INSTRUCTIONS on digitalMLPA Data Demo Set

NXtec™ D008-A1 Hereditary Cancer Panel 1

1. Purpose

This digitalMLPA data demo set serves to provide an overview of possible results that can be obtained with NXtec™ D008-A1 DMD. It contains data files of eight 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 D008-A1 data demo set

DNA Sample	Barcode used	Expected Results*
REF-Male-1	BP01-29	*Reference sample: normal ratios for all probes.*
Coriell NA05117	BP01-30	Heterozygous deletion affecting the probes for *DMD* Exon 45.
Coriell NA05123	BP01-31	Duplication affecting the probes for *DMD* Exon 45-62.
REF-Male-2	BP01-32	*Reference sample: normal ratios for all probes.*
Coriell NA05169	BP01-33	Deletion affecting the probes for *DMD* Exon 4-43.
Coriell NA10283	BP01-34	Deletion affecting the probes for *DMD* Exon 72-79.
REF-Male-3	BP01-35	*Reference sample: normal ratios for all probes.*
Coriell NA14523	BP01-36	Heterozygous deletion affecting all probes for *DMD,* the first 10 upstream probes closest to *DMD,* and the probes downstream of *DMD*. Heterozygous duplication affecting reference probe S013190 on chromosome 5.
Coriell NA23094	BP01-37	Heterozygous deletion affecting the probes for *DMD* Exon 35-43.
REF-Male-4	BP01-38	*Reference sample: normal ratios for all probes.*
Coriell NA20027	BP01-39	45,X karyotype, affecting all probes on chromosome X, including the probes for *DMD*.
Coriell NA04626	BP01-40	47,XXX karyotype, affecting all probes on chromosome X, including the probes for *DMD*.
REF-Male-5	BP01-41	*Reference sample: normal ratios for all probes.*

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

3. Analysis in Coffalyser digitalMLPA software

The FASTQ file *FASTQ digitalMLPA Demo Set D008-A1.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-yymmdd onwards (Default)*.
- Step 4a: Select the barcodes listed in Table 1.
- Step 7: Select *NXtec D008* from the product drop-down menu.
- Step 8: Select *A1-0226* 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: Check the selection box in the column *pooled DNA* for the reference samples.