# BGGN 213

## Foundations of Bioinformatics

Barry Grant

UC San Diego

http://thegrantlab.org/bggn213

HELLO
my name is
BARRY
bjgrant@ucsd.edu

HELLO
HIS ~~my~~ name is
YUANSHENG
yuz461@ucsd.edu

# Introduce Yourself!

Your preferred name,
Place you identify with,
Major area of study/research,
Favorite joke (optional)!

# Today’s Menu

| | |
|---|---|
| **Course Logistics** | Website, screencasts, survey, ethics, assessment and grading. |
| **Learning Objectives** | What you need to learn to succeed in this course. |
| **Course Structure** | Major lecture topics and specific leaning goals. |
| **Introduction to Bioinformatis** | **Introducing the *what, why* and *how* of bioinformatics?** |
| **Bioinformatics Database** | **Hands-on** exploration of several major databases and their associated tools. |

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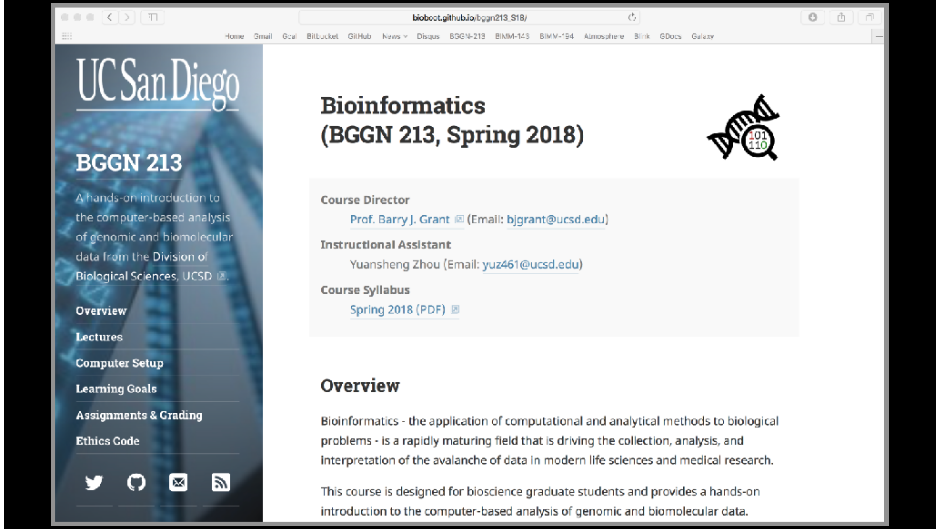
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BGGN 213

A hands-on introduction to the computer-based analysis of genomic and biomolecular data from the Division of Biological Sciences, UCSD.

Overview
Lectures
Computer Setup
Learning Goals
Assignments & Grading
Ethics Code

## Bioinformatics
## (BGGN 213, Spring 2018)

**Course Director**
Prof. Barry J. Grant (Email: bjgrant@ucsd.edu)

**Instructional Assistant**
Yuansheng Zhou (Email: yuz461@ucsd.edu)

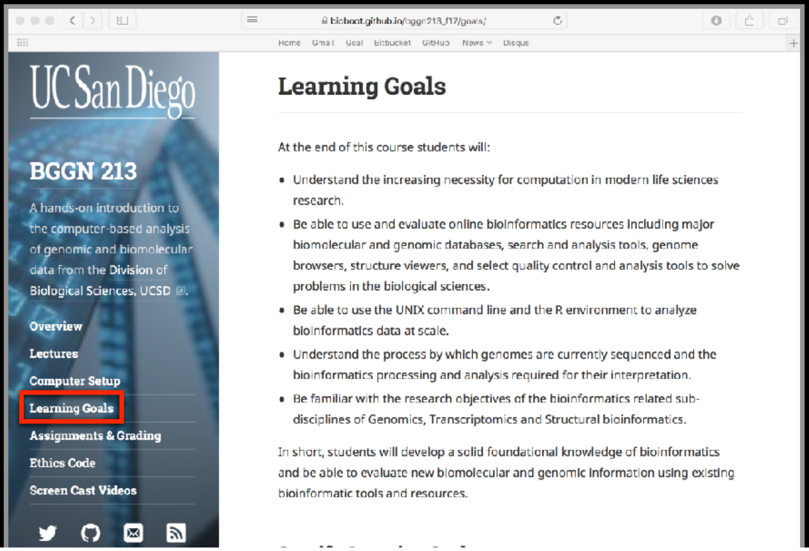
**Course Syllabus**
Spring 2018 (PDF)

## Overview

Bioinformatics - the application of computational and analytical methods to biological problems - is a rapidly maturing field that is driving the collection, analysis, and interpretation of the avalanche of data in modern life sciences and medical research.

This course is designed for bioscience graduate students and provides a hands-on introduction to the computer-based analysis of genomic and biomolecular data.

## What essential concepts and skills should YOU attain from this course?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## Learning Goals

At the end of this course students will:

- Understand the increasing necessity for computation in modern life sciences research.
- Be able to use and evaluate online bioinformatics resources including major biomolecular and genomic databases, search and analysis tools, genome browsers, structure viewers, and select quality control and analysis tools to solve problems in the biological sciences.
- Be able to use the UNIX command line and the R environment to analyze bioinformatics data at scale.
- Understand the process by which genomes are currently sequenced and the bioinformatics processing and analysis required for their interpretation.
- Be familiar with the research objectives of the bioinformatics related sub-disciplines of Genomics, Transcriptomics and Structural bioinformatics.

In short, students will develop a solid foundational knowledge of bioinformatics and be able to evaluate new biomolecular and genomic information using existing bioinformatic tools and resources.

## At the end of this course students will:

- Understand the increasing necessity for computation in modern life sciences research.
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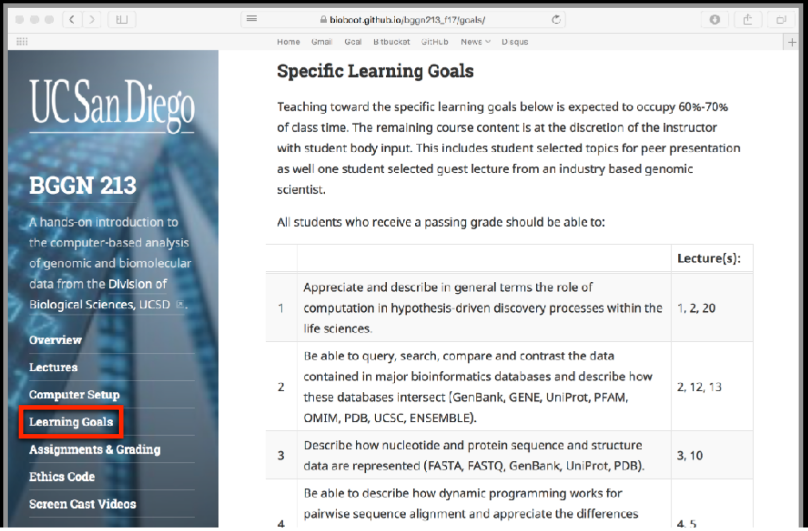
In short, you will develop a solid foundational knowledge of **bioinformatics** and be able to evaluate new biomolecular and genomic information using **existing bioinformatic tools and resources**.

# Specific Learning Goals….

What I want you to know by course end!



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## Specific Learning Goals

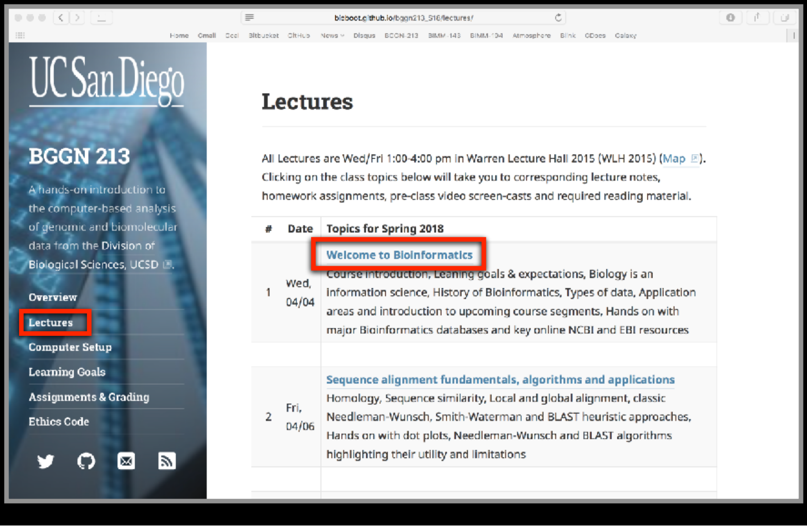
Teaching toward the specific learning goals below is expected to occupy 60%-70% of class time. The remaining course content is at the discretion of the instructor with student body input. This includes student selected topics for peer presentation as well one student selected guest lecture from an industry based genomic scientist.

All students who receive a passing grade should be able to:

| | | Lecture(s): |
|---|---|---|
| 1 | Appreciate and describe in general terms the role of computation in hypothesis-driven discovery processes within the life sciences. | 1, 2, 20 |
| 2 | Be able to query, search, compare and contrast the data contained in major bioinformatics databases and describe how these databases intersect (GenBank, GENE, UniProt, PFAM, OMIM, PDB, UCSC, ENSEMBLE). | 2, 12, 13 |
| 3 | Describe how nucleotide and protein sequence and structure data are represented (FASTA, FASTQ, GenBank, UniProt, PDB). | 3, 10 |
| 4 | Be able to describe how dynamic programming works for pairwise sequence alignment and appreciate the differences | 4, 5 |

# Course Structure

Derived from specific learning goals



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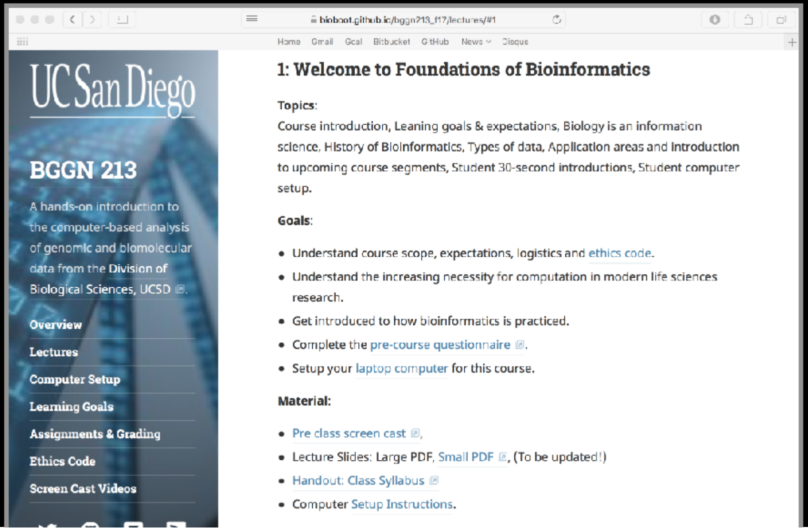
## Lectures

All Lectures are Wed/Fri 1:00-4:00 pm in Warren Lecture Hall 2015 (WLH 2015) (Map). Clicking on the class topics below will take you to corresponding lecture notes, homework assignments, pre-class video screen-casts and required reading material.

| # | Date | Topics for Spring 2018 |
|---|---|---|
| 1 | Wed, 04/04 | Welcome to Bioinformatics<br>Course introduction, Leaning goals & expectations, Biology is an information science, History of Bioinformatics, Types of data, Application areas and introduction to upcoming course segments, Hands on with major Bioinformatics databases and key online NCBI and EBI resources |
| 2 | Fri, 04/06 | Sequence alignment fundamentals, algorithms and applications<br>Homology, Sequence similarity, Local and global alignment, classic Needleman-Wunsch, Smith-Waterman and BLAST heuristic approaches, Hands on with dot plots, Needleman-Wunsch and BLAST algorithms highlighting their utility and limitations |

# Class Details

Goals, Class material, Screencasts & **Homework**



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## 1: Welcome to Foundations of Bioinformatics

**Topics**:
Course introduction, Leaning goals & expectations, Biology is an information science, History of Bioinformatics, Types of data, Application areas and introduction to upcoming course segments, Student 30-second introductions, Student computer setup.

**Goals**:

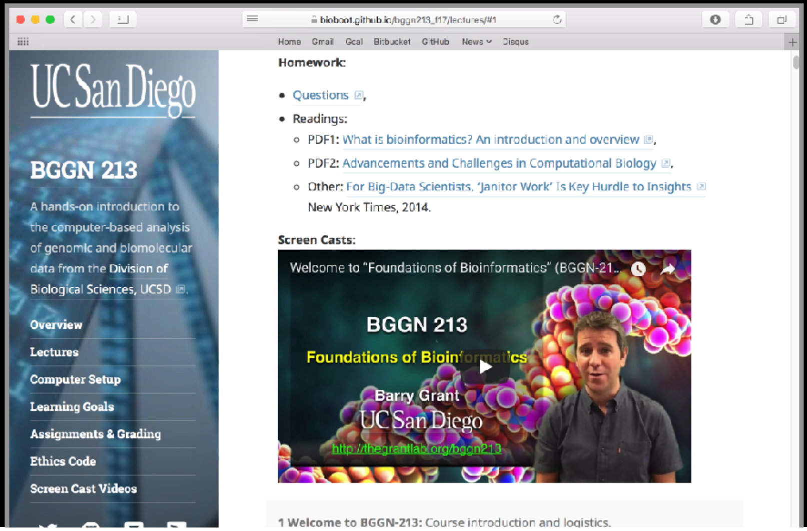
- Understand course scope, expectations, logistics and ethics code.
- Understand the increasing necessity for computation in modern life sciences research.
- Get introduced to how bioinformatics is practiced.
- Complete the pre-course questionnaire.
- Setup your laptop computer for this course.

**Material:**

- Pre class screen cast,
- Lecture Slides: Large PDF, Small PDF, (To be updated!)
- Handout: Class Syllabus
- Computer Setup Instructions.

