

# Usher's Syndrome Type I

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## Abstract

A 5 yr old female child brought by her mother, came to our OPD complaining of wandering movements in both eyes with decreased hearing since birth. On examination, retinoscopy under atropine eye drops was -2.5D in both the axes. Fundus examination with indirect ophthalmoscope showed RPE irregularities suggestive of retinitis pigmentosa. ENT examination showed sensorineural deafness. ERG and fundus photography was done.

## Introduction

Usher syndrome is a genetic disorder that is characterized by hearing impairment as well as an eye disease called retinitis pigmentosa in which vision degenerates (gets worse) over time. Some people with Usher syndrome also have balance problems.

## The naming of Usher syndrome

The syndrome was first described by Albrecht Von Graefe in 1858. Other early descriptions were provided by R. Liebreich in 1861, who commented on a relatively high frequency in Jews in Berlin, and by V. Hammerschlag in 1907.

However, the syndrome is named for Charles Usher, a British eye doctor, who was the first to recognize clearly that this condition was inherited and to emphasize the hereditary aspect of the disease. Usher's report appeared in 1914. The syndrome still bears his name.

Usher syndrome is sometimes called "Retinitis pigmentosa and congenital deafness" (or "Congenital deafness and retinitis pigmentosa.")

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## Incidence and prevalence

Usher syndrome is the most common condition (aside from aging) that affects both hearing and vision. More than half of all deaf-blind people in developed countries (such as the US, Canada, and France) have Usher syndrome.

## Inheritance

The syndrome is transmitted in families by autosomal recessive inheritance, which requires the presence of two copies of the Usher gene for the disorder to be manifest. Each parent of a boy or girl with Usher syndrome is a "carrier" with one standard and one mutated Usher gene but no sign of the syndrome. A child with the syndrome has it because he or she received two mutated Usher genes, one from each of the carrier parents.

The parents' risk with each future pregnancy is:

- 1 in 4 (25%) that the child will have two mutated Usher genes and have the syndrome
- 2 in 4 (50%) that the child will have one mutated Usher gene and one standard gene and be an unaffected carrier
- 1 in 4 (25%) that the child will have two

standard and no Usher gene.

If a normal child from an Usher family marries more or less at random (they don't marry someone from another known Usher family), the risks for their offspring of Usher syndrome are less than 1%. (Assuming a gene frequency in the general population of 1%, the risks are  $2/3 \times 1/100 \times 1/4 = 1/600$ .)

#### Case Report

A 5 year old female child brought by her mother, came to our OPD complaining of wandering movements in both eyes with decreased hearing since birth. On examination, retinoscopy under atropine eye drops was -2.5D in both the axes. Fundus examination with indirect ophthalmoscope showed RPE irregularities suggestive of retinitis pigmentosa. ENT examination showed sensorineural deafness. ERG and fundus photography was done. ERG showed bilateral diffuse reduction in the photoreceptor function (rods > cones) in the retina mid periphery. Fundus showed bilateral arterial attenuation and optic disc pallor and perivascular bony spicule pigmentation suggestive retinitis pigmentosa (Fig. 1).

#### Discussion

##### Three types of Usher syndrome

There are three different clinical types of Usher syndrome (US). They are called Usher syndrome type 1 (US1), Usher syndrome type 2 (US2), and Usher syndrome type 3 (US3).

Types 1 and 2 are more common than type 3. All types of the syndrome are inherited in the same pattern - as autosomal recessive traits.

