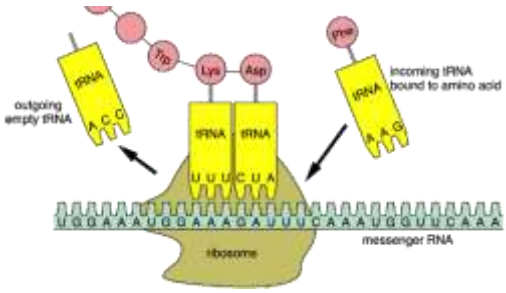
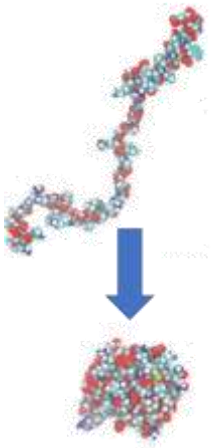
Mutations & Change Unit Summary



Each organism has a unique combination of genes in their DNA. These genes code for the assembly of proteins. Proteins determine the unique characteristics of each organism. Proteins are assembled from amino acids delivered to the ribosome by tRNA. The order of amino acids in a protein is determined by the order of bases in the mRNA copy of a gene. These bases are read codon by codon (group of 3 bases); each codon codes for a specific amino acid.

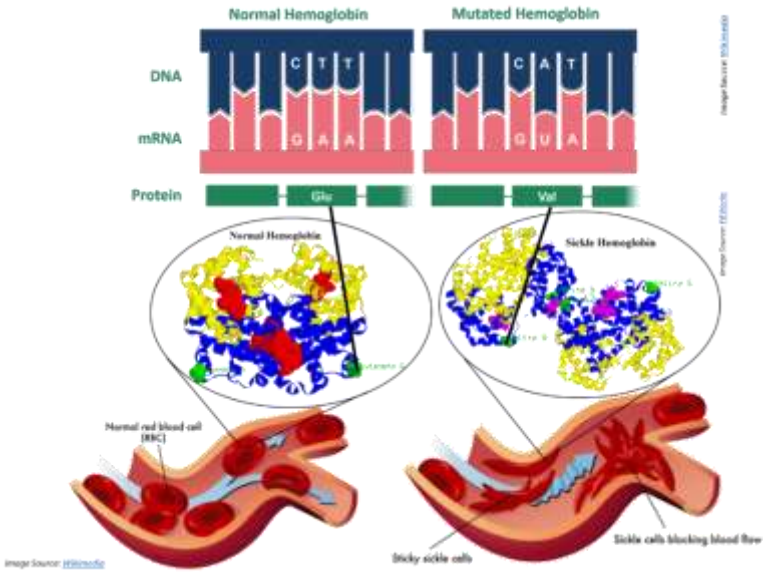
Shape & Function of Proteins



Chains of amino acids do not stay in a straight line as they leave the ribosome. Instead, they will fold into a compact blob; eventually they form a specific shape. The shape of a protein is what primarily determines its function. The properties of the amino acids in a protein determine the shape of the protein. These properties include whether individual amino acids are attracted or repelled by water, and whether they are attracted or repelled by neighboring amino acids.

Amino acids are either attracted to water (<u>hydrophilic</u>) or are repelled by water (<u>hydrophobic</u>). Hydrophilic amino acids will move to the outside of a protein to be closer to water. Hydrophobic amino acids will move inside to the center of a protein to avoid water.

Some amino acids are positively or negatively charged; others are neutral. Oppositely charged amino acids are attracted to each other; similarly charged amino acids are repelled by each other.



In addition, one specific type of amino acid, called <u>cysteine</u>, forms special bonds with other cysteine molecules. Two cysteine amino acids will move the whole chain of amino acids to bond together.

The function of a protein depends on whether a chain of amino acids folds into a correct shape as a result of these three properties. For example, hemoglobin is the protein that binds oxygen molecules on a red blood cell. Hemoglobin must form a ring shape to bind to oxygen.

