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RESEARCH ARTICLE

Msaken, Tunisia: A Common Paternal Ancestor Confirmed by Y Chromosome DNA Analysis

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Abstract

Msaken City (Tunisia) is believed to have been founded around 1360 AD by five related men who migrated from West Asia. The population would have grown with the descendants of these founders and with the arrival of other populations from different regions of Tunisia. In order to elucidate the TMRCA of the founder population and to reveal their geographic origin, 23 males from different families of Msaken were examined, using the services of commercial companies, for 12 to 440 Y chromosome Short Tandem Repeats (STR) and Single Nucleotide Polymorphisms (SNP) markers using NG testing technology. Eight samples were genotyped for SNPs to determine their Haplogroups. In order to refine the phylogeny, traditional Sanger testing was performed on one sample for 300,000 bp in Y Chr (in Walk Through the Y chromosome project). Seven samples were also tested using Next Generation Testing (BigY) covering 20 million bp of Y chromosome overlapping 85% of Gold standard region (chromosome Y positions placed on the phylogenetic tree by the YCC) using NGS instruments, HiSeq 2000 and 2500. A comparison of STR results with data from different sources and databases was made, using SQL scripts and data mining tools, to find matching haplotypes. All the STR results were found to have no more than three mismatches per 12 markers and not more than six mismatches per 67 markers and the SNP results showed that all tested samples belonged to Haplogroup J-M172 inside its subgroup J-L24. Relying on the common STR marker values, we define a Msaken-Haplotype. NG tests for our samples as well as those added to the yfull.com tree allowed us to refine the phylogeny of J-L24 and the samples were all found to belong to J-L271 Haplogroup and share 54 exclusive SNPs. The calculated Time to Most Recent Common Ancestor (TMRCA) based on NG testing, ranges between1500 and 6200 YBP showing a strong bottleneck around 5400 YBP. The variation of the collected results shows a geographic root of J-L192 in East Anatolia, present day Armenia, Azerbaijan, and West Iran. 20 to 30% of random Tunisian STR haplotypes belonging to J-M172 (J2) Haplogroup exhibit the Msaken-Haplotype.

Key Words: *Msaken DNA; Tunisia DNA; Salar DNA; China DNA; Haplogroup J2*

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1. Introduction

Y-chromosome DNA studies play a pivotal role in advancing human genomics research. This is because the Y chromosome is a unique genetic marker in that approximately 95% of its sequence does not undergo recombination with its homologue (the X chromosome) during meiosis. This characteristic makes it an invaluable tool for investigating human male-to-male genealogy by studying the mutations accumulated in its non-recombining region.

Recent advancements in third-generation genetic testing technologies have revolutionized our understanding of human migrations and the relatedness between different human populations. These tools have enabled researchers to delve deeper into the intricate tapestry of human history, uncovering hidden stories encoded within our genetic material.

In this paper, our primary objective is to investigate the deep paternal ancestry of the population in Msaken, a city located in the Sahel region of Tunisia, specifically in the East Central part. Msaken has a rich history and is believed to have ancestral ties to the Middle East. Our study aims to shed light on this historical connection by exploring the genetic variations within the Y chromosome of the contemporary Msaken population.

As of 2014, Msaken was home to approximately 70 thousand people. In the intervening years, the city has experienced population growth, and our estimates for 2023 suggest that it now houses around 100 thousand residents. Interestingly, a significant portion of individuals with roots in Msaken have migrated to Europe, particularly the French Riviera, with an estimated 20 thousand Msaken-origin individuals residing in this region [1].

Local historical accounts and oral traditions [2] link the foundation of Msaken to five related men—four brothers and their nephew—who are said to have migrated from the Arabian Peninsula. According to this tradition, their descendants initially settled in Kairouan, and after several generations, they embarked on an eastward journey, ultimately founding Msaken. This founding history is further marked by the construction of five Qasrs, known as 'Masakin al Ashraf,' symbolizing the houses of noble people (see Figure 1).

Figure 1: *Map of Tunisia showing Msaken location.*

Furthermore, historical records from Ganiage in 1860 [3] indicate that Msaken's population was approximately nine to ten thousand in that era. This historical context positions Msaken as the fourth-largest city in Tunisia during that time, trailing only Tunis, Sfax, and Kairouan and preceding Sousse.

