Change-point Detection on a Tree to Study Evolutionary Adaptation from Present-day Species

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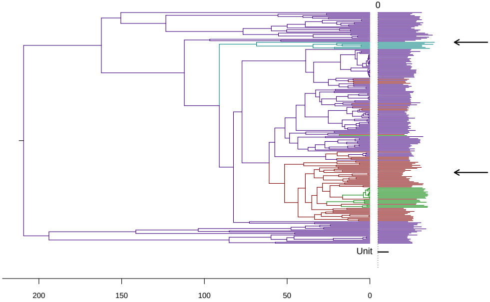
11 April 2016

Introduction



Turtles phylogenetic tree with habitats.
(Jaffe et al., 2011).

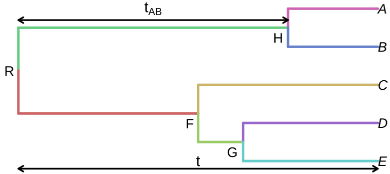
Dermochelys Coriacea

Homopus Areolatus

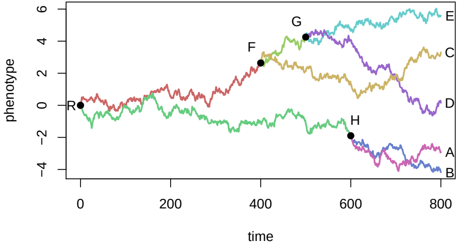
- How can we explain the diversity, while accounting for the phylogenetic correlations ?
- Modelling: a shifted stochastic process on the phylogeny.

Outline

Stochastic Process on a Tree (Felsenstein, 1985)



Only *tip* values are observed

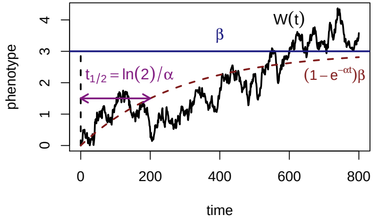


Brownian Motion:

$$\mathbb{V}\mathrm{ar}\,[A \mid R] = \sigma^2 t$$

$$\mathbb{C}\mathrm{ov}\,[A; B \mid R] = \sigma^2 t_{AB}$$

OU Modeling (Hansen, 1997)



$$dW(t) = \alpha[\beta(t) - W(t)]dt + \sigma dB(t)$$

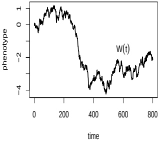
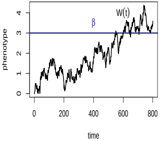
Deterministic part :

- $\beta(t)$: primary optimum, mechanistically defined.
- $\ln(2)/\alpha$: phylogenetic half live.

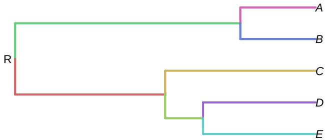
Stochastic part :

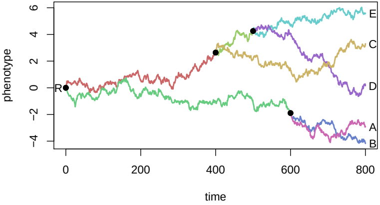
- $W(t)$: actual optimum (trait value).
- $\sigma dB(t)$ Brownian fluctuations.

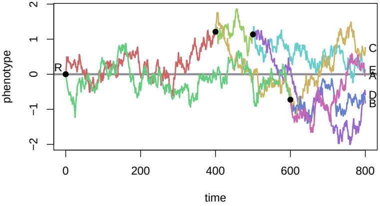
BM vs OU

	Equation	Stationary State	Variance
	$dW(t) = \sigma dB(t)$	None.	$\sigma_{ij} = \sigma^2 t_{ij}$
	$dW(t) = \sigma dB(t)$ $+\alpha[\beta(t) - W(t)]dt$	$\begin{cases} \mu = \beta_0 \\ \gamma^2 = \dfrac{\sigma^2}{2\alpha} \end{cases}$	$\sigma_{ij} = \gamma^2 e^{-\alpha(t_i+t_j)}$ $\times (e^{2\alpha t_{ij}} - 1)$

Shifts







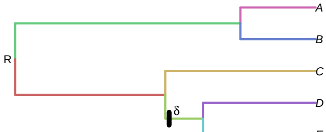
BM Shifts in the **mean**:

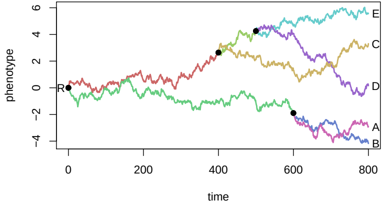
$$m_{\text{child}} = m_{\text{parent}} + \delta$$

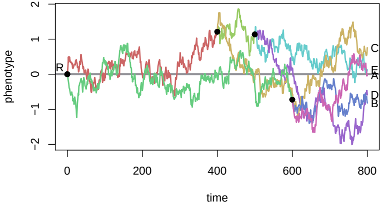
OU Shifts in the **optimal value**:

$$\beta_{\text{child}} = \beta_{\text{parent}} + \delta$$

Shifts







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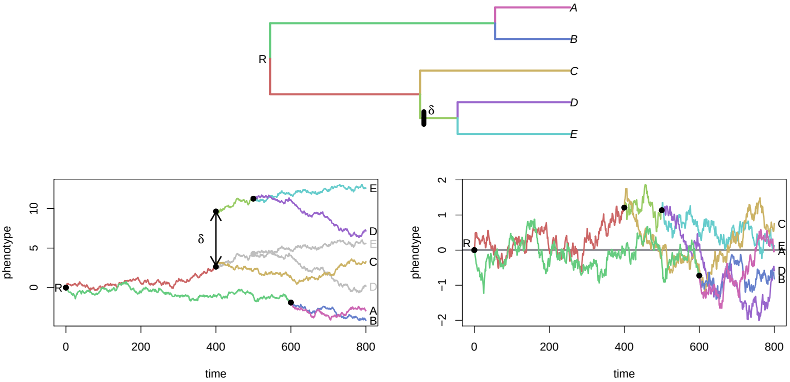

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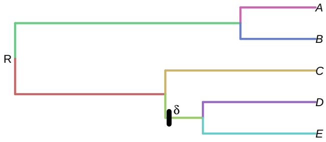

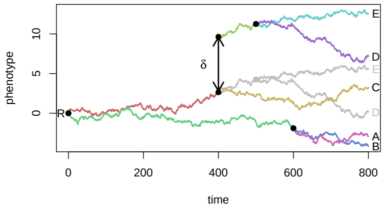

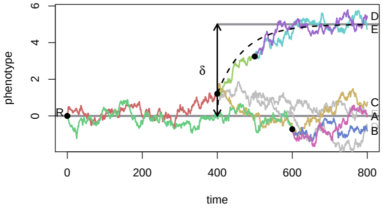

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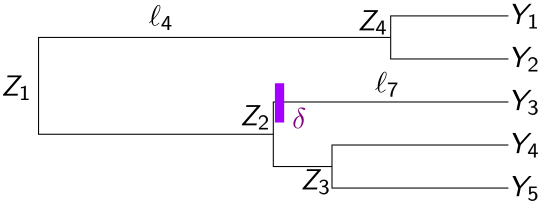
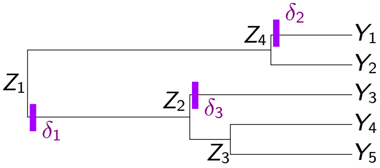
Incomplete Data Model



$$\text{BM}\quad Z_4|Z_1 \sim \mathcal{N}\left(Z_1, \sigma^2 \ell_4\right)$$
$$Y_3|Z_2 \sim \mathcal{N}\left(Z_2 + \delta, \sigma^2 \ell_7\right)$$

$$\text{OU}\quad Z_4|Z_1 \sim \mathcal{N}\left(Z_1 e^{-\alpha\ell_4} + (1 - e^{-\alpha\ell_4})\ \beta_{Z_1}, \frac{\sigma^2}{2\alpha}(1 - e^{-2\alpha\ell_4})\right)$$
$$Y_3|Z_2 \sim \mathcal{N}\left(Z_2 e^{-\alpha\ell_7} + (1 - e^{-\alpha\ell_7})(\beta_{Z_2} + \delta), \frac{\sigma^2}{2\alpha}(1 - e^{-2\alpha\ell_7})\right)$$

Linear Regression Model

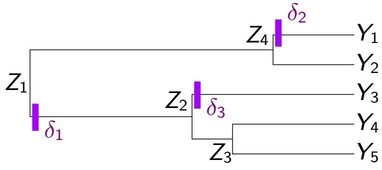


$$\Delta = \begin{pmatrix} \mu \\ \delta_1 \\ 0 \\ 0 \\ \delta_2 \\ 0 \\ \delta_3 \\ 0 \\ 0 \end{pmatrix} \qquad T\Delta = \begin{pmatrix} \mu + \delta_2 \\ \mu \\ \mu + \delta_1 + \delta_3 \\ \mu + \delta_1 \\ \mu + \delta_1 \end{pmatrix}$$

$$T = \begin{array}{c} \\ Y_1 \\ Y_2 \\ Y_3 \\ Y_4 \\ Y_5 \end{array} \begin{array}{c} \begin{array}{ccccccccc} Z_1 & Z_2 & Z_3 & Z_4 & Y_1 & Y_2 & Y_3 & Y_4 & Y_5 \end{array} \\ \begin{pmatrix} 1 & 0 & 0 & 1 & 1 & 0 & 0 & 0 & 0 \\ 1 & 0 & 0 & 1 & 0 & 1 & 0 & 0 & 0 \\ 1 & 1 & 0 & 0 & 0 & 0 & 1 & 0 & 0 \\ 1 & 1 & 1 & 0 & 0 & 0 & 0 & 1 & 0 \\ 1 & 1 & 1 & 0 & 0 & 0 & 0 & 0 & 1 \end{pmatrix} \end{array}$$

$BM: \quad Y = T\Delta^{BM} + E^{BM}$

Linear Regression Model



$$\Delta = \begin{pmatrix} \lambda \\ \delta_1 \\ 0 \\ 0 \\ \delta_2 \\ 0 \\ \delta_3 \\ 0 \\ 0 \end{pmatrix} \qquad TW(\alpha)\Delta = \begin{pmatrix} \lambda + w_5\delta_2 \\ \lambda \\ \lambda + w_2\delta_1 + w_7\delta_3 \\ \lambda + w_2\delta_1 \\ \lambda + w_2\delta_1 \end{pmatrix}$$

$$W(\alpha) = \mathrm{Diag}(1 - e^{-\alpha(h - t_{\mathrm{pa}(i)})}, 1 \leq i \leq m+n)$$

$$\lambda = \mu e^{-\alpha h} + \beta_0(1 - e^{-\alpha h})$$

$$BM: \quad Y = T\Delta^{BM} + E^{BM}$$

$$OU: \quad Y = TW(\alpha)\Delta^{OU} + E^{OU}$$

OU $\iff$ BM

Expectations

$$\mathbb{E}\left[Y \mid X_1 = \mu \right] = T \underbrace{W(\alpha)\Delta^{OU}}_{\Delta^{BM}}$$

Remark: $\mu^{BM} = \lambda^{OU} = \mu e^{-\alpha h} + \beta_0(1 - e^{-\alpha h})$

Variance

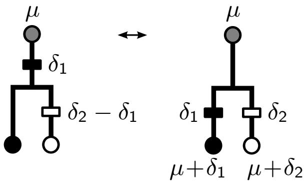
$$\mathbb{C}\mathrm{ov}\left[Y_i ; Y_j \mid X_1 = \mu \right] = \sigma^2 \times \underbrace{\frac{1}{2\alpha} e^{-2\alpha h}(e^{2\alpha t_{ij} - 1})}_{t'_{ij}}$$

OU $\iff$ BM on a re-scaled tree with $t' = e^{-2\alpha h}(e^{2\alpha t} - 1)$

Outline

Equivalencies

- Number of shifts K fixed, several equivalent solutions.



