

Chapter 3: Evolutionary mechanisms and the diversity of life

In which we consider the rather exuberant diversity of organisms and introduce the primary evolutionary mechanisms responsible for it.

In medieval Europe there was a tradition of books known as bestiaries. These were illustrated catalogs of known and imagined organisms in which it was common for particular organisms to be associated with moral lessons. "Male lions were seen as worthy reflections of God the Father, for example, while the dragon was understood as a representative of Satan on earth."⁴⁰ One can see these books as an early version of a natural theology, that is, an attempt to gain an understanding of the supernatural through lessons from and studies of natural objects. In this case, the presumption was that each type of organism was created for a particular purpose, and that often this purpose was to provide people with a moral lesson. This way of thinking grew more and more problematic as more and more different types of organisms were recognized, many of which had no obvious significance to humans. Currently, scientists have identified approximately 1,500,000 different species of plants, animals, and microbes. The actual number of different types of organisms, referred to as species, may be as high as 10,000,000.⁴¹ These numbers refer, of course, to the species that currently exist, but we know from the fossil record that many distinct species, which are now extinct, have existed in the past. So the obvious question is, why are there so many different types of organisms?⁴² Do they represent multiple independent creation events, and if so, how many such events have occurred?



Catalogued and predicted species

doi:10.1371/journal.pbio.1001127.t002

Species	Earth	Ocean
	Catalogued	Catalogued
Eukaryotes		
Animalia	953,434	171,082
Chromista	13,033	4,859
Fungi	43,271	1,097
Plantae	215,644	8,600
Protozoa	8,118	8,118
Total	1,233,500	193,756
Prokaryotes		
Archaea	502	1
Bacteria	10,358	652
Total	10,860	653
Grand Total	1,244,360	194,409
Predicted	8,750,000	2,210,000

As the true diversity of organisms was discovered, a number of observations served to undermine the early concept that organisms were created to serve humanity. The first were the number of organisms that had very little obvious importance to the human condition. This was particularly obvious in the case of extinct organisms but extended further as a result of newly discovered organisms. At the same time students of nature, known generically as naturalists, discovered many different types of upsetting and cruel behaviors within the natural world. Consider the fungus *Ophiocordyceps unilateralis*, which infects the ant *Camponotus leonardi*. The fungus takes control of the ant's behavior, causing them to migrate to positions that favor fungal growth before killing the infected ant. Similarly, the nematode worm *Myrmeconema neotropicum* infects the ant *Cephalotes*

⁴⁰ <http://www.getty.edu/art/gettyguide/artObjectDetails?artobj=304109>

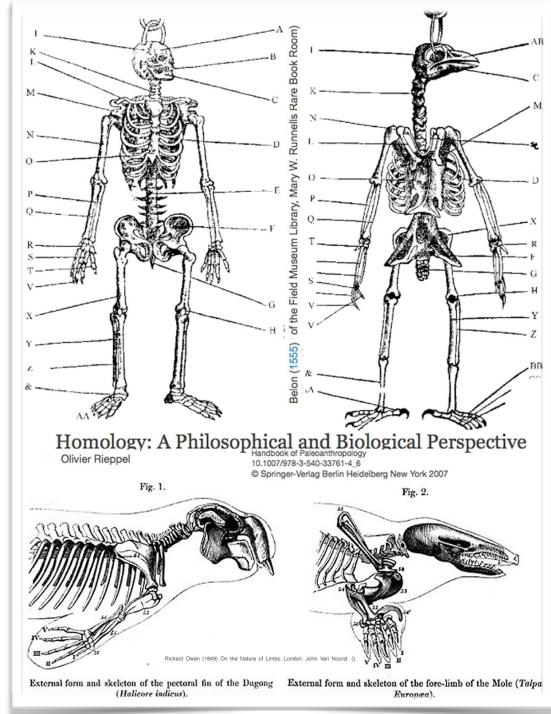
⁴¹ How many species are there on Earth and in the ocean? <http://www.plosbiology.org/article/info%3Adoi%2F10.1371%2Fjournal.pbio.1001127>

⁴² As a technical point, which we will return to, we will refer to each distinct type of organism as a species.

atratus. This leads to dramatic changes in the morphology and behavior of the ant. The ant's abdomen turns red and is held up-raised, which makes the infected ant resemble a fruit and so increases the likelihood of it being eaten by birds. The birds transport the worms, which survive in their digestive systems until they are excreted and subsequently are eaten by ants to complete the worm's life cycle.⁴³ Perhaps the most famous example of this type of behavior are the wasps of the family *Ichneumonidae*. Female wasps deposit their fertilized eggs into the bodies of various types of caterpillars, where the eggs hatch out and produce larvae that feed on the caterpillar, keeping it alive while they eat it from the inside out. Charles Darwin remarked in a letter to Asa Gray, an American naturalist, "There seems to me too much misery in the world. I cannot persuade myself that a beneficent & omnipotent God would have designedly created the *Ichneumonidae* with the express intention of their feeding within the living bodies of caterpillars, or that a cat should play with mice." Rather than presume that a supernatural creator was responsible for such gratuitously (or at least apparently) cruel behaviors, Darwin and others sought alternative, morally neutral naturalistic processes that could generate biological diversity and explain biological behaviors.



As the diversity of organisms became increasingly apparent and difficult to ignore, another broad and inescapable conclusion began to emerge from anatomical studies of organisms, many different organisms displayed remarkable structural similarities. For example, as naturalists characterized various types of animals, they found that they either had an internal skeleton (the vertebrates) or did not (the invertebrates). Comparative studies among the vertebrates revealed that there were often striking similarities between quite different types of organisms. A classic work, published in 1555, compared the skeletons of a human and a bird.⁴⁴ While many bones have changed shape and relative sizes, what was most striking is how many bones are at least superficially similar between the two. This same type of "comparative anatomy" revealed many similarities between disparate organisms. For example, the skeleton of the dugong (a large aquatic mammal) appears quite similar to that of the European mole, which tunnels underground on land. In fact, there are general skeletal similarities between all vertebrates. The closer we look, the more similarities we find. These similarities run deeper than the anatomical, they extend to the cellular and the molecular as well. So the scientific question is, what explains such similarities? Why build an organism that walks, runs, and climbs, such



⁴³ The Life of a Dead Ant: The Expression of an Adaptive Extended Phenotype: <http://www.jstor.org/stable/10.1086/603640>

⁴⁴ Belon P (1555) L'Histoire de la Nature des Oiseaux. Paris, Guillaume Cavellat

as humans, with a skeleton similar to that of a organism that flies (birds), swims (dugongs), or tunnels (moles). Are these anatomical similarities just flukes or do they imply something deeper?

Organizing organisms (hierarchically)

Carl Linnaeus (1707-1778) was the pioneer in taking the similarities between different types of organisms seriously. Based on similarities (and differences), he developed a system to classify organisms in a coherent and hierarchical manner. Each organism had a unique place in this scheme. What was, and occasionally still is, the controversial aspect of such a classification system is in deciding which traits should be considered significant and which are superficial or unimportant, at least for the purposes of classification. Linnaeus had no real theory to explain why organisms could be classified in such a hierarchical manner and could only go on observations. This might be a good place to reconsider the importance of hypotheses, models, and theories in biology. Linnaeus noticed the apparent similarities between organisms and used it to generate his classification scheme, but he had no explanatory model for why such similarities should exist (very much like Newton's law of gravitation did not explain why there was gravity). So what are the features of an explanatory model? Such a model has to go beyond just explaining, it also has to suggest observations or predict outcomes that have not yet been observed. It is these validity of these predictions that enable us to distinguish between different explanatory models. A model that makes no validated predictions is not particularly useful. A model that makes explicit predictions, even if they prove to be wrong, enables us to refine our model or force us to abandon the model and develop a new one. A model that, through its various hypotheses and their confirmation or refutation or revision, has been found to accurately explain a particular phenomena can become promoted to a theory. So this enables us to distinguish between a law and a theory. A law describes what we see but not why we see it. A theory provides the explanation for observable phenomena.⁴⁵

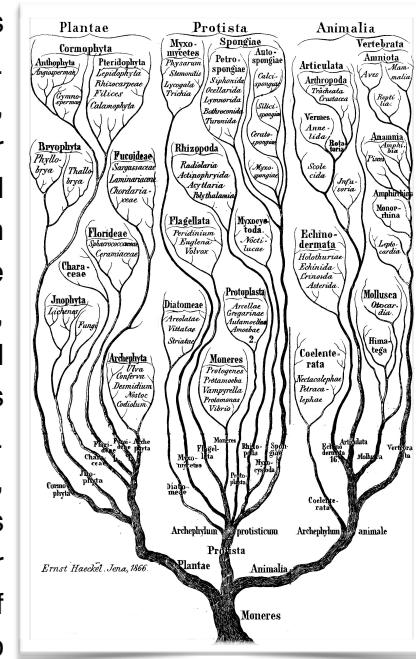
Back to Linnaeus, whose classification system placed organisms of a particular type were placed together into a species. Of course, what originally counted as a discrete type of organism was based on Linnaeus's judgement as an observer and classifier; what particularly traits he felt defined the species and distinguished it from other, similar species. The choice of these key traits was subject to debate. Based on the perceived importance and presence of particular traits, organisms could be split into two or more types (species), or two types originally considered separate species could be reclassified into a single type.

As we will see, the individual organisms that make up a species are not identical but share many traits. In organisms that reproduce sexually, there are often dramatic differences between males and females of the same species, a situation known as sexual dimorphism. In some cases, these differences can be so dramatic that without further evidence, it can be difficult to tell whether two animals are members of the same or different species. In this light the primary criteria for determining whether sexually reproducing organisms are members of the same or different species is whether they can and do successfully interbreed with one another. This criteria, that is reproductive compatibility, can be used to place species distinctions on a more empirical basis, but it cannot be used with asexual

⁴⁵ If we go back, Newton's law of gravity explained how objects behaved gravitationally, but it not why. In contrast, Einstein's theory of general relativity explained why there was gravity, and predicted behaviors that were not predicted by Newton's law.

species (such as most microbes). Within a species, there are sometimes regional differences that are distinct enough to be recognizable. Where this is the case, these groups are known as populations, races, or subspecies. While distinguishable, the organism in these groups retain the ability to interbreed and so are members of a single species.

After defining types of species, Linnaeus next grouped species that displayed similar traits into a larger group, known as a genus. While a species can be considered a natural, interbreeding population, a genus is a more artificial group. Which species are placed together within a particular genus depends on the common traits deemed important or significant by the person doing the classification. This can lead to conflicts which are generally resolved by the collection of more and more comparative data. In the Linnaean classification scheme, each organism has a unique name, which consists of its genus and species names. The accepted usage is to write out the name in italics with the genus name capitalized, for example, *Homo sapiens*. Following on this pattern, one or more genera are placed into larger, more inclusive groups, and these groups, in turn, are themselves placed in larger groups. The end result of this process is the rather surprising observation that all organisms fall into a small number of “super-groups” or phyla. We will not worry about the traditional group names, because in most cases they really do not help in our understanding of basic biology. Perhaps most surprising of all, all organisms and all phyla fall into one and only one family - all of the organisms on earth can be placed into a single unified phylogenetic “tree” or perhaps better put, bush. That this should be the case is by no means obvious. This type of analysis could have produced multiple, distinct classification schemes, but it did not.



It is worth reiterating the fact that while a species can be seen as a natural group, the higher levels of classification are based on various hypotheses, specifically that certain traits are more important or informative than others. For example, having hair, four legs, and teeth is not enough to determine unambiguously whether an organism is in the genus *Canis*, which includes wolves and coyotes, or the genus *Vulpes*, which includes foxes. This is a choice based on various lines of evidence, but nothing as distinct as whether foxes normally mate with coyotes (they do not). Because genus and more inclusive group classifications are based on arguments about the significance of various shared traits. Where scientists place a species can change. New observations can lead to the reorganization of the classification scheme, a species or a genus can be moved from one place to another, or a larger group can be divided into two or more new groups. For example consider the types of organisms commonly known as bears. There are a number of different types of bear-like organisms, a fact that Linnaeus's classification scheme explicitly acknowledged even though it never attempted to explain why. Looking at all bear-like organisms we can recognize eight types.⁴⁶ We currently consider four of these, the brown bear (*Ursus arctos*), the Asiatic black bear (*Ursus thibetanus*), the American

⁴⁶ http://en.wikipedia.org/wiki/List_of_bears

bear (*Ursus americanus*), and the polar bear (*Ursus maritimus*) to be significantly more similar to one another, based on the presence of various traits, than they are to other types of bears. We therefore placed them in their own genus, *Ursus*. We have placed each of the other bears, the spectacled bear (*Tremarctos ornatus*), the sloth bear (*Melurus ursinus*), the sun bear (*Helarctos mayalanus*), and the giant panda (*Ailuropoda melanoleuca*) in their own separate genera. Scientists consider these species more different from one another than are the members of the genus *Ursus*. That said, all of these bears clearly share a number of other traits, so we place them all in the larger group, the family Ursidae to reflect their undeniable similarities. Scientists originally considered the red panda (*Ailurus fulgens*) to be a bear, but it has now been moved into a distinct group, the Ailuridae. Both the Ursidae and the Ailuridae are part of a larger and more diverse group, the Carnivora, which includes cats, dogs, wolverines, and their relatives. A key for placing these species together is that they are all placental mammals. There are other bear-like organisms that are not bears or even members of the Carnivora group. Both the koala (*Phascolarctos cinereus*) and the extinct giant marsupial bears of the genus Proborhyaenid are marsupial mammals; their offspring are born relatively undeveloped and mature in a pouch on the mother. All marsupial mammals are more similar to one another in key ways than they are to *any* placental mammal. We consider placental and marsupial traits more significant, from a classification perspective, than the bear-like traits these organisms share. That said, both true (placental) bears and marsupial bears are placed in the larger group known as Mammalia, which includes monotreme (egg-laying), marsupial, and placental mammals. We group mammals together in part because they feed their young using a common substance, milk, secreted by the mammary glands of their mothers. We place mammals together with reptiles, birds, and fish into an even larger group known as the Chordates based on the presence of an internal skeleton and more specifically a backbone, and from there into larger and even more inclusive groups.

