Species tree estimation & the multispecies coalescent

Concatenated gene sequences - assumes every gene has same evolutionary history

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Inconsistency of Phylogenetic Estimates from Concatenated Data under Coalescence

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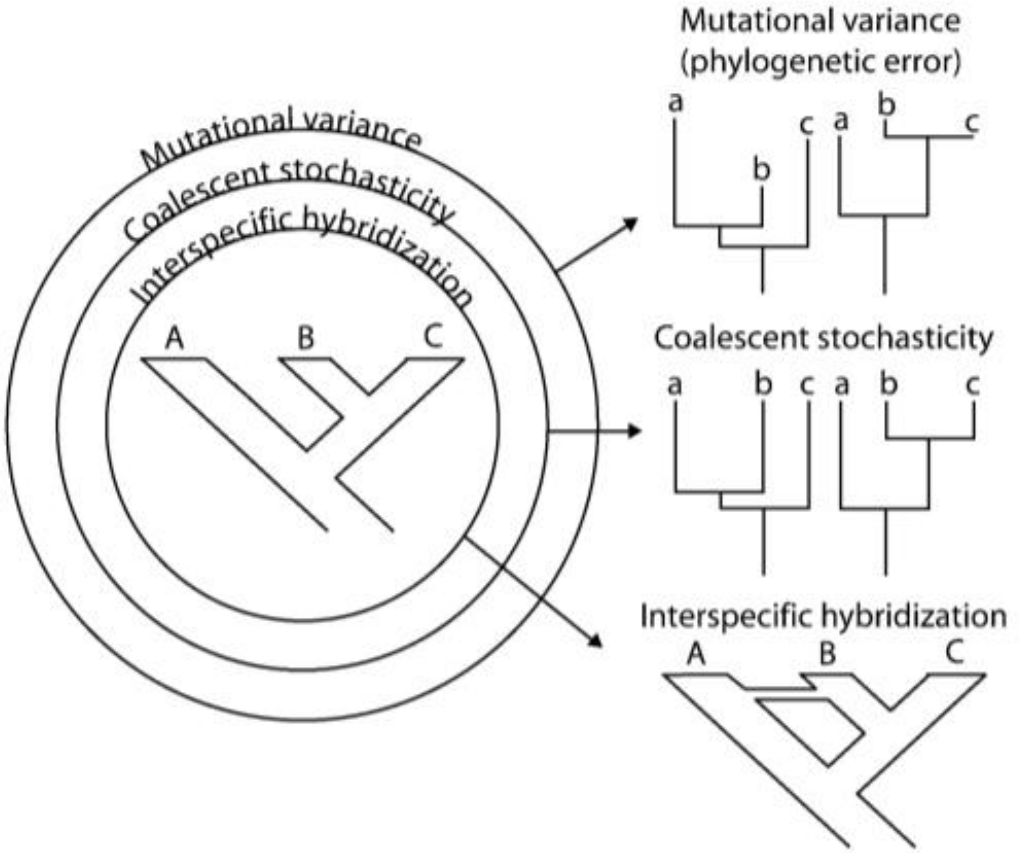
Stochasticity from:

