with hypoalbuminaemia and staphylococcal infections resembled Leiner’s disease, but he did not have the associated immunological abnormalities. The tricho-rhinophalangeal syndrome was considered because of the sparse hair and femoral head dysplasia, but he did not show the other skeletal changes of this condition. Atrichia congenita may be inherited as an autosomal recessive disorder and in view of the possibility of consanguinity in the child’s family, it seems likely that he has a rare autosomal recessive condition.

We should like to thank Dr Andrew Warin for advice on the dermatological aspects of this case.

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References


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Unknown syndrome: microcephaly, facial clefting, and preaxial polydactyly

SUMMARY We present a four year old boy with short stature, disproportionate microcephaly, developmental delay, convulsions, bilateral cleft lip and palate, and bifid right thumb.

History

Prenatal. No drugs in pregnancy. Maternal pyrexia for 24 hours at five weeks due to flu-like illness. Normal intrauterine movements.


Family. Only child of healthy Mauritian parents who are first cousins once removed. No other relevant family history.

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Medical history

Sat at eight months, began walking at 18 months. First words at four years. First convulsion at one year. Subsequent numerous convulsions which have been difficult to control with anticonvulsants. Grommets inserted at three years for ‘glue ears’.

Clinical examination

Aged four years. height 90 cm (<3rd centile), weight 11.2 kg (<3rd centile), head circumference 45.8 cm (<<3rd centile). Global developmental delay. Increased tone in lower limbs with unsteady gait. Unusual facies with flared nares and repaired bilateral cleft lip and palate (fig 1). Bilateral fifth finger clinodactyly and bifid right thumb (fig 2).

Investigations

Normal: routine haematology and biochemistry. G banded male karyotype, congenital infection screen, and CT scan. EEG (at two years): generalised spontaneous bursts of epileptic activity. (at four years) photomyoclonic response to photic stimulation.

Discussion

Non-midline facial clefting and preaxial polydactyly occur together in Meckel’s syndrome and in the pleiotropic autosomal recessive disorder described by Váradi et al.1 in which development is very severely retarded and death usually occurs in infancy or early childhood. The findings in this patient are not consistent with either of these diagnoses, nor with any other disorder known to us. Parental consanguinity suggests that this condition may show autosomal recessive inheritance.

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Reference


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