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Navigating a hidden disability: Lived experiences and challenges of adults with early stage inherited retinal diseases

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ABSTRACT

Background: Inherited retinal diseases (IRDs) are genetic conditions that typically cause vision loss in working-age adults, representing a unique hidden disability characterised by variable progression rates.
Objective: This qualitative study explored the lived experiences of adults in the early stages of IRDs, when vision loss is not outwardly apparent.
Methods: Semi-structured interviews were conducted with 15 individuals with IRDs (mean age 37 ± 17 years) with experiences of progressive vision loss in the last 10 years, exploring participants' experiences and challenges following their IRD diagnosis. Interviews were transcribed verbatim and analysed using thematic analysis method.
Results: Analysis yielded five overarching themes: 1) Adapting to the diagnosis: "The journey can be harder than the actual disease", describing challenges in coming to terms with the diagnosis. 2) Daily obstacles: "Accepting what I have, adapting where I can", describing lifestyle and behavioural changes to accommodate for changing vision. 3) A roller-coaster of emotions: "I feel like I'm not in control of the journey", highlighting emotional challenges managing the uncertainty of a variable, progressive disease. 4) Navigating society: "it's invisible, so people forget", capturing interpersonal challenges stemming from a hidden disability. 5) The road ahead: Finding an identity within uncertainty, describing struggles with identity and the future.
Conclusion: Beyond vision impairment, individuals with IRDs face numerous personal and interpersonal challenges due to the hidden nature of their disability. These challenges are not always immediately apparent, highlighting the importance of raising awareness to assist in developing targeted resources, diagnostic support, and broader societal understanding for hidden disabilities.

1. Introduction

'Hidden disability' refers to a disability that interferes with an individual's daily functioning, yet is imperceptible to others. Research across several hidden disabilities, such as autoimmune diseases[1] and mental health disorders,[2] has highlighted the psychosocial and practical

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challenges experienced by individuals, and how this is exacerbated when their disability is hidden. This can result in significant burdens from lack of public understanding, challenges accessing support and accommodations, barriers to proper diagnosis and treatment, and psychological impacts.[2,3]

Inherited retinal diseases (IRDs) represent a broad group of rare, genetic eye diseases that collectively affect around 1 in 2000 people.[4] Initial symptoms can include night blindness, tunnel vision, and central vision loss. Symptoms of IRDs typically begin in adolescence, and most IRDs cause progressive vision loss, but with substantial variation in progression rates, even among individuals with the same condition.[5] Vision loss can encompass a spectrum of impairment. Clinically, mild visual impairment is defined by the World Health Organization as visual acuity better than 6/12 (or 20/40), with many individuals still legally able to drive.[6] Moderate visual impairment ranges from visual acuity of 6/18 (or 20/60) to 6/60 (or 20/200). Severe visual impairment meets legal blindness criteria of less than 6/60 (or 20/200) visual acuity, or a visual field constricted to within 10 degrees of fixation. Most people who fit the legal blindness criteria can still see colours, shapes, and varying degrees of light. Less than 2 % of individuals with any level of visual acuity impairment have no ability to see light at all.[7] In the early stages, vision impairment may not be outwardly noticeable, leading to challenges in recognising and acknowledging the condition.

The progressive nature of IRDs means that individuals must continually adapt to their changing vision over time, which can impact various aspects of their lives, including education, employment, independence, and overall quality of life. Public perceptions around vision impairments are also highly emotive and can involve numerous misconceptions around capability, and vision loss has been ranked as equal to or worse outcome than losing hearing, speech, or a limb.[8]

In Australia, IRDs are the leading cause of blindness for working-aged adults[9] with an estimated lifetime cost of AUD $5.2 million, with 87 % being societal costs.[10] Most societal costs are attributed to lower employment rates for individuals with IRD and carers, and increased reliance on social supports.[11,12] While research has explored how vision loss impacts quality of life in individuals with IRDs, affecting emotional well-being, physical functioning, and identity,[13–18] most studies have focussed on the impact of vision loss across various disease stages. There is limited data on how individuals navigate the early stages following an IRD diagnosis when vision loss is not always apparent to others. Additionally, there is a need to better understand how individuals adapt to their progressively changing vision and how the anticipated progression influences their responses to the condition.

Understanding these experiences and unique challenges can assist in developing better resources for diagnostic support, care provision, and broader societal awareness for hidden disabilities. To begin to address this knowledge gap, we aimed to answer the following research questions from the perspective of people with lived experience.

1. What challenges are associated with early, progressive vision loss for individuals with an IRD?
2. What behavioural changes do these individuals make in response to these challenges?

