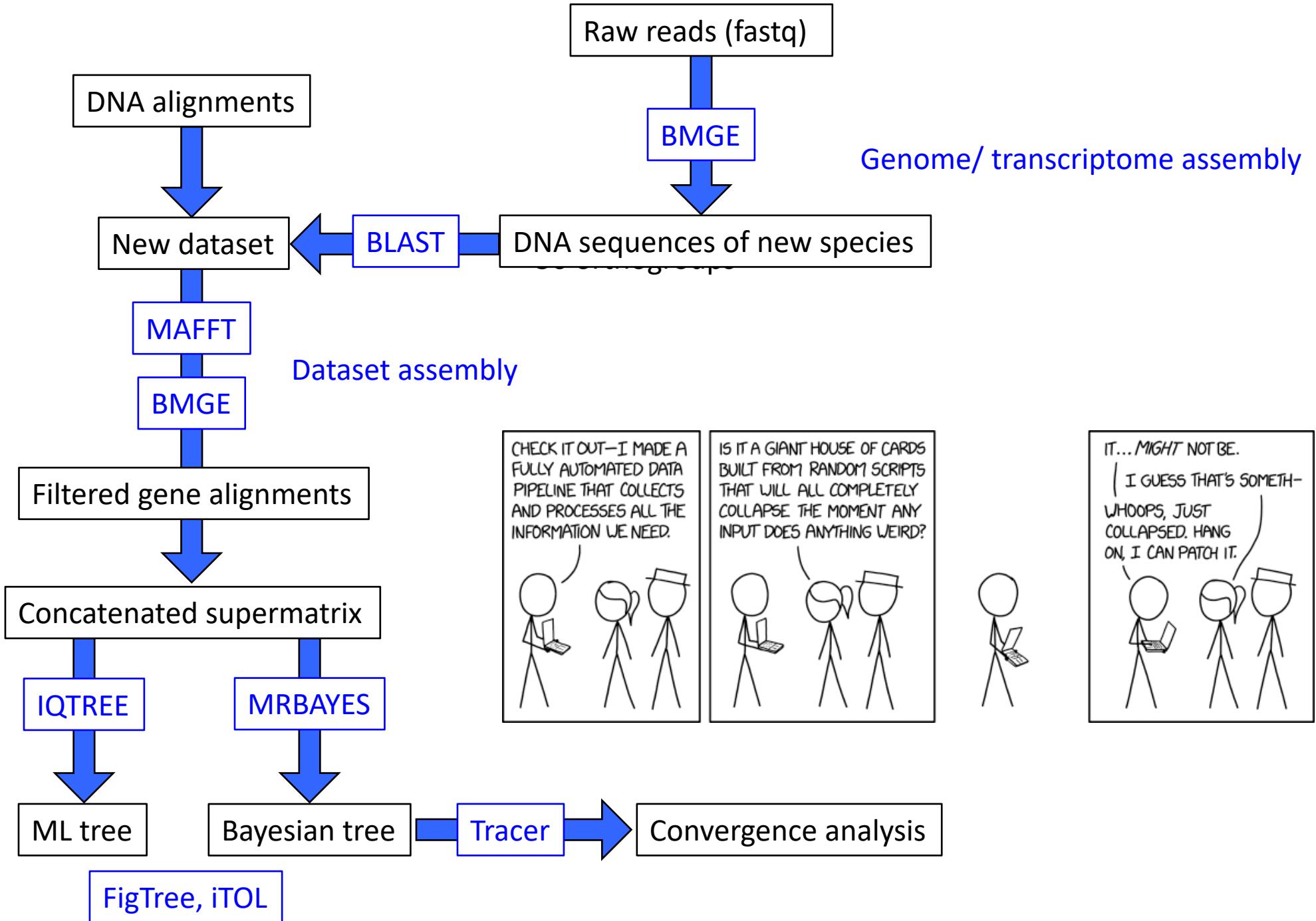


# Pipeline

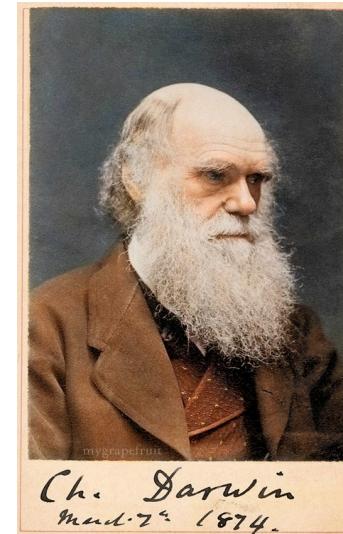
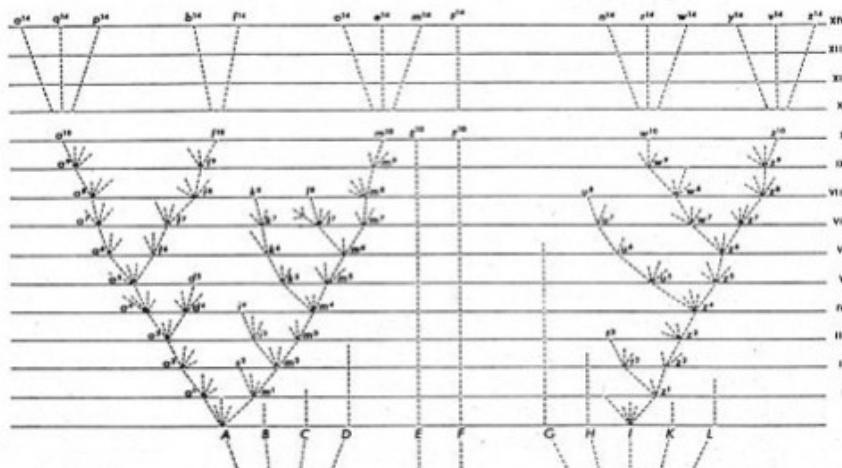
Phylogenetic analysis



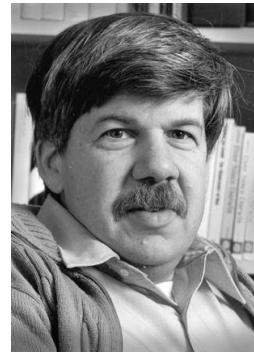
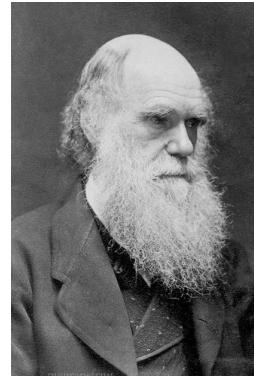


*"I am fully convinced that species are not immutable; but that those belonging to what are called the same genera are lineal descendants of some other and generally extinct species, in the same manner as the acknowledged varieties of any one species are the descendants of that species. Furthermore, I am convinced that natural selection has been the most important, but not the exclusive, means of modification."*

— Charles Darwin,  
On the Origin of Species 1859



- Ancestor-descent relationships imply we can use trees to express evolutionary relationships
- Evolution is an historical science (contingency)
- Evolution is a population process



*"Nothing in biology makes sense, except in the light of evolution.  
Without that light it becomes a pile of sundry facts - some of them  
interesting or curious but making no meaningful picture as a whole"*

– Theodosius Dobzhansky



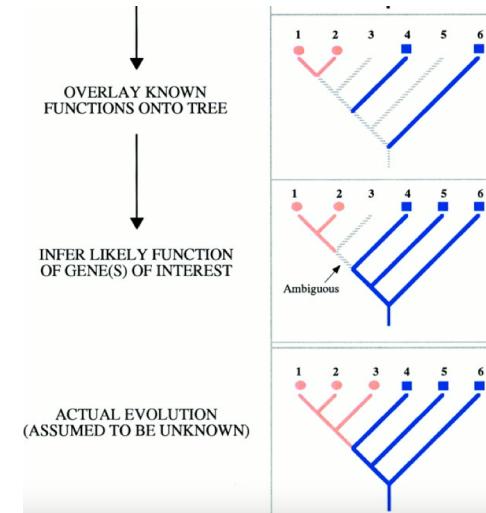
# Definitions

## Phylogenomics: Improving Functional Predictions for Uncharacterized Genes by Evolutionary Analysis

Jonathan A. Eisen<sup>1</sup>

Department of Biological Sciences, Stanford University, Stanford, California 94305-5020 USA

