Lecture 1.3

Phylogenetic Data

Phylogenetic data

1. **Data preparation**
   - Taxon and gene sampling
   - Sequence alignment (if needed)
   - Data filtering

2. **Phylogenetic inference**
   - Model selection
   - Estimation of tree
   - Further analysis and interpretation

Phylogenetic data

- **Select data to optimise signal:noise**
  - Slowly evolving markers for deep evolutionary events
  - Rapidly evolving markers for recent evolutionary events
- **Homoplasy**
  - Taxa share similarities that do not reflect evolutionary history
- **Take advantage of existing resources**

Data types

- **Sequence data**
  - Nucleotides
  - Amino acids
- **Binary data** (presence/absence of genomic features)
- **Microsatellites** (repeat numbers)
- **Single-nucleotide polymorphisms** (SNPs)
- **Reduced-representation sequences**

Morphological data

- Morphological characters from extant and extinct taxa

Current Biology

Volume 25, Issue 19, 5 October 2015, Pages R922–R929

Review

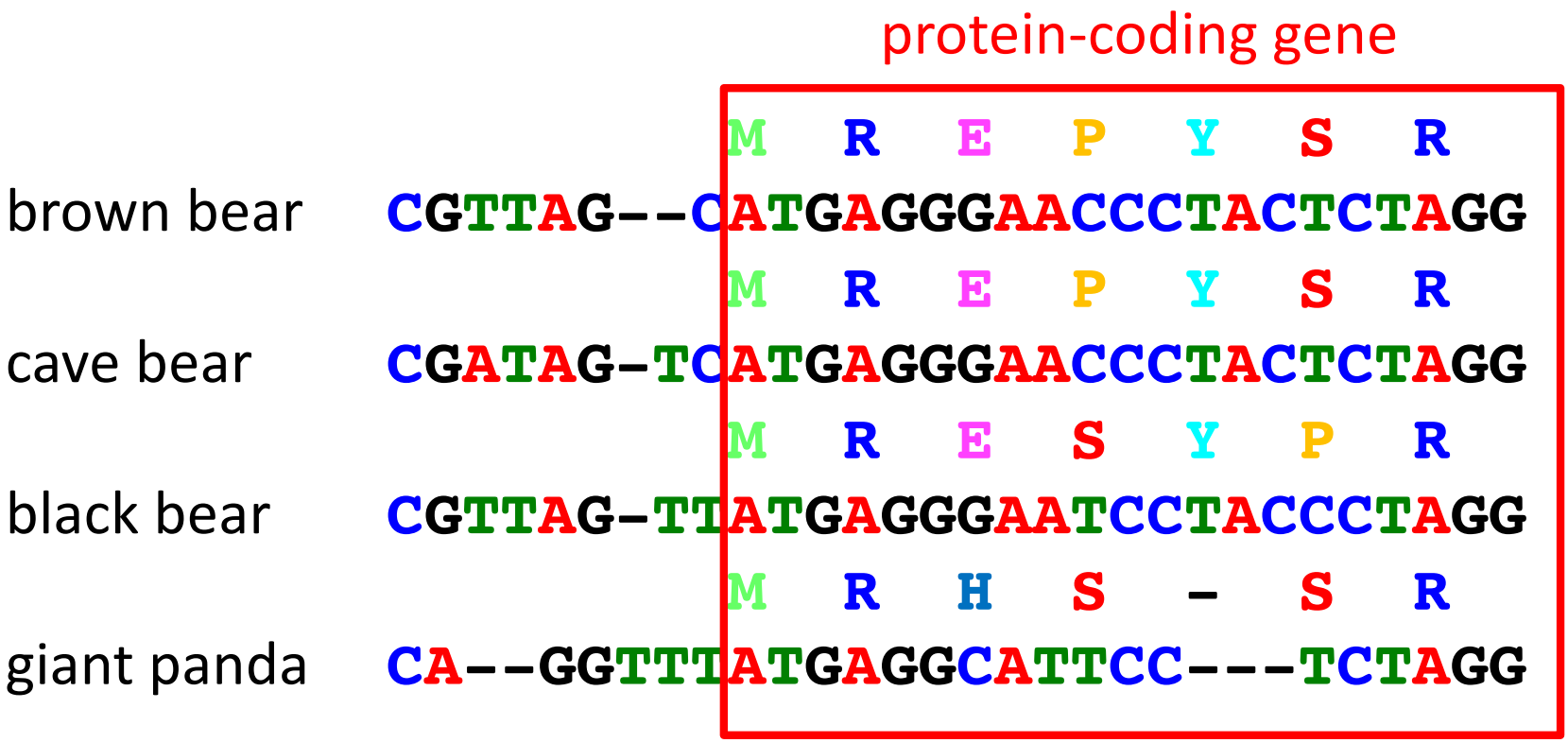
Morphological Phylogenetics in the Genomic Age

