A summary of the impact of further sight loss in people with sporadic and familial aniridia

1. Introduction

This document is a summary of research for an MSc dissertation in Genetic and Genomic Counselling, in collaboration between Cardiff University and the Aniridia Network.

The study aimed to investigate the psychosocial impact of further sight loss in aniridia, and compare the experiences of those who had inherited aniridia from a parent, with individuals who were the first in their family with the condition.

2. Literature review

A literature search was done in May 2022 and updated in May 2024 to find research that had already been done into this topic or similar topics.

There was no other research at the time about the impact of deteriorating sight specifically in aniridia, but two peer-reviewed studies have been published that look into the psychosocial impact of aniridia in general. Findings included that one of the most challenging aspects of aniridia was the experience of ocular pain, and having aniridia was found to be associated with anxiety (Landsend et al. 2023; Sigurdardottir et al. 2023).

As there was a scarcity of research into aniridia, research into Retinitis pigmentosa (RP) was also reviewed. RP is an umbrella term for a range of genetic eye conditions which affect the retina – the part of the eye which is responsible for receiving light. Although RP differs from aniridia in some important respects (in particular, people with RP are born sighted and lose sight progressively from their mid teens onwards) RP is less rare than aniridia and more research has been published into the impact of progressive sight loss in individuals with RP, so it could provide

some findings that may be transferable to aniridia. Some studies into the experience of people with RP found that having a poorer visual field and visual acuity was correlated with having a lower Vision Related Quality of Life (Sugawara et al. 2010; Azoulay et al. 2015; Sainohira et al. 2018), and sometimes with a higher likelihood of experiencing features related to depression and anxiety (Azoulay et al. 2015; Moschos et al. 2015). These studies were not conducted on the same group of individuals over time as their sight decreased, so we cannot draw a causative link between the deterioration of sight and their mental health experiences, but it does suggest some link between overall level of sight and emotional toll.

This is reinforced by some qualitative studies involving in depth interviews with people with RP such as Prem Senthil et al. (2017), which found that people's experiences of deteriorating sight had an impact on people's emotional experiences and mental health. Some examples are listed below:

- Struggling to perform important day-to-day tasks
- Concerns about disease progression, outcome and personal safety
- Facing significant emotional and psychological challenges
- Experiencing a range of visual symptoms
- Adopting different strategies to manage stressful circumstances

Given that these were identified in individuals with RP, they may not be exactly applicable to the experiences of those with aniridia but propose some potential parallels across different sight conditions and the experiences of sight loss.

The other topic that this research project examined was the comparison between the experiences of those with the familial and sporadic form of aniridia. No other research related to this question was could be found by the researcher, suggesting there may be a gap in knowledge and understanding into this.

Overall the literature review found that there was little research into the experiences of those with aniridia, and no literature that compared experiences of people who had inherited a condition, versus people who were the first in their family to have it. This highlighted the utility of the research project.

3. Methodology

Participants were recruited via the Aniridia Network newsletter, website and social media accounts. Consent was then taken via either a printed or online consent form, and one-to-one interviews were held online over zoom, with an average length of 57 minutes. These interviews were recorded, and anonymised transcripts were made of each interview.

The researcher then used an approach of thematic analysis, following Braun and Clarke (2006) on the interview transcripts, whereby the transcripts were read over and over again, with key aspects noted and then important features coded and grouped into themes.

4. Results

4.1. Participant demographics

In terms of the demographics of the participants, the split between people with sporadic and familial aniridia was relatively even, with seven participants having sporadic aniridia and six having familial aniridia. Eight of the participants were female and five were male. The age of the participants was well distributed, with most participants aged between 40 and 59. There were no participants below the age of 25 and none aged over 80.

4.2. Themes identified

Overall, four themes were identified. These will be discussed below in turn.

4.2.1. Theme One: Constant adaptation

When asked about their experiences of deteriorating sight, a common experience amongst participants was the constant adaptation that it involved. This often started with individuals realising that their sight was changing because they were no longer able to do certain things in their

day-to-day life such as reading instructions on food packaging, being able to pour a drink or seeing doorways. This often had a significant emotional toll on participants – individuals described feeling depressed and a sense of helplessness. Others described the profound impact that their sight loss had on their personal identity – one participant said that it had “taken away a lot of who I am in some ways”, whilst another described their experience of sight loss as a grieving process.

Participants explained that they coped with their changing sight by using trial and error to find alternative ways of carrying out tasks, and by using aids and adaptations such as assistive technology, magnifiers, mobility aids, hats and sunglasses, and changing lighting levels in domestic or work environments. Although these adaptations enabled people to retain their independence, the process of having to find what worked was in and of itself often very challenging for people, especially those who did not have a family member with aniridia because they had much less knowledge of what was available. Participants also sometimes experienced a tension between their identity as independent on one hand, and their reluctance to ask for help and support on the other. Participants understood that by asking for help, they could sometimes retain their ability to do certain tasks and actually maintain their independence, even if it didn’t feel like that initially.

4.2.2. Theme Two: Anxiety

The second theme that emerged from the research was anxiety, which was generally associated with two distinct areas.

The first was one area of difference between those who had inherited aniridia and people who had the sporadic form. For participants who had inherited aniridia, there was often family experience of people who had had treatment for secondary complications of their aniridia which had not gone well. This meant that other members of the family were somewhat wary of operations or interventions that they may need to have in the future, summed up by one participant in the following quote, in relation to their mother: “I might not be so fearful of the operation that will be coming up at some point if hers hadn't gone so wrong”. Anxiety related to future treatment or intervention was therefore something significant that people with inherited aniridia, especially those who were experiencing a decline in their sight, were facing.

