INSTRUCTIONS on digitalMLPA Data Demo Set

NXtec™ D028-A1 Carrier Panel 1

1. Purpose

This digitalMLPA data demo set serves to provide an overview of possible results that can be obtained with NXtec™ D028-A1 Carrier Panel 1. It contains data files of nine 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 D028-A1 data demo set

DNA Sample*	Barcode used	Expected Results**
REF-Male-1	BP01-49	*Reference sample: normal ratios for all probes.*
Coriell NA03815	BP01-50	Heterozygous deletion affecting the probes for *SMN1* Exon 7 and 8.
Coriell NA20294	BP01-51	Heterozygous duplication affecting the probes for *SMN1* Exon 7 and 8. Positive for the *SMN1* c.*3+80T>G polymorphism.
REF-Male-2	BP01-52	*Reference sample: normal ratios for all probes.*
Coriell NA14732	BP01-53	Heterozygous deletion affecting the probes for *CYP21A2*.
Coriell HG01565	BP01-54	Heterozygous deletion affecting the probes for *CFTR*. Decreased ratio for *CYP21A2* probe S023020.
Coriell NA20480	BP01-55	Heterozygous deletion affecting the probes for *HBB* upstream and Exon 1, and *HBD* Exon 3 (Hb Lepore-Baltimore deletion). Decreased ratio for *GJB2* probe S013372.
Coriell NA07945	BP01-56	Heterozygous deletion affecting the probes for *GJB6* Exon 2 and *CRYL1* Exon 4 (232 kb *GJB6/CRYL1* deletion (ClinVar ID 5548).
REF-Male-3	BP01-57	*Reference sample: normal ratios for all probes.*
Coriell HG02188	BP01-59	Positive for the *HBA2* c.427T>C Constant Spring mutation. Heterozygous duplication affecting the probe for *SMN1* Exon 8.
Coriell NA05875	BP01-60	Heterozygous deletion affecting the probes for *CLN3*.
REF-Male-4	BP01-61	*Reference sample: normal ratios for all probes.*
Coriell NA00909	BP01-62	Heterozygous deletion affecting the probes for *CTNS* Exon 1-9 (57 kb *CTNS* deletion (ClinVar ID 4445)).
REF-Male-5	BP01-64	*Reference sample: normal ratios for all probes.*

* The reference samples have been selected using SALSA® Sample DNA SD100. SD100 is not included in this demo set.
** Single probe aberrations may not be listed in the expected results.

3. Analysis in Coffalyser digitalMLPA software

The FASTQ file *FASTQ digitalMLPA Demo Set D028-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 D028* from the product drop-down menu.
- Step 8: Select *A1-1024* 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.