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Quantitative Fluorescence Polymerase Chain Reaction (QF-PCR) for Prenatal Diagnosis of Chromosomal Aneuploidies

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ABSTRACT Genomic aneuploidy is a common cause of human genetic disorders and cytogenetic analysis of metaphase karyotypes remain the standard method to identify aneuploidies and balanced translocations. Quantitative Fluorescence PCR (QF-PCR) is an alternative method in which DNA polymorphic markers on chromosomes, is used to determine the presence of different alleles**.** The assay based on the use of informative polymorphic small tandem repeat (STR) markers and the availability of parental DNA, is employed for prenatal and postnatal diagnosis of aneuploidies of chromosomes 13, 18, 21, X and Y. DNA isolated from fetal cells of amniotic fluid sample, chorionic villus sample, fetal trophoblast cells from endocervical lavage and neonatal blood are all used for the investigation of chromosomal copy number variations. The QF-PCR assay uses fluorescent labelled primers of STR markers that are analyzed after fragment length separation in capillary gel electrophoresis. The determination of the meiotic origin of aneuploidy or the post zygotic mitotic origin could also be done in most cases. Though testing of prenatal samples is complicated by limited sample quantity, variable sample quality, mosaicism and maternal cell contamination- use of parental samples and other measures can overcome most of these limitations. The QF-PCR technique serves as a very useful preliminary test to reduce parental anxiety within a short duration, and to accelerate therapeutic intervention.