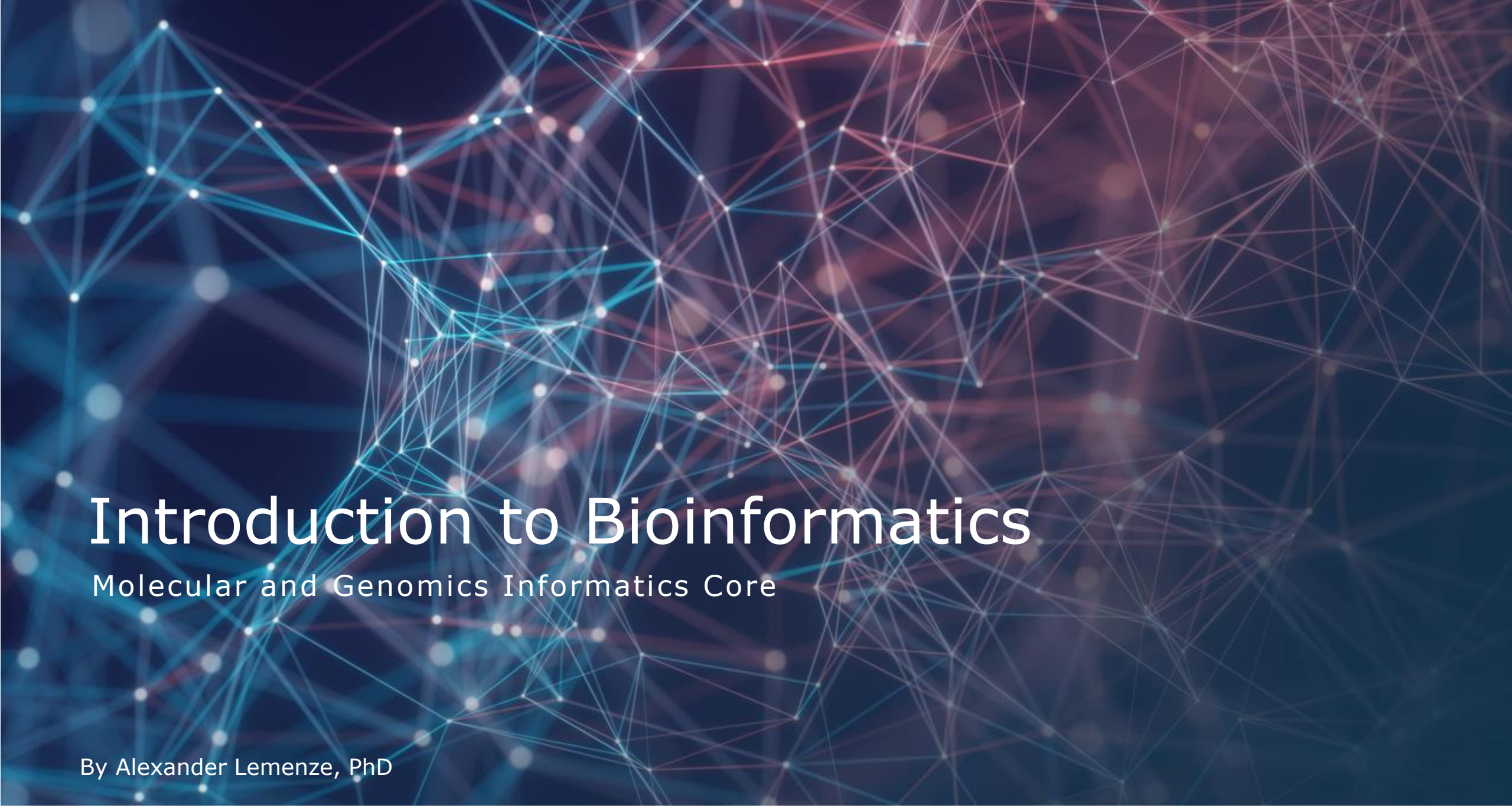


Introduction to Bioinformatics

Molecular and Genomics Informatics Core

By Alexander Lemenze, PhD

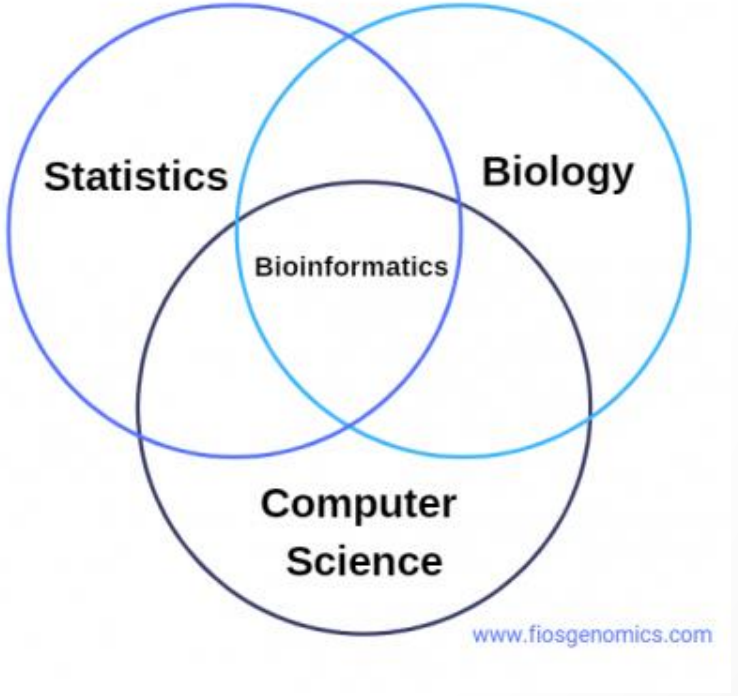
Our Journey Today

MEETING AGENDA

Computer Resources	Languages and basic access	Databases and Community
Basics computer components	What is a language	Soap box on reproducibility
Operating systems	What languages are commonly used	Common databases for use
Local and Cloud resources	How can I view my data (Genome Browsers)	Community help

What is Bioinformatics

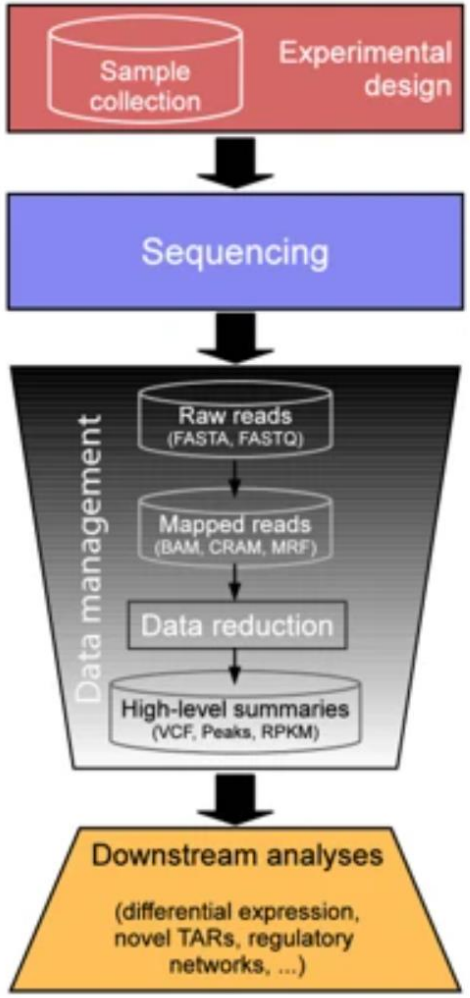
- Per google:
  - “the science of collecting and analyzing complex biological data such as genetic codes.”
- Types of “bioinformatics”
  - Biostatistician
  - Computational biologist
  - Algorithm development
  - Pipeliners

