IRD COFFEE CONNECTIONS – HALIFAX

Speakers: Dr. Johane Robitaille; Morgan Ineson; Ushany Kanagendiran; Andrew Burke; Julie Mann; Harry Nickerson

If you have questions about your diagnosis or would like support finding resources, please contact the Fighting Blindness Canada Health Information Line at healthinfo@fightingblindness.ca or by phone at 1.888.626.2995. Our team is available to help you navigate your condition, find relevant clinical trials, and connect you with trusted information and support.

Part One: Expert Discussion

Overview and Disclaimer

Morgan Ineson: Good morning, and welcome! Thank you all for joining us for Coffee Connections. My name is Morgan Ineson, and I'm the Senior Manager of Education and Engagement at Fighting Blindness Canada.

Please note that the views shared by our speakers are their own and don't necessarily reflect those of Fighting Blindness Canada. Mention of any products, treatments, or services does not constitute endorsement, and today's discussion is for educational purposes and should not replace advice from your own healthcare provider.

Dr. Johane Robitaille is a pediatric ophthalmologist at the IWK Health Centre and a professor at Dalhousie University. Her clinical and research work focuses on inherited eye conditions in children, particularly developmental retinal diseases. She is also the founder and director of the Centre for Genomics Enhanced Medicine, where she helps advance genomics research in the Maritimes. Welcome, Dr. Robitaille.

FIGHTING BLINDNESS CANADA

What Are IRDs? / IRD Basics

Morgan Ineson: Inherited retinal diseases include many different conditions—can you give us a simple overview of what IRDs are, and what they have in common?

Dr. Johane Robitaille: Retinal disorders are a group of conditions that share the trait of affecting the retina. Think of the eye like a camera: the retina is the film. It has different layers — a light-sensing layer, and layers that reorganize the captured image and send it along the optic nerve to the brain, where it's further processed into what we experience as vision. We call these conditions inherited because they're caused by defects in different genes — there are at least 350 known so far, with more being discovered. A defect anywhere along that pathway can leave the final “product,” vision, deteriorated or absent altogether.

Morgan Ineson: Depending on where the breakdown happens, people can have very different experiences with their IRD. We have a wide range of conditions represented in the room tonight — retinitis pigmentosa, Stargardt disease, BBS, Usher syndrome, X-linked RP, and others. Dr. Robitaille, can you talk about some of the common ways IRDs affect vision over time?

Dr. Johane Robitaille: Some IRDs are non-progressive — people are born with the vision they have and it stays fairly stable, though there's often very slow deterioration even in conditions considered stationary. Others progress quickly. Vision itself has different components: central vision, which we use for reading and detail, and peripheral vision, which fills in everything around it. Some conditions affect central vision first, causing problems with color and contrast; others affect peripheral vision first, causing difficulty navigating in the dark; some affect both. Some people experience flashes of light or light sensitivity. The experience varies enormously depending on which cell types and which parts of the retina are affected. Thankfully, very rapid, severe vision loss like total blindness in early childhood is not the most common outcome — most people retain some vision.

The Importance of Genetic Testing

Morgan Ineson: Since these are genetic conditions, genetic testing can be an important part of the journey. Can you talk about what genetic testing involves, and why it matters?

Dr. Johane Robitaille: Everyone with a diagnosis of a potential inherited retinal dystrophy should be offered genetic testing — if you haven't been, you should ask. It's important because there are so many subtypes, and knowing yours lets you follow relevant research. Knowing the specific defect within a gene can also tell you what to expect for your vision, since some defects have a fairly predictable course. It can affect family planning decisions, since it tells you the likelihood of passing the condition to your children. It can also uncover a broader syndrome you didn't know you had — I've had a family where a genetic diagnosis in a child revealed that a mild version of the same defect ran through generations, tied to a syndrome none of them had recognized. And at a larger scale, knowing genetic subtypes lets researchers study how a condition progresses across many patients, which is essential for designing clinical trials and knowing what to monitor.

Dr. Johane Robitaille: The first treatment for an IRD was approved by Health Canada in 2020, and in the past ten years there's been an explosion of clinical trials — some of them very promising.

Morgan Ineson: That treatment is called Luxturna, for a gene called RPE65, which affects people with Leber congenital amaurosis, or LCA. You mentioned there are many different approaches being studied — gene therapy, cell therapy, optogenetics. Can you give us an overview of where research is headed?

