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

**Related U.S. Application Data**

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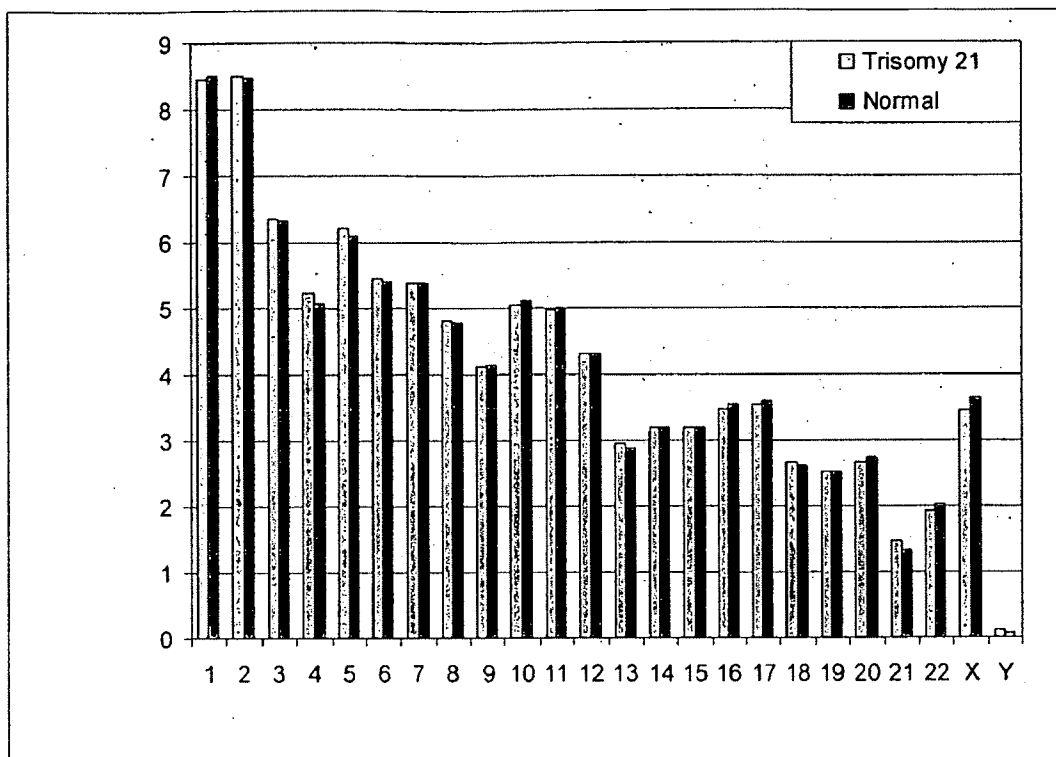
(57) **ABSTRACT**