# Homework

Goals, Class material, Screencasts & **Homework**



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**Homework:**

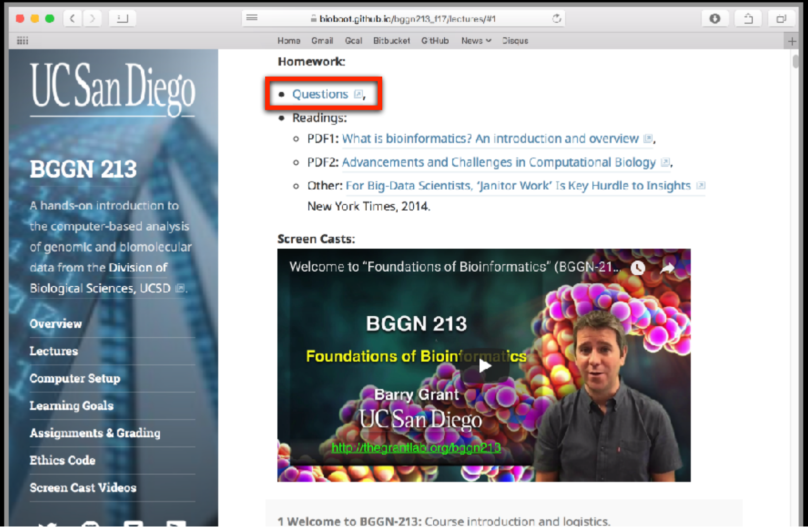
- Questions,
- Readings:
  - PDF1: What is bioinformatics? An introduction and overview,
  - PDF2: Advancements and Challenges in Computational Biology,
  - Other: For Big-Data Scientists, 'Janitor Work' Is Key Hurdle to Insights New York Times, 2014.

**Screen Casts:**

**1 Welcome to BGGN-213:** Course introduction and logistics.

# Homework

Goals, Class material, Screencasts & **Homework**



bioboot.github.io/bggn213_f17/lectures/#1

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  - PDF2: Advancements and Challenges in Computational Biology,
  - Other: For Big-Data Scientists, 'Janitor Work' Is Key Hurdle to Insights New York Times, 2014.

Screen Casts:

Welcome to "Foundations of Bioinformatics" (BGGN-21...

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Foundations of Bioinformatics

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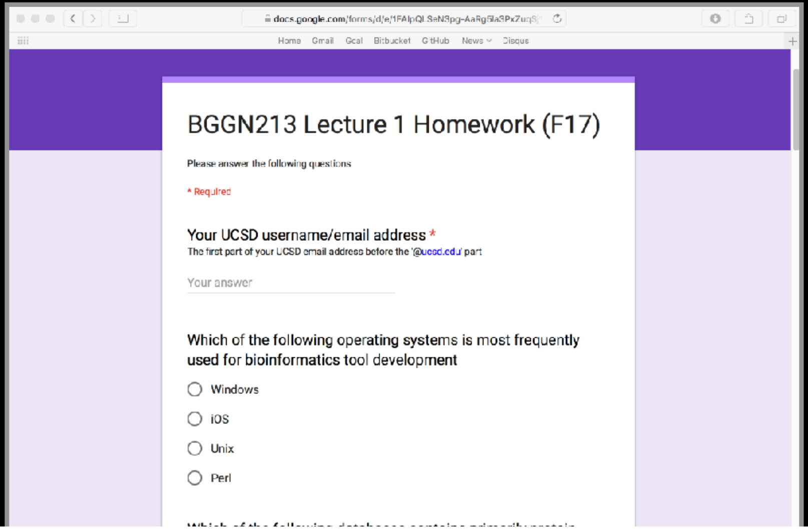
1 Welcome to BGGN-213: Course introduction and logistics.

# Homework

Goals, Class material, Screencasts & **Homework**



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BGGN213 Lecture 1 Homework (F17)

Please answer the following questions

* Required

Your UCSD username/email address *

The first part of your UCSD email address before the '@ucsd.edu' part

Your answer

Which of the following operating systems is most frequently used for bioinformatics tool development

○ Windows

○ iOS

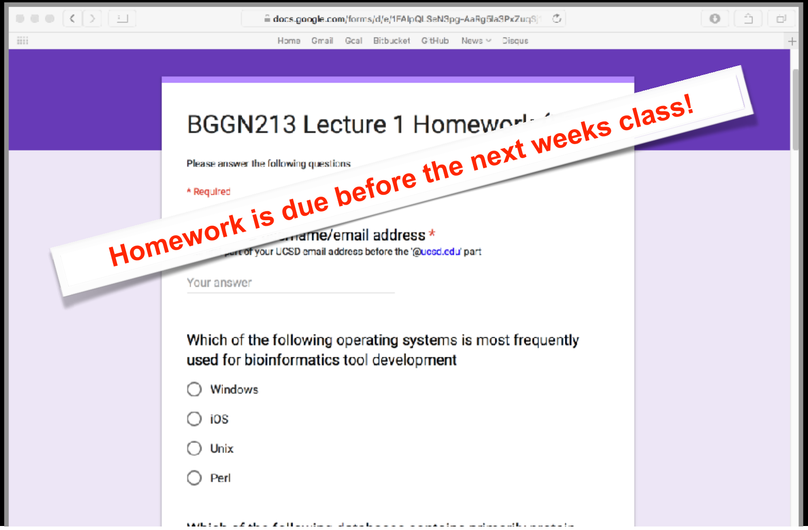
○ Unix

○ Perl

Which of the following databases contains primarily protein

# Homework

Goals, Class material, Screencasts & **Homework**



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BGGN213 Lecture 1 Homewor

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Your answer

Which of the following operating systems is most frequently used for bioinformatics tool development

○ Windows

○ iOS

○ Unix

○ Perl

Which of the following databases contains primarily protein

Homework is due before the next weeks class!

## Side Note: Why stick with this course?

**Provides a hands-on practical introduction to major bioinformatics concepts and resources.**

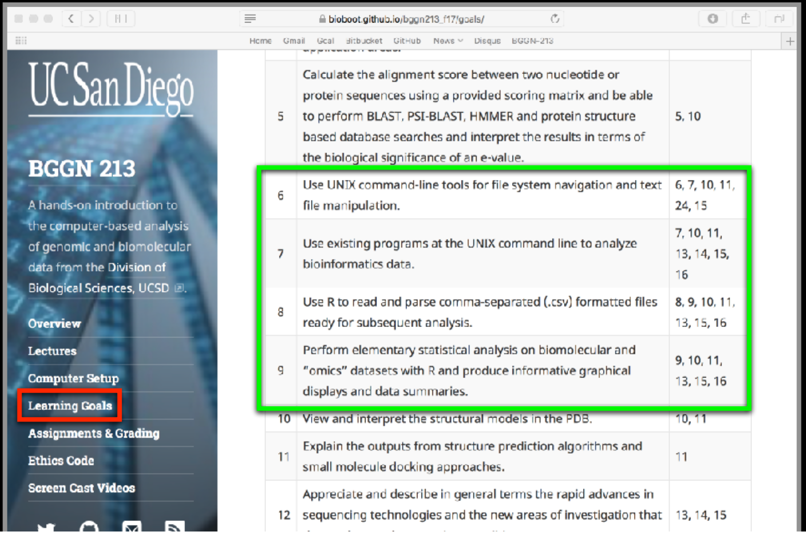
Covers modern hot topics and the intimate coupling of informatics with biology - highlighting the impact of computing advances and 'big data' on biology!

Designed for graduates in the biosciences with no programing experience or high level math skills.

Provides a hook for increasing computational and data science competencies in the biosciences - valuable high demand translational skills!

# BGGN-213 Learning Goals….

Advanced UNIX and R based learning goals



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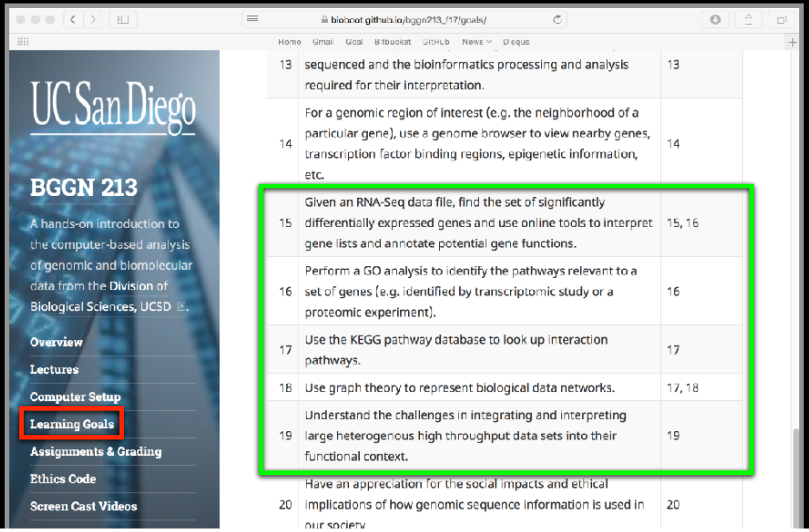
| | | |
|---|---|---|
| 5 | Calculate the alignment score between two nucleotide or protein sequences using a provided scoring matrix and be able to perform BLAST, PSI-BLAST, HMMER and protein structure based database searches and interpret the results in terms of the biological significance of an e-value. | 5, 10 |
| 6 | Use UNIX command-line tools for file system navigation and text file manipulation. | 6, 7, 10, 11, 24, 15 |
| 7 | Use existing programs at the UNIX command line to analyze bioinformatics data. | 7, 10, 11, 13, 14, 15, 16 |
| 8 | Use R to read and parse comma-separated (.csv) formatted files ready for subsequent analysis. | 8, 9, 10, 11, 13, 15, 16 |
| 9 | Perform elementary statistical analysis on biomolecular and "omics" datasets with R and produce informative graphical displays and data summaries. | 9, 10, 11, 13, 15, 16 |
| 10 | View and interpret the structural models in the PDB. | 10, 11 |
| 11 | Explain the outputs from structure prediction algorithms and small molecule docking approaches. | 11 |
| 12 | Appreciate and describe in general terms the rapid advances in sequencing technologies and the new areas of investigation that | 13, 14, 15 |

# BGGN-213 Learning Goals….

Delve deeper into "real-world" bioinformatics



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| | | |
|---|---|---|
| 13 | sequenced and the bioinformatics processing and analysis required for their interpretation. | 13 |
| 14 | For a genomic region of interest (e.g. the neighborhood of a particular gene), use a genome browser to view nearby genes, transcription factor binding regions, epigenetic information, etc. | 14 |
| 15 | Given an RNA-Seq data file, find the set of significantly differentially expressed genes and use online tools to interpret gene lists and annotate potential gene functions. | 15, 16 |
| 16 | Perform a GO analysis to identify the pathways relevant to a set of genes (e.g. identified by transcriptomic study or a proteomic experiment). | 16 |
| 17 | Use the KEGG pathway database to look up interaction pathways. | 17 |
| 18 | Use graph theory to represent biological data networks. | 17, 18 |
| 19 | Understand the challenges in integrating and interpreting large heterogenous high throughput data sets into their functional context. | 19 |
| 20 | Have an appreciation for the social impacts and ethical implications of how genomic sequence information is used in our society | 20 |

## These support a major learning objective

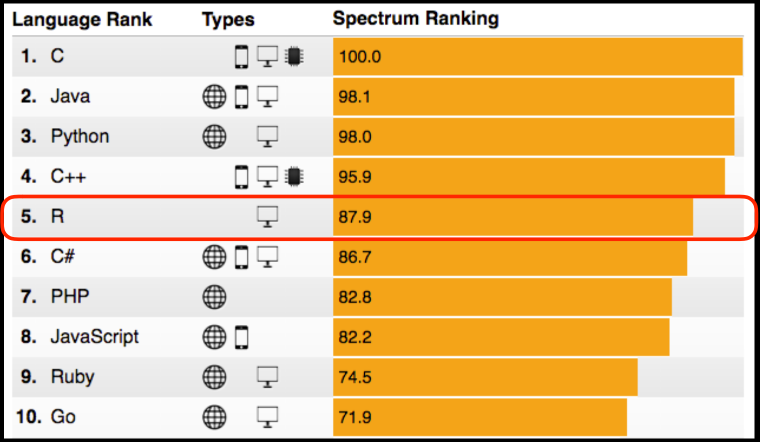
**At the end of this course students will:**

- Understand the increasing necessity for computation in modern life sciences research.
- Be able to use and evaluate online bioinformatics resources and analysis tools to solve problems in the biological sciences.
- Be able to use the R environment to analyze bioinformatics data at scale.
- Be familiar with the research objectives of the bioinformatics related sub-disciplines of Genome informatics, Transcriptomics and Structural informatics.

