

## **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)

