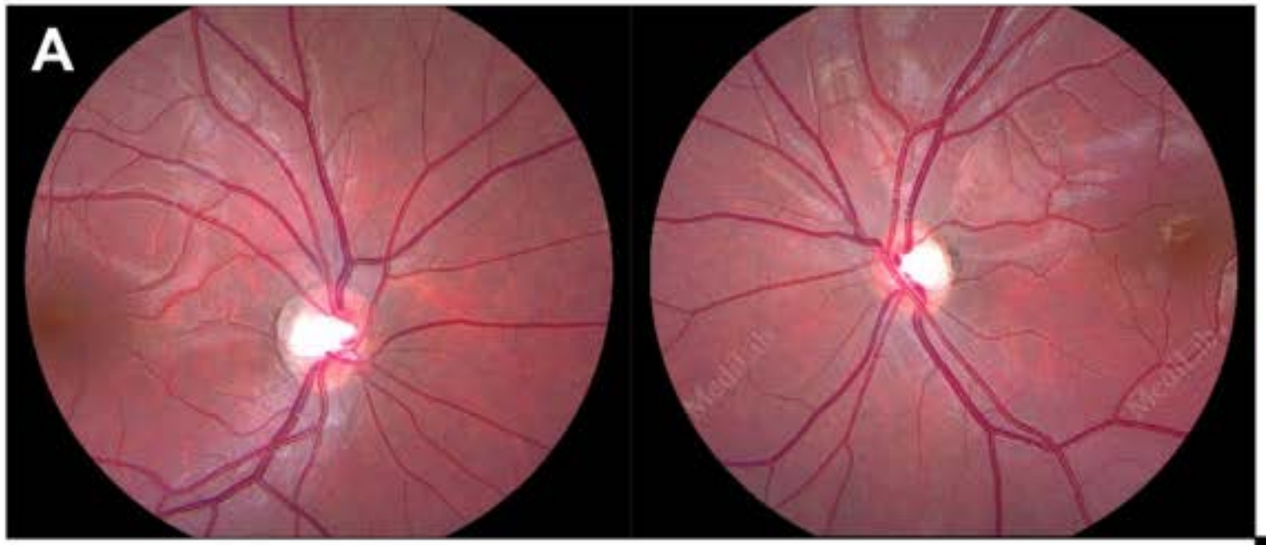
A



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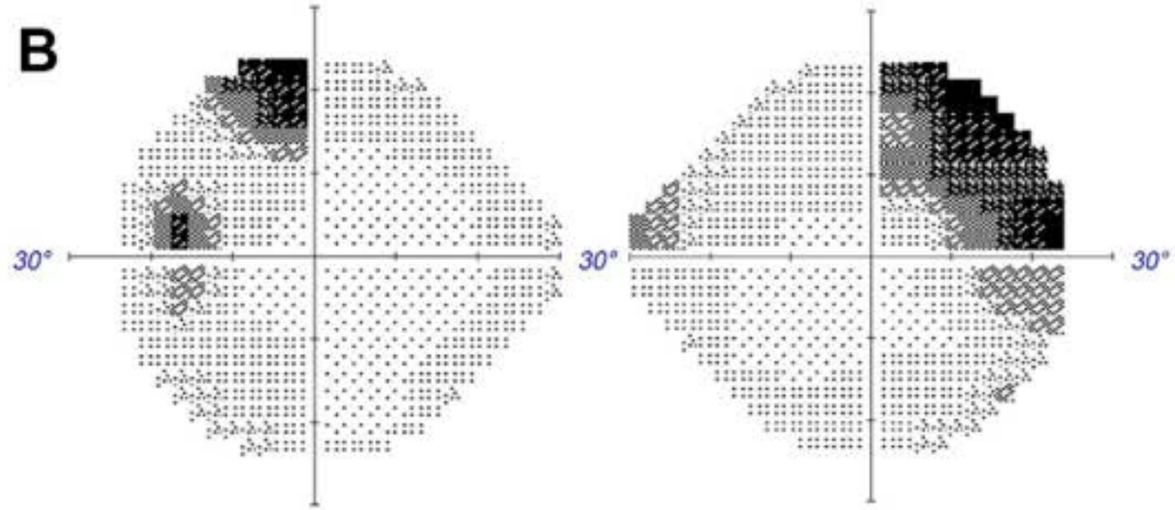
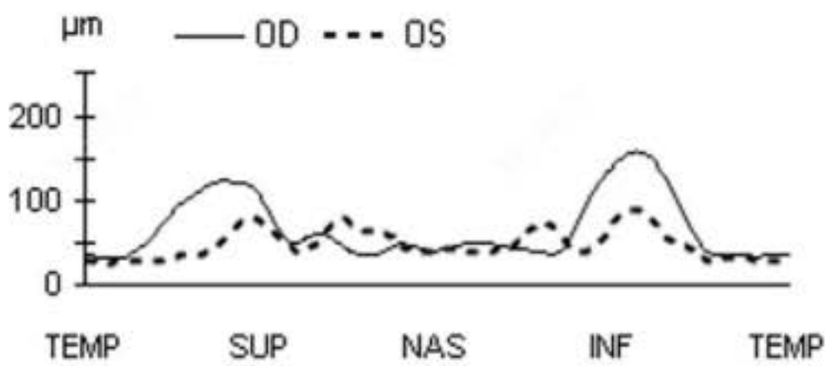