Eisen 1998 Genome Res

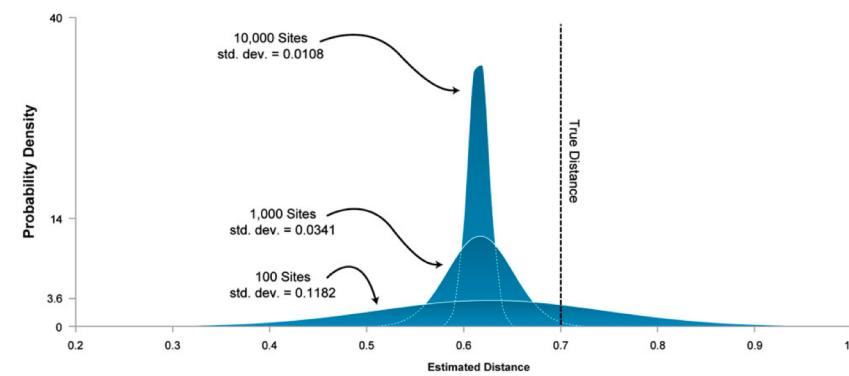


## Phylogenomics

From Wikipedia, the free encyclopedia

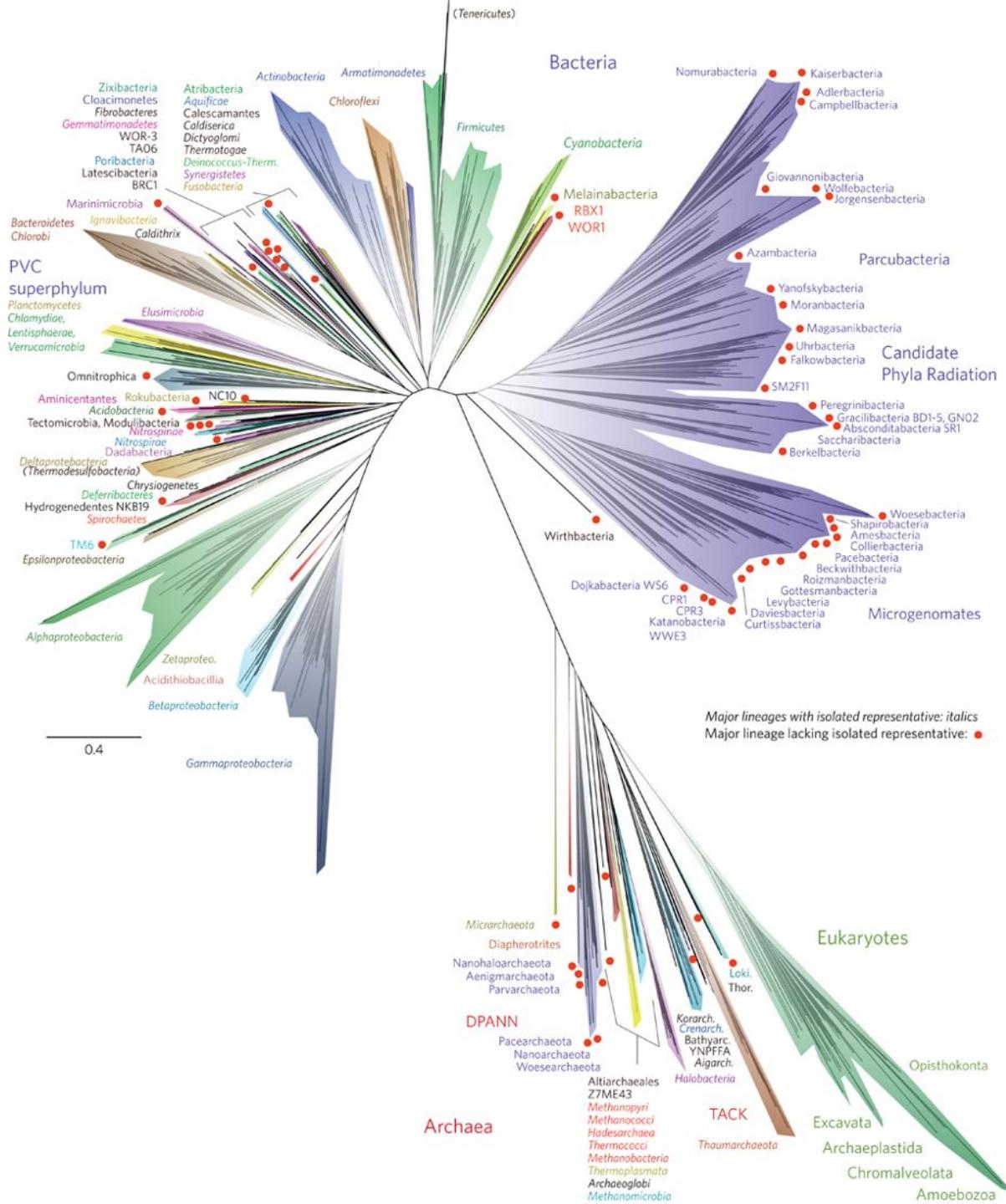
**Phylogenomics** is the intersection of the fields of [evolution](#) and [genomics](#).<sup>[1]</sup>

The term has been used in multiple ways to refer to analysis that involves [genome](#) data and evolutionary reconstructions. It is a group of techniques within the larger fields of [phylogenetics](#) and genomics. Phylogenomics draws information by comparing entire genomes, or at least large portions of genomes.<sup>[2]</sup> Phylogenetics compares and analyzes the sequences of single genes, or a small number of genes, as well as many other types of data.

