

HUMAN GENETICS

DNA Structure

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General Objectives:

1. To define DNA and understand its significance as genetic material.
 2. To identify the basic components of a nucleotide (sugar, phosphate group, nitrogenous base).
 3. To differentiate between purines (Adenine, Guanine) and pyrimidines (Cytosine, Thymine).
 4. To explain the double-helix model of DNA and the principle of complementary base pairing (A-T, G-C).
 5. To understand how nucleotides are arranged to form genes that code for specific proteins.
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DNA Structure



In this Lecture, we'll briefly explore how the double-helical structure of DNA was discovered through the work of James Watson, Francis Crick, Rosalind Franklin, and other researchers. Then, we'll take a look at the properties of the double helix itself. From the work of biochemist Phoebus Levene and others, scientists in Watson and Crick's time knew that DNA was composed of subunits called nucleotides. A nucleotide is made up of a sugar (deoxyribose), a phosphate group, and one of four nitrogenous bases: adenine (A), thymine (T), guanine (G) or cytosine (C). C and T bases, which have just one ring, are called pyrimidines, while A and G bases, which have two rings, are called purines.

DNA Structure

- A gene is a section of DNA that codes for a **protein**.
 - Each unique gene has a unique sequence of bases.
 - This unique sequence of bases will code for the production of a unique protein.
 - It is these proteins and combination of proteins that give us a unique **phenotype**.
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DNA Structure : Deoxyribonucleic acid; a nucleic acid that consists of two long chains of nucleotides twisted together into a double helix and joined by hydrogen bonds between complementary bases adenine (A), and thymine (T), or cytosine (C), and guanine(G); it carries the cell's genetic information and hereditary characteristics via its nucleotides and their sequence and is capable of self replication and RNA synthesis.



Letters form words....

Words form sentences....

***endless
combinations**



***Think of the bases of DNA
like letters.***



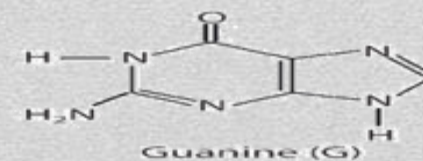
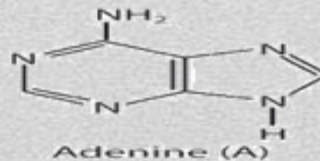
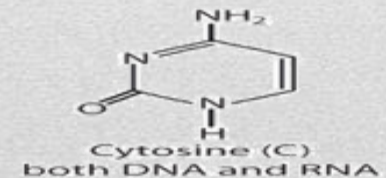
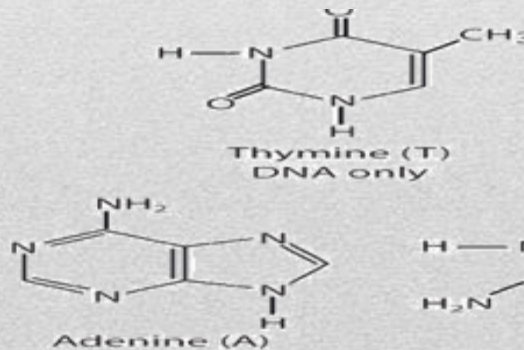
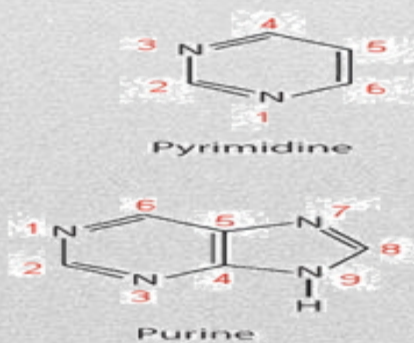
Nucleotide bases

DNA is a polynucleotide. Each nucleotide consists of a five carbon-sugar (deoxyribose).

