



ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.34

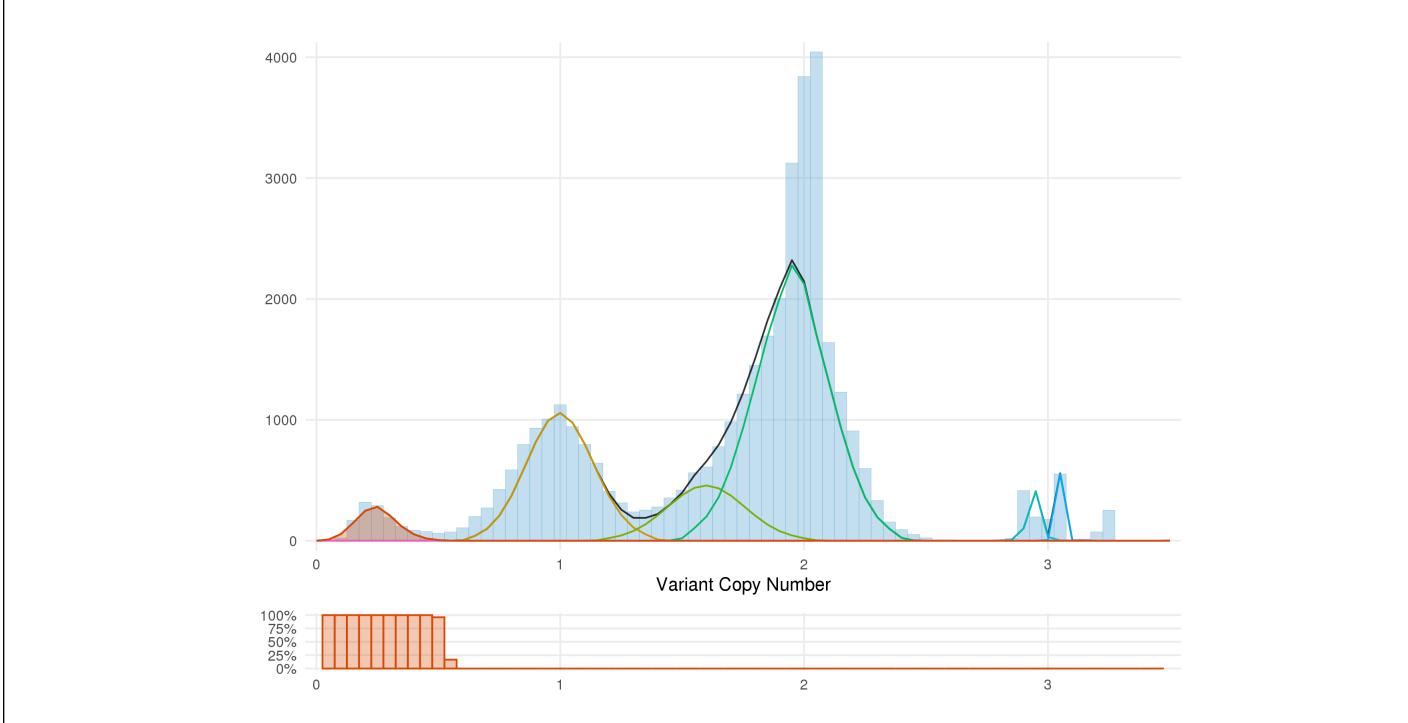
