MRC Holland

SALSA® digitalMLPA D001 Hereditary Cancer Panel 1

For Research Use Only. Not for Use in Diagnostic Procedures.

- ✓ **Reliable:** CNV detection with robust quality checks
- ✓ **Comprehensive:** extensive probe coverage of 28 clinically relevant cancer genes
- ✓ **Efficient:** decreased sample turnaround time by reducing MLPA runs
- ✓ **Specific:** CNV certainty with reduced incidental findings

SALSA® digitalMLPA D001 Hereditary Cancer Panel 1 can detect copy number variations (CNV) and 6 common mutations in **28** clinically relevant genes. CNVs in these genes are associated with hereditary predisposition to one or more of the following cancer types: breast, ovarian, colorectal, gastric, prostate, pancreatic, endometrial, and melanoma.

DNA Denaturation → Hybridisation → Ligation → Amplification (PCR) → Sequencing (Illumina Platform) → Data Analysis (Coffalyser digitalMLPA)

D001 Hereditary Cancer Panel 1 is the perfect time-saving complement to NGS sequencing for high-level CNV calling certainty. digitalMLPA is highly multiplexable, where digitalMLPA reactions and NGS libraries can be run on the same chip. This enables simultaneous CNV quantification and NGS sequence analysis. D001 Hereditary Cancer Panel 1 can replace the multiple conventional SALSA® MLPA® experiments often run on NGS-negative samples, saving time and money.

Genes covered in D001				
APC	*BRCA2*	*EPCAM*	*MUTYH*	*RAD51C*
ATM	*BRIP1*	*GREM1*	*NBN*	*RAD51D*
BAP1	*CDH1*	*MITF**	*PALB2*	*SCG5*
BARD1	*CDK4*	*MLH1*	*PMS2*	*SMAD4*
BMPR1A	*CDKN2A*	*MSH2*	*POLE**	*STK11*
BRCA1	*CHEK2*	*MSH6*	*PTEN*	*TP53*

* One probe present in D001 Hereditary Cancer Panel 1.

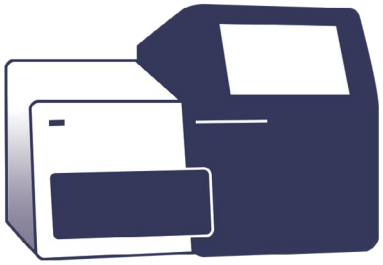
Mutation-specific probes in D001	
Gene	**Variant**
BRCA2	c.156_157insAlu exon 3⁺
CHEK2	c.1100delC
MITF	p.E318K
MSH2	10 Mb inversion (2 probes)
PMS2	Intron 7 SVA element insertion (2⁺ probes)
POLE	c.1270C>G⁺

⁺ One probe recognises the wild type sequence for the listed mutation.

Data analysis for count-based quantification is done using free software (Coffalyser digitalMLPA). Coffalyser digitalMLPA returns two reports for easy reaction quality determination and result interpretation.

Confirmation by conventional SALSA® MLPA® is possible for many of the genes present in D001 Hereditary Cancer Panel 1.

Probemix	Number of genes	Number of probes	Sample source
D001 Hereditary Cancer Panel 1	28	566	DNA derived from peripheral blood.



Breast	Ovarian	Colorectal	Gastric	Prostate	Pancreatic	Endometrial	Melanoma
ATM	*BRCA1*	*APC*	*APC*	*BRCA1*	*BRCA1*	*EPCAM*	*BAP1*
BRCA1	*BRCA2*	*BMPR1A*	*BMPR1A*	*BRCA2*	*BRCA2*	*MLH1*	*CDKN2A*
BRCA2	*BRIP1*	*CHEK2*	*CDH1*	*CHEK2*	*CDKN2A*	*MSH2*	*CDK4*
BARD1	*EPCAM*	*EPCAM*	*EPCAM*	*STK11*	*TP53*	*MSH6*	*MITF**
CDH1	*MLH1*	*MLH1*	*MLH1*	*TP53*		*PMS2*	*TP53*
CHEK2	*MSH2*	*MSH2*	*MSH2*			*PTEN*	
NBN	*MSH6*	*MSH6*	*MSH6*			*STK11*	
PALB2	*PMS2*	*MUTYH*	*MUTYH*			*TP53*	
PTEN	*RAD51C*	*PMS2*	*PMS2*				
STK11	*RAD51D*	*POLE**	*SMAD4*				
TP53	*STK11*	*SMAD4*	*STK11*				
	TP53	*SCG5/ GREM1*	*TP53*				
		STK11					
		TP53					

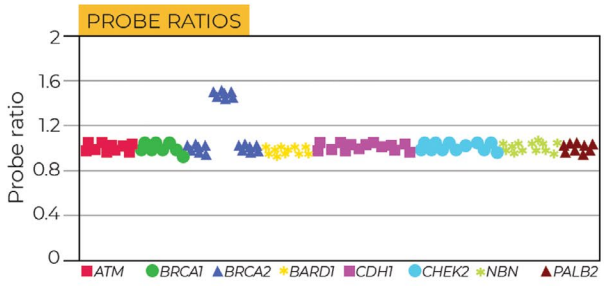
Coffalyser digitalMLPA: D001 Hereditary Cancer Panel 1

Data analysis software for clear CNV calling

- ✓ **Complimentary:** no additional costs for data analysis software
- ✓ **Simple:** FASTQ files are directly loaded into the software
- ✓ **Smart:** automatic digitalMLPA read and probemix recognition
- ✓ **Easy:** run by any user on a personal Windows 10-based computer
- ✓ **Reliable:** extensively tested and validated
- ✓ **Safe:** extensive built-in quality control by analysis of >120 control probes

Coffalyser digitalMLPA

QUALITY CHECKS	
Reaction analysis	✔ ✔ ✔ ✔ ✔
Comparative analysis	✔ ✔ ✔ ✔



Coffalyser digitalMLPA is free software developed and supported by MRC Holland for the analysis of digitalMLPA data. Coffalyser digitalMLPA automatically recognises and extracts digitalMLPA sequence reads from FASTQ files for direct use by the software. The software performs advanced data quality checks and returns a clear report with all identified aberrant regions.

* One probe included in D001 Hereditary Cancer Panel 1.

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