



ORANGE Report

SAMPLE

Test

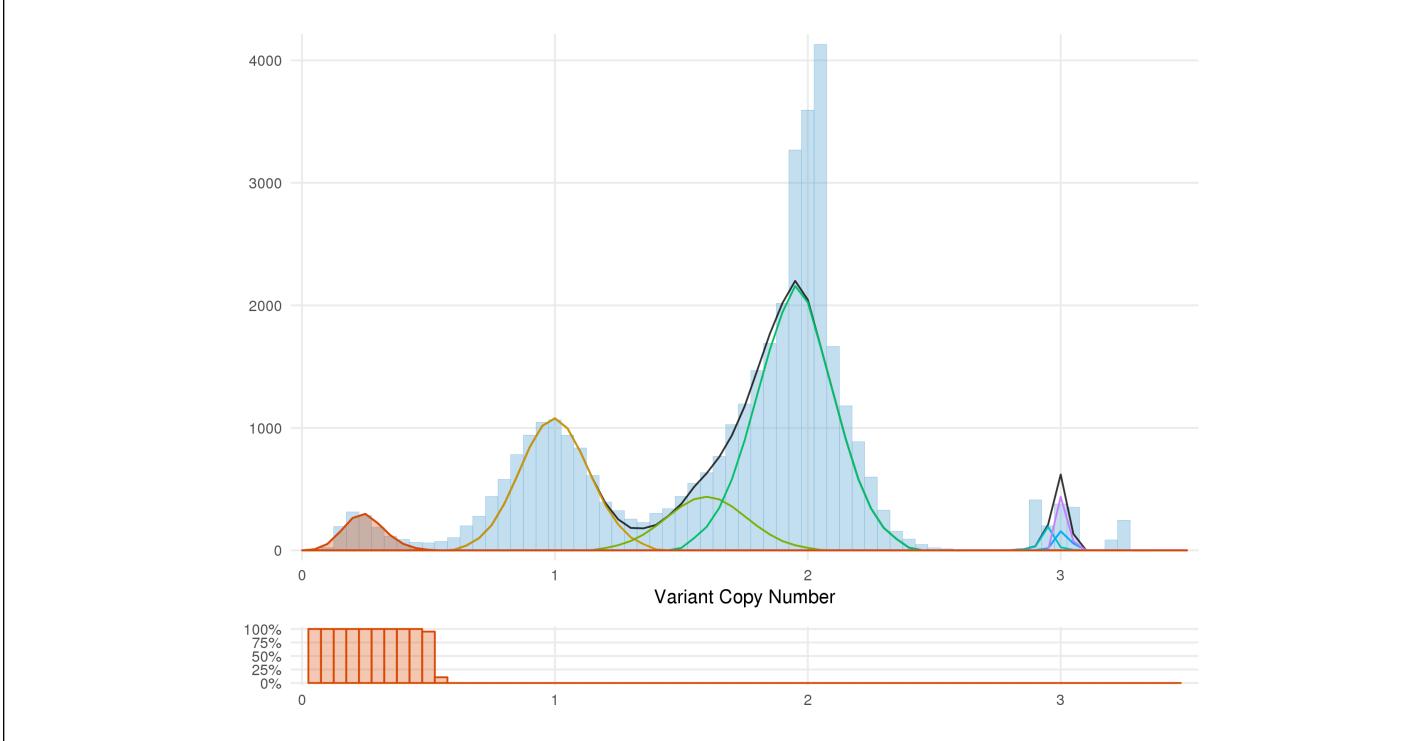
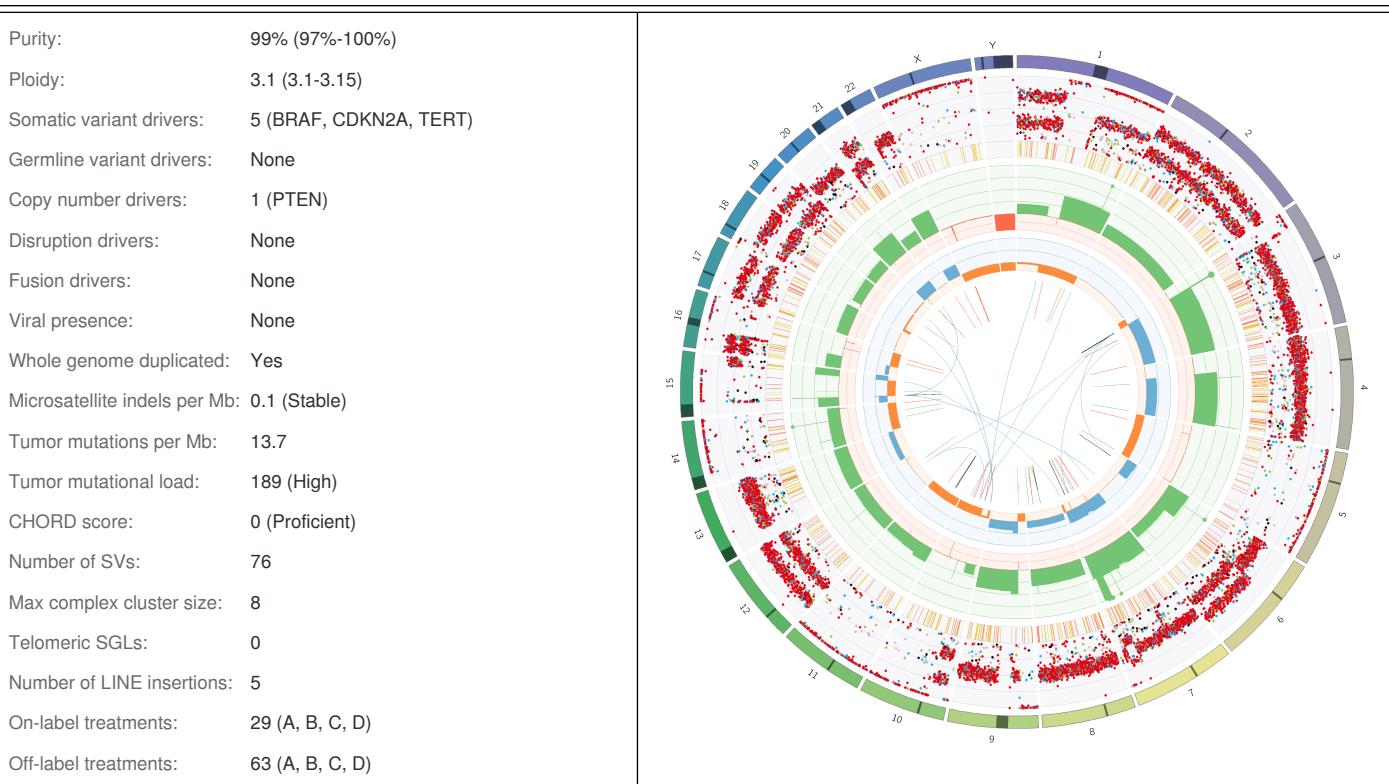
PLATINUM VERSION

