

Scheie Syndrome and Narrow Angles

BY JENNIFER S. WEIZER, MD

CASE PRESENTATION

A 37-year-old white male was referred by a community glaucoma specialist to the Kellogg Eye Center at the University of Michigan in Ann Arbor for a glaucoma evaluation. He had a history of Scheie syndrome, hyperopia, narrow angles, and elevated IOP. His maximum untreated IOP had been 28 mm Hg in both eyes 2 years earlier, at which point he began treatment with timolol 0.5% eye drops q.i.d. in both eyes. He had no ocular history of laser procedures or surgeries and had not used any other eye drops.

At presentation, the patient complained of gradually worsening vision in both eyes over the past year. Besides Scheie syndrome, his medical history was significant for asthma, sleep apnea, and allergic rhinitis. His systemic medications included an albuterol inhaler and mometasone furoate nasal spray, which he used every other day. He had no family history of glaucoma, although his sister also had Scheie syndrome.

His visual acuity was 20/50 OU with a manifest refraction of +5.00 +0.75 X 81 OD and +5.00 +1.25 X 40 OS. An external examination revealed coarse facial features and a prominent brow. Goldmann applanation tonometry

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measured 21 mm Hg OD and 16 mm Hg OS. Central corneal thickness was 657 μ m OD and 638 μ m OS. A slit-lamp examination revealed diffuse stromal haze in the corneas, peripherally shallow anterior chambers, and clear lenses in both eyes (Figure 1). Gonioscopy yielded a difficult view due to the corneal haze, but no angle structures were visible in either eye. Despite the hazy view, an undilated fundus examination showed healthy optic nerves with cup-to-disc ratios of 0.2 OU, and the retinas appeared normal in both eyes. Automated perimetry with the Humphrey Field Analyzer (Carl Zeiss Meditec, Inc., Dublin, CA) showed nonspecific diffuse depression (Figure 2).

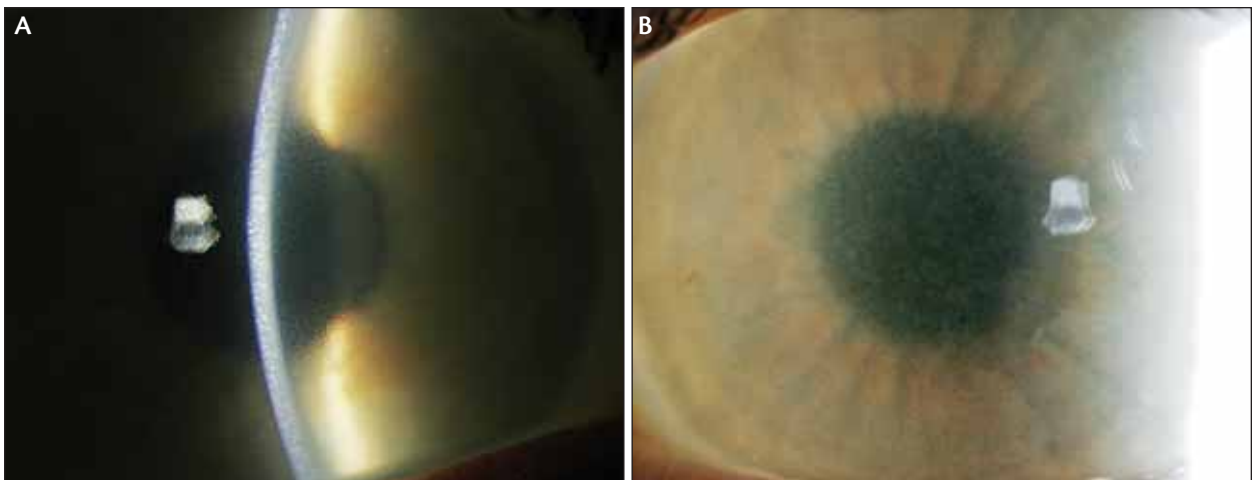


Figure 1. Slit-lamp views of the anterior segment show diffuse corneal stromal haze, a shallow anterior chamber, and a clear lens in the right (A) and left eyes (B).

