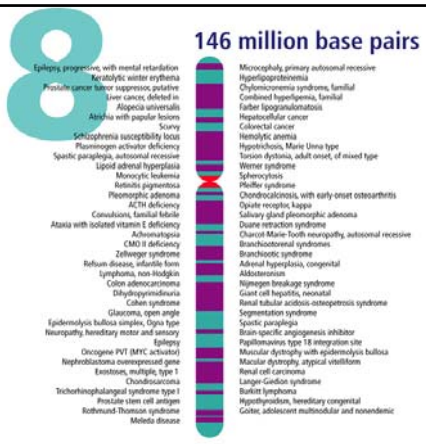
Chromosome #



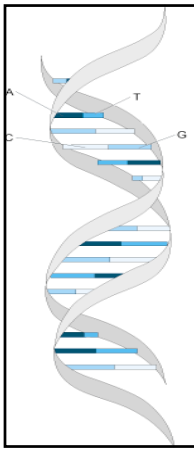
Although we all look different from one another, we are surprisingly identical at the DNA level. Compare the DNA of any two humans and it will be 99.9% the same.

Humans have approximately **three billion base pairs** (6 billion bases total) of DNA in most of our cells. This complete set of code made up of units called genes is our genome. The exact sequence of the nitrogen bases is different for everyone, which what makes each of us unique.

The reality is that only about 1.5 million bases are responsible for the differences between us. This is only **.01% of our DNA**. Yet these variations in our DNA base sequence influence most of our physical differences and many of our other characteristics.

Timeline of DNA Discoveries

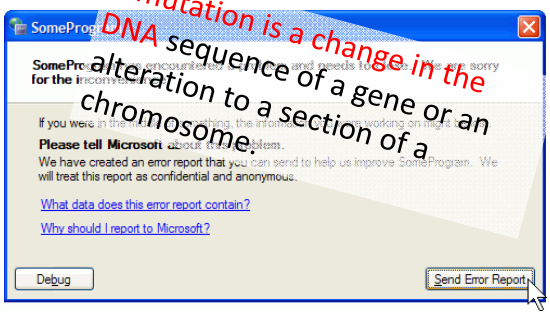
- **1953 -** Francis Crick and James Watson determine that the structure of the DNA molecule is a double helix composed of strings of nucleotides and that two parallel strands formed by sugar and phosphate molecules are joined together by the bonding of specific pairs of nitrogenous bases. They share a Nobel Prize for this in 1962.
- **1961 -** Sydney Brenner, Francois Jacob, and Matthew Meselson identify the role of Ribonucleic Acid (RNA). They determine that messenger RNA (mRNA) is the molecule that carries the genetic information from DNA in the nucleus out into the cytoplasm and that the cell ultimately uses mRNA to make specific proteins.
- **1966 -** Marshall Nirenberg and H. Gobind Khorana lead teams that crack the genetic code. They demonstrate that each of 20 amino acids is coded by a sequence of three nucleotide bases (each series of three bases is called a codon).
- **1990 -** The Human Genome Project, an international effort to sequence all of the DNA and map all of the genes in humans, is launched.
- **2000 -** A rough draft of the human genome is completed and published by the Human Genome Project and Celera. The project was planned to last 15 years, but rapid technological advances accelerated the expected completion date. Project goals are to discover all 30,000 to 40,000 human genes (the human genome) and make them accessible for further study and to determine the complete sequence of the 3 billion DNA bases in the human genome.



The code that is carried in DNA determines which amino acids will come together in what order to form a given protein.

In other words, Genes express themselves by specifying the order and type of amino acids used by a ribosome when it makes a protein.

In addition to determining what proteins are made the DNA in a cell also controls how much of each kind of protein will be made and when.



A mutation is a change in the DNA sequence of a gene or an alteration to a section of a chromosome.

Most DNA variation ("variation" is just another way to say mutation) is neutral (not beneficial or harmful), but harmful sequence changes sometimes do occur.

Changes within genes can result in proteins that don't work normally or don't work at all. Some of these changes can contribute to disease or affect how someone responds to medicine.

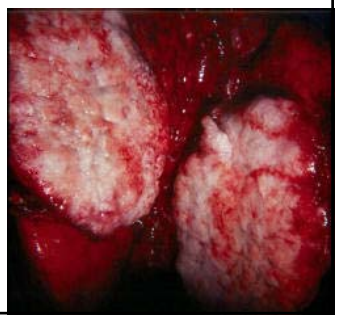
- Mutations may be passed down from parent to child (in sperm or egg cells)
- Or they may occur around the time of conception
- Or mutations may be acquired during a person's lifetime.

