Scientific Programming Practical 1 (QCB)

Introduction

Luca Bianco - Academic Year 2020-21
luca.bianco@fmach.it

Outline

- Personal introduction
- Introduction to the practical
- Hands-on practical

About me

Computer Science

Ph.D. at the University of Verona, Italy, with thesis on Simulation of Biological Systems

Research Fellow at Cranfield University - UK

Three years at Cranfield University working at proteomics projects (GAPP, MRMaid, X-Tracker...)

Module manager and lecturer in several courses of the MSc in Bioinformatics

Bioinformatician at IASMA – FEM

Currently bioinformatician in the Computational Biology Group at Istituto Agrario di San Michele all'Adige – Fondazione Edmund Mach, Trento, Italy

Collaborator uniTN - CiBio

I ran the Scientific Programming Lab for QCB for the last four years

Fondazione Edmund Mach

FEM – San Michele, Trento - Italy

Agricultural Institute

Research and Innovation Centre

Genomics, metabolomics wet labs on fruits (apple, grape, small fruits,…)

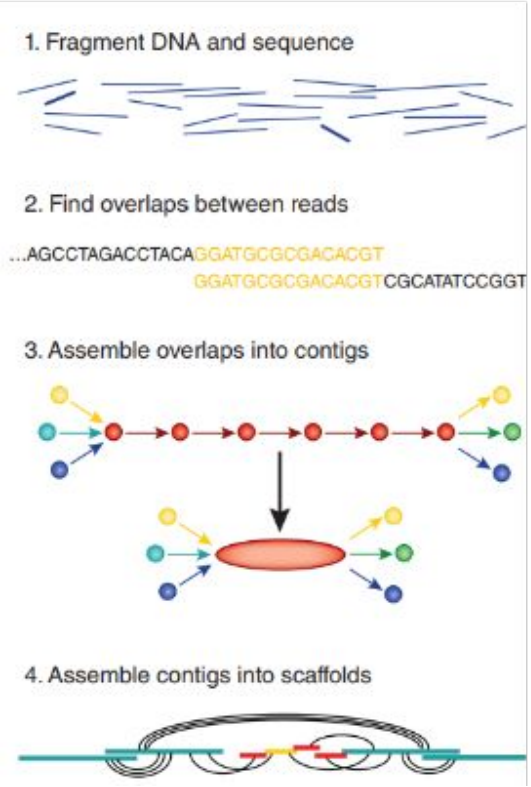
Bioinformatics and computational biology

Bioinformatics @FEM (UBC)

- Genomics
  - Assembly and annotation of complex genomes (plants, insects, etc.)
  - Development of SNP Chips for genetic screening
  - Resequencing of genomes / Variant discovery
- Metagenomics
  - Targeted metagenomic data
  - *Feature selection* algorithms
  - Algorithms for strain-level identification from un-targeted metagenomics
- Transcriptomics
  - RNA-seq data analysis, gene and pathway enrichment
  - Data integration and compilation of expression atlases
- Metabolomics
  - Data analysis pipelines for targeted and untargeted data
  - Methods for MS imaging
- Statistical data analysis
  - Integration of –omic data and analysis of correlation networks

Bix @FEM - Examples

Genome assembly



In a nutshell… (Tunis' version…)

Reads

Assembled genome

[Virgil and the Muses, Bardo Museum, Tunis]

[from M. Baker, Nature Methods, 2014]

Bix @FEM - Examples

Genome assembly of DH of Pear and Apple

Input data:

Illumina: ~60x – 100x PE information + (mate pairs for Apple)
Pacific Biosciences ~ 30x + 30x (35x only for Apple)
Bionano optical maps: ~ 600x (for both)
Hi-C: pear only
Genetic maps: integrated genetic map from 21 mapping populations (Apple only)

Output result (example for Apple):

Chromosome scale assembly

 # Contigs: 2150 for a total of **625Mb**
N50 Contigs (hybrid dbg2olc): ~ **620Kbps**
280 **Scaffolds**, for an N50 **5,6Mb**
17 chromosomes + lg0 unanchored sequences

[Daccord et al, Nature Genetics, 49, 2017; Linsmith et al., GigaScience, 2020]

