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General Program Information

1. 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 healthcare providers.

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

The primary aim of this Program is to describe the genotype-phenotype associations of inherited retinal diseases (IRDs) in My Retina Tracker Registry (MRTR) participants to benefit patients and the IRD research community. 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 to support study feasibility assessments, site selection, cohort identification, and more.
• Addressing patient barriers to IRD genetic testing and genetic counseling access in the United States.
• Allowing health care providers to order the highest quality genetic testing which has historically not been covered by health insurance but may have medical and familial implications.

New Program Overview

3. What are the major Program changes?

The updated Program is designed to accommodate your busy clinic schedule by simplifying the ordering process. Other major Program changes are outlined below:

• We will transition the laboratory partner from Blueprint Genetics to PreventionGenetics.
• Providers are required to apply with the Foundation for participation in the Program. The Program application can be submitted electronically here: https://forms.office.com/r/7YfDcucELU
• The informed consent documents for the My Retina Tracker Registry and Genetic Testing Programs and the HIPAA release form will be collected via an electronic signature platform.
• We are offering a more targeted 110-gene panel which consists of the genes that account for over 97% of the “solved” cases identified through the My Retina Tracker Genetic Testing Program since 2017.
• Free targeted familial variant testing will be available to blood relatives of participants who received an informative test result in the My Retina Tracker Genetic Testing Program.

4. Why do I have to apply for the new Program?

Any individual at the clinic who will be ordering genetic testing is required to submit an application. As part of the application process, you will provide your contact information and some general information about your clinic and agree to the terms and conditions of Program use. The application is intended to ensure compliance with Program guidelines and to facilitate the collection of up-to-date information on ordering clinics so we can direct Program communications to the correct person. The Foundation’s Program staff will monitor the genetic testing results for potential program misuse.

5. What testing options are available to my patient?

There will be two options for IRD genetic testing at no cost to the patient:
• IRD gene panel testing is for participants who have a clinically confirmed diagnosis of an IRD and no first-degree relatives tested through the Program. It is a carefully curated
110-gene panel targeting relevant genes associated with IRDs that account for over 97% of the solved cases in the My Retina Tracker Genetic Testing Program. This panel includes certain mitochondrial genes and full RPGR coverage, including the difficult-to-sequence ORF15 region.

- Targeted familial variant testing is for participants with a blood relative who received an informative genetic testing result through this Program and was tested at PreventionGenetics. The test uses targeted Sanger sequencing for the specific variant(s) identified in their family member.

Of note, additional eligibility criteria apply to each testing option. Please view the IRD Gene Panel Testing and Familial Variant Testing sections for more information.

**IRD Gene Panel Testing**

6. **Which of my patients are eligible to undergo testing with the 110-gene panel?**

Participants who undergo genetic testing with the My Retina Tracker Genetic Testing Program’s 110-gene panel must:

a. Reside in the United States or a US territory.
b. Have a clinically confirmed diagnosis of an IRD studied by the Foundation.
c. Have no first-degree relatives tested through the Program.
d. Have no biological relatives who received informative tests results through the Program.
e. Have not undergone genetic testing with a panel consisting of 32 or more IRD-related genes within the last 5 years.
f. Have not received an IRD-related molecular diagnosis from any previous genetic testing.
g. Be willing to join the My Retina Tracker Registry and share their genetic testing results, including PHI, with the Registry.

7. **Which genes are included in the 110-gene panel?**

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8. Is the new panel performed on a whole genome or whole exome backbone?

The panel is not performed on a whole genome or whole exome backbone. It is a targeted gene panel that is based on a panel-specific library of probes.

9. Why is the number of genes on the panel reduced?

Since the My Retina Tracker Genetic Testing Program was launched in 2017, we’ve learned that 110 of the genes on our original panel account for over 97% of “solved” IRD cases. By offering a more targeted panel and consequently reducing our “per test” costs, the Program, which is largely funded by philanthropic donations to the Foundation Fighting Blindness, can give more IRD patients the opportunity to receive high quality genetic testing while maintaining a comparable diagnostic yield.

