

Fastq Read Files

Bwa aln

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User Defined

Will do N=8 lanes

User Defined

Bwa sampe or Bwa samse

Bwa sampe or Bwa samse

Samtools SAM-->BAM

Samtools SAM-->BAM

Picard Sort

Picard Sort

Picard MergeSAMfiles

Will do N=8 lanes

GATK Realign Target Creator

GATK Indel Realigner

GATK Count Covariants

GATK Table Recalibrator

Picard Mark Duplicates

Required Input Files

- runparameters.ini
- samplesheet.txt
- Bwa Indexed genome
- Faidx indexed genome
- GATK FTP dbSNP VCF
- reflat file for genome
- bait intervals file
- target intervals file
- GATK regions to call file
- GATK FTP HapMap Sites
- GATK FTP OMNI Sites
- Samtools regions to call file
- GATK FTP 1000G Sites
- Cosmic VCF
- SnpEff database for genome
- List of SnpEff non-synonymous effects
- Mutated Genes in Disease list

Samtools Flag Stat

- Picard CollectAlignmentSummaryMetrics
- Picard MeanQualityByCycle
- Picard QualityScoreDistribution
- Picard CollectInsertSizeMetrics
- Picard CollectRnaSeqMetrics
- Picard CalculateHsMetrics

GATK UnifiedGenotyper "SNP"

GATK UnifiedGenotyper "INDEL"

Samtools mpileup

GATK VariantRecalibrator

Bcftools view

GATK ApplyRecalibration

GATK VariantFiltration

vcfutils.pl

SnpSift filter "INDEL"

grep -v "INDEL"

Samtools Indels

Samtools SNV

GATK VariantFiltration

GATK VariantFiltration

GATK CombineVariants

GATK CombineVariants

dbSNP GATK VariantEval

1000G GATK VariantEval

GATK CombineVariants

SnpEff eff

dbSNP
1000G
Cosmic

GATK VariantAnnotator

SnpSift filter "Intersection"

grep -v "1000G"