New Floaters in a Child Poked in the Eye

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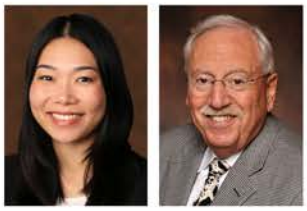


Introduction:

A 6-year-old male presented to our clinic with new onset of floaters in the left eye. He had been poked in the left eye by a finger of another child prior to the onset of the floaters. He was otherwise healthy. Family history was significant for a maternal grandmother with a retinal detachment and maternal grandfather with age related macular degeneration. Visual acuity was 20/40 in the right eye and 20/50 in the left eye. Intraocular pressures were normal in both eyes. Anterior segment exam was unremarkable. Fundus exam in the right eye was normal, while the left eye revealed mild vitreous hemorrhage with macular schisis as well as inferotemporal retinoschisis with inner retinal holes.

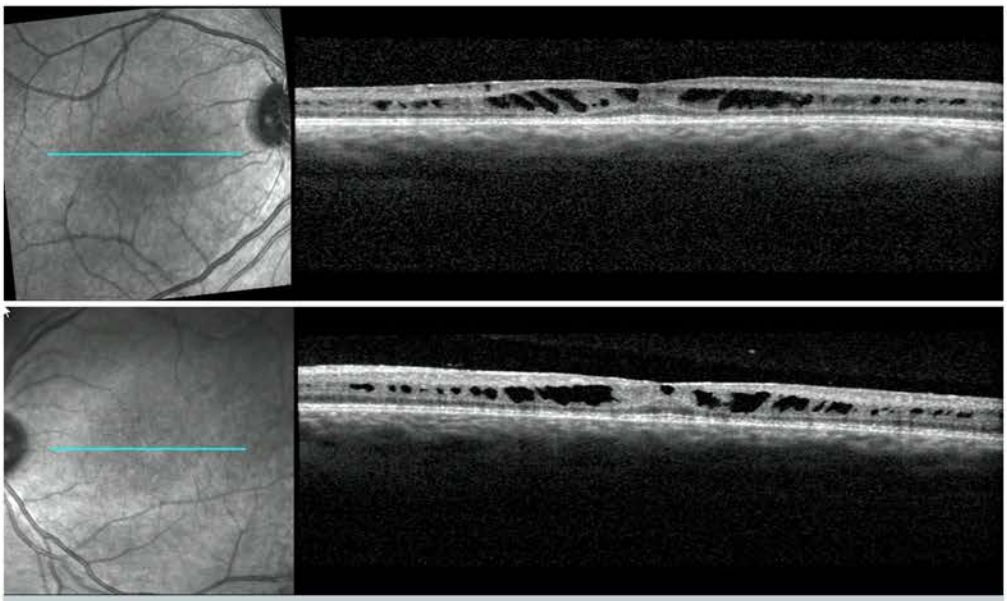


Figure 1: Optical Coherene Tomography of both eyes showed schisis cavities in the nuclear and outer plexiform layers with a blunted foveal contour.

To aid in the diagnosis, an OCT and widefield photos were obtained.

OCT of the macula showed schisis cavities in the inner nuclear and outer plexiform layers with a blunted foveal contour in both eyes. (Figure 1) The inferotemporal retina of the left eye showed similar OCT findings.

Widefield photos revealed a spoke wheel pattern in the fovea of both eyes. (Figure 2) The peripheral retina in the right eye was normal. The inferotemporal periphery of the left eye showed retinoschisis with several inner retina holes. (Figure 3)

Genetic testing revealed a hemizygous TTA>TAA nucleotide substitution which caused an amino acid change in the coding sequence of the RS1 gene. This was consistent with the diagnosis of X-linked retinoschisis.

Discussion:

X-linked retinoschisis, also known as juvenile retinoschisis, is a rare, congenital disease manifesting as bilateral splitting of the retina in males. It is caused by mutations in the RS1 gene, which encodes retinoschisin, a protein expressed by photoreceptors. Retinoschisin is involved in intercellular adhesion and cellular organization of the retina.[1] Inheritance is X-linked recessive with complete penetrance and variable expressivity.[2] Heterozygous females are rarely affected but cases have been reported.[3,4,5] The prevalence is estimated to be about 1 in 5,000 to 1 in 20,000.[2,6]

There is symmetric, bilateral macular involvement in the first decade of life, and the peripheral retina may also be affected in about half of patients.[7] Splitting of the retina was initially reported to be in the nerve fiber layer, but SD-OCT imaging has shown that splitting

most commonly involves the inner nuclear layer and sometimes the outer nuclear layer and outer plexiform layer.[1]

Patients typically present at school age with poor vision but can also present in infancy with strabismus, hyperopia, nystagmus, vitreous hemorrhage or retinal detachment. Presenting visual acuity is usually 20/60 to 20/120. Vision may worsen slightly in the first to second decades but then remains stable until the fifth to sixth decade when slow, progressive retinal atrophy may occur. If peripheral retinoschisis is present, it is usually inferotemporal, and the patient will have a corresponding absolute scotoma.[7,8]

Figure 2: Widefield photos revealed a spoke wheel pattern in the fovea of both eyes.

