SureSeq

A Sysmex Group Company

Germline 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

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Introduction

The SureSeq™ Germline 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 Germline 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 Germline 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, direct from blood samples. Facilitated by OGT's expert bait design, the hybridisation-based SureSeq Germline 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 Germline 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 gene, direct from blood samples. 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).

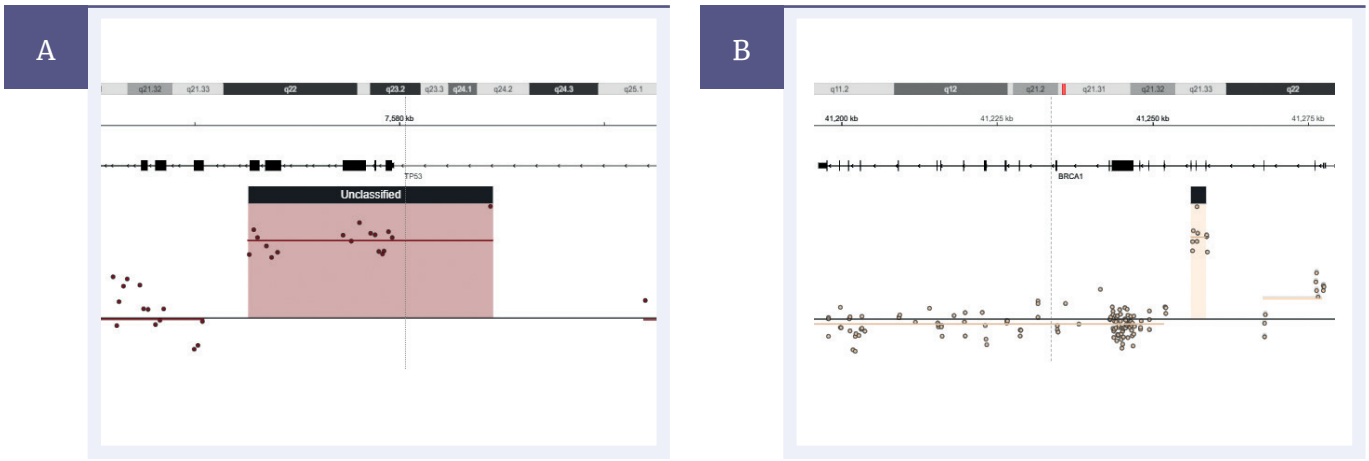

Figure 3: Detection of germline duplications in *TP53* and *BRCA1*. A *TP53* duplication of exons 2-6 (2.99kb) and B *BRCA1* duplication of exons 4-6 (2.45kb).

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 Germline 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.

The OGT Partnership

Behind every sample is a life that can be improved through the right care decisions. The OGT partnership approach is key to providing the highest level of service, working closely with you to understand your unique challenges, customising our approach to meet your exact needs.

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 Germline 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 Germline Breast Cancer + CNV Panel in numbers

Feature	Specification
Number of genes	7
Panel size	52.6kb
DNA input recommended	>250ng high quality DNA

Ordering information

Product	Contents	Cat. No.
SureSeq Germline Breast Cancer + CNV Complete NGS Workflow Solution V2 (24)	Enrichment baits sufficient for 3 x 8-sample pools. Bundle of 1 x Universal Library Preparation Kit (24) containing PCR primers and enzymes. 1 x Universal Hybridisation & Wash Kit V2 (24). 1 x Pre-PCR Universal Bead Kit (24). 1 x Post-PCR Universal Bead Kit (24). 1 x Universal Index Adapter Kit (24). Interpret NGS Analysis Software	780105-24
SureSeq Germline Breast Cancer + CNV Complete NGS Workflow Solution V2 (96)	Enrichment baits sufficient for 12 x 8-sample pools. Bundle of 1 x Universal Library Preparation Kit (96) containing PCR primers and enzymes. 1 x Universal Hybridisation & Wash Kit V2 (96). 1 x Pre-PCR Universal Bead Kit (96). 1 x Post-PCR Universal Bead Kit (96). 1 x Universal Index Adapter Kit (96). Interpret NGS Analysis Software	780105-96
SureSeq Germline Breast Cancer + CNV Panel (24)	Enrichment baits sufficient for 3 x 8-sample pools. Interpret NGS Analysis Software.	770005-24
SureSeq Germline Breast Cancer + CNV Panel (96)	Enrichment baits sufficient for 12 x 8-sample pools. Interpret NGS Analysis Software.	770005-96
Universal NGS Workflow Solution V2 (24)	Bundle of 1 x Universal Library Preparation Kit (24) containing PCR primers and enzymes. 1 x Universal Hybridisation & Wash Kit V2 (24). 1 x Pre-PCR Universal Bead Kit (24). 1 x Post-PCR Universal Bead Kit (24). 1 x Universal Index Adapter Kit (24)	770510-24
Universal NGS Workflow Solution V2 (96)	Bundle of 1 x Universal Library Preparation Kit (96), containing PCR primers and enzymes. 1 x Universal Hybridisation & Wash Kit V2 (96). 1 x Pre-PCR Universal Bead Kit (96). 1 x Post-PCR Universal Bead Kit (96). 1 x Universal Index Adapter Kit (96)	770510-96

Ordering information

UK +44 (0) 1865 856800

US +1 914 467 5285

contact@ogt.com

ogt.com

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.

Oxford Gene Technology Ltd., Begbroke Science Park, Woodstock Road, Begbroke, Oxfordshire, OX5 1PF, UK

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