

Clinical Note

Congenital aplasia of the scalp.

Congenital aplasia of the skin is a rare condition, it affects the scalp in more than two-thirds of the cases, and among those, 60-70% affect the occipital region.¹ Most of the cases are presented in the neonatal period. It usually occurs in approximately 1:10000 births. It is more common in females than males. Associated congenital anomalies are not rare, affecting more than one system,² among these congenital anomalies of the renal system and genital system are the most common.

We report a case of a 5 day old female, who had a large skin defect at the occipital region from birth. She was born by normal spontaneous vaginal delivery at hospital after 39 weeks of gestation, her father and mother are not relatives, not from the same tribe, but lived in the same district. Prenatally the mother had no history of drug intake, bleeding, polyhydramnios, or contraceptive intake. The parent had no history of any chronic illness. There was no history of any congenital anomalies from either sides. The lesion is a single one, triangular in shape, about 5x5x7.5 cm in dimension. The edges were odematous, hairless, raised and corrugated and it extend to the midline posteriorly. The floor of the lesion was a thin membrane, dusky red in color, with a normal underlying occipital bone. The surrounding skin was normal (Figure 1). The patient had associated bilateral deformed ears and Down's syndrome. The patient was admitted to the nursery department under incubator care. Investigation showed leukocytosis ($18.5 \times 10^3/\text{microliter}$), chest, abdominal and skull x-rays were normal, ultrasound of the abdomen and pelvic organs showed no abnormalities in the renal system and the internal genital organs. The patient was started on systemic antibiotics, with daily aseptic dressing. After 4 days



Figure 1 - Neonate with congenital aplasia of the occipital region and maldeveloped ear.

the floor of the lesion was covered by bright red granulation tissues. The leukocyte count dropped to $10.8 \times 10^3/\text{microliter}$. On the 5th day of admission, surgery was carried out; the skin was under-mined, the edges were trimmed, extension of the lesion at one side carried out and the defect closed by secondary sutures. Biopsy was taken from the floor and the skin edge, the floor showed granulation tissue with mild inflammatory cell infiltration, the skin edge showed hypoplastic dermis and epidermis with hypoplastic adnexae. The patient was discharged after 5 days, and follow-up after one month showed good looking surgical scar.

The exact cause of this defect is not known, familial cases had been reported suggesting an autosomal dominant inheritance, however the occurrence of this lesion in the off-spring of consanguineous parents suggests an autosomal recessive pattern of inheritance.^{1,2} The proximity of the lesion in the majority of the cases to the scalp hairwhorl, suggests that the scalp tension may be the cause, since this is the site of maximum tension during the period of gestation.¹ Vascular developmental anomalies and epidermolysis bullosa have also been suggested as possible causes for this skin defect.³ A few cases of congenital absence of the skin at the midline of the scalp had been reported in the off-spring of mothers who took methimazole for the treatment of hyperthyroidism during pregnancy, but still the role of maternal hyperthyroidism itself remains a possible cause.⁴ In some cases there was an associated bone defect, the larger the defect the deeper, sometimes even extending to the meninges and dura, and the patient may present with evidence of meningitis or sagittal sinus thrombosis.⁵ Non of the above suggested causes were detected in our patient. Biopsy from the lesion is very important in the differential diagnosis, the hypoplastic or total absence of the epidermal appendages remains a characteristic feature.⁶ Other lesions may simulate congenital aplasia of the scalp and must be distinguished from them, like iatrogenic scalp injury by scalp electrodes or forceps delivery, epidamolysis bullosa and facial dermal hypoplasia, the later shows mainly reduction in the thickness of the dermis so that the subcutaneous fat is situated almost immediately beneath the epidermis. Treatment of congenital aplasia of the scalp includes avoidance of trauma and secondary infection, later the defect can be closed by secondary sutures, rotation flap or skin graft can be used and this depends on the size of the lesion. Small defects have the tendency to heal spontaneously making surgical repair unnecessary, and the covered area can withstand trauma within a few months.

References

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ERRATUM

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