Evaluating and Managing Optic Disc Cupping in Children

How to tell when a child has glaucoma.

BY ALLEN BECK, MD

A 10-year-old boy is referred to you after a routine examination by another eye care provider revealed large cup-to-disc ratios bilaterally. His BCVA is 20/20 OU with a low myopic correction. The IOP measures 18 mm Hg OD and 19 mm Hg OS. Gonioscopy demonstrates an open angle to the ciliary body band with minimal pigmentation. Figure 1 shows the appearance of the optic nerves. What is your next step in the workup of this patient?

Answering that question requires a knowledge of the wide physiological variation in the optic disc's appearance. It is also important to understand the ocular and systemic conditions commonly associated with childhood glaucoma as well as congenital optic disc anomalies that may mimic glaucoma. This article provides tips to assist you in the diagnosis of pediatric glaucoma.

BACKGROUND

The Optic Nerve

A wide variation in the appearance of the optic nerve head is one of the challenges in diagnosing glaucoma in patients of any age. In the Baltimore Eye Survey, the optic disc area ranged from 1.15 to 4.94 mm^2 in white adult patients and from 0.90 to 6.28 mm^2 in black adult patients. The estimated average vertical cup-to-disc ratios were 0.4 to 0.5 in whites and 0.5 to 0.7 in blacks.[1] The Ocular Hypertension Treatment Study, however, found a larger horizontal or vertical cup-to-disc ratio to be a risk factor for the development of glaucoma.[2] The question is, how do you determine what is glaucoma and what is not?

Small optic discs generally have small-to-absent optic cups. Large optic discs generally have large optic cups, but they may have a greater number of nerve fibers than small discs.[3] Asymmetry of the cup-to-disc ratio greater than 0.2 or notching or focal or diffuse thinning of the
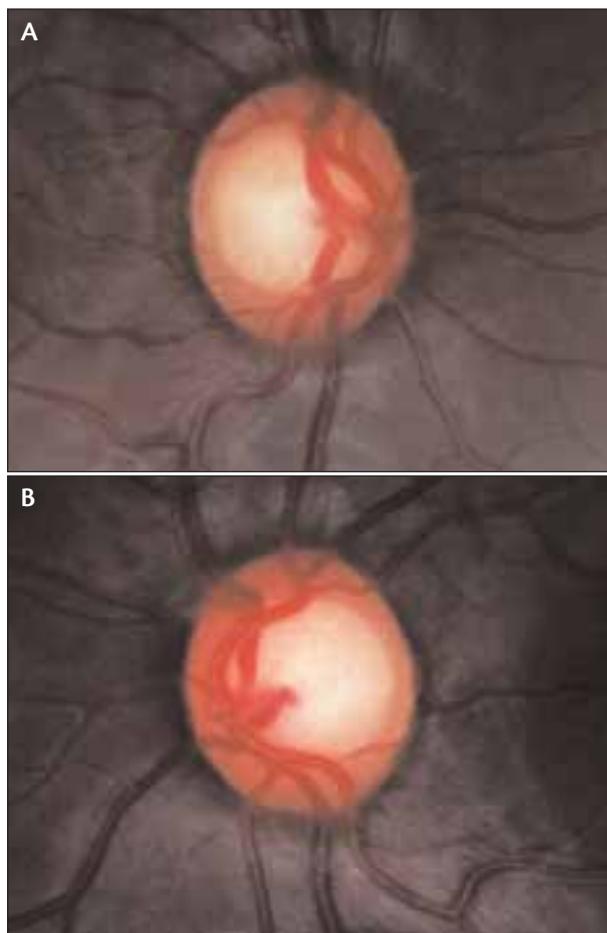

Figure 1. The patient's right (A) and left (B) optic nerves.

rim of the optic nerve is characteristic of glaucomatous changes. The ISNT rule (inferior rim thickness ≥ superior rim ≥ nasal rim ≥ temporal rim) is a useful strategy in detecting glaucomatous optic nerves.[4]

Imaging technology using optical coherence tomography or confocal scanning laser ophthalmoscopy is another way to assess a suspicious optic nerve. These modalities can provide useful information if a child is able to sit still for such testing. Studies of the thickness of the nerve fiber layer in children have confirmed the ISNT rule, but there is considerable variation in normal thickness.[5] Refractive error positively correlates to the thickness of the nerve fiber layer, with thicker rims in hyperopic eyes and thinner rims in myopic eyes.[6] The nerve fiber layer's thickness and macular volume differ significantly between normal children and those with glaucoma.[7] The absence of a normative database for patients under the age of 18 years, however, may make it difficult to differentiate normal from abnormal optic nerves. Nevertheless, even in the absence of a normative database, the user can evaluate asymmetry between eyes of the retinal nerve fiber layer with optical coherence tomography, and confocal scanning allows the measurement of disc area that can confirm a larger-sized disc that may mimic glaucoma. Further studies with optic nerve analyzers of normal children are underway, and this research will likely improve clinicians' ability to distinguish normal from abnormal optic nerves.

