Genetics 101

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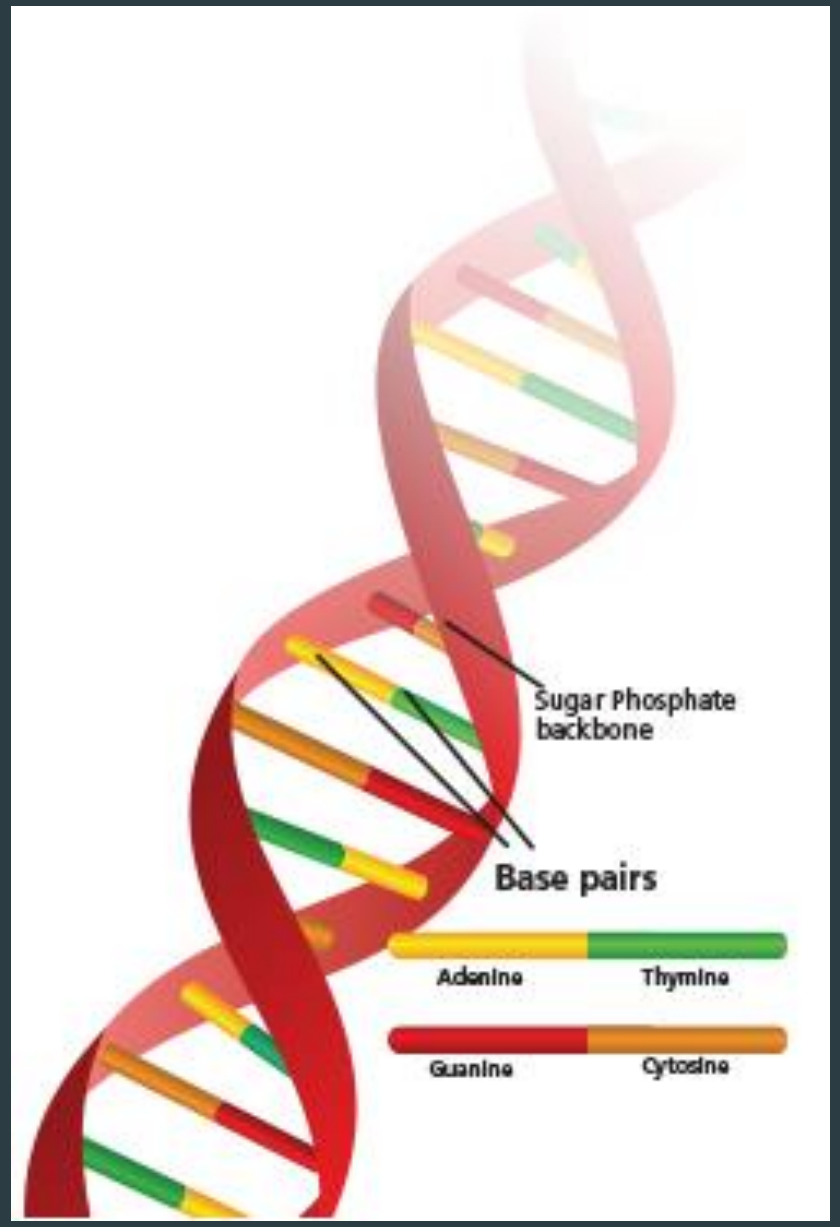
Before we get started! Genetics 101



Additional Resources

http://www.geneticheath.com/

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http://www.nlm.nih.gov/medlineplus/magazine/issues/summer13/articles/summer13pg11-12.html

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https://www.khanacademy.org/science/biology/heredity-and-genetics/v/genetics-101-part-1--what-are-genes

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http://www.genome.gov/glossary/

DNA (Deoxyribonucleic Acid)

