Congenital Scalp Defect with Distal Limb Reduction Anomalies

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Abstract. Congenital scalp defects and distal limb reduction anomalies can occur as separate entities or in combination with other anomalies. They also occur together in an apparently autosomal dominant syndrome, an example of which is described in the present paper. The literature on the subject is reviewed.

Key words: Congenital scalp effect – Limb anomalies.

Scalp defects occur as an isolated abnormality or in association with other malformations (Beresford et al., 1948; Cutlip et al., 1967; Feud et al., 1945; Hodgman et al., 1965; Johnsonbaugh et al., 1965; Lynch et al., 1970; Miller et al., 1963; Pap et al., 1970; Tisserand-Perier et al., 1953; Walker et al., 1960). Many of these reported cases were familial (Feud et al., 1945; Hodgman et al., 1965; Johnsonbaugh et al., 1965; Pap et al., 1970; Tisserand-Perier et al., 1953). The association of scalp defects with distal limb anomalies has been reported by several authors (Adams and Olivier, 1945; Burton et al., 1976; Farmer et al., 1960; Kahn et al., 1950; Scribanu et al., 1975) but so far has received little attention. Farmer (1960) described two isolated cases, whereas familial occurrence was documented in the other reports. It appears that the combination of a scalp defect with acral reduction anomalies represents a distinct genetically determined syndrome, apparently with autosomal dominant inheritance. The purpose of this paper is to call attention to this syndrome and to describe a new isolated case.

Case Report

D.G., male, is the third child in a family with three children. Family history is negative with regard to congenital malformations, neurologic abnormalities or mental handicap. Physical examination of both parents is completely normal. Diminished fetal movements were present during an otherwise normal pregnancy. Delivery, at home, occurred three weeks post-term.

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Birth weight was 2750 g. Immediately after birth a large scalp defect, with absence of the underlying bone, was noted. The parents were told that the child was anencephalic and conservative therapy was given at home. Much to the surprise of the parents the child was thrived well, his psychomotor development was normal, and spontaneous granulation and healing of the skin defect occurred within three years. Childhood and adolescence were normal and school results were excellent.

At the age of 22 years he was referred to our genetic clinic for genetic counseling. Physical examination showed a young man in perfect health (height 168 cm, weight 57 kg and head circumference 58 cm). A large scalp defect is visible on the vertex (anterior-posterior diameter 19 cm, lateral diameter 11 cm) covered by thin, atrophic skin, with absence of sweating in this area (Fig. 1). His hands and feet are small, mainly because of shortness of fingers and toes (Fig. 2 and 3). The terminal phalanges of all the fingers are rudimentary, with restricted mobility and dysplastic nails. The distal interphalangeal flexion crease is absent on the third finger of the right hand which has no nail. Both feet are short and show a slight cavus deformity. The hallucs are broad and nail-less, the right foot more severely affected than the left. The toes are stubby and a rudimentary nail is present only on the fourth toe of the left foot and the fourth and fifth toes of the right foot. Both fifth toes are proximally implanted. Cutaneous syndactyly II—III is present on the left foot and II—III—IV on the right foot. There are skin tags on the medial side of the second toe on the left and on top of the second and third toes on the right.

Radiological Investigation

Both hands show slight hypoplasia of the proximal phalanges and more pronounced shortening of the middle phalanges of fingers II to V. The broad-tipped distal phalanx of the thumb is of normal length. The terminal phalanges of all the fingers are rudimentary. There is bilateral subluxation of the hypoplastic terminal phalanges of the third fingers (Fig. 4). The proximal phalanges of toes II to V are hypoplastic and have a hour-glass appearance. The terminal phalanges of the first toe are short and broad with missing distal tufting. The fifth toe of the right foot has a small middle and terminal phalanx. Middle and terminal phalanges are absent in toes II—III—IV on the right, and toe III on the left, whereas in other toes a small bony fragment is visible (II—III—V on the left) (Fig. 5). X-ray of the skull shows a large, median bone defect of the vertex, and a smaller defect located somewhat more posteriorly (Fig. 6). No other abnormalities have been found.