# Why use R?

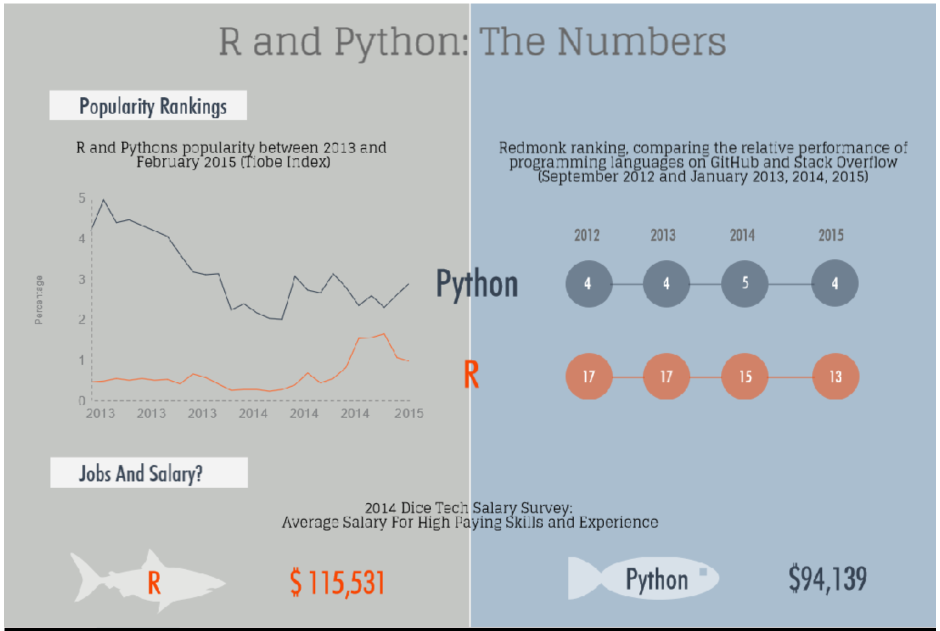
Productivity
Flexibility
Designed for data analysis

## IEEE 2016 Top Programming Languages



| Language Rank | Types | Spectrum Ranking |
|---|---|---|
| 1. C | 📱 🖥 ▦ | 100.0 |
| 2. Java | 🌐 📱 🖥 | 98.1 |
| 3. Python | 🌐 🖥 | 98.0 |
| 4. C++ | 📱 🖥 ▦ | 95.9 |
| 5. R | 🖥 | 87.9 |
| 6. C# | 🌐 📱 🖥 | 86.7 |
| 7. PHP | 🌐 | 82.8 |
| 8. JavaScript | 🌐 📱 | 82.2 |
| 9. Ruby | 🌐 🖥 | 74.5 |
| 10. Go | 🌐 🖥 | 71.9 |

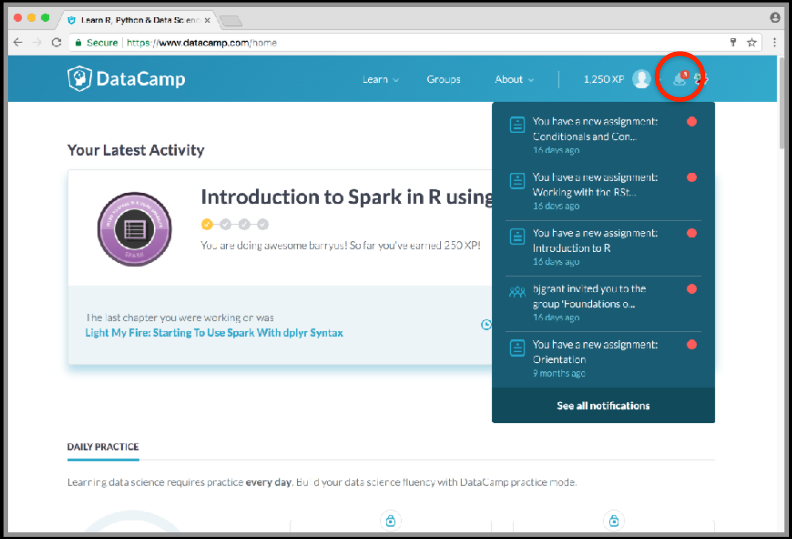
http://spectrum.ieee.org/computing/software/the-2016-top-programming-languages



http://www.kdnuggets.com/2015/05/r-vs-python-data-science.html?utm_medium=email&utm_source=flipboard

- R is the “lingua franca” of data science in industry and academia.
- Large user and developer community.
  - As of Jan 8th 2018 there are 12,039 add on **R packages** on **CRAN** and 1,473 on **Bioconductor** - more on these later!
- Virtually every statistical technique is either already built into R, or available as a free package.
- Unparalleled exploratory data analysis environment.

< https://www.datacamp.com/ >



Your Latest Activity

Introduction to Spark in R using

You are doing awesome barryus! So far you've earned 250 XP!

The last chapter you were working on was
Light My Fire: Starting To Use Spark With dplyr Syntax

You have a new assignment: Conditionals and Con...
16 days ago

You have a new assignment: Working with the RSt...
16 days ago

You have a new assignment: Introduction to R
16 days ago

bjgrant invited you to the group 'Foundations o...
16 days ago

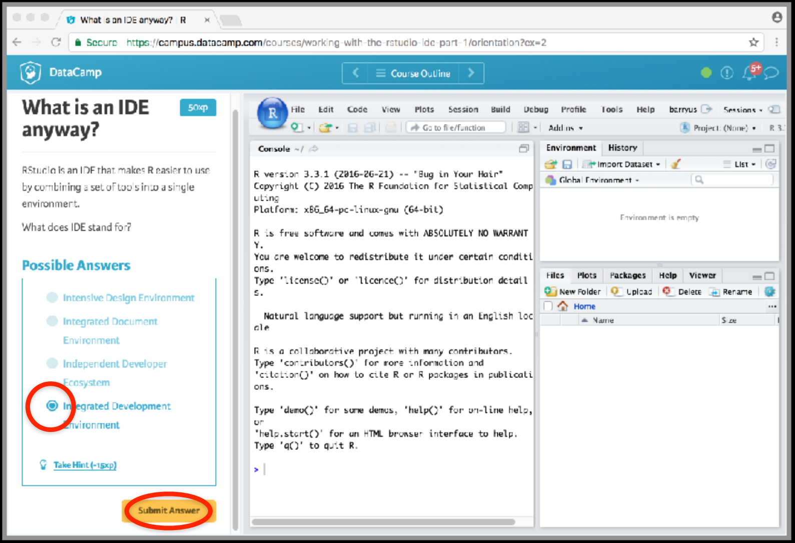
You have a new assignment: Orientation
9 months ago

See all notifications

DAILY PRACTICE

Learning data science requires practice **every day**. Build your data science fluency with DataCamp practice mode.

< https://www.datacamp.com/ >



https://campus.datacamp.com/courses/working-with-the-rstudio-ide-part-1/orientation?ex=2

**What is an IDE anyway?** 50xp

RStudio is an IDE that makes R easier to use by combining a set of tools into a single environment.

What does IDE stand for?

**Possible Answers**

- ○ Intensive Design Environment
- ○ Integrated Document Environment
- ○ Independent Developer Ecosystem
- ● Integrated Development Environment

Take Hint (-15xp)

Submit Answer

```
R version 3.3.1 (2016-06-21) -- "Bug in Your Hair"
Copyright (C) 2016 The R Foundation for Statistical Comp
uting
Platform: x86_64-pc-linux-gnu (64-bit)

R is free software and comes with ABSOLUTELY NO WARRANT
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You are welcome to redistribute it under certain conditi
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Type 'license()' or 'licence()' for distribution detail
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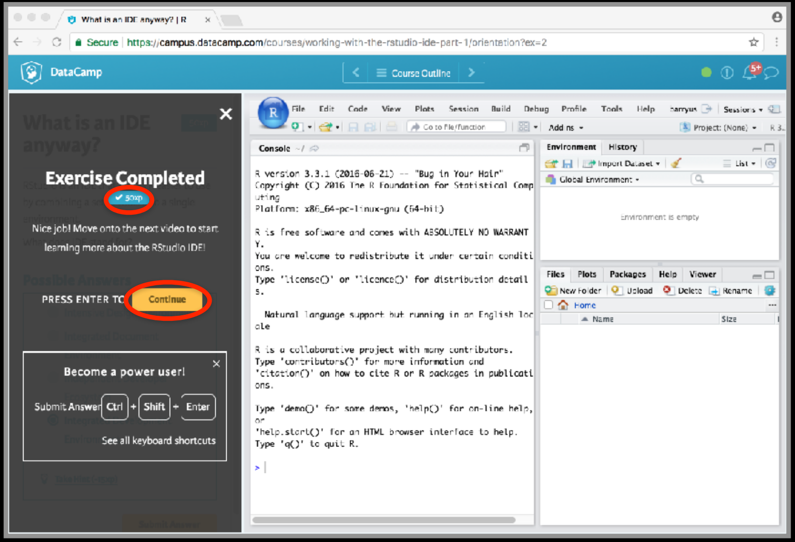
R is a collaborative project with many contributors.
Type 'contributors()' for more information and
'citation()' on how to cite R or R packages in publicati
ons.

Type 'demo()' for some demos, 'help()' for on-line help,
or
'help.start()' for an HTML browser interface to help.
Type 'q()' to quit R.

> 
```

< https://www.datacamp.com/ >



https://campus.datacamp.com/courses/working-with-the-rstudio-ide-part-1/orientation?ex=2

**Exercise Completed**

✔ 50xp

Nice job! Move onto the next video to start learning more about the RStudio IDE!

