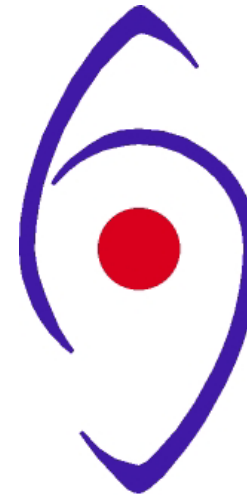


**Visual Impairment Services  
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Visual Impairment Services Highland

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**retinitis  
pigmentosa**

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## **What is Retinitis pigmentosa ?**

The name given to a group of diseases which affect the light sensitive cell in the retina called rods and cones. The rod enables us to "see out of the corner of our eyes" and also to see in poor light and dark conditions. The cone cells enable us to see colour and detail and are more closely grouped in the centre of the retina. Both types of cell may be affected by the disease. The retina slowly degenerates and loses its ability to transmit pictures to the brain. Night blindness is often an early symptom. With the loss of the rod function the field of vision is narrowed so that retinitis pigmentosa is sometimes called "tunnel vision".

## **X-Linked Retinitis Pigmentosa**

Is transmitted by female carriers to their sons. There is a 50% risk to a male child

## **Linked Retinitis Pigmentosa**

Is transmitted by female carriers to their sons. There is a 50% risk to a male child

In some cases central vision deteriorates first. In other cases vision fades between the centre and the edge of the retina. This fading area gradually grows larger until little or no vision remains. The rate of deterioration varies between individuals. Most people retain limited residual vision. Some people can have other related problems. It is not uncommon amongst people who are born deaf. There is no treatment at present to either arrest or slow down the rate of deterioration or cure it.

Retinitis pigmentosa can affect anyone who has inherited the faulty gene and symptoms may occur at any age. Usually they become apparent in young adults but often children and babies are affected.

Retinitis Pigmentosa is an inherited condition. They fall into three main groups depending on the mode of transmission.

### **Recessive Retinitis Pigmentosa**

This can strike without warning in a family with no previous history of blindness and can affect members of either sex. It is estimated that one person in 80 carries the gene for Recessive Retinitis Pigmentosa and at present there is no way to detect these carriers.

### **Dominant Retinitis Pigmentosa**

Usually appears in every generation of a family. A person with this type has a 50% risk of having an affected child with each birth.

Significant advances have, however, been made identifying various types of defective genes responsible for different kinds of Retinitis Pigmentosa (including the type where cases occur in every generation of a family) and isolated their site. It is expected that the majority of genes responsible for Retinitis Pigmentosa will have been identified and located within the next 3-4 years and the mechanism by which the rod cells die will be understood.

The first steps have therefore been taken in finding a treatment. In the future it may well be possible to take a blood sample from a pregnant mother and diagnose whether the child has a Retinitis Pigmentosa gene and insert corrected genetic material into the embryo so that the child never develops this condition.