Allelic Distribution in Population of Rajasthan, India; Inferred from 22 Autosomal STRs included in Powerplex Fusion 5C System

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Abstract

Genetic polymorphism indices and forensic parameters at 22 autosomal short tandem repeat (STR) loci were evaluated in 248 unrelated individuals of north-western Indian state Rajasthan. In total, 258 alleles were observed with an average of 11.72 alleles per locus. The most polymorphic and discriminative locus was Penta E with a value of 0.904 and 0.913 respectively. The combined power of exclusion and the combined power of discrimination were found to be 0.999999999 and 1 respectively. Additionally, the genetic relationship of the studied population with the reported Indian as well as global populations was investigated. The studied population showed genetic affinity with the previously reported population of Rajasthan and geographical close population's i.e. populations of Uttar Pradesh, Madhya Pradesh and Jharkhand. All the STR loci were found polymorphic, and the select panel of STRs was found suitable for population genetic studies and forensic analysis.

Keywords: Power plex fusion 5C system; Rajasthan; Genetic diversity; Polymorphism; STRs.

Introduction

Rajasthan is the largest Indian state by area in north-west Indian geographical province. It is bordered by five other Indian states: Punjab to the north; Haryana and Uttar Pradesh to the north-east; Madhya Pradesh to the south-east; and Gujarat to the south-west[1] (Fig. 1). The state covers an area of 342,239 square kilometres or 10.4 percent of the total geographical area of India. The geographic features of Rajasthan are the Thar Desert and the Aravalli Range, which runs through the state from south-west to north-east, almost from one end to the other, for more than 530 miles. As the history goes, Rajasthan has been a major route of human migration, which is reflected in its genetic diversity.[2] According to 2011 Census of India,

Fig. 1: Geographic location of studied population.

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Rajasthan consists of 68,548,437 people (35,550,997 males and 32,997,440 females), contribute 5.66% to the total population of the country.[3,4]

Materials and Method

In this study, we received 248 individual (147 male and 101 female) samples for routine forensic case analysis at State Forensic Science Laboratory, Jaipur, Rajasthan, India during the period between December 2019 to march, 2020. All samples are collected from generous donors with informed consent and as per the declaration of Helsinki. The sample associated information was provided along with the samples, and only unrelated individuals were selected for the study. No minor was included in this study. DNA extraction, quantification, amplification, fragment analysis and statistical evaluation was done as described in our previous publication.[5,6,7] The genetic data were subjected to statistical evaluation using various statistical software's as described in our publication.[5] Internal laboratory control standards and kit controls were used at all steps of analysis. This article follows the population data publication guidelines formulated by the journal.

Results and Discussion

The studied population showed wide range of allele frequencies from 0.002 to 0.438, which are shown in Table 1. A total of 258 alleles with the mean allele number 11.72 per locus were observed for the studied population. The locus Penta E showed the highest number of observed alleles i.e. 19 whereas locus D3S1358 showed the least number of observed alleles i.e. 6 among all the studied loci. The allele 15 of locus D22S1045 was observed as the most frequent allele among all the studied loci in the studied population. The forensic parameters analyzed for the studied population are shown in Table 2. The range of discrimination power was found between 0.851 (CSF1PO) to 0.979 (Penta E) and cumulative value for all the studied loci was found to be 1. The range of polymorphic information contents found to be 0.653 (CSF1PO) and 0.904 (Penta E). Thus the locus Penta E was observed most polymorphic and discriminatory among all the studied loci. The range of exclusion power was observed between 0.377 (D22S1045) to 0.777 (Penta E) with the cumulative value for all the studied loci was found to be 0.999999999. The observed heterozygosity ranged from 0.665 (D22S1045) to 0.891 (Penta E) for the studied population. The matching probability and paternity index for the studied loci were found to be 1.72X10-26 and 1.11 x109 respectively (Table 2).

