Spontaneous Rupture of the Lens Capsule in a Case of Marshall Syndrome

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INTRODUCTION

Marshall syndrome is a rare autosomal dominant syndrome that was described initially in 1958.1 Approximately seven multigenerational families have been reported since.2 Short stature, sensorineural hearing loss, and a characteristic facial appearance with hypoplastic nasal bones characterize the syndrome. The systemic manifestations of Marshall syndrome have been noted to overlap considerably with those of Stickler syndrome. Given this phenotypic overlap, several investigators have suggested that the two syndromes are probably allelic expressions of the same gene.3 However, other investigators believe that these two conditions are distinct. The genetic defect in Marshall syndrome has been recently identified as a mutation in the COL11A1 gene. The product of this gene is a type of collagen polypeptide (alpha 1), which has an important role in skeletal and other structural morphogenesis.4

The ocular manifestations of Marshall syndrome include myopia, vitreoretinal degeneration, cataract, and lens subluxation. The onset of spontaneous cataract with acute glaucoma has been previously described in Marshall syndrome. To date, fewer than 40 cases have been described in the literature with spontaneous rupture of the lens capsule. Only one documented case was treated with vitreous surgery.5

CASE REPORT

We examined a 10-year-old girl with a known diagnosis of Marshall syndrome since the age of 3 years. At the age of 2 years, she was noted to have an axial myopia of 22 D and bilateral lattice degeneration. Based on clinical findings of high myopia, sensorineural hearing loss, characteristic facial appearance with mid-facial hypoplasia (Fig. 1), and lack of arthropathy, the diagnosis of Marshall syndrome was made.

Recently, she presented to us with a 1-week history of nausea and vomiting. There was no history of trauma. An ophthalmic examination of the right eye revealed a hypermature cataract with lens fragments in the anterior chamber (Fig. 2). Intraocular pressure was 56 mm Hg with an open angle. Results of an examination of the anterior segment of the left eye were normal with no evidence of lens subluxation or lenticusus. The intraocular pressure was normalized with anti-glaucoma eye drops and the patient underwent cataract surgery 2 days later.

During cataract surgery, it was noted that both the anterior and the posterior capsule were ruptured.
(Fig. 3), with lens debris seen in both the anterior chamber and the vitreous. After most of the lens material was removed, a fundus examination revealed a large nuclear fragment in the posterior pole with no retinal detachment. No intraocular lens was implanted and the wound was closed with no further intervention at this stage. One week later, a standard 3-port vitrectomy was performed to remove the residual lens fragment and lens capsule. No retinal pathology was seen intraoperatively. At 6 months of follow-up, visual acuity had improved to 20/30 with +1.00 + 2.00 × 80 correction. Her intraocular pressure was 12 mm Hg. The left eye remained clear with no evidence of rupture of the lens capsule or cataract.

**DISCUSSION**

Marshall syndrome is a rare autosomal dominant condition with complex skeletal and ocular manifestations. The pathogenesis of Marshall syndrome has been found to be related to a defect in type 2 collagen, which is found in many body structures, including the lens capsule. This defect may make the lens capsule inherently weak, leading to rupture of the lens capsule either spontaneously or with minimal trauma. Prompt recognition of these conditions and appropriate surgical intervention can result in a good outcome.

**REFERENCES**