Brief Communication

Congenital Cyst of the Optic Nerve: A Case Series

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ABSTRACT

Congenital cyst of the optic nerve is a rare congenital anomaly, scarcely reported. We report a large case series from a single institute over a period of 21 years, consisting of 3 cases with rare presentations. Retrospective study of 1100 orbital cases revealed three cases with congenital cyst of optic nerve reported here. In these patients, a cyst was replacing almost all of the intraorbital portion of the optic nerve, with their unique features. Management of the cases is also discussed.

Key Words: Congenital cyst, optic nerve, orbit.

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INTRODUCTION

Cystic lesions of the optic nerve are rare and have been associated with systemic diseases and tumor so rare idiopathic. We are reporting three cases with congenital cysts which were replacing most of the intraorbital part of the optic nerve. They had smaller corneas on the affected side and all three were female infants.

CASE SERIES

Case 1

An 8-month-old female infant presented with progressive proptosis of the left eye with marked inferotemporal dystopia since birth. On gross visual assessment of the left eye, the patient could not follow light. A cystic mass was palpable on superficial palpation in supero-medial orbit. The left cornea was 2mm smaller than the right eye, and the pupil was not reactive to light. On fundoscopy, total retinal detachment was seen, obscuring the disc view. There was history of eye surgery a few months back with no record available. A Computed tomography scan showed a well-defined large intra-conal orbital cyst arising from and having a connection with the posterior pole of the globe. Although the eye ball was compressed laterally the eye ball measurements were equal in both eyes. Supero-medial trans-conjunctival orbitotomy revealed a large cystic mass replacing the optic nerve. The cyst was first drained and then excised into a small optic nerve stump was visible at the posterior end of the excised cyst. About 10 cc clear fluid was drained, which resulted in softening of the eye ball and shrinkage of the cyst. The distal portion of the superior oblique muscle was adherent to the cyst wall. The globe defect at the posterior pole was repaired. Histopathology revealed glial tissue. No malignant cells were found on cytology.

Case 2

A 6-month-old female presented with left axial proptosis since birth. The child was not following light from her left eye. Ipsilateral cornea was comparatively smaller by 2mm (figure: 1a). Left pupil was non-reactive. Extraocular movements were restricted in all gazes. On fundoscopy, the disc was pale. CT scan showed a well-defined large intra-conal orbital cyst occupying whole of the left orbit and causing

enlargement of the bony orbit with thinning and curving of the lateral wall, but no erosion (figure: 1b). Both eye balls were of the same size. No abnormality was found in her right eye. Anterolateral trans-conjunctival orbitotomy revealed large cystic mass that replaced almost all of the intra-orbital part of the optic nerve (figure: 1c). It was first drained, which did not cause softening of the globe, and then excised in toto. Histopathology revealed glial tissue. No malignant cells were found on cytology.

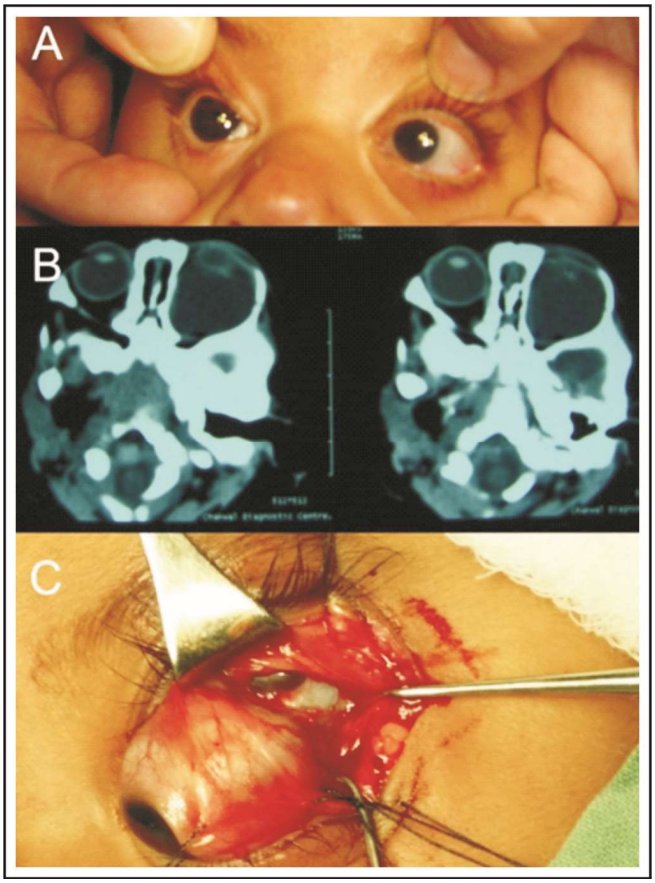

Figure 1:a: Pre-operative picture, **b**: CT scan showed a well-defined large intra orbital cyst occupying whole of orbit, **c**: Per operative picture with the cyst visible.

Case 3

Our third patient was a 5-month-old female with right inferonasal dystopia since birth. On visual assessment of her right eye, the child was not following light. Ipsilateral cornea was smaller by 2mm (figure: 2a). There was limitation of extraocular movements in all gazes. Pupil was non-reactive and fundus examination revealed a pale optic disc. Computed tomography revealed a large intra-conal cyst replacing the optic nerve (figure: 2b, c, d). Both the eye ball measurements were equal. It was excised through an anterior, trans-conjunctival lateral approach after initial drainage. It contained clear fluid. Acceptable cosmetic results were found with improved dystopia. Histopathology revealed glial tissue. No malignant cells were found on cytology.

