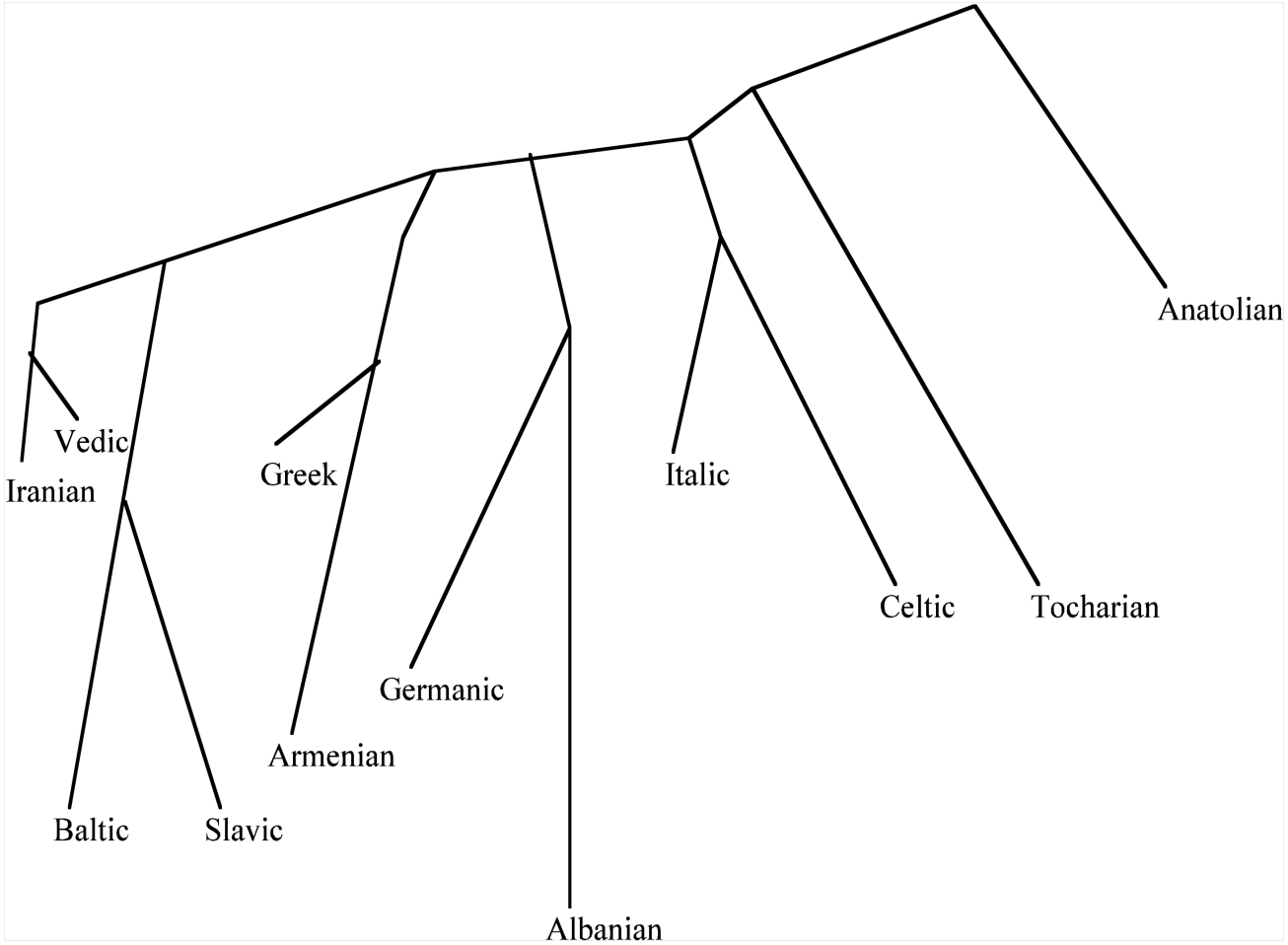
A simulation study comparing phylogeny reconstruction methods for linguistics

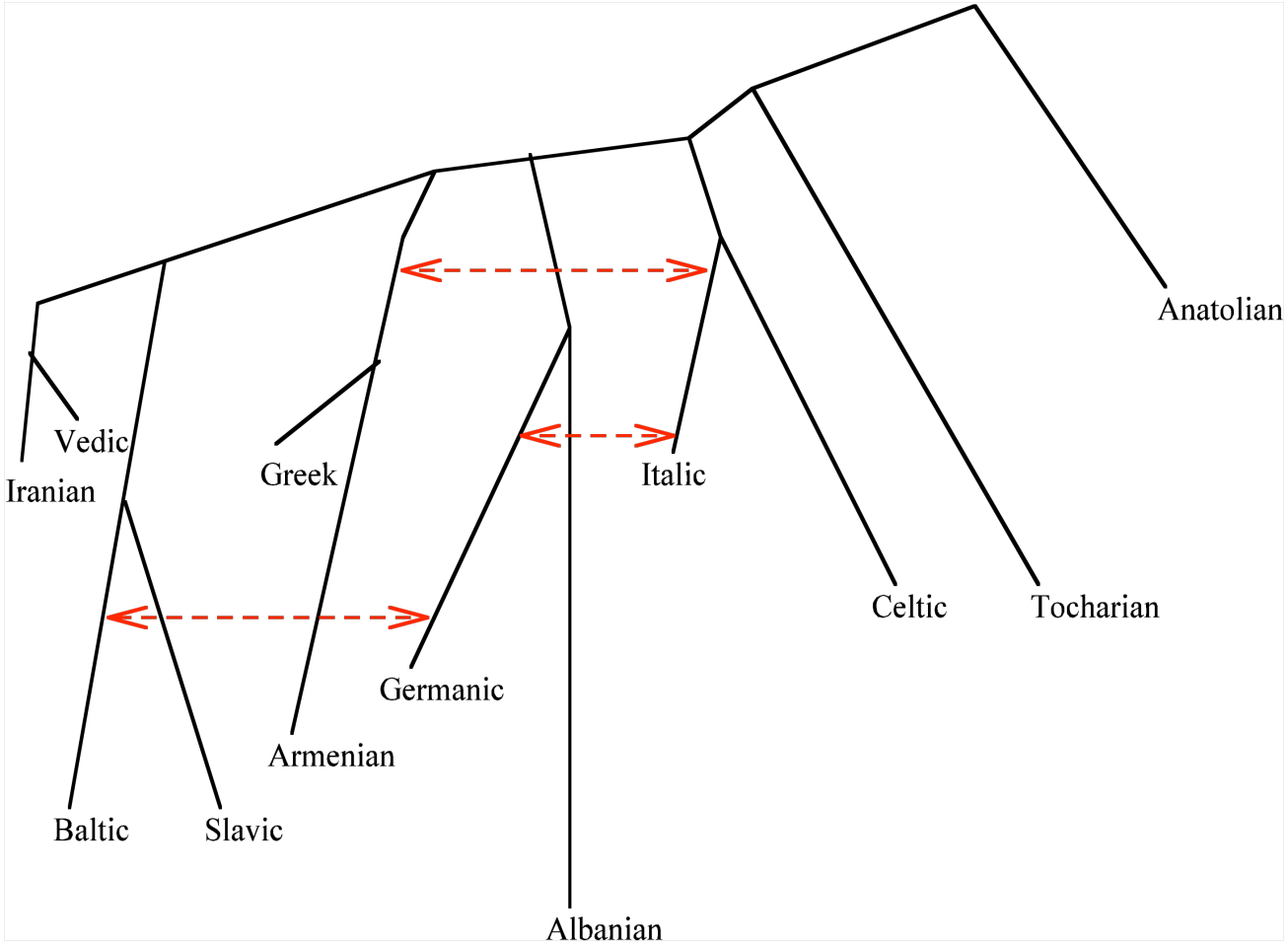
Tandy Warnow
The University of Texas at Austin
The Newton Institute for Mathematical Research

Collaborators: Francois Barbancon,
Don Ringe, Luay Nakhleh, Steve Evans

Possible Indo-European tree (Ringe, Warnow and Taylor 2000)



Phylogenetic Network for IE
Nakhleh *et al*., Language 2005



Controversies for IE history

- Subgrouping: Other than the 10 major subgroups, what is likely to be true? In particular, what about
  - Italo-Celtic
  - Greco-Armenian
  - Anatolian + Tocharian
  - Satem Core (Indo-Iranian and Balto-Slavic)
  - Location of Germanic
- Dates?
- How tree-like is IE?

Controversies for IE history

Note: many reconstructions of IE have been done, but produce different histories which differ in significant ways (e.g., the location of Germanic)

Possible issues:

Dataset (modern vs. ancient data, errors in the cognancy judgments, lexical vs. all types of characters, screened vs. unscreened)

Translation of multi-state data to binary data

Reconstruction method

The performance of methods on an IE data set (Transactions of the Philological Society, Nakhleh et al. 2005)

Observation: Different datasets (not just different methods) can give different reconstructed phylogenies.

Objective: Explore the differences in reconstructions as a function of data (lexical alone versus lexical, morphological, and phonological), screening (to remove obviously homoplastic characters), and methods. However, use a ***better basic dataset*** (where cognancy judgments are more reliable).

