Retina Consultants of Texas

RCA Research

INHERITED RETINAL DISEASES

What are Inherited Retinal Diseases (IRDs)?

IRDs are a unique and rare group of disorders of the retina (and sometimes other organs) that can result in severe vision loss. IRDs are often genetically passed down from one's parents and require one or more genes to function incorrectly. IRDs come with a large range of symptoms. A patient with one IRD can present very differently than a patient with a different IRD. You may hear them also referred to as **retinal dystrophies** or **degenerations**.

How do we classify IRDs?

IRDs come in many different forms and affect patients in various ways. Some will primarily affect the central vision; these are often called **macular dystrophies**. Others may primarily affect the peripheral vision and night vision; these are often called **rod** or **rod-cone dystrophies**. There are many classifications and names for IRDs.

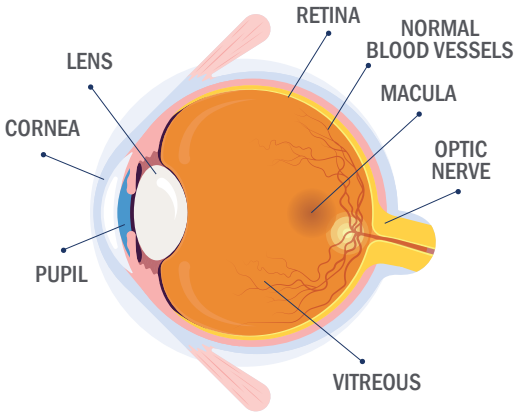
What are the most common types of IRDs?

The most common IRDs are **retinitis pigmentosa** (RP), which is a term that includes many different rod and rod-cone dystrophies, and **Stargardt disease**, which is a macular dystrophy.

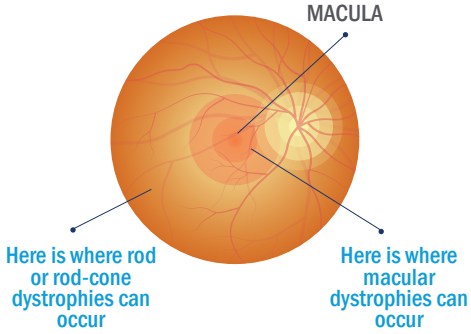
Genetic inheritance

By definition, most IRDs are passed down by one or both of a patient's parents. Defects or mutations in different genes can cause different symptoms and diseases in patients. IRDs are so complex that even the same gene mutation can cause two different types of diseases. In a disease like retinitis pigmentosa, there have been over 100 different mutations detected as possible causes of the disease.

HEALTHY EYE



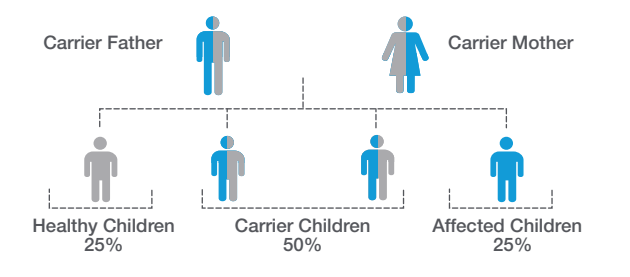
ANGLED VIEW



What are some of the common symptoms of IRDs?

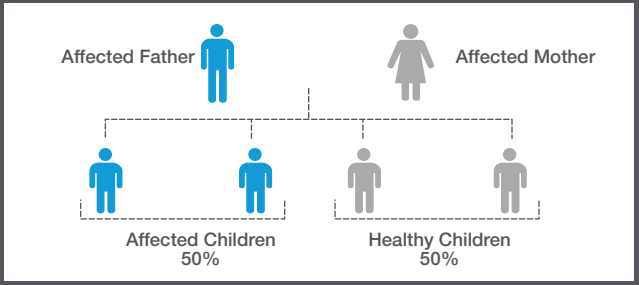
- Night blindness
- Peripheral vision loss
- Tunnel vision
- Color vision loss
- Light sensitivity

Autosomal Recessive



An autosomal recessive pattern is one where a variant gene shows symptoms only if both genes are passed on to the patient. This typically only occur if both parents are carriers, but do not have the disease. In this scenario, the probability of having an affected child is 25%.

Autosomal Dominant



An autosomal dominant pattern is one where a variant gene shows symptoms if one variant gene is passed on from one parent. In this case there is typically at least one affected person in each generation of the family. In autosomal dominant diseases, the probability of having an affected child is 50%.

Genetic testing

Considering genetic testing is important for several reasons:

- Diagnose a potential cause of vision loss
- Predict a possible future prognosis
- Counsel family members such as siblings, cousins, children, and grandchildren
- Discuss the possibility of future research, clinical trials, and gene therapy

Treating IRDs

There is currently no single treatment that can cure an IRD, however the future is bright. In 2017, the FDA approved the very first treatment for an IRD, a condition called Leber Congenital Amaurosis in patients with the RPE65 mutation.

For some IRDs, eyedrops or medicines can help with day-to-day visual function. Ask your doctor if this might help in your condition.

Research and clinical trials

Clinical trials to advance medicine and treatments are important to us, and we are heavily involved in many trials, including those for IRDs. If you are interested in participating in research, please ask your doctor. Your eligibility may depend on your genetic diagnosis as well as other clinical criteria.

If you have an IRD, you should:

- See your PCP regularly
- Always wear UV blocking eye wear in the sun
- Eat a healthy, balanced diet
- **Not** take supplements that are not approved by your retina specialist
- **Not** smoke

Scan here to watch our Ask the Expert video featuring our own Dr. Kenneth Fan.

Scan here to watch a video about what to expect at your first IRD visit.

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