

Genetic Origins and Migration Patterns of Xinjiang Mongolian Group Revealed through Y-Chromosome Analysis

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Genetic Origins and Migration Patterns of Xinjiang Mongolian Group 1 **Revealed through Y-Chromosome Analysis** 2

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22 Abstract

The Xinjiang Mongolians, located along the Silk Road, migrated westward from Northeast Asia in 23 the 13th and 14th centuries. Despite its significance, genetic studies on Xinjiang Mongolians have 24 been limited compared to other Mongolian populations. In this study, we analyzed the non-25 recombining region of the Y-chromosome in 165 Xinjiang Mongolian males using 108 biallelic 26 27 markers and 44 STRs. Our study identified prevalent haplogroups C2a1a3-F1918 (10%), C2a1a2-M48 (8%), N1a1a-M178 (5%), and R1a1a-M17 (10%) in the Xinjiang Mongolians. Additionally, 28 our study suggested a genetic affinity between Xinjiang Mongolians and Inner Mongolia Mongolian 29 populations, as well as other ethnic groups from northwest China, based on the PCA analysis. The 30 Network analysis revealed distinct branching expansion patterns in haplogroups C2a1a3a-F3796, 31 C2a1a2-M48, and N1a1a-M178, with estimated timeframes aligning with Genghis Khan's invasion 32 33 of Xinjiang in the Yuan Dynasty. Notably, our analysis of the R1a1a-M17 Network highlighted the role of Xinjiang Mongolians in the expansion of Turkic-speaking populations in Xinjiang and 34

35 surrounding regions. The integration of ancient DNA data suggested that the high frequency

- haplogroups C2a1a3a-F3796, C2a1a2-M48, and N1a1a-M178 <u>could</u> be traced back to their <u>origin</u> in
- 37 Northeast Asia. Furthermore, the estimated TMRCA of haplogroup R1a1a-M17 <u>implied</u> cultural and
- 38 genetic influences from Turkic populations during the Qagatay Khanate period. Overall, our study
- 39 provided a genetic explanation for the ethnic origin of Xinjiang Mongolians, highlighting their
- 40 migration from Northeast Asia and subsequent assimilation with the local populations in Xinjiang.

41 **1** Introduction

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Xinjiang, located in northwest China, has long been a vital hub connecting ancient China to the rest 42 of the world through the Silk Road. This region is known for its diverse ethnic makeup, with various 43 ethnic groups migrating and settling there over the years (Tian et al., 2023). One such group is the 44 Mongolian, who has a widespread presence across northern China, Mongolia, Russia, Kazakhstan, 45 and other regions (Bai et al., 2018; Wang et al., 2021). It is estimated that there are around 10 million 46 Mongolians worldwide (Bai et al., 2018), with approximately 6 million residing in China, mainly in 47 Inner Mongolia. In Xinjiang, there are about 170,000 Mongolians, with the majority concentrated in 48 Bayingol, Boltala, and Hoboksar. 49

50 Historians believe that the Mongolians can trace their roots back to the Donghu nomadic tribes (Yi, 1979; Tu, 1984). Recent genome-wide analyses of 214 ancient individuals from the Mongolian 51 Plateau and Lake Baikal region have revealed that there are at least four ancestral origins of the 52 Mongolians. Two of them can be traced back to their origins in northeastern and northern Mongolia 53 before the Bronze Age, with these populations descending from hunter-gatherer communities in 54 northeast Asia and northern Eurasia. Another ancestral source is linked to the Afanasievo culture, 55 while the fourth source is a genetic mixture of Yamnaya pastoralists and European farmers (Jeong et 56 al., 2020; Wang et al., 2021). A study using mitochondrial DNA has shown that Chinese Mongolians 57 primarily carry East Asian-specific haplogroups, with a decreasing distribution from east to the west. 58 Interestingly, some European-prevalent haplogroups have also been detected, showing an opposite 59 distribution pattern to the East Asian-specific haplogroups (Cheng et al., 2008). Furthermore, various 60 studies on Y-chromosomal haplogroups have identified C2*-M217 as the foundational paternal 61 lineage among Mongolic-speaking populations. Other haplogroups such as O2*-M122, O1b*-62 M268/P31, N*-M231, and R-M207 have also been found in certain proportions (Wei et al., 2018; 63 Zhang et al., 2018; Huang et al., 2018; Wang et al., 2021). Researchers have concluded that 64 haplogroup C2*-M217 is one of the founding paternal lineages of all Mongolic-speaking populations 65 (Wei et al., 2018). Additionally, studies have traced the origins of specific Y-chromosome 66 haplogroups, such as C3*-F3918 (updated to C2a1a1-F3918), back to ancient nomadic groups like 67 68 the Donghu (Zhang et al., 2018). Detailed phylogenetic and phylogeographic analyses have suggested the northeastern Asian origin of haplogroup C2c1a1a1-M407 among Mongolic-speaking 69 populations (Huang et al., 2018). Wang et al. have developed high-resolution Y-SNP SNaPshot 70 panels, identifying three main ancestral sources of dominant Mongolian haplogroups: local lineage 71 C2*-M217, incoming lineages from southern East Asia (O2*-M122, O1b*-P31, and N1*-CTS3750), 72 and western Eurasia (R1*-M173) (Wang et al., 2021). 73