Research Landscape: Gene Therapy, Cell Therapy & More

Dr. Johane Robitaille: When I started practicing 29 years ago, there were maybe a dozen clinical trials, mostly looking at supplements. Now we're seeing gene therapy specific to a genetic subtype, and gene therapy that isn't specific to one subtype. There's also optogenetics — if the light-sensing layer has been destroyed to the point that gene therapy can no longer rebuild it, optogenetics can turn other, still-intact layers of the retina into light-sensing tissue instead. Where all the layers are too damaged,

there's cell replacement therapy — introducing stem cells designed to regrow retinal tissue; a new Canadian trial is even repositioning a sheet of healthy retina in the eye.

There's also a promising oral supplement for Stargardt disease currently under FDA review, which could be approved as soon as 2027 — and it may end up being relevant to other subtypes too. Overall, we're moving toward treatments that last longer and that can intervene earlier, before more vision is lost. It's a very different landscape than it was even a decade ago.

Morgan Ineson: It's a really exciting time, even though it can be hard when people want a firm timeline — we don't want to overpromise. But there are several promising trials in the pipeline right now.

Dr. Johane Robitaille: And progress in one area tends to speed up others — the tools built to design one clinical trial can often be adapted for others, which shortens the runway for future trials too.

The Fighting Blindness Canada Patient Registry

Morgan Ineson: You did such a good job explaining why genetic testing matters. Can you talk about the patient registry, and what role it plays in someone's journey — for example, around clinical trial eligibility and access?

Dr. Johane Robitaille: Fighting Blindness Canada runs a patient registry, with a central site in Toronto connected to regional sites across the country — I run the one here for the Maritimes, and there are others in BC, Edmonton, Montreal, and elsewhere. The goal is to enter as many patients as possible, which gives us a picture of the genetic landscape of IRDs across Canada — which conditions are more prevalent where, and which centers should be considered for new trials. Once trials become available, we can identify and offer them to patients who may be eligible, which matters a lot since these conditions are individually rare. For RPE65 and Luxturna, for example, there are only about 50 people in all of Canada — without a registry, it would be very hard to know who and where they are.

Morgan Ineson: It's also useful when a pharmaceutical company running a trial elsewhere wants to know if there are eligible patients here. And participation is always optional — nothing is required of anyone in the registry.

Dr. Johane Robitaille: The registry also connects specialists across the country — there are really only about a dozen of us doing this work in Canada, so it acts as a kind of network. I've relied on colleagues through that network before, for instance when a patient was approached by a company about a trial with very little information provided, and I wanted a second opinion on how that had been handled.

Managing Eye Health Today

Morgan Ineson: While we wait for more treatments to be approved, what should people be doing now to manage their eye health?

Dr. Johane Robitaille: It's important to stay connected to the right resources. If you have a child, the Atlantic Provinces Special Education Authority (APSEA) supports kids in the classroom and even at home, and can connect families to adapted sports and activities. Vision Loss Rehabilitation Canada serves both kids and adults — they have a kitchen where they teach cooking with reduced vision, they assess and recommend adaptive apps and technology, and they run low vision clinics to help people stay as independent as possible. I also always tell my patients to sign up for the Fighting Blindness Canada newsletter — even if an update isn't specific to your exact condition, something relevant tends to come up eventually, and it's a reliable, accurate way to stay informed on research and events like this one.

Morgan Ineson: That's a role we can play too — appointment time with a specialist is limited, so we can help people prepare questions ahead of time, or help find resources afterward. We do have a Health Information Line, and we'll share that information with everyone after tonight.

Q&A: Is Genetic Testing Covered?

Audience member: When you talk about genetic testing, is the cost covered by the province?

Dr. Johane Robitaille: Yes — the Maritime Medical Genetics Service does genetic testing for all Maritime patients, and it's been free since 2018. There is a bit of a waitlist, though it can move faster for situations like family planning. You can be referred by your family doctor or your eye doctor. Since COVID, the counseling and consent process happens virtually, so you don't have to travel — you're only directed to the nearest location for the blood draw itself, which then ships to the lab in Halifax. As of this month, the genetic testing itself is being done in-house there as well.

Q&A: Revisiting Older Genetic Test Results

Audience member: My husband had genetic testing done around 2018. If new information has come out since about coexisting conditions, could his results be looked at again, or would he need new testing?

Dr. Johane Robitaille: Good question. Because these are such rare conditions, new information often doesn't reach family physicians directly — you're often your own best expert on your specific subtype. Unless you're followed regularly by a geneticist, you may not automatically find out about new developments. Sometimes patients discover connections themselves, through patient communities online, before we do. It's a bit of a gap, and it's worth staying engaged — following your specific subtype and reaching out proactively when something new comes up. I think of a condition like retinoblastoma, where a specific genetic type carries a risk of a later cancer elsewhere — families are given a letter to keep for exactly that reason, since the connection isn't always obvious otherwise.

