

Diagnosis and Treatment of Primary Congenital Glaucoma

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Although glaucoma in adults is a common finding, primary congenital glaucoma is not an everyday diagnosis. Diagnosis at birth occurs in only 25 percent of infants with the disease; disease onset occurs in the first year of life in more than 80 percent of all childhood glaucoma cases. If glaucoma presents after 3 or 4 years of age, it is called primary juvenile open-angle glaucoma.

In the United States, the incidence of primary congenital glaucoma is roughly one in 10,000 live births. Worldwide, the incidence ranges from a low of 1:22,000 in Northern Ireland to a high of 1:2,500 in Saudi Arabia and 1:1,250 among Gypsies in Romania. Primary congenital glaucoma is bilateral in about two-thirds of cases. There are differences in sex predilection: in North America and Europe it is more common in boys, whereas in Japan it is more common in girls.

Pathogenesis

The pathogenesis of primary congenital glaucoma is unknown and is currently described as a developmental defect. Historically, Barkan thought a thin, imperforate membrane covered the anterior chamber angle structures and blocked aqueous humor outflow. However, the hypothetical “Barkan’s membrane” has not been identified conclusively on clinical or histologic exam. The area of obstruction is now thought to be the trabecular meshwork, but the exact mechanism remains incompletely characterized.

Angle Comparisons

How the angle in healthy infants differs from the angle in healthy adults:

- The trabecular meshwork is more lightly pigmented.
- Schwalbe’s line is often less distinct.
- The uveal meshwork is translucent so that the junction between the scleral spur and the ciliary body band often cannot be clearly seen.

How the angle in infants with primary congenital glaucoma differs from the angle in healthy infants:

- The iris inserts anteriorly, often on or anterior to the scleral spur, compared with the way it inserts in the healthy infant angle.
- The stroma of the peripheral iris is hypoplastic and nonpigmented and has a scalloped appearance.

Presentation

Primary congenital glaucoma is characterized by the clinical triad of epiphora, blepharospasm and photophobia, but these symptoms are often missed. For example, epiphora may be mistaken for congenital nasolacrimal duct obstruction.

The patient usually presents because of corneal clouding and ocular enlargement. Corneal edema is due to elevated IOP and may be either subtle and gradual or sudden in onset. Corneal edema is often accompanied by curvilinear breaks in Descemet’s membrane (Haab striae), which remain for the rest of the patient’s life.

Ocular enlargement, a feature of all forms of childhood glaucoma, occurs because the immature and growing collagen of the cornea and sclera in the young eye still responds to increased IOP by stretching. All parts of the globe may stretch in response to the

elevated IOP until 3 or 4 years of age, and glaucoma-related axial myopia may be seen until the early teenage years. In healthy children, the normal horizontal corneal diameter is 9.5 to 10.5 mm for newborns and 10.0 to 11.5 mm by 1 year of age. A diameter that is more than 1.0 mm above the normal range is of concern. Glaucoma should be suspected in any child with a corneal diameter greater than 13.0 mm. The differential diagnosis is listed on the next page.

Examination

It is important to do a complete ophthalmic exam in a child suspected of glaucoma. This exam must include IOP measurement, gonioscopy, inspection of the optic nerve and refraction. Check for the child’s ability to fix and follow and for the presence of nystagmus. Examination of the cornea is crucial, with respect to the size and

Differential Diagnosis

CORNEAL EDEMA OR CLOUDING

Congenital hereditary endothelial dystrophy
Mucopolysaccharidoses I, IS, II and III
Cystinosis
Sclerocornea
Rubella keratitis
Obstetric birth trauma
Chemical injury
Idiopathic (diagnosis of exclusion)

EPIPHORA AND/OR RED EYE

Nasolacrimal duct obstruction
Viral conjunctivitis, bacterial conjunctivitis (including *Chlamydia*)
Corneal epithelial defect, abrasion

PHOTOPHOBIA

Iritis
Trauma (especially with hyphema)

CORNEAL ENLARGEMENT

Axial myopia
Megalocornea (X-linked or sporadic)

OPTIC NERVE CUPPING

(real or apparent)

