

INSTRUCTIONS on digitalMLPA Data Demo Set

NXtec™ D002-A1 Hereditary Cancer Panel 2

1. Purpose

This digitalMLPA data demo set serves to provide an overview of possible results that can be obtained with NXtec™ D002-A1 Hereditary Cancer Panel 2. It contains data files of twelve samples with different genomic profiles and five reference samples. This data demo set also allows you to familiarise yourself with Coffalyser digitalMLPA™ and the way digitalMLPA results are presented by the software. The Sample Results PDF reports that are included in this demo set offer you the possibility to have a look at the sample results without analysing the data.

2. Description of samples included

Table 1 – Samples included in the D002-A1 data demo set

DNA Sample	Barcode used	Expected Results *
Coriell HG00187	BP03-01	22q: Positive for the <i>CHEK2</i> 1100delC mutation.
Coriell NA01359	BP03-02	18q: Trisomy chromosome 18 affecting the probes for <i>ELAC1</i> and <i>SMAD4</i> .
<i>REF-Male-1</i>	BP03-03	<i>Reference sample: normal ratios for all probes.</i>
<i>REF-Male-2</i>	BP03-06	<i>Reference sample: normal ratios for all probes.</i>
Coriell NA02718	BP03-07	13q: Heterozygous deletion affecting the probes for <i>ZAR1L</i> , <i>BRCA2</i> and <i>RB1</i> .
Coriell NA03184	BP03-08	15q: Heterozygous duplication affecting the probes for <i>SCG5</i> and <i>GREM1</i> .
Coriell NA04127	BP03-09	3p: Heterozygous duplication affecting the probes for <i>VHL</i> , <i>EPM2AIP1</i> and <i>MLH1</i> .
<i>REF-Male-3</i>	BP03-10	<i>Reference sample: normal ratios for all probes.</i>
Coriell NA14626	BP03-14	17q: Heterozygous duplication affecting the probes for <i>BRCA1</i> exon 12 & intron 12.
<i>REF-Male-4</i>	BP03-15	<i>Reference sample: normal ratios for all probes.</i>
Coriell NA12519	BP03-16	7q: Triplication of chromosome 7 affecting the probes for <i>MET</i> .
Coriell NA10985	BP03-18	3q: Heterozygous deletion affecting the probes for <i>VHL</i> . 17q: Positive for the <i>HOXB13</i> G84E mutation.
<i>REF-Male-5</i>	BP03-19	<i>Reference sample: normal ratios for all probes.</i>
Coriell NA08386	BP03-20	10q: Heterozygous duplication affecting the probes for <i>ACTR1A</i> , <i>SUFU</i> and <i>TRIM8</i> .
Coriell NA17941	BP03-21	1q: Heterozygous duplication affecting the probes for <i>SDHC</i> , <i>FH</i> and <i>KMO</i> .
Coriell NA14131	BP03-23	5p: Heterozygous deletion affecting the probes for <i>SDHA</i> .
Coriell NA10074	BP03-24	14q: Heterozygous duplication affecting the probes for <i>MAX</i> , <i>DICER1</i> and <i>CLMN</i> .

* Single probe aberrations may not be listed in the expected results.

3. Analysis in Coffalyser digitalMLPA software

The FASTQ file *FASTQ digitalMLPA Demo Set D001-D2.fastq* is generated by the Illumina® NGS platform. You can use this file to analyse the samples following the procedure in chapter **3.2 Analyse Your Data** as described in the [Coffalyser digitalMLPA User Manual](#).

Specific information has to be entered into Coffalyser digitalMLPA in several steps in section 3.2.1.1 *Configure general settings*. This concerns the steps listed below:

- Step 2: Leave the barcode lot on *From lot 03-009-yyymmdd onwards (Default)*.
- Step 4a: Select the barcodes listed in Table 1.
- Step 7: Select *D002* from the product drop-down menu.

- Step 8: Select *A1-0225* from the sheet drop-down menu.
- Step 10: Select *Reference* from the drop-down menu in the column *type* for the reference samples (see Table 1). All other samples can be defined as *Test*.
- Step 11: Select *Pooled DNA source* from the drop-down menu in the column *options* for the reference samples. Leave this on *Default* for all other samples.