



**ID
YOUR
IRDSM**

**INHERITED RETINAL DISEASE
GENE TESTING INITIATIVE**

**GUIDE FOR PATIENTS
AND CAREGIVERS**

Through the **ID YOUR IRD** gene testing initiative, Spark Therapeutics aspires to help people living with certain inherited retinal diseases (IRDs) by providing access to genetic information that can empower their decisions.

This initiative is open to U.S. residents only, subject to the terms and conditions of the program.



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OVERVIEW

The ID YOUR IRD initiative explained.

Before you decide if the **ID YOUR IRD** gene testing initiative may be right for you, it helps to be informed. Here's an overview of who's involved and what happens during this gene testing initiative.

ID YOUR IRD IS SIMPLE AND FREE.*

The **ID YOUR IRD** gene testing initiative is specifically designed to be easily accessible. The genetic test and optional, confidential telephone consultation sessions with a genetic counselor are available at no cost to all eligible U.S. residents with certain inherited retinal diseases (IRDs), upon authorization by their doctor. The **ID YOUR IRD** gene testing initiative will not be able to provide genetic information to everyone with an IRD, but the initiative aims to help inform as many people as possible.

We have partnered with PreventionGenetics, a CLIA- and ISO-accredited laboratory, to provide the genetic test at no cost to you. Regardless of insurance status, **ID YOUR IRD** will be available free of charge to all eligible U.S. residents. Your information is confidential and will not be shared with anyone, nor used for any purpose, not authorized by you. Please see the terms and conditions of the **ID YOUR IRD** initiative on pages 12-13.

*Participation in the **ID YOUR IRD** gene testing initiative is subject to the terms and conditions of the program. Participants will receive genetic testing and optional, confidential genetic counseling sessions at no cost to them regardless of their insurance status. Although participants will not pay for the costs of genetic testing offered by the initiative, or the optional genetic counseling, participants are responsible for any other costs they may incur, such as doctor's visits.



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GENE TESTING INITIATIVE

WHO'S INVOLVED.

PATIENT LIAISON

Patient Liaisons are representatives from Spark Therapeutics who can help to evaluate if you may be eligible to participate in the **ID YOUR IRD** gene testing initiative. Your Patient Liaison is available to help answer your questions about the initiative and will help you understand the consent process that is necessary to participate in the initiative.

YOUR IRD DOCTOR

The doctor you consulted with for your IRD is your main **ID YOUR IRD** companion and works with you to determine your eligibility. If so, he or she authorizes your participation, and will ask you or your parent/legal guardian to fill out a consent form to enroll in this initiative. Your doctor will then order your genetic test, and contact you to discuss your genetic test results when they become available.

GENETIC COUNSELOR

If you are eligible for the **ID YOUR IRD** initiative, and your doctor authorizes your participation, Spark Therapeutics will cover the cost of optional telephone consultation sessions with a genetic counselor. Genetic counselors provide personalized care to help people understand and adapt to the medical, psychological and familial implications of genetic conditions. Your Patient Liaison from Spark Therapeutics can connect you with a genetic counselor who can help you and your doctor make informed decisions based on complex genetic information.

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INHERITED RETINAL DISEASE
GENE TESTING INITIATIVE

WHAT TO DO.

DECIDE IF THIS INITIATIVE MAY BE RIGHT FOR YOU

The **ID YOUR IRD** 4-question screener will determine if you prequalify for our gene testing initiative. If you do, one of the Patient Liaisons from Spark Therapeutics will be in touch to answer questions and discuss eligibility. In order to participate, your doctor must confirm your eligibility and authorize and order the test for you. The **ID YOUR IRD** gene testing initiative will not be able to provide genetic information to everyone with an IRD, but the initiative aims to help inform as many people as possible. Participation is subject to the terms and conditions of the **ID YOUR IRD** initiative.

SCHEDULE YOUR DOCTOR VISIT

The next step is to schedule an appointment with your doctor to discuss participating in the **ID YOUR IRD** initiative. It is your doctor's role to confirm that you are eligible, get your consent to participate, and authorize your genetic test. Your doctor will ask you or your parent/legal guardian to fill out a consent form. You will also need to provide your doctor with a blood or saliva sample, which will then be sent to PreventionGenetics, an accredited laboratory, for testing and analysis.

DISCUSS THE RESULTS

Results may take up to 6 weeks, but typically arrive within 30 days. Your doctor will walk you through your results to answer questions you and your family might have. You can also ask your Patient Liaison to put you in touch with a genetic counselor at no cost to you. All participants whose doctor orders the genetic test will receive results, which may be "Positive," "Negative," or "Inconclusive."



