Congenital scalp defects with distal limb reduction anomalies

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Scalp defects occur as an isolated abnormality or in association with other malformations, and familial cases were first reported by Feud *et al*¹ in 1945 and Tisserand-Perier² in 1953. In 1945, Adams and Oliver³ were the first to report the distinct association of scalp defects with distal limb anomalies. Later reports in the mid-1970s by Burton *et al*⁴ and Scribanu and Temtamy⁵ showed that the combination of a scalp defect with acral reduction anomalies represents a distinct, genetically determined syndrome with autosomal dominant inheritance and considerable variability in expression.

The scalp defect

Scalp defects are midline skin defects, located on the vertex or the occipital region, and frequently associated with defects of the calvarium or even the meninges. Their size may vary from a 2 to 3 mm diameter skin defect to a very extensive defect of the whole calvarium as illustrated in fig 1. In one family,⁶ the most extreme manifestation of the scalp and skull defects was a large occipital encephalocele, 3 cm in diameter, located on the vertex. In the presence of a large defect of the scalp and calvarium, perinatal and postnatal brain damage (trauma, bleeding, infection) may lead to neurological complications, for example, spasticity, hemiparesis, epilepsy, and mental retardation. These complications, however, seem to occur in less than 10% of patients.

In the first years of life spontaneous bleeding and granulation of the skin defect occur, and after the first year the defect is covered by thin, atrophic skin. In some patients, the underlying bone defect can be even larger than the skin defect. Calcification and mineralisation of the bone defect occur progressively and after some years the size of the bone defect

may be relatively much smaller than at birth (fig 2). Scalp defects can be associated with various malformations of the central nervous system, for example, asymmetrical ventricular enlargement and porencephalic cysts localised under the vertex defect (personal observation). These have been especially well documented since the advent of computerised tomography. In one of these patients the scalp defect and the underlying porencephaly were associ-

FIG 1 Large scalp defect covered by thin atrophic skin.

FIG 4 X rays of the limb reduction defects with hypoplasia of the terminal phalanges.

ated with generalised lipodystrophy and muscular pseudohypertrophy, suggesting an associated hypothalamic lesion. Moreover, in the presence of trisomy 13, the scalp defects are associated with agenesis of the corpus callosum and, more specifically, with a holoprosencephaly sequence.⁷

The distal limb anomalies

The limb anomalies mostly present as acral reduction anomalies. In affected patients, the hands and feet are relatively small because of the distal shortness of fingers and toes. The terminal phalanges of the fingers are rudimentary with restricted mobility. Nails and distal interphalangeal creases may be absent in one or more fingers (fig 3). The feet are also short and mostly show a slight cavus deformity. The reduction anomalies of the toes are similar to those present in the fingers. They are stubby and have rudimentary or absent nails. On top of the most severely affected toes skin tags may be visible (fig 3). This may lead to misclassification of this type of limb reduction anomaly as a form of congenital amputation.

The distal limb reduction defect is well illustrated on x rays (fig 4); the terminal phalanges of all fingers and toes 2 to 3 are hypoplastic or even absent. The halluces are mostly short and broad with missing distal tufting.

Although the limb anomalies are mostly minor,

the case described by Adams and Oliver³ had absent lower extremities below the midcalf region and absence of all digits and metacarpals of the right hand. This may be an indication of considerable variability in expression of this autosomal dominant syndrome which should be kept in mind when genetic counselling is given.

Two reports^{6,8} dealt with the association of congenital scalp defects and postaxial polydactyly type A. In the first report⁶ a family was described in which several members presented with the association of congenital scalp defects and postaxial polydactyly A, both with wide variability in expression. The second report of a 15 year old who presented the same association seems to confirm that the combination of both anomalies represents a new distinct entity of scalp defects and limb anomalies, inherited as an autosomal dominant trait with considerable variability in expression.

References

- Feud P, Rhodes AW, Weisz A. Hereditary skin defect in the newborn infant. *J Pediatr* 1945;27:591-4.
- Tisserand-Perier M. Transmission pendant plusieurs générations d'une aplasie cutanée circonscrite du vertex. *Bull Soc Fr Dermatol Syph* 1953;60:77-8.
- Adams FH, Oliver CP. Hereditary deformities in man. *J Hered* 1945;36:3-7.
- Burton BK, Hauser L, Nadler LJ. Congenital scalp defects with distal limb anomalies. Report of a family. *J Med Genet* 1976;13:466-8.

- ⁵ Scribanu N, Temtamy SA. The syndrome of aplasia cutis congenita with terminal transverse defects of the limbs. *J Pediatr* 1975;**87**:79–82.
- ⁶ Fryns JP, Van den Berghe H. Congenital scalp defects associated with postaxial polydactyly. *Hum Genet* 1979;**49**:217–9.
- ⁷ Miller JQ, Pickard EH, Alkan MK, *et al.* A specific congenital brain defect (arrhinencephaly) in 13–15 trisomy. *N Engl J Med* 1963;**268**:120–8.
- ⁸ Buttiens M, Fryns JP, Jonckheere P, Brouckmans-Buttiens K,

Van den Berghe H. Scalp defect associated with postaxial polydactyly: confirmation of a distinct entity with autosomal dominant inheritance. *Hum Genet* 1985;**71**:86–8.

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