Analysis of meiotic segregation, recombination and aneuploidy in sperm of a t(1;10) human heterozygous reciprocal translocation carrier using multi-color FISH for chromosomes 1, 10, 18, 21, X and Y.

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Understanding meiotic segregational behavior of the chromosomes involved in a reciprocal translocation and the segregation of unrelated chromosomes is of importance in the estimation of reproductive risk of male translocation carriers. Sperm from a t(1;10)(p22.1;q22.3) reciprocal translocation carrier was analyzed, using two multi-probe and multi-color fluorescence in situ hybridization (FISH) methods.

The first FISH method consisted of telomeric and centromeric probes on chromosome 1 plus a centromeric probe on 10 which were used to analyze segregation patterns of chromosomes involved in the translocation. The frequencies of Alternate and Adjacent I segregation was 47 % and 40 %, respectively. Sperm products from Adjacent I segregation deviated significantly from the expected 1:1 ratio ($p<0.0001$), suggesting selection against specific sperm genotypes. The frequencies of Adjacent II and 3:1 segregation were 4.8 % and 3.9 %, respectively. Overall, 53 % of the sperm were chromosomally imbalanced. Based on Adjacent II sperm products, recombination in the interstitial segments (segments between the centromere and the breakpoint) was estimated to range from 58 to 87 %. Recombination in the interstitial segment of chromosome 10 was significantly higher ($p=0.006$) than in chromosome 1, which is consistent with the difference in recombination distance of the interstitial segments between the two chromosomes.

The second FISH method was applied to quantify the aneuploidy frequency of four chromosomes not involved in the translocation: X, Y, 18 and 21. The frequencies of hyperhaploid sperm for each of the sex chromosomes and for chromosomes 18 and 21 were ~2 per 10,000. The frequency of diploid sperm was 28 per 10,000. These values are within the range seen for chromosomally normal men.

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