ANANTAM IAS · OPTIONAL NOTES

Anthropology Optional Human Population Genetics, Race and Human Variation

Paper 1 · Unit 10

The quantitative heart of biological anthropology: the Hardy-Weinberg baseline and the forces that push populations off it, the blood-group and molecular markers that map human variation, the balanced polymorphisms that prove selection acts on us, and the collapse of the biological race concept into clines and social construction.

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UPSC SYLLABUS

Mendelian population: Hardy-Weinberg law; causes and changes which bring down frequency: mutation, isolation, migration, selection, inbreeding and genetic drift. Chromosomes and chromosomal aberrations in man. Race and racism; biological basis of morphological variation; racial criteria; racial classification of human populations. Age, sex and population variation as genetic markers: ABO, Rh blood groups, HLA. Concept of human growth and development. Ecological and epidemiological anthropology.

Mendelian Population and the Hardy-Weinberg Equilibrium

Population genetics begins with a null model: what allele frequencies would look like if nothing were changing them. That model is Hardy-Weinberg, and its entire value is that real populations deviate from it, and the deviation is the signature of evolution. Get the equation and the assumptions exact, because half the marks in this unit sit here.

- A Mendelian population is a group of interbreeding, sexually reproducing individuals who share a common gene pool, the total collection of alleles in the group. The unit of study in population genetics is not the individual but this gene pool and the frequencies of alleles within it. Evolution, at this level, is simply change in allele frequencies over generations.
- The Hardy-Weinberg law (also principle or equilibrium) was stated independently in 1908 by the English mathematician G.H. Hardy and the German physician Wilhelm Weinberg. It holds that in a large, randomly mating population, the allele and genotype frequencies remain constant from generation to generation, provided no evolutionary force is acting. It is the population-genetic equivalent of Newton's first law: things stay put unless a force moves them.
- The algebra, which must be reproduced correctly. For a single locus with two alleles, A at frequency p and a at frequency q, the allele frequencies sum to one: $p + q = 1$. The genotype frequencies at equilibrium are given by the binomial expansion $(p + q)$ squared, that is p squared (AA homozygotes) plus $2pq$ (Aa heterozygotes) plus q squared (aa homozygotes), and these sum to one. Given allele frequencies, you can predict genotype frequencies, and vice versa.
- The law's practical use is estimating hidden carrier frequencies for recessive disorders. If a recessive condition has incidence one in ten thousand, then q squared equals 0.0001, so q equals 0.01, p equals 0.99, and the carrier (heterozygote) frequency $2pq$ is about 0.0198, roughly two per cent. This is how the frequency of unaffected carriers is inferred from the frequency of affected individuals, a standard exam calculation.
- The assumptions are the examinable core, because equilibrium holds only if all of them hold, and each corresponds to an evolutionary force that is absent. The population must be very large (so that random drift is negligible); mating must be random with respect to the locus (panmixia, no assortative mating or inbreeding); there must be no mutation altering allele frequencies; no gene flow or migration in or out; no natural selection (all genotypes equally viable and fertile); generations should not overlap; and the locus is assumed autosomal with equal allele frequencies in the two sexes.
- The logical pay-off, which the examiner wants stated: because equilibrium requires all these conditions, a population that departs from Hardy-Weinberg genotype proportions must be experiencing at least one evolutionary force. The law is valuable not because populations obey it, but precisely because they do not; it is the baseline against which mutation, selection, drift, migration and non-random mating are detected and measured.

KEY TERMS

Mendelian population — A group of interbreeding, sexually reproducing organisms sharing a common gene pool; the unit of study in population genetics.

Hardy-Weinberg equilibrium — The condition in which allele and genotype frequencies stay constant across generations (p squared + 2pq + q squared = 1) because no evolutionary force is acting.

Panmixia — Random mating with respect to the locus in question, one of the assumptions required for Hardy-Weinberg equilibrium.

The Forces of Evolutionary Change

Each Hardy-Weinberg assumption, when violated, names a force that changes allele frequencies. This section is that list turned into mechanisms: mutation, natural selection, genetic drift with its special cases, gene flow, and non-random mating. Attribute the drift theory to Sewall Wright and keep selection's three modes distinct.

