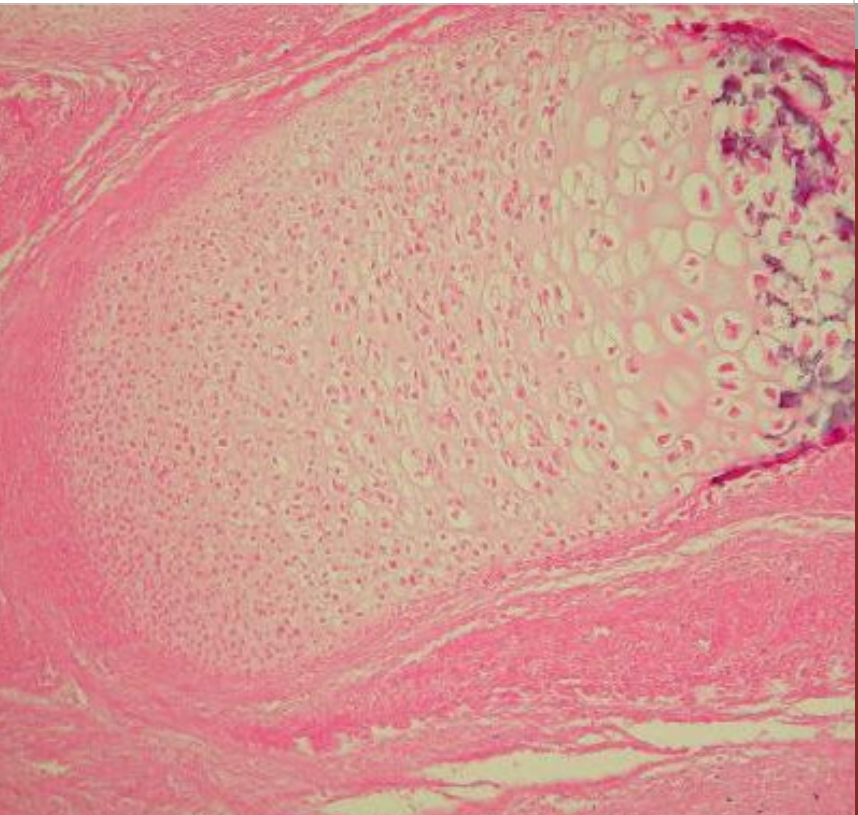
SWISS SOCIETY OF NEONATOLOGY

Aplasia cutis congenita in a surviving twin



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INTRODUCTION

Aplasia cutis congenita (ACC) is an uncommon congenital defect of the integument, usually affecting the scalp but it can involve the face, trunk and extremities. Most cases are sporadic but a familial autosomal dominant pattern of inheritance also occurs. It has also been described in association with twin pregnancies, in which one twin died during early pregnancy. We present such a case, where the surviving twin was born with multiple skin lesions.

CASE REPORT

A male infant was born at 37 4/7 by elective Cesarean section to a 31-year-old G3/P3. He was admitted to our unit with multiple skin defects. Monozygotic (chorionicity unclear) twin pregnancy was diagnosed early, but twin B died between 11 and 15 weeks of gestation. Pregnancy was otherwise uneventful and the family history was unremarkable. Apgar scores were 9, 10 and 10 at 1, 5 and 10 minutes, respectively, and arterial cord pH was 7.31.

Remnants of the twin (fetus papyraceus) were found in the placental membrane. An island of immature chondrocytes belonging to the skeleton was surrounded by dense fibrous stroma (Fig. 1).

Birth weight was 2710 g (P10-50), length 46.5 cm (P10-50), head circumference 34.3 cm (P50-90). Symmetrical skin defects were noted in the flank and

scapulae areas and a single right-sieded parieto-occipital defect was noted (Fig. 2-4). There were no limb defects and, apart from a cranial ultrasound demonstrating two small choroid plexus cysts, no other anomalies were noted.

After consultation with dermatology and pediatric surgery, a clinical diagnosis of aplasia cutis congenita (ACC) was made and conservative management was recommended. With treatment consisting of antibacterial ointment and dry dressings, the defects began to heal and epithelialize. The baby was discharged home on the sixth day of life and was seen at the dermatology clinic for a follow-up visit 2 weeks later. After daily application of ointment, the skin lesions showed continuous scarring, without functional impairment (Fig. 5).

