Morphological models and how to choose them

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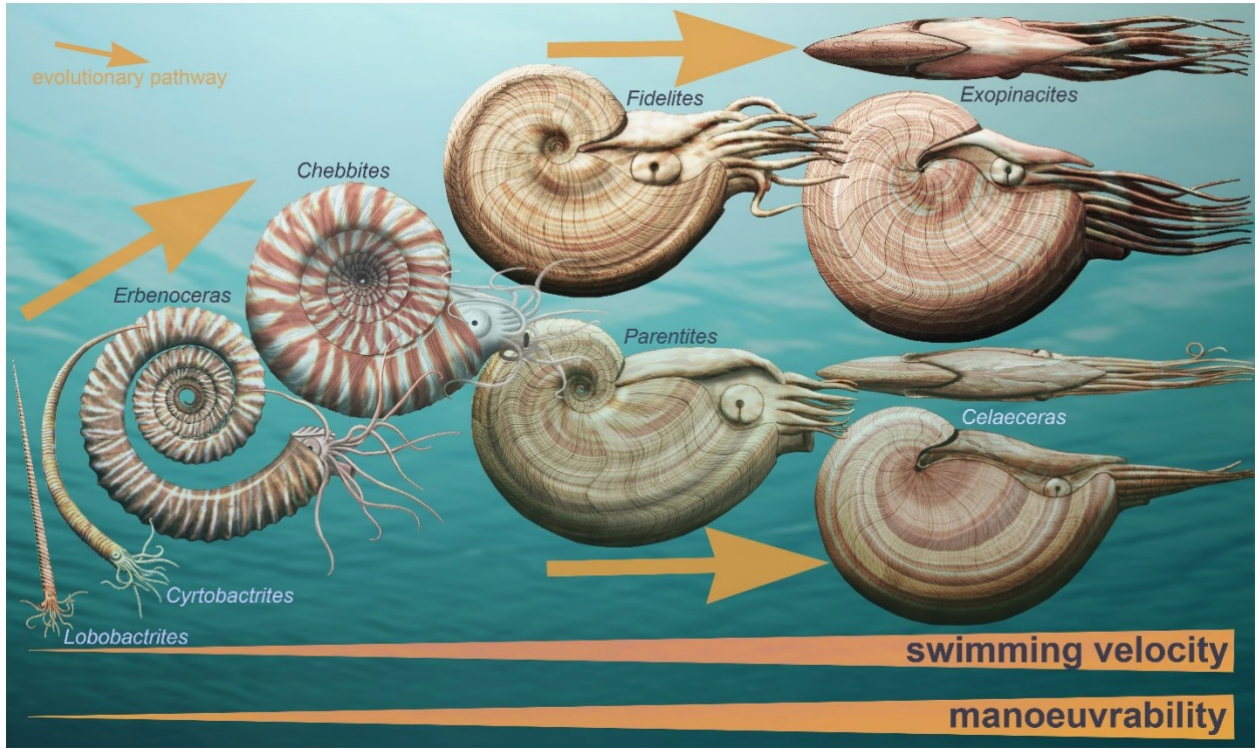
Analytical Paleobiology Workshop

13.08.2024

Morphological data

Morphological data was the original type of information used in phylogenetic analysis

Fossils can be used to provide time calibrations, helps extant phylogeny, allows us to understand evolution through time



Monnet et al 2011 BMC Ecology and Evolution

Morphological character data

Discrete Characters: Morphological data often consist of discrete characters, such as the presence or absence of certain traits, or more complex multistate traits (e.g., number of limbs, type of leaf, presence of a particular bone structure)

Continuous Characters: Some morphological data can be continuous, such as measurements of body size, length of bones, or other quantitative traits

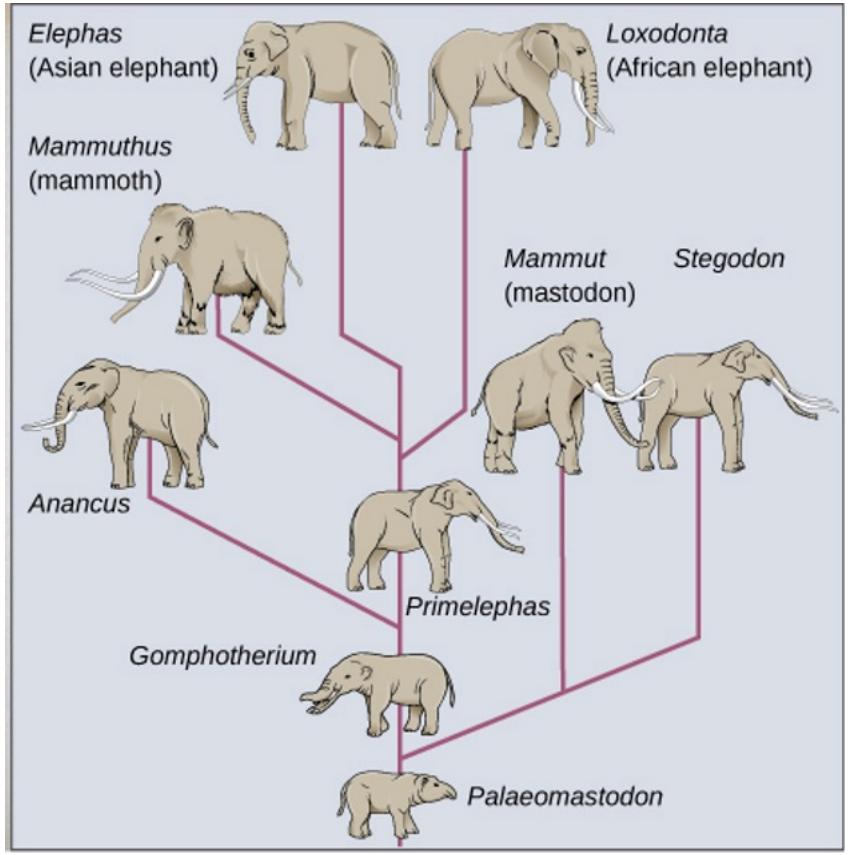


Image source

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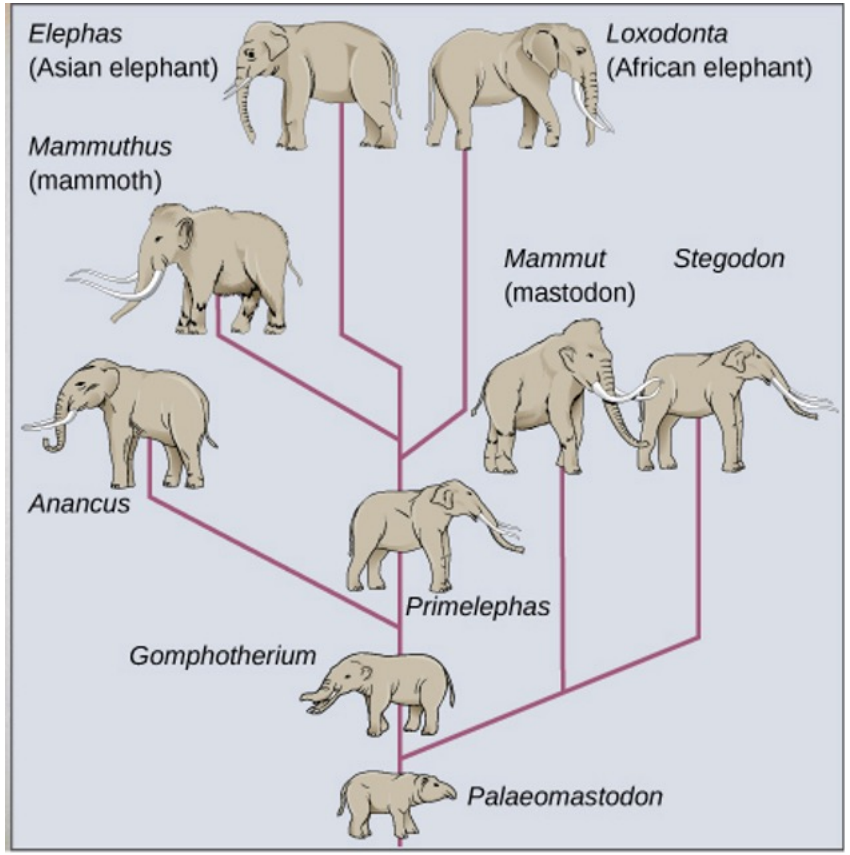

Image from https://www.zoologytalks.com/

Trait 1 Trait 28

Taxa 1 0015100l0?00-100--0000000000

0005000l0?200100--0010010000

0025000l0?200100--0?10010000

00?5?00l0?200100?-0???010110

0015000l012010004301000l1111

0015000l012010l04401110l1111

??050????2010000440?110l1111

01050?010-210000?501??010110

0002000l002101003-1110010110

0002000l00211001441121011111

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?103?0?11?1001104-0000010000

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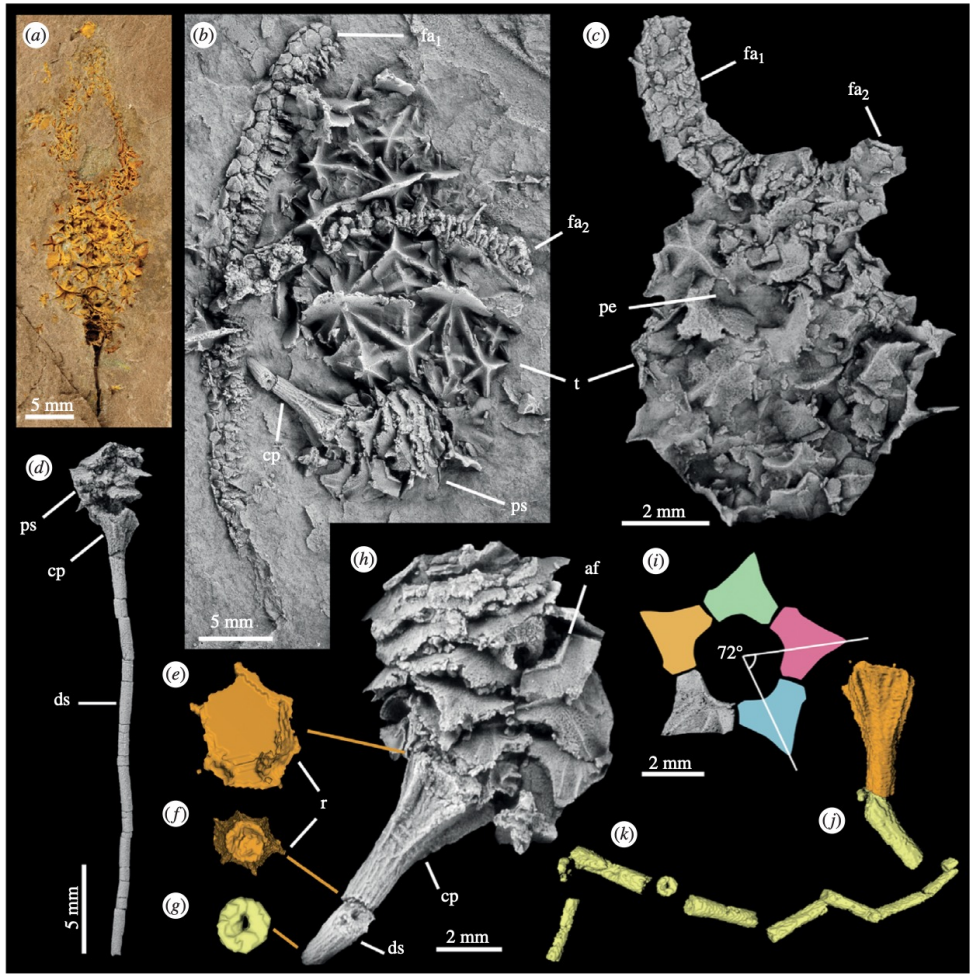
Taxa 14 1005002000101010540?00110020



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

Appendage branching pattern

Cover plate arrangement

Presence Absence

```
0015100l0?00-100--0000000000
0005000l0?200100--0010010000
0025000l0?200100--0?10010000
00?5?00l0?200100?-0???010110
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0015000l012010l044011101l111
??050????2?0l000440?11011111
01050?0l0-2l0000?501??010110
0002000l002l01003-1110010110
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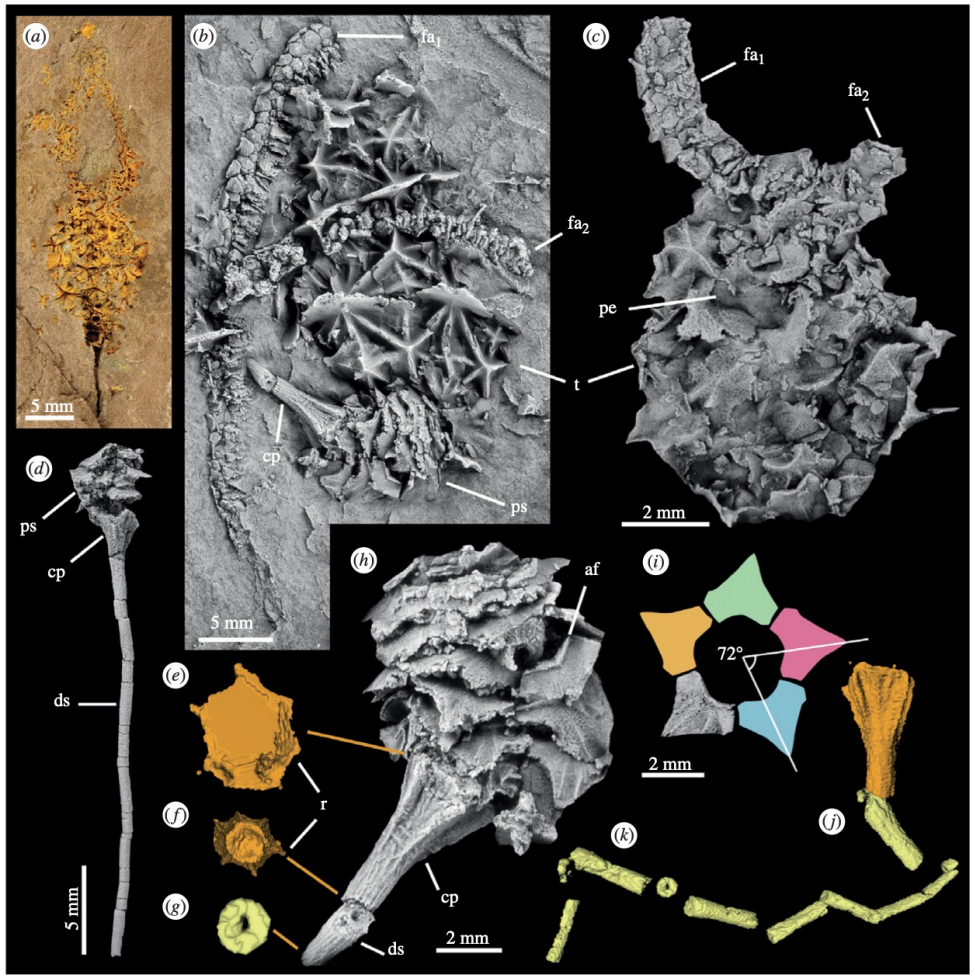
```



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Discrete character data

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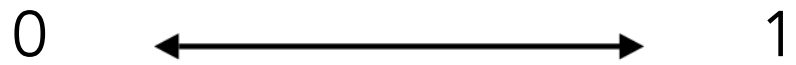
Polymorphisms	0/1/2	Used when there are variations in a traits within species
Ambiguous	0/1/2	Used when it is not clear which character trait is present in the taxon

How do we model morphological evolution?

