DIAGNOSING CANCER~~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