- Problem of over-parametrization: parsimonious configurations.

Equivalencies

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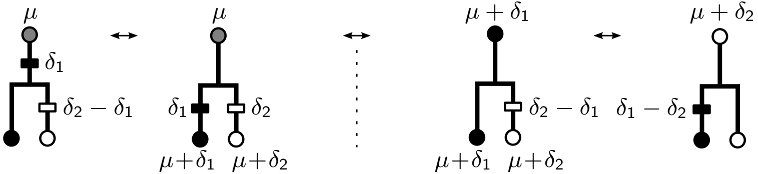
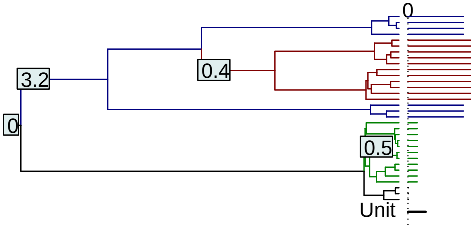


- Problem of over-parametrization: parsimonious configurations.

Process Induced Tip Coloring

Definition (Tips Coloring)

Two tips have the same color if they have the same mean under the process studied.



$$\color{green}{BM} \quad m_Y = T\Delta^{BM}$$

Parsimonious Solution : Definition

Definition (Parsimonious Allocation)

A coloring of the tips being given, a *parsimonious* allocation of the shifts is such that it has a minimum number of shifts.

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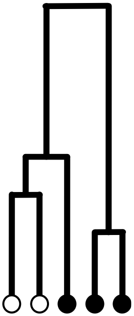

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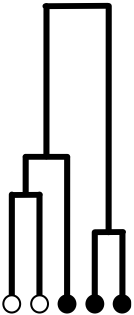

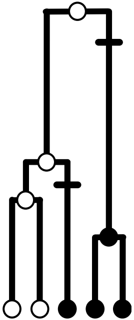

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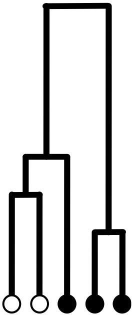
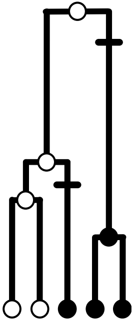
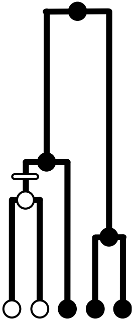




$\leq$



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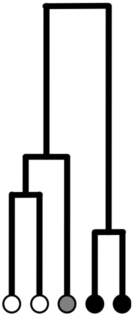

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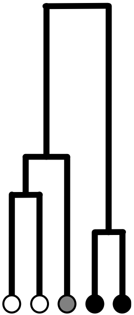

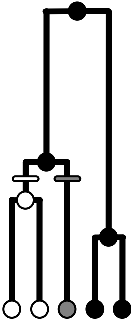

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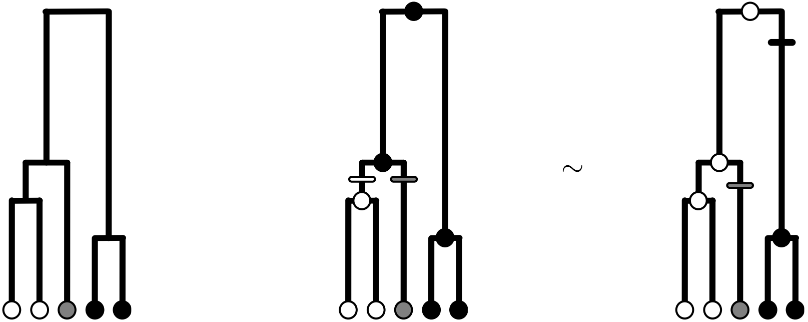


Equivalent Parsimonious Allocations

Definition (Equivalency)

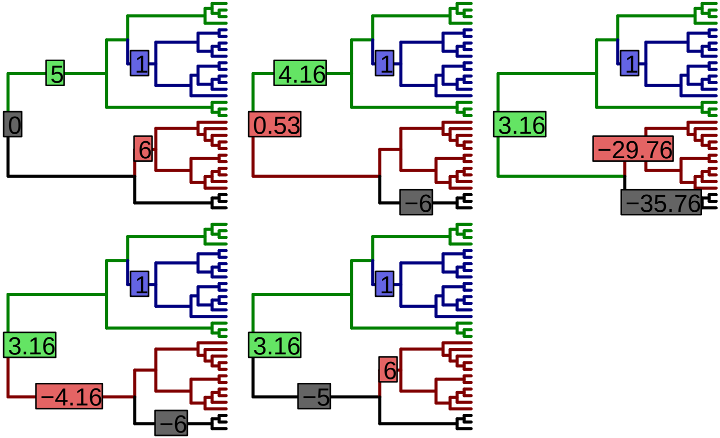
Two allocations are said to be *equivalent* (noted $\sim$) if they are both parsimonious and give the same colors at the tips.

Find one solution: Several existing Dynamic Programming algorithms (Fitch, Sankoff, see Felsenstein, 2004).

Enumerate all solutions: New recursive algorithm, adapted from previous ones (and implemented in R).

+

Equivalent Parsimonious Solutions for an OU Model.



Equivalent allocations and values of the shifts - OU.

Collection of Models

New Problem Number of Equivalence Classes: $\left|\mathcal{S}_K^{PI}\right|$?

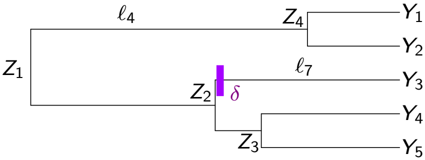
- $\left|\mathcal{S}_K^{PI}\right| \leq \binom{m+n-1}{K} = \binom{\#\text{ of edges}}{\#\text{ of shifts}}$
- A recursive algorithm to compute $\left|\mathcal{S}_K^{PI}\right|$ (implemented in R).

$\mapsto$ Generally dependent on the topology of the tree.

- Binary tree: $\left|\mathcal{S}_K^{PI}\right| = \binom{2n-2-K}{K} = \binom{\#\text{ of edges}-\#\text{ of shifts}}{\#\text{ of shifts}}$

Outline

EM Algorithm: number of shifts K fixed



$$Y_3 \mid Z_2 \sim \mathcal{N}\left(Z_2 + \delta,\ \ell_7 \sigma^2\right)$$

$$Z_4 \mid Z_1 \sim \mathcal{N}\left(Z_1,\ \ell_4 \sigma^2\right)$$

$$\log p_\theta(Y) = \mathbb{E}_\theta[\log p_\theta(Z, Y) \mid Y] - \mathbb{E}_\theta[\log p_\theta(Z) \mid Y]$$

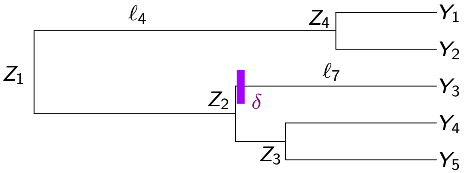
$$p_\theta(Z, Y) = p_\theta(Z_1) \prod_{1<j\leq m} p_\theta(Z_j | Z_{\text{parent}(j)}) \prod_{1\leq i\leq n} p_\theta(Y_i | Z_{\text{parent}(i)})$$

EM Algorithm Maximize $\mathbb{E}_\theta[\log p_\theta(Z, Y) \mid Y]$

E step Given θ^h, compute $p_{\theta^h}(Z \mid Y)$

M step $\theta^{h+1} = \text{argmax}_\theta\, \mathbb{E}_{\theta^h}[\log p_\theta(Z, Y) \mid Y]$

E step



Compute the following quantities:

$$\mathbb{E}^{(h)}[Z_j \mid Y], \ \mathbb{V}\mathrm{ar}^{(h)}\left[Z_j \mid Y\right], \ \mathbb{C}\mathrm{ov}^{(h)}\left[Z_j, Z_{\mathrm{parent}(j)} \mid Y\right]$$

- Using Gaussian properties. Need to invert matrices: complexity in $O(n^3)$.
- Using Gaussian properties **and** the tree structure: "Upward-Downward" algorithm. Complexity in $O(n)$.

M Step

Maximize:

$$\mathbb{E}\left[\log p_\theta(X) \mid Y\right] = -\sum_{j=2}^{m+n} C_j(\alpha, \text{shifts}) + \mathcal{F}^{(h)}\left(\mu, \gamma^2, \sigma^2, \alpha\right)$$

- μ, γ^2, σ^2: simple maximization
- Discrete location of K shifts
  - $\mapsto$ Exact and fast for the BM
- α: numerical maximization and/or on a grid
  - $\mapsto$ Generalized EM

Initialization

Shifts : Lasso regression.

$$\hat{\Delta} = \underset{\Delta}{\operatorname{argmin}} \left\{ \| Y - TW(\alpha)\Delta \|^2_{\Sigma_{YY}^{-1}} + \lambda \|\Delta_{-1}\|_1 \right\}$$

- Initialize $\Sigma_{YY}(\alpha)$, then estimate Δ with a Gauss Lasso procedure, using a Cholesky decomposition.
- λ chosen to get K shifts.

The selection strength α : Initialization using couples of tips.