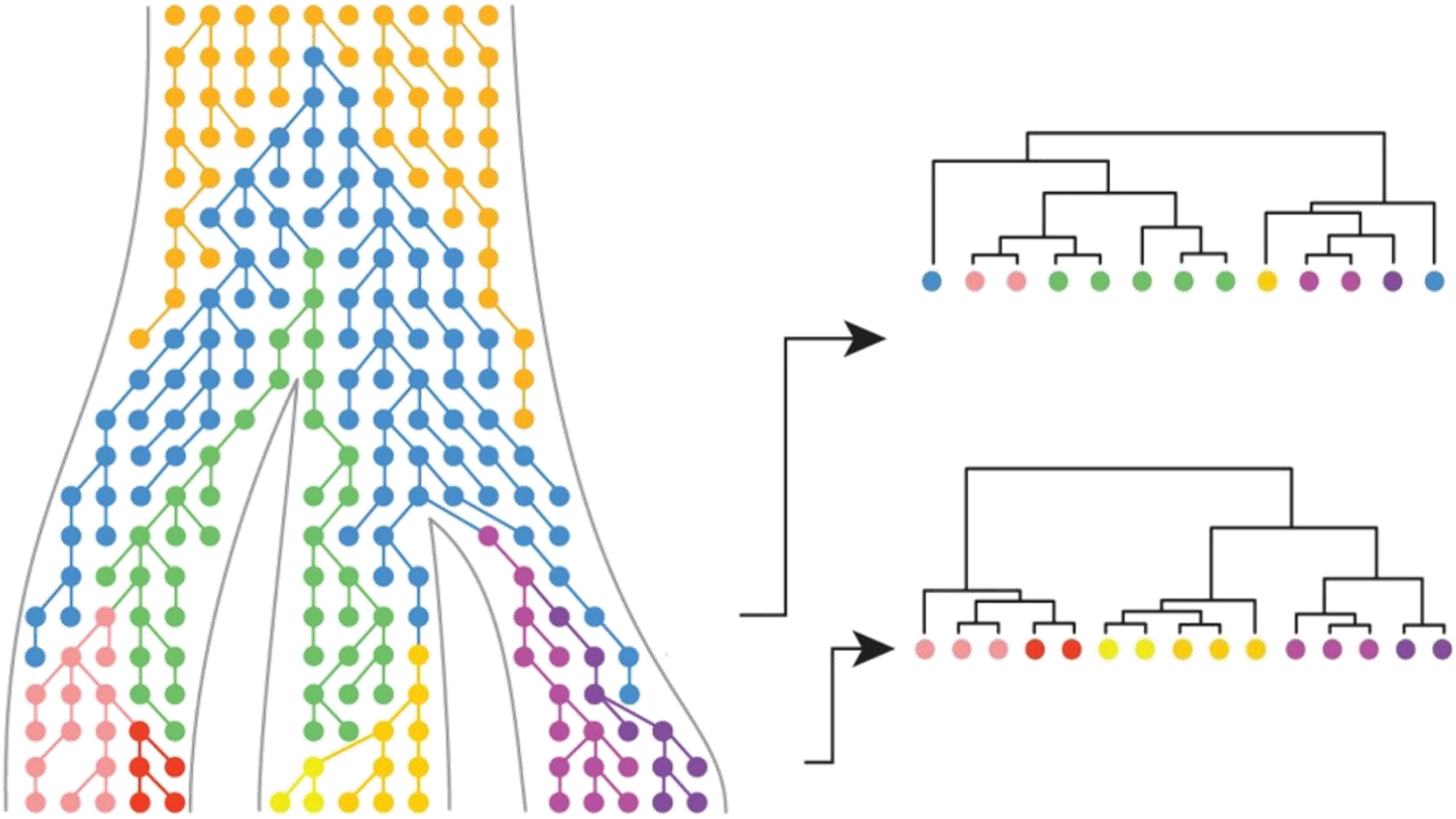


Distinguishing between sources

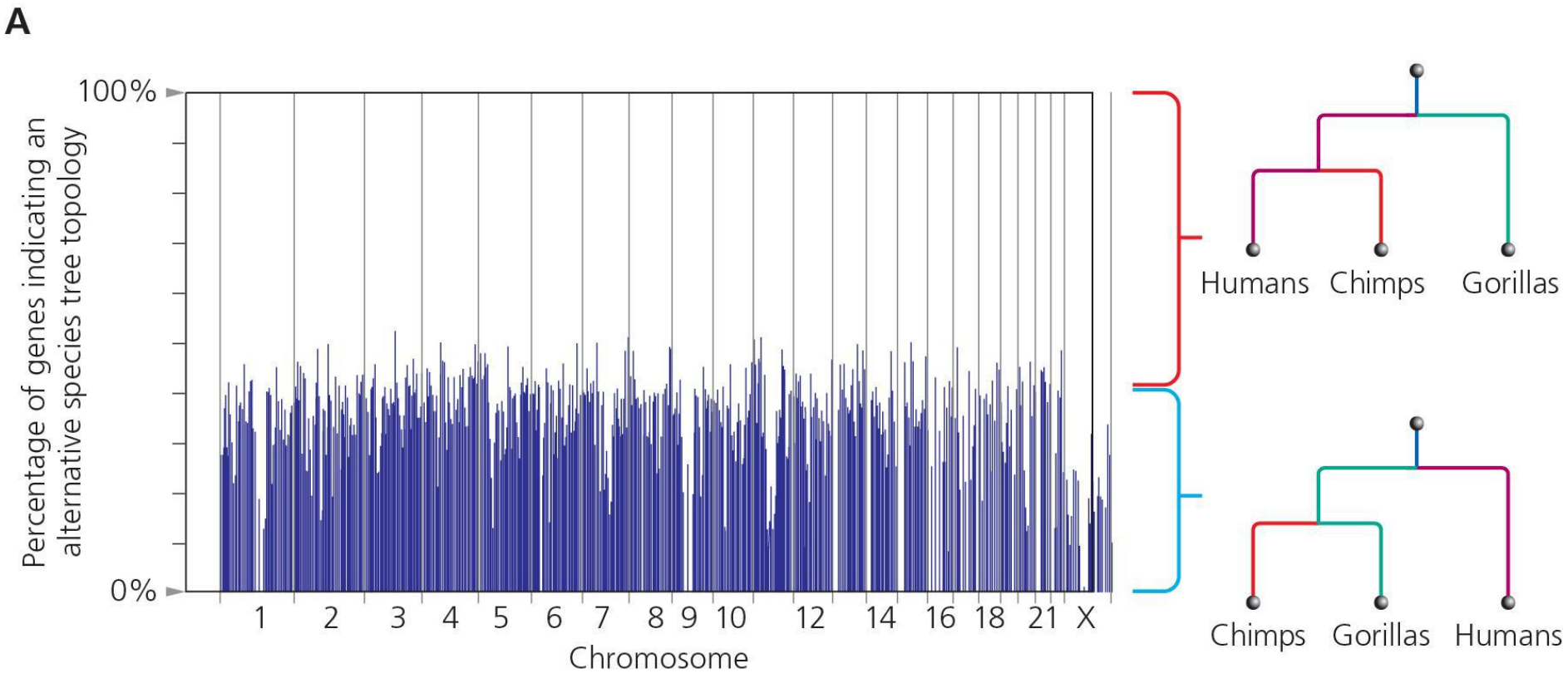
Test for mutational variance as a source of mtDNA discordance by performing a parametric bootstrap using the independently estimated species tree as the constraint tree.

