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Accurate meiotic segregation analysis in sperm from a t(1;10) heterozygous translocation carrier using multi-probe FISH.

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Understanding the meiotic segregational behavior of reciprocal translocations in man is of importance in the estimation of their reproductive risk. In this study sperm from a t(1;10) (p22.1;q22.3) heterozygous reciprocal translocation carrier was analyzed, using multi-probe and multi-color fluorescence in situ hybridization (FISH).

All segregation products could be detected and quantified accurately by hybridizing the sperm with a telomeric and centromeric probe on chromosome 1 and a centromeric probe on chromosome 10. From this analysis the incidence of the different segregation types at meiosis I (alternate, adjacent I, adjacent II and 3:1 segregation) and the frequency of meiotic recombination combined in the adjacent II segregation could be determined. The frequency of chromosomally normal and balanced sperm resulting from alternate segregation or from adjacent I segregation with recombination within the interstitial segments was 47 %. Segregation products from adjacent I or from alternate segregation with recombination within the interstitial segments was 40 %. Frequencies of sperm resulting from adjacent II without recombination was 3.0 %; with interstitial chiasmata in chromosome 1 was 0.9 %; with interstitial chiasmata in chromosome 10 was 1.3%. Overall, the frequency of chromosomally imbalanced sperm was 53%.

In a second probe combination, an centromeric probe on chromosome 8 was used together with the two chromosome 1 probes. There was no increase in the aneuploidy frequency involving chromosome 8 observed. To further analyze the aneuploidy frequencies of chromosomes not involved in the translocation, chromosomes X, Y, 18 and 21 will be studied.

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