MAYO CLINIC LABORATORIES

Test Definition: PIK3B

Cell-Free DNA PIK3CA Test, Blood

Overview

Useful For

Identification of hormone receptor positive and human epidermal growth factor receptor 2 negative (HR+/HER2-) advanced breast cancer tumors that may be eligible for treatment with targeted kinase inhibitor therapy (eg, alpelisib).

Method Name

Polymerase Chain Reaction (PCR)

NY State Available

Yes

Specimen

Specimen Type

Whole blood

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

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 cell-free DNA 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

More than 70% of breast cancers are hormone receptor (HR) positive and human epidermal growth factor receptor 2 (HER2) negative (HR+/HER2-). Approximately 40% of patients with HR+/HER2- advanced breast cancer have activating mutations in the gene *PIK3CA*, inducing hyperactivation of the alpha isoform (p110 alpha) of phosphatidylinositol 3-kinase, a key upstream component of the PI3K pathway. Mutations in *PIK3CA* are associated with tumor growth, resistance to endocrine therapy, and a poor overall prognosis.

Patients with HR+/HER2- advanced breast cancer identified to have a *PIK3CA* mutation may be eligible for treatment with targeted kinase inhibitor therapy (eg, alpelisib).

This test uses circulating tumor DNA extracted from blood to evaluate for the presence of 10 clinically actionable *PIK3CA* mutations:
E542K (c.1624G>A)
E542K (c.1633G>A)
E545D (c.1635G>T)
E545G (c.1634A>G)
E545A (c.1634A>C)
H1047Y (c.3139C>T)
C420R (c.1285C>T)
Q546E (c.1636C>G)
H1047L (c.3140A>T)
H1047R (c.3140A>G)

Reference Values

An interpretive report will be provided

Interpretation

The interpretation of molecular biomarker results includes an overview of the results and the associated diagnostic, prognostic, and therapeutic implications.

Cautions

A negative (wildtype) result does not rule out the presence of a mutation that may be present but below the limits of detection of this assay. It also does not rule out the presence of other types of alterations in the *PIK3CA* gene outside those that the assay was designed to detect.

This test is not designed to differentiate between somatic and germline alterations. Additional testing may be necessary to clarify the significance of results if there is a potential hereditary risk.

Not all tumors that have *PIK3CA* mutations will respond to targeted therapies.

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

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

Test results should be interpreted in 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.

Clinical Reference

1. Bachman KE, Argani P, Samuels Y, et al: The PIK3CA gene is mutated with high frequency in human breast cancers. Cancer Biol Ther. 2004 Aug;3(8):772-775
2. Andre F, Ciruelos E, Rubovszky G, et al: Alpelisib for PIK3CA-mutated, hormone receptor-positive advanced breast cancer. N Engl J Med. 2019 May 16;380(20):1929-1940
3. Andre F, Ciruelos EM, Juric D, et al: Alpelisib plus fulvestrant for PIK3CA-mutated, hormone receptor-positive, human epidermal growth factor receptor-2-negative advanced breast cancer: final overall survival results from SOLAR-1. Ann Oncol. 2021 Feb;32(2):208-217

Performance

Method Description

A polymerase-chain reaction (PCR)-based assay employing real-time PCR and allele-specific PCR technologies is used to test for 10 mutations within *PIK3CA* (C420R, E542K, E545A, E545D, E545G, E545K, Q546E, H1047L, H1047R, and H1047Y).(Package insert: therascreen PIK3CA RGQ PCR Kit. Qiagen; 05/2019)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

5 to 10 days

Specimen Retention Time

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

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 has been modified from the manufacturer's instructions. Its performance characteristics were 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

81309

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
PIK3B	cfDNA PIK3CA Test, Blood	60034-6

Result ID	Test Result Name	Result LOINC® Value
616647	Result Summary	50397-9
616648	Result	82939-0
616649	Interpretation	69047-9
616650	Additional Info	48767-8
616651	Specimen	31208-2
616652	Source	31208-2
616653	Released By	18771-6