

## APLASIA CUTIS CONGENITA: A REPORT OF FIVE CASES IN RIYADH

Farooq A. Bhat, MCh; Bhagwat S. Mathur, MCh, FACS;  
M.T. Hussain, MS, FRCS; Fawaz Khalil, FRCS

Aplasia cutis congenita is a rare malformation characterized by the absence of skin. We report five cases of the condition, three of them with the classical localization of the lesions in the scalp, the fourth case involving the lower extremities, and the fifth involving the trunk. A review of the literature is also presented.

### Case 1

A male baby presented with a congenital defect in the scalp. This baby was born after a full-term delivery with an insignificant antenatal history. On examination, there was an absence of scalp and underlying bone covering an area of 15 x 8 cm in the center of the scalp (vertex) (Figure 1). The defect was covered with an eschar. After routine investigations, x-ray of the skull and CT scan of the brain, the baby was operated upon for excision of the eschar and coverage with rotation flaps. There was minimal bleeding from the sagittal sinus which could be controlled easily. On the fifth postoperative day, the tips of the flaps started to get discolored. The baby was managed conservatively with dressings twice a day. After two weeks, there was a small raw area in the center of the flaps about 2 cm in size, with evidence of dural leak. The baby was put on antibiotics and the area healed within a week (Figure 2). On monthly follow-ups the child was noted to be growing well.

### Case 2

A female baby was referred from the Obstetrics Department with a congenital defect in the scalp. She was born after a full-term normal delivery with an insignificant antenatal history. On examination there was a defect in the scalp 2 x 2 cm in the occipital area. There was a thin white membrane (durameter) covering the defect which was not disturbed during examination. Margins of the defect showed deficiency of the underlying bone. Routine

possibility of a later cranioplasty, was explained to the parents.

### Case 3

This baby was referred from the Obstetrics Department with a defect in the anterior part of the scalp. It was a full thickness skin defect with a normal underlying bone measuring 5 x 3 cm in size. The defect was covered with a split-skin graft and followed up in the outpatients department. The possibility of a later excision of the scar and tissue expansion was explained to the parents.

### Case 4

This baby presented with absence of skin on the dorsum of feet. It was a bilateral defect. The defect was covered with a skin graft.

### Case 5

This baby was referred from the Pediatrics Department with a huge defect in the abdominal wall, both anteriorly and posteriorly. It was a full-term normal delivery with insignificant antenatal history. The defect was covered with split-skin graft.

### Discussion

Aplasia cutis congenita, a condition characterized by the absence of a portion of skin, and occasionally of deeper layers, was first reported by Campbell,<sup>1</sup> describing the condition as congenital ulcers of the scalp in two siblings, one of whom died from sagittal sinus hemorrhage and the other of hydrocephalus. To date, more than 500 cases have been reported in the literature. The anomaly seems to be more common in girls, primogenitures and premature neonates, and is not infrequently associated with other congenital defects. Both intrauterine and genetic factors have been implicated in the etiology of this condition.

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From the Department of Plastic Surgery, Riyadh Medical Complex, Riyadh, Saudi Arabia.

Address reprint requests and correspondence to Dr. Mathur: P.O. Box 7914, Riyadh, Saudi Arabia.

Accepted for publication 30 November 1997. Received 21 May 1997. investigations, x-ray of the skull and CT scan of the brain were done confirming the bone defect. The patient was operated upon and the defect closed primarily. The

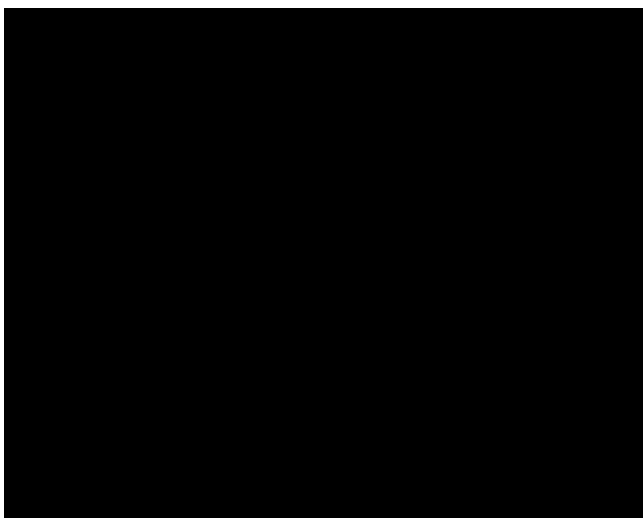


FIGURE 1. Case 1. Preoperative.

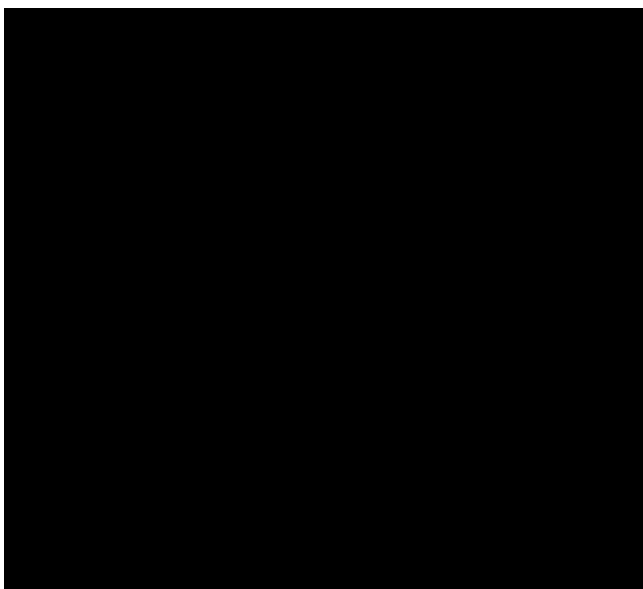


FIGURE 2. Case 1. Postoperative.

Among the intrauterine factors, amniotic adhesions and bands have been implicated as the most likely cause.<sup>2,3</sup> The scalp variety of ectopia cutis congenita is thought to be an autosomal dominant gene defect,<sup>4,6</sup> but recent evidence

suggests primary failure of differentiation. At birth, a history of obstetrical trauma or forceps delivery must be ruled out before labelling this condition. Congenital syphilis must also be excluded. A scan of the brain is useful in detecting any associated intracranial defects, such as hydrocephalus. Clinically, the neonate presents one or more sharply circumscribed surface areas where skin has failed to develop. The defect is usually small and solitary. But extensive defects have been reported resulting in early deaths.<sup>5,6</sup> The scalp is involved in 60%-70% of the cases, followed by limbs in about 20%. In the scalp, these defects usually occur over the posterior fontanelle astride the midline. Observed at birth, they are sharply demarcated and appear to be covered by a transparent membrane. After birth, the very thin membrane is easily disrupted, resulting in hemorrhage that requires hemostatic dressings. If the lesion is very small and superficial, the resulting ulceration will heal with cicatrix or keloid formation. Although the defect most frequently involves only the epidermis and dermis, there may be an associated periosteal and osseous defect underlying the lesion. Defects involving the extremities are usually symmetrical and involve only the skin as a rule. Occasionally, however, they involve the deeper structures and even the underlying muscle. Associated terminal transverse defects (Adam-Oliver syndrome) have been described.<sup>7</sup> The mainstay of treatment is control of infection and hemorrhage. Small defects may be left to epithelialize or be covered with split-skin graft. Large defects need a local flap and later treatment of scars and alopecia.

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