



CONFIGURED PRIMARY TUMOR

skin melanoma (DOID 8923)

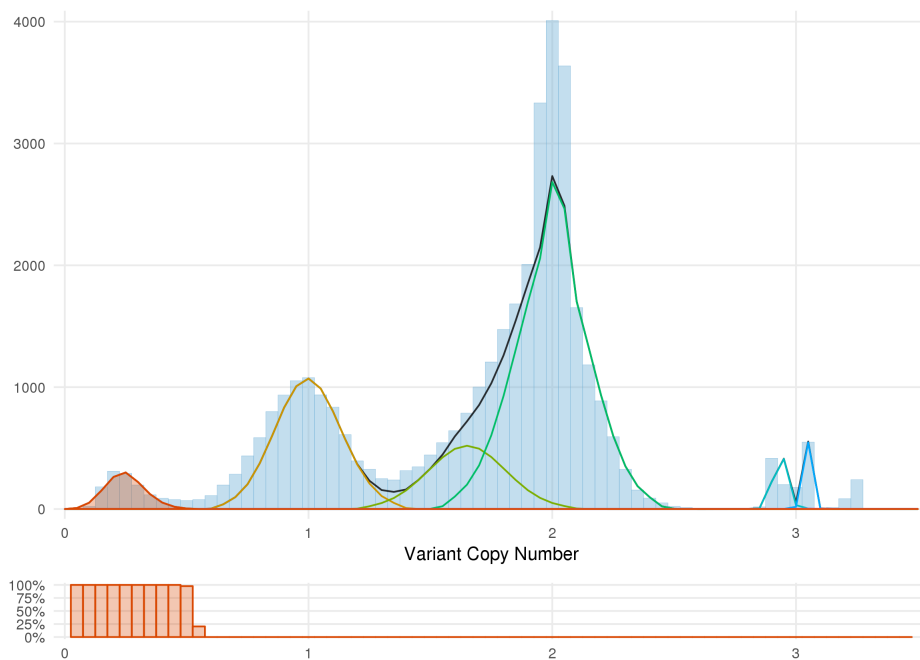
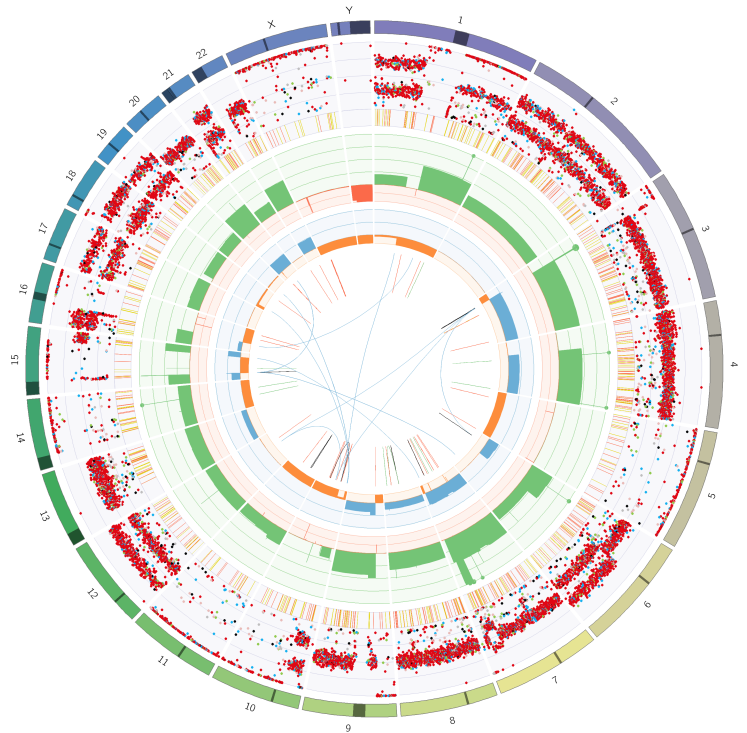
CUPPA CANCER TYPE

Melanoma (100%)

QC

PASS

Purity: 99% (97%-100%)
Ploidy: 3.10 (3.10-3.15)
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: 183 (High)
HR deficiency score: 0.0 (Proficient)
DPYD status: *1_HOM (Normal Function)
Number of SVs: 75 (Pan 22% | Skin 34%)
Max complex cluster size: 8
Telomeric SGLs: 0
Number of LINE insertions: 3





