



Hereditary Plus

OncoKit DX

Analysis of predisposition to hereditary cancer.

For diagnostic use.

Format: 48 rx / Technology: NGS / Reference: IMG-399



> Characteristics

- Panel for the study of 101 genes related to some of the most frequent hereditary cancer types: breast, ovarian, colorectal, uterine, melanoma, kidney, prostate, pancreatic, multiple endocrine neoplasia (MEN), pheochromocytoma, paraganglioma, and retinoblastoma.
- Detection of SNVs, indels, ALUs, and CNVs.
- Bioinformatic analysis using the software Datagenomics.
- It can include STID (integrated sample identification system for traceability).
- Coverage: 99% of bases covered at 50X minimum depth.
- Uniformity: 98.8% of bases covered at >20% mean coverage.
- Specificity: > 99 %
- Sensitivity: > 99 %
- Repeatability: > 99 %
- Reproducibility: > 99 %

> Added value

- Technical support from first protocol implementation, both online and on-site. All protocols will be fine-tuned in collaboration with laboratory personnel.
- Clinical support or genetic counseling, with clinical geneticists available to answer any clinical questions.
- Possibility of externalizing 10% of the samples to our laboratory at the same price during periods of excess laboratory workload or urgent deadlines.

> Specifications

- **Compatible sequencers:** Illumina MiSeq and NextSeq.
- **Number of reactions:** 48.
- **Number of samples per run:**
 - MiSeq V2 300 cycles: 16 samples.
 - NextSeq Mid Output v2.5 kit (300 cycles): 32 samples.
- **Sequencing:** Paired-end (2 x 150 cycles)
- **Sample type:** DNA from peripheral blood and saliva.
- **Amount of input DNA:** 50 ng
- **Mean coverage:** >350X
- Fully automated panel for Magnis NGS Prep System Dx equipment.
- **Target genes:**

<i>ABRAXAS1 (FAM175A)</i>	<i>BRCA2</i>	<i>FANCC</i>	<i>MAX</i>	<i>NTHL1</i>	<i>RAD51C</i>	<i>SMARCB1</i>
<i>ACD</i>	<i>BRIPI</i>	<i>FANCG</i>	<i>MCIR</i>	<i>PALB2</i>	<i>RAD51D</i>	<i>SMARCE1</i>
<i>AIP</i>	<i>CDC73</i>	<i>FANCM</i>	<i>MEN1</i>	<i>PHOX2B</i>	<i>RB1</i>	<i>SPRED1</i>
<i>AKT1</i>	<i>CDH1</i>	<i>FH</i>	<i>MET</i>	<i>PIK3CA</i>	<i>RECQL4</i>	<i>STK11</i>
<i>ALK</i>	<i>CDK4</i>	<i>FLCN</i>	<i>MITF</i>	<i>PMS2</i>	<i>RET</i>	<i>SUFU</i>
<i>APC (incl. 5' UTR)</i>	<i>CDKN1B</i>	<i>GALNT12</i>	<i>MLH1</i>	<i>POLD1</i>	<i>RINT1</i>	<i>TERF2IP</i>
<i>ATM</i>	<i>CDKN2A</i>	<i>GALNT14</i>	<i>MLH3</i>	<i>POLE</i>	<i>SDHA</i>	<i>TERT</i>
<i>ATR</i>	<i>CHEK2</i>	<i>GDNF</i>	<i>MRE11A</i>	<i>POT1</i>	<i>SDHAF2</i>	<i>TMEM127</i>
<i>AXIN2</i>	<i>GEN1</i>	<i>MSH2</i>	<i>PRKARIA</i>	<i>SDHB</i>	<i>TP53</i>	
<i>BAP1</i>	<i>CHEK2</i>	<i>GEN1</i>	<i>MSH2</i>	<i>PRKARIA</i>	<i>SDHB</i>	<i>TP53</i>
<i>BARD1</i>	<i>CTNNA1</i>	<i>GREM1</i>	<i>MSH3</i>	<i>PRSS1</i>	<i>SDHC</i>	<i>TSC1</i>
<i>BLM</i>	<i>CYLD</i>	<i>HABP2</i>	<i>MSH6</i>	<i>PTCH1</i>	<i>SDHD</i>	<i>TSC2</i>
<i>BMPRIA</i>	<i>DICER1</i>	<i>HOXB13</i>	<i>MUTYH</i>	<i>PTEN</i>	<i>SEC23B</i>	<i>VHL</i>
<i>BRCA1</i>	<i>EPCAM (incl. 3' UTR)</i>	<i>KIF1B</i>	<i>NBN</i>	<i>RAD50</i>	<i>SLX4</i>	<i>WT1</i>
	<i>LZTR1</i>	<i>NF1</i>	<i>RAD51</i>	<i>SMAD4</i>	<i>XRCC2</i>	
		<i>NF2</i>	<i>RAD51B</i>	<i>SMARCA4</i>		

- Moreover, other **intergenic regions of interest** for the calculation of CNVs in genes *EPCAM* (3' UTR) and *MSH2* (5' UTR) have been included, as well as **50 intronic regions to cover hotspots** described in genes *APC*, *ATM*, *ATR*, *BRCA1*, *BRCA2*, *CHEK2*, *FH*, *LZTR1*, *MET*, *MLH1*, *MSH2*, *NF1*, *PMS2*, *PTEN*, *RB1*, *RET*, *SDHB*, *STK11* and *TERT*.
- CE/IVD and analysis software labeling.