Frequently Asked Questions About the My Retina Tracker Genetic Testing Program
May 2024

General Program Information

What is the My Retina Tracker Genetic Testing Program?

The Foundation-sponsored My Retina Tracker Genetic Testing Program was launched in 2017 as an extension of the My Retina Tracker Registry (MRTR). It is a collaboration between Foundation Fighting Blindness, PreventionGenetics, and InformedDNA. To date, thousands of genetic tests have been ordered across 800+ participating eye healthcare providers (eye HCPs).

What are the aims of the My Retina Tracker Genetic Testing Program?

This study is designed to increase understanding of the rare Inherited Retinal Degenerations (IRDs) with a particular focus on the underlying genetic causes and how they relate to the signs and symptoms of these conditions.

In conjunction with MRTR, it also benefits the research, medical, and IRD patient communities by:

- Creating a research-ready cohort of individuals who have consented to receive targeted recruitment notifications sent by MRTR on behalf of research/clinical partners.
- Enhancing an already robust dataset which can be shared with research/clinical partners in a de-identified way to support study planning, site selection, potential participant identification, and more.
- Addressing patient barriers to IRD genetic testing and genetic counseling access in the United States.
- Allowing eye HCPs to order the highest quality genetic testing which has historically not been covered by health insurance but may produce information that can be important to affected and unaffected family members.
**Genetic Testing**

**What is genetic testing?**

Genetic testing can be used to determine the genetic cause of a person’s IRD. Using a sample of saliva or blood, clinical laboratories examine known locations in the DNA sequence (genes associated with IRDs) to determine if any changes in the DNA sequence (also known as variants or mutations) are causing the signs and symptoms in the affected person.

In the My Retina Tracker Genetic Testing Program, eligible participants will undergo genetic testing through our collaborating clinical laboratory, PreventionGenetics using the My Retina Tracker Panel. One hundred and ten genes (110) known to cause IRDs are tested for variants through the panel. If a disease-causing variant is found in a proband (first person in a family to be tested), other eligible family members can be tested for that specific variant.

**What is the role of genetic testing in IRDs?**

The precise genetic cause of a person’s IRD is important for medical care, family planning, and research that can lead to better treatments.

- **Medical management.** Genetic testing is one component of follow-up care for people with IRDs. In some cases, genetic testing may inform medical management (i.e., in IRDs that have an FDA-approved treatment). The genetic test result may also give your provider more information about your prognosis, including the symptoms you may develop or the rate of progression.

- **Cascade testing.** If the disease-causing variants in a family are identified, targeted testing for these variants is available to clarify the risk for other family members and establish carrier status for IRDs with certain inheritance patterns.

- **Family planning.** Once the disease-causing variants are identified in the family, informed decisions can be made about family planning.

- **Research.** Genetic data is essential to further IRD research as it helps advance the understanding of disease progression, provide insight into how different variants cause different disease outcomes, and allow for the development of treatments that target an underlying genetic cause. It can also help investigators identify potential participants for their research studies.
What type of genetic testing does the Program offer?

There are two options for IRD genetic testing at no cost to the patient:

- **110-gene panel.** If you are eligible to participate in the Program and do not have a blood relative who received a positive test result through this study, your eye HCP will order a genetic testing panel. This consists of 110 genes that can cause IRDs.

- **Targeted familial variant testing.** If you are eligible to participate in the Program and have a blood relative who received a positive test result through the Program (at PreventionGenetics), your eye HCP will order a targeted familial variant test. In this test, the clinical laboratory will determine whether you inherited the variant(s) that were identified in your relative. No additional genes or variants are evaluated during this test.

Who can participate?

You may qualify to participate in the Program if you meet the following criteria:

- Have a clinical diagnosis of an IRD that falls under the mission of the Foundation.
- Have not undergone genetic testing with a panel consisting of 32 or more IRD-related genes within the last 5 years, a whole genome test, or a whole exome test.
- Have not received an IRD-related molecular diagnosis from any previous genetic testing.
- Live in the United States or a US territory.

Of note, if you do not have a blood relative who received a positive result from the Program or a first degree relative tested through the program, you may qualify to undergo genetic testing with the 110-gene panel.

