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Genetic polymorphism analyses of a novel panel of 19 X-STR loci in the Chinese Uygur ethnic minority*#

Yu-xin GUO[1,2,3], Jian-gang CHEN[4], Yan WANG[5], Jiang-wei YAN[6], Jing CHEN[6], Tian-hua YAO[7], Li-ping ZHANG[4], Guang YANG[8], Hao-tian MENG[1,2,3], Yu-dang ZHANG[1,2,3], Ting MEI[4], Yao-shun LIU[4], Qian DONG[1,2,3], Bo-feng ZHU†‡[1,2,3]

([1]*Key Laboratory of Shaanxi Province for Craniofacial Precision Medicine Research, College of Stomatology, Xi'an Jiaotong University, Xi'an 710004, China*)

([2]*Clinical Research Center of Shaanxi Province for Dental and Maxillofacial Diseases, College of Stomatology, Xi'an Jiaotong University, Xi'an 710004, China*)

([3]*Research Center of Stomatology, Stomatological Hospital, Xi'an Jiaotong University, Xi'an 710004, China*)

([4]*Department of Biochemistry, Preclinical Medicine College, Xinjiang Medical University, Urumqi 830011, China*)

([5]*Criminal Police Detachment of Yili Public Security Bureau, Yili 835000, China*)

([6]*Key Laboratory of Genome Sciences, Beijing Institute of Genomics, Chinese Academy of Sciences, Beijing 100101, China*)

([7]*Department of General Dentistry, Stomatological Hospital, Xi'an Jiaotong University, Xi'an 710004, China*)

([8]*Department of Pathology and Laboratory Medicine, Loma Linda University Medical Center, California 92354, USA*)

†E-mail: zhubofeng7372@126.com

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Abstract: The population genetic data and forensic parameters of 19 X-chromosome short tandem repeat (X-STR) loci in Chinese Uygur ethnic minority are presented. These loci were detected in a sample of 233 (94 males and 139 females) unrelated healthy individuals. We observed 238 alleles at the 19 X-STR loci, with the corresponding gene frequencies spanning the range from 0.0021 to 0.5644. After Bonferroni correction (P>0.0026), there were no significant deviations from Hardy-Weinberg equilibrium. The cumulative power of discrimination in females and males, and the probability of exclusion of the 19 X-STR loci were 0.999 999 999 999 999 999 998 091, 0.999 999 999 999 966, and 0.999 999 986 35, respectively. The cumulative mean exclusion chance was 0.999 999 992 849 in deficiency cases, 0.999 999 999 999 628 in normal trios, and 0.999 999 998 722 in duo cases. The high value of the forensic parameters mentioned above revealed that the novel panel of 19 loci had important values for forensic applications in the Uygur group.

Key words: X-chromosome, Short tandem repeat (STR), Uygur, Genetic polymorphism, Forensic
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1 Introduction

At present, short tandem repeat (STR) loci are applied broadly in paternity testing and individual identification of forensic cases in forensic DNA laboratories all over the world (Rosenberg *et al.*, 2002; Deng *et al.*, 2013; Wang *et al.*, 2013; Zhu *et al.*, 2013; 2014). X-chromosome STRs (X-STRs), which contain the characteristics of both autosomal and uniparental

‡ Corresponding author
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ORCID: Bo-feng ZHU, http://orcid.org/0000-0001-7877-7387

genetic markers, are verified to be of high-efficiency in cases such as the tests of mother-son kinship, half-sisters having a common biological father without the father's DNA, or grandmother-granddaughter relationships (Liu *et al.*, 2008; Nadeem *et al.*, 2009; Luo *et al.*, 2011). Although a few panels of X-STRs have been used, these X-STR kits were not enough for forensic applications, because of insufficient X-STR loci. Thus, 19 X-STRs (DXS8378, DXS7423, DXS10148, DXS10159, DXS10134, DXS7424, DXS10164, DXS10162, DXS7132, DXS10079, DXS6789, DXS101, DXS10103, DXS10101, HPRTB, DXS6809, DXS10075, DXS10074, and DXS10135) were selected to build up a novel X-STR panel.

