



ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.33

CONFIGURED PRIMARY TUMOR

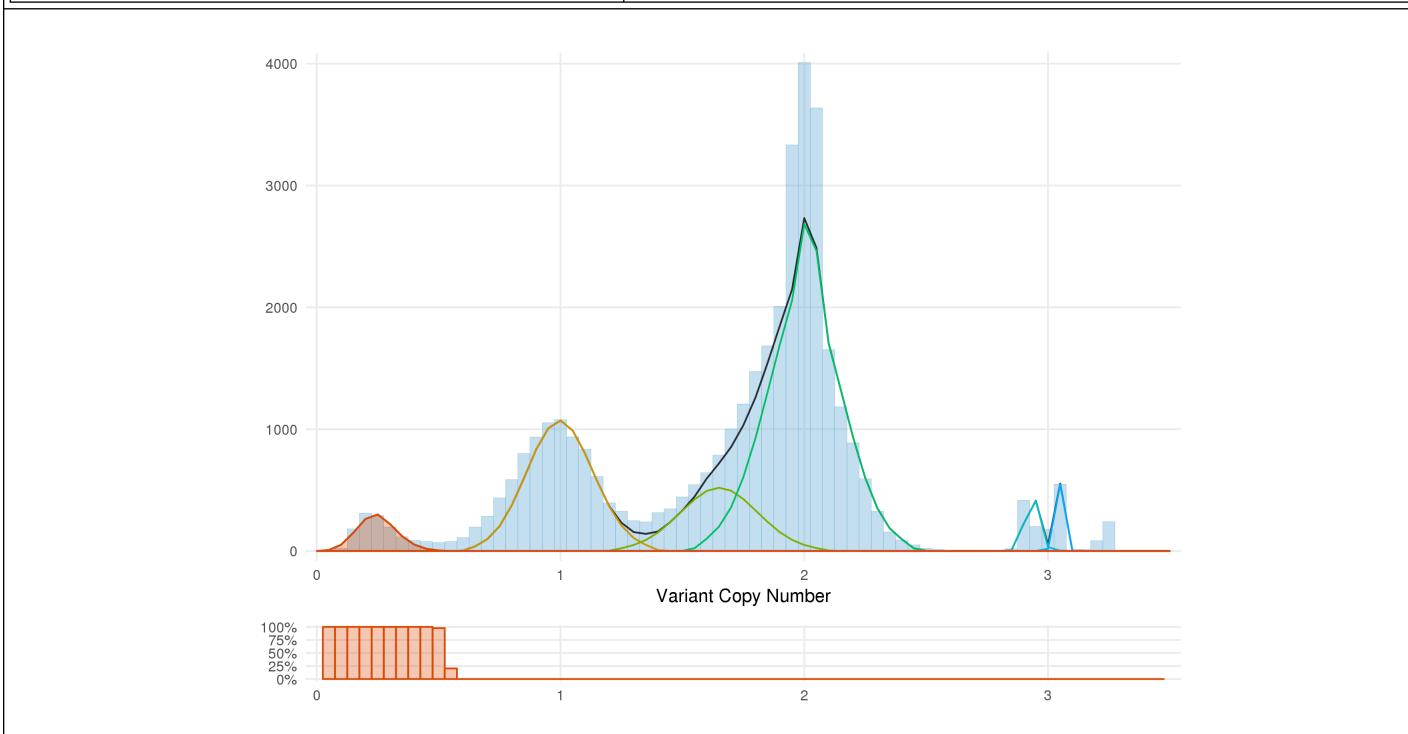
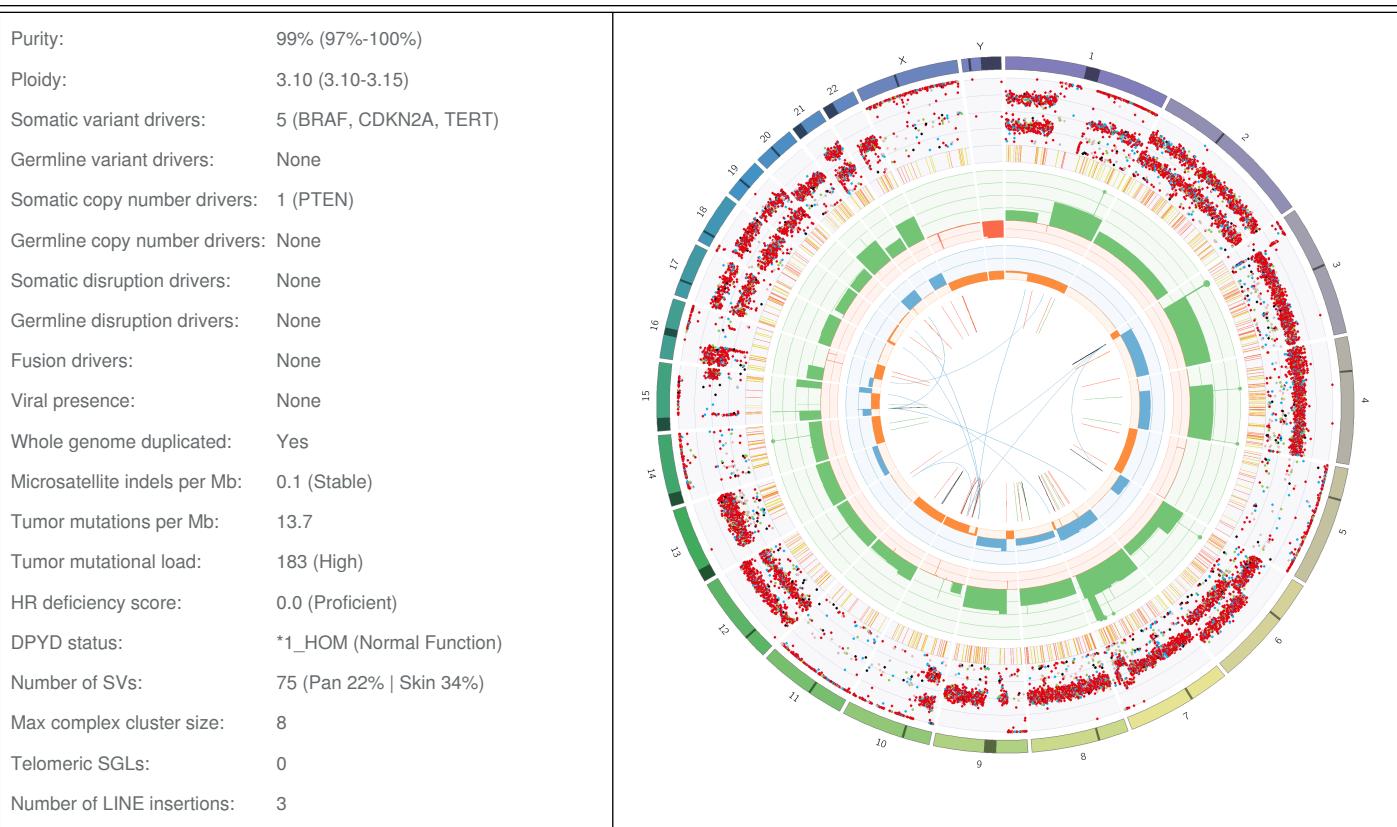
skin melanoma (DOID 8923)

