



SALSA® MLPA® is the method of choice for the detection of CNVs in genes associated with hereditary cancer. MRC Holland offers over 40 MLPA assays specifically designed to detect CNVs and select SNVs in hundreds of oncogenes.

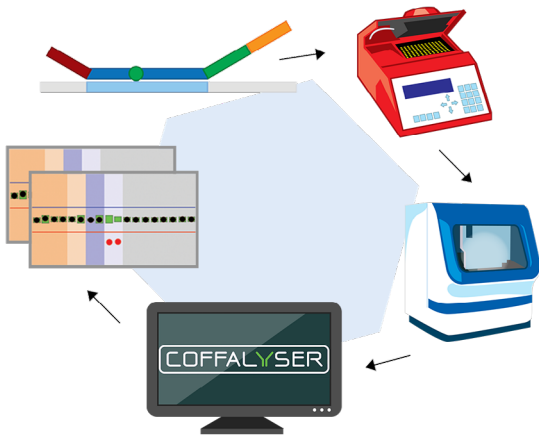
SALSA® digitalMLPA™, a more recent adaptation of the MLPA technology, combines the robustness and simplicity of MLPA with the high throughput of NGS platforms. Our SALSA® digitalMLPA Probemix D001 Hereditary Cancer Panel 1 is a useful tool to examine CNVs and targeted SNVs in 28 of the most common cancer-predisposing genes.

Both technologies share some common features that make them well-suited for germline analysis:

- Unparalleled CNV sensitivity even in complex genetic regions, such as *PMS2* and *PTEN*
- Wide CNV detection range – from whole chromosomes to single exons.
- Free software, simple analysis and clear-cut results.

## MLPA

- ✓ Multiplex PCR-based method for CNV, methylation and targeted mutation detection
- ✓ Needed: thermocycler and capillary electrophoresis device
- ✓ 40-60 targets per reaction
- ✓ 50 ng of DNA input per sample

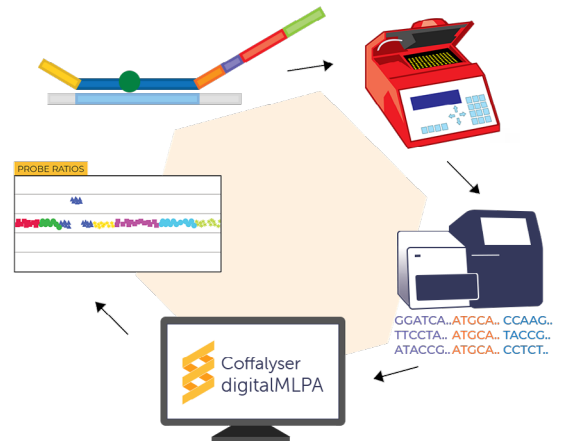


### Top-Selling Applications

<b>Hereditary Breast and Ovarian Cancer (HBOC)</b>	P002 BRCA1 P045 BRCA2-CHEK2 P041 ATM-1 / P042 ATM-2
<b>Lynch Syndrome</b>	P003 MLH1/MSH2 P008 PMS2 P072 MSH6-MUTYH
<b>Neurofibromatosis</b>	P081 NF1 mix 1 / P082 NF1 mix 2 P122 NF1-area P044 NF2

## digitalMLPA

- ✓ NGS-based MLPA for CNV, methylation and targeted mutation detection
- ✓ Needed: thermocycler and Illumina sequencing platform
- ✓ 600-1000 targets per reaction
- ✓ 20 ng of DNA input per sample



### D001 Hereditary Cancer Panel 1

APC	BRIP1	MSH2	PTEN
ATM	CDH1	MSH6	RAD51C
BAP1	CDK4	MUTYH	RAD51D
BARD1	CDKN2A	NBN	SCG5/GREM1
BMPRIA	CHEK2	PALB2	SMAD4
BRCA1	EPCAM	PMS2	STK11
BRCA2	MLH1	POLE	TP53

## Broad Hereditary Cancer Panels

SALSA® digitalMLPA™ Probemix	Target genes/regions
D001 Hereditary Cancer Panel 1	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

## Breast, Endometrial and Ovarian Cancer

SALSA® MLPA® Probemix	Target genes/regions
ME011 Mismatch Repair Genes*	Methylation profiling of MLH1, MSH2, MSH6, PMS2; EPCAM; BRAF (p.V600E)
P002 BRCA1†	BRCA1
P003 MLH1/MSH2†	MLH1, MSH2
P008 PMS2	PMS2
P041 ATM-1 / P042 ATM-2	ATM
P045 BRCA2/CHEK2†	BRCA2, CHEK2
P057 FANCD2-PALB2	FANCD2, PALB2
P072 MSH6-MUTYH	MSH6, MUTYH, MSH2, EPCAM
P083 CDH1	CDH1
P090 BRCA2†	BRCA2
P190 CHEK2	CHEK2, ATM, TP53
P225 PTEN	PTEN, PTENP1
P239 BRCA1 region	BRCA1 region
P240 BRIP1/CHEK1	BRIP1, CHEK1
P260 PALB2-RAD50-RAD51C-RAD51D	PALB2, RAD50, RAD51C, RAD51D
P489 BARD1	BARD1
P494 NBN	NBN

