IRD COFFEE CONNECTIONS: WINNIPEG

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Speakers: Morgan Ineson, Dr. Larissa Moniz, Dr. Joshua Manusow

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Part One: Expert Conversation

Overview and Disclaimer

Morgan Ineson: Good afternoon, everyone, and welcome. Thank you so much for being here. This is such a full room. My name is Morgan Ineson, and I'm the Senior Manager of Education and Engagement at Fighting Blindness Canada, and I'm so pleased to welcome you to Coffee Connections.

Before we begin, I want to note that the views shared by our speakers today are their own and do not necessarily reflect those of Fighting Blindness Canada. Mention of any products, treatments, or services does not constitute an endorsement, and, very importantly, sponsors do not influence the content of our education programs. Today's discussion is for educational purposes only and should not replace advice from your own healthcare provider.

So I will leave you in the very good hands of FBC's Director of Mission and Research, Dr. Larissa Moniz, and Dr. Manusow.

Introductions

Dr. Larissa Moniz: We really want to thank Dr. Manusow, who is not only here this afternoon, generously giving his time, but was also here this morning for our session on age-related macular degeneration. He's given us a lot of his Saturday, so thank you so much. Do you know many of the people in the room, or are a lot of them new to you?

Dr. Joshua Manusow: I definitely recognize some faces. I have a lot of patients with inherited retinal diseases, or dystrophies. I would preface this by saying that I'm technically the local expert on them, but I'm not necessarily an expert in ocular genetics. I'm a retina specialist, so it falls within the umbrella of things I see and treat. There are people whose whole job is ocular genetics, and they tend to be in larger centres like Toronto, Montreal, and even Edmonton. But because of where I trained, I was exposed to a lot of those people, and I spent a lot of time with them in Toronto. As a retina specialist, because of that experience, a lot of patients with IRDs get funneled into my clinic rather than my colleagues' clinics.

So that's my disclaimer at the beginning: I'm not an ocular geneticist by any means, but I have a special interest in ocular genetics, and I certainly follow lots of patients with inherited retinal problems. I'm probably more of an expert on macular degeneration than inherited retinal diseases, but I'm going to do my best.

Dr. Larissa Moniz: We'll do our best together. I've been doing a few of these across the country and have learned some exciting things in the last few weeks, so between the two of us, we're hopefully going to give the audience a lot of new information. I always say that if, at the end, we can't answer your specific question, especially if it's about a very specific gene mutation, we might not have the information at the top of our heads. You can always email us later, either personally or through our Health Information Line, and we will absolutely look up the information for you, and tell you about research and clinical trials that may be happening.

What Are Inherited Retinal Diseases?

Dr. Larissa Moniz: We always like to start at the beginning. A lot of the people in the room have been with Fighting Blindness Canada for a while, have been to other events, or are really well read, but there are probably others who are newer to a diagnosis. Could you tell us, at a

high level, what inherited retinal diseases are, what they have in common, and how they're different?

Dr. Joshua Manusow: That's a huge question. We could talk for hours on that. First off, I agree 100%: usually when you do one of these sessions with people who have inherited retinal diseases, they often know more than we do about them, because you do so much research and reading, and you're really the experts living with it.

But to pretend that someone in the audience may have no idea what they are, the first step is to understand what the retina is. The retina is like the film in the camera of your eye. It's what senses light. What you're looking at gets transmitted as an electrical signal to your brain, and then your brain perceives it. Within the retina, there are very specialized cells for sensing light. The retina is a complex part of the brain. It's like a layer cake. For those of us from Manitoba, it's like a vinarterta. That's how I explain it, because that's what it looks like when you look at it under a microscope. It has 10 different layers, with lots of different cells and neurons that connect to each other.

The important ones are called photoreceptors. You might have heard of rods and cones. People will say, "My rods and cones aren't working." The rods are very important in sensing black and white, telling you whether things are dark or light, and the cones are what sense color. Those are the two really important cells. Then there's something called the retinal pigment epithelium, another layer of cells that sits just underneath and helps clean up the photoreceptors. Those cells work really hard, and when they work hard, a lot of waste comes out of them, and the RPE cleans all that up. If any one of those three parts isn't working properly, vision doesn't work properly.

