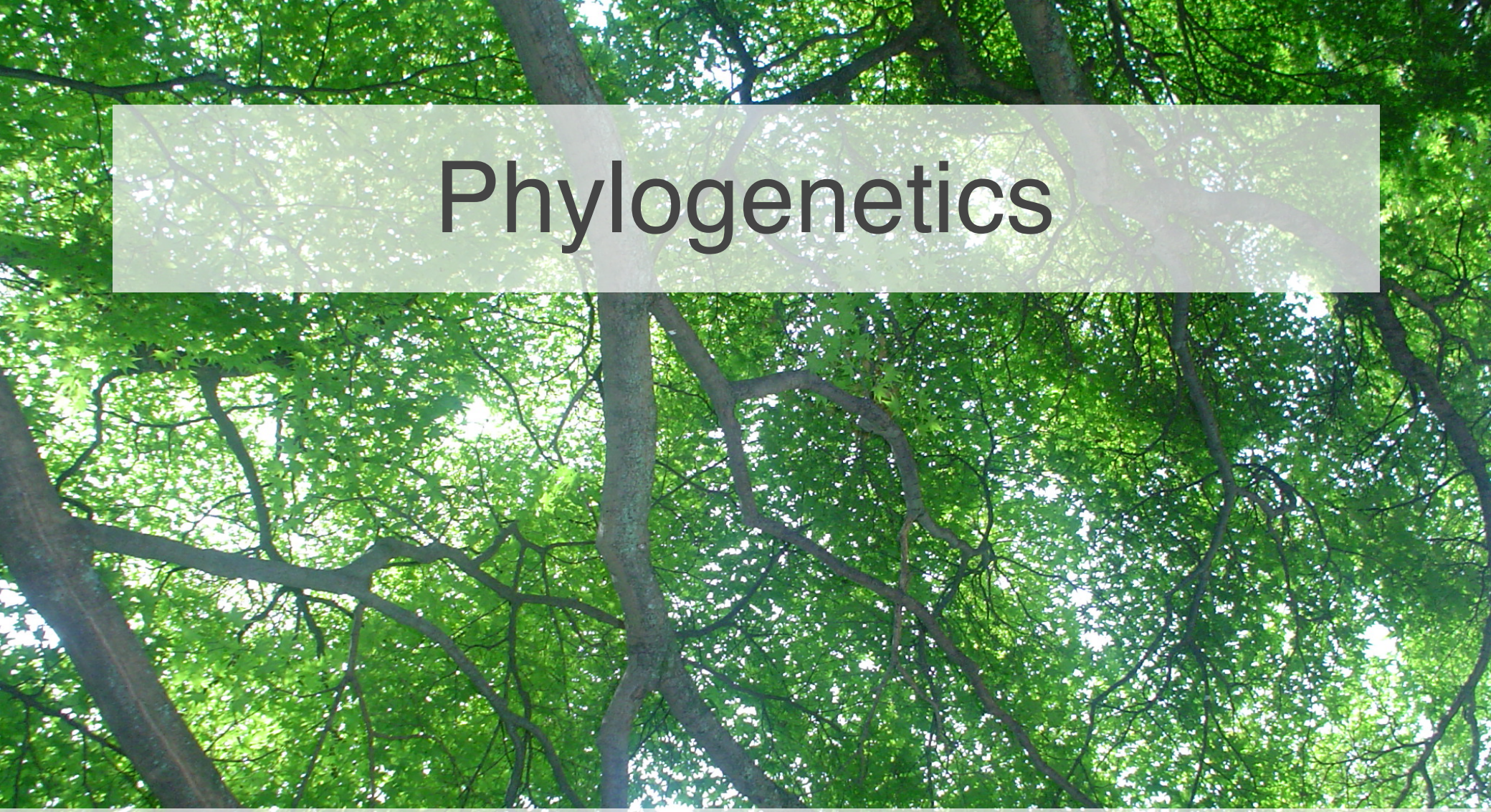
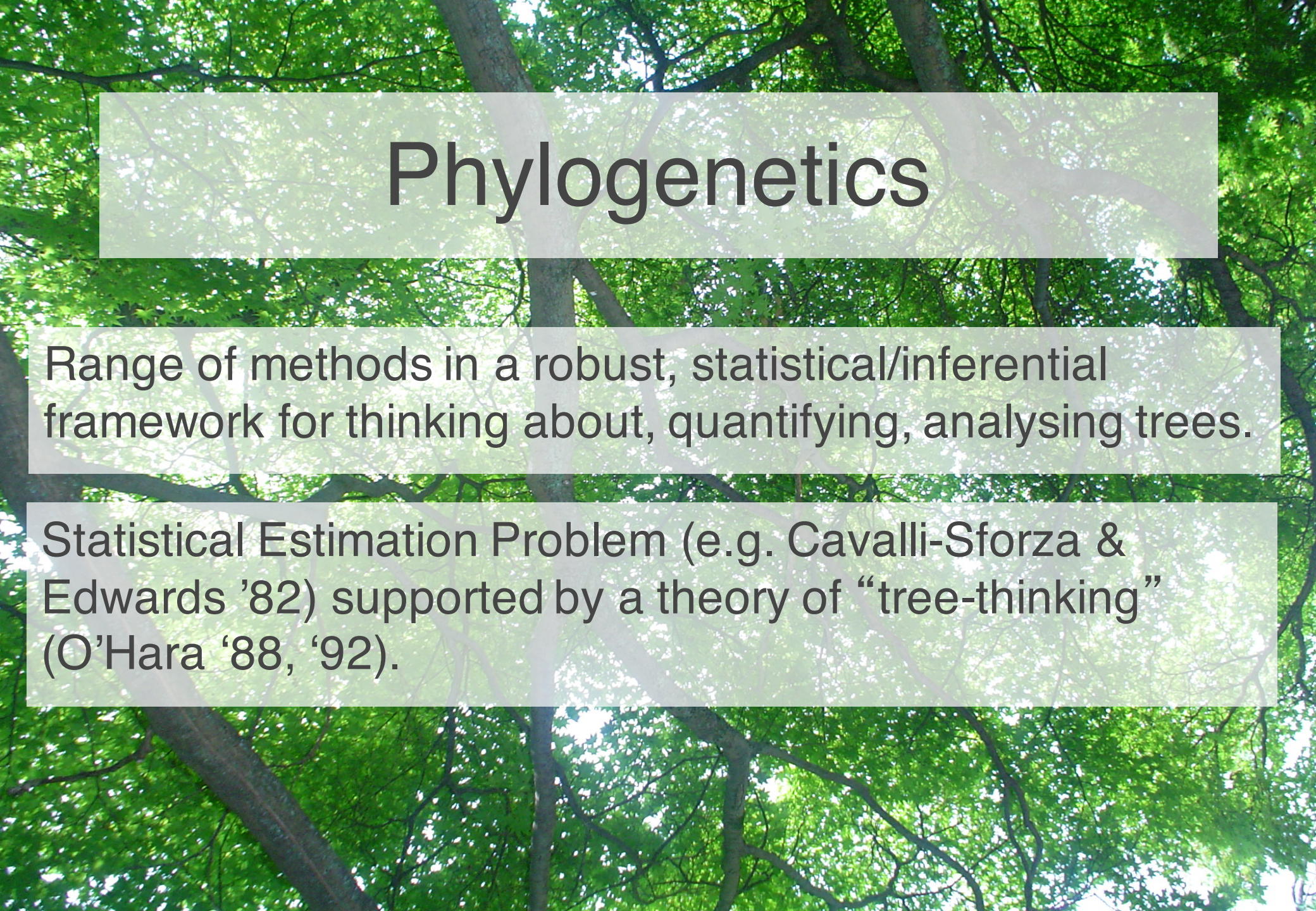
Phylogenetics



Simon J. Greenhill

ARC CENTRE OF EXCELLENCE FOR
THE DYNAMICS OF LANGUAGE

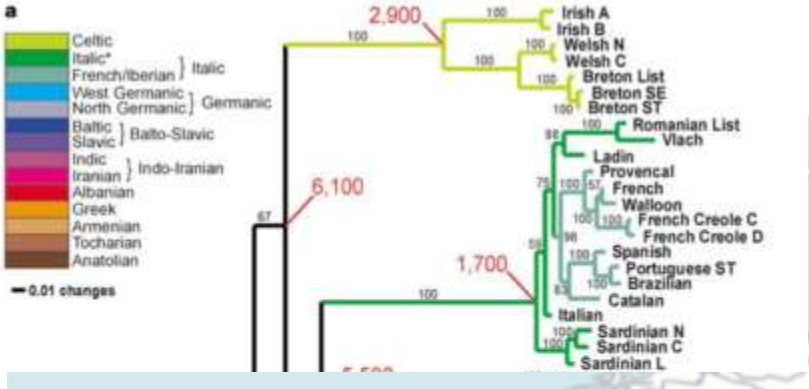
Max Planck Institute for the
Science of Human History

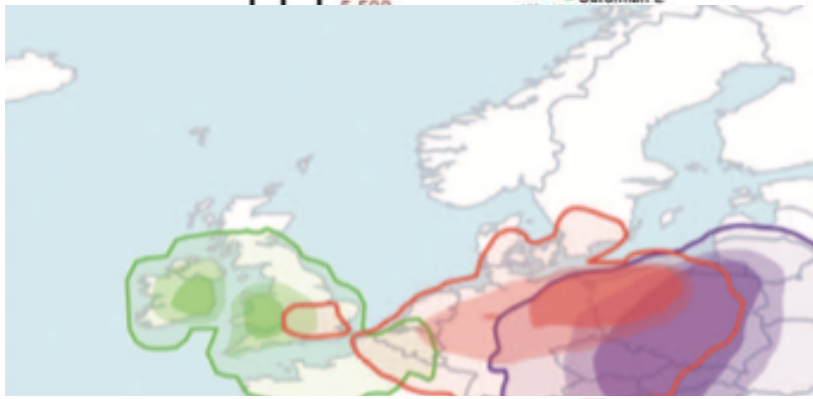


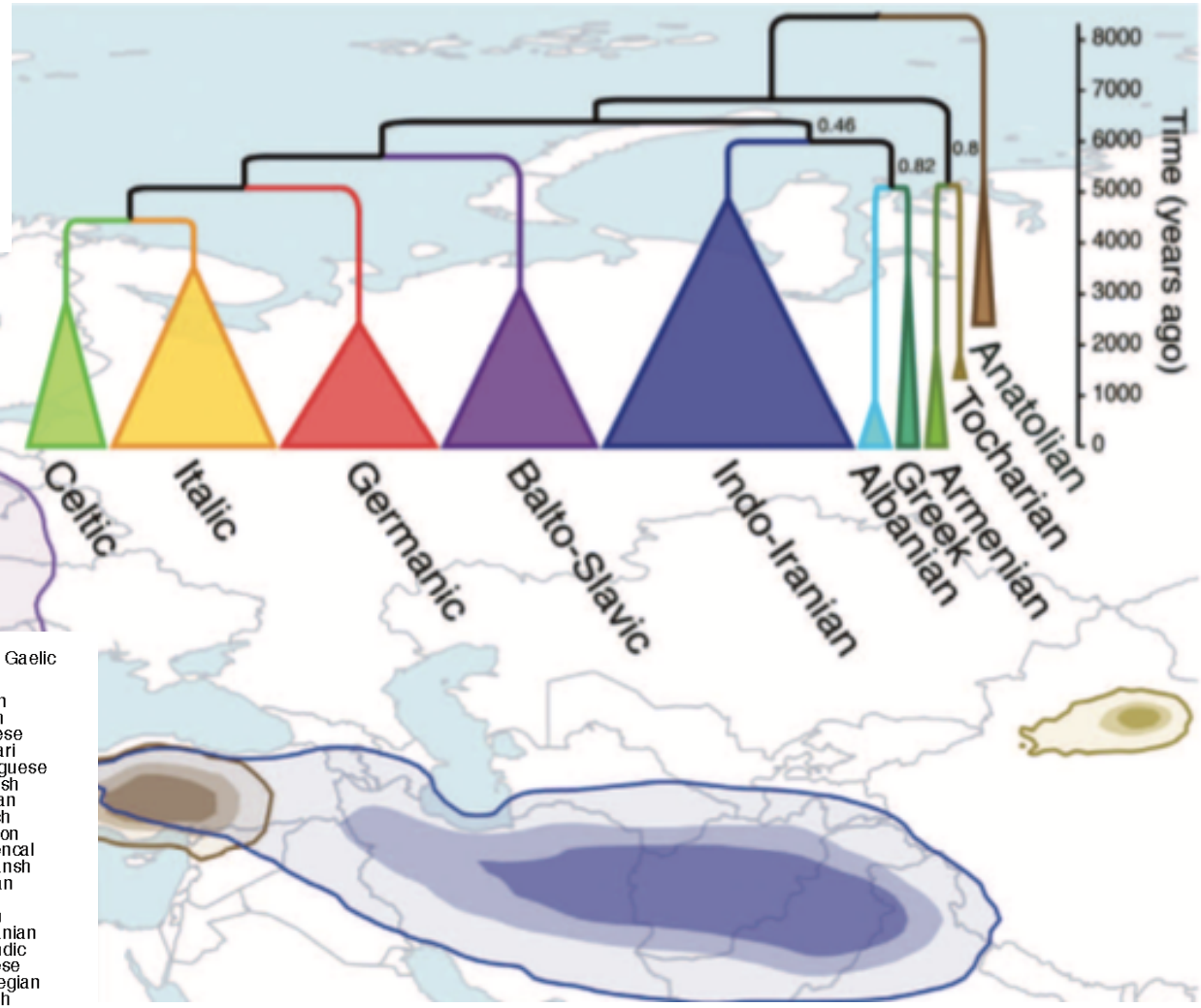
Phylogenetics

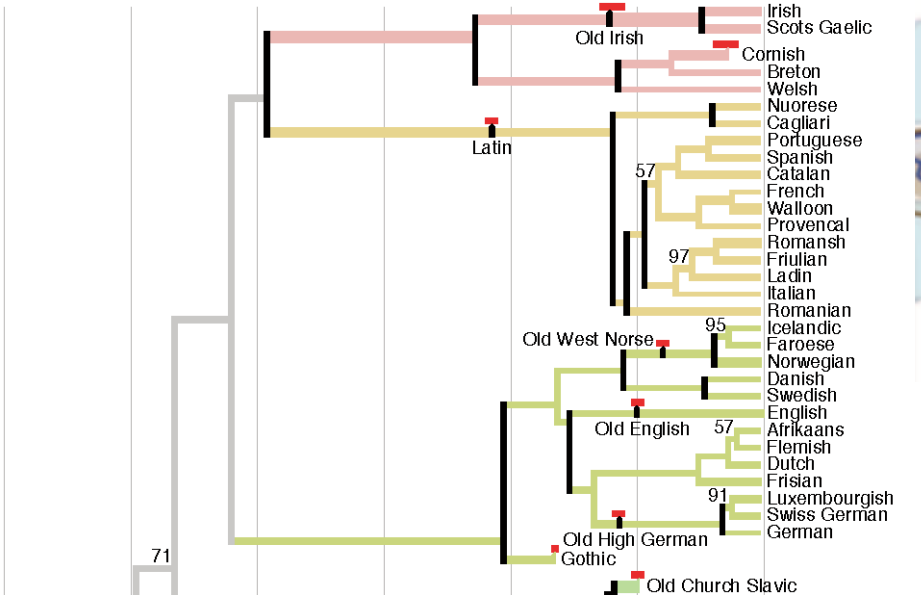
Range of methods in a robust, statistical/inferential framework for thinking about, quantifying, analysing trees.

Statistical Estimation Problem (e.g. Cavalli-Sforza & Edwards ’82) supported by a theory of “tree-thinking” (O’Hara ‘88, ‘92).