What is most significant for our purposes is *not* the particular place that an organism occupies within the classification system but rather the fact that we can place all organisms in a logical and self-consistent manner within such a system. As we will discover later on, the use of gene (DNA) sequencing methods has provided further support for this classification scheme, removing ambiguities and supporting its underlying logic. As we gather more and more data, we find that Linnaeus was correct. These is an unambiguous hierarchical relationship between organisms.

Fossils and the Linnaean system

As mentioned previously, we continue to discover new fossils and new organisms.⁴⁷ In most cases, these fossils appear to represent organisms that lived many millions to hundreds of millions of years ago but which are now extinct. Clearly there are dramatic differences between the ability of different types of organisms to become fossilized. Perhaps the easiest to fossilize are those organisms with internal or external skeletons, yet it is estimated that between 85 to 97% of such organisms are not represented in the fossil record and various studies indicate that many other types of organisms have left no fossils whatsoever.⁴⁸ Some authors have estimated that the number of organisms at the genus

⁴⁷ Your inner fish: <http://www.pbs.org/your-inner-fish/home/>

⁴⁸ The incompleteness of the fossil record: <http://www.donaldprothero.com/files/47440594.pdf>

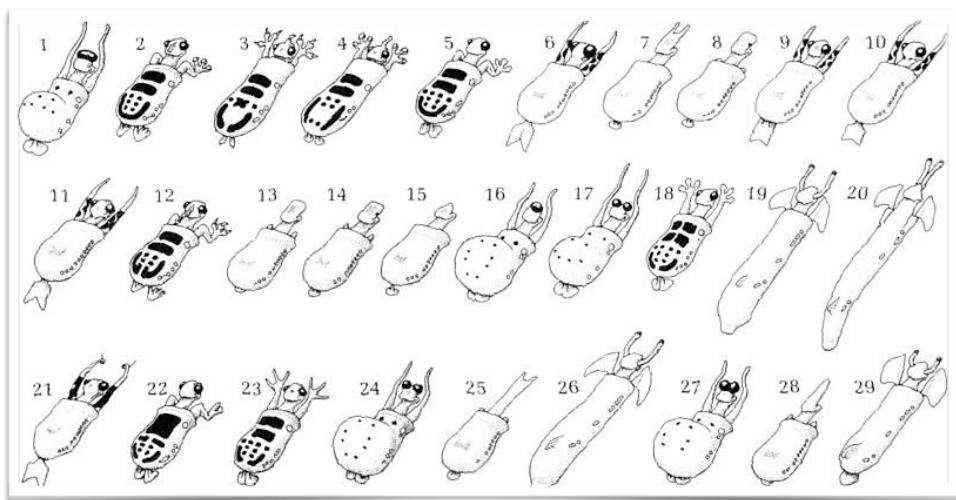
level that have been preserved as fossils may be less (often much less) than 5%.⁴⁹ For some categories of organisms, such as the wide range of microbes, essentially no informative fossils exist.

Once scientists recognized that fossils provide evidence for extinct organisms, the obvious question was do extinct organisms fit into the Linnaean classification scheme or do they form their own groups or their own separate trees? This can be a difficult question to answer, since many fossils are only fragments of the intact organism. The fragmentary nature of the fossil record can lead to ambiguities. Nevertheless, the conclusion that has emerged upon careful characterization is that we can place almost all fossilized organisms within the modern Linnaean classification scheme. There are possible exception, like the Ediacaran organisms that lived very long ago and appear structurally distinct from known living organisms. The presumption, however, is that if we had samples of these organisms for molecular analyses, we would find it that they too would fall nicely into the same classification scheme as all other organisms do.⁵⁰ For example, dinosaurs, along with modern birds, are clearly descended from a specific type of reptile, while living mammals are more closely related to a second, now extinct group, known as the “mammal-like reptiles.”

In rare cases, particularly relevant to human evolution, one trait that can be recovered from bones is DNA sequence data. For example, it has been possible to extract and analyze DNA from the bones of Neanderthals and Denisovian-type humanoid organisms, that went extinct about 30,000 years ago, and to use that information to clarify their relationship to modern humans (*Homo sapiens*).⁵¹ This type of data provides evidence for interbreeding and has led to the argument for the reclassification of Neanderthals and Denisovians as subspecies of *Homo sapiens*.

Questions to answer and ponder:

- Explain why you might expect that extinct species fit into the Linnaean classification scheme.
- What would make you decide that a particular trait was important or unimportant (secondary) from a classification perspective?
- Given the following imaginary animals →, place them in a plausible classification system and explain your reasoning.
- How could Neanderthals be a distinct species if evidence for in-breeding with *H. sapiens* exists?



⁴⁹ Absolute measures of the completeness of the fossil record: <http://www.ncbi.nlm.nih.gov/pubmed/11536900>

⁵⁰ On the eve of animal radiation: phylogeny, ecology and evolution of the Ediacara biota: <http://users.unimi.it/paleomag/geo2/Xiao&Leflammé2008.pdf>

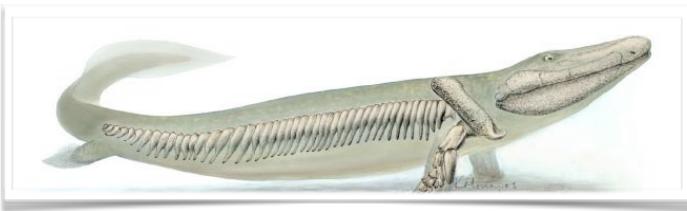
⁵¹ Paleogenomics of archaic hominins: <http://www.ncbi.nlm.nih.gov/pubmed/22192823>

The theory of evolution and the organization of life

Perhaps surprisingly, Linnaeus never proposed a plausible (or even an implausible) naturalistic explanation for why organisms should be classifiable in a hierarchical way. Why is it that birds, whales, and humans share common features, such as the organization of their skeletons, that led Linnaeus to classify them together as vertebrates? Why are there extinct organisms, known from their fossils, that share these common features, even though they are otherwise quite different? We had to wait about 100 years for a plausible model that explained why the Linnaean classification scheme actually works and can be used it to make predictions about organisms that no longer exist. Charles Darwin (1809–1882) and Alfred Wallace (1823–1913) proposed such a model, described in great detail in Darwin's book *The Theory of Evolution by Natural Selection*, originally published in 1858.

As we will see, evolutionary theory is based on a series of direct observations of the natural world and their logical implications. Evolutionary theory explains why similar organisms share similar traits and why we can place them easily into a hierarchical classification system. They are similar because they are related to one another – they share common ancestors. Moreover, we can infer that

the more different two organisms are, the longer ago this common ancestor lived. We can even begin to make plausible and empirically-supportable deductions about what those common ancestors looked like. As an example, we can predict that the common ancestor of all terrestrial vertebrates will resemble a fish with leg-like limbs. Scientists have recently discovered fossils of such an organism, *Tiktaalik*.⁵² This is



Tiktaalik roseae, an extinct fish-like organism that lived ~ 375 million years ago, is likely to be similar to the common ancestor of all terrestrial vertebrates.

just one more example of the fact that since its original introduction, and well before the mechanisms of heredity and any understanding of the molecular nature of organisms were resolved, evolutionary theory explained what was observed and made testable predictions about what would be found.

So what are the facts and inferences upon which the Theory of Evolution is based? Two foundational observations are deeply interrelated and based on empirical observations associated with plant and animal breeding and the observed behaviors of natural populations. The first is the fact that whatever type of organism we examine, if we look carefully enough, making accurate measurements of visible and behavioral traits (this description of the organism is known as its **phenotype**, we find that individuals vary with respect to one another. More to the point, plant and animal breeders recognized that the offspring of a controlled mating between individuals often had phenotypes similar to those of their parents. Certain phenotypic traits can be inherited. Over many generations, domestic animal and plant breeders used what is now known as artificial selection to generate the range of domesticated plants and animals with highly exaggerated phenotypes that we now rely on (see picture on next page). For example, beginning about 10,000 years ago plant breeders in Mesoamerica developed modern

The main unifying idea in biology is Darwin's theory of evolution through natural selection.

– John Maynard Smith

⁵² Meet *Tiktaalik roseae*: An Extraordinary Fossil Fish: <http://tiktaalik.uchicago.edu/meetTik.html>

corn (maize) by the selective breeding of variants of the grass teosinte.⁵³ All of the various breeds of dogs, from the tiny to the rather gigantic, appear to be derived from a common ancestor that lived between 19,000 to 32,000 years ago (although as always, be skeptical; it could be that exactly where and when this common ancestor lived could be revised).⁵⁴ In all cases, the crafting of specific domesticated organisms followed the same pattern. Organisms with desirable traits (phenotypes) were selected for breeding with one another. Organisms that did not have these traits were discarded and not permitted to breed. This process, carried out over hundreds to thousands of generations, led to organisms that displayed distinct or exaggerated forms of the selected trait. What is crucial to understand is that this strategy could work only if different versions of the trait were present in the original selected population and at least a part of this phenotypic variation was due to genetic, that is inheritable, factors. What these inheritable factors were was completely unclear, but we can refer to it as the organism's **genotype** (even though plant and animal breeders would never have used that term).

This implies that different organisms have different genotypes, but where those differences come from was completely unclear to early plant and animal breeders. Were they imprinted on the organism in some way based on its experiences or induced by environmental factors? Was the genotype stable or could it be modified by experience? How were genotypic factors passed from generation to generation? And how, exactly, did a particular genotype produce or influence a specific phenotypic trait. As we will see, at least superficially, this last question remains poorly resolved for many phenotypes.

So what do we mean by genetic factors?

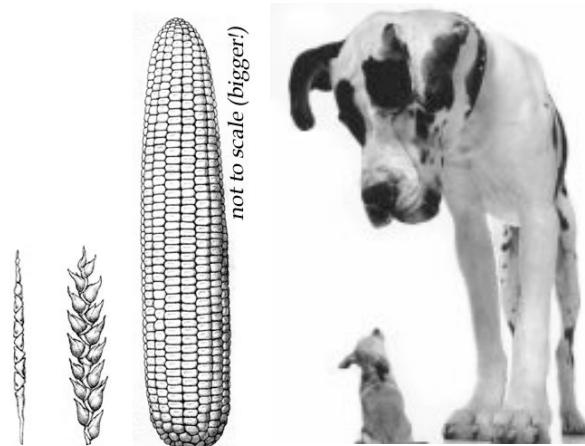


Here the answer is empirical. Traditional plant and animal breeders had come to recognize that offspring tended to display the same or similar traits as their parents. This observation led them to assume that there was some factor within the parents that was expressed within the offspring and could, in turn, be passed from the offspring to their own offspring. A classic example is the Habsburg lip, which was passed through a European ruling family for generations.⁵⁵ In the case of artificial selection, an important point to keep in mind is that the various types of domesticated organisms that are produced are often dependent for their continued existence on their human creators. This relieves them from the

⁵³ Molecular Evidence and the Evolution of Maize: <http://link.springer.com/article/10.1007/BF02860472>

⁵⁴ From wild animals to domestic pets, an evolutionary view of domestication: http://www.pnas.org/content/106/Supplement_1/9971.full

⁵⁵ 'Imperial Stigmata!' The Habsburg Lip, A Grotesque 'Mark' Of Royalty Through The Centuries!: <http://theesotericcuriosa.blogspot.com/2012/09/imperial-stigmata-habsburg-lip.html>



constraints they would experience in the wild. Because of this dependence, artificial selection can produce quite exaggerated and, in the absence of human intervention, highly deleterious traits. Just look at domesticated chickens and turkeys which, while not completely flightless, can fly only very short distances and so are extremely vulnerable to predators. Neither modern corn (*Zea mays*) or chihuahuas, one of the smallest breeds of dog, also developed by Mesoamerican breeders, would be expected to survive for long in the wild, that is, without human assistance.⁵⁶

Limits on populations

It is a given (that is, an empirically demonstrable fact) that all organisms are capable of producing many more than one copy of themselves. Consider, as an example, a breeding pair of elephants or a single asexually reproducing bacterium. Let us further assume that there are no limits to their reproduction. That is, that once born, the offspring will live a normal life-span and themselves reproduce. By the end of 500 years, a single pair of elephants could have produced ~15,000,000 living descendants.⁵⁷ Clearly if these 15,000,000 elephants then paired up to form 7,500,000 breeding pairs, within another 500 years (1000 years altogether) there would be $7.5 \times 10^6 \times 1.5 \times 10^7$ or 1.125×10^{14} elephants. Assuming that each adult elephant weighs ~6000 kilograms, which is the average between larger males and smaller females, the end result would be $\sim 6.75 \times 10^{18}$ kilograms of elephant. Allowed to continue unchecked, within a few thousand years a single pair of elephants could produce a mass of elephants larger than the mass of the Earth, an absurd conclusion. Clearly we must have left something out of our calculations! As another example, let us turn to a solitary bacterium, which needs no mate to reproduce. Let us assume that this is a photosynthetic bacterium that relies on sunlight and simple compounds, such as water, carbon dioxide, and some minerals, to grow. A bacterium is much smaller than an elephant but it can produce new bacteria at a much faster rate. Under optimal conditions, it could divide once every 20 minutes or so and would, within approximately a day, produce a mass of bacteria greater than that of Earth as a whole. Again, we are clearly making a number at least one mistake in our logic.

Elephants and bacteria are not the only types of organism on the Earth. In fact every known type of organism can produce many more offspring than are needed to replace themselves when they die. This trait is known as superfecundity. But unlimited growth does not and cannot happen for very long - other factors must constrain it. In fact, if you were to monitor the populations of most organisms, you would find that the numbers of a particular organism in a particular environment tend to fluctuate around a so-called steady state level. By steady state we mean that even though animals are continually being born and are dying, the number of organisms remains roughly constant.