Mutations can arise spontaneously during normal cell functions, such as when a cell divides, or in response to environmental factors such as toxins, radiation, hormones, and even diet.

These things are called mutagens

Nature provides us with a system of repair enzymes that find and fix most DNA errors. But as our bodies change in response to age, illness and other factors, our repair systems may become less efficient.

Uncorrected mutations can accumulate, resulting in diseases such as cancer.



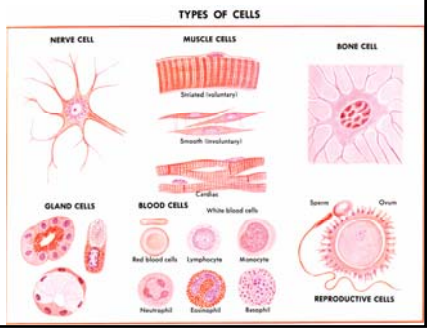
Genes: Are a sequence of DNA that code for a specific protein

Genes consist of a length of DNA that contains the instructions (the code) for making a specific protein.

Through proteins, our genes influence almost everything about us, including how tall we will be, how we process foods, and how we respond to infections and medicines.

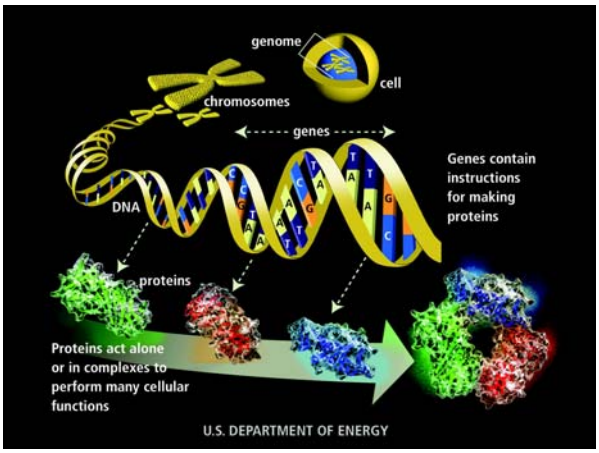
Although most of our cells have the same genes, not all genes are active in every cell.

Heart cells synthesize proteins required for that organ's structure and function;
liver cells make liver proteins



Within an individual cell, the same genes may be switched on at some times and switched off at other times.

The process of making a protein using the information stored in a gene begins with TRANSCRIPTION



Transcription

- The process that creates RNA using the coding strand of DNA as a template.
- RNA is Ribonucleic Acid
- RNA is a single stranded polymer
- RNA is used to transmit the information from the DNA in the nucleus to the ribosomes in the cytoplasm.
- RNA exits the nucleus by way of the nuclear pores.

Flash Animation

Transcription

Flash Animation

There are three kinds of RNA

- **mRNA**- Messenger RNA- contains the recipe for making protein
- **tRNA**- Transfer RNA- brings amino acids to the Ribosome from the cytoplasm
- **rRNA**- Ribosomal RNA- is what ribosomes are made from

RNA Polymerase assembles the RNA using the following RNA substitution rules:

DNA	RNA
If you have a T	You get A
If you have a G	You get C
If you have a C	You get G
If you have an A	You get U

A-T-C-G-C-G-T-A-T-G-C-A-T-A-C-T-A-G

T-A-G-C-G-C-A-T-A-C-G-T-A-T-G-A-T-C

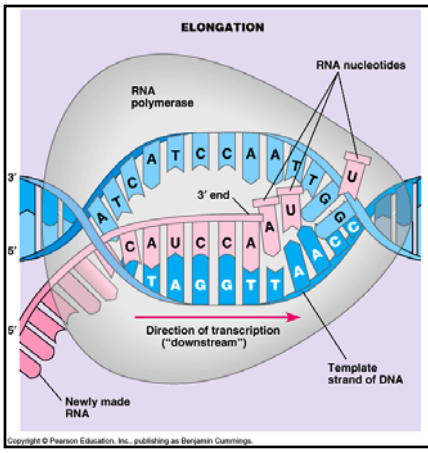
TRANSCRIPTION USES THE CODING STRAND (BLUE) TO MAKE

A-U-C-G-C-G-U-A-U-G-C-A-U-A-C-U-A-G

mRNA – Single stranded – Brings message (or recipe for a protein) to ribosome

mRNA is used in groups of 3 nucleotides called CODONS.

There are 6 codons in the strand of mRNA above



Transcription

Transcription is the process by which the information contained in a section of DNA (called a gene) is transferred to a newly assembled piece of messenger RNA (mRNA).

It is facilitated by the enzyme RNA polymerase.

