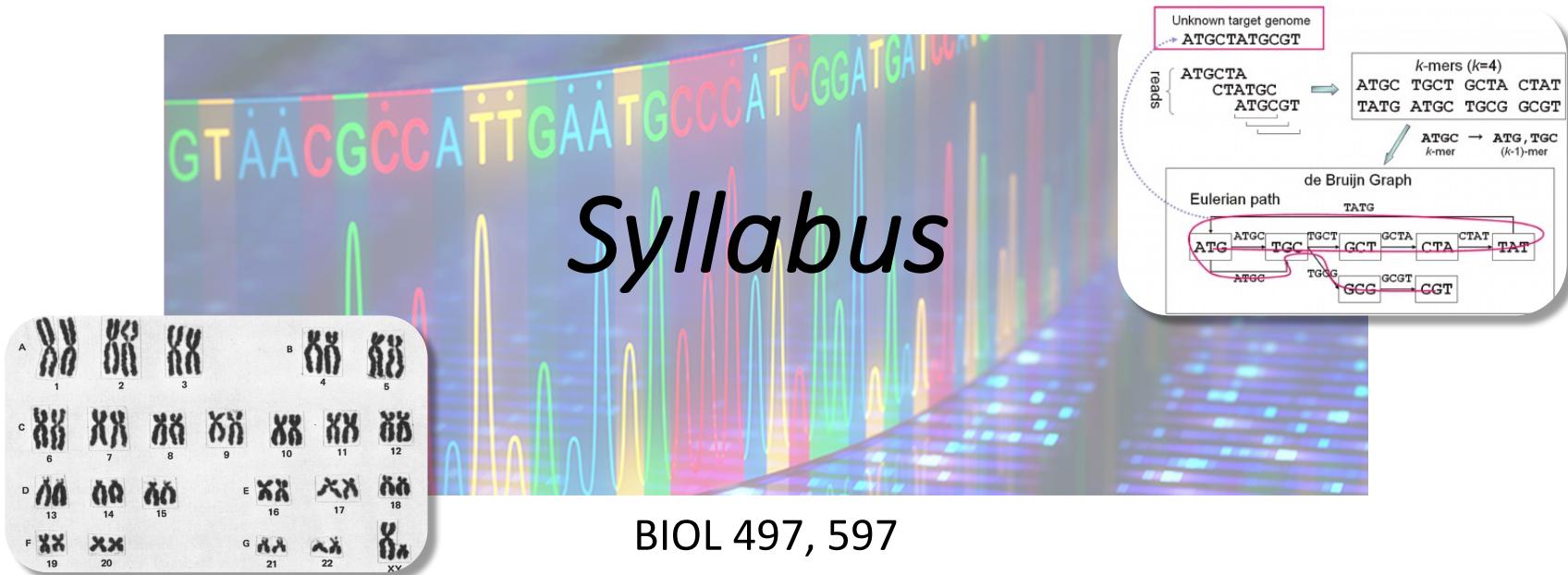


Genomics & Bioinformatics



BIOL 497, 597

Boise State University

Spring 2022



INSTRUCTOR

- **Name:** Sven BUERKI
- **Office:** Science Building, office 114
- **Email:** svenbuerki@boisestate.edu



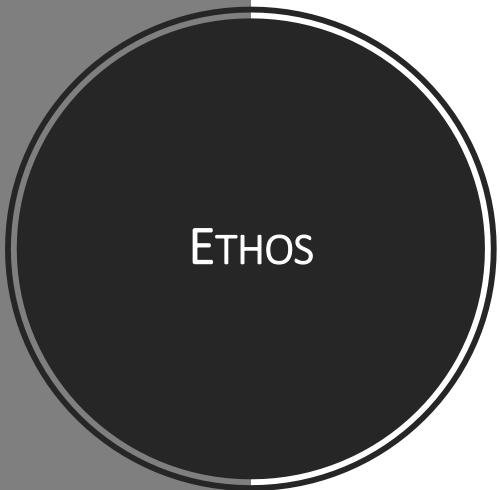
CLASS
LOCATION &
MEETING TIMES

- **Location:** SCNC149 computer room.
- **Lectures:** Wednesdays from 9:00-10:50 AM.
- **Labs:** Fridays from 9:00-10:30 AM.
- No office hours, but please contact me if you want to set an appointment.



HEALTH & SAFETY

- **Our priority is your safety!**
- The following rules apply for in-person classes:
 - ✓ Wear masks and maintain physical distancing.
 - ✓ Use sanitizing wipes to clean surfaces (desk and keyboard) at beginning and end of class.
 - ✓ Keep using the same seat throughout the semester. This information should be entered into MyBoiseState.
 - ✓ Do not attend class if you are sick.
- For more information consult the BSU campus guidelines at <https://www.boisestate.edu/coronavirus-response/>.



ETHOS

- Everyone here is smart; distinguish yourself by being kind.

Kindness in Science is an inclusive approach that fosters diversity, respect, wellbeing & openness leading to better science outcomes.



#KindnessInScience
by @jennypannell

INTRODUCTIONS



RESOURCES

<https://svenbuerki.github.io/Genomics-Bioinformatics>

Genomics & Bioinformatics

Home Chapters Mini-reports Lab. Tutorials Lexicon References

BIOL 497/597 - Genomics & Bioinformatics

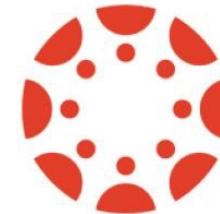
Syllabus & Timetable
Sven Buerki - Boise State University
2022-01-04

[Download pdf version](#)
[Raw data on GitHub](#)

1 Instructor

- Name: Sven Buerki
- Office: Science building, office 114 (ground floor).
- Email: svenbuerki@boisestate.edu
- Office hours: By appointment.

Shared Google Drive

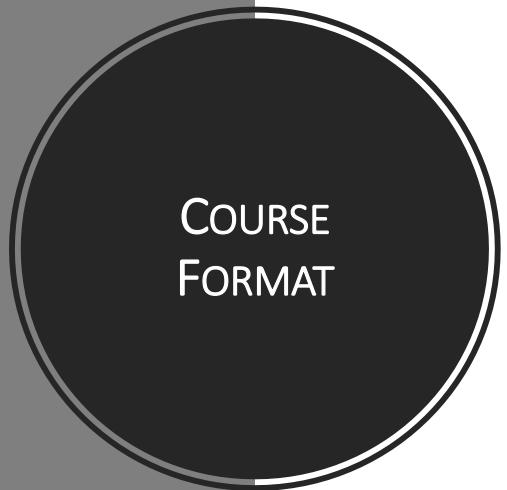


canvas

COURSE GOAL & DESCRIPTION

The goal is to provide students with the theoretical and applied knowledge in genomics and bioinformatics to sequence, assemble and annotate genomes, especially for non-model organisms.

