Assessment of multiple quantitative fluorescence PCR technique for common chromosome aneuploidies

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Objective
To evaluate the effectiveness of multiple quantitative fluorescence PCR (QF-PCR) as a rapid technique for prenatal diagnosis of common chromosome aneuploidies.

Methods
In total, 731 amniotic fluid samples of pregnant subjects, who were referred to the Women’s Hospital School of Medicine Zhejiang University, were analyzed with conventional karyotype and the QF-PCR technique by short tandem repeat (STR) markers to detect chromosomes 13, 18, 21, X and Y aneuploidies. There were 588 samples detected by single blind method, 173 samples detected by double blind method.

Results
All of the 731 amniotic fluid samples were tested in this study by QF-PCR and results were compared to the conventional cytogenetic analysis results of the same sample. In the 558 samples with single blind method 5 trisomy 21; 2 trisomy 18; 1 trisomy 13; 1 45,X; 1 47,XXY; 1 47,XYY; 1 47,XXX and 1 69,XXX were detected. In the 173 samples with double blind method 1 trisomy 21 and 1 trisomy 18. The rapid QF-PCR assay was successful to detect all aneuploidies involving chromosomes 21, 18, 13, X and Y in prenatal diagnosis, which were verified by chromosome karyotype analysis. The results showed that the total coincidence rate between QF-PCR and routine chromosome karyotype analysis was 93.75%.

Conclusion
The multiple QF-PCR is a reliable method of detecting common chromosome aneuploidies for rapid prenatal diagnosis.