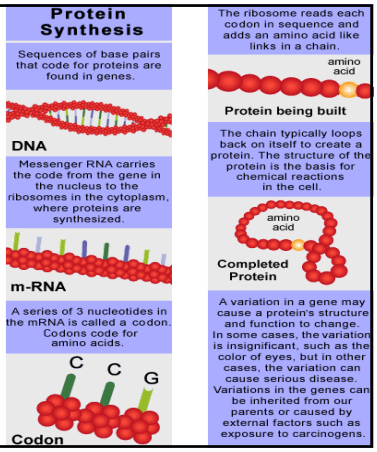
Animation for transcription

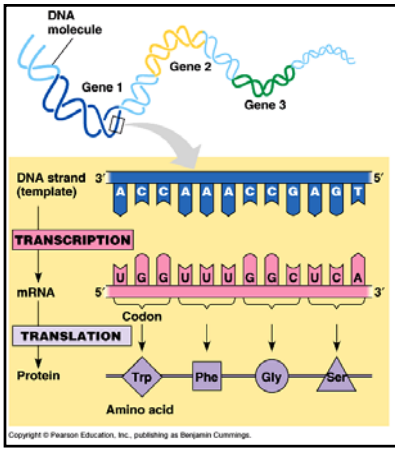
Genes act, or "express," themselves by dictating the order of amino acids used to make proteins.

The proteins made by some genes are needed by all cells, but different sets of genes may be switched on or off in different cells.

This leads to different collections of proteins being made and results in different structures, appearances and functions.

Protein Synthesis (the process of making a protein) is called translation.



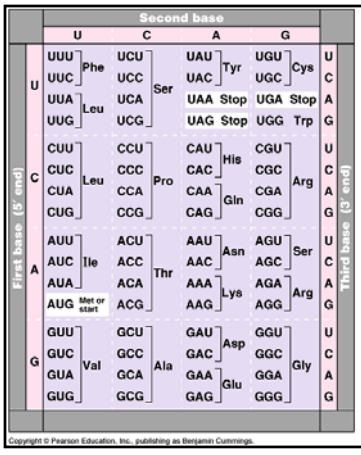


Translation

After transcription mRNA exits the nucleus via a nuclear pore and finds its way to a ribosome, where it is translated. The mRNA is read by the ribosome as triplet codons.

The mRNA codons make sure that the ribosome puts the correct amino acid in the protein.

As the amino acids are linked into a growing peptide chain, they eventually form a protein.



There are 64 possible 3 base combinations that can be formed using the 4 bases.

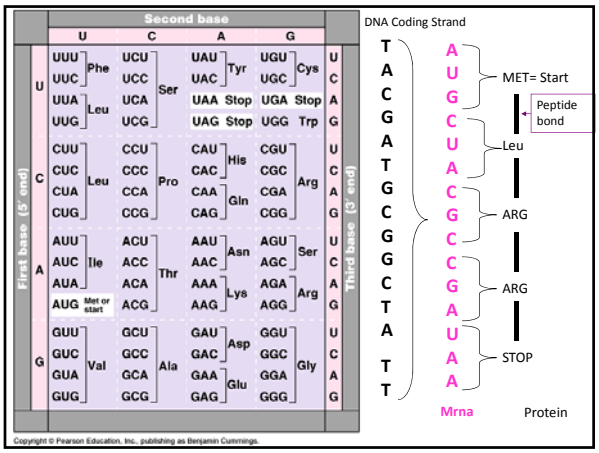
Each three base sequence is called a codon.

There are 20 amino acids that can be used to make a protein.

Because there are more codons then there are amino acids …

some codons code for the same amino acid.

This is how different sequences of mRNA can result in the same protein.



The Central Dogma

The information in DNA is used to make RNA that is used to put amino acids into a specific functional sequence called a protein.

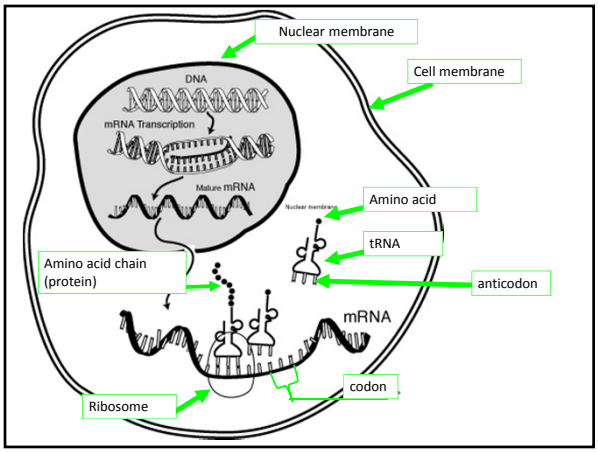
Transcription

Transcription is the process by which the information contained in a section of DNA is transferred to a newly assembled piece of messenger RNA (mRNA). It is facilitated by RNA polymerase and transcription factors.

