Q8(b)

'Genome-wide Disease Association Studies (GWAS) advanced our understanding of health and disease.' Discuss

UPSC Civil Services Mains 2025 · Anthropology GS 1 · 15 marks · Genetic imprints in human disease, genetic screening, genetic counseling, human DNA profiling, gene mapping and genome study.

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Core demand of the question

- Define GWAS in plain language
- Show how it advanced understanding of health and disease
- **Give anthropological limits:** population bias, small effects, ethics
- Link the method to genetic markers

Revision summary

GWAS tests common SNPs across the genome for association with a trait or disease.

It showed that many adult diseases are polygenic and pointed to biological pathways.

Polygenic scores are research tools, not fate.

Limits include European-ancestry bias, missing heritability, and misuse as race biology.

Environment, foetal programming and inequality still explain a large share of health.

GWAS belongs with genetic markers and with non-Mendelian inheritance.

Introduction

A genome-wide association study, or GWAS, scans many common DNA markers across the genome to see which ones are statistically more frequent in people with a disease or trait than in people without it. It advanced medicine by finding many small genetic clues at once. It did not find one gene for 'being ill'.

Body

What GWAS does

After the Human Genome Project and HapMap-style catalogues of SNPs, researchers could test hundreds of thousands of markers in large samples. Hits appear as peaks on a Manhattan plot. **Wellcome Trust** case-control studies and later consortia on height, schizophrenia, type 2 diabetes and autoimmune disease are the textbook wave. The method is an application of genetic markers at genomic scale.

GWAS is good at common variants of small effect. It is weaker at rare variants of large effect, which need sequencing families or other designs.

How understanding advanced

- **Architecture of complex disease:** Heart disease, diabetes and many psychiatric traits are polygenic, which matches non-Mendelian inheritance. Barker's foetal environment still matters; GWAS adds a layer of inherited liability, not a replacement.
- **Biology clues:** Hits can point to pathways (lipid metabolism, immune signalling) even when each SNP does almost nothing alone.
- **Pharmacogenomics and risk scores:** Polygenic scores try to rank risk. They are research tools. They are not destiny, especially when diet, infection and inequality are strong.
- **Infectious disease:** Host-genetics GWAS, including work during COVID-19, asked why severity differs. That is biological anthropology in a hospital key: variation, not blame.

Genome India and other non-European projects exist because the first decade of GWAS over-sampled European ancestry.

Limits an anthropologist must name

- **European bias:** Predictions fail or weaken in other populations. This is a scientific and justice problem.
- **Missing heritability:** Even many hits leave much family resemblance unexplained. Environment, rare variants and gene-gene interaction remain.
- **Race misuse:** A GWAS hit is not a racial essence. Lewontin's lesson still applies.
- **Ethics:** Consent, feedback of risk, and insurance fear. Tribal and small-community sampling repeats older extractive genetics unless governance is real.

GWAS advanced understanding by mapping the polygenic landscape of common disease. It did not close the biocultural file.

Flow diagram

GWAS -> Many SNPs

Many SNPs -> Complex disease map

Complex disease map -> Small effects

Complex disease map -> European sample bias

Complex disease map -> Environment still matters

Conclusion

GWAS showed that many common illnesses are influenced by many small DNA differences and pointed to biological pathways. The anthropological discussion is population bias, small effects, and the continuing force of environment and inequality. It is a powerful marker method, not a new racial science.

Facts & figures

GWAS

A study design that tests many common genomic markers for statistical association with a disease or quantitative trait.

SNP

A single-nucleotide polymorphism: a one-base difference used as the usual GWAS marker.

Polygenic score

A sum of many GWAS effects used to rank genetic liability. It is sensitive to the ancestry of the training sample.

Missing heritability

The gap between how much variation GWAS currently explains and how much family studies suggest is inherited.