Physiologic optic nerve cupping
Optic nerve coloboma
Optic atrophy
Optic nerve hypoplasia
Optic nerve malformation

clarity of the cornea and the presence of Haab striae. The anterior chamber angle of a healthy infant differs from that of a healthy adult and from that of a child with glaucoma, as described in “Angle Comparisons” (previous page). The optic nerve exam usually reveals a cup-to-disc ratio greater than 0.3 in infants with glaucoma. This ratio is usually less than 0.3 in healthy newborns. Asymmetry in the cup-to-disc ratios is also suggestive of glaucoma. Although cupping may occur rapidly in infants, it is reversible with surgical management and IOP control. This reversibility is most common in early stages of glaucoma in children, especially in the first year of life when the connective tissue of the optic nerve is relatively elastic.^{1,2}

Although measuring IOP can be challenging, this is a necessary part of the exam and can be done in the

office with a conscious, swaddled infant. Bottle-feeding may help to calm the infant. A Tono-Pen or a Perkins or Goldmann applanation tonometer may be used. In infants with healthy eyes, the IOP is in the range of 10 to 12 mmHg, and it reaches approximately 14 mmHg by age 7 or 8 years. An IOP measurement of greater than 20 mmHg in a calm resting infant raises the suspicion of glaucoma when other signs and symptoms suggest the disease. Likewise, asymmetry of more than 5 mmHg is of concern. IOP measurements taken while a child is crying are unreliable, since the Valsalva maneuver and eyelid squeezing can cause an IOP of 30 to 40 mmHg even in healthy eyes.

If it is not possible to get an IOP measurement in the office, an exam under anesthesia that includes optic nerve examination should be performed. General anesthetics can lower the IOP. However, in primary congenital glaucoma, measurements made soon after induction of general anesthesia are still usually greater than 20 mmHg, though pressure typically drops after this early stage.

For the sake of comparison, the reported IOP in a healthy infant under halothane anesthesia is 9 to 10 mmHg. It appears that conscious sedation with chloral hydrate has the least effect on IOP measurement, with no statistical significance noted in studies.³ A study using chloral hydrate found IOP in healthy children to range from 11 to 17 mmHg. Some agents are known to increase IOP, including ketamine (minimal to mild) and succinylcholine (marked).

Treatment

Medical therapy is generally used as a temporizing measure to help control IOP and clear the cornea prior to surgery. However, it may become more integral to the treatment strategy in cases where surgery is not possible due to medical reasons or when surgery is inadequate. Medical treatment can be challenging since most of the relevant medications are not approved for use in children.

Surgery. The main treatment options are surgical and include goniotomy and trabeculotomy. Surgical planning begins with a comprehensive examination under anesthesia. As long as the cornea is clear, either procedure may be performed.

In goniotomy, an incision is made across the trabecular meshwork under direct gonioscopic visualization. In trabeculotomy, the trabecular meshwork is incised by way of an external approach through Schlemm’s canal. If the cornea is hazy or opaque, trabeculotomy is the better choice.

These procedures have high success rates, up to 90 percent or greater in some series. Most infants with primary congenital glaucoma who present between 3 months and 1 year of age will have their IOP controlled by one or two angle surgeries. If IOP control is not achieved with the first surgery, at least one other angle surgery is attempted before trying another strategy. When angle surgery fails and medical therapy is inadequate, other options may be considered, such as tube implant procedures, trabeculectomy and cryoablation. Successful combined trabeculotomy-trabeculectomy has been reported.⁴ Shunt procedures were found to be more successful than trabeculectomy in patients under 2 years of age.⁴

Conclusion

The main goal in managing primary congenital glaucoma is early diagnosis and therefore early surgical treatment to normalize IOP, in order to allow possible reversal of cupping and thus to minimize the impact on vision.

1 Meirelles, S. H. *J Glaucoma* 2008;17(6):470–473.

2 Biglan, A. W. *J AAPOS* 2006;10(1):7–21.

3 Jaafar, M. S. and G. A. Kazi. *J Pediatr Ophthalmol Strabismus* 1993;30(6):372–376.

4 Ho, C. L. and D. S. Walton. *Curr Opin Ophthalmol* 2004;15:460–464.

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