Molgenis Compute based pipelines

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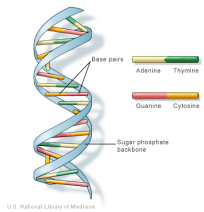
Content

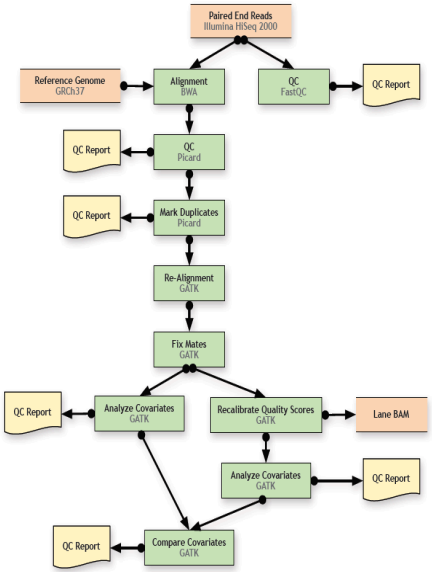
- General background
- Molgenis Compute
  - Commandline/Cluster
  - Grid
  - Cloud
- Pipelines
- LIMS
- Future work

General background

General background / example: NGS alignment workflow



HiSeq



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

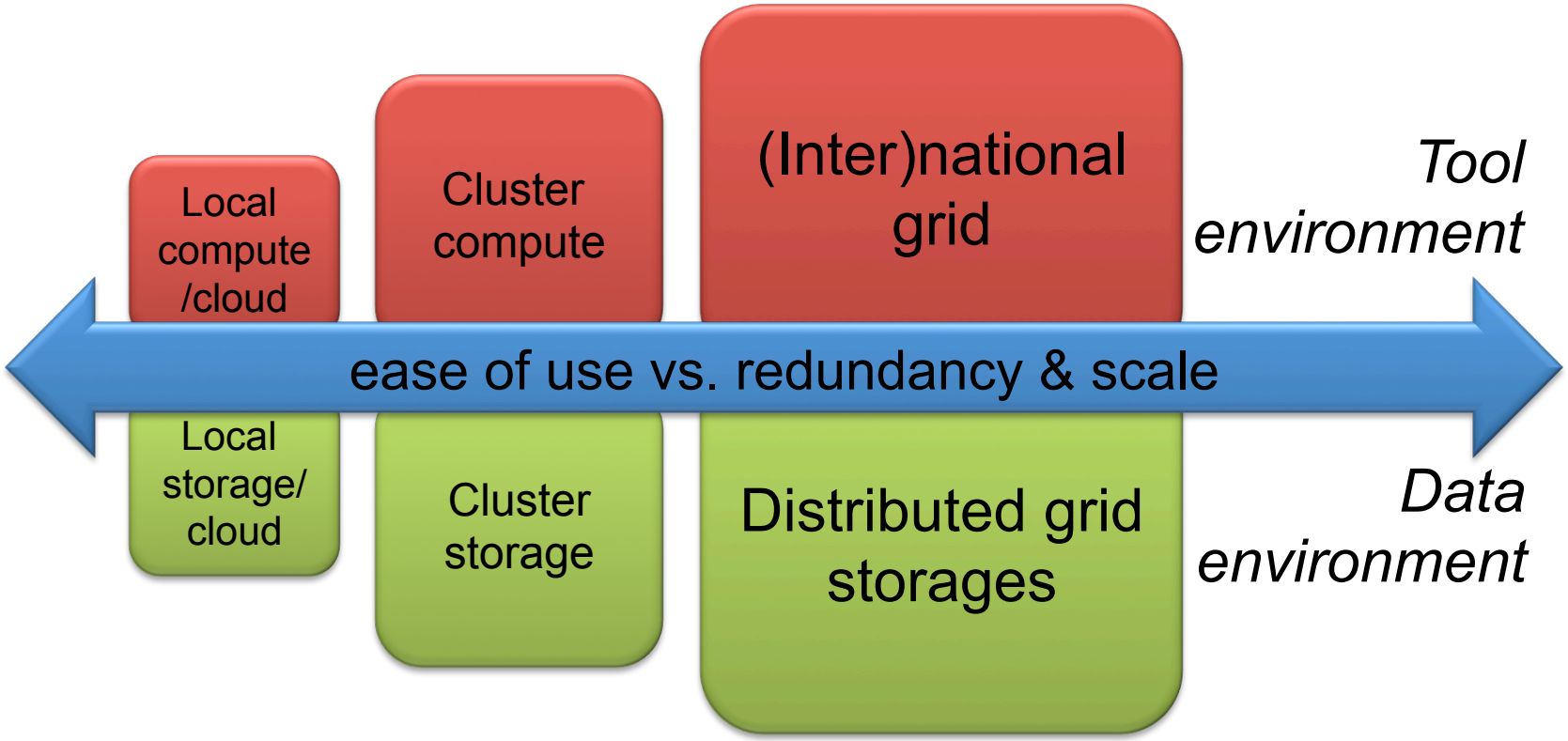
Raw data → ***alignment workflow*** → Result data

