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Case Report

Unilateral optic disc pit associated with orbital cyst in a child

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Abstract

Optic disc pit and disc coloboma are common congenital anomalies. Both have been known to be associated with an orbital cyst. We report a case of a 6 month-old child who presented with unilateral progressive proptosis. Imaging investigations revealed a well-defined, intraconal, orbital cyst located close to the posterior surface of the globe displacing the optic nerve laterally. The cyst was excised and histopathological examination showed the cyst to have a lining of glial tissue, a thin epithelial lining, over a layer of collagenous connective tissue matrix. We hypothesize that colobomatous disc abnormalities and optic disc pits are different points on the same disease spectrum and can be associated with orbital cysts. Furthermore, our case points to the possible role of imaging the optic nerve in all cases of optic disc anomalies.

Keywords: Optic disc pit, Coloboma, Orbital cyst, Proptosis, Tumour, Optic nerve

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Introduction

Congenital optic disc pit is a relatively uncommon entity. Rarely though, this condition may be associated with an accompanying orbital cyst. The embryological explanation for an optic disc pit is a developmental defect of the primary epithelial papilla and incomplete closure of the embryonic fissure.[1,2] Congenital optic disc pits are seen either alone or in combination with optic disc colobomas. This suggests that congenital optic disc pits and disc colobomas may be a part of the same pathological spectrum.[3] In this communication, we present a case of a child who had an optic disc pit along with an orbital cyst. The embryological process is reviewed here and we postulate the possible hypothesis for an underlying connection between the two conditions.

Case report

A six-month-old male child presented with outward protrusion of the left eye. The proptosis was noticed since birth, gradually increasing and painless (Fig. 1).

On examination, the right eye was normal; the left eye showed the presence of a grade II relative afferent pupillary defect. The proptosis was noted to be abaxial, non-pulsatile and had no associated bruit. The left globe was displaced inferomedially. Dilated fundus evaluation of the right eye was normal but the left eye had an optic disc pit. The media was clear, the retina was attached and normal foveal reflexes were noted. B-scan ultrasonography of the left orbit showed a large, fluid-filled cavity in the region of the optic nerve, which showed no definite connection to the globe. A computed tomography (CT) scan of the orbits showed the presence of well-defined isodense, intraconal, cystic lesion located in the superomedial orbit with the extraocular muscles being distinctly identified separately from the cyst (Fig. 2). Magnetic Resonance Imaging (MRI) showed a well-defined, intraconal, hyperintense in T2 weighted images (isointense to vitreous), cystic lesion in close relation to the posterior surface of the globe displacing the optic nerve laterally (Fig. 3). A diagnosis of an optic disc pit with an orbital cyst was made and the contents of the cyst were aspirated, which resulted in resolution of proptosis. However, this

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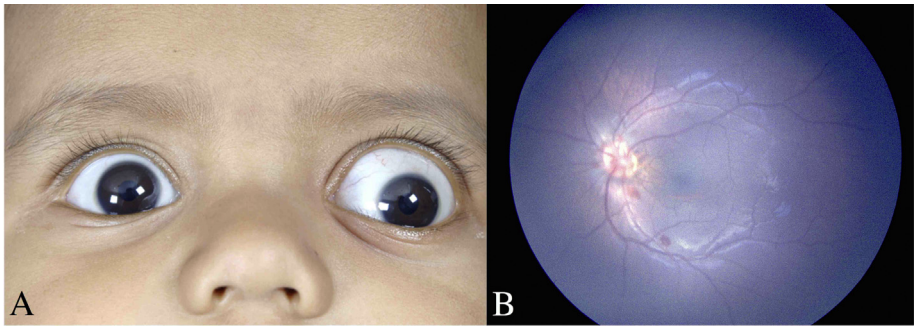

Figure 1. (A) An external photograph showing left proptosis and displacement of the globe infero-laterally; (B) Fundus photograph of the left eye with a small inferotemporal optic disc pit.