Model Selection on K

Assumption α fixed

$$Y = TW(\alpha)\Delta + \gamma E \qquad , \qquad E \sim \mathcal{N}(0, V(\alpha))$$

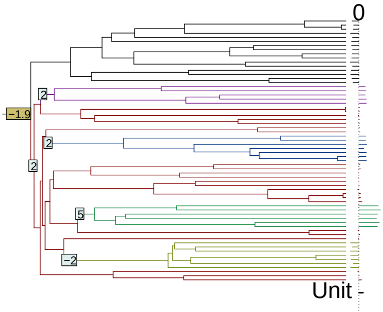
Models

$\eta \in \bigcup_{K=0}^{p-1} \mathcal{S}_K^{PI}$: Identifiable parcimonious allocations of shifts

EM Estimators

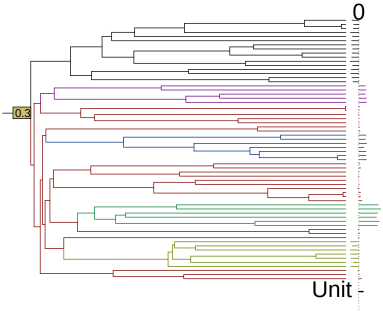
$$\hat{Y}_K = \underset{\eta \in \mathcal{S}_K^{PI}}{\operatorname{argmin}} \left\| Y - \hat{Y}_\eta \right\|_V^2$$

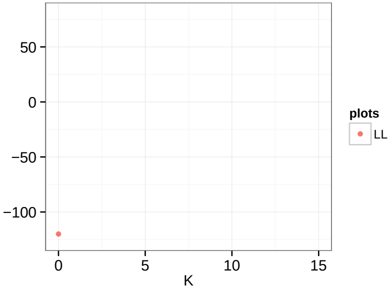
Model Selection on K



Simulated OU (α =3, γ^2 =0.1)

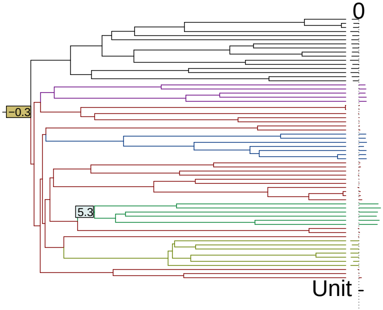
Model Selection on K

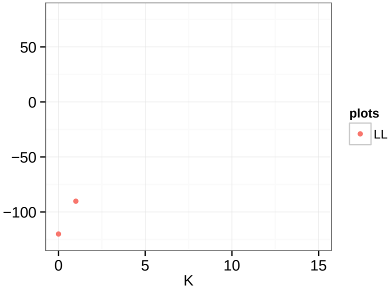




$$\hat{Y}_K = \underset{\eta \in \mathcal{S}_K^{PI}}{\operatorname{argmax}} -\frac{n}{2}\log\left(\frac{\left\|Y - \hat{Y}_\eta\right\|_V^2}{n}\right) \qquad LL = -\frac{n}{2}\log\left(\frac{\left\|Y - \hat{Y}_K\right\|_V^2}{n}\right)$$

Model Selection on K





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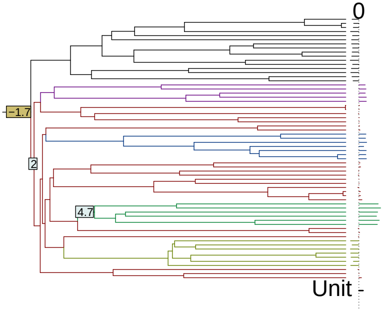
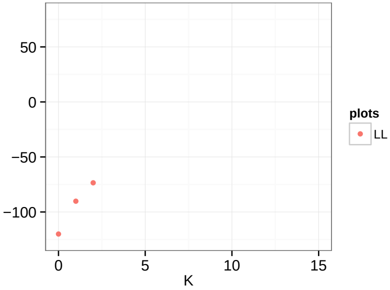
Model Selection on K

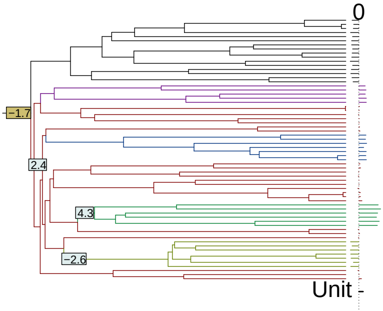


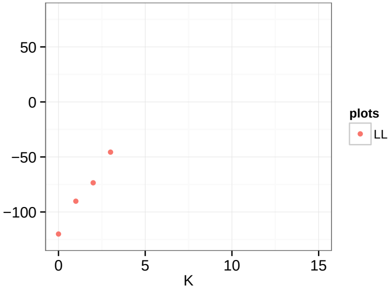


$$\hat{Y}_K = \underset{\eta \in \mathcal{S}_K^{PI}}{\operatorname{argmax}} -\frac{n}{2} \log\left(\frac{\left\|Y - \hat{Y}_\eta\right\|_V^2}{n}\right)$$

$$LL = -\frac{n}{2} \log\left(\frac{\left\|Y - \hat{Y}_K\right\|_V^2}{n}\right)$$

Model Selection on K





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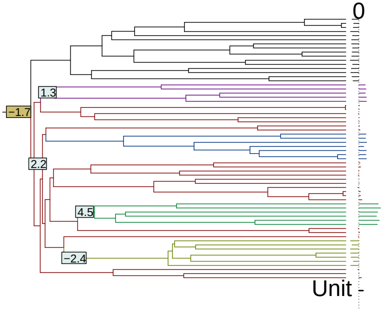
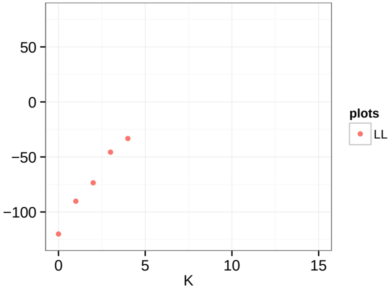
Model Selection on K





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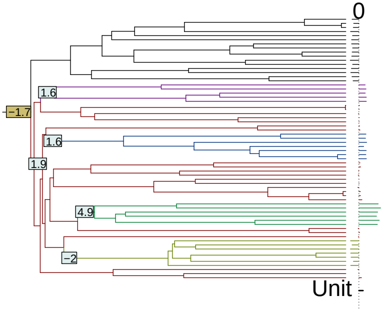
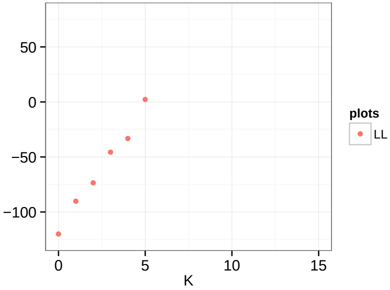
Model Selection on K





$$\hat{Y}_K = \underset{\eta \in \mathcal{S}_K^{PI}}{\operatorname{argmax}} -\frac{n}{2} \log \left(\frac{\left\| Y - \hat{Y}_\eta \right\|_V^2}{n} \right)$$

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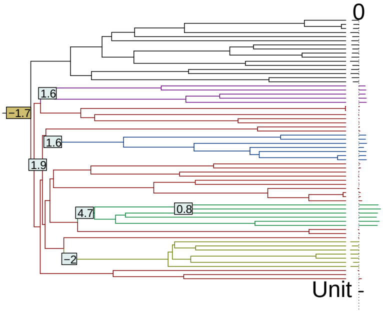
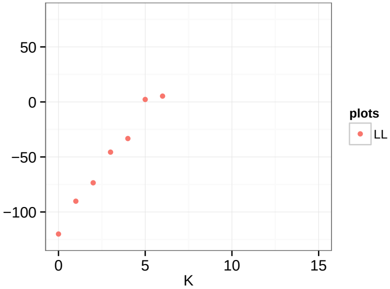
Model Selection on K





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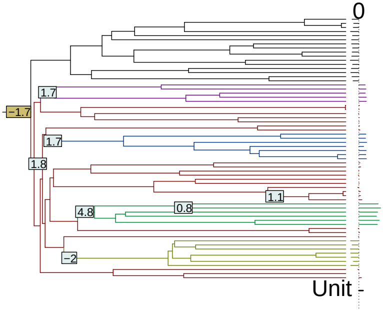
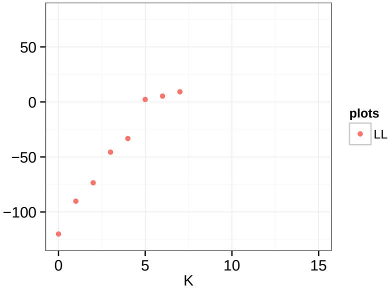
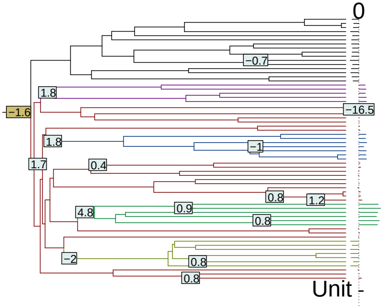
Model Selection on K

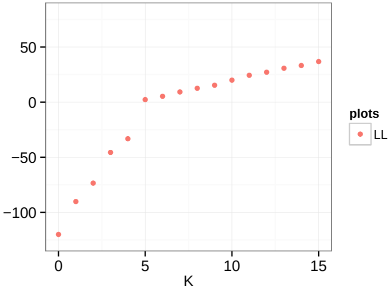




$$\hat{Y}_K = \underset{\eta \in \mathcal{S}_K^{PI}}{\operatorname{argmax}} -\frac{n}{2} \log\left(\frac{\left\|Y - \hat{Y}_\eta\right\|_V^2}{n}\right) \qquad LL = -\frac{n}{2} \log\left(\frac{\left\|Y - \hat{Y}_K\right\|_V^2}{n}\right)$$

Model Selection on K

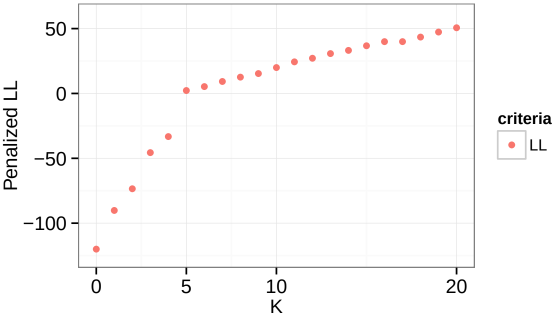




$$\hat{Y}_K = \underset{\eta \in \mathcal{S}_K^{PI}}{\operatorname{argmax}} -\frac{n}{2} \log\left(\frac{\left\| Y - \hat{Y}_\eta \right\|_V^2}{n}\right) \qquad LL = -\frac{n}{2} \log\left(\frac{\left\| Y - \hat{Y}_K \right\|_V^2}{n}\right)$$

Model Selection: Penalized Likelihood

Idea $$\hat{K} = -\underset{0 \leq K \leq p-1}{\operatorname{argmin}} \frac{n}{2} \log\left(\frac{\left\|Y - \hat{Y}_K\right\|_V^2}{n}\right) - \frac{1}{2}\operatorname{pen}'(K)$$



Model Selection: Penalized Likelihood

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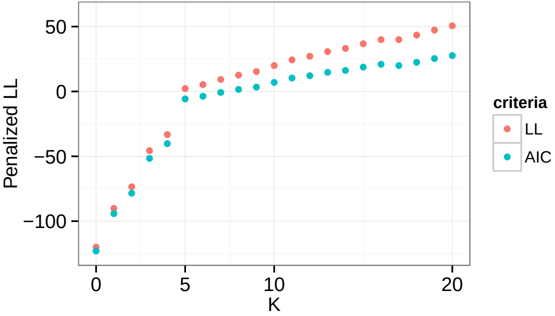
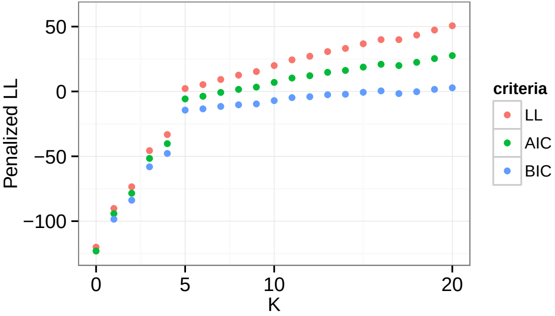