In the subsequent sections of this paper, we will have delved into our research methods, presented our findings (most of Msaken population share a common paternal ancestor who lived 1500 years ago), and discussed the implications of our study in the broader context of human migrations and population genetics in the Sahel region of Tunisia. Our results will provide valuable insights into the deep paternal ancestry of the Msaken population and its historical ties to the Middle East.

2. Material and Methods

2.1. Population studied

A total of 23 samples were collected from unrelated men from various families in Msaken and were tested by an American DNA testing company [4].

Each of these 23 samples corresponds to a unique family name, except for two samples that share the same family name: For the first of these two samples, we conducted tests exclusively on STR markers, while for the second sample, we examined both STR and SNP markers. It is noteworthy that the family names covered in this study collectively account for more than 50% of the Msaken population.

We operate on the assumption that Non-Paternity Events (NPEs) are exceedingly rare in Msaken and, more broadly, in Tunisia. This assumption is based on the conservative nature of the community and the infrequency of adoptions. Therefore, the sample size included in this study is highly indicative of the population under examination. Furthermore, we incorporated additional samples with paternal lineage originating from Msaken. This extension comprised one sample from Beni Kalthoum, a neighboring village, another from Benghazi, Libya, and one from Tunis.

Moreover, we selected and collected supplementary samples that exhibit a genetic match with the Msaken-Haplotype. These samples were subjected to testing by alternative companies such as 23andMe and YSEQ. Additionally, we compiled relevant bibliographic data pertaining to the Y DNA of the Tunisian population.

2.2. Y STR genotyping

To predict the haplogroup of each sample, we used the 12, 37,67 and 111 STR markers panels made available by FTDNA (Family Tree DNA) company (Additional Materials).

Haplogroup determination was carried out in two steps. Initially, the first eight samples were tested in 2008 by FamilyTreeDNA. Subsequently, additional samples were tested in the following years (2008-2020) using either 12, 37, 67, or 111 STR markers. All the STR results are included in the additional material.

3. Results

Among 23 samples tested from Msaken, 21 were found to belong to J-M172 Haplogroup which is parent to J-L271, and two samples were found to belong to R-Z93, which is a subgroup of R1a Haplogroup. The following is Table 1.

Table 1: *Number of samples per Qasr or group.*

Qasr (Msaken) or Place	Number of Samples	Samples Tested with NG Testing Deep SNP Testing
Nejajra	8	2
Jabliyine	2	2
Menaama	3	2
Jedidyine	2	0
Qebliyine	4	0
Qeramsa	2	0
Benghazi, Libya	1	1
Beni Kalthoum	1	0
Total	23	7

3.1. Haplogroup R1a results

Two samples were found to belong to R-Z93 Haplogroup. These samples belong to men from the Qeramsa families. These families are attested to be originating from a neighboring village (Oufim) which died out in the last centuries [2]. Ancestors from Qeramsa migrated to Msaken in the 19th Century and settled in the south-east side of Msaken.

3.1.1. Haplogroup J-M172

The rest of the samples which are 21, belong to J-M172, and more precisely to J-L271 Haplogroup among them:

- One sample from Beni Kalthoum, a village in the southern border of Msaken
- One sample from Benghazi Libya, and with a Tunisian ancestry, most probably from Msaken.
- An additional number of 6 samples from 23andme Company Results and confirmed as belonging to J-L24, and most probably to J-L271.

This company tests Y SNPs with a Y SN panel limited to L24, in the phylogeny of J-L271.
It also tests autosomal SNPs.

3.1.2. Data mining

We decided to concentrate on the outcomes linked to the prevailing Haplogroup in Msaken, which is J-M172. Upon comparing the published STR data from Tunisian sources with the STR haplotypes from Msaken, we observed that matching STRs with Msaken haplotypes were exclusively identified in Tunisia, as indicated in Table 2.

Table 2: *Samples of from published papers which are matching with Msaken.*

Origin place of matching sample	Paper	Year	No. of STR Markers Tested	Total No. of Samples	Origin of samples	Origin of Matching samples	No. of Tunisian Samples	No. of matching samples	Percentage of Matching samples/ Tunisian samples Tested
Tunis	[5]	2004	15	275	Algeria, Tunisia, Egypt	Tunis	148	2	1.35%
Sfax	[6]	2007	13	105	Sfax	Sfax	105	1	0.95%
Sousse	[7]	2014	17	220	Sousse	Sousse	220	12	5.45%

Indeed, the matching STR data is more precisely derived from the Sousse region, most likely from Msaken. To date, no matching STR results with the Msaken group have been discovered outside of Tunisia.

