Molgenis Compute based pipelines

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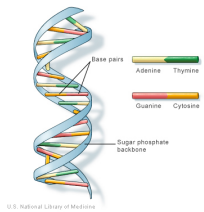
Content

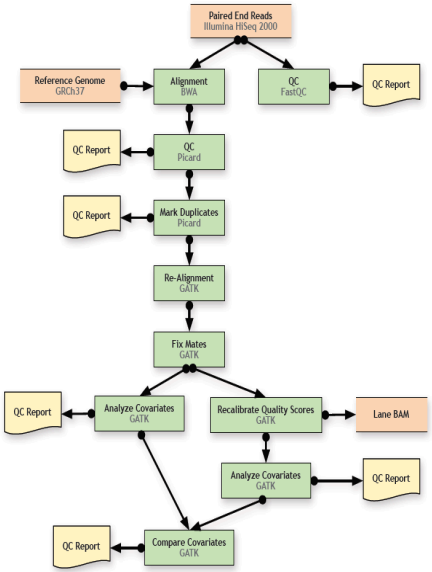
- General background
- Molgenis Compute commandline
- Molgenis ComputeDB (GUI)
- Pipelines
- Future work

General background

General background / example: NGS alignment workflow



HiSeq



Per Project:
1. Aligned reads
2. QC-reports
3. SNP lists

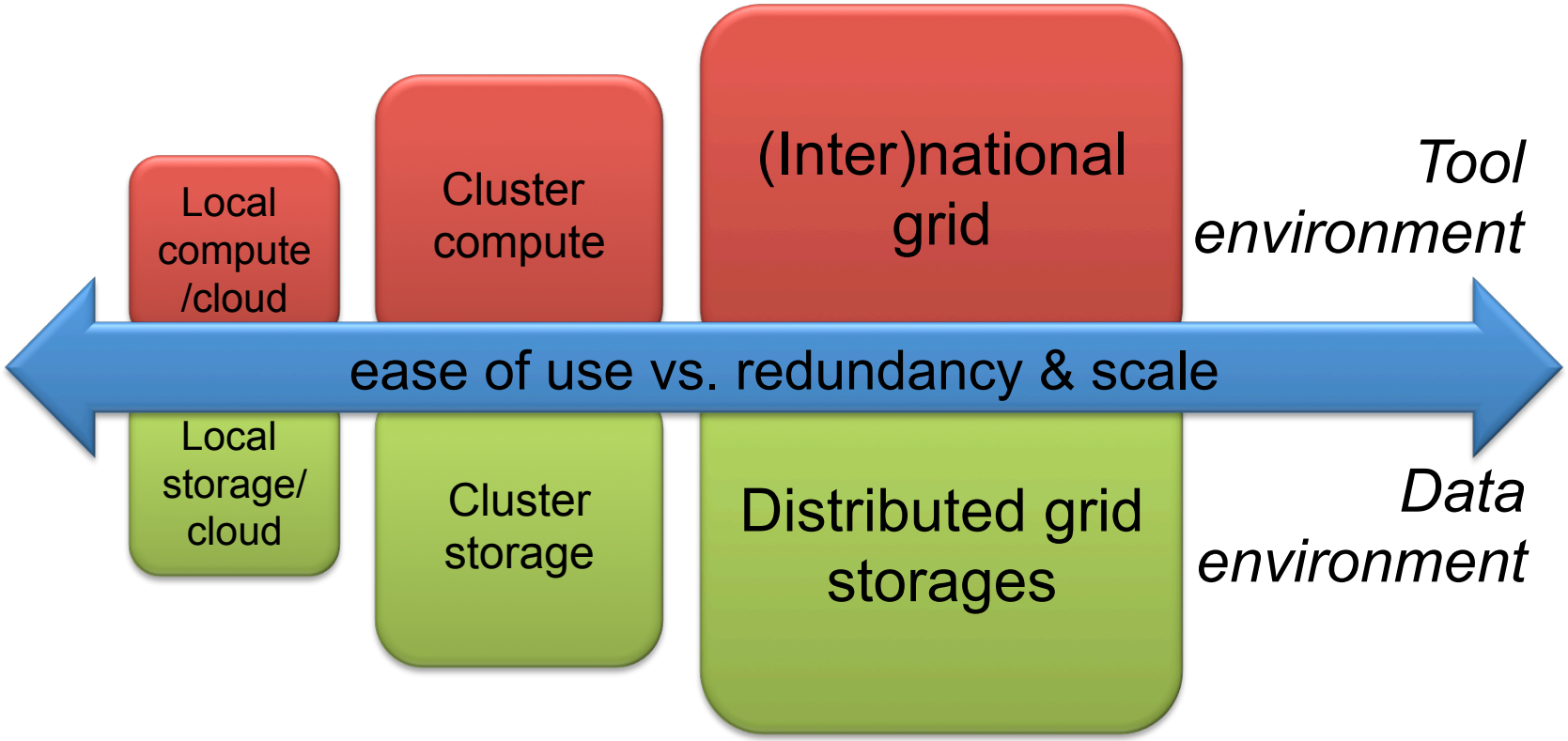
Raw data → ***alignment workflow*** → Result data