PRESS ENTER TO Continue

**Become a power user!**

Submit Answer Ctrl + Shift + Enter

See all keyboard shortcuts

```
R version 3.3.1 (2016-06-21) -- "Bug in Your Hair"
Copyright (C) 2016 The R Foundation for Statistical Comp
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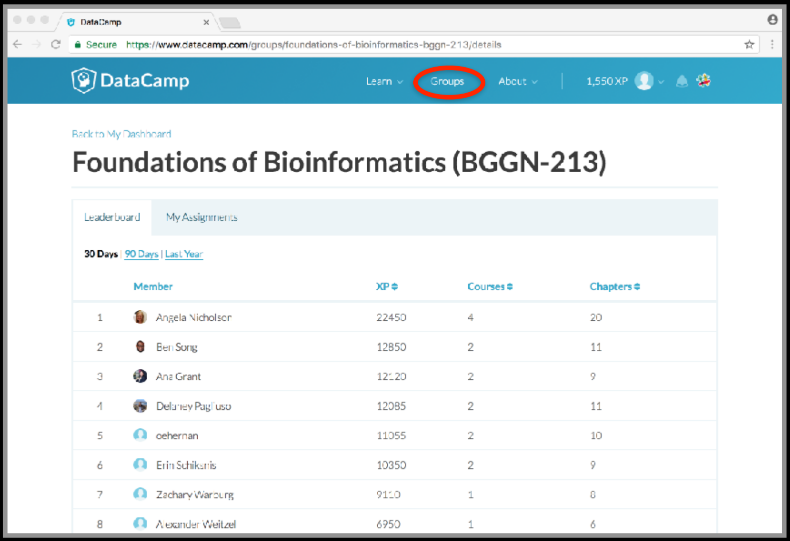

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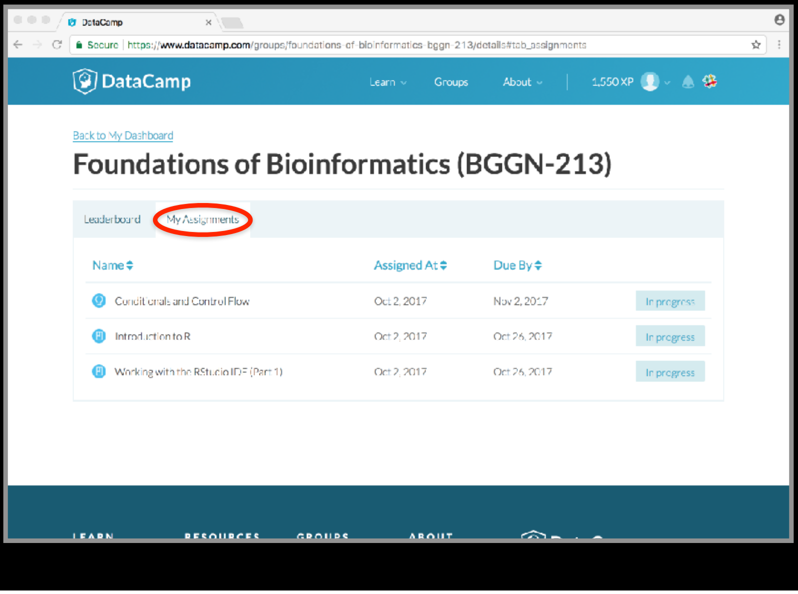
**Foundations of Bioinformatics (BGGN-213)**

Leaderboard | My Assignments

**30 Days** | 90 Days | Last Year

|  | Member | XP ⬍ | Courses ⬍ | Chapters ⬍ |
|---|---|---|---|---|
| 1 | Angela Nicholson | 22450 | 4 | 20 |
| 2 | Ben Song | 12850 | 2 | 11 |
| 3 | Ana Grant | 12120 | 2 | 9 |
| 4 | Delaney Pagliuso | 12085 | 2 | 11 |
| 5 | oehernan | 11055 | 2 | 10 |
| 6 | Erin Schiksnis | 10350 | 2 | 9 |
| 7 | Zachary Warburg | 9110 | 1 | 8 |
| 8 | Alexander Weitzel | 6950 | 1 | 6 |

< https://www.datacamp.com/ >



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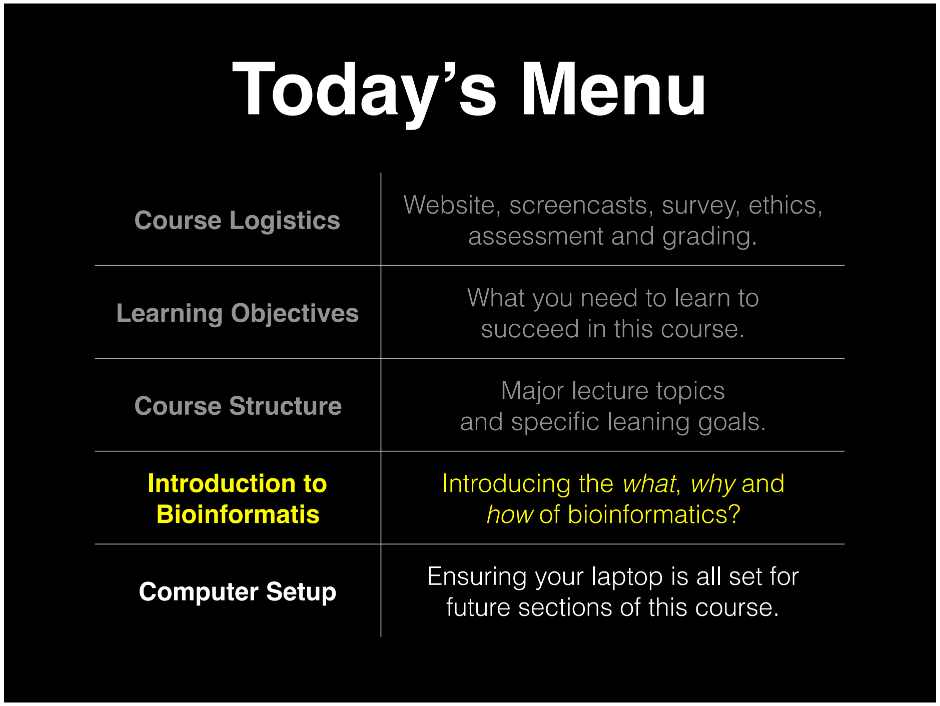
Back to My Dashboard

**Foundations of Bioinformatics (BGGN-213)**

Leaderboard | My Assignments

| Name ⬍ | Assigned At ⬍ | Due By ⬍ |  |
|---|---|---|---|
| Conditionals and Control Flow | Oct 2, 2017 | Nov 2, 2017 | In progress |
| Introduction to R | Oct 2, 2017 | Oct 26, 2017 | In progress |
| Working with the RStudio IDE (Part 1) | Oct 2, 2017 | Oct 26, 2017 | In progress |



# Today’s Menu

| | |
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| **Course Logistics** | Website, screencasts, survey, ethics, assessment and grading. |
| **Learning Objectives** | What you need to learn to succeed in this course. |
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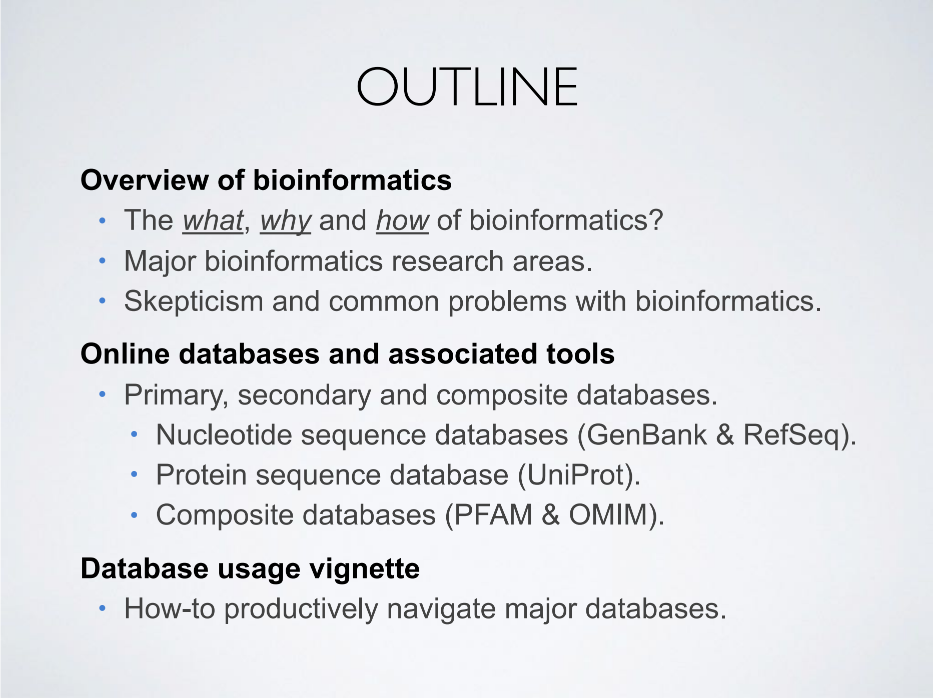
| **Computer Setup** | Ensuring your laptop is all set for future sections of this course. |



# OUTLINE

**Overview of bioinformatics**

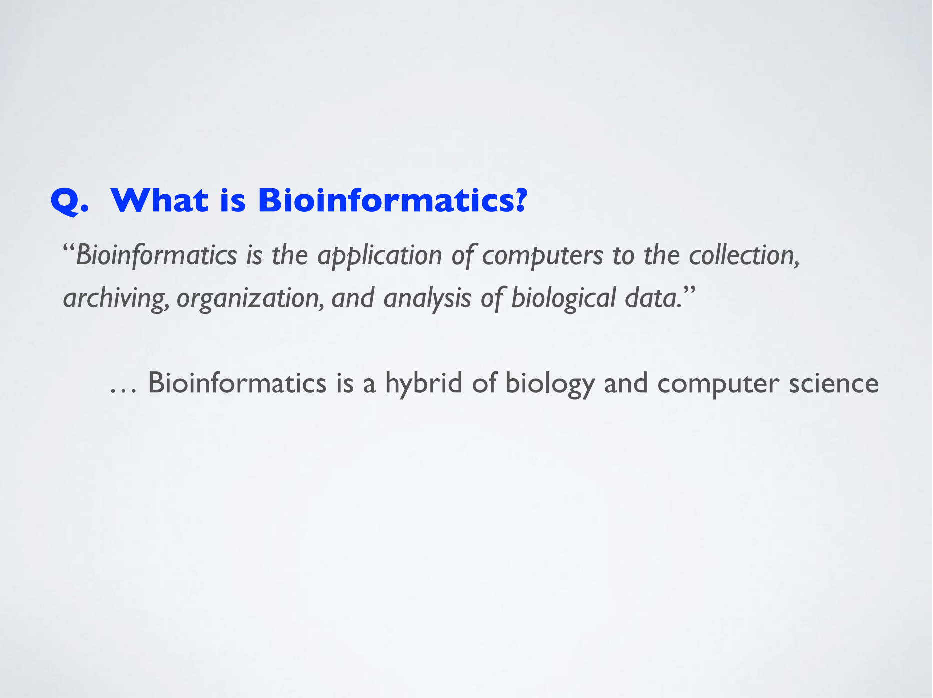
- The *what*, *why* and *how* of bioinformatics?
- Major bioinformatics research areas.
- Skepticism and common problems with bioinformatics.

**Online databases and associated tools**

- Primary, secondary and composite databases.
  - Nucleotide sequence databases (GenBank & RefSeq).
  - Protein sequence database (UniProt).
  - Composite databases (PFAM & OMIM).

**Database usage vignette**

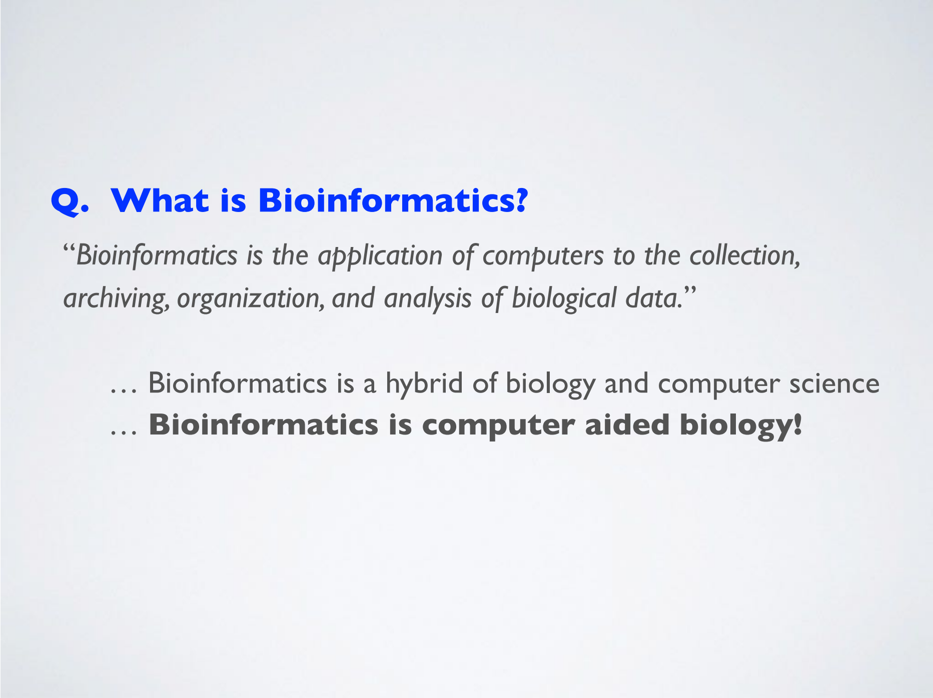
- How-to productively navigate major databases.



## Q. What is Bioinformatics?

*“Bioinformatics is the application of computers to the collection, archiving, organization, and analysis of biological data.”*

… Bioinformatics is a hybrid of biology and computer science



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… **Bioinformatics is computer aided biology!**

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… Bioinformatics is a hybrid of biology and computer science

… **Bioinformatics is computer aided biology!**

Computer based management and analysis of biological and biomedical data with useful applications in many disciplines, particularly genomics, proteomics, metabolomics, etc...

# MORE DEFINITIONS

- "Bioinformatics is conceptualizing biology in terms of **macromolecules** and then applying **"informatics" techniques** (derived from disciplines such as applied maths, computer science, and statistics) to **understand** and **organize** the information associated with these molecules, on a **large-scale**.
  Luscombe NM, *et al.* Methods Inf Med. 2001;40:346.

- "Bioinformatics is research, development, or application of **computational approaches** for expanding the use of **biological**, **medical**, **behavioral** or **health data**, including those to **acquire**, **store**, **organize** and **analyze** such data."
  National Institutes of Health (NIH) ( http://tinyurl.com/l3gxr6b )

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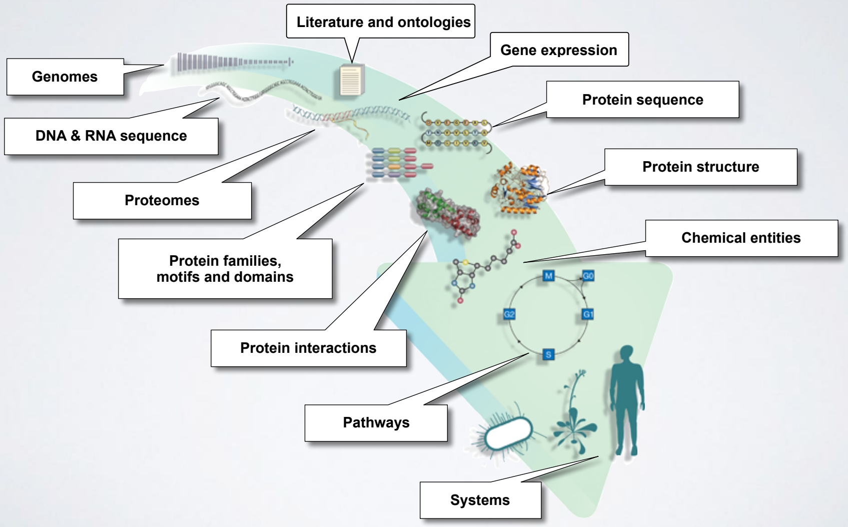
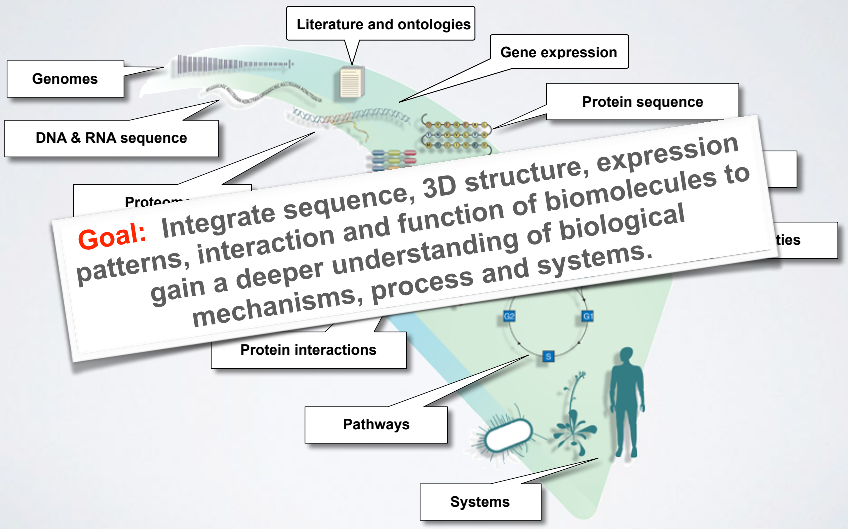
**Key Point:** Bioinformatics is Computer Aided Biology

# Major types of Bioinformatics Data

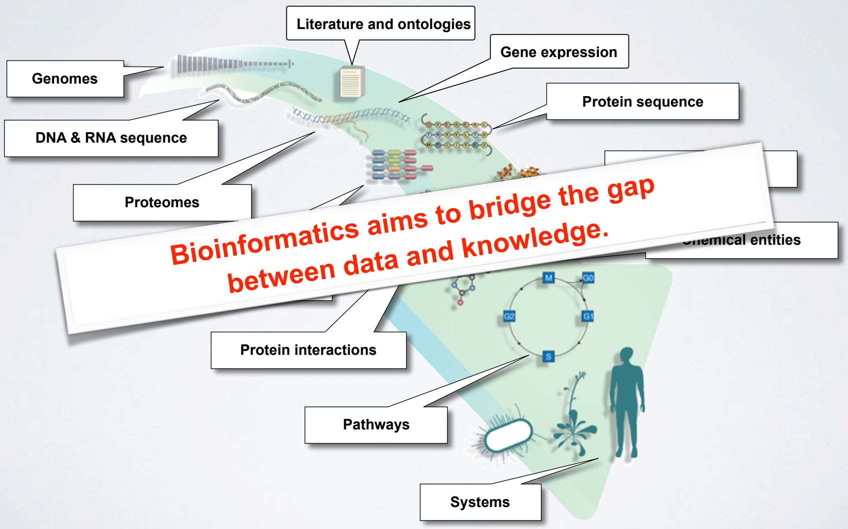


# Major types of Bioinformatics Data



**Goal:** Integrate sequence, 3D structure, expression patterns, interaction and function of biomolecules to gain a deeper understanding of biological mechanisms, process and systems.

# Major types of Bioinformatics Data



**Bioinformatics aims to bridge the gap between data and knowledge.**

# BIOINFORMATICS RESEARCH AREAS

Include but are not limited to:

- Organization, classification, dissemination and analysis of biological and biomedical data (particularly '-omics' data).
- Biological sequence analysis and phylogenetics.
- Genome organization and evolution.
- Regulation of gene expression and epigenetics.
- Biological pathways and networks in healthy & disease states.
- Protein structure prediction from sequence.
- Modeling and prediction of the biophysical properties of biomolecules for binding prediction and drug design.
- Design of biomolecular structure and function.

With applications to Biology, Medicine, Agriculture and Industry

# Where did bioinformatics come from?

Bioinformatics arose as molecular biology began to be transformed by the emergence of molecular sequence and structural data

**Recap: The key dogmas of molecular biology**

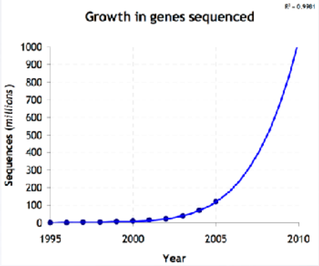
- *DNA sequence* determines *protein sequence.*
- *Protein sequence* determines *protein structure.*
- *Protein structure* determines *protein function.*
- *Regulatory mechanisms* (e.g. gene expression) determine the amount of a particular *function in space and time.*

