MAYO CLINIC LABORATORIES

Test Definition: T790M

Cell-Free DNA EGFR T790M Mutation Analysis, Blood

Overview

Useful For

Determination of EGFR T790M mutation status in blood specimens as an alternative to invasive tissue biopsies

Identification of patients with non-small cell lung cancer who harbor a T790M mutation and may benefit from specific EGFR-targeted therapies

Genetics Test Information

This test evaluates cell-free DNA (cfDNA) in peripheral blood for the presence of the *EGFR* T790M mutation in patients with non-small cell lung cancer (NSCLC) and can be used to assess eligibility for targeted therapies. Current data suggests that patients with metastatic NSCLC and the T790M mutation may benefit from T790M-targeted therapy (eg, osimertinib).

This test is **not** validated for serial monitoring of patients with cancer. This test is also **not** intended as a screening test to identify cancer.

Method Name

Droplet Digital Polymerase Chain Reaction (ddPCR)

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Ordering Guidance

This test is **not** a prenatal screening test.

This test detects only the T790M mutation in the *EGFR* gene. It does **not** detect other *EGFR* gene mutations in exons 18 through 21.

This test provides rapid detection of the *EGFR* T790M mutation in peripheral blood from non-small cell lung cancer patients as an alternative for *EGFR* analysis of tissue. For tissue testing, order EGFRS / EGFR Gene, Targeted Mutation Analysis, 51 Mutation Panel, Tumor.

Shipping Instructions

1. Samples should be transported at ambient temperature or refrigerated (4 degrees C)

2. Samples are viable for 7 days in the Streck Black/Tan Top Tube Kit (T715)

Specimen Required

Supplies: Streck Black/Tan Top Tube Kit (T715)

Specimen Volume: Two, 10-mL Streck cell-free DNA (cfDNA) blood collection tubes

Additional Information:

1. Only blood collected in Streck cfDNA tubes will be accepted for analysis.
2. Whole blood will be processed to produce platelet-poor plasma before cfDNA isolation.

Forms

If not ordering electronically, complete, print, and send an Oncology Test Request (T729) with the specimen.

Specimen Minimum Volume

One 10 mL Streck tube

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)	7 days	Streck Black/Tan top
	Refrigerated	7 days	Streck Black/Tan top

Clinical & Interpretive

Clinical Information

Epidermal growth factor receptor (EGFR)-targeted tyrosine kinase inhibitors (eg, gefitinib and erlotinib) have been approved by the US food and Drug Administration (FDA) for use in treating patients with non-small cell lung cancer (NSCLC) who previously failed to respond to traditional chemotherapy. However, the *EGFR* T790M mutation is associated with acquired resistance to tyrosine kinase inhibitor (TKI) therapy in about 60% of patients with disease progression after initial response to erlotinib, gefitinib, or afatinib. Recent data suggest that patients with metastatic NSCLC and the T790M mutation may benefit from osimertinib, an FDA-approved oral TKI that inhibits both EGFR-activating mutations and the T790M mutation.

Reference Values

An interpretive report will be provided.

Interpretation

The interpretation of molecular biomarker analysis includes an overview of the results and the associated therapeutic implications.

Cautions

Patients with a negative test result may still harbor the *EGFR* T790M mutation. Mutation testing of a tissue specimen for

EGFR mutations should be considered for patients with a negative result with this test.

The limit of detection of this assay for the detection of *EGFR* mutations is influenced by the amount of cell-free DNA in the blood. This is a biological variable that cannot be controlled.

This assay was designed to detect the following T790M mutation in the *EGFR* gene.

This test has not been clinically validated for use as a tool to monitor response to therapy or for early detection of tumors.

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.

Supportive Data

This test has been evaluated by our laboratory as an alternative to assessing paraffin-embedded tumor specimens for the *EGFR* T790M mutation in patients with non-small cell lung cancer.

Clinical Reference

1. Schwarzenbach H, Hoon DSB, Pantel K: Cell-free nucleic acids as biomarkers in cancer patients. Nat Rev Cancer. 2011 Jun;11(6):426-437
2. Ettinger DS, Wood DE, Aisner DL, et al: Non-Small Cell Lung Cancer, Version 5.2017, NCCN Clinical Practice Guidelines in Oncology. J Natl Compr Canc Netw. 2017 Apr;15(4):504-535
3. Janne PA, Yang JCH, Kim DW, et al: AZD9291 in EGFR inhibitor-resistant non-small-cell lung cancer. N Engl J Med. 2015 Apr 30;372(18):1689-1699

Performance

Method Description

Blood samples are collected in Streck Cell-Free DNA BC Tubes. Cell-free DNA is isolated from double-centrifuged plasma and assessed for the presence of the *EGFR* T790M mutation using droplet digital polymerase chain reaction analysis.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Varies

Report Available

5 to 10 days

Specimen Retention Time

Whole blood: 2 weeks (if available); Extracted DNA: 3 months

Performing Laboratory Location

Rochester

Fees & Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification

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

CPT Code Information

81235

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
T790M	cfDNA EGFR T790M Test, Blood	55769-4

Result ID	Test Result Name	Result LOINC® Value
113411	Result Summary	50397-9
113412	Result	55769-4
113413	Interpretation	69047-9
113414	Additional Information	48767-8
113415	Specimen	31208-2
113416	Source	31208-2
113417	Released By	18771-6