CUPPA CANCER TYPE

Melanoma (100%)

QC

PASS





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Somatic Findings

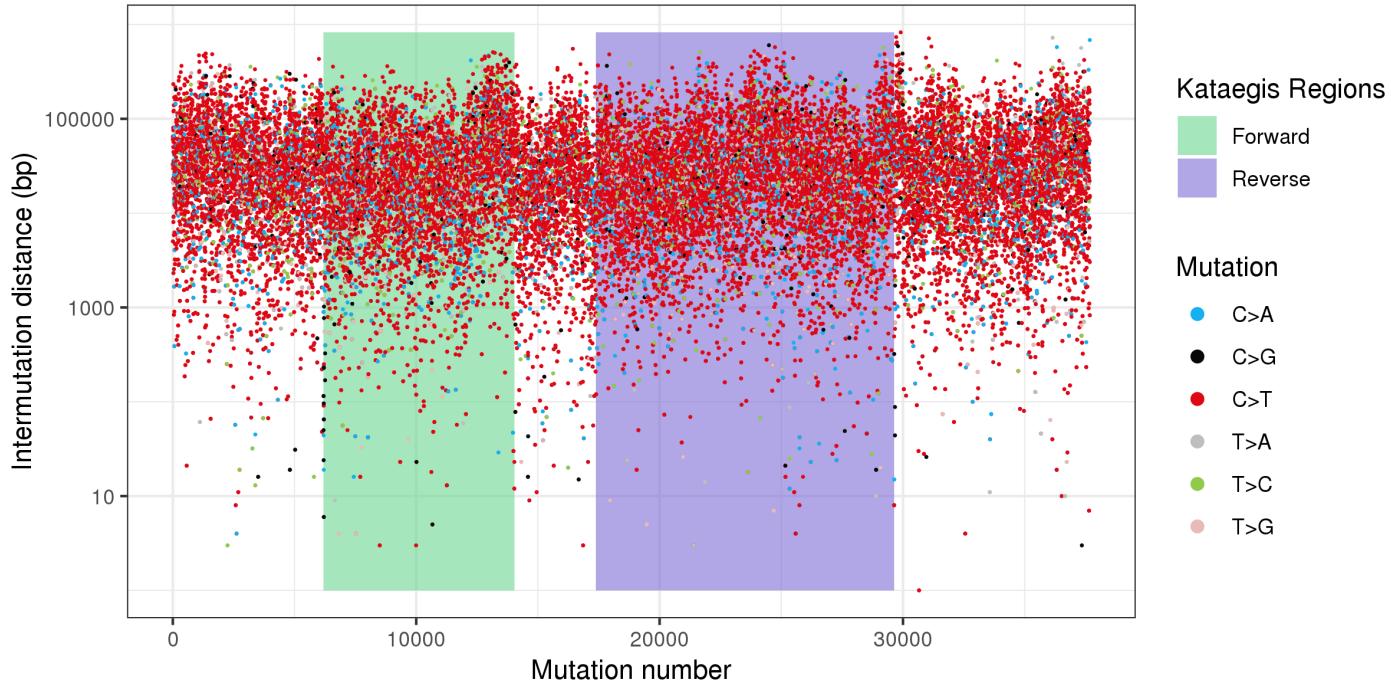
Driver variants (6)