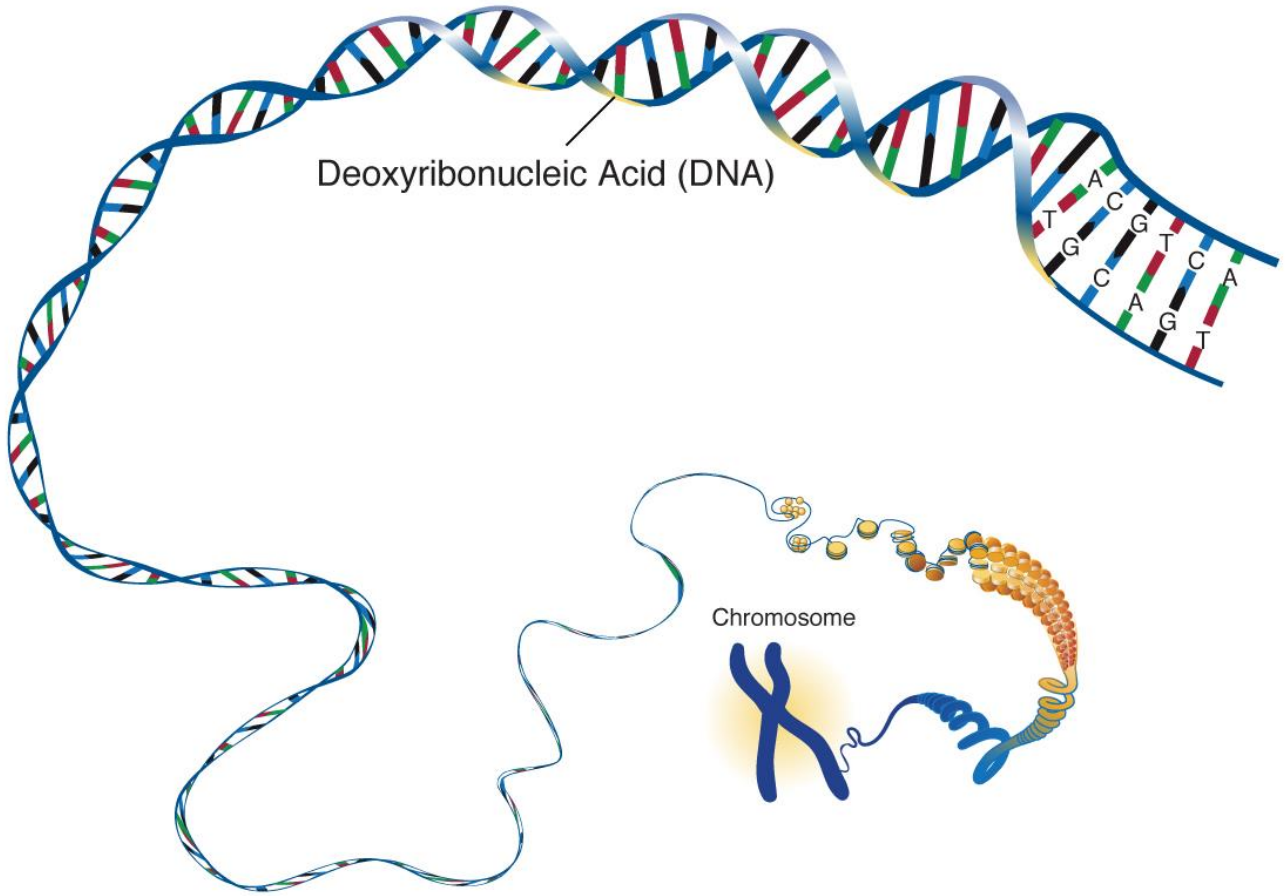

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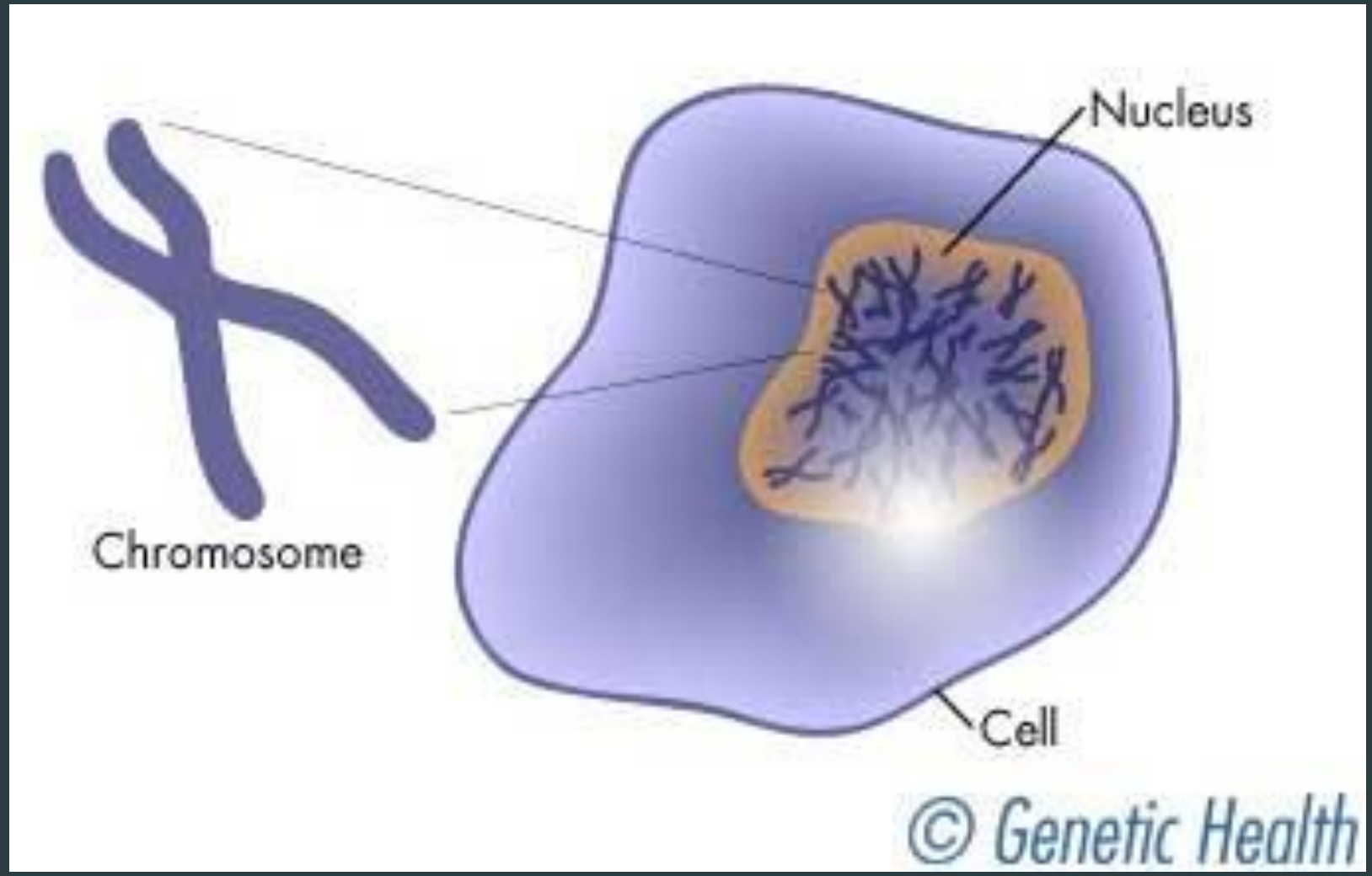
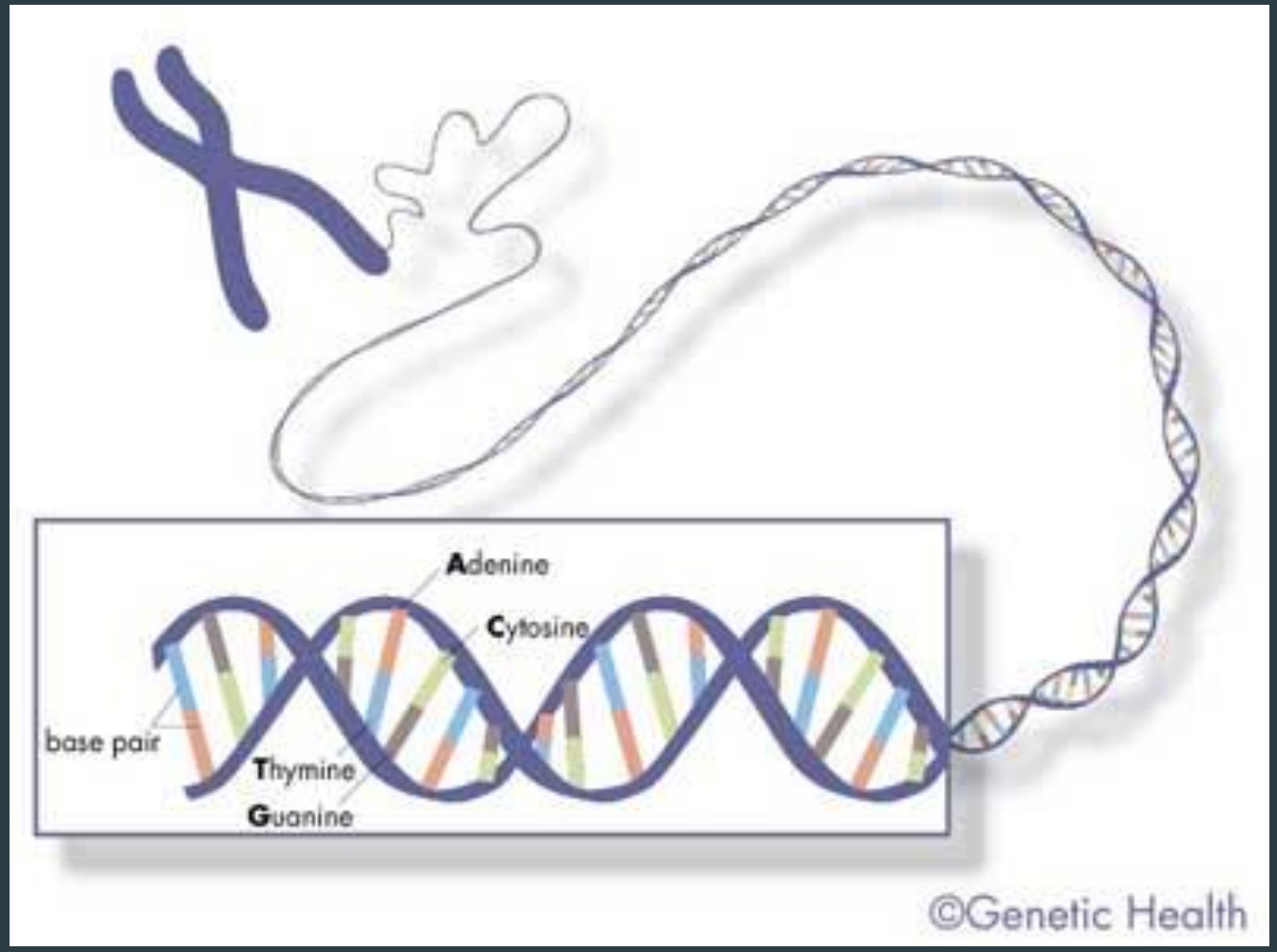
Illustration by Darryl Leja, NHGRI

What is DNA?



