

# THE IMPACT OF INHERITED RETINAL DISEASES ON CANADIAN PATIENTS: PATIENT INPUT ON LUXTURNA

Fighting Blindness Canada | Canadian Council of the Blind | CNIB Foundation | Vision Loss Rehabilitation Canada

*A Submission of Patient Input for CADTH's Review of Luxturna*

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## 1. SUBMISSION CONTEXT

This submission overviews patient experiences of inherited retinal diseases (IRDs) that were collected through an online survey that went live on March 22, 2020. Developed and hosted by Fighting Blindness Canada (FBC), the sixty-question survey is part of a broader mixed-methods research project titled VIEW IRDs (Valuation and Interpretation of Experiences with Inherited Retinal Diseases) that will include both survey data and qualitative interviews. The project received ethical approval from Advarra, a North American institutional review board with full AAHRPP accreditation. Both the survey and the broader study aim to better understand the physical, psychological, and practical challenges associated with IRDs—it can be considered a “burden of illness” or “quality of life” study—and to highlight the perspectives of Canadians who face these issues on a daily basis. Luxturna is both the first gene therapy for an ophthalmic condition and the first treatment ever for an IRD. Its arrival is a momentous occasion for all IRD patients, even if they are not candidates for the treatment. The study was developed to learn more about the impacts of IRDs, but also to ensure that the assessment of Luxturna is guided by critical insights from the Canadians who will be most affected by CADTH’s decision.

As of April 28, **the survey collected 537 responses from Canadian patients living with a variety of IRDs.** Since the survey will remain open until data saturation is reached, the results presented in this submission can be considered a preliminary look at findings, with a final form produced later this year for publication. This submission also includes two “case studies”: the first borrows from research conducted last year in the United Kingdom and Republic of Ireland—through a project called IRD COUNTS—to understand the socioeconomic implications of IRDs, applying those findings to the Canadian context (p. 17); the second summarizes a conversation that was held between FBC staff and a Canadian parent whose child received Luxturna (p. 19). The goal with the second case study is to provide a tangible and more personal sense of the impact that Luxturna and similar treatments can have on affected individuals. Similar conversations will take place when qualitative interviews are conducted later this year, though these will be more formal in nature.

Finally, this submission contains an appendix in the form of a white paper released earlier this year to mark the significance of the year 2020 for the vision loss community (p. 21). Developed by Fighting Blindness Canada (FBC), the Canadian Council of the Blind (CCB), and the CNIB Foundation, the paper pulls from over 300 survey responses from patients, caregivers, researchers, clinicians, policymakers, and more to capture some of the complexities of living with vision loss in Canada during the symbolic year 2020. It outlines key developments in policy, technology, education, labour, and other areas, providing a wide-ranging look at the challenges faced by partially-sighted Canadians, as well as opportunities for progressive change. The paper can be considered supplemental but is also germane to the review of Luxturna. White papers were also developed on the subjects of “vision research” and “equity and access to care,” both of which can be accessed online.<sup>1</sup>

Regarding the study on IRDs, while FBC is hosting the survey and running the overall project, this submission has been authored jointly by Canada’s largest blindness organizations: Fighting Blindness Canada (FBC), the Canadian Council of the Blind (CCB), the CNIB Foundation, and Vision Loss Rehabilitation Canada (VLRC), all of whom have close ties to the vision loss community and a vested interest in ensuring patient views are comprehensively integrated into the health technology assessment process.

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<sup>1</sup> <https://www.fightingblindness.ca/whitepapers/>

## 2. OVERVIEW OF INHERITED RETINAL DISEASES

Inherited retinal diseases (IRDs) are a group of rare genetic conditions that affect the retina. The result of more than 250 possible genetic mutations, they often entail degenerative vision loss and are accompanied by a range of complex physical, psychological, and economic burdens. While Luxturna treats individuals with biallelic mutations of the RPE65 gene, manifesting as either retinitis pigmentosa (RP) or Leber congenital amaurosis (LCA), a very small patient group compared to the entirety of Canadians with IRDs, this submission contains information on patient views across a broader spectrum of IRD experience. This includes RP and LCA, including those with relevant RPE65 mutations, but also choroideremia, Stargardt disease, Usher syndrome, and others. Though each disease is unique, both in terms of pathophysiology and patient experience, there are strong similarities that warrant a broader, more inclusive look at the IRD category when reviewing new treatments. It is also the case that, while Luxturna may be the first gene therapy for an ophthalmic condition, the pipeline in this area is robust, and the diseases and genotypes addressed by emerging treatments will be numerous. Our hope is that this submission will lay productive groundwork for the review of Luxturna, but also for the consideration of the many new therapies that we know are in development.

That said, **75% of our survey group specified having RP (71%) or LCA (4%),<sup>2</sup> and 7 individuals indicated an underlying mutation of the RPE65 gene identified through genetic testing.** Below are brief summaries of RP and LCA, both of which have been modified from FBC's online resource for Canadians living with eye diseases, Vision Care Pathways:

### Retinitis Pigmentosa

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Retinitis pigmentosa (RP) describes a group of genetic disorders that damage light-sensitive cells in the retina, leading to gradual vision loss over time as the cells die off. While the condition is classified as a "rare disease," it is one of the most common inherited diseases of the retina, with its combined total of possible mutations—not just RPE65—affecting approximately 1 in 4000 Canadians.<sup>3</sup> RP is often referred to as an "inherited retinal disease," meaning that it is passed along genetic lines and inherited from one's parents. Though it is usually diagnosed during childhood or adolescence, a minority of patients report symptoms later in life.

Specialized cells called photoreceptors are responsible for absorbing light and translating it into signals that are interpreted by the brain—it is these essential cells that gradually die off as a result of RP. There are two types of photoreceptor cells: rods and cones. Rod photoreceptors are responsible for peripheral and night vision, while cone photoreceptors are responsible for central, high-acuity vision as well as detail and colour. Since it is the rod cells that are first damaged by RP, peripheral and night vision are affected during the early stages of the disease, followed by a narrowing of the visual field, often referred to as a progressive form of "tunnel vision." The death of rod cells eventually affects the cone cells as well, leading to the loss of central vision and often resulting, during the later stages of the disease, in near or total blindness. The length of this process varies from individual to individual.

RP was originally considered a single disease, but after decades of research—including research funded by FBC—we now know that there are several forms of RP, and that these forms can be caused by mutations in more than

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<sup>2</sup> Percentages in this submission are rounded to the nearest whole number, unless they fall under 1%.

<sup>3</sup> <https://www.nei.nih.gov/learn-about-eye-health/eye-conditions-and-diseases/retinitis-pigmentosa>

64 genes. The gene or genes affected determine the disease type and symptoms. There are several different ways that RP can be inherited, which is usually described as the “inheritance pattern.” The different RP inheritance patterns include: autosomal dominant, autosomal recessive, and x-linked recessive. A genetic counsellor will usually discuss family history with the patient in order to determine which of these patterns is associated with the patient’s vision loss. With this information, the genetic counsellor may be able to provide a sense of how the patient’s condition will progress, and give the patient’s family information about the risks of vision loss for other family members.

Different genetic mutations can damage the retina or impair its function in different ways; for example, some mutations affect how the retina processes nutrients, while others damage the photoreceptors. It’s important to identify the specific gene and mutation, because many treatments being developed for RP will be gene or even mutation specific.

### **Leber Congenital Amaurosis**

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Leber congenital amaurosis (LCA) is a genetic disorder that causes severe visual impairment at birth or in early childhood. It was first outlined in a paper published in 1869 by the German ophthalmologist Theodor Karl Gustav von Leber, which is where the disease gets its name. Leber originally suggested that the disease is a rare form of retinitis pigmentosa (RP), and that continues to be accepted today. LCA is unique in several ways, however: the symptoms are often noticed in the first weeks or months after a child’s birth, and vision loss is more rapid and severe, as opposed to the slow progression associated with other forms of RP. The disease is less prevalent than RP as well, affecting approximately 2 to 3 per 100,000 newborns.<sup>4</sup>

LCA is also an inherited or genetic disease caused by mutations in one of at least 17 different genes. Depending on which gene or genes are mutated, different characteristics of the retina will be affected, but in all cases the retina’s ability to develop and function properly is inhibited: the photoreceptors, the light-sensitive retinal cells that convert light into messages for the brain, are no longer sending electrical signals correctly, resulting in severe visual impairment.

## **3. SURVEY RESULTS and ANALYSIS**

### **Diseases and Demographics**

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**As mentioned above, 71% of the survey group (n=537) reported having RP, while 4% reported LCA. Seven individuals (6 with LCA, 1 with RP) indicated that their disease is a result of a mutation of the RPE65 gene, though neither biallelic nor monoallelic were specified at this time.** The remainder of respondents self-identified as having Usher syndrome (8%), Stargardt disease (7%), cone-rod dystrophy (4%), cone dystrophy (3%), Leber’s hereditary optic neuropathy (3%), rod-cone dystrophy (2%), choroideremia (2%), x-lined retinoschisis (0.89%), Bardet-Biedl syndrome (0.67%), Best disease (0.44%), achromatopsia (0.22%), with the remainder (5%) listing diseases not specified in the survey, including “x-linked retinitis pigmentosa,” “late onset retinal dystrophy,” and “autoimmune retinopathy.”

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<sup>4</sup> <https://ghr.nlm.nih.gov/condition/leber-congenital-amaurosis#statistics>

Disease breakdown of respondents (%)

RP	LCA	US	SD	CRD	CD	LHON	RCD	C	XLR	BBS	BD	A	Other
71	4	8	7	4	3	3	2	2	0.89	0.67	0.44	0.22	5

Respondents were able to select multiple diseases, which is why the total percentage of diseases is above 100%. It was decided that this would provide flexibility in the way respondents describe their conditions—for instance, since Usher syndrome involves a form of retinitis pigmentosa (RP combined with hearing loss), some individuals selected both.

