

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



Reliable and specific CNV detection with a reduced chance of incidental findings



Quick turnaround time
- from DNA to sequencer in <24 hours



Minimal DNA input requirements
- 20 ng of DNA is sufficient



Simple analysis with Coffalyser digitalMLPA – no bioinformatics expertise needed

Available Panels

D001
**28 genes &
5 variants
targeted**

D002
**56 genes &
8 variants
targeted**

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, neurofibromatosis, retinoblastoma, Wilms' tumour, and more

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

<i>APC</i>	<i>BRIPI</i>	<i>MSH2</i>	<i>PTEN</i>
<i>ATM</i>	<i>CDH1</i>	<i>MSH6</i>	<i>RAD51C</i>
<i>BAP1</i>	<i>CDK4</i>	<i>MUTYH</i>	<i>RAD51D</i>
<i>BARD1</i>	<i>CDKN2A</i>	<i>NBN</i>	<i>SCG5/GREM1</i>
<i>BMPRI1A</i>	<i>CHEK2</i>	<i>PALB2</i>	<i>SMAD4</i>
<i>BRCA1</i>	<i>EPCAM</i>	<i>PMS2</i>	<i>STK11</i>
<i>BRCA2</i>	<i>MLH1</i>	<i>POLE</i>	<i>TP53</i>

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

<i>CEBPA</i>	<i>MET</i>	<i>RBI</i>	<i>SMARCB1</i>
<i>DICER1</i>	<i>NF1</i>	<i>RUNX1</i>	<i>SUFU</i>
<i>FH</i>	<i>NF2</i>	<i>SDHA</i>	<i>TMEM127</i>
<i>FLCN</i>	<i>NTHL1</i>	<i>SDHAF2</i>	<i>TSC1</i>
<i>HOXB13</i>	<i>PHOX2B</i>	<i>SDHB</i>	<i>TSC2</i>
<i>MAX</i>	<i>POLD1</i>	<i>SDHC</i>	<i>VHL</i>
<i>MEN1</i>	<i>PTCH1</i>	<i>SDHD</i>	<i>WT1</i>

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

