Introduction to Phylogenetics
Week 1

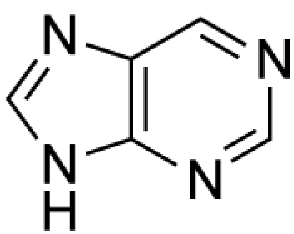
Basic Concepts of Molecular Evolution

I. Basic Concepts

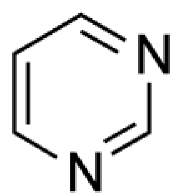
- Most life uses DNA as its genetic template
- Some viruses use RNA as the genetic template
- Evolution rate – Depends a great deal on generation time, fidelity of enzymes, fecundity and environmental stressors

I. Basic Concepts

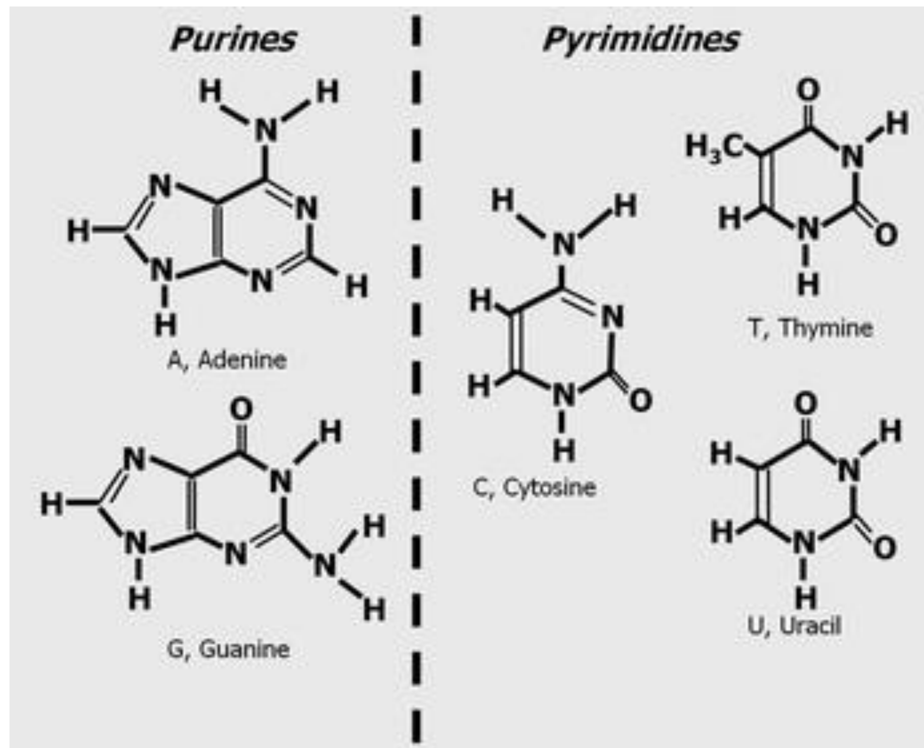
- Purines are A or G and pyrimidines are T or C (or U)



purine (R)

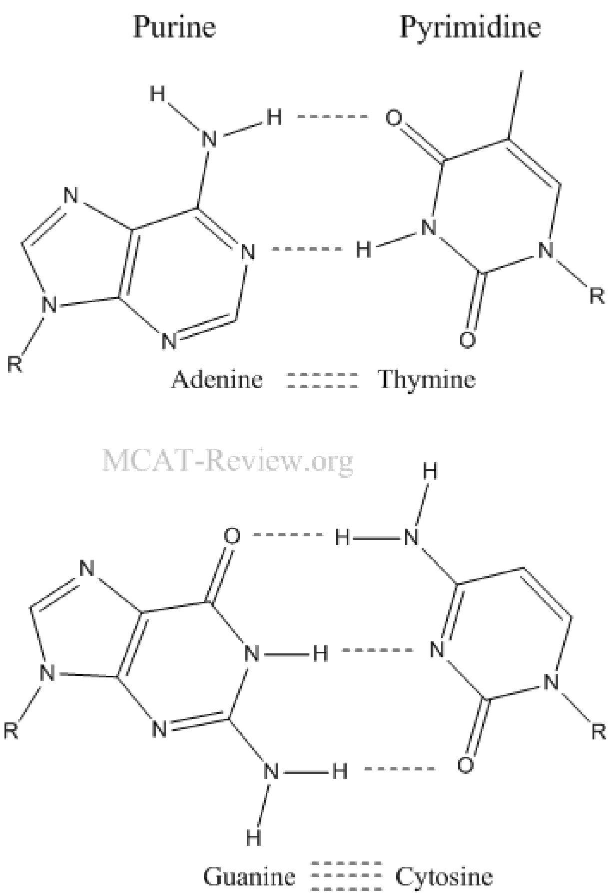


pyrimidine (Y)



I. Basic Concepts

- A=T and G≅C



Kind of a big deal when it comes to evolution

I. Basic Concepts

- Because of purine: pyrimidine base pairing, transitions are more likely to occur than transversions
  - Transitions (4): A <-> G, C <-> T
  - Transversions (8): A <-> C, A <-> T, G <-> C, G <-> T

Kind of a big deal when it comes to evolution

I. Basic Concepts

- Genetic code is degenerate

Second base of codon

First base of codon	U		C		A		G		Third base of codon
U	UUU	Phenylalanine phe	UCU	Serine ser	UAU	Tyrosine tyr	UGU	Cysteine cys	U
	UUC		UCC		UAC		UGC		C
	UUA	Leucine leu	UCA		UAA	STOP codon	UGA	STOP codon	A
	UUG		UCG		UAG		UGG	Tryptonphan trp	G
C	CUU	Leucine leu	CCU	Proline pro	CAU	Histidine his	CGU	Arginine arg	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	Glutamine gln	CGA		A
	CUG		CCG		CAG		CGG		G
A	AUU	Isoleucine ile	ACU	Threonine thr	AAU	Asparagine asn	AGU	Serine ser	U
	AUC		ACC		AAC		AGC		C
	AUA		ACA		AAA	Lysine lys	AGA	Arginine arg	A
	AUG	Methionine met (start codon)	ACG		AAG		AGG		G
G	GUU	Valine val	GCU	Alanine ala	GAU	Aspartic acid asp	GGU	Glycine gly	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	Glutamic acid glu	GGA		A
	GUG		GCG		GAG		GGG		G

Only change in AA:

30% at 3rd base
100% at 2nd base
96% at 1st base

I. Basic Concepts

- AAs can have similar chemistry

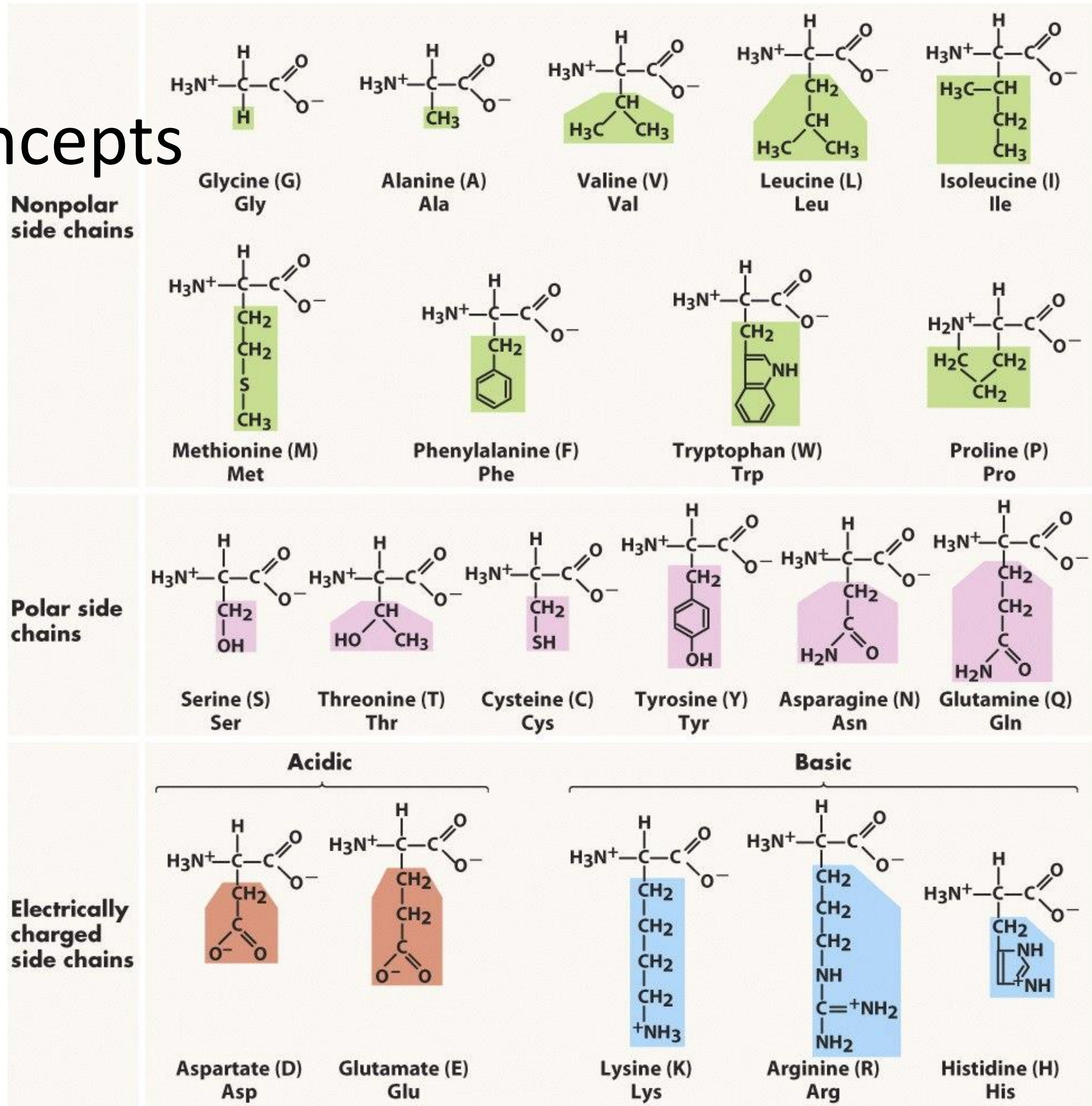


Figure 3-5 Biological Science, 2/e

I. Basic Concepts

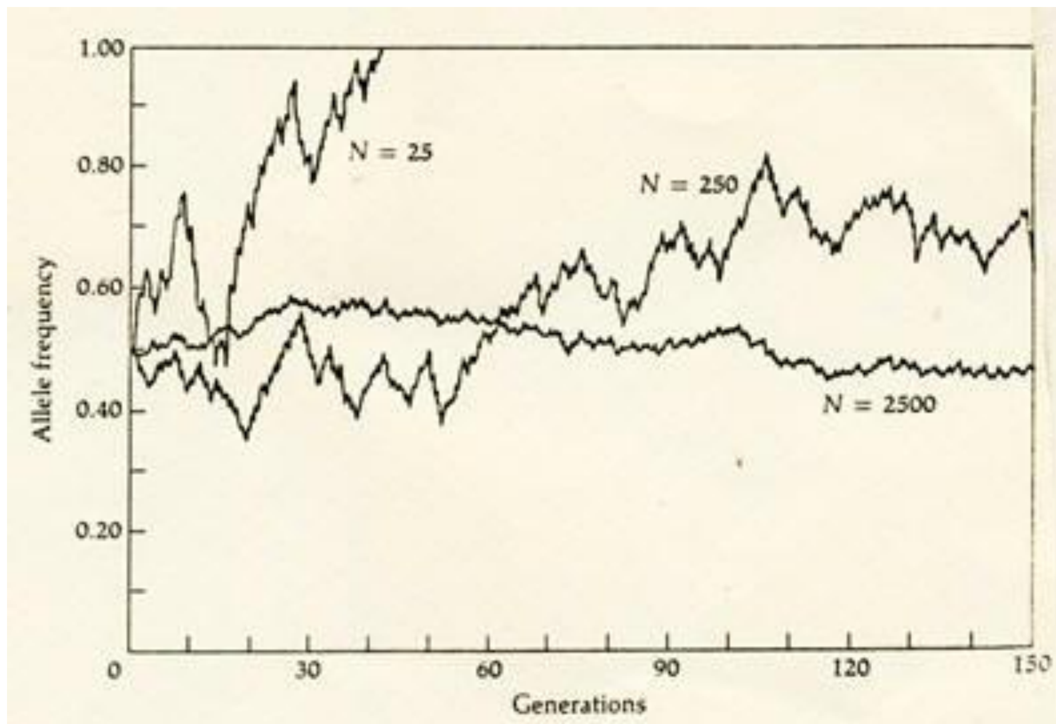
- Synonymous mutation – silent mutation
  - Generally under neutral selection
  - Can affect RNA stability
  - Can affect tRNA usage
- Non-synonymous mutation – non-silent
  - change in amino acid coding

I. Basic Concepts

- Gene duplication
- Orthologs
  - High selection pressure
- Paralogs
  - Relaxed selection pressure
  - Increased mutation rate
  - Evolution unique functions

II. Population genetics

- Mutations – polymorphisms in offspring
- Fixation rate – rate where polymorphisms reach 100%



II. Population genetics

- Many genetic changes are maintained as polymorphisms in a population
- Cannot calculate rate of fixation