Uygur is one of the important ethnic minorities in the People's Republic of China and a large proportion of Uygurs live in the Xinjiang Uygur Autonomous Region, China (Xu, 2003; Jin and Chu, 2006). It is significant to obtain the information of various genetic markers in Uygur for forensic identification and population genetics. As in the previous reports, we investigated the genetic polymorphisms of 24 Y-chromosomal STR haplotypes (Zhu *et al.*, 2014), killer cell immunoglobulin-like receptor genes (Wang *et al.*, 2012), 21 autosomal STR loci (Deng *et al.*, 2013), and HLA-A, -B, and -DRB1 loci with sequence-based typing (Shen *et al.*, 2010) in the Chinese Uygur ethnic group, but the genetic polymorphism analyses of the novel panel of 19 X-STRs mentioned above have not yet been reported until now. In the present study, we used the panel to estimate the allelic frequencies of the 19 X-STR loci, calculate the important forensically statistical parameters for each locus in a sample of 233 unrelated individuals, and evaluate the allelic frequency differentiations of these loci between the Uygur group and other groups, in order to gain a better understanding of the Uygur overall genetic background.

2 Materials and methods

2.1 Sample preparation and DNA extraction

Informed consent was obtained from all the eligible individuals, and bloodstain samples were collected from 233 unrelated healthy Uygur individuals (139 females and 94 males) living in Ili of the Xinjiang Uygur Autonomous Region, China. The criteria of sample selection were: the ancestors should be unrelated within at least three generations, come from the Uygur ethnic group, and have no family migration. The study was conducted according to the human and ethical research principles of the Stomatological Hospital, Xi'an Jiaotong University, China. After sample collection, the Chelex-100 method was used to extract genomic DNA from the bloodstain samples as described by Walsh *et al.* (2013).

2.2 PCR amplification and X-STR genotyping

Multiplex polymerase chain reaction (PCR) was performed by the AGCU X19 STR fluorescence amplification reagents (AGCU ScienTech Inc., Wuxi, Jiangsu, China) in a single PCR system in accordance with the manufacturer's instructions. PCR was performed on a GeneAmp PCR System 9700 Thermal Cycler (Applied Biosystems, Foster City, CA, USA) in a 25-μl reaction volume containing reaction mix, X19 primers, C-Taq, and sterile distilled H_2O (sdH_2O). Capillary electrophoresis was carried out on the ABI Genetic Analyzer 3500 (Applied Biosystems, Foster City, CA, USA) following the manufacturer's instructions. Alleles of 19 X-STR loci were genotyped using the GeneMapper ID software V3.2 (Applied Biosystems, Foster City, CA, USA), based on peak heights reaching a set threshold value of 50 relative fluorescence units.

2.3 Statistical analyses

The allelic frequencies of 19 X-STR loci and Hardy-Weinberg equilibrium (HWE) were analyzed by the modified Powerstate V1.2 spreadsheet (Promega, Madison, WI, USA) (Tereba, 1999). The *P*-value of HWE tests was adjusted using the Bonferroni correction (P=0.05/19=0.0026). HWE was only calculated in the female samples. Forensic statistical parameters including polymorphism information content (PIC), heterozygosity (HET), the power of discrimination in females (PDF) and males (PDM), and mean exclusion chances (MEC) were performed with ChrX-STR.org 2.0 (http://www.chrx-str.org) based on allelic frequencies. Pairwise linkage disequilibrium (LD) analysis was estimated by the Genepop Version 4.0.10 (http://genepop.curtin.edu.au). In order to measure the differences of allele frequencies among different populations, the locus-by-locus *Fst* and *P*-values were calculated based on 19 X-STR allele frequencies

using the analysis of molecular variance (AMOVA) method by ARLEQUIN Version 3.0 software (Excoffier *et al.*, 2007).