- Mutation is the ultimate source of all new genetic variation: a heritable change in the DNA, from point substitutions to chromosomal rearrangements. Mutation rates are low, of the order of one in ten thousand to one in a million per locus per generation, so mutation alone is a weak force that shifts frequencies only slightly. Its importance is qualitative: it supplies the raw material of variation on which the other forces act, and it is random with respect to fitness, that is, mutations are not directed toward adaptive need.
- Natural selection is differential reproduction and survival of genotypes according to their fitness, where Darwinian fitness means reproductive success, not strength or longevity for their own sake. Selection acts on the phenotype and thereby on the underlying alleles. It comes in three modes: directional selection favours one extreme and shifts the mean (the classic case is industrial melanism in the peppered moth, Biston betularia); stabilising selection favours the intermediate and eliminates extremes (human birth weight, where both very low and very high weights carry higher mortality); and disruptive or diversifying selection favours both extremes over the mean.
- Balancing selection maintains variation rather than reducing it, most importantly through heterozygote advantage or overdominance, where the heterozygote is fitter than either homozygote. This preserves both alleles in the population indefinitely, producing a balanced polymorphism; the sickle-cell and malaria relationship is its textbook demonstration and is treated in its own section below.
- Genetic drift is random change in allele frequency from sampling error between generations, and it is significant in small populations. By chance alone, alleles may rise, fall, become fixed or be lost, with no reference to adaptive value. Drift reduces genetic variation within a population and increases divergence between isolated populations. Its principal theorist is Sewall Wright, and it is the non-adaptive, chance counterpart to selection.
- Two special cases of drift are separately examinable. The founder effect occurs when a new population is established by a small number of colonisers who carry only a fraction, and an unrepresentative fraction, of the parent population's variation, so allele frequencies in the new group can differ sharply; the high incidence of Ellis-van Creveld syndrome among the Pennsylvania Amish, and of specific conditions among the Pingelap islanders and Ashkenazi Jews, illustrate it. The bottleneck effect occurs when a population is drastically reduced by a catastrophe and the survivors are a non-representative sample, so diversity is sharply and lastingly lowered even if numbers later recover.
- Gene flow, also called migration or admixture, is the movement of alleles between populations through interbreeding. It has two effects: it introduces new variation into the receiving population, and it makes populations more similar to one another, counteracting the divergence produced by drift and local selection.

Much human geographical variation is clinal partly because gene flow between neighbouring populations smooths differences into gradients.

- Non-random mating changes genotype frequencies without changing allele frequencies, and inbreeding is its most important form. Inbreeding, mating between relatives, increases homozygosity and so exposes deleterious recessive alleles, producing inbreeding depression; consanguineous marriage, common in some Indian communities, raises the genetic load of recessive disorders for exactly this reason. Assortative mating (choosing partners similar or dissimilar in some trait) similarly alters the distribution of genotypes. Isolation, geographical or social, reinforces drift and inbreeding by preventing gene flow.

KEY TERMS

Genetic drift — Random change in allele frequency due to sampling error, important in small populations; can fix or lose alleles by chance. Chief theorist Sewall Wright.

Founder effect — A form of drift in which a new population founded by few individuals carries an unrepresentative subset of the parent gene pool, shifting allele frequencies.

Gene flow — Transfer of alleles between populations through interbreeding; introduces new variation and reduces differences between populations.

Balanced polymorphism — The stable maintenance of two or more alleles by balancing selection, typically heterozygote advantage, as in sickle cell and malaria.

Genetic Markers and Blood Group Systems

A genetic marker is a simply-inherited, environment-proof, polymorphic trait whose frequency can be measured and compared across populations, which is exactly what population genetics needs. Blood groups were the first such markers, and they let anthropology map human variation before DNA. Attribute each system to its discoverer.

- A genetic marker is a detectable, polymorphic trait with a known and simple Mendelian mode of inheritance and little or no environmental modification, which makes it a reliable index of allele frequencies. Because their frequencies vary geographically, markers let anthropologists estimate genetic distance between populations, trace gene flow and migration history, and reconstruct population structure. Blood groups are the classic markers precisely because they meet these conditions.
- The ABO system was discovered by Karl Landsteiner in 1900 to 1901, work for which he received the Nobel Prize in 1930. Three alleles at a locus on chromosome nine, I-A, I-B and i, produce four phenotypes: A, B, AB and O; I-A and I-B are codominant and i is recessive. The antigens sit on the red cell surface and the corresponding antibodies in the serum. ABO allele frequencies vary strikingly across the world (B is relatively frequent in Asia, O reaches very high frequencies in many Native American populations, A is common in Europe), which makes ABO a workhorse of population comparison. Disease associations exist (group O with duodenal ulcer and with more severe cholera, group A with gastric cancer), of interest because they hint at past selection.
- The Rh system was discovered by Landsteiner and Alexander Wiener in 1940, named after the rhesus monkey used in the experiments. Its most important antigen is D: individuals are Rh positive if they carry it and Rh negative if they do not (genotype dd). The clinical significance is haemolytic disease of the newborn (erythroblastosis foetalis), which can arise when an Rh negative mother carries an Rh positive foetus and, sensitised in a first such pregnancy, produces antibodies that attack the red cells of a later Rh positive foetus; anti-D immunoglobulin prophylaxis now prevents it. Rh negative frequency is high among the Basques (a classic anthropological anomaly) and low in most Asian and African populations.

