

S5 Table. Example of parameter information in the GATK tool that extracts personally identifiable information

Copy Number Variant Discovery	
Tools that analyze read coverage to detect copy number variants.	
Name	Summary
AnnotateIntervals	Annotates intervals with GC content, mappability, and segmental-duplication content
CallCopyRatioSegments	Calls copy-ratio segments as amplified, deleted, or copy-number neutral
CreateReadCountPanelOfNormals	Creates a panel of normals for read-count denoising
DenoiseReadCounts	Denoises read counts to produce denoised copy ratios
DetermineGermlineContigPloidy	Determines the baseline contig ploidy for germline samples given counts data
FilterIntervals	Filters intervals based on annotations and/or count statistics
GermlineCNVCaller	Calls copy-number variants in germline samples given their counts and the output of DetermineGermlineContigPloidy
ModelSegments	Models segmented copy ratios from denoised copy ratios and segmented minor-allele fractions from allelic counts
PlotDenoisedCopyRatios	Creates plots of denoised copy ratios
PlotModeledSegments	Creates plots of denoised and segmented copy-ratio and minor-allele-fraction estimates
PostprocessGermlineCNVCalls GermlineCNVCaller	Postprocesses the output of GermlineCNVCaller and generates VCFs and denoised copy ratios
Read Data Manipulation	
Tools that manipulate read data in SAM, BAM or CRAM format.	
Name	Summary
SamToFastq (Picard)	Converts a SAM/BAM/CRAM file to FASTQ
Reference	
Tools that analyze and manipulate FASTA format references.	
Name	Summary
ComposeSTRTableFile	Composes a genome-wide STR location table used for DragSTR model auto-calibration
Short Variant Discovery	
Tools that perform variant calling and genotyping for short variants (SNPs, SNVs and Indels).	
Name	Summary
RampedHaplotypeCaller	**EXPERIMENTAL** Call germline SNPs and indels via local re-assembly of

haplotypes (ramped version)

Variant Annotations

Available to HaplotypeCaller, Mutect2, VariantAnnotator and GenotypeGVCFs.

Name	Summary
InbreedingCoeff:	Likelihood-based test for the consanguinity among samples (InbreedingCoeff)
TandemRepeat:	Tandem repeat unit composition and counts per allele (STR, RU, RPA)

SNP, single nucleotide polymorphism; SNV, single nucleotide variant; STR, short tandem repeat.