Melanoma: Deletion/Duplication Panel

Test Code: MD401
Turnaround time: 2 weeks
CPT Codes: 81228 x1, 81323 x1

Condition Description

Mutations in the \textit{p16} gene (also called \textit{CDKN2A} or \textit{INK4A}), are associated with hereditary melanoma and hereditary pancreatic cancer. Changes in the \textit{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

\textbf{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

\textbf{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

\textbf{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

\textit{Submit only 1 of the following specimen types}

\textbf{Type: Whole Blood (EDTA)}

\textbf{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

\textbf{Specimen Collection and Shipping:}

Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

\textbf{Type: DNA, Isolated}

\textbf{Specimen Requirements:}

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

\textbf{Specimen Collection and Shipping:}

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

\textbf{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.