CASE REPORTS

Epibulbar choristoma and microphthalmia: a report of two cases

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Abstract

The simultaneous ipsilateral presence of an epibulbar choristoma and microphthalmia has rarely been reported. We present two such cases, one of which is associated with bone formation, and we consider a possible pathogenetic mechanism.

The simultaneous occurrence of epibulbar choristomas and microphthalmia has been

reported¹⁻³ but is extremely rare. It is not known whether the association is causal or incidental, though intraocular involvement by epibulbar choristomas apparently without associated microphthalmia has been reported.^{4,5}

Case reports**CASE 1**

An otherwise healthy 16-year-old male presented to the Kikuyu Eye Department in Kenya with a large growth protruding through the left interpalpebral fissure (Fig 1). The lesion had been present from birth. On examination the orbit was clinically anophthalmic. The mass was excised by subtotal exenteration for cosmetic reasons.

On gross examination the lesion consisted of two lobes of tissue connected by a short pedicle. The total anteroposterior length was 32 mm. The posterior lobe was 20 mm and the anterior lobe

Figure 1 Case 1. Clinical photograph of orbital lesion protruding through the left interpalpebral fissure. Numerous hairs are present on the exposed portion of the mass.

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Figure 2 Case 1. A nodule of fibroadipose tissue covered by skin is attached by means of a short pedicle to orbital connective tissue, on the inferior aspect of which is a disorganised, microphthalmic globe. The superior part of the 'scleral' condensation surrounding the globe is deficient and corresponds to a colobomatous defect in the developing choroid and retinal pigment epithelium. (Verhoeff $\times 3.5$.)

Figure 3 Case 1. Section of the 'dermolipoma' showing adipose tissue covered by skin replete with adnexal structures. There is a mild lymphocytic infiltration of the reticular dermis. (H and E, $\times 28$.)

Figure 4 Case 1. The tissue occupying the cavity of the globe includes folds of differentiated neuroretina. (H and E, $\times 125$.)

Figure 5 Case 1. A limited anterior chamber is lined on its outer 'corneal' aspect by a Descemet's membrane which extends in part over the primitive uveal component of the anterior segment. (H and E, $\times 125$.)

Figure 6 Case 2. A large rounded mass of adipose tissue is covered by skin with sparse adnexal elements and includes a nodule of bone at the base. A microphthalmic globe is also present, the posterior segment of which is occupied by folds of detached retina. (H and E, $\times 2.8$.)

15 mm in transverse diameter. Hair-bearing, pigmented, keratinised skin covered the anterior lobe and pedicle, while the posterior portion of the lesion appeared to be a mass of orbital fat

without epithelium. Sagittal sectioning revealed a 5 mm diameter pigmented remnant of presumed ocular tissue located within the inferior part of the posterior lobe.

Histologically the anterior aspect of the specimen consisted of a pigmented, keratinising squamous epithelium covering adipose tissue subdivided by connective tissue septae (Fig 2). The epithelium displayed maturation typical of epidermis (Fig 3). Numerous pilosebaceous apparatuses, sweat glands, and rete pegs were embedded in a dense layer of subepithelial fibrous connective tissue. Within the dermis of the inferior portion of the anterior lobe was a mild chronic inflammatory infiltrate, possibly caused by prolonged rubbing of this part of the mass on the adjacent malar eminence. The adipose tissue in the anterior lobe was in continuity with similar tissue in the posterior lobe through the central pedicle. In the inferior portion of the posterior lobe lay a microphthalmic globe, the central cavity of which was filled by folds of partially gliosed neural tissue displaying frequent evidence of retinal differentiation (Fig 4). This was surrounded by a densely pigmented and richly vascularised tissue consistent with a disorganised choroid and retinal pigment epithelium, which was in turn surrounded by a coarsely organised layer of collagen. In the anterior portion of the fibrous tissue layer a Descemet's membrane was present, separated from an incompletely formed anterior chamber by a single layer of attenuated cells (Fig 5). Some sections revealed an extensive defect in the superior aspect of the 'scleral' condensation, this defect being filled with adipose tissue continuous with that present in the orbit. A large-calibre blood vessel traversed the tissue within this coloboma. Lens material was not evident in sections taken from multiple levels. Foci of dystrophic calcification and lamellar bone formation were present in the degenerate retina and primitive uvea. A well defined but gliosed optic nerve could be seen leaving the posterior part of the globe.

CASE 2

A 1-year-old female from Ghana presented with a large mass protruding from an interpalpebral fissure. This mass was excised by subtotal exenteration.

On gross examination the specimen consisted of an elongated sphere measuring 38 mm in anteroposterior length by 25 mm transversely. The anterior two-thirds of the mass was covered by pigmented, hair-bearing skin. The posterior one-third of the lesion was composed of adipose tissue. An 8 mm diameter apparent globe remnant was present in the posterior part of the specimen inferiorly. Adjacent to the globe was an 8 $\times$ 5 mm ellipse of calcified material.

