Vogel _et al_ (1970) noted a secondary constriction in the long arm of the abnormal chromosome and suggested that it represented the point of fusion between the two chromosomes participating in the tandem translocation. Similarly, it is interesting to speculate that the raised area visible in the euchromatic region of the present case also represents a point of fusion. If this is the case, a precise description of break points is possible. The translocation would be a result of a break near the centromere in q11 of the inverted chromosome, and one midway in q22 of a normal No. 21. This would result in a trisomy of the banded regions, most of the euchromatic region of q11 and a very small part of q22, along with monosomy for the distal half of q22.

The cases of Sachdeva and Vogel presented numerous features of Down’s syndrome while our case shows relatively few. Excluding mosaicism, the smaller size of our marker chromosome relative to those of Sachdeva and Vogel may account for this. We feel that the size difference is due to the amount of euchromatin present since all three cases appear to be completely trisomic for the banded regions.

**Partial trisomy 12 in a mentally retarded boy and translocation (12;21) in his mother**

**Summary.** Cytogenetic studies of an infant with malformations and a peculiar appearance showed a partial trisomy of chromosome 12. The mother carried a translocation of the distal part of chromosome 12 onto the short arm of chromosome 21, with breakpoints most likely at 12q24 and 21p11.
New cytogenetic methods have made it possible to identify chromosome abnormalities involving C-group chromosomes. Thus we present a case of partial trisomy of the long arm of chromosome 12 due to a maternal balanced translocation, between the long arm of chromosome No. 12 and the short arm of chromosome 21.

**Case history**

The propositus (Fig. 1) was born in December 1971, when the mother was 24 and the father 25 years old. The mother was an adopted child and nothing is known about her relatives. In 1969 she had a stillbirth in M.VI, a male, weight 850 g. In January 1971 she had a spontaneous abortion in M.II. Her menstrual periods were irregular with intervals up to 8 weeks, and she did not menstruate between the abortion and the third pregnancy.

This pregnancy was normal except for low oestriol values in the third trimester. When the duration of pregnancy was estimated to be 44 weeks, labour was initiated with syntecinon. Delivery in cephalic presentation was normal except for transient slow heart rate. Birthweight was 3500 g and length 54 cm; Apgar score was 6 after 1 min and 9 after 3 min.

At the age of 3 days the child was transferred from the obstetric department to the paediatric department because of peculiar appearance. His head was square-built with flattening of the top, the nose broad bridged and prominent with the tip turned downwards; the ears were low-set and poorly lobulated; the scruff of the neck was loose; the xiphoid process was prominent.

Stethoscopy of the heart revealed a systolic murmur along the left sternal border. The liver was 3–4 cm below the thoracic curvature. The scrotum was empty. Excessive hair was seen over the lower back. On both hands a simian crease was present.

On the fourth day of life a moderate neonatal jaundice was noted. There were feeding difficulties because of vomiting. During nursing generalized trembling was observed.

The child was discharged from the hospital at the age of 24 weeks. His weight was then 3560 g and there were no feeding problems.

At the age of 9 months mental and physical development was slightly retarded. He was hardly able to sit alone without support but the movement of his extremi-
ties was normal. He was interested in his surroundings. He could sit alone at 1 year of age. At the age of 2 years he could walk with slight support, but not alone. He pressed his feet down in planovalgus; his big toes were still in a hammertoe position. He still had a peculiar face and head, but less so than earlier. He had developed an angulation of his sternum, producing a deep transverse groove across the curvatures and lower part of sternum. There was no cardiac murmur or other signs of cardiac disease.

By the Vineland social maturity scale test he scored a SQ of 43.

Laboratory investigations

Haemoglobin, erythrocytes, erythrocyte volume fraction, and reticulocyte fraction were normal. Serum bilirubin at the age of 5 days was 220 μmol/l. In the urine, neither protein nor glucose was present and the sediment was normal.

At the age of 3 days radiographic examination of the thoracic and lumbar spine and the pelvis was normal. The thorax was probably normal. At the age of 9 months radiographic examination of the thorax showed signs of a congenital heart failure.

Cytogenetic Investigation

The chromosomes of the propositus, the mother, and the father were studied. With a modification of the method of Caspersson et al (1970), fluorescence studies were carried out in the propositus and the mother.

The propositus had 46 chromosomes. One chromosome, No. 21, had enlarged short arms. The short arms were non-fluorescing (Fig. 2). The other chromosomes were normal.