However, existing Y-chromosome studies of Xinjiang Mongolians primarily focused on comparing genetic variations and forensic applications across groups, leaving a gap in understanding the detailed haplogroup distribution and paternal genetic makeup of this population. For example, one study has examined the genetic diversity of Y-STR and indel markers in Xinjiang Mongolians and found their closest genetic relationship with the Gansu Dongxiang population, followed by Hulunbuir Mongols, the Daur ethnic group from Inner Mongolia, and three other ethnic minorities in Xinjiang (Liu et al.,

- 80 2020). Another study <u>has</u> found that the genetic distance between the Mongolian ethnic group and the
- Uyghur and Xibe populations in Xinjiang is relatively small compared to populations from other continents (Wei et al. 2019)
- 82 continents (Wei et al., 2019).

Previous studies have shown that different geographic divisions and ethnic groups within the
 Mongolian populations have distinct genetic backgrounds (Bai et al., 2018; Zhao et al., 2020; Wang
 et al., 2021; He et al., 2023). However, research on the Y-chromosome analysis of Xinjiang
 Mongolians is lacking. In this study, our aim is to explore the paternal genetic structure of the
 Mongolian ethnic group in Xinjiang. We will analyze 108 Y-SNPs and 44 Y-STRs from 165
 Mongolian males from Xinjiang to gain insights into the origin of Xinjiang Mongolians and their
 interactions with other local Xinjiang populations.

90 **2** Materials and methods

91 2.1 <u>Sample preparation</u>

With the informed consent from the Ethics Committee of Fudan University of Life Sciences, 165 92 bloodstain samples were collected from unrelated and healthy Mongolian males from Xinjiang. 93 Genomic DNA was extracted through the QIAamp DNA Blood Mini Kit (QIAGEN, Germany). In 94 all DNA samples, 108 Y-SNP markers were hierarchically genotyped by SNaPshot (ABI SNaPshot® 95 Multiplex Kit) as described in previous studies (Wang et al., 2014; Wen et al., 2020). The genotyping 96 of the Xinjiang Mongolian samples was conducted utilizing the GeneAmp PCR system 9,700 97 (Thermo Fisher Scientific) comprising a set of 44 microsatellite markers (Zhang, 2015; Zhang et al., 98 99 2018). The data was provided to the YHRD (Y-chromosomal haplotype reference database) and assigned the accession number YA004579 (Xinjiang, China, [Mongolian]). 100

101 2.2 <u>Y-SNP data analysis</u>

Haplogroups were categorized in accordance with the ISOGG Y-DNA Haplogroup Tree 2019. We chose 167 modern Eurasian populations, with a particular focus on Central Asian/Altaic-speaking populations, for population comparisons with a consistent level of haplogroup resolution . Meanwhile, the haplogroup frequencies were determined through the direct counting method. Additionally, a principal component analysis (PCA) was conducted based on Y-SNPs using RStudio software version 2021.09.2.

