



# ORANGE Report

SAMPLE

Test

PLATINUM VERSION

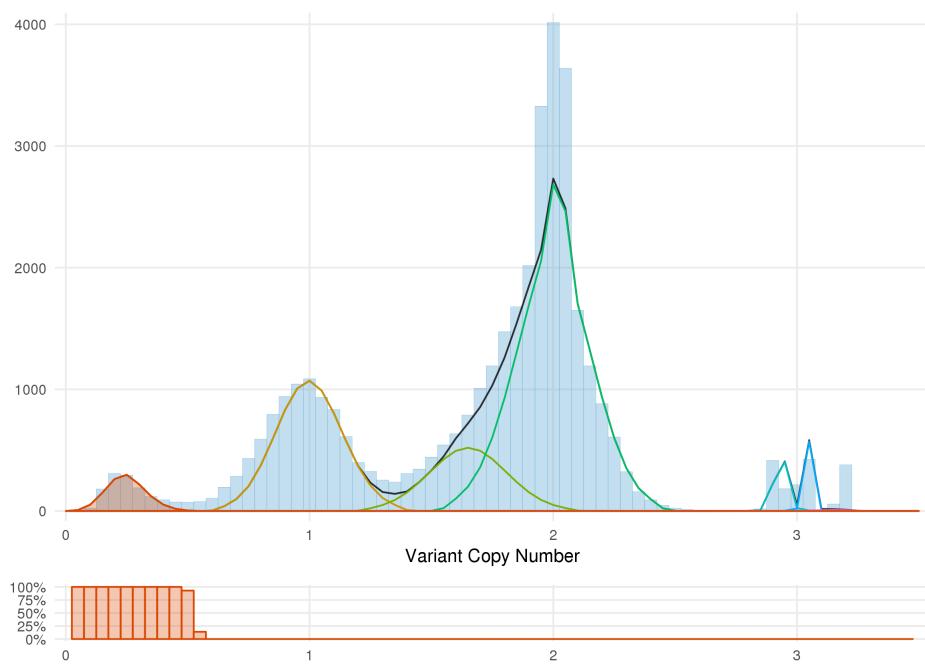
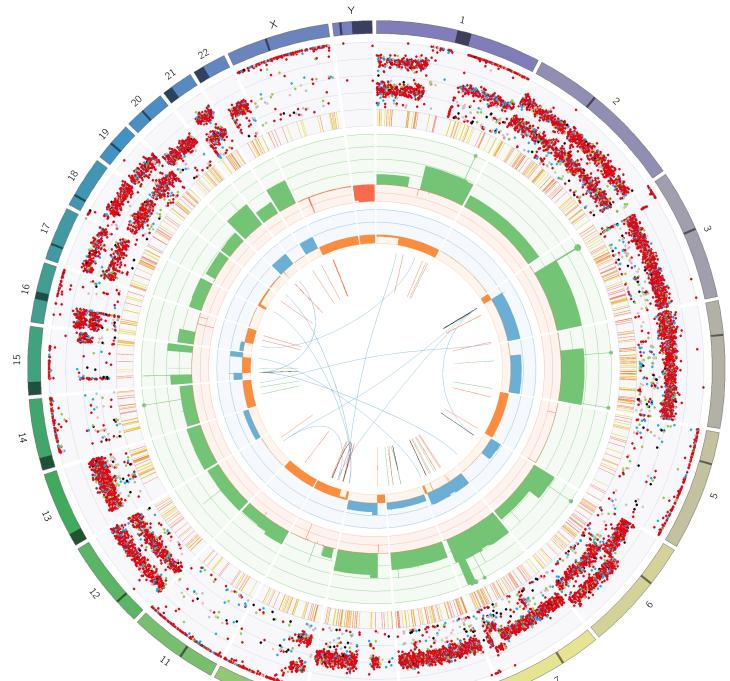
5.32

CONFIGURED PRIMARY TUMOR  
skin melanoma (DOID 8923)

CUPPA CANCER TYPE  
Melanoma (100%)

