# Usher syndrome

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## ABSTRACT

**Objective**

*To report a case of Usher syndrome at the East Avenue Medical Center.*

**Method**

*This is a case report.*

**Results**

*A 26-year-old female was diagnosed to have Usher syndrome based on the presence of retinitis pigmentosa and sensorineural hearing loss.*

**Conclusion**

*Patients with Usher syndrome associated with night blindness and deafness need appropriate supportive treatment and reassurance.*

USHER SYNDROME is an autosomal recessive congenital deafness associated with retinopathy indistinguishable from the typical retinitis pigmentosa (RP). Von Graefe first described the disorder in 1858, but it was Charles Usher, an ophthalmologist, who reported the condition as familial, representing a distinct entity. It is the most common syndrome associated with RP, with prevalence estimated at between 1.8 and 6.2 cases per 100,000.[1] It accounts for 50% of cases of those who are both deaf and blind.

Usher syndrome can be clinically subdivided into three types. Type 1 is characterized by profound congenital sensorineural deafness and resultant prelingual deafness or severe speech impairment associated with vestibular symptoms and childhood-onset retinopathy. Type 2 is characterized by congenital partial, nonprogressive deafness, absence of vestibular symptoms and milder, later onset retinopathy. Type 3 is characterized by progressive deafness starting late in the second to fourth decades, adult-onset retinopathy, and hypermetropia. Several population-based studies [1,2,3] showed that type 2 is more prevalent than type 1. The least common type 3 has been reported mostly among those of Finnish descent.[4]

We are reporting a case of type 2 Usher syndrome in a 26-year-old female who sought consultation for bilateral blurring of vision that started in childhood associated with nyctalopia and decreased hearing in both ears. Other organ systems were unremarkable. Family history revealed that maternal grandmother and aunt have the same symptoms of hearing loss and blurring of vision.

Visual acuity was 6/30 (20/100) in the right eye (OD) and counting fingers at 3 feet not improved with refraction in the left eye (OS). A 1+ posterior subcapsular cataract was present in both eyes. Fundus findings revealed generalized arteriolar attenuation of the retinal vessels with generalized intraretinal pigmentation and loss of pigment from the retinal pigment epithelium (RPE), and macular atrophy in both eyes (OU). There was waxy optic-disc pallor with distinct borders OU. Fluorescein angiography showed increased visibility of background choroidal hyperfluorescence over the whole retina especially the macular areas. The hyperfluorescence increased in intensity and then faded at later phases consistent with RPE-window defects. Standard achromatic perimetry (Octopus 101, Octopus Corp., Bern, Switzerland) showed marked constriction of the central field with ring-like central scotoma OU (Figure 1). Electroretinography (ERG) showed no recordable responses in both dark- and light-adapted conditions indicating extinguished rod and cone responses typical of retinitis pigmentosa (Figure 2). Pure tone audiometry revealed a sensorineural hearing loss on both ears with an average threshold of 41.67 dB for the right ear and 30.0 dB for the left ear.

Nyctalopia or night blindness is the hallmark symptom of RP usually present in the first or second decade of life. Narrowing of the visual field is another feature characterized by insidious progressive contraction and loss of peripheral visual field. The earliest defects on kinetic perimetry are relative scotomas in the mid-periphery usually superiorly between 30 and 50 degrees from fixation which enlarge, deepen, and coalesce to form a ring field that enlarges toward the periphery. Generally, the visual-field loss between the two eyes tends to be symmetric, most severe in the superior field as seen in this case.

A common misconception in RP is that central vision will be intact until most, if not the entire, peripheral field is lost. Central visual function, however, can be seriously affected early while significant peripheral field remains. Cystoid macular edema, diffuse retinal vascular leakage, macular preretinal fibrosis, and RPE defects in the macula can occur causing central visual loss as seen in this patient.

Visual acuity appears to be better retained in older patients with type 2 as compared to type 1 Usher syndrome. Visual acuity of 20/40 (6/12) or better at age 29 is seen in 69% of type 1 and 94% in type 2.[2]

Fundus findings may include attenuated retinal vessels, mottling and granularity of the RPE, bone spicule intraretinal pigmentation in the mid-periphery and optic-nerve-head pallor. Atrophy of the RPE and choriocapillaries leads to fundus pallor and larger choroidal vessels

become visible. In some cases, macular edema or preretinal fibrosis and bilateral atrophic macular lesions can be appreciated.