108 2.3 Y-SNP-STR data analysis

Median-joining networks were computed using the NETWORK version 10.2.0.0 program and 15 Y-109 STRs data for the common haplogroups. The Time to Most Recent Common Ancestor (TMRCA) for 110 the identified common haplogroups in both clusters was calculated using the average squared 111 distance (ASD) estimator (Sengupta et al., 2006), with a generation time set at 25 years (Wang et al., 112 2014). The genealogical mutation rate OMRS in ASD method for population variation was used for 113 the age estimates (Wang and Li, 2015). In order to ascertain TMRCA for representative genetic 114 lineages, recent expansions within the specific population were identified. Subsequently, we 115 examined samples within a network diagram, specifically those within a range of five mutation steps, 116 to investigate recent historical events within the Xinjiang Mongolians (Luo et al., 2020). 117

- 118 <u>3</u> Results
- 119 3.1 <u>Simplified phylogenetic tree of Y-chromosomal haplogroups</u>

According to the ISOGG Y-DNA haplogroup tree 2019-2020 and Y-SNP frequencies, we conducted 120 a comparative analysis of haplogroup distribution among various populations in Xinjiang, including 121 Mongolian (this study) (Supplementary Table 1), Xibe (unpublished), Hui (unpublished), Kazakh 122 (previously reported) (Wang et al., 2023), Kyrgyz (previously reported) (Wen et al., 2022), as well as 123 Hulunbuir, Hohhot, and Ordos Mongolian populations (previously reported) (Wang et al., 2021). To 124 visually represent the distribution of haplogroups across these populations, we constructed a 125 simplified phylogenetic tree (Figure 1). The figure showed that the most common haplogroups in 126 Xinjiang Mongolians were C2a1a3-F1918 (10%), R1a1a-M17 (10%), C2a1a2-M48 (8%), and 127 N1a1a-M178 (5%). 128

- The C2a1a3-F1918 haplogroup emerged as the predominant genetic lineage among the Xinjiang 129 Mongolians. It was the upstream haplogroup of C2a1a3a-F3796 and had the highest prevalence 130 among diverse groups, such as Xinjiang Kazakhs (50%), Hulunbuir Mongolians (18%), Xinjiang 131 132 Xibes (14%), Ordos Mongolians (14%), and Hohhot Mongolians (12%). C2a1a3-F1918 was widely recognized as one of the main ancestral paternal lineages among Mongolic-speaking communities 133 and spread rapidly across the Eurasian steppe during the Mongol conquests (Wei et al., 2018). 134 Therefore, the genetic impact of the Mongol Western March on the local population in Xinjiang may 135
- 136 be clearly evident.

C2a1a2-M48 was the third most prevalent haplogroup among Xinjiang Mongolians, with significant 137

distribution patterns among Xinjiang Kazakhs (10%), Hohhot Mongolians (9%), and Hulunbuir 138

Mongolians (7%). C2a1a2-M48 was the predominant paternal lineage among Tungusic-speaking 139

populations (Liu et al., 2021). 140

It was worth noting that R1a1a-M17 (10%) and its downstream subhaplogroups, namely Rlalalblala-141

M458 (2%) and Rlalalb2-Z93 (1%), were highly prevalent among the Xinjiang Mongolians and were 142 also widely distributed among other ethnic groups in Xinjiang. Specifically, these lineages were 143 common among Xinjiang Kyrgyz (48%), Xinjiang Hui (17%), Xinjiang Xibe (8%), and Xinjiang 144 Kazakh (7%). However, they were less prevalent in the Mongolian populations of Inner Mongolia, 145 with frequencies of 5% in Ordos, 3% in Hohhot, and 1% in Hulunbuir. R1a1a-M17 was widely 146 distributed among Indo-European-speaking populations (Koryakova and Epimakhov, 2014). 147

148 The N1ala-M178 haplogroup emerged as the fourth most prevalent lineage among Xinjiang

Mongolians, with significant distribution among the Hulunbuir (11%) and Ordos (7%) Mongolian 149 populations. Additionally, this haplogroup had a limited presence in both the Xinjiang Hui (4%) and 150

