IRD COFFEE CONNECTIONS – ST. JOHN'S

Speakers: Dr. James Whelan; Morgan Ineson; Ushany Kanagendiran; Ashan Kaleem; Susan Winter;

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Overview and Disclaimer

Morgan Ineson: Welcome and good morning. Thank you so much to everyone for joining us today for Coffee Connections. My name is Morgan Ineson, and I'm the Senior Manager of Education and Engagement at Fighting Blindness Canada. Fighting Blindness Canada is dedicated to supporting vision research and connecting people living with eye disease to education, trusted information, and advocacy.

Before we begin, I want to note that the views shared by our speakers today are their own and don't necessarily reflect those of Fighting Blindness Canada. Any mention of products, treatments, or services doesn't mean we're endorsing them, and sponsors do not influence the content of our program. This discussion is for educational purposes only — any decisions about your eye care should be made with your own eye doctor.

Morgan Ineson: I'd like to introduce Dr. James Whelan, Dr. Whelan is an Assistant Professor at Memorial University and Chief of the Division of Ophthalmology at Eastern Health, with a Retina Fellowship from the prestigious Wills Eye Hospital in Philadelphia. He serves as a Retina Surgeon at both Eastern Health and the Janeway Child Health Centre, focusing on the diagnosis and surgical treatment of retinal diseases across Newfoundland and Labrador.

Q&A: Symptoms of Stargardt Disease

Audience member: What are the symptoms of Stargardt disease?

Dr. James Whelan: Stargardt is an inherited retinal dystrophy that affects the pigment underneath the retina. Because the Stargardt gene, ABCA4, is so large, there are many different defects associated with the disease. Some young children lose vision very early from more aggressive genetic defects, while other forms of Stargardt don't show up until someone's 30s or 40s, presenting with decreased central vision, pigmentary changes, and sometimes changes in night vision. It's a tough one to treat, in part because the gene itself is just so big.

Q&A: Gene-Specific Approaches for Stargardt

Audience Member: What approaches are being used to work with that, given the gene won't fit inside a normal viral vector the way it does for treatments like Luxturna?

Dr. James Whelan: You'd first be tested to find your exact mutation. Because this is expensive, drug companies have narrowed in on the more common mutations that cause severe disability, and that's what they're targeting — rather than trying to treat every form of Stargardt disease, they're making very specific genetic changes so the patch can actually fit on the virus. The gene is simply too big otherwise; the patch ends up bigger than the virus meant to carry it.

Q&A: How Gene Therapy Works, and the Toll It Can Take

Audience member: Can you talk a bit more about how gene therapy works — how the virus introduces the healthy gene into the eye cells, and whether the faulty gene gets knocked out, or what the process actually looks like?

Dr. James Whelan: It's the same basic idea as any adenovirus, like when you catch the flu — your eyes water, your nose gets stuffy, your lungs are affected, because the virus has infiltrated those cells and your body reacts to it. Our hope is that if we infect a small, targeted area of the retina, we can transfer the genetic material into those cells. In gene therapy, we map out how the retina is functioning in each eye, identify the most affected

area, and inject the patch there. As those cells regenerate through the infection process, we're hoping the new genetic material takes hold across that area — currently about 750 microns. We're still in the early stages of understanding whether that effect can spread more widely.

Dr. James Whelan: It's a very cool surgery, but it's a difficult journey for patients. These treatments require a significant amount of immunosuppression — high-dose steroids, both topical and systemic — because you don't want the body fighting the treatment, and we're not yet sure whether patients need to stay on steroids indefinitely. I had a patient who went to California for a clinical trial, and two things stood out. First, his vision was actually too good — he was legally blind by visual field but had strong visual acuity, so he was functionally independent, able to work and read. Any surgery carries risk, and if something went wrong, he could have ended up worse off than before. Second, he was a young man who had just gotten married, and enrolling in the study meant agreeing not to try to have children for five years, since we don't fully understand the systemic genetic effects of these treatments yet. So while the science to build these patches is exciting, the road to actually receiving treatment can be a difficult one.

Morgan Ineson: We get questions all the time about why the criteria for clinical trials are so stringent. I think it's important to remember these aren't proven treatments yet — they're still experiments, and people volunteering are taking on real uncertainty. It's a long process. Even Luxturna took a long time to develop before it became available.

