

## Get started TALK TO YOUR FAMILY

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### TALK TO YOUR FAMILY

Talk to your family about your decision to pursue genetic testing – and options available to them, as well.

If you or your loved one has an inherited retinal disease (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 you or your family members plan for the future.
- Identifying a specific genetic mutation may help you and your family members find clinical trial opportunities to help advance research.
- A genetic test may help you and your family members gain access to supportive educational and social services.
- Knowing the mutation that causes an IRD may help you and your family members to connect with others living with the same type of vision impairment.

#### **Tips for discussing genetic testing for IRDs with your family:**

- Share with your family why you're interested in or have decided to pursue genetic testing.
- Communicate facts about genetic testing, the process and how the knowledge may benefit you and your family.
- Ask questions about family medical history related to vision loss that may help as background for your health care professional or genetic counselor.
- Ask if any other family members – of any age – with vision loss are interested in pursuing genetic testing and offer to support them in navigating the process or connect them to additional resources.

#### **After you receive your results:**

If you receive a conclusive result from your genetic test, genetic counseling provides tremendous support for individuals and families. Work with your genetic counselor to understand your results and get tips that may help you talk with your family.

## Get started TALK TO YOUR INSURER

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The cost associated with genetic testing will vary depending on which test is ordered and where it is done. Some patient advocacy organizations and research centers may offer free testing.

Your insurance may cover the costs of genetic testing. A genetic counselor may also be able to help make the case for reimbursement of a genetic test to your insurance company.

#### Questions for your insurer may include:

- Are the costs of genetic testing covered in whole or in part?
  - What is the cost of a genetic test in full?
- Does insurance cover genetic counseling?
- What is the total cost I am responsible for after I meet my deductible?
- Is a referral or pre-authorization required for genetic counseling to be covered?
- Is a referral or any pre-authorization required for the testing?

Insurance issues can be complicated and documenting conversations with your insurer may help you stay organized. That information can also be shared with your health care professional if you need support related to insurance coverage or referrals.

## Get started TALK TO YOUR HEALTH CARE PROFESSIONAL

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### ONCE YOU'VE DECIDED “EYE WANT 2 KNOW,” IT’S TIME TO ACT

Learning the specific gene mutation that causes your or your family member’s inherited retinal disease may lead to opportunities for management or participation in clinical research.

### TALK TO YOUR HEALTH CARE PROFESSIONAL

#### **How do I know if my diagnosis is an inherited retinal disease?**

Inherited retinal diseases (IRDs) are a group of rare eye disorders caused by an inherited gene mutation and can result in vision loss or blindness.

If you or your loved one has been diagnosed with a degenerative eye disease such as retinitis pigmentosa (RP), choroideremia (CHM) or juvenile macular degeneration (JMD), there will be a gradual loss of vision, eventually leading to complete blindness.

Others, with congenital conditions like Leber congenital amaurosis (LCA) or cone-rod dystrophy (CRD), are born with or experience vision loss in infancy or early childhood.

#### **What should I do if I think genetic testing is right for me?**

Talk to your health care professional (HCP) about genetic testing options.

Your doctor can provide information and guidance about genetic testing. They can also help you interpret results.

Whether you start your quest for a genetic diagnosis with a family doctor, pediatrician, ophthalmologist, retinal specialist, genetic counselor, nurse, or other medical professional, download the HCP discussion guide to help guide the conversation with your HCP.

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## Get started TALK TO YOUR HEALTH CARE PROFESSIONAL

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### **Other tips for discussing genetic testing with your HCP:**

- If you feel comfortable doing so, bring a friend or family member with you who can take notes or help you remember specific information and answers you receive from your HCP.
- Don't be afraid to ask your HCP, such as a genetic counselor, to clarify information you don't understand about genetic information or the genetic testing process.
- Some HCPs may not be as familiar with genetic testing for IRDs – directing them to [www.EyeWant2Know.com](http://www.EyeWant2Know.com) for context about your desire to pursue genetic testing may be helpful.

### **When you receive your results:**

- Discuss the test results with an expert. Depending upon the test, results typically take between 6 weeks and 4 months to come back to your health care professional. Genetic test results are sometimes unclear or hard to interpret (even for some HCPs). Your health care professional or a genetic counselor who specializes in ophthalmic conditions can help you understand what the results mean. Results may or may not identify a specific genetic mutation.
- Get connected. Once you receive and understand your results, it may be beneficial to seek out patient advocacy groups or online communities, like those highlighted on [www.EyeWant2Know.com](http://www.EyeWant2Know.com) or [www.aSharedVision.com](http://www.aSharedVision.com). You can also contribute your information to a disease registry, like [www.myretinatracker.org](http://www.myretinatracker.org).