Sample Results Report - BP02-28 - NA03184

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)	**BP02-28 - NA03184 (Undefined)**
Barcode - well (lot)	BP02-28 - D04 (03-008-yymmdd)
SNP code	10102.12011.01111.21122.12120.11121.22122.0012 v1
Gender	Male
Estimated DNA quantity	20 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\BP02-28 - NA03184.Coffa
Coffa file id	5f382c8d-14b0-466e-8acd-5b6d606e5e24
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	659	1.622.159
Unrecognised	n/a	56.074
Median total reads	n/a	2.829
Median distinct reads	n/a	1.689

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
15Q	15q12 - 15q26.3	75,1 Mb	1,45	0,03	10	S011515	CN	15-026.561	2	GABRB3	8	1,40	TooHigh	Increased2StdDev	False
						S011093	CN	15-026.621	2	GABRB3	4	1,54	TooHigh	Increased2StdDev	False
						S010048	CN	15-044.574	2	SPG11		1,47	TooHigh	Increased2StdDev	False
						S010750	CN	15-048.534	2	FBN1		1,46	TooHigh	Increased2StdDev	False
						S011094	CN	15-061.913	2	VPS13C		1,44	TooHigh	Increased2StdDev	False
						S011095	CN	15-063.558	2	USP3		1,49	TooHigh	Increased2StdDev	False
						S016623	CN	15-068.214	2	CLN6		1,54	TooHigh	Increased2StdDev	False
						S011516	CN	15-098.935	2	IGF1R		1,44	TooHigh	Increased2StdDev	False
						S013163	CN	15-101.178	2	CHSY1		1,43	TooHigh	Increased2StdDev	False
						S013299	CN	15-101.647	2	TM2D3		1,43	TooHigh	Increased2StdDev	False

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

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