Dr. James Whelan: That's right — there's been real success with Luxturna, and some countries in Europe adopted it earlier than Canada and the U.S. But some of those countries have since stopped funding it, in part because the results didn't seem to justify the cost long-term, and the effects didn't appear to last as long as hoped in every patient. If you're treating someone in their teens or 20s, you're hoping for a treatment that lasts a long time. That said, there are hundreds of patients who've had the treatment, and there are some genuinely compelling stories of it working.

Q&A: Long-Term Risks of Genetic Therapies

Audience member: Our understanding with Luxturna is that there have been some later risks — some degeneration that may not be fully attributable to the underlying eye disease itself, and some concern that the treatment could be contributing. How concerned are you about long-term risks with these genetic therapies?

Dr. James Whelan: That data is out there, and it's still relatively early. To really understand long-term effects of a treatment, you generally need very large patient numbers to prove something statistically, and we're not quite there yet with Luxturna. But there are definitely some signals in patients experiencing changes down the road that we need to keep watching closely.

Morgan Ineson: It's worth noting that Luxturna is only available for people with very specific rare mutations — RPE65, a form of Leber Congenital Amaurosis. I believe we've only had around 50 or so patients in Canada.

Dr. James Whelan: Around 50, yes. Luxturna specifically treats bi-allelic RPE65-related Leber congenital amaurosis. I personally have about 12 patients in Newfoundland with Leber congenital amaurosis, and none of them are bi-allelic — their genetic change came from just one parent rather than both. That said, some treatment centres have gone on to treat patients without bi-allelic RPE65, which is part of what's led to gene-agnostic approaches: the reasoning being that if a treatment helps establish a healthier retina generally, it might help patients with a range of different mutations, not just the one it was originally designed for. That's where RPGR gene-agnostic treatments came from.

Q&A: Slowing Progression with Supplements

Audience member: Is Stargardt centralized to a particular area, and can you talk about the supplement approach to slowing its progression?

Dr. James Whelan: Stargardt is centred in the macula, though there are some variations. On the supplement side — this is the biochemical approach. We know which enzymes and nutrients RPE cells rely on, so making sure Stargardt patients are never deficient in those may help slow disease progression. RPE cells have a very high energy demand and turn over quickly — among the highest metabolic and turnover rates of any cell in the body. If we can keep those cells well supplied, the healthy RPE population can hopefully keep the unhealthy population in check. That's the idea behind the biochemical/supplement approach.

Q&A: Steroid Duration After Treatment

Audience member: Have you seen cases of people who had Luxturna and needed to stay on steroids indefinitely?

Dr. James Whelan: I haven't referred any patients for it myself, so I haven't seen that directly. But I was part of the Luxturna treatment group when it was first approved in Canada, and we'd meet every three months to review patients and how long they'd been on steroids. With some of the signal flags coming out of treated patients now, one of the open questions is whether steroids were stopped too soon in some cases.

Morgan Ineson: It's also still a new treatment — we have to see what happens as more data comes in. Research doesn't stop once something is approved.

Q&A: Cost and Access to Treatment

Dr. James Whelan: It's interesting — we've actually never had a patient go through the Newfoundland government for this, but each Luxturna treatment, once approved, costs about $500,000 per eye, so roughly a million dollars for a patient treated in both eyes. Given the lifetime burden of vision loss, especially for people who lose vision young, the economics can still make sense from a health-system perspective.

Morgan Ineson: I believe anyone in Newfoundland with RPE65 who wanted the treatment was able to find a pathway to it through different provincial arrangements — for example, we had a patient from Nova Scotia go to Alberta to receive treatment, and it was funded.

Dr. James Whelan: Yes, but it takes time — the drug itself was approved in 2020, and it took another 18 months or so before provincial governments worked out how to fund it, given the cost and the years of development and testing behind it. It also matters that there was significant private investment behind Luxturna's development — it started with a father whose son had Leber congenital amaurosis, who funded research at the Children's Hospital of Philadelphia until it led to Luxturna, which was later brought to market with Novartis.

Dr. James Whelan: When we start a trial, we first have to do a natural history study, so we understand how the disease normally progresses over time without treatment —

that's what lets us tell whether an intervention is actually making a difference. We did a five-year study here with patients who had an RLBP1 genetic defect. Because we share patients with a site in Sweden, their trial started there with five patients. The first three were treated safely, then two more were enrolled for longer-term follow-up, but the results weren't convincing enough to bring the treatment to market — it's a very specific defect, so worldwide there might only be about 100 patients who'd be eligible, and the numbers needed to justify bringing a drug to market are a real factor. That's part of why gene-agnostic testing may gain traction — it opens up treatment to a broader group of patients with related retinal disease.

