MAYO CLINIC LABORATORIES

Test Definition: T790M

cfDNA EGFR T790M Test, 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 the 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.

Highlights

Evaluates peripheral blood for the presence of the T790M mutation in the *EGFR* gene in cell-free DNA
Rapid detection of the *EGFR* T790M mutation in non-small cell lung cancer patients as an alternative for *EGFR* analysis of tissue
Current data suggests that patients with metastatic NSCLC and the T790M mutation may benefit from T790M-targeted therapy (eg, osimertinib)

Method Name

Digital Droplet Polymerase Chain Reaction (PCR) of Plasma Cell-Free DNA

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Ordering Guidance

This test is **not** a prenatal screening test. To evaluate for the presence of common fetal chromosome abnormalities using cell-free DNA, see NIPS / Cell-Free DNA Prenatal Screen.
This test detects only the T790M mutation in the *EGFR* gene. It does **not** detect other *EGFR* gene mutations in exons 18 through 21. For that purpose, order EGFRD / Cell-Free DNA *EGFR* Exon 18, 19, 20, 21, Mutation Analysis, Blood.
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 EGFRT / *EGFR* Gene, Mutation Analysis, 29 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 (T-715)

Specimen Volume: Two 10-mL Streck Cell-Free DNA blood collection tubes

Additional Information:

1. Only blood collected in Streck Cell-Free DNA BCT tubes will be accepted for analysis. Whole blood will be processed to produce platelet poor plasma before cfDNA isolation.
2. Samples should be transported at room temperature or refrigerated (4 degrees C)
3. Samples are viable for 7 days in the Streck Cell-Free DNA BCT tube.

Forms

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

Reject Due To

No specimen collected in the proper container will be rejected.

Specimen Minimum Volume

One 10 mL Streck tube

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

EGFR-targeted tyrosine kinase inhibitors (eg, gefitinib and erlotinib) have been approved by the 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.

Interpretation

An interpretive report will be provided.

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 cfDNA 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.

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 DS, Pantel K: Cell-free nucleic acids as biomarkers in cancer patients. Nat Rev Cancer 2011;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;15(4):504-535

3. Janne PA, Yang JC, Kim DW, et al: AZD9291 in EGFR inhibitor-resistant non-small--cell lung cancer. N Engl J Med. 2015;372(18):1689-1699

Performance

Method Description

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

PDF Report

No

Performing Laboratory Location

Rochester

Fees & Codes

Test Classification

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 US Food and Drug Administration.

CPT Code Information

81235