High Performance Computing for Genomics

Part II: for Genomics

KU LEUVEN GENOMICS CORE UZ LEUVEN

Project description

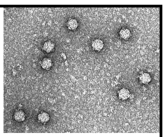
Assume this population study:
We have 2 populations, population 1 has 14 individuals, population 2 only 5.
There is no reference genome available.
Are there 'diagnostic' variations between the 2 populations.

Project parts:

- Assembly
- Mapping
- Variant calling

KU LEUVEN GENOMICS CORE UZ LEUVEN

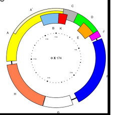
Actual data



Population 1: Sequenced PhiX in the Genomics Core
Population 2: Simulated data from the ncbi reference genome

Phi X 174:

- Bacteriophage
- First sequenced DNA virus (1977)
- Circular, single stranded DNA genome of 5386bp, GCcontent 44%
- 95% are coding genes, total of 11 genes
- Used as positive control in Illumina sequencing



KU LEUVEN GENOMICS CORE UZ LEUVEN

Project description

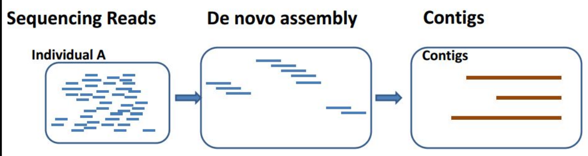
Project parts:

- **Assembly**
  Population 1 has most individuals, so best to choose one of these individuals.
- Mapping
- Variant calling

Assembly

Simple description: put overlapping reads together in order to get a contig

Used software: ABySS



Which partition would you use for the assembly?

Exercise 4

- Load Cerebro
- Load ABySS
- Check the abyss.pbs file, change the header/variables if needed
- Launch the abyss.pbs script
- Check the output of the assembly

Project description

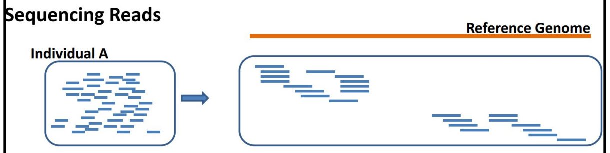
Project parts:

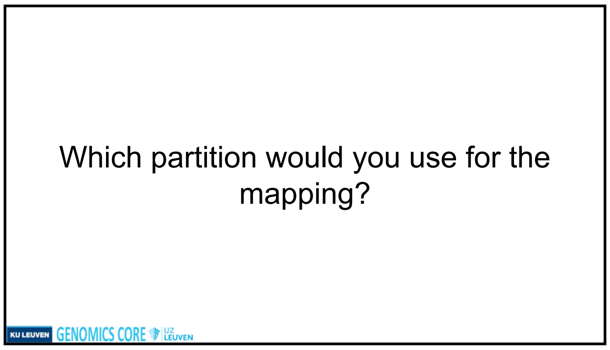
- Assembly
- **Mapping**
  19 individuals need the be mapped to the newly created reference genome
- Variant calling

Mapping

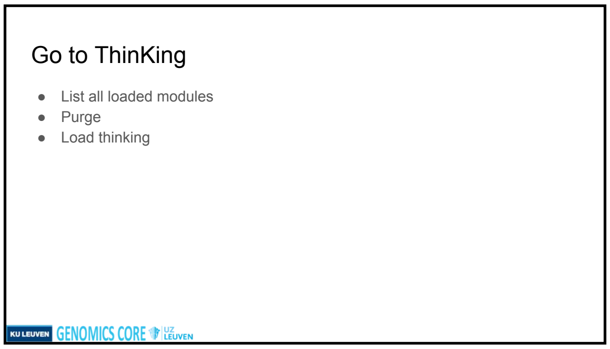
Compare your sequences against a reference in order to get the location

Used software: Bowtie2



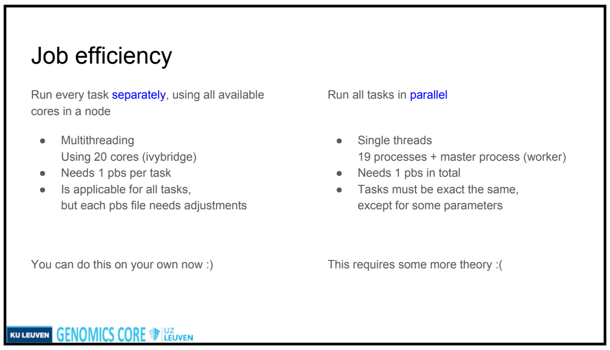


Which partition would you use for the mapping?



Go to ThinKing

- List all loaded modules
- Purge
- Load thinking



Job efficiency

Run every task separately, using all available cores in a node

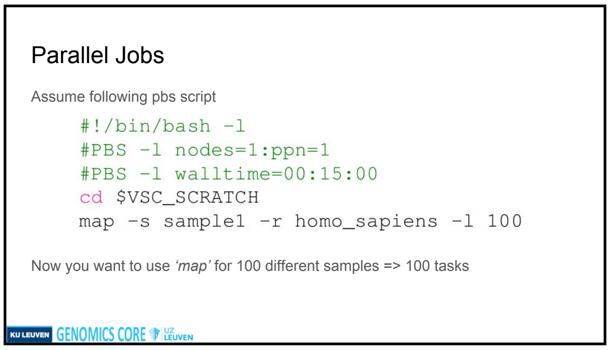
- Multithreading
  Using 20 cores (ivybridge)
- Needs 1 pbs per task
- Is applicable for all tasks,
  but each pbs file needs adjustments

You can do this on your own now :)

Run all tasks in parallel

- Single threads
  19 processes + master process (worker)
- Needs 1 pbs in total
- Tasks must be exact the same,
  except for some parameters

