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# Impact of cell free DNA analysis on invasive testing in a tertiary referral Fetal Medicine Centre

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## Objective

Impact of cell free fetal DNA (cff DNA) testing on invasive procedures (Chorion villous sampling/ Amniocentesis) in a tertiary referral fetal medicine centre and follow up of the "high risk" and "no call" reports.

## Methods

Between October 2014 and March 2017, following first or second trimester screening for aneuploidies, 1632 women chose to have either invasive or cff DNA testing. 248 (15. 1%) mothers underwent cff DNA testing in comparison to 304 (18. 62%) mothers who underwent CVS and 1080 (66. 17%) mothers who underwent amniocentesis. A comparative study was undertaken based on the indication. Fetuses with structural abnormality were excluded from all the three groups.

## Results

Cff DNA was performed in 248 mothers, of which 119 (47. 9%) was in the first trimester and 129 (52. 1%) in the second trimester. The most common indication for cff DNA testing, CVS and amniocentesis was maternal age above 35years [performed in 51(20. 5%) mothers], increased risk on combined first trimester screening [performed on 65(54%) mothers] and increased second trimester biochemistry risk [performed on 207(19%) mothers] respectively. Other indications included were presence of 2nd trimester soft markers and previous baby or family history of aneuploidy. Of 65 mothers with age above 35 years, 51 (68%) chose to have further reassurance with cff DNA testing, 4 (5. 3%) underwent CVS and 10 (1. 3%) had an amniocentesis even after a normal first trimester or a second trimester screening. Of the 273 mothers who had increased risk on the first trimester screening with a normal fetal scan, 117 (42. 8%) had amniocentesis, 91 (33. 3%) had cff DNA and 65 (23. 8%) mothers underwent CVS. Of the 229 mothers who had a "high risk" result on the second trimester biochemical screening with normal fetal scan, 207 (90. 3%) mothers had amniocentesis and 22 (9. 6%) had cff DNA testing. Out of the 248 mothers who had cff DNA testing, 5(2. 1%) results were reported as "high risk", who underwent invasive testing. Aneuploidy was confirmed in 3 fetuses (60%) (2 x T21 and 1 sex aneuploidy). There were 2 (40%) false positive results for T21. 2 more out of 248 (0. 8%) mothers with no results had a normal karyotype confirmed on amniocentesis. 241 (97. 1%) results were reported as "low risk".

## Conclusion

In the absence of "increased risk" based on first or second trimester screening, mothers are more likely to choose non invasive testing, especially when above the age of 35 years for further reassurance. However, in presence of a "high risk" result on combined first trimester or second trimester screening, majority of the mothers chose to undergo invasive testing for further reassurance. With a "high/ no" result on cff DNA testing, confirmatory test preferably, amniocentesis is mandatory as the "false positive rate" can be significantly high, as in our series it is 4/ 7 (57. 1%).