Case reports


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Trisomy 20p from maternal t(3;20) translocation

SUMMARY A case of trisomy 20p resulting from a maternal translocation t(3;20) is described. QM and BUDR banding techniques were used for its identification. A round face with oblique palpebral fissures, strabismus, cardiac and vertebral abnormalities, mild psychomotor retardation, together with poor co-ordination and speech impediment, are the most typical features of the proband.

Few cases of trisomy 20p are to be found in published reports. Recently, Centerwall and Francke (1977) documented 3 new cases in the same family and reviewed the other 10 reported cases belonging to 5 families. In all cases the trisomy was the result of anomalous segregation of balanced parental translocation.

In this paper we report a case of trisomy 20p resulting from a balanced maternal translocation t(3;20) segregating in a large family.

Case report

The subject is a female, born 1.1.74, at 38 weeks' gestation, to a gravida 1 mother, after a gravidic toxemia which complicated the end of the pregnancy. The mother and father were 23 and 31 years old, respectively, at the time of her birth. Though the child was born after a normal delivery, she had mild fetoneonatal distress. Birthweight was 2750 g. Haemodynamic studies showed Fallot's tetralogy.

On physical examination after surgery at 4 years of age she had: normal weight and height, microcephaly (<3rd centile), round face, short upward slanting palpebral fissures with downward displacement of the medial corner, moderate hypertelorism, short nose with large nares, and bilateral convergent strabismus (Fig. 1).

According to the Terman-Merrill scale, there was psychomotor retardation of 1 year compared to a normal subject which, 5 months later, appeared greatly improved (retardation of 6 months). The

Fig. 1. The proband at the age of 4½ years.
logopaedic control confirmed the presence of slight speech defects. There was also a certain lack of manual co-ordination which could be partially the result of the considerable strabismus.

Orthopaedic examination showed a mild dorsolumbar kyphosis, a slight degree of scoliosis, and also valgismus, pronation, and flat-footedness.

X-rays of the spinal column showed alteration in the structure of the vertebrae in the dorsolumbar region, characterised by rounded and irregular margins with reduced intervertebral spaces and a tendency towards partial fusion of the last thoracic vertebrae. Furthermore, at lumbar level the vertebral bodies appeared to be more developed in the anterior segment than in the posterior one. There were no alterations of the cervical vertebrae.

X-rays of the dental arches showed the absence of germs of some teeth, while those present showed a tendency to taurodontism.

Urography showed a paravertebral ectopic right kidney situated at the level of the 4th to 5th lumbar vertebrae and slightly smaller than the left. The pelvis had a bifid appearance.

CYTOGENETIC FINDINGS
Leucocyte cultures from peripheral blood were set up and the metaphases obtained were analysed according to our standard method: the same metaphases were stained first with Giemsa and then QM banded.

The proband’s karyotype, 46,XX, showed a slightly anomalous banding pattern on the short arm of a chromosome 3 (Fig. 2a). It was hypothesised that the anomalous chromosome was the result of a translocation t(3;?). The father of the proband had a normal 46,XY karyotype. The mother’s karyotype showed a reciprocal translocation between chromosome 3 and 20 (Fig. 2b). The der(3) was identical to the anomalous 3 of the daughter. The morphology and banding pattern of the deleted 20 were very

Fig. 2  (a) Proband’s chromosome pair no 3 from different metaphases, stained first with Giemsa (bottom row) and then QM banded (upper row). (b) Three partial karyotypes of the mother showing chromosomes 20 and 3 stained as in (a). (c) Three examples of maternal pairs 20 and 3 stained by BUdR technique.
similar to a chromosome 22, the satellites of this pair being very faint. For definite identification we examined metaphases in which both chromosomes 22 were involved in satellite association.

In order to obtain more detailed information about the exact breakpoints we used the BUdR technique (Dutrillaux et al., 1973) (Fig. 2c). The breakpoints were identified as 3p27 and 20p11. According to the Paris Conference (1971), the translocation can be described as follows: t(3;20) (3qter → 3p27::20p11 → 20pter; 20qter → 20p11::3p27 → 3pter). Since the proband inherited from her mother the normal 20 and the der(3), she is trisomic for 20p11→20pter and monosomic for 3p27→3pter. Cytogenetic analysis was extended to the mother’s family (Fig. 3). Individuals II.7 and III.19 were not tested because of lack of co-operation.

DERMATOGLYPHS
All digital patterns were ulnar loops except for an arch on the left 3rd digit. Total ridge count was 108 and maximal atd angle was 149°.

Discussion
Knowledge of the pathology of F group chromosomes is very scarce. As regards 20p trisomy, several explanations can be produced to account for its rarity.

1) The chromosome 20 could be prone to breakage only rarely (Aymé et al., 1976).

