Isolated accessory limb of lower eyelid with multiple dermal appendages

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ABSTRACT

We report the first case in the English literature of an isolated occurrence of accessory limb with multiple dermal appendages in a ten-month-old boy. This condition presented at birth as a limb bud below the left eyelid, multiple dermal appendages in the adjacent part of the face below the left orbit and on the upper part of the face. No anomalies of the ocular structures or central nervous system were identified. Accessory limb with multiple dermal appendages, in the absence of a congenital cystic eye, is an extremely rare condition representing a benign aberration in the developing musculoskeletal system. We present the first of such a case and endeavour to explain the embryological basis behind it.

Keywords: accessory limb bud, congenital cystic eye, dermal appendages

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INTRODUCTION

The occurrence of accessory limb buds and dermal appendages has been reported twice in association with a congenital cystic eye.[1,2] The congenital cystic eyeball develops when the primary optic vesicle fails to invaginate, while accessory limb buds and multiple dermal appendages may be the result of sequestration of parts of the developing limb buds during migration from the ventrolateral body wall at the end of the fourth week of development. We present an extremely rare occurrence of accessory limb bud and multiple dermal tags in the absence of a congenital cystic eye, and discuss the relevant embryology and outcome of this anomaly.

CASE REPORT

A ten-month-old boy presented to the outpatient department of BP Koirala Institute of Health Sciences with a limb-like projection underneath the left eyelid and multiple skin tags on the face, reported to be present since birth. A thorough history did not uncover any known teratogenic factors, nor were there any genetic disorders reported in the family. Examination revealed a 5 cm × 3 cm projection below the left lower eyelid and multiple dermal appendages on the face (Fig.1). Palpation of the lower eyelid mass was firm, non-tender, and showed limb-like contractions during crying. Five small finger-like and nipple-like projections were also noticed on the surface of the mass (Fig. 2). The left eye was normal. In addition, the patient also had multiple dermal appendages on the face (Fig. 1). The appendages measured approximately 5 mm in size, were non-tender and were covered by normal skin. No other gross anomaly was seen elsewhere on the body.

Radiographs of the skull revealed the presence of a calcified area in one of the nipples on the mass with no gross abnormalities in the intracranial structures (Fig. 3).

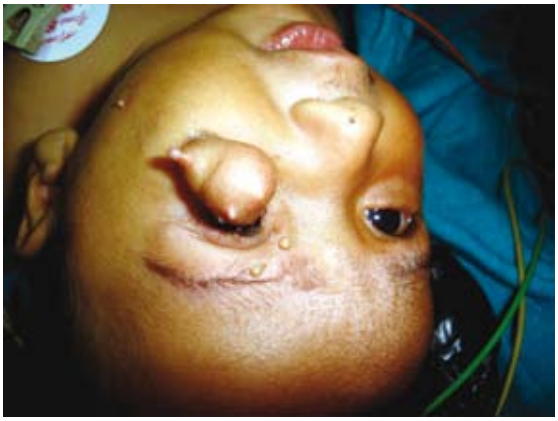

Fig. 1 Clinical photograph of the patient shows a pedunculated mass below the left lower eyelid and multiple dermal appendages.

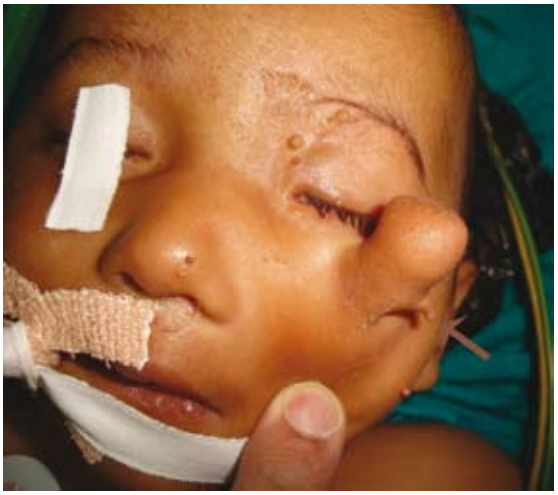

Fig. 2 Clinical photograph shows small nipple-like projections arising from the mass.

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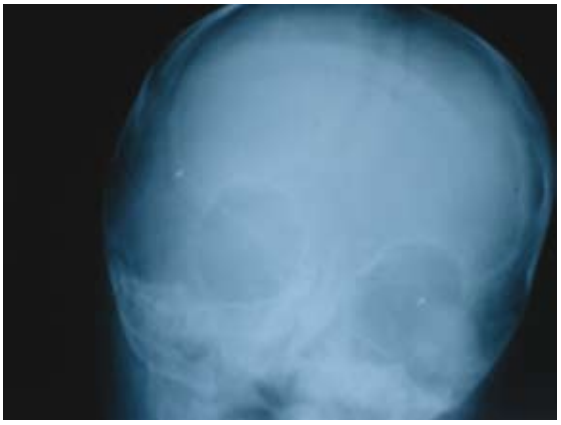

Fig. 3 Radiograph of the skull shows no obvious intracranial anomaly.

