Software & Requirements

Javier Perales-Patón
jperales@cnio.es

Fátima Al-Shahrour
[falshahrour@cnio.es]
Elena Piñeiro-Yáñez
[epineiro@cnio.es]
Pedro Fernandes
[pfern@igc.gulbenkian.pt]

Translational Bioinformatics Unit
CNIO. Madrid, Spain.

Computational Facilities

- The **genome reference** must be **indexed** only once in your computer, but it takes a long time (2-3 days). The sizes of the indexes are huge (~100 Gb). See "Before running a variant analysis for first time" at: http://rubioseq.bioinfo.cnio.es/sites/default/files/PDF/RUbioSeq-book.pdf

- You need a huge **hard disk capacity**:
  - **Human genome reference** (including indexes) : 150 Gb.
  - **Bundle of files for Variant Calling Analysis** (GATK) : 14 Gb.
  - **For each sample**:
    - Raw data : 7 - 15 Gb.
    - Intermediate files for each sample : 40 - 60 Gb (whole-exome seq).

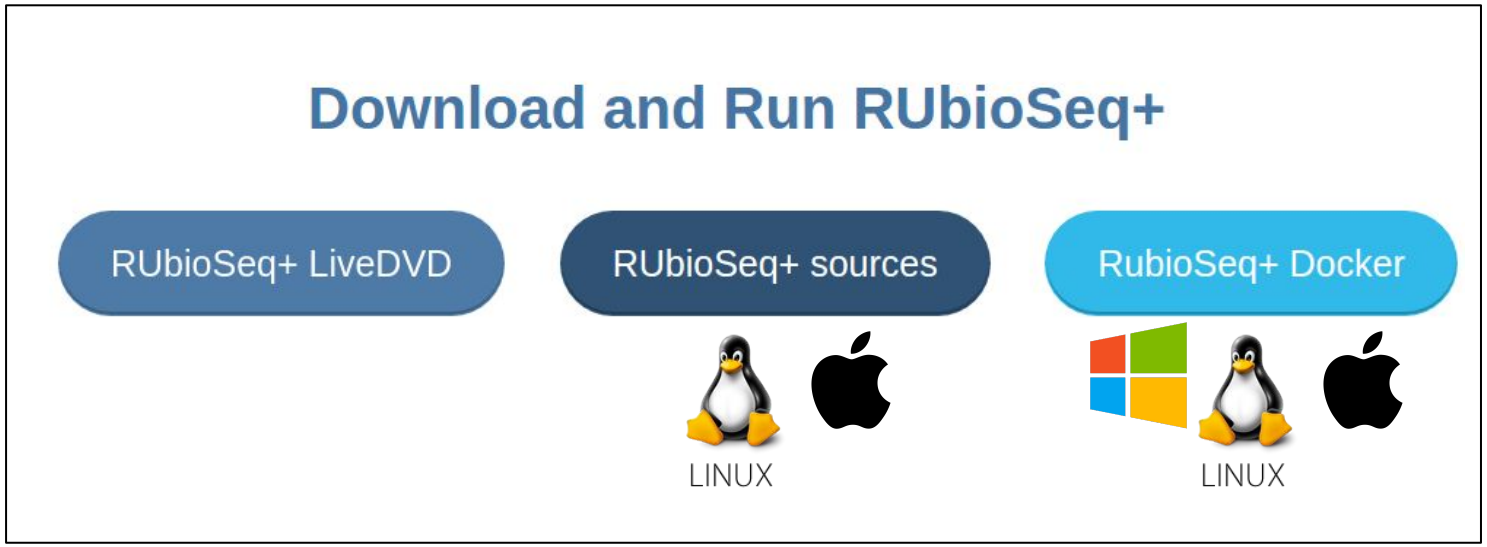
WorkStation:

- **Minimal requirements:**
  - 16 Gb RAM
  - 500 Gb hard disk
  - 8 threads ~ 8 cores.

- **Recommended requirements (multi-sample analysis, samples storage):**
  - 25 Gb RAM
  - 1 Tb hard disk (e.g. Exome-seq analysis from 50 samples → 1.5 Tb).
  - 16 threads.