Mk Model

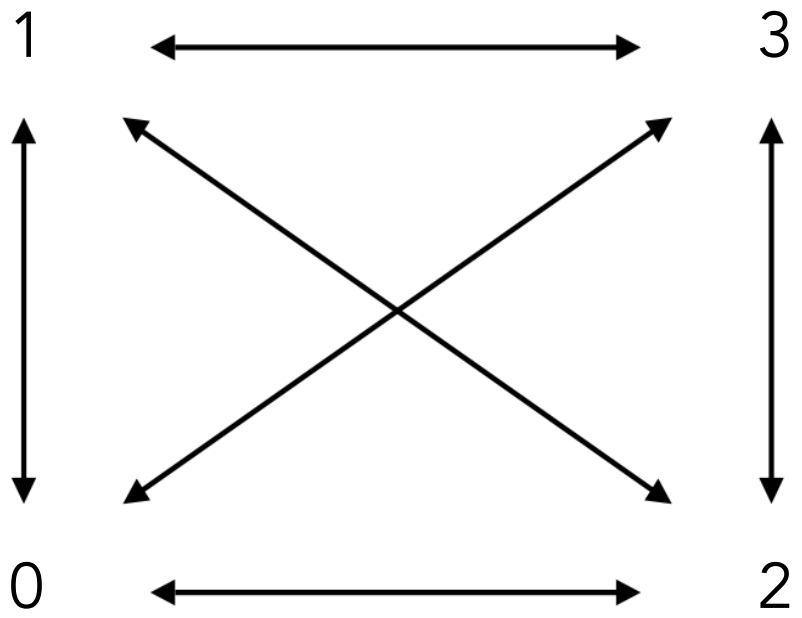
Assumes **equal transition probabilities** between states



$$Q = \begin{bmatrix} -\mu_0 & \mu_{01} \\ \mu_{10} & -\mu_1 \end{bmatrix}$$

Mk Model

K can be any number of states

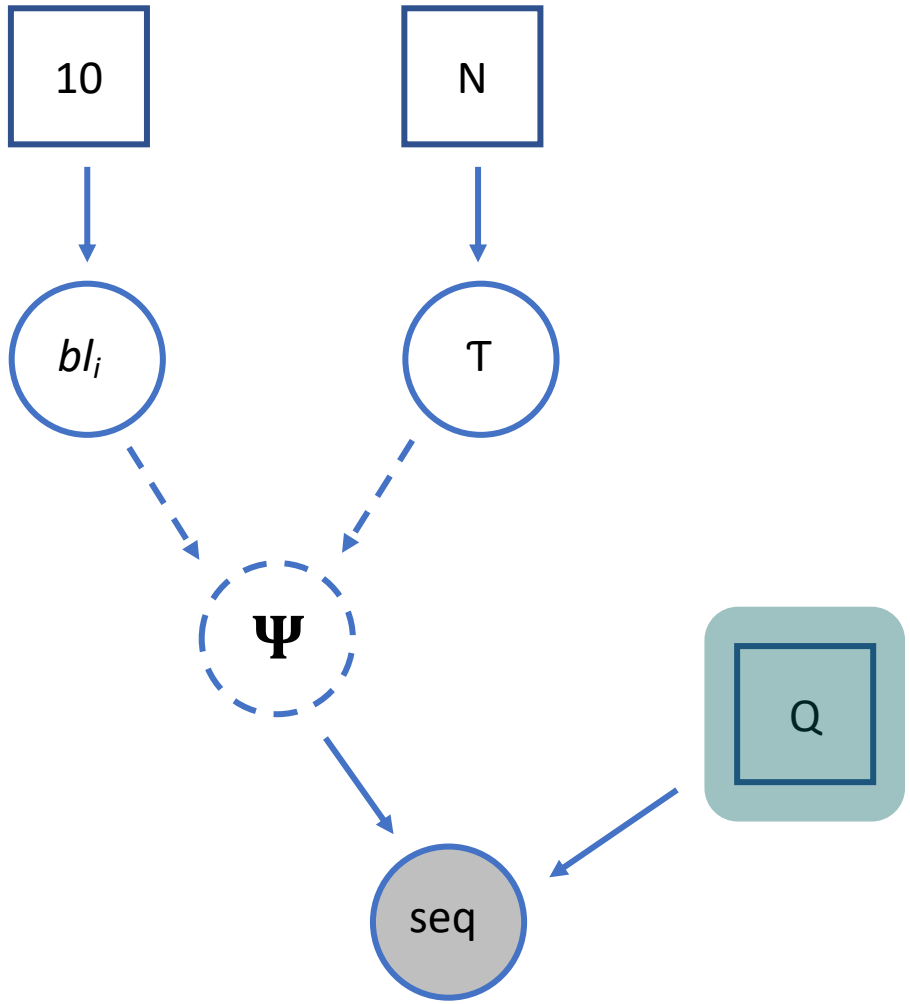


$$Q = \begin{pmatrix} -\mu_0 & \mu_{01} & \mu_{02} & \mu_{03} \\ \mu_{10} & -\mu_1 & \mu_{12} & \mu_{13} \\ \mu_{20} & \mu_{21} & -\mu_2 & \mu_{23} \\ \mu_{30} & \mu_{31} & \mu_{32} & -\mu_3 \end{pmatrix},$$

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

Mk Model

Graphical model



MkV model

What is one characteristic of morphological data that is extremely different to molecular

though there are plenty……

```
00151O010?00-100--0000000000
000500010?200100--0010010000
002500010?200100--0?10010000
00?5?0010?200100?-0???010110
00150001012010004301000111111
00150001012010104401110111111
??050?????201000440?11011111
01050?010-210000?501??010110
00020001002101003-1110010110
000200010021100144112101111
000201111-210010?-??11011121
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10050021101000l0--0?00110?20
100500200010101O540?00110020
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All varying characters

```
00151O010?00-100--0000000000
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002500010?200100--0?10010000
00?5?0010?200100?-0???010110
00150001012010004301000111111
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01050?010-210000?501??010110
000200010021010003-1110010110
0002000100211001441121011111
000201111-210010?-??11011121
?103?0?11?1001104-0000010000
1005002110100010--0?00110?20
10050020001010105 40?00110020
```

MkV model

Corrects for **ascertainment bias**

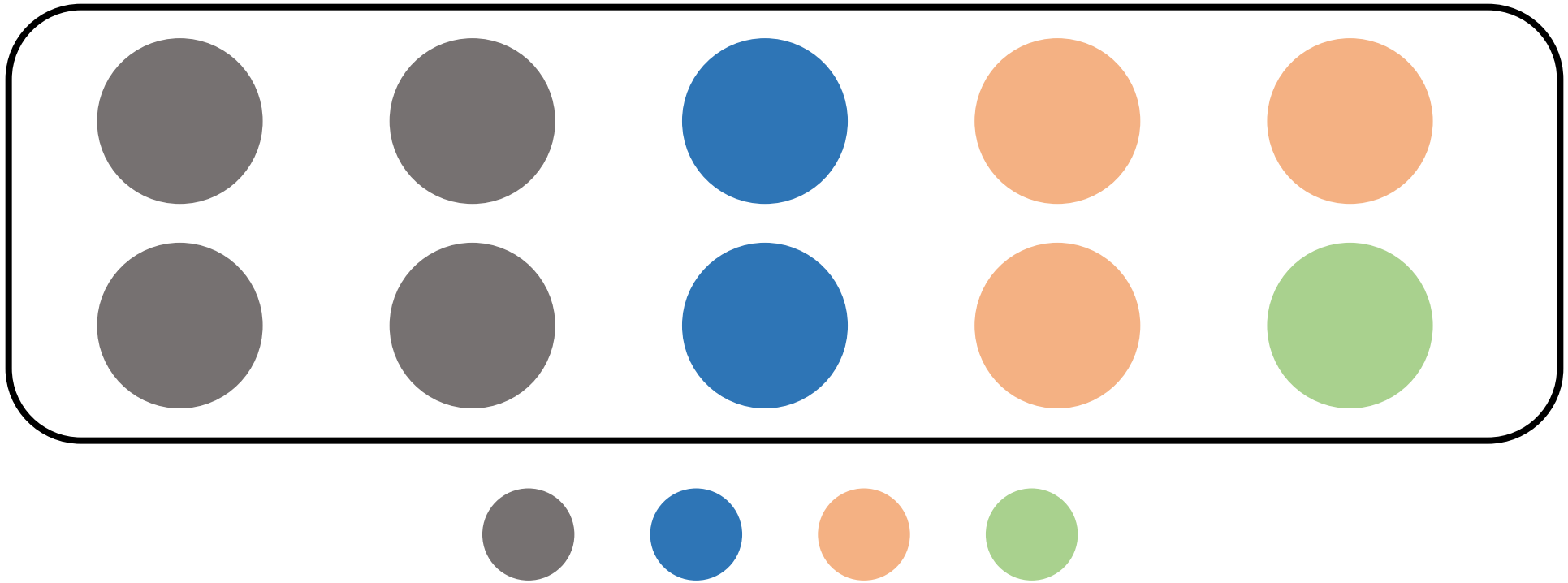
Failing to account for this can lead to **overestimations in branch lengths** and which can further lead to errors in topology!

Condition the likelihood on there only being varying site

$$\Pr(D \mid V) = \frac{\Pr(D, V)}{\Pr(V)}$$

MkV model

What is the probability for picking a certain colour ball?



$$1 = 0.4 + 0.2 + 0.3 + 0.1$$

Adapted from Paul Lewis PhyloSeminar

MkV model

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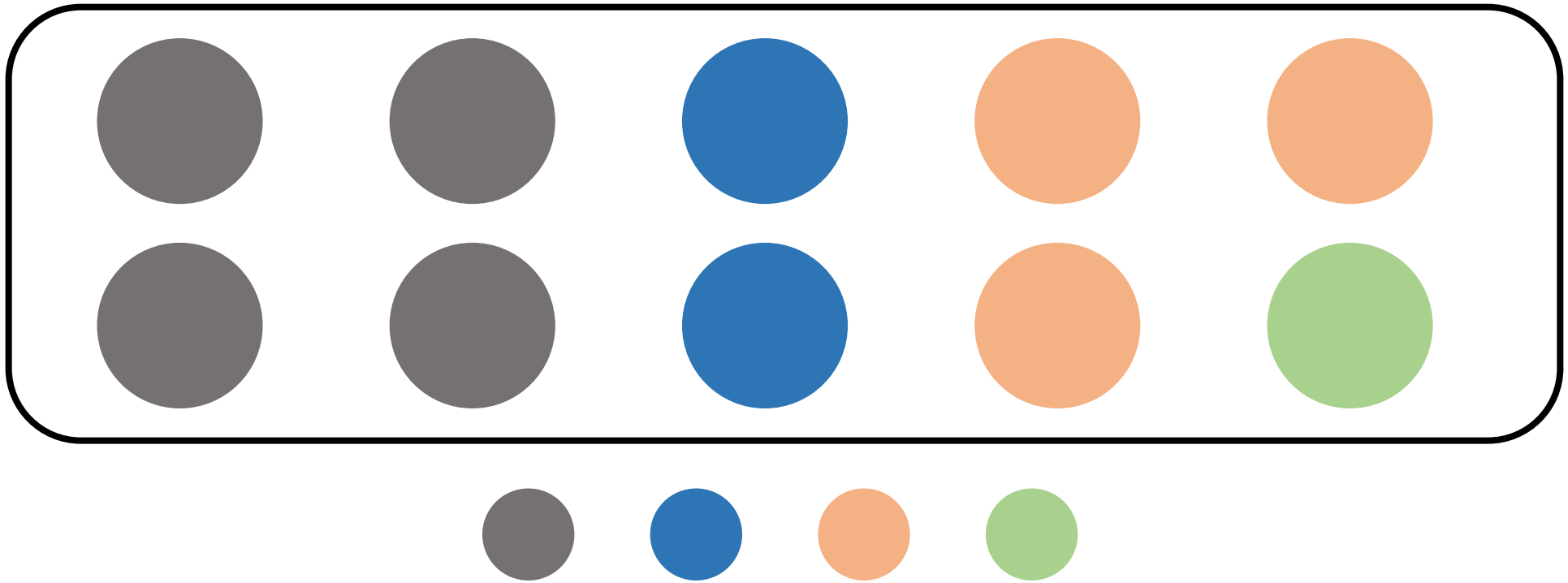

$$1 = 0.4 + 0.2 + 0.3 + 0.1$$

Probability of choosing an orange ball = 0.3

Adapted from Paul Lewis PhyloSeminar

MkV model

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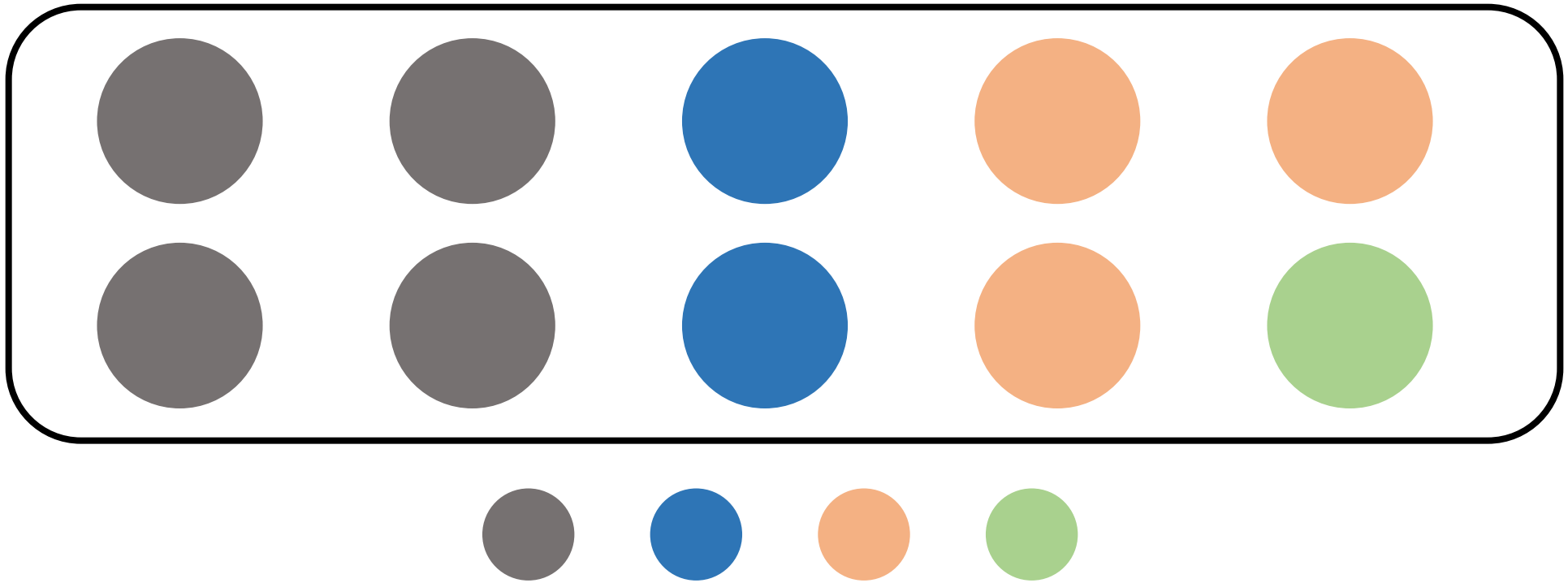

$$1 = 0.4 + 0.2 + 0.3 + 0.1$$

Probability of choosing an orange ball = 0.3

Probability of choosing an orange ball given it is not grey = 0.3/0.6 = 0.5

Adapted from Paul Lewis PhyloSeminar

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Corrects for **ascertainment bias**

Failing to account for this can lead to overestimations in branch lengths and which can further lead to errors in topology!

Probability of the data given character is variable

$$Pr(D \mid V) = \frac{Pr(D,V)}{Pr(V)}$$

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Probability that character is variable

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$$Pr(D \mid V) = \frac{Pr(D,V)}{Pr(V)}$$

Probability that character is variable

1 - Pr(character is constant)

This value, Pr(C) can be obtained using a **dummy character** having the same state for all internal nodes

MkV model

In RevBayes this is done internally and all non varying characters will be removed before the inference

In Beast you will see the dummy characters in the xml file produced from beauti