It is followed by

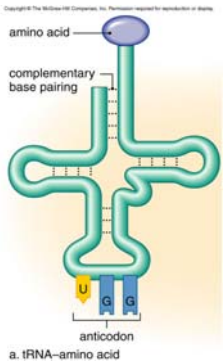
Translation

Processed mRNA exits the nucleus via a nuclear pore and finds its way to a ribosome, where it is translated. The mRNA is read by the ribosome as triplet codons, usually beginning with an AUG codon. Transfer RNA (tRNA) with the matching the anti-codon to the mRNA codon bring the correct amino acid in the sequence encoding the gene. As the amino acids are linked into the growing peptide chain, they begin folding into the correct conformation.

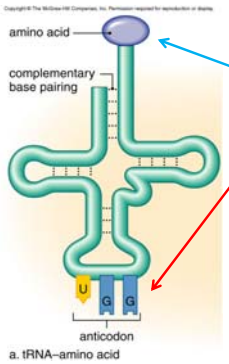


tRNA has Anticodons

Transfer RNA (tRNA) is a small RNA chain that transfers a specific amino acid to a growing polypeptide chain at the ribosome during protein synthesis (translation).



tRNA has Anticodons



Each tRNA has a site for **one amino acid to attach**.

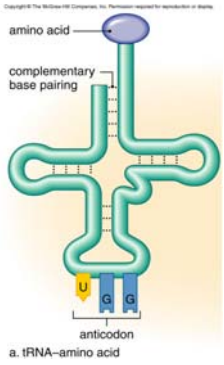
Opposite of the amino acid there is a three-base region called the **anticodon** that fits together with the corresponding three-base codon region on mRNA.

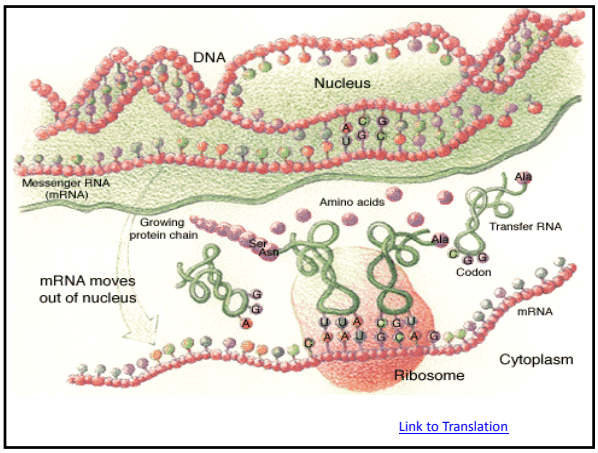
tRNA has Anticodons

Each type of tRNA molecule can be attached to only one type of amino acid but….

The genetic code contains multiple codons that specify the same amino acid.

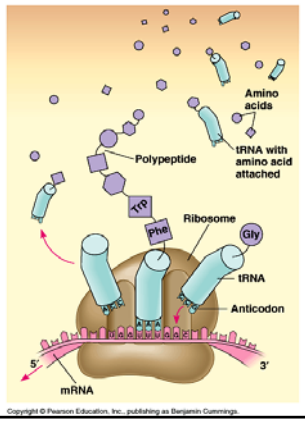
This means that multiple types of tRNA molecules each with a different anticodon may carry the same amino acid.