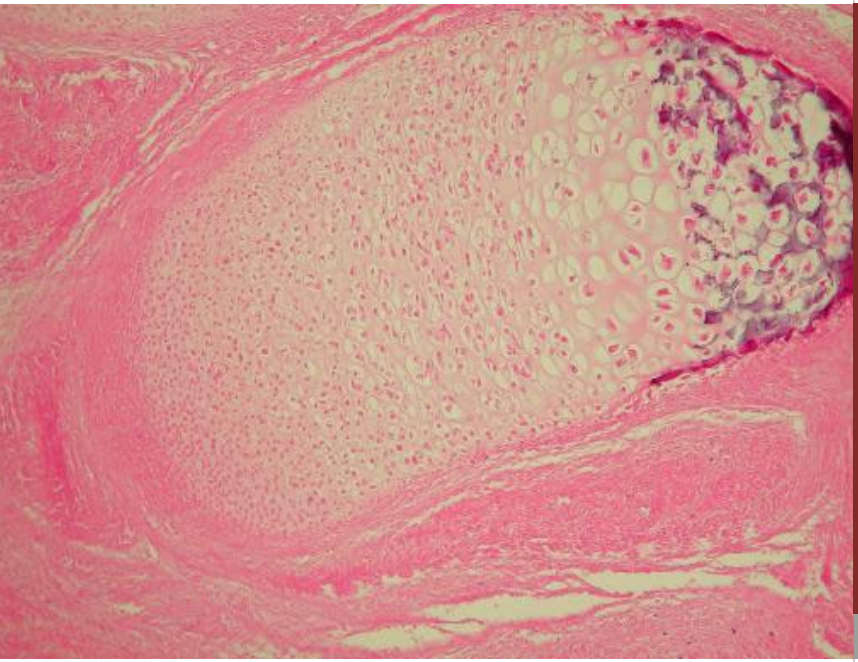

Fig. 1

Fetus papyraceus (H&E, original magnification: x20).

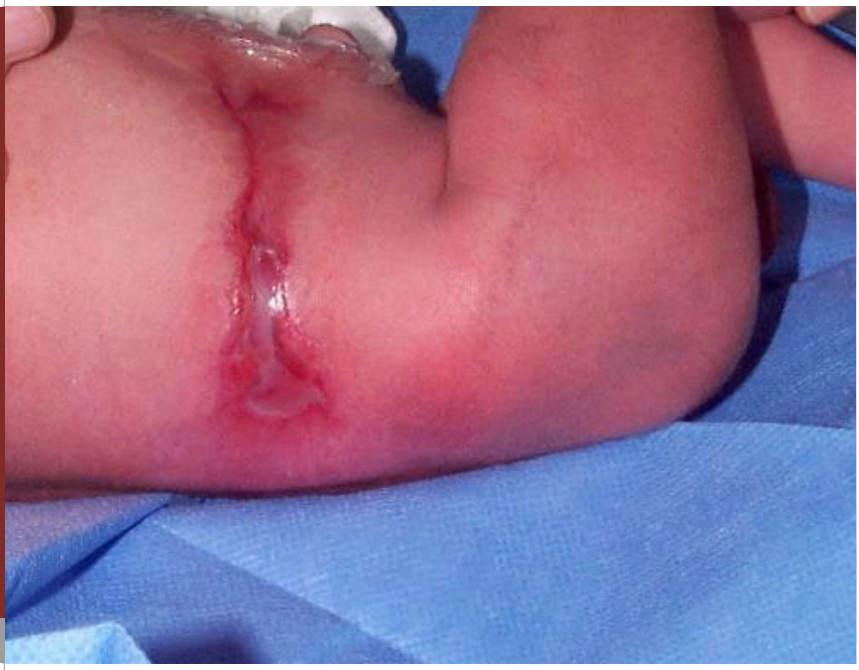

Fig. 2

Skin defect over right flank region.