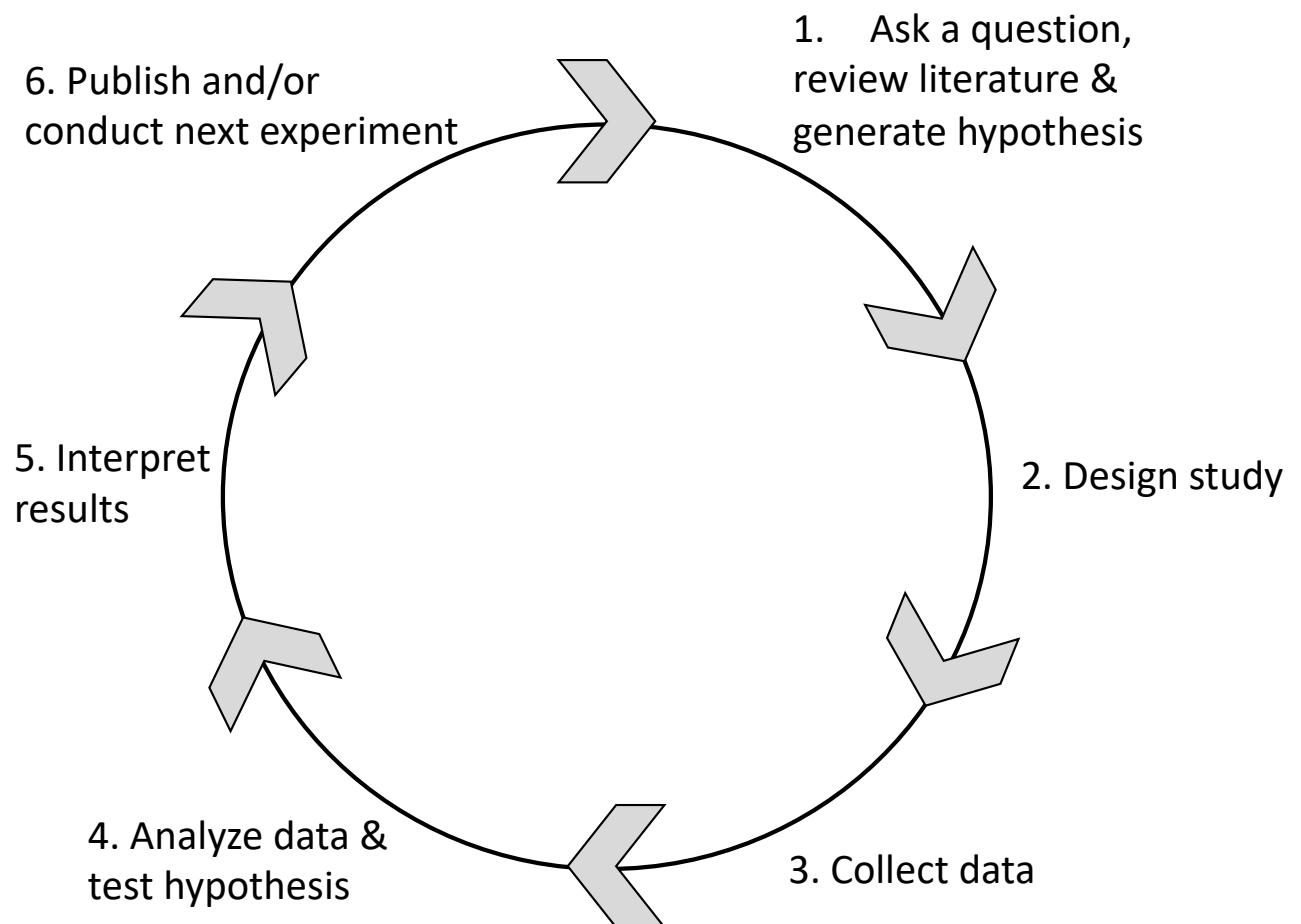


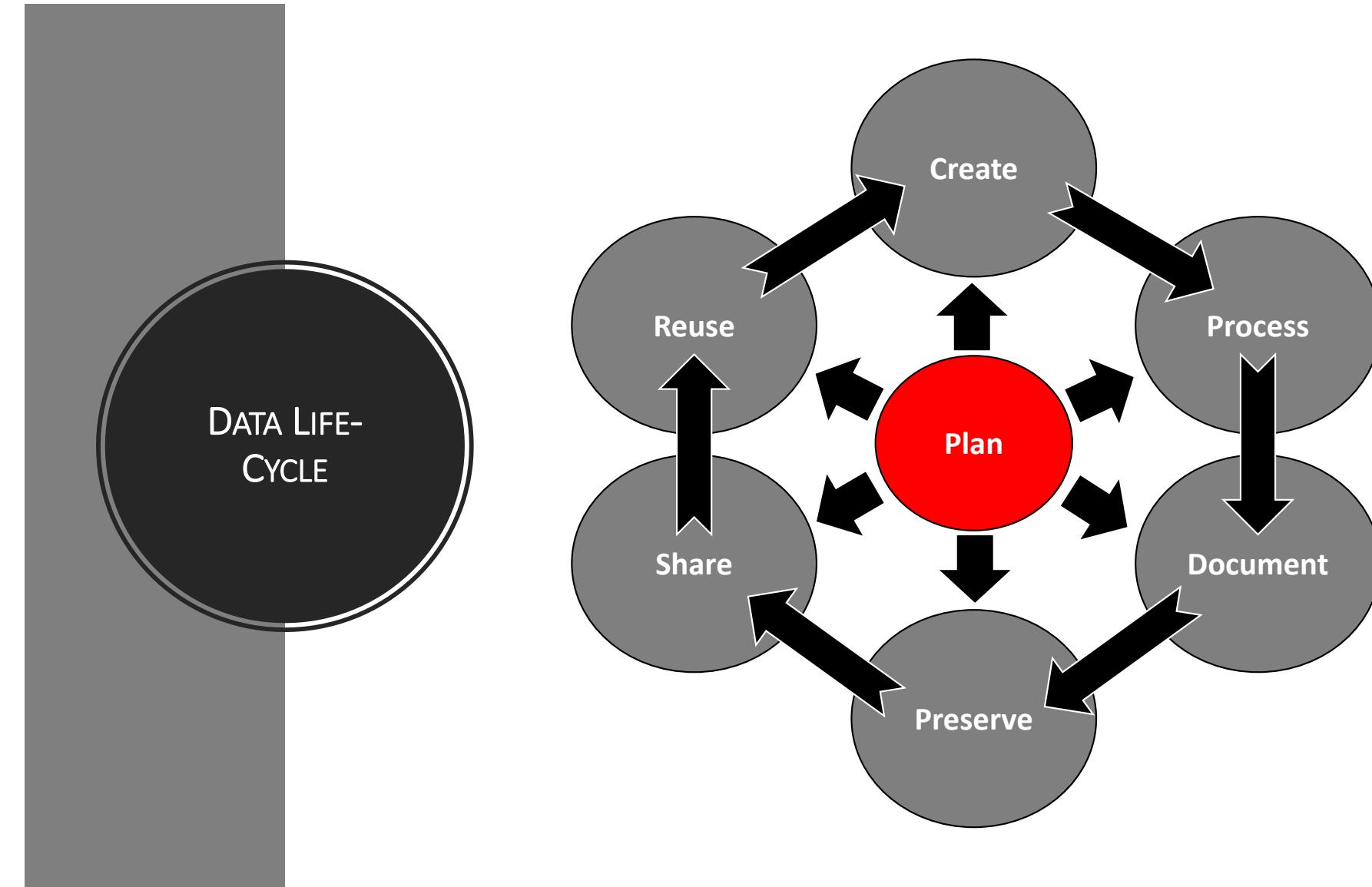


COURSE FORMAT

- This class provides a mixture of **classic lectures** together with more applied, **hands-on bioinformatic tutorials**.
- Tutorials are designed to support students in mastering theoretical genomic concepts through their implementations in bioinformatic protocols.
- Genomic field heavily relies on bioinformatic expertise, especially related to unix-based software. We will be working on computers running the **Linux operating system**. These computers will provide opportunities for students to become familiar with the **bash/shell**, **Python** and **R** computing languages.
- Lecture sessions will also serve as a platform to
 - ❖ Work on graded mini-reports.
 - ❖ Study genomic literature through a journal club.

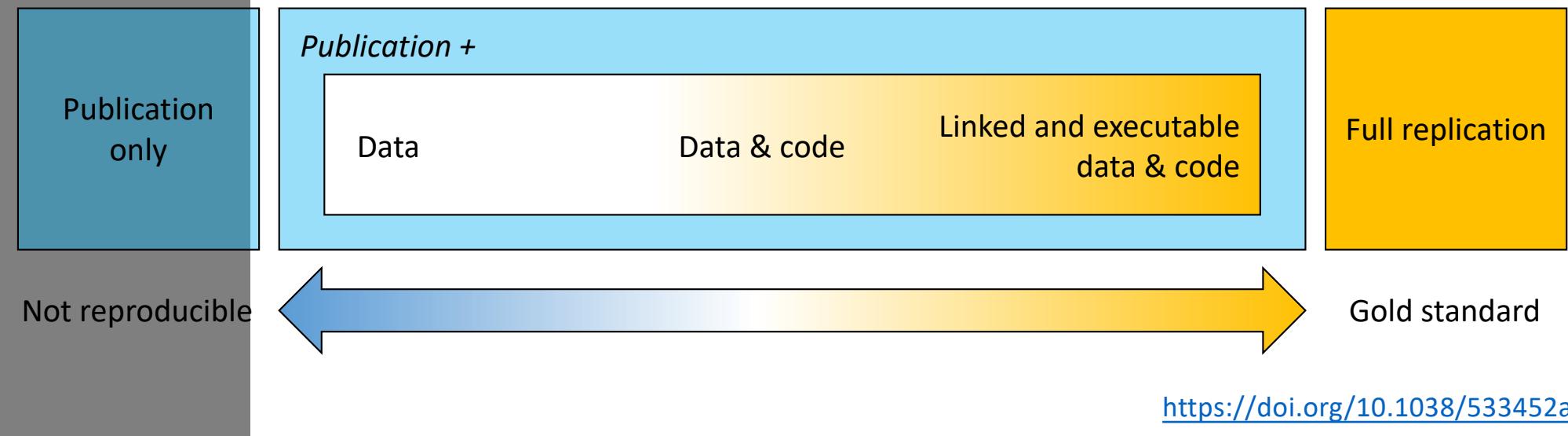
SCIENTIFIC PROCESS





REPRODUCIBLE SCIENCE

Based on a survey published in *Nature* (2016), 90% of the respondents said that **there is a reproducibility crisis in Science!**





