CURE BLINDNESS
AUSTRALIA

RETINA PIGMENTOSA

WHAT IS RETINITIS PIGMENTOSA?

Retinitis Pigmentosa is the name of a group of retinal dystrophies that cause degeneration of the retina of the eye. Retinitis pigmentosa is a disease of the eye that the affected individual is born with. The word “retinitis” derives from “retina” (a part of the eye) and “itis” (a disease). It is a disease of the retina, though not a contagious one. The word “pigmentosa” refers to an associated discoloration of the retina, which becomes visible to an eye physician on examination. For those people who find retinitis pigmentosa a difficult term to use, the shortened form “RP” serves as a simpler alternative.

WHAT IS THE RETINA?

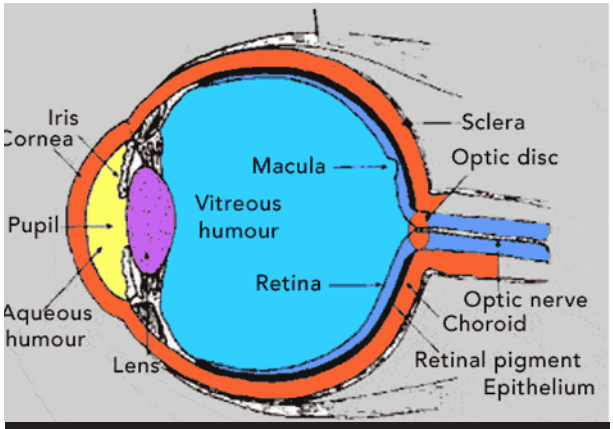
The retina is located at the back of the eye and is connected to the brain. It is made up of many millions of light-sensitive cells known as photoreceptor cells. These photoreceptor cells have the vital function of transmitting electrical impulses to the brain to enable seeing to take place.

Retinal dystrophies are caused by the gradual breakdown of these photoreceptors. Therefore it is important to understand the structure of the eye (as well as the ear in Usher Syndrome).

STRUCTURE AND FUNCTION OF THE EYE

The eye consists of several parts that resemble a camera:

- sclera - the eye’s white outer protective coat, normally seen as the “white of the eye”.
- cornea - the transparent, curved structure at the front of the eye.
- iris - the coloured part of the eye - blue, brown, green, grey etc - that can be seen through the cornea.
- pupil - the black part of the eye in the middle of the iris. It constricts or dilates according to the amount of light passing through it.
- lens - the transparent disc (with both sides being convex) immediately behind the iris and pupil.
- aqueous humour - the transparent fluid (with consistency similar to water) that circulates behind the cornea and in front of the lens.
- vitreous humour - the material (like transparent jelly) that fills the eyeball between the lens and the retina.
- retina - the light-sensitive layer of millions of nerve cells that line the back of the eyeball. The cells consist of two main groups, called rods and cones due to their appearance under the microscope.



EYE ANATOMY AND PHYSIOLOGY

- rods - more numerous, spread out over the entire retina with more toward outer edge, respond to low levels of light.
- cones - far fewer, concentrated around the retina’s centre, respond to colour and to details.
- macula - the small centre of the retina, responsible for reading vision.
- retinal pigment epithelium - This is a dark coloured layer of cells at the back of the retina responsible for providing oxygen and other nutrients to the rods and cones.

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FUNCTON OF THE EYE

When you see an object, the light travels from that object to the cornea, then passes through the aqueous humour, pupil, lens and vitreous humour to reach the retina. During this passage, the light becomes focused onto the macula.

At the macula, the light causes chemical reactions in the cones, that consequently send electrical messages from the eye to the brain. The brain recognises these messages and indicates to you that this particular object has been seen. The cones are therefore responsible for you being able to recognise colours and to read.

The rods are essential for you to see in the dark, and to detect objects to the sides, above and below the object on which you are directly focused. This function prevents you from bumping into obstacles when moving around.

All the retinal cells (rods and cones) are provided with oxygen and other nutrients from the retinal pigment cells (epithelium), which are kept supplied by the rich network of blood vessels in the choroid.

WHAT IS RESPONSIBLE FOR RP?

It is thought that one child is born with RP in approximately every 3,000 births in Australia. It is important to recognise that it is no one's fault and that RP can strike in a family with no known history of it. In fact, RP results from an imperfection in a tiny gene that causes an incorrect protein to be supplied to the retina. Over time this causes photoreceptor cells to die and progressive loss of vision results.

SYMPTOMS

Typical symptoms of RP are what are commonly referred to as "night blindness" and "tunnel vision". Night blindness refers to a reduced ability to see at night time, but more than that it entails a diminishing ability to see in dimly lit conditions generally, as well as in judging changes in level such as steps and gutters. Adjustments to changes in light intensity are typically slower and sensitivity to glare greater than is the case with the normal, healthy eye. All this results from the eye having fewer and fewer photoreceptor cells with which to transmit visual images to the brain.

The common pattern with RP is for photoreceptor cells to be lost progressively from the outer edges of the retina. This causes a progressive loss of peripheral vision, meaning side, upper and lower vision. It is as if the affected individual is viewing the world through a narrowing tunnel. Unlike a person with a normal field of vision, the person with tunnel vision cannot simultaneously look ahead and downwards or sideways without using a scanning technique by moving the eyes.

It is important to understand that with RP there is no uniform age of onset of symptoms and no uniform rate and extent of vision loss. These can vary markedly from individual to individual and are not usually able to be predicted.

However, the central vision of the person with RP may remain largely unaffected for a considerable number of years, enabling the individual to continue to read, watch television and perform other visually detailed tasks.