DNA (deoxyribonucleic acid) is a long molecule that is contained within almost all of our cells in a compartment called the nucleus. It is composed of individual units called bases. There are four types of bases, designated A (adenine), T (thymine), G (guanine), and C (cytosine)



Each DNA molecule is made of two individual strands paired together. Each strand consists of a series of the four bases. When the two strands pair up, an A on one strand is always across from a T on the other strand, and a C always pairs with a G. These A/T and G/C combination are called base pairs. The double-stranded molecule then twists like a coiled ribbon into a shape called a double helix. A piece of DNA millions of base pairs long — in conjunction with some proteins — is a chromosome.

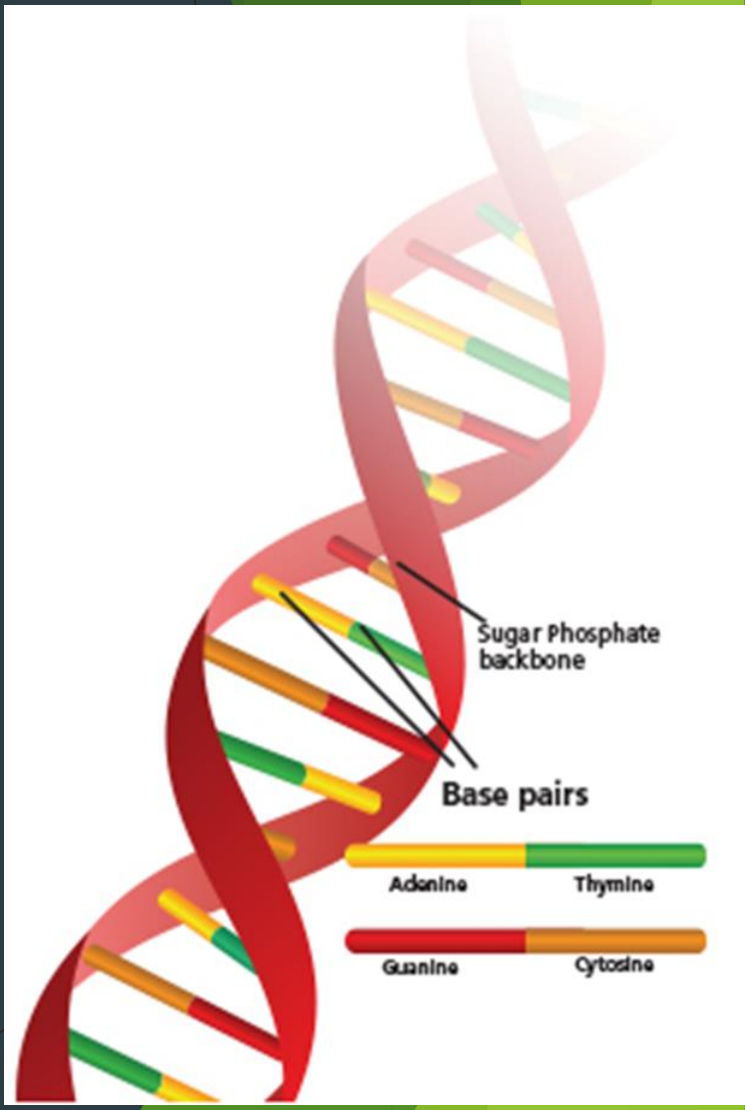
More on DNA

DNA (deoxyribonucleic acid) is the hereditary material in humans and almost all other organisms. Nearly every cell in a person's body has the same DNA. Most DNA is located in the cell nucleus (where it is called nuclear DNA), but a small amount of DNA can also be found in the mitochondria (where it is called mitochondrial DNA or mtDNA).

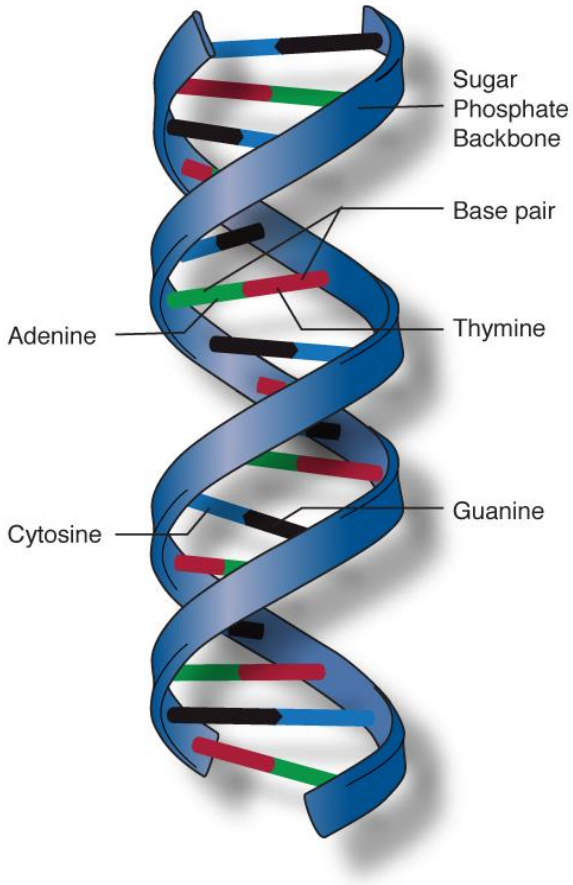
The information in DNA is stored as a code made up of four chemical bases: adenine (A), guanine (G), cytosine (C), and thymine (T). Human DNA consists of about 3 billion bases, and more than 99 percent of those bases are the same in all people. The order, or sequence, of these bases determines the information available for building and maintaining an organism, similar to the way in which letters of the alphabet appear in a certain order to form words and sentences.

DNA bases pair up with each other, A with T and C with G, to form units called base pairs. Each base is also attached to a sugar molecule and a phosphate molecule. Together, a base, sugar, and phosphate are called a nucleotide. Nucleotides are arranged in two long strands that form a spiral called a double helix. The structure of the double helix is somewhat like a ladder, with the base pairs forming the ladder's rungs and the sugar and phosphate molecules forming the vertical sidepieces of the ladder.

