



ORANGE Report

SAMPLE

Test

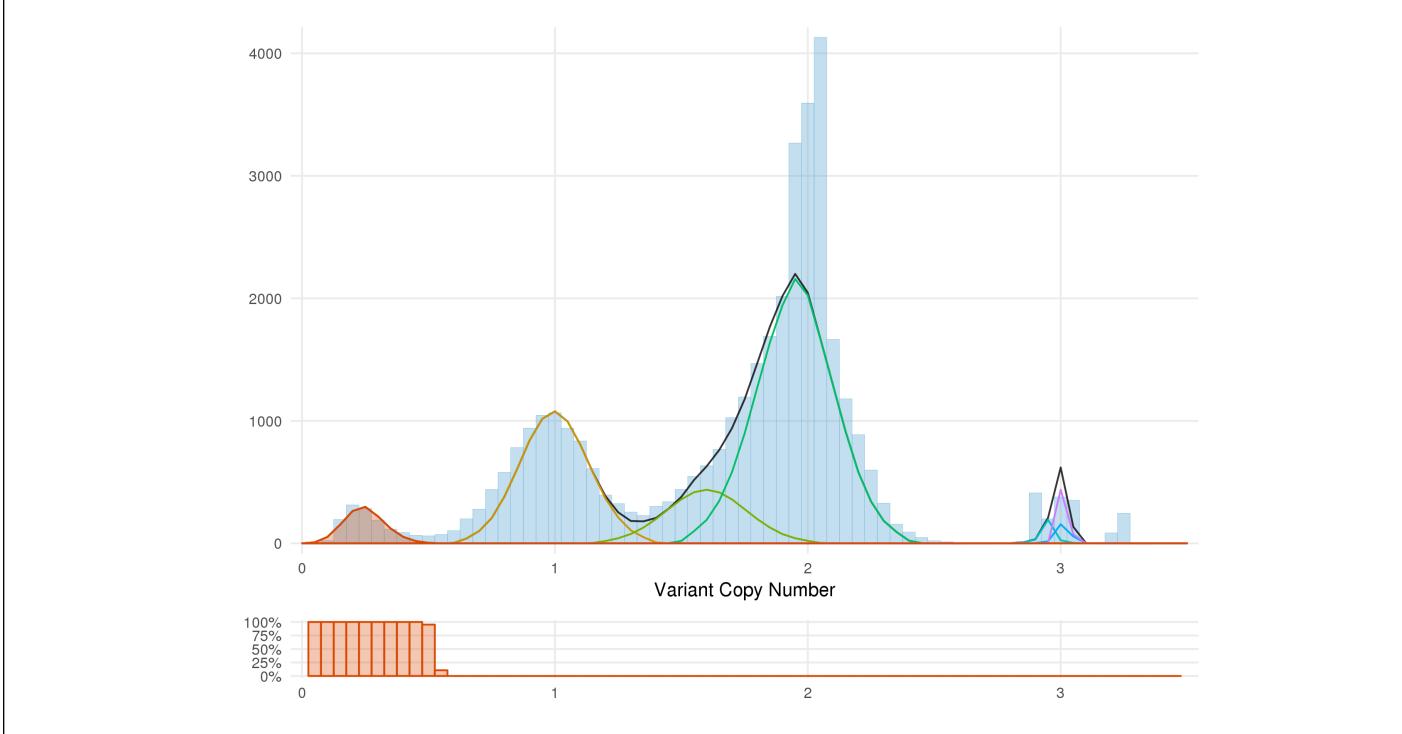