VARIANT	VCN	CN	MACN	BIALLELIC	HOTSPOT	DL	CL	PHASE ID	RNA DEPTH
BRAF p.V600E	4.1	6.0	2.0	No	Yes	100%	100%		NA
CDKN2A p.A68fs	2.0	2.0	0.0	Yes	Near	100%	100%		NA
CDKN2A (alt) p.G83fs	2.0	2.0	0.0	Yes	Near	100%	100%		NA
TERT c.-125_-124delCCinsTT	1.7	2.0	0.0	Yes	Yes	100%	100%	4621	NA
SF3B1 p.P718L	2.0	3.0	1.0	No	No	15%	100%		NA
TP63 p.M499I	1.7	4.0	2.0	No	No	0%	100%		NA

Other potentially relevant variants (2)

VARIANT	VCN	CN	MACN	BIALLELIC	HOTSPOT	DL	CL	PHASE ID	RNA DEPTH
ALB c.1059-53dupT	1.8	3.9	1.9	No	No		100%		NA
STK19 p.D89N	2.0	3.8	1.8	No	Yes		100%		NA

Kataegis plot



Driver amps/dels (1)

LOCATION	GENE	TYPE	CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
10q23.31	PTEN	partial loss	0.0	NA	NA	NA	NA	NA

Potentially interesting near-driver amps (0)

NONE



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Other regions with amps (1)

LOCATION	GENE	TYPE	CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
3p24.2	CFL1P7	full gain	12.1	NA	NA	NA	NA	NA

Regions with deletions in genes in other autosomal regions (1)

LOCATION	GENE	TYPE	CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
16q21	CNOT1	partial loss	0.0	NA	NA	NA	NA	NA

Driver fusions (0)

NONE

Other potentially interesting fusions (0)

NONE

Driver viruses (0)

NONE

Other viral presence (0)

NONE

Homozygous disruptions (0)

NONE

Driver gene disruptions (2)

LOCATION	GENE	RANGE	TYPE	CLUSTER ID	JUNCTION CN	UNDISRUPTED CN
10q23.31	PTEN	Intron 5 Upstream	DEL	68	2.0	0.0
10q23.31	PTEN	Intron 6 Downstream	DEL	68	2.0	0.0

Other potentially interesting gene disruptions (0)

NONE

Potentially interesting LOH events in case of MSI or HRD (0)

NONE

Signature allocations (12)

SIGNATURE	ALLOCATION	PERCENT
Sig2	3697.8	10%

THE TABLE CONTINUES ON THE NEXT PAGE



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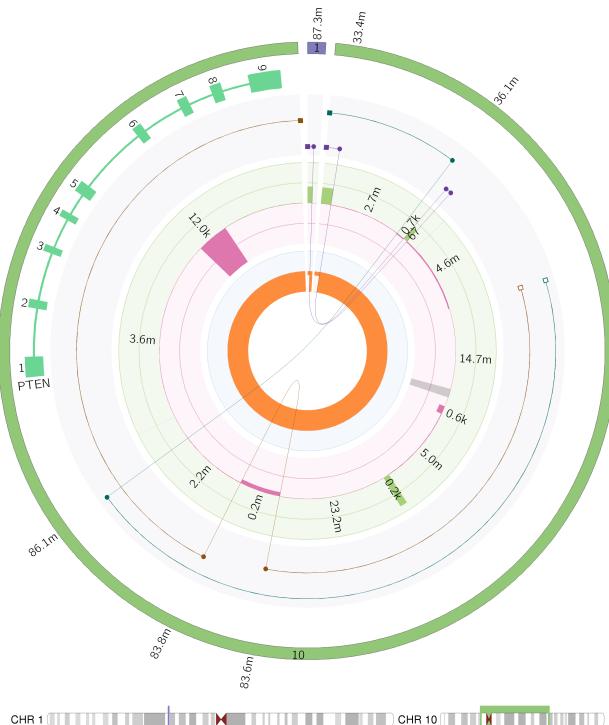
5.33