Installing RUbioSeq+ in your computer

http://rubioseq.bioinfo.cnio.es/



RubioSeq+ Docker

Quick Manual Installation

(Virtual machine, Ubuntu system)

1. First, install Docker client (depending on your Operating System):
   Follow the corresponding guidelines:
   - Ubuntu (https://docs.docker.com/installation/ubuntulinux/)
   - Mac OS X (https://docs.docker.com/installation/mac/)
   - Windows (https://docs.docker.com/installation/windows/)

2. Second, launch the Docker Image (it will download it from the Internet):
   - Windows:
     (Start Menu): Program Files > Boot2Docker

   - Unix (in terminal):
     `docker run -ti -p 0.0.0.0:8080:8080 --name RUbioSeq ubio/rubioseq:latest /bin/bash`

3. Try it out:

```
bash /opt/RUbioSeq3.7/RUbioSeq.pl --help
```

More info: http://rubioseq.bioinfo.cnio.es/rubioseq_docker

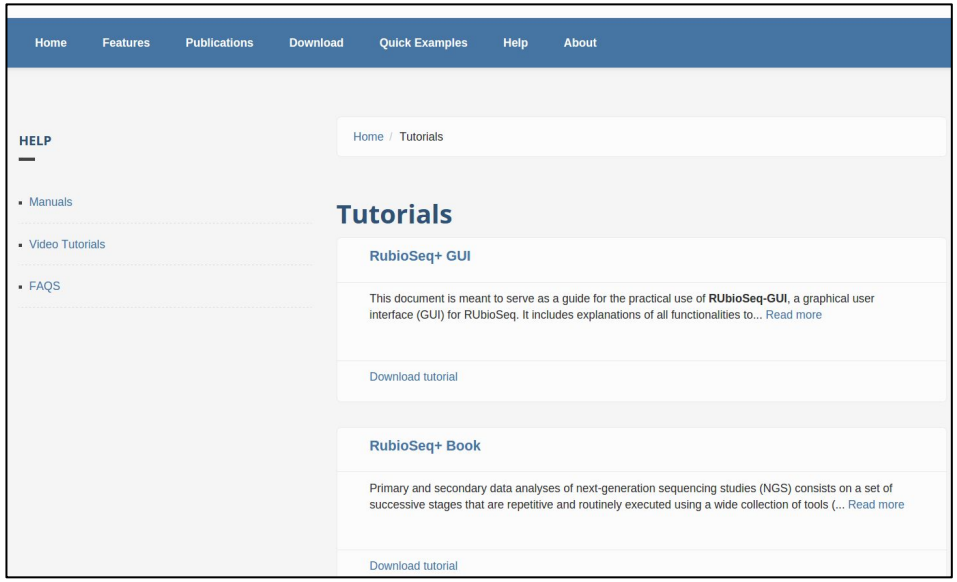
Ask for some help

RUbioSeq is under **development** by the Bioinformatics Unit at CNIO (Madrid, Spain):

- Miriam Rubio-Camarillo (mrubioc@cnio.es).
- José María Fernández (jmfernandez@cnio.es).
- Gonzalo Gómez-López (ggomez@cnio.es).

http://rubioseq.bioinfo.cnio.es/tutorials

- Video-tutorials.
- Example exercises.



RUbioSeq: a suite of parallelized pipelines to automate exome variation and bisulfite-seq analyses
Miriam Rubio-Camarillo,*,Gonzalo Gómez-López, José M. Fernández, Alfonso Valencia and David G. Pisano.
2013, Bioinformatics, 29 (13): 1687-1689

GATK's bundle from FTP repository (14 Gb)

You can download the different bundles from GATK's FTP (Broad Institute) visiting this URL with your Internet Browser:

ftp://gsapubftp-anonymous@ftp.broadinstitute.org/bundle/2.8/

1. **Genome Reference** (standard 1000 Genomes, fasta).
2. List of **Target beats or intervals** of genomic regions sequenced by the Library protocol.
3. **dbSNP** (VCF file) for a recent dbSNP release (build 138, it includes the 1000 Genomes).
4. HapMap genotypes and sites VCFs
5. **OMNI 2.5 genotypes for 1000 Genomes samples** (VCF).
6. The current best set of **known indels** to be used for local realignment); use both files:
   - 1000G_phase1.indels.b37.vcf (currently from the 1000 Genomes Phase I indel calls)
   - Mills_and_1000G_gold_standard.indels.b37.sites.vcf

Tip for home: The following UNIX command downloads the whole bundle for **hg19** in one step (~hrs) :

```
$ wget -r -nH --cut-dirs=3 --reject-regex "NA12878|CEUTrio" \

-P /path/to/your_directory/ ftp://gsapubftp-anonymous@ftp.broadinstitute.org/bundle/2.8/hg19/*
```

