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# Optic Nerve Aplasia and Microphthalmos: A Case Report

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**Abstract-**

Optic nerve aplasia (ONA) is a rare developmental anomaly characterized by the congenital absence of the optic nerve, central retinal vessels, retinal ganglion cells and optic nerve fibers. ONA is most often seen in a unilaterally malformed eye and seems to fall within a malformation complex that is fundamentally distinct from optic nerve hypoplasia, although numerous case reports in the ophthalmic literature misidentify aplasia and described hypoplasia of the optic nerve instead. There is no racial predisposition and males and females are similarly affected. Unilateral ONA is generally associated with normal brain development without evidence of an inherited factor. Family history is not consistent with Mendelian inheritance, and results of chromosome exams are within normal parameters.

This is contrasting with bilateral optic nerve aplasia cases that are far more uncommon and are usually accompanied by other central nervous system (CNS) derangements and a possible familiar history .To our knowledge, the largest series of optic nerve aplasia was published by Weiter et al., who described 11 unilateral cases of ONA and 2 atypical bilateral cases suspected to be an extreme form of hypoplasia of the optic nerve [8]. In most cases of unilateral ONA the patients display other spectrum of ocular abnormalities on the affected eye, with the most common being microphthalmos.

Histopathologic studies of 25 eyes with ONA showed microphthalmia in 20 of 25 eyes [5]. Colobomas, and anterior or posterior staphylomas are additionally present in the majority of eyes. Other clinical features may be a vestigial dural sheath; retinal dysplasia and retinal rosettes; retinal detachment; primary hyperplastic persistent vitreous; choroidal, iris or ciliary body hypoplasia; microcornea, microphakia, cataract and hypoplasia of the corneal stroma [5,8]. Esotropia of the affected eye is also commonly found. Case Report Our patient was a female infant, born from nonconsanguineous parents at 39 weeks of gestation with a birth weight of 3360 gr. Her parents’ familiar and ocular history was negative for malformations and genetic defects. The infant was the product of a full term pregnancy and elective caesarean section (due to a previous analogous caesarean section), the neonatal period was unremarkable. The girl was referred to the ophthalmologist at the age of 42 days because her parents noted an unequal size of the eyes.

The first evidences that emerged at the ophthalmological examination were microphthalmos of the right eye (16.84 mm x 17.40 mm for the right eye (OD) and 17.82 mm x 18.45 mm on the left eye) and the absence of a right pupillary afferent reflex. At the biomicroscopy the cornea appeared clear, the anterior chamber was formed, the lens was transparent and no evidence of coloboma, cataract, sclerocornea, nystagmus or strabismus was detected. At the RetCam fundus examination the ophthalmologist noted a dysplastic retina, with a complete absence of retinal vessels, optic nerve and optic disc on the OD .

A Magnetic resonance imaging (MRI) examination was performed. Coronal T1-weighted images showed the absence of the right optic nerve in the orbit and in the optic foramen, whereas the optic nerve was normal at all levels on the OS. The optic chiasm appeared asymmetric due to chiasmal hypoplasia on the right side. Coronal or axial T2- weighted images showed a decreased volume of the right optic globe and the absence of the right optic nerve whereas both optic tracts were presentchildren with partial trisomy 10q24.1-ter. The authors suggested a common specific genetic effect due to the similar

ophthalmological findings in these two patients .In a recent publication it was reported an autosomal-dominant form of nonsyndromic ONA, where the authors suggested a role of CYP26A1 and CYP26C1 in the pathogenesis of nonsyndromic ONA. The genetic source of optic nerve aplasia has been studied in mice. The recessive ocular retardation (or) mutation in the mouse has a complete penetrance in the homozygous state, and it has been linked to optic nerve aplasia, cataractous degeneration of the lens and microphthalmia .In insects, it was suggested that a mutation or inactivation on the eya gene, presented in specie of fly (Drosophila melanogaster), could be a possible explanation for the development of ONA since this gen is required for normal eye development. Moreover, as stated by the authors, function of the eya gene appears to be restricted to the eye. The authors speculated that although the eya gene is not found in mammals, perhaps eya function is conserved and an eya mutation could be responsible for ONA .Optic nerve aplasia has been previously linked with microphthalmos; however, this combination is usually reported to be in association with other ocular or systemic anomalies as contralateral microphthalmos aniridi or malformation of the anterior chamber and colobomas among others. In addition, Margo et al reported bilateral optic tracts hypoplasia on a 3-year-old girl with monocular microphthalmos who had optic nerve aplasia as revealed by mri

**References**

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