Phylogenetics

Morphological Substitution models

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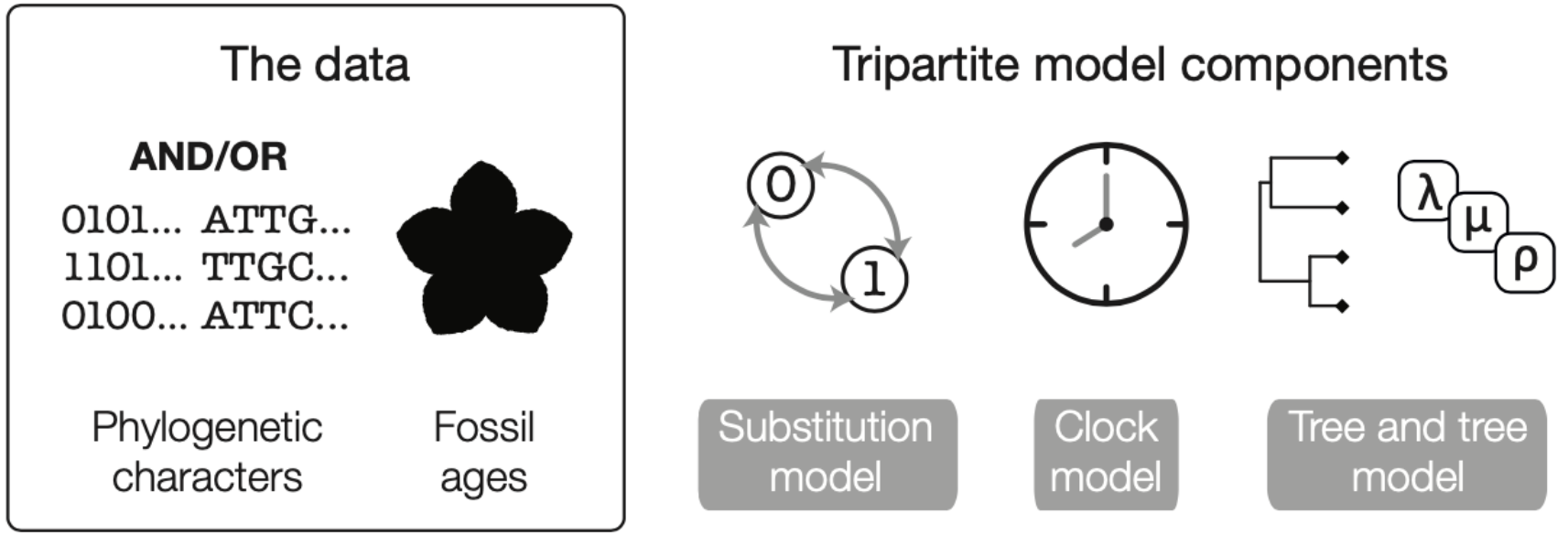
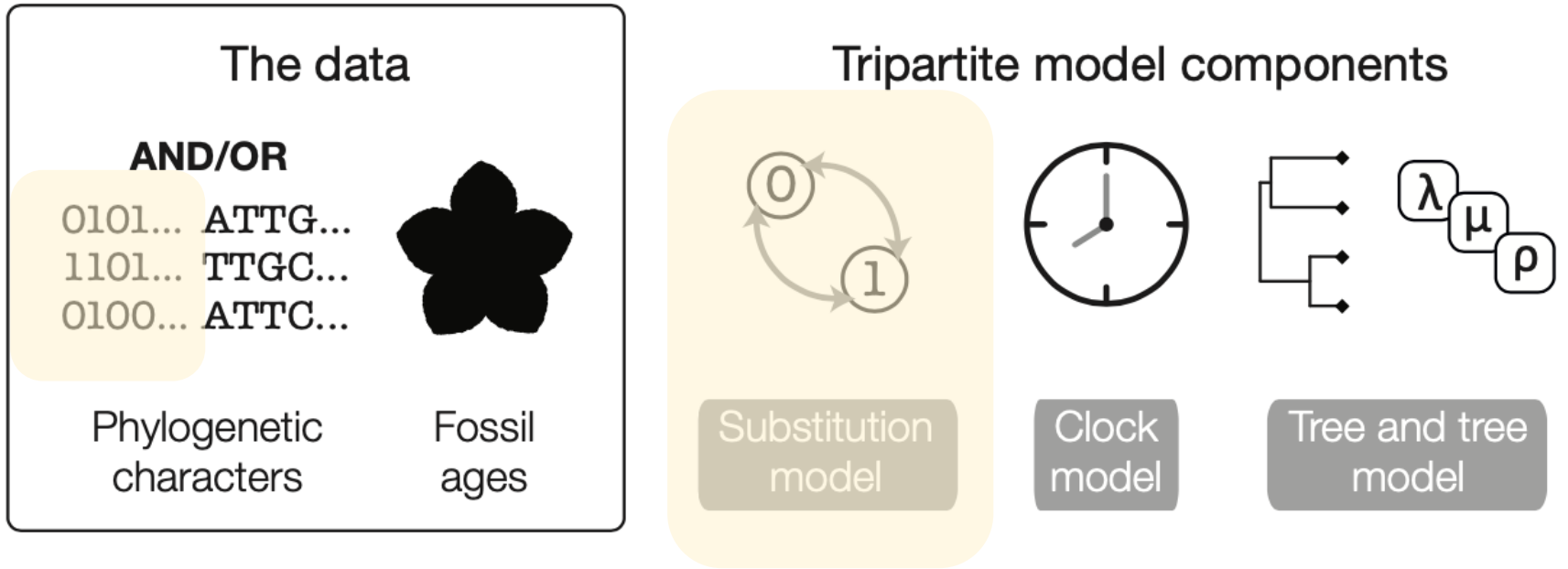
Bayesian Phylogenetic Analysis Components



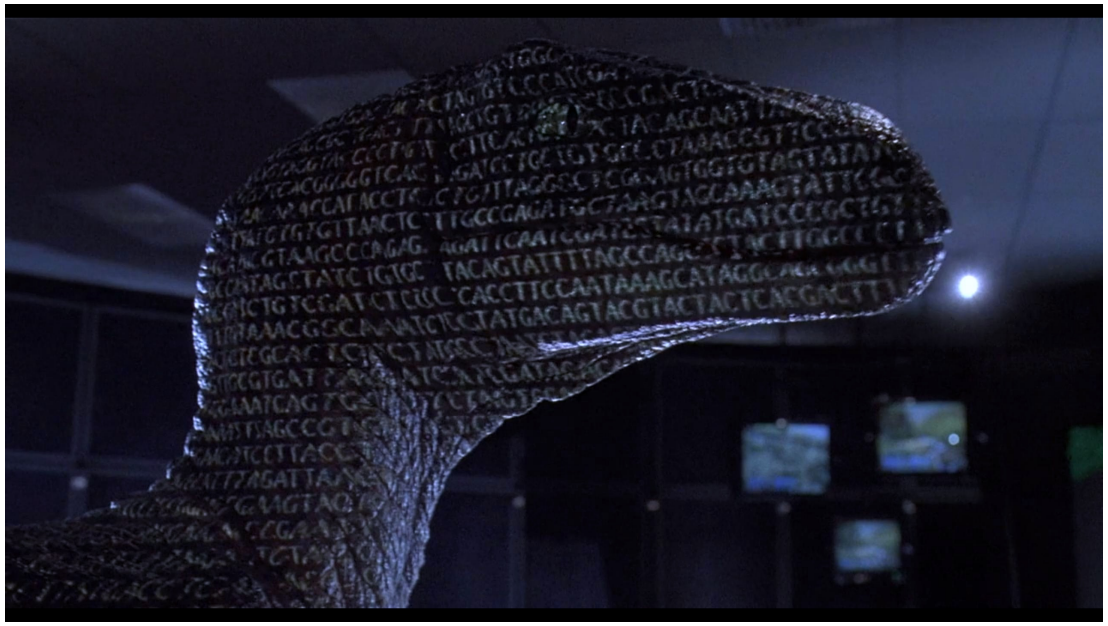
Bayesian Phylogenetic Analysis Components



Molecular Substitution models

JC substitution model

$$Q = \begin{pmatrix} -\mu_A & \mu_{AG} & \mu_{AC} & \mu_{AT} \\ \mu_{GA} & -\mu_G & \mu_{GC} & \mu_{GT} \\ \mu_{CA} & \mu_{CG} & -\mu_C & \mu_{CT} \\ \mu_{TA} & \mu_{TG} & \mu_{TC} & -\mu_T \end{pmatrix}$$



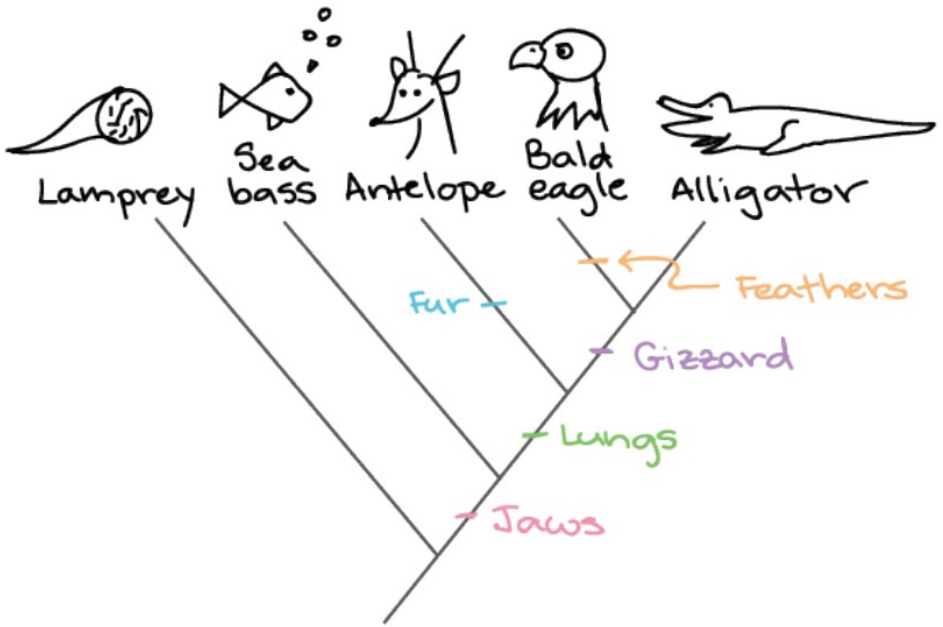
GTR substitution model

$$Q = \begin{pmatrix} * & \mu_{AG}\pi_G & \mu_{AC}\pi_C & \mu_{AT}\pi_T \\ \mu_{GA}\pi_A & * & \mu_{GC}\pi_C & \mu_{GT}\pi_T \\ \mu_{CA}\pi_A & \mu_{CG}\pi_G & * & \mu_{CT}\pi_T \\ \mu_{TA}\pi_A & \mu_{TG}\pi_G & \mu_{TC}\pi_C & * \end{pmatrix}$$