3 Results and discussion

3.1 Forensic parameter analysis

HWE was tested in female samples and no significant deviations were observed after Bonferroni correction (P>0.0026). There were no significant differences for the allelic frequencies of 19 X-STRs between male and female subgroups according to exact tests, except DXS10134, DXS7424, DXS101 and DXS10074 loci. Therefore, allelic frequencies of male and female samples in these 4 loci are shown separately, while the remaining loci are shown together. The allele frequency distributions are shown in Tables 1 and 2. Forensic statistical parameters of the 19 X-STR loci including HET, PDF, PDM, probability of exclusion (PE), paternity index (PI), PIC, and MEC are shown in Table 3. The allelic frequencies of 19 X-STR loci ranged from 0.0021 to 0.5644. The HET ranged from 0.5493 at the DXS 10164 locus to 0.9258 at the DXS10135 locus. The highest and the lowest values of PE were observed at the DXS10135 locus (PE=0.8572) and the DXS10164 locus (PE=0.2928), respectively. The PDF ranged from 0.7889 at the DXS10164 locus to 0.9908 at the DXS10135 locus, and the PDM ranged from 0.6016 at the DXS10164 locus to 0.9301 at the DXS10135 locus. The cumulative PDF, PDM, and PE of the 19 X-STR loci were 0.999 999 999 999 999 999 998 091, 0.999 999 999 999 966, and 0.999 999 986 35, respectively. In the 19 X-STRs, the highest and the lowest values of PIC were surveyed at the DXS10164 locus (PIC=0.54923) and the DXS10135 locus (PIC= 0.9258), respectively. The highest value of MEC in deficiency cases, normal trios and duo cases were observed at the DXS10135 locus (MEC=0.8585, 0.9256, and 0.8663, respectively). The lowest values were found at the DXS10164 locus (MEC=0.3595, 0.5493, and 0.4021, respectively). The cumulative MEC in deficiency cases, normal trios and duo cases were 0.999 999 992 849, 0.999 999 999 999 628 and 0.999 999 998 722, respectively. The parameters mentioned above revealed that the panel of 19 loci had great potential for forensic personal identification and forensic paternity testing.

3.2 Linkage disequilibrium analyses

It is necessary to estimate the value of LD before these STR loci are used for forensic application and population genetics. In this study, among the 171 pairwise loci of 19 X-STRs, P-values of 8 pairwise loci were found to be <0.05 in the Chinese Uygur ethnic minority. However, only one (between DXS10075 and DXS10074, P<0.0001) remained significant after Bonferroni correction (P=0.05/171= 0.0003). The value of LD is affected by various factors, for example, genetic linkage, natural selection, and demographic structure. The distance between the two markers (DXS10075 and DXS10074) is 0.021 Mbp (http://chrx-str.org), and therefore genetic linkage is likely to be a major cause of the observed LD.

3.3 Haplotype diversities

According to previously reports, there were seven linkage groups observed in the 19 X-STRs. These clusters were listed as follows: DXS10135-DXS8378-DXS10148, DXS7132-DXS10074-DXS 10075-DXS10079, DXS10103-DXS10101-HPRTB, DXS10134-DXS7423, DXS10159-DXS10162-DXS 10164, DXS6809-DXS6789, and DXS7424-DXS101. The first four clusters were obtained from a database (http://xdb.qualitype.de/xdb/linkageTable.jsf), and the remaining clusters from previous studies (Edelmann *et al.*, 2002; 2010; Szibor *et al.*, 2005). Haplotype frequencies for the seven clusters in 94 male samples are displayed in the Table S1. The most common haplotypes in the Uygur ethnic minority were observed at DXS10134-DXS7423 (H36-15) and DXS7424-DXS101 (H16-24) with a frequency of 0.1064, followed by DXS10134-DXS7423 (H36-14, H37-15) and DXS6809-DXS6789 (H32-21) at 0.0957. Haplotype diversities for the seven cluster groups were 0.9954, 0.9977, 0.9920, 0.9556, 0.9792, 0.9669 and 0.9634, respectively.

3.4 Interpopulation differentiation

Population differentiation between the Uygur and 11 other previously published groups was analyzed by the means of AMOVA based on the allelic frequencies of 9 overlapping STR loci. The *Fst* and P-values of these loci are shown in the Table S2. The results showed that statistically significant differences (P<0.05) were observed between the Uygur and

Greenlander (Tomas *et al.*, 2012) and Somali (Tomas *et al.*, 2012) at 8 loci; East Timor (Moreira *et al.*, 2015), Japanese (Uchigasaki *et al.*, 2013), Shenyang Han population (Uchigasaki *et al.*, 2013) and Dane (Tomas *et al.*, 2012) at 6 loci; Shanghai Han population (Zhang *et al.*, 2012) at 5 loci; Bhil tribal population (Shrivastava *et al.*, 2015) and Taiwanese (Chen *et al.*, 2014) at 4 loci; Malay (Samejima *et al.*, 2012) and Guangdong Han population (Zeng *et al.*, 2011) at 3 loci. The results of AMOVA demonstrated that the allelic frequency in most of these loci distributed differently among different ethnic groups. Therefore, more studies on STR loci in different ethnic groups should improve understanding of the genetic relationships between the different ethnic groups and their genetic backgrounds.

