Sample Results Report - BP02-05 - NA02030

Coffalyser digitalMLPA

General Information

Software information	Executed	Program version
Configuration	n/a	2022-10-20 12:34
Data conversion	2023-04-18 14:48	2021-10-19 09:58
Fragment analysis	2023-04-18 14:49	2021-10-19 09:58
Comparative analysis	2023-04-18 14:49	2021-10-19 09:58
Extended analysis	n/a	n/a
Reporting	2023-04-18 14:50	2021-10-19 09:58

Sample	
Sample name (type)	**BP02-05 - NA02030 (Undefined)**
Barcode - well (lot)	BP02-05 - E01 (03-008-yymmdd)
SNP code	11212.11111.10212.22012.10111.01112.21121.1110 v1
Gender	Male
Estimated DNA quantity	8 ng
Sample track probes	(none)

Experiment settings	
Experiment name (type)	Demoset D001-C1 (digitalMLPA)
Product name	D001-C1-1221 [default sheet]
Product detection	Identity detected
Digital signatures	All signatures valid
Probe catalogue	Default, Version 2022-10-20 12:34
Source files	D:\Demosets\D001-C1\Demoset_D001-C1 final.fastq

Analysis details	
Normalisation method	Individual reference probe normalisation [default]
Coffa file location	C:\D001 Demoset\20230418_143831\Demoset D001-C1\BP02-05 - NA02030.Coffa
Coffa file id	879ce25c-52e7-4afe-8d7a-15d8c3e8f5d6
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	722	1.873.022
Unrecognised	n/a	55.939
Median total reads	n/a	2.790
Median distinct reads	n/a	1.065

Probes	Expected	Detected	Unexpected
Test probes	579	579	0
Reference probes	290	290	0
Y-control probes	3	3	n/a
Mutation probes	0-5	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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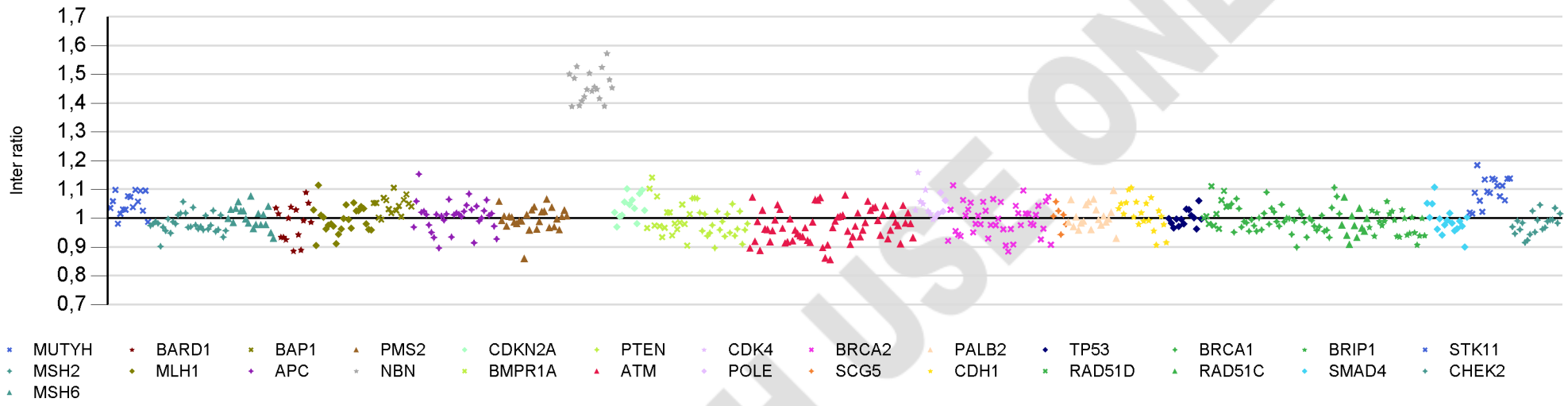
Result Summary

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
MSH2	10MBinv	S010444	Absent	MSH2 10 Mb inversion (PMID24114314); This probe consists of three oligos and has two ligation sites.
MSH2	10MBinv	S010443	Absent	MSH2 10 Mb inversion (PMID24114314); This probe consists of three oligos and has two ligation sites.
MITF	952G>A	S010555	Absent	Mutation also known as p.E318K (rs149617956).
PMS2	SVA-ins	S010670	Absent	PMS2 SVA element insertion (PMID22461402).
CHEK2	1100delC	S010117	Absent	Mutation also known as rs555607708; This probe consists of three oligos and has two ligation sites.

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
NBN	8q21.3	52,2 Kb	1,45	0,06	18	S017571	CN	08-089.934	2	NBN	16	1,50	TooHigh	Increased2StdDev	False
						S010557	CN	08-089.936	2	NBN	16	1,39	TooHigh	Increased2StdDev	False
						S017572	CN	08-089.937	2	NBN	15	1,49	TooHigh	Increased2StdDev	False
						S010559	CN	08-089.943	2	NBN	14	1,53	TooHigh	Increased2StdDev	False
						S017573	CN	08-089.946	2	NBN	13	1,39	TooHigh	Increased2StdDev	False
						S010561	CN	08-089.948	2	NBN	12	1,41	TooHigh	Increased2StdDev	False
						S010562	CN	08-089.953	2	NBN	11	1,42	TooHigh	Increased2StdDev	False
						S017574	CN	08-089.955	2	NBN	10	1,45	TooHigh	Increased2StdDev	False
						S010564	CN	08-089.959	2	NBN	09	1,50	TooHigh	Increased2StdDev	False
						S010565	CN	08-089.964	2	NBN	08	1,44	TooHigh	Increased2StdDev	False
						S017575	CN	08-089.970	2	NBN	07	1,46	TooHigh	Increased2StdDev	False
						S010567	CN	08-089.971	2	NBN	06	1,45	TooHigh	Increased2StdDev	False
						S010568	CN	08-089.978	2	NBN	05	1,42	TooHigh	Increased2StdDev	False
						S017576	CN	08-089.981	2	NBN	04	1,52	TooHigh	Increased2StdDev	False
						S010570	CN	08-089.981	2	NBN	03	1,39	TooHigh	Increased2StdDev	False
						S010571	CN	08-089.983	2	NBN	02	1,57	TooHigh	Increased2StdDev	False
						S019401	CN	08-089.985	2	NBN	01	1,48	TooHigh	Increased2StdDev	False
						S010574	CN	08-089.986	2	NBN	up	1,45	TooHigh	Increased2StdDev	False

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

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