



What is Retinitis Pigmentosa?

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Prepared by Retina Australia (Qld) Inc
Updated 2010

What is Retinitis Pigmentosa? is one of a series of booklets prepared by Retina Australia (Qld) Inc. to provide information about the most common inherited retinal dystrophies – retinitis pigmentosa, Stargardt disease, Usher syndrome and macular degeneration.

Throughout the booklets the pronoun ‘you’ is used wherever grammatically appropriate. This is to highlight that the series has been written primarily for the individual who may be facing a diagnosis of retinitis pigmentosa.

During the writing process there has been ongoing consultation with professionals who work in the field of eye disease and vision loss, persons with the particular conditions and family members. The booklets’ typeface conforms to the large print guidelines for vision impaired people produced by Blind Citizens Australia. The booklets can be also accessed at www.retinaqld.org.au. Audio versions also are available.

Retina Australia is grateful to Queensland Health (through the Statewide Health and Community Services Branch) for making a grant available to support the original development of the booklets.

Our thanks also go to Dr John Vance for his work in the preparation of the original and revised editions, to Dr Mark Donaldson MBBS (Hons) FRANZCO for his contribution to the revised edition and to Dr Darren le Broque who helped format the original booklet editions. Also to Nayomia Stibbe and Anne Housego for their work in preparation of the booklets. We also are grateful for the editorial assistance provided by Dr Rowan Porter (consultant ophthalmologist), Associate Professor Jan Lovie-Kitchin (QUT Department of Optometry) and Mrs Robyn Richards RA (NSW).

Relevant contact details, including those for each Retina Australia state or territory organisation, as well as the national body, are listed under Contacts at the back of this booklet.

Sources of information are acknowledged by way of a letter in parenthesis [e.g. (a), (b) etc] which matches a letter on the acknowledgement page at the back of this booklet

Structure and Function of the Eye

Introduction

Before discussing the condition of retinitis pigmentosa, it is important that you understand the structure and function of the eye.

Structure

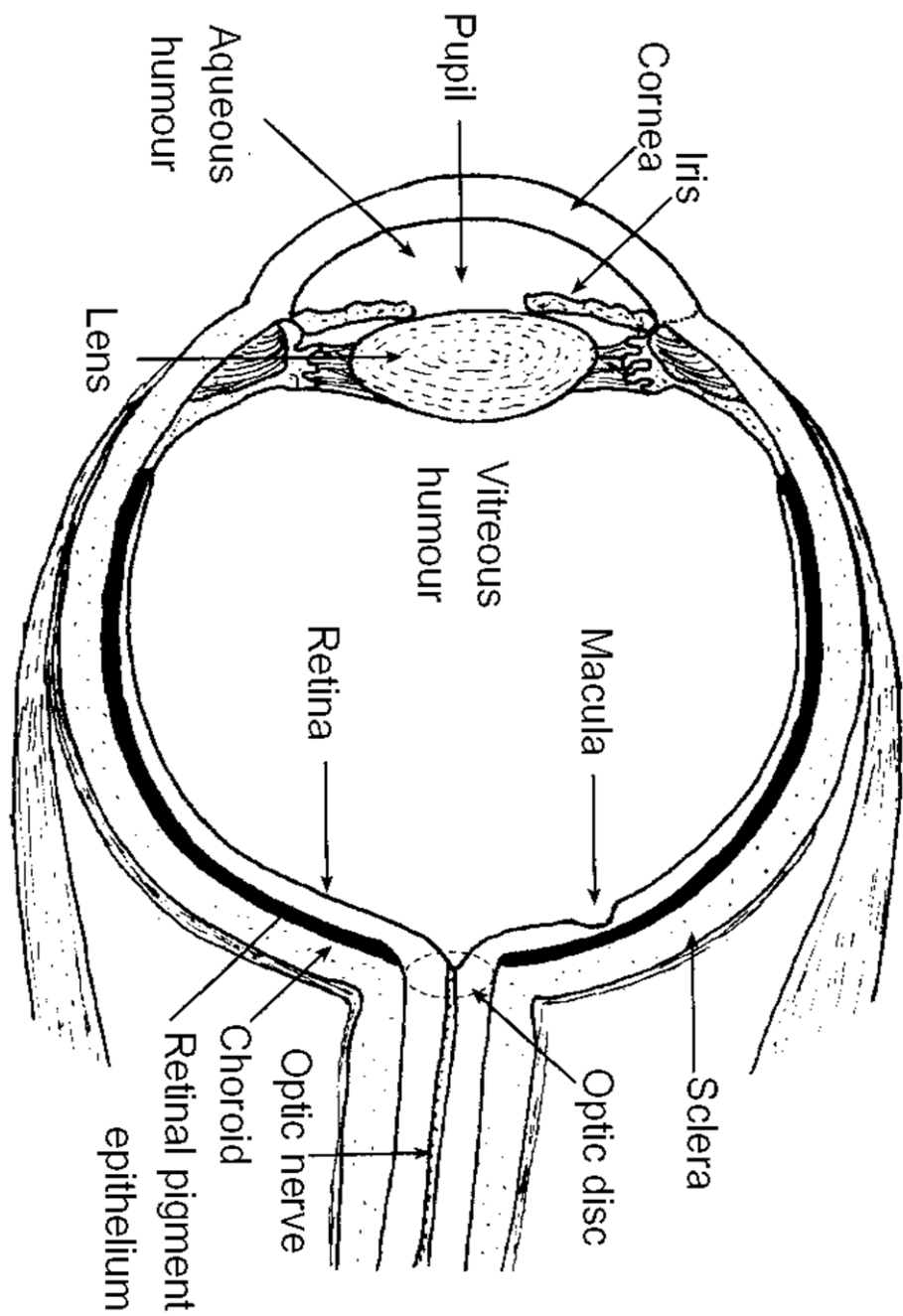
The eye consists of several parts which are somewhat similar to a camera (see diagram P.7).

