



British and Éire Association of Medical Retina Specialists

Retinitis Pigmentosa (RP) is a group of rare, inherited eye diseases that cause progressive degeneration of the retina, the light-sensitive layer of tissue at the back of the eye. The retina plays a critical role in capturing light and sending visual signals to the brain. RP affects the photoreceptor cells (rods and cones) in the retina, leading to gradual vision loss over time.

Types of Retinitis Pigmentosa

There are several types of RP, and they are classified based on the inheritance pattern:

1. Autosomal Dominant RP:

- The most common form of RP, requiring just one affected parent to pass on the condition. If one parent carries the mutated gene, there is a 50% chance of passing it on to each child.

2. Autosomal Recessive RP:

- Requires both parents to pass on a defective gene. The child must inherit two copies of the mutated gene (one from each parent) to develop the disease. This form tends to be rarer than the dominant form.

3. X-Linked RP:

- This form is passed on through the X chromosome. It is more common in males, as they only have one X chromosome (females have two, so they are usually carriers). A male child with an affected mother has a 50% chance of inheriting the condition.

Symptoms of Retinitis Pigmentosa

The symptoms of RP can vary greatly depending on the specific type and age of onset, but the condition typically causes a slow, progressive decline in vision. Common symptoms include:

1. Night Blindness (Nyctalopia):

- Difficulty seeing in low light or darkness is often one of the first signs of RP. People with RP may find it hard to navigate in dimly lit areas or during the evening.

2. Loss of Peripheral Vision (Tunnel Vision):

- RP often begins with the gradual loss of side (peripheral) vision. Over time, this can progress to tunnel vision, where the person can only see what is directly in front of them, with a narrowed field of view.

3. Decreased Central Vision:

- In the later stages of RP, central vision can also become affected, leading to blurred or lost vision in the middle of the field of view.

4. Colour Vision Impairment:

- Some individuals with RP may notice difficulty distinguishing certain colors, particularly in low-light conditions.

5. Progressive Vision Loss:

- Vision loss generally progresses slowly over many years or decades, but the speed and extent of vision loss can vary depending on the specific type of RP and the individual.

Causes and Genetic Factors

RP is caused by mutations in genes that affect the photoreceptor cells in the retina. These genes can encode proteins that are crucial for the function of rods and cones, leading to their degeneration over time. Some common genes involved in RP include RHO, USH2A, PDE6A, and RPGR, among others.

Because RP is inherited, family history plays a significant role in determining whether a person will develop the condition. Genetic testing can help identify the specific mutation responsible for the disease in individuals and their families.

Diagnosis of Retinitis Pigmentosa

Diagnosing RP typically involves several tests, including:

1. Comprehensive Eye Exam:

- The doctor will check the retina for signs of damage, such as retinal bone spicule pigmentation (dark pigment deposits in the retina), which is characteristic of RP.

2. Electroretinogram (ERG):

- This test measures the electrical responses of the retina to light stimuli. In RP, the electrical responses may be diminished or absent, indicating dysfunction of the photoreceptor cells.

3. Visual Field Test:

- A visual field test can detect areas of vision loss. People with RP typically experience a loss of peripheral vision, leading to the "tunnel vision" effect.

4. Fundus Photography:

- A special camera is used to take pictures of the retina to monitor changes or damage to the retina over time.

5. Optical Coherence Tomography (OCT):

- This imaging test provides cross-sectional images of the retina to detect thinning or damage to retinal layers.

6. Genetic Testing:

- In some cases, genetic testing may be used to identify the specific mutations responsible for RP, which can help confirm the diagnosis and determine the inheritance pattern.

Treatment Options for Retinitis Pigmentosa

Currently, there is no cure for RP, but several approaches can help manage symptoms or slow the progression of the disease:

1. Vitamin A Supplements:

- Studies have shown that high-dose vitamin A palmitate may help slow the progression of RP in some individuals, particularly in the early stages. However, this treatment is not suitable for everyone and should only be taken under the guidance of a healthcare provider, as high doses of vitamin A can be toxic.

2. Low Vision Aids:

- As vision declines, individuals with RP can benefit from devices designed to enhance remaining vision, such as magnifiers, specialized glasses, and screen readers.

3. Gene Therapy:

- Gene therapy research for RP is ongoing, and there have been some promising developments. For example, in recent years, Luxturna (voretigene neparvovec), a gene therapy for a specific type of RP caused by mutations in the RPE65 gene, has been approved by the FDA. It works by delivering a normal copy of the gene to retinal cells, improving vision in affected individuals.

4. Retinal Implants:

- Retinal implants or prosthetic devices (such as the Argus II Retinal Prosthesis System) are being explored as a way to restore some degree of vision in people with advanced RP. These implants work by bypassing damaged photoreceptor cells and stimulating the retina with electrical signals.

5. Stem Cell Therapy:

- Research into using stem cells to regenerate damaged retinal tissue is ongoing. Although still in experimental stages, stem cell therapies hold promise for potentially treating or reversing some forms of RP in the future.

6. Protection from Light:

- People with RP may be more sensitive to light, so wearing sunglasses or visors with UV protection may help reduce discomfort and further damage to the retina.

Coping with Retinitis Pigmentosa

Living with RP can be challenging due to the gradual loss of vision, but many people with the condition learn to adapt. Some strategies include:

- Adapting to changes: Using adaptive technologies, like screen readers, large print, and talking devices, can make everyday tasks easier.
- Support networks: Connecting with support groups or other people living with RP can be beneficial. Many organizations, such as the Foundation Fighting Blindness, offer resources and support for those with RP.
- Counselling: It's not uncommon for individuals with RP to experience emotional stress related to vision loss. Speaking with a counsellor or therapist can be helpful in managing these challenges.

Prognosis

The rate of progression of RP varies significantly between individuals, even within families. In general, RP tends to progress more slowly in some people, while others may experience more rapid vision loss. Most people with RP retain some level of vision throughout their lives, but the severity of the condition can differ.

While RP often leads to significant vision impairment or even blindness in later stages, there are many technological advancements and support systems available to help people adapt and maintain quality of life.

Conclusion

Retinitis Pigmentosa is a progressive genetic condition that affects the retina and leads to vision loss over time. While there is no cure, treatments such as vitamin A supplementation, low vision aids, and emerging therapies like gene therapy and retinal implants offer hope for slowing progression and improving quality of life. Early diagnosis and regular monitoring are crucial for managing the condition and maintaining vision for as long as possible.