Short reports

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Congenital skin aplasia affecting trunk

Congenital defects of skin are rare, the scalp being the most common site. A case of bilateral and symmetrical thoraco-abdominal skin defect is reported.

Case report

A male infant was born at term, weight 2·8 kg. Birth had been uneventful. Clinical examination showed no abnormality except bilateral thoraco-abdominal skin defects, each lesion measuring 5 x 4 cm. Beneath the thoracic part of the defect could be seen ribs with partially developed intercostal muscles, and deeper still the pleura and underlying lungs on both sides were visible, while beneath the abdominal part of the defect the peritoneum with underlying viscera could be seen. There was no history of such defects in the family and the pregnancy had been normal.

A policy of conservative management was adopted, the defect being dressed with Gelonet gauze after cleaning the surrounding skin with chlorhexidine. No antibiotics were given. There was gradual epithelization of the defects (Fig. 1), which was complete in 8 weeks (Fig. 2) and the baby was then discharged from hospital.

Comment

Walker et al. (1960) and Gross, Lindemayr, and Pospisil (quoted by Walker et al., 1960) reviewed a total of 275 published cases of skin aplasia, of which only 33 had skin defects affecting the trunk. Boureau (quoted by Sukarochana, 1969) reported a newborn with a scalp defect together with bilateral thoraco-abdominal skin defects; Sukarochana (1969) and Patriarca, Manzia, and Cortesi (1972) reported two cases each of thoraco-abdominal skin defects.

The cause of these skin defects remains controversial. Abt (1917) believed that adhesion of amniotic membrane to the skin of the fetus resulted in skin ulceration, but this concept is hardly tenable in the case of bilateral symmetrical defects, though a scalp defect could be explained on such a theory. Beresford and Samman (1948) ascribed these defects to failure of development of superficial vessels. Patriarca et al. (1972) reported hereditary factors in their case; no such history was available in

(a)

(b)

Fig. 1.—(a, b) Aged 1 week, epithelization beginning.
our case. Gross et al. (quoted by Walker et al., 1960) noted the decreasing number of elastic fibres in the skin as the lesion is approached from the periphery, and suggested that during development an elastic-free skin area might be overstretched, resulting in disturbed circulation in that area and consequent maldevelopment. Duhamel (1963) has given a plausible explanation; he regards aplasia of the skin in cases such as that here described as an example of defective development of the outer layer of the body wall. Agenesis of the sternum or ribs, or of the abdominal muscles are other examples.

Most of these skin defects heal well with conservative management, and the infant does well provided there are no other congenital abnormalities.

Summary
A case of bilateral, symmetrical thoraco-abdominal skin defect is described.

REFERENCES


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Hereditary pseudo vitamin D deficiency rickets in a Pakistani infant

Hereditary pseudo vitamin D deficiency rickets (Hereditäre pseudo mangelrachitis) was first described by Prader, Illig, and Heierli in 1961. The alternative name of hypocalcaemic vitamin D resistant rickets was given by them to the condition which presents during the first 2 years of life as clinical and radiological rickets with hypocalcaemia, normal or moderately reduced serum phosphate, and often mild generalized aminoaciduria. Unlike sex-linked hypophosphataemic vitamin D resistant rickets, however, there is no response to normal doses of calciferol. Daily doses of 10,000 to 40,000 units (0.25–1 mg) calciferol are required to reverse the biochemical and radiological changes. Dent, Friedman, and Watson described the first British patient with the condition in 1968, and Fanconi and Prader reviewed the condition, together with a follow-up of their original cases, in 1969.

Case report
A boy of 6 months was admitted in May 1972 for repair of an inguinal hernia. He was the second child of first generation Pakistani immigrants who are first cousins. His father came originally from the Punjab. He was a puny, miserable baby, hypotonic with only fair head control, unable to sit up without considerable support, and with marked poverty of movement. His weight was on the 3rd centile. His body was noticeably hairy with a low hairline on the forehead, and he had marked arachnodactyly, swollen epiphyses at the wrists and ankles, and a rickety rosary. He was irritable, and shortly after admission had a generalized convulsion.
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