Better datasets

- Ringe & Taylor
  - The screened full dataset of 294 characters (259 lexical, 13 morphological, 22 phonological)
  - The unscreened full dataset of 336 characters (297 lexical, 17 morphological, 22 phonological)
  - The screened lexical dataset of 259 characters.
  - The unscreened lexical dataset of 297 characters.

Differences between different characters

- **Lexical**: most easily borrowed (most borrowings detectable), and homoplasy relatively frequent (we estimate about 25-30% overall for our wordlist, but a much smaller percentage for basic vocabulary).
- **Phonological**: can still be borrowed but much less likely than lexical. Complex phonological characters are infrequently (if ever) homoplastic, although simple phonological characters very often homoplastic.
- **Morphological**: least easily borrowed, least likely to be homoplastic.

Table 1: The 24 IE languages analyzed.

Language	Abbreviation	Language	Abbreviation
Hittite	HI	Old English	OE
Luvian	LU	Old High German	OG
Lycian	LY	Classical Armenian	AR
Vedic	VE	Tocharian A	TA
Avestan	AV	Tocharian B	TB
Old Persian	PE	Old Irish	OI
Ancient Greek	GK	Welsh	WE
Latin	LA	Old Church Slavonic	OC
Oscan	OS	Old Prussian	PR
Umbrian	UM	Lithuanian	LI
Gothic	GO	Latvian	LT
Old Norse	ON	Albanian	AL

Phylogeny reconstruction methods

- Neighbor joining
- UPGMA (technique in glottochronology)
- Maximum parsimony
- Maximum compatibility (weighted and unweighted)
- Gray and Atkinson (Bayesian estimation based upon presence/absence of cognates)

Some observations

- UPGMA (i.e., the tree-building technique for glottochronology) does the worst (e.g. splits Italic and Iranian groups).
- Other than UPGMA, all methods reconstruct the ten major subgroups, as well as **Anatolian + Tocharian** and **Greco-Armenian**.
- The Satem Core (Indo-Iranian plus Balto-Slavic) is not always reconstructed.
- Almost all analyses put Italic, Celtic, and Germanic together. (The only exception is weighted maximum compatibility on datasets that include morphological characters.)

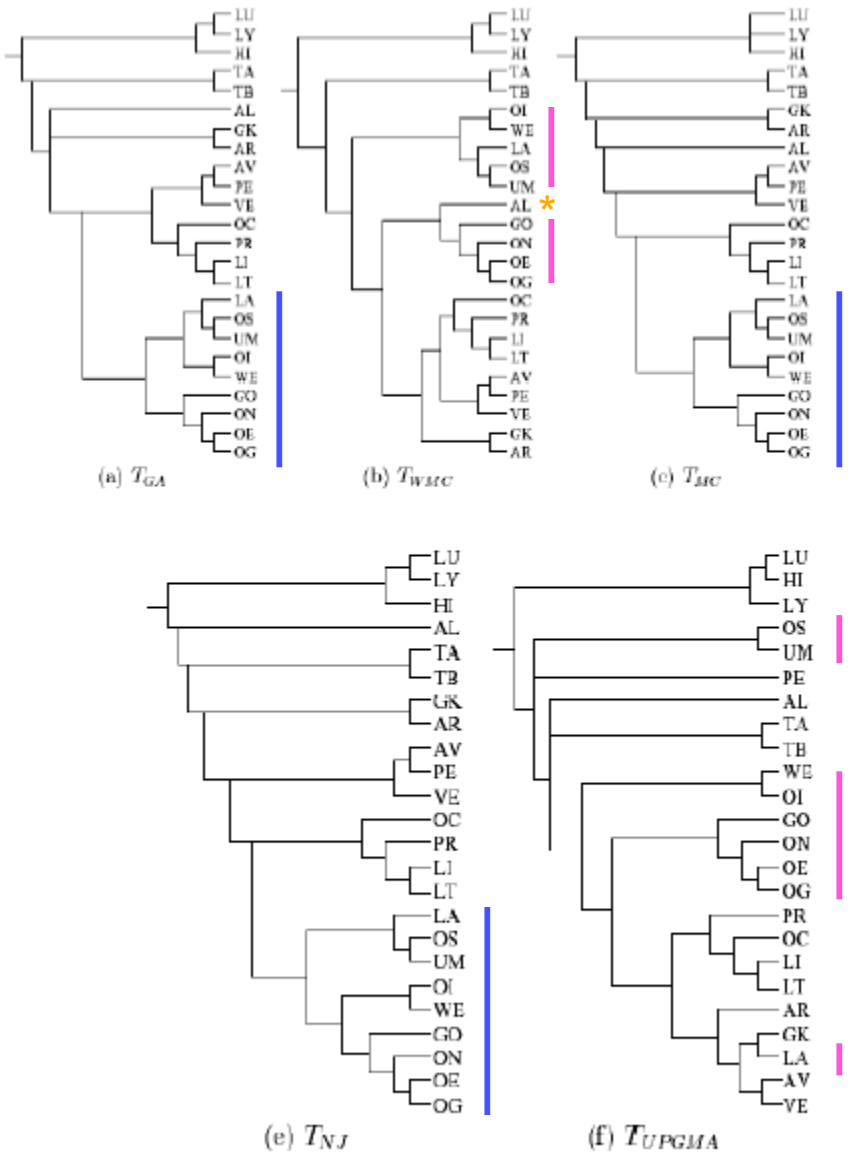


Figure 1. Five trees inferred on the screened full dataset

GA = Gray+Atkinson Bayesian MCMC method

WMC = weighted maximum compatibility

MC = maximum compatibility (identical to maximum parsimony on this dataset)

NJ = neighbor joining (distance-based method, based upon corrected distance)

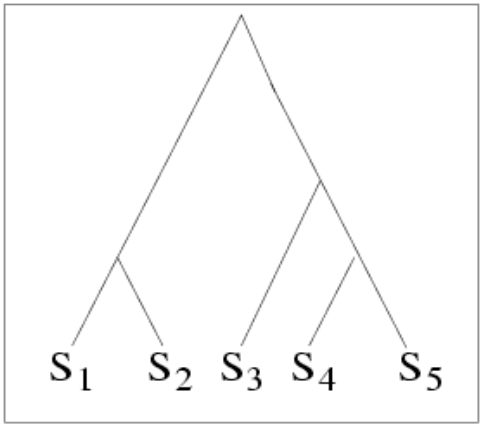
UPGMA = agglomerative clustering technique used in glottochronology.

Different methods/data
give different answers.

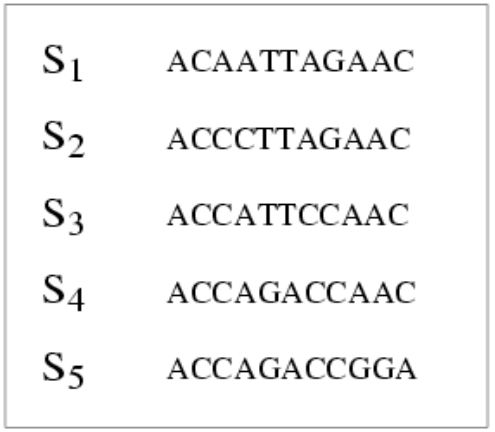
We don't know
which answer is correct.

Which method(s)/data
should we use?

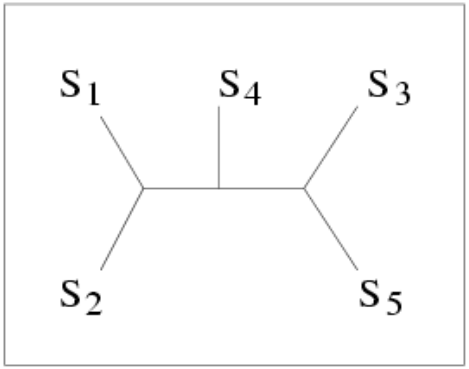
Simulation study (cartoon)



TRUE TREE

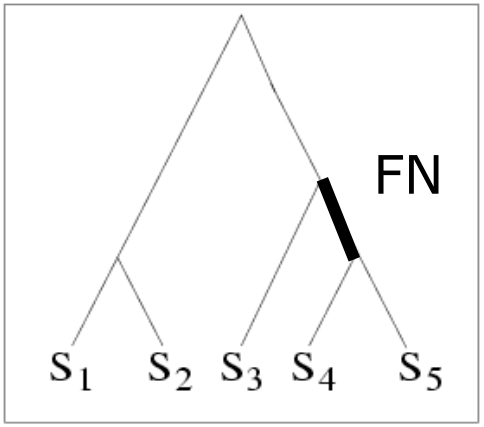


DNA SEQUENCES

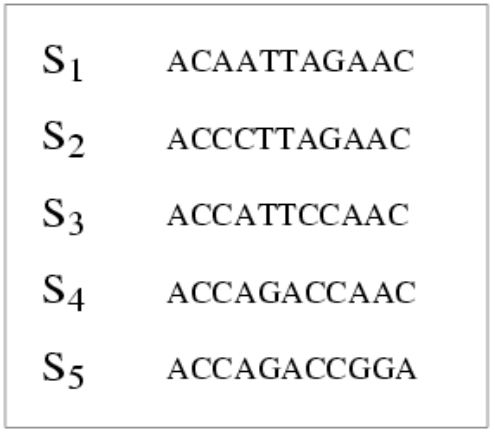


INFERRED TREE

Simulation study (cartoon)



