

Cranioectodermal dysplasia (Sensenbrenner's syndrome)

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In 1975 Sensenbrenner *et al*¹ described a new syndrome affecting two sibs, a brother and sister, who presented with a combination of skeletal, dental, and hair anomalies. The term 'cranioectodermal dysplasia' was coined by Levin *et al*² when they provided further details about these sibs and reported the clinical findings in three additional cases. The following résumé of this rare condition is based on these five children as well as published photographs of a sixth child,³ supplemented by personal experience of one unpublished case known to the author.

Clinical features

Pertinent findings in the seven cases about which information is available are summarised in the table, from which a consistent phenotype emerges.

CRANIOFACIAL

All affected children show pronounced dolichocephaly (fig 1) with sagittal suture synostosis demonstrable in over 50% of cases. Untreated this leads to marked frontal bossing with hypotelorism (fig 2). Bilateral epicanthic folds, broad nasal bridge, anteverted nares, and everted lower lip are characteristic, as is a high arched palate. Multiple oral frenula have been observed in one child.¹

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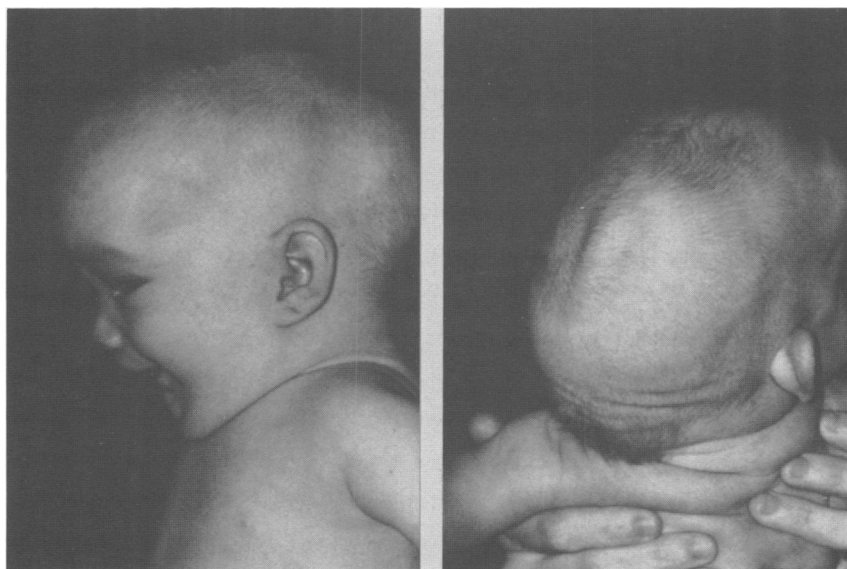


FIG 1 Lateral and posterior oblique views of a patient aged four months showing dolichocephaly and ridging of the sagittal suture.

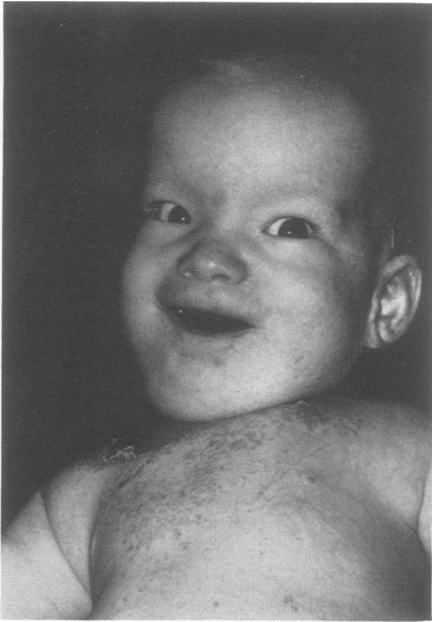


FIG 2 Facial view of the same patient aged six months showing prominence and narrowing of the forehead. Narrowing of the thorax is also apparent.

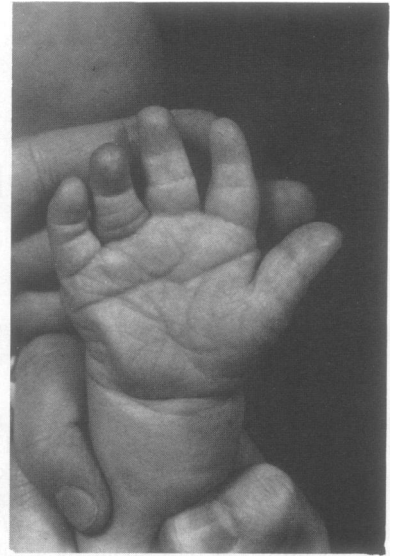


FIG 3 View of the patient's right hand showing abnormal palmar creases, brachydactyly, and fifth finger clinodactyly.

SKELETAL

The thorax is both narrow and short (fig 2) and there may be pectus excavatum. The limbs are also short, most noticeably the arms, and this is predominantly rhizomelic. The hands show brachydactyly, often with a single palmar crease and fifth finger clinodactyly (fig 3). The toes are also short and broad.

ECTODERMAL

The nails are short and stubby in keeping with the appearance of the digits, but dysplastic changes have not been reported. Scalp hair is fine, sparse, and slow growing (fig 4), and on microscopy shows absence of the central pigment core. Teeth are small and widely spaced (fig 5) and may be reduced in number. They may also show fusion and enamel dysplasia.