PROMOTING
OPEN-SOURCE
RESOURCES

ubuntu 

Google

LATEX



R



python™



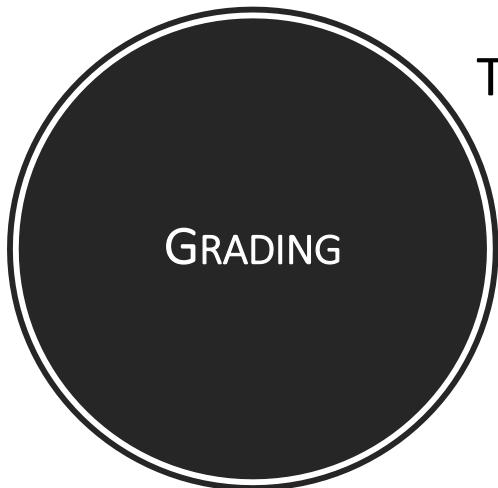
perl



GitHub



YouTube

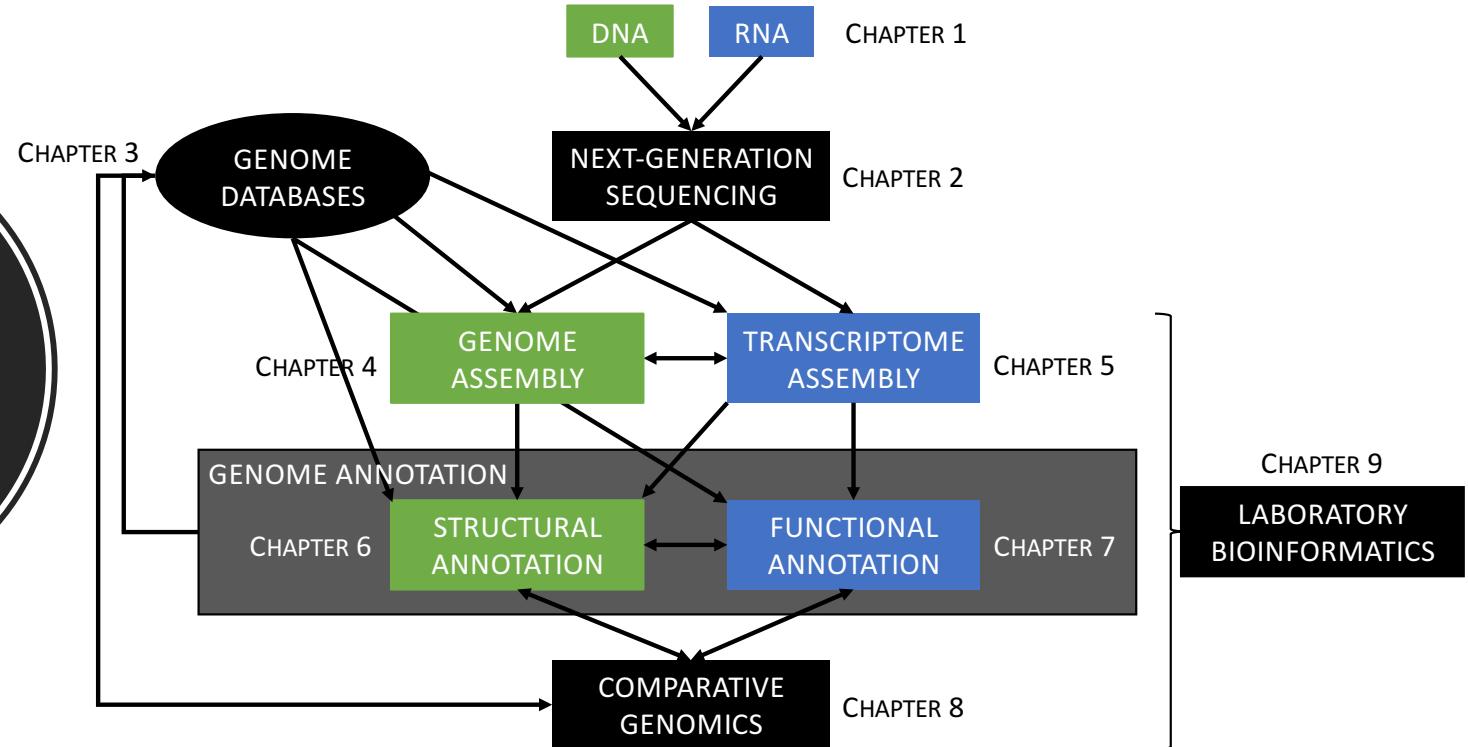


Tests conducted during this course

- 3 individual mini-reports (2x 25 points and 1x 50 points, TOTAL: 100 points).
- 1 individual oral presentation on a scientific paper (50 points).
- 1 group lab report (150 points).
- 1 group lab presentation (50 points).

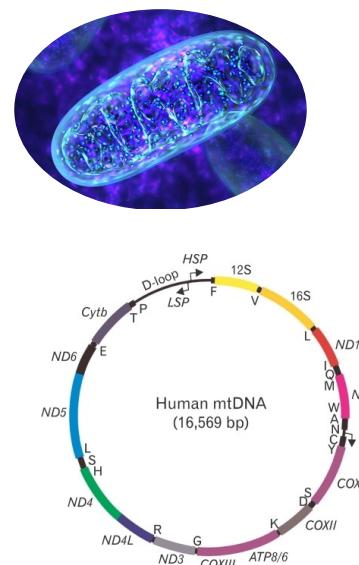
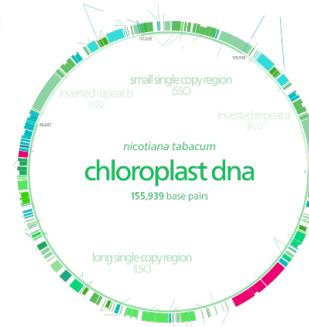
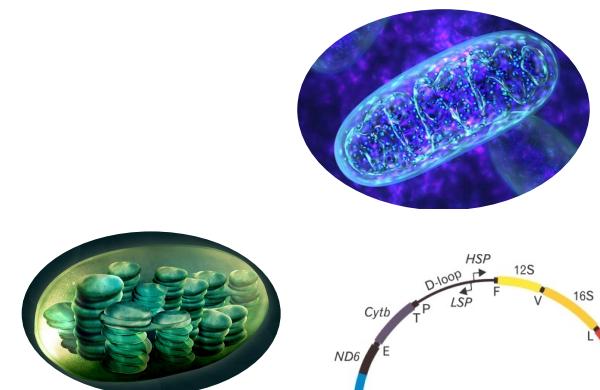
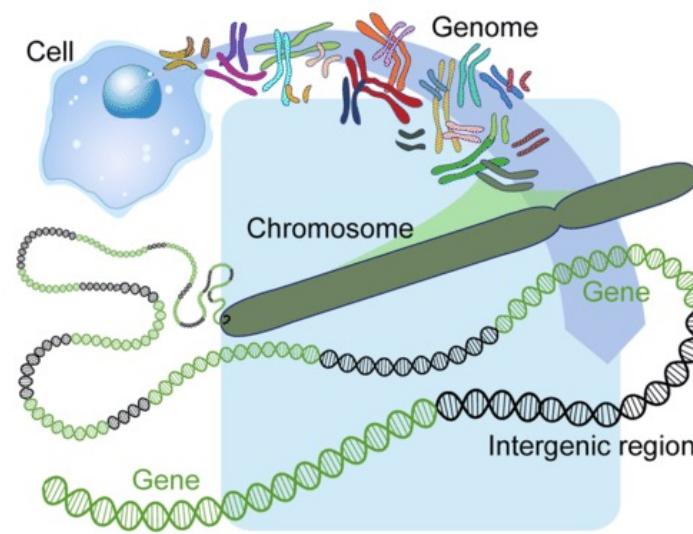
https://svenbuerki.github.io/Genomics-Bioinformatics/index.html#13_Grading_information

