Aplasia Cutis Congenita

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Abstract

Aplasia cutis congenita (ACC) is the name given to a condition characterized by a heterogeneous group of diseases which have in common a focal absence of the skin. This rare anomaly can involve the epidermis, dermis, or at times the subcutaneous tissue. We are reporting three children with ACC along with a brief review of similar reports from Saudi Arabia and the neighboring gulf states.

Case Reports

Case No. 1

A 42-year-old woman, gravida 18 and para 10 with a history of 7 abortions developed eclampsia. She had history of hypertension which was being treated orally with alphamethyldopa. A caesarian section was performed and full-term boy weighing 2.2kg was delivered. The child had a trigonal head, microphthalmia, malformed low-set ears, cleft lip and palate, and polydactyly of the fingers and toes. Bilateral cryptorchidism was present. A3 cm x 2cm ulcer was seen on the vertex (Fig. 1) . The child possibly conformed to Group 9 in the classification proposed by Frieden . No further examination or investigations could be fone as the baby died after 2 weeks due to disseminated intravscular coagulation.

Case No. 2

A 25-Year-old primigravida delivered a preterm child ate 24 weeks weighing 0.5kg. The child was kept on a ventilator due to respiratory distress syndrome. Symmetrical denuded areas were seen over the extremities (Fig. 2) and the child was classified ad Group 8 . On the third day the baby died due to intracranial hemorrhage.

Case No. 3

A 39-year-old woman, gravida 9 and para 8 delivered a normal female child with 2 nodules on the right parietal area showing the hair-collar sign (Fig. 3 & 4) . MRI was done and this revealed intracranial extension. The child was placed in group 4 of friden classification. The child was referred to a neurosurgeon for further management.

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Discussion

ACC is most often an isolated defect, but it can be associated with other physical anomalies. The heterogeneity in the extent of involvement, mode of inheritance and associated abnormalities forms the basis of the 9-group classification in ACC1.

The defects that usually affect the vertex may form early in gestation and heal before delivery leaving an alopecic scar, or may present at birth as ulcerations. The defects may however, involve any part of the skin surface. The depth of the lesions is often limited to the dermis or subcutaneous area; rarely it may extend to the periosteum, skull and dura. ACC can be associated with a number of other physical anomalies and malformation syndromes which is the reason why no classification has to date been found to be satisfactory. From the review of previous reports it has been seen that 78 per cent of patients had defects of lower limbs and 59 per cent had defects of the upper limbs2.A number of etiologic factors have been found to give rise to ACC accounting for its phenotypic heterogeneity1,3.ACC may be sporadic, autosomal dominant or recessively inherited, or result from trauma or vascular causes. Amniotic bands resulting from early rupture of the amniotic membrane have also given rise to ACC4. A teratogenic effect following exposure to antithyroid drugs has been implicated in ACC5.

Eight cases of ACC have been reported from the Gulf region. In one it was limited to the scalp6; in two others the underlying bone was affected7, and in one the scalp lesion with deficiency of bone was associated with dysmorphic facies, anophthalmia, dextrocardia and distal limb anomaly8, in two ACC presented as a defect on the dorsa of the feet and abdominal wall respectively7, and in one the ulcers involved the chest wall, and the upper and lower limbs9 , in all these reports the antenatal history was stated to be unremarkable. Both routine histopathology and electronmicroscopy done in one patient9 had revealed nothing specific for ACC as mentioned in the literature. Our patients too had diverse manifestations. One had marked deformities involving the face and extremities along with cryptorchidism and in the second it was limited to the extremities. The former may have been trisomy 13 but no chromosomal study could be arranged as the child died shortly after birth. In the last patient the hypertrichotic rim referred to as the hair collar sign, which was reported before in the mem-

branous variant of aplasia cutis, was seen around the lesion. No specific risk factor for ACC could be identified in any of our patients except for the fact that the one with multiple defects was delivered by caesarian section as the mother had eclampsia and a history of hypertension suggesting a vascular cause.

Treatment and survival in ACC depends largely on the extent of defects and the type of deformity. We feel that patients may go undiagnosed and unreported due to great diversity in the manifestations of ACC.