A variety of congenital optic disc abnormalities may mimic the appearance of glaucoma, including tilted optic discs, an optic nerve pit, and optic nerve hypoplasia. The absence of other findings to support a diagnosis of glaucoma—and the static nature of congenital optic nerve anomalies—will usually allow differentiation from a glaucomatous optic nerve. Photographic documentation of congenital optic nerve disorders during childhood can also help prevent misdiagnosis when patients become adults.

Other Signs

Fortunately, there are other clues to the diagnosis of glaucoma in children besides the appearance of the optic disc. A variety of conditions are associated with the development of glaucoma, including aphakia/pseudophakia, aniridia, Axenfeld-Rieger syndrome, Sturge-Weber syndrome, uveitis, and trauma. With the exception of trauma (for which the only finding may be angle recession on gonioscopy), the aforementioned conditions have characteristic ophthalmic findings. Primary congenital glaucoma, with its characteristic corneal enlargement, may occasionally go undiagnosed in infants due to later onset or the absence of corneal edema. Juvenile open-angle glaucoma typically has markedly elevated IOP combined with classic glaucomatous nerve and visual field findings, and a family history may also be helpful due to autosomal dominant inheritance.

Increased IOP is generally present with all forms of childhood glaucoma, and the elevation is frequently marked. Normal-tension glaucoma is a condition of older adult patients; a glaucomatous process in childhood without an associated rise in IOP is extremely rare and should be a diagnosis of exclusion.[8] Overestimating the IOP due to movement, lid squeezing, or breath-holding is a significant issue in children. The problem can be limited by engaging children in conversation and using a hand-held tonometer, which they may find less threatening than the typical slit-lamp approach.

Studies of central corneal thickness (CCT) in children have demonstrated some normal variation in the cornea during growth and development. One important point is that aphakic and pseudophakic children have corneal thickness readings that are much thicker than average, yet they develop vision loss and visual field loss with glaucomatous nerve changes at a much higher rate than other children.[9] The diagnostic value of CCT in childhood glaucoma has yet to be determined, so you should use caution when comparing these readings to adult normative information.

Visual field testing is an important way to confirm the diagnosis of glaucoma, but the information is usually less reliable in children (especially young children) than adults. It is difficult to get any useful visual field information in children until they reach the age of 6 to 8 years, and artifactual changes are the norm rather than the exception.[10,11] Artifactual issues with initial visual fields may persist until patients enter the early teenage years, and a learning curve is common. In the author's experience, Goldmann visual fields are easier for children to perform and can provide useful information until these patients are mature enough to finish an automated test successfully.

THE CASE PRESENTED

For the case presented at the start of this article, a careful history and examination can reveal any findings that may place the child at higher risk for glaucoma (Axenfeld-Rieger syndrome can be very subtle and only visible on gonioscopy). An evaluation of the patient's family members for a large cup-to-disc ratio may provide some reassurance that the cupping is physiological.

A visual field test would usually be the first test ordered, and a 10-year-old patient is generally able to provide some information on the first attempt. Do not, however, expect a clean test on the first try. If the visual field looks relatively normal, optic disc imaging or photography is usually the next step to document the

appearance of the optic nerve. Imaging of the optic nerve or nerve fiber layer provides the advantage of immediate diagnostic comparison, whereas disc photography assists with later comparisons. Be sure, however, to take into account the previously mentioned caveats regarding normative comparisons and movement. CCT should be interpreted in light of the available normative pediatric data and correlated with other factors such as a family history of glaucoma.

In this case, Goldmann visual field testing was full bilaterally. An OCT evaluation of the nerve fiber layer was within adult normative values. CCT was in the range of 530 µm OU. Repeated IOP assessments have been normal, and there has been no change in the optic discs' appearance with serial examinations.

CONCLUSION

The child with a large cup-to-disc ratio and normal IOP often represents a case of physiological variation, not incipient glaucoma. These patients are likely at increased risk of developing glaucoma, but their risk of developing the disease in the near and intermediate term may be quite low. Proper management in these cases usually involves documenting the appearance of the optic disc, with routine surveillance for elevated IOP, changes in the disc, or reproducible visual field abnormalities. ❑

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