Somatic Findings

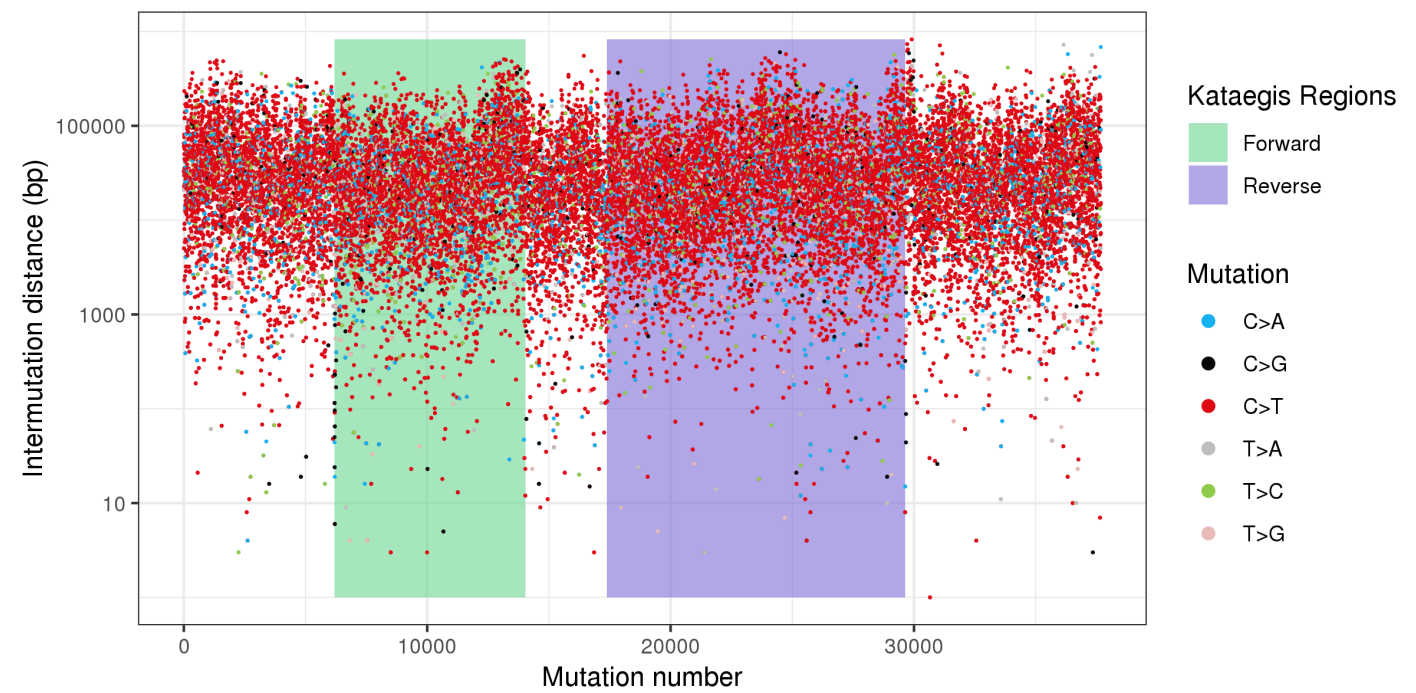
Driver variants (6)

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



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%

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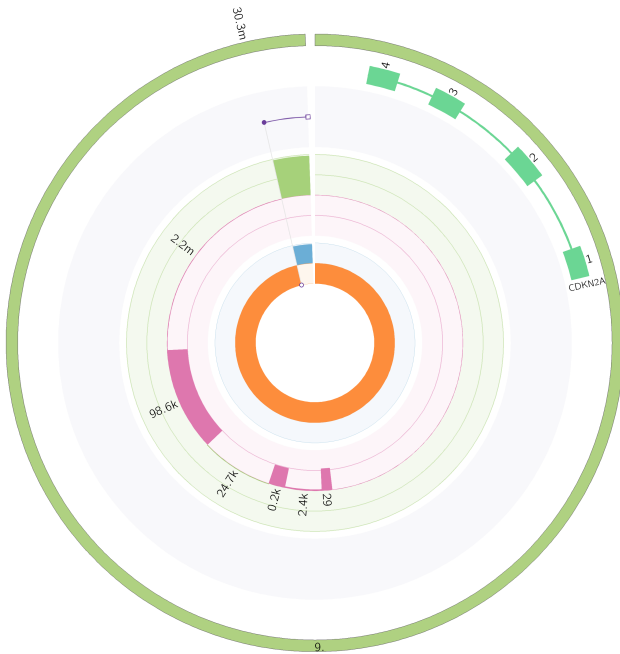
Signature allocations (12)