Penalties:

AIC $K + 3$

Model Selection: Penalized Likelihood

Idea $$\hat{K} = - \underset{0 \leq K \leq p-1}{\operatorname{argmin}} \frac{n}{2} \log \left(\frac{\left\| Y - \hat{Y}_K \right\|_V^2}{n} \right) - \frac{1}{2} \operatorname{pen}'(K)$$



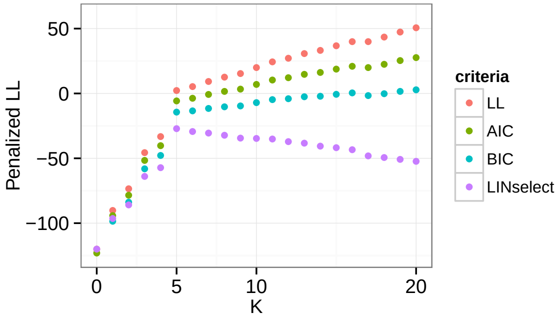
Penalties:

AIC $K + 3$

BIC $\frac{1}{2}(K+3)\log(n)$

Model Selection: Penalized Likelihood

Idea $$\hat{K} = -\underset{0 \leq K \leq p-1}{\operatorname{argmin}} \frac{n}{2} \log \left(\frac{\left\| Y - \hat{Y}_K \right\|_V^2}{n} \right) - \frac{1}{2} \operatorname{pen}'(K)$$



Penalties:

AIC $K + 3$

BIC $\frac{1}{2}(K+3)\log(n)$

LINselect $\operatorname{pen}(n, K, |\mathcal{S}_K^{PI}|)$

Model Selection on K: LINselect

Goal

$$\hat{K} = \underset{0 \leq K \leq p-1}{\operatorname{argmin}} \left\| Y - \hat{Y}_K \right\|_V^2 \left(1 + \frac{\operatorname{pen}(K)}{n - K - 1} \right)$$

Oracle

$$\inf_{\eta \in \bigcup_{K=0}^{p-1} \mathcal{S}_K^{PI}} \left\| \mathbb{E}\left[Y \right] - Y_\eta^* \right\|_V^2$$

Definition (Baraud et al. (2009))

Let D, $N > 0$, and $X_D \sim \chi^2(D)$, $X_N \sim \chi^2(N)$, $X_D \perp X_N$.

$$\operatorname{Dkhi}[D, N, x] = \frac{1}{\mathbb{E}\left[X_D\right]} \mathbb{E}\left[\left(X_D - x \frac{X_N}{N} \right)_+ \right], \quad \forall x > 0$$

$$\operatorname{Dkhi}[D, N, \operatorname{EDkhi}[D, N, q]] = q, \quad \forall 0 < q \leq 1$$

Proposition: LINselect Penalty

Proposition (Form of the Penalty and guarantees (α known))

Under our setting: $Y = TW(\alpha)\Delta + \gamma E$ *with* $E \sim \mathcal{N}(0, V)$*, define the penalty:*

$$\text{pen}(K) = A\frac{n-K-1}{n-K-2}\,\text{EDkhi}\left[K+2, n-K-2, exp\left(-\log\left|\mathcal{S}_K^{PI}\right| - 2\log(K+2)\right)\right]$$

If $\kappa < 1$*, and* $p \leq \min\left(\frac{\kappa n}{2+\log(2)+\log(n)}, n-7\right)$*, we get:*

$$\mathbb{E}\left[\frac{\left\|\mathbb{E}\left[Y\right] - \hat{Y}_{\hat{K}}\right\|_V^2}{\gamma^2}\right] \leq C(A,\kappa)\inf_{\eta\in\mathcal{M}}\left\{\frac{\left\|\mathbb{E}\left[Y\right] - Y_\eta^*\right\|_V^2}{\gamma^2} + (K_\eta + 2)\left(3 + \log(n)\right)\right\}$$

with $C(A,\kappa)$ *a constant depending on* A *and* κ *only.*

Based on Baraud et al. (2009) +

LINselect Model Selection: Important Points

Based on Baraud, Giraud, and Huet (2009)

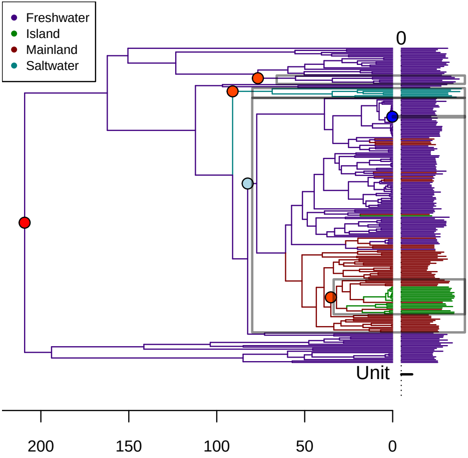
- Non-asymptotic bound.
- Unknown variance.
- No constant to be calibrated.

Novelties

- Non iid variance.
- Penalty depends on the tree topology (through $\left|\mathcal{S}_K^{PI}\right|$).

Outline

Turtles Dataset

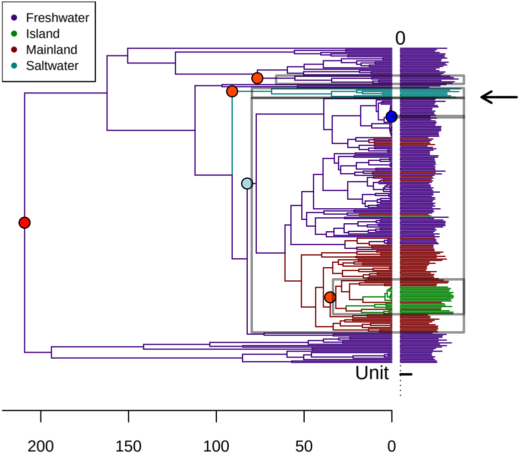


Colors: habitats.
Boxes: selected EM regimes.

	Habitat	EM
No. of shifts	16	5
No. of regimes	4	6
lnL	-133.86	-97.59
$\ln 2/\alpha$ (%)	7.44	5.43
$\sigma^2/2\alpha$	0.33	0.22
CPU t (min)	65.25	134.49

(Jaffe et al., 2011)

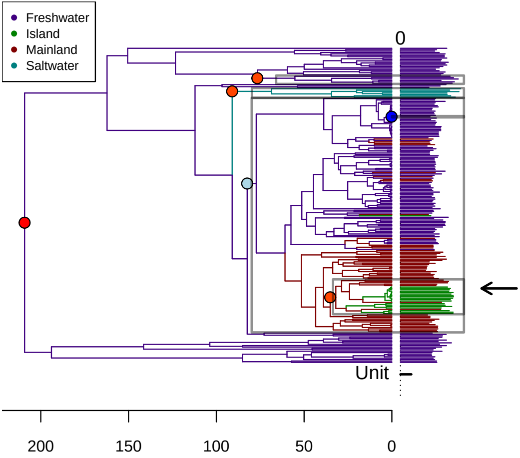
Turtles Dataset



Colors: habitats.
Boxes: selected EM regimes.

Chelonia mydas

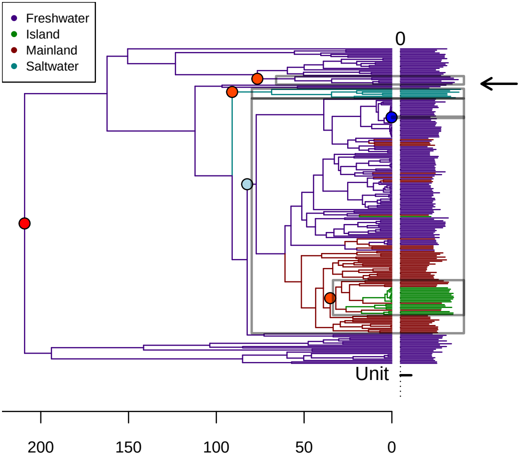
Turtles Dataset



Colors: habitats.
Boxes: selected EM regimes.

Geochelone nigra abingdoni

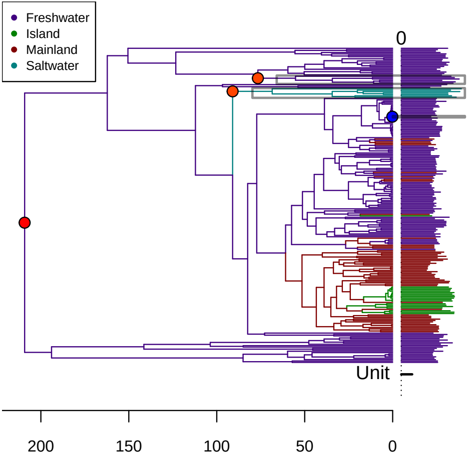
Turtles Dataset



Colors: habitats.
Boxes: selected EM regimes.

Chitra indica

Turtles Dataset

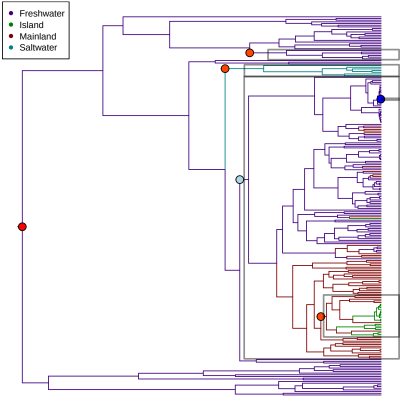


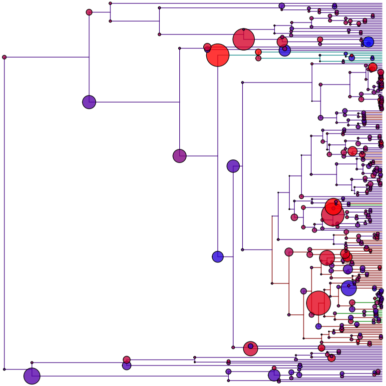
Colors: habitats.
Boxes: selected EM regimes.

	Habitat	EM(3)
No. of shifts	16	3
No. of regimes	4	4
lnL	-133.86	-113.73
$\ln 2/\alpha$ (%)	7.44	9.20
$\sigma^2/2\alpha$	0.33	0.30
CPU t (min)	65.25	134.49

(Jaffe et al., 2011)

Comparison with Bayou





Colors: habitats.
Boxes: selected EM regimes.

Colors: habitats.
Circles: posterior probability of shift.

