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Population genetic history of the ancient reindeer-herding Ewenki people

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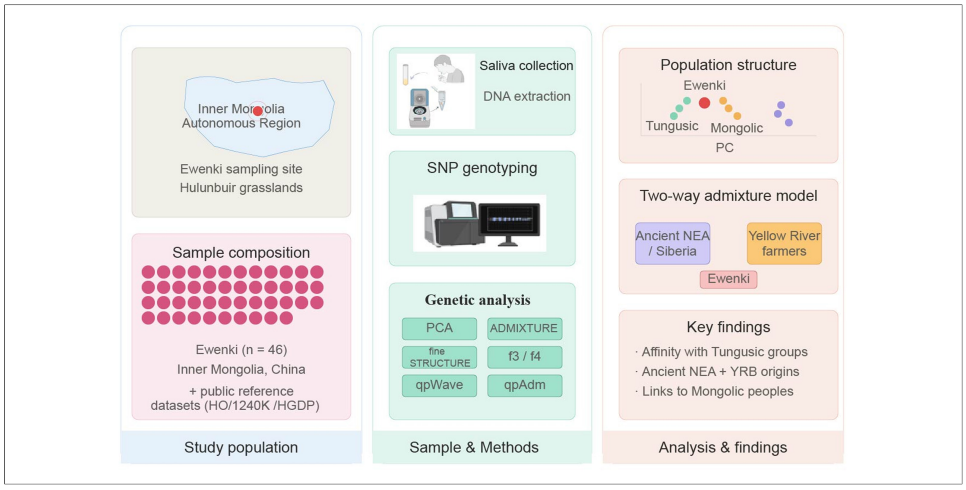

Abstract

Aim: High-quality genomic resources from underrepresented populations are essential for understanding human genetic origins, population structure, and demographic history. The Ewenki, an ethnolinguistic minority mainly inhabiting the high-latitude, cold regions of Northeast China, remain insufficiently characterized at the genome-wide level. This study aimed to investigate the population structure, ancestral composition, and demographic history of the Ewenki to provide insights into human genetic evolution in Northeast Asia.

Methods: We generated genome-wide single-nucleotide polymorphism (SNP) data from

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46 Ewenki individuals in Inner Mongolia and merged them with public modern and ancient genomic datasets. Population structure and demographic history were reconstructed through Principal component analysis (PCA), model-based ADMIXTURE, fineSTRUCTURE haplotype clustering, f3/f4 statistics, and qpWave/qpAdm modeling to infer ancestry composition and admixture events.

Results: The Ewenki occupy a genetically distinct position within Northeast Asia and show close genetic affinities with Chinese Mongolic, Tungusic-speaking, and ancient Northeast Asian populations. f3/f4 statistics revealed shared genetic drift and admixture signals linking the Ewenki to ancient Northeast Asian, Siberian, and Yellow River Basin-related populations. qpWave and qpAdm analyses further indicated that the Ewenki can be modeled primarily as a mixture of ancient Northeast Asian/Siberian-related ancestry and ancient Yellow River Basin farmer-related ancestry, reflecting long-term population interactions and admixture in Northeast Asia.

Conclusion: The Ewenki share significant genetic similarities with Tungusic-speaking populations, mainly resulting from admixture between ancient Northeast Asian groups and Yellow River Basin farmers.

INTRODUCTION

The historical underrepresentation of non-European populations in human genetic studies has constrained our understanding of global population history and the genetic architecture of complex traits, particularly for ethnolinguistically diverse groups in regions of long-standing demographic complexity[1,2]. Recent expansions of non-European genomic resources have begun to address these gaps, providing new insights into the demographic history and evolutionary trajectories of underrepresented populations across Africa, the Americas, Oceania, and Asia[3-7]. Within this broader effort, Eurasia and Northeast Asia in particular represents a critical region for closing such knowledge gaps, given its central position in major prehistoric migrations, sustained interactions among Tungusic, Mongolic, Turkic, and Sinitic populations, and its long history of forest-steppe-agricultural interface dynamics. Northeast Asia is a geographically and culturally diverse region shaped over centuries by migration, conflict, and ongoing interactions[8,9]. This region generally includes northeastern China, Japan, North Korea, South Korea, Mongolia, and the Russian Far East. Key ethnolinguistic groups include Han Chinese and various regional minorities such as Manchu, Daur, Hezhen, Nanai, Oroqen, Ewenki, Mongolians, Koreans, and Japanese, among others. Genetic studies of these underrepresented populations have provided important insights into their ancestral origins, migration patterns, genetic diversity, and population-specific health issues. These groups have experienced repeated episodes of migration, admixture, and cultural exchange along steppe, forest, and maritime routes.

The Inner Mongolia Autonomous Region (IMAR) is situated in northern China and has historically been a home to the Han and various other ethnic groups. From a geographic standpoint, its boundaries span a significant area, extending from the northeast to the southwest[10-14]. This distinctive position for examining gene flow and interactions among local ethnic communities provides ample opportunities for research in population genetics. On a global scale, the Mongolian people are widely recognized for founding the Mongol Empire during the 13th century under Genghis Khan. Historical records show that the Mongolian population had a significant impact on the genetic drift of the populations they met, especially after the empire's expansion and the conquests across a large portion of Eurasia[15]. The rise of the Mongol Empire, along with subsequent migrations and military-political integration across various periods, led to the incorporation of certain Ewenki groups into the Mongol system. The Ewenki and Mongolian peoples have historically resided in mixed settlements to the west of the Greater Khingan Mountains and within the Hulunbuir grasslands. Through their shared practices of herding, hunting, and migration, this integration

enabled coexistence with Mongol tribes, thereby facilitating gene flow[16]. Today, the Ewenki people residing in Inner Mongolia retain genomic signatures of their ancient ancestry while also showing evidence of long-term genetic interaction with surrounding populations of the Mongolian Plateau. They represent a unique and invaluable genetic marker within the pluralistic and unified framework of the Chinese nation.

The Ewenki are an indigenous Tungusic-speaking people from North Asia. They mostly live in northern China (Inner Mongolia and Heilongjiang provinces), Siberia, and Mongolia. Traditionally a reindeer-herding and forest-hunting people of the densely wooded Greater Khingan Mountains, the Ewenki have long inhabited high-latitude environments characterized by extended cold winters and limited daylight-an ecological context that, combined with their distinctive subsistence strategy, makes them an informative population for investigating the demographic history of Northeast Asian forest dwellers[17]. According to the 2020 census, their population is 34,617, with most living in the Hulunbuir region of the Inner Mongolia Autonomous Region and the Greater Khingan Mountains in Heilongjiang Province[18]. The Ewenki are among the most distinctive and northernmost ethnic minorities within the Tungusic-speaking groups in China. Historically, they inhabited forests, grasslands, and wetlands, fostering long-term contact, intermarriage, and cultural exchange with Mongolians, Han Chinese, and nearby ethnic groups. Frequent contact with neighbors has led to extensive cultural and genetic mixing. As a result, their genomes reflect a history of multi-source admixture and complex population migrations. Despite the rich demographic significance of the Ewenki, previous genetic studies on this population have been largely restricted to forensic short tandem repeat/insertion-deletion (STR/InDel) marker panels or limited mitochondrial and Y-chromosomal analyses, leaving the genome-wide population structure, fine-scale ancestral composition, and admixture history of the Ewenki insufficiently characterized. To our knowledge, no prior study has systematically integrated genome-wide single-nucleotide polymorphism (SNP) data from the Ewenki of the Inner Mongolia Autonomous Region with publicly available modern and ancient genomic datasets to reconstruct their detailed demographic history within the broader Northeast Asian context. To address these gaps, this study analyzes genome-wide data from Ewenki individuals to explore their ancient origins, migration pathways, population structure, and interactions with neighboring groups[19]. The results demonstrate how both ancient lineages and more recent gene flow have shaped the current Ewenki gene pool.