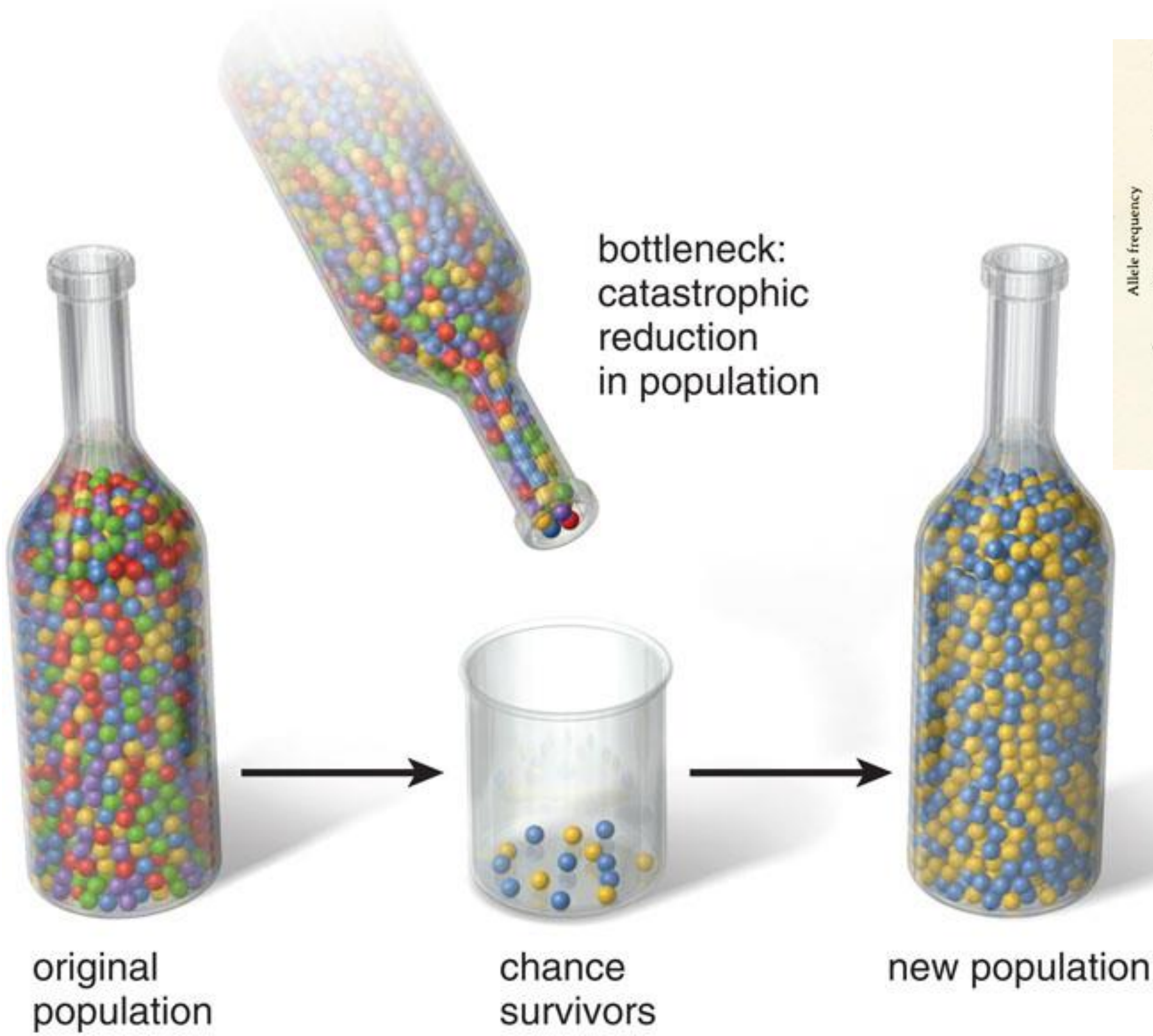
$$\frac{1}{2N} \times 2Nm = m$$

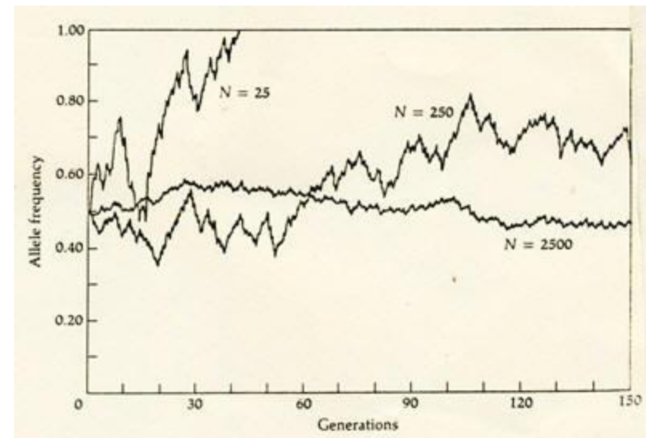
- Can use difference in genetic structure as equivalent to genetic differences

II. Population genetics

- Rate of population change
  - Underlying mutation rate
  - Generation time
  - Impact of allele on fitness
    - Positive selection
    - Negative selection
    - Balancing selection
  - Population size

II. Population genetics: Bottlenecks





Coalescent event

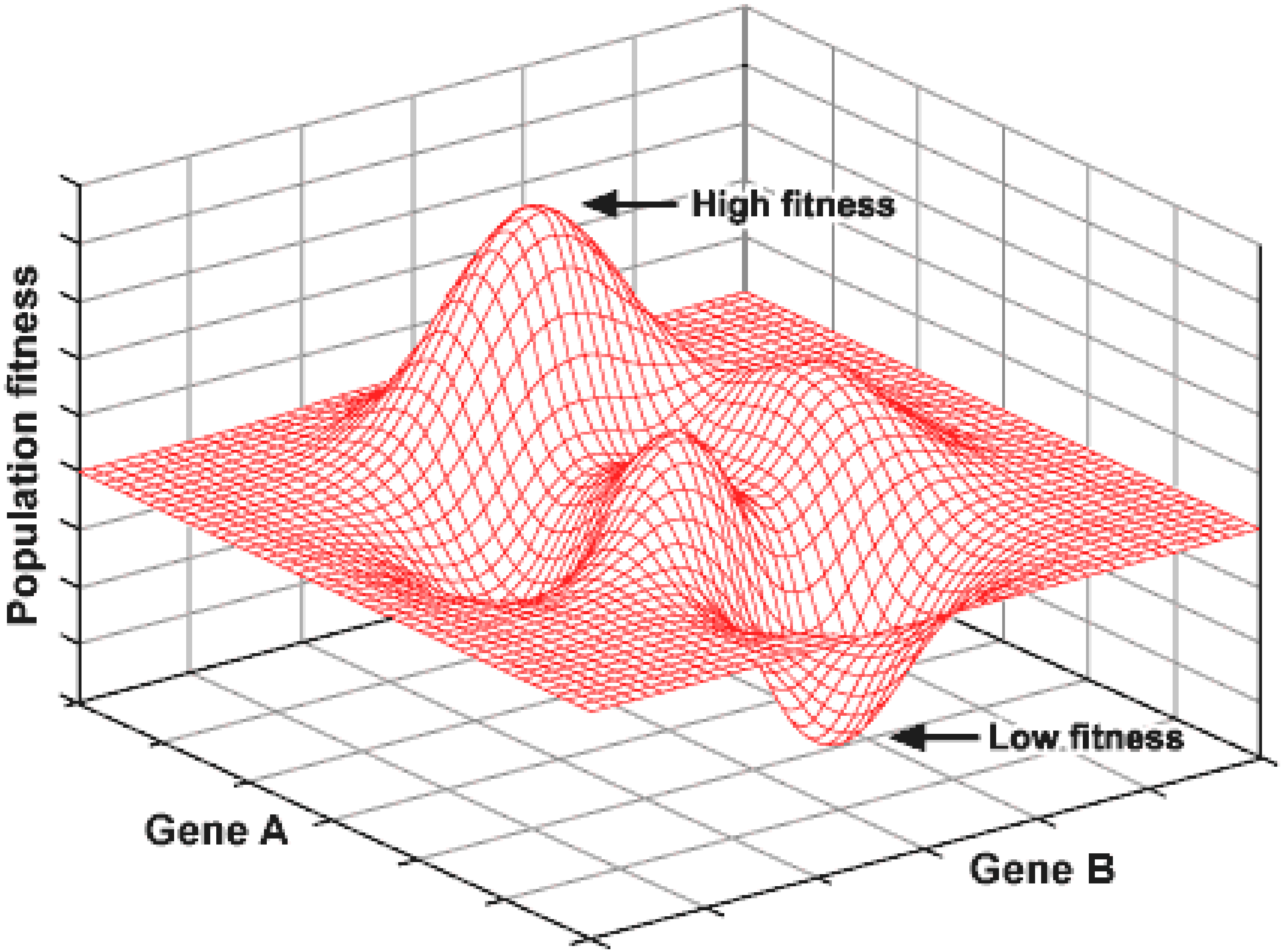
III. Evolutionary Models

- Deterministic
  - Chance events (other than environmental) do not dramatically effect allele frequencies
  - Change dependent positive/negative selection
  - Needs large population
- Stochastic
  - Random fluctuations determine allele frequency
  - Change cannot be determined by mutation rate
  - More important in small populations
  - Genetic drift (random sampling more important)