```
    <data
id="Zamora_Smith_part"
spec="Alignment"
dataType="standard">
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                            value="00250001O?200100--0?1001000012345" />
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                            value="00?5?001O?200100?-0???01011012345" />
                        <sequence id="seq_Ridersia" spec="Sequence" taxon="Ridersia" totalcount="6"
                            value="00150001012010004301000111112345" />
```

MkV model

	True Branch Length	Mk	Mkv
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)

Among-character rate variation

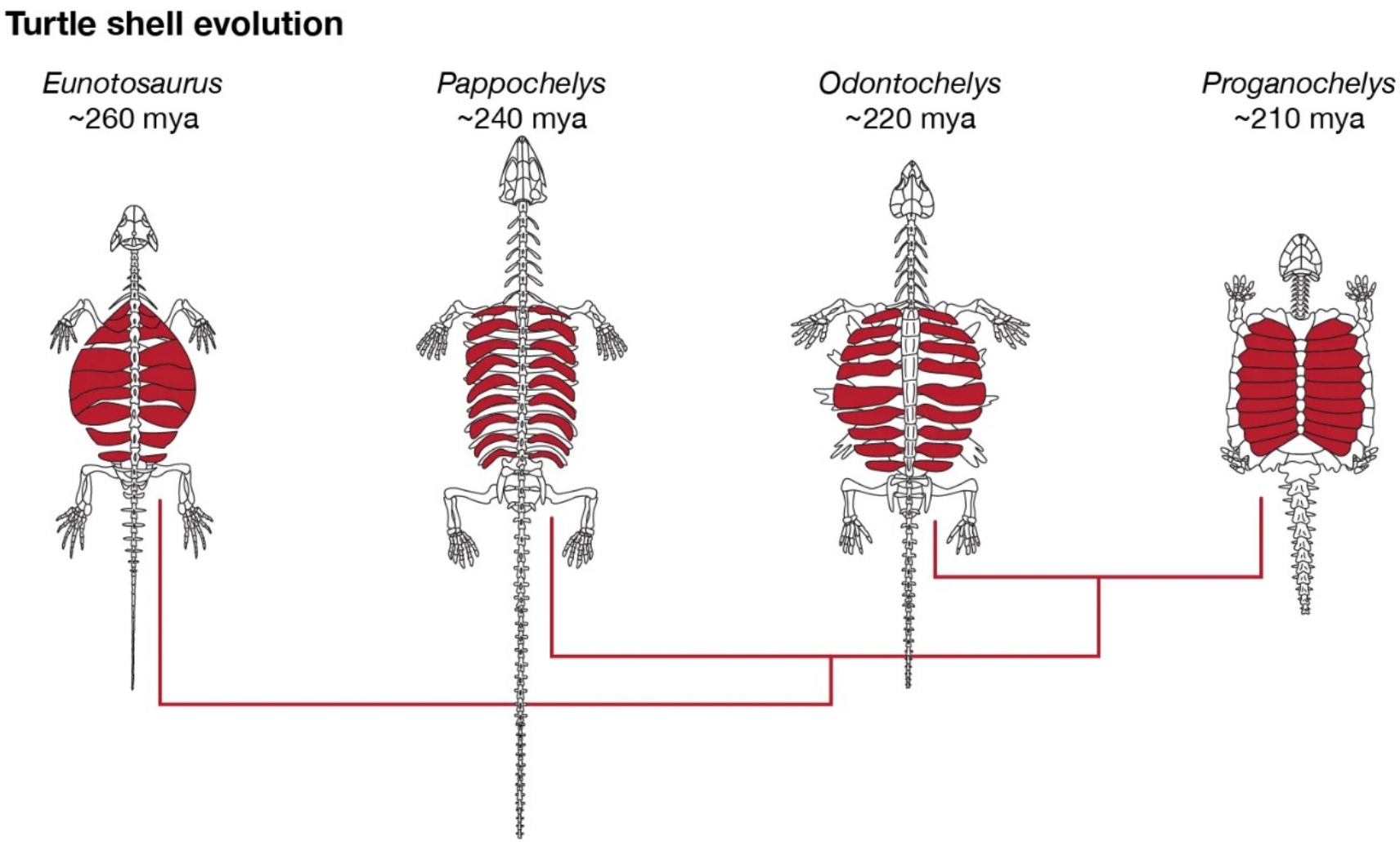

Image source

Among-character rate variation

	T1	T2
Taxa A	0	0
Taxa B	0	1
Taxa C	1	2

The transition rate will impact branch lengths

Slow rate of evolution

Fast rate of evolution

Relative to each other!

Among-character rate variation

What do we do?

	T1	T2
Taxa A	0	0
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Allow these traits to evolve at different rates:

- Specify which traits evolve fast
- Use a gamma model to account for rate heterogeneity

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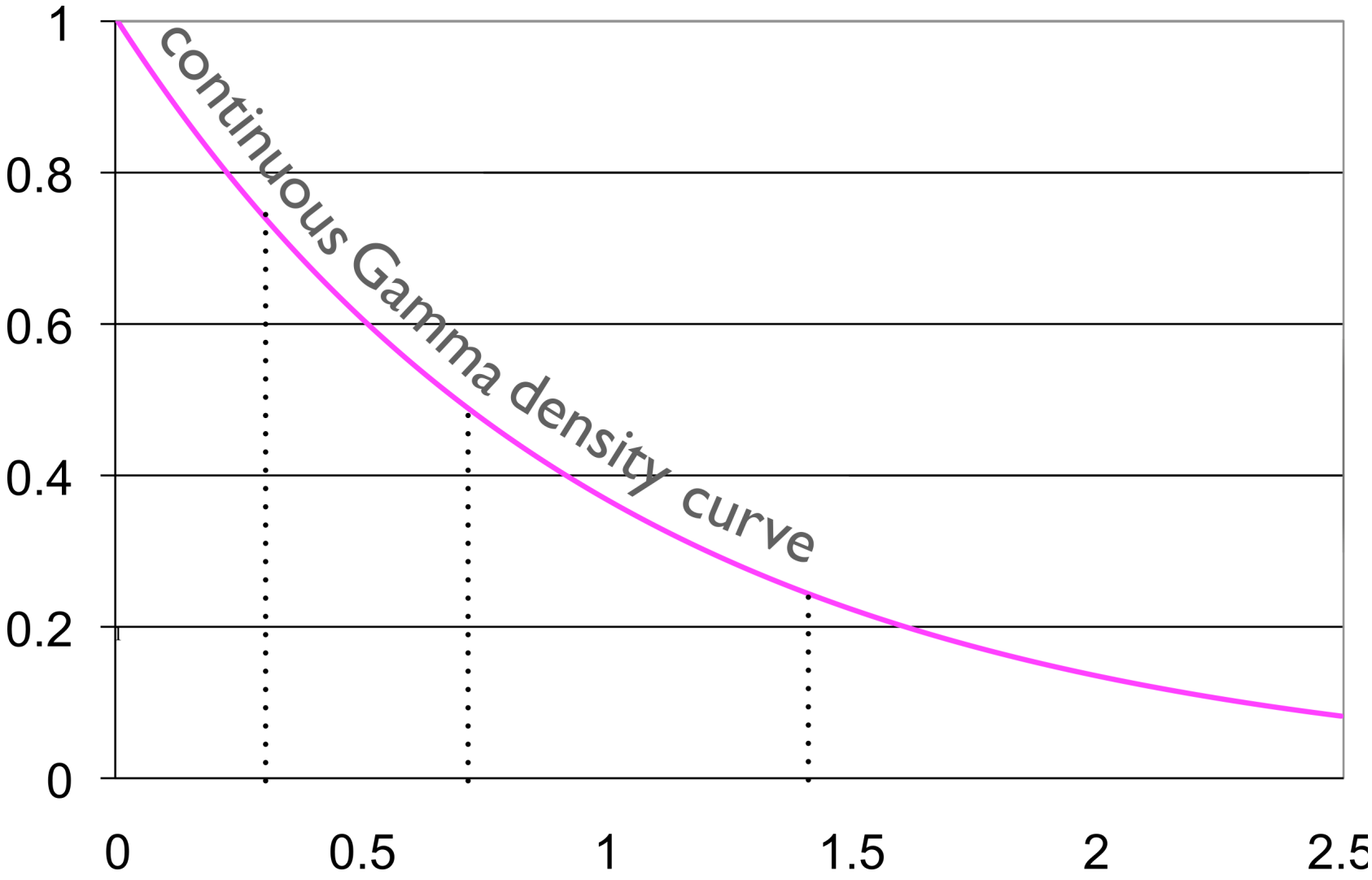
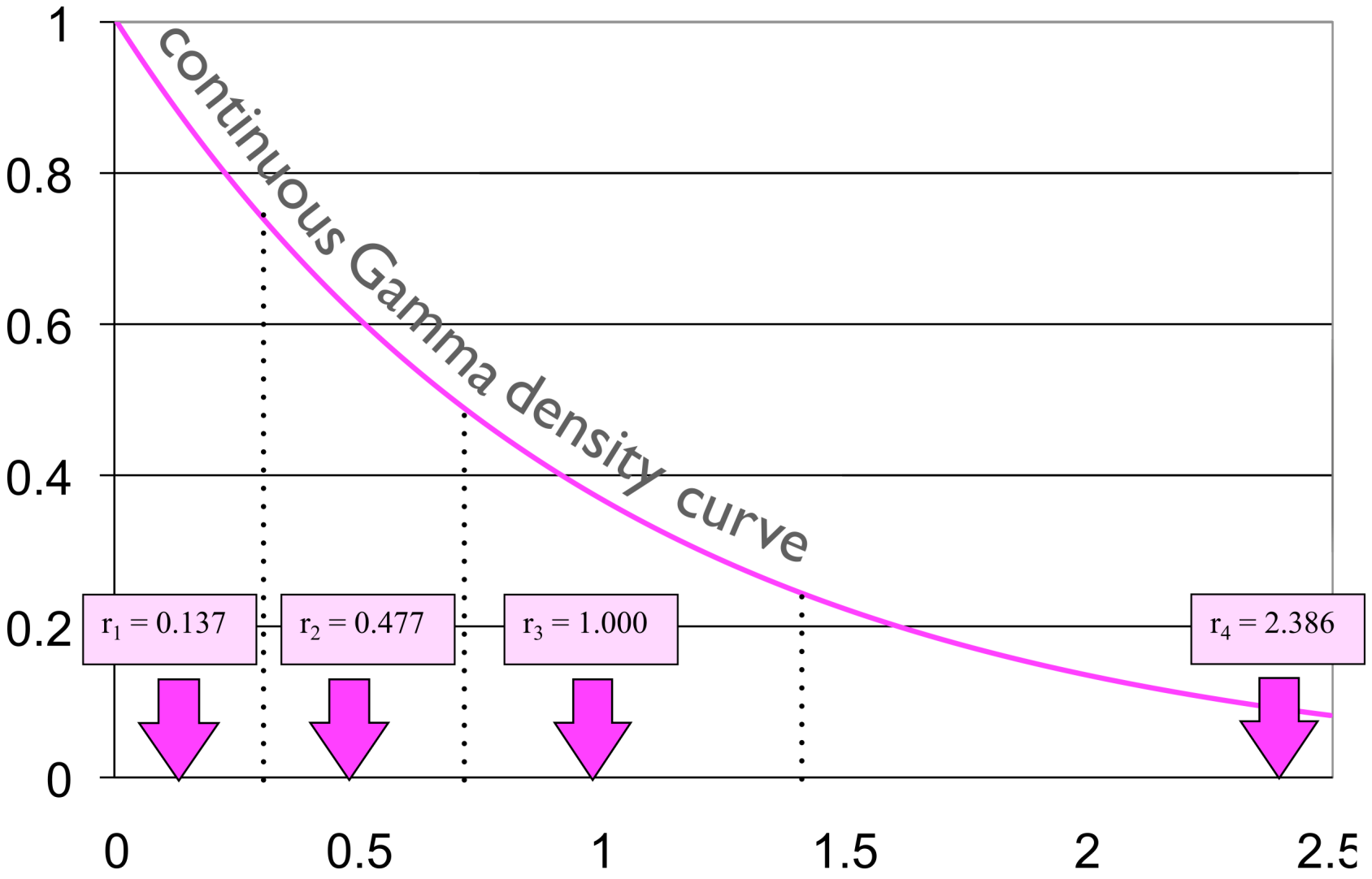
Mk(V) + Gamma



Adapted from Paul Lewis PhyloSeminar

Mk(V) + Gamma



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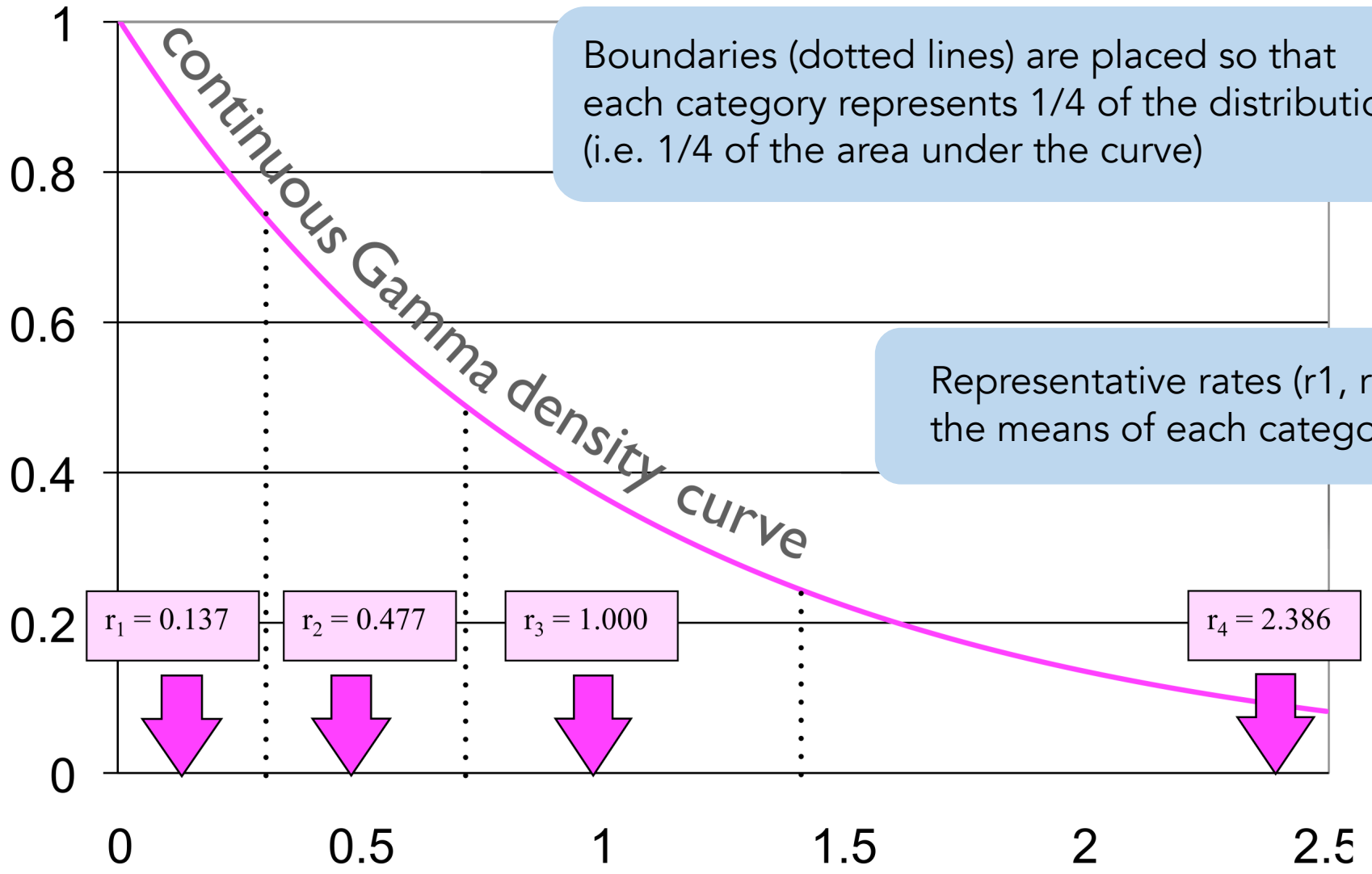


Boundaries (dotted lines) are placed so that each category represents 1/4 of the distribution (i.e. 1/4 of the area under the curve)

Representative rates (r1, r2, r3, r4) are the means of each category

Adapted from Paul Lewis PhyloSeminar

Mk(V) + Gamma

What do we do?

	T1	T2
Taxa A	0	0
Taxa B	0	1
Taxa C	1	2

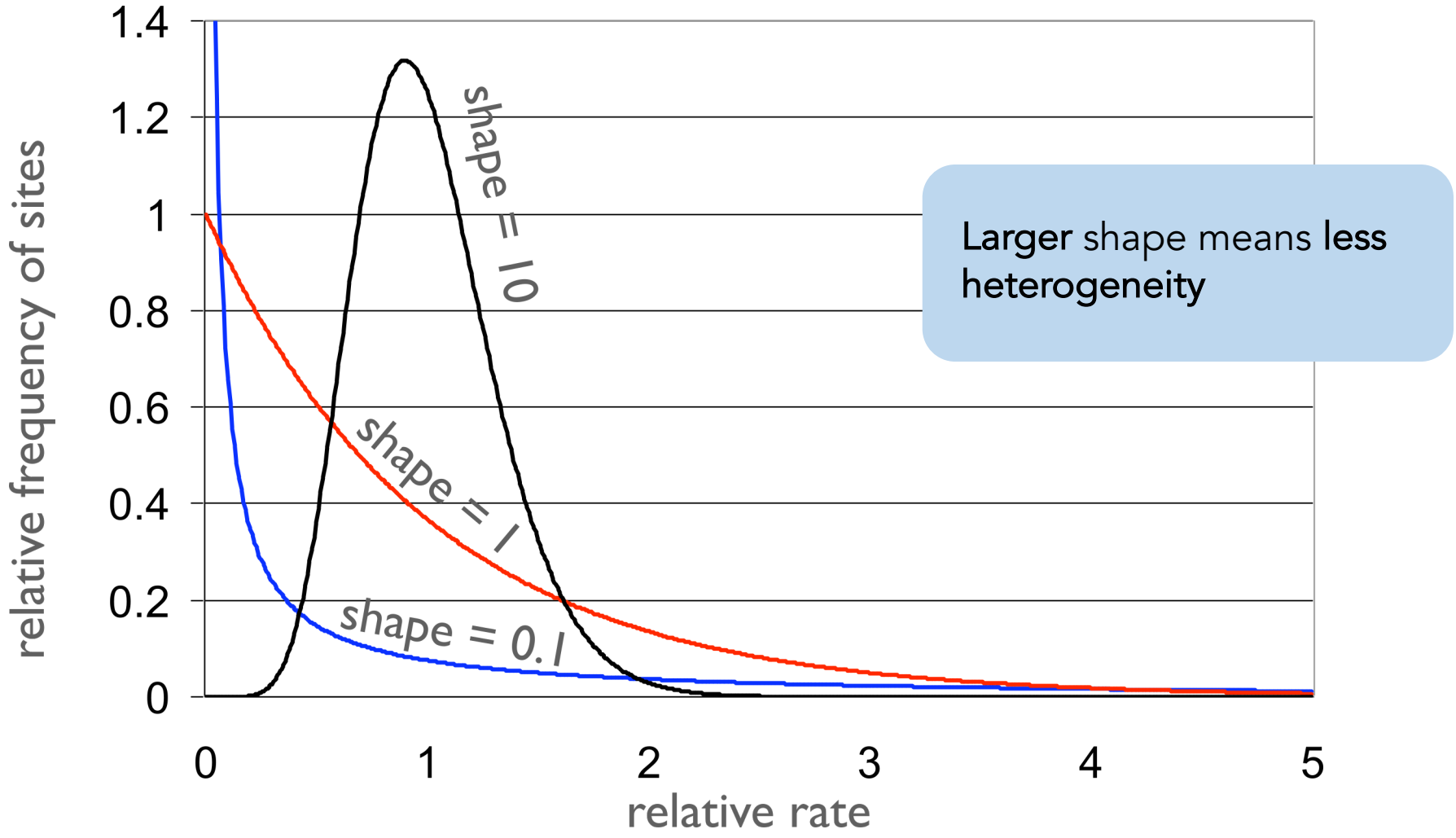
Slower R (R1,2)

Faster R (R4)

Allow each trait to evolve according to the rates drawn from the gamma distribution

One rate will fit the best and be the most influential for the likelihood calculation

Mk(V) + Gamma



Larger shape means less heterogeneity

Adapted from Paul Lewis PhyloSeminar

Partitioning Data

Grouping together parts of the alignment that have similar characteristics and or may have **evolved together** due to evolutionary pressures

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

The **defaults** in many phylogenetic software is to group by maximum observed state size

