Genetic and Environmental Factors Increase Risk of Preterm Birth

San Diego – October 24 - Infants born preterm, before the 37th week of gestation, are more likely to require longer hospitalization, suffer from long-term health concerns and have an increased rate of mortality than babies born after the 37th week. About 1 in every 8 babies (or 12%) are born preterm. While there are some known “at risk” groups, including mothers who have had a prior preterm birth and women who are pregnant with multiple fetuses, these cases account for only one-half of preterm deliveries.

From an ongoing NIH-funded molecular and genetic epidemiological study of prematurity lead by PI Dr. Xiaobin Wang, Dr. Hui-Ju Tsai and colleagues (Children’s Memorial Hospital; Northwestern University Feinberg School of Medicine; Boston University School of Medicine; University of Illinois at Chicago School of Public Health) investigated the relationship between two metabolic genes (CYP1A1 and GSTT1) and maternal smoking with preterm birth as a whole and preterm subgroups by mode of delivery, degree of prematurity, and major pregnancy complication in a large multi-ethnic population of mothers. Their study confirmed the effect of maternal smoking on preterm birth across all ethnic groups, and revealed that maternal smoking significantly increased the risk of preterm among women with high-risk CYP1A1 and GSTT1 genotypes. Such joint associations were strongest among preterm accompanied by histologic chorioamnionitis.

Two additional studies that will also be presented at the ASHG Annual Meeting, lead by PI Dr. Scott Williams have looked at the genetic factors that increase the risk of preterm birth. Drs. Digna R. Velez, Ramkumar Menon, Stephen Fortunato and colleagues at Vanderbilt University and The Perinatal Research Center in Nashville, TN completed a large-scale high-throughput candidate gene association study examining association with SNPs in 134 genes in maternal and fetal DNA involved in preterm birth molecular pathways. In maternal samples, the strongest associations were found in genes in the coagulation-complement pathway and are related to decidual hemorrhage during preterm birth. In fetuses, the single strongest effect was found in the inflammatory pathway of the IL10 receptor antagonist gene.

In an independent study, also from Vanderbilt University and The Magee-Women’s Research Institute, Kelli K. Ryckman and colleagues examined racial differences in genetic factors affecting cytokine concentrations and the interactions of this genetic variation with bacterial vaginosis. Bacterial vaginosis is known to be associated with preterm birth. Effects of genetic variants differed by race, and in Caucasians there were SNPs found that significantly affect IL1β cytokine levels, while in African Americans the strongest association was with a SNP affecting IL1α levels.

These studies help scientists to understand the complex genetic and environmental factors associated with an increased risk of preterm birth, emphasizing both genetic differences found among populations and environmental risks such as mothers’ smoking habits. With further clarification of the risks, better advice may be given to mothers so that more preterm births may be avoided.

For members of the press who would like to register for the meeting, please contact Jane Nelson, Media Relations, the American Society of Human Genetics, 9650 Rockville Pike, Bethesda, MD 20814 Tele: 301-634-7308 (em: jnelson@ashg.org) Please visit the ASHG website to find out more information on the 57th Annual Meeting at www.ashg.org. During the meeting she can be reached at the San Diego Convention Center (619-525-6413).