Sample Results Report - BP02-14 - NA14626

Coffalyser digitalMLPA

General Information

Software information	Executed	Program version
Configuration	n/a	2022-10-20 12:34
Data conversion	2023-04-18 14:49	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)	**BP02-14 - NA14626 (Undefined)**
Barcode - well (lot)	BP02-14 - F02 (03-008-yymmdd)
SNP code	22002.11002.10112.20121.00121.01011.12102.2121 v1
Gender	Female
Estimated DNA quantity	21 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\BP02-14 - NA14626.Coffa
Coffa file id	8354c1aa-85ca-43f4-8002-c01982cf9e71
Reference samples	REF-Male-1, REF-Male-2, REF-Male-3, REF-Male-4, REF-Male-5

Reads	Items	Reads
Recognised	722	1.828.648
Unrecognised	n/a	49.338
Median total reads	n/a	2.762
Median distinct reads	n/a	1.694

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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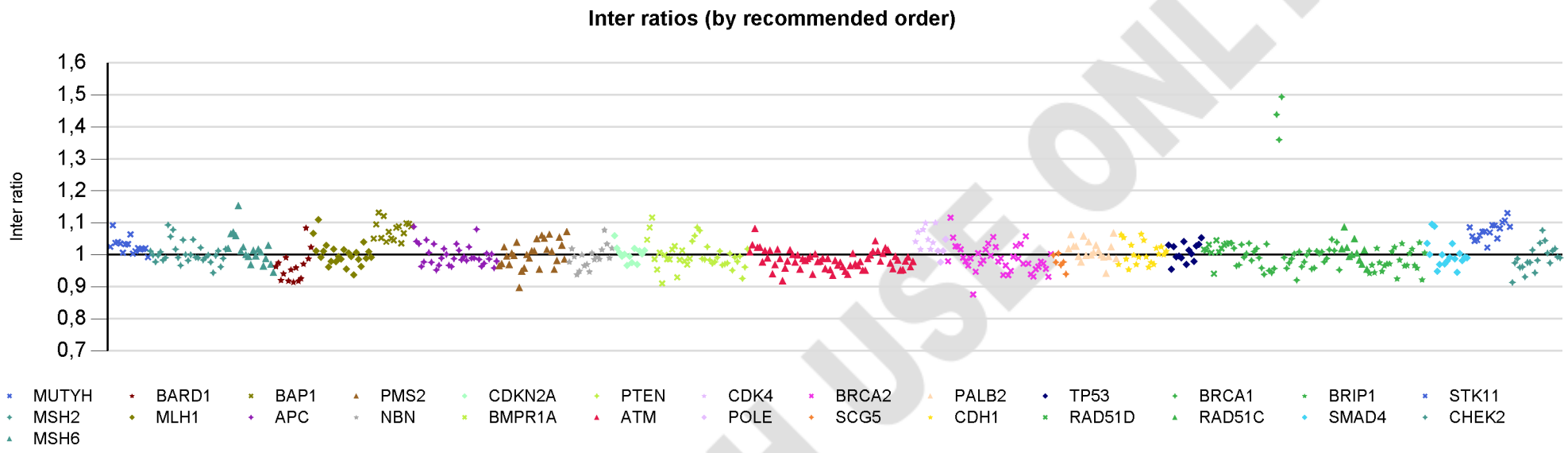
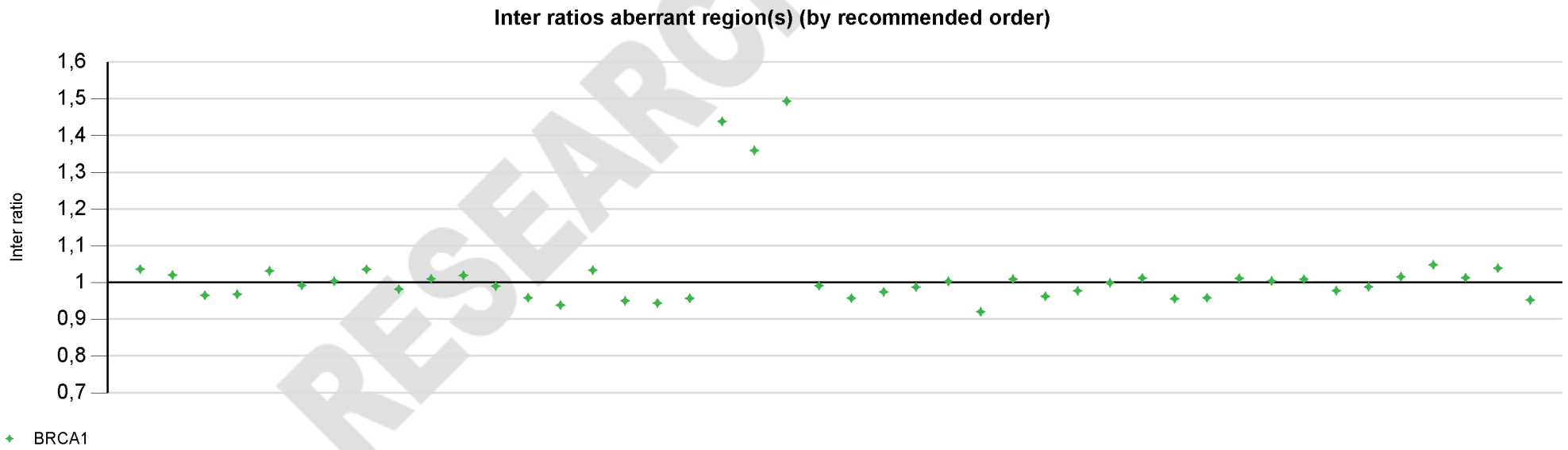
Result Summary

