MRC Holland Support

List of verified positive samples that can be used with (digital)MLPA probemixes

Inclusion

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Inclusion of a positive sample in each MLPA or digitalMLPA experiment is not required, but can be useful for the analysis of your experiments. MRC Holland has very limited access to positive samples and cannot supply such samples. You can sometimes obtain a useful positive sample from an online biorepository, such as the one from the Coriell Institute.

We have tested a large number of commercially available positive samples with our MLPA or digitalMLPA probemixes. A list of positive samples that provide useful results and their verified copy number change(s) can be found in the table below. These samples can be used as positive control samples for the specified probemix(es). You are welcome to contact us if you know of other positive samples that can be useful for one of our probemixes.

Important
The quality of cell lines can change, and these samples are not controlled by us. In addition, exon numbering may change and the table may contain inaccuracies. Therefore, all samples should be validated in-house before use.
Notes:

1. All samples can be obtained from the Coriell Institute or DSMZ unless noted otherwise. You can find these samples by searching for the sample id on the appropriate website.
2. Samples obtained from the Coriell Institute can often be diluted to 10 ng/µl upon arrival.
3. Probemixes are specified by their major version. Read more about version numbers of probemixes in this article.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
D001-C	Various	See the product description for a large number of positive samples.	
D006-A	Various	See the product description for a large number of positive samples.	

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
D007-A	Various	See the product description for a large number of positive samples.	
ME011-D	2p21-p16.3 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for EPCAM, MSH2 and MSH6, the digestion control probe at 132 nt and the reference probes at 127 and 190 nt. All MMR genes are not methylated.
		Coriell NA13451 (m)	Heterozygous deletion affecting the probes for EPCAM, MSH2 and MSH6. All MMR genes are not methylated.
	3p22.2 region	Coriell NA04127 (f)	Heterozygous duplication affecting the probes for MLH1. All MMR genes are not methylated.
		DSMZ DU-4475 (f)	Positive for the BRAF c.1799T>A (p.V600E) mutation. Moderate methylation (~50%) for 247 and 278 nt MLH1 probe targets, all other genes are not methylated.
		DSMZ SK-N-MC (f)	Heterozygous deletion affecting the probes for MLH1, the digestion control probe at 132 nt and the reference probes at 178 and 398 nt. Low methylation (10-15%) for all MLH1 probe targets, all other genes are not methylated.
	7p22.1 region	Coriell NA07081 (m)	Heterozygous duplication affecting the probes for PMS2. All MMR genes are not methylated.
		DSMZ DK-MG-UN (f)	Heterozygous duplication affecting the probes for PMS2. Heterozygous deletion affecting the digestion control probe at 132 nt and the reference probes at 238 and 398 nt. All MMR genes are not methylated.
ME012-B	various	Coriell NA00959 (m)	Heterozygous duplication affecting all MGMT probes. All MGMT probes are not methylated.
		Coriell NA05299 (f)	Heterozygous deletion affecting all MGMT probes. All MGMT probes are not methylated.
		Coriell NA20263 (m)	Heterozygous deletion affecting all MGMT probes. All MGMT probes are not methylated.
		DSMZ ACC-3 (HL-60) (f)	All MGMT probes are not methylated. Some of the reference probe targets are affected by copy number alterations.
		DSMZ ACC-180 (HEP-G2) (m)	Positive for the TERT C228T mutation. All MGMT probes are methylated. Some of the reference probe and control probe targets are affected by copy number alterations.
		DSMZ ACC-277 (DK-MG) (f)	Heterozygous deletion affecting all MGMT probes. Positive for the TERT C250T mutation. All MGMT probes are methylated.
		DSMZ ACC-405 (GMS-10) (m)	Heterozygous deletion affecting all MGMT probes. Positive for the TERT C228T mutation. All MGMT probes are methylated except the probes at 175 and 215 nt. Some of the reference probe targets are affected by copy number alterations.
		DSMZ ACC-431 (42-MG-BA) (m)	Positive for the TERT C228T mutation. All MGMT probes are methylated except the probes at 175. Some of the reference probe targets are affected by copy number alterations.
		DSMZ ACC-569 (MOLP-8) (f)	Heterozygous deletion affecting all MGMT probes. All MGMT probes are not methylated. Some of the reference probe targets are affected by copy number alterations.
		Promega Methylated Human Control DNA	Positive control - all targets are methylated.
		EpigenDx Human High Methylated Genomic DNA	Positive control - all targets are methylated.
		Sigma-Aldrich Methylated Control DNA	Positive control - all targets are methylated.
ME024-B	9p region	Coriell NA01750 (m)	Gain affecting the probes for DOCK8, GLDC, MLLT3, KLHL9, MIR31, MTAP, CDKN2A, CDKN2B-AS1 and CDKN2B. CDKN2A/2B are unmethylated; MIR31 is methylated.
		Coriell NA02819 (f)	Gain affecting the probes for DOCK8, GLDC, MLLT3, KLHL9, MIR31, MTAP, CDKN2A, CDKN2B-AS1 and CDKN2B. CDKN2A/2B are unmethylated; MIR31 is methylated.
		Coriell NA03226 (m)	Gain affecting all target probes on 9p. CDKN2A/2B are unmethylated; MIR31 is methylated.
		Coriell NA05067 (m)	Gain affecting all target probes on 9p. CDKN2A/2B are unmethylated; MIR31 is partially methylated.
		DSMZ ACC-3 (HL-60) (f)	Heterozygous loss affecting all target probes on 9p21.3.Gain affecting the reference probe at 124 nt. CDKN2A/2B are unmethylated; MIR31 is methylated.
		DSMZ ACC-29 (MOLT-16) (f)	Homozygous deletion of CDKN2A and CDKN2B. MIR31 is methylated.
		DSMZ ACC-200 (COLO-824) (f)	Gain affecting all target probes on 9p. CDKN2A/2B are unmethylated; MIR31 is methylated.
		DSMZ ACC-203 (SK-N-MC) (f)	Loss affecting the reference probe at 185 nt CDKN2A is unmethylated; CDKN2B is methylated; MIR31 is methylated.
		DSMZ ACC-255 (CADO-ES1) (f)	Homozygous loss affecting the probes for MTAP, CDKN2A and CDKN2B. MIR31 is unmethylated.
		DSMZ ACC-573 (SU-DHL-8) (f)	CDKN2A exon 1 is not methylated; CDKN2B is partially methylated; CDKN2A exon 2 and MIR31 are methylated.
		DSMZ ACC-581 (HCT-116) (m)	CDKN2A is methylated; CDKN2B is unmethylated; MIR31 is methylated.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
ME028-D	15q11/SNRPN	Coriell NA13554 (m)	Heterozygous deletion (maternal) affecting the probes for SNRPN exon 3 and exon u5; asymptomatic.
		Coriell NA13556 (f)	Heterozygous deletion (paternal) affecting the probes for SNRPN exon 3 and exon u5; Prader-Willi syndrome.
		Coriell NA20375 (m)	Heterozygous deletion affecting the probes for MKRN3, MAGEL2, NDN, SNRPN, UBE3A, ATP10A, GABRB3 and OCA2; Angelman syndrome.
		Coriell NA20408 (f)	Uniparental disomy (no copy number changes); Prader-Willi syndrome.
		Coriell NA21887 (f)	Heterozygous deletion affecting the probes for TUBGCP5, NIPA1, MKRN3, MAGEL2, NDN, SNRPN, UBE3A, ATP10A, GABRB3 and OCA2; Angelman syndrome.
		Coriell NA22397 (m)	Heterozygous duplication (maternal) affecting the probes for MKRN3, MAGEL2, NDN, SNRPN, UBE3A, ATP10A, GABRB3 and OCA2.
		The NIBSC Institute provides an excellent panel of WHO certified genomic DNA samples for Prader Willi and Angelman syndrome (catalogue number 09/140).	
ME029-B	FMR1	Coriell NA09145 (m)	FMR1 full mutation sample.
		Coriell NA09237 (m)	FMR1 full mutation sample.
		Coriell NA20231 (m)	FMR1 premutation sample.
ME031-C	20q12.32 region	Coriell NA08123 (m)	Heterozygous duplication of the maternal allele affecting the probes for STX16, GNAS-AS1, NESP55, GNASXL, GNAS-A/B, GNAS and NELFCD. Methylation ratio of ~0.33 (33%) expected for NESP55 DMR, and ~0.67 (67%) for GNAS-AS1, GNASXL and GNAS A/B DMRs.
ME032-B	7p12.2 region	Coriell NA07081 (m)	Heterozygous duplication affecting the probes for GRB10.
		Coriell NA10925 (m)	Heterozygous deletion affecting the probes for GRB10.
	7q32.2 region	Coriell NA12519 (f)	Homozygous duplication affecting the probes for MEST.
	14q32.2-q32.31 region	Coriell NA13410 (m)	Homozygous duplication affecting the probes for DLK1, MEG3, RTL1 and MIR380.
ME042-C	CDKN2A	Coriell NA02819 (f)	Gain affecting the probes for CDKN2A. IGF2 is methylated.
		Coriell NA05067 (m)	Gain affecting the probes for CDKN2A.
	MLH1	Coriell NA04127 (f)	Gain affecting the probes for MLH1.
	SOCS1	Coriell NA06226 (m)	Gain affecting the probes for SOCS1.
		Coriell NA08039 (m)	Gain affecting the probes for SOCS1. IGF2 is methylated.
	CACNA1G	Coriell NA04127 (f)	Heterozygous deletion affecting the probes for CACNA1G. CRABP1 is methylated.
	Various	DSMZ ACC-022 (REH) (f)	RUNX3, NEUROG1, CDKN2A, IGF2, CRABP1 and CACNA1G are methylated.
		DSMZ ACC-042 (697) (m)	RUNX3, NEUROG1, CDKN2A, IGF2, CRABP1 and CACNA1G are methylated.
		DSMZ ACC-255 (CADO-ES1) (f)	Homozygous deletion affecting the probes for CDKN2A. Gain affecting the probes for CACNA1G. NEUROG1, IGF2 and CRABP1 are methylated.
		DSMZ ACC-427 (DU-4475) (f)	Positive for the BRAF c.1799T>A (p.V600E) mutation. RUNX3, MLH1, NEUROG1, CDKN2A, IGF2, CRABP1 and CACNA1G are methylated.
		DSMZ ACC-573 (SU-DHL-8) (m)	RUNX3, NEUROG1, CDKN2A, IGF2, CRABP1 and CACNA1G are methylated.
		DSMZ ACC-581 (HCT-116) (m)	Gain affecting the probes for CACNA1G. RUNX3, NEUROG1, CDKN2A, IGF2, CRABP1 and CACNA1G are methylated.
ME053-A	13q12.11-q32.3 region & 17p11.2-p13.1 region & 17q21.33-q22 region	DSMZ ACC-9 (U-266) (m)	Heterozygous deletion affecting the probes for ZMYM2, CENPJ, BRCA2, N4BP2L, PCCA5, ATP1B2 and AKAP10. Gain affecting the probes for SGCA, COL1A1 and RAD51C.
	13q12.11-q32.3 region & 17p11.2-p13.1 region	DSMZ ACC-10 (K-562) (f)	Heterozygous deletion affecting the probes for ZMYM2, CENPJ, BRCA2, N4BP2L, PCCA5, ATP1B2 and AKAP10.
	BRCA2	DSMZ ACC-573 (SU-DHL-8) (m)	Gain affecting the probes for BRCA2. RAD51C is methylated.
	RAD51C	DSMZ ACC-20 (BV-173) (m)	RAD51C is methylated.
		ACC-78 (DAUDI) (m)	RAD51C is methylated.
	17q21.33-q22 region	DSMZ ACC-255 (CADO-ES1) (f)	Gain affecting the probes for SGCA, COL1A1 and RAD51C.
		DSMZ ACC-581 (HCT-116) (m)	Gain affecting the probes for SGCA, COL1A1 and RAD51C.
	Various	Zymo Research Human HCT116 DKO Non-Methylated DNA	Gain affecting the probes for SGCA, COL1A1 and RAD51C. Negative control - all targets are non-methylated.
		Zymo Research Human HCT116 DKO Methylated DNA	Gain affecting the probes for SGCA, COL1A1 and RAD51C. Positive control - all targets are methylated.
		Promega Methylated Human Control DNA	Positive control - all targets are methylated.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P002-D	BRCA1	Coriell NA14626 (f)	Heterozygous duplication affecting the probes for BRCA1 exon 13.
		Coriell NA18949 (f)	Heterozygous deletion affecting the probes for BRCA1 exon 15-16.
		Coriell NA14094 (f)	Heterozygous deletion of 40 nt in BRCA1 exon 11 affecting the probe for BRCA1 exon 11 at 382 nt.
P003-D	MLH1/MSH2	The NIBSC Institute provides a kit with 5 DNA samples containing heterozygous MLH1 or MSH2 exon deletions or amplifications (catalog number 11/218-XXX).	
P015-F	Xq28 region	Coriell NA23599 (f)	Heterozygous deletion affecting the probes for MECP2 exon 3-4.
		Coriell NA23635 (f)	Heterozygous deletion affecting all probes for MECP2 exon 3 and three for MECP2 exon 4 at 356 nt, 229 nt and 346 nt.
		Coriell NA23648 (f)	Heterozygous deletion affecting the probes for IRAK1 and MECP2 exon 4 at 260 nt, 418 nt, 292 nt, 274 nt and 154 nt.
		Coriell NA23654 (f)	Heterozygous deletion affecting the probes for MECP2 exon 3 and MECP2 exon 4 at 356 nt, 229 nt, 346 nt and 154 nt.
		Coriell NA23676 (f)	Heterozygous duplication affecting the probes for IRAK1 and MECP2.
		Coriell NA23733 (f)	Heterozygous duplication affecting the probes for L1CAM, IRAK1, MECP2 and FLNA.
		Coriell NA23734 (m)	Duplication affecting the probes for L1CAM, IRAK1, MECP2 and FLNA.
P016-C	3p25.3-p26.3 region	Coriell NA10985 (f)	Heterozygous deletion affecting the probes for CNTN6, FANCD2, BRK1, VHL, IRAK2 and GHRL.
	3p25.3 region	Coriell NA13249 (m)	Heterozygous deletion affecting the probes for VHL exon 2-3 and IRAK2.
		Coriell NA13250 (f)	Heterozygous deletion affecting the probes for VHL exon 1-2.
		Coriell NA13256 (m)	Heterozygous deletion affecting the probes for FANCD2, BRK1 and VHL.
P018-G	SHOX	Coriell NA04626 (f)	Heterozygous duplication affecting the probes for SHOX, the upstream and downstream SHOX area, AIFM and VAMP7.
		Coriell NA20212 (f)	Heterozygous deletion (~0.9 Mb) of the SHOX gene and the downstream SHOX area.
		Coriell NA20217 (m)	Compound heterozygous deletions: one from upstream of SHOX to downstream of SHOX, and one in the SHOX downstream area.
		Coriell NA20218 (f)	Compound heterozygous deletions: one from upstream of SHOX to downsteam of SHOX, and one from upstream of SHOX to the last probe of SHOX, resulting in a homozygous deletion of all probes for SHOX.
P021-B	SMN1/SMN2	See the product description for a large number of positive samples.	
P022-B	Xq22 region	Coriell NA11005 (m)	Heterozygous duplication affecting the probes for BEX4, BEX2, NGFRAP1, RAB40A, TCEAL1, MORF4L2, TMEM31, GLRA4, PLP1, RAB9B and TMSB15B.
	Xq22.2 region	Coriell NA13434 (m)	Heterozygous deletion affecting the PLP1 exon 5 probe.
P025-A	17p/CTNS/ASPA	Coriell NA06047 (m)	Large heterozygous deletion (5.6 Mb) that includes the complete ASPA gene.
P026-E	NSD1	Coriell NA01535 (f)	Heterozygous duplication affecting the probes for NSD1.
		Coriell NA04371 (m)	Heterozygous duplication affecting the probes for NSD1.
P027-C	3p25.3-p26.3 region	Coriell NA10985 (f)	Heterozygous deletion affecting the probes for CHL1, BRK1 and VHL.
