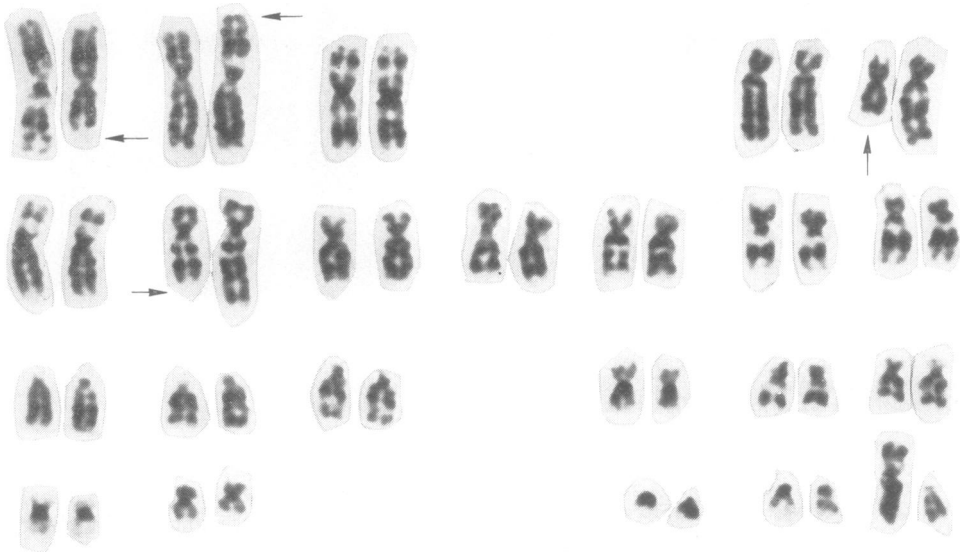

46,XY,t(1;2)(1p2p;1q2q),t(5;7)(q21;q31)

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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Reference

Paris Conference (1971). Supplement (1975): *Standardization in Human Cytogenetics*. Birth Defects Original Article Series XI, 9. The National Foundation, New York.

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.

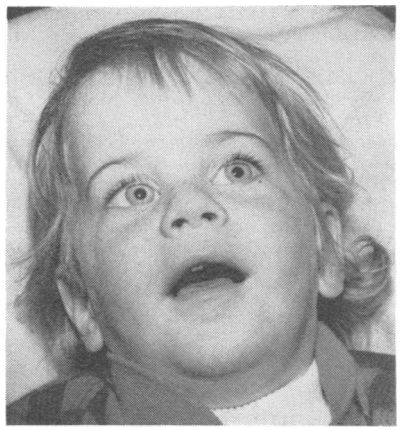

Fig. 1 *Face of patient; age 17 months.*

Case report

The propositus was born in Israel on 16 October 1974 at term. His birthweight was 2720 g, head circumference was 33·2 cm, and the Apgar score was 9. His progress was good except for some vomiting in the neonatal period. He was admitted to hospital at the age of 5 months because of bronchopneumonia and pharyngitis and a history of two previous respiratory infections. At this time it was noted that the child (Fig. 1) had slightly oblique palpebral fissures, epicanthic fold of the right eye, a somewhat flat profile, abundant neck skin, and a very mild degree of hypotonia. Partial cutaneous syndactyly of the second and third toes was present and minimal telangiectasia of the cheeks and ears was barely noticeable. The above findings were the reasons for cytogenetic investigation.

The parents and the two older sisters were normal. The father and the mother were 28 and 27 years of age, respectively, at the time of the proband's birth. They were Ashkenazi Jews and were not related.

Psychological evaluation at the age of 15 months showed a child with development within the normal range. At follow-up at the age of 26 months the development was normal.

Cytogenetic investigation

Chromosome investigations were performed on phytohaemagglutinin stimulated peripheral lymphocytes obtained from the propositus and his parents. Chromosomal banding was obtained by the ASG (Sumner *et al.*, 1971) and/or trypsin methods (Seabright, 1971).

