digitalMLPA™ NXtec™

Hereditary Cancer Panels

MRC Holland

Unparalleled CNV detection built into your NGS workflow

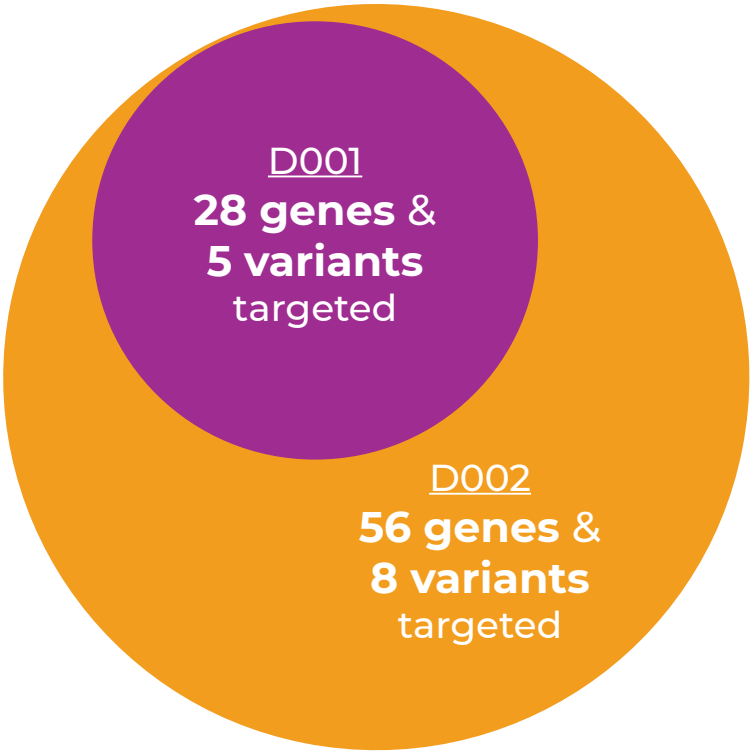
Many NGS-based techniques struggle to reliably detect CNVs, leaving critical gaps in cancer risk assessment. NXtec Hereditary Cancer Panels offer a powerful solution—providing high-resolution CNV detection with unmatched certainty, even in complex genomic regions.

digitalMLPA integrates into your existing NGS workflow—bringing clarity and confidence to your investigation into hereditary cancer risk across a wide range of cancer types.

Key Features

- High-confidence CNV detection for hereditary cancer testing
- Quick turnaround time - from DNA to sequencer in <24 hours
- Reduced chance of incidental findings
- Combine digitalMLPA with other libraries to maximise sequencing capacity
- Minimal DNA input requirements – 20 ng of DNA is sufficient
- Simple analysis with free software – no bioinformatics expertise needed

Available Panels



NXtec D001 Hereditary Cancer Panel 1

- Focused, cost-effective panel
- Targeting 28 key genes and 5 variants associated with a hereditary predisposition to breast, ovarian, colorectal, gastric, prostate, pancreatic and endometrial cancer, and melanoma

NXtec D002 Hereditary Cancer Panel 2

- Broad, expanded panel
- Coverage includes all D001 targets and additional, D002-exclusive targets
- Targeting 56 genes and 8 variants associated with a hereditary predisposition to a wide range of cancer types, including all those mentioned in D001, neurofibromatoses, retinoblastoma, Wilms' tumour, and more

Panel Coverage

NXtec D002 Hereditary Cancer Panel 2

Targeting 56 genes (all D001 targets + additional D002-exclusive targets)

NXtec D001 Hereditary Cancer Panel 1

Targeting 28 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 (p.E318K)
MSH2 Exon 1-7 inversion (PMID 24114314)
MSH2 Exon 2-6 inversion (PMID 26498247)
PMS2 Intron 7 SVA element insertion

+

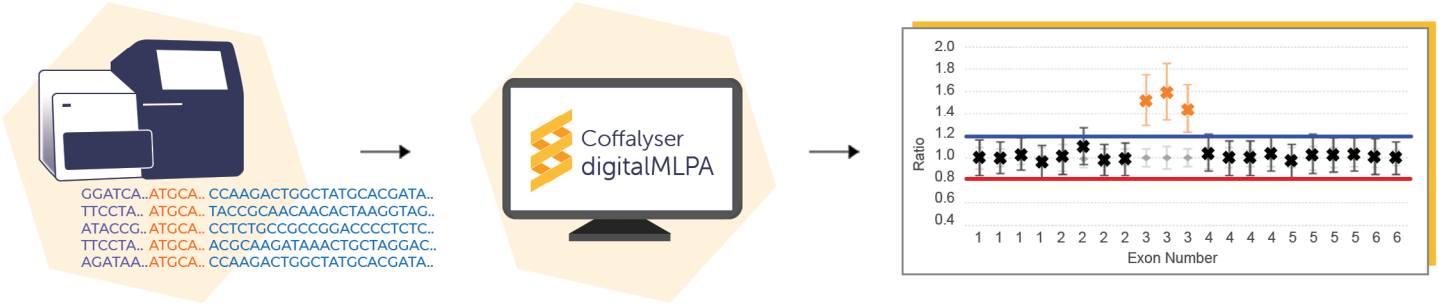
D002-exclusive targeted genes for CNV			
CEBPA	*MET*	*RB1*	*SMARCB1*
DICER1	*NF1*	*RUNX1*	*SUFU*
FH	*NF2*	*SDHA*	*TMEM127*
FLCN	*NTHL1*	*SDHAF2*	*TSC1*
HOXB13	*PHOX2B*	*SDHB*	*TSC2*
MAX	*POLD1*	*SDHC*	*VHL*
MEN1	*PTCH1*	*SDHD*	*WT1*

D002-exclusive targeted variants
FLCN c.1285dupC
FLCN c.1285delC
HOXB13 c.251G>A (p.G84E)

Data Analysis

Coffalyser digitalMLPA™

Free software that processes FASTQ files, runs QC, and delivers clear CNV reports



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Confidence in Copy Number Determination

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