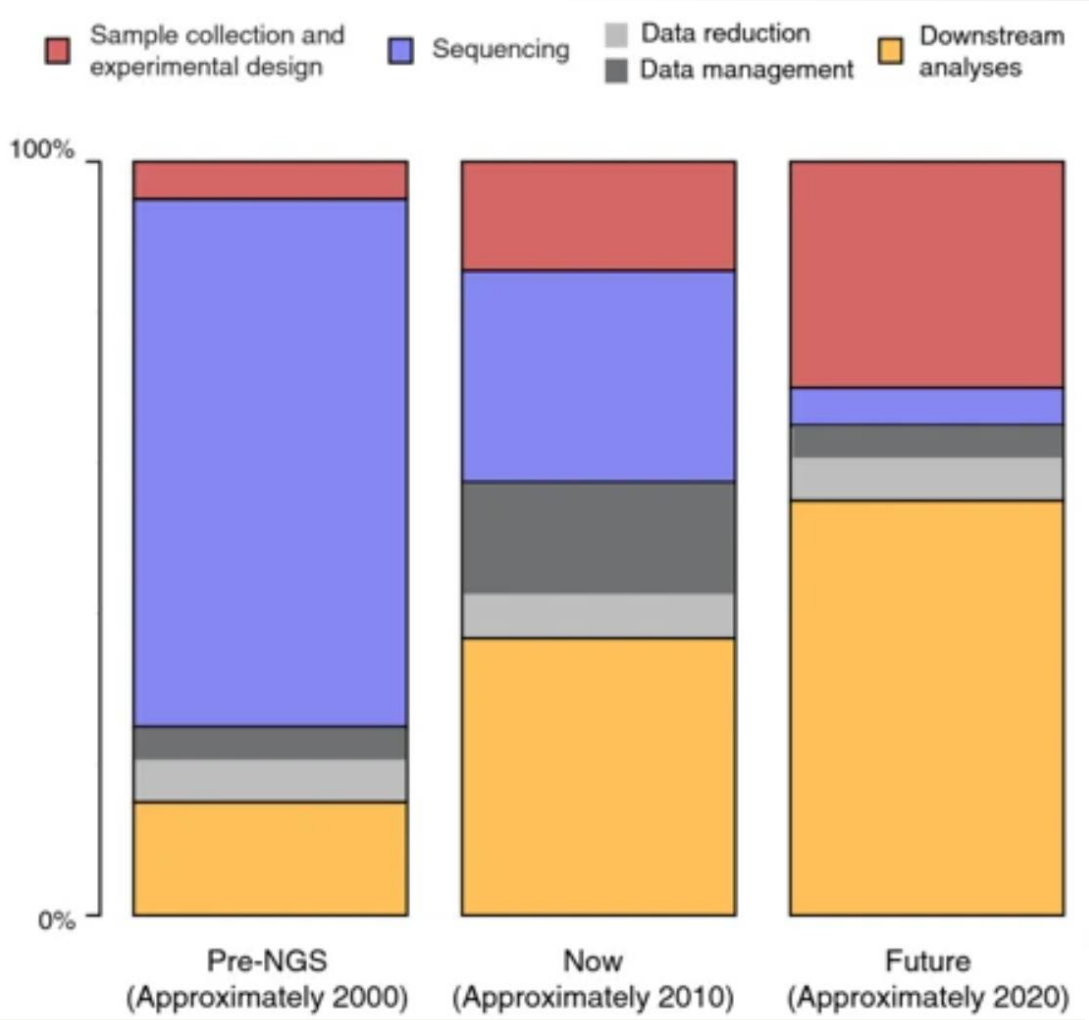


https://www.google.com/url?sa=i&url=https%3A%2F%2Fwww.fiosgenomics.com%2Fwhat-is-bioinformatics%2F&psig=AOvVaw1FTtX7ABW-pwl19ctee3fT&ust=1590504968582000&source=images&cd=vfe&ved=0CAIQjRxqFwoTCOi8iPCiz-kCFQAAAAAdAAAAABA2

