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Multiple congenital defects associated with trisomy for long arm of No. 4

Summary. The clinical and cytogenetic findings of a male infant with multiple congenital anomalies and trisomy for the distal third of the long arm of No. 4 are described. The abnormal chromosome was inherited from the mother who had a balanced translocation, t(4;9)(q31;q34). Trisomy for the long arm of No. 4 has previously been described in only 3 patients.

Case report
The propositus was born to a 31-year-old woman and her 44-year-old husband after an uneventful pregnancy and a normal delivery. The infant was thought to be 12 days past maturity by dates, though birthweight was only 2722 g. This was the mother’s fourth pregnancy (Fig. 1). The first had resulted in the birth of a ‘deformed’ male who died almost immediately. Necropsy was not performed.

The propositus was noted at birth to have a skin-covered supra-umbilical exomphalos. He had a ‘strange’ face with a suggestion of low set ears. Head circumference was 32 cm. At 1 month inffrequent motions led to rectal biopsy and the identification of Hirschspring’s disease, with aganglionosis extending to at least 10 cm of bowel.

FIG. 1. Pedigree of family showing segregation of translocation, t(4;9)(q31;q34). Year of birth is given.
Fig. 2. Propositus at 1 year 10 months.

Proximal to the anal margin. At 10 months he was noted to be grossly mentally retarded with a performance at the 3 to 4 months level. Head circumference was then well below the third centile. Skull X-ray and bone age were within normal limits. Operation for closure of the exomphalos was performed at 10 months. Mental subnormality and peculiar facial appearance led to chromosome analysis as an aid to diagnosis and genetic counselling.

On referral for chromosome analysis at 1 year 10 months it was noted that his head was very small and moderately plagiocephalic, with protrusion of the right occiput; head length 14 cm, breadth 11 cm, and circumference 43.5 cm. The anterior fontanelle was closed. Facial features were unlike those of his parents but not obviously dysmorphic (Fig. 2). Mental subnormality was obvious and severe. Though the nose had a rather wide base (inner canthic distance 2.1 cm) eyes, ears, tongue, and roof to the mouth were unremarkable. Microglossia was absent. Feet and hands were normal, as was muscle tone. The right testis was palpable and the left was in the inguinal canal. A cardiac murmur was not heard. Dermatoglyphs (Table I) were normal. A colostomy was present. Abnormality of the renal tract was not observed on later intravenous pyelogram study.

**Cytogenetic studies**

Thirty cells were counted from the lymphocyte chromosome preparations of the propositus and were stained with lacto-orcein. All had 46 chromosomes. Only 14 members of the C group were present and there was an extra chromosome in group B (Fig. 3). No other chromosome abnormality was found. The karyotype of each parent appeared normal when prepared by a conventionally orcein stained method.

Lymphocyte preparations of the propositus and both parents were 'banded' with 0.25% trypsin and stained with Leishman's (Seabright, 1971). The banded karyotype of the propositus showed the extra chromosome in group B to be a No. 9 with additional material on the long arm. Mother's banded karyotype (Fig. 4) clearly indicated a reciprocal translocation between the long arm of a No. 4 and a No. 9, t(4;9)(q31;q34). The father's karyotype was normal. The propositus is trisomic for about the distal third of the long arm of No. 4 and monosomic for the terminal part of the long arm of No. 9.

Further members of the family were examined for chromosome complement (Fig. 1) on banded preparations.

**Discussion**

Partial trisomy for the long arm of chromosome No. 4 (4q+) has previously been unequivocally identified in only three patients, all male (Table II) (Franke, 1972; Surana and Conen, 1972; Schrott et al, 1974). Schrott et al drew attention to clinical features which they thought might be indicative of 4q trisomy—in particular genital and renal abnormality. Both testes were undescended in the patient described by Surana and Conen and one only descended in that described by Schrott and his colleagues. The patient described by Franke had 'hypoplastic' external genitalia. Bilateral renal hypoplasia with hydronephrosis of the right kidney was described by Surana and Conen; a left-sided hydronephrotic kidney and urinary tract outflow obstruction were features of the patient described by Schrott et al; but the third patient had no renal abnormality.