Bix @FEM - Examples

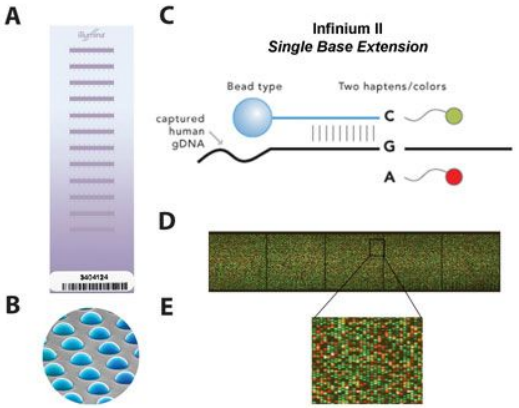
SNP-Chips development for GWAS

20K SNP Illumina Infinium II Array (reseq of 16 Apple cultivars, Illumina 30x)
487K SNP Affymetrix Axiom Array (reseq of 63 Apple cultivars, Illumina 20-30x)
600K SNP Affymetrix Axiom Array Walnut (reseq. 18 cultivars, Illumina 80x)

1. Reads alignment and filtering
2. SNP calling
3. Identification of most reliable SNPs
4. Selection of (20K) 487K target SNPs

Several Terabytes of data produced!!!!

Peach, pear and walnut done too!



[Bianco et al., PloS One, 2014; Bianco et al., the Plant Journal 2016; Marrano et al., the Plant Journal 2018]

Bix @FEM - Examples

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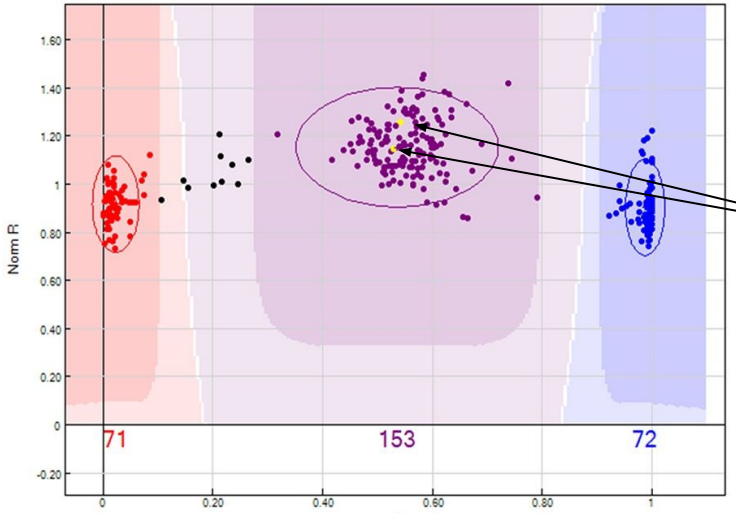
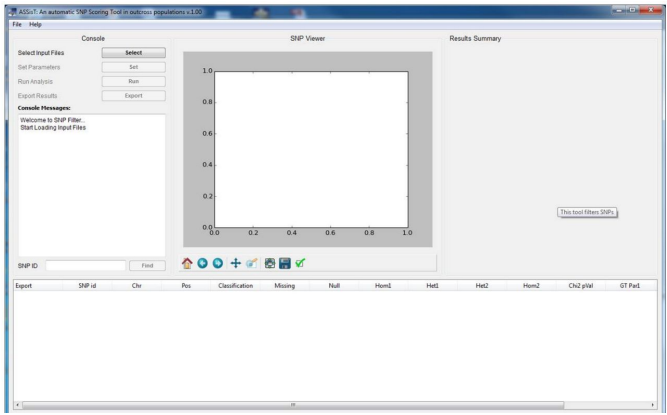


Task:
Analyze 500,000 of these…
(1 x SNP)

