Prematurity Genetics and the Environment

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Preterm birth, a major cause of neonatal mortality world-wide, has a number of etiologies. It has been difficult to quantify the maternal and fetal contributions and those of the environment. Recently, using novel twin-based study designs, we established the relative contributions of maternal and fetal genetics, common environmental factors and environmental factors unique to a pregnancy among women of European and African decent. Both maternal and fetal genetics made a contribution to gestational age at delivery for women of European decent, but unique environmental factors were the most significant factor. In women of African decent, unique environmental factors were even more significant. These findings established population-specific contributors and demonstrated the importance of gene x environmental interactions or environmental exposures alone to risk of prematurity. Consistent with these notions, we demonstrated an interaction of a maternal TNFA SNP with bacterial vaginosis and spontaneous preterm birth. We subsequently intensively studied the vaginal microbiome of women of European and African decent by sequencing 16s rDNA. Our results revealed significant differences in the vaginal microbiomes of the two groups, and identified taxa relevant to these differences. We found major community types dominated by Gardnerella vaginalis and BVAB1 that were common among African Americans. Ethnicity, pregnancy, and alcohol use correlated significantly with the relative abundance of bacterial vaginosis-associated species. Importantly, whole genome sequencing of Gardnerella, Sneathia and Mycoplasma isolates associated with bacterial vaginosis and chorioamnionitis disclosed novel genes that could potentially contribute to virulence and preterm birth. These findings suggest that characterization of microbial communities by 16s rDNA profiles alone may be insufficient to identify the specific bacteria that interact with the maternal and fetal genomes to promote preterm birth.

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