

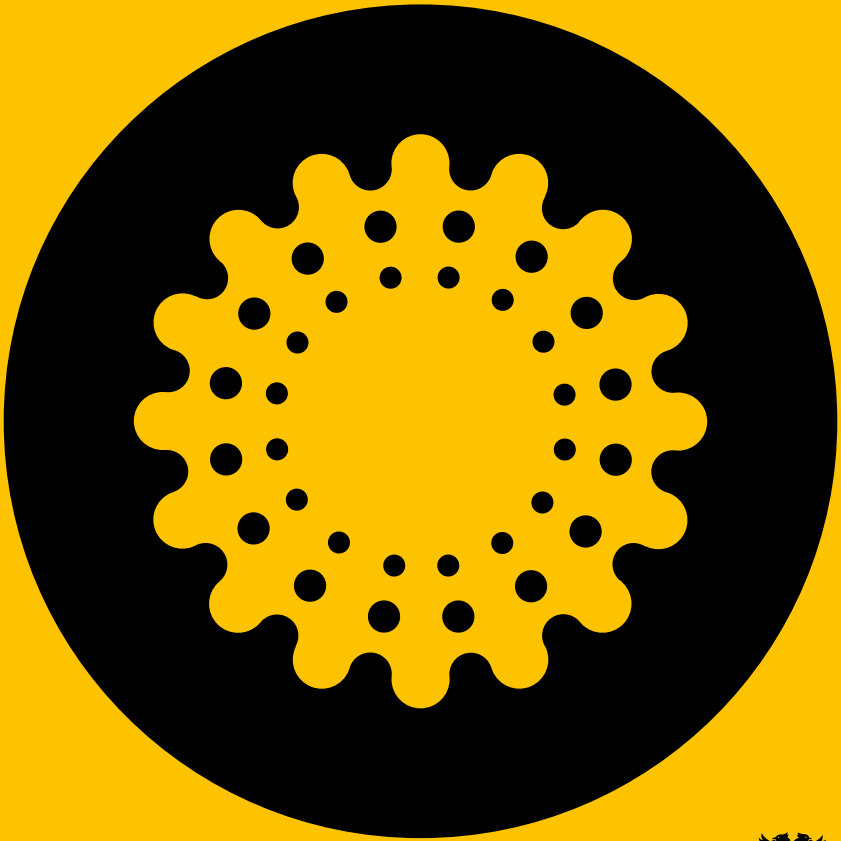
**R N I B**

**Understanding**

See differently

# Retinitis pigmentosa

and other inherited retinal dystrophies



*The ROYAL COLLEGE of*  
**OPHTHALMOLOGISTS**

## Contact us

We're here to answer any questions you have about your eye condition or treatment. If you need further information about retinitis pigmentosa or other inherited retinal dystrophies, then our Helpline is there for you.

Just give us a call on **0303 123 9999** or email us at **helpline@rnib.org.uk** and we'll be happy to speak with you.

## 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.

The series covers a range of eye conditions, and is available in audio, print and braille formats.

# Contents

- 4** What is retinitis pigmentosa?
- 6** How your eye works
- 8** What are the symptoms of RP?
- 12** What caused my RP?
- 14** How is RP inherited?
- 17** How does RP affect my family?
- 20** Can other parts of my body be affected?
- 21** What tests are used to detect RP?
- 27** What other eye conditions might I get?
- 29** Is there any treatment for RP and what research is being carried out?
- 33** Coping
- 34** Further help and support

# What is retinitis pigmentosa?

Retinitis pigmentosa (RP) is the name given to a group of inherited eye conditions called retinal dystrophies. A retinal dystrophy such as RP affects the retina at the back of your eye and, over time, stops it from working. This means that RP causes gradual but permanent changes that reduce your vision. How much of your vision is lost, how quickly this happens and your age when it begins depends on the type of RP that you have.

If you have RP, the changes to your retina can affect your peripheral vision (also known as your side vision) and make it difficult for you to see in dim light or in the dark. Your central vision can also become affected and this will make it difficult for you to see colour and do things such as reading or watching television.

The changes in your vision happen over years rather than months, and some people lose more sight than others.