		Coriell NA03503 (m)	Heterozygous duplication affecting the probes for CHL1, BRK1, VHL, PPARG and XPC.
	3p21.31-p26.3 region	Coriell NA04127 (f)	Heterozygous duplication affecting the probes for CHL1, BRK1, VHL, PPARG, XPC, MIR128-2, MLH1, CTNNB1 and RBM5.
	3q11.2-q21.1 region	Coriell NA08778 (m)	Heterozygous deletion affecting the probes for PROS1 and CASR.
	3q21.1-q29 region	Coriell NA03563 (m)	Heterozygous duplication affecting the probes for CASR, MME and OPA1.
	3q29 region	Coriell NA10175 (m)	Heterozygous duplication affecting the OPA1 probe.
		Coriell NA22976 (m)	Heterozygous duplication affecting the OPA1 probe.
	6p25.2 region	Coriell NA22770 (m)	Heterozygous duplication affecting the ECI2 probe.
	6q23.2 region	Coriell NA09367 (f)	Heterozygous duplication affecting the CCN2 probe.
	6q25.3 region	Coriell NA06802 (m)	Heterozygous deletion affecting the IGF2R probe.
		Coriell NA07994 (m)	Heterozygous duplication affecting the IGF2R probe.
	8p21.3 region	Coriell NA03255 (m)	Heterozygous duplication affecting the LZTS1 probe.
	8p12-p21.3 region	Coriell NA14485 (m)	Heterozygous duplication affecting the probes for LZTS1 and NRG1.
	8p21.3-q24.21 region	Coriell NA02030 (m)	Heterozygous duplication affecting the probes for LZTS1, NRG1, RP1, MYC and ASAP1.
	8q24.21 region	Coriell NA03999 (f)	Heterozygous deletion affecting the probes for MYC and ASAP1.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P029-C	7q11.23 region	Coriell NA13464 (m)	Williams-Beuren syndrome deletion (commonly deletion region).
P031-B & P032-B	FANCA	Coriell NA09687 (m)	Heterozygous duplication affecting the probes for FANCA and GAS8.
P033-B	PMP22	Coriell NA05167 (f)	Heterozygous duplication that includes the COX10, PMP22 and TEKT3 genes.
		Coriell NA12214 (m)	Heterozygous duplication that includes the COX10, PMP22 and TEKT3 genes.
P034-B	DMD	Coriell NA05117 (f)	Heterozygous DMD exon 45 deletion.
P034-B & P035-B	DMD	Coriell NA05123 (m)	DMD exon 45-62 duplication.
		Coriell NA23087 (f)	Heterozygous DMD exon 2-30 duplication.
		Coriell NA23094 (f)	Heterozygous DMD exon 35-43 deletion.
P035-B	DMD	Coriell NA10283 (m)	DMD exon 72-79 deletion.
P036-E	1p	Coriell NA22991 (f)	Heterozygous deletion affecting the TNFRSF4 probe.
	2p & 4q	Coriell NA00501 (m)	Heterozygous deletion affecting the probe upstream of ACP1, and heterozygous duplication affecting the TRIML2 probe.
	5p	Coriell NA14131 (f)	Heterozygous deletion affecting the PDCD6 probe.
	17p	Coriell NA06047 (m)	Heterozygous deletion affecting the RPH3AL probe.
P037-B	2p region	Coriell NA00945 (f)	Heterozygous deletion that affects all MYCN probes.
		Coriell NA01353 (m)	Heterozygous duplication that affects all MYCN and ALK probes.
		Coriell NA04409 (m)	Heterozygous duplication that affects all MYCN probes.
		Coriell NA09216 (m)	Heterozygous deletion that affects all MYCN probes.
		Coriell NA10401 (f)	Heterozygous duplication that affects all MYCN, ALK and REL probes.
	6q region	Coriell NA06802 (m)	Heterozygous deletion that affects all IGF2R and PARK2 probes.
		Coriell NA07994 (m)	Heterozygous duplication that affects all TNFAIP3, LATS1, IGF2R and PARK2 probes.
		Coriell NA09367 (f)	Heterozygous duplication that affects all SEC63 and TNFAIP3 probes.
	8p region	Coriell NA03255 (m)	Heterozygous duplication that affects all TNFRSF10A and TNFRSF10B probes.
		Coriell NA14485 (m)	Heterozygous duplication that affects all TNFRSF10A and TNFRSF10B probes.
	8p/q region	Coriell NA02030 (m)	Heterozygous duplication that affects all TNFRSF10A, TNFRSF10B, EIF3H and MYC probes.
	8q region	Coriell NA03999 (f)	Heterozygous deletion that affects all MYC probes.
	9p region	Coriell NA03226 (m)	Heterozygous duplication that affects all CDKN2A and CDKN2B probes.
	11q region	Coriell NA08618 (m)	Heterozygous duplication that affects all ATM probes.
		Coriell NA09596 (m)	Heterozygous deletion that affects all ATM probes.
	12p region	Coriell NA07981 (m)	Homozygous duplication that affects all CCND2 and LRMP probes.
	13q14 region	Coriell NA03330 (m)	Heterozygous duplication that affects all RB1, FNDC3A, KCNRG, MIR15A, DLEU2, DLEU7 and ATP7B probes.
		Coriell NA05832 (m)	Heterozygous duplication that affects all RB1, FNDC3A, KCNRG, MIR15A, DLEU2, DLEU7 and ATP7B probes.
		Coriell NA12606 (m)	Heterozygous duplication that affects all RB1, FNDC3A, KCNRG, MIR15A, DLEU2, DLEU7 and ATP7B probes.
		Coriell NA13721 (m)	Heterozygous deletion that affects all RB1, FNDC3A, KCNRG, MIR15A, DLEU2, DLEU7 and ATP7B probes.
		Coriell NA14164 (f)	Heterozygous deletion that affects all RB1, FNDC3A, KCNRG, MIR15A, DLEU2, DLEU7 and ATP7B probes.
	Various	Coriell NA02819 (f)	Heterozygous duplication that affects all CDKN2A and CDKN2B probes; heterozygous deletion that affects all CHFR probes.
		DSMZ ACC-203 (SK-N-MC) (f)	Heterozygous deletions that affects all MYCN and TP53 probes; amplification that affects all TNFRSF10A, TNFRSF10B, EIF3H and MYC probes.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P038-B	11q region	Coriell NA00959 (m)	Heterozygous duplication affecting probes for ATM, RDX, PPP2R1B and CADM1.
		Coriell NA15099 (m)	Heterozygous duplication affecting probes for ATM, RDX, PPP2R1B and CADM1.
	11q region & 14q region	Coriell NA09596 (m)	Heterozygous deletion affecting probes for ATM, RDX and IGHD.
	Coriell NA05832 (m)	Heterozygous duplication affecting probes for RB1, DLEU2, KCNRG, DLEU1 and ATP7B.	
	Coriell NA13721 (m)	Heterozygous deletion affecting probes for RB1, DLEU2, KCNRG, DLEU1 and ATP7B.	
	Coriell NA14164 (f)	Heterozygous deletion affecting probes for RB1, DLEU2, KCNRG, DLEU1 and ATP7B.	
	CD27	Coriell NA07981 (m)	Homozygous duplication affecting the probe for CD27.
	PTEN & IGHD	Coriell NA20125 (m)	Heterozygous duplication affecting probes for PTEN. Heterozygous deletion affecting the IGHD probe.
	SF3B1	DSMZ ACC-600 (HNT-34) (f)	Positive for the MSF3B1 c.2098A>G (p.K700E) mutation.
P040-B	11q region	Coriell NA00959 (m)	Heterozygous duplication of that affects all probes for the 11q region (CTTN, PICALM, ATM, DDX10, PCSK7 and NCAPD3).
		Coriell NA08618 (m)	Heterozygous duplication of part of the 11q22.3 region, affecting probes for ATM and DDX10.
		Coriell NA09596 (m)	Heterozygous deletion of part of 11q, affecting probes for PICALM, ATM and DDX10.
		Coriell NA15099 (m)	Heterozygous duplication affecting the probes for PICALM, ATM, DDX10, PCSK7 and NCAPD3.
	12p region	Coriell NA07981 (m)	Heterozygous triplication/ homozygous duplication affecting all probes on 12p (CCND2, CD27 and LRMP).
		Coriell NA08035 (m)	Heterozygous duplication affecting all probes on 12p (CCND2, CD27 and LRMP).
	13q14 region	Coriell NA02718 (f)	Heterozygous duplication affecting the probes for RB1, KCNRG, MIR15A, DLEU2, DLEU1, DLEU7 and ATP7B.
		Coriell NA03330 (m)	Heterozygous deletion affecting the probes for RB1, KCNRG, MIR15A, DLEU2, DLEU1, DLEU7 and ATP7B.
		Coriell NA13721 (m)	Heterozygous deletion affecting the probes for RB1, KCNRG, MIR15A, DLEU2, DLEU1, DLEU7 and ATP7B.
	Various	DSMZ ACC-9 (U-266) (m)	Heterozygous deletion affecting the probes for ATM, DDX10, PCSK7 and NCAPD3 on chromosome 11q, the probes for CCND2, CD27 and LRMP on chromosome 12p, the probes for RB1, KCNRG, MIR15A, DLEU2, DLEU1, DLEU7 and ATP7B on chromosome 13q, and the probes for TP53 on chromosome 17p. Some of the reference probe targets are also affected by copy number alterations.
	TP53	DSMZ ACC-203 (SK-N-MC) (f)	Homozygous TP53 exon 2 deletion, and heterozygous deletion of other TP53 exons. Some of the reference probe targets are also affected by copy number alterations.
P041-B & P042-B	11q22.3 region	Coriell NA00959 (m)	Heterozygous duplication affecting the probes for ATM.
		Coriell NA08618 (m)	Heterozygous duplication affecting the probes for ATM.
		Coriell NA09596 (m)	Heterozygous deletion affecting the probes for ATM.
		Coriell NA15099 (m)	Heterozygous duplication affecting the probes for ATM.
		Coriell HG03694 (m)	Heterozygous duplication affecting the probes for ATM intron 61 at 474 nt and 419 nt, and ATM exon 62-63.
P043-E	APC	Coriell NA14234 (m)	Heterozygous deletion affecting all APC probes.
	MUTYH	Coriell HG00097 (f), HG01095 (f), HG01519 (f), HG01685 (f), HG02224 (m), NA19789 (m), NA20522 (f) and NA20759 (m) are positive for the MUTYH c.1187G>A (p.Gly396Asp) mutation.	
		Coriell HG01918 (f)	Positive for the MUTYH c.536A>G (p.Tyr179Cys) mutation.
	15q13.3 region	Coriell NA03184 (m)	Heterozygous duplication affecting both GREM1-up probes.
P044-C	NF2	Coriell NA07106 (m)	Heterozygous duplication that affects all NF2 probes. The flanking probes are also affected.
P045-C/D	13q13.1 region	Coriell NA02718 (f)	Heterozygous deletion affecting the probes for BRCA2 and N4BP2L1.
		Coriell NA03330 (m)	Heterozygous duplication affecting the probes for BRCA2 and N4BP2L1.
		Coriell NA12606 (m)	Heterozygous duplication affecting the probes for BRCA2 and N4BP2L1.
	22q12.1 region	Coriell HG00187 (m)	Positive for the CHEK2 1100delC mutation.
P046-D	16p13 region	Coriell NA04520 (f)	Heterozygous deletion affecting the probes for TSC2 exon 1-15.
		Coriell NA06226 (m)	Homozygous duplication affecting the probes for TSC2 and PKD1.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P047-E	13q14.11-q21.1 region	Coriell NA03330 (m)	Heterozygous duplication affecting the probes for ENOX1, MED4, ITM2B, RB1, RCBTB2, DLEU1 and PCDH8. RB1 promoter (CpG106) probes: not methylated. RB1 imprinted locus(CpG85) probes: all copies methylated.
		Coriell NA12606 (m)	Heterozygous duplication affecting the probes for ENOX1, MED4, ITM2B, RB1, RCBTB2, DLEU1 and PCDH8. RB1 promoter (CpG106) probes: not methylated. RB1 imprinted locus(CpG85) probes: two copies methylated.
		Coriell NA13721 (m)	Heterozygous deletion affecting the probes for ENOX1, MED4, ITM2B, RB1, RCBTB2, DLEU1 and PCDH8. RB1 promoter (CpG106) probes: not methylated. RB1 imprinted locus(CpG85) probes: remaining copy *not* methylated.
		DSMZ ACC-009 (U-266) (m)	Heterozygous deletion affecting the probes for ENOX1, MED4, ITM2B, RB1, RCBTB2, DLEU1 and PCDH8. RB1 promoter (CpG106) probes: not methylated. RB1 imprinted locus(CpG85) probes: remaining copy methylated. Some of the reference probes are affected by CNAs.
		DSMZ ACC-163 (NCI-H929) (f)	Heterozygous deletion affecting the probes for ENOX1, MED4, ITM2B, RB1, RCBTB2, DLEU1 and PCDH8. RB1 promoter (CpG106) probes: not methylated. RB1 imprinted locus(CpG85) probes: remaining copy methylated. Some of the reference probes are affected by CNAs.
		DSMZ ACC-277 (DK-MG) (f)	Heterozygous deletion affecting the probes for ENOX1, MED4, ITM2B, RB1, RCBTB2, DLEU1 and PCDH8. RB1 promoter (CpG106) probes: not methylated. RB1 imprinted locus(CpG85) probes: remaining copy methylated. Some of the reference probes are affected by CNAs.
		DSMZ ACC-410 (MFE-28) (f)	Heterozygous deletion affecting the probes for ENOX1, ITM2B, DLEU1 and PCDH8. Homozygous deletion affecting the probes for RB1 and RCBTB2. No methylation data can be derived as both copies of RB1 gene including the CpG106 and CpG85 are deleted. Some of the reference probes are affected by CNAs.
	13q14.2-q14.3 region	DSMZ ACC-427 (DU-4475) (f)	Heterozygous deletion affecting the probes for MED4, ITM2B and DLEU1. Homozygous deletion affecting the probes for RB1 and RCBTB2. No methylation data can be derived as both copies of RB1 gene including the CpG106 and CpG85 are deleted.
	13q14.2-q21.1 region	Coriell NA14164 (f)	Heterozygous deletion affecting the probes for MED4, ITM2B, RB1, RCBTB2, DLEU1 and PCDH8. RB1 promoter (CpG106) probes: not methylated. RB1 imprinted locus(CpG85) probes: remaining copy methylated.
P049-C	ABCD1	Coriell NA17819 (m)	ABCD1 exon 6-10 deletion.
P050-C	CYP21A2	Coriell NA12217 (m)	Heterozygous CYP21A2 exon 1-4 deletion.
		Coriell NA14734 (m)	Homozygous deletion of the complete CYP21A2 gene.
P051-D	PARK2	Coriell NA21698 (m)	Heterozygous deletion affecting the probe for PARK2 exon 1.
	PARK2/LPA	Coriell NA06802 (m)	Heterozygous deletion affecting the probes for PARK2 and LPA.
		Coriell NA07994 (m)	Heterozygous duplication affecting the probes for PARK2 and LPA.
	SNCA	Coriell NA00782 (m)	Heterozygous duplication affecting the probes for SNCA.
		Coriell NA10800 (m)	Heterozygous deletion affecting the probes for SNCA.
		Coriell ND00196 (m)	Triplication affecting the probes for SNCA.
	PARK7/TNFRSF9	Coriell NA50276 (m)	Heterozygous deletion affecting the probes for PARK7 and TNFRSF9.
P051-D & P052-D	PARK2	Coriell ND01039 (m)	Heterozygous deletion affecting the probe for PARK2 exon 4.
		Coriell ND35201 (f)	Homozygous deletion affecting the probes for PARK2 exon 3 and 4.
	LRRK2	Coriell NA19750 (m)	Positive for the LRRK2 c.6055G>A (p.G2019S) mutation.
P052-D	PACRG	Coriell NA21698 (m)	Heterozygous deletion affecting the probe for PACRG exon 1.