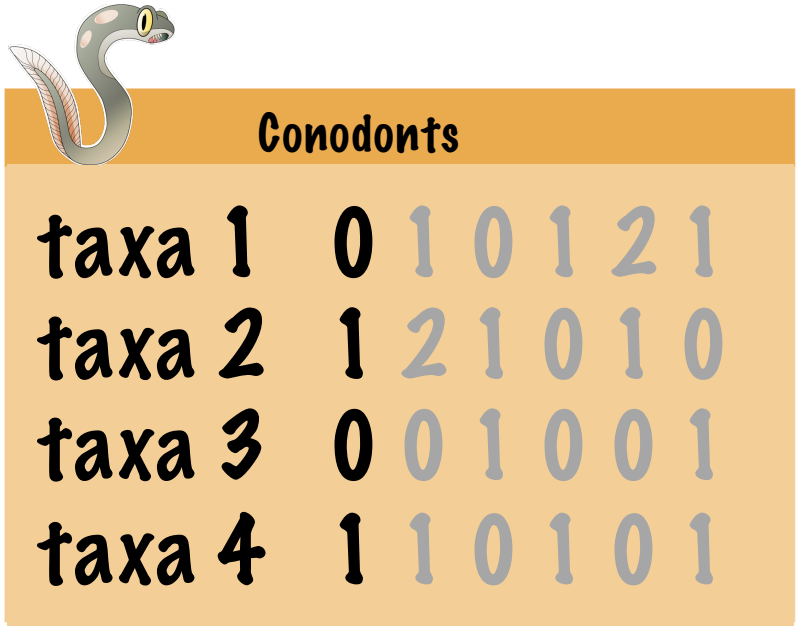
μ = substitution rate
Π = stationary frequency

Morphological data

	Lungs	Jaws	Feathers	Gizzards	Fur
taxa A	0	0	0	0	0
taxa B	1	1	0	0	1
taxa C	1	1	1	1	0
taxa D	1	1	0	1	0
taxa E	0	1	0	0	0



Issues with Morphological data



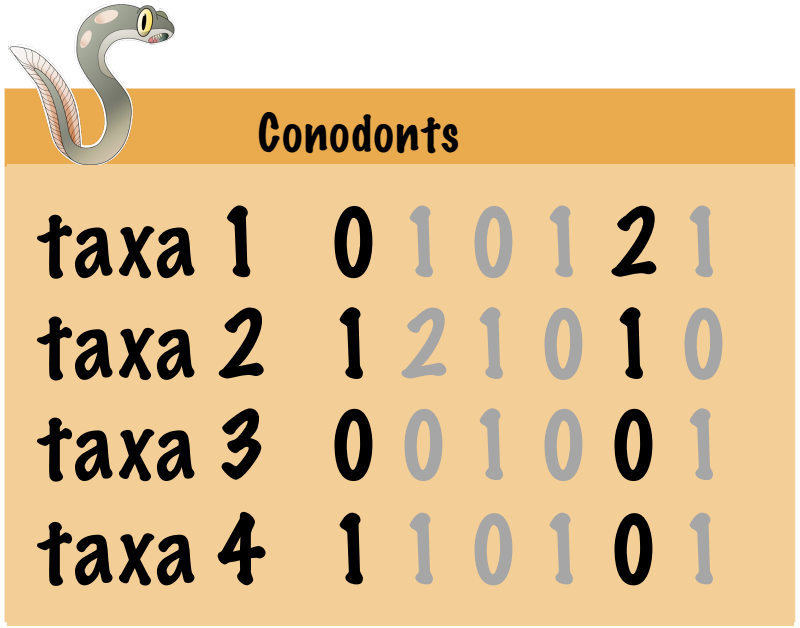
Often used to indicate presence absence data

Issues with Morphological data

Conodonts						
taxa 1	0	1	0	1	2	1
taxa 2	1	2	1	0	1	0
taxa 3	0	0	1	0	0	1
taxa 4	1	1	0	1	0	1

Multistate characters can be used to represent types of a trait

Issues with Morphological data



Trait 1		Trait 2
0	≠	0
1	≠	1

Generalising morphological data is much more difficult than molecular

Differences between molecular and morphological data to consider when modelling

Molecular data has a similar biological meaning throughout the alignment.

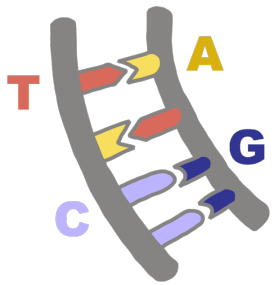
A “T” in one part of the alignment represents the same biological unit as a “T” somewhere else in the alignment.

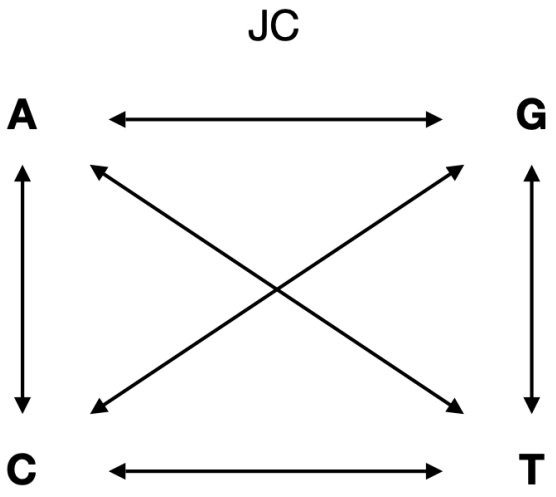
This is not the same for morphological data.

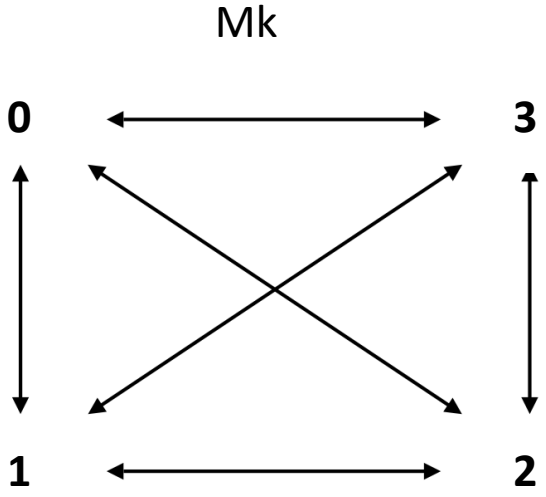
Becomes more **difficult to generalise** morphological data in any biologically meaningful way

What assumptions might you want to incorporate into a model of morphological character evolution?

Substitution models for morphological data





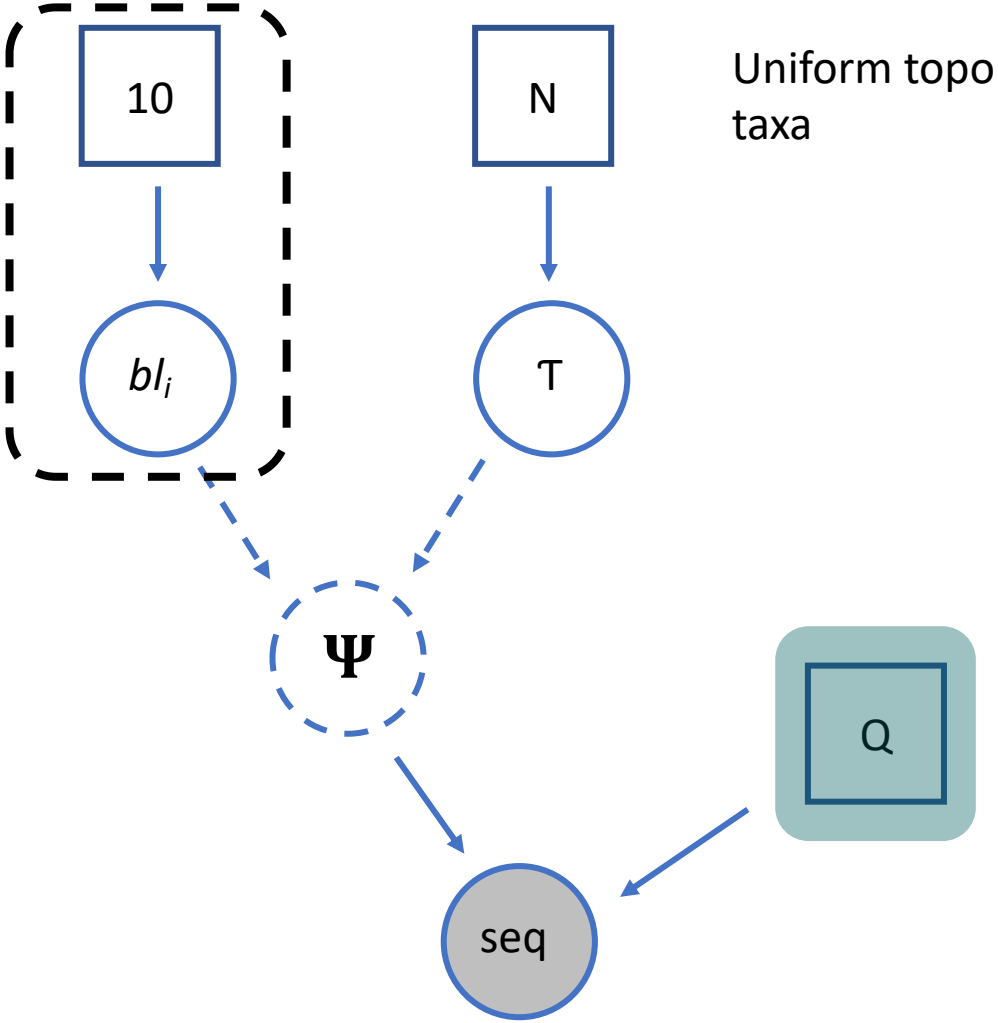


*4 state here as an example, can be any number from 2!

Line width represents the relative rate of change between different steps.

Mk Model

Exponential rate parameter of 10 on branch lengths.

