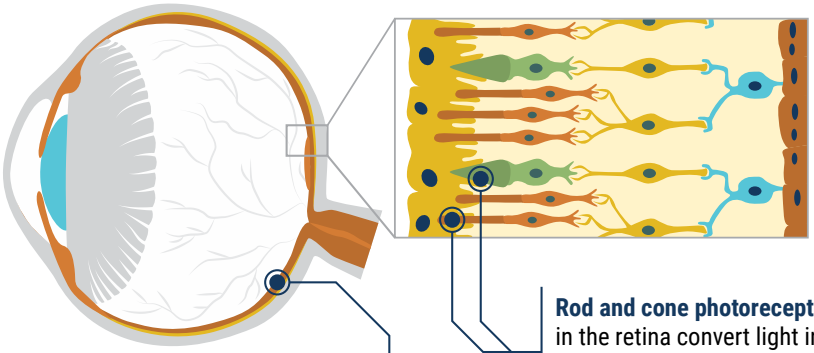
WHAT YOU SHOULD KNOW ABOUT

RETINITIS PIGMENTOSA

WHAT IS RETINITIS PIGMENTOSA?

Retinitis pigmentosa (RP) refers to a group of inherited retinal diseases, often leading to legal and sometimes complete blindness. Forms of RP and related diseases include Usher syndrome, Leber congenital amaurosis, and Bardet-Biedl syndrome, among others. An estimated 100,000 people in the U.S. have RP.



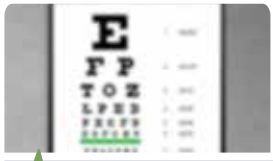
The **retina** is a thin piece of tissue lining the back of the eye.

Rod and cone photoreceptors in the retina convert light into electrical signals that the brain interprets as vision.

WHAT TO EXPECT WITH RETINITIS PIGMENTOSA:

Symptoms depend on whether **rods** or **cones** are initially involved:

Rods: In most forms of RP, rods are affected first, causing **loss of peripheral vision** and **loss of night vision**. Vision becomes more constricted over time.



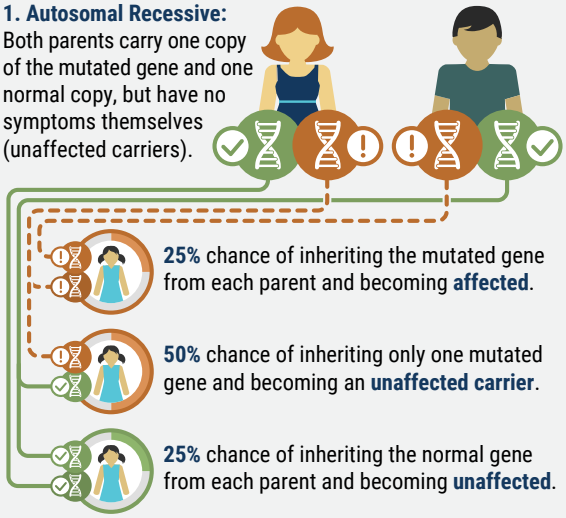
Cones: If and when the disease progresses and cones become affected, **visual acuity**, **color perception**, and **central vision** are diminished.

- Typically diagnosed in children and young adults.
- Rate of progression and degree of visual loss varies.
- Many people with RP are legally blind by age 40.
- Central visual field reduced by varying degrees.

HOW IS RETINITIS PIGMENTOSA INHERITED?

Genetic mutations can be passed from parent to offspring through one of three genetic inheritance patterns:

1. Autosomal Recessive: Both parents carry one copy of the mutated gene and one normal copy, but have no symptoms themselves (unaffected carriers).

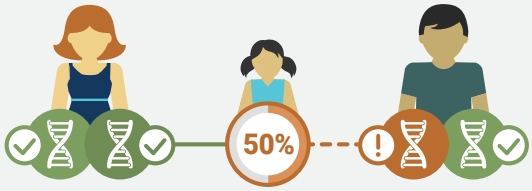


25% chance of inheriting the mutated gene from each parent and becoming **affected**.

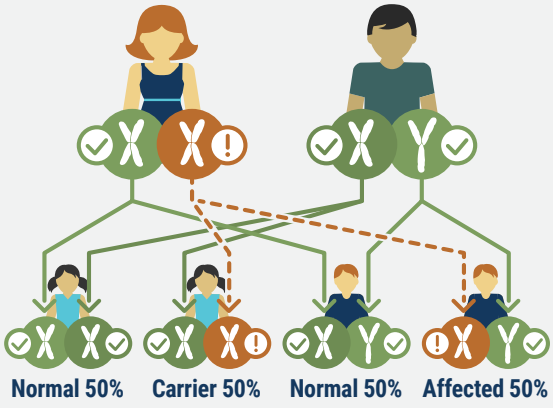
50% chance of inheriting only one mutated gene and becoming an **unaffected carrier**.

25% chance of inheriting the normal gene from each parent and becoming **unaffected**.

2. Autosomal Dominant: Only one parent is affected with one mutated copy of the gene. A child has a **50% chance** of being affected through the inheritance of the mutated gene.



3. X-linked: The mutated gene for the disease is located on the X chromosome. Female carriers have a **50% chance** of passing the disease gene to their daughters, who become carriers, and a **50% chance** of passing the gene to their sons, who are then affected by the disease.



Affected males will **never pass** an X-linked disease to their sons. However, they will always pass their affected chromosomes to their daughters who will become carriers.

FOUNDATION FIGHTING BLINDNESS

To learn more about living with Retinitis Pigmentosa, finding a specialist, genetic testing or clinical trials, visit **FightingBlindness.org** or call **888-332-3667**.