

## CHAPTER FOUR

# Trait Evolution

If the realm of phylogenetics were limited to descent from common ancestry, it would not be able to explain something as basic as the fact that living organisms are not all identical. Because evolution is about descent *with modification*, our conceptual framework needs to accommodate the phenomenon of trait evolution. *Traits* (also called characters, characteristics, or phenotypes) are features of organisms that arise through the expression of an organism's genetic makeup in a particular environment. In this chapter, we overlay trait evolution onto the concept of phylogenies offered in Chapter 3. We then discuss how changes in traits can be represented on tree diagrams and provide a brief introduction to the concept of homology.

### TRAIT EVOLUTION IN A SINGLE LINEAGE

Imagine a population of sexually reproducing plants with petals that are dark red. Figure 4.1 shows such a population. This population is very small, but the principles scale up easily. In generation two, a mutation occurs in a gene that is involved in pigment production in flowers. The mutant form of the gene (the mutant *allele*) cannot produce the red pigment. Individuals with two copies of the mutant allele (homozygotes) have yellowish-cream petals, due to the accumulation of the biochemical precursors to the red pigment.

In the figure, each organism is represented by two circles corresponding to the two alleles at the flower pigment locus. Black circles represent the functional allele, which produces red pigments, and gray circles represent the inactive alleles that produce no red pigment (and thus yield cream flowers in the homozygous condition). We will refer to the functional, red allele as the *ancestral* allele, because it was present in all members of the ancestral population. We will refer to the inactive, cream allele as the *derived* allele to communicate

the fact that it was derived from the ancestral allele by mutation. Individuals whose flowers are cream colored are surrounded by an oval.

As shown in Figure 4.1, the frequency of the derived allele increases over the course of several generations. The genetic composition of the population has changed: it has evolved. The speed with which the frequency of the cream allele increases suggests that it is favored by natural selection. However, evolution does not require selection; such changes in allele frequency can also occur by chance, a phenomenon called *genetic drift*.

In this example, the ancestral red allele is ultimately lost from the population. In population genetics terms, we would say that the population has become *fixed* for the derived allele. It is worth highlighting that the loss of the ancestral red allele, which occurs when the derived allele goes to fixation, is irreversible. If the entire population complex (comprising all populations linked by occasional gene flow) becomes fixed for the derived allele, the ancestral allele could recur only by a reverse mutation, which is highly improbable.

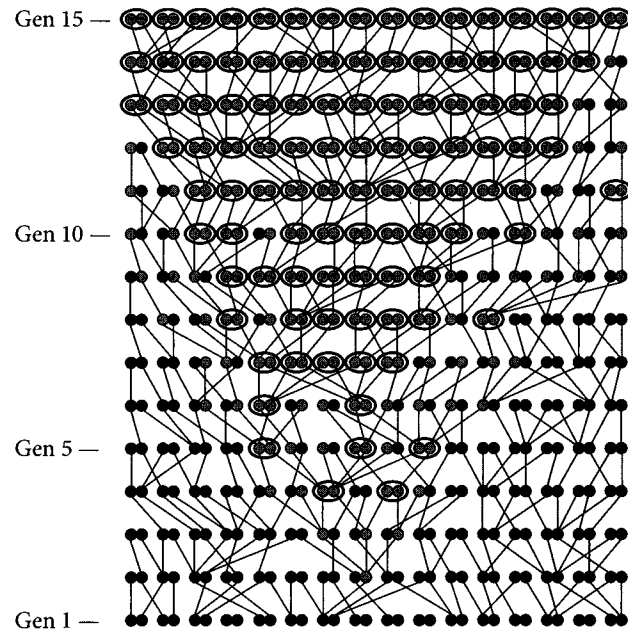


FIGURE 4.1 Fixation of a derived allele in a population over 15 generations.

Looking at the level of the organism's appearance over the generations depicted in Figure 4.1, phenotypic evolution has occurred. The population was originally fixed for the *ancestral character state*, red petals. After 15 generations the population became fixed for the *derived character state*, cream-colored petals. In short, we would say that cream-colored petals evolved from red petals.

The time from mutation to fixation of a derived character state will typically take much longer than 15 generations, especially when selection favoring the derived allele over the ancestral allele is weak (refer to population genetics or evolutionary biology textbooks for more details). Also, many derived character states require the accumulation of changes in multiple genes or multiple successive mutations at a single locus. It can take tens, hundreds, or thousands of generations to transition from a population fixed for an ancestral character state to one fixed for a derived state. Nonetheless, in the time frame of an entire phylogenetic tree, even 100,000 generations is a brief time interval. Therefore, it is often a convenient simplification when considering trees to view a derived trait as having arisen in an evolutionary instant. This is why trait evolution is most commonly depicted on phylogenetic trees as a line or bar across a branch. This does not mean that the derived character state arose and went to fixation in one generation. It simply indicates that trait evolution happened quickly relative to the rate of lineage splitting.

Many kinds of traits can evolve along phylogenetic lineages. All that is required is some degree of *heritability*, which is defined as a tendency for offspring to resemble parents. It is probably obvious that the rules governing morphological (i.e., physical) traits, would apply also to heritable behavioral, physiological, and biochemical traits. In each case the trait is the result of a heritable genetic or developmental program unfolding in a particular environmental context. But one class of trait may require special clarification: geography.

Nearly all organisms have only limited dispersal ability, which means that geographic location is a heritable trait. A squirrel in Germany is relatively unlikely to have offspring that live in Ecuador, or even in Italy. Because of this dispersal constraint, geographic locale can often be marked on trees very much like more typical traits. For example, as discussed in Chapter 1, "living in the New World" probably evolved from "living in the Old World" along the branch leading to the New World monkey clade. As this case illustrates, biologists have made good use of the phylogenetic perspective to reconstruct the paths by which different species have acquired their geographic ranges (Chapter 11).

TRAIT EVOLUTION IN A BRANCHING TREE





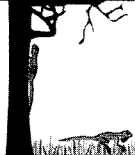


We noted above that once a mutant allele has gone to fixation, the ancestral allele is extremely unlikely to recur. This means that descendant lineages are all expected to possess the derived character state. This principle of descent with modification is all that is needed to extend our model of trait evolution from single lineages to whole branching phylogenies, whether of species or asexual clones. For example, once the ancestral mammal lineage became fixed for the production of hair, all its descendants inherited this trait. Although the color, length, texture, and distribution of hair subsequently evolved, the character state—“hair”—was retained through the many lineage-splitting events that gave rise to the approximately 5400 species of living mammals. Even dolphins and whales have some hair as fetuses.

In some cases a trait can evolve to resemble an earlier state, but this is not truly a reversal of evolution. Snakes have lost their limbs, but this does not mean that they have reverted to the state of the fishlike organisms that preceded the initial origin of walking legs: snakes do not have fins. And even in cases where a trait is indistinguishable from that of an ancestor, it is better viewed as a case of separate evolution rather than a reversal to the ancestral condition.

Before progressing further, let us clarify some terminology. In phylogenetics, it is conventional to distinguish *characters* from *character states*. A character is an attribute that potentially varies among the tips. For example, when considering mammals, hair color could be a character. Character states, in contrast, refer to alternative versions of the character that could occur in different organisms. For example, brown hair and black hair are alternative states of the character “hair color.” It is important to note that characters and character states are understood relative to the variation seen among a set of taxa. If, for example, we were looking broadly across the land vertebrates, “hair” might be a character state of the character “integumentary outgrowth,” whose alternative states might be “none,” “scales,” “feathers,” or “hair.”

To help visualize the process of trait evolution along the branches of a phylogenetic tree, consider a hypothetical example involving an ancestral lizard lineage, diversifying over time to yield six descendant species. Among the many characters that evolve somewhere on this phylogeny we track eight traits, whose ancestral character states are listed in Table 4.1. This example is based on Phylostrat, a computer program for exploring trait evolution along the branches of a phylogeny. The program is freely distributed by Jon Herron of the Univer-

TABLE 4.1 Characters and character states in lizards

	Character	Ancestral state	Derived state
	Crest on head	Absent	Present
	Colored collar	Absent	Present
	Preferred prey	Insects	Worms
	Pattern on back	Stripes	Mottled
	Tail spines	Absent	Present
	Habitat	Ground dwelling	Tree dwelling
	Tail spots	Present	Absent
	Dewlap (flap of skin under chin)	Absent	Present

sity of Washington (<http://faculty.washington.edu/herronjc/SoftwareFolder/PhyloStrat.html>).

