

Glaucoma Associated With Aniridia

An overview of treatment options.

BY SARWAT SALIM, MD

Aniridia, a rare panocular developmental disorder, is characterized by either partial or complete hypoplasia of the iris (Figure 1). Other ocular structures (including the cornea, anterior chamber angle, crystalline lens [Figure 2], retina, macula, and optic nerve) may also be affected, either at birth or later in life. Some forms of aniridia may be accompanied by systemic abnormalities. No gender or racial predilection has been observed for aniridia. The majority of cases are inherited through an autosomal dominant mode with high penetrance and variable phenotypes.¹ Approximately one-third of cases are sporadic. Deletion of the *PAX6* gene, located on chromosome 11p13, and its proximity to the Wilms tumor predisposition gene increase the risk of Wilms tumor in newborns with sporadic aniridia.

With a reported incidence of 6% to 75%,² glaucoma remains one of the most challenging features of aniridia. The higher percentage may represent an overestimation from academic glaucoma centers; a more accurate estimate may be around 50%.² Congenital glaucoma associated with buphthalmos is rare in aniridia. Rather, eyes with aniridia usually develop glaucoma in late childhood or young adulthood, as progressive anatomical changes occur in the drainage angle.³

THE ANGLE'S ANATOMY

Several mechanisms have been described for elevated IOP and secondary glaucoma, and they emphasize the role of routine gonioscopy in monitoring aniridic eyes. In some cases, the anterior chamber angle may be maldeveloped whether or not Schlemm canal is absent.^{2,4} More commonly, the angle appears to be open in infancy, with minimal to no coverage of the filtering portion of the trabecular meshwork (TM) by the iris tissue and full visualization of the scleral spur in most quadrants. Over time, changes in the angle's anatomy occur, as the rudimentary iris stump rotates and covers the TM. In addition to the optic nerve's



Figure 1. An eye with aniridia and a clear lens.



Figure 2. An eye with aniridia and a cataract.

appearance, the extent of the iris tissue's coverage of the TM helps physicians to differentiate the various stages of glaucoma and guides therapeutic intervention.³

In mild or early glaucoma, less than half of the angle may be circumferentially involved, with multiple thin extensions of the iris stroma or processes across the scleral spur onto

the TM. These processes are lightly pigmented and may be accompanied by fine blood vessels. Contraction of the tissue strands between the peripheral iris and the angle wall may be more evident in the superior angle. In moderate to severe glaucoma, most of the TM is covered by the iris tissue, which takes a parallel orientation to the angle wall, leaving the iris pigment epithelium draped across the ciliary processes. Secondary angle closure may also occur from ectopia lentis, an intumescent lens, the use of miotic agents, or surgery for cataracts or glaucoma.²

MEDICAL THERAPY AND LASER TREATMENT

The initial treatment of glaucoma associated with aniridia is usually medical therapy, specifically with aqueous suppressants. Some eyes may respond to miotic agents, which increase aqueous outflow through unobstructed TM by contracting the ciliary muscle.^{2,3} Argon laser trabeculoplasty is ineffective when performed in eyes with open angles.² Wiggins and Tomey also found that argon laser energy directly applied to the ciliary processes achieved no significant reduction in IOP.⁴ A majority of patients with aniridia eventually require surgical intervention for uncontrolled glaucoma.

SURGICAL INTERVENTION

Goniotomy

Goniotomy appears to be of limited value in advanced cases, although its role in preventing or delaying aniridic glaucoma has been described.³⁻⁵ Chen and Walton used a modified goniosurgical technique covering approximately 200° of the angle to prevent the development of glaucoma and reported a success rate of 89% (IOP < 22 mm Hg without medication) in 55 eyes of 33 patients.⁶ In the remaining 11% of patients who subsequently developed glaucoma, IOP was controlled with medical therapy. The mean age at the time of the initial surgery was 36.6 months (range, 10-113 months) with an average follow-up of about 9.5 years.