Congenital neurosensory deafness and retinitis pigmentosa might seem to have little in common pathologically. However, the photoreceptors of the retina and the hair cells in the inner ear are both ciliated neuroepithelial cells. The cilia of the inner ear and the photoreceptors may share axonal components necessary for the formation of ciliated structures and whose genes would be candidates for the defective gene in Usher syndrome.

The diagnosis of Usher syndrome must be confirmed with ERG. Other syndromes that can be associated with deafness and pigmentary retinopathy are infantile and adult Refsum's disease, Cockayne's syndrome, Bardet-Biedl syndrome, Alstrom's disease, Flynn-Aird syndrome, Friedreich's ataxia, and Keans-Sayne syndrome.

Follow-up should include assessment of visual field by standard achromatic perimetry at least every 2 years. Other work-up procedures include dark adaptometry, retinal densitometry, and ERG. Patients with RP, when tested with dark adaptometry, may show elevation of the cone segment, the rod segment, or both to varying degrees. Retinal densitometry is used to measure effective photoreceptor photopigment density where estimates can be made of the rates of photopigment regeneration. ERG responses in an RP patient can range from abnormal to undetectable. In studying the natural course of RP, Berson et al.[5] found that patients lost an average of 16 to 18% per year of remaining ERG amplitude to bright white flashes. Massof and Filkenstein[6] found that the scotopic rod-dominated ERG was affected to a much more severe degree than the photopic cone-mediated ERG in type 1 RP, whereas the scotopic and photopic ERGs were more equally abnormal in type 2 RP.

A major fallacy in the management of RP is the common assertion that the condition is untreatable; it is more accurate to say that RP is incurable. It should not be forgotten that aiding the patient to become adjusted to RP by supplying useful information and support can do a great deal of good. Periodic visual-field examination with compassionate explanation of visual-field defects can help patients appreciate the rate of progression and plan for disability. Reassurance that the changes seen are typical or usual for patient with RP often allays fears that they are losing visual function at a rate faster than expected. Correction of refractive error, cataract extraction when indicated, treatment of macular edema when present, and use of low-vision and hearing aids are appropriate treatment options to consider. This patient was given the option of using low-vision aids (magnifying lenses) and a hearing aid, and was able to finish college.