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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)







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



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



RNA Findings

QC	TOTAL FRAGMENTS	NON-DUPLICATE FRAGMENTS	DUPLICATE RATE
NA			

Genes with high expression (0)

NONE

Genes with low expression (0)

NONE

Known fusions detected in RNA and not in DNA (0)

NONE

Promiscuous fusions detected in RNA and not in DNA (0)

NONE

Potentially interesting novel splice junctions - Skipped exons (0)

NONE

Potentially interesting novel splice junctions - Novel exon/intron (0)

NONE



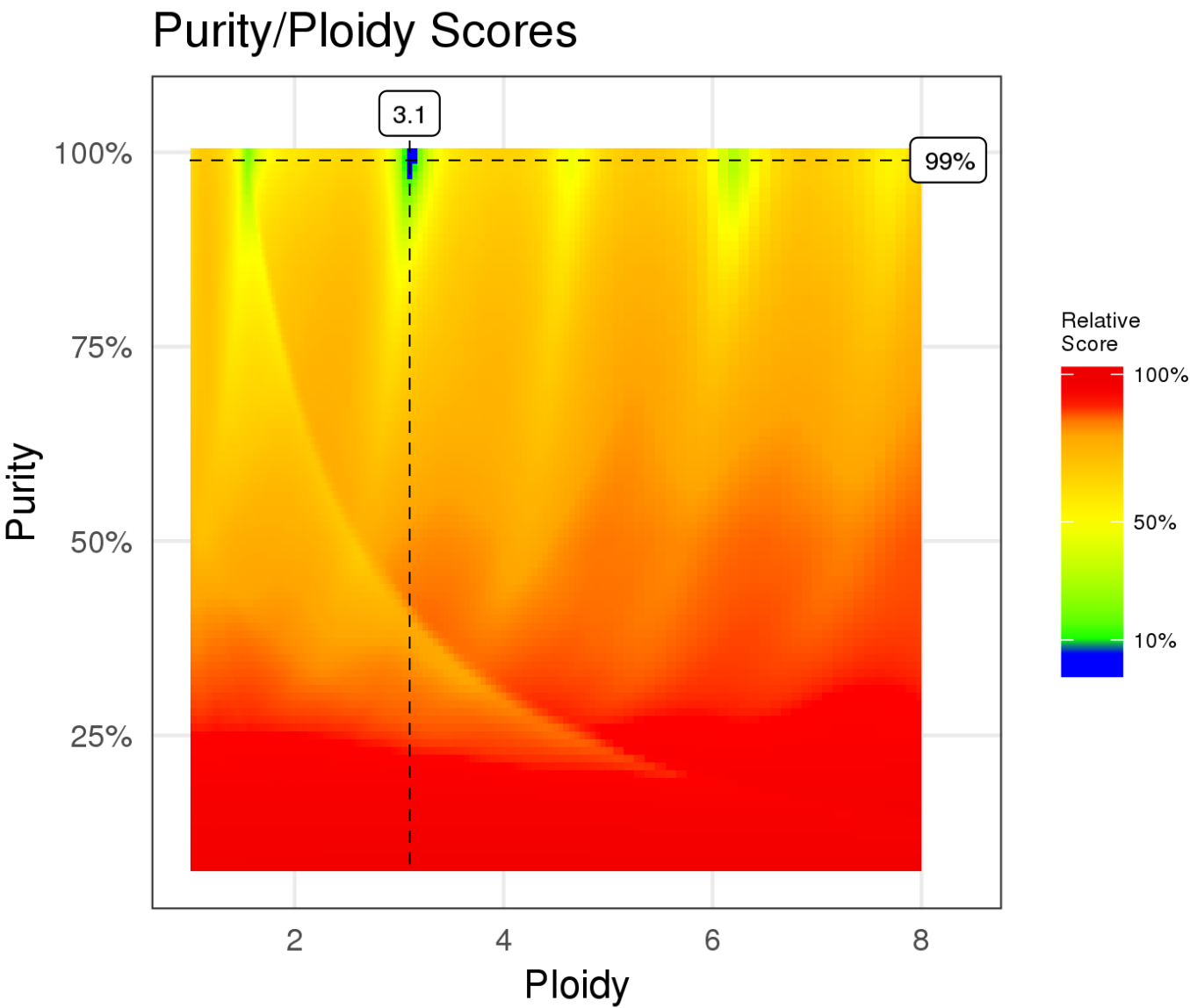
Cohort Comparison

	Acute myeloid leukemia	Anogenital	Bile duct/Gallbladder	Bone/Soft tissue: Other	Breast	Cartilaginous neoplasm	Colorectum/Appendix/Smallintestine	Esophagus/Stomach	GIST	Glioma	Head and neck: other	Kidney	Kidney-CHRC	Leiomyosarcoma	Liposarcoma	Liver	Lung: NET	Lung: Non-small Cell	Lung: Small Cell	Lymphoid tissue	Medulloblastoma	Melanoma	Mesothelium	Myeloproliferative neoplasm	Osteosarcoma	Ovary/Fallopian tube	Pancreas	Pancreas: NET	Piloeytic astrocytoma	Prostate	Salivary gland/Adenoid cystic	Skin: Other	Small intestine/Colorectum: 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%	98%	0%	0%	0%	0%	0%	0%	0%	0%	0%	2%	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%	99%	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%	1%	0%	0%	0%	97%	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%	100%	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 (24216)	32300%	1920%	3910%	97%	1240%	172000%	1680%	2390%	23000%	47500%	1770%	2950%	15800%	9880%	14000%	5740%	11400%	787%	1580%	2330%	39600%	33%	7910%	4040000%	8040%	4260%	3520%	9170%	202000%	9710%	98%	66%	14200%	22400%	742%	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%	56%	39%	30%			
SIG 4 SMOKING (2127)	618%	100%	107%	592%	122%	10000%	99%	100%	1440%	10000%	92%	123%	88600%	99%	19000%	95%	98%	39%	7%	10000%	5030%	99%	166%	10000%	225%	101%	99%	100%	10000%	154%	98%	99%	10000%	320%	93%	376%			