Why Bioinformatics

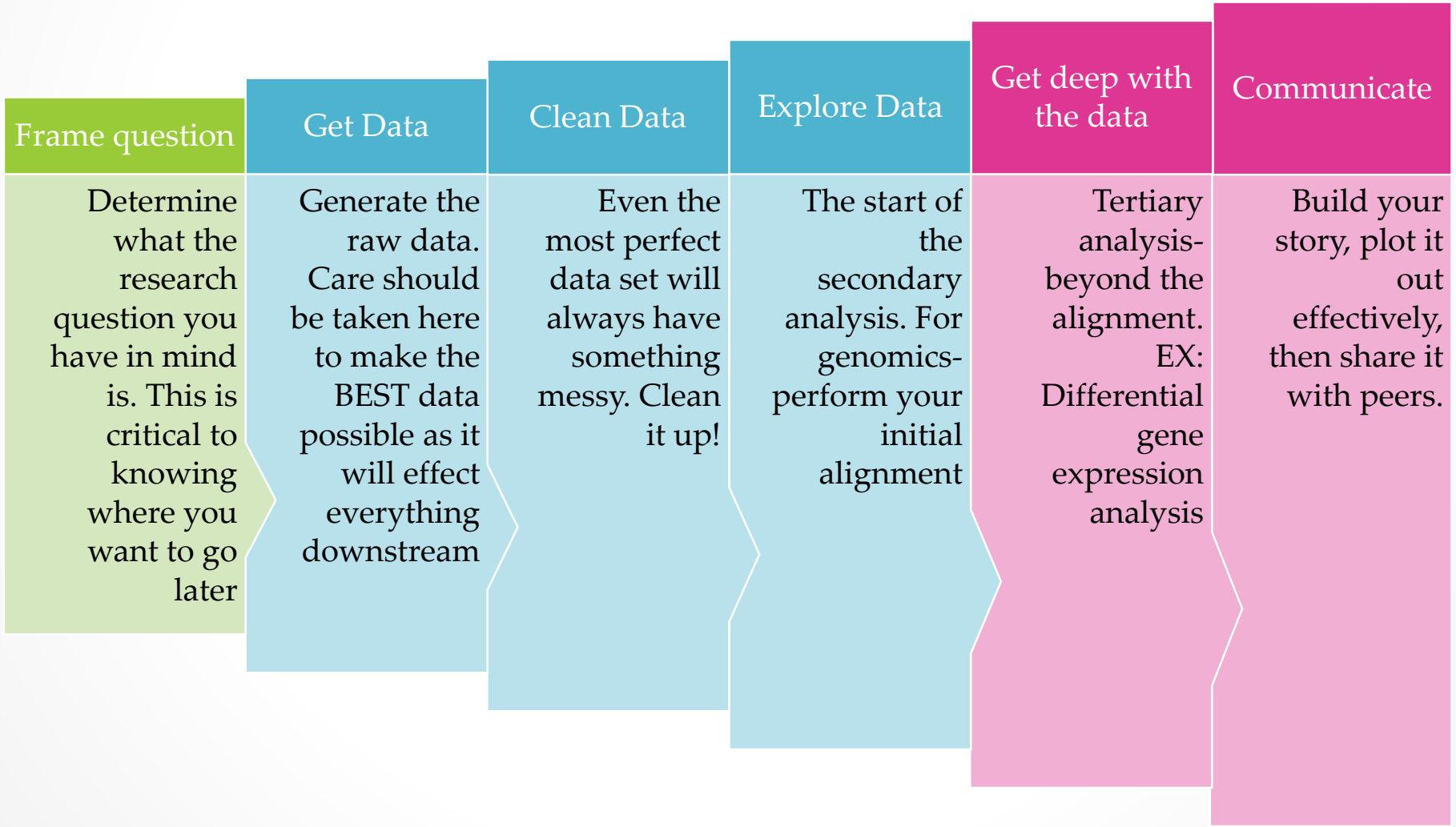
- Sequencing data is now more cost effective then ever.
- DATA DELUUGGGEEE!!





Sboner et al. Genome Biology 2011 12:125 doi:10.1186/gb-2011-12-8-125

Stages of data science



Computer Resources

The basics:

