DIAGNOSING CANCER USING GENOMIC SEQUENCING

~~DIAGNOSING FETAL CHROMOSOMAL ANEUPLOIDY USING GENOMIC SEQUENCING~~

CLAIM OF PRIORITY

[0001] The present application claims priority from and is a non-provisional application of U.S. Provisional Application No. 60/951438, entitled "DETERMINING A NUCLEIC ACID SEQUENCE IMBALANCE" filed July 23, 2007 (Attorney Docket No. 016285-005200US), the entire contents of which are herein incorporated by reference for all purposes.

CROSS-REFERENCES TO RELATED APPLICATIONS

[0002] The present application is also related to concurrently filed non-provisional application entitled "DETERMINING A NUCLEIC ACID SEQUENCE IMBALANCE," (Attorney Docket No. 016285-005210US) the entire contents of which are herein incorporated by reference for all purposes.

FIELD OF THE INVENTION

[0003] This invention generally relates to the diagnostic testing of fetal chromosomal aneuploidy by determining imbalances between different nucleic acid sequences, and more particularly to the identification of trisomy 21 (Down syndrome) and other chromosomal aneuploidies via testing a maternal sample (e.g. blood).

BACKGROUND

[0004] Fetal chromosomal aneuploidy results from the presence of abnormal dose(s) of a chromosome or chromosomal region. The abnormal dose(s) can be abnormally high, e.g. the presence of an extra chromosome 21 or chromosomal region in trisomy 21; or abnormally low, e.g. the absence of a copy of chromosome X in Turner syndrome.

[0005] Conventional prenatal diagnostic methods of a fetal chromosomal aneuploidy, e.g., trisomy 21, involve the sampling of fetal materials by invasive procedures such as amniocentesis or chorionic villus sampling, which pose a finite risk of fetal loss. Non-invasive procedures, such as screening by ultrasonography and biochemical markers, have been used to risk-stratify pregnant women prior to definitive invasive diagnostic procedures. However, these screening methods typically measure epiphenomena that are associated with the chromosomal aneuploidy, e.g., trisomy 21, instead of the core chromosomal abnormality, and thus have suboptimal diagnostic accuracy and other disadvantages, such as being highly influenced by gestational age.