4. Ancient DNA Study Reveals Human Evolution Continued During Last 10,000 Years - Geography

1. Why in News

1. Major study analysed ancient and modern human genomes understanding human evolution during last 10,000 years
2. Found natural selection continued shaping human evolution despite relatively recent timeframe
3. Key findings - Blood group genetics changes, coeliac disease susceptibility rise, skin colour adaptation, disease resistance evolution
4. Demonstrates evolution continuous and dynamic process
5. Implications for medical science, South Asian ancestry understanding

2. What is Ancient DNA?

Definition

1. Genetic material extracted from skeletal remains, teeth, bones of humans who lived thousands of years ago
2. Preserved in cool, dry conditions (permafrost, caves, arid deserts)
3. Degraded over time but sufficient for sequencing

Methodology

1. Scientists sequence ancient DNA using next-generation sequencing technologies
2. Compare with modern genomes identifying genetic changes over time
3. Statistical methods accounting for DNA degradation, contamination

Applications

1. **Understanding human migration -** Tracking population movements across continents (Out of Africa, Indo-European migrations, peopling of Americas)
2. **Evolutionary adaptations -** Identifying genetic changes in response to environmental pressures (climate, diet, altitude)
3. **Disease susceptibility -** Tracing origins of disease-related genetic variants
4. **Changes in physical and behavioural traits -** Height, skin colour, lactose tolerance, cognitive abilities

Challenges

1. DNA degrades exponentially; ancient samples highly fragmented
2. Contamination from modern DNA (excavators, lab personnel)
3. Limited samples from tropical regions (poor preservation in hot, humid climates)
4. Ethical concerns (indigenous communities' consent for studying ancestral remains)

3. Carbon Dating

Carbon-14 Dating Basics

1. Scientists use Carbon-14 (C-14) dating to determine age of ancient skeletal remains
2. Radiocarbon dating method applicable to organic materials up to ~50,000 years old

Formation of Carbon-14

1. Carbon-14 is radioactive isotope formed by interaction between cosmic rays and atmospheric nitrogen
2. Cosmic rays (high-energy particles from space) strike upper atmosphere
3. **Nuclear reaction -** Nitrogen-14 (N-14) + neutron → Carbon-14 (C-14) + proton
4. Carbon-14 enters carbon cycle; absorbed by plants through photosynthesis, animals through food chain
5. Living organisms maintain constant C-14 to C-12 ratio (equilibrium with atmosphere)

Radioactive Decay After Death

1. Amount of Carbon-14 decreases after death because radioactive decay converts it back into nitrogen
2. Organism stops absorbing new C-14; existing C-14 decays exponentially
3. **Half-life -** 5,730 years (time for half of C-14 atoms to decay)
4. After 5,730 years - 50% C-14 remains
5. After 11,460 years - 25% remains
6. After 17,190 years - 12.5% remains

Measurement and Age Estimation

1. Scientists use mass spectrometers (Accelerator Mass Spectrometry - AMS) to measure isotope ratios (C-14/C-12)
2. Compare with atmospheric C-14 levels (calibrated using tree rings, coral reefs, lake sediments)
3. Estimate age of remains based on degree of C-14 decay
4. **Formula -** Age = $(\ln(N_0/N) / \lambda)$ where N_0 = initial C-14, N = current C-14, λ = decay constant

Limitations

1. Effective range - ~500 to ~50,000 years (beyond 50,000 years, too little C-14 remains)
2. Contamination risks (modern carbon from soil, preservation chemicals)
3. Calibration needed (atmospheric C-14 levels varied historically due to solar activity, volcanic eruptions, fossil fuel burning)

4. Major Findings of the Study

Continuing Natural Selection

1. Study found natural selection continued shaping human evolution during last 10,000 years (Holocene epoch, post-Last Glacial Maximum)
2. Contrary to assumption that cultural evolution (agriculture, technology, medicine) replaced biological evolution
3. Several genetic variants increased or decreased in frequency because of environmental and disease-related pressures
4. **Selection pressures -** Climate change, agricultural transition, infectious disease epidemics, dietary shifts
5. Evolution ongoing despite relatively short timeframe (10,000 years = ~400 human generations)

Changes in Blood Group Genetics

1. Frequency of B blood-group variant increased in Western Eurasia over last 6,000 years
2. A blood-group variant declined during same period
3. **Possible explanations -**
    1. Disease resistance - Different blood groups confer resistance to different pathogens (malaria, plague, cholera, smallpox)
    2. Example - Type O resistant to severe malaria; Type A susceptible to plague
    3. Agricultural transition - Dietary changes affecting metabolism and disease susceptibility
    4. Population migrations - Indo-European steppe migrations bringing B variant to Europe
4. Blood group frequencies vary geographically today reflecting historical selection