2. Methods

This cross-sectional, qualitative study, broadly following a phenomenological framework, was approved by the University of Melbourne Human Research Ethics Committee (ID: 23825). Study procedures adhered to the tenets of the Declaration of Helsinki, and all participants provided consent prior to participation.

2.1. Participants

Eligible participants were those 18 years or older who i) were diagnosed with an IRD within the last 10 years and/or had experienced vision changes due to a progressive IRD in the past 5 years, ii) speak English, and iii) were able to provide informed consent. We excluded individuals diagnosed with a syndromic IRD (conditions that affect multiple organs or systems in addition to causing vision loss). Participants with a clinical IRD diagnosis were eligible, and they were not required to have a genetic diagnosis or to have had genetic testing.

We recruited participants through the Victorian Evolution of Inherited Retinal Diseases Natural History Registry (VENTURE) who had agreed to be contacted for qualitative and other research studies.[19] An initial email invitation was sent (in April 2022) to 20 individuals enrolled to the VENTURE registry between July and December 2021. We had invited individuals registered to VENTURE at least 4 months prior to the recruitment period to account for potential participation in other clinical studies. No follow-up email was sent. We also recruited through community-based IRD support groups (Retina Australia, Blind Citizens Australia) and referrals from ophthalmology clinics. Using purposive sampling, we recruited participants with varying gender, age at diagnosis, and years since diagnosis to capture a wide range of experiences. As we recruited via the VENTURE registry, we anticipated that of the 20 individuals contacted, ~16 (80 % response) would agree to participate, and 10–15 participants would be adequate to address our research question given the homogeneity of experiences at diagnosis due to a lack of supports and treatments.[20] As data collection and analysis occurred concurrently, the researchers determined that we had reached pragmatic saturation (i.e., that we had enough participants to meaningfully interpret themes related to the purpose of the study) by participant 15.

2.2. Data collection

We collected basic demographics (age, gender, age of onset) and interview data through 1-1 semi-structured interviews from May to November 2022. Participants had the option of conducting their interviews via video conferencing (often preferred for accessibility), via phone call, or in person. Interviewers (BM, with background in social work, and RC, with background in nursing) had experience in qualitative data collection and relevant clinical and disability research, and received input and guidance from author ACBJ with expertise in IRDs and low vision. All participants had the opportunity to review and correct their transcripts.

Our interview schedule (Appendix 1) was developed based on the research aim and designed in consultation with individuals with lived experience of vision impairment from two Australian community organisations (Retina Australia, Blind Citizens Australia). The interview guide was piloted and further refined with a community representative with IRD. During the interview, participants were encouraged to discuss any aspect of their experiences of living with an IRD.

2.3. Analysis

Interviews were audio-recorded, transcribed verbatim using an automated speech-to-text software (Otter.ai, Inc., California, United States) and manually reviewed by an investigator (BM, RC, or EM), who also de-identified all transcripts for analysis. We undertook a reflexive thematic analysis, using an inductive and exploratory coding approach, following the 6-phase guide summarised by Braun and Clarke.[21]

The initial five transcripts were reviewed by two investigators (ACBJ and BM) for data familiarisation and initial observations. These codes were discussed among investigators (ACBJ, BM, EM, RC) to identify potential collective ideas capturing information relevant to our research questions. All interviews were then coded by another investigator (RC). Codes were extracted through inductive reasoning, where data was derived directly from the participants transcripts rather than imposing predetermined frameworks. Data collection and initial coding was done concurrently which allowed for the codes to evolve.

After all interviews were complete, the codes were independently reviewed by our multidisciplinary team to identify, revise, and

consolidate potential themes. The initial codes were independently reviewed by three investigators with eye care clinical backgrounds (ACBJ, EM, BN), who engaged in reflexive discussions to develop the initial themes. The themes were then independently reviewed by a medical clinician (MB) and a behavioural scientist (EGR), who contributed additional perspectives. Throughout the review process, all investigators had access to the de-identified transcripts to verify or refine the codes, ensuring an accurate interpretation of the data. The analysis and development of the themes were refined through several meetings, ensuring a rigorous and collaborative process. Any disagreements were resolved by discussion and consensus. After final review of codes and themes, a coherent interpretation of the data was constructed (ACBJ, MB, EGR). Coding was performed in NVivo 14 (Lumivero).

3. Results

Our sample included 15 participants with an IRD (Table 1). Participants had a range of age at diagnosis (8–65 years) and time since diagnosis (1–25 years). All participants completed their interview via online video conferencing (Zoom). The median interview length was 45 min (range: 20–61 min).