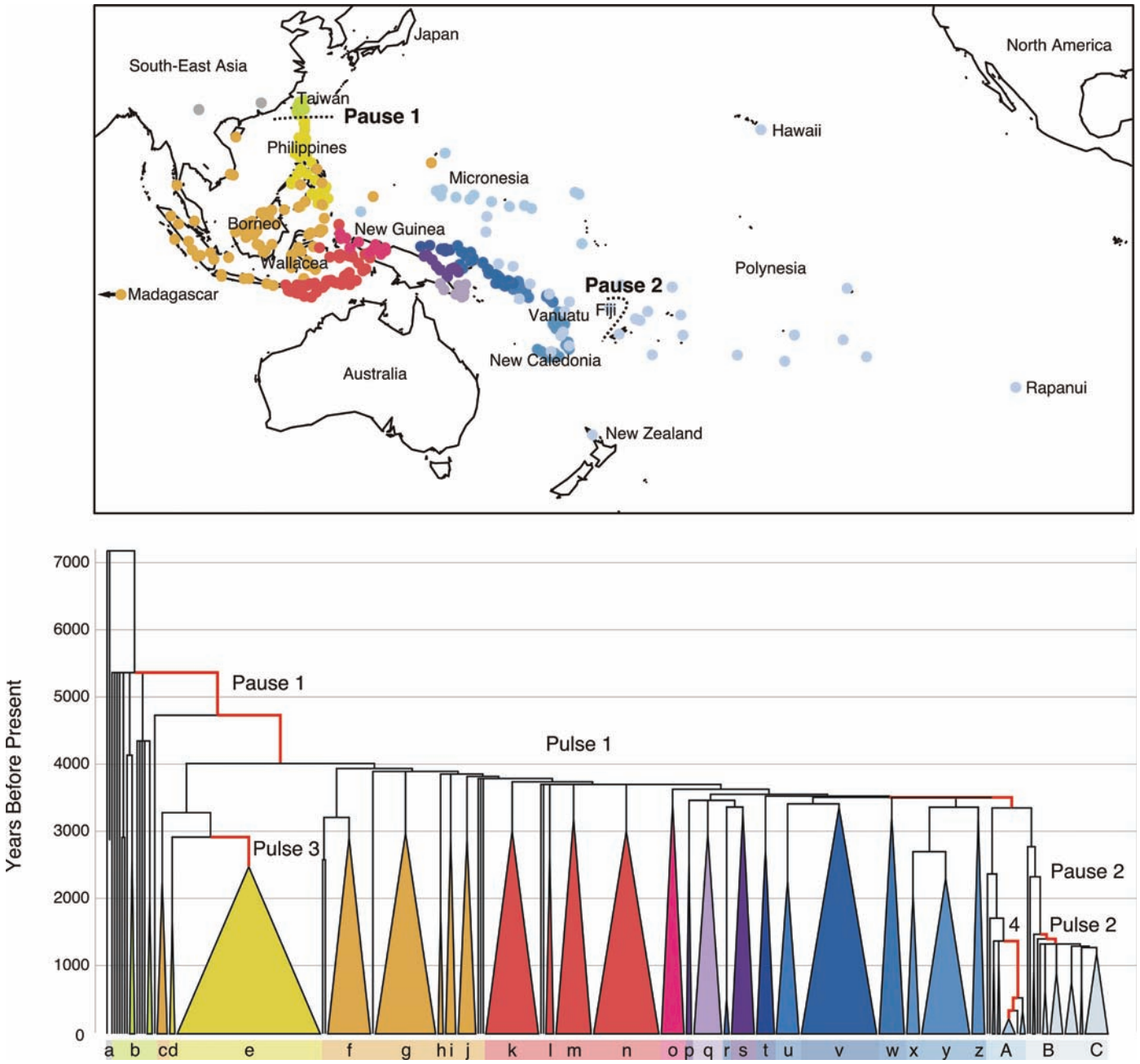




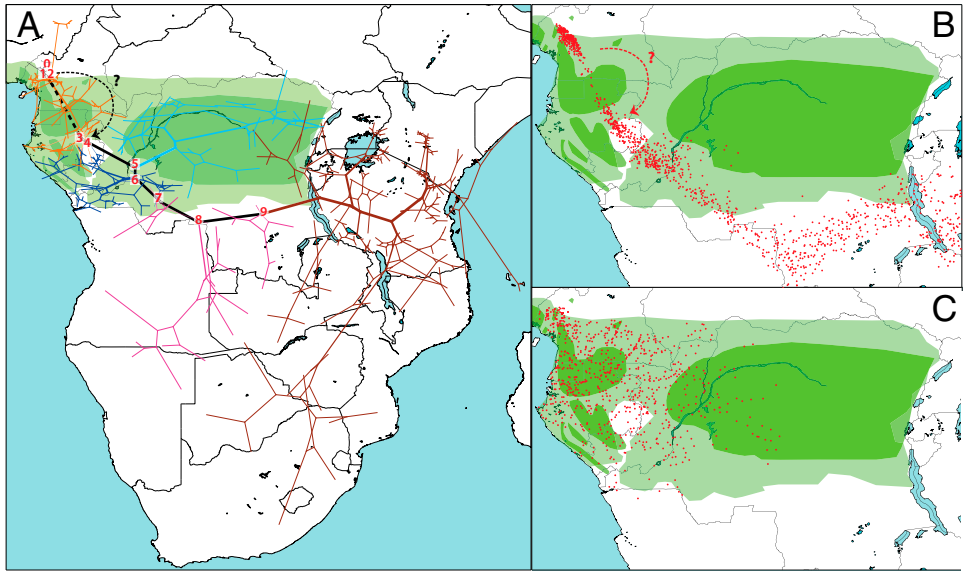


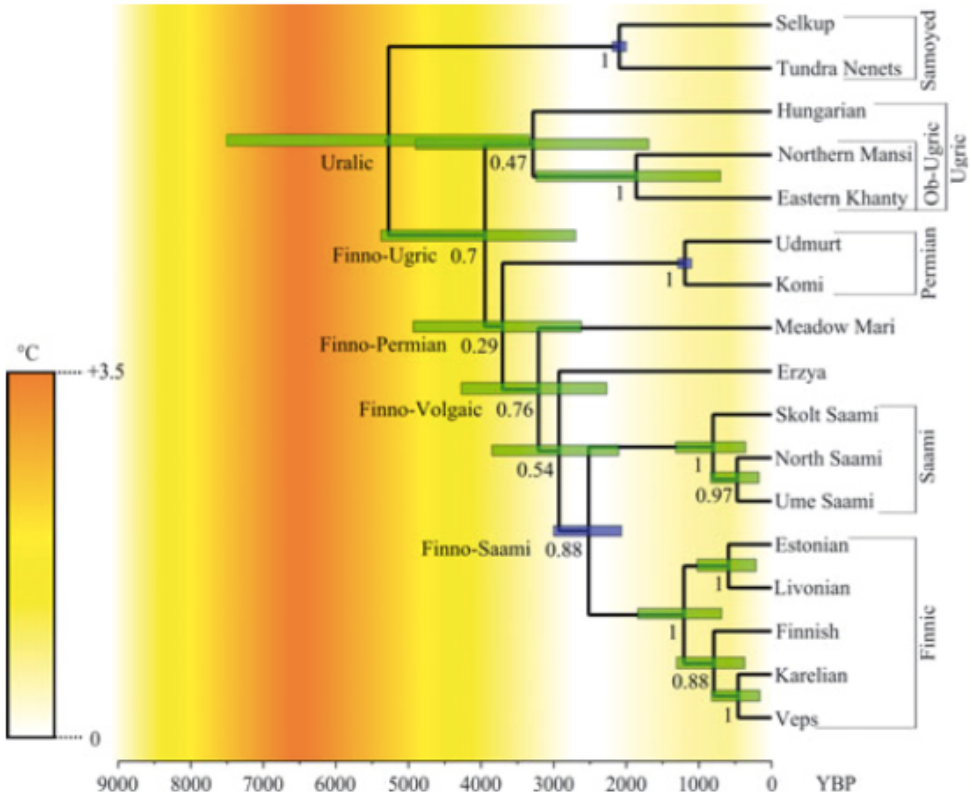


Gray & Atkinson ’03, Bouckaert et al’ 11, Chang et al ‘14.


Japan
South-East Asia
Taiwan
Pause 1
Philippines
Micronesia
Borneo
New Guinea
Wallacea
Madagascar
Australia
Pause 2
Vanuatu
Fiji
New Caledonia
North America
Hawaii
Polynesia
Rapanui
New Zealand
Years Before Present
7000
6000
5000
4000
3000
2000
1000
0
Pause 1
Pulse 1
Pulse 3
Pause 2
4
Pulse 2
a b cd e f g hi j k l m n o p q r s t u v w x y z A B C

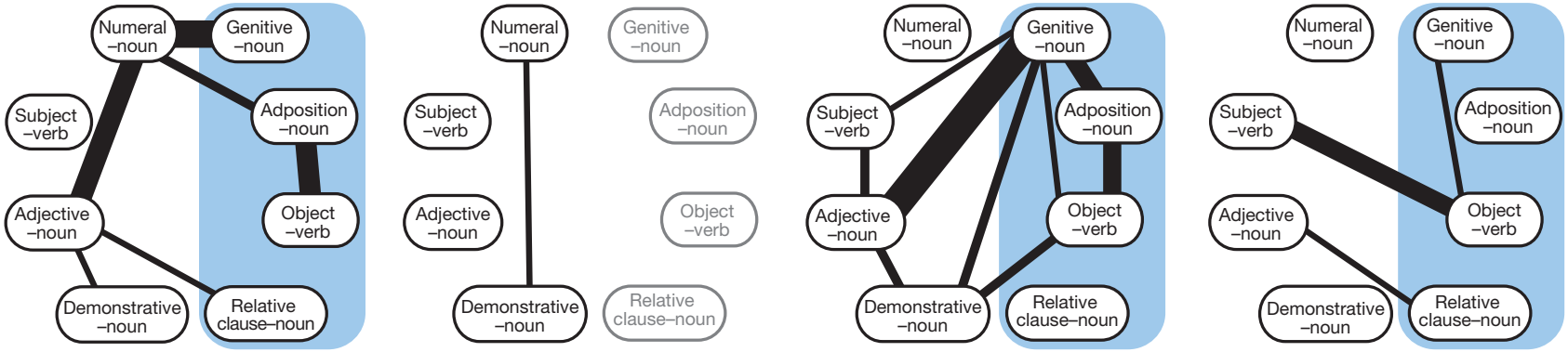
Gray et al '09



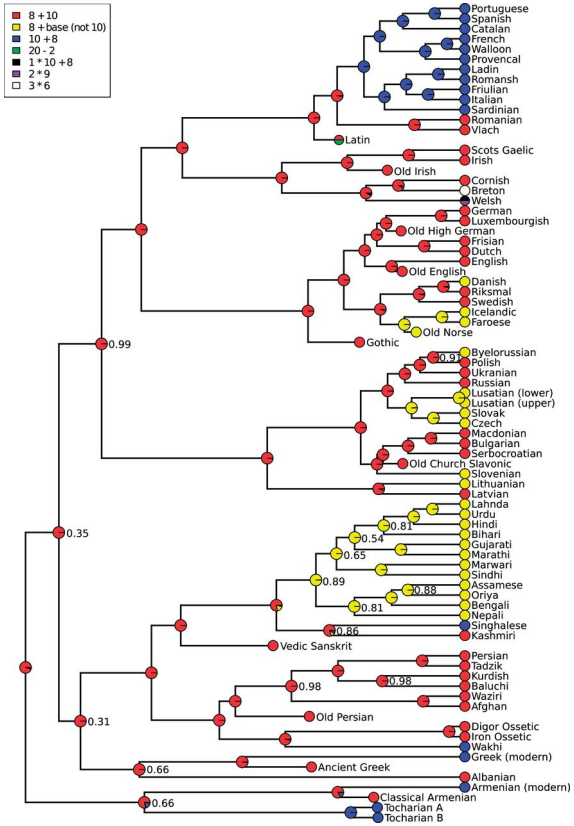


Grollemund et al. ‘15

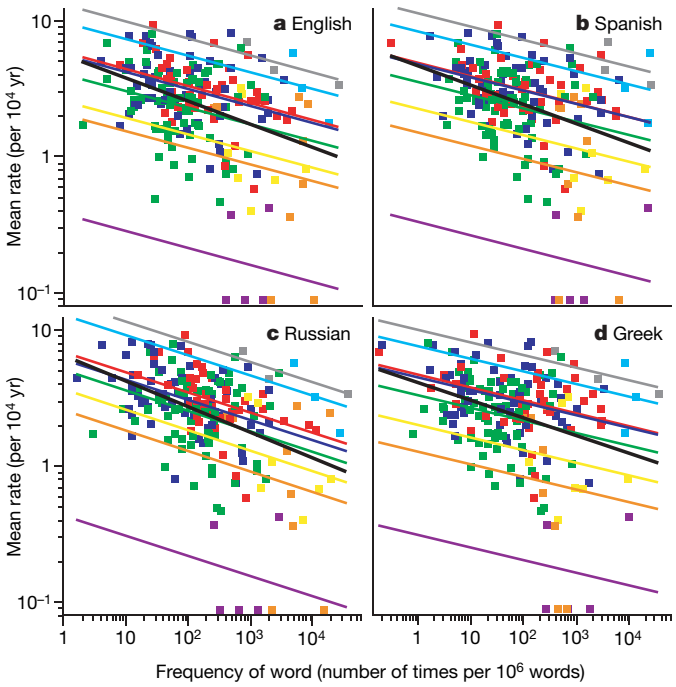
Honkola et al ‘13



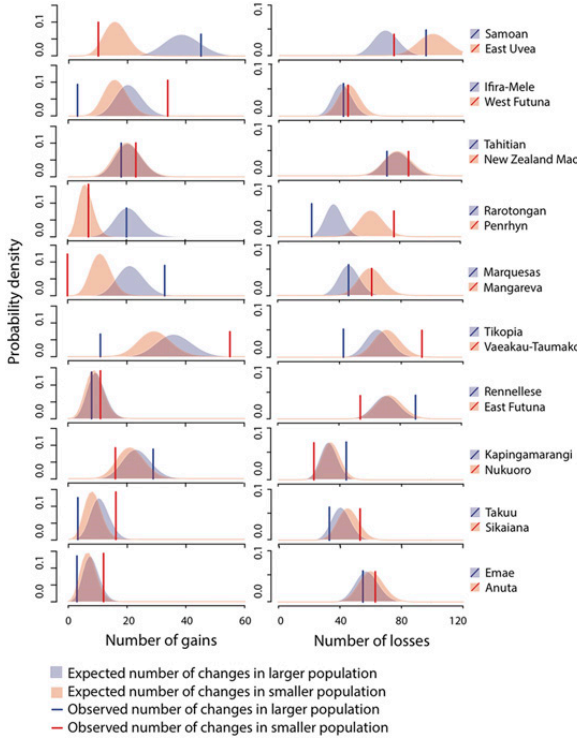
Dunn et al. ‘15



Calude and Verkerk ‘16

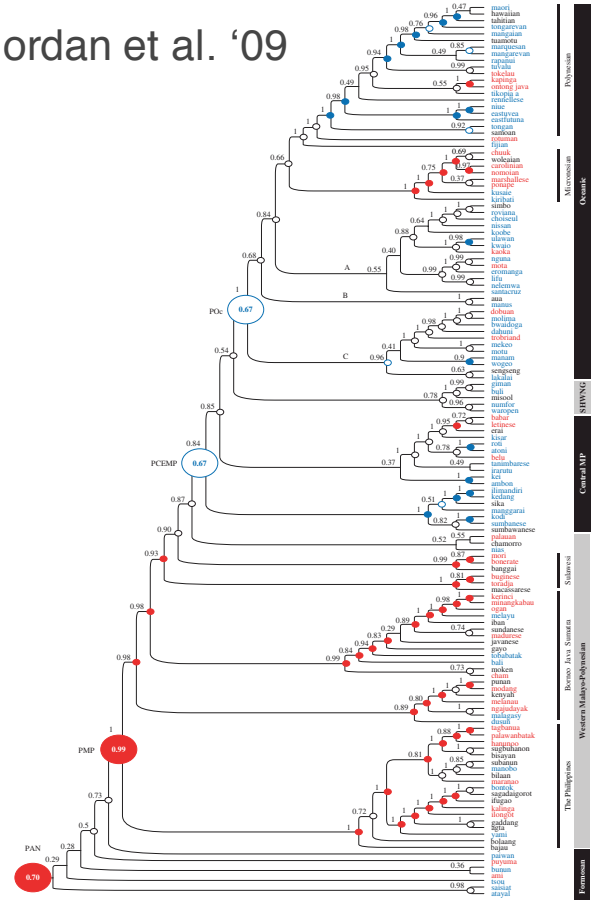


Pagel et al ’07

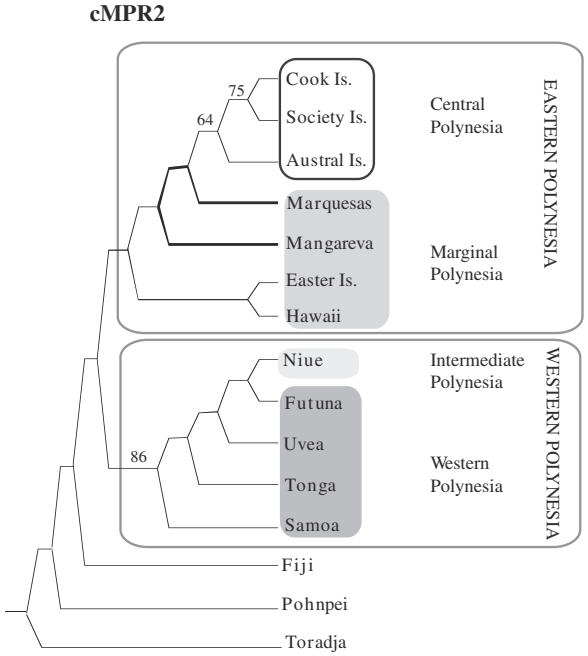


Bromham et al. ’15

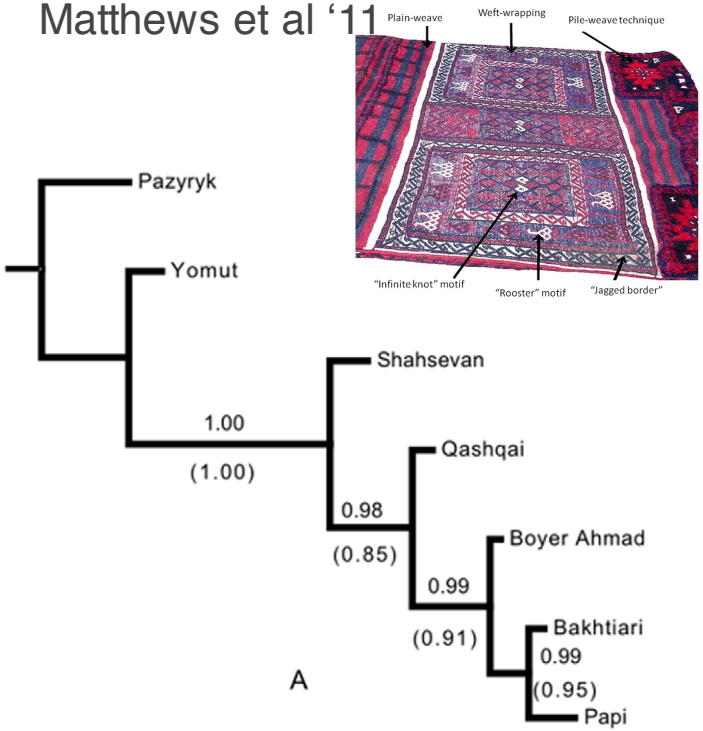
Jordan et al. ‘09

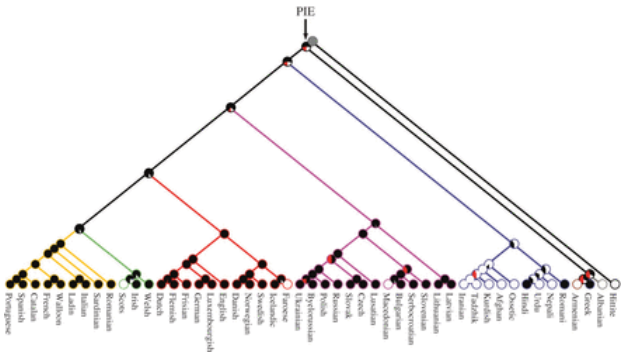


Larsen ‘11

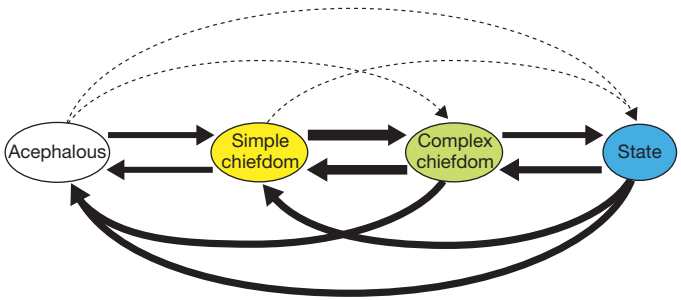


Matthews et al ‘11

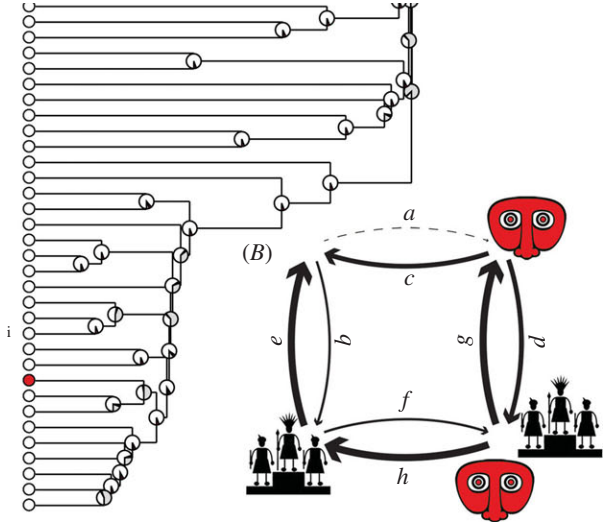




Da Silva & Tehrani ‘16



Currie et al. ‘10



Watts et al. ‘15

Controversial

"most vibrant stream of contemporary linguistics"

"Computational methodologies of this kind can only be helpful for historical linguistics"

"languages and biomolecular sequences evolve in very different ways"

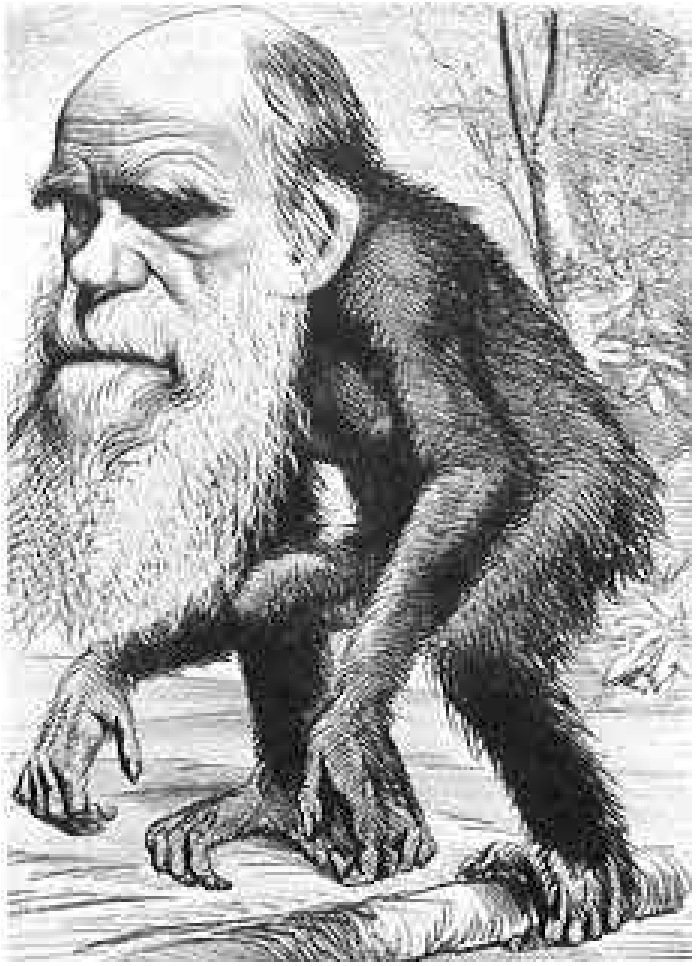
"more questions than answers"

"utter bollocks"

"biggest intellectual fraud since Chomsky"

"this isn't history, it's history put in nested boxes!"

