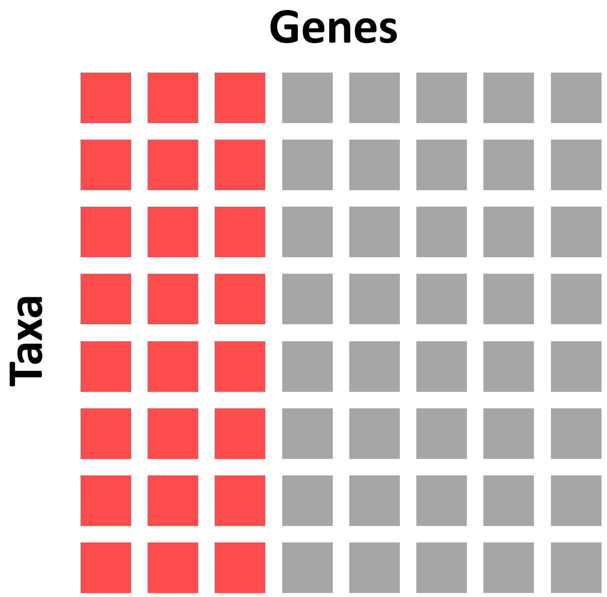
Lecture 3.2

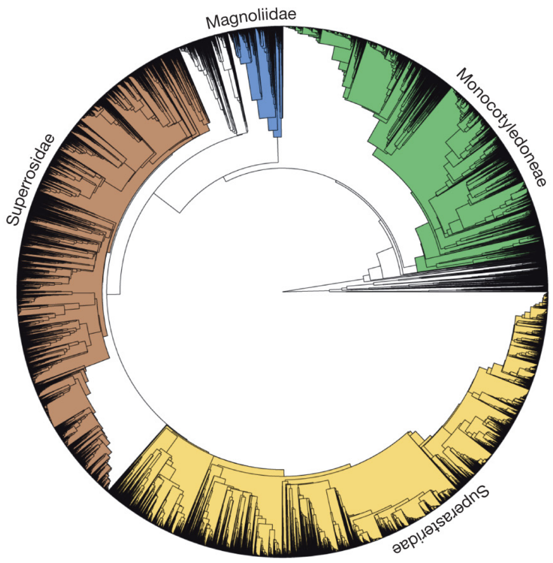
Phylogenomics

Analysing Large Data Sets

Large data sets



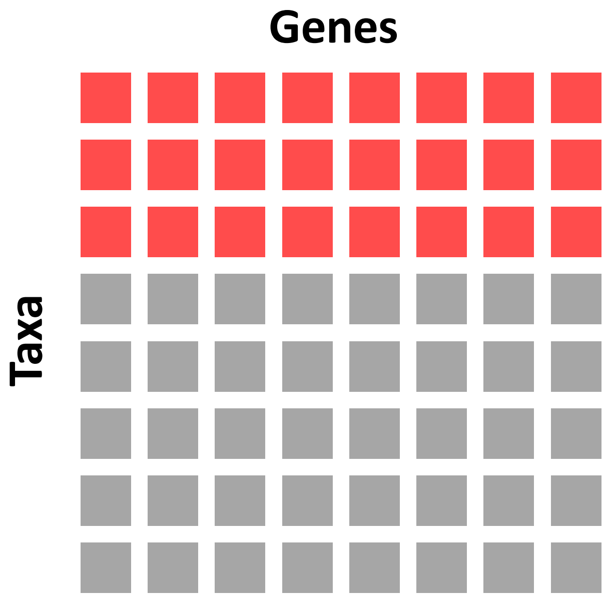
- Tree-space is extremely large
  - Efficient tree-searching heuristics



