

# Fact Sheet

## Retinitis Pigmentosa (RP)

Retrieved from: <http://www.lighthouse.org/medical/eye-disorders/retinitis-pigmentosa/>

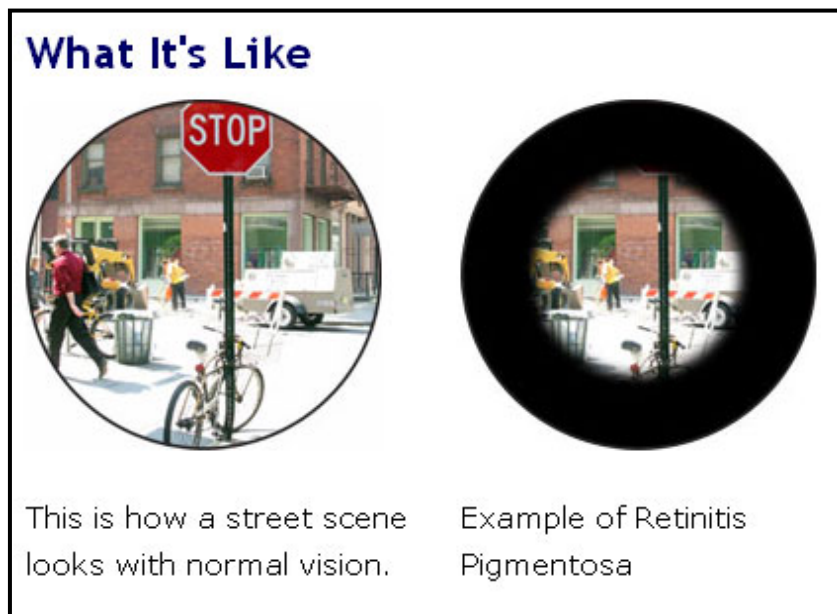
**What is Retinitis Pigmentosa (RP)?** Retinitis Pigmentosa (RP) is the name given to a group of hereditary disease of the retina characterized by progressive loss of visual field, night blindness and reduced or absent electroretinogram (ERG test) recording, which indicates that a large portion of the retina is damaged. RP causes the degeneration of photoreceptor cells in the retina. Photoreceptor cells capture and process light helping us to see. As these cells degenerate and die, individuals experience progressive vision loss.

There are two types of photoreceptor cells: rod cells and cone cells. Rod cells are concentrated along the outer perimeter of the retina. Rod cells help us to see images that come into our peripheral or side vision. They also help us to see in dark and dimly lit environments. Cone cells are concentrated in the macula, the center of the retina, and allow us to see fine visual detail in the center of our vision. Cone cells also allow us to perceive color. Together rods and cones are the cells responsible for converting light into electrical impulses that are transmitted to the brain where "seeing" actually occurs.

**Genetics of RP:** Within the nucleus of every human cell reside a host of genes. Genes are the fundamental building blocks of life. Inherited from our parents, genes carry family traits like eye and hair color, the shape of our face and even diseases like RP. Genes are like computer programs containing sets of coded instructions. Each gene instructs the cell to create a specialized protein that performs a specific task for the cell. In retinal cells, some genes encode proteins that allow the cell to process light. Other genes encode proteins that uptake nutrients and eliminate waste. Still other genes encode proteins that form the cell walls and other structures within the cell.

Sometimes, the coded instructions within a gene become altered. These alterations, known as mutations, can confer a benefit, allowing the organism to better adapt to its environment. However, mutations can also interfere with the proper encoding of a protein. The resulting protein cannot perform its job within the cell, thereby hampering the cell's well being and leading to disease.

RP can be passed to succeeding generations by one of three genetic inheritance patterns -- autosomal dominant, autosomal recessive, or X-linked inheritance. Each type of inheritance causes a different pattern of affected and unaffected family members. For example, in families with autosomal recessive RP, unaffected parents can have both affected and unaffected children. In recessive RP, there is often no prior family history. In families with the autosomal dominant RP, an affected parent can have both affected and unaffected children. In families with the X-linked type, only males are affected, while females carry the genetic trait but do not experience serious vision loss.



It is very important to remember that because RP is an inherited disorder, it can potentially affect another member of the family. If one member of a family is diagnosed with a hereditary retinal degeneration, it is strongly advised that all members of that family contact an ophthalmologist.

**Related Diseases:** Other inherited diseases share some of the clinical symptoms of RP. Some of these conditions are complicated by other symptoms besides loss of vision. The most common of these is Usher syndrome, which causes both hearing and vision loss. There are three types of Usher syndrome. Type One involves congenital deafness, Type Two involves congenital hearing loss, and Type Three involves a progressive loss. The onset of RP tends to be earlier in type one than type two than type three. Other associated syndromes related to retinitis pigmentosa include Bardet-Biedl (Laurence-Moon) syndrome, rod-cone dystrophy, choroideremia, gyrate-atrophy, Leber's congenital amaurosis, and Stargardt's disease.