Summary

	EM	Habitat	bayou
No. of shifts	5	16	17
No. of regimes	6	4	18
lnL	-97.59	-133.86	-91.54
MlnL	NaN	NaN	-149.09
$\ln 2/\alpha$ (%)	5.43	7.44	1.90
γ^2	0.22	0.33	0.16
CPU time (min)	134.49	65.25	136.81

Outline

BM Model

Data *n vectors* of p traits at the tips: $\mathbf{Y}_i = \begin{pmatrix} Y_{i1} \\ \vdots \\ Y_{ip} \end{pmatrix}$

SDE $d\mathbf{W}(t) = \mathbf{\Sigma} d\mathbf{B}_t$, rate matrix $\mathbf{R} = \mathbf{\Sigma}\mathbf{\Sigma}^T$ $(p \times p)$

Covariances $\mathbb{C}\text{ov}\left[Y_{il}; Y_{jq}\right] = t_{ij} R_{lq}$ for i, j tips, and l, q characters

$$\mathbb{V}\text{ar}\left[\text{vec}(\mathbf{Y})\right] = \mathbf{C}_n \otimes \mathbf{R}$$

Shifts K shifts $\boldsymbol{\delta}_1, \cdots, \boldsymbol{\delta}_K$ *vectors* size p

$\mapsto$ All the characters shift at the same time

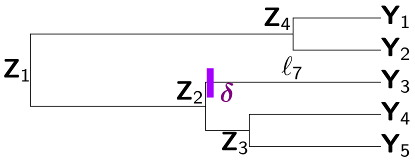
BM Model

Linear Model Representation

$$\mathrm{vec}(\mathbf{Y}) = \mathrm{vec}(\mathbf{\Delta T}^T) + \mathbf{E} \text{ with } \mathbf{E} \sim \mathcal{N}(0, \mathbf{V} = \mathbf{C}_n \otimes \mathbf{R})$$

Incomplete Data Representation

$$\mathbf{Y}_3 \mid \mathbf{Z}_2 \sim \mathcal{N}\left(\mathbf{Z}_2 + \delta,\ \ell_7 \mathbf{R}\right)$$



OU Model: General Case

Data *n vectors* of p traits at the tips: $\mathbf{Y}_i = \begin{pmatrix} Y_{i1} \\ \vdots \\ Y_{ip} \end{pmatrix}$

SDE $\mathbf{A}$ $(p \times p)$ "selection strength"

$$d\mathbf{W}(t) = -\mathbf{A}(\mathbf{W}(t) - \beta(t))dt + \mathbf{\Sigma} d\mathbf{B}_t$$

Covariances

$$\mathbb{C}\mathrm{ov}\left[\mathbf{X}_i ; \mathbf{X}_j\right] = e^{-\mathbf{A}t_i}\mathbf{\Gamma}e^{-\mathbf{A}^T t_j} + e^{-\mathbf{A}(t_i - t_{ij})}\left(\int_0^{t_{ij}} e^{-\mathbf{A}v}\mathbf{\Sigma}\mathbf{\Sigma}^T e^{-\mathbf{A}^T v} dv\right) e^{-\mathbf{A}^T(t_j - t_{ij})}$$

Shifts K shifts $\boldsymbol{\delta}_1, \cdots, \boldsymbol{\delta}_K$ *vectors* size p

$\mapsto$ On the optimal values

OU Model: $\mathbf{A}$ scalar

Assumption $\mathbf{A} = \alpha \mathbf{I}_p$ "scalar"

Stationnary State $\mathbf{S} = \frac{1}{2\alpha}\mathbf{R}$

Fixed Root For i, j tips and l, q characters:

$$\mathbb{C}\text{ov}\left[Y_{il}; Y_{jq}\right] = \frac{1}{2\alpha} e^{-2\alpha h}\left(e^{2\alpha t_{ij}} - 1\right) R_{lq}$$

$\mapsto$ Can be reduced to a BM on a re-scaled tree

EM algorithm

BM Natural generalization of the univariate case.

OU M step intractable in general.

Incomplete Data Model: Can readily handle missing data.

Model Selection

- Previous criterion cannot be applied
- Solution: "Slope Heuristic"-based method
  - Massart (2007)
    - oracle inequality with known variance
    - penalty up to a multiplicative constant
  - Baudry et al. (2012)
    - Slope-heuristic method to calibrate the constant
    - Implemented in capushe (Brault et al., 2012)

Model Selection: Toy Example

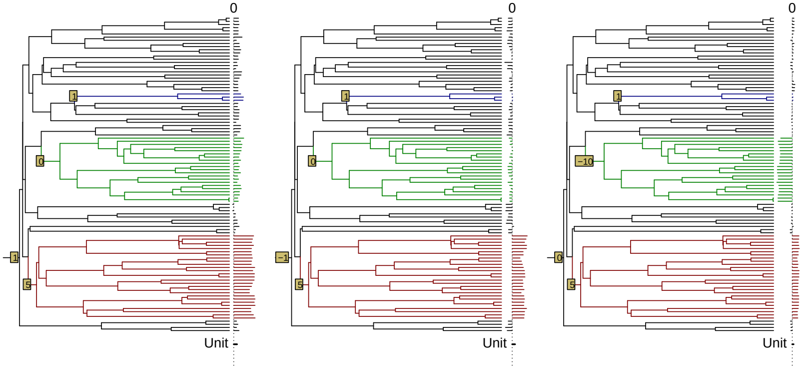


Figure: *Simulated Process.*

Model Selection: Toy Example

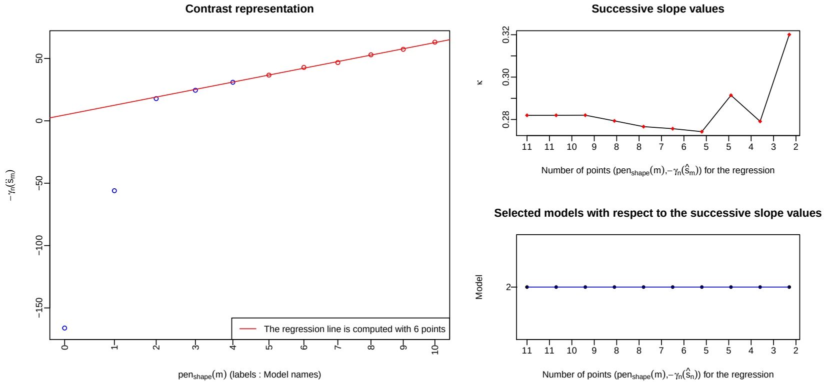


Figure: capushe *output for penalized log-likelihood.*

Model Selection: Toy Example

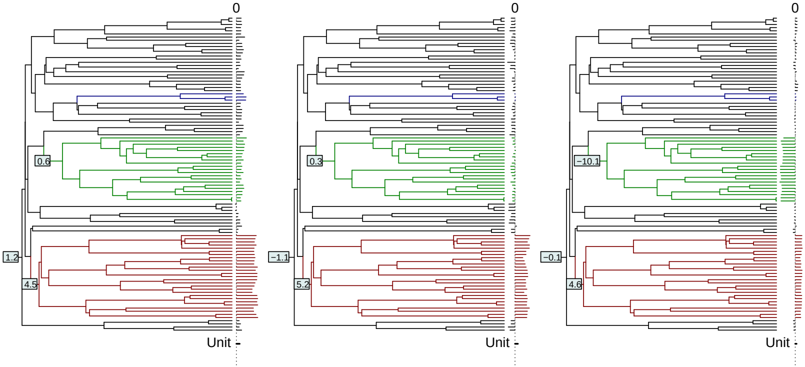


Figure: Reconstructed Process.

Conclusion and Perspectives

A general inference framework for trait evolution models.

Conclusions
- Some problems of identifiability arise.
- An EM can be written to maximize likelihood.
- Adaptation of model selection results to non-iid framework.

R codes Available on GitHub:
https://github.com/pbastide/Phylogenetic-EM

Perspectives
- Multivariate traits.
- Deal with uncertainty (tree, data).
- Use fossil records.

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Thank you for listening

Appendices

6. Inference
   - Lasso Initialization and Cholesky decomposition
   - Upward-Downward Algorithm
   - Model Selection
   - Segmentation Algorithms
   - Multivariate M
7. Identifiability Issues
   - Cardinal of Equivalence Classes
   - Number of Tree Compatible Clustering
8. Simulations Results

Cholesky Decomposition

The problem is:

$$\hat{\Delta} = \underset{\Delta}{\operatorname{argmin}} \left\{ \|Y - R\Delta\|^2_{\Sigma_{YY}} + \lambda \left|\Delta_{-1}\right|_1 \right\}$$

Cholesky decomposition of Σ_{YY}:

$$\Sigma_{YY} = LL^T \ , \ L \text{ a lower triangular matrix}$$

Then:

$$\|Y - R\Delta\|^2_{\Sigma_{YY}} = \left\| L^{-1}Y - L^{-1}R\Delta \right\|^2$$

And if $Y' = L^{-1}Y$ and $R' = L^{-1}R$, the problem becomes:

$$\hat{\Delta} = \underset{\Delta}{\operatorname{argmin}} \left\{ \left\| Y' - R'\Delta \right\|^2 + \lambda \left|\Delta_{-1}\right|_1 \right\}$$

Gauss Lasso

Let $\hat{m}_\lambda$ be the set of selected variables (including the root). Then:

$$\hat{\Delta}^{\text{Gauss}} = \Pi_{\hat{F}_\lambda}(Y') \text{ with } \hat{F}_\lambda = \text{Span}\{R'_j : j \in \hat{m}_\lambda\}$$

back

Goal and Notations

Data A process on a tree with the following structure:

$$\forall j > 1, \quad X_j | X_{\mathrm{pa}(j)} \sim \mathcal{N}\left(m_j(X_{\mathrm{pa}(j)}) = q_j X_{\mathrm{pa}(j)} + r_j, \sigma_j^2\right)$$

$$\text{BM:} \begin{cases} q_j = 1 \\ r_j = \sum_k \mathbb{I}\{\tau_k = b_j\}\delta_k \\ \sigma_j^2 = \ell_j \sigma^2 \end{cases} \qquad \text{OU:} \begin{cases} q_j = e^{-\alpha \ell_j} \\ r_j = \beta^{\mathrm{pa}(j)}(1 - e^{-\alpha \ell_j}) + \sum_k \mathbb{I}\{\tau_k = b_j\}\delta_k \left(1 - e^{-\alpha(1-\nu_k)\ell_j}\right) \\ \sigma_j^2 = \dfrac{\sigma^2}{2\alpha}(1 - e^{-2\alpha \ell_j}) \end{cases}$$

Goal Compute the following quantities, at every node j:

$$\mathbb{V}\mathrm{ar}^{(h)}\left[Z_j \mid Y\right], \mathbb{C}\mathrm{ov}^{(h)}\left[Z_j, Z_{\mathrm{pa}(j)} \mid Y\right], \mathbb{E}^{(h)}[Z_j \mid Y]$$

Upward

Goal Compute for a vector of tips, given their common ancestor:

