CASE REPORT

Usher syndrome

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ABSTRACT

To report two brothers who have Usher's syndrome. Two brothers, 24 and 26, reported to the Chittagong eye hospital and training complex with symptoms of progressive vision loss in both eyes, particularly at night. Their greatest corrected visual acuity was 6/36 in the right eye and 6/24 in the left eye, according to their ocular exams. Pigmentary retinopathy was discovered during a dilated fundus examination. Perimetry exhibited a narrowing of the peripheral visual field Sensorineural hearing loss was discovered during their systematic assessment. These are clinical trials. The Usher syndrome Type 1 was diagnosed based on the findings. Low vision aids were recommended to the patients in order to enhance their eyesight. The patient's family members were assessed for quality of life and counseled about the disease's prognosis. Early identification and treatment will assist to maintain the remaining eyesight.

Key Words: *Usher Syndrome; Pigmentary Retinopathy; Sensorineural; Hearing Loss*

INTRODUCTION

Usher syndrome is a rare genetic illness marked by retinitis pigments and sensorineural hearing loss at birth. It was named after Charles Usher, a Scottish ophthalmologist who established the disorder's hereditary aetiology and recessive inheritance pattern. It was initially reported by Von Graefe A, et al. in 1858. The clinical ophthalmic appearance of two brothers with Usher syndrome Type I is described here.

CASE REPORT

Two brothers, aged 24 and 26, reported to the Chittagong Eye Infirmary and Training Complex with symptoms of progressive vision loss in both eyes, particularly at night. Their medical history indicated a normal full-term delivery, as well as hearing loss and inability to communicate since childhood. A third sibling had never suffered any vision or hearing difficulties. In addition, none of their parents or any other near relatives had a history of a comparable disease. They had a best-corrected visual acuity of 6/36 in the right eye and 6/24 in the left eye, according to an ophthalmological examination. Both of their anterior portions were normal. Pupils were spherical, symmetrical, and responsive. In both brothers' eyes, fundus examination revealed numerous bony spicules in the retinal periphery and significant arteriolar attenuation.

Both of the optic discs were pallid. Both twins' intraocular pressures were 12mmHg in both eyes. Perimetry demonstrated shrinkage of the peripheral visual field in both eyes, as well as relative abnormalities in the paracentral area. Audiometry indicated a significant hearing loss in both ears after consulting with an ENT expert. Usher syndrome Type I was diagnosed based on the clinical symptoms. Both brothers were encouraged to have low vision aids to improve their quality of life, and the patient's family members were counseled about the disease's prognosis. Patients were recommended to see an ophthalmologist and an ENT expert regularly [1-4].

DISCUSSION

Retinitis pigmentosa and congenital sensorineural hearing loss are two symptoms of Usher syndrome. Usher syndrome is divided into three types: Types I, II, and III Type I is the most severe type of the condition, characterized by the substantial hearing loss that begins at birth and is followed by night vision problems in the first decade of life[5].

Type II is marked by significant hearing loss and the start of night vision issues later in life. With commencement in late adolescence, Type III is characterized by gradual hearing and night vision loss. RP is the predominant ophthalmic expression

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shared by all three varieties, despite variances in the auditory and vestibular function being the differentiating aspects. In our situation, the hearing loss was congenital, and each child struggled with night vision from a young age. As a result, these instances were labelled Usher type I. It impairs the patient's hearing and visual systems, making him or her functionally unable. Early diagnosis is critical for children with Usher syndrome, as it assists in patient and parent re-adjustment for good social functioning [8]. Multiple visual problems affect patients with Usher syndrome, including impaired contrast sensitivity, poor dark adaption, glare, and night blindness. These impairments might make it difficult to move, recognize objects, and do other activities of daily living. A basic penlight is highly effective for night blindness and dark adaptation challenges. For reading and writing, near vision aids such as hand-held LED magnifiers and closed-circuit TVs are useful. Appropriate genetic counselling for parents can raise awareness of the illness and assist to prevent it in the future[6-8].

CONCLUSIONS

In these circumstances, early identification and proper care will assist to retain residual eyesight as well as strengthen auditory and speech potentials. In these circumstances, an ophthalmologist's involvement involves diagnosis, therapy, and collaboration with other members of the treatment team.

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