3.2. Msaken results in the landscape of Tunisian DNA

E-M81 and J-M267 are respectively the most prevalent haplogroups in Tunisia while J-M172 Haplogroup in Tunisia is found at a frequency less than 4%; hence, J-M172 is a minor Haplogroup in Tunisia.

Zalloua *et al.* [8] found that there are some lineages in J-M172 which are most probably linked to Phoenician expansion in the Mediterranean area. Just sum up the following citation one of the haplotypes listed below and linked to Phoenician or Greek expansion, matches Msaken haplotypes (Table 3). Thus, linking the Msaken haplotypes to Phoenician or Greek expansion, is very unlikely.

Table 3: *Core haplotypes defining Y-STR haplotype groups associated with the Phoenician or Greek expansions.*

STR+	DYS19, DYS389I, DYS389b, DYS390, DYS391, DYS392, DYS393
PCS1+	14,13,16,24,10,11,12
PCS2+	14,14,17,23,10,11,12
PCS3+	13,12,18,23,10,11,13
PCS4+	14,13,17,23,10,11,12
PCS5+	14,14,16,23,10,11,12
PCS6+	14,13,16,22,10,11,12
GCS1+	13,13,17,24,10,11,13

Fadhlaoui *et al.* [7] tested 220 samples from the region of Sousse and found 18 samples belonging to J-M172. Among them, 12 are marked as J2a1h-L24 with haplotypes matching Msaken results. This seems to indicate that 67% of Sousse Samples belonging to J-M172 are of Msaken type.

3.3. TMRCA calculation

We used the Yfull method [9,10] to calculate the TMRCA based on the SNP found in seven samples for which NG testing has been made. The whole TMRCA of J-L271 is 1496 Years Before Present with 95% CI. The calculation of this TMRCA is based on six samples for which a deep SNP test has been done.

We found a sample which is an outlier of the whole group: positive for the 54 SNPs common to the whole group and negative for three SNPs. These three SNPs were found common to the six samples belonging to three Qasrs (Nejajra, Jabliyine and Menaama).

3.4. Phylogeny of haplogroup J-L271

We uploaded the NG results of our seven samples to the International Yfull Tree. The tree can be found in the following link: yfull.com/tree/j-l271.

The phylogeny we obtained (Figure 2) is concordant with the genealogy of Qasrs founder cited in the book [2] except for sample YF102634 which stands at the root of J-L271.

1. Samples in haplogroup J-FGC32615 are from two different families and belong to the same Qasr founder (AL Nejajra Qasr, J-FGC32589)
2. Samples in Haplogroup J-Y200779 are from different families all belonging to Al-Menaama Qasr. As for the original Qasr of the Libyan sample, it is not known but presumed to be the same.
3. Further tests are required to determine the SNPs common to the following Qsars: Jedidyine, Qebliyine.

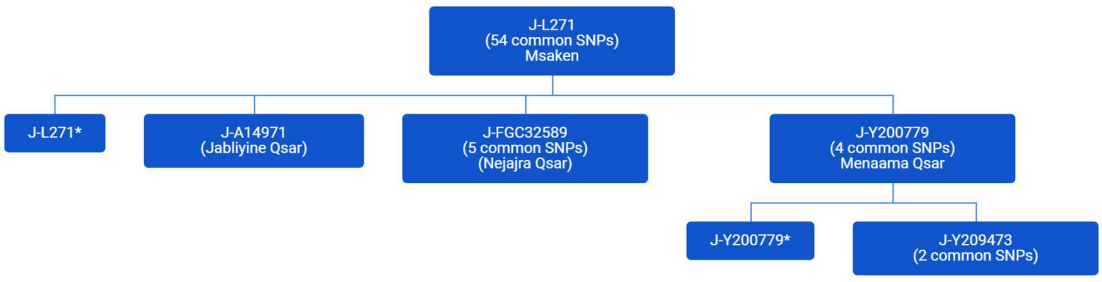

Figure 2: *J-L271 phylogeny.*

3.5. Autosomal data

Autosomal Data plays an important role in the field of human migration history. The actual data collected from our Msaken samples could be used to disentangle the following issues:

3.5.1. Factor V of Leiden

Several cases of venous thrombosis are known in many families of Msaken (personal investigation). Thanks to Autosomal DNA testing, we determined the genetic origin of this illness.

