

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

BP01-51 - Coriell NA20294

sample		identification		quality summary	
sample name	BP01-51 - Coriell NA20294	SNP id code	020110011110110001221101111101112201221v1	QC checks	Passed
sample type	Test sample	probemix lot	Identity detected	program	Ok
barcode	BP01-51	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

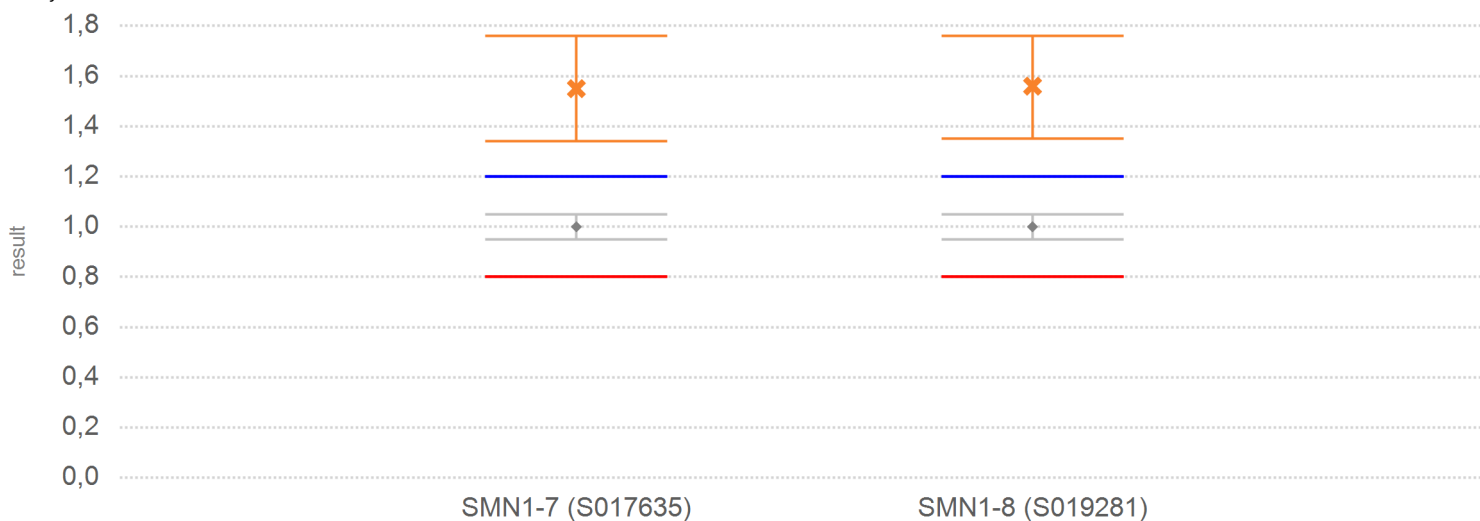
Note: The results displayed in this report are based on pre-set product-specific visualisation settings and masking (if applicable). Not all probe results may be displayed. For an overview of all results, please consult the Excel Report.

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 [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
S017635	CN	05:070,952	05q13.2	SMN1	7	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	1,55
S019281	CN	05:070,953	05q13.2	SMN1	8	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	1,56

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			-		present

analysis performed		final approval	
date		date	
signature		signature	

other		2.6.1	software versions	executed
sample id	026b8196-a662-4b8a-94fb-0d9c7fd073ca	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	BP01-51 - Coriell NA20294.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