10-100 samples

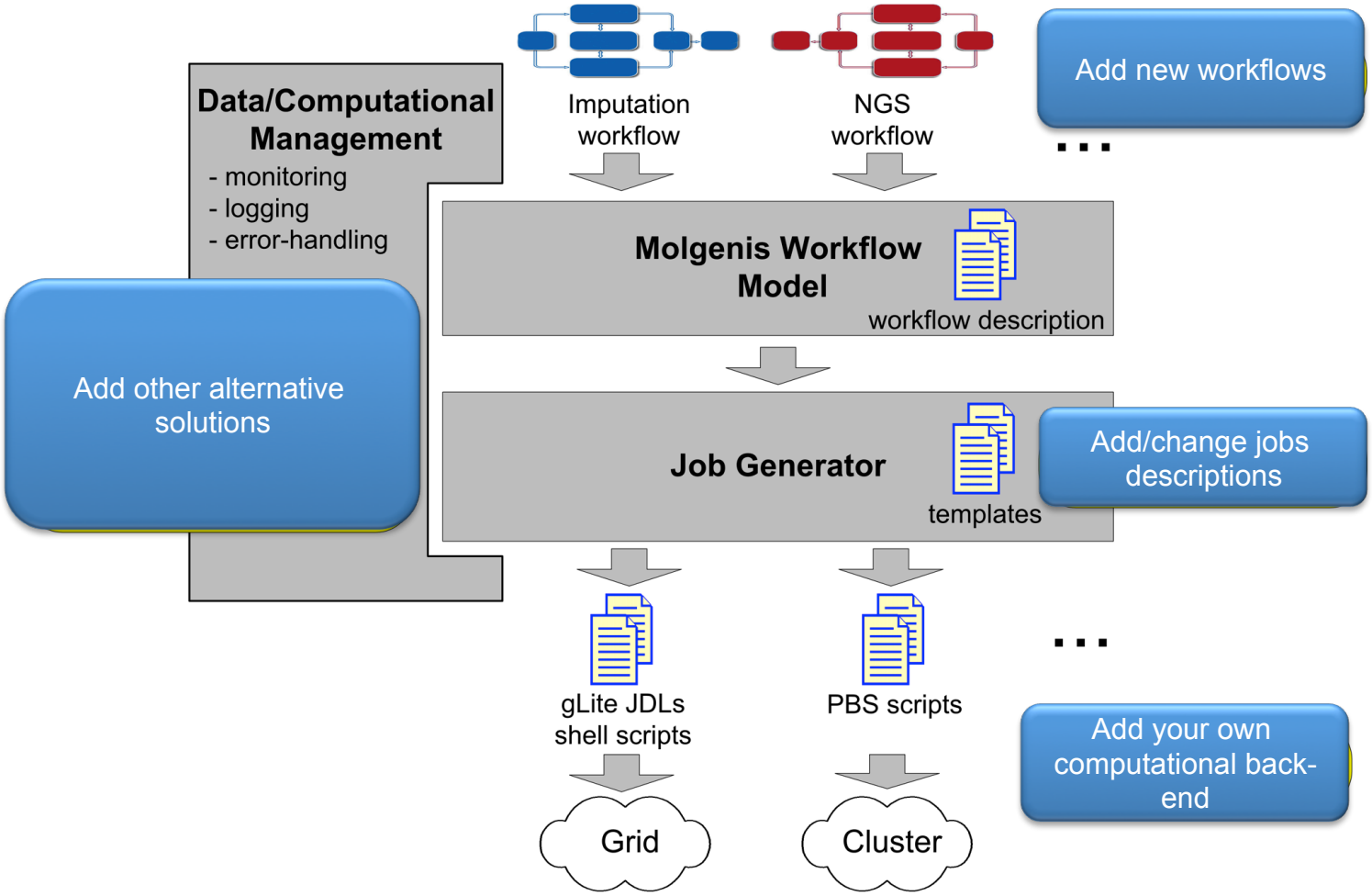
20 – 200 days

80 – 800 GB

Computational environments



Command-line generator



- Generates jobs (scripts) from model described in files
