



BIMM 194

Genomics, Big Data & Human Health

Barry Grant
UC San Diego

<http://thegrantlab.org/bimm194>

Today's Menu

Group Presentations

Introduction to student presentation assignment; rules and expectations.

Reading Guide

A 12 step guide to reading and understanding primary research articles.

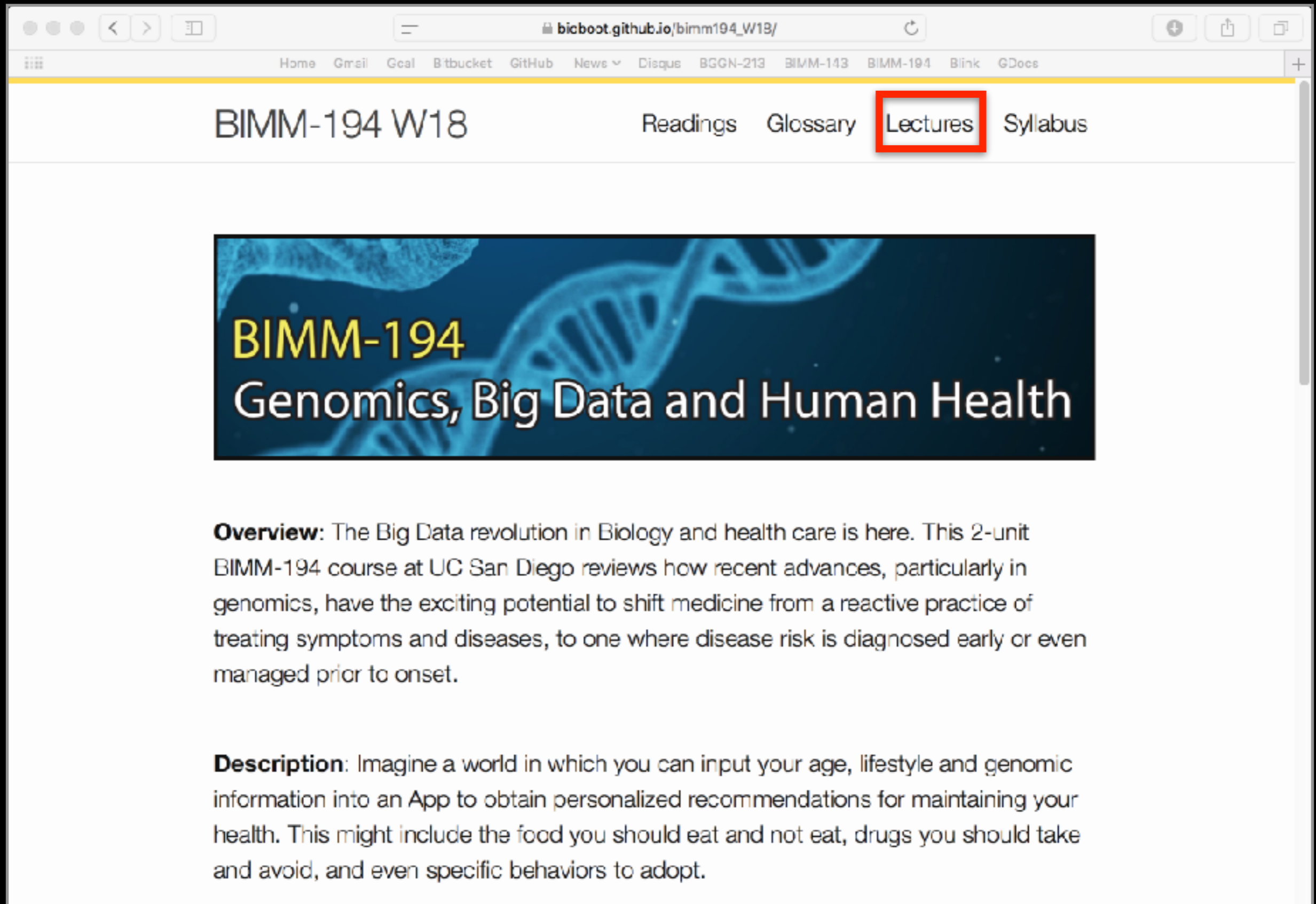
Selecting Papers

How to find and select primary research articles for presentation.

Group Get Together


Get to know your assigned group members.

