

Date: December 18, 2014

To: Valued Clients

From: Baylor College of Medicine – Whole Genome Laboratory

RE: Testing Memorandum

Effective December 20, 2014, the whole exome sequencing tests offered in our laboratory (test codes listed below) will no longer report variants in the mitochondrial genome (mtDNA). This decision was made due to the low positive mtDNA diagnostic rate in the patients we have tested. In our recent publication (Yang et al. JAMA. 2014 Nov 12;312(18):1870) describing 2000 consecutive patients referred to our laboratory for clinical exome sequencing, there was an overall diagnostic rate of 25%. However, there was only one patient who was positive for a pathogenic mutation in the mitochondrial genome.

The following test codes will no longer have the mitochondrial genome variant screening:

- **Test code 1500-Whole Exome Sequencing**
- **Test code 1530-Whole Exome Sequencing Plus CMA**
- **Test code 1595-Cancer Exome Sequencing-Constitutional Exome Interpretation**
- **Test code 1600-Trio Whole Exome Sequencing**
- **Test code 1605-Adult Screening Exome Sequencing**

If you would like to order mtDNA analysis for your patients, the following tests are available from the Medical Genetics Laboratories:

- **Test code 2055-Comprehensive mtDNA Analysis by Massively Parallel Sequencing (MitoNGSSM)**
- **Test code 2010-Advanced mtDNA Point Mutations and Deletions by Massively Parallel Sequencing (BCM-MitomeNGSSM)**

Please direct any billing questions to 713-798-3566 or email us at medgenbilling@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