



# ORANGE Report

SAMPLE

Test

PLATINUM VERSION

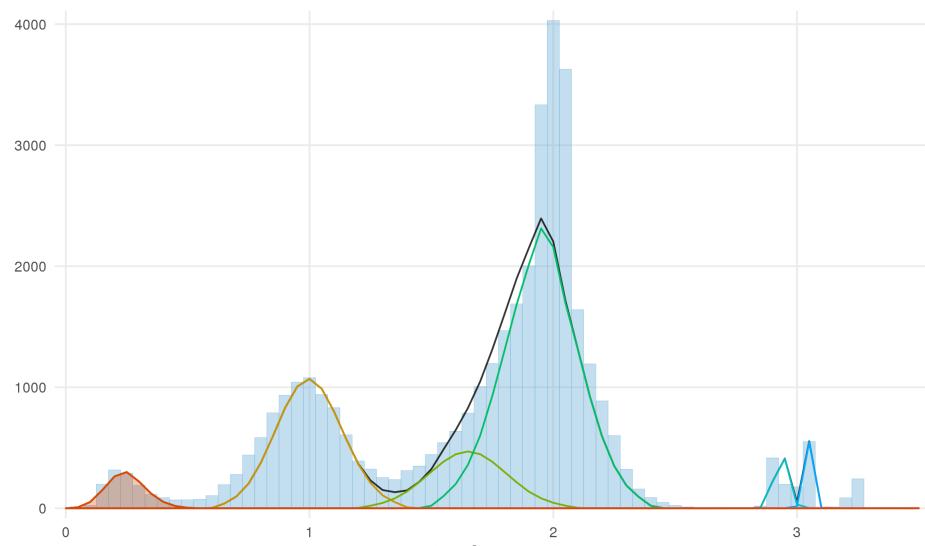
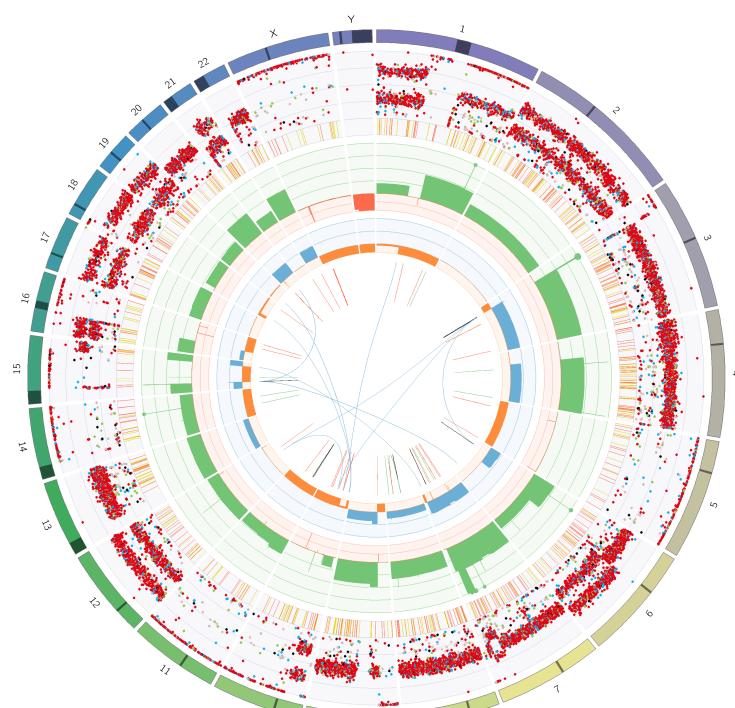
5.30

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
Copy number drivers:	1 (PTEN)
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:	72 (Pan 22%   Skin 34%)
Max complex cluster size:	8
Telomeric SGLs:	0
Number of LINE insertions:	4