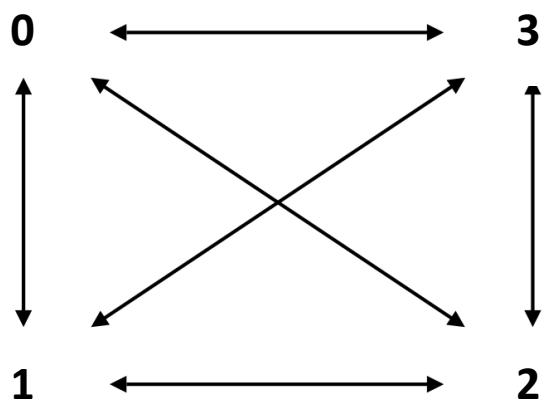


Uniform topology of N taxa

Mk model

Substitution models for morphological data

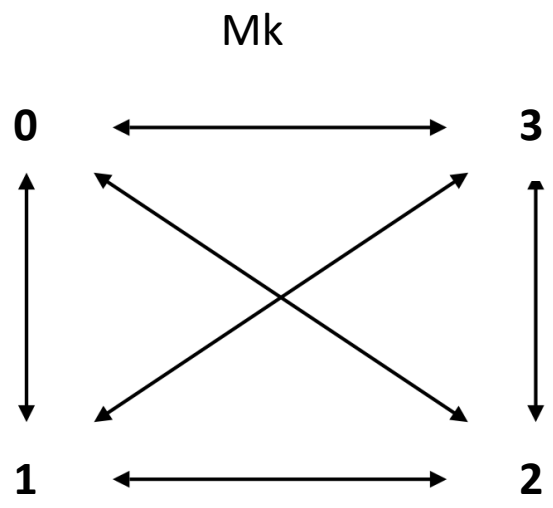
Mk



$$Q = \begin{pmatrix} -\mu_0 & \mu_{01} & \mu_{02} & \mu_{03} \\ \mu_{10} & -\mu_1 & \mu_{12} & \mu_{13} \\ \mu_{20} & \mu_{21} & -\mu_2 & \mu_{23} \\ \mu_{30} & \mu_{31} & \mu_{32} & -\mu_3 \end{pmatrix}$$

*4 state here as an example, can be any number from 2!

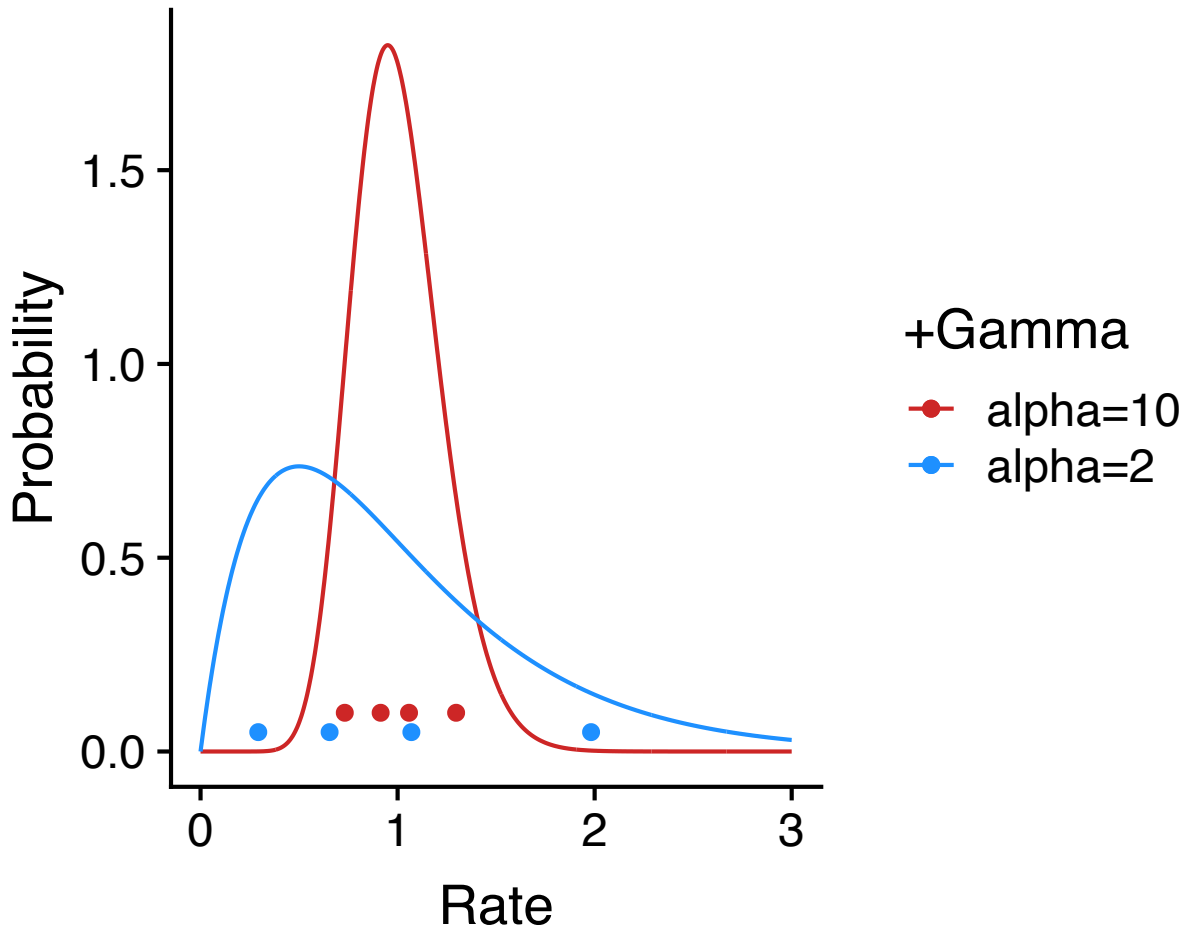
Substitution models for morphological data



We can **add extensions** to the standard Mk model in a number of ways

*4 state here as an example, can be any number from 2!

Across Site Rate Variation (+G)

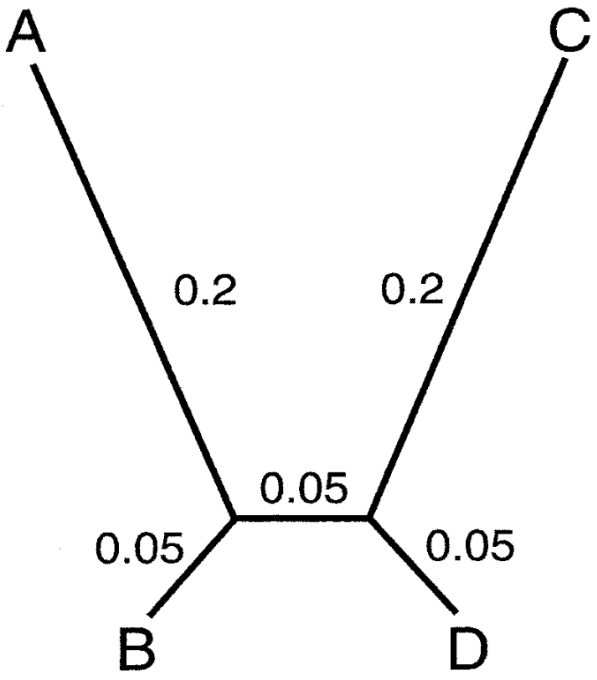


alpha = 10, the rates are similar
alpha = 2 the rates differ

This approach allows **faster evolving sites to evolve according to higher rates** and visa versa

Ascertainment Bias (V)

Conditions on the fact that all sites are variable



	True branch length	Mk (uncorrected)	Mkv (corrected)
Percent correct	—	74.0	99.8
Branch A	0.2	241,750 (±349,100)	0.206 (±0.060)
Branch B	0.05	0.43210 (±0.13756)	0.050 (±0.018)
Branch X	0.05	54.646 (±1,725.3)	0.052 (±0.023)
Branch C	0.2	143,950 (±228,910)	0.206 (±0.059)
Branch D	0.05	0.022 (±0.054)	0.051 (±0.019)

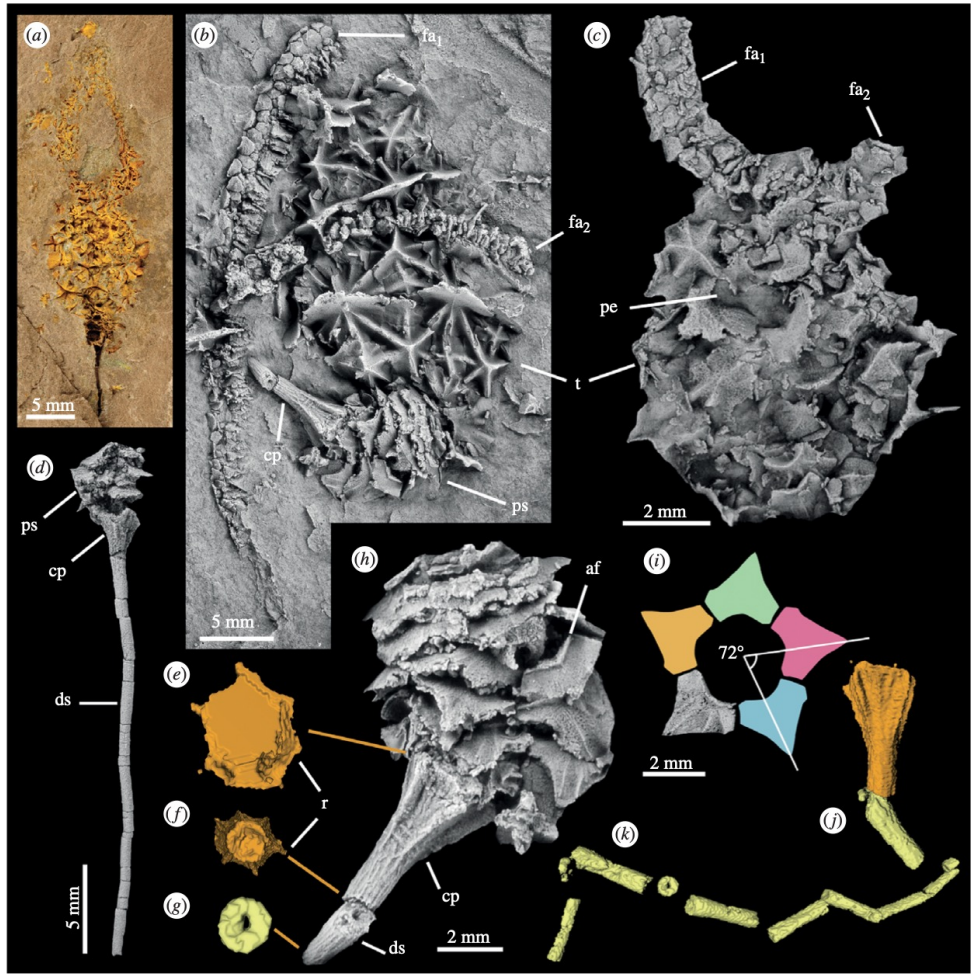
Lewis 2001

Partitioning the data

Researchers have argued that it is reasonable partition a morphological matrix by the number of character states