SIG 2 13 AID APOBEC (3842)	20000%	56%	96%	97%	78%	22900%	99%	94%	334%	99%	62%	99%	712%	243%	301%	145%	1110%	60%	93%	100%	3280%	34%	366%	15800%	98%	97%	100%	98%	42200%	100%	93%	57%	815%	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%	10000%	98%	1540%	40000%	94%	99%	94%	676%	17200%	99%	94%	93%	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%	334%	153%	164%	98%	15200%	295%	97%	58%	518%	229%	96%	202%			
SIG 10 POLE (0)	45%	23%	24%	38%	26%	50%	15%	24%	32%	49%	18%	32%	46%	42%	48%	36%	44%	38%	47%	36%	24%	48%	40%	44%	38%	44%	27%	40%	36%	35%	27%	42%	49%	36%	26%	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%	41%	-10000%	-10000%	6%	1%	0%	4%	0%	0%	3%	22%	-10000%	2%	4%	6%			
PERCENTILES																																							
SNV COUNT (37681)	1470%	93%	95%	96%	95%	2630%	64%	80%	273%	95%	90%	99%	470%	100%	324%	99%	384%	61%	37%	98%	1130%	37%	225%	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%	87%	84%	196%	33%	13%	68%	100%	69%	95%	1060%	98%	81%	41%	94%	2560%	39%	69%	42%	122%	97%	32%	52%			
TELOMERIC SGL (0)	50%	36%	38%	28%	36%	50%	39%	30%	44%	42%	39%	46%	42%	4%	26%	42%	33%	27%	41%	47%	48%	28%	44%	50%	20%	38%	35%	35%	50%	42%	39%	38%	47%	44%	34%	48%			
SIMPLE DUP 32B 200B (3)	130%	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%	39%	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%	0%	1%	0%	6%	2%	4%	0%	51%	0%	0%	0%	2%	2%	0%	5%	2%	6%	0%	0%	19%	3%	1%			
CDKN2A (1)	0%	16%	52%	25%	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%	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%	0%	5%	45%			
SF3B1 (0.15)	0%	1%	4%	0%	2%	0%	0%	1%	0%	0%	0%	0%	2%	0%	0%	3%	1%	14%	1%	0%	3%	0%	2%	0%	0%	1%	0%	3%	1%	0%	1%	0%	0%	0%	1%	0%			
TERT (1)	0%	14%	2%	5%	0%	0%	2%	0%	2%	69%	14%	9%	2%	0%	12%	15%	0%	2%	0%	0%	6%	66%	5%	0%	2%	4%	0%	0%	0%	0%	9%	38%	0%	28%	69%	1%			
TP63 (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%	1%	0%			
INDEL_ALB (1)	0%	0%	5%	2%	0%	0%	1%	0%	0%	0%	1%	1%	0%	0%	0%	0%	62%	0%	1%	2%	0%	0%	0%	0%	0%	1%	1%	0%	0%	0%	0%	3%	0%	0%	0%	0%			



