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REGIONAL PATHOLOGY SERVICES
DEPARTMENT OF PATHOLOGY AND MICROBIOLOGY
www.reglab.org

BRAF c.1799T>A (p.Val600Glu) mutation detection

This test is currently validated and recommended for mutation detection in colorectal adenocarcinomas, melanomas and papillary thyroid cancer.

CLINICAL SIGNIFICANCE

The human *BRAF* gene encodes a serine-threonine protein kinase which is a signaling mediator in the RAS/RAF/MAP kinase signal transduction cascade involved in the stimulation of cell proliferation. This specific c.1799T>A *BRAF* mutation has been identified in multiple cancers including colorectal carcinoma, melanoma and papillary thyroid cancer. In colorectal carcinoma, this mutation has been shown to render the tumor unresponsive to Epidermal Growth factor Receptor inhibitors, similar to KRAS mutations. Alternatively, the BRAF c.1799T>A (p.Val600Glu) mutation can also be used to help differentiate a sporadic microsatellite instability-high (MSI-H) colon cancer from one related to the hereditary non-polyposis colorectal cancer syndrome (HNPCC or Lynch Syndrome). Additionally, the BRAF c.1799T>A (p.Val600Glu) mutation is detected in approximately 50-60% of melanomas and also occurs in about 45% of papillary thyroid cancers where it is thought to indicate more aggressive behavior within the thyroid neoplasm.

TEST METHOD

-Pyrosequencing

PERFORMED

-Monday through Friday

REPORTED

-Report within 5 to 7 days

SPECIMEN REQUIRED

- Paraffin embedded formalin fixed tissue block (with at least 25% tumor), or 6 Unstained slides-10µm thick with ≥50% tumor (with one adjacent H&E). Minimum volume: 5mm X 5mm area if tissue is 100% tumor; 10mm X 10mm if tissue is 50%
- Tissue snap frozen is acceptable but not preferred.

SPECIMEN TRANSPORT

- Ambient Temperature: Paraffin blocks, slides
- Frozen: Snap Frozen Tissue

UNACCEPTABLE CONDITIONS

- Paraffin blocks with ≤10% tumor. Tissue fixed in heavy metal fixative or decalcified tissue.

RESULTS:

- Negative:** No BRAF c.1799T>A (p.V600E) mutation was identified in the *BRAF* gene by DNA amplification and pyrosequencing analysis.
- Positive:** A BRAF c.1799T>A (p.V600E) mutation was identified in the *BRAF* gene by DNA amplification and pyrosequencing analysis