Table 1 Allele frequency distributions in Chinese Uygur group at 15 X-STR loci with no difference between male and female samples

Allele	DXS 8378	DXS 7423	DXS 10148	DXS 10159	DXS 10164	DXS 10162	DXS 7132	DXS 10079	DXS 6789	DXS 10103	DXS 10101	HPRTB	DXS 6809	DXS 10075	DXS 10135
8	0.0043				0.0193										
9	0.0408				0.0300							0.0021			
10	0.3970				0.5644							0.0043			
11	0.3283				0.2639		0.0129					0.0708			
12	0.2017	0.0021			0.0880		0.0923					0.2940			
12.1														0.0021	
13	0.0129	0.0429			0.0343		0.2983					0.3498		0.0365	
14	0.0150	0.3476					0.3391		0.0064	0.0064		0.1845			
14.2														0.0064	
15		0.4893				0.0064	0.2146	0.0215	0.1116	0.0494		0.0858		0.0064	0.0021
15.2										0.0021					
16		0.1159	0.0021			0.0494	0.0322	0.0451	0.1373	0.2275		0.0064		0.2103	0.0043
16.1															
16.2										0.0021				0.0129	
17		0.0021				0.1824	0.0107	0.0773	0.0043	0.0837		0.0021		0.4099	0.0107
17.2						0.0043								0.0021	
17.3															
18			0.1073			0.3734		0.1116	0.0021	0.1524				0.2833	0.0322
18.1															0.0021
18.2										0.0021					
18.3														0.0021	
19			0.0730			0.2532		0.2103	0.0515	0.3605				0.0279	0.0451
19.1															0.0021
20			0.0258	0.0021		0.1116		0.3219	0.2639	0.1094					0.0923
20.1			0.0021												0.0086
20.2															0.0021
21			0.0043	0.0021		0.0193		0.1330	0.2489	0.0043					0.0923
21.1			0.0021												0.0150
22			0.0064	0.0150				0.0579	0.1352						0.1159
22.1			0.0579												0.0021
23			0.0043	0.0815				0.0215	0.0386						0.0794
23.1			0.0665												0.0086
23.2				0.0021											
24			0.0107	0.2811											0.1137
24.1			0.1288												0.0043
24.2				0.0021											
25			0.0021	0.2511							0.0043				0.0644
25.1			0.1416												
26				0.1781							0.0043				0.0536

To be continued

Table 1

Allele	DXS 8378	DXS 7423	DXS 10148	DXS 10159	DXS 10164	DXS 10162	DXS 7132	DXS 10079	DXS 6789	DXS 10103	DXS 10101	HPRTB	DXS 6809	DXS 10075	DXS 10135
26.1			0.1373												
26.2			0.0043												
27				0.1180							0.0193		0.0021		0.0451
27.1			0.0901												
27.2			0.0021								0.0343				
28				0.0665							0.0236		0.0064		0.0536
28.1			0.0665												
28.2											0.0644				
29											0.0150		0.0258		0.0451
29.1			0.0365												
29.2											0.0880				
30											0.0794		0.0172		0.0408
30.1			0.0193												
30.2											0.1180				
31											0.1159		0.1266		0.0172
31.1			0.0043												
31.2											0.1009				
32											0.1395		0.2339		0.0215
32.1			0.0043												
32.2											0.0451				
33											0.0901		0.2554		0.0064
33.2											0.0043				
34											0.0408		0.2082		0.0107
34.2											0.0043				
34.3											0.0064		0.0021		
35											0.0021		0.0923		0.0064
36													0.0279		
37													0.0021		
38															0.0021

Table 2 Allele frequency distribution in Chinese Uygur group at 4 X-STR loci with difference between male and female samples

