# "A RARE CASE OF CONGENITAL ABSENCE OF SKIN"

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**ABSTRACT**

Background: 'Congenital absence of skin' includes situations where the skin developed but has subsequently lost. At birth lesions may appear as scars or ulcers. Frieden classified them into 9 groups based on the number and presence or absence of other anomalies.

Case presentation : Here we are reporting a case with extensive congenital defects of the skin over scalp, face, neck, axilla and chest with symmetrical distribution and conjunctival xerosis, which is rarely seen. Microscopy showed moderate atrophy of epidermis with absence of adnexial structures. To our knowledge, this is the sixth case report from India of congenital absence of skin and second case from our institute.

Conclusion : The prognosis in congenital absence of skin is determined by the severity of skin lesions and underlying associated anomalies, so a regular follow up of the patient is required.

**Keywords**: Congenital absence of skin, Aplasia cutis congenita.

## INTRODUCTION

The term 'aplasia cutis congenita' (ACC) implies a failure of skin development, while the broader term 'Congenital absence of skin' includes situations where the skin develops but is subsequently lost. It was first described by Cordon in 1767.[1]

According to Western literature ACC is reported to affect 1 in every 10,000 live births and is very rare in Asian subcontinent.[1]

The common site of predilection is the midline area of the posterior scalp. At birth lesions may appear as scars or ulcers. On the scalp they may appear as parchment-like scars with alopecia. Involved areas are well-circumscribed, not inflamed, and vary in size from 0.5 to 10 cm or larger.[2]

Many theories have been considered for causation of congenital absence of skin, such as, mechanical events, amniotic bands, vascular malformation in utero, failure in the process of embryologic development and cutaneous rupture caused by pressure of cerebral development.[3]

Although usually benign, they may be associated with other physical abnormalities and syndromes. Frieden classified them into 9 groups based on the number and presence or absence of other anomalies. Nearly 86 percent belong to the first group with a solitary lesion.[4]

## CASE HISTORY

A 26-year-old boy of nonconsanguinous parents presented at BLDE Hospital, Bijapur with extensive congenital defects of the skin i.e. atrophic parchment-like scar within which there was a total absence of hair. These were present over face, neck, axilla, scalp, chest and lower extremities with symmetrical distribution and as well as conjunctival xerosis was noted which is rarely seen (Fig:1).

On physical examination there were no signs of acute distress, neurological impairment and no other organ abnormalities. Routine investigations were with in normal limits. Radiological examination and ultrasonography of abdomen revealed no abnormalities. No family history of similar condition and no history of medications or disease during pregnancy.

## PATHOLOGY

Punch biopsies were taken from the lesions over face, neck and axilla.

## MICROSCOPY:

Microscopic examination revealed moderate atrophy of the epidermis with absence of adnexial structures. Only one, atrophic hair follicles was seen with few chronic inflammatory cells in the deeper dermis. Focal areas in dermis showed increased amount of collagen (Fig:2).

**Histopathological Diagnosis**: Congenital absence of skin.

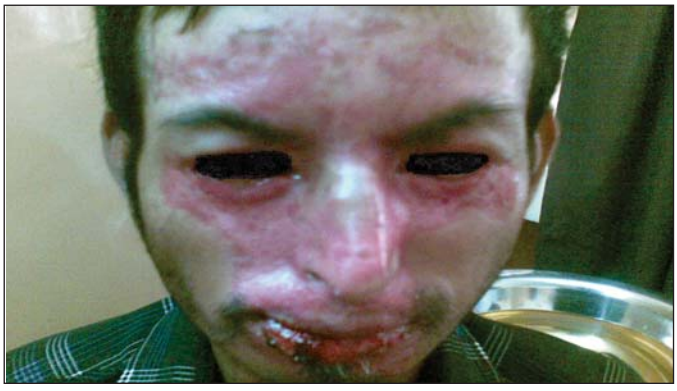

**Fig1: Photograph showing absence of skin around fore head, nose, and upper lip. Skin is replaced by sub-cutaneous tissue.**

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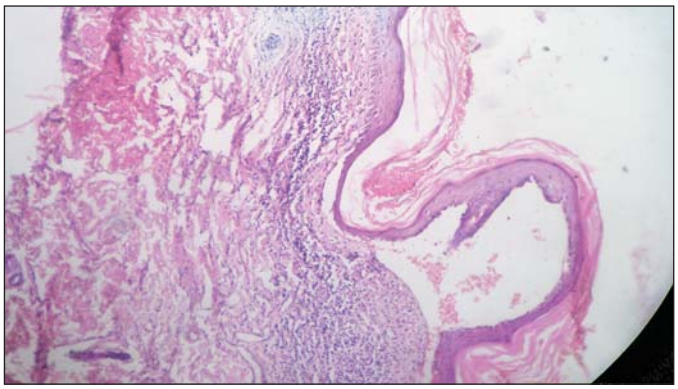

**Fig2: Microphotograph showing thinned out epidermis with absence of adnexial structures.**

**DISCUSSION:**

Congenital absence of skin includes situations where the skin develops but is subsequently lost. Aplasia cutis congenita (ACC) is a rare congenital disorder characterised by a localized absence of skin, dermal appendages, and in some cases subcutaneous tissues.[1] ACC may occur anywhere in the body; however, in 84% of cases, the defect is found in the scalp, where it is often solitary and located predominantly in the midline vertex. Non-scalp lesions may involve the trunk and/or extremities and are usually bilaterally symmetrical.[3]

The non-scalp ACC lesions are usually large size and may be associated with epidermolysis bullosa (EB). The association of ACC with EB may be a visible sign of other congenital anomalies like pyloric or duodenal atresia, ureteral stenosis, renal abnormalities, craniofacial abnormalities and nail dystrophy.

In literature, very few histological details are available; histological features vary depending on the depth of aplasia and duration. Ulcers are seen at birth. After healing, the epidermis appears flattened with proliferation of fibroblasts within a connective tissue stroma. Total absence of the epidermal appendages remains a characteristic feature.[4]

Treatment is rarely necessary as the lesion almost always heal spontaneously and rapidly. Occasionally, keloidal scarring and joint contracture can be seen. In cases with extensive involvement prevention of secondary infection, further trauma and effective temperature regulation can reduce mortality.[3]

Differential diagnosis should include transient bullous epidermolysis, congenital herpes, setleis syndrome, scalp electrode and pyoderma gangrenosum.

**CONCLUSION:**

The prognosis in congenital absence of skin and aplasia cutis congenita is determined by the severity of skin lesions and underlying associated anomalies, so a regular follow up of the patient is required.

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