



CONFIGURED PRIMARY TUMOR

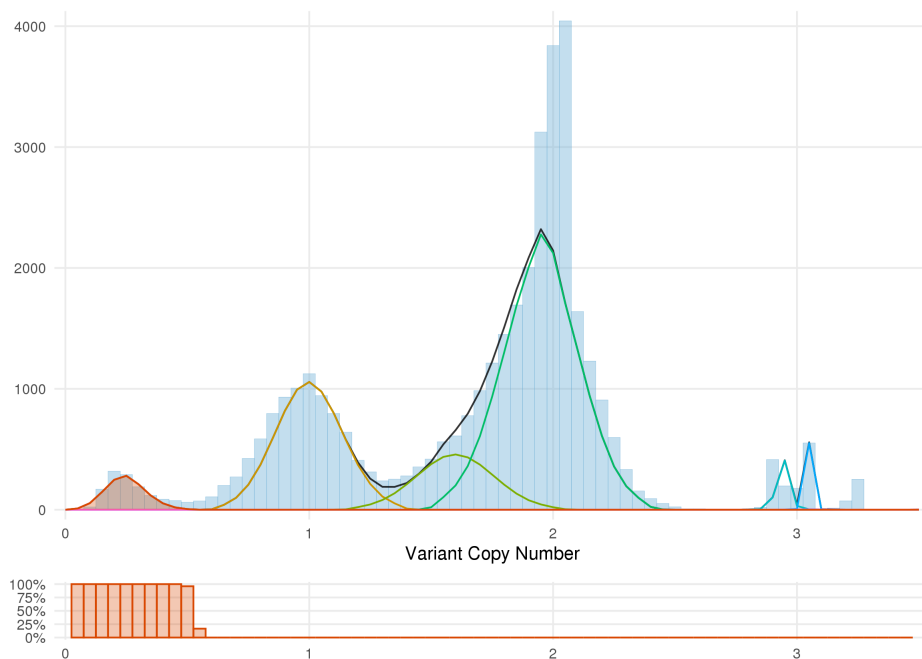
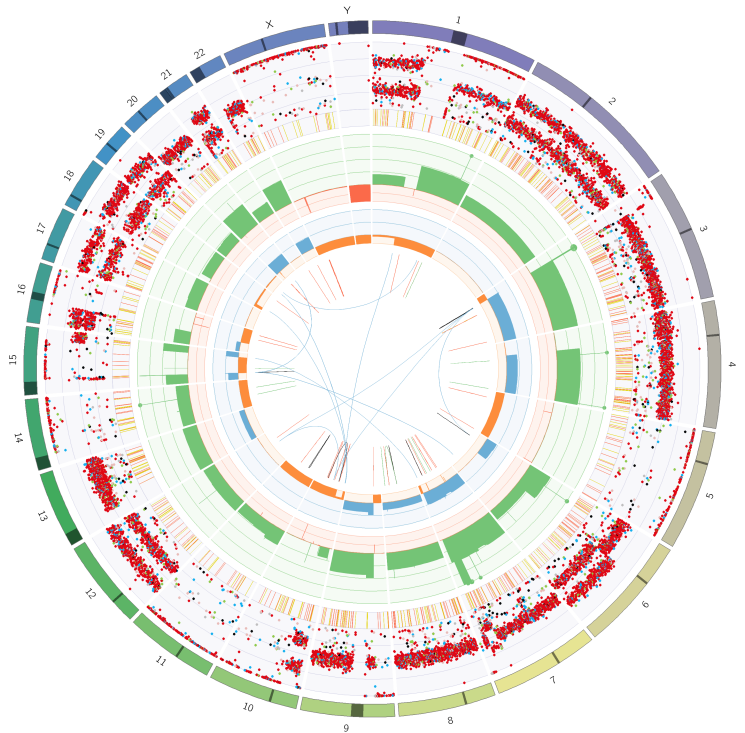
TUMOR-ONLY

QC

skin melanoma (DOID 8923)

PASS

Purity: 99% (97%-100%)  
Ploidy: 3.10 (3.05-3.15)  
Somatic variant drivers: 5 (BRAF, CDKN2A, TERT)  
Somatic copy number drivers: 1 (PTEN)  
Somatic disruption drivers: None  
Fusion drivers: None  
Whole genome duplicated: Yes  
Microsatellite indels per Mb: 0.1 (Stable)  
Tumor mutations per Mb: 13.7 (High)  
Tumor mutational load: 184 (High)