That's how I usually start explaining inherited retinal diseases: one of those cell types isn't working properly, and the reason is that there's a mutation in a gene. Genes control everything we have in our body and everything we do. Sometimes, from just bad luck, you can have a mutation in a gene that causes one of those cells to stop working. Sometimes you inherit it from a parent, or you inherit part of it from a parent, and there's a whole complex drama behind whether you have one copy of the mutated gene or two, and whether it's dominant or recessive. It's probably not that important to understand all of that, except that it's an extremely complicated system. Depending on which mutation you have, what age you get it at, and which cell type is involved, it can be a very severe thing or a very mild thing.

The other thing to remember about inherited retinal diseases, or dystrophies, is that they can affect different parts of the retina. There are macular dystrophies, and the macula is the central part of the retina that's really important for fine vision, color vision, and seeing things clearly. Some inherited retinal conditions only affect the macula, so the rest of the retina works fine and your peripheral vision is fine, but your central vision is affected. Then there's a whole bunch of diseases that are the exact opposite, where the peripheral retina gets affected first, you slowly lose your visual field, and very late it affects the central vision.

The last thing I would say is that we used to give these conditions blanket names. The most common one you would have heard of is retinitis pigmentosa, and it's okay if you still want to use that name. It's the traditional name most people know. But in reality, retinitis pigmentosa is probably 250 different diseases, with many subtypes. Some are treatable and some aren't, and they're all caused by different genetic mutations. So it's really important to know which mutation caused it. That's why we tend to call them IRDs now, for inherited retinal diseases, or we get a bit more specific and refer to the cell type involved, saying you have a rod-cone dystrophy, a rod dystrophy, or a cone dystrophy. That's important because in the future, most treatments probably won't be blanket treatments. They'll probably be gene-specific treatments. So it's important to know exactly what you have before you know whether you're a candidate for a clinical trial or a treatment.

Genetic Testing

Dr. Larissa Moniz: That's a great segue to the next question: how do you know which gene you have?

Dr. Joshua Manusow: For a long time, you didn't. I started my practice here in Winnipeg about 10 years ago, and when I first started, I would refer people to medical genetics when I diagnosed what looked like an inherited retinal dystrophy, and they would usually refuse to see them. They would say, "There's no funding for testing, and even if we tested them, what's the point? There's no treatment for these things." There might have been some reason to see a genetic counselor for family planning if someone was young, but if I had a 55-year-old who didn't plan on having kids, or didn't have kids, they would just get refused.

Then, around 2017, we had the first gene therapy come out. I believe it was the first approved gene therapy anywhere in the body, and it was for a very specific rod-cone dystrophy. That

changed everything. All of a sudden, medical genetics got very excited about getting referrals from ophthalmology, and suddenly they were seeing all of my patients, and everyone was getting tested. It would still take about two years to get in to see them, because they spend a long time with each patient and there aren't a lot of them. In some ways there was no rush, so that was okay, but it was a little frustrating. They're a bit quicker now, but it still takes some time.

Now, everyone we see that we suspect has an inherited retinal dystrophy gets genetic testing. They go and see a medical geneticist, who takes a very thorough family history, way more thorough than I would ever take in my office. They'll know more about you than you know about you. They talk about the risks and benefits of getting tested, because even though it's not a surgery or a treatment, there are all sorts of ethical considerations when testing for genes: privacy concerns, insurance concerns. They go over all of that with you and counsel you, and then ultimately you decide whether or not you want to be tested. They have a huge panel of genes, and depending on the diagnosis I give them, they test for those genes.

We have a genetic counselor who specializes in eyes here in Manitoba. Her name is Jessica Lawrence. Some of you may have worked with her already. She's not a doctor, but she works with the genetic doctors, and she gets the results and is much more aware of clinical trials than I would be. So we manage patients together. My job is to make the initial diagnosis and provide any possible treatments, because there are certain treatments that can be done, and their job is on the genetic side, linking people up with studies and things like that. In addition to the foundation, which I'm sure we'll talk about, because that's the other important part of the puzzle.

FBC's Patient Registry

Dr. Larissa Moniz: I'll do a little plug here for something we have called the patient registry. It's an inherited retinal disease patient registry, a medical directory based at different sites across the country. I think we currently have six sites: SickKids in Toronto, and sites in Montreal, Edmonton, Vancouver, and Halifax. If you don't go to one of those doctors, you can still enter the registry by contacting one of those sites.