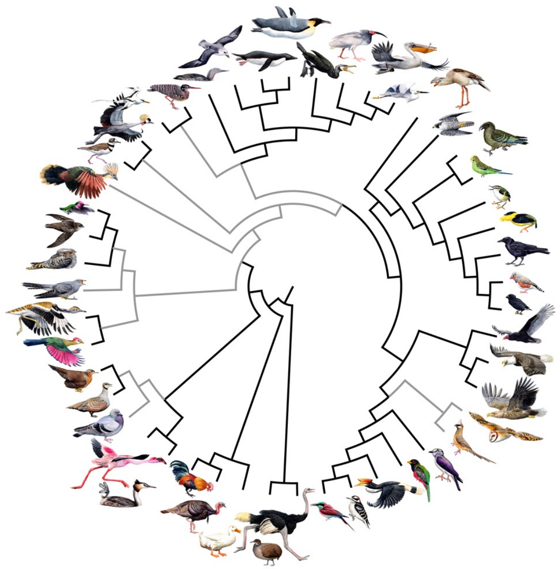
32,223 taxa
7 genes

Zanne *et al*. (2014) *Nature*

Large data sets

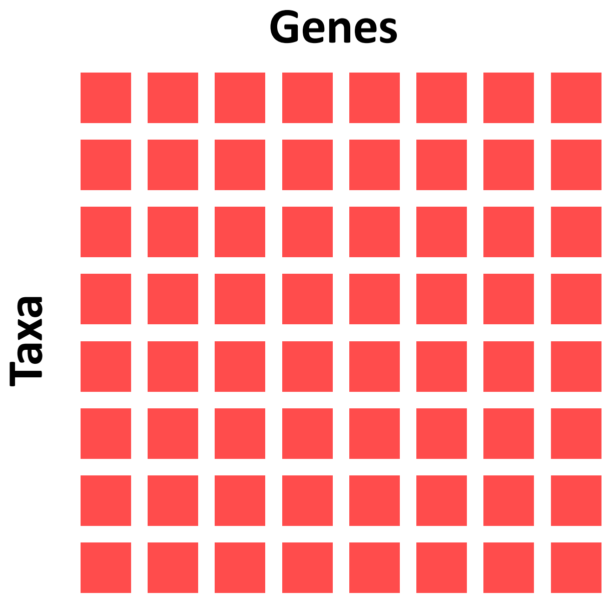


- Calculation of likelihood is expensive
  - Speed up by grouping sites with identical patterns
  - Approximate likelihood calculation
  - Multithreading/parallelisation



48 taxa
8,295 genes
Jarvis *et al*. (2014) *Science*

Large data sets

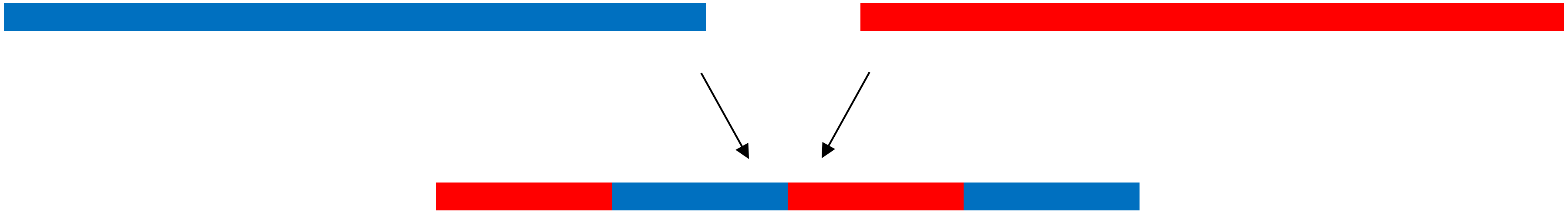


- Analysis is computationally expensive
- Consider filtering the data
  - Phylogenetic signal
  - Substitution saturation
  - Missing data
  - Model fit
  - Random subsample
- Dividing the tree into subtrees

