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Trisomy 4q32→4qter due to a maternal 4/21 translocation

Summary. The case is described of a malformed girl with partial trisomy for a segment of the long arm of chromosome 4 (4q32→4qter) due to an unfavourable segregation of a maternal reciprocal translocation t(4;21) (q32q22). The clinical comparison between the child and patients previously described by other authors does not suggest the existence of a syndrome associated with trisomy 4q+.

To date there are only four clinical descriptions of partial trisomy for the long arm of chromosome 4, the chromosome being identified by autoradiography (Surana and Conen, 1972) or by banding techniques (Francke, 1972; Chapelle et al, 1973; Schrott et al, 1974). Recently another two cases of trisomy 4q+ in a family with a translocation t(4q;18q) have been published (Knörr-Gärtner et al, 1974) but no report of the clinical picture was included. We would like to describe a patient who presents a peculiar phenotype and who is affected by trisomy 4q+ as a consequence of a familial translocation t(4q;21q).

Case report

The proposita, born 25 March 1966, was the result of a fourth pregnancy of a mother aged 40 and a father aged 43. The previous three pregnancies (Fig. 1) resulted in the birth of a normal healthy female, now aged 21, a healthy male, now aged 13, and a spontaneous abortion which occurred during the second month of pregnancy in 1965. The patient was born at term after an uneventful pregnancy and from slight recurrent vaginal bleeding. Birth weight was 9500 g (below the 10th centile); length and head circumference were not registered. The neonatal period was uneventful except for problems of nutrition, growth, and psychomotor development. At 6 months her weight was 3500 g and head circumference 36 cm; at 13 months she was able to sit, weight was 5800 g, and head circumference 39 cm. She took her first steps at 26 months; weight was 8700 g and head circumference 43 cm. She is now 8½ years and weighs 15 kg and is 112 cm tall with a head circumference of 45 cm. She does not speak and her only expressions are guttural sounds and hand gestures. Her posture and coordination are equivalent to that of a child of 2 years (Fig. 2), while her language and social growth are below this level. Discrete, generalized muscular hypotonia is present and both deep and superficial reflexes are increased. The head is small. Facies are narrow and consist of prominent frontal bossing, low hairline, small and hollowed eyes, depressed bridge of the nose which shows a large bottom and flat top, large philtrum, and

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micrognathia. An arched palate, malaligned teeth, large and prominent ears with mild peaking of the antihelix and a backward slant are also present. The third, fourth, and fifth toes are bilaterally curved and the hands show a simian crease and an absence of the digital triradius bilaterally. On the right hand, fingertips show a whorl on the 1st, 2nd, 3rd, and 4th fingers and an ulnar loop on the 5th. On the left hand the fingertips show a whorl on the 1st, 3rd, and 4th fingers, a radial loop on the 2nd, and an ulnar loop on the 5th. Routine laboratory data are normal and include: complete blood count, urine analyses, blood glucose level, serum electrolytes, serum proteins and immunoglobulins, blood urea nitrogen and creatinine, urine screening for amino acids and mucopolysaccharides, EEG and ECG. Radiology showed mild lumbar scoliosis and a bilateral 13th rib. The bone age is slightly retarded (corresponding to 7 years). Intravenous pyelography is normal.
**Cytogenetic studies**

Chromosome preparations, obtained by standard techniques from peripheral blood cultures, were treated with the reverse-staining Giemsa method (Dutrillaux and Lejeune, 1971) and the BUDR-acridine orange staining method (Dutriullass et al., 1973).

I.1, I.2, II.5, III.I.1, and III.2 were examined and found to have normal karyotypes. In all cells examined in II.6 (the mother of the proposita) there were 46 chromosomes. Only one normal chromosome 4 was present, the other one was deleted at q32. The missing segment was translocated on to the long arm of one chromosome 21. The positive R-band at the end of the deleted chromosome 4 could be band—4931. We cannot exclude, however, that this band could be the translocated terminal R-band of chromosome 21 (Figs. 3a and 3b). Using the international chromosome nomenclature (Paris Conference, 1971) the mother’s karyotype is: 46,XX,t(4;21)(q32q22).

The karyotype of the proposita (III.3) is characterized by two normal chromosomes 4 and one normal chromosome 21 while the other one is the same translocated chromosome observed in the mother (Fig. 4). The karyotype is then: 46,XX,—21, +der(21),t(4;21)(q32q22)mat.

The other members of the family were not available for testing.

**Discussion**

The clinical features of our patient show some data in common with those previously reported—that is, low birth weight, which was in our case a sign of slowed intrauterine development, ear malformation, microcephaly, micrognathia, mental retardation, growth impairment, and simian crease—but these anomalies can also be easily found in other chromosomal abnormalities.

It is not possible, to date, to conclude the existence of a syndrome associated with partial trisomy 4q+. The presence of various additional features and the lack of strict similarities of anomalies noticed in the patients can be explained by a different chromosomal segment being translocated—that is, different breakage point and/or by different chromosomes being involved in the translocation.

One point we would like to stress is that in our case the trisomic segment q32--qter is the smallest one described to date. It should be useful then to compare clinically other subjects with identical cytogenetic anomalies.

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