

Edward Syndrome with Aplasia Cutis Congenita: A Rare Case Report

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ABSTRACT

Aplasia cutis congenita (ACC) is a rare developmental malformation characterized by the absence of skin, extending to bone or dura in a localized or widespread area at birth. Mostly it's seen in the scalp with an autosomal dominant inheritance and slight female predisposition. Generally, it is found as a solitary lesion without other anomalies, but sometimes it is reported associated with other syndromes. The association is

much appreciated in case of malformations like Patau syndrome, Wolf-Hirschhorn Syndrome, Johanson-Blizzard Syndrome etc. It is not commonly seen with Edward Syndrome. Normally Edward syndrome is a rare entity and its association with ACC makes it very rare. As the survival rate of Edward syndrome is very low, these types of associations are unavailable in the literature. We present this very rare association and which was never reported before in my country.

Keywords: ACC, Edward syndrome, Trisomy

CASE REPORT

A twelve day old term female baby presented to neonatal outdoor with chief complaint of respiratory difficulties and refusal to feed. Baby was apparently alright after birth. To start with she had hurried breathing for two days and refusal to feed for one day. Mother had no history of premature rupture of membrane or prolonged fever. Baby was not given any prelacteal feeds. She was born by normal vaginal delivery. Her birth weight was 2345 grams, APGAR score on 1 and 5 minute was 7 and 9

respectively. Baby cried immediately after birth and was feeding well. On day 12 her weight was 2456 grams with head circumference 30 cms, length 48 cms. She was centrally cyanosed, lethargic, tachypnoeic, febrile and sick with all the reflexes diminished. Morphologically she had low set of ears, triangular face, small mouth, distended abdomen, overlapping of toes and fingers [Table/Fig-1,2], in scalp there was a red denuded lesion posteriorly [Table/Fig-3]. Baby had normal female genitalia with patent anus. She was having poor cry.



[Table/Fig-1]: Syndromic facies

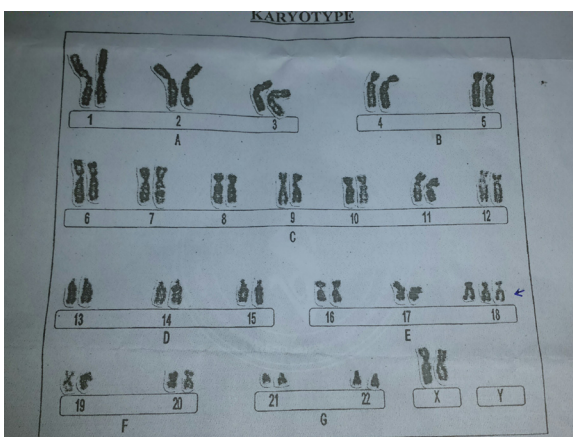


[Table/Fig-2]: Over riding of toe

On systemic examination she had respiratory rate of 76 per minute with sternal indrawing, grunting, nasal flaring. SPO₂ was 81% on admission without oxygen. On auscultation bilateral crepitation was there. Heart was 146 per minute. S1 and S2 distinctly audible with a pan systolic murmur of Grade-4 in mitral area. Abdomen was soft with no hepatosplenomegaly. Baby was given oxygen through nasal prong and worked up for further. The sepsis screen was positive having CRP 11mg/dL, MICRO ESR 20 mm/1st hr, band cell 7% and immature neutrophil 30%. Echocardiography suggested small perimembranous VSD of 5 mm. After initial stabilisation baby was treated for late onset sepsis in NICU. She was given IV antibiotics (Cefotaxime and Amikacin), oxygen, and routine warm care. The respiratory distress did not decrease with all the conventional measures. Congenital absence of skin in scalp was investigated by the dermatologists and it was diagnosed to be aplasia cutis congenita. Karyotyping was done for diagnosing cause of syndromic morphology. It came to be trisomy of 18th chromosome [Table/Fig-4] (with an arrow near trisomy). Baby had multiple episodes of apnoea on day 4 of admission. She was Intubated and kept in mechanical ventilator for 14 hours later on day 5 evening baby succumbed in NICU due to respiratory failure.