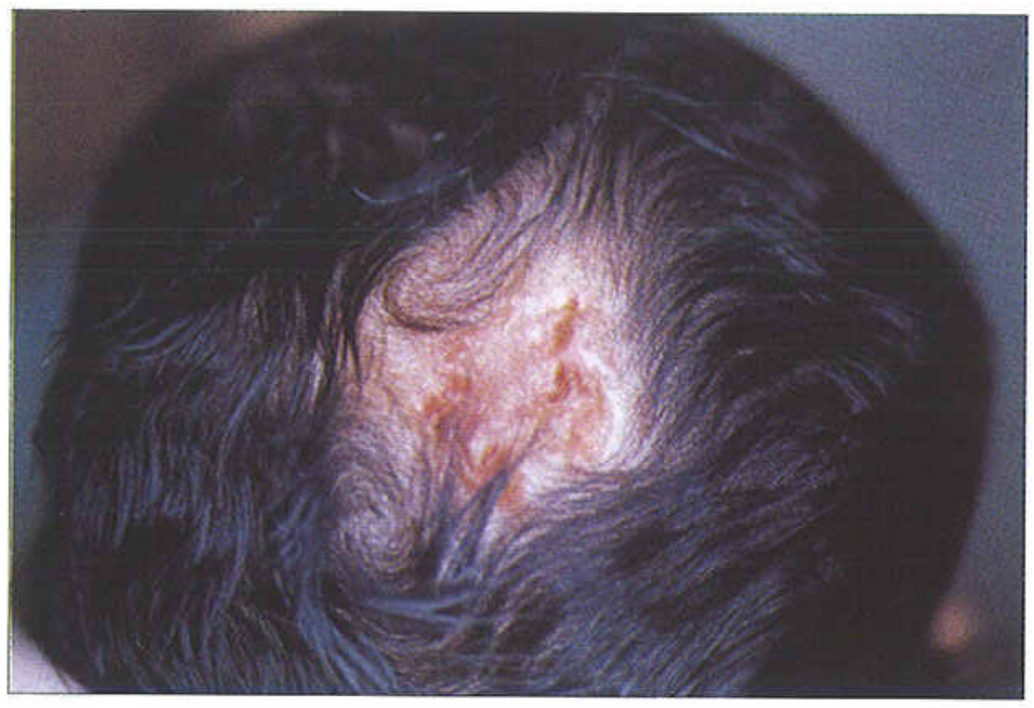

Fig.1 : Aplasia Cutis congenita in case 1

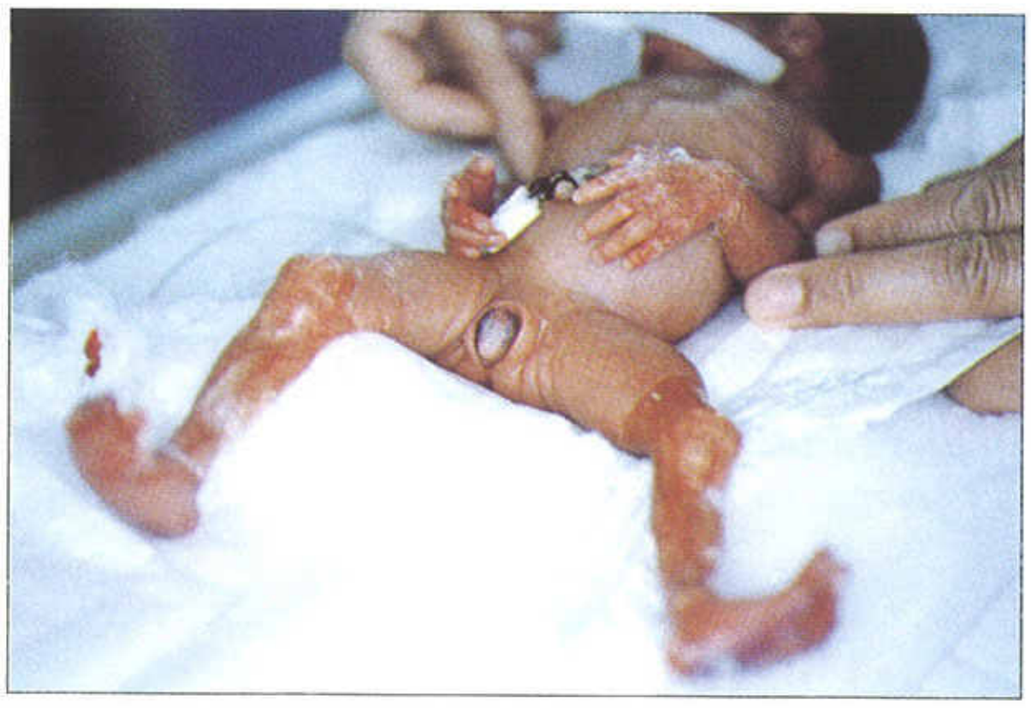

Fig.2 : Aplasia Cutis congenita in case 2

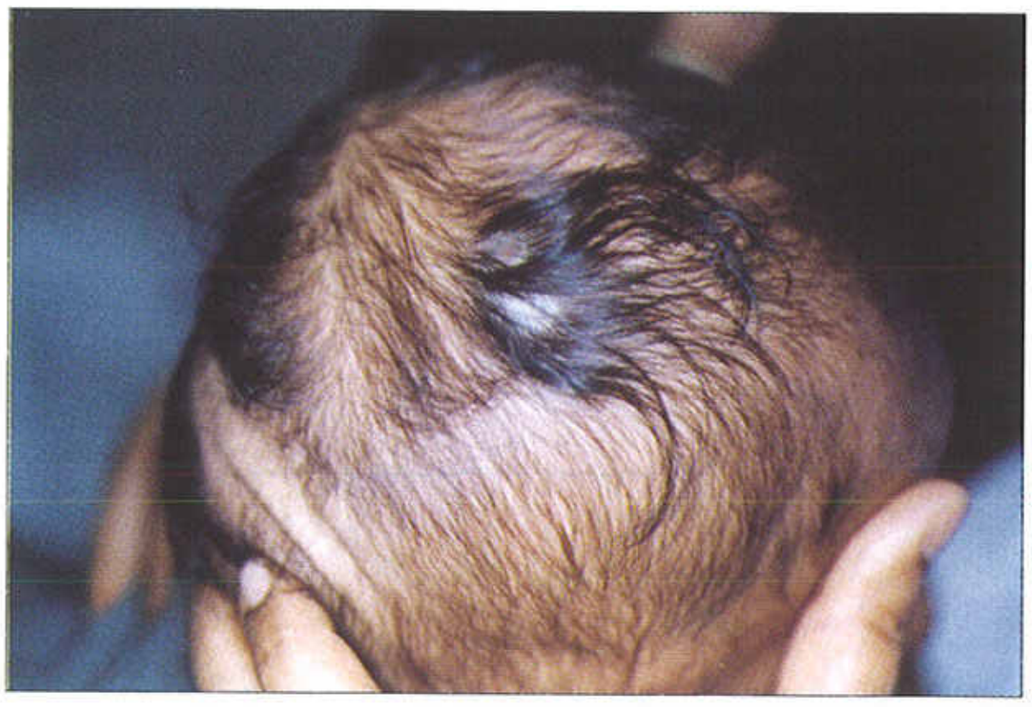

