



ORANGE Report

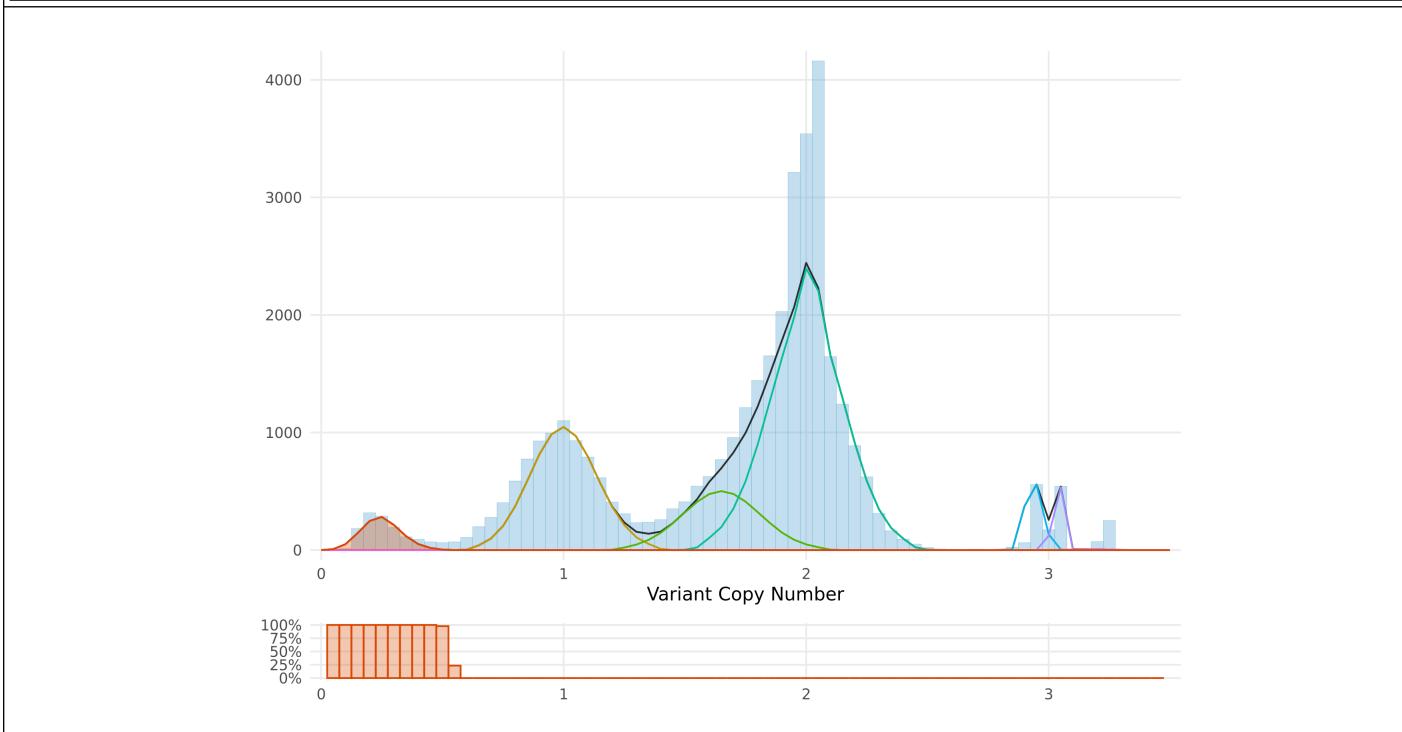
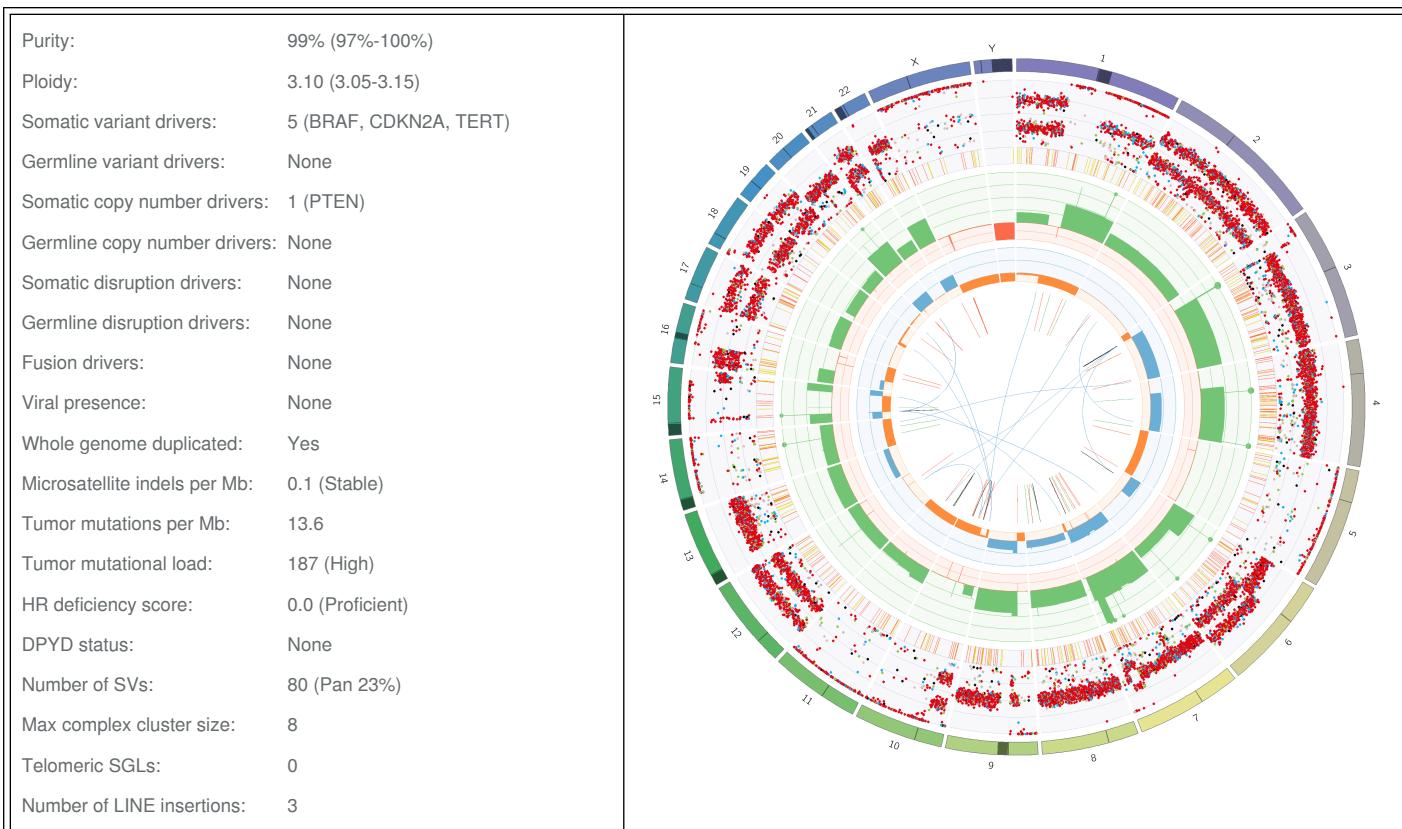
SAMPLE
COLO829v003T

