MAYO CLINIC
Mayo Medical Laboratories

TEST ID: TERT

TERT PROMOTER ANALYSIS, TUMOR

USEFUL FOR

Assisting in central nervous system tumor classificationr

CLINICAL INFORMATION

TERT gene encodes the catalytic subunit of telomerase, an enzyme complex that regulates telomere length. *TERT* promoter mutations in 2 hotspots (C228T and C250T) have been shown to increase telomerase activity and contribute to tumorigenesis by allowing cancer cells to overcome cellular senescence. Among central nervous system tumors, *TERT* promoter mutations have primarily been identified in adults, with highest frequencies in oligodendroglioma, primary glioblastoma, solitary fibrous tumor, and medulloblastoma. Although less frequent, *TERT* promoter mutations have also been observed in lower-grade infiltrating (diffuse and anaplastic) astrocytomas and ependymoma, and are rare or absent in other central nervous system tumor types. The presence of *TERT* promoter mutations have been associated with a less favorable prognosis in lower-grade (grade II/III) diffuse gliomas that lack IDH1/2 mutations and have intact 1p/19q ("IDH-wildtype astrocytomas"), and with a more favorable prognosis in prognosis in grade II/III IDH1/2-mutant and 1p/19q-codeleted diffuse gliomas ("IDH-mutant and 1p/19q codeleted oligodendrogliomas"). Assessment of *TERT* promoter mutation status in central nervous system tumors may assist in tumor classification and provide prognostically relevant information for subgroups of patients with lower-grade diffuse gliomas.

TERT gene mutations are also observed in a variety of non-central nervous system (CNS) tumor types. In hepatocellular neoplasms *TERT* promoter mutations occur frequently in hepatocellular carcinomas and are believed to be an early step in hepatocarcinogenesis. However, *TERT* promoter mutations are not specific to hepatocellular carcinoma and have been reported as a key alteration in the rare progression of hepatocellular adenomas to hepatocellular carcinomas. As such, identification of a *TERT* promoter mutation suggests a hepatocellular neoplasm with an increased risk for aggressive behavior.

INTERPRETATION

An interpretive report will be provided.

REFERENCE VALUES

An interpretative report will be provided.

ANALYTIC TIME

12 days

SPECIMEN REQUIRED

Detailed on back side of this sheet.

02/2018

SPECIMEN REQUIRED

PREFERRED

Specimen Type
Tissue block

Collection Instructions
Submit a formalin-fixed, paraffin-embedded tissue block

ACCEPTABLE

Slides
1 stained and 10 unstained slides

Collection Instructions
Submit 1 slide stained with hematoxylin and eosin and 10 unstained slides (nonbaked, charged slides preferred) with 5-micron thick sections of the tumor tissue.

Specimen Type
Cytology slide (Direct smears or ThinPrep)

Slides
1-2 slides

Collection Instructions
Submit 1-2 slides stained and coverslipped with at least 5,000 total nucleated cells

Additional Information
Cytology slides will not be returned.

SUPPORTIVE DATA

We have developed a next-generation sequencing assay to detect somatic mutations that can be used to assist in the classification and prognostication of central nervous system tumors.

This assay has been shown to be very reproducible, having a 100% concordance for intra- and interassay reproducibility experiments. All somatic mutations that had been previously identified by various other molecular methods were detected by this assay during accuracy studies. No pathogenic variants were detected in known mutation negative samples.