The Biology of Phylogenetic

Umar Ahmad, Ph.D. [ORCID: 0000-0002-3216-5171]

Bioinformatic Scientist, MOGID Laboratory, ATBUTH Faculty Member, Bauchi State University.











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What is phylogenetics ?

 Phylogenetics is the study of evolutionary relationships among biological entities – often species, individuals or genes (which may be referred to as taxa).

 The major elements of phylogenetics are summarised in this figure.



Motivation

Typically phylogeneticists study one of the following types of question:

- What are the evolutionary relationships or histories among my species/individuals/genes of interest?
- How do sequences evolve?
- Can I better describe processes of sequence evolution with a mathematical model?

Why use molecular data ?

Today, almost all evolutionary relationships are inferred from molecular sequence data. This is because:

- DNA is the inherited material
- We can now easily, quickly, inexpensively and reliably sequence genetic material
- Sequences are highly specific and are often information rich
- In rare cases, where it is not possible to obtain genetic material (e.g. in the case of certain ancient fossil samples), morphological measurements can be used to infer evolutionary relationships.

Why is phylogenetic important?

• Phylogenetics is important because it enriches our understanding of how genes, genomes, species (and molecular sequences more generally) evolve.

• Through phylogenetics, we learn not only how the sequences came to be the way they are today, but also general principles that enable us to predict how they will change in the future.

Application of phylogenetics

- Classification
- Forensics
- Identifying the origin of pathogens
- Conservation
- Bioinformatics and computing
- Adding biological meaning to data



What is a phylogeny ?

 A phylogeny, also known as a tree, is an explanation of how sequences evolved, their genealogical relationships, and therefore how they came to be the way they are today.

 One the first sketches of a phylogenetic tree was made by Charles Darwin. This famous diagram is one of his earlier sketches from a series of notes that he used to develop the idea.



A family tree

• You will probably already know how to interpret patterns of relatedness on a family tree, and it turns out that the same principles apply to phylogenetic trees more generally.



Components of phylogenies

It is important to learn how to read and interpret phylogenies. Some major components of phylogenies:-

- Topology
- Branches
- Nodes
 - Tips
 - Internal nodes
 - Root
- Confidence

Topology

The topology is the branching structure of the tree.

It is of particular biological significance because it indicates patterns of relatedness among taxa, meaning that trees with the same topology and root have the same biological interpretation.



Branches

Branches show the path of

transmission of genetic information

from one generation to the next.



Branch lengths indicate genetic change i.e. the longer the branch, the more genetic change (or divergence) has occurred.

Human	ATGTTGACTC
Mouse	ATGCTGACTC

Nodes

Nodes are the points at the ends of branches which represent sequences or hypothetical sequences at various points in evolutionary history.

Tips

Internal node

Root



(Image courtesy of Andrew Rambaut)

Let's talk about rooting a tree !



Root

The root is the most recent

common ancestor of all of the taxa

in the tree.

It is therefore the oldest part of the tree and tells us the direction of evolution, with the flow of genetic information moving from the **root**, towards the **tips** with each successive generation.



Interpreting patterns of relatedness

The key to interpreting patterns of relatedness in an evolutionary context is to trace back to the point in the tree where taxa share their **most recent common ancestor (MRCA)**.

You can practice this by finding two species of interest in a tree and tracing back along their **branches** to identify the **node** where they meet – this is their MRCA.



Stages in phylogenetics analyses



https://www.jse.ac.cn/EN/10.3724/SP.J.1002.2008.08060

List of phylogenetics software



https://evolution.genetics.washington.edu/phylip/software.html

Phylogenetics resources

EMBL-EBI offers a range of tools and resources that are relevant to the field of phylogenetics:

- Ensembl
- Ensembl genomes
- Ensembl compara
- ClustalW2 Phylogeny
- Clustal Omega and Prank
- EMBOSS Seqret



Summary

- Phylogenetics is the study of evolutionary relationships among nucleotide or protein sequences
- There are many applications of phylogenetics, including forensics, pathogen surveillance, conservation and bioinformatics
- There are several aspects of phylogenies that you need to understand in order to interpret your trees: topology, branch lengths, nodes and confidence.
- Careful interpretation is critical to understanding the biological meaning of phylogenies
- The same phylogenetic tree can be visualised in many different ways
- Evolutionary relationships can be unraveled by identifying the most recent common ancestor (MRCA) shared by species
- EMBL-EBI has several resources and tools that are relevant to the field of phylogenetics including: Ensembl, ClustalW2 Phylogeny, Clustal Omega and Prank

Learning resources

Articles

- Yang, Z & Rannala, B. (2012) Molecular phylogenetics: principles and practice. Nature Reviews Genetics. 13: 303-314.
- Whelan, S., Liò, P. & Goldman, N. (2001) Molecular phylogenetics: state-of-the-art methods for looking into the past. Trends in Genetics. 17: 262–272.
- Jukes TH, Cantor CR. (1969) Evolution of protein molecules In: Munro HN, editor., editor. Mammalian protein metabolism. 3rd ed.New York: Academic Press; p. 21–132.

Published teaching materials

- Baum D.A., Smith SD., Donovan SS. (2005) Evolution: The tree-thinking challenge. Science 310: 979-980.
- Bauldauf, SL. (2003) Phylogeny for the faint of heart: a tutorial. Trends in Genetics. 19:346-351.
- Gregory, TR. (2008) Understanding Evolutionary Trees. Evolution: Education and Outreach. 1:121-137.

Books

- Computational Molecular Evolution (2006). Ziheng Yang, Oxford University Press.
- Inferring Phylogenies (2003). Joseph Felsenstein, Sinauer.
- Molecular Evolution (1997). Wen-Hsiung Li , Sinauer.

Online resources

- Andrew Rambaut's "How to read a phylogeny" tutorial
- "Understanding Evolution" tutorials and resources, University of California Museum of Paleontology
- Joe Felsenstein's comprehensive collection of phylogenetics software

Acknowledgement



