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NON INVASIVE PRENATAL TESTING

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The landscape of prenatal testing has changed dramatically over the past five years, when the first versions of cell-free DNA sequencing hit the U.S. market. Since then the cell-free tests have gained rapid acceptance, by clinicians, payors and consumers, and the number of cell-free test providers has expanded and become that much more competitive. One result has been a sharp decrease in the rate of invasive procedures, where arrays and sequencing are leading the shift to molecular cytogenetics.
Numerous issues and controversies remain, including how broadly cell-free DNA testing should be offered, to what extent cell-free DNA testing should be extended to which sub-chromosomal or copy number coverage, and what the impact may be once non-invasive diagnostics based on isolation of fetal cells from maternal blood become commercially available.
Earlier on in the 1980's more difficult b/c were looking for whole fetal cells (very difficult to find and isolate). These cells also can persist in maternal circulation for years (ie in the next pregnancy).
- We are now looking at cell free fetal DNA.The cell free fetal DNA is thought to be placental in origin and crosses from the placenta into maternal circulation as cells break down (apoptosis) and the DNA becomes fragmented
This cell free fetal DNA leaves mom's circulation within hours of birth (so not persistent from year to year and pregnancy to pregnancy.
- Define Fetal Fraction: The proportion of DNA in maternal blood that is fetal in origin is the fetal fraction.
If you were to isolate the cell free DNA, and 90% of it was maternal in origin and the other 10% was fetal in origin, the fetal fraction of that sample would be defined as 10%.
- Fetal fraction is variable person to person, tends to be lower earlier in pregnancy, can vary with maternal weight, etc.
A metric that is often overlooked in the evaluation of NIPT test performance is the rate of test failures or no-call results. This is the percentage of patients whose results are not reported, indeterminate, or uninterpretable, because of laboratory technical issues.
Finally we will talk about new indications and what's next regarding nipt, monogenic disorders, whole genome etc.
This is really a rapid and expanding field that changes and gives the opportunity to the clinicians and the patients do go deeper in the counselling and diagnosis of several genetic conditions afecting pregnancy.

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