

Unilateral Optic Nerve Hypoplasia in a patient desiring surgical treatment for exotropia

Michael S. Floyd, MD, Christy Benson, and Susannah Q. Longmuir, MD

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Chief Complaint: 17- year-old male presents for surgical treatment of longstanding left exotropia

History of Present Illness: This patient has a longstanding sensory left exotropia and is interested in strabismus surgery. His left eye has turned out since birth. He thinks that patching was attempted when he was younger. However, he thinks the patching was not successful, because of his non-compliance as a child.

Past Ocular History: Left amblyopia and left sensory exotropia.

Past Medical History: Sinusitis and seasonal allergies

Medications: None

Allergies: No known drug allergies

Family History: Mother has a history of strabismus surgery. His paternal grandmother has diabetes.

Social History: Quit smoking 3 months ago. Smoked ½ pack per day for 7 years. He is attending a state school for boys due to delinquent behavior.

Physical Exam:

- General: well-appearing patient with no acute distress
- Best Corrected Visual Acuity:
 - OD 20/25
 - OS HM (hand motion) at 1 foot
- Extraocular Motility: -1 adduction deficit OU, superior oblique overaction OU, and inferior oblique overaction OU. Left exotropia measures 50 prism diopters at near and in the distance in central gaze.
- Pupils: 6 mm OU, brisk reactivity OU, and left afferent pupillary defect.
- Intra ocular pressure: 15 OD and 13 OU (Tonopen)
- External and anterior segment examination: WNL OU
- Confrontation visual fields: full to count fingers OD and infratemporal quadrantanopia OS.
- Dilated fundus exam: See figure 1.

Clinical Course: Refraction did not improve his visual acuity. Optic nerve optical coherence tomography (OCT) was obtained (figure 2). An MRI of the brain was obtained to rule out midline abnormalities, given his behavioral issues. The study demonstrated a hypoplastic left nerve and tract (figure 3 and 4). Otherwise normal cerebrum and cerebellum, with no midline intracranial abnormalities. The pituitary gland was present. He underwent a left medial resection and left lateral recession with an adjustable suture. Initially, his ocular alignment measured 15 prism diopters of esotropia and 2 months later measured 5 prism diopters exotropia. The patient was happy with the outcome.

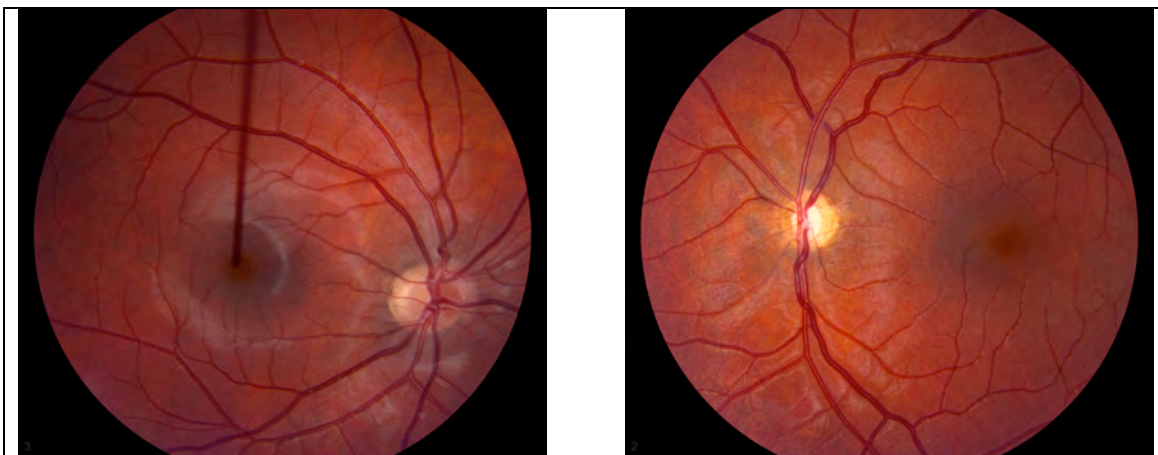


Figure 1: Color fundus photographs of this patient’s right and left eye. The left photo is a picture of the right fundus. It has a normal optic nerve with peripapillary atrophy. The vessels and macula are within normal limits. The right photo is a picture of the patient’s left fundus. The optic nerve is markedly small and pale. It has peripapillary atrophy or a “double ring” sign. The vessels and macula are within normal limits.

