



# ORANGE Report (Research Use Only)

SAMPLE

Test

PIPELINE VERSION

6.0

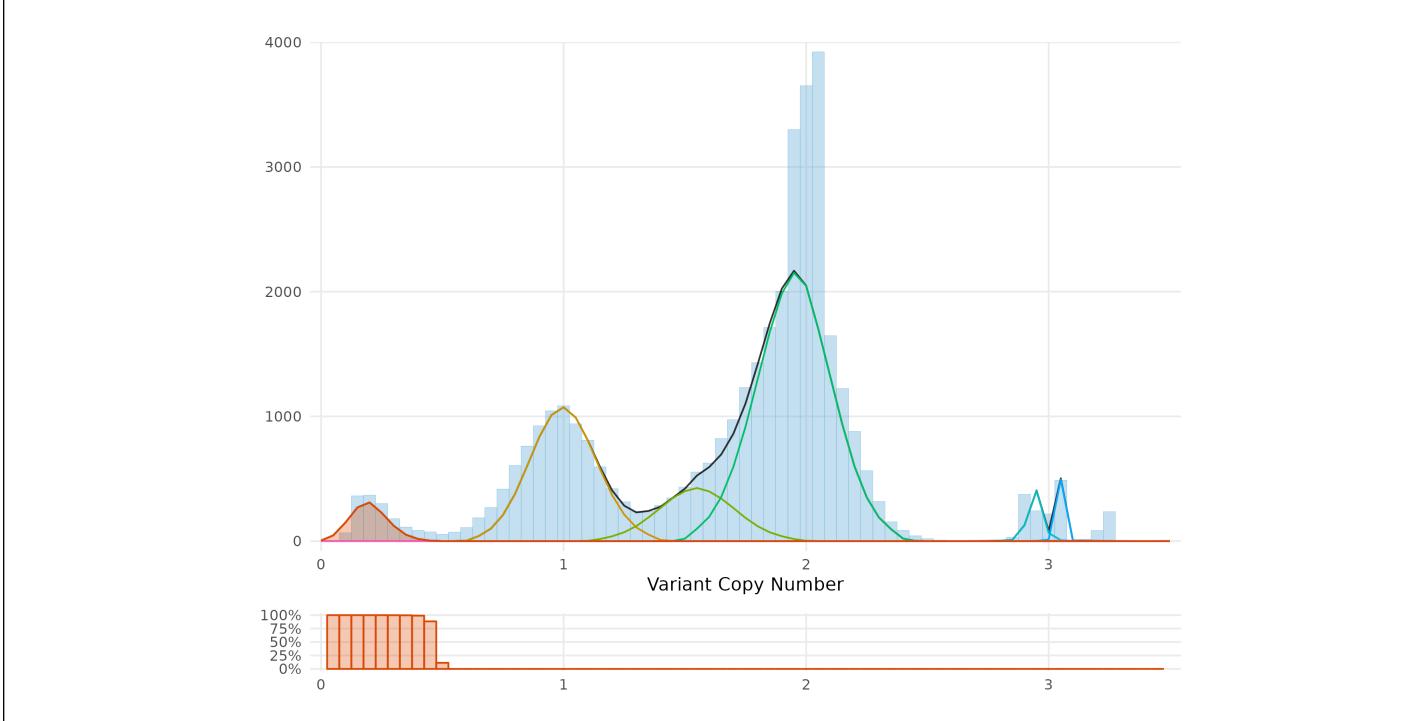
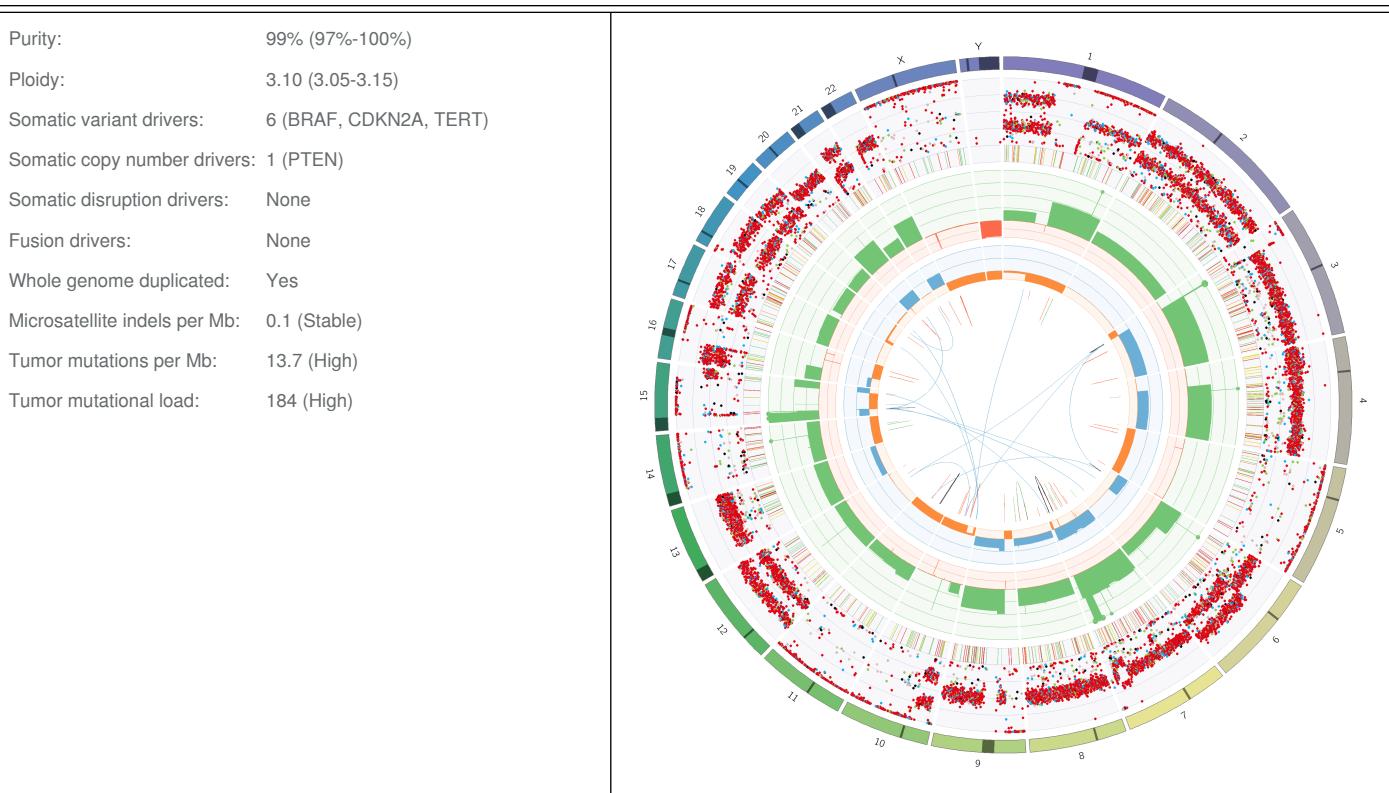
CONFIGURED PRIMARY TUMOR