The registry helps us understand more about the population of inherited retinal diseases in the country: how many people have different diseases, and how many have different genetic mutations. That's good for researchers doing IRD research, but it's also really good when new clinical trials come up. Companies will often say, "We have a clinical trial on RPGR, or on this

gene. Do you have anybody in Canada who could be part of it?" These are very rare diseases, so you don't know whether you'll be able to recruit enough people. Because we have the information in the registry, we can say, "Yes, there are maybe 10 people, or three people here and two people there." And once there are clinical trials recruiting in Canada or elsewhere, or once a new treatment is approved in Canada, the registry is a way for you to be contacted. So we highly recommend that you look into joining. It's a little more complicated since there isn't a site here yet, but maybe we should work on that.

Dr. Joshua Manusow: We have a lot of patients with these conditions, and we always recommend they go to your website to learn how to join the registry. The other side of the coin is that on your website, you have a really good list of the clinical trials that are scientifically sound. Nothing is ever 100% safe, but these are reasonable things to explore, as opposed to when you just Google things and find some doctor in some other country trying an experimental treatment, where you're really putting your vision in their hands, which can be dangerous. But yes, we should work on getting a site here. We've informally tried to set up our own reservoir of patients here, so maybe we can try to put the two together.

Dr. Larissa Moniz: Exactly. There are so many more clinical trials coming almost every month and every year, so the registry is becoming more and more important. If you want more information, we'll send it in the follow-up email, and feel free to email us or ask Dr. Manusow.

Major Themes in IRD Research

Dr. Larissa Moniz: Let's do one more big question, at a very high level, and then open it up to the crowd. I know there are going to be very specific questions coming. Can you take us through some of the big themes in IRD research right now? What are some of the main avenues?

Dr. Joshua Manusow: There are maybe three different things to think about. The first is gene-specific gene therapy, such as the drug Luxturna, which some of you may have heard of. Luxturna has a long medical name that I'll butcher, so I'll just call it Luxturna. It was the first approved gene therapy in the body, it was for the eye, and it was in 2017. It's for people with a biallelic RPE65 mutation. It's not a simple treatment. It cost, or at least it did cost, almost a million dollars. I'm not sure what it costs right now, probably similar. Whether provinces pay for it is extremely province specific.

Dr. Larissa Moniz: I think they all pay for it. Even if it's not official, if there is a patient in the province with that mutation, we've never heard of any province not paying for it.

Dr. Joshua Manusow: That's good. Initially, who was going to pay for it was a bit of an issue. It involves a pretty big surgery that can be somewhat risky, because it involves injecting the drug underneath the retina, which is not the easiest thing to do and has quite serious potential complications. But it was very promising, and it was a really good study. Rather than testing vision, and those of you who live with low vision know there aren't great ways of testing vision once you get to a certain point, they used a unique endpoint. They put people in a kind of maze with different light levels to see how they could navigate it. After the treatment, people could navigate the maze at much lower light levels than before, and that's how it got approved. It was a clever way of showing that the drug worked.

That's approved in Canada, and they do the treatments in Toronto and Montreal. Do they do them in Edmonton now too?

Dr. Larissa Moniz: Vancouver, and Edmonton.

Dr. Joshua Manusow: We don't do them here. We don't have a lot of patients, and we certainly have the expertise, but we weren't chosen as a site, although I was on the initial board looking at things across the country for it. There are lots of clinical trials coming for other genes. I won't go into them, and frankly, I don't know enough about them to go into them.

Then there's what we call gene-agnostic treatment, and it's a really interesting way to treat these conditions. Rather than trying to fix the mutation you have, which is what the first avenue does, this one says, "We don't care what mutation you have. We know your photoreceptors don't work anymore, so we're going to try to turn other cells in your retina, cells that are still working, into photoreceptors." They tried this about 30 years ago and it didn't work at all, but recently they've figured out how to start making it work. In fact, there are people in this room who have been in a trial for that already. It's interesting because it could treat a wide range of retinal problems, even ones that aren't inherited. Any problem with photoreceptors, really, if you can turn other cells into photoreceptors.