CPU

- Central Processing Unit
  - “Brains” of the computer
  - Terms associated:
    - Cores
    - Threads
    - Processors

Minimum:
24 cores

The basics:

RAM

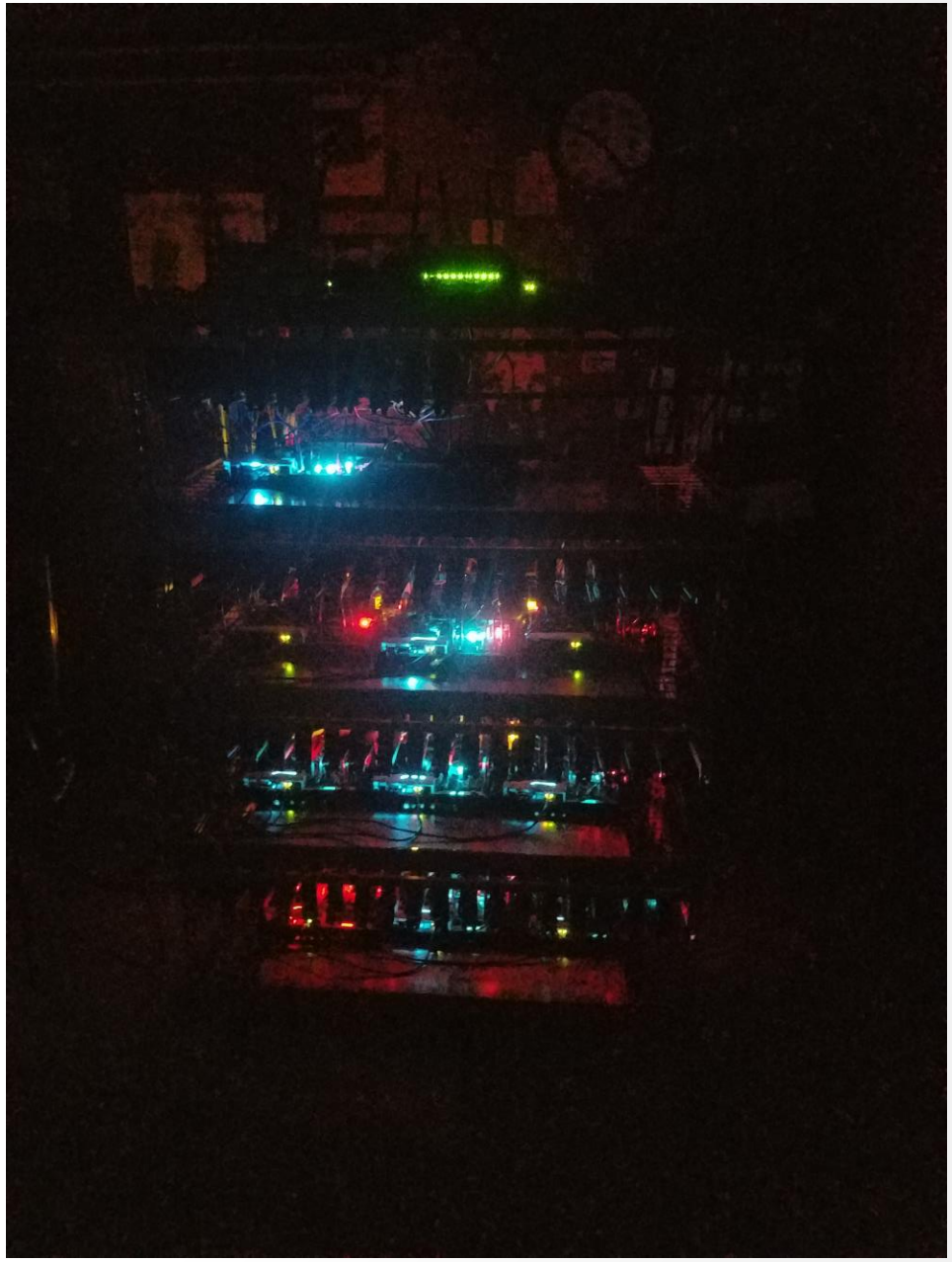
- Random-access memory
  - Thinking capacity of computer

