

Short reports

Cranioectodermal dysplasia in sibs

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Cranioectodermal dysplasia (Sensenbrenner's syndrome) is a rare genetic disorder in which abnormalities of the hair and teeth are associated with short limbs, narrow chest, dolichocephaly, and variable sagittal suture synostosis.¹ A recent review included brief details of an affected female infant.² The recent delivery of an affected brother provides further information about clinical and genetic aspects of this disorder.

The 2½ year old proband is the second child of healthy, unrelated parents. During infancy she required treatment on many occasions for life threatening respiratory tract infection associated with bronchospasm. These episodes have become less frequent and less severe. Other medical problems have included sagittal suture synostosis, febrile convulsions, and moderate bilateral myopia. Intellectual development has been normal. Her hair and nails have grown extremely slowly and her hair has never been cut (figure). At recent examination her head circumference (51.5 cm) lay above the 97th centile and her height (88 cm) fell on the 50th centile. Relevant findings included marked dolichocephaly, telecanthus, small, widely separated incisors, rhizomelia, narrow chest, protuberant abdomen, and thin, short nails.

Unconcerned by the possibility of recurrence, the parents readily agreed to regular ultrasonographic monitoring of fetal growth in a subsequent pregnancy. Biparietal diameter and limb length were consistent until 27 weeks, when humeral and femoral growth began to decelerate, resulting in a discrepancy of three to four weeks between head size and proximal limb length by delivery at 38 weeks. At birth the baby showed mild dolichocephaly with telecanthus, epicanthus, a short, narrow chest, and prominent



The affected sibs aged 2½ years and 3 months.

abdomen. The predicted limb shortening was confirmed. By 3 months, this infant had been admitted to hospital on three occasions for treatment of respiratory infection and showed clinical findings very similar to those of his sister (figure). However, no synostosis was evident on skull radiographs.

Several points emerge. Cranioectodermal dysplasia has previously been reported in a single set of monozygotic twins and in one other sib pair. This report of an affected brother and sister lends further support for autosomal recessive inheritance. Each of these sib pairs shows discordance for sagittal suture synostosis confirming that this is an inconstant feature. Finally, the absence of any demonstrable ultrasonographic abnormality until 27 weeks' gestation indicates that it could be very difficult to provide reliable prenatal diagnosis for this disorder.

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- 2 Young ID. Cranioectodermal dysplasia (Sensenbrenner's syndrome). *J Med Genet* 1989;26:393-6.