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"Adrift From the World": Exploring the Lived Experiences of Individuals Affected by an Inherited Optic Neuropathy in the United Kingdom—A Qualitative Study

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ABSTRACT

Objectives: Little is understood about the lived experiences of individuals affected by inherited optic neuropathies (IONs) in the United Kingdom. The aim of this study was to understand how autosomal dominant optic atrophy (DOA) and Leber hereditary optic neuropathy, the 2 more commonly encountered IONs, impact affected individuals and the factors contributing to their vision-related quality of life (VRQoL).

Methods: Semistructured qualitative interviews were conducted with 20 individuals with a genetic diagnosis of DOA (10 participants) or Leber hereditary optic neuropathy (10 participants) and affected by vision loss. Eligible participants were purposively sampled to achieve variation in participant age, sex, duration of visual impairment, and location in the United Kingdom. Using inductive thematic analysis, a range of themes and subthemes were developed.

Results: Participants' experiences could be broadly summarized across 4 overarching themes: (1) IONs affected all aspects of life, most notably psychosocial and emotional well-being; (2) participants learned to cope by adapting and adjusting to visual impairment, often on their own, with little external support or resources; (3) participants' identities as visually impaired people were determined by how they viewed themselves and others' reactions to their disability; and (4) good VRQoL was defined as having independence with the support of others.

Conclusions: Visual impairment due to an ION threatens the independence of affected individuals, leading to psychosocial losses and reduced emotional well-being. Despite the challenges they face, people living with an ION describe a "relatively" good VRQoL, often because of the positive impact of social support, enabling them to lead fulfilling lives.

Keywords: lived experience, mitochondrial disease, optic atrophy, quality of life.

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Highlights

- The vision-related quality of life (VRQoL) of individuals affected by inherited optic neuropathies (IONs) has previously been described using generic patient-reported outcome measures focused on visual functioning, a measure of activity limitation and participation restriction.
- Visual impairment caused by IONs impacts on all aspects of life, notably psychosocial functioning. Contextual factors contribute significantly to self-reported VRQoL. Positive interpersonal relationships and social supports enable affected individuals to maintain independence and contribute to their VRQoL.
- The impact of vision loss caused by IONs affects 3 domains: physical health (symptoms and visual functioning), mental health (cognition, affect, and behaviors), and social health (social function and relationships). Interventions for IONs should be evaluated on the impact they have on these 3 VRQoL domains.

Introduction

Inherited optic neuropathies (IONs) are a diverse group of rare eye diseases that result in significant visual impairment and reduced health-related quality of life (HRQoL), particularly in children and young adults.[1] The 2 most common IONs are dominant optic atrophy (DOA) and Leber hereditary optic neuropathy (LHON), with an estimated prevalence of 1 in 25 000 and 1 in 30 000, respectively.[2,3] Visual loss in DOA begins in the first 2 decades of life and declines steadily. In contrast, the peak age of onset of LHON is between the ages of 15 and 35 years. People affected by LHON develop devasting visual loss beginning usually with one eye, followed by the second eye weeks to months later. The long-term visual prognosis in both conditions is poor, with most patients meeting criteria for legal blindness.[3,4]

Gene therapy has emerged as a promising treatment strategy, particularly for LHON.[5] Recent clinical trials in LHON have used measures of visual functioning, a measure of disability or limitation associated with vision-dependent tasks, to determine the impact of gene therapy on vision-related quality of life (VRQoL).[6-9] VRQoL is considered a subset of HRQoL, focusing on how visual ability and symptoms affect day-to-day function and overall well-being. Little is known about the lived experiences of individuals affected by IONs and how they perceive their VRQoL to be affected by living with the condition.[10] Previous studies assessing the VRQoL of affected individuals have primarily focused on activity limitations caused by vision loss, often using patient-reported outcome measures (PROMs), such as the Visual Function Index, a 14-item questionnaire developed for people with cataracts,[11-14] and symptoms of anxiety and depression after vision loss.[12,15]

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The ophthalmology literature provides rich evidence of the impact that sight-threatening diseases can have on psychosocial well-being, especially in young adults. Diseases such as inherited retinal degenerations and uveitis affect educational attainment, employment opportunities, and social participation.[16-18] The broader psychosocial consequences of visual impairment may not be fully captured by functional vision assessments alone.

The aim of this qualitative study was to explore the lived experiences of individuals affected by DOA and LHON in the United Kingdom. To inform the development of a new ION-specific PROM, the objective of this study was to understand how IONs impact affected individuals and to identify areas to improve patient care. A secondary objective was to build a conceptual framework for how VRQoL could be measured in people with IONs.