## Somatic Findings

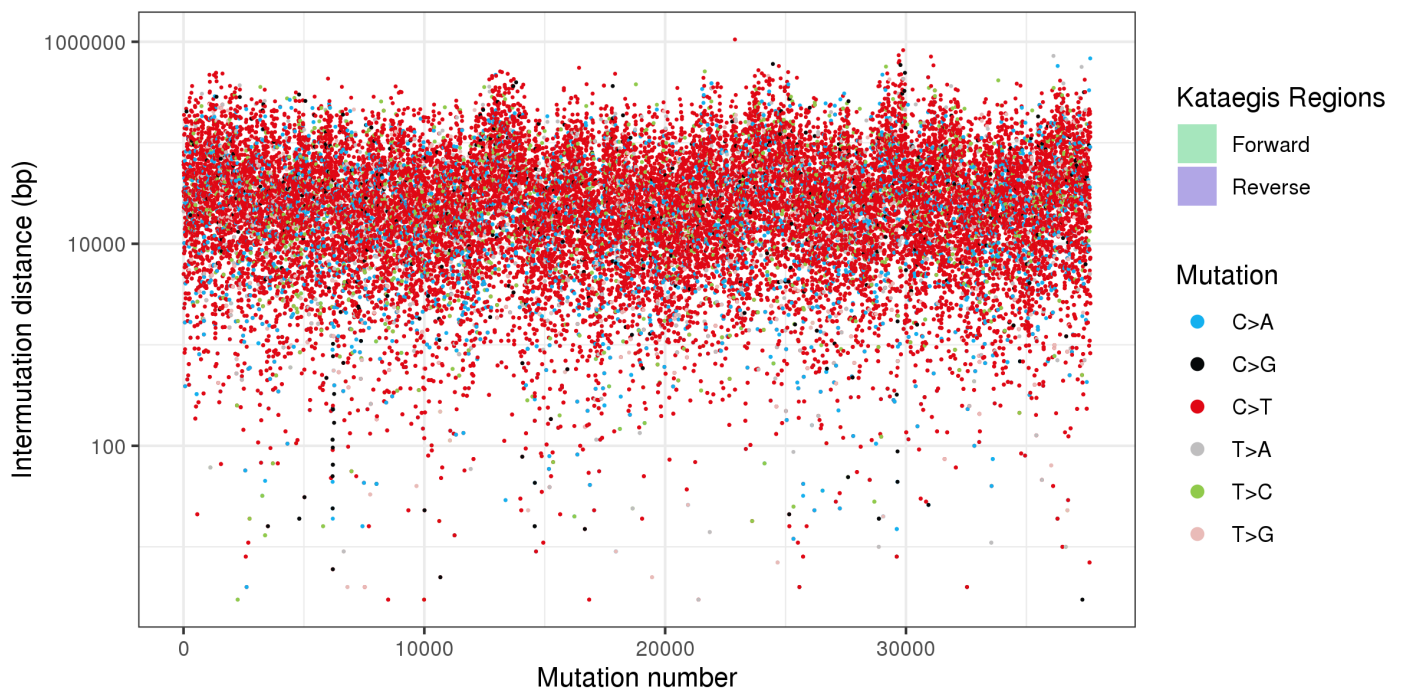
## Driver variants (6)

