A case study of Usher's syndrome

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ABSTRACT

Usher's syndrome is the most common condition that affects both hearing and vision. The major symptoms of Usher's syndrome are hearing loss and an eye disorder called retinitis pigmentosa, or RP. RP causes night-blindness and a loss of peripheral vision through the progressive degeneration of the retina. As RP progresses, the field of vision narrows—a condition known as "tunnel vision"—until only central vision remains. Many people with Usher's syndrome also have severe balance problems.

Keywords: Usher's syndrome, Retinitis Pigmentosa, Electroretinography.

INTRODUCTION

Usher's syndrome is autosomal recessive deafnesswith retinopathy similar to Retinitis Pigmentosa(RP).Usher's syndrome is classified into 3 types, on the basis of severity of hearing loss, age of onset, and rate of vision loss. Patients with Type I are born deaf, and begin to lose their vision in the first decade of life. They also have balancing difficulties, and learn to walk slowly as children, due to problems in their vestibular system. Patients with Type II are not born deaf, but have hearing loss. They do not seem to have noticeable problems with balance; they also begin to lose their vision later in their life and may preserve some vision even into middle age. Patients with Type III are not born deaf, but experience a gradual loss of their hearing and vision; they may or may not have balance difficulties.[Table 1]

CASE REPORT

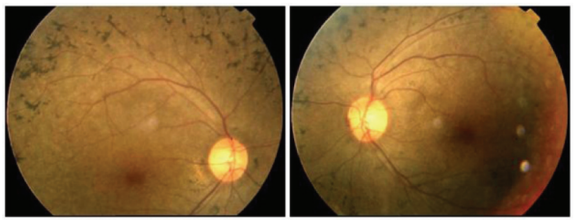
A 30year old female presented with complaints of defective vision both during the day and night since adolescence associated with deafness since childhood.Vision loss was gradual, painless, progressive in nature.Unaided Visual acuity in Both Eyes- Counting Fingers 3m.Best Corrected Visual Acuity: Right Eye: -3.50 sph 6⁄18 partialLeft Eye: -3.50 sph -1.00 cyl 160degree 6⁄18 partial.

On examination both eyes anterior segment was normal.Pupils were normal in size briskly reacting to light.On fundus examination:Both Eyes showed pale waxy disc, attenuated vessels, Dullfoveal reflex and multiple bony spicules in the periphery, which is suggestive of Retinitis Pigmentosa [Fig 1]. Patient was then sent to ENT department for evaluation of deafness.

ENT Examination: Rhinne's test- Both Ears Airconduction> Bone conduction.

Weber's test: Laterality towards left ear. Pure Tone Audiometry: Sensory neural hearing loss.

Figure 1: Fundoscopy showing changes in Usher's syndrome



DISCUSSION

Piazza et al., in astudy of 106 patients with Usher syndrome, found that 33% had type 1 and 67% had type 2 disease. No type 3 cases wereidentified. The overall prevalence rate of Usher's syndrome was estimated to 5⁄100000.Patients with type 2 report onset of nyctalopia over agreater range, up to the early 30s. Visual acuity appears to be better retained in older patients with type 2 as compared to type 1.

The electroretinography (ERG) is often profoundly abnormal to non-detectable, non-averaging techniques in all types.

Table1: Showing different types of Usher's syndrome

	TYPE 1	TYPE 2	TYPE 3
HEARING	Profound deafness in both ears from birth	Moderate to severe hearing loss from birth	Normal at birth; progressive loss in childhood or early teens
VISION	Decreased night vision before age 10	Decreased night vision begins in late childhood or early teens	Varies in severity; night vision problems often begin in teens

BALANCE	Balance problems from birth	Normal	Normal to near-normal, chance of later problems

Occasional ataxia in type 2 disease has been attributed to cerebellar atrophy.[1] Speech can be so severely affected asto be unrecognizable.Hunter et al.[2] found abnormalaxonemes in sperm from subjects with Usher syndrome andabnormal axonemes on histologic examination of retinal sectionsfrom a patient with Usher syndrome. Connor et al.[3] foundthat spermatozoa from patients with Usher syndrome type 2 had an abnormal lipid composition that was associated withreduced motility. Barronget al.[4] also found abnormal connectingcilia on electron microscopy of retinal sections from a malewith type 2 Usher syndrome. These findings suggest that boththe cilium of the inner ear and the cilium of the photoreceptorsmay share axonemal components that are necessary for formationof ciliated structures and whose genes would be candidatesfor the defective gene in Usher syndrome. A similar explanation may account for the report of olfactory disorder in somepatients with Usher syndrome.[5]

Management includes low vision aids, regular follow-up, Vitamin A supplementation of about 15,000IU/ day. The visual outcome for these patients is poor as the damage done to the retina is irreversible.She was prescribed hearing aids, and was advised regular follow-up [6,7].

CONCLUSION

The loss of both vision as well as hearing is a great burden on the patient, hence education and counselling for socio-psychological benefits to the patient and society is very important.Genetic counselling and the examination of the patient's siblings and off-springs needs to be done for early diagnosis and intervention for better visual and auditory outcomes.

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