Unknown syndrome: mental retardation with postaxial polydactyly, congenital absence of hair, severe seborrhoeic dermatitis, and Perthes' disease of the hip

SUMMARY

We report a six year old male with mental retardation, postaxial polydactyly and syndactyly, atrichia congenita totalis, severe seborrhoeic dermatitis, recurrent staphylococcal skin sepsis, and Perthes' disease of the hip. His birth may have resulted from an incestuous mating.

History

The child's mother was a 22 year old educationally subnormal primigravida, and the father was her 46 year old stepfather, who may have been her natural father. Her half brother had died at birth with malformations, and two half sisters and a half brother, all of whom were fathered by her stepfather, were educationally subnormal. There was a strong history of atopy in the stepfather's family. The pregnancy proceeded normally, and the patient was born by normal spontaneous vaginal delivery at term, weighing 2.95 kg.

Medical history

At birth he was noted to have complete atrichia with absence of eyelashes and eyebrows. There was postaxial polydactyly with six toes on each foot and syndactyly of the second and third toes (fig 1). On the left hand there were six digits, but on the right there were only four, a thumb and three fingers (fig 2). A haemangioma of the scalp was present in the right parietal region. His penis was small, but the testes were descended. He subsequently developed a severe seborrhoeic exfoliative dermatitis with hypoalbuminaemia and hepatosplenomegaly which responded to treatment with a combination of topical steroids and systemic antistaphylococcal agents, as recommended for the treatment of Leiner's disease. On stopping maintenance anti-staphylococcal therapy staphylococcal skin infections occurred on two occasions. His development was delayed and he had epilepsy. At five years he began to limp and hip x ray showed avascular necrosis of both femoral heads, with an associated metaphyseal reaction. At six years of age he remains hairless with very rough skin (fig 3a). His teeth and nails are normal. The nose is rather beaked with a prominent columella, and the left ear has an irregularly folded helix (fig 3b). The head circumference is normal and his height is above the 3rd centile and weight above the 75th centile.

Investigations

Chromosome examination with G banding showed...
normal male karyotype, 46,XY. Immunological studies, including immunoglobulins, complement, and yeast opsonisation, were all normal. CT scan of the head showed slightly dilated ventricles. X rays of the hips showed fragmentation and increased density of the femoral heads with some changes in the femoral necks. The findings were asymmetrical and were consistent with Perthes' disease.

Discussion

The combination of features in this patient has not previously been described. The skin abnormalities suggest a form of ectodermal dysplasia, but he has normal nails and teeth, and the polydactyly and Perthes' disease could not be explained by this. The severe seborrhoeic dermatitis
with hypoalbuminaemia and staphylococcal infections resembled Leiner's disease, but he did not have the associated immunological abnormalities. The tricho-rhino-falangeal 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.

Received for publication 18 June 1987.
Accepted for publication 25 June 1987.

FIG 1 Facial view of the patient aged four years.

FIG 2 View of the patient's right thumb.