What is **evolution**?



Variation

Heritability

Differential survival

When and where did 🐨👱💬 **originate**?

What **differences** are there between 🐨👱💬's?

How are 🐨👱💬 **related** to other 🐨👱💬's?

What **processes** shaped 🐨👱💬?

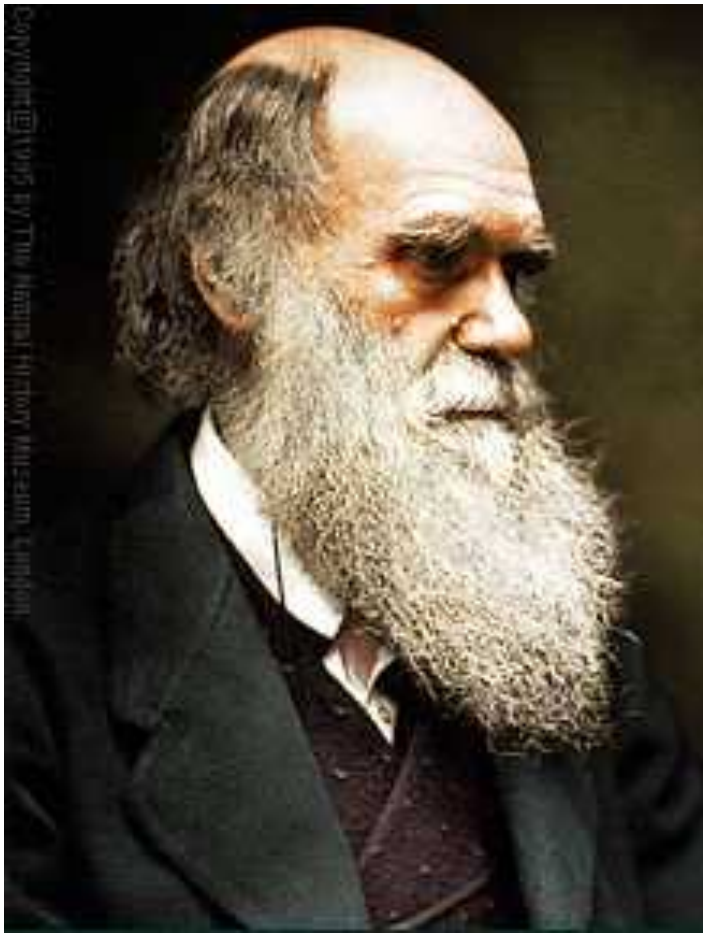
Can we infer what 🐨👱💬 were in the **past**?

Conceptual Parallels

- 🐨 Speciation
- 🐨 Homologies
- 🐨 Mutations
- 🐨 Random Drift
- 🐨 Natural selection
- 🐨 Hybridisation
- 🐨 Fossils
- 🐨 Extinction

- 👱💬 Divergence
- 👱💬 Cognates
- 👱💬 Innovations
- 👱💬 Random Drift
- 👱💬 “Social” selection
- 👱💬 Borrowing
- 👱💬 Ancient Texts
- 👱💬 Death

Descent of Man (1871)



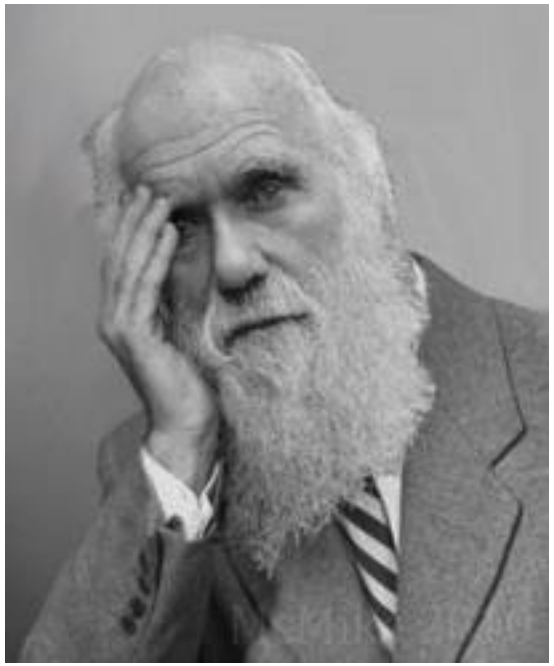
"Languages, like organic beings, can be classed in groups under groups…

Dominant languages and dialects spread widely, and lead to the gradual extinction of other tongues. A language, like a species, when once extinct … never reappears."

…

"The formation of different languages and of distinct species, and the proofs that both have been developed through a gradual process, are **curiously parallel**"

Darwin was late



~400: BCE

Socrates & Aristotle recognised Greek had changed since Homer (730 BCE)

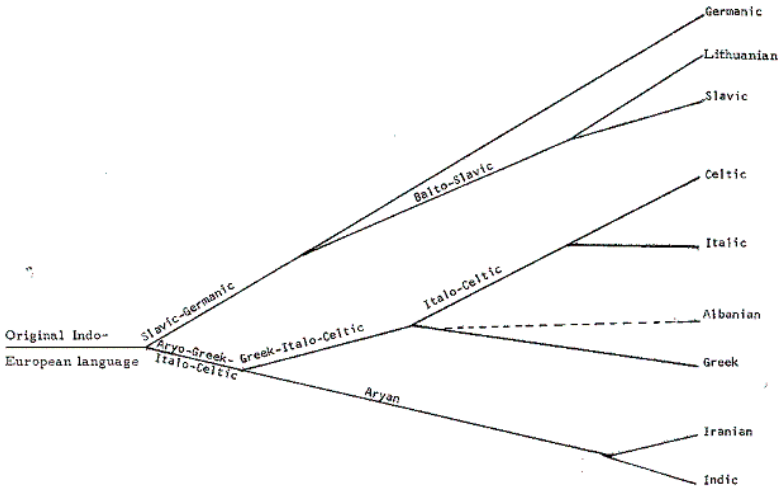
1786:

Sir William Jones: Sanskrit languages have “sprung from some common source”

1814:

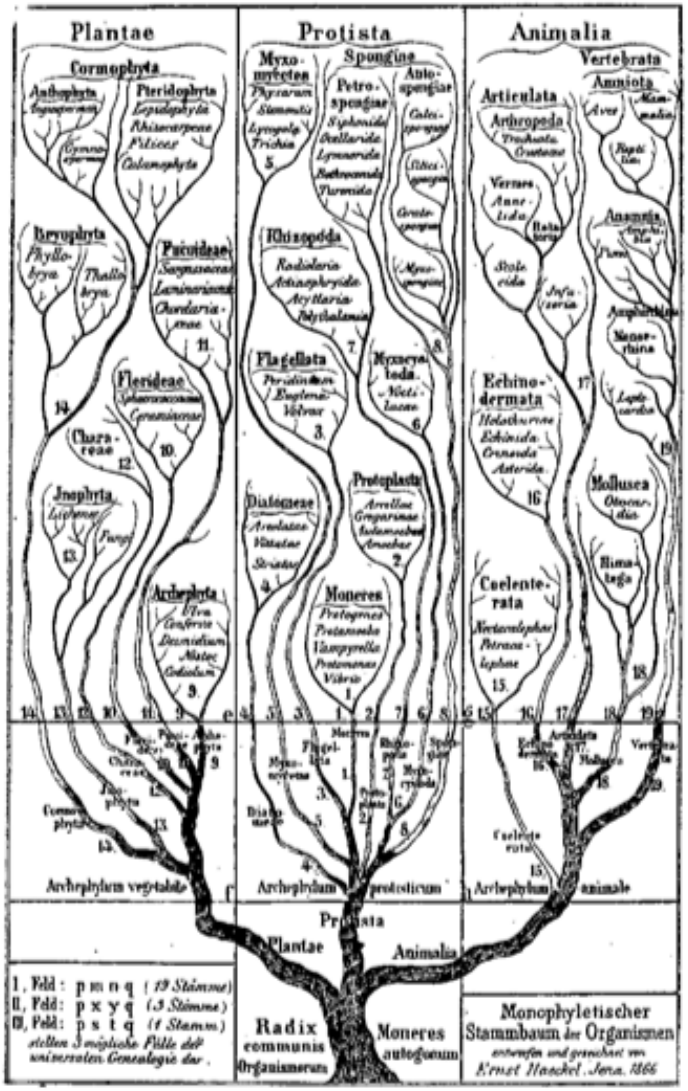
Rasmus Rask: Comparative Method

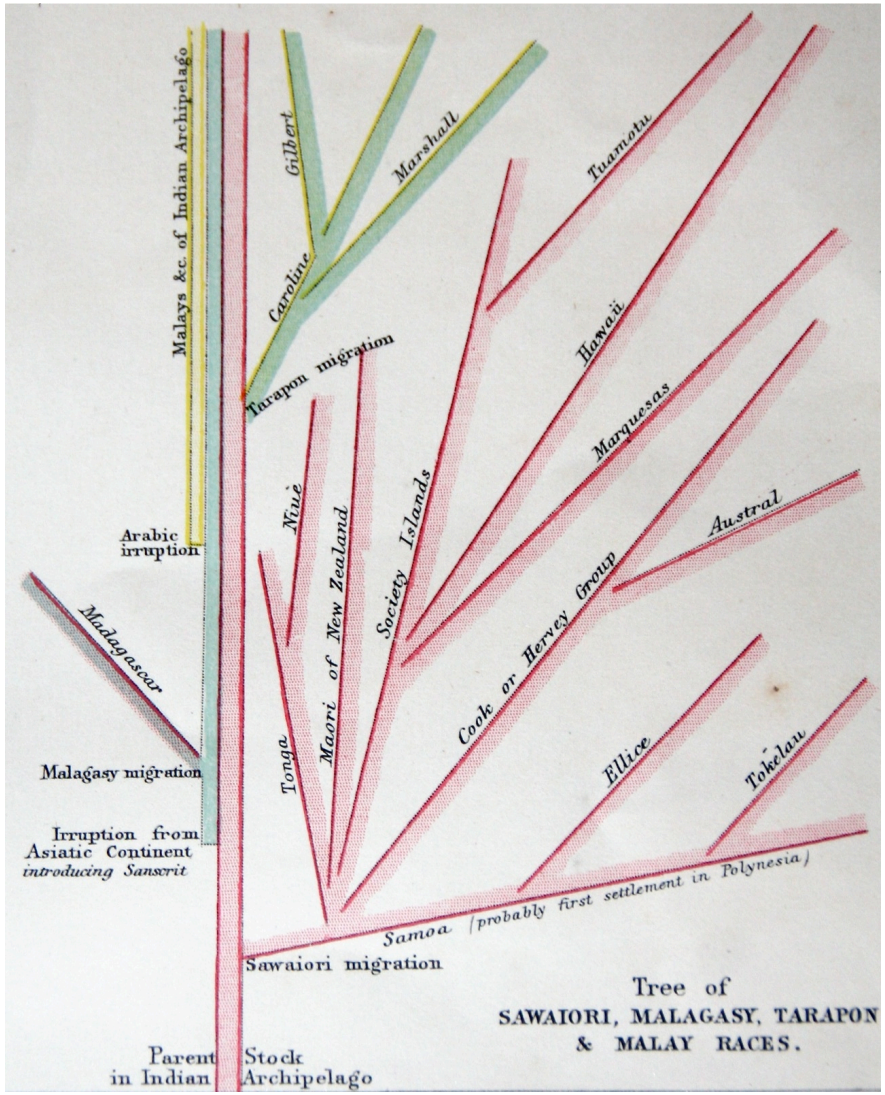
August Schleicher 1863
Darwinism Tested by the Science of Language



"same process has long been generally assumed for linguistic organisms"

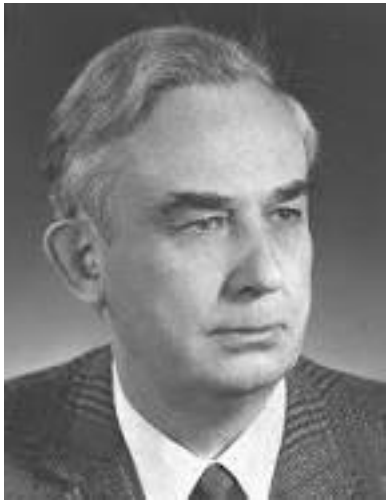
"We set up family trees of languages known to us in precisely the same way as Darwin has attempted to do for plant and animal species."


Plantae
Protista
Animalia
Cormophyta
Spongiae
Vertebrata
Amniota
Articulata
Arthropoda
Echinodermata
Coelenterata
Mollusca
Moneres
Flagellata
Rhizopoda
Diatomeae
Protoplasta
Archephyta
Archephylum vegetabile
Archephylum protisticum
Archephylum animale
Protista
Plantae
Animalia
Radix communis Organismorum
Moneres autogenum
Monophyletischer Stammbaum der Organismen
entworfen und gezeichnet von
Ernst Haeckel. Jena. 1866


Malays &c. of Indian Archipelago
Gilbert
Marshall
Caroline
Tarapon migration
Tuamotu
Hawaii
Marquesas
Austral
Society Islands
Niuë
Maori of New Zealand
Tonga
Cook or Hervey Group
Ellice
Tokelau
Samoa (probably first settlement in Polynesia)
Sawaiori migration
Arabic irruption
Madagascar
Malagasy migration
Irruption from Asiatic Continent introducing Sanscrit
Parent Stock in Indian Archipelago
Tree of SAWAIORI, MALAGASY, TARAPON & MALAY RACES.

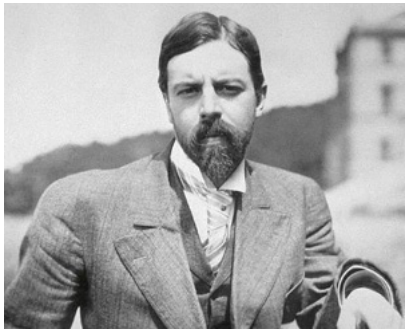
Brugmann (1884)

Importance of using “shared innovations” to identify clades and not “shared retentions”



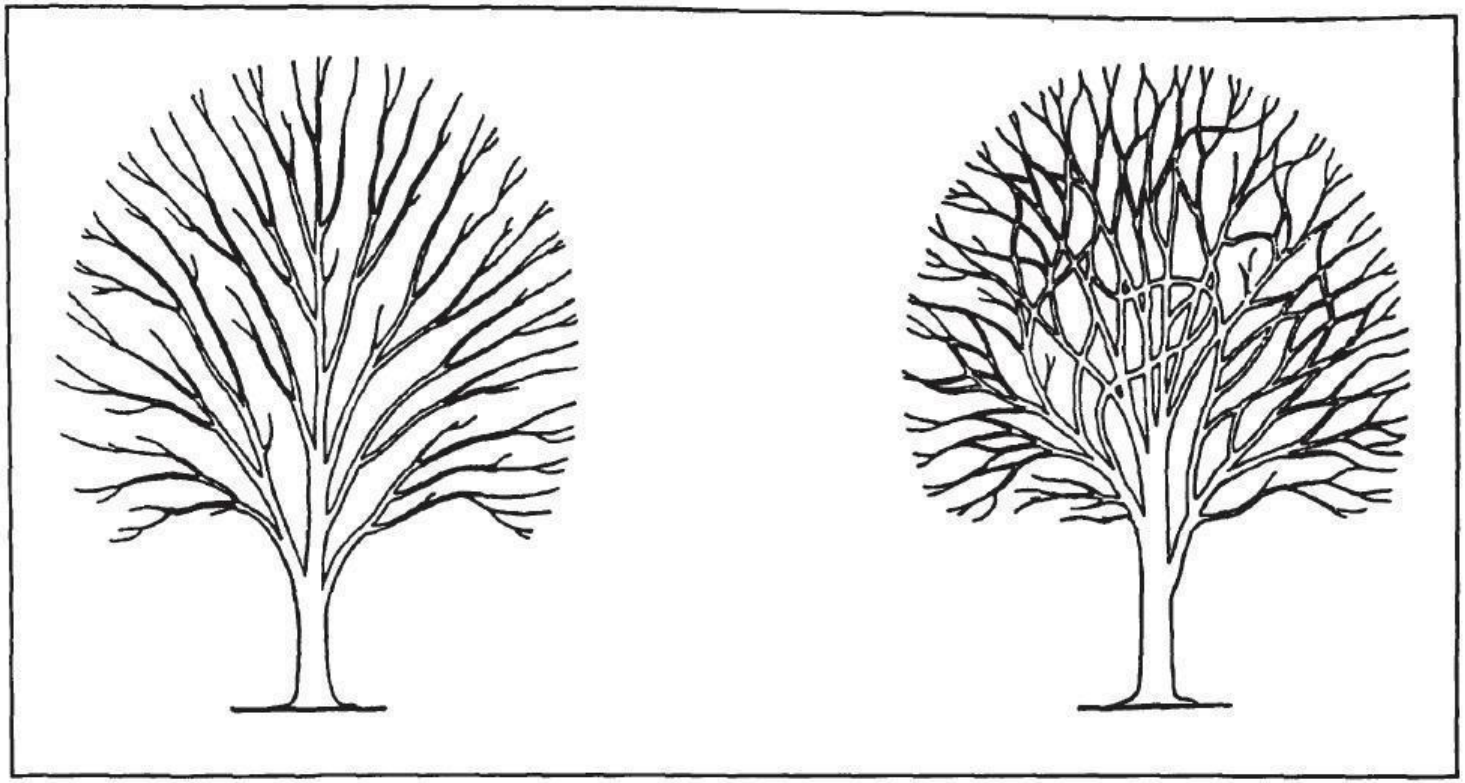
Hennig (1966)

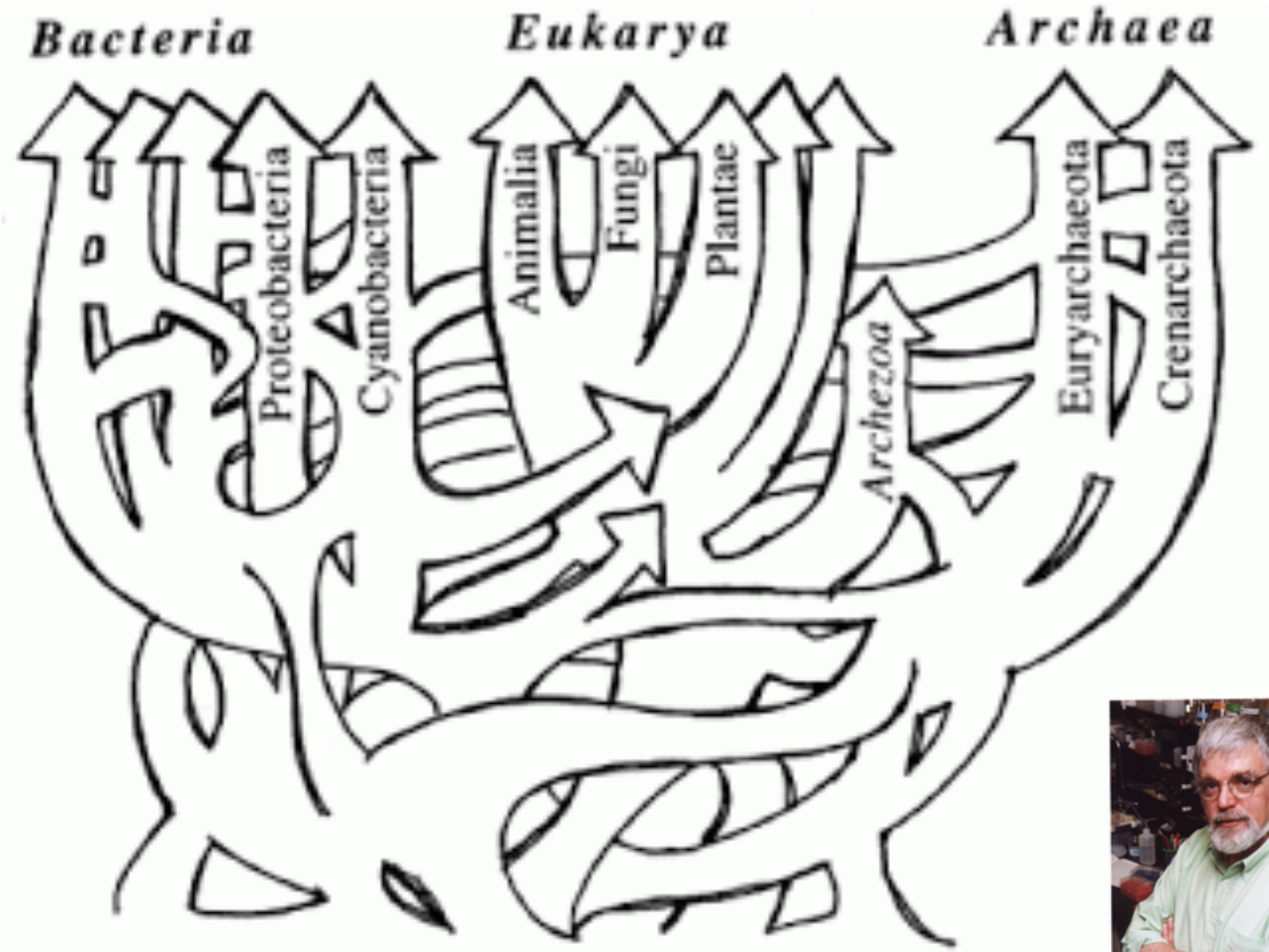
“Synapomorphies” and
“Symplesiomorphies”