Rise in Coeliac Disease Susceptibility

1. Variant of HLA-DQB1 gene associated with coeliac disease increased significantly over last 4,000 years
2. **Coeliac disease -** Autoimmune disorder where gluten consumption triggers immune attacks on small intestine in affected individuals
3. Gluten found in wheat, barley, rye (staple grains post-agricultural revolution ~10,000 years ago)
4. **Paradox -** Why did disease-causing variant increase despite agriculture?
5. **Researchers' finding -** Agriculture alone cannot explain this increase

6. **Possible explanations -**
    1. Balancing selection - HLA-DQB1 variant protects against certain infections (plague, tuberculosis) outweighing coeliac disease cost
    2. Hygiene hypothesis - Reduced pathogen exposure in agricultural societies altered immune system development
    3. Recent dietary changes - Modern gluten-heavy diets different from ancient grain consumption patterns
    4. Drift - Random genetic drift in small populations

Skin Colour Adaptation

1. Humans increasingly selected for lighter skin tones around 8,000 years ago (post-agricultural transition in Europe)
2. **Mechanism -** Lighter skin helped populations living in regions with low sunlight synthesize more vitamin D
3. **Vitamin D synthesis -** Requires UV-B radiation; darker skin (melanin) blocks UV, reducing vitamin D production
4. Low vitamin D causes rickets (bone deformities), impaired immunity, reproductive issues
5. **Agricultural intensification -** Agricultural diets poor in vitamin D (compared to hunter-gatherer diets rich in fish, animal organs containing vitamin D)
6. May have strengthened adaptation toward lighter skin
7. **Genes involved -** SLC24A5, SLC45A2, TYRP1 (depigmentation alleles increased frequency in European populations)
8. **Geographic variation -** Skin colour correlates with latitude; equatorial populations retained dark skin (UV protection), high-latitude populations lighter skin (vitamin D synthesis)

Evolution of Disease Resistance

1. CCR5-Δ32 gene variant provides resistance against HIV-1 infection
2. Frequency of this variant increased thousands of years before HIV emerged (HIV pandemic - 1980s; CCR5-Δ32 ancient)
3. **CCR5 gene -** Encodes chemokine receptor on immune cell surface; HIV uses CCR5 as entry point
4. **Δ32 deletion -** 32-base-pair deletion creating non-functional receptor; homozygous individuals (~1% Northern Europeans) nearly immune to HIV; heterozygous (~10-15%) slower disease progression
5. Ancient infectious diseases likely drove spread of this protective gene
6. **Candidates -** Bubonic plague (Yersinia pestis), smallpox (Variola virus) hypothesized to Favor CCR5-Δ32 carriers
7. Recent research suggests plague selected for CCR5-Δ32 during medieval Black Death epidemics
8. **Example of "genetic rescue" -** Variant beneficial against historical pathogen now protective against modern pandemic

Other Possible Findings (Typical in Such Studies)

1. Lactose tolerance (LCT gene) - Increased in European, African, Middle Eastern pastoralist populations post-dairy farming (~7,500 years ago)
2. Height variation - Selection for different statures in different environments
3. High-altitude adaptation - Tibetan EPAS1, Andean EGLN1 variants improving oxygen utilization
4. Immune system genes (HLA complex) - Rapid evolution responding to local pathogen environments

5. Significance of the Study

Understanding Human Adaptation

Study explains how humans adapted to -

1. **Climate change -** post-glacial warming, regional climate variations
2. **Diseases -** Epidemics of plague, smallpox, malaria shaping immune genes

3. **Agriculture -** Transition from hunting-gathering to farming (~10,000 years ago in Fertile Crescent, spreading globally)
4. **Dietary transformations -** Increased grains (gluten), dairy (lactose), reduced meat/fish
5. Demonstrates evolution is continuous and dynamic process, not static
6. Humans continue evolving despite short timeframe (evolutionary biology traditionally focuses on millions of years)

Importance for Medical Science

Ancient DNA research improves understanding of -

1. **Immunity -** Why certain populations resistant/susceptible to diseases (COVID-19 severity varied by ancestry)
2. **Genetic diseases -** Origins and spread of disease alleles (coeliac, diabetes, cardiovascular disease)
3. **Long-term health patterns -** How historical diets, lifestyles affect modern health
4. **Precision medicine -** Tailoring treatments based on ancestral genetic backgrounds
5. **Pharmacogenomics -** Drug responses varying by ancestry-specific genetic variants
6. **Public health -** Predicting disease risk in different populations

Relevance for South Asia

South Asians possess ancestry from multiple ancient populations -

1. **Iranian farmers -** Agricultural migrants from Zagros Mountains (~7,000 years ago)
2. **Steppe pastoralists -** Indo-European speakers from Eurasian steppes (~4,000 years ago)
3. **Ancient South Indians (AASI) -** Indigenous hunter-gatherers, earliest inhabitants
4. **East Asian-related populations -** Tibeto-Burman speakers in Northeast India

Complex admixture creating genetic diversity

Ancient DNA studies in India can improve understanding of -

5. **Migration -** Aryan migration debate, Harappan civilization ancestry, Austroasiatic (Munda) origins
6. **Adaptation -** Lactose tolerance in pastoral communities, malaria resistance in tribal populations
7. **Disease history -** Diabetes susceptibility (South Asians highest Type 2 diabetes rates globally), cardiovascular disease genetics