5.23

CONFIGURED PRIMARY TUMOR
skin melanoma (DOID 8923)

CUPPA PRIMARY TUMOR
Melanoma (likelihood=99.6%)

QC
PASS





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Clinical Evidence

* Treatments are reported up to a maximum evidence level of B.

On-Label Evidence

TREATMENT	RESPONSIVE EVIDENCE	RESISTANCE EVIDENCE
Cobimetinib + Vemurafenib	BRAF p.V600E (A - CGI)	
Dabrafenib	BRAF p.V600E (A - CGI)	
Dabrafenib + Trametinib	BRAF p.V600E (A - CGI, CIViC)	
Trametinib	BRAF p.V600E (A - CGI)	
Vemurafenib	BRAF p.V600E (A - CGI, CIViC)	

Off-Label Evidence

TREATMENT	RESPONSIVE EVIDENCE	RESISTANCE EVIDENCE
Dabrafenib	BRAF p.V600E (A - CGI)	
Dabrafenib + Trametinib	BRAF p.V600E (A - CGI, CIViC)	
Vemurafenib	BRAF p.V600E (A - CGI)	BRAF p.V600E (B - CIViC)
CI-1040	BRAF p.V600E (B - CIViC)	
Cetuximab	BRAF p.V600E (B - CIViC)	PTEN partial loss (B - CIViC)
Cetuximab + Irinotecan + Vemurafenib	BRAF p.V600E (B - CIViC)	
Cetuximab + Vemurafenib	BRAF p.V600E (B - CIViC)	
Chemotherapy		PTEN partial loss (B - CIViC)
Everolimus		PTEN partial loss (B - CIViC)
Fluorouracil		BRAF p.V600E (B - CIViC)
Irinotecan		BRAF p.V600E (B - CIViC)
Lapatinib + Trastuzumab		PTEN partial loss (B - CIViC)
Oxaliplatin		BRAF p.V600E (B - CIViC)
Panitumumab	BRAF p.V600E (B - CIViC)	
Panitumumab + Vemurafenib	BRAF p.V600E (B - CIViC)	
Ridaforolimus		PTEN partial loss (B - CIViC)
Selumetinib	BRAF p.V600E (B - CIViC)	
Sorafenib	BRAF p.V600E (B - CIViC)	
Temsirolimus		PTEN partial loss (B - CIViC)
Trastuzumab		PTEN partial loss (B - CIViC)