COURSE LEARNING OUTCOMES



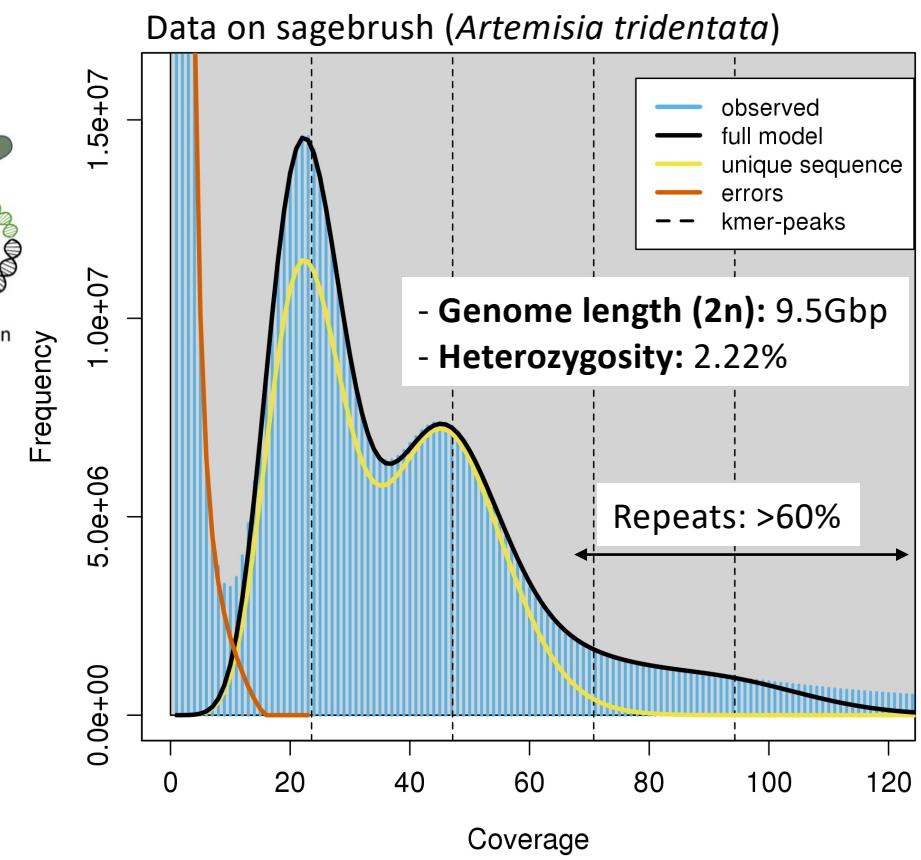
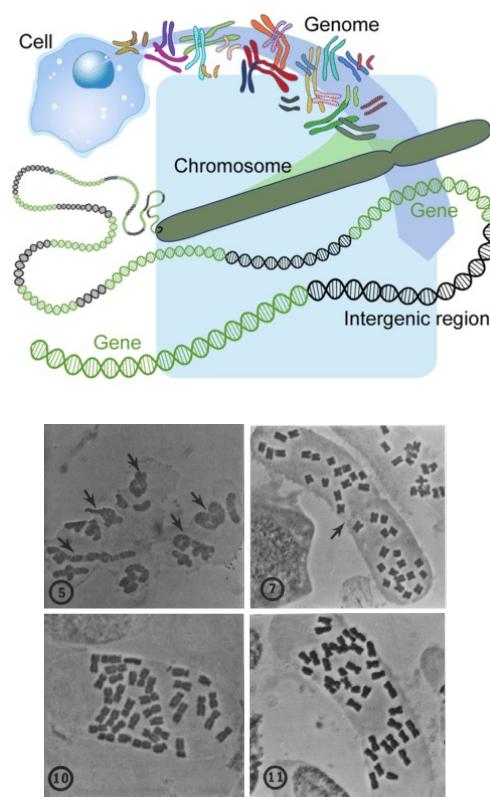
CHAPTER 1

Moving our mindset from the study of single genes (genetics) to the study of the entire genetic material in a cell (genomics).



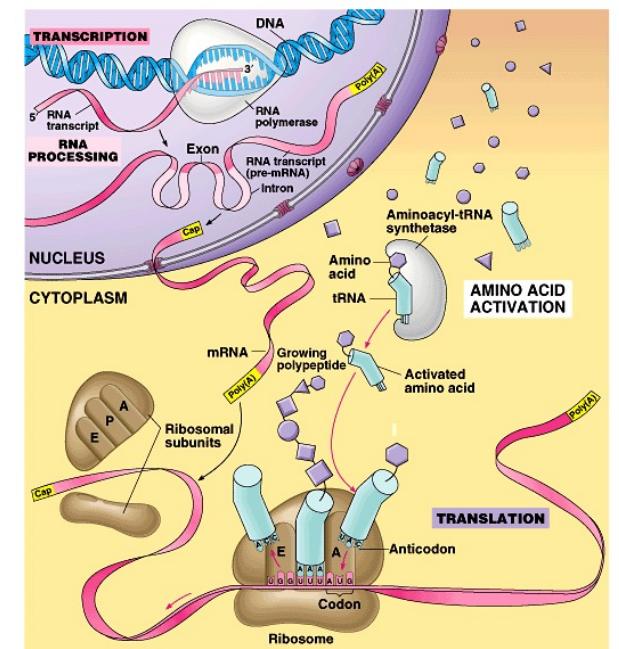
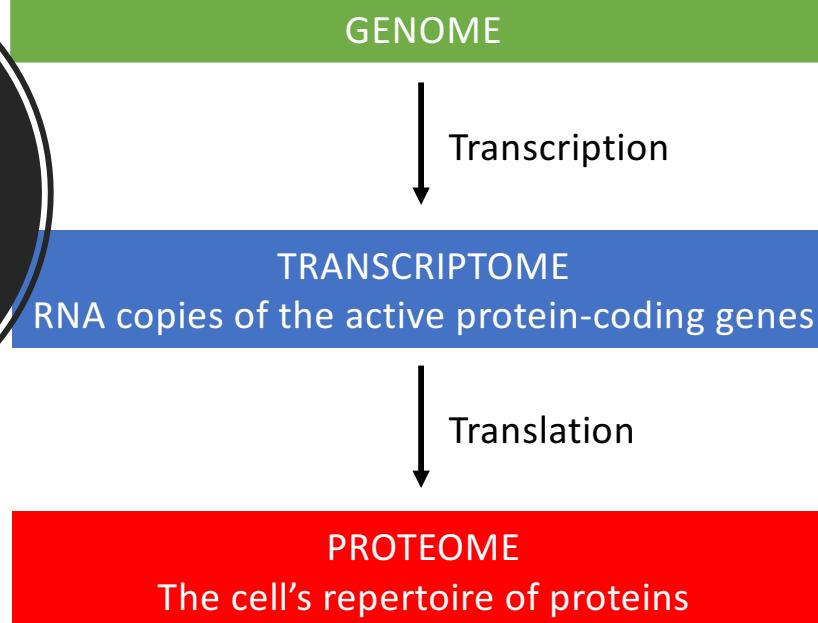
CHAPTER 1

Appreciate that **eukaryotic genomes contain extensive repetitive regions** of several different kinds. This provides a challenge for genome assembly!



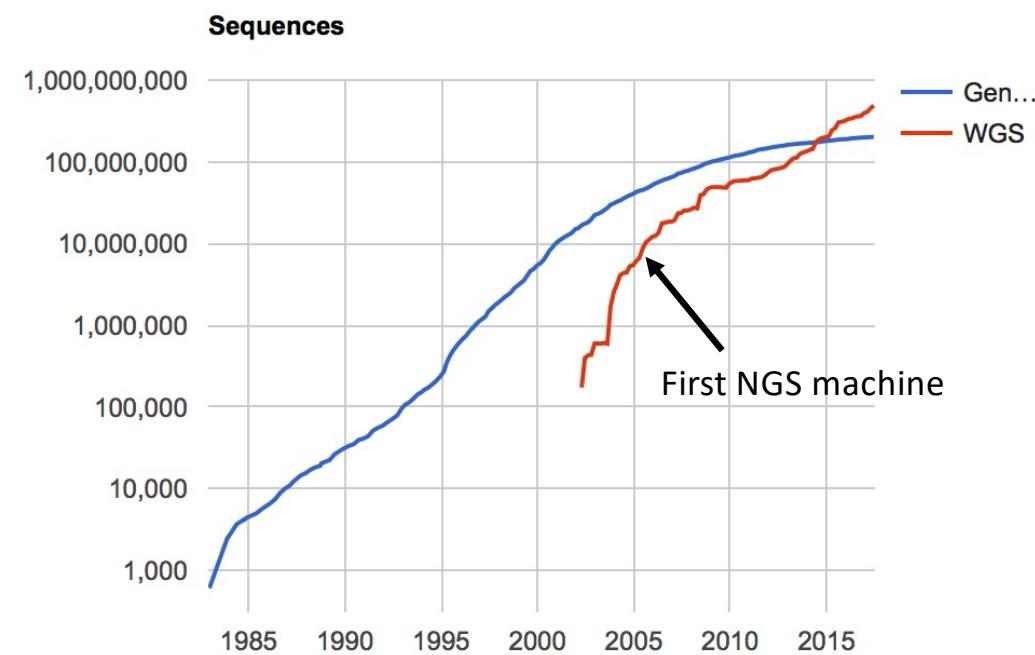
CHAPTER 1

Know the basic dogma that **DNA** is transcribed to **RNA**, which is translated to **protein**.