parents

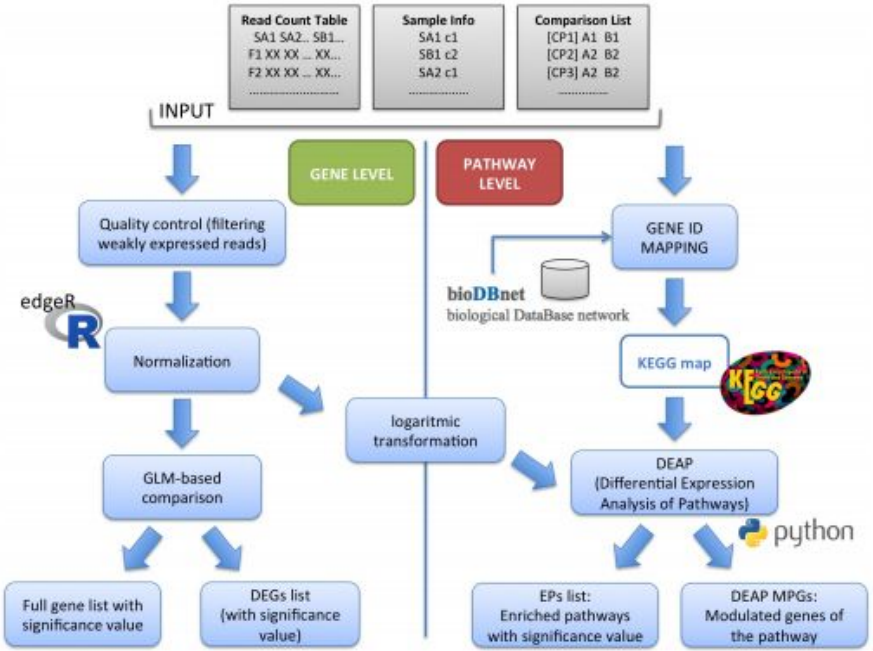
ASSIsT



[Di Guardo et al., Bioinformatics, 2015]

Bix @FEM - Examples

RNAseq data analysis with Pathway Inspector

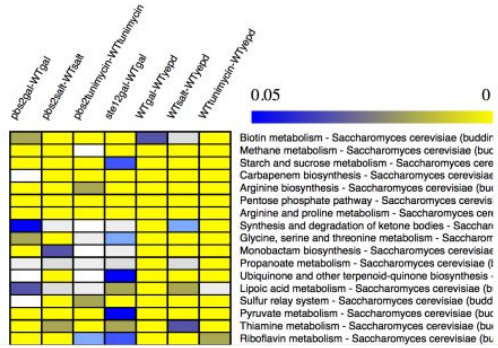


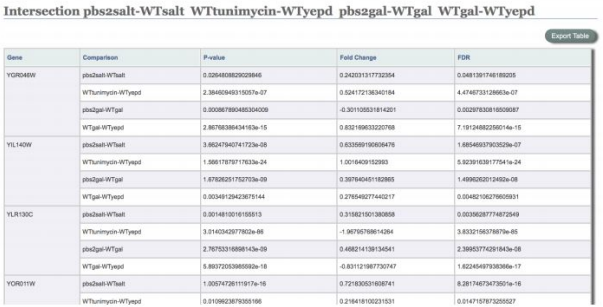
https://pathwayinspector.fmach.it

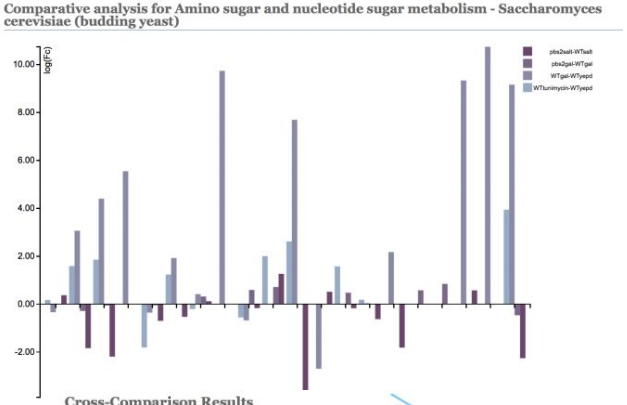
[Bianco et al., Bioinformatics, 2017]