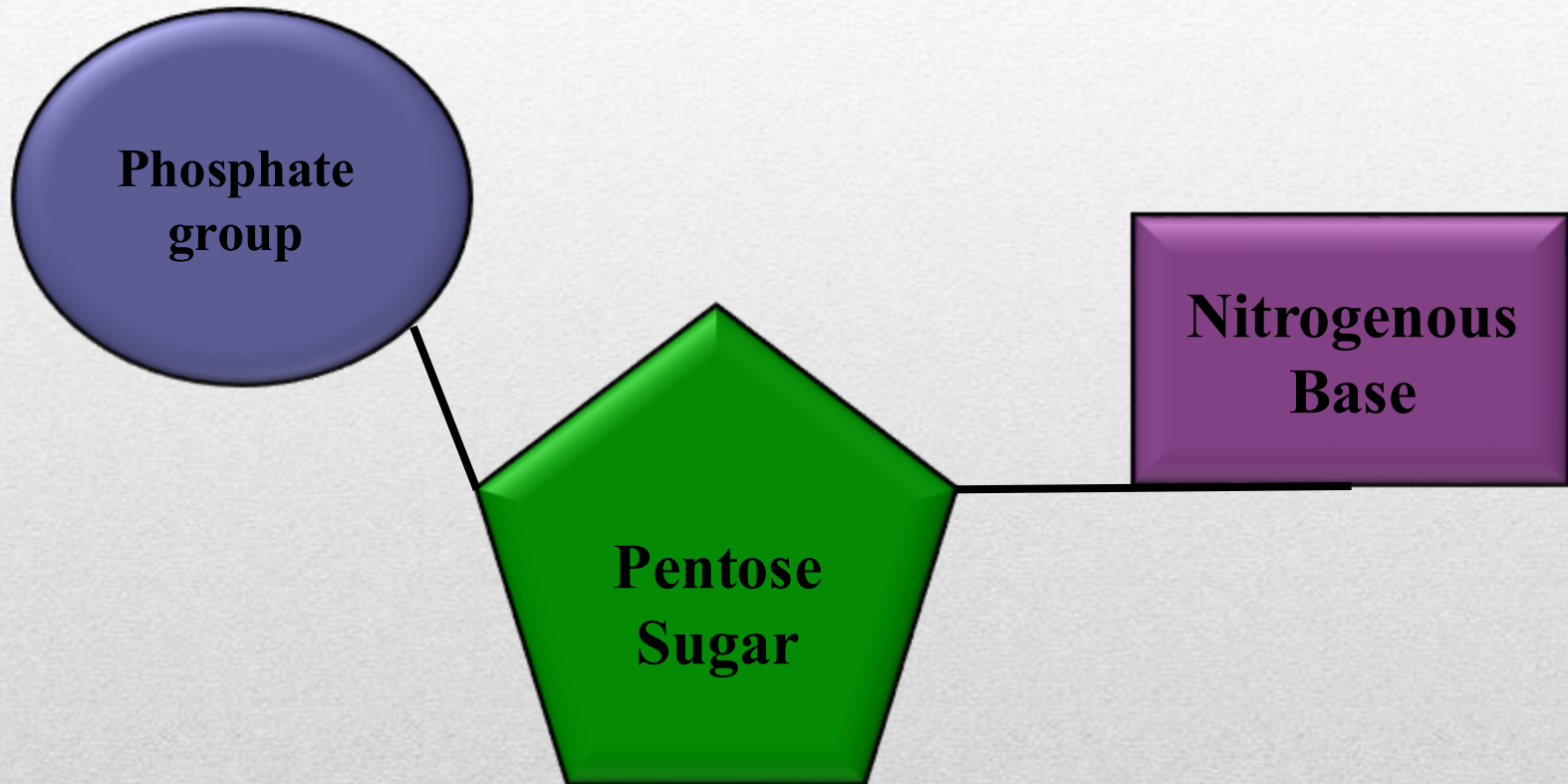
A nitrogen containing base attached to the sugar and a phosphate group. There are four different types of nucleotides founded in the DNA.

This four different types of nucleotides found in the DNA. Differing only in the nitrogenous base. The four nucleotides are Adenine (A), Guanine (G), Thymine (T) and Cytosine (C).

Adenine and Guanine are Purines. Cytosine and Thymine are Pyrimidines. Purines are larger than Pyrimidines. The deoxyribose sugar of the DNA backbone has 5 carbons and 3 oxygen's