	PARK2/PACRG	Coriell NA06802 (m)	Heterozygous deletion affecting the probes for PARK2 and PACRG.
		Coriell NA07994 (m)	Heterozygous duplication affecting the probes for PARK2 and PACRG.
	GCH1	Coriell NA10074 (m)	Heterozygous duplication affecting the probes for GCH1.
	UCHL1	Coriell NA10947 (f)	Heterozygous duplication affecting the probes for UCHL1.
	CAV1/CAV2	Coriell NA01059 (f)	Heterozygous deletion affecting the probes for CAV1 and CAV2.
		Coriell NA12519 (f)	Heterozygous triplication/Homozygous duplication affecting the probes for CAV1 and CAV2.
P056-D	17p13.1 region	DSMZ SK-N-MC (f)	Heterozygous deletion affecting the probes for POLR2a, MPDU1, ATP1B2, TP53, EFNB3 and AKAP10, and the reference probe at 471 nt. Homozygous deletion affecting the probes for TP53 exon 2. In addition, a heterozygous duplication affecting the reference probe at 135 nt.
P060-B	SMN1/SMN2	See the product description for a large number of positive samples.	
P061-D	17p	Coriell NA06047 (m)	Heterozygous 17p deletion (telomere-ASPA-TRPV1).

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P064-C	1p36 region	Coriell NA22991 (f)	1p36 deletion syndrome.
	4p16.3 region	Coriell NA22601 (m)	Wolf-Hirschhorn syndrome deletion.
	5p15 region	Coriell NA14131 (f)	Cri-du-Chat syndrome deletion.
	7q11.23 region	Coriell NA10160 (m)	Williams-Beuren syndrome deletion.
	8q24.11-q24.13 region	Coriell NA09888 (f)	Langer-Giedion syndrome deletion.
	15q11.2 region	Coriell NA21887 (f)	Angelman syndrome deletion.
	17p13.3 region	Coriell NA06047 (m)	Miller-Dieker syndrome deletion.
	17p11.2 region	Coriell NA13476 (f)	Smith-Magenis syndrome deletion.
	22q11.21 region	Coriell NA17942 (m)	DiGeorge syndrome deletion (AB, BC & CD regions).
P065-C & P066-C	FBN1	Coriell NA03184 (m)	Large heterozygous duplication (82 Mb) that includes the complete FBN1 gene.
		Coriell NA21940 (f)	Heterozygous FBN1 exon 45-47 deletion.
		Coriell NA21939 (f)	Heterozygous FBN1 exon 43-44 deletion.
	TGFBR2	Coriell NA04127 (f)	Heterozygous duplication of the complete TGFBR2 gene. This cell line has a partial trisomy of the 3p arm.
P070-B	1p	Coriell NA22991 (f)	Heterozygous deletion affecting the TNFRSF18 probe.
	2p & 4q	Coriell NA00501 (m)	Heterozygous deletion affecting the ACP1 probe and heterozygous duplication affecting the FRG1 probe.
	5p	Coriell NA14131 (f)	Heterozygous deletion affecting the CCDC127 probe.
	17p	Coriell NA06047 (m)	Heterozygous deletion affecting the RPH3AL probe.
P072-D	2p16.3-p21 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for EPCAM, MSH2, KCNK12 and MSH6, and two reference probes at 155 nt and 190 nt.
		Coriell NA13451 (m)	Heterozygous deletion affecting the probes for EPCAM, MSH2, KCNK12 and MSH6.
	MUTYH	Coriell HG00097 (f), HG01095 (f) and HG01519 (f) are positive for the MUTYH c.1187G>A (p.Gly396Asp) mutation.	
		Coriell HG01918 (f)	Positive for the MUTYH p.Y179C mutation.
P073-A	IKBKG	Coriell NA19225 (f)	Heterozygous deletion of IKBKG exon 4-10 (IKBKGexon4_10del mutation).
P077-B	13q13.1 region	Coriell NA02718 (f)	Heterozygous deletion affecting the probes for BRCA2.
		Coriell NA03330 (m)	Heterozygous duplication affecting the probes for BRCA2.
		Coriell NA12606 (m)	Heterozygous duplication affecting the probes for BRCA2.
P078-D	6q25 region	Coriell NA07994 (m)	Heterozygous duplication of ESR1.
	7p11 region	Coriell NA07081 (m)	Heterozygous duplication of EGFR.
	8p/q	Coriell NA02030 (m)	Heterozygous duplication of ZNF703, FGFR1, ADAM9, IKBKB, PRDM14, MTDH and MYC.
	8p12-p11 region	Coriell NA14485 (m)	Heterozygous duplication of ZNF703, FGFR1, ADAM9 and IKBKB.
	8q13-q24 region	Coriell NA03999 (f)	Heterozygous deletion of MYC.
	11q13 region	Coriell NA00959 (m)	Heterozygous duplication of CCND1 and EMSY.
	16q22 region	Coriell NA12074 (m)	Heterozygous deletion of CDH1.
	17q11-q25 region	Coriell NA16445 (m)	Heterozygous duplication of BIRC5.
	20q13 region	Coriell NA08123 (m)	Heterozygous duplication of AURKA.
	Various	DSMZ ACC-180 (HEP-G2) (m)	Gains of ESR1, CDH1, MED1, ERBB2, CDC6, TOP2A, MAPT, PPM1D, BIRC5 and AURKA.
		DSMZ ACC-432 (8-MG-BA) (f)	Gains of EGFR, PRDM14, MTDH, MYC, CCND1, EMSY, MAPT, PPM1D, BIRC5, CCNE1 and AURKA.
P083-D	CDH1	Coriell NA12074 (m)	Heterozygous deletion affecting the probes for CDH1 gene. The flanking probes are not affected.
		Coriell NA19092 (m)	Heterozygous duplication affecting the probes for CDH1 exon 4-16 and CHD1 downstream probe. The flanking probes are not affected.
P087-C/D	BRCA1	Coriell NA14626 (f)	Heterozygous BRCA1 exon 13 duplication.
		Coriell NA18949 (f)	Heterozygous BRCA1 exon 15-16 deletion.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P088-D	1p36.33-1p36.32 region & 9p21.3 region	Coriell NA22976 (m)	Heterozygous deletion affecting the probes for GNB1, TNFRSF14 and TP73. Heterozygous duplication of CDKN2A and CDKN2B.
	1q31.3-1q32.1 region & 9p21.3 region	DSMZ ACC-255 (CADO-ES1) (f)	Heterozygous duplication affecting the probes for CRB1 and TNNT2. Heterozygous deletion of CDKN2A and CDKN2B. Some of the reference probes are affected by CNAs.
	1p34.1-1p33 region & 1p31.3-1p21.3 region & 19q region	DSMZ ACC-397 (HT-1376) (f)	Heterozygous duplication affecting the probes for MUTYH, PRDX1, FAF1, CDKN2C, MIR101, FUBP1, GTF2B and DPYD. Heterozygous deletion affecting the probes on chromosome 19q. Some of the reference probes and denaturation control fragments and are affected by CNAs, which might lead to warnings for quality scores (e.g. RPQ, DNA denaturation and CAS)
	1p region & 9p21.3 region	DSMZ ACC-778 (LOPRA-1) (f)	Heterozygous deletion affecting the probes on chromosome 1p and the probes for CDKN2A and CDKN2B. Homozygous deletion affecting the probes for FAF1 and CDKN2C. Some of the reference probes and denaturation control fragments and are affected by CNAs, which might lead to warnings for quality scores (e.g. RPQ, DNA denaturation and CAS)
P089-B	RRM2B	Coriell NA02030 (m)	Heterozygous duplication of RRM2B.
	SUCLA2	Coriell NA02718 (f)	Heterozygous deletion of SUCLA2.
		Coriell NA03330 (m)	Heterozygous duplication of SUCLA2.
	TK2	Coriell NA19401 (f)	Heterozygous TK2 exon 1-2 deletion.
	Various	Coriell NA10401 (f)	Heterozygous duplication of MPV17, DGUOK and SUCLG1.
P090-C	13q13.1 region	Coriell NA02718 (f)	Heterozygous deletion affecting the probes for BRCA2 and N4BP2L1.
		Coriell NA03330 (m)	Heterozygous duplication affecting the probes for BRCA2 and N4BP2L1.
		Coriell NA12606 (m)	Heterozygous duplication affecting the probes for BRCA2 and N4BP2L1.
P091-D	CFTR	Coriell HG01565 (m)	Heterozygous deletion of the complete CFTR gene. The flanking probes are not affected.
		Coriell HG02461 (m)	Heterozygous CFTR exon 19-21 deletion.
		Coriell HG04131 (m)	Heterozygous CFTR exon 4-11 deletion.
		Coriell NA01059 (f)	Heterozygous deletion of the complete CFTR gene. The flanking probes are also affected.
		Coriell NA01531 (m)	Homozygous positive for the CFTR ΔF508 mutation.
		Coriell NA11277 (m)	Heterozygous positive for the CFTR ΔI507 mutation.
		Coriell NA12519 (f)	Heterozygous triplication of the complete CFTR gene (4 copies). The flanking probes are also affected.
		Coriell NA18668 (m)	Heterozygous CFTR exon 2-3 deletion; heterozygous positive for the CFTR ΔF508 mutation.
P093-C	2q32.1-q33.2 region	Coriell NA01229 (f)	Heterozygous duplication affecting all BMPR2 probes.
		Coriell NA11213 (f)	Heterozygous deletion affecting all BMPR2 probes.
	9q34.11 region	Coriell NA10186 (m)	Heterozygous duplication affecting all ENG probes and one reference probe at 290 nt.
P095-A	Chromosome 13	Coriell NA02948 (m)	Trisomy of chromosome 13 affecting all probes on this chromosome.
	Chromosome 18	Coriell NA02422 (f)	Trisomy of chromosome 18 affecting all probes on this chromosome.
	Chromosome 21	Coriell AG05024 (f)	Trisomy of chromosome 21 affecting all probes on this chromosome.
	Chromosomes X & Y	Coriell NA04375 (m)	Klinefelter variant 48, XXYY. All probes on chromosomes X and Y are affected.
P098-E	13q14.3 region	Coriell NA03330 (m)	Heterozygous duplication affecting the probes for ATP7B and ALG11.
		Coriell NA05258 (f)	Heterozygous positive for the ATP7B H1069Q mutation in exon 14.
		Coriell NA12606 (m)	Heterozygous duplication affecting the probes for ATP7B and ALG11.
		Coriell NA13721 (m)	Heterozygous deletion affecting the probes for ATP7B and ALG11.
		Coriell NA14164 (f)	Heterozygous deletion affecting the probes for ATP7B and ALG11.
P099-D	GCH1	Coriell NA05966 (m)	Heterozygous duplication affecting the GCH1 probes.
	PRRT2	Coriell NA05875 (f)	Heterozygous deletion affecting the PRRT2 probes.
	SGCE	Coriell NA10160 (m)	Heterozygous deletion affecting the SGCE probes.
P102-C/D	HBB	Coriell NA06342 (m)	Heterozygous HBG1 intron 2 to HBB intron 1 (Hb Kenya-gamma); heterozygous positive for the HBB c.20A>T mutation (HbS).
		Coriell NA20480 (m)	Heterozygous HBD intron 1 to HBB intron 1 (Hb Lepore-Baltimore).
		Coriell NA16267 (m)	Heterozygous positive for the HBB c.20A>T mutation (HbS).

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P103-C	DPYD	Coriell HG02684 (m)	Homozygous positive for the DPYD c.1905+1G>A (IVS14+1G>A) mutation.
		Coriell NA07048 (m)	Heterozygous positive for the DPYD c.2846A>T mutation.
		Coriell NA21112 (m)	Heterozygous positive for the DPYD c.1905+1G>A (IVS14+1G>A) mutation.
P105-D	7p11.2 region	Coriell NA07081 (m)	Heterozygous duplication affecting the probes for EGFR.
	9p21.3 region	Coriell NA01750 (m)	Heterozygous duplication affecting the probes for CDKN2A.
		Coriell NA02819 (f)	Heterozygous duplication affecting the probes for CDKN2A.
		Coriell NA03226 (m)	Heterozygous duplication affecting the probes for CDKN2A.
		Coriell NA05067 (m)	Heterozygous duplication affecting the probes for CDKN2A.
	10q23.31 region	Coriell NA20125(m)	Heterozygous duplication affecting the probes for PTEN.
	14q13.2 region	Coriell NA06801 (f)	Heterozygous duplication affecting the probes for NFKBIA.
	10q23.31 region 17p13.1 region	DSMZ ACC-203 (SK-N-MC) (f)	Heterozygous deletion affecting the probes for PTEN. Heterozygous deletion affecting the probes for TP53, homozygous deletion affecting the TP53 exon 2a probe. Some of the reference probes are affected by CNAs.
	4q12 region 9p21.3 region 10q23.31 region 12q14-q15 region 14q13.2 region 17p13.1 region	DSMZ ACC-237 (IGR-37) (m)	Heterozygous deletion affecting the probes for PDGFRA, CDKN2A exons 1 and 2, PTEN, CDK4, MIR26A2, MDM2, NFKBIA and TP53. Homozygous deletion affecting the probes for CDKN2A exons 3 and 4.
	4q12 region 12q14-q15 region 14q13.2 region 17p13.1 region	DSMZ ACC-243 (SAOS-2) (f)	Heterozygous duplication affecting the probes for PDGFRA. Homozygous duplication affecting the probes for CDK4, MIR26A2, MDM2 and NFKBIA. and TP53. Homozygous deletion affecting the probes for TP53 exon 2a-11. Some of the reference probes are affected by CNAs.
P114-C	KCNE1/KCNE2	Coriell NA13031 (m)	Heterozygous duplication that affects all KCNE1 and KCNE2 probes.
	KCNH2	Coriell NA01220 (f)	Heterozygous duplication that affects all KCNH2 probes.
		Coriell NA07412 (m)	Heterozygous deletion that affects all KCNH2 probes.
		Coriell NA08808 (m)	Heterozygous deletion that affects all KCNH2 probes.
		Coriell NA12519 (f)	Homozygous duplication or heterozygous triplication that affects all KCNH2 probes.
	KCNQ1	Coriell NA03435 (m)	Heterozygous duplication that affects all KCNQ1 probes.
P118-C	WT1	Coriell NA05518 (f)	Heterozygous deletion of the WT1 gene. The flanking probes are also affected.
		Coriell NA06803 (m)	Heterozygous deletion of the WT1 gene. The flanking probes are also affected.
		Coriell NA09709 (m)	Heterozygous deletion of the WT1 gene. The flanking probes are also affected.
P124-C	9q34.13 region	Coriell NA13685 (f)	Heterozygous duplication affecting the probes for TSC1.
P125-C	mtDNA	Coriell NA11605 (f)	Positive for the 3460G>A mutation (~95% mutation/~5% wild-type).
		Coriell NA11906 (f)	Positive for the 8344A>G mutation (~35% mutation/~65% wild-type)
		Coriell NA11907 (f)	Positive for the 8344A>G mutation (~98% mutation/~2% wild-type).
		Coriell NA13411 (m)	Positive for the 8993T>G mutation (100% mutation).
		Coriell NA10744 (m)	Positive for the 11778G>A mutation (100% mutation).
P137-B/C	SCN1A	Coriell NA10401 (f)	Heterozygous duplication of the complete SCN1A gene.
		Coriell NA10607 (m)	Heterozygous deletion of the complete SCN1A gene.
P140-C	HBA	Coriell CD00026 (m)	Heterozygous HBQ1-HBM deletion ($--^{SEA}$).
		Coriell CD00027 (f)	Heterozygous HBQ1-HBM deletion ($--^{SEA}$).
		Coriell GM18933 (f)	Homozygous $\alpha^{3.7}$ deletion (type D mentioned in the product description).
		Coriell HG02188 (f)	Heterozygous positive for the Hb Constant Spring mutation.
		Coriell NA02325 (f)	Heterozygous duplication affecting all target probes.
		Coriell NA09687 (m)	Heterozygous deletion affecting all target probes.