Many respondents also indicated that they live with one or more **eye-related comorbidities**: 34% of the group indicated cataracts, followed by glaucoma (7%), macular degeneration (5%), diabetic macular edema (0.51%), and diabetic retinopathy (0.26%), as well as 15% selecting “other” and providing conditions such as “stigmatism,” “lazy eye,” “keratoconus,” “dry eyes,” “pigment patch,” “born blind in right eye,” and “nystagmus.” Seeing as ocular comorbidities were self-reported in just under 50% of the overall group, it is clear that the presence of an IRD carries a significant chance of existing alongside additional ocular complications, at least for the individuals surveyed. As a result, it is clear that from a disease perspective, an IRD cannot be viewed in isolation, but rather as a single—albeit strong—factor in one’s overall visual health experience. We know from existing research that the presence of comorbidities often leads to more demanding and complex care, as well as higher levels of health systems navigation.<sup>5</sup>

In terms of **geographic distribution**, 48% of respondents specified being located in Ontario. The remainder indicated British Columbia (17%), Alberta (14%), Quebec (6%), Nova Scotia (4%), Saskatchewan (3%), Manitoba (3%), New Brunswick (1%), Newfoundland (1%), and PEI (0.68%).

Geographical breakdown of respondents (%)

ON	BC	AB	QC	NS	SK	MB	NB	NL	PE	NU	NT	YT
48	17	14	6	4	3	3	1	1	0.68	0	0	0

As shown in past submissions to CADTH, the higher concentration of respondents in ON, AB, and BC speaks to population, certainly, but also to difficulties accessing and surveying patients in less populated or urbanized areas without the use of in-person engagement at clinics and other strategies. This is an overall challenge to survey-based representativeness that has been encountered by many researchers, one that we will attempt to address through our targeting for qualitative interviews. Though the survey was offered in French, low numbers in Quebec illustrate our limited reach as organizations in that province.

In terms of **general demographic details**, the average (mean) age for the group is 51 years old. The majority of respondents indicated a biological sex of “female” (54%), the remainder “male,” while 54% of the total group indicated “female” as a current gender identity, the rest “male” (45%) and “other” (0.87%). Responding to a question about the highest level of education completed, respondents selected bachelor’s degree (26%), “some college credit, no degree” (15%), master’s degree (14%), high school degree (11%), trade/technical/vocational training (10%), associate degree (7%), professional degree (6%), doctoral degree (4%), “some high school, no diploma” (4%), “no school completed” (2%), and “nursery school to 8th grade” (2%).

<sup>5</sup> Pinazo-Durán M, Zanón-Moreno V, García-Medina J, Arévalo J, Gallego-Pinazo R, and C Nucci. Eclectic Ocular Comorbidities and Systemic Diseases with Eye Involvement: A Review. *BioMed Research International*. 2016: 1-10. <https://doi.org/10.1155/2016/6215745>.

## Vision Loss, Genetics, and Family History

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This section of the survey was designed to learn more about patients' overall experience of vision loss, as well as genetic testing and known genetic mutations. It also asks questions about knowledge of disease heredity.

The lack of night vision stood out when respondents were asked **what statement best describes their current vision**: with the option to choose multiple responses, 41% selected "no night vision." This is unsurprising, given that in RP and other IRDs the loss of night vision is a common symptom of the deterioration of rod photoreceptors, which are typically damaged before cone photoreceptors begin to die off. But responses also made it clear that those with IRDs have other ways of describing their visual experience: 34% of the group selected "good central vision," 32% selected "some useful central vision," 27% selected "some useful peripheral vision," 14% selected "some light perception," 10% selected "I still have good overall vision," 7% selected "good peripheral vision," 5% selected "no vision," and 3% selected "shadows only."

It is important to note, however, that among the 19 participants diagnosed with LCA, including the six individuals with RPE65 mutations, responses skewed more heavily towards a significant loss of visual perception in some areas. This is unsurprising, since LCA typically involves childhood onset and rapid visual deterioration. For example, 11% of respondents indicated "shadows only" (compared to 3% for the entire group) and 26% indicated "some light perception" (compared to 14% for the entire group).

Despite some indication of maintained visual acuity in overall responses—"good central vision," for example—67% of the overall group indicated being **diagnosed as legally blind** (31% indicated they are not legally blind, while 4% selected "other"). When asked **how they would describe their vision**, the largest number of respondents selected "moderate-low vision" (42%), followed by "severe-low vision" (30%), "near normal vision" (15%), "near total blindness (some light perception)" (10%), and "total blindness (no light perception)" (2%). **And here again, patients with LCA diagnoses skewed more severe in terms of vision loss**: 95% indicated being diagnosed as legally blind (compared to 67% for the overall group); and most respondents selected "severe-low vision" to describe their vision (60% compared to 30%), followed by "near total blindness (some light perception)" (21% compared to 15%), "moderate-low vision" (16% compared to 42%), and "near-normal vision" (5% compared to 15%).

When asked if they have ever **received a genetic test**, 60% of the overall group selected that they have, while 36% said "no" and 4% said "I don't know." The largest percentage of respondents indicated that they have never **met with a genetic counsellor** (35%), followed by once (24%), twice (20%), "three or more" (12%), and "I don't know/I don't remember" (8%). While genetic testing is now covered by all public health plans across Canada, there is a well-known shortage of genetic counsellors and related infrastructure, a likely factor in the large number of respondents not receiving tests or meeting with counsellors. Equipping the Canadian IRD community for the future of ocular treatments will undoubtedly involve addressing these issues.

Unsurprisingly, the percentage of respondents who are **aware of their underlying genetic mutation** adheres closely to the percentage who have received a genetic test: 63% of the overall group selected "yes" when asked if they know their mutation. Percentages for specific mutations are difficult to determine, since by necessity an open field was provided for answers. At the same time, many who selected "yes" did not fill in the actual mutation, probably because they could not recall it; indeed, some respondents provided answers such as "can't

remember” and “can’t find the document but both parents had to be a carrier.” Analysis of the indicated mutations will be completed by the time this study is submitted for publication.

When asked if they are **aware of any family members receiving genetic testing** for IRDs, 49% of the overall group responded with “no,” 41% said “yes,” and 10% said “I don’t know.” Respondents were also asked about **how many family members are affected by the same IRD that they have been diagnosed with**: most said 0 (38%), followed by 1 (21%), “5 or more” (14%), 2 (9%), “I don’t know” (7%), 3 (6%), and 4 (5%). When given the chance to **tell us something about genetic testing experiences or family history** in an open-ended field, respondents gave a range of experiences that demonstrate the complexity of these subjects. For instance, many patients emphasized the difficulty of accessing genetic testing services or receiving conclusive results:

- “I would like to be tested, but am having a hard time getting linked with a geneticist. There has to be a better system in place to make testing more available to people with IRDs.”
- “Receiving a diagnosis is not easy.”
- “It’s almost impossible to get a genetic test! I’ve tried several times.”
- “No one in my family has it. Still on the waiting list for genetics.”
- “Testing was inconclusive.”
- “After genetic testing, results are inconclusive.”

Many patients also took the question as an opportunity to detail their own knowledge of family history, including gaps or blind spots in that knowledge. What stands out in these responses is that neither family histories nor genetic testing are reliable resources for confirming an affected gene. Contemporary genetic testing is of course quite accurate, but in Canada the service is unreliable for many individuals due to barriers to access. This leaves many in the IRD community with increased anxiety and uncertainty: many don’t know where their disease comes from, and without a confirmed gene they do not know how it will impact their own families. This is an excessive burden placed on the shoulders of individuals who are already managing the complexities of their own vision loss.

## Healthcare, Support Services, and Clinical Trials

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To manage their vision loss and seek better care and potential treatments, the IRD community in Canada accesses a variety of health services and infrastructure. This section of the survey asks questions about those services and resources, including clinical trials.

When asked **which healthcare providers are accessed** to manage their IRDs (and given the chance to select multiple), most respondents chose “ophthalmologist” (66%). This was followed by “optometrist/optician” (42%), “I have not seen any healthcare providers in the last year” (19%), “family physician” (17%), “other” (11%), “habilitation, rehabilitation, or low vision specialist or service” (10%), “genetic counsellor” (10%), “sight support volunteer” (5%), “psychologist, psychiatrist, or counsellor” (5%), “specialist outpatient clinic or unit” (5%), “occupational therapist” (3%), and “admitted to hospital as inpatient” (0.26%). When asked to specify **how often they see their specified healthcare providers**, most chose “yearly” (56%), followed by “every 2 to 6 months” (22%), “every 2 years” (14%), “rarely/never” (5%), “monthly” (3%), and “weekly” (2%). When asked about their **level of satisfaction with the care they receive from these providers**, most selected “very satisfied”

(37%), followed by “satisfied” (33%), “neither satisfied nor unsatisfied” (20%), “dissatisfied” (7%), and “very dissatisfied” (2%).

