Sample Results Report - BP02-12 - NA13451

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)	**BP02-12 - NA13451 (Undefined)**
Barcode - well (lot)	BP02-12 - D02 (03-008-yymmdd)
SNP code	21022.10011.11102.11112.01122.01211.21021.?020 v1
Gender	Male
Estimated DNA quantity	22 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\BP02-12 - NA13451.Coffa
Coffa file id	fc5e91bd-db68-4aca-be58-3f680d4caf20
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	722	1.754.454
Unrecognised	n/a	47.527
Median total reads	n/a	2.660
Median distinct reads	n/a	1.668

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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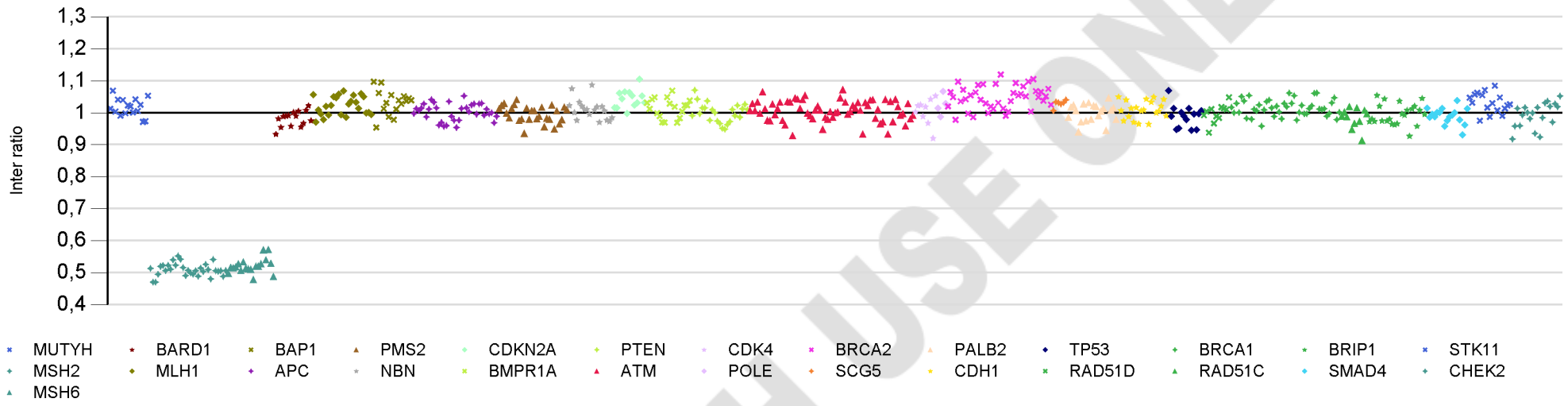
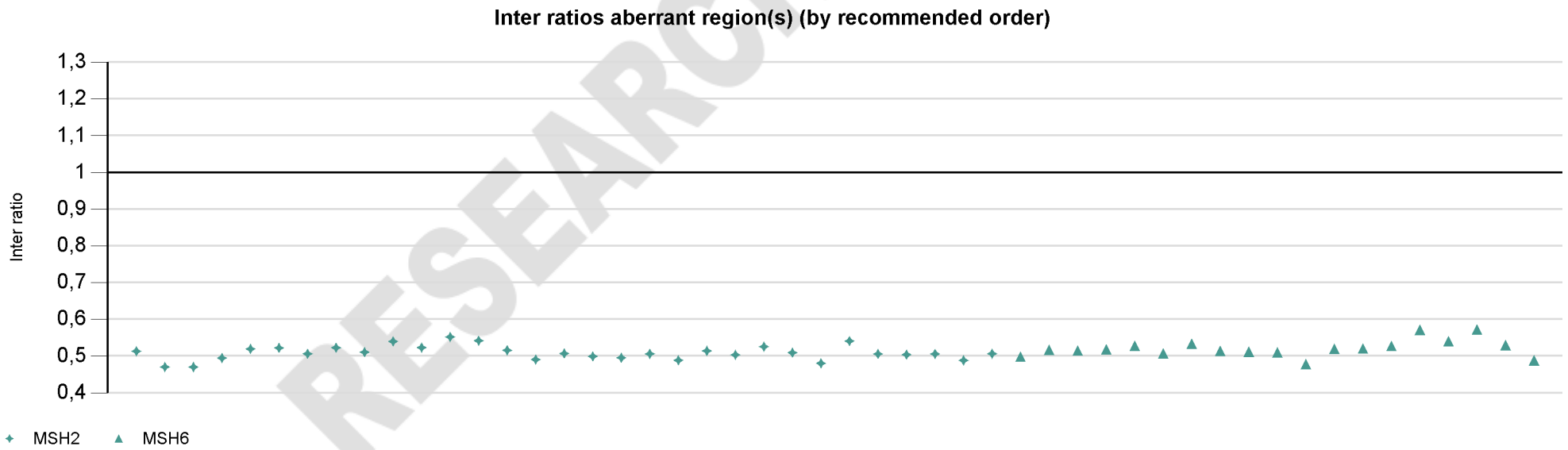
Result Summary