Bioinformatics is *now* essential for the archiving, organization and analysis of data related to all these processes.

# Why do we need Bioinformatics?

Bioinformatics is necessitated by the rapidly expanding quantities and complexity of biomolecular data

- Bioinformatics provides methods for the efficient:
  - **storage**
  - **annotation**
  - **search** and **retrieval**
  - data **integration**
  - data **mining** and **analysis**



E.G. data from sequencing, structural genomics, proteomics, new high throughput assays, *etc…*

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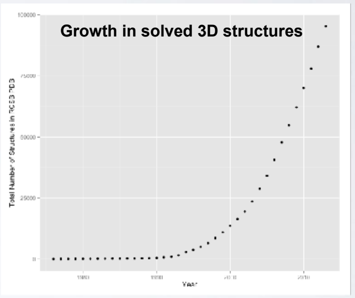
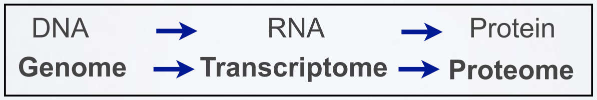


E.G. data from sequencing, structural genomics, proteomics, new high throughput assays, *etc…*

# How do we do Bioinformatics?

- A "*bioinformatics approach"* involves the application of **computer algorithms**, **computer models** and **computer databases** with the broad goal of understanding the action of both individual genes, transcripts, proteins and large collections of these entities.



x 1,000

x 100,000

# How do we *actually* do Bioinformatics?

## Pre-packaged tools and databases

- Many online
- Most are free to use
- Time consuming methods require downloading…

## Advanced tool application & development

- Mostly on a UNIX environment
- Knowledge of programing languages frequently required (*e.g.* **R**, Python, Perl, C, Java, Fortran)
- May require specialized high performance computing…

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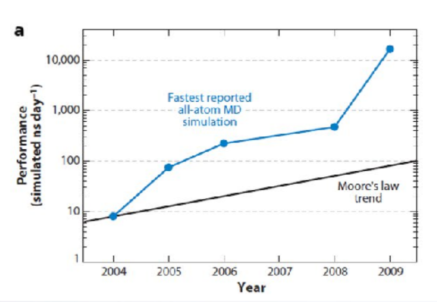
## SIDE-NOTE: SUPERCOMPUTERS AND GPUS



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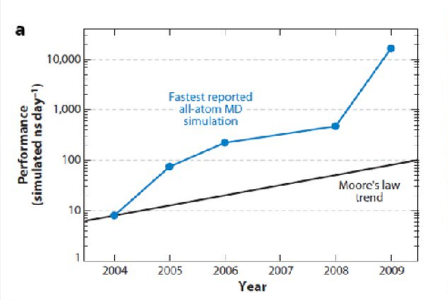
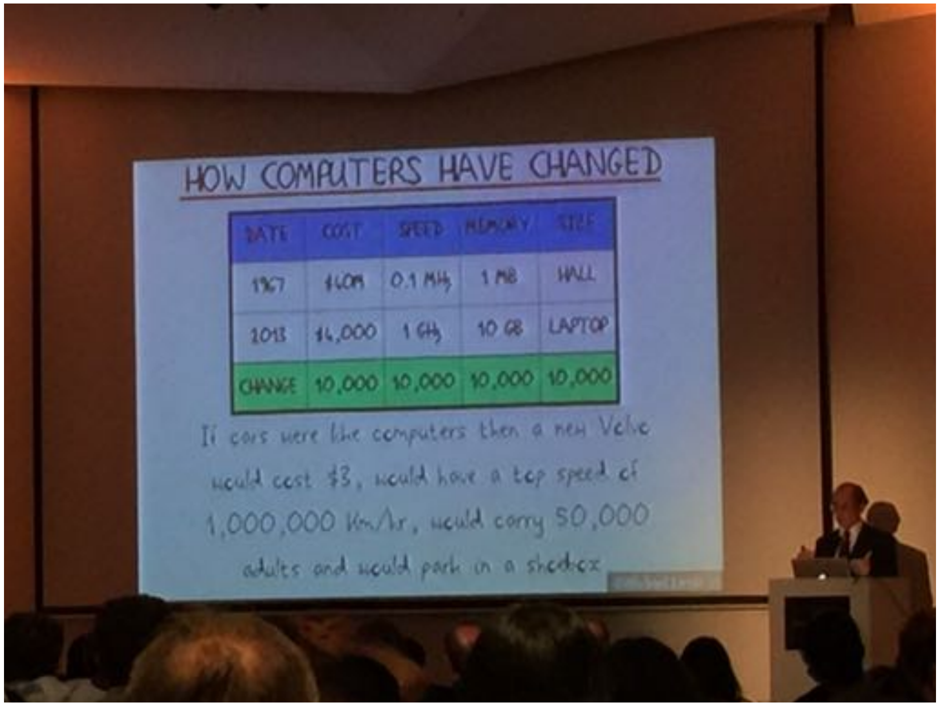


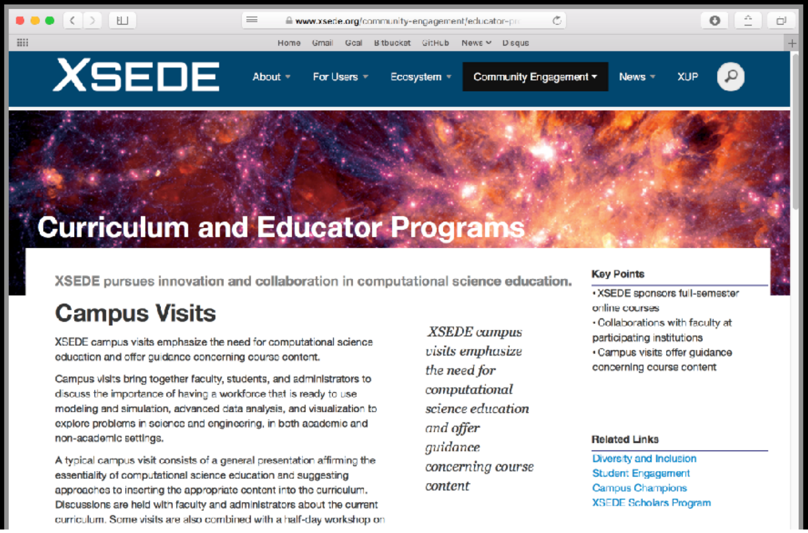


HOW COMPUTERS HAVE CHANGED

| DATE | COST | SPEED | MEMORY | SIZE |
|---|---|---|---|---|
| 1967 | $40M | 0.1 MHz | 1 MB | HALL |
| 2013 | $4,000 | 1 GHz | 10 GB | LAPTOP |
| CHANGE | 10,000 | 10,000 | 10,000 | 10,000 |

If cars were like the computers then a new Volvo would cost $3, would have a top speed of 1,000,000 Km/hr, would carry 50,000 adults and would park in a shoebox

# NSF Extreme Science and Engineering Discovery Environment (XSEDE)



## Curriculum and Educator Programs

XSEDE pursues innovation and collaboration in computational science education.

### Campus Visits

XSEDE campus visits emphasize the need for computational science education and offer guidance concerning course content.

Campus visits bring together faculty, students, and administrators to discuss the importance of having a workforce that is ready to use modeling and simulation, advanced data analysis, and visualization to explore problems in science and engineering, in both academic and non-academic settings.

A typical campus visit consists of a general presentation affirming the essentiality of computational science education and suggesting approaches to inserting the appropriate content into the curriculum. Discussions are held with faculty and administrators about the current curriculum. Some visits are also combined with a half-day workshop on

*XSEDE campus visits emphasize the need for computational science education and offer guidance concerning course content*

**Key Points**

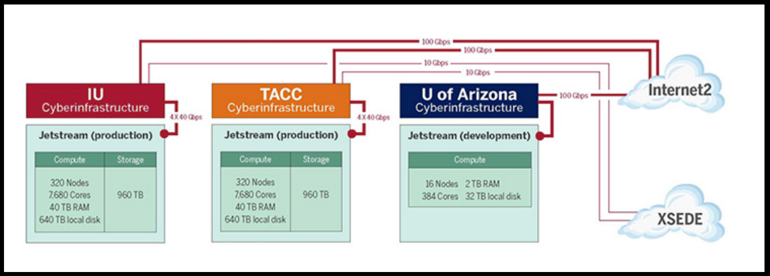
- XSEDE sponsors full-semester online courses
- Collaborations with faculty at participating institutions
- Campus visits offer guidance concerning course content

**Related Links**

Diversity and Inclusion
Student Engagement
Campus Champions
XSEDE Scholars Program

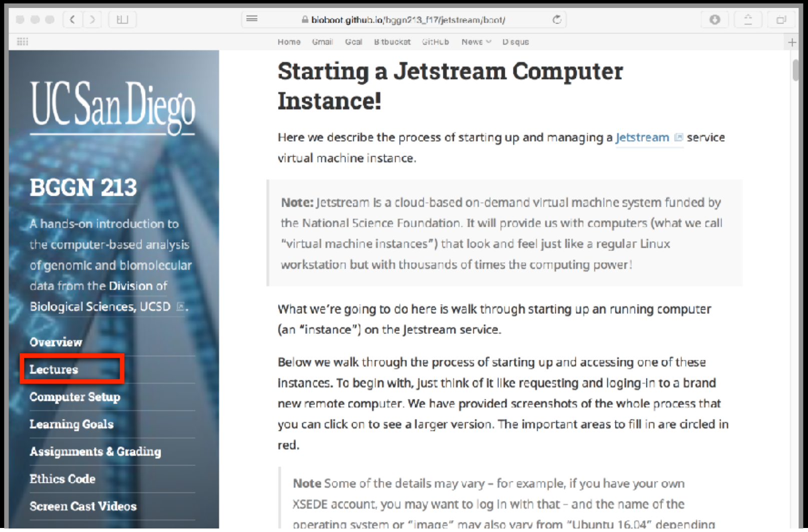
# What is *Jetstream*?

- A new cloud computing environment based at Indiana University and the Texas Advanced Computing Center (TACC) providing on-demand access to interactive computing and data analysis resources.



# Jetstream tutorials

Developed *user friendly* labs for Jetstream basics



**BGGN 213**

A hands-on introduction to the computer-based analysis of genomic and biomolecular data from the Division of Biological Sciences, UCSD.

## Starting a Jetstream Computer Instance!

Here we describe the process of starting up and managing a Jetstream service virtual machine instance.

> **Note:** Jetstream is a cloud-based on-demand virtual machine system funded by the National Science Foundation. It will provide us with computers (what we call "virtual machine instances") that look and feel just like a regular Linux workstation but with thousands of times the computing power!

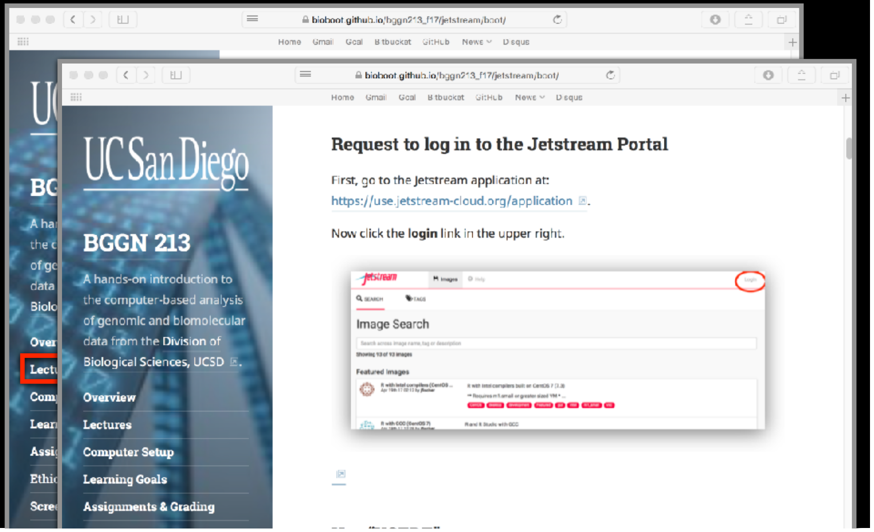
What we're going to do here is walk through starting up an running computer (an "instance") on the Jetstream service.

Below we walk through the process of starting up and accessing one of these instances. To begin with, just think of it like requesting and loging-in to a brand new remote computer. We have provided screenshots of the whole process that you can click on to see a larger version. The important areas to fill in are circled in red.

> **Note** Some of the details may vary - for example, if you have your own XSEDE account, you may want to log in with that - and the name of the operating system or "image" may also vary from "Ubuntu 16.04" depending

# Jetstream tutorials

Developed *user friendly* labs for Jetstream basics



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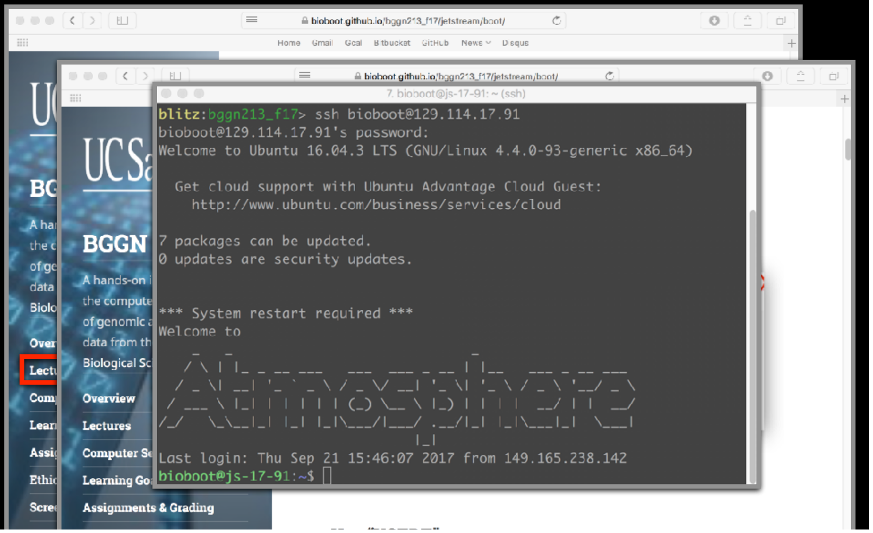
## Request to log in to the Jetstream Portal

First, go to the Jetstream application at: https://use.jetstream-cloud.org/application.

Now click the **login** link in the upper right.

## Jetstream tutorials

Developed *user friendly* labs for Jetstream basics



