Childhood Glaucoma
Associated With Developmental Ocular and Systemic Disorders

Preventing blindness requires recognizing the abnormalities associated with this group of diseases.

BY DAVID S. WALTON, MD

Childhood glaucoma is an important and demanding cause of preventable childhood blindness. Some pediatricians and other childcare workers’ unfamiliarity with the diagnostic signs of glaucoma in children can delay its recognition. In addition, parents often do not recognize the significance of the early abnormalities secondary to increased IOP such as ocular enlargement and photophobia. Childhood glaucoma also challenges eye care specialists. Its rarity means that the necessary examination techniques are practiced infrequently. Moreover, they require special instrumentation, are time consuming, and are not easily handed off to office assistants.

Upon diagnosis, the type and severity of the eye disease must be determined, and the potential for associated pediatric systemic conditions must be actively considered. Medical therapy is often less successful than in adults, and the execution of appropriate glaucoma surgical procedures in infants and young children may initially be uncomfortable even for experienced eye surgeons. Parents are typically surprised by the diagnosis of childhood glaucoma and may become overwhelmed by the logistical challenge of care as well as by their feelings of responsibility for both the condition and its delayed recognition. That said, parents are amazingly resilient, and their subsequent positive efforts to support physicians are both humbling and essential to the successful treatment of these glaucomas.

To achieve the best possible outcomes, the ophthalmologist must become familiar with the many potential types of pediatric glaucoma (see The Childhood Glaucomas) and their medical or surgical treatments. In addition, he or she must have instrumentation available for tonometry, inspection of the anterior segment, and gonioscopy.

DIAGNOSIS

The most common primary childhood glaucoma is primary congenital glaucoma (PCG). This hereditary glaucoma can be caused by 147 mutations to one gene, CYP1B1. This gene has been mapped to chromosome 2p22.2 and may be responsible for approximately one-third of the cases of PCG. The incidence of this disease ranges from 1:2,500 to 1:60,000 and is highly variable in different geographic and ethnic populations.

PCG may present at birth associated with prompt recognition of corneal anomalies and opacities secondary to increased IOP. More frequently, however, PCG is detected during the first year of life when a primary care physician or parent notices a child’s symptomatic photophobia and corneal enlargement and opacification (Figure). When the disease is less severe, however, these anterior segment abnormalities may be less obvious, leading to the late recognition of PCG. In these cases, PCG is diagnosed by the presence of decreased visual acuity, corneal enlargement, and, rarely, routine tonometry.
### THE CHILDHOOD GLAUCOMAS

#### I. Primary Developmental Glaucomas

A. Primary congenital glaucoma
   1. Newborn
   2. Infantile
   3. Late recognized

B. Juvenile open-angle glaucoma

C. Primary glaucomas associated with systemic diseases
   1. Sturge-Weber syndrome
   2. Neurofibromatosis (NF-1)
   3. Stickler syndrome
   4. Oculocerebrorenal syndrome (Lowe)
   5. Axenfeld-Rieger syndrome
   6. SHORT syndrome
   7. Hepatocerebrorenal syndrome (Zellweger)
   8. Marfan syndrome
   9. Rubinstein-Taybi syndrome
   10. Infantile glaucoma with retardation and paralysis
   11. Oculodentodigital dysplasia
   12. Glaucoma with microcornea and absent sinuses
   13. Mucopolysaccharidosis
   14. Trisomy 13
   15. Caudal regression syndrome
   16. Trisomy 21 (Down syndrome)
   17. Cutis marmorata telangiectatica congenita
   18. Warburg syndrome
   19. Kniest syndrome (skeletal dysplasia)
   20. Michel's syndrome
   21. Nonprogressive hemiatrophy
   22. PHACES syndrome
   23. Soto syndrome
   24. Linear scleroderma
   25. GAPO syndrome
   26. Roberts pseudothalidomide syndrome
   27. Wolf-Hirschhorn (4p-) syndrome
   28. Robinow syndrome
   29. Nail-patella syndrome
   30. Proteus syndrome
   31. Fetal hydantoin syndrome
   32. Cranio-cerebello-cardiac (3C) syndrome
   33. Brachmann-deLange syndrome
   34. Rothmund-Thomson syndrome
   35. 9p deletion syndrome
   36. Phakomatosis pigmentovascularis
   37. Jacobsen syndrome

D. Primary glaucomas associated with ocular anomalies
   1. Aniridia

#### a. Congenital aniridic glaucoma
   b. Acquired aniridic glaucoma

#### II. Secondary (Acquired) Glaucomas

A. Traumatic glaucoma
   1. Acute glaucoma
      a. Angle concussion
      b. Hyphema
      c. Ghost cell glaucoma
   2. Glaucoma related to angle recession
   3. Arteriovenous fistula

B. Glaucoma with intraocular neoplasms
   1. Retinoblastoma
   2. Juvenile xanthogranuloma
   3. Leukemia
   4. Melanoma of ciliary body
   5. Melanocytoma
   6. Iris rhabdomyosarcoma
   7. Aggressive iris nevus
   8. Medulloepithelioma
   9. Mucogenic glaucoma with iris stromal cyst

C. Glaucoma related to chronic uveitis
   1. Open-angle glaucoma
      2. Angle-blockage mechanisms
         a. Synechial angle closure
         b. Iris bombé with pupillary block
         c. Trabecular endothelialization
   2. Uveitic angle-closure glaucoma

D. Lens-related glaucoma
   1. Subluxation-dislocation with pupillary block
      a. Marfan syndrome
      b. Homocystinuria
      c. Weill-Marchesani syndrome
   2. Axial subluxation high myopia syndrome
   3. Ectopia lentis et pupillae
During the ocular examination, gonioscopy is most helpful to distinguish PCG from other types of pediatric glaucoma. Typically, the filtration angle anomaly is confined to the relative presence of the trabecular meshwork, scleral spur, and ciliary body regions.

**TREATMENT**

PCG patients often require glaucoma surgery. Goniosurgery is the first line, unless an advanced angle anomaly is present, as may be seen with the newborn expression of this condition.5,7 The success of goniosurgery for PCG relates to the severity of the filtration angle defect.8 When patients present at birth with cloudy corneas, elevated IOP, and iris anomalies, the disease may be complicated by filtration angle hypoplasia. Gonioscopy may be difficult in these children, and the surgeon must consider the placement of a glaucoma drainage device instead of initial goniosurgery.5 Patients with PCG recognized after 1 month of age with corneal signs secondary to the elevated IOP can be expected to do well with goniosurgery, even when the diagnosis is made later in childhood. When goniosurgery fails after 6 months of age, trabeculotomy or glaucoma drainage implants must be used.9

**CONCLUSION**

Pediatric glaucoma is unusual but an important cause of childhood blindness. Signs of the disease early in life and during eye examinations throughout childhood offer the opportunity for glaucoma’s recognition and prompt treatment. Both the medical and the surgical treatment of childhood glaucoma have advanced in the past 25 years related to the increased interest and improved training of pediatric and glaucoma specialists. Once a hopeless diagnosis, parents now bring their children for glaucoma care with an expectation of successful treatment.

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