



ORANGE Report

SAMPLE

Test

PLATINUM VERSION

5.23

CONFIGURED PRIMARY TUMOR

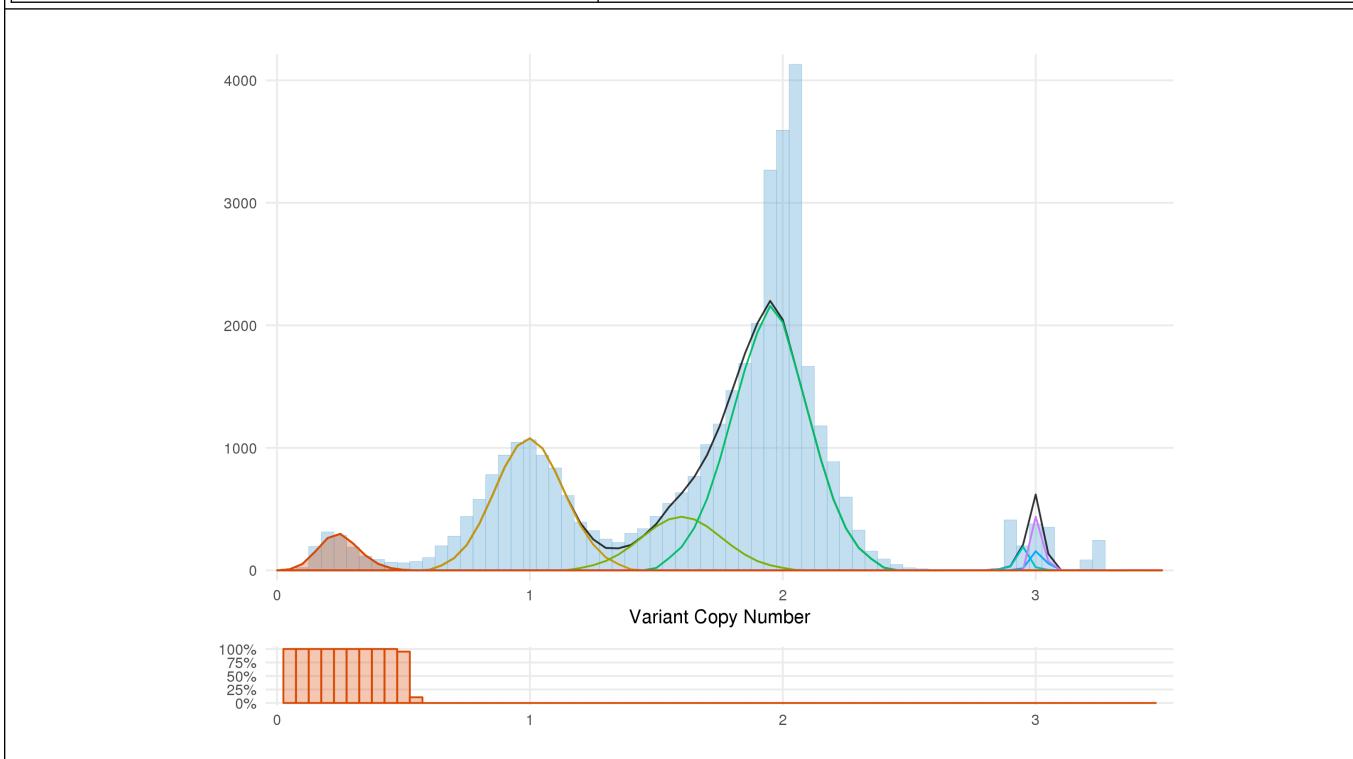
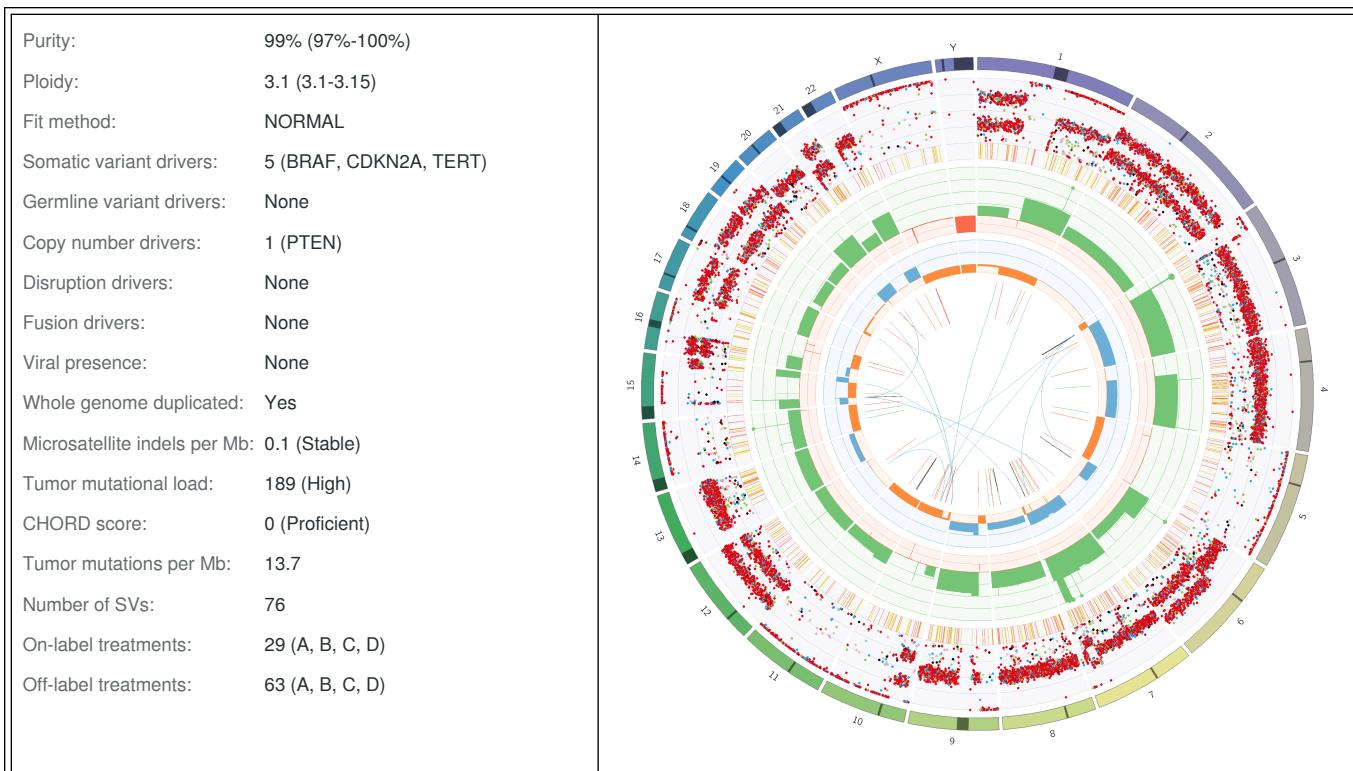
skin melanoma (DOID 8923)