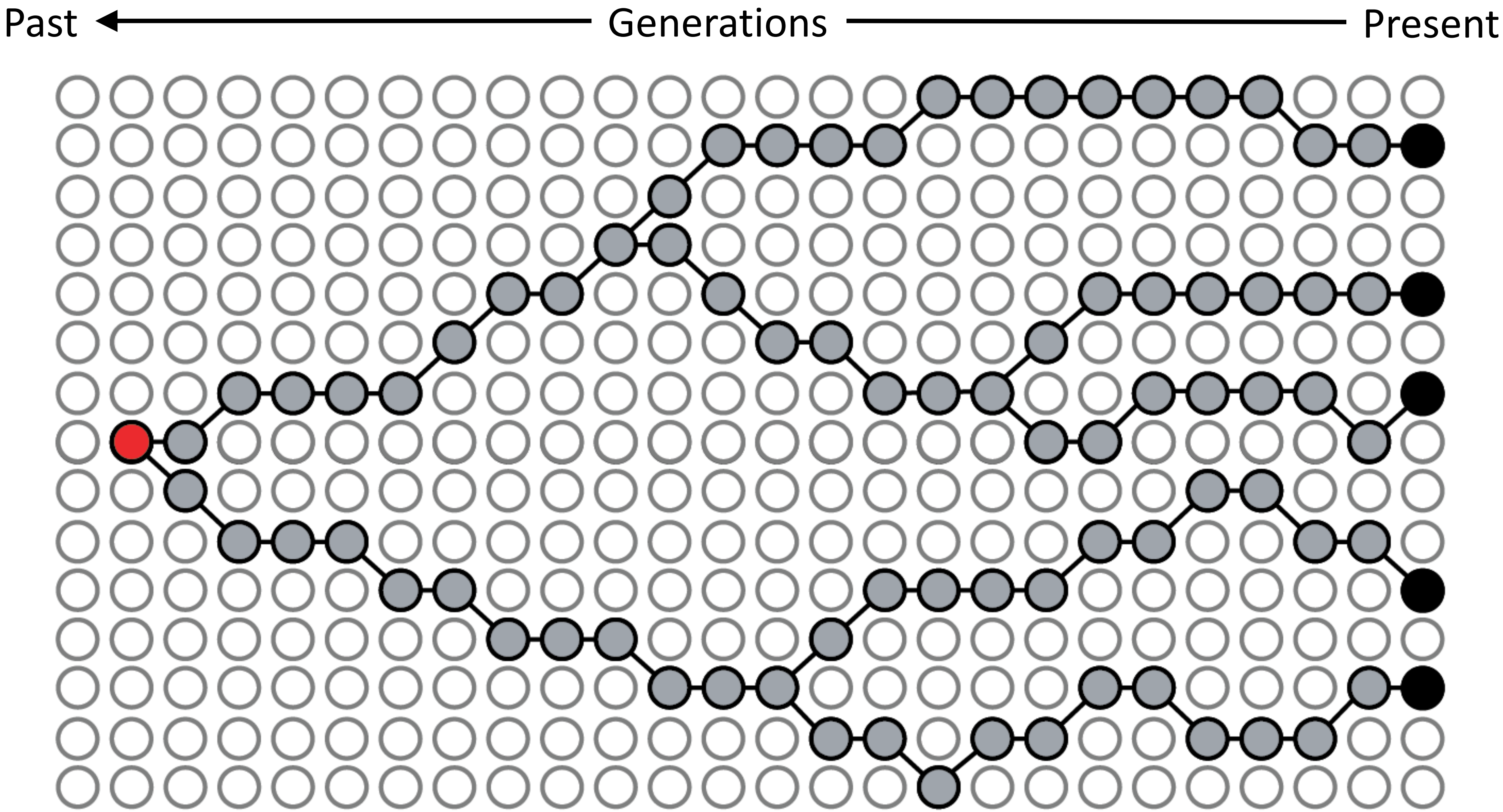
Gene Trees

Gene trees

- Many phylogenetic methods assume that there is a single tree that describes the evolution of the whole data set
- But recombination complicates this