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

## 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

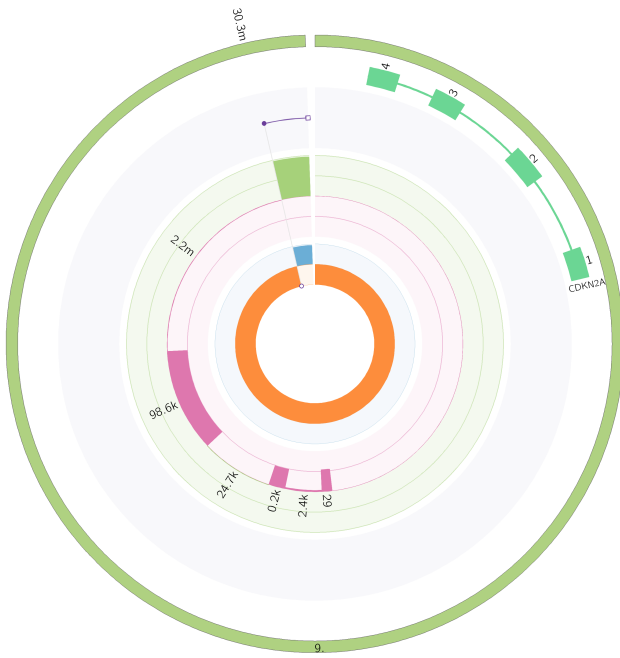
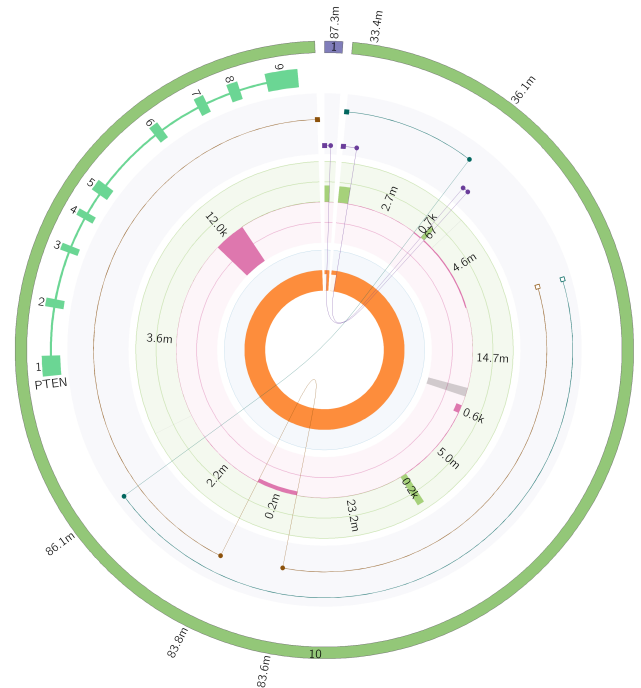
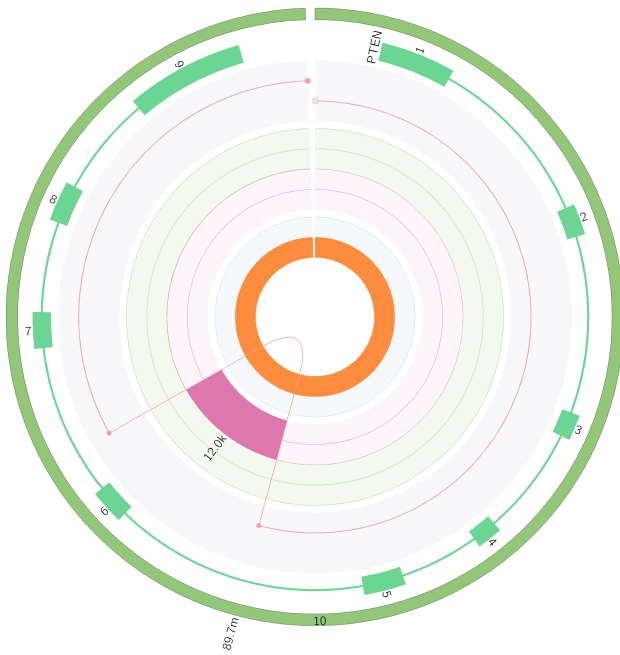
## Other potentially interesting gene disruptions (0)