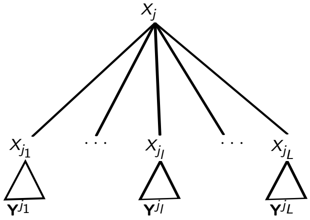
$$f_{\mathbf{Y}^j|X_j}(\mathbf{Y}^j; a) = A_j(\mathbf{Y}^j)\Phi_{M_j(\mathbf{Y}^j), S_j^2(\mathbf{Y}^j)}(a)$$

Initialization For tips: $f_{Y_i|Y_i}(Y_i; a) = \Phi_{Y_i,0}(a)$

Propagation

$$f_{\mathbf{Y}^j|X_j}(\mathbf{Y}^j; a) = \prod_{l=1}^{L} f_{\mathbf{Y}^{j_l}|X_j}(\mathbf{Y}^{j_l}; a)$$

$$f_{\mathbf{Y}^{j_l}|X_j}(\mathbf{Y}^{j_l}; a) = \int_{\mathbb{R}} f_{\mathbf{Y}^{j_l}|X_{j_l}}(\mathbf{Y}^{j_l}; b) f_{X_{j_l}|X_j}(b; a) db$$



Root Node and Likelihood At the root:

$$f_{X_1|\mathbf{Y}}(a; \mathbf{Y}) \propto f_{\mathbf{Y}|X_1}(\mathbf{Y}; a) f_{X_1}(a)$$

$$\begin{cases} \mathbb{V}\text{ar}\left[X_1 \mid \mathbf{Y}\right] = \left(\dfrac{1}{\gamma^2} + \dfrac{1}{S_1^2(\mathbf{Y})}\right)^{-1} \\ \mathbb{E}\left[X_1 \mid \mathbf{Y}\right] = \mathbb{V}\text{ar}\left[X_1 \mid \mathbf{Y}\right]\left(\dfrac{\mu}{\gamma^2} + \dfrac{M_1(\mathbf{Y})}{S_1^2(\mathbf{Y})}\right) \end{cases}$$

Downward

Compute $E_j = \mathbb{E}\left[X_j \mid \mathbf{Y}\right]$, $V_j^2 = \mathbb{V}\mathrm{ar}\left[X_j \mid \mathbf{Y}\right]$, $C_{j,\mathrm{pa}(j)}^2 = \mathbb{C}\mathrm{ov}\left[X_j ; X_{\mathrm{pa}(j)} \mid \mathbf{Y}\right]$

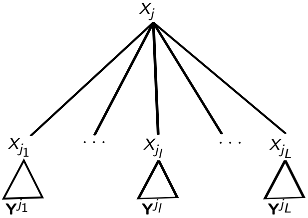
Initialization Last step of Upward.

Propagation

$$f_{X_{\mathrm{pa}(j)}, X_j | \mathbf{Y}}(a, b; \mathbf{Y}) = f_{X_{\mathrm{pa}(j)} | \mathbf{Y}}(a; \mathbf{Y}) f_{X_j | X_{\mathrm{pa}(j)}, \mathbf{Y}}(b; a, \mathbf{Y})$$

$$f_{X_j | X_{\mathrm{pa}(j)}, \mathbf{Y}}(b; a, \mathbf{Y}) = f_{X_j | X_{\mathrm{pa}(j)}, \mathbf{Y}^j}(b; a, \mathbf{Y}^j)$$

$$\propto f_{X_j | X_{\mathrm{pa}(j)}}(b; a) f_{\mathbf{Y}^j | X_j}(\mathbf{Y}^j; b)$$



Formulas

Upward

$$\begin{cases} S_j^2(\mathbf{Y}^j) = \left(\sum_{l=1}^{L} \dfrac{q_{j_l}^2}{S_{j_l}^2(\mathbf{Y}^{j_l}) + \sigma_{j_l}^2} \right)^{-1} \\ M_j(\mathbf{Y}^j) = S_j^2(\mathbf{Y}^j) \sum_{l=1}^{L} q_{j_l} \dfrac{M_{j_l}(\mathbf{Y}^{j_l}) - r_{j_l}}{S_{j_l}^2(\mathbf{Y}^{j_l}) + \sigma_{j_l}^2} \end{cases}$$

Downward

$$\begin{cases} C_{j,\mathrm{pa}(j)}^2 = q_j \dfrac{S_j^2(\mathbf{Y}^j)}{S_j^2(\mathbf{Y}^j) + \sigma_j^2} V_{\mathrm{pa}(j)}^2 \\ E_j = \dfrac{S_j^2(\mathbf{Y}^j)(q_j E_{\mathrm{pa}(j)} + r_j) + \sigma_j^2 M_j(\mathbf{Y}^j)}{S_j^2(\mathbf{Y}^j) + \sigma_j^2} \\ V_j^2 = \dfrac{S_j^2(\mathbf{Y}^j)}{S_j^2(\mathbf{Y}^j) + \sigma_j^2} \left(\sigma_j^2 + p_j^2 \dfrac{S_j^2(\mathbf{Y}^j)}{S_j^2(\mathbf{Y}^j) + \sigma_j^2} V_{\mathrm{pa}(j)}^2 \right) \end{cases}$$

back

Model Selection with Unknown Variance

Theorem (Baraud et al. (2009))

Under the following setting:

$$Y' = \mathbb{E}\left[Y'\right] + \gamma E' \quad \textit{with} \quad E' \sim \mathcal{N}(0, I_n) \quad \textit{and} \quad \mathcal{S}' = \{S'_\eta, \eta \in \mathcal{M}\}$$

If $D_\eta = \mathrm{Dim}(S'_\eta)$, $N_\eta = n - D_\eta \geq 7$, $\max(L_\eta, D_\eta) \leq \kappa n$, *with* $\kappa < 1$, *and:*

$$\Omega' = \sum_{\eta \in \mathcal{M}} (D_\eta + 1) e^{-L_\eta} < +\infty$$

$$\textit{If:} \quad \hat{\eta} = \underset{\eta \in \mathcal{M}}{\mathrm{argmin}} \left\| Y' - \hat{Y}'_\eta \right\|^2 \left(1 + \frac{\mathrm{pen}(\eta)}{N_\eta}\right)$$

$$\textit{with:} \quad \mathrm{pen}(\eta) = \mathrm{pen}_{A,\mathcal{L}}(\eta) = A \frac{N_\eta}{N_\eta - 1} \mathrm{EDkhi}[D_\eta + 1, N_\eta - 1, e^{-L_\eta}] \quad , \quad A > 1$$

$$\textit{Then:}\ \mathbb{E}\left[\frac{\left\|\mathbb{E}\left[Y'\right] - \hat{Y}'_{\hat{\eta}}\right\|^2}{\gamma^2}\right] \leq C(A, \kappa) \left[\inf_{\eta \in \mathcal{M}} \left\{ \frac{\left\|\mathbb{E}\left[Y'\right] - Y'_\eta\right\|^2}{\gamma^2} + \max(L_\eta, D_\eta) \right\} + \Omega'\right]$$

IID Framework ($\alpha = 0$)

$$\text{Assume} \quad K_\eta = D_\eta - 1 \leq p - 1 \leq n - 8, \quad \forall \eta \in \mathcal{M}$$

Then:

$$\begin{aligned}
\Omega' &= \sum_{\eta \in \mathcal{M}} (D_\eta + 1) e^{-L_\eta} = \sum_{\eta \in \mathcal{M}} (K_\eta + 2) e^{-L_\eta} \\
&= \sum_{K=0}^{p-1} \left| \mathcal{S}_K^{PI} \right| (K+2) e^{-L_K} = \sum_{K=0}^{p-1} \left| \mathcal{S}_K^{PI} \right| (K+2) e^{-(\log\left|\mathcal{S}_K^{PI}\right| + 2\log(K+2))} \\
&= \sum_{K=0}^{p-1} \frac{1}{K+2} \leq \log(p) \leq \log(n)
\end{aligned}$$

And:

$$L_K \leq \log \binom{n+m-1}{K} + 2\log(K+2) \leq K\log(n+m-1) + 2(K+1) \leq p(2 + \log(2n-2))$$

Hence, if $p \leq \min\left(\frac{\kappa n}{2+\log(2)+\log(n)}, n-7\right)$, then $\max(L_\eta, D_\eta) \leq \kappa n$ for any $\eta \in \mathcal{M}$.

Non-IID Framework ($\alpha \neq 0$)

Cholesky decomposition: $V = LL^T \quad Y' = L^{-1}Y \quad s' = L^{-1}s \quad E' = L^{-1}E$

$$Y' = \mathbb{E}\left[Y'\right] + \gamma E', \text{ with: } E' \sim \mathcal{N}(0, I_n)$$

$$S'_\eta = L^{-1}S_\eta, \quad \hat{Y}'_\eta = \mathrm{Proj}_{S'_\eta} Y' = \underset{a' \in S'_\eta}{\mathrm{argmin}} \left\| Y - La' \right\|_V^2 = L^{-1}\hat{Y}_\eta$$

$$\left\| \mathbb{E}\left[Y\right] - \hat{Y}_{\hat{\eta}} \right\|_V^2 = \left\| \mathbb{E}\left[Y'\right] - \hat{Y}'_{\hat{\eta}} \right\|^2, \quad \left\| Y - \hat{Y}_\eta \right\|_V^2 = \left\| Y' - \hat{Y}'_\eta \right\|^2$$

$$\mathrm{Crit}_{MC}(\eta) = \left\| Y' - \hat{Y}'_\eta \right\|^2 \left(1 + \frac{\mathrm{pen}_{A,\mathcal{L}}(\eta)}{N_\eta} \right) = \left\| Y - \hat{Y}_\eta \right\|_V^2 \left(1 + \frac{\mathrm{pen}_{A,\mathcal{L}}(\eta)}{N_\eta} \right)$$

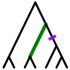
back

M Step: Segmentation

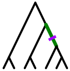
$$C_j(\alpha, \tau, \delta) = \sigma_j^{-2}\left(\mathbb{E}\left[X_j \mid Y\right] - q_j\mathbb{E}\left[X_{\mathrm{pa}(j)} \mid Y\right] - r_j - s_j\sum_k \mathbb{I}\{\tau_k = b_j\}\delta_k\right)^2$$

BM : $r_j = 0$, each cost is independent.

$$C_j^0(\alpha) = \sigma_j^{-2}\left(\mathbb{E}\left[X_j \mid Y\right] - q_j\mathbb{E}\left[X_{\mathrm{pa}(j)} \mid Y\right]\right)^2$$



$$C_j^1(\alpha, \tau, \delta) = \sigma_j^{-2}\left(\mathbb{E}\left[X_j \mid Y\right] - q_j\mathbb{E}\left[X_{\mathrm{pa}(j)} \mid Y\right] - s_j\sum_k \mathbb{I}\{\tau_k = b_j\}\delta_k\right)^2$$



Algorithm:

1. Find the K branches $j_1, \dots, j_K$ with largest C_j^0;
2. Allocate one change point in the first K branches;
3. For each of these branches, set $\delta_{j_k}^{(h+1)}$ so that $C_j^1(\tau, \delta) = 0$

M Step: Segmentation

$$C_j(\alpha, \tau, \delta) = \sigma_j^{-2} \left(\mathbb{E}\left[X_j \mid Y\right] - q_j \mathbb{E}\left[X_{\mathrm{pa}(j)} \mid Y\right] - r_j - s_j \sum_k \mathbb{I}\{\tau_k = b_j\}\delta_k \right)^2$$

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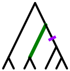
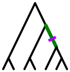


$$C_j^1(\alpha, \tau, \delta) = \sigma_j^{-2} \left(\mathbb{E}\left[X_j \mid Y\right] - q_j \mathbb{E}\left[X_{\mathrm{pa}(j)} \mid Y\right] - s_j \sum_k \mathbb{I}\{\tau_k = b_j\}\delta_k \right)^2$$



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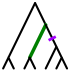

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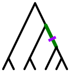

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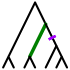

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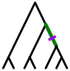

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OU : $r_j = \beta^{\mathrm{pa}(j)}$, a cost depends on all its parents.

