Species tree estimation under multispecies coalescent

Ziheng Yang

Department of Genetics, Evolution, and Environment

University College London

Outline

- Species tree estimation in the case of 3 and 4 species
- The anomaly zone
- MCMC moves that change the species tree
- Simulation and empirical results

MSC, 3 species, 3 sequences

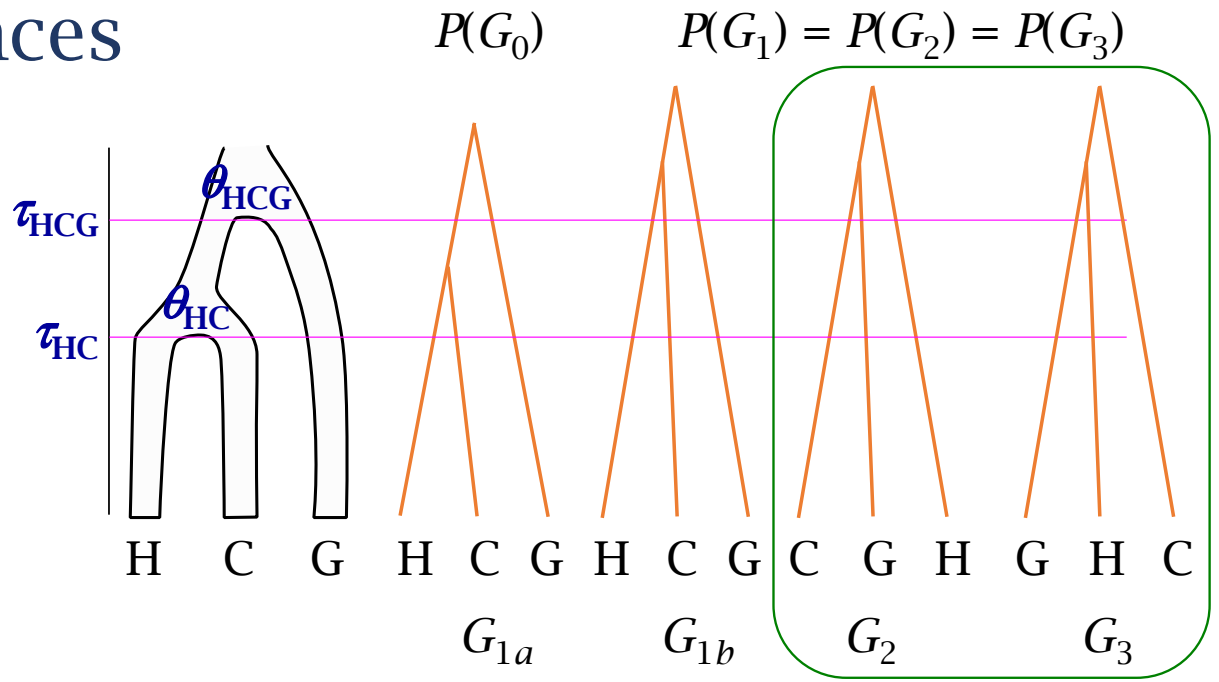
Parameters:

Speciation times:

τ_{HC}, τ_{HCG}

Population sizes:

θ_{HC}, θ_{HCG}



$$P_{\text{mismatch}} = P(G_2) + P(G_3) = \tfrac{2}{3}\left[1 - P(G_0)\right] = \tfrac{2}{3}\mathrm{e}^{-2(\tau_{HCG} - \tau_{HC})/\theta_{HC}}$$

For human-chimpanzee-gorilla, P_{mismatch} = 30%.

