Sample Results Report - BP02-23 - NA10925

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 06:53	2021-10-19 09:58

Sample	
Sample name (type)	**BP02-23 - NA10925 (Undefined)**
Barcode - well (lot)	BP02-23 - G03 (03-008-yymmdd)
SNP code	12001.01011.01111.11212.11221.02211.22110.1021 v1
Gender	Male
Estimated DNA quantity	22 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\BP02-23 - NA10925.Coffa
Coffa file id	c8eca490-702f-40a2-9f38-da5eced0c6e7
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	661	1.593.807
Unrecognised	n/a	54.014
Median total reads	n/a	2.738
Median distinct reads	n/a	1.709

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
07P	7p22.2 - 7p15.3	19,6 Mb	1,01	0,01	4	S019460	CN	07-004.158	2	SDK1		1,01	AsExpected	Equal	False
						S013184	CN	07-004.822	2	RADIL		1,00	AsExpected	Equal	False
						S011057	CN	07-022.155	2	RAPGEF5		1,01	AsExpected	Equal	False
						S011058	CN	07-023.786	2	STK31		1,05	AsExpected	Equal	False
	7p12.3 - 7p12.2	4,7 Mb	0,50	0,00	5	S011055	CN	07-045.662	2	ADCY1		0,50	TooLow	Decreased2StdDev	False
						S011056	CN	07-048.278	2	ABCA13		0,50	TooLow	Decreased2StdDev	False
						S013432	CN	07-050.377	2	IKZF1	4	0,50	TooLow	Decreased2StdDev	False
						S011485	CN	07-050.387	2	IKZF1	6	0,49	TooLow	Decreased2StdDev	False
						S011486	CN	07-050.401	2	IKZF1	8	0,51	TooLow	Decreased2StdDev	False
	7p11.2	50 b	0,96		1	S014303	CN	07-055.432	2	LANCL2		0,96	AsExpected	Equal	False

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

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