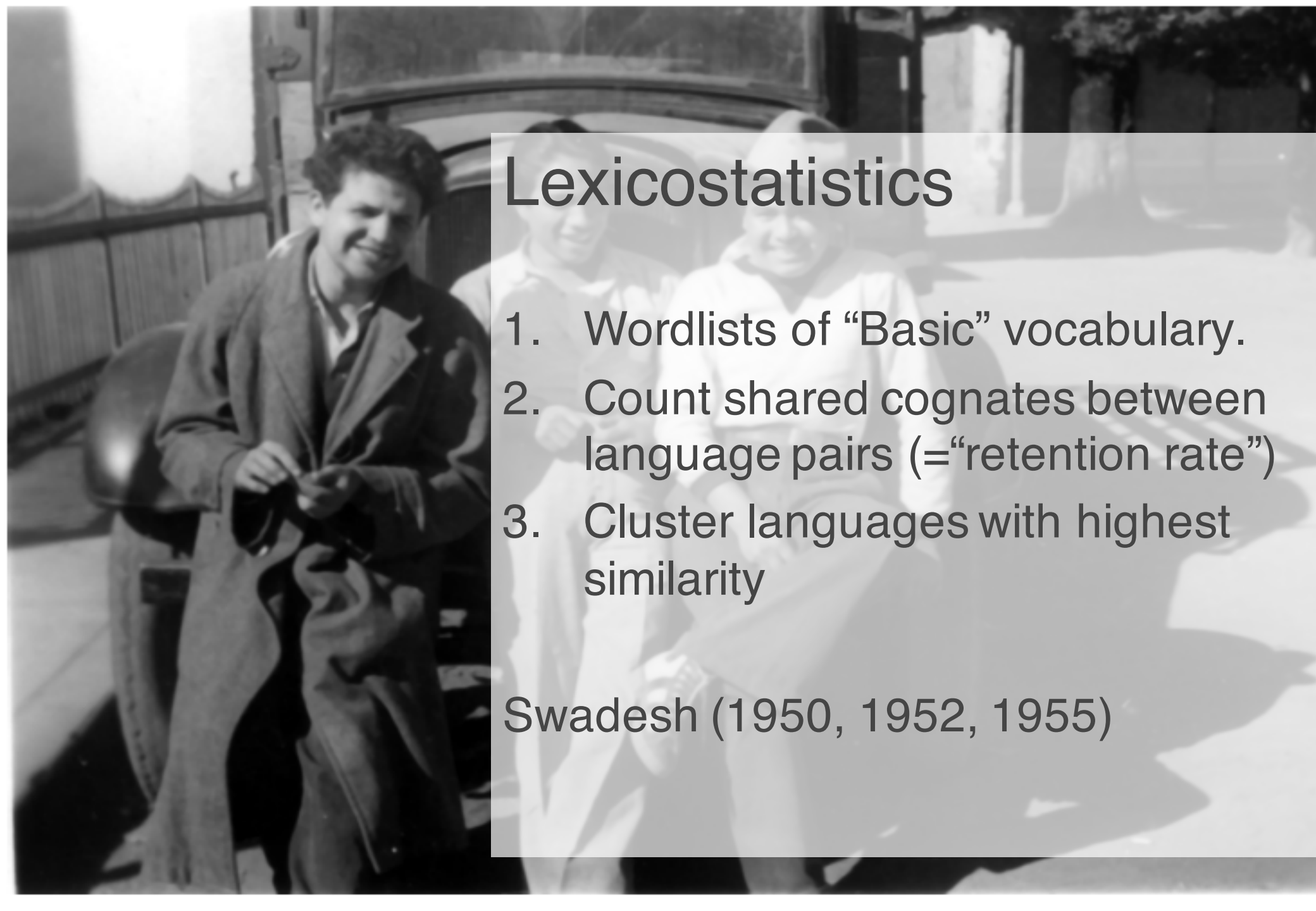
Kroeber (1948)

"The tree of life is eternally branching, and never doing anything fundamental but branching, except for the dying-away of branches. The tree of human history, on the contrary, is constantly branching and at the same time having its branches grow together again. Its plan is therefore much more complex and difficult to trace." (p.86)




Bacteria
Eukarya
Archaea
Proteobacteria
Cyanobacteria
Animalia
Fungi
Plantae
Archezoa
Euryarchaeota
Crenarchaeota


IBM



Lexicostatistics

1. Wordlists of “Basic” vocabulary.
2. Count shared cognates between language pairs (=“retention rate”)
3. Cluster languages with highest similarity

Swadesh (1950, 1952, 1955)

	Taboo	Blood	To Suck
Fijian	tabu	drā	sucu-ma
Tahitian	tapu	toto	ngote
Maori	tapu	toto	ngote
Hawaiian	kapu	koko	omo
Marquesan	tapu	toto	omo

Identified by Systematic Sound Correspondences
- e.g. Maori“t” = “k” in Hawaiian.

Elbert 1953

TABLE 2

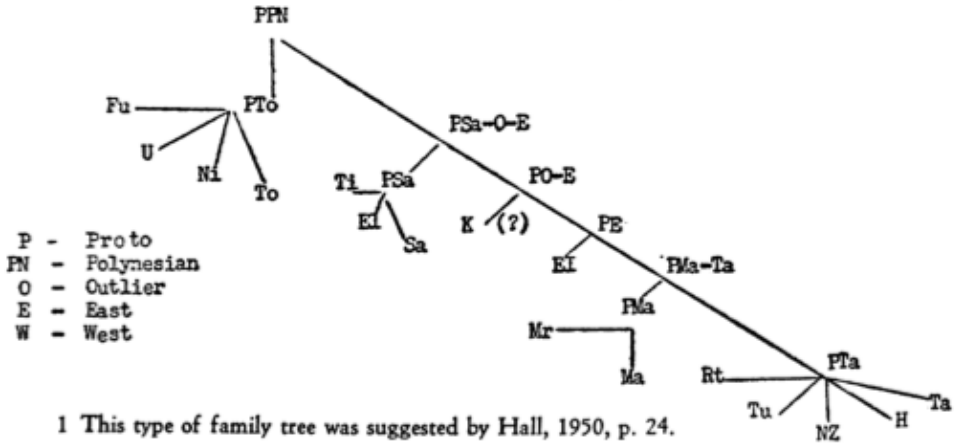
Polynesian cognate percentages

U	Ni	To	Ti	E	Sa	Si[1]	Fi[1]	OJ[1]	Nu[1]	K	EI	Mr	Ma	Rt	Tu	NZ	H	Ta	
83	62	74	83	79	74	57	62	62	54	52	54	57	56	66	62	61	58	58	Fu
	72	86	78	74	70	53	59	58	52	51	53	53	51	62	62	61	55	59	U
		64	68	61	63	50	59	49	54	49	49	55	47	56	55	54	49	51	Ni
			70	64	66	46	55	55	49	45	48	49	45	58	53	54	49	52	To
				81	76	66	66	62	59	59	62	67	63	71	68	71	67	66	Ti
					78	66	61	62	59	58	61	62	63	66	66	67	68	66	E
						64	60	61	55	53	53	55	52	67	62	57	59	60	Sa
							55	60	52	55	52	55	56	60	58	60	60	59	Si
								57	52	54	54	59	53	55	61	60	55	54	Fi
									51	54	53	53	51	53	56	55	56	53	OJ
										53	49	45	44	53	50	52	49	48	Nu
											47	49	45	54	51	51	49	50	K
												64	63	64	62	63	64	62	EI
													73	75	72	70	69	68	Mr
														73	69	67	70	67	Ma
															83	83	79	85	Rt
																79	77	83	Tu
																	71	73	NZ
																		76	H

1 Percentages based on incomplete data.

TABLE 4

A tentative family tree for Polynesia[1]



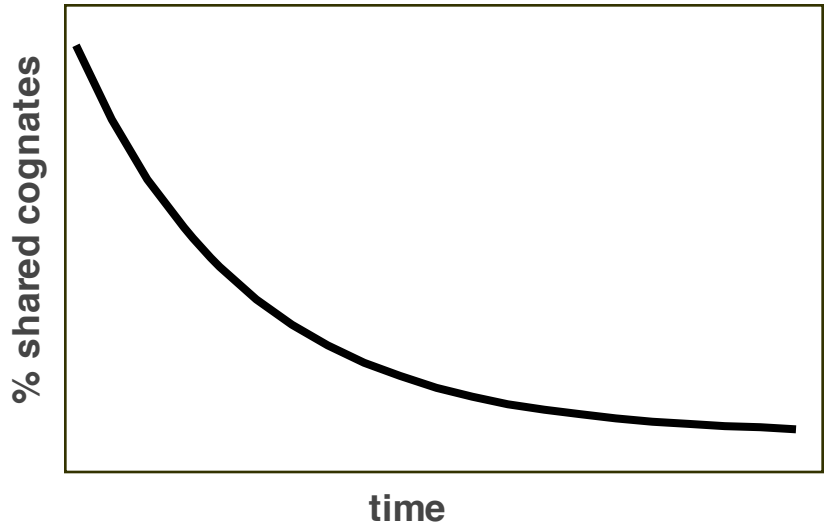
1 This type of family tree was suggested by Hall, 1950, p. 24.

Glottochronology

Loss of cognates happens at a constant rate (=radioactive decay)

19% loss per 1000 years (Lees 1953)

$$time = \frac{\log(\%sharedcognates)}{2\log(retentionrate)}$$



The Rise of Lexicostatistics…

IN THE LAST DECADE glottochronology has excited international interest and acquired a literature of its own. To the anthropologist it promises a measure of time depth for language families without documented history, and yet another linguistic example of regularity in cultural phenomena.

Hymes (1960): "Lexicostatistics so far"

"... a significant work—one which may conceivably be as revolutionary for Oceanic linguistics and culture history as was the work of Greenberg (1949–54) for the interpretation of African languages and cultures"

Murdock (1964) p.117

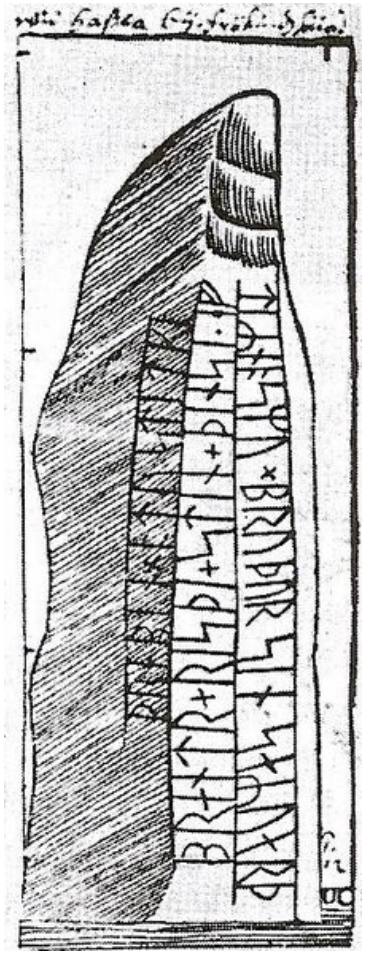
…and the fall of Lexicostatistics

Major Criticism: Universality of Rates

Old Norse & Icelandic?

- Glottochronology: 200 years.
- Reality: 1000 years

Bergsland & Vogt 1962: “Our findings clearly disprove the basic assumption of glottochronology 'that fundamental vocabulary changes at a constant rate’ ”



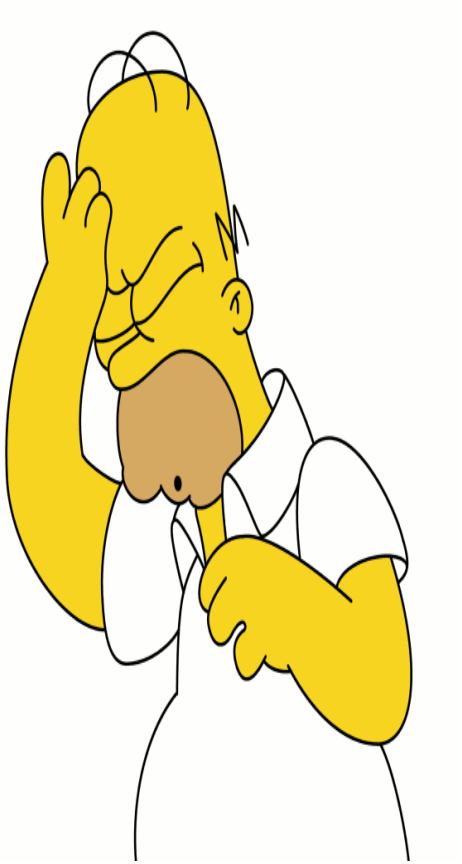
Jungner, Hugo; Elisabeth Svärdström (1940-1971). Sveriges runinskrifter: V. Västergötlands runinskrifter. Stockholm: Kungl. Vitterhets Historie och Antikvitets Akademien. ISSN 0562-8016. p. 260

Fallout.

"a tradition of hostility towards probabilistic modelling in historical linguistics" (Sankoff '73)

"In summary, glottochronology is not accurate; all its basic assumptions have been severely criticized. It should not be accepted, it should be rejected" (Campbell '04)

"Linguists don't do dates" (McMahon & McMahon '03)



U.P.G.M.A

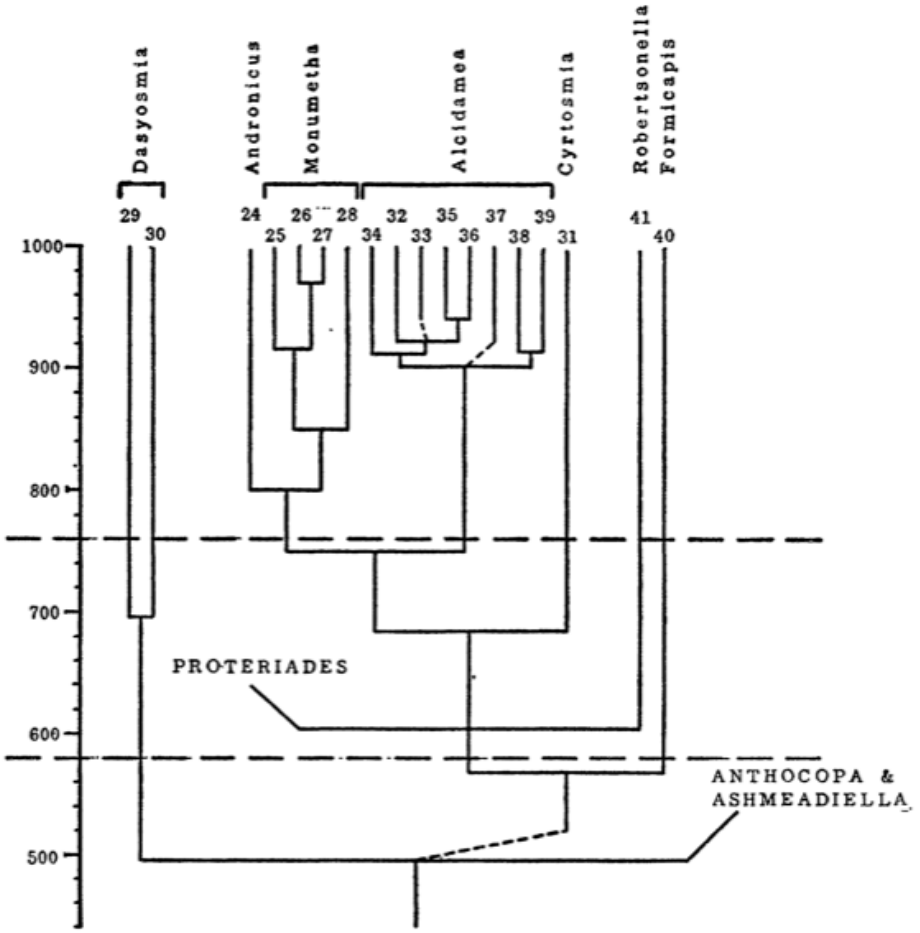


FIG. 6. Diagram of relationships for the genus *Hoplitis* obtained by the weighted variable group method.

A QUANTITATIVE APPROACH TO A PROBLEM IN CLASSIFICATION [1]

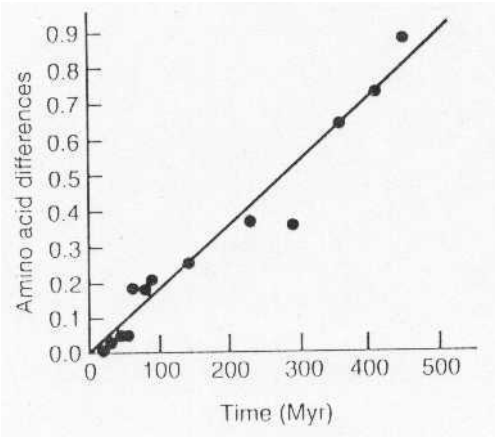
CHARLES D. MICHENER AND ROBERT R. SOKAL [2]

Department of Entomology, University of Kansas, Lawrence

Molecular Clock

Zuckerkandl & Pauling 1962

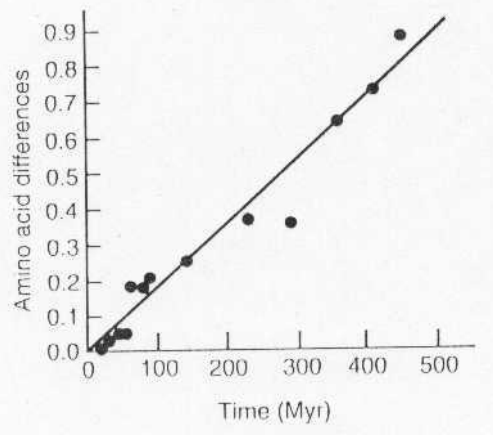
Number of AA differences were proportional to species divergence times.



Molecular Clock

Zuckerkandl & Pauling 1962

Number of AA differences were proportional to species divergence times.



Kimura 1968

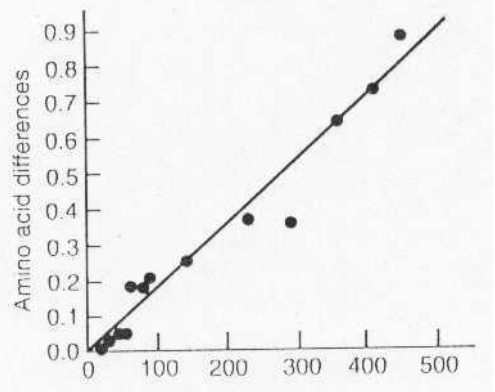
The average time taken for one base pair replacement within a genome is therefore

$$28 \times 10^6 \text{ yr} \div \left(\frac{4 \times 10^9}{300}\right) \div 1{\cdot}2 \doteqdot 1{\cdot}8 \text{ yr}$$

Molecular Clock

Zuckerkandl & Pauling 1962

Number of AA differences were proportional to species divergence times.