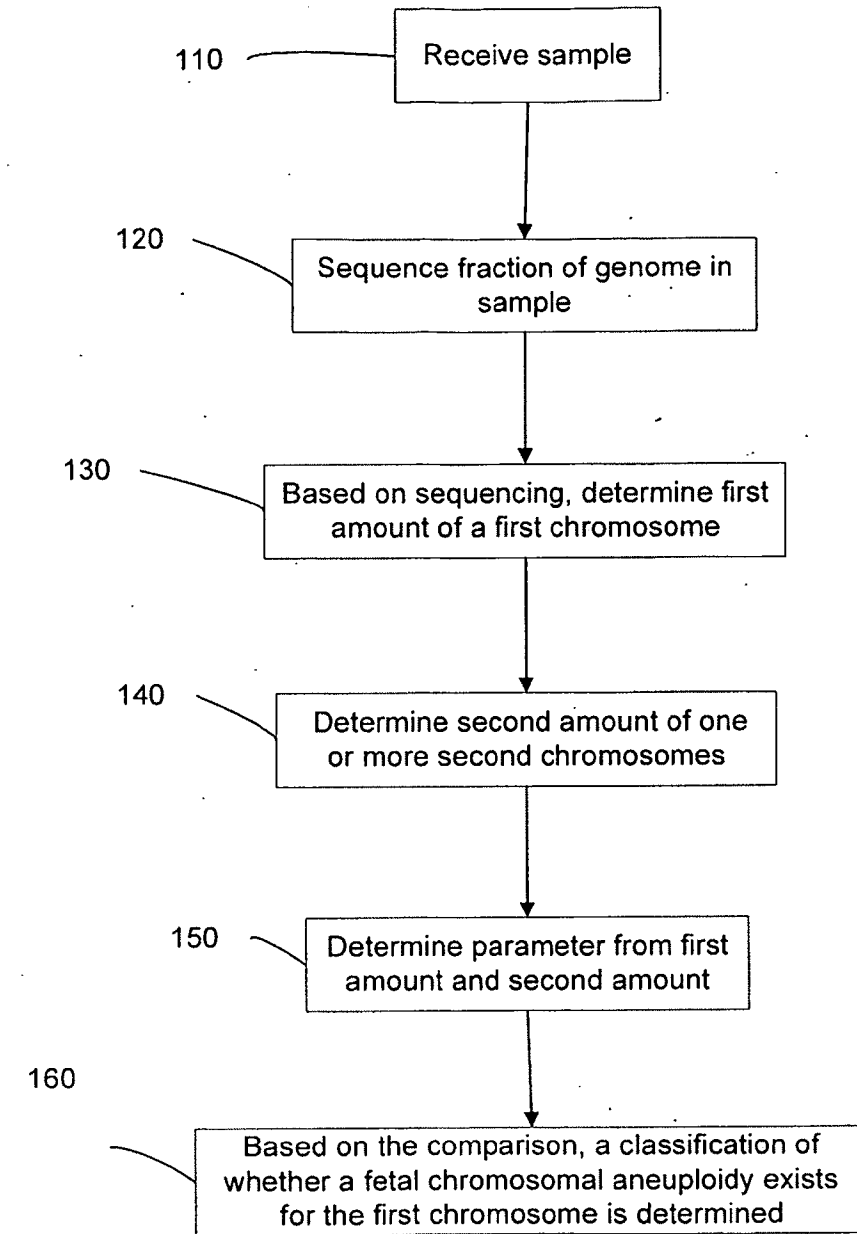
Embodiments of this invention provide methods, systems, and apparatus for determining whether a fetal chromosomal aneuploidy exists from a biological sample obtained from a pregnant female. Nucleic acid molecules of the biological sample are sequenced, such that a fraction of the genome is sequenced. 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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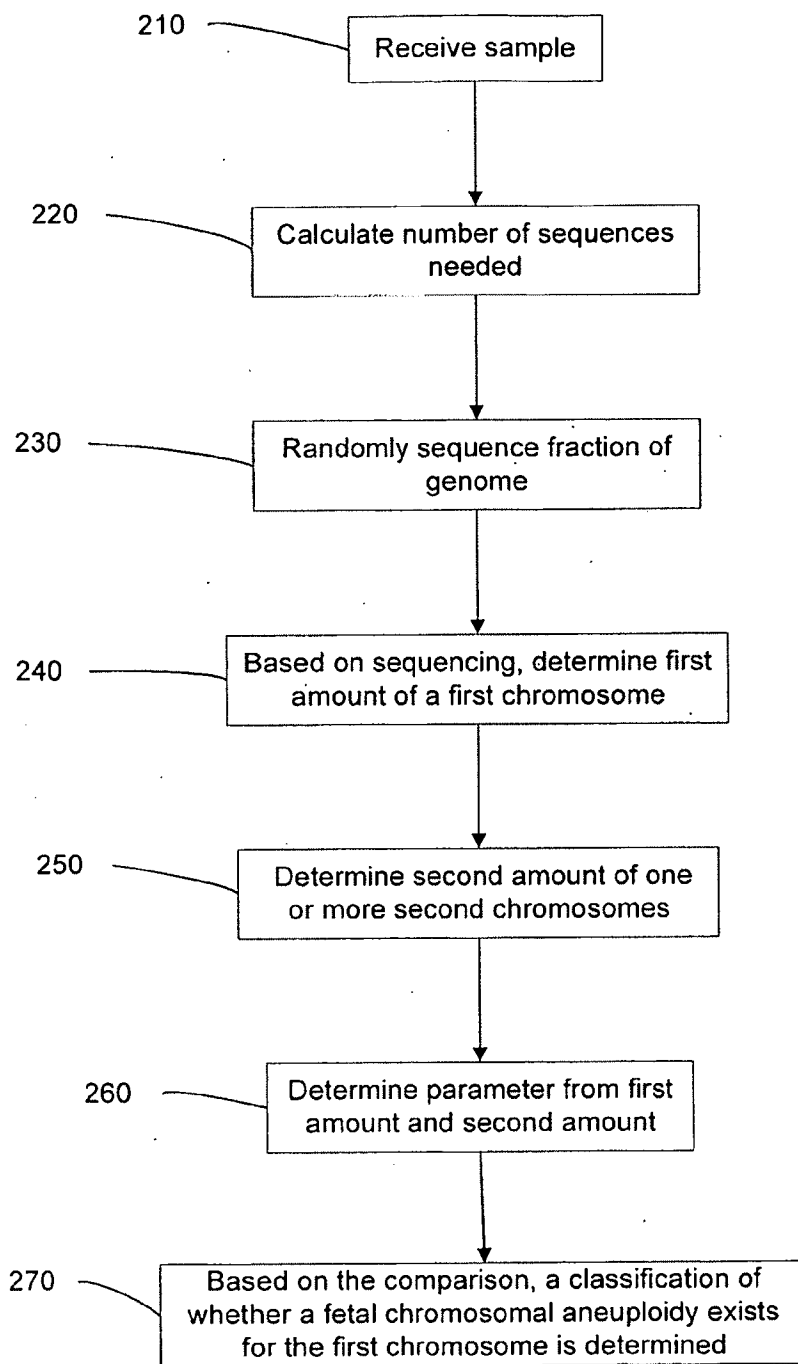
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100

