Patient ID	Patient Name		Birth Date	Sex	Age
SA00174622	NONCM, ABNORMAL		1990-10-30	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00174622	SA00174622	UNKNOWN,PROVIDER			
Account Information		**Collected**			
C7028846 DLMP Rochester		07 Mar 2025 08:00			

Neuro-Onc Panel, Mutations Only

Result

MCR

Provided diagnosis: glioblastoma

The following CLINICALLY RELEVANT VARIANT was detected:
Gene: TERT
DNA Change: c.–124C>T (also known as C228T)
Variant Allele Frequency: 37.6%

Microsatellite Instability (MSI) status: Stable (MSS)

NO reportable CLINICALLY RELEVANT SEQUENCE VARIANTS detected within the analyzed regions of the remaining tested genes.

This assay only evaluates mutations and small insertion/deletions. RNA Fusion/Transcript variant testing was not performed. If fusion testing is desired, consider Neuro-Oncology Panel, Fusions Only (Mayo Test ID: NONCR). Refer to mayocliniclabs.com or call 1–800–533–1710 for more information about specimen requirements for this test.

Interpretation

MCR

This result should be interpreted in correlation with histopathological and clinico-radiological findings and other available genetic test results.

1) TERT c.–124C>T (also known as C228T)

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].

Other TERT promoter variants have been rarely reported and have been shown or considered to be functionally significant [cBioPortal for Cancer Genomics].

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 mutations are rare or have not been reported [COSMIC, cBioPortal for Cancer Genomics].

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

Additional Information

MCR

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

REFERENCE TRANSCRIPTS

Sequence variant nomenclature is based on the following RefSeq accession numbers (build GRCh37 (hg19)):TERT NM_198253.

Performing Site Legend

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

MAYO CLINIC LABORATORIES

Patient ID	Patient Name		Birth Date	Sex	Age
SA00174622	NONCM, ABNORMAL		1990-10-30	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00174622	SA00174622	UNKNOWN,PROVIDER			
Account Information		**Collected**			
C7028846 DLMP Rochester		07 Mar 2025 08:00			

Method

MCR

Microscopic examination was performed by a pathologist to identify areas of tumor for enrichment by macrodissection.

Next generation sequencing is performed to test for the presence of microsatellite instability and a mutation within all coding regions (unless otherwise specified) and exon/intron boundaries of the following 89 targeted genes: ACVR1, AKT1, APC, ARID1A, ATRX, BAP1, BCOR, BRAF, CDKN2A, CDKN2B, CIC, CTNNB1, CYSLTR2, DAXX, DDX3X, DGCR8, DICER1, DROSHA, EED, EGFR, EIF1AX, ERBB2, FGFR1, FGFR2, FGFR3, FUBP1, GNA11, GNAQ, GNAS, H3–3A, H3–3B, H3C2, H3C3, HIST2H3C, HRAS, IDH1, IDH2, KBTBD4, KDM6A, KIT, KLF4, KRAS, LZTR1, MAP2K1, MET, MLH1, MSH2, MSH6, MTOR, NF1, NF2, NOTCH1, NRAS, NTRK1, NTRK2, NTRK3, PBRM1, PDGFRA, PIK3CA, PIK3R1, PLCB4, PMS2, POLD1, POLE, POLR2A, PPM1D, PRKCA, PRKAR1A, PTCH1, PTEN, PTPN11, RB1, SETD2, SF3B1, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SMO, SUFU, SUZ12, TCF12, TERT (Promoter), TP53, TRAF7, TSC1, TSC2, and VHL.

Variant nomenclature is based on build GRCh37 (hg19). For details about gene reference transcripts (RefSeq accession numbers), specific targeted regions of each gene, and additional information about this test, see www.mayocliniclabs.com (Test ID NONCM).

Disclaimer

1 MCR

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.

Variants of uncertain significance may be identified.

A negative result does not rule out the presence of a variant that may be present but below the limits of detection of this assay. The analytical sensitivity of this assay for sequence reportable alterations is 5% mutant allele frequency with a minimum coverage of 500X in a sample with =>20% tumor content.

Point mutations and small insertion/deletion mutations will be detected 89 genes. This test may detect single exon deletions but does not detect multi-exon deletions, duplications, or genomic copy number variants. DELINS of 1,000 bp or less are detectable with at least 50 supporting reads.

Variant allele frequency (VAF) is the percentage of sequencing reads supporting a specific variant divided by the total sequencing reads at that position. In somatic testing, VAF should be interpreted in the context of several factors including, but not limited to: tumor purity/heterogeneity/copy number status (ploidy, gains/losses, loss of heterozygosity) and sequencing artifact/misalignment [PMID: 27144058, PMID: 29769535].

This test cannot reliably determine if a variant identified in PMS2 exons 11–15 originated from PMS2 or the highly homologous pseudogene PMS2CL. In the instance that a reportable variant is detected in PMS2 exons 11–15, additional testing will be recommended in the patient report.

Rare polymorphisms may be present that could lead to false-negative or false-positive results.

The presence or absence of a variant may not be predictive of response to therapy in all patients.

Test results should be interpreted in the context of clinical, tumor sampling, histopathological, and other laboratory data. If results obtained do not match other clinical or laboratory findings, contact the laboratory for discussion. Misinterpretation of results may occur if the information provided is inaccurate and/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 treatment of tissues, such as decalcification, may cause PCR failure.

Specimen

MCR

Tissue, Tumor

Tissue ID

MCR

9898

Performing Site Legend

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

MAYO CLINIC LABORATORIES

Patient ID	Patient Name		Birth Date	Sex	Age
SA00174622	NONCM, ABNORMAL		1990-10-30	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00174622	SA00174622	UNKNOWN,PROVIDER			
Account Information		**Collected**			
C7028846 DLMP Rochester		07 Mar 2025 08:00			

Released By MCR

Cristiane M. Ida, M.D.

Received: 07 Mar 2025 12:26 **Reported:** 09 Mar 2025 18:11

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.

Performing Site Legend

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