**IDH1/2 Mutation Detection**

**Indication for Use:** Acute myeloid leukemia (AML) is a clinically and genetically heterogeneous disease that has poor prognosis. Combinations of mutations interact to drive the initiation and progression of AML and may create unique sensitivities to epigenetic-focused and other targeted or chemotherapies. Approximately 12% of AML patients have mutations in IDH2. IDH1 mutations are slightly less common. The phenotype of IDH1 and IDH2 mutant AML is similar, characterized by the gain-of-function activity which impairs cellular differentiation.

Brain tumors (gliomas) with IDH1 or IDH2 mutations have distinctive genetic and clinical characteristics, and patients with such tumors have a better outcome than those with wild type IDH genes. Mutation of IDH1 occurs early in glioma progression with somatic mutations of the R132 residue of IDH1 identified in majority (>70%) of grades II and III astrocytomas and oligodendrogliomas, as well as in secondary GBMs that developed from these lower grade lesions. Mutation analysis of closely related IDH2 revealed mutations of IDH2 residue R172 or R140, with most mutations occurring in tumors lacking IDH1 mutations.

**Testing Method:** Mutations in IDH1 and IDH2 are detected by amplicon based targeted next generation sequencing.

**Turn Around Time:** 5-7 business days

**Sample Requirements:**

**AML:**

Blood - Specimen stability: Ambient - 72 hours; Refrigerated - 1 week

- 3 ml peripheral blood in lavender top tube (EDTA)

  Note: One lavender tube of blood is sufficient for multiple DNA based tests

Bone marrow aspirates (anticoagulated with either heparin or EDTA and, if possible, placed into tissue culture medium) - Specimen stability: Refrigerated - 1 week (ship cold)

**Brain tumors (gliomas):**

The presence of adequate tumor in the material submitted for analysis should be confirmed by a surgical pathologist. A section from archival paraffin material should be confirmed to contain > 50% tumor by a surgical pathologist. If the submitted material for analysis contains < 50% of tumor, areas of predominant tumor will be microdissected to enrich for neoplastic cells.

- Formalin-fixed, paraffin-embedded tissue
- 5-6 tissue sections (please include H&E slide and a copy of pathology report)
- Cytology slides

**CPT Codes:** 81120, 81121, G0450 (88381 may apply)