DOI: 10.7860/IJNMR/2017/25196.2216

Case Report

Neonatology Section

A Rare Case of Aplasia Cutis Congenita

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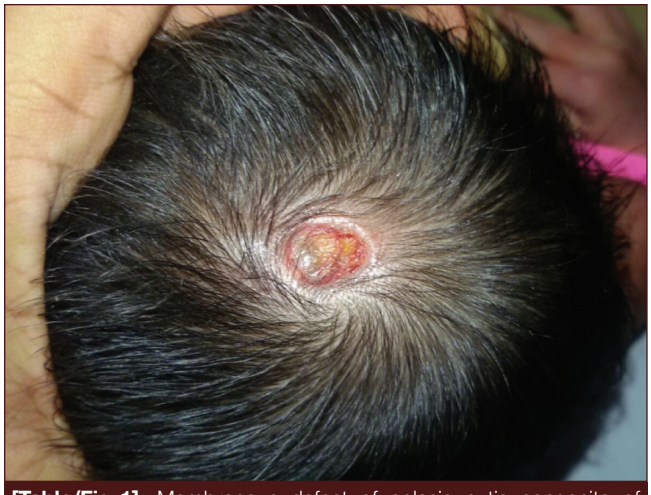
ABSTRACT

Aplasia cutis congenita is a heterogenous group of disorders characterized by the absence of a portion of skin either localized or widespread at birth. Most commonly seen on the scalp (84%), it can affect any part of the body, including the trunk and limbs. We report a case of a baby born with aplasia cutis congenital managed conservatively.

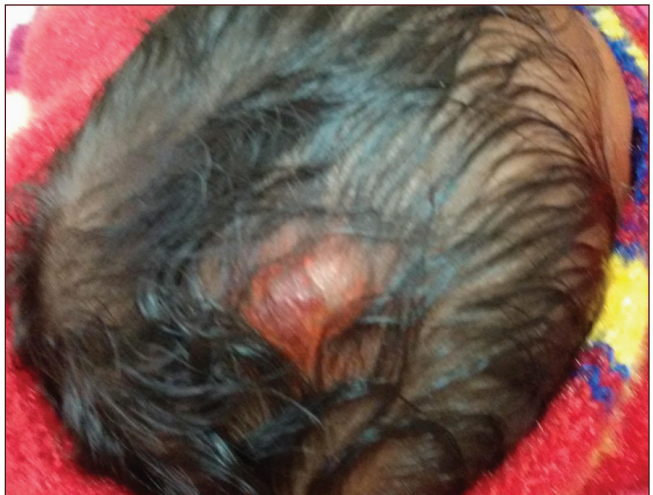
Keywords: Alopecia, Congenital malformations, Scalp

CASE REPORT

A 30-year-old booked G3P1L1A1 case was admitted 38 weeks for cesarean section in view of transverse lie and central placenta previa. Antenatal period was uneventful and there was no history of intake of teratogens or other drugs, history of consanguinity or syndromes in the family was found. Her intra-operative and postoperative periods were uneventful. The baby cried at birth, had normal APGAR scores at 1 minute and 5 minutes. On head to toe examination of the baby, a membranous defect of approximately 1x1 cm was present on the right lateral aspect of sagittal suture which bulged during crying [Table/Fig-1]. No evidence of seizures was noted. No other lesions were noted. Physical and neurological examination of the baby was normal. Intracranial ultrasonography was normal. After pediatric and dermatology opinion, a diagnosis of non-syndromic aplasia cutis congenita was made with the differential diagnosis being cutis congenital, generalized xerosis, lamellar ichthyosis and birth trauma. Sepsis screen was negative. Injectable antibiotics were given for seven days since the baby had pustular lesions on the face, neck and inguinal region. Genetic screening was not done as the facility for the same were not available and the parents were unable to afford. Mother and baby both were discharged in a satisfactory condition after proper parental counseling and the infant is on regular follow-up till date [Table/Fig-2]. Written and informed consent was taken from the parents regarding the publication of the manuscript.



[Table/Fig-1]: Membranous defect of aplasia cutis congenita of scalp.



[Table/Fig-2]: Follow-up image of the scalp after 3 months.

DISCUSSION

Aplasia cutis congenita is a rare congenital defect seen in 1 in 10,000 births [1]. Defects early in gestation usually heal before birth and may appear as atrophic, bullous, membraneous or as scales with associated alopecia but less matured effects usually present as ulceration deep down to the bone. The most common type of defect is membranous. These lesions are usually solitary situated near the vertex of the scalp lateral to the midline.

Syndromic and non-syndromic aplasia cutis congenital are the two forms of this rare disorder, the latter being more common. In syndromic type characteristic skin lesions can occur with conditions like Johanson-Blizzard syndrome, Trisomy 13 and Adams-Oliver syndrome. The etiology is still unclear though genetic components, teratogens, amniotic adhesions, drugs like-methimazole, benzodiazepines, have all been implicated [2]. A mutation in the GTPase BMS1 coding protein that delays 18S ribosomal RNA maturation and consequently reduces cell proliferation recently has been linked to aplasia cutis congenital [3]. Commonly a benign isolated defect, association of other physical anomalies or malformation syndromes has to be ruled out. Urgent intracranial ultrasonography for any associated intracranial malformations should be done. In our case, intracranial ultrasound was normal so a diagnosis of non-syndromic type was made. Dermatoscopy can be performed to determine, whether hair follicles are present, if the diagnosis is uncertain. Radially arranged hair bulbs which are visible through the translucent epidermis are characteristic [4]. Treatment usually involves a conservative approach for less severe cases. These cases generally resolve within weeks to month, but alopecia tends to persist. If lesions fail to heal or for cosmetic reasons, surgical excision and primary repair can be performed. Haemorrhage, secondary local infections, meningitis, or sagittal sinus thrombosis are the complications of aplasia cutis congenita, though rare. Genetic counseling considered only if a strong family history is present or if associated other abnormalities are there. The prognosis is usually excellent unless associated with other anomalies.

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FINANCIAL OR OTHER COMPETING INTERESTS:

None.

Date of Publishing: **Oct 01, 2017**