Sample Results Report - BP02-07 - NA02718

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)	**BP02-07 - NA02718 (Undefined)**
Barcode - well (lot)	BP02-07 - G01 (03-008-yymmdd)
SNP code	11112.11022.01102.20211.10111.01022.12210.1122 v1
Gender	Female
Estimated DNA quantity	23 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\BP02-07 - NA02718.Coffa
Coffa file id	cdeced8b-7a46-40e3-bbac-47f382ec084f
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	723	1.835.568
Unrecognised	n/a	49.522
Median total reads	n/a	2.796
Median distinct reads	n/a	1.742

Probes	Expected	Detected	Unexpected
Test probes	579	579	0
Reference probes	290	290	0
Y-control probes	0	0	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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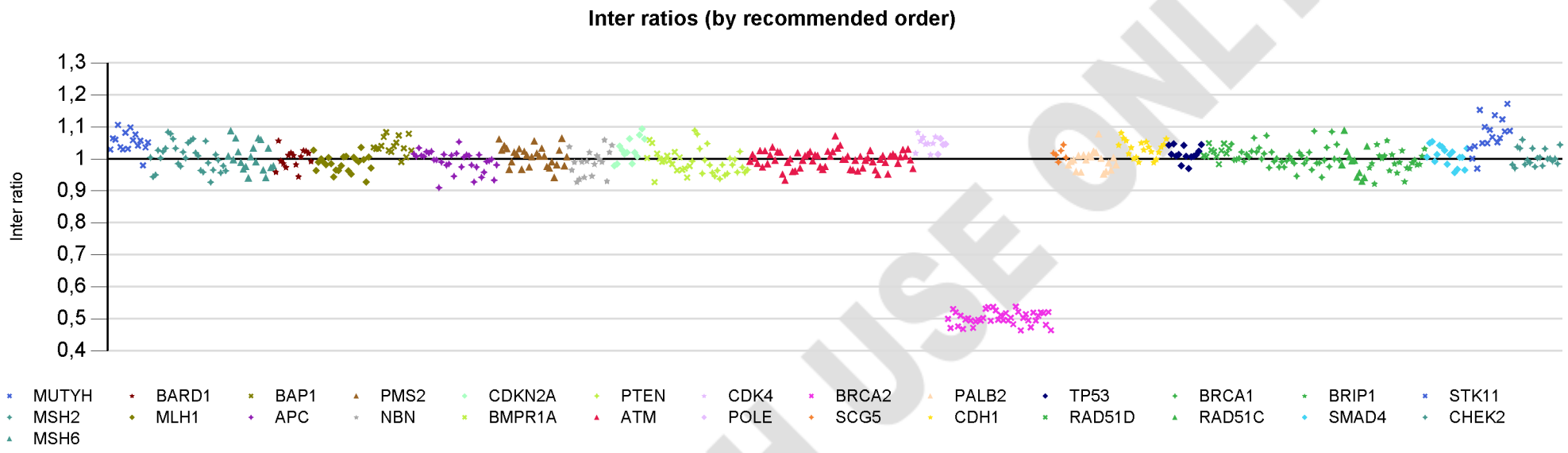
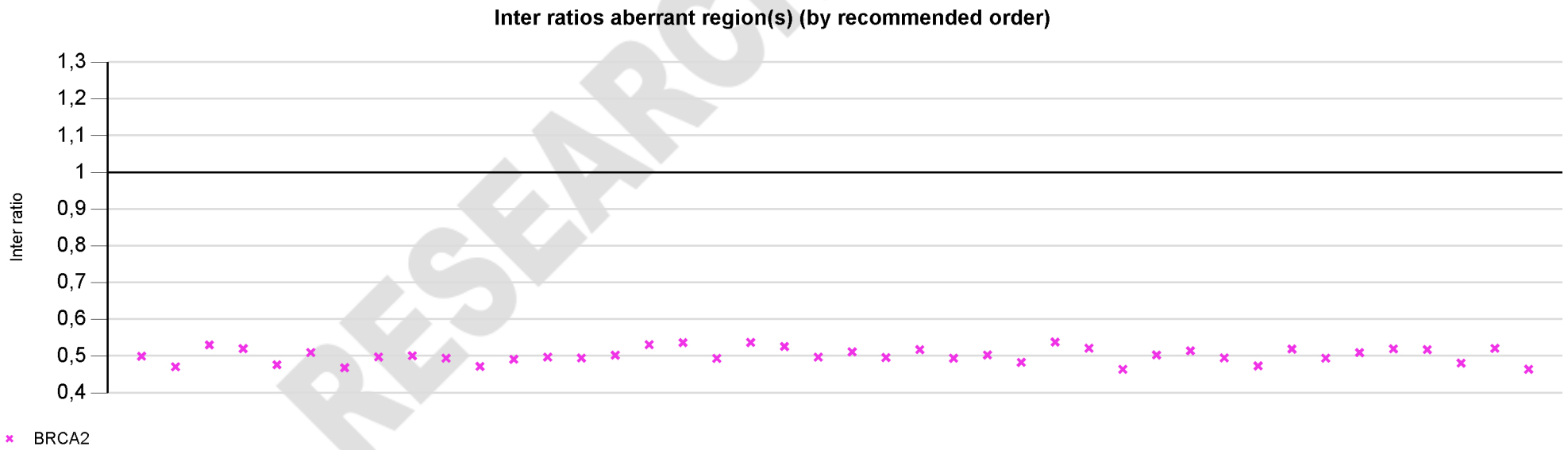
Result Summary

