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Original Article

Effect of inherited retinal diseases on Canadian patients and families: a mixed-methods study

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Objective: To understand the physical, psychosocial, and practical challenges faced by Canadian patients with inherited retinal diseases (IRDs) and their families.

Design: Mixed methods.

Participants: A total of 408 Canadians living with or caring for someone with an IRD (mean age = 51.4 ± 16.7 years) completed an online survey. Twenty cohort respondents participated in additional telephone interviews.

Methods: The online survey was comprised of questions concerning demographics, self-reported vision, genetic testing, information preferences, health care experiences, treatment goals, and disease impact on daily life. Recruitment occurred through Fighting Blindness Canada's community database. Survey dissemination also occurred via social media and not-for-profit stakeholder outreach. Subsequent to survey completion, a subset of respondents participated in semistructured telephone interviews to further elucidate illness experience.

Results: Respondents identified having 1 of more than 14 IRDs, with 72% specifying retinitis pigmentosa. Sixty-eight percent reported being legally blind, and more than 85% self-reported moderate to low vision or worse. IRDs impacted daily functioning, with 53% of respondents indicating that they affected employment or education. Psychological challenges were evident, with more than 70% worried about coping with daily life and more than 60% indicating fear and stress. Qualitative data described hopelessness around suitable work, loss of independence, and challenges with social interaction. Sixty-five percent reported a negative impact on family life. Many had not accessed social support services because of a lack of perceived need, awareness, or availability.

Conclusion: Canadian patients with IRDs report moderate to severe visual impairment, and both patients and their families describe an impact on psychosocial well-being and functioning during daily activities. Vision rehabilitation with a psychosocial approach is necessary, alongside facilitating access to emerging treatments.

Inherited retinal diseases (IRDs) are a set of genetic conditions that cause progressive retinal damage and visual impairment.[1] Disrupted retinal pigment epithelium integrity alongside photoreceptor degeneration contributes to vision loss.[1] Characterized by genetic heterogeneity, IRDs can present with autosomal dominant, autosomal recessive, or X-linked modes of inheritance, whereas other cases are sporadic or with equivocal mutations.[2] Clinical heterogeneity comes from varied visual impairment, age of onset, progression rate, and extent of systemic involvement.[3] Retinitis pigmentosa, the globally predominant IRD, causes night blindness and peripheral vision changes.[3,4] IRDs with cone degeneration present with photophobia, diminished acuity, and affected colour vision.[3] Certain IRDs implicate central vision, whereas syndromic forms have concomitant systemic involvement.[5]

Current treatment focuses on symptom management because prognosis is usually not reversible. While single-gene augmentation therapy is federally approved, most therapies remain in the research phase.[1,6]

Progressive vision loss with few treatment prospects creates a context for mental health symptom development and diminished quality of life.[7] The literature indicates that low vision increases the odds for depressive symptoms and is significantly associated with lower quality of life. Impaired vision affects independence, social capital, mobility, relationships, and activities of daily living.[7–10] Given that IRDs are hereditary and can present in childhood, impacts on emotional, social, and financial well-being extend to caregivers (often familial).[11]

Estimates in 2019 report approximately 21,000 Canadian IRD cases.[11,12] Research estimates that the Canadian cost of IRDs is between $1637.8 million and $6687.5 million, of which 66% is solely attributed to loss of well-being costs measured in disability-adjusted life-years.[11] Other prominent contributors include productivity losses and informal caregiver costs.[11] Extensive well-being costs with minimal treatment options establish the need to characterize the lived experience of patients with IRDs. There is a paucity of literature on the Canadian IRD illness experience. Consolidating a Canadian experience is integral to ensuring national access to newly developed IRD treatments. This elucidation is imperative for Canadian-based policy change and institutional investment in accessible health services, in addition to informing patient-centred clinical care.[11]

Using a mixed-methods approach, this study aims to understand the physical, psychosocial, and practical challenges faced by Canadian patients with IRDs and their

https://doi.org/10.1016/j.jcjo.2022.06.021
ISSN 0008-4182

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families to identify areas of reform from a medical, policy, and social care perspective.

Methods

Study design and participants

This sequential mixed-methods study was comprised of 408 online survey respondents who were either living with an IRD or were a caregiver completing the survey on the patient's behalf (n = 42). The latter was with a minor or when an individual required assistance. When caregivers were included in the survey process, they were encouraged to complete the survey along with the individual diagnosed with the IRD and act as an aid for data input, if needed by the individual. Respondents had to be Canadian residents at the time of survey administration with fluency in English or French. Exclusion criteria were minimized for a generalizable sample reflective of the Canadian landscape. The survey link was emailed to individuals categorized as having an IRD or having a relationship with someone with an IRD in Fighting Blindness Canada's (FBC) community database. FBC is a national not-for-profit funder of vision research. Survey dissemination also occurred via social media, electronic newsletters, and not-for-profit stakeholder organization outreach to constituents. The survey was accessible from late March to late May of 2020.

