Use of fetal genetic sequencing increases the detection rate of genetic findings

January 23 2017

In a study to be presented Thursday, Jan. 26, in the oral plenary session, at the Society for Maternal-Fetal Medicine's annual meeting, The Pregnancy Meeting, researchers with the Columbia University Medical Center in New York found that, in preliminary data, fetal genomic (whole exome) sequencing (WES) as a diagnostic test for women with pregnancies complicated by major fetal congenital anomalies increased the detection rate of genetic findings by between 10 to 30 percent.

The study, titled Whole exome sequencing in the evaluation of fetal structural anomalies: A prospective study of sequential patients used selected patients that were felt to have a high likelihood of having a fetal genetic anomaly.

In recent years, prenatal detection of fetal congenital anomalies has become increasingly more frequent, due to the adoption of routine ultrasound imaging. Simultaneously, advanced genetic testing has evolved demonstrating that an increasing proportion of these anomalies have a genetic cause. Approximately 10 years ago, chromosomal microarray analysis (CMA) was added to standard karyotyping as a prenatal diagnostic test increasing the detection rate of clinically significant cytogenetic abnormalities by 6% in cases with a single anomaly (abnormality) and 13% when multiple anomalies were present. In other words, CMA looked at cell and chromosomal disorders. These prior studies, including a multi-center National Institutes Child Health and Human Development (NICHD)-funded trial presented at a prior Society of Maternal-Fetal Medicine annual meeting, has changed

national guidelines so that CMA is now the recommended test for evaluating fetal anomalies.

While CMA has been a significant improvement, an estimated 60-70% of cases with identified fetal abnormalities still remain without a genetic diagnosis. With this current study, fetal genomic (whole exome) sequencing was evaluated as a diagnostic test for women with pregnancies complicated by major fetal congenital anomalies.

"Our preliminary data and published literature indicate that sequencing will increase the detection rate of genetic findings and this information will significantly improve patient counseling and neonatal treatment," explained Ronald Wapner, M.D., professor of obstetrics and gynecology for the maternal fetal medicine department at Columbia University Medical Center, who is presenting the study. "New associations with genes with very specific fetal phenotypes are also beginning to be uncovered," he added.

More information: Abstract #8: Whole exome sequencing in the evaluation of fetal structural anomalies: A prospective study of sequential patients, The Pregnancy Meeting, 2017.

Provided by Society for Maternal-Fetal Medicine