Starting from one ancestral lizard lineage, five branching events occurred to give rise to six terminal species. In the course of this branching process, the traits evolved from the ancestral to various derived states on different lineages, as shown in Figure 4.2. Assume that this is the true tree and the actual history of trait evolution.

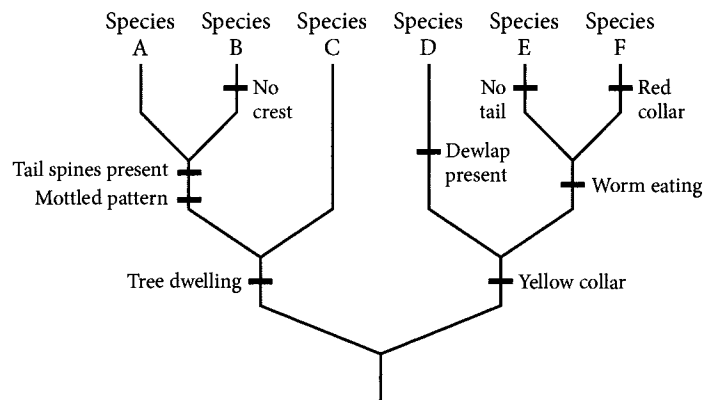


FIGURE 4.2 Trait evolution in six species of lizards. Table 4.1 lists the ancestral character states.

In the format we have chosen, character state changes are drawn with a mark at an arbitrary position along a branch. Changes marked on a lineage occurred somewhere along its length. Recall, from Chapter 3, that lineage branching primarily involves the geographic fragmentation of population lineages, which then *allows* the isolated lineages to undergo independent evolution. As a result, the evolution of derived traits that differentiate descendant lineages usually occurs after lineages have become genetically isolated. For this reason, we recommend against marking character state changes at nodes since such a placement might lead one to mistakenly assume that character evolution *caused* lineage splitting.

You should also notice that the distribution of changes is not entirely even; some branches have no changes, whereas one branch has two changes. Trait evolution is not a fully predictable phenomenon. Given enough time, some trait evolution is likely to occur in any descendant lineage, but it can occur at different rates in different lineages, and may or may not include changes in the particular subset of characters that we are considering.

In cases where multiple changes occur on the same lineage, we often do not know which happened first. The convention is to mark the two changes in an arbitrary order. For example, in Figure 4.2 the evolution of a mottled pattern is shown as having occurred before tail spines, but actually the order is probably unknown. When reading a tree that has multiple evolutionary changes marked

on a single branch, one should not assume that the order of events is accurately represented.

The figure conveys which character states occur in each of the six tips. Take the list of ancestral states, as found at the root, and then note the characters that changed on the path from the root to the tip. For example, individuals of species A have the ancestral states for five of the characters (head crest, insect eating, no collar, tail, and no dewlap) but have the derived states for three characters (living in trees, having a mottled pattern on their backs, and having spiny tails). You may find it useful to make up a list of the characteristics of the other five species and then check it against Table 4.2.

This example involves a few lizards, but the principles apply equally to the entire tree of life. The heritable features that we see in any living species, whether in morphology, physiology, biochemistry, geography, or behavior, are those traits that evolved somewhere in the species’ ancestry. If you could trace a path from the origin of life through the full branching phylogeny to any tip, the set of traits seen in that tip would be the aggregate of all the derived traits evolving along that path. Organisms can be viewed as summations over their evolutionary history.

These considerations explain why trees are such important tools for organizing information about biological diversity. Since evolution occurs along a

TABLE 4.2 Character states in each of six lizard species (Sp. A–F) based on the character mapping in Figure 4.2

Character	Character states					
	Sp. A	Sp. B	Sp. C	Sp. D	Sp. E	Sp. F
Crest on head	Present	Absent	Present	Present	Present	Present
Colored collar	Absent	Absent	Absent	Yellow	Yellow	Red
Preferred prey	Insects	Insects	Insects	Insects	Worms	Worms
Pattern on back	Mottled	Mottled	Stripes	Stripes	Stripes	Stripes
Spiny tail	Present	Present	Absent	Absent	Absent	Absent
Habitat	Tree	Tree	Tree	Ground	Ground	Ground
Tail	Present	Present	Present	Present	Absent	Present
Dewlap	Absent	Absent	Absent	Present	Absent	Absent

tree, keeping track of the tree allows us to store information about diversity in a maximally efficient way. This can be seen quantitatively in this example. With eight traits and six species, you could memorize each species' state for each character, requiring you to store 48 pieces of data. Alternatively you could memorize the tree (composed of four clades), the eight ancestral states, plus the nine character state changes (listed beside the clade with which they are associated) for a total of 21 data points. You require less than half the brain space to store these same data using the tree framework than you would if you just memorized the data table.

As the number of tips and traits increases, the advantage of tree thinking becomes even more apparent. It is certainly easier to memorize that hair is a trait of all 5400 living species of the mammal clade than to have to put a mental check by the trait hair 5400 times. Furthermore, a single tree, for example, the tree of major vertebrate groups, can help you make sense of dozens of important traits.

Once you have developed an ability to think clearly about trees, you can start building a mental sketch of the full tree of life and gradually flesh it out by attaching more and more traits of interest. Any trait present in living organisms evolved somewhere on the tree of life. Traits found in all cellular organisms (the genetic code, glycolysis, DNA, ribosomes, etc.) can be attached to the **stem lineage** of all life (i.e., the lineage leading to the last common ancestor of all living organisms). Likewise the traits of particular living species will map at some depth within the tree, which tells you how widely that trait is shared with other organisms. Some human traits, such as written language or hunting with projectiles, evolved recently and are shared with no other living species. In contrast, other human traits, such as having a cell with a nucleus, map nearer to the root of the tree of life and are shared much more widely.

## ANCESTRAL AND DERIVED CHARACTER STATES

While it is inappropriate to label an organism or taxon as either “ancestral” or “derived” (Chapter 3), this terminology is quite appropriate for character states. As discussed in relation to Figure 4.1, a derived character state is one that evolves from an ancestral character state due to fixation of derived alleles. Thus, any character with two alternative states within a certain group of organisms has a **polarity**, a direction of evolution: one state is ancestral and the other

is derived. Determining a character's polarity is important from a phylogenetic perspective because ancestral and derived character states provide different kinds of phylogenetic evidence.

One way that scientists indicate that a concept is important is to associate it with new terminology. This is illustrated well by Willi Hennig, the father of phylogenetic systematics (Chapter 2), who coined novel terms for ancestral character states, **plesiomorphies**, and derived character states, **apomorphies**. Why, you may wonder, did Hennig think that these concepts deserved their own scientific-sounding terms?

When you find that a taxon is divided into two subgroups, one with character state 1 and the other with character state 2, your conclusions about phylogenetic history depend upon whether 1 or 2 is the apomorphic (derived) character state. As Hennig emphasized, shared, derived character states (**synapomorphies**) should be associated with clades. In contrast, groups of organisms sharing ancestral character states (**symplesiomorphies**) need not be clades. Figure 4.3 is reminiscent of Figure 3.10 but adds trait evolution to the picture.

In Figure 4.3a crocodiles and birds share a derived (apomorphic) character state, marked with a black bar. For example, the trait of laying eggs in nests that are tended by the parents may be an apomorphy shared by birds and crocodiles. This indicates that these taxa both descended from the branch on which the derived state arose, consistent with them forming a clade. In Figure 4.3b, lizards and crocodiles share a plesiomorphic or ancestral state (e.g., scales), while birds have an apomorphic or derived state (e.g., feathers). Although lizards and crocodiles have identical character states, because these traits are plesiomorphic, we cannot assume that lizards and crocodiles form a clade. Indeed, we know that lizards plus crocodiles is a non-monophyletic group because of

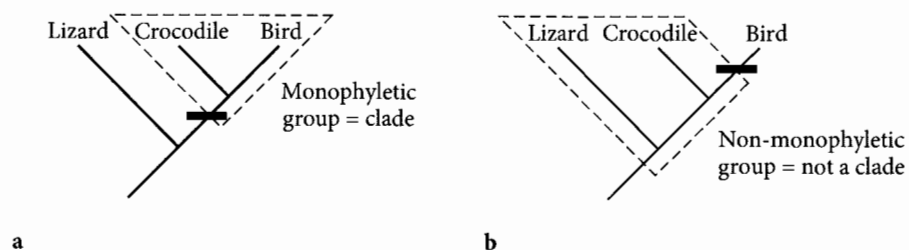


FIGURE 4.3 The association between character polarity and monophyly. Black bars indicate changes in character state.

other information, including traits whose evolutionary history resembles Figure 4.3a.