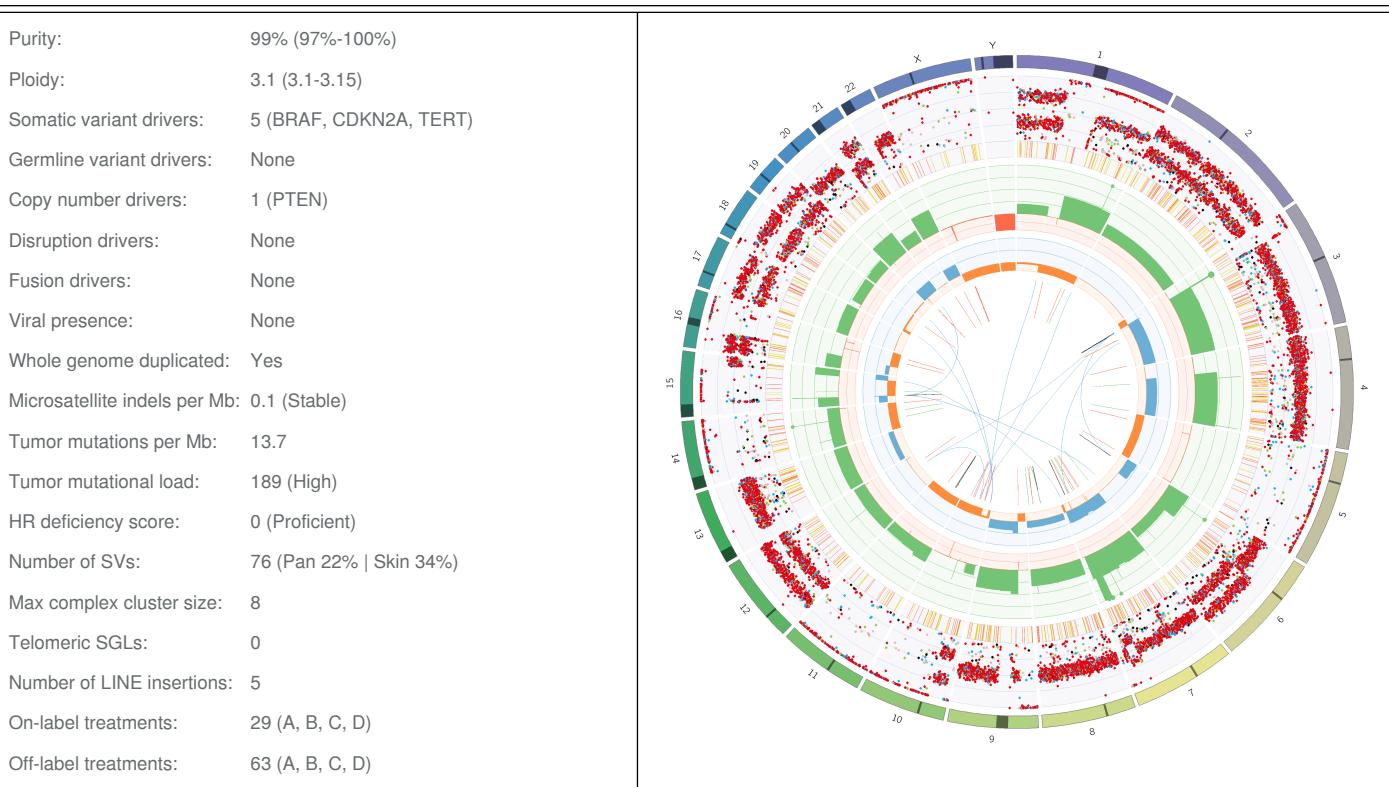
PLATINUM VERSION

