

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

BP01-60 - Coriell NA05875

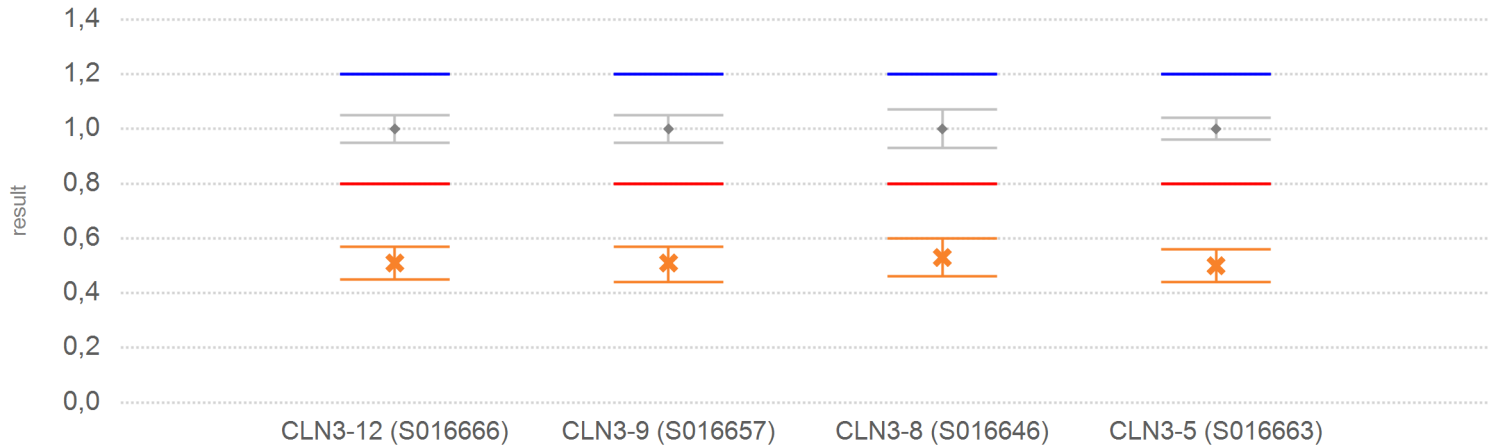
sample		identification		quality summary	
sample name	BP01-60 - Coriell NA05875	SNP id code	10212010000111121111112022020121212110v1	QC checks	Passed
sample type	Test sample	probemix lot	Identity detected	program	Ok
barcode	BP01-60	barcode lot	03-012-yymmdd	run	Ok
sex	Female	sample tracker		identity	Ok
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 (F), 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.

Juvenile Neuronal Ceroid Lipofuscinosis (CLN3-related) [1 aberrant subpanel]

CLN3 [1 aberrant segment]

There is a common CLN3 deletion region of 1.02 kb (ClinVar ID 3552). Probes S016657 and S016646 are expected to be affected in case of the 1.02 kb deletion.



Segment 1 of 1 [4 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
S016666	CN	16:028,483	16p12.1	CLN3	12	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,51
S016657	CN	16:028,486	16p12.1	CLN3	9	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,51
S016646	CN	16:028,487	16p12.1	CLN3	8	Reference	2	0,80 - 1,20	0,93 - 1,07	-	YES *	0,53
S016663	CN	16:028,489	16p12.1	CLN3	5	Reference	2	0,80 - 1,20	0,96 - 1,04	-	YES *	0,50

analysis performed		final approval	
date		date	
signature		signature	

other		2.6.1	software versions	executed
sample id	ada9e3db-4c9d-4f4e-88a4-23782d7a198d	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	BP01-60 - Coriell NA05875.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