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Letters to the Editor

Origin of sperm with extra chromosome set

Dear Sir,

We read with interest the article by Devillard *et al*. (2002), in which they report on the high frequency of various polyploidies in large-headed sperm. Examining sperm nuclei by FISH from three patients with macrocephalic, multiflagellated sperm, they also concluded that incomplete partition of homologous chromosomes/chromatids during both meiosis I and meiosis II is associated with the failure of nuclear cleavage. The combination of the two disorders determines the chromosomal constitution of the sperm nuclei in the polyploid sperm.

Although this finding may occur in some patients with serious and multiple defects of the meiosis, in our experience, the defective chromosome separation during either meiosis I or meiosis II with the failure of nuclear cleavage dominates over the other is found in most of the patients with moderately elevated frequencies of polyploid sperm. Consequently, and in line with the recent in-depth reviews of the topic from Dr Egozcue and co-workers (Egozcue *et al*., 2000, 2002), the vast majority of the polyploid cells are diploids. Diploid sperm with X and Y chromosomes originate from meiosis I due to chromosomal non-separation (M1 diploids), but may undergo meiotic II chromatid separation plus nuclear cleavage. In contrast, diploid cells with sex-identical chromosome set (XX or YY diploids) can originate only from a defective meiosis II (M2 diploids).

We have studied sperm aneuploidy frequencies in 31 moderately oligospermic men so far (Kovanci *et al*. 2001; Jakab *et al*. 2002; Kovacs *et al*. 2002) using triple-colour fluorescence in-situ hybridization (FISH) for chromosomes X, Y and 17 (mean sperm concentration 17.3×10^6/ml, range $8.0–32.7\times10^6$; ~5000–7000 sperm scored in each, a total of 193 083 sperm). Diploidy frequencies showed a considerable inter-individual variation (mean 0.594%, range 0.03–1.94). It was observed that the higher the total frequency of diploid sperm the greater was the difference between the proportion of M1 and M2 diploids. Among the 10 men with >0.5% diploidy rate there were seven (70%) with >3-fold difference between M1 and M2 diploids (M1>M2 in two cases, M1<M2 in five cases), while there was only one such patient (M1>M2) among the 21 men (4.7%) with <0.5% diploidy rate. Using Pearson correlation analysis we confirmed a very high correlation (r = 0.596; $P < 0.0005$) between the total diploidy rate and the ratios of M1/M2 (or M2/M1, if M1/M2<1) diploids. In conclusion, in sperm with higher than average diploidy rates, the dominance of either the M1 diploids or M2 diploids can always be observed. This suggests, that in the majority of the cases with moderately elevated frequencies of diploid sperm, chromosomal non-separation during meiosis I and defective nuclear cleavage during meiosis II occur separately.

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