Using this logic, Hennig and many subsequent workers placed great emphasis on identifying synapomorphies. As described in Chapter 7, polarizing character states formed a critical step in Hennigian phylogeny reconstruction. While this method for phylogenetic inference is no longer used, it is still important to keep track of character polarity because only synapomorphic character states are expected to be associated with monophyletic groups.

THE EVOLUTION OF DNA SEQUENCES

The evolution of visible (phenotypic) characters is the result of changes at the molecular level—specifically, changes in the DNA sequence of some part of the genome. While not all evolutionary changes in a DNA sequence will result in phenotypic changes (some are *silent*), almost all heritable changes in phenotypes are attributable to changes in DNA sequences. If phylogenetic trees are to provide a comprehensive framework for thinking about evolution, they must also accommodate molecular sequence evolution.

A DNA strand contains a series of nucleotides that is copied during DNA replication. Although genomic DNA occurs as a double-stranded helix, molecular evolution is typically modeled as a single strand. Because of complementary base pairing, the strand not shown can be determined entirely from the sequence of its complement. Figure 4.4 depicts one parental DNA strand of 13 bases, its two children, and its four grandchildren.

Each nucleotide position in a daughter sequence is copied from a particular position in its parent sequence. A nucleotide position in an offspring is defined to be *homologous* to that in a parent if the former was copied from the latter during DNA replication. Molecular homology of nucleotide positions is independent of the fact that copying is imperfect. A mutation occurring at a nucleotide position does not change that position's homology. For example, position number 7 is homologous in all the grandchildren despite the fact that two have A's and two have T's at this position. Pairing during DNA replication and the *tendency* for the daughter strand to match the parent strand is what defines molecular homology.

Sequence evolution involves changes in the nucleotides occupying particular positions in the sequence. As the shaded bases highlight, mutations have arisen

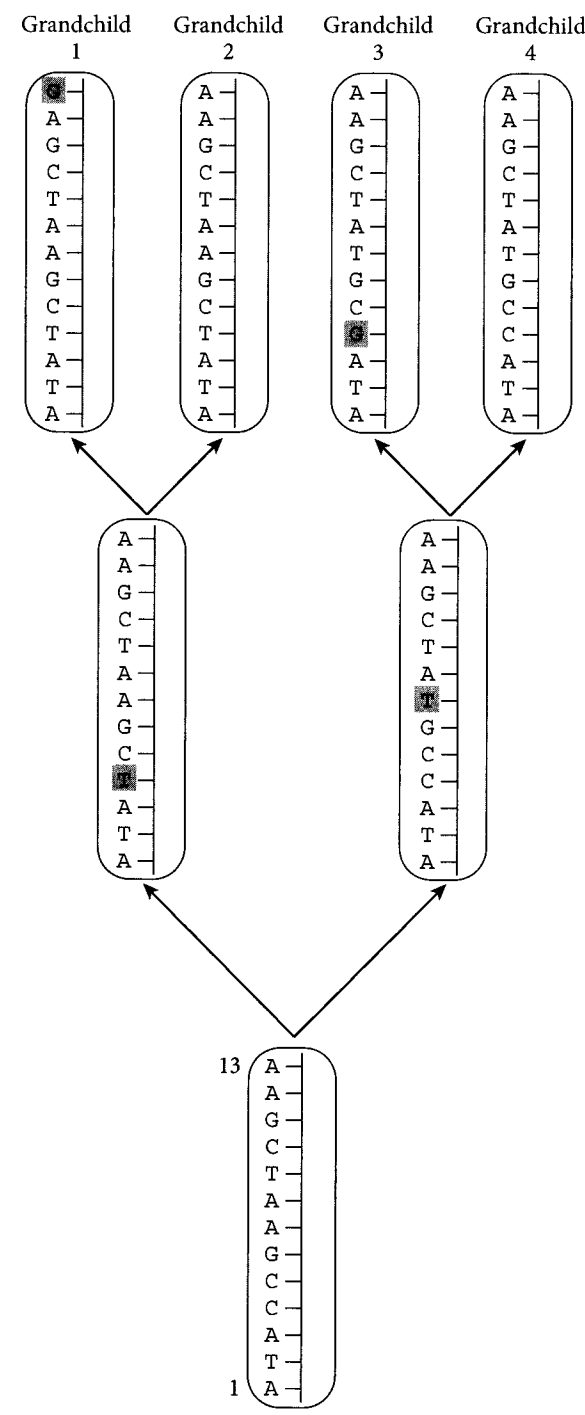


FIGURE 4.4 Changes in DNA sequences during two rounds of replication. Mutated positions are highlighted

(either during DNA replication or at another time in the life of the organisms) so that the four grandchildren have distinct sequences from one another and from the ancestral sequence. We call the change of one base into a different base at the same position a *substitution*. For illustrative purposes, we have assumed a higher rate of substitution than is typically seen in real data.

If you were given the ancestral sequence and told which changes had occurred on the lineage leading to grandchild 1, you could infer the sequence of grandchild 1. The principles here are exactly the same as for phenotypic traits such as tails and crests. That is to say, a sequence is the sum of all changes that occurred at some point during its evolution, overlaid upon the starting sequence.

The phylogenetic treatment of DNA sequences is very similar to the treatment of traits such as morphology. In the case of sequence evolution, each position in the sequence is a character and the bases occupying that position are character states. Thus, in reference to Figure 4.4, we could state that for character 13, grandchild 1 has the derived character state, G, whereas the other three grandchildren have the ancestral character state, A.

This description of molecular sequence evolution is somewhat simplified. We have ignored the fact that, as well as substitutions, previously exist-

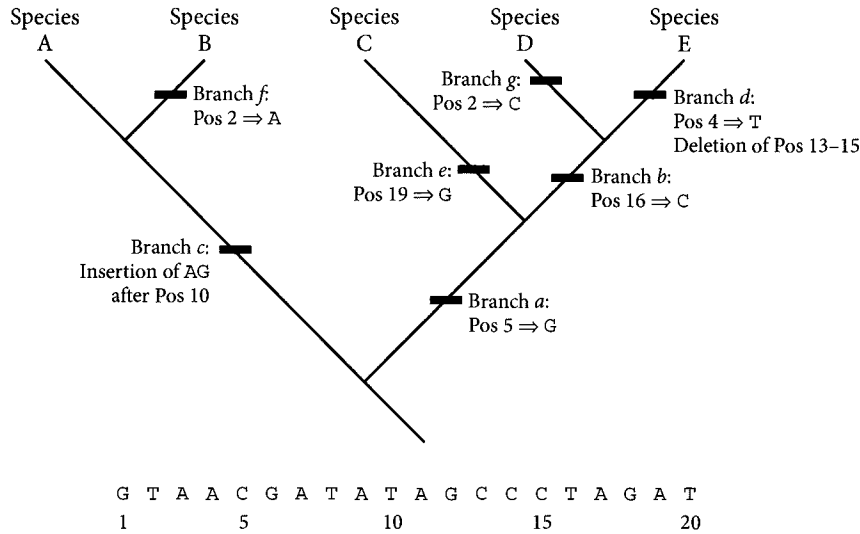


FIGURE 4.5 Example of DNA sequence evolution in five species. The ancestral sequence is provided at the bottom and subsequent changes are marked with black bars. Pos = Position.

ing bases can be deleted or new bases can be inserted into a sequence. When bases are inserted into a sequence, they have no locally homologous positions (though they may be copied from some distant part of the genome). Thus, from the point of view of this evolving sequence, these characters appeared from nowhere. An example of a *deletion* is shown on branch *d* in Figure 4.5, whereas branch *c* includes an *insertion*. If we knew the true history of insertions and deletions, we could align the sequences such that homologous positions are above one another. Chapter 7 provides more details on sequence alignment and the treatment of insertion/deletion (*indel*) events. The correctly aligned sequences from the tips of Figure 4.5 are shown in Table 4.3.

