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Grand Rounds

A 25-month-old girl with vision loss, nystagmus, and anomalous head posture

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History

A 25-month-old, Caucasian girl presented for evaluation of high myopia, decreased vision, new onset nystagmus and anomalous head posture (AHP). The parents noted decreased vision, nystagmus, photophobia, nycloptia and AHP 8–10 weeks prior to examination. The patient had an uncomplicated prenatal and birth history with subsequent normal growth and development. She was healthy other than a history of uncomplicated otitis media. There was no history of trauma, recent illness or new medications. Her family history was notable for myopia, amblyopia, and a paternal cousin diagnosed with "optic neuropathy" at age 3 years.

Examination

Initial exam revealed an alert, cooperative, age-appropriate 25-month-old girl with a left head turn of approximately 30–40 degrees. The patient was noted to have central, unsteady, and unmaintained fixation OU. The pupils were of equal size without an afferent pupillary defect. During an MRI under sedation, intraocular pressures (IOP) were 32–36 mmHg OD and 30–38 mmHg OS. Her corneal diameters were 12.5 mm OU without evidence of edema, infiltrate, or neovascularization. Haab's striae ran longitudinally in the inferior corneas of both eyes. On gonioscopy, the angle appeared anomalous with a high anterior insertion of a flat iris. The lenses and vitreous were clear OU. Dilated fundoscopic exam demonstrated a 0.9 × 0.9 cup-to-disc ratio OD and 0.9 × 0.8 cup-to-disc ratio OS. Both nerve heads showed pallor and deep cupping. An image of the left nerve is shown in Figure 1. Her refraction was −9.00 +2.00 ×180 OD and −7.00 +2.50 ×180 OS. The MRI was normal. Combination timolol-dorzolamide ophthalmic drops were initiated, trabeculotomy was scheduled, and the patient was referred for further evaluation of her nystagmus.

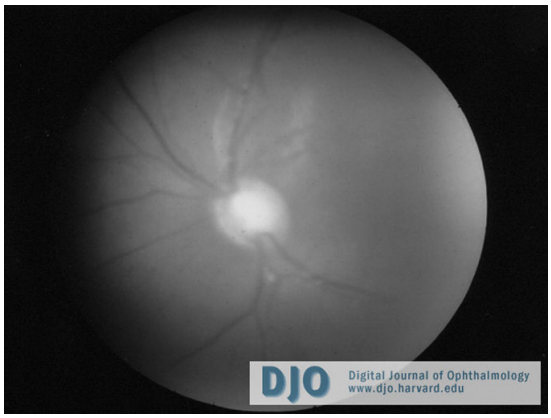

Figure 1. Fundus photograph of left optic nerve showing extreme cupping and atrophy present in both nerves at the time of diagnosis.

Additional examination revealed full versions and ductions and a comitant esotropia. The patient had a left AHP, preferring right gaze. She had variable, symmetric, conjugate, small-to-moderate amplitude, and moderate-

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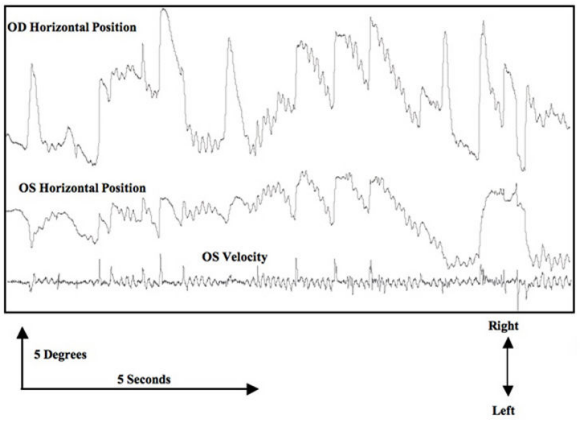

Figure 2. Ocular motility recording showing position trace OU and velocity trace OS at the time of diagnosis (right gaze is up and left gaze is down). The recording shows well-developed jerk and bidirectional jerk waveforms with periods of foveation, typical of infantile nystagmus syndrome.

to-high frequency multiplanar involuntary ocular oscillations with a null position in right gaze of 30–40 degrees. There was no latent component and saccades, pursuit, and vestibulo-ocular reflexes were otherwise normal.

