

Gene linked to preterm birth among Hispanic women

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Researchers at Yale School of Medicine have found that the gene ENPP1 is linked to preterm birth and low birth weight among Hispanic women.

Errol Norwitz, M.D., associate professor in the Department of Obstetrics, Gynecology & Reproductive Sciences at Yale, will present preliminary results from this research at the Society for Maternal Fetal Medicine Annual Meeting on February 2 in Dallas, Texas.

One out of eight babies in the United States is born prematurely—delivery prior to 37 weeks gestation. These babies don't fare as well as their full-term counterparts, especially if they are born prior to 28 weeks gestation. In many cases, it is still unclear why preterm births occur, but Norwitz said that both the genetic make-up of the mother and the genetic make-up of the baby play a role.

Norwitz and his collaborators sought to understand the mechanisms responsible for the onset of labor at term and how these mechanisms are either overwhelmed or short-circuited, leading to preterm birth. Some women, especially African-American women, are genetically predisposed to preterm births, even after taking into account socioeconomic status, demographics, underlying medical conditions and multiple pregnancies. Norwitz said that multiple genes or a single particular genetic variant—single nucleotide polymorphism—may be involved.

In his study, Norwitz and colleagues tried to tease out some of the



genetic factors that are important for preterm birth. They isolated DNA in blood samples from a largely Hispanic population of mothers with a history of preterm birth and compared them to the DNA of women who had only had full-term pregnancies. They then screened the DNA for 128 different genetic variations in 77 candidate genes. While these genetic variants have been known to cause clinical disorders, no one had ever investigated them in the context of preterm birth before.

Four polymorphisms were associated with premature birth, but—to the team's surprise—a variant of the ENPP1 gene was the one most closely linked. ENPP1 has been associated with insulin resistance, glucose intolerance and a risk of developing type-2 diabetes. In certain people, it is associated with hardening of the arteries and high blood pressure. In the context of prematurity, say the researchers, it is possible that the variant form of ENPP1 is associated with deranged energy metabolism.

"In our original study, 85 percent of the population was Hispanic," said Norwitz. "It appears that there are genetic variations unique to each ethnic population. We are now in the process of validating our findings in African-American, Caucasian and Native-American populations."

Source: Yale University

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