



ORANGE Report

SAMPLE

Test

PLATINUM VERSION

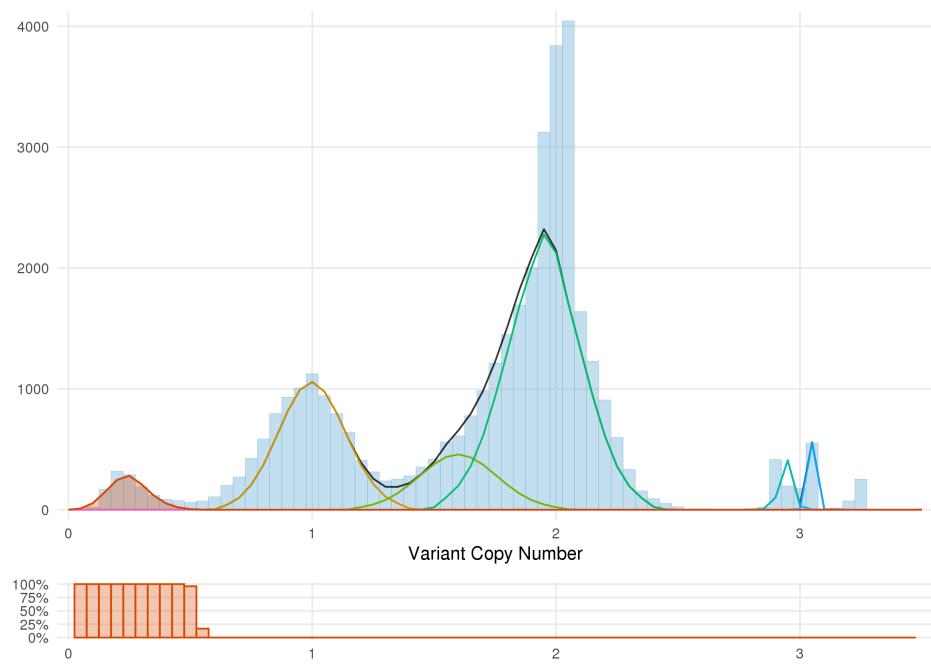
5.34

CONFIGURED PRIMARY TUMOR
skin melanoma (DOID 8923)

CUPPA CANCER TYPE
Skin: Melanoma (100%)

QC
PASS

Purity:	99% (97%-100%)
Ploidy:	3.10 (3.05-3.15)
Somatic variant drivers:	1
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 (High)
Tumor mutational load:	184 (High)
HR deficiency score:	0.0 (Proficient)
DPYD status:	*1 HOM (Normal Function)
UGT1A1 status:	*1 HOM (Normal Function)
Number of SVs:	75 (Pan 0.22 Skin 0.34)
Max complex cluster size:	8
Telomeric SGLs:	0
Number of LINE insertions:	3





ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.34

Somatic Findings

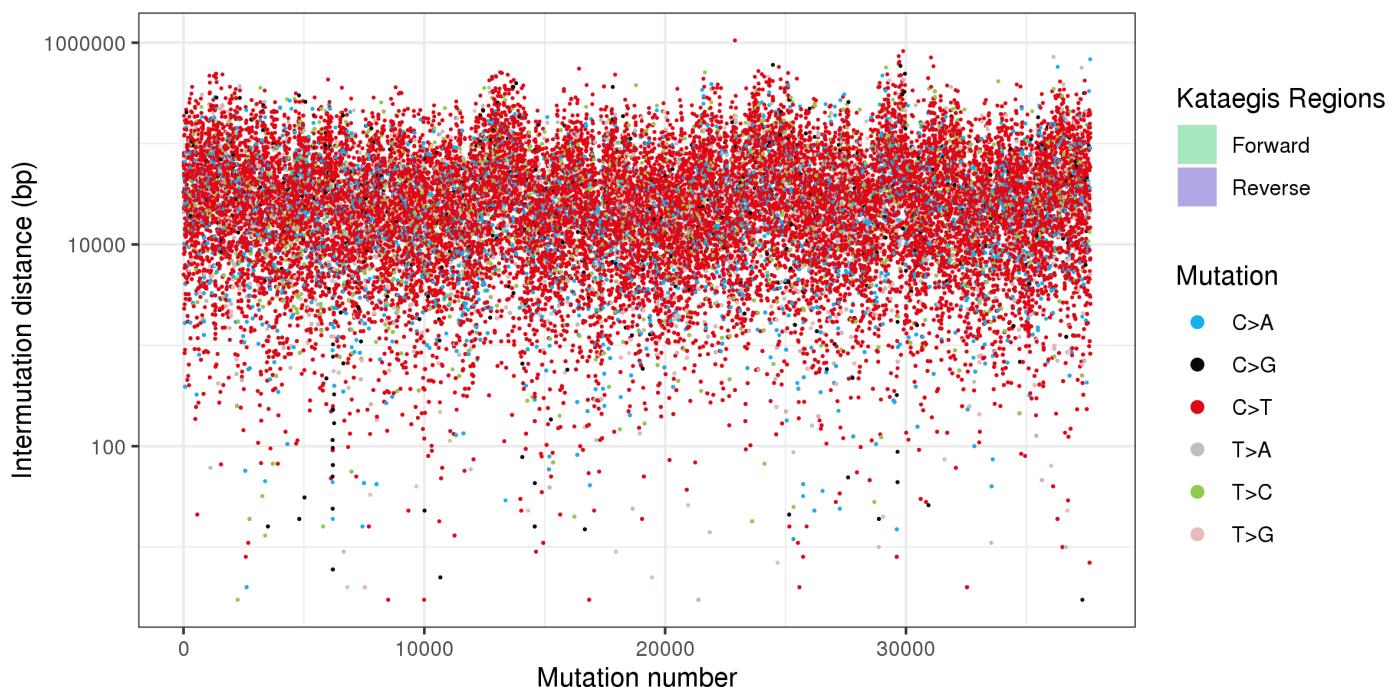
Driver variants (1)