Twenty respondents subsequently participated in telephone interviews between late July and early August. They provided consent to be interviewed during online survey completion. Participation was voluntary, and compensation was provided. Research was conducted in compliance with Advarra Institutional Review Board.

Data collection

Survey fields included demographics, self-reported vision, genetic testing, information preferences and sources, health care experiences, support services use, treatment goals, and disease impact on daily life (Appendix A, available online). A trained facilitator conducted 30-minute telephone interviews using a semistructured guide. Interviews were recorded and immediately transcribed for data analysis (Appendix B, available online).

Data analysis

Survey data were analyzed using descriptive statistics with STATA software version 10.0 (StataCorp LP, College Station, Tex.). Using conventional content analysis, qualitative data were processed by 2 researchers who independently coded interview transcripts to iteratively develop a coding dictionary, with disagreement resolved by consensus. Codes were inputted into qualitative software (QSR NVivo version 8.0; QRS International, Melbourne, Australia) to establish emerging themes.

Results

Participation

The study sample had a mean age of 51.4 years (SD = 16.4 years). Fifty percent of respondents lived in Ontario, and 88.1% of respondents resided in urban locations (Table 1).

IRD diagnosis and ocular status

Of respondents, 75.1% identified having retinitis pigmentosa (Fig. 1), and 76.9% had been diagnosed with their IRD

Table 1—Sociodemographics of study population

Characteristic	n (%)
Age (n = 340)	
19–29 y	40 (11.8)
30–39 y	54 (15.9)
40–49 y	57 (16.8)
50–59 y	65 (19.1)
60–69 y	68 (20.0)
70–79 y	49 (14.4)
>80	7 (2.1)
Biological sex (n = 341)	
Male	156 (45.7)
Female	185 (54.3)
Intersex	0 (0.0)
Gender identity (n = 338)	
Male	154 (45.6)
Female	183 (54.1)
Other (i.e. transgender, nonbinary, fluid, two-spirited)	1 (0.3)
Ethnicity (n = 344)	
Arab	10 (2.9)
Black	5 (1.5)
Chinese	5 (1.5)
Filipino	2 (0.6)
French-Canadian	28 (8.1)
Japanese	2 (0.6)
Korean	0 (0)
Latin American	7 (2.0)
South Asian	5 (1.5)
Southeast Asian	4 (1.2)
West Asian	2 (0.6)
White	237 (69.5)
Inuit	1 (0.3)
Métis	3 (0.9)
First Nations	1 (0.3)
Other (i.e. Scottish, Hungarian, Italian, mixed race)	18 (5.2)
Prefer to not answer	14 (4.1)
Province (n = 390)	
British Columbia	61 (15.6)
Alberta	52 (13.3)
Saskatchewan	13 (3.3)
Manitoba	8 (2.1)
Ontario	195 (50.0)
Quebec	29 (7.4)
New Brunswick	7 (1.8)
Nova Scotia	17 (4.4)
Prince Edward Island	3 (0.8)
Newfoundland and Labrador	4 (1.0)
Yukon	1 (0.3)
Highest level of education completed (n = 341)	
No school	1 (0.3)
Nursery school to grade 8	4 (1.2)
Some high school, no diploma	0 (0.0)
High school graduate, diploma or equivalent	36 (10.6)
Some college credit, no degree	63 (18.5)
Trade, technical, or vocational training	36 (10.6)
Associate degree	28 (8.2)
Bachelor's degree	97 (28.4)
Master's degree	45 (13.2)
Professional degree	19 (5.6)
Doctorate degree	12 (3.5)

