

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

BP01-54 - Coriell HG01565

sample		identification		quality summary	
sample name	BP01-54 - Coriell HG01565	SNP id code	220012111211022211220002010110212002211v1	QC checks	Passed
sample type	Test sample	probemix lot	Identity detected	program	Ok
barcode	BP01-54	barcode lot	03-012-yymmdd	run	Ok
sex	Male	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 (M), Reference probes, Control Probes				
masked panels					
				DNA	Ok
				hybridisation	Ok
				ligation	Ok
				polymerase	Ok
				data analysis	Ok

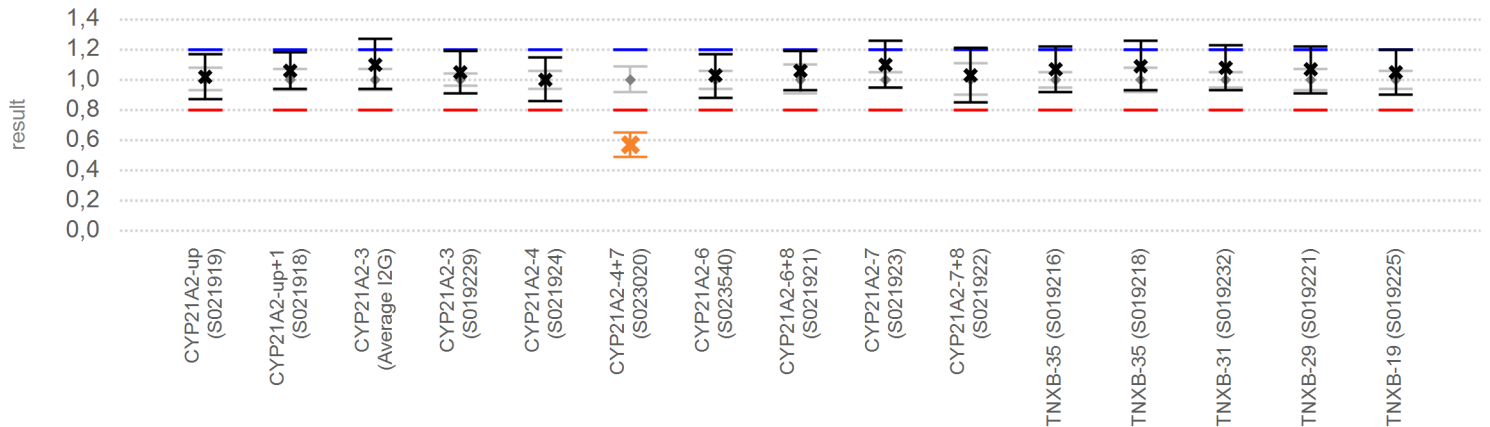
Note: The results displayed in this report are based on pre-set product-specific visualisation settings and masking (if applicable).
Not all probe results may be displayed. For an overview of all results, please consult the Excel Report.

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 [1 aberrant probe]

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	-	No	1,02
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	-	No	1,06
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	-	No	1,10
S019229	CN/WT	06:032,039	06p21.33	CYP21A2 WT c.332 339del	3	Reference	2	0,80 - 1,20	0,96 - 1,04	-	No	1,05
S021924	CN/WT	06:032,039	06p21.33	CYP21A2 WT c.518T	4	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	1,00
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 *	0,57
S023540	CN/WT	06:032,040	06p21.33	CYP21A2 WT c.713T	6	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	1,03
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	-	No	1,06
S021923	CN/WT	06:032,040	06p21.33	CYP21A2 WT c.923dup	7	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	1,10
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	-	No	1,03
S019216	CN	06:032,044	06p21.33	TNXB	35	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	1,07
S019218	CN	06:032,044	06p21.33	TNXB	35	Reference	2	0,80 - 1,20	0,92 - 1,08	-	No	1,09
S019232	CN	06:032,046	06p21.33	TNXB	31	Reference	2	0,80 - 1,20	0,95 - 1,05	-	No	1,08
S019221	CN	06:032,049	06p21.33	TNXB	29	Reference	2	0,80 - 1,20	0,93 - 1,07	-	No	1,07
S019225	CN	06:032,065	06p21.33	TNXB	19	Reference	2	0,80 - 1,20	0,94 - 1,06	-	No	1,05

