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## Eyes - retinitis pigmentosa

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### Summary

- Retinitis pigmentosa (RP) is the collective name for a range of diseases that damage the light sensitive cells of the retina and cause vision to fade.
  - There is currently no cure, and no treatments are available to slow the progression of disease.
  - Symptoms include night blindness and tunnel vision.
  - It is often difficult to accurately predict how RP will progress with each person.
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The retina is located at the back of the eye. This thin layer of light-sensitive cells sends information on shape, colour, pattern and movement to the brain via the optic nerve. There are special light sensing cells in the retina, called rods and cones (see explanation below).

Retinitis pigmentosa (RP) is the collective name for a range of genetic (hereditary) diseases that damage the retinal rod and/or cone cells and cause vision to fade. According to some estimates, about one in every 3,000 Australians is affected by RP.

The severity of symptoms, speed of progression and age of onset depend on individual factors such as the type of RP. There is no cure and no treatments are available to slow the progression of disease.

### Rods and cones explained

The retina contains millions of cells called rods and cones. They are named this because of their appearance. Rods, found in greater numbers than cones, are located across the entire retinal surface.

There is a higher concentration of rods around the periphery (edges) of the retina, which allows you to see what is above, below and to the sides of the object you are directly viewing. One of the key functions of rods is to detect low light levels.

There are far fewer cones than rods. Cones are located mainly in the centre of the retina, and help to distinguish colours and finer details.

### Symptoms of retinitis pigmentosa (RP)

Since retinitis pigmentosa is a progressive condition, the symptoms and signs worsen as time goes by. Early symptoms can include:

- poor night vision
- problems seeing things in dimly lit environments
- loss of peripheral (side) vision
- difficulty in judging changes in peripheral vision, such as curbs or steps.

### Tunnel vision

One of the first symptoms of RP is night blindness, followed by the loss of peripheral vision. Typically, light-sensitive rod cells in the peripheral retina are lost first. As more cells die, the person loses more of their peripheral vision until only the very centre of their visual field remains. This is known as tunnel vision.

In order to see what is above, below or to the sides of the focused object, the person with RP has to move their eyes. In other cases of RP, the central area of the retina is affected first, which means the person may have trouble with:

- distinguishing colours
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- reading
- recognising visual details.

## Cataracts

The lens is located behind the iris (coloured part of the eye) and pupil. This transparent disc allows the eye to finely focus. A cataract means the lens has become cloudy or opaque, which can interfere with vision.

The development of cataracts is a common complication of RP. However, it does not necessarily have great visual significance, as in most cases the retinal disease is the most important reason for the poor vision. In such cases removing the cataract by surgery does not always result in a great improvement in vision.

## Types of retinitis pigmentosa

In most cases, RP is caused by any one of more than 100 defective genes. It is thought that these flawed genes cause the formation of a wrong type of protein in the retina, which results in the death of rods and cones.

The three patterns of inheritance linked to RP include:

- **Autosomal recessive** – both parents are healthy but carry the gene, so each of their children has a 50 per cent chance of inheriting one gene and being a carrier, and a 25 per cent chance of inheriting both genes and developing RP.
- **Autosomal recessive** – this inheritance pattern is the most common type of RP. The chance of having this condition is higher if the parents are related (for example, cousins).
- **Autosomal dominant** – in this form of RP, only one parent has the gene, and is usually affected by the disease as well. Each child has a 50 per cent chance of inheriting this gene and developing RP.
- **X-linked recessive** – this is a rarer type of RP. The defective gene is carried by the (usually healthy) mother and is passed to sons only via the X chromosome. Each son has a 50 per cent chance of developing RP. Each affected male patient will transmit the gene to the next generation, but only to his daughters, who will be healthy carriers.

## Diagnosis of retinitis pigmentosa

A range of eye tests diagnoses RP. For example, the retina looks like it is covered in deposits of black pigment (which give the disease its name) when viewed through an instrument known as an ophthalmoscope.

In most cases, a confirmation of the diagnosis is given by an electro-physiological test called electro-retinogram. This test records the electric activity of the retina in a similar way to an ECG test.

It involves exposing the person to dark and light conditions and measuring the retinal electric activity via electrodes placed on the person's head or eyes. The test is painless and is sometimes repeated to monitor the disease progression.

Other tests that may be required include:

- visual field test
- colour vision testing and, sometimes
- fluorescein angiogram a special type of photography of the retina that is done after injection of dye into the arm vein.

## Treatment for retinitis pigmentosa

Unfortunately, there are no known treatments to prevent or slow the progression of RP. However, researchers around the world are constantly working on development of treatments for this common blinding condition.

## Research into retinitis pigmentosa

Although some researchers have tried to treat the disease with vitamins and certain fatty acids, the results of such studies have not shown significant effects of this treatment on a person's vision. Their role, if any, in the management of retinitis pigmentosa remains controversial.

## Gene therapy research

Researchers are continuously trying to develop treatments for RP. Since the reason for the disease is usually a faulty gene, gene therapy (introducing a healthy gene into the retina) is one of the explored areas. However, there is currently no available gene therapy for RP and it is still only a subject for intensive research.

### **Transplant research**

Attempts at transplanting healthy retinal cells into sick retinas are being made experimentally. This research has not reached a clinically-useful degree of success.

### **Retinal prosthesis**

Another type of treatment that is being explored is an implantable light-sensitive electrode (retinal prosthesis). This prosthesis would be introduced into the eye and function as a "bionic retina". This is still under development and so far has not been able to produce useful vision in blind patients.

### **Management of retinitis pigmentosa**

Suggestions include:

- Seek advice from professional organisations.
- Joining support groups may be helpful.
- Use low vision aids, such as magnifiers and lamps.
- Rearrange the furniture to reduce the risk of stumbling or bumping into things.
- Get into the habit of closing cupboard doors.
- Don't leave doors half-open - either close them or fully open them.
- Tell your family about ways in which they can help you. For example, when you are out of the house, you need warning about low-hanging tree branches or steps.
- Genetic counselling is available.

### **Progression of retinitis pigmentosa**

It is often impossible to accurately predict how RP will progress with each person. Points to remember include:

- Different people with RP may have totally different disease courses because of genetic and other differences.
- Even within the same family, there may be variability in the rate of progression of the disease between different members.
- Many people live a full, active life with RP for many years, although modifications in the workplace or job often need to be made due to progressive visual loss.

### **Usher syndrome**

Usher syndrome is a genetic disorder that includes retinitis pigmentosa and a partial or total hearing loss from birth. Usher syndrome is categorised into three broad groups according to the type and severity of symptoms. Type 1 and Type 2 account for around 10 per cent of children who are born deaf.

### **Where to get help**

- Your doctor
- Eye specialist
- **Retina Australia** (Vic) Tel. (03) 9650 5088
- **The Royal Victorian Eye and Ear Hospital** Tel. (03) 9929 8666

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