somatic (Somatic Variant Caller)

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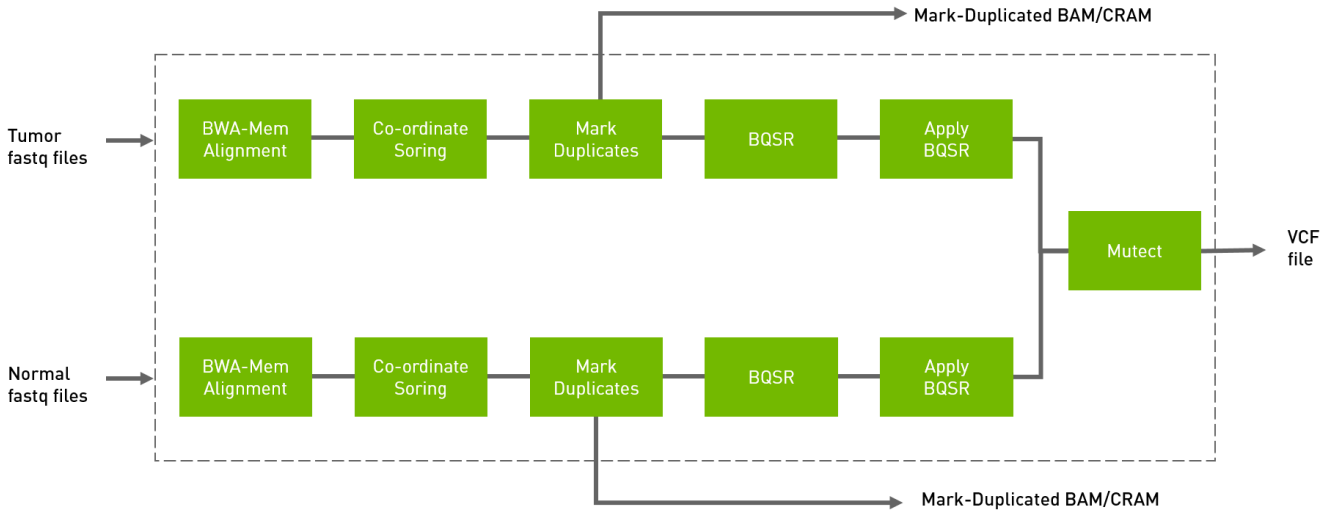
Run a somatic variant workflow.

The somatic tool processes the tumor FASTQ files, and optionally normal FASTQ files and knownSites files, and generates tumor or tumor/normal analysis. The output is in VCF format.

Internally the somatic tool runs several other Parabricks tools, thereby simplifying your work flow.



Quick Start

 # The command line below will run tumor-only analysis. # This command assumes all the inputs are in INPUT_DIR and all the outputs go to OUTPUT_DIR. docker run --rm --gpus all --volume INPUT_DIR:/workdir --volume OUTPUT_DIR:/outputdir \ --workdir /workdir \ nvcr.io/nvidia/clara/clara-parabricks:4.3.1-1 \ pbrun somatic \ --ref /workdir/${REFERENCE_FILE} \ --in-tumor-fq /workdir/${INPUT_FASTQ_1} /workdir/${INPUT_FASTQ_2} \ --bwa-options="-Y" \ --out-vcf /outputdir/${OUTPUT_VCF} \ --out-tumor-bam /outputdir/${OUTPUT_BAM} *# The command line below will run tumor-normal analysis. # This command assumes all the inputs are in INPUT_DIR and all the outputs go to OUTPUT_DIR.* docker run --rm --gpus all --volume INPUT_DIR:/workdir --volume OUTPUT_DIR:/outputdir \ --workdir

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/workdir \ nvcr.io/nvidia/clara/clara-parabricks:4.3.1-1 \ pbrun somatic \ --ref /workdir/${REFERENCE_FILE} \ --knownSites /workdir/${KNOWN_SITES_FILE} \ --in-tumor-fq /workdir/${INPUT_TUMOR_FASTQ_1} /workdir/${INPUT_TUMOR_FASTQ_2} "@RG\tID:sm_tumor_rg1\tLB:lib1\tPL:bar\tSM:sm_tumor\tPU:sm_tumor_rg1" \ --bwa-options="-Y" \ --out-vcf /outputdir/${OUTPUT_VCF} \ --out-tumor-bam /outputdir/${OUTPUT_TUMOR_BAM} \ --out-tumor-recal-file /outputdir/${OUTPUT_RECAL_FILE} \ --in-normal-fq /workdir/${INPUT_NORMAL_FASTQ_1} /workdir/${INPUT_NORMAL_FASTQ_2} "@RG\tID:sm_normal_rg1\tLB:lib1\tPL:bar\tSM:sm_normal\tPU:sm_normal_rg1" \ --out-normal-bam /outputdir/${OUTPUT_NORMAL_BAM}
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Compatible CPU Command

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# The commands below will run tumor-normal analysis. # # Run bwa mem on the tumor FASTQ files then sort the BAM by coordinates. $ bwa mem \ -t 32 \ -K 10000000 \ -Y \ -R '@RG\tID:sample_rg1\tLB:lib1\tPL:bar\tSM:sample\tPU:sample_rg1' \ ${REFERENCE_FILE} ${TUMOR_FASTQ_1} ${TUMOR_FASTQ_2} | \ gatk SortSam \ --java-options -Xmx30g \ --MAX_RECORDS_IN_RAM 5000000 \ -I /dev/stdin \ -O tumor_cpu.bam \ --SORT_ORDER coordinate # Mark duplicates. $ gatk MarkDuplicates \ --java-options -Xmx30g \ -I tumor_cpu.bam \ -O tumor_mark_dups_cpu.bam \ -M tumor_metrics.txt # Generate a BQSR report. $ gatk BaseRecalibrator \ --java-options -Xmx30g \ --input tumor_mark_dups_cpu.bam \ --output ${OUTPUT_TUMOR_RECAL_FILE} \ --known-sites ${KNOWN_SITES_FILE} \ --reference ${REFERENCE_FILE} # Apply the BQSR report. $ gatk ApplyBQSR \ --java-options -Xmx30g \ -R ${REFERENCE_FILE} \ -I tumor_cpu.bam \ --bqsr-recal-file ${TUMOR_OUTPUT_RECAL_FILE} \ -O ${OUTPUT_TUMOR_BAM} # Now repeat all the above steps, only with the normal FASTQ data. $ bwa mem \ -t 32 \ -K 10000000 \ -Y \ -R '@RG\tID:sample_rg1\tLB:lib1\tPL:bar\tSM:sample\tPU:sample_rg1' \ ${REFERENCE_FILE} ${NORMAL_FASTQ_1} ${NORMAL_FASTQ_2} | \ gatk SortSam \ --java-options -Xmx30g \ --MAX_RECORDS_IN_RAM 5000000 \ -I /dev/stdin \ -O normal_cpu.bam \ --SORT_ORDER coordinate # Mark duplicates. $ gatk MarkDuplicates \ --java-options -Xmx30g \ -I normal_cpu.bam \ -O normal_mark_dups_cpu.bam \ -M normal_metrics.txt # Generate a BQSR report. $ gatk BaseRecalibrator \ --java-options -Xmx30g \ --input normal_mark_dups_cpu.bam \ --output ${OUTPUT_NORMAL_RECAL_FILE} \ --known-
```

sites ${KNOWN_SITES_FILE} \ --reference ${REFERENCE_FILE} *# Apply the BQSR report.* $ gatk ApplyBQSR \ --java-options -Xmx30g \ -R ${REFERENCE_FILE} \ -I normal_cpu.bam \ --bqsr-recal-file ${OUTPUT_NORMAL_RECAL_FILE} \ -O ${OUTPUT_NORMAL_BAM} *# Finally, run Mutect2 on the normal and tumor data.* $ gatk Mutect2 \ -R ${REFERENCE_FILE} \ --input ${OUTPUT_TUMOR_BAM} \ --tumor-sample tumor \ --input ${OUTPUT_NORMAL_BAM} \ --normal-sample normal \ --output ${OUTPUT_VCF}

somatic Reference

Run the tumor normal somatic pipeline from FASTQ to VCF.

Input/Output file options

--ref REF

Path to the reference file. (default: None)

Option is required.

--in-tumor-fq [IN_TUMOR_FQ ...]

Path to the pair-ended FASTQ files followed by optional read group with quotes (Example: "@RG\tID:foo\tLB:lib1\tPL:bar\tSM:20"). The files can be in fastq or fastq.gz format. Either all sets of inputs have a read group, or none should have one, and it will be automatically added by the pipeline. This option can be repeated multiple times. Example 1: --in-tumor-fq sampleX_1_1.fastq.gz sampleX_1_2.fastq.gz --in-tumor-fq sampleX_2_1.fastq.gz sampleX_2_2.fastq.gz. Example 2: --in-tumor-fq sampleX_1_1.fastq.gz sampleX_1_2.fastq.gz "@RG ID:foo\tLB:lib1\tPL:bar\tSM:sm_tumor\tPU:unit1" --in-tumor-fq sampleX_2_1.fastq.gz sampleX_2_2.fastq.gz "@RG ID:foo2\tLB:lib1\tPL:bar\tSM:sm_tumor\tPU:unit2". For the same sample, Read Groups should have the same sample name (SM) and a different ID and PU. (default: None)

--in-se-tumor-fq [IN_SE_TUMOR_FQ ...]

