GENETIC TESTING FOR RETINAL DISEASES

A resource for the inherited retinal disease community
WHAT IS GENETIC TESTING?

Genetic testing is a process that identifies changes, called mutations, in a person’s genes that may cause health conditions.

- A genetic test may give people with an inherited retinal disease (IRD) and their families important health information.
- Because genetic mutations may be the cause of one or more health conditions, a genetic test may help explain how and why ocular or another disease affects a person, and/or their family members.
- For people with an IRD, genetic testing may be especially important for identifying the cause of their vision loss.
- A genetic test can be done using a blood or saliva sample.

Your healthcare professionals (HCPs)

Along their healthcare journey, people with an IRD might consult with various HCPs, including their family doctor, pediatrician, ophthalmologist, retinal specialist, genetic counselor, nurse, or other medical professional.
Genetic testing may help empower people with an IRD and their families

- For people with an IRD, a genetic test is the only way to know exactly which genetic mutation they have, which can better help them to address health issues related to that mutation.
- A genetic test may help people with an IRD and their family members plan for the future of that person’s vision and health.
- Identifying a specific genetic mutation may help people with an IRD and their family members find clinical trial opportunities.
- A genetic test may help people with an IRD and their family members gain access to supportive educational and social services.
- Knowing the mutation that causes an IRD may help people with an IRD and their family members to connect with others living with the same type of vision impairment.
A genetic test may provide one of three different results

- **Positive**: When a genetic test returns a positive result, it means the test found mutations on a specific gene that, for people with an IRD, can identify the cause of their vision impairment.
- **Negative**: A negative result means that the test was not able to identify a specific genetic cause for the disease—but this does not rule out the diagnosis of a retinal disease.
- **Inconclusive**: When a genetic test is inconclusive, it does not mean a person has no genetic mutations—it simply means that the test didn’t find what it was specifically looking for. This might be because it was looking at a gene panel that doesn’t affect that particular person. Or it might mean that the person’s mutated gene has not been identified yet. An inconclusive result sometimes means that additional testing might be needed.

What it won’t tell you

When people get a genetic test to help identify an IRD, the test will be specific only for those genes, and not reveal any risk for cancer, diabetes, or other health issues not related to retinal diseases.
WHAT DOES GETTING A GENETIC TEST INVOLVE?

1: Discuss genetic testing with your HCP. Start a conversation with your doctor, ophthalmologist, or other HCP about getting a genetic test to help identify the underlying cause of your vision loss. Your HCP can order a genetic test or refer you to a specialist to discuss it further.

2: Speak to your HCP about scheduling an appointment with a genetic counselor. Genetic counseling is a process that helps people to understand issues that arise from having an inherited medical condition—for both the person with the disease and for other members of their family. Get a referral to speak with a genetic counselor who specializes in ophthalmic conditions. He or she can explain the potential benefits of genetic testing and help you navigate the process. If you decide that genetic testing is right for you, ask your HCP or genetic counselor to order the genetic test.

3: Visit the HCP’s office to have blood or saliva collected for the test. A genetic test can be done using either a blood or saliva sample, depending on the kind of test it is and where it is performed. Your HCP will know which type of sample needs to be collected.

4: Discuss the test results with an expert. Depending upon the test ordered, results typically take between 6 weeks and 4 months to come back to your physician. Genetic test results are sometimes unclear or hard to interpret (even for some HCPs). Your physician or a genetic counselor who specializes in ophthalmic conditions can help you understand what the results mean.

Remember, results may or may not identify a specific genetic mutation. Currently, genetic testing can identify the underlying cause of disease in many patients with retinitis pigmentosa (RP) or Leber congenital amaurosis (LCA).

5: Get connected. Once you receive and understand your results, it may be beneficial to seek out patient advocacy groups or online communities. You can also contribute your information to a disease registry, like myretinatracker.org.

The process for getting a genetic test generally involves a few simple steps

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Here are some questions that people considering genetic testing often ask

What will a genetic test cost? The cost will vary depending on which test is ordered and where it is done. A person’s insurance plan may cover these costs. Some patient advocacy organizations and research centers may offer free testing. A genetic counselor may help make the case for reimbursement of a genetic test to the insurance company.

Are genetic test results confidential? Yes, like all personal health information, genetic test results are confidential. Only you, the HCP who ordered the test for you, and the laboratory will know your results. Of course, you can share the results with other individuals if you choose to do so.

Genetic testing for the IRD was done in the past. Should another test be done now? If the genetic test was more than 5 years ago and you didn’t receive a conclusive result—or any result at all—think about getting tested again. Genetic tests have improved, research is advancing, and a person may learn more from a newer test, especially with the identification of new genes.

What if I don’t have a doctor to help me order the test? For information on finding an HCP who can discuss genetic testing with you, please contact one of the resources listed on page 14.

What if I cannot understand the results? Discuss the results with your HCP, or ask for a referral to a genetic counselor who can help explain the results to you.
When it’s time to talk to your HCP, here are some topics you might want to discuss

- Do you work with a genetic counselor that I should talk to?
- Can you confirm from my medical records whether a genetic test for my IRD has been done in the past? If yes, were results received and if so, what were they?
- If I haven’t had testing completed in the past, can we discuss having genetic testing done now?
- What if I have genetic testing done and the test result is inconclusive?
- If my test comes back with a conclusive result, what are my next steps?

More information about genetic testing can be obtained from the following resources

- aSharedVision - asharedvision.com
- Foundation Fighting Blindness - fightblindness.org
- The National Society of Genetic Counselors - nsgc.org
- The National Institutes of Health - ghr.nlm.nih.gov/primer/testing/genetictesting
Resources

More information can be found at the following websites:

- aSharedVision  asharedvision.com
- Choroideremia Research Foundation  curechm.org
- Curing Retinal Blindness Foundation  crb1.org
- Foundation Fighting Blindness  fightblindness.org
- Genetic Alliance  geneticalliance.org
- Genetic Information Nondiscrimination Act  ginahelp.org
- National Society of Genetic Counselors  nsgc.org
- Sofia Sees Hope  sofiasees.org
- Spark Therapeutics  sparktx.com
- The RDH12 Fund for Sight  rdh12.org
- WonderBaby.org  WonderBaby.org

Notes

Use this page to jot down any additional items you would like to discuss with your healthcare professional.
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We would like to acknowledge and thank the following organizations for their contributions in developing this brochure:

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