

□ Study Material: Human Genome Project (HGP)

Class 12 Biology — Molecular Basis of Inheritance

1. What is the Human Genome Project?

The **Human Genome Project (HGP)** was an international scientific project started in **1990** to determine:

- The complete sequence of **human DNA**
- The number and location of **human genes**
- The organisation of the human genome
- The identification of genetic variations

The project was declared **complete in 2003**.

□ Remember:

HGP = Human Genome → Genes + DNA sequence + Genome organisation

2. Main Goals of HGP

The major goals were to:

1. **Identify all the approximately 20,000–25,000 human genes.**
2. Determine the sequence of approximately **3 billion base pairs** in human DNA.
3. Store the information in **databases**.
4. Improve tools for **data analysis**.
5. Transfer related technologies to industry.
6. Address the **ethical, legal and social issues (ELSI)** arising from the project.



3. Major Findings of HGP

① Human genome contains about 3 billion base pairs

The human genome has approximately:

3×10^9 base pairs

② Number of genes

Humans have approximately **20,000–25,000 genes**.

Earlier estimates were much higher, but genome sequencing showed that the number of protein-coding genes is much smaller than expected.

③ **Less than 2% of the genome codes for proteins**

Only a **small fraction** of the human genome consists of sequences that directly code for proteins.

④ **Humans are genetically very similar**

About **99.9% of the DNA sequence is the same among humans**.

The remaining variation contributes to differences among individuals.

□ **4. Important Features of Human Genome**

Feature	Important Fact
Total genome size	~3 billion bp
Genes	~20,000–25,000
Protein-coding portion	< 2%
Human genetic similarity	~99.9%
Chromosomes	23 pairs
Genome sequencing began	1990
HGP completed	2003

🔍 **5. Important Discoveries**

◆ **Chromosome 1**

It contains the **largest number of genes** among human chromosomes.

◆ **Y chromosome**

It contains the **fewest genes** among human chromosomes.

◆ **Repetitive DNA**

A large proportion of the human genome consists of **repetitive DNA sequences**.

These sequences may occur many times in the genome.

□ 6. Techniques Used in HGP

Two important approaches were used:

A. Expressed Sequence Tags (ESTs)

ESTs identify genes that are **expressed** as RNA.

EST → Expressed genes

B. Sequence Annotation

The genome sequence was analysed to identify:

- Genes
- Regulatory sequences
- Repetitive DNA
- Other important regions

7. Bioinformatics

The enormous amount of information generated by HGP required computers for:

- Storage
- Organisation
- Analysis
- Comparison of DNA sequences

This field combining **biology + computer science + information technology** is called **bioinformatics**.

□ Memory Trick:

HGP → Huge Data → Computers → Bioinformatics

8. ELSI

One important component of HGP was the study of:

Ethical, Legal and Social Issues (ELSI)

Examples:

- Privacy of genetic information
- Genetic discrimination

- Ownership of genetic information
 - Misuse of genetic data
 - Ethical concerns related to genetic testing
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☆ 9. Why is HGP Important?

HGP has helped scientists to:

- Understand human genetic variation
- Study genetic diseases
- Identify disease-associated genes
- Improve genetic diagnosis
- Develop new approaches to disease treatment
- Understand human evolution
- Advance personalised medicine

CBSE Exam Points

2-Mark Question

What were the two major goals of the Human Genome Project?

Answer:

1. To identify all the genes present in human DNA.
 2. To determine the complete sequence of approximately 3 billion base pairs of human DNA.
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3-Mark Question

Mention three important features of the human genome.

Answer:

- Human genome contains approximately **3 billion base pairs**.
- It contains approximately **20,000–25,000 genes**.
- Less than **2%** of the genome codes for proteins.

3-Mark Question

What is the significance of ELSI in HGP?

Answer:

ELSI stands for **Ethical, Legal and Social Issues**. It addresses concerns related to the use of genetic information, such as **privacy, genetic discrimination, ownership and possible misuse of genetic data**.

Last-Minute Revision

HGP = 1990–2003

3 billion bp

20,000–25,000 genes

<2% protein-coding

99.9% human DNA similar

EST → expressed genes

Bioinformatics → data analysis

ELSI → Ethical + Legal + Social Issues