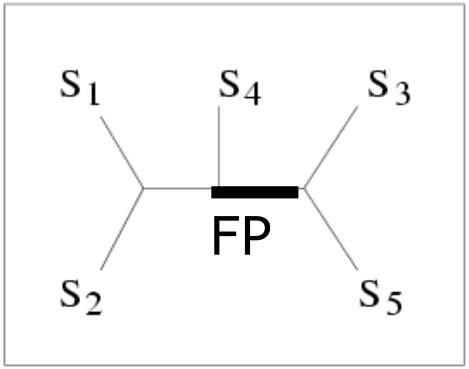
TRUE TREE



DNA SEQUENCES

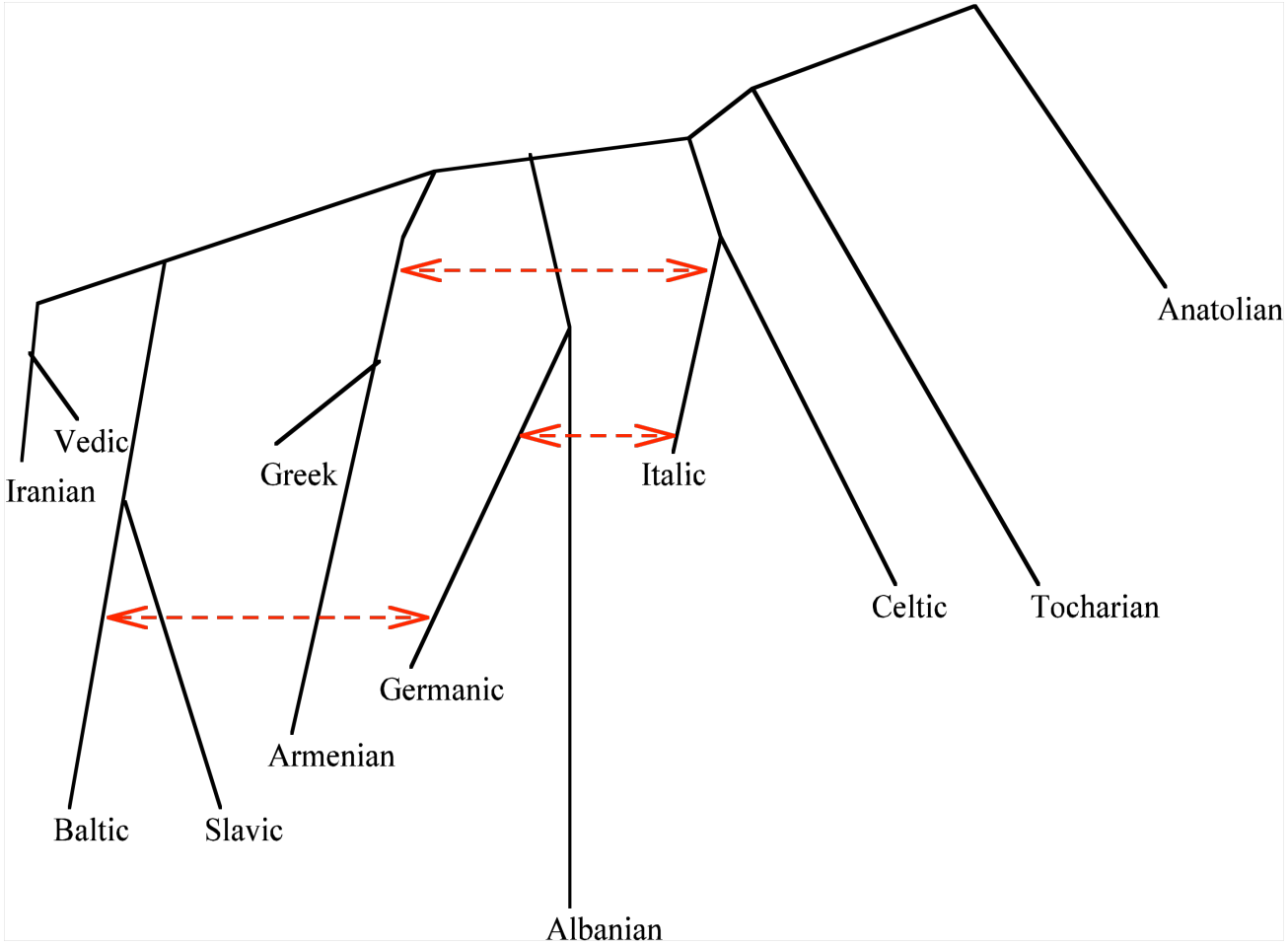
FN: false negative
(**missing edge**)
FP: false positive
(incorrect edge)