Michael S.Y. Lee[1, 2], Alessandro Palci[1, 2]

Sequence data

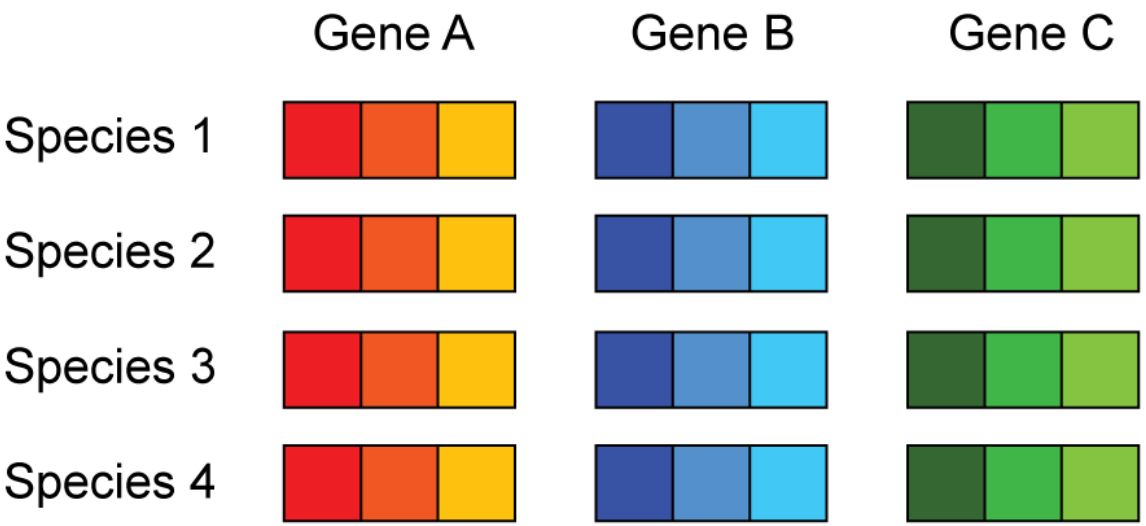
- **Coding sequences**
  - Ribosomal RNA
  - Protein-coding genes
- **Non-coding sequences**
  - Intergenic sites
  - Introns
- **Amino acid sequences**

Sequence data



Data partitioning

- Separate substitution model for each gene and codon position?



- **Biological**
  - Genome
  - Genes
  - Codon positions
  - RNA stems *vs* loops
  - Hydrophobic *vs* hydrophilic
- **Statistical**

PartitionFinder

- Too many possible partitioning schemes
  - 15 schemes for 4 genes
  - 52 schemes for 5 genes
  - 203 schemes for 6 genes

PartitionFinder: Combined Selection of Partitioning Schemes and Substitution Models for Phylogenetic Analyses

Robert Lanfear,[*,1] Brett Calcott,[1,2] Simon Y. W. Ho,[3] and Stephane Guindon[4]

