
Malini Veerappan, MD; Garrick Chak, MD; Christine Shieh, MD; Pratap Challa, MD

ABSTRACT

Introduction: Maroteaux-Lamy syndrome (MLS) is a rare progressive condition characterized by inflammation and scarring of multiple organs. Ocular complications caused by anterior segment abnormalities commonly cause visual impairment in MLS. Angle-closure glaucoma is one such complication, but there are limited data on presentation, workup, and management of this condition.

Case Presentation: This case report describes an atypical presentation of acute angle-closure glaucoma in a patient with MLS despite a prior prophylactic laser peripheral iridotomy—which would typically prevent an acute angle-closure attack—that was patent and intact at the time of angle closure.

Discussion: Because of severe congenital anterior segment crowding, high axial hyperopia, and constant accommodative demand in patients with MLS, we recommend performing two prophylactic laser peripheral iridotomies simultaneously in the same eye instead of one. The mechanism for this indication differs from that in patients at risk of acute angle-closure glaucoma because of lens zonulopathy alone. We hope that this case report may help prevent vision loss and optimize quality of life in patients with MLS who may be wheelchair-bound but are typically high functioning with normal intelligence.

INTRODUCTION

Maroteaux-Lamy syndrome (MLS), known as mucopolysaccharidosis type VI, is a rare autosomal recessive disease caused by a mutation in the ARSB gene. This gene encodes an enzyme called arylsulfatase B, which is involved in the breakdown of glycosaminoglycans (GAGs), specifically the GAGs dermatan sulfate and chondroitin sulfate. Mutations in this gene cause absent or reduced arylsulfatase B activity, which leads to an accumulation of GAGs in cell lysosomes and manifests phenotypically as progressive inflammation and scarring of multiple organ systems.1-4 External features of MLS include macrocephaly, coarse facial features, macroglossia, short stature, and limited joint mobility. Other systemic findings include atlanto-axial instability, meningeal thickening, cervical stenosis, hearing loss, cardiac valve abnormalities, and restrictive/obstructive lung disease, but normal intelligence.2

Ocular complications causing severe vision loss are common in patients with MLS. These complications include corneal clouding (GAG deposition in the cornea), retinopathy (GAG accumulation in the retinal pigment epithelium, causing photoreceptor loss), and ocular hypertension/glaucoma caused by either the open-angle (GAG deposition in the trabecular meshwork) or angle-closure type (narrow anterior chamber with thickened cornea and iris).1 Optic nerve changes, such as optic nerve atrophy, optic nerve swelling, and optic nerve sheath thickening, are also common.5

Typically, a single laser peripheral iridotomy is indicated for the pupillary block mechanism of angle-closure glaucoma (ACG). Currently, performing two laser peripheral iridotomies simultaneously is indicated for patients at risk of acute ACG because of lens subluxation causing pupillary block ACG.6 Although these patients' anatomic risk factors predispose them to a mixed mechanism of ACG, if a patient with mucopolysaccharidosis presents acutely with the pupillary block variety of ACG in the involved eye, we recommend performing two simultaneous laser peripheral iridotomies (LPIs) prophylactically, particularly in patients with MLS, who possess normal intelligence and functional potential.

CASE PRESENTATION

Presenting Concerns

A 37-year-old, wheelchair-bound, 10-diopter (D) hyperopic white woman with MLS documented by arylsulfatase B enzyme assay presented with acute, painful visual decline in her left eye with light perception visual acuity. On presentation, the patient had a history of a patent LPI in each eye and was not receiving any ophthalmic medications. The affected left eye had trace nuclear sclerosis and was notable for acute ACG with an intraocular pressure (IOP) of 60 mmHg.

Records obtained from the patient's local ophthalmologist showed that her IOP in the preceding 8 years ranged from only 10 mmHg to 16 mmHg in both eyes. At 2 months before presentation, best-corrected visual acuity in the affected eye was 20/40 with a manifest refraction of +9.50 + 1.00 × 35. In the other eye, best-corrected visual acuity was 20/25 with a +10.25 lens. The patient reported 6 months of chronically intermittent headaches.

Therapeutic Intervention and Treatment

The patient was treated with maximally tolerated medical therapy using aqueous suppressants as well as oral acetazolamide. Despite the creation of a second LPI in the affected left eye, the patient remained in pupillary block with an IOP of 38 mmHg.

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A surgical peripheral iridectomy was subsequently performed. The IOP decreased to 21 mmHg, and visual acuity improved to counting fingers. The anterior chamber (AC) deepened, but the view to the optic nerve remained hazy because of corneal edema and chronic corneal clouding. Medical therapy was continued, although pilocarpine treatment was held to avoid miotic-induced AC shallowing.

The patient's postoperative regimen included fluorometholone and 5% sodium chloride drops to facilitate corneal clearing. The timeline of events is summarized in Table 1.

Follow-up and Outcomes
Ancillary testing was performed at the postoperative visit 1 week after completion of iridectomy in the left eye (Table 2). Ultrasound biomicroscopy of the left eye revealed a shallow AC and a thickened cornea with central AC depth of only 0.38 mm, measured as the distance from corneal endothelium to the anterior iris border (Figure 1). B-scan ultrasonography demonstrated a thickened, congested sclera (Figure 2) and axial length of less than 20 mm in both eyes (19.53 mm in the right eye and 19.32 mm in the left eye). The retinal-choroidal-scleral thickness was very high,\(^7\) and there was no evidence of vitreous debris, retinal detachment, or mass or tumor.

As of this writing, the patient uses her right eye to see, has stable IOP control, and is being followed closely for signs of angle closure in the right eye.