Fundus exam reveals a spoke wheel or stellate pattern radiating from the fovea with a dome-like elevation of a layer of the retina.[8] White flecks, pigmentation, vessel sheathing, vascular attenuation, inner layer holes, vitreous veils, or subretinal linear fibrosis may be present. [2,9]

OCT findings include schisis cavities, retinal atrophy, small perifoveal cysts with larger foveal cystic spaces. Flattening of the cysts can occur after adolescence at which time the cysts may not be evident. Fluorescein angiogram may be normal in younger boys and may show late pooling of the dye in the cystic cavities without petaloid leakage in older individuals. A full field ERG may show a reduced b-wave and a preserved a-wave (negative waveform) in more than half of affected males. The diagnosis can be confirmed with genetic testing for mutations in the RS1 gene.[7]

In managing young X-linked retinoschisis patients, it is important to perform an annual dilated exam, counsel the patient to avoid head trauma and high contact sports, and to treat any amblyopia, refractive error, or strabismus if applicable. Genetic counseling should be performed as affected males will pass the pathogenic variant to their daughters but not their sons, and heterozygous females have a 50% chance of transmitting the pathogenic variant. [7] Carbonic anhydrase inhibitors may improve the schisis in up to two-thirds of patients.[1] Gene therapy clinical trials with an AVV-RS1 vector showed closure of schisis cavities in one individual but also caused ocular inflammation.[10] Another clinical trial showed that gene therapy led to increased inflammation and a lack of positive treatment signs.[7]

By the sixth to seventh decade, vision may decrease to 20/200 or worse.[7] Complications include vitreous hemorrhage, rhegmatogenous retinal detachment, neovascular glaucoma, macular dragging, or optic atrophy. Stable peripheral retinoschisis

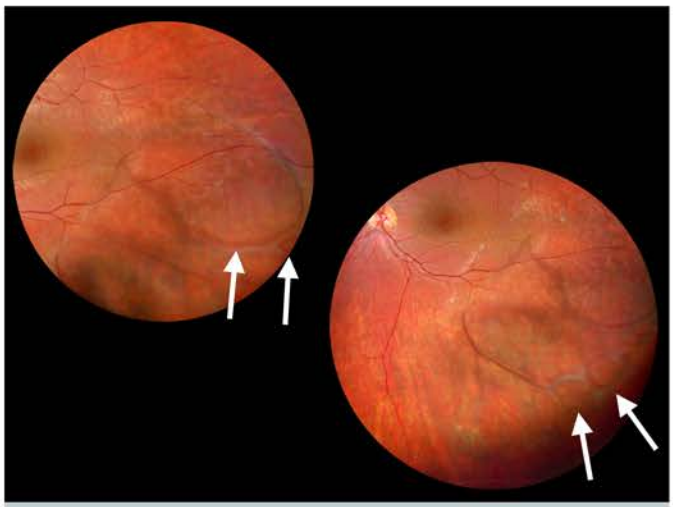
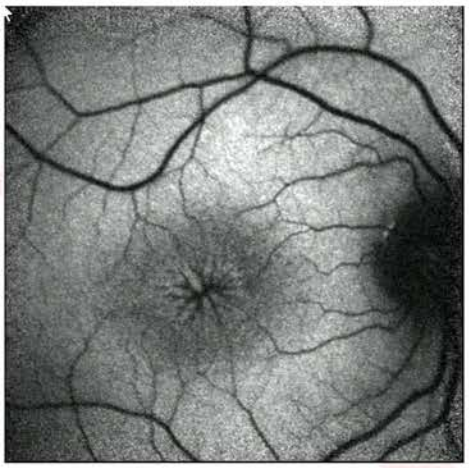
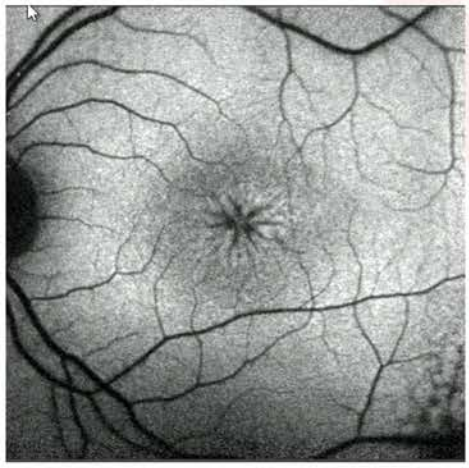


Figure 3: The left eye showed retinoschisis with several inner retina holes (arrows) in the inferotemporal periphery.

that does not threaten the macula is usually observed. Vitreous hemorrhage and RRD may require surgery. Laser in these patients is controversial.[2,10]





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