[Table/Fig-3]: Triangular facies with low set ears



[Table/Fig-4]: karyotype imaging

1 Scalp ACC without multiple anomalies
2 Scalp ACC with associated limb anomalies
3 Scalp ACC with associated epidermal and Organoid nevi
4 ACC overlying embryologic malformation
5 ACC with associated fetus papyraceous or placental infarct
6 ACC associated with epidermolysis bullosa
7 ACC localized to extremities without blistering
8 ACC caused by teratogens
9 ACC associated with malformation syndromes

[Table/Fig-5]: Frieden Classification

DISCUSSION

Aplasia cutis congenita (ACC) is a rare congenital skin condition. It was first described by Cordon in 1767, and there were more than 500 cases reported around the world with a slight more incidence in females [1]. 25% of the reported cases are familial with a vast majority (69%) showing an autosomal dominant inheritance [2]. The incidence is 1-3/2000-10000 [3]. It is congenital defect of skin which can be anywhere in body. Generally the most common site of lesion is the scalp. Specifically in the vertex of cranium as a solitary lesion [4]. However, it can be associated with other physical anomalies or as part of malformation syndromes. Syndromic association of the ACC are: Adams-Oliver Syndrome (associated with cardiac pathology, terminal transverse limb defects), Barts Syndrome (associated with blisters in oral mucosa and nail abnormalities), Trisomy 13 -Patau Syndrome (associated with characteristic facial and cardiac anomalies), Ellis-van Creveld Syndrome, Johanson-Blizzard syndrome (autosomal recessively inherited, had features of scalp ACC and other important features such as nasal alar hypoplasia, dental anomalies, pancreatic insufficiency and congenital deafness, amniotic band syndrome, Kabuki syndrome, oculocerebrocutaneous (Delleman) syndrome, scalp-ear-nipple syndrome (Finlay-Mark syndrome), Wolf-Hirschhorn syndrome, split cord malformation (SCM) [3,5]. May be seen in some other syndromes. Out of all trisomies ACC has a good association with trisomy 13 (Patau syndrome). However getting ACC with trisomy 18 (Edward syndrome) is rare entity. The lesions of ACC varied considerably. It could be linear, round, oval, stellate, or even punched out in shape. In the present case, it's a punched out shape. There are lots of cases without any aetiological factor in the literature [6]. Frieden classified the aetiology of aplasia cutis into nine types [Table/Fig-5].

The pathogenesis of ACC was not completely understood, and several hypotheses have been proposed. Some authors suggested that ACC may be correlated with neural tube defects [7], according

to some other school of thought genetic factors play an important role in the development of ACC, either chromosomal abnormalities or monogenetic inheritance expressed as autosomal dominant, autosomal recessive or X-chromosomal inherited syndromes [7]. Some authors think that the cause may be multifactorial.

In this case mother was healthy and free of any teratogenic drug intake, there was no history of amniotic band syndrome in prenatal period. She was firstborn and without any consanguinity. The ACC lesion was in scalp and was a punched out one. The baby had characteristic morphological abnormalities (microcephaly, lowset ears, triangular face, micrognathia, congenital heart disease, over riding of fingers and toes). Finally the karyotyping confirmed the diagnosis of trisomy 18 (Edward syndrome). Edwards's syndrome is a common chromosomal disorder due to the presence of an extra chromosome 18, full mosaic trisomy, or partial trisomy 18q. The live born prevalence is estimated as 1/6,000-1/8,000 [8]. Despite the well known infant mortality, approximately 50% of babies with trisomy 18 live longer than one week and about only 5-10% of children beyond the first year [8]. In this case also the baby didn't survive inspite of all the NICU Care.

Trisomy 18 and association of ACC is never reported. In our patient the baby had all the features of Edward syndrome and ACC. There may be an association of Edward syndrome and ACC. Much more research needed to sort out the true association of these two anomalies.

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