Surgery was performed on eyes in which the posterior TM was covered for more than one-half of its circumference by extensions of tissue from the peripheral iris and on eyes that demonstrated progressive coverage of the TM by iris tissue. The modified technique involved detaching the abnormal tissue between the iris and the TM with a goniotomy knife without placing an incision through the TM. No surgical complications were reported.

Trabeculotomy

The results of trabeculotomy in eyes with glaucoma associated with aniridia are conflicting. Adachi et al found a higher surgical success with trabeculotomy than with other glaucoma surgeries.⁷ They reported a qualified surgical suc-

cess of 83% in 10 out of 12 eyes over a mean follow-up period of 9.5 years after one or two trabeculotomy procedures. Of note, eyes in this series were diagnosed with glaucoma in the first year of life and had open angles on gonioscopy at the time of surgery. In contrast, Wiggins and Tomey reported trabeculotomy failures in their series.⁴

Trabeculectomy

Trabeculectomy may be performed on eyes refractory to medical treatment. Several studies have found limited success with this approach, however, and potentially higher rates of shallow or flat anterior chambers after surgery, consequently requiring meticulous postoperative observation. Grant and Walton reported failures in all eyes that underwent trabeculectomy.³ Wiggins and Tomey reported a surgical success rate of only 7% after trabeculectomy and a need for additional surgery in mainly young or aphakic patients.⁴

Other investigators have found trabeculectomy to be effective. Nelson et al reported successful outcomes after trabeculectomy in 11 of 14 eyes 1 year postoperatively.² Okada et al found trabeculectomy to be effective in 10 eyes of aniridic patients under the age of 40 years.⁸ The investigators reported that the IOP in these 10 eyes was stable from the time of surgery until it exceeded 20 mm Hg with or without glaucoma medications (approximately 14.6 months).

Tube Shunt Surgery

Glaucoma drainage devices have been shown to be effective for aniridic glaucoma.^{9,10} Arroyave et al reported a success rate of 88% 1 year postoperatively in eight aniridic eyes that received glaucoma drainage devices (mostly Baerveldt glaucoma implants [Abbott Medical Optics Inc.]).¹¹ The investigators defined surgical success as an IOP of 21 mm Hg or lower with or without medication, without additional surgery, and without visually devastating complications.

Wiggins and Tomey reported a surgical success rate of 83% (IOP ≤ 21 mm Hg with no visually significant complications) in five of six eyes that received a Molteno Implant (Molteno Ophthalmic Limited).⁴ All eyes in this study had undergone multiple ocular surgeries prior to the placement of the implant. In terms of complications, only one case of tube migration was reported and required additional surgery.

Cyclodestruction

Cyclodestructive procedures may be performed in cases refractory to medical and surgical therapy. Often, multiple treatments are required. These procedures generally are not recommended as a primary intervention due to higher rates of associated complications, including cataract formation or phthisis bulbi.

Wiggins and Tomey reported a success rate of only 25% after cyclocryotherapy in aniridic eyes.⁴ Wallace et al reported adequate IOP control in six of nine aniridic eyes after cyclocryotherapy, but 50% of the treated eyes experienced a reduction in visual acuity.¹² Wagle et al reported higher rates of complications in aniridic eyes after cyclocryotherapy when compared with other types of refractory pediatric glaucomas.¹³ In contrast, Kirwan et al found the cyclophotocoagulation diode laser to be effective and safe in children with refractory pediatric glaucoma, including five eyes with aniridia.¹⁴

CONCLUSION

Glaucoma is a common complication of aniridia and is difficult to control both medically and surgically. Routine gonioscopy is critical to guide appropriate prophylactic or therapeutic intervention. Goniosurgery, either trabeculotomy or goniotomy, may benefit young aniridic patients with glaucoma who exhibit little to no obstruction of the TM by the iris tissue.¹⁵ In general, glaucoma drainage devices may be more successful than goniosurgery or glaucoma filtration surgery in aniridic glaucoma. Cyclodestructive procedures are usually reserved for refractory cases, given the intervention's higher rates of complications. The best approach may be to prevent or delay aniridic glaucoma by prophylactic modified goniotomy in eyes that demonstrate progressive anatomical changes in the drainage angle. ■

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