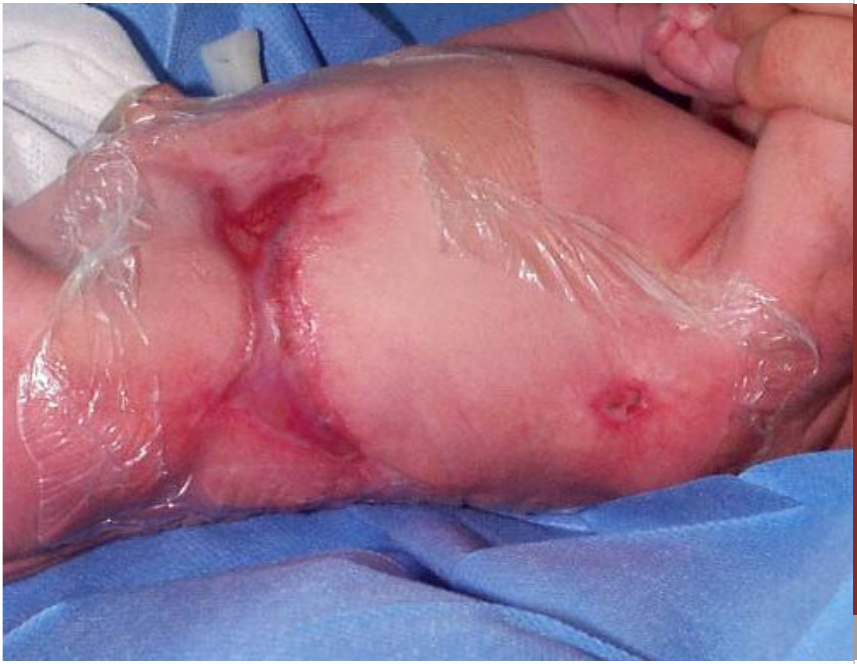

Fig. 3

Skin defect over left flank region.

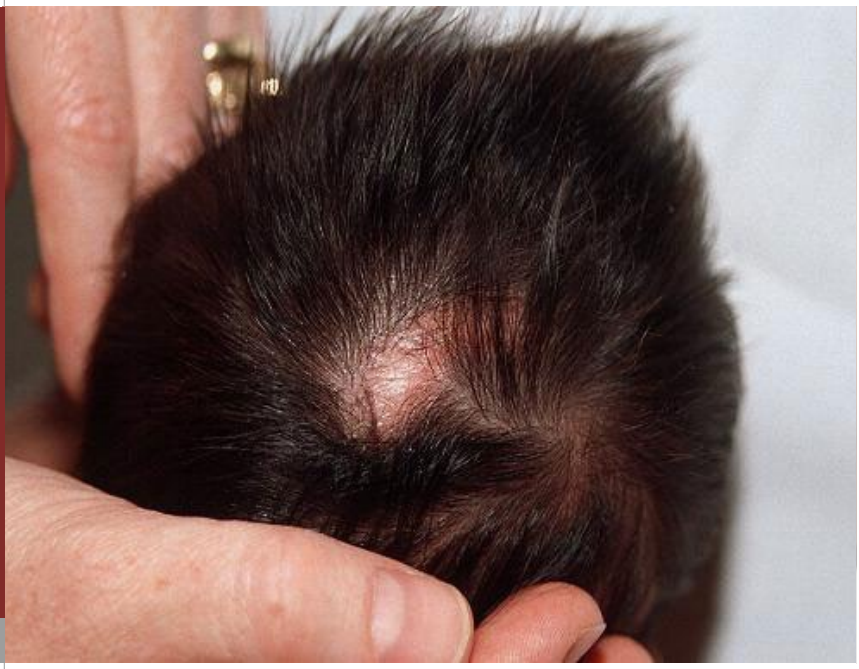

Fig. 4

Parieto-occipital scalp defect.

