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.

We thank Dr Takeki Hirano, Department of Pediatrics, University of Tsukuba, for his careful reading of the paper.

IKUKO KONDO, HIDEO HAMAGUCHI, AKIO MATSUURA, HACHIRO NAKAJIMA, AKIRA KOYAMA, AND HITOSHI TAKITA
Department of Human Genetics, Institute of Basic Medical Sciences and Department of Pediatrics, Institute of Clinical Medicine, University of Tsukuba, Ibaraki-ken;
Department of Pediatrics, Takayama Red Cross Hospital, Gifu-ken; Department of Forensic Medicine, Tokyo Medical and Dental University, Tokyo; and the Special Reference Laboratory, Tokyo, Japan.

References


Requests for reprints to Dr Ikuko Kondo, Department of Human Genetics, Institute of Basic Medical Sciences, University of Tsukuba, Sakuramura, Niihari-gun, Ibaraki-ken 300-31, Japan.

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;
Palmer et al., 1977) and 3 partial deletions (Turleau et al., 1974; Koivisto et al., 1976; Garver et al., 1976) of the long arm of this major chromosome have been reported.

Our report presents for the first time the clinical syndrome caused by a structural anomaly on the short arm of chromosome 1, which occurred de novo.

Case report

The patient, a female, born in 1962, has been under medical care from the age of 9 months for severe psychomotor retardation. The parents were not consanguineous and, at birth, the father was 28 and the mother 25 years of age. The proband, their first child, was born at 7 months and 2 weeks' gestation after an uneventful pregnancy. At birth the baby was 48 cm long and weighed 2000 g.

At the time of the first medical examination, the child was 63 cm tall and weighed 9 kg. Striking peculiarities were noticed: the face was round with a prominent, bulbous nose, a large, half-open mouth, ridged tongue, microtia, aplasia of the preauricular structures, and malformations of the pinnae (smaller auricles with rolled-over helix covering the antihelical fold, large lobulus), short neck, convex thorax, widely spaced nipples, long thin fingers, clinodactyly of the 5th finger and toe, enlargement of the first interdigital space of the foot, genu valgum, and open anterior fontanelles (2/2 cm). The child made only slow movements, and did not sit, play, or talk.

At the ages of 5, 9, and 14 years (in 1968, 1971, and 1977, respectively) the patient was re-examined. There was little growth in stature and she was over weight (Fig. 1). The last time the patient was seen, at the age of 14 years 9 months, she showed grotesque thickening of the features of the face (sheep-like), predominantly facial and truncal distribution of the adiposity, and mental retardation (IQ = 50). Signs of puberty present were pubic and axillary hair and 2nd degree Tanner breast development (Fig. 2, 3, 4).

CLINICAL INVESTIGATIONS

X-ray examination showed thickened cranial bones, especially in the vertex area; sella turcica with moderate hypotrophy of the patrulater lamella; dextroconvex dorsal scoliosis; distortions in the distal half of the metacarpals and phalanges; clinodactyly of the 5th finger and toe, and decalcification of the leg bones.

DERMATOGlyphS

Digital dermatoglyphs

The digital dermatoglyphs are shown in the Table.

Decadactyl formula, A1 L8 W1; TRC 74.

Palmar dermatoglyphs

Right hand: formula a1, b8, c11 d11 t(11); ab=11, bc=7, cd=30; patterns Hy=Lu, Th=Ar, Iio, IIIo, IVo; palmar sulcus type II, atd 73°. Left hand: formula a1, b6, c0, dlo, t'(11), ab=28; patterns Hy=M, Th=Ar, Iio, IIIo, IVo, normal flexion lines, atd=53°.

The total number of digital crests was at the lower limit of the normal control mean (-1SD). The triradius palmar line (A) opened at the root of the thumb and the axial triradius line (T) opened in the 2nd interdigital spaces of both hands. Ulnar loops on the right hand in the hypothenar area, associated with an increased atd angle, and a transitional form of palmar sulcus were present.