Nucleotides



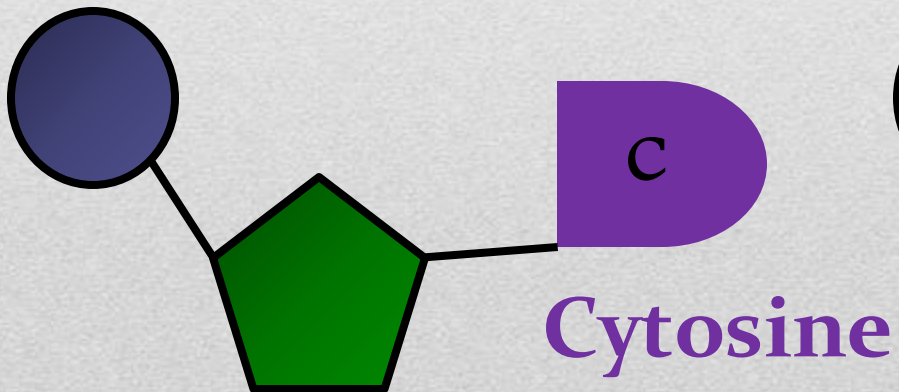
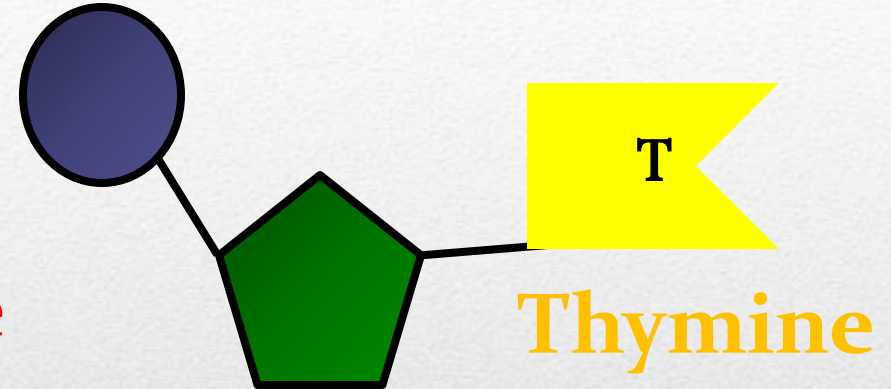
Nucleotides

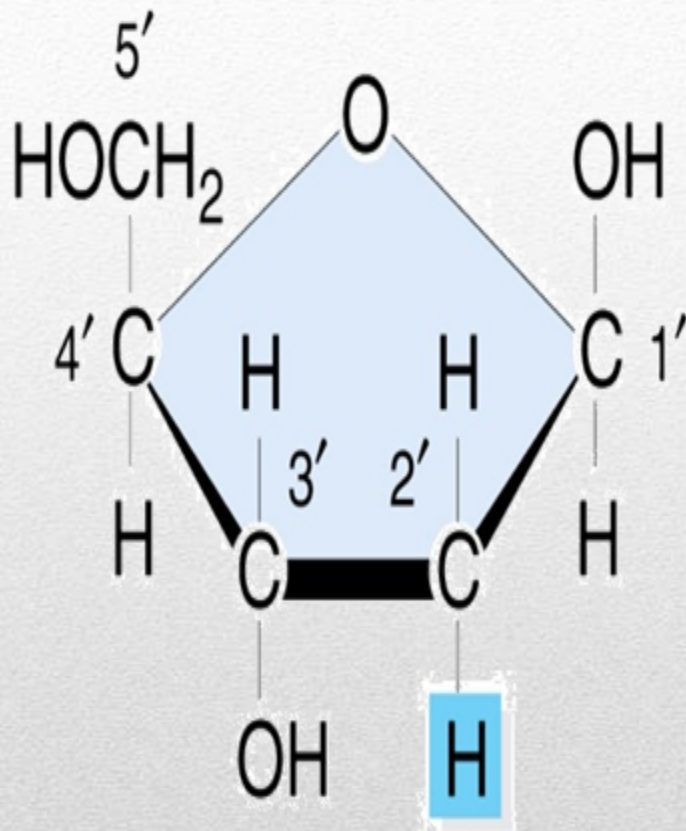
- The phosphate and sugar form the backbone of the DNA molecule, whereas the bases form the “rungs”.