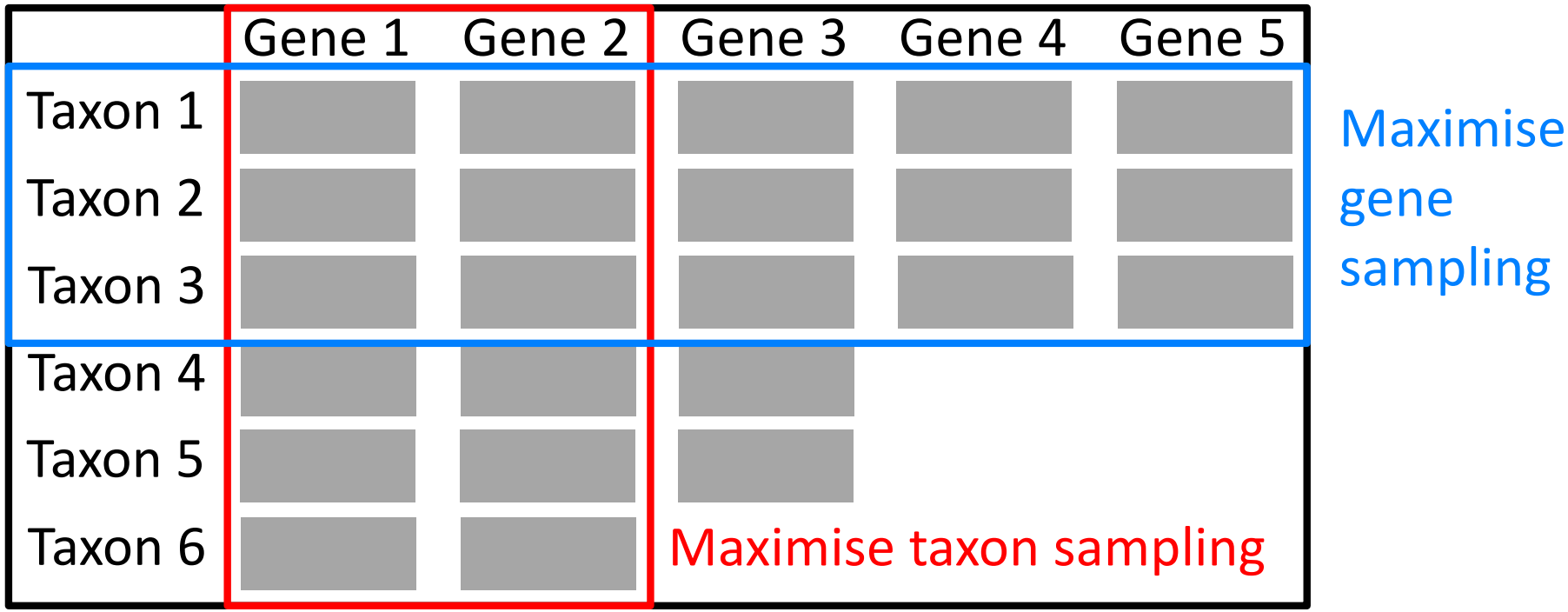
2012 – *Molecular Biology and Evolution*, 29: 1695–1701.

Gaps and missing data

- **Delete sites with any missing data**
  - Potential loss of informative data
  - Problematic in analyses of data supermatrices
- **Treat gaps as unresolved data**
  - Gap is simultaneously A, C, G, and T
  - Most common approach
- **Treat gaps as a 5th (nucleotide) or 21st (amino acid) state**
  - Not appropriate when there are long gaps
- **Code gaps as binary characters**

Gaps and missing data

- Impact of missing data remains poorly understood
- Filter data according to chosen threshold of missing data