- The MN system was discovered by Landsteiner and Philip Levine in 1927. The M and N antigens are governed by codominant alleles L-M and L-N, giving three phenotypes, M, MN and N. Because the antibodies are weak and the system is clinically almost silent, it is under little selection, which makes it an excellent neutral marker: its frequencies reflect drift and population history rather than adaptation, and it is therefore especially useful for reconstructing relationships between populations.
- The HLA system, the human leukocyte antigens encoded by the major histocompatibility complex on chromosome six, is the most polymorphic genetic system known in humans. It governs immune self-recognition and is central to tissue transplantation matching; it shows strong disease associations (HLA-B27 with ankylosing spondylitis, various HLA-DR alleles with type 1 diabetes and rheumatoid arthritis). For population genetics its extreme polymorphism, maintained by balancing selection driven by pathogen pressure, makes it a highly informative marker of population structure and history, and it is also used in forensics and parentage testing.
- Secretor status is a further Mendelian marker, governed by the FUT2 gene on chromosome nineteen. Secretors (carrying the dominant Se allele) secrete their ABO and H blood-group antigens in saliva, gastric juice and other body fluids, while non-secretors (homozygous se) do not; roughly four in five people are secretors. Beyond its use as a simple marker, secretor status matters medically because non-secretors show altered susceptibility to certain infections (for instance resistance to some noroviruses, and different urinary-tract infection risk).
- Other markers worth naming to show range: haptoglobin and transferrin serum-protein variants, the G6PD variants, haemoglobin variants (HbS, HbE, thalassaemias), and the simple Mendelian trait of PTC (phenylthiocarbamide) tasting, where the ability to taste the bitter compound is dominant and its frequency varies between populations. The general point: markers let biological anthropology quantify variation objectively, and they collectively showed, long before genomics, that human variation is continuous and does not sort into races.

KEY TERMS

Genetic marker — A polymorphic, simply-inherited, environment-proof trait used to measure allele frequencies and compare populations; blood groups are the classic examples.

ABO system — Landsteiner's blood-group system on chromosome 9, alleles I-A, I-B (codominant) and i (recessive), giving phenotypes A, B, AB, O; a key population marker.

HLA — The highly polymorphic human leukocyte antigen system on chromosome 6, governing immune recognition and transplant matching; a very informative population marker.

Secretor status — FUT2-governed trait (dominant Se) determining whether ABO/H antigens are secreted in body fluids; a Mendelian marker with infection-susceptibility effects.

Genetic and Chromosomal Disorders; the Sickle Cell-Malaria Balanced Polymorphism

This section carries the unit's most examinable single idea, the sickle-cell and malaria balanced polymorphism, which is the clearest proof that natural selection operates on living human populations. Set it out with its full chain of attribution, then cover the single-gene and chromosomal disorders as ordered lists.

- Sickle cell anaemia is caused by a single point mutation in the beta-globin gene on chromosome eleven, substituting valine for glutamic acid at the sixth position of the beta chain, which yields abnormal haemoglobin S. Linus Pauling and colleagues in 1949 identified it as the first 'molecular disease', and Vernon

Ingram shortly afterwards pinpointed the single amino-acid substitution responsible. Homozygotes (HbSS) suffer full sickle cell anaemia, in which red cells distort under low oxygen, block vessels and are destroyed, a severe and historically often fatal condition.