<http://thegrantlab.org/bimm194/>



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BIMM-194 W18 Readings Glossary **Lectures** Syllabus



BIMM-194
Genomics, Big Data and Human Health

Overview: The Big Data revolution in Biology and health care is here. This 2-unit BIMM-194 course at UC San Diego reviews how recent advances, particularly in genomics, have the exciting potential to shift medicine from a reactive practice of treating symptoms and diseases, to one where disease risk is diagnosed early or even managed prior to onset.

Description: Imagine a world in which you can input your age, lifestyle and genomic information into an App to obtain personalized recommendations for maintaining your health. This might include the food you should eat and not eat, drugs you should take and avoid, and even specific behaviors to adopt.

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bioboot.github.io/bimm194_W18/lectures/

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BIMM-194 W18 Readings Glossary **Lectures** Syllabus

Lectures

All Lectures are Friday 2:00-3:20 pm in York Hall 3010 (YH 3010) ([Map](#)). Clicking on the class topics below will take you to corresponding lecture notes, homework assignments, and required reading material.

Course introduction & review of genome fundamentals

1 Fri, 01/12/18 Introduction to the course, Overview of major learning objectives and topic areas. Human genome review: What is DNA? What is a genome? What does the genome do? How do genomes differ between individuals? How is the genome decoded? Exploring what genetic errors are and what causes them.

Genomics and cancer treatment

What is cancer and how does it arise? Example genes implicated in cancer. What has been learned from genome

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4 Fri, 02/02/18

How to read a scientific paper & Introduction to student presentation assignments

A guide for selecting, reading and understanding peer-reviewed primary research articles, How to obtain a basic understanding of a published science paper and decide whether or not it is a reputable study? How does the described work contribute to advancing the scientific knowledge base or our technical capabilities? Introduction to student presentation assignments.

A 12 step guide to reading primary research articles,
Topics for student presentation
Student group assignment and presentation dates

Student group literature presentations

Group 1 & Group 2

Each week 2 student groups of 3 students each will present selected primary literature on recent genomic advances of relevance to biomedical science and health care. Topics

<http://thegrantlab.org/bimm194/>

Group	Presentation Date	Student Last Name	Student Given Name	Email
1	Fri: 02/09/18	Lu	Ailing	ailu@ucsd.edu
1	Fri: 02/09/18	Miao	Kathleen Lin	klmiao@ucsd.edu
1	Fri: 02/09/18	Qi	Xiaoyu	x1qi@ucsd.edu
2	Fri: 02/09/18	Del Rosario	Stefania Francesca Puy	sdelrosa@ucsd.edu
2	Fri: 02/09/18	Priestley-Miliana	Christopher Dyl	cpriestl@ucsd.edu
2	Fri: 02/09/18	Trinh	Jovonny	jotrinh@ucsd.edu
3	Fri: 02/16/18	Ding	Jeffrey	jeding@ucsd.edu
3	Fri: 02/16/18	Grundman	Jennifer Ann	jagrundm@ucsd.edu
3	Fri: 02/16/18	Sang	Hae Rin	hrsang@ucsd.edu
4	Fri: 02/16/18	Cho	In Ae	iacho@ucsd.edu
4	Fri: 02/16/18	Crinklaw	Austin Matthew	acrinkla@ucsd.edu
4	Fri: 02/16/18	Kilpatrick	Sidonie Katherine	skkilpat@ucsd.edu
5	Fri: 02/23/18	Lee	Su Han	shl073@ucsd.edu
5	Fri: 02/23/18	Mamidi	Anila	amamidi@ucsd.edu
5	Fri: 02/23/18	Menon	Vaibhav Dhinu	vdmenon@ucsd.edu

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2	Fri: 02/09/18	Del Rosario	Stefania Francesca Puy	sdelrosa@ucsd.edu
2	Fri: 02/09/18	Priestley-Miliana	Christopher Dyl	cpriestl@ucsd.edu
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3	Fri: 02/16/18	Grundman	Jennifer Ann	jagrundm@ucsd.edu
3	Fri: 02/16/18	Sang	Hae Rin	hrsang@ucsd.edu
4	Fri: 02/16/18	Cho	In Ae	iacho@ucsd.edu
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4	Fri: 02/16/18	Kilpatrick	Sidonie Katherine	skkilpat@ucsd.edu
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5	Fri: 02/23/18	Menon	Vaibhav Dhinu	vdmenon@ucsd.edu
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10	Fri: 03/09/18	Shang	Jason Y	jyshang@ucsd.edu
10	Fri: 03/09/18	Um	Christopher Jae	cjum@ucsd.edu