Hudson, R. R. 1983. Evolution **37**:203-217.
Chen & Li 2001. Am. J. Hum. Genet. **68**:444-456

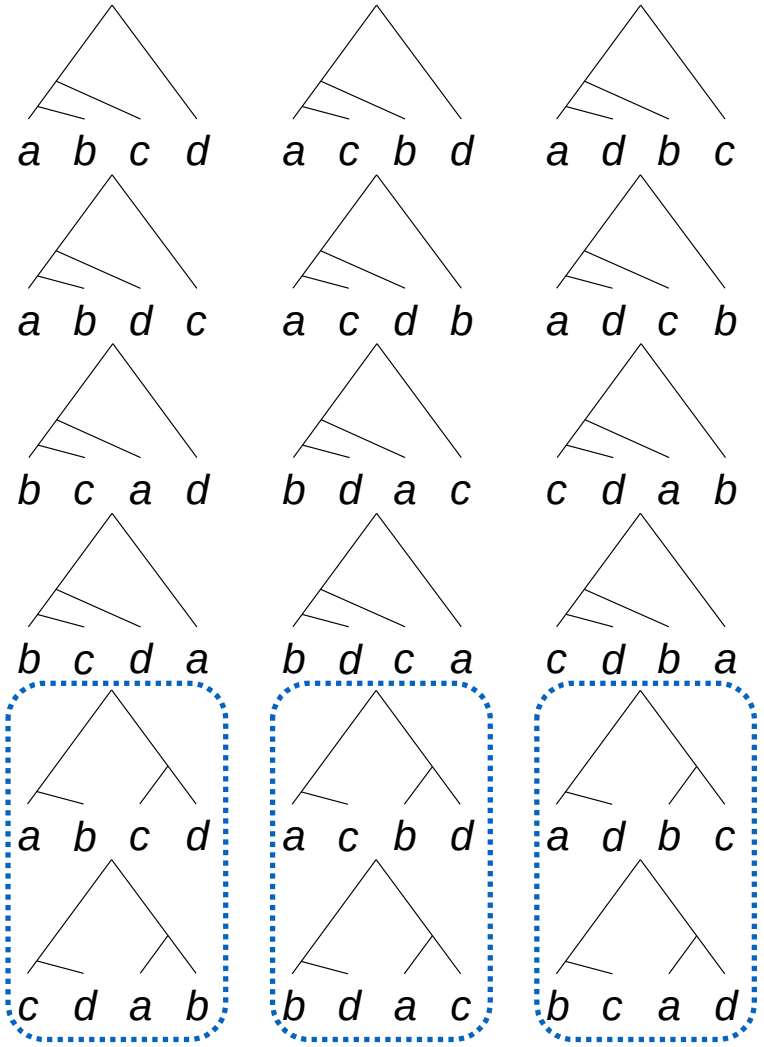
MSC: 4 sequences from 4 species

The probability for each of the 15 (rooted) gene trees is a sum over the compatible labelled histories.

$\tau_{ABCD} > \tau_{ABC} > \tau_{AB}$

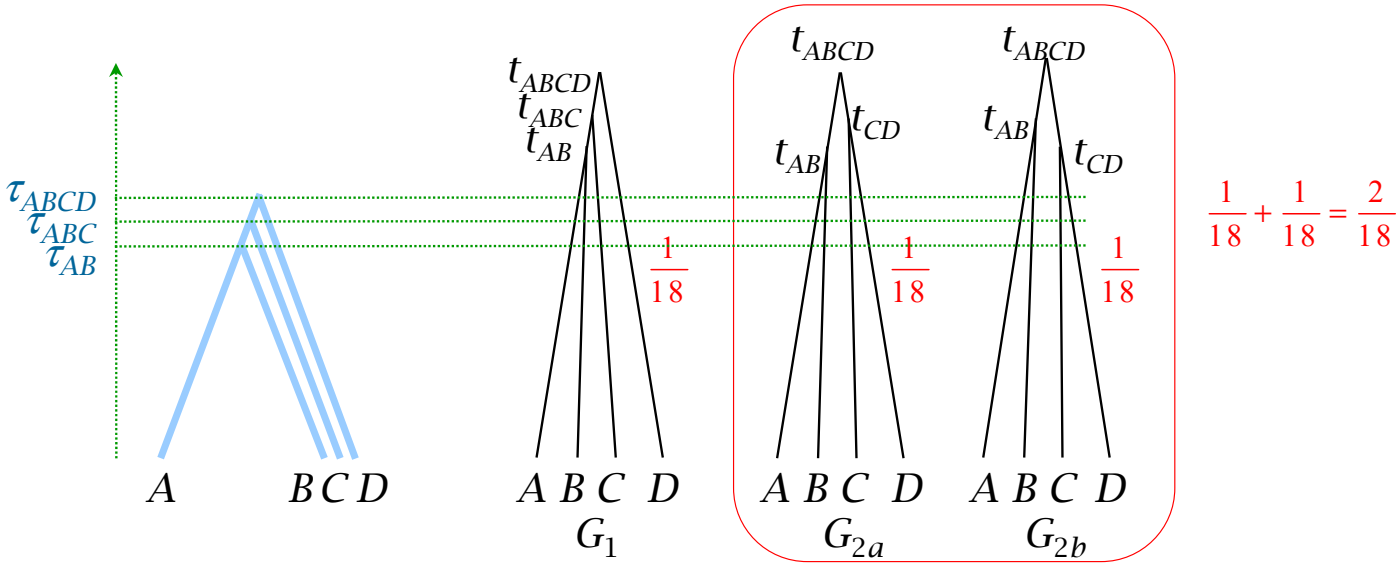
$\tau_{ABCD} \approx \tau_{ABC} \approx \tau_{AB}$