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## Somatic Findings

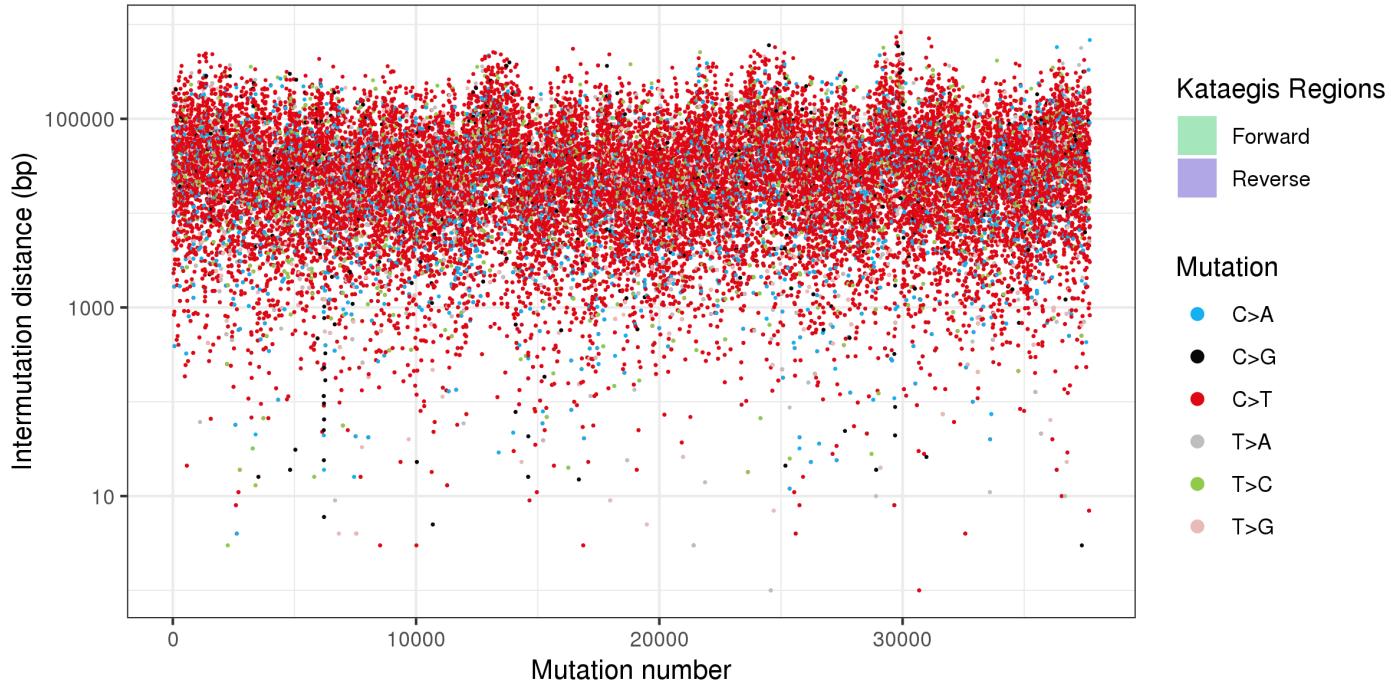
### Driver variants (6)

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 upstream	1.7	2.0	0.0	Yes	Yes	100%	100%	4410	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)

CHR	REGION	GENE	TYPE	CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
10	q23.31	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)

CHR	REGION	GENE	TYPE	CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
3	p24.2	CFL1P7	full gain	12	NA	NA	NA	NA	NA

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

CHR	REGION	GENE	TYPE	CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
16	q21	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	66	2.0	0.0
10q23.31	PTEN	Intron 6 Downstream	DEL	66	2.0	0.0