Presentations (25 min):

Based on **YOUR** review of primary literature on recent genomic advances of relevance to biomedical science and health care. Topics can be selected from the provided “[Readings](#)” online or address any of the following:

- How useful are genomic approaches to solving mystery genetic diseases?
- How can your genome directly help guide drug treatments for treating disease?
- Can genetic testing be used to predict intelligence or sports performance?
- Can genetic testing and genome editing be useful for choosing healthier embryos and producing designer babies?
- How will increased understating of epigenetics impact health care?
- How does the microbiome affect health and can it be rationally altered to improve health?
- Will having my genome sequenced affect my family members?
- Who has the right to know your genetic test results?

Rules and expectations

- Each week 2 student groups of 3 students each will present
- Topics may be selected from any of the primary articles noted in our blog posts to date or those related to our online topic list.
- Presentations should be 25 minutes in length with all group members contributing.
- Your groups PPT or PDF slides should be emailed to me by **9am on the Friday of your presentation.**
- Your chosen paper should be emailed to me no later than 12pm on the **Wednesday before your presentation.** I will post it online! **Once posted online no other group can select the same paper.**
- All audience members should read the paper before class and contribute to questions and discussions.

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Reading Guide

- As a newbie it can be easy to get frustrated by the dense, stilted writing and the unfamiliar jargon in primary research articles.
- I remember feeling this way!
- Reading and understanding research papers is a skill that every single scientist and doctor has had to learn.
- You can learn it too, but like any skill set it takes patience and practice.
- I have prepared a 12 step guide to reading primary research articles to help you: <https://tinyurl.com/bimm194-papers>

First Goal

- Your first goal should be to *obtain a basic understanding of a given scientific paper and decide whether or not it's a reputable study*
- Reading a scientific paper is a completely different process than reading an article about science in a blog or newspaper.

Recommendations

- Reading a single paper may take you a very long time at first.
 - ➔ Be patient with yourself.
 - ➔ The process will go much faster as you gain experience.
- You will have to take notes, read it multiple times, and probably go look up other papers for some of the details.

Do **NOT** read the paper in order of the presented sections

- Most primary research papers will be divided into the following sections:
 - ➔ Abstract,
 - ➔ Introduction,
 - ➔ Methods,
 - ➔ Results,
 - ➔ Discussion/Conclusions.
- The order will depend on which journal it's published in.
- Most journals also have additional files (called *Supplementary Online Information*).

Recommendation:

Follow these 12 steps for effectively reading a research paper

Step 1.

- Before you begin reading, take note of the authors and their institutional affiliations.
 - ➔ Some institutions (e.g. University of California, San Diego) are well-respected; others (e.g. the Discovery Institute) are agenda-driven.
- Also take note of the journal in which it's published. Reputable (biomedical) journals will be indexed by [Pubmed](#).
- Beware of [questionable journals](#).

Step 2.

- As you read, write down every single word that you don't understand.
 - ➔ You are going to have to look them all up
 - ➔ You won't understand the paper if you don't understand the vocabulary..

Step 3.

- Read the **introduction first**, NOT the abstract.
- When I'm choosing papers to read, I decide what's relevant to my interests based on a combination of the title and abstract.
- But when I've got a collection of papers assembled for **deep reading**, I always read the abstract LAST.
- I do this because abstracts contain a succinct summary of the entire paper, and I'm concerned about inadvertently becoming biased by the authors' interpretation of the results.

Step 4.

- Identify the **BIG QUESTION!**
- Before you focus in on the question “*what is this paper about?*”, identify the bigger question, namely:
 - “*What problem is this entire field trying to solve?*”
- This helps you focus on why this research is being done and how it might fit in to advancing the frontier of knowledge or technical capabilities.

Step 5.

- Summarize the background in five sentences or less
- Use these questions to guide you:
 - ➔ What work has been done before in this field to answer the BIG QUESTION?
 - ➔ What are the limitations of that work?
 - ➔ What, according to the authors, needs to be done next?
- Try to be be concise and really think about the context of this research. You need to be able to explain why this research has been done in order to understand it.

Step 6.