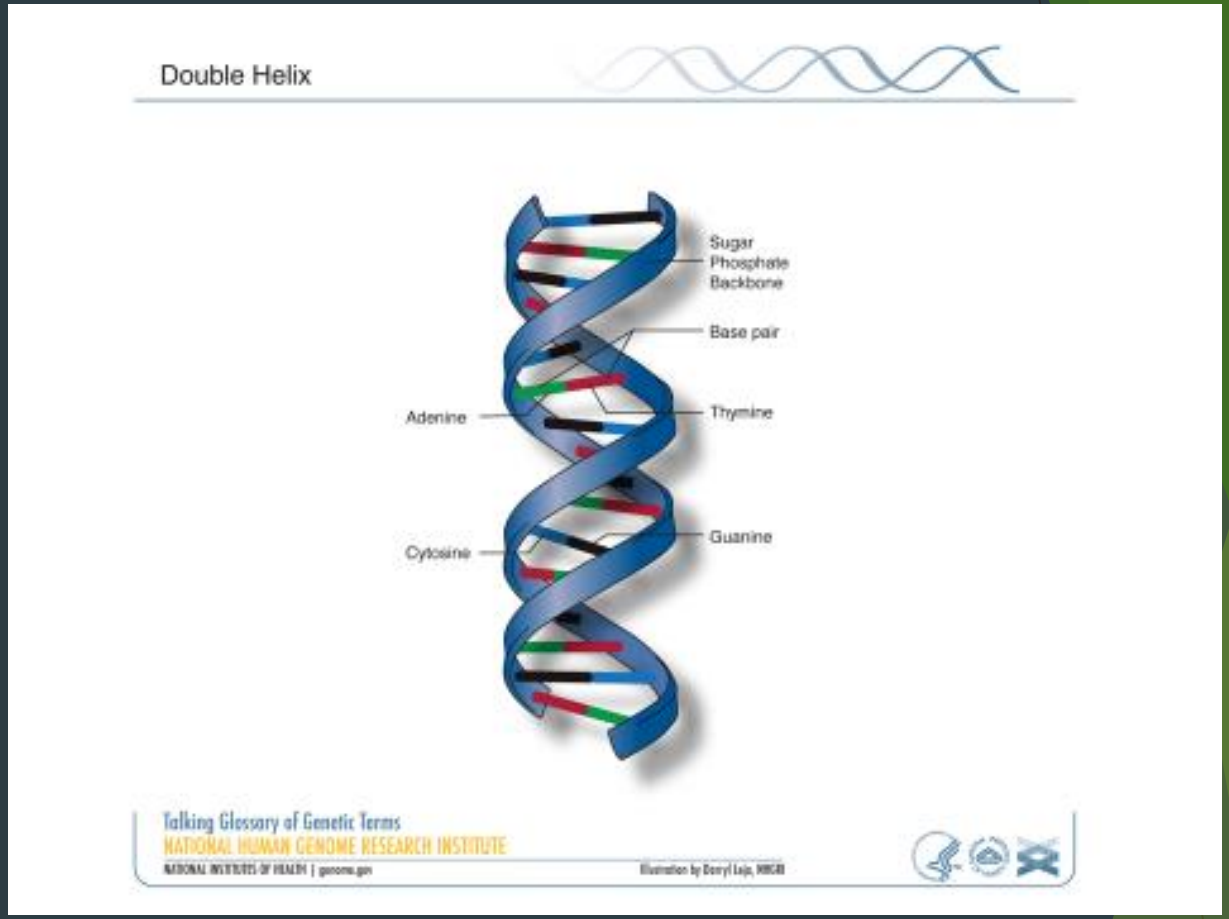
An important property of DNA is that it can replicate, or make copies of itself. Each strand of DNA in the double helix can serve as a pattern for duplicating the sequence of bases. This is critical when cells divide because each new cell needs to have an exact copy of the DNA present in the old cell.



Double Helix

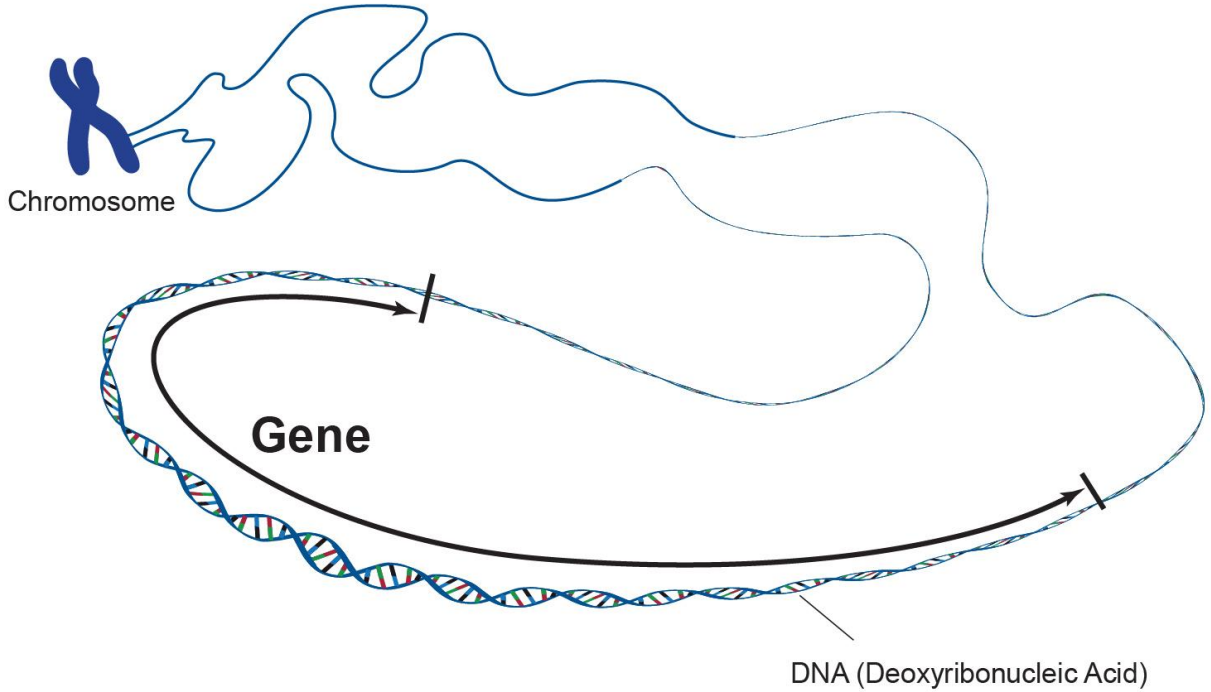


What is a Double Helix?



Double helix is the description of the structure of a DNA molecule. A DNA molecule consists of two strands that wind around each other like a twisted ladder. Each strand has a backbone made of alternating groups of sugar (deoxyribose) and phosphate groups. Attached to each sugar is one of four bases: adenine (A), cytosine (C), guanine (G), or thymine (T). The two strands are held together by bonds between the bases, adenine forming a base pair with thymine, and cytosine forming a base pair with guanine.

Gene



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What are Genes?

Each chromosome contains thousands of genes, each of which is several thousand bases long. The sequence of bases in each gene contains instructions for making a single protein.

Each protein serves a particular function in the body. For example, enzymes help us digest food, structural elements give our cells shape, and signaling molecules help the cells communicate with each other.

Additional bases that come before the genes on a chromosome tell cells when each gene should be used. For example, these sequences might contain instructions that a protein for making hair should only be made in certain skin cells, and not by other cells of the body.

A gene is the basic physical and functional unit of heredity. Genes, which are made up of DNA, act as instructions to make molecules called proteins

In humans, genes vary in size from a few hundred DNA bases to more than 2 million bases

The Human Genome Project has estimated that humans have between 20,000 and 25,000 genes

How do we inherit our genes?

Humans inherit 23 chromosomes from each of their parents for a total of 46 chromosomes. Of these, 44 are identical in men and women — these are called autosomes.

The remaining two chromosomes are called sex chromosomes, which are designated X and Y. Women inherit two X chromosomes, whereas men inherit one X chromosome from their mother and one Y chromosome from their father.

Because of the way we inherit our chromosomes, we all have two copies of every gene that is contained on the autosomes. Depending on the combination of the genes we inherit, we end up with some traits that resemble our mother and others that resemble our father.

Women have two copies of each gene on the X chromosome, while men have only the genes that they inherit from their mother on the X chromosome and only genes that they inherit from their father on the Y chromosome.

What is a gene mutation?

Each gene is made up of a series of bases, and those bases provide instructions for making a single protein.

Any change in the sequence of bases — and therefore in the protein instructions — is a mutation.

Just like changing a letter in a sentence can change the sentence's meaning, a mutation can change the instruction contained in the gene.

Some mutations have little or no effect on the protein, while others cause the protein not to function at all.