Signature allocations (12)

CONTINUED FROM THE PREVIOUS PAGE

SIGNATURE	ALLOCATION	PERCENT
Sig4	2057.0	5%
Sig7	23307.0	62%
Sig8	1969.7	5%
Sig11	1313.5	3%
Sig12	1136.2	3%
Sig17	1082.8	3%
Sig18	2066.8	5%
Sig20	248.6	1%
Sig24	287.6	1%
Sig28	514.0	1%
MISALLOC	4616.2	12%

Structural driver plots (3)





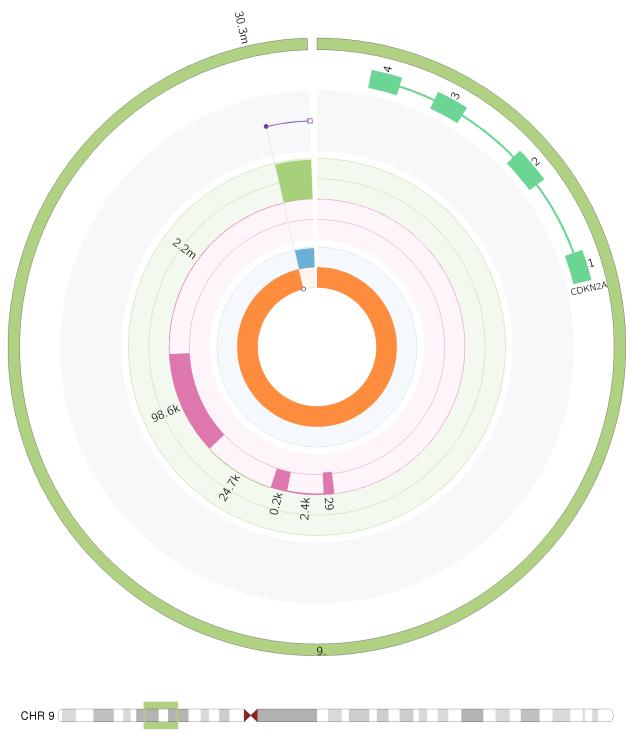
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Germline Findings

Driver variants (0)

NONE

Other potentially relevant variants (3)

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

Potentially pathogenic germline deletions (0)

NONE

Potentially pathogenic germline homozygous disruptions (0)

NONE

Potentially pathogenic germline gene disruptions (0)

NONE

Genes with missed variant likelihood > 1% (0)

NONE

Germline CN aberrations (0)

NONE

Pharmacogenetics (2)

GENE	GENOTYPE	FUNCTION	LINKED DRUGS	SOURCE
DPYD	*1_HOM	Normal Function	5-Fluorouracil;Capecitabine;Tegafur	PHARMGKB
UGT1A1	*1_HOM	Normal Function	Irinotecan	PHARMGKB



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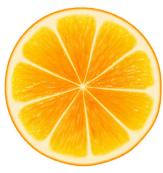
Immunology

HLA QC

QC Status: PASS

HLA Alleles (6)

ALLELE	REF FRAGS	TUMOR FRAGS	RNA FRAGS	TUMOR CN	SOMATIC #MUTATIONS
A*01:01	210	1602	0	2.0	None
A*01:01	211	1602	0	1.8	None
B*08:01	216	750	0	1.8	None
B*40:02	196	721	0	2.0	None
C*03:04	213	768	0	2.0	None
C*07:01	225	767	0	1.8	None



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RNA Findings

QC	TOTAL FRAGMENTS	NON-DUPLICATE FRAGMENTS	DUPLICATE RATE
NA			
<hr/>			
Genes with high expression (0)			
NONE			
<hr/>			
Genes with low expression (0)			
NONE			
<hr/>			
Known fusions detected in RNA and not in DNA (0)			
NONE			
<hr/>			
Promiscuous fusions detected in RNA and not in DNA (0)			
NONE			
<hr/>			
Potentially interesting novel splice junctions - Skipped exons (0)			
NONE			
<hr/>			
Potentially interesting novel splice junctions - Novel exon/intron (0)			
NONE			
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Cohort Comparison