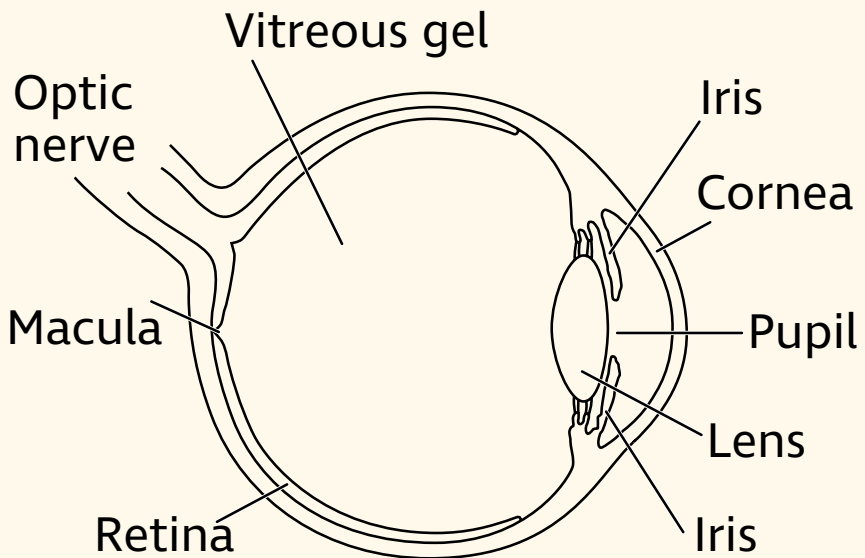
## How your eye works

When light enters your eye, it is focused onto your retina at the back of your eye. The retina has a number of layers, but the most important for vision is a layer made up of cells called photoreceptors. Photoreceptors are cells which are sensitive to light.

The macula, which is the central part of the retina, contains a few million specialised photoreceptor cells called cone cells. These cone cells work best in bright light and allow you to see fine detail for activities like reading and writing and recognising colours.

The peripheral retina is further away from the central macula. It is mostly made up of the other type of photoreceptors called rod cells. They allow us to see when light is dim and provide peripheral vision outside of the main line of sight.

RP causes damage to the rod cells in particular. Other types of retinal dystrophies may affect the cone cells which are concentrated in your macula.



# What are the symptoms of RP?

## Early symptoms

When you have a retinal dystrophy like RP, your rod and cone cells gradually stop working. Depending on the type of RP you have, you may notice your first symptoms in your early childhood or later, between the ages of 10 to 30. Some people don't have symptoms until later in life.

In RP, the first symptom you'll notice is not seeing as well as people without a sight condition in dim light, such as outside at dusk, or at night. This is often called "night blindness". People without a sight condition can fully adapt to dim light in 15 to 30 minutes, but if you have RP it will either take you much longer or it won't happen at all.

You may start having problems with seeing things in your peripheral vision. You may miss things to either side of you and you might trip over or bump into things that you would have seen in the past.

Difficulty seeing in dim light and loss of peripheral vision are signs that the rod cells in your retina are being affected by your RP.



In the more common forms of RP, your cones are not affected in the early stages so your central vision will still be good enough to recognise faces and to continue reading.

If your peripheral vision has started to change, it may mean it is no longer safe for you to drive. If you have a condition such as RP which can affect your sight in both eyes, you are required by law to report it to the Driver and Vehicle Licensing Authority (DVLA). They will assess your vision regularly to find out if your sight meets their standards.

## **Early symptoms of other retinal dystrophies**

In some other retinal dystrophies, such as cone-rod dystrophy, central vision is affected before peripheral vision. This is because the cone cells are affected first or affected more severely than the rod cells. You'll find reading and seeing detail difficult and your colour vision will be affected.

## **Later symptoms**

RP is a progressive condition, which means that your sight will continue to get worse over the years. Often, changes in sight can

happen suddenly over a short period of time. You may then have a certain level of vision for quite some time. However, there may be further changes to your vision in the future. This may mean that you have to keep re-adapting to lower levels of sight. The type of RP that you have can affect how quickly these changes develop.



As your RP progresses, you'll gradually lose your peripheral sight, leaving a central narrow field of vision, often referred to as having "tunnel vision". You may still have central vision until you're in your 50s, 60s or older. However, advanced RP will often affect your central vision and you may find reading or recognising faces difficult.

You may find that bright lights and sunlight give you problems with glare and moving between a light and a dark room can be difficult. This is because, as your RP progresses, your retinal cells become less able to adapt to changing light levels.

Your family or the people you live with can help you as your vision deteriorates by keeping your home environment free of obstacles and by putting things away in the same place so they are easy to find.