10. Does the 110-gene panel cover non-coding regions?

As with our previous panel, supplemental capture probes have been added to ensure coverage of noncoding variants that have been reported as “pathogenic or likely pathogenic” in ClinVar or as "a disease-causing mutation" in HGMD.

11. What if my patient doesn’t receive an informative genetic testing result?

If your patient receives an uninformative genetic testing result, there is still a possibility that he or she has an inherited retinal disease caused by a gene that is not on the My Retina Tracker 110-gene panel. If your patient is interested in pursuing additional testing through insurance or self-pay, PreventionGenetics and several other genetic testing labs offer larger IRD panels.

12. What is the testing strategy for the 110-gene panel?

This test is performed using Next-Generation sequencing with additional Sanger sequencing as necessary. This panel has been validated to provide 99.64% coverage of all coding exons of the genes plus 10 bases of flanking non-coding DNA in all available transcripts along with other non-coding regions in which pathogenic variants have been reported in available databases. We define coverage as ≥20X NGS reads or Sanger sequencing.

Copy number variant (CNV) analysis of the NGS data is included. This panel has been validated to detect 92.86% of CNVs involving 1 exon and 100% of CNVs involving 2 or more exons.

Please note that analysis of RPGR exon 15 (commonly referred to as ORF15) has historically been difficult due to the highly repetitive, purine-rich sequence. However, saturating levels of capture-probes in ORF15 region have now enabled 100% sequencing coverage in the majority of cases. When NGS does not achieve full coverage or there is a variant detected by NGS that needs confirmation, then a specialized chemistry Sanger sequencing is utilized.
13. What is the estimated turnaround time for the My Retina Tracker 110-gene panel?

PreventionGenetics’ standard turnaround time for panel testing is approximately three to four weeks.

Familial Variant Testing

14. Which of my patients are eligible to undergo targeted familial variant testing?

Participants who undergo genetic testing with the My Retina Tracker Genetic Testing Program’s targeted familial variant testing must:

a. Reside in the Unites States or a US territory.

b. Has a blood relative tested through the Program and received an informative genetic testing result through PreventionGenetics.*

c. Have not undergone genetic testing with a panel consisting of 32 or more IRD-related genes within the last 5 years.

d. Have not received an IRD-related molecular diagnosis from any previous genetic testing.

e. Be willing to join the My Retina Tracker Registry and share their genetic testing results, including PHI, with the Registry.

*Of note, the familial variants must be sequence-based (nucleotide substitutions and indels) and within the nuclear genome to qualify for free testing and the participant must meet the following criteria based on the inheritance pattern of the gene identified in the proband:

- Dominant conditions: Targeted testing will be available to all blood relatives on the side of the symptomatic parent. If both parents are asymptomatic, targeted parental testing will be offered to determine if the case is a de novo variant. If one of the parents has the variant, targeted testing will be available to all blood relatives on that side of the family.

- Recessive conditions: Targeted testing will be available to all first-degree relatives (parents, full siblings).

- X-linked: Targeted testing will be available to mother, full male siblings, male maternal half-siblings, and male relatives on maternal side. If there is evidence of manifesting heterozygous women in the family history or the literature, targeted testing will be offered through the same pathway as dominant genes.

Test orders and specimen for familial variant testing must be received within 180 days after the original proband report was issued.

15. What is the estimated turnaround time for familial variant testing?

PreventionGenetics’ standard turnaround time for familial variant testing is approximately four weeks.
16. How can I create an online ordering portal account with PreventionGenetics?

To place genetic testing orders, you will need to create a Program-specific portal account, by accessing the following link: https://myretinatrackerprogram.preventiongenetics.com/.

As part of the account registration process, you will enter a unique access code which will be provided by Foundation Fighting Blindness after your application has been approved. For more information on the application process, please see questions 4 and 5.

If you have questions about your application or access code, please contact Foundation Fighting Blindness at coordinator@myretinatracker.org

If you need assistance while creating your account, please contact PreventionGenetics directly at support@preventiongenetics.com.