```
blitz:bggn213_f17> ssh bioboot@129.114.17.91
bioboot@129.114.17.91's password:
Welcome to Ubuntu 16.04.3 LTS (GNU/Linux 4.4.0-93-generic x86_64)

  Get cloud support with Ubuntu Advantage Cloud Guest:
    http://www.ubuntu.com/business/services/cloud

7 packages can be updated.
0 updates are security updates.


*** System restart required ***
Welcome to
    _   _                             _
   /_\ | |_ _ __ ___   ___  ___ _ __ | |__   ___ _ __ ___
  / _ \| __| '_ ` _ \ / _ \/ __| '_ \| '_ \ / _ \ '__/ _ \
 / ___ \ |_| | | | | | (_) \__ \ |_) | | | |  __/ | |  __/
/_/   \_\__|_| |_| |_|\___/|___/ .__/|_| |_|\___|_|  \___|
                               |_|
Last login: Thu Sep 21 15:46:07 2017 from 149.165.238.142
bioboot@js-17-91:~$
```

## Skepticism & Bioinformatics

We have to approach computational results the same way we do wet-lab results:

- Do they make sense?
- Is it what we expected?
- Do we have adequate controls, and how did they come out?
- Modeling is modeling, but biology is different...

  *What does this model actually contribute?*
- Avoid the miss-use of ‘black boxes’

## Skepticism & Bioinformatics

Gunnar von Heijne in his old but quite readable treatise, *Sequence Analysis in Molecular Biology; Treasure Trove or Trivial Pursuit*, provides a very appropriate conclusion:

- “Think about what you’re doing; use your knowledge of the molecular system involved to guide both your interpretation of results and your direction of inquiry; use as much information as possible; and do not blindly accept everything the computer offers you”.
- Key-Point: **Avoid the miss-use of ‘black boxes’!**

## Common problems with Bioinformatics

Confusing multitude of tools available

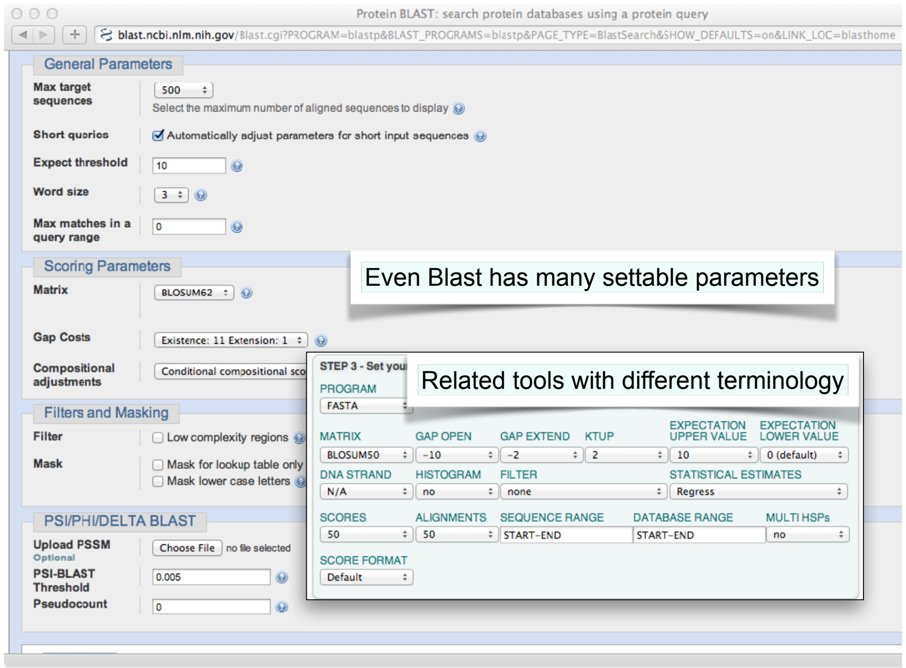
- Each with many options and settable parameters

Most tools and databases are written by and for nerds

- Same is true of documentation - if any exists!

Most are developed independently

Notable exceptions are found at the:
- **EBI** (European Bioinformatics Institute) and
- **NCBI** (National Center for Biotechnology Information)



Protein BLAST: search protein databases using a protein query

blast.ncbi.nlm.nih.gov/Blast.cgi?PROGRAM=blastp&BLAST_PROGRAMS=blastp&PAGE_TYPE=BlastSearch&SHOW_DEFAULTS=on&LINK_LOC=blasthome

**General Parameters**

- Max target sequences: 500 — Select the maximum number of aligned sequences to display
- Short queries: ☑ Automatically adjust parameters for short input sequences
- Expect threshold: 10
- Word size: 3
- Max matches in a query range: 0

**Scoring Parameters**

- Matrix: BLOSUM62
- Gap Costs: Existence: 11 Extension: 1
- Compositional adjustments: Conditional compositional sco

**Filters and Masking**

- Filter: □ Low complexity regions
- Mask: □ Mask for lookup table only
  □ Mask lower case letters

**PSI/PHI/DELTA BLAST**

- Upload PSSM (Optional): Choose File no file selected
- PSI-BLAST Threshold: 0.005
- Pseudocount: 0

Even Blast has many settable parameters

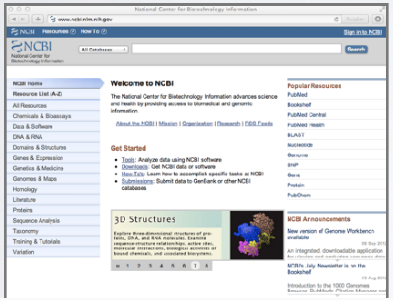
STEP 3 - Set you

| PROGRAM | | | | | |
|---|---|---|---|---|---|
| FASTA | | | | | |
| MATRIX | GAP OPEN | GAP EXTEND | KTUP | EXPECTATION UPPER VALUE | EXPECTATION LOWER VALUE |
| BLOSUM50 | -10 | -2 | 2 | 10 | 0 (default) |
| DNA STRAND | HISTOGRAM | FILTER | | STATISTICAL ESTIMATES | |
| N/A | no | none | | Regress | |
| SCORES | ALIGNMENTS | SEQUENCE RANGE | DATABASE RANGE | MULTI HSPs | |
| 50 | 50 | START-END | START-END | no | |
| SCORE FORMAT | | | | | |
| Default | | | | | |

Related tools with different terminology

# Key Online Bioinformatics Resources: NCBI & EBI

The NCBI and EBI are invaluable, publicly available resources for biomedical research



http://www.ncbi.nlm.nih.gov

The European Bioinformatics Institute

https://www.ebi.ac.uk

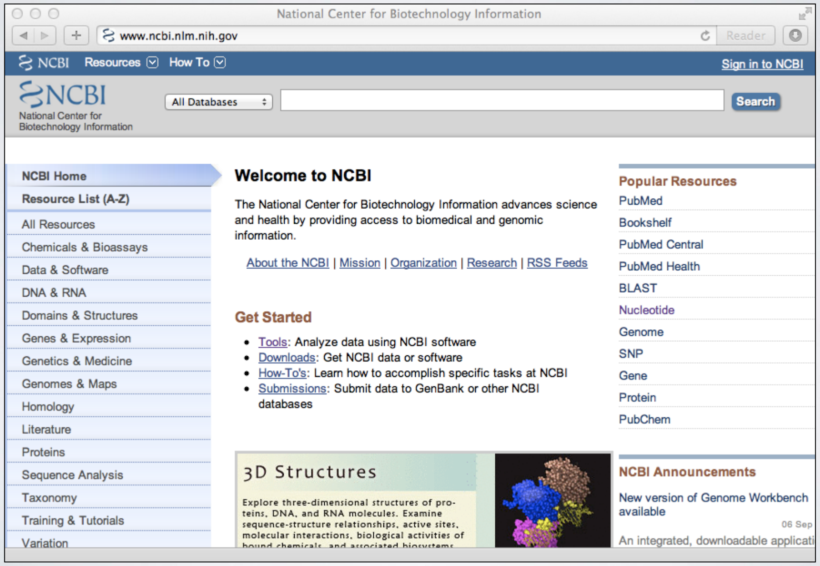
# National Center for Biotechnology Information (NCBI)

- Created in 1988 as a part of the National Library of Medicine (NLM) at the National Institutes of Health
- NCBI's mission includes:
  - Establish **public databases**
  - Develop **software tools**
  - **Education** on and dissemination of biomedical information

Bethesda,MD

- We will cover a number of core NCBI databases and software tools in the lecture

## http://www.ncbi.nlm.nih.gov



National Center for Biotechnology Information

www.ncbi.nlm.nih.gov

- NCBI Home
- Resource List (A-Z)
- All Resources
- Chemicals & Bioassays
- Data & Software
- DNA & RNA
- Domains & Structures
- Genes & Expression
- Genetics & Medicine
- Genomes & Maps
- Homology
- Literature
- Proteins
- Sequence Analysis
- Taxonomy
- Training & Tutorials
- Variation

**Welcome to NCBI**

The National Center for Biotechnology Information advances science and health by providing access to biomedical and genomic information.

About the NCBI | Mission | Organization | Research | RSS Feeds

**Get Started**

- Tools: Analyze data using NCBI software
- Downloads: Get NCBI data or software
- How-To's: Learn how to accomplish specific tasks at NCBI
- Submissions: Submit data to GenBank or other NCBI databases

3D Structures

Explore three-dimensional structures of proteins, DNA, and RNA molecules. Examine sequence-structure relationships, active sites, molecular interactions, biological activities of bound chemicals, and associated biosystems

**Popular Resources**

- PubMed
- Bookshelf
- PubMed Central
- PubMed Health
- BLAST
- Nucleotide
- Genome
- SNP
- Gene
- Protein
- PubChem

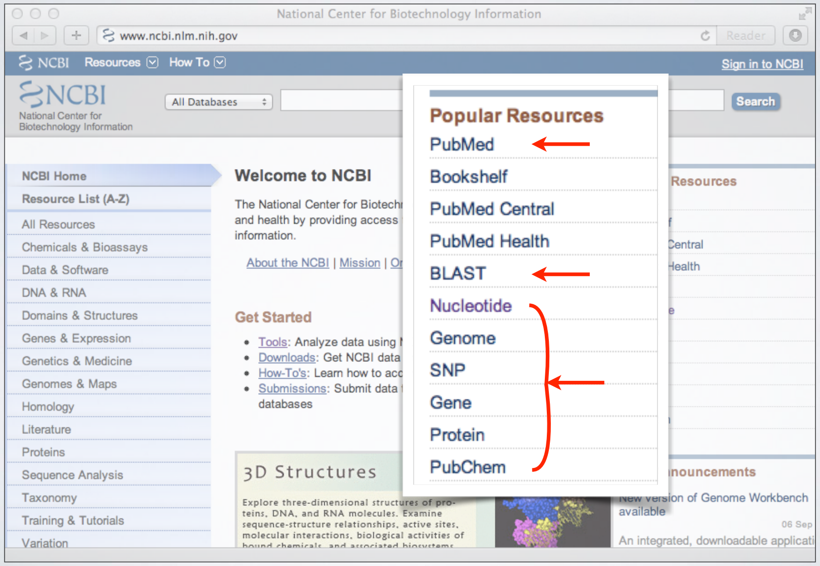
**NCBI Announcements**

New version of Genome Workbench available

06 Sep

An integrated, downloadable applicati

## http://www.ncbi.nlm.nih.gov



Welcome to NCBI

The National Center for Biotechn and health by providing access information.

About the NCBI | Mission | Or

Get Started

- Tools: Analyze data using
- Downloads: Get NCBI data
- How-To's: Learn how to acc
- Submissions: Submit data databases

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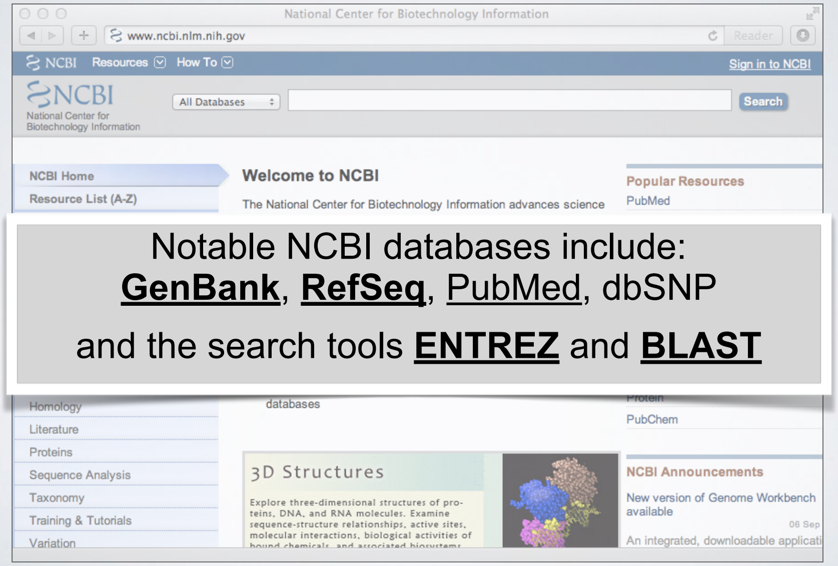

Welcome to NCBI

The National Center for Biotechnology Information advances science

databases

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06 Sep

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Notable NCBI databases include:
**GenBank**, **RefSeq**, PubMed, dbSNP

and the search tools **ENTREZ** and **BLAST**

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http://www.ncbi.nlm.nih.gov

The European Bioinformatics Institute
Find a gene, protein or chemical:
Visit EMBL.org

https://www.ebi.ac.uk

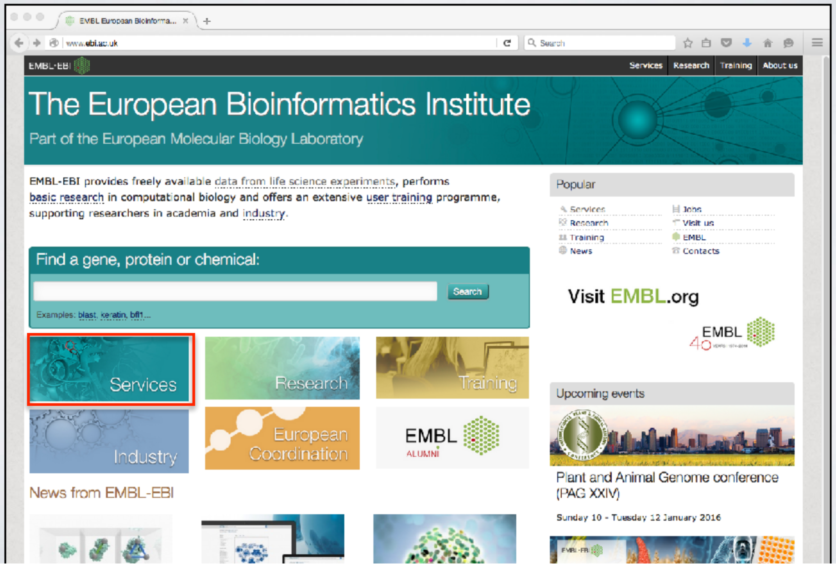
## European Bioinformatics Institute (EBI)

- Created in 1997 as a part of the European Molecular Biology Laboratory (EMBL)
- EBI's mission includes:
  - providing freely available **data and bioinformatics services**
  - and providing advanced **bioinformatics training**

United Kingdom
Ireland
France
Hinxton,UK

- We will briefly cover several EBI databases and tools that have advantages over those offered at NCBI

## The EBI maintains a number of high quality curated **secondary databases** and associated tools



The European Bioinformatics Institute

Part of the European Molecular Biology Laboratory

EMBL-EBI provides freely available data from life science experiments, performs basic research in computational biology and offers an extensive user training programme, supporting researchers in academia and industry.

Find a gene, protein or chemical:

Examples: blast, keratin, bfl1...

News from EMBL-EBI

Upcoming events

Plant and Animal Genome conference (PAG XXIV)

Sunday 10 - Tuesday 12 January 2016

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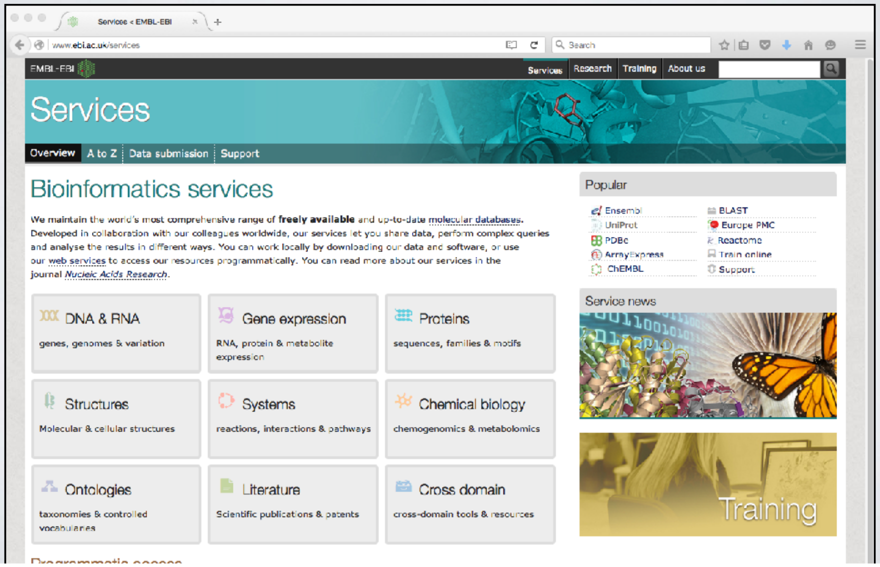

Bioinformatics services

We maintain the world's most comprehensive range of **freely available** and up-to-date molecular databases. Developed in collaboration with our colleagues worldwide, our services let you share data, perform complex queries and analyse the results in different ways. You can work locally by downloading our data and software, or use our web services to access our resources programmatically. You can read more about our services in the journal *Nucleic Acids Research*.

| | | |
|---|---|---|
| DNA & RNA<br>genes, genomes & variation | Gene expression<br>RNA, protein & metabolite expression | Proteins<br>sequences, families & motifs |
| Structures<br>Molecular & cellular structures | Systems<br>reactions, interactions & pathways | Chemical biology<br>chemogenomics & metabolomics |
| Ontologies<br>taxonomies & controlled vocabularies | Literature<br>Scientific publications & patents | Cross domain<br>cross-domain tools & resources |

Programmatic access

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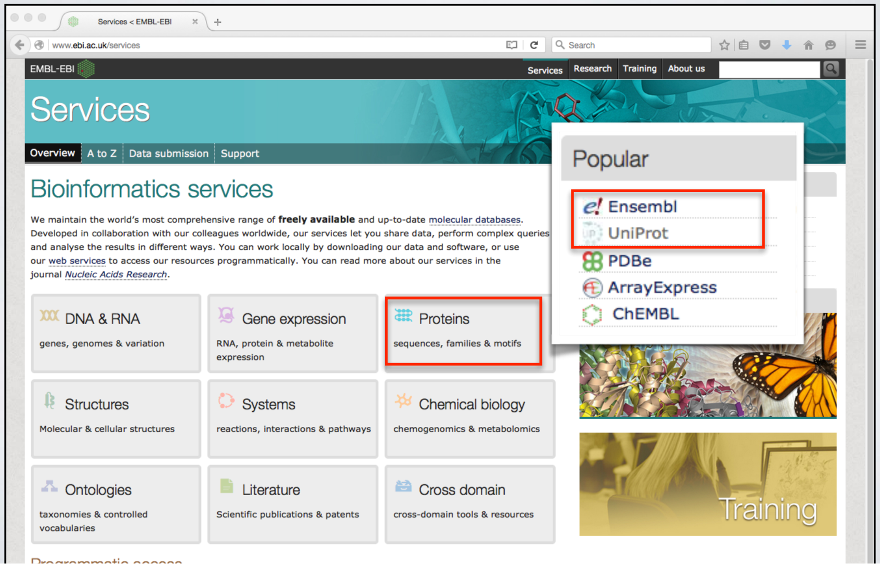

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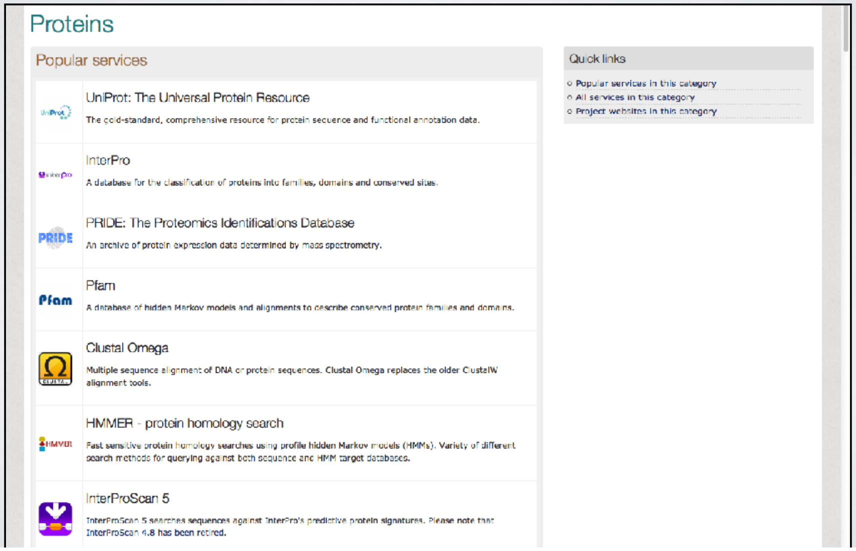
| | | |
|---|---|---|
| DNA & RNA<br>genes, genomes & variation | Gene expression<br>RNA, protein & metabolite expression | Proteins<br>sequences, families & motifs |