PLATINUM VERSION
5.34 [oncoanalyser]

CONFIGURED PRIMARY TUMOR
cancer (DOID 162)

CUPPA CANCER TYPE
Melanoma (100%)

QC
PASS





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5.34 [oncoanalyser]

Somatic Findings

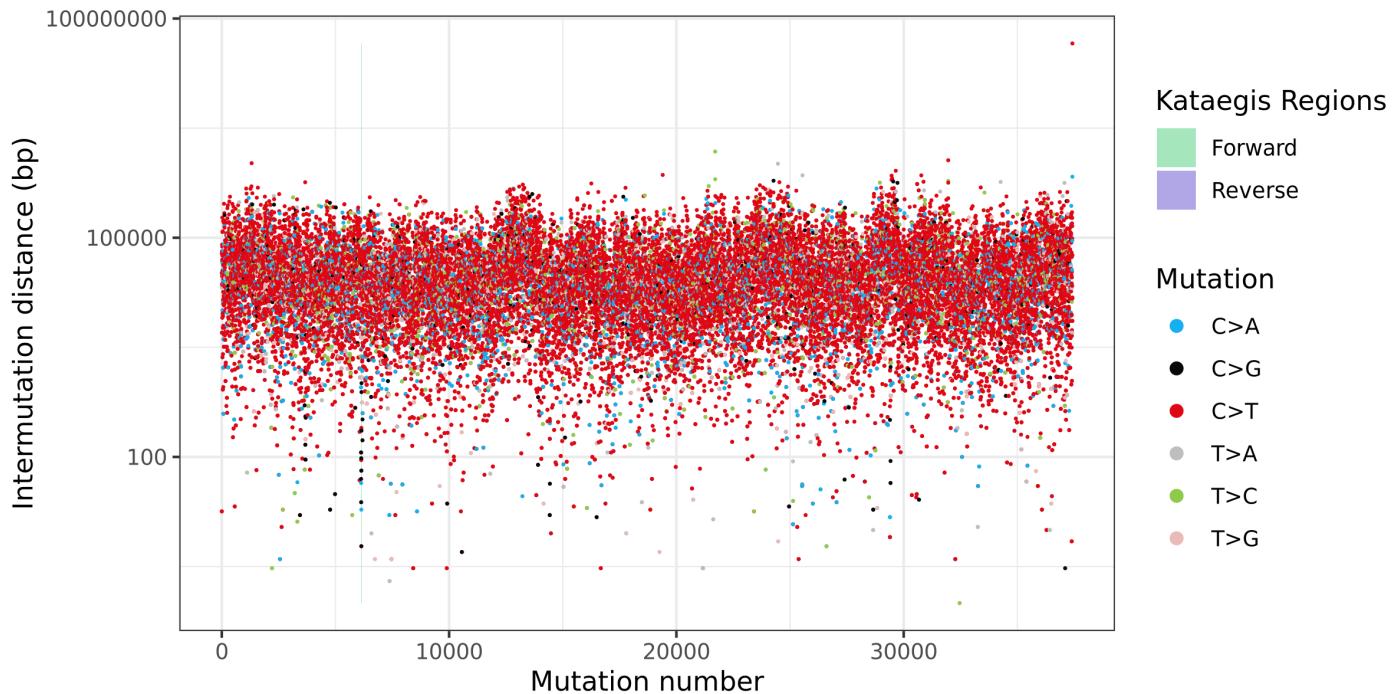
Driver variants (6)