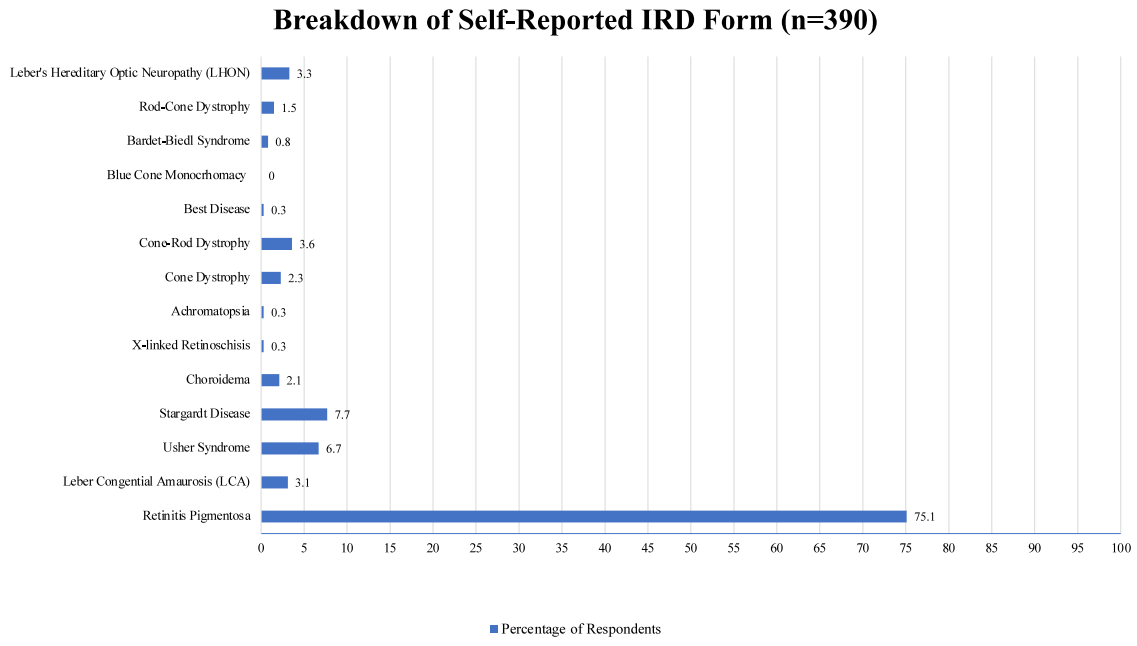

Fig. 1—Results from 390 survey respondents who self-reported having 1 of 14 inherited retinal diseases.

more than 10 years ago, with 1.8% receiving their diagnosis in the past year, suggesting that this data set represents a chronic illness experience. While 49.3% of respondents had no other ocular conditions, 33.7% had concurrent cataracts.

Self-reported vision

Of those surveyed, 68.7% self-reported legal blindness status, with 40.3% reporting no night vision; 34.6% self-reported good central vision, while 9.0% indicated good peripheral vision. When asked to self-describe vision, more than 85% of respondents described their vision as moderate to low vision or worse (Table 2).

Genetic testing

Genetic testing had been pursued by 59.3% of patients. Of these, 64% were aware of their underlying genetic mutation. More than 45% of respondents had at least 1 family member affected by an IRD, with 13.8% having 5 or more family members affected. Sixty percent reported that family members had not undergone genetic testing looking for the same IRD.

Table 2—Self-described vision (n = 389)

Self-described vision	n (%)
Near-normal vision	58 (14.9)
Moderate to low vision	169 (43.4)
Severe to low vision	114 (29.3)
Near-total blindness (some light perception)	40 (10.3)
Total blindness (no light perception)	8 (2.1)

Information sources and preferences

When asked about sources of IRD information, 71.5% reported relying primarily on their ophthalmologist and supplemental online sources (Canadian National Institute for the Blind and FBC). At diagnosis, respondents prioritized receiving information on expected illness experience, access to support services, coping with vision loss, and information on clinical trials. Yet qualitative data elucidated a need for a graduated and patient-centred approach to information sharing. One respondent shared, "I mostly needed access to services, information, and emotional support that matched the stage of my vision loss. Being flooded with 'help' that didn't match my needs and abilities was more traumatic than my diagnosis."

Interactions with care providers

More than 80% of respondents had seen a care provider for their IRD in the last year, with most reporting an annual appointment frequency. More than 70% reported being satisfied or very satisfied with their physician-led IRD care. Seeing a provider was challenging because of travel, with 30% reporting travel time lengthier than 1 hour complicated by long distances and the inability to drive independently. Importantly, 80% shared that their providers did not regularly ask about mental health and well-being during clinic visits.

Treatment goals

When asked about treatment goals, 92% prioritized improvement of overall sight, which was notably higher than the 75% who valued finding a cure. Participation in social activities and relationships was as important as

continued participation in work or school. Fifty-seven percent of respondents indicated that improving night vision and mobility would significantly improve their quality of life, and almost 75% believed that a treatment that met personal goals would significantly improve emotional well-being. Yet a pervasive qualitative theme was the belief that there are no prospects for treatment or a cure within their lifetime.

