Breast and Ovarian Cancer
Germline and Tumour testing

High confidence in **copy number determination** with **MLPA** and **digitalMLPA** to support comprehensive **BRCA & HR deficiency testing**

Genetic variants in *BRCA1*, *BRCA2*, *PALB2* and other cancer-associated genes, particularly those involved in the homologous recombination (HR) repair pathway, can give rise to cancer and influence therapeutic decisions. Both copy number variations (CNVs) and promoter methylation, especially of *BRCA1* and *RAD51C*, are emerging biomarkers for guiding clinical interventions, including the use of PARP inhibitors.

MRC Holland offers over 20 assays specifically designed for the detection of these genetic alterations, enabling the study of predisposition to breast and ovarian cancer and tumour profiles in these cancer types. These assays are based on the well-established **SALSA® MLPA®** technology and the innovative technique **digitalMLPA™**.

- Reliable CNV detection
- Multiple targets per reaction
- High-throughput solutions
- Simple data analysis

Germline analysis

digitalMLPA assay	Target genes/regions
NXtec™ D001 Hereditary Cancer Panel 1 582 probes targeting 28 genes associated with a predisposition to 8 common hereditary cancer types, and 5 variants	*APC, ATM, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, MITF* (p.E318K), *MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, POLE, PTEN, RAD51C, RAD51D, SCG5/GREM1, SMAD4, STK11, TP53*
NXtec™ D002 Hereditary Cancer Panel 2 1031 probes targeting 56 genes associated with a predisposition to a wide range of hereditary cancer types, and 8 variants	All D001 targets + *CEBPA, DICER1, FH, FLCN, HOXB13, MAX, MEN1, MET, NF1, NF2, NTHL1, PHOX2B, POLD1, PTCH1, RB1, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCB1, SUFU, TMEM127, TSC1, TSC2, VHL, WT1*

SALSA® MLPA® Probemix	Target genes/regions
P002 BRCA1	*BRCA1*
P087 BRCA1 Confirmation	*BRCA1*
P239 BRCA1 region	*BRCA1* upstream and downstream region, including *BRCA1P1*
P090 BRCA2	*BRCA2*
P045 BRCA2/CHEK2	*BRCA2, CHEK2* (+c.del1100C)
P077 BRCA2 Confirmation	*BRCA2*
P190 CHEK2	*CHEK2* (+c.del1100C), *ATM, TP53*
P240 BRIP1/CHEK1	*BRIP1, CHEK1*
P260 PALB2-RAD50-RAD51C-RAD51D	*PALB2, RAD50, RAD51C, RAD51D*
P057 FANCD2-PALB2	*FANCD2, PALB2*
P041 ATM-1	*ATM*
P042 ATM-2	*ATM*
P083 CDH1	*CDH1*
P101 STK11	*STK11*
P056 TP53	*TP53, CHEK2* (+c.del1100C)
P225 PTEN	*PTEN, PTENP1*
P489 BARD1	*BARD1*
P494 NBN	*NBN*
P003 MLH1/MSH2	*MLH1, MSH2, EPCAM*
P248 MLH1-MSH2 Confirmation	*MLH1, MSH2*
P072 MSH6-MUTYH	*MSH6, MUTYH, MSH2, EPCAM*
P008 PMS2	*PMS2, PMS2CL*