Trials

NONE



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Somatic Findings

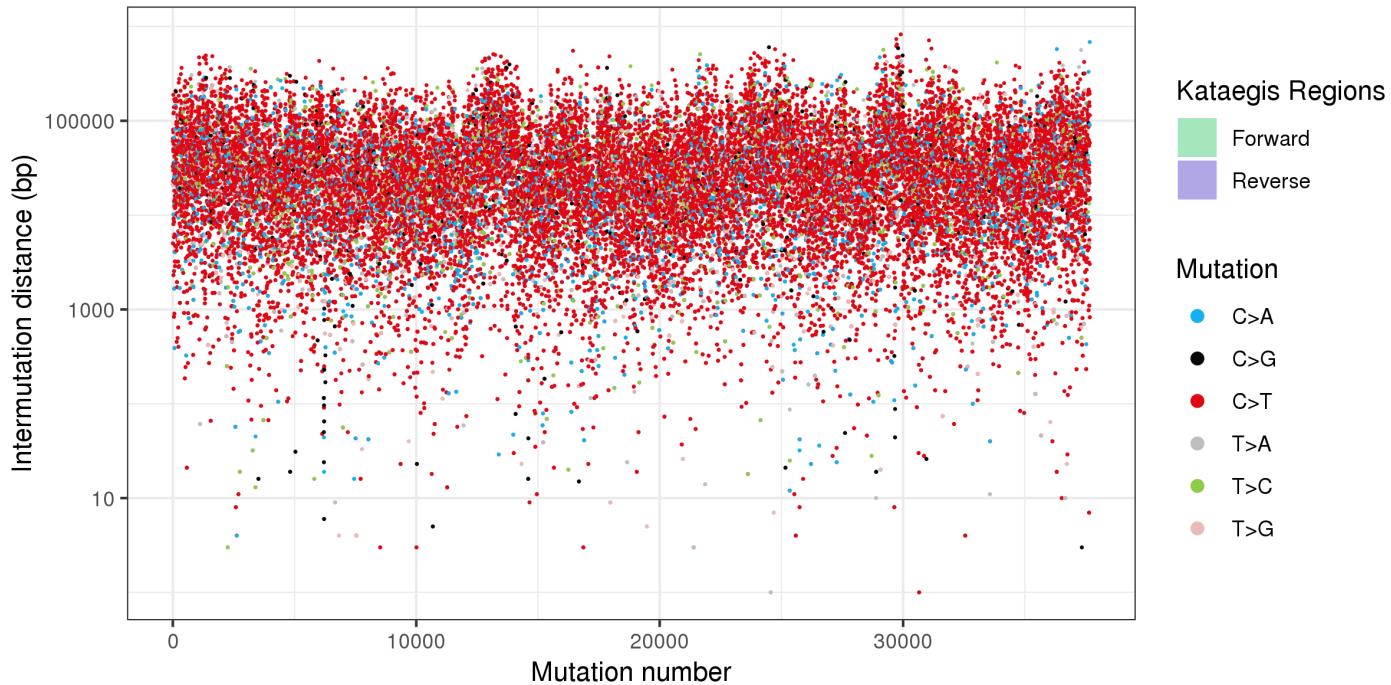
Driver variants (5)

VARIANT	VCN	CN	MACN	RNA VAF	BIALLELIC	HOTSPOT	DL	CL	PHASE ID
BRAF p.V600E	4.1	6.0	2.0	NA	No	Yes	100%	100%	
CDKN2A p.A68fs	2.0	2.0	0.0	NA	Yes	Near	100%	100%	
TERT c.-125_-124delCCinsTT	1.7	2.0	0.0	NA	Yes	Yes	100%	100%	482
SF3B1 p.P718L	2.0	3.0	1.0	NA	No	No	15%	100%	
TP63 p.M499I	1.7	4.0	2.0	NA	No	No	0%	100%	