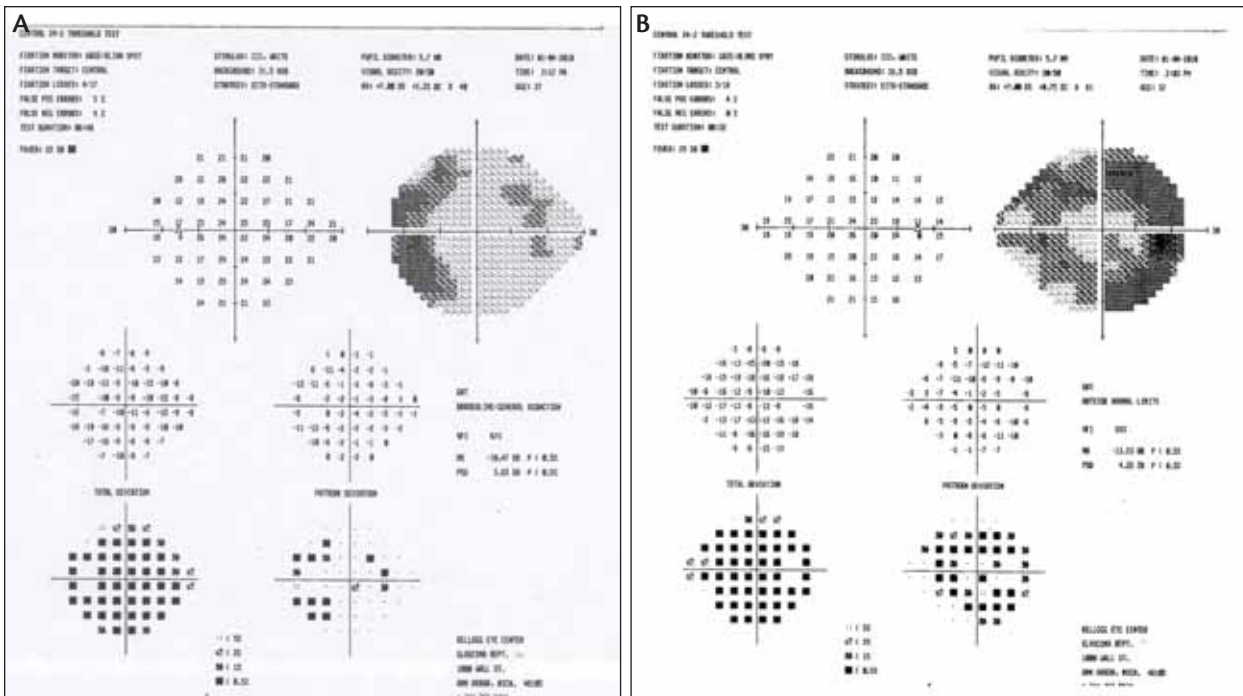


Figure 2. Humphrey Field Analyzer visual field testing shows relatively nonspecific diffuse depression in the left (A) and right eyes (B).

HOW WOULD YOU PROCEED?

- Can the patient's angles be assessed by any other method?
- How would you expect this patient's angles to respond to peripheral laser iridotomy?
- With thick corneas, presumably due to stromal deposits of glycosaminoglycans, is his IOP overestimated by Goldmann applanation tonometry?

SURGICAL COURSE

Because the patient's angles were difficult to assess with gonioscopy, I obtained anterior segment optical coherence tomography (AS-OCT) imaging (Visante OCT; Carl Zeiss Meditec, Inc.), which showed appositional narrowing for 360° in both eyes (Figure 3). I then recommended a YAG laser peripheral iridotomy in both eyes, which was per-

