

Genzyme Genetics Cystic Fibrosis Mutation Analysis Now Tests for 97 mutations

(September 14, 2005) – Genzyme Genetics, a business unit of Genzyme Corporation, announced that it has upgraded its CFplus™ cystic fibrosis (CF) mutation analysis to include testing for 97 mutations, while providing an industry-leading turnaround time. With the additional mutations, CFplus continues to be one of the most comprehensive and rapid cystic fibrosis screening tests for the ethnically diverse U.S. population. Genzyme’s CFplus mutation analysis provides 21% higher detection for Hispanics and 9% higher detection for African Americans over the standard 23 mutation panel, with detection rates of 78% and 81%, respectively. The inclusion of a new mutation, CFTRdele2,3 (21kb), is expected to improve CF detection for individuals of central and eastern European descent. These upgrades follow last year’s launch of CFplus for mouthwash samples, which made Genzyme Genetics the only national laboratory to offer CF testing for this convenient, noninvasive sample type.

“Our new cystic fibrosis test exemplifies our commitment to continued innovation and leadership in CF screening,” says Mara Aspinall, president, Genzyme Genetics. “We are pleased to offer a cystic fibrosis carrier screening test that not only tests for 97 mutations with the highest detection rates for

Caucasians, African Americans and Hispanics, but also gets results to patients faster.”

Cystic fibrosis (CF) is a disorder of mucus production primarily affecting the pulmonary, gastrointestinal and reproductive systems. Most individuals with CF require lifelong medical care and experience reduced life expectancy.

CFplus determines if a couple is at increased risk for giving birth to a child who will have cystic fibrosis. If both parents are found to be carriers, tests can also be performed during pregnancy to determine if the baby has cystic fibrosis. According to the American College of Obstetricians and Gynecologists (ACOG) Clinical and Laboratory Guidelines, “Cystic fibrosis carrier screening should be offered to couples with a family history of CF, partners of individuals with CF, and Caucasian couples of European or Ashkenazi Jewish descent planning a pregnancy or seeking prenatal care.” Additionally, according to ACOG guidelines, “...information about CF screening should be provided to patients in other ethnic and racial groups.”

“Because the U.S. has an ethnically diverse population, a pan-ethnic mutation panel is optimal to screen for CF in the U.S.,” says Stirling Puck, MD, national medical director, Genzyme Genetics. “That’s why we continually evaluate our cystic fibrosis mutation panel to ensure that it includes the most clinically relevant mutations and detection rates.”

Additional testing information

Genzyme Genetics offers several innovative options for collecting patient samples for CF testing. CFplus can be performed on whole blood and prenatal samples, as well as on noninvasive samples such as mouthwash, cheek brush and blood spot, eliminating the need for phlebotomy services and the discomfort of venipuncture for the patient. Genzyme Genetics is the only national laboratory to offer CF testing for mouthwash samples.

Additionally, to facilitate informed decision-making regarding genetic testing options, Genzyme Genetics provides patient education materials, as

well as the largest nationwide network of board-certified genetic counselors to talk with patients about their genetic risks and concerns.

In addition to an expanded CF carrier screening program, Genzyme Genetics offers a wide array of other molecular screening and testing options; an industry-leading cytogenetics program; and an extensive maternal serum screening program (which includes first trimester, integrated and second trimester screening).

About Genzyme Genetics

Genzyme Genetics is a leading, nationwide provider of high-quality, complex diagnostic testing services for physicians and their patients. With laboratories and counseling facilities located across the U.S., Genzyme Genetics offers extensive reproductive and cancer testing services, supported by innovative technology and a commitment to quality service and trusted information. Genzyme Genetics is a business unit of Genzyme Corporation.

Tel: 800 357-5744

Fax: (508) 389-5577