3.16: Genetic drift

Genetic drift is an evolutionary phenomenon that is impossible in a strict Hardy-Weinberg world, yet it 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 (meaning “wet noses”). The Strepsirrhini include 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, suppuring wounds, loss of teeth, jaundice, fever, neuropathy, and death.”

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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 gene is the root cause of vitamin C dependence in Haplorhini by putting a working copy of the gulo1 gene, for example derived from a 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 a preventative treatment for scurvy was discovered94), no new, functional gulo1 allele has appeared. Organisms do not always produce the alleles they need or that might be beneficial, such alleles must be selected from alleles already present in the population or that appear through mutation. In some cases, however, there may be no molecule pathway that can generate such an allele.

The mutant gulo1 allele appears to have become fixed in the ancestral population that gave rise to the Haplorhini ~40 million years ago. So the question is, how did we (that is our ancestors) come to loose 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 (that is, there was little or no selective pressure for the ability to make vitamin C). 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 functional 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, it is due to random effects on organismic survival, which can, in practice be difficult to distinguish from selective effects. In contrast, drift is unavoidable in sexually reproducing organisms. 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 the alleles present within the population, any particular gamete contains only a single 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. Moreover, not all gametes (something particularly true of sperm) become part of the next generation. 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 with respect to one another, 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. Of course new alleles continue to arise by mutation, but they are originally very infrequent, just one in the entire population, so unless they are strongly selected for they are likely to disappear from the population. Drift can lead to some weird outcomes. 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 BBY 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.
This use of pre-existing variation, rather than the idea that an organism invents variations in its genome as they are required, was a key point in Darwin's view of evolutionary processes. The organism cannot create the alleles it might need, nor are there any known processes 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 to work with. 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:

• 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)?

References

93 One amazing fact is that it took various navies the deaths of thousands of sailors to understand the nutritional challenges of vitamin C. https://en.wikipedia.org/?title=James_Lind

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

95 Of course, if the population is small, instead of disappearing, they could become fixed, just through genetic drift - Try the genetic drift applet (Genetic drift: http://darwin.eeb.uconn.edu/simulati...1.0/drift.html )and look for examples where an allele almost disappears and then becomes fixed; it does happen.

96 The exception involves the transfer of genes from organism to organism, a process known as horizontal gene transfer, which we will come to.

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