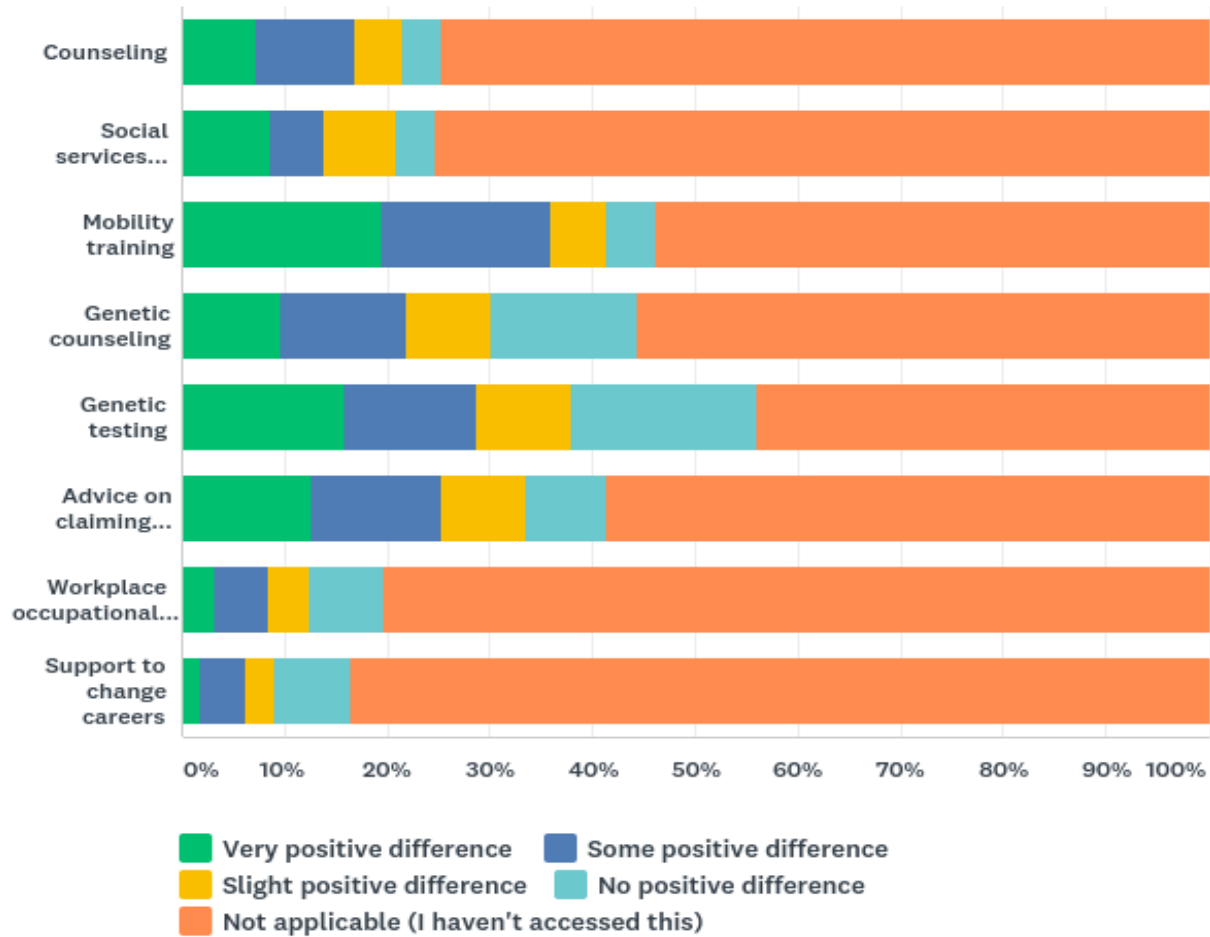
Clearly, Canadians with IRDs are accessing a wide-range of healthcare providers, and their experiences tend to be largely positive. They also meet with their providers on a relatively frequent basis, at least in the context of rare diseases with no existing treatment options. A potential care-gap emerges in the area of mental health, however. When asked **whether their eye doctors ask them about their mental well-being** during appointments, most selected “no” (70%), followed by “yes, at every appointment” (15%) and “yes, at some of my appointments” (15%). It is unrealistic to expect that eye specialists and primary care providers can simultaneously provide psychological care and counselling—nonetheless, these responses show that emotional and psychological wellness may not be addressed as often as they should be within clinical settings. And as we have already seen (and will continue to see in following sections), IRDs are psychologically burdensome, especially when tied to unpredictable sight loss (in many cases vision loss is certain, but the rate of that loss is unknown), uncertainty regarding the affected gene, and anticipated effects on family members.

Considering the lack of existing treatments for these diseases, clinical trials are a particularly important topic for the IRD community. That said, when asked if they are **aware of any clinic trails or other research specific to their IRDs**, most respondents selected “no” (59%). Considering the explosion of clinical trials for IRDs over the last several years, this does not necessarily suggest that relevant clinical trials do not exist; instead, a lack of familiarity with the clinical trial landscape could be a strong factor here, especially because we know how confounding and inaccessible that landscape can be for many patients. At the same time, only a very small percentage of respondents indicated having actually **participated in a clinical trial for their disease**: only 5% said they had. To what degree this correlates to various factors—to a lack of clinical trial infrastructure in Canada, or to access/barriers to non-Canadian clinical trials, or to a lack of information/understanding about clinical trials—requires additional insights and analysis. We hope to explore these subjects in more detail during interviews with the respondents.

To dive more deeply into the issue of support services, our survey asked participants to **rate the value of a number of different services they have accessed**, including counseling, social services, mobility training, genetic counselling, genetic testing, advice on claiming benefits, workplace occupational health support, and support to change careers. Below is a chart that outlines the array of experiences with each service, ranging from “very positive difference” at the high end to “no positive difference” at the low.



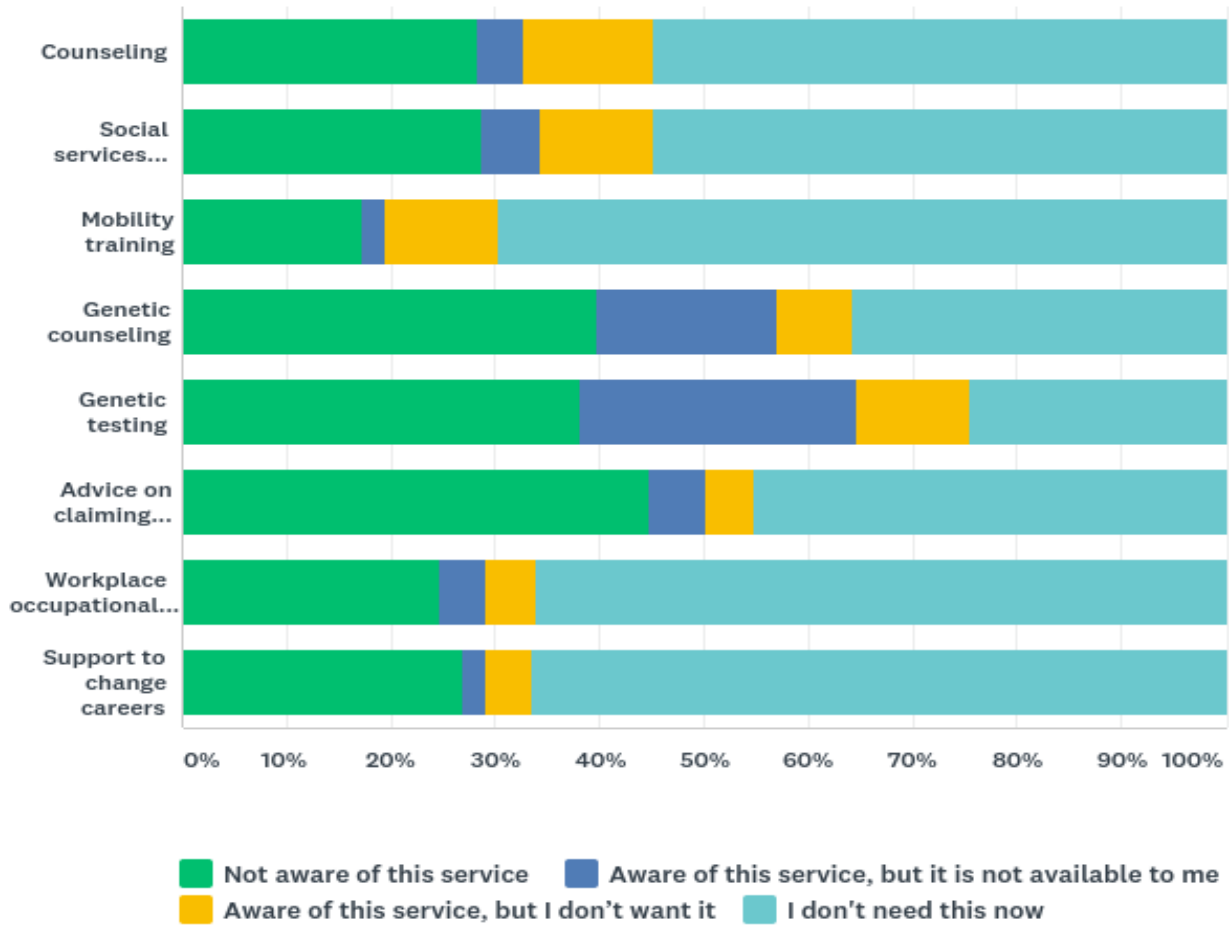
Positive and negative experiences with support services:



Interestingly, the vast majority of respondents have not accessed these services, shown by the longer orange bars that represent “not applicable (I haven’t accessed this).” For those who have, their experiences tend to vary dramatically. In the case of mobility training, for example, while 54% of the overall group have not accessed the service, those who have had largely seen a “very positive difference” (20%) or “some positive difference” (16%). In the areas of genetic counselling and genetic testing, on the other hand, the larger responses fall on the negative side: for genetic counselling, 14% selected “no positive difference,” while 18% selected the same for genetic testing. In the areas of workplace occupational health support and support to change careers, the negative trend is carried forward, with “no positive difference” selected by the largest percentage of respondents who have accessed the service. In both cases, however, very large percentages of respondents had no experience with the service: 80% for workplace occupational health support and 85% for support to change careers.

In a follow-up question, we asked about why certain services have not been accessed, allowing respondents to select “not aware of this service,” “aware of this service, but it is not applicable to me,” “aware of this service, but I don’t want it,” and “I don’t need this now.” This is useful information, especially because most respondents indicated not accessing each service.

Reasons for not accessing support services:



Most participants indicated not accessing a service because “I don’t need this now” (represented by the light blue bars), especially in relation to “mobility training” (70% of the group that did not access the service), “support to change careers” (67%), and “workplace occupational health support” (66%). A lack of awareness of the service was selected frequently as well, especially for “advice on claiming benefits” (45%), “genetic counselling” (40%), and “genetic testing” (38%). It is clear in these instances, as well as in relation to the other services, a lack of awareness is a key driver in a service not being accessed. Considering the fact that these are online-savvy participants who are connected to our organizations, interested in relevant news, etc., this is likely connected to a lack of available or digestible information regarding such services, or perhaps with a lack of interest in support services compared to emerging treatments and other topics.

**Impact on Daily Life**

It would be a mistake to conclude that the lack of a strong connection to support services such as counselling and occupational support is associated with a low impact of IRDs on daily life. In fact, responses to this section of the survey, which was designed to learn more about daily challenges and impacts, show that IRDs alter the lives of patients in ways that are varied and multifaceted.

