Understanding of and attitudes to genetic testing for inherited retinal disease – a patient perspective.

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Abstract

Background/aims: The views of people with inherited retinal disease are important to help develop health policy and plan services. This study aimed to record levels of understanding of and attitudes to genetic testing for inherited retinal disease, and views on the availability of testing.

Methods: Telephone questionnaires comprising quantitative and qualitative items were completed with adults with inherited retinal disease. Participants were recruited via postal invitation (response rate 48%), approach at clinic or the newsletters of relevant charitable organisations.

Results: Questionnaires were completed with 200 participants. Responses indicated that participants' perceived understanding of genetic testing for inherited retinal disease was variable. The majority (90%) considered testing to be good/very good and would be likely to undergo genetic testing (90%) if offered. Most supported the provision of diagnostic (97%) and predictive (92%) testing, but support was less strong for testing as part of reproductive planning. Most (87%) agreed with the statement that testing should be offered only after individual have received genetic counselling from a professional. Subgroup analyses revealed differences associated with participant age, gender, education level and ethnicity ($p<.02$). Participants reported a range of perceived benefits (e.g. family planning, access to treatment,) and risks (e.g. impact upon family relationships, emotional consequences).

Conclusion: Adults with inherited retinal disease strongly support the provision of publicly funded genetic testing. Support was stronger for diagnostic and predictive testing than for testing as part of reproductive planning.

INTRODUCTION

Inherited retinal diseases are an important cause of visual disability, leading to loss of visual field (peripheral vision), visual acuity (detailed, central vision) or both. Individual conditions are rare and the most common conditions, retinitis pigmentosa (RP) and Stargardt disease occur in only 1:3,700 and 1:10,000 individuals respectively.[1, 2]

To date, almost 250 disease-causing genes have been identified or mapped. The number of different genes involved makes genetic testing difficult but not impossible. Using the phenotype and family history to direct testing, screening for common mutations or with the use of next generation sequencing, it is possible to identify a faulty gene or mutation in 40–70% of selected cases.[3, 4]

The identification of the genetic basis of an inherited retinal disease has the potential to offer many advantages. It can provide a precise clinical diagnosis, confirm the condition is inherited and the pattern of inheritance, provide a more accurate guide to future visual function, assist marriage and family planning and may also allow patients to be added to disease registries, giving them early access to clinical trials and emerging treatments that are gene- or mutation-specific. Despite these potential benefits and support from clinicians and patient groups, the availability and uptake of genetic testing for inherited retinal diseases within the publicly funded National Health Service (NHS) are variable.[5] This may be the result of several factors, including cost, perceived clinical utility, variations in the commissioning and provision of specialised eye genetic services and a lack of evidence of demand from service users. We have completed preliminary research into these issues involving a survey of delegates at a national patient conference.[6] This study aimed to investigate these issues further by investigating perceived understanding of and attitudes to genetic testing in a larger sample of individuals with inherited retinal disease and exploring potential differences between subgroups.

MATERIALS AND METHODS

Participants: Invitation letters were sent to patients who had previously attended eye clinics in Yorkshire. Participants were also recruited from the eye clinic or via newsletters of two national charities, RP Fighting Blindness and the Macular Society. At the time of recruitment, access to diagnostic genetic testing in a clinical laboratory was not routinely available locally. Patients aged over 16 years, with a clinical diagnosis of inherited retinal disease but without a significant hearing impairment were eligible. Invitation letters were available in English and Urdu and the study information leaflet and consent forms were available in print, electronic and Braille formats. Ethical approval was received from the Leeds (East) Research Ethics Committee (10/H1306/90) and informed consent was obtained from all participants.

Questionnaire: Semi-structured, telephone questionnaires were conducted with participants. Demographic information was collected, together with both quantitative and qualitative data on the level of understanding of, attitudes to and the availability of genetic testing for inherited retinal disease. Three Likert scale items explored understanding ("Do you feel that you understand what a genetic test is?"), attitudes ("Based on what you know right now, do you consider genetic testing for inherited eye disease to be good or bad?") and willingness to undergo genetic testing ("Based on what you know right now, how likely would you be to have a genetic test for inherited eye disease if offered tomorrow?"). All used a five-point scale, with 1 indicating a strong negative response or level of support and 5 indicating a strong positive response. A further eight questions explored participants' support for the availability of genetic testing for inherited retinal disease in general and for testing in specific circumstances namely diagnostic testing, testing in children under 18 years, predictive testing, carrier status testing for reproductive planning, pre-implantation genetic diagnosis and pre-natal diagnosis. For these questions, the choice of responses was limited to "yes", "not sure" or "no". Responses to

questions were followed by prompts or further questions to clarify or expand the initial answer. Questionnaires typically lasted approximately 30 minutes, were conducted in English, Urdu, Punjabi or Mirpuri by TAW, BP or MA and recorded.