QC  
PASS

Purity:	99% (97%-100%)
Ploidy:	3.1 (3.1-3.15)
Somatic variant drivers:	5 (BRAF, CDKN2A, TERT)
Germline variant drivers:	None
Somatic copy number drivers:	1 (PTEN)
Germline copy number drivers:	None
Somatic disruption drivers:	None
Germline disruption drivers:	None
Fusion drivers:	None
Viral presence:	None
Whole genome duplicated:	Yes
Microsatellite indels per Mb:	0.1 (Stable)
Tumor mutations per Mb:	13.7
Tumor mutational load:	186 (High)
HR deficiency score:	0 (Proficient)
DPYD status:	*1_HOM (Normal Function)
Number of SVs:	73 (Pan 22%   Skin 34%)
Max complex cluster size:	8
Telomeric SGLs:	0
Number of LINE insertions:	3





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## Somatic Findings

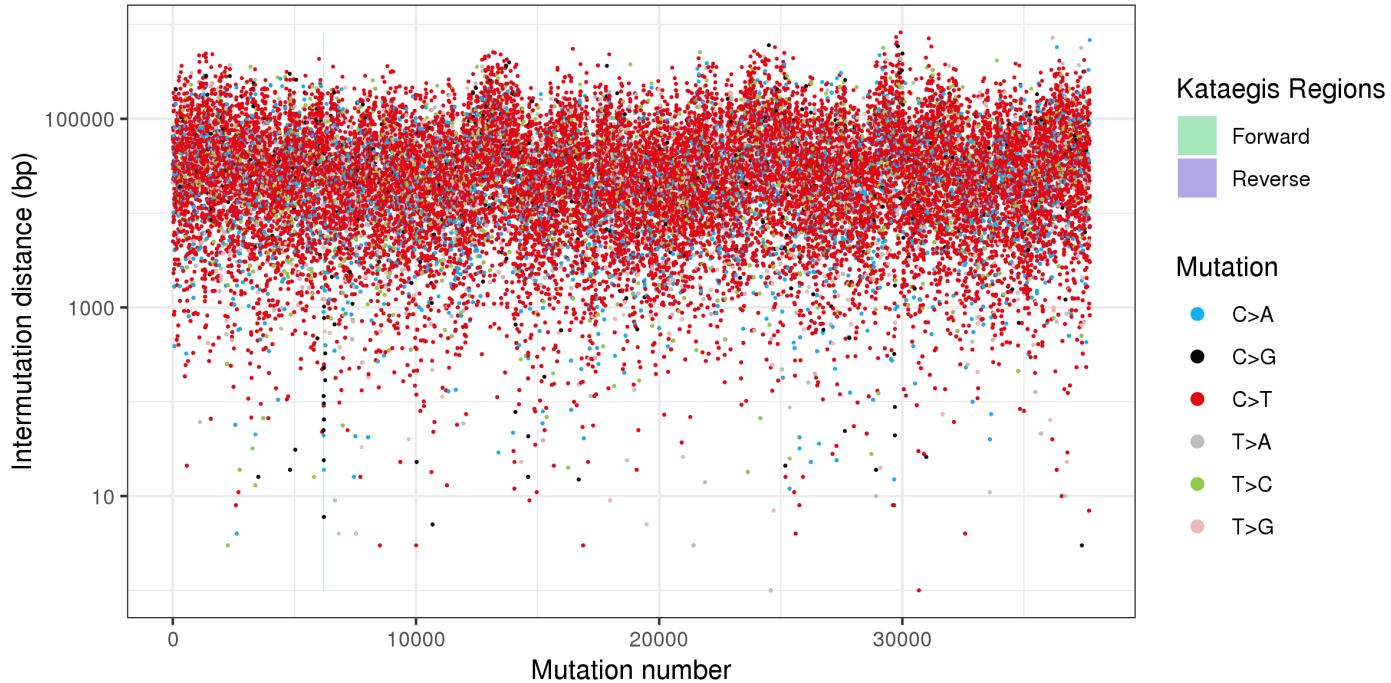
### Driver variants (6)

