



ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.34

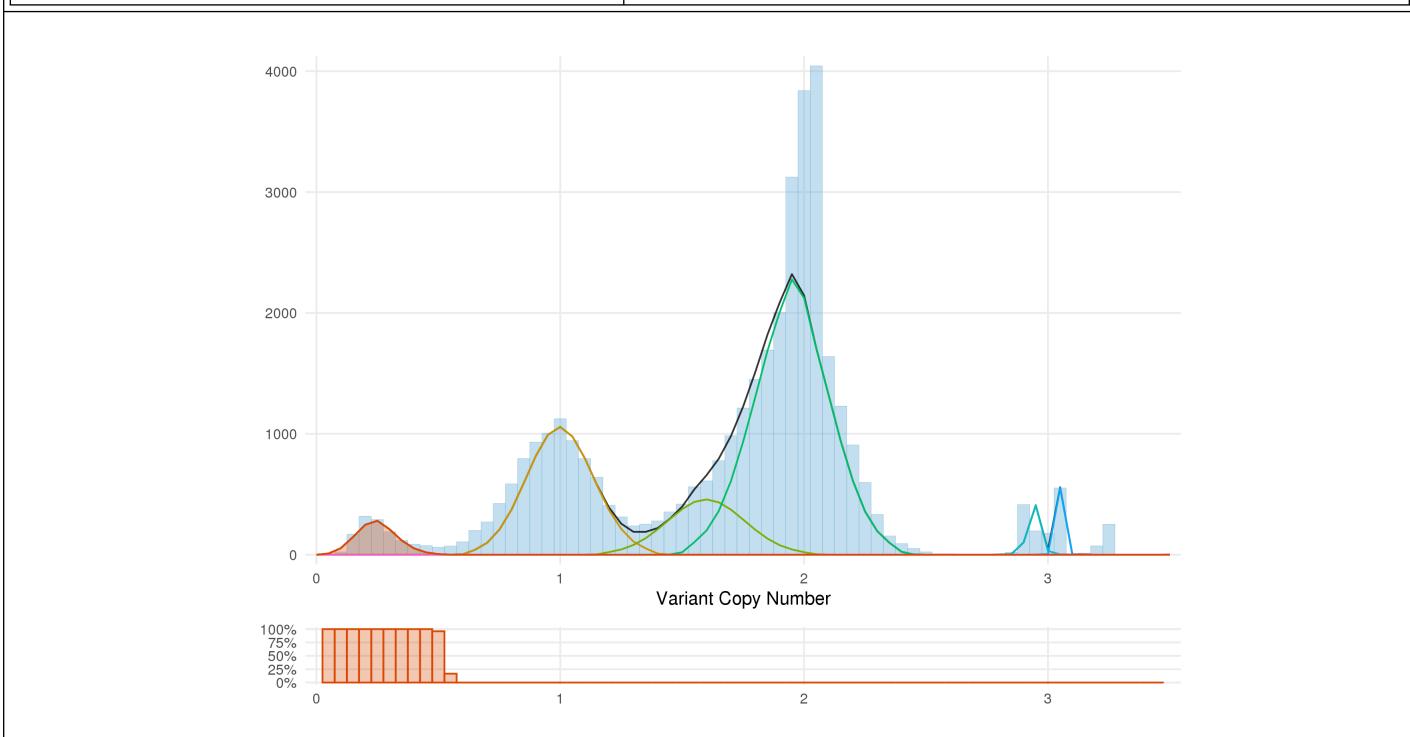
