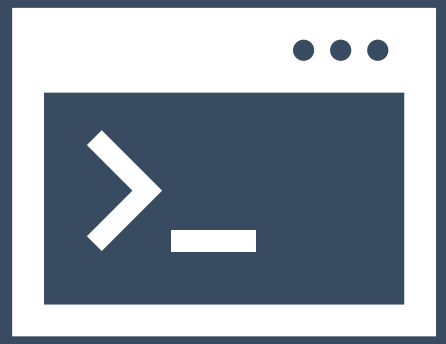
HBC
Harvard Chan Bioinformatics Core

Current Topics in Bioinformatics

https://hbctraining.github.io/Training-modules/



Harvard Chan Bioinformatics Core

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HBC consulting: bioinformatics@hsph.harvard.edu

Introductions!

HARVARD MEDICAL SCHOOL

DF/HCC
DANA-FARBER / HARVARD CANCER CENTER

HARVARD T.H. CHAN SCHOOL OF PUBLIC HEALTH

Training

- **Basic Data Skills**
  - Shell
  - R
- **Advanced Topics: Analysis of high-throughput sequencing data**
  - Chromatin Biology
  - Bulk RNA-seq
  - Differential Gene Expression
  - scRNA-seq
  - Variant Calling
- **Current Topics in Bioinformatics**

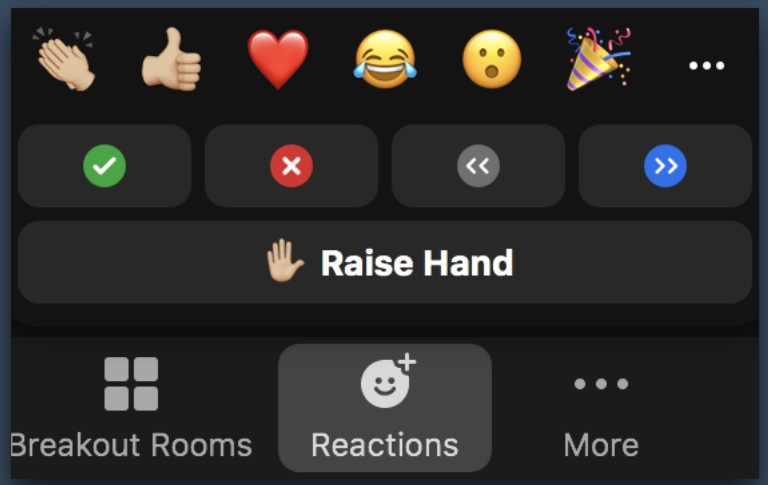
Consulting

- **Transcriptomics:** RNA-seq, small RNA-seq, scRNA-seq
- **Epigenetics:** ChIP-seq, genome wide methylation, ATAC-seq
- **DNA Variation:** WGS, resequencing, exome-seq, CNV studies
- **Functional enrichment analysis**
- **Experimental design help**
- **Grant support**