CUPPA PRIMARY TUMOR

Melanoma (likelihood=99.6%)

QC

PASS





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Clinical Evidence

* Treatments are reported up to a maximum evidence level of 'B'.

On-Label Evidence

TREATMENT	RESPONSIVE EVIDENCE	RESISTANCE EVIDENCE
Cobimetinib + Vemurafenib	BRAF p.V600E (A - CGI)	
Dabrafenib	BRAF p.V600E (A - CGI)	
Dabrafenib + Trametinib	BRAF p.V600E (A - CGI, CIViC)	
Trametinib	BRAF p.V600E (A - CGI)	
Vemurafenib	BRAF p.V600E (A - CGI, CIViC)	

Off-Label Evidence

TREATMENT	RESPONSIVE EVIDENCE	RESISTANCE EVIDENCE
Dabrafenib	BRAF p.V600E (A - CGI)	
Dabrafenib + Trametinib	BRAF p.V600E (A - CGI, CIViC)	
Vemurafenib	BRAF p.V600E (A - CGI)	BRAF p.V600E (B - CIViC)
CI-1040	BRAF p.V600E (B - CIViC)	
Cetuximab	BRAF p.V600E (B - CIViC)	PTEN partial loss (B - CIViC)
Cetuximab + Irinotecan + Vemurafenib	BRAF p.V600E (B - CIViC)	
Cetuximab + Vemurafenib	BRAF p.V600E (B - CIViC)	
Chemotherapy		PTEN partial loss (B - CIViC)
Everolimus		PTEN partial loss (B - CIViC)
Fluorouracil		BRAF p.V600E (B - CIViC)
Irinotecan		BRAF p.V600E (B - CIViC)
Lapatinib + Trastuzumab		PTEN partial loss (B - CIViC)
Oxaliplatin		BRAF p.V600E (B - CIViC)
Panitumumab	BRAF p.V600E (B - CIViC)	
Panitumumab + Vemurafenib	BRAF p.V600E (B - CIViC)	
Ridaforolimus		PTEN partial loss (B - CIViC)
Selumetinib	BRAF p.V600E (B - CIViC)	
Sorafenib	BRAF p.V600E (B - CIViC)	
Tensirolimus		PTEN partial loss (B - CIViC)
Trastuzumab		PTEN partial loss (B - CIViC)



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Trials

NONE



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Somatic Findings

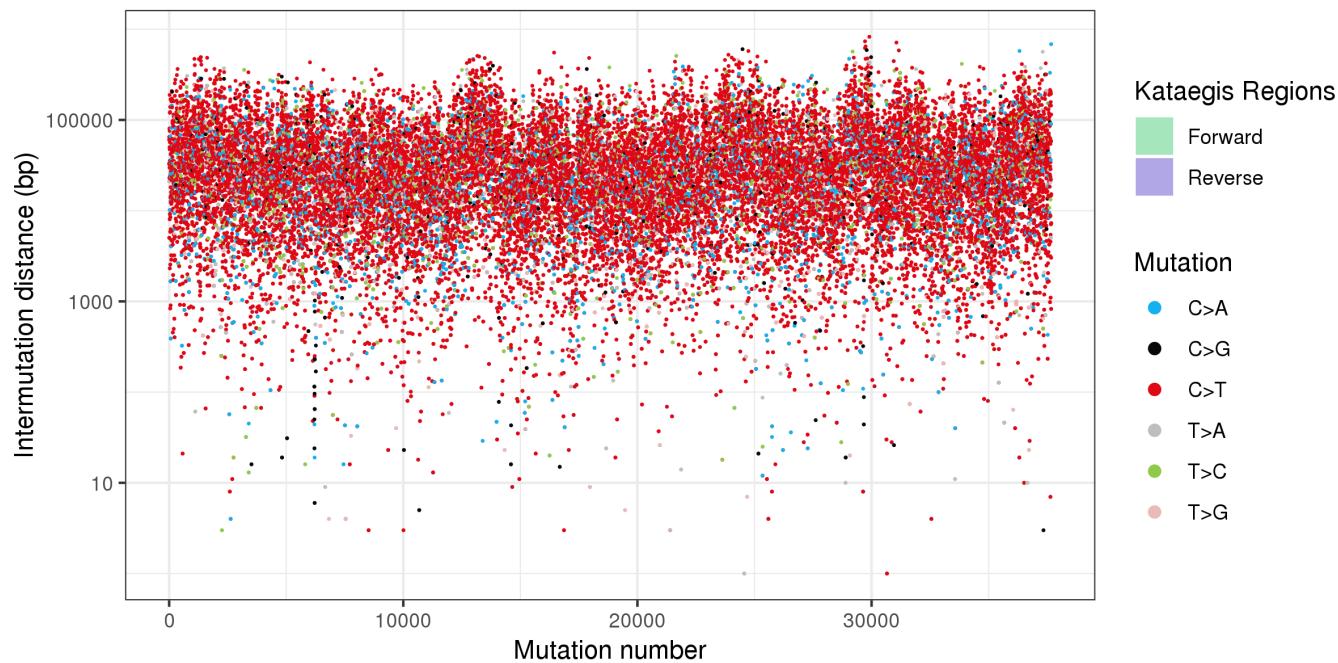
Driver variants (5)

VARIANT	CN	MACN	VCN	RNA VAF	BIALLELIC	HOTSPOT	DL	CL	PHASE
BRAF p.V600E	6.0	2.0	4.1	NA	No	Yes	100%	100%	
CDKN2A p.A68fs	2.0	0.0	2.0	NA	Yes	Near	100%	100%	
TERT c.-125_-124delCCinsTT	2.0	0.0	1.7	NA	Yes	Yes	100%	100%	482
SF3B1 p.P718L	3.0	1.0	2.0	NA	No	No	15%	100%	
TP63 p.M499I	4.0	2.0	1.7	NA	No	No	0%	100%	