Taxa A	010023
Taxa B	201102
Taxa C	112131

```
0015100010?00-100--0000000000
0005000010?200100--0010010000
0025000010?200100--0?10010000
00?5?0010?200100?-0???010110
0015000101201000430100011111
0015000101201010440111011111
??050?????2010004 40?11011111
01050?010-210000?501??010110
0002000100210100 3-1110010110
0002000100211001441121011111
000201111-210010?-??11011121
?103?0?11?1001104-0000010000
1005002110100010--0?00110?20
1005002000101010540?00110020
```

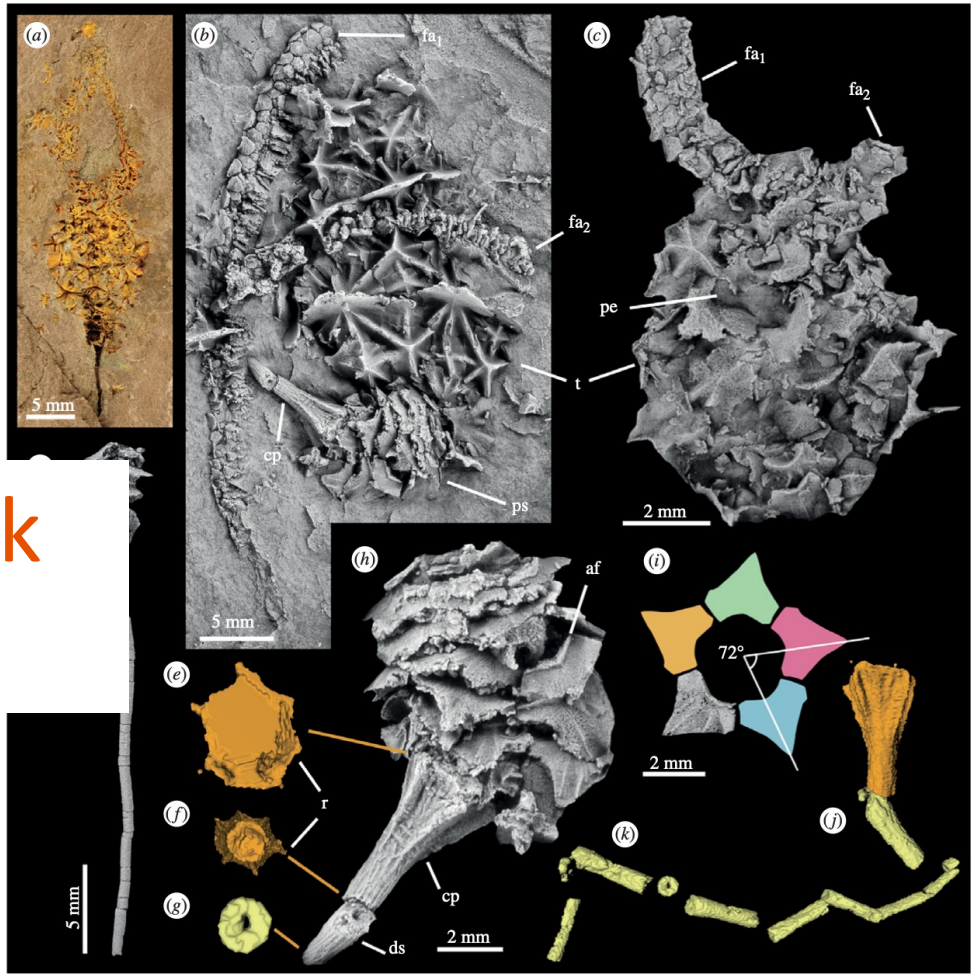


Cambrian stalked echinoderms show unexpected plasticity of arm construction
Zamora & Smith. 2012 Proc B

```
0015100 10?00-100--0000000000
0005000 10?200100--0010010000
0025000 10?200100--0?10010000
00?5?0010?200100?-0???010110
```

Can you draw the Q-matrix for an Mk model for this data set?

```
01050?010-210000?501??010110
0002000100210100 3-1110010110
0002000100211001441121011111
000201111-210010?-??11011121
?103?0?11?1001104-0000010000
1005002110100010--0?00110?20
1005002000101010540?00110020
```



Cambrian stalked echinoderms show unexpected plasticity of arm construction
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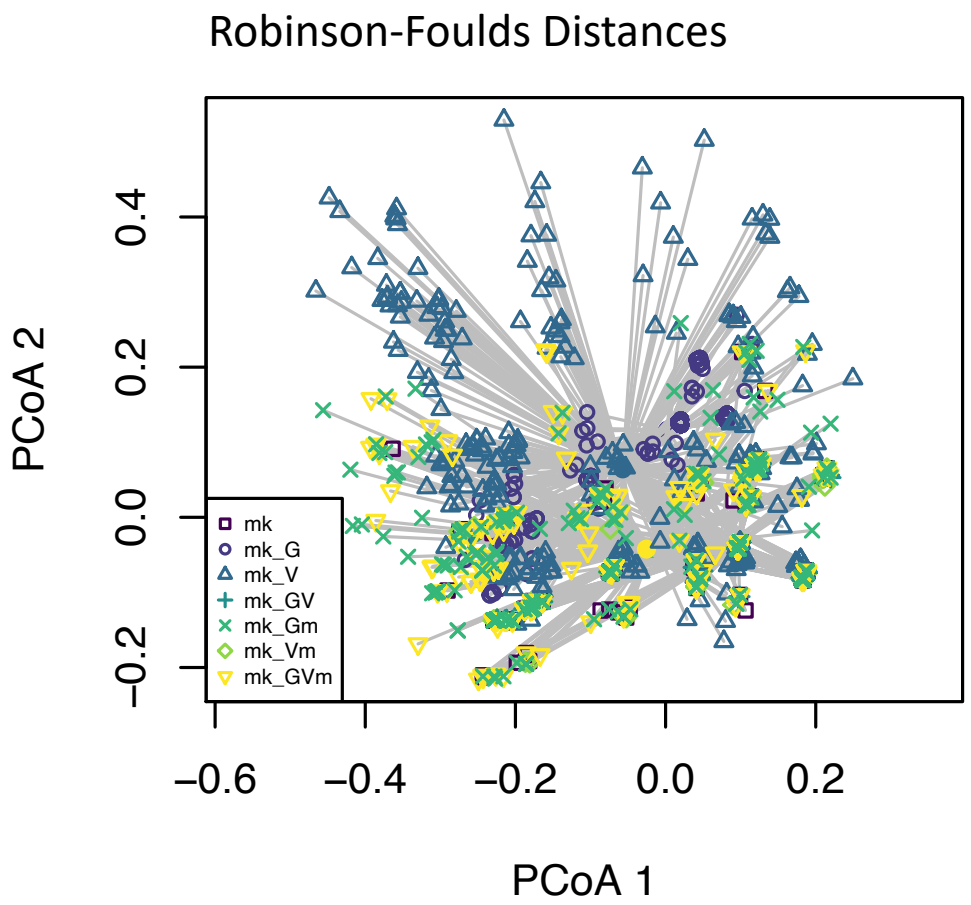
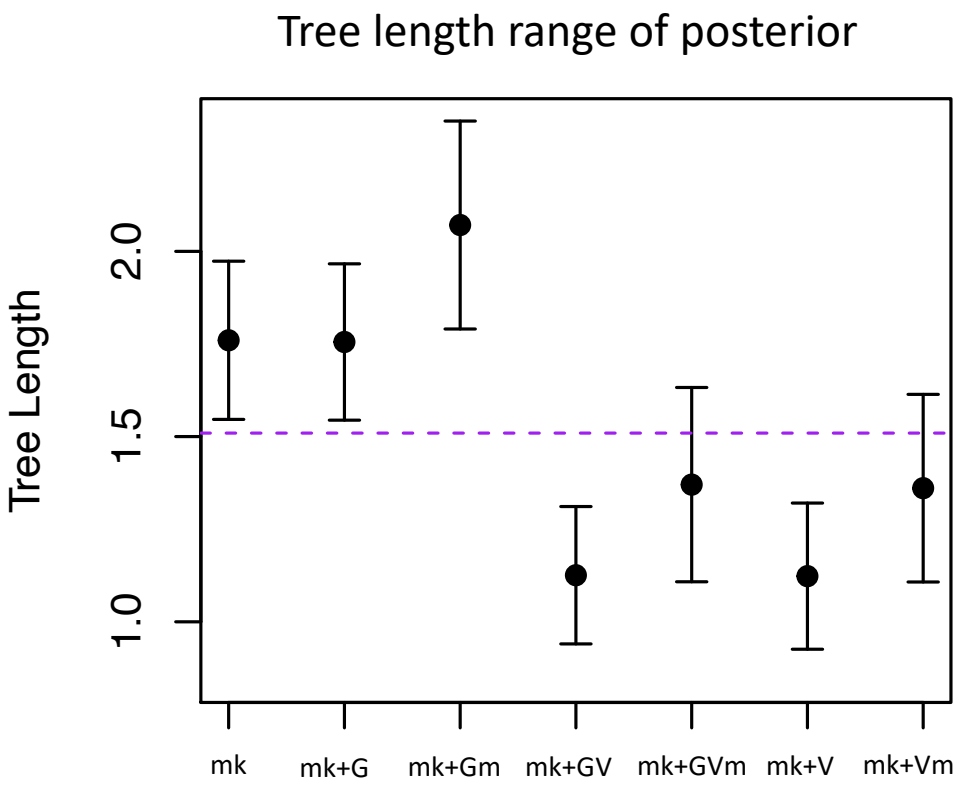
Exercise

Run an MCMC inference using **two** "versions" of the Mk model

Does changing the substitution model really matter for empirical data?