The third thing is device-based treatment. I don't mean low vision devices, though there's always a place for those. I'm talking about retinal prostheses, little implants placed in your eye

that function as an artificial retina. The first one was called the Argus implant, and I was involved with it in Toronto when I was a resident and fellow. It was a chip with an array of little electrodes. You would wear glasses with a video camera on them, the camera would turn whatever you were looking at into a pixelated array and send it into your eye, and the chip would stimulate the part of the retina that still worked and send that picture to your brain, which would interpret it. It wouldn't let you read the paper, but it could help you see a curb as you were walking, or see a doorway. Think of what a computer might see. It was a huge surgery, and the chip at the time was low resolution, but the technology has improved, and they're coming out with higher resolution chips. Elon Musk even has a brain-implantable chip, which is a totally different way of bypassing the eye and going directly to your brain. It all sounds futuristic, and it is, but if you compare what we have today to 20 years ago, it sounds like a science fiction movie.

I would also include things like stem cell treatments. They've researched that for a long time, and there's nothing really ready for prime time yet, but replacing some of the cells, or sheets of cells, that aren't working is another research avenue. So those are the three ways I would break it down.

Dr. Larissa Moniz: I'll add one more, which is a variation of the gene-agnostic approach, and that's neuroprotective. It doesn't matter which gene mutation you have. It's trying to help the cells that are still there survive longer. Some are gene therapies, but there's also a trial happening right now around the world, with some sites in Canada, looking at an oral drug like NAC, an antioxidant. So there are different ways, and different things, that are getting close to the clinic. I think it's time to open it up to questions from the audience. Yes, right there.

Audience Q&A

Age and timing of treatment

Audience Member: Is there an age at which it becomes hopeless, or is there an ideal age to try to catch these things? Do we know?

Dr. Joshua Manusow: It depends on which disease you're talking about. In general, a later diagnosis tends to be better, because it probably means you've had the disease for a long time without symptoms, so it's moving really slowly. Everyone's disease moves at a different pace.

We can't look at you and say, "You're going to fall off a cliff," or "You're going to go gradually." For most of the diseases, a later diagnosis is probably better, but that's not 100% of the time.

Dr. Larissa Moniz: And for some of the treatments you were talking about, is there a maximum age or vision limit?

Dr. Joshua Manusow: For Luxturna, for the RPE65 mutation, the initial patients we wanted to find were not just people with that mutation, but people who still had reasonable visual function. The idea was that if all the cells are completely dead, we can't bring them back to life, but maybe we can save the ones that are still around. Some of the early patients were children with something called Leber congenital amaurosis, which has that RPE65 mutation. I don't believe there was a specific visual cutoff.

With the Argus implant, when we first started doing it in Toronto, we were looking for people with bare light perception, really the end of the line. Part of the reason is that until you get to that point, the chip wasn't really better than what most people had, and there were a lot of potential complications. As the chips get better, and as we get better at implanting them, we would probably do them sooner. But for the last 10 years or so, retinal prostheses have been very late in the game. The neuroprotective treatments you'd want to do earlier, because you're trying to protect what you already have. The gene-agnostic treatments tend to be done later, because you're getting to a point where you don't have the photoreceptors anymore, and you're trying to turn cells, specifically bipolar cells, into photoreceptors.

Dr. Larissa Moniz: Neuroprotective approaches are about preserving vision, so you're probably looking at early to mid stage. When you go to the restorative therapies, like turning those cells into light-sensing cells, or stem cells, you're looking at later-stage vision loss, at least currently.

Dr. Joshua Manusow: There are also some very common therapies we use for a lot of these conditions. People with rod-cone dystrophies, what we called retinitis pigmentosa in the past, often get something called macular edema, which is a very common thing that people with diabetes, vein occlusions, and macular degeneration get as well. For those people, we use a pill called acetazolamide, or eye drops called carbonic anhydrase inhibitors, and they work really well. Sometimes people come in with an early to mid rod-cone dystrophy and suddenly their central vision goes, and they're very concerned that it's just gone. But really, they just have a little swelling in their retina, and we can give them some drops, and it gets better. So when

something changes, it's important to go see your eye doctor, because sometimes there are simple things.

There are also treatable forms of rod-cone dystrophy. There are some genetic mutations where there's a problem with vitamin A metabolism, or a problem with diet, that get treated with specialized diets. We send those patients to a molecular biologist, essentially, who gives them a specialized diet that can improve things. So it's not always about fancy gene therapies. Sometimes it's really basic, simple stuff, but that's why it's important to be properly tested, so you know what you have.

How many genes cause IRDs

Dr. Larissa Moniz: Other questions?