The mother also had 46 chromosomes. A translocation between the long arm of chromosome 12 and the short arm of chromosome 21 was found. The satellites from chromosome 21 were clearly

![Figure 2: Chromosomes 12, 19, 20, 21, 22, and Y from the propositus. Note the non-fluorescent but Giemsa-stained material on the short arm of chromosome No. 21.](image-url)
FIG. 3. Chromosomes 12 and 21 from the mother, the same cells with fluorescence microscopy and restained with Giemsa. The satellites of chromosome 21 are seen on the long arm of chromosome 12.

seen on chromosome 12 (Fig. 3). The breakpoint at chromosome 12 is most likely located at band q24, as the non-fluorescing part of the long arm of chromosome 12 is found on chromosome 21. The breakpoint on the 21 chromosome is most likely close to the centromere at p11. The chromosome abnormality in the propositus with the karyotype 46,XY,der(21)mat must have occurred on account of the translocation 46,XX,t(12;21)(q24;pl1) in the mother. The father had normal chromosomes.

Blood group, serum type, and enzyme studies are given in Table I. No abnormal segregation of the systems studied was observed.

**Discussion**

Unfortunately, the registration regulations of adoption authorities in Denmark makes it impossible to trace the family of the mother, thus preventing further gene location examinations and family studies.

The extra material of chromosome 12 must be the cause of the peculiar appearance and mental and physical retardation in the propositus. It is unlikely that loss of short arm 21 material has influence on the phenotype.

A few translocations involving chromosome 12 have been identified by new techniques. In two cases, the translocation apparently was balanced (Franke, 1972; Baheux-Morlier, Taillemite, and Roux, 1973) and in one case a trisomy for part of the short arm of chromosome 12 was found (Uchida and Lin, 1973). In another case, chromosome 7 was involved in the translocation and a partial trisomy 7 was observed (Carpentier, Rethoré, and Lejeune,
An unusual chromosomal segregation in a family with a translocation between chromosomes 3 and 12

Summary. A family is reported in which two infants were born with different types of congenital abnormalities. Chromosome studies on one of the infants showed a partial trisomy of the short arms of a No. 3 chromosome. A family study showed many balanced translocation carriers who had extra chromosomal material on the long arms of a No. 12 chromosome.

Balanced translocation carriers for a chromosomal rearrangement have the potential for producing abnormal infants and are, therefore, adding to the abnormal biological load in our society. Because of the almost unlimited chromosomal rearrangements that can occur among chromosomes, it is to be expected that this form of chromosomal mutation will greatly contribute to the number of children that are born with congenital abnormalities.

We wish to report a family in which two infants were born with different types of congenital abnormalities. Both of these infants died within the first 6 months of life. Chromosomal studies on one of the infants revealed a chromosomal abnormality suggesting a partial trisomy of short arms of a chromosome 3. The second infant who died had a different complex of congenital anomalies and we assume that his anomalies were caused by a deletion of a No. 3 chromosome.

Case reports

III.5 was a female infant weighing 1520 g. The anterior and posterior fontanelles were longer than normal. The ears were low set and she had micrognathia. The peritoneum was incompletely closed. There was a bicornuate uterus. Both large toes were displaced medially. The child lived 36 hours. The immediate cause of death was atelectasis of the lungs. The infant was born at another hospital and no chromosomal studies were done.

III.8 was a male infant born after 37 weeks' gestation and weighed 1700 g at birth. The patient's facial appearance was unusual and he was slightly jaundiced shortly after birth. The child lived for approximately 6 months and died of a congenital heart condition. Chromosomal studies which had been done at another hospital revealed that the child had 46 chromosomes with a karyotype which showed extra chromosomal material on the long arms of a No. 12 chromosome.

At necropsy examination, he had an enlarged heart with stenosis of the mitral valve. There was a deformity of the coccyx associated with a pilonidal sinus. There were congenital deformities of the lungs and kidneys. The penis was short and deformed. The testes were undescended.

Chromosomal studies

Chromosomal studies were done on both parents and their five living children (Fig. 1). The chromosomes were stained using the usual Giemsa staining technique. In addition, they were also examined by staining for G-banding and fluorescence (Fig. 2). The father's chromosomes were normal. The mother and all five children had 46 chromosomes which on karyotype analysis showed the same chromosomal rearrangement. There was a partial deletion of the short arms of a No. 3 chromosome and extra chromosomal material on the terminal end of the long arms of one of the No. 12 chromosomes (Figs. 2 and 3).
Partial trisomy 12 in a mentally retarded boy and translocation (12;21) in his mother

N. Hobolth, Petrea Jacobsen and Margareta Mikkelsen

doi: 10.1136/jmg.11.3.299

Updated information and services can be found at:
http://jmg.bmj.com/content/11/3/299

**Email alerting service**

Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

**Notes**

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/