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Original Research Paper

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APLASIA CUTIS CONGENITA – A CASE SERIES OF VARIED PRESENTATIONS

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ABSTRACT Aplasia cutis congenita (ACC) belongs to a heterogeneous group of disorder, characterised by localised absence of skin and in some cases subcutaneous tissues, involving multiple possible body locations. The most common site is the scalp (70%), however any skin site can be affected including the trunk and the extremities .

Herein we report a series of 4 cases of aplasia cutis congenita with varied cutaneous manifestations ranging from absence of epidermis to deeper defects extending upto the dermis and subcutaneous tissue. Our first case presented to us with congenital absence of skin with ulceration and eschar formation of extremities, buttocks and bullae formation over hands and lower back. Clinicopathological diagnosis was made to be aplasia cutis congenita associated with epidermolysis bullosa (Bart's syndrome). The second case presented with a single, well defined, rhomboid shaped deep ulceration with overlying eschar formation over the vertex extending upto the right parietal region of scalp, diagnosed to be a case of Type 2 Aplasia cutis congenita - Adams-Oliver syndrome. Our third case was a superficial variant of aplasia cutis congenita with linear , erythematous, blanchable atrophied parchment like scar on the back, without any underlying bony deformity. The last was a sporadic case of bitemporal aplasia cutis congenita with thinning of skin over bilateral temporal areas with overlying telangiectasia.

KEYWORDS : Aplasia cutis congenita, Bart's syndrome , Adams Oliver syndrome.

INTRODUCTION

Aplasia cutis congenita is a heterogeneous group of disorder characterised by localised absence of skin and in some cases subcutaneous tissues, involving multiple possible body locations . The most common site is the scalp (70%), however any skin site can be affected including the trunk and the extremities. [1] It was first described by Cordon in 1767 and later classified by Frieden based on the number and site of the lesions as well as presence or absence of associated malformations.

Frieden classification for Aplasia cutis congenita :

Group 1: scalp lesions without other anomalies

Group 2: scalp lesions with associated limb abnormalities – includes Adam's Oliver syndrome

Group 3: scalp lesions associated with solitary epidermal and sebaceous naevi or an epidermal naevus syndrome

Group 4: acc overlying deeper embryologic malformations such as meningomyelocele, cranial stenosis, spinal dysraphism, gastroschisis and omphalocele

Group 5: Stellate acc of trunk or limb associated with fetus papyraceus and placental infarcts

Group 6: aplasia cutis congenita associated with epidermolysis bullosa (Bart's syndrome)

Group 7: aplasia cutis congenita localised to the extremities without epidermolysis bullosa.

Group 8: aplasia cutis congenita caused by teratogens

Group 9: aplasia cutis congenita with associated malformation syndromes – Patau syndrome (trisomy 13 with large membranous defect); Wolf – Hirschhorn syndrome (deletion of short arm of chromosome 4) with midline scalp defects; Setleis syndrome with bitemporal acc and abnormal eyelashes; Johanson-Blizzard syndrome with stellate scalp defects; focal dermal hypoplasia (Goltz syndrome); and others . [2]

Apart from those in the above classification, a few other variants of aplasia cutis congenita reported are: bitemporal aplasia cutis congenita, a superficial variant involving only the epidermis. We report the following 4 cases of ACC with varied cutaneous manifestations and discuss about the prognosis and management strategies .

Case Reports

Case 1 :

A 14 day old full-term male weighing 2.4kgs born of consanguineous marriage, presented to us with congenital absence of skin, ulceration and eschar formation of both upper and lower limbs and buttocks. The perinatal history of the mother was uneventful. The first child is normal and there is no positive family history .

On general examination, pallor was seen. Systemic examination revealed Grade II systolic murmur. Hemogram revealed anemia and thrombocytopenia. Histopathological examination showed necrotic epidermis separated from the underlying dermis with supepidermal blisters.

Dermis showed dense collection of neutrophils and eosinophils alongwith increased number of capillaries. Blister cavity showed fibrin, rbcs and neutrophils.

The patient was diagnosed with Type 6 Aplasia cutis congenita with dystrophic epidermolysis bullosa - Bart's syndrome

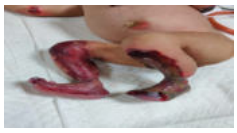

Fig. 1: A case of Bart's syndrome with large eroded areas over both lower extremities

Fig. 2: Case of Bart's syndrome with ulceration and Eschar formation over lumbosacral region

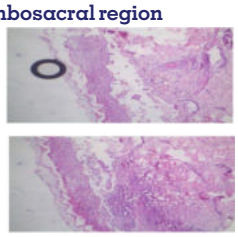

Fig. 3 : Histopathology showing separation of epidermis from the underlying dermis with subepidermal blister formation. Dense collection of neutrophils and eosinophils in the dermis.

Case 2:

A day 3 old term Male baby, born of non consanguineous marriage was brought with absence of skin over scalp since birth. There was no history of preceding fluid filled lesions over the scalp, natal tooth or neonatal seizures. There was no history of maternal intake of oral medications. Mode of delivery was normal vaginal delivery . The elder sibling did not have any abnormality.

Examination revealed the presence of single, well defined 10 cm X 7cm, rhomboid shaped 0.5cm deep ulceration with overlying eschar formation over the vertex and right parietal region of the scalp . The peripheral skin shows oedema and erythema . No obvious underlying bony defect was palpable . Anterior fontanelle was open. Brachydactyly of left foot was seen. Ophtalmological, ENT and dental examination was normal . Hemogram and biochemical investigations were normal. The patient was diagnosed as a sporadic case of Type 2 Aplasia cutis congenita - Adams-Oliver syndrome.

