SureSeq

A Sysmex Group Company

Breast Cancer + CNV Panel

Features

Unparalleled uniformity and high depth of coverage

- Reliably detect germline variants in all exonic regions

CNV detection ranging from loss of single exons to full gene deletions and duplications

- Profile your samples for CNVs in all 7 genes

Time savings

- Streamline your laboratory workflow with a single NGS assay for a comprehensive profile of all variants of interest

Complimentary Interpret NGS data analysis software

- Easy-to-use analysis solution for accurate detection of all variants in your panel

SureSeq

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Introduction

The SureSeq™ Breast Cancer + CNV Panel has been developed to provide comprehensive coverage of 7 key genes implicated in breast and ovarian cancer, including *BRCA1* and *BRCA2* (Table 1). Detecting SNVs and indels, as well as exon-level to whole gene CNVs, the SureSeq Breast Cancer + CNV Panel provides researchers with a single NGS workflow to study clinically relevant aberrations and alleviates the burden of running multiple assays.

ATM	*BRCA1*	*BRCA2*	*TP53*	*CHEK2*	*PALB2*	*PTEN*

Table 1: The SureSeq Breast Cancer + CNV panel targets all exons in 7 key genes implicated in breast and ovarian cancer, detecting SNVs and indels, as well as CNVs.

Evidence-based content and unparalleled coverage uniformity

Loss-of-function mutations in *BRCA1* and *BRCA2* have been implicated in an increased risk for breast and ovarian cancer[1,2]. Screening for germline mutations in these genes allows research into familial risk of developing breast and ovarian cancer. Facilitated by OGT's expert bait design, the hybridisation-based SureSeq Breast Cancer + CNV Panel delivers excellent coverage uniformity, allowing consistent detection of SNVs and indels (Figure 1).

Figure 1: Illustration of the excellent coverage uniformity across *BRCA1* and *BRCA2*. A *BRCA1* exons 9, 10 and 11 coverage and B *BRCA2* exons 9, 10 and 11. Depth of coverage per base (grey). Gene coding region as defined by RefSeq (blue).

Reliable CNV detection

To gain a comprehensive picture of breast and ovarian cancer, researchers often have to employ different methods for investigating SNVs, indels, and CNVs. The SureSeq Breast Cancer + CNV Panel offers reliable CNV detection in all genes covered by the panel, ranging from single-exon events up to deletions and duplications of complete genes. The panel has been fully validated on germline samples, with CNV detection 100% concordant with MLPA data, providing researchers with a single NGS assay for profiling of CNVs in *BRCA1*, *BRCA2* and 5 other key genes implicated in breast and ovarian cancer (Figures 2 - 3).

Figure 2: Detection of germline deletions in *BRCA1* and *BRCA2*. A *BRCA1* deletion of exon 20 (4.99kb) and B *BRCA2* deletion of exons 14-17 (4.21kb).