DISCUSSION
There are limited data on the presentation, workup, cause, and management of ACG in MLS, but the association has been reported in two studies.\(^5,6\) In 1981, two cases of glaucoma associated with MLS were described by Lloyd-Jones and Hitchings.\(^6\) In 1989, Cantor et al\(^7\) described four patients with MLS who had increased IOP and features of glaucoma. Two of these patients had documented ACG that required surgical treatment. One patient had narrow AC depth peripherally but relatively normal depth centrally. This patient's cornea was too opaque to visualize the angle structures. The last patient had angle closure on gonioscopy.\(^7\)

Possible mechanisms for glaucoma vary. Open-angle glaucoma in mucopolysaccharidosis has been attributed to GAG deposition in the trabecular meshwork.\(^8,9\) With ACG, intracellular and extracellular GAG accumulation has been linked to thickening of the cornea and other anterior segment structures.\(^1,10\) Specifically, GAGs are deposited in the intracytoplasmic vacuoles of macrophages in the Bowman layer, corneal stromal keratocytes, ciliary body stroma cells, and intracanalicular connective tissue cells of the trabecular meshwork, as well as extracellularly around the stromal keratocytes.\(^11\) In addition, the constant accommodative demand from high hyperopia shifts the lens-zonule plane anteriorly and increases the risk of ACG. Across different types of glaucoma, optic neuropathy may occur secondary to GAG accumulation in ganglion cells and optic nerve compression caused by thickening of the optic nerve sheath and sclera.\(^1\)

In our patient, after surgical iridectomy to treat the pupillary block component of acute ACG, the plan was to proceed with cataract extraction and lens insertion, primary posterior capsulotomy with anterior vitrectomy, and a glaucoma drainage device to treat the remaining components of ACG. A prophylactic second LPI was also performed in the contralateral eye because of the risk of acute ACG in the setting of shallow AC depth and high hyperopia. Potential challenges to additional surgery involved the patient's medical comorbidities (tracheostomy and multiple cardiac valve abnormalities) as well as surgical positioning of the patient: a posterior cervical fusion had been performed for spinal cord decompression. Furthermore, the patient's extreme axial hyperopia increased her risk of aqueous misdirection or choroidal effusion and necessitated trimming of the posterior plate of the glaucoma drainage device because short axial length increased the risk of optic nerve touch.

### Table 1. Timeline of events

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
<th>Relevant ophthalmic examination data</th>
<th>Intervention</th>
</tr>
</thead>
<tbody>
<tr>
<td>Unknown</td>
<td>Bilateral LPI</td>
<td>Unknown</td>
<td>Uncomplicated postoperative course</td>
</tr>
<tr>
<td>7/2014</td>
<td>Headaches noticed</td>
<td>IOP OS 10 mmHg</td>
<td>None; good IOP control</td>
</tr>
<tr>
<td>1/20/2015</td>
<td>Painful acute vision loss in left eye</td>
<td>IOP OS 16 mmHg</td>
<td>No drops</td>
</tr>
<tr>
<td>1/21/2015</td>
<td>Patient presented with severe headache</td>
<td>IOP OS 60 mmHg, VA OS LP</td>
<td>Laser PI OS; drops; pilocarpine, acetazolamide, brinzolamide-brimonidine, dorzolamide-timolol</td>
</tr>
<tr>
<td>1/22/2015</td>
<td>Persistent headache</td>
<td>IOP OS 38 mmHg, VA OS LP</td>
<td>Surgical PI OS</td>
</tr>
<tr>
<td>1/23/2015</td>
<td>Postoperative day 1 after surgical PI, no headaches</td>
<td>IOP OS 21 mmHg, VA OS CF</td>
<td>Continue therapy with acetazolamide, brinzolamide-brimonidine, and dorzolamide-timolol</td>
</tr>
</tbody>
</table>

* At the time of the event, before intervention.  
* Drops” indicate aqueous suppressant medical therapy.  
* CF = counting fingers; IOP = intraocular pressure; LP = light perception; LPI = laser peripheral iridectomy; NA = not available; OS = left eye; VA = visual acuity.

### Table 2. Patient parameters at 1-week postoperative visit

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Measurement</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anterior chamber depth, mm</td>
<td>OD: 1.19, OS: 0.38</td>
</tr>
<tr>
<td>Axial length, mm</td>
<td>OD: 19.53, OS: 19.32</td>
</tr>
<tr>
<td>Average keratometry, diopeters*</td>
<td>OU: 43.50</td>
</tr>
<tr>
<td>Central corneal thickness, µm²</td>
<td>OU: 565; OS: undetectable because of corneal edema</td>
</tr>
<tr>
<td>Retinal-choroidal-scleral thickness, mm²</td>
<td>OS: 2.3</td>
</tr>
</tbody>
</table>

* Average keratometry measurement obtained using a manual keratometer.  
* Central corneal thickness from a handheld pachymeter (Pachmate DGH 55, DGH Technology Inc, Exton, PA).  
* Normal retinal-choroidal-scleral thickness is 1.3 mm.  
OD = right eye; OS = left eye; OU = both eyes.

The patient presented with symptoms of acute angle-closure glaucoma. The clinical presentation was atypical for Maroteaux-Lamy syndrome (MLS), and the patient was predisposed to extreme pupillary block due to congenital anterior segment crowding.

CONCLUSION

This case highlights the importance of considering prophylactic laser peripheral iridotomy (LPI) in patients at risk for angle closure, particularly those with Maroteaux-Lamy syndrome. The use of LPI can help prevent future episodes of acute angle-closure glaucoma.

Disclosure Statement

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References