NONE

## Potentially interesting LOH events (0)

NONE

## Structural driver plots (3)





## 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

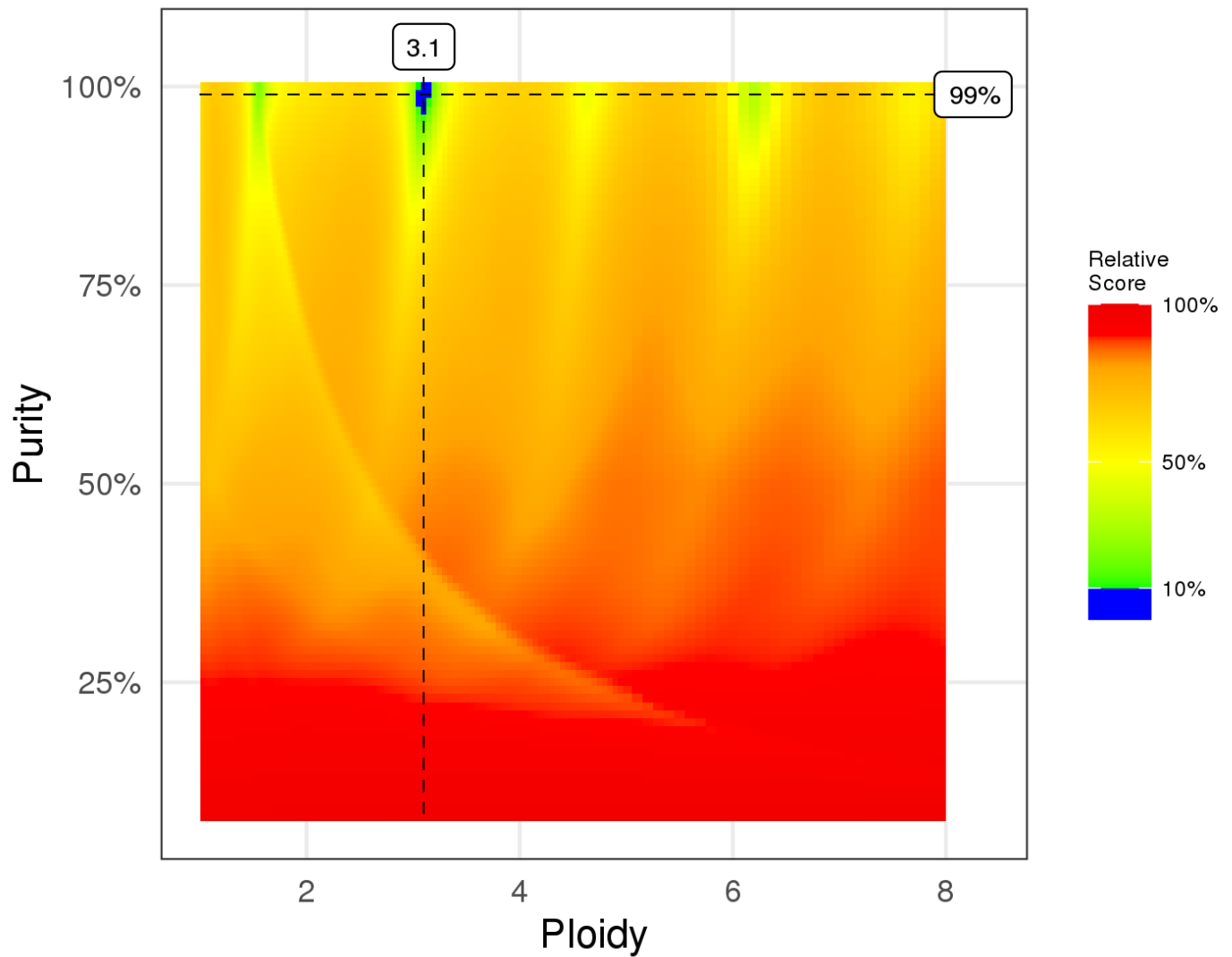
ESCAPE MECHANISM	DETECTED?
HLA-1 loss-of-function	No
Antigen presentation pathway inactivation	No
IFN gamma pathway inactivation	No
(Potential) PD-L1 overexpression	No
CD58 inactivation	No
Epigenetics driven immune escape via SETDB1	No