*A single cell of the bacterium *E. coli* would, under ideal circumstances, divide every twenty minutes. That is not particularly disturbing until you think about it, but the fact is that bacteria multiply geometrically: one becomes two, two become four, four become eight, and so on. In this way it can be shown that in a single day, one cell of *E. coli* could produce a super-colony equal in size and weight to the entire planet Earth.*

*- Michael Crichton (1969) *The Andromeda Strain**

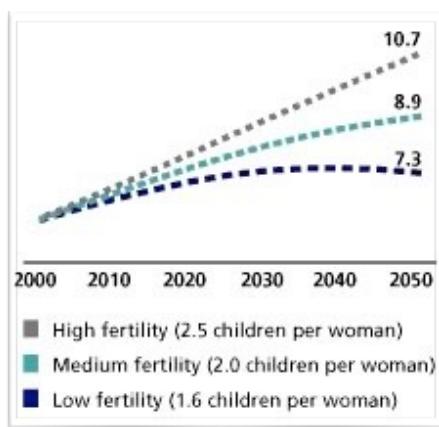
⁵⁶ How DNA sequence divides chihuahua and great dane: <http://www.theguardian.com/science/2007/apr/06/uknews.science>

⁵⁷ Darwin's elephants: <http://www.idlex.freeserve.co.uk/idle/evolution/sex/elephant.html>

So what balances the effects of superfecundity, what limits population growth? The obvious answer to this question is the fact that the resources needed for growth are limited and there are limited places for organisms to live. Thomas Malthus (1766-1834) was the first to clearly articulate the role of limited resources as a constraint on population. His was a purely logical argument. Competition between increasing numbers of organisms for a limited supply of resources would necessarily limit the number of organisms. Malthus painted a rather gloomy picture of organisms struggling with one another for access to these resources, with many living in an organismal version of poverty, starving to death because they could not out-compete others for the food or spaces they needed to thrive. One point that Malthus ignored, or more likely was ignorant of, is that organisms rarely behave in this way. It is common to find various types of behaviors that limit the direct struggle for resources. For example, in some organisms, an adult has to establish (and defend) a particular territory before it can successfully reproduce.⁵⁸ The end result of this type of behavior is to stabilize the population around a steady state level, which is a function of both environmental and behavioral constraints.

An organism's environment includes all factors that influence the organism and by which the organism influences other organisms and their environments. These include factors such as changes in climate, as well as changes in the presence or absence of other organisms. For example, if one organism depends in important ways upon another, the extinction of the first will necessarily influence the survival of the second.⁵⁹ Similarly, the introduction of a new type of organism or a new trait (think oxygenic photosynthesis) in an established environment can disrupt existing interactions and conditions. When the environment changes, the existing steady state population level may be unsustainable or many of the different types of organisms present may not be viable. If the climate gets drier or wetter, colder or hotter, if yearly temperatures reach greater extremes, or if new organisms (including new disease-causing pathogens) enter an area, the average population density may change or in some cases, if the environmental change is drastic enough, may even drop to zero, that is, certain

populations could go extinct. Environmental conditions and changes will influence the sustainable steady state population level of an organism (something to think about in the context of global warming, whatever the cause).



An immediate example of this type of behavior involves the human population. Once constrained by disease, war, and periodic famine, human population increased dramatically with the introduction of better public health and sanitation measures, a more secure food supply, and reductions in infant mortality. Now, in many countries, populations appear to be heading to a new steady state, although

⁵⁸ Territorial Defense, Territory Size, and Population Regulation: <https://iriss.stanford.edu/sites/all/files/shared/documents/Lopez-Sepulcre2005.pdf>

⁵⁹ Why the Avocado Should Have Gone the Way of the Dodo <http://www.smithsonianmag.com/arts-culture/why-the-avocado-should-have-gone-the-way-of-the-dodo-4976527/?no-ist> and Neotropical Anachronisms: The Fruits the Gomphotheres Ate: <http://www.sciencemag.org/content/215/4528/19.short>

exactly what that final population total level will be is unclear.⁶⁰ Various models have been developed based on different levels of average fertility. In a number of countries, the birth rate has already fallen into the low fertility domain, although that is no guarantee that it will stay there!⁶¹ In this domain (ignoring immigration), a country's population actually decreases over time, since the number of children born is not equal to the number of people dying. This can generate its own social stresses. Decreases in birth rate per woman correlate with reductions in infant mortality (generally due to vaccination, improved nutrition, and hygiene) and increases in the educational level and the reproductive "self-determination" (that is, the emancipation) of women. Where women have the right to control their reproductive behavior, the birth rate tends to be lower. Clearly changes in the environment, and here we include the sociopolitical environment, can dramatically influence behavior and serve to limit reproduction and population levels.

The conceptual leap made by Darwin and Wallace

What Darwin and Wallace recognized were the implications and significance of these key facts: the heritable nature of variation between organisms, the ability of organisms to reproduce many more offspring than are needed to replace themselves, and the constraints on population size due to limited environmental resources. Based on these facts, they drew a logical implication, namely that individuals would differ in their reproductive success – that is, different individuals would leave behind different number of descendants. Over time, we would expect that the phenotypic variations associated with greater reproductive success (and the genotypes associated with them) will increase in frequency within the population; they would replace those organisms with a less reproductively successful phenotype. Darwin termed this process natural selection, in analogy to the effects of artificial selection by plant and animal breeders. As we will see, natural selection is one of the major drivers of biological evolution.

Just to be clear, however, reproductive success is more, and more subtle, than survival of the fittest. First and foremost, from the perspective of future generations, surviving alone does not matter much if the organism fails to produce offspring. An organism's impact on future generations will depend not on how long it lives but on how many fertile offspring it generates. An organism that can produce many reproductively successful offspring at an early age will have more of an impact on subsequent generations than an organism that lives an extremely long time but has few offspring. Again, there is a subtle point here. It is not simply the number of offspring that matter but the relative number of reproductively successful offspring produced.

If we think about the factors that influence reproductive success, we can classify them into a number of distinct types. For example, organisms that reproduce sexually need access to mates, and must be able to deal successfully with the stresses associated with normal existence and reproduction. This includes the ability to obtain adequate nutrition and to avoid death from predators and pathogens. These are all parts of the organism's phenotype, which is what natural selection acts on. It is worth remembering, however, that not all traits are independent of one another. Often the mechanism (and

⁶⁰ Global population growth: https://www.ted.com/talks/hans_rosling_on_global_population_growth and The Joy of Stats: <http://youtu.be/bkSRLYSojo>

⁶¹ Hans Rosling: Religions and babies: <http://www.youtube.com/watch?v=ezVk1ahRF78>

genotype) involved in producing one trait also influences other traits – they are interdependent. There are also non-genetic sources of variation. For example, there are molecular level fluctuations that occur at the cellular level; these can lead genotypically identical cells to display different behaviors, that is, different phenotypes. Environmental factors can influence the growth, health, and behavior of organisms. These are generally termed physiological adaptations. An organism's genotype influences how it responds phenotypically to environmental factors, so the relationship between phenotype, genotype, and the organism's environment is complex.

Mutations and the origins of genotype-based variation

So now the question arises, what is the origin of genetic – that is inheritable-variation? How do genotypes change? As a simple (and not completely incorrect) analogy, we can think of an organism's genotype as a book. This book is also known as its **genome** (not to worry if this seems too simple, we will add needed complexities as we go along). An organism's genome is no ordinary book. For simplicity we can think of it as a single unbroken string of characters. In humans, this string is approximately 3.2 billion characters (or letters) long (~3,200,000,000). In case you are wondering, a character corresponds to a base pair, which we will consider in detail in Chapter 7. Within this string there are regions of what look like words and sentences, that is, regions that look like they have meaning. There are also long regions that appear to be meaningless. To continue our analogy, a few critical changes to the words in a sentence can change the meaning of a story, sometimes subtly, sometimes dramatically, and sometimes a change will lead to a story that makes no sense at all.

At this point we will define the meaningful regions (the words and sentences) to correspond to **genes** and the other intervening sequences as **intragenic** regions, that is, spaces between genes. We estimate that humans have approximately 25,000 genes (we will return to a molecular level discussion of genes and how they work in Chapters 7 through 9). As we continue to learn more about the molecular biology of organisms, our understanding of both genes and intragenic regions becomes increasingly sophisticated. The end result is that regions that appear meaningless can influence the meaning of the genome. Many regions of the genome are unique, they occur only once within the string of characters. Others are repeated, sometimes hundreds to thousands of times. When we compare the genotypes of individuals of the same type of organism, we find that they differ at a number of places. For example, we have found over 55,000,000 variations between human genomes and more are likely to be identified. When present within a population of organisms, these genotypic differences are known as **polymorphisms**, from the Latin meaning multiple forms. Polymorphisms are the basis for DNA-based forensic identification tests. One thing to note, however, is that only a small number of these variations are present within any one individual, and considering the size of the human genome, most people differ from one another less than 1 to 4 letters out of every 1000. That amounts to between 3 to 12 million letter differences between two unrelated individuals. Most of these differences are single characters, but there can be changes that involve moving regions from one place to another, or the deletion or duplication of a region. In sexually reproducing organisms, like humans, there are two copies of this book in each cell of the body, one derived from each of the organism's parents - organisms with two genomic "books" are known as **diploid**. When a sexual organism reproduces, it produces reproductive cells, known as sperm or eggs. Since each of these cells contains one copy of its own unique version of the genomic book, it is said to be **haploid**. This haploid genome is produced

through a complex process (known as meiosis) that leads to the significant shuffling between the organism's original parental genomes. The end result is that each new organism contains its own unique genomic book (or books). When the haploid sperm and haploid egg cells fuse a new and unique (diploid) organism is formed with its own unique pair of genomic books.

The origins of polymorphisms

So what produces the genomic variation between individuals found within current populations? Are these processes still continuing or have they ended? First, as we have alluded to (and will return to again and again), the sequence of letters in an organism's genome corresponds to the sequence of characters in DNA molecules. A DNA molecule is water (and over 70% of a typical cell is water) is thermodynamically unstable and can undergo various types of reactions that lead to changes in the sequences of characters within the molecule.⁶² In addition, we are continually bombarded by radiation that can damage DNA (although not to worry, the radiation associated with cell phones, bluetooth, and wifi is too low in energy to damage DNA). Mutagenic radiation, that is, the types of radiation capable of damaging the genome, comes from various sources, including cosmic rays that originate from outside of the solar system, UV light from the sun, the decay of naturally occurring radioactive isotopes found in rocks and soil, including radon, and the ingestion of naturally occurring isotopes, such as potassium 40. DNA molecules can absorb such radiation, which can lead to chemical changes (mutations). Many but not all of these changes can be identified and repaired by cellular systems, which we will consider later in the book.

The second, and major source of change to the genome involves the process of DNA replication. DNA replication happens every time a cell divides and is remarkably accurate but it is not perfect. Copying creates mistakes. In humans, it appears that replication creates one error for every 100,000,000 (10^8) characters copied. A proof-reading error repair system corrects ~99% of these errors, leading to an overall error rate during replication of 1 in 10^{10} bases replicated. Since a single human cell contains about 6,400,000,000 (> 6 billion) bases of DNA sequence, that means that less than one new mutation is introduced per cell division cycle. Given the number of generations from fertilized egg to sexually active adult, that corresponds to 100-200 new mutations (changes) added to an individual's genome per generation.⁶³ These mutations can have a wide range of effects, complicated by the fact that essentially all of the various aspects of an organism's phenotype are determined by the action of hundreds to thousands of genes working in a complex network. And here we introduce our last new terms for a while; when a mutation leads to change in a gene, it creates a new version of that gene, which is known as an **allele** of the gene. When a mutation changes the DNA's sequence, whether or not it is part of a gene, it creates what is known as a **sequence polymorphism** (a different DNA sequence). Once an allele or polymorphism has been generated, it is stable - it can be inherited from a parent and passed on to an offspring. Through the various processes associated with reproduction (which we will consider in detail later on), each organism carries its own distinctive set of alleles and its own unique set of polymorphisms. Taken together these genotypic

⁶² Instability and decay of the primary structure of DNA: <http://www.nature.com/nature/journal/v362/n6422/pdf/362709a0.pdf> and DNA has a 521-year half-life: <http://www.nature.com/news/dna-has-a-521-year-half-life-1.11555>

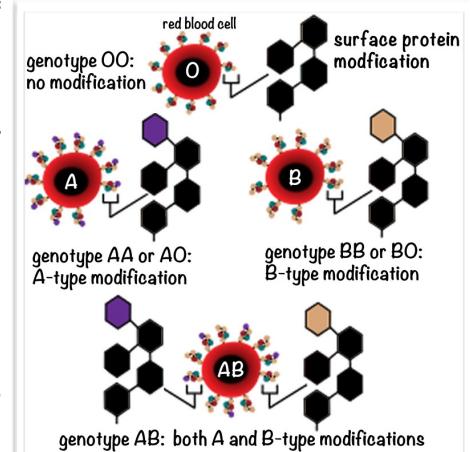
⁶³ Human mutation rate revealed: <http://www.nature.com/news/2009/090827/full/news.2009.864.html>

differences (different alleles and different polymorphisms) produce different phenotypes. The DNA tests used to determine paternity and forensic identity work because they identify the unique polymorphisms (and alleles) present within an individual's genome. We will return to and hopefully further clarify the significance of alleles and polymorphisms when we consider DNA in greater detail later on in this book.

Two points are worth noting about genomic changes or mutations. First, whether produced by mistakes in replication or chemical or photochemical reactions, it appears that these changes occur randomly within the genome. With a few notable and highly specific exceptions there are no known mechanisms by which the environment (or the organism) can influence where a mutation occurs. The second point is that a mutation may or may not influence an organism's phenotype. The effects of a mutation will depend on a number of factors, including exactly where the mutation is in the genome, its specific nature, the role of the mutated gene within the organism, the rest of the genome (the organism's genotype), and the environment in which the organism finds itself.