Kimura 1968

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Sarich & Wilson 1967

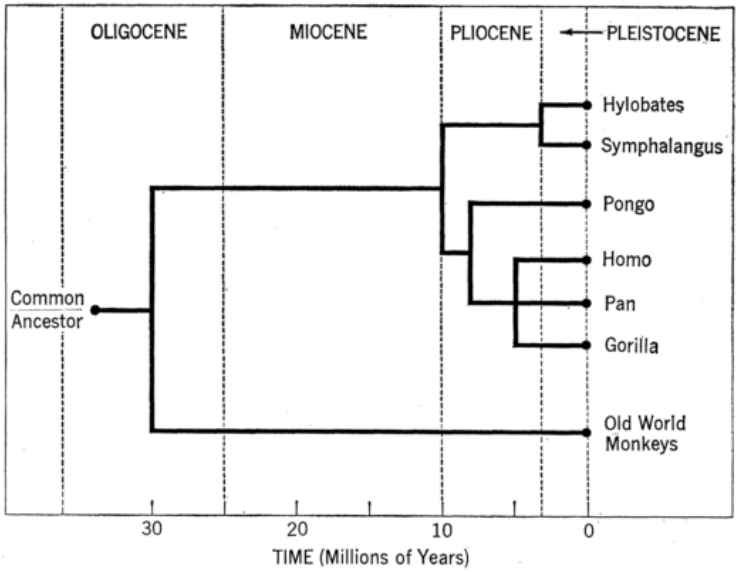

Fig. 1. Times of divergence between the various hominoids, as estimated from immunological data. The time of divergence of hominoids and Old World monkeys is assumed to be 30 million years.

Problems?

Kirsch 1969

SEROLOGICAL DATA AND PHYLOGENETIC INFERENCE: THE PROBLEM OF RATES OF CHANGE

JOHN A. W. KIRSCH

Felsenstein 1978

CASES IN WHICH PARSIMONY OR COMPATIBILITY METHODS WILL BE POSITIVELY MISLEADING[1]

JOSEPH FELSENSTEIN

Britten1986

Rates of DNA Sequence Evolution Differ Between Taxonomic Groups

ROY J. BRITTEN

The Cladistics Wars

Solutions.

Cavalli-Sforza & Edwards 1967

Phylogenetic Analysis
Models and Estimation Procedures

L. L. CAVALLI-SFORZA AND A. W. F. EDWARDS*

Yang 1993

Maximum-Likelihood Estimation of Phylogeny from DNA Sequences When Substitution Rates Differ over Sites[1]

Ziheng Yang

Sanderson 1997

A Nonparametric Approach to Estimating Divergence Times in the Absence of Rate Constancy

Michael J. Sanderson

Drummond et al. 2006

Relaxed Phylogenetics and Dating with Confidence

Alexei J. Drummond, Simon Y. W. Ho, Matthew J. Phillips, Andrew Rambaut

Joe Felsenstein

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Professor of Genome Sciences, and Professor of Biology, University of Washington, Seattle
Evolutionary biology, phylogenetic methods, population genetics
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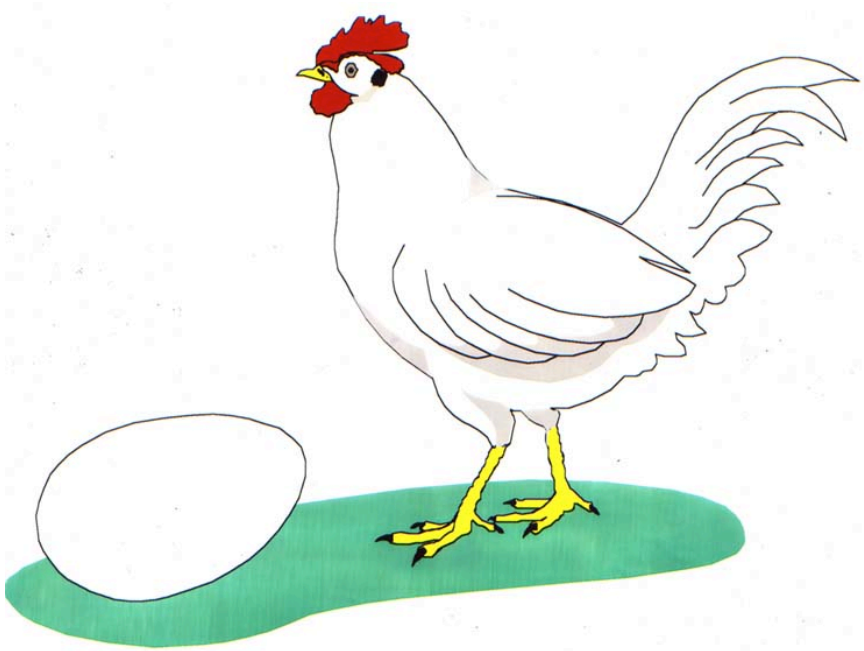
i10-index	121	73

Title 1–20	Cited by	Year
Confidence limits on phylogenies: an approach using the bootstrap J Felsenstein Evolution, 783-791	30037	1985
PHYLIP-phylogeny inference package (version 3.2) D Plotree, D Plotgram cladistics 5, 163-166	23173	1989
Evolutionary trees from DNA sequences: a maximum likelihood approach J Felsenstein Journal of molecular evolution 17 (6), 368-376	8494	1981
Phylogenies and the comparative method J Felsenstein American Naturalist, 1-15	6259	1985
Inferring phylogenies J Felsenstein, J Felenstein Sinauer Associates	3743	2004
Cases in which parsimony or compatibility methods will be positively misleading J Felsenstein Systematic Biology 27 (4), 401-410	3057	1978
Phylogenies from molecular sequences: inference and reliability J Felsenstein	2252	1988

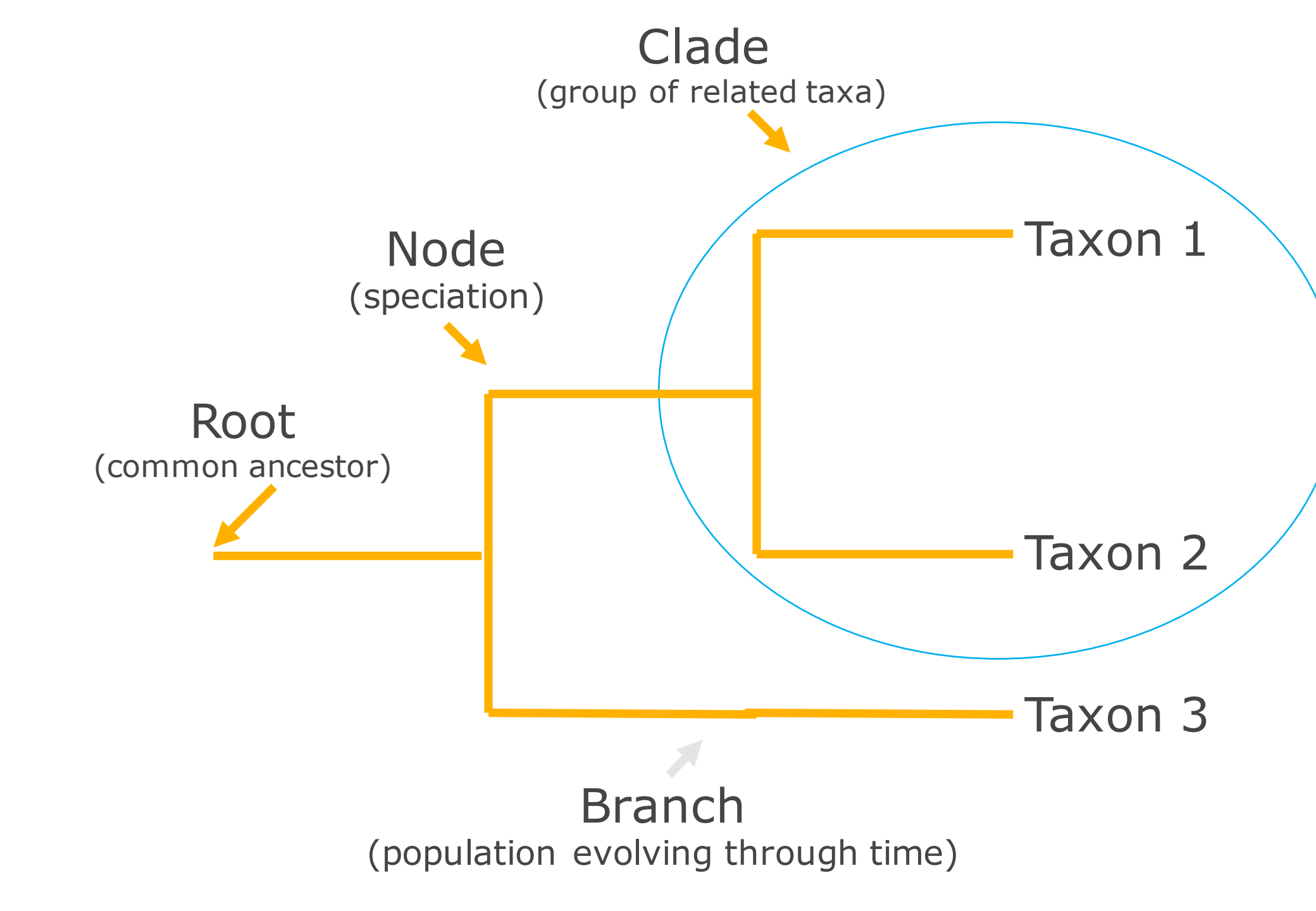
Year


Trees are
boring

Chicken or the Egg?



Shykoff & Widmer (1998)
Trends in Ecology and Evolution, 13, 158.


Clade
(group of related taxa)
Node
(speciation)
Root
(common ancestor)
Taxon 1
Taxon 2
Taxon 3
Branch
(population evolving through time)

Trees rotate around nodes

ORDER is not important. GROUPINGS are.

Chicken or the Egg?

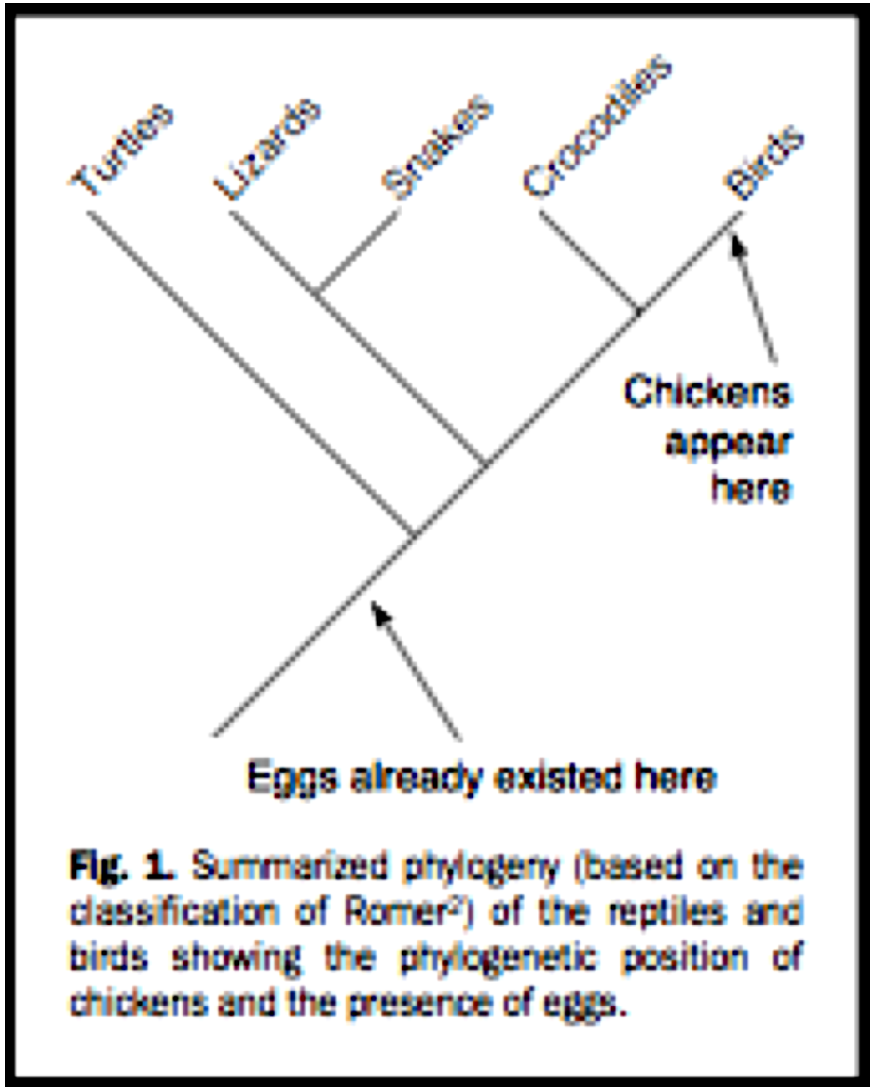
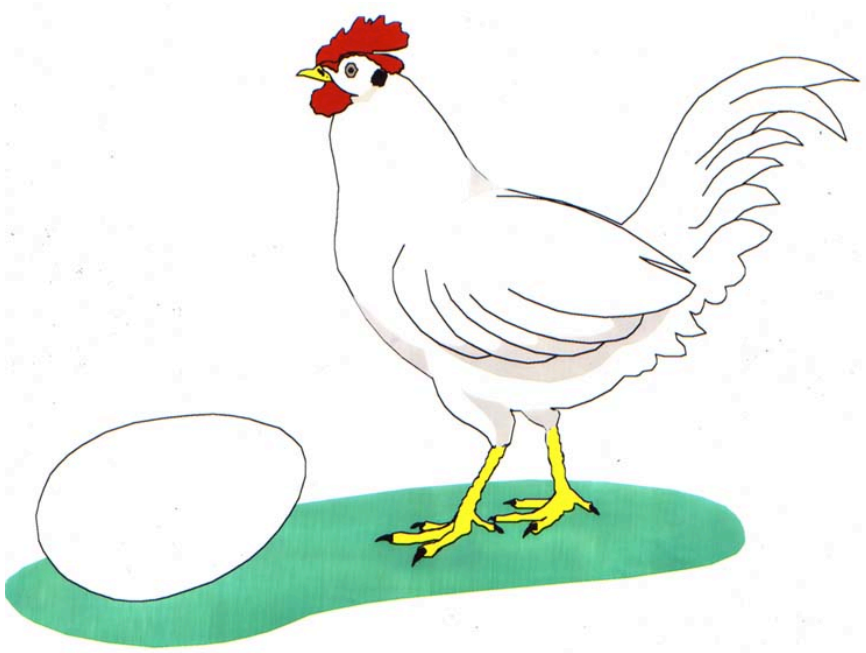


Fig. 1. Summarized phylogeny (based on the classification of Romer[2]) of the reptiles and birds showing the phylogenetic position of chickens and the presence of eggs.

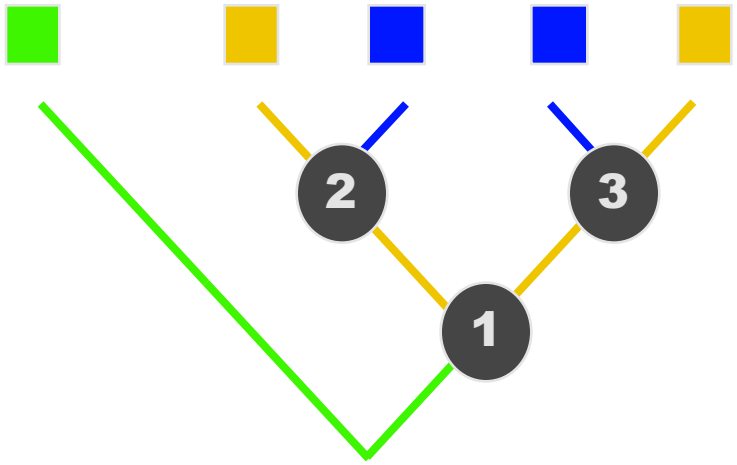


Shykoff & Widmer (1998)
Trends in Ecology and Evolution, 13, 158.

How do we **build** trees?

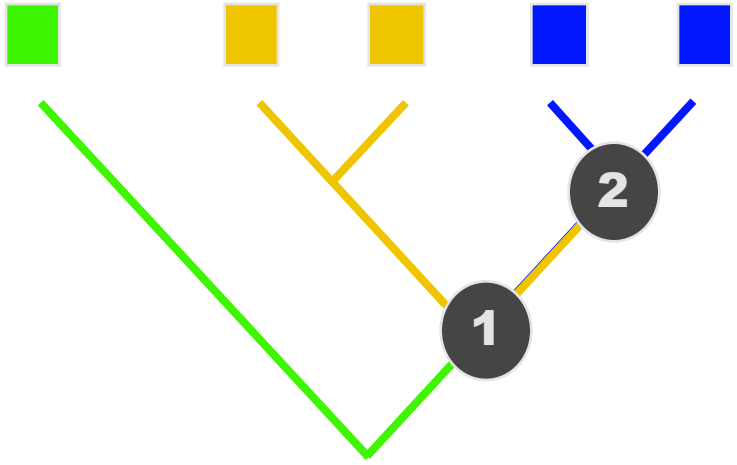
1. Maximum Parsimony.
2. Maximum Likelihood.
3. **Bayesian Phylogenetic Analyses.**

Maximum Parsimony



Aim to find “most parsimonious” tree

Smallest amount of evolution



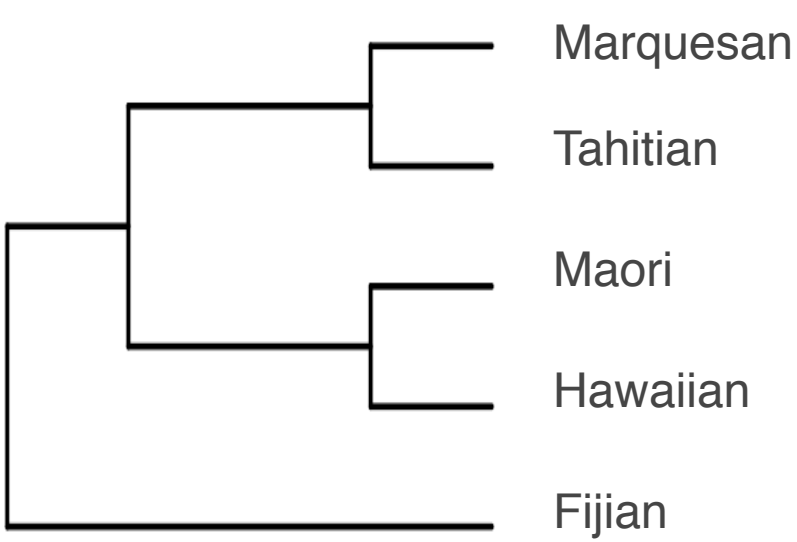
Unlikely that things should arise more than once

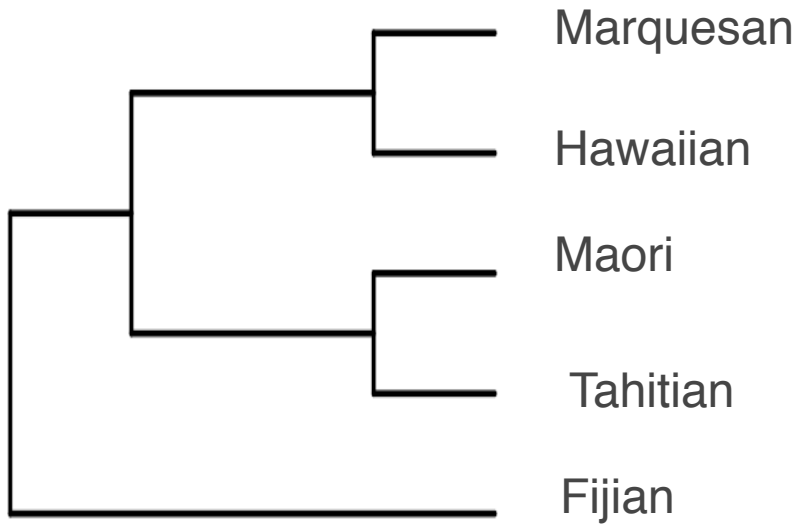
(i.e. cognates should innovate once)