Fig.3 : Aplasia cutis congarita and hair collor sign case 3

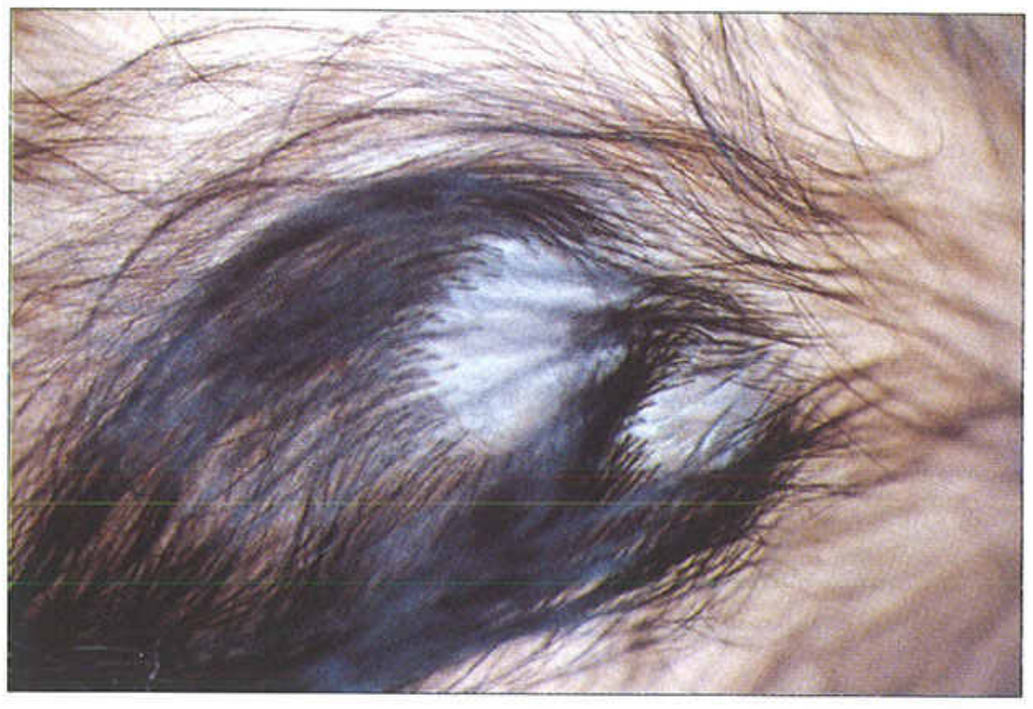

Fig.4 : Aplasia catis congenita and hair callar sign (Close up view in case No. 4)

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