A mutation in the gene for the hemoglobin protein changes a single amino acid in that sequence. This causes the entire amino acid chain to fold into a different shape, impairing its function. This results in a disease called sickle cell anemia, causing tissue damage and reduced blood flow.

Mutations

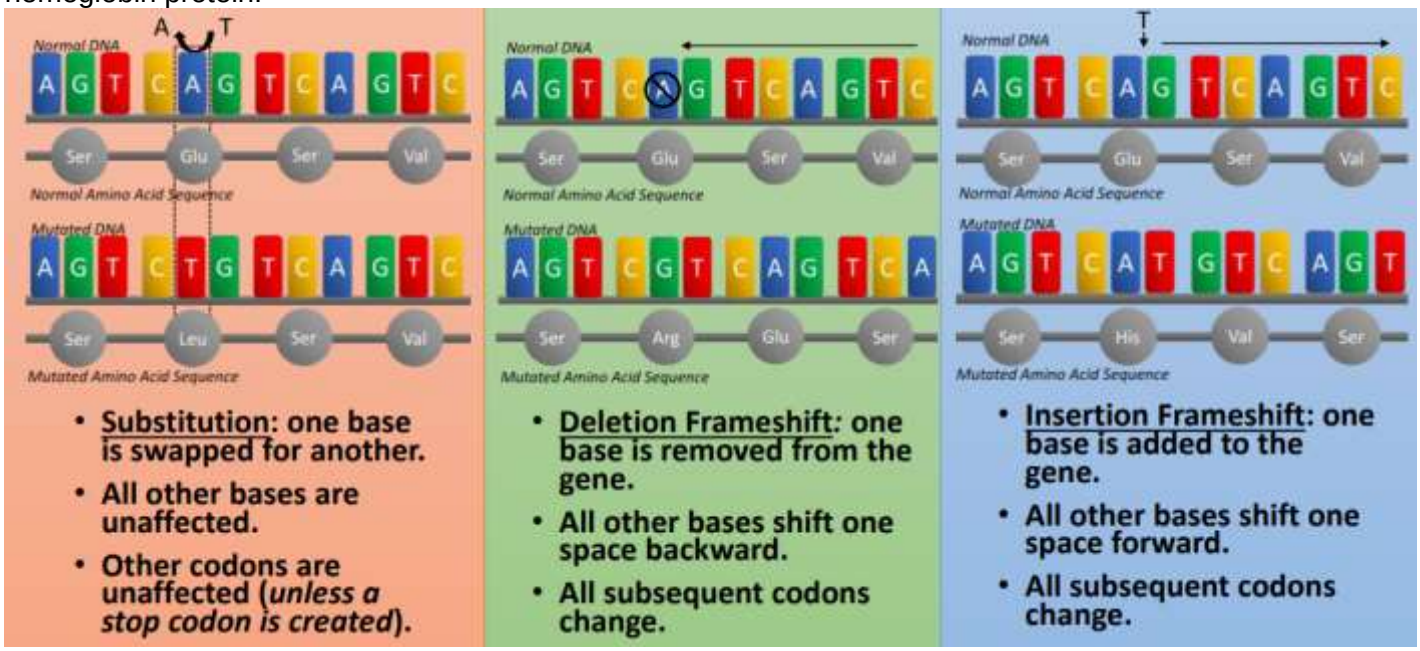
<u>Mutations</u> occur when DNA is changed. This can involve adding, deleting, or changing a base. Mutations can also occur when entire chromosomes change (such as when a chromosome is added or lost). Mutations can be categorized by when they occur (acquired or hereditary) or by what is changed (substitution or frameshift).

<u>Acquired mutations</u> occur sometime after an organism's life has begun. These mutations are only found in some cells, and usually cannot be passed on. In comparison, <u>hereditary mutations</u> occur prior to the start of an organism's life. These mutations are found in every cell of that organism and can be passed on to offspring.