This requires some more theory :(

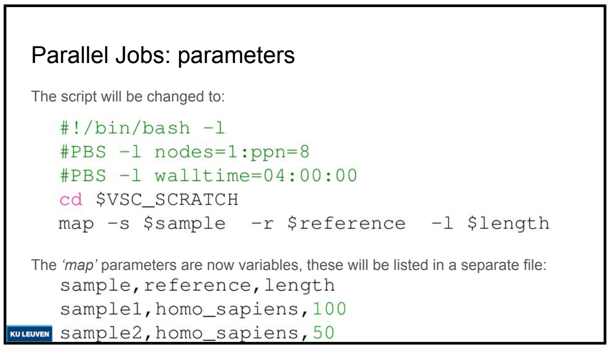


Parallel Jobs

Assume following pbs script

```
#!/bin/bash -l
#PBS -l nodes=1:ppn=1
#PBS -l walltime=00:15:00
cd $VSC_SCRATCH
map -s sample1 -r homo_sapiens -l 100
```

Now you want to use *'map'* for 100 different samples => 100 tasks



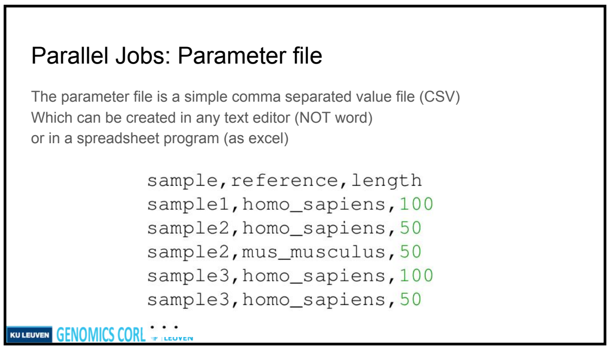
Parallel Jobs: parameters

The script will be changed to:

```
#!/bin/bash -l
#PBS -l nodes=1:ppn=8
#PBS -l walltime=04:00:00
cd $VSC_SCRATCH
map -s $sample  -r $reference  -l $length
```

The *'map'* parameters are now variables, these will be listed in a separate file:

```
sample,reference,length
sample1,homo_sapiens,100
sample2,homo_sapiens,50
```



Parallel Jobs: Parameter file

The parameter file is a simple comma separated value file (CSV)
Which can be created in any text editor (NOT word)
or in a spreadsheet program (as excel)

```
sample,reference,length
sample1,homo_sapiens,100
sample2,homo_sapiens,50
sample2,mus_musculus,50
sample3,homo_sapiens,100
sample3,homo_sapiens,50
...
```

Parallel Jobs: PBS header

Original file

```
#!/bin/bash -l
#PBS -l nodes=1:ppn=1
#PBS -l walltime=00:15:00
```

- Uses 1 core
- Walltime is 15 minutes
  100 tasks, 15 minutes each
  => 1500 minutes or 25 hours

Parallel file

```
#!/bin/bash -l
#PBS -l nodes=1:ppn=8
#PBS -l walltime=04:00:0
```

- 8 cores in use: 7 for the job, 1 for delegating the work
- Walltime is 4 hours
  15 minutes per job, 7 jobs at the time
  => 215 minutes (1500/7) or 3.56 hours
  So 4 hours is a safe time

Parallel Jobs: start a job

Load the worker module:

```
$ module load worker/1.5.0-intel-2014a
$ wsub -batch run.pbs -data data.csv
```

Use wsub instead of qsub

-batch	The pbs script (.pbs)
-data	The parameter file (.csv)

Parallel Jobs: Map Reduce

Parallel computations can be abstracted into 3 steps:

1. Preparation
2. The work items
3. Aggregating results, clean-up, …

The worker framework also supports this:

-prolog	Preparation step	e.g.: copying the reference genome to scratch
-batch	The parallel jobs	e.g.: mapping the reads to the genome
-epilog	Clean-up step	e.g.: cleaning the scratch storage

```
$ wsub -prolog split-data.sh -batch run.pbs -epilog distr.sh -
data data.csv
```

Exercise 5a

Description of the task

- Open bowtie_batch.pbs
- Change script
- Variable name?
- input/output path?
- Reference genome?

KU LEUVEN GENOMICS CORE UZ LEUVEN

Exercise 5b

- Open prolog script
- What happens?
- Check genome path
- Where is the genome stored?

KU LEUVEN GENOMICS CORE UZ LEUVEN

Exercise 5c

- Open epilog script
- What happens?

KU LEUVEN GENOMICS CORE UZ LEUVEN

Exercise 5d

- Start job on thinking
- Check mapping statistics (especially the data for the assembly)

Project description

Project parts:

- Assembly
- Mapping
- **Variant calling**
  The 2 populations have the be called together in order to find common variants (if no variant found, we want to know if it was reference, or not seen)

Homework: do population variant calling

```
$GENOME_DIR="$VSC_DATA/tutorial/denovo/abyss_output";
$FREEBAYES_OPTIONS="-m 20 -q 15 --use-duplicate-reads --ploidy 2";
$OUTPUT_DIR="$VSC_DATA/tutorial/freebayes";
mkdir -p $OUTPUT_DIR;
#insert a copy of your bam file to $SCRATCH_DIR/bams here
bamfiles="";
for i in `ls -1 -d $SCRATCH_DIR/bams/*bam`;
do
        bamfiles="$bamfiles $i";
done
freebayes --fasta-reference $GENOME_DIR/genome.fa
$FREEBAYES_OPTIONS $bamfiles > $SCRATCH_DIR/freebayes.vcf;
#insert the copy of your results to the $OUTPUT_DIR here
#Clean up your $SCRATCH_DIR here
```