Support services

Counselling, social services, mobility training, genetic counselling, advice on claiming benefits, occupational support, and support around switching careers were not used by 50% of respondents, most commonly because of perceived lack of need. However, many were unaware that social services or advice on claiming benefits was available. Of respondents using low-vision services, mobility training, genetic testing, and advice on claiming benefits were most beneficial.

Impact on occupation and education

For 44.4% of respondents, employment or school had not been affected by their diagnosis, whereas 54.8% reported altered occupational or educational functioning. Mean age and age of diagnosis were similar between those reporting unaffected versus affected occupation or education, suggesting that impact transcends stage of disease progression. When asked about how their IRD impacted their ability to perform school responsibilities, 48% reported a 7 or above out of 10, with 10 being a very severe impact.

Qualitative data indicate that occupational functioning is affected by unsuitable work environments compounded by mobility and transportation limitations. Respondents mentioned sadness, fear, and hopelessness in their search for suitable work. One participant noted, "Any previous attempt to apply for employment was marred by indifference and lack of interest in me due to my legally blind status. It has been very personally challenging. I am scared for my future." Respondents who were unable to secure employment felt forced to switch career paths, which resulted in additional training. Furthermore, maintaining workload with progressive vision loss caused fear around job security. Many participants required altered hours to ensure that work and commuting happened during daytime and to arrange for accommodations for reading small print, long breaks, and more time to complete tasks. Respondents described the need for support persons and assistive devices, with canes and magnifiers and modifications to mobile phones being the most beneficial assistive technologies. Yet this support was often unavailable or unsupported in the workplace.

Impact on daily life and mental health

Seventy-one percent of respondents reported an above-moderate negative impact on quality of life, with most severe effects experienced with mobility, leisure activities, socializing, and reading. With 68% of respondents thinking about their IRD at least once a day, respondents worried about their safety outside the home as much as coping with everyday life and progressive vision loss. For 20% of respondents, this worry extended to their safety within their homes. When asked about negative feelings, 72% reported frequent stress, and 63% reported fear.

Impact on family and social functioning

Twelve percent of respondents indicated that they live alone, 63.3% reported living with a partner, and 25.6% reported living with their children. Sixty-five percent reported a negative impact on family life. Qualitative themes suggest that this is from guilt about heritability and feeling like a burden. Many respondents discussed their inability to assume familial responsibilities and being unable to parent their children the way they had hoped to. Many felt that they were not trusted or relied on by family. Disease progression led to fiscal challenges, denial, abandonment, isolation, marital trouble, and estrangement. Respondents commented on the perpetual angst experienced by family members about the present and future. However, many respondents knew of family members with an IRD that was an integral source of support.

Seventy-four percent of respondents reported a negative impact on social life because vision loss limited independent transportation, confidence in social situations, and familiarity with new surroundings, all compounded by fear of personal safety. Many avoided all social situations, frequently turning down social invitations. Half the respondents felt stigmatized, most commonly at work and also by friends, family, and the general public.

Discussion

These data highlight the medical and psychosocial burdens IRDs present for Canadian patients and families, elucidating the need for enhanced awareness, treatment, and social support. are congruent with global IRD epidemiology in age, ethnicity, and IRD form distributions.[13,14] However, congruency with Canadian IRD epidemiology is difficult to establish given the paucity of literature, highlighting the necessity for national research such as FBC's IRD Patient Registry.[15]

Self-reported vision indicates a need for vision rehabilitation, especially with peripheral vision changes supported by previous literature.[16] The discrepancy between the 68.7% with legal blindness and the 85% who self-described moderate to low vision or worse demonstrates that objective thresholds miss nuances of visual impairment and the

importance of self-reported vision in clinical assessment of lived experience.[17,18]

Responses show that while genetic testing is pursued, education on results, which is imperative for treatment eligibility and family-based care, can improve. This also may contribute to the discrepancy between prevalence of family members with IRDs and the rate of family-based genetic testing in this study.[19,20] The literature suggests that providers and patients have a positive view of genetic testing, and when treatments are available, genetic testing is viewed more favourably.[20–22] Provider knowledge, influenced by guideline availability and accessibility of new IRD genetic research, also may contribute to pursuit of testing.[21,22]

Responses demonstrate a strong trust in ophthalmologists for IRD information and care.[23] Yet information preferences suggest that patients wish to focus on empowerment, coping, and illness experience rather than medicalized information, similar to published studies.[23] Within this data set, there is alignment between information preferences and treatment goals that also focus on care elements such as quality of life, social functioning, occupational or educational productivity, and emotional well-being. Given the reliance on ophthalmologists and the inextricable link between vision loss and mental health, improving on regular mental health checks during clinic visits may invite further discussions on the psychosocial IRD experience and treatment goals beyond sight restoration.[7,23,24]