Species tree



MSC, 4 species, anomaly zone

For species tree and parameters (τs & θs) in the anomaly zone, the most probable gene tree has a different topology from the species tree.



Species tree Three labelled histories

- For 4 seqs, there are 15 rooted tree topologies (3 balanced & 12 unbalanced) and 18 labelled histories.
- If $\tau_{ABCD} = \tau_{ABC} = \tau_{AB}$, $f(G_1) = f(G_{2a}) = f(G_{2b}) = 1/18$.
- If the τs are close, it will be possible for $f(G_1) < f(G_{2a}) + f(G_{2b})$.

The anomaly zone

- If the most probable gene tree differs from the species tree, the species tree is said to be in the **anomaly zone**.
- If the species tree is in the anomaly zone, the method of using the most common gene tree as the species tree estimate (called the *majority-vote* method) will be inconsistent (misleading).
- Anomaly zone does not exist for 3 species.
  It exists for the unbalanced species tree of 4 species, and exists for any species tree of ≥5 species.

Degnan JH, Salter LA (2005 Evolution 59:24-37)
Degnan JH, Rosenberg NA (2006 PLoS Genet. 2:e68)

MCMC samples from the posterior:
$f(S, \{\tau s, \theta s\}, \{G_i, t_i\} \mid \text{Data})$

1. Initialize S, $\{\theta s, \tau s\}$, $\{G_i, \boldsymbol{t}_i\}$.
2. Iterate
   - Change parameters (θs, τs in the model).
   - Change gene trees $\{G_i, \boldsymbol{t}_i\}$.
   - Change species tree S (by NNI, SPR, NodeSlider).
   - Save on the disk every k iterations.

S: species tree
$\{\theta s, \tau s\}$: parameters in the MSC
$\{G_i, \boldsymbol{t}_i\}$: gene trees and ages

The MCMC algorithm visits the species trees according to their posterior probabilities.