MATERIALS AND METHODS

Sample collection and ethical statement

We collected saliva samples from 46 Ewenki individuals residing in the Inner Mongolia Autonomous Region. All participants met the following inclusion criteria: (i) self-reported Ewenki ancestry for at least three generations with no known intermarriage with other ethnic groups; (ii) currently residing in the Hulunbuir region of IMAR; (iii) aged 18 years or older and in general good health at sample collection; and (iv) verified as unrelated to other participants based on KING2 kinship analysis (kinship coefficients < 0.0884, corresponding to second-degree or closer relationships), resulting in 46 unrelated individuals retained from the initial 46 samples for all subsequent population genetic analyses. All subjects provided written informed consent in accordance with the Declaration of Helsinki before participating. The study protocol was approved by the Medical Ethics Committee of West China Hospital, Sichuan University (Approval No. 2024-1463).

DNA sequencing, genotyping, and quality control

DNA was extracted and purified using the PureLink Genomic DNA Mini Kit (Thermo Fisher Scientific, USA). Genome-wide SNP genotyping was performed using the Illumina Infinium Global Screening Array (GSA) v3.0 (Illumina, San Diego, CA, USA), yielding approximately 600,000 high-quality SNPs across the genomes of all 46 individuals. Genotype calling was performed against the GRCh37 (hg19) human reference

genome to ensure compatibility with publicly available ancient and modern reference datasets (Human Origins, 1240K, and HGDP), all of which are coordinated on the same reference build. To ensure quality control, PLINK 1.9 was used with the following parameter settings: --mind 0.01, --geno 0.01, --hwe 0.001, and --maf 0.01. The minor allele frequency (MAF) threshold of 0.01 was chosen to retain low-frequency informative SNPs relevant for population structure analyses while excluding rare variants potentially affected by genotyping errors, following standard practice in population genomics studies of underrepresented populations[20]. Furthermore, family relationships were evaluated by calculating kinship coefficients using KING2. No pairs of individuals with kinship coefficients > 0.0884 (corresponding to second-degree or closer relationships) were identified, and one individual from each related pair was excluded from downstream analyses to ensure unrelatedness, resulting in all 46 individuals retained as unrelated samples for downstream analyses.

Data merging

We independently merged our newly generated Ewenki SNP data with three publicly available reference panels, resulting in three separate working datasets with distinct SNP densities and complementary analytical purposes:

(1) Human Origins (HO)_Ewenki dataset (245,657 SNPs): generated by merging with the HO panel; used for principal component analysis (PCA) and ADMIXTURE analyses, given its broad population coverage of modern Eurasian groups[21,22].

(2) 1240K_Ewenki dataset (95,353 SNPs): generated by merging with the 1240K dataset from the Allen Ancient DNA Resource (AADR v54.1.p1); used for f3, f4, qpWave, and qpAdm analyses involving ancient genomes[23,24].

(3) Human Genome Diversity Project (HGDP)_Ewenki dataset (525,456 SNPs): generated by merging with the HGDP panel; used for fineSTRUCTURE haplotype-based analyses requiring high SNP density.

All original publications contributing data to these merged panels are listed in Supplementary Table 1, in accordance with the AADR data usage guidelines.

PCA

To investigate clustering patterns within the studied and reference populations, we performed PCA using the smartPCA program in the EIGENSOFT v.6.1.4 package, based on an Illumina dataset comprising 111 modern and 77 ancient populations. Principal components were computed using the modern populations, while ancient samples were projected onto these axes by setting lsqproject: YES and shrinkmode: YES in the smartPCA parameter file. This projection-based approach mitigates potential bias from missing data and low coverage in ancient genomes, ensuring that PCA clustering accurately reflects population relationships rather than data quality differences. We applied LD pruning using the --indep-pairwise 200 25 0.4 option in PLINK 1.9. This window-based scheme (200-SNP sliding window, 25-SNP step size, $r^2 > 0.4$) was selected as a widely adopted compromise that effectively reduces marker correlation while retaining sufficient SNP density for downstream PCA and ADMIXTURE analyses. To further clarify the fine-scale structure of the populations, we performed an additional PCA on a combined, high-density Illumina dataset. The results were visualized in R version 4.5.1, where we used our in-house scripts to create a scatter plot.

Model-based unsupervised ADMIXTURE

We combined our dataset with a wide range of modern and ancient reference populations to examine the genetic characteristics of Ewenki individuals. In this analysis, we employed ADMIXTURE in its unsupervised mode[25], utilizing the standard 10-fold cross-validation (--cv = 10) to assess genetic heritage. To

handle linkage disequilibrium, we used PLINK 1.9, setting $r^2 > 0.4$ and --indep-pairwise 200 25 0.4 to ensure strong data integrity. Subsequently, we applied the admixture models across a series of specified ancestral sources (K = 2-20), running 100 bootstrap replicates using various random seeds to evaluate consistency and reliability. We selected the optimal models based on the lowest cross-validation error estimates, ensuring the most precise depiction of ancestral genetic contributions.

fineSTRUCTURE analysis

Prior to haplotype-based analysis, genotype data were statistically phased using SHAPEIT4 v4.2.2[26] with the 1000 Genomes Project Phase 3 reference panel to ensure accurate haplotype reconstruction. The phased data were then converted to ChromoPainter format, and the co-ancestry matrix was generated using ChromoPainter v2 under the linked model with default parameters. Fine-scale population structure was inferred with fineSTRUCTURE v4 using the following core parameters: -x 100000 (burn-in), -y 100000 (sampling iterations), and -z 1000 (thinning interval), with two independent Markov chain Monte Carlo (MCMC) runs to ensure convergence. The resulting maximum a posteriori (MAP) clustering tree was visualized using the built-in fineSTRUCTURE GUI tool, with branch lengths reflecting the inferred merging patterns of population clusters.

Allele-based shared ancestry estimation

Multiple analyses were performed using ADMIXTOOLS to explore the genetic composition and admixture history of the Ewenki people. Initially, we utilized the qp3pop package with its default settings and applied a block jackknife to calculate outgroup-f_3 statistics f_3 (source 1, Ewenki; Mbuti) and to evaluate shared genetic drift among 27 contemporary and 46 ancient populations from East Asia. Following this, admixture-f_3 statistics f_3 (source 1, source 2; targeted population) were calculated to detect signals of admixture in Ewenki samples derived from various modern and ancient East Asian sources. Significant admixture was identified when negative f_3 values were paired with Z scores below -3, suggesting that the targeted population was a combination of two parental groups, source 1 and source 2. Additionally, f_4 statistics in the form of f_4 (W, X; Y, outgroup) were used to assess potential indicators of admixture from different source populations into the targeted groups. For this purpose, we employed the qpDstat package in ADMIXTOOLS, including an extra parameter f_4 Mode (f_4: YES), and applied the block jackknife technique to approximate standard errors.

qpWave/qpAdm estimation

The qpWave/qpAdm software from ADMIXTOOLS was employed with default settings to determine the minimum number of ancestral populations and to compute admixture proportions. In the two-way qpAdm models, the candidate northern (Ancient Northeast Asian/Siberian) sources included AR13-10K, AR14K, Mongolia_North_N, DevilsCave_N, and Russia_Shamanka_Eneolithic, while the candidate southern (Yellow River Basin farmer-related) sources included Wadian_LN, Haojiatai_LN, Pingliangtai_LN, Lajia_LN, and Han_HGDP[27]. To improve model accuracy, we included a fixed panel of basal outgroups: Mbuti, Russia_Ust_Ishim, Russia_Kostenki14, Belgium_UP_GoyetQ116_1, Italy_North_Villabruna_HG, Israel_Natufian, Mixe, Onge, and Chokhopani. The allsnps: YES option was used to ensure comprehensive SNP inclusion.