VARIANT	VCN	CN	MACN	BIALLELIC	HOTSPOT	DL	CL	PHASE ID	RNA DEPTH
BRAF p.V640E	4.1	6.0	2.0	No	Yes	100%	100%		5/9 (56%)
CDKN2A p.A68fs	2.0	2.0	0.0	Yes	Near	100%	100%		1350/1354 (100%)
CDKN2A (alt) p.G83fs	2.0	2.0	0.0	Yes	Near	100%	100%		1350/1354 (100%)
TERT c.-125_-124delCCinsTT	1.7	2.0	0.0	Yes	Yes	100%	100%	4725	0/0
SF3B1 p.P718L	2.0	3.0	1.0	No	No	15%	100%		54/79 (68%)
TP63 p.M499I	1.7	4.0	2.0	No	No	0%	100%		0/0

Other potentially relevant variants (3)

VARIANT	VCN	CN	MACN	BIALLELIC	HOTSPOT	DL	CL	PHASE ID	RNA DEPTH
ALB c.1059-52dupT	1.8	3.9	1.9	No	No		100%		0/0
MED12 c.3210-6C>T	2.0	2.0	0.0	Yes	No		100%		1/1 (100%)
STK19 c.-66G>A	2.0	3.8	1.8	No	Yes		100%		0/0

Kataegis plot



Driver amps/dels (1)

LOCATION	GENE	TYPE	CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
chr10q23.31	PTEN	partial loss	0.0	12.9	-1.00	NA	0.03	0.3

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)
chr3p24.2	CFL1P7	full gain	12.1	0.0	-1.00	NA	0.29	NA

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

LOCATION	GENE	TYPE	CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
chr16q21	CNOT1	partial loss	0.0	38.7	-1.00	NA	0.01	0.4

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
chr10q23.31	PTEN	Intron 5 Upstream	DEL	43	1.7	0.0
chr10q23.31	PTEN	Intron 6 Downstream	DEL	43	1.7	0.0

Other potentially interesting gene disruptions (0)

NONE

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

NONE

Signature allocations (11)

SIGNATURE	ALLOCATION	PERCENT
Sig2	3784.6	10%

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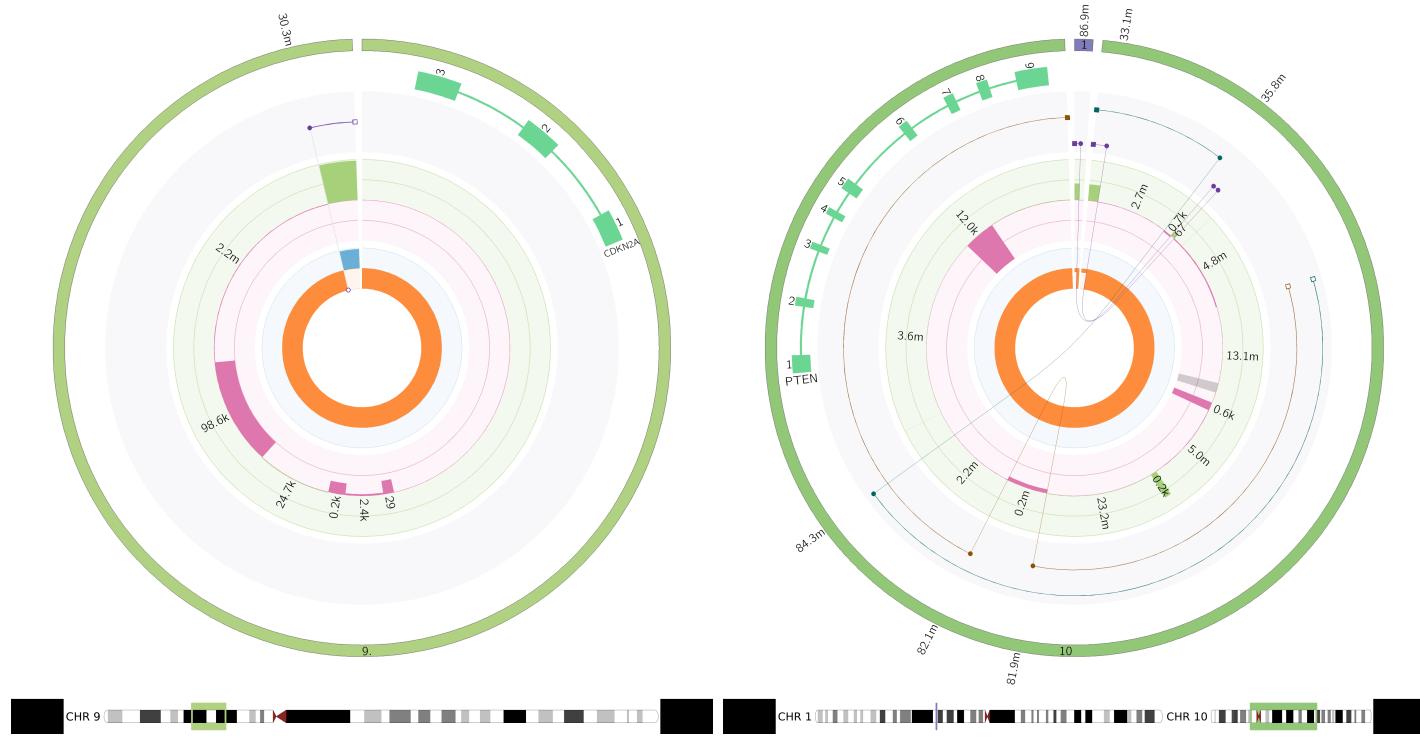


