

# Aplasia cutis congenita

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## DESCRIPTION

A male infant was delivered via normal vaginal delivery at term gestation. No abnormalities had been identified on routine antenatal ultrasound scans. There was no significant family history of medical conditions and no maternal medication use in pregnancy. Postnatally the midwifery team noticed an abnormal area of skin on the infants' back (figure 1). The infant was reviewed by the paediatric team and diagnosed with aplasia cutis congenita (ACC).

ACC is a rare congenital condition characterised by the complete or partial absence of an area of skin and less commonly bone at the time of birth.<sup>1</sup> It most commonly affects the scalp but can also affect the face, trunk and limbs.<sup>1-3</sup>

The cause of ACC is felt to be multifactorial in aetiology. Theories associated with its development include: incomplete closure of the neural tube and embryonic fusion lines; vascular insufficiency to the skin resulting from placental insufficiency; intra-uterine infections; teratogenic agents; amniotic bands and chromosomal abnormalities.<sup>2-5</sup> Nine clinically distinct subtypes have been described, characterised by the location and pattern of skin absence, associated malformations and mode of inheritance.<sup>3,4</sup>

Treatment of ACC is dependent on the size and location of the lesion. Conservative management is preferred, allowing for gradual epithelialisation of skin and formation of a hairless, atrophic scar over several weeks. Surgical closure may be required for large or multiple defects.<sup>2</sup> The prognosis of ACC is usually excellent; however, if associated with other abnormalities or malformations, the prognosis then depends on the nature and severity of those conditions.<sup>1</sup>



**Figure 1** Aplasia cutis congenita on infants back. Tape measure included for scale.

## Learning points

- ▶ Aplasia cutis congenita is a rare congenital condition affecting newborns.
- ▶ It most commonly affects the scalp but can also affect other areas of the body as demonstrated in this case.
- ▶ Majority of cases are managed conservatively, but surgical repair may be required in larger defects.

In this case, the infant was reviewed in the outpatient clinic by the plastic surgery team at 6 weeks of age. The area of skin had completely healed with no evidence of hypertrophic scarring. No surgical intervention was required. There was some evidence of hypersensitivity noted over the area of healed skin, and the parents were advised to massage emollient creams over this area to help alleviate these symptoms. The patient will have further outpatient follow-up with the plastic surgery team to monitor his progress.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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