Phakomatous choristoma of the orbit: a case report

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Abstract
We report the case of a 3-month-old infant with a rare phakomatous choristoma of the orbit. This lesion is believed to be a congenital neoplasm of lenticular anlage. The clinical, radiological, and histopathological findings are presented.

(Zimmerman first described the clinical and histopathological features of phakomatous choristoma in three infants. Since then there have been six further case reports in this rare and unusual tumour. In all these cases the lesion was described as a lower eyelid mass with occasional extension posteriorly into the orbit. In our case the lesion was located entirely posterior to the orbit septum and was confined to the anterior orbit. We present the characteristic clinical, radiological, and histopathological findings, and suggest that this lesion be added to the differential diagnosis of an anterior orbital mass in an infant.

Case report
A 9-week-old female infant was seen by an ophthalmologist with a history of a lump visible in the inferomedial aspect of the right lower eyelid (Fig 1). The lesion had been noted by the mother shortly after birth. This was associated with some swelling of the lower eyelid and epiphora. The lesion was initially thought to be a mucocele of the lacrimal sac and was treated conservatively. The lesion remained unchanged and the patient was referred to the oculoplastic clinic having undergone a computed tomography (CT) scan of the orbits.

The lesion had remained unchanged in size since it was first noted. On examination a 1 cm x 1 cm, discrete, firm, mass was palpable through the skin of the lower eyelid medially. It was situated 0.5 mm lateral to the punctum. The lesion was non-mobile, non-fluctuant, not attached to overlying skin, and there was no overlying skin discolouration. There were no abnormal pulsations and no increase in size of the lesion with a positive Valsalva manoeuvre. Pressure over the lesion was not associated with reflux of mucus or pus from the puncta. There was a mild degree of epiphora on the right side and a fluorescein dye disappearance test was prolonged. A cycloplegic refraction was per-

Figure 1 Photograph of the infant demonstrating a visible mass lesion in the inferomedial aspect of the right lower eyelid.

Figure 2 Axial CT scan demonstrating a discrete well-circumscribed homogeneous anteromedial lesion in the right orbit inferiorly.

Figure 3a Operative photograph demonstrating the anterior aspect of the lesion. The orbital septum has been opened to the lesion. Intact orbital septum is seen laterally in the expose wound.

Figure 3b Photograph of the lesion excised intact.
formed the result of which was as follows: OD + 0.5/0 + 2.00 x 120, OS + 0.50. The infant was the product of a full-term pregnancy. Delivery was normal and the baby was in good health. The remainder of the ocular examination, as well as the general physical examination, was normal.

The CT scan showed a discrete, well circumscribed, homogeneous, non-cystic anterior orbital lesion not involving the lacrimal drainage apparatus (Fig 2). There were no other orbital anomalies. Standardised echography showed the lesion to be well circumscribed and highly reflective.

A preoperative clinical diagnosis of a neoplastic lesion was made. The patient underwent an excision biopsy of the lesion via a subciliary skin incision. The lesion was found to be posterior to the orbital septum with which it was partially fused. The lesion was solid, white, and nodular with no discrete capsule (Fig 3). It was dissected from the surrounding tissues. It was adjacent to the inferior oblique muscle and did not directly involve the lacrimal drainage apparatus.

The lesion measured 10 x 8 x 4 mm. Histological examination showed the tumour to consist of dense collagenous stroma in which islands of benign cuboidal epithelium were embedded (Fig 4). These epithelial islands surrounded central eosinophilic globules, consistent with lens cortex. Some of these globules were swollen and contained benign nuclei resembling ‘bladder cells’ of human cataractous lenses. The epithelial islands were surrounded by thickened basement membrane material. Small foci of dystrophic calcification were also identified.

Discussion
The clinical findings of this case are similar to those of previously reported cases except that the lesion in this case was confined to the orbit. The phakomatous choristoma always appears to be confined to the antero-inferior aspect of the eyelid/orbit medially. This location is the most consistent feature of the lesion. The close proximity of the lesion to the junction of the lacrimal sac and nasolacrimal duct was probably responsible for the infant’s epiphora. This resolved spontaneously postoperatively.

It has been postulated that this lesion is due to some of the surface ectodermal cells, which are induced to form the lens plate and lens vesicle in the embryo, remaining external to the optic vesicle as the embryonic fissure closes. These cells would then multiply and form the phakomatous choristoma after undergoing rudimentary differentiation. Its constant inferonasal position might also be explained by the ventral nasal location of the embryonic choroidal fissure of the optic vesicle.

This is a rare and interesting tumour which has never been correctly diagnosed preoperatively in any of the previously reported cases. This case is no exception in this regard. The clinical, radiological, and echographic characteristics of this tumour together with an awareness of its existence should permit its ready differentiation from most other orbital lesions occurring in childhood in this anatomical location.

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