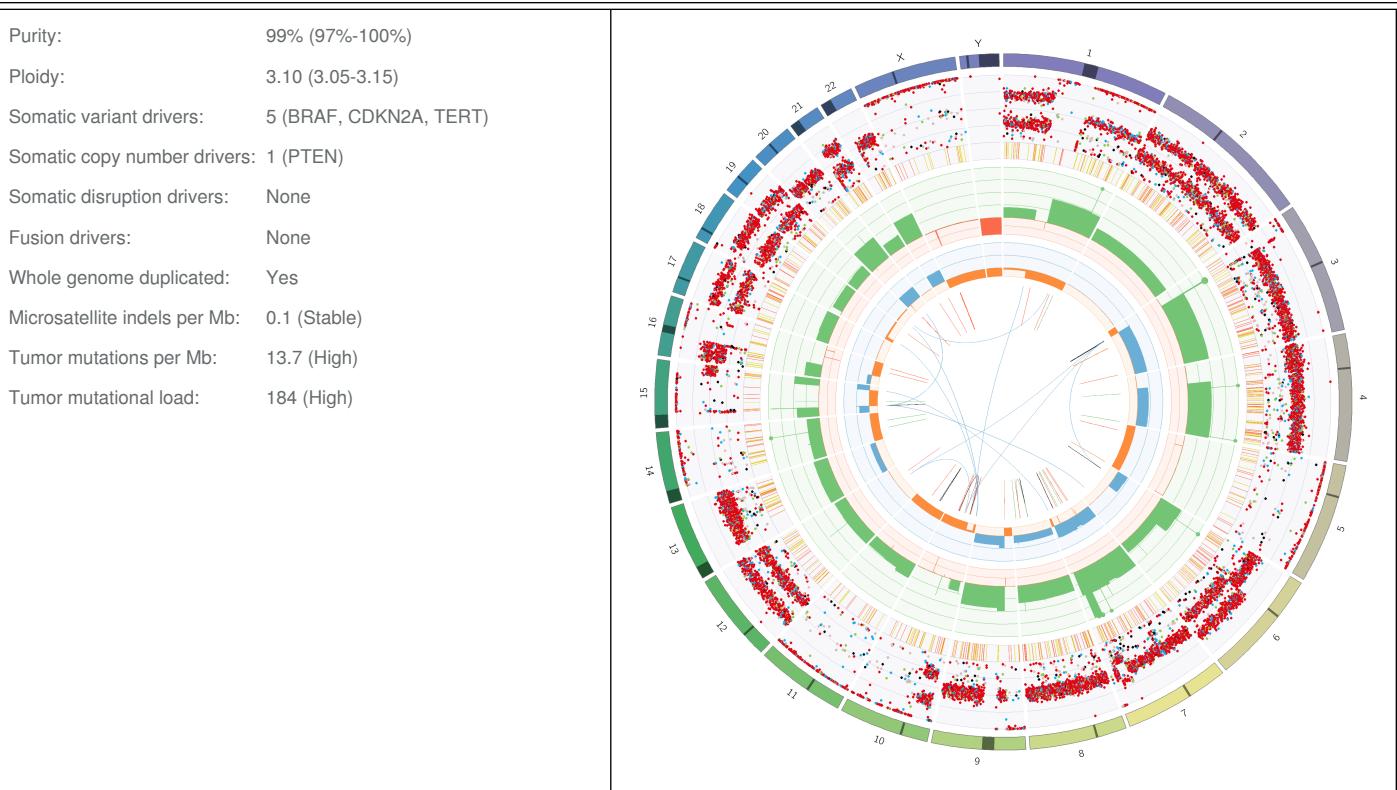
CONFIGURED PRIMARY TUMOR

