Mutations Are the Raw Materials of Evolution

By: Joel L. Carlin (*Department of Biology, Gustavus Adolphus*)

A mutation is a change in the sequence of an organism's DNA. Mutations can be caused by high-energy sources such as radiation or by chemicals in the environment. They can also appear spontaneously during the replication of DNA. Mutations generally fall into two types: point mutations and chromosomal aberrations. In point mutations, one base pair is changed. The human genome, for example, contains over 3.1 billion bases of DNA, and each base must be faithfully replicated for cell division to occur. Mistakes, although surprisingly rare, do happen. About one in every 10^{10} (10,000,000,000) base pair is changed. The most common type of mistake is a point substitution. More uncommon is the failure to copy one of the bases (deletion), the making of two copies for a single base (point duplication) or the addition of a new base or even several bases (insertion). Chromosomal aberrations are larger-scale mutations that can occur during meiosis in unequal crossing over events, slippage during DNA recombination or due to the activities of transposable events. Genes and even whole chromosomes can be substituted, duplicated, or deleted due to these errors (Figure 1).

Figure 1: Mutations in DNA sequence from seven related species of tropical fishes (data are from intron 6 of *LDH-A* gene sequenced by the author from epinepheline serranids) At top are the original sequences, at bottom are the sequences adjusted to vertically align similar DNA bases. Point substitutions are in red, and the yellow box with dashes indicates a deletion of 12 bases.

Mutations can have a range of effects. They can often be harmful. Others have little or no detrimental effect. And sometimes, although very rarely, the change in DNA sequence may even turn out to be beneficial to the organism. A mutation that occurs in body cells that are not passed along to subsequent generations is a somatic mutation. A mutation that occurs in a gamete or in a cell that gives rise to gametes are special because they impact the next generation and may not affect the adult at all. Such changes are called germ-line mutations because they occur in a cell used in reproduction (germ cell), giving the change a chance to become more numerous over time. If the mutation has a deleterious effect on the phenotype of the offspring, the mutation is referred to as a genetic disorder. Alternately, if the mutation has a positive effect on the fitness of the offspring, it is called an adaptation. Thus, all mutations that affect the fitness of future generations are agents of evolution.

Mutations are essential to evolution. Every genetic feature in every organism was, initially, the result of a mutation. The new genetic variant (allele) spreads via reproduction, and differential reproduction is a defining aspect of evolution. It is easy to understand how a mutation that allows an organism to feed, grow or reproduce more effectively could cause the mutant allele to become more abundant over time. Soon the population may be quite ecologically and/or physiologically different from the original population that lacked the adaptation. Even deleterious mutations can cause evolutionary change, especially in small populations, by removing individuals that might be carrying adaptive alleles at other genes.

Figure 2: The history of the gray treefrog, *Hyla versicolor*, is an example of mutation and its potential effects. When an ancestral *Hyla chrysocelis* gray treefrog failed to sort its 24 chromosomes during meiosis, the result was *H. versicolor*. This treefrog is identical in size, shape and color to *H. chrysocelis* but has 48 chromosomes and a mating call that is different from the original *H. chrysocelis*. © 2011 Center for Invasive Species and Ecosystem Health Courtesy of David Cappaert, Michigan State

University. All rights reserved.

Most mutations occur at single points in a gene, changing perhaps a single protein, and thus could appear unimportant. For instance, genes control the structure and effectiveness of digestive enzymes in your (and all other vertebrate) salivary glands. At first glance, mutations to salivary enzymes might appear to have little potential for impacting survival. Yet it is precisely the accumulation of slight mutations to saliva that is responsible for snake venom and therefore much of snake evolution. Natural selection in some ancestral snakes has favored enzymes with increasingly more aggressive properties, but the mutations themselves have been random, creating different venoms in different groups of snakes. Snake venoms are actually a cocktail of different proteins with different effects, so genetically related species have a different mixture from other venomous snake families. The ancestors of sea snakes, coral snakes, and cobras (family Elapidae) evolved venom that attacks the nervous system while the venom of vipers (family Viperidae; including rattlesnakes and the bushmaster) acts upon the cardiovascular system. Both families have many different species that inherited a slight advantage in venom power from their ancestors, and as mutations accumulate the diversity of venoms and diversity of species increased over time.

Although the history of many species have been affected by the gradual accumulation of tiny point mutations, sometimes evolution works much more quickly. Several types of organisms have an ancestor that failed to undergo meiosis correctly prior to sexual reproduction, resulting in a total duplication of every chromosome pair. Such a process created an "instant speciation" event in the gray treefrog of North America (Figure 2). The consequence of doubling the genome size in plants is often abnormally large seeds or fruits, a trait that can be of distinct advantage if you are a flowering plant! Most cereals that humans eat have enormous seeds compared to other grasses, and this is often due to the genomic duplications that occurred in the ancestors of modern rice and wheat and, because the mistake occurred in reproductive organs, was successfully passed on to future generations. Humans themselves have mimicked this process by interbreeding individual plants with the largest fruits and seeds in the process of artificial selection, creating many of our modern agricultural crop strains. The idea of evolution by natural selection, first described by Charles Darwin and Alfred Russell Wallace, requires differential survival due to some individuals having greater evolutionary fitness. Whether that fitness is affected by genetic disorders, venomous saliva or enlarged offspring, heritable variation can only arise by mutation. Evolution is simply not possible without random genetic change for its raw material.