Laboratory 1 – Introduction

Prepared by: J. Nunez, D. Crawford, and M. Oleksiak

**Introduction**

The genetic material of a cell is deoxyribonucleic acid or DNA. DNA is made up of chemical units known as bases (Adenine, Cytosine, Guanine, Thymine). Triplets of DNA bases code for either an amino acid or for a stop, and polymers of amino acids make up proteins. RNA is intermediate between DNA and amino acids. Thus, DNA is transcribed into RNA, and RNA is then translated into a string of amino acids, which make up a protein. DNA --> RNA --> protein is known as the central dogma of biology. Importantly, this flow of information is in only one direction. One cannot go from protein back to DNA.

Sequences of DNA associated with a particular function are known as genes. These functions can be anything from coding for a protein to regulating the expression of other genes. The combination of all the genes and other genetic material of an organism is known as its genome, and it is identical through all of its cells. However, not all genes are expressed equally throughout the body an organism. For example, your skin will express melanin in response to sunlight while your liver will express a xenobiotic metabolizing enzyme to metabolize the caffeine in your morning coffee.

Despite several cellular mechanisms to ensure faithful genome replication, mistakes happen. From ionizing radiation to simple errors in the replication machinery, DNA is constantly changing or mutating. While some mutations are harmful to the organism, others are neutral or beneficial. Some mutations appear in single cells and eventually disappear without trace, others stay in the genome, and some even create new variants of genes that may or may not be fixed in the gene pool of a population. Regardless of the nature of mutations, these genetic changes are the raw material for evolution. On a large scale, DNA variation over 3.5 billion years is responsible for the diversity of species seen on our planet. On a more local scale, DNA variation is the reason we are different from each other, *i.e.,* why we respond differently to the similar medical treatments, *etc.*

Many animals are diploid; they have two sets of chromosomes in their nuclear genome. Commonly, one set of chromosomes comes from the father and the other comes from the mother. Both chromosomes have genes with identical functions located at similar positions or locations in the genome. These positions are known as loci (singular: locus). If the genes found at a locus at each of the chromosomes are identical, the organism is said to be homozygous for said locus. However, if there are different versions of a gene at any particular locus, organism are said to be heterozygous for that locus, and the variants of the gene in questions are known as alleles. The nuclear genome, however, is not the only genome contained in most eukaryotic creatures. Eukaryotic cells contain extra-nuclear genomes in organelles such as the mitochondrion (plural: mitochondria) or chloroplasts. In contrast to the nuclear genome, the mitochondrial genome is one circular chromosome (inherited from the mother in many eukaryotes). In animals, the mitochondrial genome, contains 37 genes all involved in the biochemical process of energy generation. One of the most fundamental differences between the nuclear DNA (gDNA) and the mitochondrial DNA (mtDNA) is recombination. Recombination is the shuffling of genetic material that creates new combinations of genes. This normally takes place between the two sets of chromosomes of an organism’s nuclear DNA during replication. However, having only one chromosome, mtDNA is incapable of recombination. As a result, all genes in the mitochondria are inherited as one consolidated bundle. This is known as a haplotype.

One of the central goals of evolutionary biology is to understand the forces that drive genetic variation. This is, to understand the forces that define whether or not a mutation or an allele becomes common (increases in frequency) or becomes rare (decreases frequency) in the genomes of the members of a population, *i.e.* a gene pool. Full discussions on the forces of genetic variation are outside the scope of this handout; however, there are three important concepts that help understand these phenomena.

* Natural Selection: There are many types of selection, however, the two main forms are: (1) Positive selection occurs when certain traits (encoded by certain alleles) confer certain individuals with higher reproductive and survivability success, and as a result, that trait becomes more common in a population. (2) Negative selection occurs when certain alleles are purged out of the populations because they code for traits that are unviable for the survivability of an organism in a given environment. Selection is a dynamic process and depends on a complex relationship between genes and the environment.
* Neutral Drift: Variation in DNA sequences can occur due to random neutral effects. In fact, contrary to common belief, most variation between DNA sequences is not due to selection, but rather is due to neutral drift. Neutral drift refers to alleles that become more or less common in a population due to neutral (random) events such as random sampling. For instance, imagine a very fit organism that does not to reproduce or that dies at a young age of accidental causes. Said individual was otherwise favored by natural selection to spread his/her genes but for neutral cases its genes got expelled from the gene pool. Random events can have a large impact on the gene pool of a population (think of a hurricane wiping out 90% of a population).
* Migration and Isolation by distance (IBD): Some organisms can travel long distances seeking food and mates. Inversely, other organisms are limited in the moving capabilities or have high fidelity for certain reproduction sites. These limitations or behaviors have a direct impact in the genetic diversity of species. For instance, species with high migratory behavior tend to be different from those with low migratory behavior or high site fidelity. If two populations are separated by a long distance but their members are able to migrate between the populations, they will establish patterns of gene flow leading to considerably higher level of genetic similarity among populations. Conversely, if there is a physical barrier impeding migration or organisms cannot migrate between populations, the gene flow will be halted. As a result, populations may begin to display genetic divergence (either due to selection or drift). Divergence may lead to speciation over time.