For instance, when asked **whether their IRD has affected their employment or school status**, 54% of the study cohort said “yes.” When asked to specify why, they provided insights that demonstrate how challenging it can be to manage degenerative vision loss and also be successful in work or school settings. Respondents had the following to say:

- “I quit my master’s degree because I couldn’t keep it up. I was a mechanical engineer. With my first job in Montreal, I was demoted from an office job an operative job. I haven’t been able to find a job since 2017, and I am a musician and now trying to do it on my own, which has been hard and time consuming without any compensation or payments. I have my wife that helps me with everything but I haven’t been able to find a job that I can work at.”
- “I had to change my schooling with reduced income and couldn’t finish school as fast as other students due to barriers in the classrooms. I lost jobs and still continue to deal with discrimination with employers which has only magnified during the pandemic.”
- “I worked for 10 years in healthcare and earned a master’s degree but quit without counselling (gave up disability benefits and pension) because of fear that I couldn’t fulfill my role or find a suitable alternative.”
- “I had to leave my movie industry job due to RP. It’s been a tough slog ever since.”
- “I required assistive technology to complete all my schooling. Then, after university, many barriers to employment exist. As a result, I found a job 16 years after graduating university and it was nothing related to my university studies.”
- “I struggled all through elementary school and junior high. It wasn’t until grade 9 when I got my first CCTV and computer with a screen reader and magnifier that I finally could do my own homework without help. I got taken out of gym class instead of having adapted activities, and this affected by weight, which has become a lifelong struggle. At work, I have been discriminated against. I have failed interviews when I have used adaptive technology to complete them.”

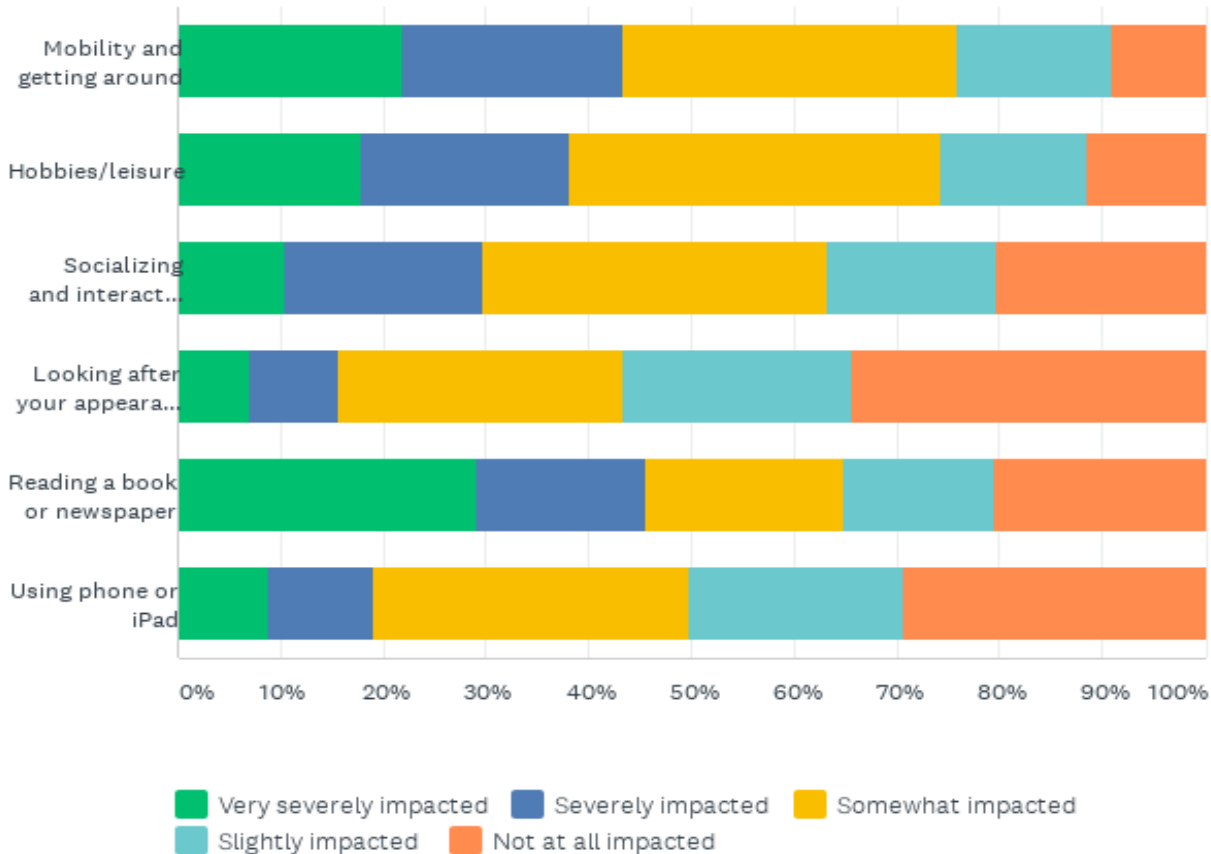
These responses—as well as many others not listed—make it clear that IRDs can strongly influence one’s experiences at school and work. This is not always the case, of course, but for those who did indicate a negative impact, the affect tended to be severe. This is supported by responses to a follow-up question. When the entire group was asked to **rate the impact of their disease on their ability to perform job or school responsibilities**, the largest number indicated the highest end of the scale: “10 very severe impact” (22%). The remainder of the cohort selected 5 (12%), 7(11%), 3 (11%), 1 (11%), 8 (9%), 6 (8%), 4 (6%), 2 (6%), and 9 (5%).

While support services may not be widely utilized by the IRD community, this is not the case with modifications or aids such as canes, magnifiers, and specialized laptops. When asked to select **the modifications or aids they utilize**, the largest percentage of the overall group indicated “cane” (45%), followed by magnifiers (42%), “books, including books with enlarged font and audiobooks” (32%), “modifications to mobile phones such as applications” (28%), modified laptops (27%), “I don’t use any additional items because I don’t need them” (22%), screen readers (21%), ergonomic adaptations (11%), “I don’t use any additional items because I don’t have access to them” (5%), and braille (2%). Many respondents also selected “other” (19%), providing answers such as “flashlight,” “Google home,” “e-sight device,” “PenFriend mic,” and “Plextalk recorder.”

These devices and aids are undoubtedly used to navigate some of the challenges of work and school that have been outlined; at the same time, many of them—a flashlight, for instance—are clearly helpful in more

“ordinary” or day-to-day contexts. We asked respondents to tell us about their difficulties in these settings by indicating **how much their eyesight interferes with a range of activities**, including “mobility and getting around,” “hobbies/leisure,” “socializing and interacting with others,” “looking after your appearance,” “reading a book or a newspaper,” and “using phone or iPad.”

Degrees to which eyesight interferes with activities:



For each activity, the majority of the overall group indicated some degree of interference, with “reading a book or newspapers” being the most severely impacted activity (29% selected “very severely impacted”). In terms of mobility, the largest group chose “somewhat impacted” (33%), but only 9% chose “not at all impacted,” showing that challenges in this area are pervasive, even if there is a wide degree of severity.

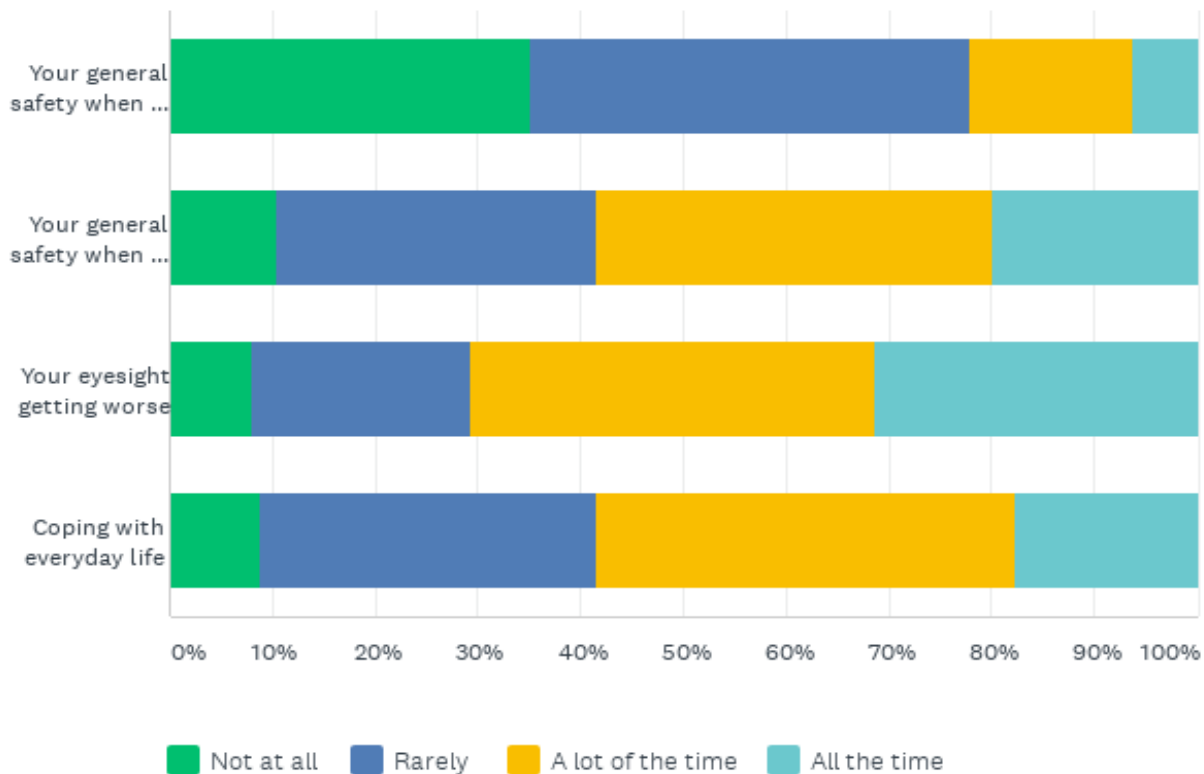
By and large, then, daily activities are challenging for the participants we surveyed. At the same time, they indicated that other, more specific areas of their lives can be challenging as well. When asked to select **what they find challenging about their IRD** and given the chance to select multiple, a large majority of respondents selected “worry that my condition might worsen in the future” (76%). This was followed by “not being able to do the daily activities I used to do” (61%), “the effect on my ability to work” (50%), “socializing” (47%), “having meaningful work and/or educational experiences” (35%), “lack of social support” (26%), and “the long wait times for appointments” (16%). 23% of respondents selected “other,” providing insights such as “uncertainty about future and the effect on activities and safety,” “seeing my child,” “raising my children,” “slow progress for a cure,” “everything is challenging,” and “difficulty dating.” These responses present a wide range of challenges,

and they show just how completely an IRD can impact one’s life, affecting not only obvious activities such as work, but the specific details of personal and social existence—seeing loved ones, dating, and more.