VARIANT	VCN	CN	MACN	BIALLELIC	HOTSPOT	DL	CL	PHASE ID	RNA DEPTH
SF3B1 p.P718L	2.0	3.0	1.0	No	No		100%		NA

Other potentially relevant variants (1)

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

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



ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.34

Regions with deletions in genes in other autosomal regions (0)

NONE

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 (1)

LOCATION	GENE	RANGE	TYPE	CLUSTER ID	JUNCTION CN	UNDISRUPTED CN
10q23.31	PTEN	Intron 5 Upstream	DEL	68	2.0	0.0

Other potentially interesting gene disruptions (0)

NONE

Potentially interesting LOH events (0)

NONE

Signature allocations (12)

SIGNATURE	ALLOCATION	PERCENT
Sig2	3702.7	10%
Sig4	2038.8	5%
Sig7	23286.3	62%
Sig8	1984.8	5%
Sig11	1313.3	3%
Sig12	1137.1	3%
Sig17	1083.2	3%
Sig18	2057.3	5%

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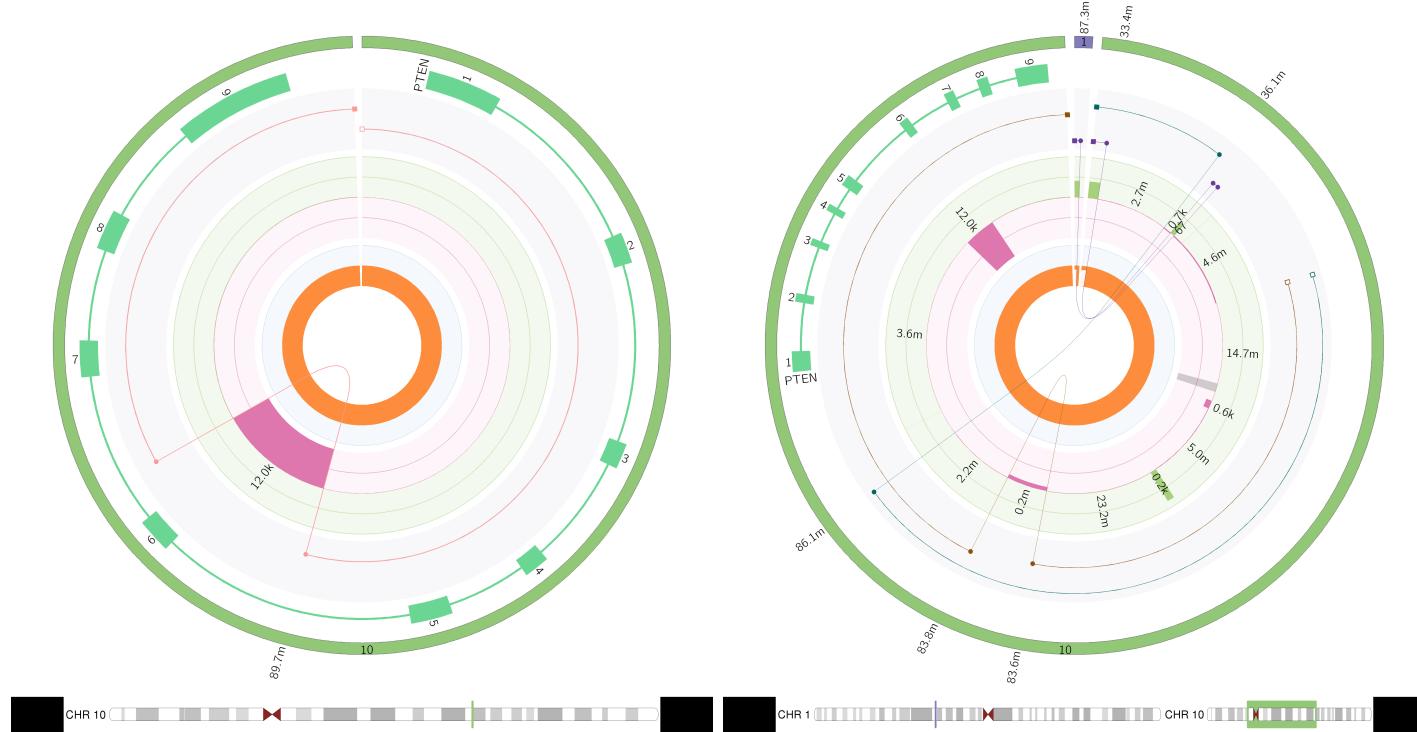