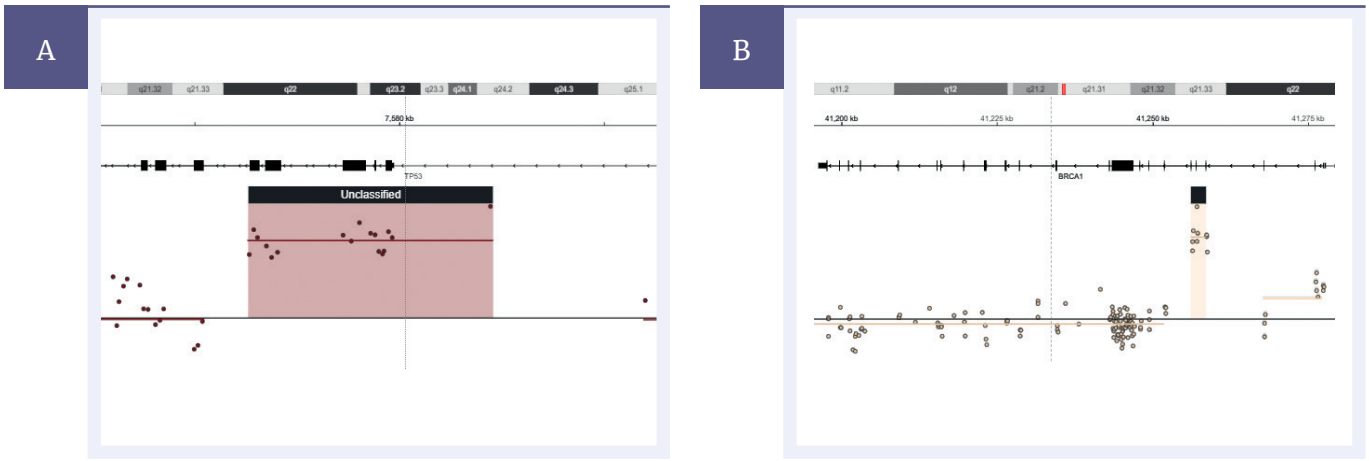

Figure 3: Detection of germline duplications in *TP53* and *BRCA1*. A *TP53* duplication of exons 2-6 (2.99kb) and B *BRCA1* duplication of exons 4-6 (2.45kb).

Complimentary Interpret NGS analysis software

Interpret is OGT's powerful and easy-to-use NGS analysis solution, facilitating analysis and visualisation of a wide range of somatic variants and structural aberrations. Designed to work seamlessly with all SureSeq panels, Interpret perfectly complements the SureSeq Breast Cancer + CNV Panel, delivering fast and accurate detection of all SNVs, indels and CNVs covered by the panel. Following detection, all variants can be easily visualised in the user-friendly variant browser, for an effortless translation of all your sequencing data into meaningful results.

Bespoke panel content

You never have to sequence genes you're not interested in and can always modify each panel to what's relevant to your research. If the SureSeq Breast Cancer + CNV Panel doesn't meet your exact requirements, you can choose from our regularly updated, expert-curated library of pre-optimised cancer content to create your ideal custom SureSeq myPanel™ Breast and/or Ovarian Cancer Panel.

The SureSeq Breast Cancer + CNV Panel in numbers

Feature	Specification
Number of genes	7
Panel size	52.6kb
Mean target coverage	>1000x
Coverage uniformity	>99% of bases at >20% of mean target coverage
DNA input recommended	>250ng high quality DNA
Samples per MiSeq® v2 run	24 samples / run

Ordering information

Product	Contents	Cat. No.
SureSeq Breast Cancer + CNV Panel (16 reactions)	Enrichment baits sufficient for 16 samples; Interpret Software	602028-16
SureSeq Breast Cancer + CNV Panel (96 reactions)	Enrichment baits sufficient for 96 samples; Interpret Software	602028-96
SureSeq NGS Library Preparation Complete Solution (16)	Bundle of 1x SureSeq NGS Library Preparation Kit (16), containing adaptors, PCR primers and enzymes, 1x SureSeq NGS Index Kit – Collection A, 1x SureSeq NGS Hyb & Wash Kit (16), 1x Dynabeads M270 Streptavidin (2ml) and 1x AMPure XP beads (10ml). Sufficient for 16 samples	500084
SureSeq NGS Library Preparation Complete Solution (48)	Bundle of 1x SureSeq NGS Library Preparation Kit (48), containing adaptors, PCR primers and enzymes, 1x SureSeq NGS Index Kit – Collection B, 3x SureSeq NGS Hyb & Wash Kit (16), 3x Dynabeads M270 Streptavidin (2ml) and 3x AMPure XP beads (10ml). Sufficient for 48 samples	500085

Ordering information

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References

1. Antoniou *et al.*, Am J Hum Genet. 2003 Sep;73(3):709.
2. King *et al.*, Science. 2003;302:643–646

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What binds us, makes us.

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