Analysis yielded five themes that captured how individuals with an IRD perceived challenges associated with early-stage progressive vision loss (Fig. 1; Table 1). Additional illustrative quotes are shown in Table 2.

3.1. Adapting to diagnosis: "the journey can be harder than the actual disease"

3.1.1. Shock of initial diagnosis

All participants shared the initial struggle of trying to process their diagnosis. Despite several participants experiencing vision loss for some time before their diagnosis, their initial response to the diagnosis was still shock. Participants commonly expressed high levels of anxiety, despair, loneliness, and feeling like the only person with the condition or a disability. One young person described feeling *"special"* as the only person in their family diagnosed with an IRD, especially when they were young and did not understand the gravity of what the condition meant.

The initial IRD diagnosis often left people grappling with how to manage their situation moving forward. Many people had no prior knowledge and little personal familiarity with IRDs, and reported feeling lost and unsure of what steps to take next after receiving a diagnosis.

> "You've been given this diagnosis, and you're pretty much … standing on the street afterwards thinking … what do we do now? What next?" (24 years old, diagnosed with Stargardt disease)

Table 1
Participant demographics.

Characteristic	Number of participants (%)
Gender	
Male	7 (47 %)
Female	8 (53 %)
Age at interview, mean (SD)	37 ± 17 years
<30 years old	6 (40 %)
30–50 years old	7 (47 %)
>50 years old	2 (13 %)
Age at diagnosis, mean (SD)	28 ± 19 years
<18 years old	6 (40 %)
18–30 years old	3 (20 %)
30–50 years old	4 (27 %)
>50 years old	2 (13 %)
Years since diagnosis, mean (SD)	9 ± 7 years
<5 years	5 (33 %)
5–10 years	5 (33 %)
10–15 years	3 (20 %)
>15 years	2 (13 %)
Inherited retinal disease diagnosis	
Photoreceptor diseases initially affecting peripheral vision[a]	12 (80 %)
Macular dystrophies initially affecting central vision[b]	3 (20 %)

[a] Included rod-cone dystrophy, Bietti crystalline dystrophy, choroideremia.
[b] Included Stargardt macular dystrophy and Best Vitelliform dystrophy.

3.1.2. Denial and avoidance

Many participants recounted avoiding their diagnosis initially, when they could function well without vision aids. Several participants shared that it took several years to process their diagnosis and move beyond this denial or avoidance, and they reflected that this delayed response may have partly been due to their initial IRD symptoms being mild and not disrupting their daily living. Some participants felt that engaging in avoidant responses served as a coping mechanism to alleviate concerns stemming from knowing they will lose vision, yet uncertainty around progression.

> "I think I prefer not knowing how bad it will get over a period of time." (46 years old, diagnosed with rod-cone dystrophy)

3.1.3. "Grieving a life that I wanted to lead"

Participants shared an immense grief for a life they had envisaged for themselves and now no longer possible post-diagnosis. For many, the grief response appeared more debilitating than any presenting symptoms at the time of diagnosis. This included *"giving up"* hobbies or losing motivation in continuing their education, increased alcoholic consumption, and social avoidance.

> "You don't know what you're going to be able to do in the future. There are so many things left uncertain. There's a lot of anxiety that comes with that, of not knowing what your future is going to look like, and how much control you have over it." (24 years old, diagnosed with Stargardt disease)

3.1.4. Acceptance

Many participants shared how they reframed their thinking to be more accepting of their diagnosis as they got older. For several participants, this acceptance felt like a long journey which guided them to an "*epiphany*". For some participants, accepting their diagnosis involved appreciating what they could see and do, rather than focusing on potential limitations. This change in perspective led to them being kinder to themselves and greater confidence around what they could achieve.

3.2. Daily obstacles: "accepting what I have, adapting where I can"

3.2.1. Ongoing daily practical challenges

Despite accepting their diagnosis, participants raised the challenge of needing to continually adapt to their changing vision. Progressive symptoms most frequently reported were night blindness, blurred vision, decline in peripheral vision, eye strain, fatigue, and issues with depth perception and glare. Many challenges participants reported facing were related to the impacts of these symptoms on daily life (e.g., difficulty navigating a new space, cooking, playing group sports) and social interactions (e.g., noticing physical social cues, like an extended hand to gesture a handshake). Reading and near vision challenges, such as not being able to track a line of text or experiencing eyestrain when reading, often affected information gathering, impacting participants' ability to thrive in a school or work setting. Many participants also reported frustrations with daily tasks requiring seeing low contrast, such as personal grooming, or the ability to differentiate different foods on a plate.

Lifestyle changes made around these daily challenges were predominantly around social interactions. Participants shared how social outings were limited to daytime, well-lit, quiet or familiar environments. Socialisation required planning in advance and engaging with technology to communicate and navigate the world.

