Effect of intervention, removal of costs, on prenatal genetic testing

September 23 2014

An intervention for pregnant women that included a computerized, interactive decision-support guide regarding prenatal genetic testing, and no cost for testing, resulted in less prenatal test use and more informed choices, according to a study in the September 24 issue of *JAMA*.

Since the introduction of amniocentesis, prenatal genetic testing guidelines have focused on identifying women at increased risk of giving birth to an infant with Down syndrome or other chromosomal abnormalities, for whom invasive diagnostic testing should be recommended. Prenatal genetic testing guidelines recommend providing patients with detailed information to allow informed, preference-based screening and diagnostic testing decisions. The effect of implementing these guidelines is not well understood, according to background information in the article.

Miriam Kuppermann, Ph.D., M.P.H., of the University of California, San Francisco, and colleagues randomly assigned 710 pregnant women to receive either a computerized, interactive decision-support guide and access to prenatal testing with no out-of-pocket expense (n = 357) or usual care as per current guidelines (n = 353). The trial was conducted from 2010-2013 at prenatal clinics and practices from throughout the San Francisco Bay area.

The researchers found that significantly fewer women who were assigned to the intervention group underwent invasive diagnostic testing compared with women randomized to the control group (5.9 percent vs

12.3 percent). The overall prenatal testing strategy used by the 2 groups also differed: women randomized to the intervention group were more likely to have no testing (25.6 percent vs 20.4 percent) or screening alone (68.5 percent vs 67.3 percent).

Also, women assigned to the intervention group had significantly higher genetic testing knowledge scores, were more likely to correctly report both the miscarriage risk of amniocentesis and their likelihood of carrying a fetus with the congenital disorder trisomy 21.

"This study's finding that women who were randomized to the intervention group were less likely to undergo testing than those who received usual care adds support to the contention that women may not be receiving adequate counseling about their options. This underscores the need for clinicians to be clear that prenatal testing is not appropriate for everyone, and to present forgoing testing as a reasonable choice," the authors write.

"If validated in additional populations, this approach may result in more informed and preference-based prenatal testing decision making and fewer women undergoing testing."

In an accompanying editorial, Siobhan M. Dolan, M.D., M.P.H., of the Albert Einstein College of Medicine/Montefiore Medical Center, Bronx, New York, writes that the finding from this study that women with better understanding of the information about various prenatal testing options were less likely to undertake invasive prenatal testing is important, and it contradicts the notion that more information is always desired.

"It is possible that the nature of prenatal testing is different than other health care decisions, but the public may be increasingly aware that the numerous medical advances of the last decade have also created greater

complexity in decision making. This finding also suggests that prenatal genetic testing decisions require a complex calculus that considers the timing of the testing, the certainty of the results, and the risks related to undergoing invasive genetic testing during pregnancy."

More information: DOI: 10.1001/jama.2014.11479
DOI: 10.1001/jama.2014.12205