## What caused my RP?

RP is a hereditary condition caused by a fault in one of the genes involved in maintaining the health of the retina. You inherit genes from your parents. Your genes give the cells in your body the instructions they need to work well and stay healthy. When a gene is faulty, it is because there is a fault in their instructions and the cells using those instructions don't work as they should. In RP, the faulty gene causes your retinal cells to stop working and to eventually die over time. Researchers have identified many of the genes that cause RP and the faults within them, but there are still other genes to discover.

There have always been different types of RP, but in the past, they've all been given the same name – retinitis pigmentosa. This happened because many of the conditions looked the same when ophthalmologists looked inside someone's eyes. However, as research developed, giving a better understanding of how our genes cause eye conditions, it became possible to tell the difference between conditions which would have all been called RP in the past. This means that some people may have their RP more accurately renamed as

a rod-cone dystrophy or cone-rod dystrophy because of the genes involved.

Other retinal dystrophies include Leber's congenital amaurosis, cone dystrophy, cone-rod dystrophies, rod-cone dystrophies, choroideremia and macular dystrophies.



## How is RP inherited?

About half of people with RP have another family member with the condition. The way RP is passed from generation to generation can tell you who in your family has had the condition, how severely your vision could be affected and the chances of your children being affected.

Genes usually come in pairs. You inherit one gene from each of your parents to make each pair. When you have children, you only pass on one gene to them. You can inherit RP in three different ways – autosomal dominant, autosomal recessive and X-linked.

### Autosomal dominant inheritance

To have autosomal dominant RP, you only need one faulty gene to have the condition. This can be inherited from either your mother or your father. Usually this parent will also have RP.

Autosomal dominant RP affects men and women equally and there tends to be a known history of the condition in your family. If you have autosomal dominant RP, there is a one in two risk of passing on the condition to each of your children.

## **Autosomal recessive inheritance**

Autosomal recessive RP requires two faulty genes, one inherited from your mother and one inherited from your father. If both your parents have one normal gene and one faulty gene, they are carriers of RP and their vision is unaffected by the condition. If both your parents pass on their faulty gene to you, you will inherit autosomal recessive RP.

If you have autosomal recessive RP, you will pass on a faulty gene to all your children. If they inherit a normal copy of the gene from their other parent, they will be carriers of RP. Because you need two copies of the faulty gene to have this type of RP, it usually appears in families without any history of the condition in other generations. This type of RP affects men and women equally.

## **X-linked inheritance**

This is a type of RP that affects men. It can be severe and result in poor sight by the age of 30 to 40.

X-linked RP relates to our sex chromosomes. Men have one X and one Y chromosome. Women have two X chromosomes. The gene relating to X-linked RP is found on an

X chromosome. This means men with a faulty gene on their X chromosome will have the condition. If a woman has a faulty gene on one X chromosome but a normal gene on the other, she is usually more mildly affected or does not have any symptoms from the condition at all. This means she is a carrier of the condition.

A man with X-linked RP will pass on the faulty gene to all his daughters but not to his sons. His daughters will be carriers of the condition but his sons won't be affected.

### **No known relative**

About half of people with RP don't know of any members of their family with the condition. This may be because their relatives were carriers of RP and haven't shown signs of the condition themselves. If there is no known relative in your family, it may not be possible to find out how your RP has been inherited without the help of genetic testing to find out which genes are faulty.



# How does RP affect my family?

## Genetic testing

Genetic testing can be carried out to try to find out if you have a faulty gene that causes RP. This can either identify the faulty gene that is causing your RP or enable you to find out if you're carrying a faulty gene that your children may inherit. There are several genetic centres around the country that carry out genetic tests and your ophthalmologist (also known as a hospital eye doctor) or your GP can refer you to one.

Genetic testing uses a blood test to look at your genes to see if they're faulty. Testing for RP and other inherited retinal dystrophies is complicated. It doesn't identify all forms of these conditions as new faulty genes are still being discovered. Ask your ophthalmologist or GP to discuss genetic testing with you.

## Genetic counselling

Genetic counselling can help you to understand the type of RP you have, how it's likely to affect you in the future and the risks of passing on the condition to any children you may have. Genetic counselling is usually advised when you have genetic testing. A genetic counsellor asks about your family tree in detail to try and understand how RP has been inherited in your family. Genetic counselling is a free NHS service. It may be provided by a specialist RP clinic or a medical genetics department. You can ask your GP or ophthalmologist to refer you to your local genetic counselling service.