While there are many similarities in the way that DNA sequence characters and other kinds of characters evolve along a tree, there is one important difference. The characters that make up a DNA sequence are physically connected in a specific order. The order in which morphological characters are listed in Table 4.2 is arbitrary, whereas the order in which the sequence characters (positions) are listed in Table 4.3 is constrained by their order in the sequence.

The physical connectedness of the positions in a DNA sequence might lead you to wonder whether their evolution is truly independent. Even under the assumption that an entire gene sequence shares the same gene tree (Chapter 6), evolutionary changes from one character state to another at one position are generally independent of character state changes at other positions in the sequence. The physical connections between adjacent positions do not, in themselves, result in nonindependent evolution. This principle explains why different positions in a DNA sequence, like different morphological characters, can be viewed as providing independent pieces of evidence on the structure of the true tree (Chapters 7 and 8).

TABLE 4.3 The sequences of the five terminal species shown in Figure 4.5

Taxon	Position																			
	1	2	3	4	5	6	7	8	9	10	10a	10b	11	12	13	14	15	16	17	18
Sp. A	G	T	A	A	C	G	A	T	A	T	A	G	A	G	C	C	C	T	A	G
Sp. B	G	A	A	A	C	G	A	T	A	T	A	G	A	G	C	C	C	T	A	G
Sp. C	G	T	A	A	G	G	A	T	A	T	-	-	A	G	C	C	C	T	A	G
Sp. D	G	C	A	A	G	G	A	T	A	T	-	-	A	G	C	C	C	C	A	G
Sp. E	G	T	A	T	G	G	A	T	A	T	-	-	A	G	-	-	-	C	A	G

## AN INTRODUCTION TO HOMOLOGY

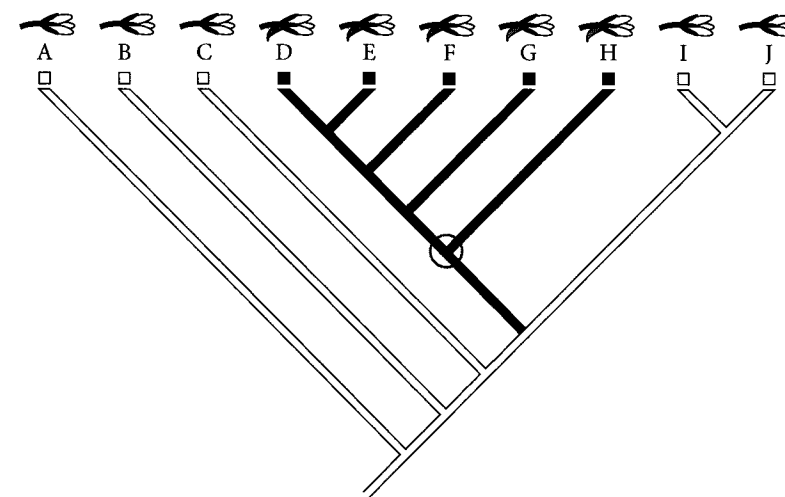
It is natural to look at two humans and note that both have the “same” characters, for example, nostrils. While that character may take on alternative states (e.g., varying in size or shape), nostrils themselves are taken to be equivalent in some deep way. Likewise, you might entertain the possibility that human nostrils are the “same” as the blowholes of whales. Conversely, you probably appreciate that the wings of insects and birds are not the “same,” despite bearing the same name—wing—and being used for flight.

The technical term used to refer to evolutionarily equivalent characters in different organisms is **homology**. As we saw earlier, nucleotide positions in two DNA molecules are considered homologous if they trace back to the same nucleotide position in an ancestor. Phenotypic traits like nostrils are not directly inherited—kids’ noses are not direct copies of their parents’ noses. Nonetheless, so long as a phenotypic trait is heritable, we can conceive of traits as coming from ancestors. Consequently, homology may be defined for phenotypic traits similarly to nucleotide positions: two traits are evolutionarily equivalent, that is, homologous, if they trace to the same trait in a common ancestor.

Given this definition of homology, the claim that nostrils and blowholes are homologous would be supported by showing that the common ancestor of whales and humans had nostrils that became modified over evolutionary time to become the human and whale versions. Likewise, the lack of homology of bird and insect wings would be supported by evidence that the last common ancestor of birds and insects did not have wings. This evidence could come from phylogenetic analysis (as in the examples below) but also from analysis of the trait’s structure and development. There are some complications, which make homology one of the most difficult concepts in evolutionary biology. However, the key point here is simply that, because homology invokes common ancestry, it is an inherently phylogenetic concept.

To clarify the relationship between phylogenetic trees and homology, we will consider an example involving nectar-containing spurs in some hypothetical flowering plant species. We will run through three possible histories of trait evolution and we will ask, In which cases is the spur of species D homologous to the spur in species H?

Figure 4.6 depicts a case in which the last common ancestor of species D and H (indicated by the circle) had a spur, and both lineages retained this spur



**FIGURE 4.6** An evolutionary scenario under which the spurs in species D and H are homologous. Branches in black ended with spurred flowers, branches in white ended with spurless flowers.

from that point on. We have used a common format for depicting character evolution in which branches are colored based on their final character state (the state at the tip or just before the branch divided into two descendant lineages). Based on this history of trait evolution, the spurs in species D and H are homologous.

In Figure 4.7, the last common ancestor of D and H (again marked with a circle) lacked spurs. Spurs were acquired independently in the lineages leading to D and H. Because the spurs in D and H arose independently, they are the products of **convergent evolution**. As a result, they are not homologous. This would be true even in the unlikely event that these independently evolved spurs utilized a similar developmental pathway to achieve their distinctive form. While similarity of development and structure provide good initial evidence that traits are homologous, homology is defined based on common ancestry, not similarity.

Figure 4.8 shows a case in which the common ancestor of D and H had a spur but, nonetheless, the traits are not homologous. This is because, on the lineage leading to D, the original spur was lost, but then a similar structure



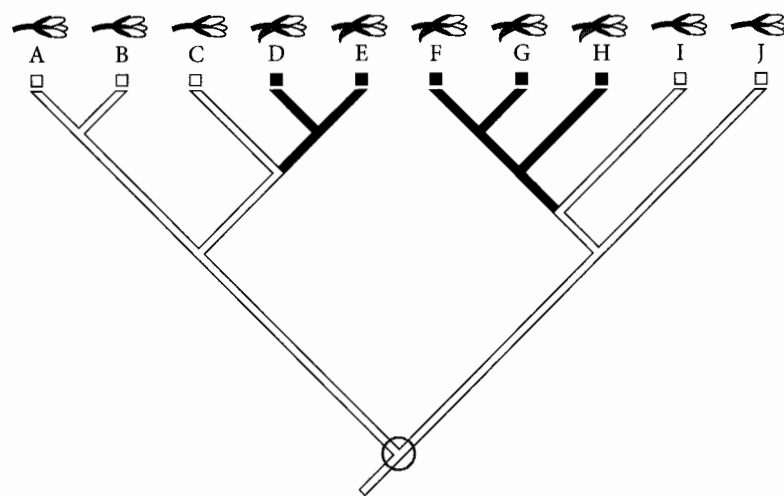


FIGURE 4.7 An evolutionary scenario under which the spurs in species D and H are not homologous due to convergence. Branches in black ended with spurred flowers, branches in white ended with spurless flowers.

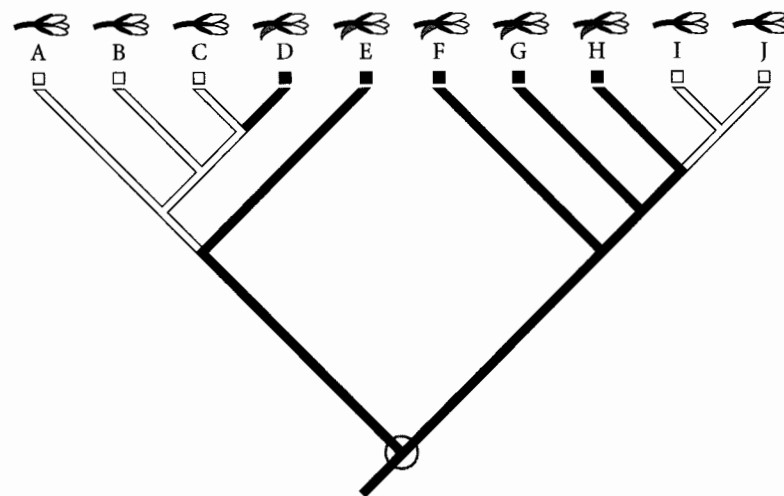


FIGURE 4.8 An evolutionary scenario under which the spurs in species D and H are not homologous due to reversal. Branches in black ended with spurred flowers, branches in white ended with spurless flowers.

evolved again. This pattern is sometimes called a *reversal*. Because the spurs in D and H trace to two different evolutionary origins, they are not homologous.

