Sample Results Report - BP02-29 - NA05832

Coffalyser digitalMLPA

General Information

Software information	Executed	Program version
Configuration	n/a	2022-10-20 12:34
Data conversion	2023-04-19 06:53	2021-10-19 09:58
Fragment analysis	2023-04-19 06:53	2021-10-19 09:58
Comparative analysis	2023-04-19 06:53	2021-10-19 09:58
Extended analysis	n/a	n/a
Reporting	2023-04-19 06:54	2021-10-19 09:58

Sample	
Sample name (type)	**BP02-29 - NA05832 (Undefined)**
Barcode - well (lot)	BP02-29 - E04 (03-008-yymmdd)
SNP code	12001.21011.10001.12201.21021.11212.02201.2012 v1
Gender	Male
Estimated DNA quantity	27 ng
Sample track probes	(none)

Experiment settings	
Experiment name (type)	D006 Demoset (digitalMLPA)
Product name	D006-D006-A1 tube AB4B0D 13-10-2022 [default sheet]
Product detection	Identity detected
Digital signatures	All signatures valid
Probe catalogue	Default, Version 2022-10-20 12:34
Source files	C:\D006-A1 Demoset\Demoset_D006-A1 final.fastq

Analysis details	
Normalisation method	Individual reference probe normalisation [default]
Coffa file location	C:\D006-A1 Demoset\20230419_064513\D006 Demoset\BP02-29 - NA05832.Coffa
Coffa file id	9e7572ee-ab7e-4d73-b416-9bd9ea369360
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	661	1.667.931
Unrecognised	n/a	51.185
Median total reads	n/a	2.842
Median distinct reads	n/a	1.851

Probes	Expected	Detected	Unexpected
Test probes	508	508	0
Reference probes	81	81	0
Y-control probes	3	3	n/a
Mutation probes	0-1	0	n/a

Quality Details

Quality Checks: Passed

Quality		
Category	**Description**	**Classification**
Reaction analysis	Read depth	Ok
	Sequence quality	Ok
	Unrecognised reads	Ok
	Sample DNA denaturation	Ok
	Hybridisation Tm	Ok
	Hybridisation completeness	Ok
	Ligation start temperature	Ok
	Ligase activity	Ok
	Ligase inactivation	Ok
	Polymerase activity	Ok
Comparative analysis	Sample DNA denaturation	Ok
	Sample DNA depurination	Ok
	Sample DNA fragment length	Ok
	Hybridisation Tm	Ok
	Hybridisation completeness	Ok
	Ligase activity	Ok
	Polymerase activity	Ok
	Reference probe quality	Ok
	Reference sample quality	Ok

RESEARCH ONLY

Coffalyser digitalMLPA

Result Summary

Note: Please refer to the 'Probe Information File' for additional information that may be crucial for correct result interpretation.

Chart(s)

RESEARCH USE ONLY

Mutation-specific probe(s)

Gene	Mutation	Probe	Mutation status	Additional information
BRAF	V600E	S011487	Absent	p.V600E = c.1799T>A

Aberrant region(s) table

Region	Chromo-somal band (hg38)	Region size	Region ratio	Stdev	Probes in region	Probe number	Probe function	Mapview (hg38) in kb	Normal copy number	Gene	Exon	Inter ratio	Within normal range	Different from reference population	Ambi-guous
13Q	13q12.3	50 b	0,96		1	S011089	CN	13-030.241	2	KATNAL1		0,96	AsExpected	Equal	False
	13q14.11 - 13q34	70,3 Mb	1,49	0,03	22	S011537	CN	13-043.413	2	ENOX1		1,45	TooHigh	Increased2StdDev	False
						S011809	CN	13-048.347	2	RB1	intr. 4	1,47	TooHigh	Increased2StdDev	False
						S011434	CN	13-048.363	2	RB1	8	1,50	TooHigh	Increased2StdDev	False
						S015864	CN	13-048.380	2	RB1	16	1,47	TooHigh	Increased2StdDev	False
						S011509	CN	13-048.477	2	RB1	26	1,55	TooHigh	Increased2StdDev	False
						S015858	CN	13-048.480	2	RB1	27	1,48	TooHigh	Increased2StdDev	False
						S011510	CN	13-048.512	2	RCBTB2		1,50	TooHigh	Increased2StdDev	False
						S011508	CN	13-049.983	2	DLEU2		1,42	TooHigh	Increased2StdDev	False
						S011538	CN	13-050.021	2	KCNRG		1,59	TooHigh	Increased2StdDev	False
						S011539	CN	13-050.049	2	MIR15A		1,47	TooHigh	Increased2StdDev	False
						S011435	CN	13-050.105	2	DLEU1	3	1,49	TooHigh	Increased2StdDev	False
						S011541	CN	13-050.310	2	DLEU1	down	1,47	TooHigh	Increased2StdDev	False
						S011543	CN	13-050.713	2	DLEU7		1,41	TooHigh	Increased2StdDev	False
						S011511	CN	13-050.957	2	RNASEH2B		1,50	TooHigh	Increased2StdDev	False
						S011540	CN	13-051.958	2	ATP7B		1,55	TooHigh	Increased2StdDev	False
						S014299	CN	13-052.436	2	VPS36		1,51	TooHigh	Increased2StdDev	False
						S011542	CN	13-052.845	2	PCDH8		1,49	TooHigh	Increased2StdDev	False
						S012572	CN	13-069.708	2	KLHL1		1,49	TooHigh	Increased2StdDev	False
						S011436	CN	13-072.762	2	DIS3	17	1,49	TooHigh	Increased2StdDev	False
						S011437	CN	13-072.775	2	DIS3	6	1,53	TooHigh	Increased2StdDev	False
						S013121	CN	13-111.218	2	ARHGEF7		1,48	TooHigh	Increased2StdDev	False
						S013151	CN	13-113.670	2	GRK1		1,51	TooHigh	Increased2StdDev	False
18Q	18q11.2 - 18q21.1	26,0 Mb	0,99	0,03	4	S011526	CN	18-023.535	2	NPC1		1,01	AsExpected	Equal	False
						S011110	CN	18-023.572	2	NPC1		1,04	AsExpected	Equal	False
						S011111	CN	18-046.506	2	LOXHD1		0,97	AsExpected	Equal	False
						S011112	CN	18-049.581	2	LIPG		0,97	AsExpected	Equal	False
	18q23	263,7 Kb	0,50	0,04	2	S013109	CN	18-079.713	2	CTDP1		0,47	TooLow	Decreased2StdDev	False

RESEARCH USE ONLY

Sample Results Report - BP02-29 - NA05832

Coffalyser digitalMLPA

Region	Chromo-somal band (hg38)	Region size	Region ratio	Stdev	Probes in region	Probe number	Probe function	Mapview (hg38) in kb	Normal copy number	Gene	Exon	Inter ratio	Within normal range	Different from reference population	Ambi-guous
18Q	18q23	263,7 Kb	0,50	0,04	2	S013126	CN	18-079.977	2	TXNL4A		0,53	TooLow	Decreased2StdDev	False

	Analysis performed	Final approval
Quality Checks: Passed	Date: Signature:	Date: Signature:

RESEARCH USE ONLY