Path to the single-ended FASTQ file followed by an optional read group with quotes (Example: "@RG\tID:foo\tLB:lib1\tPL:bar\tSM:sample\tPU:foo"). The file must be in fastq or fastq.gz format. Either all sets of inputs have a read group, or none should have one; if

no read group is provided, one will be added automatically by the pipeline. This option can be repeated multiple times. Example 1: --in-se-tumor-fq sampleX_1.fastq.gz --in-se-tumor-fq sampleX_2.fastq.gz . Example 2: --in-se-tumor-fq sampleX_1.fastq.gz "@RG\tID:foo\tLB:lib1\tPL:bar\tSM:tumor\tPU:unit1" --in-se-tumor-fq sampleX_2.fastq.gz "@RG\tID:foo2\tLB:lib1\tPL:bar\tSM:tumor\tPU:unit2" . For the same sample, Read Groups should have the same sample name (SM) and a different ID and PU. (default: None)
--in-normal-fq [IN_NORMAL_FQ ...]

Path to the pair-ended FASTQ files followed by an optional read group with quotes (Example: "@RG\tID:foo\tLB:lib1\tPL:bar\tSM:20"). The files must be in fastq or fastq.gz format. Either all sets of inputs have a read group, or none should have one; if no read group is provided, one will be automatically added by the pipeline. This option can be repeated multiple times. Example 1: --in-normal-fq sampleX_1_1.fastq.gz sampleX_1_2.fastq.gz --in-fq sampleX_2_1.fastq.gz sampleX_2_2.fastq.gz . Example 2: --in-normal-fq sampleX_1_1.fastq.gz sampleX_1_2.fastq.gz "@RG ID:foo\tLB:lib1\tPL:bar\tSM:sm_normal\tPU:unit1" --in-normal-fq sampleX_2_1.fastq.gz sampleX_2_2.fastq.gz "@RG ID:foo2\tLB:lib1\tPL:bar\tSM:sm_normal\tPU:unit2". For the same sample, Read Groups should have the same sample name (SM) and a different ID and PU. (default: None)

--in-se-normal-fq [IN_SE_NORMAL_FQ ...]

Path to the single-ended FASTQ file followed by optional read group with quotes (Example: "@RG\tID:foo\tLB:lib1\tPL:bar\tSM:sample\tPU:foo"). The file must be in fastq or fastq.gz format. Either all sets of inputs have a read group, or none should have one; if no read group is provided, one will be added automatically by the pipeline. This option can be repeated multiple times. Example 1: --in-se-normal-fq sampleX_1.fastq.gz --in-se-normal-fq sampleX_2.fastq.gz . Example 2: --in-se-normal-fq sampleX_1.fastq.gz "@RG\tID:foo\tLB:lib1\tPL:bar\tSM:normal\tPU:unit1" --in-se-normal-fq sampleX_2.fastq.gz "@RG\tID:foo2\tLB:lib1\tPL:bar\tSM:normal\tPU:unit2" . For the same sample, Read Groups should have the same sample name (SM) and a different ID and PU. (default: None)

--knownSites KNOWNSITES

Path to a known indels file. The file must be in vcf.gz format. This option can be used multiple times. (default: None)

--interval-file INTERVAL_FILE

Path to an interval file in one of these formats: Picard-style (.interval_list or .picard), GATK-style (.list or .intervals), or BED file (.bed). This option can be used multiple times. (default: None)

--out-vcf OUT_VCF

Path of the VCF file after Variant Calling. (default: None)

Option is required.

--out-tumor-bam OUT_TUMOR_BAM

Path of the BAM file for tumor reads. (default: None)

Option is required.