Quality Control

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



Flagstats

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



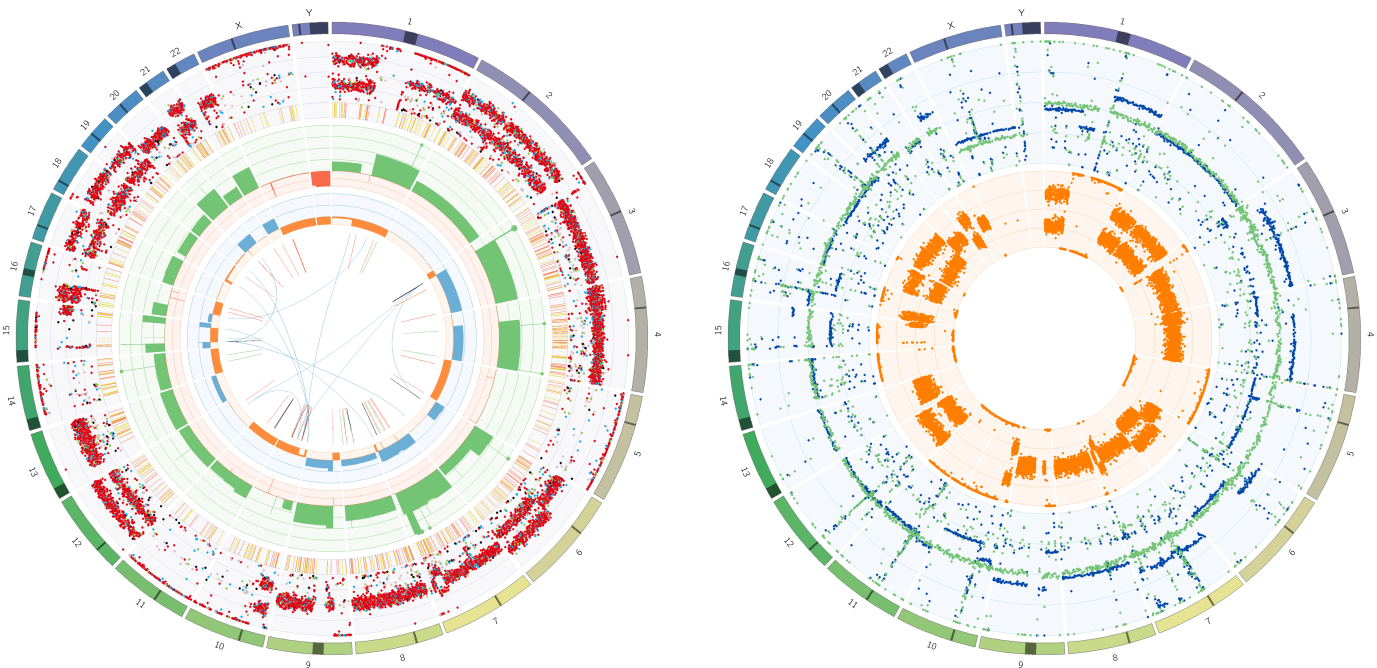
Coverage Stats

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

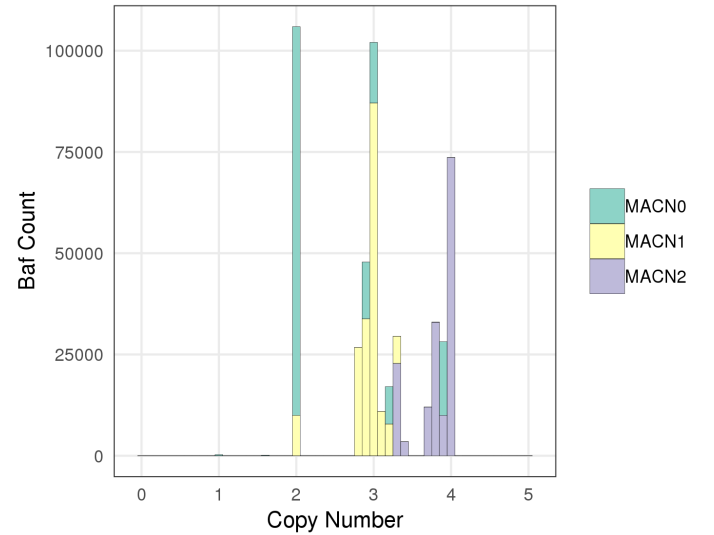
