MAYO CLINIC LABORATORIES

**NEW TEST**

Notification Date: August 11, 2023
Effective Date: September 12, 2023

# *TERT* Promoter Mutation Analysis, Droplet Digital PCR, Tumor

**Test ID:** TERTD

## Useful for:

Identifying specific mutations within the *TERT* promoter that assist in tumor diagnosis/classification

## Genetics Information:

- This test uses droplet digital PCR (ddPCR) to evaluate for the presence of the c.-124C>T (also known as C228T) and c.-146C>T (also known as C250T) somatic mutations in the promoter region of the *TERT* gene. *TERT* promoter analysis ddPCR is a highly sensitive testing platform that can detect the c.-124C>T (C228T) and c.-146C>T (C250T) hotspot mutations at low levels, which may be observed in specimens with low number or proportion (%) of tumor cells/cells of interest.
- This test cannot differentiate between somatic and germline variant origin and is not intended to assess for germline risk.

## Additional Tests:

| Test ID | Reporting Name | Available Separately | Always Performed |
|---|---|---|---|
| SLIRV | Slide Review in MG | No | Yes |

## Testing Algorithm:

- When this test is ordered, slide review will always be performed at an additional charge.
- The preferred test to assess for somatic hotspot mutations in TERT and IDH1/2 genes is IDTRT (IDTRT / IDH1, IDH2 and TERT Mutation Analysis, Next-Generation Sequencing, Tumor). If TERTD is ordered with IDHT, both tests will be canceled and reordered as IDTRT.

## Methods:

Droplet Digital Polymerase Chain Reaction (ddPCR)

## Reference Values:

An interpretive report will be provided.

## Specimen Requirements:

**This assay requires at least 5% nuclei of tumor cells/cells of interest**

- Preferred amount of tumor area with sufficient percent tumor nuclei: tissue 144 mm(2)
- These amounts are cumulative over up to 10 unstained slides and must have adequate percent tumor nuclei of tumor cells/cells of interest.
- Tissue fixation: 10% neutral buffered formalin, not decalcified
- Cytology fixatives: Cytology smears fixed in alcohol and thin preps fixed with CytoLyt

### Preferred:

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|---|---|
| **Specimen Type:** | **Tissue block** |
| **Collection Instructions:** | Submit a formalin-fixed, paraffin-embedded tissue block with acceptable percent nuclei of tumor cells/cells of interest |

### Acceptable:

| | |
|---|---|
| **Specimen Type:** | **Tissue Slide** |
| **Slides:** | 1 Stained and 10 unstained |
| **Collection Instructions:** | Submit 1 slide stained with hematoxylin and eosin and 10 unstained, nonbaked slides with 5-micron thick sections.<br>Note: The total amount of required cell nuclei can be obtained by scraping up to 10 slides from the same block. |
| **Note:** | The total amount of required tumor nuclei can be obtained by scraping up to 10 slides from the same block. |

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|---|---|
| **Specimen Type:** | **Cytology slide (direct smears or ThinPrep)** |
| **Slides:** | 1 to 3 Slides |
| **Collection Instructions:** | Submit 1 to 3 slides stained and coverslipped with a preferred total of 3000 nucleated cells, or a minimum of at least 350 nucleated cells. |
| **Note:** | Glass coverslips are preferred; plastic coverslips are acceptable but will result in longer turnaround times. |
| **Additional Information:** | Cytology slides will not be returned. |

## Specimen Stability Information:

| Specimen Type | Temperature | Time | Special Container |
|---|---|---|---|
| Varies | Ambient (preferred) | | |
| | Refrigerated | | |

## Ordering Guidance:

The preferred test to assess for somatic hotspot mutations in TERT and IDH1/2 genes is IDTRT (IDTRT / IDH1, IDH2 and TERT Mutation Analysis, Next-Generation Sequencing, Tumor). If TERTD is ordered with IDHT, both tests will be canceled and reordered as IDTRT.

## Necessary Information:

**A pathology report (final or preliminary)**, at minimum containing the following information, **must accompany specimen for testing to be performed**:

1. Patient name
2. Block number-must be on all blocks, slides, and paperwork (can be handwritten on the paperwork)
3. Tissue collection date
4. Source of the tissue

## Cautions:

- This test cannot differentiate between somatic and germline alterations. Additional testing may be necessary to clarify the significance of results if there is a potential hereditary risk.
- A negative (wildtype) result does not rule out the presence of a mutation that may be present but below the limits of detection of this assay. The analytical sensitivity of this assay for mutation detection is 1% mutant copies in a sample with 5% or more tumor cells/cells of interest.
- This test detects *TERT* promoter mutations in 2 hotspots (C228T and C250T) only. Other alterations within the *TERT* promoter are not detectable by this test.
- Rare alterations (ie, polymorphisms) may be present that could lead to false-negative or false-positive results.
- Test results should be interpreted in the context of clinical findings, tumor sampling, and other laboratory data. If results obtained do not match other clinical or laboratory findings, contact the laboratory for updated interpretation. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- Reliable results are dependent on adequate specimen collection and processing. This test has been validated on cytology slides and formalin-fixed, paraffin-embedded tissues; other types of fixatives are discouraged. Improper or other treatment of tissues, such as decalcification, may cause droplet digital polymerase chain reaction failure.

## CPT Code:

81345
88381 – Microdissection, manual

**Day(s) Performed:** Monday through Friday

**Report Available:** 8-10 days

## Questions

Contact Michelle Raths, Laboratory Resource Coordinator at 800-533-1710.