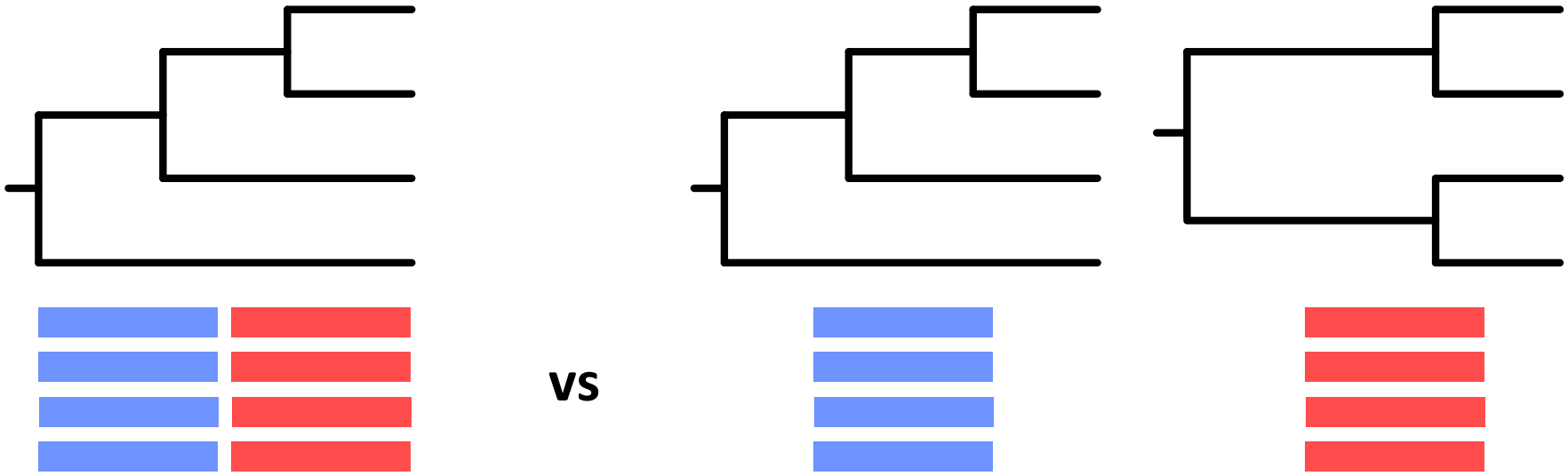
Mutational saturation

- Some sites can evolve very rapidly
  - 3rd codon positions
  - Loop regions in RNA
- Multiple hits can erode phylogenetic signal
- Various ways of testing for saturation
  (e.g., Xia's test in DAMBE)

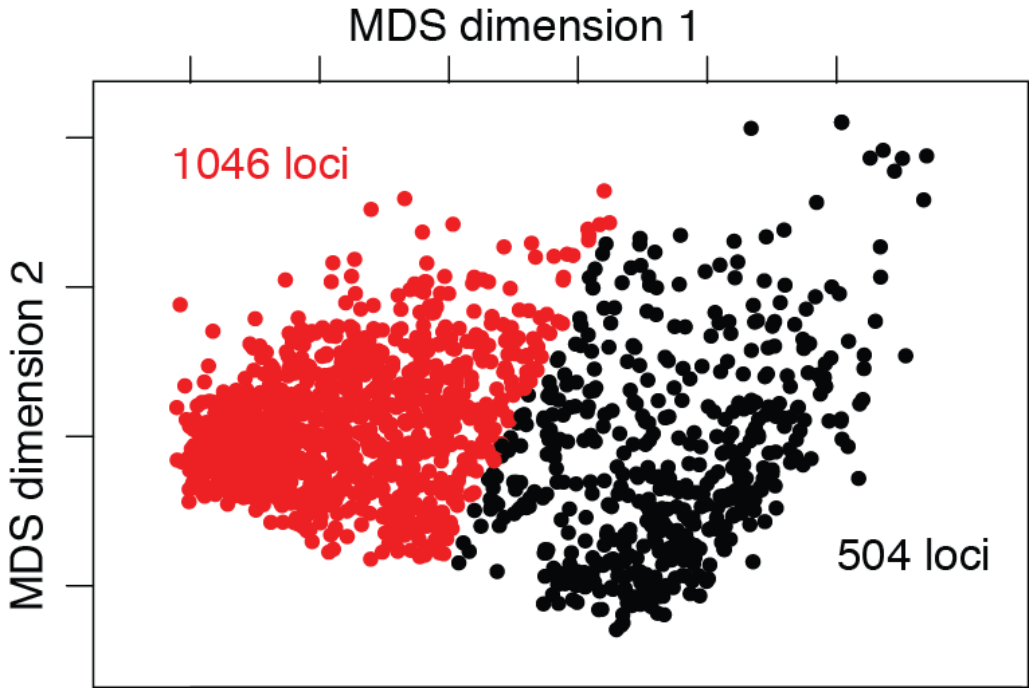
Saturated sites can be removed to improve signal:noise