5.25

CONFIGURED PRIMARY TUMOR
skin melanoma (DOID 8923)

CUPPA CANCER TYPE
Melanoma (100%)

QC
PASS





Somatic Findings

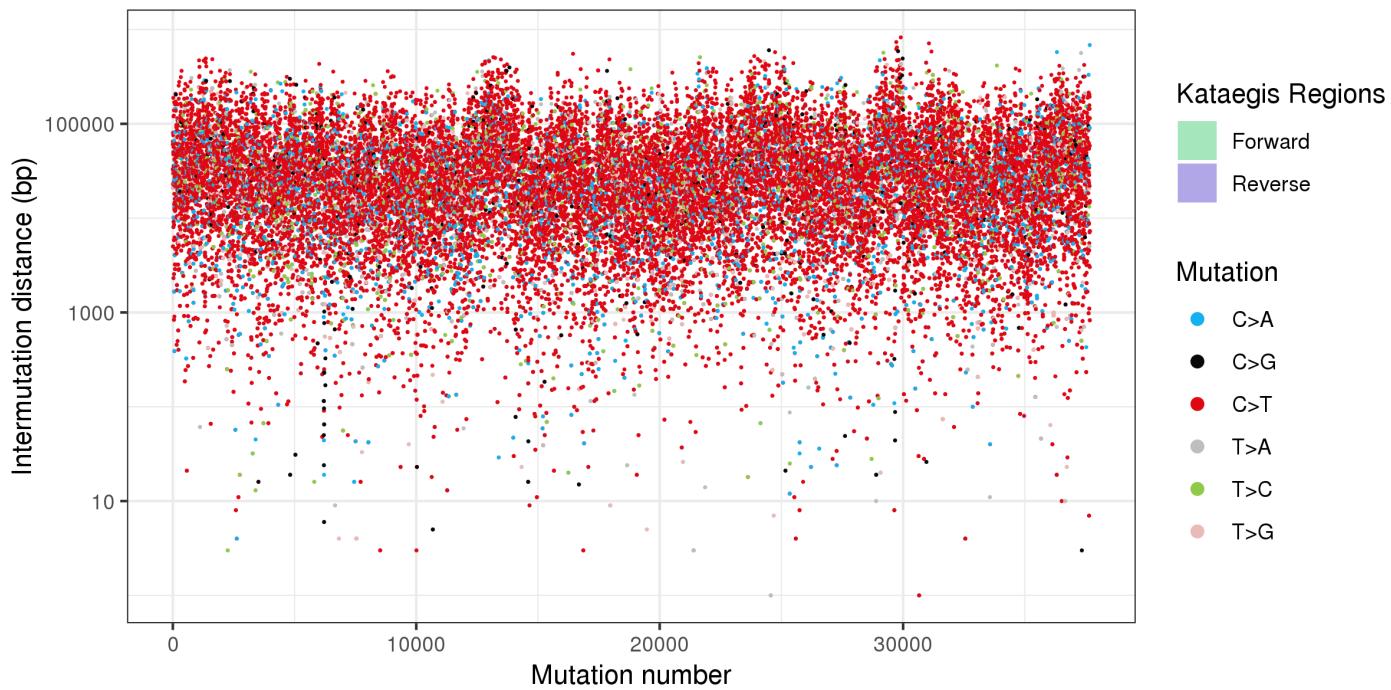
Driver variants (5)

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 (0)

NONE

Kataegis plot



Driver amps/dels (1)

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

Other regions with amps (0)

NONE

Other autosomal regions with dels (0)

NONE



ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.25

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)



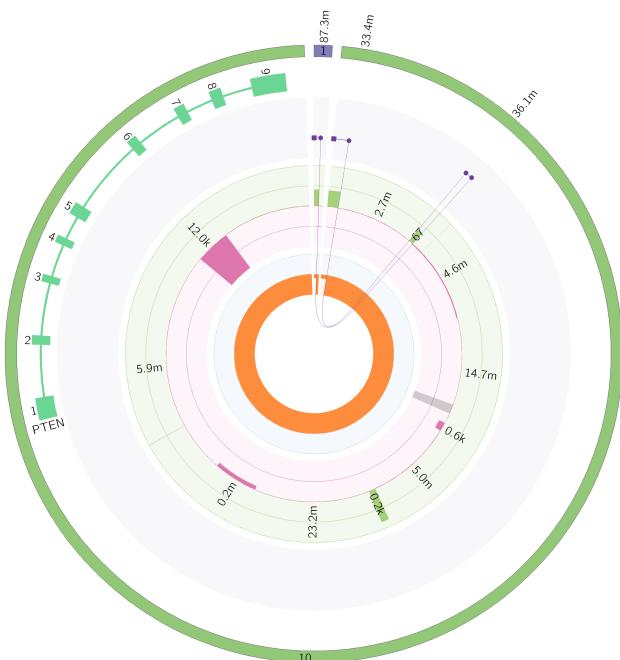
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SAMPLE

