

# BREAST CARCINOMA NEXT GENERATION SEQUENCING SUB PANEL



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Breast carcinoma is the most commonly diagnosed cancer among women in South Africa and a leading cause of cancer-related mortality. Ampath is now offering a somatic breast carcinoma specific next generation sequencing (NGS) sub panel, which incorporates various actionable biomarkers, including *BRCA1* and *BRCA2* pathogenic variants. Tumour genetic testing allows for more accurate diagnoses, risk assessments, and individualized therapies, with the potential to improve outcomes and minimize unnecessary side effects.

## BREAST CARCINOMA SOMATIC NGS TESTING SUB PANEL

Mnemonic	Details	Genes tested	Turnaround time	Sample requirements
<b>OCASUB</b>	DNA and RNA panel, including <i>BRCA1</i> and <i>BRCA2</i>	<i>AKT1, ALK, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, CTNNB1, ERBB2, ERBB4, ESR1, HRAS, MDM2, MED12, MRE11, MSI, MYB, MYBL1, NBN, NF1, NRG1, NTRK1, NTRK2, NTRK3, PALB2, PIK3CA, PTEN, RAD50, RAD51C, RAD51D, RET, STK11, TERT, TP53</i>	14 working days	Formalin Fixed Paraffin Embedded Tissue (FFPE) 8-12 normal slides (not charged) with 10 micron thick unstained recuts or 10 micron thick recuts in an Eppendorf tube or Paraffin Embedded tissue block

Biomarker	Therapeutic Implications
<i>AKT1</i>	Capivasertib (+fulvestrant)
<i>BRCA1, BRCA2</i>	Somatic <i>BRCA1</i> and <i>BRCA2</i> emerging biomarker for olaparib response Germline screening
<i>CDH1</i>	Germline screening
<i>ERBB2</i>	Anti-HER2 therapies
<i>ESR1</i>	Elacestrant
<i>NTRK1, NTRK2, NTRK3</i>	Larotrectinib and entrectinib
<i>PIK3CA</i>	Alpelisib and capivasertib
<i>PTEN</i>	Alpelisib and capivasertib
<i>RET</i>	Selpercatinib



## OTHER BREAST CARCINOMA SOMATIC NGS TESTING OPTIONS AT AMPATH

Mnemonic	Details	Genes tested	Turnaround Time	Sample requirements
<b>EGFRASSEQ</b>	DNA only panel	<i>AKT1, CDH1, CDK4, CDKN2A, CHEK2, CTNNB1, ERBB2, ERBB4, ESR1, HRAS, IDH2, PIK3CA, PTEN, TP53</i>	10 days	Formalin Fixed Paraffin Embedded Tissue (FFPE) 8-12 normal slides (not charged) with 10 micron thick unstained recuts or 10 micron thick recuts in an Eppendorf tube or Paraffin Embedded tissue block
<b>ONCOBRCA</b>	Somatic <i>BRCA1</i> and <i>BRCA2</i> genes only	<i>BRCA1</i> and <i>BRCA2</i>	14 days	Formalin Fixed Paraffin Embedded Tissue (FFPE) 8-12 normal slides (not charged) with 10 micron thick unstained recuts or 10 micron thick recuts in an Eppendorf tube or Paraffin Embedded tissue block
<b>HRD</b>	Sequencing of HRR genes and commenting on the presence of HRD	<i>ABRAXAS1, ATM, ATR, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDK12, CHEK1, CHEK2, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MRE11, NBN, PALB2, PARP1, POLD1, POLE, PPP2R2A, PTEN, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54L, RNASE2A, RNASE2B, RNASE2C, RPA1, TP53, XRCC2, XRCC3</i>	14 days	Formalin Fixed Paraffin Embedded Tissue (FFPE) 8-12 normal slides (not charged) with 10 micron thick unstained recuts or 10 micron thick recuts in an Eppendorf tube or Paraffin Embedded tissue block