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(54) Title: IDENTIFICATION OF DIFFERENTIALLY REPRESENTED FETAL OR MATERNAL GENOMIC REGIONS AND USES THEREOF

(57) Abstract: The present invention provides a novel approach for identification and characterization of differentially represented fetal or maternal genomic regions in maternal circulation. Identification of overrepresented fetal genomic regions in the maternal circulation according to the present invention permit accurate analysis of fetal DNA without the need for enrichment or purification, which provides a simpler, more accurate and efficient prenatal diagnosis in early pregnancy. The present invention is particularly useful for noninvasive prenatal diagnosis during early pregnancy (e.g., during the first trimester).



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