MAYO CLINIC LABORATORIES

1-800-533-1710

IDTRT

IDH1, IDH2, and TERT Mutation Analysis, Next-Generation Sequencing, Tumor

Patient ID	Patient Name		Birth Date	Sex	Age
SA00174978	TESTINGRNV, NORMAL		1985-01-12	M	40
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00174978	SA00174978	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		28 Mar 2025 00:00			

IDH1/2 and TERT Mutation Analysis

Result

MCR

Provided diagnosis: mild glial hypercellularity and microglial activation (preliminary diagnosis)

No reportable sequence variants were detected within the analyzed regions of the IDH1, IDH2 and TERT genes.

Interpretation

MCR

IDH1 and IDH2: No clinically relevant sequence variant detected

IDH1 and IDH2 (IDH) encode highly homologous enzymes that are involved in the citric acid cycle and other metabolic processes, playing roles in normal cellular metabolism and in protection against oxidative stress and apoptosis; Idh1 is localized to the cytoplasm and peroxisome and Idh2 is localized to the mitochondria [PMID:20513808].

IDH mutations are a diagnostic molecular biomarker for astrocytoma, IDH-mutant and 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]. For astrocytoma, IDH-mutant, the IDH1 R132C mutation is enriched in Li-Fraumeni patients [PMID:19340432], and IDH1-mutant tumors in children and young adults are enriched for germline mutations in mismatch repair genes [PMID:32895736]. IDH mutations have also been recurrently reported in medulloblastoma and meningioma [PMID:23894344, PMID:28726821, PMID:32179909].

The ABSENCE of an IDH mutation does not support but does not exclude the diagnosis of any of these tumor types. If there is evidence of a tumor, the ABSENCE of an IDH mutation indicates that this tumor is IDH-wildtype. This result should be interpreted in correlation with histopathological and clinico-radiological findings and other genetic test results.

TERT: No clinically relevant sequence variant detected

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].

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].

The ABSENCE of a TERT promoter mutation does not support but does not exclude 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.

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

Specimen

MCR

Tissue, Tumor

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

Patient ID	Patient Name		Birth Date	Sex	Age
SA00174978	TESTINGRNV, NORMAL		1985-01-12	M	40
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00174978	SA00174978	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		28 Mar 2025 00:00			

Tissue ID

MCR

12345

Method

MCR

Microscopic examination is performed by a pathologist to identify areas of tumor for enrichment by macrodissection. DNA is extracted from FFPE or cytology slides, and next generation sequencing is performed to evaluate the presence of a mutation in all coding regions and exon/intron boundaries of the IDH1 and IDH2 genes and the TERT promoter.

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

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.

DNA 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 at least 20% tumor content.

Point mutations and small insertion/deletion mutations will be detected in the IDH1 and IDH2 genes and the TERT promoter only. This test may detect single exon deletions but does not detect multi-exon deletions, duplications, larger-scale genomic copy number variants, copy neutral loss of heterozygosity (LOH), or epigenetic modifications such as promoter methylation. DELINS of 1,000 bp or less are detectable with at least 50 or more 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].

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.

Released By

MCR

HUSSAM AL KATEB

Received: 29 Mar 2025 11:00 **Reported:** 08 Apr 2025 15:05

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