Investigations

Chromosomes (G banded) showed a normal female karyotype. Radiological examination showed the bones to be generally osteosclerotic with extensive periostitis and metaphyseal flaring (figs 4 and 5). These changes were particularly marked in the skull, pelvis, and long bones. Enzymology on cultured fibroblasts from a skin biopsy was normal and in particular excluded I cell disease and GM1 gangliosidosis.

Necropsy findings

Macroscopic examination showed hypoplastic lungs. The lungs weighed half their joint expected weight of 50 g. The liver and spleen were one and a half times their expected weight, weighing 15.4 g and 11 g respectively. The adrenal glands were hyperplastic; the right and left adrenals weighed 5.8 g and 6.3 g respectively, which is almost twice their expected weight. The brain was of normal size weighing 310 g. The contours of the brain were distorted by the shape of the skull but fissures and sulci appeared normal. There were no other abnormal findings on macroscopic examination.

Discussion

In spite of a detailed search of published reports we were unable to find any cases such as the one we have described.

We would like to thank Dr V Mitchell and Dr S Gilani for their help regarding the radiological aspects of this case.

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Unknown syndrome: pachygyria, joint contractures, and facial abnormalities

SUMMARY A male infant, the offspring of a father-daughter mating, is described. He had a lethal condition consisting of brachycephaly, large fontanelles, a flat face, a small nose with thin nares, hypertelorism, small ears with cystic pinnae, camptodactyly, talipes equinovarus, and hypoplastic lungs and kidneys. The brain was very short in the anterior-posterior diameter with simplified broad convolutions (a form of pachygyria).

Received for publication 21 November 1988.
Revised version accepted for publication 25 May 1989.

Medical history

This male baby was apparently the offspring of a father-daughter mating. The parents were West Indian and the mother was a 15 year old primipara. Pregnancy and delivery were normal. Birth weight was 3200 g, crown-heel length 49 cm, crown-rump length 34 cm, and head circumference 31.5 cm (<3rd centile). The baby died at three hours of age from respiratory insufficiency.

FIG 1 The face at necropsy.

FIG 2 Lateral view of head at necropsy; note cystic ear pinna.

Clinical examination

On examination the baby was brachycephalic with an upsweep of hair on the forehead and large anterior and posterior fontanelles and wide sutures. There was hypertelorism with short palpebral fissures (fig 1). The nose was small with a depressed bridge and narrow nares. The external ears were small and the pinnae were cystic (fig 2). The hands were puffy with camptodactyly of the fingers, mild skin syndactyly, and long, thin, adducted thumbs. There was bilateral talipes equinovarus. The liver was palpable at 2 to 3 cm below the costal margin and the penis was embedded in the scrotum, although the shaft was of normal size.

Investigations

Urine amino acids, long chain fatty acids, and chromosomes (G banded) were normal. Radiographs showed small bones in the vault of the skull with extremely large fontanelles and sutures (fig 3). At necropsy there were small but normally shaped kidneys (each weighed 2 g) with scattered foci of dysplastic features consisting of dilated tubules surrounded by primitive mesenchymal tissue. There was ectopia of the left kidney which was situated near the pelvic brim and was anteriorly rotated. The lungs were hypoplastic (each weighed 13 g), and an additional left sided superior vena cava draining into the right atrium via the coronary sinus was noted. The brain had a most unusual shape being very short in the anterior-posterior diameter with simplified broad convolutions (fig 4). Histologically, the cortical ribbon was slightly thicker than normal, expanded by one, or in some areas two, sharply

FIG 4 The brain; note brachycephaly with simplified broad convolutions.

defined narrow bands of poorly cellular, though somewhat gliotic, tissue running in parallel with the surface. Although a form of pachygyria, the pattern was different from both the Miller-Dieker and Walker-Warburg types of lissencephaly. There were no other CNS malformations.

Discussion

Some of the patient's features could have been secondary to neuromuscular abnormalities and poor fetal movement, for example, the joint contractures and hypoplastic lungs. However there are significant dysmorphic facial features and the other internal malformations suggest a multiple congenital anomaly syndrome. The patient's features did not seem to fit into other syndromes with pachygyria or lissencephaly¹ and a search of the London Dysmorphology Database did not show similar cases. In view of the close consanguinity of the parents, inheritance is most likely to be autosomal recessive.

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FIG 3 Lateral skull radiograph.