

Patient ID 321	Patient Name TESTRNV, IMPLEMENTATION	Birth Date 1968-02-19	Sex M	Age 55
Order Number X100422594	Client Order Number X100422594	Ordering Physician TESTING	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 23 Jun 2023 07:00		

TERT Promoter Analysis ddPCR, Tumor

Result

MCR

Provided diagnosis: brain glioblastoma

The following CLINICALLY RELEVANT VARIANT was detected:

Gene: TERT

DNA change: c.-124C>T (also known as C228T)

Interpretation

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TERT encodes the telomerase reverse transcriptase protein and is activated through multiple mechanisms in several cancer types [PMID:26941407, PMID:18482052, PMID:9454332, PMID:3907856].

The TERT promoter c.-124C>T (also known as C228T, chr5:1,295,228 C>T) and c.-146C>T (also known as C250T, chr5:1,295,250C>T) generate a binding consensus sequence for ETS transcription factors within the TERT promoter and result in increased transcriptional activity, protein expression, telomerase activity, and telomere length as compared with wild-type TERT [PMID:23348503, PMID:23348506, PMID:26194807, PMID:26389665, PMID:27562490].

TERT promoter mutations are a diagnostic molecular biomarker for glioblastoma, IDH-wildtype and anaplastic meningioma and a supportive diagnostic molecular biomarker for oligodendroglioma, IDH-mutant and 1p/19q-codeleted [WHO Classification of Tumours Editorial Board. Central nervous system tumours. Lyon (France): International Agency for Research on Cancer, 5th ed., vol. 6, 2021]. TERT promoter mutations have also been recurrently reported in medulloblastoma, SHH-activated subgroup 4 (SHH-adult), diffuse pediatric-type high-grade glioma, H3-wildtype and IDH-wildtype subgroup RTK2, solitary fibrous tumor, choroid plexus tumors, pleomorphic xanthoastrocytoma (mainly anaplastic), astrocytoma, IDH-mutant and ependymoma (all locations) [PMID:24154961, WHO Classification of Tumours Editorial Board. Central nervous system tumours. Lyon (France): International Agency for Research on Cancer, 5th ed., vol. 6, 2021]. In other central nervous system and in peripheral nervous system tumors, TERT promoter mutations are rare or have not been reported [COSMIC, cBioPortal for Cancer Genomics].

The PRESENCE of a TERT promoter mutation supports the diagnosis of any of these tumor types and should be interpreted in correlation with histopathological and clinico-radiological findings and other genetic test results.

There are currently no known clinically approved therapies that specifically target TERT promoter mutations.

Additional Information

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CLINICAL TRIALS

Possible clinical trials of benefit for this patient can be found at the following sites:

- 1) ClinicalTrials.gov:
www.clinicaltrials.gov/ct2/search/advanced
- 2) Mayo Clinic: www.mayo.edu/research/clinical-trials/
- 3) National Cancer Institute: www.cancer.gov/clinicaltrials/search

Specimen

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Tissue, Tumor

Tissue ID

MCR

1234

Method

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Microscopic examination was performed by a pathologist to identify areas of tumor for enrichment by macrodissection. Tumor DNA was isolated and evaluated for the presence of hotspot c.-124C>T (C228T) and c.-146C>T (C250T) mutations in the promoter region of the TERT gene using droplet digital PCR analysis.

Disclaimer

1 MCR

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. If results obtained do not match other clinical or laboratory findings, please contact the laboratory for possible interpretation. Misinterpretation of results may occur if the information provided

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292



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is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results.

This assay was designed to detect only the c.-124C>T (C228T) and c.-146C>T (C250T) mutations in the promoter region of the TERT gene.

A negative result does not rule out the presence of a mutation

that may be present but below the limit of detection for this assay (approximately 1% mutant copies in a sample with ??% tumor cells/cell of interest).

Released By

Cristiane M. Ida, M.D.

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Received: 26 Jun 2023 13:03

Reported: 10 Jul 2023 11:55

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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