- Exact minimization: too costly.
- Need of an heuristic.
- Idea: rewrite as a least square:

$$\left\| D - AU\Delta \right\|^2$$

  with D a vector of size $n + m$, A a diagonal matrix of size $n + m$, Δ the vector of shifts and U the incidence matrix of the tree.
- Then use Stepwise selection or LASSO.

back

BM

Conditional laws:

$$\mathbf{X}_j \mid \mathbf{X}_{\mathrm{pa}(j)} \sim \mathcal{N}\left(\mathbf{X}_{\mathrm{pa}(j)} + \sum_{k=1}^{K} \mathbb{I}\{\tau_k = b_j\}\boldsymbol{\delta}_k, \ell_j \mathbf{R}\right)$$

Completed log-likelihood:

$$\begin{aligned} p_{\boldsymbol{\theta}}(\mathbf{X} \mid \mathbf{X}_1) &= \prod_{j=2}^{m+n} p_{\boldsymbol{\theta}}\left(\mathbf{X}_j \mid \mathbf{X}_{\mathrm{pa}(j)}\right) \\ &= \prod_{j=2}^{m+n} \frac{1}{(2\pi)^{p/2}} \left|\ell_j \mathbf{R}\right|^{-1/2} \exp\left\{-\frac{1}{2}\left\|\mathbf{X}_j - \mathbf{X}_{\mathrm{pa}(j)} - \sum_{k=1}^{K} \mathbb{I}\{\tau_k = b_j\}\boldsymbol{\delta}_k\right\|^2_{(\ell_j \mathbf{R})^{-1}}\right\} \end{aligned}$$

BM

Conditional laws:

$$\mathbf{X}_j \mid \mathbf{X}_{\mathrm{pa}(j)} \sim \mathcal{N}\left(\mathbf{X}_{\mathrm{pa}(j)} + \sum_{k=1}^{K} \mathbb{I}\{\tau_k = b_j\}\boldsymbol{\delta}_k, \ell_j \mathbf{R}\right)$$

Objective Function:

$$\begin{aligned}
-2\mathbb{E}\left[\log p_{\boldsymbol{\theta}}(\mathbf{X}) \mid \mathbf{Y}\right] =& p(m+n)\log 2\pi + p\sum_{j=2}^{m+n}\log \ell_j \\
& + (m+n-1)\log|\mathbf{R}| + \sum_{j=2}^{m+n} \ell_j^{-1}\,\mathrm{tr}\left\{\mathbf{R}^{-1}\mathbb{V}\mathrm{ar}\left[\mathbf{X}_j - \mathbf{X}_{\mathrm{pa}(j)} \mid \mathbf{Y}\right]\right\} \\
& + \sum_{j=2}^{m+n} \ell_j^{-1}\left\|\mathbb{E}\left[\mathbf{X}_j - \mathbf{X}_{\mathrm{pa}(j)} \mid \mathbf{Y}\right] - \sum_{k=1}^{K}\mathbb{I}\{\tau_k = b_j\}\boldsymbol{\delta}_k\right\|_{\mathbf{R}^{-1}}^2
\end{aligned}$$

back

General OU with $\mathbf{A}$ positive definite

Conditional laws ($\mathbf{S}$ stationary variance):

$$\mathbf{X}_j \mid \mathbf{X}_{\mathrm{pa}(j)} \sim \mathcal{N}\left(e^{-\mathbf{A}\ell_j}\mathbf{X}_{\mathrm{pa}(j)} + (\mathbf{I}_p - e^{-\mathbf{A}\ell_j})\beta_j, \boldsymbol{\Upsilon}_i = \mathbf{S} - e^{-\mathbf{A}\ell_j}\mathbf{S}e^{-\mathbf{A}^T\ell_j}\right)$$

Completed log-likelihood:

$$\begin{aligned} p_{\boldsymbol{\theta}}(\mathbf{X} \mid \mathbf{X}_1) &= \prod_{j=2}^{m+n} p_{\boldsymbol{\theta}}\left(\mathbf{X}_j \mid \mathbf{X}_{\mathrm{pa}(j)}\right) \\ &= \prod_{j=2}^{m+n} \frac{1}{(2\pi)^{p/2}} |\boldsymbol{\Upsilon}_j|^{-1/2} \exp\left\{-\frac{1}{2}\left\|\mathbf{X}_j - e^{-\mathbf{A}\ell_j}\mathbf{X}_{\mathrm{pa}(j)} - (\mathbf{I}_p - e^{-\mathbf{A}\ell_j})\beta_j\right\|^2_{\boldsymbol{\Upsilon}_j^{-1}}\right\} \end{aligned}$$

General OU with $\mathbf{A}$ positive definite

Conditional laws (**S** stationary variance):

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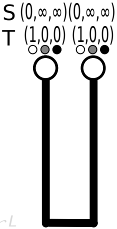
Objective Function:

$$\begin{aligned} -2\mathbb{E}\left[\log p_{\boldsymbol{\theta}}(\mathbf{X}) \mid \mathbf{Y}\right] = (m+n)p\log 2\pi &+ \sum_{j=2}^{m+n} \log|\boldsymbol{\Upsilon}_j| \\ &+ \sum_{j=2}^{m+n} \mathrm{tr}(\boldsymbol{\Upsilon}_j^{-1}\mathbb{V}\mathrm{ar}\left[\mathbf{D}_j \mid \mathbf{Y}\right]) \\ &+ \sum_{j=2}^{m+n} \left\|\mathbb{E}\left[\mathbf{D}_j \mid \mathbf{Y}\right] - \mathbf{E}_j\beta_j\right\|^2_{\boldsymbol{\Upsilon}_j^{-1}} \end{aligned}$$

(where $\mathbf{D}_j = \mathbf{X}_j - e^{-\mathbf{A}\ell_j}\mathbf{X}_{\mathrm{pa}(j)}$ and $\mathbf{E}_j = (\mathbf{I}_p - e^{-\mathbf{A}\ell_j}))$

back

Cardinal of Equivalence Classes



Initialization For tips

Propagation

$$\mathcal{K}_k^l = \underset{1 \leq p \leq K}{\text{argmin}} \left\{ S_{i_l}(p) + \mathbb{I}\{p \neq k\} \right\}$$

$$S_i(k) = \sum_{l=1}^{L} S_{i_l}(p_l) + \mathbb{I}\{p_l \neq k\} \; , \; \forall (p_1, \ldots p_L) \in \mathcal{K}_k^1 \times \ldots \times \mathcal{K}_k^L$$

$$T_i(k) = \sum_{(p_1, \ldots p_L) \in \mathcal{K}_k^1 \times \ldots \times \mathcal{K}_k^L} \prod_{l=1}^{L} T_{i_l}(p_l) = \prod_{l=1}^{L} \sum_{p_l \in \mathcal{K}_k^l} T_{i_l}(p_l)$$

Termination Sum on the root vector

back

Cardinal of Equivalence Classes

Initialization For tips

Propagation

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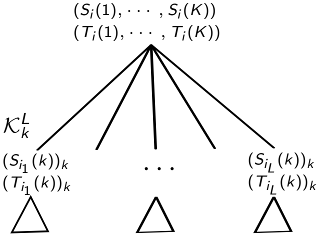
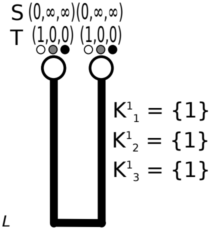


Termination Sum on the root vector

back

Cardinal of Equivalence Classes



○S(1) = 0 + 0 ; T(1) = 1 x 1
●S(2) = 0 + 1 ; T(2) = 1 x 1
●S(3) = 0 + 1 ; T(3) = 1 x 1

Initialization For tips

Propagation

$$\mathcal{K}_k^l = \underset{1\leq p\leq K}{\text{argmin}} \left\{ S_{i_l}(p) + \mathbb{I}\{p \neq k\} \right\}$$

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Termination Sum on the root vector

back

Cardinal of Equivalence Classes

Initialization For tips

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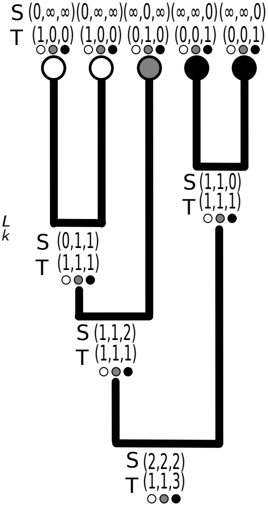
Termination Sum on the root vector

back



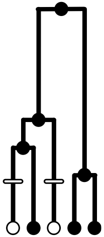
Linking Shifts and Clustering

Assumption "No Homoplasy": 1 shift = 1 new color

Proposition "K shifts $\iff$ $K+1$ clusters"

Linking Shifts and Clustering

Assumption "No Homoplasy": 1 shift = 1 new color



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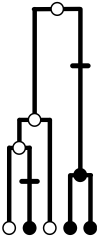
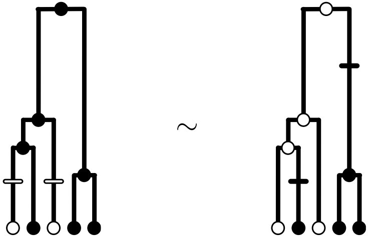


The No Homoplasy hypothesis is not respected.

Proposition "K shifts $\iff$ $K+1$ clusters"

Linking Shifts and Clustering

Assumption "No Homoplasy": 1 shift $=$ 1 new color

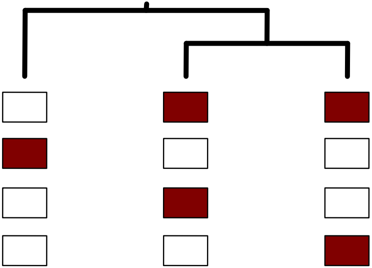


The No Homoplasy hypothesis is not respected.