One of the samples tested had more than three episodes of venous thrombosis in both legs, as well as two of his direct siblings and many cousins. We found the SNP rs6025 positive in this sample: [11] Just sum up Hence, we discovered that Factor V of Leiden was among the genetic causes of venous Thrombosis occurring in the Msaken population.

On another hand, at the Factor V of Leiden locus, a selective advantage for heterozygous, favoring immunity against *S. aureus* and *Y. pestis* strains was observed [12].

It has been reported that Tunisia faced many episodes of Plagues in the last centuries [13]. This drastically affected the demography of Tunisia and caused hundreds of thousands of deaths. It is remarkable though that Msaken's population was relatively high compared to other cities [3].

We suspect that the prevalence of Factor V of Leiden played a favorable role in the survival of the Msaken population during these plague episodes; a deeper study could be undertaken about this possible link.

- Our study showed that Msaken city population belongs in majority to J-L271, a subgroup of J-M172 (J2) which has a global frequency of 2.83% over 601 samples from academic papers (15), taking into account that the J-L271 population in Tunisia represents approximately 1% of the whole Tunisian population, we can say that approximately 35% of people in Tunisia belonging to J-M172 are from Msaken.

- This study allowed us to find 54 SNPs which are common to seven samples from Msaken and not found elsewhere so far in the world.

  The rest of the samples (16 samples) were found to be matching the STRs of these seven samples, thus we conclude that they all belong to the same haplogroup J-L271.

- The TMRCA found for the whole group is around 1500 (95% CI) which is prior to the foundation date of Msaken and hence concordant with the tradition.

- The phylogeny we retraced is concordant with the genealogy cited by Mahmoud Gazzah [2], except for an outlier sample which is inside the whole group but standing in the root. More research is required to explain this exception.

- J-L271 haplogroup has a high number of common SNPs (54) revealing a strong bottleneck, and there is no known group matching before 5400 Years BP (estimated relying on the common SNPs).

- The closest group to J-L271 is J-A16345, sharing with J-L271, one unique SNP (FT203595).

This group has its samples from South Turkey (2 samples) and one presumed sample (predicted as J-L192 as per its haplotype) from North Iraq (Mosul) (Figure 3).

- J-FT203595 is a subgroup of J-L192 (yfull.com/tree/j-l192).

Figure 3: *Maps of J-FT203595 samples.*

- J-L192 has a wide geographic distribution, and its highest distribution is found in East Anatolia [14] (Figure 4).

- We may cite one Chalcolithic (sample N°I7494) [15] (Eneolithic / Copper Age) site along the Amu Darya (ancient Oxus) river in Uzbekistan and Tepe Sapali [16] belonging to J-L192.

Figure 4: *Map of J-L192 samples; Red: J-FT203595, Green: J-FGC30635, Purple: J-L192*, Blue: Old sample.*

The Bactria Margiana Archeological Complex (BMAC) culture was clearly a pre-Indo-European culture of Central Asia, which traded tin and lapis lazuli to populations like the Sumerians in the Near East. They interacted with the contemporary Indus Valley Civilization further South. BMAC was among the first people to have ox carts and domesticated the Bactrian camel.

The autosomal DNA shows that the BMAC people were autosomally mostly like Iranian Neolithic farmers with a smaller Anatolian Neolithic component than the Western Iranian Neolithic farmers. Unlike later Chalcolithic people from Godin Tepe (Seh Gabi) in Western Iran, they are lacking any Natufian / Levantine ancestry. However, it is fascinating to notice that sample N° I7494. is autosomally far from the typical BMAC individual, and just similar to someone from Godin Tepe in Western Iran. This ancient sample may then be originating from Western Iran.

- No J-L192 sample was found in 110 ancients Near Eastern individuals spanning the Late Neolithic to Late Bronze Age, a period characterized by intense interregional interactions for the Near East [17].

- Considering the previous remarks, the place of origin of J-L192 may not be far from West Iran. This assumption makes sense and is concordant with what was found about J2a-M530 haplogroup [18] which found that Sum up.