		Coriell NA10797 (m)	Compound heterozygous $--^{SEA}/--^{FIL}$ deletions (homozygous HBA deletion).
		Coriell NA10798 (f)	Heterozygous HBQ1-HBZ deletion ($--^{FIL}$).
		Coriell NA10799 (m)	Heterozygous deletion of probes 184 nt to 400 nt ($=--^{SEA}$ deletion).
		Coriell NA19122 (f)	Homozygous $\alpha^{3.7}$ deletion (type D mentioned in the product description).
		Coriell NA19176 (f)	Heterozygous $\alpha^{3.7}$ deletion (type D mentioned in the product description).
P143-C	MPZ	Coriell NA00803 (m)	Heterozygous deletion affecting all MPZ probes.
P147-B	1p36 region	Coriell NA22995 (m)	1p36 deletion syndrome; heterozygous telomeric deletion (4.67-5.97 Mb).

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P155-E	COL3A1	Coriell NA03918 (f)	Heterozygous deletion affecting all COL3A1 probes.
		Coriell NA10401 (f)	Heterozygous duplication affecting all COL3A1 probes.
	TNXB	Coriell NA11213 (f)	Heterozygous deletion affecting all COL3A1 probes, and heterozygous duplication affecting TNXB exon 35 and CYP21A2 probes.
P156-B	GALT	Coriell NA05067 (m)	Large heterozygous duplication (6.5 Mb) that includes the complete GALT gene.
		Coriell NA01741 (m)	Homozygous deletion of the complete GALT gene. The flanking probes are not affected.
P158-D	SMAD4	Coriell NA01359 (m)	Heterozygous duplication affecting all SMAD4 probes.
		Coriell NA03623 (f)	Heterozygous duplication affecting all SMAD4 probes.
		Coriell NA07891 (m)	Heterozygous deletion affecting all SMAD4 probes.
	BMPR1A/PTEN	Coriell NA20125 (m)	Heterozygous duplication affecting all BMPR1A and PTEN probes.
P163-D/E	4p16 region	Coriell NA22601 (m)	Heterozygous deletion affecting the probes for WFS1.
	13q12 region	Coriell NA23835 (m)	Heterozygous positive for the GJB2 35delG and the 101T>C mutations.
		Coriell HG00478 (m)	Heterozygous positive for the GJB2 c.235delC mutation.
		Coriell NA12606 (m)	Large heterozygous duplication (42 Mb) affecting the probes for the GJB2 and GJB6, and flanking probes.
P165-C	SPAST	Coriell HG00500 (m)	Heterozygous duplication affecting probes for SPAST exon 4-17.
		Coriell NA10401 (f)	Heterozygous duplication affecting all SPAST probes.
P175-B	1q32.1 region	Coriell NA05347 (m)	Heterozygous duplication affecting the probes for MDM4.
	2p24.3-p23.2 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for MYCN and ALK. Some of the reference probes are affected by CNAs.
	2p24.3 region	Coriell NA00945 (f)	Heterozygous deletion affecting the probes for MYCN.
	7p11.2 region	Coriell NA07081 (m)	Heterozygous duplication affecting the probes for EGFR.
	7q31.2 region	Coriell NA01059 (f)	Heterozygous deletion affecting the probes for MET.
	7q31.2-q34 region	Coriell NA12519 (f)	Homozygous duplication/Heterozygous triplication affecting the probes for MET, SMO and BRAF.
	7q34 region	Coriell NA07412 (m)	Heterozygous deletion affecting the probes for BRAF.
	8p11.23 region & 8q24.21 region	Coriell NA02030 (m)	Heterozygous duplication affecting the probes for FGFR1 and MYC.
	8q24.21 region	Coriell NA03999 (f)	Heterozygous deletion affecting the probes MYC.
	9q34.12 region	Coriell NA13685 (f)	Heterozygous duplication affecting the probes for ABL1.
	12p13.32 region	Coriell NA07981 (m)	Homozygous duplication/Heterozygous triplication affecting the probes for CCND2.
	20q13.2 region	Coriell NA08123 (m)	Heterozygous duplication affecting the probes for AURKA.
	Xq12 region	Coriell NA03384 (m)	Homozygous duplication/Heterozygous triplication affecting the probes for AR.
	1q32.1 region & 7q34 region	DSMZ ACC-427 (DU-4475) (f)	Homozygous duplication / Heterozygous triplication affecting the probes for MDM4. Positive for the BRAF c.1799T>A (p.V600E) mutation. Ambiguous ratios are observed for a gain of 7q32.1-7q34 (including MET, SMO and BRAF genes).
	7p11.2 region & 7q31.2-q34 region & 12p13.32 region & 12q14.1 & 20q13.2 region	DSMZ ACC-573 (SU-DHL-8)(m)	Heterozygous duplication affecting the probes for EGFR, MET, SMO, BRAF, CCND2 and CDK4. Homozygous duplication / Heterozygous triplication affecting the probes for AURKA. Some of the reference probes are affected by CNAs.
P178-B	Xq28	Coriell NA02325 (f)	Heterozygous duplication affecting the probes for F8 exons 1-22.
		Coriell NA07503 (f)	Heterozygous deletion affecting the probes for F8.
P183-C	1q42.3	Coriell NA05347 (m)	Heterozygous duplication affecting the probes for EDARADD.
		Coriell NA10020 (f)	Heterozygous deletion affecting the probes for EDARADD.
	2q35	Coriell NA01229 (f)	Heterozygous duplication affecting the probes for WNT10A.
		Coriell NA10918 (f)	Heterozygous deletion affecting the probes for WNT10A.
	2q13-q35	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for EDAR and WNT10A.
P189-C	CDKL5	Coriell NA23710 (f)	Heterozygous deletion affecting the probes for CDKL5 intron 16-exon 18.
	FOXG1	Coriell NA01750 (m)	Heterozygous duplication affecting the probes for FOXG1.
		Coriell NA22765 (m)	Heterozygous deletion affecting the probes for FOXG1.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P190-D	ATM	Coriell HG03694 (m)	Heterozygous duplication affecting the probes for ATM exon 62-63.
		Coriell NA08618 (m)	Heterozygous duplication affecting all ATM probes.
	CHEK2	Coriell HG00187 (m), HG00266 (f), HG00275 (f), HG00371 (m) and NA19707 (f) are positive for the CHEK2 1100delC mutation.	
		Coriell HG00343 (f)	Heterozygous deletion affecting the probes for CHEK2 exon 9-10.
		Coriell HG01872 (m)	Heterozygous duplication affecting the probes for CHEK2 exon 3-5.
		Coriell NA07106 (m)	Heterozygous duplication affecting the probes for CHEK2 and HSCB.
P198-A	FH	Coriell NA03648 (m)	Heterozygous duplication affecting the probes for FH.
		Coriell NA05347 (m)	Heterozygous duplication affecting the probes for FH.
		Coriell NA06473 (f)	Heterozygous deletion affecting the probes for FH.
		Coriell NA10020 (f)	Heterozygous deletion affecting the probes for FH.
P199-B	HEXA	Coriell NA00502 (m)	Heterozygous positive for the HEXA c.1274_1277dupTATC (1278insTATC) and c.1421+1G>C (IVS12+1G>C) mutations.
		Coriell NA03184 (m)	Heterozygous duplication of the complete HEXA gene.
P202-C	14q32.33	Coriell NA08123 (m)	Heterozygous deletion that includes the probes on 14q32.33 (CEP170B, MTA1, CRIP2, and IGHD).
	ERG	Coriell NA09868 (f)	Heterozygous deletion that includes the complete ERG gene and its flanking probes.
	CDKN2A/2B	Coriell NA01750 (m)	Heterozygous duplication that includes CDKN2A and CDKN2B.
	IKZF1	Coriell NA10925 (m)	Heterozygous deletion that includes the complete IKZF1 gene and it flanking probes.
		Coriell NA07081 (m)	Heterozygous duplication that includes the complete IKZF1 gene and it flanking probes.
P207-C	13q34	Coriell NA03089 (f)	Heterozygous duplication affecting the probes for F7.
		Coriell NA03330 (m)	Heterozygous duplication affecting the probes for F7.
		Coriell NA03887 (f)	Heterozygous deletion affecting the probes for F7.
		Coriell NA05832 (m)	Heterozygous duplication affecting the probes for F7.
	Xq28	Coriell NA02325 (f)	Heterozygous duplication affecting the probes for F8 exon 1-14.
P213-B	REEP1	Coriell HG00246 (m)	Heterozygous REEP1 exon 3-8 triplication.
	SPG7	Coriell HG02128 (m)	Heterozygous SPG7 exon 1-7 deletion. The upstream flanking probe is also affected.
		Coriell NA21108 (f)	Heterozygous duplication of the complete SPG7 gene. The upstream flanking probe is also affected.
P221-C	CRB1	Coriell NA00214 (m)	Heterozygous deletion of the complete CRB1 gene.
	CRX	Coriell HG02397 (m)	Heterozygous duplication of the complete CRX gene.
	LCA5	Coriell HG01802 (f)	Heterozygous duplication of the complete LCA5 gene.
		Coriell NA10946 (m)	Heterozygous deletion of the complete LCA5 gene.
P225-E	PTEN	Coriell NA20125 (m)	Heterozygous duplication of the complete PTEN gene. Several flanking probes (between BMPR1A and HTRA1) are also affected.
		DSMZ ACC-50 (OPM-2) (f)	Homozygous PTEN exon 3-7 deletion. This cell line also has several other aberrations.
P226-D	SDHC	Coriell NA00803 (m)	Heterozygous deletion affecting all SDHC probes.
		Coriell NA17941 (f)	Heterozygous duplication affecting all SDHC probes.
	SDHAF2	Coriell NA20775 (f)	Heterozygous duplication affecting the probes for SDHAF2 exon 2-4.
	SDHD	Coriell NA15099 (m)	Heterozygous duplication affecting all SDHD probes.
P229-B	7p12.2 region	Coriell NA03563 (m)	Heterozygous duplication affecting the probes for OPA1.
P236-B	1q31.3 region	Coriell NA00214 (m)	Heterozygous deletion affecting the probes for CFH, CFHR1, CFHR2, CFHR3, CFHR4 and CFRH5.
		Coriell NA00501 (m)	Heterozygous deletion affecting the probes for CFHR3 and CFHR4. Homozygous deletion affecting the probes for CFHR1.
		Coriell HG01770 (f)	Heterozygous deletion affecting the probes for CFHR3 and CFHR1.
		Coriell HG01894 (f)	Heterozygous deletion affecting the probes for CFHR3 and CFHR1.
P241-E	7p13	Coriell NA07081 (m)	Heterozygous duplication affecting all GCK probes.
		Coriell NA10925 (m)	Heterozygous deletion affecting all GCK probes.
		Coriell NA10951 (f)	Heterozygous deletion affecting all GCK probes.
	17q12	Coriell NA20359 (f)	Heterozygous duplication affecting all HNF1B probes.
	20q13	Coriell NA07945 (m)	Heterozygous deletion affecting all HNF4A probes.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P242-C	PRSS1	Coriell NA11949 (f)	Heterozygous deletion affecting all PRSS1 probes.
	7q34 region	Coriell NA07412 (m)	Heterozygous deletion affecting the probes for BRAF, PRSS1 and CASP2.
		Coriell NA12519 (f)	Homozygous duplication affecting the probes for BRAF, PRSS1 and CASP2.
P244-C/D	CDKN1B	Coriell NA07981 (m)	Four copies of all probes for CDKN1B. The flanking probes are also affected.
P245-B	1p36 region	Coriell NA22995 (m)	1p36 deletion syndrome; heterozygous telomeric deletion (4.67-5.97 Mb).
	2q32-q33 region	Coriell NA11213 (f)	Glass syndrome deletion.
	3q29 region	Coriell NA11428 (f)	3q29 microduplication syndrome.
	4p16.3 region	Coriell NA04126 (m)	Wolf-Hirschhorn syndrome deletion.
	5p15 region	Coriell NA16593 (f)	Cri-du-Chat syndrome deletion (affecting one probe).
	7q11.23 region	Coriell NA13464 (m)	Williams-Beuren syndrome deletion (commonly deletion region).
	15q11.2 region	Coriell NA20375 (m)	Angelman syndrome.
	17p11.2 region	Coriell NA13476 (f)	Smith-Magenis syndrome deletion.
	17p13.3 region	Coriell NA09208 (m)	Miller-Dieker syndrome deletion.
	22q11.21 region	Coriell NA02944 (m)	DiGeorge / 22q11 syndrome deletion (region AB).
	Xq28 region	Coriell NA23675 (m)	MECP2 duplication syndrome.
		Coriell NA23676 (f)	MECP2 duplication syndrome.
		Coriell NA23635 (f)	Rett syndrome deletion (affecting one probe).
P248-B	MLH1/MSH2	The NIBSC Institute provides a kit with 5 DNA samples containing heterozygous MLH1 or MSH2 exon deletions or amplifications (catalog number 11/218-XXX).	
P250-B	4q35 region	Coriell NA00501 (m)	Heterozygous duplication affecting the probes for SLC25A4 and KLKB1.
		Coriell NA03013 (f)	Heterozygous deletion affecting the probes for SLC25A4 and KLKB1.
		Coriell NA10313 (m)	Heterozygous duplication affecting the probes for SLC25A4 and KLKB1.
	8p23 region	Coriell NA02030 (m)	Heterozygous duplication affecting the probes for PPP1R3B, MSRA and GATA4.
		Coriell NA03255 (m)	Heterozygous duplication affecting the probes for PPP1R3B, MSRA and GATA4.
		Coriell NA12721 (f)	Heterozygous deletion affecting the probes for PPP1R3B, MSRA and GATA4.
	9q34 region	Coriell NA13685 (f)	Heterozygous duplication affecting the probes for EHMT1.
	10p14 region	Coriell NA06936 (f)	Heterozygous deletion affecting the probes for GATA3, TCEB1P3 and CELF2.
	17p13.3 region	Coriell NA06047 (m)	Heterozygous deletion affecting the probes for RPH3AL, GEMIN4 and YWHAE.
		Coriell NA09208 (m)	Heterozygous deletion affecting the probes for RPH3AL, GEMIN4 and YWHAE.
	22q11.2 region	Coriell NA02944 (m)	Heterozygous deletion affecting the probes in the Cat Eye Syndrome region, and LCR-A to LCR-B region.
		Coriell NA05401 (m)	Heterozygous deletion affecting the probes in the Cat Eye Syndrome region, and LCR-A to LCR-B region with the exception of the probe for DGCR8.
		Coriell NA07215 (f)	Heterozygous deletion affecting the probes in the LCR-A to LCR-D region.
		Coriell NA10382 (m)	Heterozygous deletion affecting the probes in the LCR-A to LCR-D region.
		Coriell NA17942 (m)	Heterozygous deletion affecting the probes in the LCR-A to LCR-D region.
	22q11.2 region & 22q13 region	Coriell NA07106 (m)	Heterozygous duplication affecting the probes on chromosome 22.
	22q13.33 region	Coriell NA13284 (m)	Heterozygous duplication affecting the probes for ARSA and SHANK3.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P251-C	GABRD	Coriell NA18827 (f)	Heterozygous deletion affecting the GABRD probe.
		Coriell NA22977 (f)	Heterozygous deletion affecting the GABRD probe.
	1p36.32-p36.33 region	Coriell NA22991 (f)	Heterozygous deletion affecting the probes for GABRD and TP73.
		Coriell NA22995 (m)	Heterozygous deletion affecting the probes for GABRD and TP73.
	1p36.22-p36.31 region	Coriell NA50276 (m)	Heterozygous deletion affecting the probes for CHD5, PARK7 and KIF1B.
	1q21.2-q44 region	Coriell NA17941 (f)	Heterozygous duplication affecting the probes for LHX4, LIN9 and AKT3.
	LHX4	Coriell NA00214 (m)	Heterozygous deletion affecting the LHX4 probe.
		Coriell NA06038 (m)	Heterozygous deletion affecting the LHX4 probe.