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%	4613	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	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	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	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	71	2.0	0.0
10q23.31	PTEN	Intron 6 Downstream	DEL	71	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	3700.7	10%

THE TABLE CONTINUES ON THE NEXT PAGE



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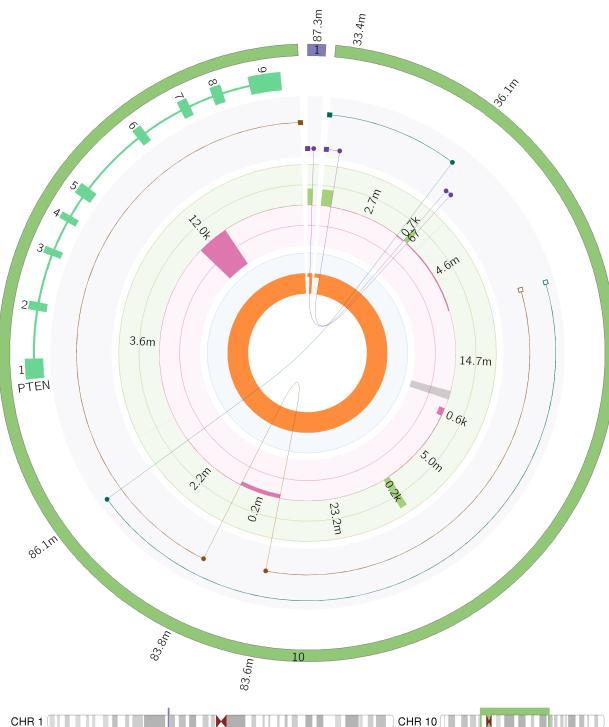
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## Signature allocations (12)

CONTINUED FROM THE PREVIOUS PAGE

SIGNATURE	ALLOCATION	PERCENT
Sig4	2059.7	5%
Sig7	23326.6	62%
Sig8	1976.7	5%
Sig11	1309.6	3%
Sig12	1137.1	3%
Sig17	1083.4	3%
Sig18	2066.2	5%
Sig20	254.8	1%
Sig24	285.9	1%
Sig28	512.3	1%
MISALLOC	4615.7	12%

## Structural driver plots (3)





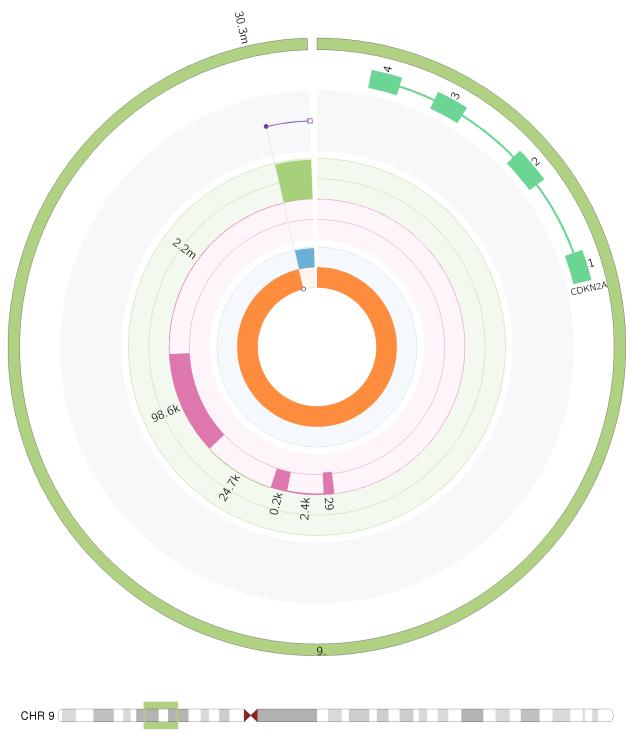
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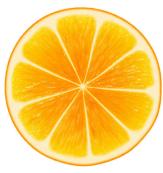
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## Germline Findings