Hence, if we suppose that J2a-M530 is J-L24 itself, from which J-L192 derived, this will mean that Msaken Qasrs ancestors are descendants from one unique man originating from West Asia, most probably West Iran or East Anatolia or Central Asia. Hopefully, with more data from this region in the future we could have a clearer picture about the geographic origin of the inhabitants of Msaken.

The historical circumstances of the migratory event should be studied more in depth considering the different possible scenarios.

3.6. The Chinese Salar sample

After the completion of this paper, we came across a study titled "A Pangenome Reference of 36 Chinese Populations" [19], published in June 2023. In this paper, an individual from XINHUA Autonomous County in China, belonging to the Salar community, was identified as sample HIFI032453D.

This individual share 23 single nucleotide polymorphisms (SNPs) out of 55 with Msaken population. One additional SNP named TY274315 common to Msaken and the Salar sample were found changing the number from 54 to 55 common snps to Msaken and the Salar Sample. Consequently, the Msaken group now only shares 32 SNPs, significantly reducing the bottleneck from 5400 to 3370 years.

The Salar people, a Turkic ethnic minority in China, predominantly speak Salar, an Oghuz language. Numbering 130,607 in the 2010 census, the Salar ethnicity emerged through the intermarriage of male Turkmen migrants from Central Asia with Amdo Tibetan women during the early Ming dynasty. They primarily reside in the Qinghai-Gansu border region, including Xunhua Salar Autonomous County, Hualong Hui Autonomous County, and other areas in Qinghai and Gansu. Additionally, there are Salars in Northern Xinjiang (in the Ili Kazakh Autonomous Prefecture). As a patriarchal agricultural society, they follow the Muslim faith. Noteworthy locations include Gansu's Lintan County, Xining, Linxia County, Qinghai's Hualong Hui Autonomous County, and Xunhua Salar Autonomous County.

It is worth noting that the J2 haplogroup is a minority among the Salars, with 24-30% carrying the O3-M122 haplogroup and 17% the R-M17 haplogroup. Therefore, it appears that the Salar sample belonging to J-FGC32618 represents a minority within the Salar community.

The discovery of this Salar sample that matches with the Msaken group opens new avenues for research to explore the common origins between Salar families residing in China and a predominant population in a distant city in Tunisia, namely Msaken (Figure 5).

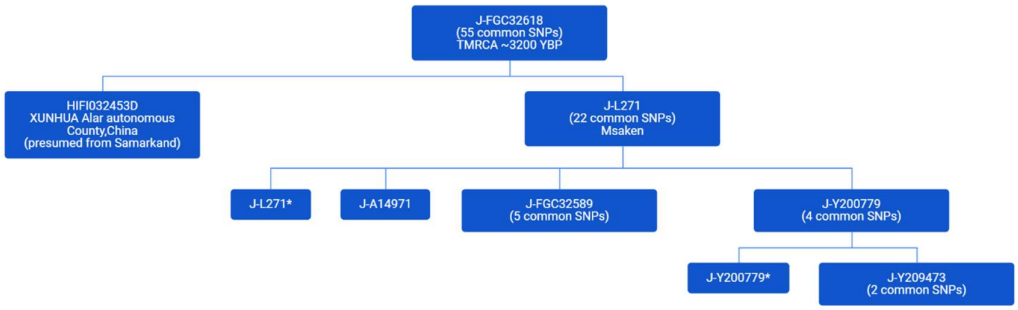

Figure 5: *Phylogeny of Msaken group plus the Salar sample.*

4. Conclusion

In summary, the results of the present study shed light on the genetic heritage of the Msaken city population. Most individuals in this region were found to belong to the J-L271 subgroup of J-M172 (J2), a genetic lineage with a global frequency of 2.83% across various academic samples. Considering that the "J-L271 population" in Tunisia represents only about 1% of the entire Tunisian population, our findings suggest that approximately 35% of Tunisian individuals who carry the J-M172 haplogroup come from Msaken.

Moreover, our research has unveiled 55 Single Nucleotide Polymorphisms (SNPs) that are exclusive to seven samples from Msaken plus an individual from XINHUA Autonomous County, belonging to Salar community and have not been identified elsewhere in the world. Among these 55 SNPS, 22 are exclusive to Msaken population and were never found elsewhere.

These findings are indicative of a distinct genetic profile in the region. Additionally, the Time to Most Recent Common Ancestor (TMRCA) for the entire group, estimated at around 1500 years ago, aligns with the historical foundation date of Msaken, underscoring the consistency with local tradition.