Impacts of substitution model on inferred parameters



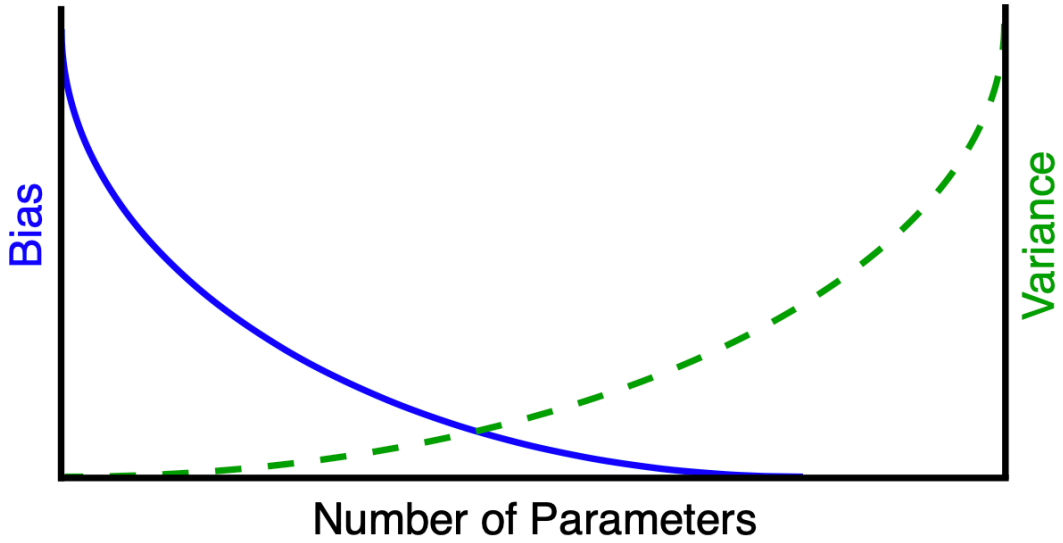


(Mulvey et al in prep.)

What does a good model look like?

To do statistical inference we need a model
What model should that be?
Our goal should be to have a model that is **complex enough** to capture "important" variation in the data, but **not be more complex** than it needs to be

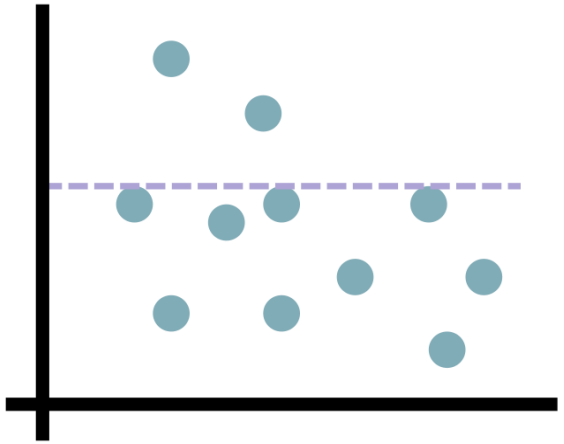
Too simple, misinterpreting the data



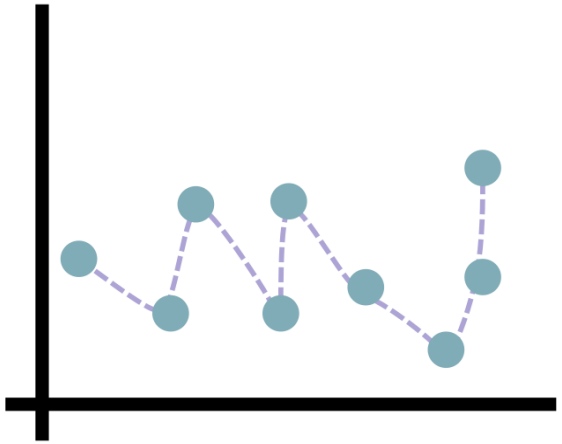
Too complicated, not enough information

What does a good model look like?

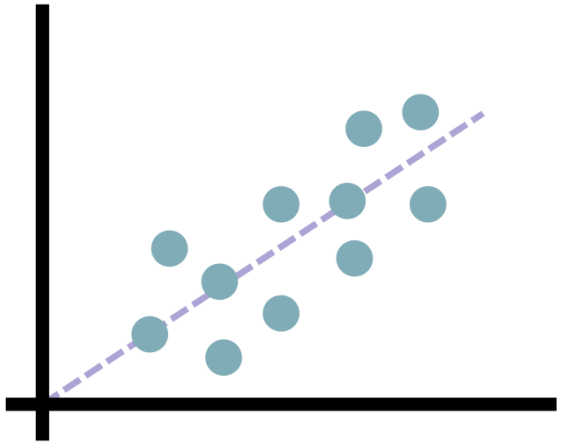
To do statistical inference we need a model
What model should that be?
Our goal should be to have a model that is **complex enough** to capture "important" variation in the data, but **not be more complex** than it needs to be



Underfitting



Overfitting



Proper fit

Model selections vs model adequacy

Model Selection and Testing

General Introduction to Model selection
Comparing relative model fit with Bayes factors

Model selection of common substitution models for one locus
Comparing relative model fit with Bayes factors

Model selection of partition models
Comparing relative model fit with Bayes factors

Model averaging of substitution models
Reversible-jump MCMC over substitution models

Introduction to Posterior Prediction
Assessing the fit of Normal distributions to trait data

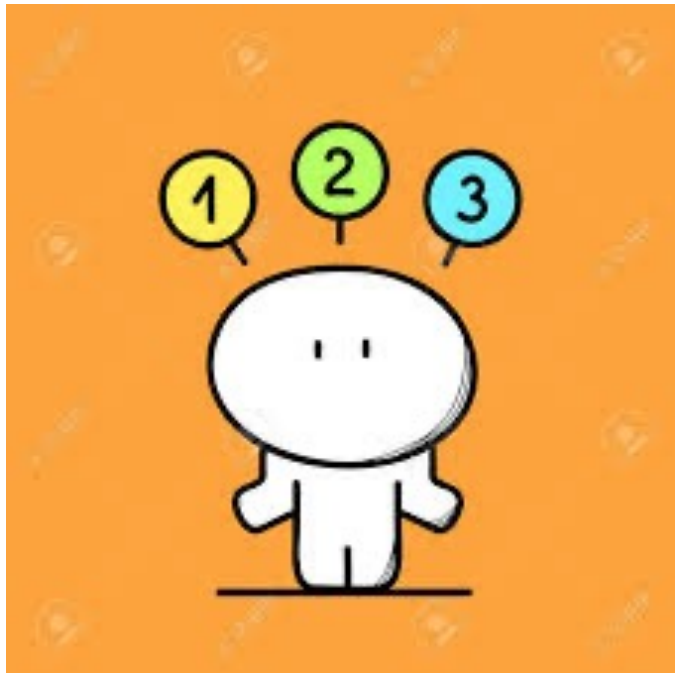
Assessing Phylogenetic Reliability Using RevBayes and P^3
Model adequacy testing using posterior prediction (Data Version).

Assessing Phylogenetic Reliability Using RevBayes and P^3
Model adequacy testing using posterior prediction (Inference Version).

Revbayes tutorials

How to choose which model to use for morphological data?

Guess
What other people have done
AIC values
BIC Values
Bayes factors :



Bayes Theorem

$$P(\text{parameters} \mid \text{data, model}) = \frac{P(\text{data} \mid \text{parameters, model})\, P(\text{parameters} \mid \text{model})}{P(\text{data} \mid \text{model})}$$

Posterior

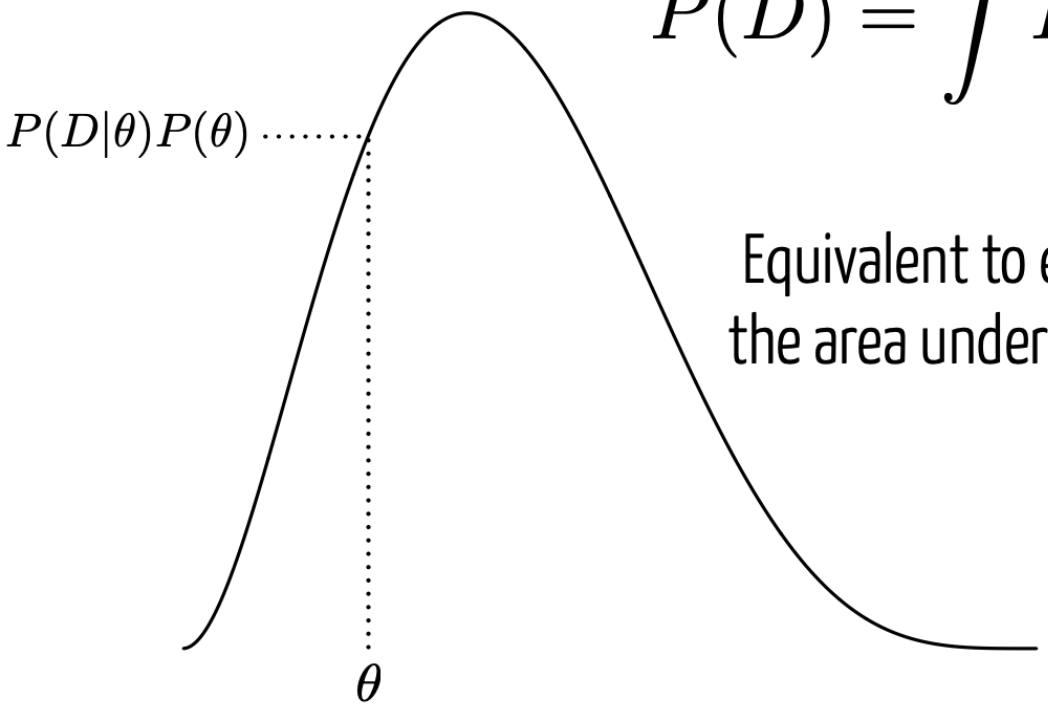
Likelihood

Priors

Marginal probability

What is the marginal likelihood.....