CYP21A2 - MUT

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

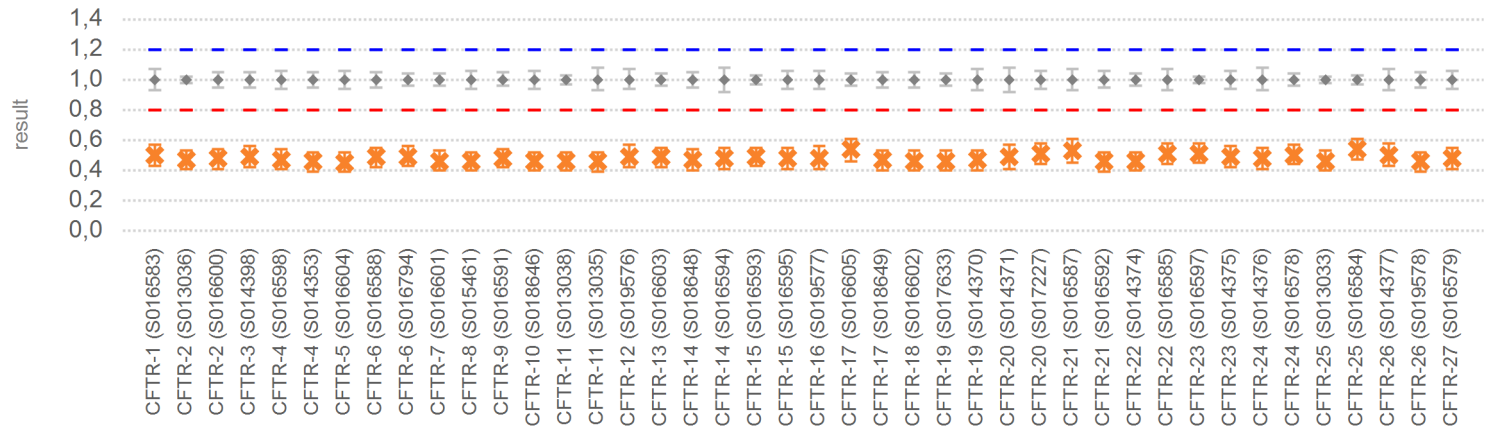
Segment 1 of 1

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

Cystic Fibrosis (CF) [1 aberrant subpanel]

CFTR [1 aberrant segment]

The CN/WT probe S013035 targets the wildtype sequence at the location of the very frequent F508del mutation (ClinVar ID 7105, rs113993960). A decreased read count for probe S013035 can also be due to the I507del (ClinVar ID 7106, rs121908745), or a deletion.



