BIMM 194

Genomics, Big Data & Human Health

Barry Grant

UC San Diego

http://thegrantlab.org/bimm194

Today's Menu

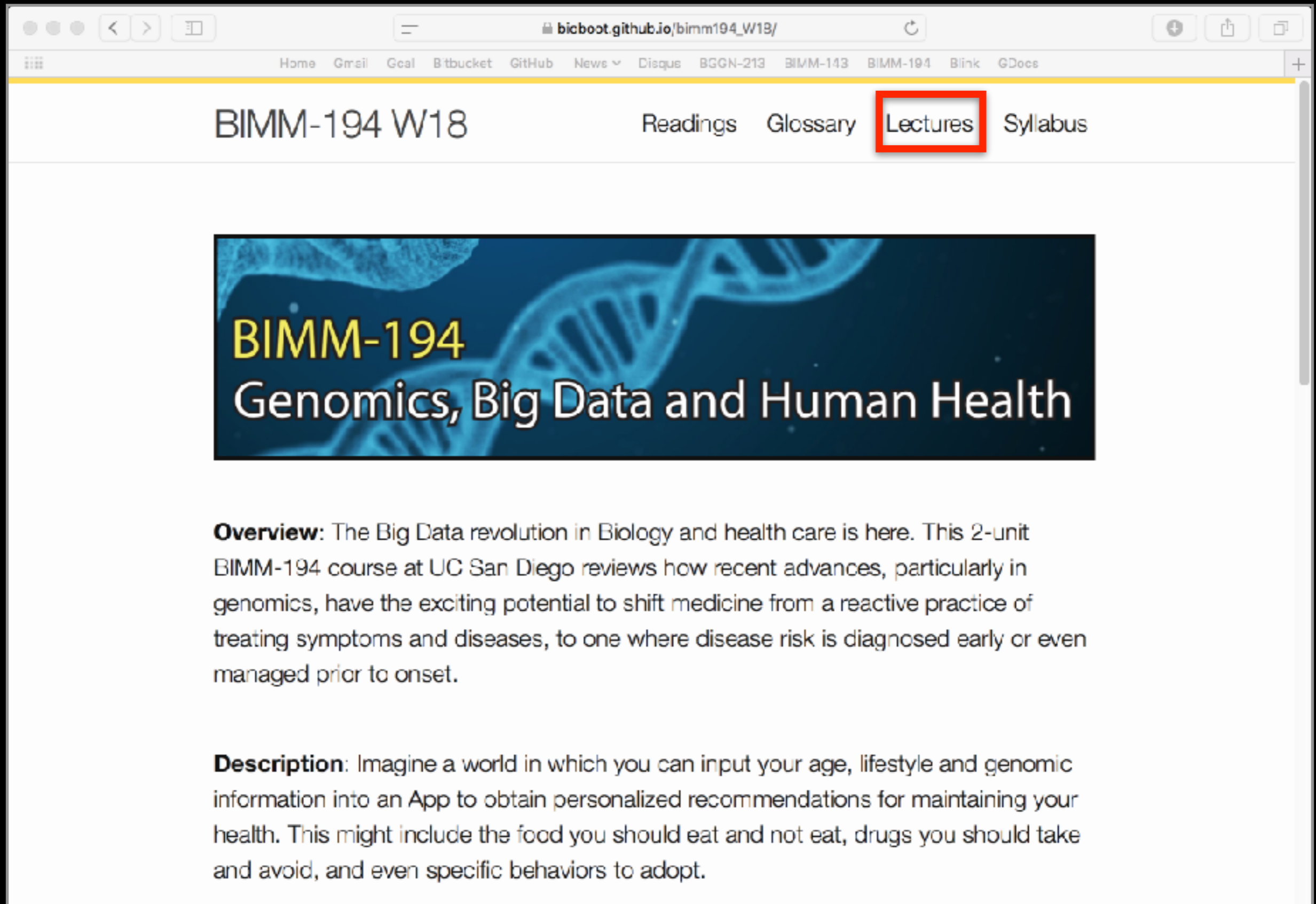|	
Group Presentations	Introduction to student presentation assignment; rules and expectations.
Reading Guide	A 12 step guide to reading and understanding primary research articles.
Selecting Papers	How to find and select primary research articles for presentation.
Group Get Together	Get to know your assigned group members.

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bioboot.github.io/bimm194_W18/lectures/

Home   Gmail   Gcal   Bitbucket   GitHub   News ∨   Disqus   BGGN-213   BIMM-143   BIMM-194   Blink   GDocs

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

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Navigation bar: Home   Gmail   Gcal   Bitbucket   GitHub   News   Disqus   BGGN-213   BIMM-143   BIMV-194   Blink   GDocs