Q&A: Measuring Whether a Treatment Is Working

Dr. James Whelan: It's all well and good to have a treatment, but you only know it's working if you have a reliable way to measure it. One challenge with the RLBP1 study was that the test we used to measure results — a dark adaptation test — takes essentially a full day to complete and isn't comfortable, since patients have to stay in the dark for a long stretch before we test their retinal response to light. The team was also trying to develop a new kind of test, more like a questionnaire, that captures a patient's ability to do things in daily life — something the older questionnaires don't really do well. That kind of outcome measurement work isn't as visible as the surgery and the gene transplantation itself, but it's critical to proving whether a treatment actually works.

Morgan Ineson: It's interesting, too — with Luxturna, one of the more innovative things they did was develop what's called a multi-luminance maze. Patients navigate a maze in a dim environment before and after treatment, and there are some striking videos of people stumbling through it beforehand and then walking through confidently afterward. That's not something regulators had traditionally considered, so it's also about finding new ways to measure real-life impact, not just visual acuity numbers, and helping regulators understand why that matters.

Dr. James Whelan: Exactly — and when you're talking about a treatment that costs roughly a million dollars per patient, having a clear way to show that it works, to Health Canada or a provincial health ministry, makes a real difference in whether it gets funded.

Q&A: Registering with FBC's Patient Database

Dr. James Whelan: I see patients regularly with inherited retinal diseases, and I encourage all of them to register their genetic information with Fighting Blindness Canada. Companies like Novartis, Roche, or Genentech are looking for large pools of patients to recruit for trials — they're not going to know there's a cluster of patients here in Newfoundland unless that information is captured somewhere. FBC can pull together numbers across the country by condition, and when a company is interested in patients with a specific mutation or disease, FBC can help connect them. So if you or someone you know has an inherited eye disease, getting tested and registering with a database like FBC's really does matter.

Morgan Ineson: We can absolutely help with that. There isn't a dedicated testing site here in Newfoundland, but there's one at the IWK in Halifax, and we can help you get your genetic information registered in that secure database. No one would ever approach you without your consent — it's just another tool in your toolkit for staying connected to what's out there and what's coming.

Q&A: Early Symptoms in Young Children

Audience member: My niece is five and keeps coming back to see the specialist — my brother has the same diagnosis. At what age do symptoms typically start showing up?

Dr. James Whelan: For patients with RLBP1 dystrophy, age five would be quite early. The main thing you might notice at that age is difficulty with night vision — they may not be able to see stars in the sky, play games like spotlight, or adjust well going into a dark movie theatre. These conditions are highly variable in how they present — even siblings with the exact same genetic defect can have very different retinal appearances.

Q&A: What Patients Can Do Now

Morgan Ineson: To wrap up — I've talked with a lot of patients living with IRDs, and it can be frustrating waiting on research. What can people do now, while we wait to see what happens?

Dr. James Whelan: You can't choose your parents, unfortunately, but there are proactive things people can do. Preschool vision testing is really important for catching a lot of ocular conditions early. If you have a positive family history, regular yearly checkups that include diagnostic retinal testing — visual fields, OCT, and similar tests — are valuable, so we can track whether there have been any changes. That matters a lot for young people making decisions about school or careers that might be harder to manage with vision loss down the road. Beyond that, we know the retina is very sensitive to smoking, so avoiding nicotine matters across all retinal diseases, not just IRDs. Most supplements on the market are formulated for macular degeneration, so they're not necessary unless that applies to you — but eating well, with plenty of antioxidants, leafy greens, and omega-3s, is good for the retina generally.

Morgan Ineson: Good for the heart, good for the eyes. And I'd add — because many of these conditions are degenerative, it's worth connecting with vision loss rehabilitation and community groups earlier rather than later. Learning to use assistive technology and resources before you strictly need them means there are more tools already in your toolbox by the time vision does change.

Part Two: Lived Experience

Introductions

Ushany Kanagendiran: Hi, everyone. My name is Ushany — I'm the Senior Coordinator of Education and Engagement at Fighting Blindness Canada, and I'm also known for being a bit quiet, so if you can't hear me, just give me a wave and I'll speak up. This session is our lived experience panel, and I have Ashan Kaleem and Susan Winter with me. Before we jump in, I'll do a quick introduction of both of them. Ashan is a Memorial University graduate student and disability rights advocate, a member of the university's accessibility steering committee, and has represented graduate students with disabilities and participated in Fighting Blindness Canada's Young Leaders Program. Susan Winter is a social worker from Newfoundland and Labrador who was diagnosed with retinitis pigmentosa in 2010, and is also the mother of two young daughters. Thank you both for being here — I'd love for you to take a minute to introduce yourselves and share when you were first diagnosed with your IRD. Ashan, let's start with you.

