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A three way translocation in mother and daughter

A woman presented with a history of six miscarriages occurring between the 6th and 14th weeks of gestation. Chromosome analysis revealed a complex rearrangement, with segments of three chromosomes translocated in 'circular' fashion: most of 2q onto 18q; some of 18q onto 11p; and the tip of 11p onto 2q. Her karyotype can be expressed as 46,XX,t(2:11;18)(2pter→2q13::11p15→11pter;11q1ter→11p15→3::18q21→18qter;18pter→18q21→2q13→2qter). A partial karyotype is shown in the figure. Because both her parents had normal chromosomes, it can be assumed that the rearrangement arose de novo. Her two pregnancies which went to term produced phenotypically normal daughters, one with the same balanced three way translocation, the other with a normal karyotype.

Complex chromosomal rearrangements are rare.1,2 In most cases where the translocation is familial, it is the mother who is the transmitting parent,1 in one family through three generations.3 Oogenesis may be a more robust mechanism of gamete formation than spermatogenesis in the presence of this particular disruptive influence. A maturation arrest at the secondary spermatocyte stage has been demonstrated in one sterile male heterozygote.2

If, at pachytene, pairing of homologous chromosomal segments is successfully achieved, this will lead to the formation of a hexavalent configuration. At the first meiotic division, then, 3:3, 4:2, 5:1, and 6:0 disjunctions are theoretically possible. The great majority of theoretically possible segregant gametes could be expected to give rise to conceptuses with such an unbalanced genetic complement that loss would occur in early pregnancy, often in very early pregnancy ('occult abortion'). Given the enormous theoretical range of gamete abnormality, and bearing in mind the modifying factor of selection against unbalanced forms, the fact that our proband had two conceptuses arising from alternate 3:3 segregation out of eight recognised pregnancies might indicate this category of chromosomal distribution to be favoured. In our advice to her 19 year old heterozygous daughter, we have indicated the high probability of her having miscarriages in the future, but have also counselled cautious optimism that the same good fortune that attended two of her mother's meioses may also dwell in her (although advising that a heterozygous son might possibly be sterile). Prenatal diagnosis would be indicated to detect an unbalanced but viable chromosomal complement.

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References

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Familial inv(1)(p36.3q12) associated with sterility

Chromosomal abnormalities are among the more important causes of reproductive failure. Three instances of familial pericentric inversion of chromosome 1 associated with sterility have been reported.1-3 In this paper we...
A three way translocation in mother and daughter.
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