MAYO CLINIC LABORATORIES

1-800-533-1710

EGFRS

EGFR Gene, Targeted Mutation Analysis, 51 Mutation Panel, Tumor

Patient ID	Patient Name		Birth Date	Gender	Age
SA00150123	TESTING, EGFRS ABNORMAL		1985-01-10	F	37
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00150123	SA00150123	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		11 Jan 2022 08:15			

EGFR Gene, Mutation Analysis, Tumor

Result Summary

MCR

MUTATION IDENTIFIED

Result

MCR

Provided diagnosis: lung non-small cell carcinoma

EGFR status: Mutant (exon 20 insertion)

This specimen is negative for all other tested mutations (listed in Method).

Interpretation

[1] MCR

ASSOCIATIONS BETWEEN EGFR MUTATIONS AND LUNG CANCER

Approximately 35% of patients with lung adenocarcinoma have a somatic mutation in the EGFR gene (1). EGFR mutations, primarily those occurring in the tyrosine kinase domain (exons 18–21), result in constitutive activation of the RAS/MAPK signaling pathway (2–4).

Current data suggest that patients with non-small cell lung cancer (NSCLC) whose tumors harbor exon 20 insertion mutations are generally resistant to the traditional EGFR inhibitors (5, 6).

Clinically approved targeted therapies are available for adult patients with NSCLC that harbor EGFR exon 20 insertion mutations. Thus, the detection of an exon 20 insertion mutation suggests that this patient may respond to such therapy (7).

REFERENCES

1. cancer.sanger.ac.uk/cancergenome/projects/cosmic/
2. Nat Rev Cancer. 2007 Mar;7(3):169–81 (PMID 17318210)
3. Int J Cancer 2011;131(5):E822–829 (PMID 22161771)
4. Nat Rev Clin Oncol. 2011 Aug 23;8(11):661–8 (PMID 21862980)
5. Lancet Oncol. 2012 Jan;13(1):e23–31 (PMID 21764376)
6. Sci Transl Med. 2013 Dec 18;5(216):216ra177 (PMID 24353160)
7. https://www.fda.gov/drugs

ADDITIONAL INFORMATION

Microscopic examination was performed by a pathologist to identify areas of tumor for enrichment by macrodissection. A qualitative PCR-based assay employing fluorescently labeled probes was used to test for the presence of specific mutations within exons 18–21 of the EGFR gene (mutations at codon G719 in exon 18; small deletions in exon 19; T790M, S768I, and small insertions in exon 20; and L858R and L861Q in exon 21). Mutation nomenclature is based on GeneBank accession number NM_005228. Due to the sensitivity of the method being used, EGFR mutations cannot be reliably detected in samples containing less than 10% tumor cells. Samples are routinely macrodissected to enrich for tumor cells, with samples less than 10% rejected from further testing. Test results should be interpreted in the context of clinical findings, tumor sampling, 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 is inaccurate or incomplete.

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

Specimen

MCR

Tissue, Tumor

Tissue ID

MCR

1234

Released By

MCR

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

Received: 11 Jan 2022 09:26 **Reported:** 20 Jan 2022 11:20

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	William G. Morice M.D. Ph.D	24D0404292