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
MSH2	2p21	106,6 Kb	0,51	0,02	31	S018400	CN	02-047.380	2	EPCAM	07	0,51	TooLow	Decreased2StdDev	False
						S018399	CN	02-047.385	2	EPCAM	08	0,47	TooLow	Decreased2StdDev	False
						S017525	CN	02-047.387	2	EPCAM	09	0,47	TooLow	Decreased2StdDev	False
						S010417	CN	02-047.387	2	EPCAM	09	0,49	TooLow	Decreased2StdDev	False
						S010418	CN	02-047.390	2	EPCAM	down	0,52	TooLow	Decreased2StdDev	False
						S010419	CN	02-047.396	2	MSH2	up	0,52	TooLow	Decreased2StdDev	False
						S017526	CN	02-047.399	2	MSH2	up	0,51	TooLow	Decreased2StdDev	False
						S010421	CN	02-047.403	2	MSH2	01	0,52	TooLow	Decreased2StdDev	False
						S017527	CN	02-047.403	2	MSH2	01	0,51	TooLow	Decreased2StdDev	False
						S018257	CN	02-047.403	2	MSH2	01	0,54	TooLow	Decreased2StdDev	False
						S010423	CN	02-047.404	2	MSH2	intr. 1	0,52	TooLow	Decreased2StdDev	False
						S010424	CN	02-047.408	2	MSH2	02	0,55	TooLow	Decreased2StdDev	False
						S017528	CN	02-047.410	2	MSH2	03	0,54	TooLow	Decreased2StdDev	False
						S019420	CN	02-047.410	2	MSH2	03	0,52	TooLow	Decreased2StdDev	False
						S010426	CN	02-047.412	2	MSH2	04	0,49	TooLow	Decreased2StdDev	False
						S017529	CN	02-047.414	2	MSH2	05	0,51	TooLow	Decreased2StdDev	False
						S010428	CN	02-047.416	2	MSH2	06	0,50	TooLow	Decreased2StdDev	False
						S017530	CN	02-047.430	2	MSH2	07	0,50	TooLow	Decreased2StdDev	False
						S010430	CN	02-047.430	2	MSH2	07	0,51	TooLow	Decreased2StdDev	False
						S017531	CN	02-047.446	2	MSH2	08	0,49	TooLow	Decreased2StdDev	False
						S010432	CN	02-047.446	2	MSH2	08	0,51	TooLow	Decreased2StdDev	False
						S010433	CN	02-047.463	2	MSH2	09	0,50	TooLow	Decreased2StdDev	False
						S017532	CN	02-047.467	2	MSH2	10	0,53	TooLow	Decreased2StdDev	False
						S010435	CN	02-047.471	2	MSH2	11	0,51	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
MSH2	2p21	106,6 Kb	0,51	0,02	31	S010436	CN	02-047.475	2	MSH2	12	0,48	TooLow	Decreased2StdDev	False
						S017533	CN	02-047.476	2	MSH2	13	0,54	TooLow	Decreased2StdDev	False
						S010438	CN	02-047.478	2	MSH2	14	0,51	TooLow	Decreased2StdDev	False
						S010439	CN	02-047.481	2	MSH2	15	0,50	TooLow	Decreased2StdDev	False
						S017534	CN	02-047.483	2	MSH2	16	0,51	TooLow	Decreased2StdDev	False
						S010441	CN	02-047.483	2	MSH2	16	0,49	TooLow	Decreased2StdDev	False
						S010442	CN	02-047.486	2	MSH2	down	0,51	TooLow	Decreased2StdDev	False
MSH6	2p16.3	31,0 Kb	0,52	0,01	19	S010671	CN	02-047.776	2	MSH6	up	0,50	TooLow	Decreased2StdDev	False
						S010672	CN	02-047.778	2	MSH6	up	0,52	TooLow	Decreased2StdDev	False
						S010445	CN	02-047.783	2	MSH6	01	0,52	TooLow	Decreased2StdDev	False
						S010446	CN	02-047.783	2	MSH6	01	0,52	TooLow	Decreased2StdDev	False
						S017535	CN	02-047.783	2	MSH6	01	0,53	TooLow	Decreased2StdDev	False
						S010448	CN	02-047.791	2	MSH6	02	0,51	TooLow	Decreased2StdDev	False
						S010449	CN	02-047.791	2	MSH6	02	0,53	TooLow	Decreased2StdDev	False
						S010450	CN	02-047.796	2	MSH6	03	0,51	TooLow	Decreased2StdDev	False
						S017536	CN	02-047.799	2	MSH6	04	0,51	TooLow	Decreased2StdDev	False
						S010452	CN	02-047.799	2	MSH6	04	0,51	TooLow	Decreased2StdDev	False
						S017537	CN	02-047.800	2	MSH6	04	0,48	TooLow	Decreased2StdDev	False
						S010454	CN	02-047.803	2	MSH6	05	0,52	TooLow	Decreased2StdDev	False
						S010455	CN	02-047.804	2	MSH6	05	0,52	TooLow	Decreased2StdDev	False
						S017538	CN	02-047.805	2	MSH6	06	0,53	TooLow	Decreased2StdDev	False
						S010457	CN	02-047.806	2	MSH6	07	0,57	TooLow	Decreased2StdDev	False
						S017539	CN	02-047.806	2	MSH6	08	0,54	TooLow	Decreased2StdDev	False
						S010459	CN	02-047.807	2	MSH6	09	0,57	TooLow	Decreased2StdDev	False
						S017540	CN	02-047.807	2	MSH6	10	0,53	TooLow	Decreased2StdDev	False
						S010461	CN	02-047.807	2	MSH6	down	0,49	TooLow	Decreased2StdDev	False

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

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