III. Evolutionary Models

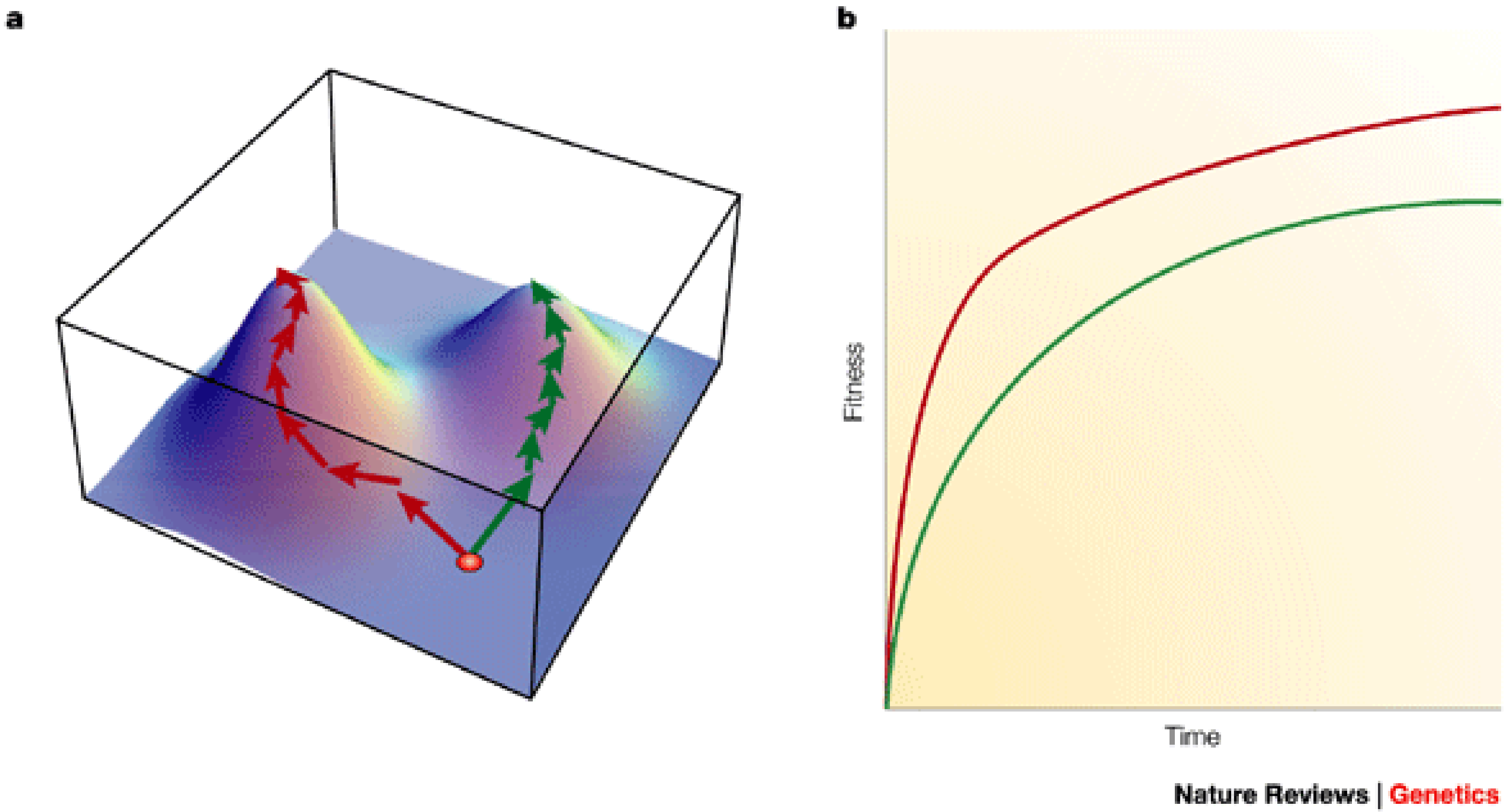
- Evolution is likely a combination of deterministic and stochastic effects
  - Mutations result in genetic variation that natural selection acts as dominant force
- Adaptive evolution – positive selective pressure on mutations
- Neutral theory – stochastic fixation of neutral/nearly neutral mutations
  - Assumes many organisms well adapted to current conditions
  - Positive selection only affects few individuals within greater population

III. Evolutionary Models



III. Evolutionary Models

- Adaptive landscapes

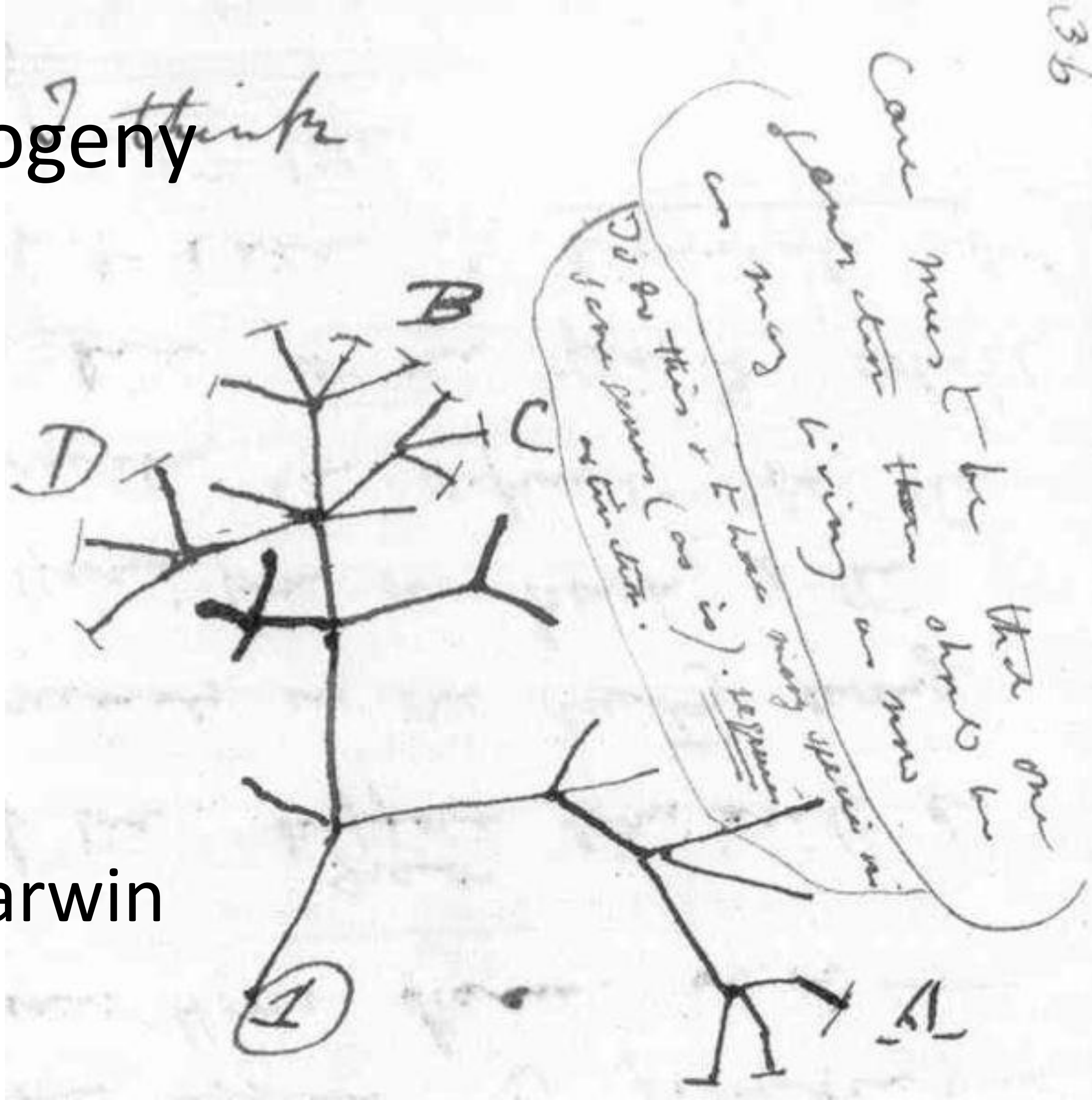


III. Phylogeny

Carl Linnaeus - Father of modern taxonomy

Began classifying plants based on structure – published *Systema Naturae* in 1735