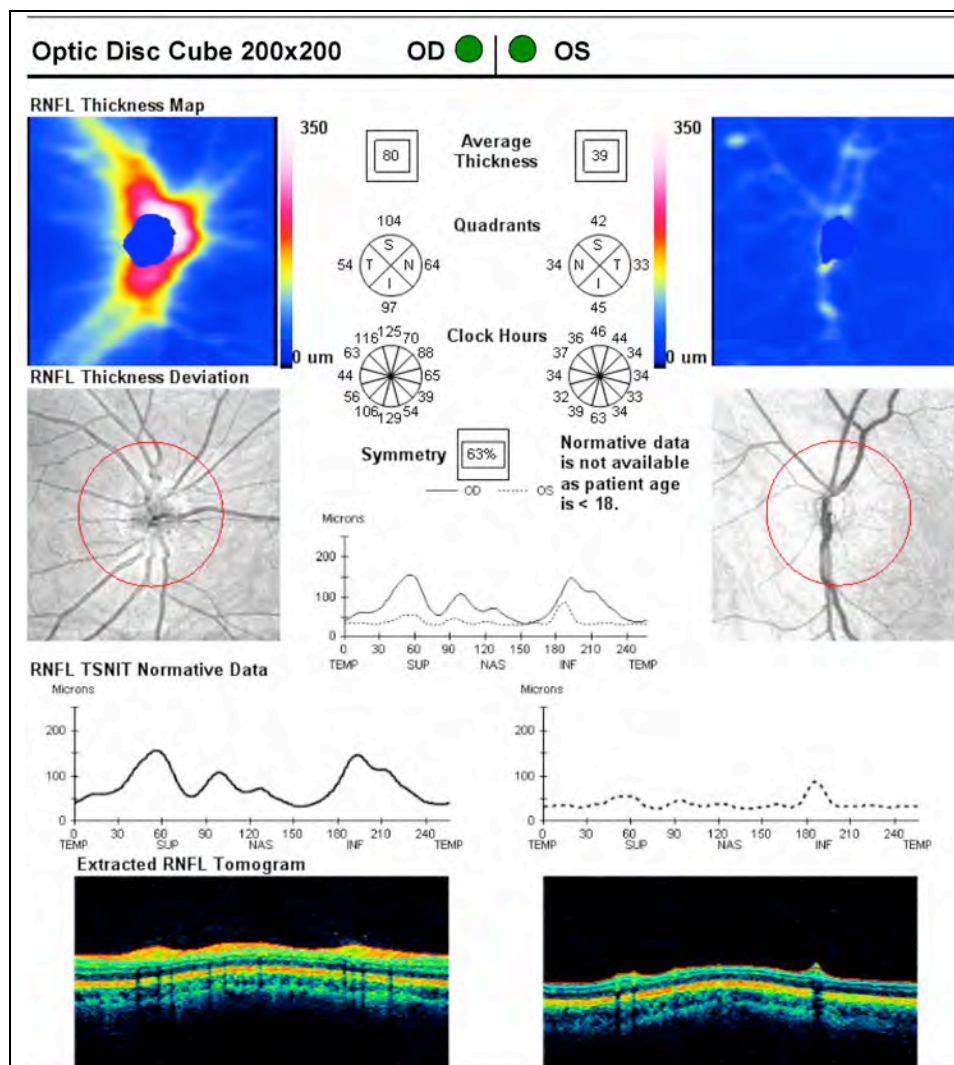


Figure 2: Cirrus OCT of bilateral optic nerves is presented. The study is of excellent quality. No normative data is available as this patient is under 18 years of age. The right optic nerve has an average thickness of 80 microns. The left optic nerve half the diameter of the right nerve and its average thickness is 39 microns.



