Sample Results Report - BP02-11 - NA07106

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)	**BP02-11 - NA07106 (Undefined)**
Barcode - well (lot)	BP02-11 - C02 (03-008-yymmdd)
SNP code	11101.11120.10011.10221.10211.12121.22121.2111 v1
Gender	Male
Estimated DNA quantity	11 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\BP02-11 - NA07106.Coffa
Coffa file id	e40a80e6-c680-4d32-b68c-f932f3783912
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	723	1.768.336
Unrecognised	n/a	83.408
Median total reads	n/a	2.601
Median distinct reads	n/a	1.239

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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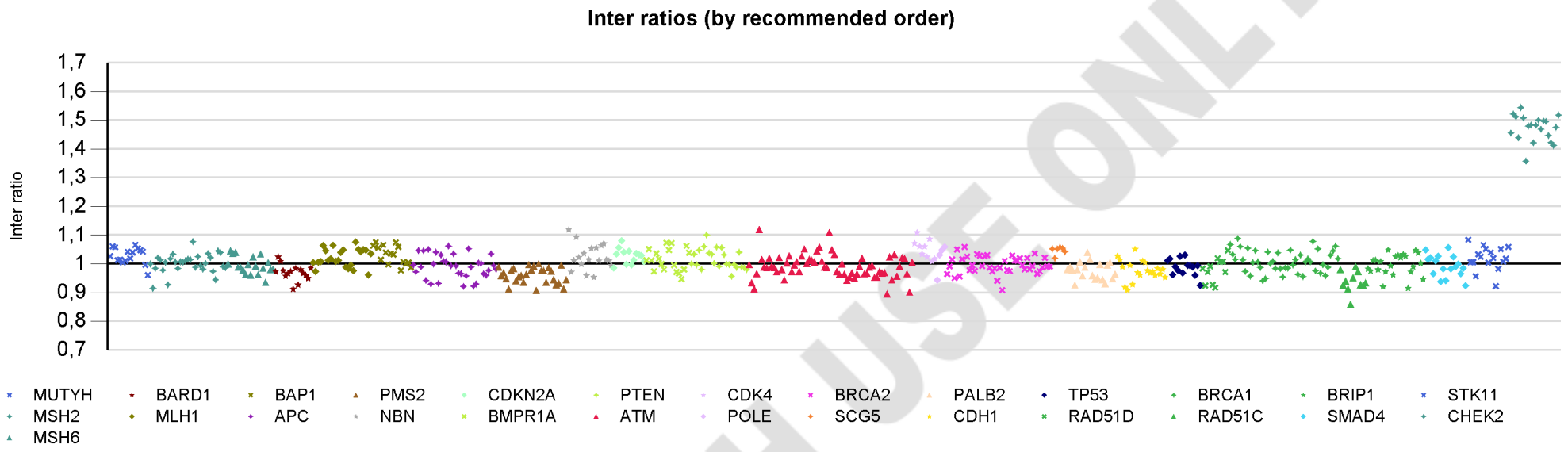
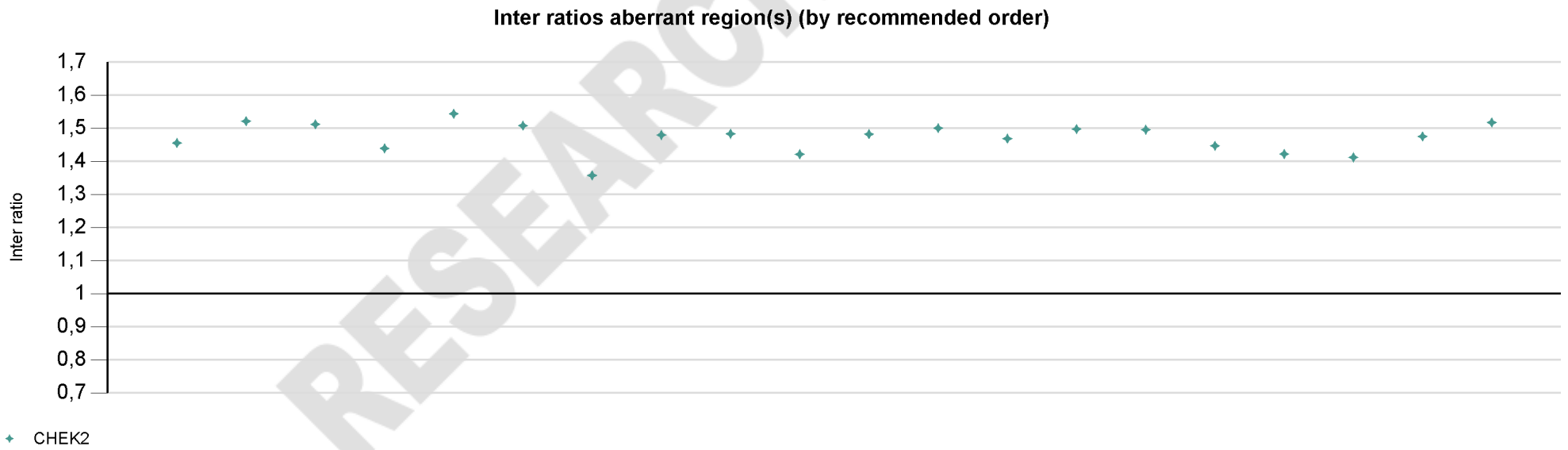
Result Summary