## Other potentially interesting gene disruptions (0)

NONE

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

NONE

## Structural driver plots (3)



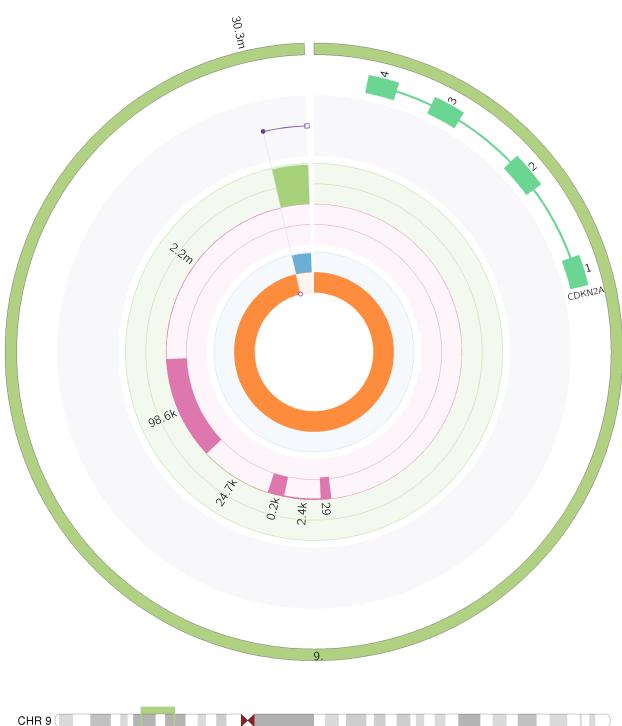
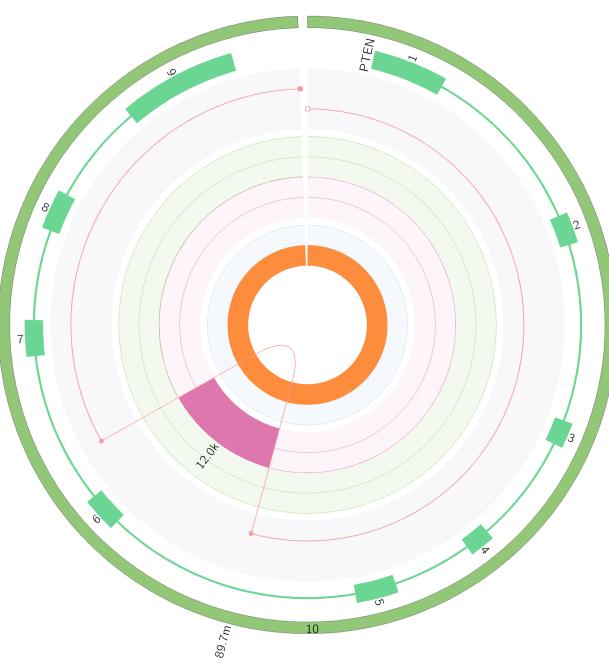
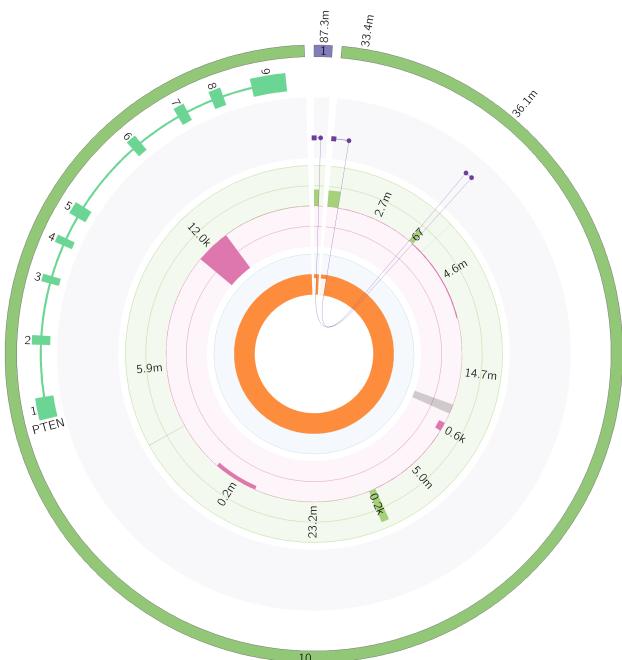
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## Germline Findings