Signature allocations (11)

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SIGNATURE	ALLOCATION	PERCENT
Sig4	2424.6	6%
Sig7	22876.3	61%
Sig8	1622.7	4%
Sig11	1518.8	4%
Sig12	1293.0	3%
Sig17	1081.9	3%
Sig18	2030.4	5%
Sig24	233.6	1%
Sig28	558.1	1%
MISALLOC	4823.5	13%

Structural driver plots (3)





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CHR 10





Germline Findings

Driver variants (0)

NONE

Other potentially relevant variants (4)

VARIANT	VCN	CN	MACN	RNA DEPTH	BIALLELIC	HOTSPOT	GENOTYPE
CYP2D6 c.506-1G>A splice	1.9	4.0	2.0	0/0	No	Yes	HET
CYP3A4 c.522-191C>T	2.4	4.0	2.0	0/0	No	Yes	HET
CYP3A4 upstream	4.0	4.0	2.0	0/0	Yes	Yes	HOM
CYP3A5 c.219-237A>G	4.0	4.0	2.0	0/0	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 (0)

NONE



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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	1609	2672	2.0	None
A*01:01	211	1609	2672	1.8	None
B*08:01	217	753	889	1.8	None
B*40:02	196	726	946	2.0	None
C*03:04	213	771	674	2.0	None
C*07:01	225	772	2040	1.8	None



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RNA Findings

QC	TOTAL FRAGMENTS	NON-DUPLICATE FRAGMENTS	DUPLICATE RATE
PASS	10240202	6511348	36%

Genes with high expression (0)

NONE

Genes with low expression (53)

GENE	TUMOR CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
DOCK8	2.0	0.1	-1.00	NA	0.00	0.0
CCSER1	3.9	0.0	-1.00	NA	0.00	0.0
IKZF1	4.0	0.0	-1.00	NA	0.00	0.0
PTPRN2	2.0	0.0	-1.00	NA	0.00	0.0
CYP2D6	4.0	0.0	-1.00	NA	0.00	0.0
HNF1A	3.0	0.0	-1.00	NA	0.00	0.0
BTK	2.0	0.1	-1.00	NA	0.00	0.0
ZNF521	2.9	0.2	-1.00	NA	0.00	0.0
KMT2C	2.0	47.6	-1.00	NA	0.00	0.2
DLG2	2.9	0.2	-1.00	NA	0.00	0.0
NIPBL	2.0	13.2	-1.00	NA	0.00	0.3
TP63	4.0	0.0	-1.00	NA	0.01	0.0
LINC01001	3.2	0.6	-1.00	NA	0.01	0.1
CASZ1	2.8	0.6	-1.00	NA	0.01	0.0
RAD50	2.0	12.8	-1.00	NA	0.01	0.3
ZRSR2	2.0	5.9	-1.00	NA	0.01	0.3
FOSL2	3.0	5.1	-1.00	NA	0.01	0.1
NSD1	2.0	19.0	-1.00	NA	0.01	0.3
WWOX	2.0	9.1	-1.00	NA	0.01	0.3
DROSHA	2.0	32.7	-1.00	NA	0.01	0.4
EYS	2.9	1.7	-1.00	NA	0.01	0.3
PRDM1	2.9	0.7	-1.00	NA	0.01	0.1
PIK3R1	2.0	9.7	-1.00	NA	0.01	0.3
EPCAM	3.0	0.0	-1.00	NA	0.01	0.0
GRIN2A	3.3	0.0	-1.00	NA	0.01	0.0
CYP3A4	4.0	0.0	-1.00	NA	0.01	0.0
TMPRSS2	3.0	0.0	-1.00	NA	0.01	0.0
CDH1	2.0	0.2	-1.00	NA	0.01	0.0
ZFHX3	2.0	8.4	-1.00	NA	0.01	0.3
KANSL1	3.1	33.1	-1.00	NA	0.01	0.4
FOXO3	2.9	13.4	-1.00	NA	0.02	0.4