Minimum:
64 Gb

The basics:

GPU

- Graphical Processing Unit
  - Usually for visual effects (hence the graphical)
  - Big for gaming and cryptocurrency
  - Can run algorithms more effectively in some cases
  - -> Process acceleration



The basics:

Hard drive

- Hard drive
  - Storage space!
- Estimated raw data per run:
  - Illumina:
    - ~20-200Gb/run
  - ONT:
    - ~2 Tb/run
    - Rich raw data

Minimum for a local machine:
10 Tb

Soap Box on Data management

Example Structure

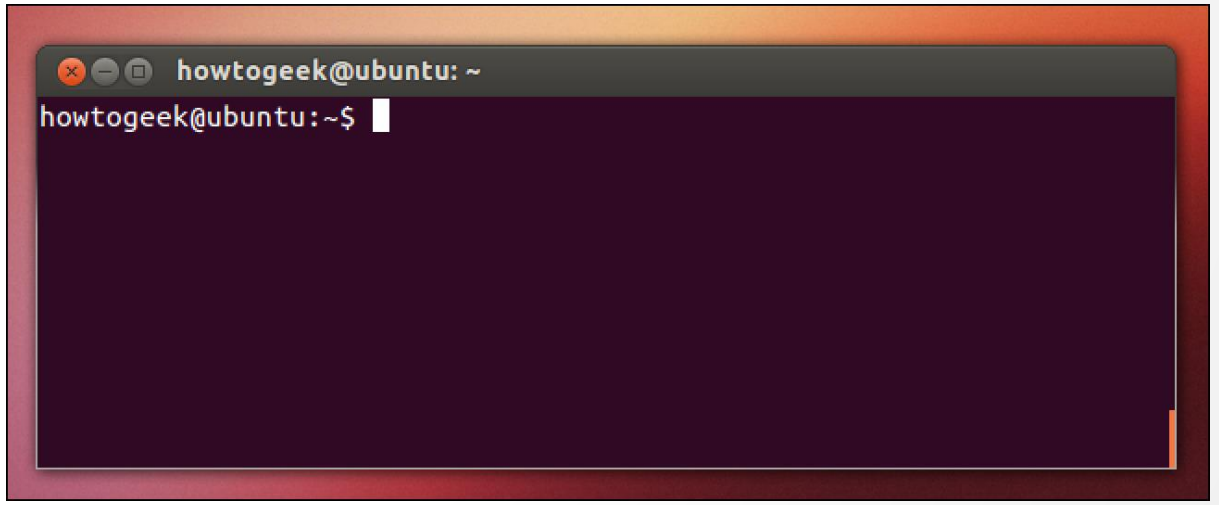
Operating Systems

- Windows
  - Infrequent for bioinformatics
  - Many tools incompatible
  - Drastically improving with virtual machines and linux integration
- Mac
  - Unix based like linux, more often used for bioinformatics and visuals.

