New York State Approves Quest Diagnostics' Fragile X Syndrome Test

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XSense(R) is the first laboratory test that may be suitable for population-based screening for Fragile X Syndrome, the leading inherited cause of mental retardation

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An accurate, faster testing option to identify female carriers and other patients with genetic abnormalities that cause Fragile X Syndrome is now available to physicians in all fifty states with the recent approval in New York. Fragile X is the leading cause of inherited mental retardation and the most common known single gene cause of autism.

XSense(R), Fragile X with Reflex, from Quest Diagnostics Incorporated (NYSE: DGX), the world's leading diagnostic company, has been approved by New York State's Department of Health. XSense is the first test for Fragile X Syndrome to be approved by New York to employ a new laboratory analysis technique that bypasses the need to perform the Southern Blot DNA analysis method in 99 percent of cases. The use of Southern Blot, which can take several days to weeks to perform, has hampered the lab industry's ability to widely provide Fragile X testing. XSense results are reported in about a week for the vast majority of patients.

With the approval, Quest Diagnostics can offer the test to physicians in New York as well as in all other U.S. states. New York is the only U.S. state with an independent regulatory review process for laboratory developed tests, which are also regulated at the federal level.

"New York's approval is significant because it means a new, highly innovative genetic analysis technique for Fragile X has fulfilled state-required quality standards that are widely regarded in the lab industry as highly rigorous," said Charles Strom, M.D., Ph.D., medical director, Genetic Testing Center, Quest Diagnostics Nichols Institute. "Fragile X can be a devastating diagnosis, given the severe disability it causes many patients. While it is highly prevalent, Fragile X is not widely tested for, due in part to technical limitations with conventional tests that our XSense technique largely surmounts."

Physicians can use XSense to aid their identification of women who, as carriers, may be unaffected or slightly affected by Fragile X syndrome, but are at risk of passing it to offspring, regardless of the father's genetics. It may also aid in the diagnosis of male and female patients using blood and other specimen types. An estimated one in 260 women are genetic carriers of Fragile X Syndrome, according to the National Fragile X Foundation, although recent research by scientists at Quest Diagnostics and other organizations suggest carrier prevalence may be even higher.

"XSense is a step forward to the day when the medical community can seriously consider the option to provide population-wide quality testing for Fragile X in much the same way we now offer population screening for other hereditary disorders," said Dr. Strom.

A couple is statistically more likely to pass Fragile X to their offspring than two common hereditary disorders, cystic fibrosis or Tay-Sachs Disease, which guidelines support for wide population screening. In most states, newborns are routinely tested for cystic fibrosis. While medical guidelines recommend Fragile X testing for some patients, such as women seeking reproductive counseling with a personal or family history of mental retardation, they do not support population-based carrier or newborn testing, in part due to technical lab-testing hurdles.

A study performed and funded by Quest Diagnostics and published in the March 2010 issue of *Genetics in Medicine*, the official publication of the American College of Medical Genetics (ACMG), found that the XSense technique showed 100% agreement with the standard widely used lab-testing method, which requires a DNA analysis technique called Southern Blot in about 20 percent of cases. Southern Blot can take several days to weeks to perform, making it generally unsuitable for high-volume testing applications. In the study, the XSense technique bypassed the need for Southern Blot in more than 99 percent of cases.

The investigators concluded that the XSense test is highly accurate, and may be suitable for high-volume population screening and diagnostic testing on a range of patients, including women and newborns.

Earlier this month, *Genetics in Medicine* published a report that found there is adequate research data to support population screening of women of childbearing age for Fragile X syndrome. The report was based on a review and analysis of eleven prior studies of Fragile X screening in women of reproductive age.

About XSense(R), Fragile X with Reflex

XSense, Fragile X with Reflex, test identifies abnormalities of the fragile X mental retardation 1 (FMR1) gene residing on the X chromosome. The number of times a certain pattern of DNA, called CGG, occurs determines whether a person has a premutation and is a carrier or has a full mutation and has the disorder. XSense employs a technique called triplet-primed polymerase chain reaction by capillary electrolysis (triplet-primed PCR-CE) to assess the number of CGG repeats without Southern Blot in about 99 percent of cases. Quest Diagnostics is the first U.S. company to publish peer reviewed data on this new Fragile X syndrome test technique and bring it to market. Quest Diagnostics offers XSense in alignment with current guidelines for Fragile X Syndrome testing.

About Quest Diagnostics

Quest Diagnostics is the world's leading provider of diagnostic testing, information and services that patients and doctors need to make better healthcare decisions. The company offers the broadest access to diagnostic testing services through its network of laboratories and patient service centers, and provides interpretive consultation through its extensive medical and scientific staff. Quest Diagnostics is a pioneer in developing innovative diagnostic tests and advanced healthcare information technology solutions that help improve patient care. Additional company information is available at www.QuestDiagnostics.com.

About the New York State Department of Health Clinical Laboratory Evaluation Program

New York approves laboratory-developed tests that are not FDA cleared before allowing them to be performed on patients in the state. In order to gain approval, labs must validate that a test performs as it is intended, based on validation data collected according to the U.S. Center for Disease Control and Prevention's Clinical Laboratory Improvement Amendments and New York State requirements. New York is the only state that independently approves laboratory-developed tests.

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