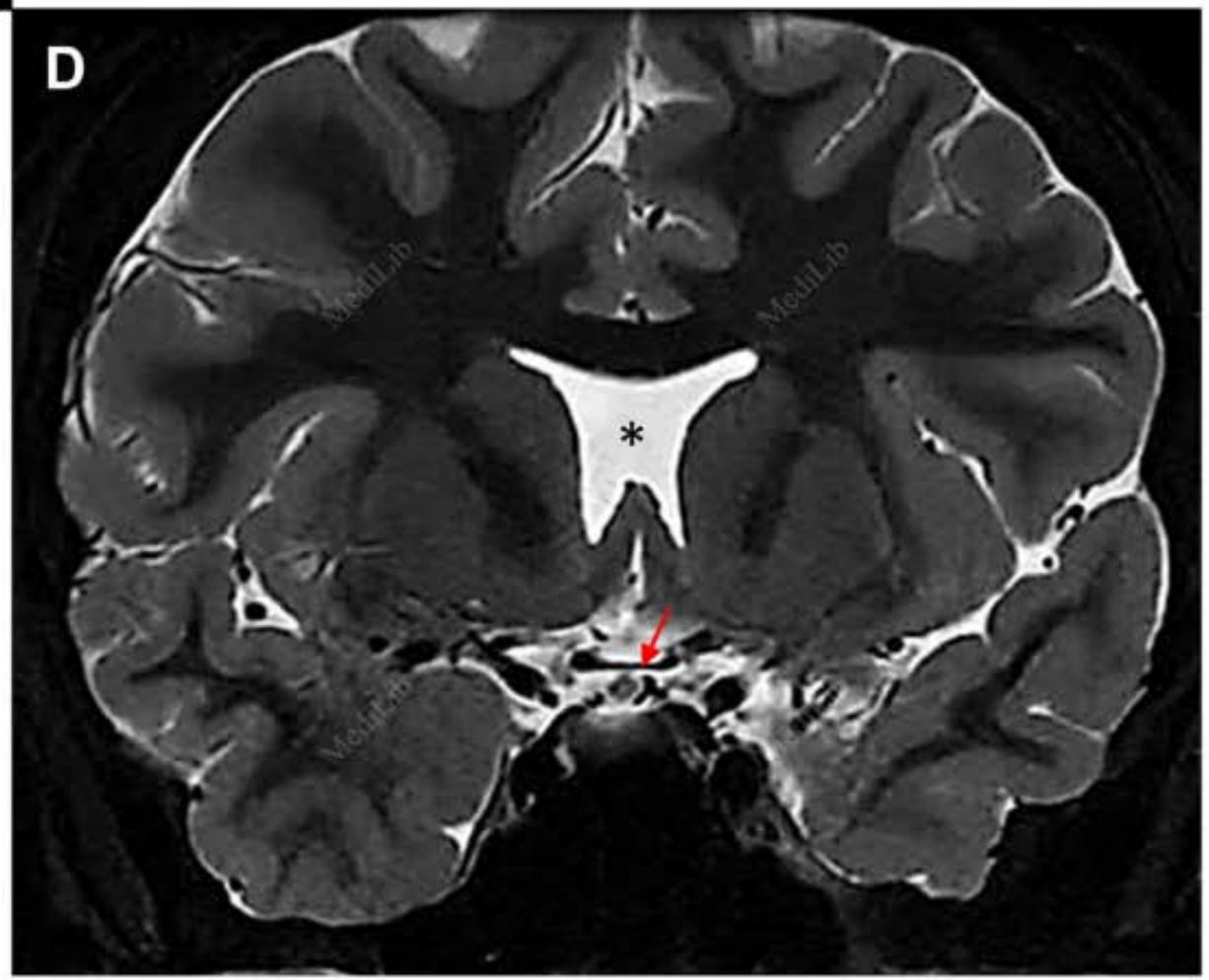
C

	OD	OS
Average RNFL Thickness	68 µm	50 µm
RNFL Symmetry	53%	
Rim Area	1.00 mm²	0.84 mm²
Disc Area	1.62 mm²	1.27 mm²
Average C/D Ratio	0.62	0.59
Vertical C/D Ratio	0.44	0.56
Cup Volume	0.280 mm³	0.195 mm³

RNFL Thickness



D



Extensive Horizontal Optic Disc Cupping as a Sign of Chiasmal Hypoplasia

A 13-year-old girl was referred for suspicion of glaucoma. Examination revealed extensive horizontal cupping of the right optic disc and mild hypoplasia of both optic nerves (**A**) with otherwise normal color vision, intraocular pressures, and neuro-ophthalmic examination. Humphrey visual fields showed bilateral superotemporal defects respecting the midline (**B**). OCT confirmed optic nerve hypoplasia, more severe in the left eye, with thinning of the nasal and temporal retinal nerve fiber layer (RNFL) bilaterally (**C**). Magnetic resonance imaging showed chiasmal hypoplasia, more severe on the left side (red arrow), and an absent septum pellucidum (asterisk) (**D**). Extensive horizontal optic disc cupping can be a sign of congenital chiasmopathy (Magnified version of Figure **A**-**D** is available online at www.aaojournal.org/).

