



Short Communication

Y-chromosomal STR analysis in the Pashtun population of Southern Afghanistan

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ABSTRACT

Afghanistan is a landlocked country in the heart of Asia and since the dawn of humankind Afghanistan has faced centuries of turmoil, strife, conflict, warfare, distress, social unrest, difficult climate, harsh terrain and due to its unique geostrategic position in Eurasia which has historically attracted commerce and conflict. It is an important stop along the Silk Road, connecting the far eastern civilizations to the western world. A 5000-year history of constant invasion. Afghanistan has been repeatedly invaded and conquered by rulers and super powers, neighboring interference in this conflict-tattered land for centuries yet rarely leading to the conquest of this rugged and challenging terrain nation. Afghans are not only shepherds, farmers and nomads but also intense fighters and fierce warriors. Currently very limited genetic studies have been performed in Afghan populations.

17 Y chromosomal short tandem repeats (Y-STRs) were analyzed in 125 unrelated Pashtun (in hindi: Pathan) males residing in the Kandahar region of Southern Afghanistan. A total of 92 unique haplotypes were observed. The predominant haplotype reached a frequency of 9.6%. The haplotype diversity was 0.987 and the discrimination capacity 73.6%. Analysis of molecular variance (AMOVA) reveals a considerable regional stratification within the country as well as between different Pashtun (Pathan) groups from Afghanistan, Pakistan and India.

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1. General information about the population

Afghanistan is a multi-ethnic society of about 30 million people. Afghanistan is strategically located between the energy rich republics of central Asia. Afghanistan is bordered by Pakistan in the south and east, Iran in the west and Turkmenistan, Uzbekistan and Tajikistan in the north, and China in the far northeast. Afghanistan has been the center of many powerful empires for the last 2000 years and in the last 30 years this nation has been in chaos due to major wars from the Soviet invasion of 1979 to their withdrawal in 1989 and from warlords to the removal of Taliban in 2001. The Hindu Kush mountain range a sub-range of the Himalayas runs diagonally from the northeast to the Southwest of Afghanistan, separating the northern provinces from the rest of the country. The country is split east to west by the Hindu Kush mountain range, rising in the east. With the exception of the southwest most of the country is covered by high snow-capped mountains and is traversed by deep valleys. Historically the Pashtun have been the most dominant in Afghanistan. The term Afghan, is generally viewed by other people in the country to

refer to the Pashtuns. The royal families of the country were Pashtun and the rest of the ethnic groups make up considerably smaller percentages, vast majority of the people are Muslim. It is estimated that as many as 40 different languages are spoken in Afghanistan. The Pashtun are the largest ethnic group comprising about 40% of the population. They live mainly in the Kandahar, Helmand, Ghazni, Qalat, Mazar e Sharif, Herat, Oruzgan and Kabul provinces of Afghanistan. Other important ethnolinguistic groups are the Tajiks, Hazara, Uzbeks, Aimaks and Turkmens, Baloch and Brahvi. Within the country there are Hindu, Sikh and tiny Jewish communities as well. These ethnic groups are distinctly unique in their different cultural characteristics, such as family beliefs, religion and languages [6].

Here we present a Y-STR population study of 125 Pashtun individuals mainly living in the Southern Afghanistan province of Kandahar and compared the haplotype data to other Pashtun (Pathan) groups living in North and South Afghanistan, Pakistan and India.

2. Extraction, PCR and typing

Blood samples from 125 unrelated Afghan males residing in Kandahar, Helmand, Qalat, Ghazni, Mazar e Sharif were collected with informed consent and DNA was extracted using a

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Table 1

Pairwise comparisons between the Pathan population.

Population	North Afghanistan [Pathan]	Peshawar, Pakistan [Pathan]	South Afghanistan [Pathan]	Uttar Pradesh, India [Afridi Pathan]	India	Iran	Pakistan
North Afghanistan [Pathan]	–	0.0824	0.0836	0.0000	0.0000	0.0001	0.0000
Peshawar, Pakistan [Pathan]	0.0053	–	0.0062	0.0000	0.0000	0.0000	0.0000
South Afghanistan [Pathan]	0.0065	0.0105	–	0.0000	0.0000	0.0000	0.0000
Uttar Pradesh, India [Afridi, Pathan]	0.0978	0.0806	0.1052	–	0.0000	0.0000	0.0000
India	0.0132	0.0111	0.0328	0.0842	–	0.0000	0.0000
Iran	0.0154	0.0136	0.0349	0.0918	0.0041	–	0.0000
Pakistan	0.0117	0.0087	0.0298	0.0866	0.0026	0.0035	–
Afghanistan [Pathan] ^a	0.0476	0.0316	0.0557	0.1064	0.0348	0.0434	0.0363

^a This study non-significant *P* values >0.05 printed in bold.

salting out procedure and quantified spectrophotometrically. All samples were genotyped for the 17 Y-chromosomal STR loci included in the AmpFISTR® Yfiler® PCR Amplification kit (Applied Biosystems, Foster City, USA), namely DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a/b, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 and GATA H4. Alleles were designated according to the latest recommendations of the International Society for Forensic Genetics [1].