- Suitable for workflows (PBS cluster) and single jobs (gLite grid)

Molgenis Compute commandline

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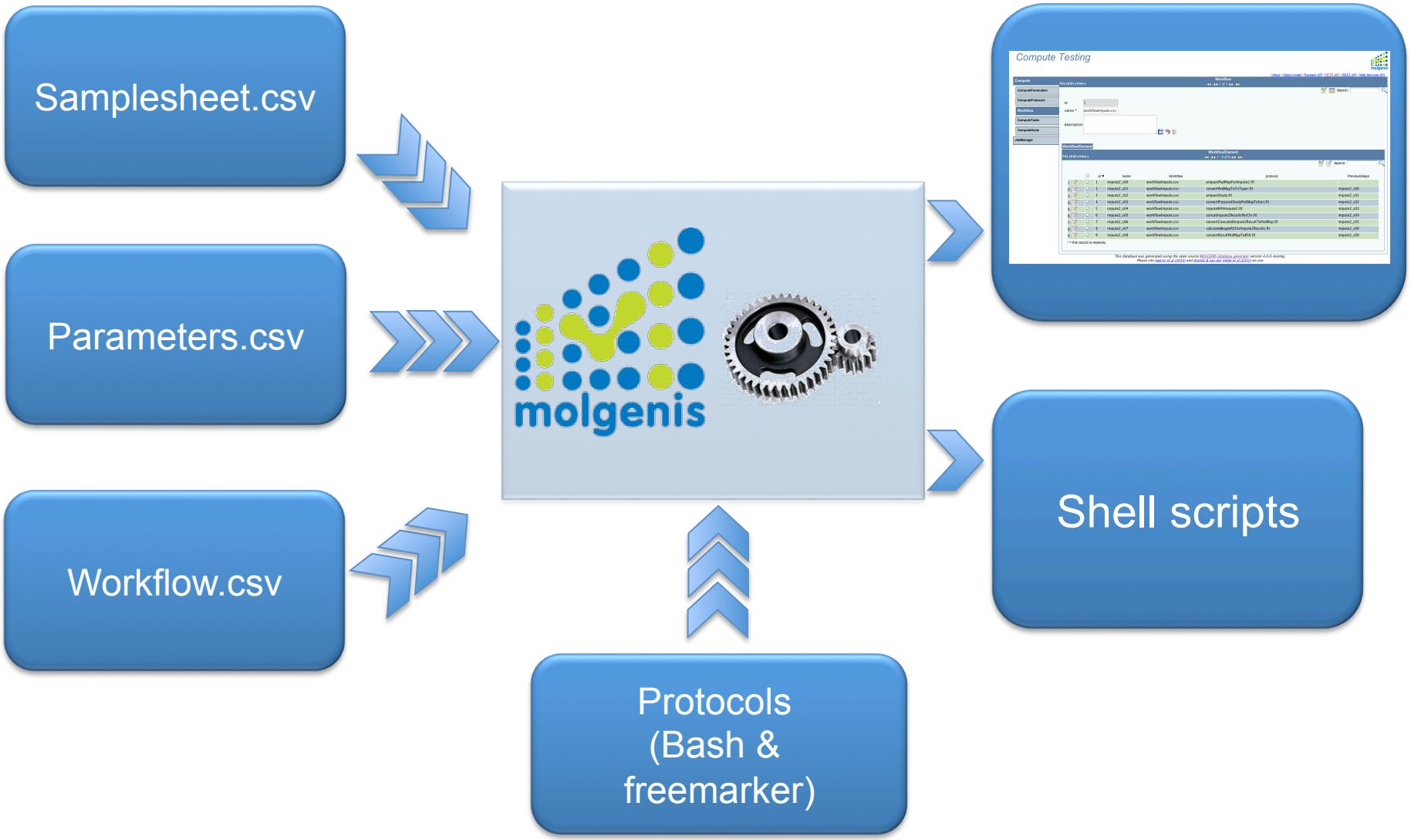
Compute overview