	Taboo	Blood	To Suck
Fijian	tabu	drā	sucu-ma
Tahitian	tapu	toto	ngote
Maori	tapu	toto	ngote
Hawaiian	kapu	koko	omo
Marquesan	tapu	toto	omo

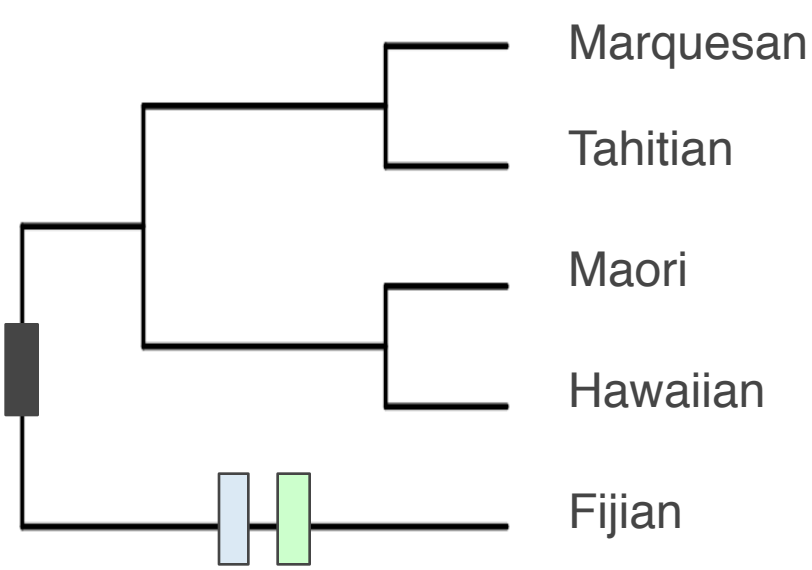
	Taboo	Blood	To Suck
Fijian	tabu	drā	sucu-ma
Tahitian	tapu	toto	ngote
Maori	tapu	toto	ngote
Hawaiian	kapu	koko	omo
Marquesan	tapu	toto	omo

Fijian	1	1	0	1	0	0
Tahitian	1	0	1	0	1	0
Maori	1	0	1	0	1	0
Hawaiian	1	0	1	0	0	1
Marquesan	1	0	1	0	0	1





Fijian	**1**	**1**	**0**	**1**	**0**	**0**
Tahitian	**1**	**0**	**1**	**0**	**1**	**0**
Maori	**1**	**0**	**1**	**0**	**1**	**0**
Hawaiian	**1**	**0**	**1**	**0**	**0**	**1**
Marquesan	**1**	**0**	**1**	**0**	**0**	**1**



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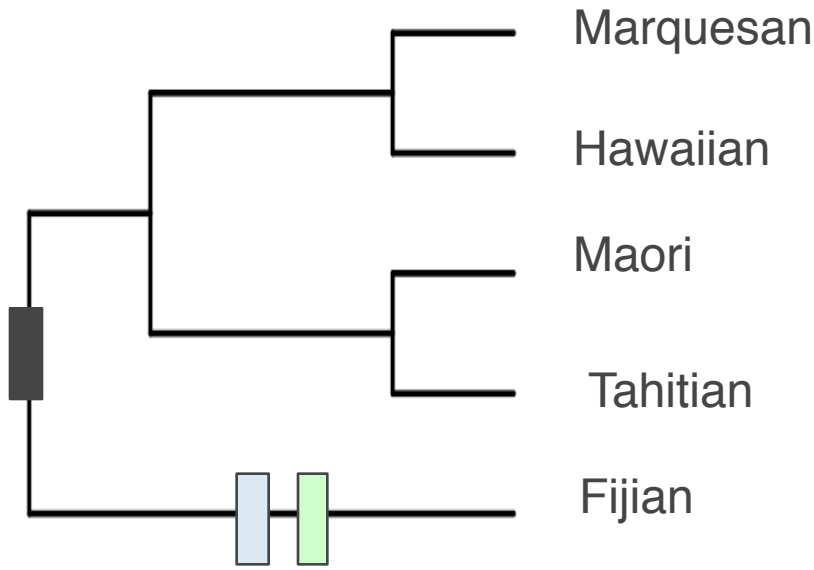
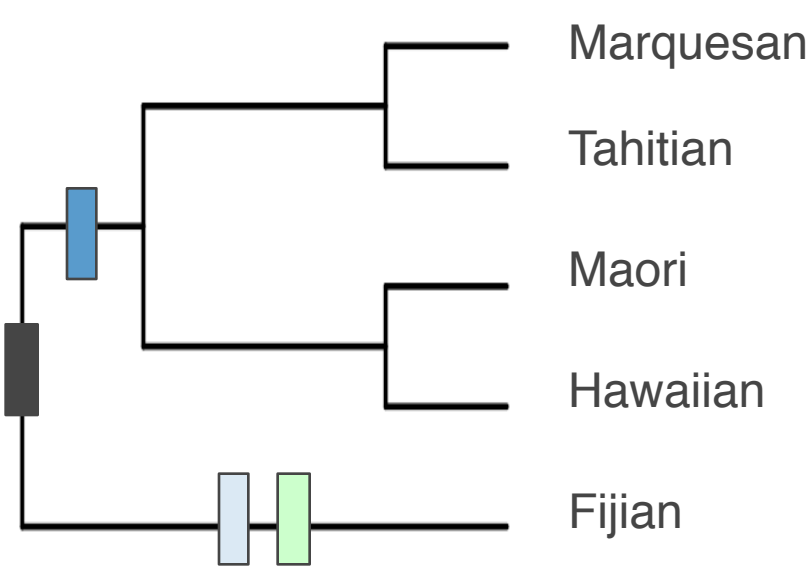


Length=3

Fijian	**1**	**1**	**0**	**1**	**0**	**0**
Tahitian	**1**	**0**	**1**	**0**	**1**	**0**
Maori	**1**	**0**	**1**	**0**	**1**	**0**
Hawaiian	**1**	**0**	**1**	**0**	**0**	**1**
Marquesan	**1**	**0**	**1**	**0**	**0**	**1**



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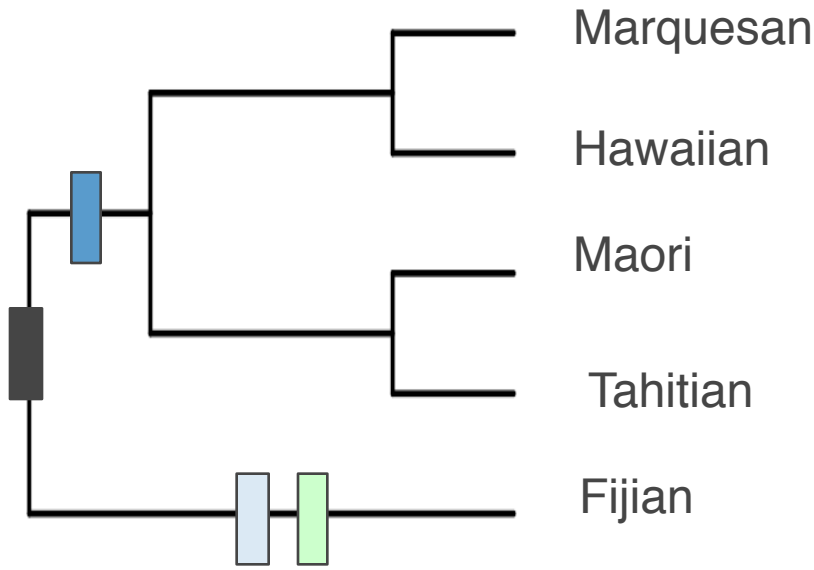
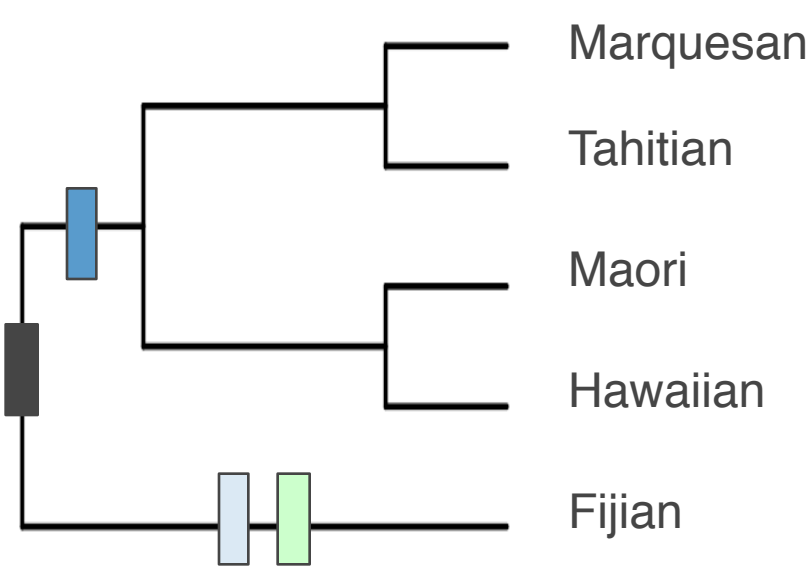
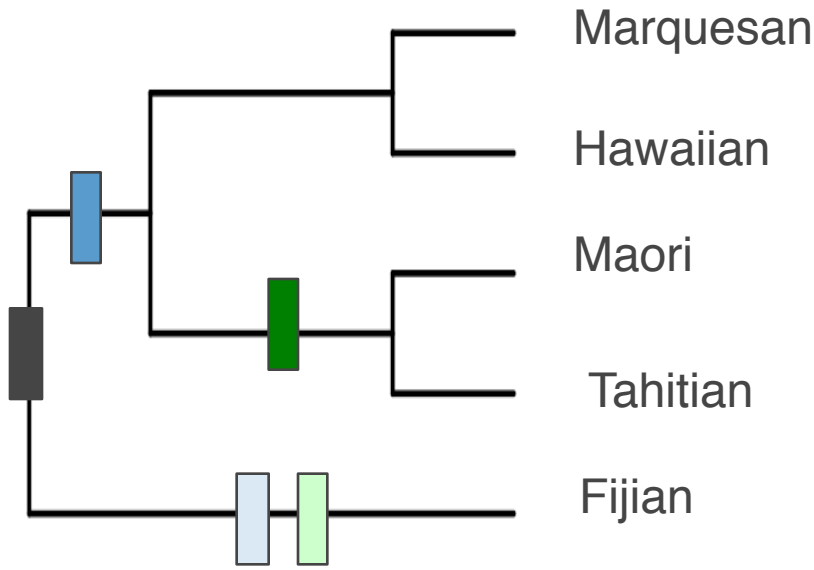


Length=4

Fijian	**1**	**1**	**0**	**1**	**0**	**0**
Tahitian	**1**	**0**	**1**	**0**	**1**	**0**
Maori	**1**	**0**	**1**	**0**	**1**	**0**
Hawaiian	**1**	**0**	**1**	**0**	**0**	**1**
Marquesan	**1**	**0**	**1**	**0**	**0**	**1**

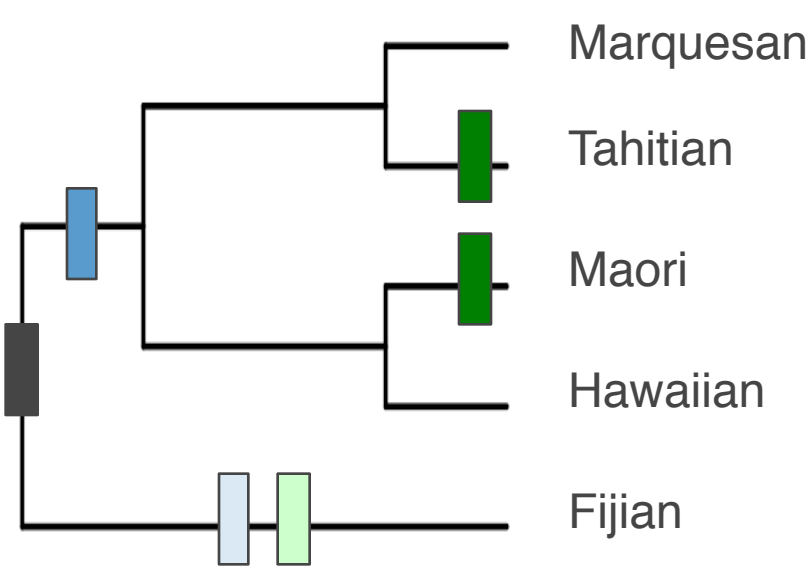




Length=4

Length=5

Fijian	**1**	**1**	**0**	**1**	**0**	**0**
Tahitian	**1**	**0**	**1**	**0**	**1**	**0**
Maori	**1**	**0**	**1**	**0**	**1**	**0**
Hawaiian	**1**	**0**	**1**	**0**	**0**	**1**
Marquesan	**1**	**0**	**1**	**0**	**0**	**1**



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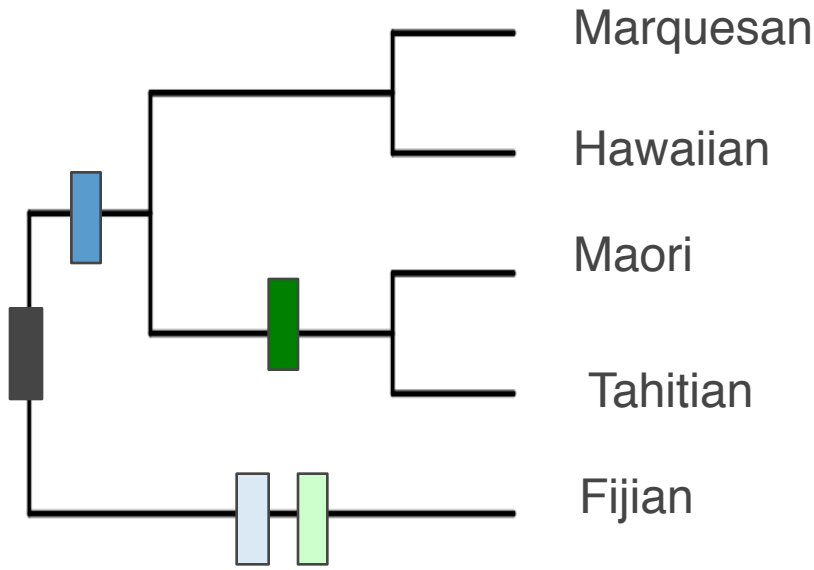
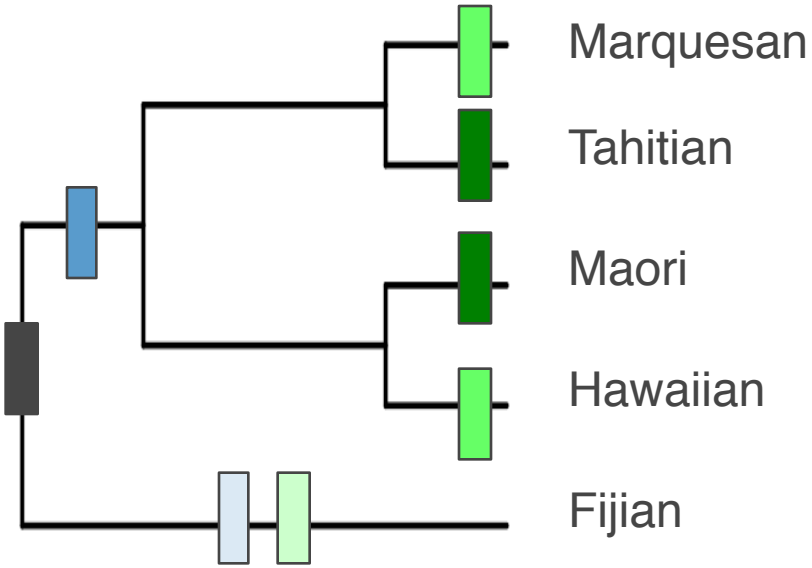
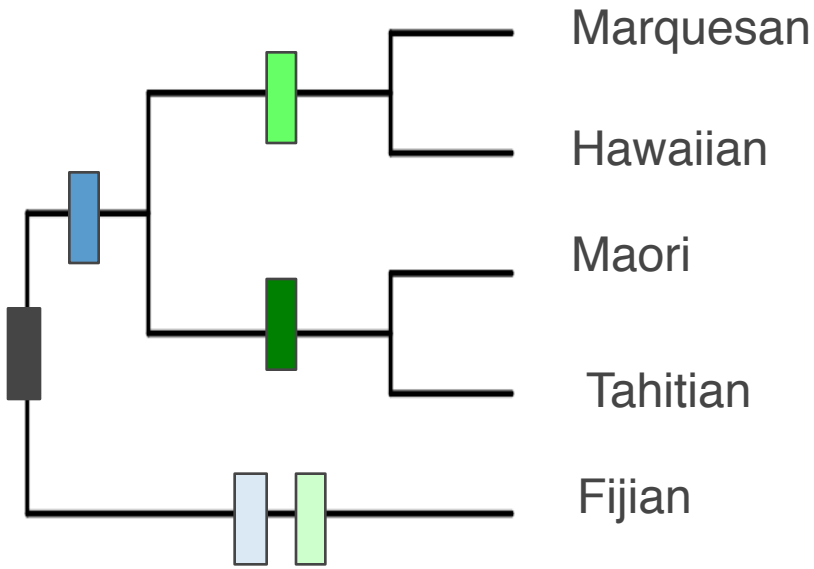


Length=5

Fijian	**1**	**1**	**0**	**1**	**0**	**0**
Tahitian	**1**	**0**	**1**	**0**	**1**	**0**
Maori	**1**	**0**	**1**	**0**	**1**	**0**
Hawaiian	**1**	**0**	**1**	**0**	**0**	**1**
Marquesan	**1**	**0**	**1**	**0**	**0**	**1**

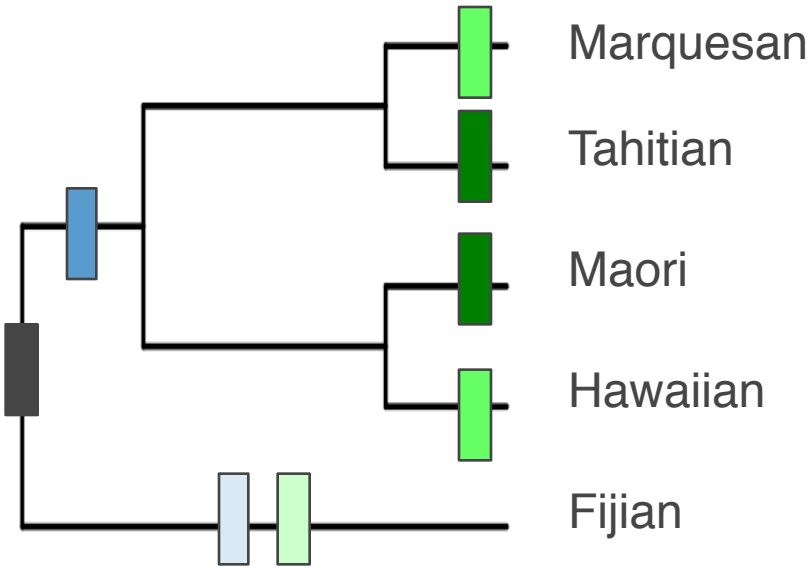


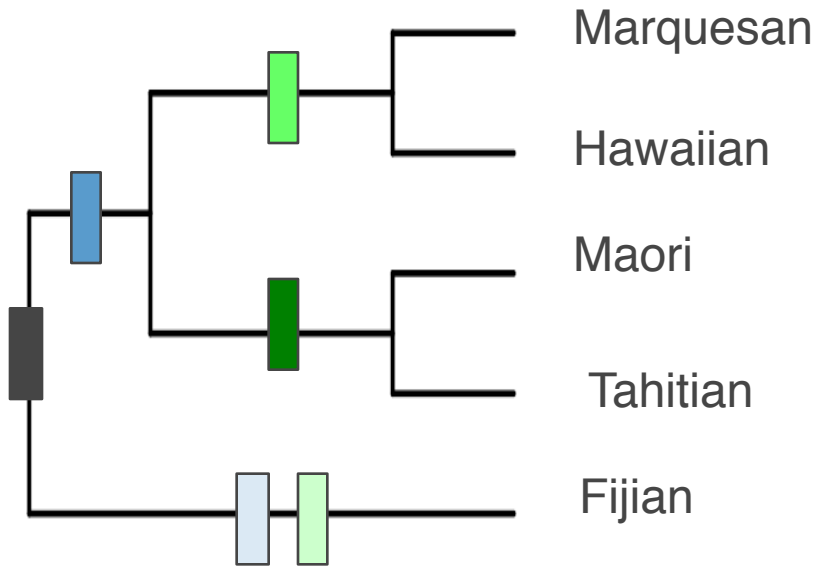


Length=8

Length=6

Fijian	**1**	**1**	**0**	**1**	**0**	**0**
Tahitian	**1**	**0**	**1**	**0**	**1**	**0**
Maori	**1**	**0**	**1**	**0**	**1**	**0**
Hawaiian	**1**	**0**	**1**	**0**	**0**	**1**
Marquesan	**1**	**0**	**1**	**0**	**0**	**1**