Xinjiang Xibe (3%) populations. 151

3.2 152 PCA analysis of Y-SNP haplotypes

At the population level, we compared the Y-SNP data of Xinjiang Mongolians with data from 167 153

modern Eurasian populations, including Mongolic, Tungusic, Turkic, Indo-European, Uralic, Sino-154

Tibetan speaking groups, and others. We conducted a principal component analysis (PCA) on these 155

datasets to analyze the genetic relationships (Figure 2, Supplementary Table 2). The linguistic 156 clusters were broadly distributed in the figure, with Xinjiang Mongolians falling within the 157

overlapping confidence intervals of Han Chinese and Mongolian populations, indicating a close 158

genetic affinity with both groups. Additionally, Xinjiang Mongolians showed significant genetic 159

relationships with populations such as Xinjiang Hui, Xinjiang Xibe, Xinjiang Kazakh, Kyrgyzstan-160

Northwest Kyrgyz, Hohhot Mongolian, Hulunbuir Mongolian, Ordos Mongolian, and others. 161

Network analysis of Y-SNP-STR haplotypes 162 3.3

163 At the individual level, we conducted an analysis and investigation of four primary lineages of

164 Xinjiang Mongolians using reduced median networks, namely C2a1a3a-F3796, R1a1a-M17, C2a1a2-

165 M48, and N1a1a-M178. Furthermore, we categorized and cross-referenced populations based on

166 their respective language families.

167 The network of haplogroup C2a1a3a-F3796 was established using 15 Y-STR profiles from 262 individuals across 16 diverse populations (Figure 3, Supplementary Table 3). The network of 168 haplogroup C2a1a3a-F3796 displayed a characteristic branching expansion pattern, featuring a 169 central founder haplotype and multiple sub-founders. The central haplotypes depicted in the image 170 predominantly consist of Kazakh populations from northwest China and Central and North Asia, 171 Xinjiang Kyrgyz populations (blue), as well as Mongolians originating from Inner and Outer 172 173 Mongolia (purple). Xinjiang Mongolians (red) tended to exhibit clustering patterns with other Mongolic-speaking populations in northern China, as well as with various ethnic groups in Xinjiang, 174 175 including Kyrgyz, Xibe, and Uygur populations. These clusters represented simultaneous small-scale expansions. By employing the ASD method, we estimated that the samples within the dashed ellipse 176 had a Time to Most Recent Common Ancestor (TMRCA) of approximately 729.76 years Before 177 Present (BP), with a standard deviation of \pm 653.89 years, while excluding steps greater than five 178 179 mutations.

180 Haplogroup C2a1a2-M48 exhibited a predominant distribution in Central and northern Asia, particularly among Tungusic-speaking and Paleo-Siberian-speaking populations. Additionally, it 181 demonstrated a significant presence in Xinjiang Mongolians. The Xinjiang Mongolians distributed in 182 the upper part of the figure (Figure 3, Supplementary Table 4), forming a star-shaped expansion 183 pattern along with the Inner Mongolia Mongolian populations (purple), Xinjiang Kazakh populations 184 (blue), and Xinjiang Uygur populations (blue), presenting an aesthetically pleasing distribution. 185 186 Excluding steps greater than five mutations, we estimated TMRCA of the most recent cluster in the right clade (samples in the dashed line), yielding a result of 765.43 ± 545.09 years ago. 187

Haplogroup N1a1a-M178 was frequently observed in populations of northern European and northern
East Asian descent. In the genetic network analysis (Figure 3, Supplementary Table 5), Xinjiang
Mongolian samples exhibited proximity to other Mongolic-speaking populations (purple), while also
displaying associations with a limited number of Turkic-speaking populations (blue). The upper left
corner of the network revealed the involvement of Xinjiang Mongolians in the expansion of
Mongolians from Inner Mongolia. Excluding steps greater than five mutations, we estimated
TMRCA of this cluster (samples in the dashed line), yielding a result of 769.85 ± 457.38 years ago.