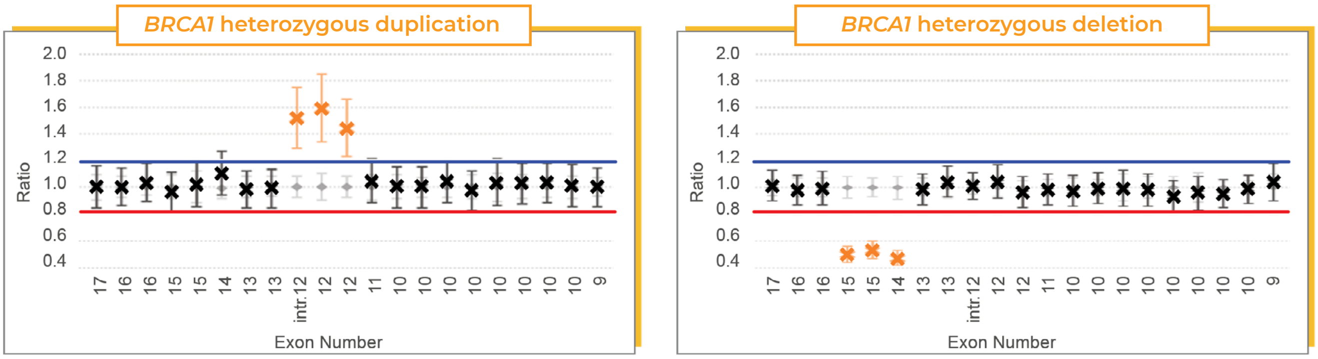
Tumour analysis

SALSA® MLPA® Probemix	Target genes/regions
P078 Breast tumour	6q (*ESR1*), 7p (*EGFR*), 8p (*ZNF703, FGFR1, IKBKB*), 8q (*MTDH, MYC*), 11q13 (*CCND1, EMSY*), 16q (*CDH1*), 17q12-q25 (*ERBB2, TOP2A, BIRC5*), 19q (*CCNE1*), 20q (*AURKA*)
P483 HER gene family	*EGFR, ERBB2, ERBB3, ERBB4*
P056 TP53	*TP53, CHEK2* (+c.del1100C)
P083 CDH1	*CDH1*
P225 PTEN	*PTEN, PTENP1*
ME053 BRCA1-BRCA2-RAD51C	*BRCA1, BRCA2, RAD51C* methylation profiling
ME011 Mismatch Repair Genes	*MLH1, MSH6, PMS2* methylation, *BRAF* p.V600E point mutation and *EPCAM* copy number profiling
ME001 Tumour suppressor mix	Methylation profiling of tumour suppressor genes

digitalMLPA™

High-throughput analysis of copy number status

- ✓ NGS-based MLPA for the detection of CNVs and targeted variants
- ✓ Only requires a thermocycler and Illumina sequencing platform
- ✓ Up to 1600 targets per reaction
- ✓ 20 ng of DNA input per sample
- ✓ Free analysis software: **Coffalyser digitalMLPA™**


BRCA1 heterozygous duplication

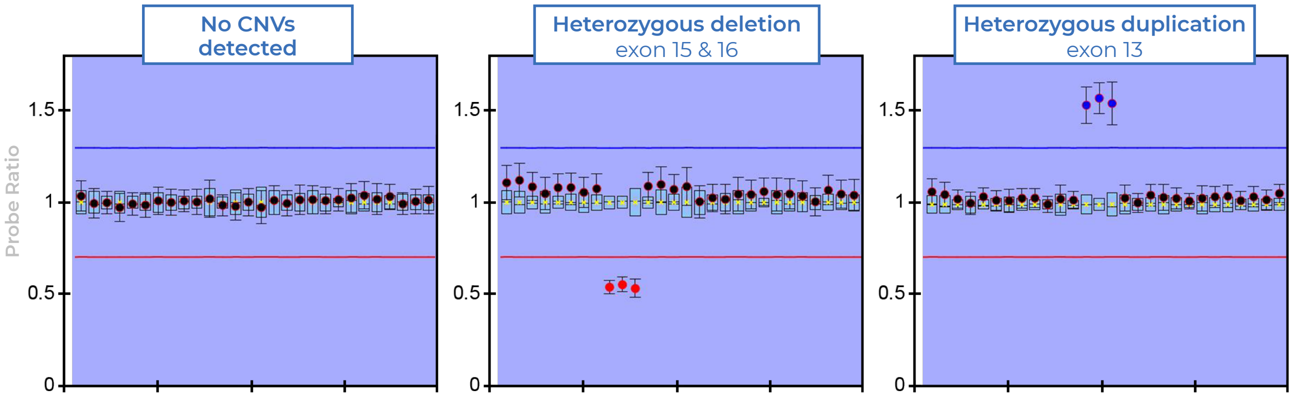
BRCA1 heterozygous deletion

Figure 1. Representation of copy number results using NXtec D002 Hereditary Cancer Panel 2 and Coffalyser digitalMLPA. Probe Ratios sorted by genomic location. A selection of data points is shown.

SALSA® MLPA®

The go-to technique for studying CNVs associated with disease

- ✓ Multiplex PCR-based method for CNV and targeted variants detection
- ✓ Only requires a thermocycler and capillary electrophoresis device
- ✓ 40-60 targets per reaction
- ✓ 50 ng of DNA input per sample
- ✓ Free analysis software: **Coffalyser.Net™**


No CNVs detected

Heterozygous deletion exon 15 & 16

Heterozygous duplication exon 13

Figure 2. Representation of *BRCA1* copy number results using SALSA® MLPA® Probemix P002 BRCA1 and Coffalyser.Net. Probe Ratios sorted by genomic location. A selection of data, from *BRCA1* exon 24 (left) to exon 7 (right), is shown.

MRC Holland

Confidence in Copy Number Determination

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