Other potentially relevant coding variants (2)

VARIANT	CN	MACN	VCN	RNA VAF	BIALLELIC	HOTSPOT	DL	CL	PHASE
HDAC2 p.R409*	2.9	1.0	0.9	NA	No	No		100%	
STK19 p.D89N	3.8	1.8	2.0	NA	No	Yes		100%	

Kataegis plot



Driver amps/dels (1)

CHROMOSOME	REGION	GENE	TYPE	COPIES
10	q23.31	PTEN	partial loss	0

Other amps (0)

NONE



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Other dels on autosomes (1)

CHROMOSOME	REGION	GENE	TYPE	COPIES
16	q21	CNOT1	partial loss	0

Driver fusions (0)

NONE

Other potentially interesting fusions (0)

NONE

Driver viruses (0)

NONE

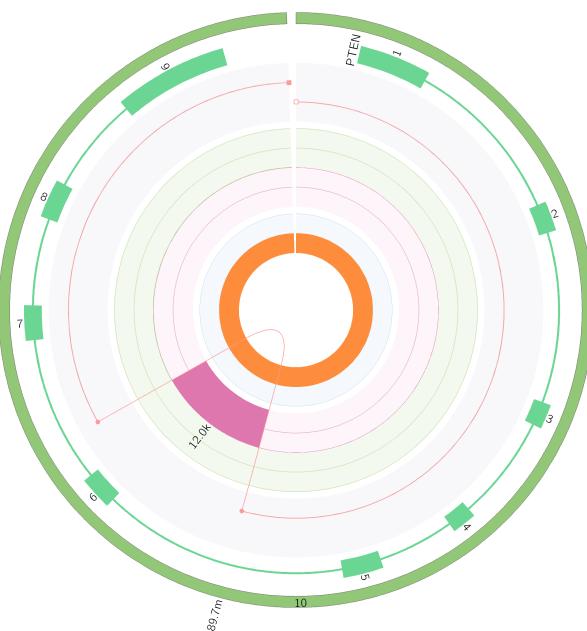
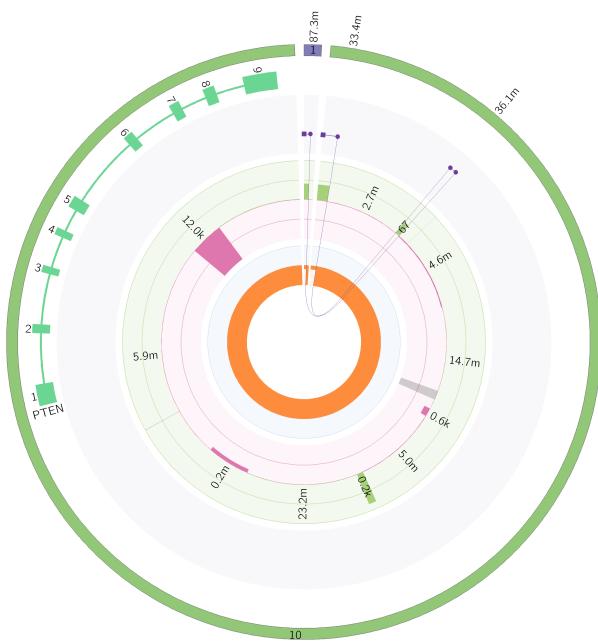
Other viral presence (0)

NONE

Gene disruptions (1)

LOCATION	GENE	RANGE	TYPE	JUNCTION CN	UNDISRUPTED CN
10q23.31	PTEN	Intron 5 -> Intron 6	DEL	2.0	0.0

Linx driver plots



CHR 1 [grey] [purple] [red] [green] CHR 10 [grey] [red] [green]

CHR 10 [grey] [red] [green]