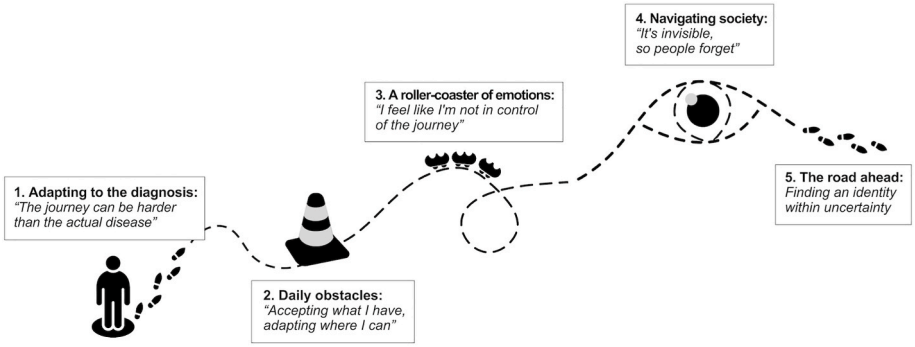

Fig. 1. Summary of key themes.

"Going out … especially in my early 20s, I was feeling a bit vulnerable. I would be going out in the evenings to places that young people go and I would have trouble navigating." (32 years old, diagnosed with rod-cone dystrophy)

3.2.2. Driving as a representation of independence

One of the most cited challenges was no longer being able to drive, or the fear of if or when this would happen. For many, the loss of a driver's license was synonymous with their perception of independence. Some individuals noted the further burden of losing their license when there was a lack of resources, beyond family and friends, to support them with transport (e.g., lack of public transport in regional/rural areas or funding schemes for ride-sharing services or taxis).

"Having to stop driving was a big one … for practical reasons, but also that was like a freedom that is really hard to give away once you've had it. I chose to stop driving … I wasn't told to. If I'm going to constantly have things taken away from me because of my vision slowly going, why wouldn't I take control where I can?" (35 years old, diagnosed with rod-cone dystrophy).

3.2.3. Balancing self-sufficiency and support

Many participants expressed a struggle between wanting to maintain independence yet becoming more reliant on family, friends, and partners. The loss of independence led several participants to feel like a burden, or that they were "not as helpful" than they previously were. Many participants acknowledged the conflicting desires of wanting to be treated "normally" yet needing others to understand and accommodate for their vision loss.

"You don't want your friends to necessarily be treating you like you need to be [pigeonholed]. On the flip side of that they were still treating me completely normally, which is not actually what you necessarily need. As someone with a visual disability, you actually do need to be treated a little bit differently insofar as you might need a little bit more help." (24 years old, diagnosed with Stargardt disease)

3.3. A roller-coaster of emotions: *"I feel like I'm not in control of the journey"*

3.3.1. Fluctuating emotions

Participants expressed a wide range of emotions - both of positive and negative valence, and high and low arousal. Common negative emotions were anxiety, anger, sadness and despair, which shifted over time, often occurring in waves and triggered by periods of worsening vision. Similarly, feelings of embarrassment and frustration would often arise when worsening vision impacted their daily functioning.

Despair was prevalent among participants. They described feeling apathy toward life, with many choosing to stop hobbies, socialising, or their education/career. For some, this further exacerbated their sense of social isolation and wavering self-confidence.

"Worry about my vision getting worse comes on me from time to time when I start talking about it. I suppose if I'm busy, I don't think about it too much. But it can come and go … I do sort of wake up with a bit of dread every morning thinking it's not going to get any better." (67 years old, diagnosed with macular dystrophy)

3.3.2. Anticipatory grief

Despite being triggered by worsening vision, several participants felt that their distress was not directly related to their vision impairment at the time, but rather in anticipation of their future. Participants experienced anxiety and feeling a lack of control from the uncertainty surrounding their disease progression, accompanied by feelings of powerlessness regarding what the future may hold.

"Not knowing a timeline. Maybe those with other [genetic diseases] have a timeline but it's scary not knowing when your vision is going to get worse. Who will be there to support me?" (24 years old, diagnosed with rod-cone dystrophy)

3.3.3. Cognitive load

Despite several participants stating the daily challenges were *"just little things",* they also expressed an immense cognitive load and time burden in needing to constantly problem-solve and adapt. This resulted in hypervigilance, leaving them feeling mentally fatigued. Participants conveyed exhaustion, lack of control, and constant heightened awareness to navigate the world and accomplish basic tasks, like getting to work or school with vision impairment. Many conveyed the draining toll of expending immense energy just trying to function independently.