	Acute myeloid leukemia	Angiomyxoma	Bile duct/CaBladder	Bone/Soft tissue: Other	Breast	Cardiogenic neoplasm	Colon/Appendix/SmallIntestine	Esophagus/Stomach	GIST	Glioma	Head and neck: other	Kidney	Kidney-ChRCC	Lemomysarcoma	Liposarcoma	Liver	Lung: NET	Lung: Non-small Cell	Lung: Small Cell	Lymphoid tissue	Medulloblastoma	Melanoma	Mesothelioma	Myeloproliferative neoplasm	Osteosarcoma	Ovary/European tube	Pancreas	Pancreas: NET	Pleocystic astrocytoma	Prostate	Salivary gland/Adenoid cystic	Skin: Other	Small intestine/Colon/rectum: NET	Thyroid gland	Urothelial tract	Uterus: Endometrium
SNV 96 PAIRWISE	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%		
GENOMIC POSITION COHORT	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%		
FEATURE	0%	0%	0%	0%	0%	0%	0%	0%	0%	1%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%		
DNA COMBINED	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%		
SEX (MALE)	58%	12%	47%	53%	1%	78%	56%	79%	65%	66%	79%	70%	56%	31%	69%	72%	50%	46%	53%	61%	59%	62%	85%	50%	46%	56%	61%	48%	100%	53%	62%	61%	43%	72%	0%	

SNV SIGNATURES

SIG 7 UV (24216)	32300%	1920%	3910%	97%	1240%	17700%	1680%	2390%	2300%	47300%	1770%	2950%	15800%	9880%	14000%	5740%	11400%	787%	1580%	2330%	39600%	33%	7910%	4040000%	8040%	4260%	3520%	9170%	202000%	9710%	98%	66%	14200%	22400%	740%	6100%			
SIG 6 MMR (0)	50%	20%	36%	43%	38%	44%	32%	39%	40%	50%	32%	43%	48%	42%	48%	43%	35%	36%	48%	42%	44%	46%	38%	44%	45%	28%	42%	38%	44%	39%	44%	48%	32%	36%	39%	30%			
SIG 4 SMOKING (2127)	61%	100%	107%	592%	122%	10000%	99%	100%	1440%	10000%	92%	123%	88600%	99%	19000%	95%	98%	93%	39%	71%	10000%	5930%	99%	166%	10000%	225%	101%	99%	100%	10000%	154%	98%	99%	10000%	320%	95%	376%		
SIG 2 13 AID APOBEC (3842)	20000%	56%	96%	97%	78%	22900%	99%	94%	334%	99%	62%	99%	712%	243%	301%	60%	93%	100%	3280%	34%	366%	15800%	98%	97%	100%	98%	42200%	100%	93%	93%	57%	81%	98%	32%	96%	81%	98%	32%	96%
SIG 17 (1121)	1300%	98%	95%	98%	94%	21600%	58%	27%	98%	10000%	97%	100%	98%	205%	366%	100%	10000%	94%	97%	89%	10500%	98%	1540%	40000%	94%	95%	94%	94%	100%	98%	97%	1440%	354%	92%	99%				
SIG 11 (1365)	2440%	100%	100%	96%	102%	5480%	99%	100%	555%	95%	99%	344%	1270%	319%	396%	248%	397%	99%	131%	100%	937%	87%	442%	1380%	234%	123%	164%	98%	15200%	295%	97%	58%	51%	222%	98%	202%			
SIG 10 POLE (0)	45%	23%	24%	38%	26%	50%	15%	24%	32%	45%	18%	32%	45%	42%	48%	36%	44%	38%	47%	36%	24%	48%	40%	44%	38%	44%	27%	40%	36%	35%	27%	42%	49%	36%	25%	36%			
SIG 1 (0)	0%	-10000%	3%	3%	2%	10000%	4%	8%	2%	0%	3%	6%	10000%	0%	10000%	23%	6%	26%	44%	4%	-10000%	10000%	6%	1%	0%	4%	0%	0%	2%	22%	10000%	2%	4%	6%					