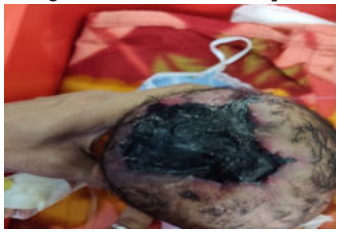

Fig. 4 : Scalp defect in a case of Adam's Oliver syndrome

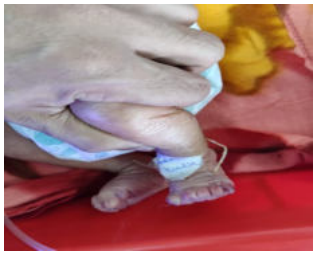

Fig. 5 : Clinical photograph showing brachydactyly in the same case of Adams- Oliver syndrome

Case 3:

A female neonate born at term presented with a single, linear, erythematous, atrophied parchment like scar on the back, which blanched on pressure. The surrounding skin was tense and shiny . No underlying bony deformity was palpable. The lesion was suggestive of superficial aplasia cutis involving only epidermis.

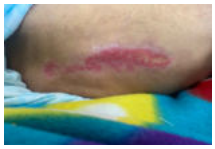

Fig. 6 : Superficial aplasia cutis congenita presenting with a linear, atrophied, blanchable erythematous parchment like scar on the back

Case 4: A day 7 old male child born at term by caesarean section after twin pregnancy presented with thinning of skin over bilateral temporal areas with overlying telangiectasia. No similar lesion was seen in the other twin.

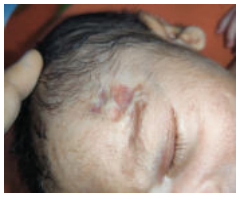

Fig. 7 : Case of bitemporal aplasia cutis congenita with defect over left temporal region.

Fig. 8 : Clinical photograph of the same baby showing defect over right temporal region.

In all the above cases, the perinatal history in the mothers were uneventful. There was no history of maternal intake of oral medications. There was no positive family history. Mode of delivery was normal vaginal delivery in all except the 4th case. General and systemic examination were normal. Hematological and Biochemical parameters were within normal limits.

DISCUSSION:

Aplasia cutis congenita is a rare congenital disorder of the skin . The 9 groups as classified by Frieden is based on the number is based on the number, location, pattern of lesions, presence or absence of associated abnormalities and mode of inheritance .

No obvious pathophysiology of ACC could be described. According to literature, there are multiple probable factors which contribute the development of acc :

- Chromosomal abnormality, especially BMS 1, a recent study implies the UBA2 gene and the SUMOylation pathway
- Trauma
- Premature amniotic membrane rupture and amniotic band formation
- Intrauterine complications such as vascular accidents or infection
- Thrombosis, vascular lesions
- Teratogens : misoprostol, benzodiazepine, valproic acid, cocaine, methotrexate, Acei, methimasole
- Nutritional or vitamin deficiencies. [3]

Majority of the cases are sporadic, but familial occurrences have also been described. Family history of a similar lesion in another sibling suggests a possible genetic factor. [4] Although in majority of cases(70%), ACC manifests as a solitary defect in the scalp, but occasionally it may present with multiple lesions (such as in our 1st case) or at other sites (involvement trunk in our 4th case). The lesions are non-inflammatory and well demarcated. Ulcerations may vary in size from 0.5 cm to 10 cm. A tuft of hair surrounding aplasia cutis may denote

underlying malformation with neural tube defect. [5] Superficial aplasia cutis involves only the epidermis. Shallow defects usually heal

in utero leaving a scar. Deeper defects can extend upto the dermis, subcutaneous tissue, and rarely periosteum, skull, or dura. Aplasia cutis may partially heal before delivery and appear as a hairless, atrophic, membranous, parchment like or fibrotic scar. Membranous aplasia cutis is a flat, white membrane overlying a defect in the skull. Distorted hair growth, known as the hair collar sign, is a marker for an underlying cranial defect such as encephalocoele, meningocoele, and brain tissue outside the skull. A rare bullous variant of aplasia cutis congenita has also been reported.

There are reports of association of aplasia cutis congenita with congenital malformations of the heart, gastrointestinal, genitourinary (such as gastroschisis or omphalocele), and central nervous systems (such as meningomyelocele or spinal dysraphism). [6]

Complications are rare but may include arterial bleeding, secondary wound infection, sagittal sinus thrombosis or fatal infection of the brain.

The management through conservative route has shown excellent healing results. It consists of regular wound cleansing and application of dressings alongwith the use of systemic antibiotics. This include physiological saline, continuous saline drips, fusidic acid cream or mupirocin ointment application and liquid paraffin embedded gauze dressings. These aim at preserving moisture, preventing desiccation, allowing granulation tissue and healing by secondary intention, thereby avoiding potential operative risks. There have been reports of even more specialized conservative treatment techniques such as the use of autologous cultured fibroblasts and keratinocytes or application of fibroblast growth factors. There is hardly any role of early surgical intervention. [7] Some cases may require multidisciplinary approach with early physiotherapy and surgical management in the form of primary wound closure, skin grafts or skin flaps.

CONCLUSION:

Aplasia cutis congenita is a rare variably expressed congenital disorder characterised by the absence of epidermis, dermis and occasionally subcutaneous tissues, involving multiple body sites, the most common location being the scalp. Majority of the cases can easily be managed in a limited resource setting. The management is conservative with daily cleaning and dressing of the lesions with frequent surgical reviews of the healing process. Surgical intervention is sometimes required and in those cases is mostly reconstructive and / or cosmetic.

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