	1q42.12-q44 region	Coriell NA05347 (m)	Heterozygous duplication affecting the probes for LIN9 and AKT3.
	AKT3	Coriell NA06473 (f)	Heterozygous deletion affecting the AKT3 probe.
	VHL	Coriell NA03503 (m)	Heterozygous duplication affecting the VHL probe.
		Coriell NA10985 (f)	Heterozygous deletion affecting the VHL probe.
	3p21.31-p25.3 region	Coriell NA04127 (f)	Heterozygous duplication affecting the probes for VHL, TGFBR2, CTNNB1, SEMA3B, RASSF1 and ZMYND10.
	CASR	Coriell NA08778 (m)	Heterozygous deletion affecting the CASR probe.
	3q21.1-q26.32 region	Coriell NA03563 (m)	Heterozygous duplication affecting the probes for CASR, ZIC1 and PIK3CA.
	3q24-q26.32 region	Coriell NA11428 (f)	Heterozygous duplication affecting the probes for ZIC1 and PIK3CA.
		Coriell NA20022 (m)	Heterozygous duplication affecting the probes for ZIC1 and PIK3CA.
	PIK3CA	Coriell NA10175 (m)	Heterozygous duplication affecting the PIK3CA probe.
	CD44	Coriell NA09709 (m)	Heterozygous deletion affecting the CD44 probe.
	PTPRJ	Coriell NA22633 (m)	Heterozygous deletion affecting the PTPRJ probe.
	11q13.2-q23.3 region	Coriell NA00959 (m)	Heterozygous duplication affecting the probes for GSTP1, CNTN5, CASP1, ATM, CADM1, KMT2A, HMBS and THY1.
	11q22.1-q22.3 region	Coriell NA08618 (m)	Heterozygous duplication affecting the probes for CNTN5, CASP1 and ATM.
		Coriell NA09596 (m)	Heterozygous deletion affecting the probes for CNTN5, CASP1, ATM.
	11q22.1-q23.3 region	Coriell NA15099 (m)	Heterozygous duplication affecting the probes for CNTN5, CASP1, ATM, CADM1, KMT2A, HMBS and THY1.
P252-D	TMEM18	Coriell NA00501 (m)	Heterozygous deletion affecting the TMEM18 probe.
	TPO	Coriell NA10951 (f)	Heterozygous duplication affecting the TPO probe.
	2p25.3-q33.1 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for TMEM18, TPO, NBAS, DDX1, MYCN, ALK, RTN4, DYSF, RPIA, SCN1A, CFLAR, CASP8 and BMPR2.
	2p24.3-p25.3 region	Coriell NA04409 (m)	Heterozygous duplication affecting the probes for TMEM18, TPO, NBAS, DDX1 and MYCN.
	2p24.3 region	Coriell NA00945 (f)	Heterozygous deletion affecting the probes for NBAS, DDX1 and MYCN.
		Coriell NA09216 (m)	Heterozygous deletion affecting the probes for NBAS, DDX1 and MYCN.
	SCN1A	Coriell NA10607 (m)	Heterozygous deletion affecting the SCN1A probe.
	2q33.1 region	Coriell NA01229 (f)	Heterozygous duplication affecting the probes for CFLAR, CASP8 and BMPR2.
		Coriell NA11213 (f)	Heterozygous deletion affecting the probes for CFLAR, CASP8 and BMPR2.
	PAFAH1B1	Coriell NA06047 (m)	Heterozygous deletion affecting the PAFAH1B1 probe.
	TOB1	Coriell NA13031 (m)	Heterozygous deletion affecting the TOB1 probe.
	17q25.3 region	Coriell NA16445 (m)	Heterozygous duplication affecting the probes for BIRC5, SECTM1 and TBCD.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P253-D	4p15.31-p16.3 region	Coriell NA03435 (m)	Heterozygous deletion affecting the probes SPON2, WSF1 and KCNIP4.
		Coriell NA10947 (f)	Heterozygous duplication affecting the probes for SPON2, WSF1 and KCNIP4.
	4p13.2-q27 region	Coriell NA00782 (m)	Heterozygous duplication affecting the probes for GNRHR and IL2.
	4q27-q35.2 region	Coriell NA00501 (m)	Heterozygous duplication affecting the probes for IL2, GLRB and KLKB1.
	4q32.1-q35.2 region & 7q36.3 region	Coriell NA10313 (m)	Heterozygous duplication affecting the probes for GLRB and KLKB1. Heterozygous deletion affecting the SHH probe.
	4q32.1-q35.2 region	Coriell NA03013 (f)	Heterozygous deletion affecting the probes for GLRB and KLKB1.
	GHRHR	Coriell NA08763 (m)	Heterozygous deletion affecting the GHRHR probe.
	7p11.2-p15.1 region	Coriell NA07081 (m)	Heterozygous duplication affecting the probes for GHRHR and EGFR.
	ELN	Coriell NA12590 (m)	Heterozygous deletion affecting the ELN probe.
	7q11.23-q21.2 region	Coriell NA10160 (m)	Heterozygous deletion affecting the probes for ELN and KRIT1.
	IMPDH1	Coriell NA12519 (f)	Homozygous duplication affecting the IMPDH1 probe.
	SHH	Coriell NA01220 (f)	Heterozygous duplication affecting the SHH probe.
	PTPRD	Coriell NA10989 (m)	Heterozygous deletion affecting the probes for PTPRD.
	9p21.3-p24.1 region	Coriell NA01750 (m)	Heterozygous duplication affecting the probes for PTPRD and CDKN2A.
	9p13.3-p24.1 region	Coriell NA02819 (f)	Heterozygous duplication affecting the probes for PTPRD, CDKN2A and DNAI1.
	9q34.13 region	Coriell NA13685 (f)	Heterozygous duplication affecting the probes for POMT1 and TSC1.
	12p11.21-p13.33 region	Coriell NA07981 (m)	Homozygous duplication affecting the probes for ERC1, CDKN1B and PKP2.
	NFKBIA	Coriell NA06801 (f)	Heterozygous duplication affecting the NFKBIA probe.
	ALT	Coriell NA09888 (f)	Heterozygous deletion affecting the ALT probe.
P256-C	FLCN	Coriell NA08146 (f), NA13476 (f), NA18319 (f), NA18320 (f), NA18322 (f), NA18324 (m), NA18326 (m) and NA20743 (m) have a heterozygous deletion that affects all FLCN probes.	
P258-C	SMARCB1	Coriell NA02325 (f)	Heterozygous duplication affecting the probes for TBX1, DGCR8, SNAP29, LZTR1, PPIL2, GNAZ, SMARCB1, SNRPD3, SEZ6L and NIPSNAP1.
		Coriell NA07106 (m)	Heterozygous duplication affecting the probes for TBX1, DGCR8, SNAP29, LZTR1, PPIL2, GNAZ, SMARCB1, SNRPD3, SEZ6L and NIPSNAP1.
		DSMZ ACC-808 (KP-363T) (m)	Heterozygous deletion affecting the probes for TBX1, DGCR8, SNAP29, LZTR1, PPIL2, GNAZ, SMARCB1, SNRPD3, SEZ6L and NIPSNAP1. Some of the reference probes are affected by CNAs.
P260-C	05q31.1 region	Coriell NA14230 (m)	Heterozygous deletion affecting the probes for RAD50.
	16p12.2 region	Coriell NA08039 (m)	Heterozygous duplication affecting the probes for PALB2.
		Coriell HG00634 (m)	Heterozygous duplication affecting the PALB2 exon 13 probe.
		Coriell HG03857 (f)	Heterozygous deletion affecting the probes for PALB2 exon 5-7.
P278-D	PCCA	Coriell NA06312 (m)	Heterozygous deletion of the complete PCCA gene.
		Coriell NA22208 (f)	Heterozygous PCCA exon 13-20 deletion.
P292-B	PCDH15	Coriell NA11672 (m)	Heterozygous deletion affecting all PCDH15 probes.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P294-C	1p36.32-p36.33 region	Coriell NA22991 (f)	Heterozygous deletion affecting the probes for TNFRSF4 and PRDM16.
	1p36.22-p36.31 region	Coriell NA50276 (m)	Heterozygous deletion affecting the probes for CHD5, CAMTA1 and KIF1B.
	1p36.22-p36.33 region & 3p14.2-p25.3 region & 10q23.31 region & 17p13.1 region	DSMZ ACC-203 (SK-N-MC) (f)	Subclonal gain (ratios around 1.3) affecting the probes for TNFRSF4, PRDM16, CHD5, CAMTA1 and KIF1B. Heterozygous deletion affecting the probes for VHL, FHIT, PTEN and TP53. Some of the reference probes are affected by CNAs.
	3p25.3 region	Coriell NA03503 (m)	Heterozygous duplication affecting the probes for VHL.
		Coriell NA10985 (f)	Heterozygous deletion affecting the probes for VHL.
	5q22.2 region	Coriell NA14234 (m)	Heterozygous deletion affecting the probes for APC.
	9p21.3 region	Coriell NA02819 (f)	Heterozygous duplication affecting the probes for CDKN2A and CDKN2B.
	9q22.32 region	Coriell NA09834 (f)	Heterozygous deletion affecting the probes for PTCH1.
	9q34.13 region	Coriell NA13685 (f)	Heterozygous duplication affecting the probes for TSC1.
	10q23.31 region	Coriell NA20125 (m)	Heterozygous duplication affecting the probes for PTEN.
	11p13 region	Coriell NA09709 (m)	Heterozygous deletion affecting the probes for WT1.
	13q13.1-q14.2 region	Coriell NA12606 (m)	Heterozygous duplication affecting the probes for BRCA2, RB1, MIR15A and DLEU1.
	13q14.2-q14.3 region	Coriell NA14164 (f)	Heterozygous deletion affecting the probes for RB1, MIR15A and DLEU1.
	16p13.3 region & 22q11.23 region	Coriell NA02325 (f)	Heterozygous duplication affecting the probes for TSC2 and SMARCB1.
	17q21.31 region	Coriell NA18949 (f)	Heterozygous deletion affecting the BRCA1 exon 15 probe.
	18q21.2	Coriell NA07891 (m)	Heterozygous deletion affecting the probes for SMAD4.
	18q21.2 region & Xq11.1 region	Coriell NA02325 (f)	Heterozygous duplication affecting the probes for SMAD4 and AMER1.
P297-D	3q29 region	Coriell NA03563 (m)	Heterozygous duplication affecting the probes for RNF168, FBXO45, PAK2 and DLG1.
		Coriell NA10175 (m)	Heterozygous duplication affecting the probes for RNF168, FBXO45, PAK2 and DLG1.
		Coriell NA11428 (f)	Heterozygous duplication affecting the probes for RNF168, FBXO45, PAK2 and DLG1.
		Coriell NA22976 (m)	Heterozygous duplication affecting the probes for RNF168, FBXO45, PAK2 and DLG1.
	15q13.1 – 15q24.2 region	Coriell NA03184 (m)	Heterozygous duplication affecting the probes for TJP1, ARHGAP11B, FAN1, TRPM1, KLF13, OTUD7A, CHRNA7, SCG5, PML, STRA6, EDC3 and SIN3A.
	16p13.11 region	Coriell NA13685 (f)	Heterozygous deletion affecting the probes for MARF1, MYH11 and ABCC6.
	16p13.11 – 16p12.1 region	Coriell NA06226 (m)	Heterozygous duplication affecting the probes for MARF1, MYH11, ABCC6, XYLT1, UQCRC2, VWA3A and CDR2.
		Coriell NA08039 (m)	Heterozygous duplication affecting the probes for MARF1, MYH11, ABCC6, XYLT1, UQCRC2, VWA3A, CDR2, PALB2 and LCMT1.
	16p12.1 – 16p11.2 region	Coriell NA05875 (f)	Heterozygous deletion affecting the probes for IL21R, ATXN2L, RABEP2, LAT, MAZ, MVP, HIRIP3 and MAPK3 .
P298-A	BRAF	Coriell NA01220 (f)	Heterozygous duplication affecting all BRAF probes.
		Coriell NA07412 (m)	Heterozygous deletion affecting all BRAF probes.
		Coriell NA08808 (m)	Heterozygous deletion affecting all BRAF probes.
		Coriell NA12519 (f)	Homozygous duplication/Heterozygous triplication affecting all BRAF probes.
	HRAS	Coriell NA03435 (m)	Heterozygous duplication affecting all HRAS probes.
	KRAS	Coriell NA07981 (m)	Homozygous duplication/Heterozygous triplication affecting all KRAS probes.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P301-B	6p22.3 region	Coriell NA12721 (f)	Heterozygous duplication affecting the probes forE2F3 and SOX4.
	6q15 region	Coriell NA01221 (m)	Heterozygous duplication affecting the MAP3K7 probe.
	6q23.2-q23.3 region	Coriell NA09367 (f)	Heterozygous duplication affecting the probes for SGK1 and MYB.
	6q23.3-q26 region	Coriell NA07994 (m)	Heterozygous duplication affecting the probes for MYB, MYCT1 and MAP3K4.
	6q26 region	Coriell NA06802 (m)	Heterozygous deletion affecting the MAP3K4 probe.
	14q23.1-q24.3 region	Coriell NA05966 (m)	Heterozygous duplication affecting the probes for OTX2 and MLH3.
	16p11.2 region	Coriell NA05875 (f)	Heterozygous deletion affecting the TGFB1I1 probe.
	16p13.3 region	Coriell NA02325 (f)	Heterozygous duplication affecting the probes for AXIN1 and MEFV.
		Coriell NA06226 (m)	Heterozygous duplication affecting the probes for AXIN1 and MEFV.
		Coriell NA08039 (m)	Heterozygous duplication affecting the MEFV probe.
		Coriell NA13284 (m)	Heterozygous duplication affecting the AXIN1 probe.
	16p13.3 region & 16q22.3-q24.3 region	Coriell NA09687 (m)	Heterozygous deletion affecting the AXIN1 probe. Heterozygous duplication affecting the probes for ZFHX3 and FANCA.
	16q22.3 region	Coriell NA12074 (m)	Heterozygous deletion affecting the ZFHX3 probe.
	17p11.2 region	Coriell NA13476 (f)	Heterozygous deletion affecting the PRPSAP2 probe.
	17p13.2-p13.3 region	Coriell NA06047 (m)	Heterozygous deletion affecting the probes for HIC1, PAFAH1B1 and ATP2A3.
	17q25.3 region	Coriell NA16445 (m)	Heterozygous duplication affecting the probes for TK1, BIRC5, ARHGDIAand RAC3.
P302-A	2p25.3-q37.3 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for TMEM18, NBAS, MYCN, ALK, RTN4, RPIA, IL1RN, RPRM, BMPR2 and ATG4B.
	2p23.2-p25.3 region	Coriell NA01353 (m)	Heterozygous duplication affecting the probes for TMEM18, NBAS, MYCN and ALK.
	2p24.3-p25.3 region	Coriell NA04409 (m)	Heterozygous duplication affecting the probes for TMEM18, NBAS and MYCN.
	2p25.3 region	Coriell NA00501 (m)	Heterozygous deletion affecting the TMEM18 probe.
	2p24.3 region	Coriell NA00945 (f)	Heterozygous deletion affecting the probes for NBAS and MYCN.
		Coriell NA09216 (m)	Heterozygous deletion affecting the probes for NBAS and MYCN.
	2q33.1-q37.3 region	Coriell NA01229 (f)	Heterozygous duplication affecting the probes for BMPR2 and ATG4B.
	2q37.3 region	Coriell NA22770 (m)	Heterozygous deletion affecting the ATG4B probe.
	3p21.31-p26.3 region	Coriell NA04127 (f)	Heterozygous duplication affecting the probes for CRBN, PPARG, CTNNB1 and RASSF1.
	3p26.3 region	Coriell NA10985 (f)	Heterozygous deletion affecting the CRBN probe.
	3p26.3 region & 3q24-q27.1 region	Coriell NA11428 (f)	Heterozygous deletion affecting the CRBN probe. Heterozygous duplication affecting the probes for ZIC1, SLITRK3 and MCCC1.
	3q13.33 region	Coriell NA08778 (m)	Heterozygous deletion affecting the CASR probe.