### Driver variants (0)

NONE

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### 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

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### Potentially pathogenic germline homozygous disruptions (0)

NONE

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### Potentially pathogenic germline gene disruptions (0)

NONE

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### Genes with missed variant likelihood > 1% (0)

NONE

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## Germline CN aberrations (0)

NONE

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## 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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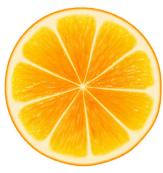
### Immunology

#### 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	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	None
C*03:04	213	768	0	2	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/>			
<b>Genes with high expression (0)</b>			
NONE			
<hr/>			
<b>Genes with low expression (0)</b>			
NONE			
<hr/>			
<b>Known fusions detected in RNA and not in DNA (0)</b>			
NONE			
<hr/>			
<b>Promiscuous fusions detected in RNA and not in DNA (0)</b>			
NONE			
<hr/>			
<b>Potentially interesting novel splice junctions - Skipped exons (0)</b>			
NONE			
<hr/>			
<b>Potentially interesting novel splice junctions - Novel exon/intron (0)</b>			
NONE			
<hr/>			



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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%	0%	56%	61%	48%	100%	53%	62%	61%	43%	72%	0%

SNV SIGNATURES																																										
SIG 7 UV (2423)	32300%	1920%	3910%	97%	1240%	17700%	1680%	2600%	2300%	47300%	1770%	2950%	15800%	9880%	14000%	5750%	11400%	787%	1580%	2340%	39600%	33%	7920%	4040000%	8050%	4260%	3520%	9180%	202000%	9720%	98%	66%	14200%	2250%	741%	6110%						
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 (2130)	619%	100%	107%	593%	122%	10000%	99%	100%	1440%	10000%	92%	123%	88700%	99%	19000%	95%	98%	93%	39%	7%	10000%	5930%	99%	166%	10000%	225%	101%	99%	100%	10000%	154%	98%	99%	10000%	320%	95%	376%					
SIG 2 13 AID APOBEC (3845)	20000%	56%	96%	97%	78%	22900%	99%	94%	334%	99%	62%	99%	712%	243%	301%	60%	93%	100%	3290%	34%	366%	15800%	98%	97%	100%	98%	42300%	100%	93%	93%	93%	57%	816%	98%	32%	96%	816%	354%	92%	99%		
SIG 17 (1121)	1300%	98%	95%	98%	94%	21600%	58%	2%	98%	10000%	97%	100%	98%	205%	366%	100%	10000%	94%	97%	89%	10500%	98%	1540%	40100%	94%	95%	94%	94%	676%	17000%	99%	94%	93%	1440%	354%	92%	99%	93%	517%	222%	98%	202%
SIG 11 (1361)	2430%	100%	100%	96%	102%	5460%	99%	10%	553%	95%	99%	343%	1270%	318%	395%	247%	395%	99%	131%	100%	934%	87%	441%	1380%	233%	123%	164%	98%	15100%	294%	97%	58%	517%	222%	98%	202%						
SIG 10 POLE (0)	45%	23%	24%	38%	26%	50%	15%	15%	24%	32%	45%	18%	32%	45%	42%	48%	36%	44%	38%	47%	36%	24%	48%	40%	44%	38%	44%	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%	22%	10000%	2%	4%	6%	6%								