## HOMOPLASY AND CONSISTENCY

**Homoplasy** applies whenever character states are found to arise more than once on a given tree. Convergence, as shown in Figure 4.7, is a kind of homoplasy in which a derived trait arises multiple times on the same tree. Similarly, reversal, as shown in Figure 4.8, is a kind of homoplasy.

Homoplasy is sometimes presented as the opposite of homology, although this is inaccurate. Homology refers to a relationship between traits in different taxa, whereas homoplasy refers to the relationship between trait variation and a specific tree. While spur evolution in Figures 4.7 and 4.8 is homoplastic, in both cases the spurs in species F and G are homologous.

The opposite of homoplasy is not homology but **consistency**. A character is said to be **consistent** with a tree if it evolved on that tree without any extra changes of character state. A character with two states (e.g., spur present vs. spur absent) is consistent with any tree that can explain the trait's evolution with one change of state.

Spur presence/absence is consistent with the tree in Figure 4.6 because there is only one change in character state. This logic can be extended to traits with more character states. In this case, consistency holds if the number of trait changes is one less than the number of character states. This ensures that each derived character state arose just once. If the number of character state changes is equal to or greater than the number of character states, then the character shows some homoplasy.

The idea of consistency is illustrated with some DNA characters in Table 4.4, all of which evolved along the tree shown in Figure 4.9. In the characters on the left, the number of character state changes on the tree is exactly one less than the number of character states observed. Therefore, these characters are consistent with the tree. In contrast, the characters on the right experienced as many or more character state changes than character states, meaning that these characters show homoplasy.

A useful way to quantify the consistency of a character with a tree is by using the consistency index, or CI. This index reports the minimum number of changes needed to explain a trait's evolution ( $L_{\min}$ ), which is one less than

the number of character states, divided by the actual number of character state changes ( $L_{obs}$ ). This is summarized in the equation  $CI = L_{min} / L_{obs}$ . For example, if the minimum number of changes to explain a character is 3 but the observed number is 4, then  $CI = \frac{3}{4} = 0.75$ . Table 4.4 provides the CI for each trait.

The CI is not the only measure of fit between character state variation and trees. You may encounter the homoplasy index, HI, which is equal to one minus the CI:  $HI = 1 - CI$ . The retention index, RI, is also widely used. This is a more complex index that corrects for the maximum number of steps that a character could have on any tree ( $L_{max}$ ). Specifically,  $RI = (L_{max} - L_{obs}) / (L_{max} - L_{min})$ . The

TABLE 4.4 Examples of DNA sequence characters that are either consistent or homoplastic (= homoplasious), given a particular tree (Figure 4.9).

Taxa	Consistent					Homoplastic				
A	A	G	T	G	G	G	C	G	A	T
B	A	G	T	G	G	G	T	T	C	G
C	A	G	T	G	C	A	T	G	G	A
D	A	G	T	T	T	A	C	T	C	T
E	A	A	C	T	A	G	C	A	A	C
F	A	G	C	C	A	G	T	A	G	G
States	1	2	2	3	4	2	2	3	3	4
Changes	0	1	1	2	3	2	3	3	4	4
CI	1.0	1.0	1.0	1.0	1.0	0.5	0.33	0.66	0.5	0.75

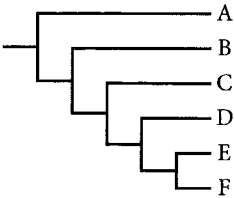


FIGURE 4.9 The tree assessed for consistency in Table 4.4.

RI is sometimes favored because, unlike the CI, it uses the full range from 0 to 1 (the minimum value of the CI varies, but is always well above 0.0).

The consistency index and related indices provide convenient measures of the fit between a character and a tree. The higher the CI or RI, the less homoplasy is implied. One application, introduced in Chapter 2, is to see if the amount of treelike structure in a complete data set is more than would be expected if the characters had not evolved up a tree. This is usually done by first finding the tree with the highest average CI for a data set (the *most parsimonious tree*; see Chapter 7). Next, this CI is compared to the CI we would expect under the scenario that the data lack any treelike structure. Chapter 9 introduces one way to conduct this analysis, the so-called permutation tail probability (PTP) test.

PARSIMONY AS A WAY TO INFER THE HISTORY OF TRAITS

So far we have discussed trait evolution as it happens in theory. We have described how genetic changes in populations yield phenotypic variation and how phenotypes evolve over time such that some traits come to characterize clades of the tree of life. However, even when we are confident in the tree, we do not have direct knowledge of the evolutionary history of traits. Rather, when we encounter statements about the evolutionary history of a character, we are dealing with *inferences*—informed conclusions guided by observational evidence.

In this section we provide a brief introduction to the use of the maximum parsimony criterion for making inferences about trait evolution. While there are more sophisticated methods now available (Chapter 10), parsimony is intuitive and provides an easy-to-understand introduction to the principles by which trait evolution can be reconstructed given some information about phylogenetic history.

As an example, let us look at the trait “wings” in insects. Figure 4.10 depicts the likely relationship among the winged insects and their closest relatives. Wings occur in adult damselflies, dragonflies, mayflies, and in the Neoptera, the large clade that includes such familiar insects as beetles, flies, bees, crickets, and butterflies. While a few neopteran lineages, such as fleas and lice, later lost wings, the ancestral condition at the base of Neoptera can be assumed to be wings present. Given this tree and the presence of wings in damselflies, dragonflies, mayflies, and Neoptera, what is the evolutionary history of wings?

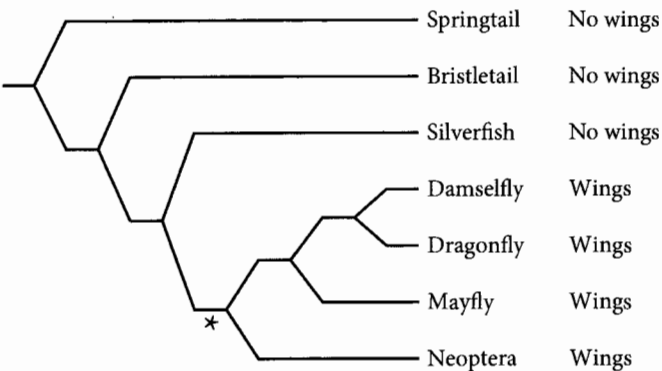


FIGURE 4.10 Phylogeny of Neoptera and related insects. Presence/absence of wings is indicated for each lineage. Based on parsimony, the most plausible hypothesis is that wings evolved once, on the branch marked with an asterisk.

There are many possible histories that could explain the distribution of wings. There could have been an ancient origin of wings at the base of the tree, followed by losses in springtails, bristletails, and silverfish; or there could have been four origins of wings, once each in dragonflies, damselflies, mayflies, and other insects; or there could have been a single origin of wings just before the last common ancestor of dragonflies, damselflies, mayflies, and Neoptera; and so on. All of these are valid explanations in that each tip has a state that is a summation over its evolutionary history. But only one of these alternatives actually happened. How can we decide which explanation is the best working hypothesis?

A simple way to pick the most plausible history is to apply the *principle of parsimony* (we will talk more about this concept in Chapter 7). The logic of parsimony may be familiar under the label Occam’s (or Ockham’s) razor, named after the fourteenth-century logician and friar William of Ockham (ca. 1288–1348), who said, “pluralitas non est ponenda sine necessitate” (plurality should not be posited without necessity). This principle holds that the simplest explanation of a phenomenon is the most likely to be true.

Suppose you are sitting at a police switchboard in a North American city and within a span of five minutes you receive two calls reporting a tiger on the loose. Theoretically this could indicate the existence of two tigers roaming the

streets. But would you really suspect this? No. More likely you would assume that there is just one tiger and two independent witnesses. The reason this is the logical conclusion is that calls reporting tigers are very rare. As a result, it is more likely that the two calls represent two manifestations of the same rare event rather than two independent, rare events.