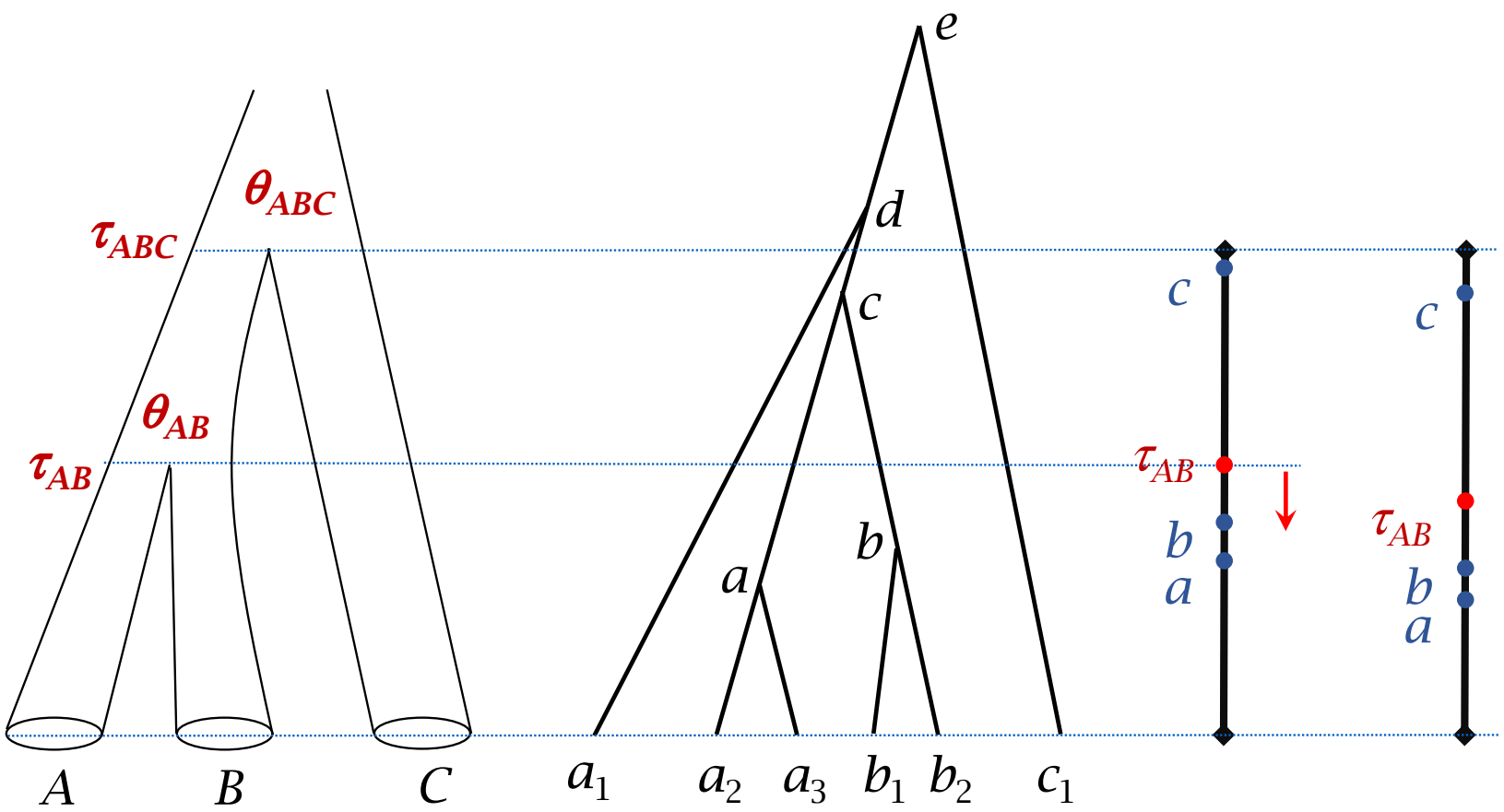
The constraint that gene trees fit inside species tree is a major issue when we design MCMC moves to change the species tree.

(a) Rubber-bound algorithm to change the species divergence time τ in the multispecies coalescent model in MCMC (Rannala & Yang 2003 Genetics)

(b) Rubber-bound with proportional scaling to change the species delimitation model in rjMCMC (Rannala & Yang 2013 Genetics)

