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Case report

"Usher syndrome Type I in an adult Nepalese male: a rare case report"

Sabin Sahu[1], Sanjay Kumar Singh[1]
[1]Sagarmatha Choudhary Eye Hospital, Lahan – 3, Siraha, Nepal

Introduction

Usher syndrome, also known as retinitis pigmentosa-dysacusis syndrome, is an extremely rare genetic disorder, characterized by retinitis pigmentosa (RP) and congenital sensorineural hearing loss. It has been estimated to account for 3-6% of the congenitally deaf population, upto 8-33% of individuals with RP and half of all cases with combined deafness and blindness (Vernon M,1969; Boughman JA et al,1983). The prevalence of Usher syndrome have been reported to range from 3.5 to 6.2 per 100,000 in different populations (Vernon M,1969; Boughman JA et al,1983; Yan D et al, 2010).

Usher syndrome is clinically variable and genetically heterogeneous autosomal recessive disorder. It is traditionally subdivided into three clinical subtypes: Types I, II and III (Yan D et al, 2010; Kremer H et al 2006). The subtypes are differentiated by the severity and progression of the hearing loss and by the presence and absence of vestibular symptoms, with visual impairment due to RP being common to all three subtypes. Type I is the most severe form with congenital profound deafness and vestibular dysfunction as well as pre-pubertal onset of progressive RP. Type II is less severe than type I and is characterized by congenital moderate to severe deafness, normal vestibular function with onset of RP in first or second decade. Type III is least common and is characterized by variable onset of progressive hearing loss, vestibular function and variable onset of RP (normal to absent) (Yan D et al, 2010).

We report a case of Usher syndrome type I in an adult Nepalese male with typical congenital profound hearing loss, and night blindness secondary to retinitis pigmentosa.

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Corresponding Author
Dr Sabin Sahu;
Ophthalmologist, Sagarmatha Choudhary Eye Hospital,
Lahan – 3, Siraha, Nepal
Tel : +977-9862223692, +977-33-560402; Fax: +977-33-560492
E mail: sabinsahu@gmail.com

Case report

A 30-year-old male presented with chief complaint of gradually progressive night blindness. His elder brother accompanying him gave a history of being deaf and dumb since early childhood. There was no history of similar illness in the family.

On examination, the patient had a visual acuity of 6/60 in both eyes improving to 6/9 with -7.0 DS/ -1.0 DC x 40^{o} correction in the right eye and 6/9 with -7.0 DS/ -0.75 DC x 40^{o} correction in the left eye. His intraocular pressures were normal in both eyes. On slit lamp examination, normal conjunctiva, clear cornea, quiet anterior chamber with normal anterior chamber depth, normal iris was seen in both eyes. Both eyes had early posterior subcapsular cataract (Figure 1). Dilated fundus examination revealed arteriolar attenuation and mid-peripheral bone-spicule pigment distribution, with normal disc and macula in both eyes (Figure 2).

Visual field testing with Goldmann perimetry showed generalized peripheral visual field constriction in both eyes. His fields were severely constricted to 15^{o} tunnel vision in right eye and 20^{o} tunnel vision in left eye with the Goldmann V4e target (Figure 3). Colour vision test on Ishihara charts and contrast sensitivity test was

normal in both eyes. Ear, Nose and Throat (ENT) consultation for assessment of hearing loss was conducted. Audiometry revealed no air-bone gap and both air and bone conduction was more than 100 db in most frequencies in both ears (Figure 4) suggestive of profound sensorineural hearing loss bilaterally. The electroretinogram (ERG) and genetic testings could not be done due to lack of the facilities in our centre.

The patient was given the optical correction and kept under regular follow up.

Figure 1. Slit lamp photograph of right eye (A) and left eye (B) showing posterior subcapsular cataract.

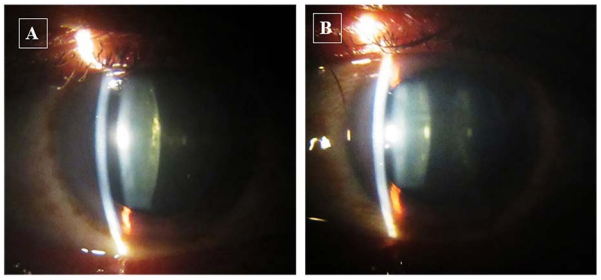


Figure 2. Fundus photograph showing arteriolar attenuation, mid-peripheral bone-spicule pigment distribution with normal disc and macula in right eye (A) and left eye (B).