Methods

The FDA Patient-Focused Drug Development guidance was used to develop study methods.[19-22] Potential participants were identified from 3 specialist clinics for people with mitochondrial diseases or IONs in Cambridge (Cambridge University Hospitals) and London (Moorfields Eye Hospital and the National Hospital for Neurology and Neurosurgery). Inclusion criteria required participants to be aged ≥16 years and visually affected by (a genetically confirmed) diagnosis of DOA or LHON. Given the rare prevalence of IONs, an initial sample of 20 participants was determined from a practical standpoint to achieve maximum variation in terms of participant characteristics. To provide the greatest source of information power,[23] eligible participants were purposively sampled to achieve variation in age, sex, ethnicity, duration of visual impairment, and location in the United Kingdom.

Qualitative interviews were conducted on Zoom (Zoom Video Communications, Inc, San Jose, CA, USA) or telephone and lasted approximately 60 minutes. All interviews were led by the first author (B.C.) and supported by 2 other authors (C.S., J.F.), following a semistructured format to encourage discussion of participants' experience of living with an ION, how their condition affected daily living and activities, and the QoL domains that were affected by their condition. The interview guide was developed with an advisory group consisting of affected individuals and their families, representatives from the Cure ADOA Foundation and UK LHON Society, and researchers with experience in PROM development and was piloted before use with study participants (see Appendix 1 in Supplemental Materials found at https://doi.org/10.1016/j.jval.2025.07.023).

Analysis

Interviews were audio-recorded and transcribed verbatim. All participants were provided a copy of their interview transcript and invited to check the accuracy and provide additional post-interview information. Transcripts were imported into the qualitative software package NVivo 12 Pro (Lumivero, Denver, CO, USA) and analyzed by reflexive thematic analysis after completion of all 20 interviews.[24,25] Separate independent coding of 3 transcripts (B.C. and C.S.) allowed initial inductive code generation and facilitated cross-check capture of codes. Complete coding was conducted for all transcripts (B.C.), with the final 2 transcripts checked again for consistency and data saturation (C. S.). Candidate themes and related subthemes were identified inductively by grouping codes into related concepts and observing clusterings of codes around central organizing concepts across the entire data set. Themes were reworked and then refined independently by 2 authors (B.C. and C.S.), who then defined the final themes in discussion with the members of the advisory group.

A conceptual framework for describing the impact of IONs on VRQoL was developed in accordance with FDA guidance.[20] The final themes and subthemes were organized into a preliminary model outlining key concepts and their hypothesized relationships to VRQoL. Similar concepts were grouped into potential domains and subdomains, drawing on both the interview data and relevant preexisting frameworks used in vision-related and chronic disease research.

Results

A total of 10 participants with DOA and 10 with LHON were interviewed (Table 1). All participants were White British, except 1 participant of mixed-race heritage. Participants with DOA tended to be older than participants with LHON. Duration of visual impairment varied across interview participants. All participants with DOA had visual impairment of at least 5 years, whereas some participants with LHON were still within the first 2 years of vision loss. Four themes were identified across the complete data set (Fig. 1).

Theme 1: Impact of ION

The impact of IONs was experienced in all aspects of life, creating challenges with vision-related activities and a diverse range of emotional and psychological responses.

Subtheme 1: Psychosocial and emotional losses

IONs principally affected participants' ability to obtain information, fulfil responsibilities at home and in the workplace, and pursue social and leisure activities. Participants described feeling isolated, left behind, or a burden on others (Table 2A).

Participants spoke about losing their freedom or independence and the difficulty of behaving how society would expect someone of their age, gender, or status to behave (Table 2B). The inability to recognize faces or facial expressions often made participants feel

Table 1. Demographic and clinical characteristics of 20 study participants.

Characteristic	DOA (*N* = 10)	LHON (*N* = 10)
Sex		
Female	4 (40%)	4 (40%)
Male	6 (60%)	6 (60%)
Genetic diagnosis		
OPA1 variant	10 (100%)	-
m.11778G>A (*MT-ND4*)	-	6 (60%)
m.14484T>C (*MT-ND6*)	-	1 (10%)
m.3460G>A (*MT-ND1*)	-	3 (30%)
Age, years		
Median (range)	51 (20-60)	28 (16-76)
16-18 years	-	3 (30%)
19-25 years	3 (30%)	2 (20%)
>25 years	7 (70%)	5 (50%)
Duration of visual loss		
<2 years	-	2 (20%)
2-5 years	-	4 (40%)
>5 years	10 (100%)	4 (40%)

Note. Values presented as *n* (%).
DOA indicates dominant optic atrophy; LHON, Leber hereditary optic neuropathy.

embarrassed in social settings, especially when asking for help or being confronted by others for appearing different (Table 2C).

Female participants frequently reported feeling guilty about the possibility of passing or having passed a genetic mutation onto their children, and the challenges that visual impairment had on their ability to be a parent (Table 2D):

Critically, participants felt that little was offered by the health system to help with the psychological and emotional impact of living with an ION, with many having to figure things out for themselves to survive. Participants used self-help strategies, sought out professional help by themselves, and highlighted the positive impact of community support and patient advocacy groups (Table 2E).