Other potentially relevant variants (3)

VARIANT	VCN	CN	MACN	RNA VAF	BIALLELIC	HOTSPOT	DL	CL	PHASE ID
ALB c.1059-53_1059-52insT	1.8	3.9	1.9	NA	No	No		100%	
HDAC2 p.R409*	0.9	2.9	1.0	NA	No	No		100%	
STK19 p.D89N	2.0	3.8	1.8	NA	No	Yes		100%	

Kataegis plot



Driver amps/dels (1)

CHROMOSOME	REGION	GENE	TYPE	CN
10	q23.31	PTEN	partial loss	0

Other regions with amps (0)

NONE



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Other autosomal regions with dels (1)

CHROMOSOME	REGION	GENE	TYPE	CN
16	q21	CNOT1	partial loss	0

Driver fusions (0)

NONE

Other potentially interesting fusions (0)

NONE

Driver viruses (0)

NONE

Other viral presence (0)

NONE

Homozygous disruptions (0)

NONE

Gene disruptions (1)

LOCATION	GENE	RANGE	TYPE	JUNCTION CN	UNDISRUPTED CN
10q23.31	PTEN	Intron 5 -> Intron 6	DEL	2.0	0.0

Structural drivers (3)

GENE	EVENT TYPE
CDKN2A	LOH_SV_TELO
PTEN	DEL
PTEN	LOH_SV_TELO

Structural driver plots (3)



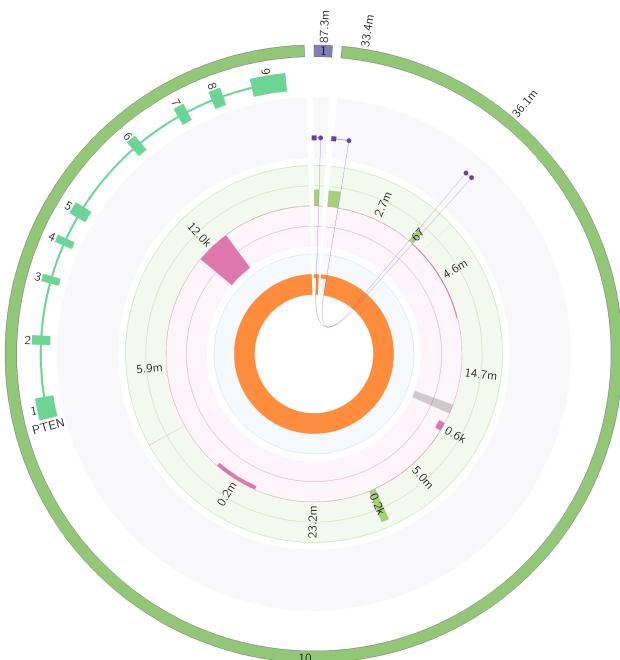
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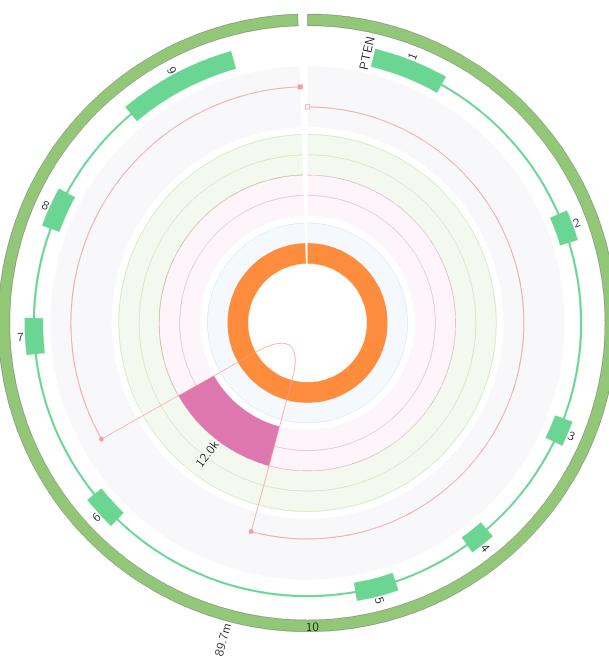
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CHR 1 ▶ CHR 10



