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Research Article

Allele Frequency of 15 Autosomal Short Tandem Repeat Loci in Iranian Population with Comparison to Some Other Population

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Abstract

Background: Short tandem repeat (STR) markers are extensively being used for human identification as well as paternity and forensic analysis of biological evidence.
Objectives: The aim of this study was to investigate the allelic frequencies and several forensic and paternity parameters of 15 autosomal short tandem repeat (STR) loci D3S1358, D16S5391, D7S820, D8S1179, D21S11, D18S51, D5S818, D13S317, FGA, THO1, TPOX, CSF1PO, vWA , D2S1338, and D19S433 in the Iranian population.
Methods: Estimation of allelic frequencies and several forensic and paternity parameters of 15 STR loci were performed with the AmpFLSTR Identifilerr kit (Applied Biosystems) for 274 unrelated individuals living in Iran.
Results: No deviation from Hardy-Weinberg equilibrium was found in any loci studied in this population. Among the 15 STR loci analyzed in the Iranian sample, the most discriminating loci were D21S11, D2S1338, D19S433, D18S51 and FGA with the highest power of discrimination. The allelic distribution also was compared to 13 other populations including 3 Iranian population living in Syria, Dubai, the USA and in Fars province and 8 population from published studies of Azerbaijan, Bolu in Turkey, Morocco, Syria, Iraq, Saudi Arabia, Turkey, East Anatolia, and Pakistan.
Conclusions: It was concluded that the population of present study had the least similarity with Azerbyjani (11 loci) and most similarity with the Iranian population in USA (15 loci).

Keywords: Allele Frequency, STR, Iran, AmpFLSTR Identifiler

1. Background

The human genome sequence is composed of many repetitive regions, and these repeat regions are found not only in the coding regions but also in the non-coding regions of the genes. More than 30% of the human genome contains these repeat regions. Short tandem repeat or STRs are one of these repeated DNA sequences dispersed across the entire human genome. STR loci are highly polymorphic markers and their repeat unit motif varies between 2 - 6 bp. Due to their high polymorphism and heterozygosity as a powerful tool in providing genetic maps, and utilized in forensics, and linkage analysis. The frequency of STR alleles, in different races, and even in certain geographical regions, show variations. Therefore, the investigation of frequency of each STR allele in each particular and specific population is essential for proper interpretation of the results (1-3).

2. Objectives

The aim of this study was to investigate the allelic distribution and several forensic parameters in Iranian population and compare these results with some neighboring population. To this end, a group of individuals referred for paternity testing were chosen for 15 STR loci, including the 13 CODIS core STR loci (D3S1358, D16S5391, D7S820, D8S1179, D21S11, D18S51, D5S818, D13S317, FGA, THO1, TPOX, CSF1PO, and vWA), in addition to D2S1338 and D19S433 were carried out on these samples. To make sure, these individuals are not in any way related, only non-consanguineous parents, as well as the excluded offsprings, were included in the study. As a result, they may be regarded as a representative of the Iranian population.

3. Methods

3.1. Sample Preparation

Peripheral blood samples were collected from 274 unrelated individuals. DNA extraction was performed according to the standard salting-out method (4). Quality and quantity of the DNA samples were evaluated by NanoDrop Spectrophotometer (Thermo Scientific). PCR amplification of 15 autosomal STR loci plus amelogenin locus for sex determination was carried out using by AmpFLSTR Identifiler kit followed by separation of the fragments on ABI Genetic Analyzer 3130 (Applied Biosystem). Raw data analyzed with GeneMapper ID software V. 3.2 (Applied Biosystem).

3.2. Statistical Analysis

Informed written consent was obtained from all individuals and the study was approved by the local ethics committee.

Several population parameters such as the power of discrimination (PD), match probability (PM), polymorphism information content (PIC), the power of exclusion (PE), and typical paternity index (TPI) were calculated by Power-Stats V. 1.2 software (Promega Corporation, USA). The observed and expected heterozygosity (HO, HE) and polymorphism information content (PIC) were computed by Arlequin V. 3.1 software. Inter-population comparison was performed using the pairwise Fst value and sequential Bonferroni correction.

