



CONFIGURED PRIMARY TUMOR

skin melanoma (DOID 8923)

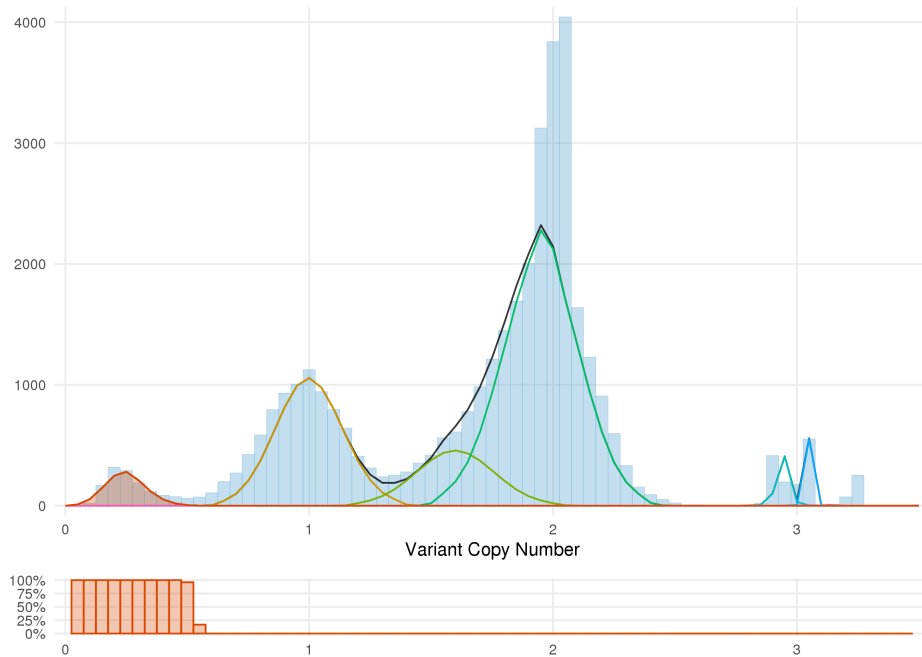
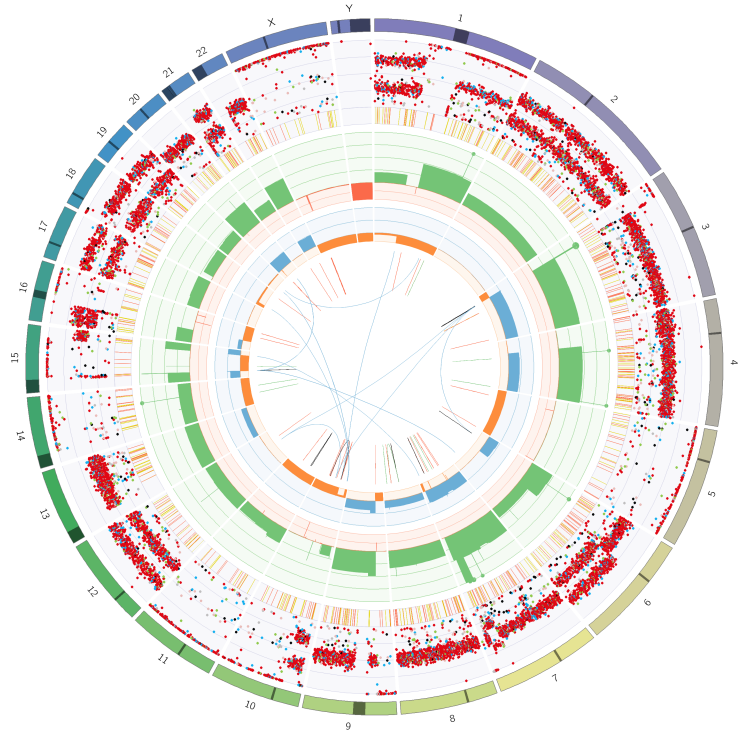
CUPPA CANCER TYPE

Skin: Melanoma (100%)

QC

PASS

Purity: 99% (97%-100%)
Ploidy: 3.10 (3.05-3.15)
Somatic variant drivers: 5 (BRAF, CDKN2A, TERT)
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