2) The small size of this segment only slightly alters the morphology of the chromosome involved in the translocation (see our case), making its morphological identification difficult. In fact, all described cases were discovered using banding techniques.

3) The clinical abnormalities of this syndrome, especially at birth, are not very serious and can escape detection.

Comparing the phenotypic features of the reported cases with ours, it should be remembered that though the duplicated segment is similar, this does not imply similarity at the molecular level. Furthermore, the deleted segment, different in all families, may also contribute to the phenotype. These arguments, together with the different genetic background, probably account for differences among patients from different families. Keeping in mind these considerations, it can be noted that the clinical picture of our patient seems to fit well enough with the most typical features of the syndrome.

A common characteristic of autosomal derangement is serious impairment of mental activity. Cases of trisomy 20p often do not show this and the small size of the 20p and/or the absence of important genes located on this segment can account for this fact. In our case, the psychomotor retardation was mainly the result of lack of physical activity and environmental stimulation because of the heart defect. The rapid progress made in only 5 months after surgery supports this hypothesis, which can only be completely verified in the future.

Examination of the pedigree shows the presence of miscarriages and deaths in the descendants of the carriers. As a general consideration we suggest that monosomy 20p does not appear to be viable and can therefore contribute in explaining these miscarriages, while the trisomy 20p could, at least partially, be represented in the dead children. Unfortunately, anamnestic inquiries yielded little information regarding the cause of death of most of the deceased children. Nevertheless, it was possible to establish that only 3 of these subjects (III.3, III.10, III.17) died of causes which were definitely not connected with the chromosome pathology in question, at ages varying from 5 to 18 years old. All the others died within the first months of life (most of them within the first week), and in some of them (III.2, III.16, IV.1) a congenital heart defect had been diagnosed.
In conclusion, it should be remembered that the diagnosis of this syndrome can only be suspected on clinical grounds alone and requires cytogenetic confirmation.

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References


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Triple mosaicism 45,XY,—18/46, XY/47,XY,+18

SUMMARY A patient with symptoms clinically resembling Edwards's syndrome is presented. Cranial asymmetry, thoracic and lumbar hemivertebræ, and an additional rib were the unusual features. The cytogenetic studies revealed the coexistence of three separate cell lines with 45,XY,—18/46,XY/47,XY,+18 complement.

Group E triple mosaicism has been reported twice in published medical reports (Backus and Darien, 1968; Bricarelli et al., 1971). These two cases presented with congenital asymmetry, scoliosis, and, in addition, Sprengel's deformity and vertebral and rib anomalies were noted in one patient (Backus and Darien, 1968). We wish to present a further case, the first to be confirmed by chromosome banding, which showed the aberration to be associated with chromosome 18.

Case report

A 17-day-old male baby was referred from the nursery because of a strangulated inguinal hernia. The baby was the second child of normal, unrelated, healthy, Jewish parents of Yemenite origin. The family history was uninformative. The mother was treated with progesterational agents from the 12th to the 20th week of pregnancy because of threatened abortion. The infant was born by caesarean section at 32 weeks because of an accidental maternal antepartum haemorrhage. The birthweight was 1370 g.

Physical examination on admission revealed an acutely ill premature baby, weighing 1400 g. Marked asymmetry of the skull as a result of prominence of the right parieta! and occipital bones was evident. Mild retrogondhia was noted. The left ear was smaller, lacking a lobulus. The index and middle fingers were flexed and overlapping and rocker-bottom feet were prominent. There was lateral displacement of the nipples and the sternum was short. Marked scoliosis of the spine with convexity to the right was present. A grade 3 systolic heart murmur was found, shown later by cardiac cathe
terisation to be because of a large ventricular septal defect and a persistent ductus arteriosus. The abdomen was distended, and an irreducible right inguinoscrotal hernia was present. The rest of the physical examination was normal.

Complete blood count, biochemical analysis, and serological tests for syphilis, cytomegalic inclusion virus, rubella, and toxoplasmosis were negative.

Under local anaesthesia, resection of a necrotic ileal loop was performed. There was initial improve
tment, but he developed congestive cardiac failure. At the age of 67 days he died of extensive bilateral pneumonia.

Additional findings at necropsy were an enlarged right cerebral hemisphere, corresponding to the asymmetry observed, absence of the left kidney, and enlargement of the right one with a double ureter. Also present was a right hemivertebra at T10 with a complete additional rib, and the first lumbar vertebra was 'Y'-shaped.

Cytogenetic studies

Cytogenetic studies, using peripheral blood lympho
cytes of the child and both his parents, were performed according to the method described by Moorhead et al. (1960). G-banding was performed by the method of Seabright (1971), modified by
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