WHAT ALL DOCTORS OF OPTOMETRY SHOULD KNOW

Expert Insights on Genetic Testing for Patients with Inherited Retinal Diseases

Genetic testing and genetic counseling are critical for patients with inherited retinal diseases (IRDs). Genetic testing is an important step in confirming patient diagnoses and informing treatment approaches. Recent advancements in genetic testing have led to new hope and new options for patients with IRDs. The standard of care for IRD patients is evolving with scientific confirmation of disease-causing genetic variants and the availability and accessibility of genetic testing. Doctors of optometry play a key role in examining, testing and caring for patients with IRDs. AOA convened a group of experts to discuss genetic testing for IRDs.

Benefits of genetic testing for IRDs

- Genetic testing is crucial for patients to understand the genetic cause of their IRD or associated syndromic condition.
- Genetic testing can be used to confirm a diagnosis, aid in determining prognosis, understand inheritance risks, and inform eligibility for clinical trials.
- Currently, there is one approved gene therapy for IRD (Luxturna™ for biallelic RPE65 mutation-associated retinal dystrophy). Genetic testing is necessary to determine eligibility for treatment.

How to identify the right patients for testing

- Doctors should always approach genetic testing with a clinical hypothesis in mind.
- Doctors and patients should recognize that the results may not always be immediately actionable but may become actionable as research progresses and treatments become available.
- Always consider the patient. Some patients may not yet fully understand or accept their IRD and the future changes they may have to their eye health and vision.
- Patients and family members may benefit from genetic counseling before and after genetic testing.
- Remember: Genetic testing is recommended for patients with IRDs and can provide valuable insight into the clinical condition and inform continued management decisions.

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Challenges to be aware of

- Currently, genetic testing may not always identify the cause of an IRD. However, knowledge about IRDs is growing. Patients with previous negative or inconclusive tests may benefit from retesting.
- Retina conditions can look very similar. Patients with the same pathogenic gene variations (mutations in the DNA) can have differing presentations, even among family members.
- There are many variants (mutations in the DNA) that are currently of uncertain significance.
- Results can change with time. As variants are reclassified, test reports may be updated. Genetic counselors and clinical geneticists can assist with test interpretation.
- Genetic test results require interpretation within the context of clinical findings and family history. A full baseline clinical evaluation and continued follow-up evaluations are indicated.

Referrals are critical

- Doctors of optometry should recommend genetic testing for IRDs and either provide testing and counseling services in-office or refer to facilities that do.
- Utilize your colleagues trained in providing low vision care for patients. Primary care optometrists should refer concurrently to low vision optometrists to help support functional vision for the patient. Know your colleagues in your community and nearby.
- Ultimately, our goal is improving the patient's quality of life.

Additional resources

- AOA Low Vision Resources
- Foundation Fighting Blindness
- EyeGene
- ClinicalTrials.gov
- eyesongenes.com

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