Prenatal tests more informative using microarray technology, researchers find

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A new method for detecting abnormalities in unborn children is providing physicians with more information to analyze the results than conventional, microscopic testing, according to two George Washington University researchers.

Elizabeth Thom, research professor of epidemiology and biostatistics, and Julia Zachary, senior research scientist, are co-authors of the lead article appearing in the current issue of the New England Journal of Medicine showing that microarray technology provides a more comprehensive result from genetic testing during prenatal care of women than the current method of testing, called karyotyping, which relies on visual analysis of the fetal chromosomes.

"Microarray analysis of blood is now standard of care when children or adults present with undiagnosed neurodevelopmental problems since there are a number of serious syndromes that involve small changes on chromosomes which are not seen through a microscope," explained Dr. Thom. "It is a natural extension to want to identify these syndromes prenatally, but research into how to interpret the data is essential."

The GW Biostatistics Center in Rockville, Md. where Dr. Thom and Ms. Zachary are both based was the coordinating center for the multi-site trial. The research was funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development and led by Columbia University Medical Center.

The trial involved 4,400 patients at 29 centers nationwide and the data took more than four years to compile. The trial's cohort consisted of women over the age of 18 but predominantly over 30, whose fetuses were shown in early screenings to be at a heightened risk for Down syndrome or to have structural abnormalities found through an ultrasound.

The trial found that microarray analysis performed as well as karyotyping in identifying common outcomes involving an abnormal number of chromosomes, which can cause genetic or developmental disorders. It also identified additional abnormalities that were completely undetected by karyotyping.

"Particularly when a prenatal ultrasound has shown anomalies it becomes very important for parents to have an explanation. When the karyotype is normal, microarray analysis will provide additional information in 6 percent of cases. This diagnosis allows parents to plan for early intervention, especially in the case of autism spectrum and other neurodevelopmental disorders," said Ms. Zachary.

Use of microarray for analysis of prenatal samples has only been done in a few laboratories in the U.S. until recently, primarily because of high cost, the difficulty in developing protocols which achieve reliable success rates in DNA extraction from uncultured prenatal samples and the limited experience by genetic counselors and physicians in interpreting the results.

Dr. Thom is a research faculty member at the School of Public Health and Health Services. The Biostatistics Center is a research facility under the Office of the Vice President for Research and serves as a coordinating center for large scale multi-center clinical trials and epidemiologic studies. Its researchers participate in major medical research programs of national and international scope, frequently leading

to major medical advances.

Provided by George Washington University