50% error rate

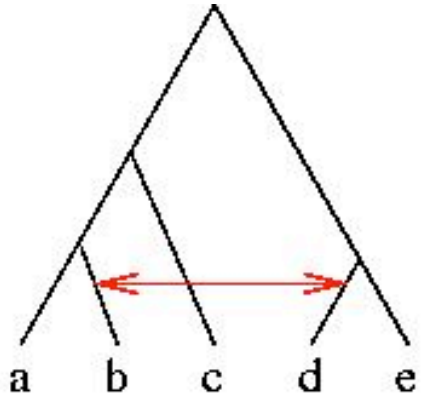


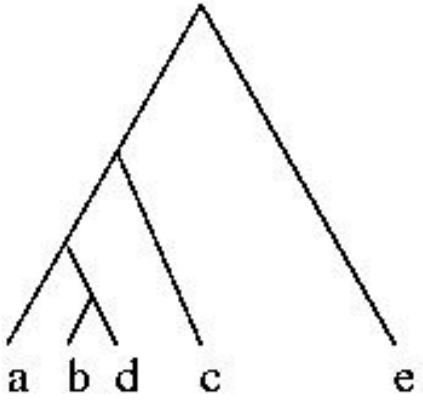
INFERRED TREE

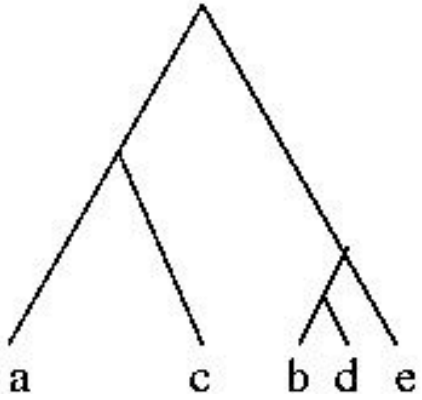
Phylogenetic Network Evolution

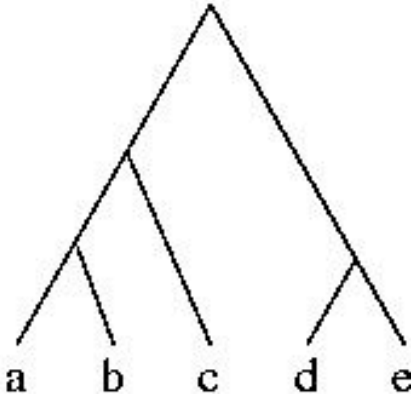


Modelling borrowing: Networks and Trees within Networks

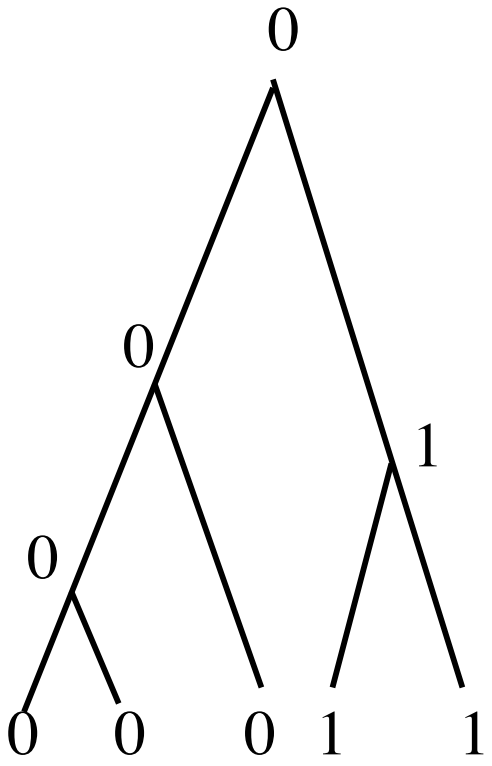




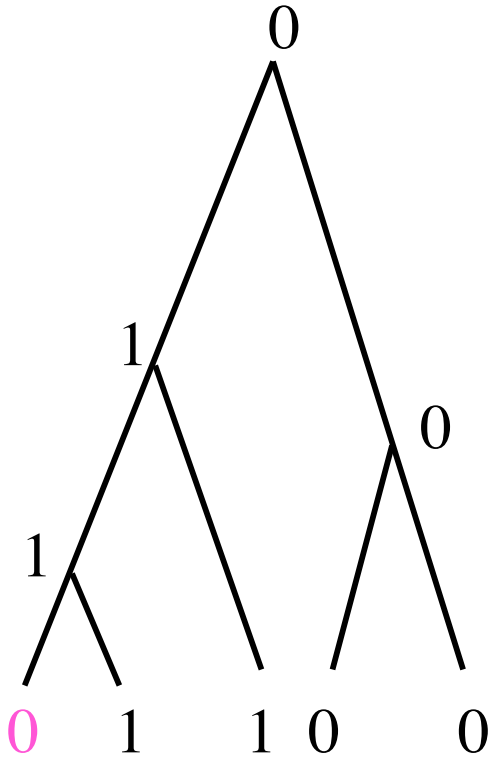




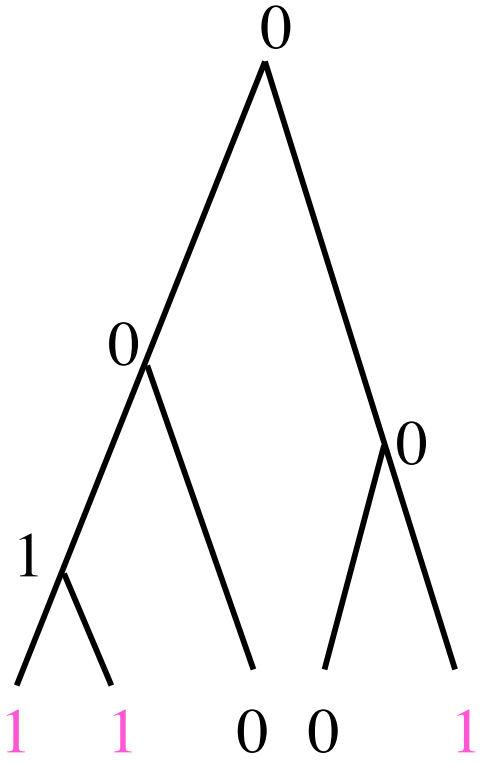
Some useful terminology: homoplasy



no homoplasy

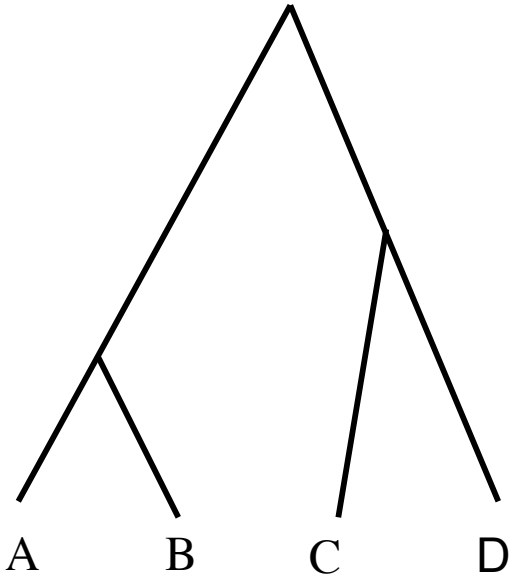


back-mutation

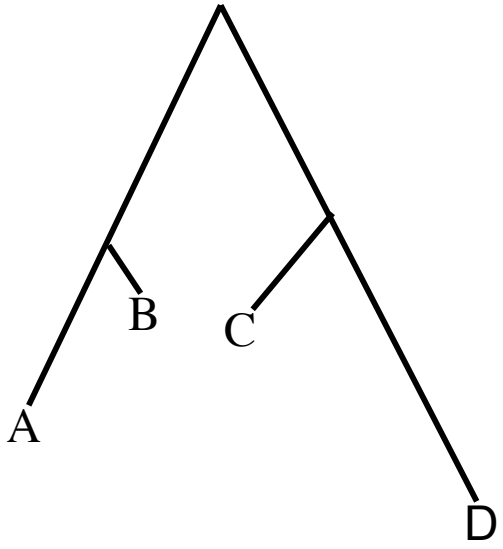


parallel evolution

Some useful terminology: lexical clock



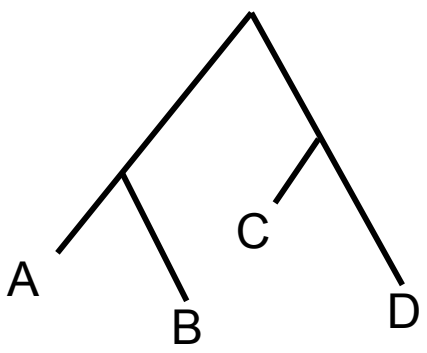
lexical clock

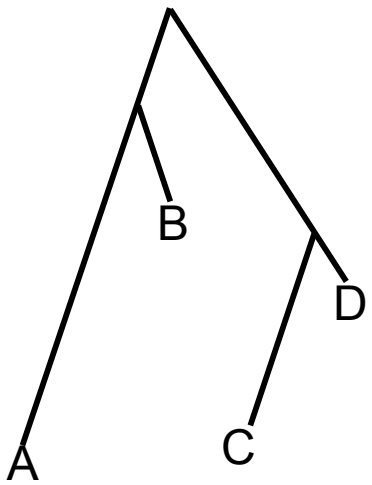


no lexical clock

edge lengths represent expected numbers of substitutions

Heterotachy = departure from rates-across-sites





The underlying tree is fixed, but there are no constraints on edge length variations between characters.

Previous simulations

- Most previous simulations of linguistic evolution had evolved characters without any borrowing or homoplasy, all under an i.i.d. assumption, and many have assumed a strong lexical clock.
- Some (notably McMahon and McMahon) had evolved characters with small amounts of borrowing but no homoplasy, on small networks (with one contact edge)

Our model
(Cambridge University Press, 2006)

"Genetic evolution"

- Characters evolve independently from each other, but under a linguistic equivalent of the Tuffley & Steel "no common mechanism" model
- We allow for a single homoplastic state, **h*.** The non-homoplastic states are indicated by **n** (or **n'**).
- If a character changes state on an edge, it either evolves into the homoplastic state h*, or innovates to a new non-homoplastic state.
- For each character **c** and tree edge **e**, there is a quintuple of probabilities: $p_{e,c}(n,n)$, $p_{e,c}(n,n')$, $p_{e,c}(n,h^*)$, $p_{e,c}(h^*,h^*)$, and $p_{e,c}(h^*,n)$.
- Binary phonological characters c satisfy $p_{e,c}(h^*,h^*)=1$ and $p_{e,c}(h^*,n)=0$. We make the mild assumption that $0<p_{e,c}(n,h^*)<1$.
- Morphological and lexical characters have an unbounded number of states, so we only require that $0<p_{e,c}(n,n')<1$.

"Borrowing" (horizontal transfer):

- Each contact edge e=(v,w) has a parameter K_e which is the probability of transmission of character states from v to w. Note that $K_{(v,w)}$ may not be equal to $K_{(w,v)}$.
- Each character **c** has a relative probability $\mathbf{B_c}$ of being borrowed, so that the *probability that character c is borrowed* across a contact edge e is $\mathbf{B_c}\ \mathbf{K_e}$

Theoretical results - I

- The model tree (but not its root or parameters) is identifiable, and can be reconstructed with high probability in polynomial time, given logarithmic number of morphological and lexical characters (extension of result by Mossel and Steel 2004 for homoplasy-free model).

Theoretical results - II

- Other statistically consistent and simpler polynomial time algorithms exist (compute all bipartitions or compute all quartet-trees), with longer sequence length requirements. These apply to the morphological and lexical characters.
- Reconstruction from binary phonological characters can be done if they evolve iid, using a distance-based approach.

Theoretical results - III

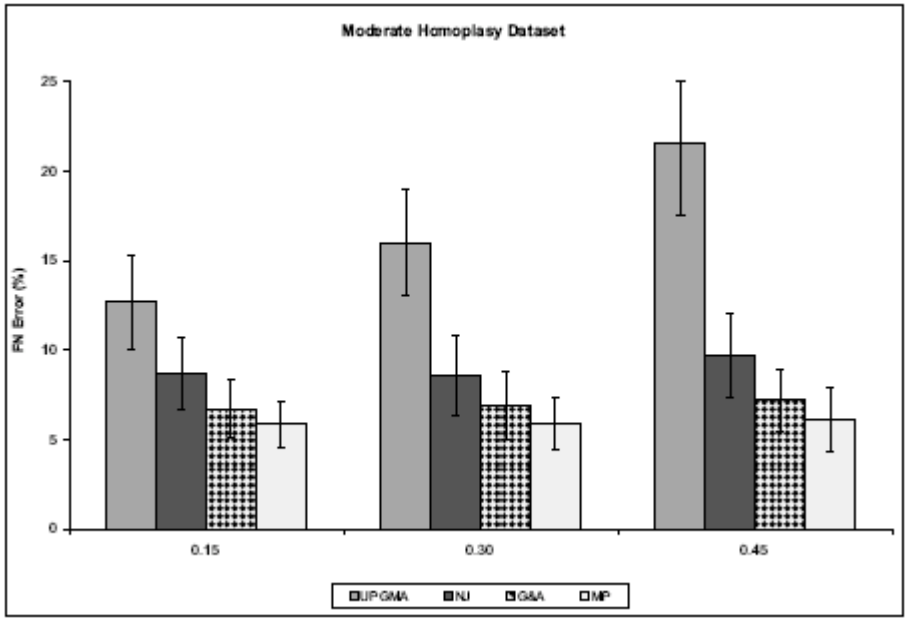
- The likelihood of the tree can be computed in linear time for each character, using a dynamic programming algorithm.