What kind of problems can mutations cause?

Some mutations result in proteins that do not function normally, and may end up causing disease. There are several ways that gene mutation can change the way a protein functions, including:

1. ***Altered function:*** Some mutations result in a protein that cannot carry out its normal function in the cell, or cannot carry out that function very well. One example of this type of mutation is sickle cell anemia. In this disorder, an altered protein in red blood cells alters the shape of the red blood cell, which causes the cell to become stuck in blood vessels. This prevents cells from carrying sufficient oxygen to the rest of the body.
2. ***Lack of protein:*** Some mutations prevent the protein from being made. One example of this type of mutation is hemophilia. In this condition, a mutation results in the absence of a protein that causes blood to clot. The result is uncontrolled bleeding in response to injury.
3. ***Change in how much protein is made:*** Some mutations cause too much or too little of a normal protein to be made. Although the protein itself functions properly, it is not present in quantities that are appropriate. One example of this is in the development of some cancers. In this case, a protein that prevents additional mutations from building up can become turned off. Without this protein, the cell accumulates mutations and becomes increasingly cancerous.

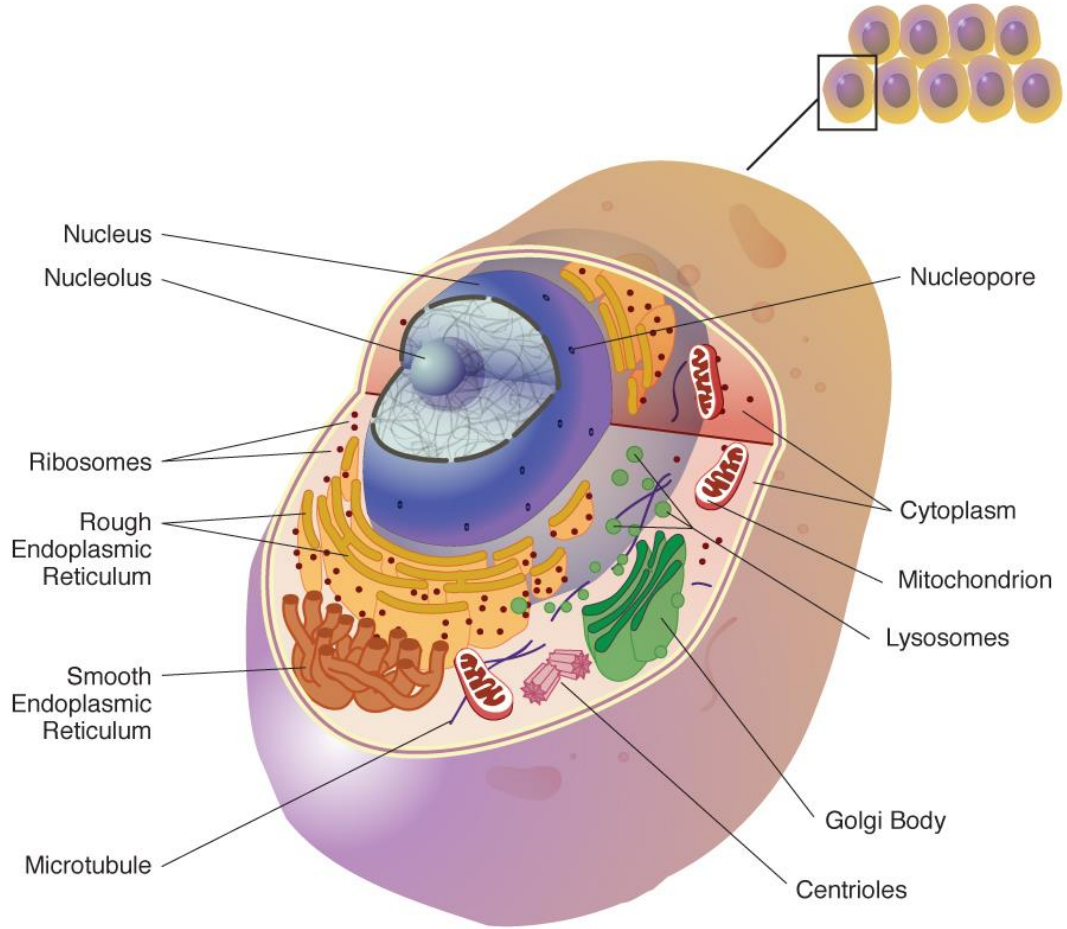
Can we overcome our genetics?

Our risk for almost any medical condition is a function of both our genes and our environment.

While we can't change our genes, we can apply our knowledge of our family medical history to predict our risk for specific problems.

This, in turn, allows us to focus on the things we can change — diet, lifestyle, screening, treatment — to ensure a long, healthy life

Cell



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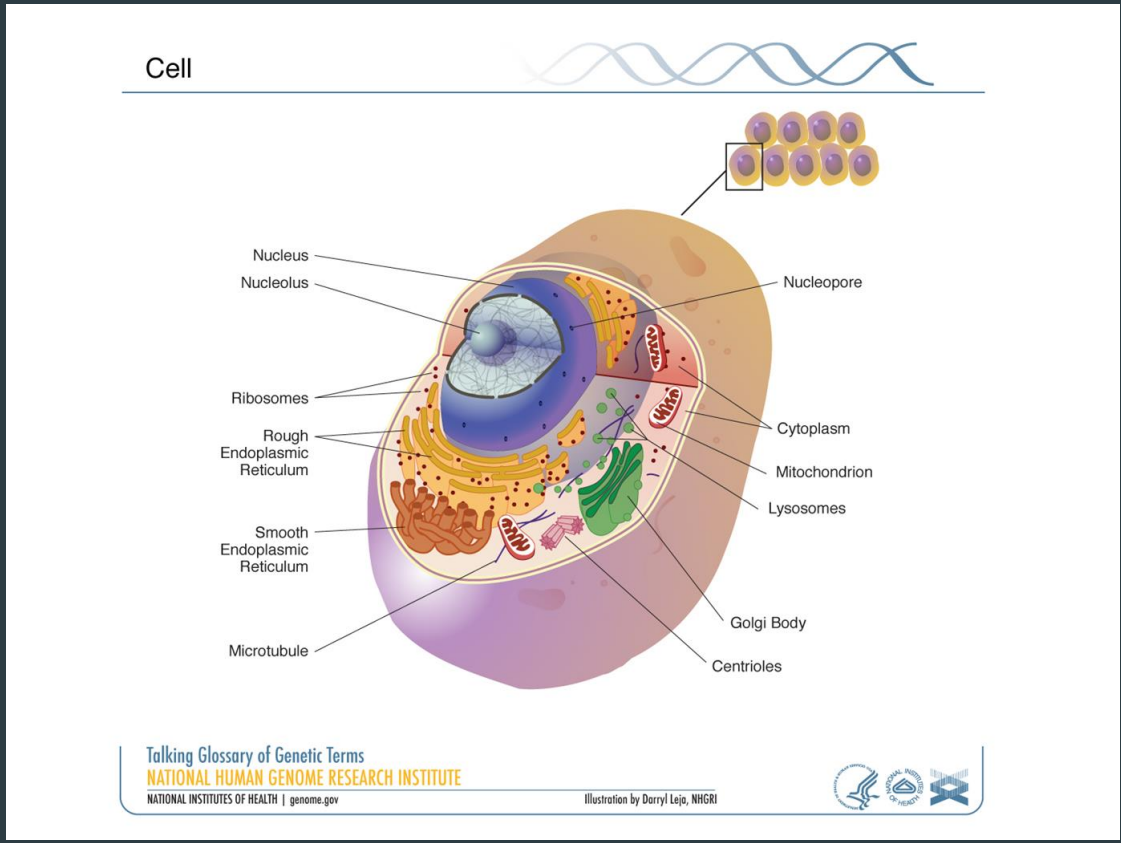
What is a Cell?



