

MOLECULAR GENETICS

Genes, DNA & Nucleic Acids

1. Gene functions

- To be preserved and transmitted
- To control various biological functions through the production of proteins (i.e., large, complex sequences of amino acids) and RNA

2. Gene structure is based on two nucleic acids:

- Deoxyribonucleic acid (DNA)
- Ribonucleic acid (RNA)

3. Nucleotides: The components of nucleic acids; made of three subunits:



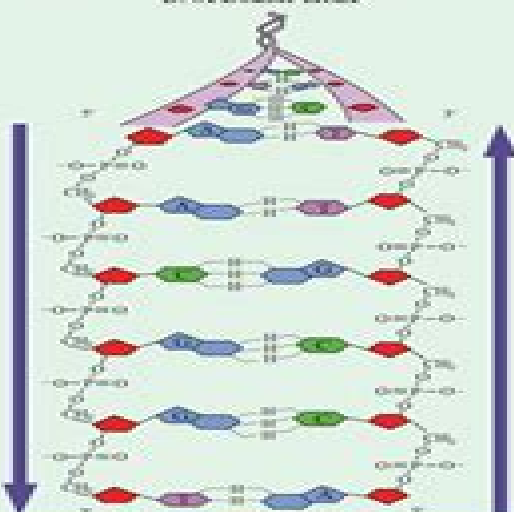
A. Sugar (deoxyribose in DNA; ribose in RNA)

B. Phosphate

C. Nitrogenous base (one of five possible bases)

- In DNA, the nucleic acid of chromosomes, four nitrogenous bases are found: adenine (A), guanine (G), cytosine (C), and thymine (T)
- RNA consists of similar bases, except uracil (U) replaces thymine (T)
- DNA is a double helix molecule; that is, it is similar to a spiral staircase or twisted ladder, with the sides formed by repeating sugar-phosphate groups from each nucleotide, and the horizontal portions (i.e., steps) formed by hydrogen bonds involving A with T or C with G
- Hereditary information: Genes found along the linear sequence of nucleotides in the DNA molecule

DNA Double Helix



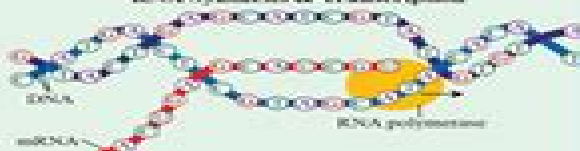
The Central Dogma

1. **Replication:** DNA is copied from other DNA by unsplicing the helix and pairing new nucleotides with the proper bases (i.e., A with T and C with G) on each separated side of the original DNA

2. Transcription

- Messenger RNA (mRNA) is copied from DNA by unsplicing a portion of the DNA helix that corresponds to a gene
- Only one side of the DNA will be transcribed, and nucleotides with the proper bases (A with U and C with G) will be sequenced to build pre-mRNA
- Segments of nucleotides called introns are removed, and the remaining segments called exons are spliced together
- The mature mRNA leaves the nucleus to be translated by the ribosomes

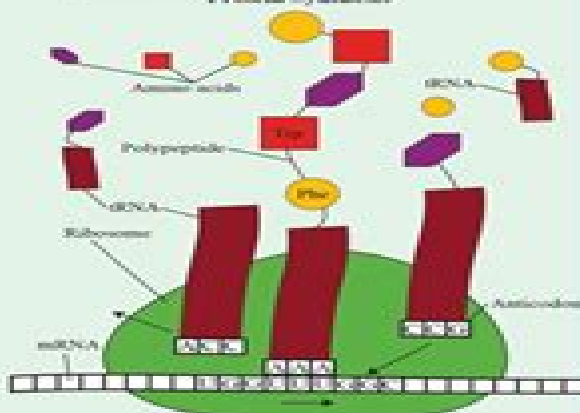
RNA Synthesis & Transcription



3. Translation

- Proteins are synthesized from mRNA by ribosomes (which are composed of ribosomal RNA, or rRNA, and proteins), which read from a universal triplet code (i.e., codons)
- The ribosomes instruct transfer RNAs (tRNAs) to bring specific amino acids in the sequence dictated by the mRNA, which in turn was built based on the sequence of nucleotides in the original gene portion of the DNA

Protein Synthesis



Mutations

A **mutation** is any random, permanent change in the DNA molecule; many mutations are harmful, some have no effect, and a few actually benefit the organism; nature selects those mutations that are beneficial or adaptive in organisms to help shape the course of evolution

POPULATION GENETICS

Genes in Populations

vs. Individuals

- Populations evolve just as species do
- Genotype:** Genetic composition of an individual
- Gene pool:** Genetic composition of a population of individuals (i.e., all alleles for all genes in a population)
- Evolution involves changes in gene pools over time; to understand changes in gene pools as populations evolve, an understanding of non-evolving populations is necessary

The Hardy-Weinberg Law

- Both allelic frequencies and genotypic ratios (i.e., gene pools) remain constant from generation to generation in sexually reproducing populations if the following conditions of equilibrium exist:

- Mutations do not occur
- There is no net movement of individuals out of or into a population
- All offspring produced have the same chances for survival, and mating is random (i.e., no natural selection occurs)
- The population is so large that chance would not alter frequencies of alleles

2. Algebraic equivalent of the Hardy-Weinberg law:

$$p^2 + 2pq + q^2 = 1, \text{ where:}$$

p = frequency of dominant allele

q = frequency of recessive allele

p^2 = AA genotype

$2pq$ = Aa genotype

q^2 = aa genotype

Example

- If a group of six individuals has nine dominant (A) alleles and three recessive (a) alleles, then $p = 9/12$ or 0.75 and $q = 3/12$ or 0.25 ; a total of 12 gametes will be produced, 9 of which will have the dominant allele and 3 of which will have the recessive allele
- Use the equation to predict the ratios of the three possible genotypes as a result of fertilization:
 - Frequency of AA genotypes is p^2 , or $(0.75)^2 = 0.56$
 - Frequency of Aa genotypes is $2pq$, or $2(0.75)(0.25) = 0.38$
 - Frequency of aa genotypes is q^2 , or $(0.25)^2 = 0.06$
- The frequencies of dominant and recessive alleles are still the same, but the specific alleles have been redistributed

Hardy-Weinberg & Natural Populations

- Few (if any) populations are in equilibrium; therefore, changes in allelic frequencies and thus gene pools do occur in natural populations
- The Hardy-Weinberg law helps identify the mechanisms of these evolutionary changes by predicting that one or more of the four conditions required are not met; that is, in natural populations:
 - Mutations occur
 - Individuals leave and enter populations
 - Nonrandom mating and natural selection occur
 - Small populations exist

Author: Randy Brooks, PhD

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