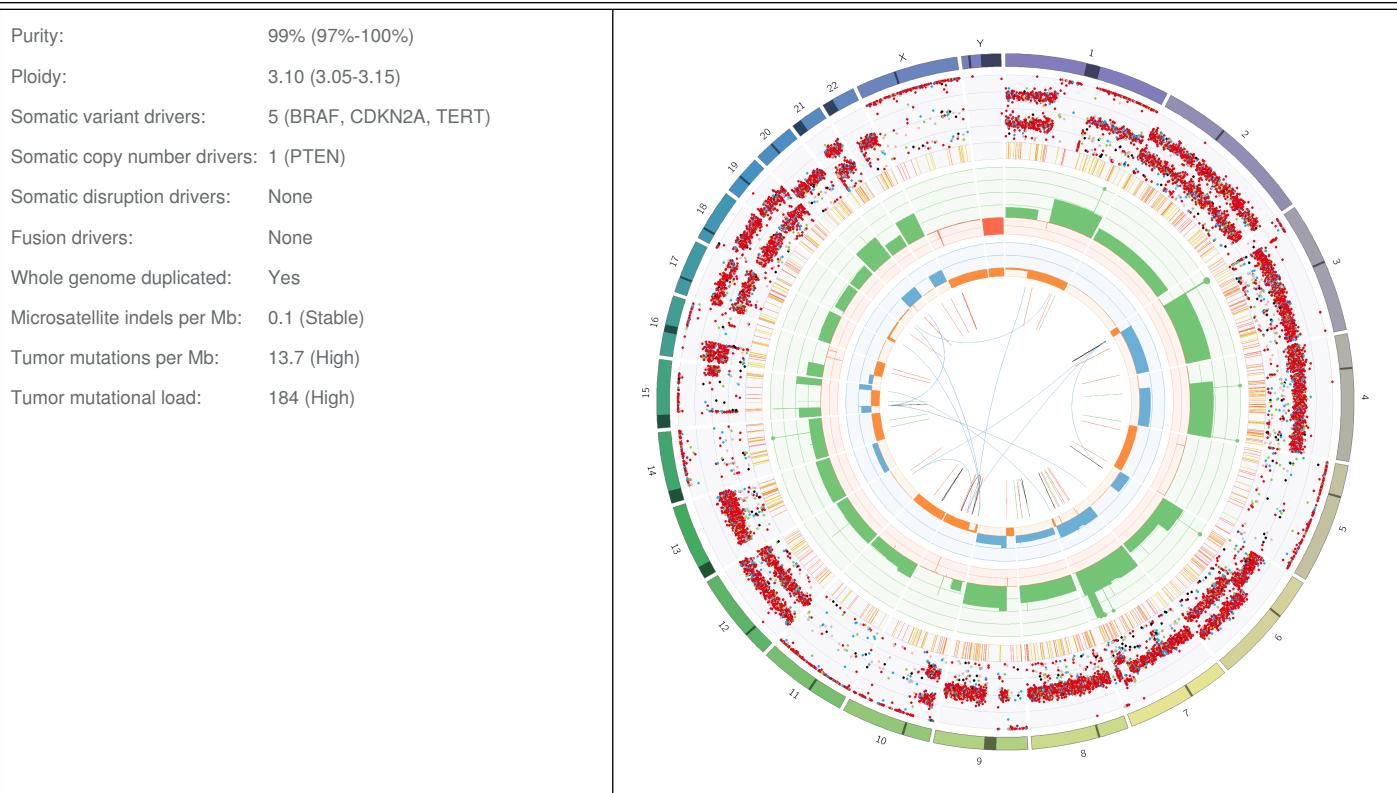
CONFIGURED PRIMARY TUMOR