"I live in constant fight or flight … and on edge all the time expecting there's something I'm not seeing. Your other senses go overboard … your brain is in overload … and it's absolutely exhausting." (49 years old, diagnosed with Bietti crystalline dystrophy)

3.3.4. Coping mechanisms

As a coping mechanism, the use of laughter and humour was a positive coping strategy that helped combat despair. Participants highly valued emotional support from their family, friends, and health professionals, who could relieve some cognitive load as they became more familiar with the condition and the challenges.

"Being able to take a really bad experience and deal with it through having a laugh about it with someone went a long way towards making me feel a lot better about myself and my situation." (24 years old, diagnosed with Stargardt disease)

Table 2
Illustrative quotes of themes and subthemes.

Theme	Subthemes	Illustrative quotes
Adapting to diagnosis: "The journey can be harder than the actual disease"	Shock of initial diagnosis	*"So the initial response in my mind was almost, like "Is this serious?" How … could this possibly be happening to me? I think I didn't really grasp the ramifications straightaway … emotionally, I wasn't able to … comprehend the scale of what was going on, because it was so far out of my experience."* (24 years old, diagnosed with macular dystrophy)
	Denial and avoidance	*"I didn't want to think about it - what would happen when I lost my eyesight. As normally, [there is slow] loss of peripheral vision towards blindness [by] around age 40. I probably sort of pushed it back and didn't want to think about what the consequences would be when I was 40. Because I was really worried about that."* (23 years old, diagnosed with choroideremia)
	"Grieving a life that I wanted to lead"	*"A lot of my emotional response was grief to a loss, which is the loss of my vision, and then all the things that you do with your vision that you don't even necessarily take for granted."* (24 years old, diagnosed with macular dystrophy)
	Acceptance	*"I'm very proud of who I am and the disability that I have. But if I was 15 or 14 … it's definitely something that I tried to hide from, not to stand out. And then also just realizing a lot of people have disabilities in high school. Just most people tend to try and hide them."* (23 years old, diagnosed with choroideremia)
Daily obstacles: "Accepting what I have, adapting where I can"	Ongoing daily practical challenges	*"I feel like I'm expending a lot of energy and just concentrating on not tripping over …. it's challenging learning a new place and going to new spaces."* (34 years old, diagnosed with rod-cone dystrophy)
	Driving as a representation of independence	*"I think that there is a big gap in our support systems when it comes to legal blindness and low vision. One of the barriers that I've noticed since I've lost my license is it's just impossible to move around without support, without great costs. When you look at the resources, they're for legally blind people, but there's not much [for] in between."* (32 years old, diagnosed with rod-cone dystrophy)
	Balancing self-sufficiency and support	*"I hate having to rely on people because I'm very independent. I think the impact of a diagnosis and finding out what it would mean now and going forward, suddenly it was a lot bigger than I thought it would be … a huge impact on my independence."* (35 years old, diagnosed with rod-cone dystrophy)
A roller-coaster of emotions: "*I feel like I'm not in control of the journey*"	Fluctuating emotions	*"It goes through seasons … I still sometimes feel in a really dark moment, but it happens less. It usually happens around a period of changing in vision if I notice it."* (32 years old, diagnosed with rod-cone dystrophy)
	Anticipatory grief	*"I feel like I'm not in control of the journey, because I know that there's a journey of decline … I know that I'm losing vision over time."* (32 years old, diagnosed with rod-cone dystrophy)
	Cognitive load	*"It is exhausting, absolutely exhausting when … you can't rely on your main sense to help you function and be part of the world and society."* (49 years old, diagnosed with Bietti crystalline dystrophy)
	Coping mechanisms	*"I'm gradually acknowledging that I'm not … perfect ….and people need to accept that imperfection. It's probably meant that I'm also more tolerant of other people's imperfections."* (75 years old, diagnosed with rod-cone dystrophy)
Navigating society: "it's invisible, so people forget."	The Invisibility Conundrum	*"It's either speak up and work through the feeling that it's vying for attention or don't speak up and take the challenge of not being understood."* (49 years old, diagnosed with Bietti crystalline dystrophy)
	Public misconceptions	*"People associate disabilities with something that they can see or like a walking stick or cane. And then they're thinking, what's this guy talking about? I don't see a guide dog or a cane."* (24 years old, diagnosed with macular dystrophy)
The road ahead: Finding an identity within uncertainty	Shifting senses of identity	*"I've grown up my entire life in with the privilege of believing that I could do whatever I wanted to do. Now I feel like I can't … You're going to have to work harder than everybody else, and probably not even get as far. Having had time to process it, you start realizing, you're just going to have to work a bit smarter than other people, not necessarily harder."* (24 years old, diagnosed with macular dystrophy)
	"Struggle with an undetermined outcome"	*"It's really full on not knowing exactly when your vision is going to decline … not knowing the timeline changes …"* (24 years old, diagnosed with rod-cone dystrophy)
	Redefining belonging and purpose	*"While it may be limiting in some areas … there's still so many things that you can do. So that's a part of the realization, …in growing up and maturing."* (23 years old, diagnosed with choroideremia)

