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(54) **NON-INVASIVE FETAL GENETIC SCREENING BY DIGITAL ANALYSIS**

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(58) **Field of Classification Search** ..... None
See application file for complete search history.

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(57) **ABSTRACT**

The present methods are exemplified by a process in which maternal blood containing fetal DNA is diluted to a nominal value of approximately 0.5 genome equivalent of DNA per reaction sample. Digital PCR is then be used to detect aneuploidy, such as the trisomy that causes Down Syndrome. Since aneuploidies do not present a mutational change in sequence, and are merely a change in the number of chromosomes, it has not been possible to detect them in a fetus without resorting to invasive techniques such as amniocentesis or chorionic villi sampling. Digital amplification allows the detection of aneuploidy using massively parallel amplification and detection methods, examining, e.g., 10,000 genome equivalents.

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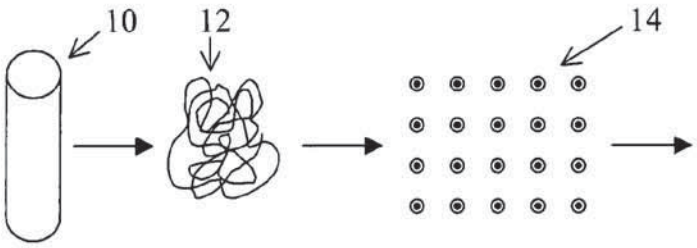

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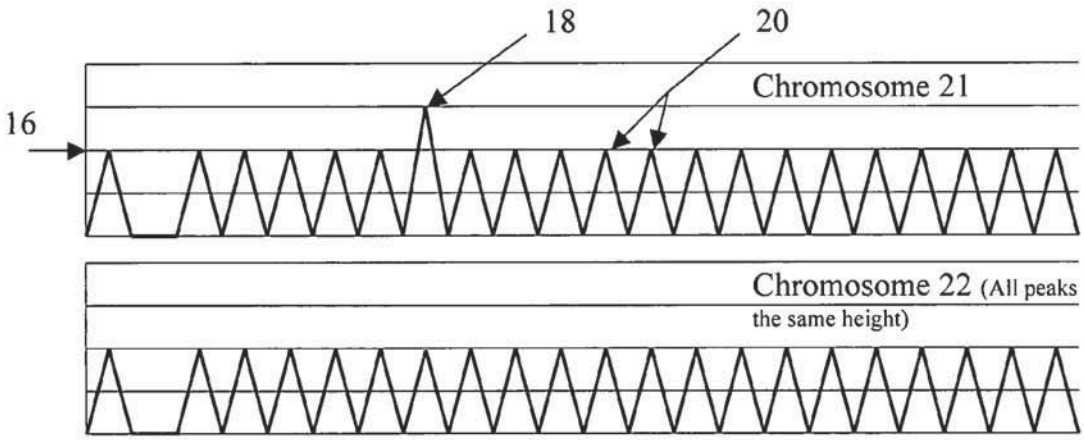
Fig. 1A



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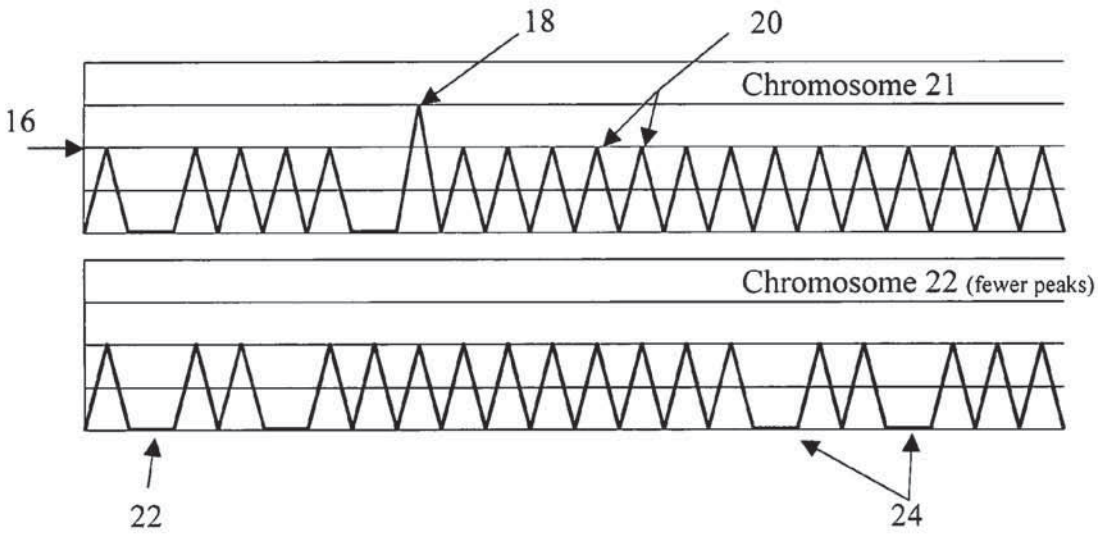

