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Contact: Kathy Beal, MBA
ACMG Media Relations Director
301-238-4582
Mobile 978-853-1810
kbeal@acmg.net

New Test May Allow Screening for Fragile X Syndrome, Reports Genetics in Medicine Simple, Rapid Test May Be Suitable for Newborn or Family Screening

A newly developed test appears highly accurate in identifying newborns with fragile X syndrome—the most common inherited cause of cognitive impairment—as well as identifying couples who are carriers of the causative gene, reports a study in the March issue of Genetics in Medicine, the official peer-reviewed journal of The American College of Medical Genetics. The journal is published by Lippincott Williams & Wilkins, a part of Wolters Kluwer Health, a leading provider of information and business intelligence for students, professionals, and institutions in medicine, nursing, allied health, and pharmacy.

The new test "overcomes the technical block to institute programs of newborn screening or population-based carrier detection for fragile X syndrome," according to the study, led by Feras M. Hantash, M.S., Ph.D., of Quest Diagnostics, San Juan Capistrano, Calif.

Quick Test May Overcome Barriers to Fragile X Screening

Fragile X syndrome, which causes intellectual disability and other abnormalities, affects about 1 in 4,000 males in the United States (Fragile X syndrome also occurs in females, but causes less severe impairment). Fragile X syndrome is caused by mutations of a gene called FMR1. The mutations are relatively common in the population—although estimates vary, 1 in 300 to 400 U.S. couples may be carriers of the abnormal genes.

Currently available tests are too expensive and time-consuming for use in routine screening of couples and newborns. As a result, testing is generally done only when there's some reason to suspect the family is at high risk, such as a previous child with fragile X syndrome.

To overcome these obstacles, the researchers developed a new test for mutations of the FMR1 gene. Using polymerase chain reaction (PCR) technology to detect abnormalities called "CGG repeats," the test detects not only full disease-causing mutations, but also more mild gene expansions called premutations.

In samples with previously identified FMR1 mutations, the new PCR test showed a distinct "stutter" pattern whenever a full or partial mutation was present. It was also capable of detecting FMR1 "mosaics," which can be difficult to detect with standard approaches. The test was further evaluated in a series of 1,275 blood samples submitted for fragile X testing. The results showed a 100 percent rate of agreement between the new test and the standard "Southern blot" test. Mutations were found in 6 of the 1,275 patients tested.

With further study, the new PCR test may open the way to more widespread testing for fragile X mutations. This could include routine screening of newborns, as the test can detect FMR1 mutations in standard newborn blood spots. Abnormal screening results would require further diagnostic evaluation, including the standard Southern blot test. The researchers estimate that the screening test could reduce the number of Southern blot tests performed by 99 percent, thus saving considerable time and money.

More research will be needed before the ACMG and other organizations issue official recommendations on prenatal screening. In addition to medical and scientific questions, studies will need to address the ethical and legal issues involved with screening for fragile X syndrome. "It certainly will be a challenge to educate the public about the complexities of fragile X testing," Dr. Hantash and colleagues write.

About the American College of Medical Genetics

Founded in 1991, the American College of Medical Genetics (www.acmg.net) advances the practice of medical genetics by providing education, resources and a voice for more than 1400 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals committed to the practice of medical genetics. ACMG's activities include the development of laboratory and practice standards and guidelines, advocating for quality genetic services in healthcare and in public health, and promoting the development of methods to diagnose, treat and prevent genetic disease. Genetics in Medicine, published monthly, is the official ACMG peer-reviewed journal. ACMG's website (www.acmg.net) offers a variety of resources including Policy Statements, Practice Guidelines variety of resources including Policy Statements, Practice Guidelines, Educational Resources, and a Medical Geneticist Locator. The educational and public health programs of the American College of Medical Genetics are dependent upon charitable gifts from corporations, foundations, and individuals. The American College of Medical Genetics Foundation (www.acmgfoundation.org) is a 501(c)(3) not-for-profit organization dedicated to funding the College's diverse efforts to translate genes into health. The Foundation is dedicated to Better Health Through GeneticsTM.

About the Rare Disease Foundation

The Rare Disease Foundation is an organization comprised of parents, clinicians and researchers working together to find solutions for children and families affected by rare disease. Through facilitating communications between expert researchers, the Rare Disease Foundation performs treatment-focused research into rare diseases. The Foundation also provides research grants to directly improve patient care. With parents and their families, we advocate for and foster the organization of the rare disease community and organize families for mutual support regardless of diagnosis.

7101 Wisconsin Avenue, Suite 1101 | Bethesda, MD 20814
Telephone: 301-718-9603 | Fax: 301-718-9604 | E-mail:acmg@acmg.net | Website: www.acmg.net
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