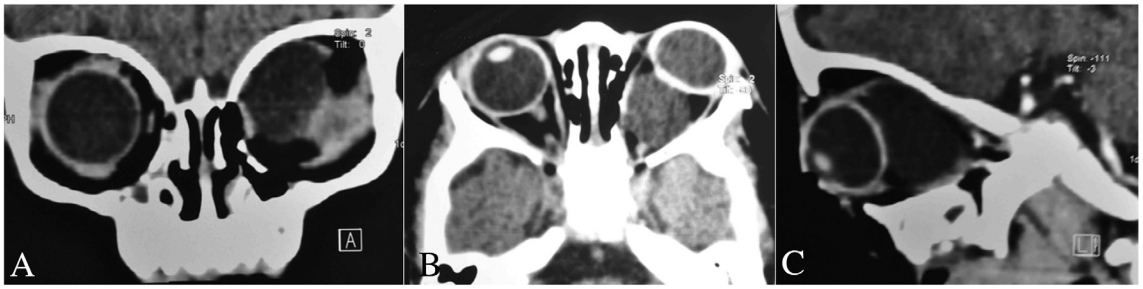

Figure 2. CT Scan images showing a well-defined isodense, intraconal, cystic lesion located in the superomedial orbit with the extraocular muscles being distinctly identified separately from the cyst. (A – coronal, B – axial and C – sagittal slices).

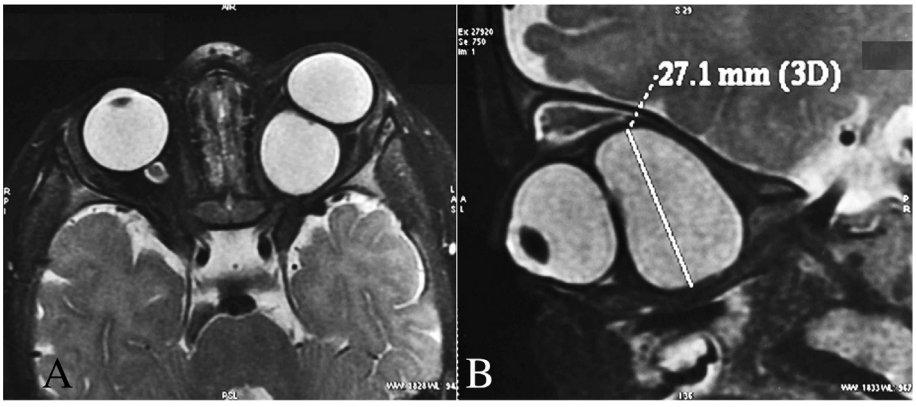

Figure 3. MRI images showing a well-defined, intraconal, hyperintense in T2 weighted images (isointense to vitreous), cystic lesion in close relation to the posterior surface of the globe displacing the optic nerve laterally. (A – parasagittal slice. B – coronal slice).

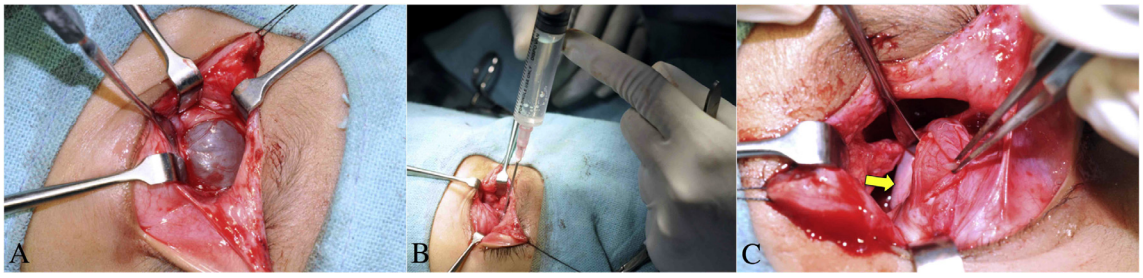

Figure 4. Intra-operative images of the surgical excision of the orbital cyst. (A) The orbital cyst in the superomedial orbit; (B) Aspiration of clear fluid from the orbital cyst; (C) Complete excision of the cyst wall (grasped within the forceps). Note the close proximity to the optic nerve (Yellow arrow).

resolution was only temporary as the swelling recurred within a week.