CHR 10 ▶ CHR 10



CHR 9 ▶ CHR 10



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Germline Findings

Driver variants (0)

NONE

Other potentially relevant variants (3)

VARIANT	VCN	CN	MACN	RNA VAF	BIALLELIC	HOTSPOT	GENOTYPE
CYP2D6 c.506-1G>A	1.8	4.0	2.0	NA	No	Yes	HET
CYP3A4 c.-392G>A	4.0	4.0	2.0	NA	Yes	Yes	HOM
CYP3A4 c.522-191C>T	2.5	4.0	2.0	NA	No	Yes	HET

Genes with missed variant likelihood > 1% (22)

GENE	MVLH	GENE	MVLH	GENE	MVLH	GENE	MVLH
AMER1	3.8%	AR	2.6%	ARAF	2.0%	ATP2B3	2.0%
ATRX	3.1%	BCOR	2.2%	BTK	3.1%	CASZ1	3.7%
DDX3X	2.7%	EIF1AX	1.9%	GATA1	1.9%	KDM5C	1.7%
KDM6A	3.3%	KRTAP5-5	1.2%	MED12	3.1%	PHF6	3.9%
RBM10	2.1%	RPS6KA3	3.7%	STAG2	2.5%	ZFX	1.5%
ZMYM3	3.0%	ZRSR2	5.1%				

Germline CN Aberrations (0)

NONE

Pharmacogenetics (1)

GENE	GENOTYPE	FUNCTION	LINKED DRUGS	SOURCE
DPYD	*1_HOM	Normal Function	5-Fluorouracil;Capecitabine;Tegafur	PHARMGKB



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Immunology

Coming soon: HLA types for patient + status in tumor

Coming soon: List of neo-epitopes with predicted binding affinity

Coming soon: Details about RNA tumor micro-environment



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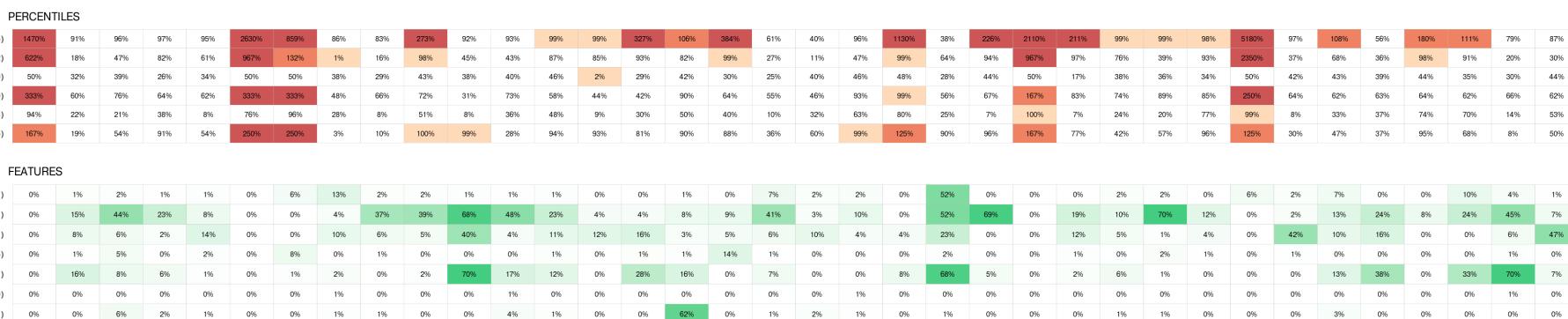
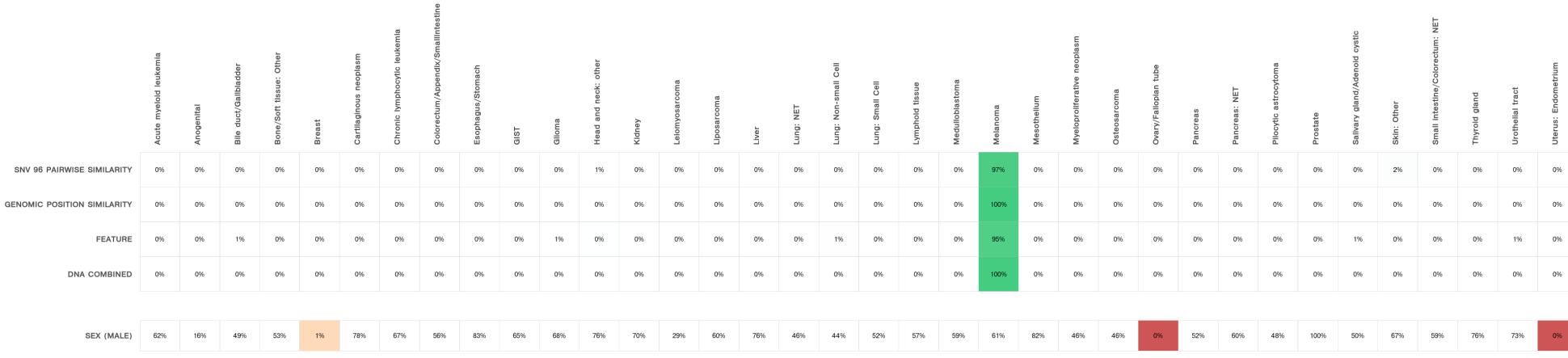
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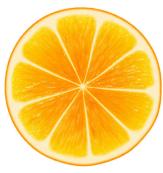
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Cohort Comparison





