

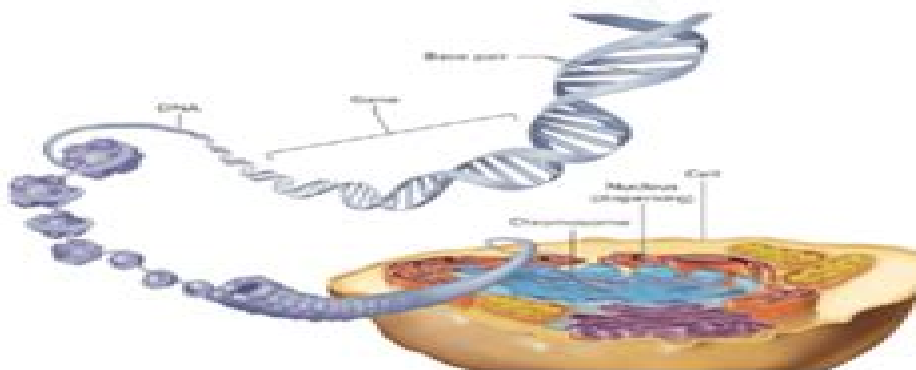
Chapter 1: Human Genome

1.1: INTRODUCING GENES AND GENOME

- **Genetics** – study of heredity, traits and their variation.
 - study of how traits are transmitted.
- **Heredity**- transmission of traits and biological information.
- **Genes**- basic unit of heredity.
 - instruct 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
- **Protein**- control the characteristics of every individual.
- **DNA (Deoxyribonucleic Acid)** - transmits information in sequence or **nitrogenous bases**.
- **Genome** – made up of complete set of genetic instructions characteristics of short an organism.
 - all of our cells contain two arm copies of genome.
- **Genomics** – scientific field that analyses genome.
 - reveals how closely related are different species.

1.2 LEVELS OF GENETICS AND GENOMICS

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



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

1.3. THREE PROCESSES OF ACCESSING GENETIC INFORMATION

- **DNA replication**- (1) chains of double helix untwist and separate.
 - (2) each half 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 RNA 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

- **20,325 protein- encoding genes**- comprise the exome.
 - Accounts: **1.5 percent** of human genome, **85 percent** of known genetic diseases
- **Alleles** ("gene variants")-same protein- encoding genes may variate in DNA base sequence from person to person that is passed to the next generation.
- **Mutation** (" changed gene")
 - alteration or change in genes, causing a disease.

1.3. MUTATION DISEASES

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

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