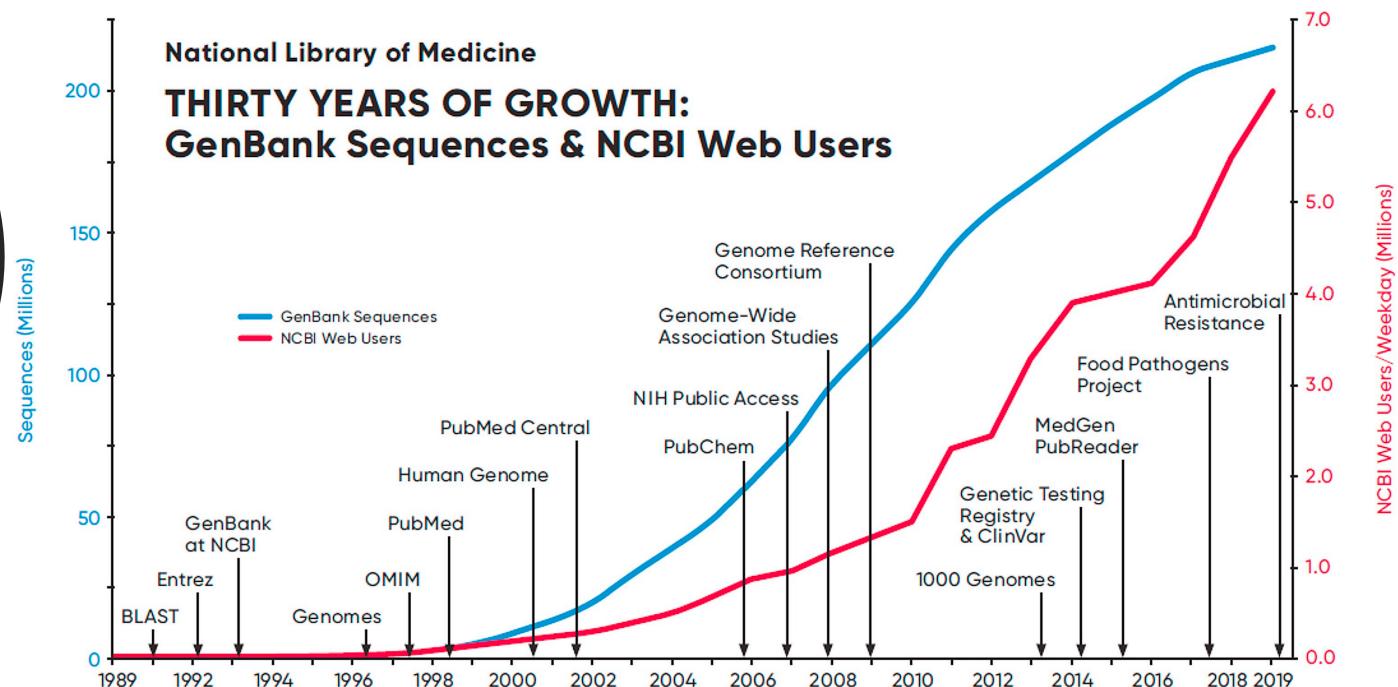
CHAPTER 1

Perfect moment to study genomics, a plethora of data have been generated in the last 15 years.



CHAPTER 1

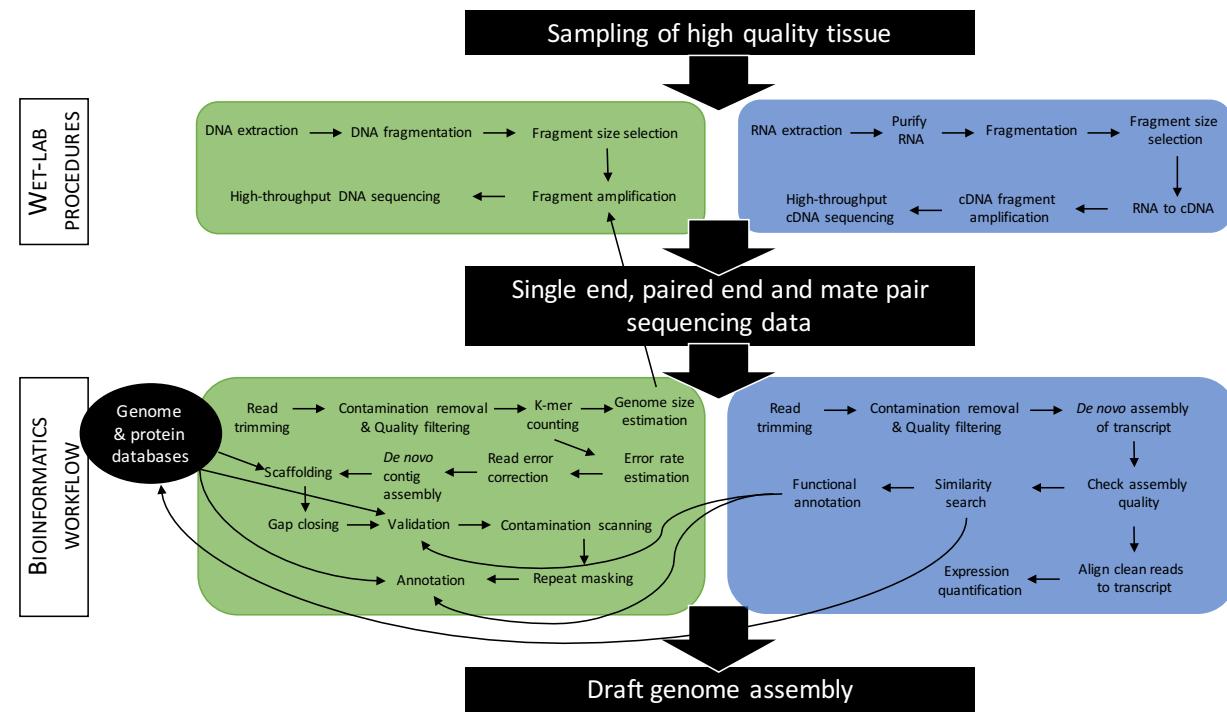
Perfect moment to study genomics, a plethora of data have been generated in the last 15 years.



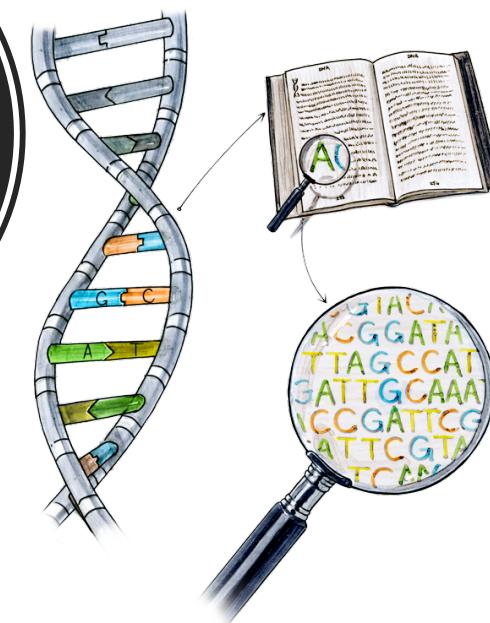
<https://doi.org/10.1016/j.gfs.2020.100411>

CHAPTER 2

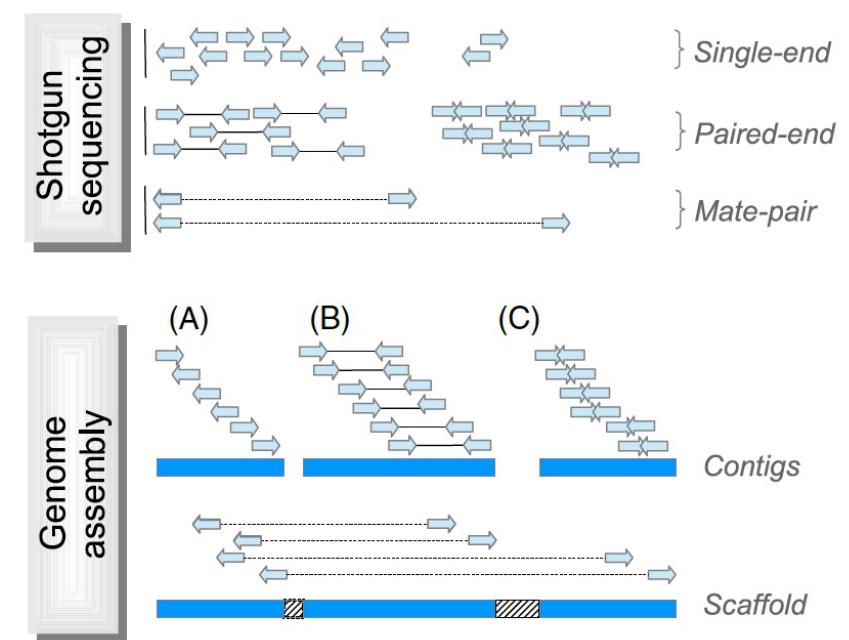
Independently on the approach to produce a whole-genome sequence, all projects share the same major steps:



CHAPTER 2



Learn the next-generation sequencing (NGS) jargon necessary to assemble and annotate genomes.