PERCENTILES

SNV COUNT (37681)	1470%	93%	95%	96%	95%	26300%	84%	80%	273%	95%	90%	99%	479%	100%	324%	99%	384%	61%	37%	98%	1130%	37%	222%	2110%	210%	99%	99%	99%	5180%	96%	97%	60%	561%	110%	81%	84%
MS INDELS TMB (0,13)	679%	35%	46%	86%	67%	1060%	2%	11%	101%	74%	52%	91%	241%	95%	97%	64%	196%	33%	15%	68%	100%	98%	81%	41%	94%	2560%	39%	69%	42%	122%	97%	32%	52%			
TELOMERIC SGL (0)	50%	36%	38%	36%	36%	50%	39%	39%	44%	42%	39%	46%	42%	41%	26%	42%	33%	27%	41%	47%	48%	28%	44%	50%	20%	38%	35%	50%	42%	39%	38%	47%	44%	34%	48%	
SIMPLE DUP 2 B 200B (3)	150%	80%	62%	62%	64%	300%	44%	49%	76%	39%	74%	26%	75%	44%	38%	62%	76%	41%	40%	91%	95%	38%	80%	100%	65%	76%	63%	99%	47%	59%	54%	77%	72%	69%	81%	
MAX COMPLEX SIZE (8)	93%	24%	14%	40%	9%	67%	32%	8%	51%	16%	26%	56%	90%	14%	18%	48%	38%	10%	29%	76%	80%	24%	9%	98%	8%	22%	20%	73%	98%	7%	36%	38%	77%	83%	16%	43%
LINE (3)	91%	13%	34%	74%	30%	150%	2%	6%	84%	74%	17%	76%	74%	77%	46%	62%	87%	24%	35%	92%	93%	72%	86%	88%	65%	29%	24%	70%	98%	13%	23%	24%	87%	78%	7%	20%

FEATURES

BRaf (1)	0%	1%	2%	0%	1%	0%	13%	2%	2%	1%	1%	1%	0%	0%	0%	1%	0%	6%	2%	4%	0%	51%	0%	0%	2%	2%	6%	0%	0%	19%	3%	1%				
CDKN2A (1)	0%	16%	52%	28%	8%	0%	4%	38%	41%	59%	49%	20%	0%	11%	7%	8%	7%	41%	6%	6%	0%	53%	70%	0%	20%	9%	74%	12%	23%	8%	10%	46%	2%			
PTEN (1)	0%	12%	5%	3%	14%	0%	11%	8%	5%	35%	4%	9%	5%	14%	10%	4%	4%	7%	10%	2%	4%	22%	0%	0%	14%	6%	2%	4%	0%	42%	9%	15%	0%	5%	45%	
SF3B1 (0,15)	0%	1%	4%	0%	2%	0%	0%	1%	0%	0%	0%	0%	0%	0%	3%	1%	14%	1%	0%	3%	0%	2%	0%	0%	1%	0%	3%	1%	0%	1%	0%	0%	0%	0%		
TERT (1)	0%	14%	2%	5%	0%	0%	2%	0%	2%	14%	9%	2%	0%	0%	12%	15%	0%	0%	2%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%		
TP53 (0,00)	0%	0%	0%	0%	0%	0%	1%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	
INDEL_ALB (1)	0%	0%	5%	2%	0%	0%	1%	0%	0%	0%	1%	1%	0%	0%	0%	0%	0%	62%	0%	1%	2%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%



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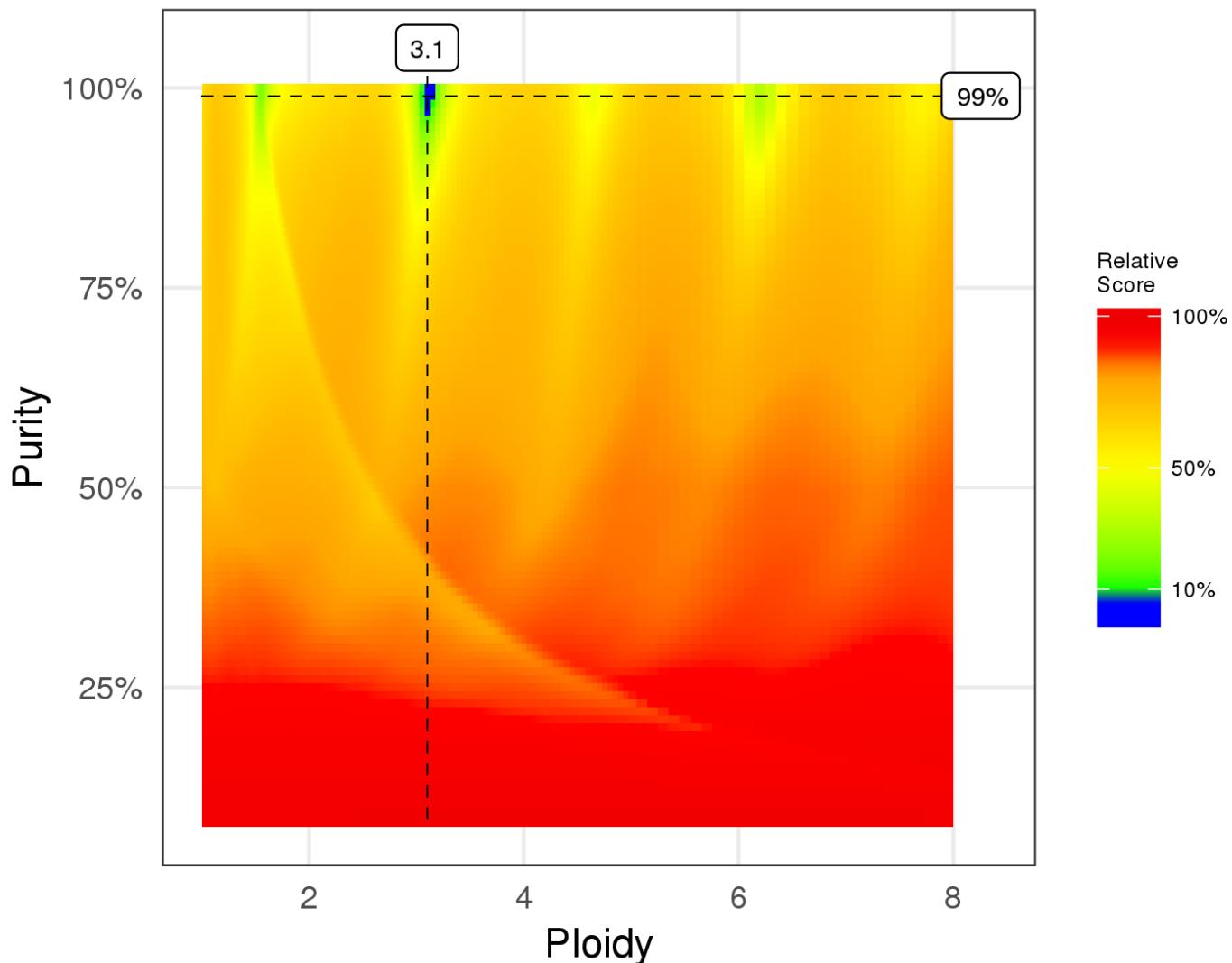
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Quality Control