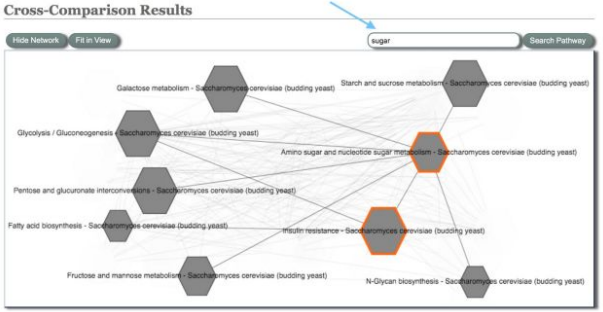
Bix @FEM - Examples

RNAseq data analysis with Pathway Inspector









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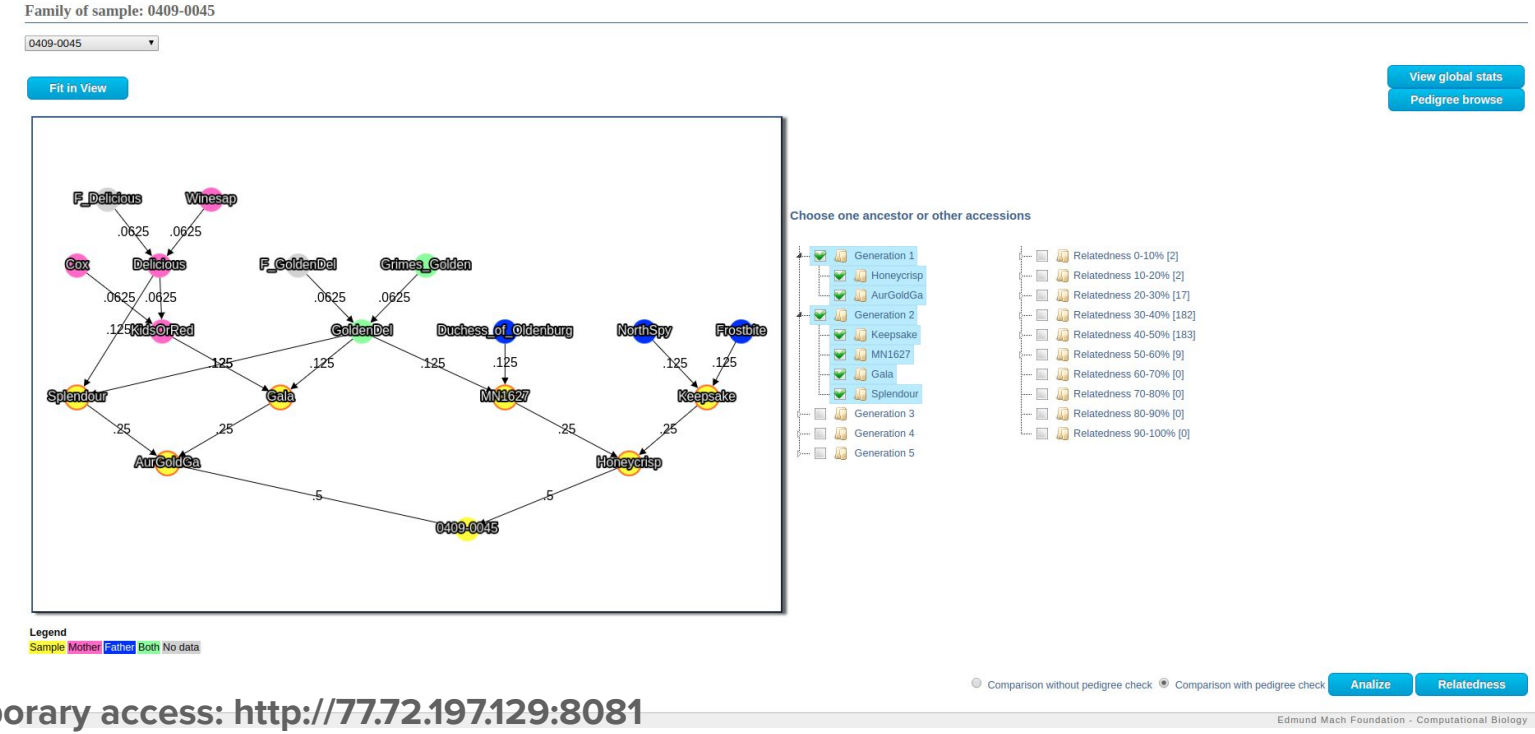
Bix @FEM - Examples