A short aside on the genotype-phenotype relationship

When we think about polymorphisms and alleles, it is tempting to assume simple relationships. In some ways, this is a residue from the way you may have been introduced to genetics in the past.⁶⁴ Perhaps you already know about Mendel and his peas. He identified distinct alleles of particular genes that were responsible for distinct phenotypes; yellow versus green peas, wrinkled versus smooth peas, tall versus short plants, etc. Other common examples might be the alleles associated with sickle cell anemia (and increased resistance to malarial infection) and the major blood types. Which alleles of the ABO gene you inherited determines whether you have O, A, B or AB blood type. Remember you are diploid, so you have two copies of each gene, including the ABO gene, in your genome, one inherited from your mom and one from your dad. There are a number of common alleles of the ABO gene present in the human population, the most common (by far) are the A, B, and O alleles. The two ABO alleles you inherited from your parents may be the same or different. If they are A and B, you have the AB blood type; if A and O or A and A, you have the A blood type, if B and O or B and B, you have the B blood type, or if you have O and O, you have the O blood type. These are examples of discrete traits; you are either A, B, AB, or O blood type – there are no intermediates. You cannot be 90% A and 10% B.⁶⁵ As we will see, this situation occurs when a particular gene determines the trait; in the case of the ABO gene, the nature of the gene product determines the modification of surface proteins on red blood cells. The O allele leads to no modification, the A allele leads to an A-type modification, while the B allele leads to a B-type modification. When A and B alleles are present, both types of modifications occur. However, most traits do not behave in such a simple way.

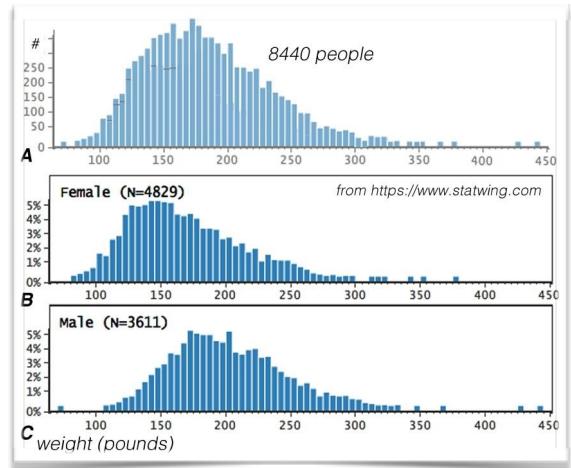


⁶⁴ We call this type of thinking didaskalogenic: <http://en.wikipedia.org/wiki/Didaskalogenic>

⁶⁵ Human blood types have deep evolutionary roots: <https://www.sciencenews.org/article/human-blood-types-have-deep-evolutionary-roots>

The vast majority of traits, however, are continuous rather than discrete. For example, people come in a continuous range of heights, rather than in discrete sizes. If we look at the values of the trait within a population, that is, if we can associate a discrete number to the trait (which one cannot always do), we find that each population can be characterized by a distribution. For example, let us consider the distributions of weights in a group of 8440 adults in the USA (see →). The top panel (A) presents a graph of the weights (along the horizontal or x-axis) versus the number of people with that weight (along the vertical or y-axis). We can define the “mean” or average of the population (\bar{x}) as the sum of the individual values of a trait (in this case each person’s weight) divided by the number of individuals measured, as defined by the equation:

$$\bar{x} = \frac{x_1 + x_2 + \dots + x_n}{n}$$



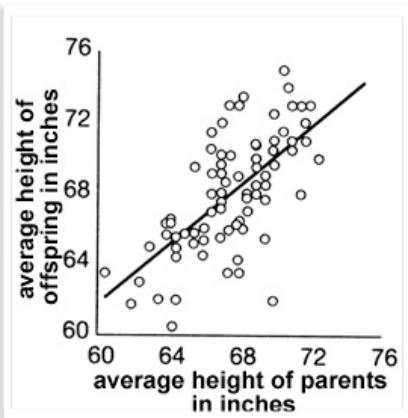
In this case, the mean weight of the population is 180 pounds. It is common to recognize another characteristic of the population, namely the median. The median is the point at which half of the individuals have a smaller value of the trait and half have a larger value. In this case, the median is 176. Because the mean does not equal the median, we say that the distribution is asymmetric, that is there are more people who are heavier than the mean value compared to those who are lighter. For the moment we will ignore this asymmetry, particularly since it is not dramatic. Another way to characterize the shape of the distribution is by what is known as its standard deviation (σ). There are different versions of the standard deviation that reflect the shape of the population distribution, but for our purposes we will take a simple one, the so-called uncorrected sample standard deviation.⁶⁶ To calculate this value, you subtract the mean value for the population (\bar{x}) from the value for each individual (x_i); since x_i can be larger or smaller than the mean, this difference can be a positive or a negative number. We then take the square of the difference which makes all values positive (hopefully this makes sense to you). We sum these squared differences together, divide that sum by the number of individuals in the population (N), and take the square root (which reverses the effects of our squaring x_i) to arrive at the standard deviation of the population. The smaller the standard deviation, the narrower the distribution - the more organisms in the population have a value similar to the mean. The larger is σ , the greater is the extent of the variation in the trait.

$$\sigma = \sqrt{\frac{1}{N} \sum_{i=1}^N (x_i - \bar{x})^2}$$

So how do we determine whether a particularly complex trait like weight (or any other non-discrete, continuously varying trait) is genetically determined? We could imagine, for example, that an organism’s weight is simply a matter of how easy it was for it to get food. The standard approach is to ask whether there is a correlation between the phenotypes of the parents and the phenotypes of the offspring. That such a correlation between parents and offspring exists for height is suggested by the graph on the next page. Such a correlation serves as evidence that height (or any other quantifiable trait) is at least to some extent genetically determined. What we cannot determine from such a

⁶⁶ http://en.wikipedia.org/wiki/Standard_deviation <http://www.mathsisfun.com/data/standard-deviation.html>

relationship, however, is how many genes are involved in the genetic determination of height or how their effects are influenced by the environment and environmental history which the offspring experience. For example, "human height has been increasing since at least the 19th century when comprehensive records first began. The mean height of Dutchmen, for example, has increased from 165cm in 1860 to a current 184cm. The spectacular rise in height probably reflects improvements in health care and diet", rather than changes in genes.⁶⁷ Geneticists currently estimate that allelic differences at more than 50 genes make significant contributions to the determination of height, with alleles at hundreds more having smaller effects that contribute to differences in height.⁶⁸ At the same time, specific alleles of certain genes can lead to extreme shortness or tallness. For example, mutations that inactivate or over-activate genes encoding factors required for growth can lead to dwarfism or giantism.



On a related didaskalogenic note, you may remember learning that alleles are often described as dominant or recessive. But the extent to which an allele is dominant or recessive is not necessarily absolute, it depends upon how well we define a particular trait and whether it can be influenced by other factors and other genes. These effects reveal themselves through the fact that people carrying the same alleles of a particular gene can display (or not display) the associated trait, which is known as its penetrance, and they can vary in the strength of the trait, which is known as its expressivity. Both the penetrance and expressivity of a trait can be influenced by the rest of the genome, that is, by which alleles of other genes are present. Environmental factors can also have significant effects on the phenotype associated with a particular allele or genotype.

Questions to answer & to ponder:

- Explain why superfecundity is required for evolution to occur.
- Why is the presence of inheritable variation important for any evolutionary model?
- How did plant and animal breeders inspire Darwin's thinking on evolution?
- From a practical point of view, what makes it possible for plant and animal breeding to produce distinctive types of organisms?
- What factors might lead to a new steady state level in the human population?
- How might the accumulation of mutations be used to determine the relationship between organisms?
- Why might the products of artificial selection not be competitive with "native" organisms?

Variation, selection, and isolation (speciation)

Darwin and Wallace's breakthrough conclusion was that genetic variation within a population would lead to altered reproductive success among the members of that population. Some genotypes, and the alleles of genes they contain, would become more common within subsequent generations because the individuals that contained them would reproduce more successfully. Other alleles and genotypes would become less common. The effects of specific alleles on an organism's reproductive

⁶⁷ "From Galton to GWAS: quantitative genetics of human height": <http://www.ncbi.nlm.nih.gov/pubmed/21429269>

⁶⁸ Genetics of human height: <http://www.ncbi.nlm.nih.gov/pubmed/19818695>

success would, of course, be influenced by the rest of the organism's genotype, its structure and behaviors (both selectable traits) and its environment. While some alleles can have a strong positive or negative impact on reproductive success, the effects of most alleles are subtle, assuming they produce any noticeable phenotypic effect at all. A strong positive effect will increase the frequency of the allele (and genotype) associated with it in future generations, while a strong negative effect can lead to the allele disappearing altogether from the population. At the same time, many alleles have more subtle, less strongly selectable effects. An allele that increases the probability of death before reproductive age is likely to be strongly selected against, whereas an allele that has only modest effects on the number of offspring an organism produces will be relatively weakly selected for.

Types of simple selection

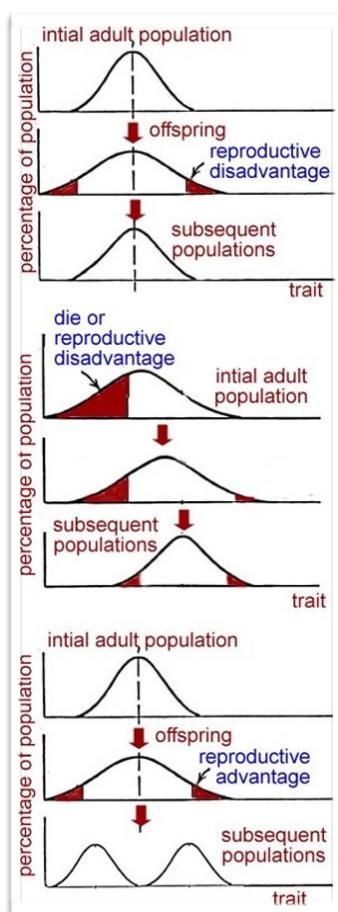
While it is something of an oversimplification (we will introduce the complexities associated with the random aspects of reproduction and the linked nature of genes shortly), we will begin with the three basic types of selection: conservative, directed, and disruptive. We start with a population composed of individuals displaying genetic variation in a particular trait. The ongoing processes of mutation continually introduces new genotypes, and their associated phenotypic effects. What is important to remember is that changes in the population and the environment can influence the predominant type of selection occurring over time, and that different types of selection may well (and most certainly are) occurring for different traits.

For each type of selection, we illustrate the effects as if they were acting along a single dimension, for example smaller to larger, or stronger to weaker, lighter to darker, slower to faster. In fact, most traits vary along a number of dimensions. For example, consider the trait of ear, paw, heart, or big toe shape. An appropriate type of graph would be a multi-dimensional surface, but that is harder to draw. Also, for simplicity, we start with populations whose distribution for a particular trait can be described by a simple and symmetrical curve, that is the mean and the median are equal. New variants, based on new mutations, generally fall more or less randomly within this distribution. Under these conditions, for selection NOT to occur we would have to make two seriously unrealistic assumptions: first that all organisms are equally successful at producing offspring, and second that each organism or pair of organisms produce only one or two (respectively) offspring. Whenever these are not the case, which is always, selective processes will occur, although the strength of selection may vary dramatically between traits.

Conservative selection: Sometimes a population of organisms appears static for extended periods of time, that is, the mean and standard deviation of a trait are not changing. Does that mean that selection has stopped? Obviously we can turn this question around, assume that there is a population with a certain stable mean and standard deviation of a trait. What would happen over time if selection disappeared?

Let us assume we are dealing with an established population living in a stable environment. This is a real world population, where organisms are capable of reproducing more, and sometimes, many more organisms than are needed to replace them when they die and that these organisms mate with one another randomly. Now we have to consider the factors that lead to the population distribution to being with: why is the mean value of the trait the value it is? What factors influence the standard

deviation? Assuming that natural selection is active, it must be that organisms that display a value of the trait far from the mean are (on average) at a reproductive disadvantage compared to those with the mean value of the trait. We do not know why this is the case (and don't really care at the moment). Now if selection (at least for this value of the trait) is acting, what happens? The organisms far from the mean are no longer at a reproductive disadvantage, so their numbers in the population will increase. The standard deviation will grow larger, until at the extreme, the distribution would be flat, characterized by a maximum and a minimum value. New mutations and existing alleles that alter the trait value will not be selected against, so they will increase in frequency. But in our real population, the mean and standard deviation associated with the trait remain constant. We can then predict selection against extreme values of the trait and can measure that selection "pressure" by following the reproductive success of individuals in the population with different values of the trait we have been considering. We would also predict that the more extreme the trait, that is, the further from the population mean, the greater its reproductive disadvantage would be, so that with each generation, the contribution of these outliers will be reduced. The distribution's mean will remain constant. The stronger the disadvantage the outliers face, the narrower the distribution will be – that is, the smaller the standard deviation. In the end, the size of the standard deviation will reflect both the strength of selection against outliers and the rate at which new variation enters the population through mutation. Similarly, we might predict that where a trait's distribution is broad, one might hypothesize that the impact of the trait on reproductive success is relatively weak.



Directed selection: Now imagine that the population's environment changes, and that it is no longer the case that the phenotype of the mean is the optimal phenotype, in terms of reproductive success. It could be that a smaller or a larger value is now more favorable. Under these conditions, we would expect that the mean of the distribution would shift toward the phenotypic value associated with maximum reproductive success over time. Once reached, and assuming the environment stays constant, conservative selection again becomes the predominant process. For directed selection to work, the environment must change at a rate and to an extent compatible with the changing mean phenotype of the population. Too big a change and the reproductive success of all members of the population could be dramatically reduced. The ability of the population to change will depend upon the variation already present within the population. While new mutations leading to new alleles are appearing, this is a relatively slow process. In some cases, the change in the environment may be so fast or so drastic and the associated impact on reproduction so severe that selection will fail to move the population and extinction will occur. One outcome to emerge from a changing environment leading to the directed selection is that as the selected population's mean moves, it may well alter the environment of other organisms.

Disruptive selection: A third possibility is that organisms find themselves in an environment in which traits at the extremes of the population distribution have a reproductive advantage over those nearer

the mean. If we think about the trait distribution as a multidimensional surface, it is possible that in a particular environment, there will be multiple and distinct strategies that lead to greater reproductive success compared to others. This leads to what is known as disruptive selection. The effect of disruptive selection in a sexually reproducing population will be opposed by the random mating between members of the population. But is random mating a good assumption? It could be that the different environments, which we will refer to as ecological niches, are physically distant from one another and organisms simply do not travel far to find a mate. The population will split into subpopulations in the process of adapting to the two different niches. Over time, two species could emerge, since whom one chooses to mate with and the productivity of that mating, are themselves selectable traits.