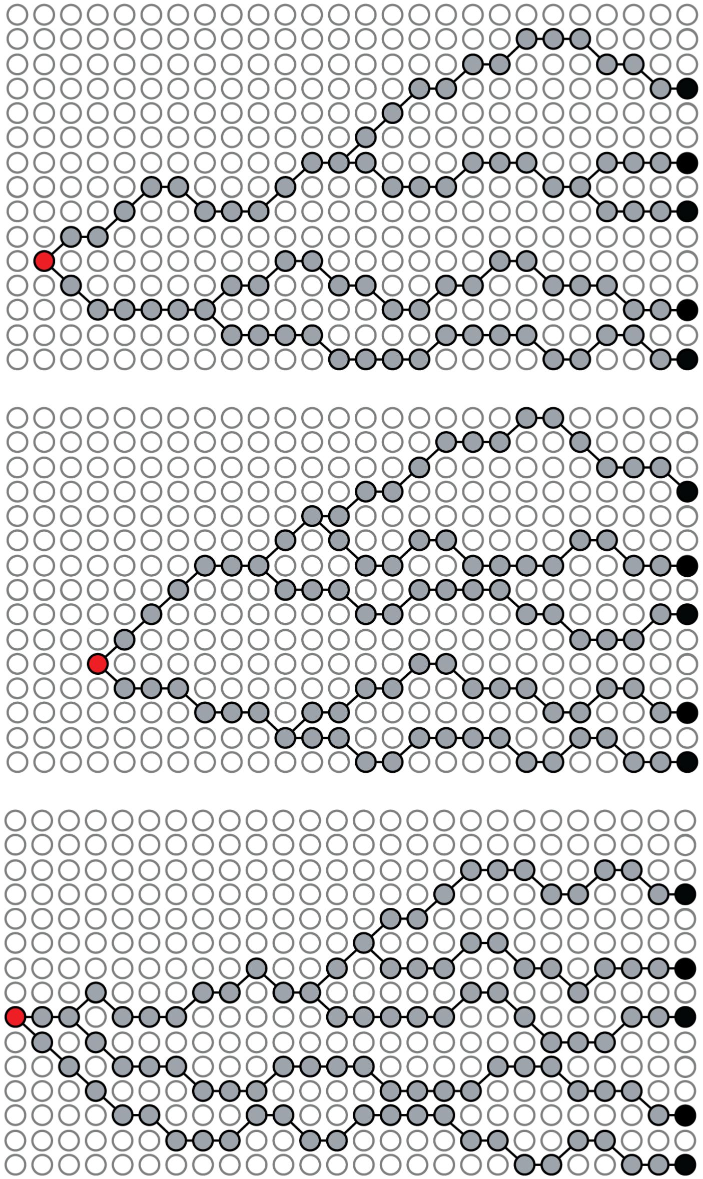


Coalescent theory

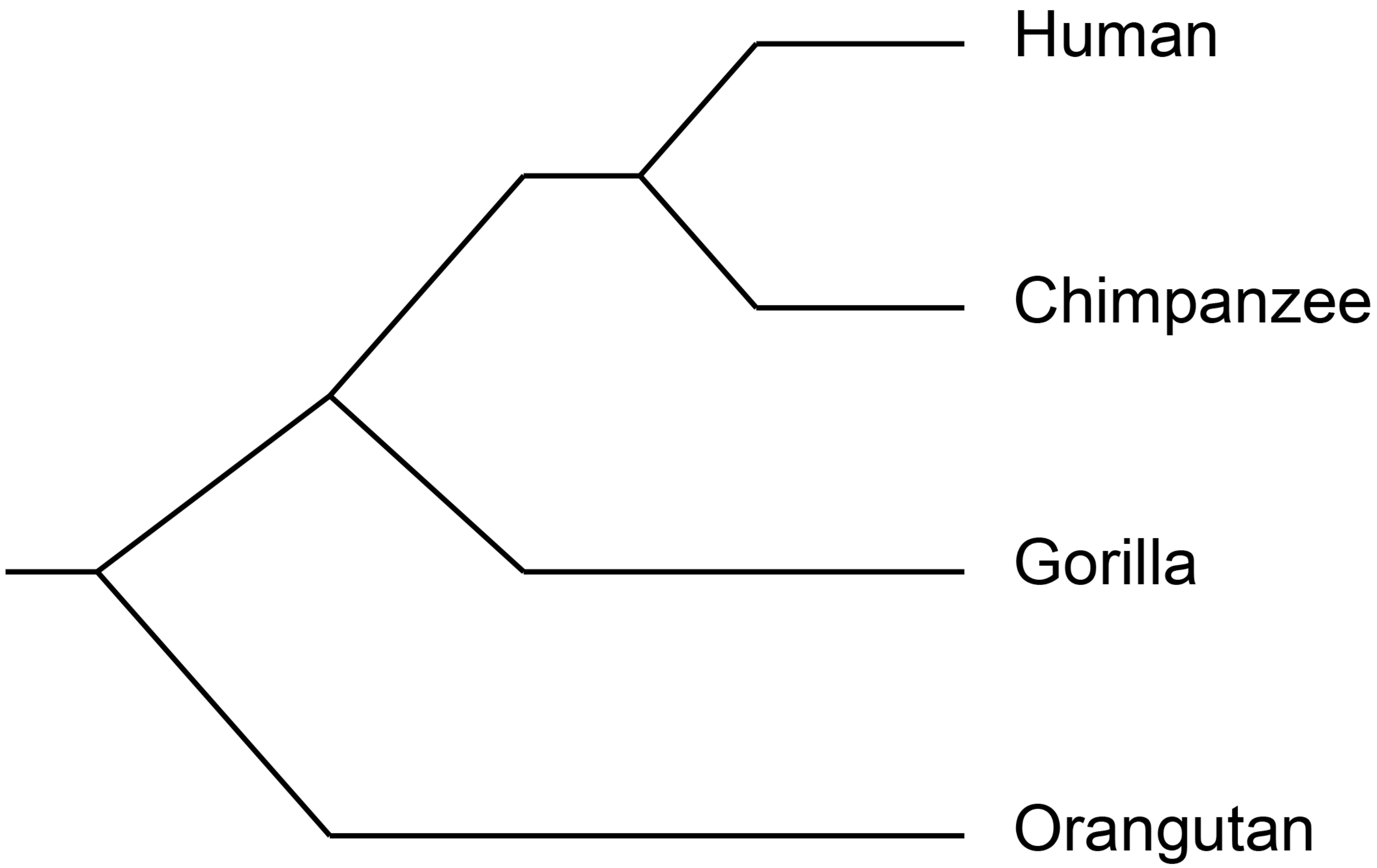


Gene trees in a species