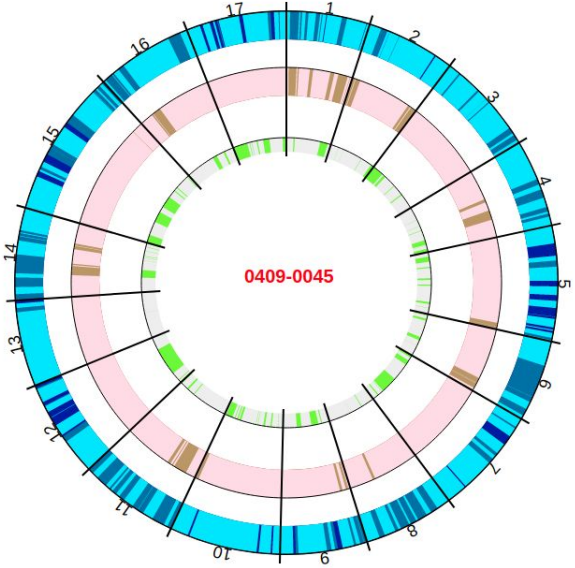
Pedigree-based haplotype visualization

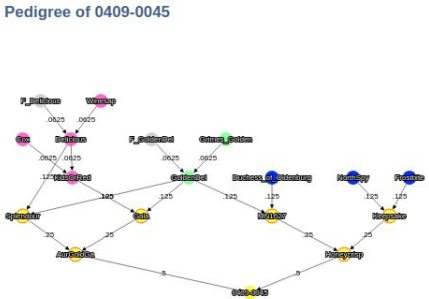


Temporary access: http://77.72.197.129:8081

Bix @FEM - Examples

Pedigree-based haplotype visualization





Cultivars	Relatedness	Color
MN1627	0.39	1CE6FF
AurGoldGa	0.55	FF4A46
Keepsake	0.42	006FA6
Gala	0.48	FFDBE5
Honeycrisp	0.52	0000A6
Splendour	0.48	B79762

Temporary access: http://77.72.197.129:8081

Bix @FEM - Examples

Sequencing and assemblying of Sars-Cov-2 samples from the Province of Trento (sponsored by Fondazione VRT)

Artic networks V3 protocol amplification & sequencing of 98, ~400bps long sequences overlapping by ~20bps

basecalling & de-multiplexing with guppy

"quasi"-real time sequencing run checking with rampart

RAMPART

post-processing and consensus calling with artic networks pipeline and IRMA assembler

ARTICnetwork

Nextstrain

down-stream analysis: multiple-sequence alignment, nextstrain, deepVariant,...

Bix @FEM - Examples

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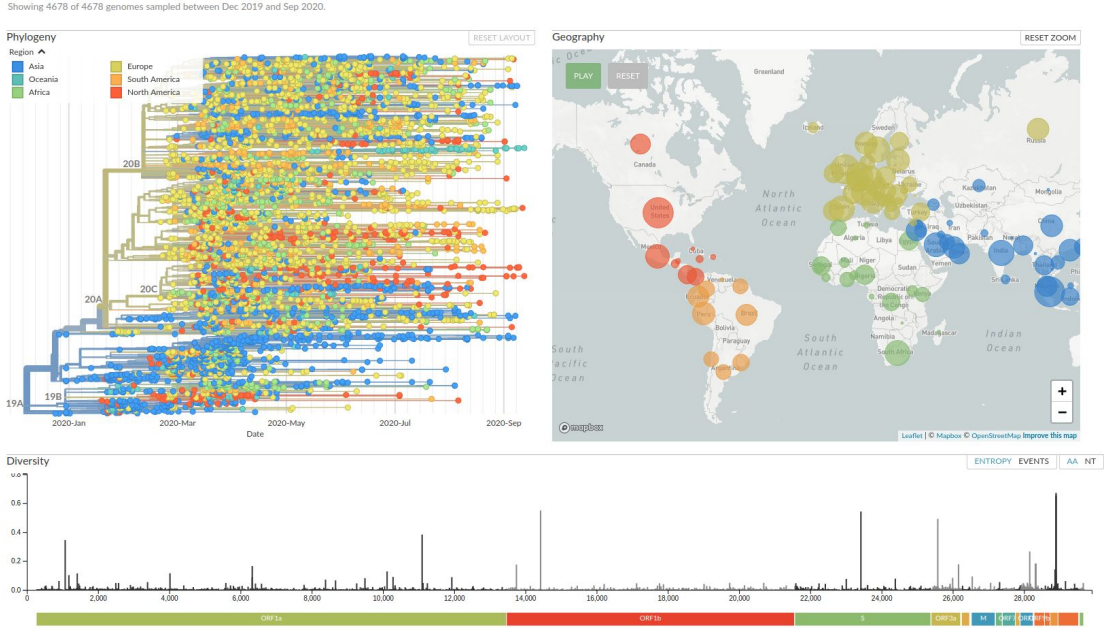


Opportunities @FEM

MSc External thesis

Are you interested in a bioinformatics project in NGS data analysis, RNA Seq, data integration?