- The balanced polymorphism is the crucial point. Heterozygotes (HbAS, sickle cell trait) are largely healthy and are relatively protected against Plasmodium falciparum malaria. In malarial regions the two homozygotes are both at a disadvantage (HbSS from anaemia, HbAA from malaria) while the heterozygote is fittest, so heterozygote advantage maintains the otherwise-lethal HbS allele at high frequency. J.B.S. Haldane advanced the 'malaria hypothesis' for haemoglobin disorders around 1949, and A.C. Allison demonstrated the sickle-malaria association empirically in the 1950s. The geographical overlap of the sickle allele with historic falciparum malaria, across sub-Saharan Africa, the Mediterranean, and parts of India including several tribal populations, is the visible evidence.
- Why examiners return to this: it is the definitive worked example of natural selection, balancing selection and gene-culture interaction in humans, all at once, and it can be linked to Livingstone's argument that the spread of agriculture and forest clearance created the mosquito breeding grounds that raised malaria pressure and thereby selected for the sickle allele. Related malaria-protective polymorphisms (G6PD deficiency, the thalassaemias, and Duffy-antigen negativity against Plasmodium vivax) reinforce the pattern.
- Phenylketonuria (PKU) is an autosomal recessive inborn error of metabolism: deficiency of the enzyme phenylalanine hydroxylase (PAH gene, chromosome twelve) causes phenylalanine to accumulate and, if untreated, produces severe intellectual disability. Its exam value is that it is the model case of genotype-environment interaction: the same genotype yields a healthy child on a phenylalanine-restricted diet and a damaged one on a normal diet, so the disorder is preventable by environmental control, detected through newborn screening.
- Chromosomal disorders divide into autosomal and sex-chromosomal aneuploidies. Down syndrome is trisomy 21, the commonest autosomal aneuploidy, usually caused by meiotic nondisjunction with risk rising sharply with maternal age; features include intellectual disability, characteristic facial morphology, hypotonia, a single palmar crease and frequent congenital heart defects. It was clinically described by John Langdon Down in 1866, and Jerome Lejeune identified the extra chromosome 21 in 1959. Trisomy 18 (Edwards) and trisomy 13 (Patau), and the 5p deletion of cri-du-chat, are the other named autosomal anomalies.
- The sex-chromosomal aneuploidies: Klinefelter syndrome is 47,XXY in males, giving tall stature, small testes, infertility, gynaecomastia and reduced testosterone, with a Barr body present because one X is inactivated. Turner syndrome is 45,X (monosomy X) in females, giving short stature, a webbed neck, gonadal dysgenesis with streak ovaries and infertility, and no Barr body. Note the diagnostic contrast that examiners like: a normal male has no Barr body, a Klinefelter male has one, a normal female has one, and a Turner female has none.
- The broader concept to frame all of this is the inborn error of metabolism, introduced by Archibald Garrod (alkaptonuria, early twentieth century), the recognition that single-gene defects can disrupt specific metabolic steps, which underlies the one-gene-one-enzyme understanding of PKU and many other recessive conditions and connects clinical genetics back to Mendelian population genetics.

KEY TERMS

Balanced polymorphism (sickle cell) — The maintenance of the HbS allele at high frequency because HbAS heterozygotes resist falciparum malaria, outweighing the loss of HbSS homozygotes; the classic proof of human natural selection.

Genotype-environment interaction (PKU) — Phenylketonuria as the model case: the same recessive genotype causes disease on a normal diet but not on a phenylalanine-restricted one, so the outcome depends on environment.

Aneuploidy — An abnormal chromosome number from nondisjunction; autosomal (Down, trisomy 21) or sex-chromosomal (Klinefelter 47,XXY; Turner 45,X).

The Concept of Race: Criteria and Historical Classifications

The race question is asked more than any other in this unit, and it must be answered in two movements: first the traditional biological concept with its criteria and historical schemes, then the modern demolition. This section is the first movement. Present the classifications as a chronology and keep the Indian schemes ready.

- In its traditional biological sense, a race is a subdivision of the human species whose members share a distinctive combination of heritable physical traits associated with a geographical region. The concept assumes that humanity sorts into a small number of relatively discrete, biologically bounded types. This is the concept the second movement will dismantle; state it clearly first, because you cannot critique what you have not set out.
- The racial criteria are the traits classifiers used, and they divide into metric (measured) and non-metric (observed). The main ones: skin colour, hair form and colour, eye colour and form (including the epicanthic fold), stature and body build, and two indices in particular, the cephalic index (the ratio of head breadth to length, coined by Anders Retzius, dividing people into dolichocephalic, mesocephalic and brachycephalic) and the nasal index (nasal breadth to height, dividing them into leptorrhine, mesorrhine and platyrrhine). Later, blood-group frequencies were added as criteria.
- The historical classifications, as a chronology. Francois Bernier (1684) is usually credited with the first classification of humans into races. Carl Linnaeus (Systema Naturae, 1758) gave four geographical varieties, Americanus, Europaeus, Asiaticus and Afer, laden with temperament stereotypes. Johann Friedrich Blumenbach (1795) produced the influential five-race craniometric scheme, Caucasian (a term he coined), Mongolian, Ethiopian, American and Malayan; a monogenist, he held all to descend from one stock by degeneration, but he ranked the 'Caucasian' as the original type.
- The twentieth-century typologists carried the scheme to its most elaborate and most criticised forms. The common three-fold division into Caucasoid, Mongoloid and Negroid became the shorthand. Earnest Hooton built a detailed hierarchy of primary races and subtypes on morphological traits. Carleton Coon, in The Origin of Races (1962), proposed five subspecies (Caucasoid, Mongoloid, Australoid, Congoid, Capoid) and the notorious thesis that these lineages crossed the threshold to Homo sapiens at different times, a polygenic claim widely condemned as scientifically unsound and as ammunition for racism.
- The move toward a populational view began within this tradition. Stanley Garn (Human Races, 1961) replaced fixed types with geographical races, local races and microraces defined as breeding populations, a shift that already pointed away from typology toward population genetics and prepared the ground for abandoning the type concept altogether.
- The Indian racial classifications are examinable in their own right. Herbert Risley (The People of India, 1915) used the nasal index to define seven types and tied them to caste, a scheme now discredited for confusing a social institution with biology. The standard textbook classification is B.S. Guha's (Census of India, 1935),

