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Validation of quantitative fluorescent-PCR for rapid prenatal diagnosis of common aneuploidies in the Chinese population

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ABSTRACT. Quantitative fluorescent polymerase chain reaction (QF-PCR) is an accurate and reliable method for rapid detection of aneuploidy; however, it is not routinely used in China. We aimed to validate QF-PCR as a means for prenatal common aneuploidy screening and to analyze the heterozygosities of short tandem repeat (STR) markers in the Chinese population. The sequences of 19 STR markers in chromosomes 21, 18, 13, X, and Y were designed; three kinds of fluoresceins were used to label the primers, and the QF-PCR detecting conditions were explored and optimized. The results of analysis of 210 prenatal samples by multiplex QF-PCR were compared with karyotyping analysis. All cases were successfully tested by QF-PCR and conventional cytogenetic analysis. QF-PCR

results were consistent with the results of cytogenetic analyses, with the exception of two cases. The sensitivity and specificity of QF-PCR to diagnose common aneuploidies were 94.74 and 100%, respectively. The heterozygosities of most of the markers were lower than reported for Western populations, but relatively similar to those of other Asian populations. We conclude that QF-PCR is able to detect the common aneuploidies for prenatal diagnosis with high detection efficacy; therefore it is suitable for rapid prenatal diagnosis and for large-scale testing in laboratories. However, we need to add new STR markers or to find alternative STR markers with high heterozygosity in order to make this technique useful for routine diagnosis.

Key words: QF-PCR; Prenatal diagnosis; Aneuploidy