skin melanoma (DOID 8923)

TUMOR-ONLY

QC

PASS





Somatic Findings

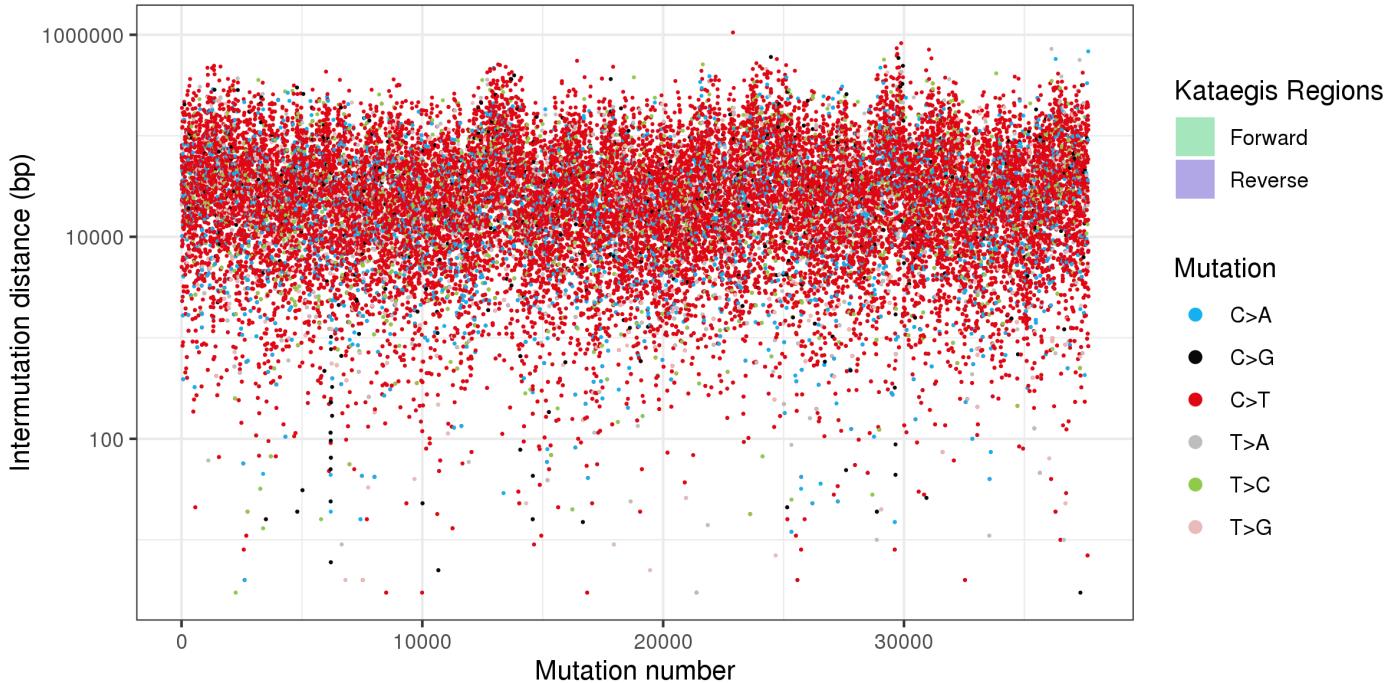
Driver variants (6)

| 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

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)