Statistical analysis

Genome-wide SNP data were filtered using PLINK v1.9, and close genetic relatedness among individuals was evaluated using KING2. Linkage disequilibrium pruning was performed prior to PCA (smartPCA in EIGENSOFT) and unsupervised ADMIXTURE analyses, with the optimal K determined by the lowest 10-fold cross-validation error. Fine-scale population structure was further examined using ChromoCombine and fineSTRUCTURE. Population genetic statistics were computed with ADMIXTOOLS: outgroup-f3 and admixture-f3 assessed shared drift and admixture signals (Z < -3 indicating significant admixture); f4

statistics evaluated asymmetric allele sharing (|Z| > 3 considered significant); standard errors were estimated by block jackknife. qpWave assessed pairwise genetic homogeneity, and qpAdm estimated two-way ancestry proportions, with model acceptability evaluated by p-values, standard errors, and biological plausibility. All visualizations were generated using R.

RESULTS

Genetic clustering of the Ewenki

We genotyped approximately 600,000 genome-wide SNPs in 46 Ewenki individuals from IMAR and merged these data with publicly available ancient and modern genomic datasets, including the HO[28-31] resource and recently published ancient DNA data[32-35]. We grouped present-day individuals by language family: Tibeto-Burman-speaking (n = 159), Sinitic-speaking (n = 138), Tai-Kadai-speaking (n = 292), Hmong-Mien-speaking (n = 91), Austronesian-speaking (n = 120), Austroasiatic-speaking (n = 196), Mongolic-speaking (n = 144), and Tungusic-speaking (n = 64) [Supplementary Table 1]. We grouped ancient individuals by geographic origin [Figure 1A]. The map in Figure 1A was generated using ArcGIS software (Esri Inc., Redlands, CA, USA). Basemap data are derived from Esri's freely available online basemap service (© Esri and its data contributors), which permits non-commercial academic use. The Ewenki fall into a position that reflects genetic input from neighboring ethnic groups, while also preserving signals of ancient ancestry related to both Siberia and the Yellow River Basin (YRB). We performed PCA to characterize patterns of genetic structure and admixture, and to evaluate relationships between the Ewenki and other modern and ancient populations in Northeast Asia. We used 111 modern populations to compute the principal components and then projected 77 ancient populations onto these axes [Supplementary Figure 1]. PC1 separates northern groups, primarily Mongolic-speaking populations, from southern groups, including Hmong-Mien (HM)- and Tai-Kadai (TK)-speaking populations [Figure 1B]. The results reveal three major genetic clusters: one formed by Mongolic-speaking populations, one by Tungusic-speaking populations, and one by Ancient Siberian groups. These clusters are closely spaced and partially overlapping. Ancient Yellow River Basin (A-YRB) farmers cluster on the opposite side, close to the Ewenki.

Population structure of the Ewenki

We investigated the ancestral composition of the Ewenki using model-based ADMIXTURE on 1,287 modern and 266 ancient individuals from Northeast Asia [Figure 2]. Cross-validation identified six ancestral sources (K = 6) as the optimal model, which showed the lowest cross-validation (CV) error [Supplementary Table 2]. The dominant ancestral components in the Ewenki were Ancient YRB (dark cyan) and Ancient Siberia (pink). These results indicate that the Ewenki carry a complex ancestry primarily derived from Ancient YRB and Ancient Siberia. In contrast, populations from Southeast Asia, southern ethnic minorities, and the Tibetan-Burmese Plateau exhibit distinct combinations of genetic components. This indicates that the genetic structure of the Ewenki primarily originates from the Siberian forest-steppe zone, with later introductions of some Central Plains agricultural lineages. This phenomenon aligns with the Ewenki's geographical distribution, their Tungusic language affiliation, and their historical nomadic-hunting pastoral lifestyle in northern regions. The target population of this study, Ewenki_InnerMongolia, is highlighted in red to distinguish it from reference populations obtained from publicly available datasets in Figure 2.

We further performed a fineSTRUCTURE analysis among Tungusic speakers and their neighbors, comprising 110 individuals from different Tungusic-speaking groups, to explore the fine-scale genetic structure of the Ewenki [Figure 3]. The results revealed two primary genetic clusters: one encompassing Ewenki-related ethnic minority groups residing in Northeast China, and the other consisting of Ulchi, Negidal, and Nanai groups dwelling in the Amur River Basin. Notably, the Ewenki, Oroqen, and Hezhen formed a tightly clustered subgroup at a shallow branching level, indicating a high degree of genetic similarity among these populations. In contrast, the Amur River Basin groups (Ulchi, Negidal, Nanai) joined