Incongruence among gene trees

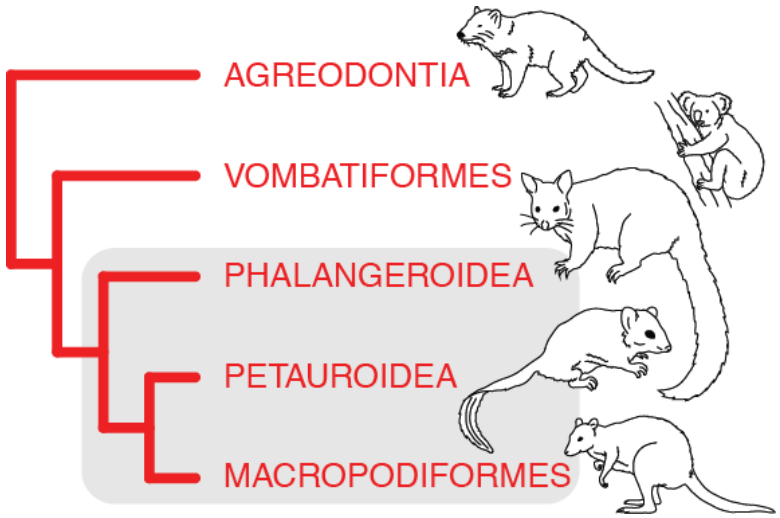
- Unlinked loci can have different gene trees
- Test for phylogenetic congruence across markers
- Partition-homogeneity (incongruence length difference) test

