Note added in proof

Since submission of this paper, Roberts and Duckett (1978) have reported a live born female infant with trisomy 16p. They also reviewed published reports. Their report modifies the opinion given in the last paragraph of this paper in which it is proposed that triplication of the short arm of chromosome 16 may be incompatible with live birth and may be responsible for fetal loss in full trisomy 16.

Reference


Two children with partial trisomy for 7p

SUMMARY A second family in which a balanced translocation between 7p and 22q is segregating is described. The clinical features of 2 children with a resulting partial trisomy for 7p are described and compared with the previously described case.

The report by Larson et al. (1977) of a family in which a 7p;22q translocation was segregating prompts us to report a similar family with 2 further children with the unbalanced karyotype and partial 7p trisomy.

Case reports

CASE 1

This case (PRU 3373) was a baby girl born in 1966 after a normal pregnancy of 40 weeks' gestation, birthweight 2.30 kg. She was hypotonic with a weak cry and multiple anomalies were noted at birth: the skull was elongated with a widely separated metopic suture; she had microphthalmos, the eyes having a mongoloid slant; ears were low set and there was micrognathia; a systolic murmur was present. There were a number of skeletal abnormalities which included arachnodactyly, flexion deformity of the wrists, talipes calcaneo-valgus, and widely separated 1st and 2nd toes. A single palmar crease was present unilaterally. She failed to thrive and died aged 8 weeks.

At necropsy, in addition to the features noted above, she had hydrocephalus and microgyria, a hypertrophic left ventricle with persistent ductus arteriosus, and some aortic stenosis proximal to the ductus. Both kidneys were small with cysts.

Chromosome analysis in 1966 using Orcein stain showed a chromosome complement of 46,XX,Gq+.

CASE 2

This case (PRU 3373), the brother of case 1, was born in 1968 at 38 weeks' gestation, birthweight 3.94 kg. His mother had had influenza at the beginning of the pregnancy. Delivery was in breech presentation and a left brachial plexus lesion was noted almost immediately. He looked similar to his sister, case 1.

When seen at the age of 6 months (by Dr S. M. Kohlinsky), his head circumference was 44 cm (+1 SD) and the occiput was prominent. He had a long face with narrow palpebral fissures, especially on the left, with a slight mongoloid slant to the eyes and epicantlic folds. The nasal bridge was broad and flat, the palate was high and arched, and the maxilla prominent with a thick lower lip. Slight weakness of the left arm and shoulder was present.

The cardiovascular, respiratory, and genitourinary systems appeared normal. Developmental level was 2½ to 3 months.

Because of his coarse facies, his urinary mucopolysaccharides were measured and found to be normal. Peripheral blood chromosome analysis using Orcein stain showed a chromosome complement of 46,XY,Gq+, similar to his sister, case 1.

He has remained extremely retarded and at the age of 9 years can sit unaided but cannot stand or walk. He says no words and is unable to feed himself. He has had one fit, but is not currently on medication.

Family studies

The mother (born in 1936) has rheumatoid arthritis and the father (born in 1926) is healthy and they are not close relations. The mother had 2 miscarriages, one at 14 and the other at 16 weeks' gestation, but 2 healthy sons were born in 1970 and in 1973. In 1966, peripheral blood chromosome analysis using Orcein stain showed the father to have a chromosome complement of 46,XY,Gq+ and in 1970 one of the healthy sons was found to have this karyotype also. The father was an only child and no further family studies have been attempted.

It seemed most likely that a balanced translocation was involved, but it was impossible to distinguish the karyotypes of the phenotypically normal family members from those of the abnormal before the development of banding techniques. Early attempts at banding in 1973 were unsatisfactory and for this reason the family decided against prenatal chromosome analysis during the final pregnancy. Chromosome analysis after birth showed the infant to have a normal male chromosome complement 46,XY.
EMI trisomy atrophy. Profound motor and other two problem usually more of degree of the brach development resulting in the two families. Involving 22q, the situation arose from the reciprocal translocation involving 22q, and in both it was paternal in origin, though, in general, derivative chromosomes are more usually maternally transmitted. Breakpoints in the two families are not identical, 7p21 in Larson’s family, and 7p15 in this family.

The clinical features have certain similarities. Profound motor and mental retardation is the major problem and the necropsy study on case 1 showed a degree of hydrocephalus and microgyria, while an EMI scan on Larson’s case showed hypoplasia and atrophy of the brain.

Other common features, such as epicanthic folds and a high arched palate, are non-specific, and our case 1 had skeletal abnormalities not noted in the other two patients. Thus, the most marked effect of the trisomy for 7p seems to be a severe failure in cerebral development resulting in gross retardation, more severe than that found in many other autosomal aneuploidies. In the family described by Larson, however, three unstudied adults were mentally retarded, and therefore thought to have the duplication, though in these cases the effect seemed less severe.

Children with deletions of 7p have been described (reviewed by McPherson et al., 1976) and the most striking clinical feature is craniosynostosis, though the breakpoints appear to be the same as in the present family. Thus, there is no evidence of type and contratype for abnormalities of this chromosome arm.

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References


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A case of Turner’s syndrome with familial balanced translocation t(1;2)(q32;q21)mat

SUMMARY The first case of Turner’s syndrome with the familial translocation not involving the X chromosome is described. The patient had a number of clinical signs of Turner’s syndrome and her karyotype was 45,X,t(1;2)(q32;q21)mat. Though it is speculated that the altered structure of a chromosome may influence meiotic disjunction of a non-homologous chromosome, our case suggests that there may be no relationship between the two chromosomal abnormalities.