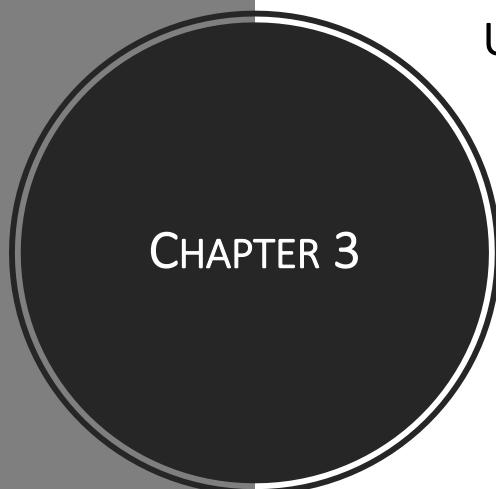
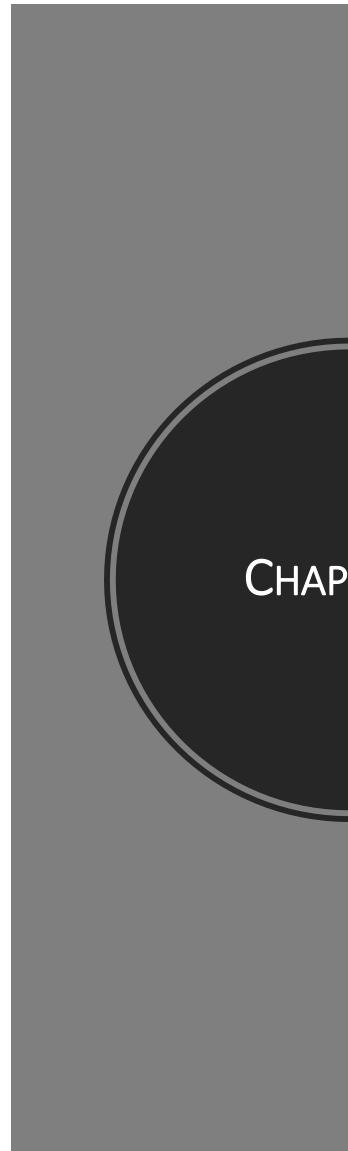


Subject of Mini-report 1

CHAPTER 2

Introduction to NGS platforms:





CHAPTER 3

Understand importance of computer science in:

- Producing raw sequence data (e.g. base-calling),
- Creating databases in molecular biology,
- Archiving and curation of data,
- Distributing data via the Internet,
- Creating information-retrieval tools to allow effective mining of the data for research application.

CHAPTER 3

Gain knowledge on major molecular biology databases, which are key to genome assembly and annotation:

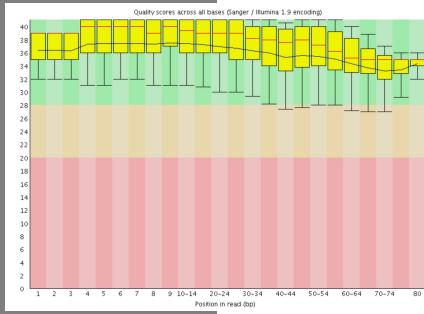
- Nucleic acid sequences databases (e.g. NCBI).
- Protein sequences databases (e.g. Swissprot).
- Gene ontology databases.
- Metabolic pathways databases (e.g. KEGG).
- Specialized annotated genome portals.



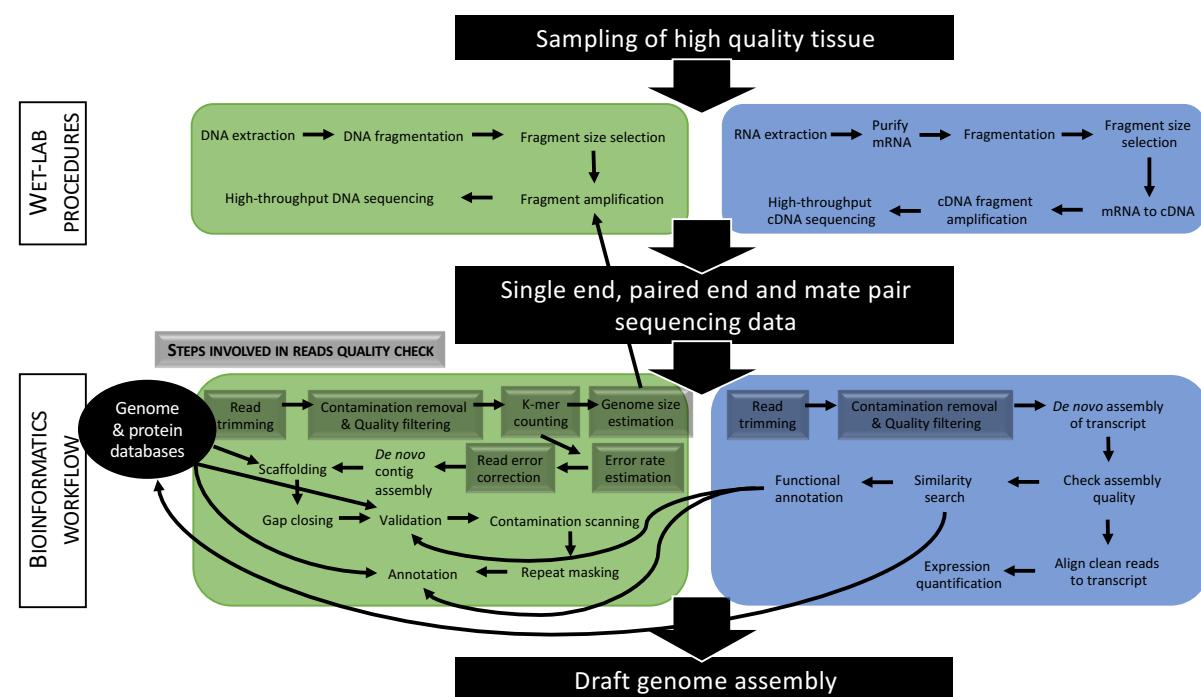
NCBI
National Center for
Biotechnology Information



CHAPTER 4

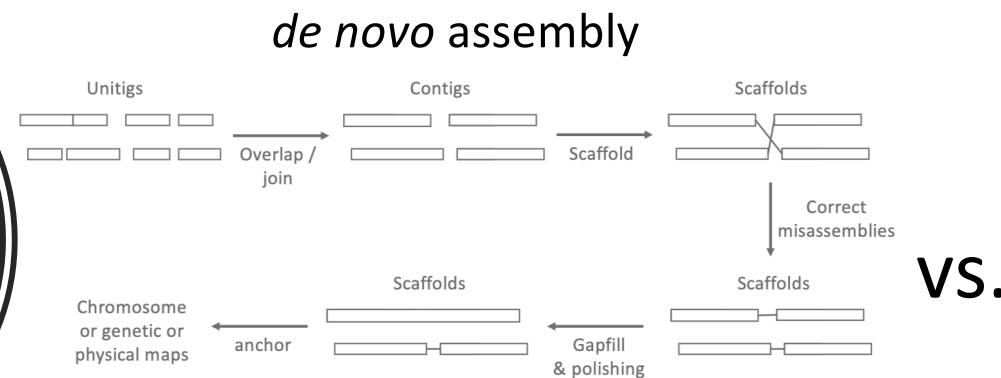


Learn how to conduct reads quality checks on raw NGS data.



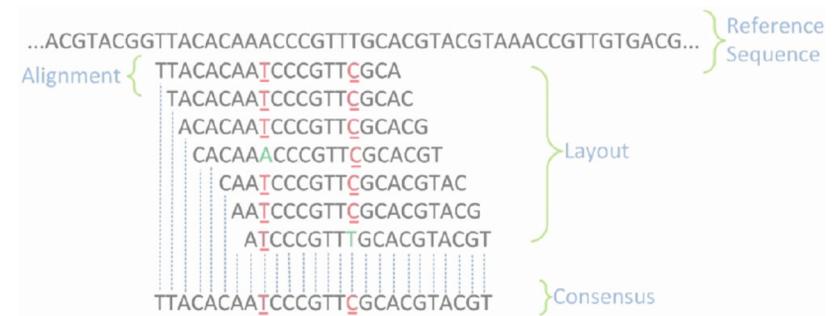
CHAPTER 4

Understand key steps involved in producing a genome assembly and best strategies to get there.



