

TO: Clients of URMCLabs

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RE: New Test: Detection of the Mutations in the Epidermal Growth Factor Receptor (EGFR) Gene

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The Molecular Diagnostics Laboratory is now approved by the New York State Department of Health to offer a test to detect mutations in exons 18-21 of the EGFR gene in non-small-cell lung cancer (NSCLC).

Particular mutations in the EGFR gene, L858R and small in-frame deletions in exon 19, are associated with a good response to the orally available EGFR inhibitors gefitinib (Iressa™) and erlotinib (Tarceva™). Another mutation, T790M, is associated with a lack of response to these drugs, and accounts for about half of the cases of acquired resistance, although it is occasionally seen prior to treatment.

The purpose of the new test is to aid in the selection of the optimal therapy for patients with NSCLC. The standard test will detect mutations only in exons 19 and 21, the locations of the mutations most clearly associated with response to the currently approved drugs. Testing of exons 18 and/or 20 is available if needed.

The results of the EGFR mutation test will be reported as presence or absence of mutation. If a mutation is present then it will be identified. This test is done on formalin-fixed paraffin-embedded tissue blocks or on fresh tissue from NSCLC. Prior to DNA preparation and analysis, the specimen will be evaluated by a surgical pathologist to ascertain that a sufficient fraction of the cells are malignant. We intend a turn-around-time of about two weeks.

If you have any questions about this new test, please contact Dr. Paul Rothberg.