

B.Sc. (Hons) Microbiology
IIIrd Year

SEMESTER: VI

ADVANCES IN MICROBIOLOGY

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Salient Features of Human Genome:

- ❑ Human genome consists the information of **24 chromosomes** (22 autosome + X chromosome + one Y chromosome); in *Homo sapiens* $2n = 2x = 46$
- ❑ The human genome contains **over 3 billion nucleotide pairs**.
- ❑ Human genome is estimated to have **about 30,000 genes**.
- ❑ Average gene consists **of 3000 bases**. But sizes of genes vary greatly, with the largest known human gene encoding dystrophin containing 2.5 million base pairs.
- ❑ Only about **3 %of the genome** encodes amino acid sequences of polypeptides and **rest of it junk (repetitive DNA)**.
- ❑ The **functions are unknown** for over **50%** of the discovered genes.

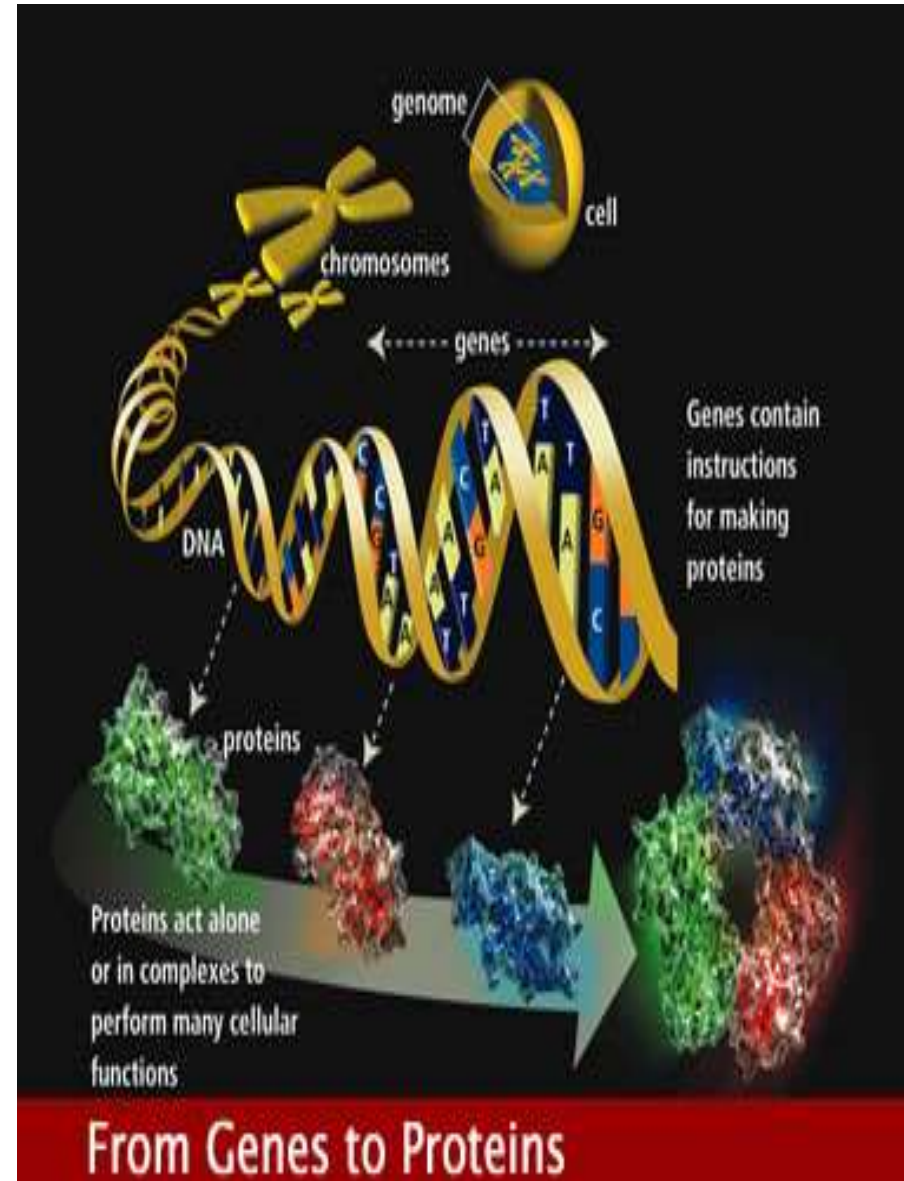
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- ❑ The repetitive sequences makeup very large portion of human genome. Repetitive sequences have no direct coding function but they shed light on the chromosome structure, dynamics and evolution.
- ❑ Chromosome **1 has most genes (2968)** and **Y chromosome has the lowest (231)**.
- ❑ Almost all nucleotide bases are exactly the same in all people. Genome sequences of different individuals differ for **less than 0.2% of base pairs**.

Most of these differences occur in the form of single base differences in the sequence. These single base differences are called **single nucleotide polymorphisms (SNPs)**. One SNP occurs at every $\sim 1,000$ bp of human genome. About 85% of all differences in human DNAs are due to SNPs.

What was Human Genome Project(HGP)

- The Human Genome Project was an international research effort to determine the **sequence of the human genome** and identify the **genes that it contains**.
- The US Human Genome Project is a 13 year effort, which is coordinated by the
 - **Department of Energy (DOE) and**
 - **National Institutes of Health (NIH).**



Milestones

- 1986 The birth of the Human Genome Project.
- 1990 Project initiated as joint effort of US Department of Energy and the National Institute of Health.
- 1994 Genetic Privacy Act: to regulate collection, analysis, storage and use of DNA samples and genetic information is proposed.
- 1996 Wellcome Trust joins the project.
- 1998 **Celera Genomics** (a private company founded by Craig Venter) formed to sequence much of the human genome in 3 years.
- 1999 Completion of the sequence of Chromosome 22-the first human chromosome to be sequenced.
- 2000 Completion of the working draft of the entire human genome.
- 2001 Analysis of the working draft are published.
- 2003 **HGP sequencing is completed and Project is declared finished two years ahead of schedule.**

Goals of Human Genome Project

1. To identify all the genes in human DNA.
2. To develop a genetic linkage map of human genome.
3. To obtain a physical map of human genome.
4. To develop technology for the management of human genome information.
5. To know the function of genes.
6. Determine the sequences of the 3 billion chemical base pairs that make up human DNA.
7. Store this information in public databases.
8. Develop tools for data analysis.
9. Transfer related technologies to the private sectors.