



# **What is Usher Syndrome?**

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## Preface

Retina Australia (Qld) has prepared several booklets to provide factual information about the most common retinal dystrophies – retinitis pigmentosa, Stargardt disease, Usher syndrome and macular degeneration. Usher syndrome is a genetic condition which causes hearing loss and progressive loss of sight due to retinitis pigmentosa (RP). For information about the problems of the eye, which occur in Usher Syndrome, referral should be made to the companion booklet “What is Retinitis Pigmentosa?”

These booklets also contain information about support groups which you may find helpful. They have been prepared following review of the current available literature, consultation with professionals who work in the field of ear disease, persons with the particular conditions and family members. The information provided is not intended to be comprehensive, but should serve as a basis for understanding the disease and for seeking further information from the literature and professionals

consulted about the disease. Possible useful websites will be listed at the end of this booklet. The first edition of these booklets were made possible by a grant from Queensland Health.. They are also available on audio CD and email as text or audio. The typeface conforms to the large print guidelines for vision impaired people. recommended by Blind Citizens Australia

Our thanks also go to Darren Le Brocque, graduate medical student, The University of Queensland, who helped format these books in their original editions, Dr John Vance for his contribution and Anne Housego for her work in preparation of this current edition. We are also grateful for the editorial assistance of Mr Bryan Pyman (consultant ENT Surgeon). More detailed information provided by Mr Pyman is available on request. Sources are acknowledged at the bottom of relevant pages.

Contact details for each state or territory organisation is at the back of the book.

# Structure and Function of the Ear

## Introduction

Before discussing Usher syndrome, it is important to have some understanding of the structure and function of both the ear and the eye. Information about the eye is contained in the companion booklet “What is Retinitis Pigmentosa?”

## Structure

The ear is divided into three (3) major parts – outer, middle and inner ear (See diagram P,7).

**Outer Ear** consists of

- **auricle**  
This is the outer part of the ear situated on either side of the head
  
- **ear canal**  
This canal, lined by skin, passes from the hole in the auricle through cartilage and bone to the tympanic membrane (ear drum).

- **tympanic membrane**

This is the thin transparent piece of skin which sits at the end of the ear canal

## **Middle Ear**

This is a small air filled bony chamber which contains three small bones connecting the tympanic membrane to the inner ear.

## **Inner Ear**

This consists of two parts:

- **cochlea**

This spiral shaped, fluid filled structure has an elastic membrane along its whole length. There are thousands of hair cells in 4 rows on top of the membrane. They fire when the hairs on their upper end bend with the resonating vibration. The bending is limited to the places where sound is resonating with the membrane. The nerves in the nerve of hearing are attached to just