If you have a blood relative who received a positive result through the Program (at PreventionGenetics), you may qualify to undergo targeted familial variant testing.

Are there any costs for genetic testing and genetic counseling?

The cost of the genetic test will be paid by the Foundation Fighting Blindness. The total cost of genetic counseling and the diagnostic test will also be paid by the Foundation Fighting Blindness when a referral for a results consultation with the Program’s genetic counseling partner is made during the test requisition process.

The cost of your routine eye HCP visit will not be paid by the Foundation Fighting Blindness.
6. What can I learn from genetic testing?

The results of the 110-gene panel could be:

- **Positive**, and may
  a. Contribute to your IRD diagnosis.
  b. Reveal carrier status for a genetic condition that your child could inherit.
  c. Have implications for other family members.

- **Negative**, and may:
  a. Be uninformative.
  b. Suggest that additional testing might be worthwhile.

- **Of uncertain significance**, and may:
  a. Lead to a suggestion that additional testing, or genetic testing of additional family members, may be helpful.
  b. Remain uncertain for the near future.
  c. Be resolved over time if additional cases like yours are documented by genetics experts.

The results of the targeted familial variant test could be:

- **Positive**, and may
  a. Contribute to your IRD diagnosis or indicate that you will likely develop symptoms of an IRD in the future.
  b. Reveal carrier status for a genetic condition that your child could inherit.
  c. Have implications for other family members.

- **Negative**, and may:
  a. Indicate that you did not inherit the variant(s) identified in your relative
  b. Suggest that additional testing might be worthwhile if you have symptoms of an IRD or other genetic condition.

*Incidental findings.* In some cases, genetic testing in family members may reveal unexpected findings. For example, the test could reveal non-paternity (someone who is not the biological father), or some previously unknown information about family relationships, such as adoption.

7. How will I receive my results?

When the test results are ready, a report will be sent to your eye HCP. If your eye HCP did not refer you for genetic counseling through this study, he or she will explain the results. It is your right to ask your provider for a copy of your test results if they do not provide it.

If your eye HCP has referred you for no-cost genetic counseling through this study, you will be contacted by our collaborating genetic counseling provider to schedule an appointment to receive your results and discuss their implications for your medical care
and family. You will receive a summary of your genetic counseling session and a copy of your test results following the session.

8. What if I receive a negative genetic testing result?

If you receive a negative test result, there is still a possibility you may have an inherited retinal disease caused by a gene that is not on the My Retina Tracker 110-gene panel. If you are interested in pursuing additional testing through insurance or self-pay, PreventionGenetics and several other genetic testing labs offer larger IRD panels. Since every patient is different, please direct questions regarding additional testing to your eye HCP who will be more familiar with your diagnosis and family history.

Genetic Counseling

9. Is genetic counseling required?

The Foundation Fighting Blindness recognizes the importance of genetic counseling as a key component of the genetic testing process. As such, the Foundation requires that all participants who undergo testing through the My Retina Tracker Genetic Testing Program with the 110-gene panel receive genetic counseling when their results are ready. This will ensure each person tested fully understands the medical and familial implications of their genetic diagnosis.

When your eye HCP orders the My Retina Tracker Panel for you, they have the following options for genetic counseling:

- The Foundation offers genetic counseling at no cost to the participant through InformedDNA. If your eye HCP selects this route, the laboratory will send a referral to InformedDNA when your results are available. Upon receipt of the referral, InformedDNA will contact you to schedule a telephone-based counseling appointment.
- If your eye HCP prefers, they or a member of their can provide genetic counseling to you.

You can discuss these genetic counseling options with your eye HCP at your appointment.

Of note, no-cost genetic counseling is not available for My Retina Tracker Genetic Testing Program participants undergoing targeted familial variant testing.
10. Will I get a copy of a genetic counseling report?

If you are referred to InformedDNA for counseling, you will receive a copy of the post-counseling report and the test results following your appointment.

When the test results are ready, a report will be sent to your doctor. If your healthcare provider did not refer you for genetic counseling through this study, he or she will explain the results. It is your right to ask your provider for a copy of your test results if they do not provide it.

Program Participation

How do I participate?

Patients who are eligible and interested in participating must first meet with their eye HCP and discuss genetic testing and genetic counseling. Then follow these steps:

- **Review the program brochure and patient FAQ.** If your eye HCP believes that you are eligible for the program, he or she will complete a webform with some general information, including your email address. Upon submission of the form, you will receive an email from the Foundation Fighting Blindness via DocuSign that includes links to a Program flyer and patient FAQ and a button for accessing the Program paperwork. Please review the Program brochure and patient FAQ to determine your interest in Program participation.

- **Click on the DocuSign button to access the Program paperwork.** If you are interested in enrolling in the Program, you can access the MRTR and MRT-GTP informed consent documents, a HIPAA release form, and InformedDNA’s Consent for Services by clicking on the DocuSign button in the email. Please direct any questions you may have regarding the Program and participation to the Foundation Fighting Blindness at coordinator@myretinatracker.org. After submitting these forms, your eye HCP will receive an email, so he or she can order the genetic test.

- **Submit a sample for genetic testing.** Based on your provider’s preference, a blood or saliva sample will be collected for testing. Your eye HCP may collect a blood or saliva sample in the office or request that a saliva collection kit be sent to your home address. In the latter case, you will collect the sample at home and return it to the lab in a self-addressed, paid envelope.

- **Meet with your eye HCP or a genetic counselor.** When your results are ready, you will meet with your eye HCP and/or a genetic counselor to receive your
results and learn more about the implications they may have for you and your family.

What is the My Retina Tracker Registry (MRTR)?

MRTR is a patient-reported research registry that gives patients and families the opportunity to advance IRD research by sharing their health data with qualified investigators, promoting awareness of their specific disease, and increasing their visibility for research studies and clinical trial opportunities. Since launch, the MRTR has grown to include more than 30,000 adults and families currently participating. This large group of genetically tested individuals has become increasingly valuable as a source for deidentified research data that can be used to support clinical trial planning and development, estimate disease prevalence, increase understanding of the patient journey and more. Of note, the Foundation is deeply committed to patient privacy, so no identifying information is ever shared with external investigators.

Do I have to join the Registry?

Yes, one of the requirements for Program participation is for patients to enroll in the Foundation’s MRTR. In joining the Registry and sharing your genetic testing results with the Foundation, you will contribute to a database that helps researchers understand IRDs. You will also benefit from receiving communications regarding relevant research studies or clinical trials that you may qualify for based on your Registry profile. The Foundation does not use the Registry for fundraising.

How will my data and sample be used?

- Genetic testing services. Our clinical laboratory partner will prepare and deliver a genetic testing report to your healthcare provider.

- Research. Your genetic testing results will be stored in your MRTR profile and used as outlined in the MRTR informed consent. However, as with all information in your MRTR Health Profile, you may request to have some or all the information in your record deleted at any time. Of note, the Foundation is deeply committed to patient privacy, so no identifying information (like your name and date of birth) is ever shared with external investigators.

The clinical genetic testing lab may use and disclose de-identified test data and results with external physicians, scientists, researchers, and pharmaceutical companies to promote research and improve the diagnosis and treatment of
IRDs. No identifiable protected health information (PHI) will be shared without a signed HIPAA release form.

- **Quality Improvement Activities that Support Genetic Testing Services.** The clinical genetic testing lab will store a portion of each blood sample collected and use it in a de-identified way for additional genetic testing or to help develop or improve genetic testing methods. If you are from the state of New York and have not provided consent for the lab to retain your sample, it will be disposed of 30 days after testing is complete.

**Will my data be kept confidential?**

All identifiable information that is obtained in connection with this Study will remain confidential. Test results and data stored in MRTR will be protected as outlined in the MRTR consent form.

The clinical laboratory partner uses technical, administrative, and physical safeguards to secure data and protect it against misuse, loss, or alteration. It also takes steps to de-identify or anonymize data in accordance with applicable laws.

The electronic signature platform will use reasonable and appropriate safeguards, including encryption in transit and at rest, and comply with the requirements of the HIPAA Security Rule with respect to electronic protected health information (ePHI), to prevent the inappropriate use or disclosure of PHI.