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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References


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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

*This work was partially supported by The Birth Defects Institute, New York State, Department of Health.
approximately 34 weeks' gestation. The mother was 29 years old and the father was 33 years old. They have a normal girl, 4 years older than this boy. The family history is negative for any birth defects or mental retardation. The pregnancy was uneventful and birthweight was 2099 g. Because of the noticeable maturation delay noted by the family physician, the child was referred for consultation at the age of 13 months and the diagnosis of psychomotor retardation was confirmed. At the age of 4 years 5 months he was examined again. His height was 96·5 cm, his weight was 17·4 kg, and his head circumference was 49 cm (−2 SD). Neurological examination demonstrated no defects, but fine manipulation was poor. He was referred to us for chromosome studies when he was 6 years old (71 months).

**Psychological Evaluation**

At 53 months his IQ was 60, and, although his other psychological test scores fell within the mildly retarded range, clinically he functioned at a moderately retarded level. His speech was intelligible; he had retained a jargon speech pattern, although he was capable of speaking sentences.

**Dermatoglyphs**

Palmer axial triradii were in t and t" position bilaterally. Simian creases were not present. A dermatogram showed the diagnostic index line to be just touching the Down syndrome bar.

**Cytogenetic Studies**

Peripheral blood leucocytes were cultured for chromosome study. Both quinacrine and trypsin-Giesma banding techniques were used to analyse the patient's chromosomes. A total of 400 cells was analysed. Unfortunately, skin biopsy was refused by the parents.

**Results**

Of the chromosomes, 51·25% showed a balanced 13;21 translocation, 47·75% showed trisomy 21 in the form of 21;21 translocation, and 1% were normal, 46,XY (figs 2, 3, 4). The karyotypes of both parents were normal.

**FIG 2** Trypsin banded karyotype showing 21:21 translocation.
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FIG 3 Trypsin banded karyotype showing 13:21 translocation.

FIG 4 Trypsin banded normal karyotype.
Discussion

Mosaicism involving more than two cell lines in Down syndrome patients has been described. However, the reports of mosaicism involving two different Robertsonian translocations are very few.

In 1965, Zellweger and Abbo\(^2\) reported a case where mosaicism was observed in a girl with Down syndrome. She had four different cell lines, balanced and unbalanced translocations involving D;D and D;G lines, as well as a normal cell line. However, pictures were not available and banding techniques were not in use at that time. Chromosomal mosaicism was observed in other members of the family also. They attributed this familial mosaicism to an autosomal dominant gene. Another case of Down syndrome with two different Robertsonian translocations (15;21 and 21;21) was reported by Atkins and Bartoscas in 1974.\(^1\) In this case and the case of Zellweger and Abbo the patients appeared to have a Down phenotype.

In the present case, approximately 50% of the patient’s blood cells were trisomic for chromosome 21 and 50% were normal (table). This could explain why he did not present with typical Down syndrome features. This again shows that, in a mosaic, when a percentage of a particular cell line is less than 1 or 2%, it may go unnoticed in a routine count of 30 cells.

In our case, we could not determine whether q21q21 was an isochromosome or a translocation. The formation of an isochromosome in one cell line and chromosomal breakage leading to q13;q21 translocation in another cell line of a normal zygote could be one of the explanations of this mosaicism. Another possibility is that the zygote started out as normal 46,XY, non-disjunction at the second mitotic division resulted in a 46/47 mosaicism (non-disjunction at the first mitotic division would result in a regular trisomy), or the zygote started out as trisomic for a chromosome 21. Anaphase lagging of a chromosome 21 at one of the first mitotic divisions can also result in a 46/47 mosaicism. Since there was no 47 cell line, chromosomal breakage might have occurred immediately in a cell with 47 chromosomes giving rise to a 21;21 translocation. Chromosomal breakage occurred also in a cell with 46 chromosomes and gave rise to a 13;21 translocation. Considering that the normal cell line represents only 1% in the distribution of three cell lines, this chromosomal breakage might have occurred in the early cleavage division. We were unable to carry out blood group studies for chimaerism.

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Autoimmune chronic active hepatitis in Down’s syndrome

SUMMARY Hashimoto’s thyroiditis, autoimmune adrenalitis, pernicious anaemia, and diabetes mellitus are all recognised associations with Down’s syndrome. In addition chronic active hepatitis (CAH) resulting from chronic hepatitis B antigenaemia is known to occur in these patients, but an association of autoimmune CAH and Down’s syndrome has not previously been described. We report a case in which Down’s syndrome was associated with autoimmune CAH, Hashimoto’s thyroiditis, and alopecia areata.

Case report

A 29-year-old man with Down’s syndrome was referred in May 1980 with a 9-month history of increasing lethargy and dyspnoea, dryness of the...
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