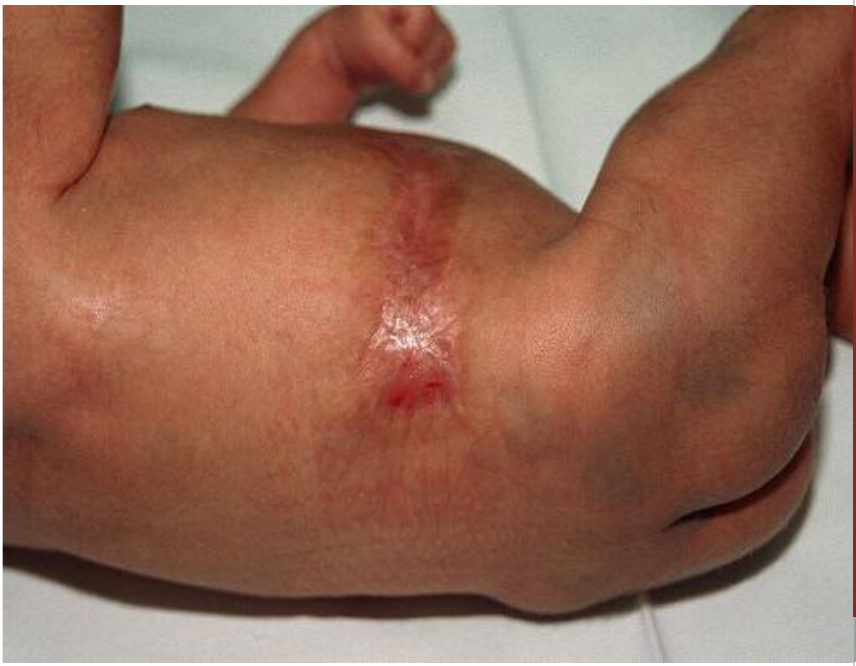

Fig. 5

Healing skin defect.

DISCUSSION

ACC represents a heterogeneous group of disorders with a focal absence of skin being the common characteristic. Frieden (1,2) and Sybert (3) have each proposed a classification of the disease according to inheritance, location of the lesions and associated anomalies. Most cases are sporadic but familial cases occur with either autosomal dominant or autosomal recessive inheritance patterns.

Many of the sporadic cases have been associated with placental infarcts or twin pregnancies, in which one twin died during the late first or early second trimester (1 – 4). The majority of the twins were monozygotic but dizygotic twins with fused placentas have also been described, supporting the theory that the pathogenesis is related to vascular disruption (5,6). Several complications in the surviving twin are known to be associated with the death of the co-twin in utero and their manifestations appear to be related to the gestational age at the time of death. Death during early pregnancy is usually associated with ischemic complications manifesting as absence of structure, atresia or hypoplasia whereas mortality in the late second or third trimester tends to be associated with embolic phenomena and coagulopathy, leading for example to neurological problems in the survivor (1,4).

The skin lesions, which may be single (70%), double (20%) or triple and more (8%) (2,7), as in our patient, are most commonly localized on the scalp (86%), but

can be seen on the trunk and extremities with a symmetrical distribution (1 – 3,7). The defect varies from a sharply demarcated weeping or granulating ulceration to an area of erosion covered by a thin friable membrane.

The most common associated anomalies are limb anomalies and include amelia, syndactyly, limb reduction defects, polydactytly, Volkmann's ischemic contracture and clubbing of the hands and feet. Bowel atresia may also be associated. Other less common associations include cleft lip and palate, porencephaly, hydrocephalus, neural tube defects, epidermolysis bullosa, ectodermal dyplasias and chromosomal anomalies such as trisomy 13 (1 – 3,7).

In the presence of a fetus papyraceus, maternal serum alphafetoprotein and amniotic fetoprotein may be elevated prenatally and amniotic acetylcholinesterase activity may be increased (8,9). However, the diagnosis is usually made at birth and is based on clinical criteria.

Histologically, there is a complete absence of the epidermis, absence or paucity of epidermal appendageal structures and variable loss of dermis and other subcutaneous tissue. In severe defects bone and dura may be absent (2,7).

Treatment is usually conservative and aimed at preventing further trauma and secondary infection. As

the lesion heals, it is replaced by grey hairless scar tissue characterized by the absence of all epidermal appendages. Most lesions heal within the first few months of life. Skin and bone grafts may be required for the reconstruction of major defects (7).

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