

USHER SYNDROME – CASE REPORT OF TWO SIBLINGS

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INTRODUCTION

Usher syndrome is an autosomal recessive disorder characterized by congenital sensory neural deafness and progressive visual loss secondary to retinitis pigmentosa. [1] Delay in diagnosis of Usher syndrome type I, the most dreadful variant of this disease, may lead to a deaf, dumb and a blind child. Two family members of Usher syndrome type I were identified and reported here.

CASE 1

An eight years old girl presented with history of progressive decrease in visual acuity especially at night since birth, decreased hearing and squint in the left eye. There was history of delayed milestones and broad based gait. She was advised hearing aids by audiologist when she was 2 years of age.

Ocular examination revealed best corrected visual acuity of 20/50 (6/24) in right eye and 20/100 (6/30) in the left. There was 40° esotropia in left eye more for near that was partially corrected with glasses. Ocular movements were of full range, but there was lack of fusion and stereopsis. Slit lamp examination revealed coronary cataract in both eyes, while fundoscopy revealed cystoid macular edema (Fig. 1) and arteriolar attenuation with peripheral pigmentary bone spicule like changes (Fig. 1)

Her speech reception threshold was monitored and response at 7/db in right ear and 80/db in left ear was found indicating profound hearing loss (Fig. 2). Her caloric test was normal. Her chromosomal analysis revealed deletion in long arm of X-chromosome in all the metaphases showing 46XXq. Her MRI brain showed normal study.

All these clinical findings confirmed the diagnosis of Usher syndrome type 1.

She was advised executive bifocal glasses and was offered squint surgery. Post operatively her eyes were straight and she was advised patching of good eye for two hours a day along with advice to do near work at that time. Tab Acetazolamide 125mg twice a day was prescribed for cystoid macular edema.

CASE 2

A nine years old boy (brother of case1) presented with history of decrease visual acuity since birth especially at night, right esotropia, and decreased hearing which was noticed since 1 year of age. There was history of delayed milestones with broad based gait. He was advised with hearing aids by audiologist when he was 2 1/2 years of age.

Ocular examination revealed best corrected visual acuity of 20/100 (6/30) in right eye and 20/80 (6/24) in left eye. There was 30° esotropia in right eye more for near with glasses. Ocular movements were of full range with V pattern esotropia and right inferior oblique over action. There was lack of fusion and stereopsis. Worth four dot test revealed right suppression. Slit lamp examination revealed few vitreous cells while fundoscopy revealed normal optic disc (Fig. 3) and arteriolar attenuation and peripheral pigmentary bone like spicule changes (Fig. 3)

On investigations his chromosomal analysis revealed no numerical or structural abnormality. His speech reception threshold was monitored and responses at 75/db in both ears were found with moderate to severe sensory neural deafness. His caloric test revealed no response in both ears with ice cold water irrigation. His MRI brain showed normal study. Based on these finding

diagnosis of Usher syndrome type 1 was made. He was advised executive bifocals and was offered squint surgery. Post operatively his eyes were straight and he was advised patching of good eye for 2hrs a day along with advice to do near work at that time.

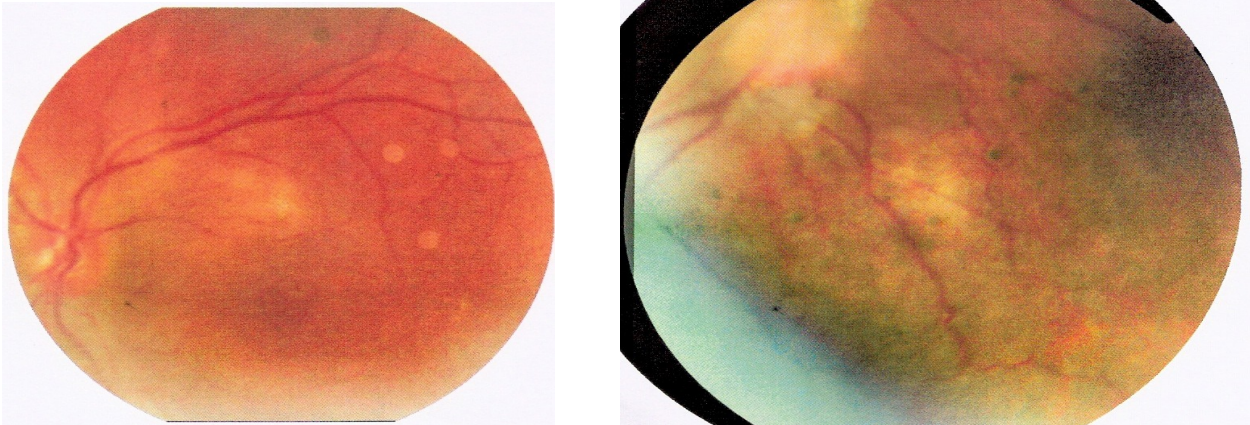


Fig. 1: Left Fundus showing cystoid macular edema and right eye in case 1 showing vascular attenuation and bone spicule like pigmentation