Data analysis: Quantitative data were analysed for the whole sample and then according to each of seven, pre-determined subgroups: age (</≥50 years), sex, ethnicity (White British/other), highest educational level (up to GCSE or O level/College or higher), sight impairment certification status, parenting status (current or planned parent/no parenting plans) and the presence/absence of other affected family members. In addition, exploratory analyses revealed that participants with congenital conditions did not differ from those with acquired visual impairment in perceived understanding, attitude, nor the likelihood of undergoing testing (although numbers in the former group were small, precluding meaningful statistical comparisons). Differences in responses to Likert scale items were analysed using the Mann-Whitney U test for independent groups. The subgroups were compared in their responses to the items concerning the availability of testing for different purposes. This analysis was conducted using Chi-square or Fisher's exact test, as appropriate. Where significant differences were observed, independence between subgroups was tested using Chi-square analysis. Due to the number of tests being conducted, a more stringent significance level of $p<.02$ was applied.

Responses to open-ended questions were coded independently by two researchers (BP, TAW). Results were compared and differences resolved by consensus. The summarized statements, transcribed verbatim from the original recordings, were analyzed using a thematic approach, a common analytical method in this area,[7, 8] and managed using NVivo8. Selected responses were chosen to illustrate the differing levels of understanding of genetic testing, attitudes to and support for genetic testing.

RESULTS

The sample comprised 200 participants with a clinical diagnosis of inherited retinal disease. The majority (n=129) were recruited following postal invitation, for which the positive response rate was 48.1%. Other participants were recruited from clinic (n=41), newsletters (n=28) or via contact with affected relatives (n=2). There were 110 females (55%) and participants' median age was 50 years (range 18-84 years). Demographic data and the most common clinical diagnoses are presented in Table 1.

Table 1: Participant demographic data

Clinical diagnosis	
Retinitis pigmentosa	90 (45%)
Stargardt disease	26 (13%)
Cone dystrophy	8 (4%)
Sorsby fundus dystrophy	6 (3%)
X-linked retinoschisis	5 (2.5%)
Best disease	4 (2%)
Choroideremia	4 (2%)
Leber congenital amaurosis	4 (2%)
Doyne honeycomb dystrophy	3 (1.5%)
Achromatopsia	2 (1%)
Oculo-cutaneous albinism	2 (1%)
Other or unspecified macular dystrophy	38 (19%)
Other generalised retinal dystrophy	8 (4%)
Age range	
≥ 50 years	101 (50.5%)
< 50 years	99 (49.5%)
Ethnicity	
White British	167 (83.5%)
British Asian	31 (15.5%)
Mixed or other ethnic origin	2 (1%)
Highest level of education	
Primary school / no qualifications	36 (18%)
O or GCSE level	55 (27.5%)
College – diploma	39 (19.5%)
University degree	48 (24%)
Postgraduate	21 (10.5%)
Not answered	1 (0.5%)
Sight impairment certification status	
Severely sight impaired	111 (55.5%)
Sight impaired	36 (18%)

Not certified	50 (25%)
Not known	3 (1.5%)
Parenting status	
Have or plan to have children	169 (84.5%)
Decision taken not to have children	31 (15.5%)
Other affected family members	
Other affected family members	110 (55%)
No other affected or known relatives	90 (45%)

Responses to the Likert scale items are presented in Figure 1. Overall, the level of self-reported understanding about genetic testing was variable: 33% of the sample reported that they had no or little understanding, while 41% perceived themselves to have a high level of understanding (Figure 1; illustrative quotations are provided in Box 1). Participants educated to College-level or beyond reported a greater level of understanding than those with lower educational attainment ($p=0.019$). Attitudes towards genetic testing for inherited eye disease were largely positive: 90% considered testing to be good/very good. Responses to the third Likert scale item (concerning willingness to undergo testing) were similarly positive, with 90% being likely/very likely to undergo genetic testing. Responses to these items did not differ significantly across subgroups. Participant views of potential benefits and risks of genetic testing are illustrated in Boxes 2-3.