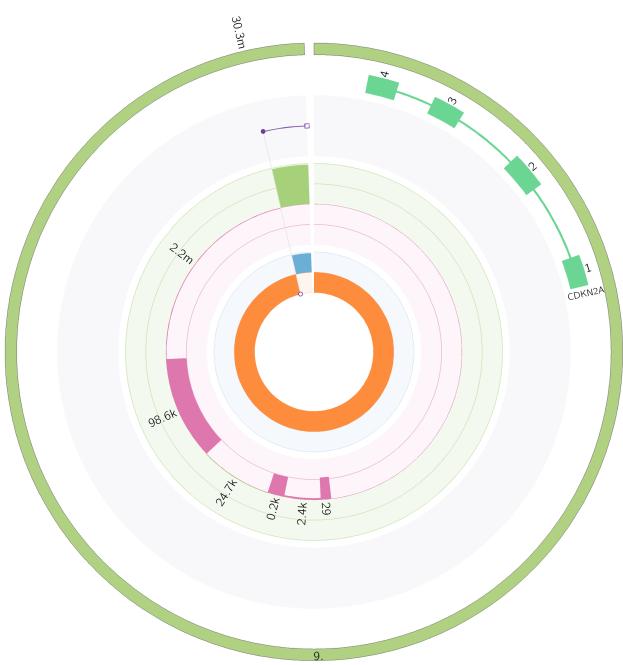
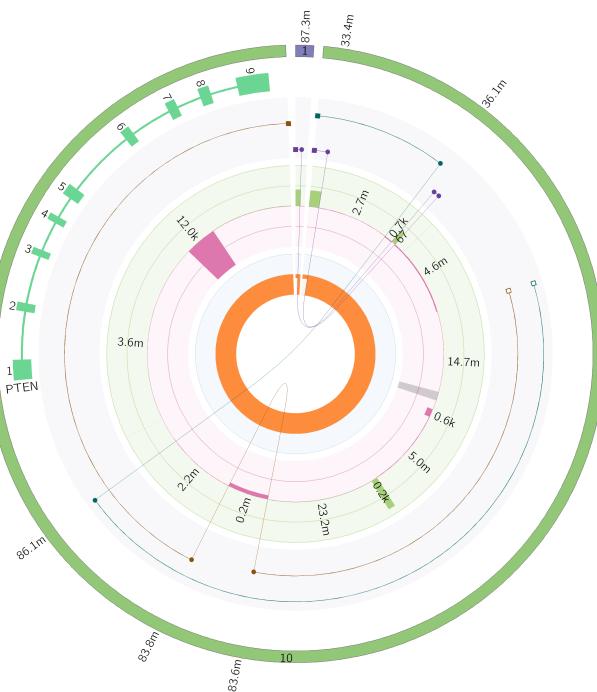
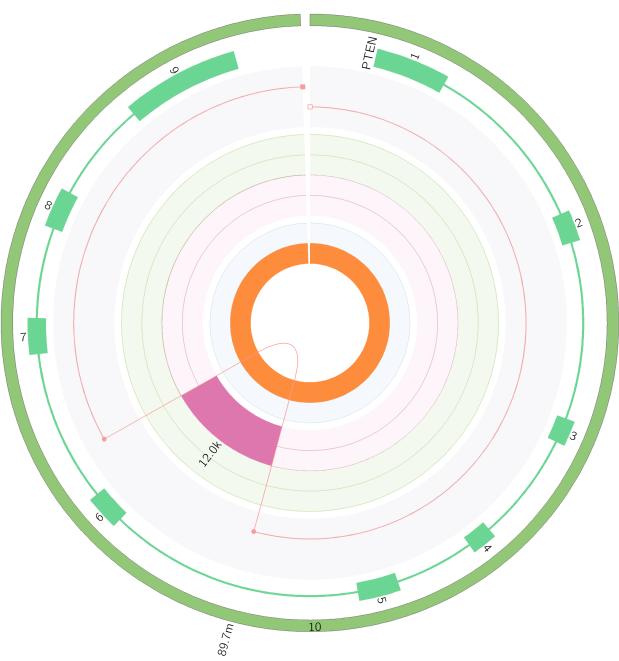
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Immunology