4. Results and Discussion

STR allele frequencies for each locus and the results of statistical analysis of their parameters are summarized in Table 1. The observed allele frequencies ranged between 0.0018 - 0.482. No deviation from Hardy-Weinberg equilibrium was detected in any of STR loci. All markers were shown a high degree of genetic polymorphism, with observed heterozygosity (HO) values ranging from 0.614 (TPOX) to 0.879 (D8S1179) and PD values ranging from 0.851 (TPOX) to 0.974 (D2S1338). PIC varied between 0.64 (TPOX) and 0.88 (D2S1338) indicating good informativeness and suitability of 15 STR loci for forensic and population study.

The most discriminating loci according to their power of exclusion (PE) and power of discrimination (PD) values were D2S1338 (PE = 0.744, PD = 0.974), D18S51 (PE = 0.722, PD = 0.972), FGA (PE = 0.635, PD = 0.963), D19S433 (PE = 0.607, PD = 0.958) and D21S11 (PE = 0.674,PD = 0.957), whereas the score of TPOX (PE = 0.315, PD = 0.851) appeared to be the lowest.

The allele frequency spectrum detected in this study were compared with the published data of 13 different studies on regional and related population including Moroccans (5), Syrians (5), Azerbaijanis (6), Iranians living in Dubai (7), Iranians living in the USA (8), Iraqis (9), Bolu (10), Turkey (11), East Anatolia (12), population of Fars province in Iran (13) and Pakistan (14) populations. The data is shown in Table 2.

According to this inter-population comparison, our population has the greatest difference with Azerbaijani population (11 loci) and Saudi Arabia population (6 loci), whereas our results are very similar to Iranian individuals residing in the USA and Iraqi population. FGA was found to be the most differentiating locus among all compared population. While the locus D5S818 did not show significant difference between different pathological purposes. In conclusion, the findings of this study could be useful as a reference for Iranian population genetic and forensic study.

Footnotes

Conflict of Interests: The authors declare no conflict of interests.

Ethical Considerations: Informed written consent was obtained from all individuals and the study was approved by the local ethics committee.

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References

1. Butler JM. *Forensic DNA typing: Biology, technology, and genetics of STR markers*. 2th ed. New York: Elsevier Academic Press; 2005.
2. Butler JM. Genetics and genomics of core short tandem repeat loci used in human identity testing. *J Forensic Sci*. 2006;**51**(2):253–65. doi: 10.1111/j.1556-4029.2006.00046.x. [PubMed: 16566758].
3. Butler JM. Short tandem repeat typing technologies used in human identity testing. *Biotechniques*. 2007;**43**(4):ii–v. [PubMed: 18019344].
4. Miller SA, Dykes DD, Polesky HF. A simple salting out procedure for extracting DNA from human nucleated cells. *Nucleic Acids Res*. 1988;**16**(3):1215. [PubMed: 3344216]. [PubMed Central: PMC334765].
5. Rakha A, Yu B, Hadi S, Sheng-Bin L. Population genetic data on 15 autosomal STRs in a Pakistani population sample. *Leg Med (Tokyo)*. 2009;**11**(6):305–7. doi: 10.1016/j.legalmed.2009.08.001. [PubMed: 19740693].
6. Abdin L, Shimada I, Brinkmann B, Hohoff C. Analysis of 15 short tandem repeats reveals significant differences between the Arabian populations from Morocco and Syria. *Leg Med (Tokyo)*. 2003;**5 Suppl 1**:S150–5. [PubMed: 12935576].
7. Nasibov E, Shahzad MS, Cengiz M, Bulbul O, Ilyas M, Jafri SS, et al. Allele frequencies of 15 STR loci using AmpFLSTR Identifiler kit in Azerbaijan population. *J Clin Pat and Forensic Med*. 2011;**2**(4):25–32.
8. Alshamali F, Alkhayat AQ, Budowle B, Watson ND. STR population diversity in nine ethnic populations living in Dubai. *Forensic Sci Int*. 2005;**152**(2-3):267–79. doi: 10.1016/j.forsciint.2004.09.133. [PubMed: 15978355].