Samplesheet.csv

ID	sampleID	Project	Machine	SequencingDate	Run	Flowcell	Lane	Barcode	SequencingT	Manufacture	Enrichmentkit
3	Sample3_2	Testproject2	SN163	130809	501	BD2BWCACXX	4	RPI 01 ATCACG	PE	Agilent	SureSelect_All_Exon_50MB_v5.1
4	Sample5_2	Testproject2	SN163	130809	501	BD2BWCACXX	4	RPI 04 TGACCA	PE	Agilent	SureSelect_All_Exon_50MB_v5.1
5	Sample5_2	Testproject2	SN163	130809	501	BD2BWCACXX	5	RPI 04 TGACCA	PE	Agilent	SureSelect_All_Exon_50MB_v5.1
6	Sample4_2	Testproject2	SN163	130809	501	BD2BWCACXX	4	RPI 08 ACTTGA	PE	Agilent	SureSelect_All_Exon_50MB_v5.1

Parameters.csv

Samplesheet.csv

Parameters.csv

queue	devel				
mem	4				
walltime	23:59:00				
nodes	1				
ppn	1				
defaultInterprete	#!/bin/bash				
stage	module load				
checkStage	module list				
WORKDIR	/gcc/				
root	${WORKDIR}				
tempDir	${WORKDIR}/groups/gaf/tmp02/tmp/				
resDir	${root}/resources/				
toolDir	${root}/tools/				
scriptDir	${toolDir}/scripts/				
gafHome	${root}/groups/gaf/				
tmpDataDir	${root}/groups/gaf/tmp02/				
allRawtmpDataDi	${tmpDataDir}/rawdata/				
allRawNgstmpDat	${allRawtmpDataDir}/ngs/				
allRunDemultiple	${tmpDataDir}/projects/${project}/${run}				
runPrefix	${sequencingStartDate}_${sequencer}_${run}_${flowcell}				

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Workflow.csv

step	protocol	dependencies
FastQC	protocols/FastQC.sh	
BwaAlign	protocols/BwaAlign.sh	
SamToBam	protocols/SamToBam.sh	BwaAlign
SortBam	protocols/SortBam.sh	SamToBam;inputSortBam=alignedBam;tmpSortedBam=tmpAlignedSortedBam;tmpSortedBamIdx=tmpAlignedSortedBamIdx;sc
MergeBam	protocols/MergeBam.sh	SortBam;inputMergeBam=alignedSortedBam;inputMergeBamIdx=alignedSortedBamIdx;tmpMergedBam=tmpSampleMergedB
MarkDuplica	protocols/MarkDuplicates.sh	MergeBam

Parameters.csv

Workflow.csv

Example snippet SortBam.sh:

```
#string inputSortBam
#string tmpSortedBam
#string tmpSortedBamIdx


#Run picard, sort BAM file and create index on the fly
java -jar -Xmx3g $PICARD_HOME/${sortSamJar} \
INPUT=${inputSortBam} \
OUTPUT=${tmpSortedBam} \
SORT_ORDER=coordinate \
CREATE_INDEX=true \
VALIDATION_STRINGENCY=LENIENT \
MAX_RECORDS_IN_RAM=2000000 \
TMP_DIR=${tempDir}
```

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Protocol structure in more detail

```
//header
      #MOLGENIS walltime=15:00 nodes=1 cores=4 mem=6gb
      #list inputMergeBam
      #string outputMergedBam
//tool management
      module load bwa/${bwaVersion}
//data management
      getFile ${indexfile}
      getFile ${leftbarcodefqgz}
//template of the actual analysis
      bwa aln \
      ${indexfile} ${leftbarcodefqgz} \
      -t ${bwaaligncores} -f ${leftbwaout}
//data management
      putFile ${leftbwaout}
```

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Data transfer

- getFile and p
  - are back-e
  - now, we
    - check i
    - do srm/
- Input

```
getFile $
```