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Genes with low expression (53)

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GENE	TUMOR CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
ELF3	3.9	0.2	-1.00	NA	0.02	0.0
PPP2R3B	2.0	5.0	-1.00	NA	0.02	0.3
PTPRD	2.0	0.2	-1.00	NA	0.02	0.0
AFDN	2.9	23.0	-1.00	NA	0.02	0.2
TCF12	2.0	55.1	-1.00	NA	0.02	0.4
LRP1B	3.0	0.0	-1.00	NA	0.02	0.0
FOXQ1	3.9	0.0	-1.00	NA	0.02	0.0
SOX9	3.1	0.4	-1.00	NA	0.02	0.0
MUTYH	2.8	9.7	-1.00	NA	0.03	0.4
SOCS1	3.3	0.5	-1.00	NA	0.03	0.1
PRKN	2.9	0.9	-1.00	NA	0.03	0.2
CEBPA	2.9	1.4	-1.00	NA	0.03	0.1
AGBL4	2.8	0.0	-1.00	NA	0.03	0.0
ATP2B3	2.0	0.0	-1.00	NA	0.03	0.0
PTEN	0.0	12.9	-1.00	NA	0.03	0.3
VHL	2.0	17.2	-1.00	NA	0.04	0.5
CREBBP	3.3	42.7	-1.00	NA	0.04	0.6
ETS2	3.0	11.4	-1.00	NA	0.04	0.2
PTPRB	3.0	3.1	-1.00	NA	0.04	0.3
KMT2D	3.0	43.4	-1.00	NA	0.05	0.5
TCF7L2	2.0	12.0	-1.00	NA	0.05	0.3
MST1R	4.0	0.5	-1.00	NA	0.05	0.0

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



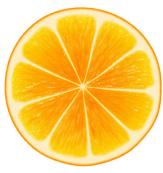
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Cohort Comparison

	Acute myeloid leukemia	Bone/Sot tissue: Other	Breast	Colon/Appendix/small intestine	Esophagus neoplasm	GIST	Glioma	Head and neck: other	Kidney	Kidney-ChRCC	Lemomysarcoma	Liposarcoma	Liver	Lung: NET	Lung: Non-small Cell	Lymphoid tissue	Medulloblastoma	Melanoma	Mesothelium	Myeloproliferative neoplasm	Osteosarcoma	Ovary/Fallopian tube	Pancreas	Pancreas; NET	Pilocytic astrocytoma	Salivary gland/Adenoid cystic	Small intestine/Colon/rectum: NET	Thyroid gland	Uterus: Endometrium	Urothelial tract							
SNV 96 PAIRWISE	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%	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%	98%	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%	98%	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%	100%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%						
EXPRESSION PAIRWISE	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%						
ALT SJ COHORT	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%						
RNA COMBINED	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	95%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%						
COMBINED	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%						
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	31800%	1890%	3850%	97%	1220%	174000%	1850%	2560%	22700%	46800%	1740%	2910%	15600%	9740%	13800%	5660%	11300%	776%	1550%	2300%	39000%	33%	7800%	398000%	7930%	4200%	3470%	9050%	19900%	9580%	98%	66%	14000%	22100%	730%	6020%	
SIG 7 UV (23805)	50%	21%	36%	43%	38%	44%	32%	39%	40%	50%	32%	43%	48%	42%	48%	43%	35%	36%	48%	42%	44%	46%	46%	38%	44%	45%	28%	43%	38%	44%	48%	32%	36%	39%	30%		
SIG 6 MMR (0)	733%	102%	127%	701%	145%	10000%	99%	105%	1710%	10000%	94%	146%	10500%	99%	22500%	97%	98%	40%	10%	10000%	5960%	100%	196%	10000%	267%	119%	99%	119%	10000%	10000%	182%	98%	100%	10000%	379%	95%	445%
SIG 2 13 AID APOBC (3952)	20600%	57%	97%	97%	78%	23500%	99%	95%	343%	99%	62%	99%	732%	250%	309%	149%	1140%	61%	94%	100%	3380%	34%	377%	16300%	98%	97%	100%	98%	43400%	100%	93%	57%	838%	98%	33%	96%	
SIG 17 (1125)	1300%	98%	95%	98%	94%	21600%	58%	27%	98%	10000%	97%	100%	98%	206%	368%	100%	10000%	94%	97%	89%	10500%	98%	1550%	40200%	94%	100%	94%	679%	17300%	99%	94%	93%	1440%	355%	92%	99%	
SIG 11 (1586)	2840%	100%	100%	96%	119%	6370%	99%	114%	645%	95%	100%	399%	1480%	370%	460%	288%	461%	100%	153%	100%	10900%	88%	514%	1600%	388%	178%	191%	98%	17600%	343%	98%	58%	602%	266%	99%	235%	
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%	27%	40%	36%	34%	27%	42%	36%	26%	36%	4%		
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%	3%	22%	-10000%	2%	4%	6%		
PERCENTILES	1460%	93%	95%	96%	95%	2610%	85%	80%	271%	95%	90%	99%	467%	100%	322%	99%	381%	60%	36%	98%	1120%	37%	223%	2100%	209%	99%	99%	5150%	96%	97%	60%	557%	109%	81%	84%		
SNV COUNT (37424)	469%	16%	29%	72%	48%	72%	1%	7%	89%	50%	32%	77%	166%	82%	93%	66%	135%	19%	3%	63%	92%	46%	94%	729%	83%	64%	20%	90%	1770%	29%	57%	36%	98%	96%	15%	22%	
MS INDELS TMB (0.09)	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%	
TELOMERIC SGL (0)	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%	80%	
SIMPLE DUP 32B 200B (3)	91%	13%	34%	74%	40%	30%	150%	2%	6%	8%	17%	76%	74%	77%	46%	62%	87%	23%	35%	92%	93%	72%	86%	88%	65%	29%	24%	70%	98%	13%	23%	24%	87%	78%	7%	20%	
MAX COMPLEX SIZE (8)	93%	24%	14%	40%	9%	67%	31%	8%	51%	16%	26%	56%	90%	14%	18%	48%	38%	10%	29%	76%	80%	24%	9%	98%	8%	22%	20%	73%	7%	36%	38%	77%	83%	16%	43%		
LIFE SIZE (3)	91%	13%	34%	74%	40%	30%	150%	2%	6%	8%	17%	76%	74%	77%	46%	62%	87%	23%	35%	92%	93%	72%	86%	88%	65%	29%	24%	70%	98%	13%	23%	24%	87%	78%	7%	20%	

