

Rapid One-day Fluorescence *In Situ* Hybridisation in Prenatal Diagnosis Using Uncultured Amniocytes and Chorionic Villi

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Abstract

Rapid assays for the detection of the common chromosomal aneuploidies using DNA probes to chromosomes 13, 18, 21, X and Y have been developed in recent years. In this study, we report our experience using fluorescence in situ hybridisation (FISH) with uncultured amniocytes from amniotic fluids (AF) and cells from chorionic villus sampling (CVS) for prenatal diagnosis in 239 assays. Only 2 ml of AF or 5mg of chorionic villi were required for FISH analysis using region-specific probes to chromosome 13, 18, 21, X and Y. Results were informative in all the assays conducted in this study. The average time to obtain a result was 24 hours. However, the results for chromosome X, Y and 18 were ready within four hours. The criteria employed for result interpretation were stringent. In a normal finding, at least 80% of all nuclei observed must have 2 discrete signals per cell. In a suspected abnormal case, a minimum of 60% of all nuclei observed must have one (monosomy X) or 3 signals (trisomy) per cell. One trisomy 13 and two trisomy 21 cases were detected. All FISH results were confirmed by karyotyping. Subsequent karyotyping of the resultant abortuses gave further confirmation of the results. The results obtained showed 100% positive and negative predictive values. This study shows that FISH can be used as a rapid routine assay in conjunction with full karyotyping analysis. Its prognostic value is especially important for patients with late referrals, parental anxiety, high-risk pregnancies identified by maternal serological screening and suspected foetal abnormalities detected by ultrasonographic examination.

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