Clinical application of QF-PCR in rapid prenatal diagnosis of common chromosomal aneuploidies

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Objectives (목적)

Quantitative fluorescent polymerase chain reaction (QF-PCR) is a rapid method for detecting chromosomal aneuploidies within a few hours. The main advantage of QF-PCR is to alleviate the anxiety of parents waiting for conventional cytogenetic results usually available in 14 days. We assessed the results of QF-PCR for detecting the three most common aneuploidies (chromosome 21, 18, 13).

Methods (연구 방법)

From February 2007 through May 2009, we performed QF-PCR on 3,701 amniotic samples and compared the results with conventional cytogenetics. The primary test with 4 short tandem repeat (STR) markers for each chromosome was performed. In chromosome of uninformative results on the primary test such as all single peaks or one diallelic pattern, secondary test with additional 3 STR markers was performed. All results of QF-PCR were informed to the parents within 24 hours after sample collection and they were recommended to wait for the result of the long-term culture.

Results (결과)

The results of 3,624 samples (97.9%) were informative. In cases of informative results, there were 54 cases of nonmosaic trisomy: 31 cases of trisomy 21, 16 cases of trisomy 18 and 7 cases of trisomy 13. The results between QF-PCR and conventional cytogenetics were concordant in 3,622 samples (99.9%) except for the two cases of mosaic trisomy 13. The results of QF-PCR were uninformative in 77 samples (0.21%) due to maternal cell contamination, bloody color of cell pellet, and confused allele ratio. The sensitivity and specificity of QF-PCR for detecting common chromosomal aneuploidies were 96.4% and 99.9%. False positive rate and false negative rate of QF-PCR for detecting common chromosomal aneuploidies were 0% and 0.05%.

Conclusions (결론)

The QF-PCR is clinically useful method for rapid analysis of common chromosomal aneuploidies with high reliability. It helps to decrease anxiety of a great majority of parents with normal fetus. However, the final cytogenetic result should be identified even in negative QF-PCR although the incidence of abnormal cytogenetic result is very low.