Somatic Findings

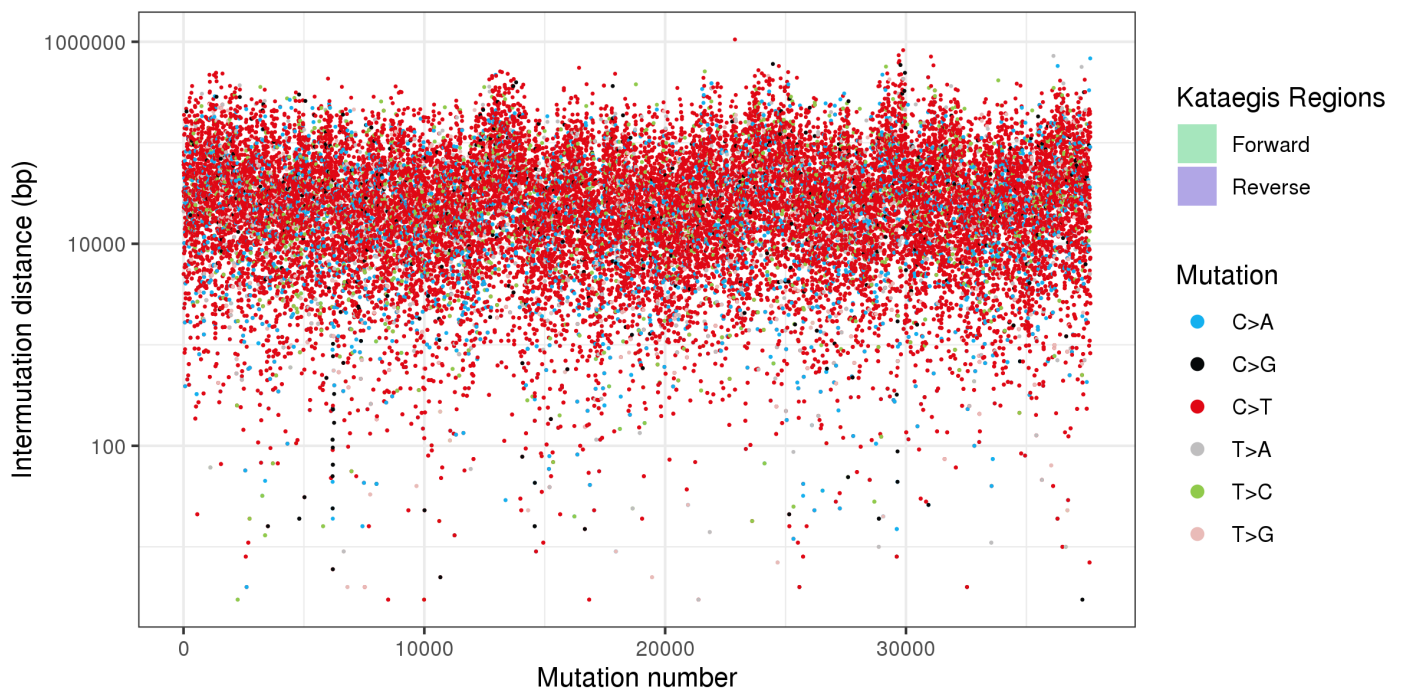
Driver variants (6)

| 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



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

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 |
|----------|------|---------------------|------|------------|-------------|----------------|
| 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

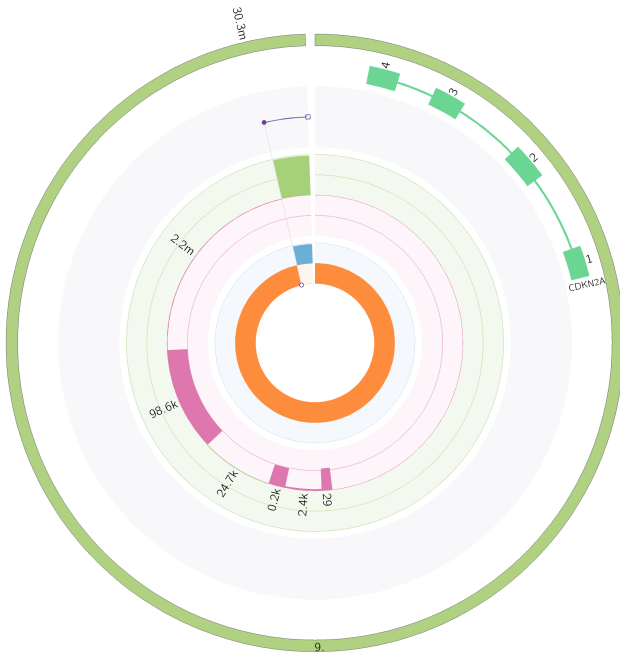
Potentially interesting LOH events (0)