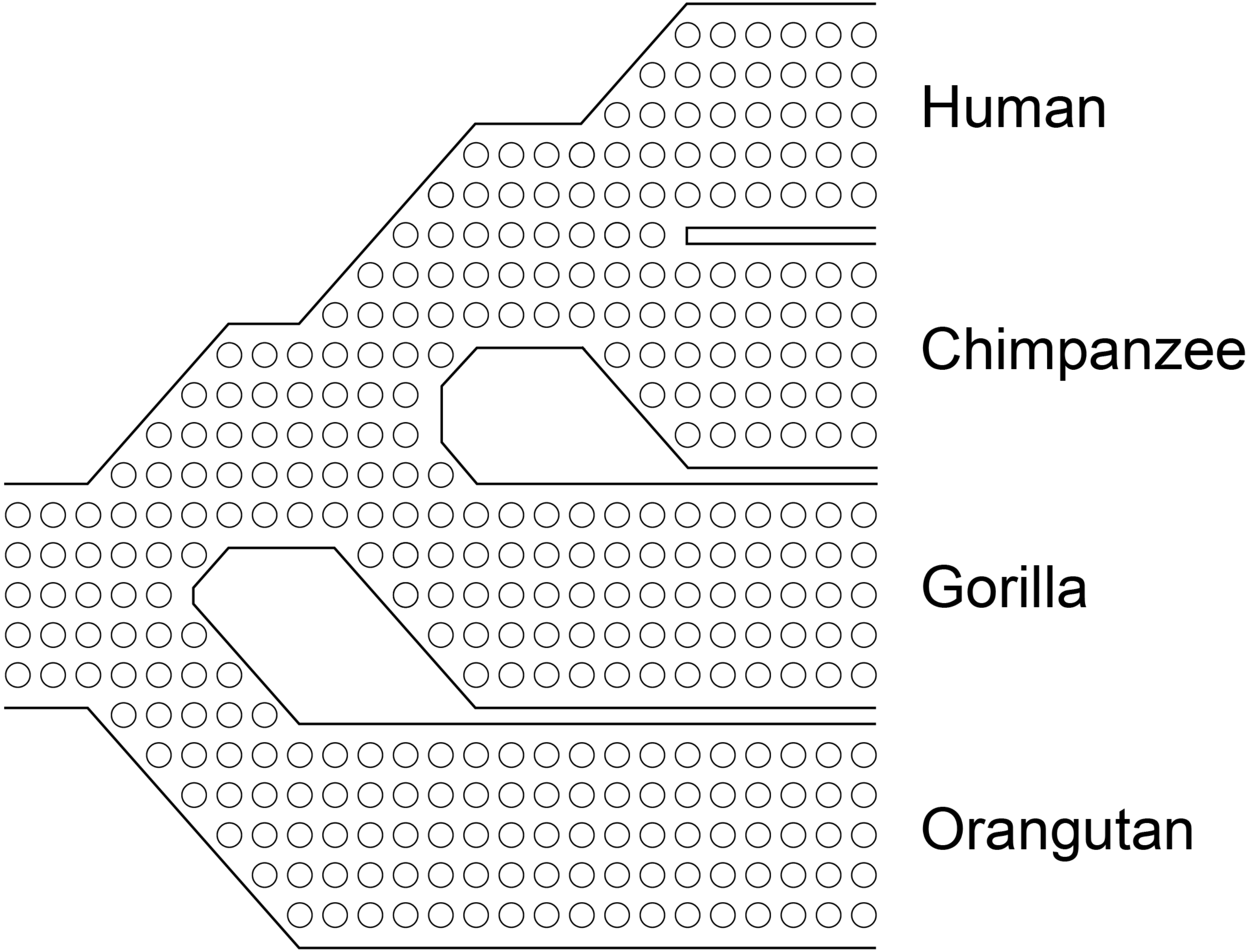
- Gene trees vary by chance among unlinked genes
  - Different trees
  - Different timescales