Acquired and hereditary mutations have multiple causes. For example, when DNA polymerase creates a duplicate copy of DNA prior to mitosis or meiosis, it can make mistakes. As a result, the copy of the DNA will have a slightly different order of bases. Mutagens can also change DNA. Mutagens are environmental factors that cause mutations. Examples include UV radiation from the sun, cigarette smoke, and X-rays.

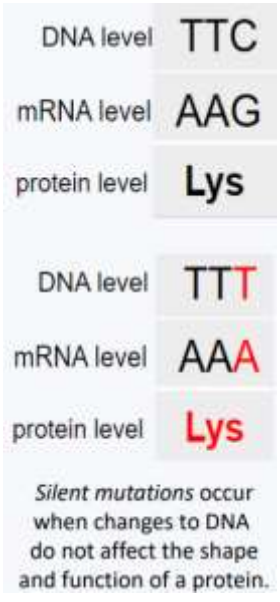
Finally, crossing over during meiosis can also cause mutations. Crossing over occurs when chromosomes exchange segments. Sometimes genes lose or gain bases because of crossing over. Because meiosis forms sperm and egg cells that create new offspring, mutations from crossing over are always hereditary.

Mutations can change DNA in multiple ways. If one base is replaced by another base, this is called a substitution mutation. In these mutations, usually only one codon is changed. For example, sickle cell anemia can occur if a T is changed to an A. This changes one amino acid, changing the shape of the hemoglobin protein.



Alternatively, if a base is inserted or deleted, this is known as a frameshift mutation. Frameshift mutations tend to have a bigger impact on protein assembly because they affect multiple codons and amino acids after the mutation. For example, double-muscling in some cattle is caused by the deletion of 11 bases in the gene for the myostatin protein. This affects most of the protein and changes all codons after the mutation.

Finally, sometimes mutations affect multiple genes at the same time. These are known as chromosomal mutations because they involve changes in the structure of a chromosome. For example, Down syndrome results from an extra copy of the 21st chromosome. The impact of these kinds of mutations varies and depends on how many genes are affected, whether gene function is interrupted, and whether any genes are gained or lost.



Natural Selection & Evolution

Whether changes from mutations are harmful, neutral, or beneficial depends on an organism's surrounding environment. Some mutations have no impact on the organism; these are known as silent mutations. Conversely, genetic diseases result from harmful mutations that disrupt bodily function.

Traits from mutations that improve an organism's ability to survive and reproduce are called adaptations. Natural selection is the process that determines whether changes from mutations increase or decrease an organism's capacity for survival and reproduction in response to their surroundings.

For example, a mutation in the *BMP12* gene caused vultures and ostriches to lose their head feathers. This was beneficial to each species because of their environmental conditions (it reduced disease among vultures and kept ostriches cooler). However, this same mutation would not beneficial for birds like penguins who depend on their head feathers for warmth. The *BMP12* mutation is not inherently helpful or harmful – environmental factors drive the natural selection that determines whether or not the mutation is helpful and becomes more common in a species.

A lack of feathers on vultures and ostriches provides benefits because of their unique environmental conditions.

Species can also change because of artificial selection. This occurs when humans select for species that provide traits that are useful for human needs (but are not necessarily helpful for their own survival). For example, human intervention changed teosinte into corn. While corn provides more benefits for human needs, it cannot survive without humans.

If enough changes occur as a result of natural selection, a group of organisms can form a new species (organisms with similar traits that can produce fertile offspring). Changes to a species over time due to natural selection is known as evolution. The pace at which a species evolves depends on four factors:

- **Sexual Reproduction**: Mating increases genetic diversity and can result in new combinations of genes and physical traits.
- **Heritable Genetic Variation**: Hereditary mutations result in new varieties of genes and physical traits that can be passed on.
- **Competition**: Competition for food, mates, survival determines whether a set of traits is advantageous for an organism in that environment.
- **Differences in Reproduction & Survival Rates**: Organisms with advantageous traits are more likely to survive and reproduce, increasing the prevalence of those traits in that environment.

