

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

BP01-53 - Coriell NA14732

sample		identification		quality summary	
sample name	BP01-53 - Coriell NA14732	SNP id code	211112202201010202112100001101221201012v1	QC checks	Passed
sample type	Test sample	probemix lot	Identity detected	program	Ok
barcode	BP01-53	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

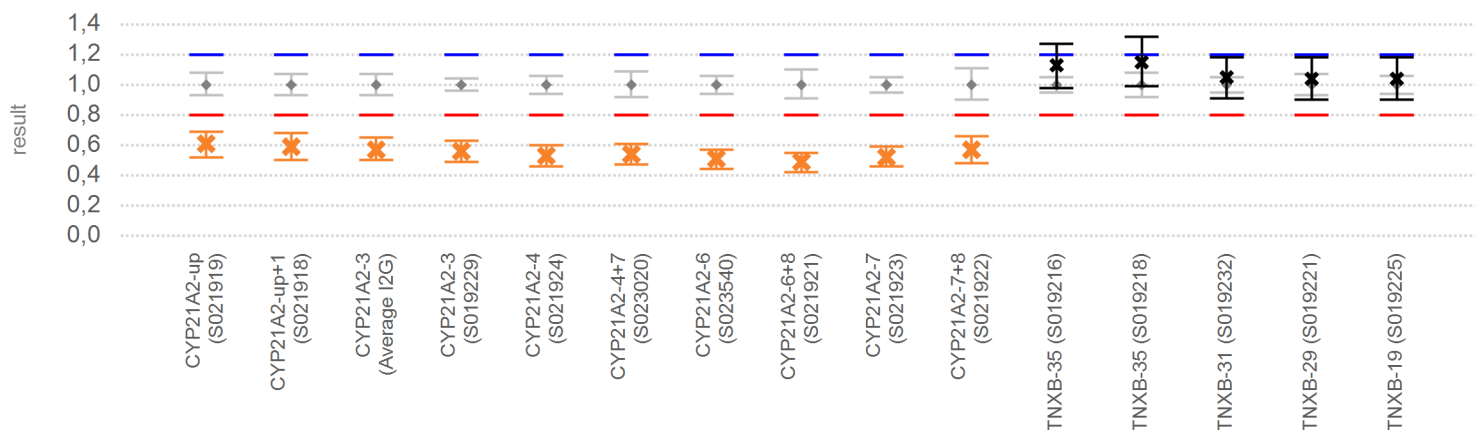
Note: The results displayed in this report are based on pre-set product-specific visualisation settings and masking (if applicable). Not all probe results may be displayed. For an overview of all results, please consult the Excel Report.

Congenital Adrenal Hyperplasia (CAH) [1 aberrant subpanel]

Genotyping CYP21A2 poses a significant challenge due to the existence of the highly homologous and inactive pseudogene CYP21A1P. Recombination between CYP21A2 and CYP21A1P can lead to the pseudogene acquiring the wildtype gene sequence, potentially obscuring the presence of pathogenic SNVs in CYP21A2. The four double wildtype probes target the wildtype CYP21A2 sequence at two locations simultaneously: one location where recombination with the pseudogene is common, and one where it is rare. The double wildtype probes are therefore specific to CYP21A2, even when CYP21A1P has acquired the wildtype sequence at one of the two targeted locations.

CYP21A2 & TNXB [1 aberrant segment]

Four double wildtype probes are included: S021918, S023020, S021921 & S021922. These probes have two ligation sites and detect the wildtype sequence at two locations simultaneously. A decreased probe read count can be due to a deletion or by the presence of one of the two (or both) mutations. The double wildtype probes require relatively long intact target DNA sequences, rendering these probes more sensitive to sample DNA fragmentation and depurination (the extent of which should be similar across all samples tested within one experiment). RPQ warnings or errors for these probes indicate issues with sample DNA fragmentation, please refer to the Product Description for more information. For each double wildtype probe, an accompanying wildtype probe is included to deduce which mutation is present in case of a decreased read count for the double wildtype probe. The four accompanying wildtype probes are S021919, S021924, S023540 & S021923 respectively. Two probes cover the CYP21A2 I2G locus, detecting the wildtype alleles A- and C. The average result of these two wildtype probes is shown by the 'Average I2G' probe. The normal copy number of this probe is 2. Only the average result is reported, not the individual I2G-A and I2G-C probe results. This probe has some residual background in absence of its specific target sequence.



Segment 1 of 1 [10 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
S021919	CN/WT	06:032,038	06p21.33	CYP21A2 WT c.-113G	Up	Reference	2	0,80 - 1,20	0,93 - 1,08	-	YES *	0,61
S021918	CN/WT	06:032,038	06p21.33	CYP21A2 WT c.-113G & c.92C	Up + 1	Reference	2	0,80 - 1,20	0,93 - 1,07	-	YES *	0,59
Average I2G	CN/WT	06:032,039	06p21.33	CYP21A2 WT c.293-13	3	Reference	2	0,80 - 1,20	0,93 - 1,07	-	YES *	0,57
S019229	CN/WT	06:032,039	06p21.33	CYP21A2 WT c.332 339del	3	Reference	2	0,80 - 1,20	0,96 - 1,04	-	YES *	0,56
S021924	CN/WT	06:032,039	06p21.33	CYP21A2 WT c.518T	4	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,53
S023020	CN/WT	06:032,039	06p21.33	CYP21A2 WT c.518T & c.844G	4 + 7	Reference	2	0,80 - 1,20	0,92 - 1,09	-	YES *	0,54
S023540	CN/WT	06:032,040	06p21.33	CYP21A2 WT c.713T	6	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,51
S021921	CN/WT	06:032,040	06p21.33	CYP21A2 WT c.713T & c.955C	6 + 8	Reference	2	0,80 - 1,20	0,91 - 1,10	-	YES *	0,49
S021923	CN/WT	06:032,040	06p21.33	CYP21A2 WT c.923dup	7	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,52
S021922	CN/WT	06:032,040	06p21.33	CYP21A2 WT c.923dup & c.1069C	7 + 8	Reference	2	0,80 - 1,20	0,90 - 1,11	-	YES *	0,57
S019216	CN	06:032,044	06p21.33	TNXB	35	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	1,13
S019218	CN	06:032,044	06p21.33	TNXB	35	Reference	2	0,80 - 1,20	0,92 - 1,08	-	No	1,15
S019232	CN	06:032,046	06p21.33	TNXB	31	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	1,05
S019221	CN	06:032,049	06p21.33	TNXB	29	Reference	2	0,80 - 1,20	0,93 - 1,07	-	No	1,04
S019225	CN	06:032,065	06p21.33	TNXB	19	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	1,04

CYP21A2 - MUT

Probe S013205 targets the c.1360C>T mutation (ClinVar ID 12159, rs6445, P454S).

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
S013205	MUT	06:032,041	06p21.33	CYP21A2 c.1360C>T	10	Reference	0			-	No	absent

analysis performed		final approval	
date		date	
signature		signature	

other		2.6.1	software versions	executed
sample id	612a8aa5-aea8-494e-b2d0-47ea3926be1f	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	BP01-53 - Coriell NA14732.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