| Structures<br>Molecular & cellular structures | Systems<br>reactions, interactions & pathways | Chemical biology<br>chemogenomics & metabolomics |
| Ontologies<br>taxonomies & controlled vocabularies | Literature<br>Scientific publications & patents | Cross domain<br>cross-domain tools & resources |

Programmatic access

## https://www.ebi.ac.uk

The EBI makes available a wider variety of **online tools** than NCBI



Proteins

Popular services

UniProt: The Universal Protein Resource

The gold-standard, comprehensive resource for protein sequence and functional annotation data.

InterPro

A database for the classification of proteins into families, domains and conserved sites.

PRIDE: The Proteomics Identifications Database

An archive of protein expression data determined by mass spectrometry.

Pfam

A database of hidden Markov models and alignments to describe conserved protein families and domains.

Clustal Omega

Multiple sequence alignment of DNA or protein sequences. Clustal Omega replaces the older ClustalW alignment tools.

HMMER - protein homology search

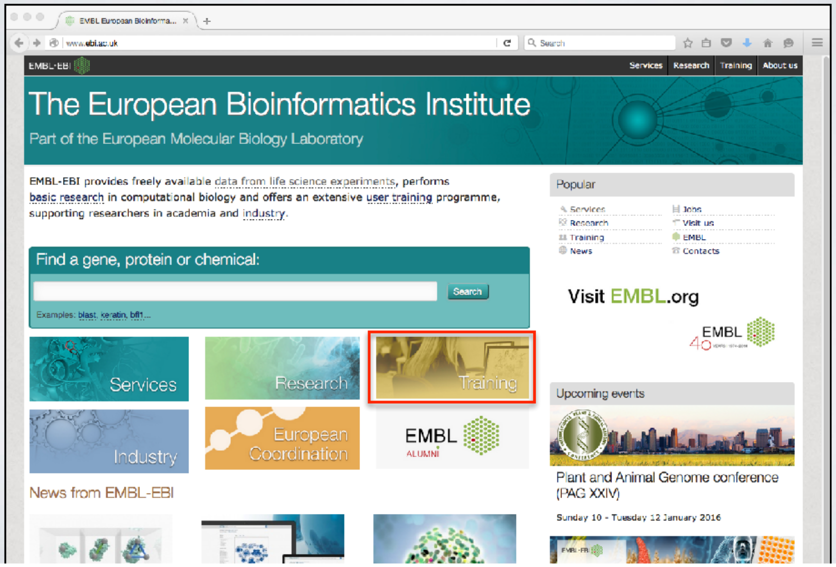
Fast sensitive protein homology searches using profile hidden Markov models (HMMs). Variety of different search methods for querying against both sequence and HMM target databases.

InterProScan 5

InterProScan 5 searches sequences against InterPro's predictive protein signatures. Please note that InterProScan 4.8 has been retired.

Quick links
Popular services in this category
All services in this category
Project websites in this category

The EBI also provides a growing selection of **online tutorials** on EBI databases and tools



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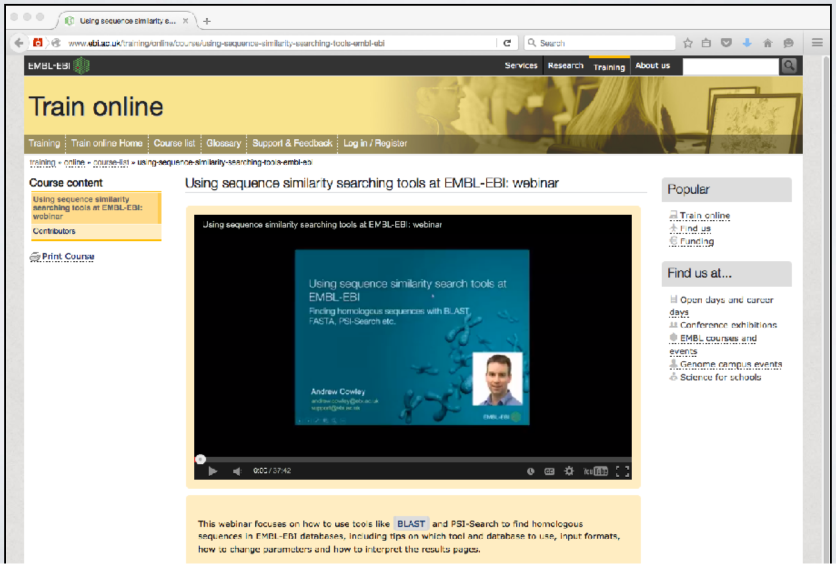


Train online

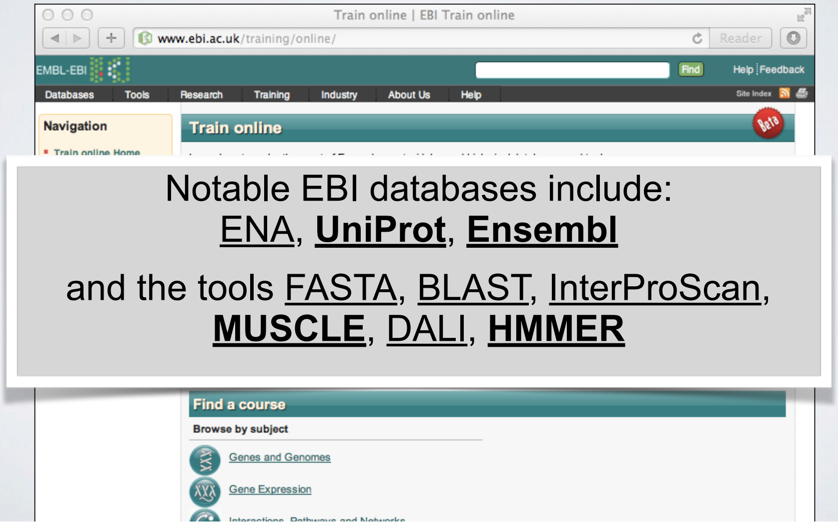
Using sequence similarity searching tools at EMBL-EBI: webinar

Using sequence similarity searching tools at EMBL-EBI

Finding homologous sequences with BLAST, FASTA, PSI-Search etc.

This webinar focuses on how to use tools like BLAST and PSI-Search to find homologous sequences in EMBL-EBI databases, including tips on which tool and database to use, input formats, how to change parameters and how to interpret the results pages.

The EBI also provides a growing selection of **online tutorials** on EBI databases and tools



Notable EBI databases include:
ENA, **UniProt**, **Ensembl**

and the tools FASTA, BLAST, InterProScan, **MUSCLE**, DALI, **HMMER**

## Next Class…

## MAJOR BIOINFORMATICS DATABASES AND ASSOCIATED ONLINE TOOLS

# Bioinformatics Databases

AATDB, AceDb, ACUTS, ADB, AFDB, AGIS, AMSdb, ARR, AsDb,BBDB, BCGD, Beanref, BioImage, BioMagResBank, BIOMDB, BLOCKS, BovGBASE, BOVMAP, BSORF, BTKbase, CANSITE, CarbBank, CARBHYD, CATH, CAZY, CCDC, CD4OLbase, CGAP, ChickGBASE, Colibri, COPE, CottonDB, CSNDB, CUTG, CyanoBase, dbCFC, dbEST, dbSTS, DDBJ, DGP, DictyDb, Picty_cDB, DIP, DOGS, DOMO, DPD, DPInteract, ECDC, ECGC, EC02DBASE, EcoCyc, EcoGene, EMBL, EMD db, ENZYME, EPD, EpoDB, ESTHER, FlyBase, FlyView, GCRDB, GDB, GENATLAS, Genbank, GeneCards, Genlilesne, GenLink, GENOTK, GenProtEC, GIFTS, GPCRDB, GRAP, GRBase, gRNAsdb, GRR, GSDB, HAEMB, HAMSTERS, HEART-2DPAGE, HEXAdb, HGMD, HIDB, HIDC, HIVdb, HotMolecBase, HOVERGEN, HPDB, HSC-2DPAGE, ICN, ICTVDB, IL2RGbase, IMGT, Kabat, KDNA, KEGG, Klotho, LGIC, MAD, MaizeDb, MDB, Medline, Mendel, MEROPS, MGDB, MGI, MHCPEP5 Micado, MitoDat, MITOMAP, MJDB, MmtDB, Mol-R-Us, MPDB, MRR, MutBase, MycDB, NDB, NRSub, 0-lycBase, OMIA, OMIM, OPD, ORDB, OWL, PAHdb, PatBase, PDB, PDD, Pfam, PhosphoBase, PigBASE, PIR, PKR, PMD, PPDB, PRESAGE, PRINTS, ProDom, Prolysis, PROSITE, PROTOMAP, RatMAP, RDP, REBASE, RGP, SBASE, SCOP, SeqAnaiRef, SGD, SGP, SheepMap, Soybase, SPAD, SRNA db, SRPDB, STACK, StyGene,Sub2D, SubtiList, SWISS-2DPAGE, SWISS-3DIMAGE, SWISS- MODEL Repository, SWISS-PROT, TelDB, TGN, tmRDB, TOPS, TRANSFAC, TRR, UniGene, URNADB, V BASE, VDRR, VectorDB, WDCM, WIT, WormPep, etc ................. !!!!

# Bioinformatics Databases

**There are lots of Bioinformatics Databases**

**For a annotated listing of major bioinformatics databases please see the online handout**

**< [Major_Databases.pdf](Major_Databases.pdf) >**

AATDB, AceDb, ACUTS, ADB, AFDB, AGIS, AMSdb, ARR, AsDb,BBDB, Beanref, BioImage, BioMagResBank, BIOMDB, BLOCKS, BovGBASE, TKbase, CANSITE, CarbBank, CARBHYD, CATH, AP, ChickGBASE, Colibri, COPE, bEST, dbSTS, DDBJ, DGP, DictyDb, CDC, ECGC, EC02DBASE, THER, FlyBase, Link, HAEMB, HotMolecBase, IL2RGbase, IMGT, Kabat, KDNA, Medline, Mendel, MEROPS, MGDB, MGI, MITOMAP, MJDB, MmtDB, Mol-R-Us, MPDB, MRR, MutBase, 0-lycBase, OMIA, OMIM, OPD, ORDB, OWL, PAHdb, PatBase, PDD, Pfam, PhosphoBase, PigBASE, PIR, PKR, PMD, PPDB, PRESAGE, PRINTS, ProDom, Prolysis, PROSITE, PROTOMAP, RatMAP, RDP, REBASE, RGP, SBASE, SCOP, SeqAnaiRef, SGD, SGP, SheepMap, Soybase, SPAD, SRNA db, SRPDB, STACK, StyGene,Sub2D, SubtiList, SWISS-2DPAGE, SWISS-3DIMAGE, SWISS- MODEL Repository, SWISS-PROT, TelDB, TGN, tmRDB, TOPS, TRANSFAC, TRR, UniGene, URNADB, V BASE, VDRR, VectorDB, WDCM, WIT, WormPep, etc ................. !!!!

# Side-note: Databases come in all shapes and sizes

Databases can be of variable quality and often there are multiple databases with overlapping content.

# Primary, secondary & composite databases

Bioinformatics databases can be usefully classified into *primary*, *secondary* and *composite* according to their data source.

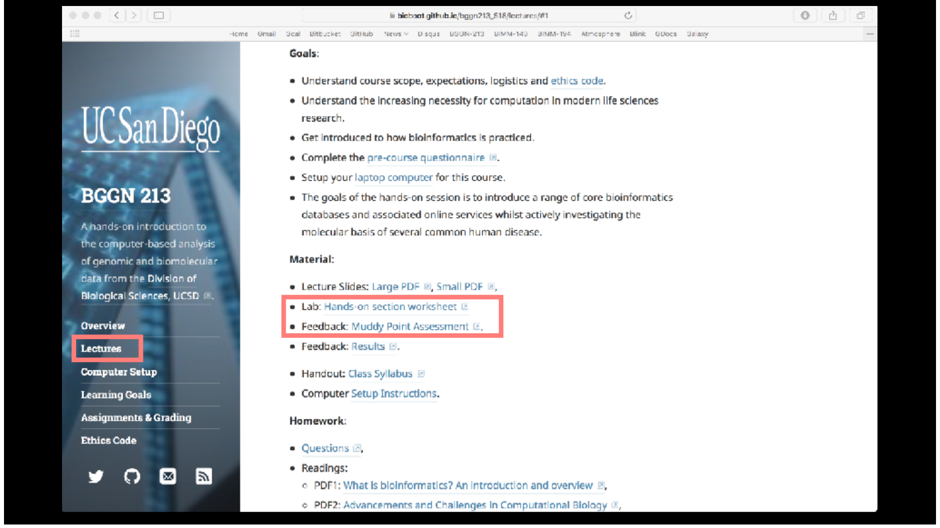
- **Primary databases** (or *archival databases*) consist of data derived experimentally.
  - **GenBank**: NCBI's primary nucleotide sequence database.
  - **PDB:** Protein X-ray crystal and NMR structures.
- **Secondary databases** (or *derived databases*) contain information derived from a primary database.
  - **RefSeq**: non redundant set of curated reference sequences primarily from GenBank
  - **PFAM**: protein sequence families primarily from UniProt and PDB
- **Composite databases** (or *metadatabases*) join a variety of different primary and secondary database sources.
  - **OMIM**: catalog of human genes, genetic disorders and related literature
  - **GENE**: molecular data and literature related to genes with extensive links to other databases.

# Today's Menu

| | |
|---|---|
| Course Logistics | Website, screencasts, survey, ethics, assessment and grading. |
| Learning Objectives | What you need to learn to succeed in this course. |
| Course Structure | Major lecture topics and specific leaning goals. |
| Introduction to Bioinformatis | Introducing the *what*, *why* and *how* of bioinformatics? |
| **Bioinformatics Database** | **Hands-on** exploration of several major databases and their associated tools. |

# Your Turn!

https://bioboot.github.io/bggn213_S18/lectures/#1



**BGGN 213**

A hands-on introduction to the computer-based analysis of genomic and biomolecular data from the Division of Biological Sciences, UCSD.

**Goals:**

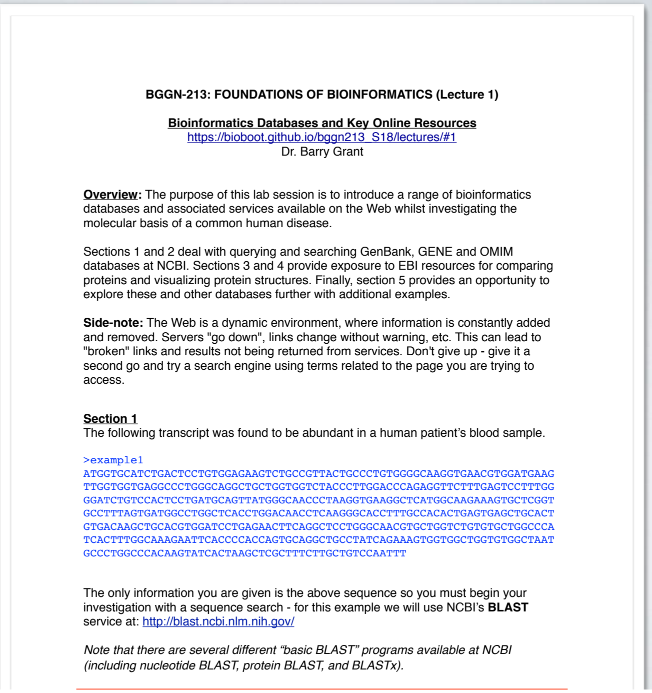
- Understand course scope, expectations, logistics and ethics code.
- Understand the increasing necessity for computation in modern life sciences research.
- Get introduced to how bioinformatics is practiced.
- Complete the pre-course questionnaire.
- Setup your laptop computer for this course.
- The goals of the hands-on session is to introduce a range of core bioinformatics databases and associated online services whilst actively investigating the molecular basis of several common human disease.

**Material:**

- Lecture Slides: Large PDF, Small PDF,
- Lab: Hands-on section worksheet
- Feedback: Muddy Point Assessment,
- Feedback: Results.
- Handout: Class Syllabus
- Computer Setup Instructions.

**Homework:**

- Questions,
- Readings:
  - PDF1: What is bioinformatics? An introduction and overview,
  - PDF2: Advancements and Challenges in Computational Biology,



**BGGN-213: FOUNDATIONS OF BIOINFORMATICS (Lecture 1)**

**Bioinformatics Databases and Key Online Resources**
https://bioboot.github.io/bggn213_S18/lectures/#1
Dr. Barry Grant

**Overview**: The purpose of this lab session is to introduce a range of bioinformatics databases and associated services available on the Web whilst investigating the molecular basis of a common human disease.

Sections 1 and 2 deal with querying and searching GenBank, GENE and OMIM databases at NCBI. Sections 3 and 4 provide exposure to EBI resources for comparing proteins and visualizing protein structures. Finally, section 5 provides an opportunity to explore these and other databases further with additional examples.

**Side-note:** The Web is a dynamic environment, where information is constantly added and removed. Servers "go down", links change without warning, etc. This can lead to "broken" links and results not being returned from services. Don't give up - give it a second go and try a search engine using terms related to the page you are trying to access.

**Section 1**

The following transcript was found to be abundant in a human patient's blood sample.