### 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 disruptions (0)

NONE

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

NONE

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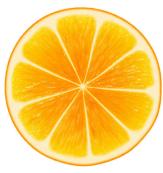
### Germline CN aberrations (0)

NONE

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### Pharmacogenetics (1)

GENE	GENOTYPE	FUNCTION	LINKED DRUGS	SOURCE
DPYD	*1_HOM	Normal Function	5-Fluorouracil;Capecitabine;Tegafur	<a href="#">PHARMGKB</a>



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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	Angiogenesis	Bile duct/Ca/bile duct	Bone/Soft tissue: Other	Breast	Cartilaginous neoplasm	Chronic lymphocytic leukemia	Colon/Rectum/Appendix/Small intestine	Esophagus/Stomach	GIST	Glioma	Head and neck: other	Kidney	Leiomyosarcoma	Liposarcoma	Liver	Lung: NET	Lung: Non-small Cell	Lung: Small Cell	Lymphoid tissue	Meningioma	Mesothelioma	Myeloproliferative neoplasm	Osteosarcoma	Ovary/Fallopian tube	Pancreas	Pancreas: NET	Pilo-cystic astrocytoma	Prostate	Salivary gland/Adenoid cystic	Skin: Other	Small intestine/colon/rectum: NET	Thyroid gland	Urinary tract	Uterus: Endometrium	
SNV 96 PAIRWISE SIMILARITY	0%	0%	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%			
GENOMIC POSITION SIMILARITY	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%	1%	0%	0%	0%	0%	0%	0%	0%	1%	0%	0%	0%	0%	0%	0%	1%	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%			
SEX (MALE)	62%	16%	49%	53%	1%	78%	67%	56%	83%	65%	68%	76%	70%	29%	60%	76%	46%	44%	52%	57%	59%	61%	82%	46%	46%	0%	52%	60%	48%	100%	50%	67%	59%	76%	73%	0%

## SNV SIGNATURES

SIG 7 UV (24251)	34700%	1920%	4140%	98%	1260%	176000%	49600%	1670%	2580%	24700%	50000%	144%	2910%	9970%	14300%	5700%	11400%	791%	1600%	2350%	39800%	34%	7890%	457000%	7990%	4330%	3580%	9450%	20100%	5660%	2930%	63%	10600%	27800%	747%	6130%									
SIG 6 MMR (0)	50%	22%	36%	44%	36%	44%	44%	31%	40%	40%	49%	27%	42%	42%	50%	42%	38%	34%	46%	42%	44%	45%	38%	40%	44%	30%	43%	38%	42%	40%	43%	48%	32%	38%	39%	22%									
SIG 4 SMOKING (2115)	664%	99%	107%	650%	120%	10000%	10000%	99%	189%	145%	10000%	93%	124%	99%	10000%	96%	97%	41%	9%	10000%	5000%	99%	169%	10000%	227%	101%	166%	127%	49700%	152%	98%	99%	10000%	322%	92%	376%	105%								
SIG 2 13 AID APOBEC (3860)	20900%	67%	103%	98%	85%	41600%	10300%	100%	98%	80%	99%	88%	152%	518%	458%	1980%	82%	125%	99%	4110%	34%	865%	16100%	99%	100%	165%	98%	35300%	225%	99%	92%	677%	96%	59%	105%	99%	52%	90%	376%	22%					
SIG 17 (1119)	1390%	97%	94%	97%	95%	22100%	208%	541%	24%	98%	10000%	97%	100%	285%	366%	100%	10000%	93%	97%	81%	10300%	97%	1560%	39300%	95%	99%	99%	18000%	99%	93%	92%	701%	353%	90%	97%	99%	92%	97%	99%	99%	99%	99%			
SIG 11 (1328)	2410%	99%	100%	97%	100%	5370%	1870%	100%	100%	542%	93%	100%	336%	98%	252%	267%	385%	99%	123%	99%	923%	87%	431%	1260%	384%	100%	161%	98%	14600%	271%	55%	55%	219%	98%	212%	99%	99%	99%	99%	99%	99%	99%	99%	99%	99%
SIG 10 POLE (0)	45%	24%	25%	37%	25%	50%	42%	15%	25%	32%	48%	16%	31%	41%	48%	37%	45%	38%	46%	30%	24%	45%	39%	42%	39%	44%	28%	42%	38%	35%	29%	40%	48%	22%	28%	41%	41%	61%							
SIG 1 (0)	0%	-1000%	4%	3%	2%	-1000%	-10000%	3%	10%	2%	1%	4%	6%	2%	-1000%	22%	7%	24%	42%	8%	-1000%	41%	-1000%	5%	2%	0%	4%	-1000%	0%	2%	22%	-1000%	8%	4%	6%										

