Case Report

Nongranulomatous anterior uveitis in a patient with Usher syndrome

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Abstract

A 34-year-old female with Usher syndrome, but no family history of similar illness, presented with complaints of vision reduction, redness, and photophobia. Biomicroscopic examination showed mildly injected conjunctivae bilateral, small, round keratic precipitates; bilateral +2 cells with no flare reaction in the anterior chamber; and bilateral posterior subcapsular cataracts. No associated posterior synchiae, angle neovascularization, or iris changes were detected; normal intraocular pressures were obtained. Fundus examination demonstrated waxy pallor of both optic nerves, marked vasoconstriction in retinal vessels, and retinal bone spicule pigment formation, with a normal macula. Electroretinography confirmed the diagnosis of retinitis pigmentosa, optical coherent tomography was normal and otolaryngology consultation was conducted.

To our knowledge, an association between Usher syndrome and bilateral nongranulomatous anterior uveitis has not been previously reported, and our purpose is to report this association.

Keywords: Retinitis pigmentosa, Usher syndrome, Uveitis: Anterior uveitis

Introduction

Usher syndrome is an association of retinitis pigmentosa (RP) and congenital sensorineural hearing loss. The exact incidence of Usher syndrome has been difficult to determine, but surveys of RP patients have suggested that about 10% are profoundly deaf, and ophthalmic examinations of children in deaf schools have revealed that approximately 6% have RP. Usher syndrome is the most common cause of deafness and blindness and is responsible for half of all cases of combined deafness and blindness. The prevalence of Usher syndrome is thought to be 3 cases per 100,000 people.1–3

Uveitis is commonly subdivided histologically and clinically into two main categories: granulomatous and nongranulomatous. Nongranulomatous uveitis typically has an infiltration of lymphocytes and plasma cells, whereas granulomatous reactions also include epithelioid and giant cells. The most common form of nongranulomatous anterior uveitis is acute anterior uveitis (AAU), which is associated with the HLA-B27 allele in half to two thirds of patients. Although the cause is usually unknown, certain ocular and systemic diseases may be the underlying cause of the iritis, or iridocyclitis.2

Here, we report after PUBMED search a case of Usher syndrome with bilateral nongranulomatous anterior uveitis, which was not reported before.

Case report

A 32-year-old female with a known case of Usher syndrome has been followed up in the clinic since the age of 17 years. At her initial hospital presentation, she had...
decreased vision, especially at night, and partial deafness. Her eye examination at that time revealed the following: visual acuity of 20/40 in the right eye and 20/30 in the left eye and intraocular pressure (IOP) measurements within a normal range in both eyes. Slit lamp examination showed the following in both eyes: a quiet conjunctiva, a clear cornea, a deep and quiet anterior chamber, a clear lens, a pale disk, arteriolar attenuation, and the presence of bone spicules (Fig. 1). The diagnosis was confirmed by electrerotinography using flash stimuli (no reproducible response could be obtained in both eyes) (Fig. 2). Visual field testing was performed, showing generalized field constriction in both eyes. The patient was referred to the Ear, Nose, and Throat (ENT) clinic for an assessment of her hearing problem. A diagnosis of Usher syndrome was confirmed in the patient.

Thirteen years later, the patient presented with complaints of redness, photophobia and blurred vision. On examination, the visual acuity was the same, the IOP measurements were within the normal range in both eyes, the conjunctivae were mildly injected, and the corneas were clear in both eyes. Bilateral +2 cells with no flare reaction in the anterior chamber (SUN working group classification) and early bilateral posterior subcapsular cataracts (Fig. 3A) were detected. Fine keratic precipitates (KPs) (i.e., corneal endothelial deposits) were found in both eyes (Fig. 3B). The iris appeared normal, and no posterior synechiae were detected. The retina was the same as before, with no evidence of vitritis, vasculitis, or retinitis. Based on these examination results, the patient was diagnosed with bilateral nongranulomatous anterior uveitis.