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Optic nerve hypoplasia is the most common congenital optic nerve anomaly. It is characterized by a reduced number of ganglion cell axons within the optic nerve, and it can occur in isolation or associated with other CNS abnormalities. When optic nerve hypoplasia occurs in conjunction with midline brain abnormalities, the absence of the septum pellucidum and agenesis or thinning of the corpus callosum, it is known as septo-optic dysplasia or de Morsier's syndrome.

Septo-optic dysplasia is a rare congenital anomaly and has a reported prevalence of 6.3 per 100,000. It has a near equal distribution among males and females,[1] and it is typically diagnosed in newborns and infants.While absence of the septum pellucidum is not associated with specific cognitive or developmental abnormalities, visual loss and hypopituitarism associated with the midline brain abnormalities contribute to significant morbidity. Early detection and treatment of endocrine deficits can be life saving.

Patients with septo-optic dysplasia may present to medical professionals for a variety of reasons. From 60 to 90 percent of these patients present with blindness or symptoms of visual impairment. Signs related to visual loss include absent fixation, searching nystagmus, visual inattentiveness and strabismus. Three-fourths of patients may have other systemic conditions such as mental retardation, epilepsy or cerebral palsy or behavioral problems such as attention deficit disorder or autism. Other developmental abnormalities include underdeveloped midfacial structures, hypoglycemia, hyperbilirubinemia, disturbance in temperature regulation, muscular hypotonia, microgenitalism, low Apgar scores and failure to thrive.

Optic nerve hypoplasia is always present in septo-optic dysplasia. The optic nerve head is often gray or pale and typically one-third to one-half normal size. Classically, the optic nerve is surrounded by a ring of visible sclera and annular pigmentation, the “double ring” sign. The outer ring is the junction of the sclera and the choroidal pigment with the lamina cribrosa, and the diameter corresponds approximately to the size of a normal disc. The inner ring is darker and represents the junction of the termination of the retinal pigment epithelium with the hypoplastic optic nerve. The disc may or may not have significant cupping, and it can have spotted intradisc pigmentation. The nerve fiber layer can be thin due to the varying degrees of absent ganglion cells and their axons. Retinal vessel tortuosity may also be present.

The visual acuity with optic nerve hypoplasia ranges from 20/20 to no light perception, with most patients having visual acuity of 20/200 or worse. Because visual acuity is determined by the integrity of the papillomacular bundle, it does not necessarily correlate with the overall size of the optic disc.Invariably, however, affected eyes will show localized visual field defects, often combined with a general constriction of the visual fields. Optic nerve hypoplasia can occur unilaterally or bilaterally, and if bilateral it can be asymmetric. When it is unilateral or bilaterally asymmetric, there can be an associated relative afferent pupillary defect.

Maternal age. Young maternal age has been associated with septo-optic dysplasia as well. The syndrome has been found to occur with increased frequency in the children of mothers who give birth in their middle to late adolescence. Maternal insulin-dependent diabetes mellitus is also associated with superior segmental optic nerve hypoplasia.

Of patients with optic nerve hypoplasia, 45 percent also have cerebral hemispheric migration anomalies (schizencephaly, cortical heterotopias) or hemispheric injury (periventricular leukomalacia, encephalomalacia), which can be detected with MRI. These abnormalities are highly predictive of neurodevelopmental deficits. Absence of the septum pellucidum alone is not predictive of visual acuity, specific cognitive abnormalities or spatial orientation deficits. Thinning or agenesis of the corpus callosum alone is not predictive of neurodevelopmental defects; however, it is frequently associated with cerebral hemispheric abnormalities.

Fifteen percent of patients with optic nerve hypoplasia demonstrate neurohypophyseal abnormalities on MRI. In normal patients, the anterior and posterior pituitary and the infundibulum appear as a bright spot on high-resolution MRI. This is thought to be due to the posterior pituitary hormones and the phospholipid content of the vesicles harboring the hormones. Endocrine dysfunction is associated with absence of the infundibulum and with posterior pituitary ectopia. Absence of the infundibulum and its surrounding portal venous system results in the inability of the hypothalamus to stimulate the anterior pituitary, resulting in anterior pituitary hormone deficiency. The posterior pituitary hormones accumulate proximal to the site of injury and form an ectopic nodule, which shows as a hyperintense nodule at the median eminence on T1-weighted images. This bright spot is where the infundibulum is normally located. Hormone secretion from the ectopic tissue is sufficient to maintain posterior pituitary function; however, ectopia is nearly always associated with anterior pituitary dysfunction.