A case of partial trisomy 3q

Summary. A case report on an infant with trisomy of the long arm of chromosome No. 3 is presented. The condition has not been described previously.

A case of presumptive trisomy 3 was reported by Sinha (1968) and a few cases of partial trisomy 3 have been reported (Clarke et al, 1964; Walzer et al, 1966; Aarskog, 1969; Rethore et al, 1972; Sachdeva, Smith, and Justice, 1974). Among these cases, three cases of Rethore and one case of Sachdeva were identified as the duplicated portion of the short arm of No. 3 chromosome by chromosome banding methods.

Case report

The proposita (Fig. 1a and b), a 1-month-old female, was born to a 26-year-old mother and a 28-year-old father. Gestation was full term with polyhydramnios. The parents were phenotypically normal but many abortions, stillbirths, and congenital malformations were found among the maternal relatives. A pedigree of the family is shown in Fig. 2 and the proposita is indicated by an arrow.

Birthweight was 2630 g, length was 48.5 cm, and head circumference was 41 cm.

The clinical features were as follows: square-shaped face, hypertelorism, small, malformed ears, prominent nasal bridge, cleft palate, micrognathia, short, webbed neck, wide-set nipples, camptodactyly, clinodactyly of the 5th fingers, hypoplastic nails, hypoplastic dermal ridge. She died at 32 days of age and necropsy showed hypoplastic cerebellum, cystic dilatation of 4th ventricles, renal cortical cyst, bicornuated uterus, and double vagina.

Cytogenetic studies

Chromosomal studies were performed on a culture of peripheral blood. The karyotype of the proposita was 46,XX,2p+ and the mother's was 46,XX,2p+3p or q-. This karyotype suggested that the proposita was partial.
Case reports

trisomy 3 and the mother was a balanced 2/3 translocation carrier.

Giemsa banding studies were then performed. The method was as follows: after flame-drying procedure, the preparations were treated with 2xSSC at 60°C for 60 minutes; rinsed with isotonic saline and treated with 0.25 per cent trypsin for 5–10 s at room temperature; rinsed twice with isotonic saline and stained for 10 minutes with Giemsa (MERCK) diluted to 2% with pH 6.8 phosphate buffer. Giemsa banding studies showed that the points of exchange were located at 2p25 and 3q21 bands (Fig. 3). The short-system designation (Paris

46,XX,rcp(2:3)(2qter→2p25::3q21→3qter;
   2pter→2p25::3q21→3pter)

Fig. 3. Partial karyotype of the carrier mother (G-banding). Points of exchange are indicated either by arrows (derivative chromosomes) or solid line (normal counterparts).
Case reports

Disclosure of the chromosome finding in this family would be,
Proposita 46,XX,der(2),t(2;3)(p25;q21)mat
Mother 46,XX,rcp(2;3)(p25;q21)
In the same studies of available relatives, two other translocation carriers rcp(2;3)(p25;q21) were found (III.6, IV.12).

Discussion
The translocation rcp(2;3)(p25;q21) was found in at least two generations. There is a possibility that the translocations of the carriers (III.6, III.12) have been inherited from their mother (II.2) but study was not possible.

Clinical features of trisomy 3, trisomy 3p, and trisomy 3q (present case) are similar to each other but some differences are recognized in the necropsy findings. Both hypoplastic cerebellum and renal cortical cyst were found in trisomy 3 and trisomy 3q (present case); however, they were not observed in trisomy 3p. The main clinical features of these cases and the present case are summarized in the Table. More cases with detailed clinical descriptions as well as cytogenetic findings are needed to establish the two clinical syndromes, trisomy 3q and trisomy 3p.