Figure 3: MRI brain Axial T1 FS post GAD demonstrating left ON hypoplasia

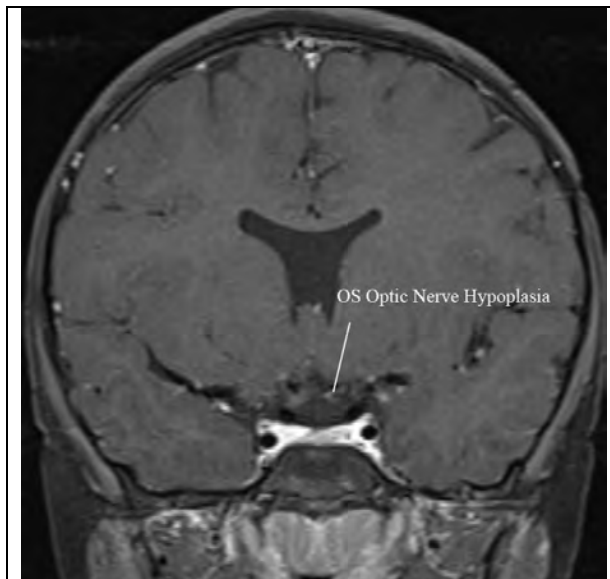


Figure 4 : MRI Brain Coronal T1 FS thin post GAD demonstrating left ON hypoplasia (note : this imaging sequence was taken from another patient with ON hypoplasia, absent septum pellucidum, and pituitary hypoplasia).

Discussion:

Optic nerve hypoplasia is a rare congenital disorder that may be an isolated finding or occur in association with another syndrome. There is an extremely wide range of severity: visual acuity in the affected eye(s) correlate to the severity of hypoplasia, falling anywhere between 20/15 to no light perception. Histologically, the retinal nerve fiber layer and ganglion cell number are decreased, while the outer retinal layer is unaffected.[1] Even if visual acuity is normal, patients with optic nerve hypoplasia can have visual field defects, most commonly in the nasal or inferior fields of vision.[2] It can be a bilateral (75% of cases[3]) or a unilateral finding and may be associated with absence of the septum pellucidum, thinning or absence of the corpus callosum, cerebral hemisphere abnormalities, anencephaly, and/or dysfunction with the hypothalamic-pituitary axis. Therefore brain imaging should be performed on all infants with a diagnosis of optic nerve hypoplasia.

Children with bilateral optic nerve hypoplasia more commonly have associated neurologic abnormalities (57%) when compared to children with unilateral optic nerve hypoplasia (32%). Failure of the retinal ganglion cell layer to differentiate is the main cause of optic nerve hypoplasia, although the true pathogenesis remains unknown.[1] A 2009 case study reported a patient found to have optic nerve hypoplasia and a pericentric inversion of upon chromosomal analysis.[4] Infants born to mothers with diabetes are at increased risk for optic nerve hypoplasia. Other known risk factors include quinine, phenytoin, or alcohol ingestion by the mother during pregnancy.

Infants with unilateral optic nerve hypoplasia with absent light perception in the affected eye usually present with strabismus and an afferent pupillary defect. For infants with bilateral optic

nerve hypoplasia, they commonly present with a course, pendular nystagmus.[2, 3] The classic finding in patients with optic nerve hypoplasia is a decreased diameter of the optic disc. Other common findings with fundus examination include optic disc pallor, vascular tortuosity, and abnormalities in the retinal vasculature. Externally, patients may have microphthalmia, aniridia, or cranial nerve IV/VI palsies. In a chart review of 100 infants with optic nerve hypoplasia, 32% were developmentally delayed, 13% had cerebral palsy, and 12% had seizures.[3]

