

Please consult the Coffalyser digitalMLPA User Manual for details on information displayed in this report.
For additional support contact us at info@mrcholland.com or visit www.mrcholland.com.

QC passed

BP01-59 - Coriell HG02188

sample		identification		quality summary	
sample name	BP01-59 - Coriell HG02188	SNP id code	121012221011102011010012210211122011000v1	QC checks	Passed
sample type	Test sample	probemix lot	Identity detected	program	Ok
barcode	BP01-59	barcode lot	03-012-yymmdd	run	Ok
sex	Female	sample tracker		identity	Ok
experiment				DNA	Ok
experiment name	D028-A1 Demo set			hybridisation	Ok
product name	NXtec D028-A1-1024 [default sheet]			ligation	Ok
analysed panels	Spinal Muscular Atrophy (SMA), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Beta-thalassemia, DFNB1 hearing loss, Alpha-thalassemia, Juvenile Neuronal Ceroid Lipofuscinosis (CLN3-related), Cystinosis, Duchenne & Becker Muscular Dystrophy (F), Reference probes, Control Probes			polymerase	Ok
masked panels				data analysis	Ok

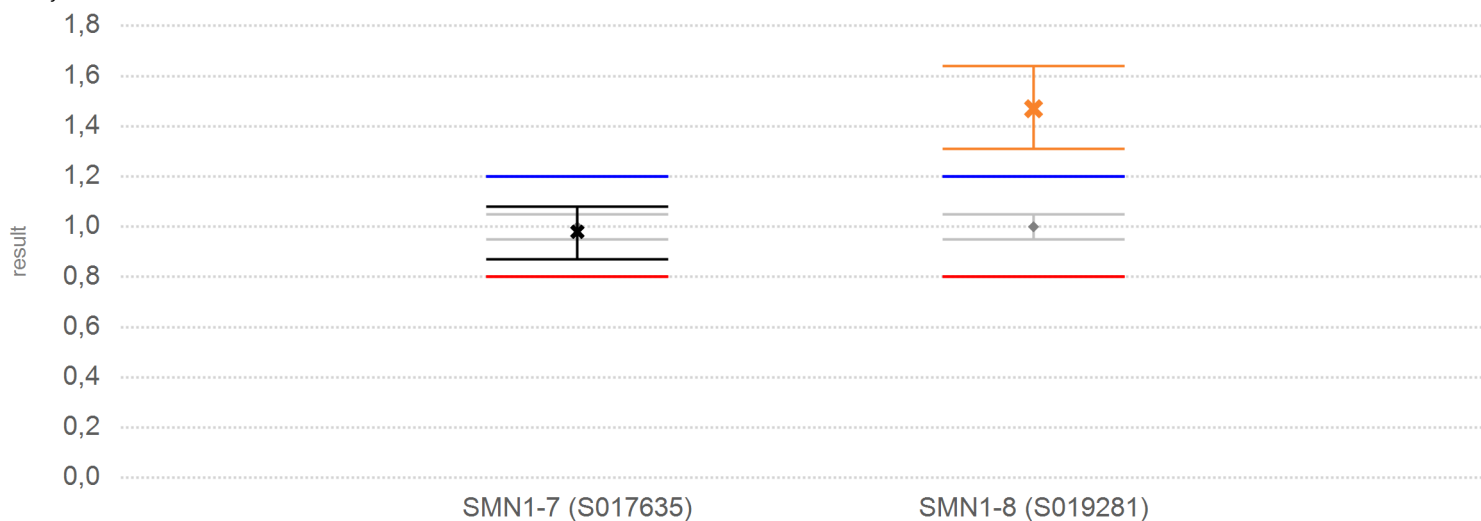
Note: The results displayed in this report are based on pre-set product-specific visualisation settings and masking (if applicable). Not all probe results may be displayed. For an overview of all results, please consult the Excel Report.

Spinal Muscular Atrophy (SMA) [1 aberrant subpanel]

The SMN1 exon numbering is the traditional exon numbering (exons 1, 2a, 2b, and 3-8) used in most scientific literature. This exon numbering is different from the MANE select transcript (NM_000344.4).