Talk to me or email me at:

luca.bianco@fmach.it

Scientific Programming Practical

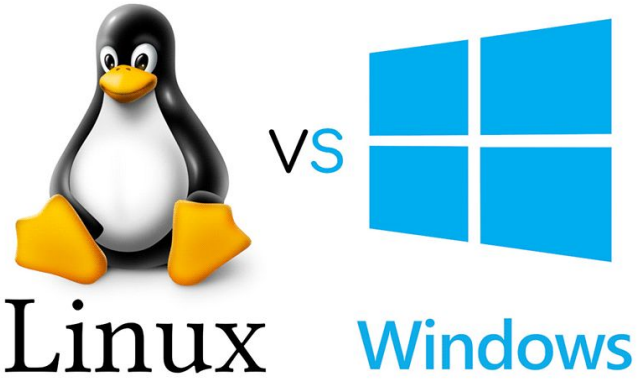
Back to business now!

Scientific Programming Practical

In this practical you will

1. Install Python 3.x (and pip)
2. Install Visual Studio Code
3. Get familiar with the Python console
4. Start using Visual Studio Code and advanced features (like debugging)
5. End the session with some exercises

Linux or Windows?



Up to you, as far as this course is concerned...

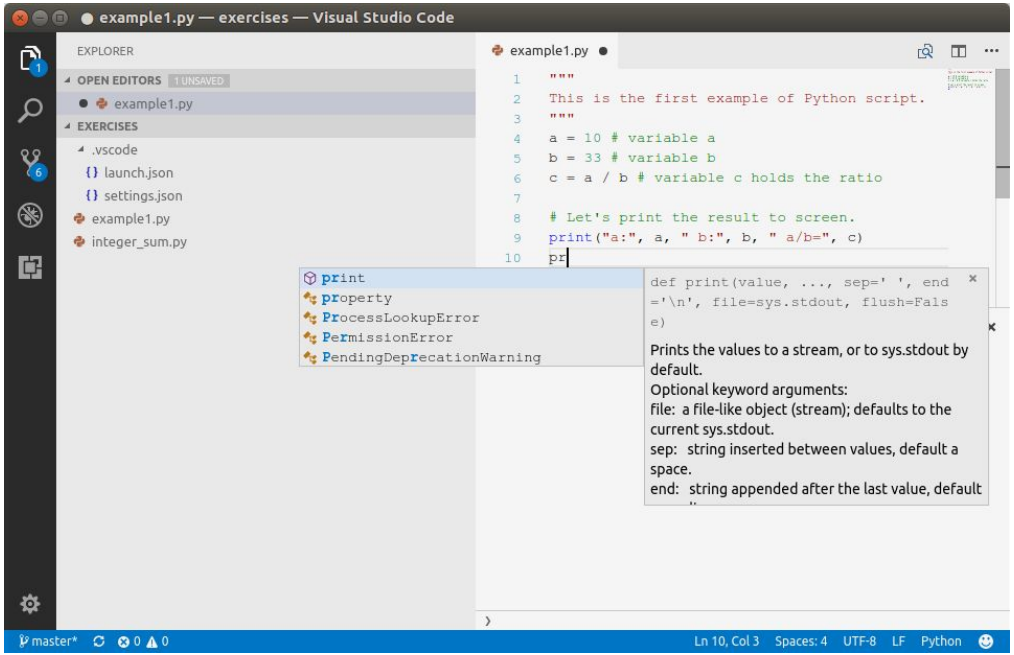
if you are looking for a career in bioinformatics I think it would be a good idea to get familiar with Linux

Scientific Programming Practical

Console VS. Integrated Development Environment (IDE)

```
biancol@bluhp:~$ python3
Python 3.5.2 (default, Aug 18 2017, 17:48:00)
[GCC 5.4.0 20160609] on linux
Type "help", "copyright", "credits" or "license" for more information.
>>>
```

```
>>> print("Hi there")
Hi there
>>> print("{} + {} = {}".format(10,20, 10+20))
10 + 20 = 30
>>>
```