	3q13.33-q27.1 region	Coriell NA03563 (m)	Heterozygous duplication affecting the probes for CASR, ZIC1, SLITRK3 and MCCC1.
	3q27.1 region & 9p24.1-q34.3 region	Coriell NA03563 (m)	Heterozygous duplication affecting the probes for MCCC1, PTPRD, CDKN2A, CDKN2B, IGFBPL1, TRPM3, ALDOB, DEC1 and EHMT1.
	7p11.2-p22.3 region	Coriell NA07081 (m)	Heterozygous duplication affecting the probes for MAFK, GHRHR and EGFR.
	7p15.1 region	Coriell NA08763 (m)	Heterozygous deletion affecting the GHRHR probe.
	7q11.23 region	Coriell NA12590 (m)	Heterozygous deletion affecting the ELN probe.
	7q11.23-q21.2 region	Coriell NA10160 (m)	Heterozygous deletion affecting the probes for ELN and CDK6.
	7q32.1 region	Coriell NA12519 (f)	Heterozygous triplication / homozygous duplication affecting the IMPDH1 probe.
	7q36.3 region	Coriell NA07412 (m)	Heterozygous deletion affecting the SHH probe.
		Coriell NA01220 (f)	Heterozygous duplication affecting the SHH probe.
		Coriell NA10313 (m)	Heterozygous deletion affecting the SHH probe.
	9p21.3-p24.1 region	Coriell NA02819 (f)	Heterozygous duplication affecting the probes for PTPRD, CDKN2A and CDKN2B.
		Coriell NA01750 (m)	Heterozygous duplication affecting the probes for PTPRD, CDKN2A and CDKN2B.
	9p13.1-p24.1 region	Coriell NA03226 (m)	Heterozygous duplication affecting the probes for PTPRD, CDKN2A, CDKN2B, IGFBPL1.
		Coriell NA05067 (m)	Heterozygous duplication affecting the probes for PTPRD, CDKN2A, CDKN2B, IGFBPL1.
	9q34.3 region	Coriell NA13685 (f)	Heterozygous duplication affecting the EHMT1 probe.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P303-A	1p36.32 region	Coriell NA22995 (m)	Heterozygous deletion affecting the TP73 probe.
	1q32.1 region	Coriell NA00214 (m)	Heterozygous deletion affecting the KIF14 probe.
	1q42.12-q44 region	Coriell NA05347 (m)	Heterozygous duplication affecting the probes for LIN9 and SH3BP5L.
	4q13.2 region	Coriell NA00782 (m)	Heterozygous duplication affecting the GNRHR probe.
	5q22.2 region	Coriell NA14234 (m)	Heterozygous deletion affecting the APC probe.
	8p23.1 region	Coriell NA10932 (m)	Heterozygous deletion affecting the PINX1 probe.
	8p23.1-q24.3 region	Coriell NA02030 (m)	Heterozygous duplication affecting the probes for PINX1, GNRH1, FGFR1, CHD7, MYC and PTP4A3.
	8p12-p21.2 region	Coriell NA14485 (m)	Heterozygous duplication affecting the probes for GNRH1 and FGFR1.
	8q24.21 region	Coriell NA03999 (f)	Heterozygous deletion affecting the MYC probe.
	8q24.3 region	Coriell NA20263 (m)	Heterozygous duplication affecting the PTP4A3 probe.
	10p15.1 region	Coriell NA06936 (f)	Heterozygous deletion affecting the KLF6 probe.
	10q23.31-q26.13 region	Coriell NA20125 (m)	Heterozygous duplication affecting the probes for PTEN, SUFU, MXI1 and DMBT1.
	10q24.32-q26.13 region	Coriell NA00959 (m)	Heterozygous duplication affecting the probes for SUFU, MXI1 and DMBT1.
	10q26.13 region	Coriell NA05299 (f)	Heterozygous deletion affecting the DMBT1 probe.
	20p11.21-p12.2 region	Coriell NA00981 (f)	Heterozygous duplication affecting the probes for JAG1 and PYGB.
	20p12.2 region	Coriell NA10608 (m)	Heterozygous deletion affecting the JAG1 probe.
	20q11.23 region	Coriell NA07945 (m)	Heterozygous deletion affecting the NNAT probe.
P305-B	2q37.3 region	Coriell NA01229 (f)	Heterozygous duplication affecting the probes for AGXT.
		Coriell NA14943 (m)	Heterozygous deletion affecting the probes for AGXT.
	9p13.2 region	Coriell NA05067 (m)	Heterozygous deletion affecting the probes for GRHPR.
P308-B	MET	Coriell NA01059 (f)	Heterozygous deletion affecting all probes for MET. Flanking probes on 7q31.2 are also affected.
		Coriell NA12519 (f)	Homozygous duplication affecting all probes for MET. Flanking probes on 7q31.2 are also affected.
	PTEN	Coriell NA20125 (m)	Heterozygous duplication affecting all probes for PTEN.
P315-C	EGFR	Coriell NA07081 (m)	Heterozygous duplication of the complete EGFR gene.
		DSMZ ACC-444 (FU-OV-1) (f)	Loss of EGFR.
P319-B	2p25.3 region & 2q13 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for TPO and PAX8.
	2q13 region	Coriell NA13590 (f)	Heterozygous duplication affecting the probes for PAX8.
	14q13.3 region	Coriell NA06801 (f)	Heterozygous duplication affecting the probes for NKX2-1.
		Coriell NA22765 (m)	Heterozygous deletion affecting the probes for NKX2-1.
	14q31.1 region	Coriell NA16593 (f)	Heterozygous deletion affecting the probes for TSHR Exon 1-8.
P323-B	12p & 12q	DSMZ ACC-200 (COLO-824) (f)	Gain affecting the probes for FOXM1, CCND2 exon 1,CDKN1B, KRAS and KIF21A. Loss affecting the probes for COL2A1, MAP3K12, CDK2, STAT6, GLI1, DDIT3, TSPAN31, CDK4 and CHFR. Some of the reference probes are also affected by CNAs.
	12q	DSMZ ACC-237 (IGR-37) (m)	Loss affecting the probes for KIF21A, COL2A1, MAP3K12, CDK2, STAT6, GLI1, DDIT3, TSPAN31, CDK4, MIR26A2, HMGA2, MDM2, YEATS4, ALX1, IGF1, PTPN11, TBX5, HNF1A, PIWIL1, RAN and CHFR. Some of the reference probes are also affected by CNAs.
		DSMZ ACC-277 (DK-MG) (f)	Amplification affecting the probes for MDM2. Some of the reference probes are also affected by CNAs.
		DSMZ ACC-517 (HCC-1143) (f)	Gain affecting the probes for STAT6, GLI1, DDIT3, TSPAN31, CDK4 and MIR26A2. Amplification affecting the probes for HMGA2, MDM2 and YEATS4. Some of the reference and flanking probes are also affected by CNAs.
		DSMZ ACC-566 (HCC-827) (f)	Amplification affecting the probes for CDK4, TSPAN31 and HMGA2 exon 1-3. Some of the reference probes are also affected by CNAs.
		Coriell NA02819 (f)	Heterozygous deletion affecting the probes for PIWIL1, RAN and CHFR.
		Coriell NA07981 (m)	Mosaic homozygous duplication affecting the probes for PFOXM1, CCND2, CDKN1B and KRAS.
		Coriell NA08035 (m)	Heterozygous duplication affecting the probes for PFOXM1, CCND2, CDKN1B and KRAS.
P324-B	22q11.21 region	Coriell NA05401 (m)	Heterozygous deletion from LCR-A to LCR-B and of the Cat Eye Syndrome region.
		Coriell NA07215 (f)	Heterozygous deletion of the typically deleted region (TDR), from LCR-A to LCR-D.
		Coriell NA10382 (m)	Heterozygous deletion of the typically deleted region (TDR), from LCR-A to LCR-D.
		Coriell NA17942 (m)	Heterozygous deletion of the typically deleted region (TDR), from LCR-A to LCR-D.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P327-B	21q11.2-q21.3 region	Coriell NA00692 (m)	Heterozygous deletion affecting the HSPA13, SAMSN1, MIR99A, BTG3, TMPRSS15, NCAM2, MIR155 and APP probes.
	21q11.2-q22.3 region	Coriell NA02571 (f)	Trisomy 21. All probes targeting chromosome 21 are affected.
	21q11.2-q21.1 region	Coriell NA03503 (m)	Heterozygous duplication affecting the HSPA13, SAMSN1, MIR99A, BTG3, TMPRSS15 and NCAM2 probes.
	21q11.2-q22.11 region & 21q22.3 region	Coriell NA05881 (f)	Heterozygous duplication affecting the HSPA13, SAMSN1, MIR99A, BTG3, TMPRSS15, NCAM2, MIR155, APP, CYYR1, ADAMTS5, BACH1, TIAM1 and PRMT2 probes.
	21q21.3 region	Coriell NA08331 (m)	Heterozygous deletion affecting the APP, CYRR1 and ADAMTS5 probes.
	21q22.13-q22.3 region	Coriell NA09868 (f)	Heterozygous deletion affecting the SIM2, HLCS, DYRK1A, KCNJ6, ERG, ETS2, PSMG1, TMPRSS2, RIPK4, TFF1, ITGB2, SLC19A1, COL6A2 and PRMT2 probes.
	21q22.11 region	Coriell NA13031 (m)	Heterozygous duplication affecting the KCNE2 probe.
P329-B	Xp22.33 / Yp11.32 region	Coriell NA03623 (f)	Heterozygous duplication affecting the probes for SHOX, CRLF2, CSF2RA, IL3RA, P2RY8, ZBED1 and CD99.
		Coriell NA04626 (f)	Heterozygous duplication affecting the probes for SHOX, CRLF2, CSF2RA, IL3RA, P2RY8, ZBED1 and CD99.
		Coriell NA09403 (f)	Heterozygous deletion affecting the probes for SHOX, CRLF2, CSF2RA, IL3RA, P2RY8, ZBED1 and CD99.
		Coriell NA13019 (f)	Heterozygous deletion affecting the probes for SHOX, CRLF2, CSF2RA, IL3RA, P2RY8, ZBED1 and CD99.
		Coriell NA14523 (f)	Heterozygous deletion affecting the probes for SHOX, CRLF2, CSF2RA, IL3RA, P2RY8, ZBED1 and CD99.
		Coriell NA20027 (f)	Heterozygous deletion affecting the probes for SHOX, CRLF2, CSF2RA, IL3RA, P2RY8, ZBED1 and CD99.
P335-C	5q33.3 region & Xp22.33-PAR1 region	Coriell NA04371 (m)	Heterozygous duplication affecting the probes for EBF1 and CSF2RA.
	7p12.2 region	Coriell NA07081 (m)	Heterozygous duplication affecting the probes for IKZF1.
	7p12.2 region & Xp22.33-PAR1 region	Coriell NA10925 (m)	Heterozygous deletion affecting the probes for IKZF1. Heterozygous duplication affecting the probes for CRLF2 and CSF2RA.
	9p13.2-p24.1 region	Coriell NA05067 (m)	Heterozygous duplication affecting the probes for JAK2, CDKN2A, CDKN2B and PAX5.
	9p21.3-p24.1 region	Coriell NA01750 (m)	Heterozygous duplication affecting the probes for JAK2, CDKN2A and CDKN2B.
		Coriell NA12722 (m)	Heterozygous duplication affecting the probes for JAK2, CDKN2A and CDKN2B. Some of the reference probes are affected by CNAs.
	12p13.2 region	Coriell NA07981 (m)	Heterozygous triplication / homozygous duplication affecting the probes for ETV6.
	13q14.2 region	Coriell NA12606 (m)	Heterozygous duplication affecting the probes for RB1.
		Coriell NA14164 (f)	Heterozygous deletion affecting the probes for RB1.
	7p12.2 region & 9p21.3 region & Xp22.33-PAR1 region	DSMZ ACC-20 (BV-173) (f)	Heterozygous deletion affecting the probes for IKZF1 (exons 1-7), CDKN2A (exon 4) and PAR1 region. Homozygous deletion affecting the probes for CDKN2A (exon 2) and CDKN2B (exon 2). some of the reference probes are affected by CNAs.
	Xp22.33-PAR1 region	Coriell NA01353 (m)	Heterozygous deletion affecting the probes for SHOX, CRLF2 and CSF2RA.
		Coriell NA09403 (f)	Heterozygous deletion affecting the probes for SHOX, CRLF2, CSF2RA, IL3RA and P2RY8.
P337-C	16p13 region	Coriell NA04520 (f)	Heterozygous deletion affecting the probes for TSC2 exon 1-15.
		Coriell NA06226 (m)	Homozygous duplication affecting the probes for TSC2 and PKD1.
P338-B	GBA	Coriell NA20273 (m)	Homozygous absence of GBA exon 10.
P348-C	ATP1A2	Coriell NA00803 (m)	Heterozygous deletion affecting all ATP1A2 probes.
	PRRT2	Coriell NA05875 (f)	Heterozygous deletion affecting all PRRT2 probes.
P351-D & P352-E	16p13.3 region	Coriell NA02325 (f)	Heterozygous duplication affecting the probes for PKD1 and TSC2.
P352-E	4q22.1 region	Coriell NA00782 (m)	Heterozygous duplication affecting all PKD2 probes.
		Coriell NA10800 (m)	Heterozygous deletion affecting all PKD2 probes.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P357-A	2p25.1 region	Coriell NA00945 (f)	Heterozygous deletion affecting all KLF11 probes.
		Coriell NA01353 (m)	Heterozygous duplication affecting all KLF11 probes.
		Coriell NA04409 (m)	Heterozygous duplication affecting all KLF11 probes.
	2p25.1-q31.3 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for KLF11 and NEUROD1.
	2q31.3 region	Coriell NA03918 (f)	Heterozygous duplication affecting all NEUROD1 probes.
	7q32.1 region	Coriell NA12519 (f)	Heterozygous duplication affecting all PAX4 probes.
	9q34.2 region	Coriell NA13685 (f)	Heterozygous duplication affecting all CEL probes.
	13q12.2 region	Coriell NA03330 (m)	Heterozygous duplication affecting all PDX1 probes.
		Coriell NA12606 (m)	Heterozygous duplication affecting all PDX1 probes.
	17q12 region	Coriell NA20359 (f)	Heterozygous duplication affecting all HNF1B probes.
P369-A	17p11.2 region	Coriell NA13476 (f)	Heterozygous deletion of the common chromosome 17p11.2 deletion/duplication region.
P370-C	3p25.1-p26.3 region	Coriell NA03503 (m)	Heterozygous duplication affecting the CRBN, SRGAP3 and RAF1 probes.
		Coriell NA04127 (f)	Heterozygous duplication affecting the CRBN, SRGAP3 and RAF1 probes.
	3p25.3-p26.3 region	Coriell NA10985 (f)	Heterozygous deletion affecting the CRBN and SRGAP3 probes.
	6q22.33-q23.3 region	Coriell NA09367 (f)	Heterozygous duplication affecting the LAMA2 and MYB probes.
	6q23.3-q24.2 region	Coriell NA07994 (m)	Heterozygous duplication affecting the MYB and PLAGL1 probes.
	7q34-q35 region	Coriell NA07412 (m)	Heterozygous deletion affecting the KIAA1549, HIPK2, MKRN1, BRAF and CNTNAP2 probes.
		Coriell NA08808 (m)	Heterozygous deletion affecting the KIAA1549, HIPK2, MKRN1, BRAF and CNTNAP2 probes.
		Coriell NA12519 (f)	Heterozygous triplication / Homozygous duplication affecting the KIAA1549, HIPK2, MKRN1, BRAF and CNTNAP2 probes.
		Coriell NA01220 (f)	Heterozygous duplication affecting the MKRN1, BRAF and CNTNAP2 probes.
	8p11.23-p12 region	Coriell NA14485 (m)	Heterozygous duplication affecting the FGFR1 and TACC1 probes.
	8p12-q13.1 region	Coriell NA02030 (m)	Heterozygous duplication affecting the FGFR1, TACC1 and MYBL1 probes.