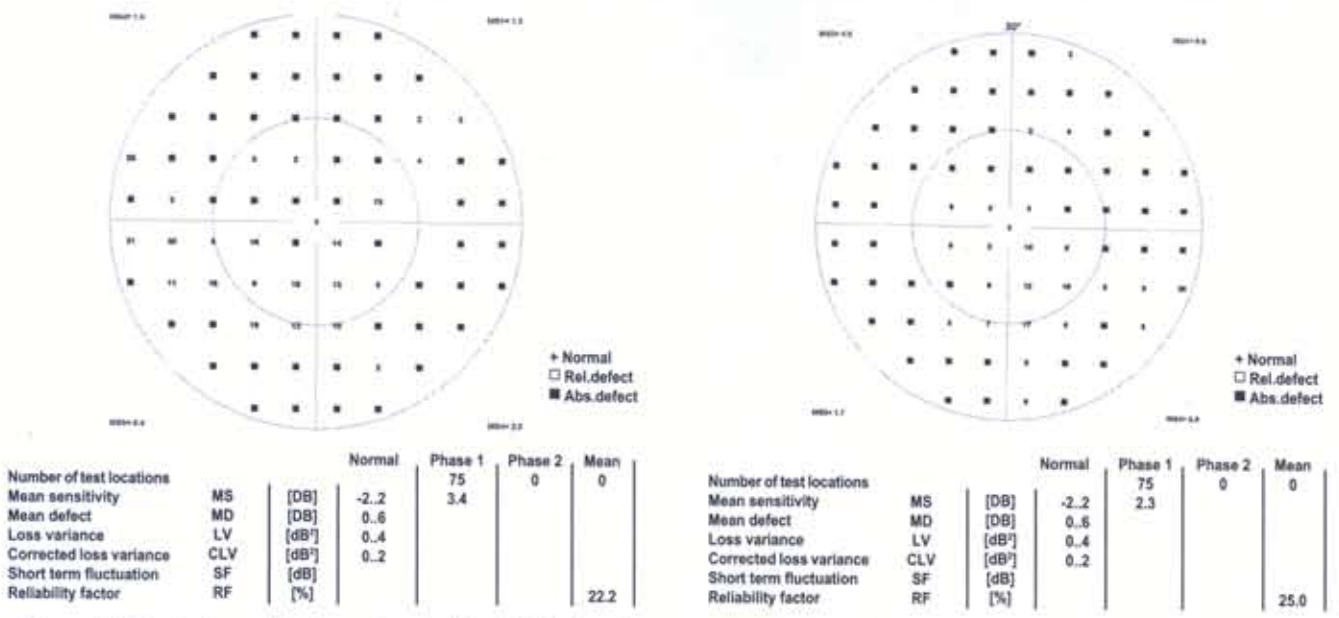


**Figure 1.** Marked constriction of central field with ring-like central scotomas in both eyes.

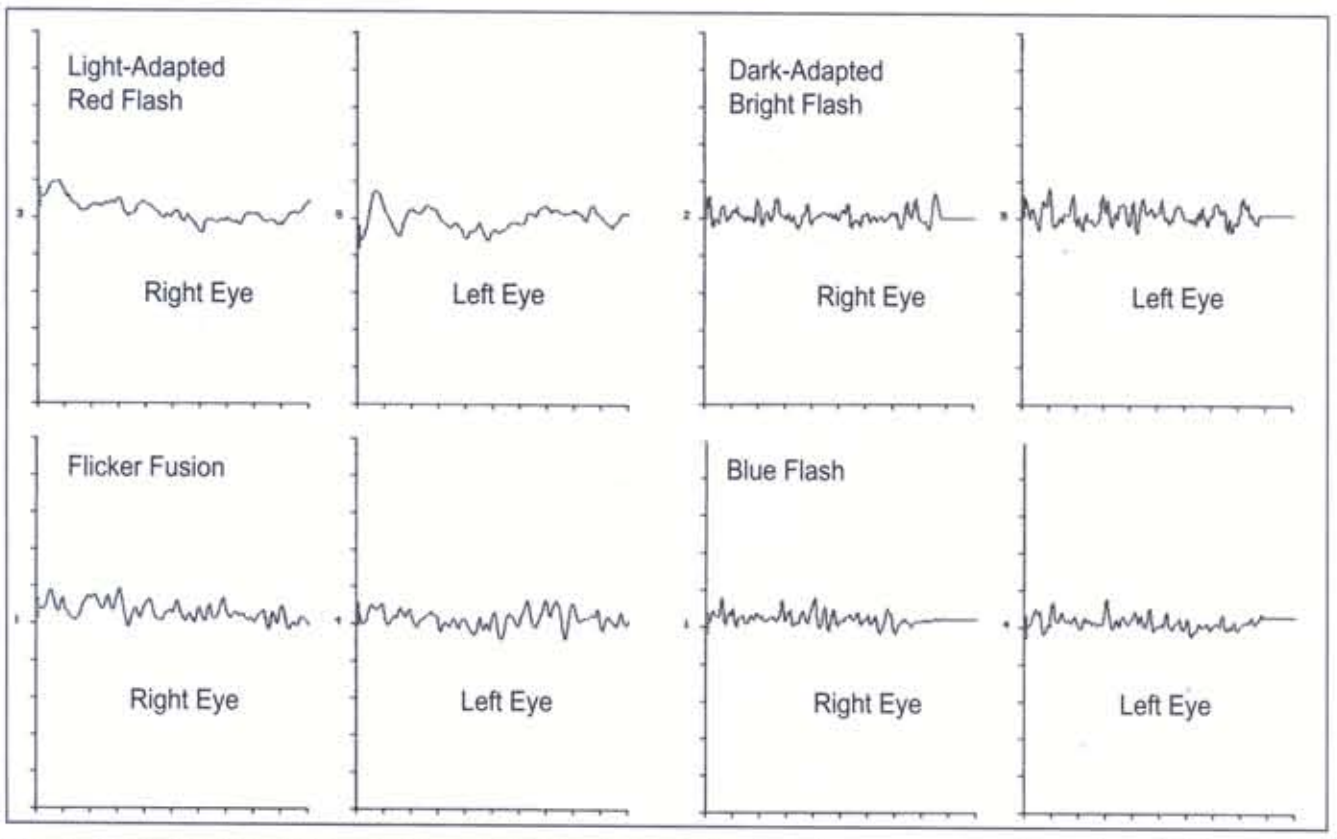


**Figure 2.** Electroretinographic findings showing extinguished rod and cone responses in dark- and light-adapted conditions.

**References**


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