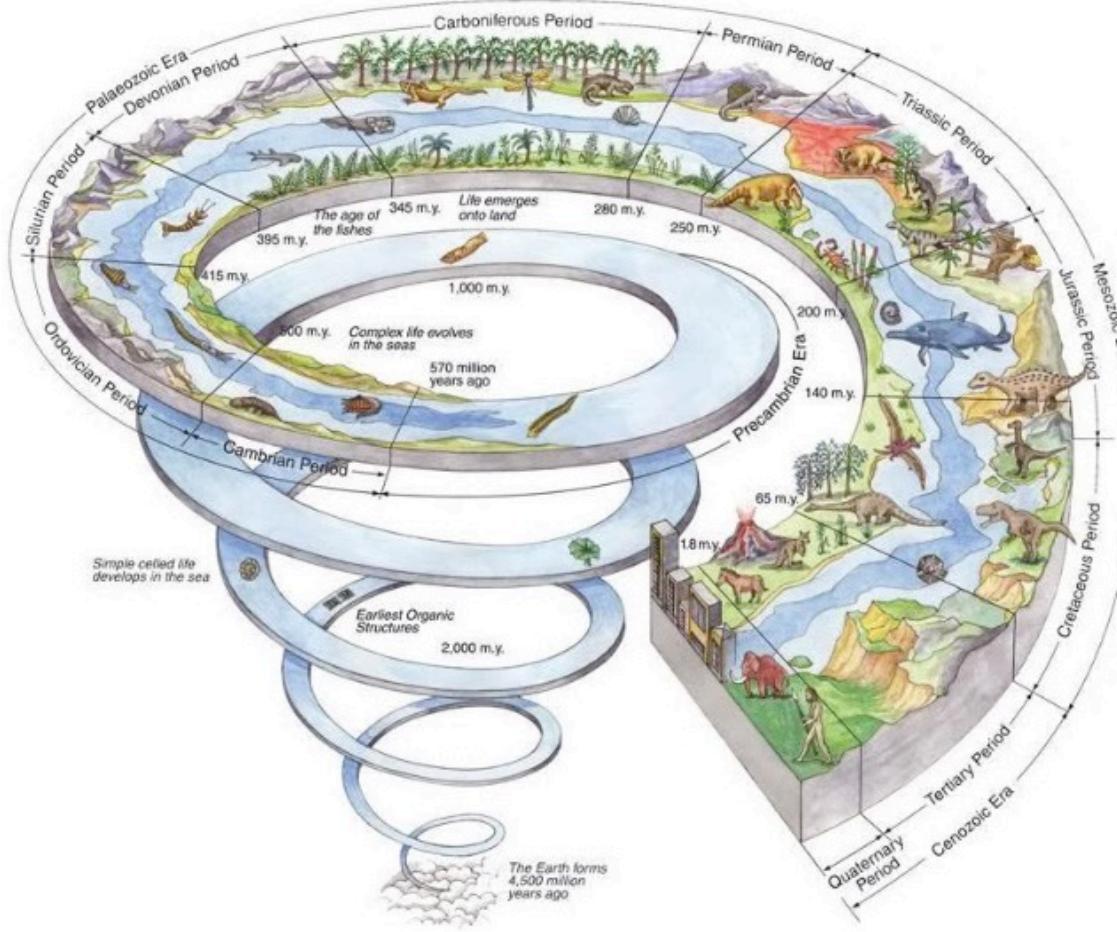


Kumar et al. 2012 Mol Biol Evol

# Tree of Life



# Phylogenomics: “molecular archaeology”



YOUR GENETIC TEST RESULTS  
ARE BACK. APPARENTLY YOU'RE  
PART OF AN UNBROKEN LINEAGE  
STRETCHING BACK BILLIONS OF  