	9p21.3 region	Coriell NA01750 (m)	Heterozygous duplication affecting the MIR31, CDKN2A and CDKN2B probes.
		Coriell NA02819 (f)	Heterozygous duplication affecting the MIR31, CDKN2A and CDKN2B probes.
		Coriell NA03226 (m)	Heterozygous duplication affecting the MIR31, CDKN2A and CDKN2B probes.
		Coriell NA05067 (m)	Heterozygous duplication affecting the MIR31, CDKN2A and CDKN2B probes.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected	
P377-B	2p23.2-p24.3 region	Coriell NA01353 (m)	Heterozygous duplication affecting the probes for ALK and MYCN.	
	2p24.3 region	Coriell NA00945 (f)	Heterozygous deletion affecting the probes for MYCN.	
		Coriell NA04409 (m)	Heterozygous duplication affecting the probes for MYCN.	
	5q33.3 region	Coriell NA04371 (m)	Heterozygous duplication affecting the probes for EBF1 and MIR146A.	
	6q21 region	Coriell NA01221 (m)	Heterozygous duplication affecting the FYN probe.	
	6q21-q23.3 region	Coriell NA09367 (f)	Heterozygous duplication affecting the probes for FYN and MYB.	
	6q23.3-q27 region	Coriell NA07994 (m)	Heterozygous duplication affecting the probes for MYB, ESR1 and SMOC2.	
	6q27 region	Coriell NA08386 (f)	Heterozygous deletion affecting the SMOC2 probe.	
	7p12.2 region	Coriell NA07081 (m)	Heterozygous duplication affecting the probes for IKZF1.	
		Coriell NA10925 (m)	Heterozygous deletion affecting the probes for IKZF1.	
	7q21.2 region	Coriell NA10160 (m)	Heterozygous deletion affecting the CDK6 probe.	
	7q31.2 region	Coriell NA12519 (f)	Heterozygous triplication/homozygous duplication affecting the MET probe.	
	7q36.2 region	Coriell NA10313 (m)	Heterozygous deletion affecting the DPP6 probe.	
	8q24.21 region	Coriell NA02030 (m)	Heterozygous duplication affecting the probes for MYC.	
		Coriell NA03999 (f)	Heterozygous deletion affecting the probes for MYC.	
	9p13.2-p21.3 region	Coriell NA03226 (m)	Heterozygous duplication affecting the probes for MTAP, CDKN2A, CDKN2B and PAX5.	
		Coriell NA05067 (m)	Heterozygous duplication affecting the probes for MTAP, CDKN2A, CDKN2B and PAX5.	
	9p21.3 region	Coriell NA01750 (m)	Heterozygous duplication affecting the probes for MTAP, CDKN2A and CDKN2B.	
		Coriell NA02819 (f)	Heterozygous duplication affecting the probes for MTAP, CDKN2A and CDKN2B.	
	10q23.31 region	Coriell NA20125 (m)	Heterozygous duplication affecting the probes for PTEN.	
	11q22.3 region	Coriell NA08618 (m)	Heterozygous duplication affecting the probes for ATM.	
		Coriell NA09596 (m)	Heterozygous deletion affecting the probes for ATM.	
	12p13.2-p13.32 region	Coriell NA07981 (m)	Heterozygous triplication/homozygous duplication affecting the probes for CCND2 and ETV6.	
		Coriell NA08035 (m)	Heterozygous duplication affecting the probes for CCND2 and ETV6.	
	13q14.2-q14.3 region	Coriell NA05832 (m)	Heterozygous duplication affecting the probes for RB1, MIR15A, DLEU2 and DLEU1.	
		Coriell NA14164 (f)	Heterozygous deletion affecting the probes for RB1, MIR15A, DLEU2 and DLEU1.	
	18p11.21 region	Coriell NA06870 (f)	Heterozygous triplication/homozygous duplication affecting the RNMT probe.	
		Coriell NA50322 (f)	Heterozygous deletion affecting the RNMT probe.	
	18p11.21-q21.2 region	Coriell NA01359 (m)	Heterozygous duplication affecting the probes for RNMT and DCC.	
		Coriell NA03623 (f)	Heterozygous duplication affecting the probes for RNMT and DCC.	
	18q21.2 region	Coriell NA07891 (m)	Heterozygous deletion affecting the DCC probe.	
	9p21.3 region & 18p11.21-q21.2 region	Coriell NA12722 (m)	Heterozygous duplication affecting the probes for MTAP, CDKN2A, CDKN2B, RNMT and DCC.	
	7p12.2-q36.2 region & 8q24.21 region & 9p24.1 region	Coriell NA23245 (f)	Heterozygous deletion affecting the probes for IKZF1, CDK6, RELN, MET and DPP6. Heterozygous duplication affecting the probes for MYC. Positive for the JAK2 p.V617F mutation.	
	7p12.2-q36.2 region & 9p24.1 region & 18q21.2 region	Horizon Discovery JAK2 p.V617F 50% reference standard	Heterozygous duplication affecting the probes for IKZF1, CDK6, RELN, MET, DPP6 and DCC. Positive for the JAK2 p.V617F mutation.	
	7p12.2-q36.2 region & 18q21.2 region	JAK2 p.V617F 0% (wild-type) reference standard	Heterozygous duplication affecting the probes for IKZF1, CDK6, RELN, MET, DPP6 and DCC.	
	P378-D	MUTYH	Coriell HG01918 (f)	Positive for the MUTYH c.536A>G (p.Tyr179Cys) mutation.
Coriell HG00097 (f), HG01095 (f), HG01500 (m), HG01685 (f), NA19789 (m) and NA20522 (f) are heterozygous positive for the MUTYH p.G396D mutation.				
15q13.3 region		Coriell NA03184 (m)	Heterozygous duplication affecting the probes for SCG5 and GREM1.	

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P380-B	1p36.33 region	Coriell NA22977 (f)	Heterozygous deletion affecting the TNFRSF18 probe.
	1p36.31-p36.33 region	Coriell NA22976 (m)	Heterozygous deletion affecting the probes for TNFRSF18 and CHD5.
	1p36.23-p36.31 region	Coriell NA50276 (m)	Heterozygous deletion affecting the probes for CHD5 and MIR34A.
	1q23.3 region	Coriell NA00803 (m)	Heterozygous deletion affecting the MPZ probe.
	1q43 region	Coriell NA06473 (f)	Heterozygous deletion affecting the SDCCAG8 probe.
	2p24.3-q36.1 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for MYCN, DYSF and PAX3.
	2p24.3 region	Coriell NA00945 (f)	Heterozygous deletion affecting the probes for MYCN.
	4q22.1 region	Coriell NA10800 (m)	Heterozygous deletion affecting the PKD2 probe.
	4q31.3 region	Coriell NA00501 (m)	Heterozygous duplication affecting the probes for FBXW7.
		Coriell NA10313 (m)	Heterozygous duplication affecting the probes for FBXW7.
	11p13 region	Coriell NA09709 (m)	Heterozygous duplication affecting the probes for WT1.
	16p13.11-p13.3 region	Coriell NA06226 (m)	Heterozygous duplication affecting the probes for CREBBP and ABCC6.
		Coriell NA08039 (m)	Heterozygous duplication affecting the probes for CREBBP and ABCC6.
	16p13.11 region	Coriell NA13685 (f)	Heterozygous deletion affecting the ABCC6 probe.
	16p11.2 region	Coriell NA05875 (f)	Heterozygous deletion affecting the VKORC1 probe.
	16q22.1 region	Coriell NA12074 (f)	Heterozygous deletion affecting the CDH1 probe.
	16q23.2-q24.3 region	Coriell NA09687 (m)	Heterozygous duplication affecting the probes for MLYCD and FANCA.
	Xp22.12-q21.2 region	Coriell NA01416 (f)	Heterozygous triplication/homozygous duplication affecting the probes for RPS6KA3, AMER1 and CHM.
P383-A	4q25 region	Coriell NA00501 (m)	Heterozygous duplication affecting the LEF1 probes.
		Coriell NA00782 (m)	Heterozygous duplication affecting the LEF1 probes.
	6q15 region	Coriell NA01221 (m)	Heterozygous duplication affecting the CASP8AP2 probes.
	6q23.3 region	Coriell NA07994 (m)	Heterozygous duplication affecting the probes for MYB and AHI1.
		Coriell NA09367 (f)	Heterozygous duplication affecting the probes for MYB and AHI1.
	7q36.1 region	Coriell NA01220 (f)	Heterozygous duplication affecting the EZH2 probes.
		Coriell NA07412 (m)	Heterozygous deletion affecting the EZH2 probes.
		Coriell NA12519 (f)	Homozygous duplication affecting the EZH2 probes.
	9p21.3 region	Coriell NA02819 (f)	Heterozygous duplication affecting the probes for MLLT3, MTAP, CDKN2A and CDKN2B.
		Coriell NA03226 (m)	Heterozygous duplication affecting the probes for MLLT3, MTAP, CDKN2A and CDKN2B.
		Coriell NA05067 (m)	Heterozygous duplication affecting the probes for MLLT3, MTAP, CDKN2A and CDKN2B.
	9q34.12-q34.13 region	Coriell NA13685 (f)	Heterozygous duplication affecting the probes for ABL1 and NUP214.
	10q23.31 region	Coriell NA20125 (m)	Heterozygous duplication affecting the probes for PTEN.
	11p13-p12 region	Coriell NA09709 (m)	Heterozygous deletion affecting the probes for LMO2, CD44, SLC1A2 and RAG2.
	18p11.21 region	Coriell NA01359 (m)	Heterozygous duplication affecting the PTPN2 probes.
		Coriell NA06870 (f)	Homozygous duplication affecting the PTPN2 probes.
		Coriell NA50136 (f)	Heterozygous deletion affecting the PTPN2 probes.
	18p11.21 & Xq25-q26.3 region	Coriell NA03623 (f)	Heterozygous duplication affecting the PTPN2, SH2D1A, PHF6 and ARHGEF6 probes.
	Xq25-q26.3 region	Coriell NA01416 (f)	Homozygous duplication affecting the SH2D1A, PHF6 and ARHGEF6 probes.
		Coriell NA20027 (f)	Heterozygous deletion affecting the PTPN2, SH2D1A, PHF6 and ARHGEF6 probes.
P385-A & P386-A	9p24.3 region	Coriell NA02819 (f)	Heterozygous duplication affecting the probes for DOCK8 and DMRT1.
		Coriell NA03226 (m)	Heterozygous duplication affecting the probes for DOCK8 and DMRT1.
		Coriell NA05347 (m)	Heterozygous deletion affecting the probes for DOCK8 and DMRT1.
		Coriell NA10989 (m)	Heterozygous deletion affecting the probes for DOCK8 and DMRT1.
P405-A/B	17p12 region	Coriell NA05167 (f)	Heterozygous duplication affecting the COX10, PMP22 and TEKT3 probes.
		Coriell NA12214 (m)	Heterozygous duplication affecting the COX10, PMP22 and TEKT3 probes.
P411-B	PPOX	Coriell NA00803 (m)	Heterozygous deletion affecting all PPOX probes.
	HMBS	Coriell NA15099 (m)	Heterozygous duplication affecting all HMBS probes.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P414-C	3q21.3-q26.2 region	Coriell NA03563 (m)	Heterozygous duplication affecting the GATA2 and MECOM probes.
	5q22.2 region	Coriell NA14234 (m)	Heterozygous deletion affecting the APC probe.
	5q33.3 region	Coriell NA04371 (m)	Heterozygous duplication affecting the MIR146A probe.
	7q21.2-q21.2 region	Coriell NA10160 (m)	Heterozygous deletion affecting the CDK6 and SAMD9L probes.
	7q22.2-q31.2 region	Coriell NA01059 (f)	Heterozygous deletion affecting the KMT2E and MET probes.
	7q36.1 region	Coriell NA01220 (f)	Heterozygous duplication affecting the EZH2 probes.
		Coriell NA07412 (m)	Heterozygous deletion affecting the EZH2 probes.
	8p12-q24.3 region	Coriell NA02030 (m)	Heterozygous duplication affecting the FGFR1, NCOA2, RUNX1T1, MYC and PTK2 probes.
	8p12 region	Coriell NA14485 (m)	Heterozygous duplication affecting the FGFR1 probe.
	8q24.21 region	Coriell NA03999 (f)	Heterozygous deletion affecting the MYC probe.
	8q24.3 region	Coriell NA20263 (m)	Heterozygous deletion affecting the PTK2 probe.
	11q23.3-q24.3	Coriell NA15099 (m)	Heterozygous duplication affecting the KMT2A, TIRAP and ETS1 probes.
	11q24.2-q24.3 region	Coriell NA09102 (m)	Heterozygous deletion affecting the TIRAP and ETS1 probes.
	12p13.1-p13.2 region	Coriell NA07981 (m)	Triplication affecting the ETV6 and CDKN1B probes.
	17q11.2-q12 region	Coriell NA02587 (m)	Heterozygous mosaic deletion affecting the NF1, SUZ12 and AATF probes.
	20q11.21-q13.12 region	Coriell NA07945 (m)	Heterozygous deletion affecting the SRC and HNF4A probes.
	Various	ACC-203 (SK-N-MC) (m)	Heterozygous deletion affecting the probes for MLH1 and TP53. Gain affecting the probes for GATA2, MECOM,FGFR1, NCOA2, RUNX1T1, MYC and PTK2. Some of the reference probes are affected by CNAs.
		ACC-554 (MOLM-13) (m)	Gain affecting the probes for FGFR1, NCOA2, RUNX1T1, MYC, PTK2, SMARCA4 and PRPF31.
		ACC-686 (KASUMI-6) (m)	Gain affecting the probes for MECOM and KMT2A exon 4. Heterozygous deletion affecting the probves for ETV6, CDKN1B and TP53. Some of the reference probes are affected by CNAs.
	IKZF1	ACC-546 (SEM) (f)	Heterozygous deletion affecting the IKZF1 probe.
P417-B	3p region	Coriell NA04127 (f)	Heterozygous duplication affecting the probes for MLH1, RMB5, RASSF1 and ZMYND10.
		DSMZ ACC-203 (SK-N-MC) (f)	Heterozygous deletion affecting all probes for BAP1. The flanking probes are also affected.
		DSMZ ACC-512 (ARH-77) (f)	Heterozygous deletion affecting all probes for BAP1. The flanking probes are also affected.
P419-B	9p21.3 region	DSMZ ACC-47 (DOHH-2) (m)	Homozygous deletion of MTAP, CDKN2A, CDKN2B and DMRTA1.
		DSMZ ACC-264 (COLO-679) (f)	Heterozygous deletion of MLLT3, MIR31, MTAP and CDKN2B and homozygous deletion of CDKN2A, as well as several other aberrations.
		Coriell NA03226 (m)	Heterozygous duplication of the 9p21.3 region and of PTENP1.
		Coriell NA05067 (m)	Heterozygous duplication of the 9p21.3 region and of PTENP1.
	CDK4	DSMZ ACC-573 (SU-DHL-8) (f)	Heterozygous duplication of CDK4.
	MITF	Coriell HG00259 (f) and HG01498 (f) are positive for the MITF c.952G>A (p.E318K) mutation.	
P425-B	1q23.3 region	Coriell NA00803 (m)	Heterozygous deletion affecting the probes for NUF2, RP11s and PBX1.
		Coriell NA06038 (m)	Heterozygous deletion affecting the PBX1 probe.
	9p24.1 region	Coriell NA10989 (m)	Heterozygous deletion affecting the JAK2 probe.
	12p13.31 region	Coriell NA07981 (m)	Heterozygous triplication / Homozygous duplication affecting the probes for CD27, VAMP1, NCAPD2 and CHD4.
	13q14.2-q22.1 region	Coriell NA05832 (m)	Heterozygous duplication affecting the probes for RB1, DLEU2 and DIS3.
		Coriell NA14164 (f)	Heterozygous deletion affecting the probes for RB1, DLEU2 and DIS3.
	15q12-q26.3 region	Coriell NA03184 (m)	Heterozygous duplication affecting the probes for GABRB3 and IGF1R.