Three cultures of the patient taken on three separate occasions were studied by trypsin or ASG banding and they all showed a balanced reciprocal translocation 46,XY,t(6;8)(q13;q22) (Fig. 2) in all

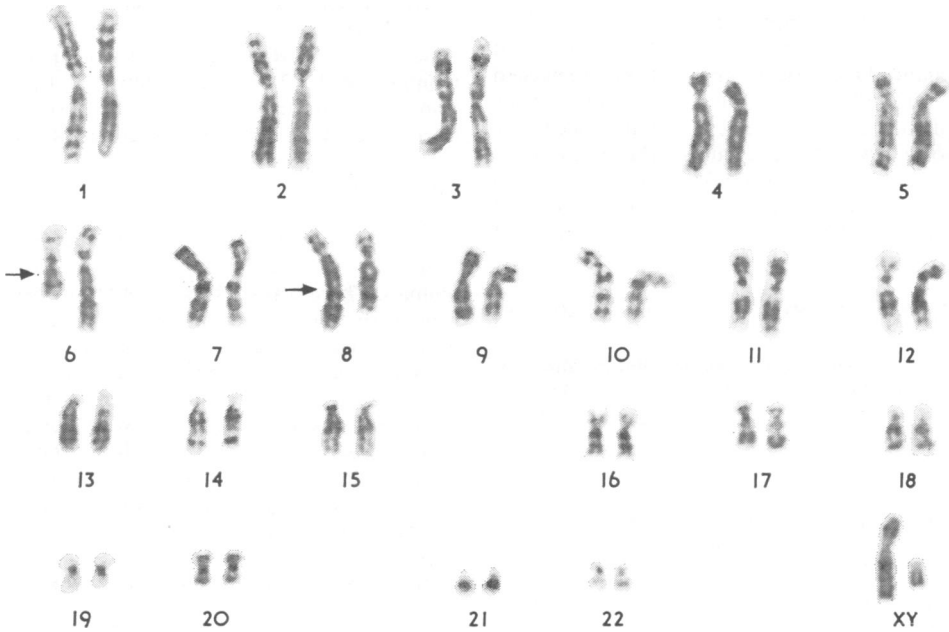

Fig. 2 *Karyotype (trypsin) of patient 46,XY,t(6;8)(q13;q22). Arrows indicate break points in the two translocation chromosomes.*

100 cells studied. Trypsin banded preparations of both parents and sisters showed normal karyotypes. Blood group findings did not reveal paternity exclusion.

Discussion

Jacobs (1974) discussed the correlation between euploid structural chromosome rearrangements and mental subnormality. While in a normal newborn survey *de novo* balanced reciprocal translocation was found in at least 1/3000, in mentally subnormal individuals it was found in at least 1/1000. She suggested that more intensive study of mutant individuals might well enable us to determine not only the proportion of euploid mutations associated with a phenotype effect but also the mechanism by which the effect is produced. This was further discussed by Jenkins *et al.* (1975). At present even though our case seems to have normal mental development we cannot but join in the hesitation expressed by Laurence and Gregory (1976) about prenatal diagnosis. They state, 'When there is a balanced translocation present, the pregnancy ought to be allowed to go to term. There may be some worry when this has arisen *de novo* in the fetus, as in the process some deletion may have occurred which is not detectable even by the banding techniques, in which case the fetus may be phenotypically abnormal.' *The Paris Conference (1971), Supplement (1975)* has also dealt in some detail with examples of *de novo* structural rearrangement. It seems that more detailed reports of *de novo* balanced reciprocal translocations are needed before a rational decision on termination can be reached, if such a fetus is detected on amniocentesis. Such reports will help to establish the distribution and frequency of points of breakage and exchange in human chromosomes; and allow a determination of the effect, if any, of parental age on the occurrence of translocations.

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A partial long arm deletion of chromosome 7: 46,XY,del(7)(q32)[1]

SUMMARY We have identified a partial deletion of the long arm of chromosome 7 in a newborn baby boy. His major anomalies were microcephaly, synbrachydactyly, diastasis recti, hypospadias, short neck, and widely spaced nipples.

With the development of the various new banding techniques, cytogenetic diagnosis of different types of chromosome deletions as well as clinical recognition of these deletion syndromes have become possible. With G-, R-, and Q-banding methods (Paris Conference, 1972), we have identified a partial deletion of the long arm of a No. 7 chromosome in a newborn male infant with multiple congenital anomalies.

Case report

The propositus (031273), was a product of the second pregnancy of a 29-year-old class A diabetic black woman. There was no history of fetal wastage in the mother or in two previous generations. The delivery was by caesarian section because of fetal distress at 35 weeks' gestation. Apgar score was 2 at 1 minute and 5 at 5 minutes. The birthweight was 1680 g and the length was 41 cm. The following congenital anomalies were noted: microcephaly (head circumference 29·5 cm) (Fig. 1a), synbrachydactyly of the right hand with hypoplastic nails on the thumb and the 5th finger (Fig. 1b), diastasis recti, first degree hypospadias, short neck, and widely spaced nipples. Both testes were descended. There was conspicuous

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