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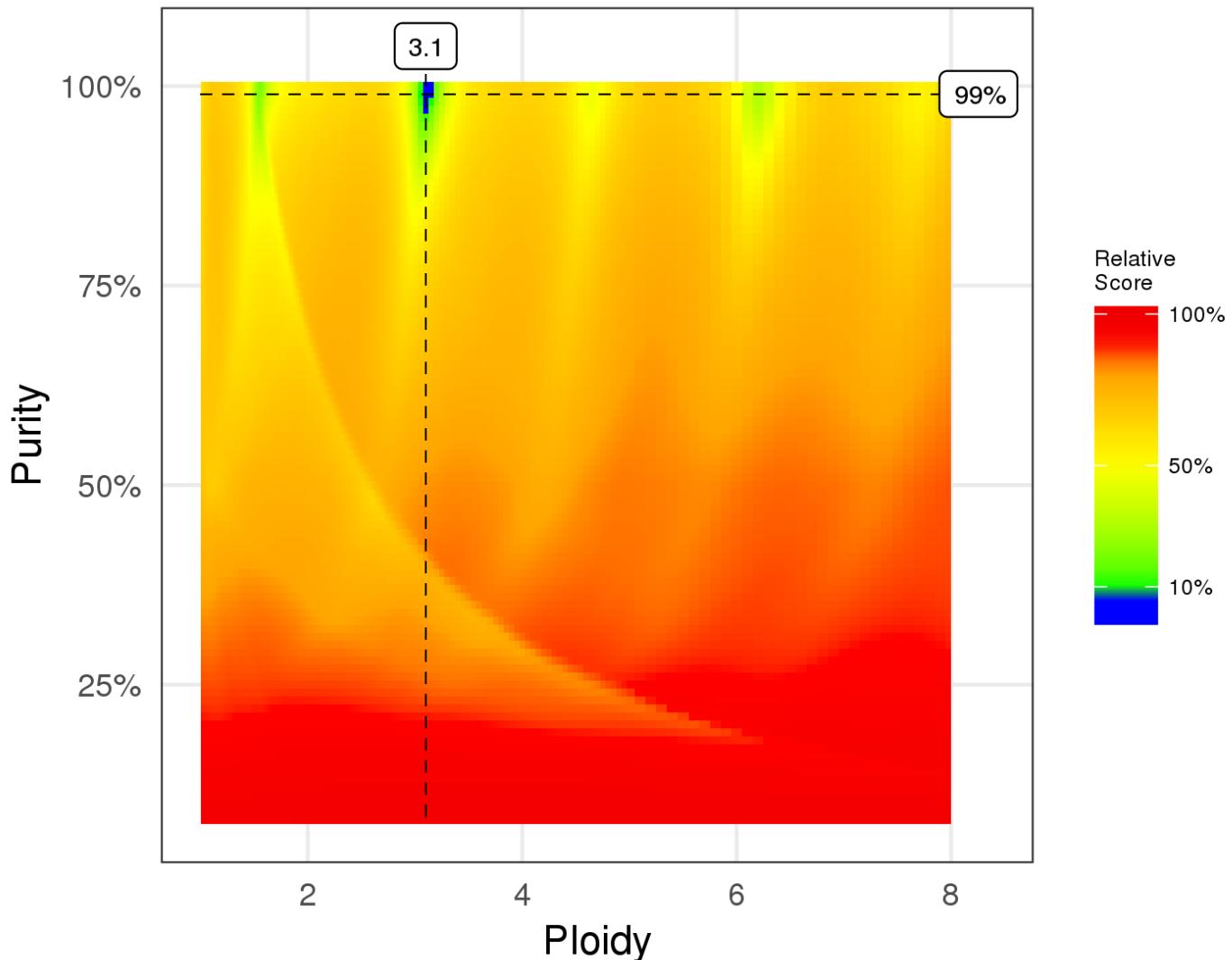
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Quality Control