Having a genetic condition in your family may cause emotional concerns. Talking to a genetic counsellor may help you and your family to discuss the eye condition in your family. Knowing the chances of passing on any condition you have can help if you are thinking about starting a family.



## Can other parts of my body be affected?

In most cases, the inherited faulty gene 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 disease, and Alström and Bardet-Biedl (BBS) syndromes, all of which cause RP-like sight loss along with other health conditions.



## What tests are used to detect RP?

If you've noticed that you're having problems seeing in dim light or at night, you should see your optometrist (also known as an optician) or your GP. Early symptoms can vary from person to person so your RP might be diagnosed at an early stage or after many years of having the condition.

An optometrist will examine your retina to detect RP. To do this, they will look into your eyes using either a special microscope called a slit lamp or an instrument called an ophthalmoscope. If you have the early signs of classic RP, there will be tiny but distinctive clumps of dark pigment around your retina.

Any changes to your peripheral vision can only be detected by a field of vision test, which your optometrist can also carry out. This test may not be offered to you routinely so if you're worried about your peripheral vision, you should ask your optometrist to check your field of vision for you.

If you've got a family history of RP and you have problems with your vision in dim light or problems when moving from light to dark, you should tell the person examining your

eyes. This will help them to carry out the most appropriate tests. If your optometrist is concerned after your eye examination, they can refer you to an ophthalmologist for more tests.

## **What tests will the hospital do?**

When you're referred to hospital, you'll be examined by an ophthalmologist. There are various tests that can diagnose RP, but it's unlikely that they'll do all of them at your first visit. These tests can also monitor how your RP changes over time. Your ophthalmologist may be able to say that you have RP when they've got the results of these tests, but it may not be possible to know exactly what type of RP you have and what the long term effects on your vision will be without genetic testing.

It's important to ask your ophthalmologist about these tests and about what the results mean for you. None of the tests are painful or cause you any harm but they may take a long time and be repetitive. Here are some of the tests you may need to undergo:

### **Examining the retina at the back of your eye**

Your retina will be examined each time you visit the hospital. You'll be given drops to dilate (widen) your pupils to allow the ophthalmologist

to see your retina clearly. These drops take about 30 minutes to work. They'll make you sensitive to light and make your vision blurry. The effects of the drops usually wear off in about six hours, though sometimes it can take overnight. It isn't safe to drive until the effects have worn off.

## **Retinal photographs, fluorescein angiograms and autofluorescence imaging**

Your retina may be photographed using a special camera. By comparing the photographs taken on different visits, your ophthalmologist might be able to monitor how your RP is changing over time.

Your ophthalmologist may ask for a more specialised set of photographs to be taken after a yellow dye called fluorescein has been injected into a vein in your arm. As the dye travels into the tiny blood vessels in your retina, a series of photographs are taken. The dye in the blood vessels will reveal the changes in your retina that can't be seen with normal photography. The fluorescein dye can make your skin look yellow for up to 24 hours. It leaves your body through your urine, which will be a deep yellow colour for about 24 hours too.

Autofluorescence imaging involves taking more pictures of the back of your eye that show another retinal change that can't be seen with normal photography. These pictures show the ophthalmologist how the layer under your retina, called the retinal pigment epithelium, is functioning. This layer helps the retina to work and if it's not working properly itself, the health of the retina will be affected.

### **Visual field test**

A visual field test uses a machine which checks how much of your peripheral vision has been affected by an eye condition. One of your eyes is covered with a patch and your chin rests on the machine, which is in a darkened room. You'll be given a button to hold in your hand and asked to look straight ahead at a central point on the machine's bowl-shaped screen. It's important to keep looking at this central point and not to move while the test is being carried out. You'll notice spots of light flash on the screen and each time you see one, you press the button you're holding. The test takes about 10 minutes for each eye and shows how much vision you have above, below and to the sides of looking straight ahead.



## **Colour vision**

To test your colour vision, you'll be asked to pick out numbers or patterns that you can see on a background of coloured dots. This test takes less than five minutes to do and shows what colours you're able to see.

## **Electro-diagnostic tests**

Electro-diagnostic tests can tell your ophthalmologist how well your retina is working. They check how your retina responds electrically to patterns and different lighting conditions. Different tests can be carried out to show the results of your retina's electrical activity. These test results will indicate which layers of your retina have been affected.