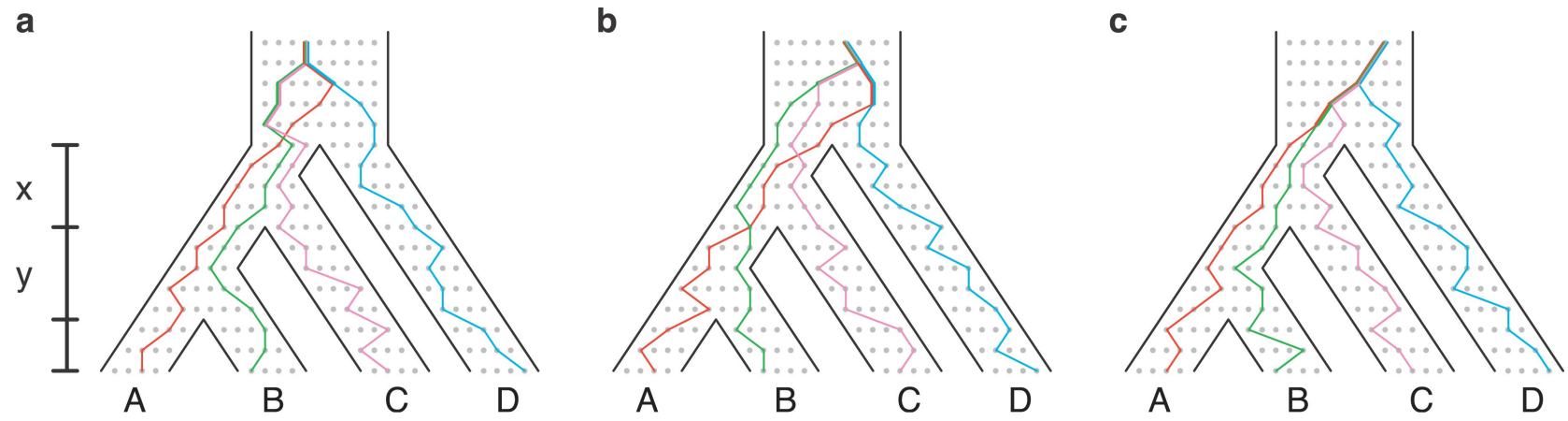
If mutational variance is unlikely, test for coalescent stochasticity as a source of mtDNA discordance by simulating genealogies on an independent estimate of the species tree.

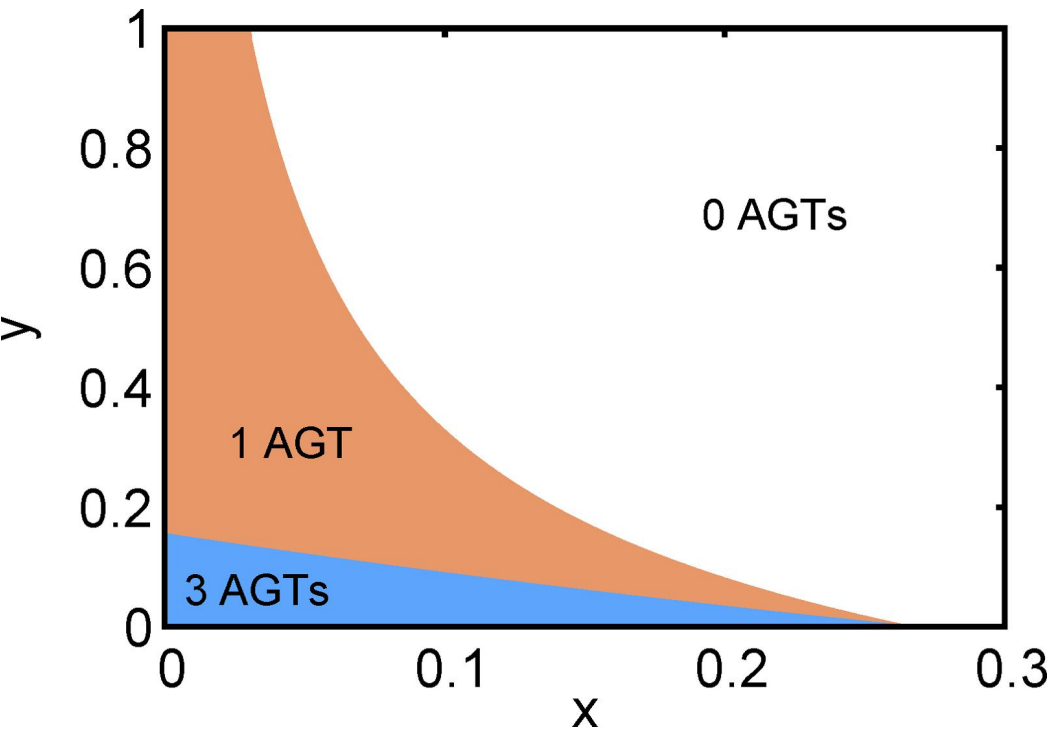
If both mutational and coalescent sources of discordance are rejected, hybridization is left as a likely explanation.



Incomplete lineage sorting


A
100%
0%
Percentage of genes indicating an alternative species tree topology
1
2
3
4
5
6
7
8
9
10
12
14
16
18
21
X
Chromosome
Humans
Chimps
Gorillas
Chimps
Gorillas
Humans





The “Anomaly zone”

AGT - Anomalous gene tree with higher likelihood than the tree species topology

ILS will result when branches are short & population sizes are large

(often mistakenly thought only to occur in recent radiations, but ancient short internal branches just as at risk!)

