Brain, CNS, and PNS Cancer: Deletion/Duplication Panel

**Test Code:** MD204  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81297 x1, 81203 x1, 81294 x1, 81300 x1, 81403 x1, 81404 x1

**Condition Description**

Approximately 5% of primary brain cancers have known hereditary factors. Specifically, Li-Fraumeni syndrome, p53 defects, neurofibromatosis, tuberous sclerosis, von Hippel-Lindau disease, Turcot’s syndrome, and familial polyposis increase the risk of brain tumors.

In 2013, an estimated 23,130 people in the United States will be diagnosed with primary malignant brain and other central nervous system (CNS) neoplasms.

References:


**Genes**

ALK, APC, ATM, MEN1, MLH1, MSH2, MSH6, NBN, NF2, PALB2, PHOX2B, PTCH1, SUFU, TP53, VHL

**Indications**

The test is indicated for:

- Individuals with a clinical or suspected diagnosis of brain, CNS, or PNS cancer.

**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: 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.

**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 24 hours of collection. Do not refrigerate or freeze.

**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.
- Brain, CNS, and PNS Cancer: Sequencing Panel.