SMN1 [1 aberrant segment]

The exon 7 difference between the SMN1 and SMN2 genes, as targeted by S017635, is the only clinically relevant difference between these two genes. The great majority of SMA carriers can be identified by the presence of a single copy of SMN1 exon 7. In 5-10% of all cases, the SMN1 exon 8 probe (S019281) will show a different copy number compared to the SMN1 exon 7 probe (S017635). In these cases, the copy number of SMN1 is only determined by the exon 7 probe. The nucleotide difference targeted by the exon 8 probe is not clinically relevant.



Segment 1 of 1 [1 aberrant probe]

probe information						comparison reference samples				results		
number	function	location (kb)	chr.band	gene	exon	type	CN exp.	range exp.	range 95%	warning / error	aberrant	result
S017635	CN	05:070,952	05q13.2	SMN1	7	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	0,98
S019281	CN	05:070,953	05q13.2	SMN1	8	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	1,47

SMN1 - SNP

The SNP probe S016793 targets the c.*3+80T>G SNP (ClinVar ID 1331398, rs143838139). Presence of this polymorphism in combination with two copies of SMN1 indicates an increased risk, depending on the ethnic background, that the person tested is a silent SMA carrier who carries two SMN1 copies on one allele and none on the other (PMID: 23788250).

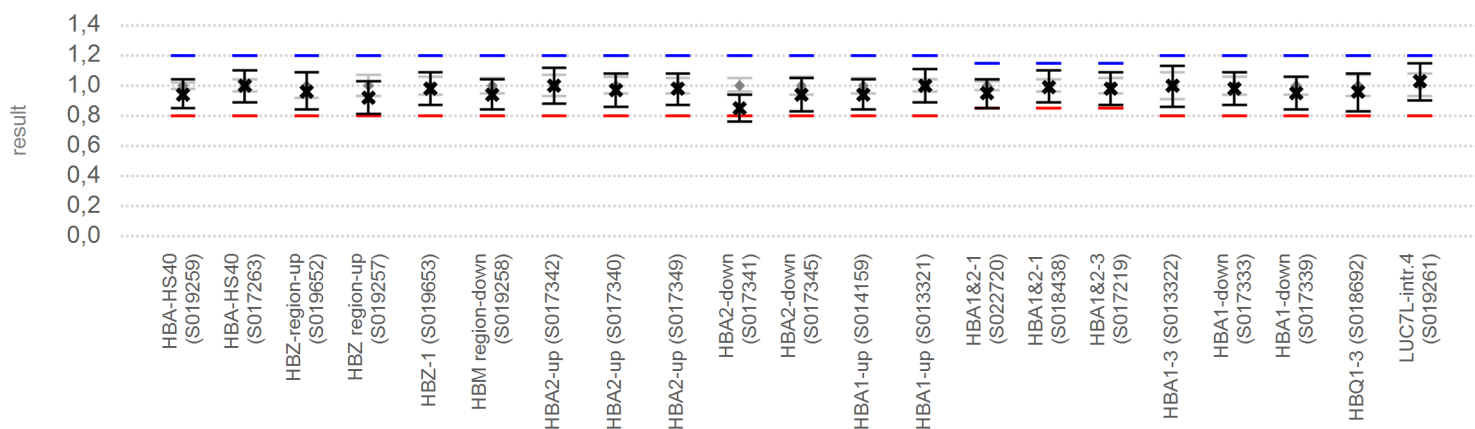
Segment 1 of 1

probe information						comparison reference samples				results		
number	function	location (kb)	chr.band	gene	exon	type	CN exp.	range exp.	range 95%	warning / error	aberrant	result
S016793	SNP	05:070,952	05q13.2	SMN1 c.*3+80T>G	7	Reference	0			-		absent

Alpha-thalassemia [1 aberrant subpanel]

Due to the very high homology and close proximity of the HBA1 and HBA2 genes, sequence exchange (gene conversion) frequently occurs. In some healthy individuals the HBA2 sequence is therefore changed into the HBA1 sequence or vice versa, without any clinical consequences. Two probe pairs target locations with very small sequence differences between HBA1 and HBA2: S017342 & S017341 and S017340 & S017345. When one of the probes in such a probe pair has a ~50% reduced probe signal (ratio 0.5), while the other has a ~50% increased probe signal (ratio 1.5), it is possible that this is a benign polymorphism due to a sequence exchange between HBA1 and HBA2 rather than a true deletion and duplication.