The Coalescent

Coalescence - MRCA of a pair of genes

Built on standard population genetics (e.g. Wright-Fisher model)

Key parameter - Effective population size

Expected # of generations to coalescence - 2N

The Coalescent

What is the probability two copies of a gene in a randomly breeding population have an ancestor 1 generation ago?

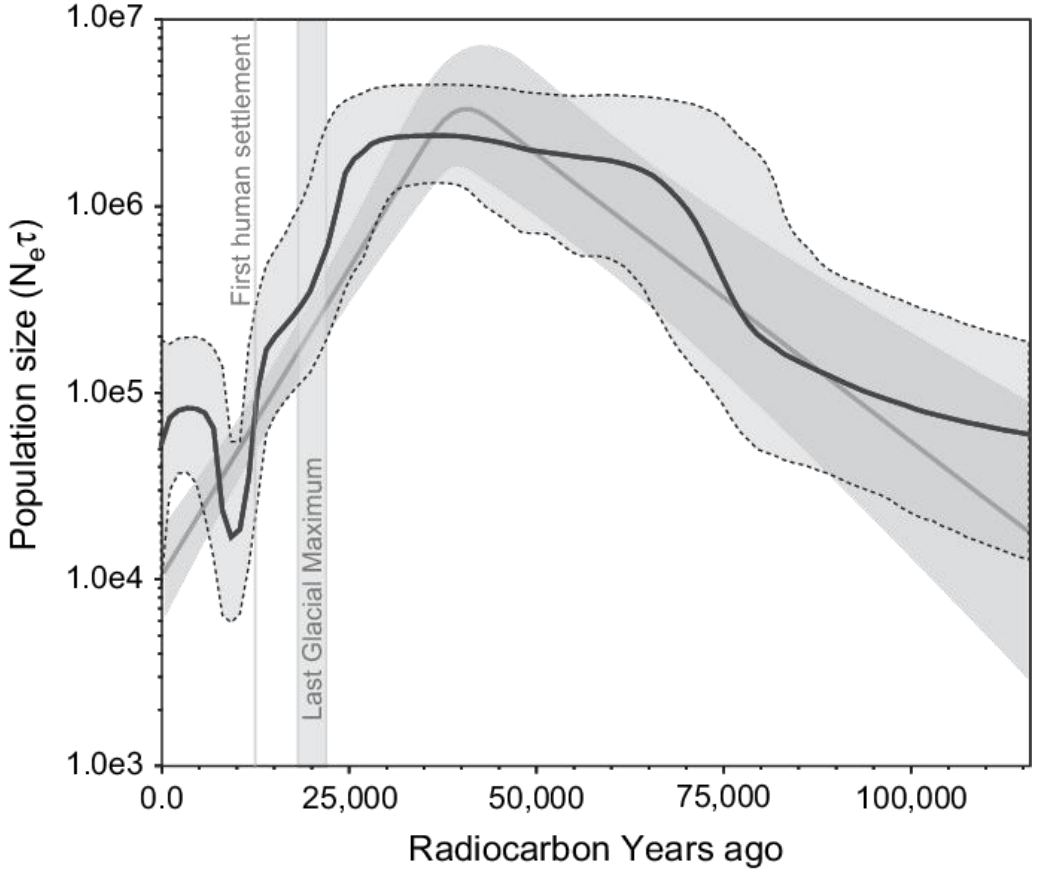
$$1/(2Ne)$$

What about j generations ago?

$$(1-1/(2Ne))^{j} * 1/(2Ne)$$

The Coalescent

Coalescence-time measured in units of *Ne*



Bayesian Coalescent Inference of Past Population Dynamics from Molecular Sequences

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Department of Zoology, University of Oxford, Oxford, United Kingdom

