

Speaker 1:

Welcome to the Eye on the Cure Podcast. The podcast about winning the fight against retinal disease from the Foundation Fighting Blindness.

Ben Shaberman:

Welcome everyone. This is Ben Shaberman, Senior Director of Scientific Outreach at the Foundation Fighting Blindness. I'm so glad you could join us for another episode of the Eye on the Cure Podcast. For this episode, you have just me, I don't have any guests, but I will be covering a pretty important and pretty popular topic, and that's the Foundation's No-Cost Genetic Testing program. Now, a lot of the information that I'll be covering today is also on our website, [fightingblindness.org](http://fightingblindness.org). There's a genetic testing section on our homepage, so if you'd rather watch a CAD video than listen to me for the next 10 or so minutes, you can learn more about the program, again, at [fightingblindness.org](http://fightingblindness.org), but if you have the courage to stay on and listen to the podcast, let's get started. So first of all, why is genetic testing often important for people with inherited retinal diseases?

Well, it really helps confirm a diagnosis. A lot of retinal diseases can be difficult to diagnose clinically, that is a doctor may not be quite certain what a patient may have. Sometimes the diseases look alike. Sometimes they present atypically, but if you can find the mutated gene causing the disease then you really have a clear diagnosis, and there are nearly 300 genes, each of which when mutated can cause an inherited retinal disease, so clarifying the diagnosis is an important reason why genetic testing is important, but it can also help the patient and the family if the gene is found, who may be at risk for the disease, what the inheritance pattern might be. And it's also a great way for patients and families to understand which trials and emerging therapies may be applicable. Though keep in mind, some therapies that are in clinical trials and coming down the pipeline won't require knowledge of the mutated gene, but if you do have that knowledge, it's often helpful.

And finally, we find that a lot of patients and families when they do have their mutated gene identified, find some affirmation or empowerment. It can be helpful emotionally just to get at the root cause of the condition that's been causing vision loss. So with the technologies that we have available today, and we use a panel from a company called Blueprint Genetics, the mutated gene is identified in about 60 to 65% of cases. So how do you go about getting a genetic test through the Foundation's program? Well, first of all, you need to be clinically diagnosed by a doctor, and just about any doctor in the US can make that diagnosis and then order the test. It could be an optometrist, a retinal specialist, an ophthalmologist, as long as they can make that clinical diagnosis, most doctors can order the test, and it's ordered directly from Blueprint.

It's ordered online, so the doctor needs to go to the Blueprint website, [blueprintgenetics.com](http://blueprintgenetics.com), go to the nucleus portal to register, and that's where he or she can order the test and order kits that can be sent to his or her office or potentially you the patient. Now keep in mind, because these conditions, these inherited retinal diseases are rare, your doctor may not be aware that even genetic testing is available for IRDs. They may know about genetic testing and may not know about the blueprint program, so the message there is you may need to be an advocate and help educate the doctor and the staff about this program so you can get them to order the test. Now, the test is pretty simple, you normally either spit in a tube or swab the inside of your cheek, then that DNA is put into a container and then it's sent off to the blueprint lab and the results come back in about four weeks.

Now, an important component of the genetic testing process is genetic counseling, and we offer no cost genetic counseling through the company InformedDNA. I'll be talking about them in a moment. If your doctor has a genetic counselor in their clinic, they can use their genetic counselor, but genetic counseling is required in this program. The doctor either needs to provide it or order it when they order

the test online. And genetic counseling is important to help you and your family understand what the results mean from the test. The reports that come back to the doctor can be pretty cryptic in many cases, and as I indicated before, while about 60 to 65% of cases yield a definitive result, there are many times where you don't get a clear result, and the genetic counselor can help you understand the path forward, if additional testing might be warranted for you or family members, and then also what clinical trials and emerging therapies might be applicable to you.

Now, we really like the counselors at InformedDNA because they really understand the inherited retinal disease space. They're very knowledgeable and very experienced in our world, and they provide their counseling over the phone, so you don't have to go anywhere. You can do it in the comfort of your own home. They're pretty flexible with hours, so it could be evenings or weekends. And what's nice about phone counseling is you can have, perhaps, another family member or friend join the call if you want to help you ask questions and take notes. So the other thing I wanted to mention is our patient registry, the My Retina Tracker Registry, and this is an important component as you go through your journey in trying to overcome and manage your inherited retinal disease. And whether you've received a genetic test, whether you know your gene or not, it's really important to be in the My Retina Tracker Registry.

Now, this is basically a database. It's very secure. The information is private, but you go to [myretinatracker.org](http://myretinatracker.org) to create the record. You or, perhaps, a family member create the record, and by creating a record and putting information about your condition in that database, you can get on the radar screen of researchers and therapy developers who are conducting studies and potentially recruiting for clinical trials. Now, keep in mind, we never share personal information with these therapy developers or these researchers. When they do a query, the personal information doesn't go back to them. When they do a query, the names and contact information are sent to the foundation to a couple of our administrators, and then we contact you, the patient or the family member to let you know that you matched the search criteria, and then it's up to you to actually contact the researcher or investigator.

Now, again, My Retina Tracker is a great way to get notified about clinical trials, but we also strongly encouraged people to visit the Clinical Trials website at [clinicaltrials.gov](http://clinicaltrials.gov). It's just about all the trials in the US and many overseas are listed on this website, and you can also go to [fightingblindness.org](http://fightingblindness.org) our website, to get the latest news on many trials. And as I said earlier, a lot of the information I covered today is on our website under the Genetic Testing tab which is at the top and center of our homepage. Now, if you have specific questions about genetic testing, you can send an email to [coordinator@myretinatracker.org](mailto:coordinator@myretinatracker.org), and our staff will try to help you.

If you have questions about the podcast, you want to reach out to me, you can do that by sending an email to [podcast@fightingblindness.org](mailto:podcast@fightingblindness.org). So thanks for taking a moment to learn about our No-Cost Genetic Testing program. It's again, available to people with inherited retinal diseases throughout the US. And good luck in managing your inherited retinal condition. And as always, thank you for joining us for another episode of Eye On the Cure. Stay tuned and hope you can join us for our next episode.

Speaker 1:

This has been Eye on the Cure. To help us win the fight, please donate [@foundationfightingblindness.org](mailto:@foundationfightingblindness.org).