Subtheme 2: Living under a cloud of uncertainty

Because of the lack of information and few opportunities to interact with other affected individuals, participants expressed concerns regarding their visual prognosis and the impact of further vision loss on their livelihood. This concern was most pronounced in participants with better vision (Table 2F).

For younger participants, uncertainty about not having family members to support them or having to live independently was worrying. Older participants focused more on the threat of giving up driving, finding new employment, retiring early, or altering retirement plans (Table 2G).

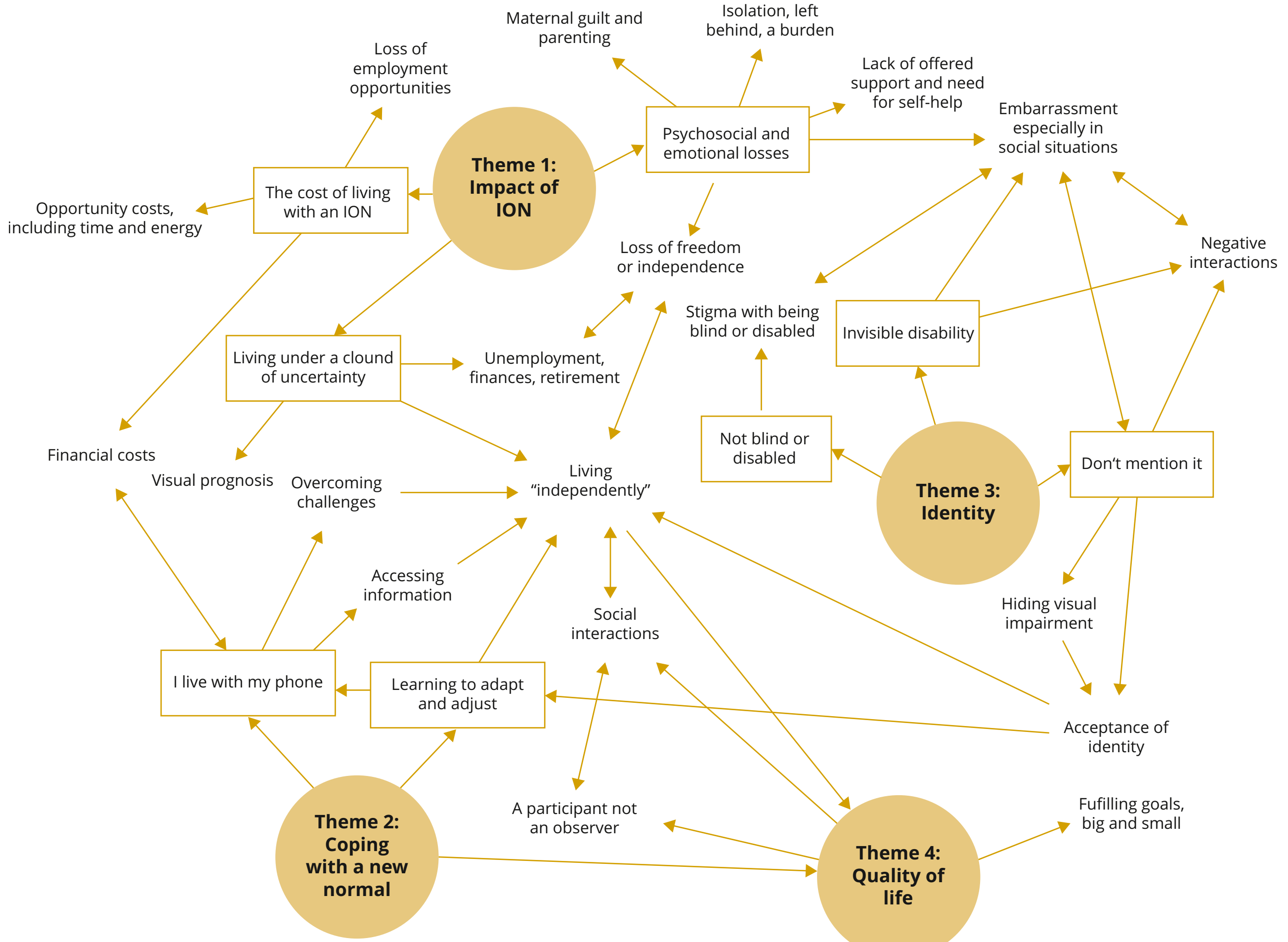
Subtheme 3: The cost of living with an ION

Living with an ION came with many costs. Participants found that low-vision aids were cumbersome and expensive to purchase. Necessary adjustments, such as traveling on public transport or using online grocery shopping imposed additional costs. Although some of this was offset by disability benefits or allowances, not all participants were able to access these consistently across the United Kingdom (Table 2H).