VISCERAL

Congenital heart disease consisting of a bicuspid aortic valve and possible cor triatriatum has been documented in one patient.² Apparent hepatosplenomegaly has been noted in three infants. This is likely to be a consequence of the short, narrow thorax rather than true visceromegaly. Liver biopsy in case 7 in the table showed no abnormality.

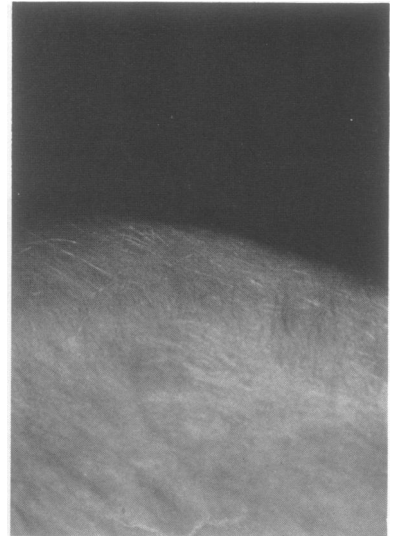


FIG 4 View of the scalp at the age of nine months showing poor growth of fine, sparse hair.

Radiographic changes

Skeletal radiographs show generalised osteoporosis with shortening of the ribs and long bones, particularly the humeri and fibulae. The distal phalanges are short and broad. The epiphyses of the long

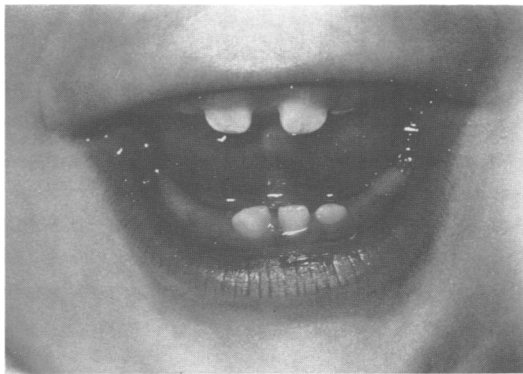


FIG 5 View of the patient's mouth aged nine months showing hypoplastic, widely spaced teeth with dysplastic enamel.

bones tend to be flattened and the femoral capital epiphyses may show delay in ossification. The vertebral bodies have convex upper and lower surfaces and in the lumbar spine the pedicles are short with less than usual widening of the interpedicular distance. The skull shows dolichocephaly, possibly in association with premature fusion of the sagittal suture.

Natural history

Growth parameters at birth are usually normal but during early childhood head circumference tends to increase disproportionately, whereas body length decreases as a reflection of the limb shortening. Information about height in older children and adults is not available. The child with congenital

heart disease (case 2 in the table) died at the age of three years, possibly as a result of recurrent chest infections, a problem which has precipitated urgent admission to hospital in case 7 on several occasions. Of the five patients described by Levin *et al.*,² three are reported to have died by the age of seven years (L S Levin, 1988, personal communication). Surgery for correction of premature sagittal suture synostosis is indicated. Intellectual development has been normal in all cases.

Differential diagnosis

The ectodermal dysplasias have been extensively reviewed by Freire-Maia and Pinheiro.⁴ Conditions showing involvement of both the skeletal and ectodermal systems include the Ellis-van Creveld syndrome, the GAPO syndrome, cartilage-hair hypoplasia, and the tricho-dento-osseous syndrome. Features which distinguish these disorders from cranioectodermal dysplasia include polydactyly and nail dysplasia in the Ellis-van Creveld syndrome,⁵ dry, fragile skin and optic atrophy in the GAPO syndrome,⁶ dwarfing and metaphyseal dysostosis in cartilage-hair hypoplasia,⁷ and curly hair and sclerosteosis in tricho-dento-osseous dysplasia.⁸ Premature fusion of the sagittal suture can be associated with several other malformations as non-specific findings⁹ and also occurs in a very large number of chromosomal and monogenic syndromes,¹⁰ none of which corresponds closely to cranioectodermal dysplasia.

Inheritance

With so few cases having been documented it is not possible to draw firm conclusions about the likely

TABLE Clinical features in patients with cranioectodermal dysplasia. Details of case 7 have not been published previously.

	Case						
	1 ¹	2 ¹	3 ²	4 ²	5 ²	6 ³	7
	(Sibs)		(MZ twins)				
Sex	F	M	F	F	M		F
Dolichocephaly	+	+	+	+	+	+	+
Sagittal suture synostosis	+	-	+	-	+		+
Short/narrow thorax	+	+	+	+	+	+	+
Palpable liver/spleen	+	+					+
Short limbs	+	+	+	+	+	+	+
Short digits	+	+	+	+	+	+	+
Single palmar creases	+	+					+
Short, thin hair	+	+	+	+	+	+	+
Small teeth	+	+	+	+	+		+
Short nails	+	+	+	+		+	+
Cardiac anomaly		+			-		-
Recurrent chest infection		+					+
Neurological findings		Hypotonia	Nystagmus	Nystagmus			
Normal intelligence	+	+	+	+	+		+

mode of inheritance. However, the reports of affected sibs¹ and monozygotic twins² are suggestive of autosomal recessive inheritance, as is the fact that case 5 in the table may have had an affected sib who died shortly after premature delivery.²

The author is grateful to Dr L S Levin for providing further details of the patients described by him in 1977.²

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