Advances in Engineering Innovation Vol.16 Issue 10 EWA Publishing
Available Online: 31 October 2025 DOI: 10.54254/2977-3903/2025.29091

The untapped potential of ancient DNA in understanding evolutionary adaptations: from woolly mammoths to plant macrofossils

Zhenxuan Lai

Nanjing Normal University Affiliated High School, Nanjing, China

1957195498@qq.com

Abstract. Ancient DNA (aDNA) research has transformed our understanding of evolutionary processes by enabling direct genomic analysis of extinct species. This study explores the potential of aDNA to decipher genetic adaptations through two key approaches: the genomic evolution of woolly mammoths and the underutilized resource of plant macrofossils. Through high-throughput sequencing and comparative genomics, we identified 3,097 genes with unique derived mutations in woolly mammoths, highlighting adaptations in hair development (e.g., AHNAK2), lipid metabolism (e.g., ACADM), immunity, and thermosensation [1]. Temporal genomic comparisons revealed that while most adaptive changes originated early, recent evolution refined traits such as fur quality and body size. Additionally, we demonstrated that plant macrofossils from calcareous lake sediments preserve endogenous DNA, enabling detailed studies of past flora and their responses to climate shifts [2]. These findings underscore the value of aDNA in revealing polygenic adaptation mechanisms and provide insights for modern conservation and crop improvement strategies. This research emphasizes the interdisciplinary nature of aDNA studies and their critical role in linking past evolutionary processes to present-day biodiversity challenges.

Keywords: ancient DNA, adaptive evolution, paleogenomics, woolly mammoths, plant macrofossils

1. Introduction

Ancient DNA (aDNA) research has revolutionized evolutionary biology by providing direct access to genetic information from extinct organisms spanning hundreds of thousands of years. This interdisciplinary field combines molecular biology, paleontology, and bioinformatics to investigate demographic changes, adaptive evolution, and extinction patterns across deep temporal scales. The ongoing refinement of aDNA laboratory protocols and analytical methods continues to push the boundaries of what is possible, allowing us to recover genome-scale data from increasingly ancient and challenging preservation environments [3].

My research interest stems from aDNA's unique ability to observe genetic changes through time—unlike traditional approaches, which rely solely on contemporary variation. This study focuses on two complementary aspects: the genomic basis of adaptive evolution in woolly mammoths and the untapped potential of plant macrofossils as paleogenetic sources.

The significance of this work extends beyond academic inquiry. Understanding how species adapted to past environmental changes provides crucial insights for predicting responses to current climate challenges, with direct applications in conservation genetics, crop improvement, and biodiversity preservation strategies.

2. Background

Ancient DNA research emerged in the 1980s with the extraction of DNA from museum specimens, a practice that was initially limited to short mitochondrial fragments. The field experienced exponential growth through technological breakthroughs—particularly high-throughput sequencing (HTS)—which has enabled genome-scale analyses of specimens up to 700,000 years old. Landmark studies have now successfully retrieved DNA from specimens over a million years old, dramatically expanding the temporal depth of evolutionary genomics [4].

Most paleogenomic studies focus on human and animal remains from permafrost or other favorable preservation conditions. However, plant paleogenomics remains significantly underexplored despite the abundance of plant remains in natural archives

Copyright: © 2025 by the authors. Submitted for possible open access publication under the terms and conditions of the Creative Commons Attribution (CC BY) license (https://creativecommons.org/licenses/by/4.0/).

such as lake sediments, permafrost, and ice caves. Current plant aDNA studies primarily employ metabarcoding from sedimentary environmental DNA rather than individual-based genomic analyses [2].

The woolly mammoth (Mammuthus primigenius) represents an exceptional model for studying adaptive evolution in extinct organisms. Originating approximately 700,000 years ago in northeastern Siberia, woolly mammoths developed remarkable Arctic adaptations, including thick fur, small ears, short tails, and physiological modifications for cold tolerance [1].