9. Shepard EM, Herrera RJ. Iranian STR variation at the fringes of biogeographical demarcation. *Forensic Sci Int*. 2006;**158**(2-3):140–8. doi: 10.1016/j.forsciint.2005.05.012. [PubMed: 15998573].
10. Barni F, Berti A, Pianese A, Boccellino A, Miller MP, Caperna A, et al. Allele frequencies of 15 autosomal STR loci in the Iraq population with comparisons to other populations from the middle-eastern region. *Forensic Sci Int*. 2007;**167**(1):87–92. doi: 10.1016/j.forsciint.2006.03.005. [PubMed: 16616444].
11. Akbasak BS, Budowle B, Reeder DJ, Redman J, Kline MC. Turkish population data with the CODIS multiplex short tandem repeat loci. *Forensic Sci Int*. 2001;**123**(2-3):227–9. [PubMed: 11728752].
12. Cakir AH, Celebioglu A, Altunbas S, Yardimci E. Allele frequencies for 15 STR loci in Van-Agri districts of the Eastern Anatolia region of Turkey. *Forensic Sci Int*. 2003;**135**(1):60–3. [PubMed: 12893137].
13. Hedjazi A, Nikbakht A, Hosseini M, Hoseinzadeh A, Hosseini SM. Allele frequencies for 15 autosomal STR loci in Fars province population, southwest of Iran. *Leg Med (Tokyo)*. 2013;**15**(4):226–8. doi: 10.1016/j.legalmed.2013.01.005. [PubMed: 23477811].
14. Osman AE, Alsafar H, Tay GK, Theyab J, Mubasher M, Sheikh NEE, et al. Autosomal short tandem repeat (STR) variation based on 15 loci in a population from the central region (Riyadh province) of Saudi Arabia. *J Forensic Res*. 2015;**6**. doi: 10.4172/2157-7145.1000267.
15. Tuğ A, Erkol Z, Çetinyurek A, Alakoç YD, Elma C, Büken B, et al. Allele distribution data for 16 short tandem repeat loci in Bolu. *Turk J Med Sci*. 2010;**40**(4):659–64. doi: 10.3906/sag-0805-63.

Table 1. Allele Frequency of 15 Autosomal STR in Iranian Population

Allele	D8S1179	D21S11	D7S820	CSF	D3S1358	THO1	D13S317	D16S539	D2S1338	D19S433	VWA	TPOX	D18S51	D5S818	FGA
5												0.0053			
5.3						0.0018									
6			0.0036			0.2589									
7			0.0396			0.2158	0.0054	0.0018				0.0162		0.0018	
8	0.0125		0.1493	0.0018		0.1169	0.1529	0.0486				0.4802	0.0018	0.0126	
8.3			0.0036			0.0018									
9	0.0054		0.08816	0.0198		0.2194	0.0881	0.1691				0.1097	0.0054	0.0701	
9.3						0.1654									
10	0.1025		0.25	0.2679		0.0162	0.0504	0.1043		0.0018		0.0737	0.0054	0.1115	
10.2										0.0144			0.0018		
11	0.0809		0.205	0.3453		0.0036	0.2859	0.2662		0.0072		0.2662	0.0306	02878	
11.2			0.2248							0.0036			0.0036		
12	0.1295			0.3093			0.3039	0.2266		0.1169		0.0467	0.1223	0.3345	
12.2			0.0306							0.0090			0.0054		
13	0.2482			0.0467	0.0054		0.0791	0.1601		0.2320	0.0054	0.0018	0.1259	0.1708	
13.2			0.0054		0.0574					0.0252			0.0108		
14	0.1960		0.1493	0.0072			0.0342	0.0197		0.2608	0.10254		0.1906	0.0162	
14.2					0.259		0.0054			0.0342			0.0036		
15	0.1655			0.0018			0.3039	0.0036	0.0036	0.1025	0.1277		0.1403	0.0018	
15.2					0.2734					0.0845	0.0018				
16	0.0539								0.0323	0.0557	0.2014		0.1241		
16.2					0.2374					0.0324			0.0036		
17	0.0054								0.1655	0.0108	0.2626		0.0827	0.0018	
17.2					0.1582					0.0054					
18					0.0072				0.117	0.0018	0.2086		0.0683		0.0054
19									0.1115	0.0018	0.0773		0.0342		0.0539
19.2					0.0018										0.0072
20					0.0054				0.1259		0.0108		0.0252		0.0845
20.2															0.0018
21									0.0395		0.0018		0.0126		0.1313
21.2															0.0072
22									0.0485						0.1906
22.2															0.009
23									0.1385				0.0018		0.212
23.2															0.0018
24									0.1043						0.1367
24.2															0.0018
25		0.0036							0.0900						0.1133

