Fluorescence in situ hybridization (FISH) as a method to detect aneuploid cells

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摘要

Abstract

OBJECTIVES: To test the sensitivity and specificity of various FISH probes for detecting male and aneuploid cells and to determine the percentage of fetal cells that must be present in a sample in order to use the probes for prenatal diagnosis. METHODS: Adult human lymphocytes were cultured and harvested. Twelve different proportions of male to female cells and 5 different proportions of trisomy 21 cells and trisomy 18 cells in euploid cells were prepared for FISH. Alpha-satellite DYZ1 was applied to detect the male cells. Chromosome 21/Down syndrome critical region cosmid and D13Z1/D21Z1 alpha-satellite probes were applied to detect trisomy 21 cells. The D18Z1 alpha-satellite probe was used to detect trisomy 18 cells. RESULTS: DYZ1 detected male cells reliably in concentrations as low as 2%. Both D18Z1 and chromosome 21/Down syndrome critical region cosmid probes could detect aneuploid cells at reasonably achievable concentrations. However, the D13Z1/D21Z1 probe was not sensitive below concentrations of 50%. CONCLUSIONS: FISH is an alternative technique for noninvasive prenatal diagnosis. The sensitivity and specificity of FISH probes may play a crucial role in the accuracy of prenatal diagnosis.