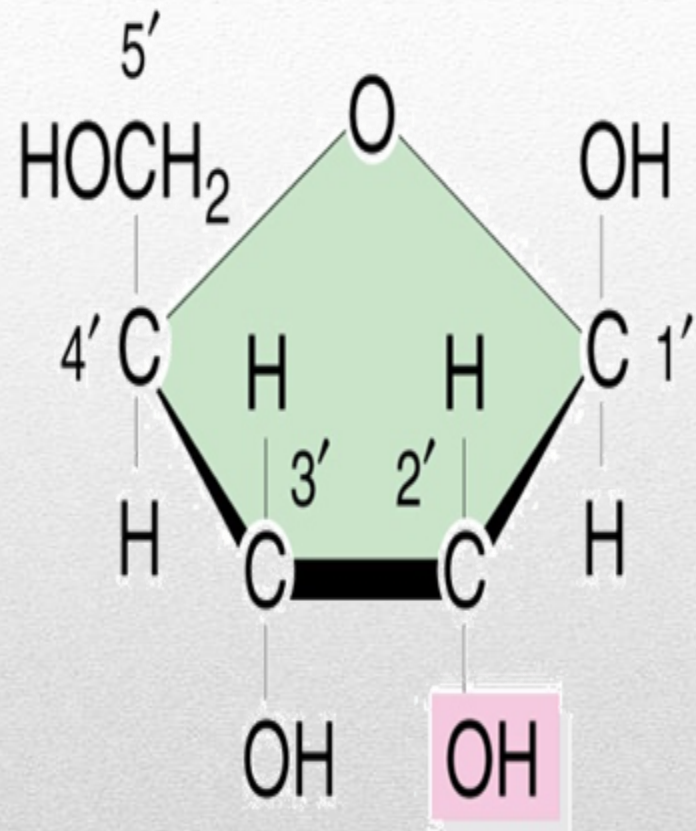
- There are four types of nitrogenous bases.
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Nucleotides