Microscopic examination showed that the lesion was covered anteriorly by mature, stratified, squamous, keratinising epithelium with a deeply pigmented basal layer. Near a reflection of tissue posteriorly the epithelium became non-keratinising, but no goblet cells were present in this part of the epithelium. Deep to the epithelium was a well-defined dermal layer

Figure 7 Case 2. Part of the choristomatous osseous tissue in the form of a shell surrounding haemopoietic marrow with sparse interlacing trabeculae. (H and E, $\times 30$.)

Figure 8 Case 2. A reasonably well formed anterior chamber is backed by iridociliary pigment epithelium which merges with a mass of detached and folded retinoglia. Subretinal exudate is also seen. (H and E, $\times 15$.)

Figure 9 Case 2. A section cut at a different level from the previous samples shows a remnant of cataractous and partially organised lens tissue. It is separated from the anterior chamber by a narrow rim of pigmented uveal tissue. (H and E, $\times 30$.)

composed of dense connective tissue, containing fine hair follicles and numerous sweat glands. The bulk of the mass consisted of mature adipose

tissue continuous posteriorly with the intraorbital fat (Fig 6). Connective tissue septa coursed through the intraorbital and intralesional adipose tissue. The calcified lesion in the posterior aspect of the specimen was found to be lamellar bone with a central cavity containing adipose tissue and bone marrow (Fig 7). Fibrous connective tissue covered the bone on three sides and the remaining surface was in direct contact with the sclera of the globe remnant at the insertion site of an extraocular muscle.

The globe remnant had a fibrous tissue coat, the anterior fibres being more regularly orientated than those situated posteriorly. A Descemet's membrane and single layer of cuboidal cells lined the posterior aspect of this immature cornea, posterior to which was an anterior chamber filled with exudate (Fig 8). The angle and iris were immature, though some evidence of trabecular tissue was present in the angle, and dilator pupillae fibres were seen in the iris. The lens was cataractous (Fig 9). The central cavity of the globe was filled with disorganised neuroglial tissue attempting to form layers. Subretinal exudate separated this from a heavily pigmented retinal pigment epithelial layer covering a well developed choroid. Foci of intraocular calcification and early dystrophic lamellar bone formation were present anterior to the lens and within the disorganised retinal tissue. At least one extraocular muscle was inserted into the globe. An apparently normal optic nerve could be seen leaving the globe posteriorly.

Discussion

Each of the cases we have presented had epibulbar masses comparable with lesions that others have called dermolipomas. However, adnexal structures, especially pilosebaceous apparatuses, have been said to be unusual in dermolipomas,^{6,7} and it may be that the lesions in question are simply epibulbar extensions of orbital fat covered by skin. Sinha and Mishra⁸ have reviewed the postulated aetiologies of dermoid choristomas. These hypotheses direct attention towards a superficial cause and do not adequately explain the extensive orbital involvement of many 'dermolipomas', including the cases we present. Conversely, the presence of mature cancellous bone in the second case, similar to a case presented by Ferry and Hein,⁹ probably argues for the genuinely choristomatous character of the masses.

The degree of development reached by the ocular tissue in the two cases differed. In the first case the presence of a Descemet's membrane, retinal differentiation sufficient to include ganglion cells, and the degree of development of the choroid and optic nerve suggest maturation to at least the 110 mm (16 weeks) stage. The failure to detect any lens tissue implies a resorptive process as it is unlikely that development of the globe could have proceeded to the level observed in its absence. The incomplete formation of the cornea and anterior chamber may also have been a consequence of an abbreviated lens presence. It is possible that maturation of the globe as a whole was arrested shortly

before the end of the fifth month, as by that time the sclera should have been approaching a definitive stage, and some attempt at a retinal circulation might have been expected. The second case showed more advanced development, probably well into the seventh month, as judged by the presence of a well defined dilator pupillae muscle and anterior chamber structures.

It is tempting to try to link the incomplete ocular development with the presence of epibulbar lesions in these cases. It could be reasoned in the first case that a putative excess of precursor adipose tissue early in embryogenesis had prevented adequate contact between the optic vesicle and the lens plate such that the lens failed to develop properly, leading to its subsequent resorption. Invagination of similar tissue into the primary optic fissure might account for the coloboma, while continued proliferation of adipose tissue could have produced the epibulbar mass.

In respect of the second case development was much more advanced, and it is difficult to ascribe the involutional changes to interference by adipose tissue lying outside the globe, though excessive proliferation of the orbital adipose

tissue in the later stages of intrauterine life could account for the epibulbar lesion. Whether this admittedly speculative attempt to explain the coincidence of microphthalmia and epibulbar choristoma has any validity is unknown.

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