# DIAGNOSING FETAL CHROMOSOMAL ANEUPLOIDY USING GENOMIC SEQUENCING


## 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.


Fig. 1