Quest Diagnostics Introduces Specialized Cystic Fibrosis Test for CFTR Gene Mutations

Quest Diagnostics has added a new genetic test to its comprehensive menu of tests for cystic fibrosis (CF). The new test detects deletions and duplications within the CF transmembrane regulator (*CFTR*) gene and is designed to help physicians identify genetic abnormalities in patients at high risk for cystic fibrosis. Developed at Quest Diagnostics Nichols Institute in San Juan Capistrano, California, the *CFTR* Gene Deletion or Duplication test broadens Quest Diagnostics' leading position in genetic testing.

The *CFTR* Gene Deletion or Duplication test provides additional information beyond that provided by traditional CF screening tests to assist physicians in diagnosing, counseling and treating individuals with a family history or clinical symptoms of CF. This assay detects primarily large deletions or insertions in the *CFTR* gene, which are generally not detected by common screening panels or extensive sequencing of the *CFTR* gene. Recent studies suggest that such rearrangements may account for 16% to 24% of mutant *CFTR* genes not identified after extensive sequencing (Audrazet et al., 2004; Chevalier-Porst et al., 2005). It can be used to diagnose CF pre- or postnatally and to diagnose atypical CF in individuals with other conditions associated with the *CFTR* gene. This test can be especially helpful for individuals with symptoms of classical or atypical CF who have fewer than 2 CFTR mutations identified via standard mutation screening. Ordering physicians have access to consultation with genetic counselors and medical staff toll-free at 866-GENE INFO (1-866-436-3463).

According to the Cystic Fibrosis Foundation, CF is a genetic disease affecting approximately 30,000 children and adults in the United States. More than 10 million Americans are carriers of a defective *CFTR* allele. Quest Diagnostics offers a carrier-screening test for CF that analyzes a patient's blood sample for all 23 mutations included in the 2004 revised guidelines from the American College of Medical Genetics. In addition, Quest Diagnostics offers the CF Complete™ test, which sequences the complete coding region of the *CFTR* gene. The new *CFTR* Gene Deletion or Duplication Test along with the CF Complete test now enables physicians to identify more rare mutations and offer carrier detection and diagnosis for both nuclear and extended family members.

About Quest Diagnostics

Quest Diagnostics is the leading provider of diagnostic testing, information and services that patients and doctors need to make better health care decisions. The company offers the broadest access to diagnostic testing services through its national 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 new diagnostic tests and advanced health care information technology solutions that help improve patient care. Additional company information is available at: **www.questdiagnostics.com**.

The "New Products" page is designed to offer you news and information from businesses serving the genetics community. We welcome you submissions. All submissions are subject to review by the Editor. For more information, contact Al Lucchesi, National Accounts Manager, Lippincott Williams & Wilkins, 530 Walnut Street, Philadelphia, PA 19106; phone 215-521-8409; fax 215-521-8411; email alucches@lww.com