Whole arm translocation t(1;13) in an infertile man

The proband is a 45 year old Algerian. He is childless after 13 years of marriage. His three brothers and two sisters are fertile. Among his five uncles and five aunts, only one of them has no children. There is no history of infectious disease or trauma. Physical examination revealed testicular hypotrophy. The male genital tract and secondary sex characteristics were normal. FSH and LH levels were raised (20.9 IU/l, 17.5 IU/l) and plasma testosterone level was normal (6.75 ng/ml). The spermogram showed azoospermia. Trichomonas and haemolytic streptococcus (group B of Lancefield) were present.

Chromosome preparations, obtained from phytohaemagglutinin stimulated peripheral blood lymphocytes, were examined after R banding, C banding, and NOR staining. The NOR bearing chromosomes were identified by R banding in the Ag stained metaphases (figure a, b).

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FIGURE (a) R banded metaphase: 46,XY,t(1;13)(q11;p13). (b) Same metaphase with Ag I technique showing NORs. (c) R banded chromosomes 1 and 13. (d) C banded chromosomes 1 and 13. (e) Partial metaphase showing the participation of translocation 1;13 chromosome in satellite association. (f) Chromosomes 1 and D and G group chromosomes. Ag I technique.

R banding revealed a translocation between the long arm of chromosome 1 and chromosome 13 (figure c). C banding showed that the breakpoint in chromosome 1 had occurred within the centromeric heterochromatin. Part of the heterochromatic region was translocated onto the short arm of chromosome 13, thus forming a dicentric chromosome (figure d). The abnormal chromosome 1 consisted of the short arm and a small region of centromeric heterochromatin. No part of chromosome 13 was apparently translocated onto chromosome 1. Ag staining showed that the translocated 1;13 chromosome retained the active NORs and was participating in satellite association (figure e, f). No chromosome studies on the family have yet been possible.

In previously published cases the whole arm translocations have been reciprocal. For example, Mikkelsen *et al*¹ using the Ag-As technique demonstrated a large silver positive block in the centromeric region of the deleted chromosome 1 and concluded that a part of the short arm of chromosome 13, including the NORs, had been translocated onto chromosome 1. The translocation studied by us, with R banding, appeared to be similar to that of Mikkelsen *et al*,¹ but Ag staining revealed that the NORs of chromosome 13 were present in the metacentric t(1;13) chromosome. From the C banding and Ag staining results, it can be concluded that the translocation is not a reciprocal one. The break in chromosome 13 leading to a fusion with the long arm of chromosome 1 must have occurred at a point above the NORs, possibly with a loss of band 13p13, generally considered to be a band with reduced activity. It would be interesting to establish through family studies if the translocation is *de novo* or inherited. A *de novo* event is more likely in view of the non-reciprocal nature of the rearrangement.

The important question is to determine the relationship between infertility and observed chromosomal anomalies in infertile men. In sex chromosome aneuploidies the association is obvious. The role of autosomal rearrangements is more difficult to explain. It seems unlikely that spermatogenic arrest in heterozygotes for chromosome rearrangements could be the result of altered expression of genes at (or near) the chromosome breakpoint, as too many genes would have to be involved in spermatogenesis. Perhaps the hypothesis of Forejt² provides a better explanation. "The following chain of events is responsible for male sterility in carriers of chromosomal rearrangements: the incomplete pairing of rearranged chromosomes at pachytene, the attraction and non-homologous association between unpaired autosomal regions and unpaired parts of X and Y chromosomes, the interference with X inactivation due to X-autosome association, transcription of 'non-permissible' gene products of the X chromosome, the breakdown of spermatogenic differentiation."

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