25.2															0.0018
26		0.0018							0.0162						0.0342
27		0.0108							0.0072						0.0054
28		0.1691													
28.2		0.0072													
29		0.2050													0.0018
29.2		0.0108													
30		0.194													
30.2		0.0179													
31		0.0647													
31.2		0.0971													
32		0.0144													
32.2		0.1439													
33		0.0018													
33.2		0.0467													
34		0.0018													
34.2		0.0018													
35		0.0072													
35.2															
36															
37															
Ho	0.879	0.838	0.809	0.739	0.813	0.79	0.761	0.805	0.875	0.805	0.816	0.614	0.868	0.794	0.82
He	0.839	0.856	0.812	0.712	0.775	0.779	0.787	0.811	0.888	0.84	0.815	0.679	0.885	0.765	0.86
P	0.2019	0.3201	0.8811	0.2227	0.0223	0.6796	0.6945	0.8989	0.8855	0.4049	0.9327	0.0476	0.9109	0.2985	0.2708
PIC	0.8172	0.8376	0.7837	0.6556	0.7363	0.7666	0.7547	0.783	0.875	0.8208	0.7871	0.6319	0.8726	0.7275	0.843
PE	0.751	0.674	0.607	0.489	0.614	0.594	0.527	0.607	0.744	0.607	0.635	0.315	0.722	0.587	0.635
MP	0.052	0.043	0.065	0.142	0.099	0.074	0.078	0.066	0.026	0.042	0.065	0.144	0.028	0.107	0.037
PD	0.948	0. 957	0.935	0.858	0.901	0.926	0.922	0.934	0.974	0.958	0.935	0.851	0.972	0.893	0.963
TPI	4.11	3.11	2.56	1.91	2.61	2.46	2.08	2.56	3.99	2.56	2.77	1.32	3.66	2.42	2.77

Abbreviations: PD, power of discrimination; PE, power of exclusion; PIC, polymorphic information content; PM, probability matching; TPI, typical paternity index.

Table 2. The Result of Comparison Between 15 Autosomal STR in Iranian Population and 13 Other Populations[a]

Variables	IR-FARS[A]	Blou[B]	Pakistan[C]	Morocco[D]	Syria[E]	Azerbaijan[F]	IR-Dubai[G]	IR-USA[H]	Iraq[J]	Saudi Arabia[K]	East Anatolia[L]	Turkey[M]
D8S1179	0.86364	0.0*	0.06364	0.23636	0.80909	0.0*	0.89091	0.54545	0.6	0.51818	0.92727	0.08182
D21S11	0.67273	-	-	0.23636	0.15455	0.0*	0.0*	0.88182	0.66364	0.05455	0.58182	0.88182
D7S820	0.33636	0.91818	0.25455	0.23636	0.1	0.0*	0.65455	0.32727	0.12727	0.02727*	0.0*	0.0*
CSF1PO	0.95445	0.2	0.51818	0.56364	0.27273	0.0*	0.82727	0.85455	0.51818	0.08182	0.83636	0.97273
D3S1358	0.96364	-	-	0.87273	0.95455	0.0*	0.8	0.64545	0.14545	0.24545	0.63636	0.76364
THO1	0.95455	0.62727	0.92727	0.0*	0.0*	0.0*	0.94545	0.67273	0.70909	0.34545	0.38182	0.90909
D13S317	0.82727	0.4	0.73636	0.2	0.63636	0.33636	0.48182	0.9	0.20909	0.0*	0.67273	0.82727
D16S539	0.04545*	0.8	0.76364	0.01818*	0.61818	0.0*	0.0*	0.09091	0.10909	0.03636*	0.26364	0.7273
D2S1338	0.99091	0.82727	-	-	-	-	-	0.85455	0.56364	0.0*	-	-
D19S433	0.92727	0.0*	-	-	-	-	-	0.48182	0.09091	0.0*	-	-
vWA	0.93636	0.0*	0.33636	0.4	0.2	0.0*	0.00909*	1.0	0.1	0.19091	-	-
TPOX	0.0*	0.02727*	0.0*	0.03636*	0.36364	0.0*	0.89091	0.77273	0.52727	0.0*	0.05455	0.01818*
D18S51	0.74545	0.09091	0.46364	0.03636*	1.0	0.0*	0.27273	1.0	0.96364	0.0*	0.94545	1.0
D5S818	0.46364	0.19091	0.08182	0.25455	0.31818	0.75455	0.99091	0.98182	0.68182	0.38182	0.79091	0.94545
FGA	0.39091	0.08182	0.2	0.0*	0.0*	0.0*	0.0*	0.44545	0.50909	0.0*	0.0*	0.40909

[a], *Indicates a significant difference. Ref: A (13), B (15), C (5), D & E (6), F (7), G (8), H (9), J (10),K (14), L (12), M (11).