QC	FIT METHOD	MEAN DEPTH	CONTAMINATION	UNS. CN SEGMENTS	DELETED GENES
PASS	NORMAL	111	0%	0	2

Purity/Ploidy Scores



Coverage Stats

	MEAN COVERAGE	SD COVERAGE	MEDIAN COVERAGE	MAD COVERAGE
Ref Sample	31.4	10.1	32	5
Tumor Sample	108.1	35.0	109	24



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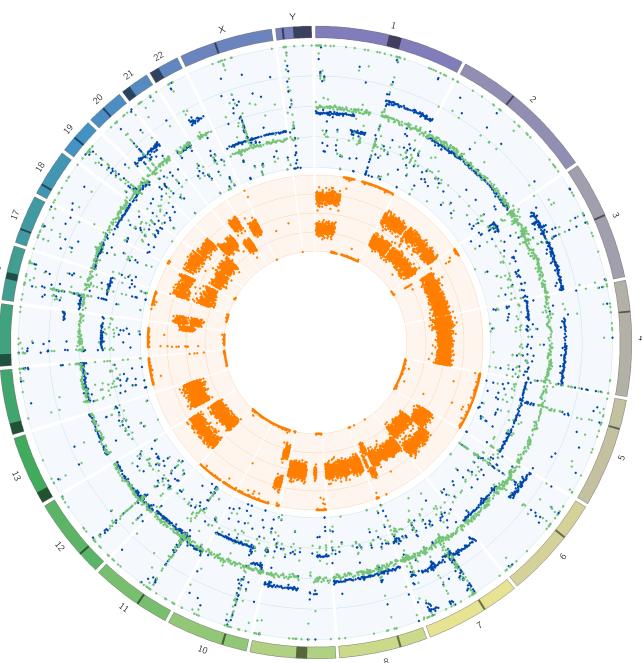
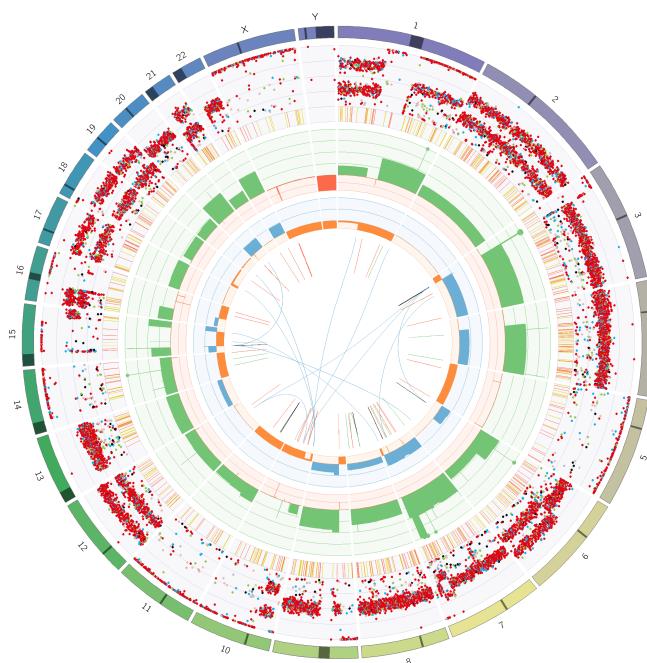
Excluded Percentages

	ADAPTER	BASEQ	CAPPED	DUPE	MAPQ	OVERLAP	UNPAIRED	TOTAL
Ref Sample	0%	0%	1%	11%	5%	1%	0%	18%
Tumor Sample	0%	0%	1%	15%	5%	1%	0%	22%

