Retinitis Pigmentosa

Fact sheet

What is retinitis pigmentosa?
Retinitis pigmentosa (RP) is the name given to a group of hereditary eye disorders. These disorders affect the retina, which is the light-sensitive tissue lining the back of the eye, in which the first stages of seeing take place. In RP, sight loss is gradual but progressive. It is unusual for people with RP to become totally blind as most retain some useful vision well into old age.

What is the retina?
The retina in your eye serves a similar purpose to a film inside a camera. Light is focused by a lens at the front of the camera onto a light-sensitive film at the back, to form a picture. In a similar way, light entering your eye is focused onto a light-sensitive tissue that lines the inside of the eye at the back. This tissue is the retina.

The retina consists of two main layers, a thin one called the pigment epithelium and a thicker one, made up of many layers of cells, called the neural retina. One particular layer in this neural retina contains many millions of cells called photoreceptors, which are able to respond to light. A few million cells called cones are concentrated in the central portion of the retina. These allow us to see fine detail and colour. Away from the central portion of the retina are about 120 million cells, which are mostly rod cells. They enable us to see when light is dim and provide peripheral vision outside of the main line of sight.

When light is focused onto these rods or cones, a small electrical charge is generated (the amount depends on the amount of light) and this charge passes down the optic nerve to the brain. As each of these cells receives a slightly different amount of light and sends a different electrical pulse, the brain is able to assemble a picture.

What causes RP?
It is now known that there are many different inherited defects causing RP. In all RP-related conditions however, the ability of the retina to respond to light is affected. The problem can be in many parts of the retina such as the rod or cone cells, or in the connections between the cells of the retina.

What are the symptoms of RP?
The most common first symptom is difficulty in seeing in poor light, for example outdoors at dusk, or in a dimly lit room. A second symptom is reduction of the visual field, in which sight is lost from the sides, or from above and below. This is often referred to as tunnel vision and means that

Retinitis Pigmentosa

Fact sheet

the rod cells, and some of the outer cone cells, have been affected first.

In some RP-related conditions central vision is lost first. The first signs of this are difficulty in reading print or carrying out detailed work. All RP conditions are progressive, but the speed at which deterioration takes place varies from one person to another.

In many types of RP, glare from bright lights is an increasing problem, although some people do not experience this until the more advanced stages.

How is RP inherited?
RP can be inherited in three ways:

- Autosomal dominant inheritance. This is the pattern of inheritance where RP is known to exist in a family, affecting both males and females. The probability of RP being passed from an affected parent to a child is exactly 50 per cent.

- Autosomal recessive inheritance. There will usually be no known history of RP in the family but if two carriers who show no obvious symptoms have a child, there is a 25 per cent chance that he or she will have RP.

- X-linked inheritance. This is a pattern of inheritance where only males develop the disease, but female members of a family are carriers. Some carriers can develop a mild form of RP. For example, if a man has X-linked RP, his sons will not develop RP, but all of his daughters will become carriers. These daughters will each have a 50 per cent chance of producing an affected son and a 50 per cent chance of daughters who will be carriers. This inheritance pattern is sometimes difficult to identify in a family where there have been no sons for several generations, as the faulty gene could have passed down a line of female carriers and then suddenly affect a male child.

When does RP develop?
There is no hard and fast rule, but in most cases the early symptoms of RP develop between the ages of 10 and 30.

Can it be detected by an eye test?

Retinitis Pigmentosa

Fact sheet

The types of RP which cause loss of central vision become apparent through an inability to read a test card, but peripheral loss, or loss of side vision, is not so obvious and a person may be able to read an optician's test card for many years.

The condition is best detected by an examination of the inside of the eye by a doctor, using an ophthalmoscope. Normally he or she would see an orange-coloured area called the fundus. When the patient has RP, the orange surface is broken by black or brown clumps of pigment.

Other tests are available which measure the area of visual field, which is still useable, and the ability to adapt to low light levels. Many more sophisticated tests are available at eye hospitals and these are explained in detail in the BRPS booklet, 'The Eye Clinic and RP'.

Do people with RP develop cataracts?
Yes, many people with RP do. Cataracts are a clouding of the lens at the front of the eye. They usually occur around middle age in people with RP. When they have reached a certain stage, an eye specialist will usually recommend their removal. The lens is then either replaced with an implant, or specially made spectacles are prescribed.

After the operation, the patient will still have RP, but if the retina has not deteriorated too far, a limited amount of vision will be restored. This is the most common origin of the 'miracle cure' stories that appear from time to time in the press.

Is RP just an eye condition?
Yes, but there are other conditions which people with RP can inherit. One example of this is Usher syndrome, where people develop the dual disability of hearing loss and retinitis pigmentosa. For more information on Usher syndrome, contact Sense on 0207-272 7774.

Can RP be treated?
Currently, no treatment is available to cure RP or arrest its progress. It is the result of incorrect instructions being passed to the body's chemistry by faulty inherited genes.

However, co-ordinated research, in many centres around the world, has been expanding for around 20 years. Many of the genes responsible for the numerous types of RP have been located and their defects identified.

Retinitis Pigmentosa

Scientists are following many lines of research, among which is the development of a safe system for introducing corrective genetic material to the appropriate cells of an eye. There is some evidence that this will take hold and assist cell regrowth.

Factsheet