New Paths in Prenatal Care

Promising Non-Invasive Screening Technology

by Rhea Maze

Most parents today can remember one of the first parenting questions they faced: Should Mom undergo an amniocentesis to check for birth defects or not? For many, it was a tough question for two reasons. In addition to the ethical dilemma a positive result could create, the test, which involves injecting a long needle into Mom's belly and extracting amniotic fluid from baby's protective sac, carries slight risks, including a less-than-1-percent chance of miscarriage.

But a test being offered to some high-risk pregnant women today could relieve at least that piece of the worrisome puzzle, allowing parents to test for gender and three main chromosomal defects, including Down syndrome, through a simple blood test for Mom. Then, parents can make a more-informed decision on whether to proceed with the more invasive, but more definitive, tests, such as amniocentesis and chorionic villus sampling (which tests a piece of placental tissue rather than amniotic fluid).

The blood test, which became available in 2011 but is not yet FDA approved, requires only a blood draw from women who are at least 10 weeks pregnant. "The test performs sequencing of cell-free DNA from the mother's blood, looking for fetal DNA and analyzing it for fetal disorders," says Dr. Richard Porreco, director of maternal-fetal medicine at Rocky Mountain Hospital for Children at Presbyterian/St. Luke's Medical Center.

Studies of the test have demonstrated detection rates for the chromosomal abnormalities (trisomy 13, 18 and 21) of greater than 98 percent and false-positive rates of less than 0.5 percent. An autosomal trisomy occurs when a baby receives three copies of a chromosome instead of the normal two. Risk increases with maternal age. Trisomy 21 is Down syndrome, which can vary in severity. Trisomy 13 and 18 are more serious, resulting in severe retardation and other often-fatal birth defects. Women who screen positive with the new blood test are recommended to confirm the results with an amniocentesis or CVS test.

Porreco routinely offers the new test to his high-risk patients and, as a result, has seen the number of invasive procedures markedly diminish, he says. He predicts the test ultimately will become routine for both high- and low-risk women. "It adds information that can be very productive for parents, and it has changed our whole approach to prenatal diagnosis." ■

DID YOU KNOW?

During the first trimester, the most common cause of miscarriage is chromosomal abnormality.