

METHOD AND SYSTEM OF DETECTING FETAL CHROMOSOMAL ABNORMALITIES

ABSTRACT

The present invention relates to the field of biotechnology. A method and system of detecting fetal chromosomal abnormalities are disclosed. The method comprises: (1) obtaining sequencing data of cell-free nucleic acid fragments and clinical phenotypic feature data from a pregnant woman to be detected, wherein the sequencing data comprise a plurality of read segments, and the clinical phenotypic feature data of the pregnant woman form a phenotypic feature vector of the pregnant woman, (2) performing window division on at least part of a chromosome sequence of a reference genome to obtain sliding windows, counting the read segments falling within the sliding windows, and generating a sequence feature matrix of the chromosome sequence; (3) inputting the sequence feature matrix into a trained machine learning model to extract a sequence feature vector of the chromosome sequence; and (4) combining the sequence feature vector and the phenotypic feature vector of the pregnant woman to form a combined feature vector, and inputting the combined feature vector into a classification detection model to obtain the fetal chromosomal abnormality state of the pregnant woman to be detected.

Fig. 1