- Usher syndrome type 1 (US1) - People with US1 are profoundly deaf from birth and have severe balance problems. Many of these individuals obtain little or no benefit from hearing aids. Most use sign language as their primary means of communication. Because of the balance problems, children with US1 are slow to sit without support and rarely learn to walk before they are 18 months old. These children usually begin to develop vision problems by the time they are ten. Visual problems most often begin with difficulty seeing at night, but tend to progress rapidly until the individual is completely blind.
- Usher syndrome type 2 (US2) - People with US2 are born with moderate to severe hearing impairment and normal balance. Although the severity of hearing impairment varies, most of these children perform well in regular classrooms and can benefit from hearing aids. These children most commonly use speech to

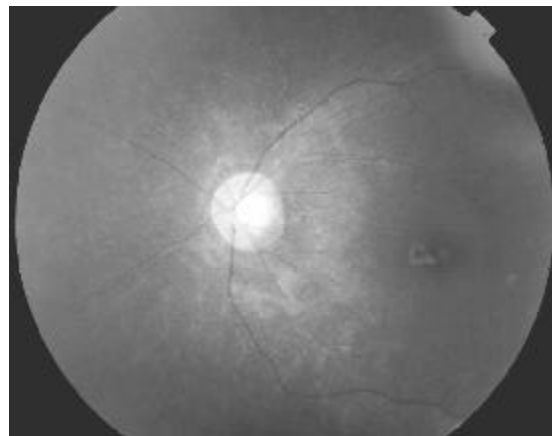
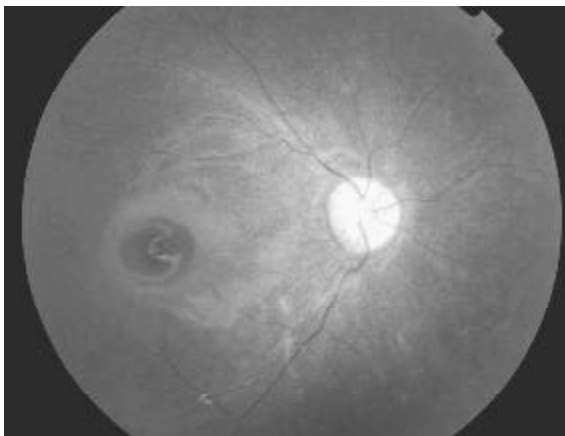


Fig. 1a and b : Fundoscopy showing bilateral arterial attenuation and pale optic disc.