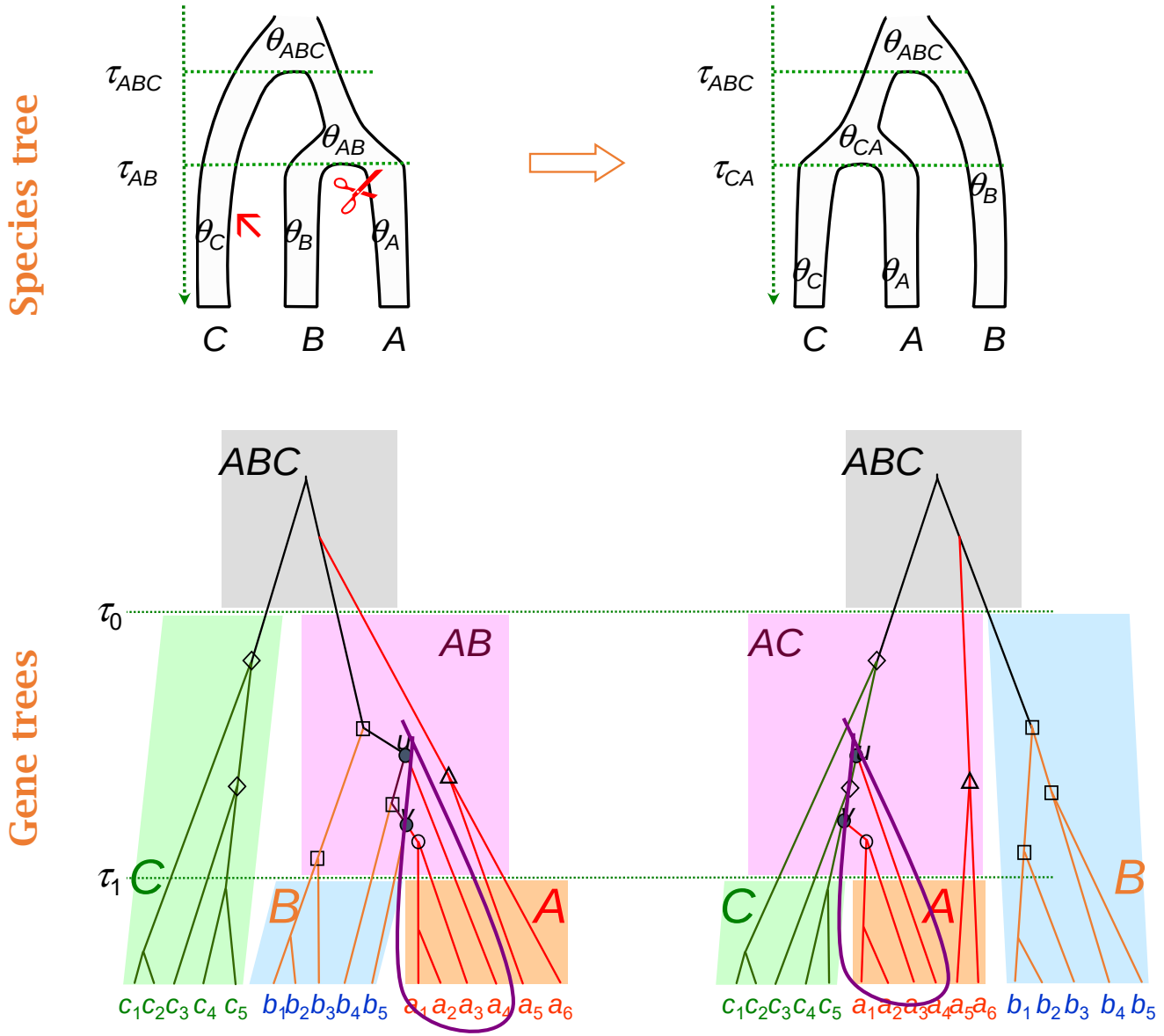
(c) NNI, SPR, & NodeSlider to change the species tree in transmodel MCMC (Yang & Rannala 2014 MBE; Rannala & Yang 2017 Syst Biol)

Rubber-band algorithm to change τ



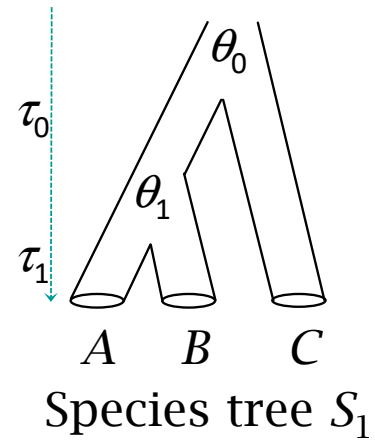
(Rannala & Yang, 2003 Genetics)

Species tree change by NNI



(Yang & Rannala 2014 MBE)