Views on the general availability of genetic testing and its use for particular purposes were examined with a series of categorical items (Figure 2). The majority of participants supported diagnostic testing as a publicly funded service: 93% felt that the NHS should offer genetic testing for inherited retinal disease. Support was strong for both diagnostic (96.5%) and predictive testing (91.5%). Only 17% of participants thought that genetic testing should be limited to adults over the age of 18 years, while 87% felt that it should be offered only after the provision of information and genetic counselling. Support for genetic testing as part of

reproductive planning was mixed: 65% were in favour of carrier status testing, 52% supported pre-implantation genetic diagnosis and 45% were in favour of pre-natal diagnosis.

Some subgroup differences emerged in responses to these items. First, age and sex effects were observed when asked whether genetic testing should be limited to those over 18. A Chi-square test indicated that responses were not equally distributed between younger and older participants (χ^2 (2, N=200) =12.24, p=0.002). Inspection of the frequency data indicated that younger participants were more likely to disagree with the statement that testing should be limited to those over 18 years (80.2%) than older participants (57.6%). Males and females also differed in their views on this issue (χ^2 (2, N=200) =7.75, p=0.020), with females more likely than males to agree with such an age limit (23.6% vs. 8.9%). To assess the independence of the age and sex effects, a further Chi-square test was conducted and the two variables were found to be independent of each other (χ^2 (1, N=200) =1.02, p=0.324).

Effects of age and education were observed in relation to the use of prenatal genetic testing. Here, younger participants were more likely to support the use of prenatal testing (χ^2 (2, N=200) =7.16, p=0.021; 50.5% vs. 39.4%), although this result did not meet our stringent level of significance. Responses were not equally distributed between groups categorised by educational attainment (χ^2 (2, N=199) =13.21, p=0.001). Those completing a higher level of education were more likely to oppose the option of prenatal testing (50.9% vs. 28.6%) and respondents with lower educational attainment reported a greater level of uncertainty around this issue (Not Sure: 22.0% vs. 8.3%.). The effects of age and education were also found to be independent (χ^2 (1, N=199) =2.18, p=0.156).

Finally, responses to the availability of carrier status testing were not equally distributed between ethnic groups (χ^2 (2, N=200)=11.69, p=0.003). Frequency data indicated that

participants of British Asian, mixed or other ethnicity were more likely to support access to carrier status testing than White British participants (90.9% vs. 59.9%).

There were no differences between subgroups according to sight impairment certification, parenting status, or the presence of other affected family members.

DISCUSSION

This study explored understanding of and attitudes to genetic testing for inherited retinal disease in a large sample of affected adults. The aim was to collect data to inform inherited retinal disease services, improve information provision and assess current demand for genetic testing.

When participants were asked to self-rate their level of understanding of genetic testing for inherited retinal disease, a wide range of responses was obtained. The only subgroup difference in perceived understanding was due to education: those educated to College level or above reported a significantly greater understanding. In general, public understanding of genetic science appears to be variable.[9, 10] Many people have difficulty explaining the meaning behind the concepts of 'genetics' and 'genes', despite being familiar with the terminology.[10] One study found that females, younger participants (18-44 years) and those with higher educational attainment were more likely to possess greater knowledge in this field.[11] However, unlike this research, these studies were all conducted with the general public. Further exploration of understanding within patient samples is warranted to assess whether knowledge is greater in those affected by genetic conditions.

The majority of participants viewed genetic testing for inherited retinal disease very positively. Support was very strong support for the provision of publicly funded diagnostic and predictive

genetic testing. However, most participants were in favour of information provision and access to genetic counselling before genetic testing. These findings are consistent with existing research in similar patient groups.[6, 12-14] Support was less strong for genetic testing as part of reproductive planning. The use of pre-implantation genetic diagnosis to achieve an unaffected pregnancy has been reported in cases of recessive Stargardt disease, severe RP and X-linked retinoschisis[4, 15] and pre-natal testing has also been reported for Leber congenital amaurosis.[3] Sizeable proportions of the current sample supported the use of genetic testing for reproductive planning purposes: 65% of participants supported carrier status testing, and 52% and 47% supported pre-implantation and prenatal genetic testing for inherited retinal disease, respectively. Similar figures have been reported elsewhere.[4, 6]

Participants' comments provided additional important information on attitudes to pre-implantation and prenatal testing. While approximately half of the sample felt that these services should be available, they would not necessarily choose to utilise them themselves. This finding may help to explain the phenomenon of high hypothetical but low actual uptake of (predictive) genetic testing.[16] This pattern has been consistently observed in populations affected by Huntington disease[17, 18] which has been considered a model of understanding the attitudes towards (predictive) testing for late-onset conditions with no treatment or cure[19] such as many inherited retinal diseases.[12] Our research suggests that when planning genetic testing services, patient attitudes should be explored in depth.