Ashan's Story

Ashan Kaleem: My parents have told me they first started noticing something was wrong when I was very young — I would turn toward sounds rather than lights. I was born in Pakistan and grew up there until I was 26. At around eight months old, my parents took me to an eye specialist, who diagnosed me with RP. So I was diagnosed quite young, even though I wasn't in a place with particularly advanced medical technology at the time — but I had attentive parents, which really helped. I had genetic testing done about three years ago, in 2023, and Fighting Blindness Canada played a big role in that — I wasn't aware of much of this beforehand. Through FBC's Young Leaders Summit in Toronto, I learned a lot about inherited retinal disease, gene therapies, and the value of genetic testing. All of that education and those connections came through Fighting Blindness Canada, and I really value that. So in 2023 I was formally diagnosed with a defect in the gene associated with Leber congenital amaurosis type 5.

Susan's Story

Susan Winter: Hi, I'm Susan. Similar to Ashan, there were some early questions — my sister and I both have RP, and we were tested heavily as kids, eyes dilated, stumbling around at dance class. But I was classed as okay until much later in life. I graduated university, came back to Newfoundland to start my career, and had my eyes tested again through my health insurance — that's when things were picked up, my driver's licence was taken away, and my career started off on a very stressful note. I don't have the most positive diagnosis story, honestly — the doctor I initially saw wasn't great, and I asked to be transferred to Dr. Whelan, who has been phenomenal in my journey since. He's talked me through all of the genetic testing, which I've since done, especially given that I'm a mom of two daughters — I also did genetic counselling. Genetic testing turned out to be very eye-opening, picking up more than just what relates to RP itself; they were able to look at other developmental considerations for my daughters, which thankfully haven't come up. So I'm a big advocate for genetic testing — mine was just blood work and a phone call with a genetic counsellor, and it was hugely important. I'd push it for everybody.

Tools, Technology & Adaptations

Ushany Kanagendiran: Thank you both. My next question: what are some tools, technologies, or adaptations that have made the biggest difference in your daily life?

Ashan Kaleem: The Meta smart glasses are something I've recently adopted. My vision has changed a lot over the last several years, and they help — I can ask "Hey Meta, what's in front of me?" and get a description of what's there. They also read my mail to me. I was always fairly stubborn about using accessible technology, which I'd actually advise everyone against — even if your vision is still fairly good, it's worth learning these tools early, which I didn't do, and I'm still adjusting. AI tools like ChatGPT and Gemini have also been genuinely useful day to day.

Susan Winter: I'm a bit of an anomaly at this point — I have RP, but I honestly can't point to a specific tool that's made a big difference for me yet, which I know is unusual to say, especially since it's degenerative. I'm planning to get ahead of that and learn some of this technology before I need it. I'm lucky enough that I still drive, though I have a daytime-only licence — my RP affects night vision and peripheral vision. I don't usually talk about my condition much; I've worked at my job for almost 17 years, and only a few people there know. I do a road test every year and see my specialist annually to monitor my peripheral vision. Day to day, the biggest thing has been having people who can drive me at night, and a supportive workplace that lets me leave earlier as it gets dark. I've had days I leave as early as 3:30 to make sure I'm home before dark, and there have been times I've been sent home in a taxi so I could leave my car behind. So far, I've been lucky.

Ushany Kanagendiran: I'd love to open it up to the audience as well — if there are any tools, technologies, or adaptations that have made a big difference for you.

Audience member: Thank you both for your bravery. We have a son with an IRD, and he benefits a lot from a magnifying dome — it sits right on the page and he moves it along as he reads. We've tried a lot of magnifiers, and that's been the most helpful for him so far.

Audience member: For piano lessons specifically, I use a goose-neck clamp with grips on either side — the kind sometimes used to hold a cell phone — to bring sheet music right up close to his eyes, which works much better than reading from farther away on a music stand.

Ushany Kanagendiran: We have some pocket magnifiers at the back with our FBC resources, and I'd also love to bring in our colleagues from Vision Loss Rehabilitation to talk a bit more about magnifiers.