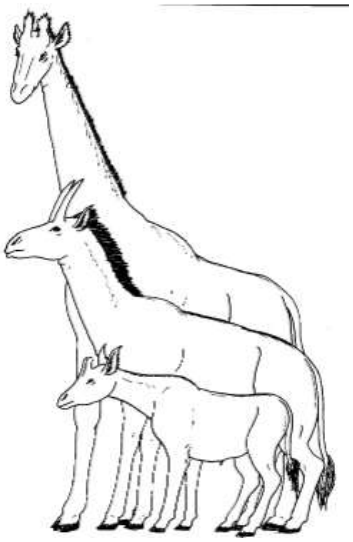
Rates of evolution can vary depending on these four factors. For example, sharks have very little competition for food and generally do not have predators that reduce their population. As a result, they have had very little change over millions of years. Alternatively, whales have had rapid evolution because they have more competition for food and can be prey for predators like sharks. Beneficial mutations had more of an impact on their rates of survival and reproduction.

There is a wide array of evidence to support the idea that natural selection drives changes that enable species to evolve over time. This evidence includes:

- **Homologous structures**: shared anatomical features across multiple species (*e.g., humans, whales, birds, & dogs all contain the same kinds of bones in their limbs due to a common ancestor*).
- **Analogous structures**: body parts with different structure but shared function (*e.g., both dolphins and sharks have dorsal fins that emerged from different random mutations in similar environments*).
- **Vestigial structures**: anatomical features that lost their original size & function (*e.g., whales still have hip bones; humans still have an appendix).*
- **DNA**: species that share recent genetic ancestors also share more similar genes.
- **Fossils**: comparing species' fossils at different points demonstrates how they have changed and evolved over time.
- **Measurable evolution** occurs in some species (*e.g., anoles, bacteria, etc.*). Evolution in these species can be measured by scientists.

Bacteria growing in this petri dish are resistant to most of the antibiotics in the white disks.

Hereditary mutations are needed for evolution to occur. These mutations occur randomly and independently from the environment. For example, a giraffe did not acquire a long neck because there were tall trees in its environment. Instead, a random mutation provided some giraffes with longer necks, and these giraffes were more likely to survive and reproduce compared to other giraffes without the mutation. Species don't change because of their environment – they change because of random mutations that happen to provide benefits in that environment.



Vocabulary

Acquired mutations occur sometime during the life of an organism and cannot be passed on to offspring.
Adaptations: traits from mutations that improve an organism's ability to survive and reproduce.
Analogous structures: body parts w/ different structure but shared function.
Artificial selection is the process in which organisms are selected for genetic traits that benefit human needs.
Chromosomal mutations affect multiple genes and involve changes in the structure of a chromosome.
Cysteine: an amino acid that forms unique bonds with other cysteines.
Domestication: when humans select for beneficial genetic changes to the extent that the organism becomes physiologically different from its wild counterparts.
Evolution: changes to a species over time due to natural selection.
Fossils: remnants of prehistoric organisms that have been preserved in the earth's crust.
Frameshift mutation: a mutation that changes every codon that occurs after the mutation due to the insertion or deletion of a base.
Hereditary mutations are those that can be passed on to subsequent generations and are present throughout the life of an organism.
Homologous structures: shared anatomical features across multiple species.
Hydrophilic: attracted to water.
Hydrophobic: repelled by water.
Mutagens: environmental factors that cause mutations.
Mutation: when DNA undergoes a permanent but unintentional change through the loss, addition, or switching of at least one base.
Natural selection: the process that determines whether changes from mutations increase or decrease an organism's capacity for survival and reproduction in response to their surroundings.
Silent mutations do not affect the shape and function of a protein.
Species: organisms with similar traits that can produce fertile offspring.
Substitution mutation: a mutation that occurs if one base is replaced by another base.
Vestigial structures: anatomical features that lost their original size & function.