The Multispecies Coalescent

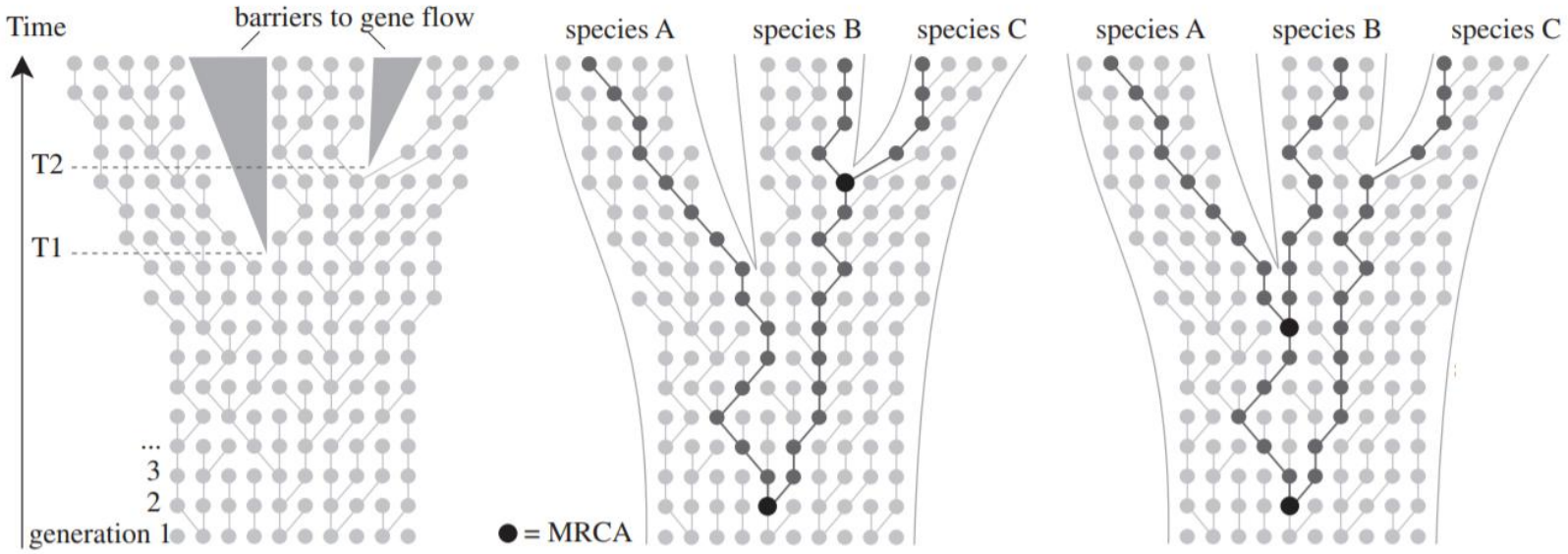
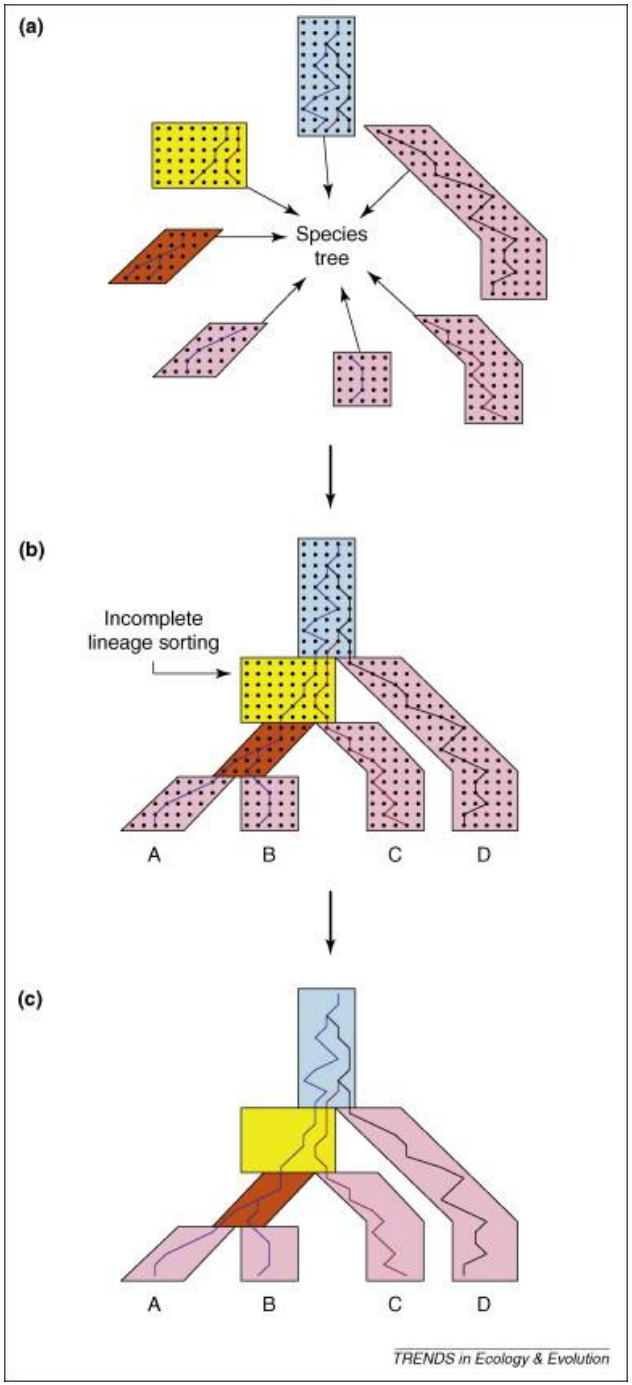


Image: Leliaert, F., Verbruggen, H., Vanormelingen, P., Steen, F., López-Bautista, J. M., Zuccarello, G. C., & De Clerck, O. (2014). DNA-based species delimitation in algae. European journal of phycology, 49(2), 179-196.



$P(D_i \mid Q_i, \pi_i, \boldsymbol{\psi_i})$ = standard likelihood of gene tree

$P(\boldsymbol{\psi_i} \mid S)$ = Likelihood of gene tree given the species tree

"AND" rule:

$P(D_1, D_2 \ldots D_n \mid S) =$

$P(D_1 \mid Q_1, \pi_1, \boldsymbol{\psi_1}) * P(\boldsymbol{\psi_1} \mid S) \times \ldots$

$\times P(D_n \mid Q_n, \pi_n, \boldsymbol{\psi_n}) * P(\boldsymbol{\psi_n} \mid S)$

Methods & software

Parsimony - “MDC” species tree that minimizes deep coalescences (can be inconsistent estimator)

ML - STEM (Kubatko & Degnan 2007). Requires gene trees to be well-estimated and clock-like