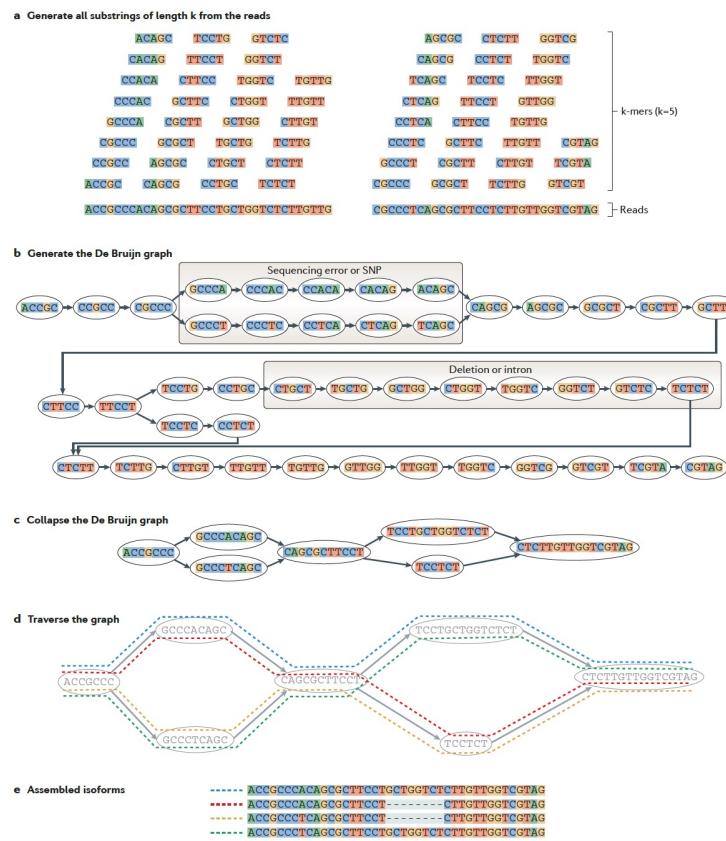
VS.

Referenced-based assembly



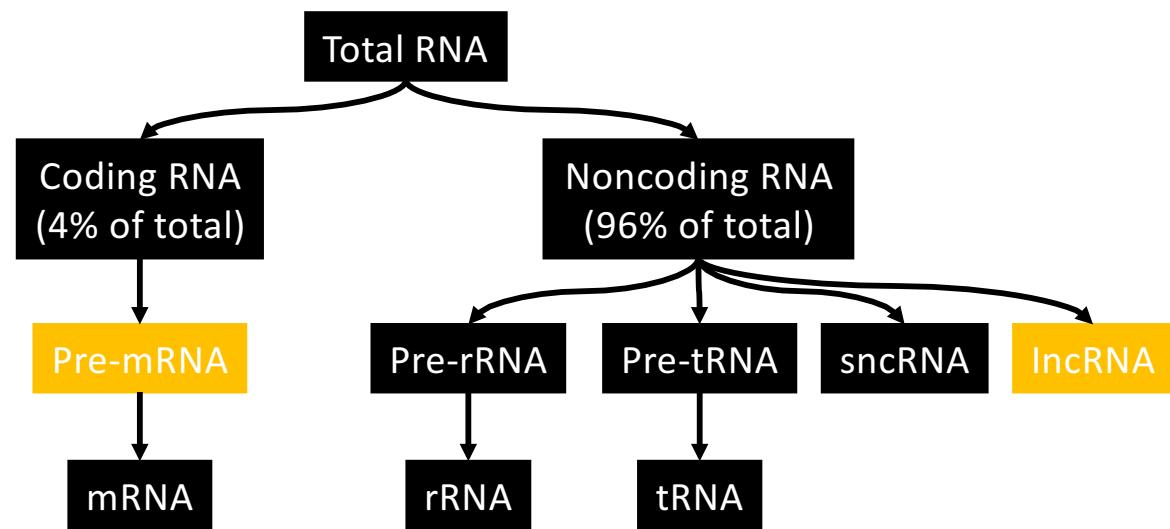
CHAPTER 4

Study de Bruijn graph procedure for *de novo* genome assembly.



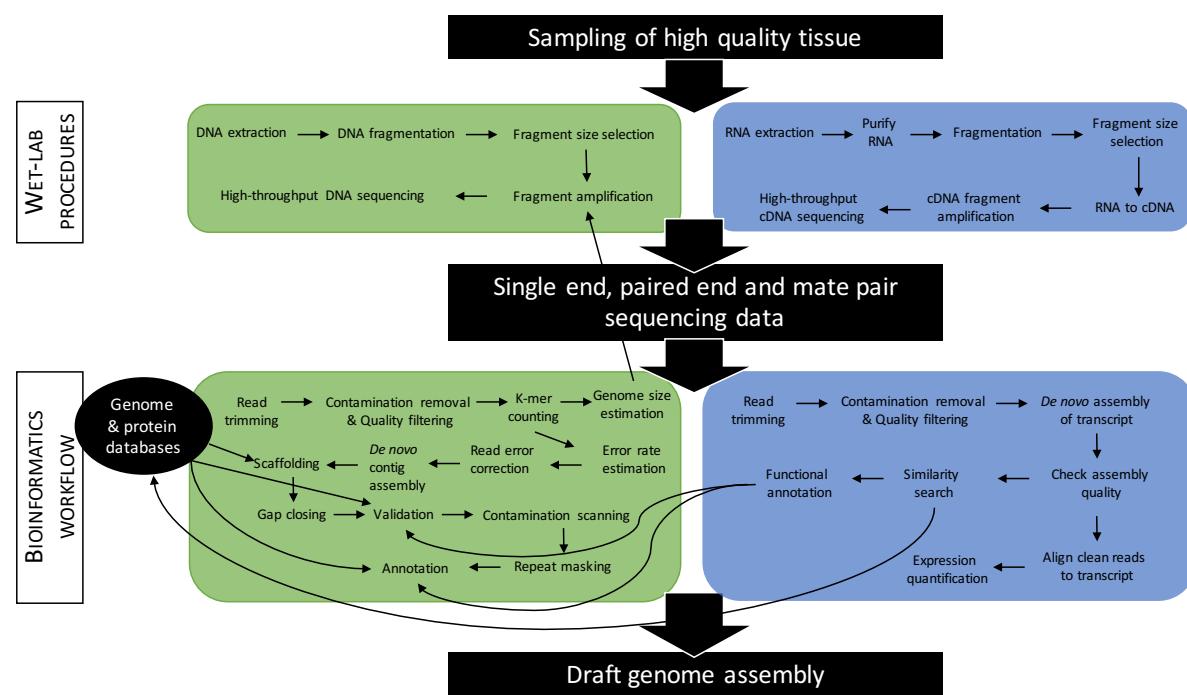
CHAPTER 5

Learn about the different types of RNA molecules in cell.



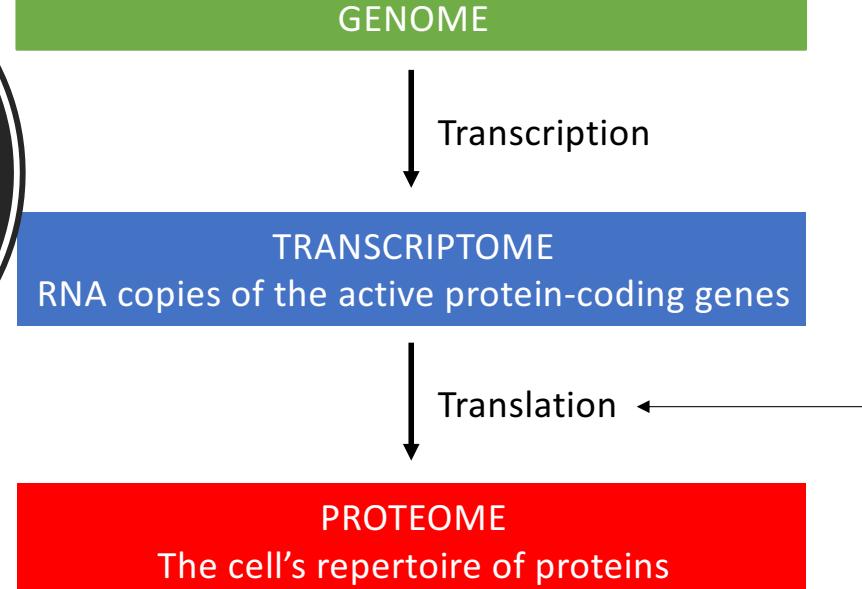
CHAPTER 5

Adapt wet-lab and bioinformatics workflow according to targeted RNAs.



