Q6(a)

What are genetic markers? Discuss their applications in understanding population variation, disease association and forensics

UPSC Civil Services Mains 2025 · Anthropology GS 1 · 20 marks · Applications of Anthropology

12. Applications of Anthropology: Anthropology of sports, Nutritional anthropology, Anthropology in designing of defence and other equipments, Forensic Anthropology, Methods and principles of personal identification and reconstruction, Applied human genetics-Paternity diagnosis, genetic counselling and eugenics, DNA technology in diseases and medicine, serogenetics and cytogenetics in reproductive biology.

Core demand of the question

- Define genetic markers
- Discuss applications in population variation
- Discuss applications in disease association
- Discuss applications in forensics
- Give named examples

Revision summary

A genetic marker is a heritable polymorphism that can be read in families or populations.

Classical markers include ABO and proteins. DNA markers include SNPs, STRs, mtDNA and Y haplotypes.

Population applications map variation and movement and undermine typological race.

Disease applications include sickle-cell and malaria, HLA, counselling, and GWAS.

Forensic applications include STR identification, mtDNA from degraded samples, and Y-lineage in mixtures.

Lewontin's within-group diversity and Jeffreys's DNA fingerprinting are central names.

Introduction

A genetic marker is a measurable hereditary character that differs among individuals and can be followed in families or populations. It may be a classical blood-group antigen, a protein variant, or a DNA site such as a SNP or STR. Markers are tools. They are not the same thing as a 'race gene'.

Body

What counts as a marker

Classical markers include ABO and Rh blood groups, HLA types, and protein polymorphisms. **Karl Landsteiner's** ABO system is still the first standard example. **DNA markers now dominate: single-nucleotide polymorphisms (SNPs), short tandem repeats (STRs), mitochondrial haplogroups, and Y-chromosome haplotypes. Alec Jeffreys** made

hypervariable DNA useful as a fingerprint. **L. L. Cavalli-Sforza** organised classical and then molecular markers into maps of population history. **Richard Lewontin** used similar variation to show that most genetic diversity is within groups, not between continental labels.

- **A good definition:** a marker is polymorphic, heritable, and readable in the lab or the pedigree.

Population variation

- **Markers describe structure:** allele frequencies, genetic distance, and clines. They helped replace typological race with population thinking. Indian anthropology used ABO, MN, PTC and later DNA to study caste, tribe and regional diversity as overlapping frequencies, not as sealed types. mtDNA and Y-chromosome markers add sex-specific histories of movement. The ethical warning is the same as for mtDNA: a haplogroup is not a civilisation.

Disease association

Some markers sit in or near genes that affect disease. Sickle-cell haemoglobin and malaria is the classic anthropological case (**A. C. Allison**). HLA associations with autoimmune disease, and later SNP associations, including GWAS, extend the same logic. Markers also guide counselling when they track a Mendelian locus in a pedigree.

Association is not always causation. Linkage disequilibrium can flag a neighbour gene. Population structure can produce false links if cases and controls are poorly matched.

Forensics

STRs from several loci identify individuals with high discrimination when the sample is good. Markers also exclude suspects and identify remains after disasters. mtDNA helps when hair shafts or degraded bone yield little nuclear DNA, because many copies exist per cell. Y markers can track a paternal line in sexual-assault mixtures. Forensic anthropology still needs bones, context and consent. DNA does not replace osteology.

DNA testing of unidentified dead, or in disputed paternity, is a public application of the same methods.

Flow diagram

Genetic markers -> Population variation

Genetic markers -> Disease association

Genetic markers -> Forensics

Population variation -> Frequencies not races

Disease association -> Sickle cell / GWAS

Forensics -> STRs and mtDNA

Conclusion

Genetic markers are heritable polymorphisms used as instruments. In populations they map variation and history. In medicine they flag disease risk and Mendelian loci. In forensics they identify and exclude. Landsteiner, Jeffreys, Cavalli-Sforza and Lewontin mark the path from blood groups to DNA, and race typology does not follow from markers.

Facts & figures

ABO blood groups

Classical serological markers described by Landsteiner. They remain a teaching example of Mendelian polymorphism used in population surveys.

Alec Jeffreys

He developed DNA fingerprinting from hypervariable regions, which became the basis of forensic STR profiling.

Richard Lewontin

He showed that most human genetic variation is found within populations rather than between continental groups.

Sickle-cell and malaria

A. C. Allison's work linked the sickle-cell allele to malaria, a classic marker-disease-environment case in biological anthropology.

STRs

Short tandem repeats used in forensic identification because they are highly variable among individuals.