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
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2	Fri: 02/09/18	Priestley-Milianta	Christopher Dyl	cpriestl@ucsd.edu
2	Fri: 02/09/18	Trinh	Jovonny	jotrinh@ucsd.edu
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3	Fri: 02/16/18	Sang	Hae Rin	hrsang@ucsd.edu
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5	Fri: 02/23/18	Mamidi	Anila	amamidi@ucsd.edu
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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	A 12 step guide to reading and understanding primary research articles.
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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.
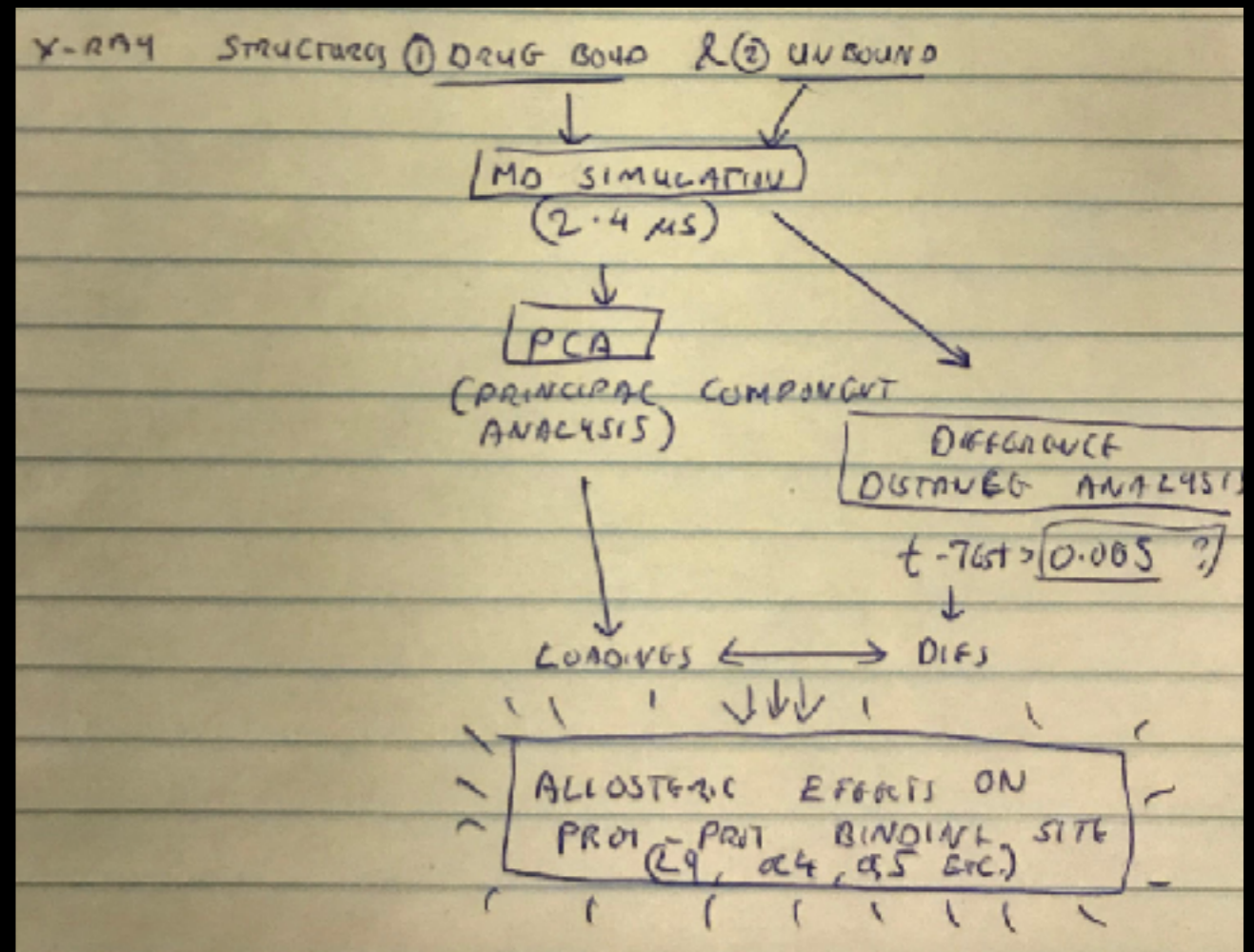
TIP: At this stage of your career you don't need to understand the methods in enough detail to replicate the experiments .

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 easer with practice, habit, and following our 12 step guide!

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((genomic[Title/Abstract]) AND Immunotherapy) AND ("2017/01/01"[Date – – PubMed – NCBI   The challenges of tumor genetic diversity. – PubMed – NCBI

PMID: 28808500   **Free PMC Article**

Similar articles

☑
7. Analysis of 100,000 human cancer genomes reveals the landscape of tumor mutational burden.

Chalmers ZR, Connelly CF, Fabrizio D, Gay L, Ali SM, Ennis R, Schrock A, Campbell B, Shlien A, Chmielecki J, Huang F, He Y, Sun J, Tabori U, Kennedy M, Lieber DS, Roels S, White J, Otto GA, Ross JS, Garraway L, Miller VA, Stephens PJ, Frampton GM.

Genome Med. 2017 Apr 19;9(1):34. doi: 10.1186/s13073-017-0424-2.

PMID: 28420421   **Free PMC Article**

Similar articles

☐
8. Resources for Interpreting Variants in Precision **Genomic** Oncology Applications.

Tsang H, Addepalli K, Davis SR.

Front Oncol. 2017 Sep 19;7:214. doi: 10.3389/fonc.2017.00214. eCollection 2017. Review.

PMID: 28975082   **Free PMC Article**

Similar articles

Recommendation:
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https://europepmc.org

https://europepmc.org

https://europepmc.org/advancesearch

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Others Search Options

- Web of Science
  ➡ https://webofknowledge.com/

- Google Scholar
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- And others…

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