## PERCENTILES

SNV COUNT (37701)	1470%	91%	96%	97%	95%	2630%	860%	86%	83%	274%	92%	93%	99%	99%	327%	106%	384%	61%	40%	96%	1130%	38%	227%	2120%	211%	99%	99%	98%	5190%	97%	108%	56%	180%	111%	79%	87%
MS INDELS TMB (0.12)	634%	18%	48%	83%	62%	985%	134%	14%	16%	99%	48%	44%	88%	85%	94%	83%	99%	27%	12%	48%	99%	64%	94%	985%	98%	78%	40%	93%	2330%	37%	68%	36%	91%	21%	31%	
TELOMERIC SGL (0)	50%	32%	39%	26%	34%	50%	50%	38%	29%	49%	38%	40%	46%	2%	29%	42%	30%	25%	40%	46%	48%	28%	44%	50%	50%	42%	43%	39%	44%	35%	30%	44%				
SIMPLE DUP 32B 20B (3)	98%	15%	26%	6%	15%	100%	100%	7%	28%	20%	4%	30%	12%	4%	6%	56%	17%	8%	8%	84%	10%	20%	93%	53%	30%	55%	48%	98%	24%	5%	7%	12%	3%	14%	28%	
MAX COMPLEX SIZE (8)	94%	22%	21%	38%	8%	76%	96%	28%	8%	51%	8%	36%	48%	9%	30%	50%	40%	10%	32%	63%	80%	25%	7%	100%	24%	20%	77%	99%	8%	33%	37%	74%	70%	14%	53%	
LINE (4)	133%	17%	52%	88%	48%	200%	200%	2%	10%	96%	98%	26%	91%	92%	77%	86%	81%	32%	52%	98%	100%	88%	95%	133%	75%	38%	52%	94%	100%	26%	41%	32%	94%	65%	6%	46%

## FEATURES

BRAF (1)	0%	1%	2%	1%	1%	0%	6%	13%	2%	2%	1%	1%	0%	0%	1%	0%	0%	7%	2%	2%	0%	52%	0%	0%	0%	2%	2%	7%	0%	0%	10%	4%	1%			
CDKN2A (1)	0%	15%	44%	23%	8%	0%	4%	37%	39%	68%	48%	23%	4%	4%	8%	9%	41%	3%	10%	0%	52%	69%	0%	19%	10%	70%	12%	0%	2%	13%	24%	8%	24%	45%	7%	
PTEN (1)	0%	8%	6%	2%	14%	0%	0%	10%	6%	5%	40%	4%	11%	12%	16%	3%	5%	6%	10%	4%	4%	23%	0%	0%	12%	5%	1%	4%	0%	42%	10%	16%	0%	6%	47%	
SF3B1 (0.15)	0%	1%	5%	0%	2%	0%	8%	0%	1%	0%	0%	0%	1%	0%	1%	1%	1%	14%	1%	0%	0%	2%	0%	0%	1%	0%	1%	0%	0%	0%	0%	0%	0%	0%	0%	0%
TERT (1)	0%	16%	8%	6%	1%	0%	1%	2%	0%	2%	70%	17%	12%	0%	28%	16%	0%	7%	0%	0%	8%	68%	5%	0%	2%	6%	1%	0%	0%	0%	0%	0%	0%	0%	0%	
TP53 (0.00)	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%	0%	0%	0%	0%
INDEL_ALB (1)	0%	0%	6%	2%	1%	0%	0%	1%	1%	0%	0%	0%	0%	0%	0%	0%	0%	62%	0%	1%	2%	1%	0%	0%	1%	1%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%