YEARS TO THE EARLY EARTH!



# Understand evolution of traits: morphology, behaviour, etc.

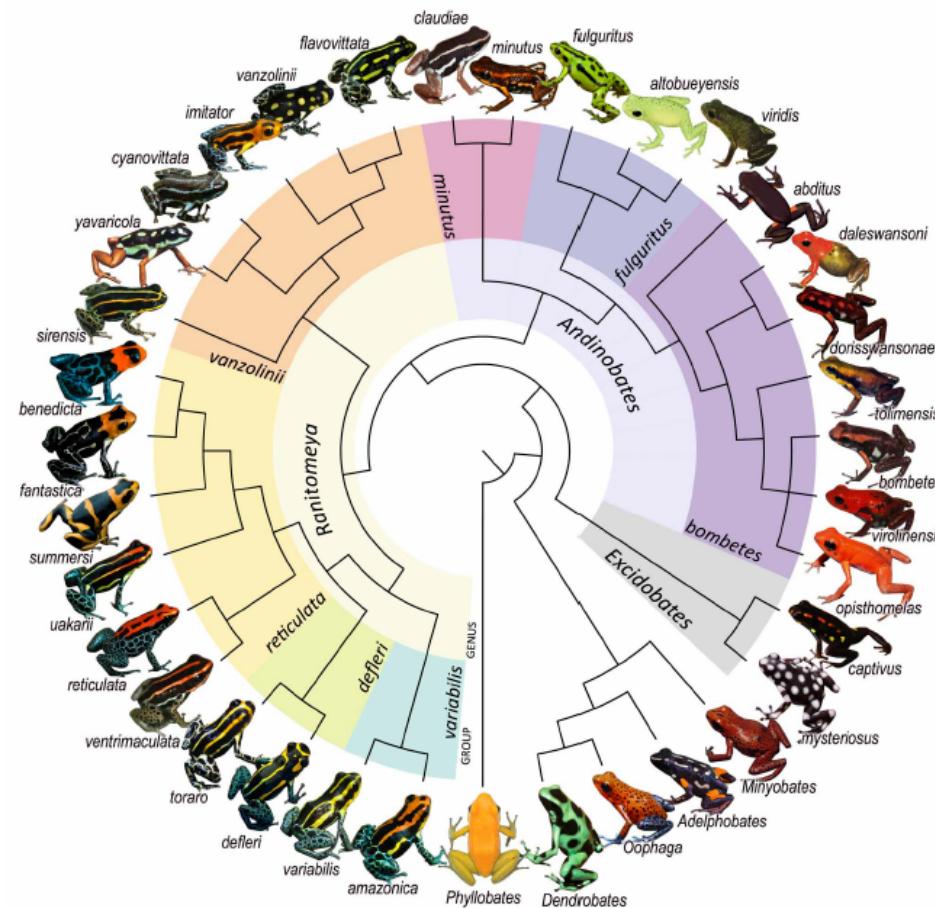
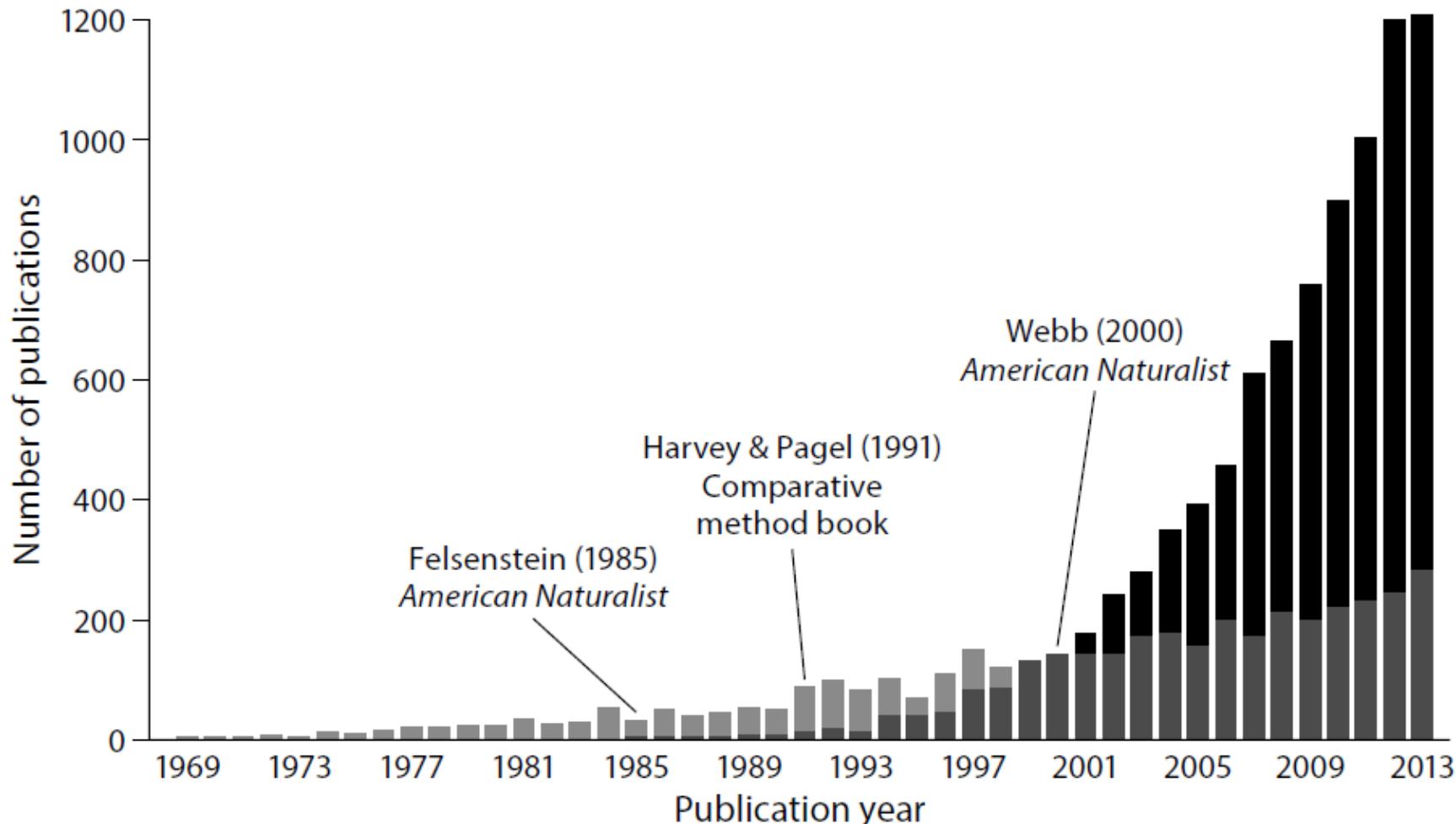
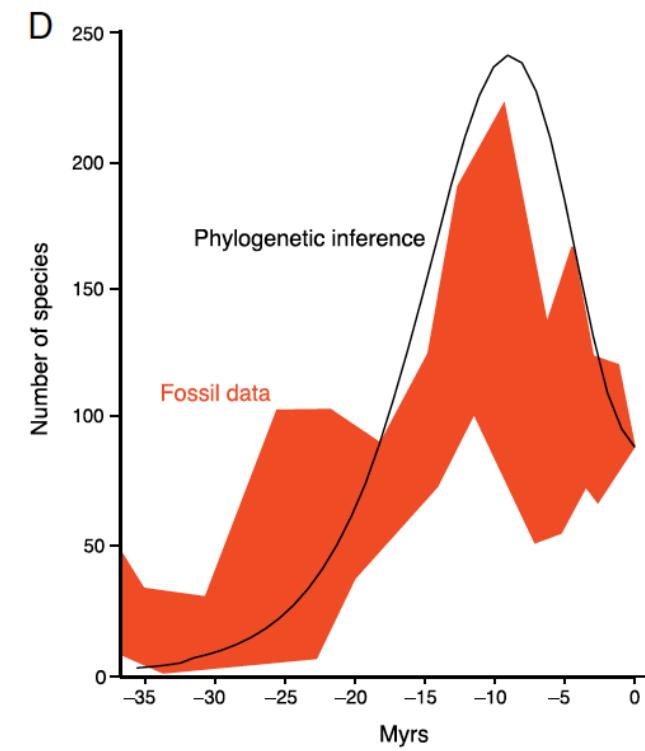
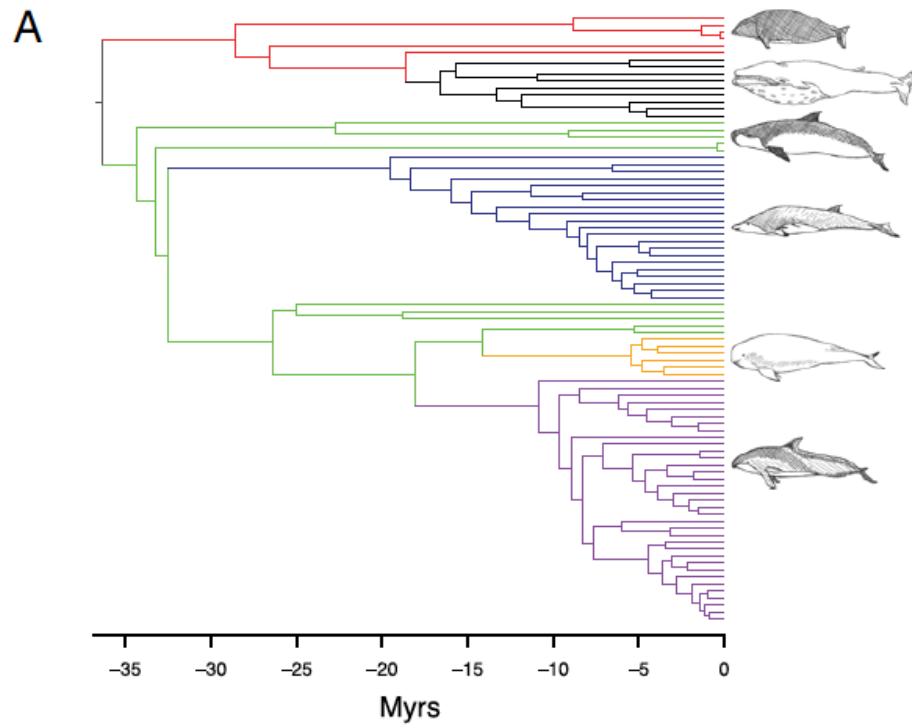