A short note on pedagogical weirdness

Many students are introduced into the field of population genetics and evolutionary mechanisms – that is, how phenotypes, genotypes, and allele frequencies change in the face of selective and environmental pressures – through what is known as the Hardy-Weinberg (H-W) equilibrium equation. Many H-W equation problems have been solved, but the question is why? From a historical perspective, the work of G.H. Hardy and Wilhelm Weinberg (published independently in 1908) resolved the question of whether, in a *non-evolving population*, dominant alleles would replace recessive alleles over time. So what does that mean? Remember (and we will return to this later), in a diploid organism two copies of each gene are present. Each gene may be represented by different alleles. Where the two alleles are different, the one associated with the expressed (visible) phenotypic trait is said to be dominant to the other, which is termed recessive.⁶⁹ Geneticists previously believed that dominant alleles and traits were somehow “stronger” than recessive alleles or traits, but this is simply not the case and it is certainly not clear that this belief makes sense at the molecular level, as we will see. The relationship between allele and trait is complex. For example, an allele may be dominant for one phenotype and recessive for another (think about malarial resistance and sickle cell anemia, both due to the same allele in one or two copies.) What Hardy & Weinberg demonstrated was that in a *non-evolving* system, the original percentage of dominant and recessive alleles at various genetic loci (genes) stays constant. What is important to remember however is that this conclusion is based on five totally unrealistic assumptions, namely that: 1) the population is essentially infinite, so we did not have to consider processes like genetic drift (discussed below); 2) the population is isolated, no individuals left and none entered; 3) mutations do not occur; 4) mating between individuals is completely random (discussed further in Chapter 4); and 5) there are no differential reproductive effects, that is, no natural selection.⁷⁰ Typically H-W problems are used to drive students crazy and (more seriously) to identify situations where one of the assumptions upon which they are based is untrue (which are essentially all actual situations).

Questions to answer & ponder:

- Why does variation never completely disappear even in the face of conservative selection?

⁶⁹ In the context of the ABO gene for blood type, A and B alleles are dominant to O, which is recessive. Neither A nor B are dominant or recessive with respect to one another.

⁷⁰ Hardy-Weinberg Equilibrium: <http://www.tiem.utk.edu/~gross/bioed/bealsmodules/hardy-weinberg.html>

- What would lead conservative selection to be replaced by directed or disruptive selection?
- Explain the caveats associated with assuming that you know why a trait was selected.
- optional exercise: virtuallaboratory on adaptation:
<http://virtuallaboratory.colorado.edu/BioFun-Support/labs/Adaptation/Adaptation.html>

Population size, founder effects and population bottlenecks

When we think about evolutionary processes from a Hardy-Weinberg perspective, we can ignore some extremely important situations that we would otherwise expect to impact populations. Things get more interesting when we take into consideration these non-exceptional processes. For example, what happens when a small number of organisms (derived from a much larger population) colonize a new environment? This is a situation, known as the founder effect, that is particularly relevant in island ecologies but also applies to pioneer populations migrating into new territories and then becoming isolated from their parent populations. Something similar happens when a large population is dramatically reduced, a situation known as a population bottleneck. Various types of environmental catastrophe, such as the appearance of a new pathogen, a new predator, or rapid climate change caused by volcanic activity, a cosmic collision, or a zombie apocalypse can cause population bottleneck. In both founder effect and population bottleneck situations, small populations become more susceptible to the effects of random fluctuations in survival and reproductive mechanisms, commonly referred to as genetic drift. In each case, given the dynamics of environmental change and population migrations, a population can come to develop unique traits through founder effects, population bottlenecks, and genetic drift. This can lead to the development of unexpected and advantageous traits that result in a selective advantage over the descendants of its parental population.

If we think of evolutionary changes as the movement of the population through a fitness landscape (the combination of the various factors that influence reproductive success), then isolation and evolutionary change of small populations can relieve, at least temporarily, the intensity of selective pressure and make possible the development and dispersal of new adaptations. For example, one effect of the major extinctions that have occurred during the evolution of life on Earth is that they provide a relaxed context for the evolution of new forms, a less densely-populated playing field, if you will. The expansion of the various types of mammals that followed the extinction of the dinosaurs is an example of one such opportunity, associated with changes in selection pressure.

Founder effects: What happens when a small subpopulation becomes isolated from its parent population? The original (large) population will contain a number of genotypes (and alleles), and if it is in a stable environment it will be governed primarily (as a first order approximation) by conservative selection. We can characterize this parental population in terms of the frequencies of the various alleles present within it. For the moment, we will ignore the effects of new mutations, which will continue to arise. Now assume that a small group of organisms from this parent population comes to colonize a new, geographically separate environment and that it is then isolated from its parental population, so that no individuals travel between the parent and the colonizing population. The classic example of such a situation is the colonization of newly formed islands, but the same process applies more generally during various types of migrations. The small isolated group is unlikely to have the same distribution of alleles as the original parent population. Why is that? It is a question of the randomness of sampling of the population. For example, if rolled often enough (or an infinite number of times), a fair

six sided (cubical) die would be expected to produce the numbers 1, 2, 3, 4, 5, and 6 with equal probabilities. Each would appear 1/6th of the time. But imagine that the number of rolls is limited and relatively small. Would you expect to get each number appearing with equal probability? You can check your intuition using this applet [[DiceExperiment](#)]. See how many throws are required to arrive at an equal 1/6th probability distribution; the number is almost certainly much larger than you would guess. We can translate this onto populations in the following way: Imagine a population in which each individual carries one of six alleles and the percentage of each type is equal (1/6th). The selection of any one individual from this population is like a throw of the die, there is an equal 1/6th chance of selecting an individual with one of the six alleles. Since the parental population is large, the removal of one individual does not appreciably change the distribution of alleles remaining, so the selection of a second individual produces a result that is independent of the first just like rolls of die and equally likely to result in a 1/6th chance to produce any one of the six alleles. But producing a small subpopulation with 1/6th of each allele (or the same percentages of various alleles as are present in the parent population) is, like the die experiment above, very unlikely. The more genetically complex the parent population, the more unlikely it is; imagine that the smaller colonizing population only has, for example, 3 members (three rolls of the die) – not all alleles present in the original population will be represented. Similarly, the smaller the subpopulation the more unlikely it is. So when a small group from a parent population invades or migrates into a new environment, it will very likely have a different genotypical profile from the parent population. This is a difference that is due not to natural selection but rather chance alone. Nevertheless, it will influence subsequent evolutionary events, first because the small subpopulation is likely to be significantly simpler genetically than the original population and so likely to respond in different ways to new mutations and environmental pressures, and second, because the exact alleles present will influence the phenotypes associated with new combinations (genotypes) and new mutations.

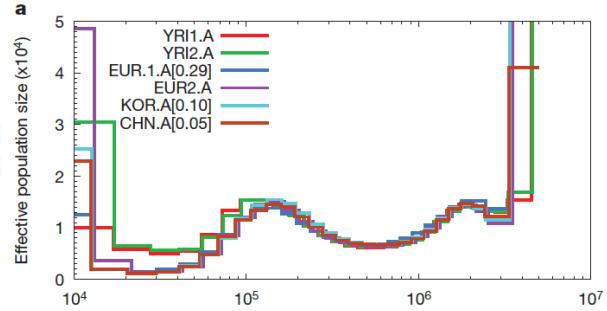
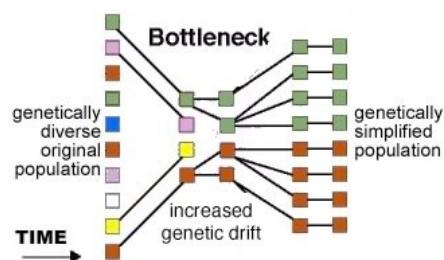
Because the human species appears to have emerged in Africa approximately 200,000 years ago, the people living in Africa represent the parent population of *Homo sapiens*. Genetic studies indicate that the African population displays a much greater genotypic complexity than do groups derived from the original African population, that is, everyone else. What remains controversial is the extent to which migrating populations of humans in-bred with what are known as archaic humanoids (such as Neanderthals and the Denisovians), which diverged from our lineage (*Homo sapiens*) approximately 1.2 million years ago.⁷¹

Population bottlenecks

A population bottleneck is similar in important ways to the founder effect. Population bottlenecks occur when some environmental change leads to the dramatic reduction of the size of a population. Catastrophic environmental changes, such as asteroid impacts, massive and prolonged volcanic eruptions (such as associated with continental drift), or the introduction of a particularly deadly pathogen, which kills a high percentage of the organisms that it infects can all create population bottleneck effects. Which organisms survive most types of bottlenecks will be random, that is unrelated to genotype (think of the immediate effects of an asteroid or the effects on a island-bound population

⁷¹ Genetic Data and Fossil Evidence Tell Differing Tales of Human Origins: <http://www.nytimes.com/2012/07/27/science/cousins-of-neanderthals-left-dna-in-africa-scientists-report.html?pagewanted=all>

when the volcanic island they inhabit blows up or mostly blows up). There is compelling evidence that such drastic environmental events are responsible for population bottlenecks so severe that they led to mass extinctions. The most catastrophic of these extinction events was the Permian extinction that occurred ~251 million years ago, during which it appears that ~95% of marine organisms and ~75% of land species died off.⁷² If most species were effected, we would not be surprised if the surviving populations experienced serious bottlenecks. The subsequent diversification of the surviving organisms, such as the dinosauria (which includes the extinct dinosaurs and modern birds) and the cynodontia, which includes the ancestors of modern mammals, including us, could be due in part to these bottleneck-associated effects, for example, through the removal of competing species or predators. A second catastrophic event occurred around 65 million years ago, which contributed to the extinction of the dinosaurs and led to the diversification of mammals, particularly the placental mammals.



In other cases, however, the effects of a bottleneck may not be random. Consider the effects of a severe drought or highly virulent bacterial or viral infection; the organisms that survive may have specific phenotypes (and associated genotypes) that increased their chances of survival. In such a case, the effect of the bottlenecking event would produce non-random changes in the distribution of genotypes (and alleles) in the post bottleneck population – these selective agents could continue to influence the population in various ways. For example, a trait associated with pathogen resistance may have other, even negative effects on phenotype, but these negative effects could be less important than the positive effect of surviving infection. In addition, the very occurrence of a rapid and extreme reduction in population size has its own effects. For example, it would be expected to increase the effects of genetic drift (see below).

We can identify extreme population reduction events such as founder effects and bottlenecks by looking at the variation in genotypes, particularly in genotypic changes not expected to influence phenotypes, mating preference, or reproductive success. These so-called neutral polymorphisms are expected to accumulate in the nonsense (intragenic) parts of the genome at a constant rate over time. The rate of the accumulation of such neutral polymorphisms is a type of population-based biological clock. Its rate can be estimated, at least roughly, by comparing the genotypes of individuals that are derived from populations in which the time of separation can be accurately estimated. For example, these types of studies indicate that the size of the human population dropped to a few thousands individuals between 20,000 to 40,000 years ago. This is a small number of people, likely to have been spread over a large area.⁷³ This bottleneck occurred around the time of the major migration of people

⁷² The Permian extinction and the evolution of endothermy: http://www.nap.edu/openbook.php?record_id=11630&page=133

⁷³ Late Pleistocene human population bottlenecks, volcanic winter, and differentiation of modern humans: http://ice2.uab.cat/argo/Argo_actualitzacio/argo_butlleti/ccee/geologia/arxius/1Ambrose%201998.pdf

out of Africa into Europe and Asia. Comparing genotypes, that is, neutral polymorphisms, between isolated populations also leads to estimates that aboriginal Australians reached Australia about 50,000 years ago, well before other human migrations⁷⁴ and that humans arrived in the Americas in multiple waves beginning around 15,000 to 16,000 years ago.⁷⁵ The arrival of humans into a new environment (another violation of the Hardy-Weinberg premises) has been linked to the extinction of a group of mammals known as the megafauna in those environments.⁷⁶ The presence of humans changed the environmental pressures on these organisms around the world.



