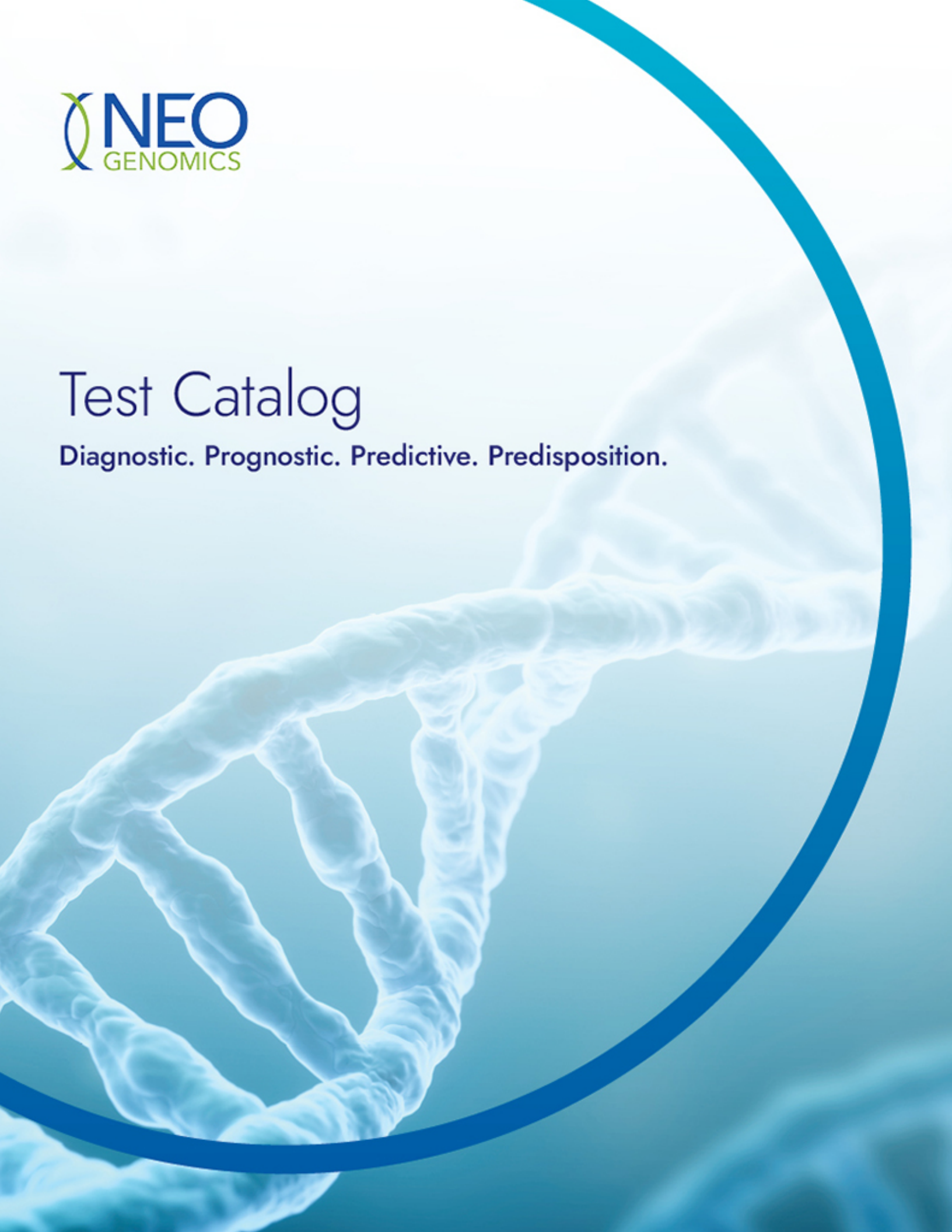
NEO GENOMICS

Test Catalog

Diagnostic. Prognostic. Predictive. Predisposition.



IDH1/IDH2 Mutation Analysis by PCR

Alternative Name
IDH1 Mutation Analysis, IDH2 Mutation Analysis

Methodology
Molecular

Test Description
Detection of mutations at IDH1 codons R132X and R100Q and at IDH2 codons R140X and R172X is performed using real-time PCR. IDH1 and IDH2 are analyzed concurrently. For solid tumors, tumor enrichment is performed before extraction.

Clinical Significance
Mutations in the enzyme isocitrate dehydrogenase 1 (IDH1) and IDH2 genes have been identified in a variety of tumors including central nervous system gliomas, cholangiocarcinoma, acute myeloid leukemia, blast-phase myeloproliferative neoplasms (MPNs) and chronic-phase primary myelofibrosis (PMF). Per professional practice guidelines, testing for IDH mutations has diagnostic and prognostic implications in the workup of gliomas and guides therapy selection in AML. AML patients with IDH mutations may respond to venetoclax-based therapy or IDH inhibitors.

Specimen Requirements
- **Peripheral blood:** 5 mL in EDTA tube.
- **Bone marrow:** 2 mL in EDTA tube.
- **FFPE solid tumor tissue:** Paraffin block is preferred. Alternatively, send 1 H&E slide plus 5-10 unstained slides cut at 5 or more microns. Please use positively-charged slides and 10% NBF fixative. Do not use zinc fixatives.

Note: NOT validated for Freeze & Hold option.

Storage & Transportation
Use cold pack for transport, making sure cold pack is not in direct contact with specimen. Slides can be packed at room temperature.

CPT Code(s)*
81120, 81121

Medicare MolDX CPT Code(s)*
81479

New York Approved
Yes

Level of Service
Global

Turnaround Time

5 days

References

1. Han S, Liu Y, Cai SJ, et al. IDH mutation in glioma: molecular mechanisms and potential therapeutic targets. Br J Cancer. 2020;122(11):1580-1589. PMID: 32291392.
2. DiNardo CD, Stein AS, Stein EM, et al. Mutant isocitrate dehydrogenase 1 inhibitor ivosidenib in combination with azacitidine for newly diagnosed acute myeloid leukemia. J Clin Oncol. 2021;39(1):57-65. PMID: 33119479.
3. Stein EM, DiNardo CD, Fathi AT, et al. Molecular remission and response patterns in patients with mutant-IDH2 acute myeloid leukemia treated with enasidenib. Blood. 2019 Feb 14;133(7):676-687. PMID: 30510081.

*The CPT codes provided with our test descriptions are based on AMA guidelines and are for informational purposes only. Correct CPT coding is the sole responsibility of the billing party.

Please direct any questions regarding coding to the payor being billed.

NeoGenomics Laboratories is a specialized oncology reference laboratory providing the latest technologies, testing partnership opportunities, and interactive education to the oncology and pathology communities. We offer the complete spectrum of diagnostic services in molecular testing, FISH, cytogenetics, flow cytometry, and immunohistochemistry through our nation-wide network of CAP-accredited, CLIA-certified laboratories.

Committed to research as the means to improve patient care, we provide Pharma Services for pharmaceutical companies, in vitro diagnostic manufacturers, and academic scientist-clinicians. We promote joint publications with our client physicians. NeoGenomics welcomes your inquiries for collaborations. Please contact us for more information.

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