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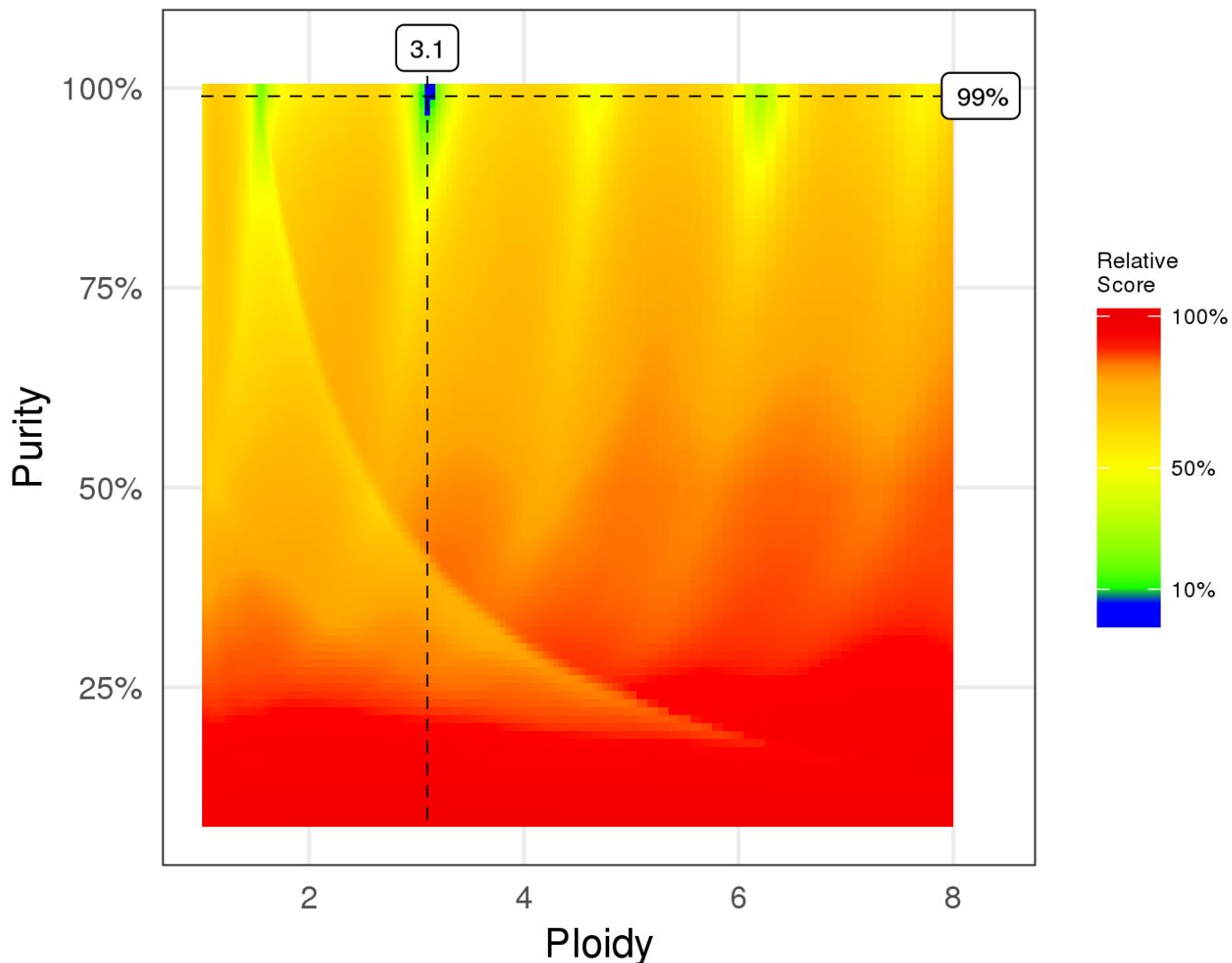
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### Quality Control