Other Remarks

The genetic data of the studied population was compared at common 15 autosomal Short Tandem Repeats (STR's) with the Indian population namely; Balmiki (Punjab),[8] Mahadev Koli (Maharashtra),[8] Oraon Chotanagpur),[9] Kora (Bengal),[10] Maheli (Bengal),[10] Central Indian Population,[11] Population of Jharkhand,[12] UP Population,[13] Tamil Population (Tamil Nadu),[14] Population of Rajasthan[5] and population of Rajathan-1,[6] and global populations namely; Americans (Basque),[15] Han population of Southern China,[16] Tibetan population (Nepal) Royal Kingdom of Bhutan Population,[17] Manchu population of China,[18] Tibet[19] and Korean population.[20] The Neighbour Joining (NJ) tree revealed the genetic relatedness between the studied and compared populations (Fig. 2). Principal Component Analysis (PCA) plot (Fig. 3) showed the graphical representation of the studied and compared populations based on Nei's genetic distances (Table 3). NJ and PCA showed the consistency in the results, in which studied population pooled with the geographical close population's with the previously reported populations of Rajasthan, Uttar Pradesh, Jharkhand and Central Indian rather than the geographical distinct populations.

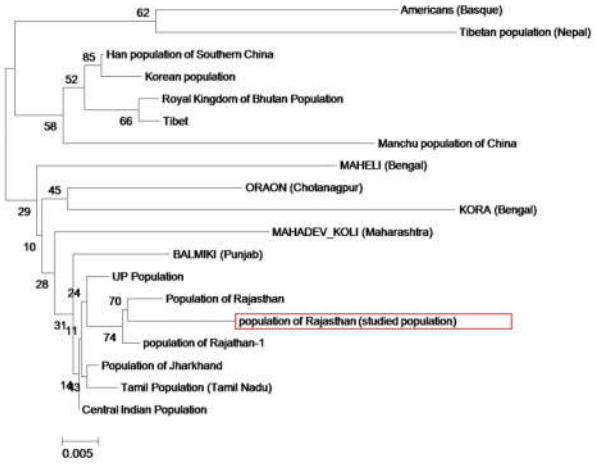

Fig. 2: NJ tree comparison based on Fst genetic distance calculated from autosomal STRs.

Table 1: Allele frequencies for the population of rajasthan (n=248).

Locus	D3S1358	D1S1656	D2S441	D10S1248	D13S317	PENTA–E	D16S539	D18S51	D2S1338	CSF1PO	PENTA–D	TH01	vWA	D21S11	D7S820	D5S818	TPOX	D8S1179	D12S391	D19S433	FGA	D22S1045
5	–	–	–	–	–	0.081	–	–	–	–	0.002	–	–	–	–	–	–	–	–	–	–	–
6	–	–	–	–	–	0.010	–	–	–	–	0.010	0.230	–	–	–	–	0.002	–	–	–	–	–
7	–	–	–	–	0.010	0.054	–	–	–	0.002	0.002	0.169	–	–	0.006	–	–	–	–	–	–	–
8	–	0.012	0.006	–	0.208	0.008	0.058	–	–	0.002	0.012	0.137	–	0.004	0.220	0.006	0.321	0.010	–	–	–	–
8.3	–	–	–	–	–	–	–	–	–	–	–	0.002	–	–	–	–	–	–	–	–	–	–
9	–	0.002		–	0.083	0.014	0.139	–	–	0.028	0.198	0.286	–	–	0.042	0.016	0.151	0.004	–	0.006	–	–
9.3	–	–	–	–	–	–	–	–	–	–	0.002	0.161	–	–	–	–	–	–	–	–	–	–
10	–	–	0.304	0.002	0.087	0.056	0.115	0.004	0.002	0.147	0.173	0.006	–	0.002	0.226	0.125	0.115	0.173	–	0.006	–	0.006
11	–	0.175	0.405	0.016	0.244	0.147	0.290	0.024	0.002	0.341	0.284	–	–	0.002	0.238	0.327	0.379	0.103	–	0.004	–	0.240
11.3	–	–	0.040	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–
12	–	0.093	0.044	0.012	0.258	0.131	0.252	0.089	0.004	0.391	0.119	–	–	–	0.224	0.335	0.022	0.073	–	0.044	–	0.006
12.2	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.014	–	–
12.3	–	–	0.004	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–
13	–	0.145	0.026	0.143	0.065	0.105	0.115	0.101	–	0.071	0.141	–	0.002	–	0.042	0.181	0.002	0.196	–	0.268	–	0.002
13.2	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.006	–	–
14	0.028	0.103	0.145	0.230	0.040	0.060	0.026	0.268	–	0.014	0.046	–	0.147	–	0.002	0.010	0.002	0.202	–	0.264	–	0.040
14.2	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.077	–	–
15	0.298	0.169	0.018	0.262	0.004	0.075	0.002	0.161	0.002	0.004	0.008	0.002	0.063	–	–	–	–	0.155	0.004	0.115	–	0.438
15.2	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.060	–	–
15.3	–	0.006	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–
16	0.308	0.155	0.002	0.278	0.002	0.083	0.002	0.125	0.002	–	0.002	–	0.212	–	–	–	–	0.067	0.008	0.071	–	0.192
16.2	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.002	–	0.042	–	–
16.3	–	0.020	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–
17	0.250	0.042	–	0.054	–	0.095	–	0.105	0.052	–	–	0.002	0.292	–	–	–	0.006	0.012	0.127	0.018	–	0.071
17.3	–	0.040	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.002	–	–	–
18	0.105	0.006	0.002	0.002	–	0.026	–	0.040	0.202	–	–	–	0.173	–	–	–	–	0.004	0.246	0.004	0.010	0.006
18.3	–	0.028	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.026	–	–	–
19	0.010	–	0.002	–	–	0.020	–	0.052	0.181	–	–	0.002	0.083	–	–	–	–	–	0.109	–	0.028	–
19.3	–	0.002	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.008	–	–	–
20	–	–	–	–	–	0.014	–	0.008	0.133	–	–	0.002	0.020	–	–	–	–	–	0.127	–	0.087	–
21	–	–	–	–	–	0.010	–	0.008	0.036	–	–	–	–	–	–	–	–	–	0.135	–	0.091	–
22	–	–	–	–	–	0.006	–	0.010	0.044	–	–	–	–	–	–	–	–	–	0.077	–	0.151	–