Drawing: Rob Warren

Genetic drift

Genetic drift is an evolutionary phenomena that is difficult to comprehend in a strict Hardy-Weinberg world and explains the fact that most primates depend on the presence of vitamin C (ascorbic acid) in their diet. Primates are divided into two suborders, the Haplorhini (from the Greek meaning “dry noses”) and the Strepsirrhini (from the Greek meaning “wet noses”). The Strepsirrhini contain the lemurs and lorises, while the Haplorhini include the tarsiers and the anthropoids (monkeys, apes, and humans). One characteristic trait of the Haplorhini is that they share a requirement for ascorbic acid (vitamin C) in their diet. In vertebrates, vitamin C plays an essential role in the synthesis of collagen, a protein involved in the structural integrity of a wide range of connective tissues. In humans, the absence of dietary vitamin C leads to the disease scurvy, which according to Wikipedia, *“often presents itself initially as symptoms of malaise and lethargy, followed by formation of spots on the skin, spongy gums, and bleeding from the mucous membranes. Spots are most abundant on the thighs and legs, and a person with the ailment looks pale, feels depressed, and is partially immobilized. As scurvy advances, there can be open, suppurating wounds, loss of teeth, jaundice, fever, neuropathy, and death.”*⁷⁷ The requirement for dietary vitamin C is due to a mutation in a gene, known as *gulo1*, which encodes the enzyme 1-gulono-gamma-lactone oxidase (Gulo1) required for the synthesis of vitamin C. One can show that the absence of a functional *gulo1* allele is the root cause of vitamin C dependence in Haplorrhini by putting a working copy of the *gulo1* gene, for example derived from the mouse, into human cells. The mouse-derived, *gulo1* allele, which encodes a functional form of the Gulo1 enzyme cures the human cells’ need for exogenous vitamin C. But, no matter how advantageous a working *gulo1* allele would be (particularly for British sailors, who died in large numbers before the discovery of a preventative treatment for scurvy was discovered, a depressing story in its own right⁷⁸), no new *gulo1* allele appeared. Organisms do not always produce the alleles they need or that might be

⁷⁴ <http://www.sciencemag.org/content/334/6052/94.short>

⁷⁵ Reich et al., 2012. Reconstructing Native American population history. Nature; DOI: [10.1038/nature11258](https://doi.org/10.1038/nature11258)

⁷⁶ <http://australianmuseum.net.au/Megafauna-extinction-theories-patterns-of-extinction> and a very interesting video: <http://youtu.be/8WZ5Q2JYbLY>

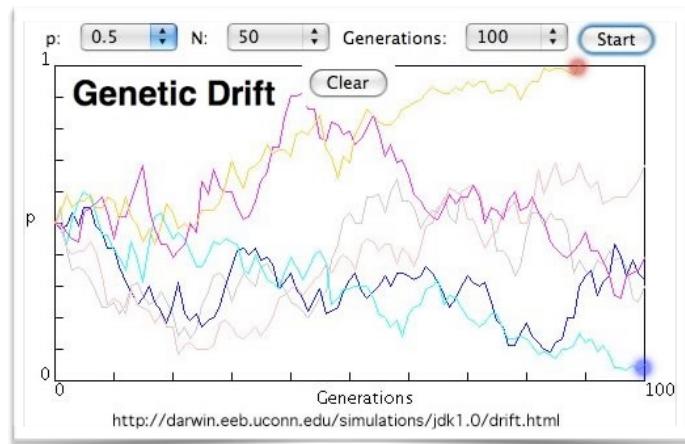
⁷⁷ One amazing fact is that it took various navies the deaths of thousands of sailors to understand the nutritional challenges of vitamin C. ADD REFERENCE

⁷⁸ <http://mentalfloss.com/article/24149/how-scurvy-was-cured-then-cure-was-lost>

beneficial, such alleles must be selected from alleles already present in the population or that appear through mutation.

This mutant allele appears to have become fixed in the ancestral population that gave rise to the Haplorrhini ~40 million years ago. So the question is, how did we (that is our ancestors) come to lose a functional version of such an important gene? It seems obvious that when the non-functional allele became universal in that population, the inability to make vitamin C must not have been strongly selected against. We can imagine such an environment and associated behavior; namely, these organisms must have obtained sufficient vitamin C from their diet, so that the loss of the ability to synthesize vitamin C themselves had little negative effect on them.

So how were function alleles involved in vitamin C synthesis lost? In small populations, non-adaptive – that is, non-beneficial and even mildly deleterious – genotypic changes and their associated traits can increase in frequency through a process known as genetic drift. In such populations, selection continues to be active, but it has significant effects only for traits (and their associated alleles) when the trait strongly influences reproductive success. While genetic drift occurs in asexual populations, due to random effects on organismic survival, it is particularly prominent in sexually reproducing species. This is because cells known as gametes are produced during the process of sexual reproduction (Chapter 4). While the cell that generates these gametes contains two copies of each gene, and each gene can be one of a number of alleles within the population, any particular gamete contains only one allele of each gene. To generate a new organism, two gametes fuse to produce a diploid organism. This process combines a number of chance events: which two gametes fuse is generally a matter of chance, and which particular alleles each gamete contains is again a matter of chance. In a small population, over a reasonably small number of generations, one or the other alleles at a particular genetic locus will be lost, and given enough time, this allelic loss approaches a certainty. In this figure, six different experimental outcomes (each line) are analyzed over the course of 100 generations. In each case, the population size is set to 50, and at the start of the experiment half the individuals have one allele and half have the other. While we are watching only one genetic locus, this same type of behavior impacts every gene for which multiple alleles (polymorphisms) exist. In one of these six populations, one allele has been lost (red dot), in the other (blue dot), the other allele is close to being lost. When a particular allele becomes the only allele within a population, it is said to have been fixed. Assume that the two alleles convey no selective advantage, can you predict what will happen if we let the experiment run through 10,000 generations? If you are feeling mathematically inclined, you can even calculate the effect of mild to moderate positive or negative selective pressures on allele frequencies and the probability that a particular allele will be lost or fixed.



Since the rest of the organism's genotype often influences the phenotype associated with the presence of a particular allele, the presence or absence of various alleles within the population can influence the phenotypes observed. If an allele disappears because of genetic drift, future evolutionary

changes may be constrained (or perhaps better put, redirected). At each point, the future directions open to evolutionary mechanisms depend in large measure on the alleles currently present in the population. For example, what happens if drift leads to the fixation of a mildly deleterious allele, let us call this allele BBY. Now the presence of BBY will change the selective landscape: mutations and/or alleles that ameliorate the negative effects of aBBY will increase reproductive success, selection pressures will select for those alleles. This can lead to evolution changing direction even if only subtly. With similar effects going on across the genome, one quickly begins to understand why evolution is something like a drunken walk across a selective landscape, with genetic drift and founder and bottleneck effects resulting in periodic staggers in random directions.⁷⁹

This use of pre-existing variation, rather than the idea that an organism would invent variations in its genome as it needed them, was a key point in Darwin's view of evolutionary processes. The organism cannot create the alleles it might need nor are there any processes known that can produce specific alleles in order to produce specific phenotypes. Rather, the allelic variation generated by mutation, selection, and drift are all that evolutionary processes have with which to work. Only a rare mutation that recreates the lost allele can bring an allele back into the population once it has been lost. Founder and bottleneck effects, together with genetic drift combine to produce what are known as non-adaptive processes and make the history of a population a critical determinant of its future evolution.

Questions to answer & ponder:

- How does the extinction of one type of organism influence the evolution of others?
- How can a founder effect/bottleneck lead to a slightly deleterious mutation becoming common in a population?
- Why is the common need of a subclass of primates for vitamin C evidence for a common ancestor?
- Consider the various ways that the individuals that fail to pass through a bottleneck might differ from those that do. How many "reasons" can you identify?
- How does selection act to limit the effects of genetic drift? Under what conditions does genetic drift influence selection?
- Describe the relative effects of selection and drift following a bottleneck?
- How is it that drift can be quantified, but in any particular experiment, not predicted?
- Does passing through a bottleneck improve or hamper a population's chances for evolutionary success (that is, avoiding extinction)?

Gene linkage: one more complication

So far, we have not worried overly much about the organization of genes in an organism. It could be that each gene behaves like an isolate object, but in fact that is not the case. We bring it up here because the way genes are organized can, in fact, influence evolutionary processes. In his original genetic analyses, Gregor Mendel (1822 – 1884) spent a fair amount of time looking for "well behaved" genes and alleles, that is those that displayed simple recessive and dominant behaviors and that acted as if they were completely independent from one another. But it quickly became clear that these behaviors are not how most genes behave. In fact, they act as if they are linked together, because they are (as we will see, gene linkage arises from the organization of genes within the DNA molecules.) So what happens when a particular allele of a particular gene is highly selected for or against, based on its effects on reproductive success? That allele, together with whatever alleles are found in genes located

⁷⁹ Genetic drift: <http://darwin.eeb.uconn.edu/simulations/jdk1.0/drift.html>

near it, are also selected. We can think of this as a by stander (or sometimes termed a “piggy-back”) effect, where alleles are being selected not because of their inherent effects on reproductive success, but their location within the genome.

Linkage between genes is not a permanent situation. As we will see toward the end of the course, there are processes that can shuffle the alleles (versions of genes) on chromosomes, the end result of which is the further away two genes are from one another on a chromosome, the more likely alleles of those genes will appear to be unlinked. Over a certain distance, they will always appear unlinked. This means that effects of linkage will eventually be lost, but not necessarily before particular alleles are fixed. For example, extremely strong selection for a particular allele of gene A will lead to the fixation of alleles at neighboring genes; similarly, strong selection against a particular allele of gene A will lead to apparent selection against alleles in neighboring genes. This effect, together with other non-selective effects, such as genetic drift, can produce mildly non-advantageous traits. It is also possible that a trait that increases reproductive success, that is the number of surviving offspring, may have other not-so-beneficial, and sometime seriously detrimental effects - the key is to remember that evolutionary mechanisms do not result in what is best for an individual organism but what in the end enhances reproductive success. In this sense, they do not select for particular genes or versions of genes but rather for combinations of genes that optimize reproductive success. In this light, talking about selfish genes, as if a gene can exist outside of an organism, makes little sense. Evolution can be a rather dispassionate and even cruel process, if you personify it.

Of course, the situation gets more complex when evolutionary mechanisms generate organisms, like humans, who feel and can object to the outcomes of evolutionary processes. How such organisms come to be and the implications of their existence are deeply complex topics. In some cases, they may be the unintended side effects of selection for a particular trait; in other cases they arise from processes known as inclusive fitness and social evolution, which we will deal with in more detail in the next chapter.

A brief reflection on the complexity of phenotypic traits

We can classify traits into three general groups. Adaptive traits are those that, when present increase the organism’s reproductive success. These are the traits we normally think about when we think about evolutionary processes. Non-adaptive traits are those generated by stochastic processes, like drift and bottlenecks. These traits become established not because they improve reproductive success but simply because they happened to be fixed randomly within the population. Some of these non-adaptive traits can in fact be deleterious only in specific situations, for example when humans with a non-functioning *gulo-1* allele attempt to live on a diet from which vitamin C is absent. Of course, if an allele is extremely deleterious (particularly if it behaves in a dominant, genetically and environmentally independent manner), it will disappear from the population due to selection. If it reappears, it is most likely to be due to a new (spontaneous) mutation that occurred within the affected individual or their parents. That said, when we consider an allele deleterious, we mean in terms of reproductive success. An allele can harm the individual organism carrying it yet persist in the population because it improves reproductive success. Similarly, an allele can be slightly positive in its effects, but again, its presence within the population is not directly due to these positive effects. Finally,

there are traits that could be seen as actively maladaptive, but which occur because they are linked, either genetically or mechanistically, to another positively-selected, adaptive trait. Many genes are involved in a number of distinct processes and their alleles can have multiple phenotypic effects. Such alleles are said to be pleiotropic, meaning they have many distinct effects on an organism's phenotype. Not all of the pleiotropic effects of an allele are necessarily of the same type; some traits can be beneficial, others deleterious. A trait that dramatically increases the survival of the young, and so their potential reproductive success, but leads to senility in older adults could well be positively selected for. In this scenario, the senility trait is maladaptive but is not eliminated by selection because it is mechanistically associated with the highly adaptive juvenile survival trait. It is also worth noting that a trait that is advantageous in one environment or situation can be disadvantageous in another. All of which is to say that when thinking about evolutionary mechanisms, do not assume that a particular trait exists independently of other traits or functions in the same way in all environments or even that its presence indicates that it is beneficial.

Questions to answer and ponder:

- Consider this quote from Charles Darwin, “*Natural selection will never produce in a being any structure more injurious than beneficial to that being, for natural selection acts solely by and for the good of each.*” How would you modify it in light of our modern understanding of evolutionary mechanisms?
- Make a model of the factors that would influence a population isolated for 100 generations from its much larger parental population, assuming that it migrated back into its original habitat.

Speciation & extinction

As we have already noted, an important fact that any biological theory has to explain is why there are millions of different types of organisms currently present on Earth. The Theory of Evolution explains this observation through the process of speciation. The basic idea is that populations of organisms can split into distinct groups; over time evolutionary mechanisms acting on these populations will produce distinct types of organisms, that is, different species.⁸⁰ At the same time, we know from the fossil record and from modern experiences that types of organisms can disappear – they can become extinct. So the question is, what leads to the formation of a new species or the disappearance of an existing one?

*So, naturalists observe, a flea has smaller fleas
that on him prey; and these have smaller still
to bite 'em; and so proceed ad infinitum.*

- Jonathan Swift

To answer these questions, we have to consider how populations behave. A population of an organism will typically inhabit a particular geographical region. The size of these regions can range from extending over a continent or more, to a small region, such as a single isolated lake. Moreover, when we consider organisms that reproduce in a sexual manner, that is, that have to cooperate with one another to produce the next generation of organisms, we have to consider how far the organism (or its gametes) can travel. The range of some organisms is quite limited, whereas others can travel significant distances. Another factor we need to consider is how an organism makes its living, that is, where does

⁸⁰ The problem is, of course, more complex and subject with asexual species (such as bacteria), but here a more Linnaean analysis based on the comparison of traits is used. Among these traits are genomic sequence.

it get the food and space it needs to successfully reproduce?

The concept of an organism's **ecological niche**, which is the result of its past evolutionary history, that is, of the past selection pressures acting within a particular environment, combines all of these factors. In a stable environment, and a large enough population, reproductive success will reflect how organisms survive and exploit their ecological niche. Over time, conservative selection will tend to optimize the organism's adaptation to its niche. At the same time, it is possible that different types of organisms will compete for similar resources. This interspecies competition leads to a new form of selective pressure. If individuals of one population can exploit a different set of resources or the same resources differently, these organisms can minimize competition with other species and become more reproductively successful compared to individuals that directly compete with that species. This can lead to a number of outcomes. In one case, one species becomes much better than the other at occupying a particular niche, driving the other to extinction. Alternatively, one species may find a way to occupy a new or related niche, and within that particular niche, it can more effectively compete, so that the two species come to occupy distinct niches. Finally, one of the species may be unable to reproduce successfully in the presence of the other and become (at least) locally extinct. These scenarios are captured in what is known as the competitive exclusion principle or Gause's Law, which states that two species cannot (stably) occupy the same ecological niche - over time either one will leave (or rather be forced out) of the niche, or will evolve to fill a different, often subtly different niche. What is sometimes hard to appreciate is how specific a viable ecological niche can be. For example, consider the situation described by the evolutionary biologist Theodosius Dobzhansky (1900-1975):

Some organisms are amazingly specialized. Perhaps the narrowest ecologic niche of all is that of a species of the fungus family Laboulbeniaceae, which grows exclusively on the rear portion of the elytra (the wing cover) of the beetle *Aphenops cronei*, which is found only in some limestone caves in southern France. Larvae of the fly *Psilopa petrolei* develop in seepages of crude oil in California oilfields; as far as is known they occur nowhere else.

