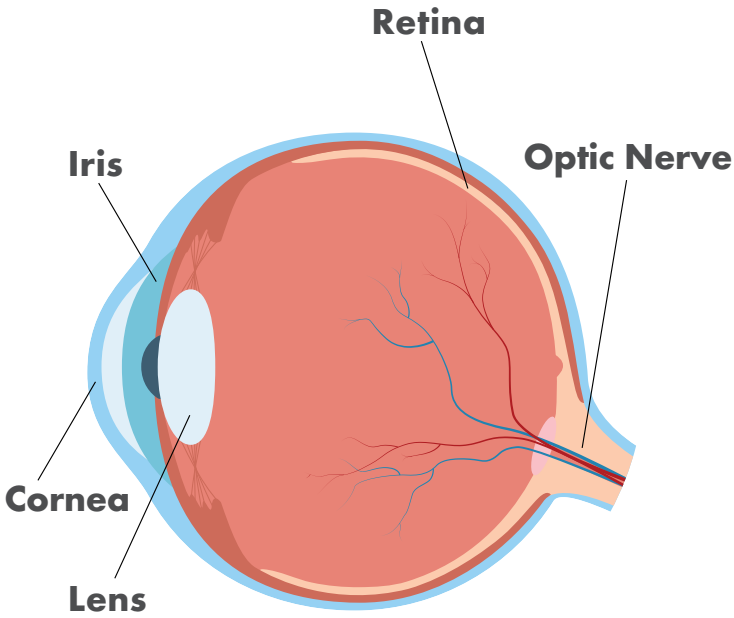
RETINITIS PIGMENTOSA

Macular Disease Foundation AUSTRALIA

Retinitis pigmentosa (RP) is the name given to a diverse group of inherited eye disorders that affect the retina at the back of the eye. RP causes permanent changes to your vision but how quickly this happens and how it changes differs widely between people. These changes may include difficulty with vision in dim light, the loss of side or peripheral vision, and sometimes reduced central vision.

If you have RP, sight loss is gradual and progresses over many years. Most people with RP keep some useful vision well into later life, although some people with RP may become blind.



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 5.5mm in diameter 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 recognising 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

Different types of RP affect the retina. The symptoms you experience depend on the way your retina is affected by RP and can be very different from person to person.

RP is a genetic condition, usually caused by a fault in the genetic information passed down from a parent. Our genes contain the instructions that tell our body how to grow, repair and renew. When a gene is faulty 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 some of the genes that, when faulty, cause RP but there is still much 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 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.

Your eye health practitioner can refer you for genetic counselling. A genetic counsellor can help you to understand the type of RP you have, how it may affect you in the long term and the risk of passing on the condition to any children you may have.

It may also be helpful to perform genetic testing, but because many of the genes causing RP have not been discovered, it may not be possible to find the specific RP gene in your case. In about half the people diagnosed with RP, it is not possible to determine how they inherited the condition.

People with RP may also wish to be entered onto the Australian Inherited Retinal Disease Register and DNA Bank, located at Sir Charles Gairdner Hospital in Perth.

Symptoms of retinitis pigmentosa

In most of the 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 (side) vision. This means that when you look straight ahead, you become less able to see things either to the side, above or below. If RP becomes severe, the peripheral vision may be very limited ("tunnel vision"). Difficulty seeing in low light and loss of peripheral vision could be signs of RP.

For some people, the loss of peripheral vision may mean it is no longer safe to drive. You are required by law to report a permanent or long-term vision condition that might affect your ability to drive. Your eye health practitioner 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 affected first. You might find it difficult to read print or carry out detailed work at this time. In these types of RP, peripheral vision is affected in the later stages.

Retinitis pigmentosa is a progressive condition, but the speed and pattern of deterioration of sight varies from one person to another. Most people with RP eventually have a very restricted visual field, leaving only a narrow tunnel of vision. By 50 years of age many 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 to become 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.

Diagnosis

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 that 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 for 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.

Examination of the retina will require drops that dilate the pupil and may leave your vision blurry for a few hours after.

Retinal photographs and fluorescein angiograms

You may have photographs taken of your retina using a special camera, sometimes using fluorescein dye. The yellow fluorescein dye is injected into a vein in your arm to show up changes in your retina that are not visible using normal photography.

Visual field test

You look straight ahead at a particular point in a bowl-shaped screen in a darkened room, and each time you spot dots of light you click a button. A visual field test usually takes less than 10 minutes for each eye.

Colour vision test

Your colour vision may be tested to check your ability to differentiate one colour from another.

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.

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 complications of retinitis pigmentosa

Some people with RP also develop cataracts, macular oedema and other eye conditions. Discuss these with your eye specialist.

Possible treatments and further research

With the exception of one gene therapy f or a specific type of RP (see below), there is no treatment or cure for RP itself. Complications such as cataract and macular oedema can be treated.

A great deal of research is taking place in the fields of gene therapy, stem cell therapy, growth factors and retinal implants that may lead to effective treatments in future. Talk to your ophthalmologist about whether you might be able to participate in clinical trials. Many of the genes causing RP and related conditions are being discovered or mapped, and this understanding of where the faults occur in the genetic information may enable potential treatments to be devised.

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.

One type of RP, caused by the RPE65 gene, can be treated by Luxturna (voretigene neparvovec) – the first gene therapy approved by the Therapeutics Goods Administration (TGA). This landmark one-time treatment was registered in Australia in 2020 and could lay the foundations for therapies treating this whole group of inherited eye disorders. As of December 2020, Luxturna is still going through the reimbursement process. Please note, MDFA does not endorse any specific treatment or therapy.

Retinal implants

Retinal implant technology works by surgically implanting an electronic chip into the back of the eye to stimulate cells in the retina. While photoreceptor cells may have died, nearby nerve cells can remain intact, and these so-called 'bionic eyes' send signals to those remaining cells that are then perceived as vision by the brain. While retinal implants are now available in the USA and Europe, they are only available in clinical trial settings in Australia.

Nutrition

Some research has shown vitamin A may slow the progression of RP. However, there is concern that too much could be harmful and therefore vitamin A supplements are not recommended.

Refsum syndrome is one of the 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.

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.

Need more information?

Learn more about macular disease at **www.mdfoundation.com.au.**

How's your macula? Take the quiz at **www.CheckMyMacula.com.au.**

You can also access our free, personalised support services and order information kits and Amsler grids by calling our National Helpline on **1800 111 709.**

MDFA has a free newsletter and you can sign up to receive invitations to education sessions and events in your area.

Macular Disease Foundation Australia is committed to reducing the incidence and impact of macular disease, by providing up-to-date information, advice and support.

National Helpline

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