

Aplasia Cutis Congenita with Ectopic Mongolian Spot in a Child of a Patient of Multiple Sclerosis: A Rare Case Report

Shiti Bose*, Niharika Jha, Emy Abi Thomas and Anuradha Bhatia

St Stephens Hospital, New Delhi, India

*Corresponding author: Shiti Bose, St Stephens Hospital, New Delhi, India, E-mail: shitibose@yahoo.in

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Abstract

Aplasia cutis congenita (ACC) is a rare heterogenous disorder which is characterized by focal absence of skin since birth. Underlying structures such as bone or dura may also be involved. ACC has been considered to be a forme fruste of a neural tube defect by many authors. It might occur in isolation or in association with certain syndromes. We report a case of a new born male with membranous type of ACC over vertex extending to the left parietal region with partial agenesis of parietal bone and ectopic mongolian spot over left ankle.

Keywords: Aplasia cutis congenital; Heterogenous disorder; Neural tube defects

Introduction

ACC is a rare congenital development disorder in which a well localized absence of epidermis, dermis and rarely subcutis occurs [1-3]. Its incidence is 1-2.8 per 10,000 live births [1,4]. Exact etiology of ACC is not known, but is considered to be multifactorial and is affected by various factors like genetics, intra-uterine trauma, infection, decreased blood supply to skin and teratogens (carbimazole, methimazole, valproic acid, non-steroidal anti-inflammatory drugs) [1,2,5,6]. ACC can occur anywhere on the body but most commonly seen on the scalp [1,5,7]. It has been known to occur in children born to mothers on antithyroid and antiepileptics; however in our case neonate's mother is a known case of multiple sclerosis and was on oral steroids and vitamin B₁₂ supplements in her first trimester. She also received a single dose of intravenous immunoglobulin (IVIG) in her first trimester.

Case Report

A dermatological consultation was sought for a new born male child who had a localized absence of skin over scalp since birth. The newborn was full term with, a birth weight of 2915 g born by normal vaginal delivery. The previous sibling was female, also born by normal vaginal delivery, and no such defects were present in this child. No family history of similar condition could be elicited.

The mother of the neonate is a known case of multiple sclerosis and was on oral steroids (1 mg/kg Tab. Prednisolone) and vitamin B₁₂ supplements in her first trimester. She also received a single dose of IVIG. There was no previous history of herpes (varicella/zoster) in the mother and her antenatal period otherwise was uneventful.

Cutaneous examination revealed a localized area of absence of skin on the left side of the scalp over vertex extending up to the parietal region. Defect was present till the level of sub-cutis and underlying bone could be seen. Haemorrhagic discharge was noted from the area. Palpation of the area showed soft tissue with underlying bone. Hair around the defect was normal (collar sign was negative). No scarring

was noticed around the defect and size of the defect was 3 × 5 cm² (Figure 1,2). A thorough physical examination including neurological examination was within normal limits.

A CT scan was done to rule out any neurological involvement, which revealed partial agenesis of parietal bone. No other organ abnormalities were found. Routine haematological and biochemistry investigations were within normal limits.

The area was treated with topical mupirocin ointment and vaseline gauze dressings along with intravenous antibiotics after taking a surgical opinion for the same. The neonate's parents were advised regular follow-up to prevent any infections or complications in future.

Hence, a diagnosis of membranous type of ACC was made. This neonate also had an ectopic mongolian spot over left ankle (Figure 3).



Figure 1: Aplasia cutis congenita on the scalp.



Figure 2: Aplasia cutis congenital.



Figure 3: Ectopic Mongolian Spot in the same neonate.

Discussion

Aplasia cutis congenita is an uncommon congenital disorder associated with localized absence of epidermis, dermis and rarely subcutis at birth. Occasionally, underlying bone or dura can also be involved. In about 80% cases scalp is involved. Maximum tensile forces during rapid development of brain might result in disruption of overlying skin at 10-18 weeks of gestation [5]. Sometimes, a collarette of dark, coarse hair is seen surrounding the defect (collar sign), which was absent in our patient. It may also involve trunk and/or extremities and are usually bilaterally symmetrical [1,2,8]. Although ACC is usually benign, it can be associated with other physical anomalies and chromosomal disorders.

Frieden classified ACC into nine types based on the number and location of lesions and associated malformations, as shown in (Table 1) [4].

Type	Area involvement/ associated anomalies	Inheritance
I	Isolated scalp involvement; may be associated with single defects	AD
II	Scalp ACC with limb reduction defects (Adams-Oliver syndrome); may be associated with Encephalocele	AD
III	Scalp ACC with epidermal nevus	Sporadic
IV	ACC overlying occult spinal dysraphism, spina bifida, or meningoencephalocele	Sporadic
V	ACC with placental infarcts	Sporadic
VI	ACC with epidermolysis bullosa	AD or AR
VII	ACC localized to extremities without blistering; usually affecting pretibial areas and dorsum of hands and feet	AD or AR
VIII	ACC caused by teratogens (e.g., varicella, herpes, methimazole)	Sporadic
IX	ACC associated with malformation syndromes (e.g., trisomy 13, deletion 4p-, deletion Xp22.1, ectodermal dysplasia, Johanson-Blizzard syndrome, Adams-Oliver syndrome)	Variable

ACC: Aplasia cutis congenita; AD: Autosomal dominant; AR: Autosomal recessive

Table 1: Frieden's classification of Aplasia cutis congenital.

ACC itself is a rare disease and its association with maternal multiple sclerosis, intake of steroids and IVIG is even more rare. Till date there have been no cases reported wherein there was a development of ACC in neonates born to mothers with multiple sclerosis or to mothers on corticosteroids. In the present case scenario a single dose of IVIG is highly unlikely to cause ACC in the neonate and multivitamins are not known to cause ACC or have any teratogenic properties in neonates. A randomized trial conducted by the Medical Research Council [9] showed a 60% reduction in risk of neural tube defects among women using folic acid supplements compared with women using multivitamins containing no folic acid. In a review done by Janssen et al., corticosteroids are relatively safe if given in low doses (Category B drug) although they are known to cause adrenal insufficiency and infection in neonates [10]. Steroids cross the placental barrier and may increase the risk of cleft palate and low birth weight when used in the first trimester [11]. The foetus receives about 10% of the maternal dose of corticosteroids. The effects of moderate to higher doses have not been determined till date [10].

A case reported by Iftikhar et al. showed the presence of extensive ACC in a neonate born to a mother with pemphigus vulgaris on treatment with low dose steroids and Azathioprine [12]. The dose of oral Prednisolone given to our patient in order to control the symptoms of multiple sclerosis was 1 mg/kg per day.

Due to lack of supporting literature it was difficult to determine as to whether either corticosteroids or maternal multiple sclerosis caused ACC in the neonate, hence a possibility of either is considered in the present scenario. To the best of our knowledge such a case has not been reported till now.

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