Short report

Parental mosaicism in de novo translocation (21q21q) Down's syndrome

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Parental mosaicism for t(21q21q) has been found in six out of 11 families in which recurrence of a de novo 21q21q translocation Down's syndrome was observed.[1-3] Therefore, Hall[1] proposed that in the parents of children with de novo 21q21q Down's syndrome the analysis of additional cells and also skin fibroblasts studies should be considered for the detection of cryptic mosaicism. Support for this suggestion comes from a family studied by us, in which the application of extensive cytogenetic investigations permitted the discovery of a very low degree parental mosaicism on the birth of the first child with apparently de novo 21q21q Down's syndrome.

The proband, the first born male of healthy, non-consanguineous parents, was referred for cytogenetic study because of clinical signs of Down's syndrome. His karyotype was 46,XY,−21,+t(21q21q). Peripheral blood cultures of the parents showed a normal karyotype in the father. However, the study of 200 cells of the mother, a 27 year old woman of normal intelligence without signs of Down's syndrome, showed one 21 trisomic cell carrying the same t(21q21q) as her affected son. Analysis of maternal lymphocytes was extended and eventually four cells out of 505 (0·8%) showed the 21q21q translocation. Two subsequent chromosome studies confirmed the presence of the trisomic cell line which showed the same very low frequency (<1%) in both blood lymphocytes and skin fibroblasts (table).

Cytogenetic findings in repeated analysis of the mother.

Examination	Tissue	46,XX	46,XX,−21, +t(21q21q)	%
1st	Blood	501	4	0·8
2nd	Blood	566	0	0·0
	Skin	574	3	0·5
3rd	Blood	998	4	0·4

In our family the parental mosaicism was detected in the absence of recurrence of t(21q21q) Down's syndrome, by examining a large number of metaphases. This approach requires only a little extra effort on the part of the laboratory; the cells need not be counted or analysed but simply inspected for t(21q21q) or normal chromosome 21. The effort is justified because of the importance for genetic counselling of the discovery of parental mosaicism. This finding, in fact, considerably modifies the recurrence risk for t(21q21q) Down's syndrome and may also dramatically change a couple's reproductive choices, particularly for those who, for moral or religious reasons, would refuse termination of a pregnancy.

Our findings reinforce the suggestion of Steinberg *et al*[3] that caution should be exercised when counselling a low recurrence risk to the parents of a child with de novo t(21q21q) Down's syndrome, and make prenatal cytogenetic studies important in any future pregnancies of these couples.

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