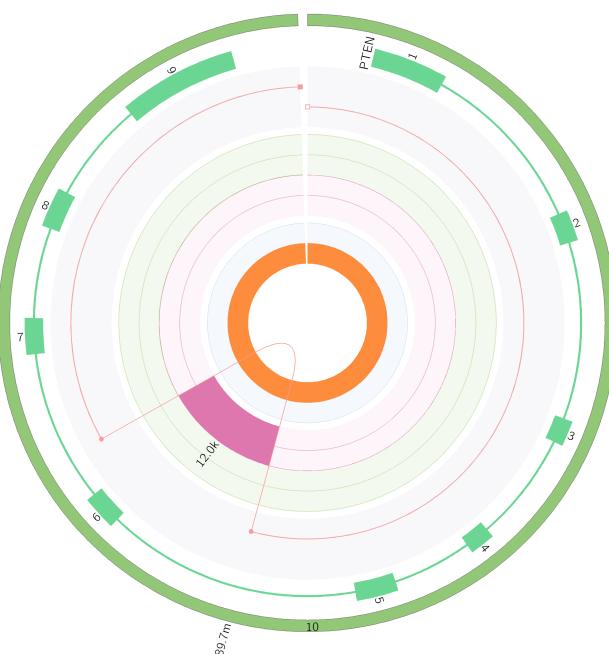
Test

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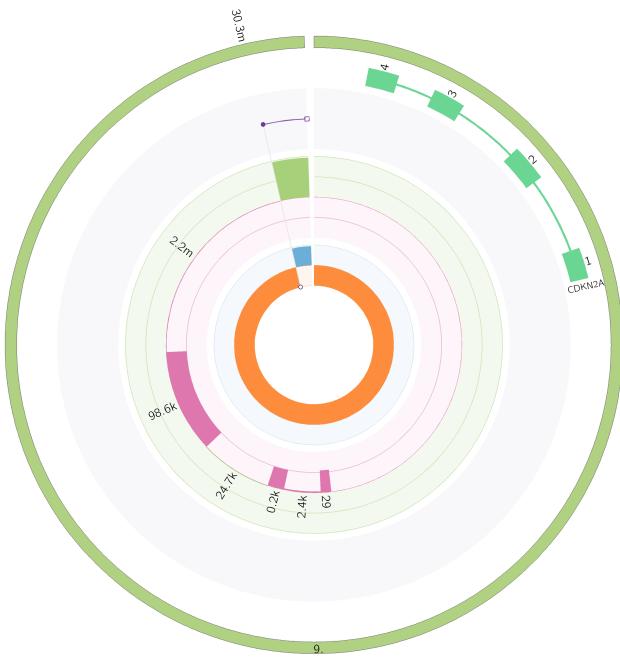
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CHR 1 CHR 10



CHR 10 CHR 10



CHR 9 CHR 10



Germline Findings

Driver variants (0)

NONE

Other potentially relevant variants (0)

NONE

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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Cohort Comparison

	Acute myeloid leukemia	Anogenital	Bile duct/Gallbladder	Bone/Soft tissue: Other	Breast	Cartilaginous neoplasm	Chronic lymphocytic leukemia	Colon/Rectum/Appendix/Small intestine	GIST	Esophagus/Stomach	Head and neck: other	Kidney	Liposarcoma	Liver	Lung: NET	Lung: Small Cell	Lymphoid tissue	Medulloblastoma	Melanoma	Mesotheli瘤	Myeloproliferative neoplasm	Osteosarcoma	Ovary/Fallopian tube	Pancreas	Pilocytic astrocytoma	Prostate	Salivary gland/Adenoid cystic	Skin: Other	Small intestine/Colon/rectum: NET	Thyroid gland	Urothelial tract	Uterus: Endometrium				
SNV 96 PAIRWISE SIMILARITY	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	1%	0%	0%	0%	0%	0%	0%	0%	97%	0%	0%	0%	0%	0%	0%	0%	2%	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%	100%	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%	1%	0%	0%	95%	0%	0%	0%	0%	0%	0%	1%	0%	0%	1%	0%					
DNA COMBINED	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	100%	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 (24240)	34700%	1920%	4140%	98%	1260%	175000%	49600%	1670%	2580%	24700%	50000%	144%	2910%	9970%	14300%	5700%	11400%	791%	1600%	2350%	39700%	34%	7890%	456000%	7990%	4330%	3570%	9450%	20100%	5660%	2930%	63%	10600%	27800%	746%	6130%		
SIG 6 MMR (0)	50%	22%	36%	44%	38%	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 SMOKEING (2106)	661%	99%	106%	64%	119%	101%	101%	99%	188%	144%	101%	93%	123%	99%	101%	96%	97%	41%	9%	101%	4980%	99%	168%	101%	226%	100%	167%	126%	49500%	151%	98%	99%	101%	320%	92%	375%	375%	105%
SIG 2 13 AID APOBEC (3858)	20900%	67%	103%	98%	85%	41500%	10300%	100%	98%	804%	99%	88%	152%	518%	376%	455%	1980%	82%	125%	99%	4110%	34%	864%	16100%	99%	100%	165%	98%	35300%	225%	99%	52%	676%	96%	59%	105%	105%	
SIG 17 (1121)	1400%	97%	94%	97%	93%	22100%	209%	55%	24%	98%	101%	97%	100%	285%	367%	100%	101%	93%	97%	81%	10300%	97%	1570%	39400%	95%	99%	96%	99%	18000%	99%	93%	92%	703%	354%	90%	97%	97%	
SIG 11 (1350)	2450%	99%	100%	97%	100%	5460%	1900%	100%	100%	551%	93%	100%	342%	98%	358%	272%	392%	99%	125%	99%	939%	87%	438%	1380%	390%	100%	163%	98%	14900%	276%	235%	55%	566%	223%	98%	218%	218%	
SIG 10 POLE (0)	45%	24%	26%	37%	26%	50%	42%	15%	25%	32%	48%	16%	31%	41%	48%	37%	45%	38%	46%	30%	24%	48%	39%	42%	39%	44%	28%	42%	38%	35%	29%	40%	48%	22%	28%	41%		
SIG 1 (0)	0%	10000%	4%	3%	2%	10000%	10000%	3%	10%	2%	1%	4%	6%	2%	10000%	22%	7%	24%	42%	8%	10000%	10000%	5%	2%	0%	4%	10000%	0%	2%	22%	10000%	8%	4%	6%				