skin melanoma (DOID 8923)

TUMOR-ONLY

QC

PASS





Somatic Findings

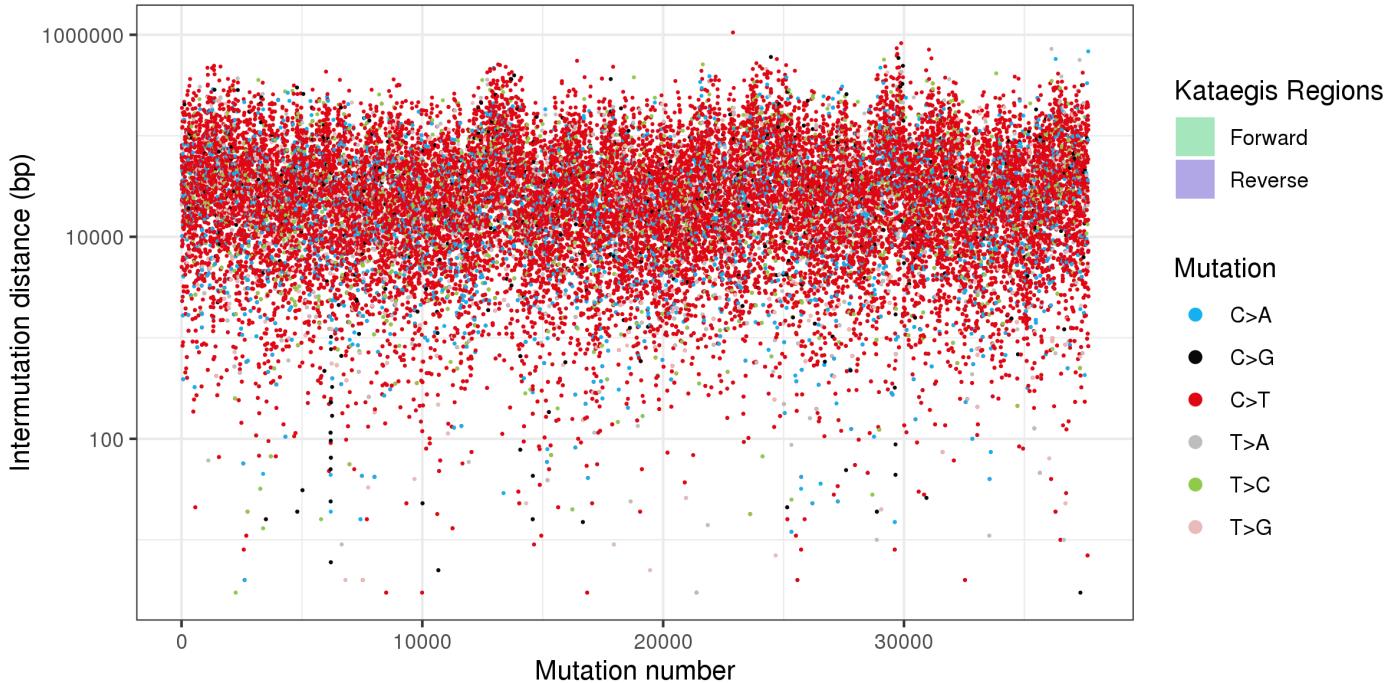
Driver variants (6)

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%	4725	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.1059-52dupT	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



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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	12.1	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	0.1	NA	NA	NA	NA	NA

Driver fusions (0)

NONE

Other potentially interesting fusions (0)

NONE

Potentially interesting in-frame fusions in case no high drivers detected

High driver likelihood events are detected in this sample, therefore this section is empty

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 (0)

NONE

Structural driver plots (3)



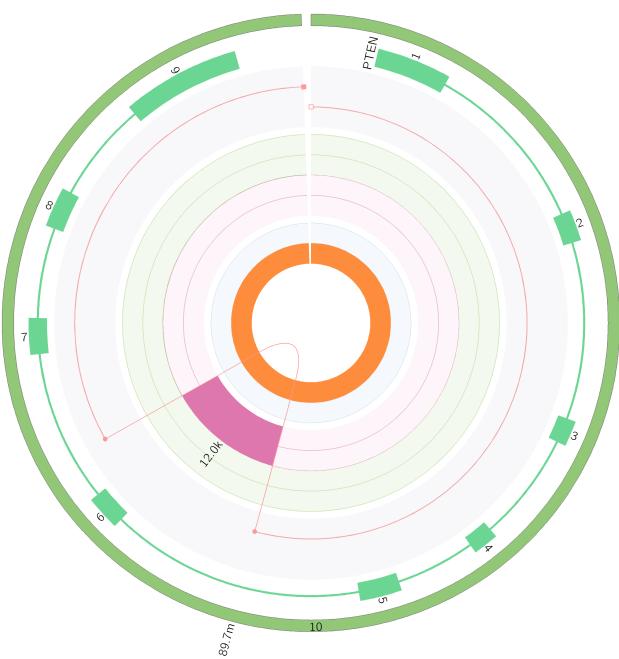
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Immunology

HLA QC

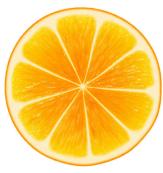
QC Status: PASS

HLA Alleles (6)

ALLELE	REF FRAGS	TUMOR FRAGS	RNA FRAGS	TUMOR CN	SOMATIC #MUTATIONS
A*01:01	NA	1602	NA	2.0	NONE
A*01:01	NA	1602	NA	1.8	NONE
B*08:01	NA	750	NA	1.8	NONE
B*40:02	NA	721	NA	2.0	NONE
C*03:04	NA	768	NA	2.0	NONE
C*07:01	NA	767	NA	1.8	NONE

