INSTRUCTIONS on digitalMLPA Data Demo Set

NXtec™ D024-A1 KaryoProfiler

1. Purpose

This digitalMLPA data demo set serves to provide an overview of possible results that can be obtained with NXtec™ D024-A1 KaryoProfiler. 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 D024-A1 data demo set

DNA Sample	Barcode used	Expected Results *
Coriell NA22976	BP04-49	**1p**: Heterozygous deletion affecting the probes for *AGRN* and *AJAP1*. **3q**: Heterozygous duplication affecting the probes for *ATP11B*, *MAP3K13*, *ST6GAL1*, *LPP*, *TPRG1*, *IL1RAP*, *MB21D2*, *ACAP2* and *DLG1*. **9**: Heterozygous duplication affecting the probes for *KANK1*, *VLDLR*, *JAK2*, *GLDC*, *PTPRD*, *LURAP1L*, *FREM1*, *BNC2*, *ADAMTSL1*, *MLLT3*, *IZUMO3*, *LINGO2*, *NFX1*, *GALT*, *PAX5*, *PIP5K1B*, *TRPM3*, *ANXA1*, *RORB*, *VPS13A*, *FRMD3*, *DAPK1*, *NXNL2*, *SYK*, *BICD2*, *FANCC*, *TDRD7*, *TGFBR1*, *ALDOB*, *SLC44A1*, *RAD23B*, *PTPN3*, *MUSK*, *KIAA1958*, *TNC*, *TRIM32*, *BRINP1*, *B3GALT9*, *RABGAP1*, *RALGPS1*, *NUP188*, *ABL1*, *OLFM1* and *NOTCH1*.
REF-Male-1	BP04-51	*Reference sample: normal ratios for all probes.*
Coriell NA03563	BP04-52	**3**: Heterozygous duplication affecting the probes for *TMEM45A*, *ALCAM*, *DUBR*, *DZIP3*, *NECTIN3*, *BOC*, *ZBTB20*, *TUSC7*, *UPK1B*, *POLQ*, *PARP14*, *UMPS*, *PLXNA1*, *RPN1*, *COL6A6*, *TMEM108*, *CEP63*, *NCK1*, *PIK3CB*, *CLSTN2*, *ATR*, *ZIC1*, *HLTF*, *MED12L*, *RAP2B*, *MME*, *LEKR1*, *GFM1*, *PPM1L*, *SLITRK3*, *SERPINI2*, *MECOM*, *TNIK*, *NLGN1*, *NAALADL2*, *TBL1XR1*, *PIK3CA*, *FXR1*, *ATP11B*, *MAP3K13*, *ST6GAL1*, *LPP*, *TPRG1*, *IL1RAP*, *MB21D2*, *ACAP2* and *DLG1*. **9p**: Heterozygous deletion affecting the probe for *KANK1*.
Coriell NA00782	BP04-53	**4p**: Heterozygous duplication affecting the probes for *ADGRL3*, *EPHA5*, *CENPC*, *RUFY3*, *AFM*, *PARM1*, *SHROOM3*, *BMP2K*, *PRKG2*, *MRPS18C*, *CDS1*, *HERC3*, *CCSER1*, *GRID2*, *BMPR1B*, *ADH4*, *PPP3CA*, *NFKB1*, *DKK2*, *SEC24B*, *ENPEP*, *ANK2*, *UGT8*, *TRAM1L1*, *MYOZ2*, *PRDM5*, *SPRY1*, *FAT4*, *JADE1*, *PCDH10*, *RAB33B*, *TBC1D9*, *GAB1* and *HHIP*.
REF-Male-2	BP04-54	*Reference sample: normal ratios for all probes.*
Coriell NA10313	BP04-55	**4q**: Heterozygous duplication affecting the probes for *SH3D19*, *TRIM2*, *LRAT*, *PDGFC*, *FNIP2*, *NPY5R*, *KLHL2*, *SPOCK3*, *PALLD*, *HPGD*, *TENM3*, *CASP3* and *FAT1*. **7q**: Heterozygous deletion affecting the probes for *RNF32* and *VIPR2*.
Coriell NA16445	BP04-57	**17q**: Heterozygous duplication affecting the probes for *MGAT5B*, *LGALS3BP*, *RPTOR*, *FN3KRP* and *TBCD*. **18p**: Heterozygous deletion affecting the probe for *ENOSF1*.
Coriell NA07891	BP04-58	**12q**: Heterozygous duplication affecting the probes for *TMEM132D*, *ADGRD1* and *ZNF10*. **18q**: Heterozygous deletion affecting the probes for *ELAC1*, *DCC*, *TCF4*, *WDR7*, *MALT1*, *CDH20*, *KDSR*, *CDH19*, *TMX3*, *GTSCR1*, *CBLN2*, *ZNF407*, *MBP* and *NFATC1*.
REF-Male-3	BP04-59	*Reference sample: normal ratios for all probes.*
Coriell NA09687	BP04-60	**16p**: Heterozygous deletion affecting the probe for *MSLN*.

		16q: Heterozygous duplication affecting the probes for *CALB2*, *ZFHX3*, *RFWD3*, *ADAMTS18*, *BCO1*, *CDH13*, *CRISPLD2*, *ZCCHC14* and *GALNS*.
REF-Male-4	BP04-62	*Reference sample: normal ratios for all probes.*
Coriell NA03435	BP04-63	**4p**: Heterozygous deletion affecting the probes for *PDE6B*, *ADD1*, *CYTL1*, *TBC1D14*, *GPR78*, *CLNK*, *CPEB2*, *LDB2* and *KCNIP4*. **11p**: Heterozygous duplication affecting the probes for *DEAF1* and *MMP26*.
REF-Male-5	BP04-64	*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 D024-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 *D024* from the product drop-down menu.
- Step 8: Select *A1-0125* 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.