195Haplogroup R1a1a-M17 was also a major haplogroup among Xinjiang Mongolians. The distribution196of Xinjiang Mongolians in the network appeared to be dispersed (Figure 4, Supplementary Table 6),197suggesting a diverse ancestral background among those belonging to the R1a1a-M17. Xinjiang198Mongolians participated in the expansion of Turkic-speaking populations in Xinjiang (cyan) as well199as in other regions (dark blue). Excluding steps greater than five mutations, we estimated TMRCA of200the most recent cluster in the right clade (samples in the dashed line), yielding a result of 552.1 \pm 201345.13 years ago.

202 **4 Discussion**

Haplogroup C2a1a3a-F3796 is renowned for being one of the ancestral paternal lineage of Mongolicspeaking populations. (Wei et al., 2018). <u>Further research has revealed that this cluster is</u> linked to the ancient Mongolian tribe Nirun (Wei et al., 2018; Zhabagin et al., 2020). Ancient DNA evidence 206 <u>has shown</u> that it <u>can</u> be found in Dornod, Mongolia during the Late Medieval period (Jeong et al.,

207 2020). Wei and Li have shown that the estimated time to the most recent common ancestor (TMRCA)

of C2a1a3a-F3796 is approximately 1000 ± 300 years old, with its origin in eastern Mongolia and the northern Greater Khingan Mountains before spreading westward (Wei and Li, 2022). <u>Previous</u>

- research has also uncovered a direct correlation between the expansion of the Mongolian empire after
- the 13th century and the dissemination of this paternal lineage (Zerjal et al., 2003). We estimated the
- 212 TMRCA of approximately 729.76 ± 653.89 , which could potentially provide further evidence to
- 213 support this speculation. After the Mongolian expansion, it seemed that the introduction of
- 214 Haplogroup C2a1a3a-F3796 into Xinjiang was observed in this group of Mongolian individuals.

Haplogroup C2a1a2-M48 represents the predominant paternal lineage in Tungusic-speaking 215 216 populations (Lell et al., 2002; Pakendorf et al., 2006, 2007), which is one of the largest demographic groups in Siberia (Liu et al., 2021). C2a1a2-M48 is widely distributed throughout Central and 217 218 Northern Asia, with higher frequencies among populations speaking Mongolic and Tungusic languages (Katoh et al., 2005; Malyarchuk et al., 2010; Balanovska et al., 2018; Liu et al., 2021). 219 However, the presence of C2a1a2-M48 in these populations does not indicate a common origin but 220 rather reflect an admixture event during the recent historical period (Liu et al., 2021). Historically, 221 222 Mongolic-speaking and Tungusic-speaking populations in the outer Baikal region were closely associated, suggesting frequent genetic exchange between them (Baodungude, 1988). This 223 224 haplogroup diverged approximately 15,000 years ago and underwent sub-branching around 4,000 years ago (Karmin et al., 2015). Its origin was conventionally associated with the inner regions of 225 226 Central and Northern Asia (Karafet et al., 2002). Previous studies of ancient DNA have shown that haplogroup C2a1a2-M48 first appears in the Songnen Plain of China before the Last Glacial 227 228 Maximum (about 20,000 years before present) and can also be found in the Songnen Plain and Russian Far East during the Neolithic Age (about 10,000 years before present) (Wang et al., 2014; 229 Mao et al., 2021, Wang et al., 2023). Since then, this haplogroup was found among Mongolians from 230 the Iron Age onwards (about 3,400 years before present) (Jeong et al., 2020). We undertook 231 calculations that suggested the TMRCA of haplogroup C2a1a2-M48, thereby implying a possible 232 link between the Xinjiang Mongolians and Genghis Khan's expedition. The spatial and temporal 233 234 distribution of haplogroup C2a1a2-M48 closely corresponded to that of haplogroup C2a1a3a-F3796. The high prevalence of haplogroup C2a1a3a-F3796 and C2a1a2-M48 in the Xinjiang Mongolians 235 might suggest a Northeast Asian origin. 236

