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Advance pre-natal screening to curb fetal mortality

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Worldwide, approximately 76,000 pregnant women and approximately half million babies die annually because of pre-eclampsia and severe preeclampsia. In India, every year roughly 12 percent (~3 Million) of pregnancies develop pre-eclampsia and this condition claims roughly five percent maternal lives.

Pre-eclampsia, in layman's language, is a pregnancy complication that causes high blood pressure, kidney damage, and other problems. Preeclampsia and subsequent disorders can lead to seizure, stroke, multiple organ failure and death of the mother and/or baby.

Incremental evidence proposes that evaluating the risk for these complications at an early stage of pregnancy can provide clinicians with opportunities to intervene in a timely manner. First trimester screening (FTS) has been emphasized in recent times for most relevant complications affecting a mother and her unborn child at 11–13 (+6) weeks of pregnancy by Fetal Medicine Foundation (FMF).

Over the last few years, we have witnessed some breakthrough technological innovations in the field of pre-eclampsia.

Pre-eclampsia is a medical calamity for the mother and baby and can lead to extra cost burden for the country. By integrating biomarkers driven preeclampsia screening and management in current clinical practice, over 95% preeclampsia can be accurately predicted and fortunately, 82-90 percent of early onset preeclampsia can be prevented with timely intervention using aspirin which is cost effective.

Early detection of the biomarkers sFlt-1 (soluble FMS-like Tyrosine Kinase) and PLGF (Placental Growth Factor) in maternal blood have shown to significantly improve risk stratification in women for pre-eclampsia evaluation.

Prenatal detection for chromosomal abnormalities have been available for more than 40 years, first by amniocentesis in the early 1970s and additionally by chorionic villus sampling (CVS) methods. Invasive prenatal diagnosis such as CVS and amniocentesis pose threats of miscarriage for all low-risk mothers. Non-Invasive Prenatal Screening (NIPS) are the most promising among the various non-invasive tests developed so far. New evidence strongly suggests the integration of Non-

Invasive Prenatal Screening using cell-free DNA (NIPS) into prenatal care after conventional screening can substantially improve the detection rate for Down syndrome (Trisomy 21)), Edwards syndrome (Trisomy 18), Patau Syndrome (Trisomy 13) and across the maternal age spectrum at 9–10 weeks gestational period. The NIPS test screens maternal blood samples for chromosome aneuploidy in fetal DNA and confers an accuracy of up to 99% on the detection of fetal chromosome aneuploidy.

Preimplantation Genetic Screening (PGS) is a test that comprehensively screens all the chromosomes (Chromosome 1-22 and the sex chromosomes) in an embryo biopsy sample for any extra or missing chromosomes, and large deletions or duplications of genetic material. PGS is recommended by many top fertility specialists as selecting embryos with the correct number of chromosomes for transfer has been shown to significantly improve implantation outcomes and pregnancy success rates.

The field of fetal medicine is expanding and the diagnostic possibilities are limitless, which has triggered a shift in paradigm of screening. Application of advance prenatal testing has revolutionized the diagnostic and treatment procedure in favor of better prediction and improved individual outcomes leading to great reduction in maternal and fetal mortality with a better quality of life for the babies who come into this world.

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