Excluded Percentages

	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%

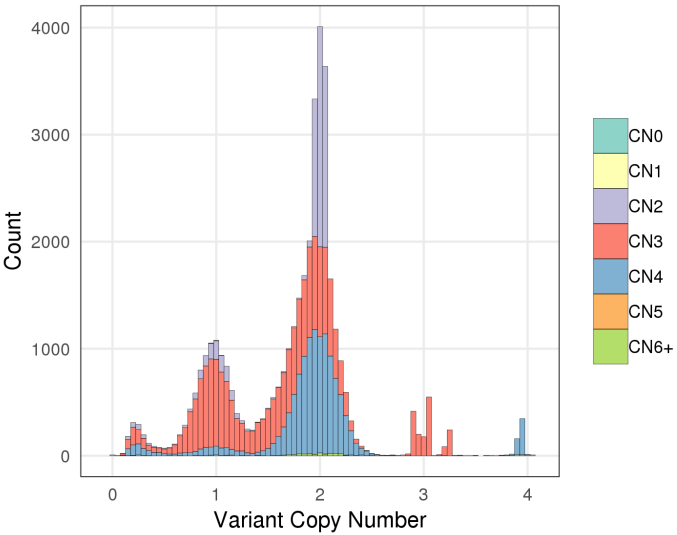
QC plots



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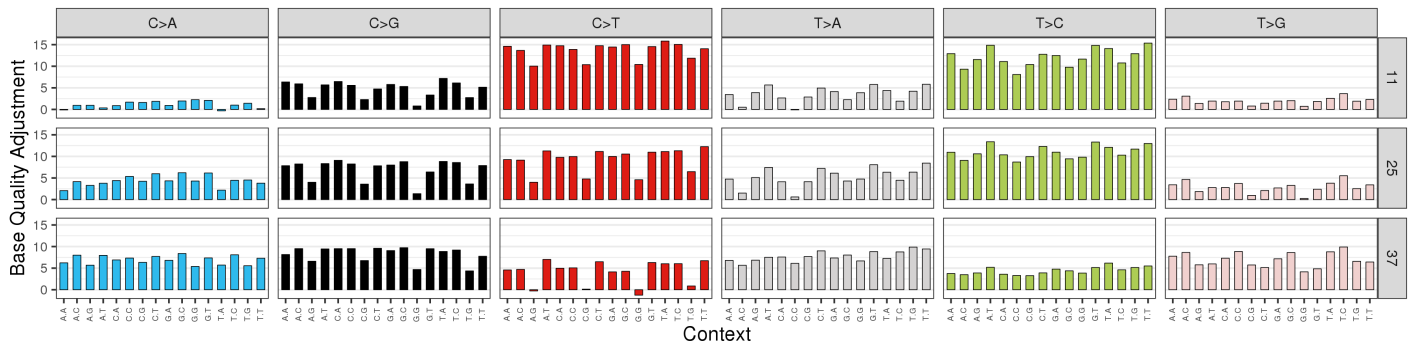


Somatic Variant Copy Number PDF





Reference Sample BQR plot



Tumor Sample BQR plot

