

# Usher Syndrome- A Rare Case

Eva Rani Tirkey (MS), Sujata Lakhtakia (MS)\*

Department of Ophthalmology, SS Medical College, Rewa, Madhya Pradesh, India

## INTRODUCTION

Usher syndrome is a rare heterogenous autosomal recessive genetic disorder with features of visual impairment due to retinitis pigmentosa and hearing loss. Other names for it include Hallgren syndrome, Usher-Hallgren syndrome, retinitis pigmentosa-dysacusis syndrome, and dystrophia retinae dysacusis syndrome.<sup>1,2</sup> Usher syndrome represents a genetically diverse condition that involves both early onset sensorineural hearing loss and retinal pathology. While reports of disease prevalence vary, the condition has been estimated to occur in three in 100,000 individuals.<sup>3</sup>

## Case Report

A 30 year old female presented to the Ophthalmology outpatient department with complaints of diminished vision in both eyes more at night since 15 years. Diminution of vision was insidious in onset, gradually progressive and painless. She also had a history of difficulty in hearing for last 3 years. There was no history of use of spectacles or hearing aids. No history of difficulty in walking. She was born out of a non-consanguineous marriage.

UCVA in both eyes was 5/60 and improved to 6/12 with -5.0DS in both eyes. Near vision and colour vision were normal. Examination of anterior segment of both eyes was found to be within normal limits; both pupils were 3 mm in size, round, regular and reactive. Ophthalmoscopy of both eyes showed clear media, waxy gliotic pallor of optic discs, severe thread like arteriolar attenuation and bone spicule retinal pigmentation in the mid periphery, characteristics of Retinitis pigmentosa (Figures 1 and 2).

Pedigree charting was done and it was found that no family members were affected. ERG showed subnormal 'a' wave and 'b' wave and visual fields examination revealed constriction of peripheral fields (Figures 3 and 4).

ENT consultation was done and audiometry showed moderate to severe sensorineural deafness in both ears.

Vestibular function was normal. ECG and all routine investigations were done and physician opinion was obtained. Putting together all these findings a diagnosis of Type II Usher's syndrome was made. This patient was prescribed refractive correction, vitamin A therapy was started and was asked to come for follow-up after 6 months.

## DISCUSSION

Charles Usher, a Scottish ophthalmologist, examined the pathology and transmission of the disease in 1914 on the basis of 69 cases and hence the condition is named after him. Richard Liebreich examined the population of Berlin for disease pattern of deafness with retinitis pigmentosa and noted it to be recessive since the cases of blind-deafness combinations occurred particularly in the sibs of blood-related marriages or in families with patients in different generations. His observations supplied the first proof for the coupled transmission of blindness and deafness since no isolated cases of either could be found in the family trees.<sup>4,5</sup> Usher's syndrome can be divided into three major groups- Type 1, type 2, type 3.

## Usher syndrome type-I

Usher I patients have difficulties maintaining their balance due to vestibular system problems. These babies are born deaf and are usually slow to develop motor skills such as walking. Worldwide, its estimated prevalence is 3 to 6 per 100,000 people in the general population and is more common in people of Ashkenazi Jewish ancestry (central

Address for correspondence: Sujata Lakhtakia,  
Department of Ophthalmology, SS Medical College, Rewa, Madhya Pradesh, India  
E-mail: meetsujata123@yahoo.co.in



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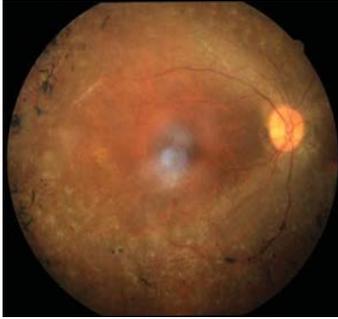
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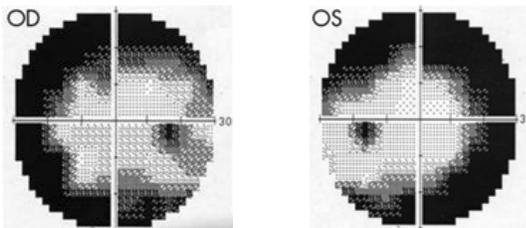
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**Figure 1:** Right eye disc pallor arteriolar attenuation and bony spicules suggestive of retinitis pigmentosa



**Figure 2:** Left eye disc pallor arteriolar attenuation and bony spicules suggestive of retinitis pigmentosa



**Figure 3:** Constriction of peripheral visual fields in both eyes

and eastern European) and in the French-Acadian populations (Louisiana).<sup>6,7</sup>

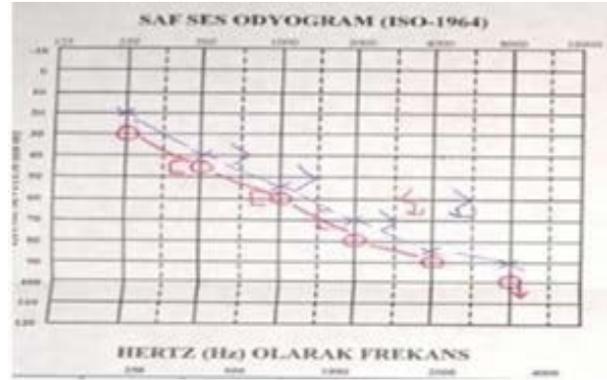
Usher type I is caused by mutations in any of the following genes: *cdh23*, *myo7a*, *pcdh15*, *ush1c*, and *ush1g*.

### Usher syndrome type-II

Usher II is characterized by hard-of-hearing and their hearing does not reduce over time with a normal vestibular system. It occurs at least as frequently as type-I, but because type-II may be underdiagnosed or more difficult to detect, it could be up to three times as common as type I. Usher syndrome type II may be caused by mutations in any of the three different genes-*ush2a*, *gpr98*, and *dfb31*.

### Usher syndrome type-III

Usher III is characterized by ‘progressive’ loss of hearing and half have vestibular dysfunction. Its incidence is highest in the Finnish population but rare in other ethnic groups. Mutations in only one gene, *clrn1* have been found in Usher



**Figure 4:** Moderate to severe sensorineural deafness

III. *clrn1* encodes clarin-1 is a protein important for developing and maintaining the inner ear and retina. But how its mutation causes hearing and vision loss, is still not clearly understood.<sup>8</sup>

### Differential Diagnosis

Other syndromes that can be associated with pigmentary retinopathy and deafness must be ruled out when considering a diagnosis of Usher’s syndrome. These include infantile and adult Refsum disease, Kearns Sayre syndrome, Cockayne syndrome, Alstrom disease, Bardet Biedl syndrome.

### Treatment

Currently, there is no cure for Usher syndrome. The best possible treatment is early identification so that educational and counseling programs can be done. Treatment will include hearing aids, assistive listening devices, cochlear implants, or other communication methods such as American Sign Language; orientation and mobility training; and communication services and independent-living training, including Braille instruction, low-vision services, or auditory training. Some ophthalmologists believe that a high dose of vitamin A palmitate may slow, but not halt, the progression of retinitis pigmentosa.<sup>9</sup>

### CONCLUSION

Investigation of Usher’s syndrome in patients will promote better rehabilitation and monitoring of patients. Consanguinity should be sought in affected individuals. Our case report shows a patient with deafness and blindness diagnosed to be Type 2 Usher’s syndrome. So, any patient with retinal degeneration suspected of even slight hearing loss must receive audiologic evaluation and treatment to minimize the effect of major sensory problems that arise from combined hearing and vision deficits.

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