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INHERITED RETINAL DISEASE
GENE TESTING INITIATIVE



ELIGIBILITY AND PREQUALIFICATION

- Complete the 4-question screener
- Talk to a Patient Liaison from Spark Therapeutics



TALK TO YOUR DOCTOR

- Schedule an appointment with your doctor to discuss ID YOUR IRD
- Eligibility confirmed and patient consent form completed
- Doctor authorizes your participation and orders your genetic test
- Doctor sends blood or saliva sample to laboratory
- Genetic counseling session via telephone (optional and confidential)



LABORATORY (4-6 WEEKS)

- Sample analyzed at PreventionGenetics, a CLIA- and ISO-accredited laboratory
- Test results sent back to your doctor



DISCUSS YOUR RESULTS

- Once received, your doctor will contact you to discuss the test results
- Genetic counseling session via telephone (optional and confidential)



ID YOUR IRDSM
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GENE TESTING INITIATIVE

What do these test results mean?

GET THE FACTS.

You have questions about your IRD. Genetic testing may provide the answer.

HOW GENETIC TESTING WORKS.

Genetic testing is a process that may identify changes or mutations in a person's genes. Because genetic mutations may cause health issues, genetic testing could help explain how and why a disease affects a person. For those with an inherited retinal disease (IRD), it may identify the cause of vision impairment.

If you had a negative or inconclusive genetic test more than 5 years ago, think about getting tested again.

Genetic tests have evolved with the identification of new genes, and one of the latest tests may provide more information. The **ID YOUR IRD** gene testing initiative will not be able to provide genetic information to everyone with an IRD, but the initiative aims to help inform as many people as possible. Participation is subject to the terms and conditions of the **ID YOUR IRD** initiative.



ID YOUR IRDSM
INHERITED RETINAL DISEASE
GENE TESTING INITIATIVE

ID YOUR IRD PROVIDES 1 OF 3 TYPES OF RESULTS.

POSITIVE

A positive result means that the test found a mutation (or mutations) on a specific gene that is known to cause an inherited retinal disease.

NEGATIVE

A negative result means that the test did not identify a genetic cause of disease - but this does not rule out the diagnosis of an inherited retinal disease. The **ID YOUR IRD** gene panel only includes a select number of genes in which mutations have been found to cause certain early-onset, rod-mediated IRDs. There are many genes in which mutations have been found to cause IRDs and additional testing may be available through your doctor.

INCONCLUSIVE

An inconclusive result (also known as a “variant of uncertain significance”) means that one or more mutations were found, but it is not yet known whether these mutations cause disease. An inconclusive test result does not rule out the diagnosis of an inherited retinal disease and, with time, research may reveal more information about these variants and their association with IRDs. Genetic counseling may help with the understanding of an inconclusive result and additional testing may also provide more information.



ID YOUR IRDSM
INHERITED RETINAL DISEASE
GENE TESTING INITIATIVE

ID YOUR IRD can only provide information about retinal diseases.

As an IRD genetic test, results can only reveal information about certain genes that may affect your vision, and cannot provide any information about other diseases such as cancer or diabetes.

Doctors help you interpret ID YOUR IRD results.

Genetic test results can sometimes be unclear or hard to interpret—but your doctor can help. They guide you through the testing journey and can answer questions about your results, or refer you to someone who can. You can also ask your Patient Liaison from Spark Therapeutics to put you in touch with a genetic counselor to discuss your results, at no cost to you.

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GENE TESTING INITIATIVE

Think ID YOUR IRD may be right for you?

ASK YOUR DOCTOR.

Asking the right questions can lead to the right conversation.

You've gotten the facts and taken the **ID YOUR IRD** 4-question screener. Now it's time to ask your doctor if **ID YOUR IRD** could be right for you. If he or she feels that it is, your doctor must authorize your participation and order the test on your behalf. Even if you're still unsure about the initiative, learning how to start the conversation with your doctor, and what to ask, could help inform your decision.

Use our Doctor Essentials as a conversation starter.

Unsure what to tell your doctor about **ID YOUR IRD**? The information contained in the Doctor Essentials guide can help. It contains information on your doctor's role, a guide for determining patient eligibility, and more.



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GENE TESTING INITIATIVE

Ask your doctor the right questions.

It's important that you and your family are informed before, during, and after the **ID YOUR IRD** initiative. A good way to ensure this happens is by asking questions. Here are some to keep in mind when talking with your doctor:

- What does getting a genetic test involve?
- Is getting a genetic test like this one right for me?
- Who has access to my information?
- Have I had a genetic test before? What were the results?
- My **ID YOUR IRD** test results were negative or inconclusive. What now?
- My **ID YOUR IRD** test results identified one or more genetic mutations causing my retinal disease. What are my options?

Genetic counselors can help.

Genetic counselors can provide valuable information during the testing initiative and can help interpret test results. Once you have enrolled in **ID YOUR IRD** and provided your blood or saliva sample, your Patient Liaison from Spark Therapeutics can help set up optional telephone consultations with a genetic counselor for you. These consultations are completely confidential.