10-100 samples

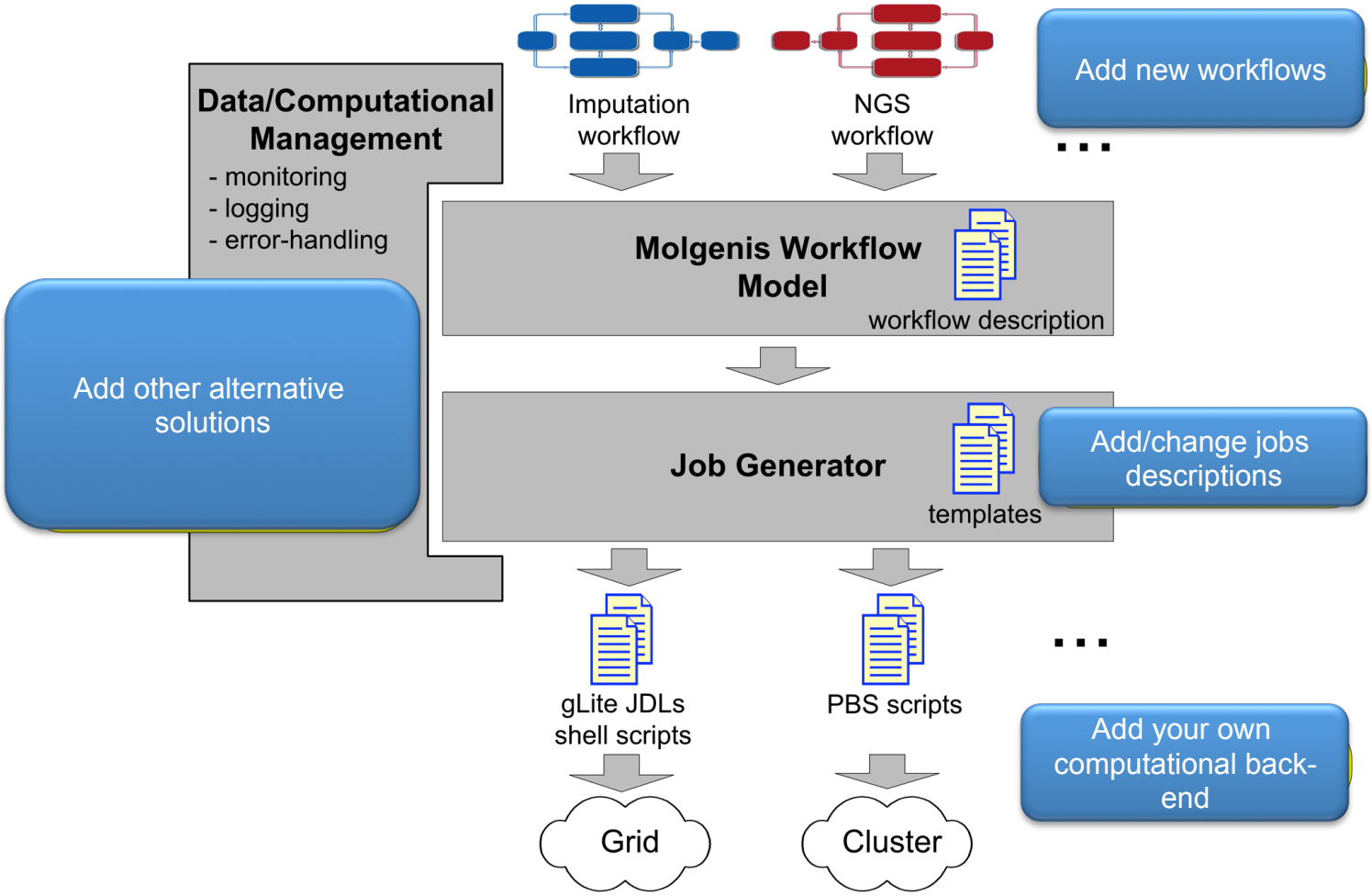
20 – 200 days

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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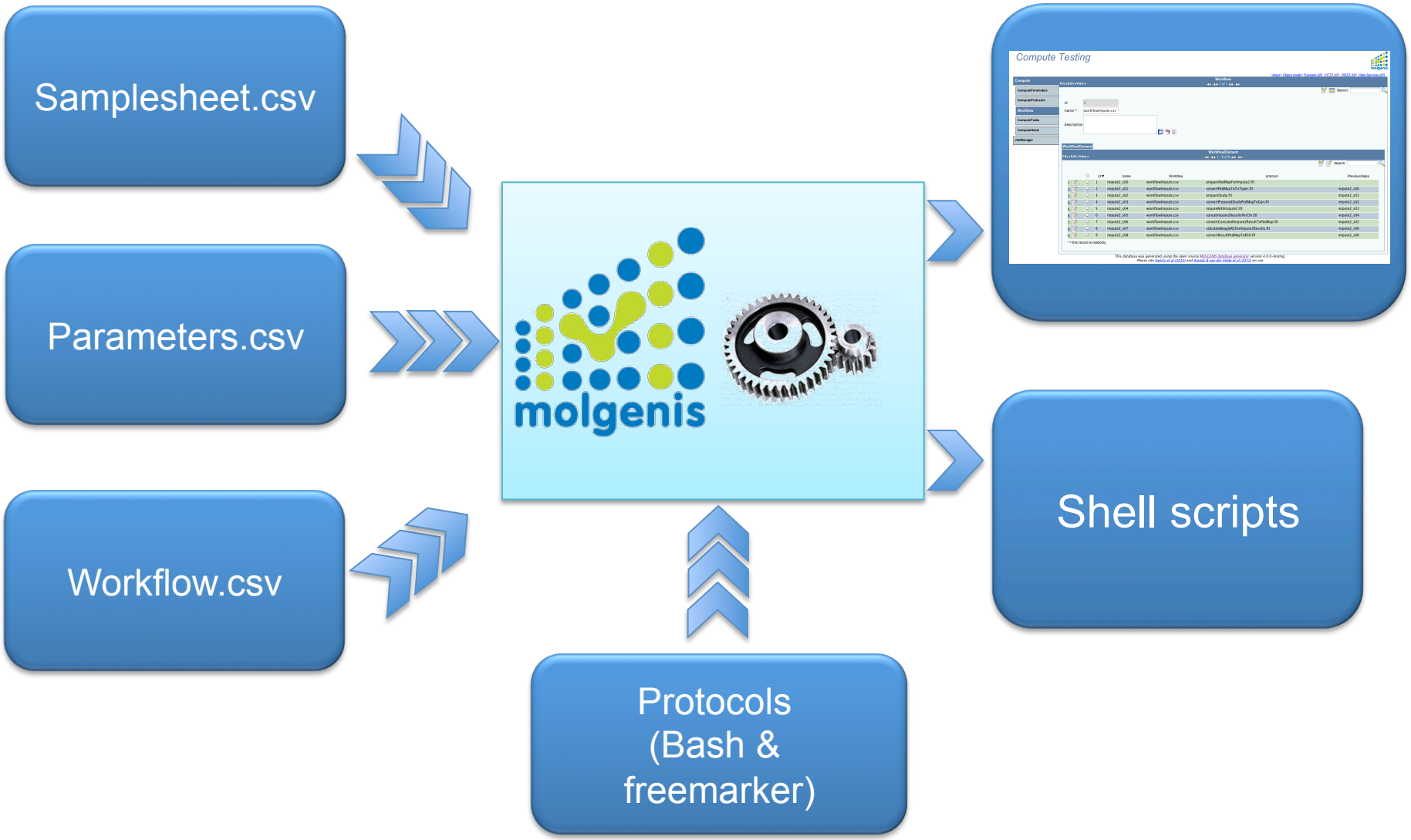
Compute overview



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}
```

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 code 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

Submission in the cluster

- No dependencies

