Sample Results Report - BP02-30 - NA00803

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 06:54	2021-10-19 09:58

Sample	
Sample name (type)	**BP02-30 - NA00803 (Undefined)**
Barcode - well (lot)	BP02-30 - F04 (03-008-yymmdd)
SNP code	10002.11011.01011.11111.00120.10121.22000.2111 v1
Gender	Male
Estimated DNA quantity	11 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\BP02-30 - NA00803.Coffa
Coffa file id	d5fcd5f9-e88f-49c3-b1c1-73333e8140c6
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	661	1.618.735
Unrecognised	n/a	79.583
Median total reads	n/a	2.771
Median distinct reads	n/a	1.263

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
01Q	1q21.1 - 1q21.3	9,3 Mb	1,06	0,03	13	S011388	CN	01-145.707	2	PDZK1		1,06	AsExpected	Equal	False
						S011389	CN	01-147.615	2	BCL9		1,03	AsExpected	Equal	False
						S011391	CN	01-150.221	2	ANP32E	7	1,08	AsExpected	Increased1StdDev	False
						S011390	CN	01-150.231	2	ANP32E	3	1,01	AsExpected	Equal	False
						S011025	CN	01-150.444	2	RPRD2		1,05	AsExpected	Equal	False
						S011394	CN	01-150.576	2	MCL1	3	1,09	AsExpected	Increased1StdDev	False
						S011396	CN	01-150.578	2	MCL1	2	1,07	AsExpected	Equal	False
						S011026	CN	01-154.002	2	NUP210L		0,98	AsExpected	Equal	False
						S011392	CN	01-154.586	2	ADAR	12	1,11	AsExpected	Increased1StdDev	False
						S011393	CN	01-154.590	2	ADAR	7	1,06	AsExpected	Equal	False
						S011459	CN	01-154.975	2	CKS1B	intr. 1	1,08	AsExpected	Increased1StdDev	False
						S011460	CN	01-154.978	2	CKS1B	2	1,00	AsExpected	Equal	False
						S011395	CN	01-154.979	2	CKS1B	3	1,08	AsExpected	Equal	False
	1q23.1 - 1q23.3	7,3 Mb	0,50	0,03	12	S021169	CN	01-157.514	2	FCRL5	17	0,54	TooLow	Decreased2StdDev	False
						S021168	CN	01-157.519	2	FCRL5	14	0,52	TooLow	Decreased2StdDev	False
						S021167	CN	01-157.520	2	FCRL5	12	0,53	TooLow	Decreased2StdDev	False
						S011399	CN	01-160.750	2	SLAMF7	3	0,49	TooLow	Decreased2StdDev	False
						S011398	CN	01-160.753	2	SLAMF7	7	0,48	TooLow	Decreased2StdDev	False
						S011397	CN	01-163.346	2	NUF2		0,45	TooLow	Decreased2StdDev	False
						S011463	CN	01-164.321	2	PBX1-AREA	Upstream of PBX1	0,51	TooLow	Decreased2StdDev	False
						S011466	CN	01-164.331	2	PBX1-AREA	Upstream of PBX1	0,49	TooLow	Decreased2StdDev	False
						S011465	CN	01-164.337	2	PBX1-AREA	Upstream of PBX1	0,49	TooLow	Decreased2StdDev	False
						S011464	CN	01-164.366	2	PBX1-AREA	Upstream of PBX1	0,53	TooLow	Decreased2StdDev	False

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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
01Q	1q23.1 - 1q23.3	7,3 Mb	0,50	0,03	12	S011462	CN	01-164.446	2	PBX1-AREA	Upstream of PBX1	0,47	TooLow	Decreased2StdDev	False
						S011461	CN	01-164.847	2	PBX1		0,52	TooLow	Decreased2StdDev	False
	1q31.3 - 1q44	48,4 Mb	0,99	0,03	4	S011027	CN	01-196.281	2	KCNT2		1,00	AsExpected	Equal	False
						S019302	CN	01-197.583	2	DENND1B		0,96	AsExpected	Equal	False
						S013049	CN	01-244.420	2	ADSS2		0,97	AsExpected	Equal	False
						S013025	CN	01-244.687	2	DESI2		1,02	AsExpected	Equal	False

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

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