- Haplogroup N1a1a-M178 is commonly found in ancient Siberian archaeological sites as well as 237 among present-day populations of Altaic, Uralic, Russian, and Chinese descent in the Siberian region 238 (Rootsi et al., 2007; Hu et al., 2015). Research by Rootsi et al. has suggested that the haplogroup 239 N1c-M46 lineage, now updated to N1a1-M46, likely originates in China and experienced population 240 bottlenecks in northern East Asia before spreading into Siberia (Rootsi et al., 2007). Ancient DNA 241 evidence have indicated that haplogroup N1a1a-M178 emerges around 7,500 years ago in Jilin, 242 China (Ning et al., 2020) and appeared in Siberia during the Bronze Age, approximately 5,000 years 243 ago (Hollard et al., 2018). The emergence of Haplogroup N1a1a-M178 in Mongolians could be 244 traced back to about 2,300 years ago (Keyser-Tracqui et al., 2004). Therefore, the prevalence of 245
- 246 <u>N1a1a-M178 in Xinjiang Mongolians might also suggest a Northeast Asian origin.</u>
- 247 Haplogroup R1a1a-M17 is one of the most common haplogroups in Xinjiang Mongolians. It is more
- 248 prevalent in Eastern Europe, South Asia, and Siberia, while less frequently observed among
- 249 individuals of Middle Eastern and East Asian descent (Balanovsky et al., 2008; Koryakova and
- 250 Epimakhov, 2014). Ancient DNA studies has consistently shown a correlation between the spread of
- 251 the paternal lineage R1a1a-M17 and the migration of Indo-European populations, estimating that this

- 252 lineage originated around 8,000 years ago during the early Holocene period (Semino et al., 2000; 253 Keyser et al., 2009; Allentoft et al., 2015; Haak et al., 2015; Damgaard et al., 2018) Haplogroup R1a1a-M17 has been identified in the Lake Baikal region of Siberia over 7,000 years ago (Moussa et 254 al., 2018). Moreover, R1a1a-M17 has also been detected in the Tarim Basin of Xinjiang during the 255 Bronze Age to the Iron Age (between approximately 6,000 and 3,000 years before present), 256 exclusively observed in male individuals from the West Eurasia, which is related to the migration of 257 258 ancient people from Europe in the early Bronze Age (at least 4,000 years before present) (Li et al., 2010). Given the widespread distribution of haplogroup R1a1a-M17 across different regions and time 259 periods, we hypothesized that individuals in the Xinjiang Mongolian population might have genetic 260 connections to Siberia or West Eurasia. Our network and computational TMRCA analyses hinted at 261 the possibility that Xinjiang Mongolians contributed to the population expansion of other groups, 262 such as the Xinjiang Kyrgyz, following their westward migration. These might be attributed to the 263 process of Turkic cultural assimilation among the Mongolians during the Chagatai Khanate period 264 (Liu, 1994). 265 Haplogroup O-M175 is a significant component of the Y-chromosome gene pool in East Asians, 266 making up approximately 75% of Chinese paternal lineages (Yan et al., 2011). This haplogroup is 267 268 also prevalent in Southeast and East Asian populations (Poznik et al., 2016). Wei et al. have found that among Mongolic-speaking populations, the frequency of Haplogroup O*-M175 is 18.929% (Wei 269 270 and Li, 2022). In our study, we observed a high frequency of the subhaplogroup O*-M175 in Xinjiang Mongolians, with O2*-M122 and O1b*-M268 being the most common subgroups,
- Xinjiang Mongolians, with O2*-M122 and O1b*-M268 being the most common subgroups,
 accounting for 17% and 8% respectively. Previous research on haplogroups O1-M119 (now updated to O1a-M119), O2-M95 (now updated to O1b1a1a-M95), and O3-M122 (now updated to O2-M122)
 has suggested that northern populations originate from the south and migrated northward after the
 initial Paleolithic settlement of East Asia around 3 million years ago (Bing et al., 2000; Wen et al., 2004; Shi et al., 2005; Wang et al., 2013). These might explain the widespread presence of O2*M122 and O1b*-M268 among Xinjiang Mongolians.