- **sclera** – the white material which we normally see as the white of the eye. It is the eye's outer protective coat.
- **Cornea** – the transparent, curved structure at the front of the eye
- **iris** – through the cornea can be seen the iris, which is that part of the eye which gives it colour – blue, brown, green, grey etc. It is a circular muscle which responds to bright light by contracting, making the pupil smaller.
- **pupil** – in the middle of the iris is the pupil, a transparent aperture which appears black.

The pupil constricts in response to bright light and dilates in low levels of light.

- **lens** – situated immediately behind the iris and pupil is a transparent disc with both sides being convex. The lens focuses light onto the retina. It divides the eye into two parts – anterior and posterior chambers.
- **anterior chamber** – a fluid with the consistency of water circulates around this chamber. This fluid is called the **aqueous humour**.
- **posterior chamber** – this is filled by the **vitreous humour** which has the appearance of transparent jelly.
- **retina** – situated at the back of the eye. It consists of millions of nerve cells which are divided into two main groups – rods and cones. They are so described because of their appearance under a microscope.
- **cones** – concentrated around an area of the retina called the **macula**.

- **rods** – although some are placed near the macula, the majority of rods are spread out to cover the rest of the retina.
- **retinal pigment epithelium** – a dark coloured layer of cells underlying the retina responsible for providing oxygen and other nutrients to the rods and cones.
- **choroid** – located behind the retina, it consists of a large network of blood vessels which transport oxygen and other nutrients to the retinal pigment cells.
- **optic disc** – the nerve cell connections from all the rods and cones travel to the optic disc, a small yellow oval structure which can be seen through an ophthalmoscope (see P.13).
- **optic nerve and beyond** – the optic disc is the front part of the optic nerve which passes from the eye to make connections throughout the brain.



Function

When you look at an object, the light travels from that object initially to your cornea. The light then passes through the aqueous humour, pupil, lens and vitreous humour to reach your retina. During this passage, most of the light becomes focused on your macula while the remainder reaches the more peripheral part of your retina.

At your macula, the light causes chemical reactions in the cones, which consequently send messages from your eye down the optic nerve to the brain. Your brain then recognises these messages and indicates to you that you have seen the particular object. Only cone cells are capable of distinguishing fine detail, such as is required for complex visual tasks including reading print. Your cones are also responsible for your being able to recognise colours. Cones require good lighting to function properly.

The light which strikes the periphery of your retina makes contact with the rods. Rods are essential in enabling us to see in very low levels of illumination. Likewise they provide peripheral vision. When fully functioning they detect

movement and objects to the sides, above and below the object on which one is directly focused. This function alerts us to movement around us and prevents us from bumping into obstacles when we move around.

Retinal cells are provided with oxygen and other nutrients from the retinal pigment cells which are kept supplied by the rich blood circulation through the choroid.

How is Vision Tested?

There are many procedures to test different visual functions. Only those which are relevant to retinal disease will be discussed here. Some tests can be carried out by professionals such as nurses, special education teachers or general practitioners, while others may require more specialist knowledge or equipment such as an optometrist or ophthalmologist may possess.

Visual acuity

Visual acuity is a measure of how clear visual images are and thus how much detail can be discriminated. A test of visual acuity is therefore essentially a test of macular or cone function.

Visual acuity is tested using a Snellen chart which has lines of random letters which get smaller with each line. You are seated 6 metres from the chart and are then asked to read from the top line down. The size of the letters on the bottom line is designed so that a person with normal vision will be able to distinguish between them. Thus, on the Snellen scale a person sitting 6 metres from the chart who can successfully read the bottom line has vision described as 6/6 – that is normal vision.

The letter sizes of the other lines (working from bottom to top) are such that an individual with normal vision could distinguish between these letters at a distance of 9, 12, 18, 24, 36 and 60 metres respectively. If you have decreased visual acuity, and you are seated at 6 metres from the chart, you may only be able to read the larger letters. Hence your visual acuity may be described as 6/9, 6/12, 6/18, 6/24, 6/36, or if only the top line

can be read, 6/60. (In Australia this is the cut-off point for legal blindness – see P.14)

If you are unable to distinguish the top line of letters you may be asked to move closer to the chart, perhaps to 3 metres or 1 metre away. If you can read the top line at 3 metres your visual acuity is described as 3/60; or from 1 metre, 1/60.

If your visual acuity is less than 1/60 you may be asked to detect whether there is or is not an object present in your field of vision. If you can do this (without having to identify it) it is said that you have 'form perception'. If no form perception is present then a nearby torch may be switched on or off while you sit in a darkened room. If you can detect this then you are said to have 'light perception only'.

While diminished visual acuity may reflect abnormalities of the eye, it is important to realize that it can also reflect abnormalities of the visual pathways to the brain or the brain itself.

Visual Fields

Visual field testing assesses the functioning of the peripheral retina. Field testing can be carried out in a number of informal and formal ways. In all cases the test revolves around you focussing on a central point and then being asked to detect an object being introduced from the periphery of your vision. For example, a formal test might involve asking you to focus on a small white point on a dark screen. A small light may be then moved in from outside your line of sight and you are asked to say when you first see the light. This procedure is repeated several times to the right, left, above and below each eye. Your results are then marked on a chart and compared with what a person with normal vision would be expected to see. A normal visual field is described as being about 140° in every direction.

Decreases in visual fields may also reflect abnormalities of the eye or the visual pathways to the brain or the brain itself.

Ophthalmoscopy

The ophthalmoscope is used to examine the internal structures of the eye, including to detect changes in the lens or retina due to various disease processes.