We can use the same principle to make an inference about the evolution of wings. Gaining or losing wings during evolution is rare. Therefore, the most plausible explanation of the observed pattern is the one that requires the fewest evolutionary origins (or losses) of wings. Given the tree shown in Figure 4.10, the most parsimonious scenario is that wings evolved once on the branch marked with an asterisk. Thus, it is most parsimonious to assume that the wings of dragonflies, mayflies, damselflies, and Neoptera are homologous and that their sister group, silverfish, does not have any winged ancestors.

A single origin of wings is the most parsimonious explanation because it can explain the distribution of wings among the tips with only a single change in character state. This reconstruction is the one that maximizes the consistency index. In contrast all other scenarios require additional gains and/or losses of wings and are associated with more homoplasy and lower consistency indices.

Figure 4.11 considers four other scenarios for the evolution of wings. Gains of wings are marked with a plus sign (+) and losses of wings are marked with a minus sign (–). The cases to the right involve four changes of state (four gains or one gain and three losses), whereas the cases to the left involve two changes of state (two gains or one gain and one loss). Because these scenarios involve more than one change of character state, they are, by definition, less parsimonious than the hypothesis of a single origin of wings. Extending this logic, it is also fair to say that the two scenarios on the right are less likely than the two on the left because they require the occurrence of even more rare events (and have even lower CIs).

In the case of wing evolution in insects, there is a single reconstruction of character history that is more parsimonious than any other. However, sometimes two or more alternative reconstructions of character history are equally parsimonious: each involves the same number of evolutionary events. For example, consider the evolution of echolocation, an ability to “see” by emitting sounds and listening for echoes, in bats.

Echolocation is well developed in the narrowly defined Microchiroptera (“microbats”) as well as the horseshoe bats and their relatives, but is absent

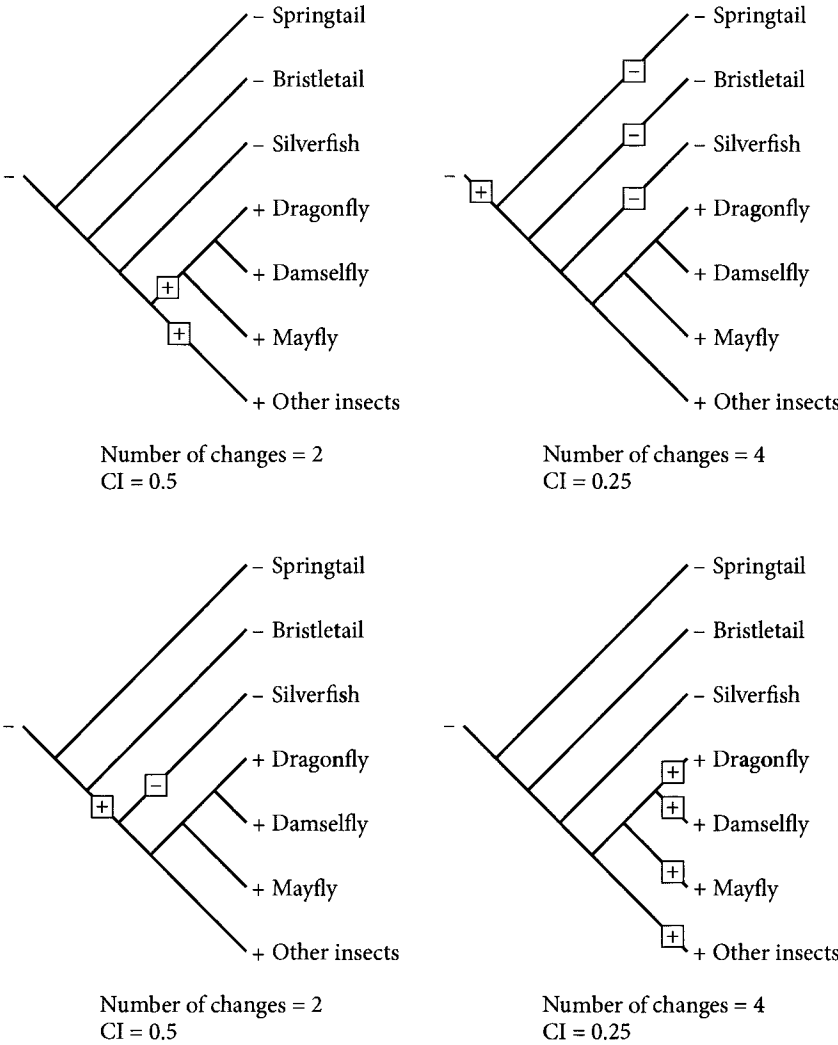


FIGURE 4.11 Four of the possible scenarios for the evolution of wings in insects. The presence of wings in the tips is indicated “+” and their absence by “-.” Gains and losses of wings under each scenario are indicated using the same convention. The number of changes and the consistency index are indicated for each scenario.

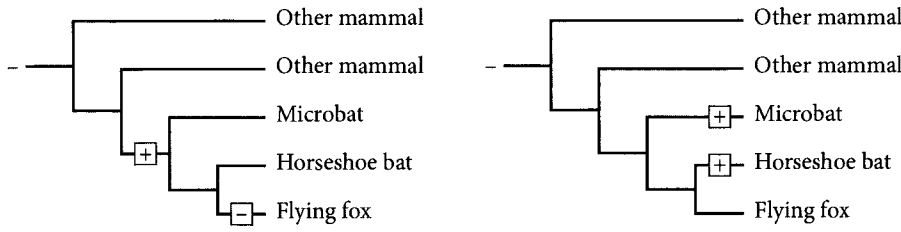


FIGURE 4.12 Two equally parsimonious scenarios for the evolution of echolocation in bats. Gains of echolocation are indicated with “+” and losses with “-.”

from the flying fox (fruit bat) group. Given the phylogeny shown in Figure 4.12 (from Teeling et al. 2005), there is no way to explain the evolution of echolocation with fewer than two evolutionary state changes. But there are two equally parsimonious explanations. There could have been a single origin of echolocation close to the origin of bats, followed by a loss of this ability in the flying foxes. Or there could have been separate origins of echolocation in “microbats” and horseshoe bats. The principle of parsimony in its most basic form would not allow us to favor one or the other of these two explanations.

With further information about echolocation in different kinds of bats you might develop a preference for one hypothesis or the other. For example, if you concluded that independent gains of echolocation are highly improbable, whereas secondarily losing echolocation is more likely, you might favor the evolutionary scenario on the left. But if all you go on is simple parsimony, and if you assume that this tree is correct, both scenarios are equally plausible.

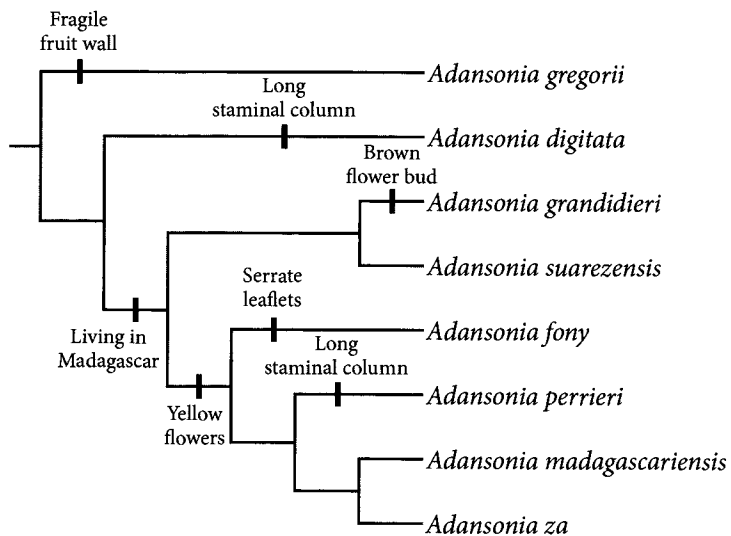
In this chapter we have explored the evolution of traits along the branches of phylogenetic trees. It should now be clear that by using parsimony (and more sophisticated methods to be introduced in Chapter 10) we can combine trees with knowledge of the distribution of character states among tips to learn about the history of trait evolution. Equally importantly, knowledge of trait evolution forms the basis of phylogenetic inference. By understanding the evolution of morphological and molecular characters, we are able to develop rigorous methods to reconstruct phylogenetic trees that are reasonable hypotheses of evolutionary history. We delve into these methods starting in Chapter 7.

