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Q8(b)

Define genetic polymorphism. Give details of its types with suitable examples

UPSC Civil Services Mains 2015 · Anthropology GS 1 · 15 marks · Concept of genetic polymorphism and selection, Mendelian population, Hardy-Weinberg law

9.3 Concept of genetic polymorphism and selection, Mendelian population, Hardy-Weinberg law; causes and changes which bring down frequency-mutation, isolation, migration, selection, inbreeding and genetic drift. Consanguineous and non-consanguineous mating, genetic load, genetic effect of consanguineous and cousin marriages.

Core demand of the question

- Define genetic polymorphism. Give details of its types with suitable examples
- Write about 200 words for 15 marks

Revision summary

Polymorphism is two or more common alleles at a locus.

Balanced types include sickle cell and malaria.

ABO, Rh, and HLA are serological polymorphisms.

SNPs and STRs are DNA types used in medicine and forensics.

Most human genetic variation is polymorphic within populations.

Introduction

Genetic polymorphism is the stable presence of two or more alleles at a locus, each at a frequency too high to be only recurrent mutation. A common teaching cut-off is 1 percent.

Body

Definition

- **Ford** used polymorphism for discrete forms in a population. In humans it now covers blood groups, enzymes, and DNA sites.
- It is not a rare disease allele. It is a normal variation that population genetics can count.

Types and examples

- **Balanced polymorphism:** heterozygote advantage. **Sickle haemoglobin** in malaria belts, shown by **Allison**, and **thalassaemia** in parts of India and the Mediterranean.
- **Transient polymorphism:** a rising advantageous allele not yet fixed, or a falling one.
- **Chromosomal polymorphism:** inversions in *Drosophila* were the classic; humans show copy-number and inversion variants too.
- **Protein / serological:** **ABO**, **Rh**, **HLA**, **G6PD**.
- **DNA polymorphism:** **SNPs**, **STRs** used in forensics, **Alu** insertions.
- **Restricted vs widespread:** PTC tasting, lactase persistence as gene-culture polymorphism.

Meaning

- Polymorphism feeds **Harris** enzyme work and **Lewontin**'s point that most variation is within groups.

Flow diagram

Genetic polymorphism -> Balanced sickle

Genetic polymorphism -> SNP STR

Genetic polymorphism -> ABO Rh HLA

Conclusion

Genetic polymorphism is common allelic variation. Types run from balanced sickle-cell systems to SNPs and STRs, and they are the raw material of human diversity.

Facts & figures

Allison

Linked sickle-cell heterozygotes to malaria protection in East Africa.

ABO

The first widely typed human polymorphism, still used in transfusion.

STR

Forensic multiplex markers; a DNA polymorphism panel.

Lewontin

Showed that most allozyme diversity is within, not between, classical races.