

BRAF Mutation

Test Description:

Detection of *BRAF* exon 15 mutations in melanoma.

Indication:

Mutations in the *BRAF* gene are found in ~50-60% of melanomas with *BRAF* V600E as the most common mutation type (90%) followed by V600K, V600D, and V600R (1,2). Targeted therapy using selective *BRAF* inhibitors (i.e. PLX4032) may be effective in cases with *BRAF* mutations (3-5). However, not all tumors reveal such correlation and, therefore, the results of mutational testing should be interpreted in the context of the histologic findings and the patient's clinical history.

1. Fecher LA et al. Toward a molecular classification of melanoma. *J Clin Oncol.* 2007 20;25(12):1606-20.
2. Willmore-Payne et al. Human malignant melanoma: detection of BRAF- and c-kit activating mutations by high-resolution amplicon melting analysis. *Hum Pathol* 2005 36:486-93.
3. Smalley KS et al. Genetic subgrouping of melanoma reveals new opportunities for targeted therapy. *Cancer Res* 2009 69:3241-4.
4. Sondak et al. Targeting mutant BRAF and KIT in metastatic melanoma: ASCO 2009 meeting report. *Pigment Cell Melanoma Res* 2009, 22:386-387.
5. Yang H, et al. RG7204 (PLX4032), a Selective BRAFV600E Inhibitor, Displays Potent Antitumor Activity in Preclinical Melanoma Models. *Cancer Res.* 2010 Jun 15. [Epub ahead of print].

Methodology:

H&E slides are reviewed by a pathologist to confirm the specimen adequacy and select an area of tumor for the molecular analysis. Tumor cells are manually microdissected for DNA isolation. PCR-based DNA Sanger sequencing is used for detection of mutation.

Loci Tested:

BRAF exon 15

Performed: Mon-Fri

Reported: Within 5 business days of receipt.

Specimens Required:

1 H&E and 6 blank slides or frozen tissue.

Shipment Must Include:

- 1 H&E and 6 blank slides of each block or frozen tissue
- MAP lab requisition form
- Patient's pathology report