While experts agree that preterm birth remains a major health challenge with causes that are numerous, complex and not always well understood, multiple sources of data support a genetic predisposition as a major factor in the etiology of preterm birth. For example, preterm births tend to recur in mothers, with the most likely time for a preterm birth being the same week as the first preterm birth. A family history of preterm birth is a key risk factor, such that women whose mothers and sisters have had a preterm delivery are more likely to enter labor prematurely. Rates of preterm birth vary among continental populations, consistent with genetic variation arising from their evolutionary histories, also indicating a role for genetics in preterm birth.

WHAT GENES CAN TELL US ABOUT PRETERM BIRTH

With a focus on understanding these genetic determinants, Dr. Scott Williams, Professor of Epidemiology and Biostatistics and Genetics and Genome Sciences at Case Western Reserve University and Dr. Ge Zhang, Associate Professor of Genetics at Cincinnati Children’s Hospital Medical Center are leading the research efforts of Theme 2, called Genetics of Unique Human Populations.

"Approximately 50% of preterm birth has no clear medical cause, and evidence strongly suggests that genetic factors contribute to some of these cases,” says Dr. Williams, an accomplished human geneticist with expertise in population studies. “In this work, we are trying to understand how genetic variation impacts the biological processes that underlie preterm birth.”

The researchers will use the latest genetic sequencing and analysis technology to identify genes associated with preterm birth and look for variations between normal populations and those prone to prematurity.

“As part of this study, we intend to compare African, Asian and European ancestries, and look at how gene variations that are most different in these populations contribute to human disease risk,” says Dr. Williams. “If we can identify those genes that contribute to the pathways of premature birth, we might be able to predict the risk and also identify new targets for which to develop therapies, or identify biomarkers that can be screened for as an indicator of preterm birth.”

Dr. Williams and Dr. Zhang, along with program director Dr. Louis Muglia and the research teams at Cincinnati Children’s and Case Western, will also use family-based whole exome analysis to identify variants in those families at risk for spontaneous preterm birth. Investigators at the University of Iowa, under the direction of Dr. Kelli Ryckman, Assistant Professor of Epidemiology and Pediatrics, are also involved in the project.

The overriding goal of Theme 2 is to identify candidate genes for further testing through collaboration with Theme 1 and Theme 3 investigators. Once strong genetic associations for preterm birth are determined, the information will be used to feed studies linking genetic indicators to environmental factors such as lifestyle, health influences and socioeconomic traits.

SIMILAR ADVANCES TO SICKLE CELL?

“Using genetics to prevent preterm birth may be a long-term goal,” Dr. Williams adds. “However, as an example, people with the gene that causes sickle cell anemia are living longer, in part due to an understanding of the genetic processes that
Moms and babies in the U.S. are facing an urgent health crisis:

- In this country 1 in 10 babies is born prematurely each year.
- Worldwide 15 million babies are born prematurely each year.
- Premature birth and its complications are the largest contributors to infant death in the United States and globally.
- More than 380,000 babies are born prematurely in the U.S. each year.
- In addition to the human toll, the societal cost of premature birth is more than $26 billion in the U.S. per year.
- Women of color are up to 50 percent more likely to give birth prematurely and their children can face a 130 percent higher infant death rate.
- In this country black women have maternal death rates over three times higher than women of other ethnicities.
- More than 20 percent of premature babies are born to black women—that’s 1 in 5 babies.
- Employers pay 12 times as much in health care costs for premature/low birthweight babies compared to babies born without these complications.

Because premature birth has many possible causes, each PRC is charged with exploring a different transdisciplinary research target that is likely to be crucial to the prevention of premature birth. To accomplish its goal the Ohio Collaborative has developed five interrelated theme areas, each bringing together renowned thinkers, researchers, physicians and top academics to focus on key aspects of the underlying causes of preterm birth.

I’m more hopeful now than I’ve been at any time in my life, says Dr. Muglia. We have tools we didn’t have just five to 10 years ago. It took decades to sequence the first genome, now we can do it in two days, providing an overwhelming amount of data. And with the rapid advances in computational power we now have the capability to analyze the data in ways we never could before.”

THEME 2 LEADERS

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For more information on how you can be a part of this effort please contact: 914.997.4492

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