Short Subjects

Vitreous Hemorrhage as the Initial Manifestation of X-Linked Retinoschisis in a 9-Month-Old Infant

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Abstract. A 9-month-old infant presented with progressive esotropia, bilateral vitreous hemorrhages, bullous retinoschises, and peripheral retinal detachments. X-linked retinoschisis was diagnosed on the basis of electroretinogram findings. We report a case of vitreous hemorrhage as the initial presentation of X-linked retinoschisis in one of the youngest patients discussed in the literature.


INTRODUCTION

X-linked juvenile retinoschisis is a rare disorder of retinal development that results from a split in the superficial layers of the retina, between the inner limiting membrane and the nerve fiber layer.¹ The average incidence of X-linked retinoschisis is approximately 1 case per 10,000 individuals.² Various phenotypic variations can occur. In 1997, the gene responsible for X-linked juvenile retinoschisis was identified and mapped to a segment on the short arm of the X chromosome (Xp22.2).³ Since then, more than 100 mutations of this gene, termed XLRS1, have been reported.⁴

Clinically, juvenile retinoschisis develops early in life. Bilateral retinal changes are noted. Spoke-wheel, cystic-like schisis changes are present in the fovea that coalesce with time.⁵ Foveal schisis is reported to be present in 70% to 100%⁶-⁸ of patients, whereas peripheral retinoschisis is present in 50% to 60% of patients.⁶,⁸,⁹ Most patients present with progressive visual impairment that is usually detected in school between 5 and 6 years of age.⁴ A few present in infancy with squint, nystagmus, and highly elevated bullous retinoschisis.⁵,⁹ The mean age at onset of symptoms is 4 to 5 years⁴ and a bimodal distribution is seen at onset. Patients who manifest squint and nystagmus are usually seen in infancy at a mean age of 1.8 years.⁶ Patients who have only poor vision as a symptom of X-linked retinoschisis are seen later in life at a mean age of 6.7 years.⁶ Later stages include complications such as vitreous hemorrhage, choroidal sclerosis, and retinal detachment.¹ Vitreous hemorrhage associated with juvenile retinoschisis usually occurs in older children. A literature review yielded an age range of 18 months to 15 years.⁴,¹⁰-¹² We report a case of vitreous hemorrhage in a 9-month-old infant due to X-linked juvenile retinoschisis.

CASE REPORT

A 9-month-old male infant born at term presented with esotropia. His medical history was unremarkable. His parents had noted progressive esotropia and a white pupillary reflex since he was 6 months old. They had also noted that he rubbed his eyes constantly during this time.

Ocular examination revealed alternating esotropia. He could not fix and follow from either eye. He did, however, respond to light in both eyes. Red reflex was absent. No afferent pupillary defect was present. Examination under anesthesia revealed normal anterior segments with no synechiae or anterior chamber reaction. The lenses were clear bilaterally. Yellow vitreous debris was noted in both eyes. Fundus examination was limited due to old, diffuse vitreous hemorrhage. The view to the superior fundus was much clearer. There, bullous retinoschisis with vitreous membranes and a shallow retinal detachment was noted in both eyes.
Ultrasonography of both eyes revealed bulbus retinoschisis with shallow retinal detachments. Moderate vitreous debris was present in both eyes. Electroretinogram of the left eye revealed an electronegative response with reduced b-wave disproportionate to a-wave changes, suggesting the diagnosis of X-linked retinoschisis or Goldman–Favre disease. An electroretinogram could not be performed on the right eye because the patient was uncooperative.

No family history of retinoschisis, retinal detachment, or poor vision was elicited. The parents underwent a dilated retinal examination that revealed no gross evidence of retinoschisis or any subtle foveal changes. Surgical intervention was recommended for diagnostic and therapeutic management. The patient underwent pars plana vitrectomy, scleral buckle, and lensectomy in the right eye. Pars plana lensectomy was performed to avoid peripheral bulbus retinoschisis. Vitreous membranes were noted to cause tractional retinal detachment. No retinal tears were present. A scleral buckling component 41 (Mira, Inc., Uzbridge, MA) was used to relieve peripheral traction. The left eye underwent pars plana vitrectomy and a scleral buckling component 41 after 2 weeks. The retinal detachments resolved in both eyes. However, large inferotemporal retinoschisis has remained bilaterally. Aggressive alternating patch therapy has been used for amblyopia. At the most recent examination, both eyes were noted to fix and follow with the infant preferring the right aphakic eye.

**DISCUSSION**

Most patients with X-linked retinoschisis present during the preschool years. They are usually identified when they have difficulty reading secondarily to foveal abnormalities. Occasionally, patients are seen early in life due to amblyopia-induced deprivation strabismus. Our patient’s early presentation with leukocoria was unusual. The parents had noted a white pupillary reflex at approximately 6 months of age.

Leukocoria is a rare but serious symptom that may present at birth or develop during infancy. It occurs with several ocular disorders, retinoblastoma being the most common and urgent because it can be life threatening. Other possible causes, in order of frequency, include persistent hyperplastic primary vitreous, Coats’ disease, ocular toxocariasis, retinopathy of prematurity, and retinal hamartomas. In our case, the etiology of bilateral leukocoria was organized vitreous hemorrhage due to X-linked retinoschisis, a rare etiology in infancy. Vitreous hemorrhage is uncommon in the first year of life and is mainly seen in association with retinopathy of prematurity, shaken baby syndrome, or retinoblastoma. Typically, vitreous hemorrhage as a sign of X-linked retinoschisis occurs later in life, usually after the disorder has been diagnosed.

The most severe vision-threatening complications of X-linked retinoschisis include retinal detachment and vitreous hemorrhage. Various small series have reported the incidence of vitreous hemorrhage associated with X-linked retinoschisis to be 21% to 40%. Multiple inner retinal holes are present in these patients. Retinal vessels present in the inner layer bridges cross the schisis cavity. Disruption of these unsupported vessels due to vitreous traction can lead to intraschisis or vitreous hemorrhage. It usually resolves spontaneously without causing a loss of vision.

Vitreous hemorrhage stimulates the formation and contraction of vitreous membranes that can lead to tractional retinal detachment. If outer retinal holes occur in the presence of the inner retinal holes in the nerve fiber layer, retinal detachment can occur. The incidence of retinal detachment ranges from 0% to 22%. Current treatment of retinal detachment includes scleral buckling, vitrectomy, or a combination of both, with varying success rates.

If vitreous hemorrhage is diagnosed without an associated retinal detachment, spontaneous resolution usually occurs and frequent ultrasonographic examinations are performed. However, in younger patients, there is concern over the development of amblyopia and surgical intervention is necessary to remove vitreous blood.

**REFERENCES**


