

ABSTRACT

The present invention relates to the field of biotechnology. Disclosed is a method for detecting a fetal genotype on the basis of a haplotype. The method comprises: 1) performing cfDNA sequencing on the plasma sample of pregnant woman to be tested, wherein the haplotyping of the mother and the father is known, and obtaining sequencing reads; 2) aligning the sequencing reads to a reference genome, and estimating the depth distribution and mutation of the sample on the basis of the alignment results; 3) with regard to four types of allelic loci, calculating the concentration of the fetal sample according to the minor allele frequency, wherein the four types of allelic loci comprise a locus where the mother is homozygous and the fetus is homozygous, a locus where the mother is homozygous and the fetus is heterozygous, a locus where the mother is heterozygous and the fetus is homozygous, and a locus where the mother is heterozygous and the fetus is heterozygous; and 4) with regard to the genotype combinations AAAB, ABAA and ABAB of the mother and the father, inferring the fetal SNP and InDel genotypes. The method of the present invention can be used for genome-wide single-site variant detection, provides accuracy and effectively reduces the sequencing depth.

FIG. 1