NONE

Signature allocations (12)

| SIGNATURE | ALLOCATION | PERCENT |
|-----------|------------|---------|
| Sig2 | 3702.7 | 10% |

THE TABLE CONTINUES ON THE NEXT PAGE





Germline Findings

Driver variants (0)

NONE

Other potentially relevant variants (3)

| VARIANT | VCN | CN | MACN | RNA DEPTH | BIALLELIC | HOTSPOT | GENOTYPE |
|--------------------------|-----|-----|------|-----------|-----------|---------|----------|
| CYP2D6 c.506-1G>A splice | 1.8 | 4.0 | 2.0 | NA | No | Yes | HET |
| CYP3A4 c.522-191C>T | 2.4 | 4.0 | 2.0 | NA | No | Yes | HET |
| CYP3A4 upstream | 4.0 | 4.0 | 2.0 | NA | Yes | Yes | HOM |

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 | GENOTYPE | FUNCTION | LINKED DRUGS | SOURCE |
|--------|----------|-----------------|-------------------------------------|--------------------------|
| DPYD | *1_HOM | Normal Function | 5-Fluorouracil;Capecitabine;Tegafur | PHARMGKB |
| UGT1A1 | *1_HOM | Normal Function | Irinotecan | PHARMGKB |



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 |



Test

5.34

Metric value

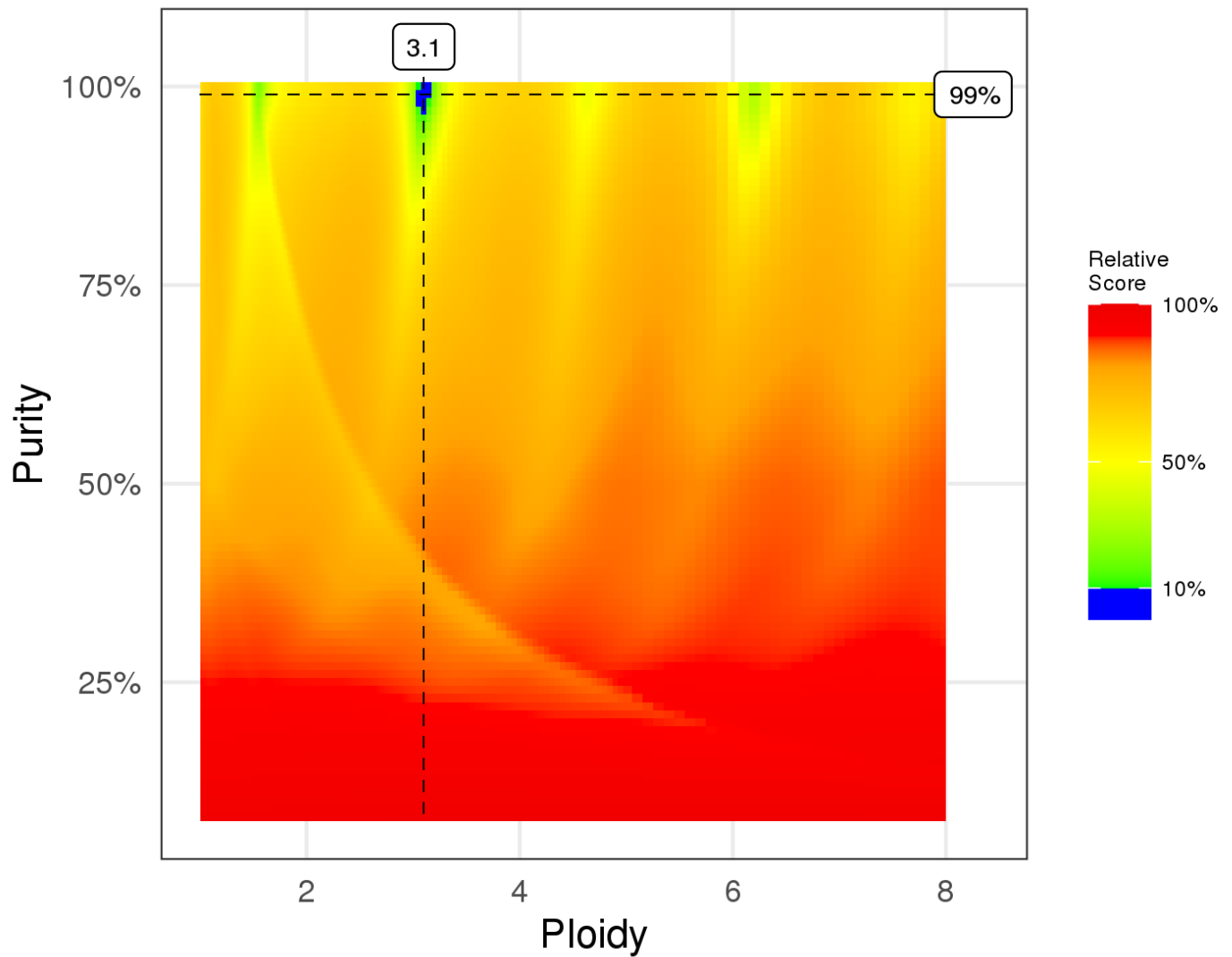
1.00