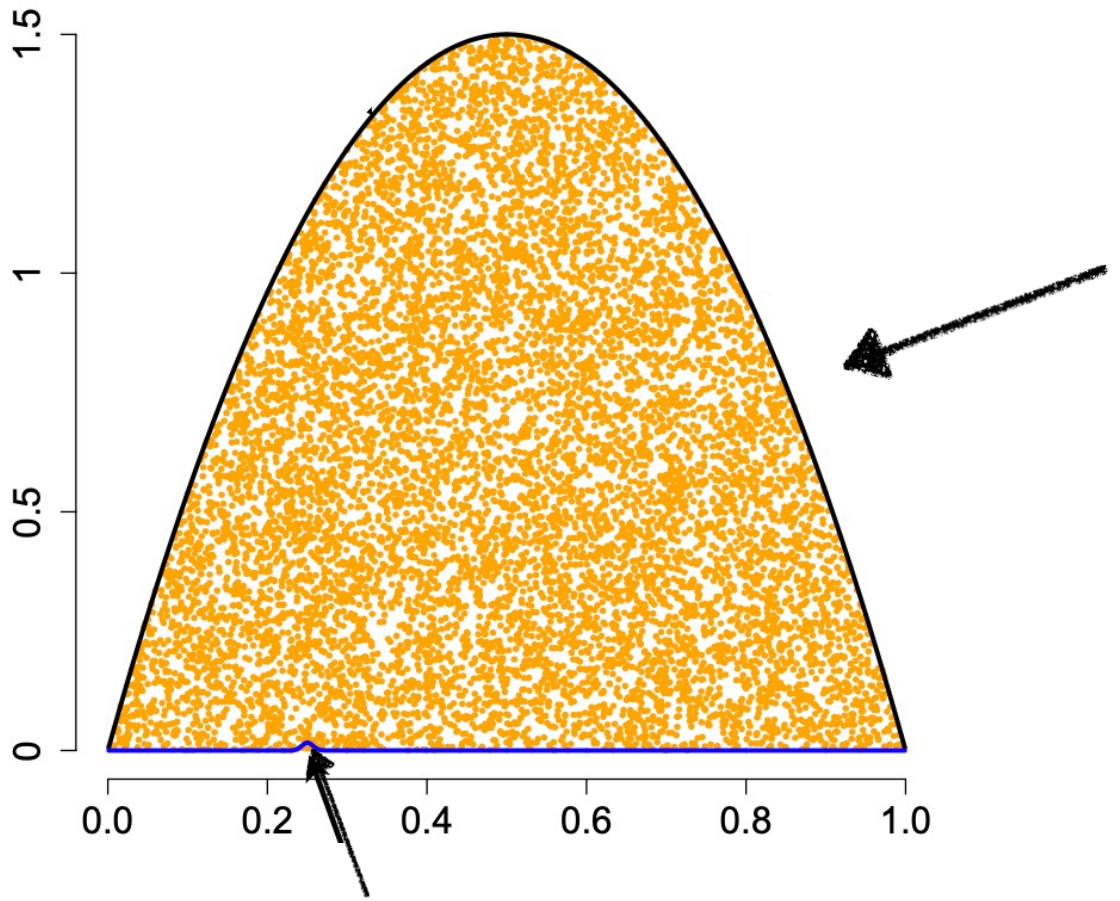
$$P(D) = \int P(D|\theta)P(\theta)$$



Equivalent to estimating the area under this curve.

Paul Lewis - Workshop on Molecular Evolution 2016

How can we estimate the marginal likelihood

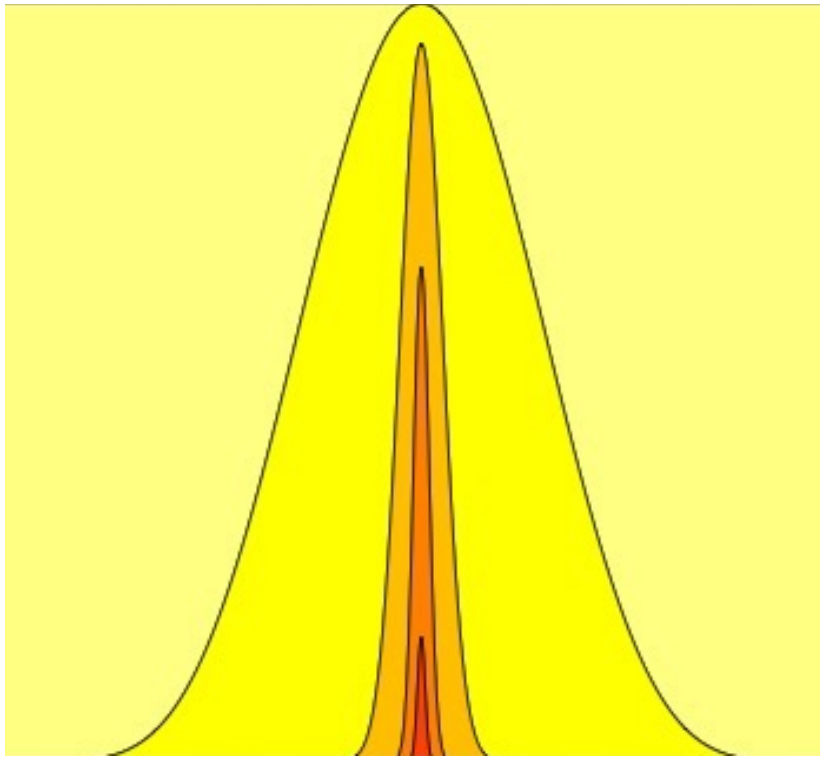


Prior distribution

Marginal likelihood

Paul Lewis - Workshop on Molecular Evolution 2016

Stepping Stone



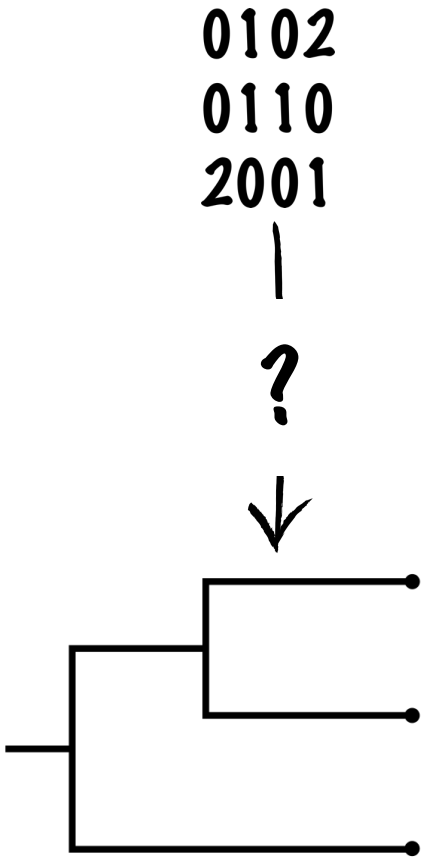
Keep estimating smaller and smaller sections until you get down to the marginal likelihood

Model selection doesn’t work well for morphological data. This is because the Mk model doesn’t have any free parameters but a partitioned model will always return a higher likelihood, so its not possible to distinguish between unpartitioned and portioned models.

Model selection vs. Model adequacy

Take a bunch of different models and test which is the *best*

Gives the **relative** fit



Assess whether a model is capturing the evolutionary dynamics that generated the data

Gives the **absolute** fit

Model Adequacy

We know that none of our models are really true. Can we be sure that the chosen model captures the salient features of the evolutionary process and provides reliable inferences?

Could the model and priors plausibly have given rise to the data?

Allows us to ask whether **any** of our models are doing a good job describing the evolutionary processes that produced our data.

Posterior Predictive Simulations

Empirical Data	
taxa 1	0 1 0 1 2 1
taxa 2	1 2 1 0 1 0
taxa 3	0 0 1 0 0 1
taxa 4	1 1 0 1 0 1

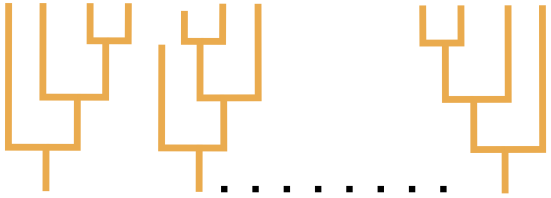
Höhna et al 2017

Posterior Predictive Simulations

Empirical Data	
taxa 1	0 1 0 1 2 1
taxa 2	1 2 1 0 1 0
taxa 3	0 0 1 0 0 1
taxa 4	1 1 0 1 0 1

1)

Standard MCMC inference while sampling from the posterior



Posterior Predictive Simulations

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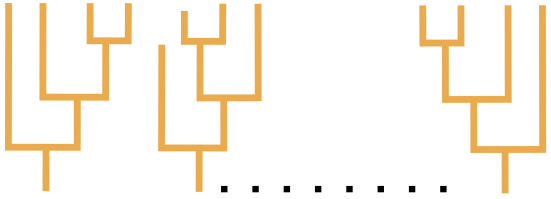

2) Using the information sampled in 1) generate new data sets

Simulated Data 1

taxa 1	1 0 0 1 2 1
taxa 2	1 2 1 0 2 0
taxa 3	0 1 0 1 1 1
taxa 4	1 0 0 1 0 1

Simulated Data 2

taxa 1	1 1 0 1 2 1
taxa 2	1 1 1 0 1 0
taxa 3	0 1 1 1 0 1
taxa 4	1 2 0 1 0 1

. . . .

Simulated Data n

taxa 1	1 1 0 1 2 1
taxa 2	1 1 1 0 1 0
taxa 3	0 1 1 1 0 1
taxa 4	1 2 0 1 0 1

Posterior Predictive Simulations

Empirical Data

taxa 1	0 1 0 1 2 1
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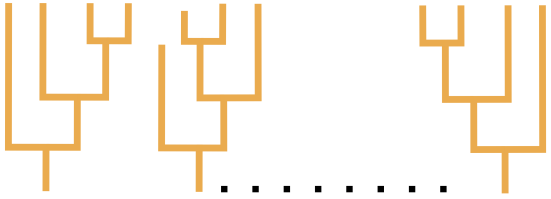
1) Standard MCMC inference while sampling from the posterior



