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JOURNAL ARTICLE

Fluorescence in situ hybridization analysis of sex-chromosome mosaicism in azoospermic men

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A retrospective study was carried out to determine the prevalence of sex-chromosome mosaicism among azoospermic men and to evaluate the feasibility of using fluorescence in situ hybridization (FISH) technique to assess mosaicism and the origin of marker chromosomes. Nine hundred eighty patients with azoospermia who were referred to a male infertility clinic at a university hospital were karyotyped by G-banding using peripheral blood lymphocyte (PBL) metaphase spreads. When sex chromosome mosaic karyotype was detected, FISH analyses using sex chromosome-specific probes were performed. Seventeen of 980 patients showed evidence of sex chromosomal mosaic karyotype or mosaicism with marker chromosomes by G-banding studies of PBLs. Ten patients showed mosaicism in the number of sex chromosomes and 7 showed mosaicism with marker chromosomes. All 17 patients agreed to undergo FISH analysis. FISH confirmed mosaicism in 88.2% of these patients (15 of 17). Low-frequency mosaicism showing a frequency of less than 10% by G-banding was proved to be nonmosaicism by FISH. Marker chromosomes detected in 7 patients were proved to be derived from the Y chromosome by FISH analyses. From these data the prevalence of sex chromosome mosaicism confirmed by FISH analysis is 1.5% (15 of 980 patients). FISH analysis should be applied when mosaicism shows a frequency of less than 10% by the G-banding technique. Also, FISH analysis is indicated when a marker chromosome is detected by G-banding.

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