We thank Dr Arthur Rodriguez and Ms Candace Thompson for their assistance.

M. B. JENKINS and L. BOYD
Human Genetics Unit
Minnesota Department of Health
717 Delaware Street S.E.
Minneapolis, Minnesota 55440
U.S.A.

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

Summary. The clinical and cyto- genetic 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 infrquent motions led to rectal biopsy and the identification of Hirschsprung's disease, with aganglionosis extending to at least 10 cm

![Fig. 1. Pedigree of family showing segregation of translocation, t(4;9)(q31;q34). Year of birth is given.](http://jmg.bmj.com/ on June 20, 2017 - Published by group.bmj.com)
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-

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**TABLE I**

**DERMATOGLYPHS OF PROPOSITUS**

<table>
<thead>
<tr>
<th>Digital patterns*</th>
<th>Right</th>
<th>Left</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>W</td>
<td>W</td>
</tr>
<tr>
<td>2</td>
<td>U</td>
<td>W</td>
</tr>
<tr>
<td>3</td>
<td>U</td>
<td>W</td>
</tr>
<tr>
<td>4</td>
<td>W</td>
<td>W</td>
</tr>
<tr>
<td>5</td>
<td>U</td>
<td>W</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Palmar axial tirradii</th>
<th>t</th>
<th>t</th>
</tr>
</thead>
<tbody>
<tr>
<td>A, b, c, and d palmar tirradii</td>
<td>Normal position</td>
<td>Normal position</td>
</tr>
</tbody>
</table>

| Hallucal patterns | High scoring loop | High scoring loop |

* U = ulnar loop; W = whorl.
Case reports

Fig. 3. Karyotype of propositus.

Fig. 4. Banded karyotype of mother of propositus.
tion of segments of varying length, and that chromosomal deficiency (different in each patient) may further complicate the gene imbalance, the present authors see no clear indication of a distinctive pattern of clinical abnormality associated with 4q+. All four patients are male and in each case unbalanced chromosome abnormality followed segregation from a mother with a reciprocal translocation.

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M. Issa, A. M. Potter, and C. E. Blank
From the Department of Zoology, Damascus University; and the Centre for Human Genetics, Sheffield

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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.

Polyploidy 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.
Multiple congenital defects associated with trisomy for long arm of No. 4.
M Issa, A M Potter and C E Blank

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