MRC Holland
SALSA®
digitalMLPA™

INSTRUCTIONS digitalMLPA Data Demo Set

SALSA® digitalMLPA™ Probemix
D006-A1 Multiple Myeloma

1. Purpose

This digitalMLPA data demo set serves to provide an idea of possible results that can be obtained with SALSA® digitalMLPA™ Probemix D006-A1 Multiple Myeloma. 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 data 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 D006-A1 data demo set

DNA Sample	Barcode	Expected Results
Coriell NA07981	BP02-17	**12p**: Heterozygous triplication/homozygous duplication affecting the probes for *WNK1, CACNA2D4, TSPAN9, LTBR, CD27, VAMP1, NCAPD2, CHD4, ETV6, BCL2L14, LRP6, BORCS5, CDKN1B, GPRC5D, AEBP2, REP15, MANSC4, PTHLH, TMTC1*.
REF-Male-1	BP02-18	*Reference sample: normal ratios for all probes.*
Coriell NA06226	BP02-20	**16p**: Heterozygous duplication affecting the probes for *DECR2, IFT140, TXNDC11, TNFRSF176* and *CPPED1*.
REF-Male-2	BP02-21	*Reference sample: normal ratios for all probes.*
Coriell NA05067	BP02-22	**9p**: Heterozygous duplication affecting the probes for *DOCK8, JAK2, GLDC, FREM1, BNC2, FBXO10* and *DCAF10*. **14q**: Heterozygous deletion affecting the probes for *IGHD* and *IGHM*.
Coriell NA10925	BP02-23	**7p**: Heterozygous deletion affecting the probes for *ADCY1, ABCA13* and *IKZF1*.
Coriell NA09102	BP02-24	**11q**: Heterozygous deletion affecting the probes for *NTM, JAM3* and *NCAPD3*.
Coriell NA02944	BP02-26	**20p**: Heterozygous duplication affecting the probes for *RBCK1, RSPO4, TGM6, TRMT6, PLCB4, RIN2* and *APMAP*. **22q**: Heterozygous deletion affecting the probes for *GAB4, CECR2* and *HIRA*.
REF-Male-3	BP02-27	*Reference sample: normal ratios for all probes.*
Coriell NA03184	BP02-28	**15q**: Trisomy chromosome 15 affecting the probes for *GABRB3, SPG11, FBN1, VPS13C, USP3, CLN6, IGF1R, CHSY1* and *TM2D3*.
Coriell NA05832	BP02-29	**13q**: Heterozygous duplication affecting the probes for *ENOX1, RB1, RCBTB2, DLEU2, KCNRG, MIR15A, DLEU1, DLEU7, RNASEH2B, ATP7B, VPS36, PCDH8, KLHL1, DIS3, ARHGEF7* and *GRK1*. **18q**: Heterozygous deletion affecting the probes for *CTDP1* and *TXNL4A*.
Coriell NA00803	BP02-30	**1q**: Heterozygous deletion affecting the probes for *FCRL5, SLAMF7, NUF2, PBX1*-area and *PBX1*.
REF-Male-4	BP02-31	*Reference sample: normal ratios for all probes.*
REF-Male-5	BP02-32	*Reference sample: normal ratios for all probes.*

3. Analysis in Coffalyser digitalMLPA software

You can analyse the samples with Coffalyser digitalMLPA using either the FASTQ file or the Coffa files provided.

3.1. Analysis using FASTQ file

The FASTQ file *FASTQ digitalMLPA Data Demo Set D006-A1.fastq.gz* was generated by the Illumina® NGS platform. It is provided in a zip archive, which can be imported directly in Coffalyser digitalMLPA without the need to unpack the archive first. It is recommended to use this file if you wish to do the complete analysis from start to finish. Follow the procedure for creating a new experiment as described in the *Coffalyser digitalMLPA User Manual*.

In several steps described in the User Manual, you are required to enter specific information into Coffalyser digitalMLPA:

- Step 6: Select the barcode lot on *03-xxx-yymmdd ; Illumina standard*.
- Step 8: Select the barcodes listed in Table 1.
- Step 11: Select *digitalMLPA* from the experiment type drop-down menu.
- Step 12: Select *D006* from the product drop-down menu.
- Step 13: Select *D006-A1 tube AB4B0D 13-10-2022* from the sheet drop-down menu.
- Step 15: 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 16: 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.

3.2. Analysis using Coffa files

Alternatively, you can perform an analysis using the Coffa files included [1]. A Coffa file is a file (*.coffa) containing the sequencing output and several analysis properties of a single digitalMLPA reaction. Analysis using the Coffa files is faster than using the FASTQ file.

For instructions, consult the *Coffalyser digitalMLPA User Manual* for the procedure on reanalysis of an experiment using Coffa files.

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[1] The Coffa files have been generated with Coffalyser digitalMLPA version 221020.1234. They may not be compatible with newer versions of the software.