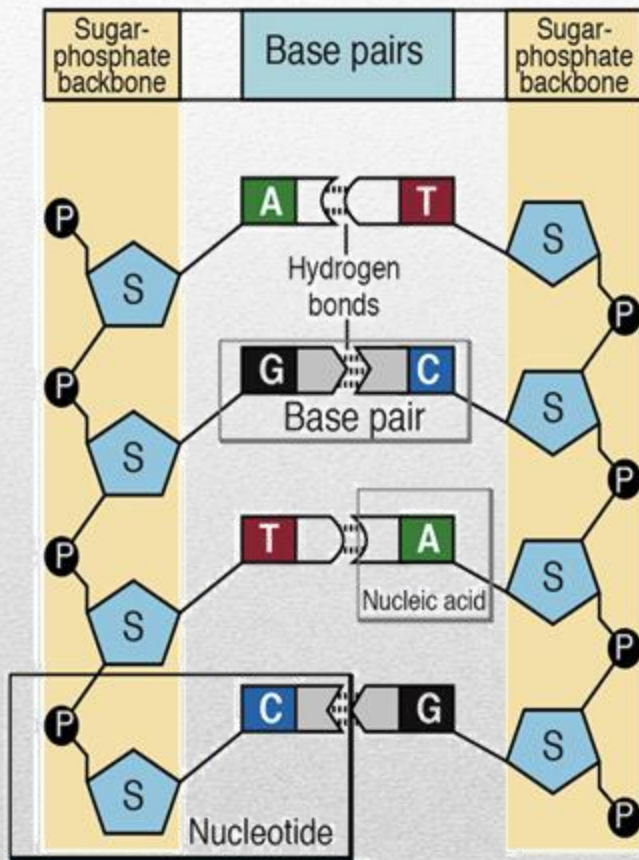


Deoxyribose

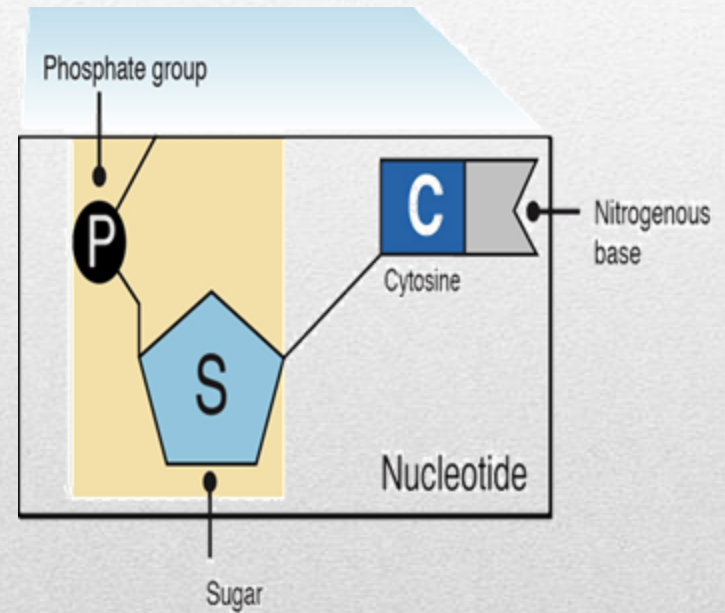


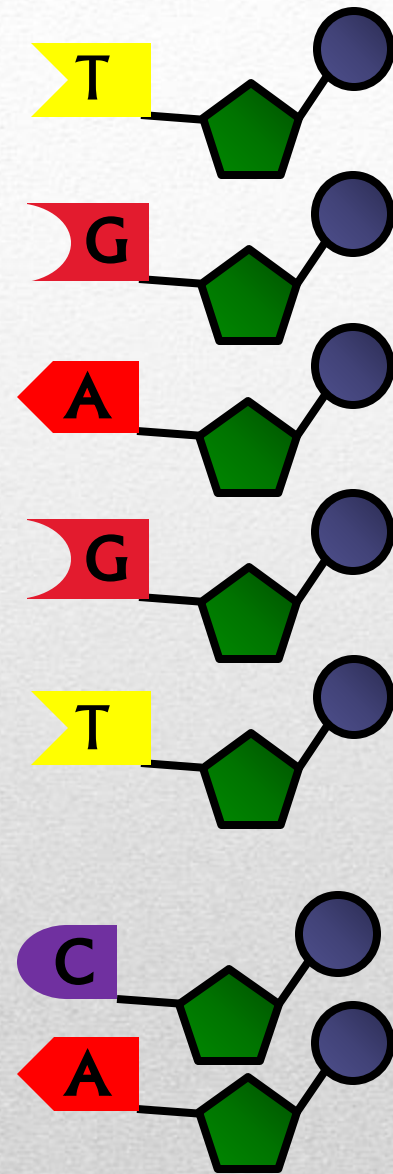
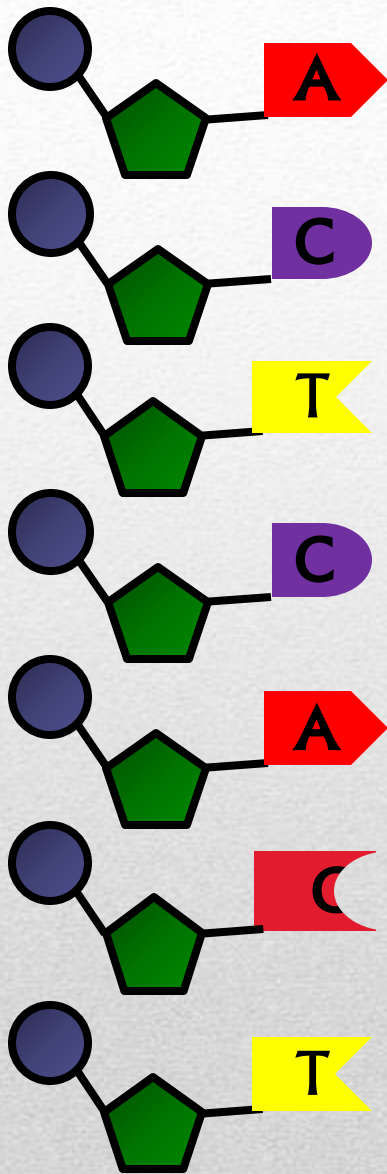
Ribose

Deoxyribonucleic Acid (DNA)