Species tree

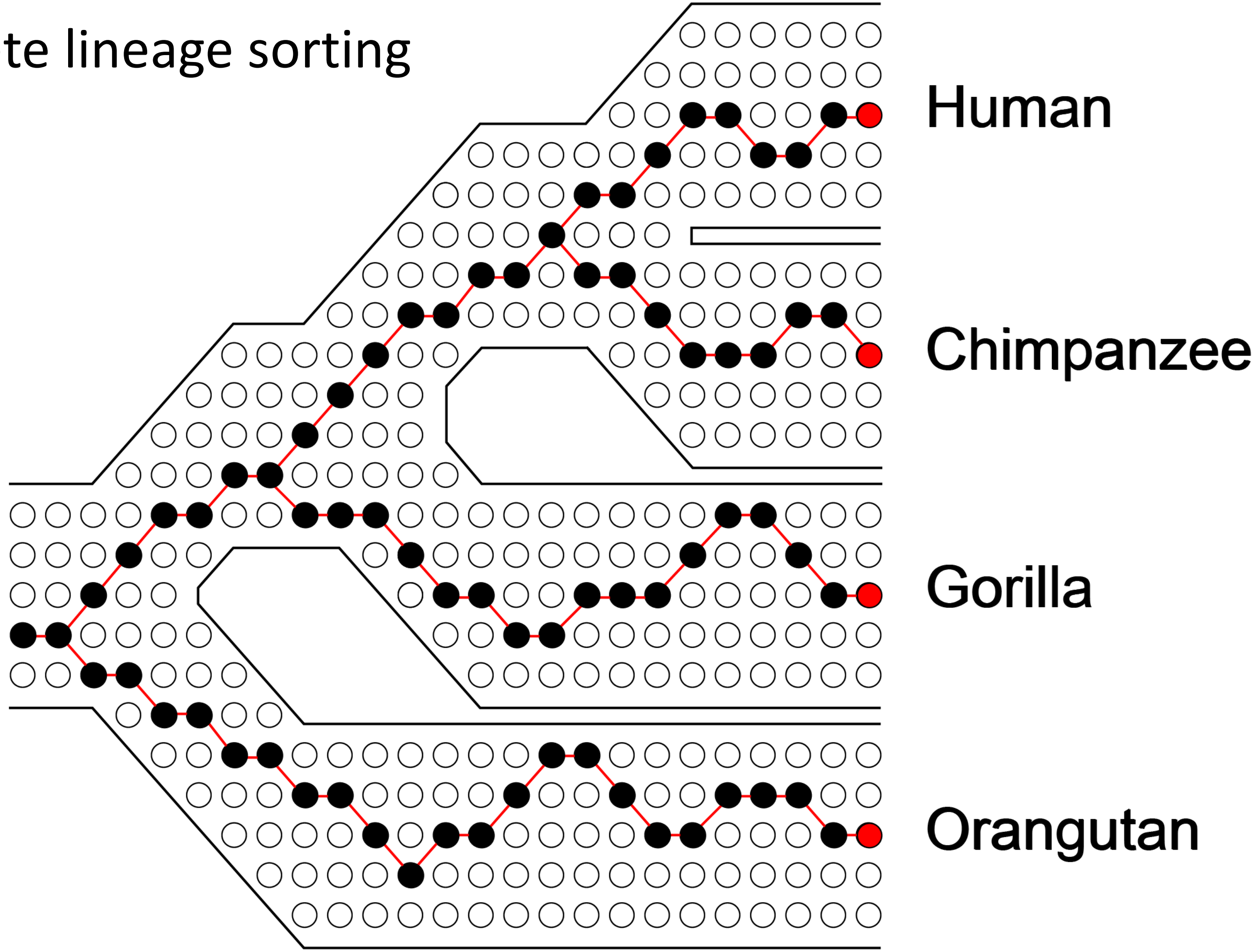


Multispecies coalescent



Gene tree (concordant)

