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Optic Nerve Aplasia (Uniateral)

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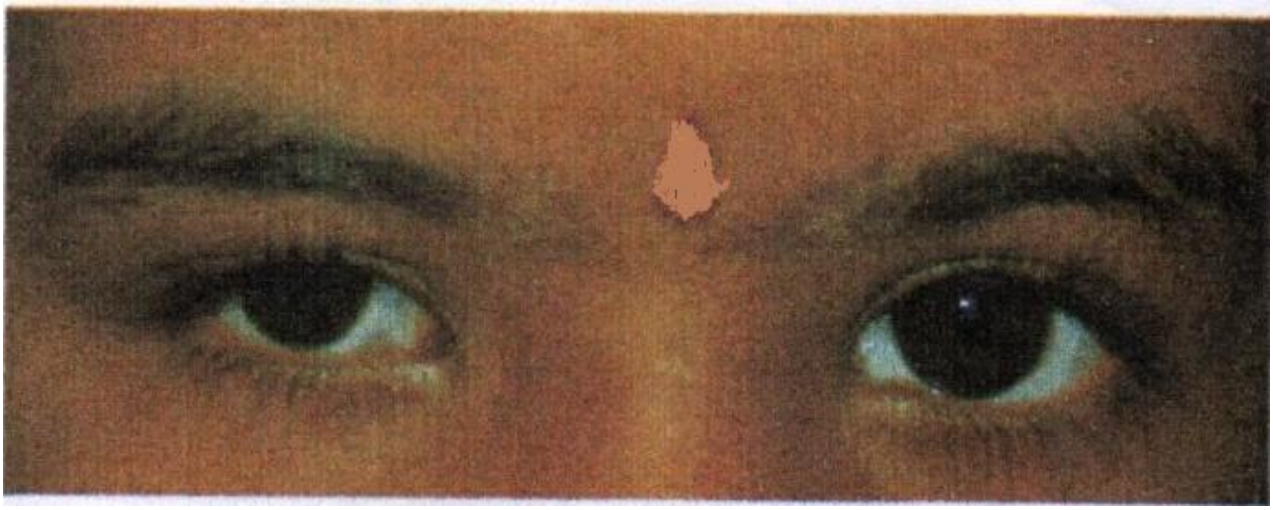
Abstract
Optic nerve aplasia is a rare developmental anomaly consisting of absence of the optic nerve, retinal blood vessels and ganglion cells. ONA may be an isolated finding or associated with congenital ocular and non-ocular abnormalities. It has frequent association with microphthalmos and other malformations confined to the involved eye. Unilateral ONA is generally associated with normal brain development, while bilateral ONA is usually accompanied by other central nervous system (CNS) abnormalities.
Keywords: *Microcornea, Enophthalmos, Magnetic resonance imaging (MRI), Optic disc, ONA, VEP.*

Introduction

The unilateral veriety tends to be more benign, most comonally associated with unilateral microphthalmos. Aplasia of the optic nerve is a very rare ocular abnormality. In aplasia there must be complete absence of the optic nerve (including the optic disc), retinal ganglions and nerve fiber layers and central retinal vessels. Histologically there is absence of the optic nerve, retinal ganglion cells and, nerve fiber layer and optic nerve vessels. Magnetic resonance imaging (MRI) in unilateral aplasia show microphthalmos, absence of optic nerve, optic chiasma tracts on the side of aplasia.



Shows the right eye of the child having microcornea with mild enophthalmos

Case Report

A 5 year old boy came in our OPD with parents noticing his right eye to be smaller than right eye since birth. Family history was negative for ocular or other birth defects. He was the first child born out of a non consanguineous marriage, term pregnancy and normal delivery with a birth weight of 3000 grams. The right eye had microcornea. Opthalmic examination revealed that there was no light perception in the right eye. The right pupil did not react to direct light stimulation and there was no consensual reaction. The right eye had a normal iris, normal colour pattern, anterior chamber depth, and lens was clear. Gonioscopic examination showed open angles in both eyes. Dilated fundus examination the right eye showed absence of optic disc and central retinal vessels. There was marked choroidal tessellation with multiple areas of chorioretinal atrophy. The left eye fundus examination revealed normal disc, macula, retinal vessels and normal peripheral retina. On A-scan ultrasound, the mean axial length was 19.8 mm in the right eye and 21.5 mm in the left eye. MRI scan of orbits and brain revealed remnants of ON sheaths with some glial tissue were present as a thin cord in posterior orbit. On right side the optic chiasma appeared asymmetric due to chiasmal aplasia. There was mild enopthalmos of the right globe.

Signs and Symptoms

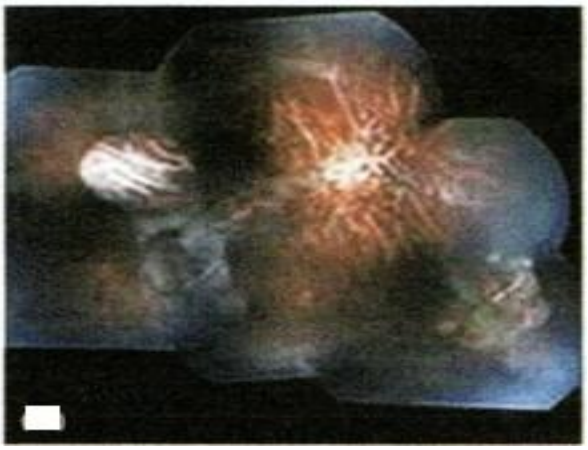
Fundus photograph of left eye showing normal disc, macula, retinal vessels and normal peripheral retina.

Anterior segment abnormalities with ONA included anterior segment dysgenesis, microphthalmos, cataract, sclerocornea, microcornea, hypoplasia of the corneal stroma, corneal edema independent to intraocular pressure or due to glaucoma. Vision is no light perception (NLP) with no direct or consensual papillary response to light and a positive RAPD.

Pathology

Other reported abnormalities include the presence of retinal pigment epithelium over the area of the optic disc, remnants of the dural sheath, rudimentary retinal vessels entering the posterior pole in a chaotic manner. Absance of ganglion cells, optic nerve fibers and retinal vessels in eyes with ONA have been described before. The inner layers of the retina were more markedly hypoplastic than the outer layers.

Embryology and Pathogenesis



Right eye montage photograph of the fundus showing absence of the optic disc and retinal vessels Numerous large round to oval, circumscribed, whitish to yellow areas of retinochoroidal hypopigmentation are visible on the posterior pole.

The optic nerve develops from the optic stalk, the original connection between the optic vesicle and the forebrain. The pathogenesis of ONA remains unknown. Any problem in formation of retinal ganglion cell (RGC) axons or their guidance, exemplified by deranged Netrin and EPH/ephrin molecule formation as guidance molecules, may seriously impact the development of optic nerve.

Genetics

The information regarding the genetic basis in ONA is limited. Mutation in PAX6 and OTX2 have been documented [1241] Meireetal[12]

Systemic Anomalies

Unilateral ONA is generally associated with normal brain development, while most bilateral cases have CNS derangement. ONA and its chorioretinal lacuna can also overlap with the autosomal-dominant microcephaly-lymphedema-chorioretinal dysplasia syndrome.

Conclusion

Diagnosis of optic nerve abnormalities in children requires a thorough ophthalmic examination and proper ancillary testing. A thorough systemic evaluation along with neuroimaging is recommended in children with ONA to rule out CNS malformations and endocrinological abnormalities. Neuroimaging such as MRI may be of some diagnostic value for documenting ONA and associated conditions.

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