Allele	DXS10134		DXS7424		DXS101		DXS10074	
	Female	Male	Female	Male	Female	Male	Female	Male
7							0.0471	
8							0.0290	
10							0.0036	
11			0.0362					
12			0.0326					
13			0.0109	0.1158				
14			0.1449	0.0526			0.0145	
15			0.3696	0.2474	0.0181	0.0316	0.0652	0.0316
16			0.2681	0.4000			0.2065	0.2737
17			0.1051	0.1526			0.2609	0.0895
17.3							0.0036	
18			0.0254	0.0211	0.0145	0.0632	0.2391	0.1579
19			0.0072	0.0105	0.0254		0.1159	0.3421
20					0.0072	0.0105	0.0145	0.1053
21					0.0181	0.0316		
22					0.0254	0.0211		
23					0.1051	0.1053		

To be continued

Table 2

Allele	DXS10134		DXS7424		DXS101		DXS10074	
	Female	Male	Female	Male	Female	Male	Female	Male
24					0.2500	0.2211		
25					0.1558	0.2263		
26					0.1449	0.1368		
27					0.1232	0.1053		
28					0.0616	0.0211		
29					0.0362	0.0158		
30					0.0145			
31	0.0108					0.0105		
32	0.0288	0.0213						
33	0.0612	0.0851						
34	0.0971	0.0851						
35	0.1511	0.1064						
35.3	0.0036							
36	0.2086	0.2553						
36.1	0.0036							
36.3	0.0036							
37	0.1655	0.1915						
37.2	0.0036							
37.3	0.0216	0.0106						
38	0.1439	0.0638						
38.2	0.0036	0.0106						
38.3	0.0072	0.0106						
39	0.0468	0.0532						
39.3	0.0108	0.0319						
40	0.0036							
40.3	0.0108	0.0106						
41	0.0036							
41.3	0.0036	0.0213						
42.3	0.0036	0.0213						
43.3	0.0036	0.0213						

Table 3 Forensic efficiency parameters of 19 X-STR loci in Chinese Uygur group

Allele	PIC	HET	PE	PI	PDF	PDM	MEC1	MEC2	MEC3
DXS8378	0.6350	0.6948	0.4158	0.1541	0.8482	0.6919	0.4324	0.6350	0.4899
DXS7423	0.5556	0.6272	0.3213	0.1878	0.7901	0.6245	0.3536	0.5556	0.4097
DXS10148	0.8969	0.9086	0.8051	0.0476	0.9832	0.9047	0.8069	0.8966	0.8202
DXS10159	0.7729	0.8044	0.6009	0.0995	0.9323	0.8010	0.6102	0.7725	0.6481
DXS10134	0.8563	0.8079	0.7330	0.0654	0.9700	0.8692	0.7407	0.8559	0.7606
DXS7424	0.7281	0.7661	0.5319	0.1186	0.9090	0.7628	0.5545	0.7281	0.5950
DXS10164	0.5493	0.6042	0.2928	0.1992	0.7889	0.6016	0.3596	0.5493	0.4021
DXS10162	0.7091	0.7511	0.5061	0.1261	0.8977	0.7479	0.5260	0.7091	0.5716
DXS7132	0.6960	0.7434	0.4932	0.1299	0.8883	0.7402	0.5058	0.6960	0.5566
DXS10079	0.7867	0.8132	0.6172	0.0951	0.9408	0.8097	0.6358	0.7867	0.6666
DXS6789	0.7897	0.8181	0.6264	0.0927	0.9408	0.8146	0.6354	0.7897	0.6696
DXS101	0.8437	0.8623	0.7119	0.0707	0.9651	0.8586	0.7206	0.8437	0.7428
DXS10103	0.7426	0.7769	0.5509	0.1132	0.9178	0.7736	0.5725	0.7425	0.6113
DXS10101	0.9036	0.9145	0.8170	0.0447	0.9850	0.9105	0.8187	0.9036	0.8304
HPRTB	0.7032	0.7479	0.5008	0.1276	0.8933	0.7447	0.5168	0.7032	0.5649
DXS6809	0.7839	0.8139	0.6184	0.0948	0.9375	0.8104	0.6260	0.7839	0.6621
DXS10075	0.6548	0.7082	0.4362	0.1474	0.8627	0.7051	0.4584	0.6547	0.5112
DXS10074	0.7821	0.8120	0.6150	0.0957	0.9369	0.8085	0.6250	0.7821	0.6604
DXS10135	0.9258	0.9341	0.8572	0.0350	0.9908	0.9301	0.8585	0.9256	0.8663

