Q1(e)

Hemoglobin in health and disease.

UPSC Civil Services Mains 2024 · Anthropology GS 1 · 10 marks · Epidemiological Anthropology

9.8 Epidemiological Anthropology: Health and disease. Infectious and non-infectious diseases, Nutritional deficiency related diseases.

Core demand of the question

- Explain haemoglobin structure and physiological role
- Describe normal developmental forms and important variants
- Connect haemoglobin disorders to health, ecology and population variation
- Use precise anthropological examples without genetic determinism

Revision summary

HbA is α2β2; HbF is α2γ2 and has higher oxygen affinity.

Haemoglobin carries oxygen and participates in carbon-dioxide and acid-base physiology.

HbS polymerises when deoxygenated; HbSS causes disease while HbAS may protect against severe falciparum malaria.

Thalassaemia is deficient globin-chain production, not an abnormal iron molecule.

HbE, HbC and thalassaemia distributions record selection, drift and migration.

Anaemia also has nutritional, infectious and inflammatory causes, so genotype is not the whole diagnosis.

Introduction

Haemoglobin is the iron-containing tetramer in red blood cells that transports oxygen and contributes to carbon-dioxide and acid-base regulation. Its molecular variants connect physiology, Mendelian inheritance, malaria ecology, migration and public health, making it a classic marker in biological anthropology.

Body

Normal function and variation

Adult HbA is mainly two alpha and two beta globin chains (α2β2), each carrying haem. HbA2 is α2δ2, while fetal HbF (α2γ2) has higher oxygen affinity and supports placental transfer. After birth, globin-chain expression switches from gamma toward beta. Iron deficiency reduces haemoglobin concentration but is not a globin-gene disorder.

Health and disease

- **Sickle-cell disease:** A missense variant in HBB substitutes valine for glutamic acid in beta-globin. Deoxygenated HbS polymerises, distorting cells and causing haemolysis, vaso-occlusion and organ injury. HbSS is disease; HbAS is usually a healthy trait.

- **Thalassaemias:** Reduced alpha- or beta-chain production causes imbalance, anaemia and ineffective erythropoiesis. Severity ranges from silent carrier states to transfusion-dependent beta-thalassaemia major.
- **Other variants:** HbE is frequent in parts of eastern and Southeast Asia and may interact severely with beta-thalassaemia. HbC occurs especially in West Africa. Their geographic distributions reflect mutation, drift, migration and selection.
- **Acquired states:** Anaemia may follow iron, folate or B12 deficiency, malaria, hookworm, inflammation or blood loss. Excess haemoglobin or erythrocytosis at altitude can aid oxygen carriage but also increase blood viscosity.

J. B. S. Haldane proposed that thalassaemia geography related to malaria; **A. C. Allison** supplied direct evidence for HbAS advantage against severe falciparum malaria. This does not mean "malaria causes sickle mutation": malaria changes the fitness of pre-existing variants.

In India, sickle variants occur in several central, western and southern populations, while beta-thalassaemia and HbE have different regional concentrations. Screening must combine electrophoresis or HPLC with counselling and must avoid turning population association into caste or tribal stigma.

Flow diagram

Globin genes -> Haemoglobin tetramer

Haemoglobin tetramer -> Oxygen transport

Globin genes -> HbS HbE HbC variants

Globin genes -> Thalassaemia: reduced chain output

Malaria nutrition altitude -> Population pattern and health

Conclusion

Haemoglobin is simultaneously a respiratory molecule and an evolutionary record. Chain switching, structural variants and production defects explain disease; malaria, migration, nutrition and care explain their unequal distribution and consequences.

Facts & figures

HbA and HbF

Major adult HbA is α2β2. Fetal HbF is α2γ2 and binds oxygen more strongly, facilitating placental oxygen transfer.

HbS mechanism

A beta-globin missense variant promotes polymerisation of deoxygenated HbS, leading to sickling, haemolysis and vaso-occlusion in HbSS disease.

Thalassaemia

A quantitative defect in alpha- or beta-globin production. Clinical severity depends on the affected genes and partner allele.

Malaria selection

Haldane framed the hypothesis; Allison showed a survival advantage of sickle trait in falciparum-malaria environments.

Indian relevance

Sickle variants, beta-thalassaemia and HbE have differing regional and community frequencies; screening requires consent and non-stigmatising counselling.