

Phylogenetic tree (phylogeny)

- *Darwin and classification:* In the Origin, Darwin said that descent from a common ancestral species could explain why the Linnaean system is hierarchical. Species in the same genus had a common ancestor more recently than species in different genera. The only figure in the Origin illustrated his idea. He introduced the idea of a phylogenetic tree.
- *A phylogenetic tree represents the history of most groups of species:* For a phylogenetic tree to be a good description of the history of a group, the time it takes for new species to form (represented by the splitting of lineages) has to be very short compared to how long each species was present. The phylogeny of humans, chimps, gorillas and bonobos is typical and thought to be roughly accurate.
- A few groups of plants, such as those in the genus *Clarkia*, do not have a tree-like history. The history of *Clarkia* and other such groups is reticulate (net-like) in which some species have two ancestors.
- *Information in a phylogenetic tree:* The pattern of branching indicates ancestry. The branch lengths indicate times.
- *Cladogram:* A cladogram summarizes the branching pattern but contains no information about times. Be sure to understand the difference between a phylogenetic tree and a cladogram.
- *A phylogenetic tree represents the pattern of evolution:* A tree is a convenient summary of what actually happened. If you could replay the webcast of life, you could discover the tree that describes the true history of a group of species.

Phylogenetic trees and classification

- *Darwin and taxonomy:* Darwin did not address the practical problem of how to use evolutionary history to classify species into higher groups. The problem was then and is still that, although there is a phylogenetic tree that describes the history of most groups, the correct tree is usually not known.
- With the advent of DNA sequencing, much effort is going into inferring the phylogenies of all major groups. The Tree of Life project (www.tolweb.org) is an international effort to join phylogenetic trees of different groups into a single tree that describes the overall pattern of evolution.
- *Terminology:* A monophyletic group is one that contains an ancestral species and all of its descendants. Mammals are a monophyletic group. A monophyletic group is often called a clade. A paraphyletic group is one that contains an ancestral species and some but not all of its descendants. The family Pongidae

(orangutans, chimpanzees, bonobos and gorillas) is paraphyletic. It does not include modern and extinct humans which are descended from a pongid ancestor.

- *Phylogenetic systematics*: The overall goal is to make higher categories reflect evolutionary history by requiring that all taxa be monophyletic.

In a phylogenetic classification, species in the same genus had a most recent common ancestor more recently than species in different genera in the same family, and so on.

Finding the best cladogram

- The problem is to determine the cladogram that describes the ancestry of a group.
- A simple method and one that is easy to apply is parsimony, which is the only one discussed in this course. Other methods, including maximum likelihood, rely on statistical models that make it possible to infer branch lengths as well the branching pattern.
- *Parsimony*: Parsimony assumes that changes in characters or the appearance of new characters occur very rarely. The underlying idea is to choose the tree that requires the fewest changes.
- Counting evolutionary transitions requires that homologous characters be distinguished from analogous characters. Wings in different species of bats are homologous. Wings in bats and wings in birds are analogous.
- A shared derived character is a character that is shared by every species in a group but is not shared with any species outside the group. The assumption of parsimony is that a shared derived character arose once in the ancestor of the group. Monophyletic groups can be distinguished by identifying shared derived characters.
- In order to distinguish between shared primitive characters and shared derived characters, one or more outgroups are needed. Wings are a shared derived character of bats; fur is a shared primitive character.
- *Theory and practice*: In theory, it should be possible to always find the correct cladogram by identifying the right set of shared derived characters. In practice, that is almost never possible because, for a given set of characters and species, there is no cladogram that perfectly fits in the sense that only one change is needed for each character.
- Shared derived characters are homologous. Characters that arose independently in different groups are analogous. The problem is that without knowing the cladogram, it may be difficult to know which characters are homologous and which are analogous. That is true whether the characters are morphological characters or differences in DNA sequence.

DNA sequences provide many characters

- There is increasing use of DNA sequences of gene from different species to infer phylogenetic trees.
- *Molecular clocks*.
- Population genetics tells us that on average differences in DNA sequence evolve at a roughly constant rate.
- Different genes evolve at different rates, which makes them useful for analyzing species that diverged at different times in the past.
- Ribosomal RNA evolves very slowly. The recognition that Archaea and Bacteria were quite different first came from the analysis of ribosomal RNA sequences. Once the groups were distinguished, phenotypic differences between them were then found. Table 27.2 summarizes the differences, but it will not be necessary to know those differences for this course.
- Mitochondria in animals contain small circular DNA molecules (about 16,000 bases in length) that are inherited maternally. Animal mtDNA evolves more rapidly than nuclear DNA, so it is useful for examining variation within species and between species that diverged recently. Mitochondrial DNA of mammoths was sequenced recently. A comparison with the mtDNA of African and Asian elephants shows that the mammoth is slightly more closely related to the Asian elephant than to the African elephant.

Using cladograms to understand evolution

- *Mapping changes on a cladogram*: If the cladogram is known, then the minimum number of transitions of a character can be inferred by using the principle of parsimony. The true number of transitions may be greater, but for many purposes the minimum number is sufficient.
- Wings in mammals evolved only once. Characteristics associated with eating ants and termites evolved three times in mammals.
- The number of times a character changes indicates how difficult it is for natural selection to cause that change. Wings in insects evolved only once. The loss of wings occurred independently in many groups, including fleas, stick insects, parasitic lice and many groups of cave-living insects.

