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 karyo-
type containing two apparently unrelated recip-
ciprocal 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 develop-
ment. He is the only child of nonconsanguineous
parents, born at term to a 21-year-old woman after
an uncomplicated pregnancy and delivery. Birth-
weight 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 were
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 nor-
mal. 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.

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 tem-
poral scalp and prominent epicanthal folds (Fig. 1).
General physical examination was otherwise normal.
Crani; nerves, reflexes, strength, and tone were nor-
mal. 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, FeCl3, 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 quina-
crine fluorescence showed 46 chromosomes in all of
Case reports

46,XY,t(1;2)(p12;p1q2q), t(5;7)(q21;q31)

Fig. 2 Karyotype 46,XY,t(1;2)(p12;p1q2q), 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)(p12;p1q2q), 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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Reference


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.
Two reciprocal translocations associated with microcephaly and retardation.
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