#### 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 aids align and link to form protein.

#### 1.2 THE HUMAN GENOME

- 20,325 protein- encoding genescomprise 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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Kerry L. Hull, Barbara Janson Cohen

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