Ancillary Testing

Oculographic recording showed constant, symmetric, multiplanar involuntary oscillations, which varied from 1.5–7 Hz with jerk, asymmetric pendular and bidirectional jerk slow phases (Figure 2).

Treatment

Over the next 18 months, the patient underwent 360 degrees of unsuccessful trabeculotomy on each eye followed by uncomplicated placement of Baerveldt implants in both eyes and a bilateral medial rectus recession. Throughout the course of these interventions, her vision continued to improve and her exam stabilized. Her IOP 18 months after presentation was 10–12 mmHg OD and 15–17 mmHg OS. The cup-to-disc ratio was 0.4–0.6 OU, and her refraction was −4.00 +1.00 ×180 OD and −5.00 +2.00 ×180 OS. Visual acuity using HOTV optotypes was 20/70 OU, and the AHP, nystagmus and esotropia were decreased.

Diff DX

Infantile nystagmus and compensatory head posturing may be associated with any process that results in visual deprivation such as media opacities, optic nerve disease, retinal abnormalities, or abnormalities of the afferent visual pathways. It is also associated with metabolic disease, drug use, or structural disease of the brainstem and cerebellum. Careful patient history revealed no birth or childhood trauma, patient medications, drug or alcohol use by the mother, or other congenital disease. Since the child was otherwise developmentally normal, serious metabolic disease or neurodegenerative disorder was highly unlikely. Detailed examination demonstrated no opacities of the lens or vitreous, the globe was structurally normal, and the retina appeared normal. A normal MRI ruled out any structural brain disease.

In this case, detailed examination of the patient revealed elevated intraocular pressures, severe myopia, Haab's striae, and enlarged optic cups with pallor and deep cupping. The combination of these findings point to advanced infantile glaucoma.

Diagnosis

The discovery of primary congenital glaucoma and decreased vision as the cause of an AHP and infantile nystagmus syndrome (INS) is unusual. Many disorders that affect vision and visual development in the first few weeks to months of life (e.g., cataracts, aniridia, achromatopsia and foveal hypoplasia) result in INS.[1–4] Despite the unusual presenting complaint, her exam was consistent with advanced primary congenital glaucoma. Goniotomy or trabeculotomy remains the preferred first surgical approach. Both procedures achieve greater than 80% success, but this number is markedly reduced in patients younger than 2 months or older than 2 years.[5] The use of drainage devices is reserved for difficult cases, where success rates may approach 95%.[5–7] Surgical success was achieved in this case with Baerveldt implants following failed trabeculotomies.

The reduction in the patient's IOP to an acceptable range had profound effects on her vision and ocular anatomy. Her myopia was significantly reduced and the optic discs demonstrated reversal of cupping. This latter finding is common in congenital glaucoma and differentiates it from the adult glaucomas. As her vision improved the nystagmus and AHP decreased. The development of INS in this patient is a result of poor ocular motor calibration due to associated glaucomatous sensory system deficits (optic neuropathy, myopia, corneal irregularities and strabismus).

INS is the most predominant type of ocular oscillation associated with visual deprivation.[2,4,8] Although the exact etiology of INS is unknown, it likely caused by abnormal communication between the developing sensory and motor systems leading to a defect of ocular

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motor calibration. Patients with INS typically present with a horizontal nystagmus characterized by a slow and fast phase that increases in intensity with fixation and decreases with sleep, inattention or convergence. The oscillations change direction about a neutral position and a null point in eccentric gaze manifests as an AHP.[9] Diagnosis is confirmed by oculography that demonstrates waveforms having slow phases that display an increasing velocity exponential curve associated with periods of foveation. AHP is strongly associated with INS.[9] Treatment of AHP due to an eccentric gaze null zone associated with INS is directed towards returning the head to a more normal position and decreasing the intensity of the oscillation. In this case, treatment of the glaucoma, myopia and strabismus improved vision and resulted in a decrease in the nystagmus and the AHP. Recognition of congenital glaucoma in this patient who presented with new-onset nystagmus and AHP was essential in preventing eventual blindness.

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