$$\begin{bmatrix} -\mu_0 & \mu_{01} \\ \mu_{10} & -\mu_1 \end{bmatrix}$$

$$\begin{bmatrix} -\mu_0 & \mu_{01} & \mu_{02} \\ \mu_{10} & -\mu_1 & \mu_{12} \\ \mu_{20} & \mu_{21} & -\mu_2 \end{bmatrix}$$

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If we have presence (1) absence (0) traits partitioning will always be a logical approach: what would transitioning to state 2 in this scenario even mean?

We should be cautious for traits describing a trait – just because we do not observe a state 2 can we be absolutely certain there never was one?

Justifying partitioning schemes is very important as they have a major impact on inference results

Other morphological models

Ordered Characters

Ordered characters can be placed in an order so that transitions only occur between adjacent states.

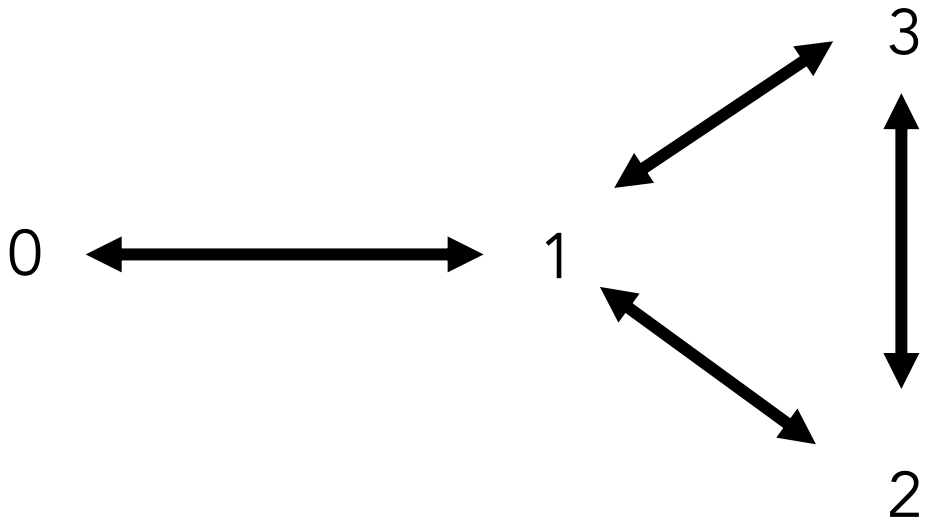
For example, "**intermediate**" species that are somewhere in between limbed and limbless – for example, the "mermaid skinks" (Sirenoscincus) from Madagascar, so called because they lack hind limbs. An ordered model might only allow transitions between limbless and intermediate, and intermediate and limbed; it would be impossible under such a model to go directly from limbed to limbless without first becoming intermediate.

For unordered characters, any state can change into any other state.

Ordered Characters

All characters ordered:

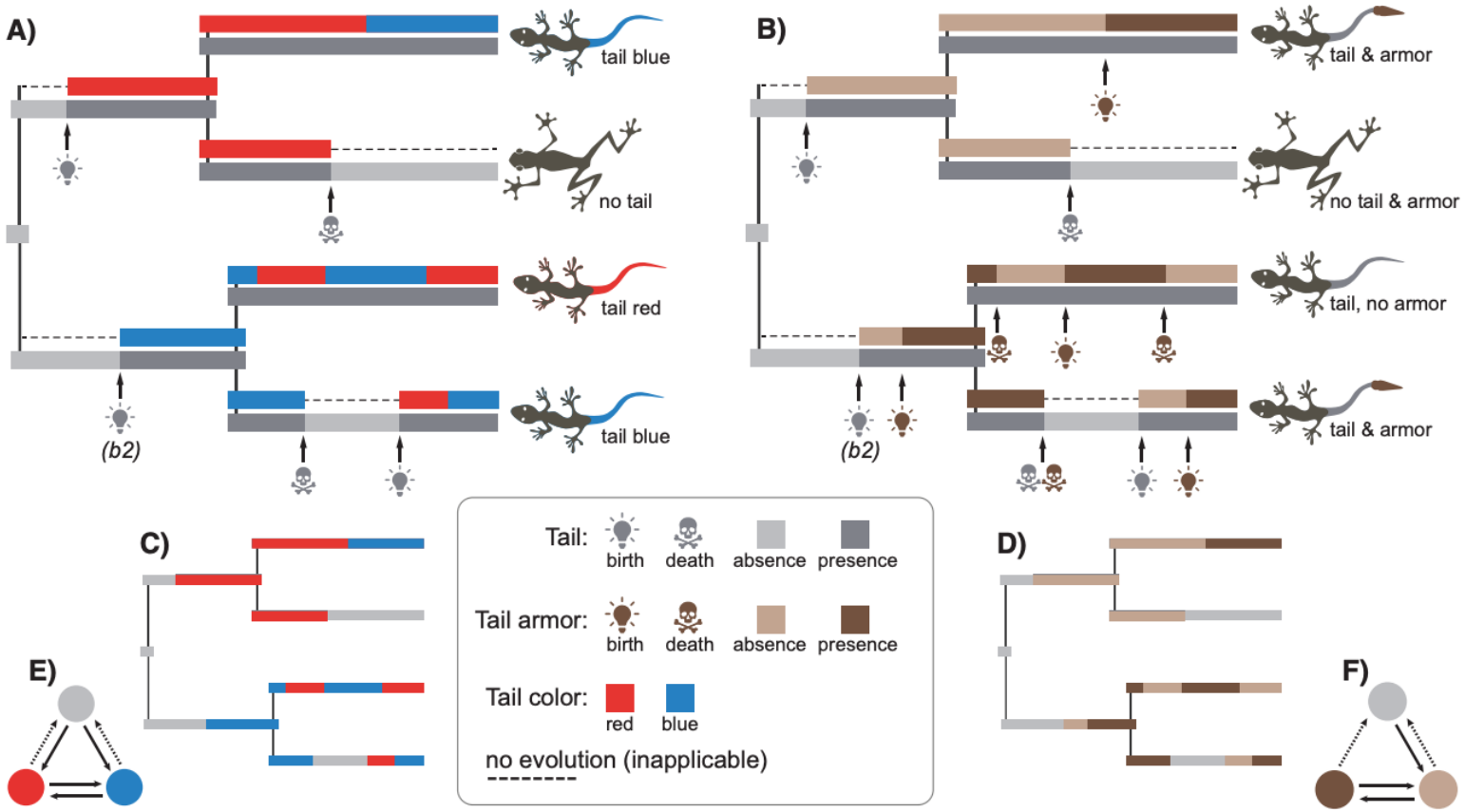
Specific characters ordered:



Embedded dependency model

Markov models for phylogenetic inference with anatomically dependent (inapplicable) morphological characters

Non-applicable characters only considered when they are present (1)



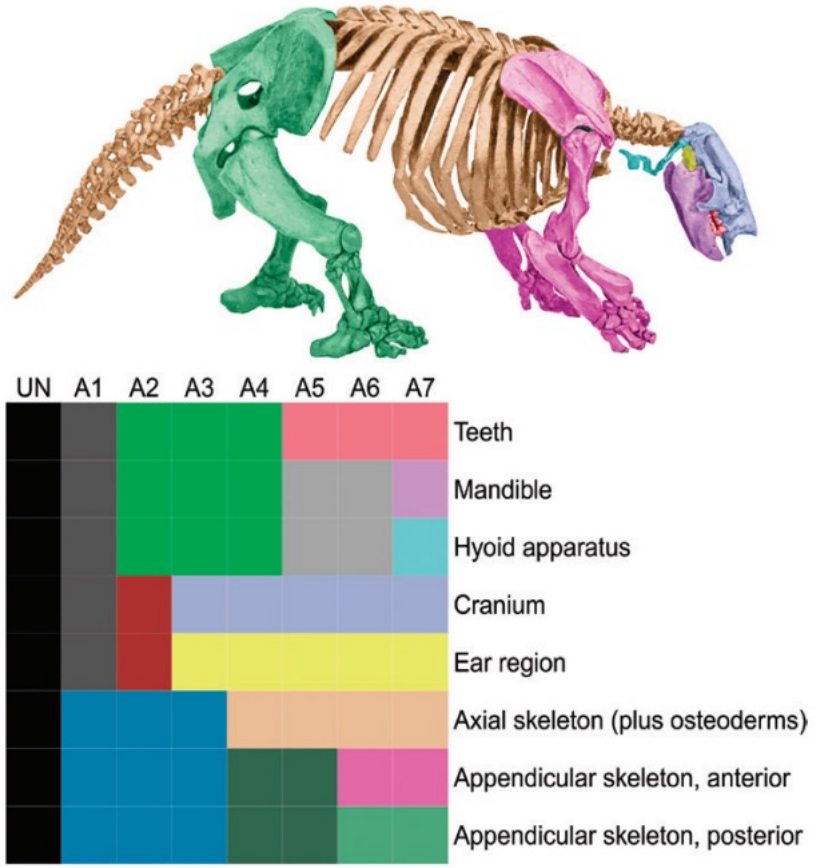
Tarasov 2023 Systematic Biology

Alternative Partitioning schemes

Reassessing the phylogeny and divergence times of sloths (Mammalia: Pilosa: Folivora)

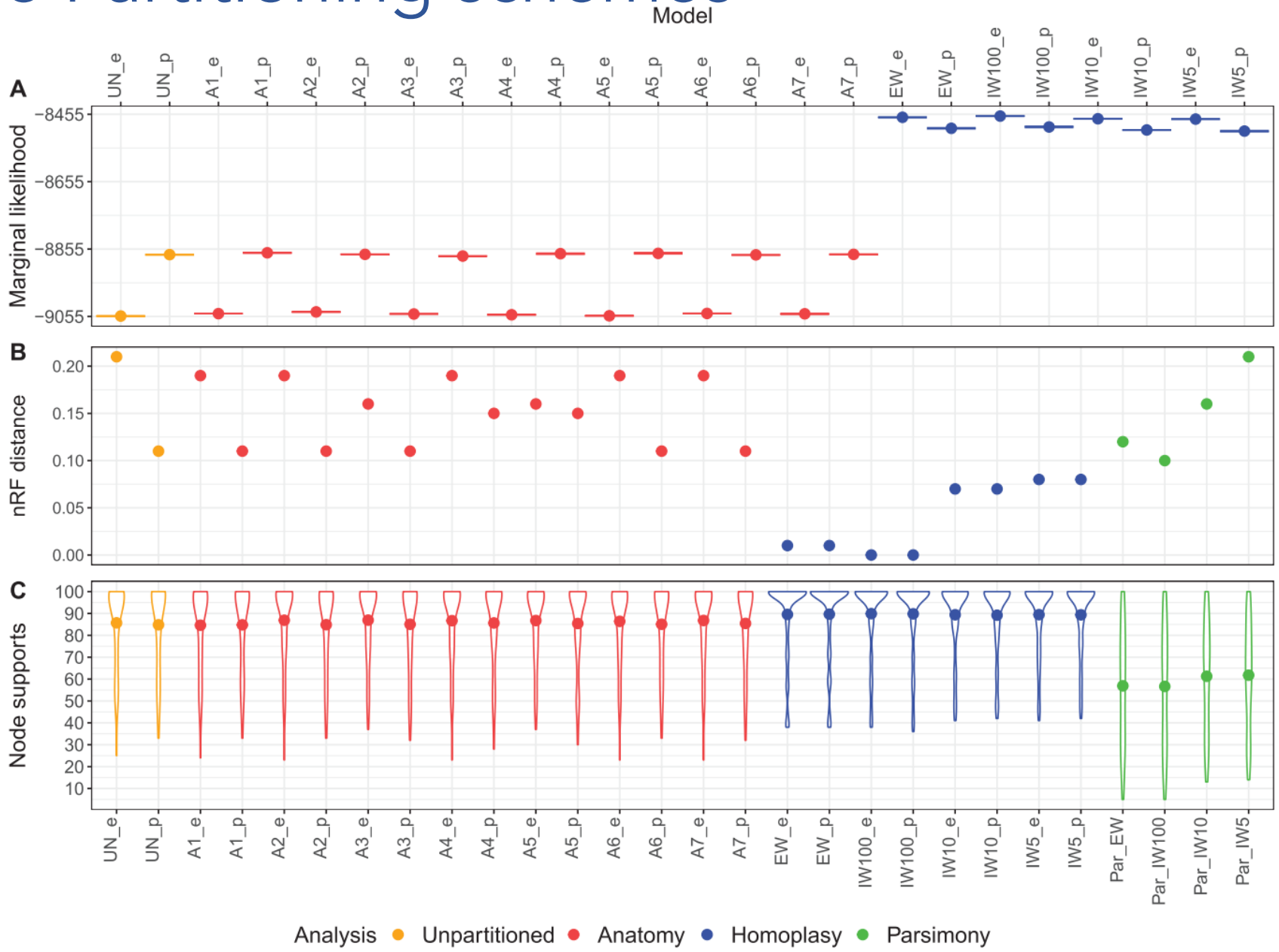
Characters can be groups based on anatomical region

Other criteria such as the degree of homoplasy present in a character was explored in this study – and found to be a better fit using Bayes factors



Casali et al 2022 Zoological Journal of the Linnean Society

Alternative Partitioning schemes



Casali et al 2022 Zoological Journal o the Linnean Society

Challenges with morphological data

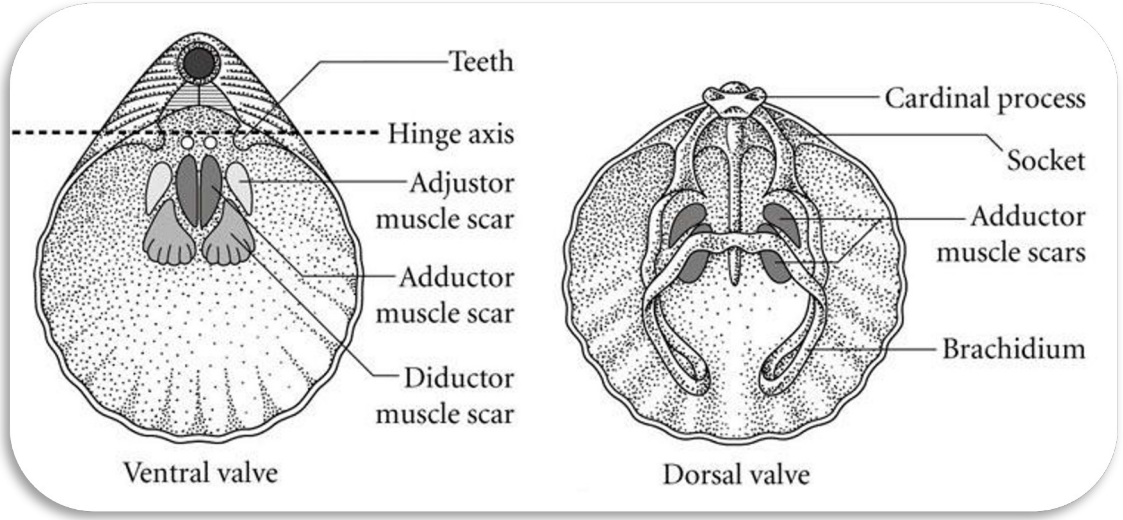
Generalising assumptions across different traits is often not possible

Modelling special characters in matrices

