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

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

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