The tests you may be offered include the electroretinogram (ERG), the pattern electroretinogram (PERG) and the electro-oculogram (EOG). The ERG shows how your retinal cells are working and you'll usually have to sit in a darkened room or wear dark goggles for about 20 to 30 minutes to start with. Then you'll be given anaesthetic eye drops before being asked to wear a special contact lens on your eye. During this test, you'll be shown flashing lights and the

response of your retina will be recorded on an electrical trace or plot.

The ERG tests the whole of your retina but the PERG uses a checkerboard pattern to check how your macula, at the centre of your retina, is working.

The EOG shows how the rods and cones and the retinal pigment epithelium behind them are working.

These tests are usually carried out by the electrodiagnostics department of the eye clinic and you should ask the staff to explain exactly what will happen for each test before they're carried out. The tests are painless and straightforward, but may involve having your eyes dilated and/or numbed, a tiny electrode being placed on your eye and a sensor on your skin.

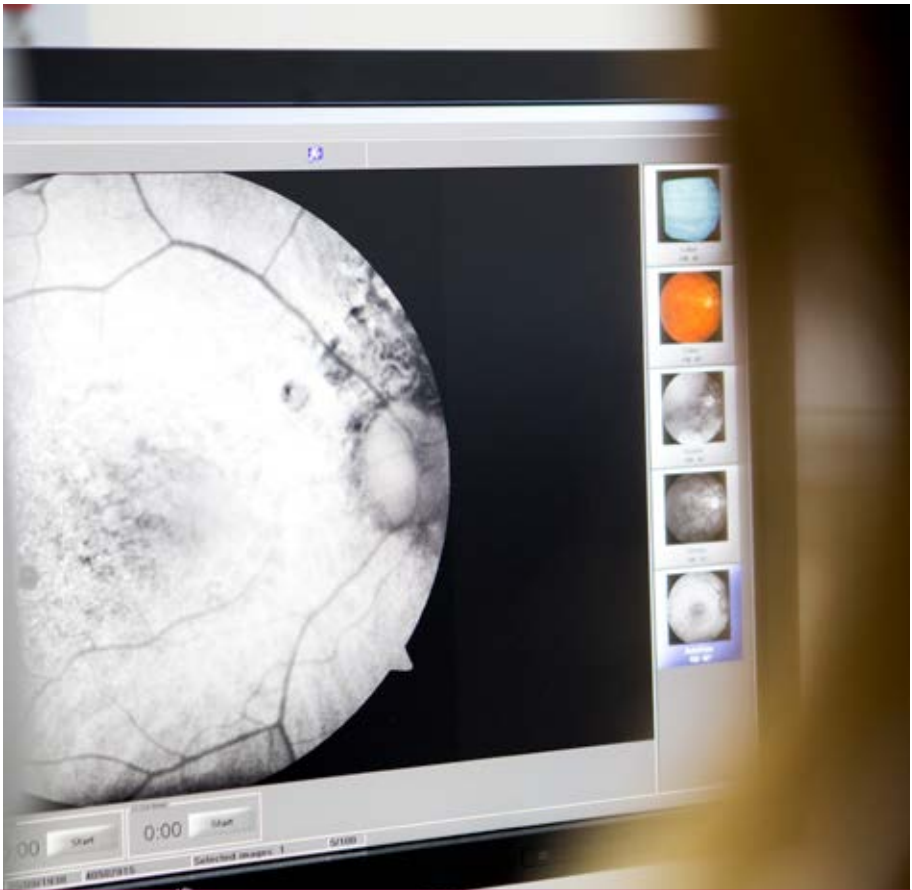
## What other eye conditions might I get?

Some people with RP also develop cataracts. A cataract is a clouding of the lens in your eye. Your lens sits just behind your iris, the coloured part of your eye. When you have RP, you may develop a cataract as young as your 20s, but it's more often picked up in your 40s. Your ophthalmologist may recommend that you have your cataract removed, particularly if it's making your remaining useful vision misty. Your cataract can be removed and replaced with an artificial lens made of plastic or silicone. For more information about cataracts or to order a copy of our publication called Understanding Cataracts, call our Helpline on **0303 123 9999**.

Many people find that although they still have sight loss due to their RP, their useful remaining vision is of a better quality after their cataract has been removed.

Some people with RP develop macular swelling, known as oedema. The macula is at the centre of your retina and you use it to see fine detail and colour. If the blood vessels near your macula leak, they can make your macula swollen. This can blur and distort

your central vision. Macular oedema can happen occasionally after cataract surgery. It can be diagnosed with a scan of the macula using a technique called optical coherence tomography (OCT).