https://bioinformatics.sph.harvard.edu/

Odds & Ends

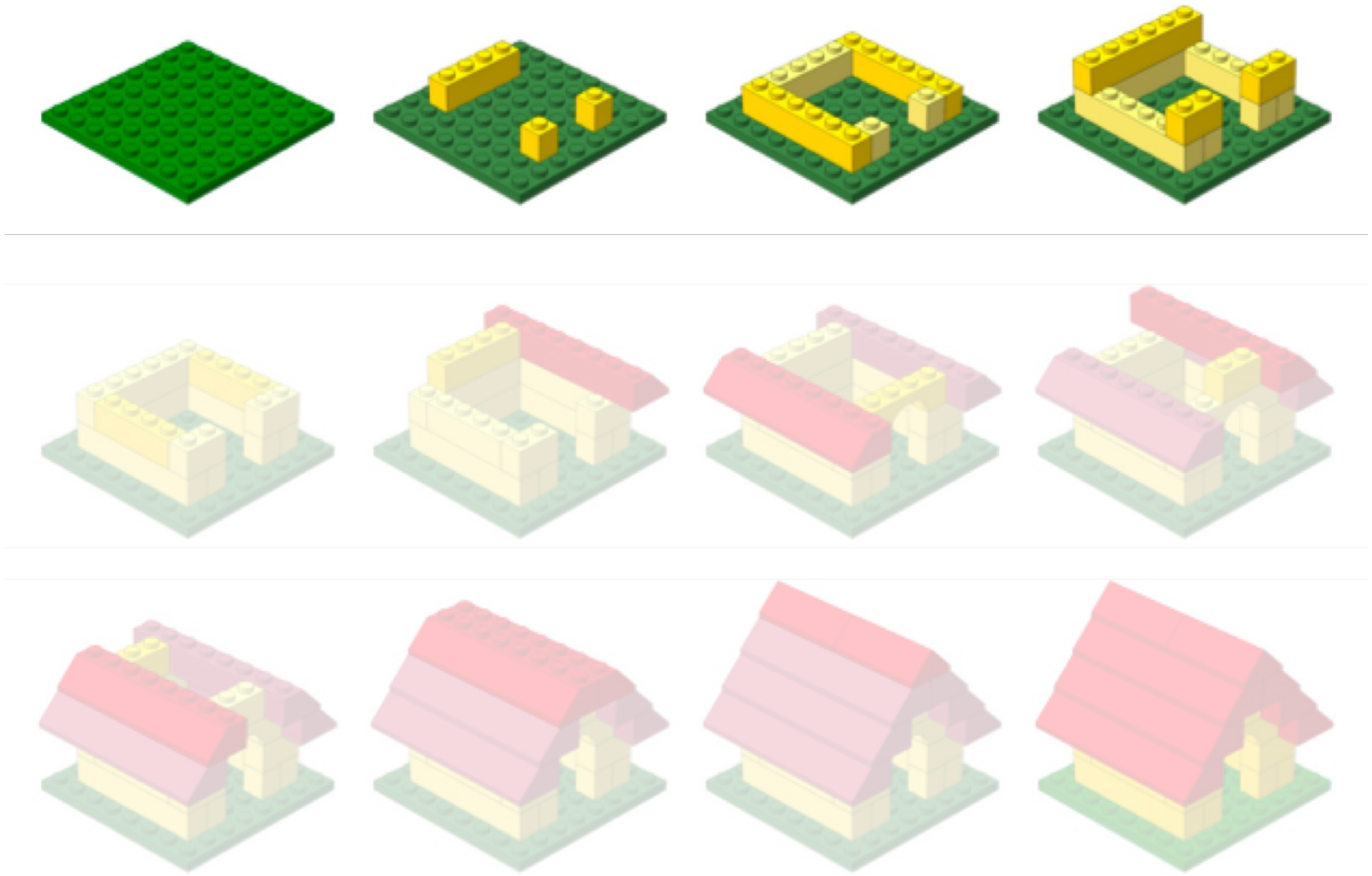
- Quit/minimize all applications that are not required for class
- Are you all set?
  -  = "agree", "I'm all set"
  -  = "disagree", "I need help"



Odds & Ends

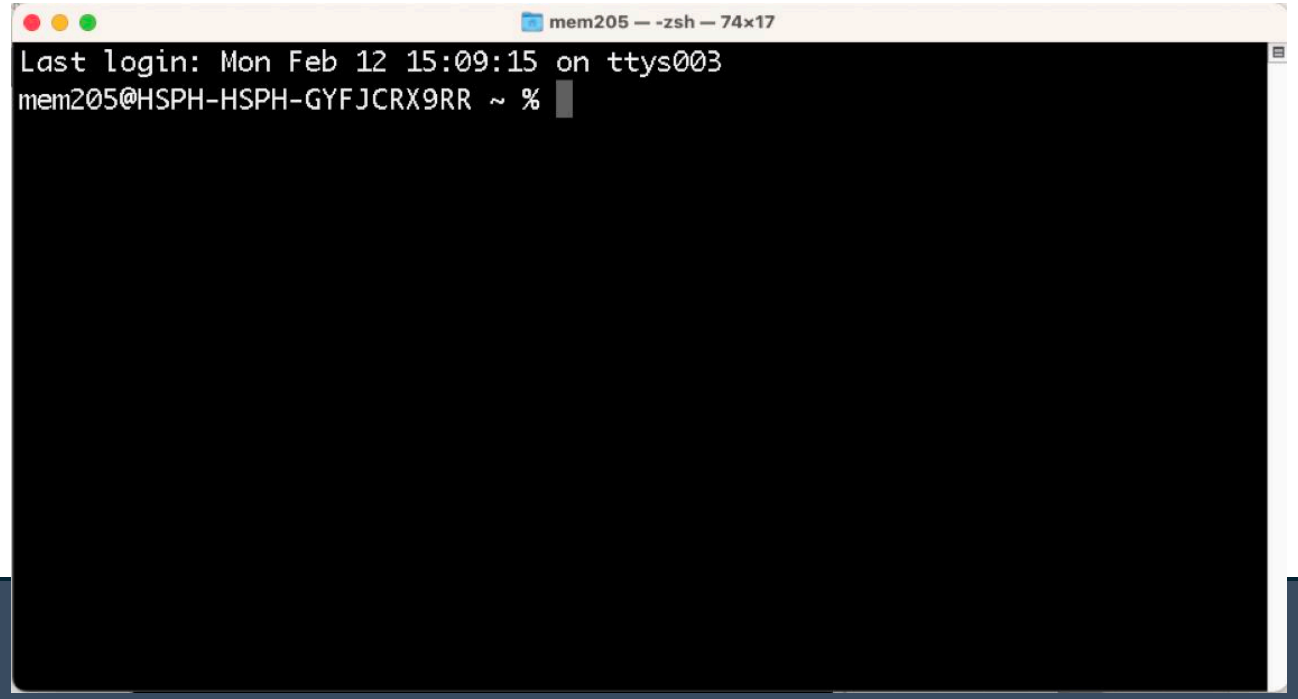
- Questions for the presenter?
  - Post the question in the Chat window OR
  - 