Of interest in our study was stronger support for carrier status testing and reproductive planning in British Asian participants. This may reflect greater awareness of the risk in communities in which inherited retinal disease is more common.[20] For some participants, genetic status might be one of the considerations when arranging a marriage. Others have explored genetic testing issues in similar populations[21, 22] who might be marginally more

affected by autosomal recessive genetic conditions due to a proportion of consanguineous marriages.

Several common themes emerged in describing the potential benefits of testing. Frequently cited benefits included greater understanding and knowledge about the genetic basis of the condition, as well as early access to emerging therapies. Participants also reported benefits to family members and future generations, as well as to society in general. They were often aware of limited personal benefit but felt that the information gained from testing may contribute to treatments of others in the future.

Participants were also asked about potential negative consequences of genetic testing. Several suggestions were offered, although a substantial number of respondents reported that they did not consider there to be any drawbacks. Reported disadvantages included the potential impact upon family relationships (i.e. feelings of guilt from passing a condition on, or blame in those who have inherited it) and the potential for results to be used to terminate pregnancies or increase insurance premiums. A substantial number of participants felt anxiety about their future. Many had ethical considerations. Emotional consequences of a result were frequently mentioned as disadvantageous. Other studies provide context to this finding. Mezer et al.[12] reported emotional distress in 57% of affected adults and their family members when recollecting their own predictive testing as children. By contrast, an investigation of a large family undergoing testing for hereditary myocilin glaucoma found no adverse impacts of predictive testing, five years following initial counselling[23] and a systematic review of various genetic conditions also showed no long-term sequelae either for carriers or non-carriers.[24]

Some limitations of the study must be acknowledged. Our sample was self-selecting and it is therefore possible that participants were more motivated and held more favourable attitudes

towards genetic testing than those who were invited but opted not to participate. Due to the recruitment methods employed, it is also acknowledged that study participants were currently engaged with the healthcare system and/or voluntary organisations. Several participants pointed out that whereas they wanted to know and were accepting of their diagnosis, other family members were not, and preferred ignorance of their genetic status. It is therefore to be expected that they may hold views different to those held by the participants in our research. Nevertheless, strengths of the study include the large number of affected individuals, with a range of clinical diagnoses and demographic characteristics.

Individuals with inherited retinal disease express strong support for the provision of genetic testing, particularly diagnostic and predictive testing. Most are aware of a number of possible benefits but not the potential negative consequences. There is a need for the provision of information, in a format accessible to those with visual impairment, and access to genetic counselling before testing and this would be in keeping with patients' expectation. Our results indicated that information may be most effectively targeted toward less educated individuals, who reported lower levels of understanding and greater uncertainty around prenatal testing. However, support for genetic testing for inherited retinal disease is not universal and many participants (typically those with a higher level of education) were in favour of access for others but not by themselves, particularly in relation to prenatal testing for reproductive planning.

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Box 1: Understanding of genetic testing

Understanding of testing
The quantitative results (see Figure 1) showed that participants' perceptions of their understanding of genetic testing and what it involved was variable. This was further illustrated by their comments:

"To be honest I don't have any idea…I don't really know what you do. I'd probably find it interesting if I did know. Do you look at chromosomes?" [ID141]

"Yes, it's presumably to try and find out about the genes that cause the hereditary problem" [ID195]

"I've had a blood test. I've had eye tests and all sorts. I'm not sure I've ever had a genetic test" [ID182]

Several participants expressed uncertainty about the technical terms involved and would have appreciated a simplified explanation:

"Always big words, tend to baffle normal people don't they?... It would be better to sort of understand it better in layman's terms" [ID012]

A lack of understanding was often not a concern however, with many participants content with their current level of knowledge:

"They did discuss things like that [autosomal dominant, recessive etc.] but I don't know huge amount about…er, genetic testing"

Interviewer: What more would you like to know?
"Nothing really" [ID089]

Box 2: Advantages of genetic testing

Advantages/benefits of genetic testing
All participants were asked to outline what they considered to be the benefits of genetic testing for inherited retinal disease, and all were able to provide examples, including those who were not particularly in favour of being tested themselves.