Figure 1. Mimetic forms of *Ranitomeya imitor* and its Müllerian co-mimics.  
Top row, the mimic *Ranitomeya imitor*: left, "Varadero" blotched morph; right, striped morph. Bottom row, the models: left, the aptly named *R. fantastica*; right, *R. variabilis*. Photos courtesy of Evan Twomey.

# Testing hypothesis in Ecology with phylogenies

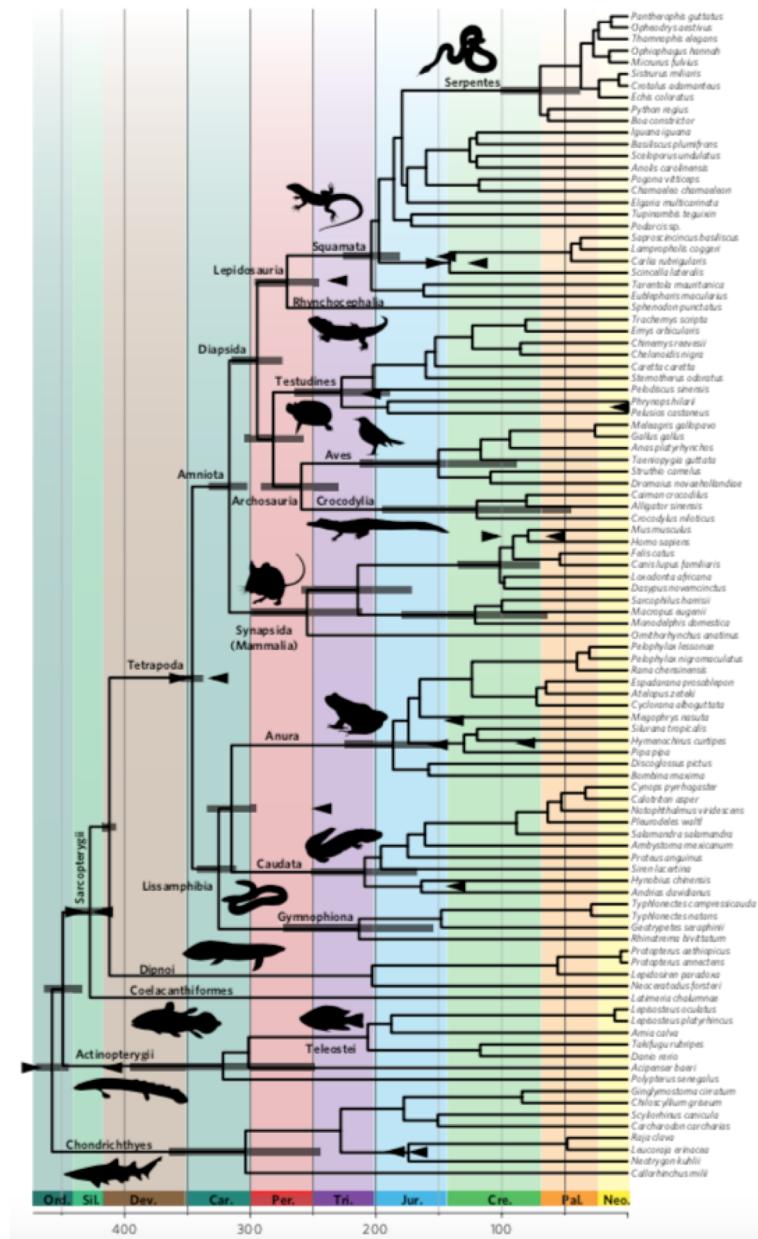


# Testing hypothesis: Macroevolution

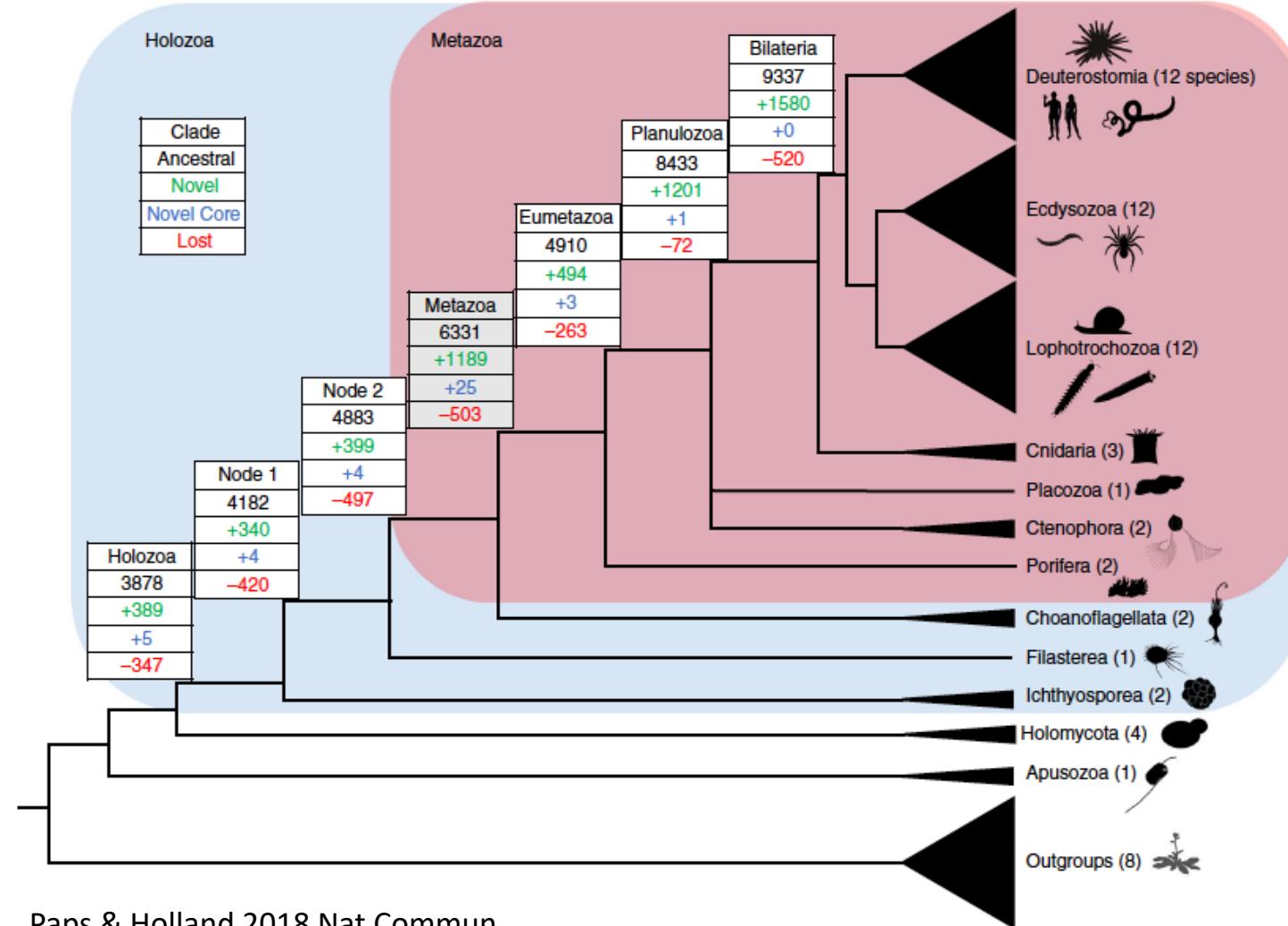


Morlon et al. 2011 PNAS

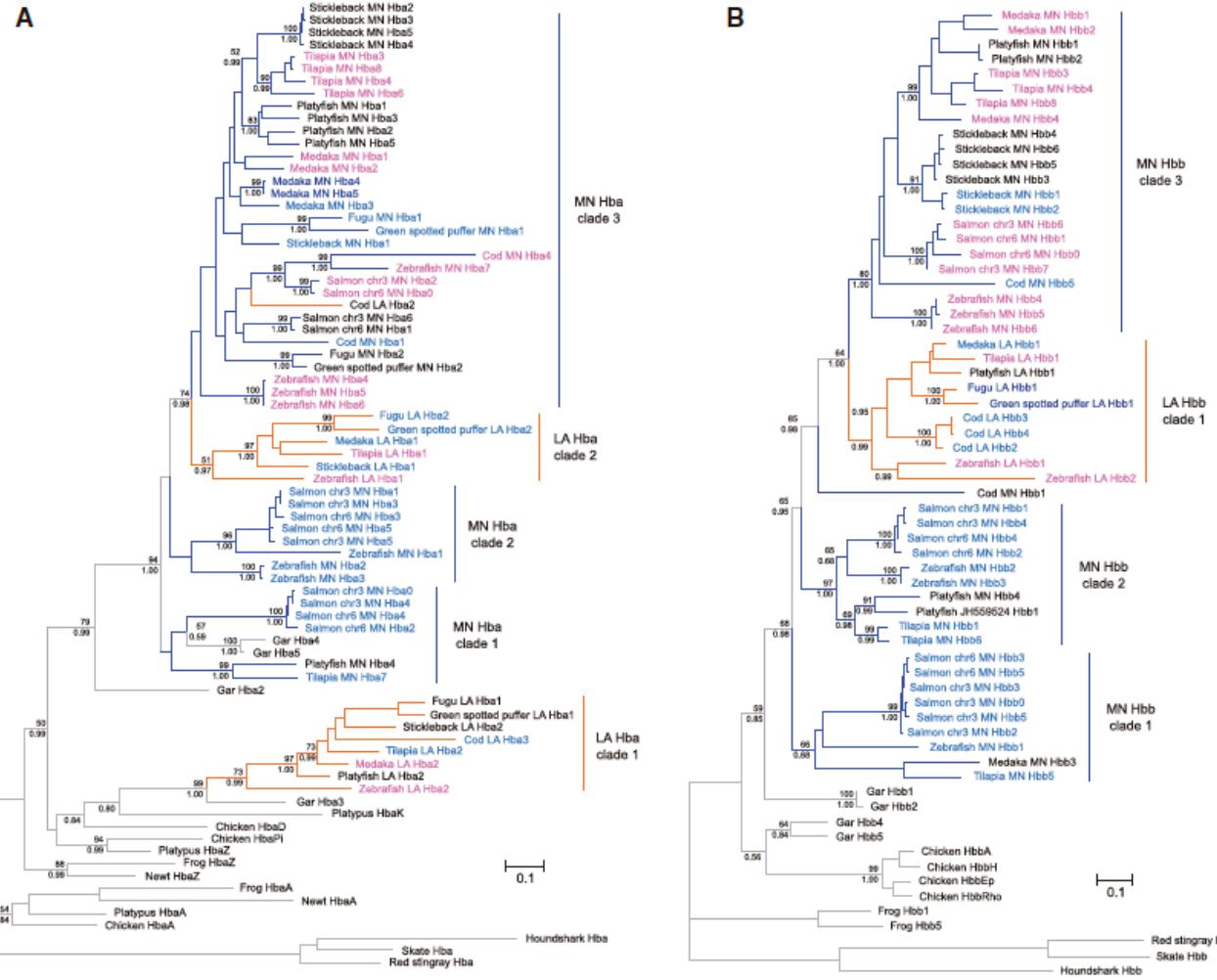
# Molecular clock



# Evolution of genomes and gene families



# Evolution of genomes and gene families



Opazo et al. 2013  
Mol Biol Evol

# Reconstruction of ancestral character states

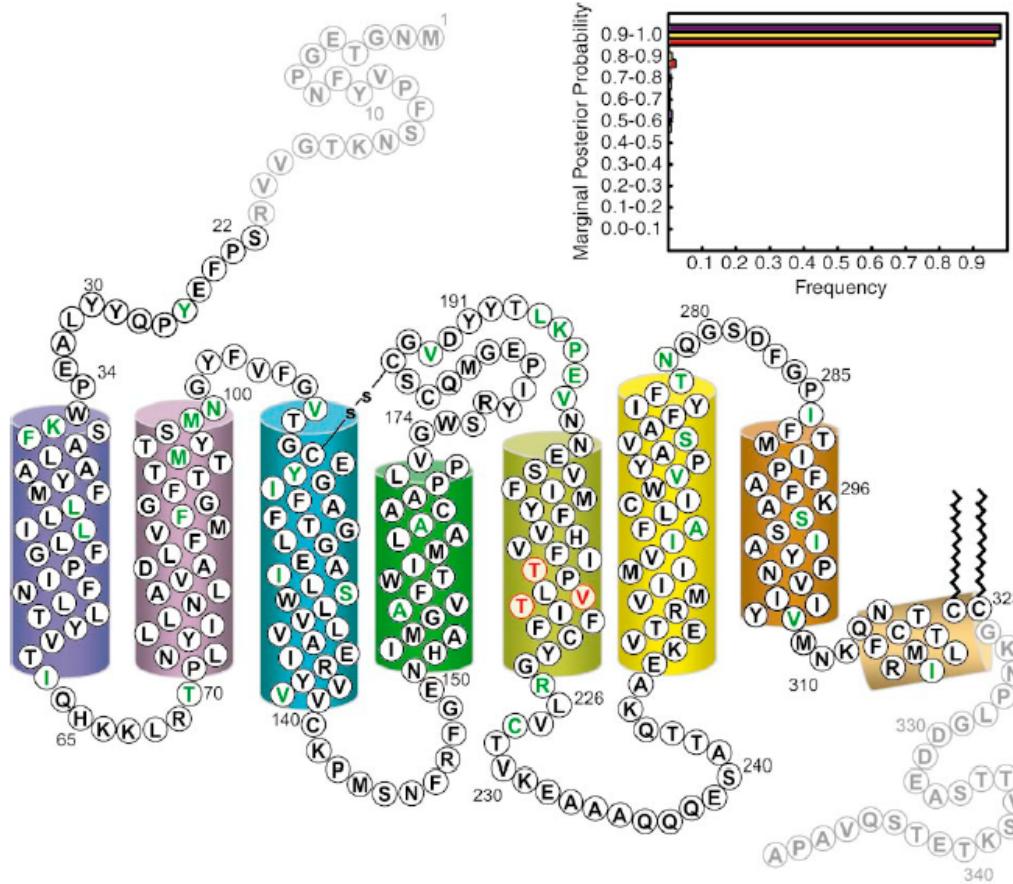
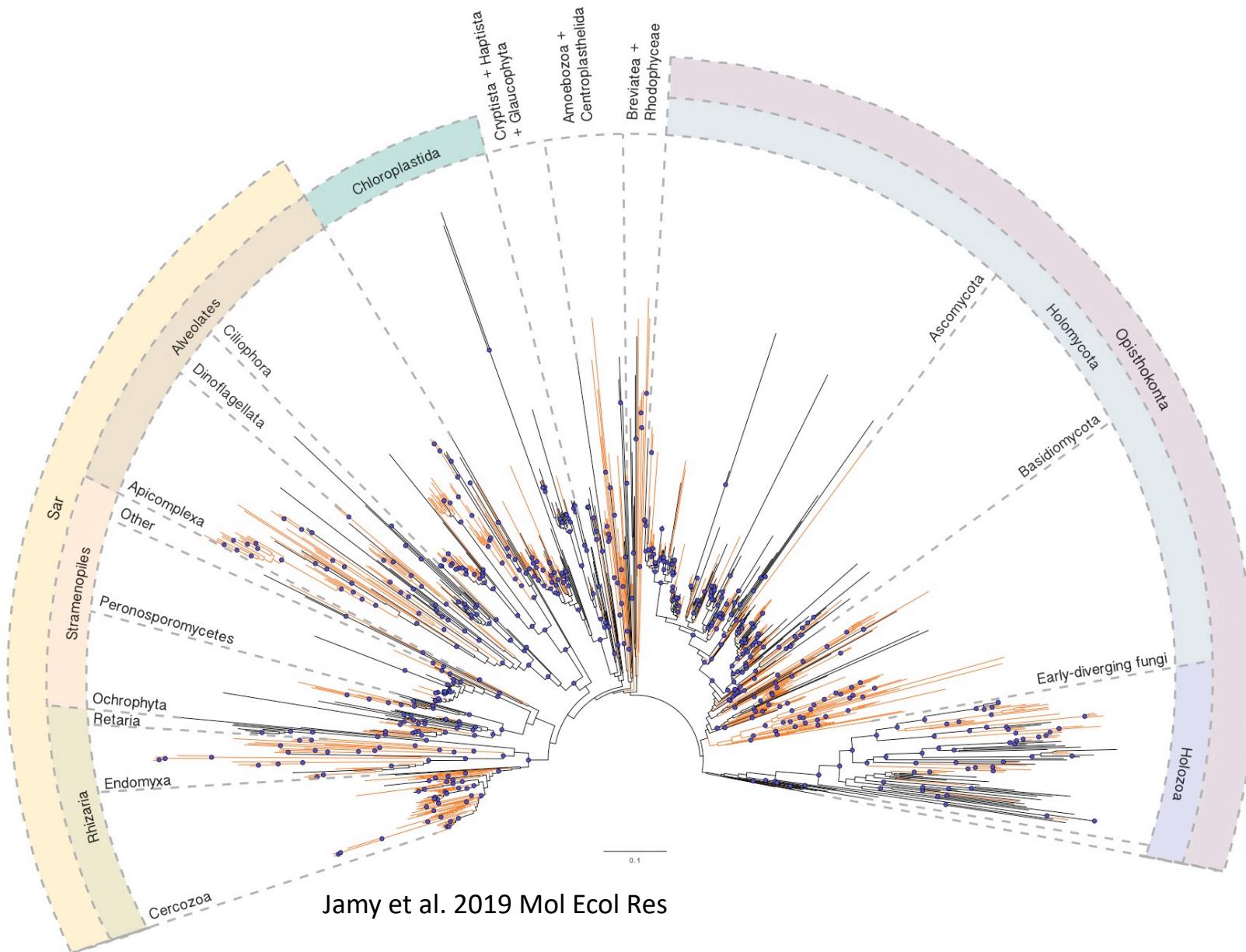


FIG. 2.—Protein sequence of the reconstructed ancestral archosaur rhodopsin drawn as a schematic on the basis of the crystal structure for bovine rhodopsin (Palczewski et al. 2000). Sites that differ among the best three reconstructions, for which variants were also synthesized, are marked in red. The 38 residues that differ from bovine or alligator rhodopsin are marked in green. Inset, frequencies of marginal posterior probabilities calculated for the most likely amino acid reconstruction at each site of the ancestral archosaur rhodopsin sequence. These posterior probabilities, which represent the likelihood of having reconstructed the correct amino acid, under the assumptions of the model used, were calculated for the best-fitting nucleotide (HKY+ $\Gamma$ , purple bars), codon (F61+ $\Gamma$ , yellow bars), and amino acid (Jones+F+ $\Gamma$ , red bars) models.

