**IDH1 and IDH2 Mutations**

**Test Description:**
Detection of mutations in the IDH1 and IDH2 genes in brain tumors.

**Indication:**
Mutations in either isocitrate dehydrogenase enzyme isoforms 1 (IDH1) and 2 (IDH2) genes have recently been identified in a large proportion of diffuse astrocytomas (70-90%), oligodendrogliomas (69-94%), oligoastrocytomas (78-100%), and secondary glioblastomas (82-88%)(1-7). Primary glioblastomas showed low frequency of IDH1/IDH2 mutations (0-5%). These mutations are not found in non-neoplastic conditions that can often mimic gliomas (e.g. radiation changes, viral infections, infarctions, etc.) (7). Approximately 90% of all mutations occur in codon 132, exon 4 of the IDH1 gene, and less frequently in codon 172, exon 4 of the IDH2 gene (4). It appears that patients with tumors harboring IDH1 and IDH2 mutations have better outcome than those with wild-type IDH genes (4, 6).


**Methodology:**
H&E slides are reviewed by a pathologist to confirm the specimen adequacy and select an area of tumor for the molecular analysis. Tumor cells are manually microdissected for DNA isolation. Real time PCR and post-PCR melting curve analysis is used for detection of mutations. Positive results are confirmed by sequencing analysis.
Loci Tested:  
*IDH1* and *IDH2*

Performed:  Mon-Fri

Reported:  Within 5 business days of receipt.

Specimens Required:  
1 H&E and 6 blank slides or frozen tissue.

Shipment Must Include:  
- □ 1 H&E and 6 blank slides of each block or frozen tissue
- □ MAP lab requisition form
- □ Patient’s pathology report