What is “legal blindness”?

In Australia a person with a permanent visual impairment is classed as legally blind according to the following guidelines:

- visual acuity on the Snellen Scale, after correction by suitable lenses, must be less than 6/60 in both eyes, **or**
- visual field constriction must be to within 10 degrees of fixation in the better eye, irrespective of corrected visual acuity, **or**
- a combination of visual defects resulting in the same degree of visual impairment as that occurring in either of the above points.

Legal blindness entitles a person to a Disability Support Pension – Blind; or an Aged Pension – Blind. Neither is means tested. (a)

What is Retinitis Pigmentosa (RP)?

RP is the name given to a group of hereditary diseases which cause progressive damage to the retina. The rods, and occasionally the cones or retinal pigment epithelium begin to degenerate, or die, and this causes the person's vision to diminish. There is currently no effective medical treatment.

When optometrists or ophthalmologists use an ophthalmoscope to look into the eye of a person with RP, they usually see scattered areas of pigment across the retina. By contrast in a normally sighted person, the pigmented cells are covered by the living nerve cells – the rods and cones. The pigmented appearance gives rise to the name – retinitis pigmentosa.

What are the symptoms of RP?

Age of Onset

Symptoms commonly start in the late teens or early twenties (sometimes even in childhood). The symptoms of RP become more severe with age. When other members of the family are also affected, the rate of progression is usually similar

within the family. It is not uncommon for individuals to have to significantly modify life arrangements (eg. work; hobbies) by about age 40, due to the gradual deterioration of their vision.

Night Blindness

One of the earliest symptoms of RP is difficulty seeing at night or in dimly lit places. However, night blindness is not always disease related and certainly it can be a symptom of a number of diseases other than RP. Clearly, it is very important to seek medical advice.

Decreased visual fields

As individuals with RP increase in age, they experience a reduction in visual fields (peripheral vision). Initially they may simply appear to become increasingly clumsy, bumping into people and objects they fail to see. Perhaps they will have some difficulty keeping their place when reading.

The impact of ever reducing visual fields may be best appreciated by way of two examples, one relating to relatively near vision and the other to distance vision. When looking at the floor from a

height of 1.5 metres, a person with 10 degrees of vision can see within a circle 26 centimetres across, whereas a person with 140 degrees of vision can see within a circle about 2 metres across. This can certainly make a big difference when looking for an object dropped on the floor! (b)

The second example relates to driving. If a typical two-lane road is taken to be 12 metres wide, a person with 10 degrees of vision can see from one side of the road to the other only by looking 70 metres down the road (without scanning from side to side). By comparison, a person with 140 degrees of vision can see from one side of the road to the other at only 10.5 metres. For the person with 10 degrees of vision travelling at 60kph, a period of 4 seconds will elapse between an object (eg. a clear intersection) being seen (70 metres away) and the person with RP actually reaching that intersection, during which time a car could have entered the intersection!

Macular changes

On **rare** occasions, the cells of the macula also die which leads to compromised central vision - visual acuity may be reduced to the point where letters can no longer be distinguished. Colour identification may be also lost.

Cataracts

While the actual incidence is not known, it has been observed that individuals with RP are more likely to develop cataracts than are members of the general population. (i.e. the normally clear lens becomes opaque. This will reduce visual acuity, particularly in bright light. The opaque lens can be surgically removed and replaced by a clear plastic lens. However the extent to which surgery improves vision depends on a number of factors including how far retinal changes have advanced. This is a matter which needs to be discussed in detail with an ophthalmologist.

Variations in everyday functioning

Glare

You may have noticed that your vision is better at some times than others. There are a number of factors which might account for this. Firstly if you have cataracts you will have almost certainly found that you are particularly sensitive to glare and bright light. (Cataracts act like a dirty windscreen – when you are facing away from the light the windscreen seems clear; but when you turn into the light the windscreen suddenly seems very dirty and uncomfortable to look through).

Even if you do not have cataracts you may find glare a problem – most individuals with RP find this, although it is not clear exactly why this is so. Indeed even subtle changes in light conditions affect individuals differently. Some individuals with RP feel they are able to see better on cloudy days, while others feel they do not see as well.

It is also important to identify that all people function less well when fatigued, ill or under emotional stress, and that this is most apparent when the task requires maximum effort for optimum performance.

Finally it is important to remember that all people have days when, for no apparent reason, they seem to function more effectively.

What causes RP?

In normally sighted people, the rods and cones die off very slowly so that only some elderly people have difficulty in seeing. However, in people with RP, it is known that rods, in particular, and, to a lesser extent, cones, die at a much faster rate. The cause of this accelerated cell death is not known at present. It is clear, however, that genetic changes result in either the presence of abnormal proteins or enzymes within the retina and that visual fields are affected more than the visual acuity.

There are hundreds of different genetic variations, which explains why every family is different.

Diagnosis

The optometrist or ophthalmologist will usually make the diagnosis of RP on the basis of

- Symptoms of night blindness and visual field loss
- Examination of the eye
- Family history
- Special tests such as visual acuity, visual fields and ERG.

In the future, genetic blood tests may become more easily available.

Is it only vision which is affected in people with RP?

Some people with RP are also born with hearing impairment (Usher syndrome). There is another booklet available which discusses the specific issues of this condition. There are also a number of other conditions, such as Refsum syndrome and Bardet-Biedl syndrome, which may occur in rare cases of RP. Clearly in this situation advice may need to be sought from a number of medical specialists, including geneticists.

Can RP cause blindness?

Blindness (or “Black Blind”), to the layman, indicates a complete loss of all sight. Fortunately this is very rare, and, although some people with RP become blind in this sense, most will retain at least limited vision. Depending on the degree of this limitation, the person may, or may not be legally blind.

