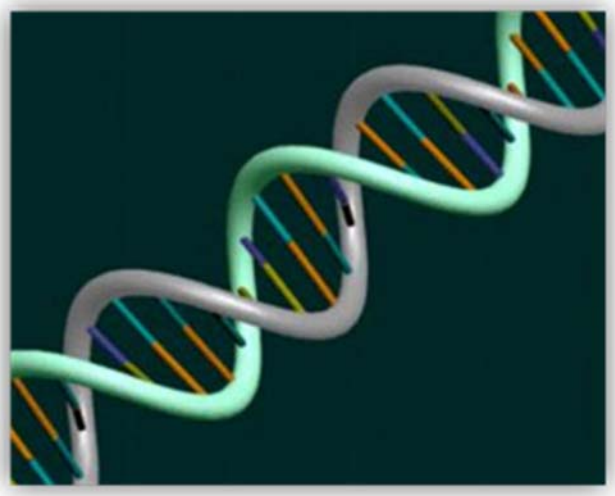


GENE MUTATION

You have learned the structure of DNA and RNA, and how DNA and RNA work in transcription and translation with the goal of making a protein. We will now discuss some of the mistakes that can happen with DNA and the sequence it holds which may affect the organism. A **mutation** is any change in the structure or amount of genetic material (DNA or RNA). When a mutation occurs in DNA it affects the transcription of the RNA sequence which then affects the translation of the protein. Mutations may harm the organism, help the organism, or have no effect on the organism and go unnoticed. A mutation can be beneficial, as in modifying an organism to better adapt to an environment, or harmful, as in negatively changing a structure or function within an organism.

We have already discussed large scale mutations that concern chromosomes. Now, we will examine smaller scale mutations which may have a great effect on the organism. You should recall, in DNA replication, how *DNA polymerase* acts as a proof-reading enzyme to correct any mistakes that happened in the replication process. Sometimes, DNA polymerase will miss a mistake and let it pass to the daughter cells.

Possible changes in the DNA, or mutations, can happen naturally as an accidental change, or can be increased in likelihood by environmental factors. A **mutagen** is anything that causes a mutation. Some environmental factors that act as mutagens are different forms of radiation (ultraviolet & X-rays), some chemicals (benzene), and some biological agents (viruses & bacteria).

Mutations can occur in any type of cell. A mutation that affects the *gametes* (sperm and egg) is known as **germ-cell mutation**. Since this type of mutation alters the DNA of the cells that will produce the zygote, these mutations are passed on to the offspring. A germ-cell mutation will

not affect the organism itself, only the future organism it may possibly produce. A mutation that affects any of the *body cells* (any other cell besides a gamete) is known as a **somatic mutation**. A somatic mutation will not be passed on to the offspring, but will affect the organism with the mutation.

Mutations that result as a change in the DNA Sequence				
Mutation Type		**Point Mutation (Substitution)**	**Insertion**	**Deletion**
Mutation Description		Involves a change of a single nucleotide.	Involves a nucleotide that is added into the DNA sequence.	Involves a nucleotide that has been removed from the DNA sequence.
Mutation Example	***Original DNA Sequence***	A-C-G-T	A-C-G-T	A-C-G-T
	Mutated DNA Sequence	A-C-**T**-T	A-C-G-**C**-T	A-G-T

Mutations that result as a change in the Gene				
Use the "Original DNA sequence", "mRNA sequence", and "amino acid sequence" below as a comparison for the different types of mutations listed below.				
Original DNA Sequence (as codons)	CGG	GGG	TCA	CCT
mRNA Sequence (from transcription)	GCC	CCC	AGU	GGA
Amino Acid Sequence (from translation)	Alanine	Proline	Serine	Glycine
Silent Mutation	**A DNA mutation that has no effect on a gene's function or amino acid coding.** Notice the point mutation that took place in the third nucleotide of the first codon. Changing the third nucleotide, in this case, had no effect on the originally intended amino acid as it is still coding for alanine.			
Mutated DNA Sequence	CGT	GGG	TCA	CCT
New mRNA Sequence	GCA	CCC	AGU	GGA
New Amino Acid Sequence	Alanine	Proline	Serine	Glycine

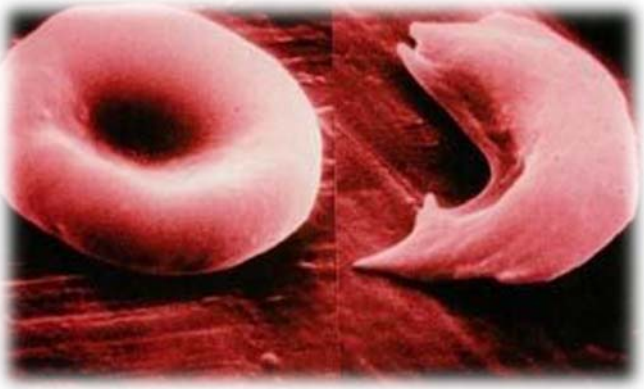
Missense (Replacement) Mutation	**A DNA mutation that changes one codon to code for a different amino acid.** Notice the point mutation that took place in the second nucleotide of the third codon. Changing the second nucleotide, in this case, has an effect on the originally intended amino acid. This mutation now calls for the codon to code for isoleucine instead of serine.			
Mutated DNA Sequence	CGG	GGG	TAA	CCT
New mRNA Sequence	GCC	CCC	AUU	GGA
New Amino Acid Sequence	Alanine	Proline	Isoleucine	Glycine
Frameshift Mutation (Due to insertion)	**A DNA mutation that shifts the entire DNA sequence from the point of the mutation forward.** Notice the insertion mutation that took place in the first nucleotide of the second codon. Adding a nucleotide, in this case, has an effect on the originally intended amino acid and every amino acid from that point onward.			
Mutated DNA Sequence	CGG	AGGG	TCA	CCT
New mRNA Sequence	GCC	UCCC	AGU	GGA
New Amino Acid Sequence	Alanine	Serine	Glutamine	Tryptophan
Frameshift Mutation (Due to deletion)	**A DNA mutation that shifts the entire DNA sequence from the point of the mutation forward.** Notice the deletion mutation that took place in the first nucleotide of the second codon. Deleting a nucleotide, in this case, has an effect on the originally intended amino acid and every amino acid from that point onward.			
Mutated DNA Sequence	CGG	GG	TCA	CCT
New mRNA Sequence	GCC	CC	AGU	GGA
New Amino Acid Sequence	Alanine	Proline	Valine	Unknown until third nucleotide is identified
Nonsense Mutation	**A DNA mutation that changes one codon to a stop codon.** Notice the point mutation that took place in the first nucleotide of the fourth codon. Changing the first nucleotide, in this case, has an effect on the originally intended amino acid. This mutation now calls for a stop codon to suddenly stop the translating of this code.			
Mutated DNA Sequence	CGG	GGG	TCA	ACT
New mRNA Sequence	GCC	CCC	AGU	UGA
New Amino Acid Sequence	Alanine	Proline	Isoleucine	STOP