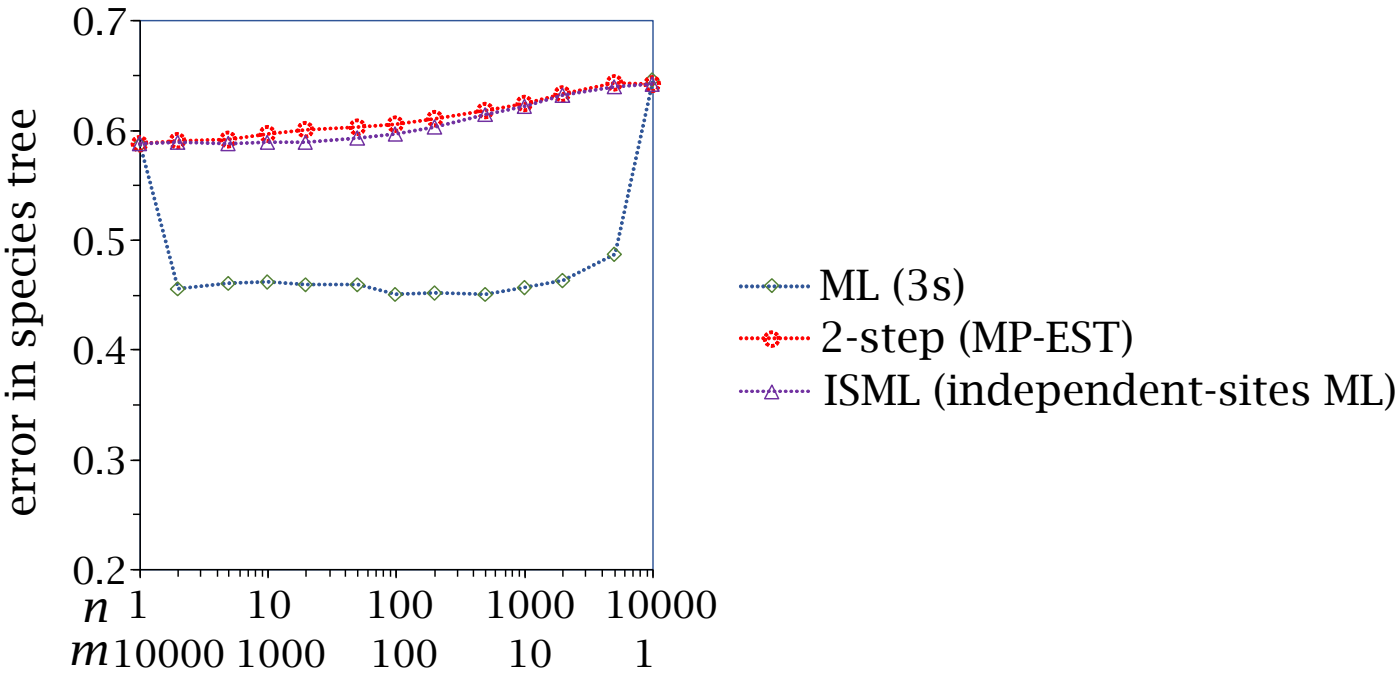
Species tree estimation in the case of 3 species



Species tree S_1

$\tau_0 = 0.02$, $\tau_1 = 0.019$,
$\theta_0 = 0.01$, $\theta_1 = 0.05$

(c) $nm = 10^4$ fixed, n sites m loci



There is more information in 2 genes each of 5000 sites than in 10000 independent sites.

Zhu T, Yang Z. 2021. Complexity of the simplest species tree problem. Mol Biol Evol. 10.1093/molbev/msab009

Full likelihood methods are much more efficient when the species tree is hard.