```
#
##alignment_21
#

# Skip this step if step finished already successfully
if [ -f alignment_21.sh.finished ]; then
        skip alignment_21.sh
        echo "Skipped alignment_21.sh"
else
        # Build dependency string
        dependenciesExist=false
        dependencies="-W depend=afterok"
        if ! $dependenciesExist; then
                unset dependencies
        fi

        alignment_21=$(qsub -N alignment_21 $dependencies alignment_21.sh)
        echo $alignment_21
        sleep 0
fi


#
##imputation_0
#

# Skip this step if step finished already successfully
if [ -f imputation_0.sh.finished ]; then
        skip imputation_0.sh
        echo "Skipped imputation_0.sh"
else
        # Build dependency string
        dependenciesExist=false
        dependencies="-W depend=afterok"
                if [[ -n "$alignment_0" ]]; then
                        dependenciesExist=true
                        dependencies="${dependencies}:$alignment_0"
                fi
        if ! $dependenciesExist; then
                unset dependencies
        fi

        imputation_0=$(qsub -N imputation_0 $dependencies imputation_0.sh)
        echo $imputation_0
        sleep 0
fi
```

- Dependencies

Molgenis Compute for the grid

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Login

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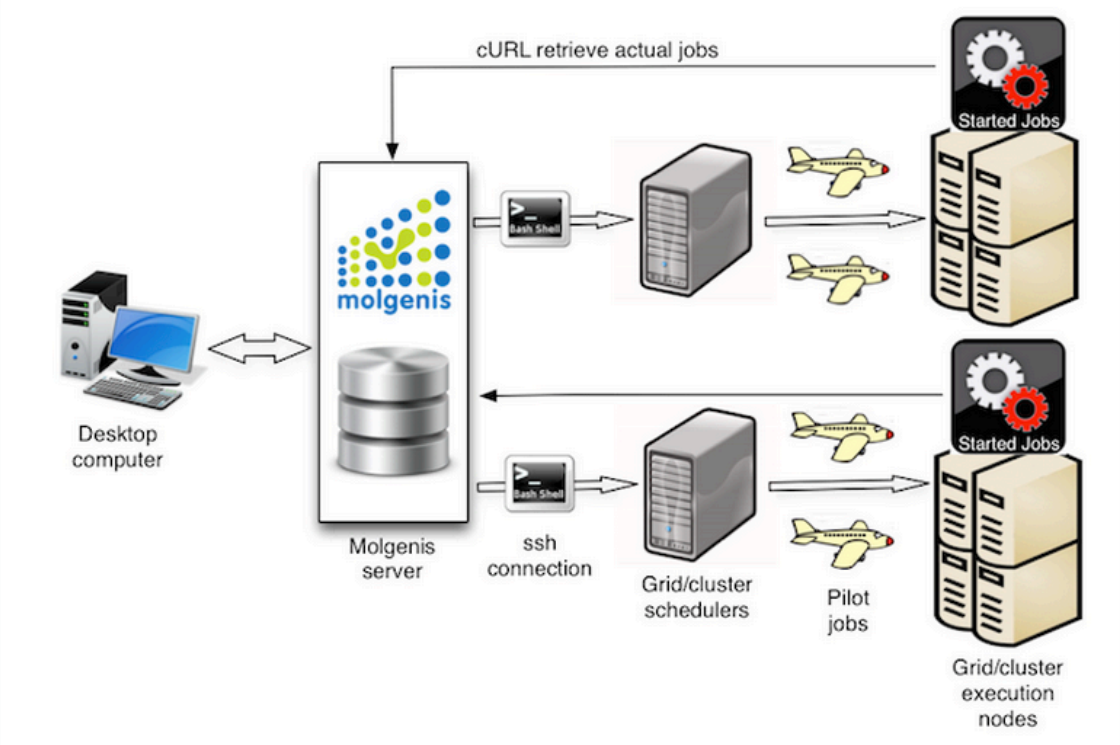
Home

Sign in

Welcome to Molgenis Compute5db Platinum Grid Edition



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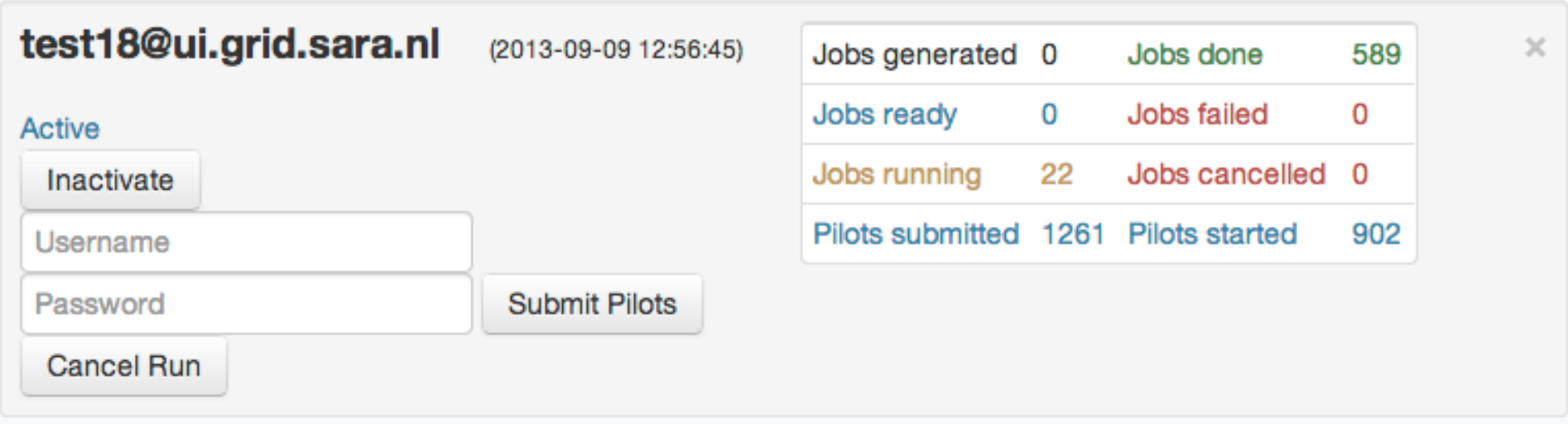
Dashboard of one run in the grid



- Main features:
  - using pilot jobs (overview of pilots)
  - all running jobs can be cancelled (pilot asks server, if it is cancelled before the final data transfer)
  - all grid sites are synchronized using module system

Technical details of execution in the grid (1)

1. *Pilot* is sent to the scheduler from MOLGENIS *Server*

