



**RETINA**  
australia

### **Retinitis pigmentosa (RP)**

Retinitis pigmentosa (RP) refers to a group of inherited diseases that affect the photoreceptor (light sensing) cells responsible for capturing images from the visual field. These cells line the back of the eye in the region known as the retina. People with RP experience a gradual decline in their vision because the two types of photoreceptor cells – rod and cone cells – die.

Rod cells are present throughout the retina, except for the very centre, and they help with night vision. Cone cells are also present throughout the retina but are concentrated in the central region of the retina (the macula). They are useful for central (reading) vision and for colour vision.

In RP, the rod cells, and eventually the cone cells stop working, causing vision loss; however, many people with RP retain useful central vision well into middle age.

### **What are the symptoms of RP?**

Rod cells are usually initially involved as previously mentioned, and difficulty seeing in dim light, including transitioning from light to dark and vice versa, is one of the earliest symptoms experienced. There can be a very variable range in the onset of RP. Some people are diagnosed in childhood while others are not affected until they are adults.

The condition is slowly degenerative, but the rate of progression and degree of visual loss can vary from person to person and even among affected members of the same family. It is therefore very difficult to predict what an individual's vision will be like at a specific time in the future. Both eyes are usually affected in a similar way.

### **What is the cause of RP and how is it inherited?**

In Australia, it is estimated that one in 3,000 people has RP, which equates to approximately 8,500 individuals. RP is one of the most complicated genetic conditions of all, and over 50 different genes have been identified to be causative for various forms of RP. There are various inheritance patterns for RP, including autosomal dominant (30-40%), autosomal recessive (50-60%) and X-linked (5-15%).

Approximately 50% of RP patients will have a history of at least one other family member being affected. 50% of patients will not have a family history of the condition. While their RP is still caused by a gene alteration, it might not be possible to determine the inheritance pattern in these patients. The autosomal dominant form of disease tends to follow a milder course with maintenance of preserved vision well into late middle age. The X-linked form is the most severe and central vision may be lost by the third decade.

RP is a genetic disease, but cases with no family history also commonly occur. If a family member is diagnosed with RP, it is strongly advised that other members of the family also have an eye exam by an eye doctor (ophthalmologist) who is specially trained to detect retinal diseases.

## **What treatments are available?**

Maximising the remaining vision that an individual has is a crucial first step to take. There are many new low vision aids including telescopic and magnifying lenses. The wide range of assistive technologies for people with visual impairments provides plenty of choice for users at all stages of sight loss, and this technology has also removed many barriers to education and employment.

There are, as of yet, no proven or effective cures for RP, although research in this area has recently accelerated. The term RP represents an extremely varied number of diseases, as scientists have now identified more than 50 genes that can have mutations causing RP. A number of these were discovered in Australian individuals by the team at the Australian Inherited Retinal Disease Register (AIRDR) based in Perth, Western Australia, which has been given funding grants by Retina Australia over many years. It is likely that mutations in more than 100 different genes will eventually be identified in coming years.

Retina Australia has also given many grants to Australian researchers studying different aspects of retinal degeneration (in 2008, 2009, 2010, 2011, 2012, 2013, 2016 and 2017), as well as other studies which will add to a body of knowledge about inherited retinal diseases which will also advance research into RP. Typically, each person with RP only has damage in one pair of genes, and gene therapy to replace defective genes by inserting healthy genes into the retina via harmless viruses is being explored in clinical trials for a small number of RP genes.

Scientists have also begun to treat animals with many other forms of RP with this approach and several more treatment trials in humans are expected to begin in the near future. Each of these hoped for new treatments will be specific for the gene responsible for the individual patient's form of RP.

The early establishment of an affected person's genetic status is vital if he or she is to have the opportunity to take full advantage of these emerging therapies. The Australian Inherited Retinal Disease Register and DNA Bank is actively used by clinicians and researchers to identify participants who may be suitable for emerging gene-specific clinical trials, to improve our understanding and treatment of inherited retinal diseases, and to facilitate clinical counselling of patients.

Another research area currently being explored is the area of promising drug treatments which aim to preserve the function of the rod and cone photoreceptor cells, thereby keeping them alive for longer. Many of these drugs are re-purposed drugs, which may have already been approved for a different disease and are now being tested for their effectiveness in RP. It is estimated that as few as 5% of cone cells need to be preserved by such a treatment in order to have a huge impact on quality of life by the maintenance of a small but significant amount of central vision. Gene therapy and drug therapies hold huge promise to treat individuals at an early to mid-stage of disease progression, where there are still some viable rod and cone cells present.

For individuals who may have lost a significant portion or all of their vision, there are other technologies that are being investigated, such as stem cell therapies and retinal implant technologies. Stem cell technology holds great potential to replace retinal cells that have already died due to degeneration. Scientists are currently working on replacing two different cell types by stem cell therapy – retinal pigment epithelium (RPE) cells and photoreceptor cells. RPE cells are a special type of cell that support the photoreceptor cells, but are not responsible for “seeing”, therefore it is hoped that replacement of RPE cells will help the retina function better, prevent further vision loss, and help nourish surviving retinal cells. This cell type would need to be replaced in time to help support a retina that is still working. Efforts at transplanting stem cell-derived photoreceptor cells are at an even earlier stage of research, however a number of recent animal studies have shown the potential to restore function in the eye, which may pave the way for human studies in the future.

Retinal implants are a form of biomedical technology currently being developed for RP. A number of these implants have shown success in delivering a form of artificial vision to individuals with total vision loss due to RP. When all or most of the photoreceptor cells have died, they can theoretically be replaced by an electronic microchip that brings a visual image to the remaining cells of the retina. These microchips electronically signal the remaining retinal cells which pass the signal down the optic nerve for processing as a visual image by the brain. At the moment, these devices do not restore natural vision, but can help to restore mobility, by allowing an individual to see a difference in light and dark to the point where they can tell how to walk through a doorway.

In 2014 Retina Australia made a funding grant for the creation of a battery of tests called LoVADA (Low Vision Assessment of Daily Activities) to assist with testing the bionic eye, and subsequently in 2015 made a grant to support a trial of the bionic eye. Despite the lack of current treatments for RP, it is still very important to continue having regular general eye check-ups. This is because people with RP are still at risk for other kinds of eye problems that can affect the general population and may be treatable. RP patients tend to develop cataracts at an earlier age than the non-RP population and can do very well from cataract surgery, although the visual outcome obviously depends on the severity of the retinal degeneration. Regular visits to your eye doctor can also make you aware of current advances as we learn more about RP.