skin melanoma (DOID 8923)

TUMOR-ONLY

QC

PASS





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## Somatic Findings

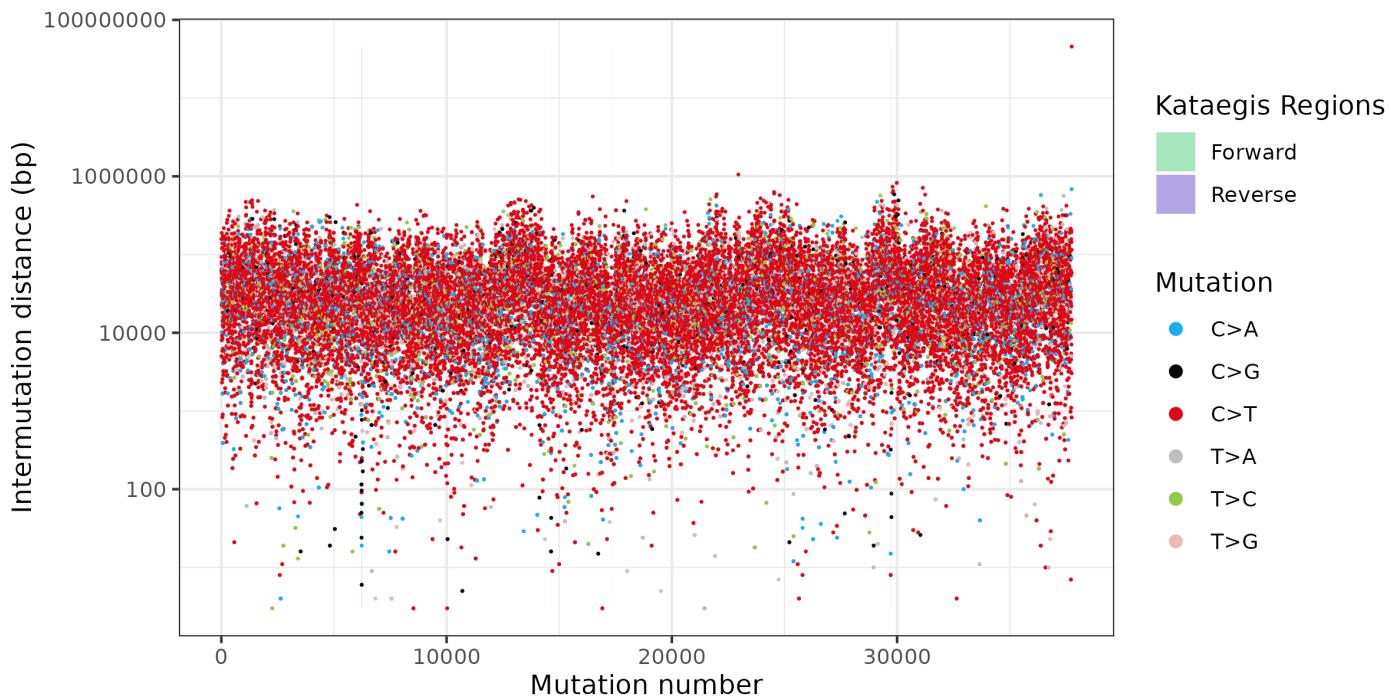
### Driver variants (7)

VARIANT	VCN	CN	MACN	BIALLELIC	HOTSPOT	DL	CL	PHASE ID	RNA DEPTH
BRAF p.V600E	4.1	6.0	2.0	2%	Yes	100%	100%		NA
CDKN2A p.A68fs	2.0	2.0	0.0	100%	Near	100%	100%		NA
CDKN2A (alt) p.G83fs	2.0	2.0	0.0	100%	Near	100%	100%		NA
TERT c.-125_-124delCCinsTT	1.7	2.0	0.0	92%	Yes	100%	100%	4304	NA
HDAC2 p.R409*	0.9	2.9	1.0	2%	No	34%	100%		NA
SF3B1 p.P718L	2.0	3.0	1.0	2%	No	15%	100%		NA
TP63 p.M499I	1.8	4.0	2.0	2%	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	2%	No		100%		NA
STK19 p.D89N	2.0	3.8	1.8	2%	Yes		100%		NA

## Kataegis plot



### Driver amplifications and homozygous deletions (1)

LOCATION	GENE	TYPE	CN	TPM	PERC (TYPE)	FC (TYPE)	PERC (DB)	FC (DB)
10q23.31	PTEN	partial del	0.0	NA	NA	NA	NA	NA

### Potentially interesting near-driver amplifications (0)

NONE



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### Other regions with amplifications (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 del	0.0	NA	NA	NA	NA	NA