Many participants were concerned by the loss of employment opportunities, loss of earning potential, and the threat of early retirement affecting their finances. Some participants had to stop working altogether because driving was an integral part of their occupation (Table 2I).

Opportunity costs were also evident, most notably time and energy. Participants spoke about living a structured routine, having to plan ahead, and needing more time to complete tasks or activities. This meant that participants tended to stick to the same routine in familiar environments, with the people who were able to support them. Participants emphasized the loss of spontaneity and ability to do things on their own (Table 2J).

Figure 1. Thematic map summarizing the lived experiences of people affected by IONs. Four major themes (black circles) and subthemes (white rectangles) are connected conceptually by keywords and key phrases (borderless text).



IONs indicates inherited optic neuropathies.

Table 2. Participant quotations representing major themes and subthemes.

Themes	Subthemes	Quotations
Impact of inherited optic neuropathies	Psychosocial and emotional losses	(A) Marjorie, 76 years old, LHON Socially, emotionally…being adrift from the world, I think, is quite a useful phrase. The world is going on around you, but you're not quite in it […] or the world is going on around you, and your opportunities for engaging with it […] you have to make much more of an effort to join into things. (B) Janine, 43 years old, LHON: [Describes holiday] And we used the buses a lot and I would always have to…I had him with me but otherwise I would always have to say to somebody, what number is this please or what number is this train or even what platform do I need to be on, I would have to ask somebody to take me to the right platform to get on the train. It is the same thing, it always stems back to the independence, doesn't it, and not feeling like you're functioning as a proper adult should be doing. (C) Natalie, 24 years old, DOA: Or people think, you know, like I'm staring at them in a rude way or whatever when I'm not, I just physically can't see them, like I can't see their facial expressions, I can't…do you know what I mean, so then people get the wrong impression, or the wrong idea about me […] And that's, obviously, quite embarrassing or, you know, it just doesn't feel very good. […] And it's so embarrassing especially when I'm walking down…not with my friends, you know, that's fine, they understand, but say like if you just see someone you just know, walking down the street and they're like smiling at you or whatever and you don't realise until they're right in front of you and by that point they've passed you. And then you just like you've been well rude, but you actually haven't I just didn't see them. (D): Fran, 17 years old, LHON: […] I think there's obviously a bit of an element of guilt with it because I obviously inherited it from one of my parents so then there's that sort of, you know, my mum in a way feels like it's her fault, which it obviously isn't, it's genetic so you can't do much about it. But I think that that probably plays into it a little bit, and then also, just things I wouldn't notice at the time but looking back on, it feels like they were a lot more lenient with certain things. […] (E): Grant, 55 years old, LHON: For a long time, when I was given the diagnosis […] I went through the stage of seeing a lot of medical professionals, and then, it got to the stage where, right, that's it, I don't really need to be reviewed every year. This is it. And I don't think I saw an eye specialist for a long, long time; many, many years. I've been back in touch with the RNIB and Sight Support, the local sight support charity and the Guide Dog Association. […] I am sort of back in the loop of adaptation, needing support and adapting once again. […] I think, for me, until recently, and I have to emphasise recently, I didn't feel that I was given a lot of information about LHON and I didn't really know how to find it out myself. And, now I feel a lot more comfortable because I do have that information. And, I do, you know, I have become aware of the LHON Society quite recently and, you know, I do read their information and I've found that really useful.
	Living under a cloud of uncertainty	(F) Sally, 20 years old, DOA: Yeah, I suppose I also have this thing of will someone want to date me because of it. And also, the fact that because it's genetic, do I want children, how will I go about that? Will my vision get a lot worse? Do I want to travel now so I can see everything, so later in life I think, oh no, do I need to do everything now before it gets worse because it's so uncertain. (G) Phillip, 50 years old, DOA: […] I do worry anyway as to how long I've actually got left of my working life. Obviously, you've interviewed my brother, and he's several years older than me, and no longer works. As soon as you get to that stage, you're thinking, how long have I got left to pay the mortgage, how long have I got left to assist with the children that are still growing up. It becomes very scary, and you start to have to plan for what you hope is worst case scenario. But of course, you might be going on a lot longer in terms of your working life. Now, at the moment, I'm actually thinking, at 55, I may not be able to go and do a full week's work. And that's what I've got to plan round.
	The cost of living with an ION	(H) Janine, 43 years old, LHON: Yeah, so I use that and I use CCTV magnifier, as I say, I have one at home and one at work. But that's for reading and writing underneath because I can…I read about probably newspaper headline size print. So I can use that for reading and writing and looking at recipes or instructions on anything. So that's fine. But again, it's restricted because I've got that at home, so then they're…I mean, you'll have seen, I suppose they're not something you can carry around with you because they're huge. […] It's not something you can put in your bag. And expensive. The one I'm using at home, I think it's getting a bit temperamental and I'm thinking, oh, don't go because they can be like…they're over a thousand pounds. […] And you don't get help with them, you have to buy it. You get help with the one from work and as I say, I work for local government so they can afford to buy you them. But if you worked for a smaller private company, they're not funded. (I) Alan, 52 years old, DOA: [Discussing previous job as a taxi driver] Yeah, it was quite depressing at the time like, 'cause I had to stop work and stop driving. Obviously, like there's no money coming in, well my wife works, but she was only part time and that, so that was quite challenging, yeah and benefits and things like that, but yeah, it was quite a depressing time actually for about six months. (J) Grant, 55 years old, LHON: […] the biggest thing I find is you have to have structure in your life when you've got a disability or something, LHON; in my situation I have to have structure, I have to plan ahead […] And it's like…the ability to just be able to do things on the spur of the moment is absolutely out of the question.