# Is there any treatment for RP and what research is being carried out?

While much progress has been made in the past few years in the understanding of the genes involved in RP, there is currently no cure or treatment which can slow down or stop RP from getting worse. Ongoing research may lead to a treatment in the future, but it may be a number of years before a tried and tested treatment for RP is produced. The types of treatment which are being researched at the moment include:

## 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 a new gene that works properly. Normal genes are injected into the affected retina using a harmless virus to carry the genetic material. The hope is that the affected cells then begin to work correctly and the damage is either stopped or reversed. Gene therapy relies on knowing which gene is faulty. In many cases of RP, the faulty genes are still to be discovered.

However, in recent clinical trials, there's already been some success in using gene therapy to improve vision for people with conditions similar to RP. It is hoped that gene therapy will be successful in treating RP in the future.

## **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 specialised cells that the body cannot easily replace. Stem cells are cells that can divide many times and can replace damaged or missing cells in different organs and tissues of the body. If stem cells can be turned into the specialised retinal cells, it may be possible to replace the cells that have been damaged in RP.

## **Growth factors**

Growth factors are chemicals that support cells to grow and repair. Research groups are working on using growth factors to treat retinal disease.

## **Artificial vision**

When RP has caused severe visual loss, it's possible that damaged retinal cells could be replaced by electronic implants. These implants

are placed on or beneath the retina to stimulate the remaining retinal cells. When the remaining retinal cells are stimulated and the optic nerve is healthy, a signal may be passed along the optic nerve to the brain allowing the person to see patterns of light or outlines of objects.

These implants don't bring your vision back or stop your vision from getting worse. You'd still need the other aids you have, such as your cane or your guide dog. Artificial vision systems are still being investigated and there isn't a system which is able to be easily implanted which returns high levels of vision. No artificial systems are currently available on the NHS.

## **Nutrition**

There's no evidence that suggests taking vitamin supplements or having a particular diet will help you avoid sight loss if you have RP. In the past there has been some debate about vitamin A and whether people with RP should take vitamin A supplements. However, research has found that vitamin A supplements do not protect sight for people with RP. Taking large doses of vitamin A can be bad for your health and should be discussed with your GP and ophthalmologist.

People with a condition called Refsum's syndrome, which includes RP-like changes to the retina, should have a special diet which avoids something called phytanic acid which is found in dairy, beef, lamb and fatty fish such as tuna, cod and haddock. If you have been diagnosed with Refsum's syndrome, you should see a dietician for help with your diet.





## Coping

It's completely natural to be upset when you've been diagnosed with a retinal dystrophy like RP and it's normal to find yourself worrying about the future and how you'll manage with the change in your vision.

It can sometimes be helpful to talk about these feelings with someone outside your circle of friends or family. At RNIB, we can help with our telephone Helpline and our Sight Loss Counselling team. Your GP or social worker may also find a counsellor for you if you feel this might help.

Your eye clinic may also have a sight loss adviser (also known as an Eye Clinic Liaison Officer or ECLO), who can be on hand to provide you with further practical and emotional support about your eye condition.

Retina UK is a UK charity that gives advice and support to anyone affected by RP and other retinal dystrophies. They're able to provide up to date information about the latest research being carried out. Their national Helpline number is **0845 123 2354**.

## Further help and support

Having RP means that you'll eventually lose sight, but there are things you can do to make the most of your remaining vision. This may mean making things bigger or smaller, using brighter lighting or using colour to make things easier to see. We have a lot of helpful information about living with sight loss as well as making the most of the sight that you have. You can find out more by calling our Helpline.

Ask your ophthalmologist, optometrist or GP about low vision aids, such as a magnifier, and ask for a referral to your local low vision service.

You should also ask your ophthalmologist whether you're eligible to register as sight impaired (partially sighted) or severely sight impaired (blind). Registration can act as your passport to expert help and sometimes to financial concessions. Even if you aren't registered, a lot of this support is still available to you.

Local social services should be able to give 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.



If you have questions about anything you've read in this publication, please get in touch with us.

Our Helpline is your direct line to the support, advice and services you need. Whether you want to know more about your eye condition, buy a product from our shop, join our library, find out about possible benefit entitlements, or be put in touch with a trained counsellor, we're only a call away.

