

# Understanding retinitis pigmentosa



supporting blind and  
partially sighted people



**RCOphth**

# RNIB's Understanding series

The Understanding series is designed to help you, your friends and family understand a little bit more about your eye condition.

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**Understanding glaucoma**

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**Understanding retinal detachment**

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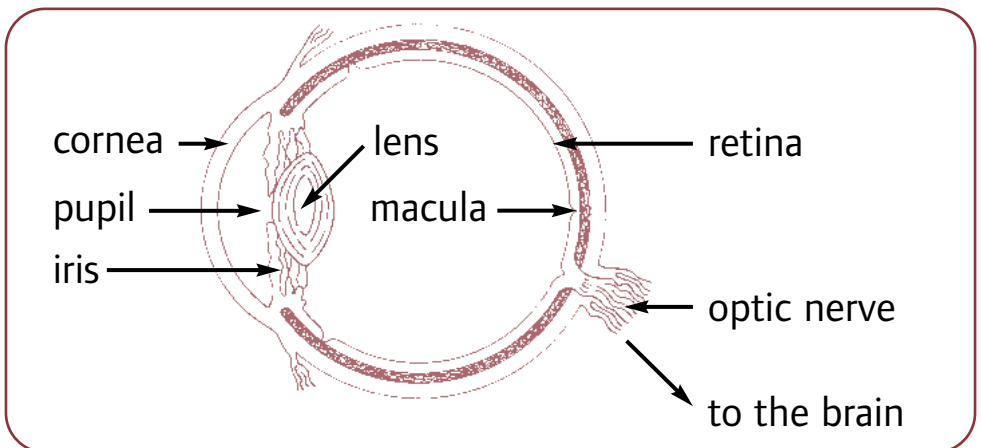
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# About retinitis pigmentosa

Retinitis pigmentosa (RP) is the name given to a diverse group of inherited eye disorders. These eye conditions affect a part of your 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 loss of your side or peripheral vision.

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

New research is constantly changing our understanding of RP. The British Retinitis Pigmentosa Society (RP Fighting Blindness) website and leaflets offer current updates with detailed explanations of these issues.



# How your eye works

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

The retina has two main layers, a thin one called the pigment epithelium and a thicker one, called the neural retina. The neural retina contains many millions of cells called photoreceptors and these cells convert light into electrical signals that travel to the brain.

Light is focused onto a tiny area of the central retina called the macula. This specialised area of the retina is about the size of a pinhead. The macula contains a few million specialised photoreceptors called cone cells. These cone cells work best in bright light levels and allow you to see fine detail for activities such as reading and 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

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 and RP later affects the central cone cells. 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 off. 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 Congenital 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. Sometimes, other parts of the body are also affected. One example of this is Usher syndrome, where people develop both hearing loss and sight loss. 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:

### **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.

### **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.

### **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 is likely to 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. Genetic counselling is a free NHS service. 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 is carried out in regional genetics centres and your eye specialist (ophthalmologist) could refer you to one. 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.

# Symptoms

## Early symptoms

In most of the more common forms of RP, the first symptoms occur between childhood and the age of 30. The first symptom you usually notice is that you find it 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 about 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 a sign that the peripheral rod cells are being affected by RP.

For some people, the early loss of peripheral vision may mean it is no longer safe for you to drive. You are required by law to report a condition which might affect your sight and you need to report a diagnosis of RP to the DVLA so they can carry out regular tests.

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

All RP conditions are progressive, 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 who have RP find the glare from bright lights and sunlight becomes an increasing 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

Most people first experience problems in low light levels and this may prompt them to see their optometrist (optician) or general practitioner (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. By looking into your eye with a piece of equipment called an ophthalmoscope, an optometrist can examine the back of the eye. 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. This is quite an early sign.

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 field of vision 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 field of vision 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 a specialist eye doctor (ophthalmologist) at a hospital for more detailed testing.