Audience Member: How many different genes cause the disease? I think you mentioned a number earlier.

Dr. Joshua Manusow: There are about 250 genes that we know of with mutations that cause inherited retinal problems, and for RP, the rod-cone dystrophies, there are about 100. Clinically, they all resemble each other, and there's a lot of overlap. There are a few mutations where I can look at a patient and say, "I bet you have a mutation in the ABCA4 gene," and I'll send a referral to the medical geneticist saying I think this person has an ABCA4 dystrophy, and nine times out of 10 I'm right just by looking at it. But there are lots of times when I can tell a patient has a rod-cone dystrophy from the testing I've done, but I have no clue which of the 100 genes it is. Sometimes you have a clue if they're really young. In that case, they do a big panel and cast a very wide net looking for mutations.

Not infrequently, someone clearly has a rod-cone dystrophy, a junior resident could diagnose it, and we send them to genetics, and genetics can't find any mutation. They'll find something called a variant of unknown significance, which means there's something different in a gene, but that gene isn't known to cause that problem. Then people get confused and ask, "Do I not have this disease?" My answer is, no, you do. It's just that the geneticists aren't smart enough yet to know what is causing it. So there are more than 100 genes. We just don't know all of them yet.

Retesting after genetic testing

Dr. Larissa Moniz: If somebody has a variant of unknown significance, or went for genetic testing but didn't get a result, are they allowed to get genetic testing again in Manitoba after a number of years? Do you know how it works?

Dr. Joshua Manusow: They are. I have patients who were tested around 10 years ago with variants of unknown significance, and we resend them. I don't think there's a specific yearly cutoff. It's up to the medical geneticist to apply to the government for funding for each test, so they would know better, but it's really on the advice of the geneticist, and they're pretty good stewards of the healthcare system. They won't test you every six months, but if it's been five or six years, and there are new techniques and new genes that have been found, they're certainly willing to do that.

We have a population here where we discovered a new retinal dystrophy. There's a large family who all thought they had macular degeneration. It really didn't look like macular degeneration to me, and eventually we showed them that they actually have a new form of macular dystrophy that had been discovered in Europe. They were from the same part of Europe, and we have a huge population with it here. When they were initially genetically tested, nothing came up, because they didn't know about that gene.

Dr. Larissa Moniz: Researchers are constantly learning and finding new genes, so that's something to keep in mind if you went for genetic testing a number of years back. If you went last year, it's probably too early, but it might be an opportunity. There was a question first at the table, yes, the gentleman in the suit jacket.

Gene-agnostic clinical trials

Audience Member: What about OCU400 and OCU410? Are they for all mutations?

Dr. Larissa Moniz: Do you know about it?

Dr. Joshua Manusow: I don't know a lot about that one. I know a little bit because I was talking to Dr. Matt Benson in Edmonton, who is involved in that clinical trial.

Dr. Larissa Moniz: It's currently in a phase three clinical trial. It's a gene therapy that is gene agnostic, so it's trying to preserve the photoreceptors that are still there. They're testing it in two populations: people who have mutations in the RHO gene, which is a very common mutation, and people with any other gene mutation. I think right now, for the clinical trial, it's RP with any gene mutation. So there's a gene-agnostic population and an RHO population. Results are looking promising from the phase one and two studies. It's now in phase three, and they're hoping to have results, I think, next year. So that's something to keep an eye on, because it is a gene-agnostic therapy.

Cost to patients

Audience Member: Cost-wise for the patient, do they pay each time they go to see one of these specialists? How does that work?

Dr. Joshua Manusow: Like to see a geneticist?

Audience Member: Yes, and down the road, if you're referred to another place.

Dr. Joshua Manusow: Generally, most of the testing, and most of the people you would be seeing, are part of the provincial healthcare plan. In Manitoba, the one thing people with IRDs get at most appointments that isn't covered is OCT. An OCT is a special type of scan, almost like an X-ray of your retina, and we're a bit of an outlier in the country in that it's not a medically insured service. Depending on which clinic you go to, you might get charged for that. But testing at medical genetics is all covered, and when you see an ophthalmologist, the exam and the pictures are all covered. It's really just that OCT test in Manitoba.

We were also chatting about low vision devices. Unfortunately, those are not covered in Manitoba, whereas in Ontario, they have some coverage. There are certain programs, and I'm not the one to talk about them. The CNIB people can talk about them. They sometimes cover it for working-age people. But there's a cost for that.

