GATK Test Protocol

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Abstract

Analysis scripts of GATK pipeline

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Protocol

GATK3 scripts

Step 1.

alignment

SOFTWARE PACKAGE (CentOS -)

bwa, 0.7.15-r1140

cmd COMMAND (CentOS - 7.3.1611)

bwa mem -R "@RG\tID:NA12878-test\tSM:NA12878-test\tLB:NA12878-test\tPL:illumina\tCN:BGI" - M -

t 48 ${refpath}/human_g1k_v37.fasta ${datapath}/ERR194147_1.fastq.gz ${datapath}/ERR194147_2.fastq.gz | samtools view -S -b -o ${alignment}/na12878.raw.bam - alignment$

EXPECTED RESULTS

\${alignment}/na12878.sort.bam

GATK3 scripts

Step 2.

rmdup

SOFTWARE PACKAGE

cmd COMMAND

samtools index \${alignment}/na12878.sort.dup.bam
bamindex

EXPECTED RESULTS

\${alignment}/na12878.sort.dup.bam

GATK3 scripts

Step 3.

BQSR

SOFTWARE PACKAGE (CentOS -)

GATK, 3.7

cmd COMMAND

java -Xmx250g -jar $\frac{genomeAnalysisTK.jar -R {refpath}/human_g1k_v37.fasta -T BaseRecalibrator -nct 48 -I <math>\frac{1s}{alignment}/na12878.sort.dup.bam -knownSites {refpath}/dbsnp_138.b37.vcf -o {alignment}/na12878.recal_data.grp BOSR$

GATK3 scripts

Step 4.

Printreads

SOFTWARE PACKAGE (CentOS -)

GATK, 3.7

cmd COMMAND

java -Xmx250g -jar \${software}/GenomeAnalysisTK.jar -R \${refpath}/human_g1k_v37.fasta T PrintReads -nct 7 -filterNoBases -I \${alignment}/na12878.sort.dup.bam BQSR \${alignment}/na12878.recal_data.grp -o \${alignment}/na12878.sort.recal.bam
Printreads

GATK3 scripts

Step 5.

Variation

SOFTWARE PACKAGE (CentOS -)

GATK, 3.7

cmd COMMAND

```
java -Xmx250g -jar ${GATK}/GenomeAnalysisTK.jar -l INFO -
R ${refpath}/human_g1k_v37.fasta -T HaplotypeCaller -rf BadCigar -
I ${alignment}/na12878.final.bam -o ${variation}/na12878.vcf.gz -nct 48 -
D ${refpath}/dbsnp_138.b37.vcf
Variation
```