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22.2	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.014	–
23	–	–	–	–	–	0.004	–	0.004	0.181	–	–	–	–	–	–	–	–	–	0.085	–	0.183	–
23.2	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.008	–
24	–	–	–	–	–	–	–	–	0.095	–	–	–	–	–	–	–	–	–	0.030	–	0.240	–
24.2	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.010	–
25	–	–	–	–	–	–	–	–	0.050	–	–	–	–	–	–	–	–	–	0.012	–	0.137	–
26	–	–	–	–	–	–	–	–	0.012	–	–	–	–	–	–	–	–	–	0.004	–	0.028	–
27	–	–	–	–	–	–	–	–	–	–	–	–	–	0.014	–	–	–	–	–	–	0.010	–
28	–	–	–	–	–	–	–	–	–	–	–	–	0.004	0.169	–	–	–	–	–	–	0.002	–
29	–	–	–	–	–	–	–	–	–	–	–	–	–	0.159	–	–	–	–	–	–	–	–
29.2	–	–	–	–	–	–	–	–	–	–	–	–	–	0.002	–	–	–	–	–	–	–	–
30	–	–	–	–	–	–	–	–	–	–	–	–	0.002	0.167	–	–	–	–	–	–	–	–
30.2	–	–	–	–	–	–	–	–	–	–	–	–	–	0.044	–	–	–	–	–	–	–	–
31	–	–	–	–	–	–	–	–	–	–	–	–	–	0.073	–	–	–	–	–	–	–	–
31.2	–	–	–	–	–	–	–	–	–	–	–	–	–	0.145	–	–	–	–	–	–	–	–
32	–	–	–	–	–	–	–	–	–	–	–	–	–	0.004	–	–	–	–	–	–	–	–
32.2	–	–	–	–	–	–	–	–	–	–	–	–	0.002	0.145	–	–	–	–	–	–	–	–
33.2	–	–	–	–	–	–	–	–	–	–	–	–	–	0.056	–	–	–	–	–	–	–	–
34.2	–	–	–	–	–	–	–	–	–	–	–	–	–	0.010	–	–	–	–	–	–	–	–
35.2	–	–	–	–	–	–	–	–	–	–	–	–	–	0.002	–	–	–	–	–	–	–	–

Table 2: Forensic parameters for the population of Rajasthan (n=248).