PERCENTILES

SNV COUNT (37675)	1470%	91%	96%	97%	95%	2630%	859%	86%	83%	273%	92%	93%	99%	99%	327%	106%	384%	61%	40%	96%	1130%	38%	226%	2110%	99%	99%	98%	5180%	97%	108%	56%	180%	111%	79%	87%	
MS INDELS TMB (0.12)	622%	18%	47%	82%	61%	967%	132%	1%	16%	98%	45%	43%	87%	85%	93%	82%	99%	27%	11%	47%	99%	64%	94%	967%	97%	76%	39%	93%	2350%	37%	68%	36%	98%	91%	20%	30%
TELOMERIC SGL (0)	50%	32%	39%	26%	34%	50%	50%	38%	29%	43%	38%	40%	46%	2%	29%	42%	30%	25%	40%	46%	48%	28%	44%	50%	17%	38%	36%	34%	50%	42%	43%	39%	44%	35%	30%	44%
SIMPLE DUP 32B 2008 (10)	333%	60%	76%	64%	62%	333%	333%	48%	66%	72%	31%	73%	58%	44%	42%	90%	64%	55%	46%	93%	99%	56%	67%	167%	83%	74%	89%	85%	250%	64%	62%	64%	62%	66%	62%	
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%	7%	24%	20%	77%	99%	8%	33%	37%	74%	70%	14%	53%
LINE (5)	167%	19%	54%	91%	54%	250%	250%	3%	10%	100%	99%	28%	94%	93%	81%	90%	88%	36%	60%	99%	125%	90%	96%	167%	77%	42%	57%	96%	125%	30%	47%	37%	95%	68%	8%	50%

FEATURES

BRAF (1)	0%	1%	2%	1%	1%	0%	6%	13%	2%	2%	1%	1%	0%	0%	1%	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%	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%	0%			
SF3B1 (0.15)	0%	1%	5%	0%	2%	0%	8%	0%	1%	0%	0%	0%	0%	1%	0%	1%	1%	14%	1%	0%	0%	2%	0%	0%	1%	0%	1%	0%	1%	0%	0%	1%	0%	0%	1%	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%	13%	38%	0%	33%	70%	7%			
TP53 (0.00)	0%	0%	0%	0%	0%	0%	0%	1%	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%	1%	0%	
INDEL_ALB (1)	0%	0%	6%	2%	1%	0%	0%	1%	1%	0%	0%	4%	1%	0%	0%	0%	62%	0%	1%	2%	1%	0%	0%	0%	1%	1%	0%	0%	3%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%



