NEO GENOMICS

866.776.5907, option 3

Molecular Genetics

PIK3CA Mutation CDx - Tissue

Client 1234
Sample Client

Address
City, ST 99999
Phone: (555) 555-5555
Fax: (555) 555-5555

Patient Name: **Patient, Sample**
Patient DOB / Sex: **01/01/1954 / F**
Specimen Type: **Tissue**
Body Site: **Left Breast**
Specimen ID: **X99-99**
MRN: **9999999**
Reason for Referral: **Breast Cancer**

Ordering Physician(s): **Sample Doctor, MD**
Treating Physician(s): **Sample Doctor, MD**
Accession / CaseNo: **9999999 / XXX99-999999**
Collection Date: **06/01/2020**
Received Date: **10/16/2020 04:01:00 PM EDT**
Report Date: **10/22/2020 04:08:22 PM EDT**

Interpretation:

PCR analysis demonstrates the PRESENCE of sensitizing mutation(s) in the PIK3CA gene, which predicts an increased likelihood of response to the PIK3CA inhibitor PIQRAY® (alpelisib).

Results:

Test	Result
PIK3CA Mutation CDx - Tissue	Positive
Mutation(s) Detected	p.E542K

Clinical Significance:

Somatic mutations of Phosphatidylinositol-4,5-Bisphosphate 3-Kinase Catalytic Subunit Alpha (PIK3CA) are associated with longer progression-free survival in patients with metastatic hormone receptor-positive and HER2-negative breast cancer when treated with Piqray® (alpelisib) and fulvestrant vs. fulvestrant alone (11.0 months vs 5.7 months) [1-2]. Approximately 40% of patients with advanced hormone receptor-positive breast cancer harbor a PIK3CA mutation [3].
NOTE: Due to tumor heterogeneity and sampling, tumor specimens containing mostly non-malignant tissue and stroma may have mutation levels below the limit of detection for this assay. Poor DNA quality resulting from improper fixation and storage of archival paraffin samples may cause assay failure.
QIAamp® is a registered trademark of the QIAGEN Group.

Methodology:

Following pathologist identification of tumor-rich areas based on hematoxylin and eosin (H&E) staining, DNA is extracted from microdissected formalin-fixed paraffin-embedded (FFPE) tissue sections using the QIAamp® DSP DNA FFPE tissue kit. The therascreen PIK3CA assay is a real time qualitative PCR assay for the detection of 11 mutations in the Phosphatidylinositol-4-,5-Biphosphate 3-Kinase Catalytic Subunit Alpha (PIK3CA) gene (Exon7: p.C420R, Exon 9: p.E542K, p.E545A, p.E545D [c.1635G > T only], p.E545G, p.E545K, p.Q546E, and p.Q546R, and exon 20: p.H1047L, p.H1047R and p.H1047Y). The therascreen PIK3CA companion diagnostic is FDA-approved to aid in identifying breast cancer patients who may be eligible for treatment with PIQRAY (alpelisib).

References:

References:
1. Andre F, Ciruelos EM, Rubovszky G et al. Alpelisib + fulvestrant for HR+, HER2- advanced breast cancer: Results of the Phase III SOLAR-1 trial. Presented at the European Society for Medical Oncology (ESMO) 2018 Congress (Abstract LBA3_PR) on October 20, 2018.
2. Sabine V, Crozier C, Brookes C, et al. Mutational analysis of PI3K/AKT signaling pathway in tamoxifen exemestane adjuvant multinational pathology study. Journal of Clinical Oncology. 2014;32:2951-2958.

Electronic Signature

Sample Doctor, MD, Pathologist

The Technical Component Processing and Analysis of this test was completed at Genoptix Rutherford, 2110 Rutherford Road, Carlsbad, CA / 92008 / 800-755-0802 / CLIA # 05D1018666. The Professional Component of this test was completed at NeoGenomics Florida, 12701 Commonwealth Drive, Suite 5, Fort Myers, FL / 33913 / 866-776-5907 / CLIA # 10D0998082. / Medical Director(s): Sample Director.
The performance characteristics of this test have been determined by NeoGenomics Laboratories. This laboratory is CLIA certified to perform high complexity clinical testing.
Images that may be included within this report are representative of the patient but not all testing in its entirety and should not be used to render a result.
The CPT codes provided with our test descriptions are based on MolDX and AMA guidelines and are for informational purposes only. Correct CPT coding is the sole responsibility of the billing party. Please direct any questions regarding coding to the payer being billed.