Proposition "K shifts $\iff$ $K+1$ clusters"

Definitions

- $\mathcal{T}$ a rooted tree with n tips
- $N_K^{(\mathcal{T})} = |\mathcal{C}_K|$ the number of possible partitions of the tips in K clusters
- $A_K^{(\mathcal{T})}$ the number of possible *marked* partitions



Partitions in two groups for a binary tree with 3 tips

Difference between $N_2^{(\mathcal{T}_3)}$ and $A_2^{(\mathcal{T}_3)}$:

- $N_2^{(\mathcal{T}_3)} = 3$: partitions 1 and 2 are equivalent
- $A_2^{(\mathcal{T}_3)} = 4$: one marked color ("white = ancestral state")

General Formula (Binary Case)

If $\mathcal{T}$ is a binary tree, consider T_ℓ and $\mathcal{T}_r$ the left and right sub-trees of $\mathcal{T}$. Then:

$$\begin{cases} N_K^{(\mathcal{T})} = \sum_{k_1+k_2=K} N_{k_1}^{(\mathcal{T}_\ell)} N_{k_2}^{(\mathcal{T}_r)} + \sum_{k_1+k_2=K+1} A_{k_1}^{(\mathcal{T}_\ell)} A_{k_2}^{(\mathcal{T}_r)} \\ A_K^{(\mathcal{T})} = \sum_{k_1+k_2=K} A_{k_1}^{(\mathcal{T}_\ell)} N_{k_2}^{(\mathcal{T}_r)} + N_{k_1}^{(\mathcal{T}_\ell)} A_{k_2}^{(\mathcal{T}_r)} + \sum_{k_1+k_2=K+1} A_{k_1}^{(\mathcal{T}_\ell)} A_{k_2}^{(\mathcal{T}_r)} \end{cases}$$

We get:

$$N_{K+1}^{(\mathcal{T})} = N_{K+1}^{(n)} = \binom{2n-2-K}{K} \quad \text{and} \quad A_{K+1}^{(\mathcal{T})} = A_{K+1}^{(n)} = \binom{2n-1-K}{K}$$

Recursion Formula (General Case)

If we are at a node defining a tree $\mathcal{T}$ that has p daughters, with sub-trees $\mathcal{T}_1, \ldots, \mathcal{T}_p$, then we get the following recursion formulas:

$$\begin{cases} N_K^{(\mathcal{T})} = \displaystyle\sum_{\substack{k_1+\cdots+k_p=K \\ k_1,\ldots,k_p\geq 1}} \prod_{i=1}^{p} N_{k_i}^{(\mathcal{T}_i)} + \sum_{\substack{I\subset[\![1,p]\!] \\ |I|\geq 2}} \sum_{\substack{k_1+\cdots+k_p=K+|I|-1 \\ k_1,\ldots,k_p\geq 1}} \prod_{i\in I} A_{k_i}^{(\mathcal{T}_i)} \prod_{i\notin I} N_{k_i}^{(\mathcal{T}_i)} \\ A_K^{(\mathcal{T})} = \displaystyle\sum_{\substack{I\subset[\![1,p]\!] \\ |I|\geq 1}} \sum_{\substack{k_1+\cdots+k_p=K+|I|-1 \\ k_1,\ldots,k_p\geq 1}} \prod_{i\in I} A_{k_i}^{(\mathcal{T}_i)} \prod_{i\notin I} N_{k_i}^{(\mathcal{T}_i)} \end{cases}$$

No general formula. The result depends on the topology of the tree.

back

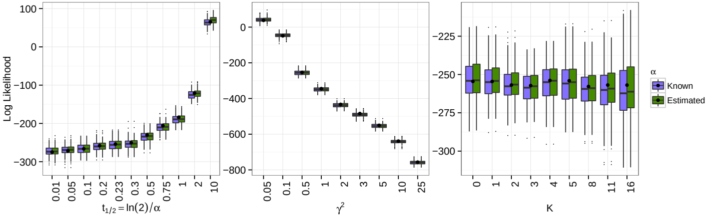
Simulations Design (Uyeda and Harmon, 2014)

- Topology of the tree fixed (unit height, $\lambda = 0.1$, with 64, 128, 256 taxa).
- Initial optimal value fixed: $\beta_0 = 0$
- One "base" scenario $\alpha_b = 3$, $\gamma_b^2 = 0.5$, $K_b = 5$.
- $\alpha \in \log(2)/\{0.01, 0.05, 0.1, 0.2, 0.23, 0.3, 0.5, 0.75, 1, 2, 10\}$.
- $\gamma^2 \in \{0.3, 0.6, 3, 6, 12, 18, 30, 60, 150\}/(2\alpha_b)$.
- $K \in \{0, 1, 2, 3, 4, 5, 8, 11, 16\}$.
- Shifts values $\sim \frac{1}{2}\mathcal{N}(4, 1) + \frac{1}{2}\mathcal{N}(-4, 1)$
- Shifts randomly placed at regular intervals separated by 0.1 unit length.
- $n = 200$ repetitions : 16200 configurations.

CPU time on cluster MIGALE (Jouy-en-Josas):

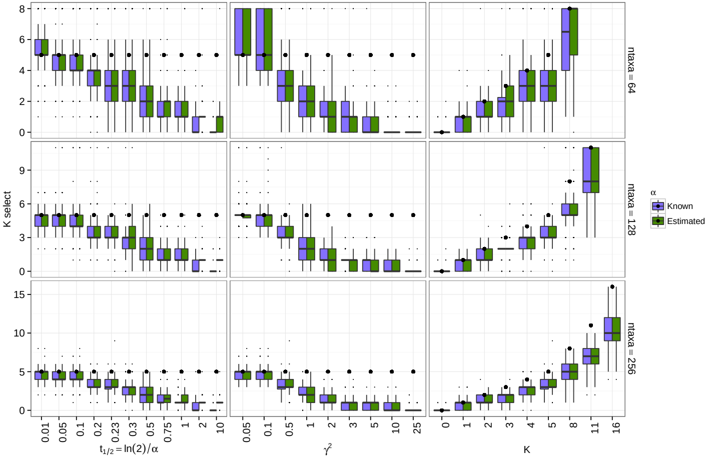
- α known: 6 minutes per estimation (66 days in total).
- α unknown: 52 minutes per estimation (570 days in total).

Log-Likelihood

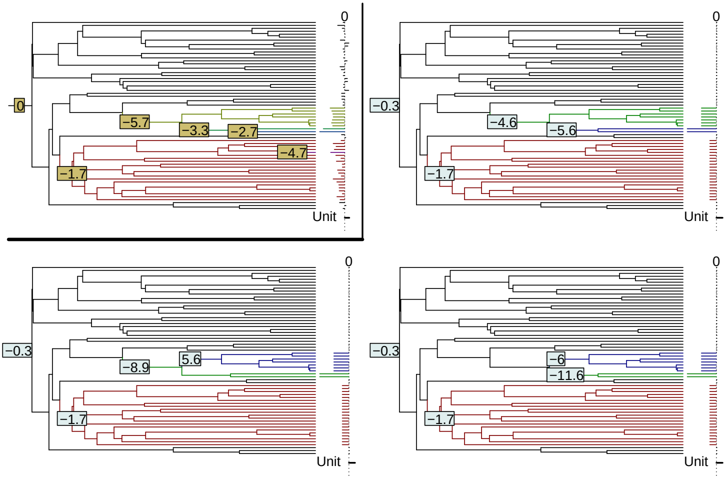


Log likelihood for a tree with 256 tips. Solid black dots are the median of the log likelihood for the true parameters.

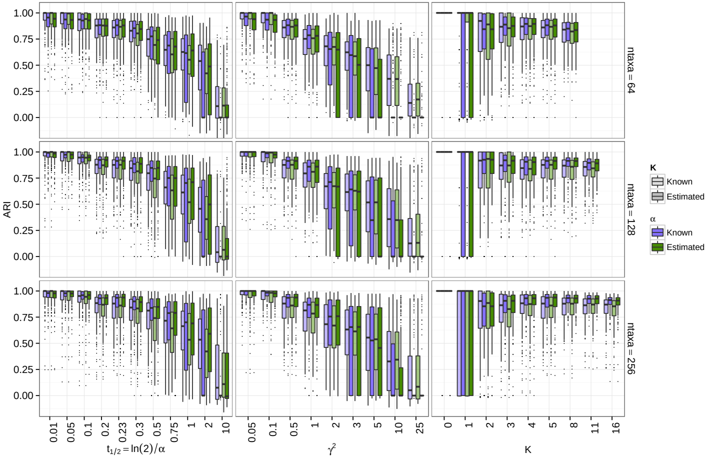
Number of Shifts



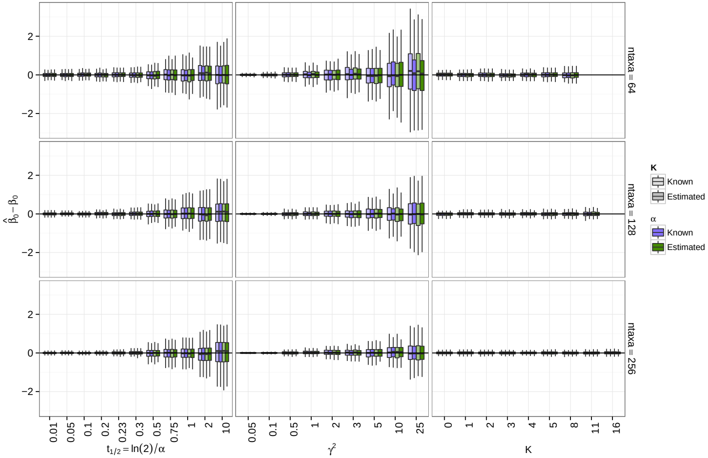
One Example


0
−5.7
−3.3
−2.7
−4.7
−1.7
0
Unit
−0.3
−4.6
−5.6
−1.7
0
Unit
−0.3
−8.9
5.6
−1.7
0
Unit
−0.3
−6
−11.6
−1.7
0
Unit

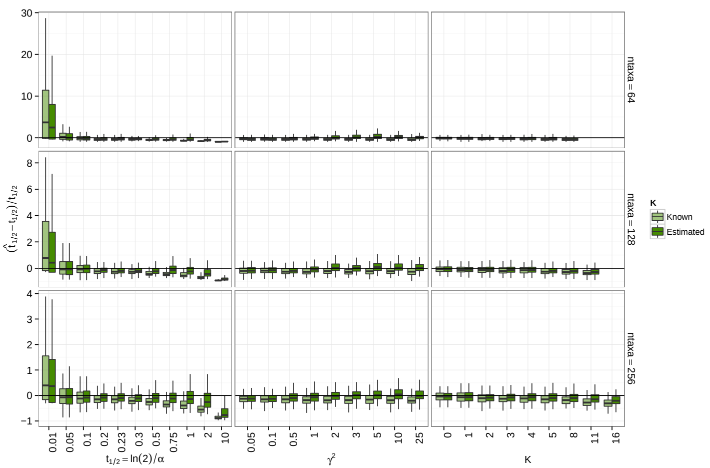
Adjusted Rand Index



Parameters: β_0



Parameters: α



Parameters: γ^2