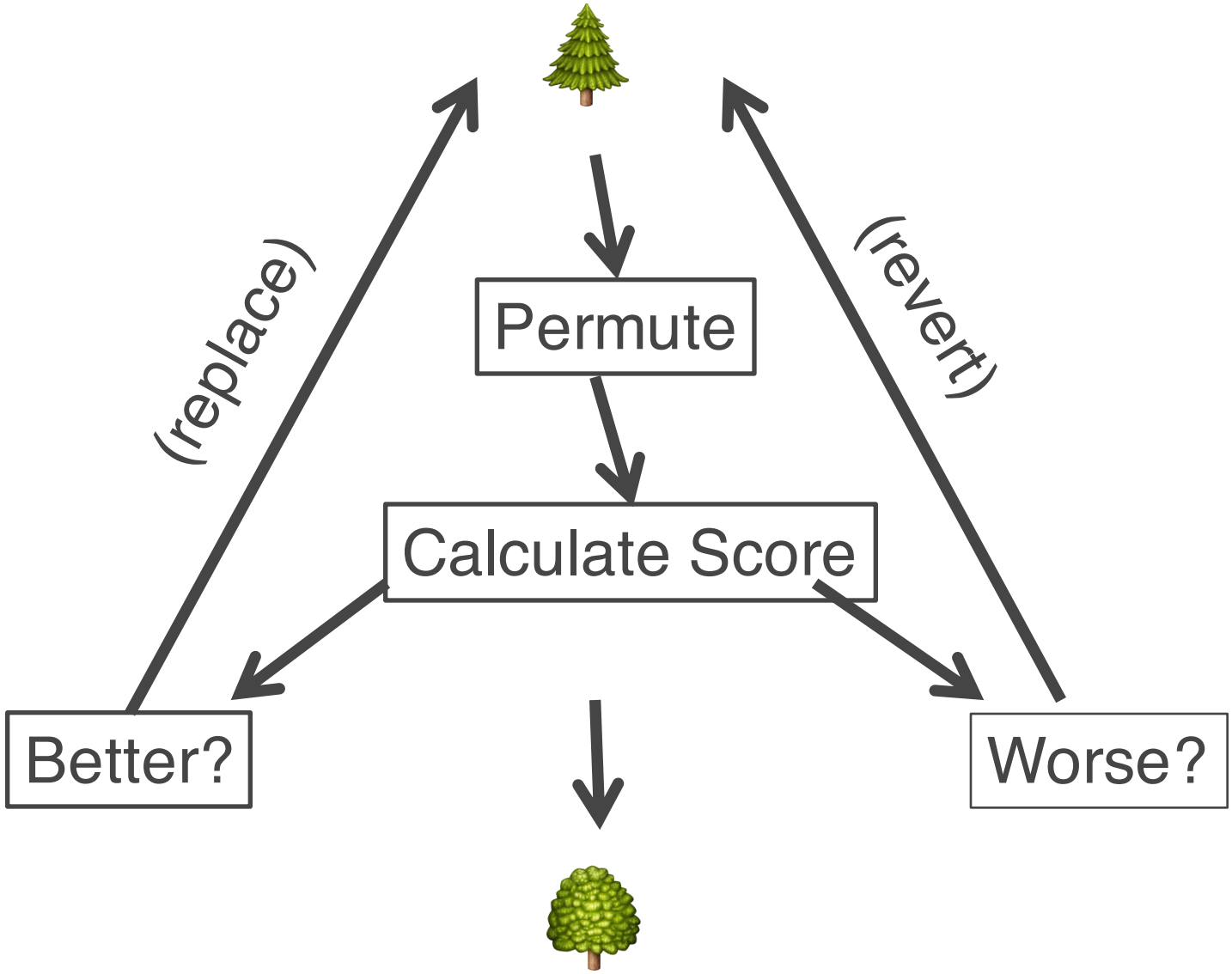


Length=8

Length=6

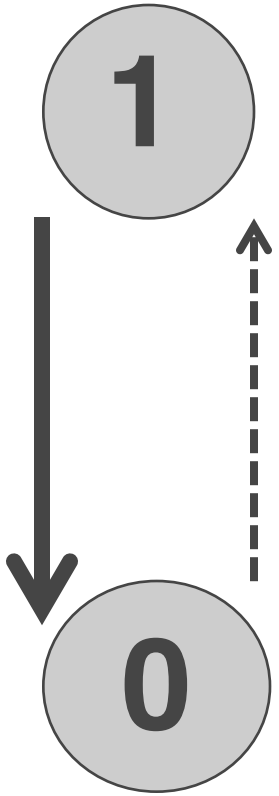
Fijian	**1**	**1**	**0**	**1**	**0**	**0**
Tahitian	**1**	**0**	**1**	**0**	**1**	**0**
Maori	**1**	**0**	**1**	**0**	**1**	**0**
Hawaiian	**1**	**0**	**1**	**0**	**0**	**1**
Marquesan	**1**	**0**	**1**	**0**	**0**	**1**

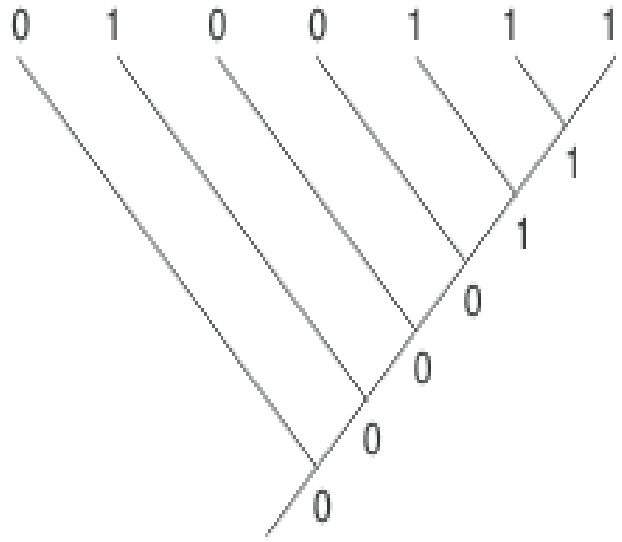
Algorithm



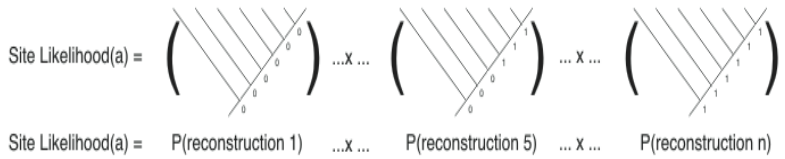
Maximum Likelihood

- Builds on Max. Parsimony
- Stochastic **model** of change.
- **Likelihood** = fit of data to tree under a model.
  - Very small number = log(Lh)
  - Closer to zero = better fit.





$L(a) = P(0\rightarrow 0|b1) \times P(0\rightarrow 0|b2) \times P(1\rightarrow 1|b3) \times P(1\rightarrow 0|b4) \times P(0\rightarrow 0|b5) \times P(0\rightarrow 0|b6) \times P(0\rightarrow 0|b7) \times P(0\rightarrow 1|b8) \times P(1\rightarrow 1|b9) \times P(1\rightarrow 1|b10) \times P(1\rightarrow 1|b11) \times P(1\rightarrow 1|b12)$



Lh =
P of being in state 0, and staying state 0 on branch 1.

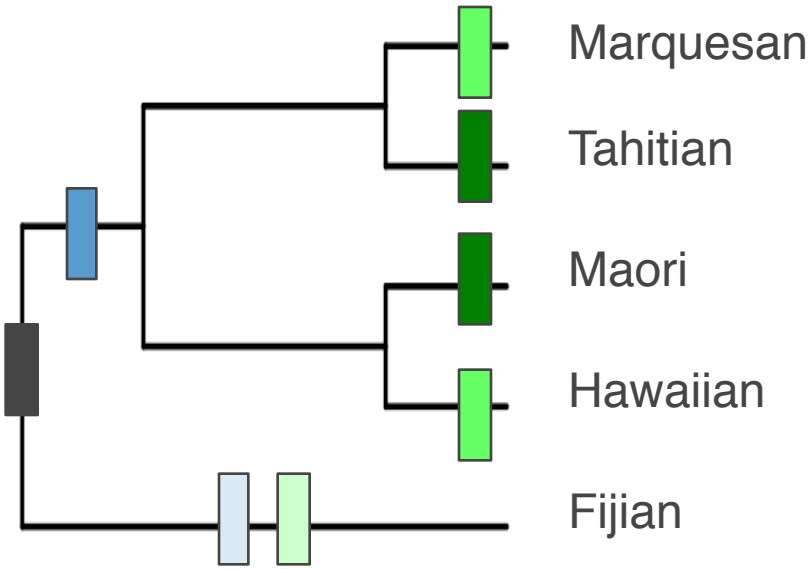
x

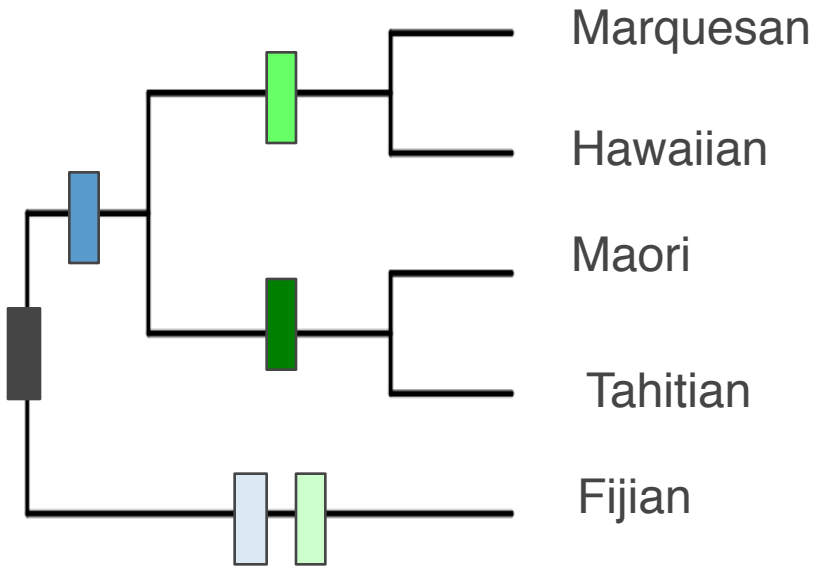
P of being in state 0, and staying state 0 on branch 2.

x

P of being in state 0, and staying state 0 on branch 2.

…. etc



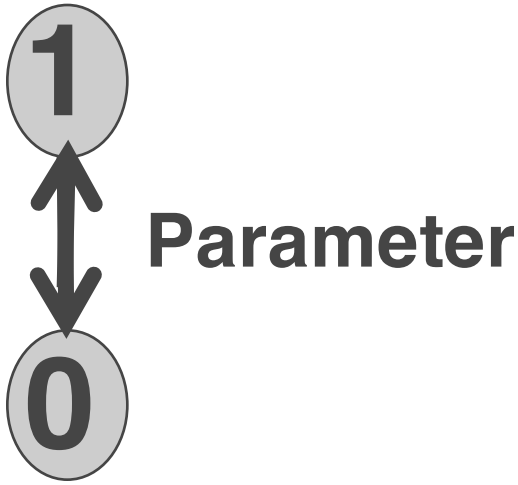


Ln(L)= -14.804

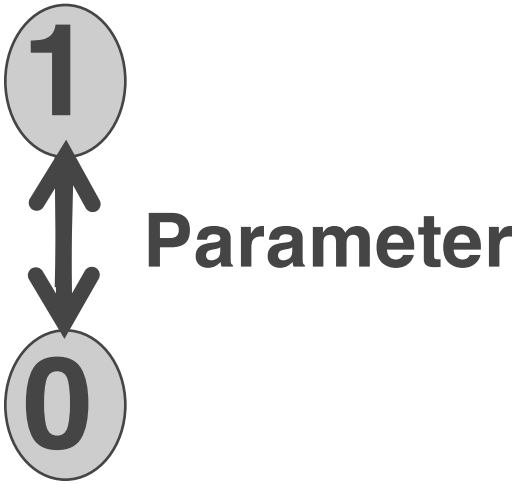
Ln(L)= -12.007

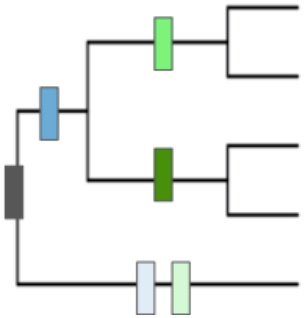
Fijian	**1**	**1**	**0**	**1**	**0**	**0**
Tahitian	**1**	**0**	**1**	**0**	**1**	**0**
Maori	**1**	**0**	**1**	**0**	**1**	**0**
Hawaiian	**1**	**0**	**1**	**0**	**0**	**1**
Marquesan	**1**	**0**	**1**	**0**	**0**	**1**

