Prenatal Diagnostic Testing

Description: This report discusses improvements in invasive procedures (karyotyping vs. cytogenetics), and the rise of noninvasive screening methods and their impact on prenatal healthcare. Although covering standard invasive procedure, this report focuses on the noninvasive technologies either on the market or in development.

The first chapter of this report discusses microarray-based cytogenetics and how it compares to karyotyping. Karyotyping is often the standard method used to detect chromosomal aneuploidies, but with microarray-based procedures even more aneuploidies can be detected with the same efficacy. Either method can be used in sync with noninvasive procedures that show positive screening results for aneuploidies, but microarray analysis may be more useful for copy number variations that often go undetected with karyotyping.

Highlighting noninvasive methods, several companies have come out with novel technologies that allow them to analyze fetal DNA without risk of injury to the mother or fetus. Amniocentesis and chorionic villus sampling are common methods used to analyze fetal DNA for aneuploidies but at the risk of miscarriage and infection. With noninvasive procedures, screening can be done for common aneuploidies and further analysis can be done on women with only positive results. This not only decreases the amount of unnecessary invasive testing but also the amount of unnecessary stress levels of many expecting mothers. Companies participating in this section include:

- Sequenom
- Verinata Health
- Ariosa Diagnostics
- Natera
- Silicon Biosystems

Other areas discussed in this report include preeclampsia and preterm labor. Preeclampsia is a common condition in women and often isn't detected until later in pregnancy. It can lead to preterm labor, which may be fatal for both the fetus and the mother. So far, a test to screen for the likelihood of preeclampsia does not exist. Detecting the risk of preeclampsia early in pregnancy can allow the expecting mother to take necessary action to avoid preterm labor at all costs, decreasing any complications that may severely affect the fetus including death.

Companies highlighted in this section include:

- Metabolomic Diagnostics
- NX PharmaGen

In addition to covering several aspects of prenatal diagnostics, this report also includes specifications of particular instruments, exclusive interviews with CEOs, CSOs, and Vice Presidents of the participating companies. Specific areas of focus include company goals, customer feedback, challenges encountered, competitive advantage, platform pricing, and future endeavors. A survey capturing the demographics of the prenatal industry is presented at the end of the report, specifically highlighting organizational background, areas of research, challenges encountered, and applications in development.

Also included is a directory of companies that were and were not interviewed for this report.

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References

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