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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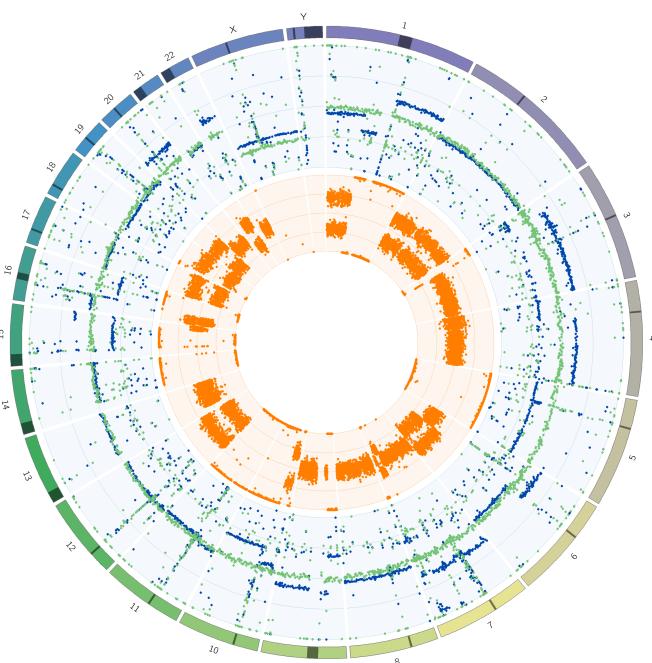
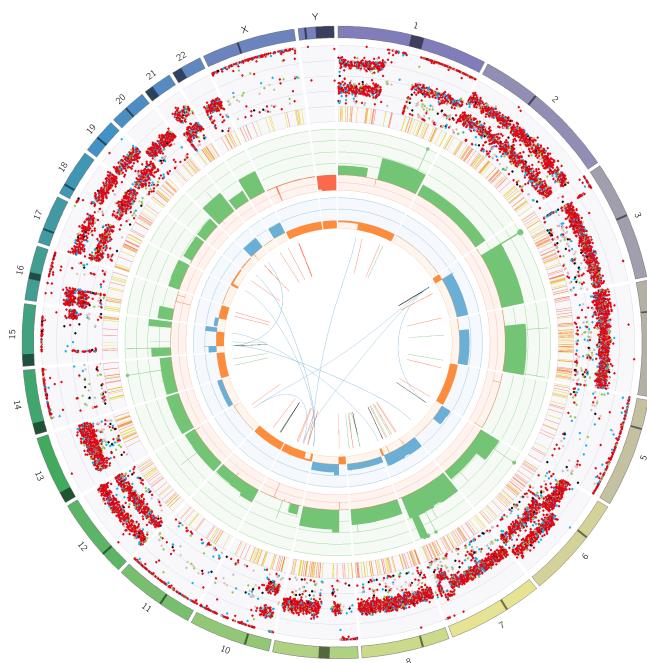
### Coverage Stats

