



Test Catalog

Diagnostic. Prognostic. Predictive. Predisposition.



TERT Promoter Mutation Analysis

Alternative Name

TERT Promoter Mutation

Methodology

Molecular

Test Description

Bi-directional Sanger sequencing is performed using PCR primers designed to target mutations in the promoter region of TERT.

Clinical Significance

TERT gene promoter mutations lead to constitutive activation and expression. This in turn leads to replication and proliferation of cancer cells. Mutations in the TERT promoter are found in approximately 70% of melanomas, 80–90% of glioblastoma multiforme, 60% of hepatocellular carcinoma, 60% of bladder cancer, 70% of basal cell carcinoma, 50% of cutaneous squamous cell carcinoma and up to 30% of thyroid cancers. In thyroid cancers, TERT promoter mutations are detected in approximately 10% of papillary, 40% of poorly differentiated, and 70% of anaplastic carcinomas. In papillary thyroid carcinomas, the co-presence of mutations in the TERT promoter region and BRAF are associated with significantly more aggressive disease and shorter survival.

Similarly, TERT promoter mutations in melanoma are associated with more aggressive disease, especially when associated with a BRAF mutation.

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)*

81345

Medicare MoIDX CPT Code(s)*

81479

New York Approved

No

Level of Service

Global

Turnaround Time

14 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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