ORANGE Report

SAMPLE

Test

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5.25

Clinical Evidence

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

Applicable on-label evidence

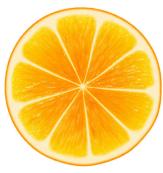
TREATMENT	RESPONSIVE EVIDENCE	RESISTANCE EVIDENCE
Cobimetinib + Vemurafenib	BRAF p.V600E (A - CGI)	
Dabrafenib	BRAF p.V600E (A - CGI)	
Dabrafenib + Trametinib	BRAF p.V600E (A - CGI, CIViC)	
Trametinib	BRAF p.V600E (A - CGI)	
Vemurafenib	BRAF p.V600E (A - CGI, CIViC)	

Applicable off-label evidence

TREATMENT	RESPONSIVE EVIDENCE	RESISTANCE EVIDENCE
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)	
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)
Vemurafenib		BRAF p.V600E (B - CIViC)

Applicable trials

NONE



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Test

PLATINUM VERSION

5.25

Other potentially interesting on-label evidence

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

Other potentially interesting 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)	
Chemotherapy		PTEN partial loss (B - CIViC)

Other potentially interesting trials

NONE



ORANGE Report

SAMPLE

Test

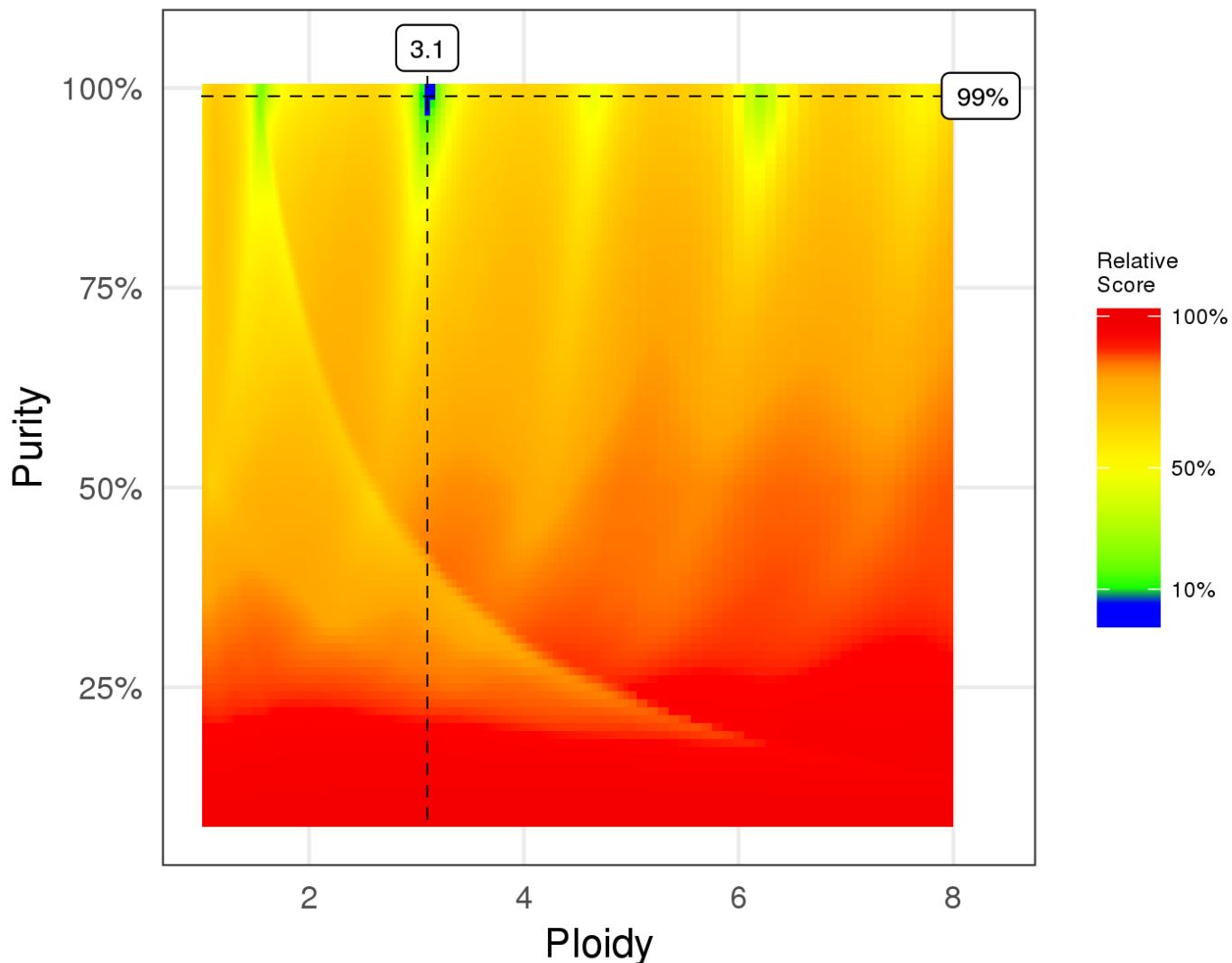
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Quality Control

