

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

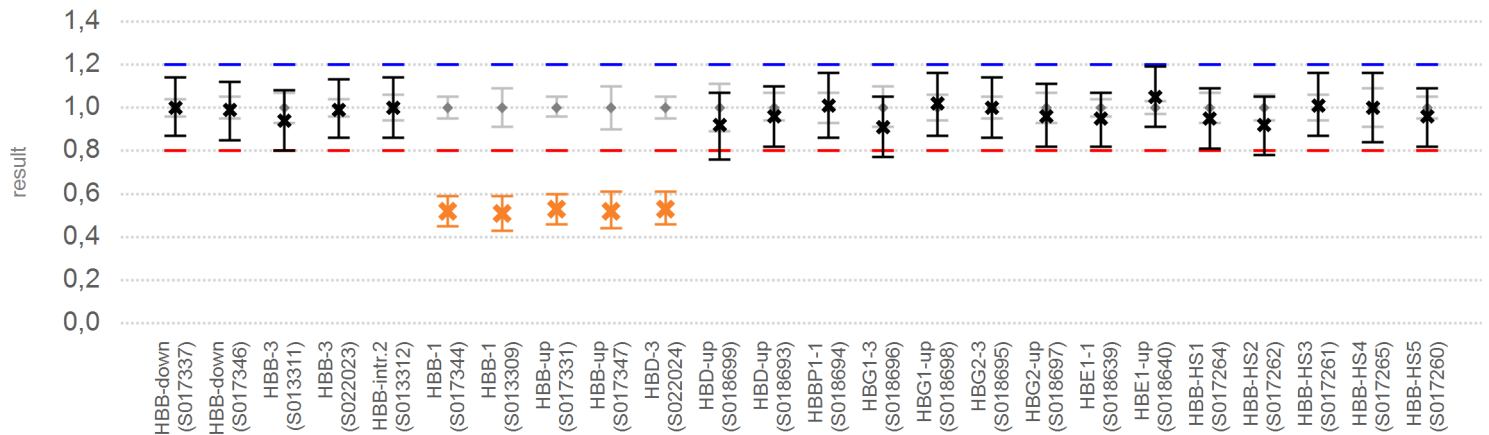
BP01-55 - Coriell NA20480

sample		identification		quality summary	
sample name	BP01-55 - Coriell NA20480	SNP id code	220100112022210110220122111212112102212v1	QC checks	Passed
sample type	Test sample	probemix lot	Identity detected	program	Ok
barcode	BP01-55	barcode lot	03-012-yymmdd	run	Ok
sex	Male	sample tracker		identity	Ok
experiment					
experiment name	D028-A1 Demo set				
product name	NXtec D028-A1-1024 [default sheet]				
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 (M), Reference probes, Control Probes				
masked panels					
				DNA	Ok
				hybridisation	Ok
				ligation	Ok
				polymerase	Ok
				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.

Beta-thalassemia [1 aberrant subpanel]

HBB region [1 aberrant segment]



Segment 1 of 1 [5 aberrant probes]