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 |



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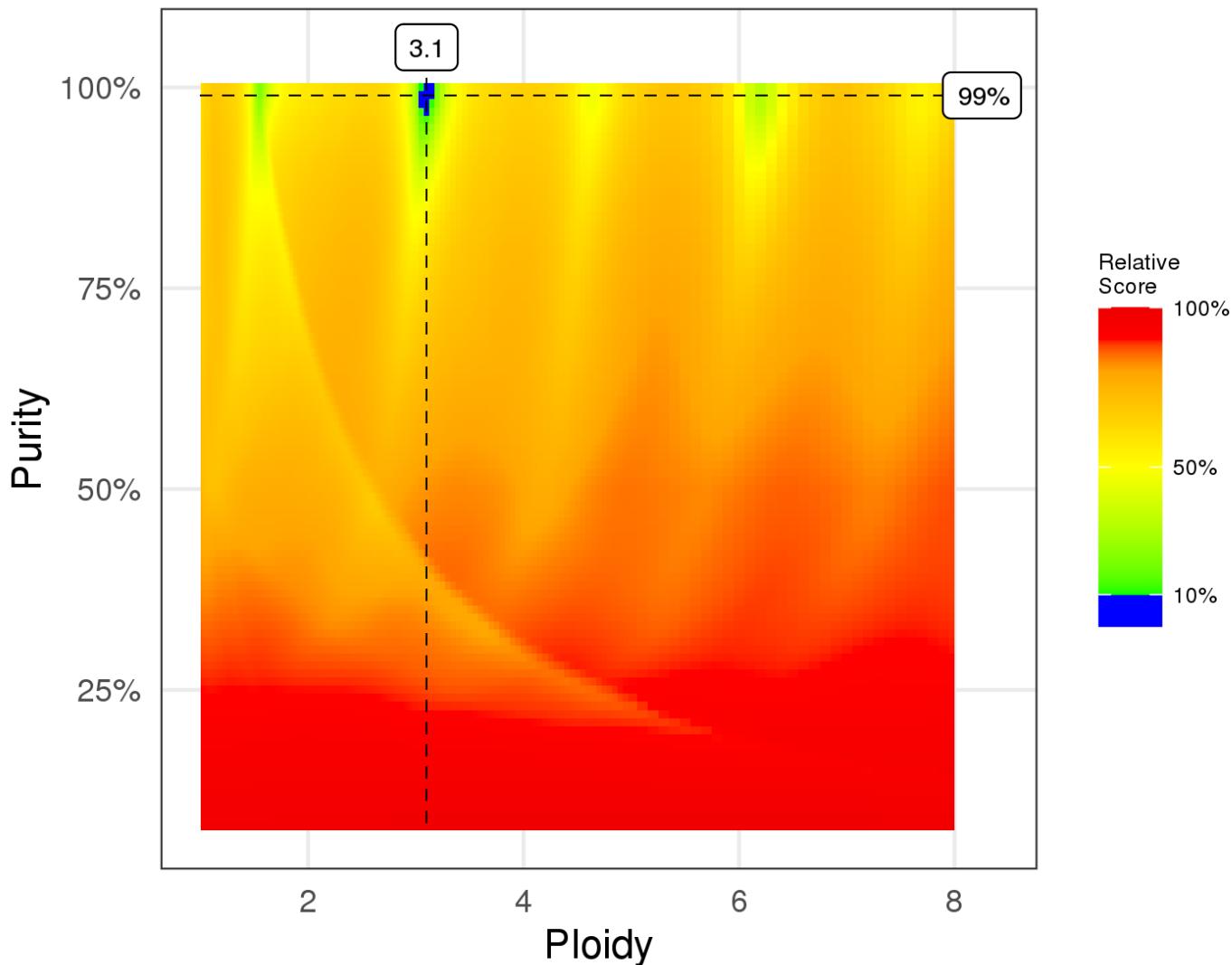
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Quality Control