```
glite-wms-job-submit -d $USER … $HOME/maverick.jdl
```

2. *Pilot* asks the *Server* for *Job* todo

```
curl  … -F status=started -F backend=ui.grid.sara.nl \
http://$SERVER:8080/api/pilot > script.sh
```

3. *Pilot* starts *Job* in the computation element (CE) of the grid

```
bash -l script.sh 2>&1 | tee -a log.log &
```

Technical details of execution in the grid (2)

4. *Pilot* sends pulse and *Job's* update to *Server*

```
while [ 1 ] ; do
…
check_process "script.sh"
CHECK_RET=$?
if [ $CHECK_RET -eq 0 ];
then
        …
        curl … -F status=nopulse -F log_file=@inter.log …
elif
       …
        curl … -F status=pulse -F log_file=@inter.log …
```

Technical details of execution in the grid (3)

5. Job asks server, if it is cancelled before final data transfer

```
RESULT=‘curl ... -F pilotid=${pilotid} –F status=is_cancel \
http://$SERVER:8080/api/pilot‘ ...

if [ "$RESULT" = "not_cancelled" ];
then
      srmcp file:///$localFile $remoteFile
...
fi
```

MOLGENIS Compute for the cloud

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Dashboard for the grid and cloud runs

test01@ui.grid.sara.nl (2014-05-06 12:22:31)

Active

Inactivate

Username

Password

Submit Pilots

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

test02@openstack01.gcc.rug.nl (2014-05-06 12:23:06)

Ready to run

Username

Password

Start

Jobs generated	0	Jobs done	0
Jobs ready	100	Jobs failed	0
Jobs running	0	Jobs cancelled	0

- Main features:
  - on demand virtual machine (VM) invocation, if jobs should be executed
  - Pre-installed VM's in OpenStack platform

Main steps in running in the cloud

1. Get token from the cloud **(keystone/rest)**
2. Select VM image to run analysis **(nova/rest)**
3. Start Server **(nova/rest)**
4. Associate Server with the external IP address **(nova/rest)**
5. Select and mount Disk to the Server **(nova/rest + ssh)**
6. Start analysis with **(ssh + curl)**
7. Analysis reports back **(curl)**
8. Move results from VM Disk to NFS server **(scp)**

Next plans with the cloud

- Now, we use only OpenStack platform
  - Later, add OpenNebula
- Now, we test on 10 VM's (max 64 CPU's) and 10 disk (10 * 100GB)
  - Later, scale up testing
- Now, we run only one workflow
  - Later, create more specific VM's for specific workflows
- Now, error handling is absent
  - Later, cover most possible errors in the cloud
- Now, we run on only one OpenStack server
  - Later, add more sites and exchange VM images between them, if needed

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

LIMS

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LIMS