III. Phylogeny



Charles Darwin

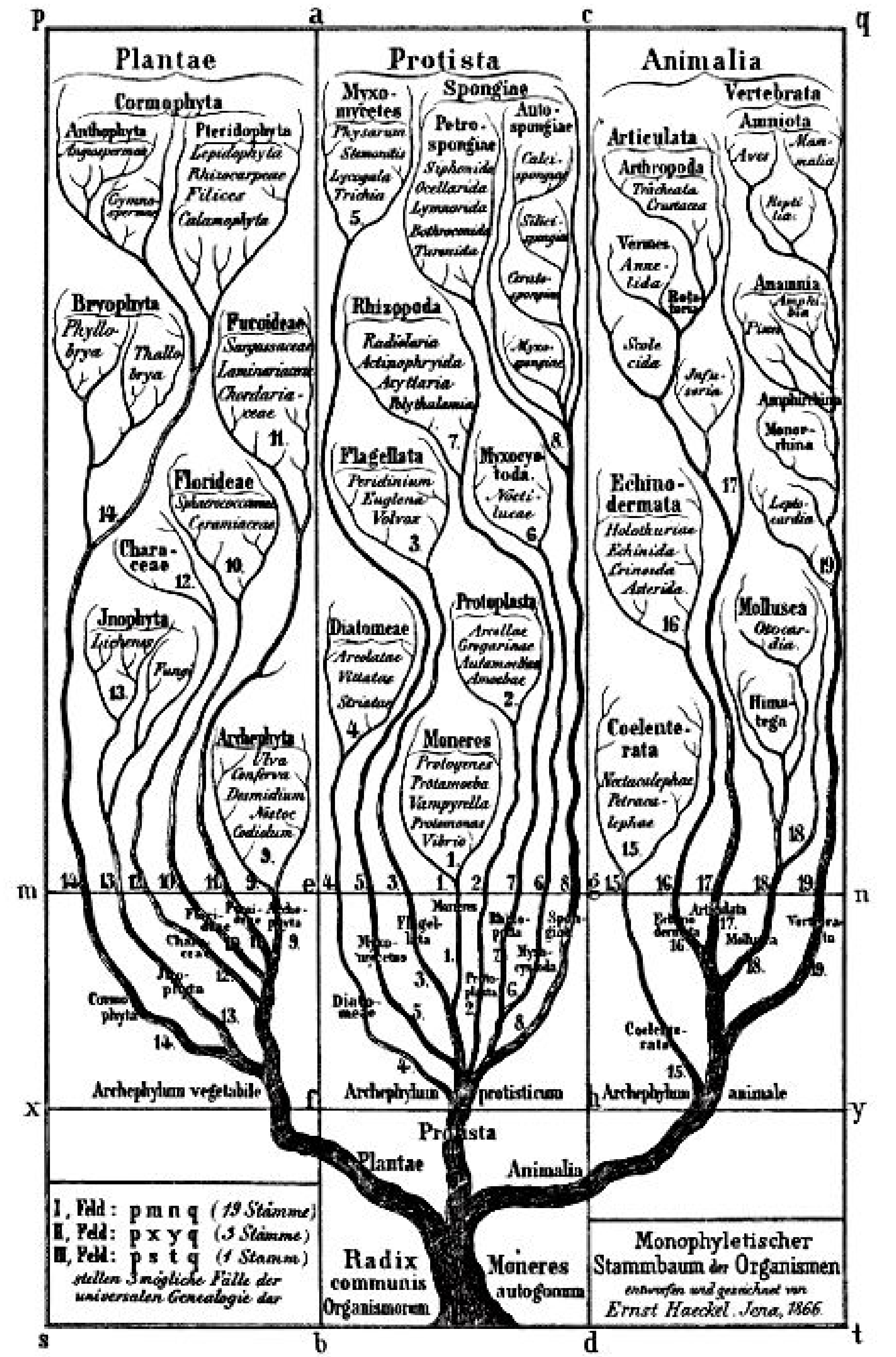
c. 1837

III. Phylogeny

Haeckle - First to include microorganisms within a tree

Created the group – Monera

Five-kingdom Tree of Life



IV. Molecular Phylogeny

- Determine relationships between genes/gene fragments
- Homologous regions need to be aligned (program)
- Missing sequence data must be trimmed (program)
- Determine appropriate evolutionary model to use (program)
- Generate phylogenetic tree (program)
- Test tree (program)

IV. Molecular Phylogeny

Nucleotide position	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
Microbe 1	A	C	U	G	A	C	U	C	A	U	A	G	A	U	C
Microbe 2	A	G	U	G	A	G	U	C	A	G	A	C	A	U	C
Microbe 3	U	C	U	G	G	G	U	C	A	G	A	C	A	U	C
Microbe 4	U	G	U	G	G	U	C	C	A	U	A	C	A	U	C

4/15 = 0.27
4/15 = 0.27
5/15 = 0.33
4/15 = 0.27
5/15 = 0.33
3/15 = 0.20

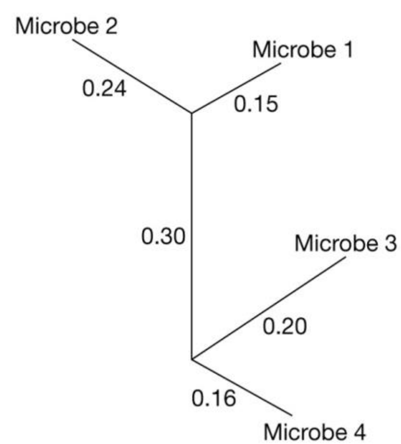
(a) Sequence alignment and analysis

Microbe	1	2	3	4
1	1.0	0.27	0.27	0.33
2		1.0	0.27	0.33
3			1.0	0.20
4				1.0

(b) Calculated evolutionary distance

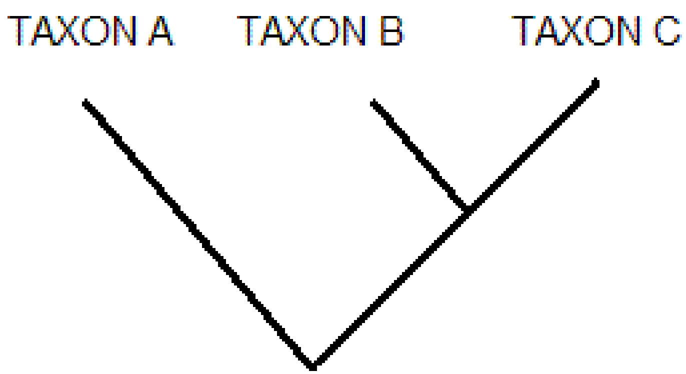
Microbe	1	2	3	4
1	1.0	0.32	0.32	0.44
2		1.0	0.32	0.44
3			1.0	0.26
4				1.0

