

Study shows that new DNA test to identify Down syndrome in pregnancy is ready for clinical use

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A new DNA-based prenatal blood test that can strikingly reduce the number of risky diagnostic procedures needed to identify a pregnancy with Down syndrome is ready to be introduced into clinical practice. The test can be offered as early as 10 weeks of pregnancy to women who have been identified as being at high risk for Down syndrome. These are the results of an international, multicenter study published online today in the journal *Genetics in Medicine*. The study, the largest and most comprehensive done to date, examined almost 1,700 pregnancies at high risk of chromosomal abnormalities, 212 of which were affected by Down syndrome.

The research was led by Jacob Canick, PhD, and Glenn Palomaki, PhD, of the Division of Medical [Screening](#) and Special Testing in the Department of [Pathology](#) and Laboratory Medicine at Women & Infants Hospital and The Warren Alpert Medical School of Brown University, and included scientists at Sequenom, Inc. and Sequenom Center for Molecular Medicine, San Diego, CA, and an independent academic laboratory at the University of California at Los Angeles.

The [test](#) identified 98.6% of the [Down syndrome](#) pregnancies, while only 0.2% of the normal pregnancies were mistakenly called positive. The test rarely failed to provide a clinical interpretation (0.8%). These findings, along with the detailed information learned from testing such a large number of samples, demonstrate that the new test will be highly

effective when offered to women considering invasive testing.

"With current screening methods, about one in every 30 women offered a follow-up invasive diagnostic procedure - amniocentesis or chorionic villus sampling (CVS) - will be found to have a [pregnancy](#) with Down syndrome. We expect the DNA-based test to more accurately determine which women should be offered invasive diagnostic testing. As a result, most of the pregnancies referred for amniocentesis or CVS will be found to have Down syndrome," said Dr. Canick.

Dr. Palomaki added, "If this new test is used as we've described, nearly all women with a normal pregnancy could avoid an invasive diagnostic procedure and its associated anxiety, cost, and potential for fetal loss."

Down syndrome, also called trisomy 21, is a chromosomal disorder that includes mental retardation, characteristic facial features, and, often, heart defects, and affects one in 550 babies born each year in the US. Down syndrome occurs when each cell in an individual has three rather than the usual two copies of chromosome number 21. Current prenatal [screening tests](#) for Down syndrome combine maternal age with information from the measurement of maternal serum markers and ultrasound markers in the first and second trimesters of pregnancy. While these tests can detect up to 90% of Down syndrome cases, they also incorrectly identify 2% to 5% of normal pregnancies as positive. The new DNA-based test will reduce this "false positive" rate while maintaining the detection rate.

"Prenatal screening and diagnosis of Down syndrome has been part of routine prenatal care for decades, and it is estimated that nearly two-thirds of all pregnant women in the US are currently screened," said Dr. Canick. "It is possible that with the availability of this new DNA-based test, more women will opt for screening because of the increased safety resulting from far fewer amniocentesis and CVS procedures being

performed." The US Centers for Disease Control and Prevention estimated in 1995 that about one in every 200 invasive [diagnostic procedures](#) will cause a pregnancy miscarriage.

This industry-sponsored project, awarded to Drs. Canick and Palomaki and Women & Infants Hospital in 2008, enrolled 4,500 women at 27 prenatal diagnostic centers throughout the world. Women & Infants also served as one of the enrollment centers under the direction of maternal-fetal medicine specialist and director of Perinatal Genetics, Barbara O'Brien, MD.

"Screening tests, by their nature, do not diagnose, but rather offer information about the chances that a pregnancy may be affected by a [genetic](#) abnormality. For years we have relied on screening tests that have had a fairly significant false positive rate because that was the best screening available," said Dr. O'Brien. "But having access to a DNA-based test that can be done early in pregnancy will give us more information so that we can better guide which patients should consider diagnostic testing."

Women & Infants Hospital has been an international center for prenatal screening research. For more than three decades, Drs. Canick and Palomaki have collaborated with others in developing and improving screening tests for Down syndrome and other fetal abnormalities. In 1988, Drs. Canick and Palomaki were involved in the development of triple marker screening. The team was able to convert its findings into prenatal screening tests now used throughout the world. Dr. Canick's lab in 1998 was the first in the US to offer quad marker screening and in the past decade was the laboratory center for the National Institute of Health (NIH) funded FASTER Trial which compared first and second trimester screening.

It was announced today that one version of this laboratory-developed

test, MaterniT21, has been validated through clinical studies and is now available through the Sequenom Center for Molecular Medicine, a CLIA-certified and CAP-accredited laboratory. Harry F. Hixson Jr., PhD, chairman and CEO of Sequenom, Inc., said, "We have been fortunate to partner in the clinical study and are proud to offer the service to assist specialists and [high-risk](#) patients in making more informed decisions about their pregnancy."

Provided by Women & Infants Hospital

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