Rapid prenatal diagnosis of common chromosome aneuploidies using quantitative fluorescent (QF)-PCR: 10 years experience in a center from the Republic of Macedonia

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INTRODUCTION

The quantitative fluorescent (QF) PCR of selected small tandem repeat (STR) markers enables rapid and accurate prenatal diagnosis of the aneuploidies of chromosomes 21, 18, 13, X and Y.

Here, we present our results of the use of QF-PCR for prenatal detection of common chromosomal aneuploidies in 2200 pregnancies at risk performed in a period of 10 years.

MATERIALS AND METHODS

The prenatal diagnosis was performed on genomic DNA isolated from fetal cells collected by amniocentesis or chorionic villus samples. All samples were analyzed by at least four STR markers on each of chromosomes 21, 18 and 13. In addition, amelogenin, TAF9, SRY and STR markers on X chromosome were used for the determination of the gender and sex chromosome aneuploidies (Figure 1).

Maternal blood samples were analyzed in all blood contaminated amniotic samples and in most chorionic villi samples. In most instances the QF-PCR analysis was performed as a stand-alone test. It was also used in the prenatal cases for monogenic diseases to control for maternal contamination of the fetal material.

RESULTS

Figure 2 shows the results of the detection of aneuploidies of chromosomes 13, 18, 21, X and Y by QF-PCR in 2200 prenatal samples. No discordant results were obtained when cytogenetic analysis was performed in addition to QF-PCR. In one case the discordant sex between ultrasonography and QF-PCR was due to sex reversal subsequently confirmed by cytogenetic analysis.

Polymorphic duplications involving STR markers D13S631, D21S1441, D18S978 or D18S535 were detected in seven fetuses; in all fetuses the duplications were inherited from one of the parents.

The parental origin of the aneuploidy was determined in 64 cases (Figure 3). The origin was maternal in the majority of the autosomal trisomies (all except one trisomy 21) and paternal in most of the sex chromosome aneuploidies (four of the five monosomies X and three of the four XXY syndrome cases).

Triple X syndrome was detected in one woman with a fetus with trisomy 18.

CONCLUSIONS

The QF-PCR method is an efficient, rapid and reliable method for prenatal diagnosis of the most common chromosome aneuploidies. In addition, it can provide information about the origin of the aneuploidy and maternal contamination of the fetal material.

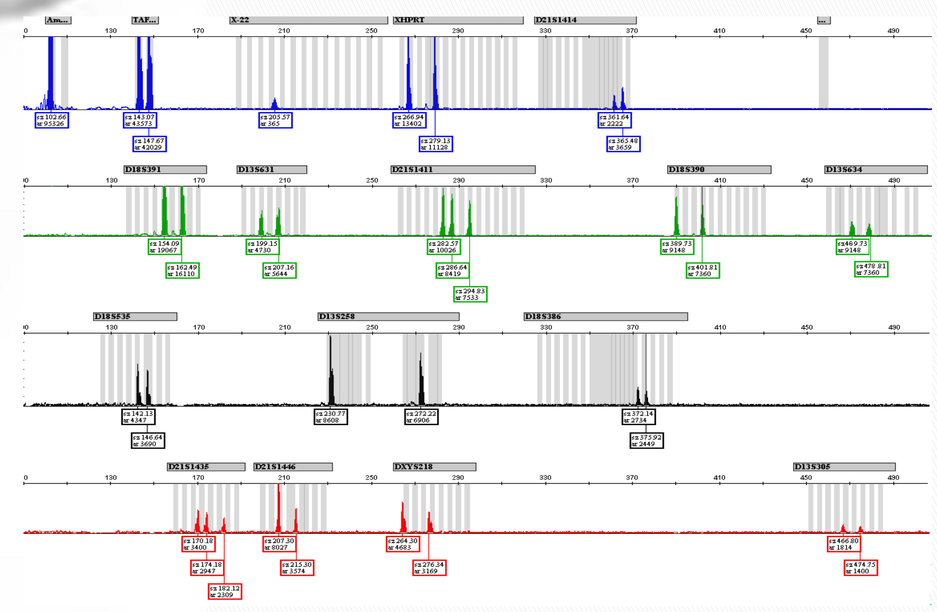

Figure 1. Electrophoreogram of a multiplex QF PCR from a female fetus with trisomy 21.

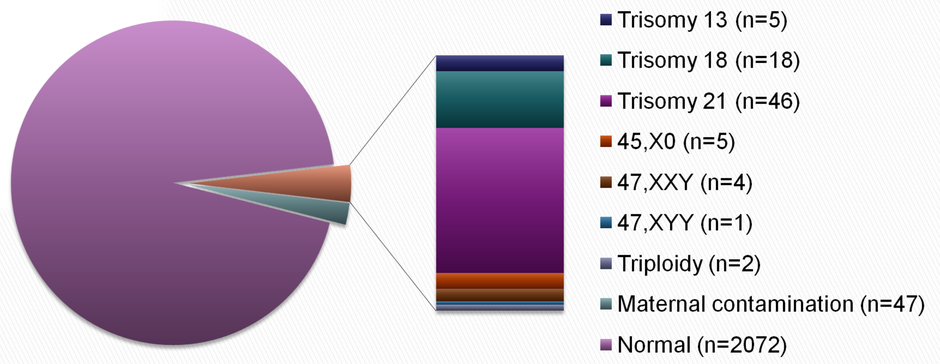

Figure 2. Results of the QF-PCR analysis for the detection of the most common chromosome aneuploidies in a total of 2200 prenatal samples.

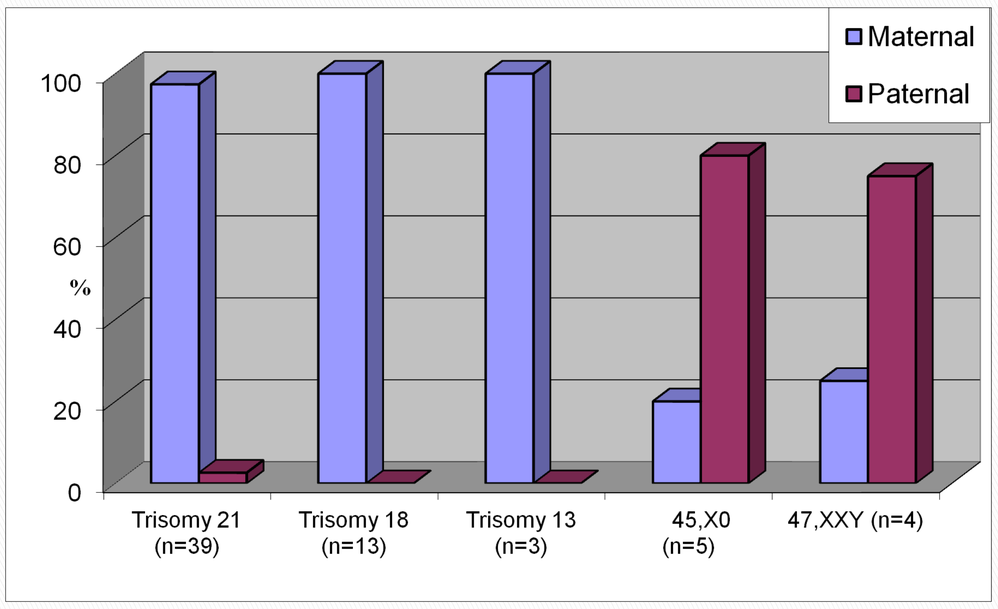

Figure 3. Parental origin of 64 prenatally diagnosed aneuploidies of chromosomes 21, 18, 13 and X.

ACKNOWLEDGEMENT

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