digitalMLPA™ NXtec™

# D001 Hereditary Cancer Panel 1

MRC Holland

- ✓ **Reliable:** CNV detection with robust quality checks
- ✓ **Comprehensive:** 582 probes targeting 28 hereditary cancer-related genes and 5 variants
- ✓ **Efficient:** decreased sample turnaround time by reducing SALSA® MLPA® runs
- ✓ **Specific:** CNV certainty with reduced incidental findings

digitalMLPA assay NXtec D001 Hereditary Cancer Panel 1 can detect copy number variation (CNV) in 28 genes associated with a hereditary predisposition to one or more of the following cancer types: breast, ovarian, colorectal, gastric, prostate, pancreatic, endometrial, and melanoma.

This digitalMLPA assay is the perfect time-saving complement to NGS sequencing. digitalMLPA ensures a higher level of confidence in CNV calling than using NGS alone, and analysis is done using free, easy-to-use software – so no bioinformatic skills are needed.

| 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* | | | | | |


* One *MITF* probe included; specific for c.952G>A mutation.

## NXtec D001 Hereditary Cancer Panel 1 Targets

Besides CNV detection, NXtec D001 Hereditary Cancer Panel 1 also allows for the detection of 5 common variants in hereditary cancer-related genes.

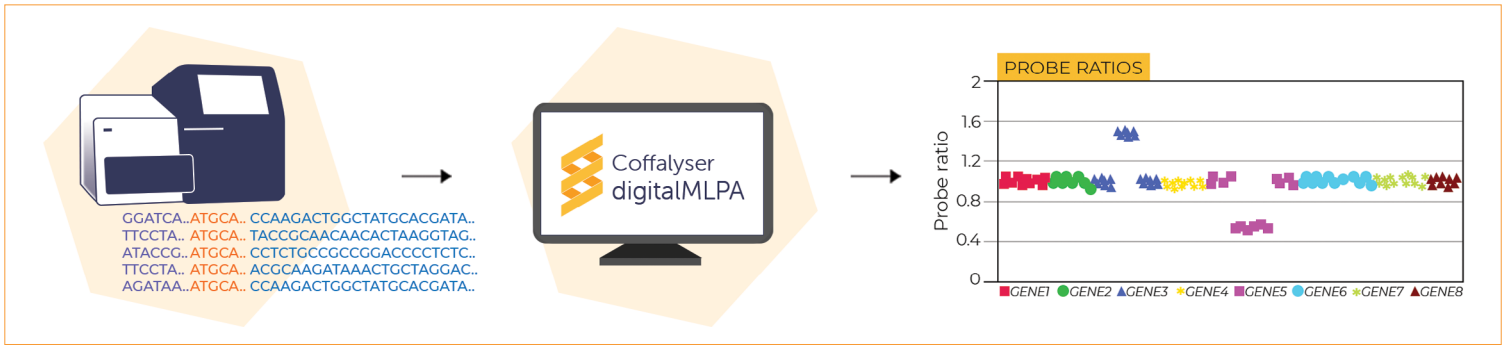
| Targeted genes for CNV | | | |
|---|---|---|---|
| *APC* | *BRIP1* | *MSH2* | *PTEN* |
| *ATM* | *CDH1* | *MSH6* | *RAD51C* |
| *BAP1* | *CDK4* | *MUTYH* | *RAD51D* |
| *BARD1* | *CDKN2A* | *NBN* | *SCG5/GREM1* |
| *BMPR1A* | *CHEK2* | *PALB2* | *SMAD4* |
| *BRCA1* | *EPCAM* | *PMS2* | *STK11* |
|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

| Targeted variants |
|---|
| *CHEK2* c.1100delC |
| *MITF* c.952G>A |
| *MSH2* Exon 1-7 inversion (PMID 24114314) |
| *MSH2* Exon 2-6 inversion (PMID 26498247) |
| *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 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.