3.4. Navigating society: "it's invisible, so people forget"

3.4.1. The invisibility conundrum

For some participants, having an invisible disability provided an opportunity to hide their condition. This was a strategy commonly used by individuals during adolescence in a desire to feel "normal". In contrast, some participants were frustrated of constantly having to advocate for themselves and remind people of their condition. Many participants reported a poor understanding from others about the nuances of partial sight and varying degrees of vision loss, and there were times that participants felt a need to exaggerate their "blindness" to feel their condition was appropriately recognised by others.

> "I have to spell things out more and I have to act more blind to be validated." (49 years old, diagnosed with Bietti crystalline dystrophy)

3.4.2. "I don't see a guide dog or a cane"

A common challenge reported was that many people in the public did not associate a normal appearance with a disability or did not comprehend that people were still able to see with some limitations. This was frequently reported by younger individuals with IRD.

> "Being young can be a hindrance as well … people look at you as 'oh, they're young, they seem to be fit and healthy.' People don't associate visual disabilities with people that are healthy." (24 years old, diagnosed with Stargardt disease)

3.5. The road ahead: finding an identity within uncertainty

3.5.1. Shifting senses of identity

Many participants recalled how their diagnosis impacted their transition into adulthood, questioning previously envisioned career paths and instilling fears about living a fulfilling life. The unpredictable trajectory about IRDs often intensified anxiety about future family and career prospects, creating an identity crisis and a sense of a limited future.

> "I didn't feel like [my diagnosis] happened at an age where I could manage it. I wasn't really interested in pursuing a long life or a life where I needed to be well rounded in other areas." (32 years old, diagnosed with rod-cone dystrophy)

Some people lost their ability to engage in hobbies, such as reading or knitting, and activities deeply tied to one's identity. This loss had a significant negative impact on an individual's sense of identity and future life narratives.

> "It [reading novels] was one of my favourite things to do, and the thought of not being able to read, it's one of those things where they're … parts of your identity, that you start to worry are going to get wiped away, or [be] taken from me." (24 years old, diagnosed with Stargardt disease)

3.5.2. "Struggle with an undetermined outcome"

Participants highlighted a multitude of concerns and uncertainties about their career, family, relationships and the future. Some have pivoted to jobs better suited to accommodate their vision. Many also worried about having limited job opportunities due to the need for workplace modifications, being perceived as a safety concern, and facing discrimination based on their disability. Some found disability employment services lacking, and raised concerns about generating sufficient income and financially supporting a family, making workforce entry and retention extremely challenging and intimidating.

> "When I am blind, I won't be able to [do the job I do now]. I don't know what to do, no one told me about it," (33 years old, diagnosed with rod-cone dystrophy)

Worries regarding the future also included concerns about finding a romantic partner who may have to become their carer, feeling less desirable due to their disability, worrying about being a burden on loved ones, and passing on the condition to their children.

> "And I used to be very worried with things like, what if nobody loves me? And it's really silly, but what if no one wants to commit to a future with someone who might go blind in the future?" (24 years old, diagnosed with rod-cone dystrophy)

3.6. Redefining belonging and purpose

Even after accepting their diagnosis, many participants commented on the need to continually readapt their self-identity, capabilities, and cultivate resilience as their vision and lives changed. Some participants expressed hope in newer technologies providing new possibilities and accessible options for people with vision impairments.

Several participants commented on the impact of knowing, or not knowing, others who had a vision impairment. Participants who had seen a family member with an IRD continue to live a fulfilling life appeared to have more positive perceptions of their future. Many participants highlighted the profound impact that positive representation and guidance from others with vision loss can have in shaping their outlook and ability to envision a meaningful future despite challenges.

> "I didn't see many people with disabilities growing up and I certainly didn't see many people with vision issues." (32 years old, diagnosed with rod-cone dystrophy)

4. Discussion

Our qualitative study explored the lived experiences of individuals navigating the challenges of early vision loss among people with an IRD. Our findings highlighted the multifaceted personal and societal challenges associated with navigating a vision impairment, exacerbated by its nature as a "hidden" disability.