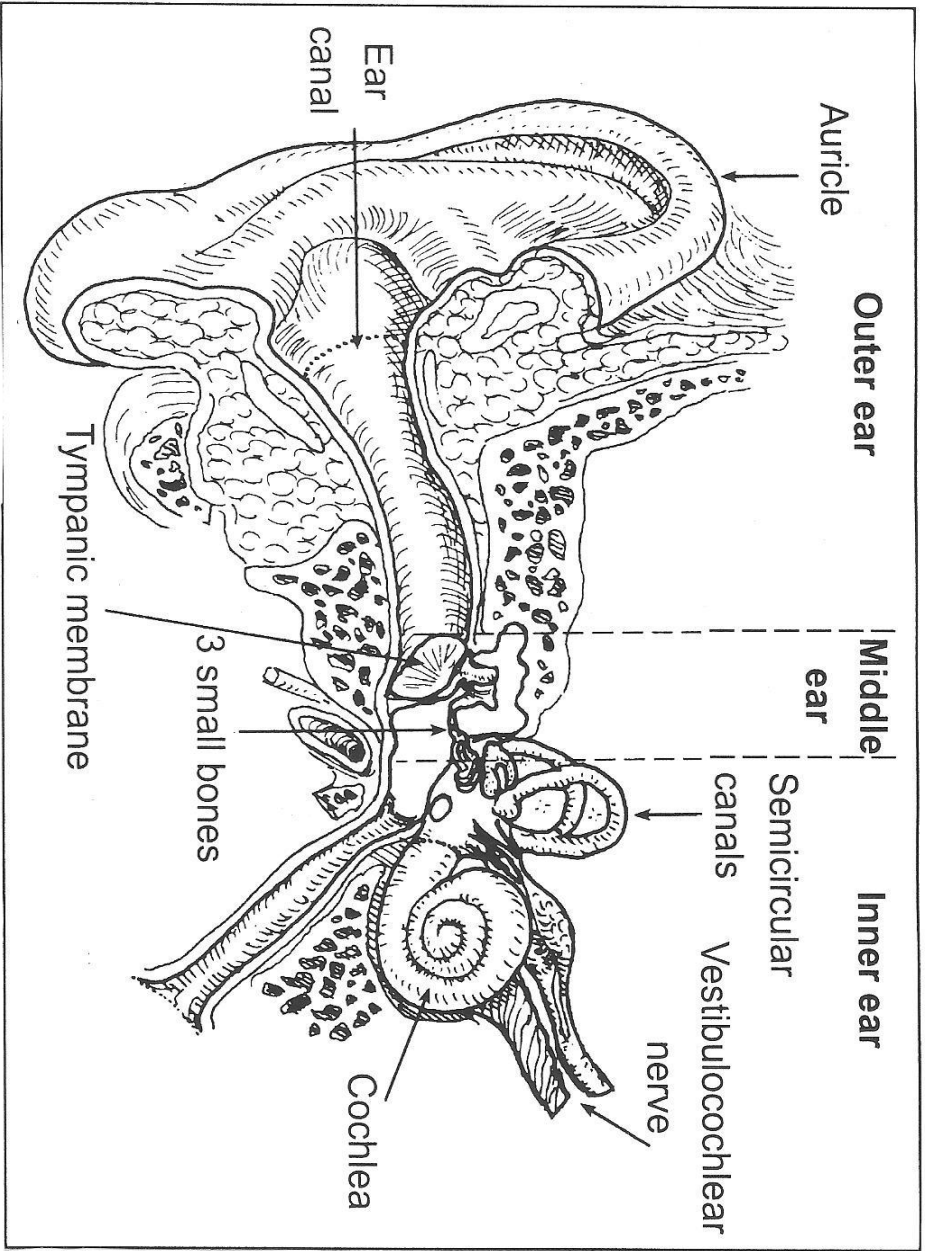
one hair cell. The brain learns about which sounds because of the association of the place of the membrane's resonance, the limited number of hair cells that fire and the dedication of the nerve fibres to just one hair cell. This association is called tonotopic coding of sound.

- **semi circular canals**

These structures are also filled with fluid

- **Vestibulo-cochlear nerve**

This nerve is connected to both the cochlea and semi circular canals. Each nerve fibre in the cochlear part of this nerve is attached to just one hair cell. This nerve conducts messages to the hearing centre in the brain.



## **Function of the ear**

When a sound is present it sets up vibrations in the ear which pass through the ear canal to the tympanic membrane and the three small bones. Together these structures increase the force of the vibrations entering the cochlea. The hair cells bend with this resonating vibration and stimulate its attached nerve fibre. This then passes information about the sound to that part of the brain which deals with hearing.

## **Causes of hearing loss**

There are two major types of hearing loss.

- **Conductive hearing loss**

This occurs when there is disruption of the air and/or bone vibration in the outer or middle ear and may add to the problem of sensorineural deafness if it is present.. It can be caused by problems such as wax in the outer ear, infection in the outer ear or infection in the middle ear.

- **Sensorineural deafness**

This occurs when there is damage to the cochlea or the vestibule-cochlear nerve and there are many possible causes. Usher syndrome is a genetic disorder. Deafness may not be present at birth and can be progressive but not necessarily profound or total in severity. In a family with the same pedigree, the severity of the hearing or vision impairment is not predictable.