How quickly does a person’s vision diminish?

Each individual must be evaluated by an optometrist or ophthalmologist, because the progression of RP is so variable. Most people will have a gradual progression of symptoms. Occasionally however, there may be a sudden deterioration.

Do any levels of light have an adverse effect on RP?

There is no scientific evidence that normal light levels worsen the symptoms of RP. Individuals with RP and other retinal degenerations are encouraged to protect their eyes from long exposure to bright light as a precaution until more

is learnt. In dim light, eyes may be used to the degree possible without fear of worsening the condition.

How common is RP?

RP affects about 10,000 people in Australia. In various overseas studies, it has been found to occur in approximately 1 in 3000 people. It occurs in all ethnic groupings.

How is RP inherited?

RP is always considered an inherited disease and hence the determination of a family history is very important.

There are five methods of inheritance. The three major forms - autosomal recessive, autosomal dominant and X-linked recessive will be discussed in detail. The two rare forms of inheritance – digenic and mitochondrial - will be noted briefly.

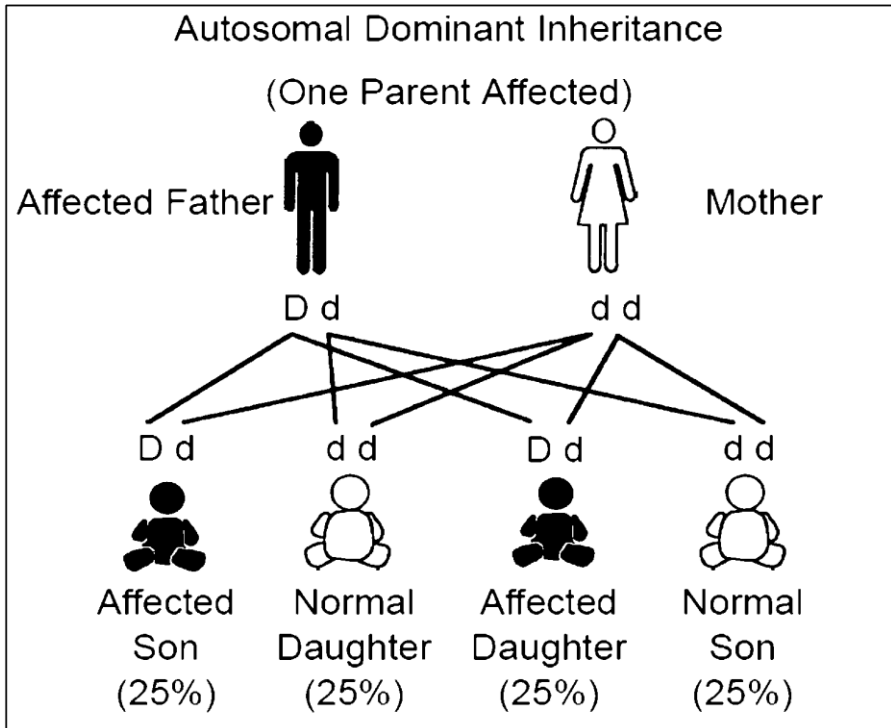
Within the major forms of inheritance, there are many possible single genes which produce the disease. Over 100 different pedigrees have been identified and the actual genes causing these abnormalities have been isolated in nearly 40.

In a significant proportion of families, only one person can be identified as having RP. Thus, without specific gene identification (currently primarily a research tool), the inheritance of RP in that person cannot be identified with certainty. This situation is defined as isolated (or simplex) RP. As more tests become available, fewer people will be allocated to this category.

The following diagrams demonstrate how the various forms of inheritance take place.

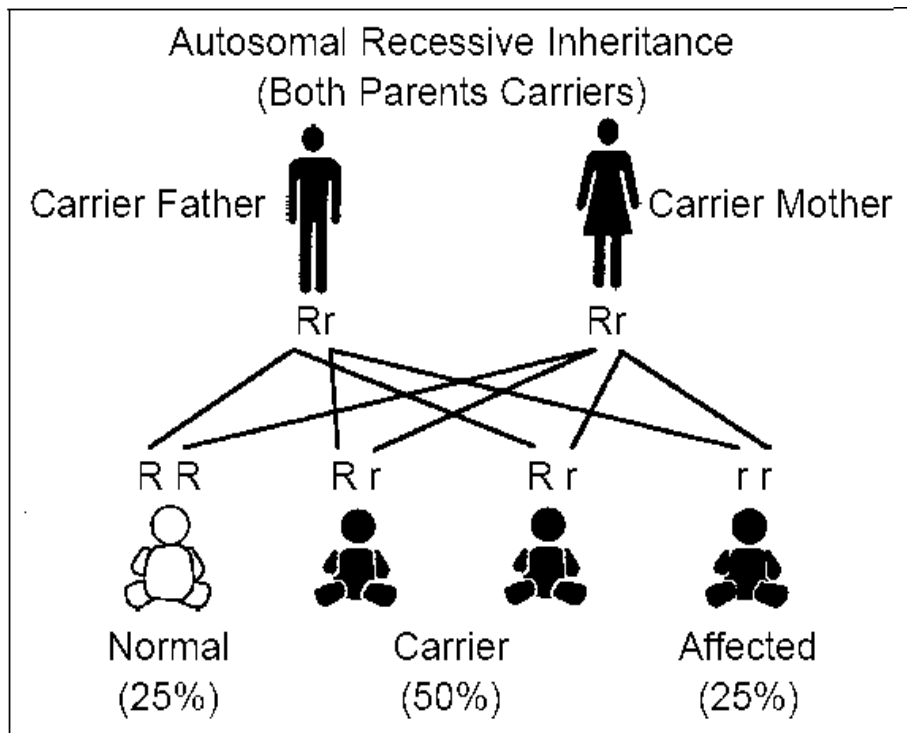
Autosomal Dominant Inheritance

One parent is affected and, for each pregnancy, there is a 50% chance (1 in 2) that the child will be affected. Males and females are equally affected. The diagram shows how the dominant gene “D” is passed from the affected father to half his children.