	15q12 region	Coriell NA20375 (m)	Heterozygous deletion affecting the GABRB3 probe.
	15q26.3 region	Coriell NA03255 (m)	Heterozygous deletion affecting the IGF1R probe.
	16q23.1 region	Coriell NA09687 (m)	Heterozygous duplication affecting the WWOX probe.
P426-A	2p21 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for PPM1B, SLC3A1, PREPL and CAMKMT.
		Coriell NA13451 (m))	Heterozygous deletion affecting the probes for PPM1B, SLC3A1, PREPL and CAMKMT.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P429-C	2q11.2 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for STARD7, TMEM127 and ITPRIPL1.
	5p15.33 region	Coriell NA14131 (f)	Heterozygous deletion affecting the probes for CCDC127, SDHA, PDCD6 and TERT.
		Coriell NA14523 (f)	Heterozygous duplication affecting the probes for CCDC127, SDHA, PDCD6 and TERT.
	14q23.1-q24.3 region	Coriell NA05966 (m)	Heterozygous duplication affecting the probes for COTX2, MAX, RDH12 and NPC2.
P433-A	ARID1B	Coriell NA06802 (m)	Heterozygous deletion affecting the probes for ARID1B.
		Coriell NA07994 (m)	Heterozygous duplication affecting the probes for ARID1B.
P437-B	3q26.2	Coriell NA10175 (m)	Heterozygous duplication affecting the probes for TERC.
		Coriell NA11428 (f)	Heterozygous duplication affecting the probes for TERC.
		Coriell NA20022 (m)	Heterozygous duplication affecting the probes for TERC.
	5p15.33	Coriell NA14523 (f)	Heterozygous duplication affecting the probes for TERT.
		Coriell NA14131 (f)	Heterozygous deletion affecting the probes for TERT.
	21q22.12	Coriell NA01201 (f)	Heterozygous deletion affecting the probes for RUNX1.
	3q21.3 region 3q26.2 region 5p15.33 region 19q13.1 region	DSMZ MFE-280 (f)	Homozygous duplication affecting the probes for GATA2. Heterozygous duplication affecting the probes for TERT. Amplification affecting the probes for TERC and CEBPA. Some of the reference probes are affected by CNAs.
	3q21.3-3q26.2 region	DSMZ SK-N-MC (f)	Heterozygous duplication affecting the probes for GATA2 and TERC.
P440-A	4q35.2 region	Coriell NA00501 (m)	Heterozygous duplication affecting the probes for F11.
		Coriell NA03013 (f)	Heterozygous deletion affecting the probes for F11.
		Coriell NA10313 (m)	Heterozygous deletion affecting the probes for F11.
	13q34 region	Coriell NA03089 (f)	Heterozygous duplication affecting the probes for F10.
		Coriell NA03330 (f)	Heterozygous duplication affecting the probes for F10.
		Coriell NA03887 (f)	Heterozygous deletion affecting the probes for F10.
		Coriell NA05832 (f)	Heterozygous duplication affecting the probes for F10.
		Coriell NA06312 (m)	Heterozygous deletion affecting the probes for F10.
		Coriell NA08254 (m)	Heterozygous deletion affecting the probes for F10.
P445-A	Xp11.3 region	Coriell NA10636 (f)	Heterozygous duplication affecting the probes for KDM6A.
P446-A	GALC	Coriell NA04372 (m)	Heterozygous GALC exon 11-17 deletion (30 kb).
		Coriell NA04517 (m)	Homozygous GALC exon 11-17 deletion (30 kb).
P451-B	16p13.3-p12.1	Coriell NA06226 (m)	Gain of 16p13.3-p12.1, affecting probes for TSC2, CREBBP, ABAT, ABCC1 and UQCRC2.
		Coriell NA08039 (m)	Gain of 16p13.3-p12.1, affecting probes for CREBBP, ABAT, ABCC1, UQCRC2 and PALB2.
	16p11.2	Coriell NA05875 (f)	Deletion of 16p11.2, affecting the probe for VKORC1.
	16q22.1-q23.1	Coriell NA12074 (m)	Deletion of 16q22.1-q23.1, affecting probes for CDH1, TXNL4B, DHX38, ZFHX3 and BCAR1.
P453-A	GAA	Coriell NA11661 (m)	Heterozygous GAA exon 18 deletion.
		Coriell NA16445 (m)	Large heterozygous duplication (6.5 Mb) that includes the complete GAA gene.
P456-A	EVC/EVC2	Coriell NA22601 (m)	Heterozygous deletion of the complete EVC and EVC2 genes.
P460-A	SMN1/SMN2	Coriell NA00232 (m)	SMN1: 0 copies; SMN2: 2 copies; g.27134T>G: 0 copies; g.27706-27707delAT: 0 copies.
		Coriell NA03815 (m)	SMN1: 1 copy; SMN2: 1 copy; g.27134T>G: 0 copies; g.27706-27707delAT: 0 copies.
		Coriell HG01773 (f)	SMN1: 1 copy; SMN2: 4 copies; g.27134T>G: 0 copies; g.27706-27707delAT: 0 copies.
		Coriell NA19984 (m)	SMN1: 2 copies; SMN2: 1 copy; g.27134T>G: 1 copy; g.27706-27707delAT: 1 copy.
		Coriell NA20294 (f)	SMN1: 3 copies; SMN2: 1 copy; g.27134T>G: 1 copy; g.27706-27707delAT: 1 copy.
		Coriell HG02882 (f)	SMN1: 3 copies; SMN2: 1 copy; g.27134T>G: 2 copies; g.27706-27707delAT: 2 copies.
		Coriell NA19235 (f)	SMN1: 4 copies; SMN2: 0 copies; g.27134T>G: 3 copies; g.27706-27707delAT: 3 copies.
P461-B	15q15.3 region	Coriell NA03184 (m)	Heterozygous duplication affecting the probes for PPIP5K1, CKMT1B, STRC, CATSPER2, CKMT1A, STRCP1 and PDIA3.
		Coriell NA20317 (f)	Heterozygous duplication affecting the probes for STRC and CKMT1A. Heterozygous deletion affecting the probes for STRCP1 and CKMT1B.
		Coriell NA20511 (m)	Heterozygous deletion affecting the probes for CKMT1B, STRC and CATSPER2.
	16q12.2 region	Coriell NA08039 (m)	Heterozygous duplication affecting the probes for METTL9, OTOA and UQCRC2.
		Coriell NA13031 (m)	Heterozygous deletion affecting the probes for METTL9 and OTOA.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P466-A	1p22.1-q31.3	Coriell NA00214 (m)	Heterozygous deletion affecting the probes for TROVE2, GLRX2, CDC73, B3GALT2, LINC01031 and KCNT2.
	1q31.2	DSMZ ACC-9 (U-266) (m)	Heterozygous deletion affecting the probes for TROVE2, GLRX2 and CDC73 exon 1-2.
P470-A	CLN3	Coriell NA05875 (f)	Large heterozygous deletion (4.8 Mb) that includes the complete CLN3 gene.
		Coriell NA20381 (f)	Heterozygous CLN3 exon 7-8 deletion.
	CLN6	Coriell NA03184 (m)	Large heterozygous duplication (82 Mb) that includes the complete CLN6 gene.
P472-A	10q24.32 region	Coriell NA00959 (m)	Heterozygous duplication affecting the probes for ACTR1A, SUFU and TRIM8.
		Coriell NA08386 (f)	Heterozygous duplication affecting the probes for ACTR1A, SUFU and TRIM8.
		Coriell NA20125 (m)	Heterozygous duplication affecting the probes for ACTR1A, SUFU and TRIM8.
		DSMZ ACC-203 (f)	Heterozygous deletion affecting the probes for ACTR1A, SUFU and TRIM8.
		DSMZ ACC-259 (m)	Heterozygous deletion affecting the probes for ACTR1A, SUFU and TRIM8.
		DSMZ ACC-569 (m)	Heterozygous deletion affecting the probes for ACTR1A, SUFU and TRIM8.
P473-A	17p/CTNS/ASPA	Coriell NA06047 (m)	Large heterozygous deletion (5.6 Mb) that includes the complete CTNS and SHPK genes.
P474-A	9p24 region	Coriell NA02819 (f)	Heterozygous duplication affecting the CD274, PDCD1LG2 and JAK2 genes and all flanking probes.
		Coriell NA03226 (m)	Heterozygous duplication affecting the CD274, PDCD1LG2 and JAK2 genes and all flanking probes.
		Coriell NA05067 (m)	Heterozygous duplication affecting the CD274, PDCD1LG2 and JAK2 genes and all flanking probes.
		Coriell NA10989 (m)	Heterozygous deletion affecting the CD274, PDCD1LG2 and JAK2 genes and the VLDLR and SMARCA2 flanking probes.
		Coriell NA14946 (m)	Heterozygous deletion affecting the CD274, PDCD1LG2 and JAK2 genes and the VLDLR and SMARCA2 flanking probes.
	JAK2	Coriell NA13480 (f)	Heterozygous JAK2 exon 23-24 duplication.
P475-A	7q31.1 region	Coriell NA01059 (f)	Heterozygous deletion affecting the probes for FOXP2.
		Coriell NA12519 (f)	Homozygous duplication affecting the probes for FOXP2.
P476-A	ZNRF3	Coriell NA02325 (f)	Heterozygous duplication of the ZNRF3 gene. The flanking probes are also affected.
		Coriell NA07106 (m)	Heterozygous duplication of the ZNRF3 gene. The flanking probes are also affected. This cell line has a partial trisomy of chromosome 22.
P478-A	SMARCE1	DSMZ ACC-410 (MFE-280)	Gain of SMARCE1 and all flanking probes.
P479-A	TCF12	Coriell NA03184 (m)	Heterozygous duplication affecting all TCF12 probes.
P480-A	4p16.3 region	Coriell NA00343 (m)	Heterozygous deletion of the 4p16.3 region, affecting all target probes.
		Coriell NA04126 (m)	Heterozygous deletion of the 4p16.3 region, affecting all target probes.
		Coriell NA22601 (m)	Heterozygous deletion of the 4p16.3 region, affecting all target probes.
P481-A	16p11.2 region	Coriell NA05875 (f)	Heterozygous deletion affecting the probes for ZNF843, ARMC5 and TGFB1I1.
	17p11.2, 17q23.2-q24.2 region	DSMZ ACC-49 (L-363) (f)	Heterozygous duplication affecting the probes for FLCN (17p arm), BRIP1, AXIN2, SLC16A6, ARSG, WIPI1, PRKAR1A and FAM20A, and one reference probe at 250 nt.
P482-A	DICER1	Coriell NA10074 (m)	Heterozygous duplication affecting all DICER1 probes. Flanking probes are also affected.
		Coriell NA13410 (m)	Heterozygous duplication affecting all DICER1 probes. Flanking probes are also affected, except the RPGRIP1 probe.
		DSMZ ACC-49 (L-363) (f)	Deletion affecting all DICER1 probes. Flanking probes are also affected, except the RPGRIP1 probe. Some of the reference probes are affected by CNAs.

Probemix(es)	Gene(s)	Sample ID & sex (m/f)	Copy number changes detected
P483-A	2p22.3-q34 region	Coriell NA10401 (f)	Heterozygous duplication affecting the probes for SPAST, CPS1, ERBB4 and IKZF2.
	2q34 region	Coriell NA01229 (f)	Heterozygous duplication affecting the probes for CPS1, ERBB4 and IKZF2.
		Coriell NA10918 (f)	Heterozygous deletion affecting the probes for ERBB4 and IKZF2.
	7p11.2 region	Coriell NA07081 (m)	Heterozygous duplication affecting the probes for VSTM2A, EGFR and LANCL2.
	2p22.3 region 7p11.2 region 7q22.3 region 12q13.2 region 17p11.2-q12 region 17q12 region	DSMZ ACC-410 (MFE-280) (f)	Heterozygous duplication affecting the probes for SPAST (2p22.3), VSTM2A, EGFR and LANCL2 (7p11.2), and PGAP3, ERBB2 and MIEN1 (17q12). Heterozygous deletion affecting the SLC26A4 probe (7q22.3). Amplification affecting the probes for RPS26, ERBB3 and PA2G4 (12q13.2), and FLCN and WSB1 (17p11.2-q12). Some of the reference probes are affected by CNAs.
	17q11.1-12 region	DSMZ ACC-589 (JIMT-1) (f)	Homozygous duplication affecting the probes for WSB1. Amplification affecting the probes for PGAP3, ERBB2 and MIEN1. Some of the reference probes are affected by CNAs.
P490-B	ADA2	Coriell NA02944 (m)	Heterozygous deletion affecting all ADA2 probes.
		Coriell NA16362 (m)	Heterozygous duplication affecting all ADA2 probes.
P492-A	12q24.33 region	Coriell NA01535 (f)	Heterozygous deletion affecting all POLE probes.
		Coriell NA02819 (f)	Heterozygous deletion affecting all POLE probes.
		Coriell NA07891 (m)	Heterozygous deletion affecting all POLE probes.
P494-A	8q21.3 region	Coriell NA02030 (m)	Heterozygous duplication affecting all NBN probes.
		Coriell NA03134 (m)	Heterozygous deletion affecting all NBN probes.
P496-A	NIPBL	Coriell NA14523 (f)	Heterozygous duplication affecting the NIPBL probe.
	NPM1	Coriell NA04371 (m)	Heterozygous duplication affecting the probes for NPM1.
	IKZF1	Coriell NA07081 (m)	Heterozygous duplication affecting the probes for IKZF1.
	7q22 region	Coriell NA01059 (f)	Heterozygous deletion affecting the probes for CUX1 and KMT2E.
	EZH2	Coriell NA07412 (m)	Heterozygous deletion affecting the probes for EZH2.
		Coriell NA12519 (f)	Homozygous duplication affecting the probes for EZH2.
	NUP98	Coriell NA03435 (m)	Heterozygous duplication affecting the NUP98 probe.
	SLC6A5	Coriell NA05518 (f)	Heterozygous deletion affecting the SLC6A5 probe.
	ATM	Coriell NA09596 (m)	Heterozygous deletion affecting the probes for ATM.
	Various	DSMZ ACC-220 (KASUMI-1) (m)	Heterozygous deletion affecting the probes for TP53. Positive for the ASXL1 c.1934dupG mutation. Some of the reference probes are affected by CNAs.
		DSMZ ACC-686 (KASUMI-6) (m)	Gain affecting the probes for KMT2A exon 2-7. Heterozygous deletion affecting the probes for TP53. Some of the reference probes are affected by CNAs.
		DSMZ ACC-386 (EOL-1) (m)	Gain affecting the probes for KMT2A exon 2-7. Some of the reference probes are affected by CNAs.
		DSMZ ACC-775 (UOC-M1) (m)	Heterozygous deletion affecting the probes for CTNNA1, IKZF1, CUX1, KMT2E, EZH2, NUP98, SLC6A5 and TP53. Gain affecting the probes for ATM, UBE4A, KMT2A, TMEM25 and ETS1. Some of the reference probes are affected by CNAs.
		DSMZ ACC-15 (ML-2) (m)	Heterozygous deletion affecting the probes for SLC6A5, KMT2A exon 9-36, TMEM25 and ETS1. Some of the reference probes are affected by CNAs.
		DSMZ ACC-542 (NOMO-1) (f)	Gain affecting the probes for NIPBL and CUX1. Heterozygous deletion affecting the probes for KMT2E, EZH2 and KMT2A exon 9. Some of the reference probes are affected by CNAs.
P520-A	JAK2	Horizon Discovery provides a JAK2 p.V617F reference standard (catalog number HD649; HD652 is a wild type reference standard).	
		The NIBSC Institute provides a JAK2 p.V617F WHO reference panel (catalog number 16/120).	
	KIT	AccuRef Quan-Plex™ NGS Reference Standard Genomic DNA (catalog number ARF-1001G-1) can be used as reference for the KIT p.D816V mutation; Onco-Ref™ Genomic DNA Reference Standard HCT116 WT (catalog number ASO-6052-1) can be used as wild-type control.	

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