## **How is Hearing Loss Tested?**

### **Distinguishing between conductive and sensory neural loss**

The vibrating tuning fork is held at the opening of the ear canal. The person is asked to indicate if they can hear the sound. Then the fork, still vibrating, is placed on the bone behind the ear and again the person is asked whether they can hear the sound. If the sound is louder when the tuning fork is applied to the bone then the person has a conductive hearing loss. For sensory-neural

deafness, the sound from the fork will be diminished both at the ear canal and bone.

## **Audiogram**

Each ear is tested separately with the ear not being tested covered with a sound proof earpiece. A series of sounds are produced by a machine to the ear being tested. The sounds vary according to their pitch or frequency (cycles/second) and loudness (decibel level). The loudness of the sound heard is recorded for the frequency levels from 250 to 8000 cycles per second. In people with normal hearing measurements of 20 decibels or less are recorded. As the loudness of the sound has to be increased in order to be heard by the patient, decibel levels of 80 – 100 may be needed, thus indicating severe hearing loss.

## **Other ear tests, such as electrical tests for balance**

These are less commonly performed and consultation with an ENT specialist would be necessary before proceeding.

## **What is Usher syndrome?**

In brief, RP causes degeneration of the retina. The retina is the light sensitive tissue inside the eye in which the first stages of “seeing” take place. Often the first symptom is night blindness followed by narrowing side vision leading to what is called “tunnel vision”.

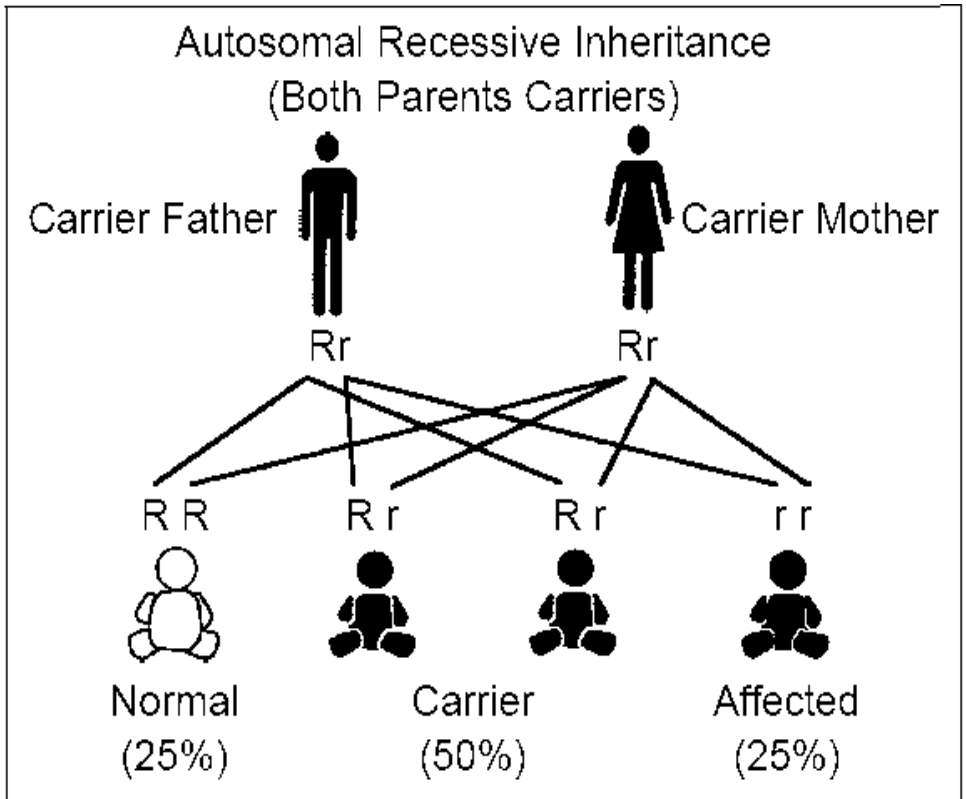
## **How do people with Usher syndrome become deaf?**

For reasons which are as yet unknown, the hair cells, which vibrate in the cochlear fluid, are damaged. Thus, when a sound is made near the auricle, it will pass through the canal to the middle ear, but subsequent transmission of vibration within the cochlea will be faulty and hence the sound will be inadequately passed along the vestibulo-cochlear nerve or recognised by the brain.