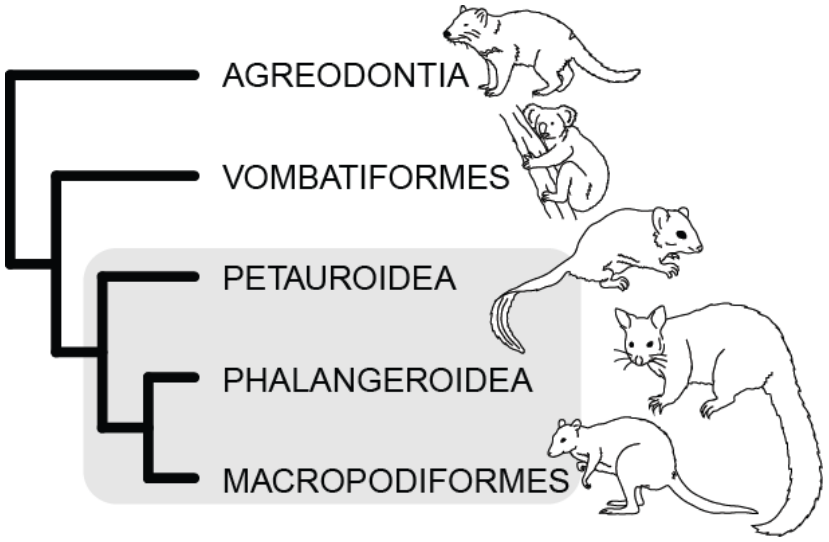


Incongruence among gene trees

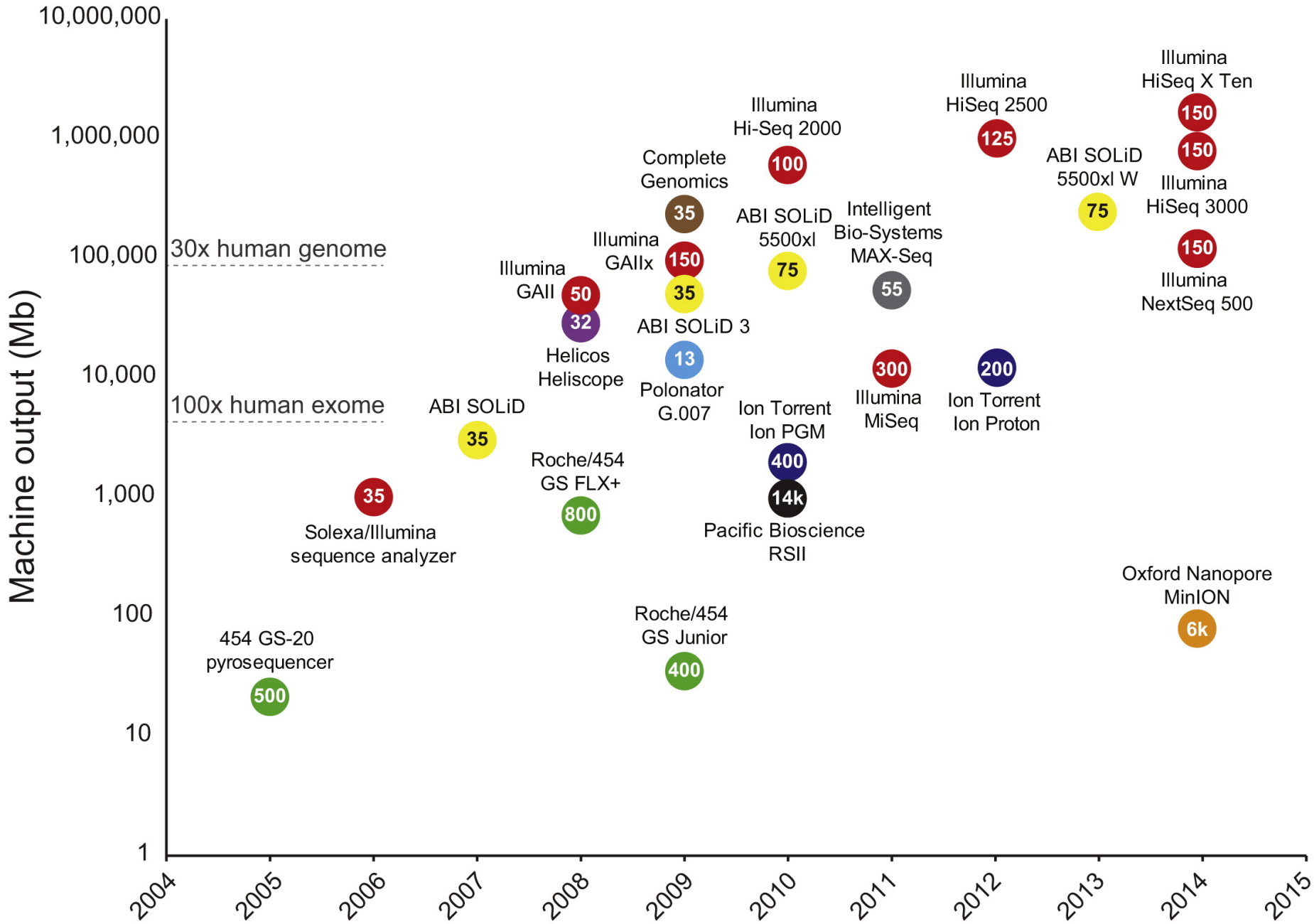


Duchene *et al.* (2018)
Syst Biol





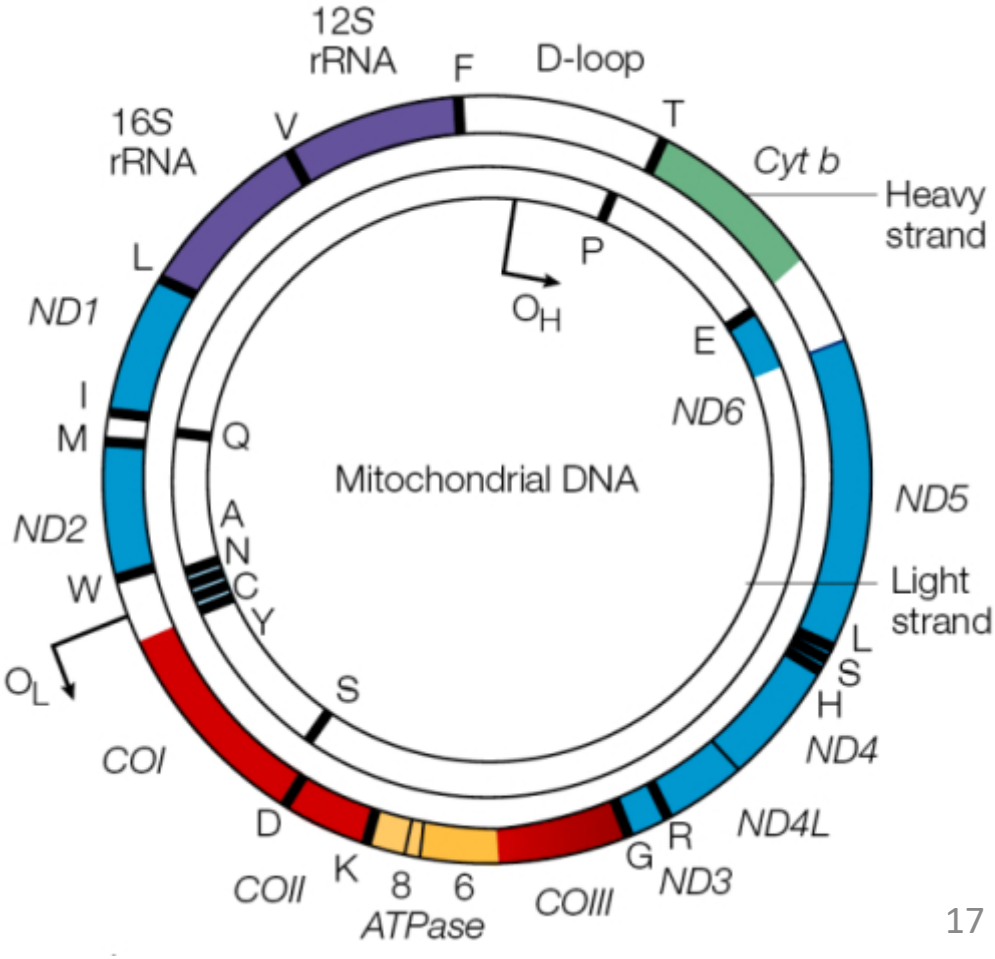
High-Throughput Data



Reuter *et al.* (2015) *Molecular Cell*

Mitochondrial genomes

- Maternally inherited
- Protein-coding genes (e.g., *COI*)
- RNA genes (e.g., *12S*, *16S*)
- Control region



Single-nucleotide polymorphisms

- Single sites sampled from throughout the genome
- More common in intraspecific (population) studies

- Issues to consider:
  - **Recombination**
    SNPs are usually unlinked so they are likely to have different (gene) trees
  - **Ascertainment bias**
    SNPs are selected for variability and this can mislead estimates of population sizes, rates, and other parameters

Reduced-representation sequences

- Markers identified by cutting genome with restriction enzymes
- Process creates binary data and short sequences
