

Chapter 1: Human Genome

1.1. INTRODUCING GENES AND GENOME

- **Genetics** – study of heredity, traits and their variations.
 - study of how traits are transmitted.
- **Heredity**: transmission of traits and biological information.
- **Genes**: basic unit of heredity.
 - inherit cell's production of proteins.
 - consist of long DNA.
 - protein-encoding genes are scattered among 3.2 billion DNA bases in each set of chromosomes.
- **Proteins** – control the characteristics of every individual.
- **DNA (Deoxyribonucleic Acid)** – hereditary information is packaged in nitrogenous bases.
- **Genomes** – made up of complete set of genetic instructions characteristic of each cell organism.
 - all of our cells contain two sets copies of genomes.
- **Genetics** – scientific field that analyzes genomes.
 - reveals how closely related are different species.

1.2 LEVELS OF GENETICS AND GENOMICS

- **DNA (Deoxyribonucleic Acid)**
 - Shape: double helix
 - Consist of: phosphate sugar and phosphates
 - Function: provides information to manufacture specific proteins



- 4 types of nitrogenous bases:
 - Adenine (A) and Thymine (T)
 - Cytosine (C) and Guanine (G)
 - Each set of these complementary bases is a code for a particular amino acid.
- **Amino Acid** – building blocks of protein.
 - align and link like-into bonds forming protein.

1.3. THREE PROCESSES OF ACQUIRED GENETIC INFORMATION

- **DNA replication** – (1) chains of double helix untwist and separate.
 - (2) each part builds a new partner chain from free DNA bases.
- **Transcription** – (3) copies the sequence of one strand of DNA molecule into mRNA.
- **Translation** – (4) each 3 DNA bases in a row attract another type of RNA, that functions as a connector, bringing in a particular amino acid.
 - (5) Amino acids align and link to form protein.

1.2 THE HUMAN GENOME

- **20,125 protein-encoding genes** comprise the genome.
 - Accounts: 1.5 percent of human genome, 8.5 percent of known genetic diseases.
- **Alleles** ("gene variants") – some proteins – encoding genes may vary in DNA base sequence from parent to parent that is passed to the next generation.
- **Mutations** ("changed gene") – alteration or change in genes, causing a disease.

1.1. MUTATION DISEASES

- **Cystic Fibrosis** – inherited disease
 - Change of a "C" to "T" inserts amino acid rather than the amino acid glutamic which results in protein becoming abnormal.
 - **Cystic Fibrosis Transmembrane**

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