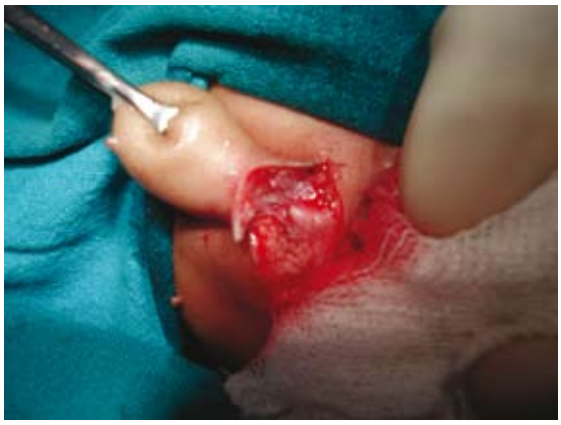

Fig. 4 Operative photograph of the accessory limb shows a bony spur.

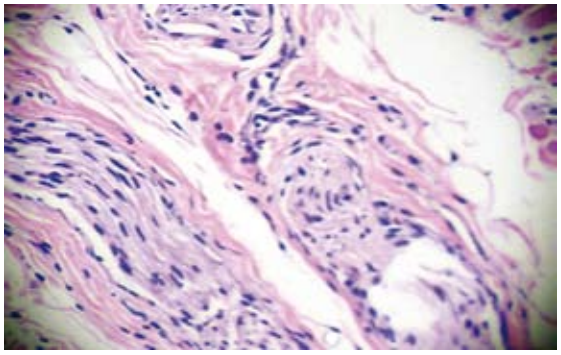

Fig. 5 Photomicrograph of the excised accessory limb shows prominent muscle tissue and fat (Haematoxylin & eosin, x 100).

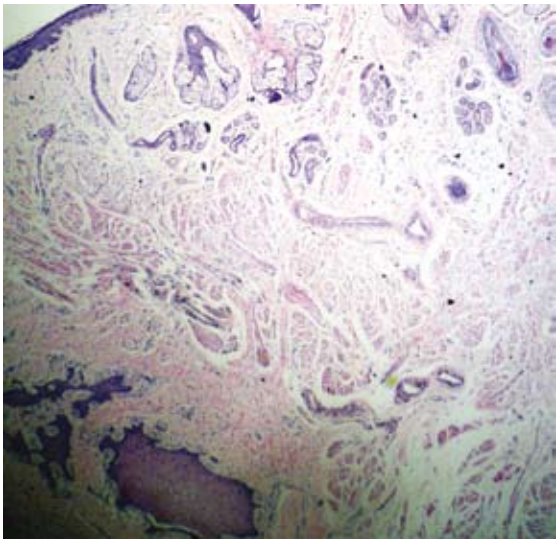

Fig. 6 Photomicrograph of the excised accessory limb shows areas of muscle, fat, cartilage and bone (Haematoxylin & eosin, x 50).

Other laboratory investigations were within normal limits. Based on the above findings, a diagnosis of accessory limb with multiple dermal appendages was made. The patient underwent surgery after requisite preoperative tests. During surgery, the accessory limb was found to be connected to the inferior orbital margin by a bony spur (Fig. 4). Excision of the accessory limb was done; care was taken to preserve the eyelids. The dermal appendages were not removed. Histological examination of the resected limb bud disclosed presence of striated muscle, fat, areolar tissue and skin (Figs. 5 & 6).

DISCUSSION

Congenital cystic eyes with accessory limb, dermal appendages or skin tags are extremely rare entities, with only four known cases reported in the literature.[1-4] However, the presence of such an accessory limb with multiple dermal appendages, in the absence of any ocular anomaly, to the best of our knowledge, has not been reported to date. Although Rice et al had reported a case of congenital cystic eye with the presence of accessory limb in a similar location,[2] and Dollfus et al had reported a case of congenital cystic eye associated with skin tags present near the inner canthus and at the root of nose,[3] there have been no known reports of an isolated accessory limb in the absence of a congenital cystic eye. The exact aetiology of this malformation is not known, although a few theories have been postulated.

At the end of the fourth week of development, limb buds become visible as outpocketings from the ventrolateral body wall. Initially, they consist of a mesenchymal core derived from the somatic layer of the lateral plate mesoderm.[5] Sequestration of these developing limb buds during lateral migration may lead to the formation of accessory limbs or dermal appendages in various parts of the body, or as seen in our case, even on the face. The other theory proposes that the limb bud formation is located in specific sites along the body wall of the developing embryo by a combination of fibroblast growth factor (FGF) and wingless/int-1 (Wnt) signalling.[6] Before limb bud initiation, FGF10 expression in the mesenchyme is localised precisely by the expression of Wnt3 produced from the surface ectoderm. The initiation of limb bud formation thus is accomplished by coordinate regulation

by the Wnt and FGF pathways. Wnt3 expression at ectopic sites in the body, or lack of coordination between the Wnt and FGF pathways, may therefore result in accessory limb bud formation in unusual sites, like in our case, the face.

The presence of a congenital cystic eye carries a relatively poor prognosis as it is often associated with more severe central nervous system (CNS) abnormalities, such as agenesis of the corpus callosum, basal encephalocoele and midbrain deformities. Other associated deformities include facial clefting, saddle nose, nostril malformations, choanal atresia, sphenoid bone malformations, microphallus with hydrocoele, hypoconvex fingernails on short stubby fingers and bifid thumb.[7-10] However, in this case, the isolated occurrence of the accessory limb, in the absence of other ocular and CNS anomalies, should carry a better prognosis. In a previously-published report, the predilection of the congenital cystic eyeball and its associated anomalies for the left side was highlighted.[11] Why only the left side of the face is affected is yet to be explained.

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