For the treatments, there's a huge cost, and no one expects someone to pay a million dollars. We were saying Luxturna has been covered. The retinal implants were also about a million dollars. When we did them in Toronto, a donor had donated $10 million for 10 of them to be

done, and we did the 10, and after that, that was kind of it. I don't believe any of the governments paid for it.

Dr. Larissa Moniz: Ontario covered it, specifically for RP, for a bit. But then the company went out of business.

Dr. Joshua Manusow: So it fell off.

Dr. Larissa Moniz: I do think that is going to be a challenge as new treatments come, because they are so expensive. But the assumption is that the provincial governments will have to pay for them, because there's no way that a person could. In Ontario, at least, when Luxturna was first approved by Health Canada, it took a really long time for the provinces to negotiate the price. It took a couple of years between the price negotiation and the provinces agreeing and setting up all their surgeries. In between, a few people got their private insurance to pay for it. We know at least two people who got treatment through private insurance, but I don't know how common that will be.

Dr. Joshua Manusow: Probably not common. The problem with these treatments is that they are very expensive, and they're expensive for maybe a good reason, depending on your point of view. These are what we call orphan drugs for orphan diseases. They're relatively rare, especially when you're talking about specific genes, and it's very expensive to do the research to bring one of these drugs to market. If you have a macular degeneration drug, which can also be expensive, you're going to use it for millions of people, so the cost of creating that drug gets spread over all of those people. But if you have a treatment that only maybe 1,000 people in the world will have, and it costs $100 million to bring to market, they have to make that money back somehow. Of course, they're also trying to make a profit, which is a whole other story about big pharma. But they are very expensive, and getting people to pay for them is very difficult. This isn't just a problem in ophthalmology. It's a problem in medicine altogether, because there are lots of really severe diseases that are rare that we want to be able to treat, but the economics behind it are very difficult.

Is gene-agnostic the future?

Audience Member: Would you say that gene-agnostic is the way of the future?

Dr. Joshua Manusow: It seems like that right now, and that's the exciting thing. But I don't know if it's necessarily the way of the future. I think it'll work for some people, and for others, you're going to need the gene-specific one. I don't think you can lump all the diseases into one.

Dr. Larissa Moniz: I like to think of the gene-agnostic therapies as really useful in the short term, before we have the money or time to develop gene-specific therapies. The gene-specific ones have just started. I imagine in 10, 15, or 20 years, we may be able to make them much more quickly and much more cheaply. Who knows what it will bring? But until that point, the gene-agnostic therapies will help many people.

Audience Member: Based on what you just said, would a gene-agnostic therapy prevent someone from having the gene therapy down the road?

Dr. Larissa Moniz: At this point, when I talk to clinicians, they don't think that would be a problem. If you have had one of these therapies, you probably aren't eligible for a clinical trial, because in a clinical trial they're trying to figure out what is working with that one therapy. But once it's a treatment, it probably won't prevent you. You do sometimes have to worry that if you've had one gene therapy and then you put another one in, there could be an immune reaction, and there is some research taking place about that. That's something you would need to talk to your doctor about. For your specific condition, they might say there's a gene therapy coming up soon, so maybe you should wait, or for others they might say it's going to be so long that it's probably better to go ahead.

Dr. Joshua Manusow: The gene-specific therapies are trying to correct the actual problem, whereas the gene-agnostic therapy is kind of like...

Audience Member: A bridge.

Dr. Joshua Manusow: Exactly, a bridge. It's not really a cure. And really, the gene-specific ones are not really a cure either. They're just trying to slow things down and hopefully preserve what you have.

Accessing specialists and supports in rural Manitoba

Audience Member: I have a different question about accessing supports. We've had genetic testing, but then nothing. We don't have an ophthalmologist, and we don't see a specialist. We're rural. How do you go about doing that? We're just told to go see your optometrist, who has very little knowledge.

Dr. Joshua Manusow: First, I'd question how you got genetic testing without seeing an ophthalmologist first. That's interesting.

Audience Member: I think it was an optometrist who sent it in, to be honest.

Dr. Joshua Manusow: Which is fine. Optometrists are certainly allowed to do that. It's been my experience that they tend to want patients to have seen a retina specialist first. There's also a test called an ERG, an electroretinogram, that they often want done to confirm that it's a rod dystrophy before they do the gene testing.