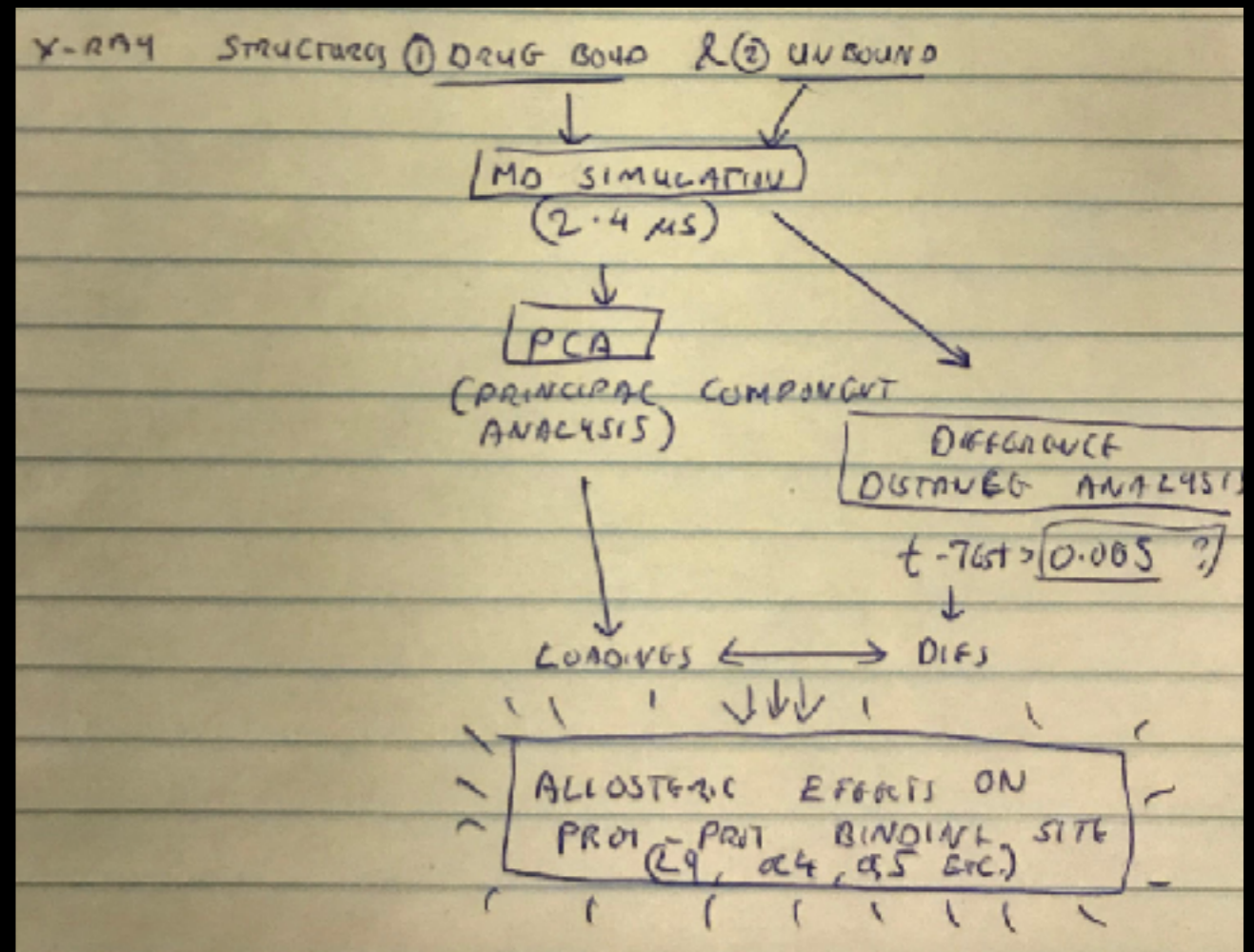
- Identify the **SPECIFIC QUESTION(S)** addressed in this paper
 - ➔ What exactly are the authors trying to answer with their research?
- There may be multiple questions, or just one. Write them down.
- If it's the kind of research that tests one or more hypotheses, identify it/them and write them down.

Step 7.

- Identify the approach as stated in the introduction section.
- What are the authors going to do to answer the SPECIFIC QUESTION(S)?
- We will find more fine grained details of the approach in the methods section addressed in Step 8.

Step 8.

- Now read the methods section and draw a diagram for each experiment, showing exactly what the authors did.
- Include as much detail as you need to fully understand the work.



TIP: At this stage of your career you don't need to understand the methods in enough detail to replicate the experiments .