CHAPTER 5

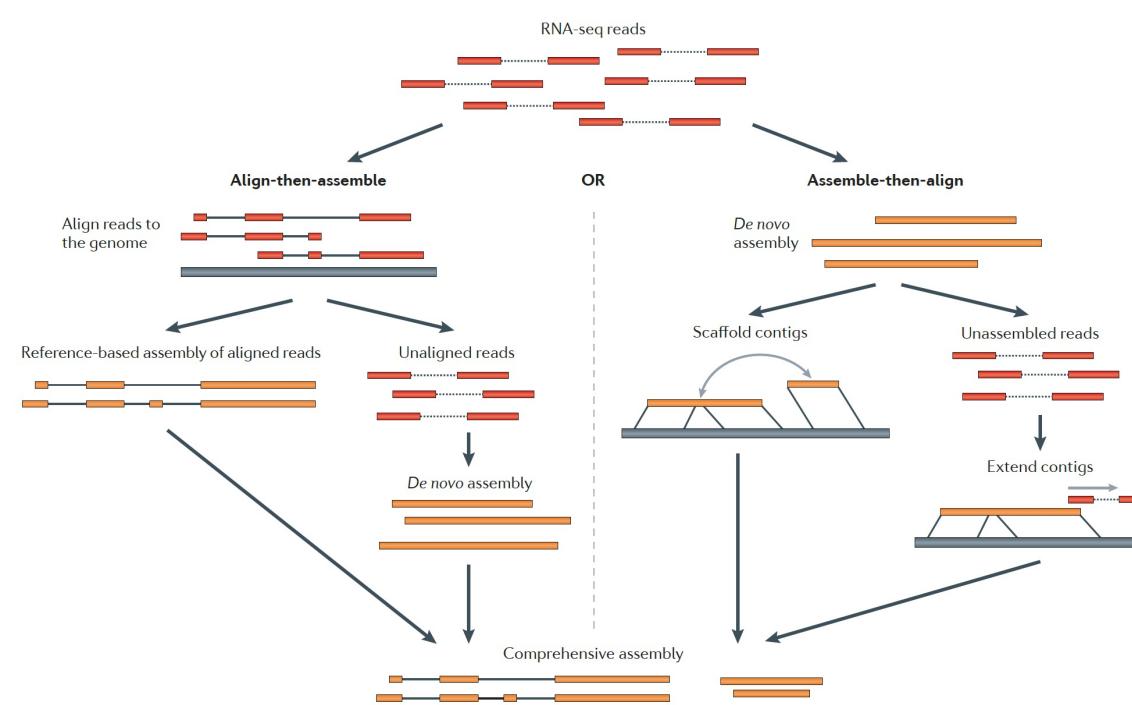
Study the link between the transcriptome and proteome via the genetic code.

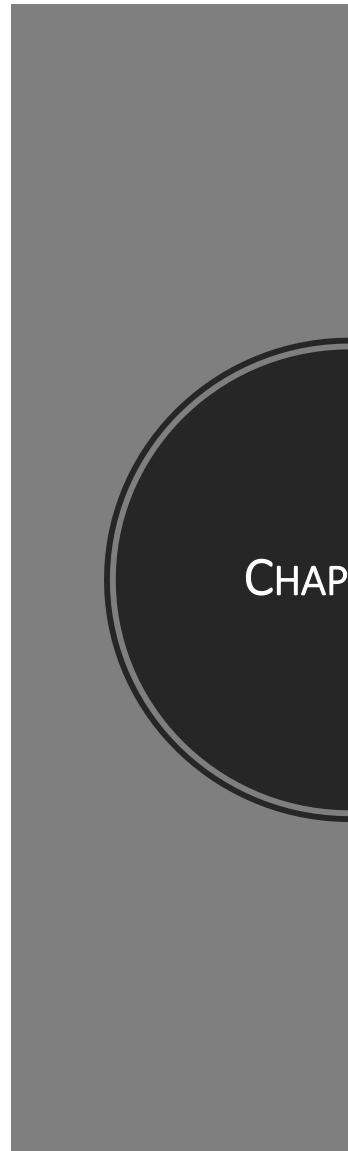


UUU	Phe	UCU	Ser	UAU	Tyr	UGU	Cys
UUC		UCC		UAC		UGC	
UUA		UCA		UAA	Stop	UGA	Stop
UUG		UCG		UAG		UGG	Trp
CUU		CCU		CAU	His	CGU	
CUC	Leu	CCC		CAC		CGC	
CUA		CCA		CAA	Gln	CGA	
CUG		CCG		CAG		CGG	
AUU		ACU		AAU	Asn	AGU	Ser
AUC	Ile	ACC		AAC		AGC	
AUA		ACA		AAA	Lys	AGA	
AUG	Met	ACG		AAG		AGG	Arg
GUU		GCU		GAU	Asp	GGU	
GUC		GCC		GAC		GGC	
GUA	Val	GCA	Ala	GAA	Glu	GGA	Gly
GUG		GCG		GAG		GGG	

CHAPTER 5

Understand key steps involved in **producing a transcriptome assembly** and best strategies to get there.





CHAPTER 6

- Learn what we exactly mean by the term "genome annotation".
- Know the following key stages of the structural genome annotation process:
 - Repeat identification.
 - Evidence alignment (map transcriptome on genome).
 - *Ab initio* ("from the beginning") gene prediction.
 - Evidence-driven gene prediction (use external info to improve prediction of gene annotations).

CHAPTER 7

Be aware of challenges to obtain accurate data on gene functions.

Research article | Open Access

Massive parallel sequencing of mRNA in identification of unannotated salinity stress-inducible transcripts in rice (*Oryza sativa* L.)

Hiroshi Mizuno [†], Yoshihiro Kawahara [†], Hiroaki Sakai, Hiroyuki Kanamori, Hironobu Wakimoto, Harumi Yamagata, Youko Oono, Jianzhong Wu, Hiroshi Ikawa, Takeshi Itoh and Takashi Matsumoto 

[†]Contributed equally

BMC Genomics 2010 11:683

<https://doi.org/10.1186/1471-2164-11-683>

Received: 20 April 2010 | Accepted: 2 Decem-

A study on the response of rice to salt stress discovered 649 genes that were missing from the rice annotation!

CHAPTER 7

Study and compare available pipelines to conduct automated genome annotations.



Last Software Update

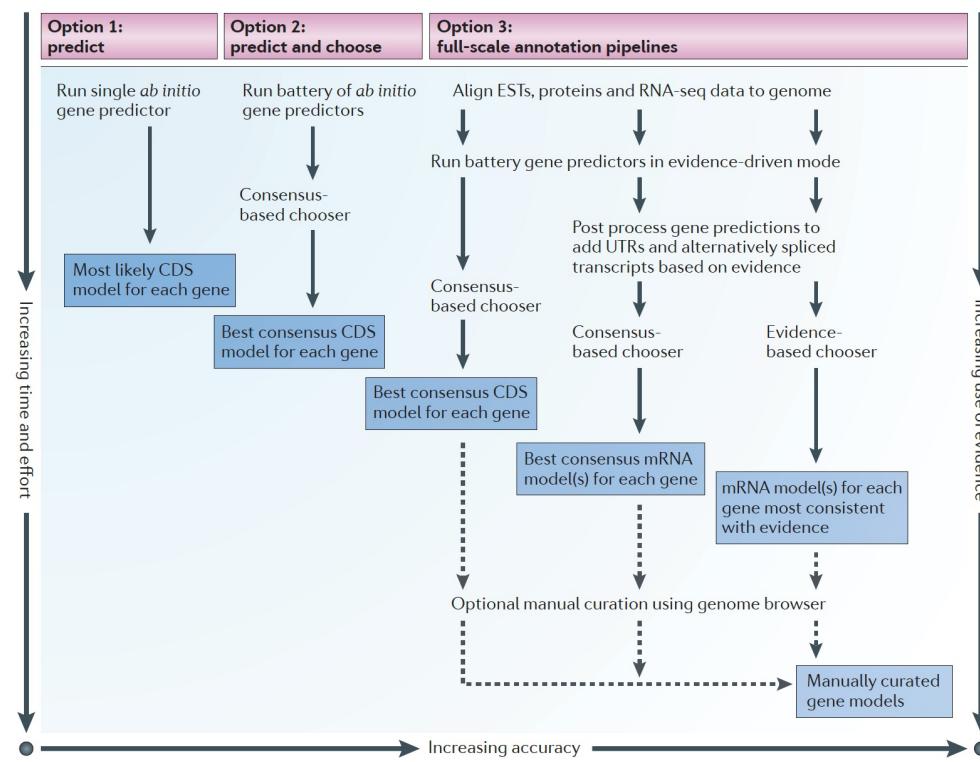
v2.31.9 (Dec 16, 2016)

Overview

MAKER is a portable and easily configurable genome annotation pipeline. Its purpose is to allow smaller eukaryotic and prokaryotic genome projects to independently annotate their genomes and to create genome databases. MAKER identifies repeats, aligns ESTs and proteins to a genome, produces ab-initio gene predictions and automatically synthesizes these data into gene annotations having evidence-based quality values. MAKER is also easily trainable: outputs of preliminary runs can be used to automatically retrain its gene prediction algorithm, producing higher quality gene-models on subsequent runs. MAKER's inputs are minimal and its outputs can be directly loaded into a GMOD database. They can also be viewed in the Apollo genome browser; this feature of MAKER provides an easy means to annotate, view and edit individual contigs and BACs without the overhead of a database. MAKER should prove especially useful for emerging model organism projects with minimal bioinformatics expertise and computer resources.

CHAPTER 7

Review approaches to assess annotation quality.



CHAPTER 8

- Learn to read and present a scientific paper reporting new genomic or transcriptomic data.
- Become an expert on a specific area of comparative genomics and share it with your peers.