Another contributing factor to anxiety or worry about future treatment was negative healthcare experiences that participants had had. Anger, lack of empathy and ignorance about aniridia were some of the feelings that participants had experienced after accessing ophthalmology healthcare.

The other source of anxiety for participants was to do with the inherent uncertainty around their sight in the future. Unanswerable questions around the extent and timing of future sight loss and the impact it might have on various aspects of their lives such as caring for family members, work, and social activities were challenging for participants to cope with. This was summed up in the following quote:

- “So, that was a big thing that I kind of had to adjust to… the future at some point will be that I have very limited sight... and I don't know how long that would be. No one can tell me, I don't know whether it’s gonna be five years or 20 years.”

Some participants felt very fearful about the prospect of losing their sight completely. Others felt that completely losing their sight could, in some ways, be preferable to the constant uncertainty that they had to live with because there would at least be some stability or sense of an end to their journey once they were completely blind.

Some participants who did not have family members with aniridia expressed gratitude that they had not known about the possibility of future sight loss earlier, as they had not had the uncertainty hanging over them when they were younger.

One way of coping with the anxiety participants felt around their future sight was proactively channelling energy into preparing for future sight loss and “futureproofing” aspects of their life, such as researching different support options, learning Braille or becoming familiar with how to use screen readers.

Anxiety about future sight loss did not affect all participants, however, and some felt that they did not worry too much about what the future held for their sight. These participants had often had quite a stable level of sight in their adult lives.

4.2.3. Theme Three: A changing relationship with the outside world

The third theme related to how participants' changing levels of sight affected their relationship with the outside world. This occurred in three different ways.

Firstly, leaving the house became more complicated, especially going out alone or going to places they had not been to before. This meant that participants went to places that were more familiar, or went out with other people, meaning they could be less spontaneous about where they went and when. Personal safety was also a concern for participants, and others had comments from members of the public about their disability which discouraged them from going out. There was also some frustration about how various aspects of the external environment were not well adapted for people with a visual impairment, such as narrow pavements, lack of safe road crossings, or the increase in number of self-service checkouts.

Secondly, several participants highlighted that work was a part of their lives that had been affected by their changing levels of sight. Some participants had had to stop work altogether due to their changing sight, whilst others felt that because of their sight loss, and the unpredictability of it, committing to a consistent work routine would be very difficult. Many participants had also had positive experiences with their employers, who had been very supportive during their sight loss. The key element here was flexibility, allowing participants to attend healthcare appointments, access treatment or take time off as and when they needed to.

Thirdly, participants highlighted that their approaches towards asking for help when they were out and about had changed. Some participants who had inherited aniridia from a parent felt that they found it easier to ask for help from strangers because they had grown up with a parent who did this. For other participants, requesting help was not something that came naturally, so they had to force themselves out of their comfort zone, or even reported that they had become more outgoing or less reserved in their personalities as a result.

4.2.4. Theme Four: Support

The final key theme that cropped up when participants were asked about their further sight loss was support. This was one area where the experiences of participants with familial and sporadic aniridia differed significantly, most obviously in people's experiences of knowing about and navigating available support. Whether they had sporadic or familial aniridia themselves, in general participants felt that those growing up in a family who were fully sighted was a disadvantage in one way because the level of knowledge and experience around what was helpful or areas of support was much lower. There was a sense with support that "you didn't know what you didn't know", and some participants with sporadic aniridia highlighted the loneliness and isolation arising from being the only ones in their family who had to cope with being visually impaired (VI), comparing the process of finding out about and accessing support to a battle. In contrast, one participant with familial aniridia described being "native" in terms of his awareness of available support.

More broadly, participants with sporadic and familial aniridia highlighted that their families were very valuable sources of support, both practically and emotionally. However, some participants felt a need to protect their family members from how difficult they felt emotionally, and others described sometimes feeling envious of other family members such as parents who had aniridia but had better sight sometimes, meaning that there wasn't an entirely straightforward relationship between participants and their families.

Unfortunately one common thread amongst participants was how inadequate existing support was felt to be, particularly financial support and emotional support. Many participants had found the accessing financial support, such as Personal Independence Payments, time consuming and incredibly emotionally challenging as a process. Whilst counselling was thought to be potentially of great benefit for participants, too often it was very limited, either in terms of the structure or level at which it was available, or in terms of counsellors that lacked skills, experience or empathy when supporting people with a VI.

Many participants felt that their most useful source of support was the VI community. Peer support was particularly felt to be invaluable on a practical level, through exchanging advice or ideas about managing sight loss, and on an emotional level in terms of the feeling of solidarity people felt.

5. Conclusions

In conclusion, this research project contributed new findings to three different areas of understanding: the psychosocial impact of aniridia as a condition, the psychosocial impact of deteriorating vision, and the varied experiences of those with sporadic and familial forms of a genetic condition. The study found that although there are some similarities between aniridia and other genetic eye conditions, what makes aniridia distinct is the uncertainty of its progression, and the complexity of treatment of further causes of sight loss. These can lead to increased anxiety, and potentially demands constant adjustment of an individual to their varying sight. Support, especially from family members and others in the VI community, can be of great benefit, though the experiences of those with sporadic and familial aniridia can differ significantly in their awareness of and access to support.

The insights of this study have implications for counselling, genetics and ophthalmic professionals working with people with aniridia. Of particular importance are increased awareness of extra information and support for people with sporadic aniridia, and the need to acknowledge the challenging nature of further sight loss, given the unpredictable progression and treatment complexities that aniridia can result in.

6. Acknowledgements

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The full dissertation manuscript can be obtained by request from Cardiff University Library service. Email: library@cardiff.ac.uk.

7. References

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