Isolated eyelid coloboma in association with complex choristoma in a newborn: A case report

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Abstract:

Ocular choristomas are rare lesions that have been reported at the conjunctiva, sclera, orbit, or intraocularly with significant potential for visual disturbance. The complex type of choristomas shows a mixture of different cartilaginous, glandular, and muscular tissue in addition to fat. We present a patient with an associated eyelid coloboma and complex choristoma. A 12-day-old baby boy was referred to our hospital with an upper medial eyelid coloboma affecting almost two-thirds of the eyelid length with an additional sub-brow mass since birth. The baby also had secondary findings to the eyelid coloboma defect: temporal conjunctival symblepharon, vascularized cornea, and inferior pannus. The patient underwent an upper eyelid reconstruction with excisional biopsy of the sub-brow mass, which was diagnosed as a complex choristoma. This is the first case of an eyelid coloboma-associated with complex choristoma without any other systemic associations.

Keywords:

Complex choristoma, dermoid cyst, eyelid coloboma, orbit, sub-brow mass

Introduction

Choristomas are benign congenital tumors with stationary or minimal growth potential, which have a normal tissue found in an abnormal location.[1] The majority of the ocular choristomas are reported at the conjunctiva, sclera, orbit, or intraocular such as the iris, ciliary bodies, retina, choroid, and optic nerve. However, it is still considered a rare tumor, and the cause of the disease is not well known yet. Although it is rarely clinically significant due to its benign nature, choristoma can be cosmetically worrisome and medically concerning when it is developed inside the eye because it can cause severe visual and functional issues. The most common of type of choristomas are dermoids and lipodermoids.[2] In addition, some studies reported cases of complex choristomas described as a mixture of different tissues such as cartilage, lacrimal gland, muscles, and adipose tissues.[1] These tumors appear as a mass and can only be identified using histopathological methods.

Incomplete closure of the fetal fissure during organogenesis can result in congenital ocular colobomas. Many eye structures can be involved in such conditions, including eyelids, iris, lens, ciliary body, choroid, retina, as well as the optic nerve. Congenital eyelid coloboma is an eyelid defect ranging from a small marginal hole to a full-thickness defect in around one out of every 10,000 births. It can involve one or all four lids, unilaterally or bilaterally. In most cases, congenital eyelid coloboma involves the upper eyelid, more specifically between the medial and middle third of the upper eyelid.[3] In a few cases, complex choristoma was reported in association with different coloboma structures such as optic disc coloboma and chorioretinal coloboma.[4,5] Only one case was found to report an eyelid complex choristoma associated with eyelid coloboma.[6] In this case study, we report the clinical presentation and histopathology of a patient with eyelid coloboma and a mass found to be complex choristoma.

Case Report

A healthy full-term 12-day-old baby was referred to King Abdulaziz University Hospital

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in Riyadh with left upper eyelid coloboma and a sub-brow mass, which was thought to be a dermoid cyst. The patient had no family history of any ocular disorders. The left eye examination showed evidence of an upper medial eyelid coloboma, involving almost two-third of the eyelid length with a sub-brow mass on top of it. The examination showed temporal conjunctival symblepharon, vascularized cornea with pannus inferiorly [Figure 1a and b]. The remaining ocular examination of both eyes was normal. The patient was referred to a general pediatrician to rule out associated syndromes. Simple mass excisional biopsy with upper eyelid reconstruction was performed. Release of conjunctival adhesions with symblepharon ring placement to prevent further formation of new adhesions was also done. The mass had no attachments to the underlying structures. The patient underwent magnetic resonance imaging (MRI) of the brain and orbit with intravenous contrast; the MRI showed a well-defined 1 cm × 0.6 cm left upper eyelid lesion without intraorbital or intracranial extension, likely representing a small dermoid/epidermoid due to the presence of proteinaceous content or cholesterol without significant enhancement postcontrast administration. The orbital cavities were unremarkable, and there was no structural brain abnormality [Figure 1c].

Histopathology of the mass showed a complex choristoma containing the characteristic dense wavy collagen fibers, fat, areas of smooth muscle fibers, neural tissues, and immature cartilage [Figure 2a-c].

The patient was otherwise healthy with no syndromic features or other systemic problems. The patient had smooth postoperative course with cosmetically and functionally acceptable eyelid appearance following reconstruction.