278 By analyzing Y-chromosome data of the Xinjiang Mongolians and considering historical contextual information, we can make more robust inferences about their migratory history. In the 13th century, 279 the Mongol Empire, a nomadic nation thriving on the Mongolian Plateau, founded an extensive land 280 empire that reached into Central Asia, including present-day Xinjiang (Howorth, 1888; Man, 2014). 281 The influence of this historical legacy can still be seen in the diverse cultural and ethnic landscape of 282 283 modern Xinjiang. Genghis Khan's conquests began in the early 1200s, and by the time of his death in 284 1227, he had already laid the foundations for a vast empire. His successors, particularly Kublai Khan and Hulagu Khan, continued to expand the empire. During the westward expansion, the Mongols 285 conquered Central Asia, including the Khwarazmian Empire, Persia, Anatolia, and parts of the 286 Caucasus (Weng, 1985; Han, 1986). They also underwent significant cultural assimilation through 287 interactions with societies in Central Asia. Central Asia is linguistically diverse, with multiple 288 289 language families present within its borders (Dalizabu, 2006). As a result, a discernible genetic component from other language families can be observed among Xinjiang Mongolians. 290

291 **5** Conclusion

Based on the combined <u>analyses</u> of ancient DNA studies focusing on haplogroup<u>s</u> C2a1a3a-F3796, C2a1a2-M48, and N1a1a-M178, as well as the calculated TMRCA of the haplogroups, it <u>could</u> be concluded that the Xinjiang Mongolian<u>s</u> had <u>the</u> origin in Northeast Asia. Additionally, the analysis of haplogroup R1a1a-M17 <u>provided</u> further evidence of their integration with neighboring populations since their migration from Northeast Asia to Xinjiang.

There was a significant genetic similarity between the Xinjiang Mongolians and present-day 297 Mongolians in Inner Mongolia, as well as genetic interactions with indigenous populations in 298 Xinjiang such as Uygur, Kyrgyz, Xibe, and Kazakh. However, caution should be exercised when 299 estimating TMRCA due to inherent errors in estimation. Overall, our study provided genetic 300 evidence supporting the complex paternal genetic composition of Xinjiang Mongolians through 301 admixture. To further enhance our understanding of the genetic structure and historical background 302 303 of Xinjiang Mongolians, it is imperative to conduct further investigations using diverse genetic markers and incorporating genetic evidence from other geographically distinct Mongolian 304 populations. 305

306 6 Conflict of Interest

307 <u>The authors declared that the research was conducted</u> in the absence of any commercial or financial 308 relationships that could be construed as a potential conflict of interest.

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489 Captions

Figure 1 Frequencies of Y-chromosomal haplogroups and geographic locations of Xinjiang Mongolians and other reference modern populations. The figure depicts the geographical locations of the Xinjiang Mongolians and the reference population, while illustrating the investigation of Ychromosome haplogroups in this study along with their corresponding frequencies. The branches indicate the names of the haplogroups, while the marker names are presented according to ISOGG Y-DNA Haplogroup Tree 2019.

496 | Figure 2 Genetic relationships of Xinjiang Mongolians with other reference modern populations
497 from the principal component analysis (PCA) based on Y-chromosome haplogroups. Supplementary
498 Table 2 provides comprehensive information regarding the populations included in this study.

499 Figure 3 A reduced median joining network was constructed for haplogroup C2a1a3a-F3796, R1a1a-

500 M17, and C2a1a2-M48. Haplotypes are visually represented by circles, whose sizes are proportionate

501 to the number of individuals they represent. The connecting lines between the circles indicate

502 mutational distance, with the shortest line representing a single mutational step.

Figure 4 A Reduced Median joining network of haplogroup R1a1a-M17. Haplotypes are visually depicted as circles, where the sizes of the circles correspond to the number of individuals they represent. The lines connecting these circles indicate mutational distance, with the briefest line denoting a single mutational step.

Figure 01.TIF



D1a1a1 D1a1b

C2ala C2bl

Nlala Nla2b Nlb Olala Olb Olblala O2 O2alalalal O2alb

> O2a2a O2a2 O2a2l O2a2b

> > Q1;