Molecular phylogenetics can be used to understand recent evolutionary history.

- *HIV*: By assuming a molecular clock, the origin of HIV in humans is dated to the 1930s.
- *mtDNA*: Because mtDNA is maternally inherited, the phylogeny of a sample of mtDNA is a tree that represents the ancestry of females.
- *Out-of-Africa*: The most recent common ancestor of all human mtDNA was present in a female who lived between 100,000 and 200,000 years ago, “mitochondrial Eve.” This female was **not** the only ancestor of modern humans

living at that time. Other individuals present at the same time were also ancestors but not of the mtDNA.

- A phylogenetic tree of mtDNA from individuals from human populations shows that individuals from Africa are found throughout the tree, while individuals from other geographic regions are generally found in only small parts of the tree.
- These two conclusions (the young age of the most recent common ancestor and the appearance of African mtDNA throughout the tree) together support the out-of-Africa theory, that all modern humans are descended from a population that lived in Africa less than 200,000 years ago and later dispersed to Europe, Asia and Australia in the past 100,000 years.
- If the out-of-Africa theory is true, then populations of *Homo erectus* represented by fossils that are 500,000 to 1,000,000 years old (Peking Man, Java Man and others found throughout Europe and Asia) went extinct. Only the African population of *H. erectus* left descendants.
- *Neanderthals*: Neanderthals (*Homo neanderthalensis*) lived in Europe and Asia from 300,000 years ago until 30,000 years ago. Between 60,000 and 30,000 years ago they lived in the same general areas as Cro-Magnons, which were modern humans classified as *H. sapiens*. There is no archaeological evidence of contact or interbreeding between the two species.
- MtDNA from Neanderthals and humans had a common ancestor roughly 600,000 years ago. MtDNA extracted from 8 different Neanderthal bones all have three differences from the human reference sequence.¹ No Neanderthal-type mtDNA has been found in any of the thousands of mtDNAs sampled from modern humans, which means that there is no evidence of gene flow from Neanderthals to modern humans.
- Recently, approximately 1 mb of the nuclear genome of a Neanderthal bone from Croatia has been obtained. These sequences have confirmed the conclusion about the time of divergence of the human and Neanderthal lineages and still show no evidence of gene flow.

Forensic use of phylogenetics

- In 2002, a doctor in Louisiana was accused of deliberately infecting someone with HIV by injecting blood from an HIV-positive patient. The victim developed AIDS and died. The doctor was convicted of second degree murder.
- Part of the evidence used in the case was from phylogenetic analysis. HIV from the victim, from the patient alleged to be the source, and from HIV-positive people in the same community were sequenced and a phylogeny was inferred. HIV from the victim was shown to be on the same part of the tree as HIV from the alleged source and quite separate from the HIV from other HIV-positive individuals in the community².

¹ D. Serre et al. PLoS Biology, March 2004

² <http://www.pnas.org/cgi/content/full/99/22/14292>

Sample questions

Q1. What statement provides the best definition of a monophyletic group?

- A. A group of species in the same genus.
- B. A group containing an ancestral species and all of its descendants.
- C. A group containing an ancestral species and some of its descendants.
- D. A group containing an ancestral species and none of its descendants.
- E. A group containing descendants of two ancestral species that are in different taxonomic groups.

Q2. What statement provides the best definition of a phylogenetic classification system?

- A. Taxonomic groups are all polyphyletic.
- B. Taxonomic groups are all paraphyletic.
- C. Taxonomic groups are all monophyletic.
- D. Taxonomic groups are all homologous.
- E. Members of taxonomic groups are all found in the same geographic area.

Q3. The table below contains the nucleotides found at 7 sites in the sequences from four different species. Using the principle of parsimony to choose the best cladogram for these four species. In the cladogram you inferred, which one of the following groups is a monophyletic if species IV is the outgroup?.

	Site						
Species	1	2	3	4	5	6	7
I	T	G	C	A	T	G	G
II	T	A	C	A	C	A	C
III	A	A	C	G	C	G	G
IV	A	A	T	G	T	A	G

- A. I & III
- B. I & IV
- C. II & IV.
- D. II & III
- E. None of the above

Q4. The Pongidae is not a monophyletic family because

- A. Bonobos and chimps are in different genera.
- B. There is more than one species of Pongidae.
- C. Humans are classified as being in a different family.
- D. The divergence of different species in the Pongidae was very recent.
- E. Some species of Pongidae are sexually dimorphic.

Q5. Which one of the following would you regard as the best evidence that there was substantial gene flow between Neanderthals and modern humans?

- A. The nuclear DNA of modern humans is similar to the mtDNA of modern humans.
- B. Some mtDNAs from modern humans are closer to the mtDNA of Neanderthals than they are to mtDNAs of other humans.
- C. There is less sequence variation among mtDNAs of modern humans than we previously thought.
- D. There is more sequence variation among mtDNAs of modern humans than has been found.
- E. There is more sequence variation among mtDNAs of Neanderthals than has been found.

Q6. Which statement best describes the evolution of wings in insects?

- A. Wings originated once and were lost many times.
- B. Wings originated many times and were lost many times.
- C. Wings originated many times and were lost only once.
- D. Wings in insects are homologous to wings in birds.
- E. None of the above.

Correct answers: b, c, e, c, b, a