Sample Results Report - BP02-22 - NA05067

Coffalyser digitalMLPA

General Information

Software information	Executed	Program version
Configuration	n/a	2022-10-20 12:34
Data conversion	2023-04-19 06:52	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 06:53	2021-10-19 09:58

Sample	
Sample name (type)	**BP02-22 - NA05067 (Undefined)**
Barcode - well (lot)	BP02-22 - F03 (03-008-yymmdd)
SNP code	01002.11011.20201.02112.10211.11122.10101.2112 v1
Gender	Male
Estimated DNA quantity	16 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\BP02-22 - NA05067.Coffa
Coffa file id	fa4f1688-28e5-4743-9de9-0c2d5a711d83
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	660	1.688.307
Unrecognised	n/a	70.082
Median total reads	n/a	2.845
Median distinct reads	n/a	1.525

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

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Result Summary

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

Chart(s)

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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
09P	9p24.3 - 9p13.2	37,4 Mb	1,56	0,04	7	S013022	CN	09-000.428	2	DOCK8		1,56	TooHigh	Increased2StdDev	False
						S015454	CN	09-005.078	2	JAK2		1,54	TooHigh	Increased2StdDev	False
						S013009	CN	09-006.559	2	GLDC		1,51	TooHigh	Increased2StdDev	False
						S011072	CN	09-014.813	2	FREM1		1,46	TooHigh	Increased2StdDev	False
						S011071	CN	09-016.583	2	BNC2		1,56	TooHigh	Increased2StdDev	False
						S011069	CN	09-037.518	2	FBXO10		1,65	TooHigh	Increased2StdDev	False
						S011070	CN	09-037.842	2	DCAF10		1,58	TooHigh	Increased2StdDev	False
14Q	14q11.2 - 14q32.33	84,1 Mb	1,01	0,01	9	S011090	CN	14-021.394	2	CHD8		1,01	AsExpected	Equal	False
						S011091	CN	14-053.073	2	DDHD1		0,98	AsExpected	Equal	False
						S011092	CN	14-054.702	2	SAMD4A		1,01	AsExpected	Equal	False
						S011512	CN	14-074.480	2	NPC2		1,00	AsExpected	Equal	False
						S013378	CN	14-102.003	2	DYNC1H1		1,01	AsExpected	Equal	False
						S011513	CN	14-102.870	2	TRAF3	3	1,00	AsExpected	Equal	False
						S020522	CN	14-102.903	2	TRAF3	11	1,06	AsExpected	Equal	False
						S013391	CN	14-103.590	2	COA8		1,02	AsExpected	Equal	False
						S013358	CN	14-105.454	2	MTA1		1,10	AsExpected	Increased1StdDev	False
	14q32.33	18,6 Kb	0,52	0,02	3	S011441	CN	14-105.838	2	IGHD	6	0,54	TooLow	Decreased2StdDev	False
						S011813	CN	14-105.854	2	IGHM	4	0,52	TooLow	Decreased2StdDev	False
						S011812	CN	14-105.856	2	IGHM	1	0,50	TooLow	Decreased2StdDev	False

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

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