PIC, polymorphism information content; HET, heterozygosity; PE, probability of exclusion; PI, paternity index; PDF, power of discrimination in female; PDM, power of discrimination in male; MEC1, mean exclusion chances (MEC) in deficiency cases, mean exclusion chance for deficiency cases; MEC2, MEC in normal trios, mean exclusion chance for normal trios; MEC3, MEC in duo cases, mean exclusion chance for duo cases

4 Conclusions

In summary, the 19 X-STR loci were used to investigate the genetic polymorphisms of the Uygur ethnic minority and the population differentiations between the Uygur group and other populations. The value of cumulative PDF, PDM, and PE showed that these 19 X-STR loci could be used as complement for the application of autosomal STR. Moreover, these results provided basic data for population genetics and forensic science research.

Compliance with ethics guidelines

Yu-xin GUO, Jian-gang CHEN, Yan WANG, Jiang-wei YAN, Jing CHEN, Tian-hua YAO, Li-ping ZHANG, Guang YANG, Hao-tian MENG, Yu-dang ZHANG, Ting MEI, Yao-shun LIU, Qian DONG, and Bo-feng ZHU declare that they have no conflict of interest.

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2008 (5). Informed consent was obtained from all volunteers for being included in the study.

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List of electronic supplementary materials

Table S1 Haplotype diversities of seven clusters in Uygur male individuals (n=94)

Table S2 *Fst* and *P*-values for allele frequency distribution between Uygur ethnic group and the other groups previously published

中文概要

题　目：新疆维吾尔族人群 19 个 X-STR 新组合基因座遗传多态性研究

目　的：研究新疆维吾尔族人群 19 个 X 染色体短串联重复序列（X-STR）基因座和他们组成的 7 组连锁基因座的单倍型多样性，评价 19 个 X-STR 新组合位点的多态信息量和累积个体识别力。为群体遗传学和法医学的应用基础研究提供数据支持；并比较维吾尔族和 11 个民族在共有的 X-STR 基因座的遗传学差异。

创新点：首次应用一个新的复合扩增检测体系，研究 19 个 X-STR 基因座新的组合（DXS8378、DXS7423、DXS10148、DXS10159、DXS10134、DXS7424、DXS10164、DXS10162、DXS7132、DXS10079、DXS6789、DXS101、DXS10103、DXS10101、HPRTB、DXS6809、DXS10075、DXS10074 和 DXS10135）在新疆维吾尔族的遗传多态性。

方　法：从 233 个新疆维吾尔族无关、健康个体的血痕中提取基因组 DNA。应用一个新的复合扩增体系，同时对 19 个 X-STR 基因座进行扩增，用毛细管电泳进行基因扫描和分型。系统分析和评价这些 X-STR 基因座常用的各种法医学参数及应用价值；并对 7 组连锁的基因座组成的单倍型进行分析。基于分子方差分析的方法对新疆维吾尔族和其他 11 个民族共有的 X-STR 基因座进行比较研究，探寻这些群体在等位基因频率分布上的差异。

结　论：研究 19 个 X-STR 基因座共发现 238 个等位基因，相应的基因频率分布在 0.0021~0.5644；女性累积个体识别力为 0.999 999 999 999 999 999 998 091，男性为 0.999 999 999 999 966，累积非父排除率为 0.999 999 986 35。分子方差分析的结果显示新疆维吾尔族人群与格陵兰和索马里人群差异最大，有 8 个基因座存在差异；与马来西亚和关中汉族人群差异最小，仅 3 个基因座存在差异。结果表明：这 19 个 X-STR 基因座多态性高、且具有较高的累积个体识别力，可很好地应用于法医学及群体遗传学研究，也为新疆维吾尔族人群的遗传背景的研究提供基础资料。

关键词：X 染色体；短串联重复序列（STR）；维吾尔族；基因多态性；法医学