

What is retinitis pigmentosa?

Retinitis pigmentosa, also known as RP, refers to a group of inherited diseases causing retinal degeneration. The retina is a thin piece of tissue lining the back of the eye. It converts light into electrical signals which the brain interprets as vision. People with RP experience a gradual decline in their vision, because photoreceptor cells in the retina degenerate.

Forms of RP and related diseases include Usher syndrome, Leber congenital amaurosis, and Bardet-Biedl syndrome, among others.

What are the symptoms?

Symptoms depend on whether rods or cones are initially involved. In most forms of RP, rods are affected first. Because rods are concentrated in the outer portions of the retina and are triggered by dim light, their degeneration affects peripheral and night vision. When the disease progresses and cones become affected, visual acuity, color perception and central vision are diminished.

Night blindness is one of the earliest and most frequent symptoms of RP. People with mainly cone degeneration,

however, first experience decreased central vision and reduced ability to discriminate colors and perceive details.

RP is typically diagnosed in adolescents and young adults. It is a progressive disorder. The rate of progression and degree of visual loss varies from person to person. Most people with RP are legally blind by age 40, with a central visual field of less than 20 degrees in diameter. It is a genetic disorder and, therefore, is almost always inherited.

How is RP inherited?

An estimated 100,000 people in the U.S. have RP, mainly caused by gene mutations (variations) inherited from one or both parents. Mutated genes give the wrong instructions to photoreceptor cells, telling them to make an incorrect protein, or too little or too much protein. (Cells need the proper amount of particular proteins in order to function properly.) Mutations in dozens of genes have been linked to RP.

Genetic mutations can be passed from parent to offspring through one of three genetic inheritance patterns — autosomal recessive, autosomal dominant, or X-linked.

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In **autosomal recessive RP**, both parents carry one copy of the mutated gene, but have no symptoms themselves. Children have a 25 percent chance of being affected by inheriting a mutated copy from each parent.

In **autosomal dominant RP**, usually one parent is affected and is the only parent with a mutated gene. A child has a 50 percent chance of being affected through the inheritance of the mutated gene from the parent.

In families with **X-linked RP**, the mother carries the mutated gene on an X chromosome, and her sons have a 50 percent chance of inheriting the condition. Daughters have a 50 percent chance of becoming carriers and aren't usually affected. However, some daughters are affected — sometimes mildly, sometimes severely.

If a family member is diagnosed with RP, it is strongly advised that other members of the family also have an eye exam by a physician who is specially trained to detect and treat retinal degenerative disorders. Discussing inheritance patterns and family planning with a genetic counselor can also be useful.

What testing is available?

Genetic testing is available for RP. It

helps assess the risk of passing the disorder from parent to offspring. It also helps with attaining an accurate diagnosis. A patient with an accurate diagnosis is in a better position to keep track of new findings, research developments, and treatment approaches. A genetic diagnosis can also help a person qualify for clinical trials. More information on genetic testing is available at www.FightBlindness.org.

Extensive information on RP research and clinical trials of RP treatments, as well as low vision resources, are available at www.FightBlindness.org

What treatment is available?

While there are limited therapies for RP, the Foundation is supporting several promising avenues of research, including gene, stem cell and drug therapies.

For the latest research advances for RP, refer to the Foundation publication *Retinitis Pigmentosa: Research Advances*.