RP and related diseases are rare and difficult to accurately diagnose. Only a specialist can properly distinguish between the subtle clinical features of these diseases. Therefore, it is important that patients who are symptomatic see an ophthalmologist who specializes in retinal degenerative diseases.

### **Symptoms**

- Normal visual acuity in early stages, possibly -- but not usually -- progressing to no light perception
- Donut shaped visual field loss progressing to severe constriction (loss of peripheral vision)
- Night blindness / Poor light-dark adaptation
- Decreased response to magnification
- Need for more light

The most common feature of all forms of RP is a gradual degeneration of the rods and cones. Most forms of RP first cause the degeneration of rod cells. These forms of RP, sometimes called rod-cone dystrophy, usually begin with night blindness. Night blindness is somewhat like the experience normally sighted individuals encounter when entering a dark movie theatre on a bright, sunny day. However, patients with RP cannot adjust well to dark and dimly lit environments.

As the disease progresses and more rod cells degenerate, individuals most often lose their peripheral vision. People with RP often experience a ring of vision loss in their mid-periphery with small islands of vision in their very far periphery. Others report the sensation of tunnel vision, as though they see the world through the opening of a straw. Many individuals with RP retain a small degree of central vision throughout their life.

Other forms of RP, sometimes called cone-rod dystrophy, first affect central vision. Individuals first experience a loss of central vision that cannot be corrected with glasses or contact lenses. With the loss of cone cells also comes disturbances in color perception. As the disease progresses, rod cells also degenerate causing night blindness and loss of peripheral vision.

Symptoms of RP are most often recognized in children, adolescents and young adults, with progression of the disease continuing throughout the individual's life. The pattern and degree of visual loss are variable.

**Diagnosis:** These special tests can be used to help diagnose RP:

**Acuity Tests:** These tests measure the accuracy of a person's central vision at specific distances in specific lighting situations.

**Color Testing:** These tests can help determine the status of the cone cells, the retinal cells that interpret color.

**Visual Field Testing:** This test uses a machine to measure how much peripheral vision a person has.

**Dark Adaptation Test:** This test will measure how well one's eyes adjust to changes in lighting and can help the doctor better understand the current function of the retina's rod cells, which are the retinal cells responsible for night vision.

**ERG Test:** The ERG (electroretinogram test) records the electrical currents produced by the retina due to a light stimulus. The intensity and speed of the electrical signal becomes reduced as the photoreceptor cells degenerate.

**Risk Factors Associated with RP:** Recent research findings suggest that in some forms of RP, prolonged, unprotected exposure to sunlight may accelerate vision loss. RP is an inherited, genetic disease. It is caused by mutations in genes that are active in retinal cells. Gene mutations are programmed into a person's cells at the time of conception. RP is not caused by injury, infection or exposure to any toxic substance.

**What Can Be Done to Reduce Risk:** Reducing exposure to sunlight is important for keeping the eye protected. However, since RP is an inherited disorder and it runs in families, the disease is not preventable. If someone in your family is diagnosed with a retinal degeneration, it is strongly advised that all members of the family contact an eye care professional.

**Treatment:** As yet, there is no known cure for RP. However, intensive research is currently under way to discover the cause, prevention, and treatment of RP. Researchers have identified some of the genes that cause RP. It is now possible, in some families with X-linked RP or autosomal dominant RP, to perform a test on genetic material from blood and other cells to determine if members of an affected family have one of several RP genes. Source: The Foundation Fighting Blindness (FFB)

[www.blindness.org](http://www.blindness.org)

## **Resources**

**RP International** PO Box 900, Woodland Hills, CA 91365 Phone: (818) 992-0500 Fax (818) 992-3265 E-mail: [info@rpinternational.org](mailto:info@rpinternational.org)

**The Foundation Fighting Blindness** 11435 Cronhill Drive, Owings Mills, MD 21117-2220 Phone: (800) 683-5551 (TDD) Fax: (410) 363-2393 Retinitis Pigmentosa

**National Retinitis Pigmentosa Foundation, Inc.** 11350 McCormick Rd., #800, Hunt Valley, MD 21031-1002 Phone: 800/638-5683 TTY

**For more information about the CO Services for Children and Youth with Combined Vision and Hearing Loss Project and the project's services specific to Colorado children who are deafblind, please contact:**

Tanni Anthony  
Phone: 303-866-6681  
Email: [anthony\\_t@cde.state.co.us](mailto:anthony_t@cde.state.co.us)

Gina Quintana  
Phone: 303-866-6605  
Email: [quintana\\_g@cde.state.co.us](mailto:quintana_g@cde.state.co.us)

Colorado Department of Education Fax: 303-866-6767  
Exceptional Student Leadership Unit Web Page Address:  
1560 Broadway, Suite 1175 <http://www.cde.state.co.us/cdesped/Deafblind.asp>  
Denver, CO 80202

*Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Updated: 10/10*