Congenital scalp defects: aplasia cutis congenita

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The authors discuss the rare lesion of congenital scalp defect, both partial and full thickness. The majority occur in children who are otherwise normal, but a few are seen in children who have many concomitant anomalies. A high percentage of the multiple anomaly cases were found to have trisomy 13-15. Etiology of these lesions is not clear, but there seems to be an inherited component. Treatment of otherwise normal children is conservative for the smaller lesions, with excision and primary closure when possible for the larger ones.

Keywords: congenital scalp defects, aplasia cutis congenita, congenital anomalies

Partial agenesis or neonatal ulceration of the skull and scalp is rare. These defects have been variously designated as congenital ulcer of the newborn, Streeter's spots, and aplasia cutis congenita. They most commonly occur in the midline, but parietal and posterior auricular lesions also occur. Lesions vary in size from pinpoints to over 8 cm in diameter and may be single or multiple. In about 10% of cases there is an associated skin defect elsewhere. The lesion may be described as a moist ulcerated defect with a base varying in depth from subcutaneous to the level of the arachnoid. They may occur in infants who are otherwise normal but also may accompany multiple congenital anomalies. Occurrence of the defects in siblings and in several generations of the same family suggests an inherited component.

This lesion was first reported by Campbell in 1826 as a congenital ulcer of the cranium. His patient died shortly after birth because of fatal hemorrhage from the superior sagittal sinus. Since then there have been about 250 cases reported. In the past few years we have seen 12 such patients.

Summary of Cases
We classified our 12 patients into two groups based on the layers involved. Group 1 patients had a partial thickness defect involving the scalp only, Group 2 patients had a full thickness absence of scalp, peristeum, skull, and dura.

Partial Thickness Defect
Case 1. This newborn white girl was the product of a full-term uncomplicated delivery (Fig. 1). She had five scalp defects ranging in diameter from 0.5 to 1 cm in the midline and biparietal regions. Neurological examination and skull films were normal. There were no other congenital anomalies. The two larger lesions were treated by full thickness excision.
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FIG. 1. Five defects ranging in diameter from 0.5 to 1 cm in newborn child. The two larger lesions were treated by full thickness excision of the defect and underlying galea followed by primary closure.

FIG. 2. Identical bilateral defects in one of identical twin girls. Treatment was by excision.

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Cases 2 and 3. These baby girls were identical twins, admitted because of bilateral parietal defects 1 cm from the midline, and 0.5 cm in diameter (Fig. 2). Neurological examination and skull films were normal. There were no associated congenital anomalies. Treatment was by excision and closure.

Case 4. This white baby boy had two defects; one parietal measuring 1.5 × 1 cm, and the other postauricular and about 2 mm in diameter. Neurological examination and skull films were normal. There were no associated anomalies. The larger lesion was excised; the smaller granulated and healed uneventfully.

Case 5. This newborn white girl had a 2 × 3 cm defect in the occipital area. Skull films and neurological examination were normal. She also had a cleft lip and palate. Treatment was by excision and primary closure.

Case 6. This newborn girl had trisomy D syndrome with a large vertex defect measuring 3 × 5 cm. She also had a midline cleft lip and palate, polydactyly, bilateral colobomas, holoprosencephaly, congenital heart disease, and renal anomalies. She died shortly after admission (Fig. 3).

Case 7. This newborn white girl had several small midline defects in the parietal area. She was a microcephalic child with hypotelorism, bilateral colobomas, and bilateral simian creases. Chromosome analysis demonstrated trisomy D. Treatment was by dressing changes.

Case 8. This 2-year-old black girl had been born with multiple lesions of the scalp. She also was severely retarded, and had a lumbar-sacral lipomyelomeningocele, malrotation of the colon, bilateral corneal opacities, congenital glaucoma, renal anomalies, and congenital heart disease. Chromosome analysis demonstrated trisomy D. The lesions had granulated in and healed spontaneously.

Group 2: Full Thickness Defect

Case 9. This newborn white girl had a 3 × 5 cm lesion in the midline-midparietal area (Fig. 4). Sagittal sinus and cortex were visible through the arachnoid. The neurological examination was normal. There were no associated anomalies. Treatment was by mobilization of the skin and primary closure.

Case 10. This newborn white boy had a 3 × 5 × 6 cm triangular-shaped defect at the vertex which when first seen was almost completely covered with a thick scar (Fig. 5). There was a large venous bleb just off the midline. Neurological examination was normal; there were no other congenital anomalies. Because of the thickness of the eschar, this lesion was initially treated with dressings. However, when bleeding occurred from the venous bleb, the child was taken to the operating room and the lesion was repaired. Surgicel was placed over the bleb and a periosteal flap swung over this. Skin flaps were then rotated to cover the defect.