- Examples include RADseq and DArTseq

- Issues to consider:
  - **Recombination**
    Markers are usually unlinked so they are likely to have different (gene) trees
  - **Missing data**
    Typically a large proportion of missing data

Leaché *et al.* (2015) *Syst Biol*

Transcriptomes and exon capture

- Large panels of protein-coding loci
- Sequences are easier to align
- Good for inferring deep relationships

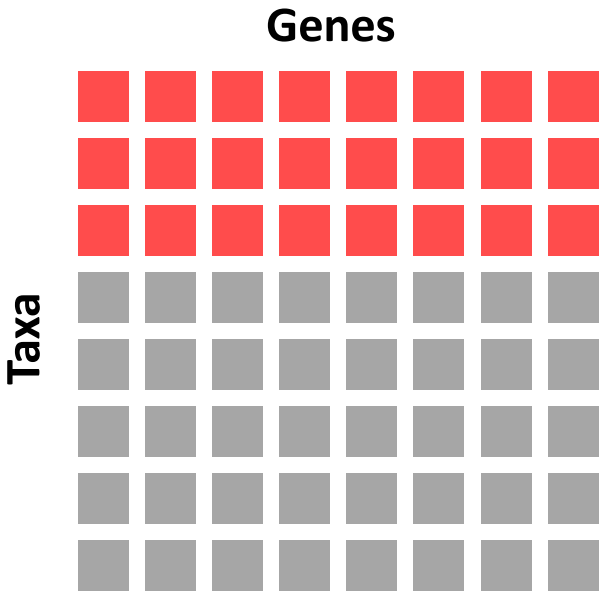
- Issues to consider:
  - **Variability**
    Might not be much variation at the population level
  - **Selection**
    Differences in selection will lead to rate differences across exons

Whole genome sequencing

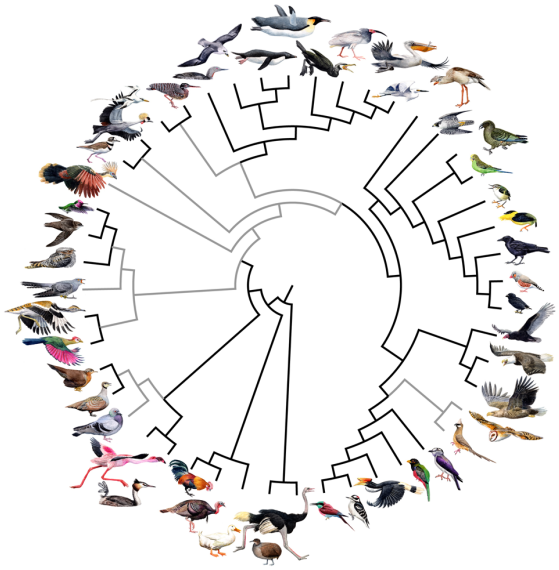
- Typically NOT (yet) the entire genome
- Many challenges: Jarvis et al *Science* 2014 >400 years of computing using a single processor
- **Issues to consider**
  - Single-copy genes
  - Selectively neutral
  - Unlinked loci

