

Table SI. Analytical tools used for next-generation sequencing data processing and variant analysis.

Software/tool	Version	Use	Supplier
bcl2fastq	v2.20	BCL→FASTQ, demultiplexing	Illumina
One-loop-trimmer	v1.1	Adapter removal, low-quality filtering	Burning Rock
BWA	v0.7.10	BWA-MEM reference genome alignment	Heng Li/BWA
GATK MarkDuplicates	v4.0	PCR duplicate identification and removal	Broad Institute
Sambamba	v0.8.0	BAM indexing and processing	Sambamba project
ABRA2	v2.22	Local realignment	ABRA2
VarDict	v1.5.1	SNV/INDEL calling	VarDict
VarScan	v2.4.3	False-positive filtering	VarScan
BRQC	-	Sampling QC	Burning Rock
BrasAnnotation	v4.3.0	Variant annotation	Burning Rock
BRVCS	-	Pathogenicity classification	Burning Rock
BRCNV	v4.3.0	CNV calling	Burning Rock
MarkSV	v0.3.1	SV calling	Burning Rock

BCL, base call file; FASTQ, a text-based format for storing nucleotide sequences; BWA-MEM, Burrows-Wheeler Aligner-Maximal Exact Match; PCR, polymerase chain reaction; BAM, binary alignment map; SNV, single-nucleotide variant; INDEL, insertion and deletion; QC, quality control; CNV, copy number variation; SV, structural variation.

Table SII. Quality control acceptance thresholds for tumor-only sample analysis.

Metric	Pass	Warning	Fail
Unique depth, x	≥ 250	50-<250	<50
Coverage uniformity, %	≥ 90	<90	N/A
Q30 ratio, %	≥ 80	75-<80	<75
Mean depth, x	≥ 500	100-<500	<100
Mapping ratio, %	≥ 90	<90	N/A

N/A, information not available.