PERCENTILES																																				
SNV COUNT (37713)	1470%	93%	95%	96%	95%	26300%	85%	60%	274%	95%	90%	99%	479%	100%	324%	99%	384%	61%	37%	98%	1130%	37%	222%	2120%	210%	99%	99%	5190%	96%	97%	60%	561%	110%	81%	84%	
MS INDELS TMB (0,12)	660%	34%	44%	85%	66%	10300%	2%	11%	100%	70%	50%	91%	234%	92%	96%	63%	191%	32%	12%	68%	100%	67%	95%	10300%	98%	80%	39%	94%	2490%	38%	69%	42%	118%	97%	30%	50%
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%	79%	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%	84%	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%	1%	0%	6%	2%	4%	0%	51%	0%	0%	2%	2%	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%	0%	3%	23%	8%	10%	46%	2%		
PTEN (1)	0%	12%	5%	3%	14%	0%	11%	6%	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%	0%	0%	
TERT (1)	0%	14%	2%	5%	0%	0%	2%	0%	2%	6%	14%	9%	2%	0%	12%	15%	0%	0%	2%	0%	0%	66%	5%	0%	2%	4%	0%	0%	0%	9%	38%	0%	28%	6%	1%		
TP53 (0,00)	0%	0%	0%	0%	0%	0%	1%	0%	0%	1%	2%	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%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%



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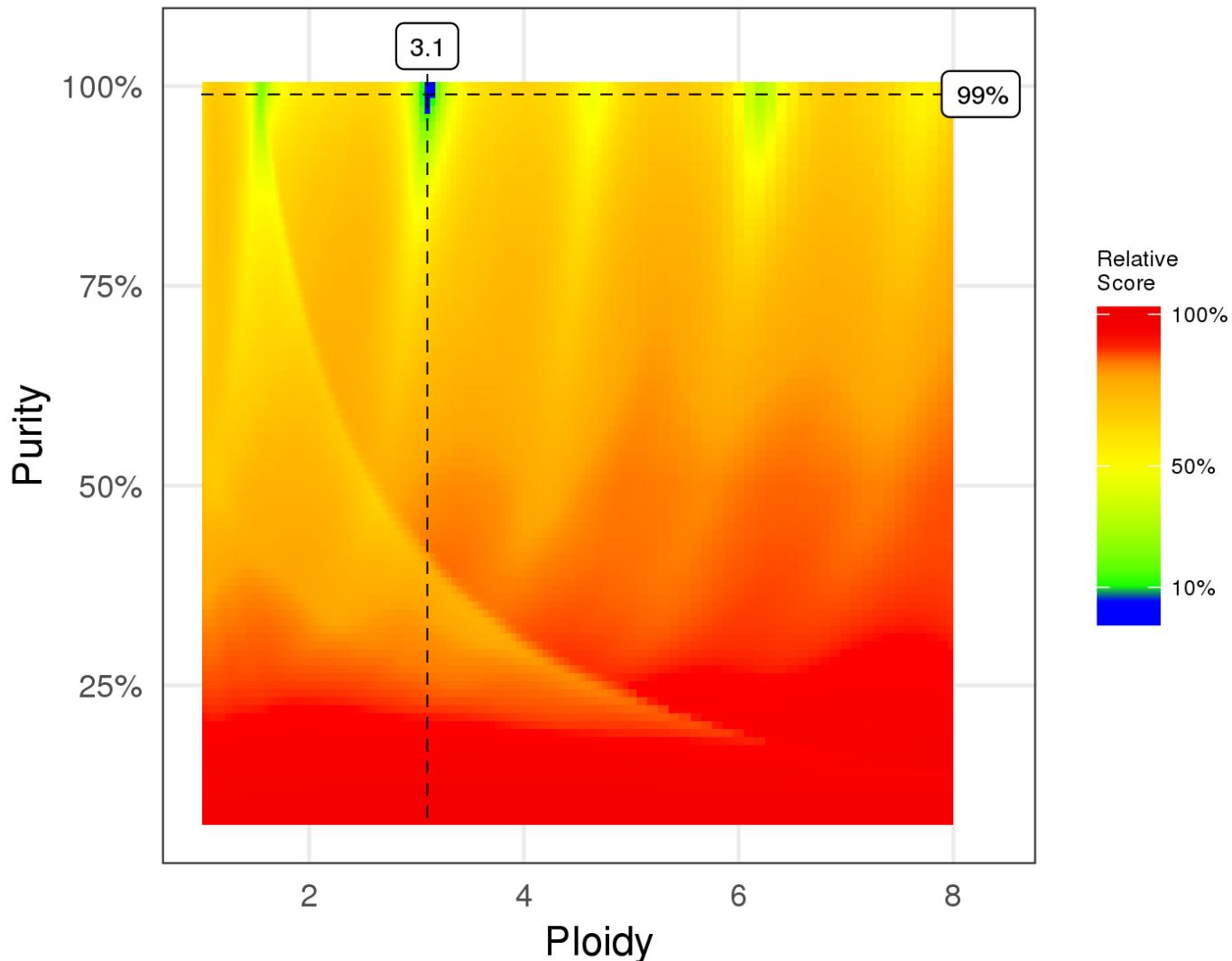
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### Quality Control