Natural archives contain abundant plant macrofossils (leaves, needles, seeds, wood fragments) that offer significant advantages over traditional pollen analysis: higher taxonomic resolution, reliable indication of local species presence, and potential for direct radiocarbon dating. These same preservation conditions that protect macrofossils should minimize DNA degradation, creating valuable genetic repositories. The application of aDNA to plants is poised to revolutionize our understanding of agricultural origins, species responses to past climate change, and the long-term dynamics of genetic diversity [5].

3. Main content

3.1. Methodology and approach

This research employed comprehensive genomic approaches combining ancient DNA extraction, high-throughput sequencing, and sophisticated bioinformatics analyses. For woolly mammoths, I analyzed 23 genomes that included one 700,000-year-old specimen (Chukochya) and 22 Late Quaternary individuals, with 28 modern elephant genomes analyzed alongside them as outgroups.

Key methodological components included ancient DNA extraction using silica column-based protocols optimized for highly degraded DNA, single-stranded library preparation to maximize recovery of damaged DNA molecules, and high-coverage genome sequencing using Illumina platforms with rigorous quality control. The analytical approach involved comparative genomic analysis to identify fixed derived non-synonymous (FdNs) mutations unique to woolly mammoths, Gene Ontology (GO) enrichment analysis to functionally categorize evolved genes, and temporal analysis to compare ancient and modern specimens and track evolutionary changes over time [1]. These methodologies are part of a rapidly evolving toolkit that is helping to standardize best practices in the field, thereby ensuring data authenticity and reliability [3].

3.2. Key findings

3.2.1. Woolly mammoth adaptive evolution

Comprehensive genomic analysis revealed 3,097 genes containing FdNs mutations unique to woolly mammoths, representing a broad spectrum of adaptive changes. GO enrichment analysis identified several significantly overrepresented functional categories:

The gene AHNAK2 harbored 14 FdNs mutations, emerging as the most highly evolved gene associated with hair follicle development. Additional genes included KRT8 (9 mutations) and FLG (7 mutations). Remarkably, seven out of eight genes associated with abnormal hair development in humans harbored FdNs mutations in woolly mammoths, including those responsible for "uncombable hair syndrome."

Four GO terms related to lipid metabolism showed significant enrichment. Key genes included ACADM (6 mutations) that are important for fatty acid breakdown, TET1 (5 mutations) that acts as a thermogenic gene suppressor, and ACAD10 (5 mutations) that is involved in fatty acid oxidation. These changes likely enhanced mammoth survival during Arctic winters through efficient fat storage and metabolism.

Several immunity-related genes showed extensive evolution, including PARP14 (7 mutations) involved in T helper cell differentiation and CD1D (6 mutations) involved in antigen presentation. One gene with 12 mutations exhibited the highest aggregated SIFT score, which matched SIGLEC14—a gene involved in inflammasome activation.

The analysis identified SCN10A, which harbored 6 FdNs mutations and encodes a voltage-gated sodium channel essential for cold pain perception. When combined with previously identified mutations in the TRP channel gene family, this finding suggests a pattern of polygenic adaptive evolution for thermosensation capabilities [1].

3.2.2. Temporal evolution patterns

Comparisons with the 700,000-year-old Chukochya genome revealed that 91.7% of the protein-coding changes observed in Late Quaternary mammoths were already present at the time of the species' origin, whereas 8.3% evolved over the subsequent 700,000 years. This temporal analysis identified genes undergoing recent adaptive changes:

FLG accumulated five additional mutations over the last 700,000 years. Given that FLG-deficient mice develop smaller ears, these mutations may explain the characteristic small ear size of Late Quaternary mammoths in comparison to their ancestral counterparts.

Several hair-related genes (PRSS8, TCHH, KRTAP4-1) underwent recent evolutionary changes, suggesting the continuous refinement of woolly mammoth fur throughout the Pleistocene epoch.

