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CASE REPORT

Aplasia Cutis Congenita

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KEYWORDS

Aplasia Cutis Congenita
Congenital Absence of Skin
Congenital Scars
Cutis Aplasia
Scalp Defect

ABSTRACT

Aplasia cutis congenita is a rare congenital disorder characterised by localised absence of skin at birth, most commonly affecting the scalp. It usually presents as a solitary lesion, but can be multiple or associated with several malformation syndromes. Here, we report a case of a neonate with a well-demarcated scalp defect, highlighting its clinical features and management.

INTRODUCTION

Aplasia cutis congenita is a congenital anomaly characterised by the localised or widespread absence of skin. The condition predominantly affects the scalp (in 80–90% of cases) but can also involve the trunk and extremities. The aetiology could be multifactorial, including genetic, teratogenic and mechanical factors. Aplasia cutis congenita is classified into nine subtypes based on specific syndromes, such as Adams-Oliver syndrome and anomalies associated with fetus papyraceus, as well as placental infarction. Diagnosis is clinical, and treatment depends on the lesion size, with conservative management preferred for minor defects and surgical intervention for larger or complicated cases.

CASE REPORT

A 20 days old female neonate (delivered via normal vaginal delivery) presented to our OPD with a discrete ovoid defect covered by a membrane on the scalp since birth. Antenatal history in the mother was uneventful and suggested no maternal infections, trauma or drug exposures. No similar complaints were reported in the family. At birth, weight, vitals and routine investigations were found to be normal. On examination, a single well-defined, oval plaque measuring 2x3 cm with a thin atrophic surface and mild erythema was noted on the vertex of the scalp. No signs of infection, discharge or active inflammation were present. Neurological and systemic examinations were normal. After ruling out the possible differentials of forceps injury, use of scalp electrodes, birth trauma, nevus sebaceous, epidermolysis bullosa and Goltz syndrome, a final diagnosis of Aplasia cutis congenita was made based on typical clinical features. Parents were counselled that the condition has an excellent prognosis and can be managed effectively with conservative management (Figure 1).

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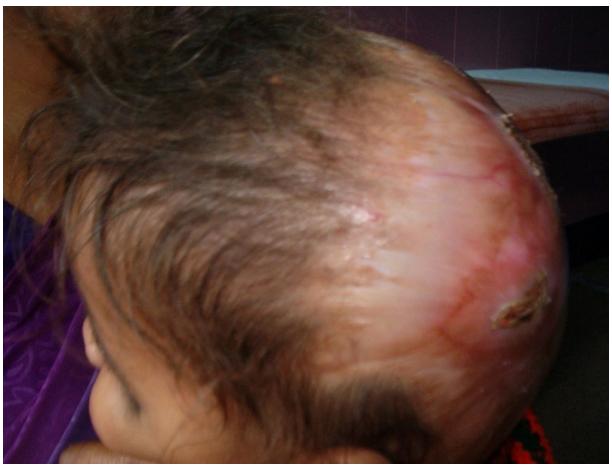

Figure 1. Aplasia cutis congenita.

DISCUSSION

Aplasia cutis congenita is typically an isolated defect, but it can be associated with certain genetic conditions [1]. The exact cause is unknown, but vascular disruptions, intrauterine trauma and genetic mutations have been implicated. Histopathological features reveal an atrophic, flattened epidermis, replacement of the dermis by loose connective tissue, and an absence of adnexal structures. Dermoscopy shows telangiectatic vessels and starburst-like hair follicles [2]. Scalp aplasia cutis congenita is divided into membranous and non-membranous forms. Membranous aplasia cutis typically presents as a punched-out oval or round defect covered by a thin, translucent, glistening epithelial membrane. Lesions of membranous aplasia cutis may have a "hair collar sign", which refers to a ring of long, dark terminal hair encircling the lesion. This is a marker of cranial dysraphism, and its presence mandates careful examination of the infant. Small lesions (<3 cm) are managed conservatively with dressing, topical antibiotics and emollients. Whereas large lesions (>3 cm or with exposed dura) may need surgical intervention to prevent complications such as infection or haemorrhage. In complicated cases (if bone involvement is suspected), imaging (ultrasound or MRI) is recommended [3]. In the absence of nail changes and with no lesions over frictional areas, diagnoses such as Bart's syndrome and epidermolysis bullosa are ruled out in this child.

CONCLUSION

Aplasia cutis congenita is a physical finding at birth that may result from any intrauterine event that disrupts skin development [4]. Most commonly, it affects the scalp. It has no single underlying cause and appears to be the end result of several distinct pathologic processes, including a form fruste of a neural tube defect. In the most common form (membranous aplasia cutis), oval, sharply marginated atrophic macules are seen on the midline of the posterior scalp. It is always hairless, and when it heals, it usually leads to atrophic scars. Aplasia cutis congenita is treated conservatively with local wound care. Surgical revision of the scar can be performed electively later in childhood or adolescence to improve cosmesis [5,6].

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