a parent. The products would then be true isochromosomes by Darlington's (1939) definition. Secondly, a reciprocal translocation may have occurred as a result of breaks at or near the centromere during the first division of meiosis between two No. 18 chromosomes followed by nondisjunction. In a recent report (Salamanca-Gómez, and Armendares, 1975) an 'isochromosome' of the long arm of No. 17 appears to be caused by this type of event. A marker indicated the non-sister origin of each arm.

Transverse division of one centromere seems more likely than breakage and reunion at appropriate positions between two No. 18 chromosomes. With support of the G and C band analysis, we favour isochromosome formation as the mechanism of formation of the two metacentric chromosomes in this report.

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Partial 18 trisomy (with 47 chromosomes) resulting from a familial maternal translocation

SUMMARY A newborn female infant presented with the classical picture of 18 trisomy syndrome. Her karyotype was 47,XX,+der(18)t(12;18)(q24;q21)mat. The mother was a balanced reciprocal translocation carrier and so too was one of the two maternal uncles of the proposita, indicating that the translocation was already present in one of the grandparents who were not available for examination. This family suggests that triplication of the distal part of the long arm of chromosome 18 is not necessary to produce Edwards' syndrome.

Tertiary trisomy resulting in partial trisomy 18 in a child of a balanced D/E translocation carrier mother has been described by Gleissner *et al.* (1970), but at that time the break points were not defined.

Fig. 1 Pedigree.

Fig. 2 The *proposita* showing typical features of 18 trisomy syndrome: (A) Note hypertonicity evident in clenched hands and crossed legs; short sternum; wide spaced nipples; protruding calcaneum. (B) and (C) Low set, slanted auricle; micrognathia; short neck with skin folds. (D) Short hallux; hypoplasia of nails. (E) Clenched hand with thumb and index finger overlying the third; narrow fingernails. (F) Haemangioma in lumbar area.

This report describes a case with typical features of trisomy 18 indicating that triplication of the distal part of the long arm of chromosome 18 is not necessary for the production of the syndrome.

Case report

The *proposita* (Fig. 1 and 2) was born after 42 weeks of pregnancy, on 10 October 1976. The father and mother were 39 and 32 years of age, respectively, at the time of her birth. Before the pregnancy the mother took contraceptive pills (Metrulen) but stopped when she decided to become pregnant and she conceived after a single menstrual period. During pregnancy she had pharyngitis in the 5th month of pregnancy and otitis in the 9th month for which she received symptomatic treatment.

The delivery was normal, birthweight 3350 g; head circumference 33 cm; the placenta weighed 550 g; Apgar score 10. Multiple malformations were suggestive of trisomy 18 (Fig. 2). A haemangioma was noted in the lumbar area, and x-ray examination demonstrated a hemivertebra at L3. Intravenous

pyelography revealed right ectopic kidney. Poor sucking capability necessitated nasogastric tube feeding but even with this the child failed to thrive, and died in hospital at the age of 5 months. Dermal ridge patterns were compatible with trisomy 18.

Cytogenetic studies and family report

Trypsin banded chromosomes (Seabright, 1971) were studied from PHA stimulated lymphocytes.

The *proposita* was a tertiary trisomic, 47,XX,+der(18)t(12;18)(q24;q21)mat (Fig. 3); in all, 100 cells were counted. Fifty maternal cells had the karyotype 46,XX,t(12;18)(q24;q21) (Fig. 4). Two maternal relatives also carried the balanced translocation (Fig. 1). The maternal grandparents were Jewish and not related and both were born in Tripoli, North Africa. The grandfather I.1 and the grandmother I.2 were 30 and 18 years of age, respectively, at the time of the birth of the mother but were not available for cytogenetic investigation. Two sisters of the mother II.3 and 4 died at the age of 9 months and 2

Fig. 3 Karyotype of the proposita (trypsin) 47,XX,+der(18)t(12;18)(q24;q21)mat.

days, respectively, and the only detail known was that II.4 was born prematurely after 7 months of pregnancy.

Blood group markers were not informative in this family.

Discussion

The problem of reciprocal translocation and 3:1 meiotic disjunction resulting in 47 or 45 chromosome offspring has been reviewed by Lindenbaum and Bobrow (1975). The present case seems to be the first tertiary trisomy segregant 3:1 of the type 47,XX,+der(18)t(12;18)(q;q). In tertiary trisomies the extra chromosome was always the shorter of the two derivative chromosomes and this was the case in the proposita.

Fig. 4 Partial karyotype (trypsin) of mother of proposita balanced reciprocal translocation carrier 46,XX,t(12;18)(q24;q21). Arrows indicate break points in the two translocation chromosomes.

The exact part of chromosome 18 responsible for the expression of Edwards' syndrome when present in triplicate is still unknown. From the phenotype-karyotype correlation of the proposita it seems justified to conclude that triplication of the distal third of the long arm of chromosome 18 is not necessary to produce Edwards' syndrome.

The mother (II.1) and her brother (II.5) were advised about the possibility of prenatal diagnosis in future pregnancies.

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