



Test Catalog

Diagnostic. Prognostic. Predictive. Predisposition.





PDGFRA Mutation Analysis

Alternative Name

PDGFR Alpha Mutation Analysis

Methodology

Molecular

Test Description

Bi-directional sequencing of exons 12 and 18 of the PDGFRA (platelet-derived growth factor alpha) gene. These exons are mutation hotspots that account for the majority of PDGFRA mutations detected in gastrointestinal stromal tumors (GISTs) including the common TKI-resistance mutation D842V. Solid tumor enrichment is performed before extraction.

[PDGFRA Amplification](#) by FISH is available for detection of PDGFRA amplifications, which occur in approximately 30% of pediatric and 20% of adult high-grade astrocytomas, and may have prognostic and/or therapeutic implications.

[PDGFRA Rearrangement](#) by FISH is also available for detection of FIP1L1-PDGFR fusion, which may be found in myeloid/lymphoid neoplasms with eosinophilia. Bone marrow and peripheral blood are acceptable specimens.

Clinical Significance

PDGFRA mutations are found in soft-tissue sarcomas including gastrointestinal stromal tumors (GISTs). Identification of mutations is informative for sensitivity or resistance to tyrosine kinase inhibitor (TKI) therapy.

Specimen Requirements

- **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.

Storage & Transportation

Use cold pack for transporting block during summer to prevent block from melting. Slides can be packed at room temperature.

CPT Code(s)*

81314

New York Approved

No

Level of Service

Global

Turnaround Time

10 days

*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.

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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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