Also, the fact that most participants indicated that they worry about their condition worsening suggests a significant emotional or psychological strain. This concern is persistent and prolonged, seeing as it stems from a disease that progresses over years and decades. Our respondents provided responses that show this kind of burden in other areas as well. When asked **how often they think about their IRD and/or vision loss**, respondents selected “a lot (once per day)” (35%), “all the time (multiple times per day)” (31%), and “a little (once a week)” (30%). Only 3% of participants indicated “not at all.” IRDs are physically challenging, then, but they present a considerable psychological burden as well, one that we can assume is often connected to the anxiety over worsening sight that was mentioned above.

To explore this notion further, we asked participants to specify **how often their eyesight makes them concerned about specific issues** over the course of an average month.<sup>6</sup> Issues included “your general safety when in your home,” “your general safety when out of your home,” “your eyesight getting worse,” and “coping with everyday life.”

Degrees of concern over eyesight in relation to certain issues:



Again, responses emphasized the notion of worsening eyesight, with 40% of the group indicating that they are concerned “a lot of the time” over an average month, and 31% expressing concern “all the time” over the same

<sup>6</sup> All timeframes in the survey were abstracted in this way—i.e. general or average periods as opposed to recent or specific ones—to avoid a situation where the unusual circumstances of the COVID-19 crisis skew responses.

period. The notion of “coping with everyday life” also stands out: 41% of the group selected that they are concerned “a lot of the time.” Similarly, 38% of the group expressed the same level of concern—“a lot”—in relation to the idea of “your general safety when out of your home.” The picture that emerges in these responses is of a patient group that is prone to worry and anxiety in relation to daily coping, leaving the home, and especially the progressive loss of vision. All of this amounts to a considerable psychological impact for many in the IRD community.

An emphasis on anxiety was also clear when we asked **which emotions and circumstances the group has experienced as a result of their IRDs**: 71% of the group selected “anxiety,” and 73% of the group selected “stress,” which is closely related. Other selected responses included fear (64%), anger (62%), loss of confidence (58%), isolation (47%), employment barriers (43%), loneliness (41%), discrimination (37%), and lack of self-worth (34%). It is worth noting that “employment barriers” and “discrimination” are emphasized here once again; they also stood out in responses to the question about work or school status. Also worth noting is that “loss of confidence,” “loneliness,” and “lack of self-worth” can be seen as overlapping concepts, all contributing to a general sense of depression and a negative impact on mental health. A significant amount of research has already been done to show that there is a strong link between vision loss and mental health issues.<sup>7,8,9</sup> This is borne out in our survey results, with many respondents showing that they have suffered emotionally and psychologically as a result of their IRDs.

Psychological effects are not endured in isolation. Instead, they ripple outwards to impact how one feels and acts within a broader social setting; we explored this social quality in our survey in a number of ways. When asked how their **IRD has affected their social life**, for instance, most respondents indicated that it has had an effect: 50% selected “it has a slightly negative effect,” while 24% selected “it has a very negative effect.” The remainder of the cohort (25%), selected that “it has not affected my social life.” A number of respondents also provided added clarification through open-ended responses. One participant explained that “Everything is linked. I don’t have a job so I don’t have contact with many people. My social life has been affected by it.” Another wrote that “I’ve withdrawn from a lot of relationships and activities due to difficulty with transportation and not feeling safe to go out when it’s dark.” Others described how they have managed to maintain a robust social life as their vision degrades, but it is clear that for those who are struggling in this area, the dangers of anxiety, isolation, loneliness, and depression are acute. Many respondents touched on this directly, such as the individual who wrote “depression and anxiety. Can’t have a driver’s license so I’m isolated.”

Family members are of course an integral part of one’s social circle, and we saw that IRDs can have negative effects here as well. When asked **whether their IRD has affected their family**, most indicated that “it has a slightly negative effect” (46%), followed by “it has not affected my family” (35%) and “it has a very negative effect” (16%). Some open-ended responses picked up the theme of concern over genetic inheritance (“my

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<sup>7</sup> Zhang X, Bullard K, Cotch M, Wilson M, B Rovner, *et al.* Association between depression and functional vision loss in persons 20 years of age or older in the United States, NHANES 2005-2008. *JAMA Ophthalmol.* 2013; 131.5: 573-81. doi: 10.1001/jamaophthalmol.2013.2597.

<sup>8</sup> Kempen G, Balleman J, Ranchor A, van Rens G, and G Zijlstra. The impact of low vision on activities of daily living, symptoms of depression, feelings of anxiety and social support in community-living older adults seeking vision rehabilitation services. *Quality of Life Research.* 2012; 21: 1405-1411. <http://dx.doi.org/10.1007/s11136-011-0103-5>.

<sup>9</sup> Bernbaum M, Albert S, Duckro P, and W Merkel. Personal and family stress in individuals with diabetes and vision loss. *Psychodynamics and Psychopathology.* 1993; 49.5. [https://doi.org/10.1002/1097-4679\(199309\)49:5<670::AID-JCLP2270490509>3.0.CO;2-F](https://doi.org/10.1002/1097-4679(199309)49:5<670::AID-JCLP2270490509>3.0.CO;2-F).

daughters are a little worried about the possibility of also having RP” and “it is inherited and passed down maternally therefore will likely continue to manifest itself in our family”), while others articulated a more generalized apprehensiveness about the future, a theme we have seen in other parts of the survey (“Worry about the future” and “Mostly just the worry about the future since we know his vision will worsen perhaps to blindness”).

We also asked respondents about **discrimination and stigma they may have experienced**. When asked if they have been treated differently as a result of their IRD, many indicated that they have, responding “yes, by others” (42%) and “yes, by my employer” (16%). When specifying “other,” some respondents indicated that individuals in the educational system have treated them differently, usually in a negative fashion; some wrote about co-workers and others in the workplace; and others described experiences with “strangers” or the “general public.” It is clear in these and other responses that the experience of stigma can be pervasive for some, extending from members of their closest social circles to the public at large. Once more, there were individuals without any negative experiences in this area, but for those who face challenges and barriers when it comes to the perceptions of others, it can exact a difficult and complex toll, one that is not easily managed.

## Treatment Goals

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Considering that many with an IRD experience emotional or psychological burdens—whether these are connected to fear over sight loss, concern for family members, social isolation, discrimination, or other experiences—it is important to understand how these feelings relate to perceptions about emerging or potential treatments. With this in mind, we asked participants to describe **how their emotional well-being would change if there was a new treatment for their IRD that met their treatment goals**. Perhaps unsurprisingly, most indicated that “my emotional well-being would improve significantly” (74%); the remainder of the cohort selected “my emotional well-being would improve slightly” (20%) and “my emotional well-being would not change” (6%).

Of course, treatment goals differ from individual to individual. We asked the cohort to specify the **goals they have in mind for the treatment of their IRD**, with the chance to select multiple. The largest group of respondents expressed that they would like a treatment to “improve my sight overall” (85%), followed by “cure my condition” (66%), “improve my night vision” (59%), “improve my mobility at night” (52%), “allow me to participate in social activities” (49%), “allow me to enjoy my personal relationships” (36%), “allow me to work” (29%), and “allow me to go to school” (9%). A number of respondents also chose “other” (22%), with some providing a sense that a stabilization of vision would be highly desirable: for instance, “even just stop the progression if it can’t be reversed,” “stabilize my vision so it doesn’t get worse,” “happy if it does not get worse,” “keep my vision from getting worse,” and “even if there was a way to just halt the progress of my RP from where it is now, I would be thrilled.” We know from clinical data that has been released that Luxturna is not a cure for blindness. Instead, it shows efficacy in halting vision loss for many individuals. These and other testimonials suggest that a treatment option that can do this, halt a progressive decline in sight, is highly desirable in this patient community.

At the same time, we know that night vision is significantly improved for patients receiving Luxturna. To gauge their level of interest in a treatment option that addresses night vision, we asked respondents **if their quality of life would be improved by a treatment that only improves their night vision and mobility at night**. Most

selected that it would, that either “my quality of life would improve significantly” (58%) or “my quality of life would improve slightly” (32%), while the remainder specified that “my quality of life would not change” (11%). The responses demonstrate that improvements in night vision and mobility are considered valuable for the vast majority of IRD patients, even if they are not accompanied by improvements or effects in other areas.

## Summary

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Responses to our survey paint a detailed picture of the IRD community in Canada. The data show that many in the group are living with decreased night and peripheral vision, and that visual impairment tends to be more severe in patients with LCA, including those with RPE65 mutations. At the same time, it appears that a majority of IRD patients in Canada are considered legally blind, and since their diseases are inherited and involve progressive vision loss, they are often concerned about the impact on their families and the looming possibility of blindness, demonstrating a considerable psychological burden. Many in the community—over half of our surveyed group—have received genetic testing and are aware of their mutation, though far fewer individuals have met with a genetic counsellor.

Unsurprisingly, most Canadians with IRDs see an ophthalmologist to help manage their disease, and in other cases an optometrist or optician. Regardless of the provider, patients tend to be satisfied with the care they receive, though mental health considerations do not often come up during these interactions. Regarding clinical trials, a significant majority of our group have not participated in one, though a large number—roughly half—have heard about trials that are relevant to their disease.

The community does not rely heavily on support services such as counselling, social services, and mobility training, either because they are not aware of such services or do not think they would benefit from them. For those who do access support services, however, their experiences are largely positive. Genetic testing and genetic counseling are exceptions in this regard, since many struggle to access these resources in the first place. Although support services are not widely utilized, specialized aids and modifications are used by a majority of the group, including canes, audiobooks, magnifiers, phones, laptops, and other assistive and adaptive technologies.