HBA region



Segment 1 of 1

probe information						comparison reference samples				results		
number	function	location (kb)	chr.band	gene	exon	type	CN exp.	range exp.	range 95%	warning / error	aberrant	result
S019259	CN	16:000,114	16p13.3	HBA-HS40	-	Reference	2	0,80 - 1,20	0,98 - 1,02	-	No	0,94
S017263	CN	16:000,114	16p13.3	HBA-HS40	-	Reference	2	0,80 - 1,20	0,96 - 1,04	-	No	1,00
S019652	CN	16:000,144	16p13.3	HBZ region	Up	Reference	2	0,80 - 1,20	0,92 - 1,09	-	No	0,96
S019257	CN	16:000,149	16p13.3	HBZ region	Up	Reference	2	0,80 - 1,20	0,93 - 1,07	-	No	0,92
S019653	CN	16:000,153	16p13.3	HBZ	1	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	0,98
S019258	CN	16:000,167	16p13.3	HBM region	Down	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	0,94
S017342	CN	16:000,170	16p13.3	HBA2	Up	Reference	2	0,80 - 1,20	0,93 - 1,07	-	No	1,00
S017340	CN	16:000,170	16p13.3	HBA2	Up	Reference	2	0,80 - 1,20	0,95 - 1,06	-	No	0,97
S017349	CN	16:000,172	16p13.3	HBA2	Up	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	0,98
S017341	CN	16:000,174	16p13.3	HBA2	Down	Reference	2	0,80 - 1,20	0,96 - 1,05	-	No	0,85
S017345	CN	16:000,175	16p13.3	HBA2	Down	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	0,94
S014159	CN	16:000,175	16p13.3	HBA1	Up	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	0,94
S013321	CN	16:000,176	16p13.3	HBA1	Up	Reference	2	0,80 - 1,20	0,96 - 1,04	-	No	1,00
S022720	CN	16:000,177	16p13.3	HBA1&2	1	Reference	4	0,85 - 1,15	0,97 - 1,03	-	No	0,95
S018438	CN	16:000,177	16p13.3	HBA1&2	1	Reference	4	0,85 - 1,15	0,96 - 1,04	-	No	0,99
S017219	CN	16:000,177	16p13.3	HBA1&2	3	Reference	4	0,85 - 1,15	0,95 - 1,05	-	No	0,98
S013322	CN	16:000,178	16p13.3	HBA1	3	Reference	2	0,80 - 1,20	0,91 - 1,09	-	No	1,00
S017333	CN	16:000,178	16p13.3	HBA1	Down	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	0,98
S017339	CN	16:000,180	16p13.3	HBA1	Down	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	0,95
S018692	CN	16:000,181	16p13.3	HBQ1	3	Reference	2	0,80 - 1,20	0,93 - 1,07	-	No	0,96
S019261	CN	16:000,206	16p13.3	LUC7L	Intr. 4	Reference	2	0,80 - 1,20	0,93 - 1,08	-	No	1,03

HBA region - MUT [1 aberrant segment]

ClinVar ID 15624, rs41464951, *143Q, Hb Constant Spring. Note that this probe cannot distinguish HBA1 from HBA2. Thus, presence of probe signal may also be caused by a sequence change in the HBA1 gene.

Segment 1 of 1 [1 aberrant probe]

probe information						comparison reference samples				results		
number	function	location (kb)	chr.band	gene	exon	type	CN exp.	range exp.	range 95%	warning / error	aberrant	result
S022719	MUT	16:000,174	16p13.3	HBA2 c.427T>C	3	Reference	0			-	Yes	present

analysis performed		final approval	
date		date	
signature		signature	

other		2.6.1	software versions	executed
sample id	f4faf532-1313-4768-a8c2-6dd5205099ca	configuration	2026-05-29 15:30:00	
barcode collection	From lot 03-009-yyymmdd onwards (Default)	data conversion	2026-08-07 20:15:36	2026-09-11 05:57:44
normalisation options	individual probe normalisation	fragment analysis	2026-08-07 20:15:36	2026-09-11 05:57:46
file name	BP01-59 - Coriell HG02188.Coffa	comparative analysis	2026-08-07 20:15:36	2026-09-11 05:57:46
source file(s)	C:\Temp\D028 Demo set\FASTQ digitalMLPA Demo Set D028-A1.fastq	result analysis	2026-08-07 20:15:36	2026-09-11 05:57:46
		reporting	2026-08-07 20:15:36	2026-09-11 05:57:47