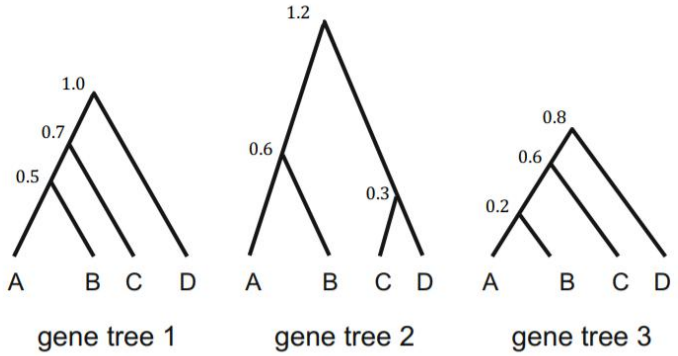
Bayesian - BEST, *BEAST, BPP. Bayesian approaches that integrate over uncertainty in gene trees. Great models...but complex and hard to converge!

$$P(S \mid D) \propto \int_G \left(\prod_{i=1} P(d_i \mid g_i) P(g_i \mid S)\right) P(S) dG,$$

Other methods

Concordance analysis- BCA/BUCKy. Semi-parametric clustering of gene trees into “concordance blocks” without regard to process

Summary methods- Uses properties of multispecies coalescent to summarize gene trees. STAR/STEAC/GLASS



gene tree 1 gene tree 2 gene tree 3

(b)

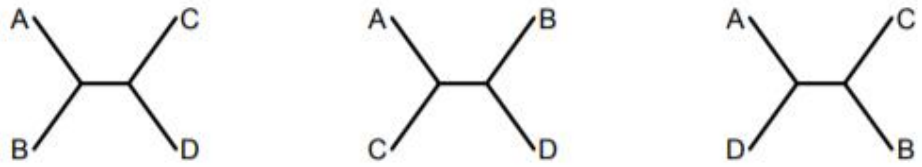
	A	B	C	D
A	--	0.2	0.6	0.8
B	0.2	--	0.6	0.8
C	0.6	0.6	--	0.3
D	0.8	0.8	0.3	--

topology of the GLASS tree

Other methods

Quartets approaches: ASTRAL/SVDQuartets

Avoids calculating full likelihood, instead focuses on site patterns over 4 taxon combinations. Good for SNPs and genomic scale data