Signature allocations (12)

CONTINUED FROM THE PREVIOUS PAGE

SIGNATURE	ALLOCATION	PERCENT
Sig20	253.1	1%
Sig24	289.0	1%
Sig28	514.4	1%
MISALLOC	4611.5	12%

Structural driver plots (3)





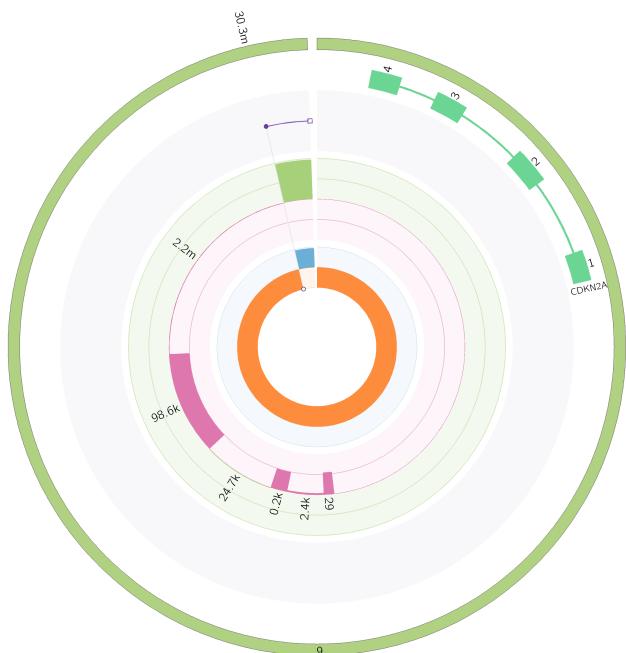
ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.34





ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.34

Germline Findings

Driver variants (0)

NONE

Other potentially relevant variants (1)

VARIANT	VCN	CN	MACN	RNA DEPTH	BIALLELIC	HOTSPOT	GENOTYPE
CYP3A4 c.522-191C>T	2.4	4.0	2.0	NA	No	Yes	HET

Potentially pathogenic germline deletions (0)

NONE

Potentially pathogenic germline LOH events (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	HAPLOTYPE	GENOTYPE	FUNCTION	LINKED DRUGS	SOURCE
DPYD	*1	HOM	Normal Function	5-Fluorouracil;Capecitabine;Tegafur	PHARMGKB
UGT1A1	*1	HOM	Normal Function	Irinotecan	PHARMGKB



ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.34

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	NA	2.0	NONE
A*01:01	211	1602	NA	1.8	NONE
B*08:01	216	750	NA	1.8	NONE
B*40:02	196	721	NA	2.0	NONE
C*03:04	213	768	NA	2.0	NONE
C*07:01	225	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



