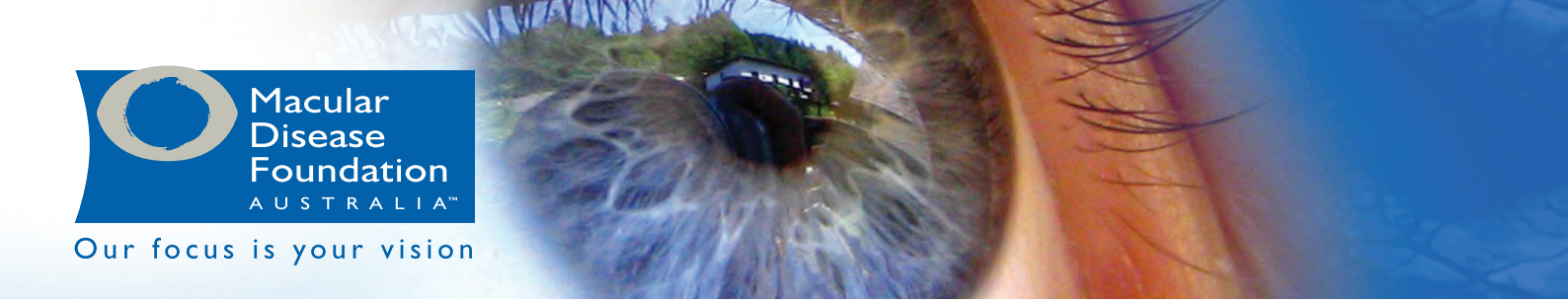

Macular Disease Foundation AUSTRALIA™

Our focus is your vision

Retinitis pigmentosa

Retinitis pigmentosa (RP) is the name given to a diverse group of inherited eye disorders which affect a part of the eye called the retina. RP causes permanent changes to your vision but how quickly this happens and how it changes differs between people. These changes may include difficulty with vision in dim light or the dark and the loss of side or peripheral vision.

If you have RP, sight loss is gradual but progresses over many years. Some people with RP might become blind but most people with RP keep some useful vision well into old age.

How the eye works

Light passes through the cornea at the front of your eye, and is focused by the lens onto your retina. The retina is a delicate tissue that lines the inside of your eye. The retina converts the light into electrical signals that travel along the optic nerve to your brain. The brain interprets these signals to “see” the world around you.

Light from the object you are looking at directly is focused onto a tiny area of the retina called the macula at the back of the eye. The macula is about 4mm across and is responsible for detailed central vision and most colour vision. It contains a few million specialised photoreceptors called cone cells. These cone cells work best in bright light and allow you to see fine detail for activities such as reading, recognising faces, driving, writing and to recognise colours.

The rest of the retina, called the peripheral retina, is mostly made up of the other type of photoreceptor called rod cells. Rod cells enable us to see when light is dim and provide peripheral vision. Peripheral vision is what you can see to the sides and above and below when you are looking at something straight ahead.

Causes of retinitis pigmentosa

All types of RP affect the retina. The retinal cells gradually stop working and eventually die. In most cases, the peripheral rod cells are affected first while the central cone cells tend to be affected later. The symptoms you experience depend on the way your retina is affected by RP and can be very different from person to person.

Almost all types of RP are inherited, caused by a fault in the genetic information passed down from a parent. The genes we inherit contain the instructions that tell our body how to grow, repair and renew. When a gene is faulty these instructions are faulty and the cells using those instructions do not work as they should. In RP, the faulty

genes cause the retinal cells to stop working and eventually die. Researchers have found many of the genes which, when faulty, cause RP but there is still work to be done to discover them all.

As there are many genes that can cause the retinal cells to stop working, there are many different types of RP. This is why RP is described as a group of inherited retinal disorders. RP is often mentioned alongside other eye conditions with similar genetic causes and effects on vision, such as Leber's congential amaurosis, cone and cone-rod dystrophies, and choroideremia.

RP can also be associated with other problems such as hearing loss. These rare conditions are referred to as RP syndromes.

