

GENETIC TESTING & RETINAL CONDITIONS 101



VISION HEALTH
ADVOCACY COALITION



Perspective from
Kari Branham, MS, CGC



What are retinal diseases?

The eye's retina is like the film in a camera. It captures an image and transmits it to the brain. Retinal diseases are vision conditions that impact the retina's ability to function. Depending on the specific impact, these diseases can lead to central or peripheral vision loss.

Retinal diseases may have different causes. Some are genetic conditions that occur when a change or mutation in a gene stops the gene from working properly. These kinds of conditions are called inherited retinal diseases.



Why conduct genetic testing for retinal conditions?

There are lots of reasons to consider having genetic testing done. It can help confirm or identify the specific diagnosis. It can also determine whether other members of the family have the same condition. In addition, genetic testing can help determine if there are treatments that may be effective in treating the condition.



What are the benefits of genetic testing?

To treat a condition effectively, providers and patients have to know what they're up against. Genetic testing helps take that first step. It gives patients a deeper understanding of their condition. Tests can sometimes identify genetic conditions that impact not only the retina, but also other parts of the body. These deeper underlying conditions may go unnoticed without genetic testing.

And genetic testing also helps target treatment specifically to a patient, leading to personalized, patient-centered care. For example, tests can help providers understand what kind of treatments may or may not be valuable for their specific condition. Gene therapy trials, for example, target a specific gene or genetic mutation. These treatments can only help when patients have a certain genetic condition.



Genetic testing also helps target treatment specifically to a patient, leading to personalized, patient-centered care.



What's preventing patients from accessing genetic testing?

A lack of awareness and access. Being aware of genetic testing and how it can help is one of the first big hurdles for patients. Many don't know that genetic testing is available, could help better explain a vision condition or could even point them in the direction of treatment.

Another hurdle is finding someone who can order the test and properly interpret its results. While more ophthalmologists and optometrists are ordering testing, the field is still new enough that answering the question "What comes next?" is still tricky.



By improving access to testing and the right care, patients with retinal conditions will be able to find the treatment that is best for them.



How can policymakers help improve access to genetic testing for vision conditions?

Ensuring that patients have access to testing is important. Some patients may have access to genetic testing through a sponsored program or a research study. That type of testing, however, may not be accessible for everyone. Increasing insurance coverage for genetic testing is an important step to improving access.

And once patients have access to these tests, they need them explained properly. Beyond just access to genetic tests, patients need access to genetic counseling. By improving access to testing and the right care, patients with retinal conditions will be able to find the treatment that is best for them.



VISION HEALTH
ADVOCACY COALITION

[VisionHealthAdvocacy.org](https://www.VisionHealthAdvocacy.org)