FURTHER READING

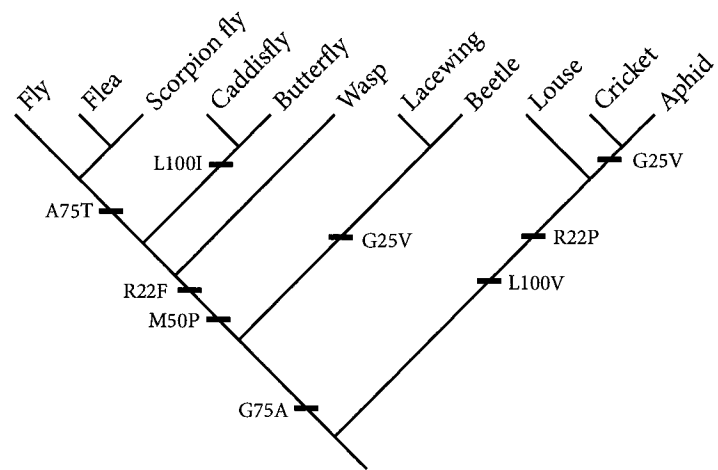
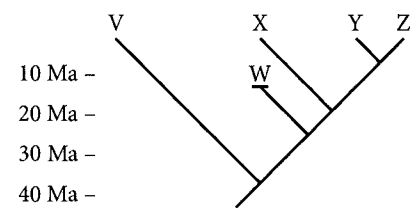
Trait evolution: Maddison and Maddison 2000  
Molecular evolution: Page and Holmes 1998; Graur and Li 2000  
Character polarity: Hennig 1966  
Homology (conceptual issues): Patterson 1982; Wagner 1989; Abouheif 1997; Mindell and Meyer 2001; Hall 2003; Scotland 2010  
Homoplasy and consistency: Farris 1989; Kitching et al. 1998; Wake et al. 2011  
Parsimony: Sober 1983, 1991

CHAPTER 4 QUIZ

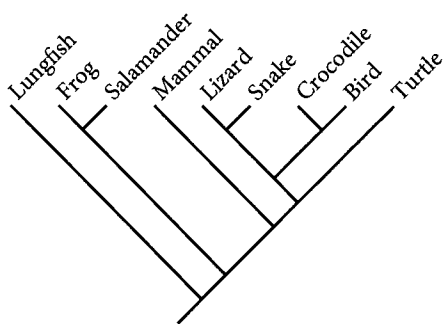
- On a phylogenetic tree, where are character state changes typically marked?  
a. at the root    b. at nodes    c. along branches    d. at the tips  
e. character state changes are not shown on trees
- Consider a plausible phylogeny of the baobab trees, genus *Adansonia*. On the basis of this tree, which species lives in Madagascar, has woody (not fragile) fruit walls, white (not yellow) flowers, and green (not brown) flower buds?  
a. *A. gregorii*    b. *A. digitata*    c. *A. grandidieri*    d. *A. suarezensis*  
e. *A. fony*



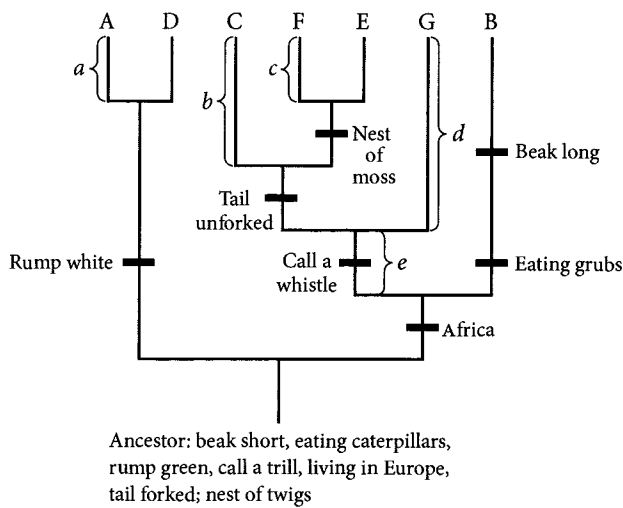
- Which statement is best supported by the chronogram?  
a. Traits occurring in species W will be more ancient than those occurring in species Z  
b. Traits occurring in species Z will tend to be more advanced than those occurring in species Y  
c. Traits in species X will tend to be more similar to the traits of species W than to the traits of species Z  
d. Traits occurring in species W and X will tend also to occur in Z  
e. V has the most plesiomorphic character states
- Consider a plausible phylogeny for some of the major insect groups. Along the branches are marked changes in a hypothetical protein sequence. The marks list the amino acid position in the protein that changed, flanked by the standard one-letter codes for the amino acids before the change (to the left of the number) and after the change (to the right of the number). For example, L122S means that position 122 changed from a leucine (L) to a serine (S). Based on this tree, what amino acids does a butterfly have at positions 50, 75, and 100?



5. Consider the following three facts: (1) the tree shown is correct; (2) The amnion (a membrane that surrounds the embryo) evolved once and was never lost; (3) A snake and a turtle have an amnion. Which of the following *must* also be correct?
- a. Salamanders have amnions
  - b. Birds have amnions
  - c. Mammals have amnions
  - d. Lungfish do not have amnions
  - e. Mammals do not have amnions

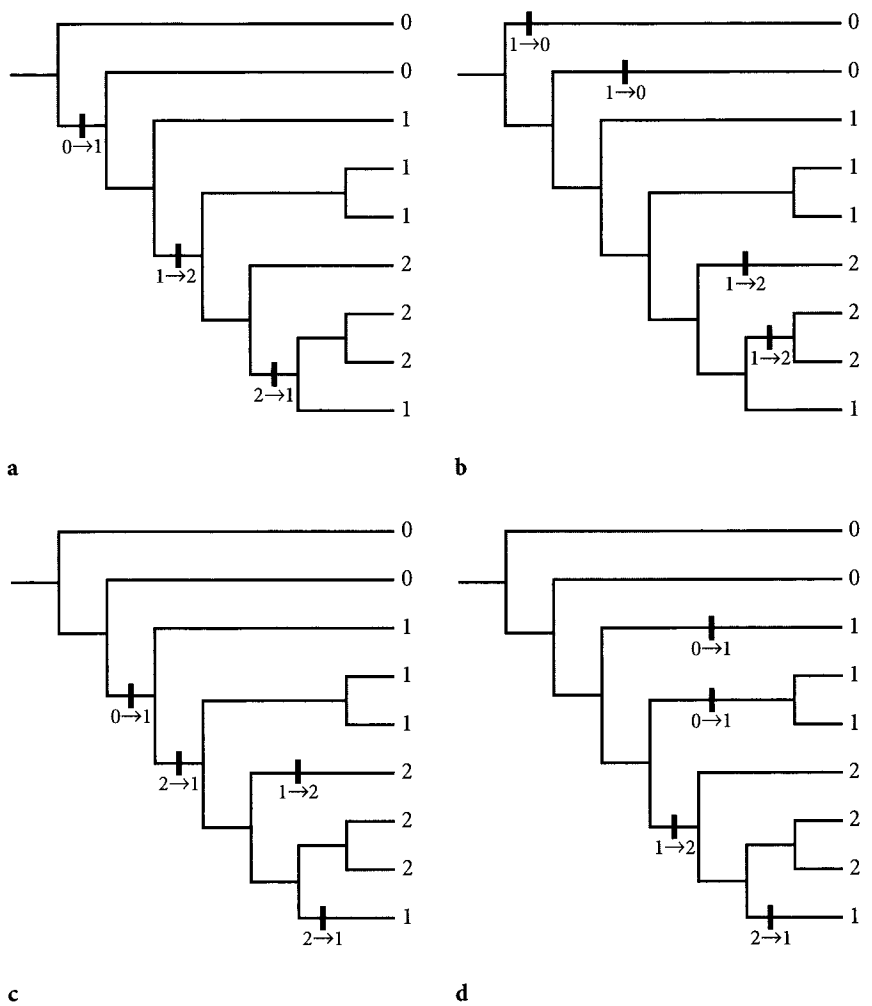


Questions 6–7 refer to the tree for a hypothetical group of birds.

