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Partial Trisomy of the Long Arm of Chromosome No. 7*

Summary. A case report on an infant with trisomy of the distal third of the long arm of chromosome No. 7 is presented.

The report of a child with 46,XY,14q+ karyotype is presented. The extra segment on the long arm of chromosome No. 14 was identified as the distal one third of the long arm of chromosome

No. 7. This case thus has a partial trisomy of the long arm of chromosome No. 7, a condition which has not been described previously.

Case Report

The propositus, a 5-month-old male, with a history of recurrent pneumonia and failure to thrive, was born to a 39-year-old, gravida 4 para 4, mother and a 44-year-old father. Both parents had been married before, and the propositus is the only product of the present marriage. Family history was negative for abortions, stillbirths, or congenital malformations. Gestation was full term, with a breech presentation, and polyhydramnios. A Cesarean section was performed because of an obstetric problem in a previous pregnancy. Birth weight was 3450 g. There were early respiratory and feeding difficulties. Recurrent pneumonias over the first four months of life necessitated almost continuous hospitalization.

The propositus was first seen at the Children's Hospital of Los Angeles at the age of 4½ months. He weighed 3450 g. His height was 51.5 cm and head circumference was 41 cm. He had an asymmetric head, with prominent occiput, more so on the right side. The anterior fontanelle was widely patent (8 x 10 cm). The

FIG. 1. The propositus.

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eyes were prominent and the nose was small. The ears were low set, with deeply cupped outer helices. There was micrognathia, and a large incomplete cleft palate not involving the alveolar ridge. The small chin and small nose contrasted with the cranium which appeared large in comparison with the facial structures (Fig. 1). The chest was symmetrical. Coarse and fine rales, and wheezes were heard over the entire lung fields. There was a midprecordial, grade II/IV pansystolic murmur consistent with ventricular septal defect. The liver edge was palpable 3 cm below the right costal margin. The spleen was not enlarged. The external genitalia were normal, with both testicles descended. There was a pilonidal dimple. The feet showed syndactyly of the right three middle toes. The finger and toe nails were hyperconvex. The baby was generally hypertonic, and occasionally turned opisthotonic when aroused. Radiography showed bilateral pneumonia, cardiomegaly, and diastasis of the symphysis pubis. The oesophagram suggested the presence of an aberrant right subclavian vessel.

**Cytogenetic Studies**

Leucocyte cultures from peripheral blood showed a 46 chromosome count with one long D chromosome in all 30 cells examined. A quinacrine fluorescence study (Caspersson et al, 1970) showed the long D chromosome to be No. 14, with an extra segment on its long arm (Fig. 2). The fluorescence pattern on that segment matched that on the distal part of the long arm of chromosome No. 7.

Chromosome analysis of the father's blood showed a balanced translocation of the distal one-third of the long arm of chromosome No. 7 to the long arm of chromosome No. 14 (see Fig. 2). No evidence for a reciprocal translocation of the telomeric end of No. 14 could be visualized on the long arm of the deleted No. 7. The mother's karyotype was normal.

The short-system designation (Paris Conference, 1971) for the chromosome findings in this family would be:

**Father:** 46,XY,t(7;14)(q31;qter).

**Propositus:** 46,XY,der(14),t(7;14)(q31;qter).

**Dermatoglyphic Studies**

A study of the patient (Fig. 3) showed whorls on all 10 fingers, transitional flexion creases on both hands, and no signs of dysplasia or ridge disruptions.

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**Fig. 2.** Partial karyotypes showing chromosomes Nos. 7 and 14 of the father (a), and the propositus (b).

**Fig. 3.** Dermatoglyphics of the propositus.
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Right hand:
Left hand:

The father’s palms showed six ulnar loops on fingers 1, 3, and 5 of both hands, radial loops on both index fingers, and whorls on the fourth fingers.

Right hand:
9.7.5°.5′: 38°. Au.A.A.:O.O.L.O.
Left hand:
The mother had whorls on all 10 fingers.

Right hand:
9.0.7.3u: 42°. Au.A.A.:O.O.O.O.
Left hand:
9.7.3u: 40°. Au.A.A.:O.O.O.O.

Discussion

The clinical findings in the patient described in this report include, among others, such features as asymmetrical skull, low set ears, micrognathia, cleft palate, short neck, hyperconvex nails. These findings have been described in association with several other chromosome abnormalities. Although none seems to be specific for partial 7q trisomy, it is possible, as other patients are identified, that some combinations of these features may prove to be diagnostic.

The observations provided in the present report illustrate that the banding techniques can localize ‘breakage’ points in the chromosomes involved in a translocation (see Fig. 2). It is noted that the distal major band on the long arm of No. 7—band 7q31 (Paris Conference, 1971)—is located on the long arm at about two-thirds of its length from the centromere. The proximal portion of this band is retained on the deleted chromosome No. 7. It is almost certain, based on this pattern, that the breakage point on chromosome No. 7 occurred within this band. A close study of the banding patterns of the two chromosomes, did not reveal a definite reciprocal translocation of the telomeric end of chromosome No. 14 to chromosome No. 7 (see Francke, 1972 and Shaw, 1972). However, a reciprocal translocation involving segments below the level of resolution cannot be excluded.

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Male Pseudohermaphroditism in a Child with Down’s Syndrome*

Summary. A case of male pseudohermaphroditism in a child with Down’s syndrome is reported. The patient had infantile testis, ambiguous genitalia, and no apparent internal female genitalia indicating a failure of either gonadal function or end-organ response.

Down’s syndrome, the most common of the chromosomal abnormalities, has been reported in association with a number of seemingly unrelated sex chromosome aneuploidies, of which the most frequent is the double euploid 48,XXY,+G (Hamerton, 1971). Although there has been one report of a female with Down’s syndrome and congenital adrenal hyperplasia (Srivuthana et al, 1971) we are unaware of any published cases of Down’s syndrome associated with male pseudohermaphroditism. We will describe such a child.

Case Report

The proposita is the first child of 25-year-old parents. She was born at term by a spontaneous vaginal delivery after a four-hour labour and weighed 3115 g. The