FEATURES



ORANGE Report

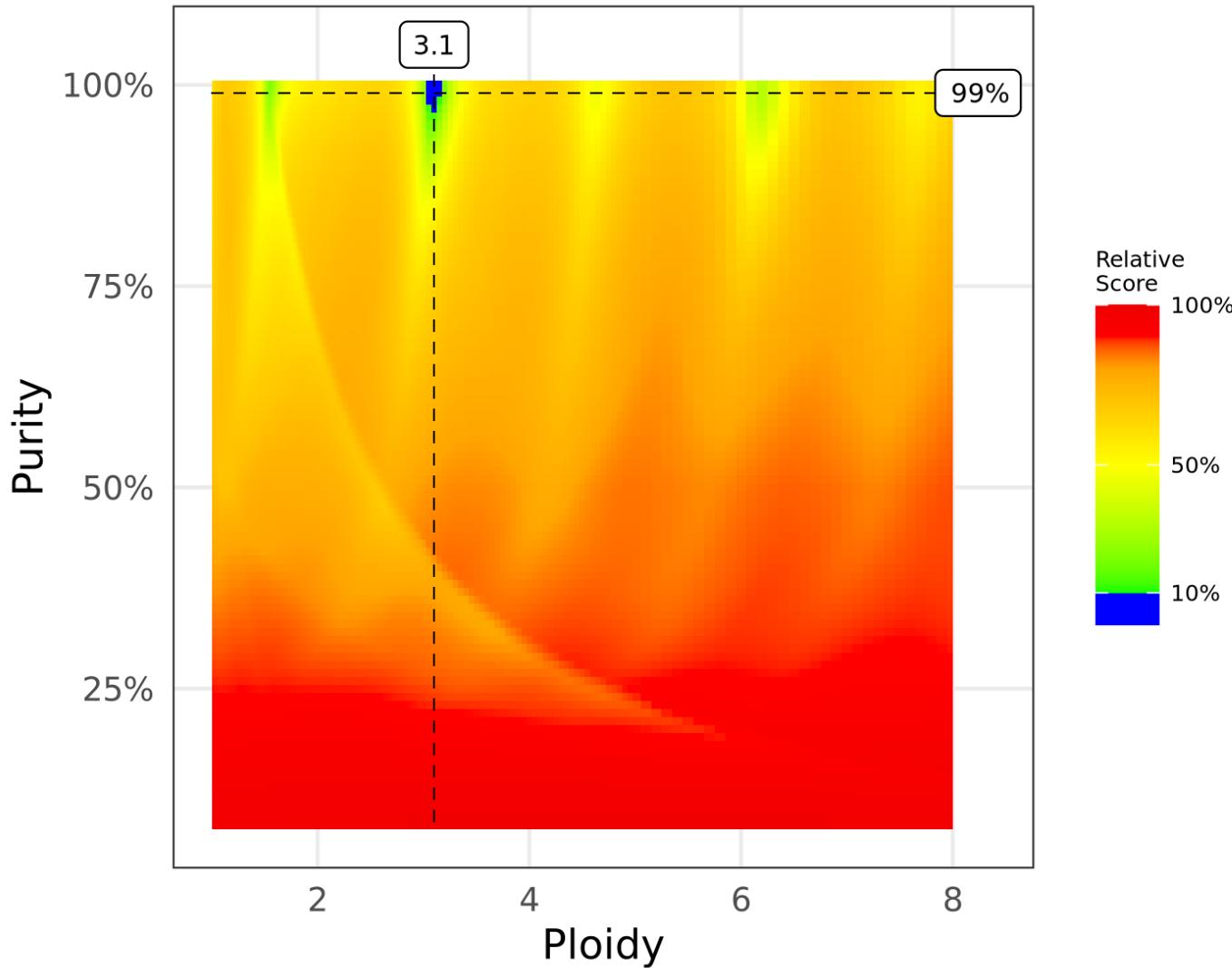
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Quality Control

