

Exercise 6

Ewa Szczurek
MIM UW

November 10, 2015

Exercise 1. SNP calling with SAM tools and Galaxy.

- Access source files for today.
- Pileup the mapped reads in the BAM file

```
NGS:SAM Tools>Mpileup
```

```
BAM file: The BAM file you generated
```

```
Using reference genome: hg19
```

```
Genotype Likelihood Computation: Perform a genotype likelihood computation
```

```
Perform INDEL calling: Do not perform indel calling
```

```
Set advanced options: Advanced
```

```
List of regions or sites on which to operate: chr20.bed
```

```
Execute
```

- Mpileup creates a BCF (binary VCF) file which contains the list of variations in a compressed format.
- Convert BCF to VCF
 - Download the BCF file
 - In the samtools directory

```
bcftools view DOWNLOADED_FILE.BCF > Mpileup_chr20.vcf
```

Homework 1. No homework as of today!