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
BRCA1	17q21.31	31,4 Kb	0,99	0,05	18	S017605	CN	17-043.045	2	BRCA1	24	1,04	AsExpected	Equal	False
						S010133	CN	17-043.046	2	BRCA1	24	1,02	AsExpected	Equal	False
						S017432	CN	17-043.046	2	BRCA1	24	0,97	AsExpected	Equal	False
						S010135	CN	17-043.048	2	BRCA1	23	0,97	AsExpected	Equal	False
						S010136	CN	17-043.049	2	BRCA1	22	1,03	AsExpected	Equal	False
						S013435	CN	17-043.049	2	BRCA1	intr. 21	0,99	AsExpected	Equal	False
						S017433	CN	17-043.051	2	BRCA1	21	1,00	AsExpected	Equal	False
						S010138	CN	17-043.057	2	BRCA1	20	1,04	AsExpected	Equal	False
						S013437	CN	17-043.057	2	BRCA1	20	0,98	AsExpected	Equal	False
						S010139	CN	17-043.063	2	BRCA1	19	1,01	AsExpected	Equal	False
						S017434	CN	17-043.064	2	BRCA1	18	1,02	AsExpected	Equal	False
						S013440	CN	17-043.068	2	BRCA1	17	0,99	AsExpected	Equal	False
						S010141	CN	17-043.068	2	BRCA1	17	0,96	AsExpected	Equal	False
						S010142	CN	17-043.071	2	BRCA1	16	0,94	AsExpected	Equal	False
						S017435	CN	17-043.071	2	BRCA1	16	1,03	AsExpected	Equal	False
						S010144	CN	17-043.074	2	BRCA1	15	0,95	AsExpected	Equal	False
						S019424	CN	17-043.076	2	BRCA1	14	0,94	AsExpected	Equal	False
						S010145	CN	17-043.077	2	BRCA1	14	0,96	AsExpected	Equal	False
	17q21.31	310 b	1,44	0,08	3	S017436	CN	17-043.082	2	BRCA1	intr. 13	1,44	TooHigh	Increased2StdDev	False
						S010147	CN	17-043.082	2	BRCA1	13	1,36	TooHigh	Increased2StdDev	False
						S010148	CN	17-043.082	2	BRCA1	13	1,49	TooHigh	Increased2StdDev	False
	17q21.31	39,2 Kb	0,99	0,03	23	S010149	CN	17-043.091	2	BRCA1	12	0,99	AsExpected	Equal	False
						S017437	CN	17-043.092	2	BRCA1	11	0,96	AsExpected	Equal	False
						S010151	CN	17-043.092	2	BRCA1	11	0,97	AsExpected	Equal	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
BRCA1	17q21.31	39,2 Kb	0,99	0,03	23	S017438	CN	17-043.092	2	BRCA1	11	0,99	AsExpected	Equal	False
						S010153	CN	17-043.093	2	BRCA1	11	1,00	AsExpected	Equal	False
						S010154	CN	17-043.093	2	BRCA1	11	0,92	AsExpected	Equal	False
						S017439	CN	17-043.094	2	BRCA1	11	1,01	AsExpected	Equal	False
						S010156	CN	17-043.094	2	BRCA1	11	0,96	AsExpected	Equal	False
						S017440	CN	17-043.095	2	BRCA1	11	0,98	AsExpected	Equal	False
						S010158	CN	17-043.096	2	BRCA1	10	1,00	AsExpected	Equal	False
						S017441	CN	17-043.097	2	BRCA1	09	1,01	AsExpected	Equal	False
						S010160	CN	17-043.100	2	BRCA1	08	0,96	AsExpected	Equal	False
						S013447	CN	17-043.100	2	BRCA1	08	0,96	AsExpected	Equal	False
						S017442	CN	17-043.104	2	BRCA1	07	1,01	AsExpected	Equal	False
						S010162	CN	17-043.105	2	BRCA1	06	1,00	AsExpected	Equal	False
						S010163	CN	17-043.106	2	BRCA1	05	1,01	AsExpected	Equal	False
						S013451	CN	17-043.116	2	BRCA1	intr. 3	0,98	AsExpected	Equal	False
						S017443	CN	17-043.116	2	BRCA1	03	0,99	AsExpected	Equal	False
						S010165	CN	17-043.124	2	BRCA1	02	1,02	AsExpected	Equal	False
						S010166	CN	17-043.125	2	BRCA1	intr. 1a	1,05	AsExpected	Equal	False
						S017444	CN	17-043.125	2	BRCA1	1a	1,01	AsExpected	Equal	False
						S010168	CN	17-043.126	2	BRCA1	up	1,04	AsExpected	Equal	False
						S010169	CN	17-043.130	2	BRCA1	up	0,95	AsExpected	Equal	False

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

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