Our simulation study (in press)

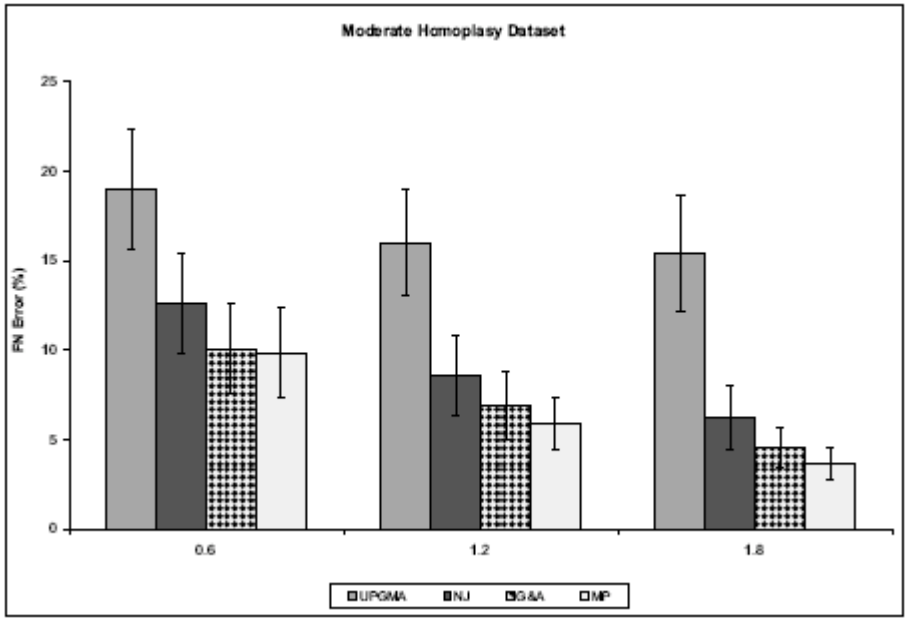
- Model phylogenetic networks: each had 30 leaves and up to three contact edges, and varied in the *deviation from a lexical clock.*
- Characters: we had up to 360 lexical characters and 60 morphological characters, each type with two rates for homoplasy and borrowing estimated from our “screened” and “unscreened” IE data. We also varied the degree of heterotachy (deviation from the rates-across-sites assumption).
- Performance metric: We compared estimated trees to the “genetic tree” wrt the missing edge rate.

Observations

1. Choice of reconstruction method does matter.
2. Relative performance between methods is quite stable (distance-based methods worse than character-based methods).
3. Choice of data does matter (good idea to add morphological characters).
4. Accuracy only slightly lessened with small increases in homoplasy, borrowing, or deviation from the lexical clock.
5. Some amount of heterotachy helps!



(i)



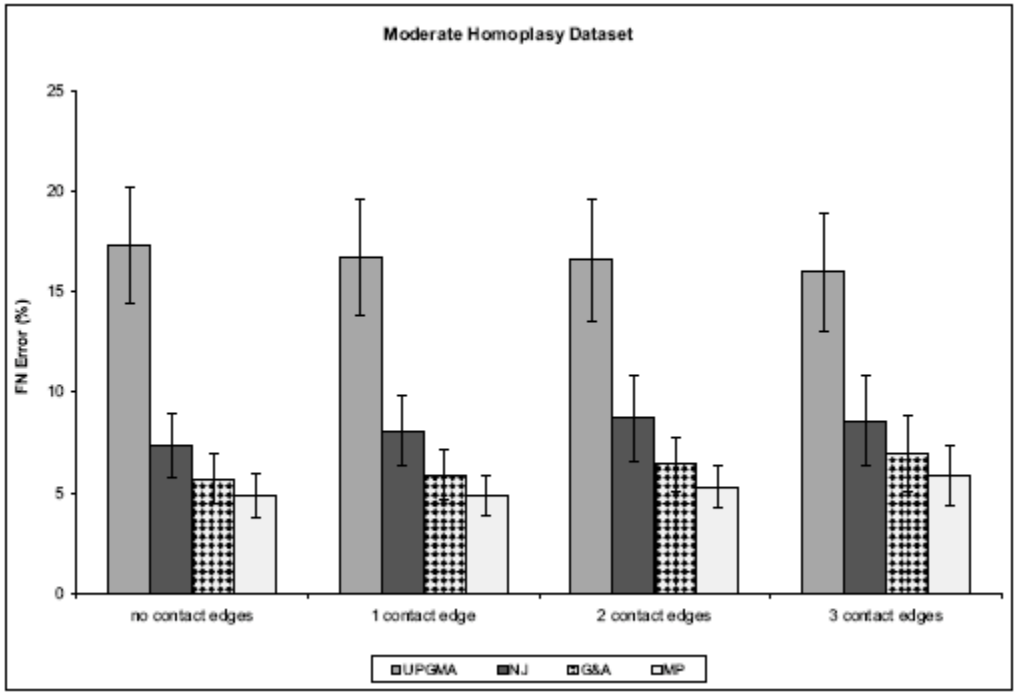
(ii)

Relative performance of methods on **moderate homoplasy** datasets under various model conditions:

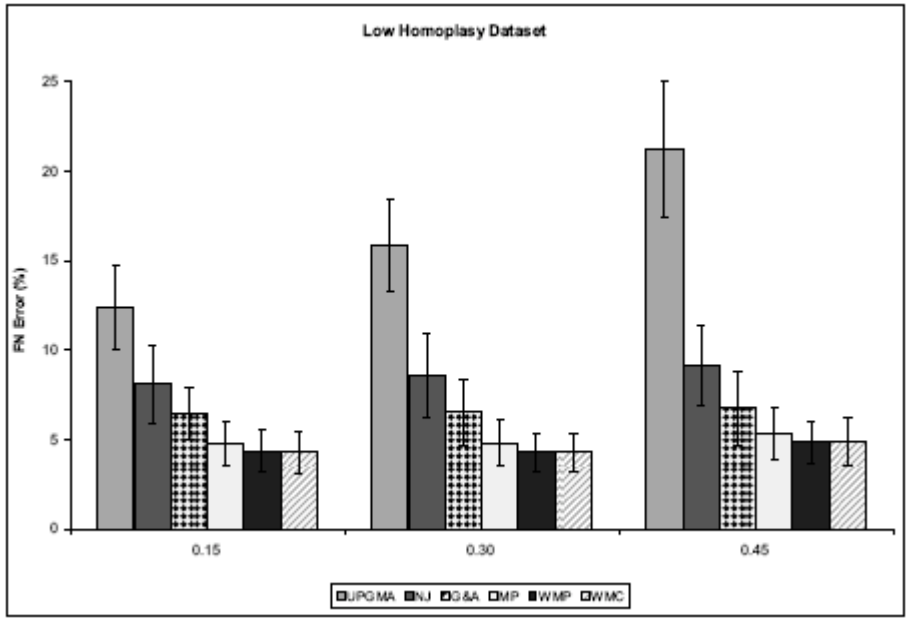
(i) varying the deviation from the lexical clock,

(ii) varying heterotachy, and

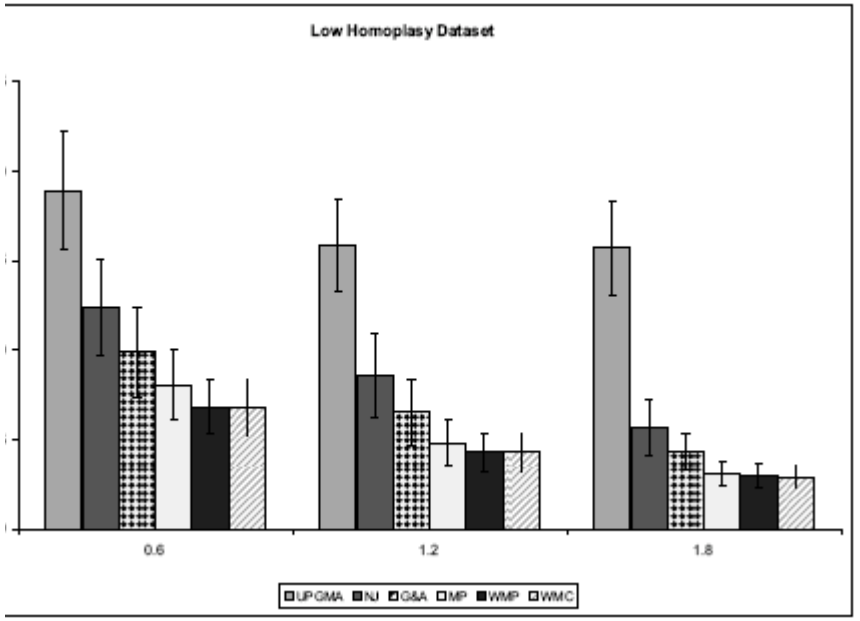
(iii) varying the number of contact edges.