A common response was that testing would provide additional information, which may be beneficial for multiple reasons. For example, it might confirm a diagnosis, lead to further treatment options or aid understanding and remove uncertainty. For others, even if it didn't result in new treatments, the basic act of gaining knowledge was beneficial.

"Information is power, if people know what's going on then they're actually in a position to try to do something about it and it stands to … increase whatever options are available, whatever treatments are out there" [ID108]

"I'd have more understanding and less fear in dealing with the disease" [ID030]

"…definitely a good thing…the more you know the better" [ID145]

While participants understood that there were no curative treatments presently available, many expressed the view that genetic testing might help them in the future if and when new treatments became available:

"If it means you're a candidate for gene therapy or something, then that is a good thing…you want to be first in the queue if it does happen" [ID205]

Or if not them personally, then future generations:

"[I]t might not help me at my age, but if it helps people like my daughter or younger people, if it's got some sort of cure or prevention for the future then that's obviously going to be good isn't it?" [ID051]

Many participants felt that genetic testing could prove helpful when considering reproductive options as it would indicate the likelihood of children being affected.

"It's extra information. It can help clarify for people if they are thinking about having a family what the possible risks are for their children. It may at some point be able to give people a more definite diagnosis and a clearer prognosis" [ID205]

Box 3: Disadvantages of genetic testing and attitudes depending on circumstances

Attitudes depending on circumstances

While the majority of participants were in favour of testing being available, several reported that they wouldn't necessarily find it useful themselves. Its utility may relate to being at a particular stage of life, and outside of this it may not provide any noticeable benefit:

"I don't know how you could use the information to be honest. I suppose the only way it's beneficial is if…you actually are in a relationship with someone…and wanting to have children…Apart from that I can't see it being of any use to me whatever" [ID206]

Another participant explained that because of the potential significance of the result upon reproductive options, they would only want to have a test when they were in a couple and could receive the information together:

"I wouldn't want [a genetic test]…until I was at the point where I was beginning to think about starting a family…it's a challenging thing to explain to a partner and I'd actually rather go through it…with that individual, rather than knowing the result now and having the responsibility of making them aware of it" [ID205]

A number of participants also believed that it was important to provide genetic testing to identify potential carriers, use in selecting the embryos and prenatal diagnosis but they themselves would not use it:

"I think it should be available as a choice. Wouldn't be one I would go for but I think it can be

for other people to choose." [ID205]

Disadvantages/negative aspects of testing

While many participants stated that they could not think of any disadvantage of genetic testing for inherited retinal disease, several negative aspects were suggested. For many, the emotional impact of receiving bad news was the main disadvantage of testing (although this may be transient):

"I would be an emotional wreck for a while until I got my head around it" [ID107]

Another disadvantage frequently cited was the potential impact of the results upon family relationships. Two parents who had undergone genetic testing described the impact of receiving confirmation that their condition had been passed to their children:

"I found it very hard to deal with it. I was getting the bad news along with my daughter and I went out on a guilt trip because I had passed it on" [ID052]

"It's an awful thought actually, to know that you've passed it on… I know people say it could've been something worse, but…" [ID103]

Others raised the possibility of children blaming their parents:

"Obviously your parents…I'm not into this blaming…whose fault it was or anything, just to find out where it's come from… That's probably the negative…you end up…blaming somebody for…what you've got…the disease you've got" [ID014]

Many participants raised the issue of eugenics and genetic testing being used as a rationale for treating individuals differently. For example,

"I wouldn't want to use it for selective breeding. My view is that everybody is equal…and everybody's life is of value. So I wouldn't want…genetic testing to be used to…treat some people as of lower value…than others" [ID073]

Issues concerning insurance were also frequently reported:

"It would worry me greatly if insurance companies get access to the information and could adjust their premiums by what comes out of a genetic test. That's the bad side of it I think" [ID170]

Figure legends

Figure 1 Participant understanding of, attitude to, and willingness to undergo genetic testing for inherited retinal disease

Figure 2 Participant views on the general availability of genetic testing and its use for particular purposes