Blazing Trails in the Genetics of Preterm Births

Preterm birth is the leading cause of death of babies and children under age 5. Researchers in the Perinatal Institute at Cincinnati Children's want to change that and are making progress in doing so. A primary goal of theirs is to reduce infant mortality. One of the ways they do so is by exploring how to prevent preterm deliveries.

"We can't predict which women are at risk for preterm births. These early deliveries usually happen for unknown reasons," explains Louis Muglia, MD, PhD, former co-director of the Perinatal Institute and former director of the Center for Prevention of Preterm Birth. Work in his laboratory seeks to discover the genetic mechanisms when birth happens and how those are disrupted during preterm birth. He and his team investigate the genetics of mother and baby and the factors responsible for increasing the risk in women to have preterm births when no other risk factors are present.

A Revolutionary Discovery

This team had the breakthrough discovery that identified the first six gene regions in the mother's genome that predicted risk of preterm birth. "With the identification of these genetic regions, we hope to devise a precision medicine approach to every pregnancy," says Muglia. "Ultimately we'd like to get to the point where we can use the mother's genetic information, social determinants and health behaviors to put together a composite in the electronic medical record that would help minimize the chances for having a baby born prematurely."

The team received grants from the March of Dimes and the Bill & Melinda Gates Foundation to continue that research internationally in diverse populations, with the goal of identifying even more of these gene regions in the near future and determining how they work. In addition, Muglia has participated in research that has identified the first loci in the fetal genome that is associated with a baby's risk for preterm birth, and they are investigating how selenium deficiency correlates with preterm birth. Preliminary results are promising.

A Solid Partnership

Muglia and leadership in the Perinatal Institute are nurturing the collaboration between the Divisions of Neonatology and Human Genetics. "This partnership allows us to bring together two worlds, so we can use rapid whole genome sequencing technology to aid in taking care of critically ill children in the NICU as well as the revealing genes for prematurity," he says. "It gives us the opportunity to build a unique program in perinatal genetics at Cincinnati Children's, so we can deliver optimal care to preterm babies while at the same time reducing the number of mothers who deliver prematurely."

Grant funding awarded

$3.5 million	$12 million	$2.5 million
The Bill & Melinda Gates Foundation	March of Dimes Prematurity Research Centers	Human Placenta Project

As of January 1, 2020 Louis Muglia, MD, PhD, is the President and Chief Executive Officer at the Burroughs Welcome Fund.