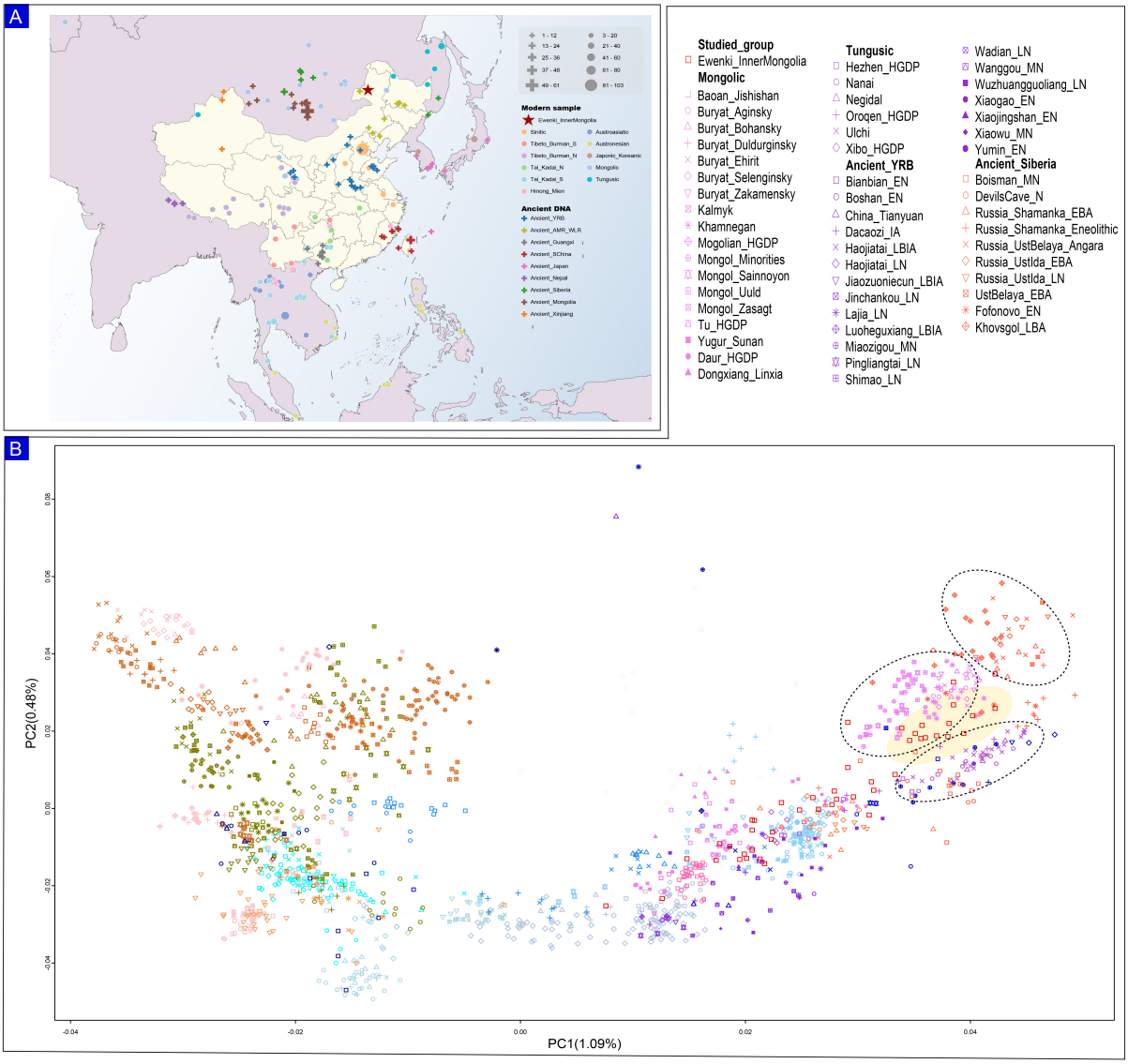

Figure 1. The geographical distribution and population patterns of the Ewenki. (A) The map displays the locations of recently gathered samples in comparison to reference populations [Supplementary Table 1]; (B) A principal component analysis (PCA) was conducted on the genetic characteristics of both contemporary and historical Northeast Asians, utilizing the combined Human Origins (HO) datasets. Distinct colors and unique shapes represent populations associated with various language families. YRB: Yellow River Basin; AMR: Amur River; WLR: West Liao River.

this cluster only at a deeper, more basal branching level, reflecting greater genetic differentiation between the two subgroups within the broader Tungusic-speaking complex.

Shared genetic drift and genetic composition of the Ewenki

We characterized the genetic relationships and ancestry composition of the Ewenki using outgroup f_3 statistics of the form f_3(source 1, Ewenki; Mbuti), which measured drift and thus allele sharing with a wide panel of modern and ancient reference populations. The Ewenki show elevated affinity with Tungusic and Mongolian populations [Figure 4A]. We then evaluated potential ancestral sources of the Ewenki using admixture-f_3 statistics of the form f_3 (source 1, source 2; Ewenki), testing different modern and ancient Northeast Asian populations as sources. We detect significant admixture signals ($Z < -3$) for pairs involving Sino-Tibetan and Altaic-speaking groups [Figure 4B]. Among ancient references, they share more alleles with the Ancient Siberian and Ancient Yellow River Basin Farmer groups [Figure 4C]. Using ancient populations, we also observe substantial signals ($Z < -3$) for admixture between Ancient Siberian herders and Ancient Yellow River Basin Farmers [Figure 4D]. Together, the admixture-f_3 results indicate that the Ewenki

derived from multiple ancestral sources and experienced complex historical interactions with neighboring groups. The detailed f3-statistics, including f3 values, standard errors, Z-scores, and *P*-values, are provided [Supplementary Tables 3 and 4].

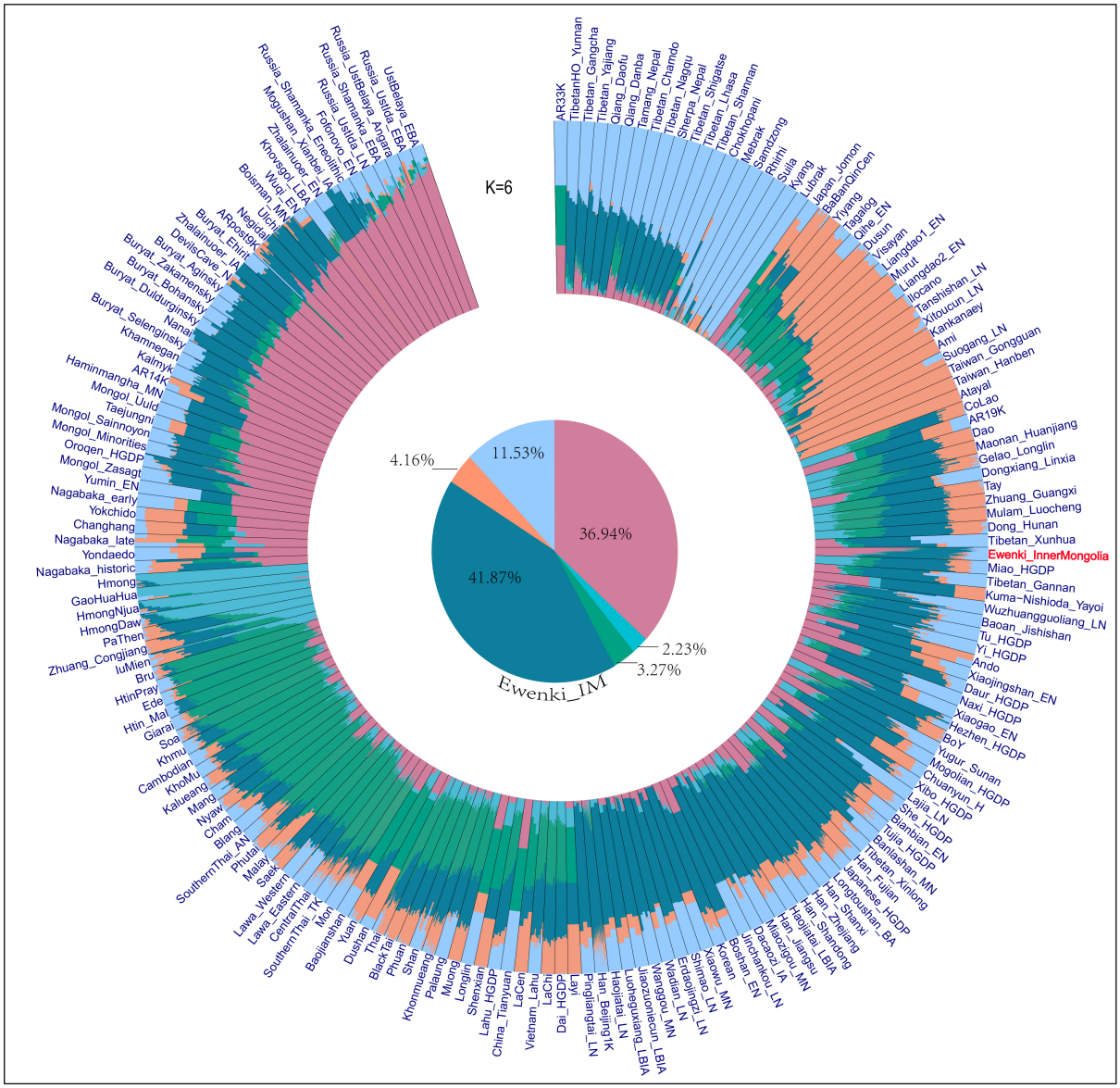

Figure 2. Model-based ADMIXTURE analysis of population structure. Ancestry proportions for K = 6 components were estimated in ancient and present-day Northeast Asian groups, with different colors indicating distinct ancestral components.