Q&A: Later-Onset and Variable Progression

Audience member: My condition is milder and had a later onset than is typical for it. Does that pattern tend to hold — milder, slower progression — or does it really just depend on the individual?

Dr. Johane Robitaille: It's quite variable. For some genetic subtypes, the course is fairly predictable; for others, even within the same gene, the specific variant and its location can change the picture a lot. For X-linked conditions, women who carry the

gene often have milder disease, sometimes none at all. Genetic counseling is really the best way to find out where your specific mutation fits within the known range for that subtype — and for very rare subtypes with only a handful of published cases, that range may simply not be well established yet.

Q&A: Supplements and Gene Therapy for RP

Audience member: What condition is the supplement you mentioned meant to treat?

Dr. Johane Robitaille: That one is for Stargardt disease — it targets a buildup of vitamin A byproducts that damages a layer of the retina, and it works by significantly slowing disease progression. It's possible a treatment developed for one condition could eventually help a related one, since some genes sit close together functionally. There's also a gene therapy trial underway for RPGR, a common cause of RP, and the data so far looks very likely to meet the bar for approval — though it's hard to give a firm timeline. There's a separate gene therapy, not specific to one gene, aimed more broadly at protecting and stabilizing the retina across several causes of RP; that one is still a few years out, since RP itself tends to progress slowly and needs to be followed over a long period to show an effect. But it's shown real promise so far.

Q&A: Follow-Up Information on Rare Subtypes

Audience member: My husband was initially diagnosed with retinitis pigmentosa, then later told it was rod-cone dystrophy, based on genetic testing done some years ago. Is there a way to get more information or follow-up now, especially with young grandchildren in the picture?

Dr. Johane Robitaille: Genetic counselors typically don't follow up after the initial diagnosis — they tell you what's known at that time. Staying connected with Fighting Blindness Canada is the best way to hear about new information as it comes out for your specific subtype. If your grandchildren are adults and want to know their own risk or status, they can be referred to the Maritime Medical Genetics Service directly; if they're still children, that would go through their parents.

Morgan Ineson: If you have the name of the specific subtype, you're also welcome to write to our Health Information Line. We can do a deeper dive to see what's out there and help connect you to relevant resources.

Q&A: Cataracts and RP — Is There a Connection?

Audience member: I was born with cataracts, and later diagnosed with RP at 61 after years of undiagnosed night vision problems. Is there a correlation between being born with cataracts and later developing RP?

Dr. Johane Robitaille: It would be quite rare for the two to be directly connected, though it's not impossible if there's an underlying syndrome. Congenital cataracts are inherited in about a third of cases, and the cause is unknown in roughly another third, so it's possible yours were genetic as well, coincidentally alongside the RP. More commonly, we see it the other way around — people with RP tend to develop cataracts earlier than usual, often in their 40s rather than their 70s. Knowing your specific genetic subtype would let you confirm whether there's a real connection in your case.

Audience member: Is age a factor in general — does a child and an adult with the same disease progress differently?

Dr. Johane Robitaille: It's less about age and more about where the disease is at when a diagnosis is made and a treatment becomes available. Someone with a slower, later-onset form may do quite well with treatment even as an adult, while a very young child with a severe, fast-progressing form is in a more urgent position. Timing relative to the disease matters more than age itself.

Q&A: Luxturna — Customization and Access

Audience member: For Luxturna and RPE65 — are there different versions being developed, almost like a customized delivery, and separately, why has only about one person locally received this treatment so far? Is that about cost, or limited rollout?

Dr. Johane Robitaille: Yes, there are several ongoing trials working on improved versions of the delivery system — the part of gene therapy that carries the new gene into the retina. The goals are to improve delivery efficiency, reduce inflammation (everyone currently needs steroids after treatment), and expand the range of genes that can be delivered, since the current Luxturna carrier can't hold a gene as large as the one behind Stargardt disease. As for access — RPE65 is very rare, with only around 50 people in Canada. Once Health Canada approves a drug, it still has to go through a separate national pricing review before individual provinces can negotiate coverage, and that process took a few years for Luxturna, since it was the first gene therapy of its kind. One of my patients' families was very proactive and worked with Fighting Blindness Canada to help move that process along, and had the surgery done in Edmonton as soon as it became available.