In our patient, also a male, the penis appears normal though only one testis was identified, and that lies in the inguinal canal. Renal abnormality was not apparent. Exomphalos and Hirschsprung's disease were not noted in the 3 patients earlier described; though they were prominent features in our patient.

In all 4 patients, facial features, though dysmorphic, are not very abnormal. While it is appreciated that in each patient 4q+ is complicated by duplica-
Case reports

Fig. 3. Karyotype of propositus.

Fig. 4. Banded karyotype of mother of propositus.
### TABLE II
COMPARISON OF CLINICAL FEATURES AND CYTOGENETIC FINDINGS IN 4 EXAMPLES OF TRISOMY FOR LONG ARM OF No. 4

<table>
<thead>
<tr>
<th></th>
<th>Franke</th>
<th>Surana and Conen</th>
<th>Schrott et al</th>
<th>Propositus</th>
</tr>
</thead>
<tbody>
<tr>
<td>Face</td>
<td>Small palpebral fissures; epicanthal folds; prominent occiput</td>
<td>Hypertelorism and low-set malformed ears</td>
<td>Minor anomalies of nose and right ear; mild micrognathia</td>
<td>Moderately dysmorphic; hypertelorism; left-set ears</td>
</tr>
<tr>
<td>Genitalia</td>
<td>'Hypoplastic external genitalia'</td>
<td>Undescended testes; at necropsy right testis was abdominal, left not located</td>
<td>Right testis undescended</td>
<td>Left testis only identified; in inguinal canal</td>
</tr>
<tr>
<td>Renal tract</td>
<td>Not mentioned</td>
<td>Bilateral renal hypoplasia; hydropneophrotic right kidney</td>
<td>Hydropneophrotic left kidney; outflow obstruction</td>
<td>Normal</td>
</tr>
<tr>
<td>Additional clinical features</td>
<td>Mental subnormality; limitation of motion in neck and hips; alive at 4 years</td>
<td>Short neck with loose skin folds posteriorly; supernumerary digit right hand; bilateral rockerbottom deformity; dilated rectosigmoid area; but normal biopsy findings; ? tetralogy of Fallot; died at 7 months; necropsy revealed pulmonary stenosis, ventricular septal defect, right ventricular hypertrophy, and overriding aorta</td>
<td>Low birthweight; microcephaly, mental subnormality; poor muscle tone; bilateral simian creases with (\text{atd}) angles in (t^r) position; alive at 30 months</td>
<td>Microcephaly; mental subnormality; exomphalos; Hirschsprung’s disease; simian creases not present; (\text{atd}) angles not in (t^r) position; alive at 40 months</td>
</tr>
<tr>
<td>Cyogenetic findings</td>
<td>46,XY,20q+ mat; trisomy distal half long arm No. 4; partial monosomy long arm No 20</td>
<td>46,XY,18q+ mat; trisomy distal half long arm No. 4; partial monosomy long arm No. 18</td>
<td>46,XY,13q+ mat[t(4;13)(q26 or q27;q34)]; trisomy distal third long arm No. 4; monosomy terminal part long arm No. 13</td>
<td>46,XY,9q+ mat[t(4;9) (q31;q34)]; trisomy distal third long arm No. 4; monosomy terminal part long arm No. 9</td>
</tr>
</tbody>
</table>

Tetraploidy in a liveborn infant

**Summary.** A congenitally malformed infant with a tetraploid chromosome complement who survived to 1 year of age is reported. The relationship of the polyplody and the anomalies is discussed.

Polyplody is generally well tolerated in plants and invertebrate animals but appears to be lethal or sublethal in man and other mammals. Niebuhr (1974) collected data on 275 cases of triploid abortuses and 18 cases of triploid fetuses surviving 28 weeks of gestation. He noted that the 8 infants who lived more than a few days were all 2n/3n mosaics. There has been great variability among the malformations reported in these patients. Tetraploidy is less common and only 16 tetraploid abortuses have been reported (Geneva Conference, 1966; Hamerton, 1971) none of which contained a formed embryo. We wish to report a congenitally malformed infant with a tetraploid chromosome complement who survived to 1 year of age.

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