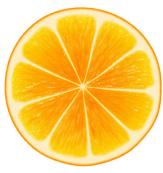
QC	REF GENOME	FIT METHOD	MEAN DEPTH	CONTAMINATION	UNS. SEGMENTS	DELETED GENES
PASS	V37	NORMAL	111	0%	0	4

Purity/Ploidy Scores



Flagstats

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



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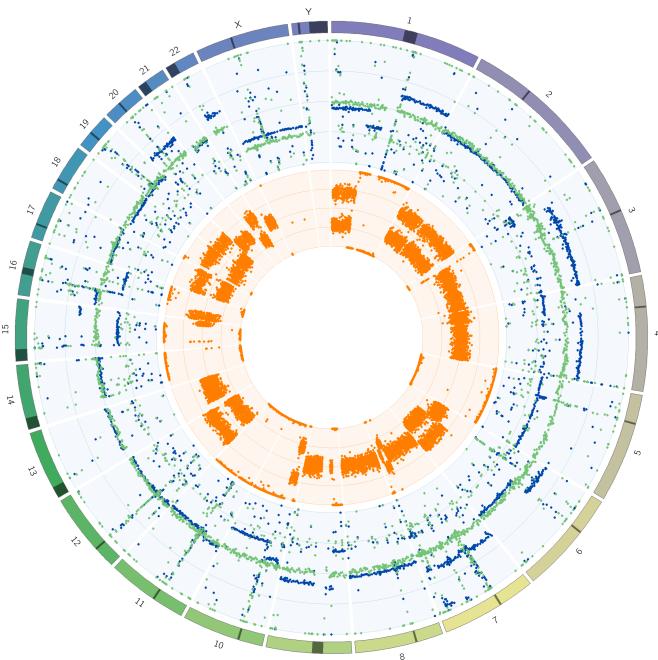
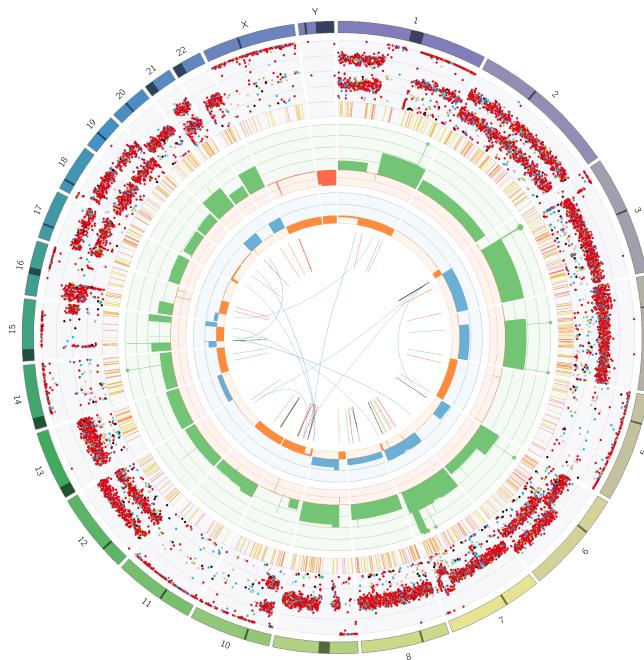
Coverage Stats

