GenOnc Breast Cancer Panel

GENOME EXPLORATIONS
A DIVISION OF COMPASS LABORATORY SERVICES LLC

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Introduction

GenOnc Breast Cancer Targeted Panel is a collection of multiplexed PCR primer assays for targeted enrichment of the coding (exonic) regions of the 44 genes most commonly mutated in human breast cancer samples. Mutations in these oncogenes and tumor suppressor genes are often relevant for tumor classification, and warrant extensive investigation to enhance the understanding of carcinogenesis. Breast cancer is a heterogeneous disease, and each tumor subtype has a specific prognosis and molecular mechanism. These molecular mechanisms include point mutations, potentially rendering the gene of interest inactive or hyperactive. For example, PIK3CA (p110α), a PI3K catalytic subunit, is commonly mutated in a variety of breast cancer subtypes. One common activating mutation for PIK3CA is H1047R. This mutation can cause an increase in PI3K/PTEN/AKT signaling, a common carcinogenic mechanism. There are hundreds of additional identified PIK3CA mutations, and many have functional consequences. In addition, there are clearly many other genes that are highly mutated in breast cancer. Therefore, sequencing analysis is an efficient method to examine a tumor sample for multiple potentially mutated genes. This panel narrows the focus to the most relevant mutated genes in breast cancer, using a variety of resources such as recent whole genome/exome sequencing studies from scientific networks including the Cancer Genome Atlas. Curated databases such as the Cancer Gene Census and COSMIC (Catalogue of Somatic Mutations in Cancer) are also used.

GenOnc Breast Cancer Panel Genes

Carcinoma:
BAP1, CCND1, EP300, ESR1 (ERα), MAP3K1 (MEKK1), MDM2, MYC, PBRM1, PCGF2 (RNF110), WEE1, ZBED4

Adenoid Cystic Carcinoma:
PIK3CA (p110α), PTEN

Basal (Triple-Negative) Carcinoma:
BRCA1, BRCA2, CDH1, CDKN2A (p16INK4), EGFR, ERBB2 (HER2), ERBB3, EXOC2, FGFR1, ITCH, MLL3, MUC16, NEK2, PIK3CA (p110α), PIK3R1, PTEN, PTGFR, RB1, TP53

Ductal Carcinoma:
AKT1, ATM, BRCA1, BRCA2, CDH1, CDKN2A (p16INK4), EGFR, ERBB2 (HER2), FGFR2, GATA3, MLL3, MUC16, PIK3CA (p110α), PIK3R1, PTEN, TP53

Ductolobular Carcinoma:
PIK3CA (p110α)

ER-PR-Positive Carcinoma:
ACVR1B, AKT1, CBFB, CDH1, ERBB3, EXT2, PIK3CA, PPM1L, PTEN, SEPT9, TP53

HER-Positive Carcinoma:
CDH1, FBXO32, IRAK4, ITCH, MLL3, MUC16, NCOR1, PIK3CA, PTGFR, TP53, TRAF5

Lobular Carcinoma:
AKT1, CDH1, ERBB2 (HER2), PIK3CA, RB1, TP53

Luminal Carcinoma:
CDH1, CDKN2A (p16INK4), GATA3, MAP2K4, PIK3CA, PTEN, TP53

Medullary Carcinoma:
TP53

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Carcinoma in situ:
Ductal & Lobular Carcinoma in situ: CDH1
Ductal Carcinoma in situ: AKT1, CDH1, PIK3CA (p110α), TP53
Lobular Carcinoma in situ: CDH1

Hyperplasia
Atypical Ductal Hyperplasia: PIK3CA (p110α), PTEN
Atypical Lobular Hyperplasia: CDH1
Ductal Epithelial Hyperplasia-Hyperplasia of Usual Type: PIK3CA (p110α)

Other Tumors
Fibrosis: PIK3CA (p110α)
Neoplasm: TP53
Papilloma: AKT1
Phyllodes Tumor: RET, TP53