ORANGE Report

SAMPLE

Test

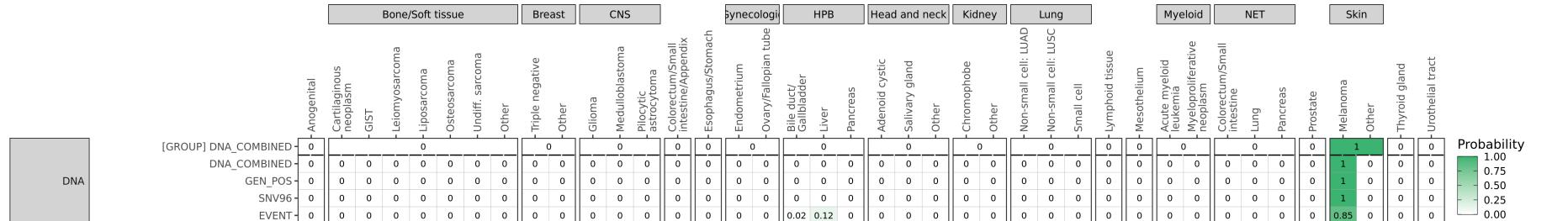
PLATINUM VERSION

5.34

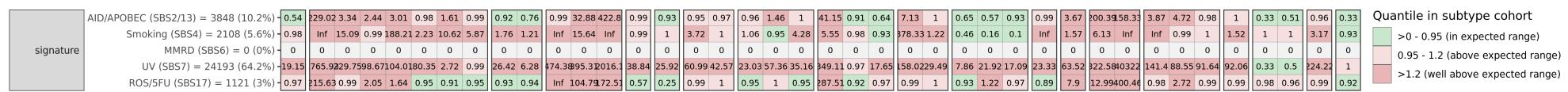
Cohort Comparison

Probabilities by classifier

Cancer group (strips) and subtype (label)

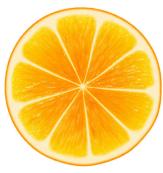


SNV96: Mutational signatures



EVENT: Feature contributions





ORANGE Report

SAMPLE

Test

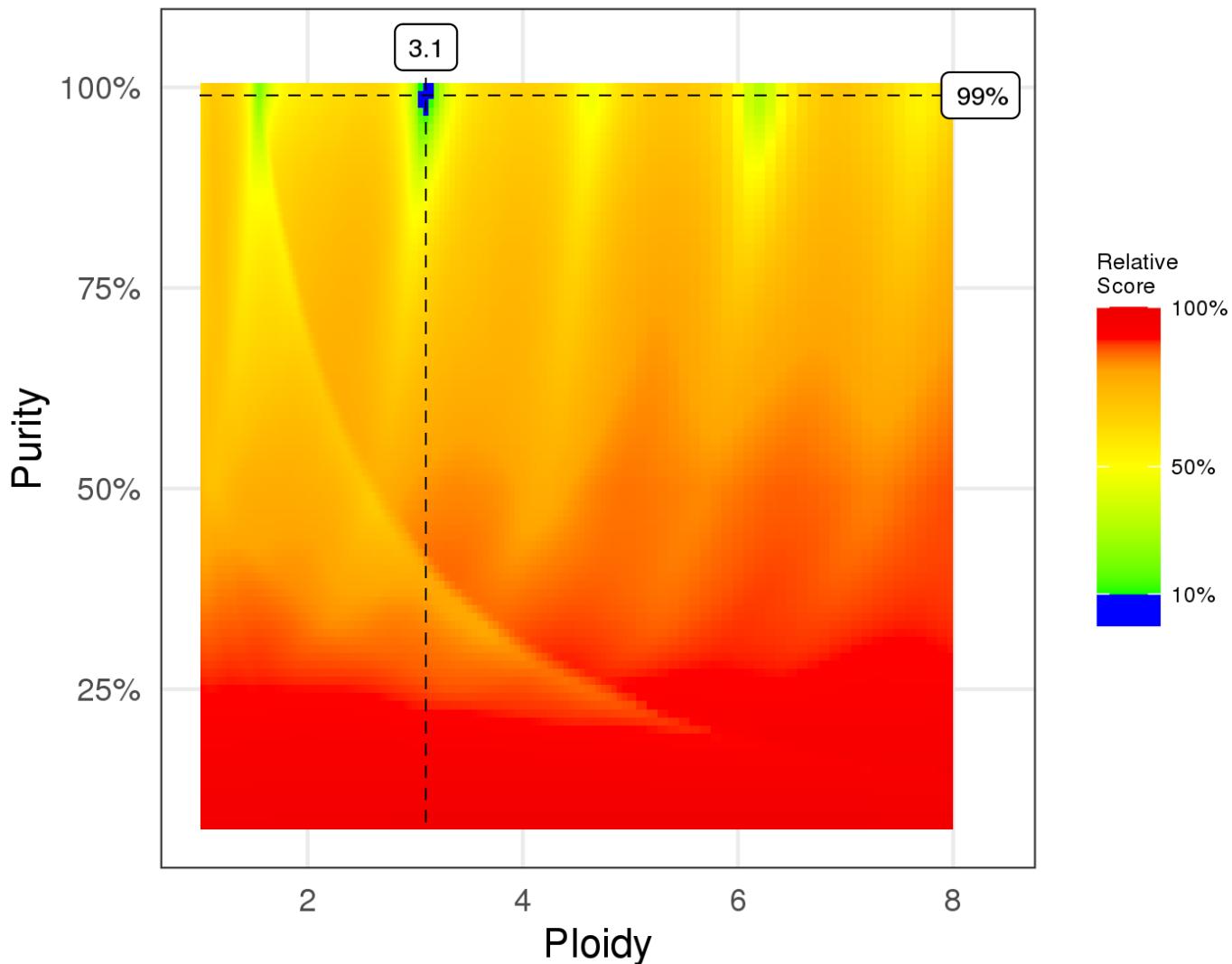
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5.34

