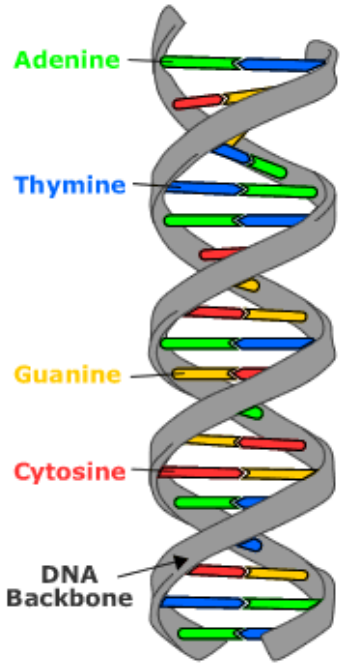
Name: _____ Tribe:_____ Date: _____

DNA: The molecular basis of mutations

Since mutations are simply changes in DNA, in order to understand how mutations work, you need to understand how DNA does its job. Your DNA contains a set of instructions for "building" a human. These instructions are inscribed in the structure of the DNA molecule through a genetic code. It works like this:



DNA is made of a long sequence of smaller units strung together. There are four basis: A, T, G, and C. These letters represent: adenine, thymine, guanine, and cytosine.

The sequence of these bases encodes instructions. Some parts of your DNA are control centers for turning genes on and off, some parts have no function, and some parts have a function that we don't understand yet. Other parts of your DNA are genes that carry the instructions for making proteins — which are long chains of amino acids. These proteins help build an organism.

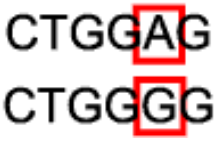
Protein-coding DNA can be divided into codons — sets of three bases that specify an amino acid or signal the end of the protein. Codons are identified by the bases that make them up — in the example at right, GCA, for guanine, cytosine, and adenine. The cellular machinery uses these instructions to assemble a string of corresponding amino acids (one amino acid for each three bases) that form a protein. The amino acid that corresponds to "GCA" is called alanine; there are twenty different amino acids synthesized this way in humans. "Stop" codons signify the end of the newly built protein.

After the protein is built based on the sequence of bases in the gene, the completed protein is released to do its job in the cell.

Types of mutations

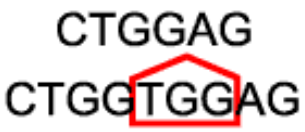
There are many different ways that DNA can be changed, resulting in different types of mutation. Here is a quick summary of a few of these:

Substitution Mutation
A substitution is a mutation that exchanges one base for another (i.e., a change in a single "chemical letter" such as switching an A to a G). Such a substitution could:



1. Change a codon to one that encodes a different amino acid and cause a small change in the protein that is produced. For example, sickle cell anemia (blood disorder) is caused by a substitution in the beta-hemoglobin gene, which alters a single amino acid in the protein that is produced.
2. Change a codon to one that encodes the same amino acid and causes no change in the protein produced. These are called silent mutations.
3. Change an amino-acid-coding codon to a single "stop" codon and cause an incomplete protein. This can have serious effects since the incomplete protein probably won't function.

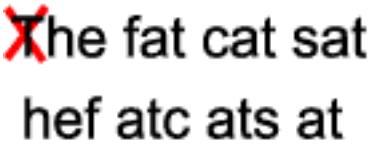
Insertion Mutation
Insertions are mutations in which extra base pairs are inserted into a new place in the DNA.



Deletion Mutation
Deletions are mutations in which a section of DNA is lost, or deleted.

Frameshift
Since protein-coding DNA is divided into codons three bases long, insertions and deletions can alter a gene so that its message is no longer correctly coded. These changes are called frameshifts.

For example, consider the sentence, "The fat cat sat." Each word represents a codon. If we delete the first letter and code the sentence in the same way, it doesn't make sense.



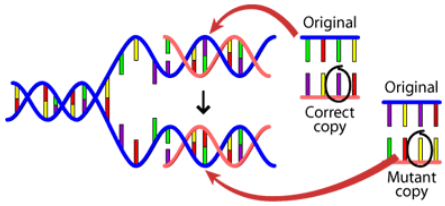
In frameshifts, a similar error occurs at the DNA level, causing the codons to be coded incorrectly. This usually generates proteins that are as useless as "hef atc ats at" is uninformative.

There are other types of mutations as well, but this short list should give you an idea of the possibilities.

The causes of mutations

Mutations happen for several reasons.

1. **DNA fails to copy accurately**
   Most of the mutations that we think matter to evolution are "naturally-occurring." For example, when a cell divides, it makes a copy of its DNA — and sometimes the copy is not quite perfect. That small difference from the original DNA sequence is a mutation.



2. **2. External influences can create mutations**
   Mutations can also be caused by exposure to specific chemicals or radiation. These agents cause the DNA to break down. This is not necessarily unnatural — even in the most isolated and pristine environments, DNA breaks down. Nevertheless, when the cell repairs the DNA, it might not do a perfect job of the repair. So the cell would end up with DNA slightly different than the original DNA and hence, a mutation.

The effects of mutations

Since all cells in our body contain DNA, there are lots of places for mutations to occur; however, some mutations cannot be passed on to offspring and do not matter for evolution. Somatic mutations occur in non-reproductive cells and won't be passed onto offspring.

The only mutations that matter to large-scale evolution are those that can be passed on to offspring. These occur in reproductive cells like eggs and sperm and are called germ line mutations.

Effects of germ line mutations

A single germ line mutation can have a range of effects:

1. **No change occurs in phenotype.**
   Some mutations don't have any noticeable effect on the phenotype of an organism.
2. **Small change occurs in phenotype.**
   A single mutation causes cat's ears to curl backwards slightly.
3. **Big change occurs in phenotype.**
   Some really important phenotypic changes, like drug resistance in insects, are sometimes caused by single mutations. A single mutation can also have strong negative effects for the organism.