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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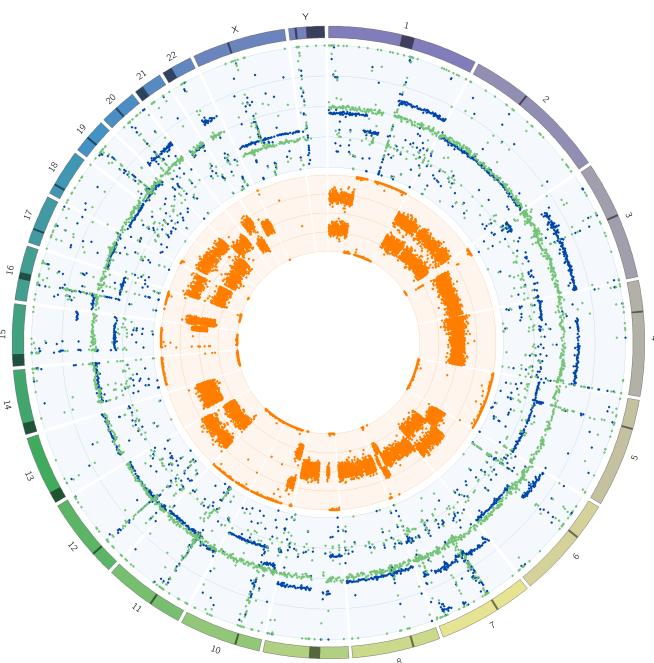
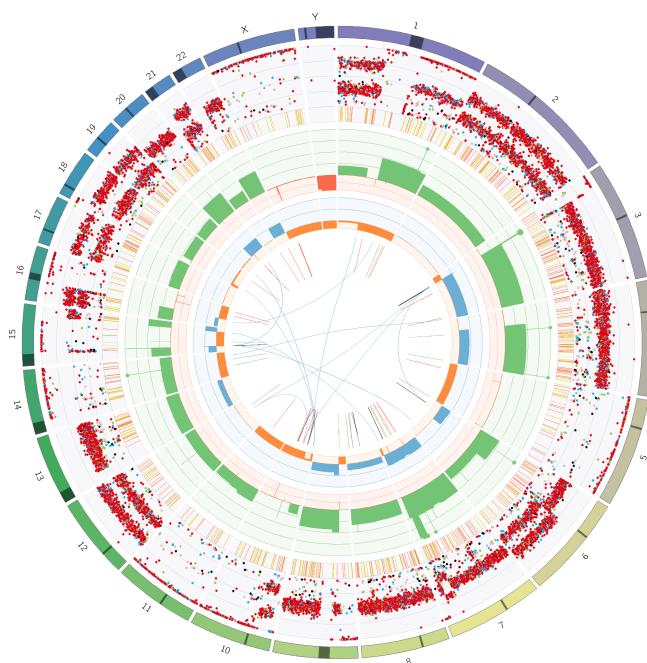
### Coverage Stats