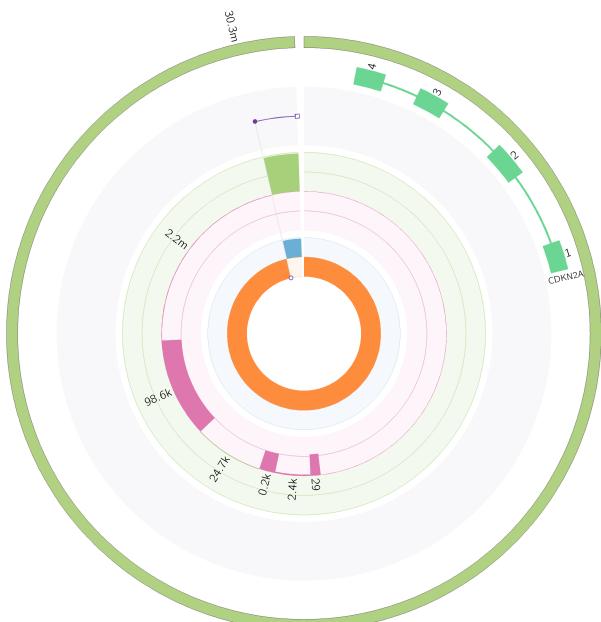
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Germline Findings

Driver variants (0)

NONE

TODO: Add other potentially other germline variants

Genes with missed variant likelihood > 1%

GENE	MVLH	GENE	MVLH	GENE	MVLH	GENE	MVLH
AMER1	3.8%	AR	2.6%	ARAF	2.0%	ATP2B3	2.0%
ATRX	3.1%	BCOR	2.2%	BTK	3.1%	CASZ1	3.7%
DDX3X	2.7%	EIF1AX	1.9%	GATA1	1.9%	KDM5C	1.7%
KDM6A	3.3%	KRTAP5-5	1.2%	MED12	3.1%	PHF6	3.9%
RBM10	2.1%	RPS6KA3	3.7%	STAG2	2.5%	ZFX	1.5%
ZMYM3	3.0%	ZRSR2	5.1%				

TODO: Add Germline CN aberrations

TODO: Add PEACH



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Immunology

TODO: Add HLA types for patient + status in tumor

TODO: Add list of neo-epitopes with predicted binding affinity

TODO: Add details about RNA tumor micro-environment



Cohort Comparison

TODO: Add cohort predictions for genomic position, signature, driver, expression and alternate splicing

