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Case Series

A rare case series of congenital absence of skin – Aplasia cutis congenita

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Abstract

Aplasia cutis congentia is a rare heterogenous group of disorder with focal absence of the skin and its underlying structures with a varying depth ranging from the epidermis to even subcutis, bone or even dura. Depending upon the onset & duration of insult during pregnancy it can present as a open wound that needs prompt treatment or a healed scar. This has a rare occurrence with upto 500 cases reported in literature. In this article, we report a rare case series of aplasia cutis congenita and review of literature.

Keywords: Aplasia cutis congenita congenital anomaly, Focal absence of skin, ACC, Aplasia cutis, Hair collar sign.

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1. Introduction

Aplasia cutis congenita (ACC) is a congenital malformation with focal absence of the tissues with varying severity of depth ranging from the epidermis, dermis, subcutaneous fat, bones (20%) and even dura.[1] This was first described by Cordon in year 1767 and the later by Campbell in the year 1826. ACC most commonly has sporadic inheritance however autosomal dominant and recessive patterns have also been described.[2] The incidence of aplasia cutis in consanguineous marriage is 7% when compared to non-consanguineous marriage which is 2% with a male: female ratio of 7:5.[1-5]

2. Case Series

2.1. Case 1

A 30 days old healthy male baby born of full term normal vaginal delivery with a birth weight of 2 kgs came with the complaints of hair loss and ulceration over the scalp since birth. Antenatal period was uneventful. History of onset of bullae over the scalp that ruptured to form non-healing ulcers.

Examination revealed a 4X3 cm atrophic plaque extending from the occipital to the parietal region of the scalp. There were two ulcers with central crusting situated over the lambdoid suture. The surrounding skin showed erythema and telangiectasia (**Figure 1**). There was a history of bleeding on trivial trauma. Gram stain showed gram positive cocci in clusters suggestive of staphylococcus aureus. Patient's parents were unwilling for biopsy. Hence, histopathological examination could not be done. CT & MRI scan was done and was found to be normal. Patient was managed conservatively with oral and topical antibiotics & hydrogel dressing.

2.2. Case 2

A 2-month-old healthy male baby, born of full term normal vaginal delivery with a birth weight of 2.7 kgs was brought to the dermatology OPD with the complaints of patchy hair loss over the vertex since birth. Antenatal period was uneventful. There was no history of similar complaints in his family.

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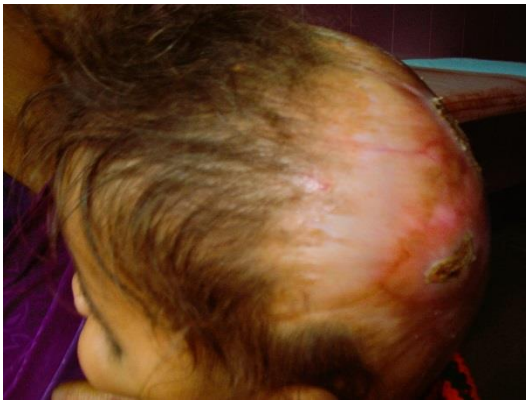

Figure 1: Shows ulcerative plaques with surrounding erythema and telangiectasia over the scalp.

On examination: A single erythematous, round atrophic plaque of size 3x3 cm with a rim of terminal hairs encircling (hair collar sign) over the margin was seen (**Figure 2**). Surrounding skin and hair was normal and there was no signs of inflammation.

MRI scan was done and was within normal limits. Skin biopsy (3 mm) was done and it showed absence of the epidermis with dermal fibrosis and absence of dermal appendages which was consistent with aplasia cutis congenita. Since the baby was asymptomatic and the scar tissue was healthy. The parents were reassured.