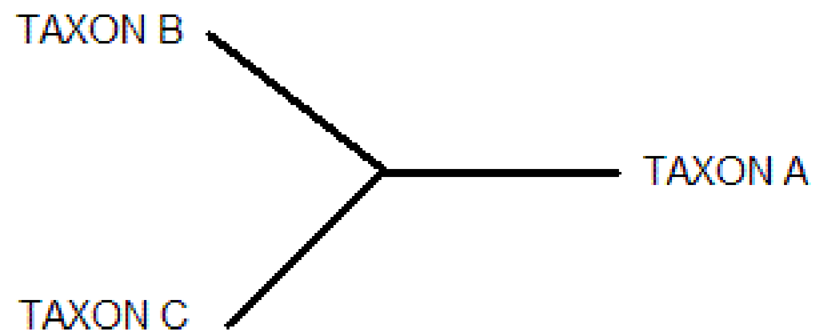
(c) Corrected evolutionary distance

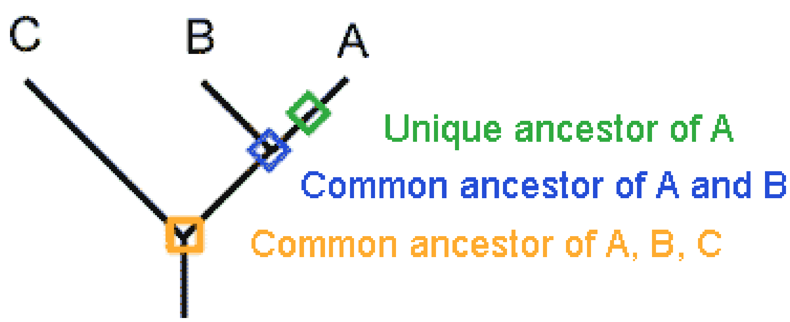


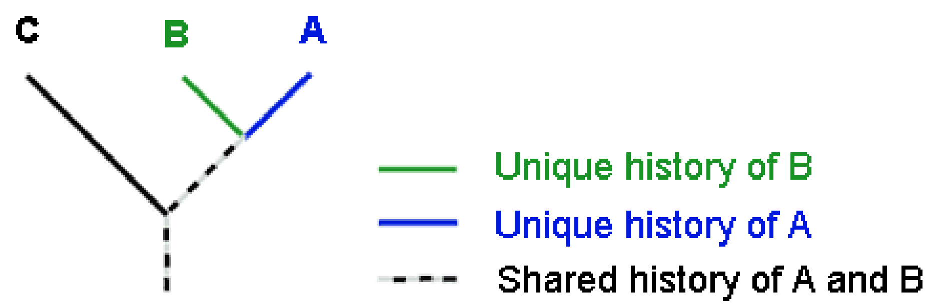
(d) Phylogenetic tree

IV. Molecular Phylogeny









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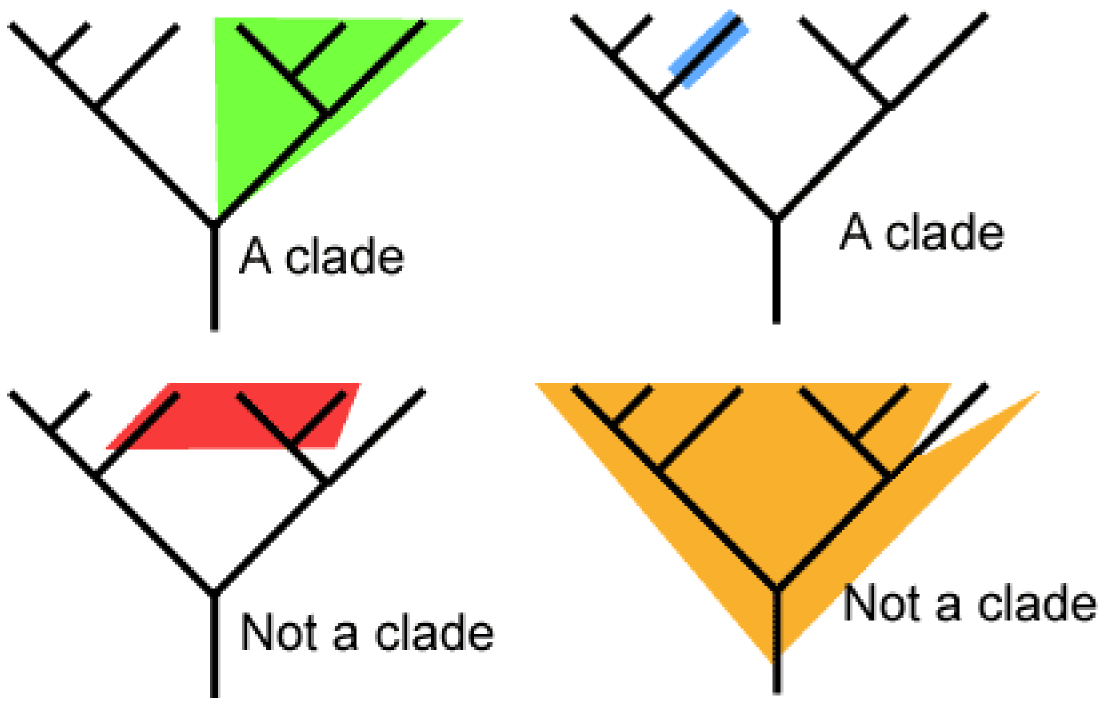