RUbioSeq+ sources

Quick Manual Installation

Dependencies (Ubuntu system)

rubioseq

1. Install **java 1.7** (http://www.webupd8.org/2012/01/install-oracle-java-jdk-7-in-ubuntu-via.html):

```
sudo add-apt-repository ppa:webupd8team/java

sudo apt-get update

sudo apt-get install oracle-java7-installer
```

2. Install **python, then the following python modules:**

```
sudo apt-get install python-dev
sudo pip install Numpy
sudo pip install Scipy
```

3. Install **perl and these perl modules:**

(For Bioperl, see “Installing using CPAN” at http://www.bioperl.org/wiki/Installing_BioPerl_on_Unix)

```
sudo cpan DBI DBD::mysql XML::LibXML Carp FindBin File::Basename
sudo cpan File::Spec File::Copy Getopt::long Class::Inspector
```

4. Library **libmysqlclient:**

(For Bioperl, see “Installing using CPAN” at http://www.bioperl.org/wiki/Installing_BioPerl_on_Unix)

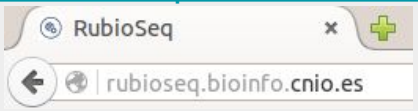
```
sudo apt-get install libmysqlclient-dev
```

Quick Manual Installation

Dependencies (Ubuntu system)

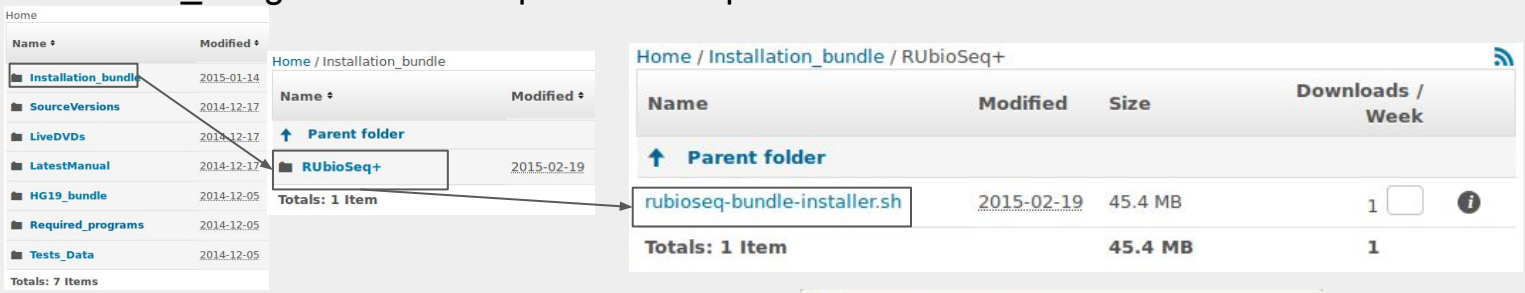
Download the installer:

1. Open  **Firefox**, and **visit** the **RUbioSeq's Home page** (www.rubioseq.bioinfo.cnio.es):



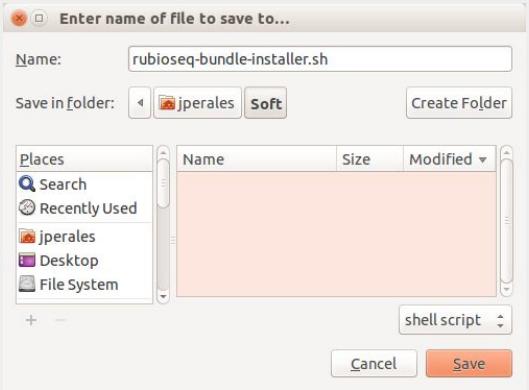
2. Click on Download from the menu options.

3. Navigate through the directory structure and download the automatic installer following:
   Installation_bungle > RUbioSeq+ > rubioseq-bundle-installer.sh



4. Select **Save File**.
   Locate your Home.
   Create Folder in your Home called **Software**
   (/home/USERNAME/Software)

   Save the file in said directory.



