

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.

* 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-yyymmdd 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.