Discussion

Periocular complex choristomas occur mostly as isolated lesions. It can be correlated as well to several types of syndromes including Goldenhar syndrome and linear sebaceous nevus syndrome.[7] The majority of previously reported complex choristomas arise from the conjunctiva with predilection to arise from an inferior-temporal location in 66.6% of cases. Choristomas are the most common conjunctival tumors in the epibulbar area and can affect the cornea, limbus, and rare locations, such as the lateral canthus. Isolated eyelid complex choristoma containing ectopic cilia and lacrimal gland tissue has been also reported.[1] Eyelid complex choristomas may accompany other ocular lesions including an epibulbar complex choristoma,[8] choroidal osteoma, and an epidermal nevus syndrome.[6] In our case, the patient had no systemic association. However, eyelid coloboma has been described in a retrospective case series of five patients with epibulbar rather than an anterior orbital complex choristoma.[7]

An anterior orbital complex choristoma near the medial canthus has been reported masquerading as dacryocystocele.[9] However, on computed tomography (CT) and MRI, the appearance was rather biphasic with variable signal intensity because of the heterogenous components of the choristoma, while dermoid cysts would show "fat" density due to the internal composition of the contents, which will be suppressed with fat suppression sequence confirming the diagnosis.[9,10] In our case, the MRI was suggestive of an anterior orbital dermoid because of the well-defined borders. It has been reported that intraorbital dermoids would mostly

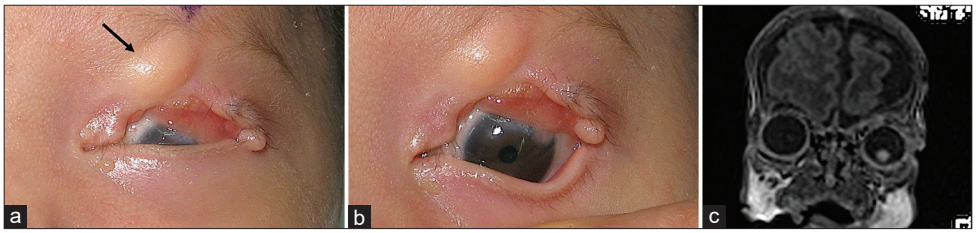

Figure 1: (a and b) An upper eyelid defect involving more than two thirds of the eyelid in conjunction with a supermedial firm sub-brow mass (black arrow) and a lateral cyst with a temporal conjunctival symblepharon. (c) The corresponding coronal T2-weighted magnetic resonance image of the eyelid coloboma region (without contrast) showing hypointense lesion in the superior anterior orbit

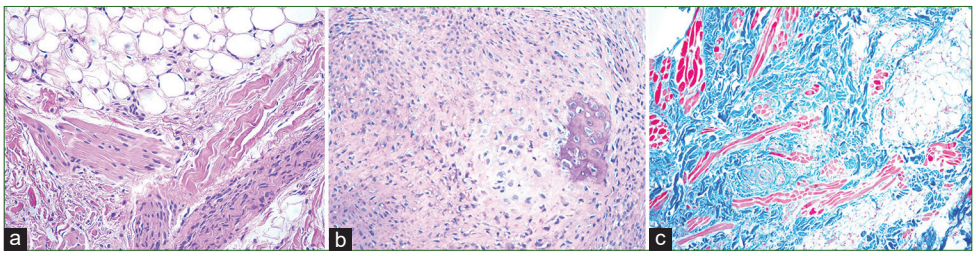

Figure 2: The histopathological appearance of the excised mass showing the typical dense collagen fibers, fat, smooth muscle, and neural tissue (a, H and E stain, ×200) and the cartilage component (b, Periodic acid–Schiff, ×200). The Masson trichrome stain highlighting some muscle fibers (c, ×100)

show this well-defined margin on CT images, but borders can be less frequently ill-defined as well.[10] The fact that our patient did not have any syndromic features has lowered our clinical suspicion of the presence of a choristoma rather than a dermoid and the histopathological diagnosis was actually unexpected. Spano *et al*. reported the association of upper eyelid coloboma with a choristoma (lower lid dermolipoma) in an infant with craniofacial anomaly (Tessier no 0–1 craniofacial cleft) and described a specific surgical technique for management.[11] Smith *et al*. have nicely conducted an observational study of 55 cases of eyelid coloboma, in which one-third only were isolated, while the rest were associated with either systemic (craniofacial) abnormality and/or ocular anomalies.[12] However, all the cases of associated choristoma in their series were limbal dermoids and dermolipomas and not an eyelid complex choristoma.[12] Although complex choristoma is mainly considered as a benign subtle disease managed by observation in small asymptomatic lesions, surgical excision can be done for cosmesis and averting amblyopia. Progression in size has been reported as the patient reaches puberty in approximately 40%.[7,13] As colobomas carry the risk of corneal involvement and amblyopia, surgical timing and techniques are considered crucial to prevent such complications. Given that our patient had a large eyelid defect with early superior corneal involvement and symblepharon, early surgical correction and postoperative follow-ups were pivotal to prevent further complications.

In conclusion, we report the first case of isolated combined eyelid coloboma and adjacent complex choristoma in the absence of any systemic associations. These cases should be genetically investigated and followed up for any emerging systemic abnormalities appearing later in childhood.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand 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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Conflicts of interest

There are no conflicts of interest.

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