12 cause retinal dysplasia and severe familial exudative vitreoretinopathy (FEVR). *Investigative Ophthalmology & Visual Science*, *53*(6), 2873–2879. https://doi.org/10.1167/iovs.11-8629

Salvo, J., Lyubasyuk, V., Xu, M., Wang, H., Wang, F., Nguyen, D., … Chen, R. (2015). Next-generation sequencing and novel variant determination in a cohort of 92 familial exudative vitreoretinopathy patients. *Investigative Ophthalmology & Visual Science*, *56*(3), 1937–1946. https://doi.org/10.1167/iovs.14-16065

Sundaram, V., Moore, A. T., Ali, R. R., & Bainbridge, J. W. (2012). Retinal dystrophies and gene therapy. *European Journal of Pediatrics*, *171*(5), 757–765. https://doi.org/10.1007/s00431-011-1615-2

Taber, J. M., Aspinwall, L. G., Kohlmann, W., Dow, R., & Leachman, S. A. (2010). Parental preferences for CDKN2A/p16 testing of minors. *Genetics in Medicine*, *12*(12), 823–838. https://doi.org/10.1097/GIM.0b013e3181f87278

Tang, M., Sun, L., Hu, A., Yuan, M., Yang, Y., Peng, X., & Ding, X. (2017). Mutation spectrum of the LRP5, NDP, and TSPAN12 genes in Chinese patients with familial exudative vitreoretinopathy. *Investigative Ophthalmology & Visual Science*, *58*(13), 5949–5957. https://doi.org/10.1167/iovs.17-22577

Wertz, D. C., Janes, S. R., Rosenfield, J. M., & Erbe, R. W. (1992). Attitudes toward the prenatal diagnosis of cystic fibrosis: Factors in decision making among affected families. *American Journal of Human Genetics*, *50*(5), 1077–1085.

Yang, L. H., Purdie-Vaughns, V., Kotabe, H., Link, B. G., Saw, A., Wong, G., & Phelan, J. C. (2013). Culture, threat, and mental illness stigma: Identifying culture-specific threat among Chinese-American groups. *Social Science and Medicine*, *88*, 56–67. https://doi.org/10.1016/j.socscimed.2013.03.036

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