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taxa 4	1 0 0 1 0 1

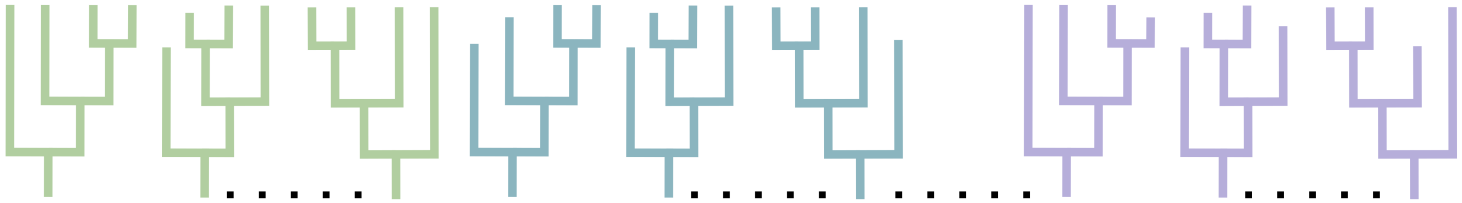
Simulated Data 2

taxa 1	1 1 0 1 2 1
taxa 2	1 1 1 0 1 0
taxa 3	0 1 1 1 0 1
taxa 4	1 2 0 1 0 1

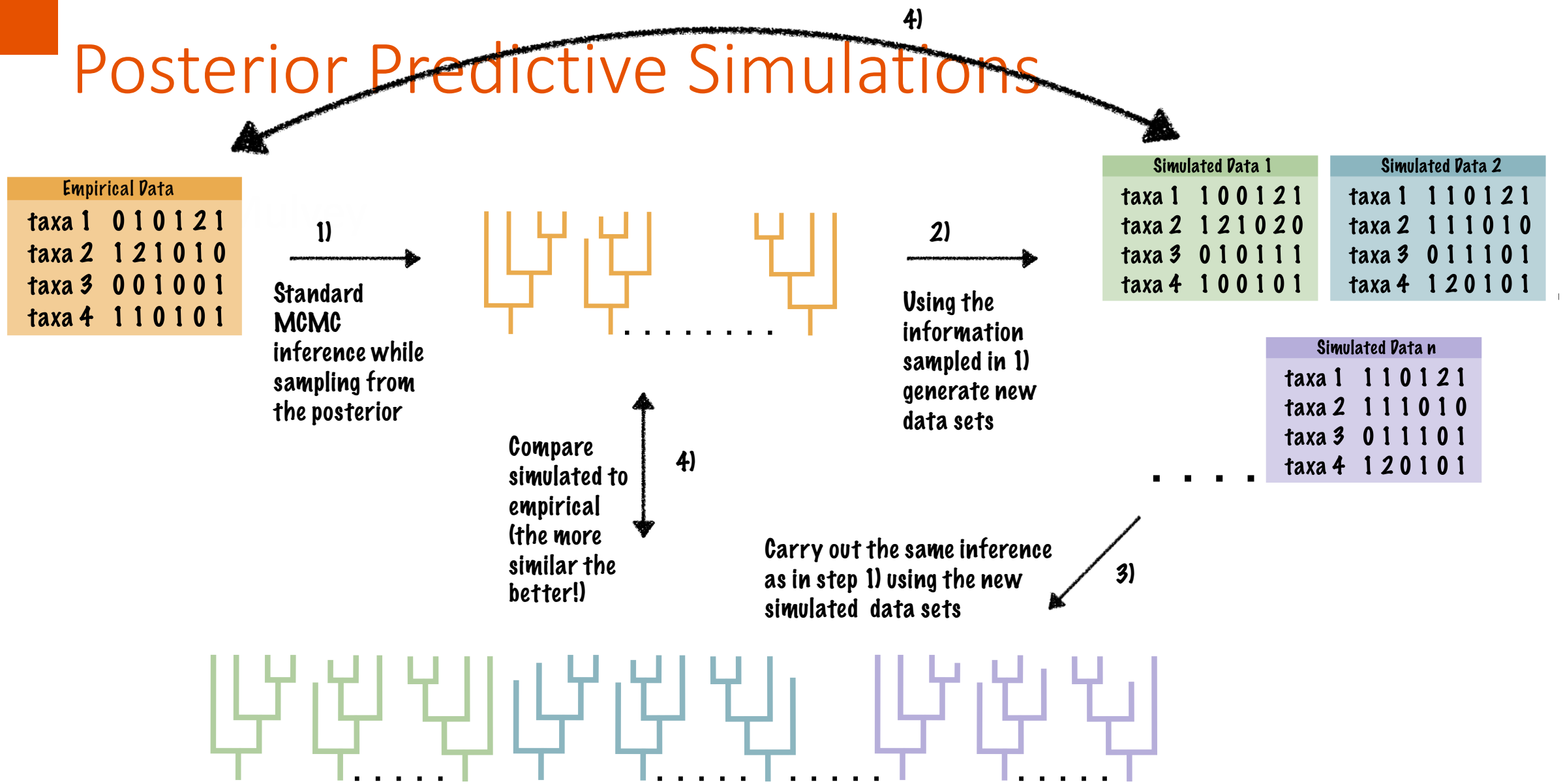
Simulated Data n

taxa 1	1 1 0 1 2 1
taxa 2	1 1 1 0 1 0
taxa 3	0 1 1 1 0 1
taxa 4	1 2 0 1 0 1

3) Carry out the same inference as in step 1) using the new simulated data sets



Posterior Predictive Simulations



Test Statistics

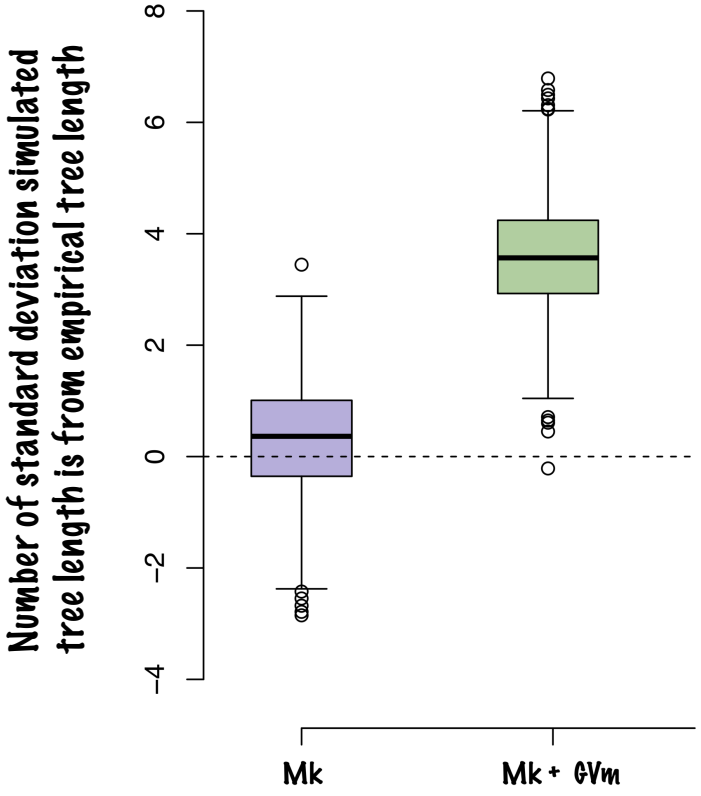
How can we compare trees and morphological matrices?

Need to get test statistics that compare the difference.

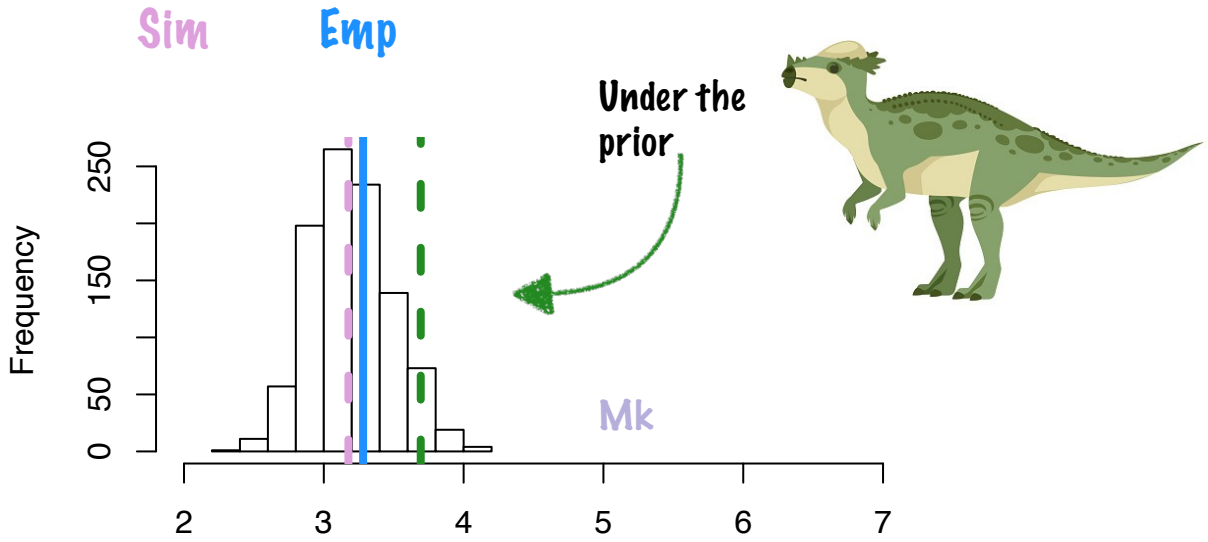
More work has been done for molecular data – easier to compare.

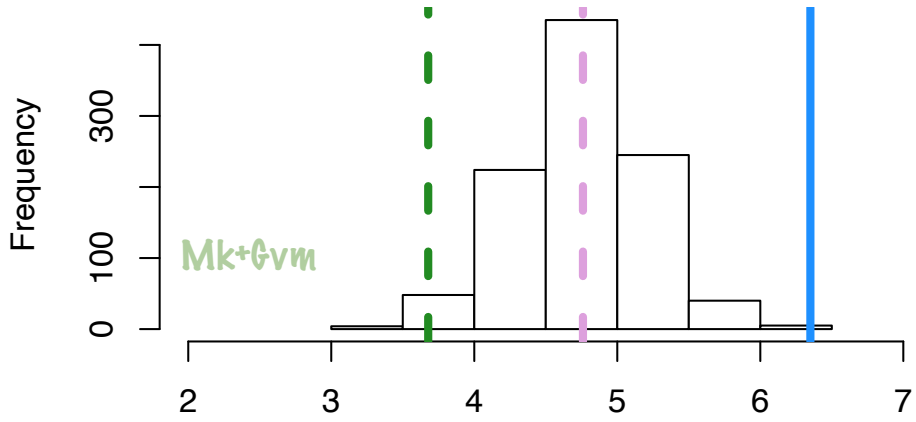
To compare simulations to empirical data we use effect sizes.

