

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.

<u>PDGFRA Amplification</u> 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.

<u>PDGFRA Rearrangement</u> by FISH is also available for detection of FIP1L1-PDGFRA 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

Level of Service

Global

Turnaround Time

10 days

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

^{*}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.

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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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