# DBT-NIBMG seeks to understand role of maternal genome in birth Outcome

New Delhi, Feb 22: Preterm birth (PTB), defined as live birth before 37 completed weeks of gestation, is a major cause of neonatal and infant mortality worldwide. Every year about 15 million babies are born preterm worldwide, among which India alone contributes 3.5 million (23.4%) cases.

Preterm infants suffer from a number of health complications like respiratory disorders, cardiovascular disorders, infections, feeding difficulties, visual and hearing problems and learning disabilities. They are also at increased risk of disorders like obesity, metabolic syndrome, hypertension, type 2 diabetes in their early life. Moderate to late preterm infants comprise a larger proportion of the preterm infants. Thus, their health complications contribute substantially to the global health burden.

Preterm birth is influenced by both environmental and genomic risk factors. Twin studies demonstrated that heritability estimates of PTB ranges between 17% to 40%. It is interesting to note that PTB is defined by the genome of two individuals - the mother and the fetus. Epidemiological studies provided evidence of increased maternal genetic contribution to PTB as compared to fetal genetic contribution.

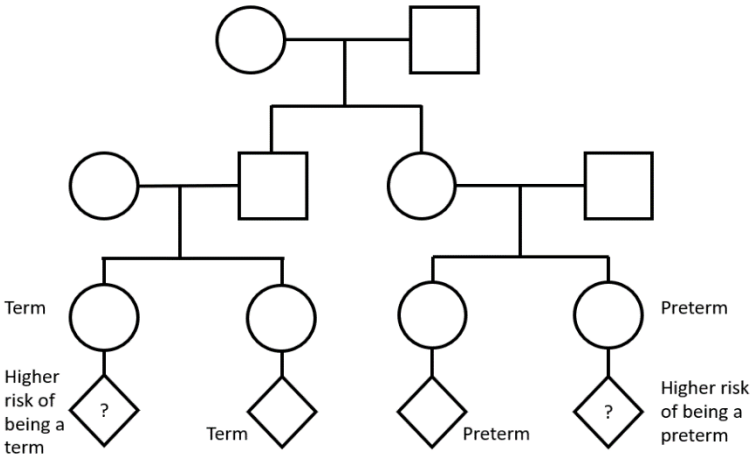


Fig : Family study and intergenerational study providing evidence of genetic contribution to preterm birth. Women whose sisters have delivered a preterm baby, have increased risk of delivering a preterm baby as compared to those women whose sisters have delivered at term. Moreover, women who themselves were born preterm have higher risk of delivering a preterm baby as compared to the mothers who themselves were born at term.

Most studies undertaken till date conducted candidate-gene studies which depend upon a prior hypothesis where genes are selected based on suspected functional relevance to a phenotype. Since, there is still no clear idea about all of the biological pathways that lead to PTB, hypothesis-free genome-wide approach might be able to capture all the potential loci throughout the entire genome which otherwise might be missed by candidate gene approach.

Although South-Asian countries contribute a substantial number of preterm births globally, no genetic studies have been conducted there yet. Since genomic biomarkers are stable as compared to other biochemical biomarkers and has the potential to predict risk of preterm birth even before pregnancy,

researchers at DBT-National Institute of Biomedical Genomics (DBT-NIBMG) are conducting a genome-wide association study in pregnant women enrolled in GARBH-Ini (interdisciplinary Group for Advanced Research on BirtH outcomes- DBT INdia Initiative) cohort, India with a definitive outcome (term and preterm), to identify maternal genomic variants that are associated with preterm birth. This will help to identify those women who are at high risk of delivering preterm and personalized care and medical attention can be provided to them to prevent preterm birth.

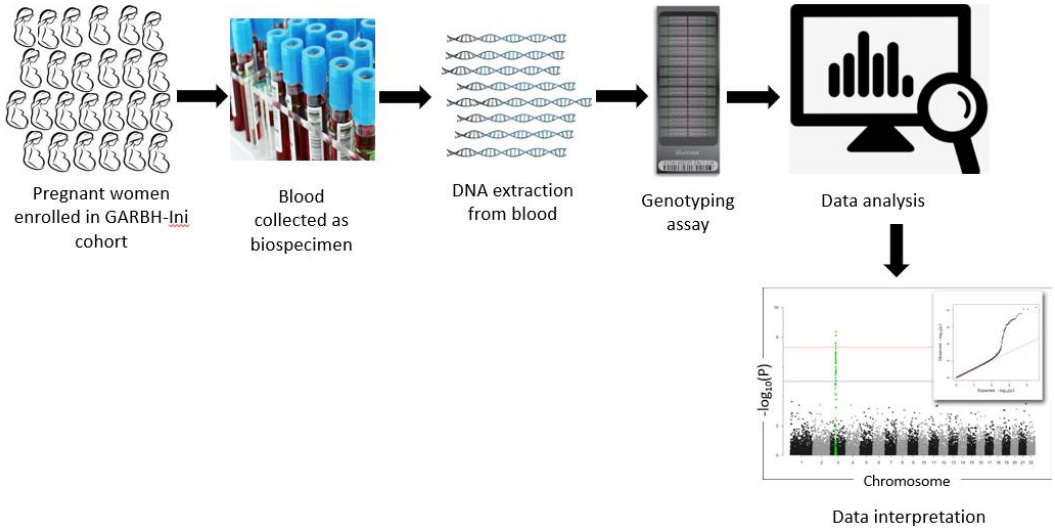


Fig 2: Genome-wide association study in GARBH-Ini cohort to identify genetic biomarker of preterm birth

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