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Fig 3. Group 1 defect. A large 3 × 5 cm defect in the vertex of the skull of a child with trisomy D syndrome.

Fig 4. Group 2 full thickness defect. The sagittal sinus and cortex are visible through the arachnoid.

Case 11. This newborn white boy had multiple congenital anomalies, including trisomy D shown by chromosome analysis. There was an irregular horseshoe-shaped defect in the midline-midparietal area with cortex visible through the arachnoid (Fig. 6). Associated anomalies included choanal atresia, micrognathia, and supranumerary digits on both hands and the left foot. Treatment was with dressings. The child died shortly after birth.

Case 12. This newborn white boy had a 6-cm horseshoe-shaped defect in the midline. Cerebral cortex was visible under the arachnoid. The child also had bilateral congenital hip dislocation, choanal atresia, ambiguous genitalia, imperforate anus, and laryngomalacia. Treatment was with dressings only, and the child died.

Discussion

The majority of our cases fell into our Group 1 classification. There was a slight predominance of females (7:5), as reported by others.4,21 These lesions do occur in children who are otherwise normal and have no demonstrable brain defect.3 However, they are also found in patients who are severely retarded with multiple concomitant anomalies. We have seen cleft lip and palate, polydactyly, colobomas, congenital heart disease, holoprosencephaly, simian creases, lipomyelomeningocele, glaucoma, renal anomalies, choanal atresia, ambiguous genitalia, and laryngomalacia. Four of our 12 cases had trisomy 13–15. Agenesis of the skin in remote locations occurs in about 10% of some reported series.9,18,20 We have seen none.

Histological Appearance

Microscopic section of the lesions excised in Group 1 showed that none contained the stratified squamous epithelium characteristic of scalp. The outer surface was covered by a layer of flat, thin cells. The remainder of the lesion was a collagenous matrix with signs of hemorrhage. There were no flat cells, inflammatory cells, nor skin appendages, such as hair follicles or sebaceous glands.

Etiology

Many theories have been proposed for the etiology of this lesion. Hoffman9 in 1885 postulated that intrauterine amniotic adhesions formed between the amnion and the skull early in development. With the accumulation of amniotic fluid these bands are torn apart, leaving the skin defect. This theory seems difficult to accept. It is unlikely that such an adhesion would occur in siblings or in successive generations; it is also unlikely that the lesion would occur in the same location in most of the reported cases. For the same reasons, trauma is an unlikely explanation. Pressure necrosis of the scalp during labor has been postulated,1 but is not confirmed by microscopic studies.
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Greig postulated an arrest of midline development in these children; Walz postulated a defective closure of the neural tube. This may be an example of the neuroschistic process described by Padget in which there is incomplete healing and fusion of the nourishing mesoderm. A healthy mesoderm seems to be necessary for proper development and maintenance of the ectoderm.

Treatment

Treatment of this lesion, as in other congenital anomalies, is based on the total clinical problem. If the child is severely damaged, no surgical treatment is given; chromosome analysis is carried out routinely to aid in this decision. In patients who are felt to be normal but have large scalp defects, therapy consists of primary closure of galea and skin with excision of the lesion when possible. The smaller lesions heal spontaneously with minimal scarring and no surgical therapy is indicated.

Surgical intervention is directed to prevent hemorrhage (Case 10), to prevent infection, to protect the underlying brain, and to improve appearance. In the Group 2 lesions, hemorrhage and infection (meningitis) are the major immediate complications. Campbell's first case died from exsanguination; our Case 10 had a massive hemorrhage from the sagittal sinus, which was controlled with pressure and subsequently repaired. Infection was not a problem in our series but has been in other cases.

As suggested by others we have excised the margins of the lesion or the entire lesion when possible, and closed primarily. When a large area of bone is missing, pericranium is rotated to fill the defect. We have not used split thickness skin grafts. To avoid a large area of alopecia, hair-bearing skin is rotated over the defect. We have been able to protect the brain adequately in all necessary cases. Eleven years later, the patient in Case 9 had only a small 0.5 cm defect visible on x-ray, while the defect in Case 10 had completely filled in with bone 1 year following periosteal flap rotation. We have not seen an excessively large defect requiring a plastic cap as described by Matson.

References

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