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### Lo et al.

#### (54) DIAGNOSING FETAL CHROMOSOMAL ANEUPLOIDY USING MASSIVELY PARALLEL GENOMIC SEQUENCING

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### (57) ABSTRACT

Embodiments of this invention provide methods, systems, and apparatus for determining whether a fetal chromosomal aneuploidy exists from a biological sample obtained from a pregnant female. Nucleic acid molecules of the biological sample are sequenced, such that a fraction of the genome is sequenced. Respective amounts of a clinically-relevant chromosome and of background chromosomes are determined from results of the sequencing. A parameter derived from these amounts (e.g. a ratio) is compared to one or more cutoff values, thereby determining a classification of whether a fetal chromosomal aneuploidy exists.





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FIG. 3B

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FIG. 4A



FIG. 4B

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