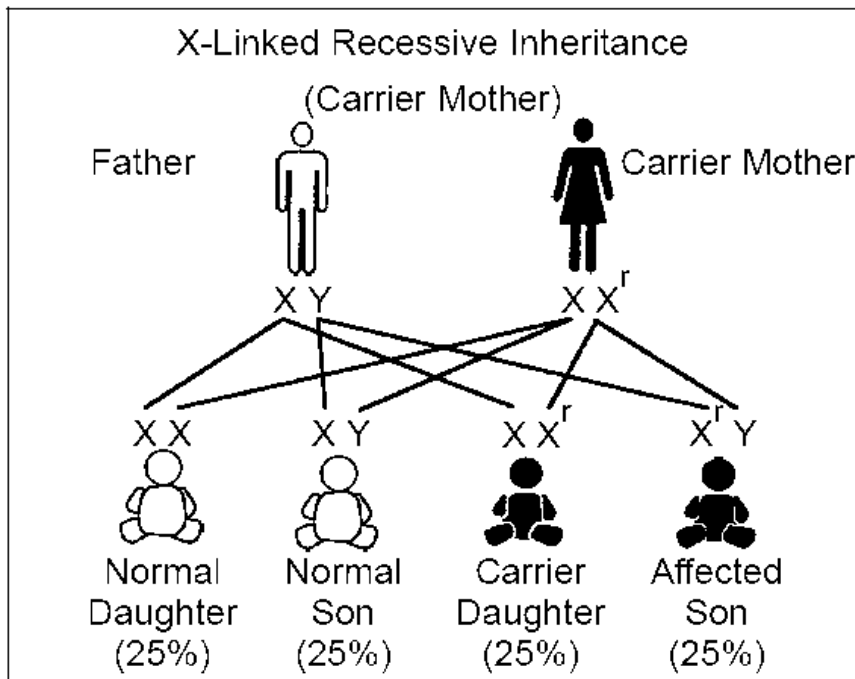
Autosomal Recessive Inheritance

Each parent (carrier) will need to carry the abnormal gene for one of their children to acquire RP. Carriers are not usually affected by the abnormal gene. Each child (male or female) will have a 25% chance (1 in 4) of being affected. The diagram shows how the recessive gene “r” is transmitted from both parents to 25% of their offspring.

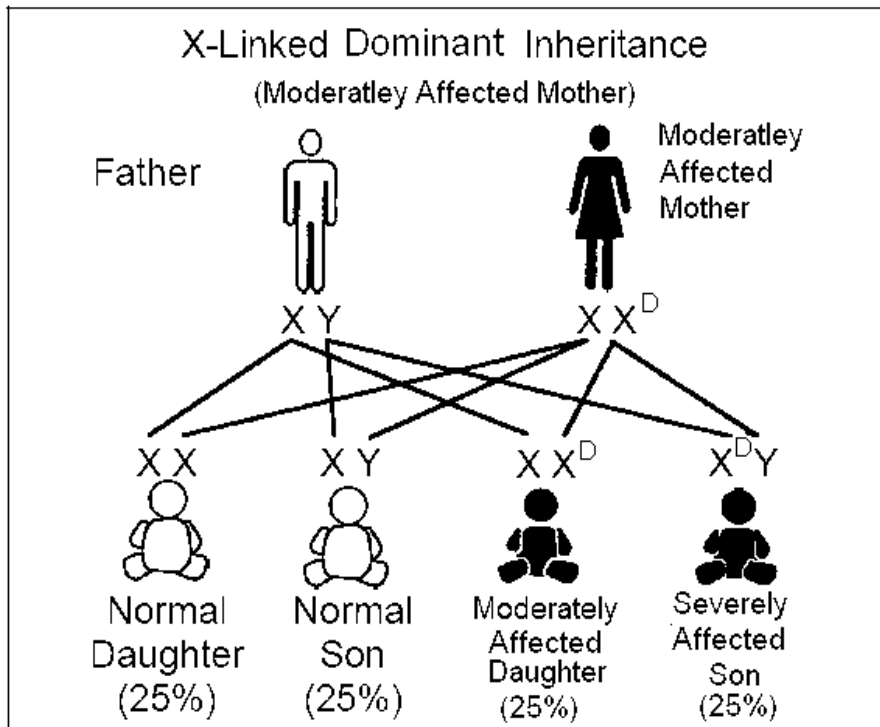


X-Linked Inheritance

This can be either recessive or dominant. In “X-linked” recessive inheritance, the mother is the carrier. If the father does not have RP, there is a 50% chance (1 in 2) that each of her sons will have RP. There is a 50% chance (1 in 2) that her daughters will be carriers. The diagram shows how the X-linked recessive gene “X^r” is passed from the carrier mother to her children.



In “X-linked” dominant inheritance, the mother will usually have mild to moderate symptoms of RP. There is 50% chance (1 in 2) that each of her sons will have RP and they will be more severely affected than their mother. There is a 50% chance (1 in 2) that each of her daughters will also be affected at about the same level of severity as the mother. The diagram shows how the X-linked dominant gene “X^D” passed from the mother to daughter and son.



Digenic Inheritance

This is a rare phenomenon where each parent is autosomal recessive for a different gene which has the ability to produce RP. Each child then has a 25 % chance (1 in 4) of having both these genes and hence produce a unique form of RP.

