Lecture 1.3

Phylogenetic Data

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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 characters**

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Review

Morphological Phylogenetics in the Genomic Age

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

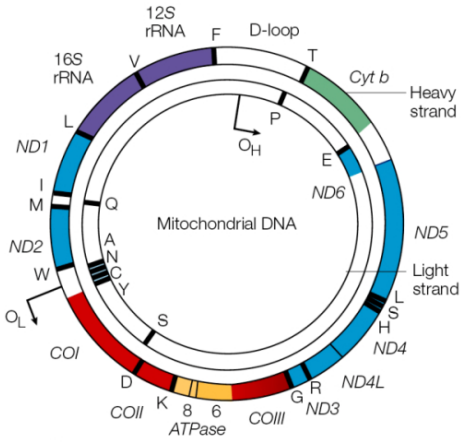
Sequence Data

Sequence data

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

Commonly used DNA sequence loci

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



Commonly used DNA sequence loci

- **Nuclear genome**
  1. **Microsatellites (STRs)**: highly polymorphic, mutation model, size homoplasy
  2. **EPICs**: highly variable, indels
  3. **Anonymous loci**: highly variable, no ascertainment bias, neutral?
  4. **NPCLs**: conserved, lack indels, 'ancient' events
  5. **rRNA 'arrays'**: tandemly duplicated, variable conservation

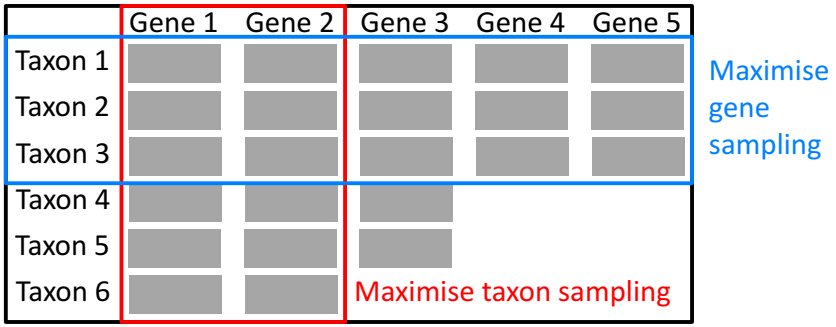
['array' = 3 rDNA coding segments (18S, 5.8S, 28S), 2 internal transcribed spacer elements ITS-1 & 2, external transcribed spacer (ETS) and non-transcribed spacer (NTS)]

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



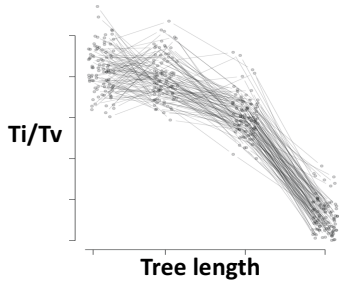
Mutational saturation

- 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. ISS – DAMBE)

Saturated sites can be removed to improve signal:noise

Mutational saturation

- **Plot transitions and transversions**
  - Transitions occur more frequently than transversions
  - Can plot in various ways



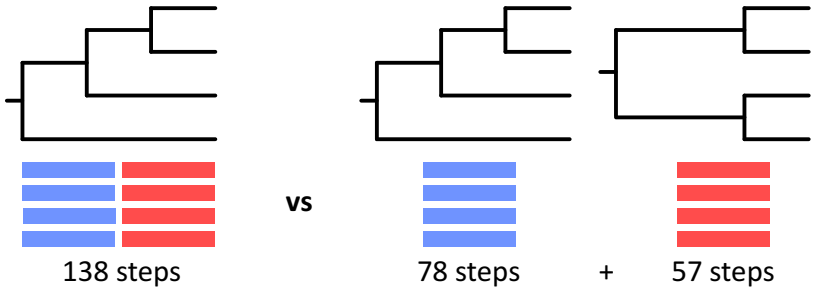
- **Xia's method (I_{SS})**
  - When sequences are fully saturated (I_{SS} = 1), expected base frequencies at each site are equal to the global frequencies
  - Compare I_{SS} with critical value calculated via simulation

Duchêne, Ho, & Holmes (2015) *BMC Evol Biol*

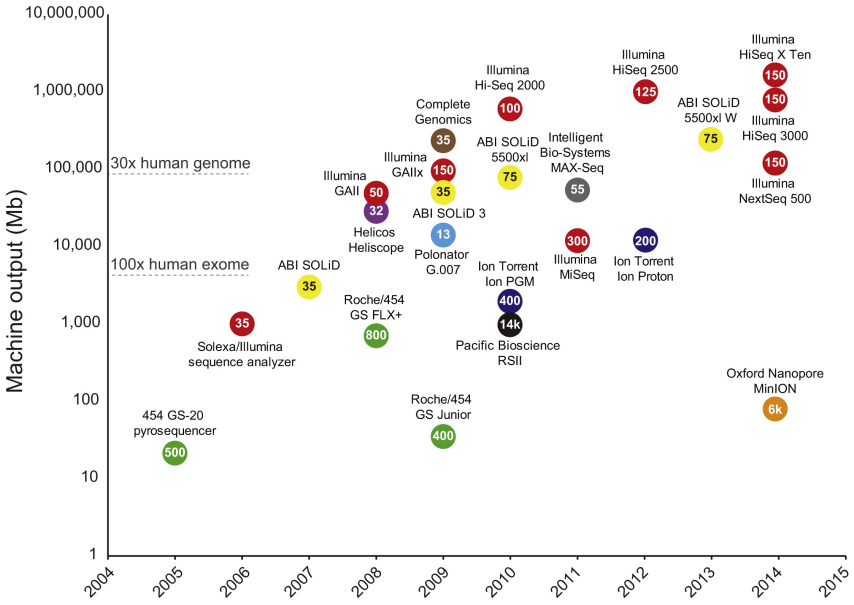
Xia *et al.* (2003) *Mol Phylogenet Evol*

Partition-homogeneity test

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



High-Throughput Data



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

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

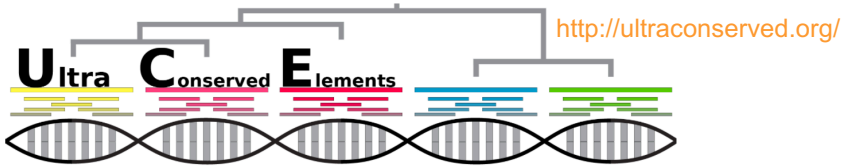
Diversity Arrays Technology
IT'S A BULLSEYE WITH DArT

- 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*

Ultra-conserved elements

- Genomic elements perfectly conserved in mammals (>200 bp)
- Gene 'deserts' – long range regulators?
- Flanking regions: phylogenetic markers varying in conservation
- Purifying selection?
  - Increase rate of lineage sorting: gene trees ≈ spp. trees
  - Inference of historical demographic parameters problematic – N_e



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**
  - Loci are single copy?
  - Selectively neutral?
  - Sampled loci have independent gene trees?
  - Historical recombination?

Useful references