```
00151O010?00-100--0000000000
000500010?200100--0010010000
002500010?200100--0?10010000
00?5?0010?200100?-0???010110
0015000101201000430100011111
0015000101201010440111011111
??050?????201000440?11011111
```

Challenges with morphological data

Morphological matrices are often quite small:
- Collection is very time consuming
- Number of characters available can be very small depending on the group



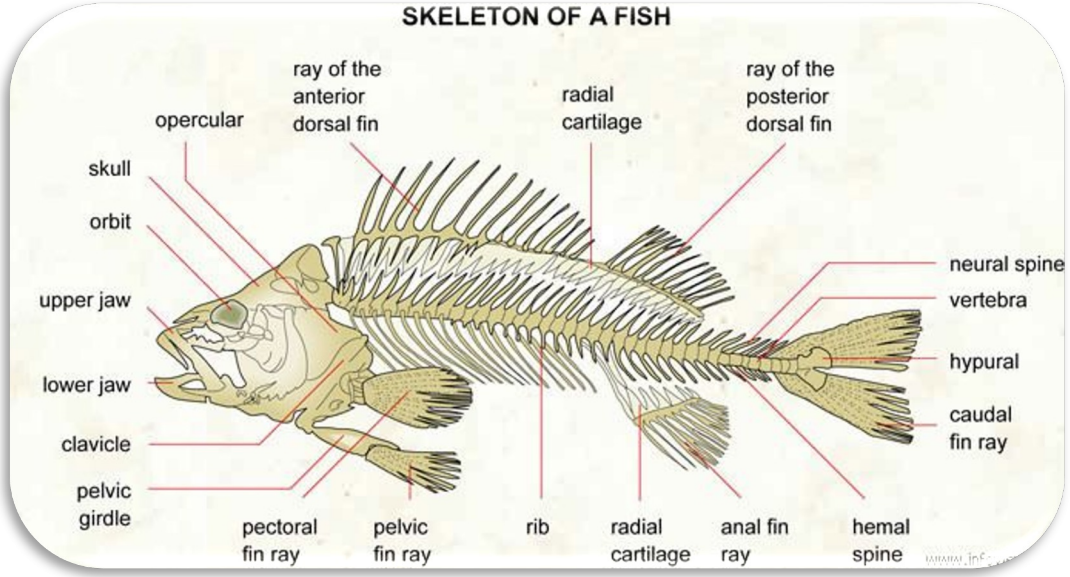
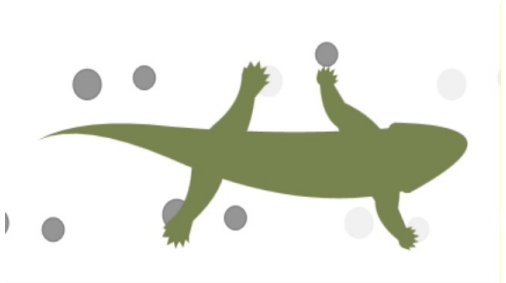


Image source

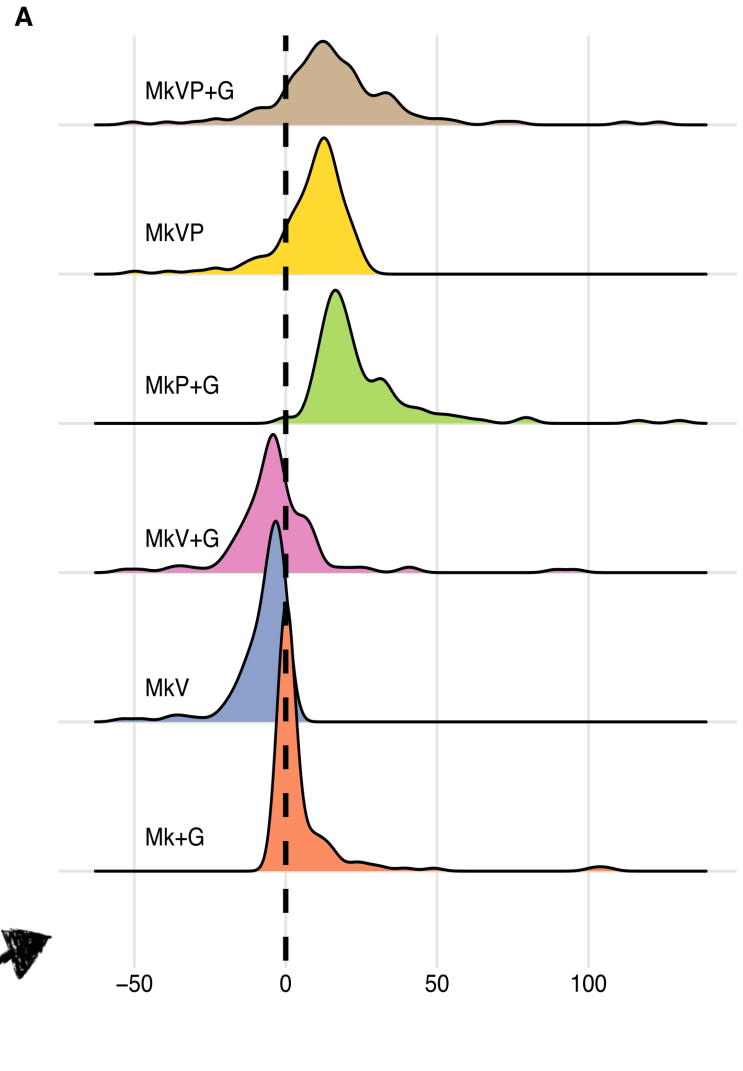
Model impact on key parameter estimates



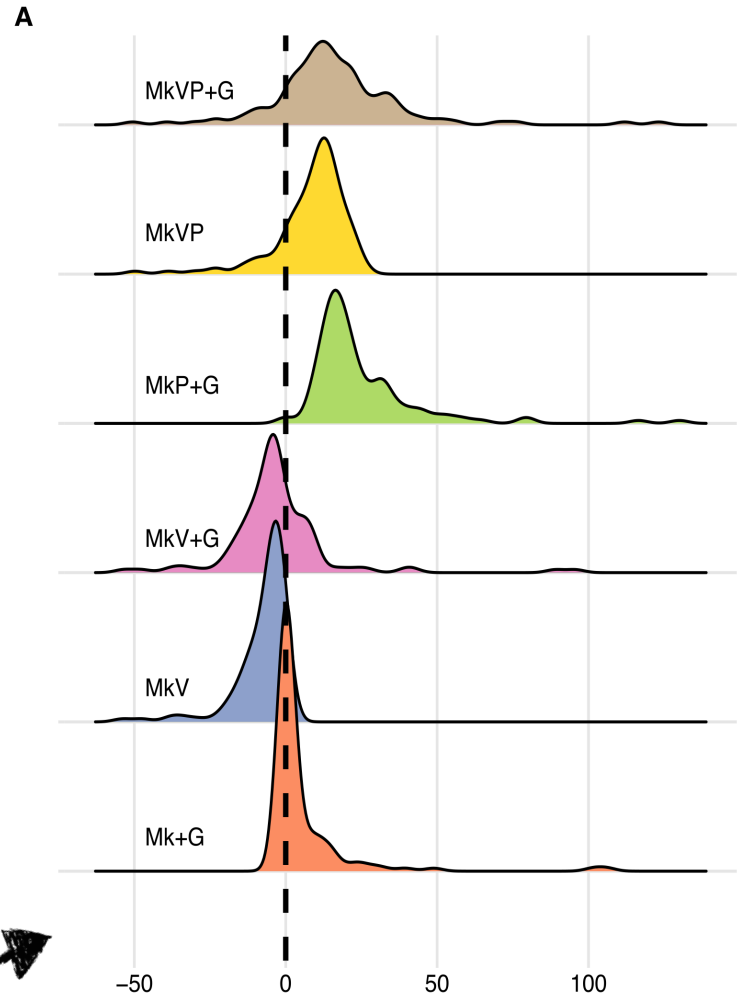
Example of 114 empirical **tetrapod** matrices

Looked at the impact on:

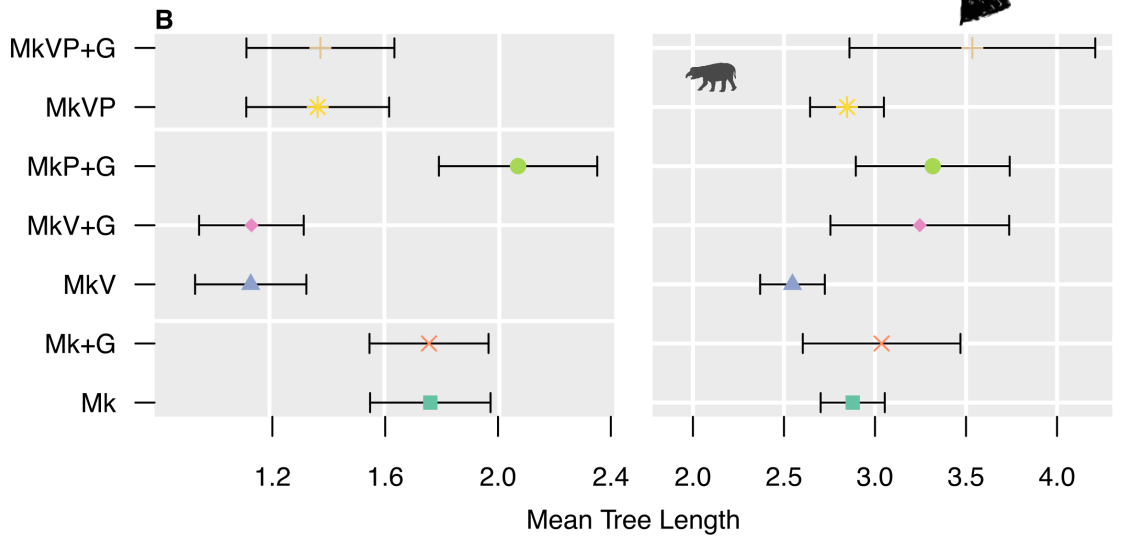
- branch lengths **(evolutionary distances)**
- Tree topology **(species relationships)**



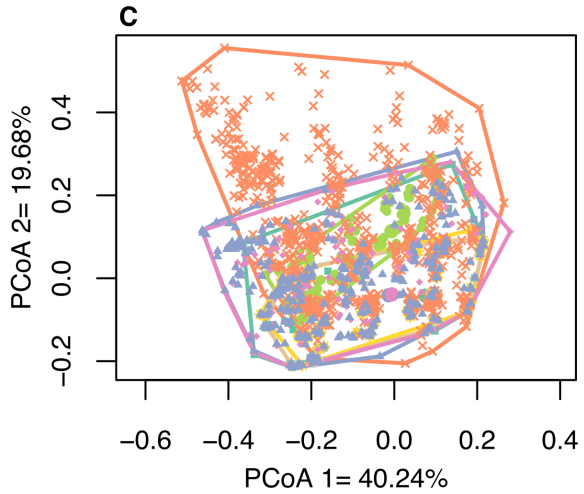
Percentage difference in tree length relative to Mk model

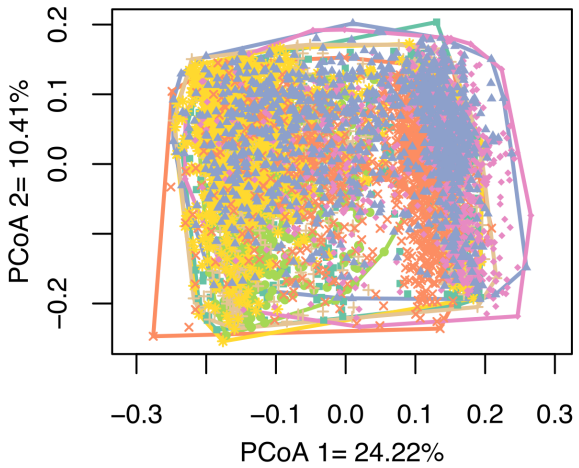


Percentage difference in tree length relative to Mk model



Tree length of two different data sets





Rf distances of two data sets

How do we choose a model?

Model selection

Bayes factors are commonly used to determine the **relative fit** between model.

It relies on comparing the marginal likelihoods approximated from different models.

The ML measures the average fit of a model to our data.

We use MCMC to avoid calculating this number as it is computationally expensive and often not directly possible.

Model selection

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

Posterior

Likelihood

Priors

Marginal likelihood

Marginal likelihood

Marginal probability of the data (denominator in Bayes' rule) is the expected value of the likelihood with respect to the prior distribution.

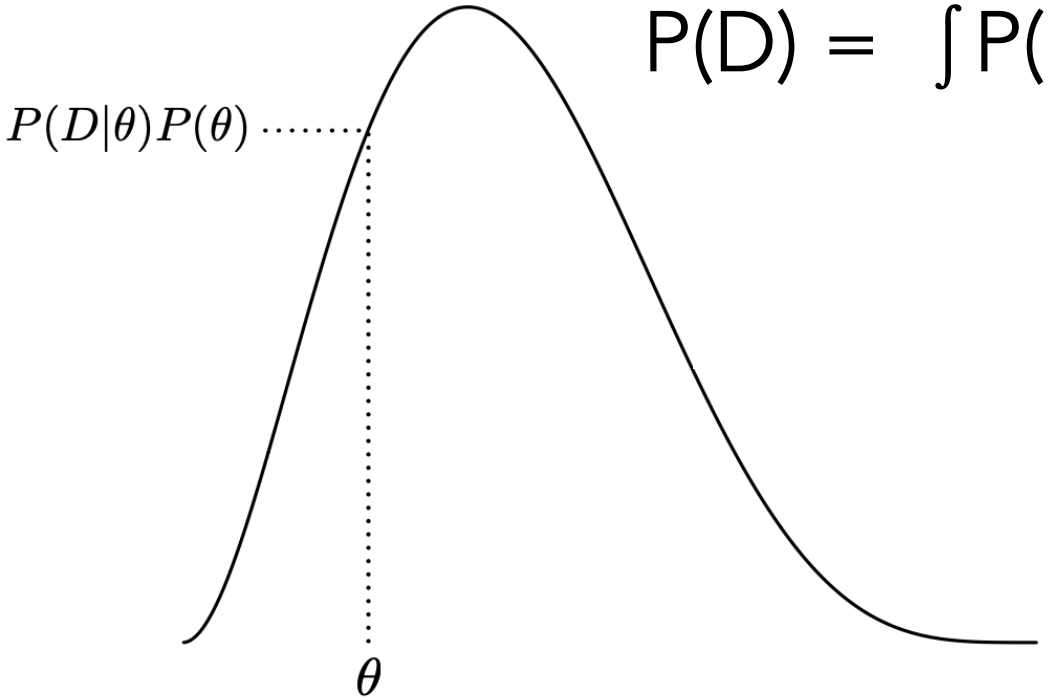
If likelihood measures model fit, then the marginal likelihood measures the average fit of the model to the data over all parameter values.

What is the expected value?