While it is tempting to think of ecological niches in broad terms, the fact is that subtle environmental differences can favor specific traits and specific organisms. If an organism's range is large enough and each individual's range is limited, distinct traits can be prominent in different regions of the species' range. These different subpopulations (sometimes termed subspecies or races) reflect local adaptations. For example, it is thought that human populations migrating out of the equatorial regions of Africa were subject to selection based on exposure to sunlight in part through the role of sunlight in the synthesis of vitamin D.⁸¹ In their original ecological niche, the ancestors of humans were thought to hunt in the open savannah (rather than within forests), and so developed adaptations to control their body temperature - human nakedness is thought to be one such adaptation (although there may be aspects of sexual selection involved as well, discussed in the next chapter). Yet, the absence of a thick coat of hair also allowed direct



⁸¹Genetics of skin color: <http://humanorigins.si.edu/evidence/genetics/skin-color/modern-human-diversity-skin-color>
image sources: <http://hmg.oxfordjournals.org/content/18/R1/R9.full>

exposure to the UV-light from the sun. While UV exposure is critical for the synthesis of vitamin D, too much exposure can lead to skin cancer. Dark skin pigmentation is thought to be an adaptive compromise. As human populations moved away from the equator, the dangers of UV exposure decreased while the need for vitamin D production remained. Under such condition, allelic variation that favored lighter skin pigmentation (but retaining the ability to tan, at least to some extent) appears to have been selected. Genetic analyses of different populations have begun to reveal exactly which mutations, and the alleles they produced, occurred in different human populations as they migrated out of Africa. Of course, with humans the situation has an added level of complexity. For example, the human trait of wearing clothing certainly impacts the pressure of "solar selection."

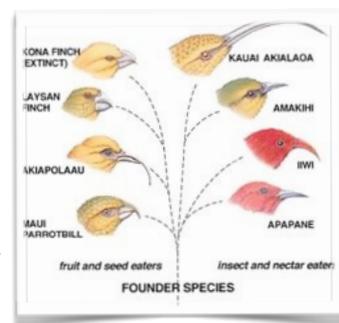
A number of variations can occur over the range of a species. Differences in climatic conditions, pathogens, predators, and prey can all lead to local adaptations, like those associated with human skin color. For example, many species are not continuously fertile and only mate at specific times of the day or year. When the range of a species is large, organisms in geographically and climatically distinct regions may mate at somewhat different times. As long as there is sufficient migration of organisms between regions and the organisms continue to be able to interbreed and to produce fertile offspring, the population remains one species.

Mechanisms of speciation

So now we consider the various mechanisms that can lead to a species giving rise to one or more new species. Remembering that species, at least species that reproduce sexually, are defined by the fact that they can and do interbreed to produce fertile offspring, you might already be able to propose a few plausible scenarios. An important point is that the process of speciation is continuous, there is no magic moment when one species changes into another, rather a new species emerges over time from a pre-existing species. Species are populations of organisms at a moment in time, they are connected to past species and can produce new species.

Perhaps the simplest way that a new species can form is if the original population is physically divided into isolated subpopulations. This is termed allopatric speciation. By isolated, we mean that individuals of the two subpopulations no longer mingle with one another, they are restricted to specific geographical areas. That also means that they no longer breed with one another. If we assume that the environments inhabited by the subpopulations are distinct, that they represent distinct sets of occupied and available ecological niches, distinct climate and geographical features, and distinct predators, prey, and pathogens, then these isolated subpopulations will be subject to different selection pressures, different phenotypes (and the genotypes associated with them) will have differential reproductive success. Assuming the physical separation between the populations is stable, and persists over a significantly long length of time, the populations will diverge. Both selective and non-selective processes will drive this divergence, and will influence by exactly what new mutations arise and give rise to alleles. The end result will be populations adapted to specific ecological niches, which may well be different from the niche of the parental population. For example, it is possible that while the parental population was more a generalist, occupying a broad niche, the subpopulations may be more specialized to a specific niche. Consider the situation with various finches (honeycreepers) found in the

Hawaiian islands.⁸² Derived from an ancestral population, these organisms have adapted to a number of highly specialized niches. These specializations give them a competitive edge in feeding off particular types of flowers [→]. As they specialize, however, they become more dependent upon the continued existence of their host flower or flower type. It is little like a fungus that can only grow on a particular place on a particular type of beetle, as well discussed earlier. We begin to understand why the drive to occupy a particular ecological niche also leads to vulnerability, if the niche disappears for some reason, the species adapted to it may not be able to cope, that is, be able to effectively and competitively exploit the remaining niches, and may become extinct. It is a sobering thought that current estimates are that greater than 98% of all species that have or now live on Earth are extinct, presumably due in large measure in changes in or the disappearance of their niche. You might speculate (and provide a logical argument to support your speculation) as to which of the honeycreepers illustrated above would be most likely to become extinct in response to environmental changes.⁸³ In a complementary way, the migration of organisms into a new environment can produce a range of effects as new competitions for existing ecological niches get resolved. If an organism influences its environment, the effects can be complex. As noted before, a profound and global example is provided by the appearance of photosynthetic organisms that released molecular oxygen (O_2) as a waste product early in the history of life on Earth. Because of its chemical reactivity the accumulation of molecular oxygen led to loss of some ecological niches and the creation of new ones. While dramatic, similar events occur on more modest levels all of the time, particularly in the microbial world. It turns out that extinction is a fact of life.



Gradual or sudden environmental changes, ranging from the activity of the sun, to the drift of continents and the impacts of meteors and comets, leads to the disappearance of existing ecological niches and appearance of new ones. For example, the collision of continents with one another leads to the formation of mountain ranges and regions of intense volcanic activity, both of which can influence climate. There have been periods when Earth appears to have been completely or almost completely frozen over. One such snowball Earth period has been suggested as playing an important role in the emergence of macroscopic multicellular life. These processes continue to be active today, with the Atlantic ocean growing wider and the Pacific ocean shrinking, the splitting of Africa along the Great Rift Valley, and the collision of India with Asia. As continents move and sea levels change, organisms that evolved on one continent may be able to migrate into another. All of these processes combine to lead to extinctions, which open ecological niches for new organisms, and so it goes.

At this point you should be able to understand that evolution never actually stops. Aside from various environmental factors, each species is part of the environment of other species. Changes in one species can have dramatic impacts on others as the selective landscape changes. An obvious example is the interrelationship between predators, pathogens, and prey. Which organisms survive to

⁸² Hawaiian honeycreepers and their tangled evolutionary tree: <http://www.theguardian.com/science/punctuated-equilibrium/2011/nov/02/hawaiian-honeycreepers-tangled-evolutionary-tree>

⁸³ The Perils of Picky Eating: Dietary Breadth Is Related to Extinction Risk in Insectivorous Bats: <http://www.plosone.org/article/info%3Adoi%2F10.1371%2Fjournal.pone.0000672>

reproduce will be determined in large part by their ability to avoid predators or recover from infection. Certain traits may make the prey more or less likely to avoid, elude, repulse, discourage, or escape a predator's attack. As the prey population evolves in response to a specific predator, these changes will impact the predator, which will also have to adapt. This situation is often called the Red Queen hypothesis, and it has been invoked as a major driver for the evolution of sexual reproduction, which we will consider in greater detail in the next chapter (follow the footnote to a video).⁸⁴

As the Red Queen said to Alice ... "Here, you see, it takes all the running you can do to keep in the same place"
-Lewis Carroll, *Through the Looking Glass*

Isolating mechanisms

Think about a population that is on its way to becoming specialized to fill a particular ecological niche. What is the effect of cross breeding with a population that is, perhaps, on an adaptive path to another ecological niche? Most likely the offspring will be poorly adapted for either niche. This leads to a new selective pressure, selection against cross-breeding between individuals of the two populations. Even small changes in a particular trait or behavior can lead to significant changes in mating preferences and outcomes. Consider Darwin's finches or the Hawaiian honeycreepers mentioned previously. A major feature that distinguishes these various types of birds is the size and shapes of their beaks. These adaptations represent both the development of a behavior – that is the preference of birds to seek food from particular sources, for example, particular types of flowers or particular size seeds – and the traits needed to successfully harvest that food source, such as bill shape and size. Clearly the organism has to first display the behavior that makes selection of the physical trait beneficial. This is a type of loop, where behavioral and physical traits are closely linked. You can ask yourself, would a giraffe have a long neck if it did not like (want to) to eat the leaves of tall trees?

Back to finches and honeycreepers. Mate selection in birds is often mediated by song, generally males sing and females respond (or not). As beak size and shape change, so the song produced also changes.⁸⁵ This change is, at least originally, an unselected trait that accompanies the change in beak shape, but it can become useful if females recognize and respond to songs more like their own. This would lead to preferential mating between organisms with the same trait (beak shape). Over time, this preference could evolve into a stronger and stronger preference, until it becomes a reproductive barrier between organisms adapted to different ecological niches. Similarly, imagine that the flowers a particular subpopulation feeds on open and close at different times of the day. This could influence when an organism that feeds on a particular type of flower is sexually receptive. You can probably generate your own scenarios in which one behavioral trait has an influence on reproductive preferences. If a population is isolated from others, such effects may develop but are relatively irrelevant. They become important when two closely-related but phenotypically distinct populations come back into contact. Now matings between individuals in two different populations, sometimes termed hybridization, can lead to offspring poorly adapted to either niche. This creates a selective

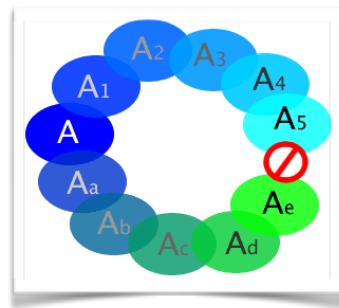
⁸⁴ The Red Queen: http://www.pbs.org/wgbh/evolution/library/01/5/l_015_03.html

⁸⁵ Beaks, Adaptation, and Vocal Evolution in Darwin's Finches: <http://bioscience.oxfordjournals.org/content/54/6/501.short> and Vocal mechanics in Darwin's finches: correlation of beak gape and song frequency: <http://jeb.biologists.org/content/207/4/607.short>

pressure to minimize hybridization. Again, this can arise spontaneously, such as the two populations mate at different times of the day or year or respond to different behavioral queues, such as mating songs. Traits that enhance reproductive success by reducing the chance of detrimental hybridization will be preferentially chosen. The end result is what is known as reproductive isolation.⁸⁶ Once reproductive isolation occurs, what was one species has become two. A number of different mechanisms ranging from the behavioral to the structural and the molecular are involved in generating reproductive isolation. Behaviors may not be “attractive,” genitalia may not fit together, gametes might not survive, or embryos might not be viable - there are many possibilities.

Ring species

Ring species demonstrate a version of allopatric speciation. Imagine populations of the species A. Over the geographic range of A there exist a number of subpopulations. These subpopulations (A_1 to A_5) and (A_a to A_e) have limited regions of overlap with one another but where they overlap they interbreed successfully. But populations A_5 and A_e no longer interbreed successfully – are these populations separate species? In this case, there is no clear cut answer, but it is likely that in the link between the various populations will be broken and one or more species may form in the future. Consider the black bear, *Ursus americanus*. Originally distributed across North America, its distribution is now much more fragmented. Isolated bear populations are free to adapt to their own particular environments and migration between populations is limited. Clearly the environment in Florida is different from that in Mexico, Alaska, or Newfoundland. Different environments will favor different adaptations. If, over time, these populations were to come back into contact with one another, they might or might not be able to interbreed successfully - reproductive isolation may occur and one species might become many.



Sympatric speciation

While the logic and mechanisms of allopatric speciation are relatively easy to grasp (we hope), there is a second type of speciation, known as **sympatric speciation**, which was originally more controversial. It occurs when a single population of organisms splits into two reproductively isolated communities within the same physical area. How could this possibly occur, what would stop the distinct populations from in-breeding and reversing the effects of selection and nascent speciation? Recently a number of plausible mechanisms have been identified. One involves host selection.⁸⁷ In host selection, animals (such as insects) that feed off specific hosts may find themselves reproducing in distinct zones associated with their hosts. For example, organisms that prefer blueberries will mate in a different place, time of day, or time of year than those that prefer raspberries. There are blueberry and raspberry niches. Through a process of disruptive selection (see above), organisms that live primarily on a

⁸⁶ Beak size matters for finches' song: http://news.nationalgeographic.com/news/2004/08/0827_040827_darwins Finch.html

⁸⁷ Sympatric speciation by sexual selection: <http://www.ncbi.nlm.nih.gov/pubmed/10591210?dopt=Abstract&holding=npg>
Sympatric speciation in phytophagous insects: moving beyond controversy? <http://www.ncbi.nlm.nih.gov/pubmed/11729091?dopt=Abstract&holding=npg>

particular plant (or part of a plant) can be subject to different selective pressures, and reproductive isolation will enable the populations to more rapidly adapt. Mutations that reinforce an initial, perhaps weak, mating preference can lead to what known as reproductive isolation - as we will see this is a simple form of sexual selection.⁸⁸ One population has become two distinct, reproductively independent populations, one species as become two.

Questions to answer & ponder:

- Make a model of interactions of how non-adaptive factors could influence species formation.
- Describe the (Darwinian) cycle of selection associated with the development of the giraffe's neck.
- Provide a scenario that would explain why a small population associated with allopatric speciation would either speed evolutionary change or lead to extinction?
- Which comes first, the behavior or the ability to carry out the behavior?
- Make a model of the various effects of isolating mechanisms on allele frequencies between once isolated populations.
- How would you model the process by which an asexual organism would be assigned to a specific species?
- How would you go about determining whether an organism, identified through fossil evidence, was part of a new or a living species?
- How would you determine whether two species are part of the same genus?