Mitochondrial disorders

Mitochondria are small parts of the cells which may have DNA present. Changes in DNA can cause a range of conditions which may include RP. This is a very rare situation and clearly the help of geneticists and other specialists is then essential.

It is clear from the complicated nature of the inheritance of RP, that it is important to seek expert genetic advice. This can be obtained through the clinical genetic services of each state and referral can be made through your general practitioner, ophthalmologist or optometrist.

Treatment

For most people, there is no treatment available at the present state of knowledge. However, there are some rare conditions for which treatment may be helpful. It is beyond the scope of this booklet to discuss these further, but if RP is associated with other symptoms, clearly a specialist consultation is necessary.

Vitamin A

Vitamin A was proposed as a cure for RP in the early 90's. It has become clear that some people with RP definitely benefit from this, but they are probably in a minority. Before taking vitamin A, it is important to seek medical advice and have regular blood tests, as too much can cause serious complications. Vitamin A should not be used in children under the age of 18 or in those suffering Stargardt disease.

Sunglasses

Glare is a major problem for many with RP. It has been shown that sunglasses which contain lenses that specifically block out the blue component of

light, are effective in reducing glare and may be very beneficial. (1)

It is important to discuss this with optometrist to try various types of lenses and to determine which is best for the individual. The spectacles are commonly called “blue block” sunglasses.

Maintaining Independence

Low Vision Devices

Does a progressive loss of vision mean that people will become increasingly dependent? The vast majority can maintain their independence if they maximise their surrounding support systems. Many devices, services and techniques are available which provide a person with a vision impairment increased mobility and independence. Special lenses, orientation and mobility training and illumination are some examples. The ability to

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1. C.E. Reme. Recommendations of Protective Eyewear for Patients Suffering from Degenerative Retinal Diseases. Laboratory of Retinal Cell Biology, University Eye Clinic, Zurich, Switzerland. (2001).

function independently can be learned just as speaking, writing and walking. The local agencies for people with a vision disability will give aid and advice on all aspects of daily living. Retina Australia in your state has a list of agencies which may help any particular needs.

Low vision devices help people obtain the most use of their remaining vision. They may be optical lenses, magnifiers and telescopes, or non-optical devices such as lamps, large print and digital recorders. Clearly, new computer products are being developed on a regular basis and some can be very helpful. To determine which aids will assist you, it may be necessary to obtain a thorough low vision evaluation from a specialist in the field. Most states have Low Vision Clinics with specialized staff such as optometrists, occupational therapists, specially trained nurses and orientation and mobility instructors. Information about these clinics can be obtained from the Retina Australia office in your state.

Orientation and Mobility

It is clear that a key component of independence is one's ability to move around with as little help as possible. Driving - People with RP are usually able to drive in the early stage of the disease. However, once their visual fields have decreased, it may be important to consider giving up driving. Clearly, this is not an easy decision and should be made in consultation with family, friends, general practitioners, optometrists or ophthalmologists. It should be pointed out that planning to use free public transport and taxis (half priced if on a blind pension) can be a reasonably effective alternative. The Federal Government has published a booklet which gives advice to optometrists, general practitioners and ophthalmologists regarding this.

(1)

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1. Assessing Fitness to Drive – guidelines and Standards to Health Professionals in Australia – 2nd Edition. 2002. Austroads.

White Cane Training

Independence, orientation and mobility may be enhanced by white cane training. This is a skill taught by specially trained instructors from the Guide Dogs Association in each state. Individual training will help people to be orientated at their home, work and to explore transport options and recreational facilities.

Dog guides

These can also be used to increase orientation and mobility, but negotiation, planning and training needs to take place with the local Guide Dog association.

Why should I continue to see my optometrist or ophthalmologist?

When the diagnosis of RP is made, people are often told that there is nothing that can be done to cure the disease. While this may be true at our present state of knowledge, there are many factors which need to be considered to ensure the optimum use of your remaining vision. Visits to the

optometrist or ophthalmologist may therefore continue to be important for the following reasons:

Follow up appointments

A follow up appointment is important to answer questions that arise once the shock of the initial diagnosis has passed.

Obtaining a second opinion

It is common when one receives the devastating news that you have an incurable eye disease, to be very angry and to not believe the person who has given you this news. It is perfectly reasonable to seek another opinion to ensure the correct diagnosis, and to reassure you that all avenues of history, examinations and tests have been undertaken.

Monitor the progression of the disease

Even though there is no cure, understanding the progression of the disease may help in planning lifestyle changes, such as work changes. As the condition slowly progresses in most cases, it will

be necessary to upgrade visual devices and supports.

Prescription of spectacles to help improve residual vision

In people with normal vision, visual acuity and reading ability change with age and spectacles are often required. The same situation occurs in people with RP as they age and it is important that optimal magnification is achieved. Prescription of sunglasses to reduce glare may also be important.

Referral for the Disability Support Pension (DSP - Blind) when sight deteriorates further.

Although being declared legally blind can have some negative consequences, the DSP (Blind) is not means tested and attracts other features, such as free train and bus travel, half fare taxi vouchers and help with rates and other service bills.

Assessment of other family members

The recognition or exclusion of RP in other family members is important in determining the inheritance pattern and ensuring that all family

members have optimum access to diagnosis and support.

Referral to Low Vision Clinic or orientation and mobility support

This has been referred to in previous sections.

Referral to local support agencies such as Retina Australia

Information is provided about these agencies later in this booklet.

Diagnosis and removal (when necessary) of cataracts.

Cataracts commonly occur in RP and further interfere with already impaired vision. Removal of the lens with the cataract and replacement with a plastic lens can thus improve residual vision.

Diagnosis of other eye conditions which may make vision worse, such as glaucoma and diabetes.

There is no reason why these two diseases could

not develop as the person with RP ages.