### Potentially interesting chromosomal rearrangements

CHROMOSOMAL REARRANGEMENT	DETECTED?
1q trisomy	No
1p19q co-deletion	No

### 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
10	PTEN	Intron 5 Upstream	DEL	73	2.0	0.0
10	PTEN	Intron 6 Downstream	DEL	73	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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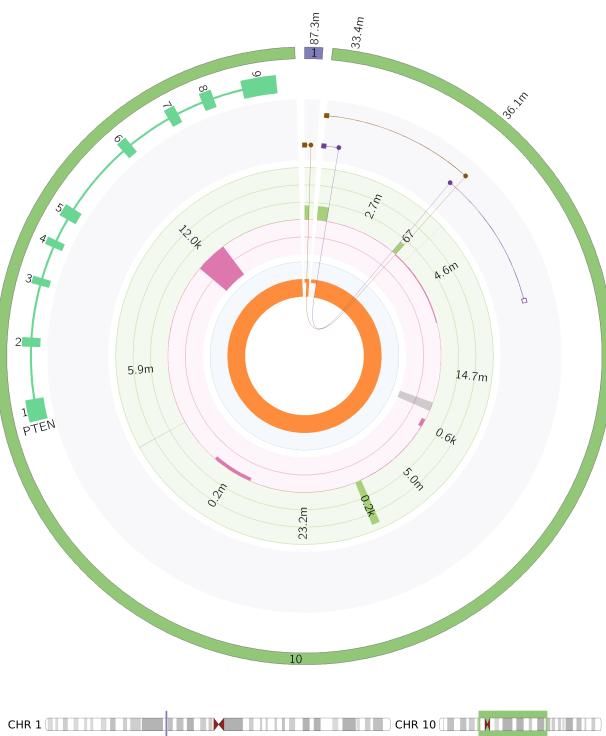
Test

Pipeline Version

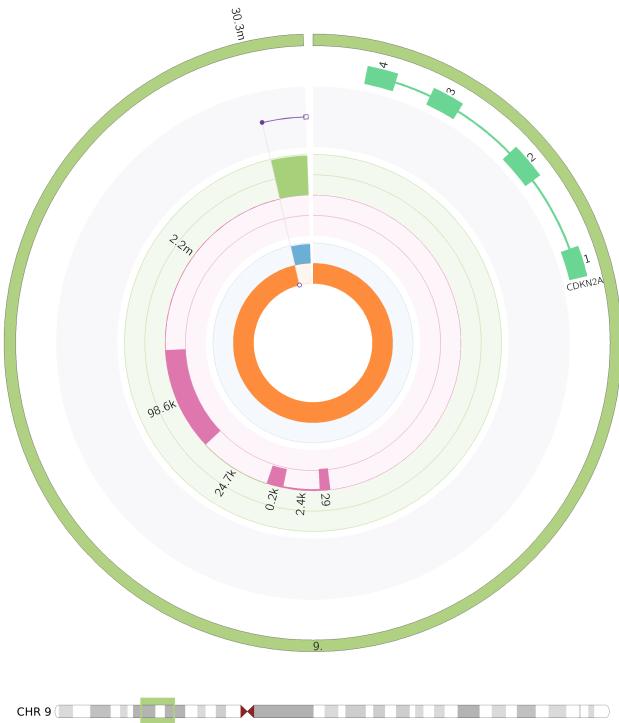
6.0



CHR 10



CHR 1



CHR 9



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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	1609	NA	2.0	NONE
A*01:01	NA	1609	NA	1.8	NONE
B*08:01	NA	753	NA	1.8	NONE
B*40:02	NA	726	NA	2.0	NONE
C*03:04	NA	772	NA	2.0	NONE
C*07:01	NA	772	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