RP syndromes

In most cases, the inherited gene defect only affects the eyes, however, sometimes other parts of the body are also affected. One example of this is Usher syndrome, where people develop loss of both hearing and sight. Others include Refsum, Alström, and Laurence-Moon-Bardet-Biedl (LMBB) syndromes.

How is RP inherited?

RP often runs in families and is often classified by the way it is passed from generation to generation. The type of inheritance tells us who in the family has had the condition in the past, the likely severity of the RP when it occurs and the chances of children being affected in the future. RP can be inherited in three different ways:

1. Autosomal dominant inheritance

Autosomal dominant RP affects men and women equally and there tends to be a known history of the condition in the family. This form of RP is less severe than the other two listed below and the first signs of it tend to appear at around 30 years of age.

2. Autosomal recessive inheritance

Autosomal recessive RP also affects men and women equally but there may be little or no known history of the condition in either family in the past. This form of RP tends to show first signs between 30 and 40 years and tends to cause more severe sight loss.

3. X-linked inheritance

This is a pattern of inheritance that affects mostly men. Female members of a family are carriers of the faulty gene but rarely develop the full condition, although some carriers can develop a mild form of RP. If there have been no boys in the family in the last few generations then there may be no history of the condition. This type of RP affects vision severely and can result in very poor vision by the age of 40.

No known relative

In about half of diagnosed cases of RP there does not seem to be any previously affected relatives. Relatives will have passed on the faulty genetic information but may have not developed symptoms themselves. In such cases it may not be possible to determine which of the three types of inheritance have caused the RP.

Genetic counselling

Genetic counselling aims to help you understand the type of RP you have, how it may affect you in the long term and the risks of passing on the condition to any children you may have. A genetic counsellor asks about your family tree in detail to try and understand how RP has been inherited through the generations. It may be provided by a specialist RP eye clinic or a medical genetics department. You can ask your GP or ophthalmologist to refer you to your local genetic counselling service.

Genetic testing

You may also be offered genetic tests to try and work out which genes are faulty. Testing for RP is complex and is not useful or possible yet for all types of RP. Ask your ophthalmologist or genetic counsellor to discuss testing with you and provide a referral.

You should also consider being added to the Australian Inherited Retinal Disease Register, maintained at Sir Charles Gairdner Hospital in Perth:

P: (08) 9346 2866 or
W: www.scgh.health.wa.gov.au/Research/InheritedRetinal.html

Early symptoms of retinitis pigmentosa

In most of the more common forms of RP, the first symptoms occur between childhood and the age of 30. You will usually initially notice that it is difficult to see in poor light, such as outdoors at dusk, or in a dimly lit room. This is often referred to as “night blindness”. While most people find it takes their eyes up to 20 minutes to adapt to dim light, if you have RP it will either take much longer or it won’t happen at all.

A second symptom is the loss of some of your peripheral vision or peripheral visual field. This means that when you’re looking straight ahead you become less able to see things either to the side, above or below. Difficulty seeing in low light and loss of peripheral vision are signs that the peripheral rod cells are being affected by RP.

For some people, the early loss of peripheral vision may means it is no longer safe to drive. You are required by law to report a permanent or long-term vision condition which might affect your ability to drive. Your doctor will provide you with advice about your ability to drive safely as well as a letter or report to take to your relevant state motor vehicle authority.

In some RP-related conditions, central vision is lost first because the central cone cells are affected first. You might find it difficult reading print or carrying out detailed work at this time. In these types of RP, peripheral vision is affected in the later stages.

Later symptoms of retinitis pigmentosa

Retinitis pigmentosa is a progressive condition, but the speed and pattern of deterioration of sight varies from one person to another. For most people, the first effect of RP is the gradual loss of peripheral vision. This means that you can start to miss things slightly to the side of you or trip over or bump into things you would have seen in the past. Most people with RP eventually have a very restricted visual field, leaving only a narrow tunnel of vision.

Most people with RP retain useful central vision through their twenties, which means the ability to read and recognise faces is not greatly affected. By 50 years of age most people's central vision is affected to the extent that reading is a problem without the help of a magnifier.