Models



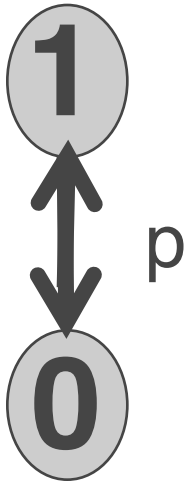
Models

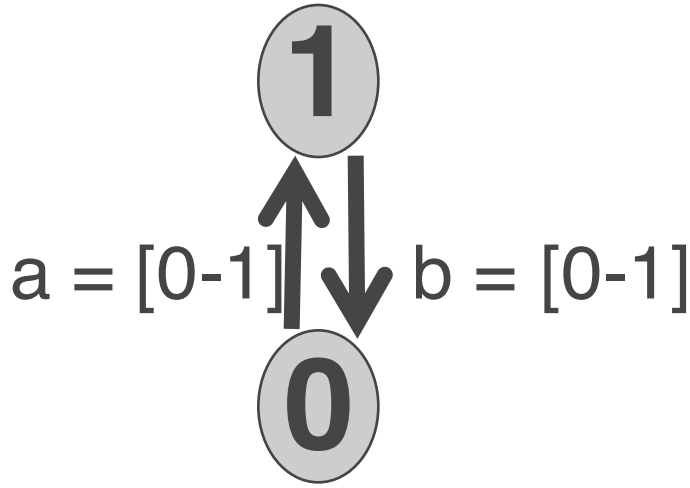


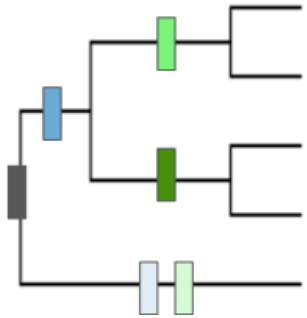


Ln(L)= -12.007

Models

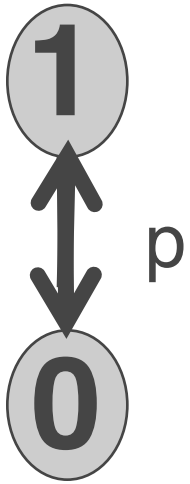


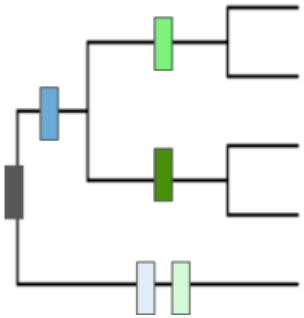




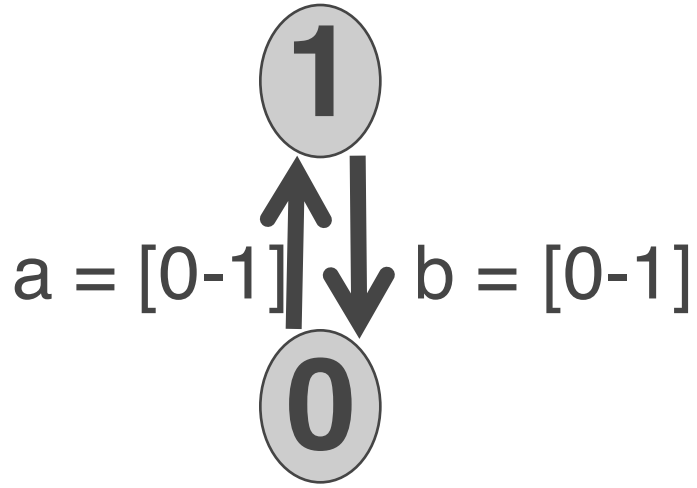
Ln(L)= -12.007

Models

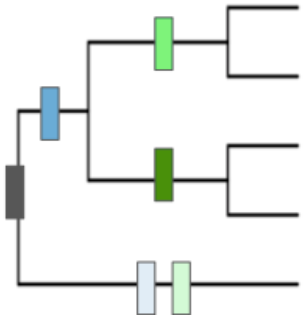




Ln(L)= -12.007



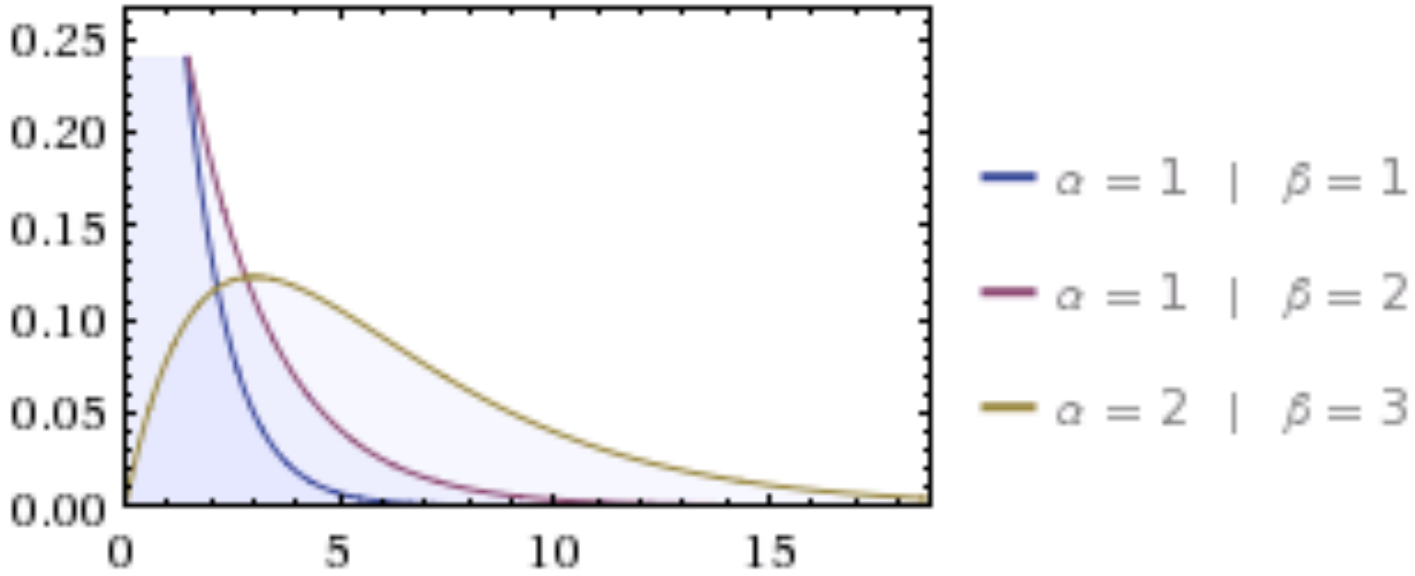
a = 0.92, b = 0.08

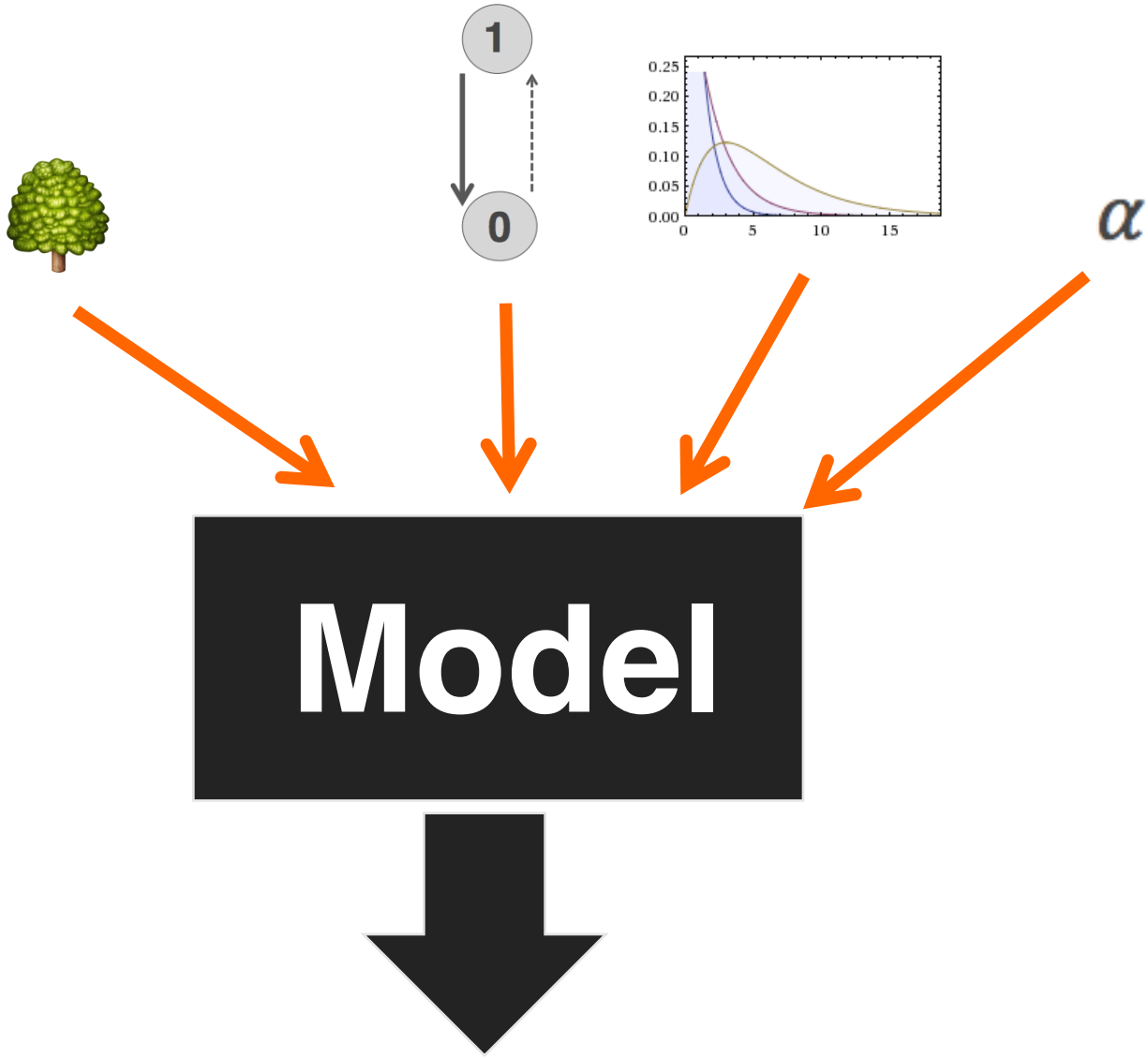


Ln(L)= -9.072

Rate Variation - Gamma

- Gamma Distribution
- One parameter, α, controls the shape
- Estimate the best value for α using Lh.




1
0
0.25
0.20
0.15
0.10
0.05
0.00
0
5
10
15
α
Model
Likelihood score

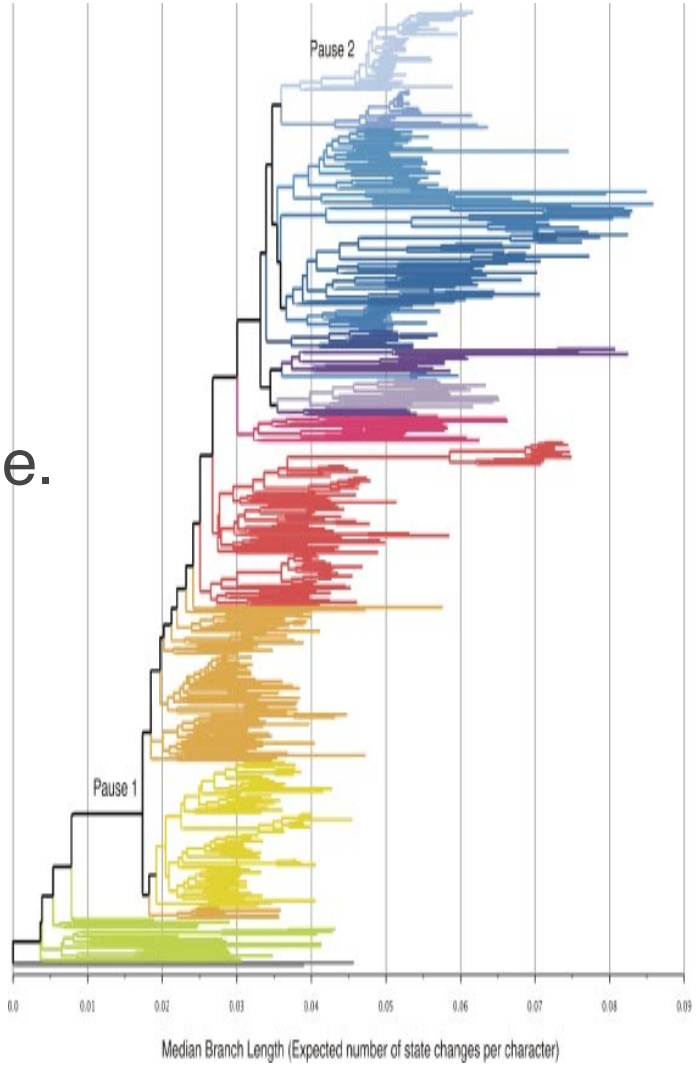
Linguists don’t do dates.

Phylogenetic Dating

ML: Estimate how long branches are.

(number of changes per cognate set)

Awesome: Not a global retention rate (i.e. glottochronology) but a **per-language** estimate of the amount of change.



Convert Rates to Dates

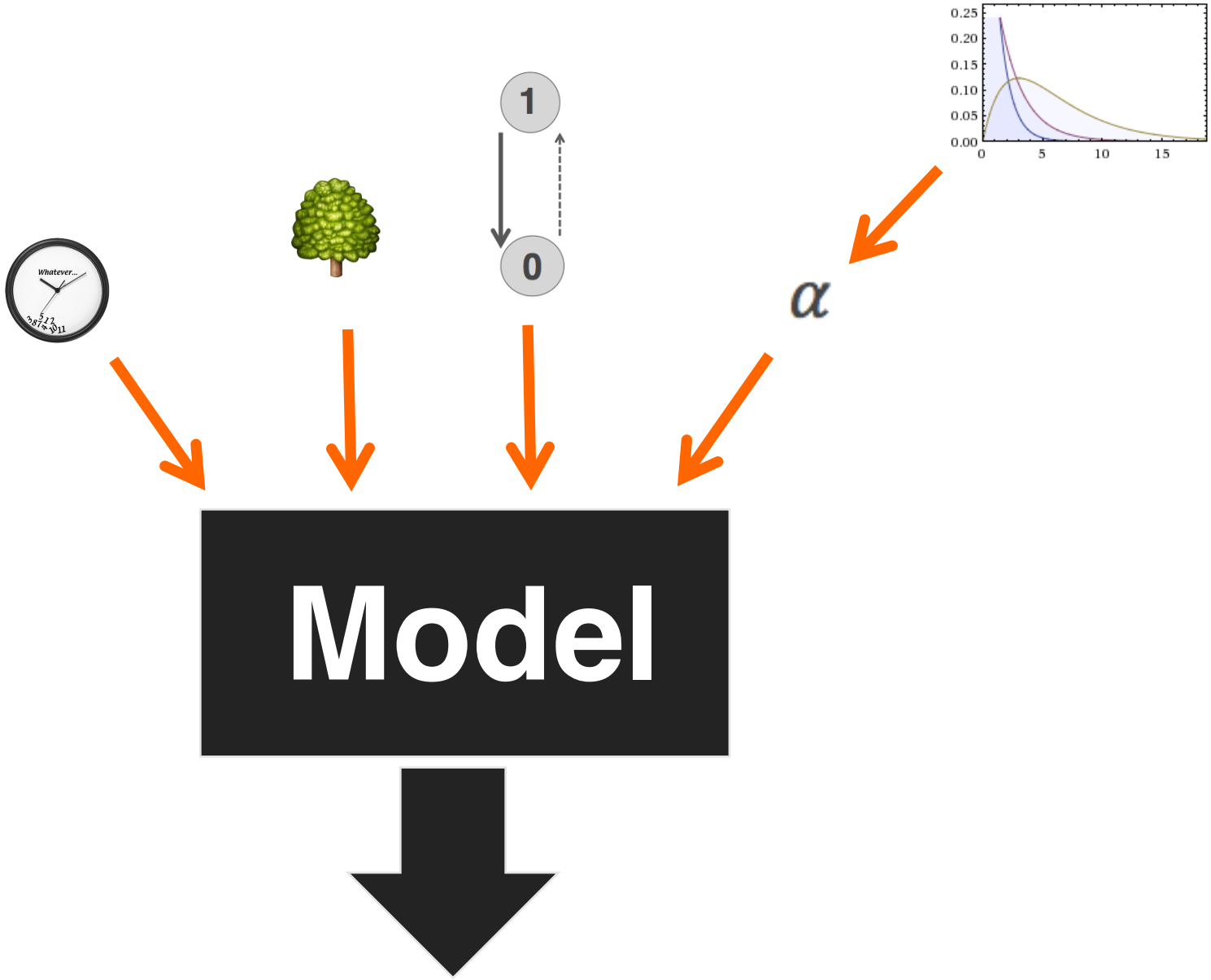
- Use (pre)historical information to calibrate nodes
- e.g. Archaeology suggests initial settlement was..
- e.g. Historical evidence says that X and Y were separate at time Z
- Smooth rates over these calibrations

Strict Clock

- One rate for all languages.
- No variation
- = glottochronology

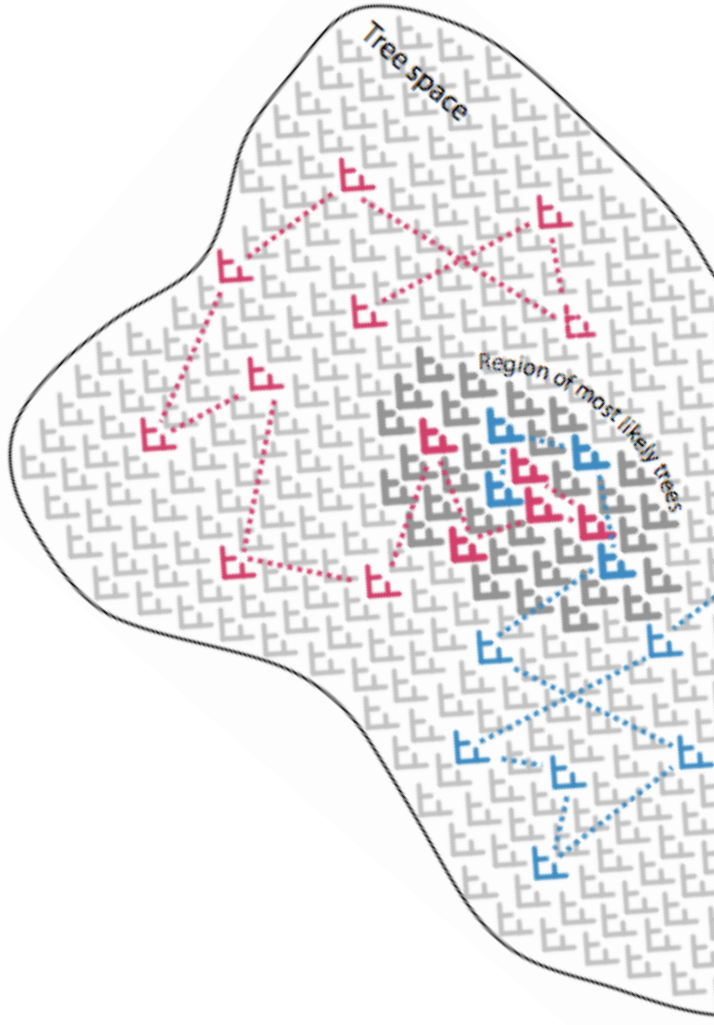
Relaxed Clock

- Allows rate to vary across branches.
- Rates are drawn from a parametric distribution with parameters estimated from the data


0.25
0.20
0.15
0.10
0.05
0.00
0
5
10
15
1
0
α
Whatever...
Model
Likelihood score

Bayesian Phylogenetics

1. Data & Model & Tree
2. Calculate the **Likelihood** of that tree
3. Modify The Tree or a Model Parameter
4. Repeat (MCMC “Walk” through treespace)



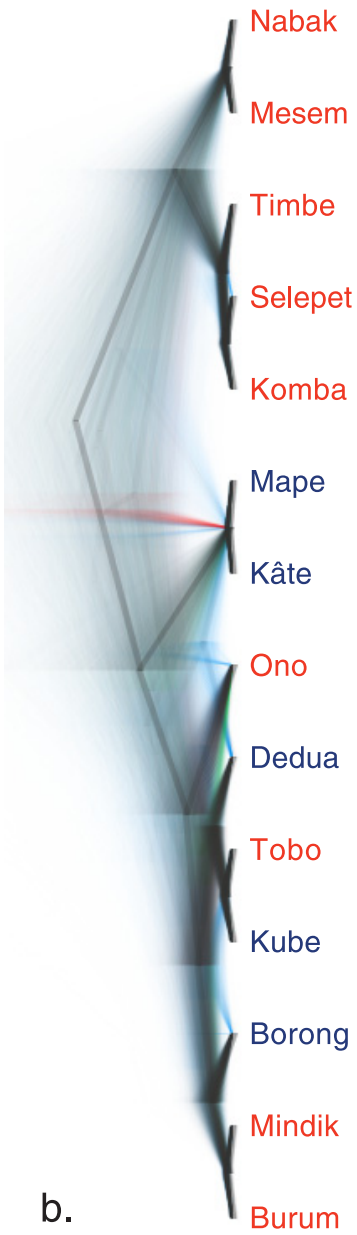
-> samples best fitting trees from all possible trees.

->”Posterior Probability Distribution”

Posterior Probability Distribution

- Not just 1 tree but sample
- Uncertainty, conflicting signal come out.

- Simulations: robust to reasonable levels of borrowing (15% / 1000 y)



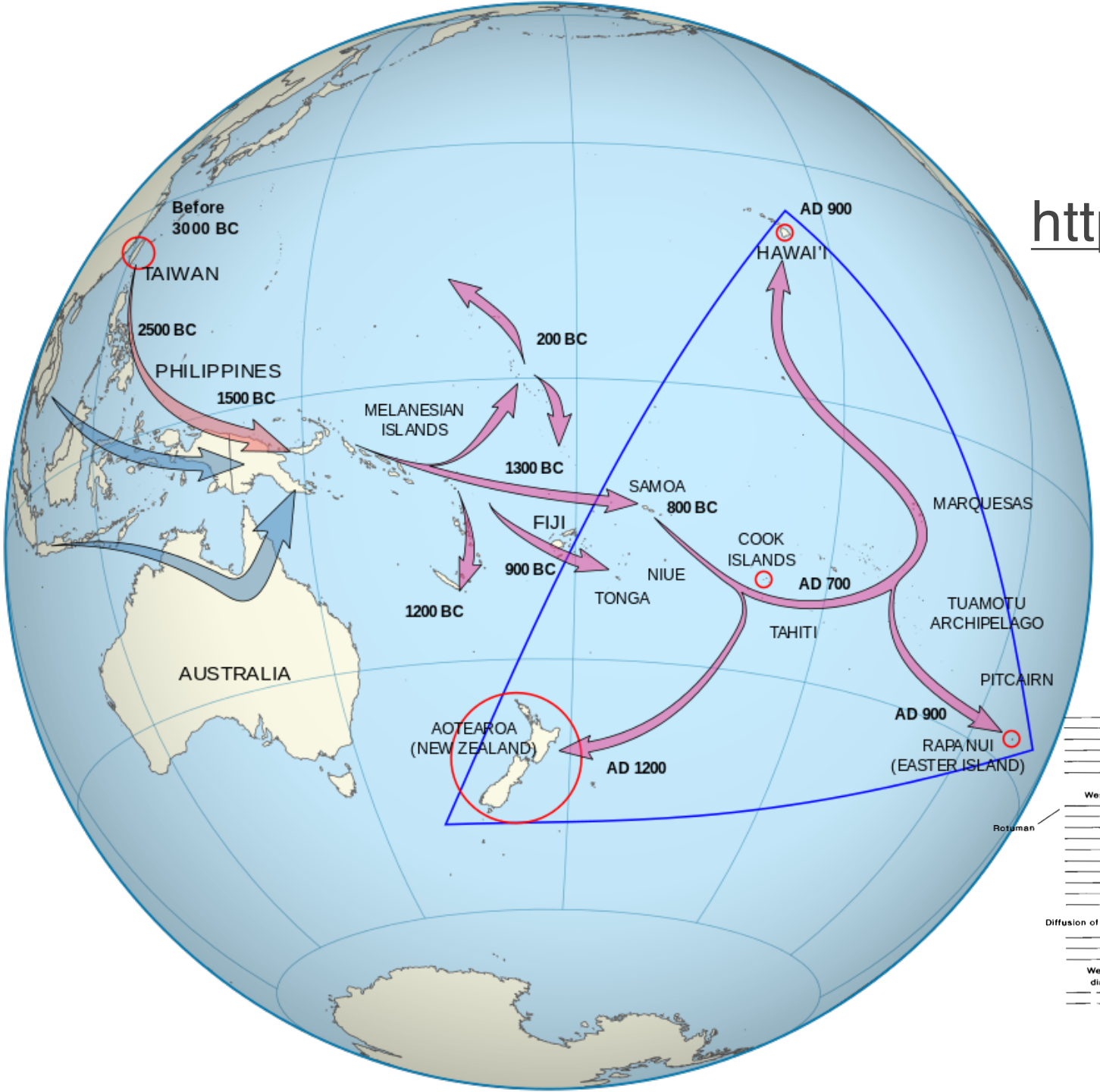
b.

Summary

- Many conceptual parallels across disciplines.
- Grew out of many of the same concerns found in Linguistics/Anthropology (innovations vs retentions, rate variation, conflicting signal, etc).
- Currently are incredibly powerful tools for answering certain types of questions (not all!).
- Range of methods available. Bayesian P.M. 👍
- Fun.

Note: H.O.P by Fiona on Bayesian methods

Questions?



DATA
http://bit.ly/252YNMj