As the mother of the proposita was a balanced 2/3 translocation carrier, the proposita was trisomic for the segment to the long arm of No. 3 chromosome (3q21→3qter) and also deficient for the terminal portion of the short arm of No. 2 chromosome (2p25→2pter). It may be that some clinical findings are the result of this minute loss of chromosome material from the short arm of No. 2 chromosome. Walzer (1966) reported 6 cases of partial trisomy 3. They were accompanied by deficiency of the terminal portion of the short arm of No. 5 chromosome. In these cases a cat cry was not noted. Aarskog (1969) described 2 cases of partial trisomy 3 that were accompanied by deficiency of the long arm of No. 18 chromosome. He reported that these cases resembled the 18q— syndrome. In cases of double autosomal aneuploidy, these usually have the features of both conditions though the more serious condition usually masks the less serious (Hamerton, 1971). In partial trisomy which is accompanied by deficiency of other chromosomes, the clinical features may be affected by the latter.

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REFERENCES

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TABLE

MAIN CLINICAL FEATURES OF PRESENT CASE, TRISOMY 3p AND TRISOMY 3

<table>
<thead>
<tr>
<th>Trisomy 3q</th>
<th>Trisomy 3p</th>
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<tbody>
<tr>
<td>Died at 32 days</td>
<td>Died at 32 days</td>
<td>Died at 32 days</td>
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<tr>
<td>Hypoplastic cerebellum</td>
<td>Hypoplastic cerebellum</td>
<td>Hypoplastic cerebellum</td>
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<tr>
<td>Cystic dilatation of 4th ventricles</td>
<td>Cystic dilatation of 4th ventricles</td>
<td>Cystic dilatation of 4th ventricles</td>
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<tr>
<td>Square-shaped face</td>
<td>Square-shaped face</td>
<td>Square-shaped face</td>
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<tr>
<td>Hypertelorism</td>
<td>Hypertelorism</td>
<td>Hypertelorism</td>
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<td>Malformed ears</td>
<td>Malformed ears</td>
<td>Malformed ears</td>
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<tr>
<td>Cleft palate</td>
<td>Cleft palate</td>
<td>Cleft palate</td>
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<tr>
<td>Micrognathia</td>
<td>Micrognathia</td>
<td>Micrognathia</td>
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<td>Prominent nasal bridge</td>
<td>Prominent nasal bridge</td>
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<tr>
<td>Short neck</td>
<td>Short neck</td>
<td>Short neck</td>
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<tr>
<td>Camptodactyly</td>
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<td>Camptodactyly</td>
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<td>Congenital heart disease</td>
<td>Congenital heart disease</td>
<td>Congenital heart disease</td>
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<td>Renal cortical cysts</td>
<td>Renal cortical cysts</td>
<td>Renal cortical cysts</td>
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<tr>
<td>Bicornuate uterus</td>
<td>Bicornuate uterus</td>
<td>Bicornuate uterus</td>
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<tr>
<td>Hypoplastic dermal ridge</td>
<td>Hypoplastic dermal ridge</td>
<td>Hypoplastic dermal ridge</td>
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<tr>
<td>Other signs:</td>
<td>Other signs:</td>
<td>Other signs:</td>
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<tr>
<td>High frequency of whorls on finger</td>
<td>High frequency of whorls on finger</td>
<td>High frequency of whorls on finger</td>
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<tr>
<td>Oesophageal atresia, mesenterium</td>
<td>Oesophageal atresia, mesenterium</td>
<td>Oesophageal atresia, mesenterium</td>
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<tr>
<td>communis</td>
<td>communis</td>
<td>communis</td>
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<tr>
<td>Low birthweight, congenital deformities of lung and</td>
<td>Low birthweight, congenital deformities of lung and</td>
<td>Low birthweight, congenital deformities of lung and</td>
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<tr>
<td>kidneys, undescended testis, small penis</td>
<td>kidneys, undescended testis, small penis</td>
<td>kidneys, undescended testis, small penis</td>
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<tr>
<td>Simian lines</td>
<td>Simian lines</td>
<td>Simian lines</td>
</tr>
<tr>
<td>Small eyes, low-set ears, inguinal hernias, Meckel's diverticulum, malrotation of gut, undescended testis</td>
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</tbody>
</table>

46,XY,inv(7;3)(q31;p21)(26)
46,XY,inv(7;3)(q31;p21)(26)
46,XY,inv(7;3)(q31;p21)(26)