Morgan Ineson: And for gene therapy generally, there needs to be enough healthy retinal tissue left for it to work — so timing in a person's disease course matters a great deal for whether treatment can help.

Closing — Expert Session

Morgan Ineson: Thank you so much to Dr. Robitaille for joining us and for answering all of your questions tonight. If you think of something afterward, you can always reach out to our Health Information Line. We're going to take a short break, and we'll be back shortly with our community panel.

Part Two: Lived Experience

Introductions

Ushany Kanagendiran: Welcome back, everyone. My name is Ushany, and I'm the Senior Coordinator of Education and Engagement at Fighting Blindness Canada. I have a wonderful panel with me tonight — Andrew Burke, Julie Mann, and Harry Nickerson — who are going to talk about navigating life with an IRD.

Andrew is a lawyer with Stewart McKelvey in Halifax and a longtime volunteer with Fighting Blindness Canada, with more than 30 years of involvement including 15 years

on the Board of Directors and six as Board Chair; he now serves on our Advisory Council.

Julie works in the Department of Pharmacology at Dalhousie University, and since her diagnosis in 2015 has focused on strengthening her own self-care; she lives with her husband and their pets.

Harry is a Grade 12 student from Halifax, diagnosed with retinitis pigmentosa at age 10; he represents Canada in goalball internationally, and created a free app called “As I See It” to help people understand and communicate the experience of vision loss.

Ushany Kanagendiran: I'd love to start with each of you introducing yourselves and sharing when you were first diagnosed. Harry, let's start with you.

Harry's Story

Harry Nickerson: I'm Harry, 17, just starting my last year of high school. I was diagnosed with retinitis pigmentosa at age 10. I used to be a competitive gymnast, and the way I was diagnosed was that when I lost my first significant chunk of vision, I started having unexplained accidents in the gym, and my coaches noticed.

Andrew's Story

Andrew Burke: My experience is probably a bit different from others here — I've never not had RP. My parents noticed I was night-blind and had poor peripheral vision when I was small. I was officially diagnosed around age 12, and got a genetic diagnosis in my 40s. My field of vision is about four degrees, but I've maintained my central reading vision, so far so good there.

Julie's Story

Julie Mann: I'm Julie. I was diagnosed about ten years ago with retinitis pigmentosa — it came as a surprise, found through a referral after a routine appointment. When I mentioned I was also hearing impaired, my doctor recognized the combination and ordered genetic testing, which confirmed Usher syndrome, type 2A, a slower-progressing form. It made sense in hindsight, since I was born with hearing loss but my

vision symptoms didn't show up until my late 20s. Looking back, I realized I'd already started avoiding night driving before I even had a diagnosis — my husband hated riding with me at night because I'd lose the lines on the road. Eventually it was recommended I stop driving altogether, and I surrendered my license, which was very hard. My journey started in Ontario; after diagnosis we moved to St. John's, and later to Halifax.

Tools, Technology & Adaptations

Ushany Kanagendiran: Thank you all so much. My next question: what are some tools, technologies, or adaptations that have made the biggest difference in your daily life? We'll go in reverse order, starting with Julie.

Julie Mann: When I was first diagnosed, I was referred to CNIB and a vision loss social worker, and at first I thought I didn't need any of it. But when he asked whether I'd been having falls, the answer was yes — several in the past year. That led to orientation and mobility training with a white cane. It took me about five years before I used it every day, but now I can't leave the house without it. I started with a blue cane just for curbs and gradually worked my way up to using it fully — it's made an enormous difference on the days my vision is worse.

Andrew Burke: Since I've always had these challenges, I learned to adapt very early, so I've never relied on much technology. I use a white cane sometimes, mostly in busy places like airports, more for identification than navigation — if I bump into someone, they apologize to me instead of the other way around. I do use some software that helps me find my cursor on my computer screen, since I can still read normal text once I locate it. Otherwise, it's mostly coping skills built up over a lifetime — sticking to familiar routes, and learning where the obstacles are.

Harry Nickerson: I'm similar to Andrew — I've adapted to using my vision as effectively as I can. I'll sometimes use a cane in an unfamiliar space, especially airports, where it also means extra assistance. I created my app, As I See It, so I could show people what my vision is actually like, since it's hard to explain otherwise — it uses your phone's camera to simulate different eye conditions through a filter. I also use a magnifier occasionally for tests at school.

Ushany Kanagendiran: We also have pocket magnifiers on our FBC table if anyone wants to take a look. Does anyone else in the room want to share a tool or technology that's made a difference for them?