when the presenter asks for questions
  - Let the Moderator know
- Technical difficulties with software?
  - Start a private chat with the Troubleshooter with a description of the problem



Learning R

What is shell?



- Shell is a program that allows users to control Unix/Linux OS with text commands

Terminology

- **Unix/Linux** - The operating systems of High Performance Computers (HPC)

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- **Unix/Linux** - The operating systems of High Performance Computers (HPC)
- **Shell** - A program that allows users to control Unix/Linux OS with text commands
- **Bash** - The most prevalent kind of shell

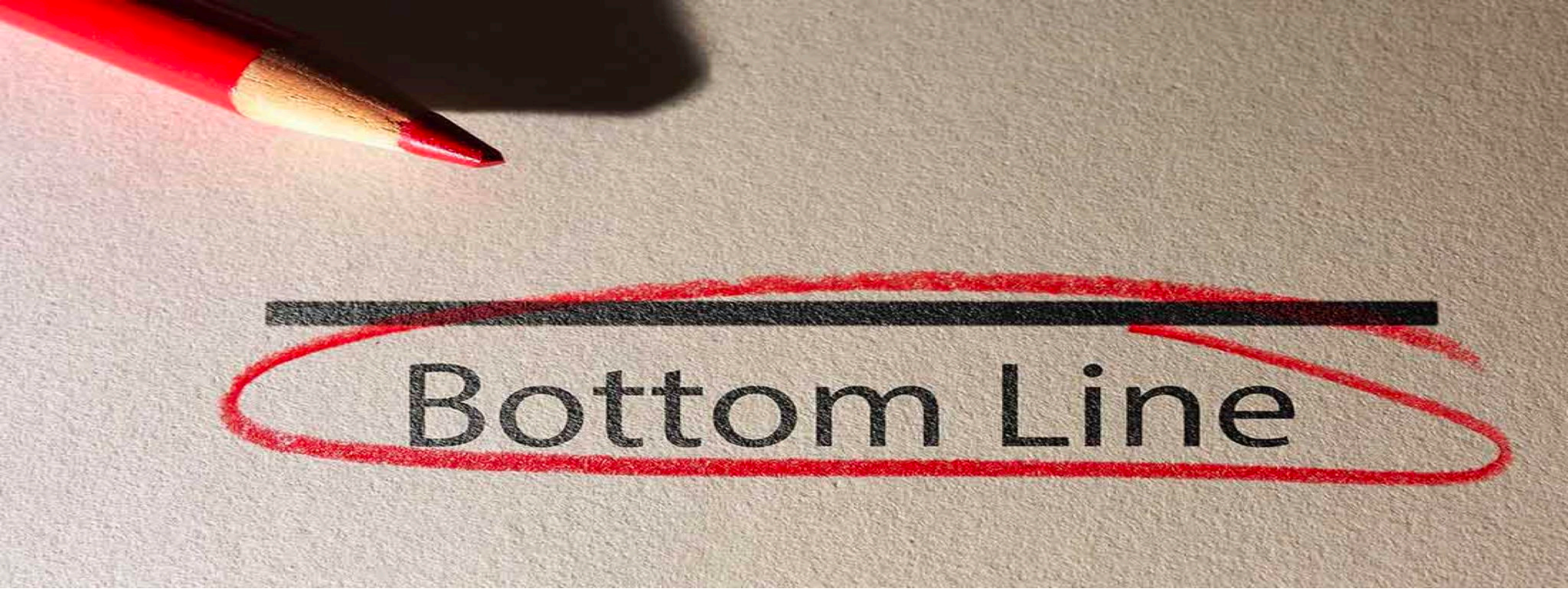

Image source: Balboa Capital Blog

If you plan to process raw high throughput sequencing data yourself, you will need to learn shell.

