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(54) Title: METHODS AND COMPOSITIONS FOR NONINVASIVE PRENATAL DIAGNOSIS OF FETAL ANEUPLOIDIES

(57) Abstract: The invention provides methods and compositions for noninvasive prenatal diagnosis of fetal aneuploidies. A large panel of differentially methylated regions (DMRs) have been identified. Certain of these DMRs are hypomethylated in adult female blood DNA and hypermethylated in fetal DNA, whereas others are hypermethylated in adult female blood DNA and hypomethylated in fetal DNA. Moreover, DMRs that are hypomethylated in adult female blood DNA and hypermethylated in fetal DNA have been shown to accurately predict a fetal aneuploidy in fetal DNA present in a maternal blood sample during pregnancy. In the methods of the invention, hypermethylated DNA is physically separated from hypomethylated DNA, preferably by methylated DNA immunoprecipitation.