Psychosocial data demonstrating a moderate to severe impact on quality of life are similar to those of previous studies, only reinforcing the necessity for physician-led regular well-being checks that complement vision care.[16,25–28] Other studies report anxiety, fear, and loss of confidence to be prominent psychological challenges, similar to this data set.[16,25–28] Published literature similarly establishes predominant challenges around mobility, leisure, and engaging in social activities.[16,25–28]

Published literature demonstrates that patients prioritize sight restoration and regaining independence, similar to this cohort, whose participants prioritized engagement in social and professional life.[23] Yet the literature suggests that patients with IRDs globally feel unhopeful about treatment prospects.[29] Research demonstrates that the priority for researchers, patients, and eye health professionals is innovation of therapies that slow down disease progression rather than determining a cure, which is in alignment with Canadian cohort treatment goals.[29]

Results establish an underutilization of support services, attributed to a lack of awareness and (or) a perceived lack of need. Yet the collected data on IRD impact on daily life demonstrate a discrepancy with this perceived lack of need. The low frequency of mental health and well-being checks during clinical encounters could indicate fewer encouraging discussions vis-à-vis accessing services and supports. This underutilization may reflect the siloed nature of clinical care and community-based support services, highlighting the need for patient navigators and liaisons. Further, this perceived lack of need may be attributed to acceptance of an illness trajectory and an unwillingness to engage in rehabilitation, further highlighting the importance of well-being in IRD care.[23]

The subjective, patient-centred survey responses in these data are unique, complementing the objective, medicalized outcomes emphasized in literature. This mixed-methods data set is an important piece of advocacy for IRD patients and their families, an underrepresented and marginalized Canadian patient population who are particularly underserved in the context of development of and access to new treatments.[11]

This study has limitations; because the study used a survey and respondents had a chronic illness experience, data are subject to recall bias and response bias. Furthermore, the use of caregivers during data collection may have introduced heterogeneity into the response pool. Of the 408 surveys, only 42 were completed with the help of caregivers (10.2%). Of the 42 surveys from caregivers, 29 indicated that the individual was legally blind, and therefore, it was likely that the caregiver was necessary to facilitate the data-collection process. Although caregivers were asked to aid in data collection without using their own perceptions, there is no way to ensure that this occurred. Caregiver support was used in this study to include patients with severe visual impairment who were unable to access screen-based technology, as well as respondents who may not have been able to complete a survey on their own, with attempts to capture a real-world, generalizable data cohort. Given the lower power in the near-normal vision and total blindness groups, and to maintain the integrity of a real-world sample, stratification of the analysis was not pursued to mitigate ascertainment bias. Selection bias is present because compensation was provided, and recruitment likely captured individuals who were more engaged with their vision loss or who were healthy enough to participate. Because survey dissemination occurred through FBC's community, recruitment may have captured members of the IRD community who were referred to FBC for networks and services to improve quality of life and therefore were more knowledgeable of and engaged with existing supports at the time of survey completion. Importantly, the survey required proficiency in English or French. Further, demographics data suggest that most respondents had received a high school diploma or higher, introducing bias related to socioeconomic status and suggesting that respondents may have been higher functioning.

Although we received no "other," "none of the above," or free responses around IRD subtypes, it is possible that the respondents beyond the 390 who indicated 1 of the 14 listed subtypes were currently undergoing genetic testing or may not have known their IRD subtype. The survey and interviews did not inquire about other syndromic sensory deficits and (or) comorbid chronic illnesses that could confound quality-of-life responses. In addition, 50% of respondents had other eye conditions in addition to their IRD diagnosis, which could confound visual outcomes. Finally, the survey

tool and interview guide used self-reported measures that are not validated.

Conclusion

Canadian patients with IRDs report moderate to severe visual impairment, and both patients and their families report a substantial impact on psychosocial well-being and functioning during daily activities. Vision rehabilitation with a psychosocial approach focused on functional improvement, mental health, personal safety, and quality of life needs to be prioritized alongside facilitating access to emerging treatments.

Supplementary materials

Supplementary material associated with this article can be found in the online version at doi:10.1016/j.jcjo.2022.06.021.

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Footnotes and Disclosure

The authors have no proprietary or commercial interest in any materials discussed in this article.

This study was funded by Fighting Blindness Canada (FBC).

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Originally received Nov. 28, 2021. Final revision Apr. 5, 2022. Accepted Jun. 28, 2022.

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