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Prenatal Test Presents Dilemmas to Expectant Mothers

Knowledge isn't always power, some women say, study finds
(*this news item will not be available after 12/27/2012)
Friday, September 28, 2012

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FRIDAY, Sept. 28 (HealthDay News) -- A sophisticated genetic test sometimes used during pregnancy can't always predict if chromosomal abnormalities will cause problems in children, leading some mothers to label the information "toxic knowledge" they wish they hadn't received, a small new study shows.

Researchers from three universities found that expectant mothers receiving bad news about a genetic test called a DNA microarray -- more often used after birth to identify chromosomal problems in children with unexplained delays or defects -- reported mostly negative responses, ranging from feeling blindsided to needing support to digest the information and make critical decisions about their pregnancies.

The women's reactions challenge the notion that knowledge is power, especially when that knowledge pertains to ambiguous information about an unborn baby's health, said study author Barbara Bernhardt, a genetic counselor and clinical professor of medicine at the Hospital of the University of Pennsylvania.

"I think we need to have better information readily available to patients and providers . . . to make the decisions they need to make in a timely manner," Bernhardt said. "We also need additional education of obstetricians and midwives to feel more comfortable talking to patients about it and counseling them about the results."

The study was published online recently in the journal *Genetics in Medicine*.

Standard DNA testing offered to pregnant women uses tests such as amniocentesis and chorionic villus sampling, which involve "karyotyping" to identify common abnormalities such as Down syndrome. But DNA microarrays can detect smaller-scale chromosomal changes that can signal future problems such as autism or congenital disorders, although the test can't necessarily predict how severe the problem will be or even if a gene variant will produce any discernible conditions in the child.

Some of the 54 study participants, 23 of whom were interviewed at least six months after childbirth or pregnancy termination, had learned from ultrasound or other tests that their fetus had abnormalities. But for those whose prior tests had come back "normal," learning their baby had genetic variants of unknown significance sent some of them into a tailspin.

Bernhardt's team identified five key responses that described the women's experiences, including considering the microarray results toxic knowledge they wish they hadn't learned. Many of the women accepted the testing -- which costs between $1,500 and $3,000 -- because it was offered at no charge to them, which they felt was "an offer too good to pass up."

Unlike children who are tested using DNA microarrays to help diagnose existing problems, children tested prenatally may never develop health or developmental issues arising from their genetic variants. "We discovered in the course of the study that some of the parents have the same [chromosomal] deletion or duplication as their fetus" but never suffered any ill effects, said Bernhardt, also co-director of the Penn Center for the Integration of Genetic Healthcare Technologies.

However, "the horse is out of the barn," she added. "This test is there, it's available, which is why we have to provide good information and careful counseling."

Sandra Darilek, a genetic counselor and clinical instructor at Baylor College of Medicine in Houston, said the vast majority of results presented by microarray testing are straightforward, but every genetic condition presents differently, from mild to severe.

"In a lot of cases, it's an incredibly useful test because if they get a normal result, it can be very reassuring for a patient," Darilek said. "But it also depends on the point of view the patient is coming from. Some want all the information possible and are willing to take uncertainty, and some want straightforward information."

Because technology is continually advancing, ambiguity will always be part of newer medical tests, Darilek noted.

"I don't know that uncertainty will ever be completely absent from prenatal testing at all until we have a handle on our complete genome," she added. "But with the platform of testing we currently have, if it would stay static, within a few years we'll have an even better grasp on some of these [genetic] variants that are new to us now. But it's going to be an ever-evolving situation."

SOURCES: Barbara Bernhardt, M.S., genetic counselor, Hospital of the University of Pennsylvania, Philadelphia, and co-director, Penn Center for the Integration of Genetic Healthcare Technologies; Sandra Darilek, M.S., genetic counselor and clinical instructor, Baylor College of Medicine, Houston; Sept. 6, 2012, *Genetics in Medicine*, online

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