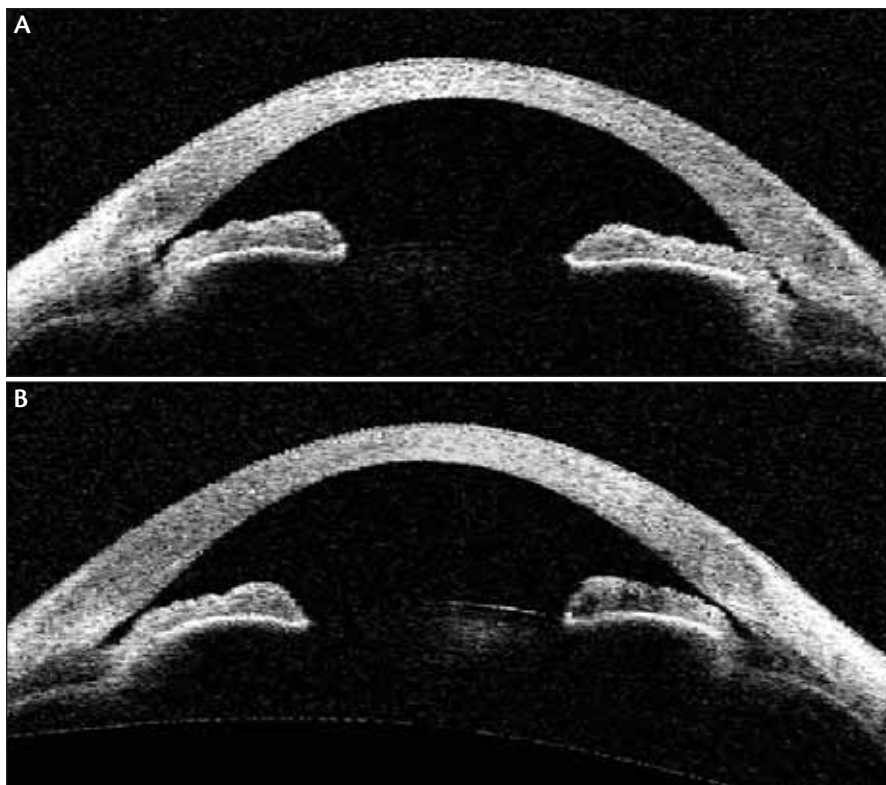


Figure 3. AS-OCT of the patient's right (A) and left (B) eyes shows appositional narrowing of the angle, which was present for 360°.

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formed without complication despite the cloudy corneas. Postoperatively, the patient administered prednisolone acetate 1% eye drops q.i.d. for 5 days in each eye. His timolol was continued.

OUTCOME

Repeat AS-OCT showed no significant change in angle anatomy in either eye. Likewise, the patient's IOP remained stable after the laser treatment. At this point, there was no significant elevation in IOP in either eye with pupillary dilation. Primarily because of his healthy optic nerves but also because his visual fields were not definitely glaucomatous, I instructed the patient to continue using timolol and continued to observe him. He was referred for a corneal evaluation as well, but given his relatively good visual acuity, corneal transplantation was deferred.

DISCUSSION

Scheie syndrome is one of a rare group of disorders known as *mucopolysaccharidosis* (MPS) in which abnormal deposits of glycosaminoglycans (GAGs) are deposited throughout the body, including in the tissues of the eye.¹ The incidence of all types of MPS is approximately one in 20,000 live births.² The genetic defects in the various types of MPS produce specific abnormal lysosomal enzymes, which are unable to properly metabolize GAGs. In Scheie syndrome, the abnormal enzyme is alpha-L-iduronidase, and the inheritance of this disease is autosomal recessive. Patients with Scheie syndrome typically suffer from skeletal, cardiac, and respiratory problems, although their phenotypic manifestations tend to be less severe than in the other subtypes of MPS.^{1,2}

Typical ocular findings in MPS include corneal opacification, ocular hypertension, glaucoma, retinopathy, and abnormalities of the optic nerve.¹ Strabismus is not uncommon.³ Corneal clouding occurs because of an accumulation of GAGs in the corneal stroma. Ocular hypertension and glaucoma are thought to develop via three mechanisms. The first is crowding of the anterior chamber angle caused by the thickening of ocular structures due to deposits of GAGs, and angle-closure

glaucoma by this mechanism has been described.⁴ The second mechanism is decreased aqueous outflow due to an accumulation of GAGs in the trabecular meshwork. Finally, goniodysgenesis has been noted in patients with MPS as well.⁵ Retinopathy in MPS is caused by GAGs deposited in the retinal pigment epithelium and in the photoreceptor layer. Optic disc swelling and atrophy can occur due to glaucoma or because of the compression of axons by dura and sclera thickened by GAG accumulations.¹

This patient's hyperopia, which is related to the narrowing of his anterior chamber angles, is also likely to be at least partly associated with Scheie syndrome.¹ His history of elevated IOP is probably affected by his thick corneas, which contain deposits of GAGs. It is not clear how GAGs in the corneal stroma affect corneal hysteresis, but it seems likely that his IOP is overestimated because of his increased corneal thickness. Studies have shown that corneal opacification in MPS is positively correlated with IOP and central corneal thickness.^{1,3} In MPS patients, however, having higher IOP even with thicker, more opaque corneas was associated with more advanced glaucomatous optic neuropathy.³

The utility of laser peripheral iridotomy in eyes with narrow angles associated with MPS has not been well studied. In this case, the angle configuration did not seem to widen after laser treatment, but it is possible that a patent peripheral iridotomy might help to reduce his risk of developing angle-closure glaucoma, whether acute or chronic. Because of the rarity of MPS, studying its association with glaucoma in a large case series in order to make recommendations about treatment is a difficult task. Observing these patients over time is the best opportunity for learning about the ocular complications from which they can suffer. □

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