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Development and Validation of the "KBC-Aneuquick-V2": A QF-PCR assay for rapid prenatal diagnosis of aneuploidies

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Abstract:

Introduction:

QF-PCR is a novel, fast, cost-effective and reliable molecular technique based on PCR amplification using fluorescent primers for prenatal diagnosis of chromosomes 21, 18, 13, X and Y aneuploidies. DNA extracted from amniotic, chorionic villus samples and blood can be used as the genomic material in this method. In a multiplex assay specific STR markers for each chromosome amplified which their peaks represent the number of chromosomes. Commercial kits considered European and American populations. In some cases suspicious results had been seen in Iranian population.

Method:

"KBC-Aneuquick-V2" is a novel kit was prepared in accordance with the Iranian population allele frequencies. This kit consisted of 24 markers which have a wide range of heterozygosity and covers the entire length of each chromosome and critical regions. Markers and their primers were chosen considering the CNVs (Copy number variations) and SNPs (Single nucleotide Mutation) to avoid false-positive or false-negative results.

More than 200 unrelated individuals were tested using the "KBC-Aneuquick-V2" and Aneufast kit in parallel, results were compared with karyotyping.

Result:

In the all cases same results were obtained from "KBC-Aneuquick-V2" and "Aneufast" kits. Only in a case of XX/XY mosaicism, none of the QF-PCR kit was able to detect chromosomal mosaicism.

Discussion:

Amplification of 24 markers in only one reaction, make the users able to get the results quickly and reliably. "KBC-Aneuquick-V2" contains 3 segmental duplication markers (XY and 7X), 4 STR markers for each of Chr 13 and Chr18, 6 markers for ChrX, 6 for Chr21 and 2 markers for ChrY chromosome.

Keywords: Aneuploidy, Prenatal diagnosis, Quantitative fluorescent-polymerase chain reaction (QF-PCR), Trisomy, KBC-Aneuquick-V2