## **How do people get Usher syndrome?**

It is a genetic disorder, inherited in an autosomal recessive manner. This means that each parent (carrier) will need to carry the abnormal gene for one of their children to acquire Usher syndrome.

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.



## Types of Usher Syndrome

There are three types of Usher syndrome – 1, 2 and 3 with 1 having the most severe hearing loss and 3 the least. The following table summarises the main hearing, vision and balance symptoms.

	<b>Type 1</b>	<b>Type 2</b>	<b>Type 3</b>
<b>Hearing</b>	Profound deafness in both ears from birth	Moderate to severe hearing loss from birth	Normal at birth; progressive loss in childhood or early teens
<b>Vision</b>	Decreased night vision and tunnel vision before age 10	Decreased night vision and tunnel vision begins in late childhood or teens	Varies in severity; night vision and tunnel vision problems often begin in teens
<b>Vestibular function (balance)</b>	Balance problems from birth	Normal	Normal to near-normal, chance of later problems

## **Type 1**

Because of the profound hearing loss hearing aids may not help. Balance problems may show up as being slow to sit or walk. As vision problems occur later in childhood, it is important that diagnosis is made early to ensure the most useful educational help can be provided.

## **Type 2**

Most will find hearing aids very helpful and can use normal speech. Vision problems occur later in teens or early twenties.

## **Type 3**

Hearing aids are usually needed by mid – late teens. Vision symptoms of RP occur from puberty.

## **What is the incidence of Usher syndrome?**

Figures from the USA and Scandinavia suggest that 3 to 6% of all people born deaf have this condition. In Victoria, a recent survey of people with RP showed that 8% have Usher syndrome. Clearly, it is important to consider Usher syndrome in anyone who is young and is having considerable hearing problems which may well be evident before the symptoms of RP develop.

## **How does Usher syndrome affect people?**

Loss of hearing, balance and vision affect communication, mobility and everyday living. It can make independent living more difficult to achieve.

- **Nature of hearing loss**

The hair cells in the cochlea are damaged in Usher syndrome. The deafness has two important features. Firstly the ability to hear high frequencies is diminished so you would need a hearing aid to detect sounds such as 's', 't', 'p', 'th', 'ff', or 'k'. Secondly distortion of hearing, even if you are wearing a hearing aid, will make it difficult to distinguish 'seat' from 'thief' or 'peak'.

Lip reading may be more difficult because of the associated RP.

- Nature of the vision loss

Night vision loss and tunnel vision are described in the companion book “What is RP?”

## **Management of deafness in Usher syndrome**

It is important that a team of ENT surgeon, audiologist and speech therapist are engaged at an early age to help support the child and family. Also an optometrist and ophthalmologist should be involved in caring for the vision issues.

- Progressive audiological assessment

Neonatal screening for deafness takes place in most states. Tests to monitor the progress of the hearing loss will be necessary to determine the time to provide further intervention.

- Early fitting of hearing aids

Consultation with the ENT surgeon and audiologist will be important when deciding when to use hearing aids. Clearly the earlier this occurs, often as

early as three months, the better chance of developing understandable speech.

- Communication

The use of both sign language and oral communication should be encouraged. This will ensure that the ability to communicate uses both the available hearing and visual cues while awaiting the application of hearing aids or cochlear implant.

- Cochlear implant

For some people an implant may be a possible option. In this situation, special tests of hearing, consultation with an ENT surgeon and consideration of other social and personal factors will be important. Implantation as early as twelve months gives best results.

## **What can be done?**

Most Australian states have hearing testing programs for the newborn. Hence hearing loss in young babies (Type 1) is likely to be detected then. A common early symptom may also be delay in speech development. Early identification is clearly important to ensure the best educational programme.

