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Evaluation of the reliability of SE33 locus in forensic DNA identification in Turkish Population

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Abstract

Forensic DNA studies is a powerful tool in criminal investigations and paternity test. In recent years, STR (Short Tandem Repeat) DNA loci are being used more and more in forensic identification all over the world. In this context, polymorphism rates and frequency of the SE33 region, which is an autosomal STR locus, is aimed to be determined about its reliability. Evaluations were made on the samples of 500 individuals who did not have close relatives. Genomic DNA isolation was carried out by DNA isolation kits (EZ-1 investigator kit and Power Plex Fusion 6C Direct kit) previously by the authorized staff of İzmir Forensic Medicine Group Presidency. SE33 STR locus amplification was achieved with the Powerplex Fusion 6C kit from the isolated DNA. In the study, homozygote genotype rate was 5.4%, heterozygote genotype rate was 94.6%, power of discrimination (PD) 0.992 and matching probability (PM) was 0.008. The lowest frequency alleles were determined as 7,10,16.3,17.2,23,24,29, 34.2,35.2,37 alleles with 0.1%. It is seen that SE33 STR locus is a strong marker in forensic identification compared to other studies in terms of power of discrimination and matching probability.

Keywords: GSE33 loci, STR, polymorphism, forensic identification

Introduction

SE33 was first reported as an STR marker neighboring to a beta actin related pseudogene (ACTBP2, also historically used as the STR locus name) on chromosome 6. [1]. The repetition set was determined as (AAAG)n [2,3]. In 1993, extended sequencing of the region was removed and concluded that the variation in SE33 was more extensive than previous studies showed. [4].

SE33 has been incorporated into several STR kits because it's high polymorphism, it's power of discrimination and the fact that more than 140 alleles have been reported to this day [5-7]. STR analysis is widely used in forensic identification. STR DNA analysis have a great importance in polymorphism and population genetics studies for reasons such as specific discrimination and reliability.

In order to use a genetic marker safely in forensic sciences, it is necessary to know the polymorphic properties such as homozygot, heterozygot states and allele frequency of a certain population.

Many autosomal loci are used in forensic identification. SE33 STR locus is one of the most distinguishing regions among these locus. Forensic identification conventionally started with the 13 CODIS core STR loci [8]. However, STR loci with high discriminatory power were needed to prevent errors for cases such as increased regional polymorphism similarity rates and close consanguineous marriages in Turkey. In addition to the high rate of consanguineous marriages, it is necessary to identify the parents of individuals who may arise from incestuous relationships. It has been reported that SE33 is a strong marker for identification in many regions outside our country [9-11]. This situation needs to be tested in forensic cases in our country.

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On the other hand considering the data of the strbase.nist.gov website; SE33 has the highest mutation rate when that compared to other STR loci. Therefore, we wanted to examine the forensic parameters of SE33 and especially to evaluate the data of our country's population. The lack of large-scale data on SE33 in the Turkish population and the fact that previous studies have been studied with a low number of samples encouraged us to conduct this study. Our aim with this study is to reveal the success of SE33 in forensic identification in terms of Turkey. This study is to examine whether SE33 STR locus is a distinctive marker and to continue its use in forensic identification. It is aimed to reveal allele frequency values, heterozygot genotype, homozygot genotype frequency values of this locus.

Materials and Methods

This study is a retrospective study and we analyzed a total of 500 unrelated individuals to calculate the population allele frequency. analysis completed data were obtained from Izmir Forensic Medicine Group Presidency, Department of Biology, Republic of Turkey. Data taken from applicants between January and June 2018. This study was done on the archive data of the results. Laboratory work has not been conducted using biological material.

DNA isolation and PCR

For DNA isolation, authorized staff of İzmir Forensic Medicine Group Presidency followed the manufacturers recommended protocols for EZ-1 investigator DNA isolation kit (Qiagen Hilden, Germany) and Power Plex Fusion system (Promega, Madison, WI, USA). For PCR amplification authorized staff of Izmir Forensic Medicine Group Presidency followed the manufacturers recommended protocols for PowerPlex_ Fusion system (Promega, Madison, WI, USA).

Genotyping

Amplification and fluorescence detection were performed with the PowerPlex® Fusion 6C System kit. WEN Internal Lane Standart 500 size standard (Promega, Madison, WI, USA) were used for capillary electrophoresis of Power Plex Fusion PCR Amplification kit. The electrophoresis was run on a ABI 3500xl genetic analyzer. Allele indications were named according to the recommendations of the DNA Commission of the International Society of Forensic Genetics (ISFG) [12,13]

Analysis of data

The data were analyzed using the Gene Mapper v1.4 software and Microsoft Excel (Microsoft). Genotyping results were studied twice for reliability and results were compared. The data were obtained from İzmir Forensic Medicine Group Presidency, Biology Department and this place is a member organization of German DNA profiling (GEDNAP). In this context, periodic verification studies are carried out.

Statistical analysis

Statistical analysis including, allelic and genotype frequencies, power of discrimination (PD), Matcing probabilty (MP), Polymorphism information content (PIC), Power of exclusion (PE), Typical paternity index (TPI) were performed using the Promega Power Stats excel software for 500 unrelated individuals.