A cell is the basic building block of living things.

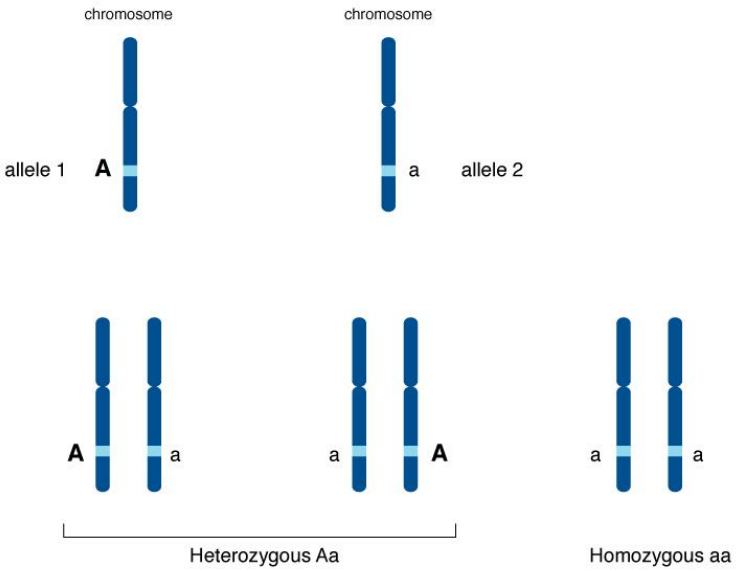
All cells can be sorted into one of two groups: eukaryotes and prokaryotes.

A eukaryote has a nucleus and membrane-bound organelles, while a prokaryote does not.

Plants and animals are made of numerous eukaryotic cells, while many microbes, such as bacteria, consist of single cells.

An adult human body is estimated to contain between 10 and 100 trillion cells.

Allele



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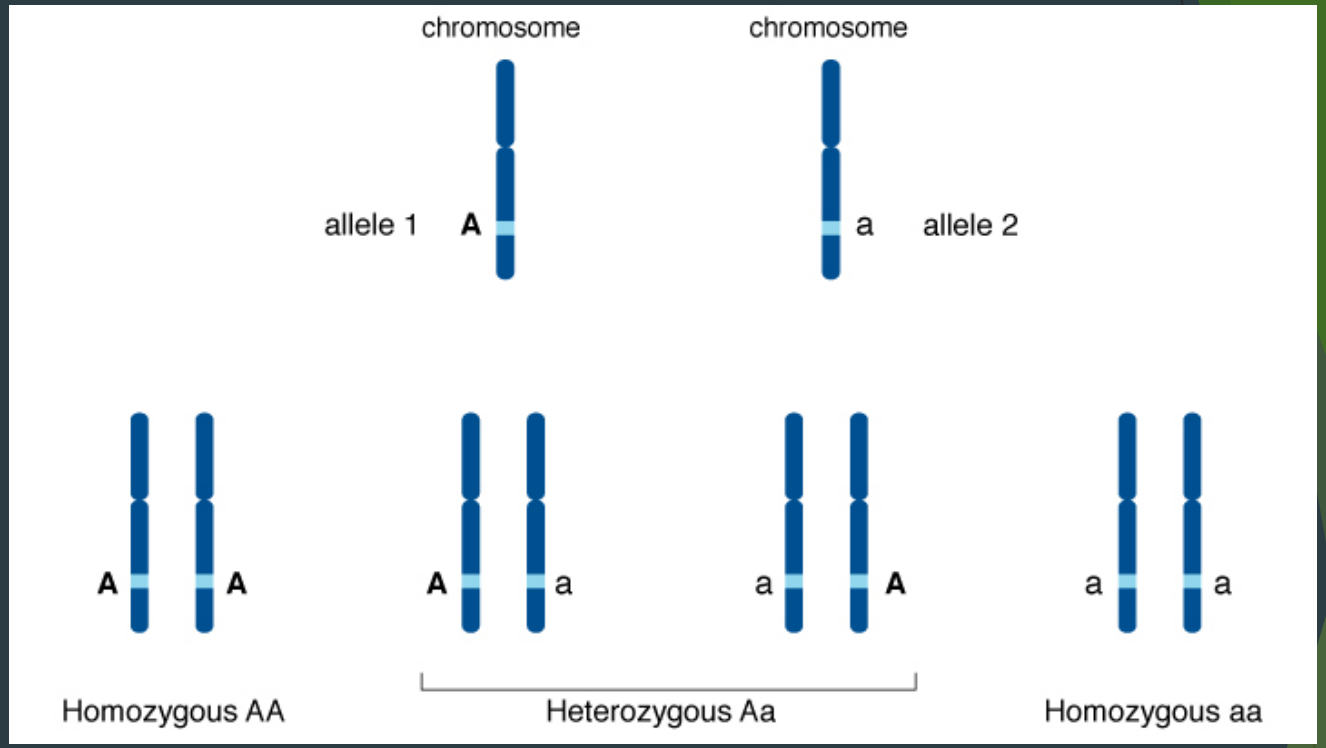
What is an Allele?



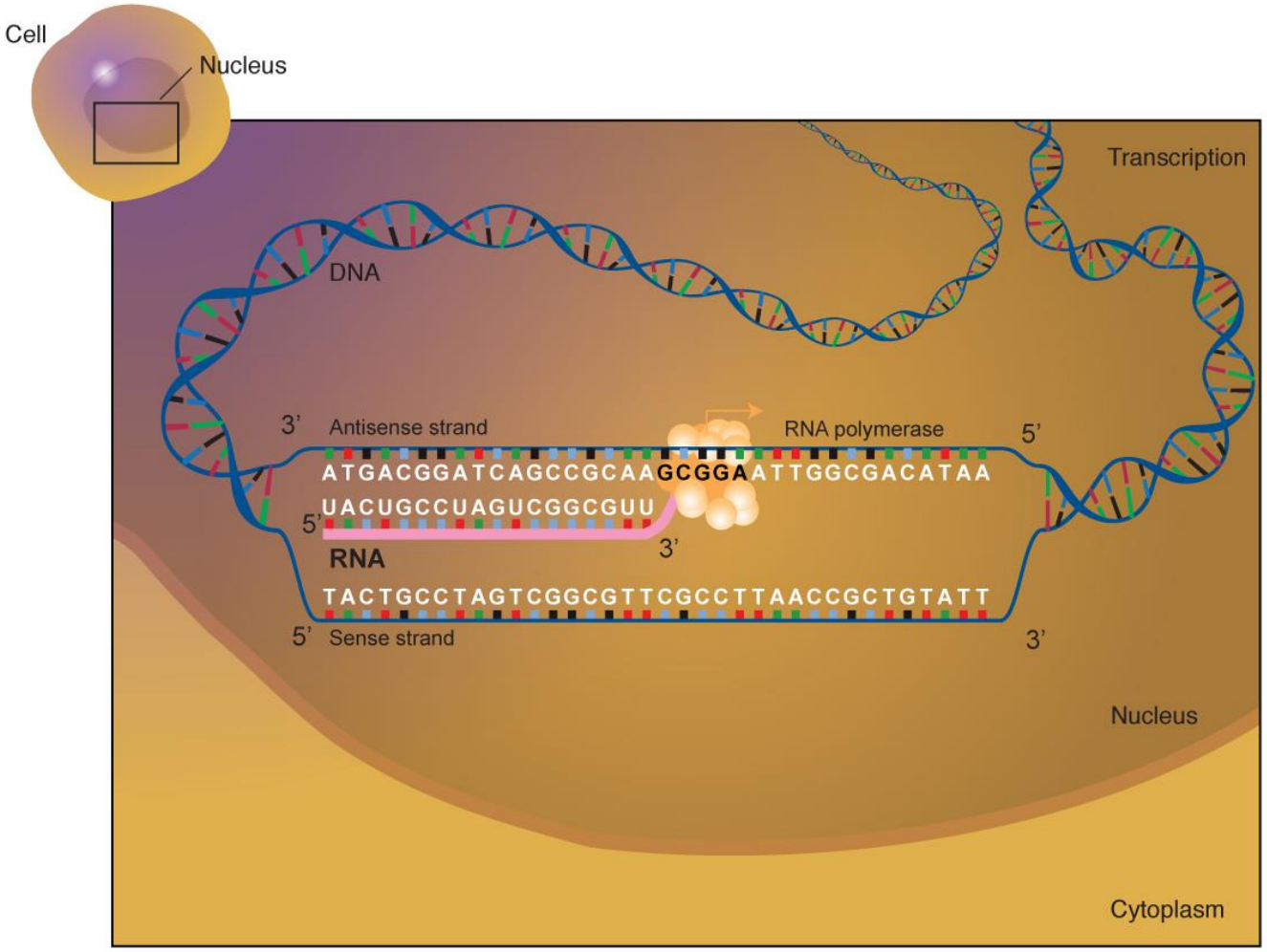
An allele is one of two or more versions of a gene.

