Rarus Congenitus Anomalia- A Rare Congenital Anomaly of Optic Nerve Aplasia

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Abstract:

Aplasia of the optic nerve is an extraordinarily rare congenital anomaly that affects one or both optic nerves associated with the absence of the central retinal vessel and ganglion cells. We present a case of unilateral optic nerve aplasia in a 37 weeks old neonate . On examination , the child had microcornea and unilateral optic nerve aplasia.

Case report:

This is a case of a 37 weeks old neonate who was delivered by lower segment caesarian section at the gestational age of 33 weeks. The weight of the neonate at birth was 2.2 kg . After delivery the child was kept in NICU for 2 days where oxygen was given . Single surface phototherapy was done for 5 hours followed by phototherapy for 2 days at a local hospital . After the neonate had stabilised hewas sent to our hospital for screening for Retinopathy of prematurity.

On anterior segment examination of right eye, microcornea was found.

On fundus examination no clear disc was seen . Anomalous vessels were present at the posterior pole. There were extensive colobomatous areas all around with retinal dysplasia (As seen in figure 1)

On anterior segment examination of left eye the anterior segment was within normal limit and on fundus examination , retina was found to be mature . (As seen in figure 2)

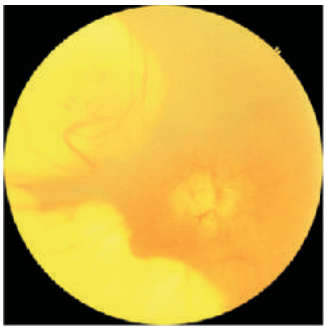


Figure 1: Image of the Right eye fundus showing no evidence of optic nerve head. Image taken on Nethra

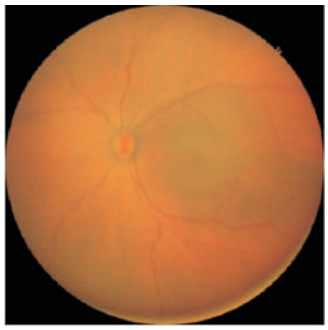
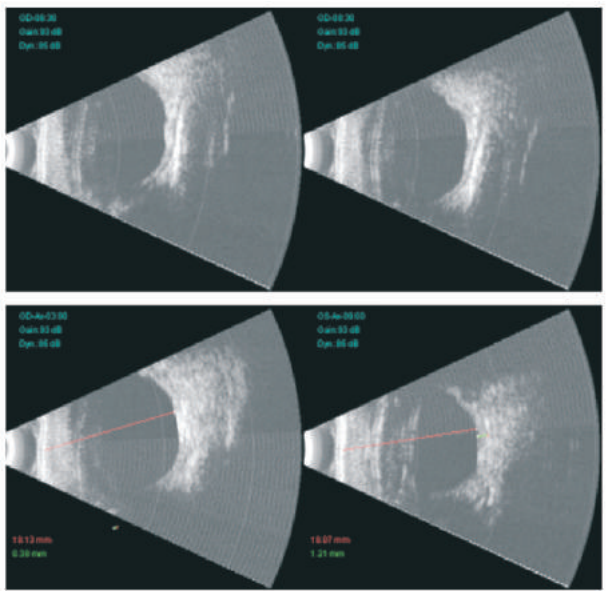


Figure 2: Left eye fundus showing mature retina . Images taken on Nethra forus camera

On B-Scan- There was no evidence of ONH shadow

Neonate's parents were advised to undergo a CT-scan for the neonate . When the parents reviewed with the report the



Comments: RE–Minimal Inferiror RC Coloboma involving disc & macula. ONH shadow not seen, ON absent, Vit cavity clear. No e/o RD or CD.
LE–Anatomically normal posterior segment.
AXL-18.07mm & ON normal. No e/o RD or CD.

next day it said–Increase in AP diameter of the right globe and thinning of posterior coat and aplastic right optic nerve noticed. Rest of the optic pathway was normal and left optic nerve was normal. Thus confirming the diagnosis of optic nerve aplasia in Right eye.

Discussion:

ONA is a rare developmental anomaly characterised by the absence of optic nerve and disc, ganglion cells and nerve fibre layeralong with retinal blood vessels.[1,2]

Earlier it was attributed as failure of the mesoderm to enter the foetal fissure and provide vascularisation of the retina and nerve tissue.[1]

Later reports suggested that ventral invagination of the optic vesicle caused nerve fibre misdirection and secondary atrophy.[3]

Yanoff and colleagues postulated a primary failure of ganglion cells to develop and send out axons as the cause of ON agenesis.[2]

Thus ONA can be due to defective formation of the embryonal fissure, failure of the mesenchymal anlage of the hyaloid system to enter the embryonal fissure or primary agenesis of the retinal ganglion cells.

Family history is not consistent with the mendelian inheritance and results of chromosomal examination in cases of ON aplasia are normal. Males and females are similarly affected.[5]

While unilateral ONA is rarely associated with brain or developmental anomalies , bilateral ONA is frequently associated with intracranial abnormalities. [5,7]

True ON aplasia is characterised clinically by blindness (no light perception), absent disc , absent central and branch retinal vessels and afferent pupillary defect.[6,7]

Histopathological examination to look for the absence of ganglion cells and nerve fibre layer along with the presence of vestigial dural sheath may help to differentiate true ON aplasia from severe ON hypoplasia.[7]

The commonly associated ocular anomalies include microphthalmos, microcornea , ptosis , squint, iris hypoplasia, irido-fundal coloboma and persistent hyperplastic primary vitreous.[7]

Conclusion:

Unilateral ON aplasia is a rare condition with poor visual prognosis and rarely associated with CNS anomalies. Clinical and radiological features can help differentiate it from other developmental anomalies of the eye ball.

Declaration of patient consent:

The authors certify that they have obtained all appropriate patient consent forms . In the form the patients has/have given his/her /their consent for his/her /their images and other clinical information to be reported in the journal. The patients understood that their names and initials will not be published and due efforts will be made to conceal their identity , but anonymity cannot be guaranteed.

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Nil

Conflicts of interest

There are no conflicts of interest

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Steroid and Antibiotic Injection at the time of Cataract Surgery could prevent DME

This analysis evaluated the effect of intravitreal triamcinolone acetonide–moxifloxacin at the time of cataract surgery on diabetic macular edema (DME). Their retrospective chart review of 64 patients with preexisting diabetic retinopathy found that mean visual acuity at 4 to 12 weeks postoperatively ranged between 0.32 and 0.43 logMAR. Central macular thickness did not change significantly after surgery. They conclude that triamcinolone and moxifloxacin stabilize macular thickness after cataract surgery and can be considered for patients with DME. *Journal of Cataract & Refractive Surgery, September 2020*