Other Significance

1. **Cultural heritage -** Connecting modern populations to ancient civilizations (Harappan, Vedic, etc.)
2. **Linguistic origins -** Linking genetic ancestry to language families (Indo-European, Dravidian, Austroasiatic, Tibeto-Burman)
3. **Caste and tribal genetics -** Understanding genetic structure of endogamous groups
4. **Forensic applications -** Identifying unknown remains, solving historical mysteries

6. Concerns

Risk of Misinterpretation

1. Genetic findings can be oversimplified or misused to justify racial or cultural superiority
2. **Historical precedent -** Eugenics movements, Nazi racial ideology misused genetics

Modern risks -

3. Claiming genetic basis for intelligence, criminality, social success
4. Nationalistic narratives distorting migration histories (Aryan invasion/migration politicized in India)
5. Ethnic supremacy claims ("pure" ancestry myths)
6. **Reality -** Human genetic variation is continuous, not discrete racial categories
7. No genetic basis for superiority; traits polygenic (many genes), environmentally influenced

Limited Geographical Representation

Most ancient DNA studies concentrated in Europe and Western Eurasia

Reasons -

1. Better DNA preservation (cool, dry climates in Northern Europe, Siberia)
2. Historical research focus and funding (European/American institutions)

3. Accessibility of archaeological sites

Large regions remain underrepresented -

4. **South Asia -** Despite large population, few ancient genomes (Rakhigarhi Harappan genome major breakthrough)
5. **Africa -** Tropical climates degrade DNA; despite being human origin continent, limited ancient data
6. **Southeast Asia -** Hot, humid conditions; complex island geography
7. **Latin America -** Improving but still limited (Inca, Maya, pre-Columbian genomes)
8. **Consequences -** Eurocentric bias in understanding human evolution; missing diversity

Other Concerns

1. **Ethical issues -** Indigenous communities' consent for studying ancestral remains; repatriation demands
2. **Cultural sensitivities -** Religious beliefs about disturbing dead; Western vs indigenous knowledge systems
3. **Commercialization -** Direct-to-consumer genetic testing companies overselling ancestry tests
4. **Privacy -** Ancient DNA databases could be misused for surveillance, discrimination
5. **Sampling bias -** Over-representation of elite burials (rich graves better preserved); missing common people's genetics

7. Way Forward

Expand Geographic Coverage

1. Increase ancient DNA research in underrepresented regions (South Asia, Africa, Southeast Asia, Latin America)
2. Develop methods for extracting DNA from poorly preserved remains (tropical climates)
3. Build local capacity (labs, trained personnel) in Global South countries
4. International collaborations ensuring equitable partnerships

Ethical Guidelines

1. Obtain informed consent from descendant communities before studying ancient remains
2. Involve indigenous communities in research design, interpretation
3. Repatriate remains respectfully after study (where communities request)
4. Transparent data sharing with source communities

Combat Misuse

1. Science communication emphasizing complexity, avoiding oversimplification
2. Countering racist, supremacist narratives misusing genetics
3. Educational programs on human genetic diversity
4. Interdisciplinary approaches (genetics + archaeology + linguistics + anthropology)

South Asia Priorities

1. Large-scale ancient DNA projects across Indian subcontinent (Harappan, Vedic, historical periods)
2. Study genetic diversity within India (tribal populations, caste groups, regional variations)
3. Understand disease genetics (diabetes, cardiovascular disease) in evolutionary context
4. Preserve archaeological sites, skeletal collections for future research

Technological Advances

1. Improve DNA extraction techniques (degraded samples, contamination reduction)
2. Lower sequencing costs making large-scale studies feasible
3. Develop computational methods handling complex admixture
4. Ancient pathogen DNA (palaeomicrobiology) revealing historical epidemics

8. Conclusion

Ancient DNA study analysing genomes from last 10,000 years reveals natural selection continued shaping human evolution through blood group changes, coeliac disease susceptibility rise, lighter skin adaptation for vitamin D synthesis, and CCR5-Δ32 disease resistance evolution. Significance includes

understanding human adaptation to climate-disease-agriculture-diet, medical science applications in immunity-genetic diseases-health patterns, and South Asian relevance with Iranian farmer-steppe pastoralist-AASI-East Asian ancestry. Concerns include misinterpretation risks justifying racial superiority and limited geographic representation (Europe-focused, underrepresenting South Asia-Africa-Southeast Asia). Way forward requires expanding geographic coverage, ethical guidelines with community consent, combating genetic misuse, South Asian ancient DNA projects, and technological advances in degraded sample extraction ensuring equitable, ethical research transforming understanding of human history-health-adaptation.

Source - **https -//www.thehindu.com/sci-tech/science/revealed-how-humans-evolved-in-the-last-10000-years-alone/article70988163.ece# -~ -text=Ancient%20genes%**