## Skin Cancer

SALSA® MLPA® Probemix	Target genes/regions
ME024 9p21 CDKN2A/2B region*	CDKN2A/2B, MTAP, MIR31, PAX5 and methylation profiling of CDKN2A, CDKN2B
P041 ATM-1 / P042 ATM-2	ATM
P417 BAP1	BAP1
P419 CDKN2A/2B-CDK4	CDKN2A/2B, CDK4, MITF (p.E318K)
P472 SUFU	SUFU

## Pancreatic Cancer

SALSA® MLPA® Probemix	Target genes/regions
ME024 9p21 CDKN2A/2B region*	CDKN2A/2B, MTAP, MIR31, PAX5 and methylation profiling of CDKN2A, CDKN2B
P003 MLH1/MSH2†	MLH1, MSH2
P045 BRCA2/CHEK2†	BRCA2, CHEK2
P090 BRCA2†	BRCA2
P419 CDKN2A/2B-CDK4	CDKN2A/2B, CDK4, MITF (p.E318K)

## Hematological Malignancies

SALSA® MLPA® Probemix	Target genes/regions
P031 FANCA mix 1 P032 FANCA mix 2	FANCA
P057 FANCD2-PALB2	FANCD2, PALB2
P113 FANCB	FANCB
P212 DBA	RPL5, RPL11, RPL35A, RPS17, RPS19, RPS26
P327 iAMP21-ERG	iAMP21, RUNX1, ERG
P437 Familial MDS-AML	GATA2 (+p.R398W, p.T354M), TERC, TERT (+p.A1062T), CEBPA, RUNX1

\* For this probemix, a separate MLPA confirmation probemix exists. A confirmation probemix contains independent probes recognising different targets in the same exons/genes as the primary probemix.

## Broad Cancer-Predisposing Syndromes

SALSA® MLPA® Probemix	Target genes/regions
ME011 Mismatch Repair Genes*	Methylation profiling of MLH1, MSH2, MSH6, PMS2; EPCAM; BRAF (p.V600E)
P016 VHL	VHL
P017 MEN1	MEN1
P044 NF2	NF2
P046 TSC2†	TSC2
P056 TP53	TP53, CHEK2 (+c.del1100C)
P057 FANCD2-PALB2	FANCD2, PALB2
P067 PTCH1	PTCH1
P081 NF1 mix 1 / P082 NF1 mix 2	NF1
P101 STK11	STK11
P118 WT1	WT1, AMER1
P122 NF1-area	NF1-area
P124 TSC1	TSC1
P215 EXT	EXT1, EXT2
P225 PTEN	PTEN, PTENP1
P226 SDH	SDHB, SDHC, SDHD, SDHAF1, SDHAF2
P244 AIP-MEN1-CDKN1B	AIP, MEN1, CDKN1B
P258 SMARCB1	SMARCB1
P308 MET	MET, PTEN, LRRK2
P417 BAP1	BAP1
P429 SDHA-MAX-TMEM127	SDHA, MAX, TMEM127
P437 Familial MDS-AML	GATA2 (+p.R398W, p.T354M), TERC, TERT (+p.A1062T), CEBPA, RUNX1
P455 LZTR1	LZTR1
P466 CDC73	CDC73
P478 SMARCE1	SMARCE1
P481 PRKAR1A-ARMC5	PRKAR1A, ARM5
P482 DICER1	DICER1

## Gastrointestinal Cancer

SALSA® MLPA® Probemix	Target genes/regions
ME011 Mismatch Repair Genes*	Methylation profiling of MLH1, MSH2, MSH6, PMS2; EPCAM; BRAF (p.V600E)
P003 MLH1/MSH2†	MLH1, MSH2
P008 PMS2	PMS2
P043 APC	APC
P072 MSH6-MUTYH	MSH6, MUTYH, MSH2, EPCAM
P083 CDH1	CDH1
P158 JPS	BMPR1A, SMAD4, PTEN
P190 CHEK2	CHEK2, ATM, TP53
P378 MUTYH	MUTYH, SCG5/GREM1
P492 POLD1-POLE	POLD1, POLE

## Other Cancers

SALSA® MLPA® Probemix	Target genes/regions
P003 MLH1/MSH2†	MLH1, MSH2
P047 RB1*	RB1
P067 PTCH1	PTCH1
P256 FLCN	FLCN
P472 SUFU	SUFU

\* This probemix also includes probes that determine the methylation status of a target. + Copy number detection for the whole gene and additional point mutation(s).