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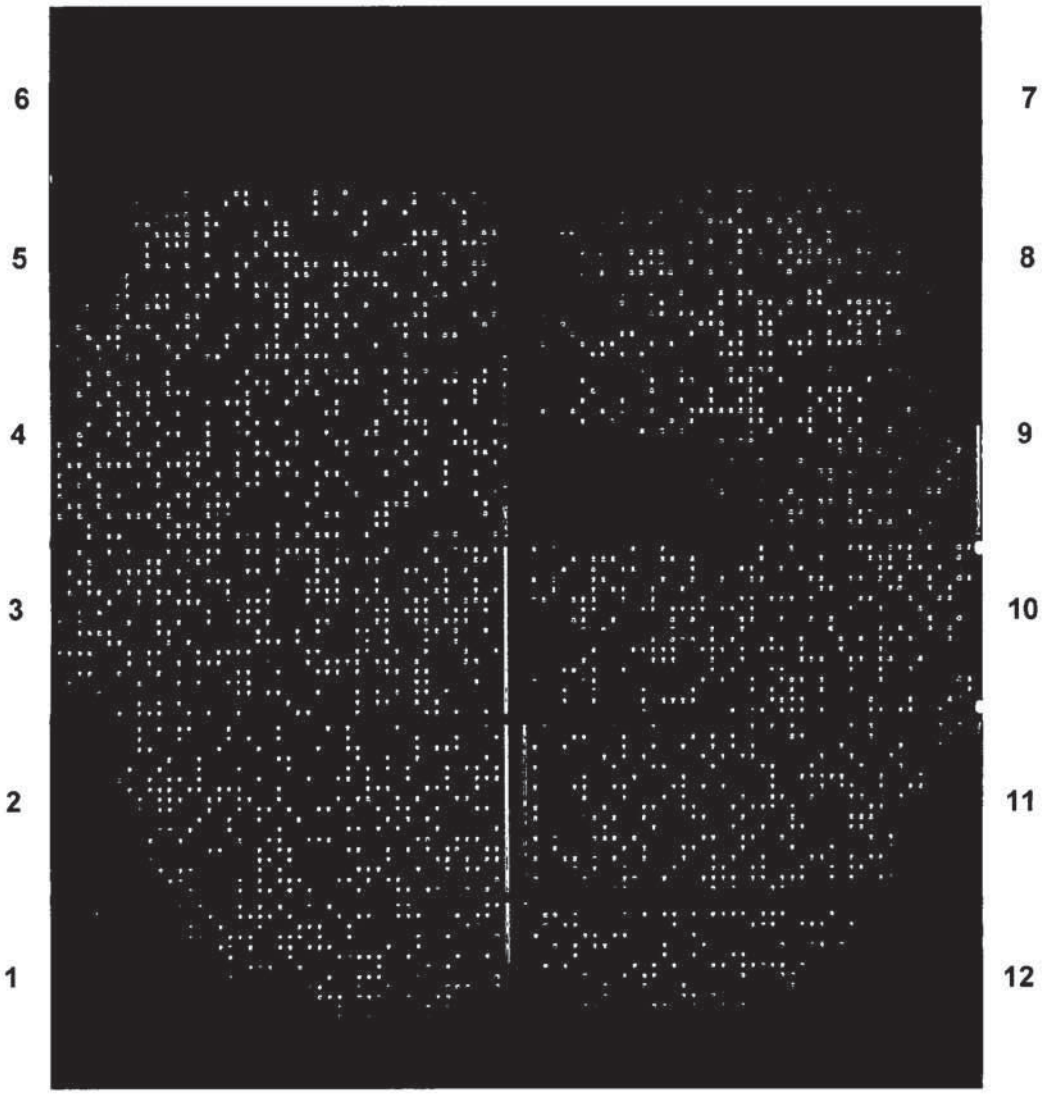

Fig. 2

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