(iii)



(i)



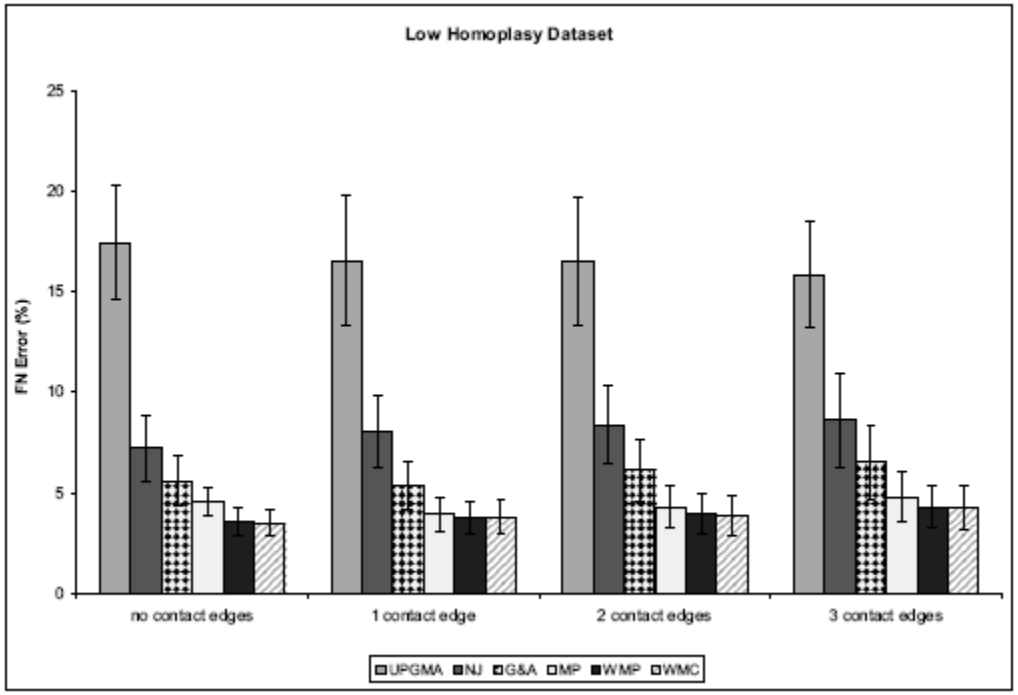
(ii)

Relative performance of methods for **low homoplasy datasets** under various model conditions:

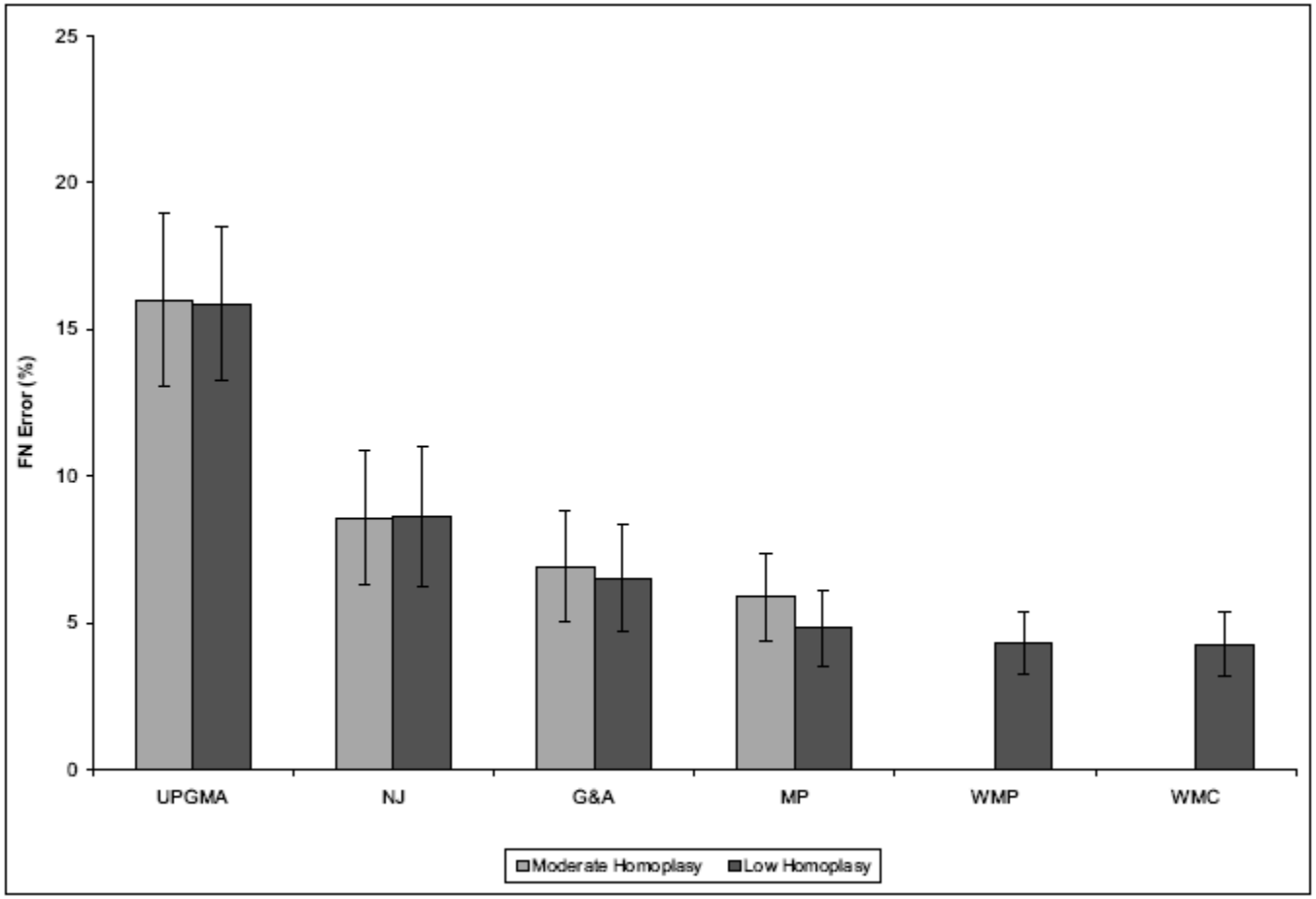
(i) Varying the deviation from the lexical clock,

(ii) Varying the heterotachy, and

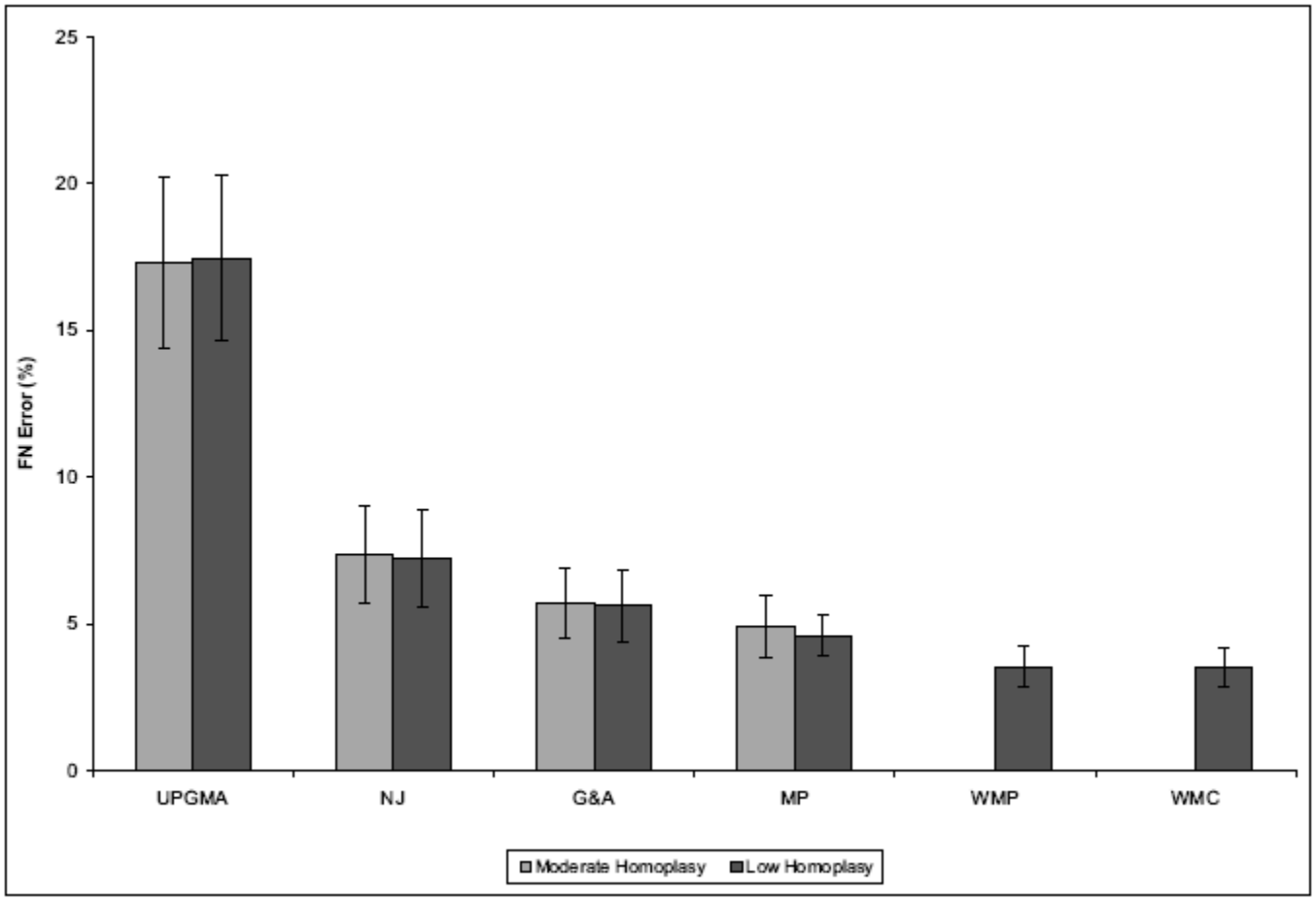
(iii) Varying the number of contact edges.