3. Quality control

The CEMB laboratory in Lahore (Pakistan) where this study has been carried out, participated in the Y-STR haplotype reference database (YHRD) [2] quality assurance exercise by typing the 17 Yfiler® loci for blind samples (certificate dated May 09, 2011). The haplotype data were sent prior to submission to the YHRD for evaluation. We received the YHRD accession number YA003701 for the population study “Southern Afghanistan [Pathan]”. All data are uploaded to release 37 of the YHRD.

4. Data analysis

Haplotype diversity (*h*) was computed according to Nei [3]. Discrimination capacity (DC) was estimated as the proportion of different haplotypes in the population sample. We used the website www.yhrd.org/Analyse to perform AMOVA variance analysis (Φ_{st} statistics taking the single step-mutation model into account) including significance tests for all pairwise comparisons. The Pashtun (Pathan) population from this study was compared to the North and South Afghanistan Pashtuns (YHRD accession numbers YA003702 and YA003703 published in [4]), Pathans from Pakistan (YA003218 published in [5]) and Afridi Pathans from Uttar Pradesh in India (YA003686 submitted to YHRD by the Central Forensic Science Laboratory, Kolkata, India). In addition full population sets were used for AMOVA, i.e. 1885 haplotypes from India, 444 from Iran and 718 from Pakistan. Since all haplotype overlap in only 7 markers (DYS19, DYS389I-II, DYS390, DYS391, DYS392 and DYS393) this format was chosen for the analysis. Results were visualized in an MDS plot as available from the YHRD website (Fig. 1).

5. Results

Haplotype data and allelic frequencies are presented in the online available [Supplementary Material Tables S1 and S2](#). 92 different unique haplotypes were identified in the sample which corresponds to a discrimination capacity (DC) of 73.6%. The haplotype diversity was 0.987. The most frequent haplotype occurs 12 times (9.6%) and does not occur in the YHRD (release 37

from June 21, 2011) with 97,575 haplotypes. Nevertheless, a reduction to the core haplotype consisting of 9 loci (minimal haplotype) mainly returns matches from the neighboring populations in India, Pakistan, Iran and Azerbaijan indicating the historical relatedness of populations in this Central Asian region. In order to analyze the relationship of Pathan populations living in Afghanistan, Pakistan and India as well as with other population from the neighboring countries including Iran, we compared 7-locus haplotypes via AMOVA with published data from the YHRD. In total, 3498 haplotypes were compared. All pairwise comparisons between the Pathan population analyzed here and the other groups were significant at the 5% level but a closer vicinity was found to the Pathan populations of Peshawar (Pakistan) and North Afghanistan. Pairwise Φ_{st} and *P* values are shown in Table 1.

Our study is consistent with previous analyses of the Afghan male population [4] demonstrating the strong effect of population substructures in this region. Our study also shows the inhomogeneity of the Pashtun (Pathan) ethnic group which is widely dispersed across Afghanistan, Pakistan and India. Notably the Indian Afridi Pathans show a large genetic distance (Fig. 1). The stratified population structure must be observed by construction of a national population database for Afghanistan whether planned for autosomal or linear markers.

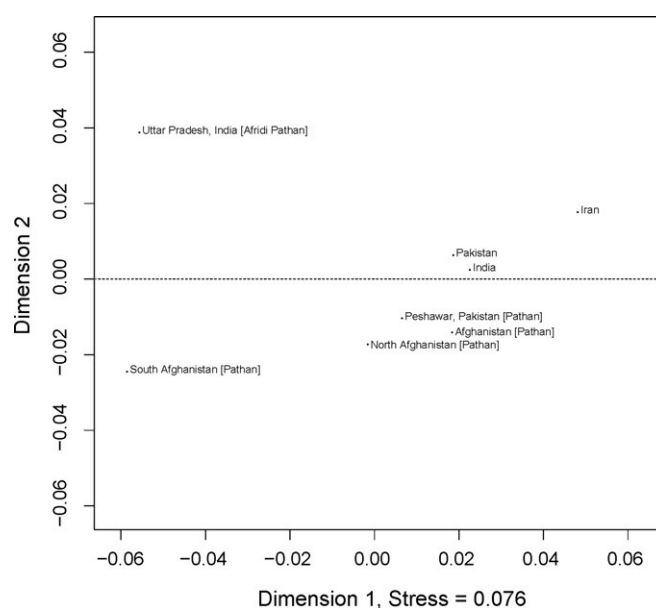


Fig. 1. MDS plot showing Pathan population of Afghanistan, India and Pakistan compared with each other.

Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at doi:[10.1016/j.fsigen.2011.10.005](https://doi.org/10.1016/j.fsigen.2011.10.005).

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