BCM
Baylor College of Medicine

MGL
MEDICAL GENETICS LABORATORIES

Date: 4/2012

To: Valued Clients

From: BCM Medical Genetics Laboratories

RE: CHANGE IN CYSTIC FIBROSIS TEST METHODOLOGY

What has changed

Effective May 1st, 2012, the **CFTR-Related Disorders Mutation Panel** (Test Code 6014) will be replaced by a new panel with an increased amount of mutations detected. The new mutation panel (Test code 7600) will test for the presence of 89, instead of 49, CFTR pathogenic mutations, including the 23 common mutations recommended by American College of Medical Genetics (ACMG). There will be **NO price change** associated with this added value**.** The new methodology has been approved for testing by the New York State Department of Health. Please use the updated requisition (as attached) to submit your future test requests.

Descriptions of New Methodology

Genomic DNA from test samples are amplified and hybridized to both normal and mutant fluorescent probes targeting the 89 mutation regions. Results are analyzed using quantitative imaging methods and analytical software to assist in determining if a given mutation is detected in the test sample. Reflex testing: If the R117H mutation is detected, analysis of the polythymidine variations (5T, 7T and 9T) at the intron 8 branch/acceptor site of the CFTR gene is performed. If the 5T variant is detected, then reflexive analysis of the adjacent polyTG tract is also performed.

We thank you for your business and look forward to continue providing you with the highest quality genetic testing services.

Sincerely,

Sean Y. Kim
General Manager
Medical Genetics Laboratories
Baylor College of Medicine