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.
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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
BRCA2	13q13.1	85,7 Kb	0,50	0,03	42	S010170	CN	13-032.314	2	BRCA2	up	0,50	TooLow	Decreased2StdDev	False
						S019409	CN	13-032.315	2	BRCA2	01	0,47	TooLow	Decreased2StdDev	False
						S017445	CN	13-032.316	2	BRCA2	01	0,53	TooLow	Decreased2StdDev	False
						S010173	CN	13-032.316	2	BRCA2	02	0,52	TooLow	Decreased2StdDev	False
						S019422	CN/WT	13-032.319	2	BRCA2	03	0,48	TooLow	Decreased2StdDev	False
						S017446	CN	13-032.319	2	BRCA2	03	0,51	TooLow	Decreased2StdDev	False
						S010176	CN	13-032.325	2	BRCA2	04	0,47	TooLow	Decreased2StdDev	False
						S010177	CN	13-032.326	2	BRCA2	05	0,50	TooLow	Decreased2StdDev	False
						S017447	CN	13-032.326	2	BRCA2	06	0,50	TooLow	Decreased2StdDev	False
						S010179	CN	13-032.327	2	BRCA2	07	0,49	TooLow	Decreased2StdDev	False
						S010180	CN	13-032.329	2	BRCA2	08	0,47	TooLow	Decreased2StdDev	False
						S017448	CN	13-032.331	2	BRCA2	09	0,49	TooLow	Decreased2StdDev	False
						S010182	CN	13-032.333	2	BRCA2	10	0,50	TooLow	Decreased2StdDev	False
						S010183	CN	13-032.333	2	BRCA2	10	0,49	TooLow	Decreased2StdDev	False
						S010184	CN	13-032.333	2	BRCA2	10	0,50	TooLow	Decreased2StdDev	False
						S017449	CN	13-032.336	2	BRCA2	11	0,53	TooLow	Decreased2StdDev	False
						S010186	CN	13-032.337	2	BRCA2	11	0,54	TooLow	Decreased2StdDev	False
						S017450	CN	13-032.337	2	BRCA2	11	0,49	TooLow	Decreased2StdDev	False
						S010659	CN	13-032.338	2	BRCA2	11	0,54	TooLow	Decreased2StdDev	False
						S010189	CN	13-032.339	2	BRCA2	11	0,53	TooLow	Decreased2StdDev	False
						S017451	CN	13-032.339	2	BRCA2	11	0,50	TooLow	Decreased2StdDev	False
						S010191	CN	13-032.340	2	BRCA2	11	0,51	TooLow	Decreased2StdDev	False
						S017452	CN	13-032.341	2	BRCA2	11	0,50	TooLow	Decreased2StdDev	False
						S010193	CN	13-032.341	2	BRCA2	11	0,52	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
BRCA2	13q13.1	85,7 Kb	0,50	0,03	42	S017453	CN	13-032.345	2	BRCA2	12	0,49	TooLow	Decreased2StdDev	False
						S010195	CN	13-032.347	2	BRCA2	13	0,50	TooLow	Decreased2StdDev	False
						S017454	CN	13-032.355	2	BRCA2	14	0,48	TooLow	Decreased2StdDev	False
						S010197	CN	13-032.357	2	BRCA2	15	0,54	TooLow	Decreased2StdDev	False
						S010198	CN	13-032.358	2	BRCA2	16	0,52	TooLow	Decreased2StdDev	False
						S017455	CN	13-032.363	2	BRCA2	17	0,46	TooLow	Decreased2StdDev	False
						S010200	CN	13-032.363	2	BRCA2	18	0,50	TooLow	Decreased2StdDev	False
						S010201	CN	13-032.370	2	BRCA2	19	0,51	TooLow	Decreased2StdDev	False
						S017456	CN	13-032.371	2	BRCA2	20	0,50	TooLow	Decreased2StdDev	False
						S010203	CN	13-032.377	2	BRCA2	21	0,47	TooLow	Decreased2StdDev	False
						S010204	CN	13-032.379	2	BRCA2	22	0,52	TooLow	Decreased2StdDev	False
						S017457	CN	13-032.380	2	BRCA2	23	0,49	TooLow	Decreased2StdDev	False
						S010206	CN	13-032.380	2	BRCA2	24	0,51	TooLow	Decreased2StdDev	False
						S010207	CN	13-032.395	2	BRCA2	25	0,52	TooLow	Decreased2StdDev	False
						S017458	CN	13-032.397	2	BRCA2	26	0,52	TooLow	Decreased2StdDev	False
						S010209	CN	13-032.398	2	BRCA2	27	0,48	TooLow	Decreased2StdDev	False
						S010210	CN	13-032.399	2	BRCA2	27	0,52	TooLow	Decreased2StdDev	False
						S010211	CN	13-032.399	2	BRCA2	27	0,46	TooLow	Decreased2StdDev	False

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

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