- A** Adenine
- T** Thymine
- C** Cytosine
- G** Guanine



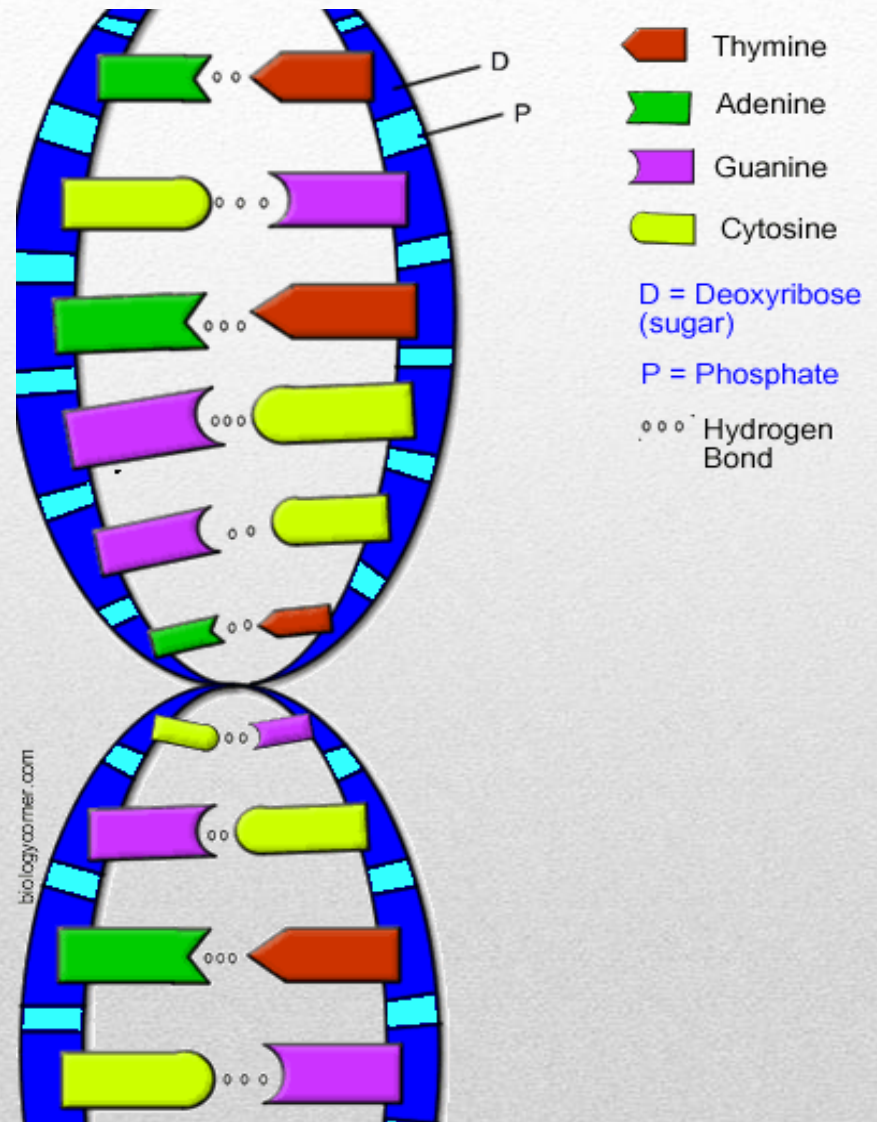


Base-Pair Rule

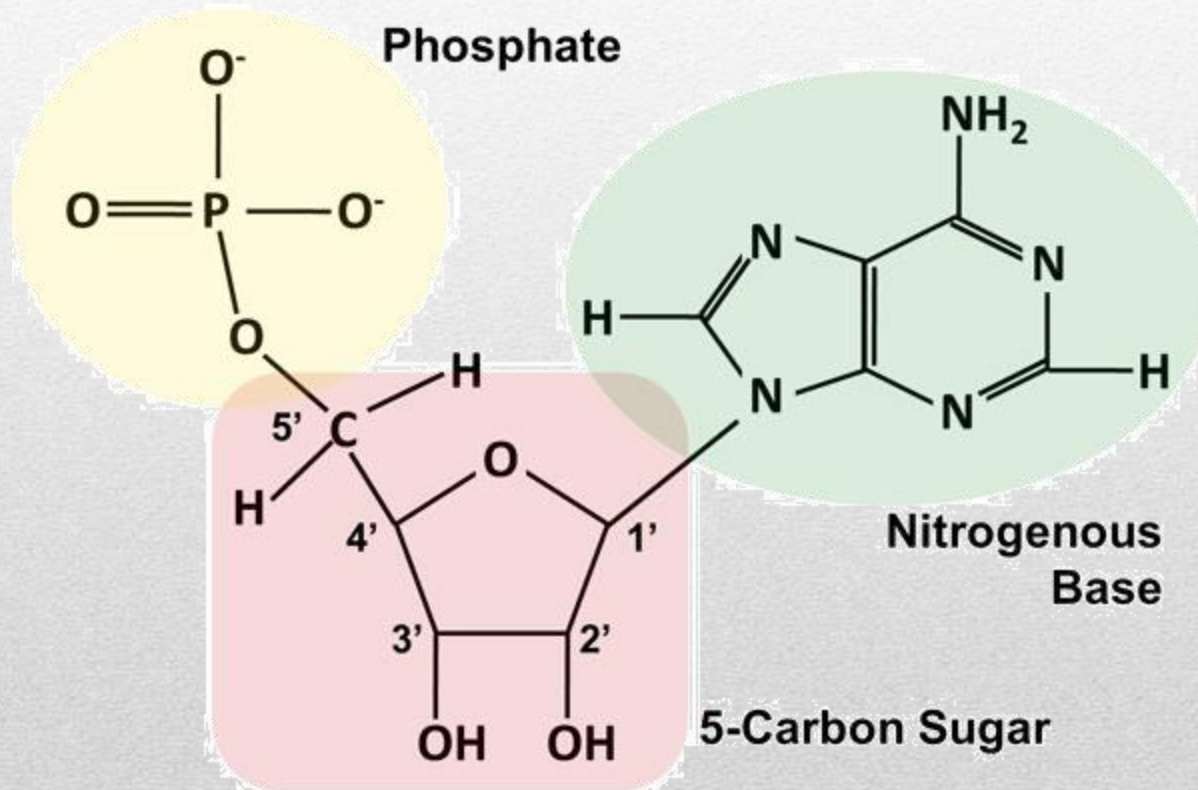
Adenine \rightleftharpoons Thymine

Guanine \rightleftharpoons Cytosine

The sides of the DNA ladder are phosphate & sugar held together by hydrogen bonds



The carbon atoms are numbered 1',2',3',4'. And 5'. The hydroxyl groups on the 5'- and 3'- carbon link to the