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

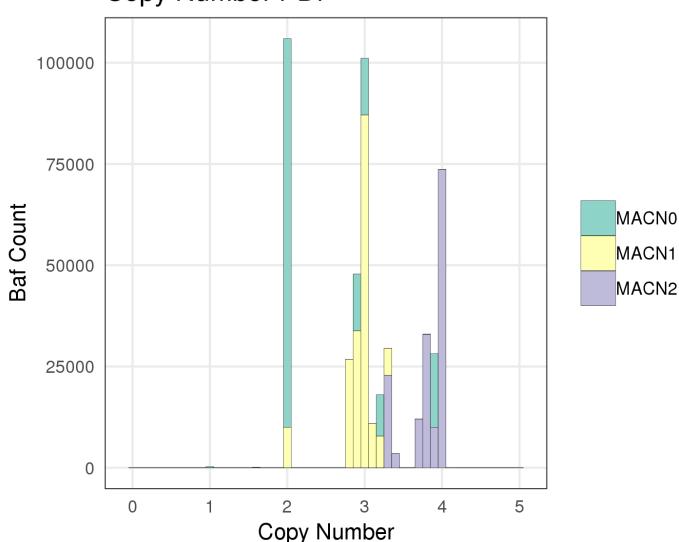
### Excluded Percentages

	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%

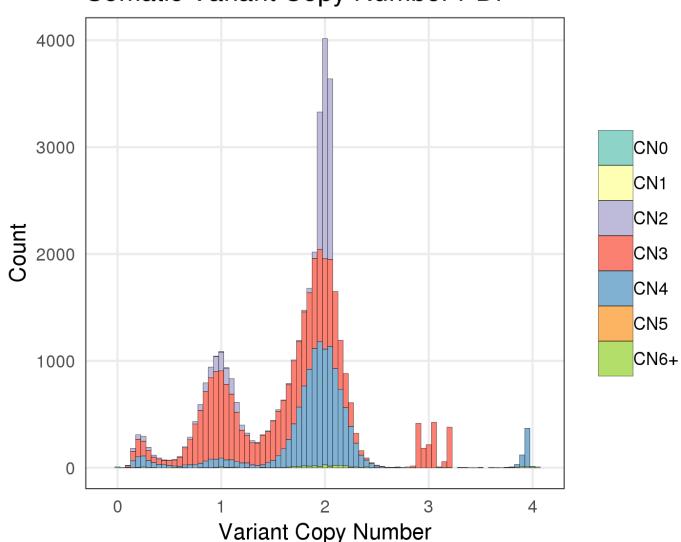
### QC plots



### Copy Number PDF



### Somatic Variant Copy Number PDF





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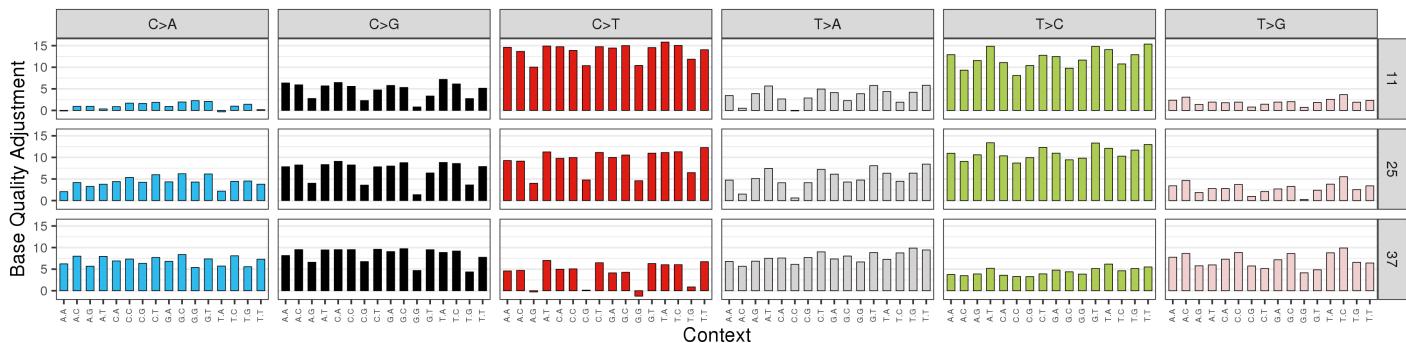
SAMPLE

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## Reference Sample BQR plot



## Tumor Sample BQR plot

