Patient ID	Patient Name		Birth Date	Sex	Age
SA00174997	TESTINGRNV, ABNORMAL		1955-03-02	F	70
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00174997	SA00174997	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		28 Mar 2025 00:00			

MayoComplete Lung Cancer Panel

Result

MCR

Provided diagnosis: lung small cell neuroendocrine carcinoma involving lymph node

The following CLINICALLY RELEVANT VARIANT was detected:
Gene: EGFR
DNA Change: c.2239_2251delinsC (Exon 19)
Amino Acid Change: p.L747_T751delinsP (Leu747_Thr751delinsPro)
Variant Allele Frequency: 59%

Microsatellite Instability (MSI) status: Stable (MSS)

The following VARIANTS OF UNCERTAIN SIGNIFICANCE were detected:
Gene: EGFR
DNA Change: c.624G>T (Exon 5)
Amino Acid Change: p.Q208H (Gln208His)
Variant Allele Frequency: 12.9%

Gene: EGFR
DNA Change: c.764G>A (Exon 7)
Amino Acid Change: p.R255Q (Arg255Gln)
Variant Allele Frequency: 68.2%

No other reportable sequence variants or rearrangements were detected within the analyzed regions of the tested genes listed in the method description.

Interpretation

MCR

1) EGFR c.2239_2251delinsC (p.L747_T751delinsP) (Exon 19)

EGFR encodes the Epidermal growth factor receptor (Egfr), a receptor tyrosine kinase that functions as an oncogene. Egfr activates signaling pathways, such as the Ras/Raf/MAPK and PI3K pathways, and stimulates the cell to grow and divide [PMID:17000658]. Amplification, mutation, and overexpression of EGFR may cause excessive proliferation and tumor formation [PMID:18337605, PMID:22127113].

EGFR mutations have been reported in 6–35% of lung samples [COSMIC, cBioPortal for Cancer Genomics].

FDA approved targeted therapy is available for patients with an advanced or metastatic NSCLC whose tumors harbor EGFR c.2239_2251delinsC (p.L747_T751delinsP) [fda.gov/drugs].

MSI: Stable (MSS)
NO evidence of microsatellite instability was detected suggesting the presence of normal DNA mismatch repair function within the tumor. Current data suggest that advanced stage solid tumors with intact mismatch repair (MSS) are less likely to be responsive to treatment with immunotherapies such as anti-PD-1 therapies [PMID 28596308, PMID 29355075].

VARIANTS OF UNCERTAIN SIGNIFICANCE

Multiple variants of uncertain significance were detected. The variants were not observed at significant frequency in population germline/cancer somatic variant databases nor predicted to be functionally significant based on variant type/in silico algorithm results. Additionally, there was no convincing published evidence of cancer association. Therefore, the clinical significance of the detected variants is unknown.

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)):EGFR NM_005228.

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
SA00174997	TESTINGRNV, ABNORMAL		1955-03-02	F	70
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00174997	SA00174997	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		28 Mar 2025 00:00			

Specimen MCR

Tissue, Tumor

Tissue ID MCR

12345

Method MCR

Microscopic examination is 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 and exon/intron boundaries of the following 12 targeted genes: ALK, BRAF, EGFR, ERBB2, HRAS, KRAS, MDM2, MET, NRAS, RET, ROS1, and STK11.

A qualitative PCR-based assay is performed to detect rearrangements (fusions) within the ALK, ROS1 and RET genes, MET exon 14 skipping, and expression imbalance for ALK, ROS1, RET, NTRK1, NTRK2 and NTRK3 genes.

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 MCLNG).

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 ALK, BRAF, EGFR, ERBB2, HRAS, KRAS, MDM1, MET, NRAS, RET, ROS1, and STK11 genes 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. This test does not detect point mutations, insertion/deletion mutations, large single or multi-exon deletions or duplications, or genomic copy number variants for the NTRK1, NTRK2, and NTRK3 genes.

Gene fusions (rearrangements) and expression imbalance involving the ALK, ROS1, RET, NTRK1, NTRK2, and NTRK3 genes only will be detected.

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.

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
SA00174997	TESTINGRNV, ABNORMAL		1955-03-02	F	70
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00174997	SA00174997	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		28 Mar 2025 00:00			

Released By MCR

Ying-Chun Lo, M.D., Ph.D.

Received: 29 Mar 2025 15:26 **Reported:** 07 Apr 2025 17:08

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