The following video focuses on mutations that can occur in DNA, which take place when an issue occurs with a nucleotide. Missense and nonsense mutations occur when one nucleotide is simply changed to another. Frameshift mutations are the result of a nucleotide being inserted or deleted, altering the rest of the codons. Synonymous substitutions are mutations in which a nucleotide is changed, but it does not impact the meaning of the codon.

QuickTime DNA Mutations (06:02)

We can use the statement **“the fat cat sat”** to help you understand some of these mutations. A *missense (replacement) mutation* would have a letter change in a word that would therefore change the word, as in changing the “a” in “cat” to an “e”. Changing the word now results in a strange unknown meaning **(“the fat cet sat”)**, just as changing an amino acid would change the protein. A *frameshift mutation (due to insertion)* would have a letter added to a word, which would shift the letters in all of the three letter words from the point of the addition forward. This is demonstrated by adding the letter “a” in front of “fat” **(“the afa tca tsa t”)**. A *frameshift mutation (due to deletion)* would have a letter removed from a word, which would shift the letters in all of the three letter words from the point of the deletion forward. This is demonstrated by removing the letter “f” from “fat” **(“the atc ats at”)**. A *nonsense mutation* would have one of the words replaced with a period (stop codon) which ends the statement abruptly with no meaning as in replacing “cat” with a period **(“the fat.”)**.

Sickle Cell Anemia Research



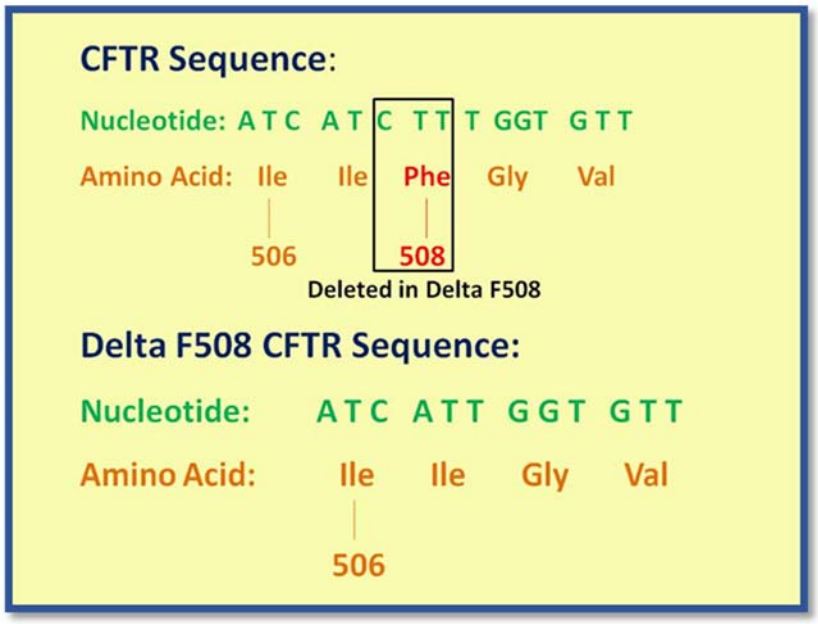
Sickle Cell Anemia is a genetic disease caused by a point mutation which results in a missense mutation. The mutation cause the original codon of GAG to change to GTG on chromosome 11. The change in one base of the codon goes from coding for the amino acid glutamic acid to the amino acid valine which dramatically changes the protein found in the blood. Visit the link below to learn more about sickle cell anemia and how it affects an individual.

http://learn.genetics.utah.edu/content/disorders/singlegene/

QuickTime Genetic Disease: What is Sickle Cell Anemia?

Cystic Fibrosis Research

Cystic Fibrosis is a genetic disease caused by many different mutations. There are different classes of cystic fibrosis based on what type of mutation is involved. The most common mutation found in about 70% of cystic fibrosis genes is a deletion mutation (known as Delta F508). The deletion involves removing three nucleotides (or one codon) which coded for phenylalanine. Visit the link below to learn more about cystic fibrosis and how it affects an individual.



http://learn.genetics.utah.edu/content/disorders/singlegene/

QuickTime Cystic Fibrosis (03:20)

Unit 17 Worksheet Gene Mutations

UNIT VOCABULARY REVIEW

Click on the **Quizlet icon** below to access the quizlet.com vocabulary flash cards. Review the vocabulary before completing your assessment.

Quizlet

Now answer questions 1 through 20.