# Classification, metagenomics



# Study of natural selection

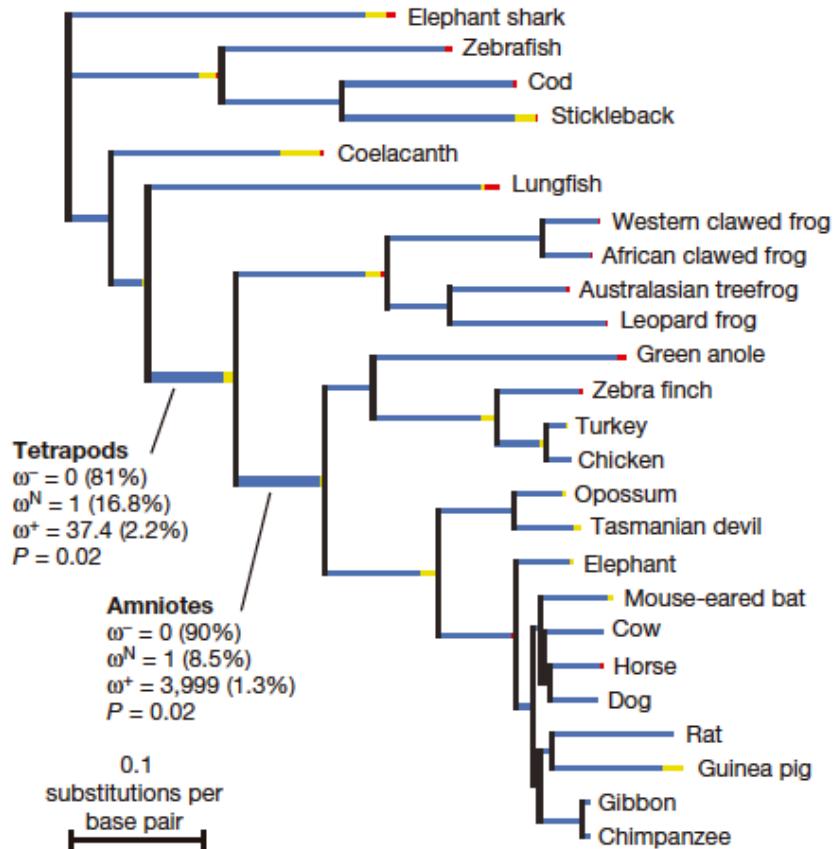
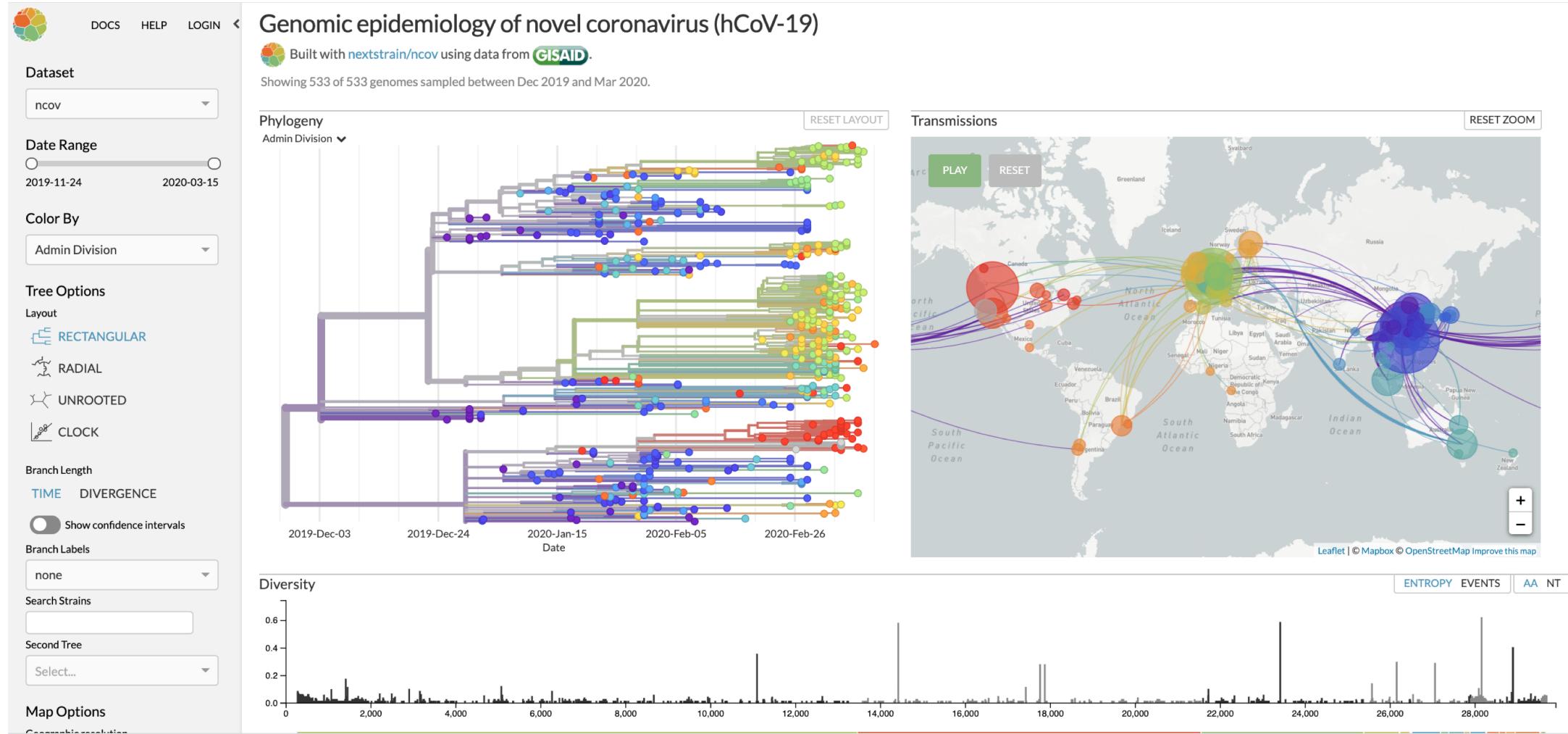


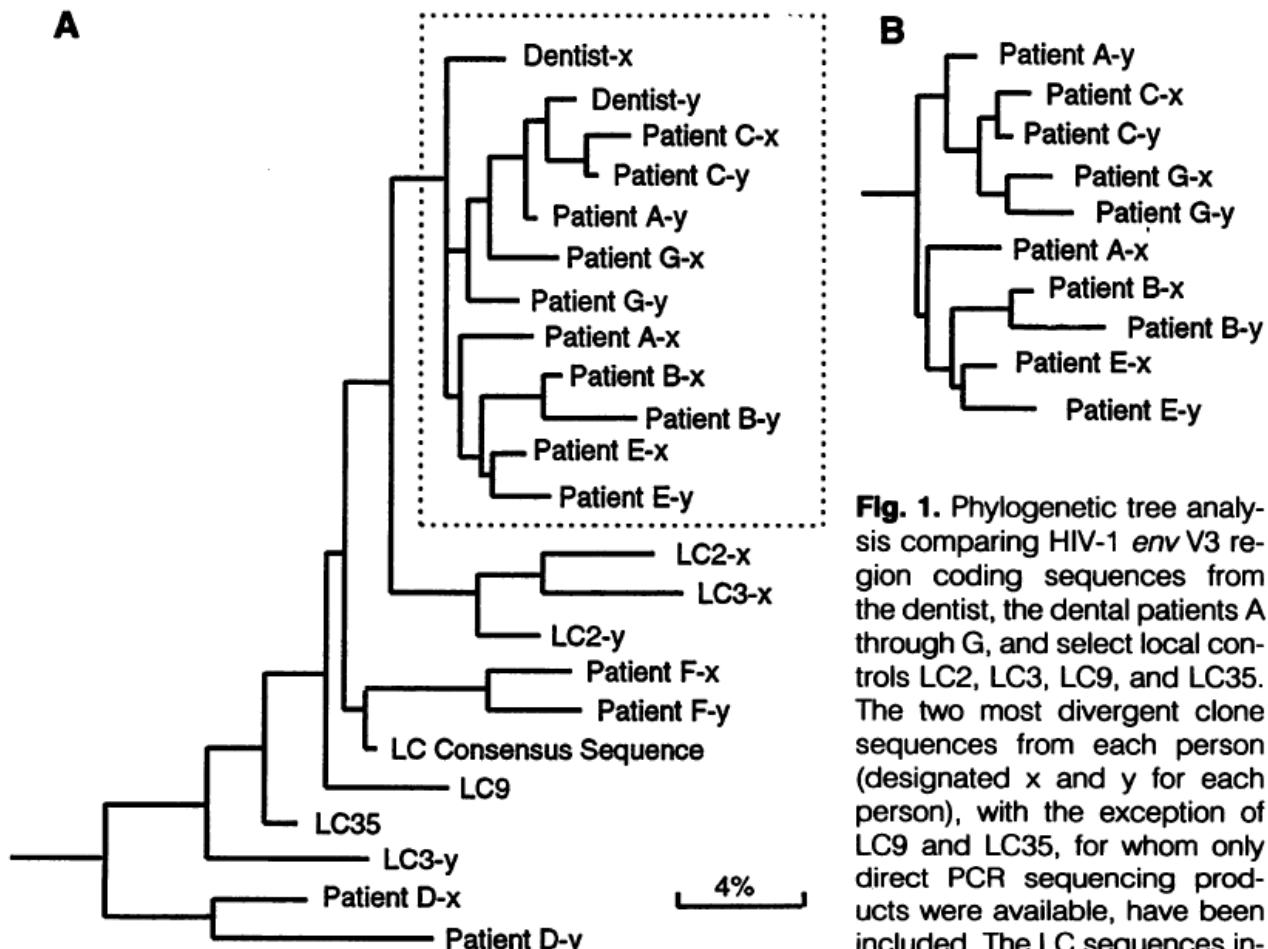
Figure 3 | Phylogeny of *Cps1* coding sequences is used to determine positive selection within the urea cycle. Branch lengths are scaled to the expected