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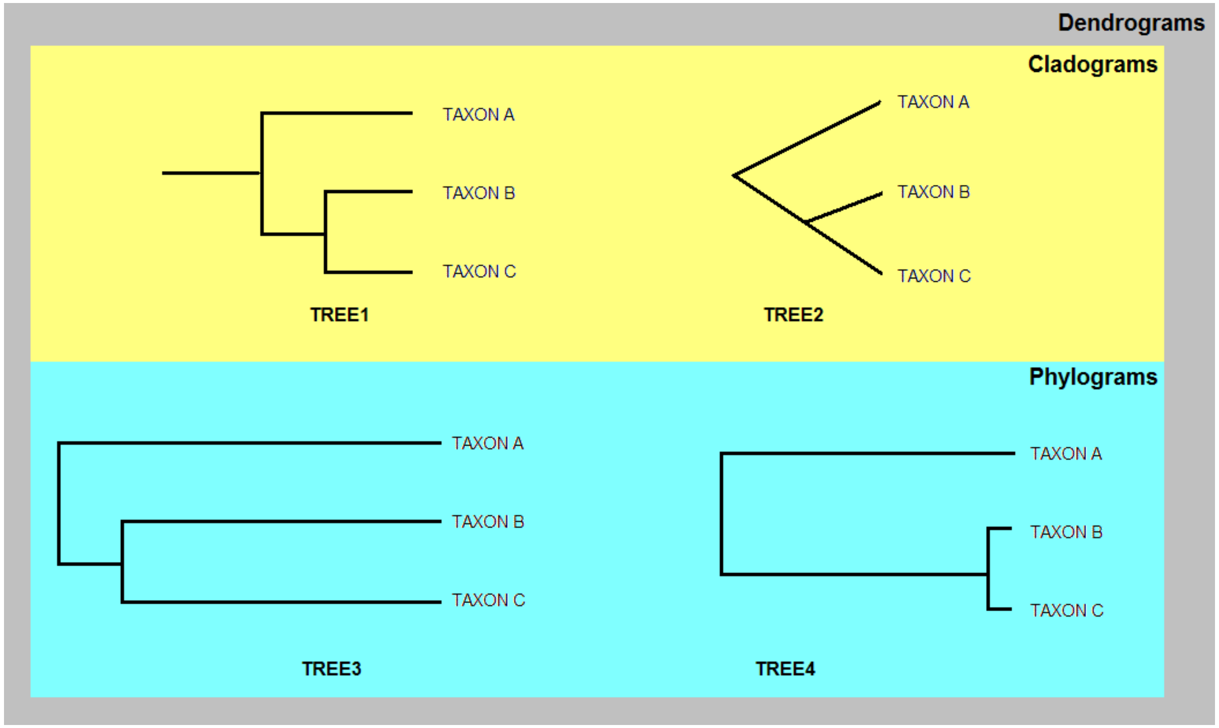
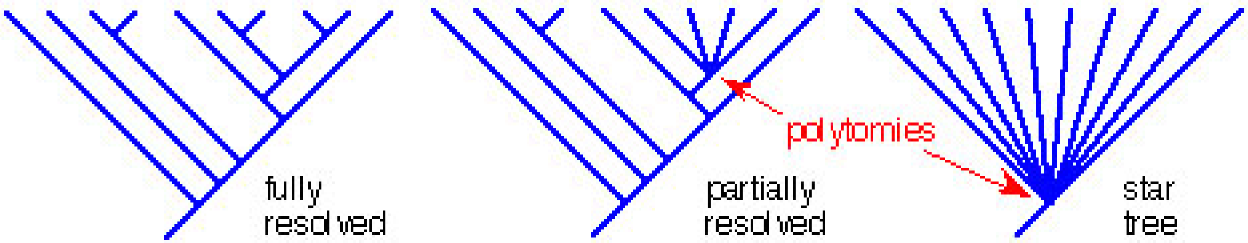
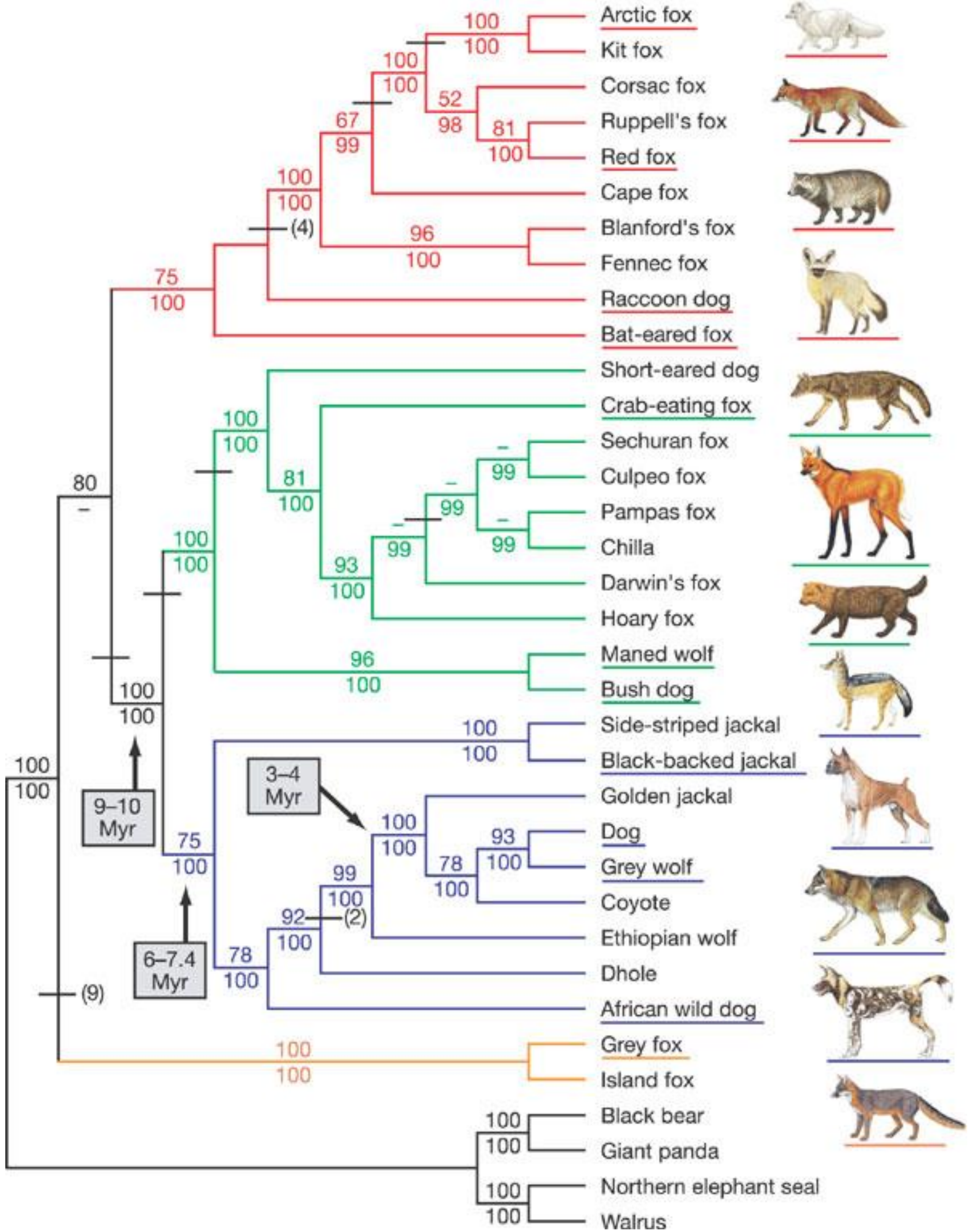
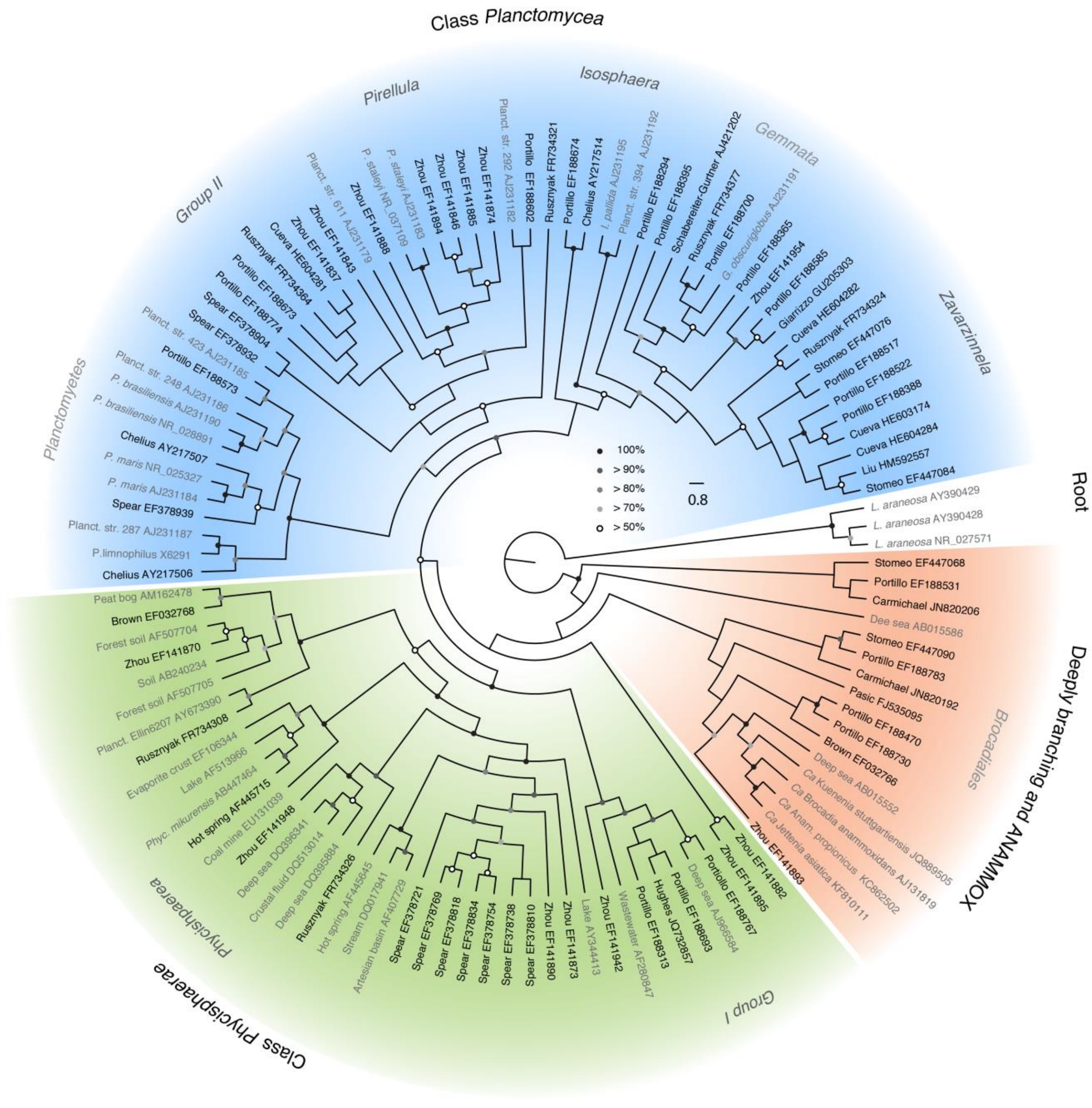


