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A complex balanced rearrangement involving four chromosomes in an azoospermic man

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SUMMARY A complex chromosomal rearrangement involving chromosomes 1, 5, 10, and 12 is described. The patient was an infertile, phenotypically normal male. Cytogenetic analyses of his parents showed that the complex translocation arose de novo. Testicular histology showed spermatogenic arrest.

Complex chromosomal rearrangements involving exchanges between three or more chromosomes are not frequent¹ and their association with male infertility is rare.² In the present paper we describe an azoospermic man with a karyotype showing balanced translocations between four chromosomes.

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Case report

A 33 year old azoospermic man was referred to our laboratory. He was born after an uneventful pregnancy and was the only child of the family. At the time of his birth his mother was 23 and his father 32 years old. There was no history of miscarriages. Apart from his infertility he was phenotypically normal.

TESTICULAR HISTOLOGY

The histology of the testes appeared normal. The tubules showed Sertoli and spermatogenic cells but complete absence of spermatozoa was noted. Leydig cells appeared to be well represented. According to the classification of Chandley *et al*³ the biopsy was classified as grade 2.

FIGURE G banded partial karyotype showing derivative chromosomes 1, 5, 10, and 12. Arrows point to the breakpoints on the ideograms corresponding to the chromosomes.

CYTOGENETIC STUDIES

Chromosome analysis of the patient was performed on peripheral blood lymphocytes. Using standard staining (G, C, and T banding methods) all cells showed only one normal homologue of chromosomes 1, 5, 10, and 12, together with four derived chromosomes (figure). The proband's karyotype was identified as a complex balanced translocation between chromosomes 1, 5, 10, and 12 which can be represented as: 46,XY,t(5;1;10;12)(5qter→5p13::12q24→12qter;1pter→1q42::5p13→5pter;10pter→10q24::1q42→1qter;12pter→12q24::10q24→10qter). The karyotype of both parents was normal.

Discussion

Complex balanced rearrangements in azoospermic males are rare events. Single balanced translocations have often been related to infertility in males. The infertility, in some cases, is due to spermatogenic arrest caused by the complexity of meiotic configurations.⁴

Unfortunately there is no information about the meiotic behaviour of complex chromosomal rearrangements. However, we can assume that, in the present case, the four derivative chromosomes

would have pairing difficulties leading to serious meiotic disturbances. Therefore one can speculate about the possibility that spermatogenic arrest and consequent azoospermia are directly attributable to the complex translocation detected in the patient and not to a chance association of both unrelated phenomena.

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A child with a recombinant of chromosome 8 inherited from her carrier mother

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SUMMARY A female child with mental retardation and dysmorphic features was found to have a duplication deficiency of chromosome 8: rec(8)dup q,inv(8)(p23q24), a recombinant product derived from a familial pericentric inversion, inv(8)(p23q24)mat. Clinical features of this previously undescribed inversion product are compared with other reported cases of partial trisomy for the distal long arm of chromosome 8, since this segment is thought to be primarily responsible for the phenotypic features of the trisomy 8 syndrome.

Since the introduction of banding techniques, pericentric inversions in human chromosomes have

been recognised with increasing frequency. Not all inversions give rise to unbalanced products and each must be assessed independently in order to calculate specific risk figures. In general, the risk of a recombinant chromosome being produced depends upon the size and position of the inversion and the particular segment involved.

There have been nine published cases of pericentric inversions of chromosome 8.¹⁻⁹ These fall into two main categories, distinguished by their arm ratios, risk of abnormal offspring, and coincidentally by their geographical distribution. The first group, which forms acrocentric type chromosomes inv(8)(p23q11),^{1,2} inv(8)(p23q12),³ and inv(8)(p11q24)⁴ found in the Scottish and Finnish populations, have no documented unbalanced progeny, while the second, which forms a metacentric type chromosome, inv(8)(p23q22),⁵⁻⁹ reported in several families of Mexican-American ancestry, has