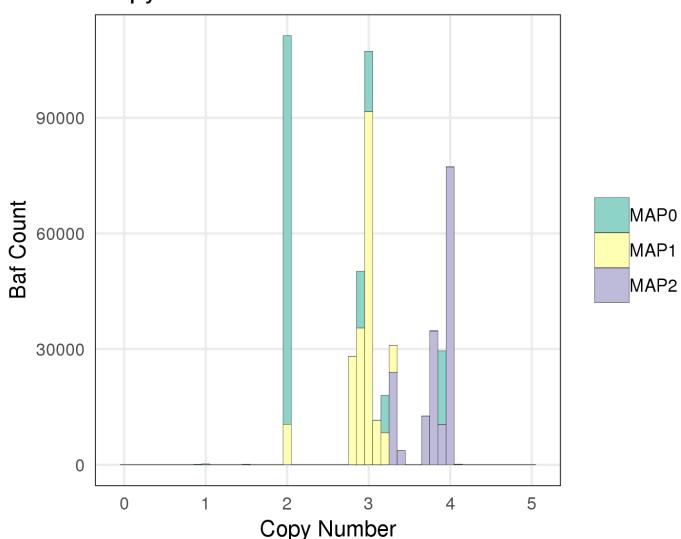
Flagstats

	UNIQUE RC	SECONDARY RC	SUPPLEMENTARY RC	MAPPED PROPORTION
Ref Sample	740406212	0	5742696	99%
Tumor Sample	2671674230	0	21477075	100%

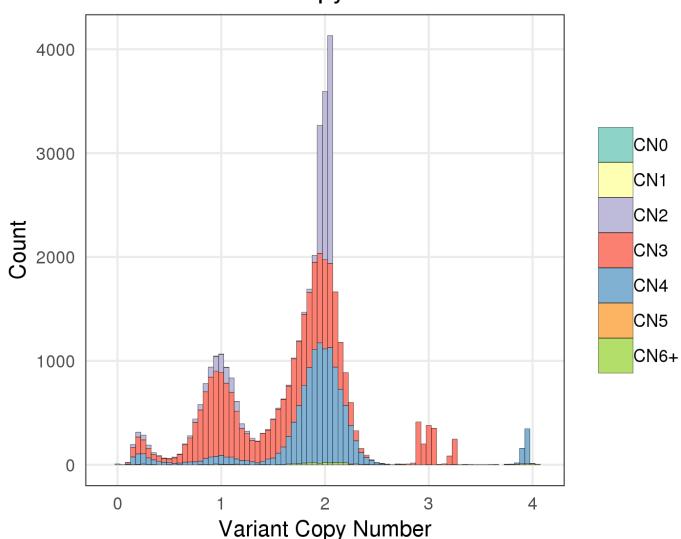
QC plots



Copy Number PDF



Somatic Variant Copy Number PDF





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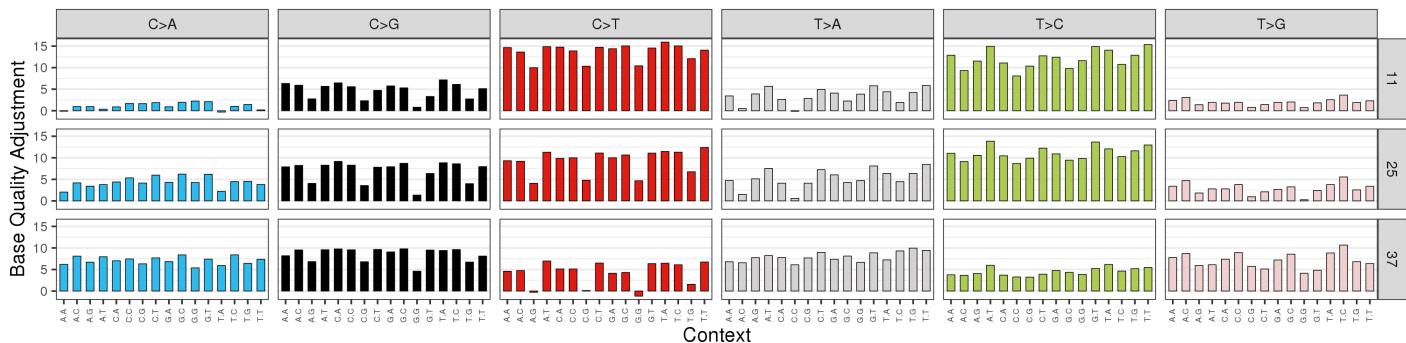
SAMPLE

Test

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Reference Sample BQR plot



Tumor Sample BQR plot