Analysing Large Data Sets

Large data sets

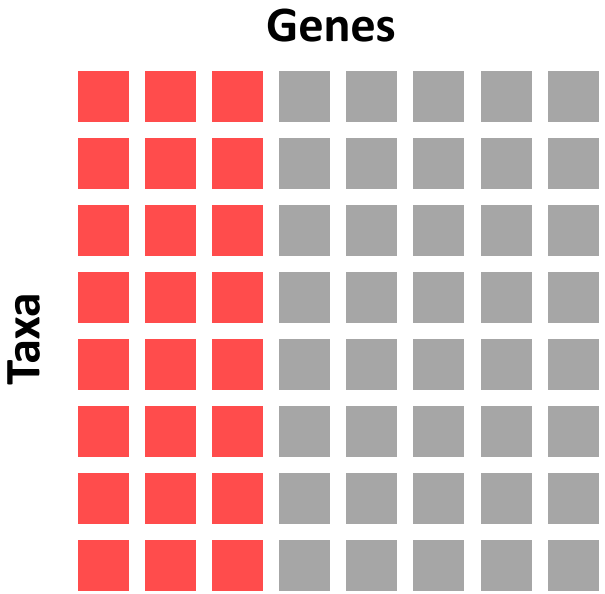


- Calculation of likelihood is expensive
  - Speed up by grouping sites with identical patterns
  - Approximate likelihood calculation
  - Multithreading/parallelisation

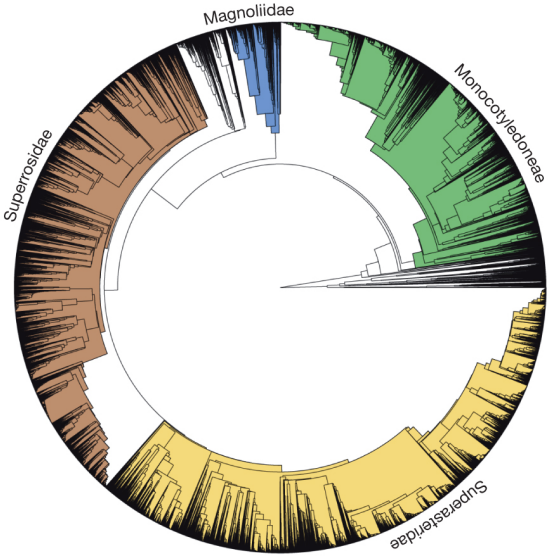


48 taxa
8,295 genes
Jarvis *et al*. (2014) *Science*

Large data sets



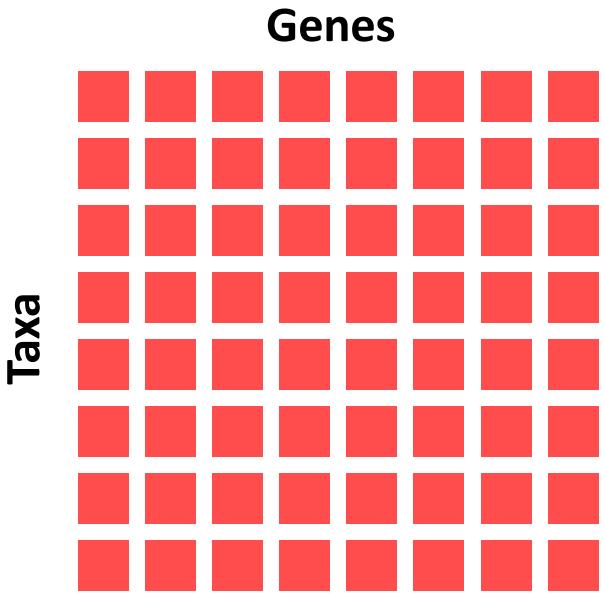
- Tree-space is extremely large
  - Efficient tree-searching heuristics



32,223 taxa
7 genes

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

Large data sets



- Analysis is computationally expensive
- Consider filtering the data
  - Phylogenetic signal
  - Mutational saturation
  - Missing data
  - Model fit

Useful references