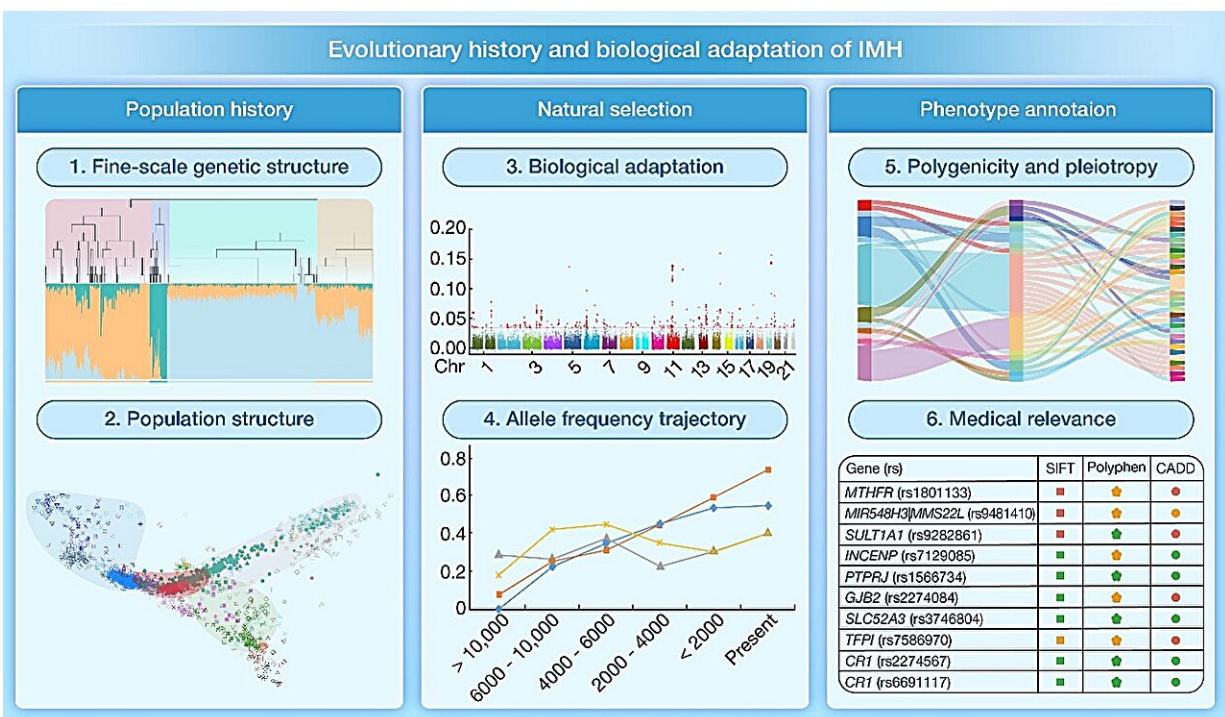


Research team reconstructs evolutionary history and biological adaptation of Han Chinese people on the Mongolian Plateau

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Demographic history reconstruction suggested that additional gene flow from Altaic people influenced the northernmost Han Chinese population. Complex biological adaptation signatures associated with metabolic biological functions were identified. The evolutionary trajectory of metabolic-adaptive genes suggested that diet shifts associated with agriculture have promoted rapid biological adaptation. Credit: Guanglin He, Institute of Rare Diseases, West China Hospital of Sichuan University, Sichuan University

A Chinese research team analyzed 5,583 modern and ancient individuals from an integrated genomic dataset to reconstruct the population evolutionary history and biological adaptation of the Han Chinese population across the Mongolian Plateau.

Their findings, [published](#) in *hLife*, demonstrate strong genetic homogeneity within the Han Chinese population on the Mongolian plateau, influenced by [gene flow](#) from surrounding Altaic-speaking populations.

Using various computational biology methods, the team characterized a comprehensive landscape of biological adaptations, identifying adaptative signatures related to complex metabolism traits. Additionally, a pan-ancestral analysis of ancient and modern genomes reconstructed the evolutionary trajectory of key mutations in genes related to fatty acid and folate metabolism over the past 50,000 years.

Comprehensive characterization of the effect of the complex demographic processes and natural selection pressures on the patterns of human genetic diversity is essential for understanding the human history of ethnolinguistically different populations and the evolutionary trajectory of the molecular genetic basis of adaptive traits and complex diseases.

"Large-scale population-specific ancient and modern genomic resources provide a direct spatiotemporal research window for elucidating the origin, migration, admixture, biological adaptation, and the geographical origins and diffusion trajectories of the genetic architecture of human diseases," says Chao Liu, an Academician at the Chinese Academy of Engineering and co-corresponding author from the Anti-Drug Technology Center of Guangdong Province.

Recent population [genetic studies](#) on the specific ancestry of ancient and

modern genomes in Western Eurasian populations have revealed the differential genetic contributions of late Upper Paleolithic European hunter-gatherers, Neolithic Anatolian agriculturalists, and Bronze Age Eurasian steppe pastoralists to the genetic basis of the complex traits such as waist-to-hip ratio and height in modern Europeans.