Many people with RP find the glare from bright lights and sunlight starts becoming a problem. The retinal cells become less able to adapt to changing light levels and it becomes more difficult to use your vision when you move between a light and a dark room.

Eye tests - initial

Most people first experience problems in low light levels and this may prompt them to see their optometrist or GP. Because the onset of the various symptoms vary from person to person, some people may have their condition diagnosed at an early stage while other people's RP may go undetected for many years.

An optometrist can examine your retina to detect RP. Normally, they would see the orange red of the healthy retina and the blood vessels that supply it. When someone has RP, the shape of the blood vessels is affected and the orange surface is interrupted by tiny clumps of black or brown pigment.

The types of RP which cause loss of central vision tend to be detected by a routine eye test at an early stage. Loss of central vision makes the letter chart harder to see. The more common symptom of peripheral field loss, or loss of side vision, is not so obvious and this can only be detected by a visual field test. Most optometrists can carry out this test but may not do so routinely. If you have any concerns about your peripheral vision then you should ask your optometrist for a visual field test.

If you have a family history of RP or you have had problems with your vision in the dark, or when moving from light to dark, you need to make this clear to the person testing your eyes. This will help them to devise the most appropriate set of tests for you. If after an eye test there is cause for concern the optometrist can refer you to an ophthalmologist for more testing.

Eye tests - further investigation

If you have been referred to the ophthalmologist, a set of tests can be done to diagnose RP. The testing process varies from person to person and may take more than one visit. The ophthalmologist may be able to say that you have RP after the first few sets of tests but it is often not possible in the early stages of the condition to define exactly what form of RP you have or what the likely long term effects will be on your vision.

It is important to ask your ophthalmologist to talk you through the tests and the results at each stage. None of the tests are painful but they can take a long time and be repetitive. You may be asked to have some or all of the following tests:

Examination of the back of the eye

An examination of the retina is always carried out. You will be given eye drops to dilate (enlarge) your pupils to allow the ophthalmologist to see the back of your eye clearly. The dilating drops take about 30 minutes to work. They will make you sensitive to light and cause your vision to be blurry. The effects of the drops usually wear off in about six hours though sometimes it can take overnight. It is not safe to drive until the effects have worn off.

Retinal photographs and fluorescein angiograms

You may have photographs taken of your retina using a special camera. This photograph of the retina can be used for comparison during future visits as an additional way of tracking the progress of the RP. Your ophthalmologist may also ask for a more specialised set of photographs to be taken using fluorescein dye. The yellow fluorescein dye is injected into a vein in your arm. It travels into the tiny blood vessels in your retina, and a series of photographs are taken. The dye in the blood vessels shows up changes in your retina that are not visible using normal photography. The fluorescein dye can make your skin look yellow for up to 24 hours. The dye is passed through the urine, which will be a deep yellow colour for about 24 hours.

Visual field test

A visual field test checks whether your peripheral vision has been affected. You look straight ahead at a particular point in a bowl-shaped screen in a darkened room. Each time you spot dots of light you click a button. The test takes about 10 minutes for each eye and tests what you notice to the sides, above and below when you are looking straight ahead.

Colour vision test

Your colour vision may be tested. You will be asked to look at a booklet that shows numbers composed of different coloured dots. The numbers are printed within different coloured dotty backgrounds. This quick and straight forward test shows which colours you are able to distinguish from each other.

Electro-diagnostic tests

Electro-diagnostic tests may be needed to investigate how your retina is working. The electrical activity of the retina is measured under different lighting conditions and this then identifies layers of the retina that are not working properly.

These tests include the Electroretinogram (ERG), Electro-oculogram (EOG) and the Pattern Electroretinogram (PERG). Each test records electrical activity producing a trace or plot. When these plots are compared to the plot from a retina without RP your eye specialist can see which layers of your retina have been affected.

Each test has a specific procedure and you should ask the staff to explain exactly what will happen before you start. The tests are painless but may involve having your eyes dilated and/or numbed, a tiny electrode being placed on your eye and a sensor on your skin.