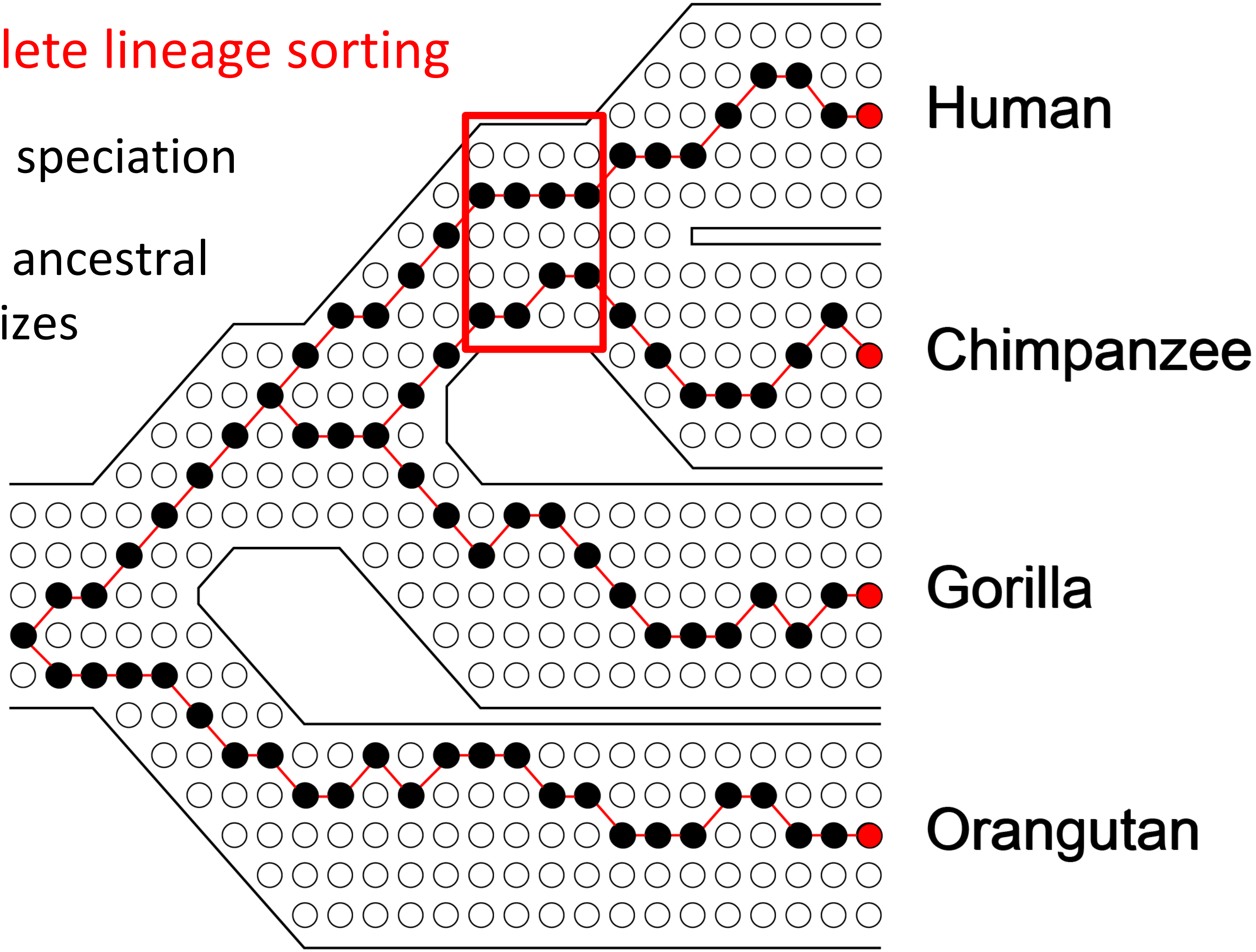
Complete lineage sorting



Gene tree (discordant)

Incomplete lineage sorting

- Rapid speciation
- Large ancestral pop sizes



Incongruence among gene trees

- Phylogenetic analyses of genome-scale data sets must deal with incongruence among gene trees

  - **Incomplete lineage sorting**
  - Different direction and strength of selection
  - Stochastic variation in the mutational process
  - Biases in nucleotide composition

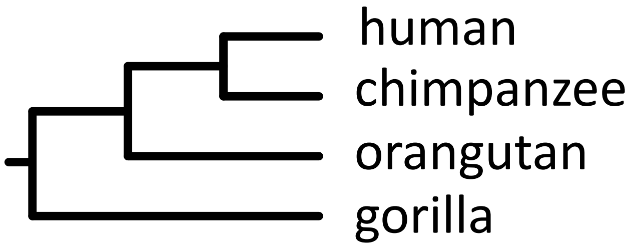
Inferring the species tree

- **Incomplete lineage sorting** can lead to gene trees that do not match the species tree
- We can infer the species tree from multiple gene trees even when they are incongruent
- Three approaches
  1. Consensus
  2. Concatenation
  3. Coalescent

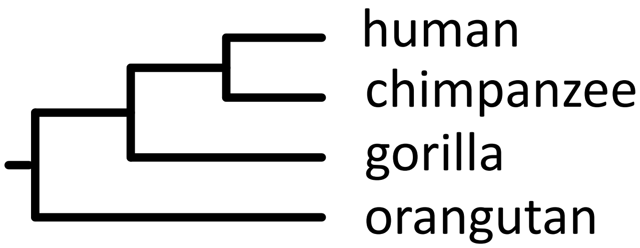
Species tree

1. **Consensus**

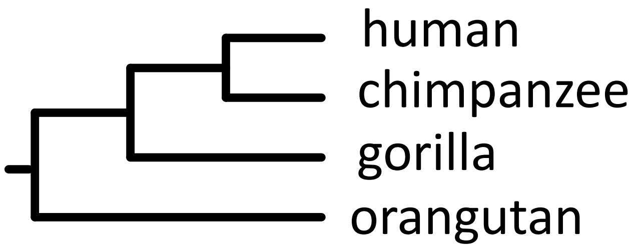
   Estimate genealogy from each gene and find the consensus