Audience member: Adaptive technology can be very expensive, and unlike Ontario, Nova Scotia doesn't offer much government support for it. What's worked for me is looking specifically for free apps — there are people who build and share them at no cost, like Be My Eyes, and that's often the only realistic option given the lack of funding.

Audience member: In our house, my husband set up all our light switches and the thermostat through smart-home devices connected to voice assistants, so he doesn't have to search for a switch or read a small display — it's been a genuinely useful tool for us.

Audience member: There are apps that do all kinds of things. One I use regularly reads incoming mail to tell me who it's from, and another lets me point my phone at a room and, if I've added someone's photo, it will tell me who's nearby. There's an app for pretty much every phone at this point, most of them free.

Audience member: I use one called Lookout that helps read documents and other printed text.

Audience member: I use tinted glasses — mine happen to be red, though I know they also come in yellow, depending on the light spectrum you're trying to filter. I can't really go without them at work anymore.

Audience member: On the non-technology side — I used to be a cyclist and can't ride solo anymore, so I got a tandem bike. I also rollerblade, using a stick that a friend holds onto while skating just ahead of or beside me. Not high-tech, but it works.

Mairin (Vision Loss Rehabilitation Canada): There's a lot coming out quickly in this space, which can feel overwhelming, so it's worth remembering that just because something is new technology doesn't mean it will work for you specifically — the same way a light filter that helps one person might not help another. A proper low vision or photophobia assessment can help identify what will actually work for you. Lately, people have been excited about smart glasses like Ray-Ban Meta, and CCTVs (closed-circuit

magnification systems) remain something of a gold standard. There are also specialized bioptic lens systems with autofocus for distance viewing. And beyond dedicated low-vision tools, most smartphones already have built-in accessibility features — magnifier apps and so on — worth exploring even before turning to specialized devices.

Support Systems

Ushany Kanagendiran: How has your support system helped you navigate your eye disease, and what would you want someone newly diagnosed, or feeling overwhelmed, to know?

Andrew Burke: I'm married with four kids, so I always have people around — and they're generally more responsive when I ask for help with my eyes specifically than with everyday requests, which I suppose is something. For people who are newly diagnosed, or parents of a newly diagnosed child, the first thing I tell them is to look at the Fighting Blindness Canada website, since there's a lot of good information there. Beyond that — there are many ways to manage vision loss, but you do have to be your own advocate.

Harry Nickerson: I've always had a very supportive family and circle of friends. When I was diagnosed, no one — not doctors, family, coaches — ever told me I had to stop gymnastics; that was always going to be my own decision. I eventually stopped, but because of an unrelated injury, not my vision. People are usually more willing to help than you'd expect — I think people are often just afraid to offer because they don't want to seem presumptuous, but if you ask, most people are glad to help.

Julie Mann: Ten years ago, when I was diagnosed, I was already navigating life with hearing loss, so this felt like one more thing added to an already full plate. My husband has been incredibly supportive, coming to every appointment and being patient while I resisted using a cane at first. My parents also struggled — they felt guilty, in a way that sometimes made it more about their own grief than about supporting me, which created some tension. My advice for anyone newly diagnosed is: let people ask what you need, and respect your own pace in accepting help. I've also built my own support network over time, through Vision Loss Rehabilitation, CNIB, and mentorship through Fighting Blindness Canada.

Harry Nickerson: I'll add — my family was proactive too. Right after my diagnosis, my doctor, Dr. Robitaille, connected us with APSEA, and through them I found goalball, a sport created after World War II to help rehabilitate blinded veterans. That community has become one of the best things in my life, and it's let me travel the world with close friends I wouldn't have met otherwise.

Andrew Burke: I'd add to what Harry said about deciding for himself when to stop gymnastics — I was once asked on a panel what my own parents were thinking when I was young, and when I actually asked them, they said they simply let me try whatever I felt comfortable trying, and let me be the one to decide if something wasn't working. I played basketball, hockey, soccer, and golf growing up, some more challenging than others without peripheral vision, but I managed. That's probably a message more for parents than for someone newly diagnosed themselves.

Harry Nickerson: To add to that — a diagnosis can be a real limitation on some things, but often it's just a label, and you can keep doing what you love. That was true for me with gymnastics — my coach told me flatly there were no worries on his end, and that it was entirely my call how far I wanted to take it.

Closing

Ushany Kanagendiran: Thank you all so much for sharing tonight — I know it takes real strength and vulnerability to talk about your experience this openly, and we're so grateful.

Morgan Ineson: Thank you, Ushany, and thank you to Andrew, Julie, and Harry. And thank you all for joining us today.