

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

BP01-56 - Coriell NA07945

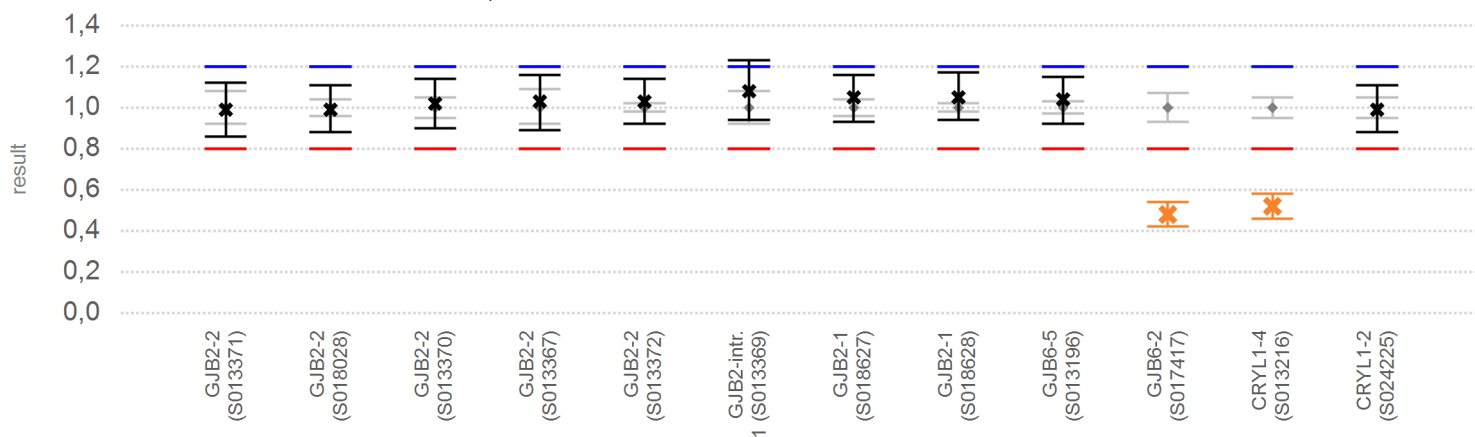
sample		identification		quality summary	
sample name	BP01-56 - Coriell NA07945	SNP id code	221120001120100121212220201021220111021v1	QC checks	Passed
sample type	Test sample	probemix lot	Identity detected	program	Ok
barcode	BP01-56	barcode lot	03-012-yymmdd	run	Ok
sex	Male	sample tracker		identity	Ok
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 (M), 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.

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 [2 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
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	0,99
S018028	CN/WT	13:020,189	13q12.11	GJB2 WT c.235delC	2	Reference	2	0,80 - 1,20	0,96 - 1,04	-	No	0,99
S013370	CN/WT	13:020,189	13q12.11	GJB2 WT c.167delT	2	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	1,02
S013367	CN/WT	13:020,189	13q12.11	GJB2 WT c.101T	2	Reference	2	0,80 - 1,20	0,92 - 1,09	-	No	1,03
S013372	CN/WT	13:020,190	13q12.11	GJB2 WT c.35delG	2	Reference	2	0,80 - 1,20	0,98 - 1,02	-	No	1,03
S013369	CN	13:020,190	13q12.11	GJB2	Intr. 1	Reference	2	0,80 - 1,20	0,92 - 1,08	-	No	1,08
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	1,05
S018628	CN	13:020,193	13q12.11	GJB2	1	Reference	2	0,80 - 1,20	0,98 - 1,02	-	No	1,05
S013196	CN	13:020,223	13q12.11	GJB6	5	Reference	2	0,80 - 1,20	0,97 - 1,03	-	No	1,04
S017417	CN	13:020,231	13q12.11	GJB6	2	Reference	2	0,80 - 1,20	0,93 - 1,07	-	YES *	0,48
S013216	CN	13:020,440	13q12.11	CRYL1	4	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,52
S024225	CN	13:020,512	13q12.11	CRYL1	2	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	0,99

analysis performed		final approval	
date		date	
signature		signature	

other		2.6.1	software versions	executed
sample id	ab1e435f-93b4-4e86-becc-40ab959335e7	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 05:57:42
normalisation options	individual probe normalisation	fragment analysis	2026-08-07 20:15:36	2026-09-11 05:57:46
file name	BP01-56 - Coriell NA07945.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