The types of services required by a person with Usher syndrome will depend on age and severity of the hearing and vision problems, but will normally include:

- Audiological assessment at birth and membership of a team of ophthalmology, ENT and speech pathologist specialist for at least annual assessment. Assessment by an ear, nose and throat specialist to ensure the correct diagnosis of the hearing loss
- Assessment by an ophthalmologist to assess the presence and severity of RP.
- Special hearing and vision tests (for example, ERG) to confirm the diagnosis.
- Audiological support including fitting of hearing aids, when necessary.
- Progressive evaluation of hearing changes by ENT surgeon so that cochlea implant can be done if necessary
- Speech assessment and therapy as necessary.
- Genetic counselling. It is important that the genetic nature of Usher syndrome is fully explained to the parents and child. Also,

there are several other genetic conditions in which deafness and RP occur. Therefore, these need to be excluded or confirmed.

- Educational training which, at different ages, may include:
  - preschool and school assessment and placement
  - independent living training
  - career guidance
- Rehabilitation counselling which may include:
  - family support
  - orientation and mobility training
  - communication aids

## **Who needs to know about Usher syndrome**

It is clear that members of both the vision impaired and hearing impaired communities need to be aware of this condition to provide support to affected members.

Special consideration and understanding is important because both senses of vision and hearing are affected to a greater or lesser degree.

Advice about mobility and supporting aids may need to be modified to ensure maximum benefit.

## **Research**

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 the work has been reviewed by other scientists working in that field. 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 makes available around \$200,000 a year for Australian research. Any contribution over \$2 is tax deductible.

## **Useful Contact Information**

People who wish to learn more about Usher syndrome should consult their ENT surgeon and geneticist. Literature on several websites may be useful. These include:

- National Institute of Deafness and communicable Disease (NIDCD).  
[www.nidcd.nih.gov/hearing/usher.asp](http://www.nidcd.nih.gov/hearing/usher.asp)
  
- Sense for deaf blind people  
[www.sense.org.uk/what\\_is\\_deafblindness/usher\\_syndrome](http://www.sense.org.uk/what_is_deafblindness/usher_syndrome)
  
- Foundation Fighting Blindness (FFB)  
[www.blindness.org](http://www.blindness.org)

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

Retina Australia (toll free) 1800 999 870 will connect you with your own state based organisation.

### **Retina Australia**

Website:

[www.retinaaustralia.com.au](http://www.retinaaustralia.com.au)

Email:

[admin@retinaaustralia.com.au](mailto:admin@retinaaustralia.com.au)

Freecall: 1800 999 870

### **Retina International**

Website: [www.retina-international.org](http://www.retina-international.org)

### **Retina Australia (Queensland)**

Phone: 07 300 300 65

Facsimile: 07 300 300 65

Free call (outside of Brisbane): 1 800 000 999

E-mail: [admin@retinaqld.org.au](mailto:admin@retinaqld.org.au)

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Office located in Brisbane city.

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**Tasmania** (as for Victoria)

**Retina Australia (Vic) Inc**

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(03) 9650 5088

**Retina Australia (NSW)**

[admin@retinaaustraliansw.com.au](mailto:admin@retinaaustraliansw.com.au)

(02) 9744 7738

**Retina Australia (WA) Inc**

[hazel@rawa.com.au](mailto:hazel@rawa.com.au)

(08) 9388 1488

**Retina Australia (SA) Inc**

[sara@retinaaustralia.com.au](mailto: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](mailto:optometry.enquiries@qut.edu.au)  
Website: [www.hlth.qut.edu.au/opt/research/lowvision.jsp](http://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](mailto:admin@guidedogsqld.com.au)

Website: [www.guidedogsqld.com.au](http://www.guidedogsqld.com.au)

## **Vision Devices and Rehabilitation**

### 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](http://www.visionaustralia.org)

Email: [info@visionaustralia.org](mailto:info@visionaustralia.org)