TODO: Add detailed cohort incidence per driver

TODO: Add detailed cohort incidence per signature



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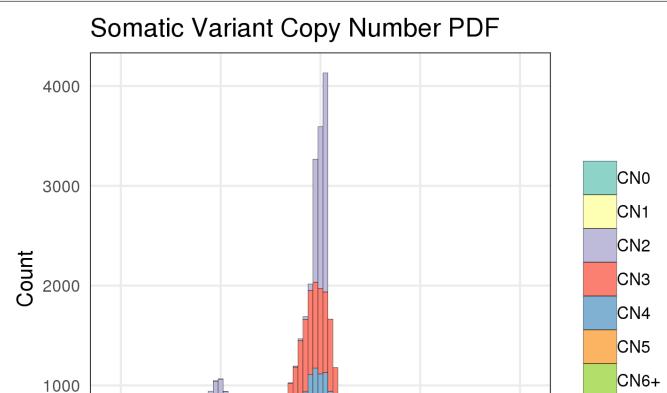
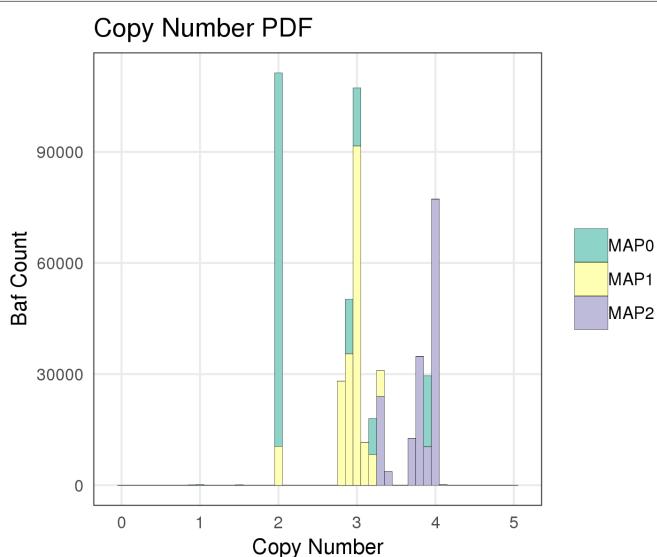
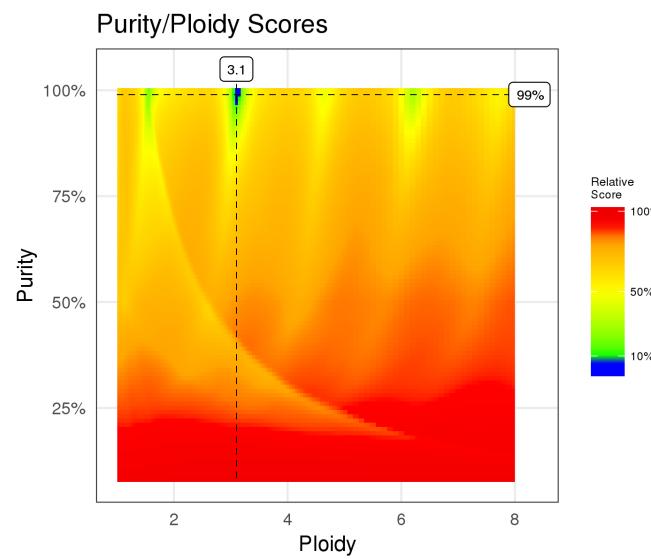
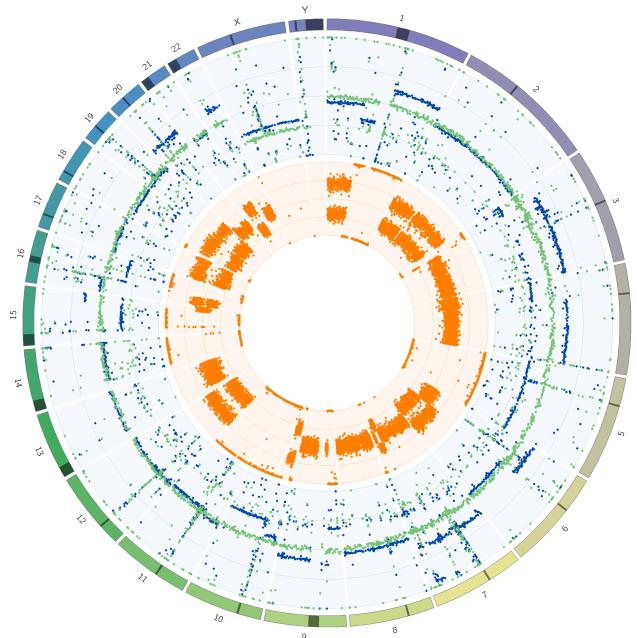
5.23

Quality Control

QC	AMBER MEAN DEPTH	CONTAMINATION	UNS. CN SEGMENTS	DELETED GENES
PASS	111	0%	0	2

TODO: Add Metrics / Flagstats

Purple QC plots



SAGE Reference Sample BQR plot



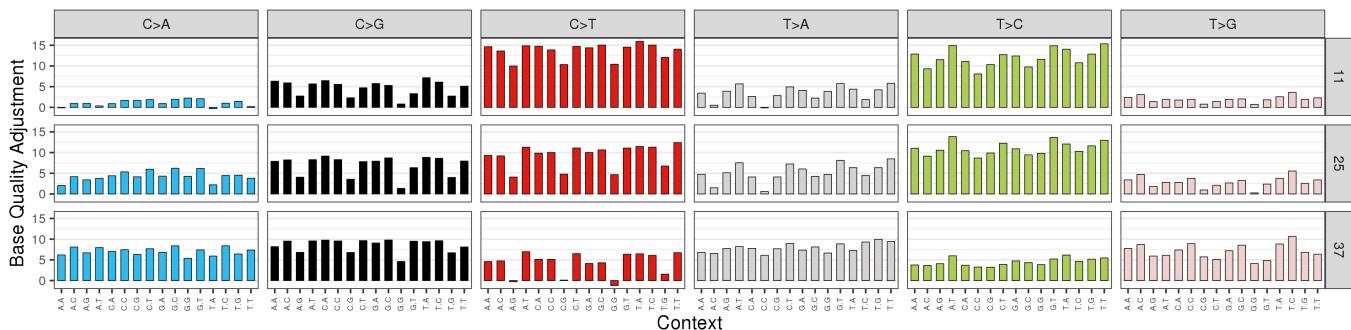
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SAGE Tumor Sample BQR plot