To detect the difference in gene flow between Ewenki and northern Han Chinese/Mongolia, we calculated f_4 (Han_North/Mongolia, Ewenki_InnerMongolia; Tungusic, Mbuti). The results indicated that the Ewenki exhibit a stronger affinity to Tungusic populations, such as the Ulchi, Negidal, and Nanai, compared to either Han North or Mongolian groups [Supplementary Tables 5-7]. Overall, the data support a genetic profile for the Ewenki that is firmly rooted in Tungusic ancestry, with an additional affinity for Sino-Tibetan groups when contrasted with Mongolians. To detect the difference in gene flow between Ewenki and ancient_YRB/ancient_Siberia/ancient_Northeast Asia (NEA), we calculated f_4 (ancient_YRB/ancient_Siberia, Ewenki_InnerMongolia; ancient_NEA, Mbuti). The results indicated that the Ewenki exhibit significantly

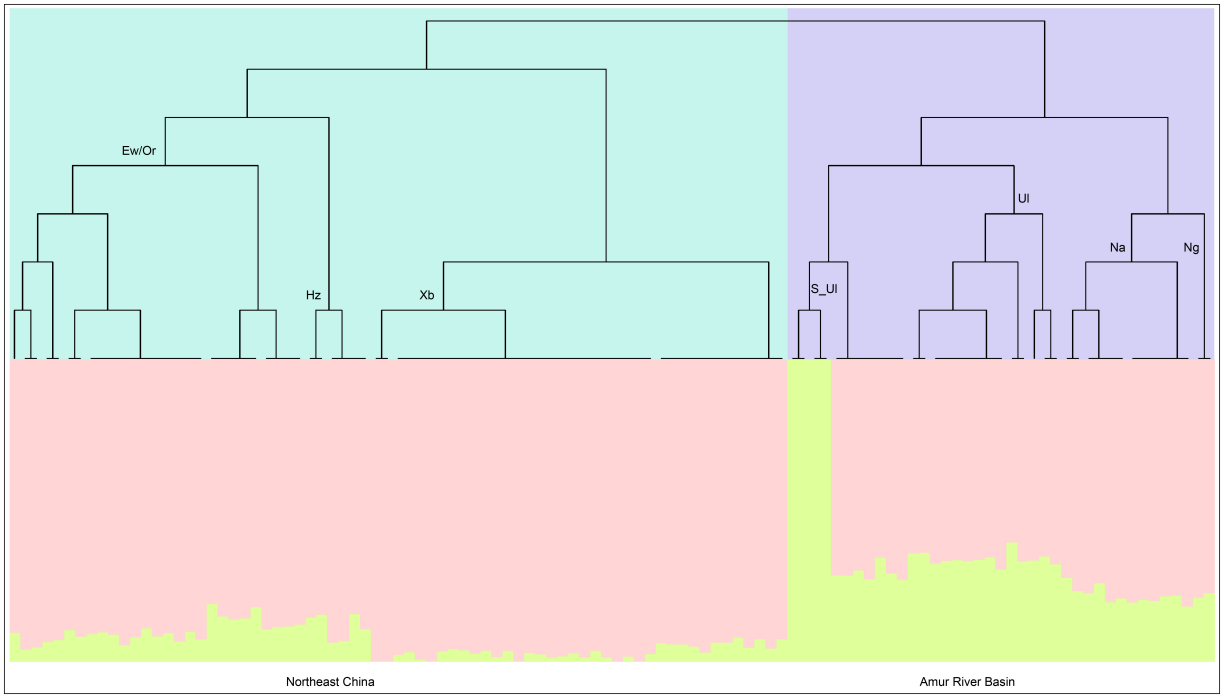

Figure 3. Population structure of the model-based fineSTRUCTURE. The fineSTRUCTURE-generated maximum a posteriori (MAP) tree illustrates clustering trends among 110 individuals from seven distinct groups in Northeast Asia. Adjacent to the tree, the ADMIXTURE plot for the same datasets represents two predefined ancestral sources. Abbreviations are as follows: Ew: Ewenki; Or: Oroqen; Hz: Hezhen; Xi: Xibo; Ul: Ulchi; Na: Nanai; Ng: Negidal.

greater genetic drift with Ancient Northeast Asian (ANA) and ancient Siberian populations than with ancient YRB farmers [Supplementary Tables 8-10]. We performed f_4 statistics in the form of f_4 (ancient_YRB, Ewenki_InnerMongolia; ancient_Siberia, Mbuti) to formally explore possible differentiated gene flow between Ewenki and ancient Siberia/ancient_YRB. Numerous tests showed strongly negative Z-scores, indicating a pronounced affinity of the Ewenki towards ancient Siberia relative to Neolithic Age farmers from the YRB. Conversely, when Y was set to ANA, comparisons with ancient Siberian groups often showed significantly negative values. This indicates that the Ewenki are genetically closer to ANA sources than to ancient Siberian populations. Collectively, these findings support substantial ANA ancestry in the Ewenki, stronger than that observed in ancient YRB farmers and ancient Siberia.

Admixture scenarios and gene flow in the Ewenki

Based on the evidence of gene flow within Ewenki populations, we initially applied qpWave on paired groups to formally test whether each pair of populations could be modeled as descending from the same number of ancestral source populations relative to a fixed set of outgroups, thereby assessing pairwise genetic homogeneity [Figure 5A]. The pairwise qpWave analysis was then expanded to include 72 populations to investigate the demographic relationships between the Ewenki and other ancient and modern groups. The results showed a strong genetic affinity between the Ewenki and ANA populations. Additionally, populations labeled as A-Siberia groups appeared genetically uniform, supporting either a recent common origin or significant admixture among ANA groups. Next, we applied qpAdm to estimate admixture proportions using predefined northern (Ancient Northeast Asian/Siberian) and southern (Yellow River Basin farmer) ancestral sources [Figure 5B and Supplementary Table 11]. In the model pairing Mongolia_North_N (northern source) with Han_HGDP (southern source), the Ewenki derived 79.8% of their ancestry from the northern source and 20.2% from the southern source. Similarly, in the model pairing AR13-10K (northern source) with Han_HGDP (southern source), the northern contribution reached 74.7%, with the remaining