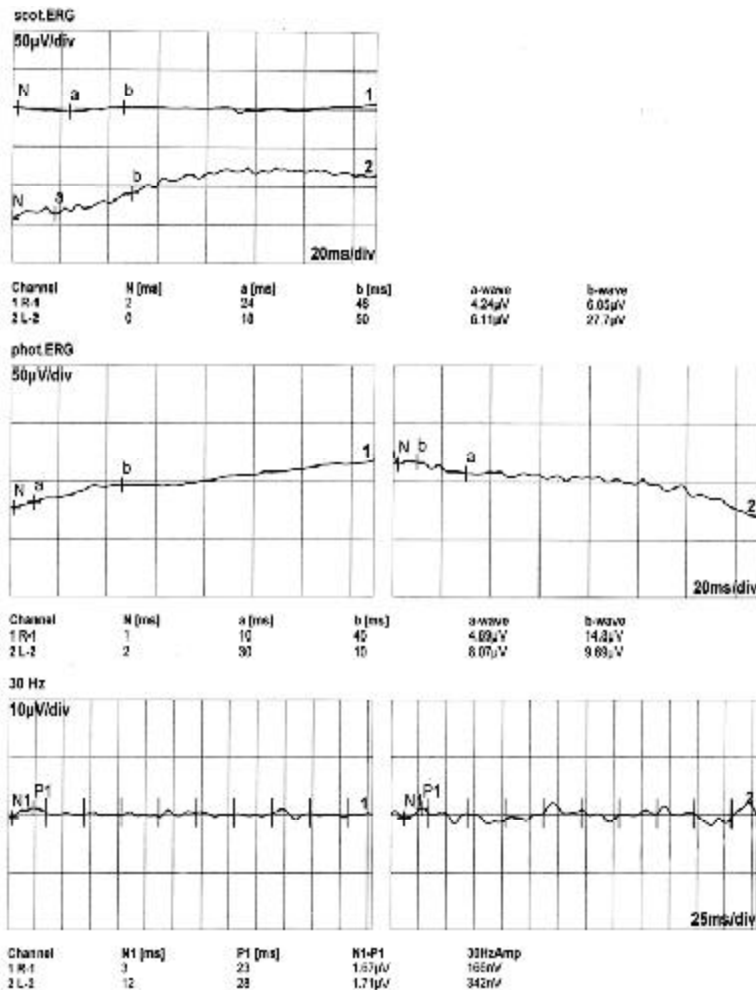


Fig. 2 : ERG of the patient

communicate. Retinitis pigmentosa, which is a degeneration of the retina (the part of the eye that receives images of objects), is characterized by blind spots that begin to appear shortly after the teenage years. The visual problems in US2 tend to progress more slowly than the visual problems in US1. When an individual's vision deteriorates to blindness, his or her ability to read speech from the lips is lost.

- Usher syndrome type 3 (US3) — Children born with US3 have normal hearing and

normal to near-normal balance. Hearing worsens over time. Children develop noticeable hearing problems by their teenage years and usually become deaf by mid to late adulthood. Retinitis pigmentosa in the form of night blindness usually begins sometime during puberty. Blind spots appear by the late teenage years to early adulthood. By mid adulthood, the individual is usually blind.

### Diagnosis of Usher's Syndrome

Hearing loss and retinitis pigmentosa are rarely found in combination other than from

Usher syndrome. Finding them together is strong presumptive evidence for Usher syndrome.

Special tests such as electronystagmography (ENG) to detect balance problems and electroretinography (ERG) to detect retinitis pigmentosa help detect Usher syndrome early (Fig. 2).

Early diagnosis is important in order to begin special educational training programme to help the individual deal with the combined hearing and vision difficulties.

### **Treatment**

The best treatment depends on early identification to begin educational programme. The exact nature of these educational programme depend on the severity of the hearing and vision impairments as well as the age and abilities of the individual.

Typically, people with Usher syndrome benefit from adjustment and career

counselling; access to technology such as hearing aids, assistive listening devices or cochlear implants; orientation and mobility training; and communication services and independent living training that may include Braille instruction, low vision services, or auditory training.

### **References**

1. Jack J. Kanski clinical ophthalmology 6th edition 2007 Pg 667/ Elsevier
2. Paediatric Ophthalmology Havley, 1983 WB Saunders's company Page 75,611,1073.
3. De Haas EB, *et al.* usher's syndrome with special reference to heterozygous manifestations *Docum. Ophthal* 1970; 28 : 166.
4. Fishman GA. Usher's syndrome visual loss and variations in clinical expressivity. *Perspect Ophthal* 1979; 3 : 77.
5. Sondheimer S, Fishman GA, Young RS, Vasquez VA. Dark adaptation testing in Usher's Syndrome *British Journal of Ophthal* 1979; 63 : 547.
6. Vernon M. Usher's syndrome Deafness and progressive blindness Clinical cases Prevention theory and literature survey. *J Chron Dis* 1969; 22 : 133.

#### **Let There be Sleep – On Time**

Shantha Rajaratnam and colleagues findings show that, like melatonin, melatonin analogues – largely because of their regulatory effects on the circadian system – can be highly effective for treating the range of symptoms that accompany transmeridian air-travel or shift-work as well as other circadian-rhythm sleep disorders.

An important point for the effects of melatonin analogues is to understand that they are not standard hypnotic drugs that resemble benzodiazepines and their derivatives. Melatonin-like compounds amplify day-night differences in alertness and sleep quality, and so exert a modest (compared with benzodiazepines) sleep-promoting effect. In view of the negative effects of benzodiazepines (addiction, dependence, etc) and the absence of these negative effects with melatonin analogues, an important educational goal for public-health authorities is to change the opinion of consumers about the validity of any sleep-promoting therapy.

**Daniel P Cardinali, Diego A Golombek, The Lancet, 2009; 373 : 439-40.**