A uveitis workup was performed, including a complete blood count (CBC), erythrocyte sedimentation rate (ESR), C-reactive protein level, antinuclear antibody test, HLA-B27 test, HLA-B5 test, angiotensin-converting enzyme (ACE) test, a chest X-ray, computerized tomography of the chest, tuberculin or purified protein derivative (PPD) test, Venereal Disease Research Laboratory (VDRL) test, and toxoplasma serology. The results of all these tests were within normal limits. The patient was treated with a topical corticosteroid, prednisolone acetate1% (Pred Forte), 4 times a day and was followed up in the clinic. Steroid tapering was carried out with subsequent follow-up.

The patient has received regular follow-up examinations in the uveitis clinic for the past 3 years. During this time period, she has had 4 episodic exacerbations of bilateral nongranulomatous anterior uveitis and has responded nicely to topical steroid treatment each time. Her best-corrected visual acuity at the most recent follow-up examination was 20/80 in the right eye, which improved to 20/60 with pinhole testing, and 20/100 in the left eye, which improved to 20/60 with pinhole testing. The decreased visual acuity was attributed to the presence of +2 posterior subcapsular cataract in both eyes. No active inflammation has occurred during the past 3 months. The patient is currently undergoing treatment with topical prednisolone acetate1% (Pred Forte) once daily for both eyes. Neither vitritis nor macular edema has been detected. She is booked for cataract surgery in 2 months, providing that her condition remains under control and an exacerbation of uveitis does not occur.

Discussion

Usher syndrome is the name given to the association of RP and congenital sensorineural hearing loss, whether partial or profound. Ophthalmologists should be attuned to patients with RP who present with a nasal intonation to their speech or wear hearing aids and should ask these patients when

Figure 1. Color fundus photos showing classic features of retinitis pigmentosa (RP), pale disk, arteriolar attenuation, and bone-spicule retinal pigment epithelial changes in both eyes.

Figure 2. Electroretinograms (ERGs) for both eyes, showing that no reproducible response could be obtained.
a particularly stronger association of FHU with Usher syndrome type 2; the causes for this significant association are not yet clear, but patients with RP and those with uveitis exhibit several common clinical and laboratory inflammatory features. In addition, Turan-Vural et al.\textsuperscript{7} reported a case of Usher syndrome in association with unilateral FHU.

Lichtinger et al.\textsuperscript{7} also demonstrated that some patients with RP have circulating B cells that are reactive with the retinal antigens, especially the S antigen. This finding may explain the tendency for immune reactions in patients with RP, which could increase their susceptibility for developing uveitis.\textsuperscript{10} A study by Chowers et al.\textsuperscript{8} described the association between FHU and RP and showed that patients with RP can develop autoimmune reactions to anterior chamber antigens that bear some similarity to retinal antigens, leading to the clinical manifestation of FHU. Other studies have also described the association between RP and FHU.\textsuperscript{11–14}

In our case, all possible causes of anterior uveitis were excluded, either by clinical examinations or by negative investigations. A herpetic cause of iridocyclitis is less likely in this case because of the presence of bilaterality, which is rare in herpetic uveitis. In addition, no herpetic corneal signs and no glaucoma or iris atrophy were detected. The patient also possessed normal corneal sensation. Fuchs’ heterochromic iridocyclitis is less likely in this case because the KPs were round (not stellate shaped), there was no evidence of heterochromia, and, more importantly, there was a complete response to topical steroid treatment, which is not typical in cases of FHU.

To the best of our knowledge, this is the first reported case of Usher syndrome associated with chronic bilateral nongranulomatous anterior uveitis. Therefore, it is important for ophthalmologists to recognize that, although rare, nongranulomatous anterior uveitis can be found in association with RP and, more specifically, with Usher syndrome. This may influence the follow-up and management plan for these patients, especially if complications have occurred.

Conflict of interest

The authors declared that there is no conflict of interest.

References


