

Familial Aplasia Cutis Congenita of the Scalp: A Case Report and Review

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Abstract

We report three cases of aplasia cutis congenita of the scalp affecting two siblings and their mother, suggesting group 1, or autosomal dominant aplasia cutis congenita not associated with multiple abnormalities. A review of the clinical features and literature concerning this heterogenous and rare condition is presented.

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Key words: Absence of skin, Autosomal dominant inheritance, Congenital, Scalp defects

Introduction

Aplasia cutis congenita (ACC) is a rare condition in which localised or widespread areas of skin are absent at birth. Diagnosis of this condition is primarily clinical, and the histologic appearances vary. Several distinct subtypes occur, characterised by the location and pattern of skin absence, the presence of associated malformations, and the mode of inheritance.¹ In most cases, ACC is confined to the scalp, although other areas of the body may be involved. There is considerable variation in size, shape and surface characteristics. We describe two siblings and their mother who have group 1 ACC, or autosomal dominant ACC affecting the scalp without multiple abnormalities. This family of 3 cases represented the only case of familial ACC amongst a total of 12 cases seen at the Middle Road Hospital and National Skin Centre from 1986 to 1996.

Case Report

A 1-year-old Chinese male was referred at the age of three months for an alopecic area in the midline of the occiput which had been present since birth. It was otherwise asymptomatic and was not increasing in size. He was born at term by vacuum-assisted vaginal delivery due to poor maternal effort. There was no antenatal or perinatal events of significance and no maternal history of ill-health or drug ingestion. The child was otherwise well, with no other medical history of note.

The patient's mother gave a history of having a similar bald patch on the scalp present since birth, and had noticed the same condition affecting her second child, a 5-year-old daughter. Her first child was unaffected. The pedigree is as shown in Figure 1. Neither the patient's

mother or her affected sibling had any medical or obstetric history of note, and both have been in good health.

Physical examination of the patient showed a brownish alopecic area measuring 0.8 by 0.8 cm located over the midline of the scalp in the occipital area. This was covered with overlying scar tissue, and the hair surrounding this area was noted to be thicker and coarser. The child also had a portwine stain surrounding this area (Fig. 2). The patient's mother had a similar defect located over the same area measuring 0.5 by 0.8 cm (Fig. 3). The patient's affected older sister was noted to have a similar alopecic patch with overlying scar tissue formation over the occiput as well (Fig. 4). All three were otherwise well, with no other abnormalities on clinical examination.

The diagnosis was explained to the mother and no further treatment was advocated as the defects had healed.

Discussion

Aplasia cutis congenita or congenital absence of skin is a heterogenous group of disorders. It is an uncommon condition. Diagnosis is primarily clinical, and it is characterised by solitary or multiple, sharply margined, raw defects. A recent review of 12 cases of ACC seen at the Middle Road Hospital and National Skin Centre in Singapore over an 11-year period from 1986 to 1996 showed that the scalp was the commonest site of involvement, occurring in 8 of the cases. This family represented the only case of familial ACC seen. There is scant literature on ACC from Southeast Asia, and the dozen cases reported in the review in Singapore do not represent the true incidence or frequency of the disease

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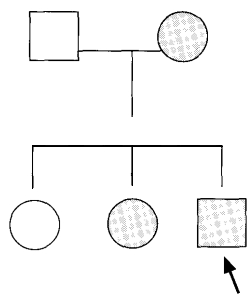


Fig. 1. Pedigree.



Fig. 2. Midline occipital scalp defect with surrounding coarse hairs.



Fig. 3. Similar midline occipital scalp defect in patient's mother.

locally, as there are cases which remain undetected because the lesions are asymptomatic or regarded as a minor developmental abnormality and ignored. The aetiology remains unknown.

In Demmel's² review of ACC, the scalp was the commonest site of involvement, with nearly 86% of the solitary lesions occurring there. Lesions may be ulcerated, bullous, cicatricial or covered with a tough, translucent membrane and occasionally extend to the bone or dura. These defects may also be circular, elongate, stellate, or triangular in shape, and of variable depth.⁴ Less often, other body parts are involved, with or without associated defects. These rare lesions of the trunk and limbs are often extensive and usually symmetric.⁴

The classification proposed by Frieden¹ divides the



Fig. 4. Midline occipital scalp defect in patient's older sister.

TABLE I: CLASSIFICATION OF ACC ACCORDING TO FRIEDEN¹

Group	Definition	Inheritance
1	Scalp involvement without other multiple abnormalities; may be associated with single abnormalities, e.g. cleft lip/palate, mental retardation or patent ductus arteriosus	AD or sporadic
2	Scalp ACC with limb abnormalities, e.g. limb reduction defects, syndactyly	AD
3	Scalp ACC with epidermal/organoid naevi; e.g. naevus sebaceous	Sporadic
4	ACC overlying embryological malformations; any site can be involved e.g. spina bifida	Depends on condition
5	ACC with placental infarcts	Sporadic
6	ACC associated with epidermolysis bullosa	AD or AR
7	ACC localised to extremities without blistering; usually pretibial areas and dorsum of hands and feet	AD or AR
8	ACC associated with teratogens, e.g. varicella and herpes simplex infections, methimazole	Not inherited
9	ACC associated with malformation syndromes; e.g. trisomy 13, ectodermal dysplasias	Varies depending on syndrome

ACC: aplasia cutis congenita; AD: autosomal dominant; AR: autosomal recessive

group of ACC into 9 subtypes (Table I). Our patient has group 1 ACC. By definition, this does not include cases of ACC with multiple abnormalities, but several isolated abnormalities have been reported. These include cleft lip and palate in association with occipital scalp ACC,⁵ transoesophageal fistula in a case of temporal ACC,⁵ mental retardation,⁶ and patent ductus arteriosus associated with scalp defects.⁷

To our knowledge, this is the first case of familial ACC reported locally. In 1922, Goldsmith⁸ described a family with ACC and underlying defects in the parietal skull bones affecting 16 members in 5 generations. Itin and Pletscher⁹ reviewed the literature on familial ACC in 1988 and found a dozen case reports of familial ACC,

some of which extended over 5 generations. To these, they added a family with ACC affecting 6 members over 3 generations. Most of the cases were inherited in an autosomal dominant fashion, although there were a few cases which demonstrated autosomal recessive inheritance. These cases were all of European origin.

The cause of familial ACC is a subject of much speculation. The scalp defects are probably not attributable to a single cause, and there is no unifying theory for its pathogenesis. ACC is a physical finding denoting that disruption of skin development has occurred in-utero. The causes of this disruption vary, and include compromised vasculature to the skin, trauma, teratogens, and in the case of familial ACC, genetic factors.

The size of our patient's scalp defect measured 0.64 cm², but such defects can range from 0.5 cm² to an area as large as 100 cm². The larger defects tend to be deeper and are more often associated with haemorrhage, venous thrombosis, and meningitis.⁴ Management of ACC is dependent on the extent of the defects. Surgical treatment is required to close large defects and prevention of skin infections is important in the early stage. All cases of ACC should have a detailed obstetric and family history obtained, as well as a thorough physical examination and screening of family members.

The physical examination should be geared towards the ectodermal structures, looking for abnormalities of the hair, teeth and other areas of the skin as well as the central nervous system.

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