

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

BP01-62 - Coriell NA00909

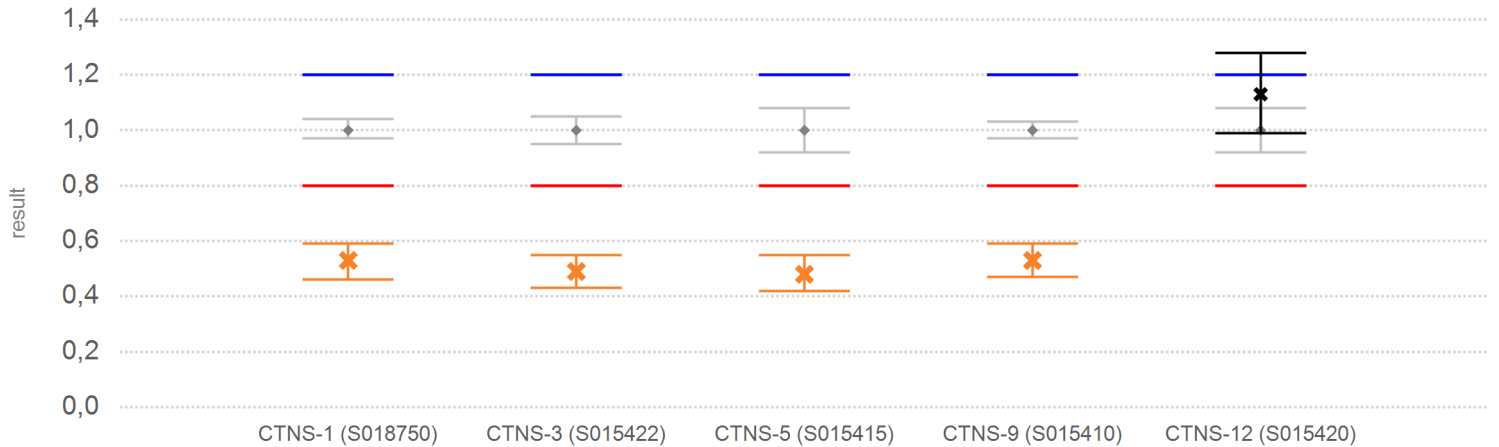
sample		identification		quality summary	
sample name	BP01-62 - Coriell NA00909	SNP id code	111022102101100222210122000102210011121v1	QC checks	Passed
sample type	Test sample	probemix lot	Identity detected	program	Ok
barcode	BP01-62	barcode lot	03-012-yymmdd	run	Ok
sex	Female	sample tracker		identity	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.

Cystinosis [1 aberrant subpanel]

CTNS [1 aberrant segment]

There is a common CTNS deletion region of 57 kb (ClinVar ID 4445). Probes S018750, S015422, S015415 and S015410 are expected to be affected in case of the 57 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
S018750	CN	17:003,637	17p13.2	CTNS	1	Reference	2	0,80 - 1,20	0,97 - 1,04	-	YES *	0,53
S015422	CN	17:003,640	17p13.2	CTNS	3	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,49
S015415	CN	17:003,649	17p13.2	CTNS	5	Reference	2	0,80 - 1,20	0,92 - 1,08	-	YES *	0,48
S015410	CN	17:003,657	17p13.2	CTNS	9	Reference	2	0,80 - 1,20	0,97 - 1,03	-	YES *	0,53
S015420	CN	17:003,661	17p13.2	CTNS	12	Reference	2	0,80 - 1,20	0,92 - 1,08	-	No	1,13

analysis performed

final approval

date	date
signature	signature

other		2.6.1	software versions	executed
sample id	644089b2-3e5a-43c2-9b1f-9eee83b92d99	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	BP01-62 - Coriell NA00909.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