

ABSTRACT

The present invention relates to a method for determining copy number variation in a mixture of nucleic acids, which are known or believed to be different in terms of the amount of one or more target sequences, and more particularly to a method for determining copy number variation, which comprises a bioinformatic analysis method and a statistical analysis method, in order to analyze variability between chromosomes and sequences.

The method for determining variation according to the present invention may be used to chromosomal copy number variation which is associated with or believed to be associated with fetal diseases. Chromosomal copy number variations that may be determined according to the method of the present invention may include trisomy and monosomy of any one or more of chromosomes 1-22, X and Y, polysomy for the full-length nucleic acid sequence, and deletion and/or duplication of any one or more sequence fragments of chromosomes, and thus the method of the present invention is useful for the analysis of fetal gender and copy number variation.