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
S017337	CN	11:005,225	11p15.4	HBB	Down	Reference	2	0,80 - 1,20	0,96 - 1,04	-	No	1,00
S017346	CN	11:005,225	11p15.4	HBB	Down	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	0,99
S013311	CN	11:005,226	11p15.4	HBB	3	Reference	2	0,80 - 1,20	0,93 - 1,07	-	No	0,94
S022023	CN/WT	11:005,226	11p15.4	HBB WT c.316-3C	3	Reference	2	0,80 - 1,20	0,96 - 1,04	-	No	0,99
S013312	CN	11:005,226	11p15.4	HBB	Intr. 2	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	1,00
S017344	CN	11:005,227	11p15.4	HBB	1	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,52
S013309	CN	11:005,227	11p15.4	HBB	1	Reference	2	0,80 - 1,20	0,91 - 1,09	-	YES *	0,51
S017331	CN	11:005,227	11p15.4	HBB	Up	Reference	2	0,80 - 1,20	0,96 - 1,05	-	YES *	0,53
S017347	CN	11:005,228	11p15.4	HBB	Up	Reference	2	0,80 - 1,20	0,90 - 1,10	-	YES *	0,52
S022024	CN	11:005,233	11p15.4	HBD	3	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,53
S018699	CN	11:005,235	11p15.4	HBD	Up	Reference	2	0,80 - 1,20	0,89 - 1,11	-	No	0,92
S018693	CN	11:005,238	11p15.4	HBD	Up	Reference	2	0,80 - 1,20	0,94 - 1,07	-	No	0,96
S018694	CN	11:005,243	11p15.4	HBBP1	1	Reference	2	0,80 - 1,20	0,93 - 1,07	-	No	1,01
S018696	CN	11:005,248	11p15.4	HBG1	3	Reference	2	0,80 - 1,20	0,91 - 1,10	-	No	0,91
S018698	CN	11:005,251	11p15.4	HBG1	Up	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	1,02
S018695	CN	11:005,253	11p15.4	HBG2	3	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	1,00
S018697	CN	11:005,256	11p15.4	HBG2	Up	Reference	2	0,80 - 1,20	0,93 - 1,07	-	No	0,96
S018639	CN	11:005,270	11p15.4	HBE1	1	Reference	2	0,80 - 1,20	0,96 - 1,04	-	No	0,95
S018640	CN	11:005,270	11p15.4	HBE1	Up	Reference	2	0,80 - 1,20	0,97 - 1,03	-	No	1,05
S017264	CN	11:005,276	11p15.4	HBB-HS1	-	Reference	2	0,80 - 1,20	0,93 - 1,07	-	No	0,95
S017262	CN	11:005,281	11p15.4	HBB-HS2	-	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	0,92
S017261	CN	11:005,285	11p15.4	HBB-HS3	-	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	1,01
S017265	CN	11:005,288	11p15.4	HBB-HS4	-	Reference	2	0,80 - 1,20	0,91 - 1,09	-	No	1,00
S017260	CN	11:005,291	11p15.4	HBB-HS5	-	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	0,96

HBB region - MUT

Three (intronic) mutation-specific probes are included, covering the following mutations: S018382 targets c.316-106C>G (ClinVar ID 15457, rs34690599, IVS II-745 C>G). S018564 targets c.316-197C>T (ClinVar ID 15458, rs34451549, IVS II-654 C>T). S018554 targets c.93-21G>A (ClinVar ID 15454, rs35004220, IVS I-110 G>A).