An individual inherits two alleles for each gene, one from each parent.

If the two alleles are the same, the individual is homozygous for that gene. If the alleles are different, the individual is heterozygous.

Though the term allele was originally used to describe variation among genes, it now also refers to variation among non-coding DNA sequences.

RNA (Ribonucleic Acid)



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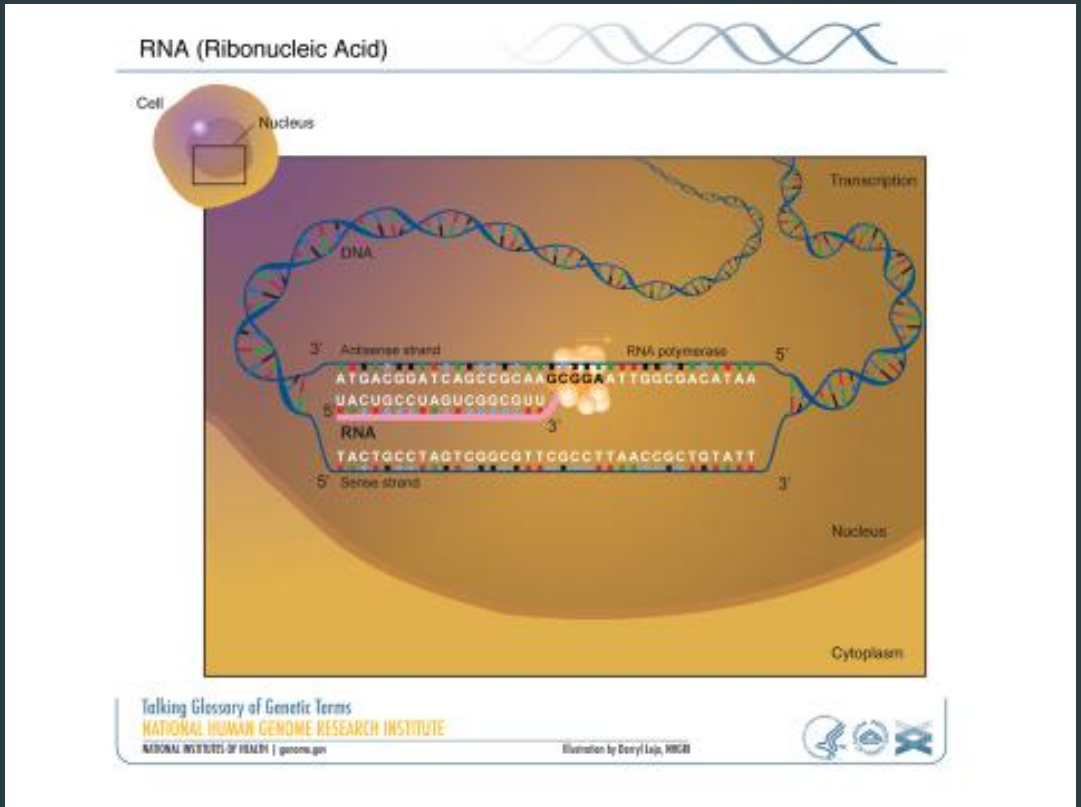
What is RNA?

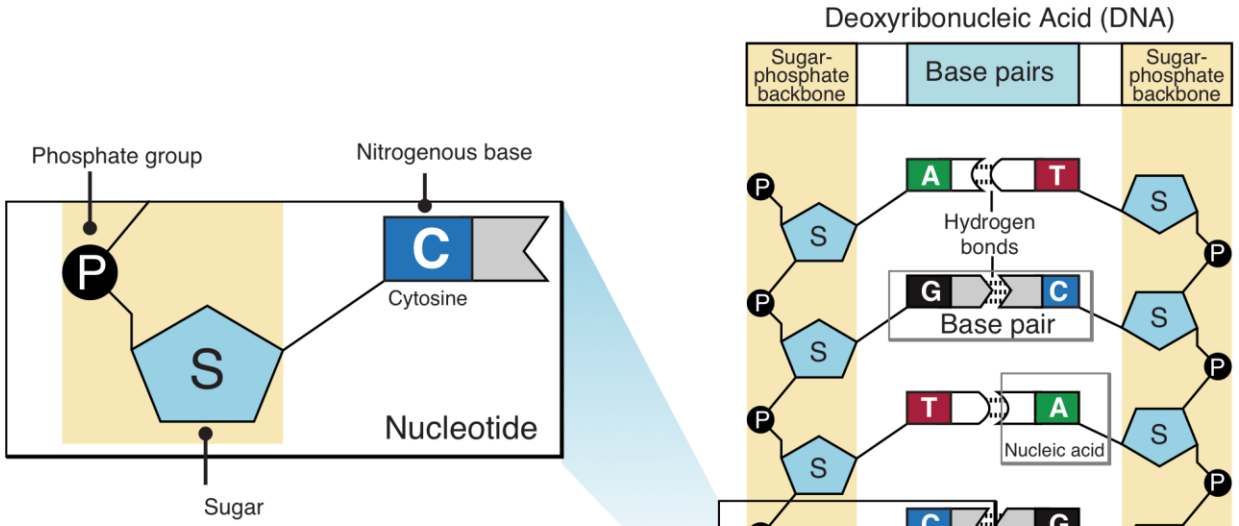


Ribonucleic acid (RNA) is a molecule similar to DNA. Unlike DNA, RNA is single-stranded. An RNA strand has a backbone made of alternating sugar (ribose) and phosphate groups. Attached to each sugar is one of four bases--adenine (A), uracil (U), cytosine (C), or guanine (G). Different types of RNA exist in the cell:

1. messenger RNA (mRNA)
2. ribosomal RNA (rRNA)
3. transfer RNA (tRNA).