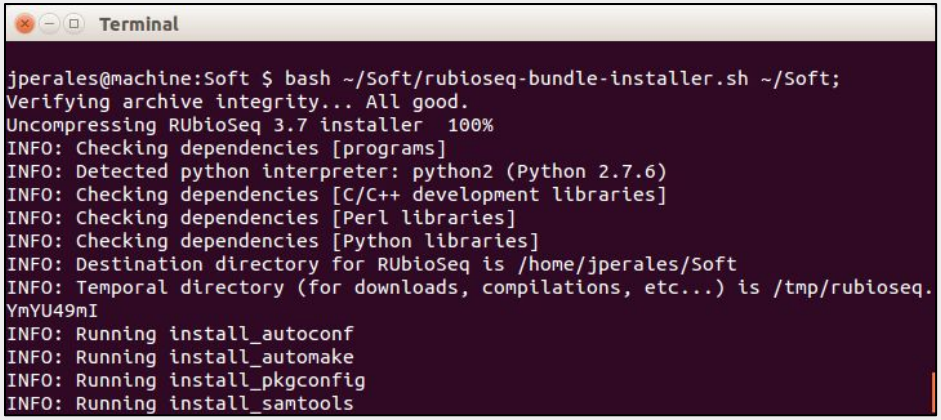
Quick Manual Installation

Dependencies (Ubuntu system)

Run the installer:

1. Open a terminal
2. Execute a bash **command**:

```
bash /home/YOUR_USER/Software/rubioseq-bundle-installer.sh
/home/YOUR_USER/Software/;
```

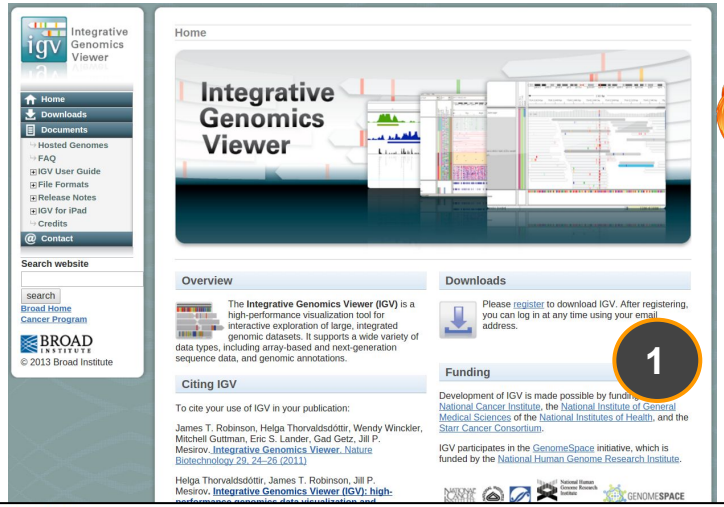


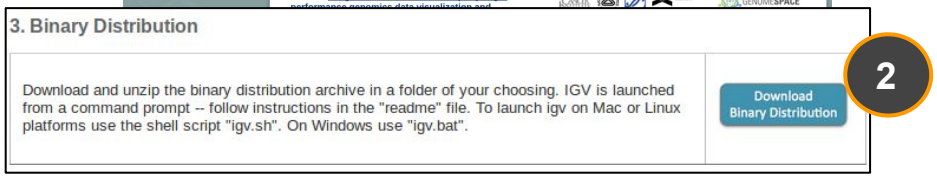
This will also configure the **PATHS** to the programs.

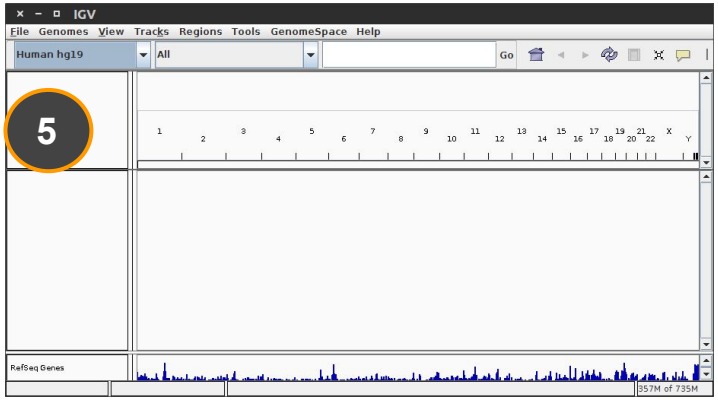
3. When it finishes, you can try it out using:

```
bash /home/YOUR_USER/Software/RUbioSeq3.7/RUbioSeq.pl --help
```

Integrative Genomics Viewer (IGV)







Open Firefox.
https://www.broadinstitute.org/igv/

1. Download section: Register and Fill out the form.

2. Download the Binary distribution: file.zip
Alternatively, you can use this link:
http://data.broadinstitute.org/igv/projects/downloads/IGV_2.3.66.zip

3. Extract the Zip file.

4. Go to the new Directory. Click on igv.jar