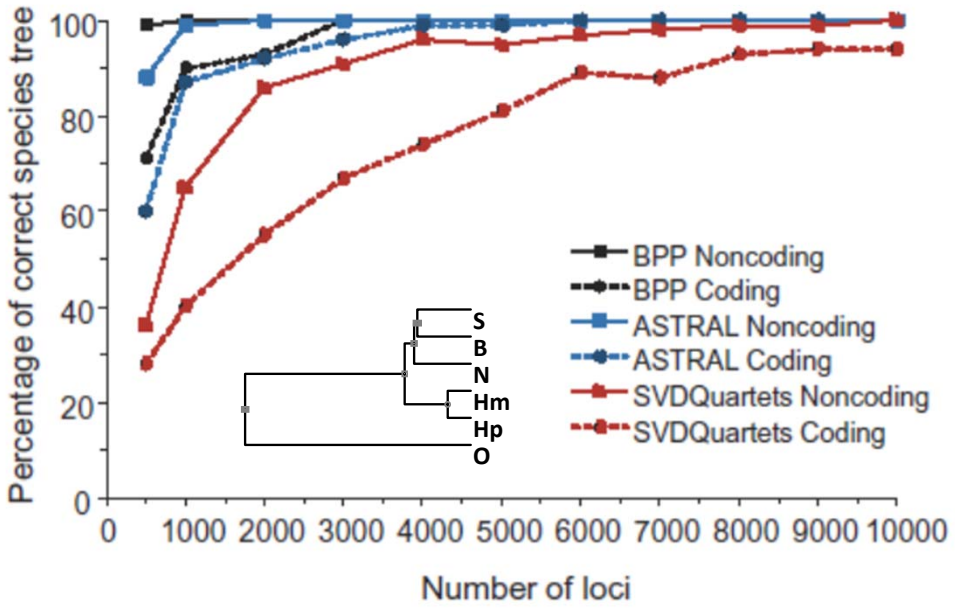


FIG. 6. The percentage of correct species trees in simulated data sets

(Shi & Yang 2018. Mol Biol Evol 35:159-179)

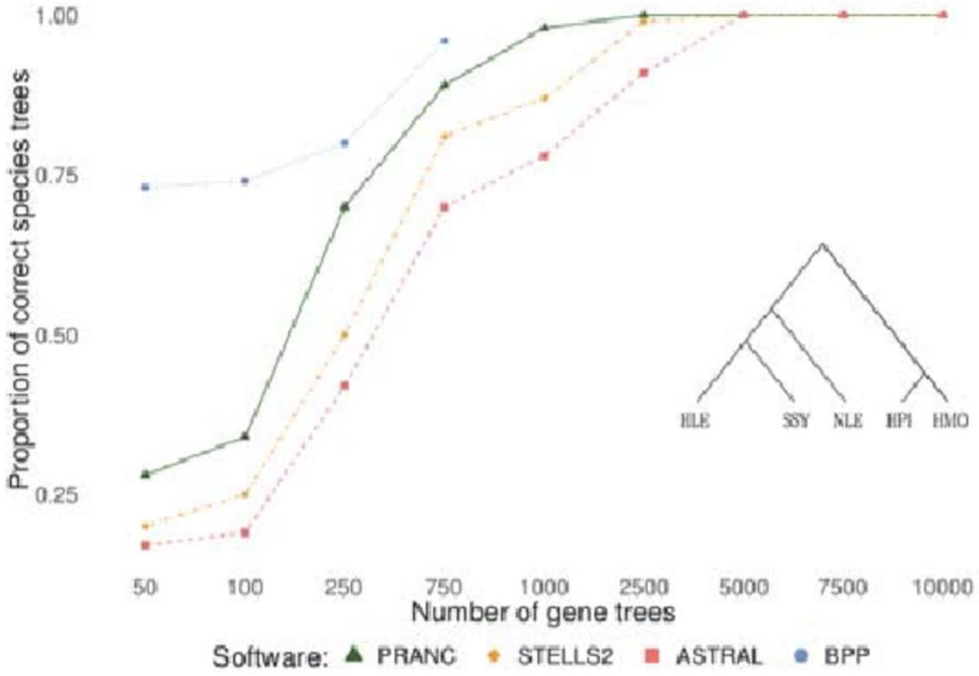


Fig. 1. The proportion of correct species trees in a gibbon dataset obtained by four different

(Kim A, Degnan J. 2020. *Bioinformatics* 10.1093/bioinformatics/btaa605)

Summary

- MSC (with and without introgression) is a powerful framework for analysis of genomic sequence data from closely related species.
- Full likelihood methods account for genealogical variations across the genome (among loci), as well as phylogenetic uncertainties in gene trees.
- In challenging species tree problems, summary methods have far poorer statistical properties but they run much faster.
- Deep coalescence and introgression are major challenges to species tree inference for shallow phylogenies. Deep trees are even harder.

References

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