



APLASIA CUTIS CONGENITA: A CASE REPORT

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ABSTRACT

Aplasia cutis congenita (ACC) is a congenital disease characterized with absence of skin usually on the scalp. No definite etiology is known however, multiple contributing factors such as intrauterine infection, fetal exposure to cocaine, heroin, alcohol or antithyroid drugs, teratogens, vascular disruption, genetic causes have been implicated. We report a neonate with an isolated lesion on the scalp in the form of a superficial ulcer which was covered with a thin membrane. There was no other genetic defect. Skin defect was managed conservatively and healed leaving an atrophic scar.

KEY WORDS: Aplasia cutis congenita, scalp defect, conservative management.



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INTRODUCTION

APLASIA CUTIS CONGENITA (ACC)

It is a rare congenital disorder characterized by a localized absence of epidermis, dermal appendages and in some cases subcutaneous tissues. It was first described by Cordon in 1767¹. ACC may affect any part of the body, however in 84% of cases; it involves the scalp that presents as solitary lesion located predominantly in the midline vertex². Non-scalp lesion may involve the trunk or extremities and are usually bilaterally symmetric³. Asymmetric distribution has been also reported⁴. At birth, the appearance of the lesion may vary from superficial erosion to a deep ulcer with the affected area covered with a thin and transparent membrane. Lesion occurs early in pregnancy; it may heal before delivery leaving a congenital atrophic alopecic scar. It may be associated with fetus papyraceus, epidermolysis bullosa, pyloric or duodenal atresia, renal abnormalities, ureteral stem oxis, nail dystrophy and cranio-facial abnormalities^{5,6}. Factors like intra uterine trauma, amniotic bands and drugs such as methimazole have been implicated⁷. Syndromes such as Adams-Oliver syndrome, SCALP Syndrome (Nevus

sebaceous, CNS malformations, ACC, limbal dermoid, pigmented nevus), Opitz syndrome and chromosomal disorders are associated with such lesion⁶. The main complication of larger defects includes infections, bleeding and thrombosis which may lead on to fatal outcome. Prompt diagnosis and appropriate treatment is mainstay of treatment which is critical for avoiding the adverse outcomes. Reported management is conservative and surgical. Allogenic dermal graft and cultured epithelial autografts have also been used to reconstruct the defects^{8,9}. Authors report such an infant managed conservatively.

CASE REPORT

A one day old full term male neonate with appropriate growth for gestational age, (birth weight 3255 gms, length 51c.m. and head circumference 33c.m.) presented with skin defect on scalp measuring 2x4c.m. (Fig.1). Physical examination showed no evidence of any other abnormalities. Urine and blood tests for, kidney and liver function were within normal limits. Lesion healed within one month leaving an alopecic scar (Fig. 2). Child is healthy and there is no other abnormality detected even on follow up.

Legends to figures



Figure 1
Skin defect with alopecia 2x4cm. at birth



Figure 2
Healed atrophic skin with alopecia 02 years after birth

DISCUSSION

ACC is a rare condition with an underlying mechanism that remains to be fully elucidated. Canter et al (2004) reviewed the literature & found 500 cases of ACC⁸. Recently also Piotr Brzezinski, Tudor Pinteala et al 2015 presented a case and reviewed the literature on ACC¹⁰. Sangeeta Velaskar 2015 has also reviewed the congenital absence of skin in IADVL Text book of dermatology¹¹. This congenital skin disorder can

present as an isolated condition, as in the present case or can co-exist with other genetic syndromes. Maternal cigarette smoking is known to be associated with the development of ACC⁸. Many drugs like methimazole, diclofenac sodium, valproic acid, marijuana and cocaine are also reported to be associated with its etiology¹². Individuals affected by ACC most commonly exhibit a unique circular defect on the scalp. Rarely it may involve wide spread areas including chest and flanks¹³. Skin defects are the primary presenting feature however, it

can also be accompanied by pulmonary as well as other congenital malformations, such as lumbo sacral sinus tracks, faun tail nevus and spinal cord anomalies^{6,7,14}. An ACC scalp defect can be an indicator of internal organ involvement with increased risk of complications, leading to poor outcome¹⁵. C. Moss' & H. Shahidullah 2013 describe Frieden types 5, 6 and 8, where skin was probably present at an early stage but was subsequently damaged and lost¹⁶. Management includes prevention and treatment of its complications e.g. infection, bleeding, electrolyte imbalance that results from epidermal water loss, nutritional deficiency from chronic blood loss and pain of wound dressing^{6,14}. The selection of treatment depends on the condition of the infant and presence of complications. Multiple treatment regimens have been reported in literature by earlier workers however, most cases of ACC associated with FP can be healed with conservative treatment¹⁵. Any evidence of cerebral damage bleeding and infections should be carefully monitored when the patient is being managed conservatively with repeated dressing changes. A multidisciplinary approach can be useful. Treatment for ACC is controversial which may be either conservative with dressings or surgery and combination of both^{8,9}. Dressing option may be moist dressing, burn cream

dressing with silver sulfadiazine and epidermal growth factor containing dressing. Skin grafting and local flaps are the predominant surgical treatment options for ACC. Skin graft is indicated for large facial defects that are possibly at risk of sagittal sinus thrombosis and cerebral hemorrhage¹⁷. Surgical management is reserved for large and deep lesions that do not heal with conservative treatment

CONCLUSION

Aplasia cutis congenita (ACC) is a rare disorder of defect in components of skin usually affecting the scalp. It may be solitary or associated with other congenital abnormalities. Knowledge of co-existing genetic syndromes is essential to manage them properly. Conservative management is recommended for smaller lesions and larger defects require skin grafts and local flaps. Newer dressing materials with epidermal growth factor may show some promise for the future.

CONFLICT OF INTEREST

Conflict of Interest declared none.

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