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

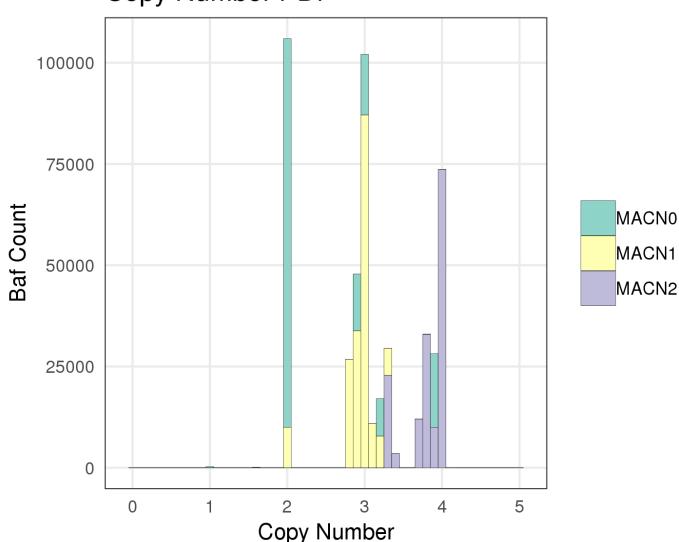
Excluded Percentages

	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%

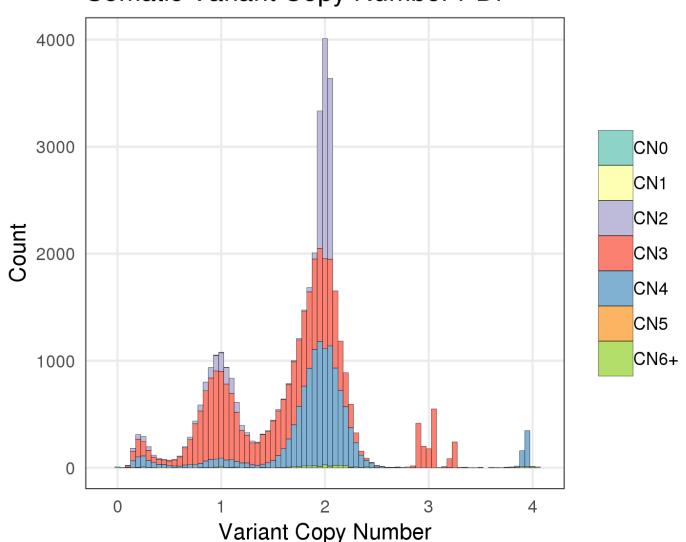
QC plots



Copy Number PDF



Somatic Variant Copy Number PDF





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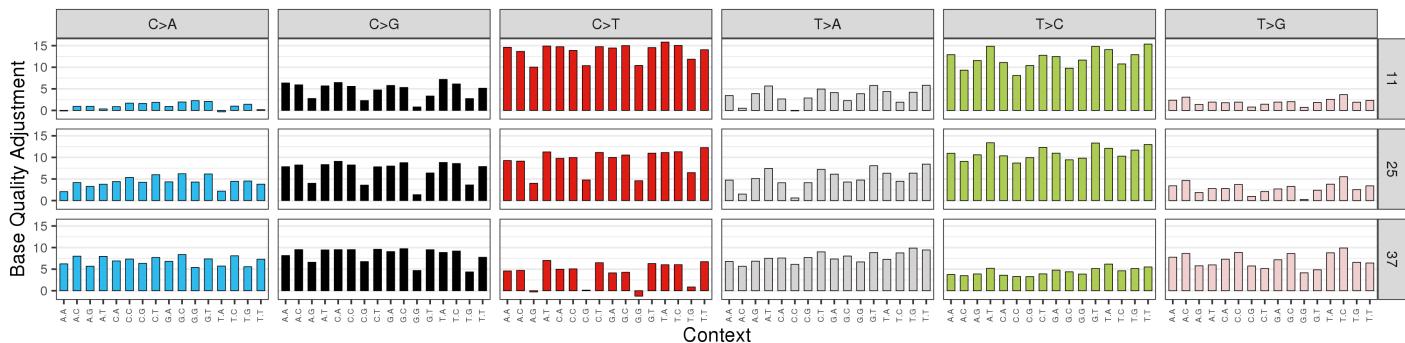
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Reference Sample BQR plot



Tumor Sample BQR plot