Genetic Immune Escape

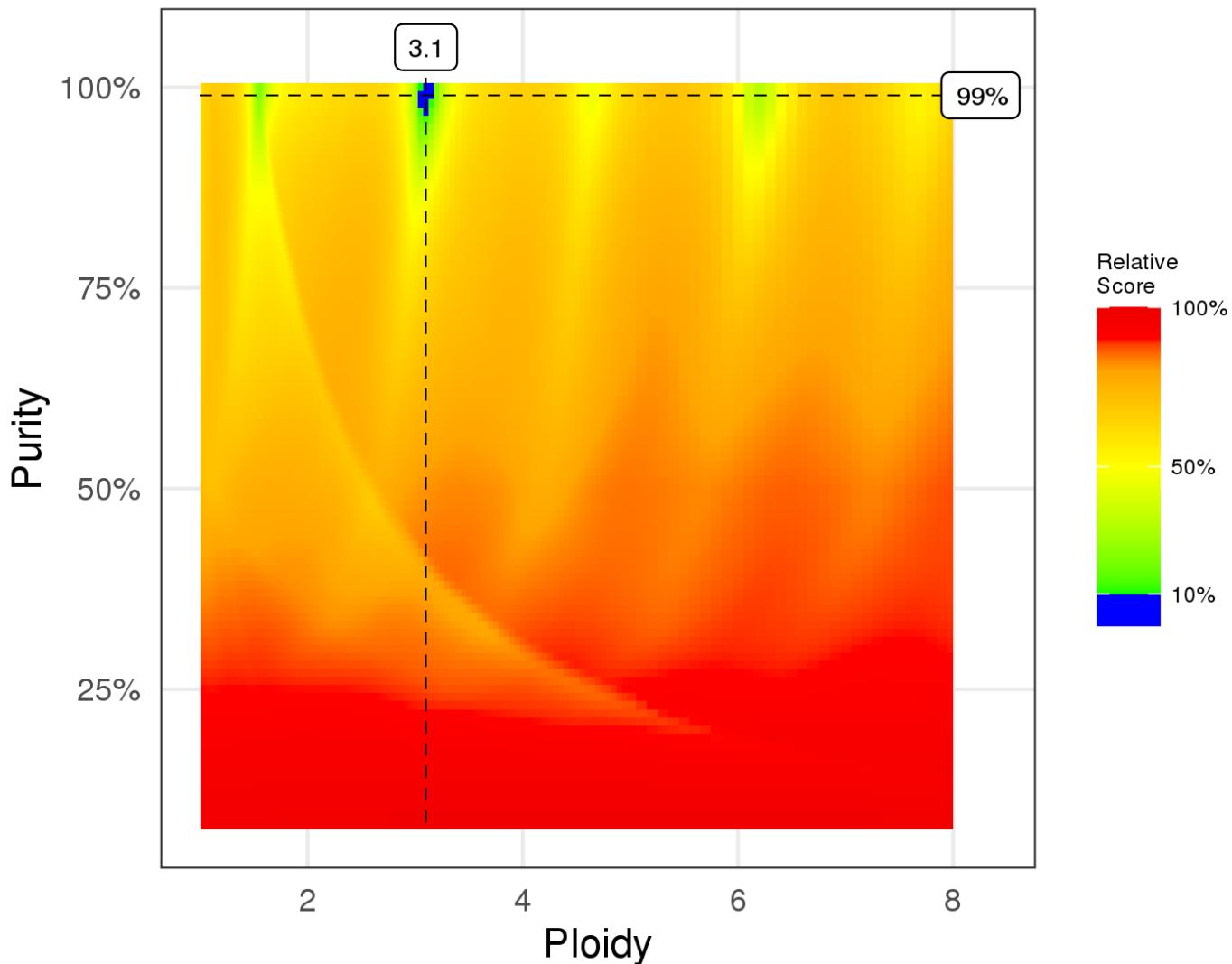
ESCAPE MECHANISM	DETECTED?
HLA-1 loss-of-function	No
Antigen presentation pathway inactivation	No
IFN gamma pathway inactivation	No
(Potential) PD-L1 overexpression	No
CD58 inactivation	No
Epigenetics driven immune escape via SETDB1	No