Audience Member: He had his testing about 25 years ago, in Winnipeg, and then everything was just left. We had the genetic testing just before COVID, and now it's, what do we do? That's one of the reasons we came here. We were told, "This is how you are. Live with it. There's no treatment. There's no cure."

Dr. Joshua Manusow: To a certain extent, that is true at the moment, but there's probably a kinder way to say it. You can always ask your optometrist, or your family doctor, to refer you to a retina specialist. We all take referrals from family doctors and optometrists, and we take referrals from the geneticist. If you somehow got in to see the geneticist and they did your testing and bypassed us altogether, and you want to see one of us, tell medical genetics. I'm sure they'd be happy to send something the other way, because they're so used to us bothering them. There are six retina specialists in the city now, and we all see people with these conditions.

When you come, bring the report with you. Most people had their genetic testing through the same place, Health Sciences Centre, so we can often just contact genetics. If it's someone who had testing a long time ago and hasn't seen genetics in a long time, I'll often send them back and say, "Maybe you should go talk to them, because things may have changed." They may be

aware of new trials, or they may want to retest you, because maybe you had a variant of unknown significance, or they couldn't figure it out.

Audience Member: We have a new word. It's not RP. It's [unclear]-dermia.

How often to be seen

Dr. Larissa Moniz: How often would you see your patients who have an inherited retinal disease if there's no other problem?

Dr. Joshua Manusow: If there's no other problem, I usually say once a year. It might be overkill, and probably every two years is okay, but I find that if I say two years, I don't see you for four years. So I like to at least try to make an appointment in a year, and that tends to stretch out a little. If you have something active going on, like macular edema, or we're trying to treat something, we'll see you more often. People with these diseases also get other eye problems. You can get glaucoma, you can get macular degeneration, you can get all the same things anyone can, and then we'll see you more often because you might need to be followed. But once a year is generally my standard.

Usher syndrome

Dr. Larissa Moniz: The gentleman in the green sweatshirt at the back first.

Audience Member: I have Dr. Duker here in Winnipeg, who's excellent. He says two years. He doesn't seem to think one year. My specific question is that I was genetically tested, and it's Usher syndrome, with my specific genes. I'm wondering whether there have been any developments, because it's been since 2018 or 2019.

Dr. Joshua Manusow: I'm not aware of a specific one. Are you aware of any specific ones for Usher?

Dr. Larissa Moniz: I am, but it's specifically for Usher 2A. For Usher 2A, there is a gene-editing therapy in a phase three clinical trial right now by a company called Sepul Bio, which was originally called ProQR. There are Canadian sites in Montreal and either Edmonton or Toronto. I'm not sure what the second site is.

Audience Member: I'm registered with Children's Hospital. Should I contact them if there's something going on?

Dr. Larissa Moniz: Is that Children's Hospital in Winnipeg or in Toronto?

Audience Member: Toronto. That's why I registered.

Dr. Joshua Manusow: Then you're probably part of the registry.

Dr. Larissa Moniz: You're part of the registry. They would contact you if there were something, because a lot of these trials only need two or three people, so it doesn't mean they're contacting everyone. But the good thing is that if it becomes an approved treatment, they will contact you. And it's specifically for Usher 2A. There are lots of types of Usher, so I don't know if that's yours.

Audience Member: It's just vision. It's not hearing.

Dr. Larissa Moniz: Okay. The gene therapy I'm talking about is just for the vision, yes.

Audience Member: Thank you.

Moving from pediatric to adult care

Dr. Larissa Moniz: The lady in the black sweater, yes.

Audience Member: If you're seeing a pediatric ophthalmologist, is there a point where you move to an adult one? How does that work?

Dr. Joshua Manusow: The pediatric ophthalmologists in Winnipeg are really good, and they have a lot of experience with IRDs, so they do most of the diagnoses. They'll also send patients to me if there's a question they're not entirely sure about, for a second opinion. But patients generally get followed by the pediatric team until they're 18, and once they turn 18, they refer them back to me, and then we follow them.

Audience Member: And that would be on an annual basis?

Dr. Joshua Manusow: Every one to two years, depending on who you see. My partner sees everyone every two years. I agree with that, and I think every two years is good. It's just that I find when I make an appointment for someone in two years, I don't see them for four years, so I tend to err on the safe side.