1. You need more resources than what is available on your laptop

- Sequence data files are LARGE
- Processing these data require increased CPU and memory
- High performance compute clusters have the necessary resources!

2. Many bioinformatics tools are only available as command-line tools

10XGenomics/
cellranger

10x Genomics Single Cell Analysis

10x GENOMICS™

staraligner

gatk4

Salmon 1.10.2

SAMtools

bedtools

3. Many genomics filetypes are binary

- Binary files are not human readable
- Binary files need an interpreter

4. There are many useful commands that can help work with enormous data files

- Commands for easily viewing files: less, cat, head, tail

```
0  ##gff-version 3.2.1
1  ##sequence-region ctg123 1 1497228
2  ctg123 . gene            1000  9000  .  +  .  ID=gene00001;Name=EDEN
3  ctg123 . TF_binding_site 1000  1012  .  +  .  ID=tfbs00001;Parent=gene00001
4  ctg123 . mRNA            1050  9000  .  +  .  ID=mRNA00001;Parent=gene00001;Name=EDEN.1
5  ctg123 . mRNA            1050  9000  .  +  .  ID=mRNA00002;Parent=gene00001;Name=EDEN.2
6  ctg123 . mRNA            1300  9000  .  +  .  ID=mRNA00003;Parent=gene00001;Name=EDEN.3
7  ctg123 . exon            1300  1500  .  +  .  ID=exon00001;Parent=mRNA00003
8  ctg123 . exon            1050  1500  .  +  .  ID=exon00002;Parent=mRNA00001,mRNA00002
9  ctg123 . exon            3000  3902  .  +  .  ID=exon00003;Parent=mRNA00001,mRNA00003
10 ctg123 . exon            5000  5500  .  +  .  ID=exon00004;Parent=mRNA00001,mRNA00002,mRNA00003
11 ctg123 . exon            7000  9000  .  +  .  ID=exon00005;Parent=mRNA00001,mRNA00002,mRNA00003
12 ctg123 . CDS             1201  1500  .  +  0  ID=cds00001;Parent=mRNA00001;Name=edenprotein.1
13 ctg123 . CDS             3000  3902  .  +  0  ID=cds00001;Parent=mRNA00001;Name=edenprotein.1
14 ctg123 . CDS             5000  5500  .  +  0  ID=cds00001;Parent=mRNA00001;Name=edenprotein.1
15 ctg123 . CDS             7000  7600  .  +  0  ID=cds00001;Parent=mRNA00001;Name=edenprotein.1
16 ctg123 . CDS             1201  1500  .  +  0  ID=cds00002;Parent=mRNA00002;Name=edenprotein.2
17 ctg123 . CDS             5000  5500  .  +  0  ID=cds00002;Parent=mRNA00002;Name=edenprotein.2
18 ctg123 . CDS             7000  7600  .  +  0  ID=cds00002;Parent=mRNA00002;Name=edenprotein.2
19 ctg123 . CDS             3301  3902  .  +  0  ID=cds00003;Parent=mRNA00003;Name=edenprotein.3
20 ctg123 . CDS             5000  5500  .  +  1  ID=cds00003;Parent=mRNA00003;Name=edenprotein.3
21 ctg123 . CDS             7000  7600  .  +  1  ID=cds00003;Parent=mRNA00003;Name=edenprotein.3
22 ctg123 . CDS             3391  3902  .  +  0  ID=cds00004;Parent=mRNA00003;Name=edenprotein.4
23 ctg123 . CDS             5000  5500  .  +  1  ID=cds00004;Parent=mRNA00003;Name=edenprotein.4
24 ctg123 . CDS             7000  7600  .  +  1  ID=cds00004;Parent=mRNA00003;Name=edenprotein.4
```

5. Automation is the name of the game

- Launch many jobs with one command
- Code is used and reused to iterate tasks over multiple files
- Parallelization to complete tasks using multiple cores and increase speed!

This could be you watching your analysis run!