Marginal likelihood

P (data | model)

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



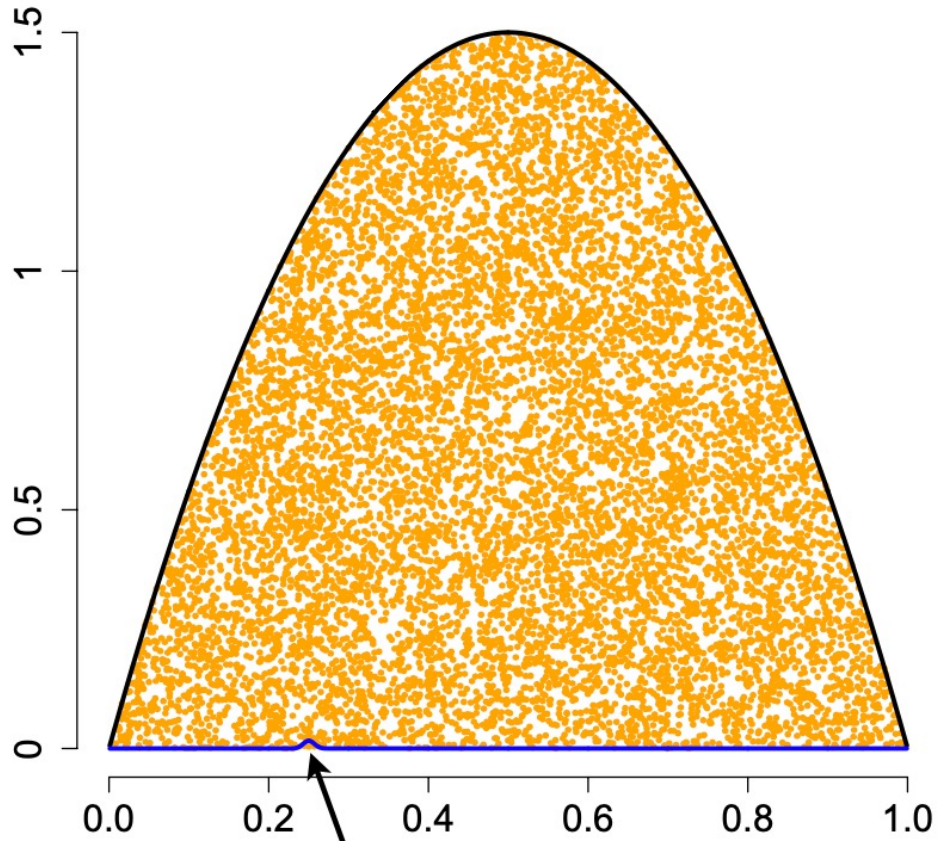
The marginal likelihood is used to evaluate the overall fit of the model to the data, integrating over all parameter values.

Adapted from Paul Lewis PhyloSeminar

Marginal likelihood

P (data | model)

Very small, single number between the posterior distribution and the prior



Adapted from Paul Lewis PhyloSeminar

Approximating the marginal likelihood

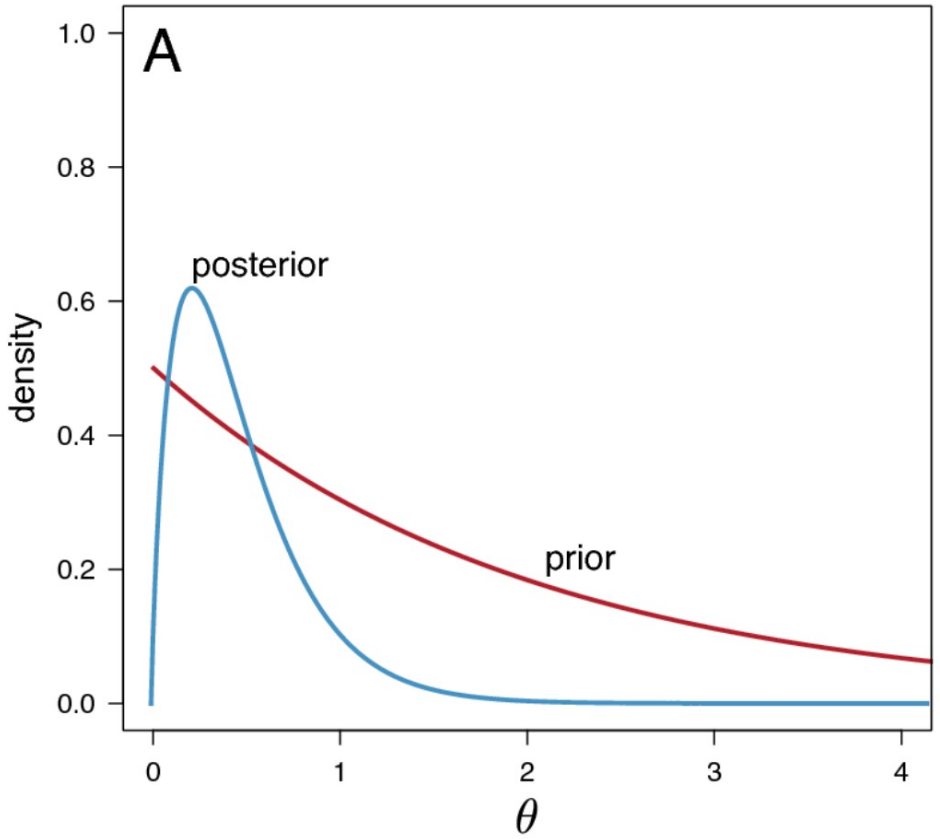
There are two common algorithms to do this:

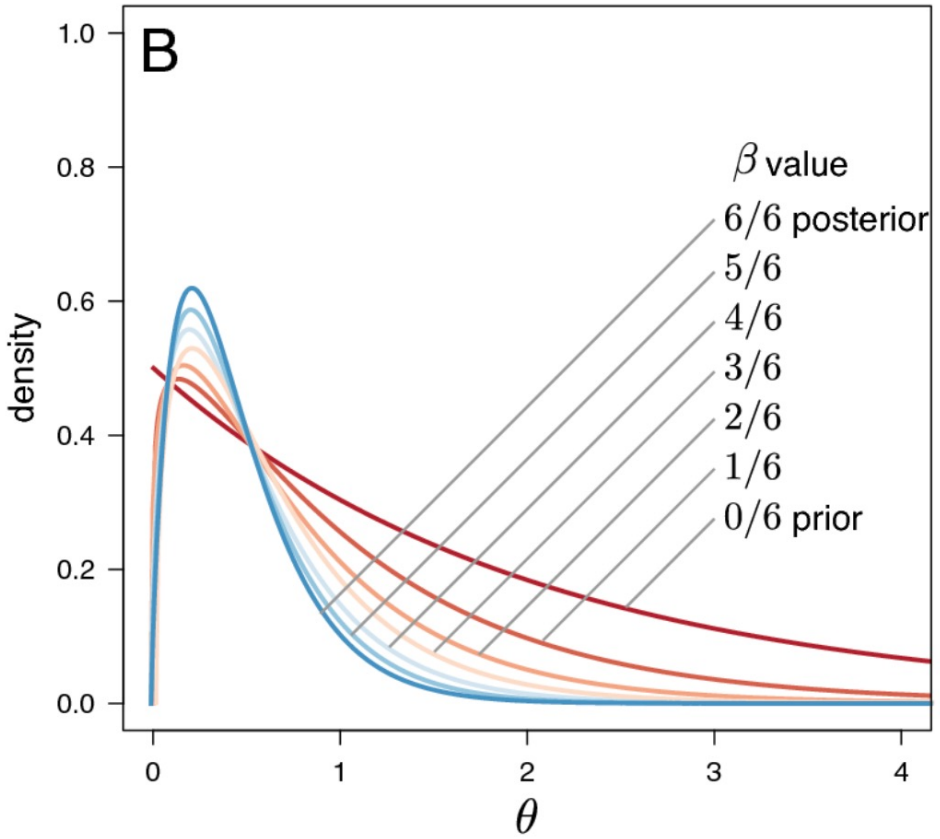
- Stepping stone
- Path sampling

Both of these approaches are computationally expensive

Stepping-stone algorithms are like a series of MCMC simulations that iteratively sample from a specified number of distributions that are discrete steps between the posterior and the prior probability distributions.

Stepping stone algorithm





β = 1 this is the posterior distribution

β = 0 this is the prior distribution

Image source

Bayes factors

$$B_{01} = \frac{P(D \mid M_0)}{P(D \mid M_1)} = \frac{\text{Marginal likelihood for model } M_0}{\text{Marginal likelihood for model } M_1}$$

Bayes factors

$$B_{01} = \frac{P(D \mid M_0)}{P(D \mid M_1)} = \frac{\text{Marginal likelihood for model } M_0}{\text{Marginal likelihood for model } M_1}$$

Marginal likelihoods are often on the log scale so the Bayes factor can be calculated as:

$$\log B_{01} = \log P(D \mid M_0) - \log P(D \mid M_1)$$

Interpreting Bayes factors

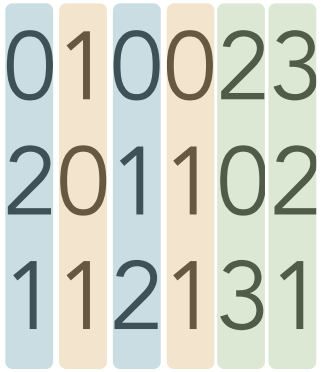
Strength of evidence	$BF(M0,M1)$	$log(BF(M0,M1))$
Negative (supports M_1)	<1	<0
Barely worth mentioning	1 to 3.2	0 to 1.16
Substantial	3.2 to 10	1.16 to 2.3
Strong	10 to 100	2.3 to 4.6
Decisive	>100	>4.6

Table source

Exercise:
Use stepping stone to determine the fit between models

Issues with Bayes factors

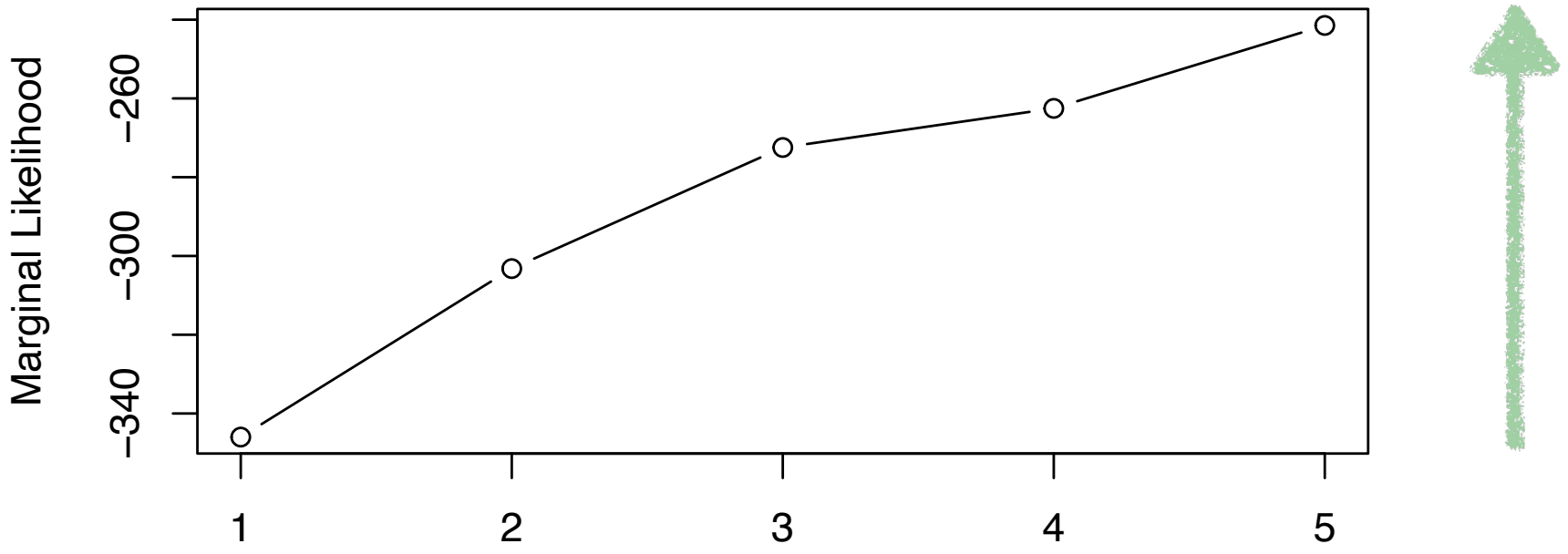