However, you are not ready to move on to the results until you can explain the basics of the methods to someone else.

Step 9.

- Now it is time to read the **results section** and write a paragraph or two that summarizes the results for each experiment, each figure, and each table.
 - ➔ Don't yet try to decide what the results mean, just write down what they are.
- You'll find that, particularly in good papers, the majority of the results are summarized in the figures and tables
 - ➔ Typically one major figure (or figure panel such as A, B, C etc.) per major experiment.

TIP: Some typical things to pay attention to in the results section include:

- Any time the words “significant” or “non-significant” are used. These have precise statistical meanings. Read more about this [here](#).
- If there are graphs, do they have error bars on them? For certain types of studies, a lack of confidence intervals is a major red flag.
- The sample size. Has the study been conducted on 10, or 10,000 people? (For some research purposes, a sample size of 10 is sufficient, but for most studies larger is better).

Step 10.

- Before moving on to the conclusions/discussion section ask yourself:
 - ➔ Do the results answer the SPECIFIC QUESTION(S)?
 - ➔ What do you think there results mean?
- Don't move on until you have thought about this
 - ➔ It is a good habit to start forming your own interpretations first

Step 11.

- Now it is time to read the **conclusion/discussion** sections.
 - ➔ What do the authors think the results mean?
 - ➔ Do you agree with them?
 - ➔ Can you come up with any alternative way of interpreting them?
 - ➔ Do the authors identify any weaknesses in their own study?
 - ➔ Do you see any that the authors missed? (Don't assume they're infallible!)
 - ➔ What do they propose to do as a next step? Do you agree with that?

Step 12.

- Finally it is time to go back to the beginning and read the **abstract**.
 - ➔ Does it match what the authors said in the paper?
 - ➔ Does it fit with your interpretation of the paper?

Final wrap-up:

- After you have finished your 12 steps it can be very informative to find out what others say about this paper.
- Who are the acknowledged experts in this particular field?
- Do they have criticisms of the study that you haven't thought of, or do they generally support it?
- Here's a place where I do recommend you use google!
- But do it last, so you are better prepared to think critically about what other people say.

Summary

- Reading the primary literature can be a significant amount of work for early stage scientists.
- It will however get much easier with practice, habit, and following our 12 step guide!

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The screenshot shows the PubMed website interface. At the top, there is a navigation bar with links for Home, Gmail, Gcal, Bitbucket, GitHub, News, Disqus, BGGN-213, BIMM-143, BIMV-194, Blink, and GDocs. Below this is a blue header with the NCBI logo and dropdown menus for Resources and How To. The main content area features the PubMed logo and the text "National Library of Medicine National Institutes of Health". A search bar is present, with a dropdown menu currently set to "PubMed". A red box highlights the "Advanced" search option. Below the search bar is a large banner with a background image of a bookshelf and a tablet. The banner text reads: "PubMed comprises more than 28 million citations for biomedical literature from MEDLINE, life science journals, and online books. Citation links to full-text content from PubMed Central and publisher websites are provided when available." Below the banner are three columns of links. The first column, titled "Using PubMed", includes links for "PubMed Quick Start Guide", "Full Text Articles", and "PubMed FAQs". The second column, titled "PubMed Tools", includes links for "PubMed Mobile", "Single Citation Matcher", and "Batch Citation Matcher". The third column, titled "More Resources", includes links for "MeSH Database", "Journals in NCBI", and "Clinical Trials".

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The screenshot shows the PubMed Advanced Search Builder interface. At the top, there are navigation links: PubMed Home, More Resources, and Help. The main heading is "PubMed Advanced Search Builder". Below this, a search query is displayed in a text box, highlighted with a red border: `((genomic[Title/Abstract]) AND immunotherapy) AND ("2017/01/01"[Date - Publication] : "3000"[Date - Pub`. Below the query is an "Edit" link. The "Builder" section contains four rows of search criteria, each with a field selector, a text input, and a "Show index list" link. The first row has "Title/Ab:" selected and "genomic" entered. The second row has "AND" selected, "All Field" selected, and "immunotherapy" entered. The third row has "AND" selected, "Date - F" selected, "2017/01/01" entered in the first date field, "to" selected, "present" entered in the second date field, and "Show index list" link. The fourth row has "AND" selected, "All Field" selected, and an empty text input field. At the bottom, there is a "Search" button and a link to "Add to history".