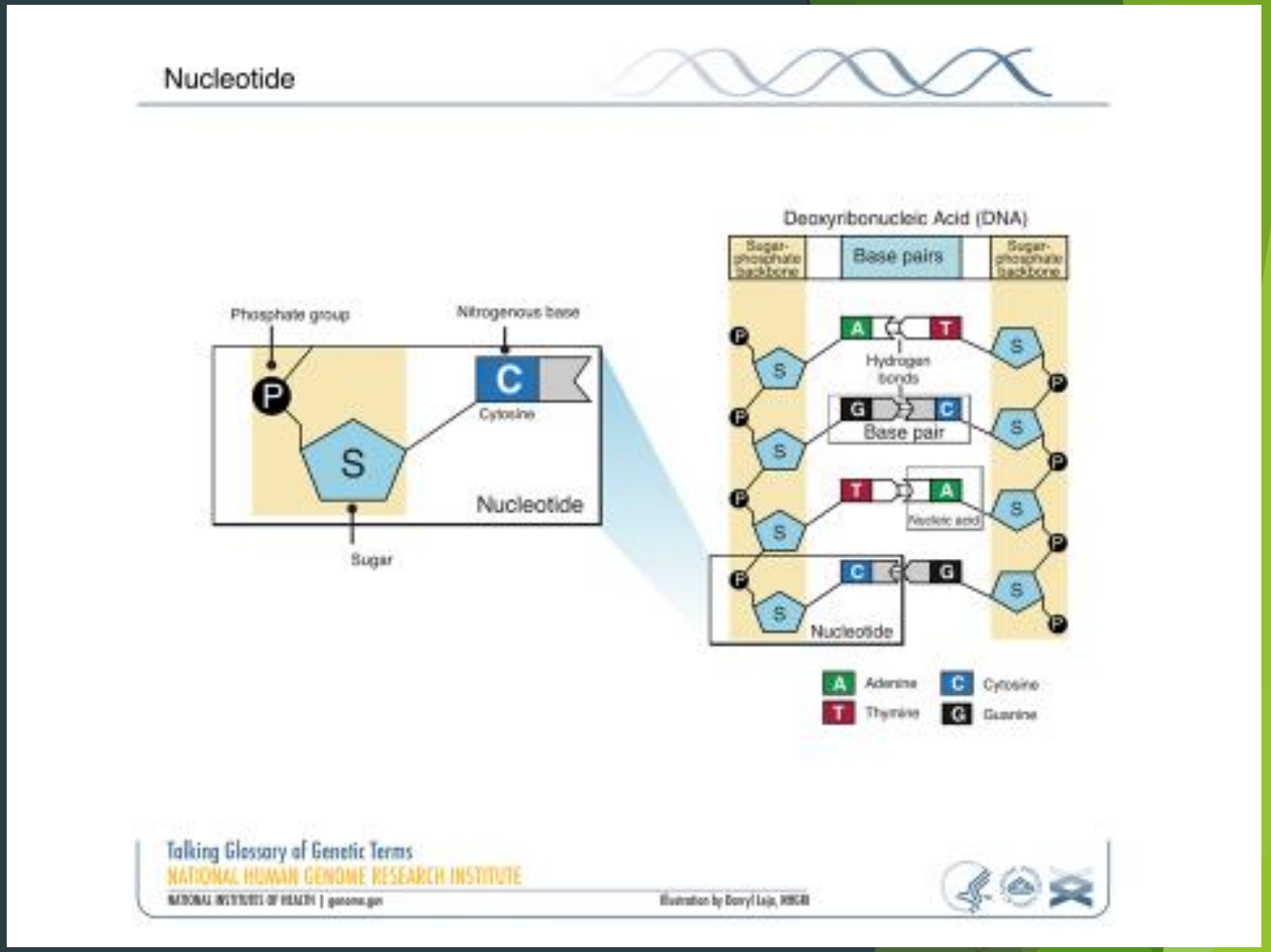
More recently, some small RNAs have been found to be involved in regulating gene expression.

Nucleotide


Phosphate group
Nitrogenous base
P
S
C
Cytosine
Nucleotide
Sugar

Deoxyribonucleic Acid (DNA)
Sugar-phosphate backbone
Base pairs
Sugar-phosphate backbone
A
T
Hydrogen bonds
G
C
Base pair
T
A
Nucleic acid
C
G
Nucleotide
Adenine
Cytosine
Thymine
Guanine

What is a Nucleotide?



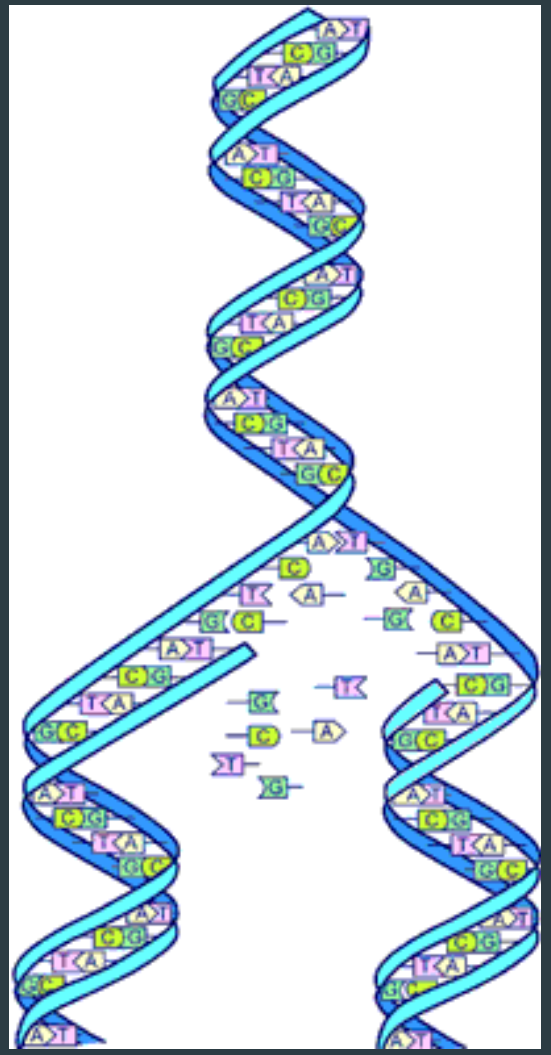
A nucleotide is the basic building block of nucleic acids.

RNA and DNA are polymers made of long chains of nucleotides.

A nucleotide consists of a sugar molecule (either ribose in RNA or deoxyribose in DNA) attached to a phosphate group and a nitrogen-containing base.

The bases used in DNA are adenine (A), cytosine (C), guanine (G), and thymine (T). In RNA, the base uracil (U) takes the place of thymine.

Human Genome Project (1990-2003)



DNA replication

Findings of the Human Genome Project

The Human Genome Project began in planning in 1984 and started in 1990 with goal of:

1. Sequencing and identifying all three billion chemical units in the human genetic instruction set
2. Finding genetic roots of disease and then developing treatments

It is considered a Mega Project because the human genome has approximately 3.3 billion base-pairs. With the sequence in hand, the next step was to identify the genetic variants that increase the risk for common diseases like cancer and diabetes

Key findings of the draft (2001) and complete (2004) genome sequences include:

1. There are approximately 20,500 genes in human beings, the same range as in mice
2. The human genome has significantly more segmental duplications (nearly identical, repeated sections of DNA) than other mammalian genomes. These sections may underline the creation of new primate-specific genes
3. At the time when the draft sequence was published fewer than 7% of protein families appeared to be vertebrate specific