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

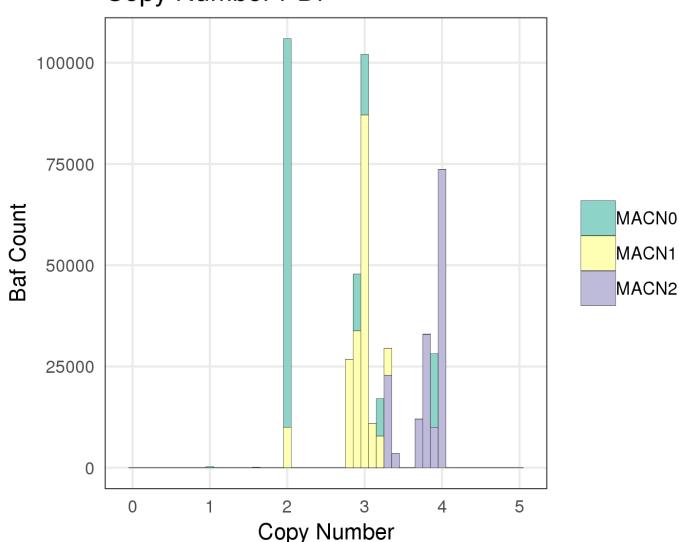
### Excluded Percentages

	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%

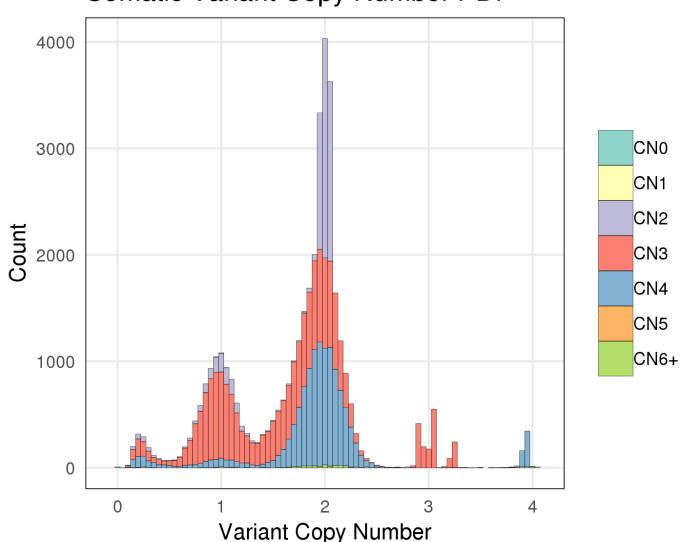
### QC plots



### Copy Number PDF



### Somatic Variant Copy Number PDF





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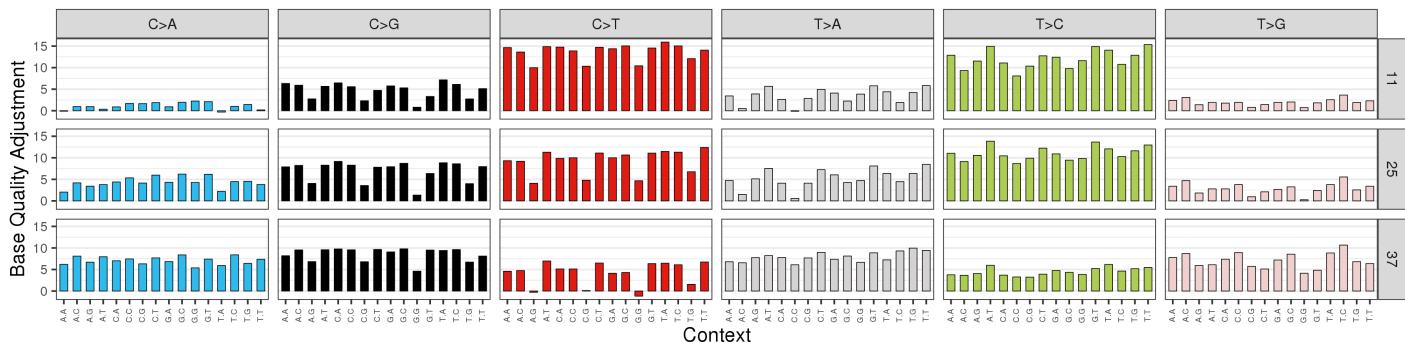
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### Reference Sample BQR plot



### Tumor Sample BQR plot