R lala R lalalblala R lalalb2 R lb R lblalb R lblalblala2 R2a

	Xinjiang					Inner Mongolia		
	N=120	N=223	N=209	N=298	N=165	N=240	N=226	N=213
	Xinjiang	Xinjiang	Xiniiang	Xiniiang	Xinjiang	Hohhot	Hulunbuir	Ordos
Y SNP	Xibe	Hui	Kazakh	Kyrgyz	Mongolian	Mongolian	Mongolian	Mongolian
M145	0.00	0.00	0.00	0.00	0.00	0.02	0.00	0.01
M174	0.02	0.00	0.00	0.03	0.00	0.00	0.00	0.00
M15	0.00	0.00	0.00	0.00	0.01	0.00	0.00	0.00
N1	0.00	0.04	0.00	0.00	0.02	0.02	0.00	0.01
P47	0.00	0.03	0.05	0.00	0.02	0.03	0.03	0.03
M96	0.00	0.01	0.00	0.01	0.00	0.00	0.00	0.00
M130	0.00	0.00	0.00	0.01	0.02	0.00	0.00	0.00
F1396	0.00	0.00	0.03	0.26	0.01	0.03	0.03	0.05
M48	0.00	0.00	0.10	0.00	0.08	0.09	0.07	0.03
F1918	0.14	0.00	0.50	0.00	0.10	0.12	0.18	0.14
F2613	0.01	0.04	0.00	0.00	0.04	0.03	0.04	0.07
M407	0.00	0.00	0.01	0.00	0.04	0.05	0.08	0.04
M89	0.00	0.00	0.00	0.00	0.04	0.00	0.00	0.00
M201	0.00	0.05	0.01	0.01	0.00	0.00	0.00	0.02
M258	0.00	0.01	0.01	0.00	0.01	0.00	0.02	0.00
M304	0.01	0.13	0.02	0.06	0.02	0.02	0.01	0.03
M20	0.00	0.00	0.00	0.01	0.00	0.00	0.00	0.00
M272	0.01	0.00	0.00	0.00	0.01	0.00	0.00	0.00
M231	0.06	0.00	0.01	0.00	0.01	0.04	0.04	0.02
M178	0.03	0.04	0.02	0.01	0.05	0.02	0.11	0.07
P43	0.00	0.03	0.03	0.02	0.01	0.00	0.00	0.00
F2930	0.02	0.04	0.00	0.00	0.01	0.02	0.02	0.01
M1354	0.00	0.00	0.00	0.01	0.00	0.00	0.00	0.00
P203	0.05	0.01	0.00	0.00	0.01	0.01	0.00	0.00
M268	0.04	0.02	0.00	0.00	0.04	0.05	0.04	0.04
M95	0.01	0.04	0.00	0.00	0.04	0.00	0.02	0.00
M122	0.08	0.01	0.00	0.00	0.00	0.00	0.01	0.01
M121	0.01	0.00	0.00	0.00	0.01	0.00	0.00	0.00
IMS-JST002611	0.02	0.01	0.00	0.01	0.02	0.05	0.03	0.03
F11		0.06	0.00	0.00	0.04	0.10	0.10	0.06
- P201	0.00	0.00	0.00	0.00	0.01	0.02	0.00	0.02
- M/	0.01	0.00	0.00	0.00	0.01	0.00	0.00	0.00
P104	0.01	0.00	0.00	0.00	0.01	0.00	0.00	0.01
M117	0.13	0.03	0.01	0.00	0.01	0.00	0.00	0.00
F444	0.13	0.07	0.00	0.00	0.04	0.08	0.04	0.07
M242	0.03	0.00	0.05	0.00	0.02	0.03	0.00	0.03
M25	0.03	0.03	0.01	0.00	0.00	0.03	0.00	0.00
M346	0.00	0.00	0.00	0.00	0.01	0.00	0.01	0.00
M173	0.00	0.01	0.02	0.00	0.04	0.00	0.00	0.01
M17	0.00	0.02	0.06	0.00	0.00	0.00	0.00	0.00
M458	0.00	0.02	0.00	0.00	0.02	0.00	0.00	0.00
Z93	0.08	0.01	0.00	0.00	0.02	0.03	0.00	0.05
M343	0.00	0.01	0.00	0.05	0.02	0.04	0.00	0.03
M269	0.00	0.00	0.00	0.00	0.02	0.00	0.00	0.00
P312	0.01	0.04	0.00	0.00	0.01	0.00	0.00	0.00
M124	0.00	0.02	0.00	0.01	0.01	0.00	0.02	0.00
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Figure 04.JPEG