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Table 2. Continued

Themes	Subthemes	Quotations
Coping with a new normal	Learning to adapt and adjust	(K) Angela, 48 years old, DOA: […] And they're concerned that I'm on the verge of not able to drive, and that's my main worry for my eyesight is not being able to drive, because I rely on that going to and from work, taking my children to different activities. But up until last year when I got the blood results, I just thought I had poor eyesight, I didn't know it was hereditary. (L) Katrina, 60 years old, DOA: When this first happened, I lived in Scotland, and I lived in the middle of nowhere and I absolutely loved it. I could drive to wherever I wanted to be. When this happened, a few years afterwards we moved. We specifically chose this little town, [name of town], because it has the tiny, little train station that's walkable. So I live in the town centre specifically, and that little train station takes you either to [name of city] which is the fast train to London, or one line goes all the way to Birmingham, to Birmingham International Airport, and the other one goes to Stansted Airport. So from this little town I can go anywhere in the world, myself, completely independently without needing a lift anywhere. That's important to me.
	I live with my phone	(M) Philip, 50 years old, DOA: […] I can't read menus in lots of restaurants, my mobile phone becomes a magnifying glass. I use the camera and I pinch zoom. I'm doing that all the time. In lots of different ways. In fact, to preparing a meal in the kitchen. If I buy a packet meal that's got cooking instructions, I can't see those, so the phone becomes a magnifying glass. I use it day in day out like that.
Identity	Not blind or disabled	(N) Bruce, 32 years old, LHON: […] Like there really is not a lot of help at all and when you do get benefits, you feel like a bad person, you're like, oh, should I be having this? I know I'm blind but I'm still not physically impaired. Should I be at work? Should I be doing something? You do feel bad for having what you're allowed to have.
	Invisible disability	(O) Simon, 54 years old, DOA: [Talking about vision at work] It's a bit irritating really, because from other people's perception, it's like there's nothing wrong. It's a bit like a hidden disability. [Talking about reactions from other people when he has had to explain his situation] Some people are a bit dismissive, as if you're just sort of, you know, moaning about nothing. Other people just don't understand it. And then other people are quite interested and want to know more, and then they sort of get it. […]
	Don't mention it	(P) Daniel, 23 years old, LHON: Aye, I'd be worried asking strangers, now. But it's just, whenever people would say something to you, like, if they see you holding the phone up close to you, and they say, jeezo, look how close you're holding that phone. And it's embarrassing, like. (Q) Gregory, 23 years old, DOA: [Feelings when talking about diagnosis] No. I don't feel embarrassed by it. I would only feel embarrassed if people start, like, reacting strangely to what I say, and then I think, then again it's just one of those things that's more of a reflection of them than of me.
Vision-related quality of life		(R) Nathan, 17 years old, LHON: I think being able to just do things that you want to do with relative ease. Like, being able to do things that you want to do and not having to hold you back by too much. (S) Christopher, 58 years old, DOA: And then social interaction as well, so I've got not a huge network of friends but, you know, friends I can count on and discuss things with. And it's that social interaction, I think, that is probably the most important thing both at work and socially. This is where I think, you know, anybody with a disability of any sort, for me, I mean I'm sure there's probably been lots of literature written about this, but I suspect that those who cope best with their disability are the ones with a good supportive network of friends and family. Who treat them as they would want to be treated. And those who don't have that will…yeah, will not have such good quality of life or well-being.

Theme 2: Coping With a New Normal

Participants demonstrated a resolve to cope by adapting and adjusting to a new life with visual impairment, often with little structured support, training, or education. Adapting and adjustment to visual impairment meant developing new or different skills, learning to use an aid, advanced preparation or planning, finding a different approach to a problem, and becoming confident in asking for help.

Subtheme 1: Learning to adapt and adjust

The rapidity of vision loss in LHON meant that participants had to adapt and adjust to significant visual impairment, while navigating a period of emotional devastation and grief. For participants with DOA, a history of vision problems from childhood often necessitated small adaptations or adjustments. Concerted efforts to adapt to significant visual impairment were prompted by the diagnosis of DOA being confirmed; deterioration in vision exhausting previous adaptations, or the threat of not being able to drive (Table 2K).