phosphate groups to form the DNA backbone. The deoxyribose sugars are joined at both hydroxyl groups to phosphate groups in ester links. Known as “phosphodiester” bonds.



Nucleotides

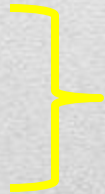
- Each base will only bond with **one** other specific base.

- Adenine (A)
- Thymine (T)



Form a base pair.

- Cytosine (C)
- Guanine (G)

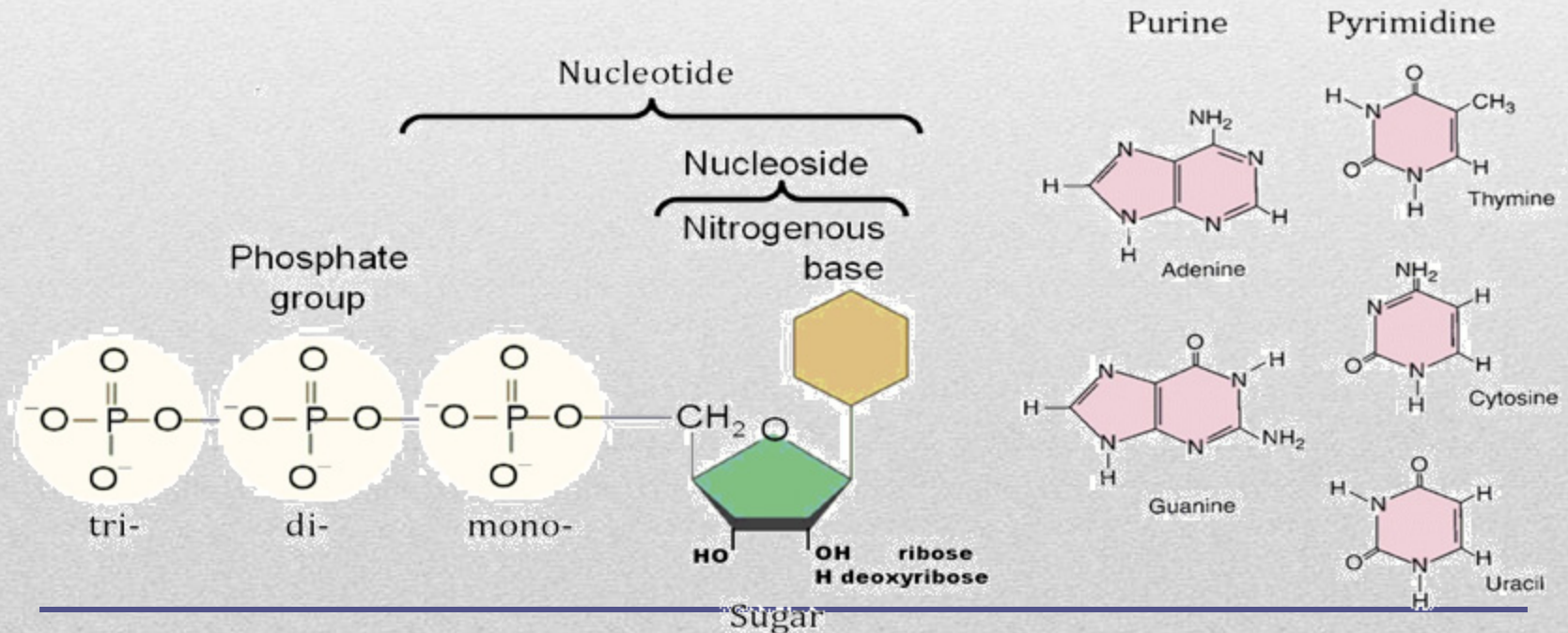


Form a base pair.