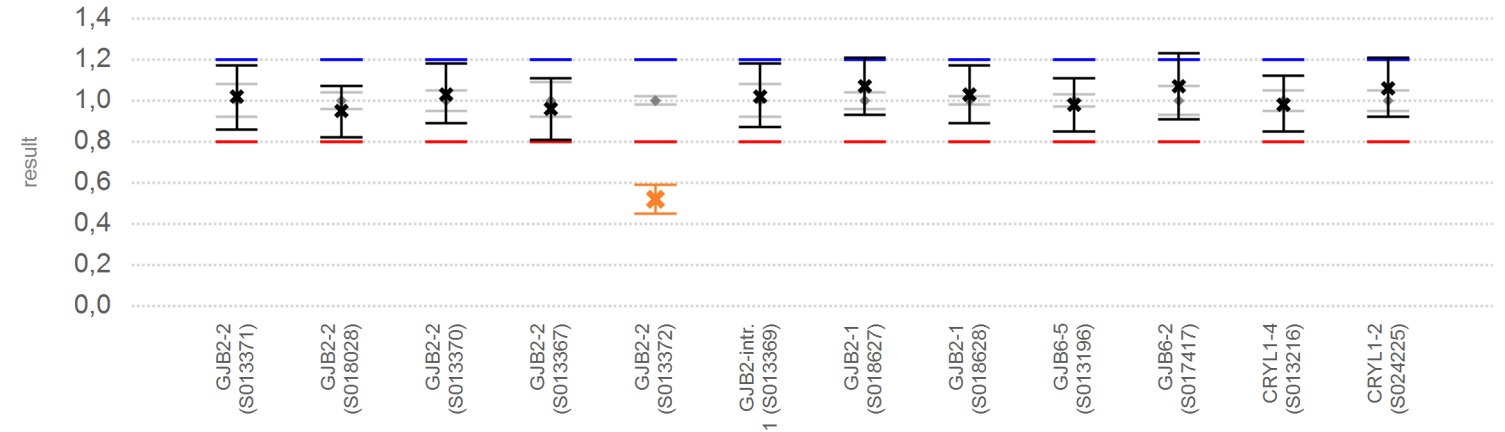
Segment 1 of 1

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
S018382	MUT	11:005,226	11p15.4	HBB c.316-106C>G	Intr. 2	Reference	0			-	No	absent
S018564	MUT	11:005,226	11p15.4	HBB c.316-197C>T	Intr. 2	Reference	0			-	No	absent
S018554	MUT	11:005,227	11p15.4	HBB c.93-21G>A	2	Reference	0			-	No	absent

DFNB1 hearing loss [1 aberrant subpanel]

GJB2 & GJB6 & CRYL1 [1 aberrant segment]

There are two common GJB6 deletion regions: 232 kb (ClinVar ID 5548) and 309 kb (ClinVar ID 5546). Probes S017417 and S013216 are expected to be affected in case of the 232 kb deletion. Probes S017417, S013216 and S024225 are expected to be affected in case of the 309 kb deletion.



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
S013371	CN/WT	13:020,189	13q12.11	GJB2 WT c.313_326del	2	Reference	2	0,80 - 1,20	0,92 - 1,08	-	No	1,02
S018028	CN/WT	13:020,189	13q12.11	GJB2 WT c.235delC	2	Reference	2	0,80 - 1,20	0,96 - 1,04	-	No	0,95
S013370	CN/WT	13:020,189	13q12.11	GJB2 WT c.167delT	2	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	1,03
S013367	CN/WT	13:020,189	13q12.11	GJB2 WT c.101T	2	Reference	2	0,80 - 1,20	0,92 - 1,09	-	No	0,96
S013372	CN/WT	13:020,190	13q12.11	GJB2 WT c.35delG	2	Reference	2	0,80 - 1,20	0,98 - 1,02	-	YES *	0,52
S013369	CN	13:020,190	13q12.11	GJB2	Intr. 1	Reference	2	0,80 - 1,20	0,92 - 1,08	-	No	1,02
S018627	CN/WT	13:020,193	13q12.11	GJB2 WT c.-23+1G	1	Reference	2	0,80 - 1,20	0,96 - 1,04	-	No	1,07
S018628	CN	13:020,193	13q12.11	GJB2	1	Reference	2	0,80 - 1,20	0,98 - 1,02	-	No	1,03
S013196	CN	13:020,223	13q12.11	GJB6	5	Reference	2	0,80 - 1,20	0,97 - 1,03	-	No	0,98
S017417	CN	13:020,231	13q12.11	GJB6	2	Reference	2	0,80 - 1,20	0,93 - 1,07	-	No	1,07
S013216	CN	13:020,440	13q12.11	CRYL1	4	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	0,98
S024225	CN	13:020,512	13q12.11	CRYL1	2	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	1,06

analysis performed					final approval				
date					date				
signature					signature				

other		2.6.1	software versions	executed
sample id	c14c9b15-924c-4076-8c63-b536284166c3	configuration	2026-05-29 15:30:00	
barcode collection	From lot 03-009-yymmdd onwards (Default)	data conversion	2026-08-07 20:15:36	2026-09-11 05:57:42
normalisation options	individual probe normalisation	fragment analysis	2026-08-07 20:15:36	2026-09-11 05:57:46
file name	BP01-55 - Coriell NA20480.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