Devon in Sight Eye Condition Fact Sheet 23

Retinitis Pigmentosa (RP)

Introduction

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.

Photograph: Visual representation of Retinitis Pigmentosa

If you have Retinitis Pigmentosa, 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.

What are the symptoms of Retinitis Pigmentosa?

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.

What causes Retinitis Pigmentosa?

Retinitis Pigmentosa 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.

How is Retinitis Pigmentosa 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.

To have **Autosomal Dominant inheritance**, 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 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 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 Retinitis Pigmentosa affect my family?

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 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.

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. It is hoped that gene therapy will be successful in treating RP in the future.

Stem cell therapy: 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. 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.

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. We're here to support you every step of the way, and to answer any questions you may have – just get in touch with RNIB Sight Loss Advice Service. https://www.rnib.org.uk/advice

Having RP means that you'll eventually lose your 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. A low vision assessment can explore how to make the most of your sight. Your GP, optometrist or ophthalmologist can refer you to your local low vision service for an assessment. You can also find out tips for making the most of your sight by downloading our guide:

You should also ask whether you are 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.

https://www.rnib.org.uk/eye-health/registering-your-sight-loss

Retina UK works to support those affected by inherited progressive sight loss and funds medical research. https://retinauk.org.uk/

What to do next?

If you are experiencing any of the above symptoms or your sight loss deteriorates, please contact your GP/Optician/Accident & Emergency Department at your local Hospital.

If you need any further advice call our Helpline: 01392 876 666

Devon in Sight is a member of the **Helpline Partnership Community**

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Support Available from Devon in Sight

- **Information, Advice and Guidance**
- **Demonstrations of Low Vision Aids & Daily Living Equipment**
- **Assistive Technology Solutions**
- **Our holistic needs assessment (Sight Loss MOT)**
- **Training for people with central vision loss (Skills for Seeing)**

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Devon in Sight is not a medical organisation; therefore we can only provide general information that is not intended to be a substitute for a proper medical assessment. Our information is not intended to be used for individual cases. If you have a specific question about your eye condition, we recommend that you consult an eye care professional.

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Devon in Sight Contact Information

Devon in Sight
Your local sight loss charity

Helpline 01392 876 666
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Devon in Sight is a Registered Charity No 1140978

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