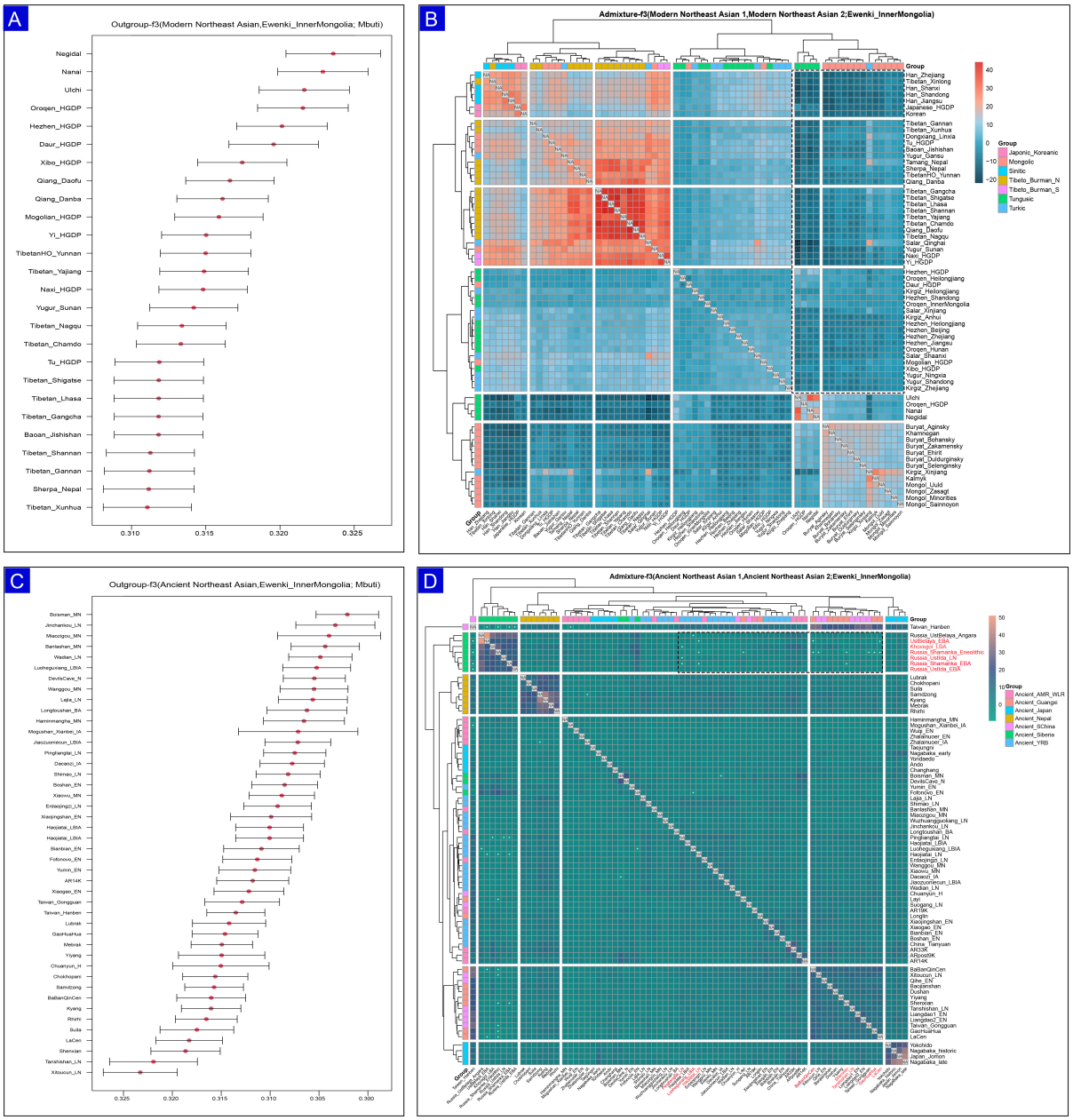

Figure 4. Outgroup and admixture-f3 statistics for the Ewenki. (A) Outgroup-f3 values of the form f3 (Modern Northeast Asian, Ewenki; Mbuti) were calculated using the HO_Illumina datasets to quantify the shared genetic drift with contemporary Northeast Asians; (B) Admixture-f3 tests of the form f3 (Modern Northeast Asian 1, Modern Northeast Asian 2; Ewenki) were employed to detect signals of Ewenki ancestry derived from pairs of modern Northeast Asian populations; (C) Outgroup-f3 statistics of the form f3 (Ancient Northeast Asian, Ewenki; Mbuti) were computed to evaluate the genetic affinities between the Ewenki and ancient Northeast Asian groups; (D) Admixture-f3 analyses of the form f3 (Ancient Northeast Asian 1, Ancient Northeast Asian 2; Ewenki) identify significant mixture events; notably negative values (Z < -3) suggest that the Ewenki can be modeled as a blend of two ancestral sources (source 1 and source 2), which are highlighted with an asterisk (*) to denote statistically significant admixture.

25.3% from the southern source. When the later Neolithic Wadian_LN was used as the southern source (paired with Russia_UstIda_EBA as the northern source), the southern contribution ranged from 67.3% to 71.6%, with the remainder attributable to the northern source. Together, these results suggest that the Ewenki primarily originated from interactions between Ancient Northeast Asian/Siberian populations and Yellow River Basin farmer-related ancestries. The detailed qpWave results, including p-values and ranks, are provided [Supplementary Table 12].

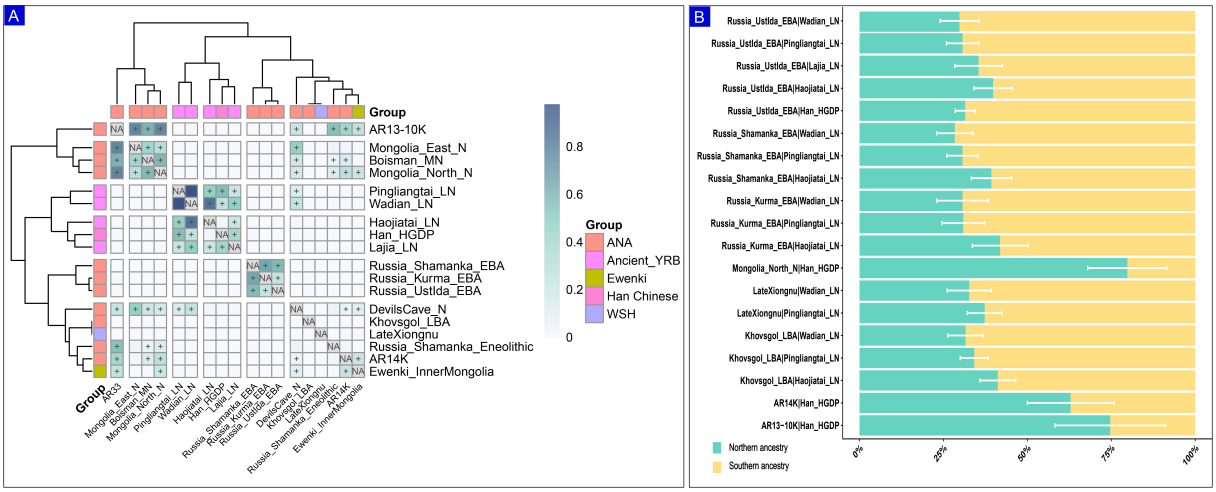

Figure 5. Population relationships and genetic affinity. (A) Pairwise qpWave tests highlight both genetic differentiation and shared ancestry among ancient populations. Rank-0 *P*-values greater than 0.05 indicate that two reference groups are genetically homogeneous ("++"), while p-values exceeding 0.01 are marked with "+"; (B) The qpAdm-inferred ancestry proportions for the Ewenki are presented. Two-way models support contributions from both northern and southern ancestry, with white error bars representing the standard error of each estimate. YRB: Yellow River Basin; ANA: Ancient Northeast Asian; WSH: Western Steppe Herders.