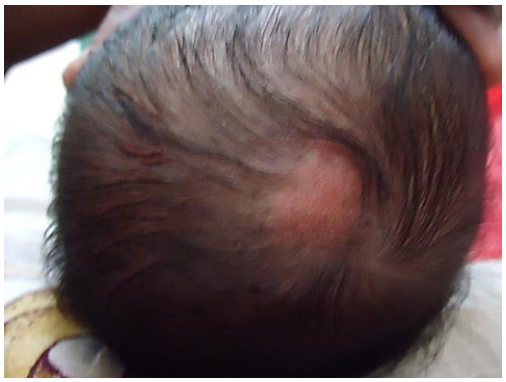

Figure 2: Shows an erythematous atrophic plaque with hair collar sign

2.3. Case 3

A 2 months old healthy baby born of full term normal vaginal delivery with a birth weight of 1.8 kgs born via full term normal vaginal delivery came with the complaints of a single alopecic erythematous patch of size 3X 3 cm with central atrophy over the left parietal region of the scalp since birth. There was no history of bleeding or ulceration. Antenatal period was uneventful. Dermoscopy showed absence of hair follicles and telangiectasia. MRI was found to be normal. Since the baby was asymptomatic and the scar tissue was healthy, the parents were reassured. Biopsy couldn't be done as the parents were not willing.

Figure 3: Shows atrophic alopecic erythematous patch over the parietal region of scalp

3. Discussion

ACC also known as cutis aplasia or epitheliogenesis imperfecta is usually focal & solitary. The most common site affected is the vertex of the scalp (70%). The reason behind this is the tensile force exerted due to the rapid growth of the brain over the vertex during its expansion in around 10-15 weeks causing disruption of the underlying structures. This presents as oval, round, linear or stellate hairless atrophic plaque. Sometimes there is presence of a ring of long hairs encircling the margin of the defect known as the *"hair collar sign"* as shown in **Figure 2**. The significance of the sign is that it is a potential indicator of neural tube defect.[6-8,9]

Though rare, it can be multiple or widespread (also known as systemic involving >90% of body surface area with extracutaneous congenital anomalies). Most of the babies with widespread involvement have multiple congenital anomalies and are still born. Clinically. It can be of two types like – membranous (covered with thin, translucent glistening membrane) or non-membranous. Depending upon the duration, its manifestations vary. For example, if the time of insult is in early gestation, it presents as a healed atrophic scar and if the insult occurs in later gestation it presents with bulla, erosions, fissures or ulcers with a granulating base. Frieden classified aplasia cutis congenita into 9 types in 1986 and is still accepted and widely used as shown in **Table 1**.[10] The etiology of aplasia cutis congenita is unknown however various risk factors have been associated with its occurrence as shown in **Table 2**.

Histopathology of ACC shows complete or near complete absence of the skin and the appendageal structures. Healed lesions show flattened epidermis with absent adnexa and dermal fibrosis in which the dermal collagen are arranged in a compact, more parallel configuration with ectatic vessels.[11] Dermoscopic examination reveals absence of follicular structures with telangiectatic vessels.[12]

Table 1: Shows Frieden's classification of aplasia cutis

Frieden classification of ACC types	
1	Aplasia cutis located over the scalp with no other anomalies
2	Aplasia cutis located on the scalp with associated limb anomalies like 1. Limb malformations (Adams-Oliver syndrome) 2. Hypoplasia or aplasia of distal phalanges 3. Vascular malformations, hair or nipple abnormalities, fibromas
3	Aplasia cutis of scalp with associated epidermal nevi, neurological or ophthalmologic abnormalities
4	Aplasia cutis accompanied by embryologic deformities such as omphalocele, leptomeningeal angiomatosis, cranial stenosis, porencephaly, meningomyelocele, spinal dysraphism or gastroschisis
5	Aplasia cutis congenita along with foetus papyraceous, placental infarct or extensive aplasia cutis over trunk and limbs.
6	Aplasia cutis and epidermolysis bullosa involving the lower extremities
7	Aplasia cutis with no epidermolysis bullosa involving extremities
8	Teratogen associated ACC – Herpes simplex, Varicella, intrauterine infections and drugs during pregnancy such as methimazole or carbimazole
9	Aplasia cutis accompanied by congenital malformations such as Patau syndrome (Trisomy 13), Wolf-Hirschhorn (4p deletion), Seteis syndrome Johanson-Blizzard syndrome, Goltz syndrome, ADAM complex, Kabuki syndrome Delleman syndrome, Finlay-Mark syndrome, XY gonadal dysgenesis.