--out-normal-bam OUT_NORMAL_BAM

Path of the BAM file for normal reads. (default: None)

--mutect-bam-output MUTECT_BAM_OUTPUT

File to which assembled haplotypes should be written in Mutect. (default: None)

--out-tumor-recal-file OUT_TUMOR_RECAL_FILE

Path of the report file after Base Quality Score Recalibration for tumor sample. (default: None)

--out-normal-recal-file OUT_NORMAL_RECAL_FILE

Path of the report file after Base Quality Score Recalibration for normal sample. (default: None)

--mutect-germline-resource MUTECT_GERMLINE_RESOURCE

Path of the vcf.gz germline resource file. Population vcf of germline sequencing containing allele fractions. (default: None)

--mutect-alleles MUTECT_ALLELES

Path of the vcf.gz force-call file. The set of alleles to force-call regardless of evidence. (default: None)

Tool Options:

-L INTERVAL, --interval INTERVAL

Interval within which to call bqsr from the input reads. All intervals will have a padding of 100 to get read records, and overlapping intervals will be combined. Interval files should be passed using the --interval-file option. This option can be used multiple times e.g. "-L chr1 -L chr2:10000 -L chr3:20000+ -L chr4:10000-20000". (default: None)

--bwa-options BWA_OPTIONS

Pass supported bwa mem options as one string. The current original bwa mem supported options are -M, -Y and -T e.g. --bwa-options="-M -Y" (default: None)

--no-warnings

Suppress warning messages about system thread and memory usage. (default: None)

--filter-flag FILTER_FLAG

Don't generate SAM entries in the output if the entry's flag's meet this criteria. Criteria: (flag & filter != 0) (default: 0)

--skip-multiple-hits

Filter SAM entries whose length of SA is not 0. (default: None)

--min-read-length MIN_READ_LENGTH

Skip reads below minimum read length. They will not be part of the output. (default: None)

--align-only

Generate output BAM after bwa-mem. The output will not be co-ordinate sorted or duplicates will not be marked. (default: None)

--no-markdups

Do not perform the Mark Duplicates step. Return BAM after sorting. (default: None)

--fix-mate

Add mate cigar (MC) and mate quality (MQ) tags to the output file. (default: None)

--markdups-assume-sortorder-queryname

Assume the reads are sorted by queryname for Marking Duplicates. This will mark secondary, supplementary, and unmapped reads as duplicates as well. This flag will not impact variant calling while increasing processing times. (default: None)

--markdups-picard-version-2182

Assume marking duplicates to be similar to Picard version 2.18.2. (default: None)

--monitor-usage

Monitor approximate CPU utilization and host memory usage during execution. (default: None)

--optical-duplicate-pixel-distance OPTICAL_DUPLICATE_PIXEL_DISTANCE

The maximum offset between two duplicate clusters in order to consider them optical duplicates. Ignored if --out-duplicate-metrics is not passed. (default: None)

-ip INTERVAL_PADDING, --interval-padding INTERVAL_PADDING

Amount of padding (in base pairs) to add to each interval you are including. (default: None)

--standalone-bqsr

Run standalone BQSR. (default: None)

--max-read-length-fq2bamfast MAX_READ_LENGTH_FQ2BAMFAST

Maximum read length/size (i.e., sequence length) used for bwa and filtering FASTQ input (Argument only applies to --fq2bamfast) (default: 480)

--min-read-length-fq2bamfast MIN_READ_LENGTH_FQ2BAMFAST

Minimum read length/size (i.e., sequence length) used for bwa and filtering FASTQ input (Argument only applies to --fq2bamfast) (default: 10)

--max-mnp-distance MAX_MNP_DISTANCE

Two or more phased substitutions separated by this distance or less are merged into MNPs. (default: 1)

--mutectcaller-options MUTECTCALLER_OPTIONS

Pass supported mutectcaller options as one string. The following are currently supported original mutectcaller options: -pcr-indel-model <NONE, HOSTILE, AGGRESSIVE, CONSERVATIVE>, -max-reads-per-alignment-start <int>, (e.g. --mutectcaller-options="-pcr-indel-model HOSTILE -max-reads-per-alignment-start 30"). (default: None)

--initial-tumor-lod INITIAL_TUMOR_LOD

Log 10 odds threshold to consider pileup active. (default: None)

--tumor-lod-to-emit TUMOR_LOD_TO_EMIT

Log 10 odds threshold to emit variant to VCF. (default: None)

--pruning-lod-threshold PRUNING_LOD_THRESHOLD

Ln likelihood ratio threshold for adaptive pruning algorithm. (default: None)

--active-probability-threshold ACTIVE_PROBABILITY_THRESHOLD

Minimum probability for a locus to be considered active. (default: None)

--no-alt-contigs