locus	**D3S1358**	**D1S1656**	**D2S441**	**D10S1248**	**D13S317**	**PENTA-E**	**D16S539**	**D18S51**	**D2S1338**	**CSF1PO**	**PENTA-D**	**TH01**	**vWA**	**D21S11**	**D7S820**	**D5S818**	**TPOX**	**D8S1179**	**D12S391**	**D19S433**	**FGA**
GD	0.743	0.873	0.719	0.779	0.812	0.913	0.804	0.854	0.860	0.705	0.815	0.793	0.808	0.867	0.792	0.734	0.718	0.848	0.864	0.828	0.851
PIC	0.696	0.858	0.673	0.741	0.785	0.904	0.775	0.837	0.842	0.653	0.788	0.760	0.780	0.850	0.757	0.686	0.669	0.828	0.848	0.806	0.832
PM	0.123	0.034	0.133	0.086	0.071	0.021	0.081	0.040	0.044	0.149	0.071	0.077	0.070	0.037	0.078	0.122	0.133	0.048	0.039	0.052	0.047
PD	0.877	0.966	0.867	0.914	0.929	0.979	0.919	0.960	0.956	0.851	0.929	0.923	0.930	0.963	0.922	0.878	0.867	0.952	0.961	0.948	0.953
Hobs	0.786	0.851	0.754	0.726	0.806	0.891	0.798	0.847	0.823	0.758	0.827	0.786	0.843	0.851	0.758	0.702	0.750	0.839	0.855	0.782	0.867
He	0.741	0.871	0.717	0.777	0.811	0.911	0.802	0.852	0.858	0.703	0.814	0.792	0.807	0.865	0.790	0.732	0.717	0.846	0.862	0.826	0.849
PE	0.574	0.696	0.517	0.469	0.611	0.777	0.596	0.689	0.642	0.524	0.649	0.574	0.681	0.696	0.524	0.431	0.510	0.673	0.704	0.567	0.729
TPI	2.340	3.351	2.033	1.824	2.583	4.593	2.480	3.263	2.818	2.067	2.884	2.340	3.179	3.351	2.067	1.676	2.000	3.100	3.444	2.296	3.758
P value	0.176	0.001	0.563	0.003	0.000	0.000	0.000	0.003	0.000	0.189	0.000	0.067	0.006	0.000	0.043	0.001	0.111	0.005	0.000	0.001	0.074

GD- Gene diversity, PIC- Polymorphic informatin content, PM- matching probability, Hobs- Observed heterozygosity, He- Expected heterozygosity, PE-Power of exclusion, TPI-Typical paternity index, P value- Hardy weinberg equilibrium test

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CPD	1
CPE	0.999999999
CPI	1.11×10^{9}
CPM	1.72×10^{-26}

Table 3: Nei's Da distance matrix between studied and compared populations.

	BALMIKI (Punjab)	MAHADEV_KOLI (Maharashtra)	ORAON (Chotanagpur)	KORA (Bengal)	MAHELI (Bengal)	Central Indian Population	Population of Jharkhand	UP Population	Americans (Basque)	Han population of Southern China	Tibetan population (Nepal)	Royal Kingdom of Bhutan Population	Manchu population of China	Tamil Population (Tamil Nadu)	Tibet	Korean population	Population of Rajasthan	population of Rajathan-1	population of Rajasthan (studied population)
BALMIKI (Punjab)	0																		
MAHADEV_KOLI (Maharashtra)	0.045	0																	
ORAON (Chotanagpur)	0.05	0.057	0																
KORA (Bengal)	0.073	0.087	0.078	0															
MAHELI (Bengal)	0.06	0.072	0.074	0.094	0														
Central Indian Population	0.015	0.032	0.03	0.066	0.046	0													
Population of Jharkhand	0.019	0.035	0.032	0.064	0.05	0.002	0												
UP Population	0.019	0.04	0.04	0.067	0.051	0.003	0.005	0											
Americans (Basque)	0.066	0.09	0.094	0.122	0.086	0.056	0.062	0.059	0										
Han population of Southern China	0.036	0.054	0.047	0.074	0.071	0.021	0.024	0.024	0.075	0									
Tibetan population (Nepal)	0.093	0.11	0.095	0.129	0.111	0.078	0.079	0.082	0.075	0.064	0								
Royal Kingdom of Bhutan Population	0.048	0.064	0.051	0.086	0.081	0.029	0.031	0.033	0.082	0.012	0.057	0							
Manchu population of China	0.078	0.092	0.077	0.105	0.097	0.061	0.065	0.063	0.118	0.048	0.103	0.056	0						
Tamil Population (Tamil Nadu)	0.017	0.037	0.038	0.066	0.057	0.007	0.006	0.009	0.066	0.03	0.084	0.038	0.07	0					
Tibet	0.046	0.061	0.059	0.09	0.077	0.025	0.032	0.033	0.082	0.013	0.055	0.006	0.058	0.038	0				
Korean population	0.043	0.062	0.052	0.083	0.078	0.027	0.033	0.032	0.082	0.006	0.069	0.02	0.054	0.036	0.018	0			
Population of Rajasthan	0.026	0.045	0.045	0.08	0.059	0.012	0.014	0.012	0.065	0.033	0.088	0.038	0.071	0.016	0.039	0.031	0		
population of Rajathan-1	0.021	0.043	0.041	0.074	0.055	0.008	0.011	0.011	0.064	0.028	0.086	0.035	0.068	0.012	0.034	0.026	0.008	0	
population of Rajasthan (studied population)	0.035	0.053	0.056	0.088	0.07	0.022	0.026	0.025	0.072	0.039	0.094	0.044	0.079	0.026	0.045	0.038	0.02	0.018	0