Amemiya et al.  
2013 Nature

# Epidemiology!



# Epidemiology, criminology, medicine



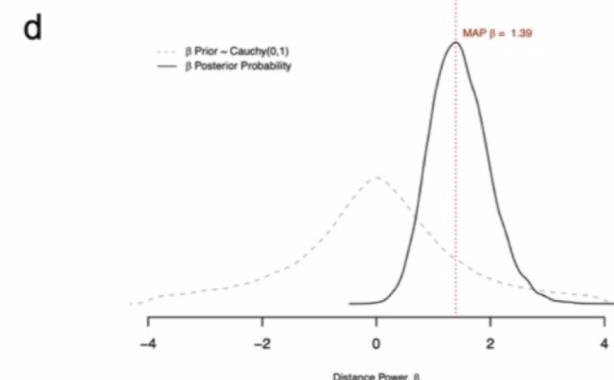
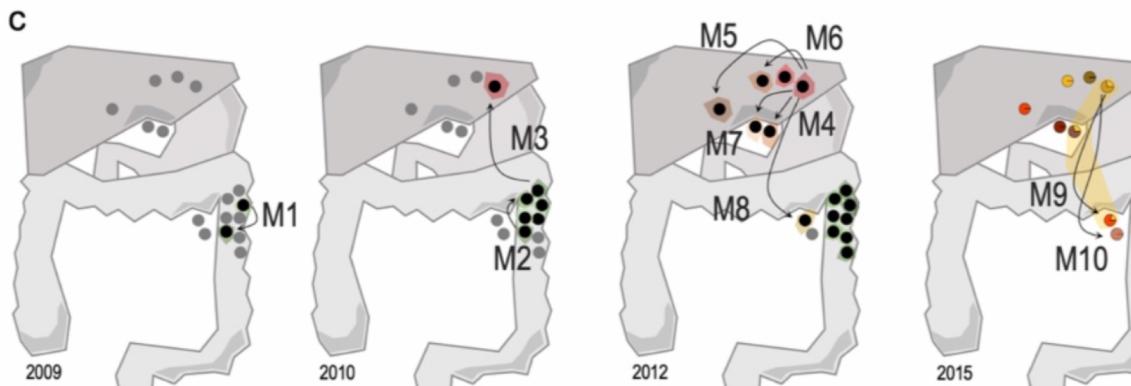
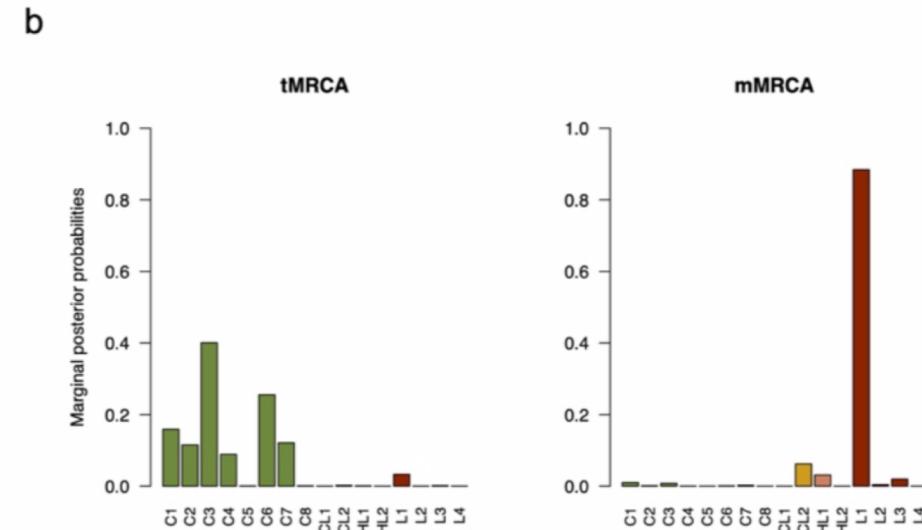
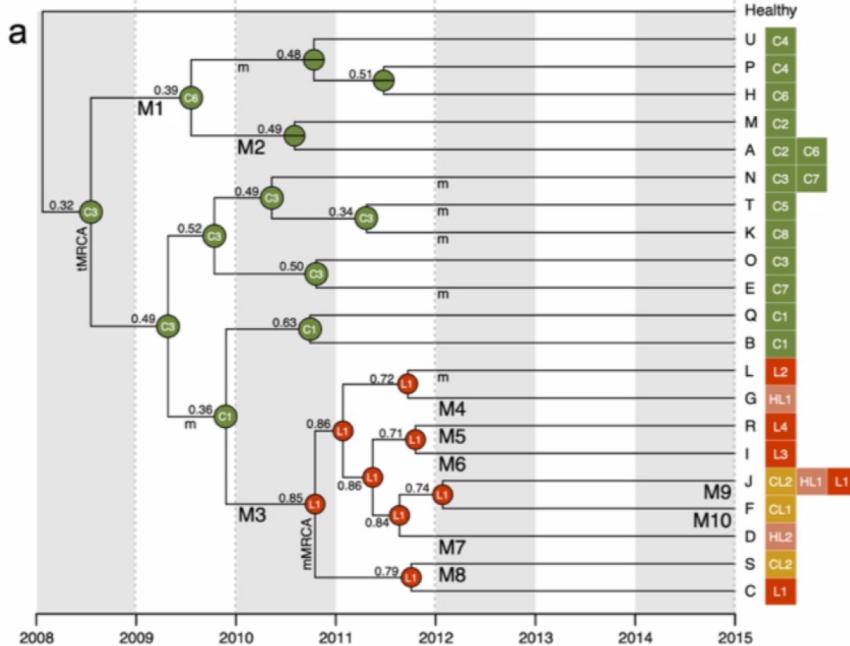
**Fig. 1.** Phylogenetic tree analysis comparing HIV-1 *env* V3 region coding sequences from the dentist, the dental patients A through G, and select local controls LC2, LC3, LC9, and LC35. The two most divergent clone sequences from each person (designated x and y for each person), with the exception of LC9 and LC35, for whom only direct PCR sequencing products were available, have been included. The LC sequences included were those found by

Ou et al. 1992  
Science

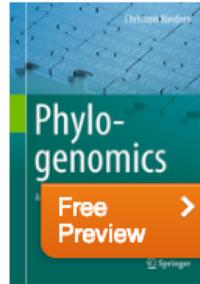
# Cancer genomics

## Rapid evolution and biogeographic spread in a colorectal cancer

Joao M Alves<sup>1,2,3</sup>, Sonia Prado-Lopez<sup>1,2,3</sup>, Jose Manuel Cameselle-Teijeiro<sup>4,5</sup>, David Posada<sup>\*1,2,3</sup>



# Further reading



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## Phylogenomics

An Introduction

Authors: Bleidorn, Christoph

Highly readable overview on the main theoretical and practical aspects of Phylogenomics

### Books

- **Phylogenetics in the Genomics Era.** 2020. Scornavacca, Delsuc, Galtier Ed.  
<https://hal.archives-ouvertes.fr/hal-02535070/>
- **Phylogenomics: An Introduction.** 2017. Bleidorn C. Springer. 222 pp.
- **The Phylogenetic Handbook,** 2<sup>nd</sup> Ed. 2009. P Lemey, M Salemi and A-M Vandamme (eds). New York: Cambridge University Press. 723 pp.
- **Molecular evolution: A statistical approach.** 2014 Yang Z. New York: Oxford University Press. 512 pp.

### Reviews

- Holder M and Lewis PO (2003) Phylogeny estimation: Traditional and Bayesian approaches. *Nat Rev Genet* 4:275–284
- Yang Z, Rannala B (2012) Molecular phylogenetics: Principles and practice. *Nat Rev Genet* 13:303-314.
- Irisarri I, Zardoya R. 2013. Phylogeny reconstruction. In: *eLS*. Chichester: John Wiley & Sons, Ltd.
- Irisarri I, Zardoya R. 2013. Phylogenetic hypothesis testing. In: *eLS*. Chichester: John Wiley & Sons, Ltd.