which identified six main racial elements in the Indian population: Negrito, Proto-Australoid, Mongoloid, Mediterranean, Western Brachycephals and Nordic. D.N. Majumdar and J.H. Hutton worked in the same anthropometric tradition. Present Guha as the reference scheme while noting that it too rests on the typological assumptions the modern position rejects.

KEY TERMS

Racial criteria — The heritable traits used in racial classification: skin, hair and eye form and colour, stature, and the cephalic and nasal indices; metric (measured) and non-metric (observed).

Cephalic index — Retzius' ratio of maximum head breadth to length, classifying skulls as dolichocephalic (long), mesocephalic (medium) or brachycephalic (broad).

Guha's classification (1935) — The standard Indian racial scheme: six elements, Negrito, Proto-Australoid, Mongoloid, Mediterranean, Western Brachycephals and Nordic.

The Modern Position: Race Has No Sound Biological Basis

This is the second movement and the high-value conclusion of the whole race question. The mainstream anthropological position is that traditional races are not valid biological units, and it rests on specific arguments and named authors. Deploy them in order: clines, non-concordance, Lewontin's apportionment, and the social-construction thesis.

- The core claim: human biological variation is real, patterned and often adaptive, but it is not organised into discrete races, and the traditional race concept has no sound biological basis. Note the balance the examiner rewards: this rejects races as biological types, not the reality of human variation, and it holds that 'race' survives as a social construct with real and harmful consequences even though it fails as biology.
- The clinal argument is Frank Livingstone's, in the essay 'On the Non-Existence of Human Races' (1962), summarised in his line 'There are no races, there are only clines.' A cline is a gradient along which a trait varies gradually and continuously across geography. Human traits vary clinally, without the sharp boundaries that discrete races would require; where you draw a racial line is arbitrary, since the variation is a gradient, not a set of steps.
- The non-concordance argument follows: different traits have different geographical distributions and do not co-vary. A classification by skin colour cuts the species differently from one by blood-group frequency, which cuts it differently again from one by head form. Because the criteria disagree, no consistent set of races emerges; the classification you get depends entirely on which trait you privilege, which shows the races are artefacts of the classifier's choice, not natural divisions.
- The genetic apportionment argument is Richard Lewontin's, 'The Apportionment of Human Diversity' (1972). Analysing variation at blood-group and protein loci, he found that about eighty-five per cent of human genetic variation lies within local populations, about eight per cent among populations within a so-called race, and only around six to seven per cent among the major races. The conclusion: racial classification accounts for a tiny fraction of human genetic diversity and has almost no taxonomic value; most human variation is between individuals, not between races. (A.W.F. Edwards later objected, in 'Lewontin's Fallacy' (2003), that the correlation structure across many loci does permit statistical assignment, but the anthropological mainstream continues to hold that traditional discrete races are not biologically valid.)
- The social-construction thesis is Ashley Montagu's, Man's Most Dangerous Myth: The Fallacy of Race (1942), which argued that race is a social myth rather than a biological reality and proposed replacing 'race'

with 'ethnic group'. Montagu was central to drafting the UNESCO Statement on Race (1950, revised 1951), which declared race a social myth and rejected any biological ranking of populations; the American Anthropological Association's Statement on Race (1998) restated the position that race is a recently invented social and cultural construct, not a biological given, while insisting that racism has real effects.

- Two supporting points seal the argument. First, modern humans are an evolutionarily young and genetically homogeneous species (the recent Out of Africa expansion), with the greatest genetic diversity found within Africa, so there has been neither time nor isolation enough for deep racial divergence. Second, the visible traits used to define races are adaptive responses to local environments and cross racial lines: skin pigmentation tracks ultraviolet radiation through the vitamin D and folate balance (Nina Jablonski), following Gloger's rule; body form follows Bergmann's rule (larger bodies conserve heat in the cold) and Allen's rule (shorter extremities in the cold); nose form tends to be narrower in cold, dry air. These clinal adaptations explain the variation far better than racial types do.
- Hold the distinction between race and racism as the closing point. The biological race concept is rejected; racism, the ideology that ranks human groups and asserts some are innately superior, is a social and political reality that produces measurable harm regardless of biology's verdict. Anthropology's mature position is that studying human biological variation and rejecting the race concept are the same task, not opposed ones.

