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Fig. (a) Partial karyotype of father showing balanced reciprocal translocation; (b) partial karyotype of case 2 showing partial trisomy for 7p.

In 1974, re-evaluation of the family using G-banding identified the translocation. Balanced family members had chromosome complements 46,XY,t(7;22)(p15;q13) (Fig. a). Case 2 had the unbalanced karyotype 46,XY,der(22),t(7;22)(p15; q13)pat (Fig. b) and therefore had a duplication (partial trisomy) of the distal part of 7p. Case 1 can be assumed to have had the same unbalanced karyotype, since she had the derived chromosome 22.

Discussion

The only other case of 7p duplication (partial trisomy) of which we are aware is that described by Larson et al. (1977), and there are strong similarities between the two families. In both, the unbalanced situation arose from a 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.
To our knowledge, 10 cases of Down's syndrome coexisting with a familial translocation involving chromosomes other than 21 have been reported (Hamerton et al., 1963; Tenconi et al., 1974; Oikawa et al., 1977). In addition, 6 cases of Klinefelter's syndrome and a case of 18 trisomy syndrome with a familial translocation involving different chromosomes have also been described (Chryso stomidou et al., 1971; Sparagana and Smith, 1975).

Having carried out cytogenetic studies on 146 patients with aneuploidy, 9 patients with balanced or unbalanced familial translocations and all of their parents, we found only one case of aneuploidy coexisting with a familial balanced translocation. This patient's karyotype was 45,X,t(1;2)(q32;q21)mat.

The purpose of this paper is to present this case and to discuss whether the familial balanced translocation might have influenced non-disjunction of the X chromosome in this case.

**Case report**

The proband, weight 2100 g and length 43 cm, was born after a gestation of 39 weeks. The mother was 28 years old and the father was 34. This was the mother's second pregnancy and it was complicated by a threatened abortion which was treated with bed rest and oestrogenic agents. The first pregnancy ended in a spontaneous abortion in the first trimester. The infant was noted to have epicanthic folds, a short nose with depressed nasal bridge, short neck, haemangioma on her trunk, short fingers with hypoplastic nails, unilateral simian crease, and lymphoedema of the dorsum of the hands and feet.

At 5 months of age, examination of lymphocytes and skin fibroblasts with Giemsa banding showed the karyotype 45,X,t(1;2)(q32;q21) (Fig.). The mother's karyotype was 46,XX,t(1;2)(q32;q21). The father and the maternal grandparents had normal karyotypes. The patient and both parents were Xg(a)+.

At present, she is 13 months of age; her weight is 8860 g which is normal for her age, but her length is 69.5 cm (under 2 SD). Developmental milestones were normal up to the age of 13 months.

**Discussion**

A number of cases of Down's syndrome who had an inherited balanced translocation not relating to chromosome 21 have been reported. Some authors suggested a correlation between a translocation not involving chromosome 21 and non-disjunction of 21 (Hamerton et al., 1963; Oikawa et al., 1977). Other authors, however, have questioned this suggestion (Tenconi et al., 1974). The frequency of balanced translocations among Down's syndrome cases was similar to that in normal newborn infants (Lundsteen et al., 1974), suggesting that 21 trisomy and familial translocation in the same person only occur by chance. In the case of Klinefelter's syndrome associated with D/D translocation, Sparagana and Smith (1975) also suggested that there was no...
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Table Cases of familial translocations with aneuploidy not involving translocated chromosomes

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>No of reported cases</th>
<th>Expected frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td>Down’s syndrome</td>
<td>10 cases</td>
<td>1/350,000</td>
</tr>
<tr>
<td>Klinefelter’s syndrome</td>
<td>6 cases</td>
<td>1/500,000</td>
</tr>
<tr>
<td>18 trisomy syndrome</td>
<td>1 case</td>
<td>1/2,500,000</td>
</tr>
<tr>
<td>Turner’s syndrome</td>
<td>1 case</td>
<td>1/340,000</td>
</tr>
</tbody>
</table>

relationship between the two chromosomal aberrations.

To our knowledge, the present case is the first report of Turner’s syndrome with a familial balanced translocation not involving the X chromosome. Turner’s syndrome occurs in about 1 in 6800 live births. Cases with balanced translocations occur in about 1 in 500 live births and most cases originate from familial translocation carriers (Bochkov et al., 1974; Hamerton et al., 1975; Nielsen and Sillesen, 1975). Hence, the expected likelihood of both abnormalities occurring in the same female should be about 1 in 3,400,000 live births through chance alone. From a similar calculation the expected occurrence of Down’s syndrome, Klinefelter’s syndrome, and 18 trisomy syndrome with a familial balanced translocation should be about 1 in 350,000, 1 in 500,000, and 1 in 2,500,000, respectively. When the number of reported cases of Down’s syndrome with familial translocations are taken as a standard, the number of reported cases of Turner’s syndrome and other aneuploidies with familial translocation in publications seem to match the expected frequency (Table). In addition, non-disjunction of the X chromosome in Turner’s syndrome generally occurs in paternal gametogenesis (Sanger et al., 1971). Though we could not determine the origin of the X chromosome, it is likely that in this case the non-disjunction also occurred in paternal gametogenesis. There seems to be no relationship between the two chromosomal abnormalities in the present case.

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References


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De novo interstitial deletion del(1)(p21p32)

SUMMARY A girl aged 14 years 9 months, overweight, with severe psychomotor retardation, short stature, a sheep-like face, malformed ears, skeletal and dermatoglyphic abnormalities, and partial deletion of the short arm of chromosome 1 is presented. The karyotype was 46,XX,del(1) (qter→p22:p32→pter).

Structural anomalies of chromosome 1, compatible with the development to term of the fetus, occur quite rarely. Until now, 5 partial trisomies (Neu and Gardner, 1973; Van den Berghe et al., 1973; Norwood and Hoehn, 1974; Garver et al., 1976;
A case of Turner's syndrome with familial balanced translocation t(1;2)(q32;q21)mat.

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