Responses to our survey show that people living with vision loss due to IRDs experience a great degree of stress and other negative experiences during “normal” times. At the same time, a study recently conducted by the CCB, “the Impact of the COVID-19 Pandemic on Canadians Who are Blind, Deaf-Blind, and Partially-Sighted,” revealed that people with vision loss are suffering from significant additional stress due to COVID-19, and have become more isolated and lonely than usual.<sup>10</sup> In short, the overall impact of COVID-19 has been to amplify the difficulties of living with vision loss. It is fully expected that the pandemic will have a lasting impact on the vision loss community, and that additional stress will make life even more challenging for people with IRDs. One of the issues identified in the CCB study, for example, is that the pandemic has made it difficult for many patients to see their eye doctors, resulting in concern that they may lose more vision as a result.

But even outside of the extenuating circumstances of COVID-19, IRDs impact the daily lives of Canadians in a number of ways. Almost half of the surveyed group told us that their progress at work or school has been

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<sup>10</sup> <http://ccbnational.net/shaggy/2020/05/05/covid-19-survey-results/>



affected negatively by their condition, with a large portion of these individuals specifying a severe impact on their ability to be successful in these contexts. Many of those who live with an IRD believe that their disease makes common or day-to-day activities difficult as well, especially reading, general mobility, and leisure activities. Combined with concerns over family members and worsening eyesight, these challenges are suggestive of not only a physical but an emotional burden. This notion is supported by the fact that many in the IRD community think about their disease often, showing that it intrudes frequently into their psychological lives. Anxiety was flagged as a significant factor in relation to this, as well as stress, fear, anger, and other emotions. These are issues that extend beyond one's isolated mental health as well, with many in the group experiencing a negative impact on their social lives and the lives of their family members, though the degree varies in both cases.

If a treatment were to emerge, most Canadians we surveyed believe their emotional well-being would improve significantly, especially if the treatment were to recover some overall sight, cure the condition entirely, or improve night vision and mobility at night. In fact, even if a treatment only enhanced vision and mobility at night, most Canadians from our group believe their overall quality of life would change for the better.

#### **4. CASE STUDY 1: THE SOCIOECONOMIC IMPLICATIONS OF INHERITED RETINAL DISEASES (IRD COUNTS)**

Our survey results provide insights into the physical, psychological, and social impacts of IRDs. The data are unable, however, to tell us anything about the economic impacts of this group of diseases, since socioeconomic analysis is outside of the study's scope. That said, Fighting Blindness Canada has partnered with other organizations, including Retina International and the Foundation Fighting Blindness in the U.S., to launch a multi-country investigation into the economic effects of IRDs.

Retina International completed a first stage of this project last year, focused on the U.K. and Republic of Ireland, called IRD COUNTS and conducted by Deloitte Access Economics: reports on both the U.K.<sup>11</sup> and ROI<sup>12</sup> were released last year, followed by a publication in *Clinical Ophthalmology* this year.<sup>13</sup> Launching later this year, this second stage of the endeavor will extend that work into Canada and the U.S., providing a much-needed assessment of the various cost impacts of IRDs, both at individual and national levels. Of course, that assessment will not be concluded in time for CADTH's review of Luxturna. It is fair, however, to look towards the IRD COUNTS numbers from the U.K., and country that provides a fair comparator in terms of health care systems and GDP per capita, for a projected sense of the cost burden of IRDs for Canada's economy.

The socioeconomics of IRDs must be anchored in an accurate picture of prevalence, and Deloitte provided as much for the U.K. last year, producing an estimated number of cases for the "major" IRDs:

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<sup>11</sup> <http://www.retina-international.org/wp-content/uploads/2019/11/cost-of-illness-report-uk.pdf>

<sup>12</sup> <http://www.retina-international.org/wp-content/uploads/2019/11/cost-of-illness-final-report-roi.pdf>

<sup>13</sup> Galvin O, Chi G, Brady L, Hippert C, Del Valle Rubido M, *et al.* The Impact of Inherited Retinal Diseases in the Republic of Ireland (ROI) and the United Kingdom (UK) from a Cost-of-Illness Perspective. *Clin Ophthalmol.* 2020; 14: 707-719. <https://doi.org/10.2147/OPHTH.S241928>.

Table 5.1: Prevalence of IRDs in the UK (2019) by condition

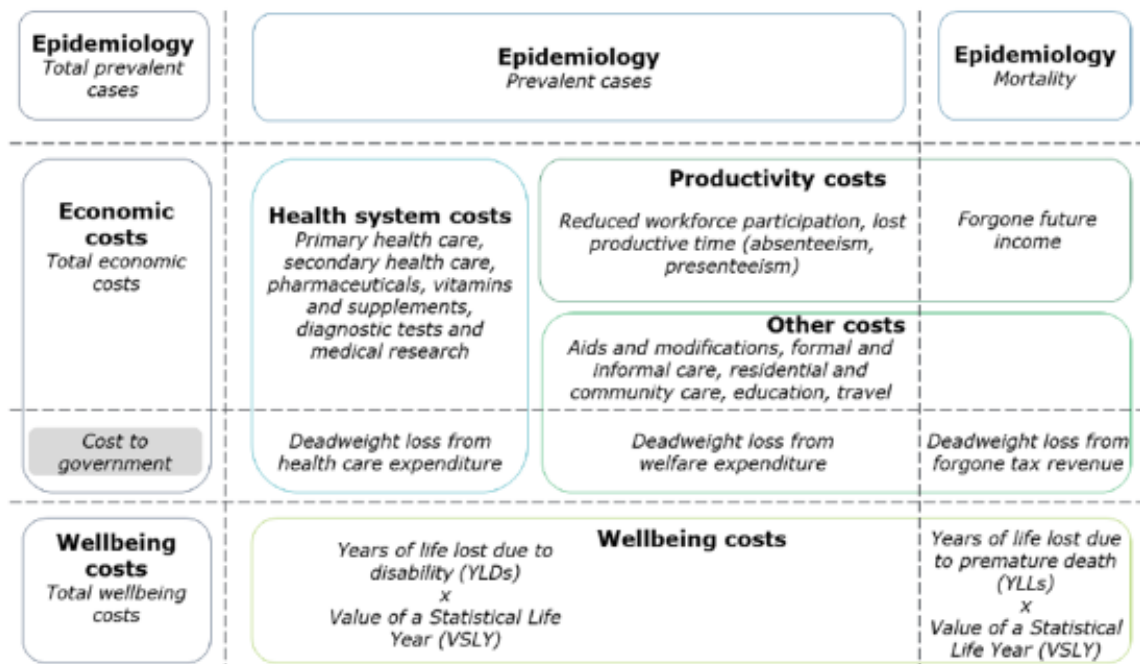
IRD	Rate (%)	Cases	Proportion (%)
RP	0.0154	10,324	49.6
Usher syndrome	0.0039	2,590	12.4
Stargardt	0.0032	2,109	10.1
LCA/EOSRD	0.0024	1,587	7.6
Best disease	0.0015	1,003	4.8
Cone dystrophy	0.0013	876	4.2
Cone-rod dystrophy	0.0012	812	3.9
Achromatopsia	0.0011	733	3.5
XLRS	0.0007	483	2.3
Choroideremia	0.0004	298	1.4
<b>Total IRDs</b>	<b>0.0311</b>	<b>20,814</b>	<b>100.0</b>

Source: Deloitte Access Economics analysis based on published literature.  
 Note: Components may not sum to totals due to rounding.

A straight projection from this prevalence breakdown to Canada’s population, which is roughly half (56%) that of the U.K.’s, suggests that our own IRD community—at least in terms of the diseases presented in Deloitte’s analysis, could total 11,737 (roughly, of course, since this is an extrapolation from population only, ignoring a range of factors). Following the same crude extrapolation, 5822 Canadians would have RP and 895 would have LCA. Genetic mutations were not part of the IRD COUNTS prevalence analysis, so determining a population size for the RPE65 group is not possible with these numbers alone.

Applied against these prevalence numbers, Deloitte used a cost-of-illness model to determine the economic impact of IRDs, including health system costs, productivity costs, and wellbeing costs, including years of life lost due to disability (YLDs), which, though not the same as the quality adjusted life year (QALY) metric deployed in many HTA models, do provide a metric for comparison. Since this is a study of economic disease burden, YLDs are more appropriate: they are measuring the amount of life lived in disability, whereas QALYs are more appropriate when measuring the amount of health or life gained through an intervention. The model is broken down in the following overview:

Figure 3.1: Cost-of-illness model overview



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By applying their cost-of-illness model to disease prevalence, Deloitte estimated that in total, IRDs carried an economic cost of £523.3 million in 2019 in the U.K. This is divided across both health system and productivity costs (£327.2 million) as well as wellbeing costs (£196.1 million).

Based on these results, we can develop a rough estimate of the economic burden in Canada by adjusting for population and currency: this translates to a total cost for the year 2019 of \$508.9 million, \$318.2 million of which are economic costs and \$190.7 million of which are wellbeing costs. The numbers may differ somewhat when we extend this analysis to Canada in a more comprehensive fashion later this year, but we could very well be looking at costs that are not far off this simple extrapolation.

## 5. CASE STUDY 2: A CONVERSATION WITH A CANADIAN PARENT WHOSE CHILD RECEIVED LUXTURNA

Last month, staff at Fighting Blindness Canada had the opportunity to “interview” a member of the IRD community whose child was treated with Luxturna. This was not a research-oriented interview, but rather a more informal conversation to learn as much about the parent’s experiences as possible, with the shared understanding that anonymized details would be used for this submission. We are able to disclose, however, that the parent and child live in Quebec—this is relevant, seeing as they were able to access Luxturna through a special case made to that government.

The parent shared that the child began showing signs of visual impairment very early on, at 2-months-old. The child was not tracking objects, smiling, or reacting to visual cues. Severe nystagmus began to develop at about five-months. After being referred to an ophthalmologist, MRI, OCT, and a range of visual testing began almost immediately, following by a genetic test and a confirmed diagnosis at 10-months-old: LCA as a result of a biallelic

RPE65 mutation. Genetic testing went very smoothly, and it was only a week or two before the results arrived. Although samples were taken at a hospital in Quebec, the parent paid for these to be shipped to an external lab.