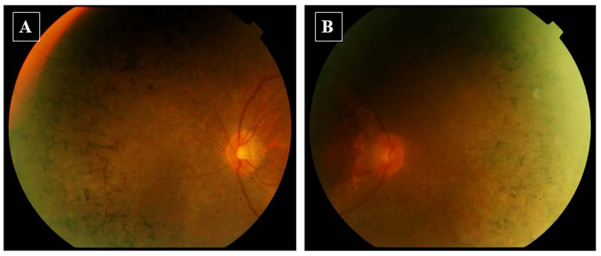


Figure 3. Visual field testing with Goldmann perimetry showing severely constricted fields to 15o tunnel vision in right eye (A) and 20° tunnel vision in left eye (B) with the Goldmann V4e target.

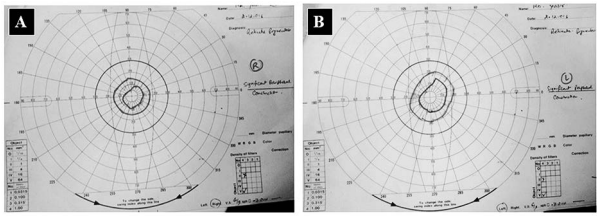
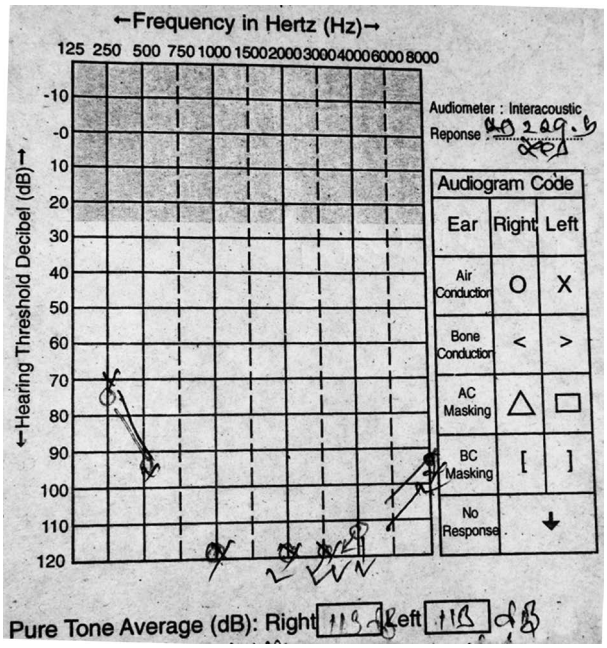


Figure 4. Pure tone audiometry showing no air-bone gap and both air and bone conduction more than 100 db in most frequencies in both ears.



Discussion

Usher syndrome represents the most common cause of inherited deafness and blindness. It is a genetically heterogeneous condition with variable clinical presentation. It typically presents with profound sensorineural hearing loss and progressive visual impairment secondary to retinitis pigmentosa. The clinical classification based on onset and severity of hearing loss, vestibular function and retinitis pigmentosa is traditionally used to determine various types of Usher syndrome (Yan D et al, 2010; Kremer H et al 2006). However, molecular genetic analysis is used, when available, to guide the investigation of these cases. Genetic testing has determined nine different genes responsible for Usher syndrome (Yan D et al, 2010). Inherited in autosomal recessive pattern, mutations in these genes can lead to significant functional impairment, so early detection and accurate diagnosis is essential.

In our case, there was profound congenital hearing loss due to which patient had unintelligible speech and difficulty with night vision started in early childhood. There was presence of early posterior subcapsular cataract in both eyes. It has been shown that patients with Usher syndrome are susceptible to visual loss from posterior subcapsular cataracts, atrophic appearing or cystoids macular lesions and epiretinal membranes, which can be seen in patients with retinitis pigmentosa (Edwards A et al, 1998). Visual acuity appears to be better retained in older patients with type II Usher syndrome compared to type I. However, no difference in the prevalence of posterior subcapsular cataracts has been noted between the two types (about 50% in both types) (Piazza L et al, 1986). The electrophysiologic test ERG which can detect sub-clinical cases of RP and has been described as early predictor of the disease (Piazza L et al, 1986; Janaky M et al, 2007) could not be done due to lack of the facilities in our centre. A report of two Nepali siblings, aged 13 and 16, with Usher syndrome type I has been earlier reported (Sah RP et al, 2015). We report a case of Usher syndrome type I in adult Nepalese patient with typical congenital profound hearing loss, speech disability and night blindness secondary to retinitis pigmentosa.

The multiple visual, auditory and speech impairments in Usher syndrome patients make them functionally handicapped. Early evaluation of deaf and dumb children for detection of Usher syndrome can help in early interventions to prevent multiple visual, auditory and speech impairments. Those diagnosed at early age require comprehensive care of different medical impairments. A great deal of educational and socio-psychologic interventions is thus necessary to help them maintain independence and productivity (Tamayo ML et al, 1997).

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