



Aplasia Cutis Congenita of Scalp and Back: A Rare Entity

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Abstract

Aplasia cutis congenita of scalp is a rare, life-threatening birth defect. It may present as an isolated absent scalp skin, or with a combination of absent skull and absent skin in other body regions. We report a newborn with a very rare presentation of combination aplasia cutis congenita of scalp and lumbo-sacral region and discuss the differentials and management strategies.

Introduction

Aplasia cutis congenita is a rare, life-threatening birth defect having a circumscribed area of absent skin. 80-90% cases involve the scalp in a well demarcated and non-inflammatory fashion [1]. Of these, most are superficial involving only the epidermis and dermis but some are full-thickness skin defects with absent epidermal appendages. 80% occur near the vertex and 20% are associated with an underlying cranial bone defect [2]. Rarely, other regions of the body may be involved simultaneously.

We report a newborn with a very rare presentation of aplasia cutis congenita of scalp and lumbo-sacral region.

Case Presentation

A female weighing 2.35 kgs was born by full-term vaginal delivery to a G3P2, 25 year old mother. Antenatal period was uneventful and no record of any antenatal ultrasound was available. The newborn had an 8 X 6 cms full thickness scalp defect with attenuated but intact dura, exposing the underlying sagittal sinus. The head circumference was 35 cms. Another 3 X 1.5 cms area of partial agenesis of the skin was present over the lumbo-sacral region (Figure 1A). Other systemic examination was essentially normal.

After prognostication and evaluation of surgical vs. non-surgical management options, parents chose conservative treatment with moist gauze dressings and denied any further investigations, including imaging.

Discussion

The pathophysiology of in utero skin disruption resulting in aplasia cutis congenita is yet unexplained. The hypotheses include intrauterine vascular accidents, viral pathogens, pressure and amniotic adhesions. Aplasia cutis can occur as a benign isolated defect or as part of a syndrome, sequence or association (eg. Goltz Gorlin syndrome, Tetrasomy 12p, Trisomy 13). Consanguinity, maternal drug intake and genetic abnormalities (TGF-3 β -II receptor gene, BMS 1, EOGT missense mutation) may explain the concurrent occurrence in scalp with other regions of the body [3]. Frieden's classification for aplasia cutis congenita is based on the number and location of the lesions and the presence or absence of associated malformations and consists of 9 groups [4]. Mortality in cases of aplasia cutis congenita has been reported in 12 – 55% and it usually occurs during the first week of life [5]. Of these, nearly 20% occur from bleeding. The exposed dura quickly becomes dry and desiccated, developing cracks which may spread into major dural veins causing exsanguinating bleed [6]. Dural defects may also cause herniation of brain with mechanical injury. Local infection, meningitis, sepsis, CSF leak and thrombosis of superior sagittal sinus are the other challenges for survival [3].

The treatment of this rare condition is controversial. Non-surgical management is preferred by many for small defects with intact calvarium. This entails keeping the dura continuously moist with either gauze dressing and saline drips or antiseptic ointments. The epithelialization is very gradual and may take several months, during which the patient is at continued risk for complications [3].

Surgical management includes full-thickness rotational flaps covering the scalp defect in the

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Figure 1: (A). Aplasia cutis congenita of scalp (white arrow) and lumbo-sacral region (red arrow). (B). Anencephaly with absent scalp, skull and forebrain (blue arrow) (Different patient).

first 24 hrs of life. Such aggressive intervention has been shown to minimize hospital stay by reducing the time to secure wound closure. It is therefore cost-effective with a low rate of complications and appealing functional and aesthetic outcomes. Underlying dural defect, when present, requires simultaneous closure with periosteal flaps. However, calvarial defects do not always warrant a bone graft as early scalp closure prompts ossification. Persistent bone defects beyond 3-4 years age may need cranial reconstruction. Composite closure of defects using bone and skin grafts in a single-stage has also been described, but without much success.

Hydrocephalus is present in only a small percentage of patients with aplasia cutis congenita. But intracranial hypertension may arise from tight closure of dural or scalp defects or even a tight bandage post-operatively. Growing brain of the child may exert undue force on the undersurface of the flaps, causing delayed wound breakdowns [3].

Aplasia cutis of the scalp can rarely be associated with lesions on other parts of the body like face, trunk or limbs. To the best of our knowledge, the association of a scalp with lumbo-sacral lesion has not been previously described. These skin defects present in other parts of the body are known to heal by spontaneous contraction and epithelialization. However, a small percentage may have large defects requiring primary closure with regional flaps or using tissue expanding techniques [3]. A multi-disciplinary approach involving neurosurgeons, pediatric surgeons and plastic surgeons is therefore important in the care of these children.

Aplasia cutis congenita of the scalp must be distinguished from anencephaly which is the congenital absence of scalp, skull and forebrain (Figure 1B). A stage in the evolution of anencephaly is

exencephaly or acrania characterized by complete or partial absence of skull bones, with complete but abnormal development of brain tissue. Anencephaly results if the protruding brain of exencephaly deteriorates. However, if it persists and is covered by skin or epithelium, encephaloceles are formed. Another confusing term in the same spectrum is mero-anencephaly (meroacrania) referring to only a partial absence of brain and calvarium. Relatively small skull and scalp defects may therefore be present in either least severe forms of mero-anencephaly or most extreme forms of microcephaly with encephalocele [7].

Conclusion

The index case represents a very rare combination of aplasia cutis congenita of scalp and lumbo-sacral region. A good knowledge of the condition and treatment options is essential for optimal case management. Multi-disciplinary team approach is required for evaluation of surgical versus non-surgical treatment, with the aim of protecting the brain from trauma and achieving long term aesthetic outcomes.

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