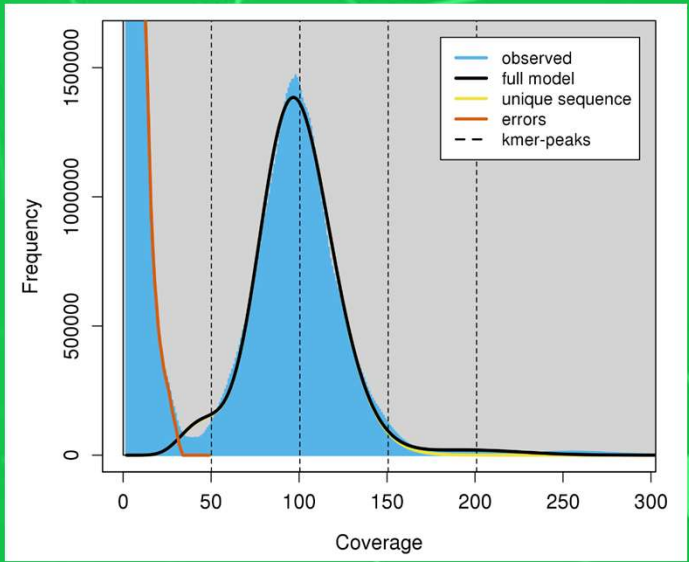
Centre for Genomic Research

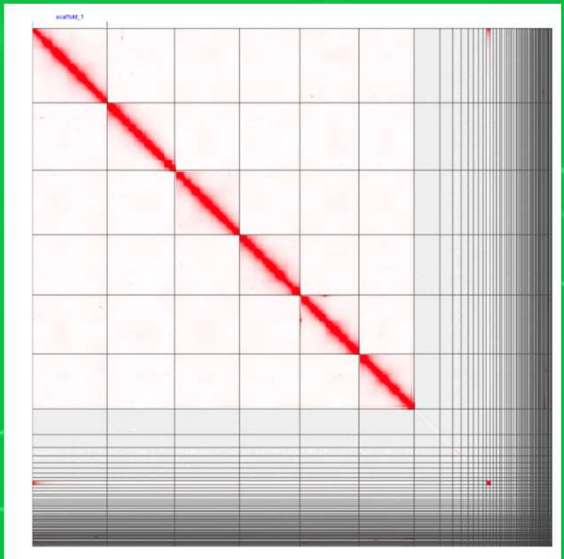
UKRI Natural Environment Research Council

NEOF NERC ENVIRONMENTAL OMICS FACILITY

wellcome sanger institute

Eukaryote Genome Assembly – day 2





UNIVERSITY OF LIVERPOOL

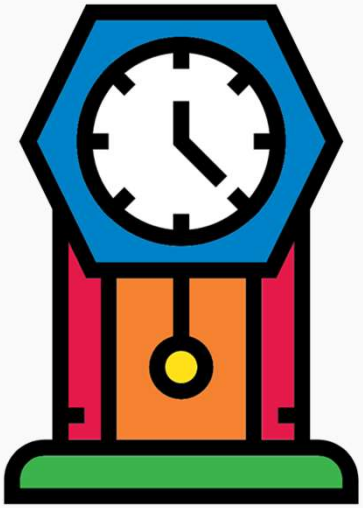
The University Of Sheffield.

Format & Schedule

This intro
Bookdown
Theory
Practice
Exercises
Optional materials

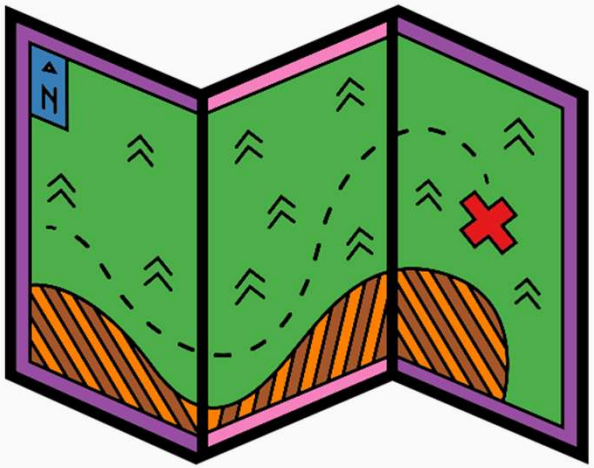
Work at your own pace
We are here to help
Time with breaks in between

- 10:00-11:15
- 11:30-12:30
- 13:30-14:45
- 15:00-16:00

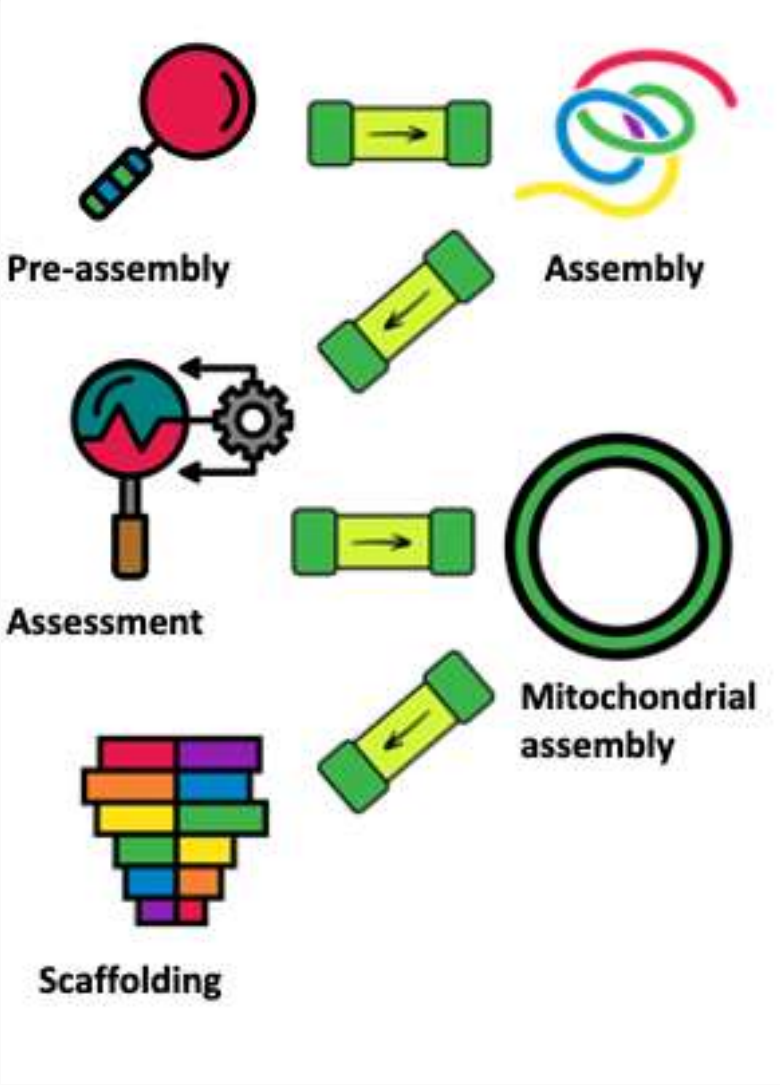


Outline for Today

- Mitochondrial genome assembly
- Scaffolding

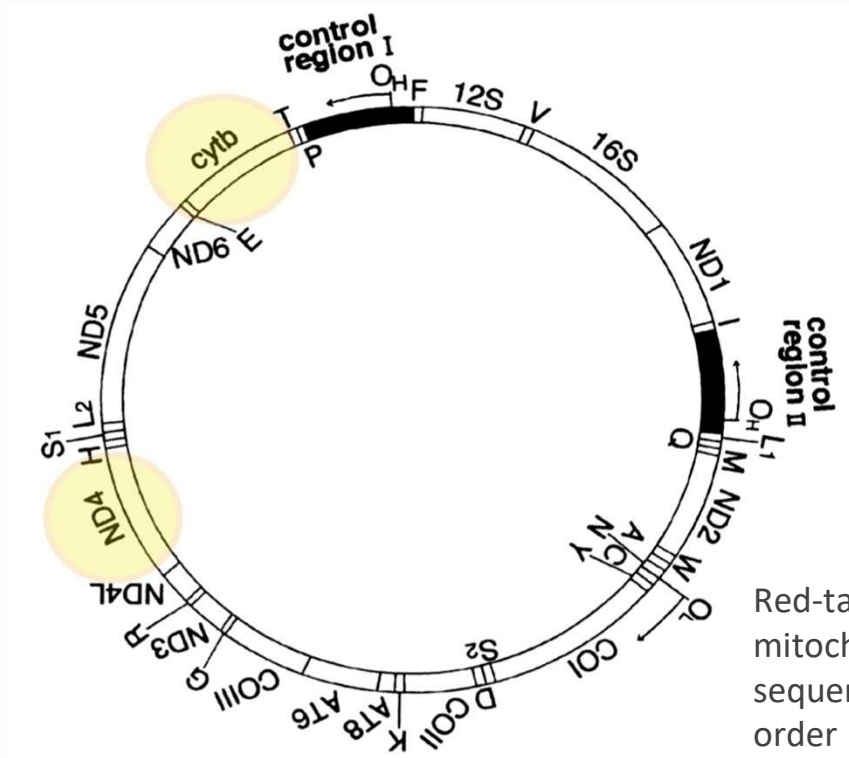


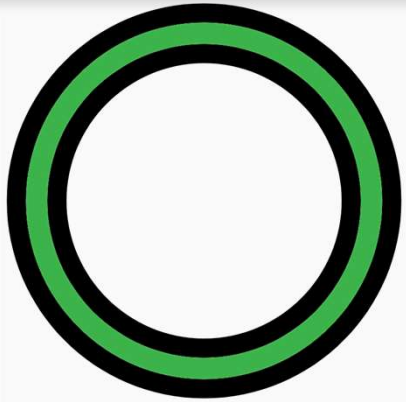
Genome assembly workflow



Mitochondrial genome assembly

- Circular genome
- Key genes for respiration
- Haploid
- Usually maternally inherited
- ‘By-product’ of WGS
- knowledge of gene content & structure

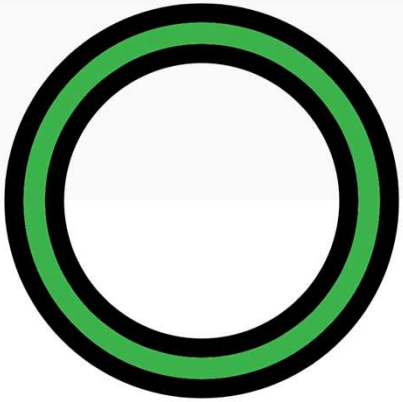




Red-tailed boa, *Boa constrictor,* mitochondrial DNA (mtDNA) sequence, showing the typical order of mtDNA genes found in snakes (Dong & Kumazawa, 2005).

Mitochondrial genome assembly

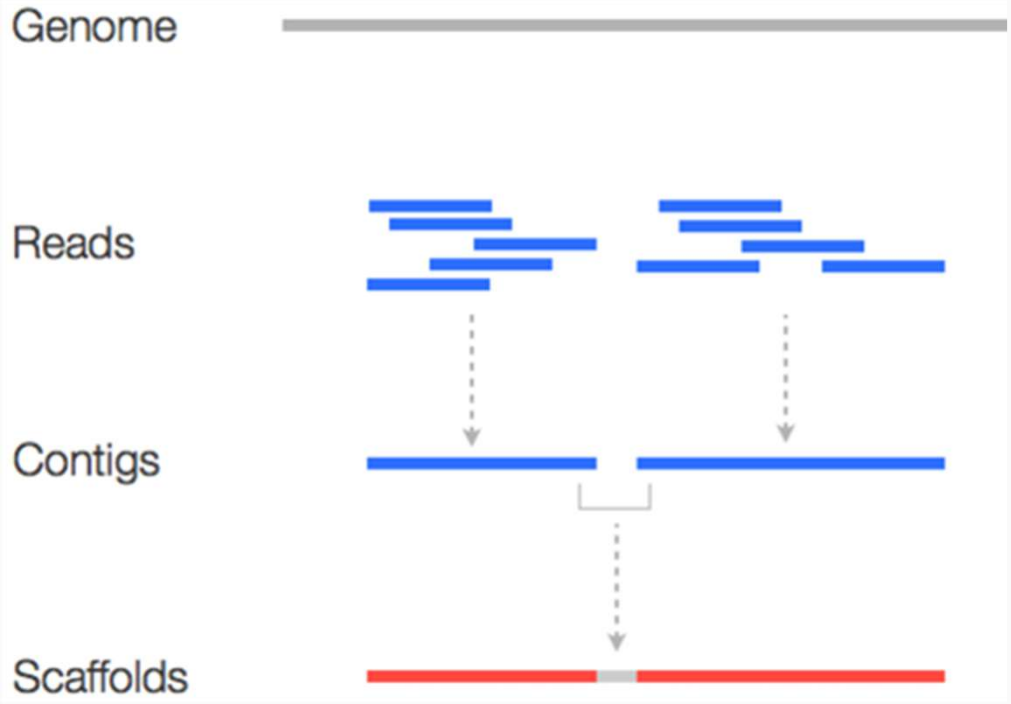
- **MitoHiFi**
- Assembles mitogenomes from PacBio HiFi reads
- DToL team
- Separates NUMTs (mito DNA in nuclear genome)
- Circularised & annotated mitogenome



DToL: https://portal.darwintreeoflife.org/
MitoHiFi https://doi.org/10.1101/2022.12.23.521667
MitoFinder https://doi.org/10.1111/1755-0998.13160
ARWEN https://doi.org/10.1093/bioinformatics/btm573

Genome assembly - scaffolding

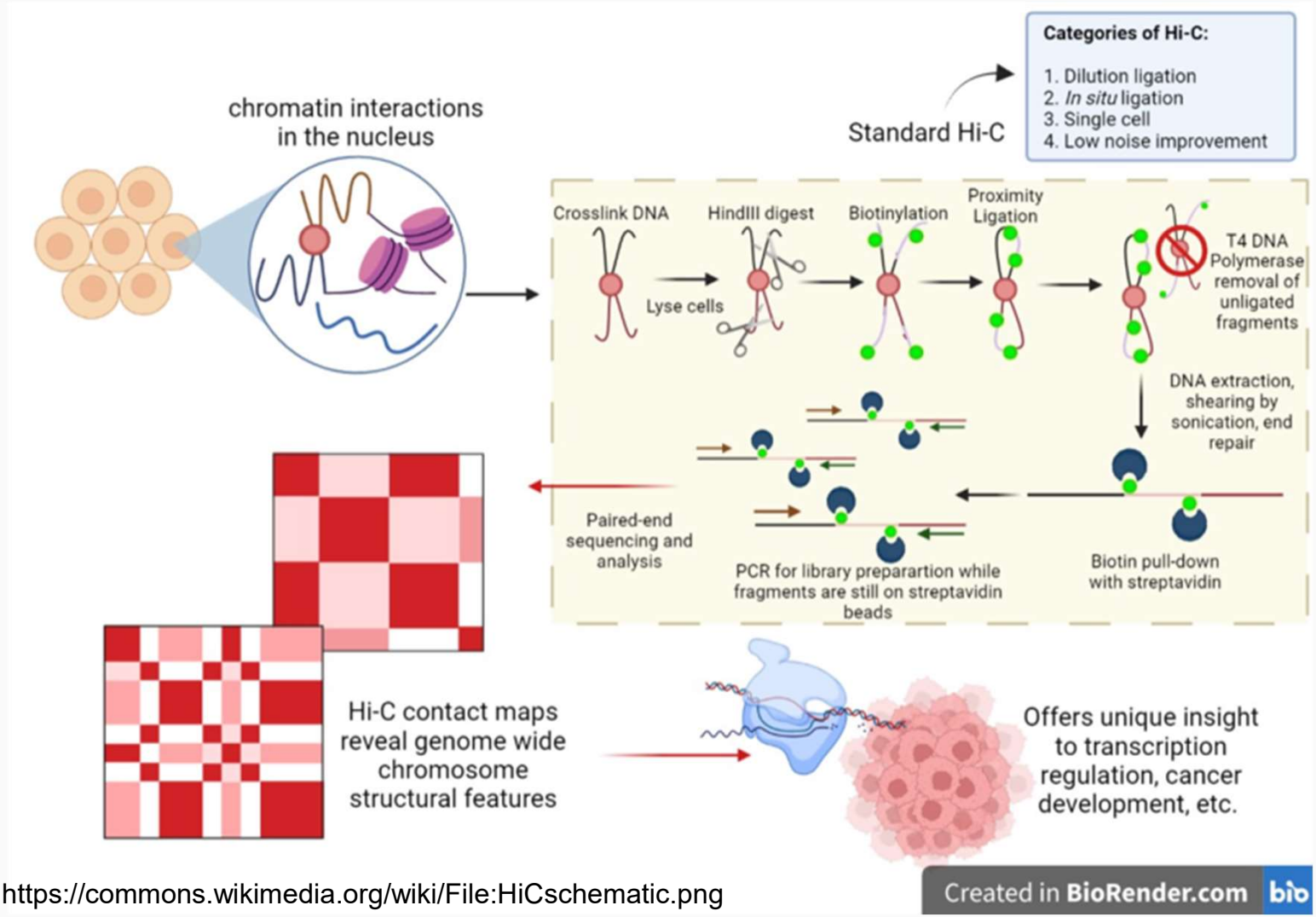
Assembling reads into contigs
Contig = contiguous sequence
Perfect contig = chromosome/plasmid/etc



PacBio HiFi & Hifiasm

Illumina Hi-C & yahs

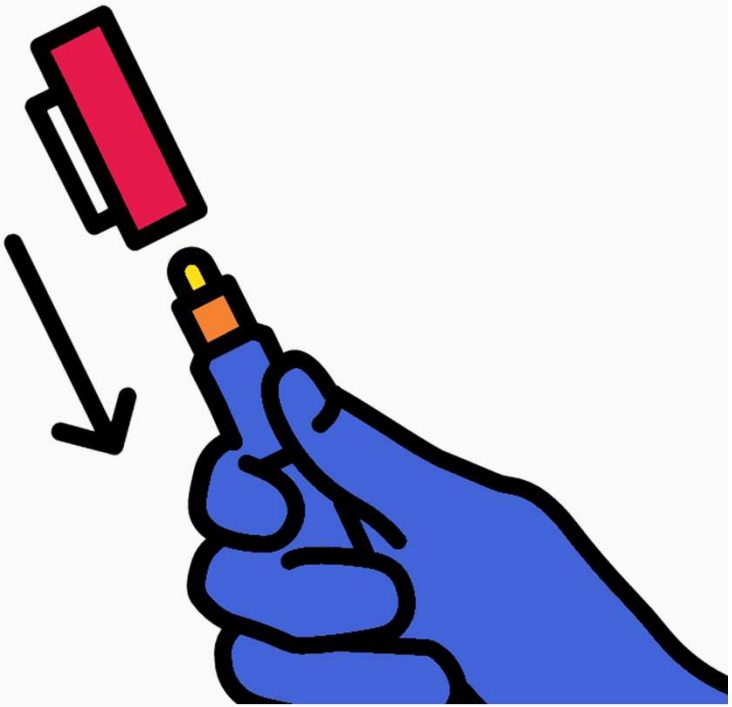
Hi-C library preparation

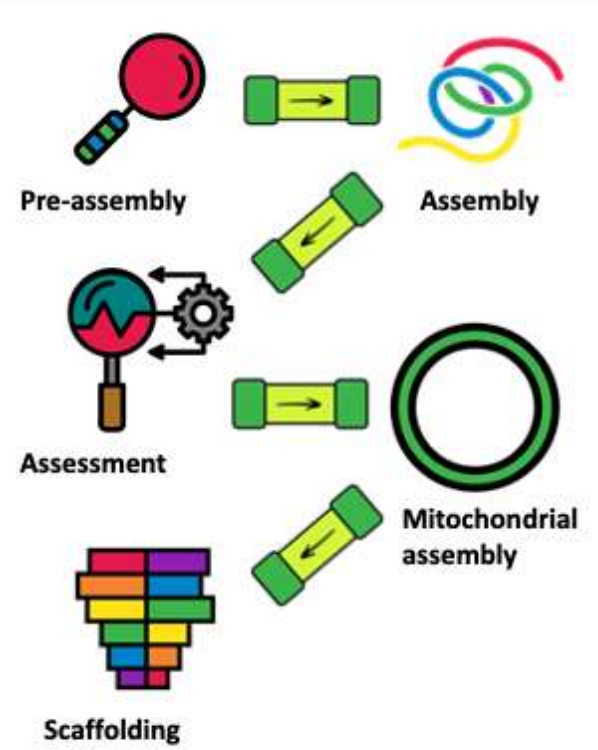


https://commons.wikimedia.org/wiki/File:HiCschematic.png

Recap

- Mitochondrial assembly
  - MitoHiFi
- Scaffolding
  - Hi-C & yahs





Reminders and Tips

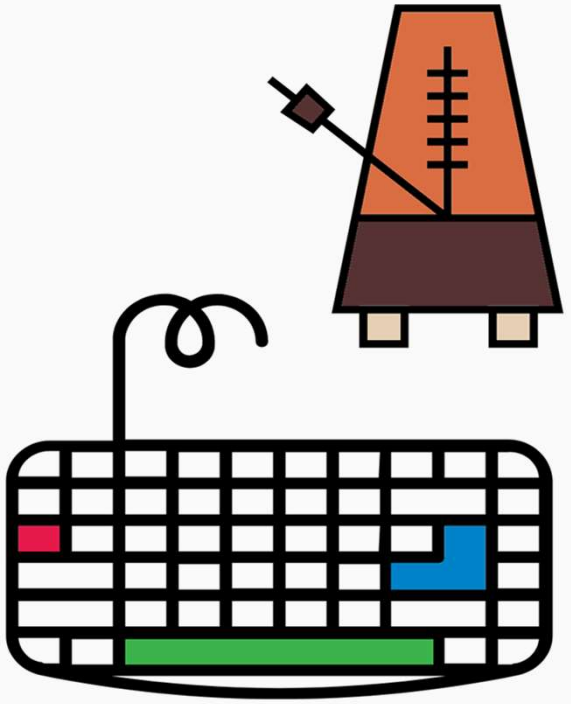
Work at your own pace

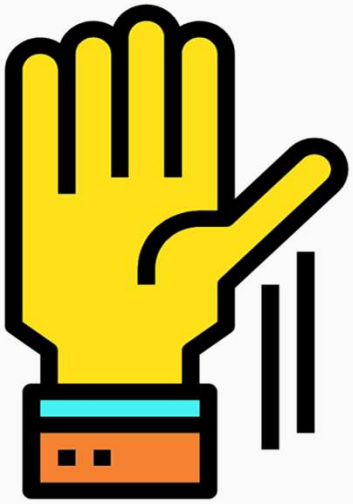
Typos

Ask questions

Breaks are important

Tab, space, and enter





During sessions

Zoom - Ask via microphone if no question currently being asked/answered

Slack - Ask questions via the channel or ask to go into a zoom breakout room with one of us

WebVNC - We can connect to your webVNC to see and help with issues.

Breakout rooms upon request

Thank you!

Questions?