FIG. 1



200

FIG. 2

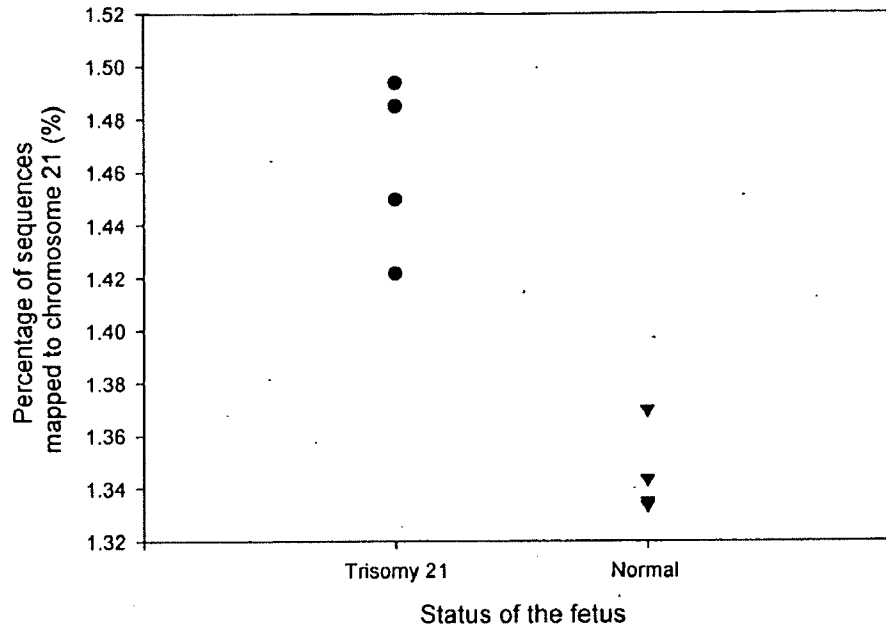


FIG. 3A

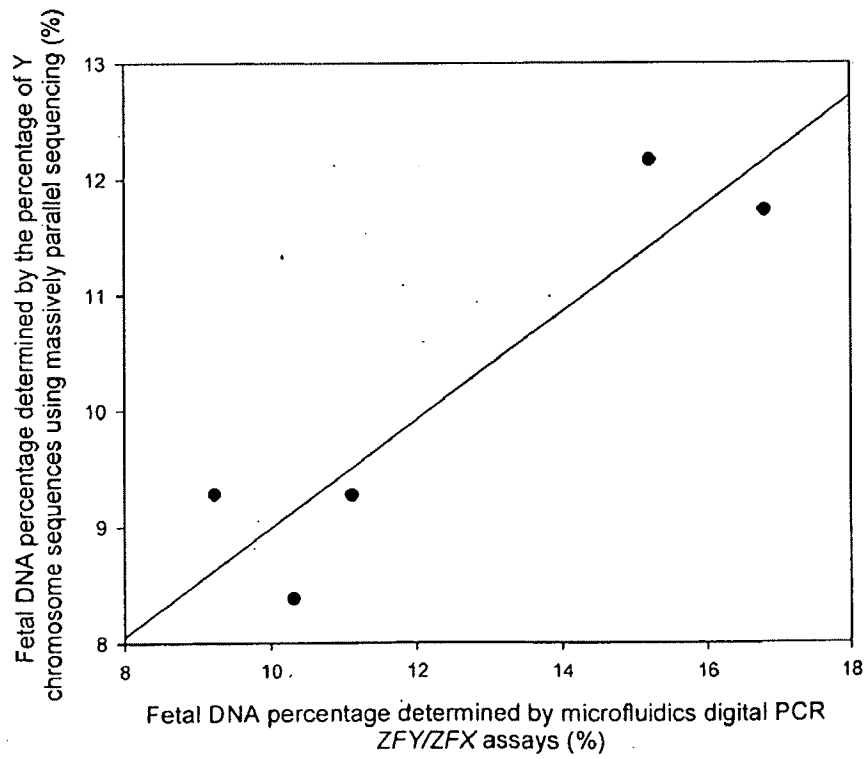


FIG. 3B