- Linux
  - Flavors of Ubuntu, Centos, Redhat, etc etc
  - Unix based. Most common platforms for bioinformatics.

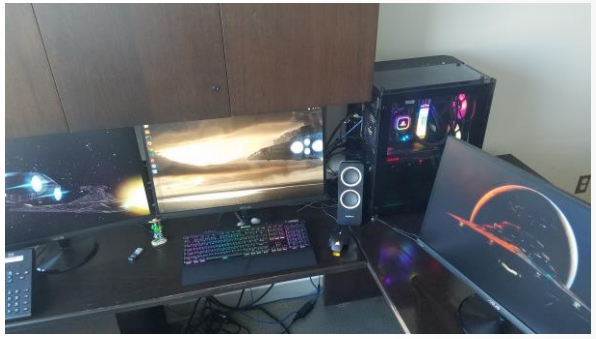
GUI vs CLI

- Graphical User Interface
  - Pretty
  - Easier to use
  - Slower
  - Limited to presets
- Command Line Interface
  - Not so pretty
  - Much faster
  - Flexible



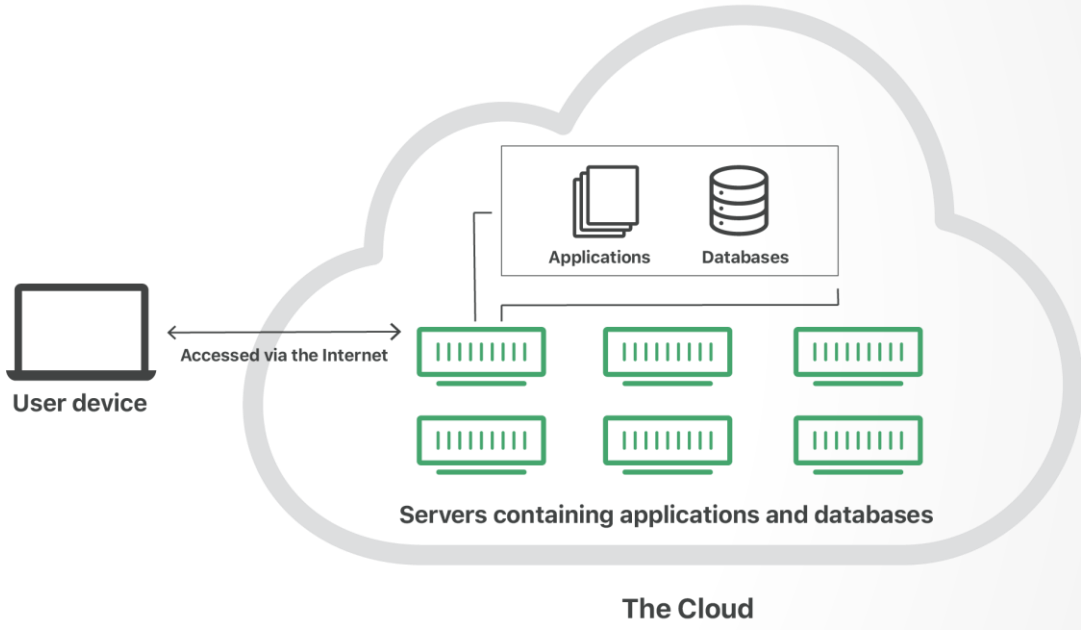
Local Resources

- Workstations
  - Easy to maintain personal set ups
  - Laptop portability
  - Expensive to have full bore station for each member
    - Usually defaults to a mid range station per person
- High Performance Compute Cluster
  - Intensive computer power
  - Scalable to needs
  - Shared amongst investigators to reduce downtime
  - Amarel @ Rutgers OARC



What is the Cloud

- Decentralized and distributed compute and storage that are accessed through the internet



https://www.cloudflare.com/learning/cloud/what-is-the-cloud/

Cloud Resources

- Amazon Web Services
  - AWS
- Google Cloud Platform
  - GCP
- Microsoft Azure Cloud
  - Azure

Languages and Basic tools

What is a programming language

- Computers speak in 1’s and 0’s
- Allows for logic to be constructed
  - Functions
  - Targets/objects
  - Data structures