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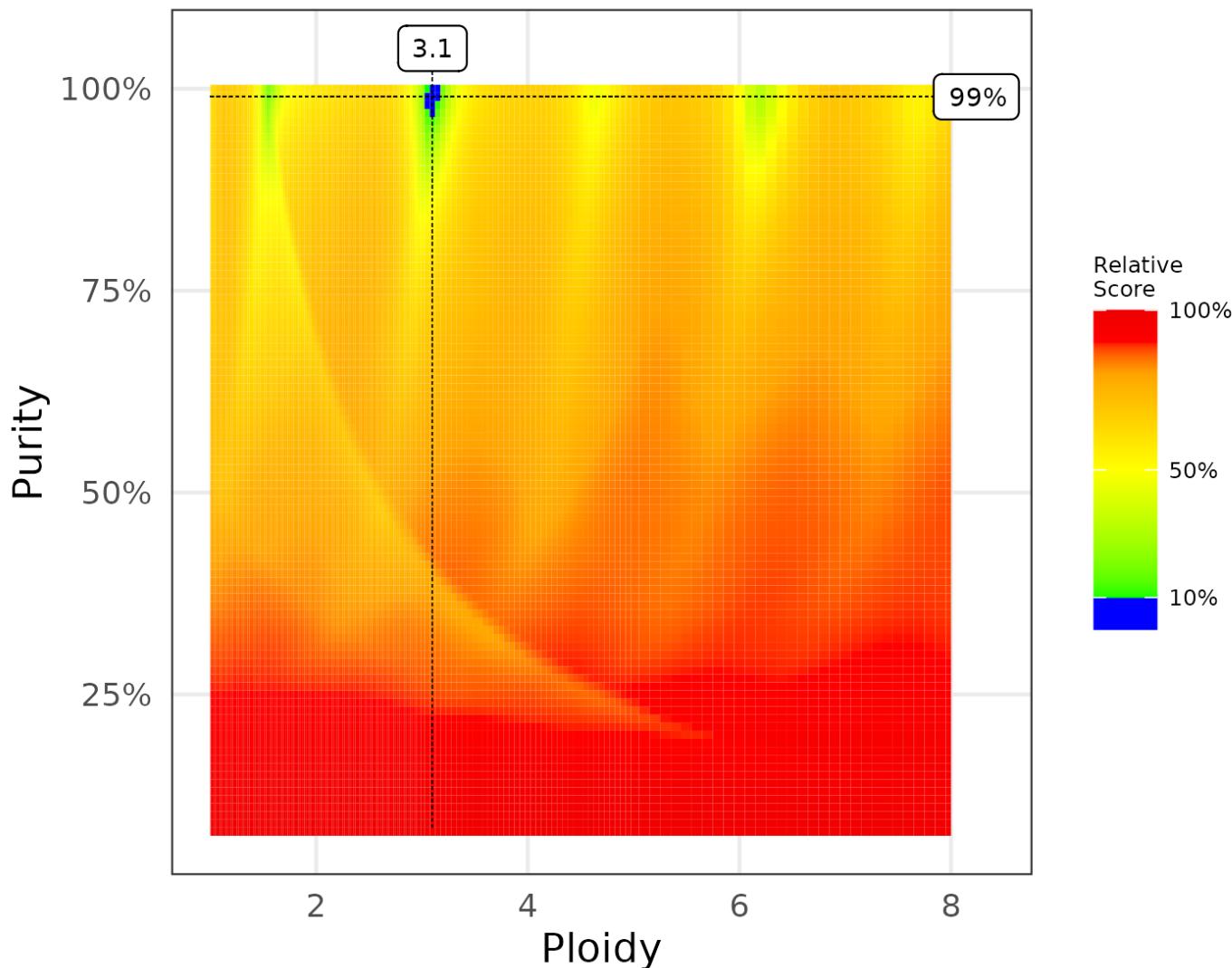
Pipeline Version

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### 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	2513301754	0	3179178	100%