0.75
0.50
0.25
0.00



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



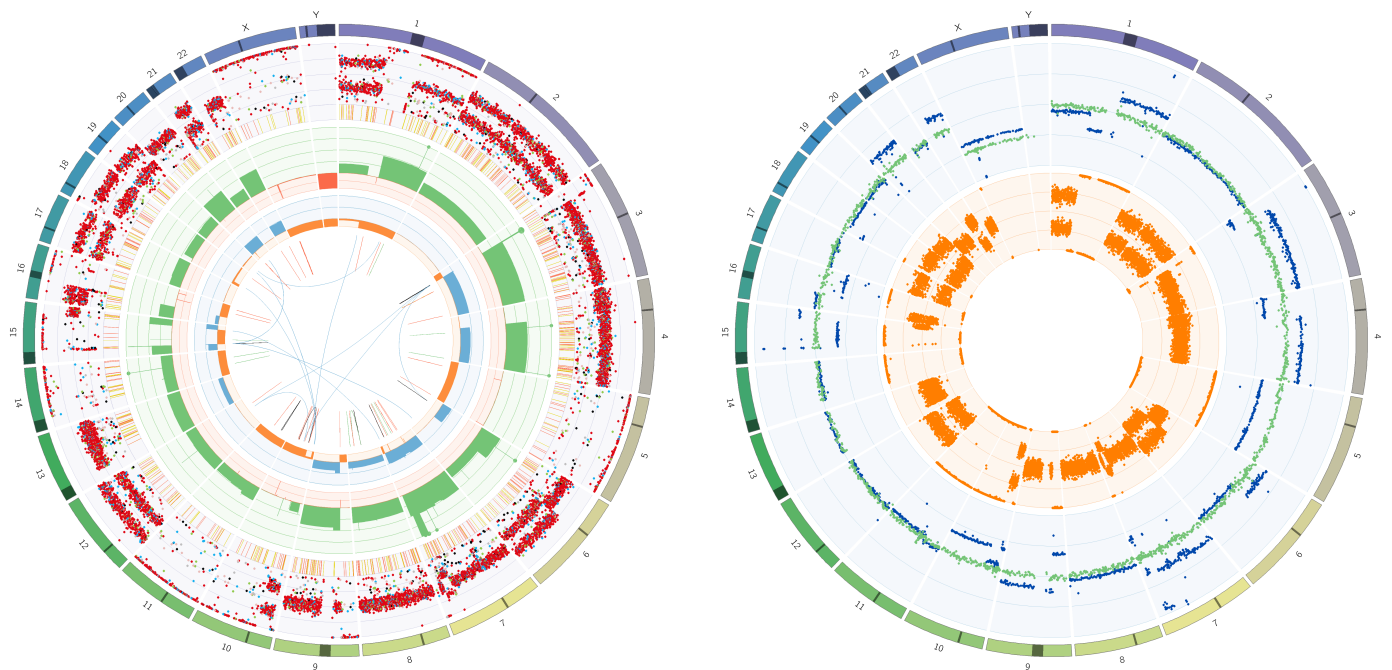
Coverage Stats

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

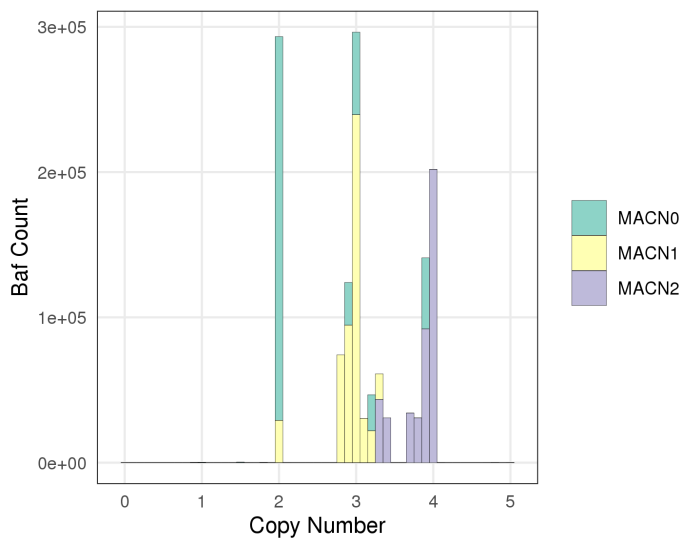
Excluded Percentages

| | 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% |

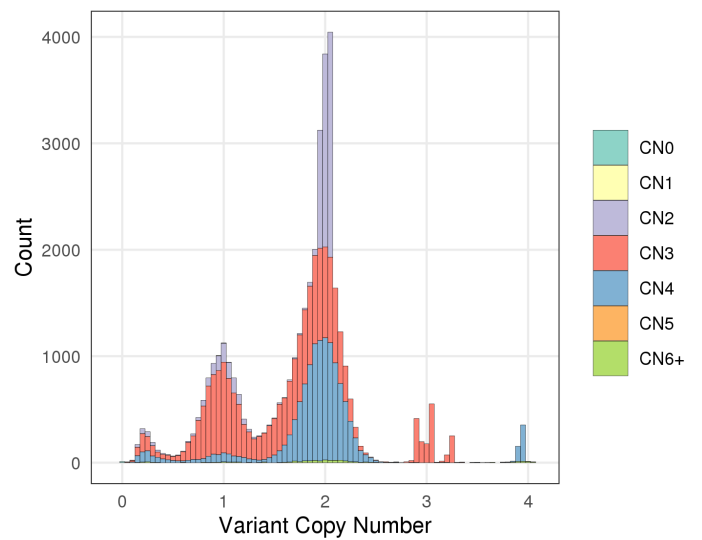
QC plots



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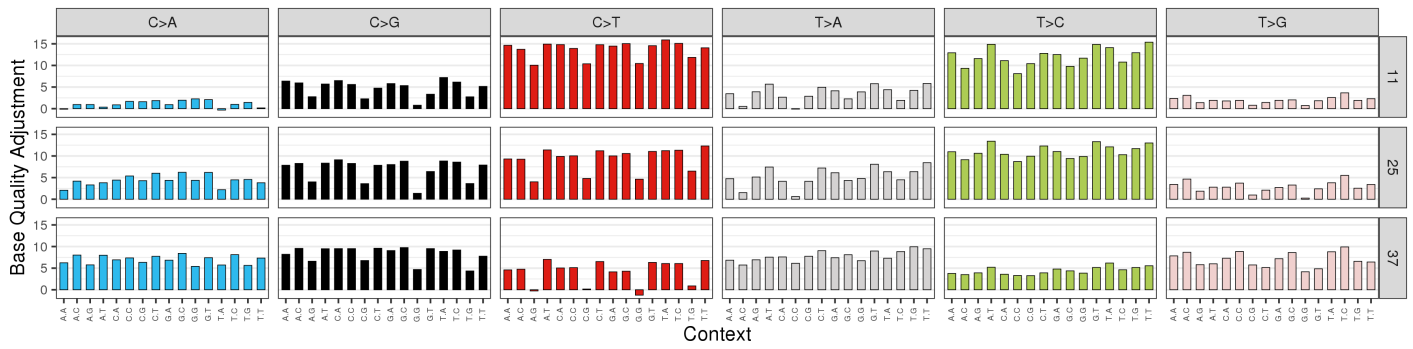


Somatic Variant Copy Number PDF





Reference Sample BQR plot



Tumor Sample BQR plot