IRDs are a unique hidden disability given the progressive nature of the disease yet uncertainty around the rate of progression. Our findings align with prior studies showing that vision loss from IRDs significantly impacts physical functioning, psychological well-being, and social functioning.[13–17,22] Similar physical and identity challenges have been described for other causes of vision loss, such as adventitious blindness, and in age-related macular degeneration and glaucoma commonly affecting older adults.[22–25] In contrast to these conditions, we highlight that a distinct challenge of navigating early-stage IRD is uncertainty stemming from their unclear disease progression. Compared to conditions diagnosed in early childhood or in older adults, these challenges can be particularly overwhelming for IRDs that are often diagnosed in the second or third decade of life, a critical developmental period where individuals explore their sense of self and identity.[26] This uncertainty amplifies concerns regarding shifting identity, anticipatory grief, and future outlook.[27] These findings align with a 2024 Australian Priority Setting Partnership (PSP) for IRDs,[28] which found that the IRD community placed value on understanding what the anticipated progression for their condition is, as well as any environmental and lifestyle factors that can influence disease progression. The strong desire for this information may be particularly relevant in the early-stages post-diagnosis, highlighted by the PSP identifying information and psychosocial needs at diagnosis a top priority.

Studies in young people with moderate to severe vision loss from various causes have found that vision impairment amplifies the dynamic changes and challenges young people experience during

adolescence.[26,29] Consistent with these findings, we show that in IRD, even before experiencing significant vision loss, the inevitability of knowing future vision loss will occur impacts relationships and developing independence. This can affect workforce choices and participation, stemming from a sense of a limited future due to impending vision loss. This finding is consistent with impacts on academic progress, student retention, and higher education progression seen in young adolescents with other hidden disabilities, such as neurological disorders and learning disabilities.[3] In the U.S., labour force participation was most burdensome for people with vision loss between 19 and 64 years of age, over medical and care costs. These findings emphasise the need for advocacy within workplaces to account for the unique challenges of vision impairment, to provide more equitable opportunities.[30,31] Increased representation of hidden disabilities in the media can also serve as aspirational examples to help shape a positive future outlook for young people.

Early stage IRDs tend not to reveal outward signs connected to an impairment, particularly if vision impairment is mild and vision aids are not needed. The feeling of being "blind but not blind enough", often challenged individuals with IRDs in communicating their needs and seeking support. Furthermore, in many countries, impairment severity needs to meet the definition of legal blindness before individuals may be qualified to receive financial support.[32] These findings emphasise the need for better public awareness about the spectrum of vision loss, as well as support frameworks that address the paradoxical concept of having vision loss appropriately recognised and accommodated, while also reducing the associated social stigma of being different.

People with IRDs have dual burdens of living with a genetic condition and vision loss, and a complex range of perceived medical, psychosocial, and practical healthcare needs.[29] In early stages, avoidant responses were often used as an coping mechanism to alleviate concerns about disease progression. However, avoidance can also delay diagnosis acceptance and postpone seeking vision and psychological support.[33] Psychosocial support is crucial from the time of diagnosis throughout the entire disease journey.[11,34] The desire for further research to develop effective psychosocial support for both individuals with an IRD and caregivers was also highlighted as a top priority for individuals with IRDs in a Australian IRD PSP.[28] Our findings emphasise the need for a dynamic IRD support program tailored to the stage of the participant's condition, considering both emotional acceptance of the diagnosis and visual needs. This support should be adapted to meet the evolving needs of IRD participants and caregivers as their vision impairment and psychosocial acceptance evolve.

Due to the anonymous nature of our study, we did not capture socioeconomic demographics like income and education levels, and we were unable to capture the experiences of individuals who did not speak English. Our targeted recruitment through existing IRD channels and purposive sampling may limit the generalizability of our findings, as our participants may have been more engaged in their healthcare. We did not limit participants by their time since diagnosis to capture a range of experiences, including long-term impacts, and account for the slow progression rate of some IRDs; however, with over half of the sample diagnosed more than 5 years ago, there is a possibility of recall bias. We did not collect information on participants' vision impairment levels, which could have provided additional context about participants' current visual abilities. Self-reported vision levels may not directly reflect disease severity or coping experiences, and individuals with similar impairment can adapt differently. While this study only explored the impact of a clinical IRD diagnosis, receiving a genetic diagnosis, which frequently occurs later for most people with IRDs,[35] can also influence how the condition is perceived. A potential area for future research is exploring how the timing of genetic testing influences the perception and experience of early-stage IRDs. While we attempted to reduce subjectivity in data interpretation by having multiple researchers from multidisciplinary fields contribute to theme development, we acknowledge that individual values and experiences can influence the collection and analysis of data.