Basic Language

Unix/Bash

- Standard operations
- First line scripting


```
BASH(1)                                General Commands Manual                                BASH(1)

NAME
       bash - GNU Bourne-Again SHell

SYNOPSIS
       bash [options] [command_string | file]

COPYRIGHT
       Bash is Copyright (C) 1989-2013 by the Free Software Foundation, Inc.

DESCRIPTION
       Bash  is  an sh-compatible command language interpreter that executes commands read from the standard input or
       from a file.  Bash also incorporates useful features from the Korn and C shells (ksh and csh).

       Bash is intended to be a conformant implementation of the Shell and Utilities portion of the IEEE POSIX speci-
       fication (IEEE Standard 1003.1).  Bash can be configured to be POSIX-conformant by default.

OPTIONS
       All  of  the   single-character  shell options documented in the description of the set builtin command can be
       used as options when the shell is invoked.  In addition, bash interprets the  following  options  when  it  is
       invoked:

       -c        If  the  -c  option  is  present,  then  commands  are  read from the first non-option argument com-
                 mand string.  If there are arguments after the command string, they are assigned to  the  positional
                 parameters, starting with $0.
       -i        If the -i option is present, the shell is interactive.
       -l        Make bash act as if it had been invoked as a login shell (see INVOCATION below).
       -r        If the -r option is present, the shell becomes restricted (see RESTRICTED SHELL below).
 Manual page bash(1) line 1 (press h for help or q to quit)
```

Basic Language

Python

- Higher level programming language
- General purpose and focused on readability
- Most common uses- full programs and data wrangling

Basic Language

R

- Another higher level language
- Primarily for statistical and graphical operations
- Most common uses then- maths and plotting

Basic Tools

Viewing your data

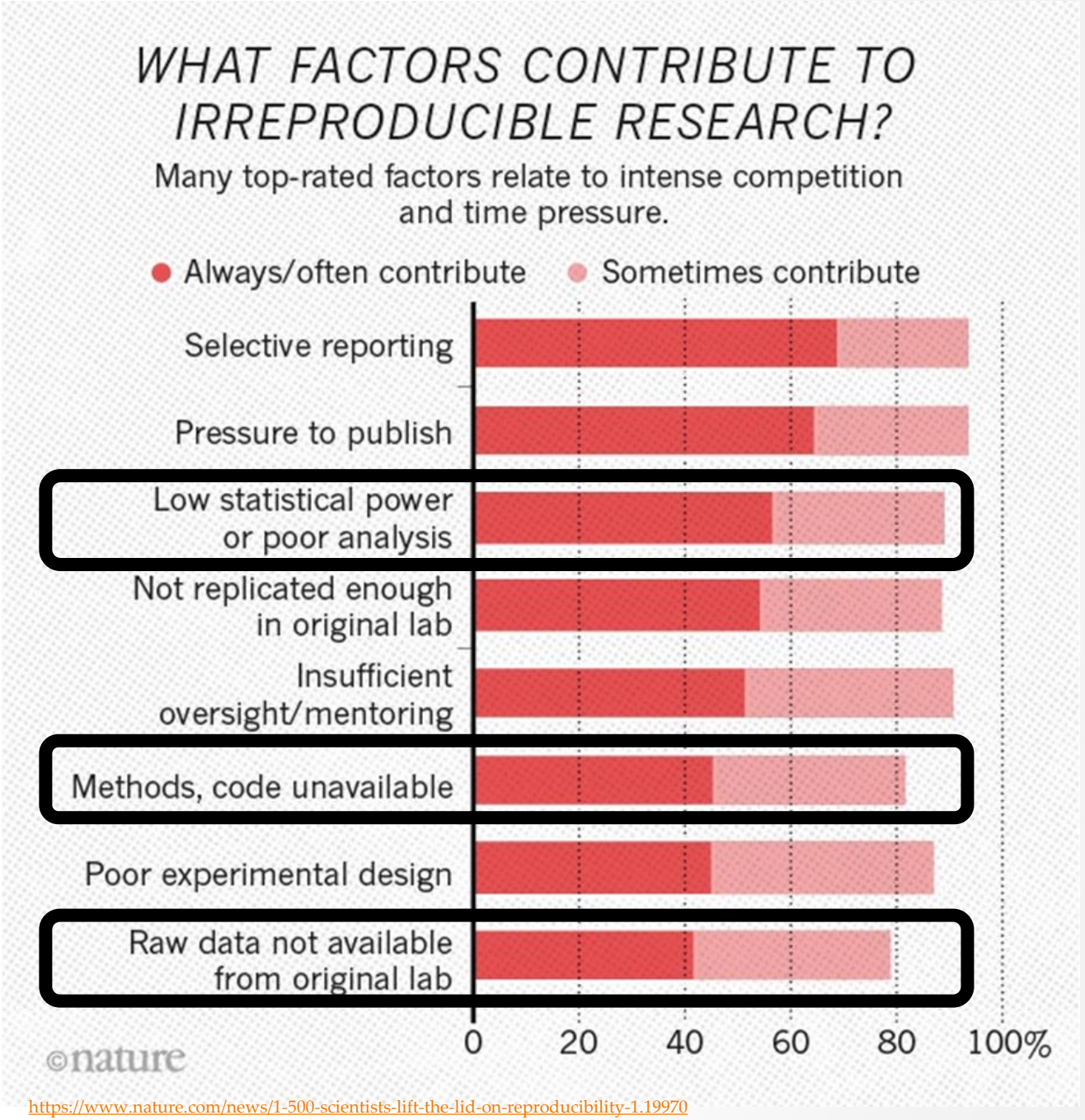
- Genome Browsers
  - Interactive Genome Browser (IGV) by the Broad
  - University of California Santa Cruz (UCSC) Genome Browser.