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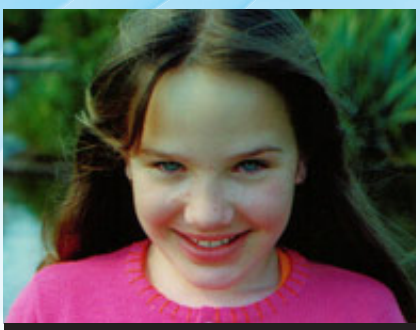
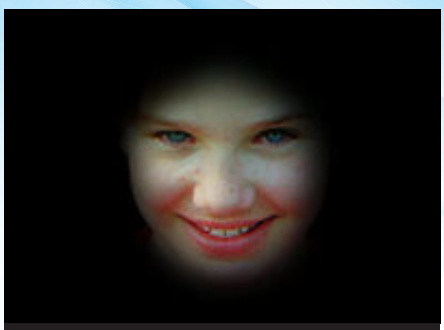


When Richelle's family look at her they see her like this



When Richelle looks at herself in the mirror this is what she sees

TREATMENT

At present, there is no treatment to halt the progression of any retinal dystrophies. No convincing scientific evidence demonstrates any benefits for individuals with RP from the many unproven treatments that have been attempted. However scientists investigate even the most remote possibilities.

There is no scientific evidence that normal light levels worsen the symptoms. However, individuals with retinal degenerations are encouraged to protect their eyes from long exposure to bright light as a precaution. In dim light, they may use their eyes to the degree possible without fear of worsening their condition.

When only one person in the family is affected, it is difficult to decide how the condition is inherited. Many hereditary diseases may affect the retina, each with symptoms. Conditions include Retinitis Pigmentosa (RP), Stargardt disease, Usher Syndrome, Best disease, Cone Rod Dystrophy and Choroideremia.

In order to determine the likelihood of your children and other members of your family being affected, you should consult your doctor or seek genetic counselling. Because retinal dystrophies usually run in families, all family members are encouraged to have a thorough eye examination.Professional genetic counselling is generally advisable.

CATARACTS AND RP

It is not unusual for people with RP to develop a cataract, a haziness of the lens of the eye.
When cataracts significantly interfere with vision, it may be advisable to remove them surgically.
Whether surgery improves vision often depends on how far the retinal changes have advanced.
The usefulness and implications of surgery should be discussed with an ophthalmologist.

GENETIC INHERITANCE

Whereas other genetically transmitted diseases are the result of a single gene defect, RP results from a large and as yet unknown number of gene defects. So far around a hundred gene defects have been found to cause RP.

However, there are three main genetic pathways through which a child can come to be born with RP and the probability of this happening varies according to which of the genetic pathways applies.

Two of them are usually identifiable through a recurrent family history of RP but the third and by far the most prevalent one is not.that the child will be affected. Males and females are equally affected.

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GENETIC INHERITANCE

In simple terms RP is inherited in different ways:-

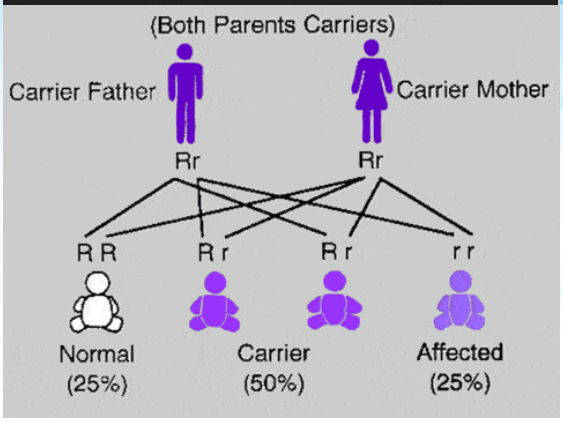
Autosomal Recessive Inheritance Pattern

This is the most common form of RP. This form means each parent is a carrier (have the gene present, but DNA testing is still unavailable) but is not affected by the disease themselves. Each of their children will have a 25% chance of being affected, with males and females having an equal chance of being affected.

The diagram indicates how the recessive genes are passed from the two carrier parents to their children.

The mutant RP gene is represented by “r” and the normal gene by “R”.

AUTOSOMAL RECESSIVE INHERITANCE

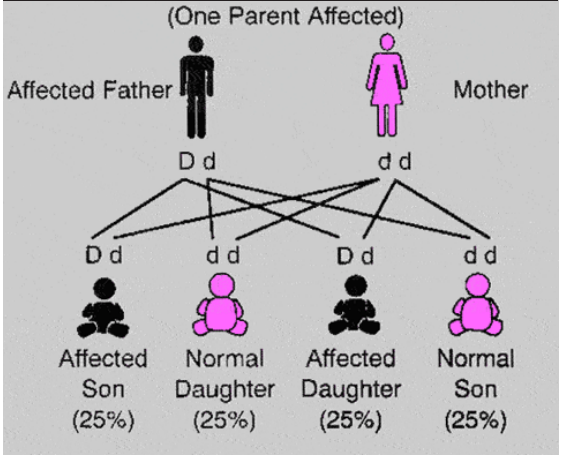


Autosomal Dominant Inheritance Pattern

In this type of RP, one parent is affected and each pregnancy has a 50% chance that the child will be affected. Males and females are equally affected.

The diagram indicates the four possible combinations of genetic information that may be passed on by the parents. The mutant dominant gene is represented by “D” and the normal gene by “d”.

AUTOSOMAL DOMINANT INHERITANCE



X-linked Recessive Inheritance Pattern

In this type of RP, one parent is affected and each pregnancy has a 50% chance that the child will be affected. Males and females are equally affected.

The diagram indicates the four possible combinations of genetic information that may be passed on by the parents. The mutant dominant gene is represented by “D” and the normal gene by “d”.

X-LINKED RECESSIVE INHERITANCE