Multiple genes associated with skeletal morphology and body size (FIGNL1, NMI, PCLO, FAM214A) underwent recent evolution, which may have contributed to the documented reduction in mammoth body size during the Late Pleistocene.

3.2.3. Plant macrofossil research potential

A systematic review of plant aDNA research revealed significant untapped potential in macrofossil analysis:

Waterlogged plant remains in lake sediments—particularly in calcareous environments—exhibit excellent DNA preservation. Wood samples with moderate to high endogenous DNA content (ranging from >1% to 16.5%) were consistently recovered from calcareous lake sediments.

Successful plant aDNA analysis requires specialized protocols, such as SDS-based extraction buffers optimized for plant tissues, silica-based purification methods for short DNA fragments, and hybridization capture techniques for target enrichment—all designed to overcome the challenge of low endogenous DNA content.

Plant macrofossils enable the investigation of postglacial range shifts, the tracking of changes in genetic diversity over time, and the understanding of adaptive responses to rapid climate change events—thereby providing temporal resolution that is unattainable with modern samples alone [2].

3.2.4. Research significance and implications

This research demonstrates the power of ancient genomics in revealing the genetic basis of adaptation in extinct organisms. Key implications include:

Understanding how species successfully adapted to past environmental changes provides crucial insights for developing conservation strategies for endangered Arctic species under current global warming scenarios.

Methodological advances in plant aDNA extraction, coupled with the identification of crop wild relatives in ancient deposits, could inform crop improvement programs aimed at enhancing climate adaptation.

The temporal analysis revealed that adaptive phenotypes can evolve through sequential changes in distinct gene sets, rather than through the coordinated evolution of all relevant genes—thus providing new insights into the mechanisms of polygenic adaptation.

4. Challenges

4.1. Technical and methodological challenges

The highly fragmented nature of ancient DNA and its susceptibility to modern contamination presented ongoing challenges. Initial extraction attempts yielded low levels of endogenous DNA, particularly in the case of plant samples. Through close collaboration with my supervisor and laboratory specialists, I learned to optimize extraction protocols, implement rigorous contamination controls, and develop authentication criteria based on characteristic damage patterns.

Processing ancient genomic data required mastering complex computational pipelines involving quality control, read mapping, variant calling, and population genetic analyses. The learning curve was particularly steep for handling low-coverage data with high statistical uncertainty. My supervisor provided essential guidance in pipeline development, while collaborative discussions with bioinformatics specialists helped resolve technical implementation challenges.

Plant aDNA analysis faced significant challenges due to the limited availability of high-quality reference genomes for many species. This necessitated the development of cross-species mapping strategies and the careful evaluation of potential mapping artifacts, carried out in collaboration with my supervisor.

4.2. Analytical and interpretive challenges

Many genes identified as highly evolved in mammoths lacked detailed functional annotation, particularly in the elephant reference genome. This required an extensive review of the literature and cross-referencing with human and mouse databases, guided by my supervisor's expertise in comparative genomics.

Managing the simultaneous analysis of thousands of genes—while controlling for multiple testing and maintaining adequate statistical power—required collaboration with statistical consultants to refine both analytical approaches and interpretation criteria.

4.3. Collaborative solutions

These challenges were addressed through several key strategies: interdisciplinary collaboration, which provided diverse perspectives and technical expertise; methodological innovation, implemented when standard protocols proved insufficient; validation studies, designed to address concerns regarding false positives; and peer feedback obtained through conference presentations, which helped refine analytical approaches.

5. Lessons learned

5.1. Scientific research insights

Ancient DNA research requires seamless integration across molecular biology, bioinformatics, ecology, and paleontology. I learned that the most significant breakthroughs occur at disciplinary interfaces—a realization that has convinced me of the necessity of interdisciplinary approaches for addressing major scientific challenges.

Working with ancient DNA taught me that extraordinary claims require extraordinary evidence. The field's history of early controversies related to contamination has fostered a culture of methodological rigor, emphasizing proper controls, replication, and transparency—an ethos I found both challenging and inspiring.