The way we **partition data** for morphological data is different to molecular



Unpartitioned everything in Q-matrix of size 4

Partitioning the data puts characters into correctly sizes Q-matrix

Issues with Bayes factors



Data set with 6 states

As the number of partitions increases, so does the likelihood

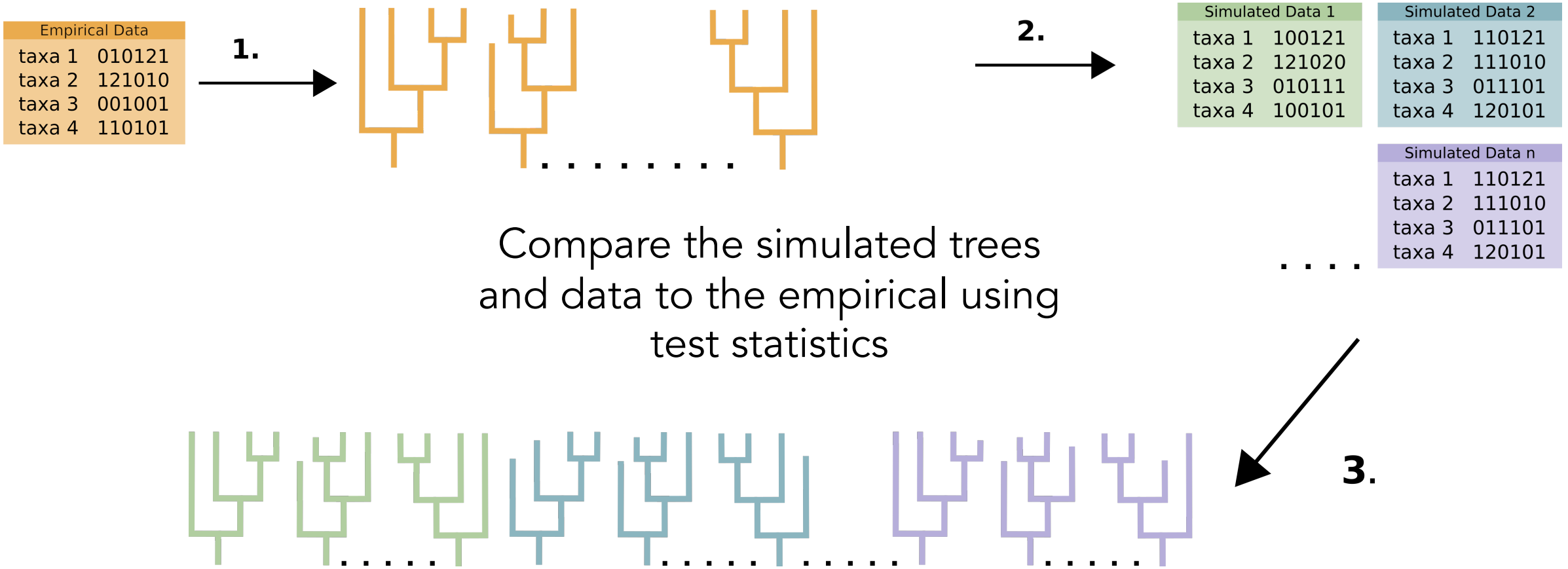
Model Adequacy

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

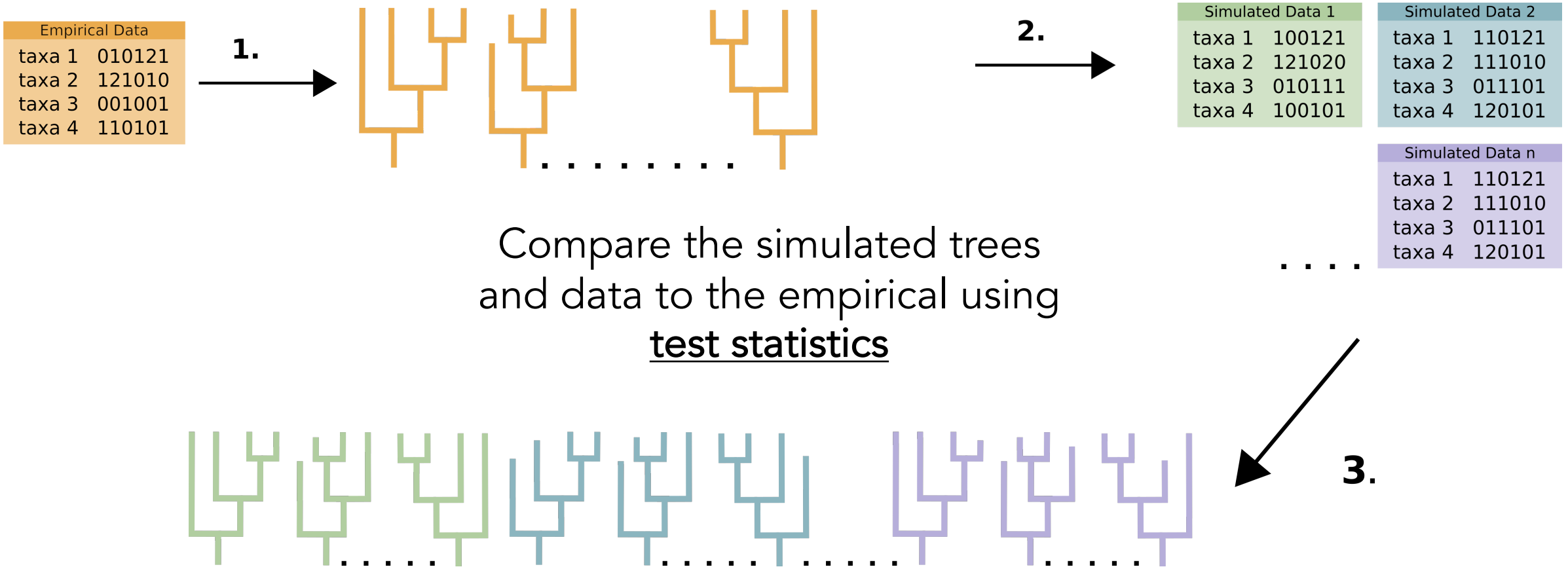
Gives the **absolute fit**

One approach is **Posterior Predictive Simulations**

Posterior Predictive Simulations



Posterior Predictive Simulations



Test Statistics

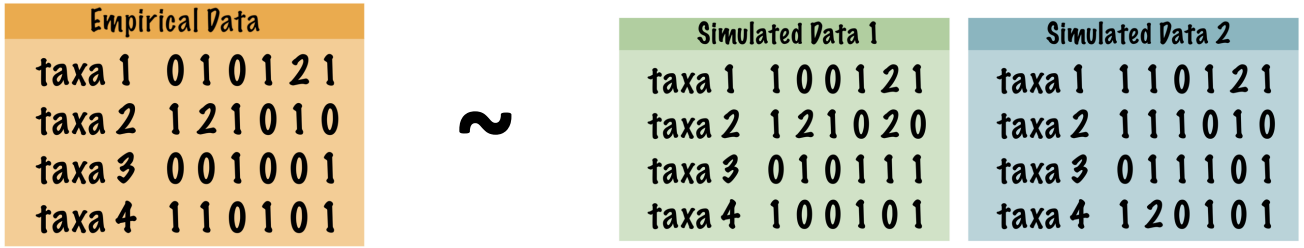
A test statistic is a **numerical summary** of data.

A value that captures the characteristic of you data.

For PPS we have 3 categories:

Data-based, inference-based, mixed

Test Statistics



Data test stats

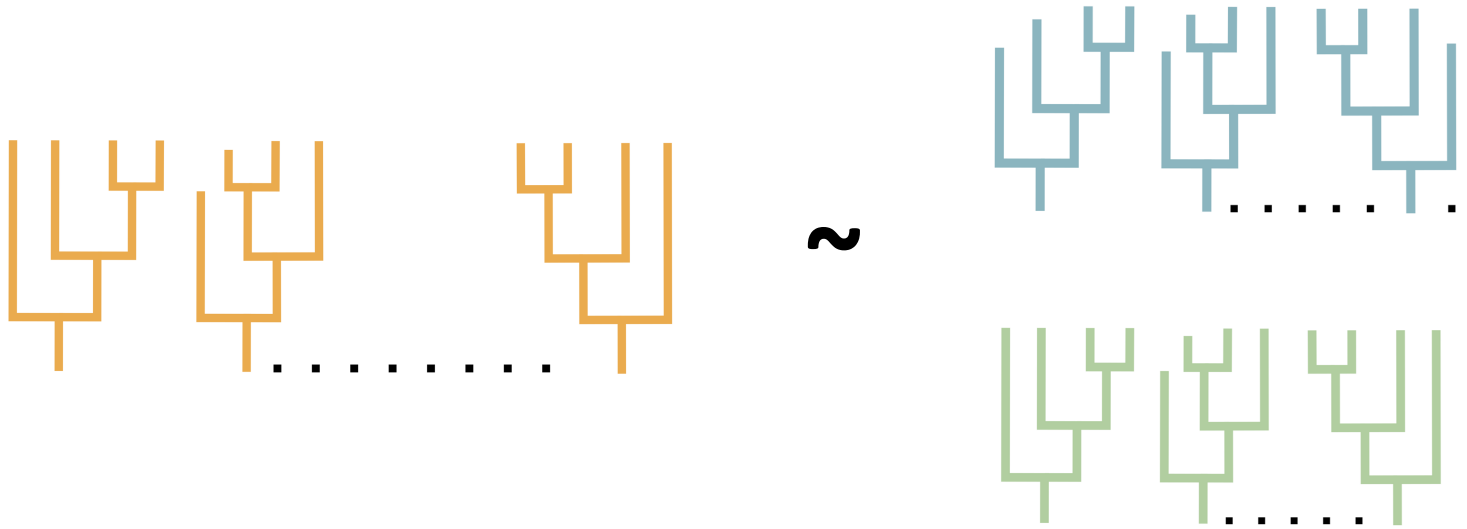
Disparity is a measure of the **range or significance of morphology** in a given sample of organisms

i. Gowers Coefficient
ii. Generalised Euclidean Distance

Test Statistics

Tree test stats

i. Tree length: sum of the branch lengths.
ii. Robinson Foulds: topological uncertainty within the posterior



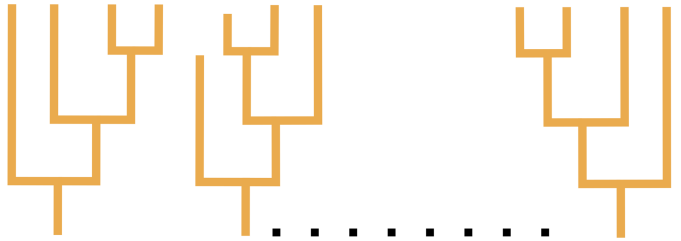
Test Statistics

Mixed test stats

i. Consistency Index
Measures the amount of **homoplasy** (convergent evolution) in a data set

ii. Retention index
Measures the amount of **synapomorphies** in a data set

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



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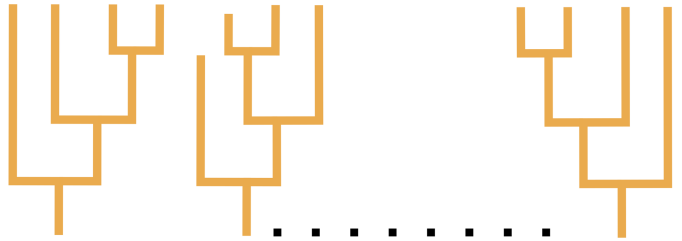
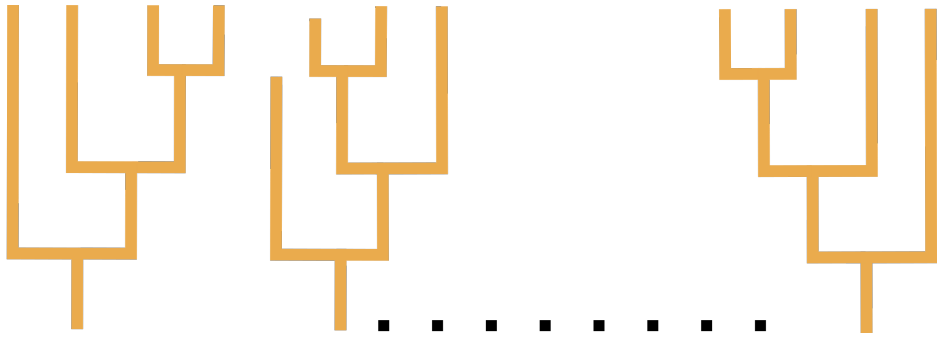
Simulated Data 1	
taxa 1	1 0 0 1 2 1
taxa 2	1 2 1 0 2 0
taxa 3	0 1 0 1 1 1
taxa 4	1 0 0 1 0 1

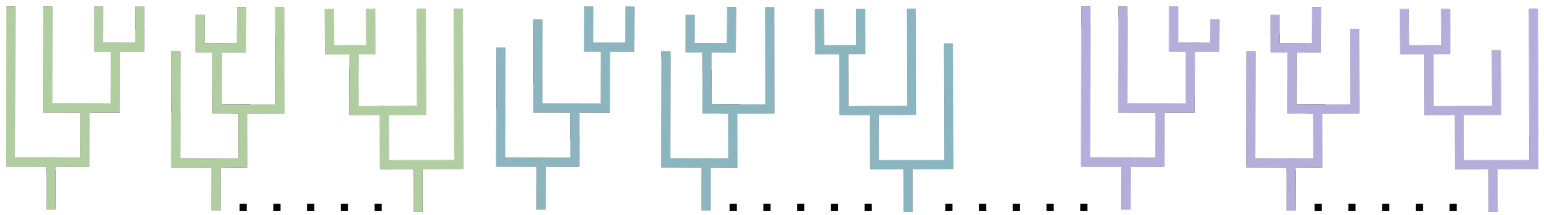


Test Statistics

Take Robinson Foulds Distance



1 empirical RF value



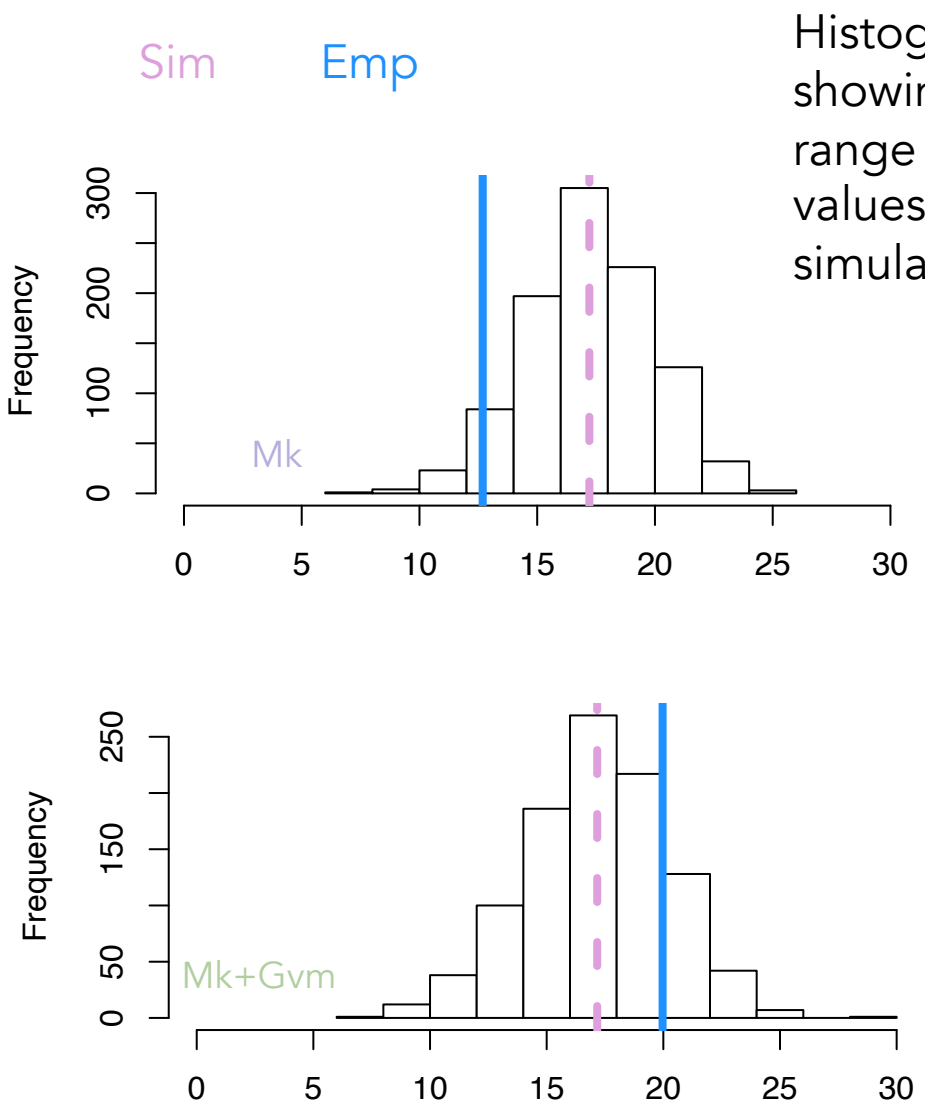
Sim 1 RF value

Sim 1 RF value

Sim n RF value

Test Statistics

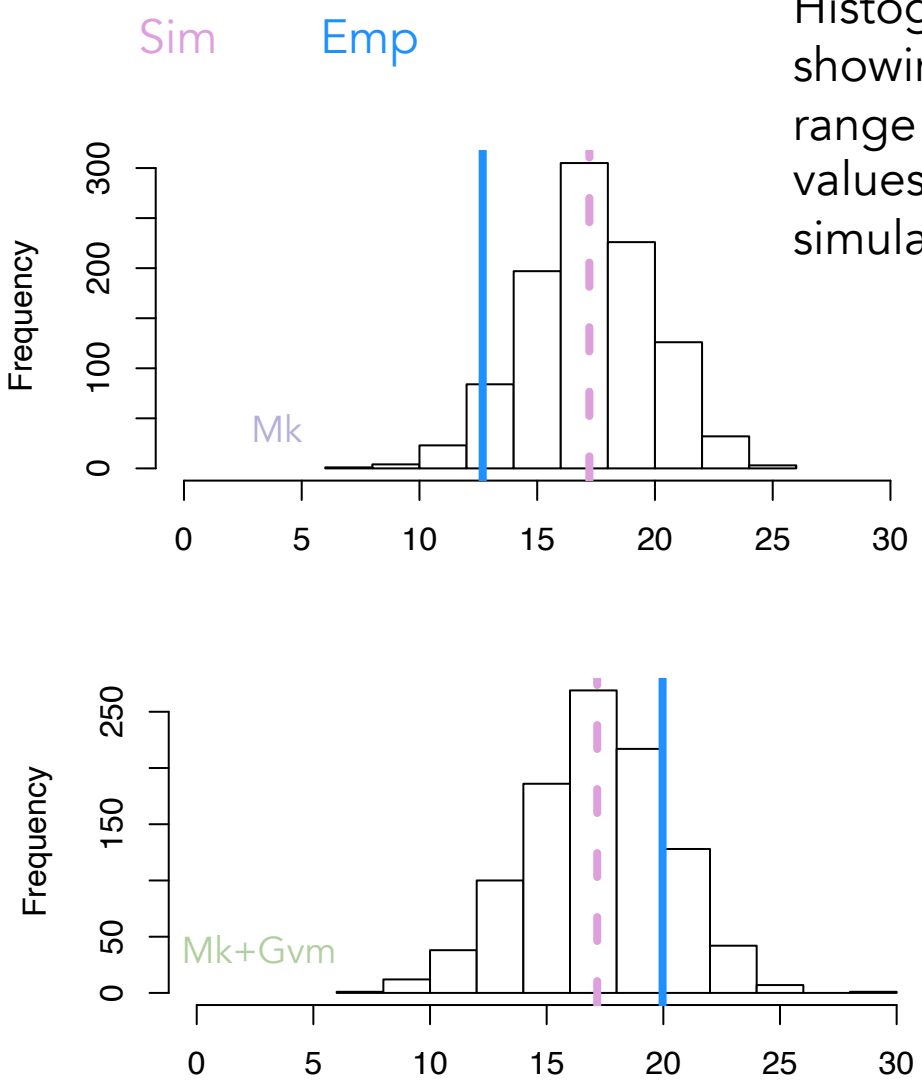
