```
INPUT_DIR=          # Directory path for the original fastq files
OUTPUT_DIR=         # Directory path for the outputs of the analysis
OUTPUT_NAME=        # Name for analysis. Choose a single name without spaces
LIST_SAMPLES=       # File directory for a list with samples and individual id, separated by space. Use LIST_SAMPLES=none if no list is
                      available.
ANALYSIS=           # ANALYSIS=1 (for WGS) or ANALYSIS=2 (for RNAseq)
EXTENSION_SE=       # For single-end fastq files, write the extension of original fastq files. If paired- end files, le" blank (EXTENSION_SE= ).
EXTENSION_PE1=      # For paired-end fastq files, write the extension of PE1 fastq files. If single-end files, le" blank (EXTENSION_PE1= ).
EXTENSION_PE2=      # For paired-end fastq files, write the extension of PE2 fastq files. If single-end files, le" blank (EXTENSION_PE2= ).
```

```
ASSEMBLY=       # Give a name for the reference genome. Eg ASSEMBLY=hg38.
REFERENCE=      # File directory for reference genome (*.fa). The reference genome must be uncompressed file and not (*.fa.gz).
VARIANTS=       # File directory for variants file (*.vcf.gz) for the reference genome. Use VARIANTS=none if no variants is available.
                  The variants must be (*.vcf.gz) and not (*.vcf).
ANNOTATION=     # File directory for annotation file (*.gtf) for the reference genome. Use ANNOTATION=none if no annotation is available.
                  The annotation must be uncompressed file and not (*.gtf.gz)
```

```
STEP_QC_TRIM=          # Activate quality control and adapter trimming step? STEP_QC_TRIM=yes or STEP_QC_TRIM=no (Default yes)
STEP_ALIGNMENT=        # Activate alignment step? STEP_ALIGNMENT=yes or STEP_ALIGNMENT=no (Default yes)
STEP_GENECOUNT=        # Activate gene count step? STEP_GENECOUNT=yes or STEP_GENECOUNT=no (Default: yes for RNAseq
                         and no for WGS)
STEP_MDUP=             # Activate mark duplicated read step? STEP_MDUP=yes or STEP_MDUP=no (Default: yes). It includes
                         splitncigar step for RNAseq
STEP_BQSR=             # Activate base quality score recalibration step? STEP_BQSR=yes or STEP_BQSR=no (Default: yes)
STEP_VCF_CALL=         # Activate variant calling step? STEP_VCF_CALL=yes or STEP_VCF_CALL=no (Default: yes)
STEP_VCF_FILTER=       # Activate vcf filtering step? STEP_VCF_FILTER=yes or STEP_VCF_FILTER=no (Default: yes)
STEP_VCF_COMBINE=      # Activate combine filtered snps from different vcfs? STEP_VCF_COMBINE=yes or STEP_VCF_COMBINE=no
                         (Default: yes)
STEP_VCF_ANNOTATE=     # Activate annotation of vcf files? STEP_VCF_ANNOTATE=yes or STEP_VCF_ANNOTATE=no (Default: yes)
```

```
ALIGNER_PROGRAM=    # Choose programs for alignment step. If more than one program are chosen, use ‘,’ between them.
                      (Options: bbmap, bowtie, bowtie2, bwa, bwa2, gsnap, hisat2, star)
CALLER_PROGRAM=     # Choose programs for variant calling step. If more than one program are chosen, use ‘,’ between them.
                      (Options: bcftools, freebayes, freebayes-parallel, gatk4, gatk4GenomicsDBImport, gatk4CombineGVCFs,
                                platypus, varscan)
GENECOUNT_PROGRAM=  # Choose programs for gene count step. If more than one program are chosen, use ‘,’ between them.
                      (Options: htseq, featurecounts). Use GENECOUNT_PROGRAM=none if you do not want gene counts.
FEATURE_TYPE=       # Any feature of the third column of gtf file. (Default FEATURE_TYPE=exon).  Use FEATURE_TYPE=none if
                      you do not want gene counts.


CALL_BY_CHROM=      # Should variant calling be done by chromosome? CALL_BY_CHROM=no or CALL_BY_CHROM=yes (Default no).
MIN_READ_LENGTH=    # Minimum read length to be kept after adapter trimming (Default 36).
MAX_READ_LENGTH=    # Maximum read length to be kept after adapter trimming (Default 150).
DOWN_SAMPLING_FASTQ= # Down sampling the fastq files. (Default 0). DOWN_SAMPLING_FASTQ=0 means no reads sampling from
                      fastq files. Any number between 0.01 and 0.99 means to sampling the fraction of reads from fastq files. Any
                      number equal or greater than 1 means to sampling the number of reads from fastq file.
MIN_DEPTH=          # Minimum locus coverage for the filtering step (Default 3).
COMBINE_VCF=        # COMBINEVCF=none : do not combine different vcfs.
                    # COMBINEVCF=partial : If it was used more than one program for variant calling, combine common SNPs that
                      appeared AT LEAST in two different vcf files.
                    # COMBINEVCF=full : If it was used more than one program for variant calling, combine common SNPs that
                      appeared IN ALL the vcf files.
THREADS=            # Number of threads for analysis.
BATCH=              # Number of samples to be processed at same time.
KEEP_LOG=           # Keep the log files in the output folder? KEEP_LOG=no or KEEP_LOG=yes (Default no).
KEEP_INTERMEDIATE_DATA=   # Keep intermediate fastq and bam files in the output folder? KEEP_INTERMEDIATE_DATA=no
                            or KEEP_INTERMEDIATE_DATA=yes (Default no).
```