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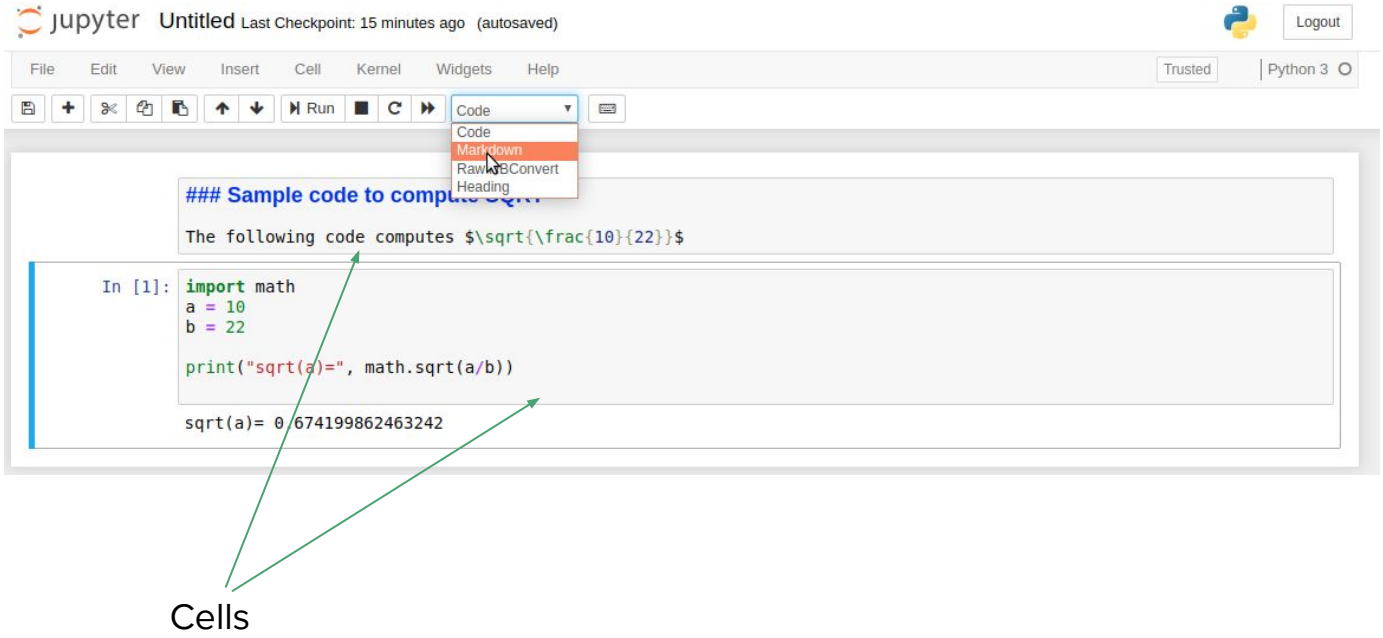
The debugger

Notebooks and Jupyter

“Jupyter is a web-based interactive development environment for python/R.. notebooks, code, and data.”

Notebooks contain both the **code**, some **text describing the code** and the **output of the code execution**,

Jupyter is becoming the de-facto standard for writing technical documentation.



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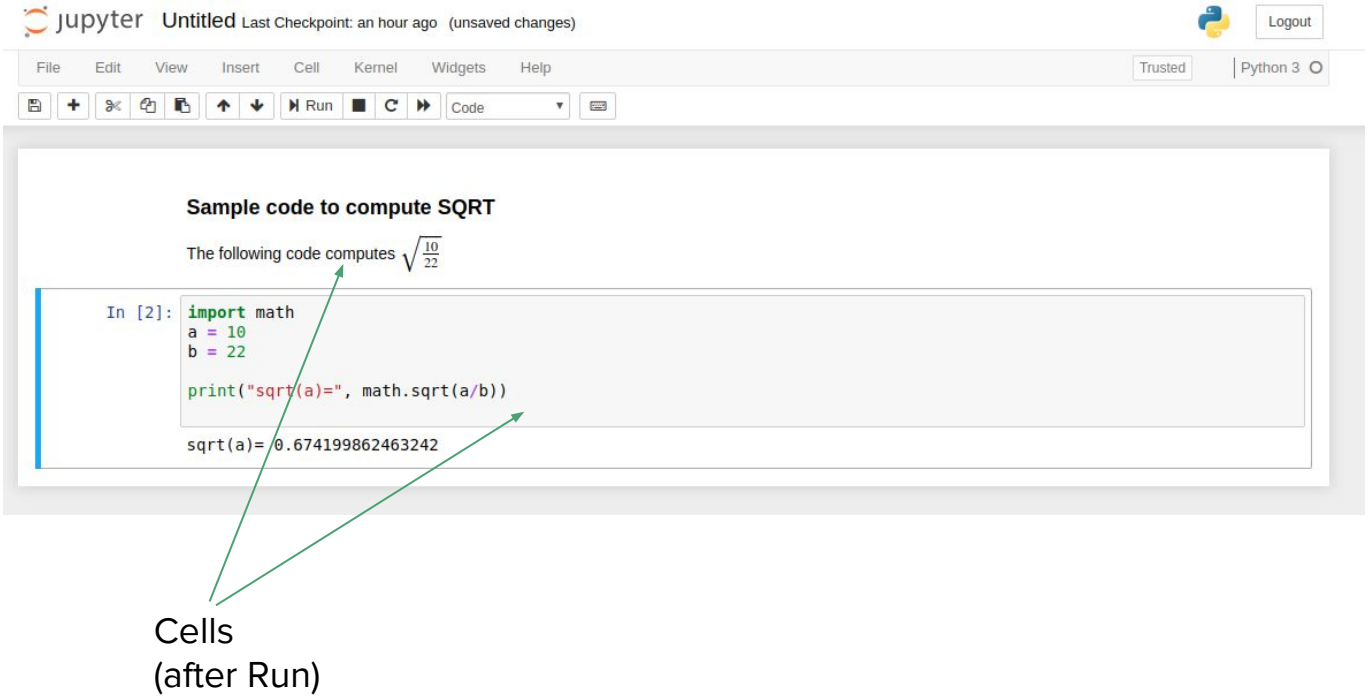
A cell can be executed by clicking on **Run**