```
>example1
ATGGTGCATCTGACTCCTGTGGAGAAGTCTGCCGTTACTGCCCTGTGGGGCAAGGTGAACGTGGATGAAG
TTGGTGGTGAGGCCCTGGGCAGGCTGCTGGTGGTCTACCCTTGGACCCAGAGGTTCTTTGAGTCCTTTGG
GGATCTGTCCACTCCTGATGCAGTTATGGGCAACCCTAAGGTGAAGGCTCATGGCAAGAAAGTGCTCGGT
GCCTTTAGTGATGGCCTGGCTCACCTGGACAACCTCAAGGGCACCTTTGCCACACTGAGTGAGCTGCACT
GTGACAAGCTGCACGTGGATCCTGAGAACTTCAGGCTCCTGGGCAACGTGCTGGTCTGTGTGCTGGCCCA
TCACTTTGGCAAAGAATTCACCCCACCAGTGCAGGCTGCCTATCAGAAAGTGGTGGCTGGTGTGGCTAAT
GCCCTGGCCCACAAGTATCACTAAGCTCGCTTTCTTGCTGTCCAATTT
```

The only information you are given is the above sequence so you must begin your investigation with a sequence search - for this example we will use NCBI's **BLAST** service at: http://blast.ncbi.nlm.nih.gov/

*Note that there are several different "basic BLAST" programs available at NCBI (including nucleotide BLAST, protein BLAST, and BLASTx).*

# YOUR TURN!

- There are five major hands-on sections including:

1. BLAST, GenBank and OMIM @ **NCBI** [~35 mins]
2. GENE database @ **NCBI** [~15 mins]

— BREAK —

3. UniProt & Muscle @ **EBI** [~25 mins]
4. PFAM, PDB & NGL [~30 mins]

— BREAK —

5. Extension exercises [~30 mins]

- Please do answer the last review question (**Q19**).
- We encourage discussion and exploration!

## YOUR TURN!

- There are five major hands-on sections including:

| | End times: |
|---|---|
| 1. BLAST, GenBank and OMIM @ **NCBI** | [10:45 am] |
| 2. GENE database @ **NCBI** | [11:00 am] |
| — BREAK — | — 11:10 am — |
| 3. UniProt & Muscle @ **EBI** | [11:35 am] |
| 4. PFAM, PDB & NGL | [12:05 pm] |
| — BREAK — | — 12:15 am — |
| 5. Extension exercises | [12:45 pm] |

- Please do answer the last review question (**Q19**).
- We encourage discussion and exploration!

## SUMMARY

- Bioinformatics is computer aided biology.
- Bioinformatics deals with the collection, archiving, organization, and interpretation of a wide range of biological data.
- There are a large number of primary, secondary and tertiary bioinformatics databases.
- The NCBI and EBI are major online bioinformatics service providers.
- Introduced Gene, UniProt, PDB databases as well as a number of 'boutique' databases including PFAM and OMIM.

## HOMEWORK

https://bioboot.github.io/bggn213_S18/lectures/#1

☑ Complete the **initial course questionnaire**:

☑ Check out the "**Background Reading**" material online:

☑ Complete the **lecture 1 homework questions**: