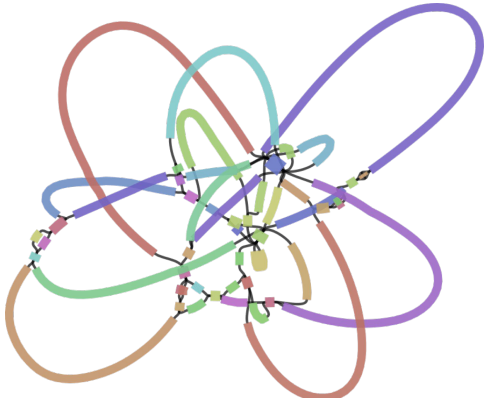
Genome assembly: where do I start?

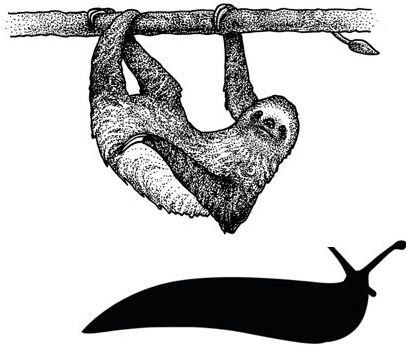
And where do I go once I have contigs and scaffolds

Marcela Uliano-Silva

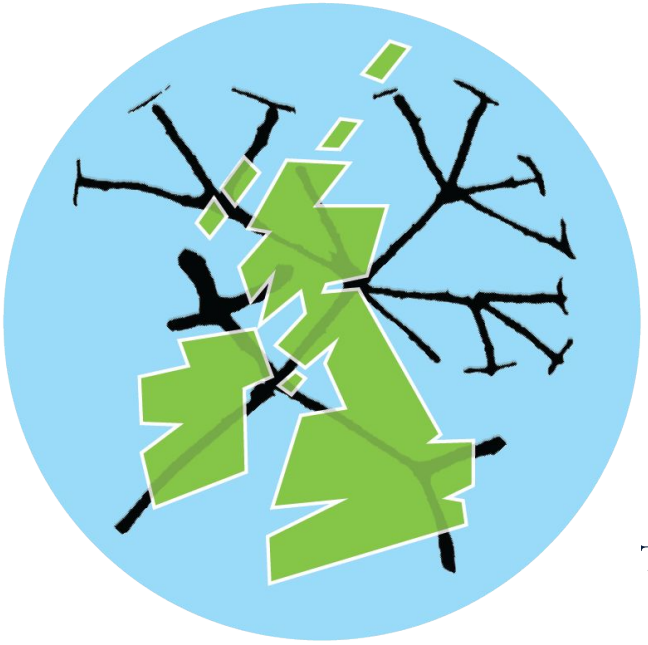


Who am I?

- Senior Bioinformatician - Tree of Life, Wellcome Sanger Institute, Cambridge, UK
- Associate Professor (Prof II) - Nord University, Bodø, Norway
- **Churchill College Teaching Fellow - University of Cambridge**



- Horizon2020 Marie Curie PostDoc Fellow (2017-2019), IZW, BenGenDiv, Germany
- PhD in Biophysics (2017) - IBCCF UFRJ, Brazil
- MSc in Biophysics (2013) - IBCCF UFRJ, Brazil
- BSc in Biology (2010) - UFSC, Brazil
- TED Fellow



wellcome sanger institute

Royal Botanic Gardens Kew

Earlham Institute

Royal Botanic Garden Edinburgh

THE UNIVERSITY of EDINBURGH

EMBL-EBI

UNIVERSITY OF CAMBRIDGE

NATURAL HISTORY MUSEUM

wellcome

UCD DUBLIN

NatureScot NàdarAlba
Scotland's Nature Agency
Buidheann Nàdair na h-Alba

Wytham WOODS
the natural place for science

UNIVERSITY OF OXFORD

CABI

ZSL

THE MARINE BIOLOGICAL ASSOCIATION

National Museums Scotland

My two main areas of research

Software development for high-quality genome assembly

Software | Open Access | Published: 18 July 2023

MitoHiFi: a python pipeline for mitochondrial genome assembly from PacBio high fidelity reads

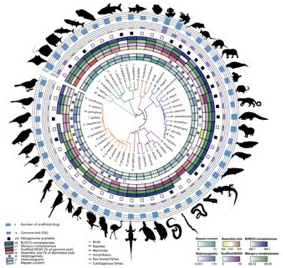
Marcela Uliano-Silva, João Gabriel R. N. Ferreira, Ksenia Krasheninnikova, Darwin Tree of Life Consortium, Giulio Formenti, Linelle Abueg, James Torrance, Eugene W. Myers, Richard Durbin, Mark Blaxter & Shane A. McCarthy

BMC Bioinformatics **24**, Article number: 288 (2023) | Cite this article

Scalable, accessible and reproducible reference genome assembly and evaluation in Galaxy

Delphine Larivière, Linelle Abueg, Nadolina Brajuka, Cristóbal Gallardo-Alba, Bjorn Grüning, Byung June Ko, Alex Ostrovsky, Marc Palmada-Flores, Brandon D. Pickett, Keon Rabbani, Agostinho Antunes, Jennifer R. Balacco, Mark J. P. Chaisson, Haoyu Cheng, Joanna Collins, Melanie Couture, Alexandra Denisova, Olivier Fedrigo, Guido Roberto Gallo, Alice Maria Giani, Grenville MacDonald Gooder, Kathleen Horan, Nivesh Jain, Cassidy Johnson, Heebal Kim, Chul Lee, Tomas Marques-Bonet, Bria Arang Rhie, Simona Secomandi, Marcella Sozzoni, Tatiana Tilley, Marcela Uliano-Silva, M Beek, Robert W. Williams, Robert M. Waterhouse, Adam M. Phillippy, Erich D. Jarvis, N Schatz, Anton Nekrutenko & Giulio Formenti — Show fewer authors

Nature Biotechnology **42**, 367–370 (2024) | Cite this article



Comparative genomics: origins (ancestral linkage groups) and diversification of phenotypes

Caecilian Genomes Reveal the Molecular Basis of Adaptation and Convergent Evolution of Limblessness in Snakes and Caecilians

Vladimir Ovchinnikov,[1,†] Marcela Uliano-Silva,[2,†] Mark Wilkinson,[3] Jonathan Wood,[2] Michelle Smith,[4] Karen Oliver,[4] Ying Sims,[2] James Torrance,[2] Alexander Suh,[5,6] Shane A. McCarthy,[2,7] Richard Durbin,[2,7,*] and Mary J. O'Connell[1,*]

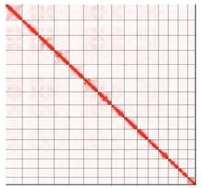
JOURNAL ARTICLE

A chromosome-level assembly supports genome-wide investigation of the DMRT gene family in the golden mussel (*Limnoperna fortunei*)

João Gabriel R. N. Ferreira, Juliana A. Americo, Danielle L. A. S. do Amaral, Fábio Sendim, Yasmin R. da Cunha, Tree of Life Programme, Mark Blaxter, Marcela Uliano-Silva, Mauro de F. Rebelo Author Notes

GigaScience, Volume 12, 2023, giad072, https://doi.org/10.1093/gigascience/giad072

Published: 30 September 2023 **Article history**



Darwin
TREE of LIFE

Tree of Life: Major Projects

★ **Darwin Tree of Life Project**
- 70,000 species from Britain and Ireland
  Phase 1: ~8,000 species by 2026

★ **AEGIS**
- Ancient Environmental Genomics Initiative for Sustainability
  1,300 genomes

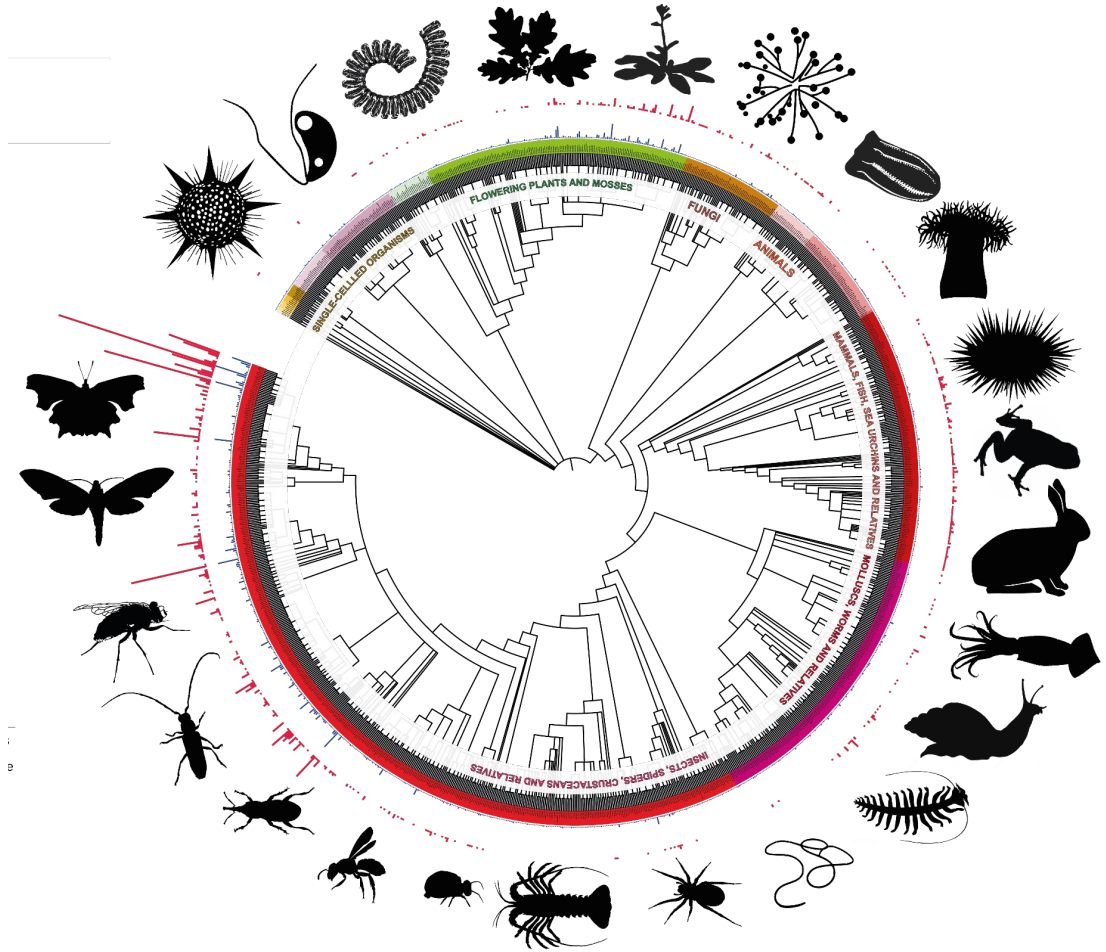
★ **Aquatic Symbiosis Genomics**
- Genomics of marine and freshwater symbioses
  500 symbiotic systems

★ **PSYCHE**
- Reference genomes for all Lepidoptera of Europe
  Phase1: 2000 genomes

★ **Vertebrate Genomes Project**
- VGP Phase 1 (ordinal) and Phase 2 (family)

★ **European Reference Genome Atlas**
- Sequencing the all species in the European continent
  Initial phase: ~200 genomes

★ **Earth BioGenome Project**
- Working to deliver Phase 1 (family) goals, and to
  "***sequence all life for the future of life***"



In December 2025, the Tree of Life programme released

3,571

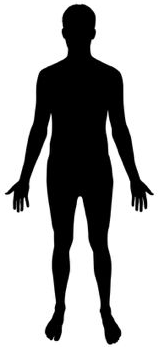
reference genome assemblies to INSDC

Genomes assembled from

- 550.3 Tb PacBio HiFi
- 2108.3 Tb Hi-C
- 80.8 Tb RNA-Seq (3341 species)

Genome assembly: what is my goal?

- Understand variation in populations (disease-related SNPs etc…)



Genome re-sequencing
Assembly by mapping to a reference

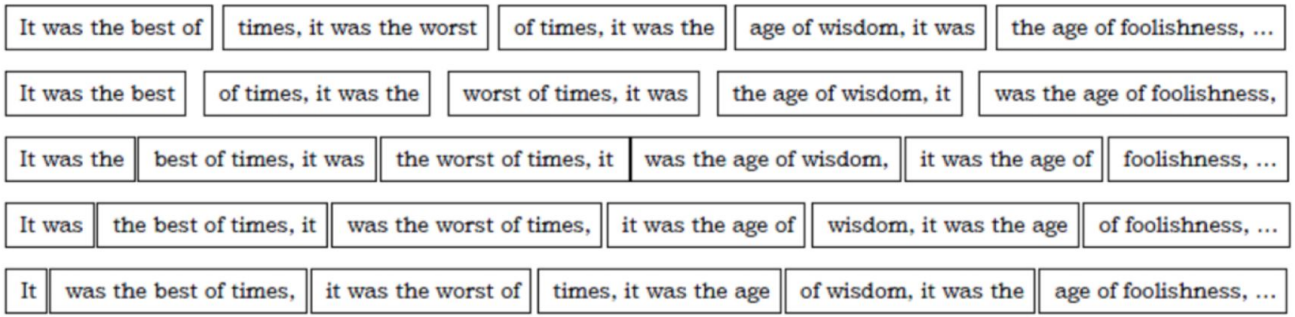
- Study the molecular profile of a species never before sequenced (evolutionary studies etc..)

De novo assembly

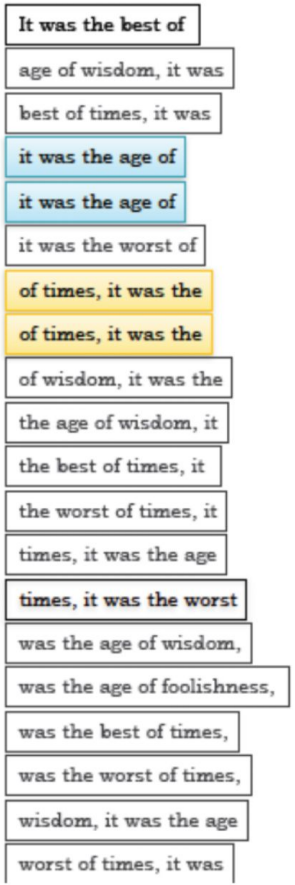
Genome assembly

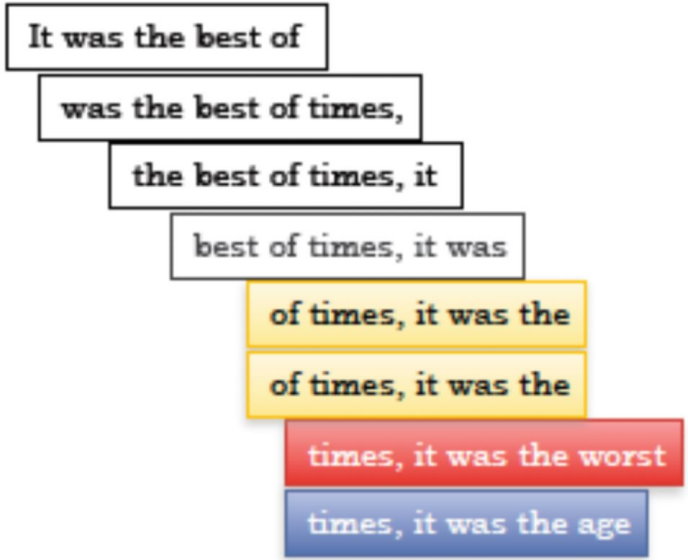
Let's try to reconstruct the sentence bellow (our genome) from some fragments (reads):

- *It was the best of times, it was the worst of times, it was the age of wisdom, it was the age of foolishness, …*



Genome assembly



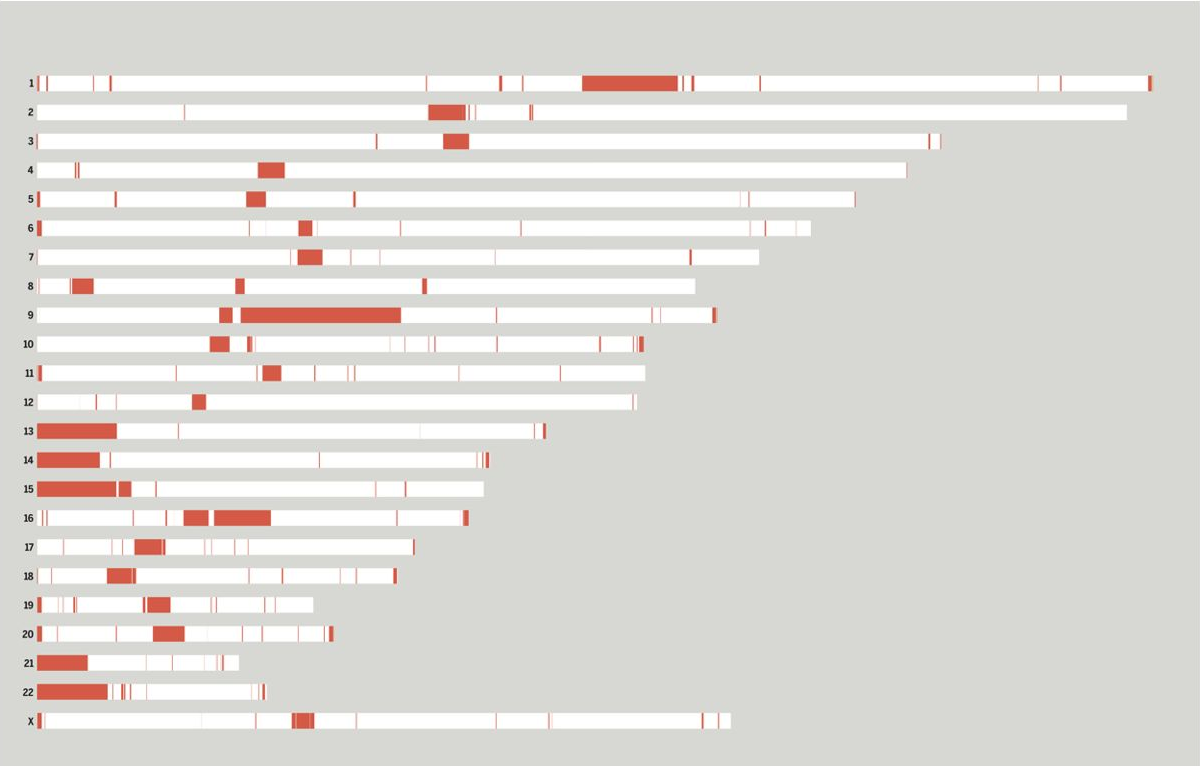


What is the next word, 'world' or 'age'?

What are eukaryotic genomes made of?

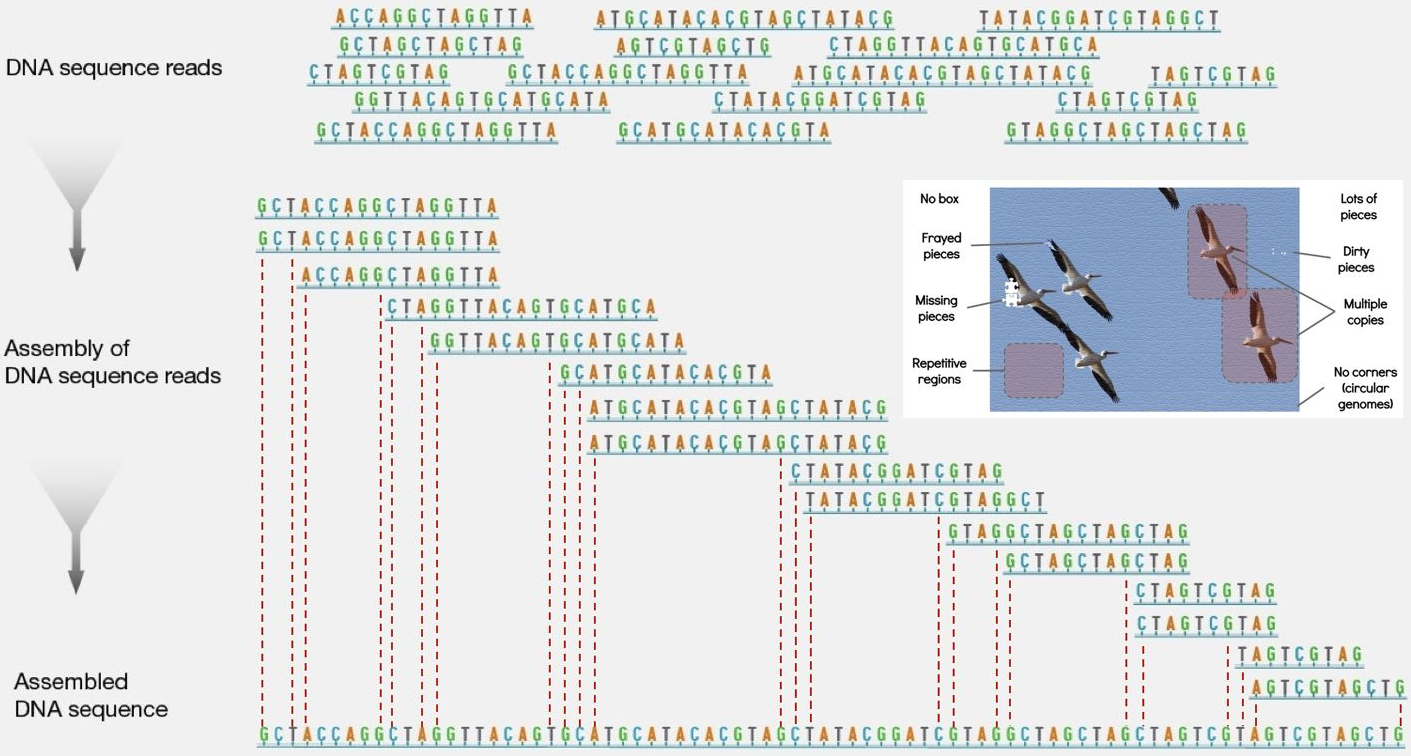
- Genes (exon, introns)
- Repetitive elements

1. Mobile elements (transposons)
2. Centromeres (tandem arrays of repeat sequence studded with transposable elements)
3. Telomeres (tandem arrays of simple repeats
4. Segmental duplications
5. rRNAS

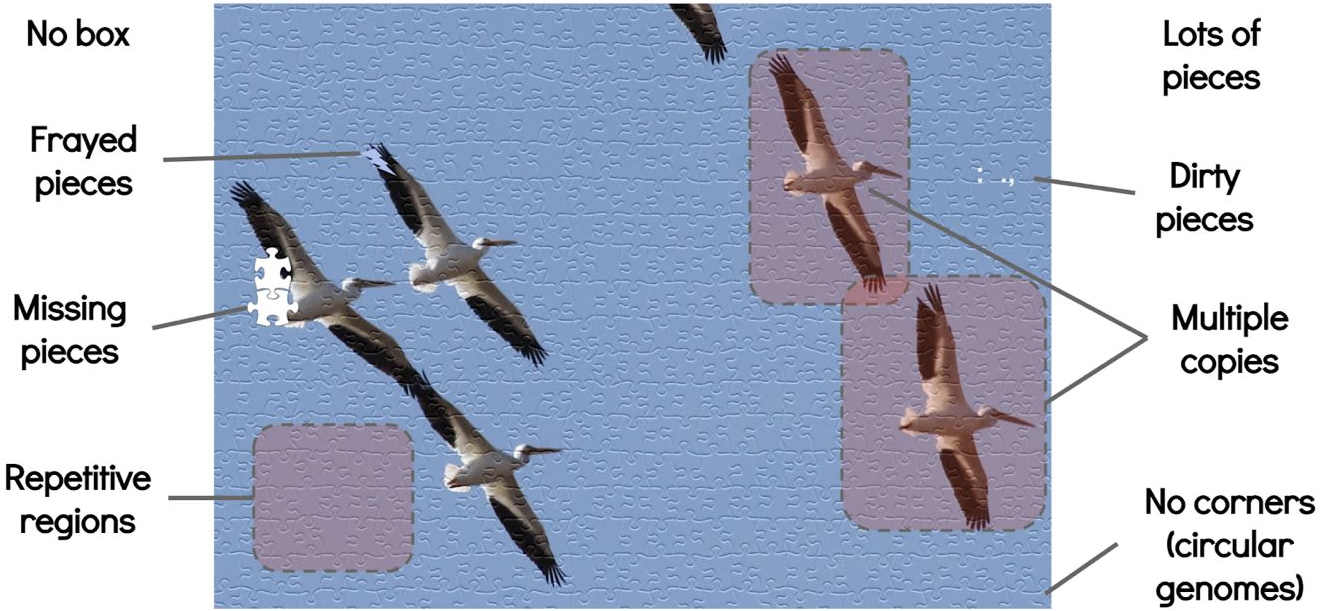


Complete human genome T2T-CHM13v2.0

The Naïve Genome Assembly Approach

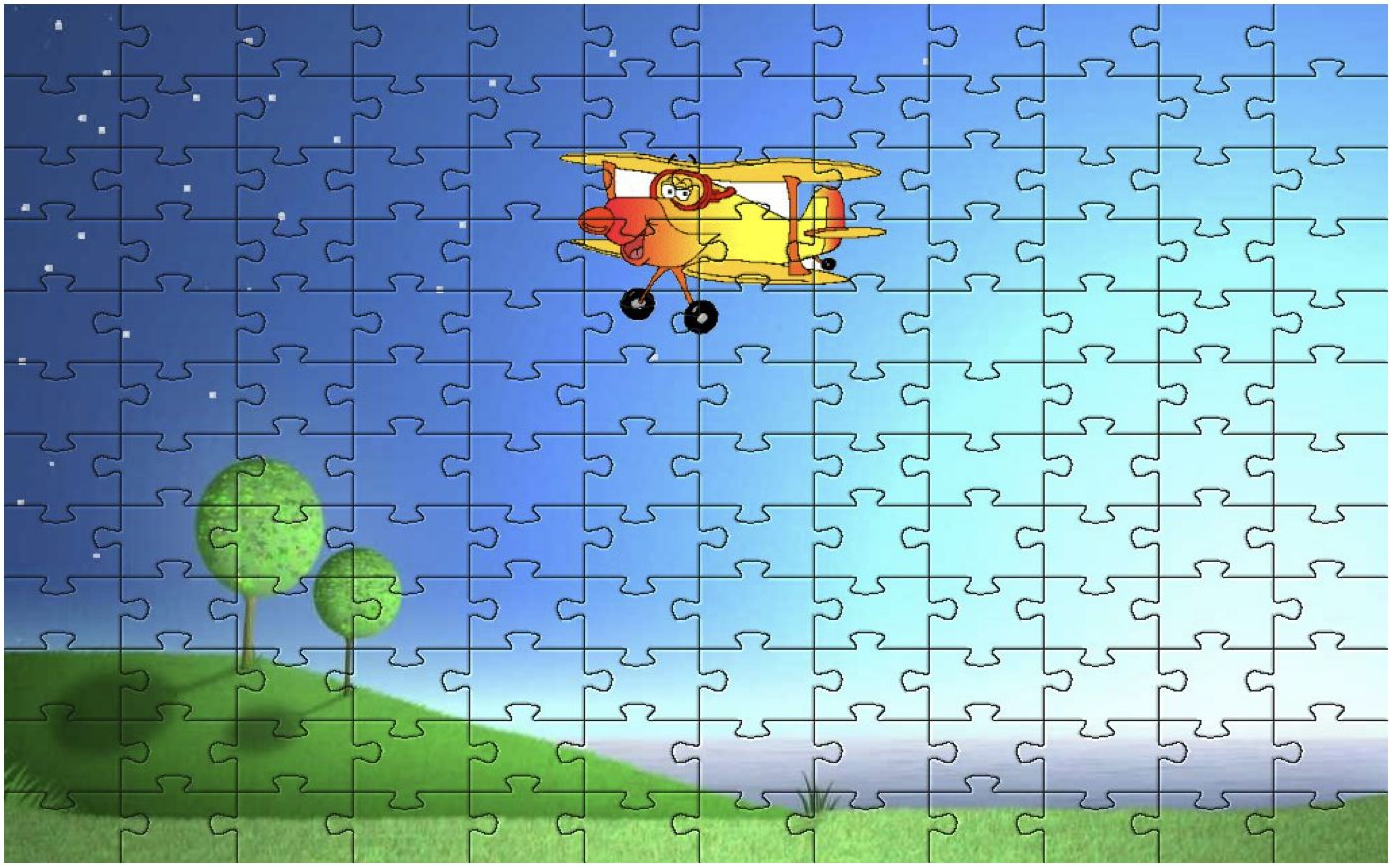


What makes a jigsaw puzzle hard?

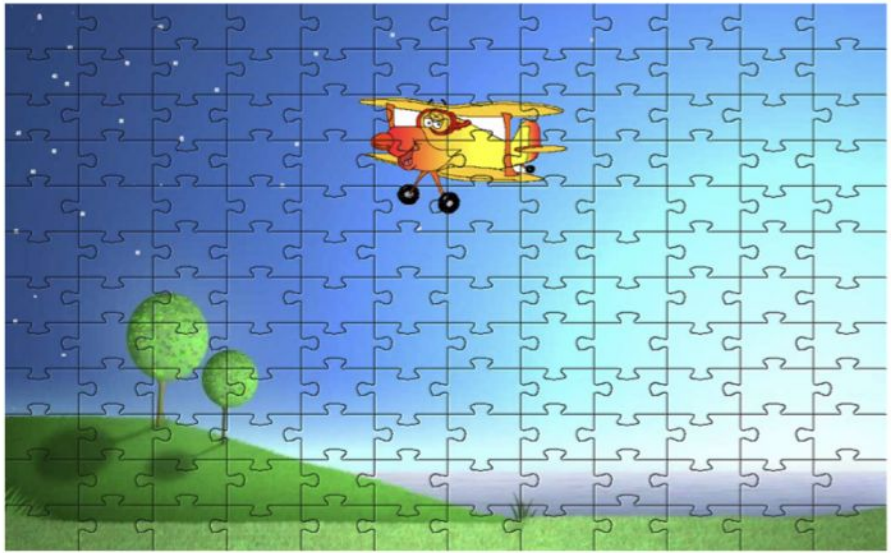


- What helps? Larger pieces (read length); fewer dirty or frayed pieces (errors in reads). fewer repeats and copies…

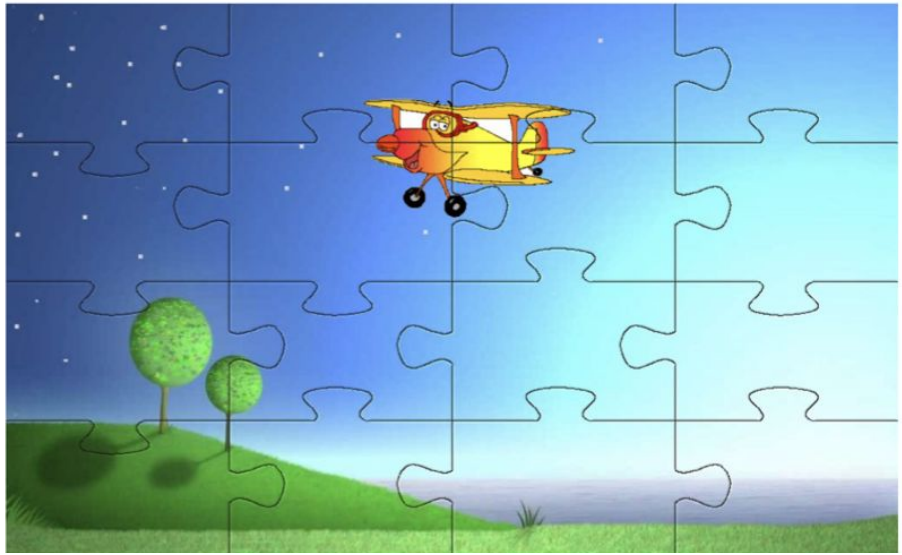
I'M A EUKARYOTIC GENOME – THE BLUE AND GREEN ARE MY REPEATS

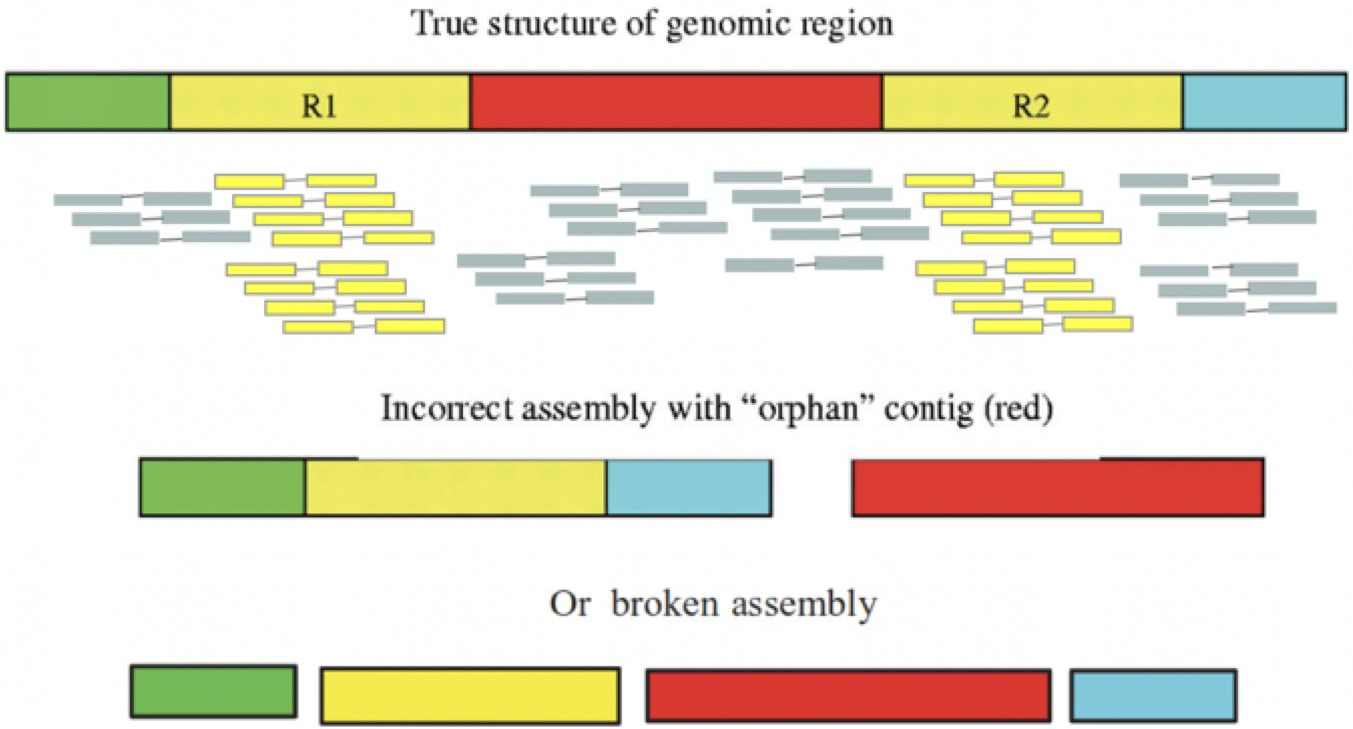


Genome assembly with short reads



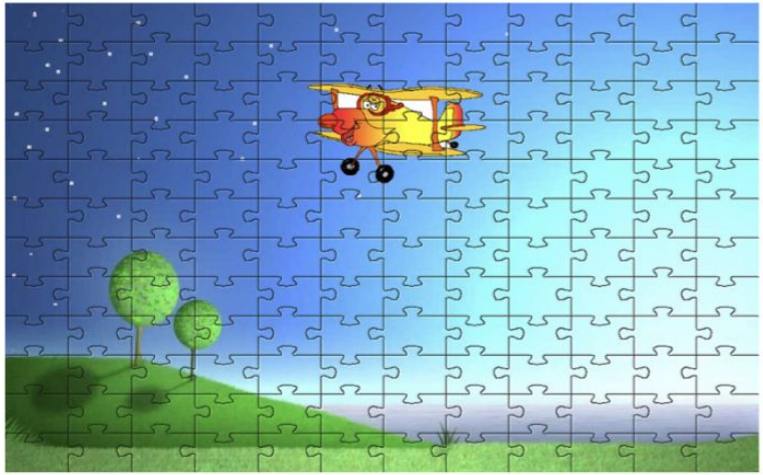
Genome assembly with long reads



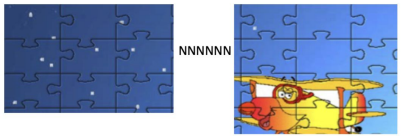


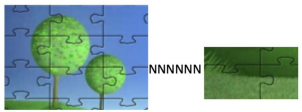
Modified from Salzberg S L , and Yorke J A Bioinformatics 2005;21:4320-4321

Genome assembly with short reads

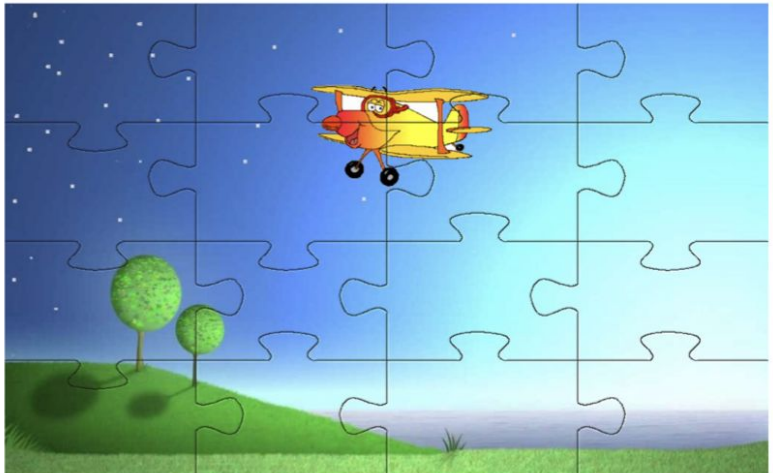


- Incomplete assemblies
- Chimeras
- Many errors





Genome assembly with long reads



- Complete assemblies
- High accuracy for biological inferences

I WANT TO TALK TO YOU ABOUT LONG READ SEQUENCING

Genome sequencing and assembly project: long reads

nature reviews genetics

https://doi.org/10.1038/s41576-024-00718-w

Review article

Check for updates

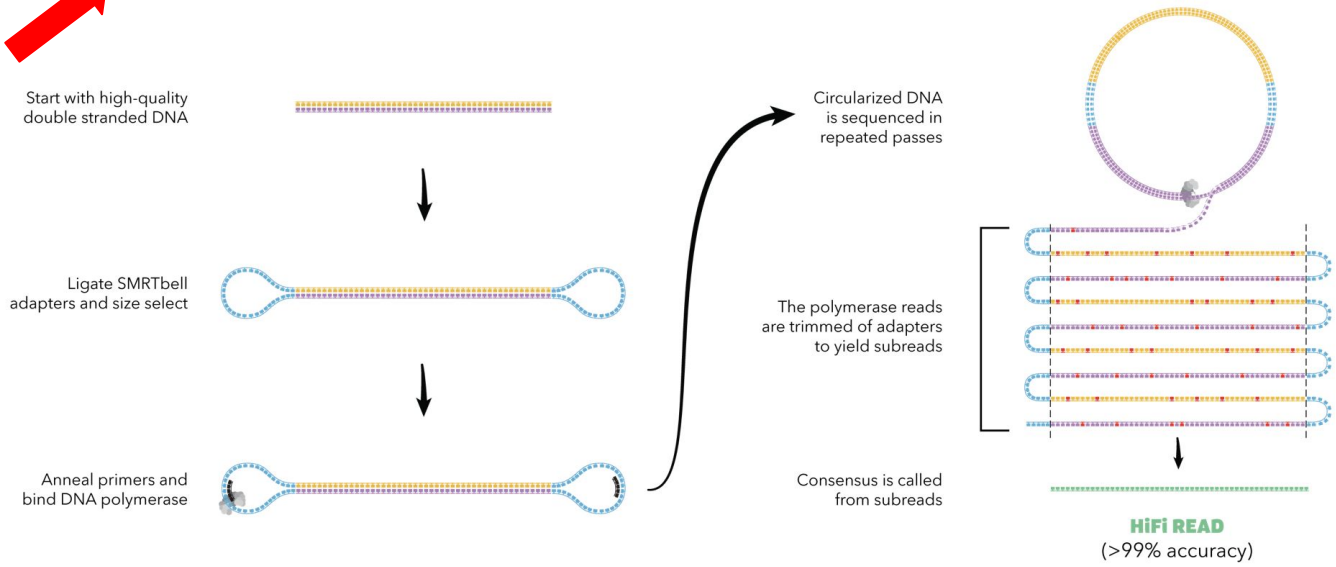
Genome assembly in the telomere-to-telomere era

Heng Li [1,2] & Richard Durbin [3]

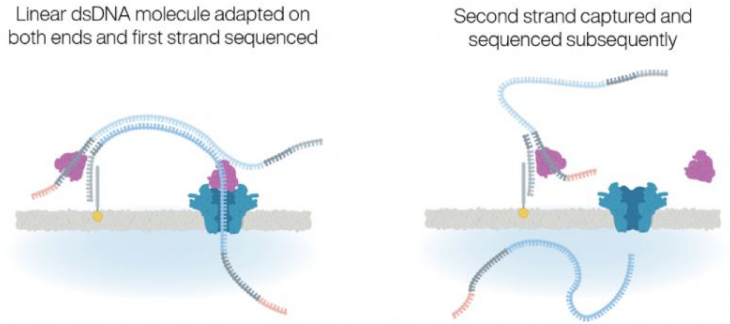
Table 1 | Common data types for high-quality assembly

Data type	Technologies	Description	Roles
Accurate long reads	PacBio HiFi, ONT duplex	>10 kb in length; error rate <0.5%	Initial assembly graph construction; phasing over heterozygous variants that are less than 10 kb apart
Ultra-long reads	ONT ultra-long	>100 kb in length; error rate <10%	Resolving tangles; phasing through homozygous regions over 100 kb in length
Trio data	Short-read	Standard whole-genome shotgun sequencing of parents	Whole-genome phasing
Long-range data	Hi-C, Pore-C, Strand-seq	Information over 1 kb to over 10 Mb in length	Chromosomal phasing; chromosome-scale scaffolding

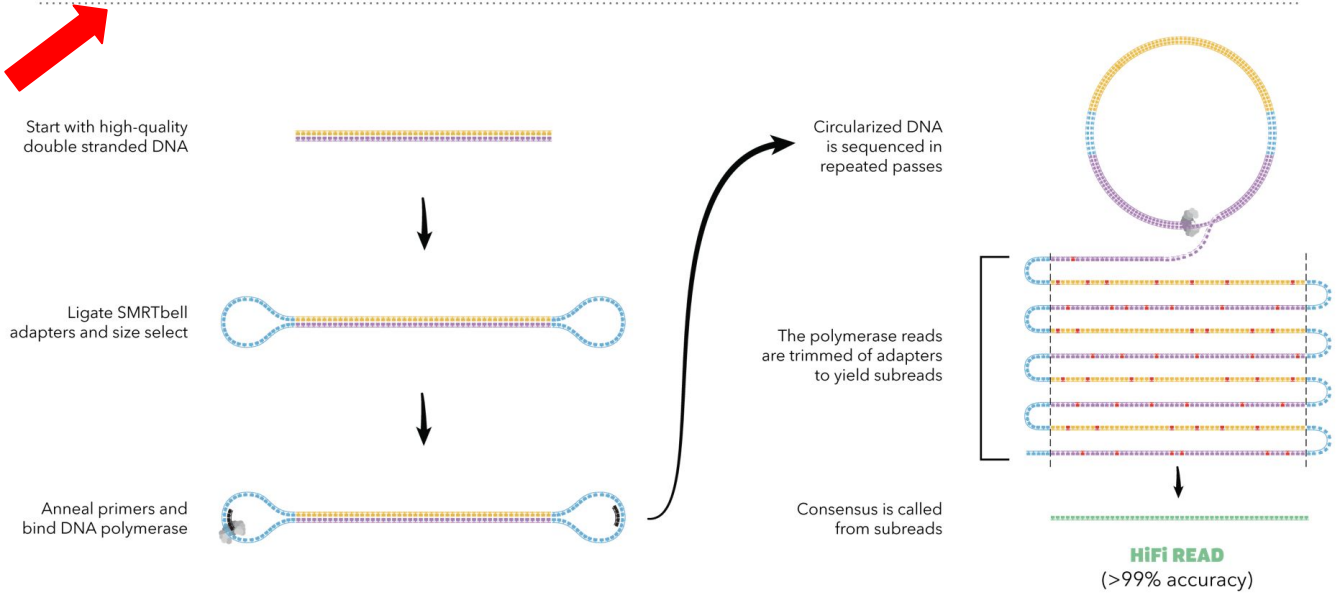
PACBIO HIFI READS



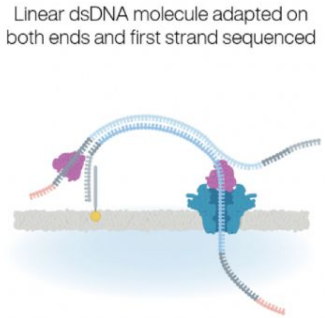
- **Nanopore Duplex**
  - >10 kb reads
  - 99.9% (Q30) read quality
  - 99.999% (Q50+) assembly quality

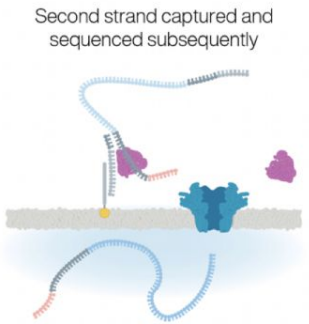


PACBIO HIFI READS



- **Nanopore Duplex**
- 99.9%
- (Q30+) assembly





All you need

Nanopore R10 + Hifiasm

- Pacbio HiFi: 25x coverage of both haplotypes
- Nanopore R10: 40x both haplotypes (polishing needed? Still to be determined)

YOUR GENOME ASSEMBLY PROJECT STARTS IN THE LAB

High Molecular Weight DNA extraction is key

No one-size-fits-all protocol!

JOURNAL ARTICLE

On the path to reference genomes for all biodiversity: laboratory protocols and lessons learned from processing over 2,000 species in the Sanger Tree of Life

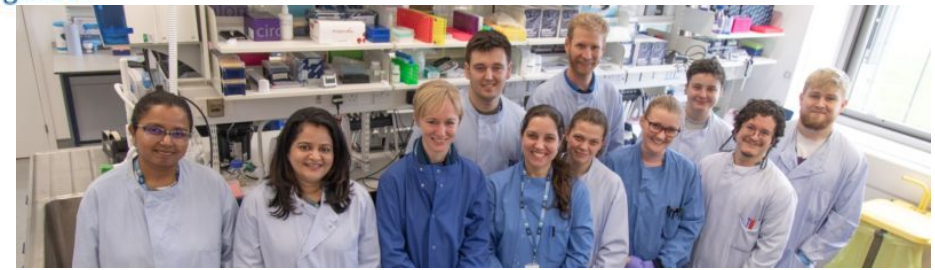
Caroline Howard, Amy Denton, Benjamin W Jackson, Adam Bates, Jessie Jay, Halyna Yatsenko, Priyanka Sethu Raman, Abitha Thomas, Graeme Oatley, Raquel Vionette do Amaral, Zeynep Ene Göktan, Juan Pablo Narváez Gómez, Isabelle Clayton Lucey, Elizabeth Sinclair, Michael A Quail, Mark Blaxter, Kerstin Howe, Mara K N Lawniczak

GigaScience, Volume 14, 2025, giaf119, https://doi.org/10.1093/gigascience/giaf119
Published: 23 October 2025 **Article history**

https://academic.oup.com/gigascience/article/doi/10.1093/gigascience/giaf119/8300236#537550287

protocols.io

Tree of Life Core Lab



OXFORD (GIGA)n SCIENCE

GigaScience, 2022, **11**, 1–13
DOI: 10.1093/gigascience/giac068
RESEARCH

VGP VERTEBRATE GENOMES PROJECT
A PROJECT OF THE G10K CONSORTIUM

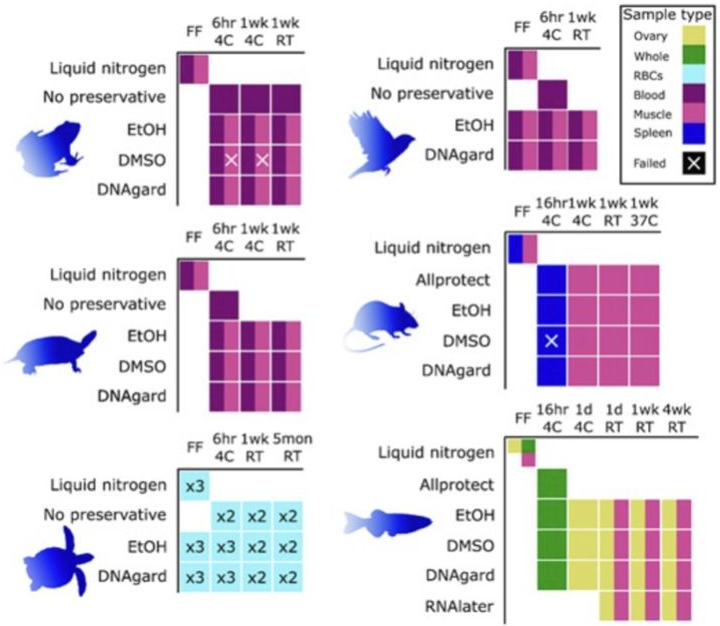
Benchmarking ultra-high molecular weight DNA preservation methods for long-read and long-range sequencing

Hollis A. Dahn [1,†], Jacquelyn Mountcastle [2,†], Jennifer Balacco [2], Sylke Winkler [3], Iliana Bista [4,5], Anthony D. Schmitt[6], Olga Vinnere Pettersson [7], Giulio Formenti [2], Karen Oliver[4], Michelle Smith [4], Wenhua Tan [3], Anne Kraus[3], Stephen Mac[6], Lisa M. Komoroske [8], Tanya Lama [8], Andrew J. Crawford [9], Robert W. Murphy [1], Samara Brown [2], Alan F. Scott [10], Phillip A. Morin [11], Erich D. Jarvis [2,12] and Olivier Fedrigo [2,*]

No one-size-fits-all protocol!

slack Channel: all.things.up.to.assembly

e 1:



I EXTRACTED HMW DNA: WHAT DO I DO NOW?

Darwin Tree of Life

Our recipe working across the Tree of Life:

Chromosome level genomes

- 25x Pacbio HiFi
- 100x Hi-C (Arima/Qiagen)

T2T (Telomere to Telomere) genomes

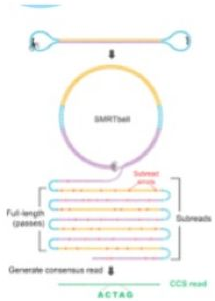
- The above plus 25x ONT Ultra Long (>100Kb reads)

DToL Current Pipeline

MitoHiFi

For mitochondria genome assembly

- Sequencing technologies: PacBio HiFi + HiC (Arima or Qiagen)



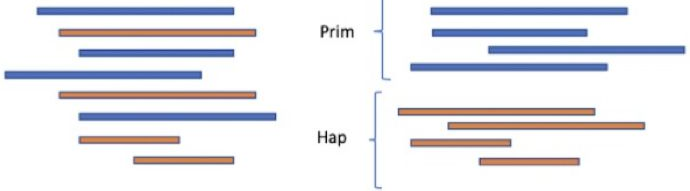
1- Kmer Jellyfish/ GenomeScope, asmstats, smudgeplot (se possível poliploide)

Assembly

Haplotype separation

Hicanu or Hifiasm

Purge dups



2 - asmstats, BUSCO, merqury

3 - asmstats, BUSCO, merqury

Scaffolding

Yahs scaffolding (Arima or Qiagen HiC)

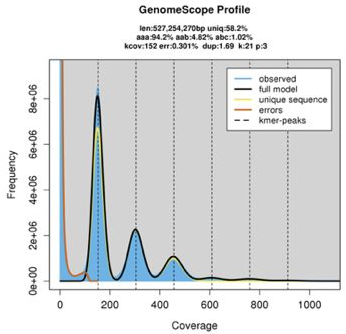
4 - asmstats, BUSCO, merqury, HiC heatmap

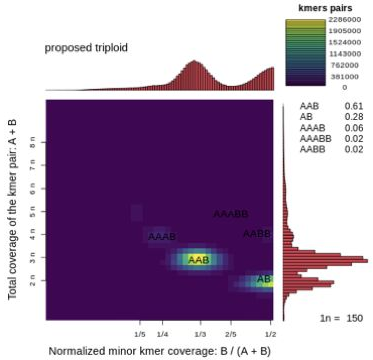
Curated assembly

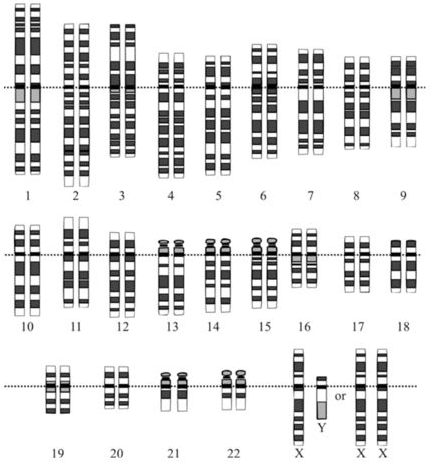
5 - asmstats, BUSCO, merqury, HiC heatmap

Key considerations to start your genome assembly project

- Genome size (flow cytometry, Kmer analysis, GoaT) **https://goat.genomehubs.org/**
- Heterozygosity (kmer analyses: jellyfish, genomescope)
- Repetitive content (kmer analyses: jellyfish, genomescope)
- Ploidy (kmer analyses: smudgeplots)

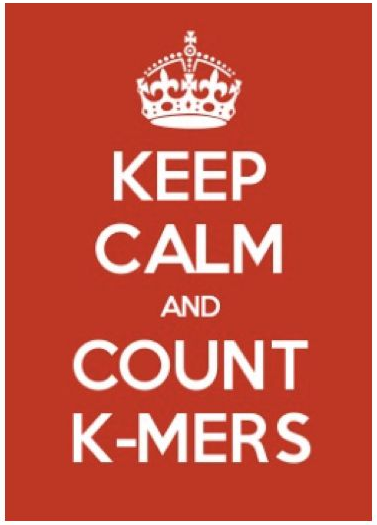




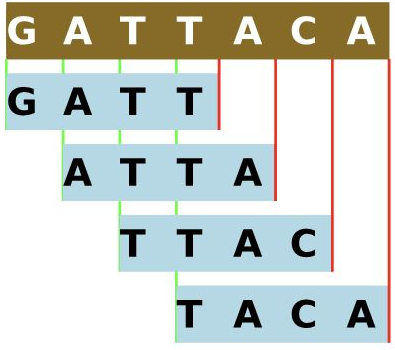


I HAVE HIGH-QUALITY DATA (ILLUMINA, PACBIO HIFI, DUPLEX NANOPORE)

I WILL do a kmer analysis first thing



KMER ANALYSIS



WHAT ARE K-MERS ?

- In biology, k-mers are unique subsequences of a sequence of length k

So, by way of example, the sequence `ATCGATCAC` contains the following *3-mers* (*k-mer* of size 3):

```
Sequence: ATCGATCAC
3-mer #0: ATC
3-mer #1:  TCG
3-mer #2:   CGA
3-mer #3:    GAT
3-mer #4:     ATC
3-mer #5:      TCA
3-mer #6:       CAC
```

APPLICATIONS OF K-MER ANALYSIS

- Genome assembly: K-mers used to construct De Brujin graphs
- Detect bacterial contamination on eukaryotic genome assembly (CG content discrepancies)
- Correcting NSG data
- Detect horizontal gene transfers
- Identification of CpG Islands
- **Estimation of genome size and heterozygosity**
- Genome assembly k-mer completeness

WHY ARE K-MERS SO POPULAR?

"Decomposing a sequence into its *k-mers* for analysis allows this set of fixed-size chunks to be analysed rather than the sequence, and this can be more efficient." (Bernardo Cavijo)

https://bioinfologics.github.io/post/2018/09/17/k-mer-counting-part-i-introduction/

COUNT AND HISTO

Counting *k-mers* in a (small) genome

We will start with an easy example first: the phi-X174 genome has 5386 bp and is a simple non-repetitive genome.

We can use `kat hist` to count *27-mers* on the genome and check how many times each *27-mer* appears (we start with `k = 27` because KAT uses that as default):

```
$ kat hist -o phiX.hist phiX.fasta
```

Checking the `phiX.hist` histogram (A.K.A. kmer spectrum) file, every *27-mer* in the genome appears only once. After the header lines starting with #, every line has a copy number (A.K.A. frequency) and a number of *k-mers*.

```
# Title:27-mer spectra for: phiX.fasta
# XLabel:27-mer frequency
# YLabel:# distinct 27-mers
# Kmer value:27
# Input 1:../genomes/phiX.fasta
###
1 5360
2 0
3 0
4 0
...
```

Bernardo Cavijo's post

COUNT AND HISTO

```
$ kat hist -o phiX_9mer.hist -m 9 phiX.fasta
```

Then the `phiX_9mer.hist` file looks like this:

```
# Title:9-mer spectra for: phiX.fasta
# XLabel:9-mer frequency
# YLabel:# distinct 9-mers
# Kmer value:9
# Input 1:phiX.fasta
###
1 4972
2 189
3 8
4 1
5 0
6 0
7 0
8 0
9 0
...
```

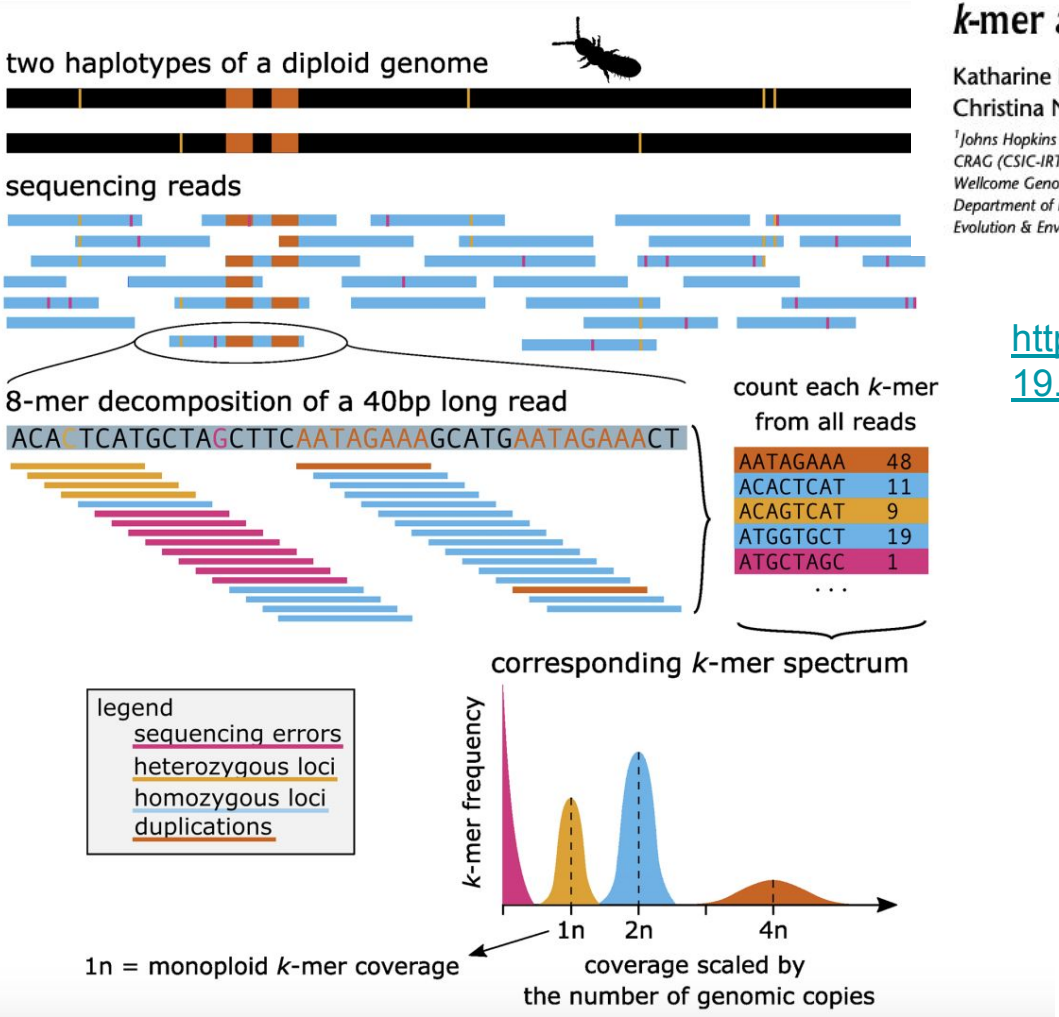
```
$ kat hist -o phiX_8mer.hist -m 8 phiX.fasta
```

Now the histogram file looks like this:

```
# Title:8-mer spectra for: phiX.fasta
# XLabel:8-mer frequency
# YLabel:# distinct 8-mers
# Kmer value:8
# Input 1:phiX.fasta
###
1 4159
2 491
3 67
4 8
5 1
6 0
7 0
8 0
9 0
```

Here, only **4159** *8-mers* are *unique*, out of **4726** *distinct 8-mers*, that are present in the genome's **5377** *total 8-mers*.

Bernardo Cavijo's post



k-mer approaches for biodiversity genomics

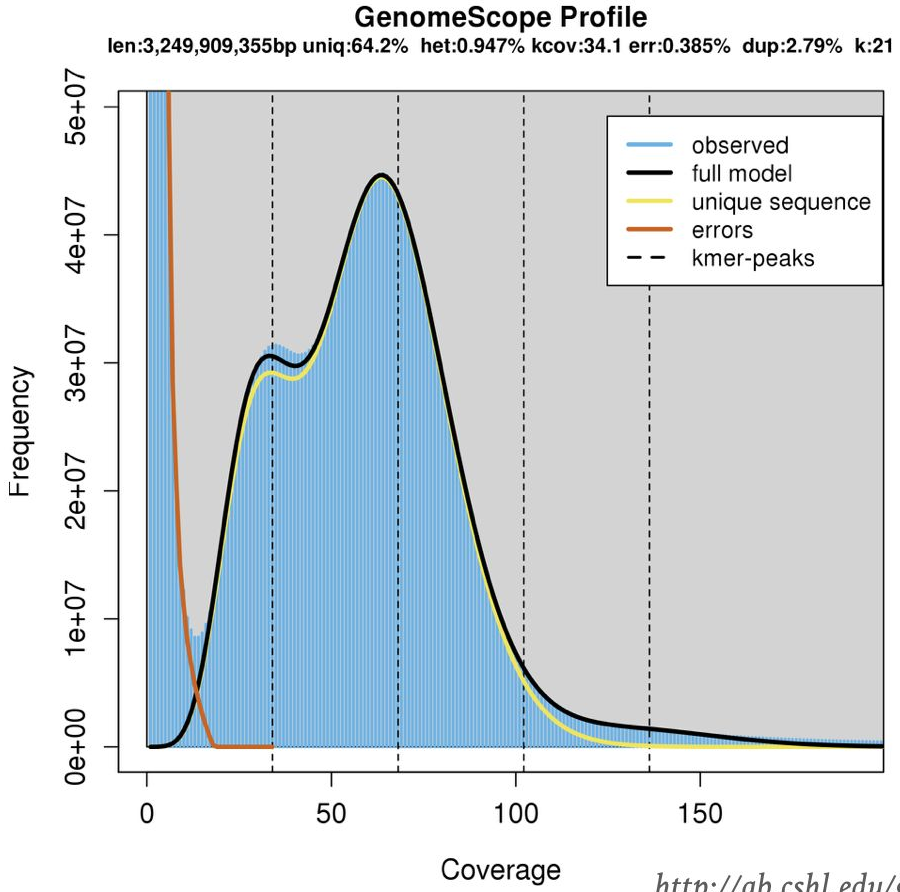
Katharine M. Jenike,[1] Lucía Campos-Domínguez,[2] Marilou Boddé,[3] José Cerca,[4,6] Christina N. Hodson,[5] Michael C. Schatz,[1] and Kamil S. Jaron[3]

[1]*Johns Hopkins University, School of Medicine, Baltimore, Maryland 21205, USA;* [2]*Centre for Research in Agricultural Genomics, CRAG (CSIC-IRTA-UAB-UB), Campus UAB, Cerdanyola del Vallès, 08193 Barcelona, Spain;* [3]*Tree of Life, Wellcome Sanger Institute, Wellcome Genome Campus, Hinxton, Cambridge CB10 1SA, United Kingdom;* [4]*Center for Ecological and Evolutionary Synthesis, Department of Biosciences, University of Oslo, 0313 Oslo, Norway;* [5]*University College London, UCL Department of Genetics, Evolution & Environment, London, WC1E 6BT, United Kingdom*

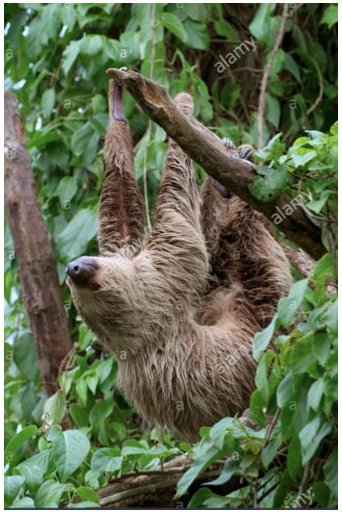
https://genome.cshlp.org/content/35/2/219.full

GENOME RESEARCH

A TYPICAL KMER PLOT FOR A DIPLOID SPECIES



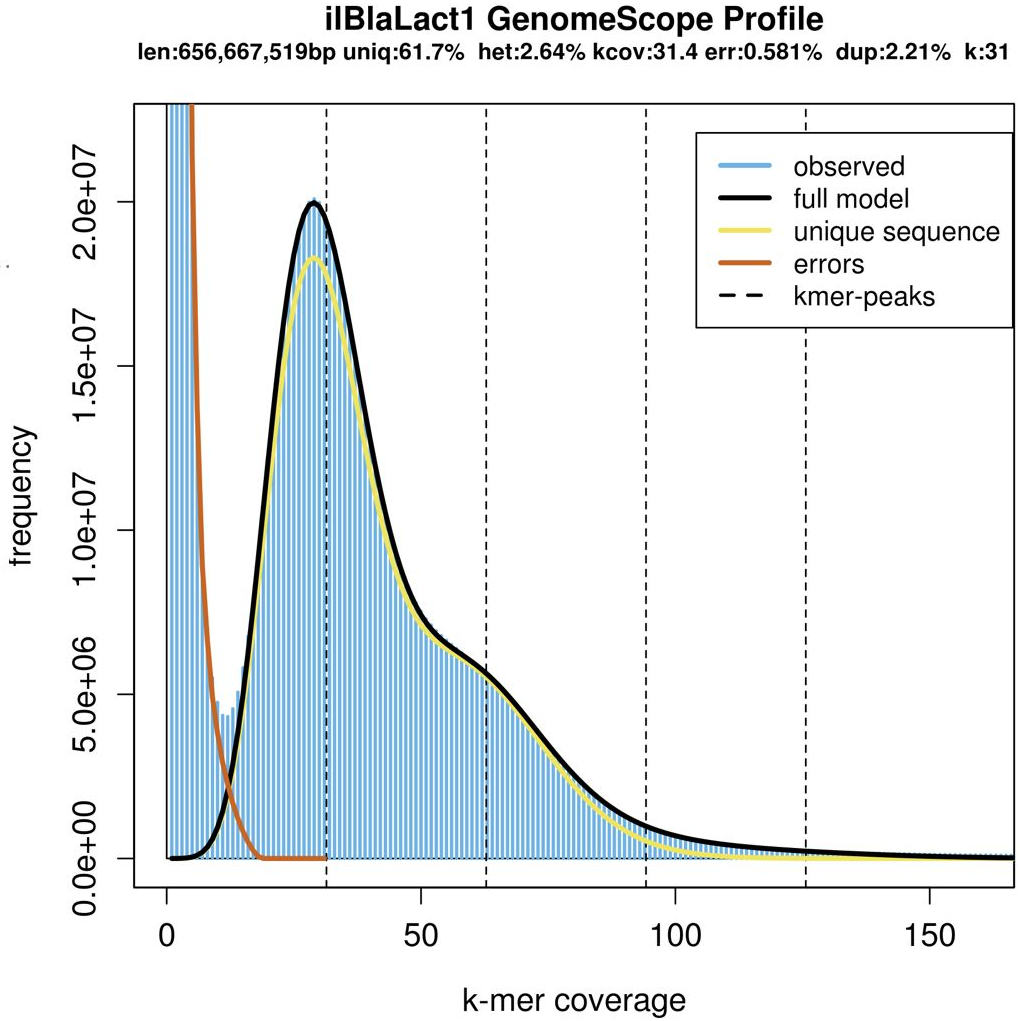
Choloepus didactylus (VGP)



http://qb.cshl.edu/genomescope/analysis.php?code=bVuZNlhwn2tVCHhRN71I

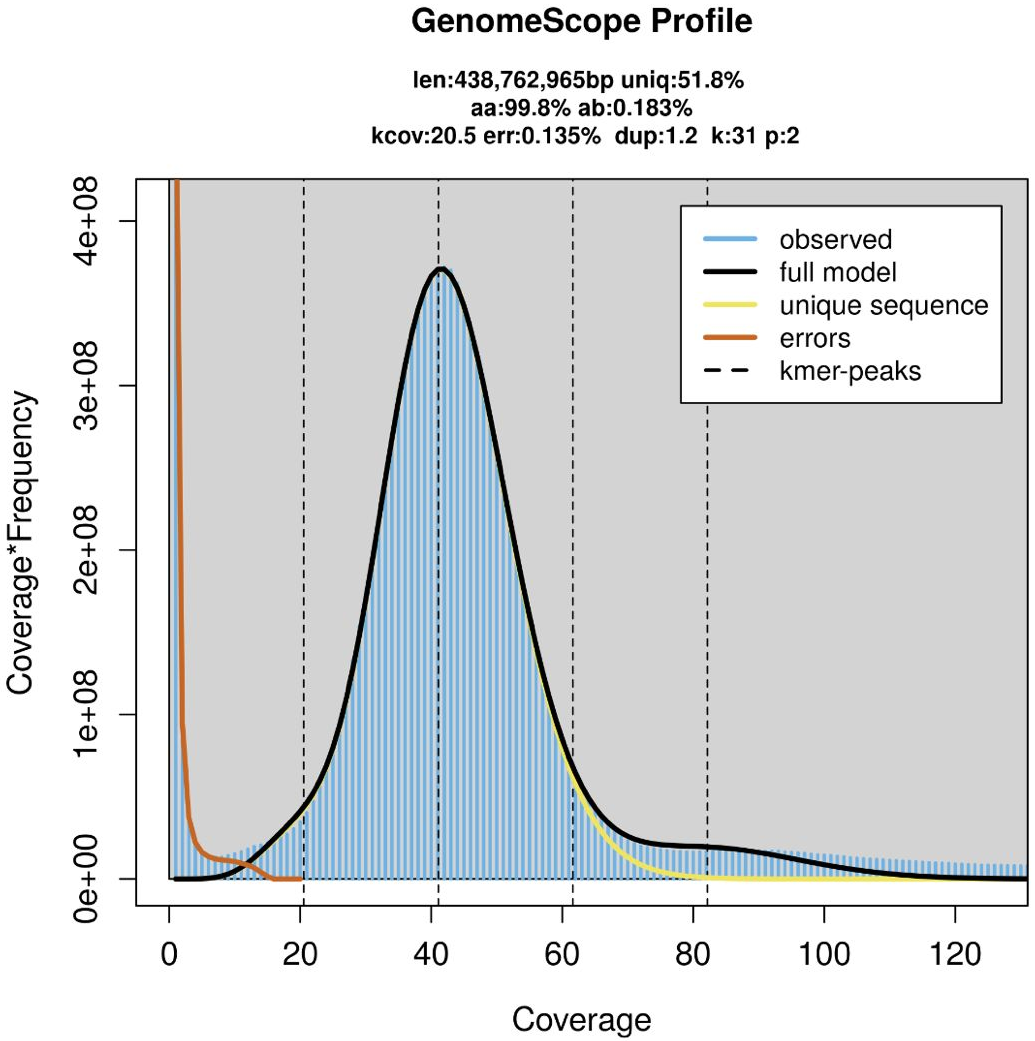
A TYPICAL KMER PLOT FOR A DIPLOID SPECIES WITH HIGH HETEROZYGOSITY

Blastobasis lacticolella (DToL)



A TYPICAL KMER PLOT FOR A DIPLOID SPECIES WITH LOW HETEROZYGOSITY

Urtica urens

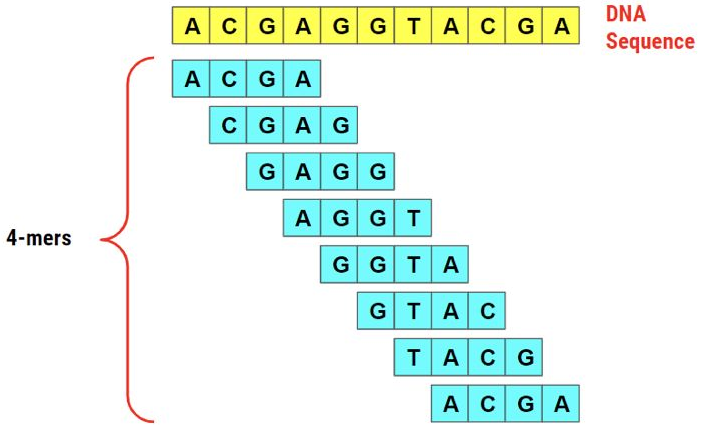


KMER SIZE

Choosing *k*: specificity vs. Sensitivity

- Using a *k* that is too small will result in many unrelated sequences being composed of the same *k-mers*, in a textbook case of specificity loss because there being very few possible *k-mers* of that size. In the extreme of the small *k*, k=1 only distinguishes two *canonical k-mers*: A and C. 1-mer analysis is incredibly popular in biology, but it is best known by the name of *GC content analysis*.

- Using extremely large *k* values would sacrifice many of the benefits and sensitivity of *k-mer* analyses in the first place. (Bernado Cavijo's post)

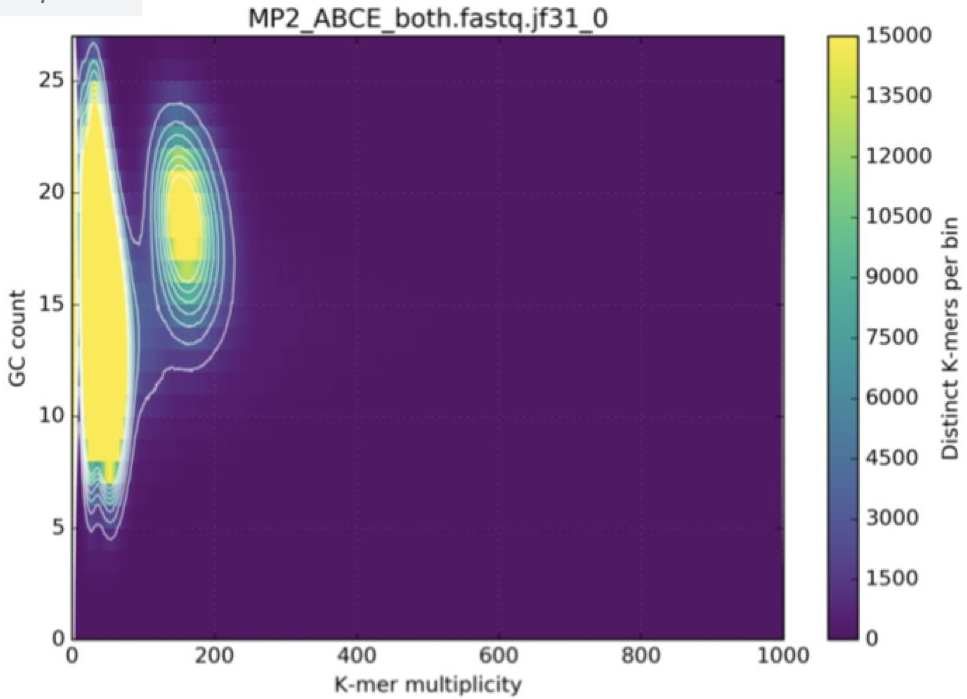


Why do we chose k=31 so often?

One reason is: it is specific enough that a large number of them are unique both in mammalian-sized genomes and in bacterial genome databases.

SPOTTING BACTERIAL CONTAMINATION: KMER AND ITS GC CONTENT

github.com/TGAC/KAT



You can use KAT to plot this!

README.md

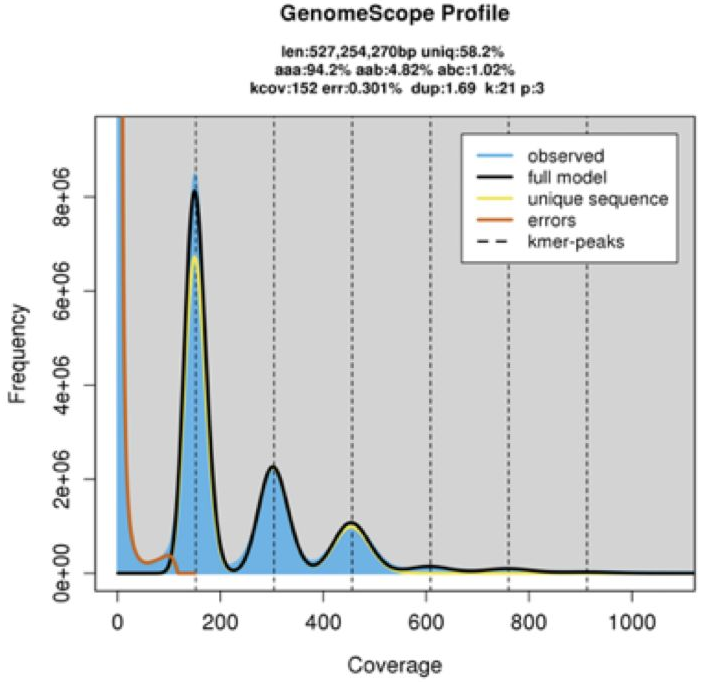
KAT - The K-mer Analysis Toolkit

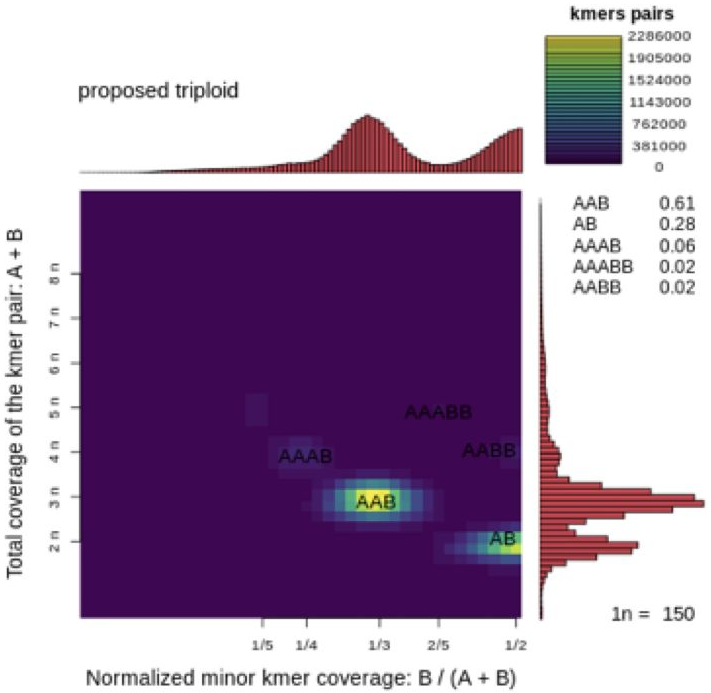
KAT is a suite of tools that analyse jellyfish hashes or sequence files (fasta or fastq) using kmer counts. The following tools are currently available in KAT:

- **hist**: Create an histogram of k-mer occurrences from a sequence file. Adds metadata in output for easy plotting.
- **gcp**: K-mer GC Processor. Creates a matrix of the number of K-mers found given a GC count and a K-mer count.
- **comp**: K-mer comparison tool. Creates a matrix of shared K-mers between two (or three) sequence files or hashes.
- **sect**: SEquence Coverage estimator Tool. Estimates the coverage of each sequence in a file using K-mers from another sequence file.

Tubastraea tagusensis

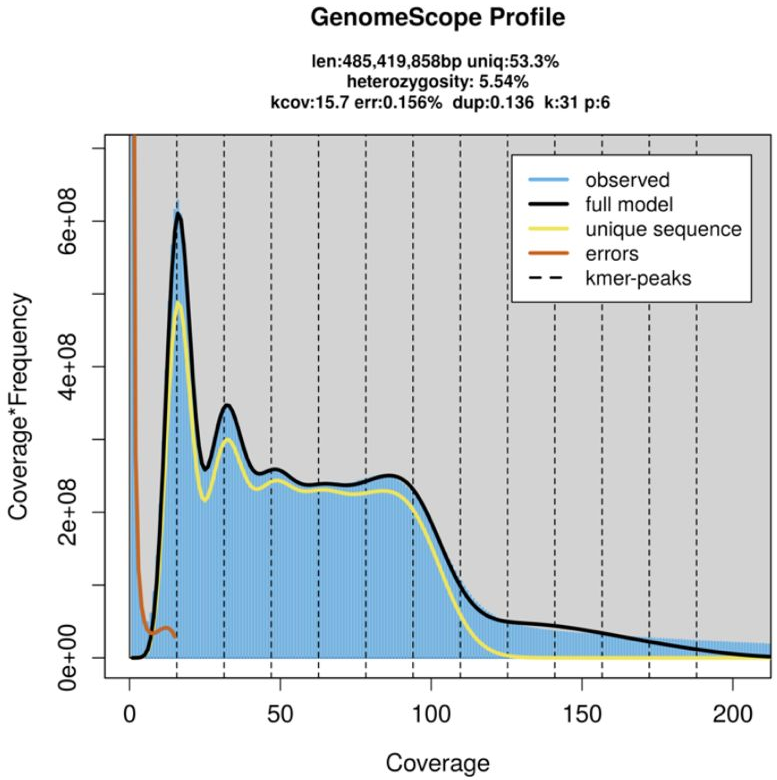
KMER PROFILE FOR A TRIPLOID SPECIES



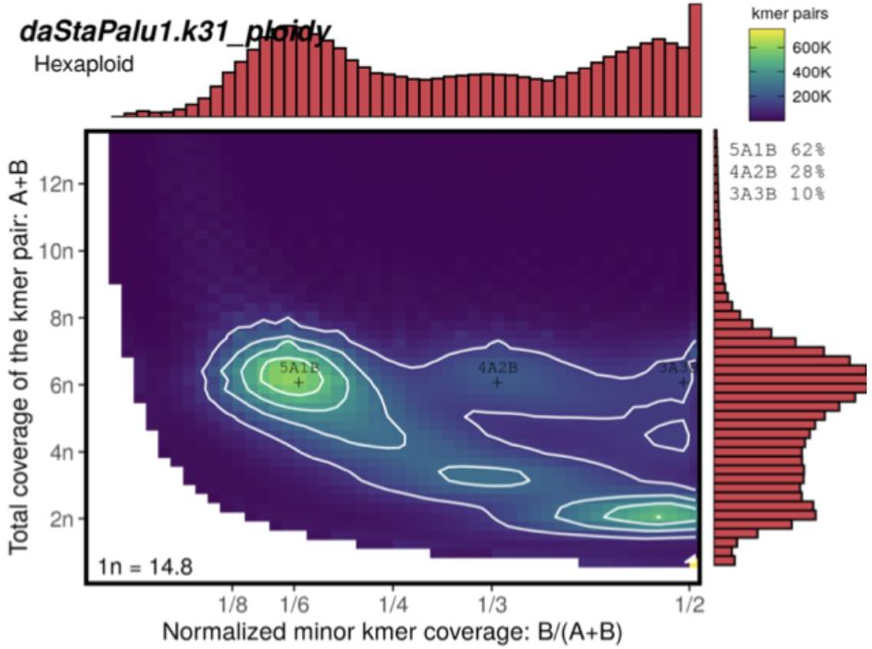


KHMER PROFILE FOR A POLYPLOID SPECIES

pacbio daStaPalu1 GenomeScope 2.0 linear plot



Ploidy stack plot daStaPalu1

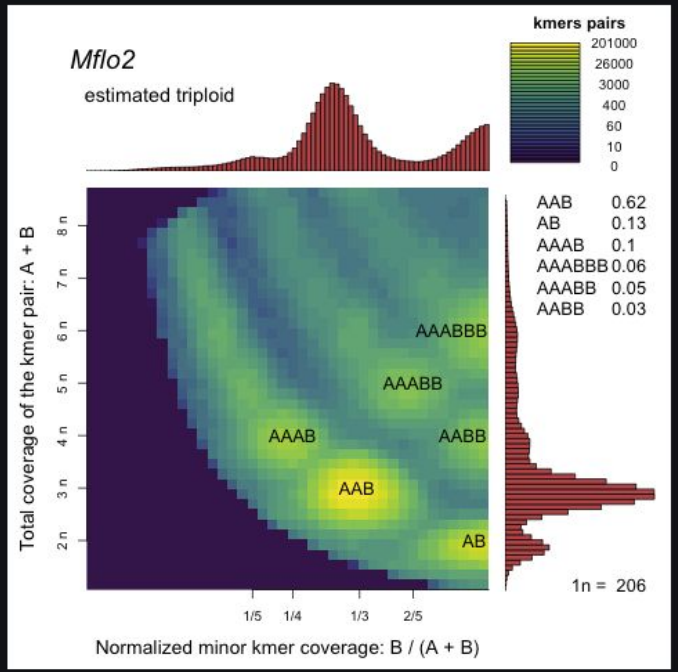


https://github.com/KamilSJaron/smudgeplot

How smudgeplot works

This tool extracts heterozygous kmer pairs from kmer count databases and performs gymnastics with them. We are able to disentangle genome structure by comparing the sum of kmer pair coverages (CovA + CovB) to their relative coverage (CovB / (CovA + CovB)). Such an approach also allows us to analyze obscure genomes with duplications, various ploidy levels, etc.

Smudgeplots are computed from raw or even better from trimmed reads and show the haplotype structure using heterozygous kmer pairs. For example (of an older version):



1. What are heterozygous kmers? Are these kmer pairs with a close but not perfect sequence match?

Yes. Right now heterozygous kmers are those that:

- are exactly one SNP from each other (for instance AATCA ACTCA)
- form a unique pair (i.e. there are no other kmers one SNP away from them. for instance ATGATCA ATGCTCA ATGGTCA would be discarded - they three not two)

Like this we heavily subsample the genome, but so far this was very sufficient to sample enough heterozygous kmers to see the genome structure.

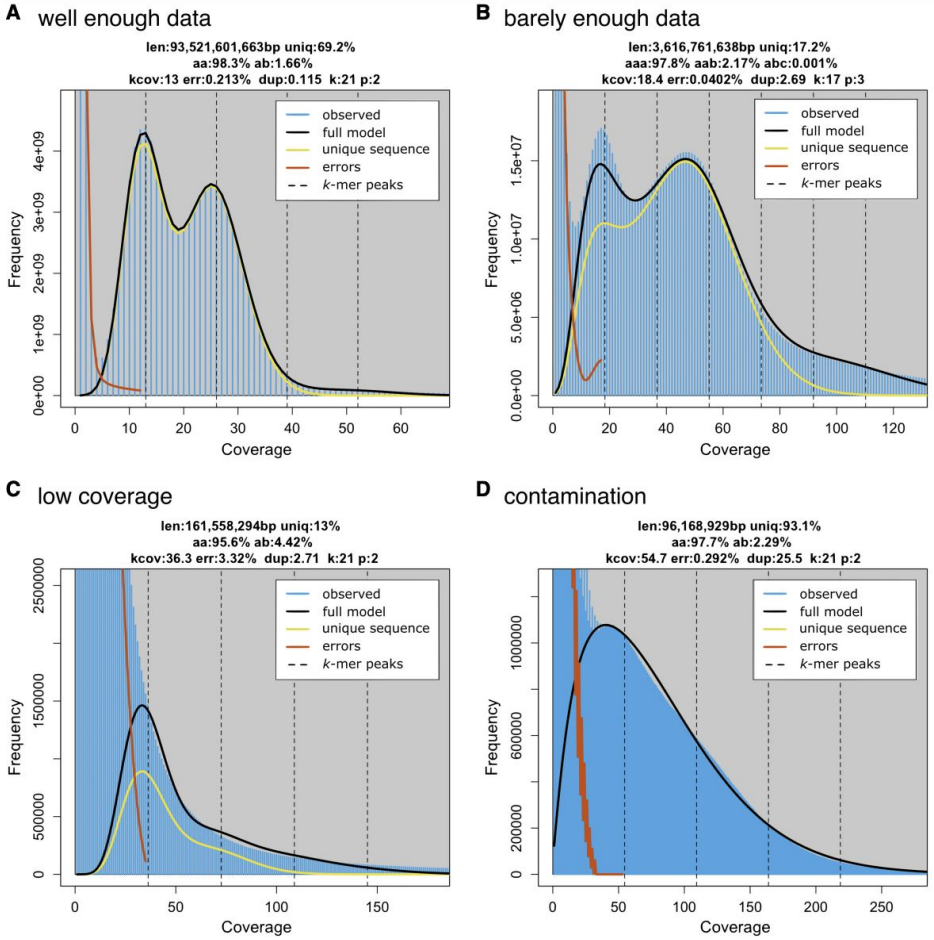


Figure 2. Examples of *k*-mer spectra. (*A*) *Viscum album:* a diploid spectra with enough data to observe two distinct peaks and fit a model that accurately reflects genomic features despite the large size of the genome. (*B*) *Procambarus virginalis:* *k*-mer spectra of a sample with low coverage, barely sufficient for a model fit. Notably, we used $k = 17$ to increase the *k*-mer coverage and make the model fit possible. (*C*) *Allium schoenoprasum*: The sequencing coverage of this data set is approximately 1×. Error *k*-mers and genome *k*-mers are completely blended; as a consequence, the model did not converge to meaningful estimates. (*D*) *Hypsibius dujardini*: a heavily contaminated sample of a tardigrade.

k-mer approaches for biodiversity genomics

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Common signatures of *k*-mer spectra

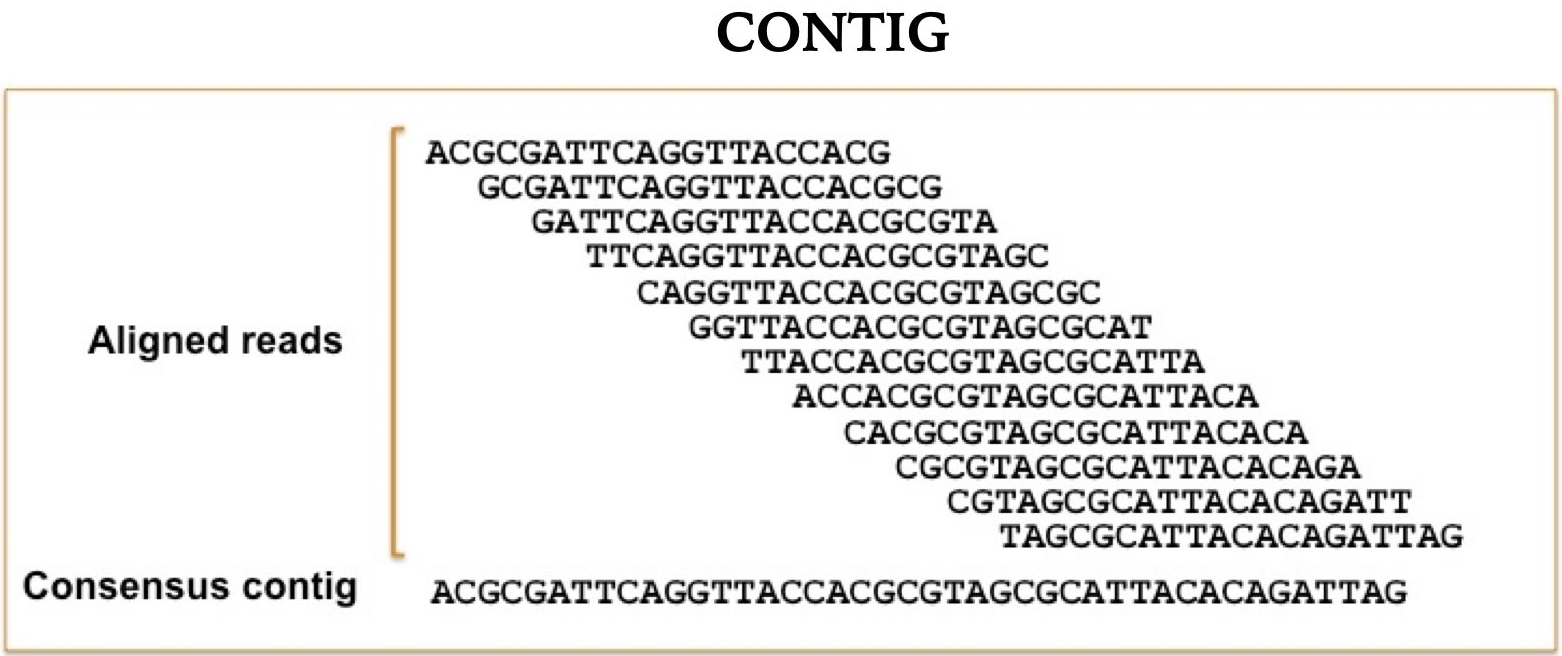
To generate a high quality reference of a diploid genome it is recommended to sequence at least 25-30x coverage of long reads, or more generally 15x per haplotype [49,50]. Even a simple visual inspection of *k*-mer spectra is valuable to quickly assess if this coverage is achieved. Such coverage should generate a *k*-mer spectrum that shows distinct coverage peaks as demonstrated by the European mistletoe *Viscum album* (**Figure 3A**). Sequencing data without sufficient coverage will have poorly defined peaks, because the homozygous and heterozygous genomic peaks will be blended at the left side of the coverage plot. If the peaks are still visible, it might be possible to fit a meaningful genome model, like in the case of the crayfish *Procambarus virginalis* (**Figure 3B;** data from [51]).

Usually, something already known about the species we sequence; in particular ploidy or genome size that were previously assessed via cytogenetic techniques. Confronting prior knowledge with the estimates derived from the *k*-mer spectra is often helpful in identifying potential problems in the data. In the case of the crayfish, there is a nearly perfect match of genome size estimate from the *k*-mer spectra and flow cytometry [51], supporting that the model converged well. Very low sequencing coverage or elevated rates of errors, leads to blending of peaks; genomic *k*-mers become indistinguishable from error *k*-mers. This is visible in the chive *Allium schoenoprasum* (**Figure 3C**) where the model (black line) does not fit the data (blue histogram) well. In such cases, the estimated values are just artefacts of a poor convergence. The predicted genome size is much lower than what we would expect in *Allium* genus, where other species have genomes ranging from 8.4-13.4Gbp [52], and the coverage is much higher than what we would expect from a spectrum of this shape. Coverage problems are usually resolvable with additional sequencing, while high error rates may require a different sequencing technology and/or library preparation.

MORE ON SMUDGEPLOTS

https://www.youtube.com/watch?v=8vuNSvrAIoA

KNOWING THE CHALLENGE, YOU GO AND BUILD CONTIGS WITH ASSEMBLERS



Check point

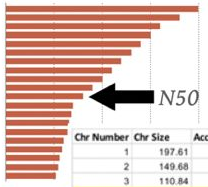
DOES MY ASSEMBLED SIZE CORRESPONDS WITH MY ESTIMATED GENOME SIZE?

Genomics is a game of going back and forth

Basic Assembly Metrics

- Total assembled sequence length
- Number of sequences (contigs and scaffolds)
- Average length (contigs and scaffolds)
- Largest/smallest (contigs and scaffolds)
- N50 = X means 50% of the genome is in sequences larger than X
- NG50 (N50 scaled by the expected genome size)
- Number of gaps

N50 = what is the smallest contig at 50% of genome?



Chr Number	Chr Size	Accum Size	Genome % Coverage
1	197.61	197.61	18.82%
2	149.68	347.29	33.07%
3	110.84	458.13	43.63%
4	91.32	549.45	52.32%
Z	82.53	631.98	60.18%
5	59.81	691.79	65.88%
7	36.74	728.53	69.37%
6	36.37	764.9	72.84%
8	30.22	795.12	75.71%
9	24.15	819.27	78.01%
10	21.12	840.39	80.03%
12	20.39	860.78	81.97%
11	20.2	880.98	83.89%
13	19.17	900.15	85.72%
14	16.22	916.37	87.26%
20	13.9	930.27	88.58%
15	13.06	943.33	89.83%
18	11.37	954.7	90.91%
17	10.76	965.46	91.94%
19	10.32	975.78	92.92%
27	8.08	983.86	93.69%
33	7.82	991.68	94.43%
21	6.84	998.52	95.08%
W	6.81	1005.33	95.73%
24	6.49	1011.82	96.35%
23	6.15	1017.97	96.94%
31	6.15	1024.12	97.52%
26	6.06	1030.18	98.10%
22	5.46	1035.64	98.62%
28	5.12	1040.76	99.11%
25	3.98	1044.74	99.48%
16	2.84	1047.58	99.76%
30	1.82	1049.4	99.93%
32	0.73	1050.13	100.00%
MT	0.02		
Total	1050.15		

Quality metrics in genomics

- **N50: half of the genome is assembled in scaffolds that are the N50 size, or larger**

Chr Number	Chr Size	Accum Size	Genome % Coverage
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19	10.32	975.78	92.92%
27	8.08	983.86	93.69%
33	7.82	991.68	94.43%
21	6.84	998.52	95.08%
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30	1.82	1049.4	99.93%
32	0.73	1050.13	100.00%
MT	0.02		
Total	1050.15		

Scaffold N50

@ Chromosome level

N50 = 91Mb

Assembled size= 1Gb

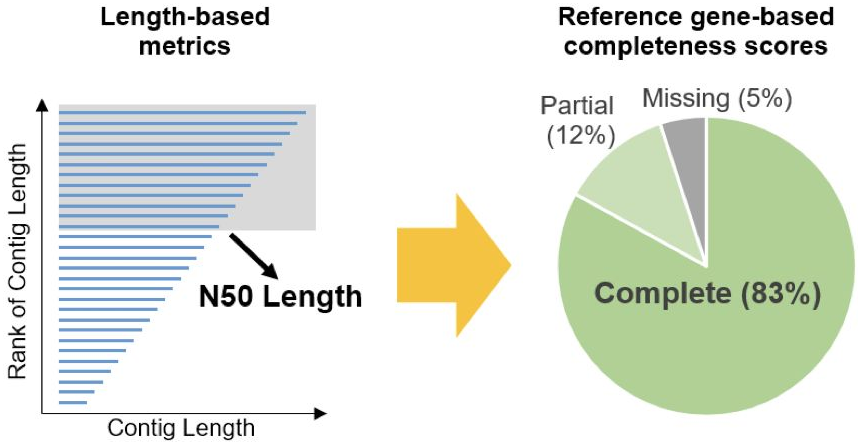
How many scaffolds= 32

BUSCO v3

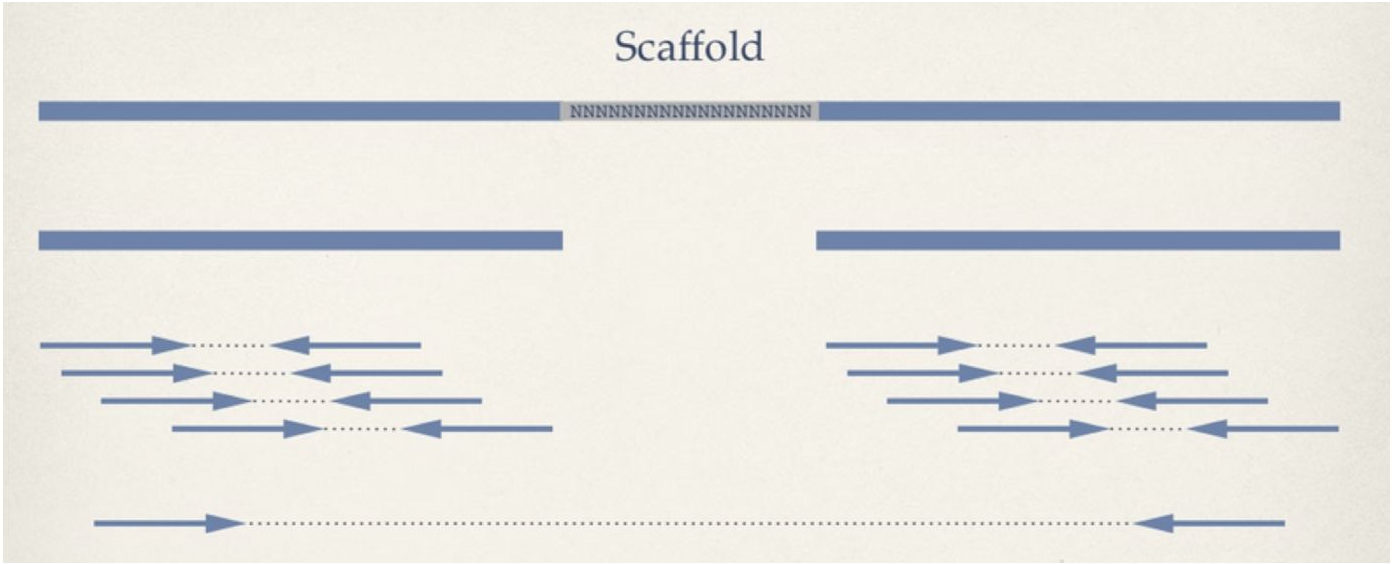
Assessing genome assembly and annotation completeness with
Benchmarking **U**niversal **S**ingle-**C**opy **O**rthologs

- The quality metrics for genome assembly should not be only the ones related to contiguity, rather, the composition of the genes present in the assembly is also crucial

More accurate assessment for genome assembly!



Scaffolding methods



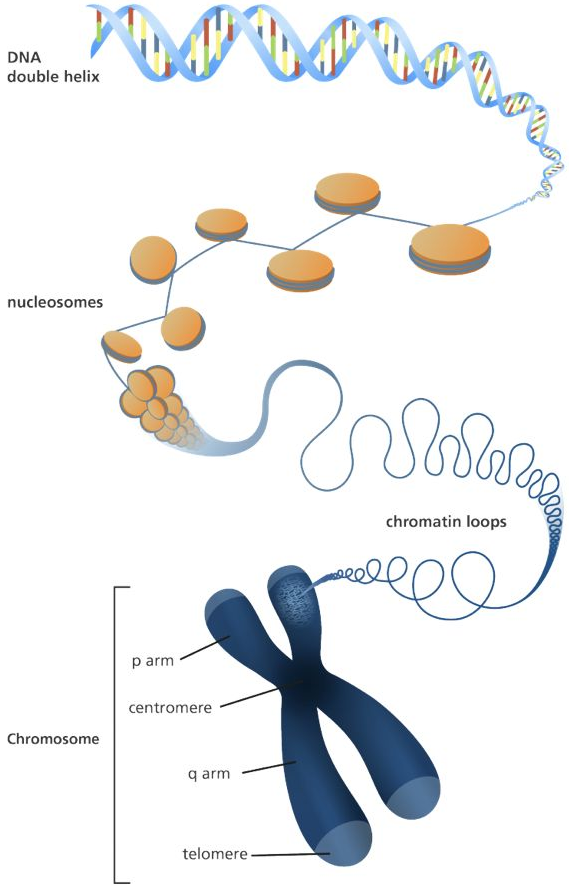
Scaffold: joining and orienting contigs

*Scaffolding methods: mate-pairs (blerg), optical maps (bionano), **Hi-C**, Nanopore UltraLong reads*

HOW DO I BUILD UP SCAFFOLDS AND CHROMOSOMES?

Hi-C and Ultralong Nanopore

The human genome consists of over 3 billion nucleotides and is contained within 23 pairs of chromosomes. ***If the chromosomes were aligned end to end and the DNA stretched, the genome would measure roughly 2 meters long. Yet the genome functions within a sphere smaller than a tenth of the thickness of a human hair (10 micron)***. … the genome does not exist as a simple one-dimensional polymer; instead the genome folds into a complex compact three-dimensional structure. (Lajoie et al 2015)

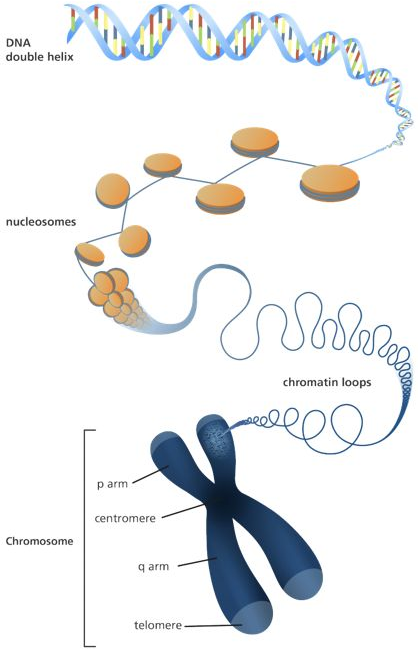


Chromosome conformation

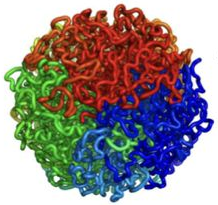
➤*The organisation of the chromatin in the nucleus is extremely relevant to biological function at the gene level as well as the global nuclear level.*

➤*The study of the packaging and organisation of chromatin in the nucleus will shed light on:*

- *the spatial aspects of gene regulation*
- *chromosome morphogenesis*
- *genome stability*
- *genome transmission*
- *biophysics of chromatin*
- *pathologies related to genome instability or nuclear morphology*



Published in final edited form as:
Science. 2009 October 9; 326(5950): 289–293. doi:10.1126/science.1181369.



Hi-C

Comprehensive mapping of long range interactions reveals folding principles of the human genome

Erez Lieberman-Aiden[1,2,3,4,*], **Nynke L. van Berkum**[5,*], **Louise Williams**[1], **Maxim Imakaev**[2], **Tobias Ragoczy**[6,7], **Agnes Telling**[6,7], **Ido Amit**[1], **Bryan R. Lajoie**[5], **Peter J. Sabo**[8], **Michael O. Dorschner**[8], **Richard Sandstrom**[8], **Bradley Bernstein**[1,9], **M. A. Bender**[10], **Mark Groudine**[6,7], **Andreas Gnirke**[1], **John Stamatoyannopoulos**[8], **Leonid A. Mirny**[2,11], **Eric S. Lander**[1,12,13,†], and **Job Dekker**[5,†]

[1] Broad Institute of Harvard and MIT, Cambridge, Massachusetts 02139, USA.

[2] Division of Health Sciences and Technology, MIT, Cambridge, Massachusetts 02139, USA.

[3] Program for Evolutionary Dynamics, Department of Organismic and Evolutionary Biology, Department of Mathematics, Harvard University, Cambridge, Massachusetts 02138, USA.

[4] Department of Applied Mathematics, Harvard University, Cambridge, Massachusetts 02138, USA.

[5] Program in Gene Function and Expression and Department of Biochemistry and Molecular Pharmacology, University of Massachusetts Medical School, Worcester, Massachusetts 01605, USA.

[6] Fred Hutchinson Cancer Research Center, Seattle, Washington 98109, USA.

[7] Department of Radiation Oncology, University of Washington School of Medicine, University of Washington, Seattle, Washington 98195, USA.

[8] Department of Genome Sciences, University of Washington, Seattle, Washington 98195, USA.

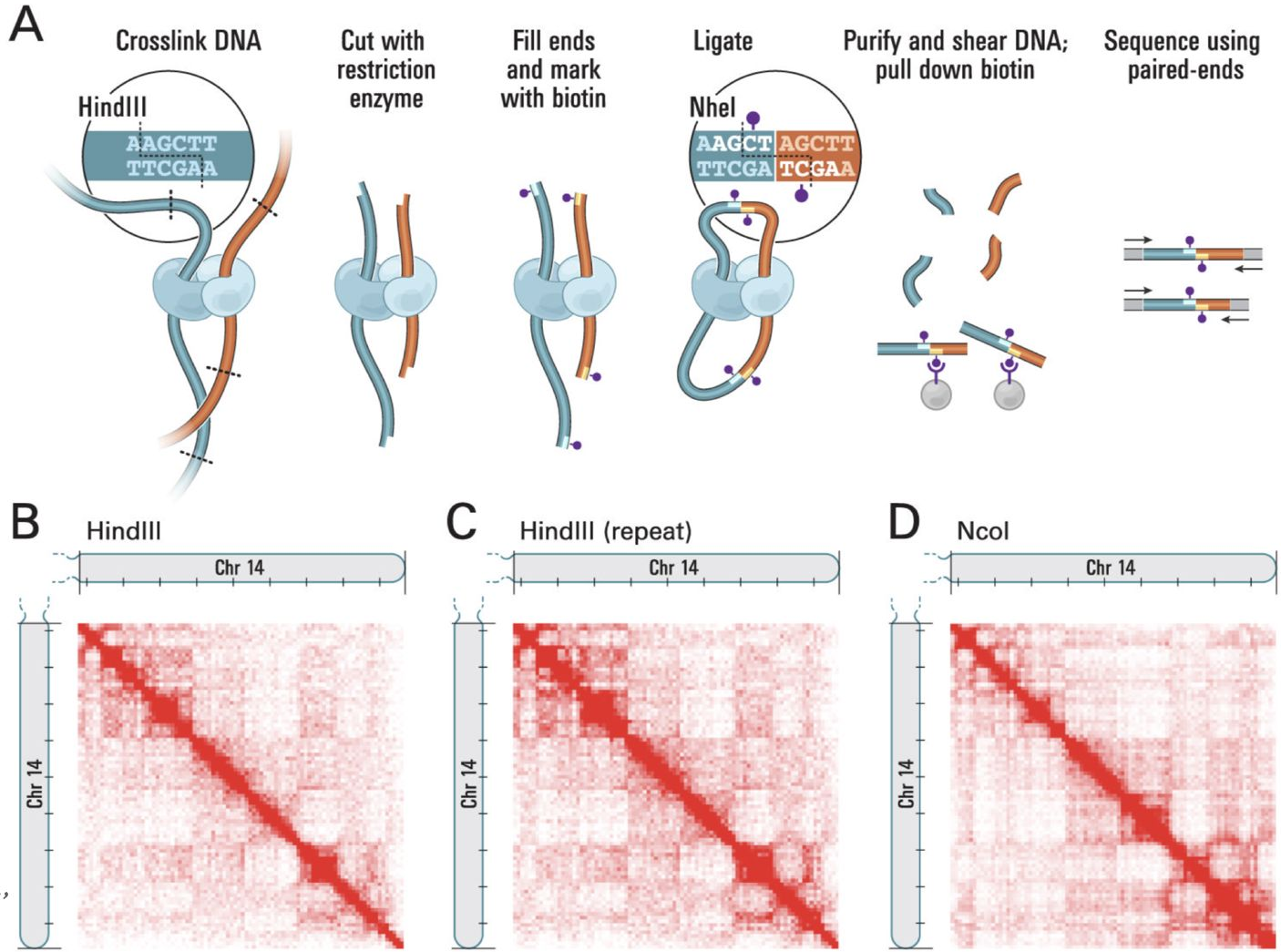
[9] Department of Pathology, Harvard Medical School, Boston, Massachusetts 02115, USA.

[10] Department of Pediatrics, University of Washington, Seattle, Washington 98195, USA.

[11] Department of Physics, MIT, Cambridge, Massachusetts 02139, USA.

[12] Department of Biology, MIT, Cambridge, Massachusetts 02139, USA.

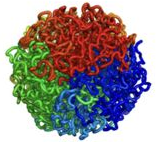
[13] Department of Systems Biology, Harvard Medical School, Boston MA 02115.



Lieberman-Aiden et al., 2010

HI-C

- Intrachromosomal contact probability is on average much higher than interchromosomal.
- Interaction probability rapidly decays with increasing genomic distance.



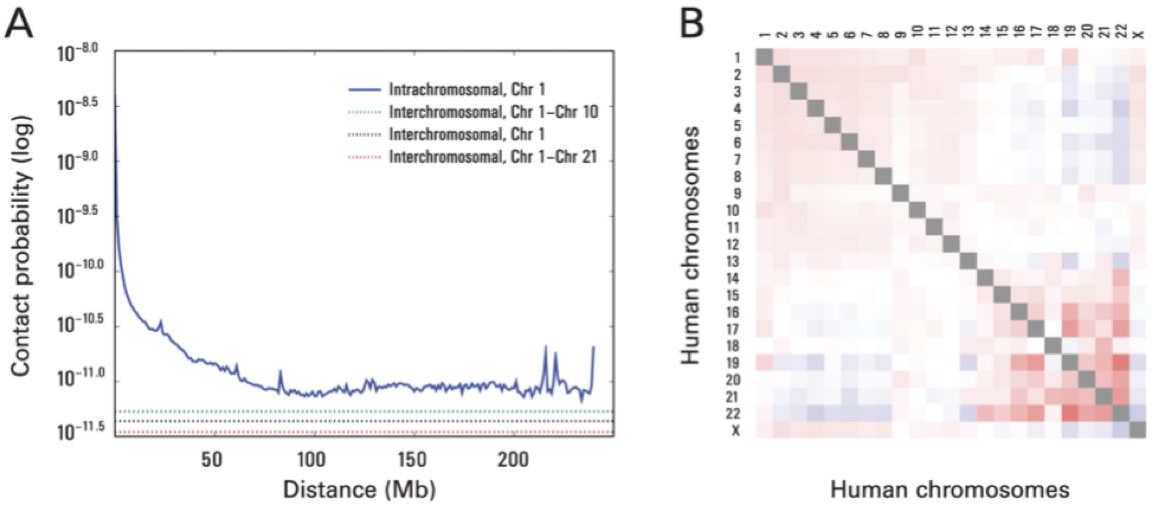
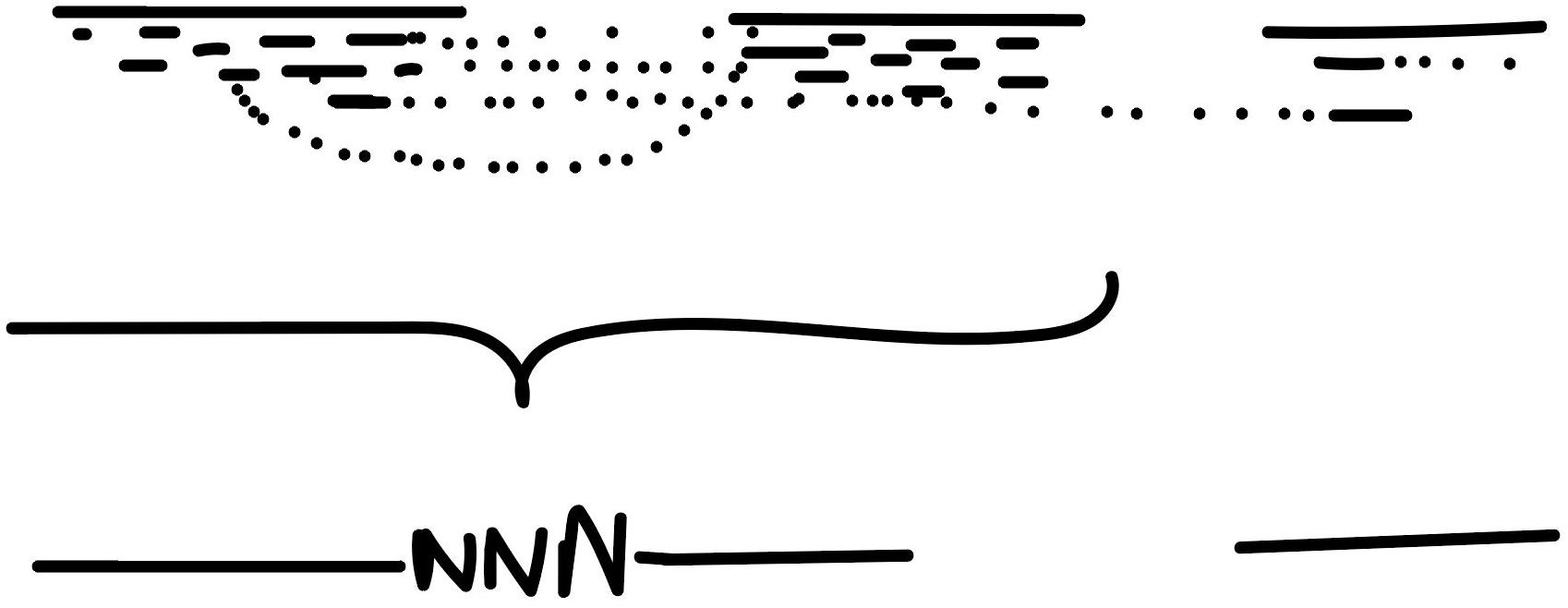


Fig. 2.
The presence and organization of chromosome territories. (**A**) Probability of contact decreases as a function of genomic distance on chromosome 1, eventually reaching a plateau at ~90M (blue). The level of interchromosomal contact (black dashes) differs for different pairs of chromosomes; loci on chromosome 1 are most likely to interact with loci on chromosome 10 (green dashes) and least likely to interact with loci on chromosome 21 (red dashes). Interchromosomal interactions are depleted relative to intrachromosomal interactions. (**B**) Observed/expected number of interchromosomal contacts between all pairs of chromosomes. Red indicates enrichment, and blue indicates depletion (up to twofold). Small, gene-rich chromosomes tend to interact more with one another.

HOW TO DO HI-C SEQUENCING

- You have a protocol for Hi-C extraction
- This is sequenced as short Illumina reads
- You map the Hi-C data to your built contigs (Arima Mapping pipeline or BWA men -5SP)
- Ran YaHS and/or ~~Salsa~~ for scaffolding
- Build and look at Hi-C HeatMaps



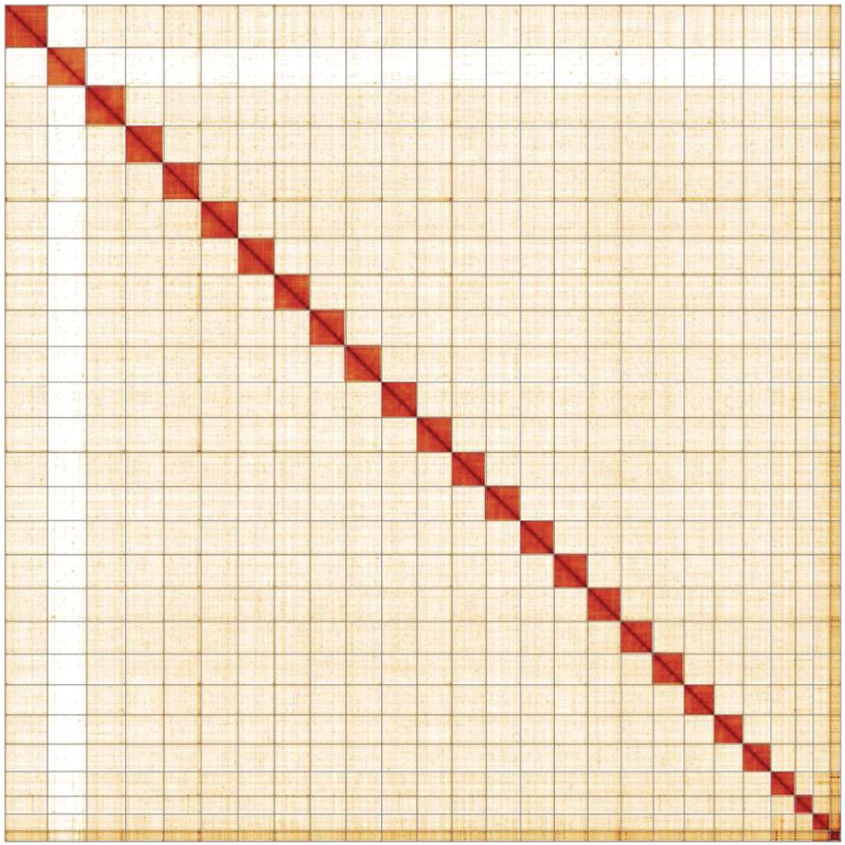
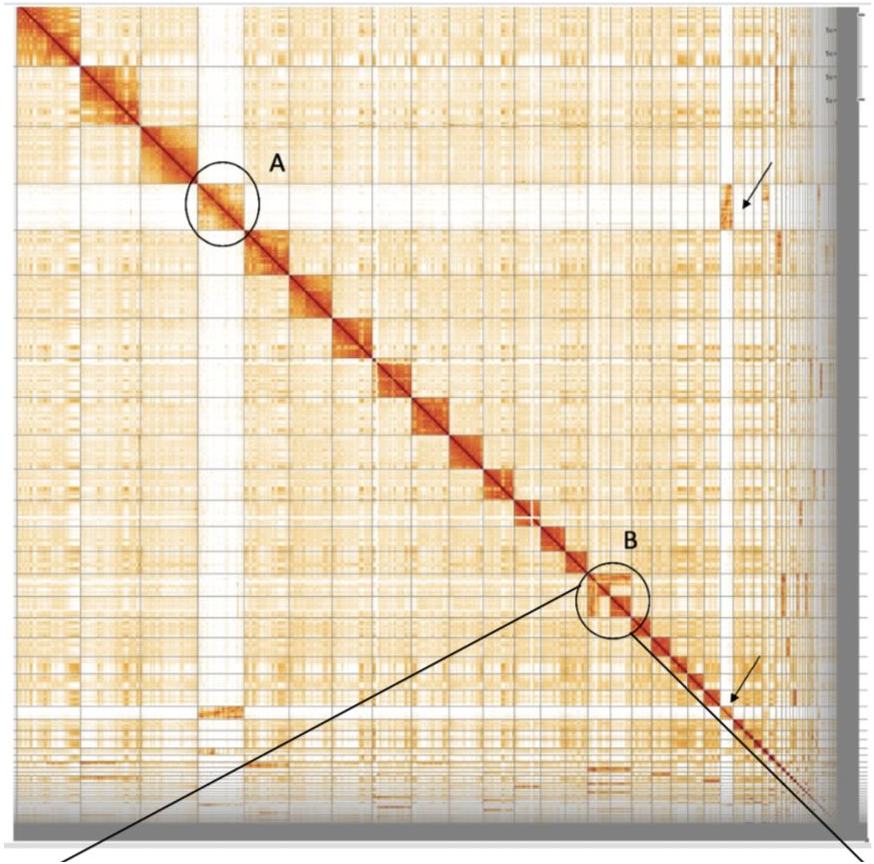


Figure 5. Genome assembly of *Pieris rapae*, ilPieRapa1.1: Hi-C contact map.

Hi-C contact map of the ilPieRapa1.1 assembly, visualised in HiGlass. Chromosomes are given in size order from left to right and top to bottom.

YOU DO MORE THAN SCAFFOLDING WITH HI-C: YOU SEE BIOLOGY



Choloepus didactylus VGP

Non-curated output

3.2 Gb, 281 scaffolds, N50 = 161 Mb

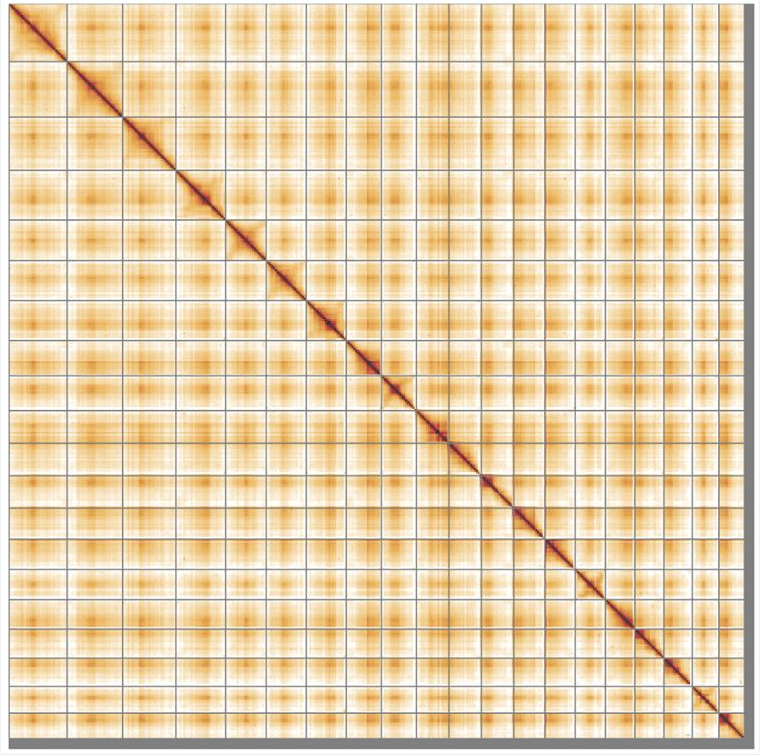


Figure 5. Genome assembly of *Ilex aquifolium*, drIleAqui2.1: Hi-C contact map of the drIleAqui2.1 assembly, visualised using HiGlass.

YaHS: yet another Hi-C scaffolding tool

Chenxi Zhou[1,2], **Shane A. McCarthy**[1,2], **and Richard Durbin**[1,2,*]

[1] Department of Genetics, University of Cambridge, Downing Street, Cambridge, CB2 3EH, UK
[2] Wellcome Sanger Institute, Wellcome Genome Campus, Hinxton, Cambridge, CB10 1SA, UK
[*] Correspondence: rd109@cam.ac.uk

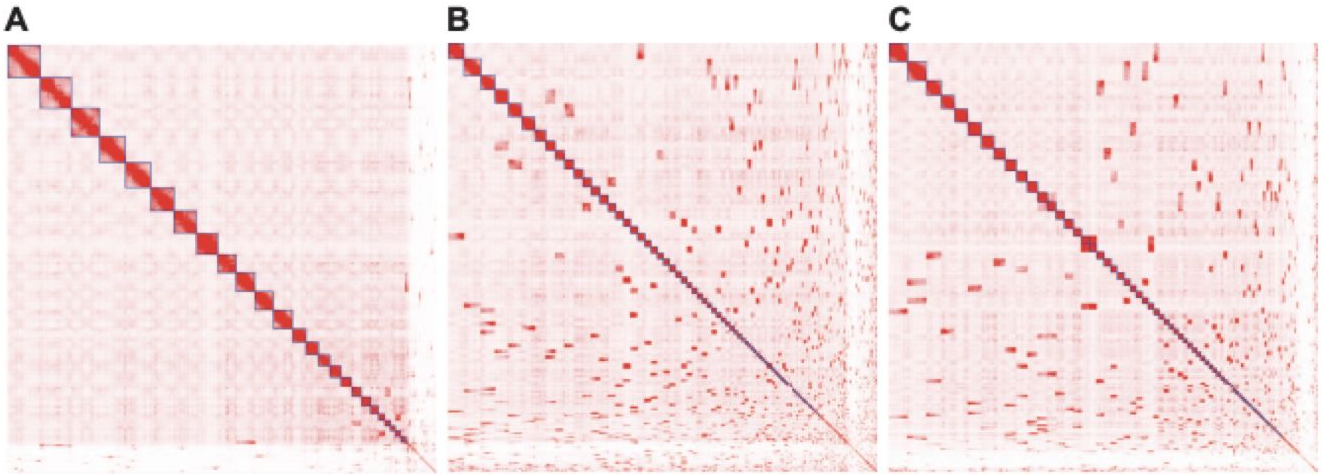


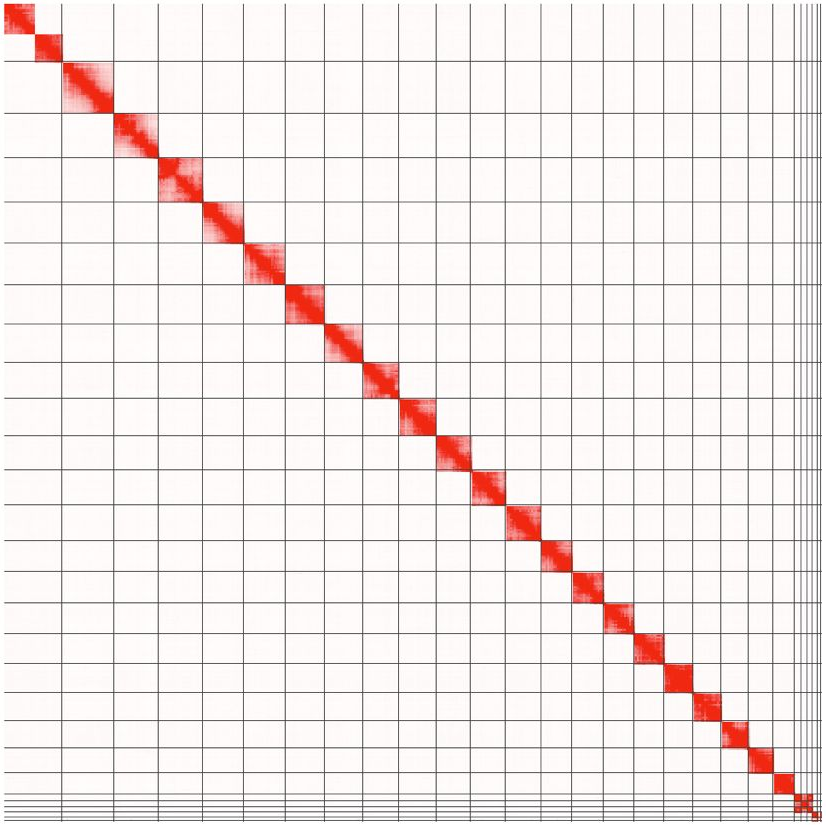
Figure 1. Hi-C contact maps of genome assemblies constructed with YaHS (**A**), SALSA2 (**B**) and pin_hic (**C**) for the simulated T2T data without contig errors. The blocks highlighted with blue squares in diagonal line are scaffolds. The contact maps were plotted with Juicebox (Durand *et al.*, 2016).

HI-C: DETECTING MISASSEMBLES

Look at me!!!!

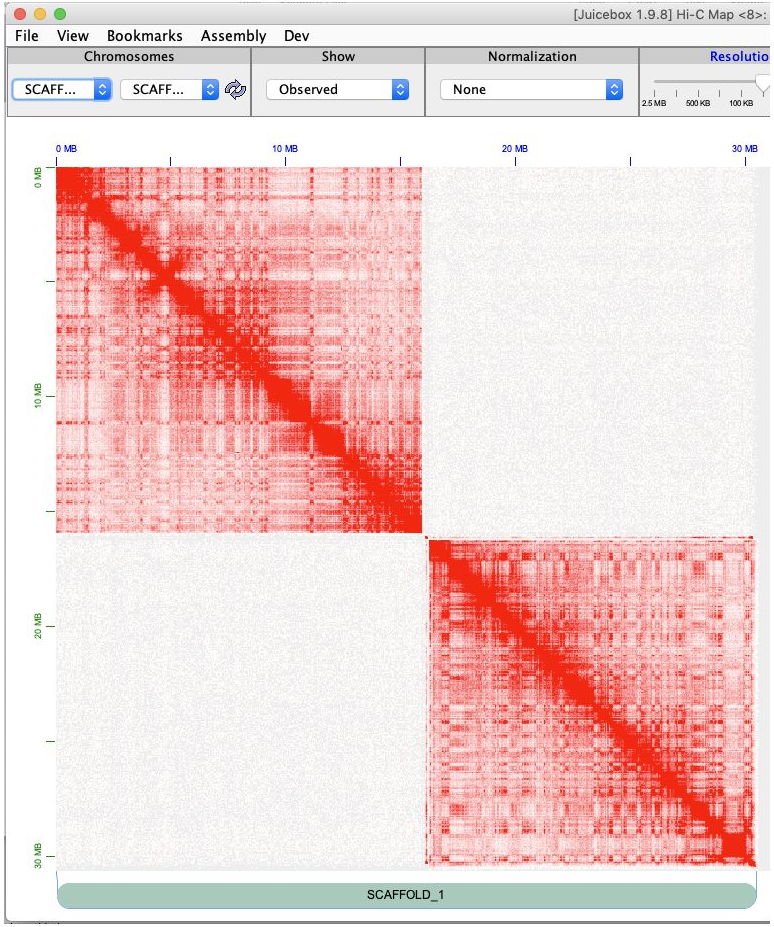
Lycaena phlaeas - ilLycPhla1

Darwin TREE of LIFE



HI-C: DETECTING MISASSEMBLES

Lycaena phlaeas - ilLycPhla1

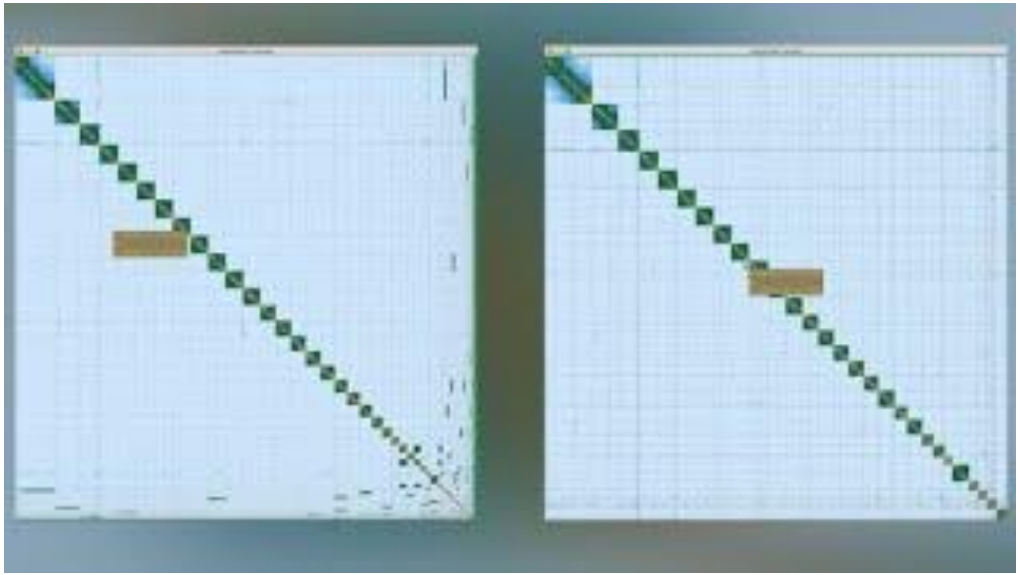


Darwin TREE of LIFE

Resources for the Sanger Grit curation team

https://gitlab.com/wtsi-grit/rapid-curation/-/blob/main/Interpreting_HiC_Maps_guide.pdf

https://gitlab.com/wtsi-grit/rapid-curation/-/blob/main/PretextView%20-%20Tutorial.pdf



https://www.youtube.com/watch?v=3IL2Q4f3k3I

Phase 1 VGP Genomes: 1st data release of 15 genomes, 14 species

Mammals (4 species)


GREATER HORSESHOE BAT


SPEAR-NOSED BAT


CANADIAN LYNX


PLATYPUS

Birds (3 species) 4 genomes)


ANNA'S HUMMINGBIRD


ZEBRA FINCH (male) (female)


KAKAPO


Dedicated to Jane, the Kakapo parrot

Reptiles (1 species)


GOODE'S DESERT TORTOISE

Amphibians (1 species)


TWO-LINED CAECILIAN

Fishes (5 species)


FLIER CICHLID


EASTERN HAPPY

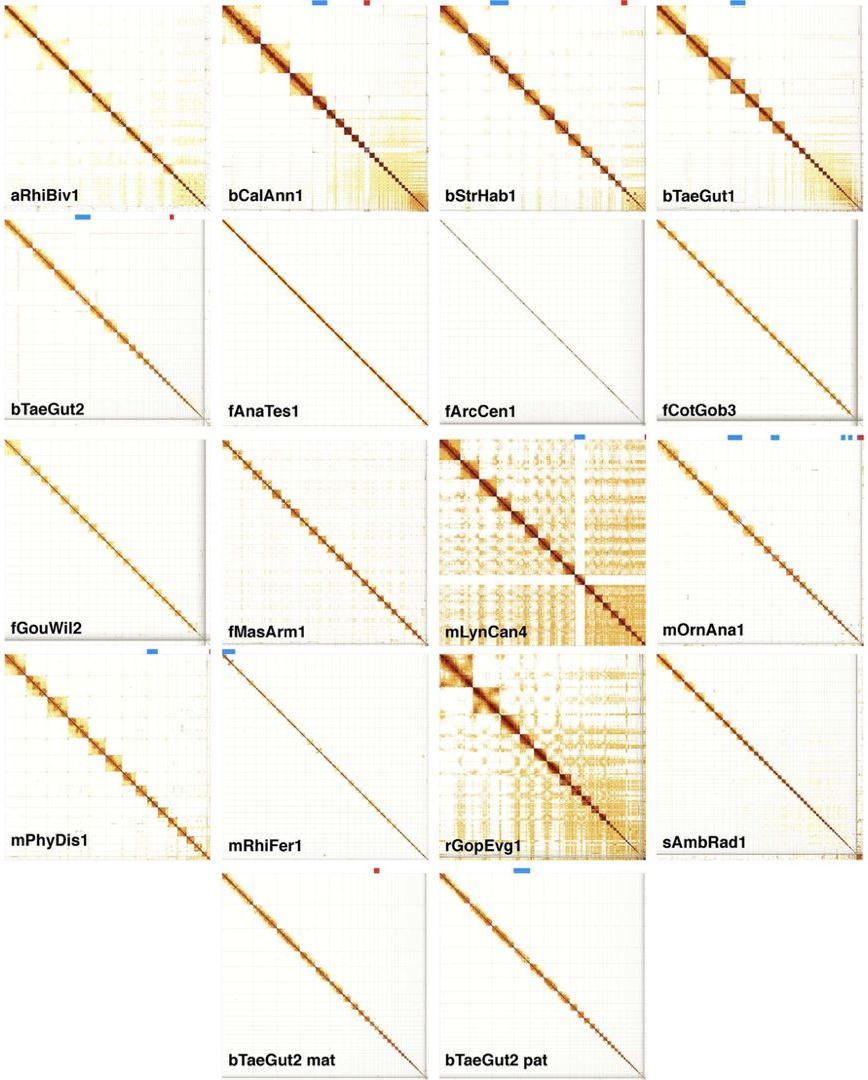

CLIMBING PERCH


TIRE TRACK EEL


BLUNT-SNOUTED CLINGFISH

thorny skate

six major vertebrate classes, with a wide diversity of genomic characteristics.



Article | Open Access | Published: 28 April 2021

Towards complete and error-free genome assemblies of all vertebrate species

Arang Rhie, Shane A. McCarthy, Olivier Fedrigo, Joana Damas, Giulio Formenti, Sergey Koren, Marcela Uliano-Silva, William Chow, Arkarachai Fungtammasan, Juwan Kim, Chul Lee, Byung June Ko, Mark Chaisson, Gregory L. Gedman, Lindsey J. Cantin, Francoise Thibaud-Nissen, Leanne Haggerty, Iliana Bista, Michelle Smith, Bettina Haase, Jacquelyn Mountcastle, Sylke Winkler, Sadye Paez, Jason Howard, Sonja C. Vernes, Tanya M. Lama, Frank Grutzner, Wesley C. Warren, Christopher N. Balakrishnan, Dave Burt, Julia M. George, Matthew T. Biegler, David Iorns, Andrew Digby, Daryl Eason, Bruce Robertson, Taylor Edwards, Mark Wilkinson, George Turner, Axel Meyer, Andreas F. Kautt, Paolo Franchini, H. William Detrich III, Hannes Svardal, Maximilian Wagner, Gavin J. P. Naylor, Martin Pippel, Milan Malinsky, Mark Mooney, Maria Simbirsky, Brett T. Hannigan, Trevor Pesout, Marlys Houck, Ann Misuraca, Sarah B. Kingan, Richard Hall, Zev Kronenberg, Ivan Sović, Christopher Dunn, Zemin Ning, Alex Hastie, Joyce Lee, Siddarth Selvaraj, Richard E. Green, Nicholas H. Putnam, Ivo Gut, Jay Ghurye, Erik Garrison, Ying Sims, Joanna Collins, Sarah Pelan, James Torrance, Alan Tracey, Jonathan Wood, Robel E. Dagnew, Dengfeng Guan, Sarah E. London, David F. Clayton, Claudio V. Mello, Samantha R. Friedrich, Peter V. Lovell, Ekaterina Osipova, Farooq O. Al-Ajli, Simona Secomandi, Heebal Kim, Constantina Theofanopoulou, Michael Hiller, Yang Zhou, Robert S. Harris, Kateryna D. Makova, Paul Medvedev, Jinna Hoffman, Patrick Masterson, Karen Clark, Fergal Martin, Kevin Howe, Paul Flicek, Brian P. Walenz, Woori Kwak, Hiram Clawson, Mark Diekhans, Luis Nassar, Benedict Paten, Robert H. S. Kraus, Andrew J. Crawford, M. Thomas P. Gilbert, Guojie Zhang, Byrappa Venkatesh, Robert W. Murphy, Klaus-Peter Koepfli, Beth Shapiro, Warren E. Johnson, Federica Di Palma, Tomas Marques-Bonet, Emma C. Teeling, Tandy Warnow, Jennifer Marshall Graves, Oliver A. Ryder, David Haussler, Stephen J. O'Brien, Jonas Korlach, Harris A. Lewin, Kerstin Howe ✉, Eugene W. Myers ✉, Richard Durbin ✉, Adam M. Phillippy ✉ & Erich D. Jarvis ✉ -Show fewer authors

Nature **592**, 737–746 (2021) | Cite this article

I have my assembly, how do I know its correct?

Final checks:

- Does final assembled size corresponds to predicted genome size?
- How are my general metrics? How is my BUSCO?
- How does my Hi-C heatmap looks like? Clean? Correct karyotype? Any scaffolding mistakes?
- Reads coverage and **Merqury (important!)**

Minimal supplementary materials for your genome paper: (i) kmer plot of your data (genomescope plot), (ii) general assembly stats, (iii) merqury plots, (iv) busco and (v) HiC heatmap.

HOW TO IDENTIFY RETAINED HAPLOTIGS? PURGING AND MERQURY!!!!!

Bioinformatics, 36(9), 2020, 2896–2898
doi: 10.1093/bioinformatics/btaa025
Advance Access Publication Date: 23 January 2020
Applications Note

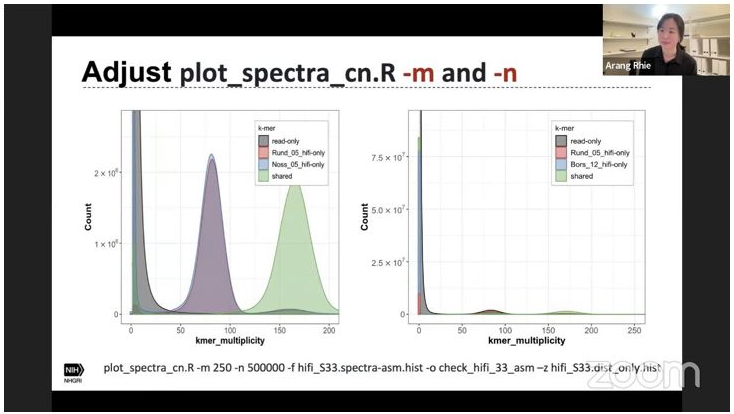
OXFORD

Genome analysis

Identifying and removing haplotypic duplication in primary genome assemblies

Dengfeng Guan[1,2], Shane A. McCarthy [2], Jonathan Wood[3], Kerstin Howe [3], Yadong Wang[1,*] and Richard Durbin [2,3,*]

[1]Department of Computer Science and Technology, Center for Bioinformatics, Harbin Institute of Technology, Harbin 150001, China, [2]Department of Genetics, University of Cambridge, Cambridge CB2 3EH, UK and [3]Wellcome Sanger Institute, Wellcome Genome Campus, Cambridge CB10 1SA, UK



Rhie *et al. Genome Biology* (2020) 21:245
https://doi.org/10.1186/s13059-020-02134-9

Genome Biology

METHOD Open Access

Merqury: reference-free quality, completeness, and phasing assessment for genome assemblies

Arang Rhie*, Brian P. Walenz, Sergey Koren and Adam M. Phillippy

* Correspondence: arang.rhie@nih.gov
Genome Informatics Section, Computational and Statistical Genomics Branch, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD, USA

Abstract

Recent long-read assemblies often exceed the quality and completeness of available reference genomes, making validation challenging. Here we present Merqury, a novel tool for reference-free assembly evaluation based on efficient k-mer set operations. By comparing k-mers in a de novo assembly to those found in unassembled high-accuracy reads, Merqury estimates base-level accuracy and completeness. For trios, Merqury can also evaluate haplotype-specific accuracy, completeness, phase block continuity, and switch errors. Multiple visualizations, such as k-mer spectrum plots, can be generated for evaluation. We demonstrate on both human and plant genomes that Merqury is a fast and robust method for assembly validation.

Keywords: Genome assembly, Assembly validation, Benchmarking, K-mers, Haplotype phasing, Trio binning

Research | Open access | Published: 27 September 2022

Widespread false gene gains caused by duplication errors in genome assemblies

Byung June Ko, Chul Lee, Juwan Kim, Arang Rhie, Dong Ahn Yoo, Kerstin Howe, Jonathan Wood, Seoae Cho, Samara Brown, Giulio Formenti, Erich D. Jarvis ✉ & Heebal Kim ✉

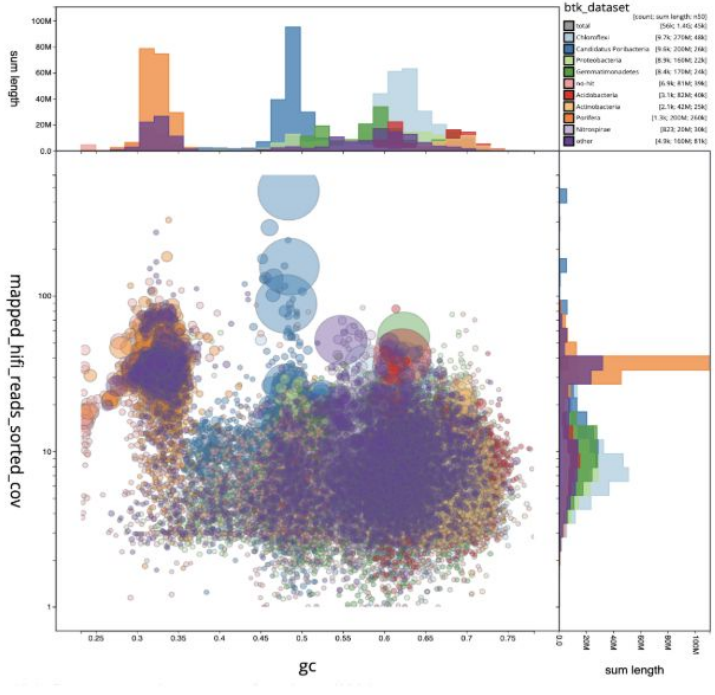
Genome Biology **23**, Article number: 205 (2022) | Cite this article

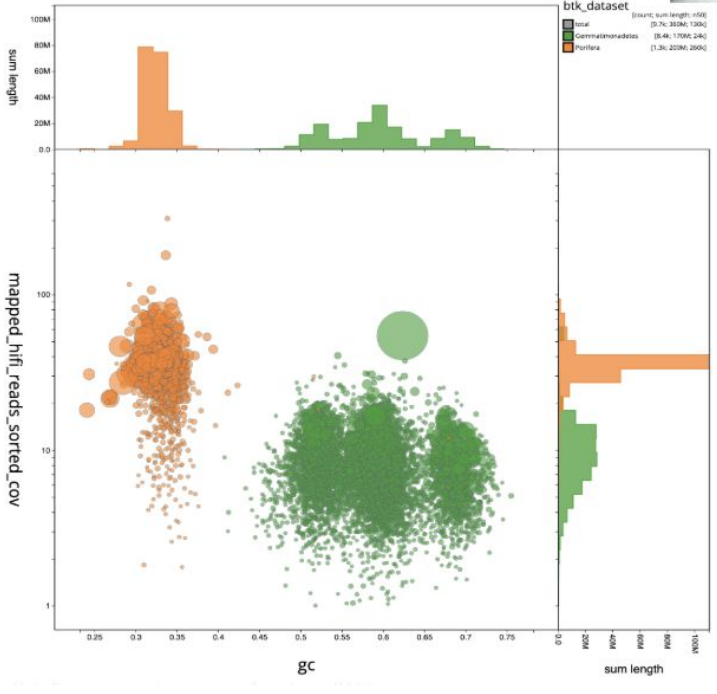
“Whole genome alignments revealed that 4 to 16% of the sequences are falsely duplicated in the previous assemblies, impacting hundreds to thousands of genes. These lead to overestimated gene family expansions.

The main source of the false duplications is heterotype duplications, where the haplotype sequences were relatively more divergent than other parts of the genome leading the assembly algorithms to classify them as separate genes or genomic regions.” Kim et al, 2022

More than the target species…

BlobToolKit analysis of ***Xestospongia muta*** (giant barrel sponge) odXesMuta1
github.com/blobtoolkit

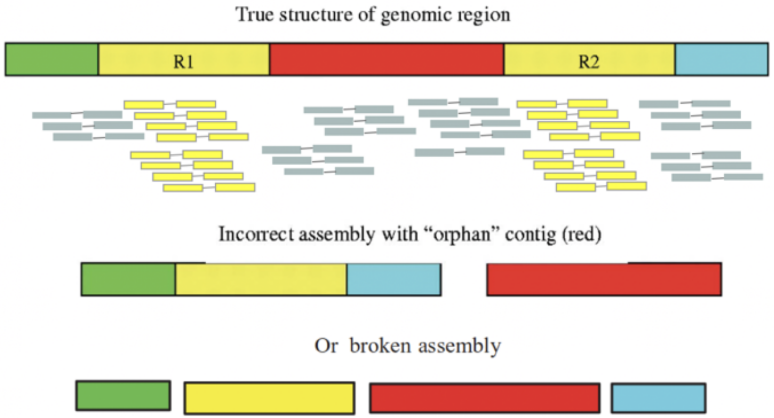




Long reads will always outperform short reads

You can do good science with short reads assemblies, but you must know the limitations so you don't make erroneous, unlikely predictions with it.

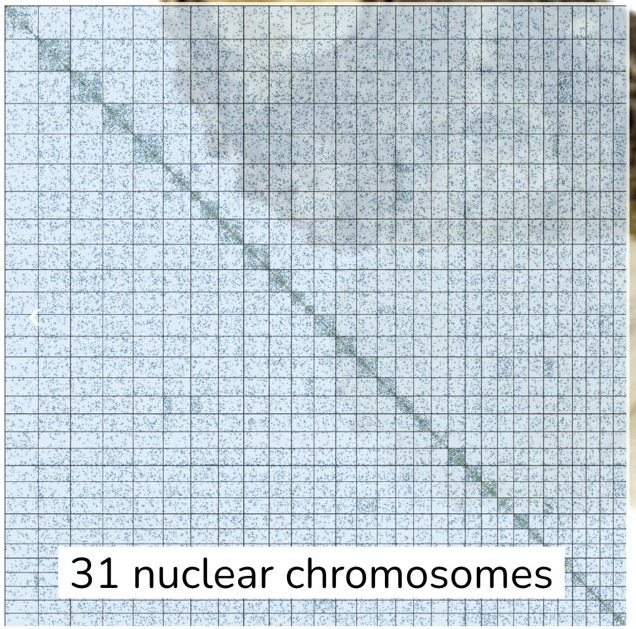
- Challenges to understand genome duplication: paralogs collapsed, genes fragmented, wrong synteny (chimeric scaffolds)
- Gene family expansion and retraction analyses over or underestimated
- Difficulty studying repeats, telomeres, centromeres, chromosome architecture



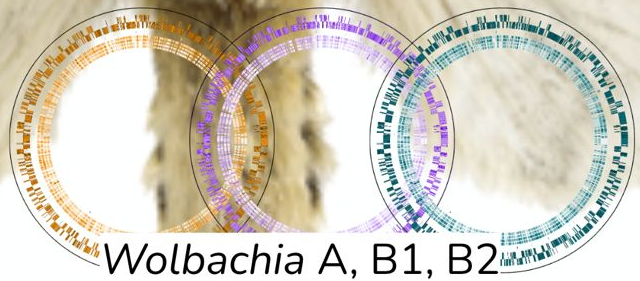
Modified from Salzberg S L , and Yorke J A Bioinformatics 2005;21:4320-4321

Assembling genomes of target *and* cobiome

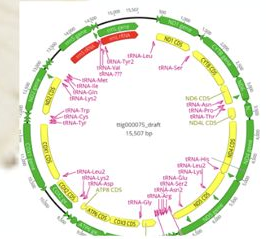
Phalera bucephala: one moth, five genomes



31 nuclear chromosomes



Wolbachia A, B1, B2



mitochondrion

Emmelien Vancaester PLoS Biology, Jan 2023 *https://doi.org/10.1371/journal.pbio.3001972*

wellcome sanger institute

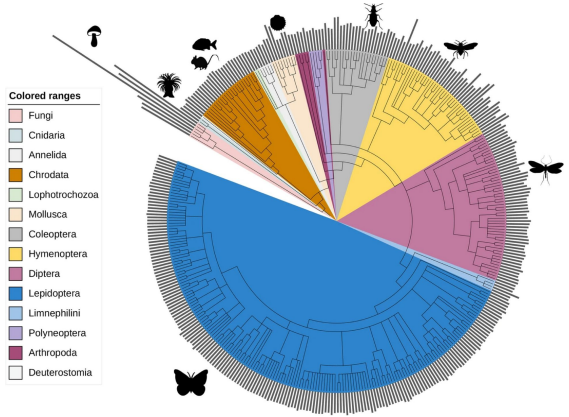
Mito and chloroplast assembly with Long Reads

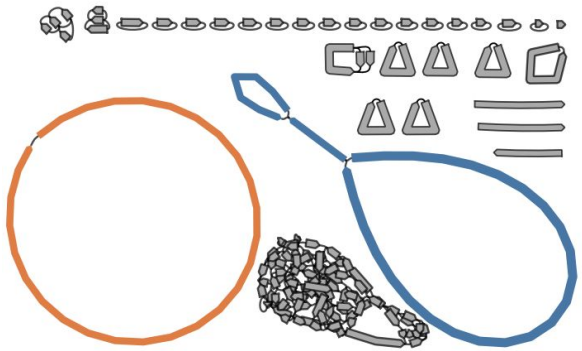
Software | Open access | Published: 18 July 2023

MitoHiFi: a python pipeline for mitochondrial genome assembly from PacBio high fidelity reads

Marcela Uliano-Silva ✉, João Gabriel R. N. Ferreira, Ksenia Krasheninnikova, Darwin Tree of Life Consortium, Giulio Formenti, Linelle Abueg, James Torrance, Eugene W. Myers, Richard Durbin, Mark Blaxter & Shane A. McCarthy

BMC Bioinformatics **24**, Article number: 288 (2023) | Cite this article





Oatk: a de novo assembly tool for complex plant organelle genomes

Chenxi Zhou[1,2], Max Brown[2,3], Mark Blaxter[2], The Darwin Tree of Life Project Consortium[2], Shane A. McCarthy[1,2], and Richard Durbin[1,2,*]

[1]Department of Genetics, University of Cambridge, Downing Street, Cambridge, CB2 3EH, UK
[2]Wellcome Sanger Institute, Wellcome Genome Campus, Hinxton, Cambridge, CB10 1SA, UK
[3]Faculty of Science and Engineering, Anglia Ruskin University, East Road, Cambridge, CB1 1PT, UK
[*]Correspondence: rd109@cam.ac.uk

Open data release

Project portals	https://portal.darwintreeoflife.org/ https://portal.aquaticsymbiosisgenomics.org
Raw data & assembly progress	https://tolqc.cog.sanger.ac.uk/
Genome Notes	https://wellcomeopenresearch.org/gateways/treeoflife
Ensembl annotation browser	https://projects.ensembl.org/darwin-tree-of-life/
Interactive genome viewer	https://blobtoolkit.genomehubs.org
Global coordination (GoaT)	https://goat.genomehubs.org https://goat.genomehubs.org/projects/DTOL https://goat.genomehubs.org/projects/ASG https://goat.genomehubs.org/projects/PSYCHE

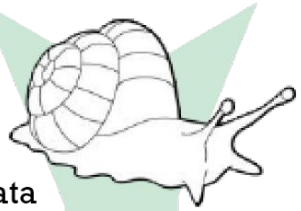
Open data release

Tree of Life QC
e!Ensembl
BlobToolKit

Project portals	https://portal.darwintreeoflife.org/ https://portal.aquaticsymbiosisgenomics.org
Raw data & assembly progress	https://tolqc.cog.sanger.ac.uk/
Genome Notes	https://wellcomeopenresearch.org/gateways/treeoflife
Ensembl annotation browser	https://projects.ensembl.org/darwin-tree-of-life/
Interactive genome viewer	https://blobtoolkit.genomehubs.org
Global coordination (GoaT)	https://goat.genomehubs.org https://goat.genomehubs.org/projects/DTOL https://goat.genomehubs.org/projects/ASG https://goat.genomehubs.org/projects/PSYCHE

Slides here

sample acquisition and metadata recording

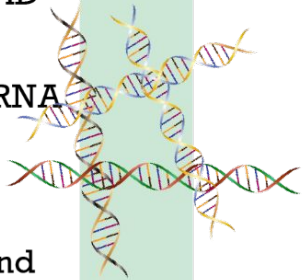


morphological ID

Cornu aspersum

DNA barcode validating ID

DNA and RNA extraction

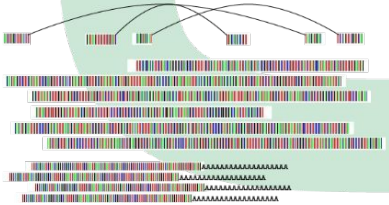


genome and transcriptome sequencing

Hi-C

long read

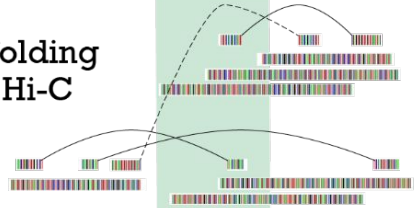
RNASeq



database submission ENA

curation

scaffolding with Hi-C



primary assembly of long reads

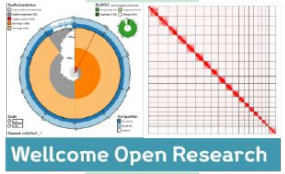
Tree of Life QC

data quality assurance

e!Ensembl

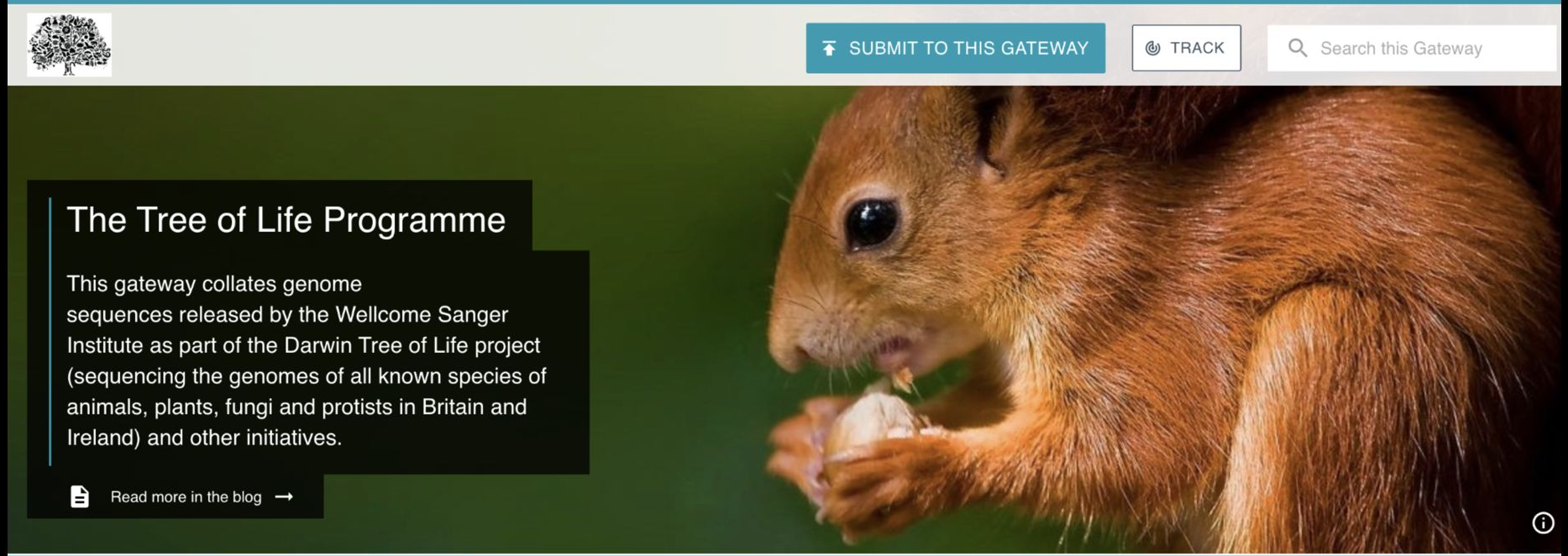
gene finding and feature annotation

open data portals



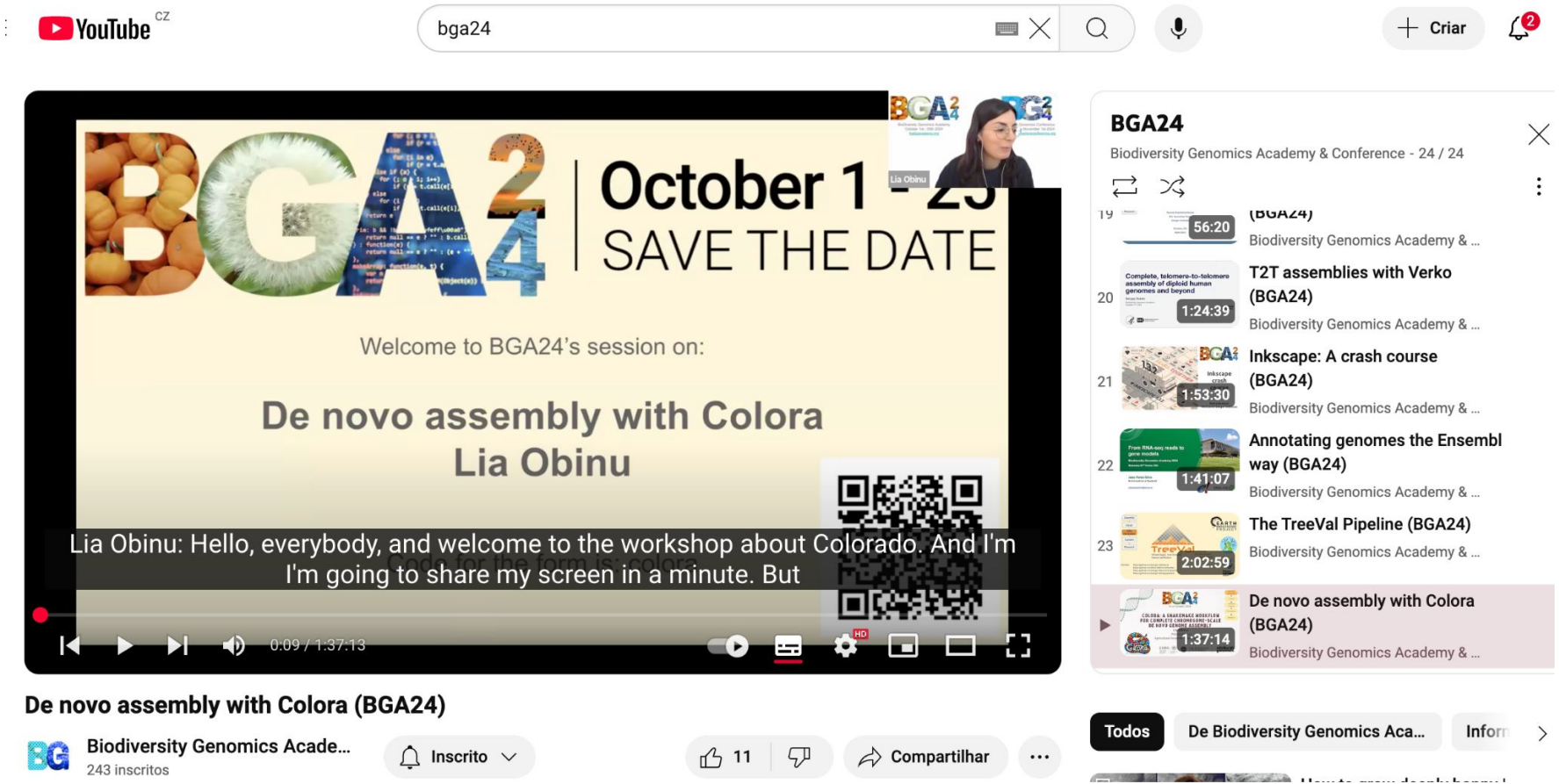
publication of genome note

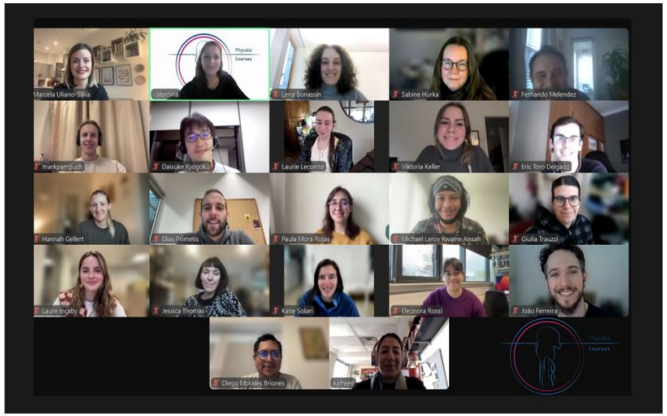
https://wellcomeopenresearch.org/treeoflife



1497 papers in the collection so far

Studying further: Biodiversity Genomics Academy

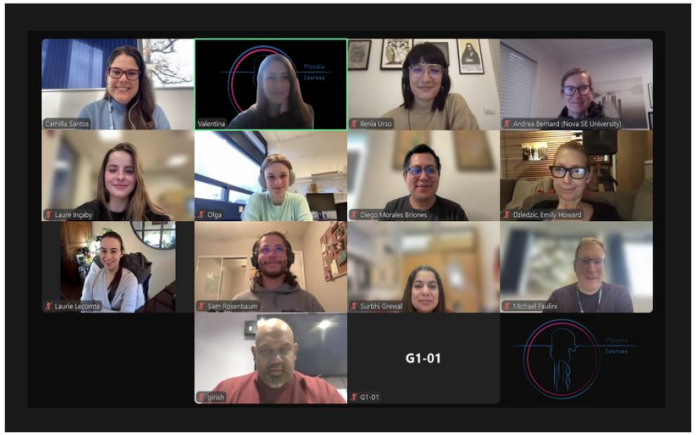




5TH EDITION GENOME ASSEMBLY USING PACBIO AND HI-C

ONLINE, 4-8 NOVEMBER 2024

20 attendees
16 research institutes
14 different countries



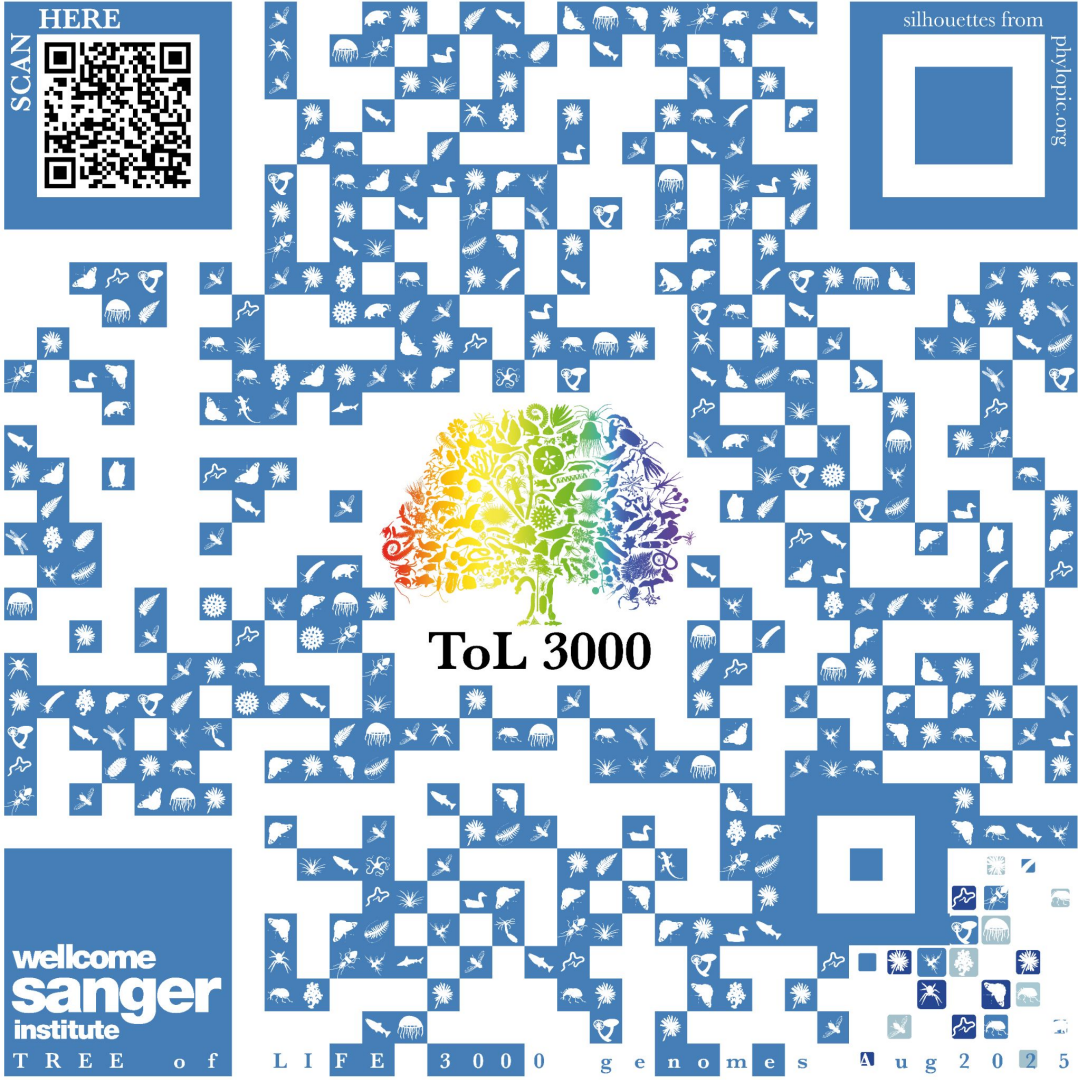
GENOME MANUAL CURATION

ONLINE, 11-15 NOVEMBER 2024

11 attendees
9 research institutes
8 different countries

Physalia Courses

physalia-courses.org



In December 2025, the Tree of Life programme released

3,571

reference genome assemblies to INSDC

Genomes assembled from

- 550.3 Tb PacBio HiFi
- 2108.3 Tb Hi-C
- 80.8 Tb RNA-Seq (3341 species)

It takes a village…

Tree of Life teams - Mark Blaxter
ToL Sample Management, ToL Core Lab, ToL Assembly, GRIT, Delivery & Operations, ToL faculty teams

Anna Kovalevskaia
Priyanka Sethu Raman
Thomas Mathers
Sarah Pelan
Manuela Kieninger
Julia Gries
Dominic Absolon
Zeynep Goktan
Barnaby Dingemans
Witold Morek
Cibele Sotero-Caio
Jessie Jay
Meyer Marco
Maneno Baravuga
Ksenia Krasheninnikova
Abitha Thomas
Jo Wood
Downie Jim
Eerik Aunin
Remi Clare
Eva van der Heijden
Adam Bates
Alex Makunin
Haoyu Niu
Noah Gettle
Anushka Mittal
Martin Wagah
Kerstin Howe
Luke Wilson
Katie Woodcock
Nicol Rueda
Halyna Yatsenko
Rebecca O'Brien
Witwicka Alicja
Seri Kitada
Victoria Mckenna
Molly Carter
Elizabeth Sinclair
Guoying Qi
Karen Brooks
Roz Malik
Edward Moulsdale
Lyndall Pereira da Conceicoa
Karin Näsvall
Beth Yates
Fiona Teltscher
Sunil Dogga
Camilla Santos
Amy Denton
Cibin Sadasivan Baby
Andrew Varley
Ian Still
Jemma Salmon
Joana Meier
Mark Blaxter
James Torrance
Logan Howat
Radka Platte
Clothilde Chenal
Francis Totanes
Manuel Batista
Erna King
Claudia Weber
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Matthieu Muffato
Chafin Tyler
Nathan Riley
Ying Sims
Damon-Lee Pointon
Iszy Clayton-Lucey
Amjad Khalaf
Ashish Mittal
Graeme Oatley
Kiernan Harding
Shane McCarthy
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Ore Francis
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Emmelien Vancaester
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Richard Challis
Ben Jackson
Kamil Jaron
Marcela Uliano-Silva
Charlotte Wright
Arif Maulana
Martha Mulongo
Raquel Vionette Do Amaral
Aleksandra Bliznina
Joanna Collins

Team301
All faculty teams

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SciOps, especially the Long Read Team and R&D

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& hundreds of engaged naturalists

ASG, VGP, ERGA
and other collaborators and collectors

Obrigada! Thank you! Grazie!

mu2@sanger.ac.uk