

Abstract

METHOD AND DEVICE FOR DETERMINING CHROMOSOMAL ANEUPLOIDY AND CONSTRUCTING CLASSIFICATION MODEL

Provided is a method for determining whether a fetus has chromosomal aneuploidy, the method comprising: the method comprises: (1) obtaining nucleic acid sequencing data from a pregnant woman sample; (2) determining a fetal fraction of the pregnant woman sample and an estimated fraction by a predetermined chromosome based on the nucleic acid sequencing data; (3) determining a first feature based on a difference between the estimated fraction by a chromosome to be tested and the estimated fraction by a second comparison chromosome, and determining a second feature based on a difference between the estimated fraction by the chromosome to be tested and the fetal fraction; and (4) determining whether the fetus has an aneuploidy for the chromosome to be tested based on the first feature and the second feature by using corresponding data of control sample, wherein the control sample comprises a positive sample and a negative sample, the positive sample has an aneuploidy for the chromosome to be tested, and the negative sample does not have an aneuploidy for the chromosome to be tested.

[No Suitable Figure]