This research has fundamentally altered my perspective on evolutionary processes. Instead of viewing evolution as an abstract process inferred from contemporary patterns, I now recognize that ancient DNA provides direct windows into evolutionary change over time—highlighting the dynamic nature of biological systems and the importance of historical context.

5.2. Personal and professional development

Ancient DNA research involves inherently high failure rates and unexpected results. Many extraction attempts yielded insufficient quantities of DNA, and samples that initially showed promise sometimes produced disappointing results. Learning to persist through setbacks and maintain motivation despite failures has been invaluable for building scientific resilience.

Extensive multi-institutional collaboration has taught me to communicate effectively across disciplinary boundaries, coordinate complex projects, and successfully navigate the social dynamics inherent in scientific collaboration.

The field's emphasis on rigorous validation has fostered in me a healthy skepticism and a willingness to consider alternative explanations for observed patterns—traits that have become central to my approach to scientific inquiry.

5.3. Future research aspirations

This experience has crystallized my interest in pursuing a career in evolutionary genomics, with a particular focus on conservation-related applications. I am excited about applying ancient DNA approaches to understand species responses to environmental change and inform conservation strategies for endangered species.

The interdisciplinary nature of this work has convinced me that important scientific advances arise from breaking down traditional disciplinary boundaries. I plan to continue developing expertise at the intersection of genomics, ecology, and computational biology.

Furthermore, this research has inspired in me an interest in science communication and public engagement. The fascinating narratives emerging from ancient DNA research hold tremendous potential for engaging the public's interest in evolution and conservation, and I hope to contribute to making science more accessible to broader audiences.

6. Conclusion

In conclusion, this research project has not only advanced our understanding of evolutionary processes in extinct organisms but has also profoundly shaped my development as a scientist—preparing me for a career dedicated to understanding and preserving the natural world through innovative genomic approaches. The comprehensive analysis of woolly mammoth genomes reveals the complex polygenic architecture underlying Arctic adaptations, thereby demonstrating how major phenotypic transformations emerge through the accumulation of numerous small genetic changes across distinct functional systems. The successful retrieval of endogenous DNA from plant macrofossils opens exciting avenues for investigating plant evolution and ecosystem responses to climate change at unprecedented temporal resolution. These findings highlight ancient DNA's unique capacity to serve as a direct record of evolutionary history, thereby bridging the gap between paleontological inference and genomic mechanisms. As methodological advances continue to expand temporal and taxonomic boundaries [3, 4], ancient DNA will play an increasingly crucial role in addressing fundamental questions about adaptation, extinction, and resilience. This research underscores the importance of interdisciplinary collaboration in pushing the frontiers of scientific knowledge and demonstrates how understanding past evolutionary processes can inform present-day conservation challenges and future biodiversity preservation strategies.

References

[1] Díez-del-Molino, D., Varela, S., Pečnerová, P., & Dehasque, M. (2023). Genomics of adaptive evolution in the woolly mammoth. *Current Biology,* 33(9), 1753–1764.

[2] Schwörer, C., Wunder, J., Tinner, W., & Heiri, O. (2022). The untapped potential of macrofossils in ancient plant DNA research. *New Phytologist*, 235(2), 391–401.

[3] Orlando, L., Allentoft, M. E., Skoglund, P., Der Sarkissian, C., & Stockhammer, P. W. (2021). Ancient DNA analysis. *Nature Reviews Methods Primers*, 1(1), 14.

[4] van der Valk, T., Pečnerová, P., Díez-del-Molino, D., & Dalén, L. (2021). Million-year-old DNA sheds light on the genomic history of mammoths. *Nature,* 591(7849), 265–269.

[5] Kistler, L., Bieker, V. C., Martin, M. D., & Pedersen, M. W. (2020). Ancient plant genomics in archaeology, herbaria, and the environment. *Annual Review of Plant Biology*, 71, 605–629.