

Research supports expanded use of cell free DNA prenatal testing

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A DNA-based prenatal blood test used to screen pregnancies for Down syndrome and similar chromosome abnormalities in high-risk women has moved a step closer to use in the general pregnancy population. Researchers at Women & Infants Hospital of Rhode Island, a Care New England hospital, have published a study in *Genetics in Medicine* that shows that this non-invasive test can be effectively and appropriately offered to all pregnant women, regardless of maternal age or risk factors, through primary obstetrical care providers.

The research, "The clinical utility of DNA-based screening for fetal aneuploidy by primary obstetrical care providers in the general pregnancy population," was led by Glenn Palomaki, PhD, Edward M. Kloza, MS, CGC, Elizabeth Eklund, MS, and Geralyn Messerlian, PhD, of the Division of Medical Screening and Special Testing in the Department of Pathology and Laboratory Medicine at Women & Infants Hospital of Rhode Island and The Warren Alpert Medical School of Brown University, as well as maternal-fetal medicine specialist Barbara M. O'Brien, MD, formerly of Women & Infants Hospital. This independent study was funded by a grant from Natera, Inc. (San Carlos, CA) and the DNAFirst test was primarily based on Natera's Panorama offering.

"We already know that DNA-based screening is highly effective. This study enabled us to look at its implementation in the general population to determine how best to educate professionals and patients," said Dr. Palomaki.

The study aimed at determining the knowledge and satisfaction of women who chose the DNAFirst screening test as part of routine prenatal care. Of the approximately 2,700 women in Rhode Island who chose DNAFirst screening, a subset with specific test and demographic characteristics was contacted. These women participated in a 15 minute structured telephone interview about their experience.

"We developed patient education materials and trained the providers on speaking with their patients about the DNAFirst test. The providers and patients were then surveyed concerning their knowledge about the test, how they made their decision about the [test](#), and their overall satisfaction," explained Dr. Palomaki. "Ultimately, we found that the materials were highly effective for both the providers and the patients."

Women & Infants Hospital has been an international center for prenatal screening research. For more than three decades, under the leadership of the late Jacob Canick, PhD, the faculty in the Division of Medical Screening and Special Testing has led research to develop and improve screening tests for Down syndrome and other fetal abnormalities. In 2011, Dr. Palomaki and colleagues published the first external validation study of next generation sequencing of circulating cell free DNA in maternal plasma to identify common chromosome abnormalities.

"The current study results will be utilized by policy-makers, professional organizations and insurance [providers](#) when deciding how and to whom DNA-based prenatal [screening](#) will be offered," said Dr. Palomaki.

Provided by Care New England

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