Resources

All material regarding practicals will be found here:

http://qcbsciprolab2020.readthedocs.io

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Scientific Programming for QCB

Download: PDF EPUB HTML

General Info

The contacts to reach me can be found at this page.

Timetable and lecture rooms

Due to the current situation regarding the Covid-19 pandemic, Practicals will take place ONLINE this year. They will be held on **Mondays from 14:30 to 16:30** and on **Wednesdays from 11:30 to 12:30**.

Practicals will use the Zoom platform (https://zoom.us/) and the link for the connection will be published on the practical page available in this site a few minutes before the start of the session.

This first part of the course will tentatively run from **Wednesday, September 23rd, 2020 to Monday, November 2nd, 2020**.

Moodle

In the moodle page of the course you can find announcements and videos of the lectures. It can be found here.

Zoom links

The zoom links for the practicals can be found in the Announcements section of the moodle web page. To get you started quickly, I report them here:

Join Zoom Meeting https://unitn.zoom.us/j/97253388646

Meeting ID: 972 5338 8646 Passcode: 794500

Slides

Slides of the practicals will be available on the top part of each practical page.

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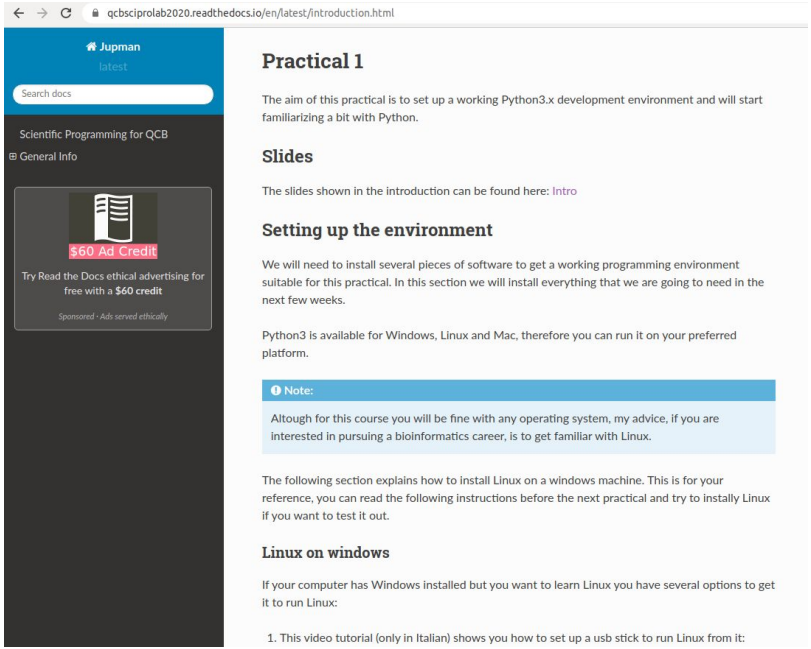
Any questions?

If not, please go to:

https://qcbsciprolab2020.readthedocs.io/latest/introduction.html



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