DISCUSSION

Genome-wide SNP genotyping and whole-genome sequencing have emerged as powerful complementary approaches for reconstructing population history and genetic structure[36-38], especially in the underrepresented American[39,40], Oceanian[41], African[42], and some Asian populations[43]. In this study, we leveraged genome-wide SNP array genotyping to characterize the demographic history of the Ewenki, taking advantage of high-density SNP coverage and ready compatibility with publicly available reference panels[44-47]. Future genomic research that combines ancient DNA, high-density SNP data, and larger sample sizes will help refine this understanding[48-51]. The substantial Ancient Siberian hunter-gatherer ancestry detected in our ADMIXTURE analysis (~36.94%, Figure 2) is consistent with the Ewenki long-term occupation of the boreal forest zone of Northeast Asia, where their traditional reindeer-herding and forest-hunting subsistence strategy reflects an enduring biocultural continuity with these ancient northern lineages[52-56]. The substantial co-occurring Yellow River Basin farmer ancestry (~41.87%) suggests that this northern subsistence base was overlaid by later interactions with agriculturally based populations, shaping the Ewenki hybrid ecological-genetic identity[57-59]. Throughout their prolonged migration and dispersal, the Ewenki people maintained a high degree of genetic continuity with ancient Northeast Asian and Siberian populations, while continuously intermarrying and exchanging genes with neighboring groups, including the Mongols, Daur, Oroqen, and Han Chinese. This process gradually formed their multi-origin mixed genetic structure. From a population-genetics perspective, the Ewenki ethnic group retains deep ancestral signals from the northern branch of the Tungusic language family, while also incorporating genetic components from ancient agricultural populations of the Yellow River Basin and from nomadic groups of the steppe[60-63]. Their contemporary genome reflects a population history marked by gradual southward migration from the ancient forest zones of Northeast Asia to the Inner Mongolian Plateau, alongside continuous admixture and evolution within the multi-ethnic interaction framework of China's northern frontier regions[64-66].

To investigate the population structure and admixture history of the Ewenki ethnic group, we conducted PCA and Admixture analyses[67]. PCA revealed that the Ewenki cluster is closely related to Tungusic, Mongolian, and ancient Siberian populations, suggesting close genetic affinities [Figure 1B]. This phenomenon can primarily be attributed to the Ewenki's classification as a Tungusic group, their geographical proximity to Inner Mongolia, and subsequent interactions with local populations[68]. Admixture

analysis under a 6-source model showed the Ewenki's predominant ancestral components consist of 41.87% ancient YRB farmers and 36.94% Siberian hunter-gatherers [Figure 2]. During the Neolithic period, the development of agriculture in the Yellow River Basin exerted significant pressure for population expansion[69]. In response to these pressures, some farmers from the region migrated northward. Similarly, ancient Siberian populations in the Amur River Basin, who depended on hunting and fishing, gradually migrated westward as material resources became depleted[70,71]. When these two groups met in the north, large-scale gene flow occurred over time. As a result, the Ewenki people inherited ancestral components from both the ancient YRB farmers and ancient Siberia. Through refined analysis of population structure, the Ewenki ethnic group shows significant genetic homogeneity with the Oroqen, Hezhen, and Xibe ethnic groups in Northeast China [Figure 3]. This finding suggests that this part of the Tungusic population may have diverged from their early ancestors, migrated southwestward and developed genetic adaptations conducive to their survival in a specific environment. In the Amur River Basin, the Negidal, Ulchi, and Nanai populations demonstrated a need to aggregate more closely with the Ewenki group [Figure 3]. This phenomenon may be attributed to the more conservative nature of these groups, which have consequently retained a greater proportion of ancient Siberian ancestry components[72].

To further validate the genetic relationships between the Ewenki and other ethnic groups, we conducted f_3-statistics [Figure 4]. In the outgroup-f_3 analysis, compared with other modern populations, the Ewenki showed higher f_3 values with Negidal, Nanai, Ulchi, Oroqen, and Daur, indicating closer genetic relationships with these groups. This shows that the Ewenki belong to the Tungusic-speaking populations, which share common ancestral origins. Additionally, the higher f_3 value with Daur suggests a certain degree of genetic affinity between the Ewenki and the Mongolic-speaking populations. Compared with other ancient populations, the Ewenki showed higher f_3 values with Boisman_MN, Jinchankou_LN, and Miaozigou_MN, indicating closer genetic affinities with these groups[73]. This result reflects the Ewenki's ancestral components from both ancient Siberian and ancient Yellow River Basin populations. In admixture analyses of modern populations, significant Z-scores were observed between the Tungusic/Mongolic and Sino-Tibetan groups, suggesting that the Ewenki derive from gene flow between Altaic-related and Sino-Tibetan-related sources. For ancient populations, significant Z-scores between ancient Yellow River Basin farmers and ancient Siberians further confirm that the Ewenki derived from the admixture of these two ancestral populations. The results from the f_4-statistics further indicate that the gene flows of the Ewenki people primarily derive from an ancient northern ancestry component related to ancient Northeast Asia and ancient Siberia [Supplementary Tables 8-10]. Additionally, a small contribution from ancient YRB farmers has been superimposed. This finding is consistent with a complex population history shaped by interactions between northern Tungusic-Mongolic groups and more southerly farmer-related lineages[74]. To assess the genetic relationships between the Ewenki and other ancient and modern populations, we first applied qpWave to formally test pairwise homogeneity relative to a fixed set of basal outgroups. The results revealed that the Ewenki could be modeled as homogeneous with several ANA populations (e.g., AR13-10K, AR14K, DevilsCave_N) and Mongolia_North_N (rank-0 $P > 0.05$), indicating that these populations share a common set of ancestral source populations relative to the outgroups [Figure 5A]. To further quantify the specific ancestry proportions in the Ewenki genome, we then applied qpAdm modeling using paired northern and southern source populations [Figure 5B]. The ratios were 39.8% Russia_UstIda_EBA to 60.2% Haojiatai_LN, 79.8% Mongolia_North_N to 20.2% Han_HGDP, and 74.7% AR13-10K to 25.3% Han_HGDP, indicating that the Ewenki primarily derived from admixture between ancient Northeast Asians and ancient Yellow River Basin farmers. Therefore, our findings suggest that Ewenki's primary ancestry comes from ancient Northeast Asian groups and from early farmers of the Yellow River Basin. During the Neolithic, the expansion of agriculture and population growth in the Yellow River Basin likely prompted northward movement and interaction with local ancient North Asian groups. Later periods, characterized by frontier contact and conflict, facilitated intermarriage and cultural exchanges with Daur, Oroqen, Mongol, and Han

groups[75]. These processes created regional differences within the Ewenki in language, demographic history, and genomic makeup. Overall, the Ewenki represent a complex mixture shaped by deep Northeast Asian ancestry and Yellow River Basin farmer ancestry, followed by long-term interaction along historical border zones.

The genetic admixture patterns inferred in this study align well with multiple lines of cross-disciplinary evidence concerning Northeast Asian population dynamics[76]. The deep ancestral component shared between the Ewenki and ancient Siberian/Northeast Asian populations is consistent with archaeological evidence of continuous boreal forest hunter-gatherer occupation since the late Pleistocene, exemplified by sites such as Devil's Cave and the Boisman culture, paralleling the traditional Ewenki subsistence economy preserved into recent centuries[77]. The Ancient Northeast Asian-Yellow River Basin farmer admixture detected in our qpAdm models corresponds to the Neolithic-to-Bronze Age expansion (~6,000-4,000 BP) of millet agriculture from the Central Plains into the West Liao River and Mongolian Plateau regions-a process also reflected in the linguistic record, as the Tungusic homeland has been traced to the West Liao River basin. Subsequent historical episodes including the Xiongnu, Mongol, and Manchu confederations facilitated population movement and intermarriage between forest-dwelling Tungusic groups and steppe pastoralists, accounting for the genetic affinity observed between the Ewenki and Mongolic-speaking populations on the Hulunbuir grasslands[78].