The phylogenetic analysis has revealed a strong concordance with the genealogy recorded by M. Gazzah [2], excepting the case of one intriguing outlier sample situated within the broader group. This outlier warrants further investigation to elucidate the reasons behind its distinctive genetic position.

Notably, the J-L271 haplogroup boasts a relatively high number of common SNPs (32 SNPS), implying a significant bottleneck in its evolutionary history. It is also crucial to highlight that no known group shares a common ancestry with J-L271 before approximately 3370 years ago, based on shared SNPs.

Further expanding the genetic landscape, our study identifies the closest genetic group to J-L271 as J-FGC32618 shared with the Chinese Salar sample then J-A16345 sharing together a unique SNP (FT203595). The distribution of J-FT203595 samples reveals their presence in South Turkey and in a presumed sample from North Iraq (Mosul). J-FT203595 is a subgroup of J-L192, which exhibits a widespread geographic distribution, with East Anatolia as a focal point.

Considering the absence of J-L192 samples in a broader study [19] of ancient Near Eastern individuals, we postulate that the origin of J-L192 may not be far from West Iran, in alignment with previous genetic research. This implies that the ancestors of Msaken's inhabitants likely descended from a common origin in West Asia, possibly West Iran or East Anatolia or Central Asia. The presumed origin of the Salars from Samarkand suggests that a common origin between the population of Msaken and the Salar subgroup might be not far from Samarkand.

While the present study has unveiled essential insights into the genetic history of Msaken, it is vital to conduct further research to explore the historical circumstances surrounding this migratory event, considering various possible scenarios. The genetic information presented here provides a foundation for deeper investigations into the region's past and the origins of its people as well as for regional health issues.

- The number of samples tested was limited to 23 over a 15-year period due to a lack of dedicated project funding.

Most of the expenses were covered by the paper's author, with anonymous contributors to the cost of four samples and for which we express our gratitude.

Ethical Declarations

We diligently confirm that every sample donor has furnished informed consent for the utilization of their samples in DNA analysis.

Acknowledgement

I wish to offer a special recognition to the memory of Mr. Mahmoud Gazzah, whose enduring passion for researching the history of Msaken ignited my own love for this endeavor. His legacy lives on through the precise historical insights and information he generously shared, enriching our understanding of Msaken's history.

We extend our heartfelt gratitude to Dr. Soufia Mourali-Chebil, University of El Manar, Tunis for her invaluable assistance in meticulously reviewing and providing constructive feedback on the document. Her expertise and insights significantly enriched the quality of this work.

Special Recognition: We would like to express our sincere appreciation to Angela Cone, a dedicated citizen scientist. In 2008, her collaborative efforts were instrumental in refining the initial STR and SNP tests. Angela's invaluable contributions have played a crucial role in the development of this project.

I would like to express my gratitude to David Huen and Ted Kandell, who assisted me in the discovery of the Salar sample, and whose contribution was crucial in updating the phylogeny of the Msaken population.

We would like to express my sincere gratitude to the generous sample donors; their invaluable contributions were crucial in facilitating this research.

Additional Materials

Shared SNP in Haplogroup J-L271 :32 SNPS, Group Msaken exclusively:

FGC32585, FGC32586, FGC32587, FGC32588, FGC32590, FGC32593, FGC32601, FGC32602, FGC32605, FGC32606, FGC32607, FGC32608, FGC32612, FGC32616, FGC32617, FT100578, FT285557, FT285647, FT285772, FT285807, FT285883, FT285954, FTA47671, L271, Y16816, Y17249/Y14318, Y200791/FT286021, Y200847/FT286791, Y200996, Y329282, Z14898, FGC32594.

Shared SNP in Haplogroup J-FGC32618 :23 SNPS, Group Msaken plus the Salar Chinese Sample:

Y201103/FT286350, FGC32595, FGC32596, FGC32598, FGC32599, FGC32603, FGC32609, FGC32610, FGC32613, FGC32614, FGC32618, FGC37142, FT285573, FT285576, FT285593, FT285717, FT286255, TY274315, Y223570, Y223571, Y329282, Y49543, Z34061.

STR Data link:

https://docs.google.com/spreadsheets/d/1_aLNxoHwLbxDvbVB3v1IU3h-Jr3Uef_z90It3rzCtjQ/edit?pli=1#gid=0

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