### Coverage Stats

	MEAN COVERAGE	SD COVERAGE	MEDIAN COVERAGE	MAD COVERAGE
Tumor Sample	107.6	34.9	109	23



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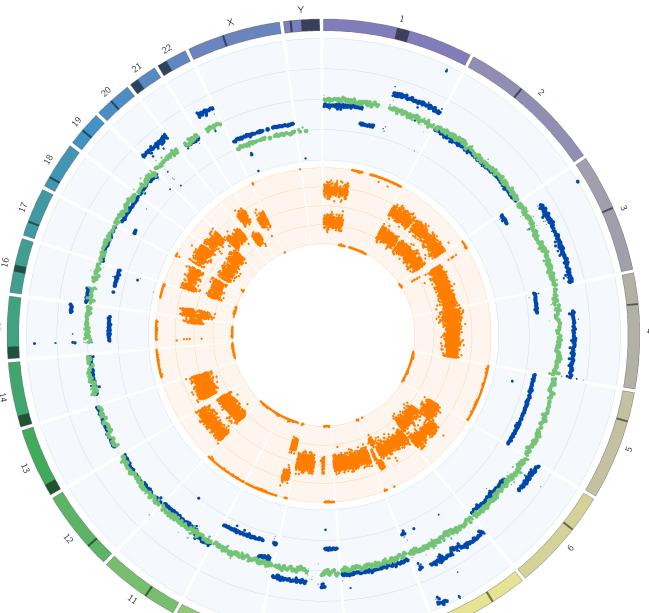
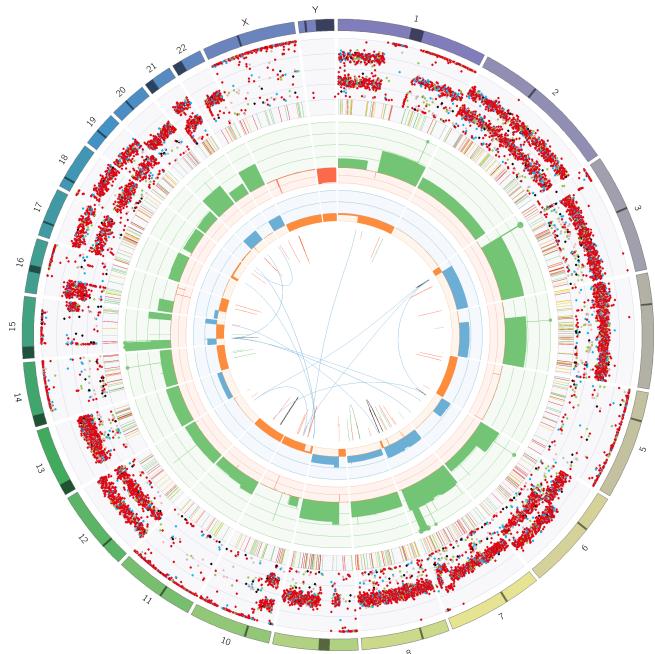
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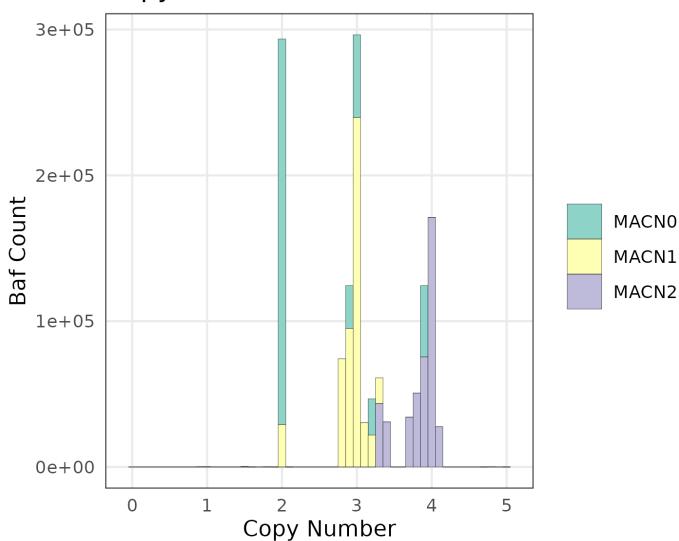
### Excluded Percentages