17. How do I order a test in the updated Program?

You can order a test in the updated Program in five easy steps:

1. **Share program information with your patient.** Complete the webform provided by the Foundation. Upon submission, your patient will receive an email from the Foundation that includes links to a study flyer and patient FAQ. If the patient elects to move forward with enrollment, they can click on the “Review Documents” button within the email to access the My Retina Tracker Registry and Genetic Testing Program Consents, a HIPAA release form, and the InformedDNA Consent for Services.

2. **Download the patient’s executed HIPAA release form.** When your patient submits the electronic forms, you will receive an email that includes a copy of the executed HIPAA release form. Please download this form as it is required for submission of the Program’s test requisition form.

3. **Complete the test requisition form in your Program-specific PreventionGenetics ordering portal.** Access the Program-specific ordering portal to complete the test requisition form. In addition to providing information about your patient’s clinical presentation, you will need to indicate your preference for genetic counseling and upload the HIPAA release form.

4. **Choose where the sample will be collected.** Based on your preference, you can collect the sample for genetic testing in your clinic or request that a sample collection kit be sent to your patient’s home address as provided in the test requisition form.

5. **Access your patient’s test report:** Test results will be ready approximately 3-4 weeks after the laboratory receives the sample. You can access the report by logging into your ordering portal or by requesting a faxed copy on the test requisition form.
18. **What types of samples will be accepted for testing?**

As part of the testing process, your patient will need to provide a saliva or blood sample. Specimen requirements and shipping information can be found on PreventionGenetics’ website at the following link: https://www.preventiongenetics.com/ClinicalTesting/specimenRequirements.

Of note, test order and specimen for familial variant testing must be received within 180 days after the original proband report was issued.

19. **How do I order PreventionGenetics’ sample kits for sample collection in my clinic?**

Blood or saliva samples can be collected in clinic based on the preference of the provider. To request sample collection kits for your clinic, please visit the following link on PreventionGenetics’ website: https://www.preventiongenetics.com/kits.

If you currently have PreventionGenetics kits on hand, please check the expiration date on the kit to ensure that the collected sample will be valid for testing.

20. **How do I order a PreventionGenetics’ sample kit to be sent to my patient’s home?**

Saliva samples can be collected in your office, or you can request that a sample collection kit be sent to your patient’s home address in the test requisition form. For the latter, your patient will collect their sample using instructions provided in the kit and then return the sample directly to the laboratory in a self-addressed, paid envelope.

21. **How will I receive the results?**

You can access your patient’s test report by logging into your Program-specific ordering portal. You can also request to receive the report by fax on the test requisition form.

22. **Can I see the results of tests ordered by other providers in my practice?**

If providers within a practice are ordering in individual portals, they have the option of setting up “autoshare” in their ordering portal. This allows the test results to be shared across multiple ordering portals so that more than once clinician has access to the result.

For more information on this option, please access the following link: https://preventiongenetics.wistia.com/medias/79krlyue3u

23. **Will the format of PreventionGenetics’ test reports be different from those I’ve received from Blueprint Genetics.**

Each lab has a slightly different format for their test reports and the information included in the report may vary. You can find a sample of PreventionGenetics’ test reports here: https://www.preventiongenetics.com/ClinicalTesting/TestCategory/sampleReports
24. Who do I reach out to with questions about ordering a test?

For questions about portal creation, the genetic testing process, and your patient’s test results contact PreventionGenetics at support@preventiongenetics.com. They have customer service agents who are ready to help and can also connect you with staff genetic counselors, if needed.

My Retina Tracker Registry

25. Does my patient have to join the Registry?

Yes, one of the requirements for Program participation is for patients to enroll in the Foundation’s My Retina Tracker Registry (MRTR). The goals of MRTR are to support the understanding and research of inherited retinal dystrophies (IRD). In joining the Registry and sharing their genetic testing results with the Foundation, participants contribute to a database that helps researchers understand the relationships between genotype and phenotype, prevalence, and distribution of the diseases. Participants also benefit from receiving communications regarding relevant research studies or clinical trials that they may qualify for based on their Registry profile. The Foundation does not use the Registry for fundraising.