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
Ref Sample	740406212	0	5742696	99%
Tumor Sample	2671674230	0	21477075	100%



ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.34

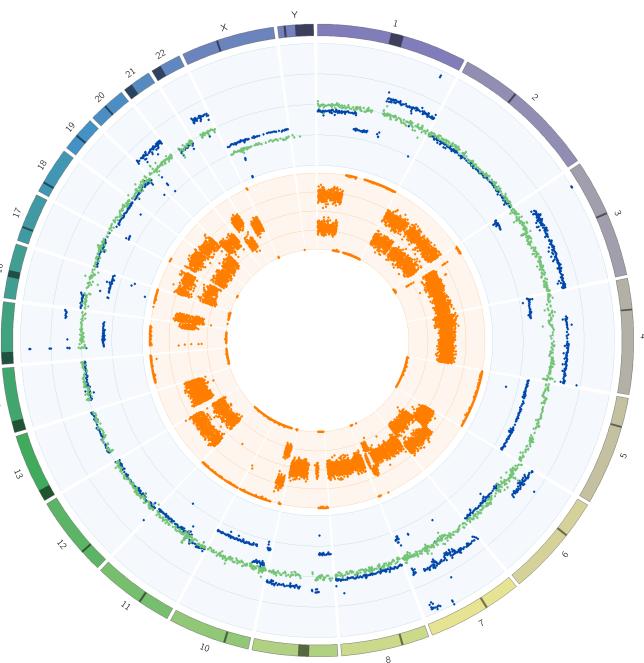
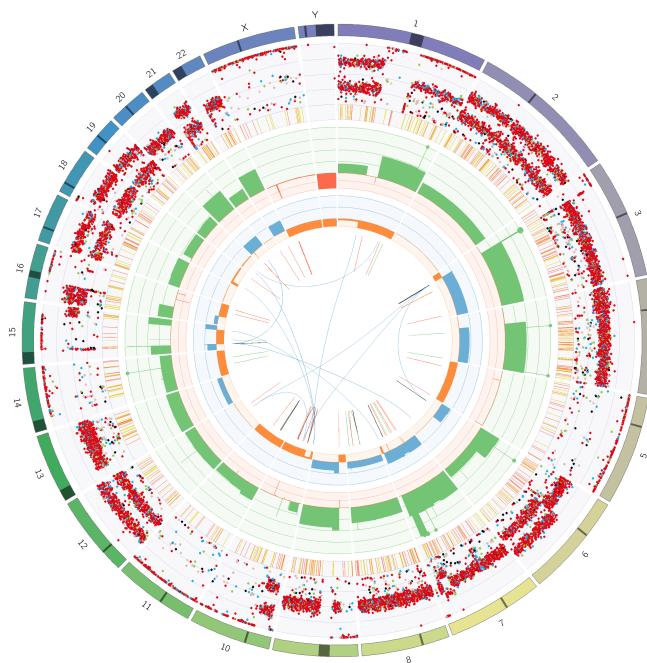
Coverage Stats

	MEAN COVERAGE	SD COVERAGE	MEDIAN COVERAGE	MAD COVERAGE
Ref Sample	31.4	10.1	32	5
Tumor Sample	108.0	35.0	109	24