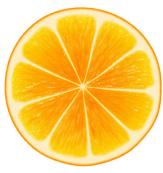
QC	FIT METHOD	MEAN DEPTH	CONTAMINATION	UNS. CN SEGMENTS	DELETED GENES
PASS	NORMAL	111	0%	0	2

Purity/Ploidy Scores



Flagstats

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



ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.25

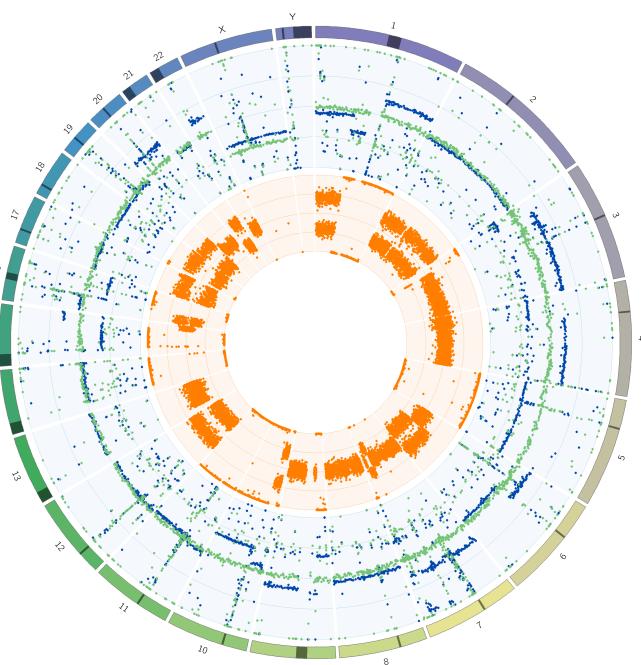
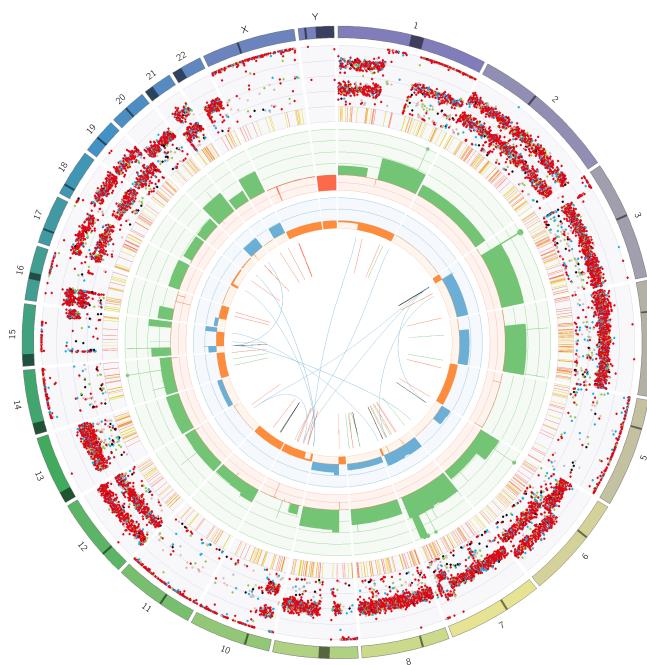
Coverage Stats