IV. Molecular Phylogeny

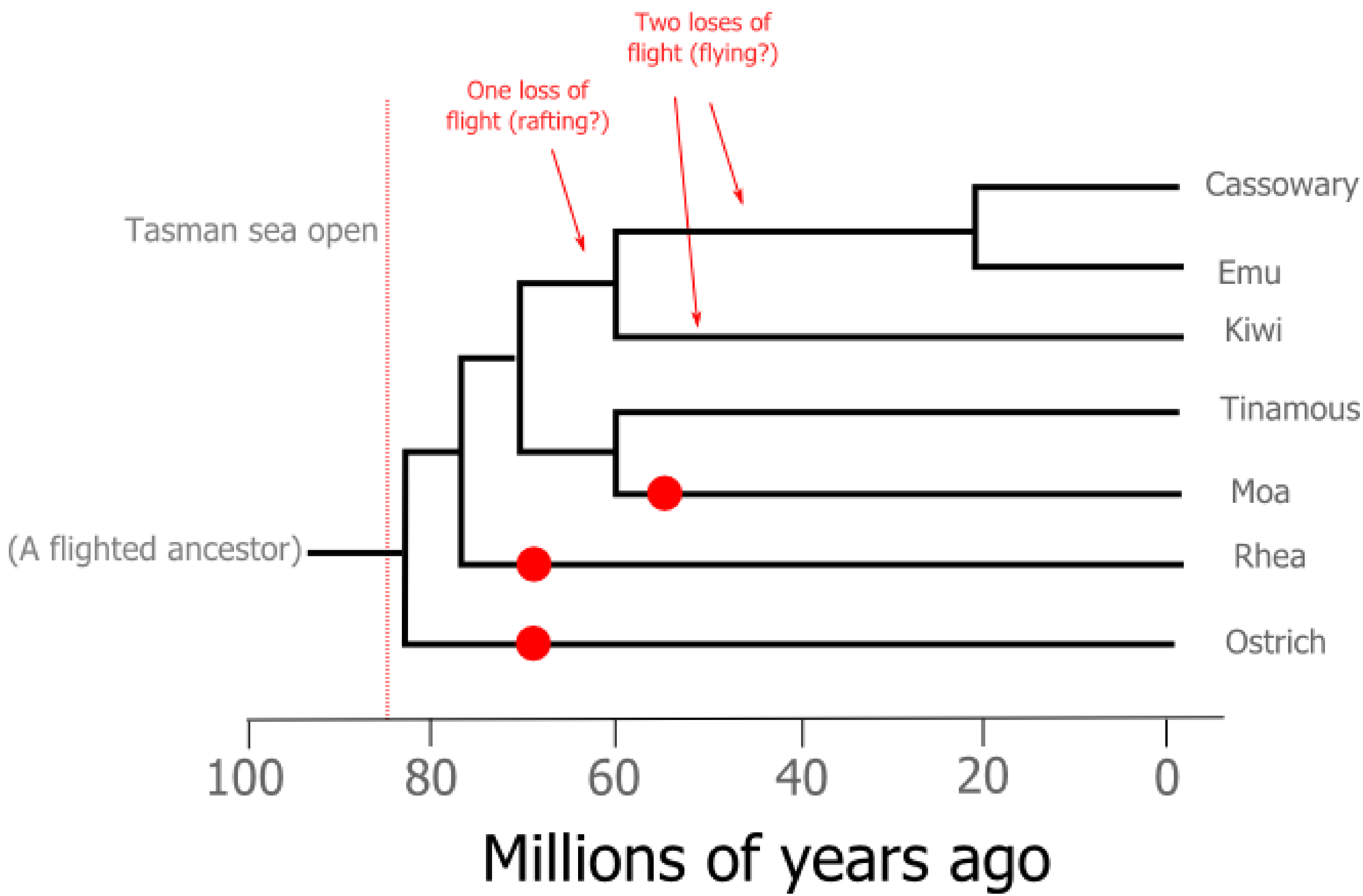




Arctic fox
Kit fox
Corsac fox
Ruppell's fox
Red fox
Cape fox
Blanford's fox
Fennec fox
Raccoon dog
Bat-eared fox
Short-eared dog
Crab-eating fox
Sechuran fox
Culpeo fox
Pampas fox
Chilla
Darwin's fox
Hoary fox
Maned wolf
Bush dog
Side-striped jackal
Black-backed jackal
Golden jackal
Dog
Grey wolf
Coyote
Ethiopian wolf
Dhole
African wild dog
Grey fox
Island fox
Black bear
Giant panda
Northern elephant seal
Walrus
9–10 Myr
6–7.4 Myr
3–4 Myr
(4)
(2)
(9)



Class Planctomycea
Pirellula
Isosphaera
Gemmata
Zavarzinnela
Group II
Planctomyetes
Root
Deeply branching and ANAMMOX
Brocadiales
Class Phycisphaerae
Phycisphaerea
Group I
100%
> 90%
> 80%
> 70%
> 50%
0.8


Two loses of
flight (flying?)
One loss of
flight (rafting?)
Tasman sea open
Cassowary
Emu
Kiwi
Tinamous
Moa
Rhea
Ostrich
(A flighted ancestor)
100
80
60
40
20
0
Millions of years ago

I. Definitions

- Synonymous mutation
- Non-synonymous mutation
- Transition
- Transversion
- Indels
- Positive selection
- Neutral selection
- Neutral theory
- Stochastic effects
- Allee effects