The optic nerve forms in human embryos at around 6 weeks gestation. Extension of the optic nerve fibers to the chiasm is complete by 8 weeks gestation.[2] Embryologic development of the corpus callosum and septum pellucidum occurs roughly at the same time as the optic nerve, hence the increased rate of coexisting anomalies. When optic nerve hypoplasia occurs with absence of the septum pellucidum, it is known as septo-optic dysplasia (de Morsier syndrome). Only about 10% of children with optic nerve hypoplasia have septo-optic dysplasia, and about 50% of these children have associated endocrine abnormalities.[3] Even though endocrine abnormalities are a rare finding in patients with optic nerve hypoplasia, all patients should be seen by an endocrinologist to rule out the possibility of potentially fatal endocrine complications such as sudden infant death due to thermoregulation problems, dehydration, and congenital hypothyroidism. The growth charts of patients with de Morsier syndrome should be monitored closely because of the potential for decreased release of growth hormone. Optic nerve hypoplasia can also be associated with cerebrohepatorenal syndrome of Zellweger, an extremely rare leukodystrophy characterized by decreased peroxisomes in renal, liver, and brain cells.[2]

Patients with unilateral optic nerve hypoplasia may have normal to severely diminished acuity. Those with poor vision may have strabismus. Treatment options include observation, a pirate patch, prism, corrective spectacles to minimize the appearance, and surgery. Primarily, the surgical site should be the affected eye and the surgeon should consider altering the standard surgical measures, as patients with neurologic deficits do not have a normal response to surgery.[5, 6]

Diagnosis: Optic Nerve Hypoplasia

Differential diagnosis for a child with a hypoplastic nerve:

- Axial myopia
- Myelinated nerve fibers
- Optic nerve drusen
- Scleral crescent
- Tilted disc

<p>EPIDEMIOLOGY</p> <ul style="list-style-type: none"> • 2/100,000 infants[2] • 75% of affected infants have bilateral involvement • Risk factors include maternal diabetes, phenytoin, quinine, and alcohol consumption during the first trimester 	<p>SIGNS</p> <ul style="list-style-type: none"> • Strabismus (unilateral) • Course, pendular nystagmus (bilateral) • Absence of corpus callosum or septum pellucidum • Developmental delay • Pallor and/or decreased diameter of optic disc, vascular tortuosity, and abnormalities in the retinal vasculature • External eye: microphthalmia, aniridia, CN IV/VI palsies
<p>SYMPTOMS</p> <ul style="list-style-type: none"> • Normal to severely depressed visual acuity • Visual field defects 	<p>TREATMENT</p> <ul style="list-style-type: none"> • Low vision consult • Referral to an endocrinologist • Consider MRI of the brain

References

1. Zeki SM, Dutton GN: Optic nerve hypoplasia in children. *The British journal of ophthalmology* 1990, 74(5):300-304.
2. Ouvrier R, Billson F: Optic nerve hypoplasia: a review. *Journal of child neurology* 1986, 1(3):181-188.
3. Garcia ML, Ty EB, Taban M, David Rothner A, Rogers D, Traboulsi EI: Systemic and ocular findings in 100 patients with optic nerve hypoplasia. *Journal of child neurology* 2006, 21(11):949-956.
4. Cheung RT, Wong AM: Optic nerve hypoplasia associated with chromosome 9 inversion. *Canadian journal of ophthalmology* 2009, 44(5):610-611.
5. Pickering JD, Simon JW, Lininger LL, Melsopp KB, Pinto GL: Exaggerated effect of bilateral medial rectus recession in developmentally delayed children. *Journal of pediatric ophthalmology and strabismus* 1994, 31(6):374-377.
6. Habet-Wilner Z, Spierer A, Glovinsky J, Wygnanski-Jaffe T: Bilateral medial rectus muscle recession: results in children with developmental delay compared with normally developed children. *J AAPOS* 2006, 10(2):150-154.

Additional Reading

- Braverman R. Optic Nerve Aplasia. In: Hay W, editor. Current Diagnosis and Treatment: Pediatrics. 17th ed. New York: Lange; McGraw Hill Medical; 2009.
- Lambert SR, Hoyt CS, Narahara MH. Optic nerve hypoplasia. Survey of ophthalmology. [Review]. 1987;32(1):1-9.
- Skarf B, Hoyt CS. Optic nerve hypoplasia in children. Association with anomalies of the endocrine and CNS. Archives of ophthalmology. 1984;102(1):62-67.

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