molgenis | Catalog | Data Explorer | Upload | Entities | BiobankConnect | Workflow Data Entry | Background | Entity Explorer | Admin | Account | Sign o

Choose a dataset: GAF list 2014-05-12 04:00

rch data values

a item filters

a item selection

- contact
- validationLog
- labStatusPhase
- labStatusComments
- lastUpDate
- *sequencingStartDate*
- *run*
- *flowcell*
- *lane*
- *barcodeMenu*
- *seqType*
- *prepKit*
- *capturingKit*
- *arrayFile*
- *arrayID*
- *GAF_QC_Name*
- *GAF_QC_Date*

zard

Data | Aggregates | Charts

internalSampleID	project	sequencer	sequencingStartDate	run	flowcell	lane	barcodeMenu	seqType	prepKit	capturingKit
1674	NIPTValidatie	SN163	130423	0493	BC1BFCACXX	8	RPI 10 TAGCTT	SR	NEB	None
1675	NIPTValidatie	SN163	130423	0493	BC1BFCACXX	8	RPI 11 GGCTAC	SR	NEB	None
1676	NIPTValidatie	SN163	130423	0493	BC1BFCACXX	8	RPI 12 CTTGTA	SR	NEB	None
1677	LDM_Klaas	SN163	130905	0510	BH7D9JADXX	1,2	MON 05 AAGACG	PE	Mondrian	SureSelect_All_Exon_50MB
1678	LDM_Klaas	SN163	130905	0510	BH7D9JADXX	1,2	MON 06 CCTCGG	PE	Mondrian	SureSelect_All_Exon_50MB
1679	LDM_Klaas	SN163	130905	0510	BH7D9JADXX	1,2	MON 07 GGATGT	PE	Mondrian	SureSelect_All_Exon_50MB
1680	LDM_Klaas	SN163	130905	0510	BH7D9JADXX	1,2	MON 08 TTCGCT	PE	Mondrian	SureSelect_All_Exon_50MB
1681	HaloPlex	SN163	130521	0494	AC20FTACXX	8	HP8 56 GTACGCAA	PE	Haloplex	None
1682	HaloPlex	SN163	130521	0494	AC20FTACXX	8	HP8 60 TAGGATGA	PE	Haloplex	None
1683	HaloPlex	SN163	130521	0494	AC20FTACXX	8	HP8 62 TCCGTCTA	PE	Haloplex	None
1684	HaloPlex	SN163	130521	0494	AC20FTACXX	8	HP8 64 TGAAGAGA	PE	Haloplex	None
1685	SCA_C	SN163	130710	0498	AD2AMUACXX	1	AGI 01 ATCACG	PE	Agilent	SureSelect_All_Exon_50MB
1686	SCA_C	SN163	130710	0498	AD2AMUACXX	1	AGI 08 ACTTGA	PE	Agilent	SureSelect_All_Exon_50MB
1687	SCA_C	SN163	130710	0498	AD2AMUACXX	1	AGI 10 TAGCTT	PE	Agilent	SureSelect_All_Exon_50MB
1688	SCA_C	SN163	130710	0498	AD2AMUACXX	1	AGI 11 GGCTAC	PE	Agilent	SureSelect_All_Exon_50MB
1689	SCA_C	SN163	130710	0498	AD2AMUACXX	2	AGI 02 CGATGT	PE	Agilent	SureSelect_All_Exon_50MB
1690	SCA_C	SN163	130710	0498	AD2AMUACXX	2	AGI 04 TGACCA	PE	Agilent	SureSelect_All_Exon_50MB
1691	SCA_C	SN163	130710	0498	AD2AMUACXX	2	AGI 07 CAGATC	PE	Agilent	SureSelect_All_Exon_50MB
1692	SCA_C	SN163	130710	0498	AD2AMUACXX	2	AGI 16 AAAGCA	PE	Agilent	SureSelect_All_Exon_50MB
1693	SCA_C	SN163	130809	0501	BD2BWCACXX	1	AGI 01 ATCACG	PE	Agilent	SureSelect_All_Exon_50MB

1083 data items found

« 1 ... 31 32 33 34 35 ... 55 »

Download as

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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

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