

TO: Clients of URMCLabs

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RE: New Test: Detection of the BRAF V600E Mutation

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As of June 28th, 2010, the Molecular Diagnostics Laboratory is now approved by the New York State Department of Health to offer a test to detect the V600E mutation in the BRAF gene.

This mutation is found in melanoma, papillary thyroid carcinoma, adenocarcinoma of the colon and rectum, and occasionally in other cancers.

The immediate purpose of the new test is to aid in the selection of the optimal therapy for patients with colorectal cancer. Colorectal cancer patients whose tumors have a BRAF V600E or a KRAS mutation do not respond to cetuximab or panitumumab, monoclonal antibodies to the epidermal growth factor receptor. The goal is to avoid the expense and possible toxicity of giving this therapy to patients who will not benefit from it. In this situation the BRAF mutation test will be offered as a reflex after a normal KRAS mutation test.

The new test may be useful in other systems for a variety of purposes including predicting prognosis in patients with papillary thyroid carcinoma and selecting patients for drugs which target BRAF in melanoma and other malignancies.

The results of the BRAF mutation test will be reported as presence or absence of the V600E mutation. This test is done on formalin-fixed paraffin-embedded tissue blocks or on fresh tissue. 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.