

BRAF V600 Mutation Testing

Test Code: BRAF

Use: Evaluation of tumor tissue for BRAF V600 mutations (including V600E, V600K, V600D, and V600R) is useful to predict response to BRAF-inhibitor therapy and is also useful in the workup of possible Lynch Syndrome.

Clinical Significance: *BRAF* mutations have been described in numerous neoplasms including melanomas, colorectal carcinomas, non-small cell lung carcinomas, and hematopoietic neoplasms (e.g. hairy cell leukemia). Tumors positive for mutations in *BRAF* codon 600 may respond to targeted therapy with BRAF-inhibitors. In a colorectal carcinoma with microsatellite instability (MSI), detection of a *BRAF* mutation indicates that the tumor is likely sporadic and not associated with Lynch Syndrome.

Results of the test must be considered in correlation with clinical information and other pathologic data. Results of this test alone should not be used to diagnosis malignancy. The test is not designed for minimal residual disease detection.

Methodology: Tissue sections are reviewed and assessed for tumor viability and cellularity. Following microdissection of tumor tissue, DNA is extracted for testing. Allele specific real time PCR is then used to test for mutations of codon 600 of the *BRAF* gene. The test is designed to detect p.Val600Glu (V600E), p.Val600Lys (V600K), p.Val600Asp (V600D), and p.Val600Arg (V600R). Acceptable results from positive and negative controls are necessary for interpretation.

Reference Range: No mutation detected

Reportable Range: Positive or No mutation detected

Assay Availability: Batched weekly

Results Reported: 7 days

Specimen: Formalin fixed paraffin embedded tissue sections representing tumor OR cytology cell block sections representing tumor.

Volume: One H&E slide and at least 5 unstained slides.

Storage: Tissue blocks can be stored at room temperature.

Causes for Rejection: Insufficient tumor cellularity (<10%); decalcified specimens; specimens treated with Bouin's, B5, or other fixatives.

Laboratory Contact: For further information, please call the Molecular Diagnostics

Laboratory at (501) 526-6439.