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A Double Aneuploid Mosaic: Trisomy 13 and XXY*

We have examined a liveborn male who showed physical features of trisomy 13 syndrome and was

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found to have 47,XY,13+/48,XXY,13+ mosaicism. To our knowledge, this is the first report of trisomy 13 and an XXY sex chromosome complement coexisting in a liveborn individual.

Case Report

The patient, a Caucasian male, was born to a 17-year-old primigravida mother and a 19-year-old father. The father had a son by a prior union. The remainder of the pedigree was unremarkable. Both parents had taken lysergic acid diethylamide (LSD) 2 years before the pregnancy. The infant was the product of an apparently normal 36-week gestation and vertex delivery. Birth weight was 1480 g, length 42 cm, and head circumference 29 cm. The anterior fontanelle was large; the sagittal suture was 1 cm in each diameter. There was a bony defect 1.6 × 2 cm in the parieto-occipital region through which the meninges were seen (Fig. 1). The parietal bones overlapped the frontal and occipital bones. The face had an inverted triangular shape with a broad forehead and hypoplastic left eye (Fig. 2). The right eye was normal. The nose was large and prominent, the mouth was small and the ears were low set. The neck showed redundant skin folds. There was a ventral hernia 1.5 cm above the umbilicus. Except for an undescended left testis, the genitalia were normal for a male infant. Both hands had a simian crease and an extra ulnar digit. The feet were rocker bottom in shape and showed a wide space between the 1st and 2nd toes. The remainder of the physical examination was normal. The infant was alert, cried vigorously, and showed no abnormal neurological findings. Shortly after admission he became cyanotic and was given antibiotics for suspected

FIG. 1. Scalp view of the propositus.

Fig. 2. Side view of the propositus.

septicaemia. He developed tonic seizures and died at 10 hours of age.

In addition to the clinical findings, necropsy examination revealed a hypoplastic left optic nerve about half the size of the right optic nerve. The heart, which was normal in size, had a 2 mm defect between the left atrium and sinus venosus and a high ventricular septal defect. There was an extra left common carotid artery. Also

present were hypoplastic thymus, malrotation of the intestines, a Meckel's diverticulum, and accessory spleens. There were only 10 ribs on each side. Microscopic examination revealed aspiration pneumonitis, focal pancreatitis, and thickening of the renal tubular membrane. The thymus showed lymphoid depletion, and the bone marrow revealed a marked granulocytic proliferation.

Chromosome Analysis

Chromosome analysis (Fig. 3) was done from the patient's blood, skin, and fascia. All of the cells examined from the peripheral blood showed trisomy D (47,XY,D+). Chromosome analysis of skin and fascia showed another cell line with an extra chromosome in the C group; 48,XXY,C+,D+ (Table I).

TABLE I
CHROMOSOME ANALYSIS

Tissue	47,XY,13+ (no. of cells)	48,XXY,13+ (no. of cells)	Total Cells Analysed
Blood	18	0	18
Skin	63	8 (11%)	71
Fascia	34	3 (8%)	37

One thousand cells from each of the tissue cultures were examined for sex chromatin. Fourteen cells in the skin culture (1.4%) and 47 cells in the fascia culture (4.7%) were sex chromatin positive. Sex chromatin nuclei were also observed in sectioned

FIG. 3. Karyotype from a fibroblast cell (skin) showing 48,XXY,13+.

adrenals. Autoradiography of skin fibroblast culture demonstrated that the cell line with 48 chromosome included a distinctly late-labelled X chromosome and an extra D chromosome labelling as expected for No. 13. The labelling in the 47 chromosome cell line also confirmed the extra D to be No. 13. No euploid cells were found. Chromosome analysis of peripheral blood from each parent was normal.

Discussion

Pergament and Kadotani (1965) found trisomy D and XXY in a tissue culture from the limb-bud of an early spontaneous abortion. Although our patient is the first reported liveborn infant with trisomy 13 and an XXY sex chromosome complement, we are aware of another such infant through a personal communication (C. B. Francisco and C. Herzon). That infant lived 2 hours and also had physical features consistent with trisomy D₁ syndrome. It is noteworthy that the double aneuploidy in our patient would have been undetected if only peripheral blood had been examined.

There are at least 4 possibilities for the mechanism by which an individual can be mosaic for single and double aneuploidy. The first is fertilization between a normal gamete and a gamete with an extra chromosome resulting in a trisomic zygote; ie, 47,XY,13+, followed by a subsequent non-disjunction producing a double aneuploid line (48,XXY,13+). The other cell line of 46,Y,13+ is presumably non-viable. A second possibility entails an initially euploid zygote followed by 2 non-disjunctional events resulting in 46,XY/47,XY,13+/48,XXY,13+ cell lines. This explanation is less satisfactory since a normal euploid cell line was not found in our patient. A third possibility is the fertilization of 2 gametes each with an extra chromosome, resulting in a doubly aneuploid zygote; namely, 48,XXY,13+. A later non-disjunction could then result in one cell line trisomic for only the No. 13 chromosome and another cell line of 49,XXX,Y,13+, presumably non-viable. A fourth possibility is that of anaphase lag occurring in a double aneuploid 48,XXY,13+ zygote, producing 48,XXY,13+/47,XY,13+ mosaicism. However, the fact that 48,XXY,13+ is the minor cell line is against it. The simplest explanation involving the fewest number of division errors is the first possibility.

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A Case of Partial 14 Trisomy 47,XY,(14q-)+ and Translocation t(9p+;14q-) in Mother and Brother*

D group trisomy (13-15) was first described by Patau *et al* in 1960. Since then at least 126 cases with cytological confirmation have been reported (Taylor *et al*, 1970). The phenotypic features have been tabulated and considerable variations noted. In some of these cases the extra chromosome was identified by autoradiographic techniques (Giannelli, 1965) and recently in 2 cases by quinacrine fluorescence (D. A. Miller *et al*, 1971) as a No. 13. The rest have been presumed to be 13 trisomy because of the clinical similarity of the phenotype. A syndrome associated with trisomy of chromosome 14 has not been described. We report here a case trisomic for a large part of chromosome 14 identified by quinacrine fluorescence.

Family studies revealed a translocation, t(9p+;14q-), involving the long arm of chromosome 14 and the short arm of chromosome 9 in the mother and an older sib of the propositus, both phenotypically normal. The case represents transmission, presumably by non-disjunction, of the structurally abnormal chromosome 14 (14q-) from the translocation carrier mother producing a child partially trisomic for 14 [47,XY,(14q-)+].

Family History

The pedigree is detailed in Fig. 1. The propositus (III.3) was the product of the 3rd pregnancy of a

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