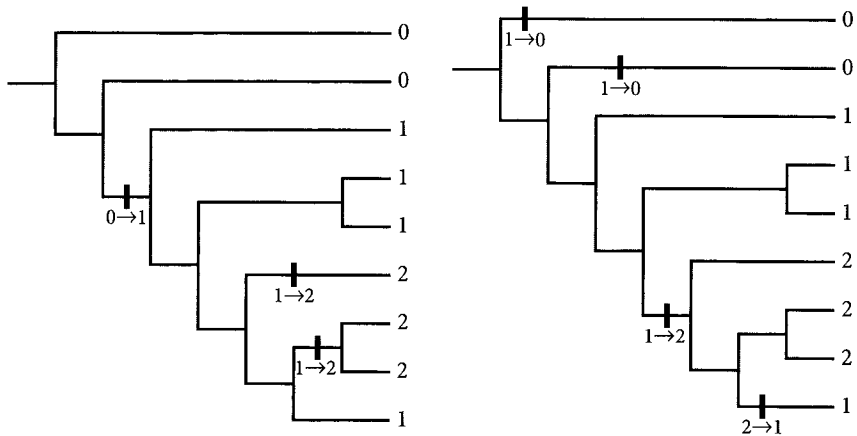


6. Which of the following sets of traits do you expect to find in bird E?
- a. Rump white, Africa, call a whistle, tail unforked, beak long
  - b. Eating caterpillars, rump green, Europe, nest of moss
  - c. Nest of moss, forked tail, rump white, Africa
  - d. Beak short, eating caterpillars, rump green, tail unforked
7. Which of the labeled branches (a, b, c, d, or e) contain at least some individuals with the following combination of traits: eating caterpillars, rump green, Africa, and call a trill?

8. The four trees each depict a history of trait evolution for a character with three characters states, 0, 1, and 2. The character states at the tips are shown, as well as each transition. Which of these four provides a complete and plausible history of trait evolution?



9. The four scenarios for trait evolution all validly explain the observed trait variation on this tree. Which is most parsimonious?



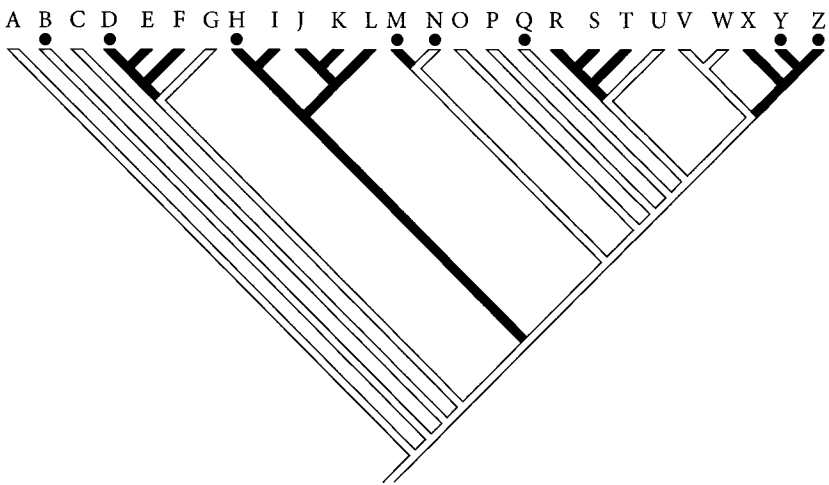
**a**

**b**

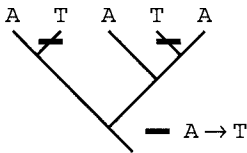
C

**d**

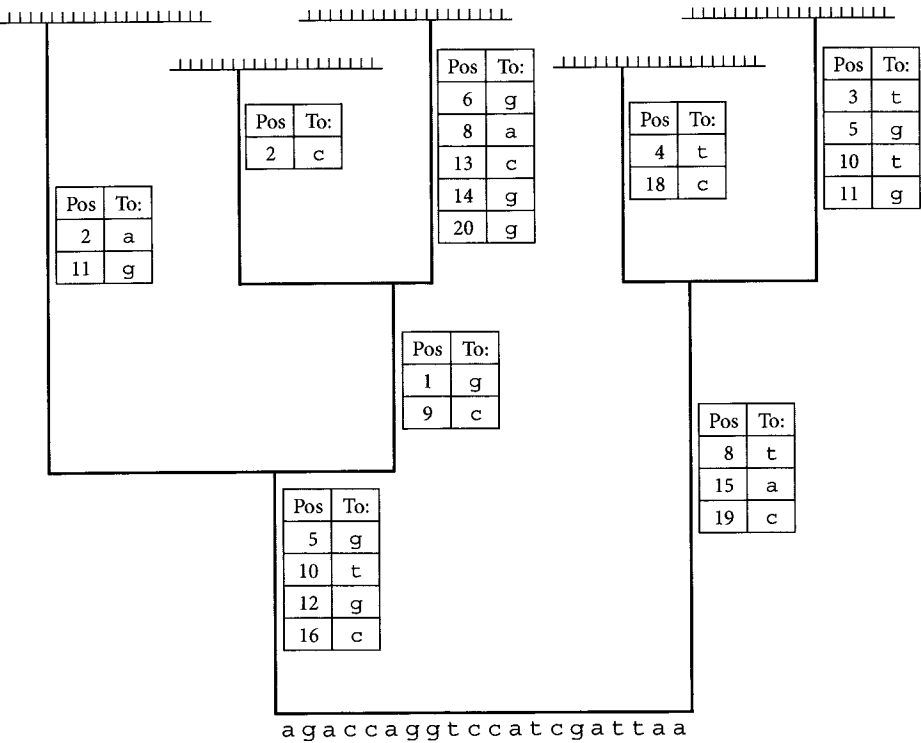
Questions 10–11. In the tree, white branches depict lineages that live in Africa, whereas the others live in South America. The taxa marked with a black circle have pink flowers, whereas the rest have yellow flowers.



10. Using parsimony, how many times did pink flowers evolve?  
a. 1    b. 3    c. 4    d. 6    e. 8
11. How many of those origins of pink flowers occurred in populations that were living in Africa?  
a. 0    b. 1    c. 3    d. 4    e. 6
12. This tree shows the state of each tip for a particular position in a DNA molecule. What is the consistency index for this position?  
a. 0.2    b. 0.25    c. 0.4    d. 0.5    e. 2



13. The tree shows an ancestral sequence of 20 nucleotides. Along the branches the changes are shown. Deduce the sequences of the five living species.



14. The tree shown for Questions 6–7 has one branch with two trait changes shown, beak long and eating grubs. Why should one be careful about assuming, on the basis of this diagram, that the diet trait changed before the beak trait? What additional information might support the conclusion that indeed beaks lengthened after the transition to grub eating?
15. Trait evolution is often represented with a line drawn across a branch of a phylogenetic tree. Describe the evolutionary phenomena that are represented by this line.
16. Dogs have a tiny appendage above their paw, the so-called dewclaw. If we had a time machine, how could we determine definitively whether this body part is homologous to the human thumb? Assume that if we look at a parent and its offspring we can unambiguously identify the structures in each that are homologous.

# Relatedness and Taxonomy

Following the acceptance of evolutionary theory in the nineteenth century, *taxonomy*, the science of establishing and using biological classifications, acquired a clear mission to represent evolutionary relationships. In so doing taxonomy became a branch of *systematics*, the study of the evolution of biological diversity. Nonetheless, it took another hundred years for all of the implications of that merger to become clear. First, taxonomists had to clarify that evolutionary relatedness should be defined in terms of common ancestry. Second, because common ancestry is captured in phylogenetic trees, taxonomists had to adapt to the idea that their classification systems should mirror the tree of life. That is to say, classification should follow the rule that the more recently two organisms last shared a common ancestor, the more closely they should be classified. And third, systematics needed to develop the tools to build reliable trees. In short, it took biologists over a hundred years to recognize that classification, relatedness, and trees are really three sides of the same coin, so to speak.

In this chapter, we clarify the concept of relatedness and show that it applies equally to family trees (pedigrees) and phylogenetic trees. Then we discuss the common confusion between relatedness and similarity and explain why modern taxonomy focuses on phylogenetic relatedness rather than similarity. We explore *nomenclature*, the rules regulating the names of taxa, and show why the concept of taxonomic rank is not meaningful in a phylogenetic context. We end the chapter with a brief discussion of phylogenetic nomenclature, a new, but controversial, approach that attaches names to clades rather than to ranked taxa.

## THE CONCEPT OF RELATEDNESS

The concept of relatedness as applied to the branches of a phylogenetic tree mirrors that used in discussions of human familial relationships. In both cases, the