QC	REF GENOME	FIT METHOD	MEAN DEPTH	CONTAMINATION	UNS. SEGMENTS	DELETED GENES
PASS	V38	NORMAL	111	0%	0	4

Purity/Ploidy Scores



Flagstats

	UNIQUE RC	SECONDARY RC	SUPPLEMENTARY RC	MAPPED PROPORTION
Ref Sample	806150780	0	718342	97%
Tumor Sample	2992264274	0	2574171	97%



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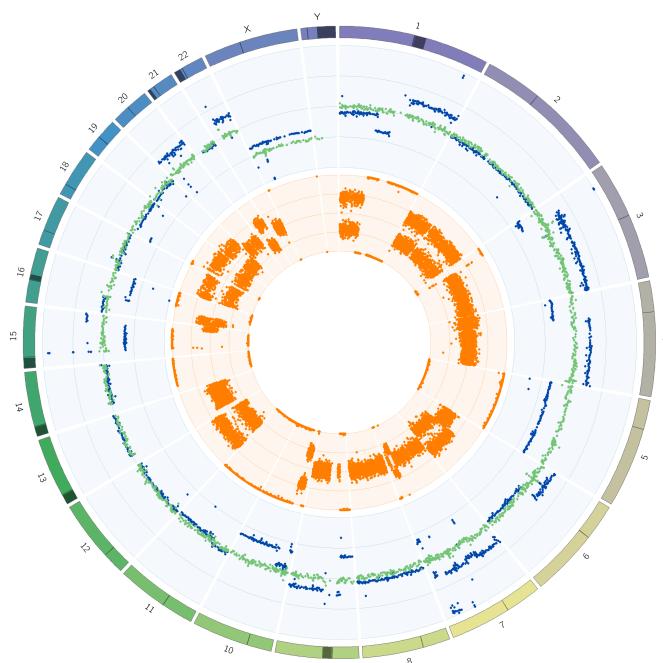
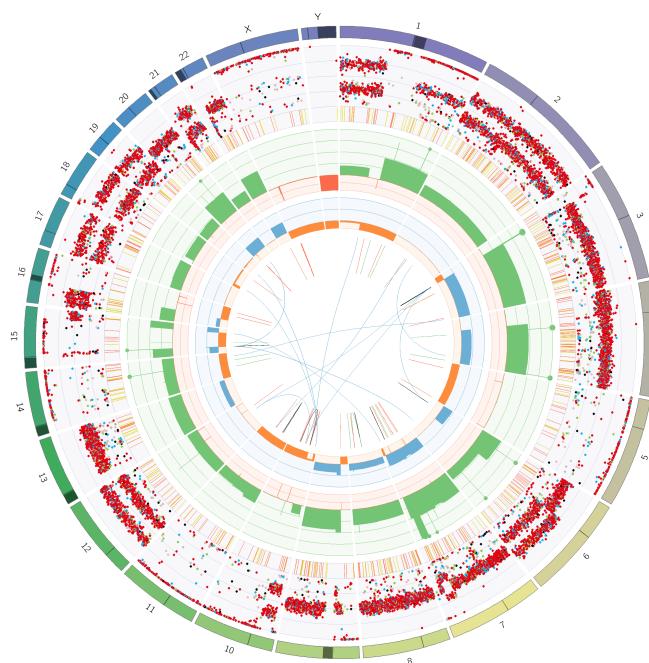
Coverage Stats

	MEAN COVERAGE	SD COVERAGE	MEDIAN COVERAGE	MAD COVERAGE
Ref Sample	27.4	8.8	28	5
Tumor Sample	89.8	31.7	92	20