Nucleosides and Nucleotides

A nucleoside is one of the four DNA base covalently attached to C1' position of the sugar. The sugar in deoxynucleosides is 2'-deoxyribose. The sugar in ribonucleosides is ribose.

A nucleotide is a nucleoside with one or more phosphate groups covalently attached to the 3'- and/or 5'-hydroxyl group(s).



DNA double strand (Double Helix)

- The DNA is normally a double stranded macromolecular. Two polynucleotide chains, held together by weak thermodynamic force.
 - Two DNA strands form a helical spiral, winding around a helix axis in a right-handed spiral.
 - The two polynucleotide chains run in opposite directions.
 - The sugar-phosphate backbone of the two DNA strands wind around the helix axis like railing of a spiral staircase.
 - The bases of the individual nucleotides are on the inside of the helix, stacked on top of each other like the steps of a spiral staircase.
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Base pairing

In Watson and Crick's model, the two strands of the DNA double helix are held together by hydrogen bonds between nitrogenous bases on opposite strands. Each pair of bases lies flat, forming a "rung" on the ladder of the DNA molecule.

Base pairs aren't made up of just any combination of bases. Instead, if there is an A found on one strand, it must be paired with a T on the other.

Similarly, an G found on one strand must always have a C for a partner on the opposite strand. These A-T and G-C associations are known as **complementary base pairs**.

Base Pair Rule

One side: A T A T C A T G C G G G

Other side: T A T A G T A C G C C C

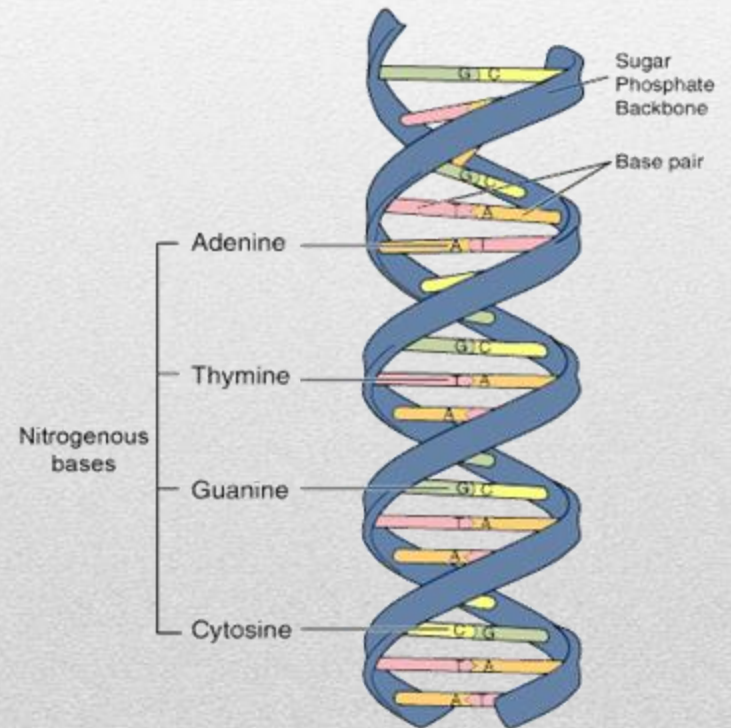


Image adapted from: National Human Genome Research Institute.

Base pairing explains Chargaff's rules, that is, why the composition of A always equals that of T, and the composition of C equals that of G. Where there is an A in one strand, there must be a T in the other, and the same is true for G and C. Because a large purine (A or G) is always paired with a small pyrimidine (T or C), the diameter of the helix is uniform, coming in at about 22 nanometers. Although Watson and Crick's original model proposed that there were two hydrogen bonds between the bases of each pair, we know today that G and C form an additional bond (such that A-T pairs form two hydrogen bonds total, while G-C pairs form three).

