A 68-Year-Old Female with Blindness and Deafness

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Introduction:

A 68-year-old female presents as a consult for retinal evaluation for ongoing poor vision for a couple of weeks from a possible retinal dystrophy in both eyes. The patient's past medical history is significant for cochlear implants in both ears for sensorineural hearing loss. Her family history is notable for a sister with blindness and father with retinal detachment. Her medication history is non-contributory.

Exam:

Best corrected visual acuity was hand motion in both eyes. Intraocular pressures were within normal limits. No afferent pupillary defect was appreciated. Visual fields were constricted peripherally and extraocular motility was full in both eyes. Anterior segment exam was notable for a posterior chamber intraocular lens in both eyes.

Dilated fundus examination revealed waxy pallor of both optic nerves with diffuse bone spicules and RPE changes peripherally and a central island of retinal tissue in the macula (Figure 1).

Discussion:

Given the progressive pigmentary retinopathy associated with deafness and family history of blindness, the patient was given a diagnosis of Usher syndrome, an autosomal recessive condition with deafness and retinopathy indistinguishable from retinitis pigmentosa.[1]

Prevalence ranges between 1.8-6.2 cases in 100,000 with 50% of individuals with blindness and deafness being diagnosed with Usher syndrome. It was described by

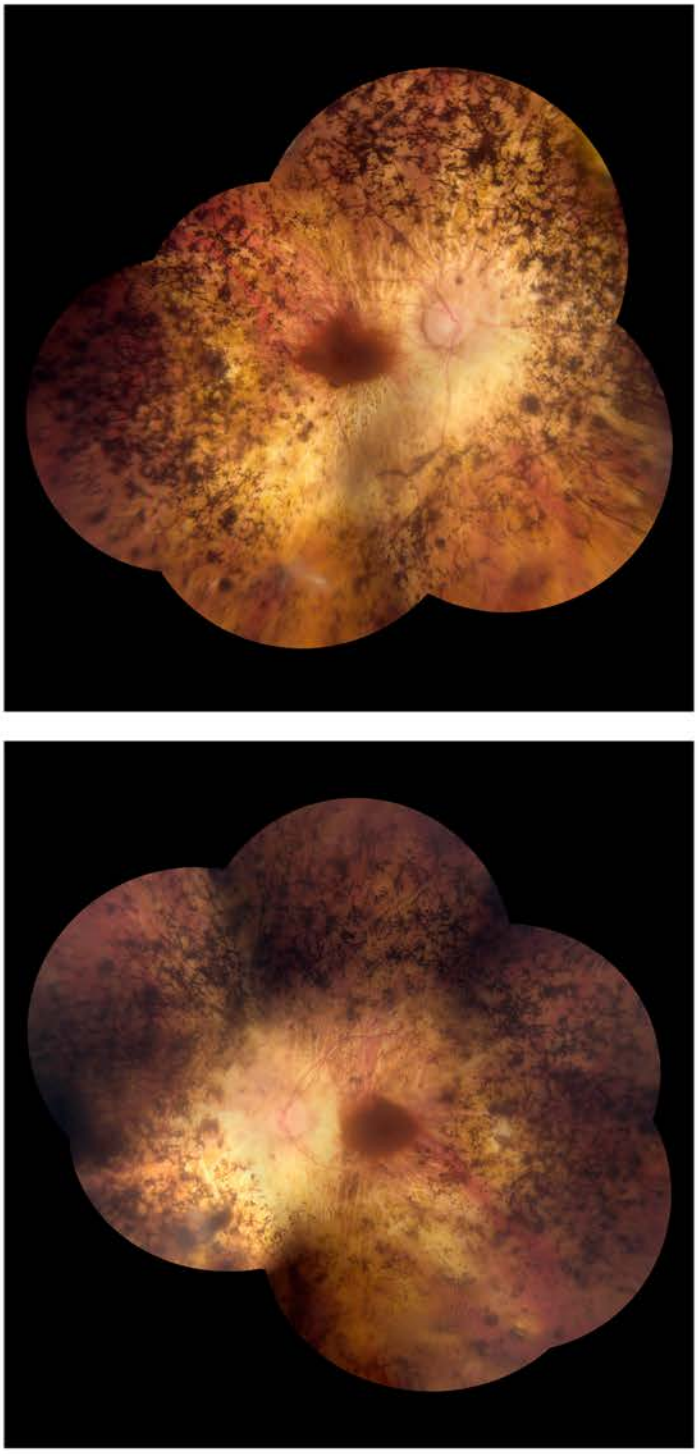

Figure 1. Dilated fundus examination with waxy pallor of both optic nerves and diffuse bone spicules and RPE changes peripherally and a central island of retinal tissue in the macula.

the British ophthalmologist Charles Usher who recognized the familial nature of the condition. He reported predominantly two types based on severity of deafness, age of onset and progression of vision loss. Type 1 Usher syndrome has symptoms of night blindness occurring in the 1st-2nd decade with profound deafness resulting in speech impairment and vestibular dysfunction. On the other hand, Type 2 Usher Syndrome has later onset night blindness in the 3-4th decades with partial deafness.[1]

One theory as to the pathogenesis of the syndrome is thought to be related to mutations in USH genes that affect the morphogenesis of hair cell bundles in the inner ear as well as in the connecting cilium of the outer segments of photoreceptors. However, there are conflicting reports in the literature regarding this theory.[2]

The diagnosis is primarily clinical. There is little utility of imaging except if macular edema is suspected. In the case of pediatric patients, genetic testing and ERG, which is abnormal in Usher syndrome, can be used to confirm the diagnosis and counsel the patient's family.[1]

Unfortunately, there is little at this time that can be done to prevent progression of vision loss in patients with Usher syndrome. However, there is a clinical trial underway to see if a viral based delivery of the MYO7A gene could treat patients with type 1B Usher syndrome.[3] Given the dual impairment of deafness and decreased vision, a great deal of support is needed for these patients to maintain their ability to remain independent. As in our patient, early implantation of cochlear implants have shown success in allowing young children to hear and support speech development.[4] Because receptive and expressive language is most closely correlated with the precocity of cochlear implantation, the diagnosis of Usher syndrome should be established as early as feasible to optimize speech therapy.

References:

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