

Abstract

Provided are a method and a device for detecting genetic mutations and a kit for genotyping of pregnant women and fetuses. The method comprises: performing high-throughput sequencing on cell free DNA in pregnant women's peripheral blood to obtain sequencing data; aligning the sequencing data with reference genome to obtain SNP sites; performing mixed genotyping at each SNP site to obtain target genotypes for each SNP site; and identifying mutant alleles that cause the gene mutations from the genotype of the fetus in the target genotypes. The method provided by the present invention does not require separate sequencing of samples from fathers or mothers, and realizes that all the possible gene mutations of the fetus can be detected by using only the sequencing data of the maternal peripheral blood, which provides convenient and diversified services for fetal genetic testing.

Accompanying Drawing of the Abstract

Step A: perform high-throughput sequencing on cell-free DNA extracted from pregnant women's peripheral blood to obtain sequencing data



Step B: align the sequencing data with reference genome to obtain SNP sites



Step C: perform mixed genotyping for each SNP site using Bayesian model and an initial fetal concentration f to obtain a mixed genotype with the maximum probability among seven mixed genotypes for each SNP site, and take the mixed genotype with the maximum probability as the target mixed genotype of each SNP site



Step D: identify the mutant alleles that cause the gene mutation of the fetuses according to the genotype of the fetus in the target mixed genotypes