- Generated o

```
getFile $V
imputatio
chr20.map
```

```
#------- data transfer

getRemoteLocation()
{
    ARGS=($@)
    myFile=${ARGS[0]}
    remoteFile=srm://srm.grid.sara.nl/pnfs/grid.sara.nl/data/bbmri.nl/RP2${myFile:`expr length $TMPDIR`}
    echo $remoteFile
}

getFile()
{
    ARGS=($@)
    NUMBER="${#ARGS[@]}";
    if [ "$NUMBER" -eq "1" ]
    then

    myFile=${ARGS[0]}
    remoteFile=`getRemoteLocation $myFile`

    # 1. myPath = getPath( myFile ) will strip off the file name and return the path
    mkdir -p $(dirname "$myFile")

    # 2. cp srm:.../remoteFile myFile
    echo "srmcp -server_mode=passive $remoteFile file:///$myFile"
    srmcp -server_mode=passive $remoteFile file:///$myFile
    chmod 755 $myFile

    else
    echo "Example usage: getData \"\$TMPDIR/datadir/myfile.txt\""
    fi
}

putFile()
{
    ARGS=($@)
    NUMBER="${#ARGS[@]}";
    if [ "$NUMBER" -eq "1" ]
    then
    myFile=${ARGS[0]}
    remoteFile=`getRemoteLocation $myFile`
    echo "srmrm $remoteFile"
    srmrm $remoteFile
    echo "srmcp -server_mode=passive file:///$myFile $remoteFile"
    srmcp -server_mode=passive file:///$myFile $remoteFile
        returnCode=$?

        echo "srmcopy: ${returnCode}"

        if [ $returnCode -ne 0 ]
        then
            exit 1
        fi
    else
    echo "Example usage: putData \"\$TMPDIR/datadir/myfile.txt\""
    fi
}
```

Generated back-end independent script

```
//header
#MOLGENIS walltime=15:00 nodes=1 cores=4 mem=6
//tool management
module load bwa/0.5.8c_patched
//data management
getFile $WORKDIR/resources/hg19/indices/human_g1k_v37.fa
getFile $WORKDIR/groups/gcc/projects/cardio/run01/rawdata/
121128_SN163_0484_AC1D3HACXX_L8_CAACCT_1.fq.gz
//template of the actual analysis
bwa aln \
human_g1k_v37.fa 121128_SN163_0484_AC1D3HACXX_L8_CAACCT_1.fq.gz -t 4 \
-f 121128_SN163_0484_AC1D3HACXX_L8_CAACCT_1.bwa_align.human_g1k_v37.sai
//data management
putFile $WORKDIR/groups/gcc/projects/cardio/run01/results/
121128_SN163_0484_AC1D3HACXX_L8_CAACCT_1.bwa_align.human_g1k_v37.sai
```

Best practices

- Always output to temporary files and check return codes of software

```
#Get return code from last program call
returnCode=$?

echo -e "\nreturnCode MergeBam: $returnCode\n\n"

if [ $returnCode -eq 0 ]
then
    echo -e "\nMergedBam finished succesfull. Moving temp files to final.\n\n"
    mv ${tmpMergedBam} ${finalMergedBam}
else
    echo -e "\nFailed to move MergeBam results to ${finalMergedBam}\n\n"
    exit -1
fi
```

- Create md5sums

Molgenis ComputeDB (GUI)