KEY TERMS

Cline — A gradient of continuous, gradual change in a trait across geography; Livingstone's argument that human variation is clinal, not divided into discrete races.

Non-concordance of traits — The fact that racial criteria (skin, blood group, head form) have different distributions and do not co-vary, so classifications depend arbitrarily on which trait is chosen.

Lewontin's apportionment — The 1972 finding that about 85 per cent of human genetic variation is within local populations and only about 6 to 7 per cent among major races, undercutting racial taxonomy.

Human Growth and Development

Growth is a smaller but recurring topic, so control the core vocabulary and the distinctive features of the human pattern rather than trying to cover everything. Separate growth from development from maturation, and know the stages, the curves and the factors.

- Distinguish the three terms precisely. Growth is a quantitative increase in size or mass; development is a qualitative process of differentiation and increasing functional complexity; maturation is progress toward the mature adult state. A child grows taller (growth) and also acquires new capacities and proportions (development); the two proceed together but are not the same.
- The stages of the human life cycle: the prenatal period (germinal, embryonic and foetal phases), infancy, childhood, the juvenile phase, adolescence (marked by puberty and the adolescent growth spurt), adulthood, and senescence. The human pattern has distinctive features: a markedly prolonged childhood and delayed maturity relative to other primates, and a pronounced adolescent growth spurt that is essentially unique to humans, both linked to the demands of a large brain and extensive cultural learning.
- Growth is described by two curve types: the distance curve, which plots size attained against age (a rising sigmoid), and the velocity curve, which plots the rate of growth against age and reveals the rapid infant growth, the steady childhood phase and the adolescent peak. Richard Scammon's classic scheme showed that different tissue systems follow different growth trajectories: general (body, sigmoid), neural (brain,

growing fast and early and nearly complete in childhood), genital (reproductive, growing late at puberty), and lymphoid (peaking in later childhood then regressing).

- The factors affecting growth are genetic, hormonal (growth hormone, thyroid hormones and, at puberty, the sex hormones), nutritional, and environmental or socio-economic (disease load, altitude, sanitation, family circumstances). Nutrition and health status are the most powerful non-genetic influences, and growth measures are therefore used as sensitive indicators of a population's welfare.
- The secular trend is an examinable observation: over the past century and more, populations with improving nutrition and health have shown increasing adult stature and earlier sexual maturation across generations, direct evidence that growth is environmentally plastic and not fixed by genes alone.
- Methods of studying growth: longitudinal designs (following the same individuals over time, best for individual trajectories), cross-sectional designs (measuring different individuals of different ages at one time, faster but blind to individual variation), and mixed-longitudinal designs. Growth is assessed by anthropometry (stature, weight, circumferences, skinfolds), by skeletal or bone age (from the ossification and fusion of bones on radiographs) and by dental age. Related to physique are Sheldon's somatotypes, the endomorph, mesomorph and ectomorph components, and bioevents such as menarche and menopause serve as markers of developmental and reproductive timing.

KEY TERMS

Growth vs development — Growth is quantitative increase in size or mass; development is qualitative differentiation and increasing functional complexity; the two proceed together.

Adolescent growth spurt — The rapid acceleration of growth at puberty, essentially unique to humans and linked to the prolonged human childhood and large brain.

Secular trend — The generational increase in adult stature and earlier maturation with improving nutrition and health, showing growth is environmentally plastic.

Human Ecology and Epidemiological Anthropology

The unit closes with two applied fields that connect biology to environment and to culture. Ecological anthropology studies human adaptation to environments; epidemiological or medical anthropology studies disease in its biocultural context. Name the theorists and the classic gene-culture cases, because those cases are what elevate an answer.

- Human ecology, or ecological anthropology, studies the relationship between human populations and their environments, biological and cultural. Its founding theorist is Julian Steward, whose cultural ecology analysed how environment shapes the 'cultural core' of subsistence and social organisation and grounded his theory of multilinear evolution. Roy Rappaport's Pigs for the Ancestors (1968), on the Tsembaga Maring of New Guinea, showed ritual regulating the ecological balance between people, pigs and land, exemplifying the ecosystem approach; Marvin Harris's cultural materialism interpreted practices such as the Indian sacred cow and food taboos as ecological adaptations.
- Human adaptation to environmental stress is the biological side of the field, and adaptation operates on several levels: genetic adaptation (heritable change over generations), developmental acclimatisation (irreversible adjustment during growth), physiological acclimatisation (reversible short-term adjustment) and cultural or behavioural adaptation (clothing, shelter, technology). The stresses studied include heat, cold, solar radiation and, notably, high-altitude hypoxia, where Andean, Tibetan and Ethiopian highland populations have arrived at different adaptive solutions (higher haemoglobin and larger chest capacity in

one case, more efficient oxygen delivery in another), a striking demonstration of multiple genetic routes to the same problem.