Segment 1 of 1 [42 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
S016583	CN	07:117,480	07q31.2	CFTR	1	Reference	2	0,80 - 1,20	0,93 - 1,07	-	YES *	0,50
S013036	CN	07:117,504	07q31.2	CFTR	2	Reference	2	0,80 - 1,20	0,98 - 1,02	-	YES *	0,47
S016600	CN	07:117,504	07q31.2	CFTR	2	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,48
S014398	CN	07:117,509	07q31.2	CFTR	3	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,49
S016598	CN	07:117,531	07q31.2	CFTR	4	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,47
S014353	CN/WT	07:117,531	07q31.2	CFTR WT c.489+1G	4	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,46
S016604	CN	07:117,534	07q31.2	CFTR	5	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,45
S016588	CN	07:117,535	07q31.2	CFTR	6	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,49
S016794	CN/WT	07:117,535	07q31.2	CFTR WT c.723_743+1del	6	Reference	2	0,80 - 1,20	0,96 - 1,04	-	YES *	0,49
S016601	CN	07:117,537	07q31.2	CFTR	7	Reference	2	0,80 - 1,20	0,96 - 1,04	-	YES *	0,46
S015461	CN/WT	07:117,540	07q31.2	CFTR WT c.1040G	8	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,46
S016591	CN	07:117,542	07q31.2	CFTR	9	Reference	2	0,80 - 1,20	0,96 - 1,05	-	YES *	0,48
S018646	CN	07:117,549	07q31.2	CFTR	10	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,46
S013038	CN	07:117,560	07q31.2	CFTR	11	Reference	2	0,80 - 1,20	0,97 - 1,03	-	YES *	0,46
S013035	CN/WT	07:117,560	07q31.2	CFTR WT c.1521_1523del	11	Reference	2	0,80 - 1,20	0,93 - 1,08	-	YES *	0,46
S019576	CN/WT	07:117,588	07q31.2	CFTR WT c.1624G	12	Reference	2	0,80 - 1,20	0,94 - 1,07	-	YES *	0,49
S016603	CN	07:117,590	07q31.2	CFTR	13	Reference	2	0,80 - 1,20	0,96 - 1,04	-	YES *	0,49
S018648	CN/WT	07:117,592	07q31.2	CFTR WT c.1820_1903del	14	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,47
S016594	CN	07:117,593	07q31.2	CFTR	14	Reference	2	0,80 - 1,20	0,92 - 1,08	-	YES *	0,48
S016593	CN	07:117,595	07q31.2	CFTR	15	Reference	2	0,80 - 1,20	0,97 - 1,03	-	YES *	0,49
S016595	CN	07:117,595	07q31.2	CFTR	15	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,48
S019577	CN	07:117,603	07q31.2	CFTR	16	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,48
S016605	CN	07:117,604	07q31.2	CFTR	17	Reference	2	0,80 - 1,20	0,96 - 1,04	-	YES *	0,54
S018649	CN/WT	07:117,604	07q31.2	CFTR WT c.2859_2890del	17	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,47
S016602	CN	07:117,607	07q31.2	CFTR	18	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,46
S017633	CN	07:117,611	07q31.2	CFTR	19	Reference	2	0,80 - 1,20	0,96 - 1,04	-	YES *	0,46
S014370	CN	07:117,611	07q31.2	CFTR	19	Reference	2	0,80 - 1,20	0,93 - 1,07	-	YES *	0,47
S014371	CN/WT	07:117,612	07q31.2	CFTR WT c.3140-26A	20	Reference	2	0,80 - 1,20	0,92 - 1,08	-	YES *	0,49
S017227	CN	07:117,612	07q31.2	CFTR	20	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,51
S016587	CN	07:117,615	07q31.2	CFTR	21	Reference	2	0,80 - 1,20	0,93 - 1,07	-	YES *	0,53
S016592	CN	07:117,615	07q31.2	CFTR	21	Reference	2	0,80 - 1,20	0,95 - 1,06	-	YES *	0,46
S014374	CN/WT	07:117,628	07q31.2	CFTR WT c.3528del	22	Reference	2	0,80 - 1,20	0,96 - 1,04	-	YES *	0,46
S016585	CN	07:117,628	07q31.2	CFTR	22	Reference	2	0,80 - 1,20	0,93 - 1,07	-	YES *	0,51
S016597	CN	07:117,642	07q31.2	CFTR	23	Reference	2	0,80 - 1,20	0,98 - 1,02	-	YES *	0,51
S014375	CN/WT	07:117,643	07q31.2	CFTR WT c.3846G	23	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,49
S014376	CN/WT	07:117,653	07q31.2	CFTR WT c.3909C	24	Reference	2	0,80 - 1,20	0,93 - 1,08	-	YES *	0,48
S016578	CN	07:117,653	07q31.2	CFTR	24	Reference	2	0,80 - 1,20	0,96 - 1,04	-	YES *	0,50
S013033	CN	07:117,665	07q31.2	CFTR	25	Reference	2	0,80 - 1,20	0,98 - 1,02	-	YES *	0,46
S016584	CN	07:117,665	07q31.2	CFTR	25	Reference	2	0,80 - 1,20	0,97 - 1,03	-	YES *	0,54
S014377	CN	07:117,665	07q31.2	CFTR	26	Reference	2	0,80 - 1,20	0,93 - 1,07	-	YES *	0,50
S019578	CN	07:117,666	07q31.2	CFTR	26	Reference	2	0,80 - 1,20	0,95 - 1,05	-	YES *	0,46
S016579	CN	07:117,667	07q31.2	CFTR	27	Reference	2	0,80 - 1,20	0,94 - 1,06	-	YES *	0,48

analysis performed

final approval

date	date
signature	signature

other		2.6.1	software versions	executed
sample id	3552bc2d-be0b-43f6-abe3-579f17beb7de	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	BP01-54 - Coriell HG01565.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