Sample Results Report - BP02-26 - NA02944

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)	**BP02-26 - NA02944 (Undefined)**
Barcode - well (lot)	BP02-26 - B04 (03-008-yymmdd)
SNP code	21002.11020.10012.20012.10110.00222.20112.2121 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\BP02-26 - NA02944.Coffa
Coffa file id	5463e1ba-fc58-4fb4-b062-e505b9f11498
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	662	1.701.335
Unrecognised	n/a	55.720
Median total reads	n/a	2.897
Median distinct reads	n/a	1.702

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
20P	20p13 - 20p11.21	24,5 Mb	1,55	0,01	7	S013134	CN	20-000.427	2	RBCK1		1,61	TooHigh	Increased2StdDev	False
						S013131	CN	20-000.968	2	RSPO4		1,51	TooHigh	Increased2StdDev	False
						S013070	CN	20-002.431	2	TGM6		1,54	TooHigh	Increased2StdDev	False
						S011121	CN	20-005.942	2	TRMT6		1,55	TooHigh	Increased2StdDev	False
						S011120	CN	20-009.420	2	PLCB4		1,53	TooHigh	Increased2StdDev	False
						S011119	CN	20-019.976	2	RIN2		1,55	TooHigh	Increased2StdDev	False
						S019481	CN	20-024.970	2	APMAP		1,55	TooHigh	Increased2StdDev	False
22Q	22q11.1 - 22q11.21	2,4 Mb	0,50	0,02	3	S014304	CN	22-016.992	2	GAB4		0,52	TooLow	Decreased2StdDev	False
						S014302	CN	22-017.505	2	CECR2		0,50	TooLow	Decreased2StdDev	False
						S011529	CN	22-019.409	2	HIRA		0,48	TooLow	Decreased2StdDev	False
	22q11.23 - 22q13.33	26,0 Mb	1,04	0,06	12	S020170	CN	22-023.792	2	SMARCB1	2	1,06	AsExpected	Equal	False
						S020171	CN	22-023.794	2	SMARCB1	3	1,10	AsExpected	Equal	False
						S020169	CN	22-023.803	2	SMARCB1	5	1,09	AsExpected	Equal	False
						S011530	CN	22-023.817	2	SMARCB1	6	1,10	AsExpected	Equal	False
						S011531	CN	22-023.825	2	SMARCB1	7	1,08	AsExpected	Equal	False
						S011532	CN	22-029.604	2	NF2		1,01	AsExpected	Equal	False
						S014300	CN	22-029.749	2	ZMAT5		1,06	AsExpected	Equal	False
						S014298	CN	22-031.557	2	SFI1		0,99	AsExpected	Equal	False
						S011533	CN	22-033.338	2	LARGE1		1,00	AsExpected	Equal	False
						S011534	CN	22-041.158	2	EP300		0,97	AsExpected	Equal	False
						S013069	CN	22-046.338	2	TRMU		1,03	AsExpected	Equal	False
						S013118	CN	22-049.788	2	BRD1		1,02	AsExpected	Equal	False

RESEARCH USE ONLY

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

RESEARCH USE ON