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

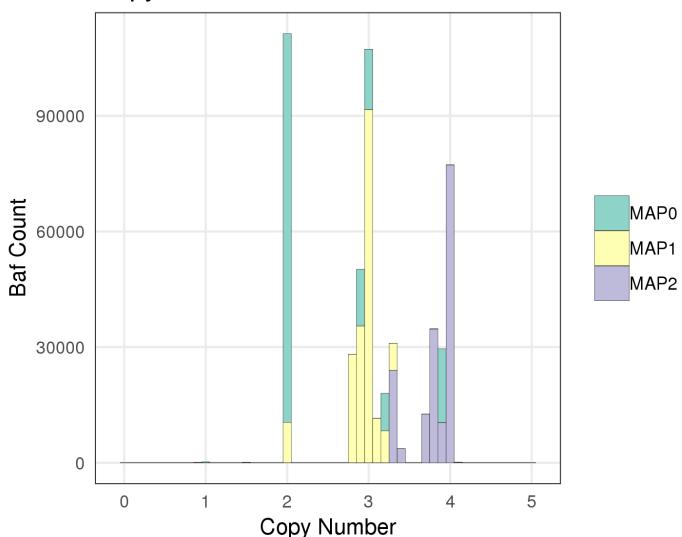
Excluded Percentages

	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%

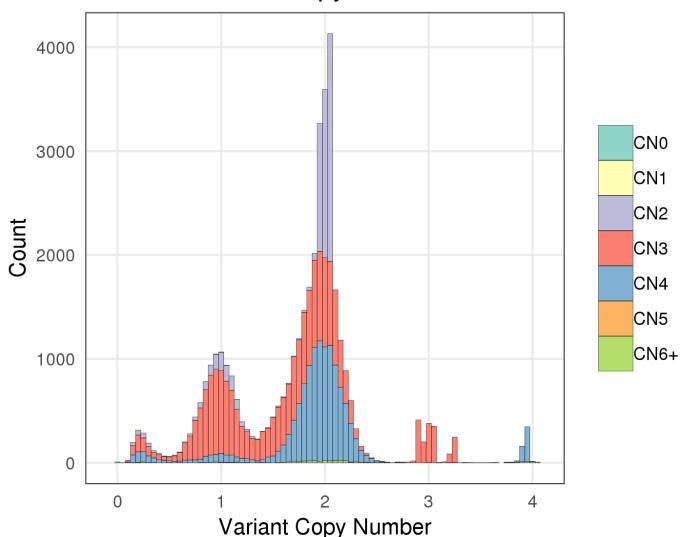
QC plots



Copy Number PDF



Somatic Variant Copy Number PDF





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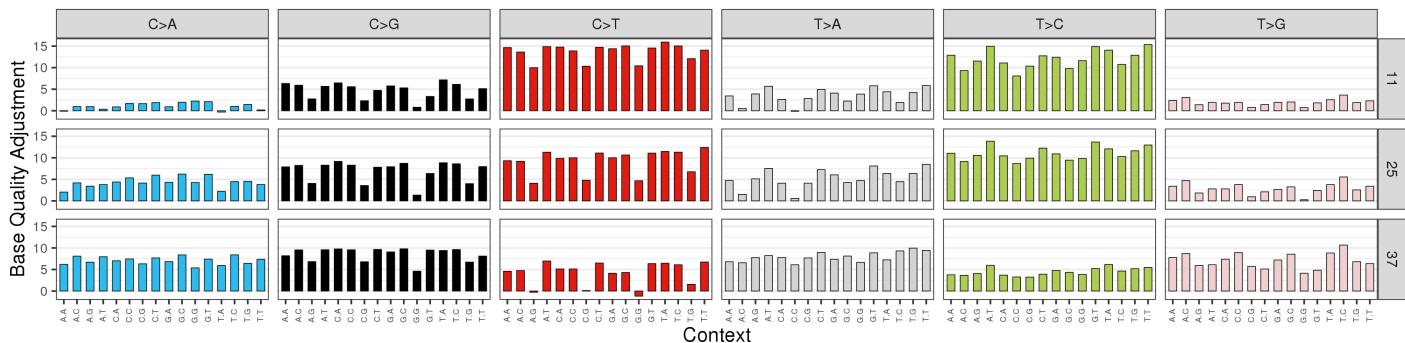
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PLATINUM VERSION

5.25

Reference Sample BQR plot



Tumor Sample BQR plot