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Pilot jobs

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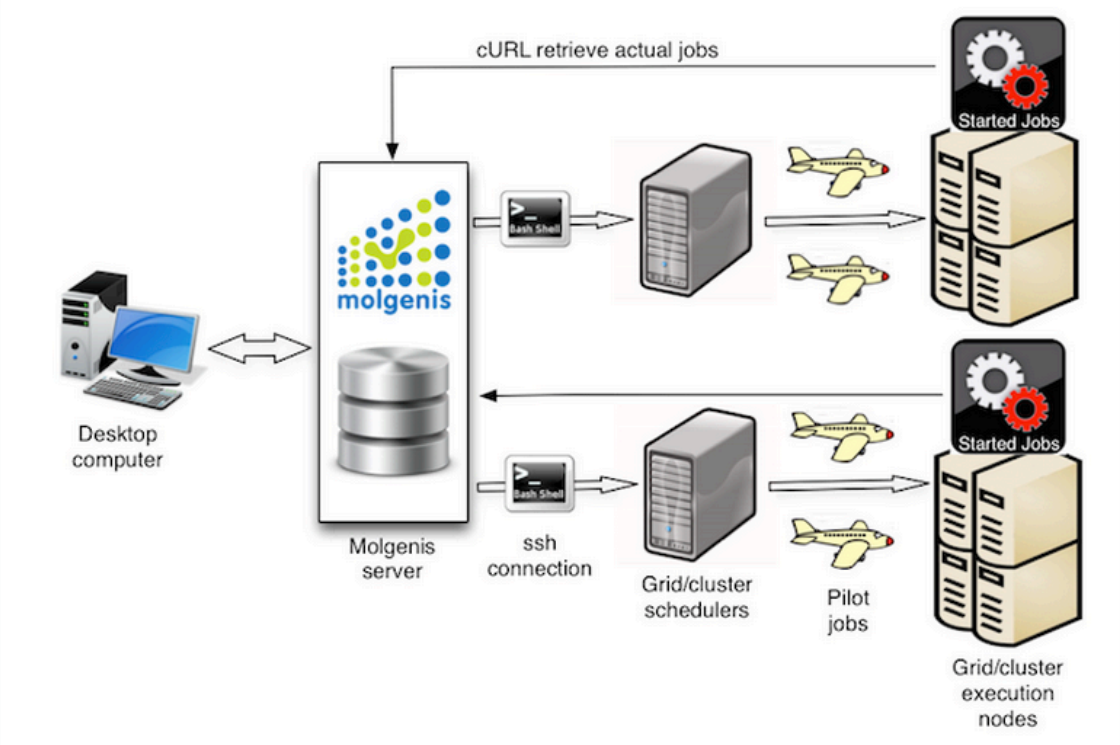
Home

Sign in

Welcome to Molgenis Compute5db Platinum Grid Edition



This database was generated using the open source MOLGENIS database generator version 4.0.0-testing. Please cite Swertz et al (2010) or Swertz & Jansen (2007) on use.

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Login

test18@ui.grid.sara.nl (2013-09-09 12:56:45)

Active

Inactivate

Username

Password

Submit Pilots

Cancel Run

Jobs generated	0	Jobs done	589
Jobs ready	0	Jobs failed	0
Jobs running	22	Jobs cancelled	0
Pilots submitted	1261	Pilots started	902

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Dashboard

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Pilot dashboard | Runs | Tasks | Parameters | Backends | Sign out

Main / Compute

test11@ui.grid.sara.nl (2013-12-20 14:15:56)

Not active

Activate

Cancel Run

Jobs generated	2	Jobs done	0
Jobs ready	0	Jobs failed	0
Jobs running	0	Jobs cancelled	0
Pilots submitted	0	Pilots started	0

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Pipelines

Supported analyses

- NGS variant calling pipeline
- RNA-seq analysis
- Liftover genome builds
- … and many more pipelines

- Out-of-the-box imputation pipeline
  - Automatic installation of tools and reference datasets
  - Liftover, phasing and imputation in 3 commands

Future work

Future work

- Finish implementation new NGS pipeline
- Collect and show more statistics to user in dashboard/commandline
- Add more workflows
- Unique *job/workflow* interface to run in different computational back-ends
  - Cluster (PBS, SGE, etc.)
  - Grid (gLite)
  - Cloud (OpenStack) <- Ongoing
- Interface with Galaxy (and other WMS?)
- Visual analytics of data/workflows/runs

Future work

- Explore easybuild
  http://hpcugent.github.io/easybuild/

Contributors: GCC team

- Compute development
  - George Byelas
  - Martijn Dijkstra
- Pipeline development and NGS analysis:
  - Pieter Neerincx
  - Gerben van der Vries
  - Lennart Johansson
  - Roan Kanninga

Github repositories

- Molgenis Compute (& tutorial)
  - https://github.com/molgenis/molgenis-pipelines/
- Imputation out-of-a-box
  - https://github.com/molgenis/molgenis-imputation
- Pipelines
  - https://github.com/molgenis/molgenis-pipelines/tree/master/compute5
- NGS (Under development)
  - https://github.com/molgenis/molgenis-pipelines/tree/master/compute5/NGS_alignment_SNP_calling