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
CHEK2	22q12.1	56,1 Kb	1,48	0,04	20	S010112	CN	22-028.688	2	CHEK2	15	1,46	TooHigh	Increased2StdDev	False
						S010113	CN	22-028.689	2	CHEK2	intr. 14	1,52	TooHigh	Increased2StdDev	False
						S019413	CN	22-028.689	2	CHEK2	14	1,51	TooHigh	Increased2StdDev	False
						S019416	CN	22-028.694	2	CHEK2	13	1,44	TooHigh	Increased2StdDev	False
						S010114	CN	22-028.694	2	CHEK2	13	1,54	TooHigh	Increased2StdDev	False
						S017426	CN	22-028.695	2	CHEK2	12	1,51	TooHigh	Increased2StdDev	False
						S010116	CN	22-028.696	2	CHEK2	11	1,36	TooHigh	Increased2StdDev	False
						S010118	CN	22-028.697	2	CHEK2	10	1,48	TooHigh	Increased2StdDev	False
						S017427	CN	22-028.700	2	CHEK2	09	1,48	TooHigh	Increased2StdDev	False
						S010120	CN	22-028.704	2	CHEK2	08	1,42	TooHigh	Increased2StdDev	False
						S017428	CN	22-028.710	2	CHEK2	07	1,48	TooHigh	Increased2StdDev	False
						S011000	CN	22-028.712	2	CHEK2	06	1,50	TooHigh	Increased2StdDev	False
						S017429	CN	22-028.719	2	CHEK2	05	1,47	TooHigh	Increased2StdDev	False
						S010124	CN	22-028.725	2	CHEK2	04	1,50	TooHigh	Increased2StdDev	False
						S017430	CN	22-028.725	2	CHEK2	03	1,50	TooHigh	Increased2StdDev	False
						S010126	CN	22-028.734	2	CHEK2	02	1,45	TooHigh	Increased2StdDev	False
						S019411	CN	22-028.735	2	CHEK2	02	1,42	TooHigh	Increased2StdDev	False
						S010127	CN	22-028.742	2	CHEK2	01	1,41	TooHigh	Increased2StdDev	False
						S017608	CN	22-028.742	2	CHEK2	01	1,48	TooHigh	Increased2StdDev	False
						S010129	CN	22-028.744	2	HSCB	02	1,52	TooHigh	Increased2StdDev	False

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

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