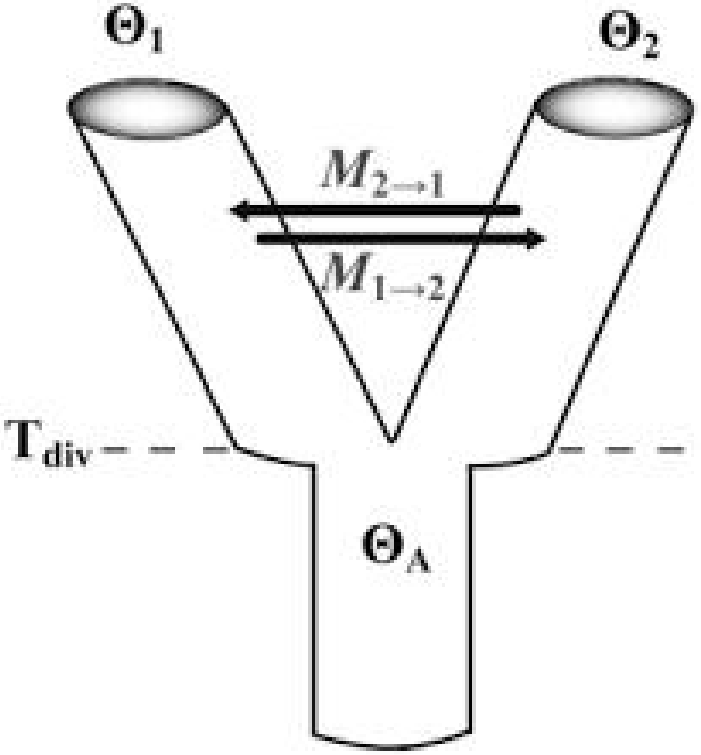


Species Tree Inference Summary – Comparison of Methods

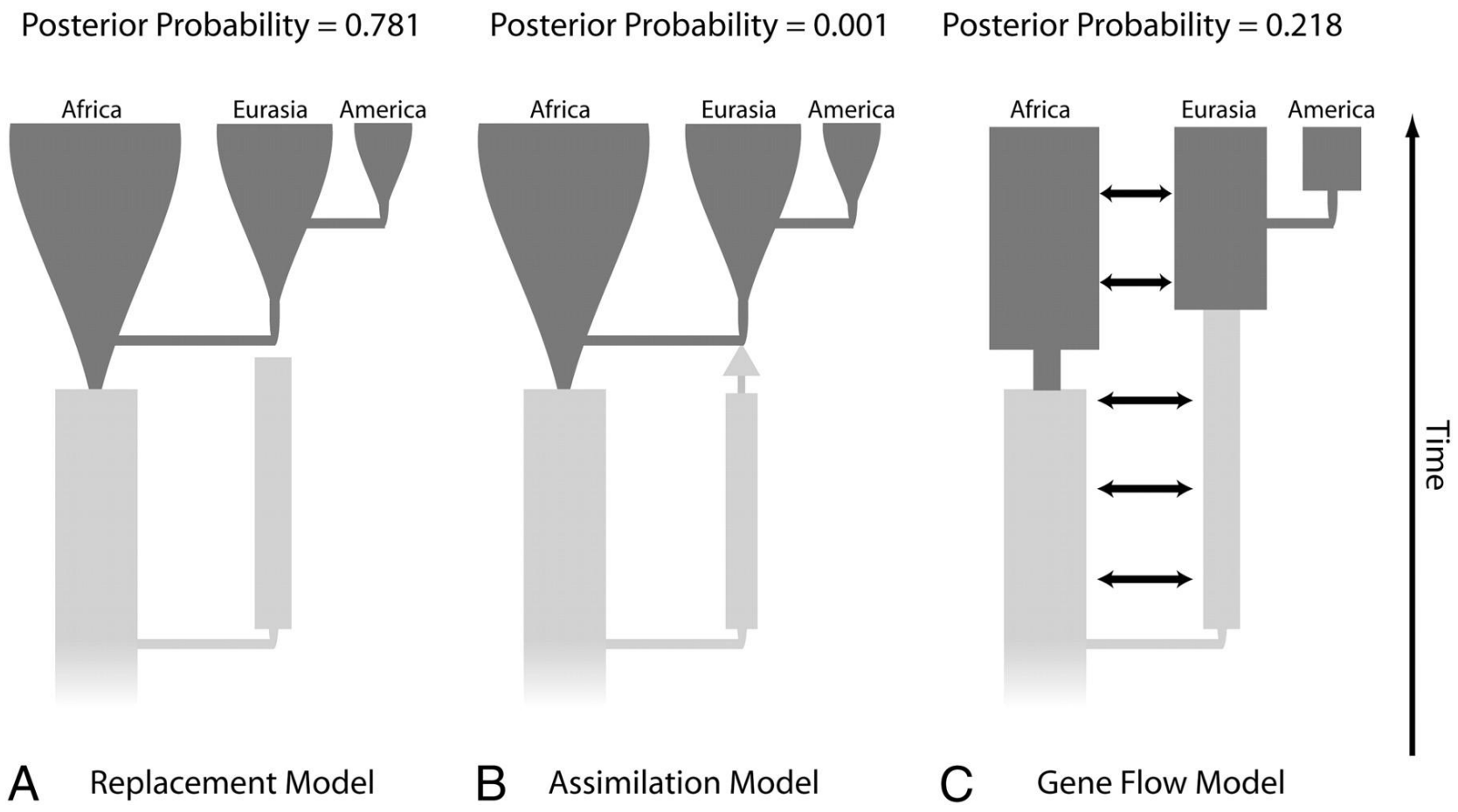
Software	Data Type	Measure of Uncertainty	Computation Time	Models Included
BEST	multilocus	posterior probability	long; can be run in parallel	coalescent; all reversible substitution models
*BEAST	multilocus	posterior probability	intermediate; can be run in parallel	coalesent; all reversible substitution models; relaxed clock; variable population sizes
BPP	multilocus	posterior probability	long	coalescent; JC69 model only; species delimitation
SVDQ	multilocus; SNP	bootstrap	short	coalescent; all reversible substitution models; parameter estimation ?
SNAPP	biallelic SNP; AFLP	posterior probability	long; can be run in parallel	coalescent; two-state substitution model; Bayes factor delimitation
ASTRAL	unrooted gene trees	bootstrap	short given gene trees	no specific model assumed
MP-EST	rooted gene trees	bootstrap	short given gene trees	coalescent model

Adding gene flow...

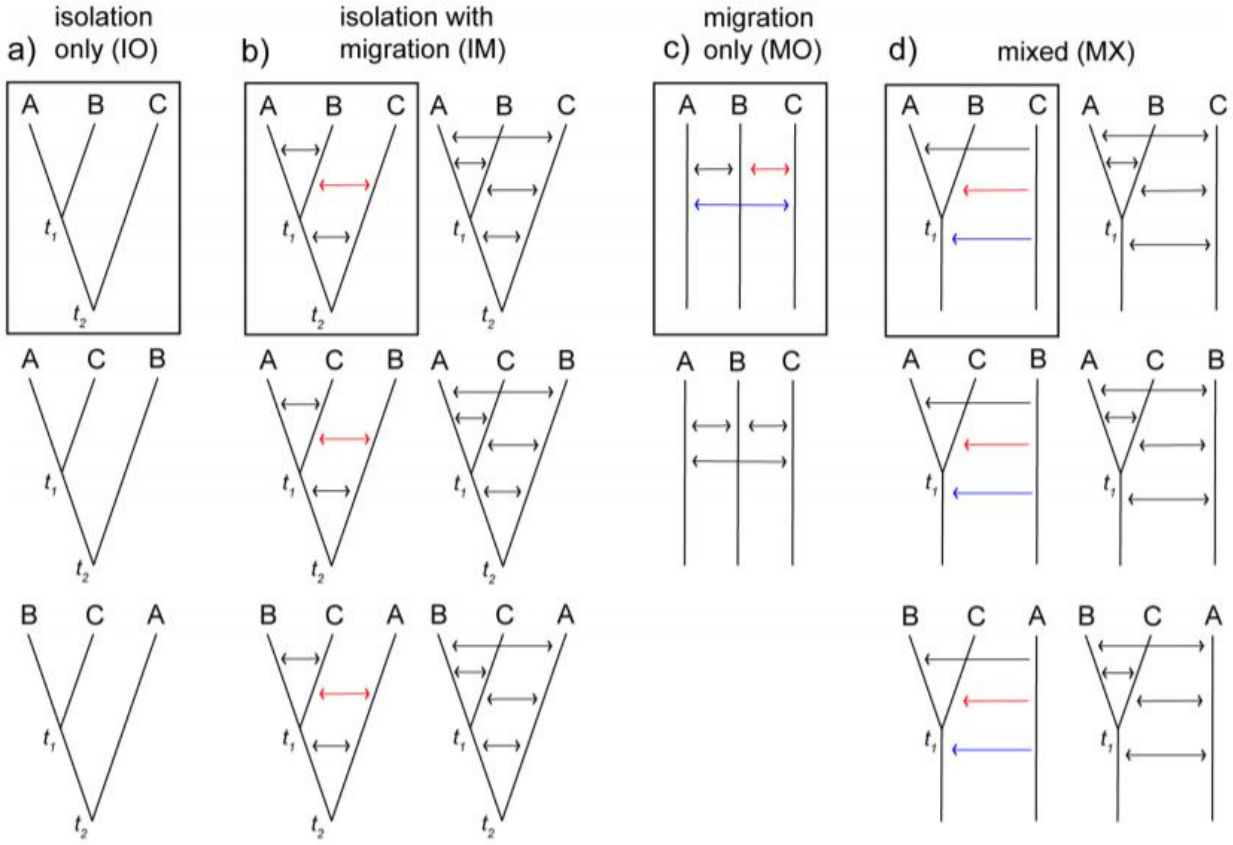
Limited to a small number of species (IM & IMa)