PubMed Home More Resources Help

PubMed Advanced Search Builder

`((genomic[Title/Abstract]) AND immunotherapy) AND ("2017/01/01"[Date - Publication] : "3000"[Date - Pub`

[Edit](#)

Builder

Title/Ab:	genomic	-	Show index list
AND	All Field	immunotherapy	- Show index list
AND	Date - F	2017/01/01 to present	- Show index list
AND	All Field		- + Show index list

[Search](#) or [Add to history](#)

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The screenshot shows the PubMed website interface. At the top, the URL www.ncbi.nlm.nih.gov/pubmed is visible in the browser's address bar. Below the navigation bar, the search query `((genomic[Title/Abstract]) AND immunotherapy) AND ("2017/01/01"[Date - - PubMed - NCBI])` is entered in the search box and highlighted with a red border. The search results page displays the following information:

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- Text availability:** Abstract, Free full text, Full text
- Format:** Summary
- Sort by:** Best Match
- Per page:** 20
- Filters:** Managed
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- Filters activated:** Free full text. [Clear all](#) to show 222 items.
- Search results list:**
 - [Current state of immunotherapy for non-small cell lung cancer.](#)
1. Malhotra J, Jabbour SK, Aisner J.
Transl Lung Cancer Res. 2017 Apr;6(2):196-211. doi: 10.21037/tlcr.2017.03.01. Review. Erratum in: [Transl Lung Cancer Res. 2017 Oct;6\(5\):612.](#)
- Titles with y terms:** Hyperprogressive Immunotherapy [Clin], Immunotherapy [Nat Rev], Genomic an

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7. [Analysis of 100,000 human cancer genomes reveals the landscape of tumor mutational burden.](#)
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- [Cancer Evolution during Immunotherapy.](#)
Andrews MC, Wargo JA
Cell [01 Nov 2017, 171(4):740-742]
Cited: 0 times (PMID:29100071)
- [Tumor and Microenvironment Evolution during Immunotherapy with Nivolumab.](#)
Riaz N, Havel JJ, Makarov V, Desrichard A, Urba WJ, Sims JS, Hodi FS, Martín-Algarra S, Mandal R, Sharfman WH, Bhatia S, Hwu WJ, Gajewski TF [...] Chan TA
Cell [11 Oct 2017, 171(4):934-949.e15]
Cited: 3 times (PMID:29033130)
- [Synthetic RNA-Based Immunomodulatory Gene Circuits for Cancer Immunotherapy.](#)
Nissim L, Wu MR, Pery E, Binder-Nissim A, Suzuki HI, Stupp D, Wehrspaun C, Tabach Y, Sharp PA, Lu TK
Cell [18 Oct 2017, 171(5):1138-1150.e15]

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7	Fri: 03/02/18	Eskandar	Joy Samir	jeskanda@ucsd.edu
7	Fri: 03/02/18	Grudzien	Jessica Lauren	jgrudzie@ucsd.edu
7	Fri: 03/02/18	Ni	Haowei	hani@ucsd.edu
8	Fri: 03/02/18	Alvarez Alvarez	Brenda Belen	bbalvare@ucsd.edu
8	Fri: 03/02/18	Halim	Dylan Patrick	dphalim@ucsd.edu
8	Fri: 03/02/18	Tubb	Helena May	htubb@ucsd.edu
9	Fri: 03/09/18	Maeda	Meg Ying	mmaeda@ucsd.edu
9	Fri: 03/09/18	Park	Sewon	swp020@ucsd.edu
9	Fri: 03/09/18	Wang	Jingjun	jiw158@ucsd.edu
10	Fri: 03/09/18	Pandya	Shivam Ramesh	spandya@ucsd.edu
10	Fri: 03/09/18	Shang	Jason Y	jyshang@ucsd.edu
10	Fri: 03/09/18	Um	Christopher Jae	cjum@ucsd.edu

Rules and expectations

- Each week 2 student groups of 3 students each will present
- Topics may be selected from any of the primary articles noted in our blog posts to date or those related to our online topic list.
- Presentations should be 25 minutes in length with all group members contributing.
- Your groups PPT or PDF slides should be emailed to me by **9am on the Friday of your presentation.**
- Your chosen paper should be emailed to me no later than 12pm on the **Wednesday before your presentation.** I will post it online! **Once posted online no other group can select the same paper.**
- All audience members should read the paper before class and contribute to questions and discussions.

Thats all folks!