26. How do I collect consent for the Registry?

In our updated Program, the ordering provider is no longer required to review the informed consent with their patient and collect his or her signature. The Foundation will provide a link to the Program webform. After the provider submits a completed webform, the patient will receive an email from DocuSign that includes links to the study flyer and a patient FAQ. If the patient elects to move forward with enrollment, they can access and complete the Study consent forms, HIPAA release form, and InformedDNA Consent for Services by clicking on the DocuSign button. Any Program-related questions your patient may have should be directed to the Foundation Fighting Blindness.

When your patient submits their online forms, you will receive a copy of the executed HIPAA release form. Uploading this form into the test requisition form is a requirement for submission.

Genetic Counseling

27. 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 that the participant fully understands the medical and familial implications of their genetic diagnosis.

When you submit the test requisition form you will need to select one of two options for genetic counseling:
The Foundation offers genetic counseling at no cost to the participant through InformedDNA. If you select this route, the laboratory will send a referral to the collaborating genetic counseling provider when your patient’s results are available. Upon receipt of the referral, the genetic counseling provider will contact your patient to schedule a tele-genetic counseling appointment.

If you or a member of your staff would prefer to provide genetic counseling to your patient, you can indicate this on the form.

Of note, no-cost genetic counseling is not available for My Retina Tracker Genetic Testing Program participants undergoing targeted familial variant testing. The ordering provider or their clinic staff will be expected to share the results with the patient and counsel accordingly.

28. What happens if I change my mind regarding the mode of counseling after submitting the test requisition form?

Unfortunately, we are not able to accommodate changes to the selected mode of genetic counseling once a test requisition form has been submitted.

29. If I refer my patient to IDNA for counseling, how soon will the patient be contacted for their counseling session?

Patients will be contacted by InformedDNA, generally within 48-72 hours after the results are available.

30. Will I get a copy of a genetic counseling report?

Yes, once InformedDNA finishes the genetic counseling session both the ordering provider and the patient will receive a copy of the post-counseling report.

Transition Information

31. Do I need to submit this to my IRB?

The My Retina Tracker Genetic Testing Program is covered under a protocol that has been reviewed and approved by WCG IRB. Our IRB does not require centers ordering genetic testing as part of the Program to undergo a separate IRB review. However, we are happy to provide a copy of our protocol for your IRB, if needed.

32. What if I’m part of the Foundation’s Genetic Testing Study?

Given our new consenting and ordering process, we will no longer have two programs (My Retina Tracker Genetic Testing Study vs Open Access). While our IRB does not require centers ordering genetic testing as part of the Program to undergo a separate IRB review, we are happy to provide a copy of our protocol for your institutional review board, if needed.

33. When is the last day that an order can be submitted to Blueprint Genetics?
July 25th, 2024. After this date both My Retina Tracker program panels (Open Access and IRB) will no longer be orderable in Nucleus.

34. When is the last day to resolve order issues (e.g. missing consent, unclear orders, eligibility issues, etc.)?

August 16, 2024. Orders on hold that are still unresolved after this date will be cancelled.

35. When is the last day that a sample can be submitted to Blueprint Genetics?

August 16th, 2024. Incomplete orders after August 16 will be cancelled. Samples received after this date will not be tested.

36. What will happen to the MRT data collected on tests performed at Blueprint Genetics?

Data on tests performed at Blueprint Genetics as part of the My Retina Tracker Genetic Testing Program will be stored in the Foundation’s My Retina Tracker Registry database if the patient consented to join the Registry at the time of testing.

Providers will continue to have access to the test results in their Nucleus accounts for as long as the account remains active. For data security reasons, Nucleus accounts will be inactivated if the provider does not change the password once per year. A reminder email is sent 30 days and 7 days before the password is due to change. To ensure uninterrupted access to past results, it is recommended that providers download and save their results from Nucleus to a separate and secure location.

Nucleus accounts can be reactivated by contacting Blueprint Genetics Customer Support at support.us@blueprintgenetics.com.