

RIK3CA Breast Cancer

Order GTC Breast Cancer Profile

The FDA has approved Piqray (alpelisib) tablets, to be used in combination with the FDA-approved endocrine therapy fulvestrant, to treat postmenopausal women, and men, with hormone receptor (HR)-positive, human epidermal growth factor receptor 2 (HER2)-negative, PIK3CA-mutated, advanced or metastatic breast cancer following progression on or after an endocrine-based regimen.

FDA approval of the drug was based on detecting one of 11 frequent mutations in PIK3CA. Based on our data, these 11 mutations constitute only 62.83% of the mutation we detect when Next Generation Sequencing (NGS) is used.

Abnormalities in PIK3CA have been reported in approximately 30% of patients with breast cancer. Most of the reported mutations are gain of function. In addition to mutations, PIK3CA gene amplification has been reported in 10% of breast cancers. Based on our database more than 126 different mutations can be seen in PIK3CA gene when NGS is used for testing. Genomic Testing Cooperative (GTC) offers comprehensive PIK3CA mutation profiling as a part Breast Cancer profiling. GTC Breast Cancer Molecular profile includes mutations analysis and copy number variation testing of 40 different genes. In addition, tumor mutation burden (TMB) and microsatellite instability (MSI) testing results are also reported with this profile.

Tested Genes									
MSI	TMB	AKT1	APC	ATM	ATR	ATRX	BRAF	BRCA1	BRCA2
CDH1	CDKN2A	EGFR	EPCAM	ERBB2	ERBB3	ERBB4	ESR1	IDH1	IDH2
KIT	KMT2A	KMT2C	KMT2D	KRAS	MAP2K4	MAP3K1	MLH1	MSH2	MSH6
MTOR	NF1	NOTCH1	NOTCH2	NRAS	PDGFRA	PIK3CA	PIK3R1	PMS2	PTEN
RB1	SF3B1	TP53							

Specimen Requirements:

-FFPE: 1 H&E slide and 6-8 unstained slides, 5-7 microns of tissue fixed with 10% NBF fixative. Please circle tumor for microdissection. Alternatively, the FFPE block can be sent for tumor circling and cutting at our laboratory.

Shipping:

Ship using cold pack. The cold pack should not directly contact specimen. Ship as soon as sample collected with overnight delivery.

Turn Around Time:

5 - 7 days