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FAMILIAL EXTRA BISATELLITED MICROCHROMOSOME AND DOWN SYNDROME

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Authors report on a Down infant, whose chromosome complement includes an extra small bisatellited marker chromosome. This marker was also found in four healthy members of the family.

INTRODUCTION

The detection of an accessory small marker chromosome may present a difficult diagnostic problem and especially familial cases may call attention to new aspects of the question /2, 3, 4, 5, 6, 7/. Recently we observed a girl with trisomy 21 who had an additional bisatellited marker chromosome which was also found in four healthy family members in three generations.

CASE REPORT

The newborn female infant was the first child of the healthy, nonconsanguineus parents. The mother aged 22 years, the father 21 years. The proposita's birth weight was 2900 g, length 47 cm. She had a typical phenotype of Down's syndrome.

Analysis of her lymphocyte mitoses revealed regular trisomy 21 plus an accessory small chromosome in each of the 50 cells examined. This marker was smaller than the members of the G-group; in 70% of the mitoses it was associated to the satellites of one of the acrocentrics. An attempt for its identification was done by means of C and G-banding, DA/DAPI and NOR methods, and it was regarded as inv dup 15p (Fig. 1).

The same marker was identified in four out of ten healthy, mentally normal family members (Fig. 2). At least 20 mitoses were analysed in each case and no mosaicism was detected.

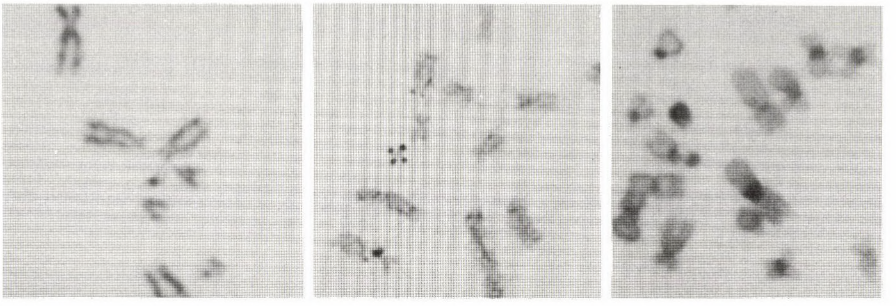

Fig 1. G-banded (left), silver-stained (middle) and C-banded (right) marker chromosome

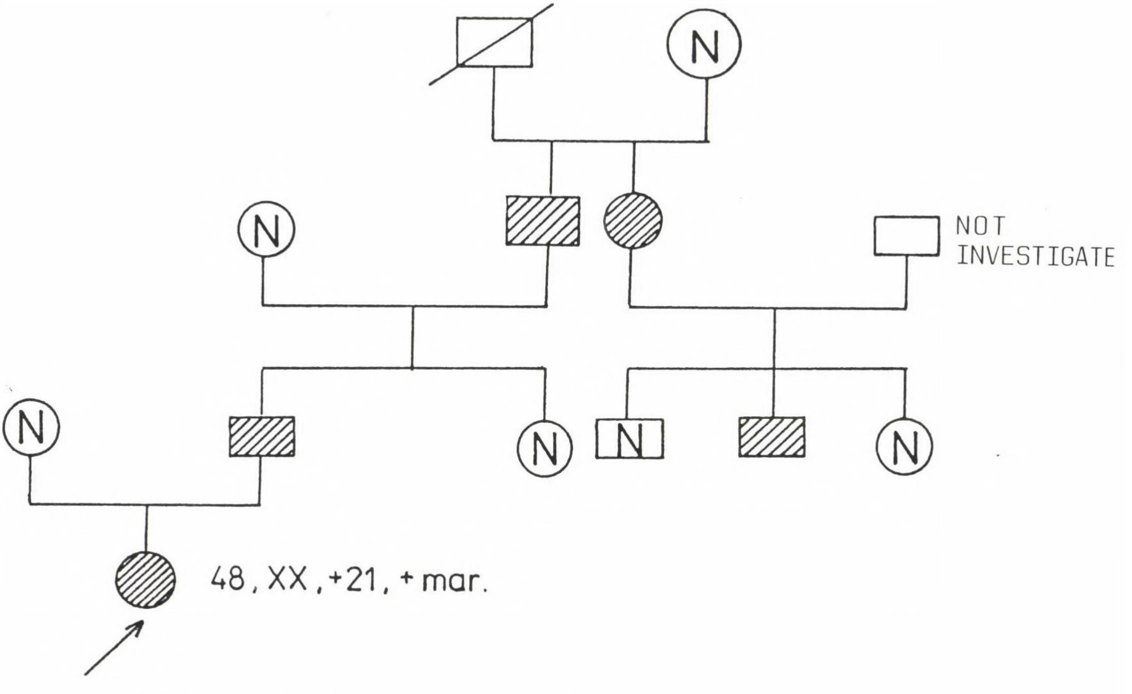

Fig. 2. Pedigree of the family

DISCUSSION

The significance of supernumerary marker chromosomes was extensively reviewed by Buckton et al /1/. The familial case reported here confirms most of their conclusions, however, we call attention to two particular points:

1. The simultaneous occurrence of trisomy 21 and inv. dup /15/ may be merely incidental, but it also may suggest that the presence of a marker chromosome may interfere with the normal process of chromosome pairing and disjunction of acrocentric chromosomes.
2. The present case provides further evidence for the assumption that the risk of abnormality to a fetus carrying a marker is minimal if the phenotypically normal carrier parent has that marker in a non-mosaic form.

REFERENCES

1. Buckton KE, Spowart G, Newton MS, Evans HJ: Forty four probands with an additional "marker" chromosome. Hum Genet 69: 353, 1985
2. Carrasco JJL, Otero GA, Vilar MMC, et al: Small marker chromosomes in a series of 1000 prenatal diagnoses by amniocentesis. Ann Genet 33: 40, 1990
3. Djalali M: The significance of accessory bisatellited marker chromosomes in amniotic fluid cell culture. Ann Genet 33: 141, 1990
4. Neri G, Cagiano MD, Serra A, et al: Properties and significance of a small marker chromosome in amniotic fluid cells. Clin Genet 18: 253, 1980
5. Romain DR, Columbano-Green L, Smythe RH, Dukes PC: Discovery of an inherited bisatellited metacentric microchromosome in amniotic cell culture. Clin Genet 16: 183, 1979
6. Sachs ES, Van Hemel JO, Den Hollander JC, Jahoda MGJ: Marker chromosomes in a series of 10 000 prenatal diagnoses. Cytogenetic and follow-up studies. Prenat Diagn 7: 81, 1987

7. Yip MY, Mark J, Hultén M: Supernumerary chromosomes in six patients. Clin Genet 21: 397, 1982

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