BCM
Baylor College of Medicine

MgL
MEDICAL GENETICS LABORATORIES

Date: 09/2013
To: Valued Clients
From: BCM Medical Genetics Laboratories
RE: Advanced mtDNA Point Mutations and Deletions by Massively Parallel Sequencing (BCM-MitomeNGS^{SM})

Effective January 3, 2014, the Medical Genetics Laboratories at Baylor College of Medicine will supplant test codes 3000, 3005, and 3006 with test code 2010.

The discontinued test codes are:

- Test Code 3000 - Mitochondrial DNA Common Mutations and Deletions Screen
- Test Code 3005 - Mitochondrial DNA Point Mutation Quantification
- Test Code 3006 – Reflex test for positive point mutations detected by test code 3000

Test Code 2010 - Advanced mtDNA Point Mutations and Deletions by Massively Parallel Sequencing (BCM-MitomeNGS^{SM}) is superior to the discontinued tests in many respects. The new features of the Advanced mtDNA Point Mutations and Deletions by Massively Parallel Sequencing (BCM-MitomeNGS^{SM}) are:

- Targets 36 common Point mtDNA mutations vs. 12 targeted mutations in the discontinued test code 3000.
- Simultaneous detection of Point mutations and their heteroplasmy levels.
- NGS technology provides more robust and accurate heteroplasmy measurement than the ARMS qPCR methodology utilized by the discontinued test codes 3005 and 3006.
- Accurate detection of heteroplasmies as low as 1.5%.
- Detection of single and multiple mtDNA deletions with much higher sensitivity than Southern blot analysis.
- Simultaneous detection of mtDNA deletions and breakpoints. Unlike Southern analysis, both the exact size and breakpoints of single deletions are obtained and reported.

Test comparison

Test code	2010	3000	3005/3006
Methodology	NGS	ASO and Southern Blot	ARMS qPCR
Number of mutations tested	36	12	1
Heteroplasmy quantification	Best	None	Good
Deletion detection	Best	Good	None
Report deletion breakpoint	Yes	No	No

Please direct all questions regarding this change to client services at 713-798-6555 or email us at genetictest@bcm.edu. Please visit our website for any additional notices or postings.

Thank you for your valued business. We look forward to providing you with the highest quality genetic testing services.

Sincerely,

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