Multiple factors contributed to successful adaptation and adjustment, including younger age at onset, better residual vision, and access to opportunities. The latter was particularly important, with participants living in metropolitan regions or areas with established transportation links afforded more opportunities (Table 2L).

Subtheme 2: I live with my phone

All participants utilized assistive technology to some extent in their daily lives. For most participants, their mobile phone became an essential aid for overcoming many daily challenges. Photographing and zooming in on an image was a simple but effective strategy that nearly all participants reported doing. Mobile phones and computers also enabled participants to access information (eg, looking up train or bus timetables) and provide an alternative solution for challenging tasks (eg, online grocery shopping instead of looking for price tags) (Table 2M).

Theme 3: Identity

Participants' identities as visually impaired people were determined by how they viewed themselves and how others responded to them.

Subtheme 1: Not blind or disabled

All participants had some degree of residual vision and often did not label themselves as "blind" or "disabled" because both terms were perceived as negative and associated with stigma. Although many participants were registered with a Certificate of Visual Impairment (CVI) as sight impaired or severely sight impaired, as per the Royal National Institute of Blind People criteria, some felt guilty about accessing or using services (Table 2N).

Subtheme 2: Invisible disability

Participants spoke about the general public's lack of awareness or tolerance of people with invisible disabilities. With their residual vision, they could still function independently. However, in situations when they needed others to be considerate or when requesting help from others, disclosing their visual impairment resulted in a range of responses, including unnecessary sympathy, others being overly helpful or condescending, and even their visual impairment being challenged or not being believed (Table 2O).

Subtheme 3: "Don't mention it"

Many participants concealed their visual impairment by trying to appear or behave as "normal" to avoid drawing attention to themselves and avoid the stigma associated with being blind or having a disability. Participants spoke negatively about being treated differently, experiencing uncomfortable social interactions, or receiving unwanted questions or attention (Table 2P).

Participants chose to reveal their visual impairment when they required information, help, or consideration from others. Rejecting negative attitudes about visual impairment, especially external societal attitudes, appeared to be a substantial contributor to success in adapting and adjusting (Table 2Q).

Theme 4: What Is Good VRQoL?

For most participants, good VRQoL meant having the independence to do what they liked, when they wanted, enabling them to fulfil their goals, eg, provide for their family, enjoy leisure pursuits or hobbies, have meaningful social relationships, or achieve a career goal (Table 2R). Despite the challenges they faced, participants still reported that their VRQoL was good. The most important factor contributing to VRQoL was social interactions. Participants spoke about the family members, friends, and colleagues in their lives and the impact these individuals had on improving their VRQoL. Having a network of support around participants enabled them to fulfil the goals they had, within reason, and enabled them to be an active participant in the world around them rather than a passive observer (Table 2S).

Discussion

The aim of this study was to explore the lived experiences of individuals living with an ION in the United Kingdom to better understand the impact of DOA and LHON on affected individuals. Participants in this study reflected on the impact of visual impairment around specific examples of activity limitation or participation restriction in their day-to-day lives. However, the discussions framed the effects of these limitations and restrictions on psychosocial functioning and emotional well-being. Key issues explored by participants included the loss of social skills, identity, status, and roles in society, and descriptions of feeling embarrassed, guilty, isolated, a burden on others, and left behind. Living independently with the support of others and having positive interpersonal relationships were important factors affecting participants' appraisal of their VRQoL.

Our findings are consistent with Ferguson and de Abreu,[26] who found that psychosocial losses associated with visual impairment were a key aspect of the lived experiences of men living with LHON in the United Kingdom. The onset of visual impairment disrupts how a person feels about themselves and their ability to independently act within the world, evoking frustration, stress, and reduced confidence and self-esteem. The experiences of study participants also mirror those with other inherited eye conditions. Despite being a diverse group of conditions, many inherited eye conditions are early onset, untreatable, progressive, and result in significant visual disability.[27] Disease-related factors and cultural factors modify the impact of inherited eye conditions, including psychological well-being (mental health, coping, and identity) and social well-being (relationships, socioeconomics, stigma, and independence).[27] Indeed, the World Health Organization's biopsychosocial model of disability recognizes that impairment, activity limitation, participation restriction, and the interactions between these components are moderated and mediated by contextual (environmental and personal) factors.[28] Contextual factors may be responsible for some individuals with impairment developing activity limitation or participation restriction and, in turn, poor HRQoL.[29]

In this study, contextual factors appeared to moderate the experiences of participants and their perceived VRQoL. Participants who coped well with visual impairment tended to be in environments that enabled them to derive a sense of meaning, purpose, or direction in their life, and have positive meaningful connections with others. These findings are consistent with those of an earlier study exploring emotional response to vision loss in individuals living with LHON.[15] Four contextual factors (work, education, romantic relationships, and family) were identified as determinants of recovery from initial sadness after vision loss. When vision loss resulted in loss of relationships, independence, and work or educational training, the extent and duration of sadness after vision loss was considerably worse.[15]