Hypothesis testing



Search among all possible models...
PHRAPL (Jackson et al. 2017)



Some general thoughts...

Scaling multispecies coalescent to genomic scale is hard, adding more data doesn't necessarily improve estimation

Filter genes to those with strong phylogenetic signal

Interrogate your data

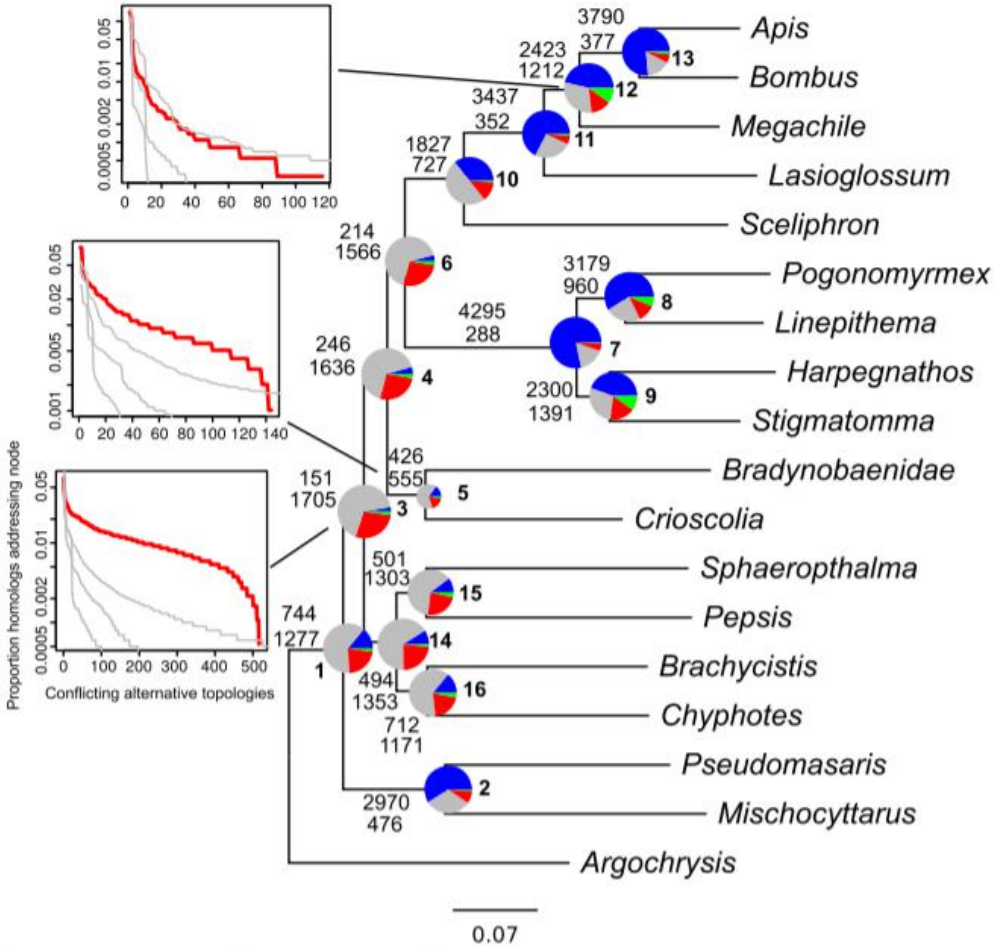


Fig. 2 Combined ML (species tree) topology for Hymenoptera, with summary of conflicting and concordant homologs. For each branch, the top number indicates the number of homologs concordant with the species tree at that node, and the bottom number indicates the number of homologs in conflict with that clade in the species tree. The pie charts at each node present the proportion of homologs that support that clade (blue), the proportion that support the main alternative for that clade (green), the proportion that support the remaining alternatives (red), and the proportion that inform (conflict or support) this clade that have less than 50 % bootstrap support (grey). The histograms show, for three nodes, the proportion of the total homologs that support each conflicting alternative resolution for the clade in question, sorted from largest to smallest. Grey lines represent distributions of conflicting alternative resolutions based on coalescent simulations generated with three tree heights. The histograms for other nodes are presented in Additional file 2: Figure S5