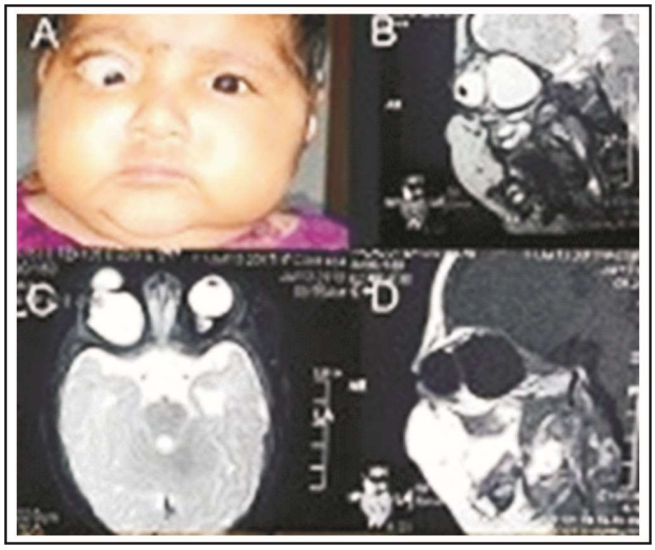

Figure 2: a: Preoperative picture, **b, c, d:** CT scan showing cystic orbital part of optic nerve.

DISCUSSION

Congenital abnormalities of the optic disc may include, size abnormalities, like optic disc aplasia, hypoplasia, megalopapilla and optic disc cupping in prematurity.[1] MEDLINE search revealed only few isolated cases of optic nerve cysts, reported so far. Kim et al,[2] reported a case of an optic nerve cyst involving distal end of the intra-orbital part of the optic nerve while in the current series, the cyst involved almost all of the intra-orbital part of the optic nerve. There was no associated corneal pathology in their case and the patient was male. Whereas all our cases were females and ipsilateral corneas were smaller by 2mm. Holland et al,[3] also reported an optic nerve cyst but unlike the current series, it was associated with a cystic eye ball. Shankar et al, reported bilateral retrobulbar cyst with associated optic disc colobomas.[4] Optic disc coloboma with an associated cyst in the optic nerve sheath was also reported by Wiggins et al.[5]

Optic nerve in previously reported cases was intact while the intraorbital part of the optic nerve is entirely

replaced by a cyst in our series. Optic canal part of the optic nerve was intact. The only explanation for cyst involving only the intraorbital part seems to be the loose space available for the cyst to expand within the orbit. The intracanalicular portion of the optic nerve was not biopsied by us or by other observers in the past. We do not know whether the axon migration is normal or there is a developmental abnormality beyond orbit.

At 6 weeks of embryological development, closure of fissure starts from the center extending anteriorly towards the optic cup and posteriorly towards the optic stalk.[6] During week 7, axons begin to grow in to the optics talk and development of the optic nerve is completed by the eighth week.[7] It is believed that during 6^{th} to 8^{th} week of development, instead of axon growth and cell migration in to the optics talk, it is replaced by fluid giving rise to an optic nerve cyst. The cause may be any developmental anomaly, genetic event or environmental factor, which is still a matter of debate.

CONCLUSION

Complete replacement of the intraorbital part of the optic nerve by congenital cyst has not been reported before to the best of our knowledge. It is important to rule out secondary causes of optic nerve cysts before labeling a cyst as benign. Our study shows female preponderance and association of ipsilateral smaller cornea in such cases.

Conflict of Interest: Authors declared no conflict of interest.

REFERENCES

1. **Denis D, Hugo J, Beylerian M, Ramtohul P, Aziz A, Matonti F, et al.** Anomalies congénitales de la papille [Congenital abnormalities of the optic disc]. J Fr Ophtalmol. 2019;**42(7):**778-789. French. Doi: 10.1016/j.jfo.2018.09.011.
2. **Kim KM, Kang SJ, Kim DS, Chi JG, Kim SM.** Congenital intraorbital optic nerve cyst. Case report. J Neurosurg. 1999;**91(2):**325-327. Doi: 10.3171/jns.1999.91.2.0325.
3. **Holland L, Haridas A, Phillips G, Sullivan T.** Congenital cystic eye with optic nerve. BMJ Case Rep. 2015;**2015:**bcr2015210717. Doi: 10.1136/bcr-2015-210717.
4. **Shankar LG, Khadia A, Gayam K, Priya S, Moutappa F.** An eye behind an eye: A rare association of bilateral optic disc coloboma with retrobulbar cyst. Indian J Ophthalmol. 2020;**68(8):**1656-1657. Doi: 10.4103/ijo.IJO_1957_19.
5. **Wiggins RE, von Noorden GK, Boniuk M.** Optic nerve coloboma with cyst: a case report and review. J Pediatr Ophthalmol Strabismus, 1991;**28(5):**274-7. Doi: 10.3928/0191-3913-19910901-10.
6. **Barishak YR.** Embryology of the eye and its adnexae. Dev Ophthalmol. 1992;**24:**1-142. PMID: 1628748.
7. **Tawfik HA, Dutton JJ.** Embryologic and Fetal Development of the Human Orbit. Ophthalmic Plast Reconstr Surg. 2018;**34(5):**405-421. Doi: 10.1097/IOP.0000000000001172.

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