- Epidemiological anthropology, closely tied to medical anthropology, studies the distribution and determinants of health and disease in populations with explicit attention to culture, behaviour and ecology, rather than treating disease as a purely biological event. It asks how cultural practices, social organisation and environment shape who falls ill and why.
- The classic cases are gene-culture and behaviour-disease links. Kuru, a fatal prion disease among the Fore of New Guinea, spread through mortuary cannibalism, was traced by D. Carleton Gajdusek (Nobel Prize) with the cultural transmission route documented by Shirley Lindenbaum, a textbook example of a cultural practice driving an epidemic. Lactase persistence, the retention of the ability to digest milk sugar into adulthood, evolved to high frequency specifically in dairying populations, the paradigm case of gene-culture coevolution. And Frank Livingstone's account of sickle cell links agriculture, forest clearance, mosquito breeding, malaria and the selection of the sickle allele into a single biocultural chain.
- A conceptual distinction from medical anthropology belongs here: Arthur Kleinman's separation of disease (the biomedical pathological process, the etic category) from illness (the patient's and community's culturally shaped experience of being unwell, the emic category), together with his notion of explanatory models. Ethnomedicine, the study of indigenous medical systems and healing, and nutritional anthropology, are further branches, and the concept of an epidemiological transition (the shift from infectious to chronic and lifestyle diseases as societies develop) frames population-level change.
- The applied relevance, which is worth a closing line: this biocultural, ecological understanding of disease is directly useful in designing public-health interventions that fit local practice, and in India specifically in addressing tribal and rural health, where sickle cell, G6PD deficiency, nutritional deficiency and the interaction of poverty, environment and culture with disease are precisely the problems these fields were built to analyse.

KEY TERMS

Cultural ecology — Julian Steward's approach to how environment shapes the cultural core of subsistence and social organisation, the basis of his multilinear evolution.

Gene-culture coevolution — The reciprocal shaping of genes and culture, as in lactase persistence evolving in dairying populations, or agriculture raising malaria pressure and selecting the sickle allele.

Disease vs illness (Kleinman) — Disease is the biomedical pathological process (etic); illness is the culturally shaped experience of being unwell (emic); a core distinction in medical anthropology.

Last-mile revision

- Mendelian population = interbreeding group sharing a gene pool. Hardy-Weinberg (Hardy and Weinberg, 1908): p + q = 1 and p squared + 2pq + q squared = 1; allele and genotype frequencies stay constant absent evolutionary force. It is the null model; deviation signals evolution.
- Use HWE to find carriers: if recessive incidence is q squared, carrier frequency is 2pq. Incidence 1/10000 gives q = 0.01 and carriers about 2 per cent.
- HWE assumptions (each a force when violated): large population (no drift), random mating (no inbreeding/assortative), no mutation, no gene flow, no selection, non-overlapping generations, autosomal locus with equal allele frequencies in sexes.
- Forces of change: mutation (ultimate source of variation, weak alone, random to fitness); natural selection (directional, stabilising, disruptive; balancing/heterozygote advantage maintains polymorphism); genetic drift (random, small populations, Sewall Wright); founder and bottleneck effects; gene flow (introduces variation, homogenises populations); non-random mating and inbreeding (change genotype not allele frequencies, expose recessives).
- Genetic marker = polymorphic, simply inherited, environment-proof trait. ABO (Landsteiner 1900, chromosome 9, I-A/I-B codominant, i recessive); Rh (Landsteiner and Wiener 1940, D antigen, haemolytic disease of newborn); MN (Landsteiner and Levine 1927, neutral, best for population history); HLA (chromosome 6, most polymorphic, transplant matching, disease associations); secretor (FUT2, chromosome 19).
- Sickle cell: point mutation in beta-globin (chromosome 11), glutamic acid to valine at position 6; Pauling 1949 'molecular disease', Ingram found the substitution. HbSS = disease, HbAS = malaria resistance. Heterozygote advantage in malarial zones = balanced polymorphism. Haldane's malaria hypothesis (1949), Allison's demonstration (1950s). The proof selection acts on humans.
- PKU: recessive, phenylalanine hydroxylase deficiency (chromosome 12); model of genotype-environment interaction (preventable by diet); newborn screening; inborn error of metabolism (Garrod).
- Chromosomal: Down (trisomy 21, nondisjunction, maternal age; Langdon Down 1866, Lejeune 1959); Klinefelter (47,XXY male, tall, infertile, one Barr body); Turner (45,X female, short, webbed neck, no Barr body). Barr-body contrast is examinable.
- Race (traditional biology): geographical subgroup sharing heritable physical traits. Criteria: skin, hair, eye, stature, cephalic index (Retzius), nasal index; metric and non-metric.
- Historical classifications: Bernier (1684, first), Linnaeus (1758, four varieties), Blumenbach (1795, five races, coined 'Caucasian'), Caucasoid/Mongoloid/Negroid shorthand, Hooton (typology), Coon (1962, five races, discredited polygenic thesis), Garn (populational: geographical/local/microraces). India: Risley (nasal index, caste, discredited), Guha (1935, six elements, the reference scheme).
- Modern position: races are not valid biology, but variation is real and 'race' is a real social construct. Livingstone (1962): no races, only clines. Non-concordance: criteria disagree, so classification is arbitrary. Lewontin (1972): about 85 per cent of variation within populations, only 6 to 7 per cent among races.
- Montagu (1942): race is a social myth; drove the UNESCO Statement on Race (1950). AAA 1998 statement: race is a social construct, racism is real. Humans are young and homogeneous (Out of Africa, most diversity within Africa). Visible traits are clinal adaptations (skin/UV, Bergmann, Allen, nose form). Distinguish race (rejected) from racism (real).