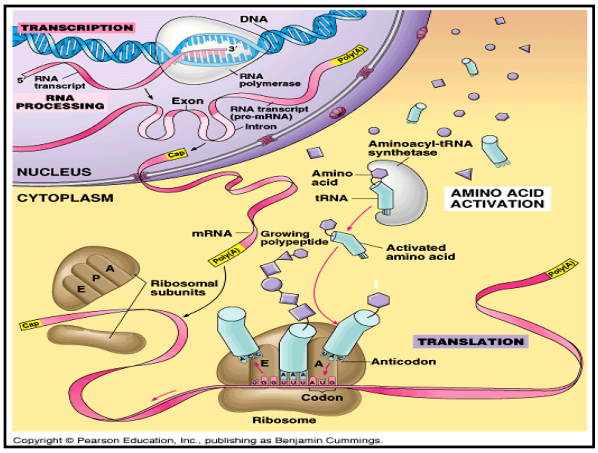


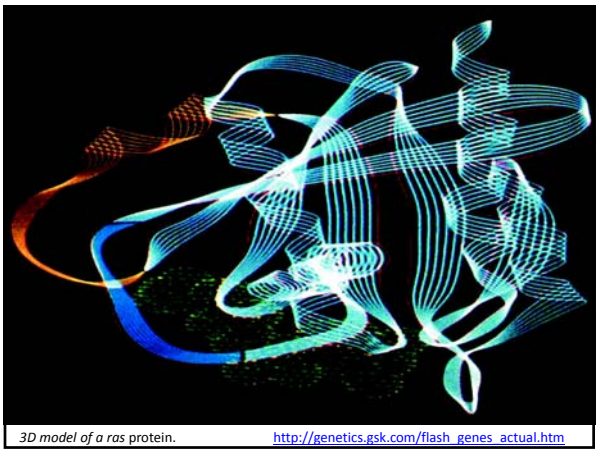


Link to Translation

Figure 17.12 Translation: the basic concept

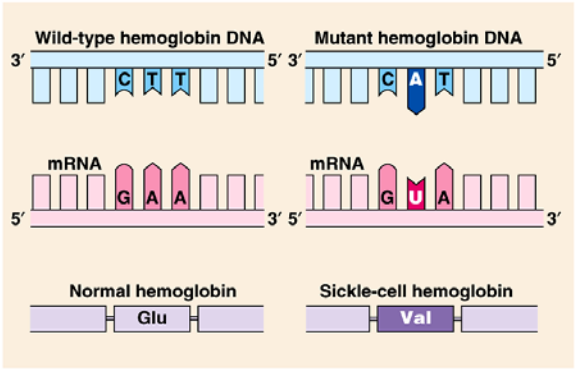






3D model of a ras protein. http://genetics.gsk.com/flash_genes_actual.htm

Mutations Alter the Codons



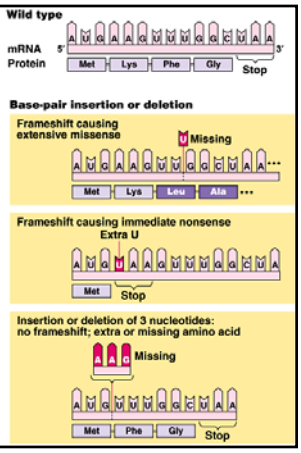
Point mutations and base-pair insertion or deletion

A Point Mutation is when one single base is altered. Will only change one codon.

A Base Insertion is when one is added

A Base Deletion is when one base is removed

Insertions and Deletions are worse for protein synthesis as they will change all of the codons from the point of the mutation forward.



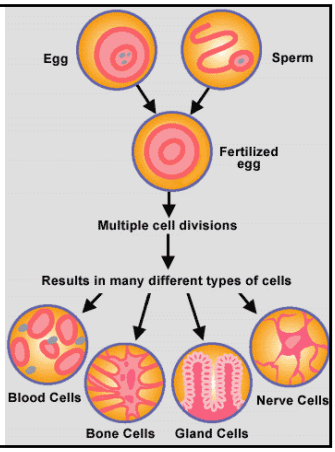
Why do children so often resemble their parents?

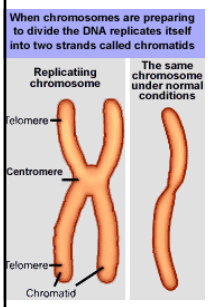
Why do some brothers and sisters share similar traits, while others are very different?

To a large degree, it's a function of the genes (which are the basic units of heredity) they have in common.

How does this happen?

To understand that we will need to find out a little bit more about what genes are and how we inherit them.





A chromosome is a long strand of DNA, packaged together with proteins and other kinds of molecules. Each chromosome has a centromere, which plays an important role during cell division and also divides each chromosome into a short arm and a long arm. Scientists can tell different chromosomes apart based on their size, the relative lengths of their arms, distinctive staining patterns, and other characteristics.

Humans have two types of chromosomes: sex chromosomes and autosomes. Two sex chromosomes determine the sex of an individual, and they are called the X chromosome and the Y chromosome.

If you are female, you have two Xs, and if you are male, you have one X and one Y (although there are genetic conditions in which this varies). The autosomes comprise the other 22 chromosomes. The longest of the autosomes is referred to as chromosome 1, the next largest as chromosome 2, and so on, down to the smallest autosomes, chromosomes 21 and 22.

Each cell nucleus contains two copies of each autosome (44 chromosomes), plus two sex chromosomes (either two Xs or an X and a Y) for a total of 46. With few exceptions, the chromosomes and genes found within any two cells of your body will be identical.

The mystery as to why you resemble your family members is solved by discovering how you inherited your chromosomes from your parents.