Databases and Community

Reproducibility

- Critically important for science as a whole

Bioinformatics plays into several major facets



https://www.nature.com/news/1-500-scientists-lift-the-lid-on-reproducibility-1.19970

Common databases

NCBI

- National Center for Biotechnology Information
  - Hosts end point data
  - Like Pubmed!
    - And a variety of other data sets
  - Genome references
  - BLAST

Common databases

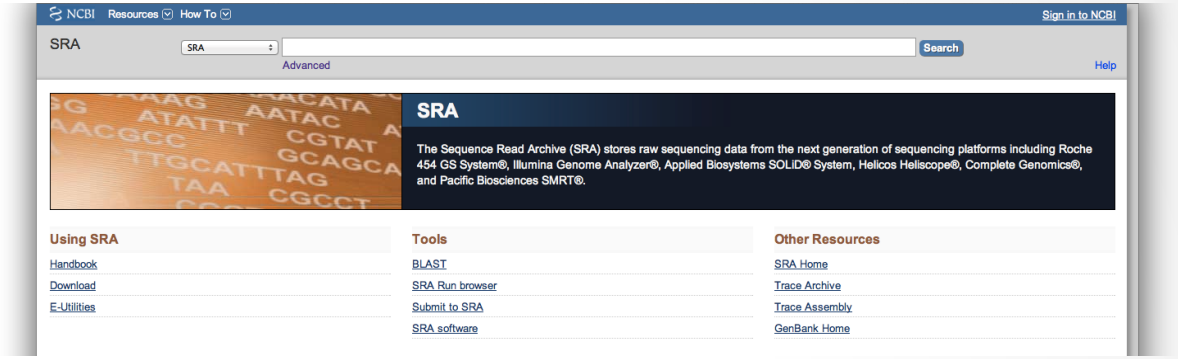
GEO

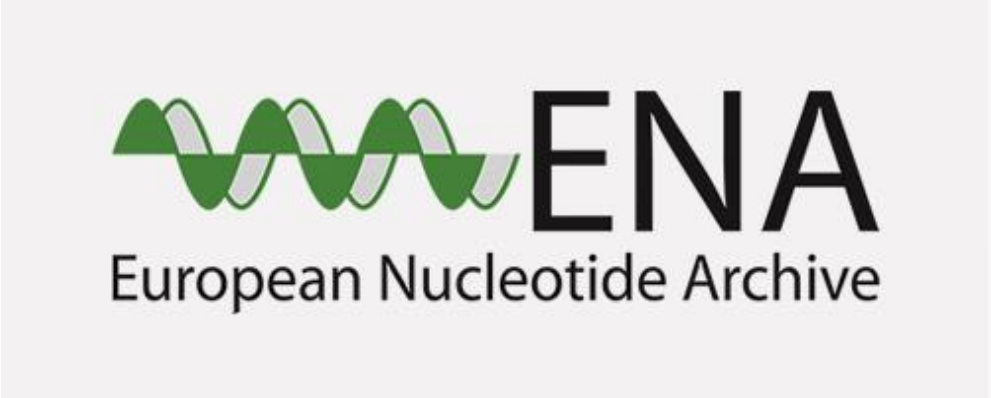
- Gene Expression Omnibus
  - Hosted by NCBI
  - Repository for experiments of expression data
  - “Easily” queriable
  - Currently >3.6 million individual samples

Common databases

SRA/ENA

- Short Reads Archive
  - NCBI hosted
- European Nucleotide Archive
  - European Bioinformatics Institute
    - Also hosts ENSEMBL





Community

Reach out for help

- Biostars
- Stackoverflow
- Google!

DOCTORS:

PROGRAMMERS:


THANK YOU