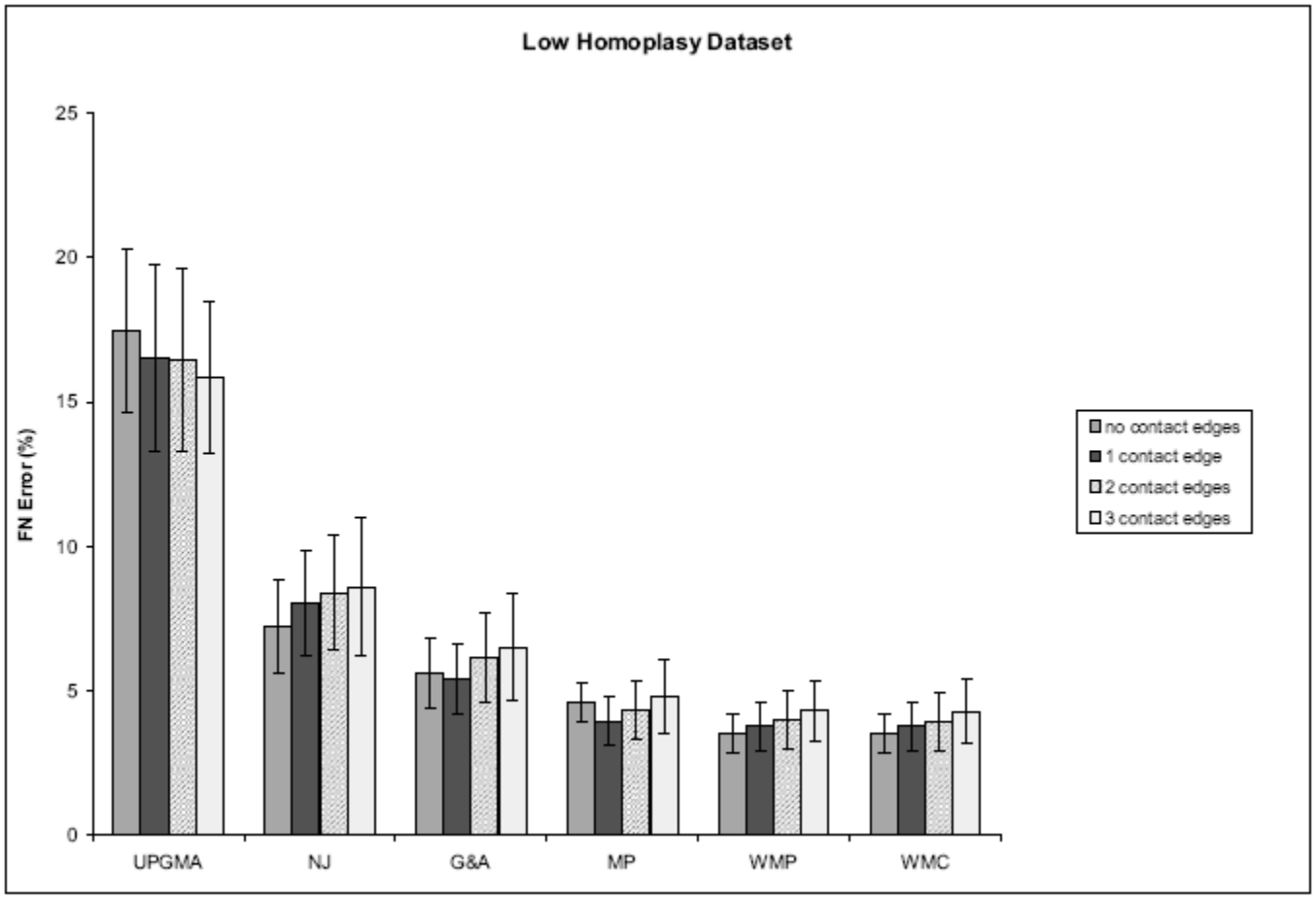
(iii)



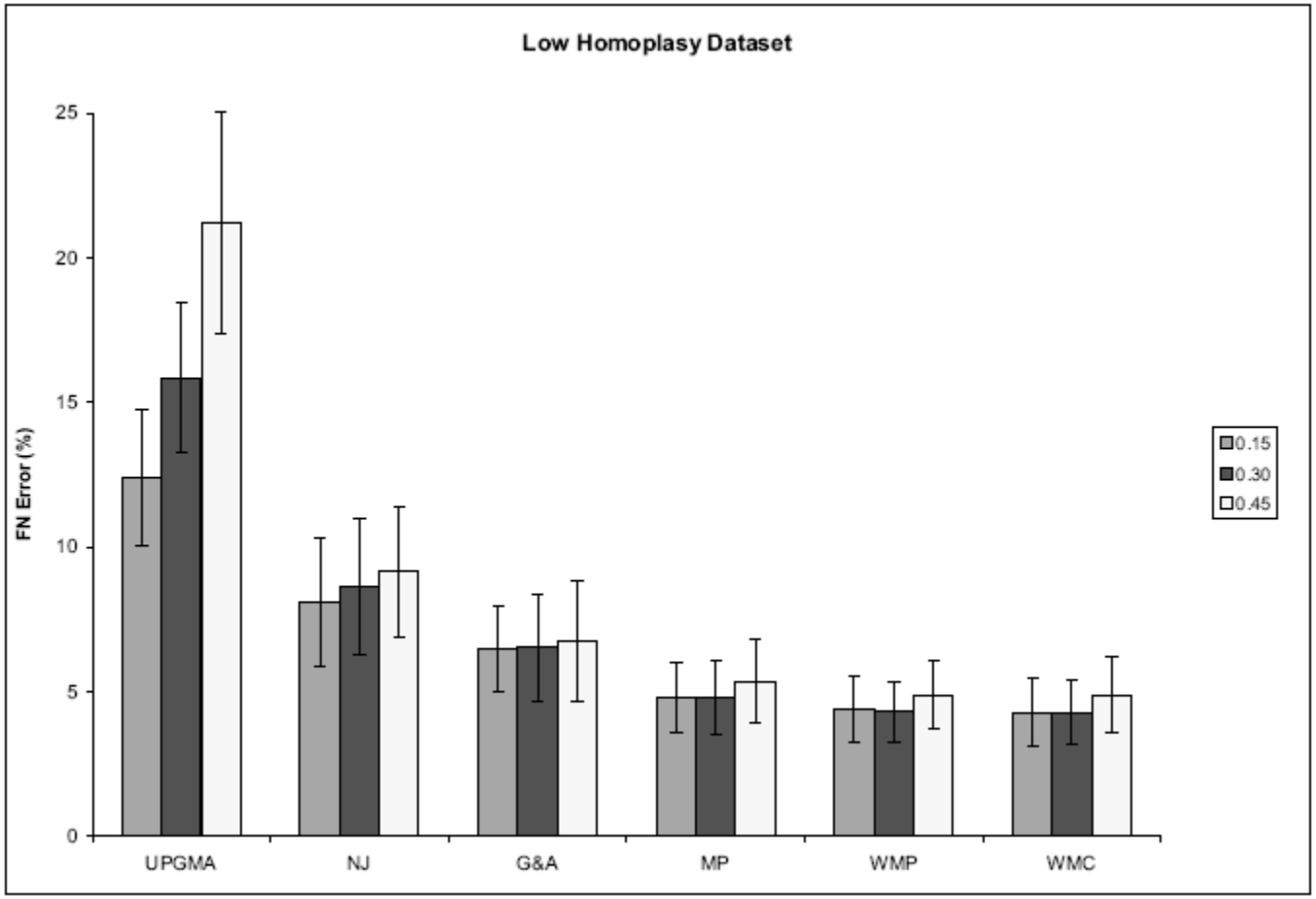
Impact of homoplasy for characters evolved down a network with three contact edges under a moderate deviation from the lexical clock and moderate heterotachy.