Other ocular complications of retinitis pigmentosa

Some people with RP also develop cataracts. Cataracts are a clouding of the natural lens of the eye, which is located just behind the iris (coloured part of the eye). They usually occur around middle age in people with RP. An eye specialist may recommend that you have the cataract removed, particularly if the cataract is interfering with your remaining useful vision. The lens affected by the cataract is removed and is either replaced with an implanted artificial lens and/or spectacles are prescribed to focus your vision correctly. After a cataract operation, you still have RP but, if the retina has not deteriorated too far, a limited amount of vision will be restored.

Some people with RP develop macular edema. This is when the blood vessels near the macula leak and make the retina swell. This blurs and distorts the central vision. Macular edema can occasionally occur after cataract surgery and very rarely it can happen spontaneously.

Research into retinitis pigmentosa

Currently, there is no known cure or treatment for RP or associated retinal disorders. Many research groups around the world are working on different aspects of the condition with the aim of developing treatments. Many of the genes causing RP and related conditions are being discovered (or mapped) and it is this understanding of where the faults occur in the genetic information that may enable potential treatments to be devised.

It is possible that eventually one or more treatments may be devised which will combine the knowledge gained from some of the following avenues of research. Most of the work so far has only been carried out in the laboratory.

Gene therapy

Once a faulty gene causing RP has been identified, gene therapy aims to replace the faulty gene within the affected retinal cells with new genes that work properly. The new genetic material, usually carried by a harmless virus, is injected directly into the affected area of the retina. The hope is that the cells then begin to work correctly and the damage is either stopped or reversed. This method relies on the gene causing the problem being known but in many cases of RP the faulty gene or genes are yet to be discovered.

Stem cell therapy

The body contains many different types of cells, and some are more specialised than others. The retinal cells affected by RP are very specialised cells that the body cannot easily replace. Stem cells are cells that can divide (differentiate) into other cell types and they have the potential to replace damaged or missing retinal cells. The aim of stem cell research is to see if stem cells can be persuaded to differentiate into retinal cells which can then be injected into the retina, replacing the damaged cells.

Growth factors

Growth factors are chemicals that support cells to grow and repair themselves. Research groups are working on the potential uses of growth factors in the treatment of retinal disease in the hope that damaged cells can be repaired or protected from damage.

Retinal implants

For people who have lost all vision from RP, new retinal implants (so-called ‘bionic eyes’) are now available in the USA and in parts of Europe. These can provide very basic vision to help identify basic shapes and movement. They do not currently allow reading. More information is at: www.2-sight.eu/en/

Nutrition

It has been suggested by some research that vitamin A may have a beneficial effect for people with RP. This research has been questioned and the positive effects observed were very slight. This type of treatment is not currently being prescribed by most ophthalmologists. Taking vitamin A can have other issues and should be discussed with your GP and ophthalmologist. Other studies are investigating the benefits of mixtures of nutritional supplements which have an anti-oxidant effect.

Refsum syndrome is one of these rare situations where RP is known to be affected by nutrition. Strictly adhering to a diet that excludes or is low in phytanic acid is beneficial in Refsum syndrome. Phytanic acid is in dairy products, beef and lamb, and fatty fish such as salmon, mackerel, sardines, and cod. Ask to be referred to a dietitian if it is recommended that you follow a restricted diet, so that you can be sure to get the nutrients you need.

Research updates

Because research and new theories change quickly, stories about potential cures or treatments for sight loss often appear in the newspapers, on television and on the internet. Such stories are often over-simplified and occasionally misleading. Regular authoritative updates on current research are available on the RP Fighting Blindness website (www.rpfightingblindness.org.uk).

Managing vision loss

When managing vision loss, a key priority is maintaining quality of life and independence. Contacting a low vision organisation can be helpful as they can work with you to assess your individual needs and determine which aids and technologies can help. There are many excellent solutions to help you live well with low vision.

Macular Disease Foundation Australia Resources

Macular Disease Foundation Australia has developed a comprehensive range of publications on macular degeneration, diabetic eye disease and other macular diseases. Information and advice on living well with vision loss is also available. Call the Foundation for a free information kit or to register to receive newsletters and invitations to attend education sessions and events.

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