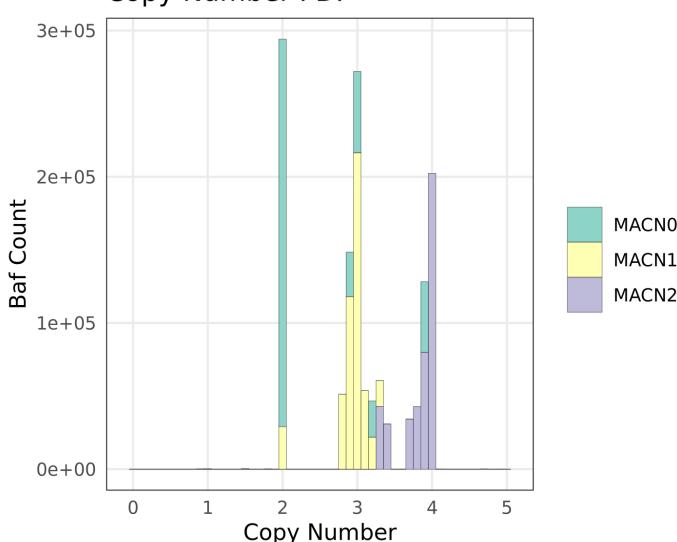
Excluded Percentages

	ADAPTER	BASEQ	CAPPED	DUPE	MAPQ	OVERLAP	UNPAIRED	TOTAL
Ref Sample	0%	0%	0%	20%	3%	0%	0%	24%
Tumor Sample	0%	0%	0%	27%	3%	1%	0%	31%

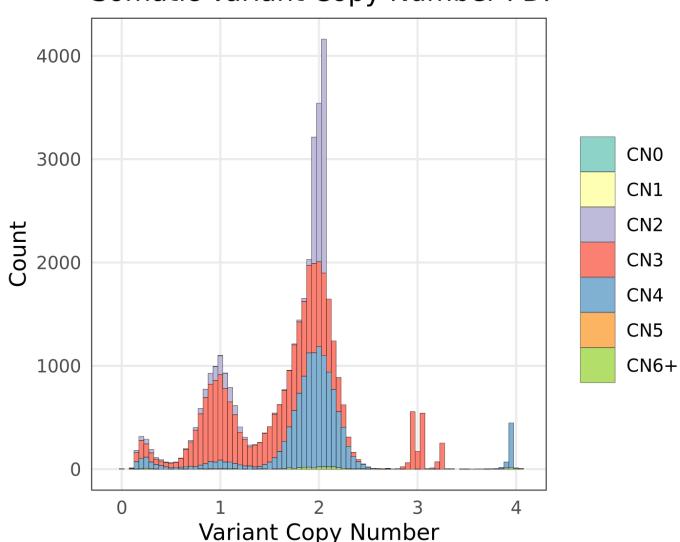
QC plots



Copy Number PDF



Somatic Variant Copy Number PDF



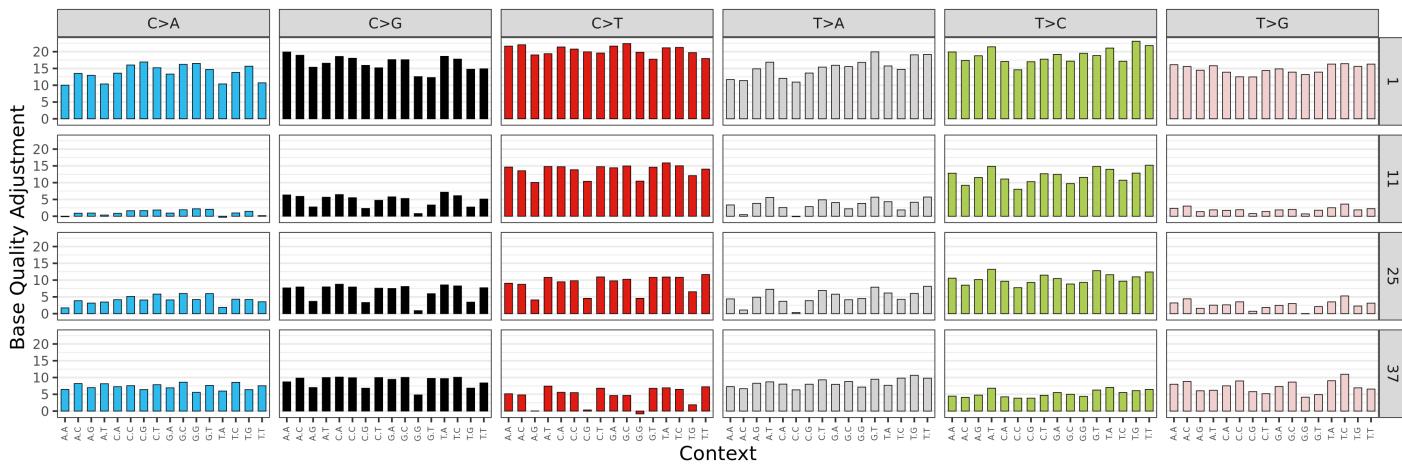


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Reference Sample BQR plot



Tumor Sample BQR plot