Glaucoma is treatable, but if it is not detected early enough, may cause even more deterioration in vision. Diabetes can be controlled with diet and medication, and treatment may halt the progress of eye deterioration.

Living with RP

You may have been told recently perhaps, or you may have known for some years, that you have RP. This diagnosis may explain the months or years of not being able to see properly in the dark, of bumping your head, or stumbling and falling over objects you did not see.

Then you are told that, as yet, there is no cure for RP and that you have to face the prospect of slowly getting worse. What then? Slow loss of sight is a very difficult thing to live with, especially when you may not receive the immediate understanding offered to people with total loss of vision. Indeed, many people will not believe that you have a problem because you have no apparent signs of a vision disability.

The first and hardest step towards living positively with a vision impairment is accepting it. For people

with RP, that means knowing the extent and limits of your vision and using intelligently the visual clues you receive. A normally sighted person must do this too, in certain circumstances. The driver who plunges into a fog has two alternatives — he can decide that he cannot see a thing, panic and stop, which might cause an accident. Or he can see (even if it is only the nearside curb) and move cautiously down the road. The person with RP can panic or move down the road.

Accepting that you have an impairment of vision is never easy. You may go through times of despair and of feeling resentful and bewildered. All these reactions are quite understandable, especially as the very nature of RP makes adjustment difficult. But the way in which you feel about RP will determine the type of life you and your family will share from day to day. Try to ignore it and you will experience constant reminders that it is there. Write yourself off as totally incapable and you will be missing out on many of life's enriching experiences. Tackle each new problem steadily as it arises, using clues from all your senses — smell, touch, hearing and balance — and you will find that you can live more positively with RP.

It is common to experience emotional distress after receiving a diagnosis of RP or other retinal degeneration

It is not unusual to experience fear and confusion. Some people accept the situation more quickly than others; some experience a period of temporary depression before they can accept and adjust to the condition. In addition to depression, it is not uncommon to also feel denial, anger and frustration. Sometimes, just knowing that you are not alone can be helpful. Throughout the world, there are over 3 million people affected, and it is estimated that around 10,000 people in Australia have RP. You may wish to contact your local Retina Australia group where you will have the opportunity of meeting others and sharing your experiences.

What about employment?

Many people with RP can continue to lead productive lives and some may continue to pursue a desired career goal. As problems, or potential problems, are identified, they can often be solved by the use of mechanical aids, extra training or job modifications. Vocational counselling offered through educational institution, government

rehabilitation departments or agencies for people with vision disabilities, can be most beneficial in planning, changing or maintaining a career.

If a child is diagnosed as having a retinal degenerative condition such as RP, when should the parents tell her/him?

There are no guidelines for explaining to children about retinal degenerations, because they have varying needs for information depending on age and maturity. Although most are very perceptive and quickly sense when there is a problem, they may only be capable of absorbing a little information at a time. Answer questions as frankly and positively as possible, without offering more answers than your child has requested. Help them to realistically understand that some of their limitations may be due to their vision impairment, without using this condition as a crutch or excuse for not reaching a reasonable goal. It may be very helpful to discuss this with other parents of vision impaired children, who have usually had similar experiences.

Does pregnancy have an effect on RP?

There is no convincing evidence that pregnancy affects RP, but the matter requires further research.

Everyone is different

RP can manifest itself in different ways. For some, loss of vision is slow. There will be only slight loss, over perhaps ten years. Others may have periods of rapid loss, often with years in between of no apparent decline. Still others have been aware of impaired vision from childhood or teens, when they had difficulty with ball games, especially at dusk. Some will lose peripheral sight but will retain near perfect central vision, while others will develop blurred central vision.

With so much variation in the symptoms and effects, it is not surprising that the public find RP difficult to understand. If the person with RP can still read, can gaze into the distance and pick out landmarks and can respond to a smile, it is hard to appreciate that she/he may have an impairment of vision. It is most important for mutual understanding, especially within the family and workplace, that the normal sighted person should

recognise and comprehend the difficulties involved. For example, a person suffering from the early stages of RP may have almost perfect day vision, but at night or in brilliant sunshine, or in changing light conditions, that same person may react as if almost totally blind.

Equally difficult to understand is the loss of visual fields. As people with normal sight stare straight ahead, they can recognize objects on either side, above and below the point on which their eyes are fixed. This faculty is very useful, enabling people to be aware of and cope with, more than that which is directly in front, thus keeping them alert and free from danger. How often do we use the expression “I saw it out of the corner of my eye”? It can be appreciated, therefore, that losing the boundaries of one’s vision can be very alarming.

How can one help?

How can others best help people with RP? Don’t be over-protective but observe what they can do without help. Try to give warnings of hazards such as steps (and say whether they are going up or down), posts or overhanging trees. Within the home, listen to any suggestions about how to

arrange the house, such as keeping the floor clear of obstacles, not moving the furniture around, shutting cupboard doors etc.

If you are the one who has RP, explain what you can do without help. If the family is always tense and over-protective, this can be morale destroying. When helping your family to understand your vision impairment, use incidents that are particularly relevant to them: e.g. when you have just tripped over your son's football in the hall, make a point about putting things away (and try not to let it turn into a nagging match). Living with RP can be hard for all members of a family and the one who has the condition can do much towards making life happier by helping family and friends to understand.

Are there other retinal degenerations besides RP?

There are many other hereditary diseases which may affect the retina. The list is long and not all are mentioned here. Symptoms will also vary. Conditions include Stargardt disease (see companion booklet), Best disease, cone-rod dystrophy and choroideremia. Age-related macular

degeneration may, in some people, have a hereditary component (see companion booklet). Clearly, consultation with an ophthalmologist or optometrist is essential to ensure that the correct diagnosis is made. Referral to a geneticist may also be important. Retina Australia seeks to provide information and support for all hereditary retinal degenerations.