The pediatric ophthalmologists are the people who trained me. I was their resident and fellow, so I have a relationship with them, and they tend to send people like your son back to me when they're done. We've even done some treatments here for older kids, where we got hold of a gene therapy drug to inject, so they didn't have to travel back and forth. It's not something we do regularly, but we certainly work with them.

Testing in young children

Audience Member: If a child is tested at a quite young age and everything comes back normal, is it possible for something to show up later in life?

Dr. Joshua Manusow: The gene mutation, or the actual problem with the eye?

Audience Member: She was quite young, and everything came back normal. Is that something that could change?

Dr. Joshua Manusow: It shouldn't, from the genetic side, unless she had a gene mutation that they didn't test for, one we're not aware of. I don't want to say she could never develop some form of it, but if they know the gene you have and she doesn't have that one, then it would be pretty unlikely.

Access to new treatments

Audience Member: I know that OCU400 or OCU410 will be available in the second quarter of 2027. If I'm seeing you as my doctor, when the treatment is available, are you the one who puts us on the list for treatment, or how does it go?

Dr. Joshua Manusow: It depends on whether we have access to the treatment in Manitoba. If it's something we can do here, then of course. If it's only being done in Edmonton or only in Toronto, then you would have to be referred to one of those centres to have it done.

Dr. Larissa Moniz: We have time for probably one or two more questions before we head to a break. Dr. Manusow, are you able to stay around for another half hour? He'll be here for the break, so if you have questions you want to ask him personally, you can do that too. Over here.

Testing by optometrists and ophthalmologists

Audience Member: I'm on my fifth ophthalmologist, and I have a question. I see an optometrist and I see an ophthalmologist. I have testing here and testing there, and I ask some questions, but the patient doesn't always know what is what.

Dr. Joshua Manusow: Your optometrist might be doing different tests than your ophthalmologist. For instance, I mentioned the OCT test. Some people in Manitoba always bring that up, because they pay for it with their optometrist and they pay for it with their ophthalmologist, and it's frustrating to feel you're paying twice. You might be getting it done for glaucoma with your optometrist, but when you see me, I'm doing it on your retina, in a totally different part. It's almost the equivalent of having an X-ray of your hip and then an X-ray of your arm. It's the same test, but of a different part, used for a different reason. That said, there certainly is duplication, and that is an issue.

The other issue is that the retina is a very dynamic thing. If you have testing done with your optometrist in June and then see me in September, what it looked like in June is not necessarily what it will look like in September. So I'm always going to want to do a new scan, and when I see you again in December, I'll want to do another one, because it won't look the same. When you talk about costs, every clinic does it differently. We charge once a year for these tests, and once you've paid, you don't pay again until the next year, even if we have to do it 12 more times. Other clinics charge every single time, maybe less than we charge for the year. There's no standardized model, but we recognize that it's frustrating. It's also very difficult to share the testing. Sometimes the optometrist will do one of these scans and fax me a grainy black and white picture, and I can't do anything with that, and I wouldn't base any recommendations on it.

Audience Member: At the Misericordia, I never paid for anything all those years.

Dr. Joshua Manusow: At the Misericordia, that's a different story, because in hospital everything is covered, but outside of hospital nothing is. To make it even more complicated, now some of it isn't covered at the Misericordia, depending on whether you're seeing a private

ophthalmologist or a public one. For instance, if you see me and get an OCT done at the red desk at the Misericordia, you don't pay for it, because that's my public clinic. But if you see my colleague who works at the green desk, who's also a retina specialist, you would pay for the OCT at the green desk. He also works at the red desk, and when he's at the red desk, you wouldn't pay. It's very confusing for all of us, because the ophthalmologists they've brought in are renting space as if it's a private office and not part of the hospital. It would be nice if the province would just pay for it. It would solve a lot of problems, but yes, it's a frustrating thing.

Dr. Larissa Moniz: I think we're going to take a break here. On the number of tests, I don't know a lot about all the different types, but we have a really good article on our website where we talked to a research associate who works at a big eye clinic in Toronto called the Kensington Eye Clinic. He wrote about the different types of tests you might have if you're going to a retina specialist, the OCT, the ERG, what they all mean, and why somebody would do them. That will come to you in the follow-up email, with all the links and information you need.

Closing

Dr. Larissa Moniz: I want to thank Dr. Manusow for joining us once again.

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