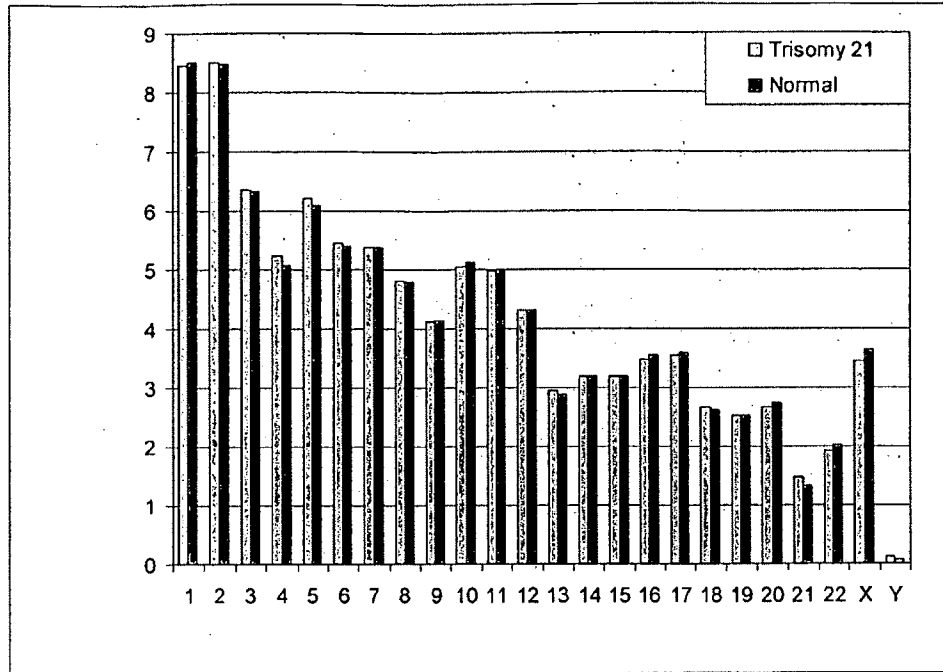


FIG. 4A

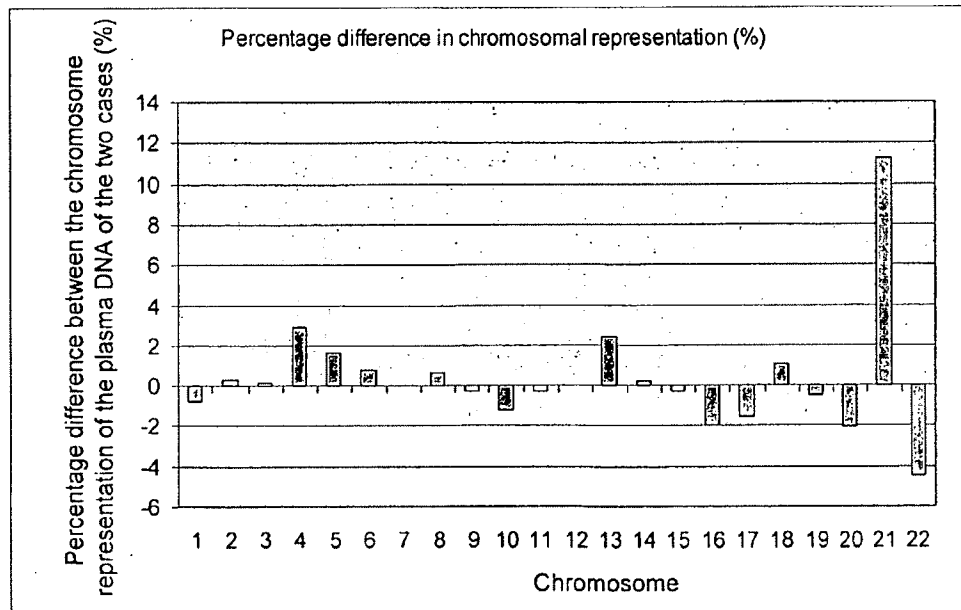


FIG. 4B

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