Bilateral Retinal Problem in a Patient With Alport Syndrome.

Sundeep K. Kasi  
*Thomas Jefferson University*

Murtaza K Adam  
*Thomas Jefferson University*

David S Ehmann  
*Thomas Jefferson University*

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A man in his late 20s with a history of Alport syndrome presented on referral for a bilateral retina problem after experiencing right-sided herpes zoster ophthalmicus. He denied risk factors for human immunodeficiency virus, and serologic testing results were negative. He had completed treatment for keratouveitis and reported some residual blurring of his vision in the right eye. His ocular history was unremarkable. His uncorrected visual acuity was 20/60 OD and 20/40 OS. Pupillary responses, confrontation visual fields, motility, external examination, and intraocular pressure were normal. Slitlamp examination revealed superficial keratitis and anterior stromal haze in the right eye but otherwise was normal. Fundus examination (Figure, A) revealed a poor foveal light reflex with an irregular-shaped area of red discoloration in the fovea and surrounding subtle flecking of the retina in both eyes. No other abnormalities were found on ophthalmoscopic examination. Spectral-domain optical coherence tomography (Figure, B) showed an irregular foveal contour with trace epiretinal membrane formation, with no evidence of foveoschisis or outer retinal changes. Fundus autofluorescence was normal.

**WHAT WOULD YOU DO NEXT?**

A. Perform pars plana vitrectomy with internal limiting membrane peeling

B. Perform aqueous/vitreous tap for viral polymerase chain reaction and inject foscarnet intravitreally

C. Observe and repeat spectral-domain optical coherence tomography in 6 months to 1 year

D. Perform intravitreal injection of ocriplasmin
Dystrophy, corneal clouding owing to accumulation of mucopolysaccharides, conjunctival telangiectasia, anterior lenticonus, cataract, and retinal flecks as well as macular holes. These findings are often absent in childhood and become more apparent with aging.

While dot and fleck retinopathy is the most common ocular sign of Alport syndrome (85%), a range of macular pathology from lamellar macular holes to giant macular holes have been reported as well as vitelliform lesions and foveal and midperipheral retinoschisis.

It is suspected that these macular pathologies may be secondary to abnormalities in type IV collagen, causing weakness of the internal limiting membrane. Others postulate that abnormal type IV collagen in the Bruch membrane may allow collection of intraretinal fluid within the retina prior to hole formation or that an anomalous vitreoretinal interface may also contribute to vitreomacular interface abnormalities.

Although the development of these macular abnormalities is not well understood, a few observational case reports have shown that lamellar macular holes can transform into full-thickness macular holes. Interestingly, Shah et al documented the coalescence of multiple small macular holes into a single large macular hole. Progressive vision loss is a distinct possibility in patients with Alport syndrome with early macular changes such that monitoring with fundus examination as well as optical coherence tomography might be appropriate in some cases where disruption of the outer retina or full-thickness macular hole formation may improve with internal limiting membrane peeling.

**Patient Outcome**

The patient was informed of the association of Alport syndrome and lamellar macular hole and asked to return with any new symptoms of metamorphopsia or reduced vision. He was followed up, and repeated refraction was performed after the corneal haze improved with best-corrected visual acuity of 20/30 OU.

**REFERENCES**