Melanoma: Sequencing Panel

Test Code: MM401
Turnaround time: 4 weeks
CPT Codes: 81216 x1, 81272 x1, 81287 x1, 81311 x1, 81321 x1, 81404 x1, 81405 x1

**Condition Description**

Mutations in the p16 gene (also called CDKN2A or INK4A), are associated with hereditary melanoma and hereditary pancreatic cancer. Changes in the p16 gene increase cancer risk, making a melanoma diagnosis up to 50 times more likely by age 50. Mutations in other genes have also been implicated in the development of melanoma.

**Genes**

BAP1, BRCA2, CDK4, CDKN2A, KIT, MGMT, NRAS, PTEN, RB1, TP53, WRN

**Indications**

The test is indicated for:

- Individuals with a clinical or suspected diagnosis of melanoma.

**Methodology**

**Next Generation Sequencing**: In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

**Detection**

**Next Generation Sequencing**: Clinical Sensitivity: Unknown. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions/duplications will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical/biochemical phenotype.

Analytical Sensitivity: ~99%.

**Specimen Requirements**

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 60 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**Special Instructions**

This test is for germline mutation analysis. DNA isolated from FFPE tumor samples is not suitable for this test.
Related Tests

- Hereditary Cancer Syndrome: Sequencing Panel.
- Melanoma: Deletion/Duplication Panel.