# Hospital 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:

## Examining the back of your eye

An examination of the retina is always carried out. You will be given eye drops to dilate 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 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 the retina that are not visible with 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 the bowl-shaped screen of a visual field machine 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**

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 dotted 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.



These tests are usually carried out by the electrodiagnostics department of the eye department. 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 and straight forward 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

Some people with RP also develop cataracts. Cataracts are a clouding of the natural lens at the front of your eye, which is located just behind the iris (the coloured part of your 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. For more information, please see our “Understanding cataracts” leaflet.

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 oedema. This is when the blood vessels near the macula leak and make the retina swell. This blurs and distorts the central vision. Macular oedema can occasionally occur after cataract surgery and very rarely it can happen spontaneously.

# Treatment

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 research into stem cell therapy is to see if stem cells injected into the retina can be persuaded to differentiate into retinal 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.

## **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 specialists. Taking Vitamin A can be bad for your health 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 tuna, cod, and haddock. Ask to be referred to a dietician 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.brps.org.uk](http://www.brps.org.uk).

# Coping

Being diagnosed with an eye condition can be very upsetting. You may find that you are worried about the future and how you will manage with a change in your vision. All these feelings are natural.

Some people may want to talk over some of these feelings with someone outside their circle of friends or family. RNIB can help you with our telephone Helpline and our emotional support service. Your GP or social worker may also be able to help you find a counsellor if you think this would help you.

RP Fighting Blindness also offers information and advice for people affected by RP and is an excellent source of the latest research regarding RP and support for people diagnosed with RP and their families. Their national RP Helpline number is 0845 123 2354.

## Help to see things better

Having RP can cause serious changes to vision in the long term, but much can be done to help you make the most of your remaining vision and adapt to any changes.

There are lots of things that you can do to make the most of the vision you have. This may mean making things bigger, using brighter lighting or using colour to make things easier to see. Ask your ophthalmologist, optician or GP to refer you to your local low vision service, which can provide you with magnifiers to help with reading and advice on lighting to help make the most of your sight.

Local social services should also be able to offer you information on staying safe in your home and getting out and about safely. They should also be able to offer you some practical mobility training to give you more confidence when you are out.

Our Helpline can also give you information about low vision clinics and the help available from social services on 0303 123 9999. They can also offer advice if you have any difficulties accessing these services. Our website [www.rnib.org.uk](http://www.rnib.org.uk) offers lots of practical information about adapting to changes in your vision and products that make everyday tasks easier.

# Useful contacts

## **Royal National Institute of Blind People**

105 Judd Street, London WC1H 9NE

t: 0303 123 9999

helpline@rnib.org.uk

www.rnib.org.uk

## **RP Fighting Blindness**

(The British Retinitis Pigmentosa Society)

PO Box 350, Buckingham MK18 1GZ

t: 01280 821 334

RP Helpline 0845 123 2354

info@brps.org.uk

www.brps.org.uk

## **Royal College of Ophthalmologists**

17 Cornwall Terrace, London NW1 4QW

t: 020 7935 0702

www.rcophth.ac.uk

## **Sense**

101 Pentonville Road, London N1 9LG

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# Information sources

We do all we can to ensure that the information we supply is accurate, up to date and in line with the latest research and expertise.

The information used in RNIB's Understanding series of leaflets uses:

- Royal College of Ophthalmologists guidelines for treatment
- clinical research and studies obtained through literature reviews
- information published by specific support groups for individual conditions
- information from text books
- information from RNIB publications and research.

For a full list of references and information sources used in the compilation of this leaflet email [publishing@rnib.org.uk](mailto:publishing@rnib.org.uk) or call 020 7391 2006.

# RNIB Helpline

0303 123 9999

helpline@rnib.org.uk

If you, or someone you know, is living with sight loss, we're here to help.

This leaflet has been produced jointly by the Royal College of Ophthalmologists and Royal National Institute of Blind People.

© RNIB and RCOphth  
RNIB reg charity no. 226227  
RCOphth reg charity no. 299872



Printed September 2010. Review date September 2011.

ISBN: 978 1 85878 730 5

PR10879