Robinson Foulds Distance



Histograms showing the range of RF values for all the simulated data

Test Statistics

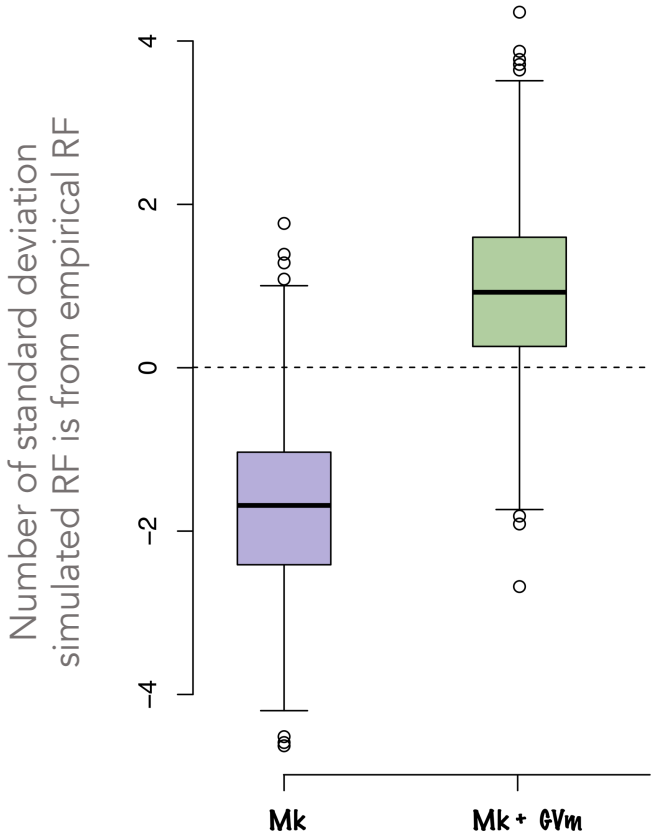
Robinson Foulds Distance



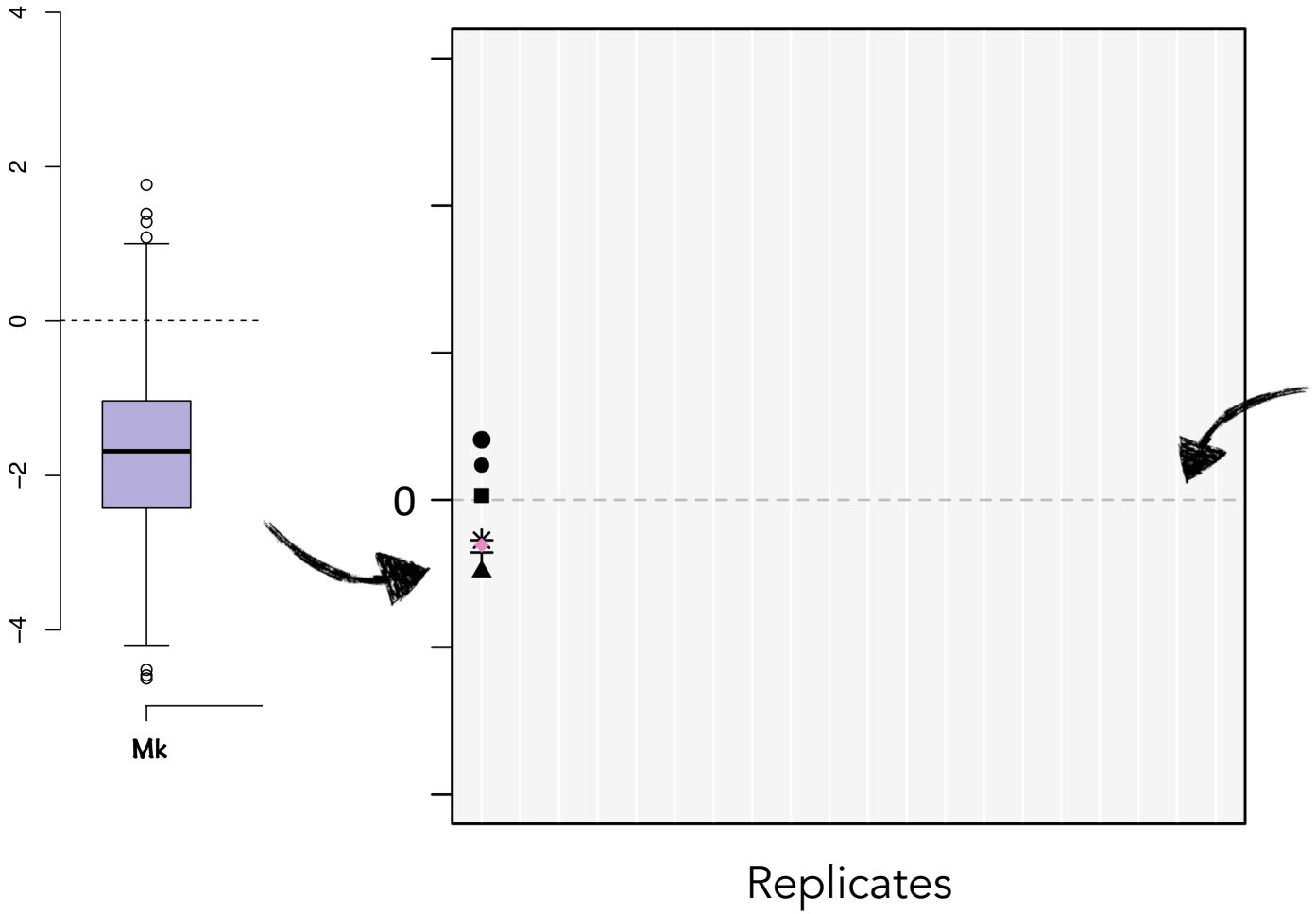
Histograms showing the range of RF values for all the simulated data

We can use this to calculate **effect sizes**

$$\frac{\text{Empirical TS - SimTs}}{\text{Sd(All Sim TS)}}$$

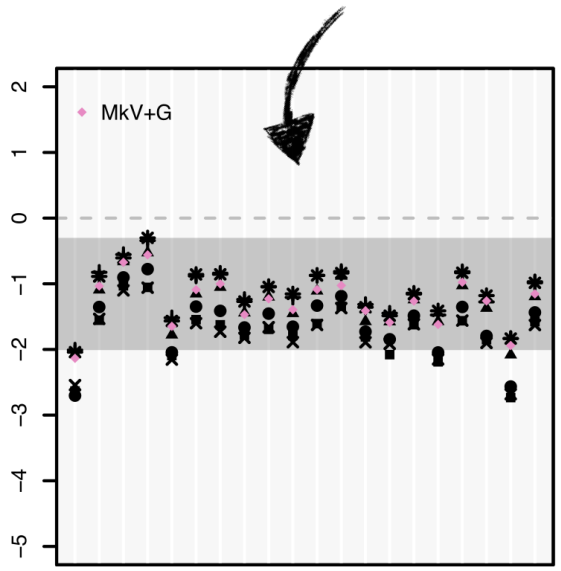


Effect Sizes



Data-based test stats

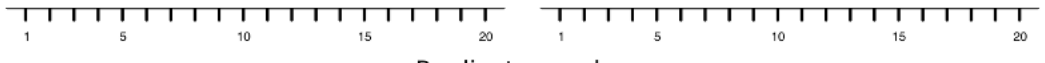
Closer to zero the better the model



Simulated under the MkV+G model:

Simulated under the MkVP+G model:

We don't see the correct model consistently closest to zero



Grower's Coefficient

Generalised Euclidean Distances

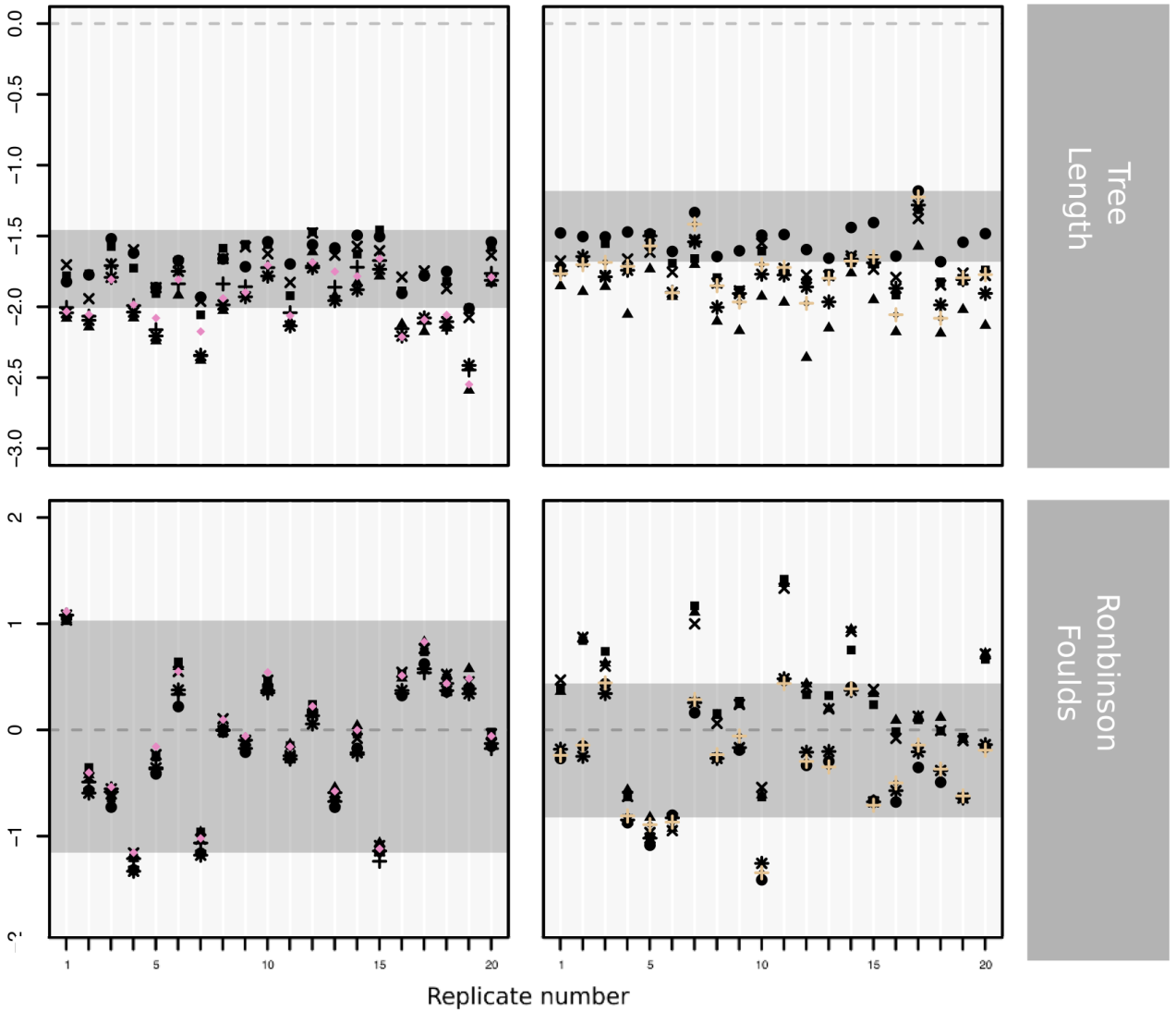
These test statistics **are not informative** about the correct model

Tree-based test stats

Closer to zero the better the model

Simulated under the MkV+G model:

Simulated under the MkVP+G model:



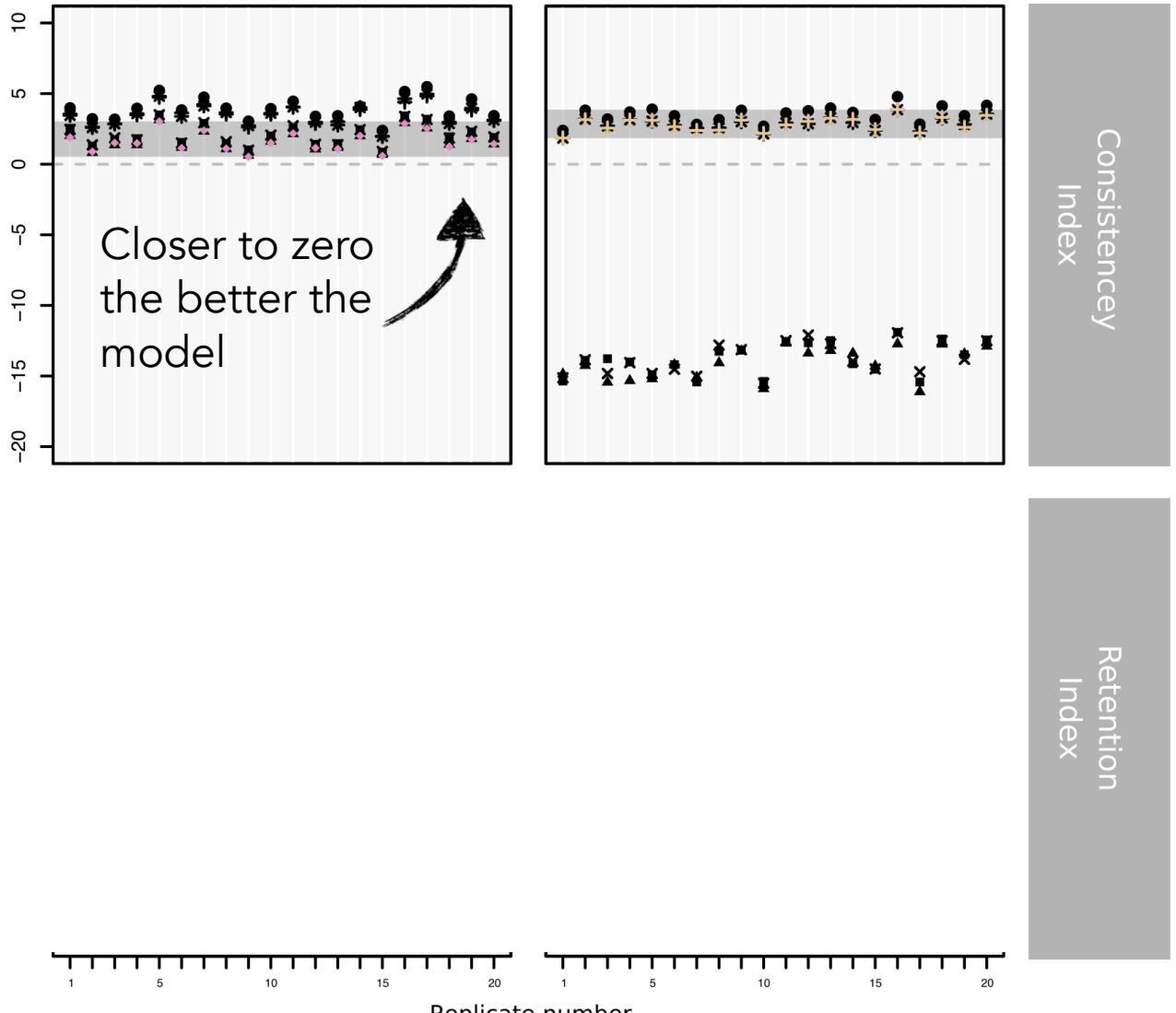
We don't see the correct model consistently closest to zero

These test statistics **are not informative** about the correct model

Mixed test stats

Simulated under the MkV+G model:

Simulated under the MkVP+G model:



We do see the correct model consistently closest to zero

These test statistics **are informative** about the correct model

Empirical data sets

MkVP+G

MkVP

MkP+G

MkV+G

MkV

Mk+G

Mk

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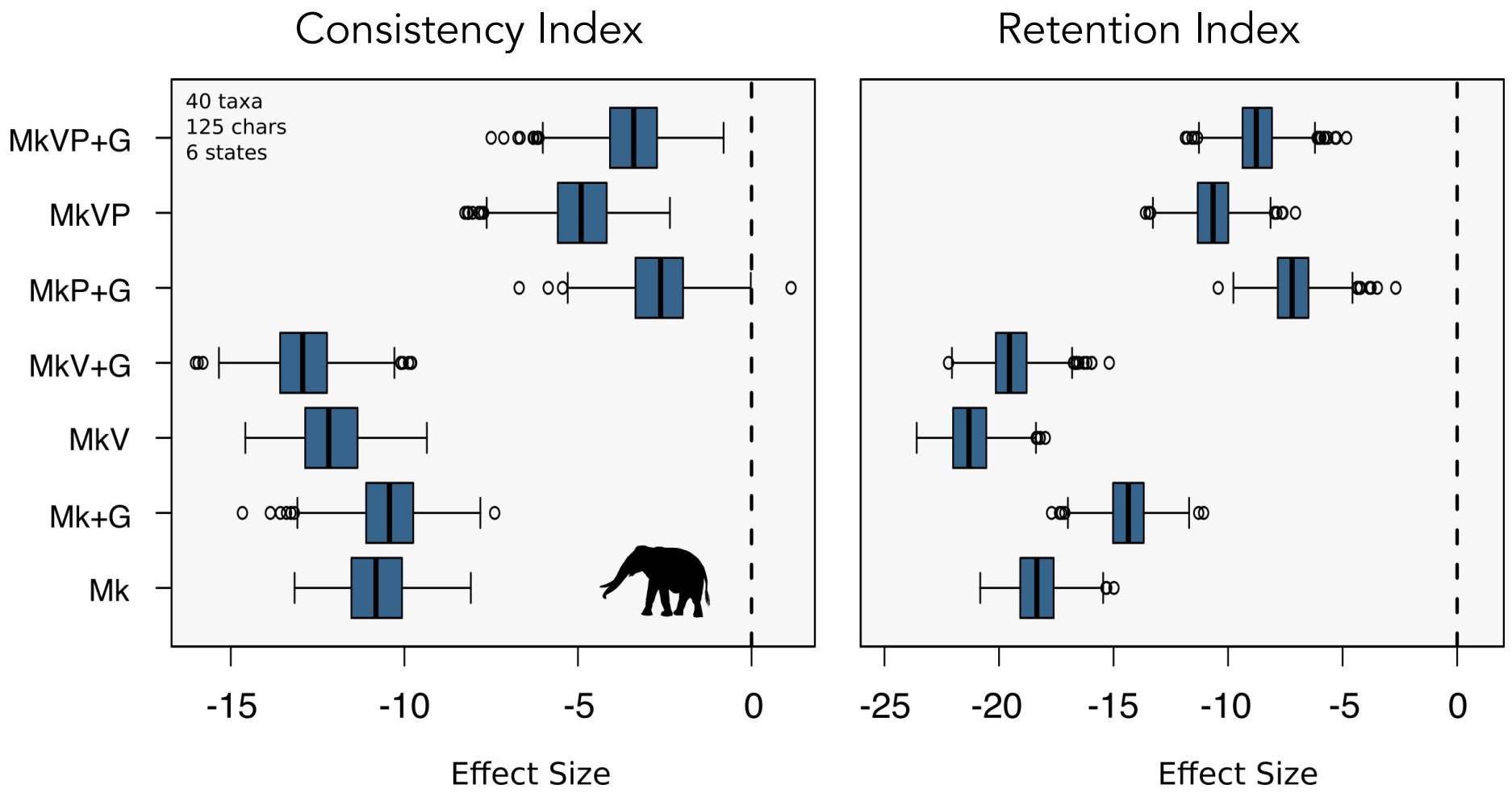
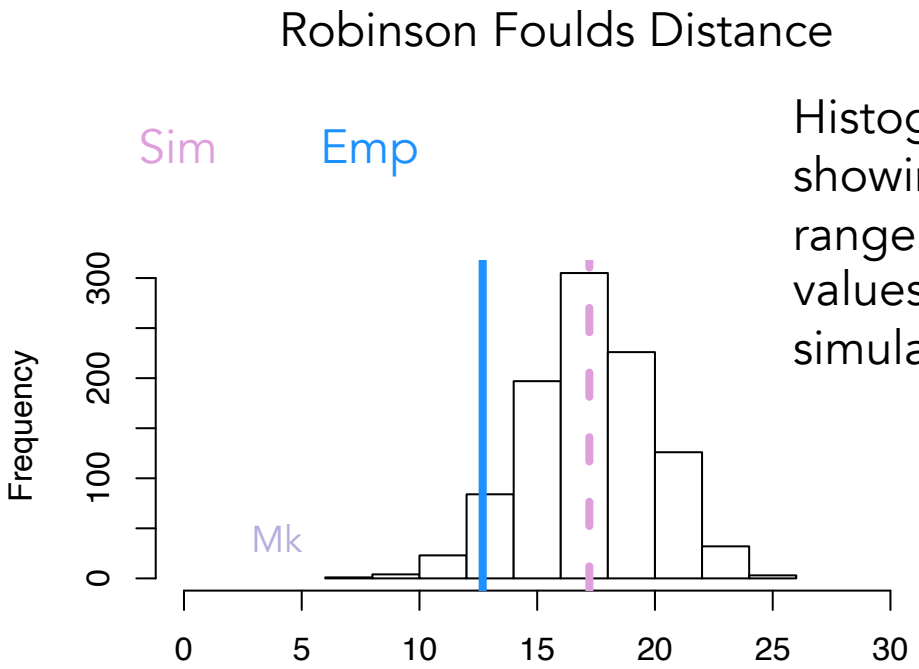
Empirical data sets



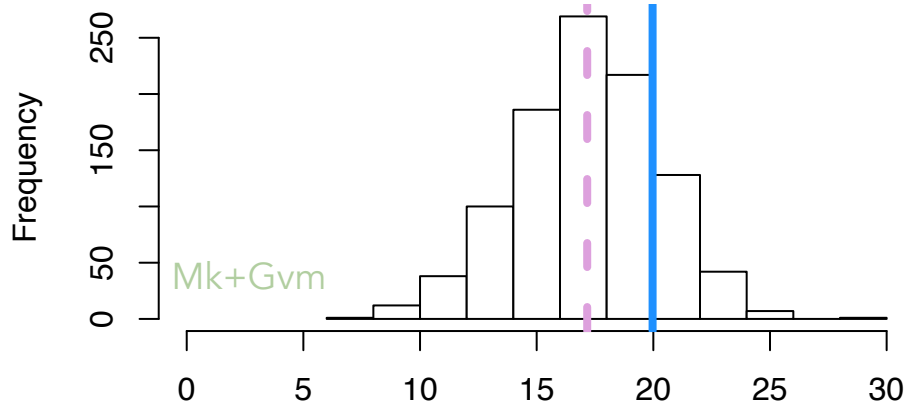
No models are adequate

Posterior Predictive P-values



Histograms showing the range of RF values for all the simulated data

Are these values significantly different from each other?



We will also calculate the P-values in R (look at the midpoint value)

Exercise 2:
Use model adequacy to determine the absolute fit of a model