| 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 |



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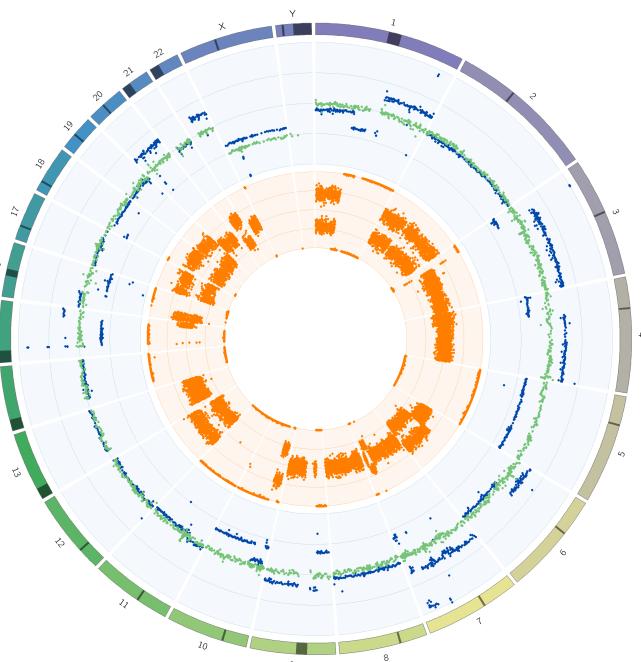
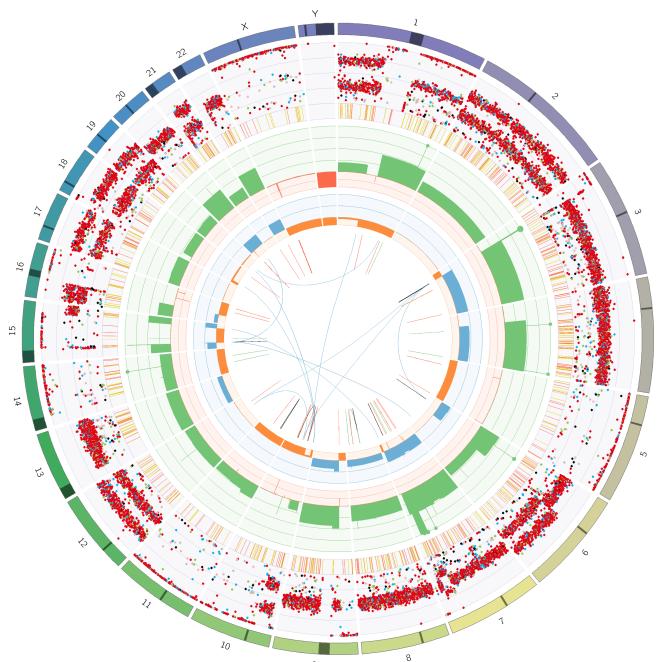
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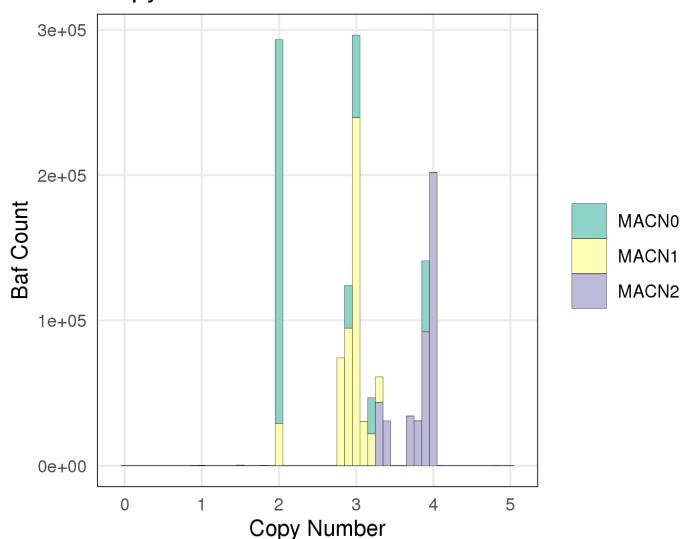
Excluded Percentages

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

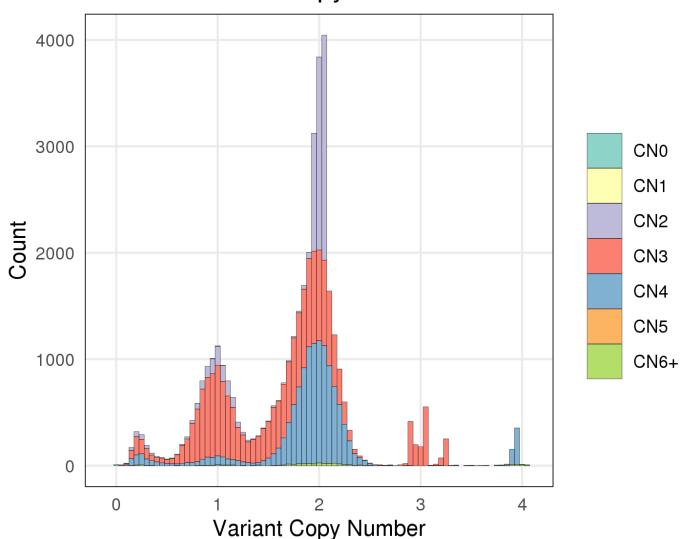
QC plots



Copy Number PDF



Somatic Variant Copy Number PDF



Tumor Sample BQR plot



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