isolated cases it is possible that some autosomal recessive ones have been misinterpreted as sporadic.

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Two reciprocal translocations associated with microcephaly and retardation

SUMMARY The first case is reported of a karyotype containing two apparently unrelated reciprocal translocations, involving chromosomes 1, 2, 5, and 7. It is suggested that the patient's psychomotor retardation and microcephaly may be the result of the loss of a small amount of chromosomal material accompanying these translocations.

This report describes a 2-year-old white boy with psychomotor retardation, microcephaly, and two apparently balanced reciprocal translocations, one between chromosomes 1 and 2, the other between 5 and 7.

Case report

The patient was admitted to Babies Hospital for assessment of delayed speech and motor development. He is the only child of nonconsanguineous parents, born at term to a 21-year-old woman after an uncomplicated pregnancy and delivery. Birthweight was 3500 g; the head circumference was not recorded. The mother took no medication during pregnancy and had only one *x*-ray, a diagnostic abdominal film, during the sixth month. There was no other known exposure to potential mutagens or teratogens during gestation, nor during the life of either parent. The family history is non-contributory except for a paternal aunt with epilepsy and delayed speech development. The patient had no feeding problems, and height and weight growth were normal. He sat alone at 12 months and crawled at 15 months. At 2 years he walked poorly with assistance and had no consistently recognizable speech. He had had no seizures.

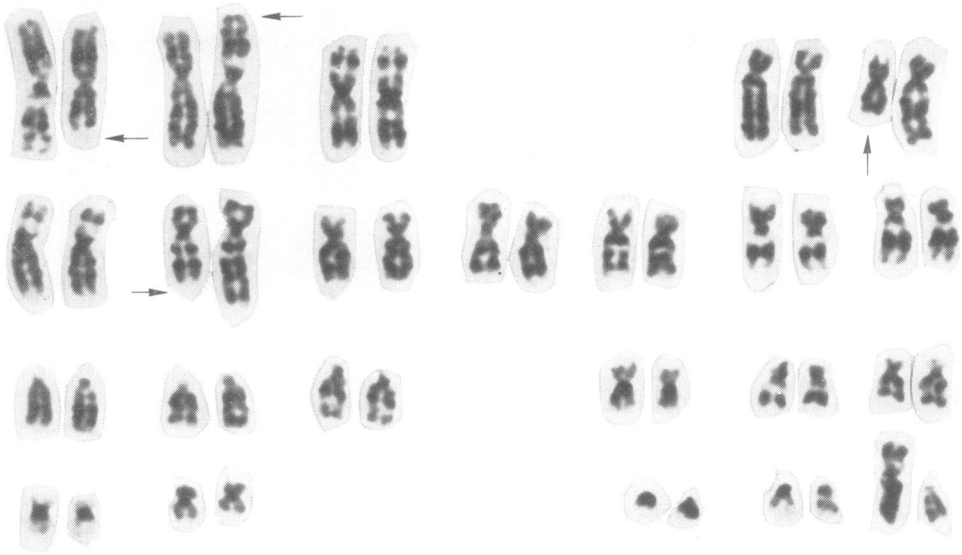
Fig. 1 *The patient at 2 years, showing prominent epicanthal folds.*

When admitted to hospital at age 2 years his weight was 14.6 kg (90th centile for age), his height was 98 cm (above the 97th centile), and his head circumference was 47 cm (below the 3rd centile). He had a diffuse angiomatous lesion on the right temporal scalp and prominent epicanthal folds (Fig. 1). General physical examination was otherwise normal. Cranial nerves, reflexes, strength, and tone were normal. Behaviour was conspicuously hyperactive.

Denver Developmental Screening Test confirmed performance at the 10- to 15-month level in all measurements. Complete blood count, urine analysis, bone age, serum thyroxine, and urine metabolic screen (amino acids, $FeCl_3$, DNPH) were all normal. Skull *x*-ray films showed only the clinically apparent microcephaly. Electroencephalogram showed a right occipital spike focus and asymmetric spindling.

CYTOGENETIC STUDIES

Karyotype analysis using Giemsa banding and quinacrine fluorescence showed 46 chromosomes in all of



46,XY,t(1;2)(1p2p;1q2q),t(5;7)(q21;q31)

Fig. 2 *Karyotype 46,XY,t(1;2)(1p2p;1q2q),t(5;7)(q21;q31).*

20 cells counted, with 4 abnormal chromosomes, replacing 1, 2, 5, and 7 (Fig. 2). These were interpreted to be the result of two reciprocal translocations, one an exchange at the centromere between chromosomes 1 and 2, the other an exchange between the long arm of 5 and the long arm of 7, with the chromosome complement expressed as: 46,XY,t(1;2)(1p2p;1q2q), t(5;7)(q21;q31) (Paris Conference (1971), Supplement (1975)). Both parents had normal karyotypes. Analysis of chromatid exchanges using BudR and Giemsa staining showed no abnormal patterns in the patient or either parent.

Discussion

To our knowledge this is the first report of a karyotype with two apparently unrelated reciprocal translocations. It is quite possible that loss of a small amount of chromosomal material has accompanied these translocations and is responsible for the child's psychomotor retardation and microcephaly.

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De novo balanced reciprocal translocation 46,XY,t(6;8)(q13;q22)

SUMMARY A 5-month-old infant was examined because of minor multiple malformations. He was found to have a *de novo* balanced reciprocal translocation 46,XY,t(6;8)(q13;q22). On follow-up at the age of 17 months his mental development was found to be within normal limits.

Hamerton *et al.* (1975) have found 0·08% balanced reciprocal translocations in a newborn population study; the large majority of them were familial.

The present report is that of a child with a *de novo* balanced reciprocal translocation 46,XY,t(6;8)(q13;q22) discovered because of minor congenital malformations. We do not know of any previous report of a translocation involving those two break points.