Familial pericentric inversion of chromosome 13 resulting in duplication 13q22→qter

SUMMARY A 10-year-old male with a recombinant (13)dup q chromosome is reported. The recombinant chromosome originated from a maternal pericentric inversion which was present in two of his four normal sibs. A segregation analysis of 60 pregnancies from which one of the parents was a carrier of inv(13) showed a significant predominance of males among the viable offspring.

Cases with duplication of a distal 13q segment have led to the detection of familial pericentric inversions

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Fig 1  Family pedigree including years of birth.

Fig 2  Proband at 10 years of age.
were short and broad and he had tapering fingers with hyperconvex nails. Palmar and plantar dermatoglyphs corresponded with the cases of distal trisomy 13q originating from familial translocations, inversions, or de novo previously reviewed by Habedank.\(^1\) In the carrier mother some special features were present on the palms.

Computerised tomography showed moderate enlargement of the ventricular system. Laboratory findings included a normal fetal haemoglobin value and no increased nuclear projections of neutrophils. The patient and both parents had an esterase D type 1–1.

Chromosome analysis on peripheral blood lymphocytes by G banding, C banding after Q banding, and NOR silver staining showed a chromosome complement of 46,XY,13p± in the proband. One chromosome 13 carried an additional segment 13q22–qter on its short arm. Its C banded centromeric region was distinctly smaller and more weakly stained than those present in the other D chromosomes and the NOR silver staining was negative.

The family study revealed that the mother and two brothers carried an inv(13)(p11q21) with Ag positive NORs and distinct satellites on the distal band q21 of the long arm. Its constitutive heterochromatin was distributed between the centromeric region and the distal end of the long arm (fig 3).

The father, two sibs, and a child of a carrier brother had normal karyotypes.

### TABLE Segregation analysis of inv(13) carriers

<table>
<thead>
<tr>
<th>Authors/position in pedigree</th>
<th>Abortions</th>
<th>Unbalanced offspring</th>
<th>Carriers</th>
<th>Normal karyotype</th>
</tr>
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<tbody>
<tr>
<td></td>
<td>M</td>
<td>F</td>
<td>M</td>
<td>F</td>
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<tr>
<td>Carrier mothers Parrington and Edwards(^2)</td>
<td>2</td>
<td>1</td>
<td>1</td>
<td>2</td>
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<tr>
<td>Hauksdottir et al(^3)</td>
<td>III.3</td>
<td>1</td>
<td>1</td>
<td>1</td>
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<tr>
<td></td>
<td>III.4</td>
<td>3</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td></td>
<td>III.9</td>
<td>1</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td></td>
<td>IV.5</td>
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<td>1</td>
<td></td>
</tr>
<tr>
<td>Taysi et al(^4)</td>
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<td>1</td>
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<td>2</td>
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<tr>
<td></td>
<td>III.2</td>
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<td>1</td>
<td></td>
</tr>
<tr>
<td>Escobar et al(^5)</td>
<td>This report</td>
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<tr>
<td></td>
<td>Total</td>
<td>8</td>
<td>5</td>
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<td>Carrier fathers Hauksdottir et al(^3)</td>
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<td>1</td>
<td>2</td>
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<tr>
<td>Koske-Westphal et al(^7)</td>
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<td>2</td>
<td>–</td>
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<td>Total (all carriers)</td>
<td>11</td>
<td>7</td>
<td>3</td>
<td>11</td>
</tr>
</tbody>
</table>
Case reports

Discussion

The segregation analysis of 60 pregnancies shows that among all viable offspring from inv(13) carriers there is a significant predominance of males (table). This observation suggests a selection against female offspring. However, the reason for this selection remains obscure.

This paper is dedicated to Professor Doctor H Schönenberg, Aachen, on his 65th birthday.

The author gratefully acknowledges the excellent assistance of Mrs Eva-Maria Bergmann and Heidi Schuster, and thanks Professor A Rodewald, Homburg, for the dermatoglyphic and esterase D studies, and Dr Roebruck, Aachen, for the statistical calculation of the segregation analysis.

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Two Robertsonian translocations in a boy with mental retardation*

SUMMARY An 8-year-old boy with mental retardation was found to be a mosaic, showing three different cell lines, 46,XY/46,XY,−21,+t(q21q21)/45,XY,−13,−21,+t(q13q21) in cultured peripheral blood cells.

Reports of structural mosaicism involving two different Robertsonian translocations are very few.1 2 We report here a very unusual case of a boy with no clinical signs of Down syndrome carrying two different Robertsonian translocations and a normal cell line.

Case report

The patient (fig 1) was born in January 1968 after

![Patient at 6 years of age: full face and profile.](image)

*This work was partially supported by The Birth Defects Institute, New York State, Department of Health.

Requests for reprints to Professor M Habedank, Klinikum RWTH, D-5100 Aachen, Federal Republic of Germany.

References

1 Habedank M. Partial trisomy 13q21−qter de novo due to a recombinant chromosome rec(13)dup q. Hum Genet 1979;52:91−9.

Requests for reprints to Professor M Habedank, Klinikum RWTH, D-5100 Aachen, Federal Republic of Germany.

Note: photographic artefacts (speckled pattern).
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