Melanoma: Deletion/Duplication Panel

Test Code: MD401  
Turnaround time: 2 weeks  
CPT Codes: 81228 x1, 81323 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, NRAS, PTEN, RB1, TP53, WRN

Indications

The test is indicated for:

- Individuals with a clinical or suspected diagnosis of melanoma.

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

Detection

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood (EDTA)

Specimen Requirements:
EDTA (Purple Top)
Infants and Young Children (2 years of age to 10 years old): 3-5 ml
Older Children & Adults: 5-10 ml
Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

Type: DNA, Isolated

Specimen Requirements:
Microtainer
3µg
Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:
Refrigerate until time of shipment in 100 ng/µL in 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: Sequencing Panel.