46,XY,der(12),t(3p12q)mat
46,XY,der(12),t(3p12q)mat
46,XY,der(12),t(3p12q)mat

46,XY,der(2),t(2;3)(p25;q21)mat
46,XY,der(2),t(2;3)(p25;q21)mat
46,XY,der(2),t(2;3)(p25;q21)mat

46,XY,inv(7;3)(q31;p21)(26)
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46,XY,inv(7;3)(q31;p21)(26)

46,XY,inv(7;3)(q31;p21)(26)
46,XY,inv(7;3)(q31;p21)(26)
46,XY,inv(7;3)(q31;p21)(26)

46,XY,der(2),t(2;3)(p25;q21)mat
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46,XY,der(2),t(2;3)(p25;q21)mat
46,XY,der(2),t(2;3)(p25;q21)mat

46,XY,inv(7;3)(q31;p21)(26)
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46,XY,inv(7;3)(q31;p21)(26)
A case of twin chimerism

Summary. A case of twin chimerism is presented and shown by cytogenetic studies, red cell grouping, and white cell HL-A typing. The sex of each twin is confirmed by examination of buccal smears and their chimeric state is confirmed by non-reactivity in the mixed lymphocyte culture system. The results of these investigations are discussed.

Chorionic vascular anastomoses are usually present between dissimilar bovine twin embryos, and when Owen (1945) published his work on 'Immunogenetic consequences of vascular anastomoses between bovine twins' it was realized that primordial red cells belonging to one twin take root in the other twin. This occurrence is rare in the human situation. The first example of human chimerism in twins was reported by Dunsford et al (1953). It was discovered when the propositus Mrs McK was found to have a mixture of two kinds of blood. Since this first case there have only been 12 further examples reported, 8 of these cases of twin chimeras are excellently summarized by Race and Sanger (1968). Gundolf described a Danish pair (1970), Ducos et al (1970) a French pair, Crookston et al (1970) a Canadian pair, and Kasser and Nennstiel (1971) a German pair.

The case to be presented differs from the majority of previously reported cases in that it was discovered after cytogenetic studies and not through blood grouping. As there was no mixture involving the ABO blood group system this example of twin chimerism could easily have been missed.

Case report

AK and PK are a pair of dissimilar, unlike sexed twins. At the age of 2 months the girl, AK, was suspected of having hypothyroidism; this was confirmed at 3 months. She responded to appropriate treatment and is now euthyroid. The boy, PK, has shown a normal development pattern, apart from bearundescended testes, which are still impalpable at the age of 21 months. In addition to the hypothyroidism AK was noticed to have an odd facies, and for this reason a chromosome analysis was carried out.

Cytogenetic studies

Chromosome studies from short-term lymphocyte cultures were carried out on three separate occasions on the twins, and once on their elder female sib, and parents. Chromosome analyses of the parents and sib yielded normal results. A total of 100 cells from each twin were karyotyped, and the results of these studies can be seen in Table I.

<table>
<thead>
<tr>
<th>TABLE I</th>
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<tr>
<td><strong>KARYOTYPE ANALYSIS AND BARR BODY STATUS</strong></td>
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<tr>
<td>No. of Cells Counted</td>
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<tr>
<td>AK</td>
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<tr>
<td>PK</td>
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</table>

Buccal smears were made on several different occasions to try to ascertain with certainty the sex of each twin.

Red cell grouping: secretor status and HL-A typing

Standard serological techniques were used in the determination of the red cell groups and secretor status. HL-A typing of the lymphocytes was carried out using the lymphocyte cytoxicity technique. The results are shown in the Fig.

Despite the extensive red cell grouping carried out, a chimeric mixture showing mixed field agglutination was only obtained with anti-s sera. Separation of the two populations was attempted with difficulty, as the anti-s serum only reacted by the indirect antiglobulin technique. 26% of AK's red cells and 10% of PK's red cells had the genotype Sa. 74% of AK's and 90% of PK's red cells had the genotype SS. Both twins were shown to have two populations of lymphocytes which could be separated into major (strong reacting) and minor (weak reacting) groups. Taken together these two populations showed all the HL-A antigens detectable in the mother and father, giving further confirmation of