Quality Control

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

Purity/Ploidy Scores



Flagstats

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

Coverage Stats

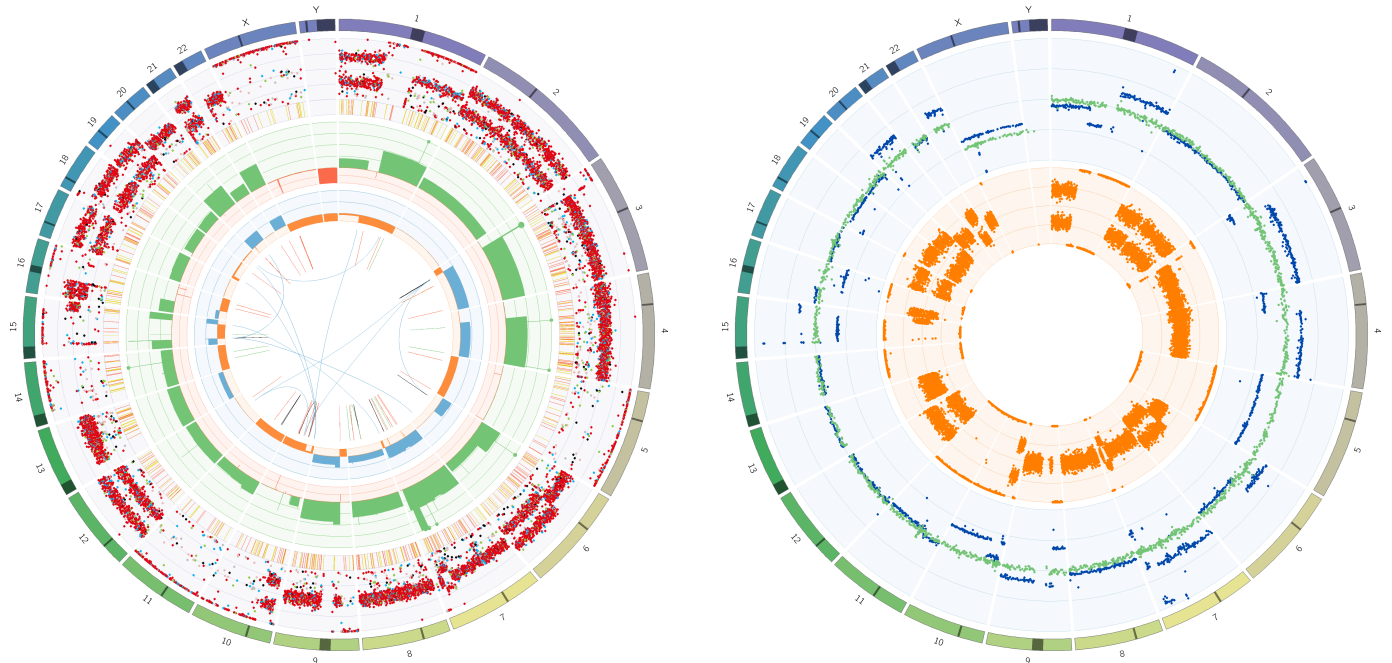
	MEAN COVERAGE	SD COVERAGE	MEDIAN COVERAGE	MAD COVERAGE
Tumor Sample	108.0	35.0	109	24



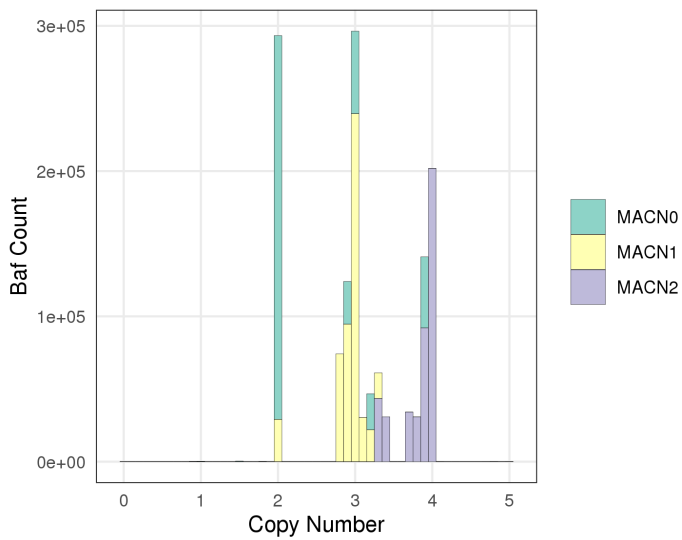
Excluded Percentages

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

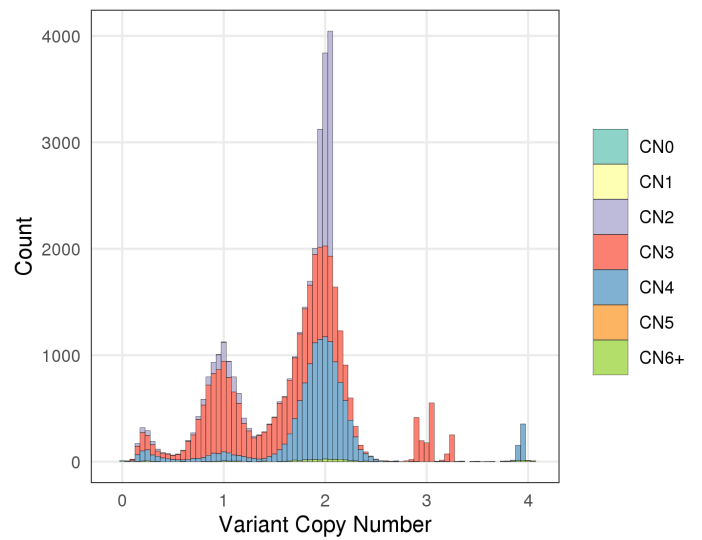
QC plots



Copy Number PDF



Somatic Variant Copy Number PDF



Tumor Sample BQR plot