Test Statistics

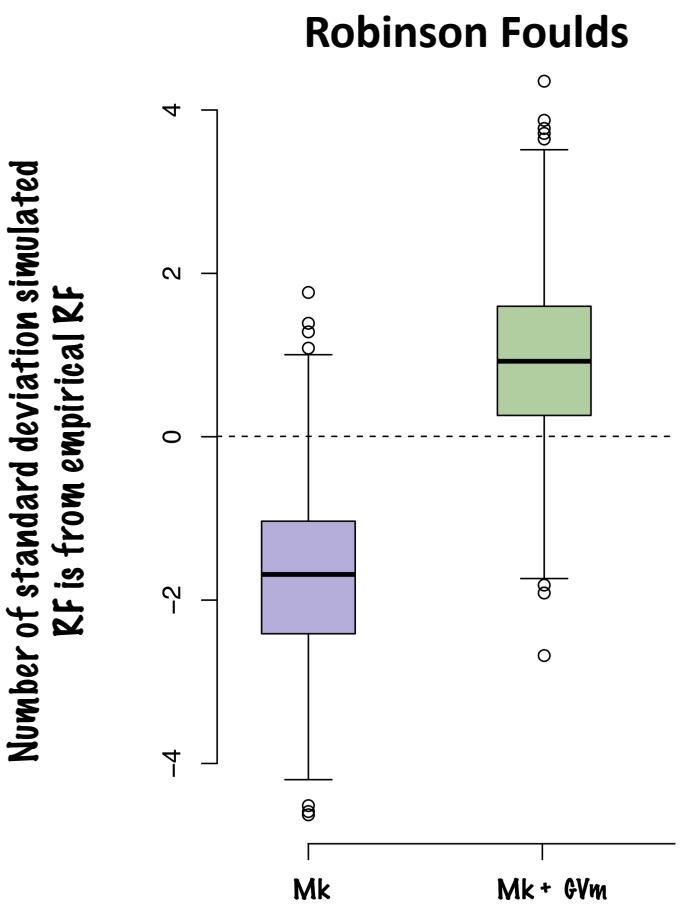


over estimated using the more complex model

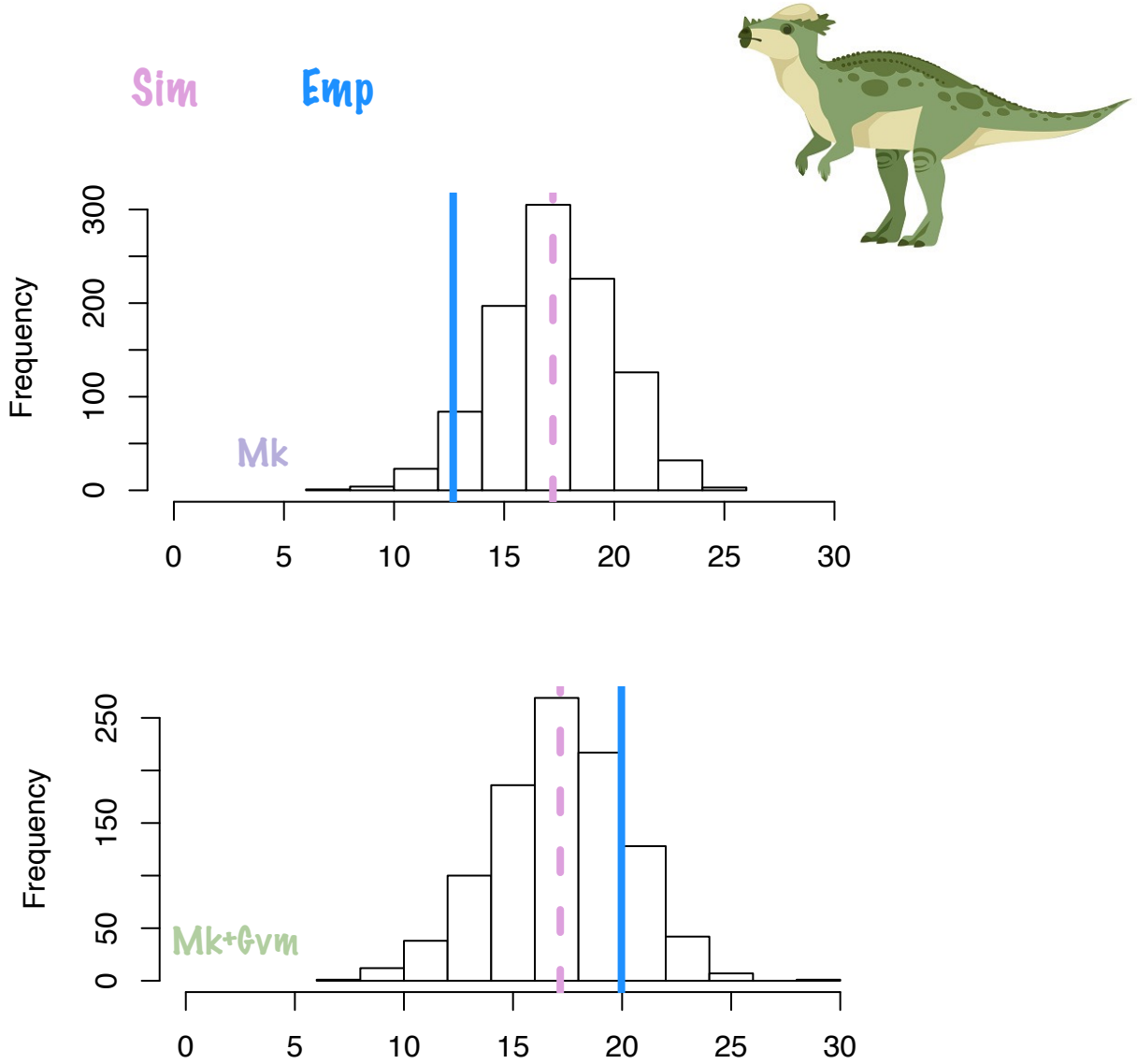




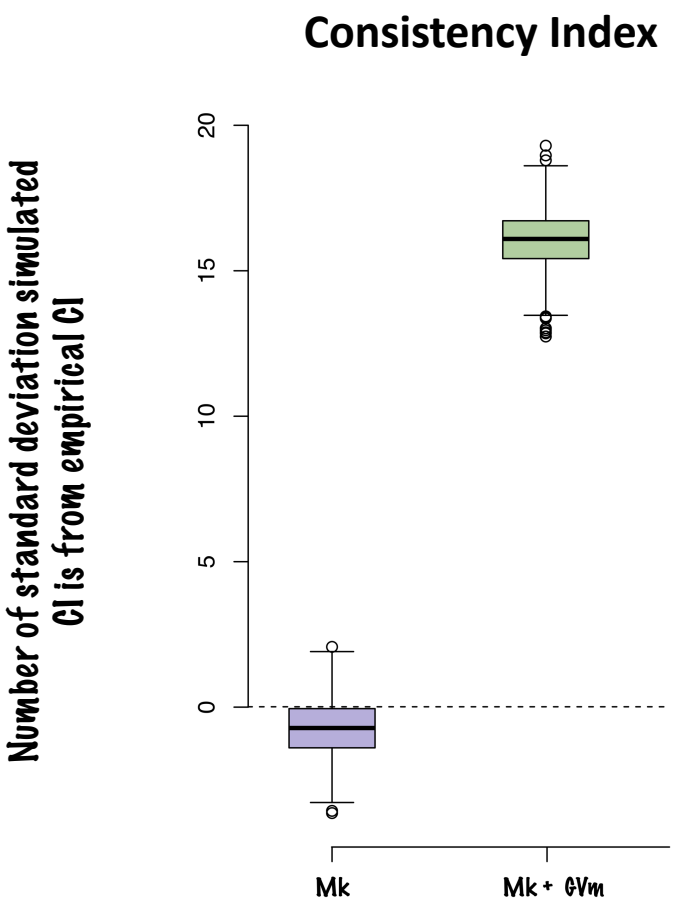
Test Statistics



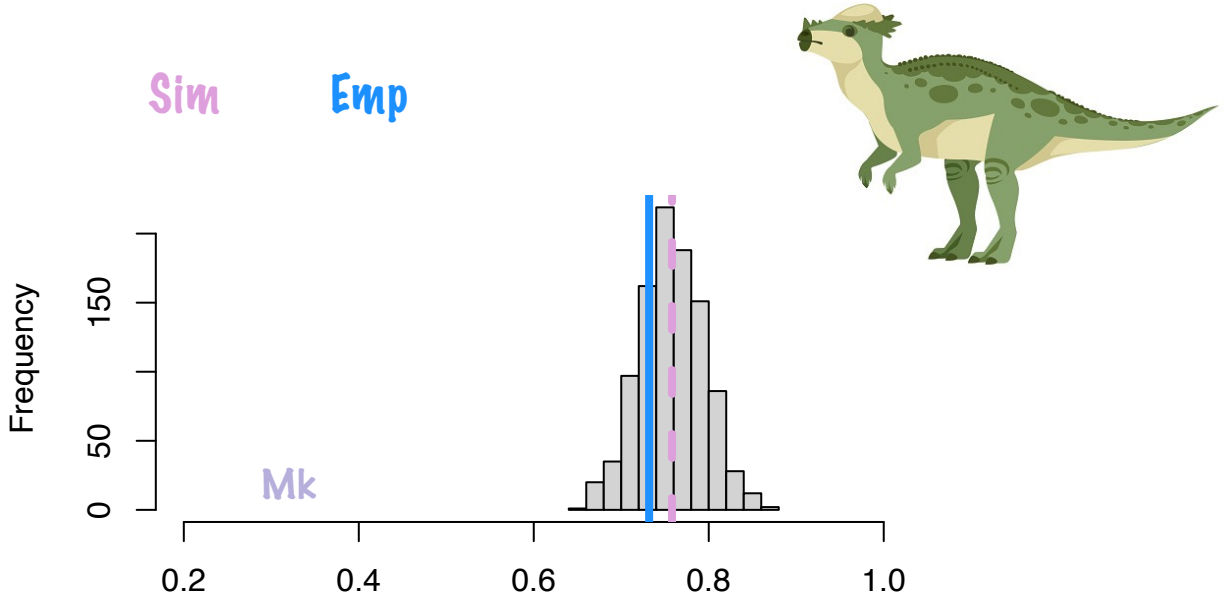
Both models produced similar RF results

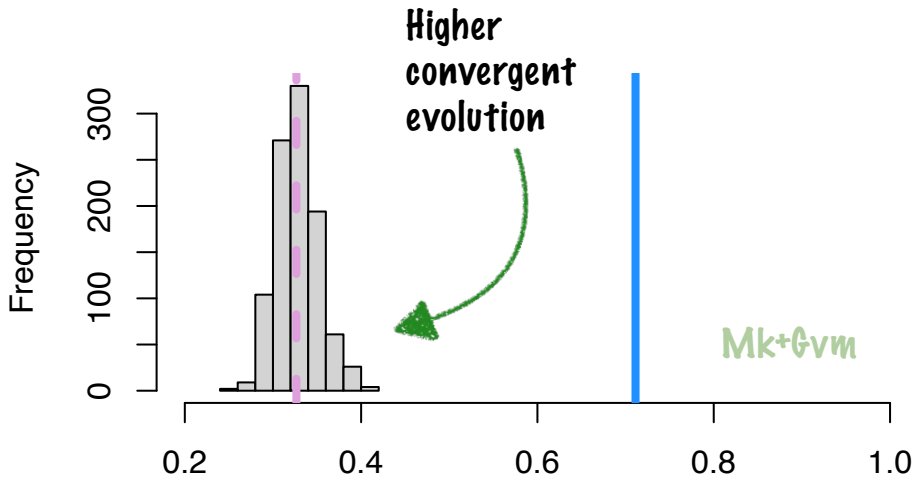


Test Statistics



The more complex **over** estimated convergent evolution





More test statistics

Tree length
Robinson Foulds
Consistency Index
Retention Index
Hamming distances
Multiple distance metrics

Exercise

Check if either of the two models you chose for exercise 1 fit your data using a model adequacy approach