Autosomal Translocation in a Mentally Retarded Male Child with 46,XY,t(2q-;13q+) Complement: Case Report and Review*

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Human chromosome No. 2 is very often involved in chromosomal rearrangements and there are already over 30 reports. Five of them (Lisco and Lisco, 1967; Reisman and Kasahara, 1968; Ricci, Dallapiccola, and Cotti, 1968; Wurster et al, 1969; Davison, Bedford, and Dunn, 1970) deal with an exchange between the long arms of one No. 2 and a D group chromosome. The present report describes a translocation between chromosomes No. 2 and 13 found in the blood lymphocytes of a 25-month-old male child with mental retardation and minor congenital malformations.

Case Report

History. This 25-month-old boy was investigated because of abnormal appearance and mental retardation. He was born at term (weight 3487 g) after a normal pregnancy to a 35-year-old mother. Routine chest radiography was performed during the 3rd month of gestation. There was no history of drug ingestion or viral infection. The parents, both French-Canadian, were in good health and nonconsanguineous with no relevant family history. One sib, a girl aged 5 years, was physically and intellectually normal.

At the age of 13 months this boy was admitted to hospital for a right inguinal herniorrhaphy. He weighed 9960 g, was 73 cm in height, and showed a normal stature. He was slightly retarded in development. He could stand alone but could not walk or speak words. His reactions to surroundings appeared normal and his behaviour was that of a 9-month-old infant.

Examination revealed a bizarre facies and a dysmorphic head (Fig. 1). The palpebral fissures were small with deep orbits (enophthalmia), bilateral epicanthic folds and hypertelorism. The eyebrows joined over the bridge of the nose which was depressed and small. The lower extremity of the nasal bone was bifid. The ears

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FIG. 1. The patient at age 25 months.
were large and pointed but normally placed. There was slight microretrognathism. The hard palate was elevated and the gums were thickened. The hands were thin with elongated fingers, a single transverse palmar crease, an atrophic hypothenar area and some articular hypermobility. The electroencephalogram and electrocardiogram were normal. Chest and skull radiographs and intravenous pyelograms were non-contributory. Urinary and blood amino-acid chromatography were normal. Retinal examination revealed generalized pallor of the papilla. In the macula region there was a lack of reflection of the fovea centralis and loss of pigment.

**Cytogenetic Analysis.** Chromosome preparations were made from short-term microcultures of heparinized whole blood, and enlarged photomicrographs of metaphases were examined for numerical and structural anomalies. Autoradiographic study was performed on cultured lymphocytes after addition of $^3$H thymidine 3½ hours before termination of the culture. The air-dried cells were stained and coated with Ilford L-4 photographic emulsion which was developed after a 4-day exposure.

Examination of 90 metaphases from the propositus revealed a modal number of 46 with XY pattern. In all cells, a chromosome No. 2 was missing, an extra chromosome was present in the C (6–12–X) group and one D (13–15) group chromosome had an unusually long arm (Figs. 2 and 3). Autoradiographic analysis revealed heavily labelling of the proximal half of the long arm of the long D group chromosome and light labelling of the distal half, indicating the presumed translocation of part of chromosome No. 2 was the distal half of the long arm. It was concluded that the labelling pattern of the abnormal D group chromosome was otherwise characteristic of autosome No. 13 and that the extra chromosome was the remainder of the missing No. 2. The chromosomes of the parents and sister of the propositus were normal.

**Discussion**

Our report is the sixth in which a portion of the long arm of a chromosome No. 2 is translocated to the distal end of the long arm of a group D chromosome, but it is the first in which a No. 13 is involved. The abnormal chromosome described by Reisman and Kasahara (1968) was identified as a No. 14, and that of Wurster et al (1969) as a No. 15 autosome. Autoradiographic studies were not described by Lisco and Lisco (1967), Ricci et al (1968) and Davison et al (1970) about their t(2q–;Dq+) observations (Table I).

The effect of this chromosomal rearrangement on the phenotype cannot easily be determined. The infertile female described by Lisco and Lisco (1967) probably had a balanced chromosomal rearrangement. The lethal congenital abnormalities in the

![Fig. 2. Metaphase from lymphocyte of the propositus. Arrows point to the abnormal autosome 2 and 13.](http://jmg.bmj.com/)

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infant reported by Ricci et al (1968) were undoubtedly related to her partial trisomy No. 2 and her phenotypically normal mother had a balanced translocation. If similarity of congenital defects and presence of a 2:D translocation in most reported cases is not coincidental then material lost from chromosome No. 2 should be considered more important than that lost from the D chromosome, since the abnormal D chromosomes involved in the cases of Reisman and Kasahara (1968), Wurster et al (1969), and this report are different.

Translocation of a part of the long arm of chromosome No. 2 to the long arm of autosomes other than the D group has also been reported. A
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TABLE I

PHENOTYPIC AND FAMILY CHARACTERISTICS OF 6 PATIENTS WITH (2q-;Dq+) TRANSLOCATION

<table>
<thead>
<tr>
<th>Case</th>
<th>Sex and Age</th>
<th>Translocation</th>
<th>Phenotypic Anomalies</th>
<th>Remarks</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lisco and Lisco (1967)</td>
<td>F, 3 yr</td>
<td>(2q-;Dq+)</td>
<td>Slight hypothyroidism and infertility</td>
<td></td>
</tr>
<tr>
<td>Ricci et al (1968)</td>
<td>F, 45 dy</td>
<td>(Dq+)</td>
<td>Microcephaly, hypertelorism, depressed nasal bridge, microretrognathia, low-set ears, systolic murmur, hypotonia, death at 75 days</td>
<td>Balanced (2q-;Dq+) translocation in the normal mother</td>
</tr>
<tr>
<td>Reisman and Kasahara (1968)</td>
<td>F, 27 mth</td>
<td>(2q-;14q+)</td>
<td>Head asymmetry, hypertelorism, epicanthic folds, microretrognathia, depressed nasal bridge, bilateral colobomata, joint hypermobility, extra digits, hypotonia, mental retardation, simian creases</td>
<td>Normal chromosomes in mother, presumptive father and 3 sibs,</td>
</tr>
<tr>
<td>Wurster et al (1969)</td>
<td>F, 12 yr</td>
<td>(2q-;15q+)</td>
<td>Motor-coordination clumsiness, speech retardation, mental deficiency</td>
<td>Normal chromosomes in parents and twin sister</td>
</tr>
<tr>
<td>Davison et al (1970)</td>
<td>F, 3 yr</td>
<td>(2q-;Dq+)</td>
<td>Speech and mental retardation</td>
<td>Normal chromosomes in parents</td>
</tr>
</tbody>
</table>