Path-specific local ancestry analyses of ancient and modern genomes in Europe have shown that the spread of Western steppe populations 5,000 years ago shaped the genetic susceptibility to multiple sclerosis in modern European populations, with higher prevalence in the north and lower in the south.

However, there is a relative scarcity of research based on ancient and modern genomic databases to dissect the geographical origins and evolutionary trajectories of diseases or biological adaptive traits in East Eurasians.

"To address this gap in eastern Eurasian populations and provide deeper insights into the [evolutionary history](#) of ancestrally different eastern Eurasian and the evolutionary origin of the population-specific genetic basis of complex biological traits, we explored ancestral composition and characterized the evolutionary trajectory of the adaptive traits in Han Chinese populations using an integrated modern and ancient genomic database," says Guanglin He, corresponding author from Sichuan University.

Population history

The Han is the largest ethnic group globally, distributed across China in diverse ecological environments and dietary patterns. The Mongolian Plateau in northern East Asia is home to both Sinitic-speaking Han and Altaic-speaking populations.

Historical records document various regimes in the region, including the Xiongnu (209 BCE–98 CE), Xianbei (386–534 CE), Türkic (552–742 CE), Uyghur (744–840 CE), and Khitan (916–1125 CE). Archaeological and genetic evidence suggests an extensive cultural and genetic interaction between the Han and these minority ethnic groups.

Mengge Wang, the co-first author from Sichuan University, said, "Population-specific genomic resources are important for reconstruction of population history and improve human health equity in precision medicine. Additionally, the population interaction on the Mongolian Plateau is very interesting, and we have done early research on the genetic origin and population history of Mongolian people.

"In this project, we mainly carried out population genetics focused on the population history and biological adaptation for the Han Chinese population on the Mongolian plateau, and found many interesting stories related to the evolution of key adaptative mutations."

Scientists comprehensively characterized the demographic history and biological adaptation of Han Chinese individuals on the Mongolian Plateau using the allele frequency spectrum and haplotype-resolved fragments.

The study revealed a pronounced genetic homogeneity among Han populations across various regions of the Mongolian Plateau. Compared to their counterparts from the Central Plains, the Han inhabitants of the Mongolian Plateau exhibit greater genetic drift with Altaic-speaking populations.

Advanced admixture modeling confirmed that the gene pool of the Han population on the Mongolian Plateau has been influenced by gene flow from populations related to Altaic-speaking groups.

Biological adaptation

The study identified natural selection signals linked to the cold environment of the Mongolian Plateau, shifts in subsistence strategy, and immunity changes under pathogen exposures through various computational modeling and detection methods.

"We performed multiple complementary statistical analyses, including allele frequency-based population branch statistics (PBS), pairwise fixation index (F_{ST}) estimates, haplotype-based cross-population extended haplotype homozygosity (XP-EHH), and the integrated haplotype score (iHS), to detect natural selection signals in the Han population of the Mongolian Plateau. The most significant were candidate genes related to metabolism, FADS, and MTHFR," says Xiangping Li from Kunming Medical University, the study's first author.

The FADS gene family encodes fatty acid desaturase enzymes, which regulate the synthesis of polyunsaturated fatty acids. The MTHFR gene encodes methylenetetrahydrofolate reductase, essential in the folate cycle.

Researchers used genomic resources with high spatiotemporal resolution from both ancient and modern samples to reconstruct the evolutionary trajectories of the most salient natural selection signals, [rs174550 (FADS1) and rs1801133 (MTHFR)], over tens of thousands of years.

The findings suggest that the frequency of the advantageous allele rs174550-T increased steadily after its emergence, stabilizing around 0.54.

Similarly, the allele rs1801133-A, which appeared in millet agricultural populations around 10 thousand years ago, reached high frequencies in regions near 40 degrees north latitude due to dietary shifts associated

with agriculture. In Chinese populations, the frequency of rs1801133-A declines continuously from north to south.

The development of cereal agriculture in the Near East's Fertile Crescent and millet farming in the Yellow River basin of East Asia accelerated the adaptation and stabilization of genetic diversity patterns linked to metabolic genes. Phenotypic association analysis of adaptive signals revealed polygenic adaptation and pleiotropy patterns in complex traits within the Han population of the Mongolian Plateau.

The advent of wheat and barley agriculture in the Near East and millet farming in the Yellow River basin spurred rapid adaptation and stabilization of genetic diversity in metabolic genes. Phenotypic association analyses of bio-adaptive signals revealed polygenic adaptation and pleiotropic patterns in complex traits within the Han population of the Mongolian Plateau.

"Our study enhances the understanding of how complex population genetic backgrounds and demographic events influence the genetic determinants of the disease and phenotypes, potentially advancing personalized precision medicine," says Liping Hu of Kunming Medical University.

More information: Xiangping Li et al, Evolutionary history and biological adaptation of Han Chinese people on the Mongolian Plateau, *hLife* (2024). [DOI: 10.1016/j.hlife.2024.04.005](https://doi.org/10.1016/j.hlife.2024.04.005)

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