



Informatics on High-throughput Sequencing Data

(Summer Course 2020)

Day 19



SAM tools

- ▶ `./samtools mpileup -v -u -f /Users/sarael-metwally/Documents/Summer/bwa/wu_0.v7.fas sample.sorted.bam > sample.vcf`
- ▶ `./samtools mpileup -g -f /Users/sarael-metwally/Documents/Summer/bwa/wu_0.v7.fas sample.sorted.bam > sample.bcf`

bcf tools

Download and install → BCFtools

vcfutils.pl is part of bcf tools

<http://www.htslib.org/download/>

create download directory for BCFtools

```
mkdir bcf tools_download
```

```
cd bcf tools_download/
```

download latest version

```
wget https://github.com/samtools/bcf tools/releases/download/1.10.2/bcf tools-1.10.2.tar.bz2
```

```
tar -jvxf bcf tools-1.10.2.tar.bz2
```

```
cd bcf tools-1.10.2
```

```
make
```

```
make prefix=$HOME/tools/bcf tools install # path where to install BCFtools
```

add path to your .bashrc file

```
export PATH=$HOME/tools/bcf tools/bin/:$PATH
```

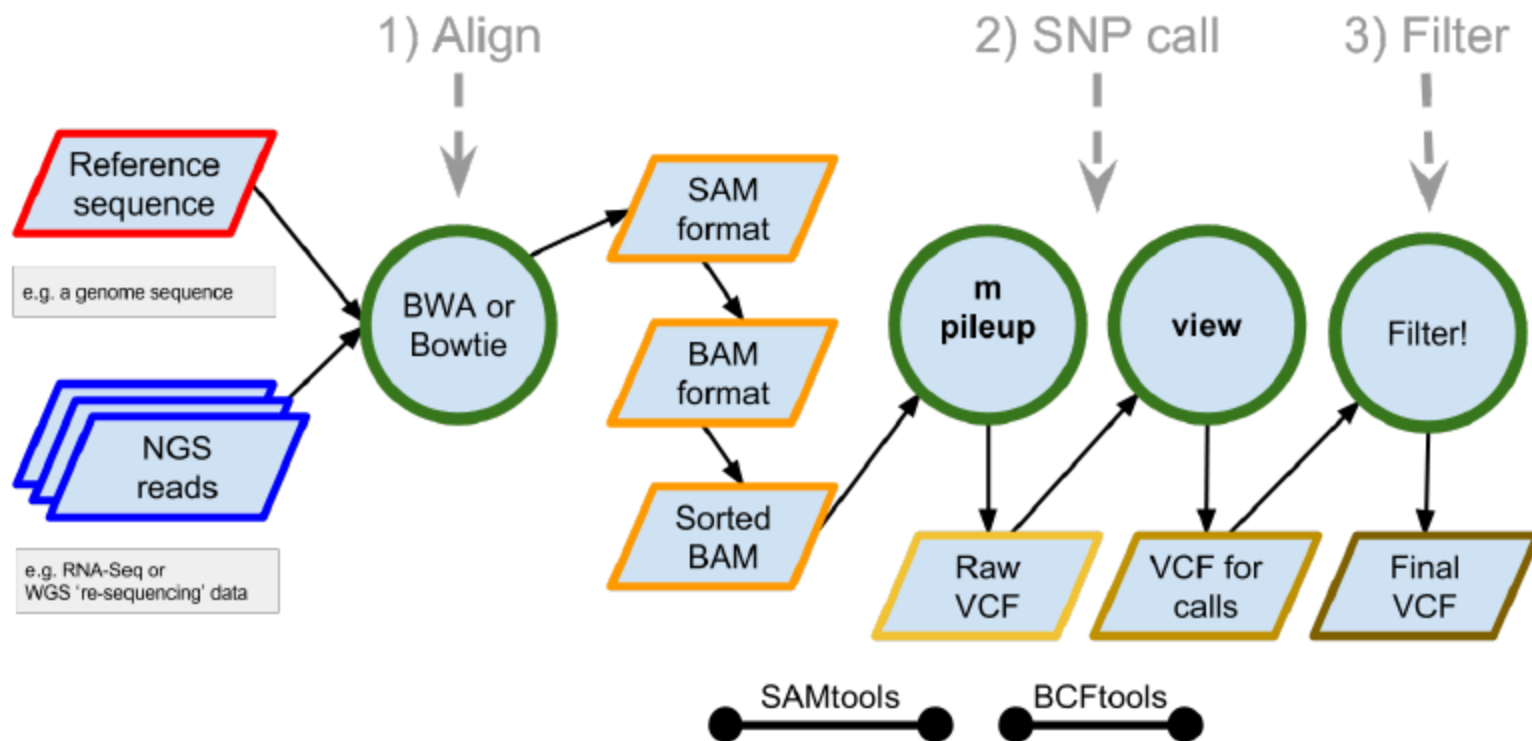
<http://www.htslib.org/download/>

bcf tools

- ▶ `./bcftools view sample.bcf`
- ▶ `./bcftools call -v -m -O v -o sample.vcf sample.bcf`

Variant Call Pipeline

Pipeline overview



Slides credit: EMBL-EBI

Variant Call Pipeline

- ▶ `./samtools tview sample.sorted.bam ref.fasta (g,arrows,?,q, Goto: CP000819.1:50)`
- ▶ `./bcftools mpileup -O b -o results.raw.bcf -f ref.fasta sample.sorted.bam`
- ▶ `./bcftools call --ploidy 1 -m -v -o results.vcf results.raw.bcf`
- ▶ `./vcfutils.pl varFilter results.vcf > results.filter.vcf`

Thanks!

// | ?