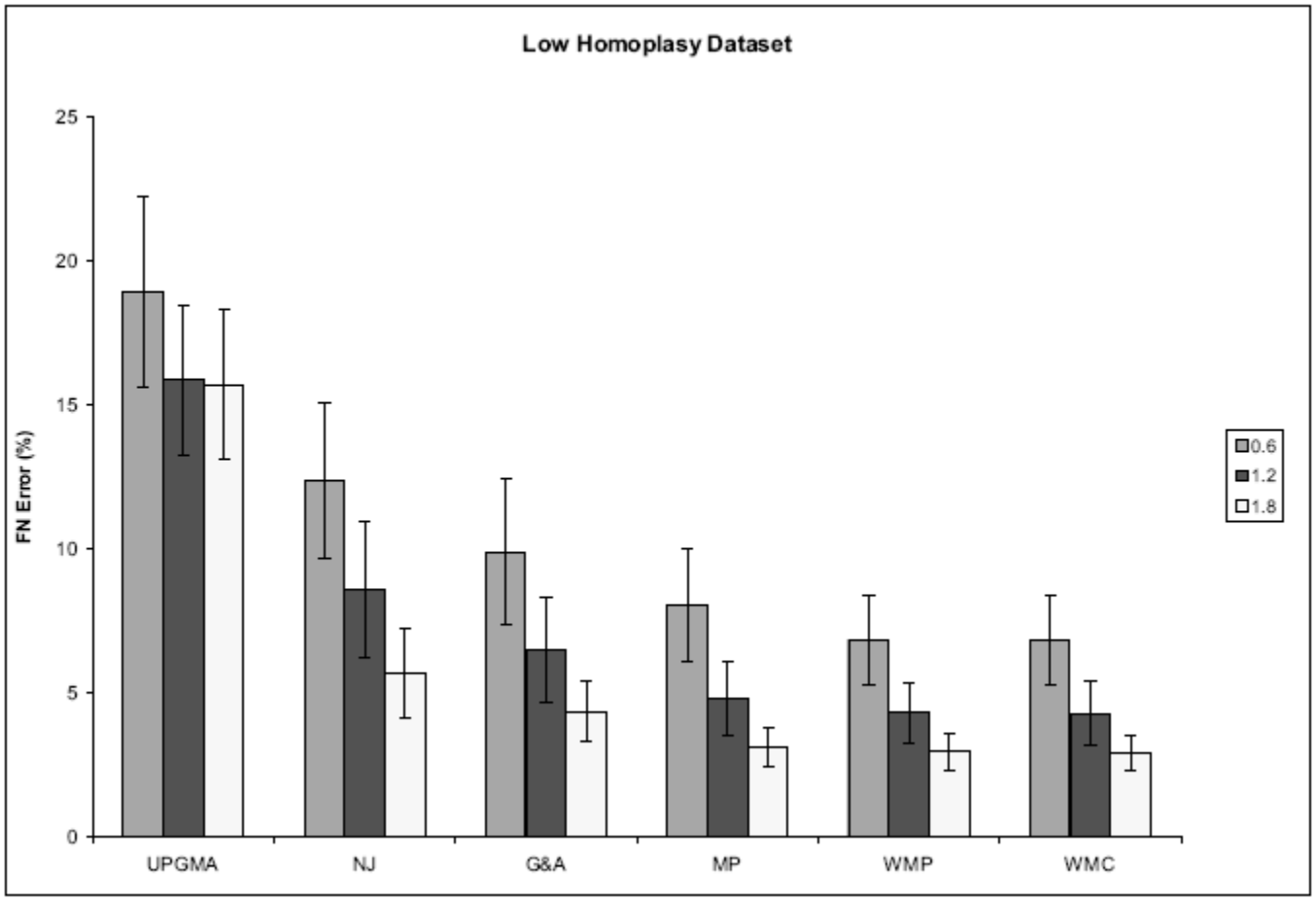
Impact of homoplasy for characters evolved down a tree under a moderate deviation from a lexical clock and moderate heterotachy. *(Our weighting is inappropriate for "unscreened" data.)*



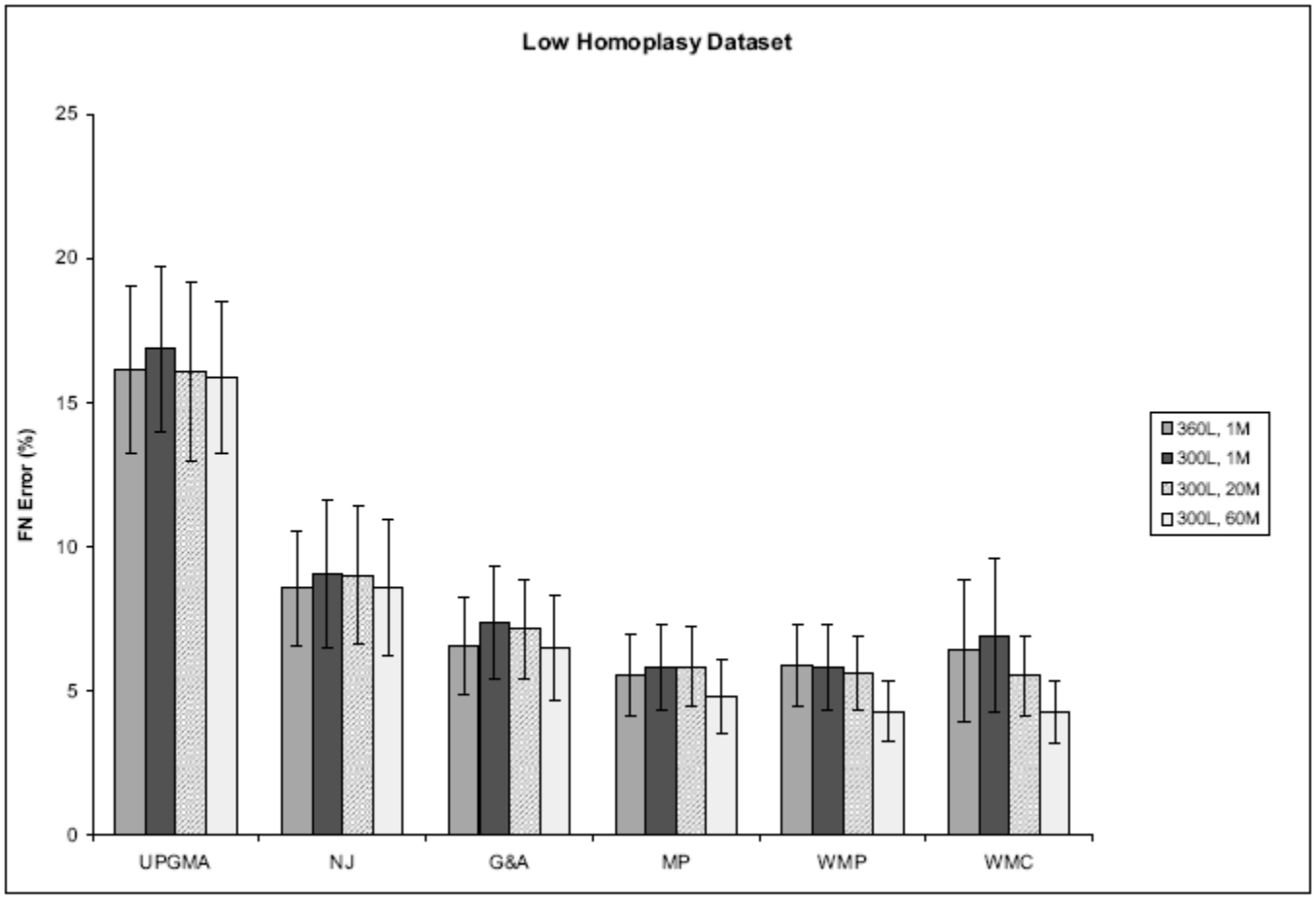
Impact of the number of contact edges for characters evolved under low homoplasy, moderate deviation from a lexical clock, and moderate heterotachy.



Impact of the deviation from a lexical clock (from low to moderate) for characters evolved down a network with three contact edges under low levels of homoplasy and with moderate heterotachy.



Impact of heterotachy for characters evolved down a network with three contact edges, with low homoplasy, and with moderate deviation from a lexical clock. Heterotachy increases with the parameter.



Impact of data selection for characters evolved down a network with three contact edges, under low homoplasy (“screened data"), moderate deviation from a lexical clock, and moderate heterotachy.

Observations

1. Choice of reconstruction method does matter.
2. Relative performance between methods is quite stable (distance-based methods worse than character-based methods).
3. Choice of data does matter (good idea to add morphological characters).
4. Accuracy only slightly lessened with small increases in homoplasy, borrowing, or deviation from the lexical clock.
5. Some amount of heterotachy helps!

Future research

- We need more investigation of methods based on stochastic models (Bayesian beyond G+A, maximum likelihood, NJ with better distance corrections), as these are now the methods of choice in biology. This requires ***better models of linguistic evolution*** and hence ***input from linguists***!

Future research (continued)

- Should we screen? The simulation uses low homoplasy as a proxy for screening, but real screening throws away data and may introduce bias.
- How do we detect/reconstruct borrowing?
- How do we handle missing data in methods based on stochastic models?
- How do we handle polymorphism?

For more information

- Please see the Computational Phylogenetics for Historical Linguistics web site for papers, data, and additional material **http://www.cs.rice.edu/~nakhleh/CPHL**

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- Collaborators: Don Ringe, Steve Evans, Luay Nakhleh, and Francois Barbancon.