After the diagnosis, the parent became aware of clinical trials for Luxturna but learned that the child was too young, not meeting the eligibility criteria of 3-years-old. By the time the child turned 3, Luxturna was already approved by the FDA. The parent turned to the Quebec government for assistance, making the case that access to Luxturna was necessary because no other treatment was available in Canada. The process was largely “parent-driven,” as described during the interview, but after a fast and largely streamlined process, the child was provided with full coverage to receive the treatment in the U.S., including not only coverage for the injections but for transportation, lodging, and other associated costs.

The parent described the impact of Luxturna on the child’s vision as substantial: “It’s huge. It’s still huge to this day.” Beforehand the child was very light sensitive, and could not see well or at all in dark or dim settings. The child also struggled with day vision in the form of gaps or blind spots that made it difficult to read, play with siblings, and to identify objects or people. The parent explained that “it’s kind of like Swiss cheese. There were holes in what [the child] could see, even in good lighting.” The post-treatment results were almost immediately noticeable. The parent described the child’s ability to “identify things much better,” expressing that the child “now sees the world in a completely different way.” The parent also described a positive effect on the child’s confidence. Whereas beforehand the child would respond to a friend’s greeting with a simple “hi,” the child now recognizes the friend and feels confident enough to reply with his or her name. An overall and extensive improvement to the child’s confidence and self-reliance was emphasized a number of times during the conversation.

The impact of the treatment was pronounced on the parent as well. Before treatment, the parent spent a substantial amount of time assisting with daily activities such as getting dressed and schoolwork, as well as maintaining relationships with teachers to ensure the child was receiving the appropriate amount of attention and care. Now, at school, the child is considerably more independent, and only relies, for example, on a magnifying lens for reading on rare occasions. And at home the child is much more self-sufficient, playing independently and being active in a way that is on par with siblings. The parent told us that “it’s just become so much easier.”

The parent was clear that the child’s vision is not perfect by any means. The treatment does not impact acuity in a significant manner, so the primary changes involve improvement to vision in low-light conditions, and to the “Swiss cheese” gaps that make it difficult to see during the day. Despite this, the parent told us that “there’s been such a big change from what it was, you sometimes forget about the things they still can’t do.”

The parent was also realistic about the uncertain longevity of the drug, explaining that as with other patients, improvements may very well plateau for the child or even diminish after a certain period of time. The parent still considers the treatment to be life-changing, however, because it bought a certain number of years of improved vision for the child, who can now see “the moon and the stars” for the period of time that Luxturna’s positive effects last. We were also told that Luxturna is invaluable because it provides a window of maintained vision that may allow the child to qualify for a new treatment, whether that be another dose of Luxturna or a different intervention. There was a strong sense of optimism during the conversation in relation to this idea: Luxturna

may not be a cure for blindness, but it improves and prolongs vision for those with the relevant mutation, putting them in a position to be eligible for future treatments.

## **6. CONCLUSION: TOWARDS THE EQUITABLE INTEGRATION OF INNOVATIVE TREATMENTS FOR IRDS**

From a patient-oriented perspective, our national and collective goal should be to treat Canadians living with IRDs safely and effectively, and to improve their lives with the resources that are currently available. From what patients have told us, and from the clinical data that has played a role in the drug's integration into public health systems in Europe and the U.S., it is clear that Luxturna has an important role to play in this endeavor. From a historical perspective, it is also apparent that Luxturna represents an important first step for the quickly-materializing treatment landscape for IRDs.

CADTH's review of this drug is a crucial step in the equitable integration of Luxturna into Canada's health system. At the same time, it will also signal to patients, policymakers, and industry what the future of innovative treatments for IRDs looks like in this country. It is not hyperbolic to say that the review will set a precedent, one that will impact our country's access to a pipeline of gene and stem cell therapies for ophthalmic conditions for years to come. We believe that these treatments should be available to Canadians regardless of the many social and economic factors that too often block access to treatment. To this end, we hope that this submission has provided you with important patient perspectives that can anchor the review process in the lived experiences of Canadians, as well as a concrete sense of the economic and social burdens entailed by diseases that have an enormous impact on the lives and futures of those affected by them.

As organizations that represent patients with IRDs and other eye diseases, our overarching goal is to contribute meaningfully to the discussion and potential implementation of new treatments in this space—in particular, to guide that discussion along lines that are patient-centered, that focus on optimal and equitable outcomes, and that recognize the expertise of patients with lived experience of IRDs and their value in the approval process of new treatments.

We look forward to continuing to work with CADTH to support Canadians living with IRDs, and to advance our collective understanding of how these diseases impact their lives.

## APPENDIX: VISION 2020 WHITE PAPER (LIVING WITH VISION LOSS)

### LIVING WITH VISION LOSS

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This paper was developed by Fighting Blindness Canada, the Canadian Council of the Blind, and the CNIB Foundation with feedback from members of the Canadian vision loss community. It outlines themes in the area of living with vision loss, as well as recommended actions for government, industry, and other stakeholders. The recommendations—bolded throughout the paper—provide a general framework for policy and advocacy activities in 2020 and beyond. What final form a recommendation takes, who it is directed towards, and in what context it is articulated will be determined by each stakeholder.

#### *Introduction*

A visual acuity measurement of 20/20 is often associated with “perfect” vision, a kind of universal standard for unimpaired sight. But a significant number of Canadians live with vision that is not represented by this norm. In fact, over 1.5 million live with a seeing disability,<sup>14</sup> and research has shown that, due to ageing and other factors, that number is in the process of doubling over a 30-year period that began in 2007. This could lead to national health care costs of over \$30 billion per year.<sup>15</sup>

At the same time, the experiences of the growing number of Canadians with vision outside the 20/20 ideal are far from uniform or consistent, especially in cases of visual impairment and blindness. As a result, though members of the vision loss community share much in common, their experiences of vision loss and blindness are incredibly diverse. They are shaped by factors that include age, geography, economic status, disease type, genetic history, and more, making it difficult to speak of vision loss as a single or unifying phenomenon.

The so-called “burdens” of vision loss are also varied, encompassing social marginalization, employment barriers, strain on families and caregivers, and a host of other issues. And since vision is one of the key senses, impairment can affect a number of day-to-day enjoyments, including reading, watching movies and television, cooking, looking at photographs, and more. Developed out of survey responses from members of the vision loss community that highlight these and other issues, this paper is designed to capture some of the complexities of vision loss in Canada during the symbolic year 2020. It also aims to highlight opportunities for progressive change in policy, technology, education, labour, and other areas.

#### *Thinking and Speaking About Vision Loss: Misconceptions, Biases, and Discrimination*

Living with vision loss often means being perceived as blind, regardless of the status of one’s vision. This is especially the case for those who use white canes or show physical symptoms. These and other signs are often interpreted as a complete lack of sight. This is of course a fundamental misconception of vision loss, which is varied and highly personal.

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<sup>14</sup> Morris, S., Fawcett, G., Brisebois, L. et al. *A demographic, employment and income profile of Canadians with disabilities aged 15 years and over, 2017*. Statistics Canada; 2018

<sup>15</sup> Cruess, A.F., Gordon, K.D., Bellan, L. et al. Mitchell, S., & Pezzullo, M.L. The cost of vision loss in Canada. 2. Results. *Can J Ophthalmol* 46(4), 315-318 (2011) doi: 10.1016/j.jcjo.2011.06.006

It is also the case that our language and terminology fall short of capturing the diversity of vision loss. The word “disabled” has been inadequate for some time, though it has been recontextualized in disability studies, but terms such as “blind” and “impairment” are also far from being broadly accepted. And though this paper uses “vision loss” as a kind of catch-all, this too falls short of encompassing the range of experiences associated with impaired sight. **As we continue to modify and evolve our ways of speaking about vision, it is important to take these considerations into account, and to work collectively to find our way forward in language and discourse.**

Our linguistic practices do not exist in a vacuum. They are tied to biases, misconceptions, and forms of discrimination that affect many with vision loss. Several community members have stressed that the most pronounced among these is the notion that those with impaired sight are incapable of being productive members of society, and that visual impairment is a kind of cognitive impairment. Although there is a potential link between visual impairment and age-related cognitive decline,<sup>16</sup> and although those with vision loss face added barriers to employment and social inclusion, they overcome these barriers regularly, and their ability to do so demonstrates the opposite of cognitive impairment. There is also a widespread misconception that, to quote one community member, the lives of the visually impaired are “horrible and joyless,” and that those with vision loss should be pitied, all of which is far from the truth.

Unfortunately, whether we are conscious of them or not, these and other stigmas tend to have the largest impact on children and young people, many of whom require mentorship and guidance to persevere. When they do, they often emerge as community leaders with unique perspectives and a strong sense of empathy. When they are unsupported, on the other hand, they can fall through the cracks, especially when faced with discrimination in their schools and places of employment. This can lead to depression, isolation, lack of self-worth, and more. **Community and mentorship programs are vital to overcoming these problems, as are counselling services that address the psychosocial impacts of vision loss, and health navigation services that connect individuals to resources and supports.**<sup>17</sup> For those who have acquired vision loss during working age and lost their jobs, their independence, and more, support programs are integral to managing their transition into being partially sighted and facing all of the challenges that follow.

Educating the public is important in this regard, as is finding new ways to facilitate interaction among those who are fully sighted and partially sighted. Public awareness campaigns have shown success in this context.<sup>18</sup> Many in the vision loss community would like to see an expansion of these and similar initiatives. **Whether it be on social media or in some other forum, finding new ways of thinking and speaking about vision loss, as well as ways of sharing stories and experiences, will be crucial to undercutting the discrimination that creates tangible obstacles for those with partial sight.**

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<sup>16</sup> Chen, S.P., Bhattacharya, J., and Pershing, S. Association of Vision Loss with Cognition in Older Adults. *JAMA Ophthalmology* 135(9), 963-970 (2017) doi:10.1001/jamaophthalmol.2017.2838

<sup>17</sup> Zhang, X., Bullard, K., Cotch, M. *et al.* Association between depression and functional vision loss in persons 20 years of age or older in the United States, NHANES 2005-2008. *JAMA Ophthalmology* 131(5), 573-581 (2013): doi:10.1001/jamaophthalmol.2013.2597

<sup>18</sup> Janiszewski, R, Heath-Watson, S.L., Semidey, A.Y. *et al.* The low visibility of low vision: increasing awareness through public health education. *Journal of Visual Impairment & Blindness* 100(1), 849-861 (2006) doi.org/10.1177/0145482X0610001S08

### *Working with Vision Loss: Employment and Marginalization*

When our thinking and speaking about vision loss fail to capture lived experience, stereotypes can lead to marginalization. This occurs in a variety of settings, but the job market in particular demonstrates what happens when misconceptions and indifference flourish. Too many Canadians with vision loss are under or unemployed.<sup>19</sup> This is due to a shifting and precarious job market, in part, but it also results from the added barriers and challenges faced by those with vision loss when seeking and maintaining meaningful employment.

