**TERT Promoter Mutation for Glioma Prognosis**

*TERT* promoter mutation, in concert with *IDH1/2* mutation status and chromosome 1p/19q FISH results, impact prognosis in patients with diffuse glioma.

**Pathobiology:**

The enzyme telomerase is required for maintenance of telomere length in dividing cells. Normal cells typically lack telomerase, but in some cancers, telomerase is activated by point mutation within the *TERT* gene promoter. The mutation is typically located 124 or 146 base pairs upstream of the transcriptional start site (known as -124C>T and -146C>T, or colloquially called C228T and C250T).

In glioma, the combination of *TERT* promoter, *IDH1/2*, and chromosome 1p/19q mutation status can identify prognostically-distinct molecular subgroups. In particular, grade 2 or 3 glioma harboring *TERT* promoter mutation, wild-type *IDH1/2*, and lacking 1p/19q deletion has a very poor prognosis comparable to that of glioblastoma (grade 4).

*TERT* promoter mutation is seen in many other cancer types, and studies are underway to assess impact on patient management.

**Test Indication:** Prognosis of grade II or III glioma.

**Specimen Requirements:**

This assay uses genomic DNA extracted from paraffin-embedded tumor tissue. Ten unstained slides from a diagnostic block should be submitted, along with an H&E stained slide marked by the pathologist to indicate the most tumor-rich region (at least 50% tumor nuclei required). After macrodissection and DNA extraction, the *TERT* promoter region is analyzed by Sanger sequencing. Results at positions -124C>T and -146C>T are interpreted by a pathologist.

**References:**


**To consult a pathologist** about indications for testing or the significance of a result, call the Molecular Genetics Lab at (984) 974-1825 or Dr. Gulley at (919) 843-4595. E-mail: margaret_gulley@med.unc.edu