Lori Mercer (Vision Loss Rehabilitation Canada – Newfoundland and Labrador): Hi everyone, I'm Lori, with Vision Loss Rehabilitation Canada. There's a wide range of low-tech and high-tech tools out there — everything as simple as a tactile bump added to an appliance to make a microwave or stove easier to operate, right up to magnifiers, including ones with speech output that can read text aloud and let you adjust contrast and lighting. We also have a full team offering low vision assessments, so we can help identify what type of magnification or adaptive device would actually be most useful for someone. My own role is in independent living — if you're having any difficulty with day-to-day tasks like cooking, cleaning, or housekeeping, we can talk through strategies and adaptations. We also offer counselling and mobility support for anyone having trouble navigating at home, at work, or in the community.

Lori Mercer (Vision Loss Rehabilitation Canada – Newfoundland and Labrador): If anyone would like more information, you can visit us online at visionlossrehab.ca, connect with us directly, or ask your doctor for a referral — we're happy to help with any level of vision loss that affects day-to-day life. All of our services are free, with the one exception being if someone is recommended a specific magnifier or device following a low vision assessment, which they'd cover themselves. We travel across the province, with additional specialists in central Newfoundland and out west, so we're able to visit people in their homes and communities if they're not able to come into an office.

Audience member: CNIB was great for me as well — they supported me through my cane training, which I don't need day-to-day, only after dark. I'm a nurse, working as a director now, and I'm similar to you both — my central vision is good, but everything else, and my night vision, isn't. I keep a flashlight in every room, one in my car, and one in my office. I also had special lighting put in at home, since certain lighting works better depending on the eye disease — for me, it removed shadowing. I put little stick-on lights inside my closets and pantries, small things that make a real difference. At work, I actually use a smaller screen rather than a larger one, since I can really only take in what's directly in front of me, and I adjust size, contrast, and colour settings instead. I do some speaking now on working with disability in healthcare, so I share strategies like this often. The identification cane has probably been the single biggest thing for me, even though I don't strictly need it to get around — when I'm out somewhere like the

supermarket and I'm slower because I'm scanning for things, having the cane visibly in hand makes people a lot more patient with me.

Support Systems and Advice for the Newly Diagnosed

Ushany Kanagendiran: One final question for our panel — what advice would you offer someone who's newly diagnosed, or feeling a bit overwhelmed, and what has your support system looked like?

Ashan Kaleem: That's something I'm still working on myself. I recently took a course on being upfront and confident about disability rather than over-explaining it. If people are curious about your condition — and here, people are almost always curious, which comes from a good place — it's okay to just say you're not comfortable talking about it. I used to say "I don't see very well," which people often didn't quite understand; now I just say "I'm blind," which is simpler and gets me the help I need more directly. Self-advocacy matters a lot too — don't rely on the system, or on people remembering to follow up on your behalf. As an example: my genetic testing could have taken two years here in Newfoundland through the regular system. Instead, thanks to Fighting Blindness Canada, I knew to reach out to SickKids Hospital in Toronto directly. A genetic counsellor there pointed me toward a sponsored testing program, I spoke with my family doctor, and what could have taken two years was done in three weeks — just a spit test sent away, with results back in under a month. My other piece of advice is to take away the shame — if you have a child with this condition, be proud of them, bring them everywhere, don't hide them. That's what my parents did for me, which is a big part of why I don't have stage fright today. Don't take on a victim mindset — that gives control of your life over to other people or to the system. It's easier said than done, but it starts from within.

Susan Winter: I've kept fairly quiet about my RP diagnosis outside of close family, friends, and coworkers, but I do believe in not being a victim, and talking about it — my own kids talk about it very openly now, in a normalized way, like "mom needs the flashlight because she can't see the same way you can." When I was newly diagnosed, it was overwhelming — I'd just graduated and started my career, was told the job required a driver's licence, and then told I couldn't drive, so it felt like everything was pulled out from under me. There will always be people who say something negative, or make jokes, but it takes time, and I'm getting better at talking about it. If someone jokes, I've learned to lean into it a bit — I'm known as "Cinderella" because I have to be home

before dark, so I'll joke back rather than let it sting. I also think genetic testing matters a lot as a parent — I actually put off having children out of fear, until my doctor told me not to be so hesitant about it. We've had our daughters tested, and I think having those conversations early, framing this as a difference rather than only a disability, helps take some of the negativity out of it. It's been a learning curve for me — I spent years somewhat in denial — but my advice would be to try to move past the negativity faster than I did. There's so much support out there, and so many paths still open to people.

Closing

Ushany Kanagendiran: Thank you both so much for your vulnerability and for sharing your experience today.

Morgan Ineson: Thank you both, Ashan and Susan. Thank you all so much for spending your afternoon with us.