The child subsequently underwent an orbitotomy, which was performed through a vertical lid-split approach. Intraoperatively, a large clear fluid filled cyst was noted superomedially in the orbit displacing the globe infero-laterally (Fig. 4). The cyst could be dissected off all surrounding tissues, however the base was not dissected owing to its close relation to

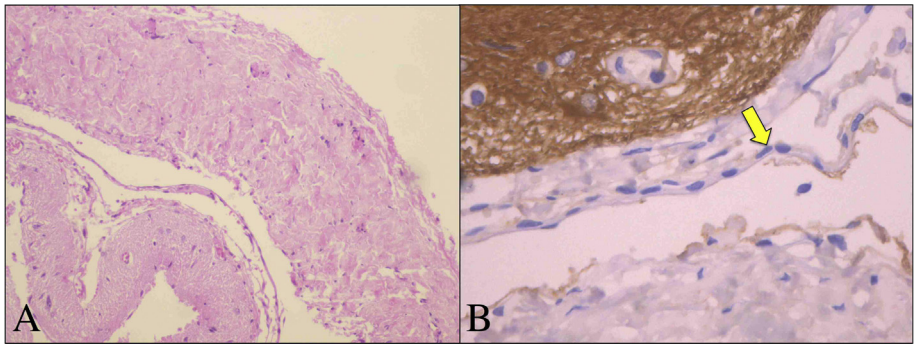

Figure 5. Histopathological examination of the excised specimen. (A) Histopathological examination of the specimen showed a thin epithelial lining, overlying a layer of collagenous connective tissue matrix. Over the epithelial cell layer, a layer of loosely arranged fibrillary astrocytic glial tissue is present. (Haematoxylin-Eosin stain, 10× magnification); (B) The glial which showed strong immunopositivity on staining with antibodies to glial fibrillary acidic protein (GFAP). Note the thin epithelial layer underneath (Yellow arrowhead). (Glial fibrillary acidic protein; 40× magnification).

the optic nerve. The cyst contents were then aspirated into a 10 mL syringe and the cyst wall collapsed; following which the entire cyst wall was excised in-toto. Histopathological examination of the specimen showed a thin epithelial lining, overlying a layer of collagenous connective tissue matrix (Fig. 5). However, over the epithelial cell layer, a layer of astrocytic glial tissue was noted which showed strong immunopositivity on staining with antibodies to glial fibrillary acidic protein (GFAP). There was no recurrence and no change was seen in the structural integrity of the globe, intraocular pressures or retinal morphology at follow up.

Discussion

An optic disc pit is said to be a herniation of dysplastic neuroectodermal tissue into a collagen-walled excavation, extending backward through a lamina cribrosa defect into the subarachnoid space.[2] Congenital optic disc pits are seen either alone or in combination with optic disc colobomas. This suggests that congenital optic disc pits and disc colobomas may be a part of the same pathological spectrum.[3] Imaging studies that have evaluated congenital optic disc pits with swept-source optical coherence tomography have reported the possibility of a communication of fluid between the optic disc pit and the sub-arachnoid space along the retrobulbar optic nerve.[3] Theodossiadis and colleagues have also hypothesized along the same lines that the simultaneous appearance of optic nerve cyst and optic disc pit is the mechanism of communication between the sub-arachnoid space with optic disc pit and from there into the subretinal space.[4] In our case, however, there was no subretinal fluid noted. The nature of the fluid within the cyst could have been either liquefied vitreous or cerebrospinal fluid. The aspirated material, in our case was sent for analysis, however, the biochemical parameters did not match those of CSF. We believe it is important that fluid aspirated in cases such as these should be sent for analysis to ascertain the origin of the fluid.

Co-existent orbital cysts with co-existent optic disc pits have been reported in the literature.[4,5] Shields have defined a colobomatous cyst to typically be a neuroectodermal lined mass that protrudes through a coloboma in the wall of a microphthalmic eye.[6] Dhir et al have earlier reported a case of 69 year old male with unilateral optic disc pit and orbital cyst in an eye with normal axial length.[7] Touitou et al. have also reported an orbital cyst associated with optic disc pit.[8] However, none of the reported orbital cysts have been histopathologically examined. The inner lining of neuroglial tissue, which was demonstrated histopathologically and confirmed with immunohistochemical staining in our case possibly, adds further weight to the theory that colobomatous disc abnormalities and optic disc pits are different points on the same disease continuum.

Furthermore, our case points to the possible role of imaging the optic nerve in all cases of optic disc anomalies.

Conflict of interest

The authors declared that there is no conflict of interest.

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