Groups for people with RP and similar conditions

The special difficulties caused by RP have led, in recent years, to the formation of RP groups in all mainland Australian states and territories and in many countries throughout the world. More recently, these groups have broadened their scope to embrace all hereditary retinal diseases and to some extent macular degeneration. Hence, the names of groups have changed to Retina Australia (Queensland), Retina Australia and Retina International. The state organisations within Australia are independent, self-help groups of people with RP and related conditions, their families and friends, whose aims are to assist people in coping with RP and to encourage research.

What services are offered by local Retina Australia organizations?

All state Retina Australia groups seek to foster support, information, awareness, advocacy, research and fundraising within their state. In practical terms, these goals –

- Seek all practical means to assist those affected and their families and to give such assistance as necessary and when requested.
- Disseminate information to professionals and non-professionals about RP and related conditions.
- Exchange information with each other and Retina International.
- Interact with Retina Australia in providing funds for research and biennial congresses.
- Ascertain the cause and means to cure or arrest the deterioration in RP and other retinal dystrophies.
- Interact with all other agencies who provide services for people with vision disabilities
- Provide advice to the government and wider community.

Retina Australia is responsible for distributing funds for research following recommendations of its Research Advisory Committee. It is also responsible for an Australian biennial congress. Contacts with Retina International are also important.

Research

Intensive research has been fostered by the world's RP groups and is making headway in unravelling the mysteries of RP and will hopefully soon lead to effective treatment and ultimately a cure.

Current approaches to research include -

- Understanding the structure and function of the retinal cells and their interaction with connections to the brain.
- Modification of the diseased retinal cells by genetic manipulation.
- Replacement of the diseased retinal cells with stem cells- derived either from embryos or adults.
- Halting or reversing the disease process through pharmacological means.
- Insertion of electronic equipment to function as a bionic eye.

Significant valuable research in Australia is contributing to the global effort in trying to overcome RP and other degenerative eye diseases. Australian research is funded by Retina Australia, through its grants program, and the National Health and Medical Research Council (NH&MRC).

Information about current research can be obtained through the local Retina Australia office or the various research websites. It is important to appreciate, however, that much information on the Internet may not have been examined with appropriate scientific rigor, so it is always important to check the source and to determine whether other scientists working in that field have reviewed the work. Retina Australia can put you in touch with local experts to help evaluate the merits of any research project.

Funding for Research

Retina Australia receives donations from each of the state bodies and allocates funds towards Australian research. Any contribution over \$2 is tax deductible.

(a) Social Security Act, Section 95 - Qualification for DSP - permanent blindness.

http://www.fahcsia.gov.au/guides_acts/ssq/ssguide-1/ssguide-1.1/ssguide-1.1.p/ssguide-1.1.p.210.html

(b) Much of the information in this section is derived from Retinitis Pigmentosa Overview, Gene Reviews, **www.genetests.org**. The data in this booklet is derived from the September 2005 update by RA Pagon and SP Daigen.

Contact List for further information

If you require support or more information about services or research, the following may be of help to you.

Retina Australia

Website: www.retinaaustralia.com.au

Email: admin@retinaaustralia.com.au

Freecall: 1800 999 870

Retina Australia (Queensland)

Phone: 07 300 300 65

Facsimile: 07 300 300 65

Free call (outside Brisbane): 1 800 000 999

E-mail: admin@retinaqld.org.au

Postal address: P.O. Box 16295

City East Qld 4002

Retina Australia (ACT) Inc
Ra1act@retinaaustralia.com.au

Tasmania (as for Victoria)

Retina Australia (Vic) Inc
support@retinavic.org.au
(03) 9650 5088

Retina Australia (NSW)
admin@retinaaustraliansw.com.au
(02) 9744 7738

Retina Australia (WA) Inc
hazel@rawa.com.au
(08) 9388 1488

Retina Australia (SA) Inc
sara@retinaaustralia.com.au
(08) 8362 1111

Northern Territory
(as for South Australia)

Genetic Advice

Queensland Clinical Genetic Service
Royal Children's Hospital and District Health
Service

Address: Back Road
HERSTON QLD 4029

Phone: 07 3636 1686

Facsimile: 07 3636 1987

Vision Rehabilitation

QUT Vision Rehabilitation Centre

Address: QUT Kelvin Grove Campus
Victoria Park Road

KELVIN GROVE QLD 4059

Phone: 07 3864 5743

07 3864 5695

Facsimile: 07 3864 5665

Email: optometry.enquiries@qut.edu.au

Website:

www.hlth.qut.edu.au/opt/research/lowvision.jsp

Orientation and Mobility

Guide Dogs (Queensland)

Address: 1978 Gympie Road
BALD HILLS QLD 4036
Phone: 07 3261 7555
Facsimile: 07 3261 7500
Email: admin@guidedogsqld.com.au
Website: www.guidedogsqld.com.au

Vision Devices and Rehabilitation

and for information on support groups in your area

Vision Australia

Street Address: 373 Old Cleveland Road,
Coorparoo Qld 4151

Postal Address: PO Box 1637,
Coorparoo DC Qld 4151

Phone: 1300 84 74 66

Website: www.visionaustralia.org
Email: info@visionaustralia.org

For further information

Retina International

Website: www.retinainternational.org

Foundation Fighting Blindness

<http://www.blindness.org>

Royal Victorian Eye & Ear Hospital

<http://www.eyeandear.org.au/healthinfo/rp/rpwhatis.asp> will provide links to reputable sources of information

Scottish Sensory Centre – RP

<http://www.ssc.education.ed.ac.uk/resources/vi&multi/eyeconds/RetPig.html>

British RP Society

<http://www.brps.org.uk/index.php?tl=aboutrp>