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. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

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

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: 10 µg

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: Sequencing Panel.