



## Mains Practice Question

**Q.** How has the Human Genome Project contributed to our understanding of genetic diseases and the development of targeted therapies? (150 words)

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### Approach

- Start with a brief introduction that explains what the Human Genome Project (HGP) was.
- List some of the ways that the HGP has contributed to our understanding of genetic diseases and the development of targeted therapies.
- You can conclude with a summary of the main points and a mention of some of the challenges that the HGP has raised for the future of medicine and society.

### Introduction

The Human Genome Project (HGP), which was officially launched in 1990 and completed in 2003, has had a profound impact on our understanding of genetic diseases and the development of targeted therapies. It was a landmark scientific endeavor that aimed to sequence the entire human genome and identify all the genes that encode the instructions for life.

### Body

The HGP has contributed to our understanding of genetic diseases and the development of targeted therapies in several ways, such as:

- It **enabled the discovery of many disease-causing variants in the human genome**, which can be used for diagnosis, prognosis, and risk assessment of various genetic disorders.
  - For example, the HGP helped identify the faulty alleles that are associated with cystic fibrosis, breast cancer, Huntington's disorder, and many other conditions.
- It **facilitated the development of somatic gene therapies, which involve modifying the DNA of a patient's cells to treat or cure a disease**. Somatic gene therapies have been successfully used to address HIV, sickle-cell disease, and transthyretin amyloidosis, and could potentially improve the treatment of many cancers and other diseases.
- It **provided the basis for the design of targeted therapies, which are drugs or other agents that specifically target the molecular defects that drive a disease**. Targeted therapies can be more effective and less toxic than conventional therapies that affect healthy cells as well as diseased ones.
  - For example, the HGP helped identify the genomic alterations that are involved in various types of cancer, and enabled the development of drugs that target these alterations.
- It **paved the way for the use of next-generation sequencing technologies, which can rapidly and accurately sequence the DNA** of individual patients or tumors, and reveal their unique genomic profile.
  - This information can be used to tailor the diagnosis, treatment, and prevention of diseases according to the patient's genetic makeup.

### Conclusion

The HGP was a remarkable achievement that has transformed our knowledge of human biology and disease. It has also raised new ethical, social, and legal challenges that need to be addressed by the scientific community and society at large. The WHO has issued new guidelines for overseeing human genome editing to ensure its safe, effective, and ethical use for the benefit of all.

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