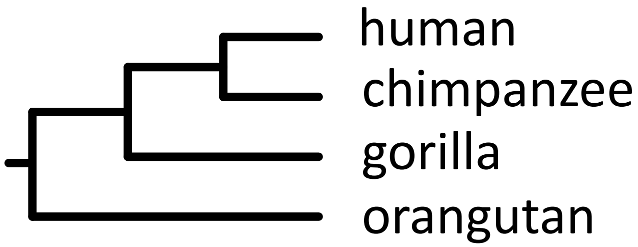
Gene 1



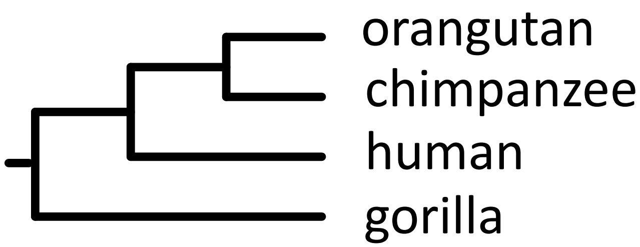
Gene 2



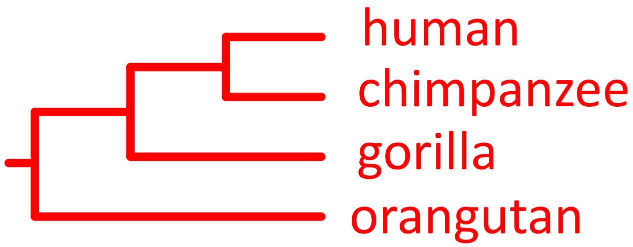
Gene 5



Gene 4



Gene 5



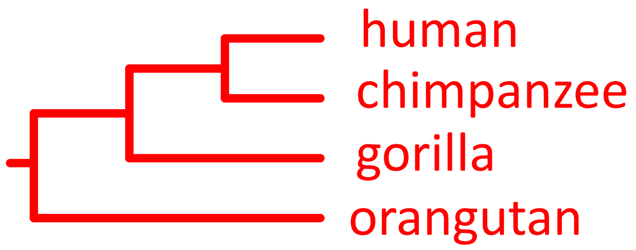
Consensus

But the most frequent gene tree does not always match the true species tree ("anomaly zone")

Analysing multiple loci

2. **Concatenation**

   Assume that all genes share the same evolutionary history



All genes

But this ignores the occurrence of different gene trees

Species tree

3. **Coalescent-based methods**
   Estimate the species tree based on gene trees

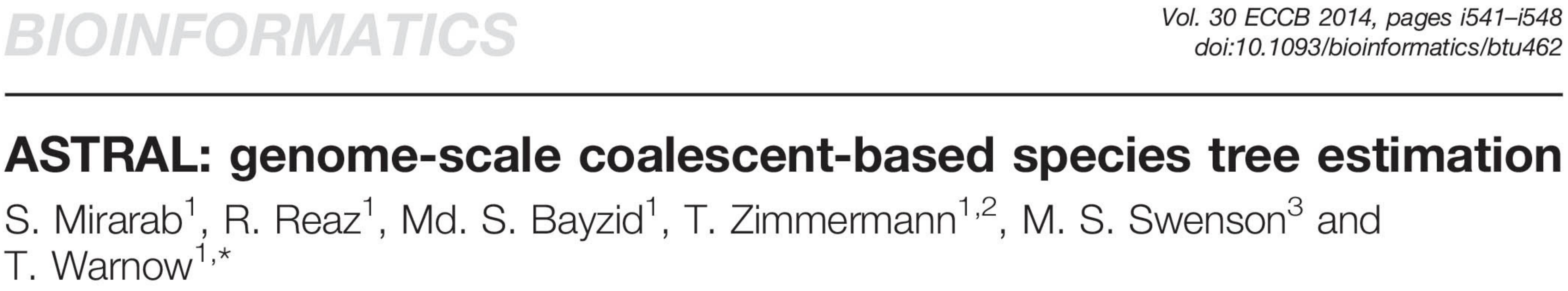
- Gene trees are independent realisations of a stochastic process (the coalescent) on the same species tree

- Various methods

  - Multispecies coalescent in **BEAST*
    Joint estimation of gene trees and the species tree

  - Summary coalescent in *ASTRAL*

ASTRAL



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ASTRAL: genome-scale coalescent-based species tree estimation

S. Mirarab[1], R. Reaz[1], Md. S. Bayzid[1], T. Zimmermann[1,2], M. S. Swenson[3] and T. Warnow[1,*]

- **A**ccurate **S**pecies **TR**ee **AL**gorithm
- Finds the species tree with the highest agreement with ‘quartets’ among the gene trees
- Can analyse a genome-scale data set in seconds or minutes

Inferring the species tree

- Choosing between concatenation and coalescent-based approaches
- **Shallower timescales**: gene trees inferred accurately but incomplete lineage sorting is important
- **Deeper timescales**: gene trees are inferred less accurately (homoplasy) but incomplete lineage sorting is less important
- The choice of methods to use should be informed by the largest sources of error

Bryant & Hahn (2020) In: *Phylogenetics in the Genomic Era*

Useful references

- **Lineage sorting in apes**
  Mailund *et al*. (2014) *Annu Rev Genet*, 48: 519–535.
- **Estimating phylogenetic trees from genome-scale data**
  Liu *et al*. (2015) *Annals New York Acad Sci*, 1360: 36–53.
- **The concatenation question**
  Bryant & Hahn (2020) In: *Phylogenetics in the Genomic Era*.