Table 2: Showing the risk factors of aplasia cutis congenita

Chromosomal abnormality	• BMS1(Ribosome biogenesis factor)[3] • Ubiquitin-like modifier-activating enzyme 2 • SUMOylation pathway[4](post translational modification of small ubiquitin-like modifiers-SUMO)	
Trauma	• Focal pressure necrosis[5]	
Defect in embryological development	• Incomplete fusion of the mesoderm[6] • Neural tube defects • Ectodermal dysplasia	
Amniotic irregularities	• Rupture of amniotic membrane forming amniotic bands	
Intrauterine complications	Infections	• Varicella • Herpes simplex • TORCH infections • Typhoid[7]
	Vascular complications	• Thrombosis • Vascular accidents
Teratogenic drugs	• Methimazole • Misoprostol • Methotrexate • Benzodiazepine • Valproic acid • ACE inhibitors • Cocaine	
Fetus papyraceous	This is more commonly associated with truncal aplasia cutis. When there is a multiple gestation and there is death of a fetus at ~10-12 weeks, the dead fetus releases thrombogenic substances that can cause placental infarction or DIC(disseminated intravascular coagulation) that causes vascular insufficiency leading to defective development of the underlying structures causing aplasia cutis	

Evaluation in the form of imaging techniques should be done to rule out the involvement of deeper structures. The blood investigations are within the normal range. Imaging techniques like ultrasonogram & MRI should be done to see the depth of involvement of the underlying structures. Though not specific, High alpha fetoprotein and positive acetylcholinesterase have been associated with aplasia cutis congenita in maternal serum.[13]

Due to the absence of the protective structures like the skin and soft tissues, patients are easily susceptible to trauma and excessive bleeding & non healing ulcers which must be properly taken care of to prevent complications like hemorrhage and secondary infections. Management of aplasia cutis congenita depends on its size, location, depth of involvement and associated involvement.[14] In cases of small defects with erosion, ulcer or bulla. Prompt treatment with appropriate antibiotics should be initiated as infections form an important cause of death in aplasia cutis. Other conservative managements like use of autologous cultured fibroblasts and keratinocytes, application of fibroblast growth factors to accelerate wound healing.[15]

If it is a larger lesion(>10 cm), it is recommended to get it surgically repaired by primary wound closure , skin grafting , flaps closures (rotational flaps, L- flaps, free or muscle flaps based on the defect). In case of bone defects use of bone grafts or cranial vault reconstruction can be done.

The complications of aplasia cutis are related to the depth, size and site of the lesion. If case of a large lesion (i.e >10 cm), it takes a longer time to heal and the chances of infection, bleeding and the probability of skull defects is higher. If the lesion is located over the sagittal sinus, the chances of haemorrhage is greater. Other complications include meningitis, psychomotor retardation and paresis.

Prognosis is usually good when it's a small defect limited to skin without involvement of deeper underlying structures. If it is a larger defect and care is not taken properly, death can occur due to infection or haemorrhage. Systemic aplasia cutis involves greater than 90% of body surface area with complete loss of skin, its appendages and internal organs is fatal.[16]

4. Conclusion

Aplasia Cutis Congenita (ACC) is a rare congenital disorder characterized by the absence of skin and underlying structures, often presenting as solitary or multiple lesions. While the etiology remains unknown, various risk factors have been associated with its occurrence. Prompt diagnosis and management are crucial to prevent complications such as infection, bleeding, and mortality. Treatment approaches vary depending on the size, location, and depth of the lesion, ranging from conservative management to surgical interventions. Understanding the clinical presentation, classification, and potential complications of ACC is essential for providing optimal care and improving patient outcomes.

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6. Conflict of Interest

None.

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