Quality Control

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

Purity/Ploidy Scores



Flagstats

	UNIQUE RC	SECONDARY RC	SUPPLEMENTARY RC	MAPPED PROPORTION
Tumor Sample	2671674230	0	21477075	100%

Coverage Stats

	MEAN COVERAGE	SD COVERAGE	MEDIAN COVERAGE	MAD COVERAGE
Tumor Sample	108.0	35.0	109	24



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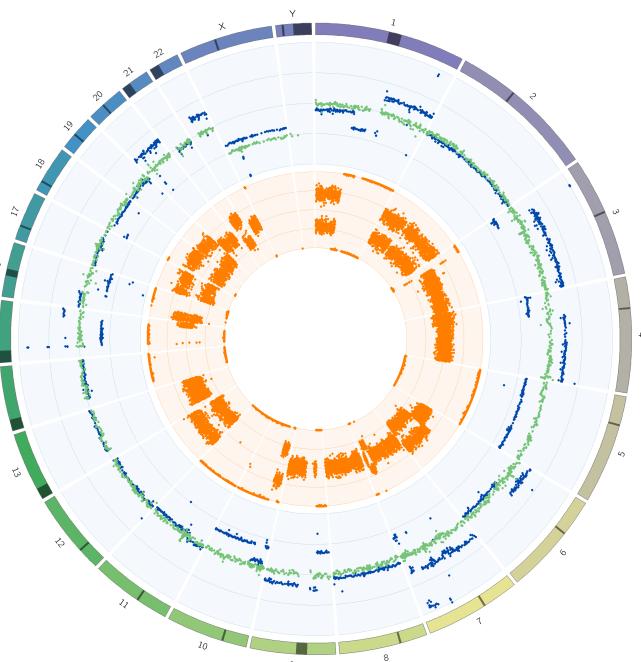
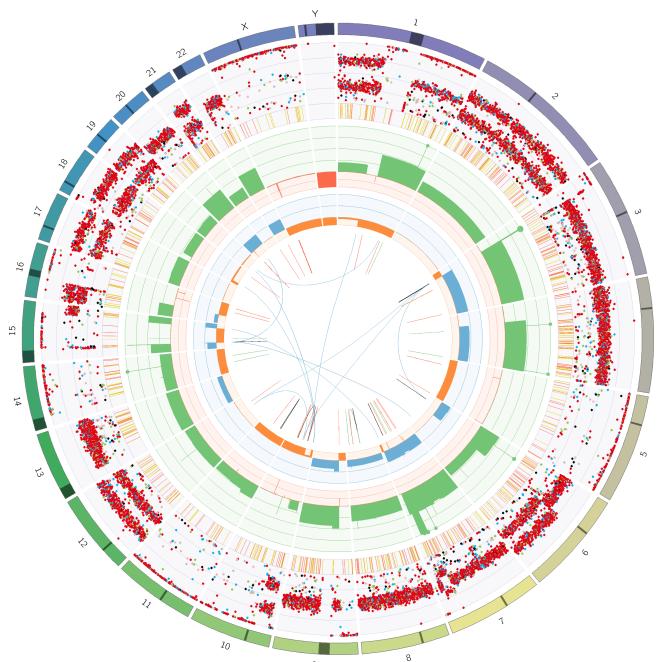
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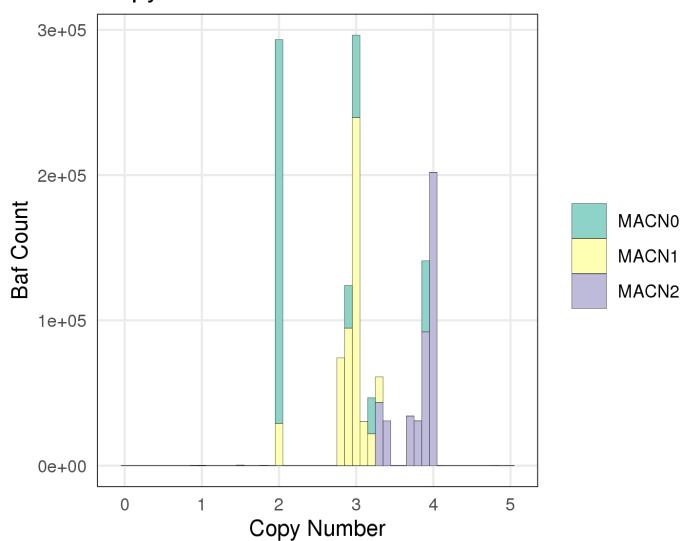
Excluded Percentages