	ADAPTER	BASEQ	CAPPED	DUPE	MAPQ	OVERLAP	UNPAIRED	TOTAL
Tumor Sample	0%	0%	0%	15%	2%	1%	0%	18%

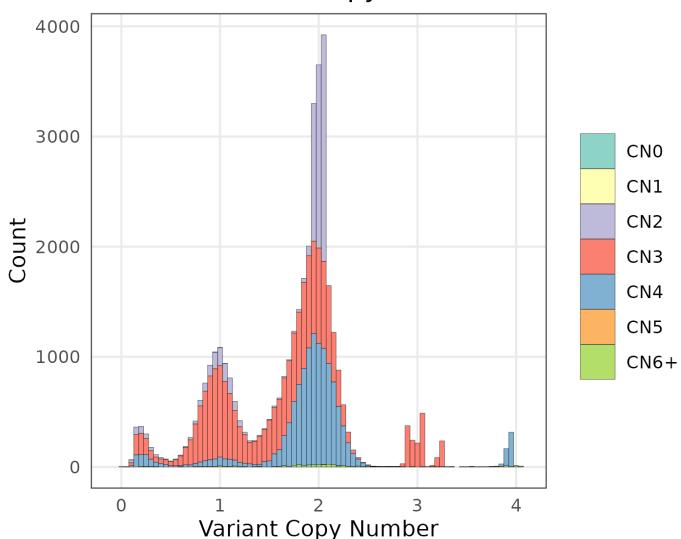
### QC plots



### Copy Number PDF



### Somatic Variant Copy Number PDF



### Tumor Sample BQR plot



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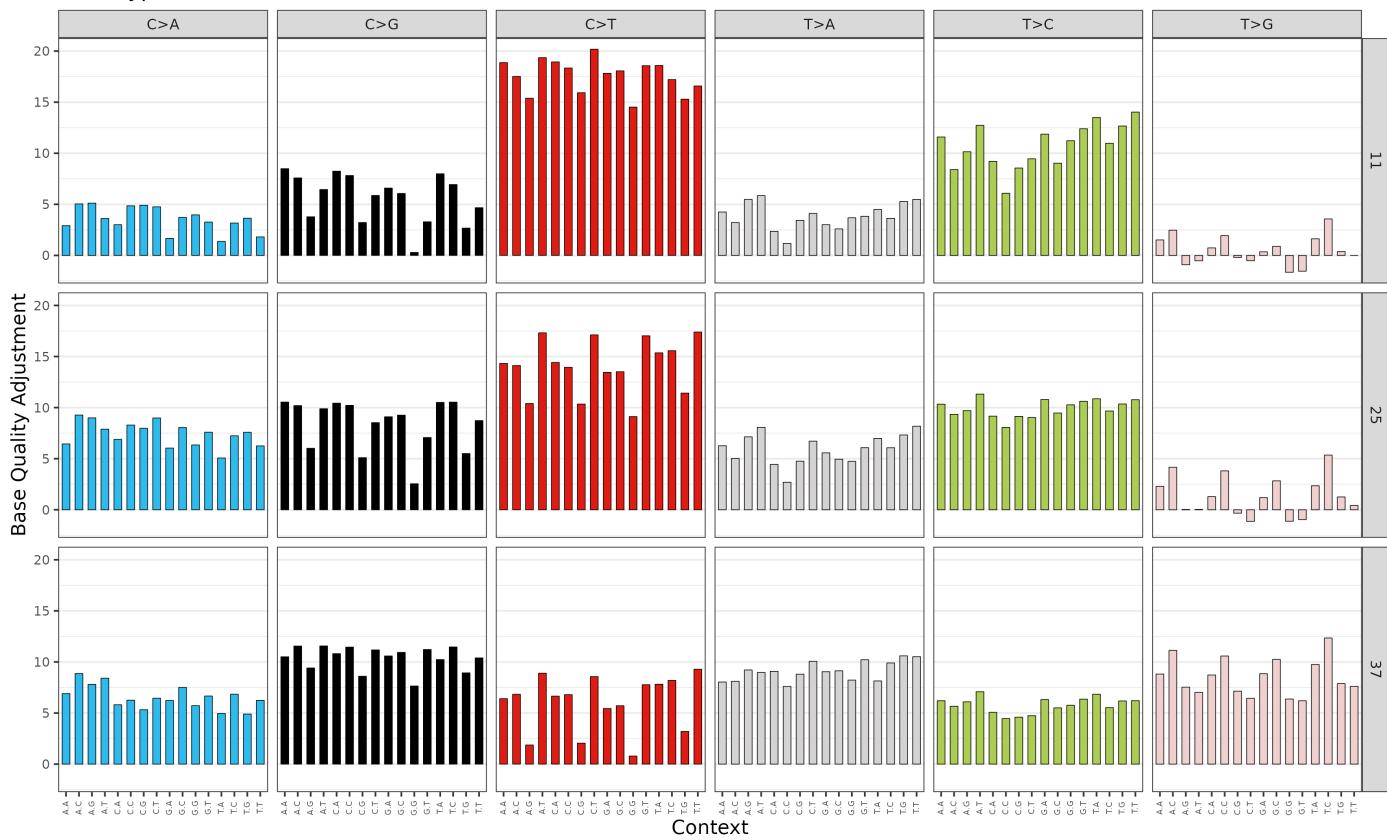
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Read type: NONE



Read type: SINGLE