Table Digital dermatoglyphs

<table>
<thead>
<tr>
<th></th>
<th>I</th>
<th>II</th>
<th>III</th>
<th>IV</th>
<th>V</th>
<th>Formula</th>
</tr>
</thead>
<tbody>
<tr>
<td>Right hand</td>
<td>W</td>
<td>Lu</td>
<td>Ar</td>
<td>Lu</td>
<td>Lu</td>
<td>pattern L3 A1</td>
</tr>
<tr>
<td></td>
<td>13</td>
<td>3</td>
<td>0</td>
<td>13</td>
<td>14</td>
<td></td>
</tr>
<tr>
<td>Left hand</td>
<td>Lu</td>
<td>Lu</td>
<td>Lu</td>
<td>Lu</td>
<td>Lu</td>
<td>pattern L5</td>
</tr>
<tr>
<td></td>
<td>4</td>
<td>6</td>
<td>6</td>
<td>7</td>
<td>8</td>
<td></td>
</tr>
</tbody>
</table>
Fig. 2  The patient at the age of 9 years 3 months (left) and 14 years 9 months (right).

Fig. 3  Facies of patient.
CYTOGENETIC FINDINGS

An X chromatin study showed 18% positive cells. There was an interstitial deletion of the short arm of an A1 chromosome. The chromosomal formula established with the help of GTG bands was 46, XX, del(1)(p22p32) or, in more detail, 46,XX, del(1)(qter→p22::p32→pter) (Fig. 5, 6). Cytogenetic findings in the younger sister and the parents of the patient were normal.

Discussion

Partial monosomies and trisomies, through the gene imbalance that they induce, are the most frequent causes of spontaneous abortion and interruption of pregnancy. According to Boué (1977) only 6.7% of products of conception with structural abnormalities
anomalies of the chromosomes survive the period of embryonal development. Most of them are eliminated before the mother is aware that she is pregnant and few embryos are brought to term. Yet, there is enough evidence to suggest that a fetus may survive even when over 5% of the haploid genome is triplicated, whereas small autosomal deficiencies are not compatible with complete embryonal development. Norwood and Hoehn (1974), in describing a trisomy for a segment of the long arm of an A1 chromosome (2 to 4 bands or 3-2% of the haploid genome), studied pregnancies in bearers of balanced translocations involving this arm. They found an increased incidence of spontaneous abortions (7/34) and fetuses born dead (2/34).

In our case, the deletion of an area representing approximately 0.98% of the haploid genome allowed the pregnancy to progress to 7 ½ months, and the birth and survival for as long as 14 years 9 months of a child with several abnormalities. Most of these abnormalities (severe mental retardation, small stature, facial dysmorphism, and anomalies of the auricles and extremities) are often encountered in chromosomal disorders. Their association may be characteristic, but confirmation will be obtained only after further similar cases have been reported. The general aspect of the face, prominent bulbous nose, large mouth, retrognathism, clinodactyly of the 5th finger and toe, misshapen, low set ears, and dextroconvex scoliosis are similar to the ‘sheep-like face’ syndrome first described by Wiedemann and Tolksoff (1973) in a 10-year-old girl and encountered again by Schönenerben and Habedank (1974) in 3 other children with severe mental retardation. While in the first case, using traditional staining methods, a chromosomal anomaly was detected (karyotype 46,XX,2q+), the latter 3 cases failed to show cytogenetic anomalies though banding methods were used.

We thank Dr Martha Ciocirnache for the dermatoglyphic examination.

M. Bene, Daniela Duca-Marinescu, Doina Ioan, and C. Maximilian
Children’s Hospital, Brasov; and the Institute of Endocrinology, Bucharest, Romania

References

Requests for reprints to Dr C. Maximilian, Institute of Endocrinology, 71279 Bd. Aviatorilor 34-36, R-76 134 Bucharest, Romania.