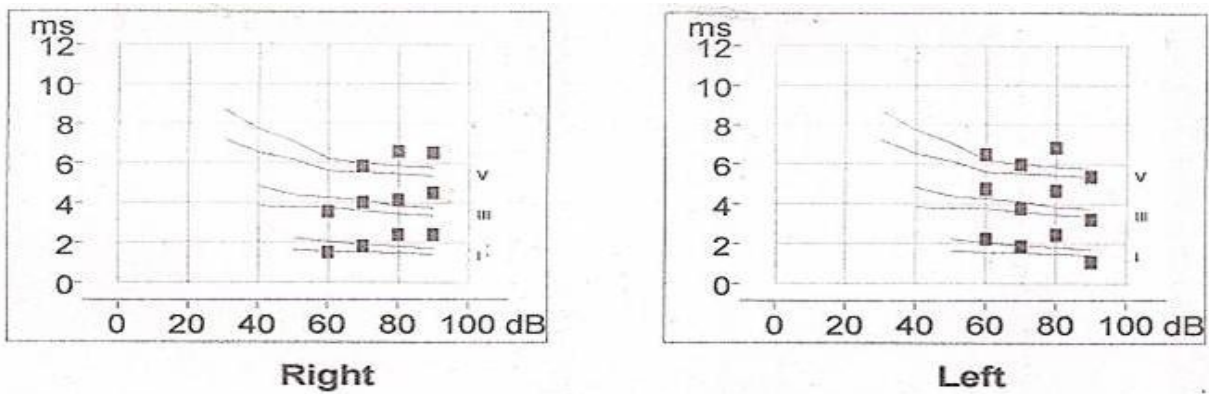


Fig. 2: Audiometric analysis of case 1 showing severe degree of hearing impairment in both ears

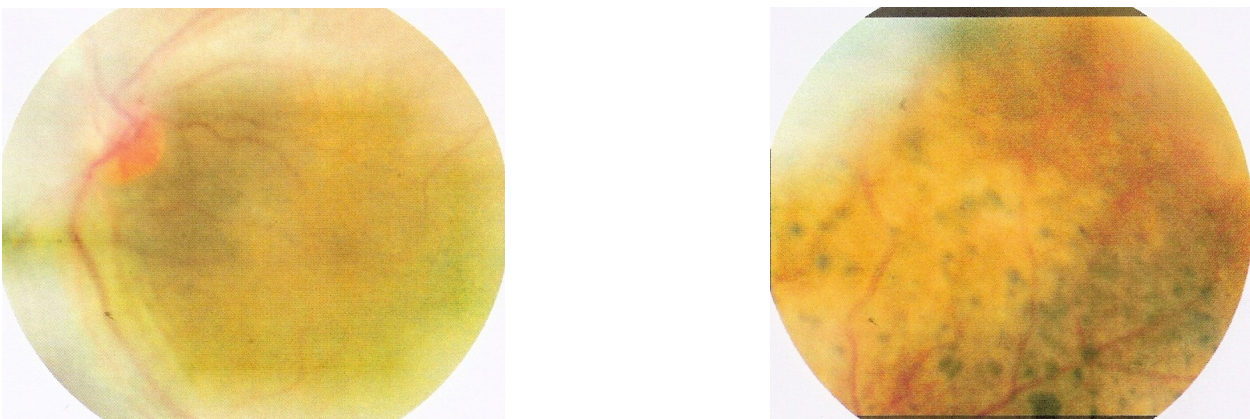


Fig. 3: Left Fundus showing normal optic disc and macula and in case 2 left fundus showing vascular attenuation and bone spicule like pigmentation

DISCUSSION

Usher syndrome is a rare autosomal recessive disorder characterized by congenital sensory neural deafness and progressive visual loss secondary to pigmentary

retinopathy, [1]. Usher was the first to recognize its hereditary nature, [2] and about 3- 6% of congenital deaf population is considered to have Usher syndrome [3].

Three clinical types are recognized. Patients with Usher syndrome type 1 are characterized by profound congenital sensory neural deafness, impaired speech development, and congenital absence of vestibular function with visual symptoms developing early. Most type 1 patients have a mutation at 11q13.5 [4]. Patients with Usher syndrome type- 2 demonstrate a congenital non-progressive hearing loss which is milder in the low frequencies with normal vestibular function. Usher syndrome type 2 has been located to 1q [5]. Cases of Usher syndrome type-3 show normal hearing development but profound deterioration occurring between the first and fourth decade, with normal speech development and variable vestibular function [6].

Progressive pigmentary retinopathy develops early in all clinical types, and the combination of visual and vestibular impairment causes marked functional disability, although a percentage of patients maintain central visual acuity of 6/60 or better until the fifth or sixth decade [7].

In this case report the kids were siblings born with age difference of one year. It was the aggressive efforts of parents and treating physicians that both kids were living a near normal life. Hearing aids were advised at 2 years of age and speech therapy was done in early life that prevented the children to become mute.

It is clinically important in children with profound hearing loss diagnosed by

otolaryngologists to seek the diagnosis of Usher syndrome by ophthalmic consult. All patients of Usher syndrome should receive cochlear implants at an earlier age along with speech therapy for maximal development of their skills to decrease sensory isolations in their future lives.

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