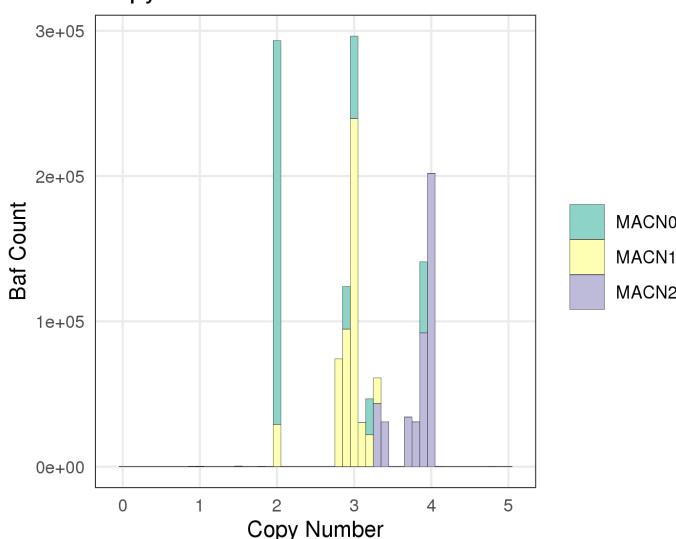
Excluded Percentages

	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%

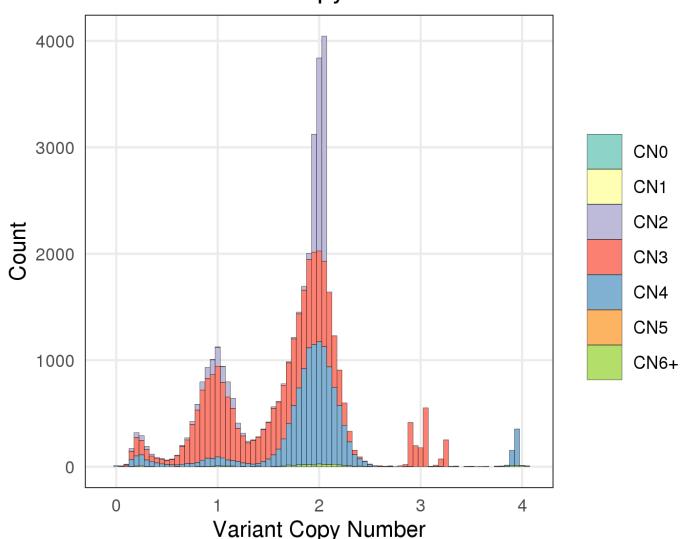
QC plots



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Somatic Variant Copy Number PDF





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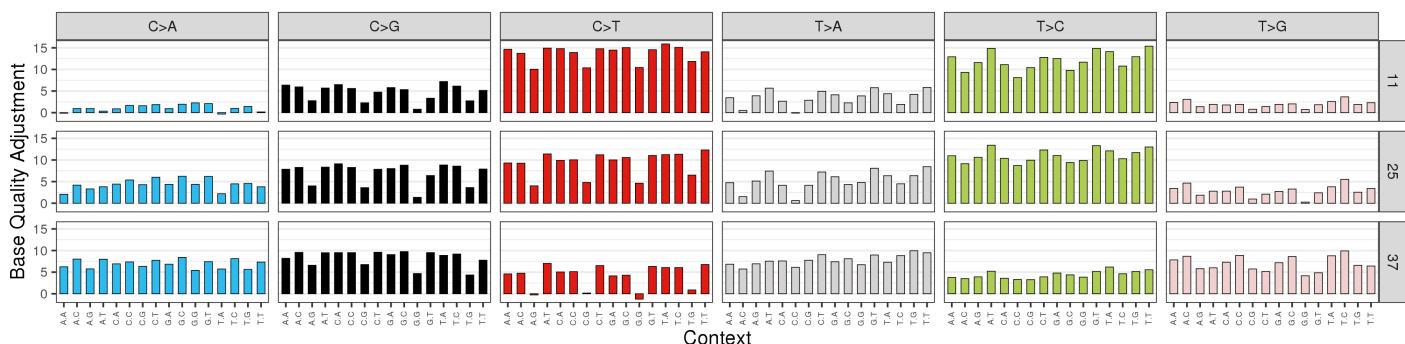
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Test

PLATINUM VERSION

5.34

Reference Sample BQR plot



Tumor Sample BQR plot

