# Detection of aneuploidies in spontaneous abortions by quantitative fluorescent PCR with short tandem repeat markers: a retrospective study

**F.F. Coelho[1], F.K. Marques[2], M.S. Gonçalves[1], V.C.O. Almeida[3], E.C.C. Mateo[2] and A.C.S. Ferreira[2]**

[1]Departamento de Genética Molecular, Instituto Hermes Pardini, Vespasiano, MG, Brasil
[2]Pesquisa & Desenvolvimento, Instituto Hermes Pardini, Vespasiano, MG, Brasil
[3]Core Lab, Instituto Hermes Pardini, Vespasiano, MG, Brasil

Corresponding author: F.K. Marques
E-mail: fabiana.marques@labhpardini.com.br

Genet. Mol. Res. 15 (3): gmr.15038617
Received March 9, 2016
Accepted June 22, 2016
Published September 23, 2016
DOI http://dx.doi.org/10.4238/gmr.15038617



**ABSTRACT.** Approximately 10-15% of all pregnancies end in spontaneous abortions. Many factors can lead to embryonic loss; however, it has been well established that over 50% of all miscarriages result from chromosomal abnormalities, primarily aneuploidies (>96%). Identifying the cause of miscarriage can significantly reduce the psychological stress in women, and enable better genetic counseling for a future pregnancy. Quantitative fluorescent polymerase chain reaction (QF-PCR) has been previously used in the study of chromosomal abnormalities. In this retrospective study, the frequency of aneuploidy in samples of 130 miscarriages undergone by patients

(age average: 34.1 ± 4.6 years) at our institution was determined by QF-PCR using short tandem repeat markers. The gender of the miscarriage cases was determined by amplifying the amelogenin locus (70 males and 60 females). Seventy-one of these cases (54.6%) presented aneuploidies such as trisomy, monosomy, triploidy, and double trisomy. Trisomy 22 was the most common aneuploidy (present in 14 cases), followed by trisomy 15, trisomy 16, and monosomy X. We also observed monosomy at chromosomes X and 21 and a case with multiple aneuploidies at chromosomes 16 and 22. The most common aneuploidies associated with miscarriages were detected by QF-PCR; therefore, we concluded that QF-PCR is a rapid and reliable method for the detection of aneuploidy, and can be used as an accessory to the widely used karyotype analysis.

**Key words:** Spontaneous abortions; Aneuploidy; QF-PCR; STR markers