Q7(b)

What is a multifactorial trait? Illustrate your answer with suitable human examples.

UPSC Civil Services Mains 2024 · Anthropology GS 1 · 15 marks · Mendelian genetics in man-family study, single factor, multifactor, lethal, sub-lethal and polygenic inheritance in man.

9.2 Mendelian genetics in man-family study, single factor, multifactor, lethal, sub-lethal and polygenic inheritance in man.

Core demand of the question

- Define a multifactorial trait and its genetic architecture
- Explain threshold and continuous models
- Illustrate with suitable normal and disease traits in humans
- State methods, gene-environment interaction and anthropological limits

Revision summary

A multifactorial trait combines many genetic variants with environment and development.

Fisher explained continuous variation through many small Mendelian effects.

Height, pigmentation, BMI and blood pressure are continuous examples.

Cleft lip, neural-tube defects, diabetes and hypertension can use a liability-threshold model.

Twin, family and GWAS designs estimate components but depend on population and environment.

Yajnik's thin-fat phenotype illustrates developmental and nutritional interaction in India.

Introduction

A multifactorial trait results from many genetic variants acting with environmental and developmental factors. It usually lacks a simple Mendelian pedigree: the phenotype may be continuously distributed, like stature, or cross a liability threshold, like cleft lip.

Body

Architecture and examples

R. A. Fisher's quantitative genetics showed how many Mendelian loci of small additive effect can generate a continuous curve. Phenotypic variance includes genetic, environmental and interaction terms. Height, body mass, skin pigmentation and blood pressure are continuous multifactorial traits. Height reflects polygenic potential plus childhood nutrition and infection; pigmentation combines variants including **SLC24A5** and **MC1R** with UV-related selection and tanning.

For dichotomous conditions, the **liability-threshold model** proposes an unobserved continuous liability. Disease appears when genetic and environmental load crosses a threshold. Cleft lip/palate, neural-tube defects, type 2 diabetes and essential hypertension are

standard examples. Recurrence is higher among close relatives and after a severe or multiple case, but no 3:1 ratio follows.

Twin, adoption and family studies estimate recurrence and heritability; genome-wide association studies identify many loci, usually of small effect. **C. S. Yajnik's** Indian "thin-fat" phenotype research illustrates developmental mismatch: constrained foetal growth combined with later calorie abundance raises metabolic risk. Lactase persistence and dairying offer a longer-term gene-culture interaction, though the phenotype has major-locus effects and should not be confused with a purely polygenic trait.

Multifactorial does not mean vaguely "many causes". Models must specify exposure and population. Heritability changes with environment, polygenic scores transfer poorly across ancestries, and social inequality can be mislabelled genetic risk.

Flow diagram

Many genetic variants -> Liability or quantitative value

Environment and development -> Liability or quantitative value

Gene-environment interaction -> Liability or quantitative value

Liability or quantitative value -> Continuous trait

Liability or quantitative value -> Threshold crossed?

Threshold crossed? -> yes Disease phenotype

Conclusion

- **Multifactorial traits are the normal terrain of human biological variation:** many alleles, development and lived environment jointly make the phenotype. Their study requires quantitative and biocultural models, not Mendelian determinism or a genes-versus-culture choice.

Facts & figures

Liability threshold

A continuous, unobserved susceptibility becomes a categorical disorder only after a threshold is crossed.

R. A. Fisher

Showed mathematically that multiple Mendelian loci can account for apparently continuous biometric traits.

C. S. Yajnik

Indian research linked small size with relatively high adiposity and later metabolic risk, highlighting developmental mismatch.

GWAS caution

Most common-trait hits have small effects and ancestry-specific prediction; association is not individual destiny.