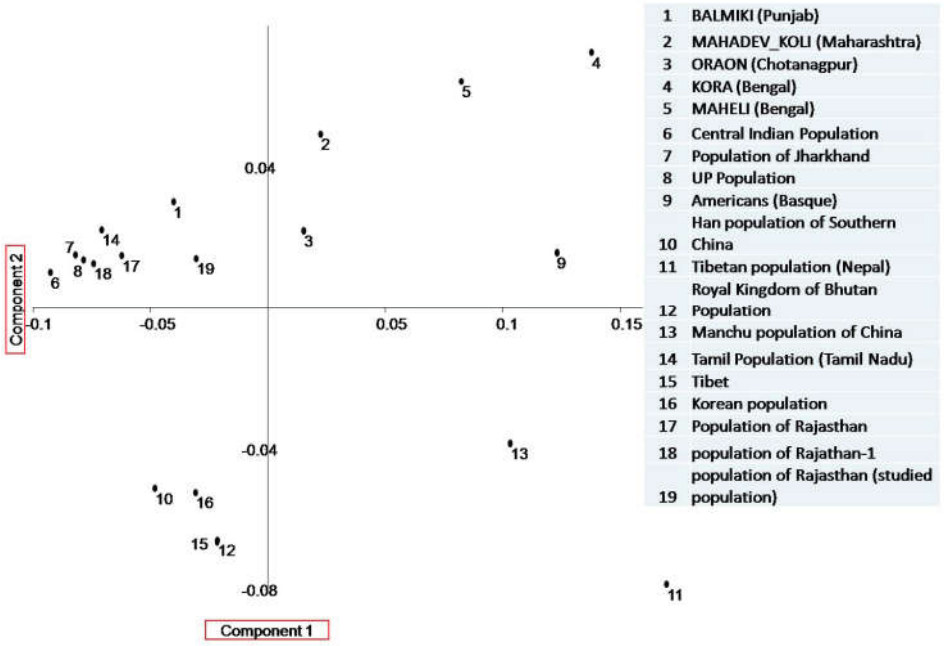

Fig. 3: PCA plot comparison based on Fst genetic distance calculated from autosomal STRs.

Conclusion

The locus Penta E was observed as the most polymorphic among the studied loci. The genetic data of this study enrich the DNA data bank and useful forensic application as well as anthropological studies.

Conflict of Interest: The authors have declared no conflict of interest.

Role of funding source: No funding source for this study.

Ethical Statement: The study was conducted in compliance with ethical standards and written inform consent was obtained from the donors and followed the declarations of Helsinki.

Author's contribution: RK, AK and RK (Kumawat) designed the study, RK (Kumawat) and AT analyzed the samples, RK did the quality check and statistical analysis. RK (Kumawat) and RK wrote the manuscript. AK and AT reviewed the manuscript. All authors read and approved the final manuscript.

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