Education plays a central role in this: the 2017 Canadian Survey on Disability showed that 24% of those living with a seeing disability had not graduated high school. In the same survey, Canadians with sight loss reported a long list of issues that directly or indirectly affected their educations, including feeling left out, being bullied or avoided, changing schools, additional expenses, and lack of assistive devices or support services.<sup>20</sup> These factors play a role in the career trajectories of partially sighted Canadians, and could contribute to high numbers in under and unemployment.

Accessibility is a key concern in the area of employment. Several workplaces have implemented successful initiatives, but the majority are either lagging or noncommittal. This must change, and the burden of showing how cannot be placed on employees to advocate on a case-by-case basis. **One way forward is to design a set of ethical standards for workplace accessibility—this can be tiered according to workplace size or category, but the details should be driven forward by the vision loss community.** Both accessible and assistive technologies should be central to any new guidelines, the former being widely usable “out of the box” for a variety of people, the latter more specialized to assist those with specific disabilities.

The Accessible Canada Act, which came into force in 2019, is a step in the right direction, but its guidelines are for government workplaces and those within the federally regulated private sector. Unfortunately, many of the country’s least accessible work environments fall outside of the Act’s purview, where the majority of Canadians work. **Continued dialogue and consultation are necessary to improve the Act and apply its regulations more broadly.**

Incentivization programs were flagged by the community as important moving forward as well, since these could help address not only accessibility concerns but other forms of marginalization. **Many members of the community would like to see federal and provincial stakeholders develop initiatives that reward employers for meeting high standards in accessibility, for hiring employees with blindness or low vision, for implementing diversity agendas, and for achieving other progressive goals.** Such programs could be integrated into a set of national and far-reaching guidelines that help establish a truly barrier-free Canada.

### *Living with Vision Loss in Canada: Accessibility and Privacy*

The issue of accessibility extends beyond employment. For instance, though transportation is a barrier to employment, especially when employers require a valid driver’s licence, it is also a barrier to accessing hospitals

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<sup>19</sup> Gold, D., Simson, H. Identifying the needs of people in Canada who are blind or visually impaired: preliminary results of a nation-wide study. *International Congress Series* 1282, 139-142 (2005) doi.org/10.1016/j.ics.2005.05.055

<sup>20</sup> Morris, S., Fawcett, G., Brisebois, L. *et al.* *A demographic, employment and income profile of Canadians with disabilities aged 15 years and over, 2017.* Statistics Canada; 2018



and medicine, grocery stores and restaurants, social and community functions, and a variety of other services and resources. **To combat isolation and ensure those with vision loss can stay connected to people and amenities, public transportation must be robust, affordable, and accessible.** This is especially the case for those in rural and remote communities who faced the largest barriers in this area. Improvements in public transportation could also alleviate strain on caregivers, many of whom drive those they care for to places of employment, health care facilities, community functions, and elsewhere. Some caregivers do this to help offset the substantial costs associated with the regular use of paid transportation services, including cabs, that they consider necessary to fill the gap left by inadequate public service.

Personal technologies—especially smart phones, which have started to replace more traditional assistive technologies—have emerged as invaluable devices for many, whether it be in facilitating navigation and transportation or engaging in online conversations and enjoying books.<sup>21</sup> But the buy-in cost for these and other devices can be extraordinary, blocking many in the vision loss community from enjoying their benefits. **New partnerships and programs that bring the advantages of accessible and assistive technologies to as many Canadians as possible should be a priority.** One example has already been provided by the World Blind Union. By working with the manufacturer, they were able to deliver a refreshable Braille reader, the Orbit, at a fraction of the cost of previously marketed products. In 2020 and beyond, we should look to this and similar collaborations to guide our own efforts. Alongside these endeavours, **it is important that we think of accessibility and inclusiveness as foundations in our collective projects, not as extraneous additions.** In software development, for instance, both Microsoft and Apple have shown the value of building accessibility into their products as central, integrated features, not something that is tacked on afterwards to fix an oversight. This is especially important in relation to vision loss, which is often accompanied by other functional limitations, including chronic pain, limitations in flexibility and dexterity, hearing loss, mental health issues, and learning disabilities.

The same philosophy can be applied to our public spaces. **Advancements have been made in some cities with tactile walking surfaces, accessible pedestrian signals, and other innovations, but these should now be standardized in building codes and within other legal, administrative, and policy frameworks. Improvements in signage, railings, doorways, parking, and other areas have been helpful for Canadians with low vision, but they need to be embraced as core aspects of urban planning, design, and law.** There is a sense that progress is being made in this regard, but also that it is happening slowly, too slowly in many cases. There is also a perception that physical disabilities are prioritized over sensory ones, leaving those with visual and other sensory impairments to “figure it out on their own.”

By and large, Canada is considered “average” by the vision loss community regarding accessibility compared to other countries. This leaves a marked space for improvement, and for the country to initiate widespread reforms that could, if prioritized, generate a model for inclusivity. Such reforms should be advanced in the spirit of the PPH model (Processus de production du handicap), widely used in Quebec, which recognized that those with disabilities find themselves in “situations de handicap,” and that it is these social situations that produce

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<sup>21</sup> Martiniello, N., Lehane, C., Eisenbarth, W. *et al.* Exploring the use of smartphones and tablets among people with visual impairments: Are mainstream devices replacing the use of traditional visual aids? *Assistive Technology* (2019)  
DOI:10.1080/10400435.2019.1682084

disability. If we fix these situations and contexts—an inaccessible walkway, for example—disability either disappears or is markedly alleviated.

Living with vision loss in this country means confronting barriers to accessibility on a regular basis, but it also means having to carefully consider the issue of privacy. Given the existence of biases, barriers to employment, marginalization, and other issues, many Canadians with low sight keep the details of their vision to themselves. Their reasons for doing so are entirely valid, but the societal drivers that lead them to feel that this is necessary must be combatted. More specifically, protections should be put in place to guarantee their privacy, especially when it comes to employment, insurance, and health care.

The Genetic Non-Discrimination Act is one example of such a protection for those with inherited diseases. It currently prohibits Canadian companies and insurers from requiring genetic tests or denying services based on genetic information. This protects those with genetic conditions from having their genetic histories held against them when seeking employment or services, or when signing contracts with companies. Knowing they are protected in this way also gives many Canadians peace of mind when seeking genetic testing; for many, this is an essential step towards accessing new treatments and being included in clinical trials. The Act was passed into law, but it is being appealed by the Quebec government on the basis of its constitutionality.

**Ensuring that the Genetic Non-Discrimination Act remains law is a key step towards securing privacy and protection for Canadians with inherited vision loss. In 2020 and beyond, similar legal safeguards should be fought for to protect the Canadian vision loss community against systematic discrimination.**

#### *Living Together with Vision Loss: Families, Caregivers, and Collaboration*

Though vision loss can lead to isolation, it is also the case that it regularly impacts the lives of those connected to it indirectly, particularly the family members, loved ones, and other caregivers who provide support to those living with vision loss. In this sense vision loss is not only a personal or subjective experience, but also a shared one that moves across a network of individuals, families, and communities.

In many cases, family members bear the largest burden of support. In the case of diseases with high treatment demands such as wet age-related macular degeneration (wet AMD) and diabetic macular edema (DME), this often means providing regular transport to and from appointments with eye specialists and blocking off time for waiting rooms—sometimes an entire day.<sup>22</sup> This can lead to financial strain and lost productivity, factors that are now calculated in many socioeconomic studies of eye diseases. In cases of severe vision loss, families and other caregivers sometimes function as a second set of eyes, a near-constant presence to ensure the safety and health of those they love.

This is especially true of the caregivers supporting those with special needs: children with vision loss, seniors, individuals with comorbidities, and others. The needs of any child are extensive, but a parent of a child with a congenital disease—Leber congenital amaurosis (LCA), for example, which leads to severe visual impairment in infancy—faces a set of particularly daunting challenges, ones that often require dramatic personal and career

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<sup>22</sup> Gonder, J.R., Walker, V.M., Barbeau, M. *et al.* Costs and quality of life in diabetic macular edema: Canadian burden of diabetic macular edema observational study (C-REALITY). *Journal of Ophthalmology* 2014(6):939315  
doi:10.1155/2014/939315

changes to overcome them. **Stress, fatigue, financial stability, and mental health are fundamental concerns here, and it is important that we find new and better ways to mitigate these and other strains placed on the individuals who provide care.**

This is important in the face of the growing need for caregivers and the growing demands placed on them. For example, as Canada's population ages, the number of citizens with age-related eye diseases will grow. Our population of caregivers will grow in parallel. And as work becomes increasingly precarious and social supports and pensions continue to disappear, there is a pronounced danger that caregivers, especially primary ones, will become overwhelmed. **Envisioning and implementing support mechanisms for our support-providers has never been more important.**

Canadian caregivers are exemplary in many ways, but one thing they show in particular is the value of collective and community-driven action. Support groups and community networks are integral to the work of caregiving, providing avenues for knowledge sharing, collaboration, and much more. Interestingly, it is exactly this kind of collaboration that will be necessary going forward—not only in the more personal instances of caregiving, but in the broader spheres of policy, law, governance, and health technology. Whether it be accessibility programs, advancements in research, innovative health policies, or new educational initiatives, collaboration and partnerships from a variety of groups will be essential. This includes government, industry, academia, health professionals, patients, patient groups, and many more. We should look to the tireless and co-operative work of Canada's caregivers for inspiration in these collective endeavours.