This study underscores the impact that societal attitudes and beliefs have on the ability of visually impaired people to successfully adjust to their disability.[30] Many study participants described feeling uncomfortable using aids (eg, a white cane) or assistive technology in public or avoiding interactions with strangers even when they required assistance, due to negative beliefs about visual impairment. Buying into negative societal beliefs can detrimentally shape their hopes, aspirations, and expectations, and can inhibit their ability to experience a healthy, well-integrated life.[31] Participants also held dual identities: a hidden and frequently concealed identity as a visually impaired person requiring others to be mindful and considerate, and a "normal" identity to avoid the stigma associated with visual impairment. Applying Ryff's model of psychological well-being to the experiences of participants in this study,[32] conflict between the 2 identities adversely affects autonomy and self-acceptance. Indeed, participants who were less concerned by the negative reactions of others when seeking assistance or using assistive technology or aids, demonstrated high levels of autonomy and self-acceptance and tended to have better environmental mastery.

Practical Implications

Several factors provide opportunities to improve the ability of individuals affected by IONs to cope with visual impairment and to enhance VRQoL after a diagnosis of DOA or LHON. Personal factors are important in determining how an affected individual copes with visual impairment. Interactions with health care professionals at the time of diagnosis, in particular the way that the diagnosis is delivered and the information that is provided, can have a lasting impact on affected individuals and how well they are able to come to terms with their visual impairment.[33] Provision of information about the condition and prognosis, no matter how negative, is important for affected individuals.[33]

Similar to other rare diseases, there is a lack of standardized, coordinated care for people living with IONs,[34] including referral to low-vision services, sensory support services, and voluntary organizations or charities, and navigating the welfare benefits they are eligible for with CVI registration. Access to these services provides an opportunity to address the barriers or challenges that prevent individuals from maintaining gainful employment, education or training, or fulfilling social roles and responsibilities. In the United Kingdom, Eye Clinic Liaison Officers (ECLO, also known as sight loss advisors) often take on the role of care coordination by streamlining the process of CVI registration; improving access to relevant support services, by referring and signposting appropriately and according to the needs of patients; and by providing emotional and practical support.[35] Furthermore, ECLO have an important role in care continuity by providing follow-up beyond the medical clinic,[35] particularly because individuals with IONs are frequently discharged from the eye clinic by their ophthalmologist because nothing can be done for them clinically.

Supporting affected individuals with the psychological and emotional impact of ION is critical and inadequately addressed by existing services. ECLO have a crucial role in providing emotional support to individuals affected by visual impairment.[35] However, ECLO services vary across the UK regions, with the lowest availability in England and significant delays from diagnosis to first visit.[36] Emotional support and counseling service may also be delivered within an integrated low-vision service, but low-vision practitioners alone are not enough to provide the required psychological support for visual impairment.[37] This study also highlights the vital role of voluntary organizations and charities in supporting people living with IONs and their families, in particular organizations such as the Cure ADOA Foundation and UK LHON Society. These organizations allow affected individuals to feel part of a community and gives them hope.

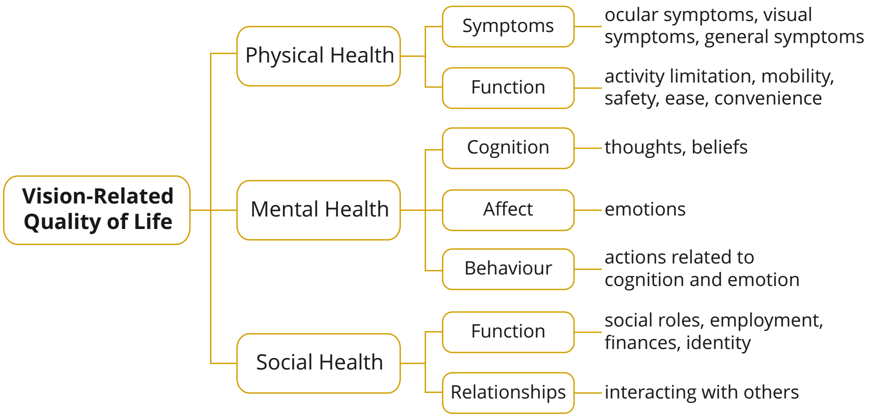
Finally, negative societal beliefs about visual impairment and disability still exist. Although the Equality Act (2010) was designed to protect people living with a disability from discrimination and to create a more equal society, people living with visual impairment, continue to face direct and indirect discrimination due to the invisible nature of their disability.[38] Recent campaigns, particularly during the coronavirus pandemic, such as the Hidden Disabilities Sunflower, have raised awareness of invisible disabilities,[39] but much more can be done to challenge perceptions and stereotypes of what it means to be visually impaired, in particular, what a person with visual impairment looks like.