ID YOUR IRDSM
INHERITED RETINAL DISEASE
GENE TESTING INITIATIVE

Want to learn more about IRDs and genetic testing?

RESOURCES

ID YOUR IRD resources.

Whether you're interested in the **ID YOUR IRD** gene testing initiative or just searching for more information on inherited retinal disease (IRD) genetic testing, the following resources may be able to help.

IRD and genetic testing information

- Foundation Fighting Blindness - www.fightblindness.org
- My Retina Tracker - www.myretinatracker.org
- aSharedVision - www.asharedvision.com
- Retina International - www.retina-international.org
- RetNet - www.sph.uth.edu/retnet
- National Society of Genetic Counselors – www.aboutgeneticcounselors.com
- Genetic Alliance - www.geneticalliance.org; www.ginahelp.org
- National Institutes of Health Genetic Testing - www.ghr.nlm.nih.gov/primer/testing/geneticctesting



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INHERITED RETINAL DISEASE
GENE TESTING INITIATIVE

THE ID YOUR IRD INITIATIVE TERMS AND CONDITIONS

ID YOUR IRD is a genetic testing initiative (“the Initiative”) supported by Spark Therapeutics, Inc. Your participation in the Initiative and use of Spark Therapeutics content and services is subject to the terms of the agreement between you and Spark Therapeutics set forth in these Initiative Terms and Conditions, which incorporate by reference the Spark Therapeutics general Terms of Use and Privacy Policy. You may accept these Initiative Terms and Conditions by signing the authorization provided by your healthcare professional to participate in genetic testing and share your genetic test results with Spark Therapeutics. You may not participate in the Initiative if you do not accept these Initiative Terms and Conditions.

To be eligible to participate in the Initiative, you must (1) be a US resident at the time you are tested and receive your test results; (2) have a type of inherited retinal disease (IRD) that makes you eligible for the Initiative as determined by your healthcare professional; (3) have the approval of your healthcare professional to have the genetic test; and (4) authorize in writing that your healthcare professional and the genetic testing laboratory selected by Spark Therapeutics may test the genetic sample you provide and share your name, contact information and information regarding your condition, diagnoses, and results of your genetic testing (collectively, “Your Information”) with Spark Therapeutics. If you are under the age of 18, you must have the approval of your legal guardian to participate in the Initiative.

The genetic test provided under the Initiative requires you to provide a saliva or blood sample to your healthcare professional. Your sample will be analyzed by a genetic testing company selected by Spark Therapeutics, and the results will be provided to your healthcare professional and to Spark Therapeutics. The genetic testing company that performs the test is independent from Spark Therapeutics and Spark Therapeutics has no control over or influence on how the test is conducted. You understand and agree that by participating in the Initiative, Spark Therapeutics will process, use and disclose Your Information only as permitted by your written authorization and the Spark Therapeutics Privacy Policy.

Through the Initiative, you will be offered an optional opportunity to discuss your genetic test results by telephone with a genetic counselor. If you choose this option, any advice provided by the counselor is independent of Spark.



ID YOUR IRDSM
INHERITED RETINAL DISEASE
GENE TESTING INITIATIVE

If you choose to participate in the Initiative, you will not be responsible for the costs of the genetic test itself or genetic counseling offered under the Initiative. Please be aware, however, that you will be responsible for any other costs that may be incurred as a result of participating in the Initiative, including but not limited to the costs of visits or consultations with your healthcare professional in connection with the genetic test or the testing results.

The Initiative and any content or services provided by Spark Therapeutics are for informational and educational use only. The Initiative does not test for every gene mutation, nor are the results of the testing performed in connection with the Initiative intended to be comprehensive. The results of the genetic test provided to your healthcare provider by the testing company may be: (1) positive (if the gene associated with the IRD has been identified by the test); (2) negative (if no genetic cause of the IRD has been identified by the test); or (3) inconclusive (if the test identified a genetic mutation, but it is unknown whether the identified mutation causes an IRD). You should consult with your own healthcare professionals about your diagnoses, genetic testing, and genetic testing results. Spark Therapeutics does not provide medical advice, and the results of the Initiative are not intended to be used by you for any diagnostic purpose or as a substitute for professional medical advice. Spark Therapeutics does not endorse, warranty, or guarantee the effectiveness of any specific course of action, resources, tests, physicians or other healthcare professionals, drugs, biologics, medical devices, products, procedures, opinions, or other information that may be offered to you or become available to you through the Initiative. Reliance on any information provided by Spark Therapeutics is solely at your own risk.

By participating in the Initiative, you understand and agree that you acquire no rights in any research or commercial products that may be developed by Spark Therapeutics and/or its collaborating partners. You specifically understand that you will not receive compensation for any research or commercial products that include or result from Your Information.

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