SALSA® digitalMLPA™ Probemix D001 Hereditary Cancer Panel 1

- ✓ **Reliable:** CNV detection with robust quality checks
- ✓ **Comprehensive:** 584 probes targeting 28 clinically relevant oncogenes
- ✓ **Efficient:** decreased sample turnaround time by reducing SALSA® MLPA® runs
- ✓ **Specific:** CNV certainty with reduced incidental findings

SALSA® digitalMLPA™ Probemix D001 Hereditary Cancer Panel 1 can detect copy number variation (CNV) in 28 clinically relevant oncogenes. 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.

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					

D001 Hereditary Cancer Panel 1 is the perfect time-saving complement to NGS sequencing. SALSA® digitalMLPA™ ensures a higher level of confidence in CNV calling than using NGS alone, and allows for multiplexing as digitalMLPA reactions and NGS libraries can be run on the same flow cell.

Analysis is done using free, easy-to-use software – so no bioinformatic skills are needed.

* One *MITF* probe included; specific for p.E318K mutation.

D001 Hereditary Cancer Panel 1 Targets

Besides CNV detection, D001 Hereditary Cancer Panel 1 also allows for the detection of 4 common mutations in clinically relevant oncogenes.

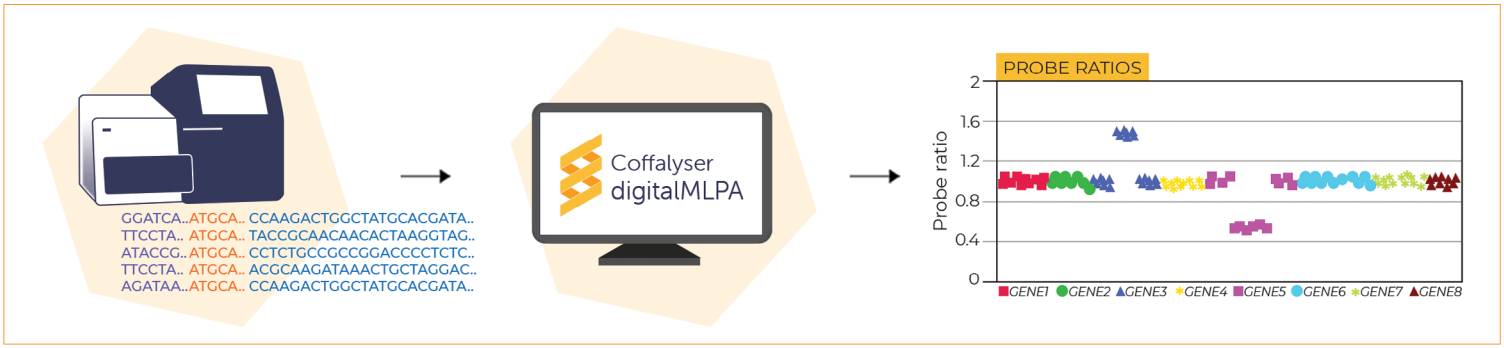
Targeted genes for CNV			
APC	*BRIP1*	*MSH2*	*PTEN*
ATM	*CDH1*	*MSH6*	*RAD51C*
BAP1	*CDK4*	*MUTYH*	*RAD51D*
BARD1	*CDKN2A*	*NBN*	*SCG5/GREM1*
BMPR1A	*CHEK2*	*PALB2*	*SMAD4*
BRCA1	*EPCAM*	*PMS2*	*STK11*
BRCA2	*MLH1*	*POLE*	*TP53*

Targeted mutations	
Gene	**Variant**
CHEK2	c.1100delC
MITF	p.E318K
MSH2	10 Mb inversion
PMS2	Intron 7 SVA element insertion

Coffalyser digitalMLPA™
Data analysis software for clear CNV calling

- ✓ **Simple:** FASTQ files are directly loaded into the software
- ✓ **Smart:** automatic digitalMLPA read and probemix recognition
- ✓ **Reliable:** extensively tested and validated
- ✓ **Safe:** thorough built-in quality control

Coffalyser digitalMLPA



Coffalyser digitalMLPA™ is free and easy-to-use software developed by MRC Holland and built specifically for the analysis of digitalMLPA data. The software automatically recognises and extracts digitalMLPA sequence reads from FASTQ files. This is followed by advanced data quality checks, and the return of a clear report displaying all detected aberrant regions.

MRC Holland

Confidence in Copy Number Determination

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