Signs of evolution: homology and convergence

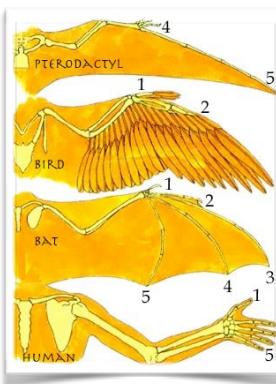
When we compare two different types of organisms we often find traits that are similar. On the basis of evolutionary theory, these traits can arise through either of two processes: the trait could have been present in the ancestral population that gave rise to the two species or the two species could have developed the traits independently. In this latter case, the trait was *not* present in the last common ancestor shared by the organism. Where a trait was present in the ancestral species it is said to be a **homologous** trait. If the trait was not present in the ancestral species but appeared independently within the two lineages, it is known as an **analogous** trait that arose through evolutionary **convergence**.

For example, consider the trait of vitamin C dependence, found in Haplorrhini primates discussed above. Based on a number of lines of evidence, we conclude that the ancestor of all Haplorrhini primates was vitamin C dependent and that vitamin C dependence in Haplorrhini primates is a homologous trait. On the other hand, Guinea pigs (*Cavia porcellus*), which are in the order Rodentia, are also vitamin C dependent, but other rodents are not. It is estimated that the common ancestor of primates and rodents lived more than 80 million years ago, that is, well before the common ancestor of the Halporrhini, and because other rodentia are vitamin C independent, that this common rodent/primate ancestor was itself vitamin C independent. We conclude that vitamin C dependence in Guinea pigs and Halporrhini are analogous traits.

As we look at traits, we have to look carefully, structurally, and more and more frequently in the 21st century, molecularly (genotypically) to determine whether they are homologous or analogous, that is the result of evolutionary convergence. Consider the flying vertebrates. The physics of flight (and many other behaviors that organisms perform) are constant. Organisms of similar size face the same aerodynamic and thermodynamic constraints. In general there are only a limited number of physically

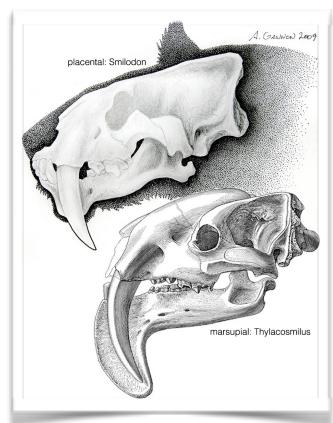
⁸⁸ The sexual selection: <http://www.youtube.com/watch?v=JakdRczkmNo>

workable solutions to deal with these constraints. Under these conditions different populations that are in a position to exploit the benefits of flight will, through the process of variation and selection, end up with structurally similar solutions. This process is known as convergent evolution. Convergent evolution occurs when only certain solutions to a particular problem are evolutionarily accessible.



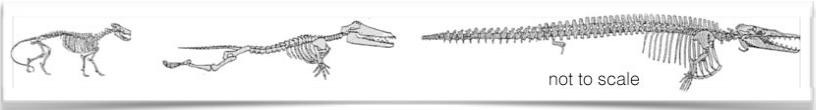
Consider the wing of a pterodactyl, which is an extinct flying reptile, a bird, and a bat, which is a flying mammal. These organisms are all tetrapod (four legged) vertebrates – their common ancestor had a structurally similar forelimb, so their forelimbs are clearly homologous. Therefore evolutionary processes (using the forelimb for flight) began from a similar starting point. But most tetrapod vertebrates do not fly, forelimbs have become adapted to different functions. An analysis of tetrapod vertebrate wings indicates that they took distinctly different approaches to generating wings. In the pterodactyl, the wing membrane is supported by the 5th finger of the forelimb, in the bird by the 2nd finger, and in the bat, by the 3rd, 4th and 5th fingers. The wings of pterodactyls, birds, and bats are clearly analogous structures, while their forelimbs are homologous.

As another example, the use of a dagger is an effective solution to the problem of killing another organism. Variations of this solution have been discovered or invented independently many times, with similar dagger-like teeth evolving independently (that is from ancestors without such teeth) in a wide range of evolutionarily distinct lineages. Consider, for example, the placental mammal Smilodon and the marsupial mammal Thyacosmilus [→]; both have similarly-shaped highly elongated canine teeth. Marsupial and placental mammals diverged from a common ancestor ~160 million years ago and this ancestor appeared to lack such teeth, as do most mammals. While teeth are a homologous feature of Smilodon and Thyacosmilus, elongated dagger-like teeth are analogous structures, the result of convergent evolution for this trait.



The loss of traits

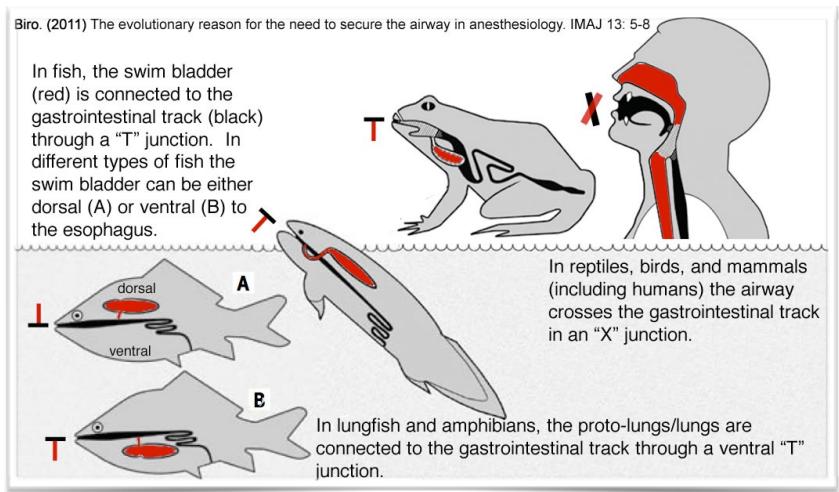
A major challenge when trying to determine the relationship between organisms based on anatomy has been to determine whether similar traits indicate common ancestry, that is whether the trait justifies putting two organisms into the same group, or whether it represents two independent solutions to a common problem, and so is irrelevant when it comes to placing an organism in a classification scheme. The loss of traits can confuse or complicate the positioning of an organism in a classification scheme. As organisms adapt to a specific environment and lifestyle, traits once useful can become irrelevant and may be lost (such as the ability to synthesize ascorbic acid). A classic example is the reduction of hind limbs during the evolution of whales. Another is the common loss of eyes often seen as populations adapt to environments in



which light is absent. The most dramatic case of loss involves organisms that become obligate parasites of other organisms. In many cases, these parasitic organisms become completely dependent on their hosts for many essential functions, and they can become quite simplified even though they are in fact highly evolved. For example, they lose many genes as they become dependent upon the host. The loss of traits can itself be an adaptation if it provides an advantage to organisms living in a particular environment. This fact can make it difficult to determine whether an organism is primitive (that is, retains ancestral features) or highly evolved.

Signs of evolutionary history

Evolution is an ongoing experiment in which random mutations are selected based on the effects of the resulting phenotypes on reproductive success. As we have discussed, various non-adaptive processes are also involved, which can impact evolutionary trajectories. The end result is that adaptations are based on past selective pressures and i) are rarely perfect and ii) may actually be outdated, if the environment the organisms live in has changed. One needs to keep this in mind when one considers the differences associated with living in a pre-technological world on the African savannah in small groups and living in New York City. In any case, evolution is not a designed process that reflects a predetermined goal but involves responses to current constraints and opportunities - it is a type of tinkering in which selective and non-selective processes interact with pre-existing organismic behaviors and structures and is constrained by cost and benefits associated with various traits and their effects on reproductive success.⁸⁹ What evolution can produce depends on the alleles present in the population and the current form of the organism. Not all desirable phenotypes (that is, leading to improved reproductive success) may be accessible from a particular genotype, and even if they are, the cost of attaining a particular adaptation, no matter how desirable to an individual, may not be repaid by the reproductive advantage it provides within a population. As an example, our ability to choke on food could be considered a serious design flaw, but it is the result of the evolutionary path that produced us (and other four-legged creatures), a path that led to the crossing of our upper airway (leading to the lungs) and our pharynx (leading to our gastrointestinal system). That is why food can lodge in the airway, causing choking or death. It is possible that the costs of a particular "imperfect" evolutionary design are offset by other advantages. For example, the small but significant possibility of death by choking may, in an evolutionary sense, be worth the ability to make more complex sounds (speech) involved in social communication⁹⁰.



⁸⁹ Evolutionary tinkering: <http://virtuallaboratory.colorado.edu/Biofundamentals/lectureNotes/Readings/EvolutionTinkering.pdf>

⁹⁰ How the Hyoid Bone Changed History: <http://www.livescience.com/7468-hyoid-bone-changed-history.html>

As a general rule, evolutionary processes generate structures and behaviors that are as good as they need to be for an organism to effectively exploit a specific set of environmental resources and to compete effectively with its neighbors, that is, to successfully occupy its niche. If being better than good enough does not enhance reproductive success, it cannot be selected for (at least via natural selection) and variations in that direction will be lost, particularly if they come at the expense of other important processes or abilities. In this context it is worth noting that we are always dealing with an organism throughout its life cycle. Different traits can have different values at different developmental stages. Being cute can have important survival benefits for a baby but be less useful in a corporate board room (although perhaps that is debatable). A trait that improves survival during early embryonic development or enhances reproductive success as a young adult can be selected for even, if it produces negative effects on older individuals. Moreover, since the probability of being dead (and so no longer reproductively active) increases with age, selection for traits that benefit the old will inevitably be weaker than selection for traits that benefit the young (although this trend can be modified in organisms in which the presence of the old can increase the survival and reproductive success of the young, for example through teaching and babysitting). Of course survival and fertility curves are also changing in response to changing environmental factors, which change selective pressures. In fact, lifespan itself is a selected trait, since it is the population not the individual that evolves.⁹¹

We see the evidence for various compromises involved in evolutionary processes all around us. It explains the limitations of our senses, as well as our tendency to get backaches, need hip-replacements, and our susceptibility to diseases and aging.⁹² For example, the design of our eyes leaves a blind spot in the retina. Complex eyes have arisen a number of times during the history of life, apparently independently, and not all have a blind spot. We have adapted to this retinal blind spot through the use of saccadic movements because this is an evolutionarily easier fix to the problem than rebuilding the eye from scratch (which is essentially impossible). An "intelligently designed" human eye would presumably not have such an obvious design flaw, but because of the evolutionary path that led to the vertebrate eye, it may simply have been impossible to back up and fix this flaw. More to the point, since the vertebrate eye works very well, there is no reward in terms in reproductive success associated with fixing this flaw. This is a general rule: current organisms work, at least in the environment that shaped their evolution. Over time, organisms that diverge from the current optimal, however imperfect, solution will be at a selective disadvantage. The current vertebrate eye is maintained by conservative selection (as previously described).

Homologies provide evidence for a common ancestor

The more details two structures share, the more likely they are to be homologous. In the 21st century, molecular methods, particularly complete genome sequencing, have made it possible to treat gene sequences and genomic organization as traits that can be compared. Detailed analyses of many

⁹¹ Methusaleh's Zoo: how nature provides us with clues for extending human health span: <http://www.ncbi.nlm.nih.gov/pubmed/19962715> and Why Men Matter: Mating Patterns Drive Evolution of Human Lifespan: <http://www.plosone.org/article/info%3Adoi%2F10.1371%2Fjournal.pone.0000785>

⁹² <http://www.pbs.org/wgbh/nova/evolution/what-evidence-suggests.html>

different types of organisms reveals the presence of a common molecular signature that strongly suggests that all living organisms share a large numbers of homologies, which implies that they are closely related; that is, that they share a common ancestor. These universal homologies range from the basic structure of cells to the molecular machinery involved in energy capture and transduction, information storage and utilization. All organisms

- use double-stranded DNA as their genetic material;
- use the same molecular systems, transcription and translation, to access the information stored in DNA;
- use a common genetic code, with few variations, to specify the sequence of polypeptides (proteins);
- use ribosomes to translate the information stored in messenger RNAs into polypeptides; and
- share common enzymatic (metabolic) pathways.

Anti-evolution arguments

The theory of evolution has been controversial since its inception largely because it deals with issues of human origins and behavior, our place in the Universe, and life and its meaning. Its implications can be quite disconcerting, but many observations support the fact that organisms on Earth are the product of evolutionary processes and these processes are consistent with what we know about how matter and energy behave. As we characterize the genomes of diverse organisms, we see evidence for the interrelationships, observations that non-scientific (creationist) models would never have predicted and do not explain. That evolutionary mechanisms have generated the diversity of life and that all organisms found on Earth share a common ancestor is as well-established as the atomic structure of matter, the movement of Earth around the Sun, and the solar system around the Milky Way galaxy. The implications of evolutionary processes remain controversial, but not evolution itself.

Scientific knowledge is a body of knowledge of varying degrees of certainty—some most unsure, some nearly sure, but none absolutely certain ... Now we scientists are used to this, and we take it for granted that it is perfectly consistent to be unsure, that it is possible to live and not know.
- Richard Feynman.

...it is always advisable to perceive clearly our ignorance.— Charles Darwin.

Questions to answer & to ponder:

- Justify the assumption that the mutations in Haplorrhini primates and guinea pigs were independent events?
- What typical mammalian traits have whales lost during their evolution?
- Model the factors that would influence the evolution whales back to a terrestrial lifestyle.
- Generate a model by which you could classify a trait as primitive or derived?
- How does the loss of a trait or convergent evolution complicate lineage analysis?
- If all organisms are descended from a common ancestor, what can we say about the diversity of pre-biogenic systems that existed before that ancestor?
- What conditions can lead to a complex organism becoming simpler?
- If the environment were constant, would extinction or evolution occur?
- In what ways can an organism direct its evolution?
- What are the benefits and drawbacks of a high degree of specialization for a species?
- How might the types of changes that lead to reproductive isolation be beneficial (overall) even if they were mildly deleterious?

- How do we know that a species is a species if we do not directly observe whether it can interbreed with other organisms?
- Consider Hawaiian honey creepers; which is most likely to become extinct and why?
- What testable predictions emerge from "intelligent design creationism"?
- Under what environmental conditions would a generalist be favored over a specialist?
- What benefit(s) could be linked to the loss of eyesight or other "advanced" traits?