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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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of chromosome 13. De novo rearrangements between the homologous chromosomes 13 seem to be rare.¹ Another case of duplication 13q (distal) originating from a maternal pericentric inversion is presented here.

Case report

The proband, a male, was born in 1969, the fifth child of healthy non-consanguineous parents. At the time of birth his mother was 37 and his father 38 years old. The other four children were normal. There was no family history of abortions, birth defects, or mental retardation (fig 1).

At the age of 10 years his height was 113 cm, below the 3rd centile. He had severe psychomotor retardation, spastic diplegia of the legs, and myoclonic and akinetic seizures. The facial appearance was characteristic of partial trisomy for the distal segment of 13q (fig 2). The thumbs and big toes

FIG 2 Proband at 10 years of age.

FIG 1 Family pedigree including years of birth.

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 Hadedank.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.

FIG 3 D group chromosomes from the inversion carrier mother and the proband with 46,XY,rec(13)dup q, inv(13)(p11q21)mat. G and C banding (after Q banding) and NOR silver staining.

TABLE Segregation analysis of inv(13) carriers

	Authors/position in pedigree	Abortions	Unbalanced offspring		Carriers		Normal karyotype	
			M	F	M	F	M	F
Carrier mothers	Parrington and Edwards ²	2	1	1			2	
	Hauksdóttir <i>et al</i> ³							
	III.3	1	1			1	3	
	III.4	3	1		2		2	1
	III.9	1		1				1
	IV.5						2	
	Taysi <i>et al</i> ⁴							
	II.6				2	1	1	
	III.2	1		1				
	Escobar <i>et al</i> ⁵		1			1		
Carrier fathers	This report	II.3	1		2		1	1
	Total	8	5	/ 3	6	3	11	/ 3
	Hauksdóttir <i>et al</i> ³							
	II.1	1			2			
	II.3	1				2		
	II.9					1	1	1
	III.1				1		1	
	III.2	1			1		1	
	IV.10					1		
	Surana and Conen ⁶		1			1	1	
Total (all carriers)	Koske-Westphal <i>et al</i> ⁷		1		1			
	This report	III.4						1
	Total	3	2	/ -	5	/ 5	4	/ 2
	Total	11	7	/ 3	11	/ 8	15	/ 5

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.

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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

FIG 1 Patient at 6 years of age: full face and profile. Note: photographic artefacts (speckled pattern).

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