Ignore commonly known alternate contigs. (default: None)

--genotype-germline-sites

Call all apparent germline site even though they will ultimately be filtered. (default: None)

--genotype-pon-sites

Call sites in the PoN even though they will ultimately be filtered. (default: None)

--force-call-filtered-alleles

Force-call filtered alleles included in the resource specified by --alleles. (default: None)

--tumor-read-group-sm TUMOR_READ_GROUP_SM

SM tag for read groups for tumor sample. (default: None)

--tumor-read-group-lb TUMOR_READ_GROUP_LB

LB tag for read groups for tumor sample. (default: None)

--tumor-read-group-pl TUMOR_READ_GROUP_PL

PL tag for read groups for tumor sample. (default: None)

--tumor-read-group-id-prefix TUMOR_READ_GROUP_ID_PREFIX

Prefix for ID and PU tag for read groups for tumor sample. This prefix will be used for all pair of tumor FASTQ files in this run. The ID and PU tag will consist of this prefix and an identifier which will be unique for a pair of FASTQ files. (default: None)

--normal-read-group-sm NORMAL_READ_GROUP_SM

SM tag for read groups for normal sample. (default: None)

--normal-read-group-lb NORMAL_READ_GROUP_LB

LB tag for read groups for normal sample. (default: None)

--normal-read-group-pl NORMAL_READ_GROUP_PL

PL tag for read groups for normal sample. (default: None)

--normal-read-group-id-prefix NORMAL_READ_GROUP_ID_PREFIX

Prefix for ID and PU tags for read groups of a normal sample. This prefix will be used for all pairs of normal FASTQ files in this run. The ID and PU tags will consist of this prefix and an identifier that will be unique for a pair of FASTQ files. (default: None)

Performance Options:

--fq2bamfast

Use fq2bamfast as the alignment tool instead of fq2bam (default: None)

--gpuwrite

Use one GPU to accelerate writing final BAM. (default: None)

--gpuwrite-deflate-algo GPUWRITE_DEFLATE_ALGO

Choose the nvCOMP DEFLATE algorithm to use with --gpuwrite. Note these options do not correspond to CPU DEFLATE options. Valid options are 0 and 3. Option 0 is faster while option 3 provides a better compression ratio. (default=0) (default: None)

--gpusort

Use GPUs to accelerate sorting and marking. (default: None)

--use-gds

Use GPUDirect Storage (GDS) to enable a direct data path for direct memory access (DMA) transfers between GPU memory and storage. Must be used concurrently with *--gpuwrite*. Please refer to Parabricks Documentation > Best Performance for information on how to set up and use GPUDirect Storage. (default: None)

--memory-limit MEMORY_LIMIT

System memory limit in GBs during sorting and postsorting. By default, the limit is half of the total system memory. (default: 62)

--low-memory

Use low memory mode (default: None)

--num-cpu-threads-per-stage NUM_CPU_THREADS_PER_STAGE

Number of CPU threads to use per stage. (default: 8)

--bwa-nstreams BWA_NSTREAMS

Number of streams per GPU to use; note: more streams increases device memory usage (Argument only applies to --fq2bamfast) (default: 4)

--bwa-cpu-thread-pool BWA_CPU_THREAD_POOL

Number of threads to devote to CPU thread pool *per GPU* (Argument only applies to --fq2bamfast) (default: 16)

--mutect-low-memory

Use low memory mode in mutect caller. (default: None)

--run-partition

Turn on partition mode; divides genome into multiple partitions and runs 1 process per partition. (default: None)

--gpu-num-per-partition GPU_NUM_PER_PARTITION

Number of GPUs to use per partition. (default: None)

--num-htvc-threads NUM_HTVC_THREADS

Number of CPU threads to use. (default: 5)

Common options:

--logfile LOGFILE

Path to the log file. If not specified, messages will only be written to the standard error output. (default: None)

--tmp-dir TMP_DIR

Full path to the directory where temporary files will be stored.

--with-petagene-dir WITH_PETAGENE_DIR

Full path to the PetaGene installation directory. By default, this should have been installed at /opt/petagene. Use of this option also requires that the PetaLink library has been preloaded by setting the LD_PRELOAD environment variable. Optionally set the

PETASUITE_REFPATH and PGCLOUD_CREDPATH environment variables that are used for data and credentials (default: None)
--keep-tmp

Do not delete the directory storing temporary files after completion.

--no-seccomp-override

Do not override seccomp options for docker (default: None).

--version

View compatible software versions.

GPU options:

--num-gpus NUM_GPUS

Number of GPUs to use for a run. GPUs 0..(NUM_GPUS-1) will be used.

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