This study enhances our understanding of human population history in Northeast Asia and highlights the Ewenki's unique genetic profile. Their distinctive combination of ANA and Ancient Yellow River Basin Farmer ancestry demonstrates how human diversity remains in the genome and can be uncovered through modern sequencing technologies. mtDNA data indicate that the Ewenki share haplogroups with northern Chinese populations, a pattern resembling that seen in Mongolian groups and pointing to close genetic ties among them[79]. Genetic affinity with the Daur minority has also been documented. Although direct identification of adaptive signals lies beyond the scope of this demographic study, the Ewenki prolonged residence in extreme northern environments characterized by long, cold winters and limited daylight likely imposed selective pressures on physiological traits such as energy metabolism and circadian regulation. Future selection scan analyses [e.g., integrated Haplotype Score(iHS), Cross-Population Extended Haplotype Homozygosity(XP-EHH), Population Branch Statistic(PBS)] building on the demographic framework established here will be required to formally characterize these adaptive signals. Further research is necessary to clarify the genetic basis of environmental adaptation, including possible parallels with high-latitude adaptation.

Limitations

This study has several limitations that must be acknowledged. First, the sample size of the target population is relatively small, which limits the statistical power and presents challenges in comprehensively addressing the genetic diversity within this ethnic minority. the geographical scope of the sampling within the Ewenki population is limited, as it includes only individuals from the Inner Mongolia Autonomous Region. While these data offer valuable insights into the genetic structure of this underrepresented group, they may not comprehensively capture the genetic diversity present among all Ewenki subgroups, particularly those residing in Heilongjiang and Siberia[80]. The Ewenki are geographically dispersed across three principal regions-the IMAR of China, the Greater Khingan Mountains of Heilongjiang Province, and the Siberian regions of the Russian Federation - and historically encompass several culturally and linguistically distinct branches, including the Solon, Tungus, and Yakut Ewenki. Previous uniparental and limited autosomal studies suggest that geographic separation and divergent contact histories may have shaped substructure within this ethnolinguistic group. Heilongjiang Ewenki populations, which have maintained closer contact with Oroqen, Hezhen, and Manchu groups, may carry relatively higher proportions of Tungusic and ancient

Northeast Asian ancestries with weaker Mongolic-related signals than the IMAR samples examined here. Siberian Evenki groups, particularly those in the Russian Far East and Eastern Siberia, are hypothesized to retain a larger fraction of Ancient Siberian hunter-gatherer ancestry with reduced Yellow River Basin farmer-related contributions[81]. The IMAR Ewenki may therefore occupy an intermediate genomic position, reflecting long-term coexistence with Mongolic-speaking populations on the Hulunbuir grasslands and demographic influences from agricultural expansion in the Central Plains. Accordingly, while our qualitative conclusion - that the Ewenki derive primarily from admixture between Ancient Northeast Asian/Siberian and Yellow River Basin ancestral sources - likely reflects a shared ancestral foundation across subgroups, the specific quantitative ancestry proportions and admixture timing may differ across regions. We therefore caution against direct extrapolation of our quantitative estimates to Heilongjiang or Siberian Ewenki populations without further validation. Future studies incorporating samples from these underrepresented subgroups, ideally combined with whole-genome sequencing and regional ancient DNA, will be essential for building a comprehensive pan-Ewenki demographic framework[82,83]. Second, this study primarily focuses on the population genetic history of the Ewenki ethnic group, with particular emphasis on analyzing population structure and ancestral relationships. However, the analysis regarding natural selection and medical aspects is relatively insufficient, as it fails to comprehensively integrate phenotypic data and disease information to explore their biological adaptability in greater depth. Consequently, we are unable to thoroughly evaluate the fine-scale demographic history or the functional and biomedical implications associated with Ewenki-specific variants. Finally, all of our genetic inferences are based on contemporary Ewenki individuals and external ancient genomes, as we do not have access to ancient DNA directly linked to ancestral Ewenki groups[84,85]. This limitation constrains the temporal resolution of our demographic reconstructions and necessitates that we infer past events indirectly. Specifically, the absence of direct Ewenki ancestors among the available ancient reference genomes constrains the precision of timing for the inferred admixture events. Future studies incorporating linkage-disequilibrium-based dating methods such as Distribution of Ancestry Tracts of Evolutionary Signals (DATES), in combination with regional ancient DNA, will be essential to pinpoint specific gene flow episodes and refine the demographic chronology of the Ewenki. Future studies should incorporate high-coverage whole-genome sequencing of broader Ewenki subgroups across Inner Mongolia, Heilongjiang, and Siberia; recovery of ancient DNA from regional archaeological contexts to anchor demographic timelines; and functional investigation of candidate adaptive variants related to cold-climate physiology, lipid metabolism, and circadian regulation, which would together construct a comprehensive pan-Ewenki framework integrating demographic history with biological adaptation[86].

CONCLUSIONS

The Ewenki are a genetically unique Tungusic-speaking group in Northeast Asia whose current ancestry reflects both deep regional roots and repeated contact episodes. By combining genome-wide SNP data from Ewenki individuals with modern and ancient Eurasian references, we demonstrate that the Ewenki primarily derive from Ancient Northeast Asian lineages, with additional ancestry related to early Yellow River Basin farmers. Formal tests (f_3, f_4), qpWave, and qpAdm analyses support a model where the Ewenki resulted from admixture between Ancient Siberian/Ancient Northeast Asian sources and agricultural populations from the Yellow River Basin, followed by long-term interactions with neighboring Tungusic and Mongolic groups. These findings refine the demographic history of the Ewenki, emphasize their strong genetic ties to other Tungusic-speaking populations, and show that small and understudied ethnolinguistic minorities hold crucial insights into population structure, migration, and adaptation in Northeast Asia.

DECLARATIONS

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Authors' contributions

Contributed equally to this work: Wei D, Zhang L

Conceived and designed the study: Liu C, Wang M, Hai X, He G
Developed the methodology: Mengge Wang, Guanglin He
Conducted analysis manuscript: Duan S, Feng Y, Chen J, Sun Q, Luo L

Availability of data and materials

The supplementary materials include all the information. The genome-wide SNP genotype data generated in this study, together with the corresponding processed VCF and PLINK-format files, have been deposited in the Genome Sequence Archive (GSA) for Human at the National Genomics Data Center, China National Center for Bioinformation. For further information and requests for resources, please contact the corresponding authors.

AI and AI-assisted tools statement

Not applicable.

Conflicts of interest

Wang M and He G are members of the Junior Editorial Board of the *Journal of Translational Genetics and Genomics*. They were not involved in any aspect of the editorial process for this manuscript, including reviewer selection, manuscript handling, and decision-making. The other authors declare that they have no conflicts of interest.

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Ethical approval and consent to participate

The present study strictly adhered to the ethical guidelines of the Declaration of Helsinki and was approved by the Medical Ethics Committees of West China Hospital, Sichuan University (2024-1463). All subjects provided informed consent before participating.

Consent for publication

Not applicable.

Supplementary Materials

Supplementary Materials

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