- Growth (quantitative size) vs development (qualitative complexity) vs maturation. Human pattern: prolonged childhood, adolescent growth spurt (near-unique). Distance and velocity curves; Scammon's four tissue curves (general, neural, genital, lymphoid). Secular trend shows environmental plasticity.
- Human ecology: Steward (cultural ecology, cultural core, multilinear evolution), Rappaport (Tsembaga, ecosystem), Harris (cultural materialism). Adaptation levels: genetic, developmental, physiological (acclimatisation), cultural. High-altitude hypoxia: Andean vs Tibetan vs Ethiopian solutions differ.
- Epidemiological/medical anthropology: kuru and mortuary cannibalism (Gajdusek, Lindenbaum); lactase persistence and dairying (gene-culture coevolution); sickle cell and agriculture (Livingstone). Kleinman: disease (etic) vs illness (emic). Applied to tribal and rural health in India.

Common errors to avoid

- Stating the Hardy-Weinberg equation without its assumptions, or vice versa. The two are one answer: equilibrium holds only if population is large, mating random, and mutation, migration and selection absent; deviation from p squared + 2pq + q squared proportions is the evidence of an evolutionary force.
- Confusing genetic drift with natural selection. Drift is random change from sampling error in small populations, non-adaptive; selection is directional, fitness-based, adaptive. Founder and bottleneck effects are special cases of drift, not of selection.
- Saying inbreeding changes allele frequencies. It changes genotype frequencies (raising homozygosity and exposing recessives) but not allele frequencies; only selection, drift, mutation and gene flow change allele frequencies.
- Muddling the blood-group discoverers and dates. ABO is Landsteiner (1900 to 1901); Rh is Landsteiner and Wiener (1940); MN is Landsteiner and Levine (1927). MN is the neutral marker best for reconstructing population history precisely because it is under little selection.
- Describing sickle cell only as a disease and missing the balanced polymorphism. The examinable point is heterozygote advantage against falciparum malaria maintaining the HbS allele; without the malaria link the answer loses its whole significance as proof of human selection.
- Getting the Barr-body pattern wrong. A normal male has none, Klinefelter (XXY) has one, a normal female has one, Turner (XO) has none. The rule is the number of Barr bodies equals the number of X chromosomes minus one.
- Presenting historical racial classifications as settled science. Set them out as a discredited typological tradition, and note that even Guha's Indian scheme rests on the type assumptions the modern position rejects. Risley's caste-race equation is the standard example of the error.
- Answering 'is race biological?' with a flat yes or the slogan 'race is a social construct' alone. The full answer holds all three: traditional biological races are invalid (clines, non-concordance, Lewontin), human variation is nonetheless real and adaptive, and race persists as a consequential social construct while racism is real.
- Citing Lewontin without the numbers or the caveat. About 85 per cent of variation is within populations and only 6 to 7 per cent among races; and Edwards' 'Lewontin's fallacy' objection exists, though the anthropological mainstream still rejects discrete biological races.
- Treating human ecology and epidemiological anthropology as descriptive add-ons. Their value is the biocultural cases (kuru and cannibalism, lactase persistence and dairying, sickle cell and agriculture) that show genes, culture and environment shaping each other, and their direct application to tribal and rural health.