5. Conclusions and future directions

Our study contributes to the limited literature on the challenges navigating progressive vision loss, from the perspective of individuals

Table 3
Suggested areas of support for individuals with inherited retinal diseases (IRD).

Domain	Suggestion
Clinical and diagnostic support	• For clinicians, tailoring management and recommendations to a patients' functional level and psychological readiness for change that is also age appropriate. •Supporting the parents and carers of individuals diagnosed with an IRD to understand the possible implications for the future. •Integrating low vision rehabilitation services and assistive technology training as early as an individual could accept, to equip them with the skills and tools for maintaining independence as vision changes. •Providing clinician training on communication skills, particularly regarding delivering news of diagnosis. •Developing follow-up information on assistive tools, networks, and resources to facilitate ongoing support and access to relevant services after the initial diagnosis.
Multidisciplinary networks	•Fostering multidisciplinary networks to ensure clinicians have knowledge of and can refer to available support services and professionals for patients and their families, and to facilitate interdisciplinary communication. Networks may include: o Primary eye care providers (optometrists) oSpecialists eye care providers (ophthalmologists, including IRD specialists) oGenetic counsellors, geneticists oOccupational or vocational therapists oPsychologists, counsellors, and mental health professionals oOther medical and allied health professionals for syndromic conditions oPatient organisations, community support groups, and research teams, as additional sources of support for clinicians and families
Psychological support	•Offering psychological support to assist individuals and families in coping with the emotional and psychological challenges of adjusting to an IRD diagnosis. •Developing specific counselling programs tailored for addressing the practical and psychosocial challenges associated with IRDs and the genetic nature of these conditions.
Peer support	•Strengthening social and peer support networks, which can serve as valuable resources for individuals with IRDs and their families to share experiences, coping strategies, and practical advice. •Promoting the sharing of personal stories and experiences of individuals living with IRDs, which can be especially helpful for individuals who are newly diagnosed, fostering more positive perceptions of their future and strategies for managing their condition.
Community support	•Advocating for additional resources and support for transportation and mobility services, including a system that is transferable across states and territories. •Working with educational institutions to ensure appropriate accommodations and support services are available for individuals living with IRDs in schools and Universities, such as assistive technology and accessible materials and orientation and mobility training. •Advocating for improvements to existing employment services programs to ensure that providers are able to effectively work in partnership with people who are vision impaired to support their equitable participation in the workforce. •Facilitating access to IRD research studies and clinical trials, enabling individuals to explore emerging treatment options and contribute to advancing scientific knowledge.

with an early stage IRD. In addition to practical challenges, we found a significant emotional toll stemming from the uncertainty surrounding disease progression and additional cognitive load and adaptation needed to navigating daily practical obstacles. The invisibility of IRDs amplified social challenges. These lived realities underscore the importance of tailored support addressing the unique needs of individuals with vision impairment, and advocacy for awareness that vision loss exists on a spectrum.

Drawing from participant insights and existing literature,[27,36] we have suggested several areas for developing support interventions for individuals living with IRDs (Table 3). Our findings emphasise the need for increased awareness, understanding, and inclusive support to tackle the distinct challenges faced by those with non-visible impairments to improve their wellbeing and quality of life.

Data access

The corresponding author (ACBJ) has full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis.

CRediT authorship contribution statement

Mariam Bakir: Writing – review & editing, Writing – original draft, Formal analysis. **Eden G. Robertson:** Writing – review & editing, Writing – original draft, Visualization, Methodology, Formal analysis. **Ruofei Trophy Chen:** Writing – review & editing, Project administration, Investigation, Formal analysis, Data curation. **Bao N. Nguyen:** Writing – review & editing, Formal analysis, Data curation. **Bronwyn McFadyen:** Writing – review & editing, Project administration, Methodology, Investigation. **Eve Makrai:** Writing – review & editing, Investigation, Formal analysis. **Leighton Boyd:** Writing – review & editing, Methodology, Funding acquisition. **Rosemary Boyd:** Writing – review & editing, Methodology, Funding acquisition. **Sally Karandrews:** Writing – review & editing, Methodology, Funding acquisition. **Lauren N. Ayton:** Writing – review & editing, Methodology, Funding acquisition. **Alexis Ceecee Britten-Jones:** Writing – review & editing, Writing – original draft, Validation, Supervision, Software, Resources, Project administration, Methodology, Investigation, Funding acquisition, Formal analysis, Data curation, Conceptualization.

Data sharing

The codebook containing non-identifiable data is available from the corresponding author (ACBJ) upon reasonable request for ethically approved projects.

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Declaration of competing interest

None.

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Appendix A. Supplementary data

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