Discussion

Zellweger and Abbo (1963) described different translocations in the cells of the same individual with Down's syndrome. Their case was a girl with mosaicism including four different cell lines. One line contained a balanced D/D translocation, an unbalanced D/G translocation and a 45,X sex-chromosome constitution. Another cell line contained a balanced D/D translocation. A third line contained an unbalanced D/G translocation with a 45,X sex-chromosome constitution. A fourth line was normal. Chromosomal mosaicism was observed in several other members of the family. The findings were attributed to an autosomal dominant gene.

In the case under discussion the presence of a balanced 15/21 translocation in one cell line and an unbalanced 21/21 translocation in another line effectively results in normal/trisomy-21 mosaicism. The possibility of an isochromosome for the long arm of a No. 21 cannot be excluded. The relationship of these two different translocations to one another, if any, is unknown. The mother's history of five spontaneous abortions suggests some predisposing factor for the chromosomal abnormalities. If a translocation were present in a parental gonad and inherited one would expect all of the cells in the child to have the same abnormality. A plausible explanation for the findings could be postzygotic chromosomal breakage within two different cells in an early cleavage division with the resulting translocations. The cause for the breakage is unknown.

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References


48,XXX, + 18 double trisomy*

Summary. An infant who died at 127 days with gross congenital deformities is described. Cytogenetic analysis showed double trisomy of 18 and X which was confirmed by autoradiography and fluorescent banding techniques.

Double trisomies are not of common occurrence in man. The most prevalent double trisomy involving one of the sex chromosomes is a 48,XXY,+G male having Klinefelter's and Down's syndromes. Only four cases involving X and 18 have been reported (Uchida and Bowman, 1961; Haas and Lewis, 1966; Ricci and Borgatti, 1963; Engel et al, 1967).

This report concerns another case of 48,XXX, + 18 double trisomy in a female infant.

Case history

A female infant was born on 14 January 1972 after full-term normal pregnancy to a 22-year-old mother, who had a normal 34-year-old child from her previous pregnancy. The father was 21 years old. The birth weight was 1560 g. There were several gross malformations. The head was long with a pronounced occiput, the ears were low set, and the right pinna was deformed with the absence of an external meatus. The index fingers of both hands were flexed and curved over the middle fingers and there were bilateral palmar creases. The chest was shield-shaped. There was a loud systolic murmur present over the precordium. The child had rocker bottom deformity of both feet. The genitalia were unusual with part of the labia giving the appearance of a clitoris. The baby died of cardiac arrest at 127 days of age. No necropsy was performed.

Materials and methods

Chromatin mass. Squamous cell smears were prepared from inside the cheeks. Barr bodies were counted in 500 cells.

Chromosome analysis. Leucocyte chromosomes from the peripheral blood samples (two) were prepared according to Moorhead et al (1960). Fifty cells were counted from each sample. Skin fibroblast chromosome slides were also prepared and a sample of 30 well-spread metaphases was taken and analysed.

Autoradiography. Autoradiographs were prepared from 72-hour cultures and from skin culture chromosomes according to the method described by Schmid (1963).

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Fluorescent banding. Slides prepared from 72-hour leucocyte cultures and skin fibroblast cultures were stained for quinacrine mustard according to the method of Dosik, Madahar, and Ehlin (1972).

Results

Of 500 cells analysed from the buccal smears for sex chromatin, 80% of the cells had Barr bodies of which 48% were double bodies and 32% were single Barr bodies; the remaining 20% of the cells were negative for sex chromatin.

Chromosome analysis from leucocyte cultures showed the presence of 48 chromosomes in all the cells. The skin fibroblast culture cells also had 48 chromosomes.

Autoradiographic analysis of the chromosomes from leucocyte and skin fibroblast cultures revealed two late-replicating X chromosomes and three 18 chromosomes with late-replicating long arms (Fig. 1).

Discussion

The first double trisomy, a male with 48,XXY, + G chromosome constitution was described by Ford et al (1959). Hamerton (1971) has summarized the most commonly found double trisomies and reported the expected incidence to be about 0.3 x 10^{-5} live born babies.

The presence of trisomy 18 in combination with trisomy X has been previously reported only four times. In the present case the infants had typical 18 trisomy features. It appeared that the extra X
chromosome had not contributed to any phenotypic abnormalities in the infant. This is consistent with the finding that triple-X females are phenotypically normal. The two late-replicating X chromosomes demonstrated by autoradiography are in accordance with Lyon’s hypothesis of genetically inert X chromosomes (Lyon, 1961).

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