Developing a Conceptual Framework for Assessing Vision-Related QoL in IONs

Measures of VRQoL must provide a complete assessment of the impact of IONs and the effectiveness of treatment on critical components of quality of life.[40] VRQoL is a complex trait that encompasses domains that are important to people affected by visual problems. In ophthalmology, a set of 10 quality-of-life domains have been identified previously as being important to patients with eye conditions, including activity limitation, mobility, visual symptoms, ocular surface symptoms, general symptoms, emotional well-being, social participation, economic, health concerns, and convenience.[41,42] In our study, visual symptoms, ocular surface symptoms, and general symptoms were infrequently discussed by participants, whereas the impact of IONs on activity limitation, emotional well-being, and social participation were discussed in detail.

Adapting the Patient-Reported Outcomes Measurement Information System (PROMIS) Domain Framework[43] and the psychosocial model previously described by D'Amanda et al,[27] we conceptualize the impact of vision loss caused by IONs as affecting 3 core domains: physical health, mental health, and social health (Fig. 2). Our conceptual framework aligns with the

Figure 2. Conceptual framework for vision-related quality of life in IONs. Self-reported vision-related quality of life in people affected by IONs is reflected across 3 core domains (physical health, mental health, and social health), each consisting of several subdomains. Constructs related to each subdomain are presented to the right of the figure.



IONs indicates inherited optic neuropathies.

World Health Organization's definition of health as the "state of physical, mental, and social well-being and not merely as the absence of disease and infirmity."[44] The physical health domain includes [physical] function (eg, activity limitation or difficulty with mobility) and disease-related symptoms (eg, glare and light sensitivity). The mental health domain captures psychological and emotional factors related to individuals' thoughts and beliefs (eg, feeling like a burden), affect (eg, feeling embarrassed or low), and behaviors (eg, reluctance to ask for help or use a white cane). The social health domain includes the impact of IONs on social function (eg, social roles and responsibilities, self-identity, and finances) and relationships (eg, interacting with others).

From a practical standpoint, an individual with a new onset ION might have a poor self-reported VRQoL due to negative attributes in all 3 domains as they learn to cope with "a new normal." However, an individual with chronic ION, who has adapted well to vision impairment, might report an excellent VRQoL because of positive attributes in the mental health and social health domains, despite having low vision and significantly impaired function on the physical health domain. The development of an ION-specific PROM that assesses the 3 domains would enable the impact of new therapies to be assessed comprehensively.

Strengths and Limitations

A key strength of this study was the use of in-depth interviews with participants who varied in age and time since diagnosis. To improve qualitative research rigor, participants were sampled purposively to obtain a diversity of patient voices and to improve information power. The reflexivity of the researcher plays a critical role in both the collecting and analyzing of the data. Although the lead researcher conducting the interviews and initial analysis (B.C.) was a fully sighted male clinician with knowledge about IONs, the interviews and analysis were conducted with the support of a female nonclinician without prior knowledge of IONs (C.S.), and a male lay member of the research team affected by LHON (J.F.). Involvement of multiple team members from different backgrounds helped us to avoid introducing a clinical bias and be open to uncertainty and adaptability within the analytical process.

A limitation of this study is its generalizability. Participants were recruited from clinics at large tertiary hospitals. Patients seen at tertiary hospitals may have more complex disease necessitating regular clinical follow-up, or they may be more motivated to participate in research because they have a particular viewpoint or perspective. Another limitation was the underrepresentation of black and minority ethnic populations, who have historically experienced poorer health and barriers to accessing health services. Further research is required to determine the generalizability of our findings to an ethnically diverse UK population.

Conclusions

Visual impairment due to an ION threatens the independence of affected individuals in all aspects of their lives, leading to psychosocial losses and reduced emotional well-being. The provision and use of assistive technology and aids eases the activity limitation and participation restriction caused by visual impairment. However, these are not always useful or effective and frequently come with financial and opportunity costs. The impact of visual impairment on the ability of affected individuals to fulfil previously defined social roles and responsibilities adversely affects their self-identity. This is further aggravated by social beliefs about visual impairment and the negative attitudes of sighted people. Despite the challenges they face, people living with an ION in the United Kingdom describe a relatively good VRQoL, often because of the positive impact of social supports allowing them to lead fulfilling lives. The psychological well-being of people living with an ION can be further enhanced by provision of accurate information about the condition, including prognosis, and improving coordination of care after diagnosis, including early proactive referral to support services, CVI registration, and provision of emotional support.

Author Disclosures

Author disclosure forms can be accessed below in the Supplemental Material section. The views expressed are those of the author(s) and not necessarily those of the NHS, the NIHR, or the Department of Health.

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