	ADAPTER	BASEQ	CAPPED	DUPE	MAPQ	OVERLAP	UNPAIRED	TOTAL
Tumor Sample	0%	0%	1%	15%	5%	1%	0%	22%

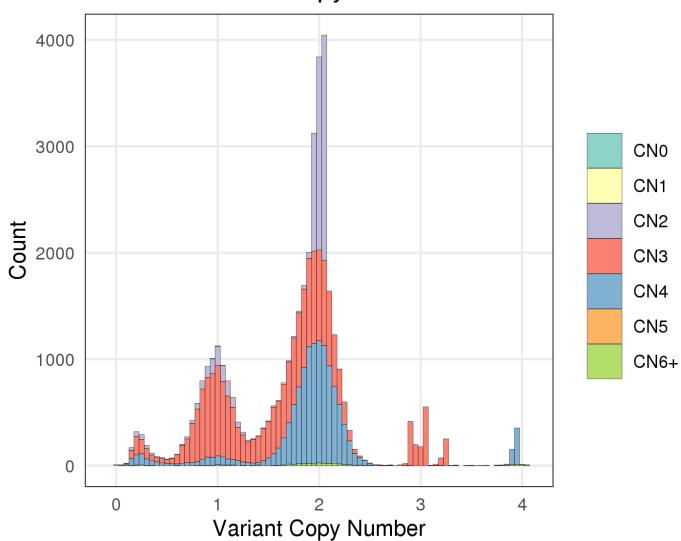
QC plots



Copy Number PDF



Somatic Variant Copy Number PDF



Tumor Sample BQR plot



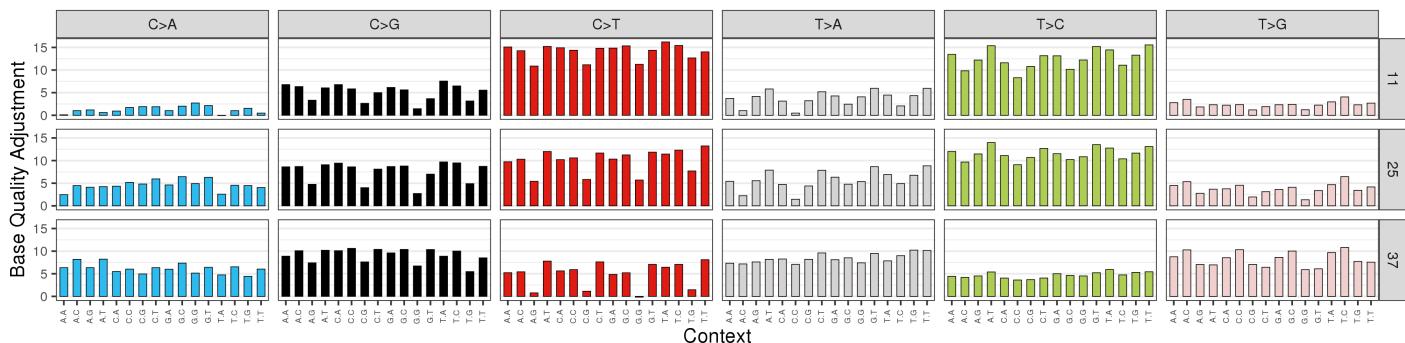
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Tumor detection requirements

At least one requirement needs to be met to indicate detection of tumor, in case of highly diploid candidate solutions (proportion >=0.97)

Tumor maximum diploid proportion	0.02
Number of hotspot mutations	3
Number of hotspot structural variants	0
Sum of small variants allele read counts	2273196
Sum of structural variant tumor fragment counts (excluding single breakends)	4908
Sum of B-allele frequency points in germline diploid regions regions with tumor ratio < 0.8 OR > 1.2	675344