It's also a way for you to join RNIB Connect, our community for anyone affected by sight loss. RNIB Connect is free to join and you'll have the chance to meet other people with similar experiences in our helpful, welcoming and supportive community.

Give us a call today to find out how we can help you.

**RNIB Helpline**  
**0303 123 9999**  
**[helpline@rnib.org.uk](mailto:helpline@rnib.org.uk)**

We're ready to answer your call Monday to Friday 8am to 8pm and Saturday 9am to 1pm.

You can also get in touch by post or by visiting our website:

**RNIB, 105 Judd Street  
London WC1H 9NE  
[rnib.org.uk](http://rnib.org.uk)**

## **Other useful contacts**

### **Retina UK**

Wharf House, Stratford Road,  
Buckingham MK18 1TD

Helpline **0845 123 2354**  
**[helpline@retinauk.org.uk](mailto:helpline@retinauk.org.uk)**  
**[retinauk.org.uk](http://retinauk.org.uk)**

### **Sense**

101 Pentonville Road  
London N1 9LG

Text **0300 330 9256**  
**[info@sense.org.uk](mailto:info@sense.org.uk)**  
**[www.sense.org.uk](http://www.sense.org.uk)**

### **Driver and Vehicle Licensing Authority (DVLA)**

Drivers' Medical Enquiries

Swansea SA99 1TU

**0300 790 6806**  
**[www.dvla.gov.uk](http://www.dvla.gov.uk)**

## **We value your feedback**

You can help us improve this publication by letting us know what you think about it. Please complete and return the form opposite to:

### **RNIB**

Eye Health Information  
105 Judd Street  
London  
WC1H 9NE

You can also email us at  
[\*\*eyehealth@rnib.org.uk\*\*](mailto:eyehealth@rnib.org.uk)

Please include your contact details if you're requesting information.



1. Where did you receive your copy of this publication?

---

---

---

---

---

---

---

---

2. Did you find the information easy to read and understand? Please give details of anything you feel could be improved.

---

---

---

---

---

---

---

---

---

---



3. Is there any information you would have found helpful, that was missing?

---

---

---

---

---

---

---

---

---

---

4. Do you have any other comments about this publication or any aspect of your contact with RNIB?

---

---

---

---

---

---

---

---



# Information sources

RNIB and The Royal College of Ophthalmologists 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.

This publication uses information from:

- The Royal College of Ophthalmologists' guidelines for treatment
- clinical research and studies obtained through literature reviews
- specific support groups for individual conditions
- medical text books
- RNIB publications and research.

For a full list of references and information sources used in the compilation of this publication, email **eyehealth@rnib.org.uk**.

# About The Royal College of Ophthalmologists

The Royal College of Ophthalmologists champions excellence in the practice of ophthalmology and is the only professional membership body for medically qualified ophthalmologists.

The College is unable to offer direct advice to patients. If you're concerned about the health of your eyes, you should seek medical advice from your GP or ophthalmologist.

**[rcophth.ac.uk](http://rcophth.ac.uk)**



U.S. GOVERNMENT PRINTING OFFICE: 1963 O - 350-000

FOR TESTING VISUAL ACUITY (Snellen) TYPE  
BY ORDER OF THE DIRECTOR, FEDERAL BUREAU OF SURVEY

SNELLEN  
TYPE

+18- R N O V S -18-

+16- Z C R D H -16-

+14- N V S O K -14-

+12- DRZKO -12-

+10- SNHCV -10-

+08- CRVSZ -08-

+06- VKCNH -06-

+04- SVKDN -04-

+02- KDHZC -02-

+00- HZCOR -00-

-02- QKDN -02-

-04- ZENH -04-

-06- KVVV -06-

-08- VVVV -08-



The Bureau of Survey  
Department of Health, Education and Welfare

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

**RNIB Helpline  
0303 123 9999  
helpline@rnib.org.uk**

The Sight Advice FAQ answers questions about living with sight loss, eye health or being newly diagnosed with a sight condition. It is produced by RNIB in partnership with a number of other sight loss organisations. [sightadvicefaq.org.uk](http://sightadvicefaq.org.uk)

This leaflet has been produced jointly by RNIB and The Royal College of Ophthalmologists.

Printing of this booklet was supported by a grant from Novartis Pharmaceuticals UK Ltd, who had no influence on the content.

**Produced date October 2017  
Review date October 2020**

**PR10879  
ISBN 978-1-85878-768-8  
Ed 1**