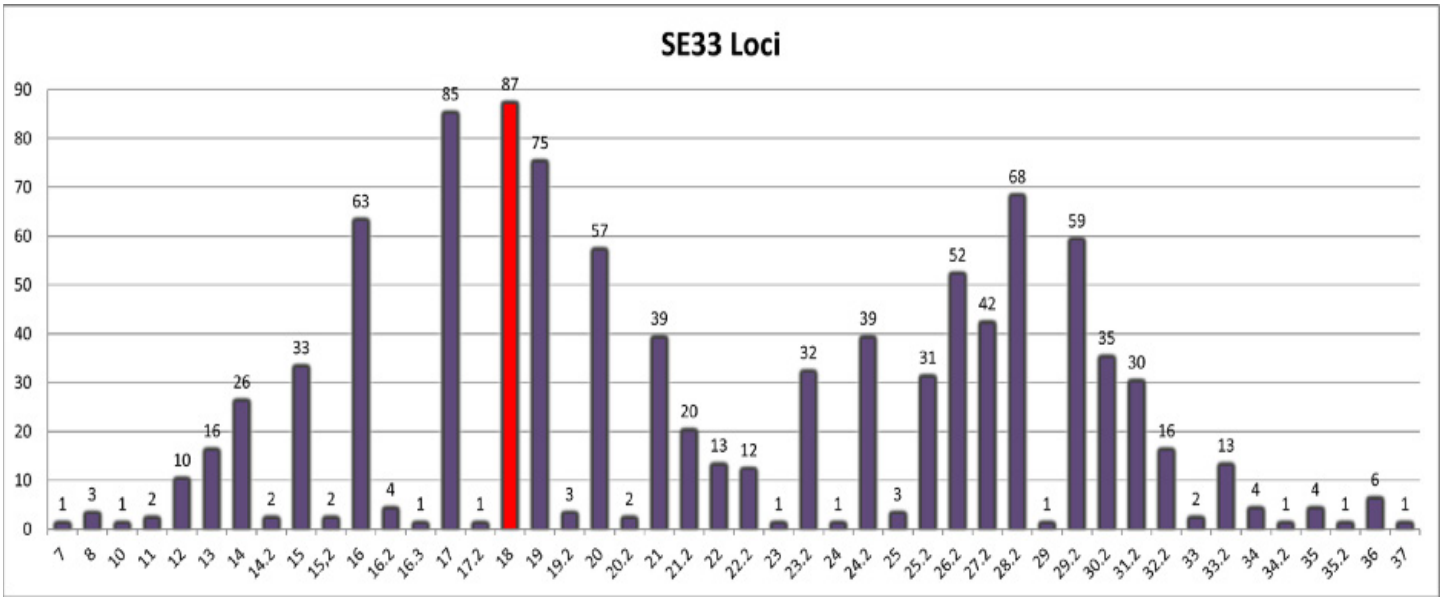

Figure 1. Distribution of detected alleles. Among a total of 1000 alleles, 18 alleles have been detected 87 times and it is the most common allele

Table 1. Distribution of number and frequency values of all homozygot genotypes

Homozygotes Genotype	n:500	%
16 - 16	2	0.4%
16.2 - 16.2	1	0.2%
17 - 17	5	1%
18 - 18	3	0.6%
19 - 19	3	0.6%
21 - 21	1	0.2%
23.2 - 23.2	1	0.2%
26.2 - 26.2	2	0.4%
27.2 - 27.2	2	0.4%
28.2 - 28.2	1	0.2%
29.2 - 29.2	3	0.6%
31.2 - 31.2	1	0.2%
33.2 - 33.2	2	0.4%
Heterozygotes Genotype	473	94.8%

Total	500	
Homozygotes	27	5.4%
Heterozygotes	473	94.6%

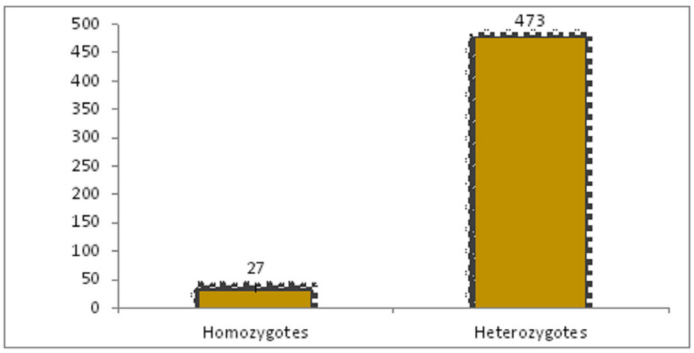

Figure 2. Table showing the number of homozygous and heterozygous 500 individuals

Table 2. Distribution of Forensic and Paternity parameters for the SE33 locus among all individuals

Forensic and Paternity Statistics	
PM (Matching Probability)	**0.008**
PD (Power of Discrimination)	**0.992**
PIC (Polymorpfic information content)	**0.95**
PE (Power of Exclusion)	**0.89**
TPE (Typical Paternity Index)	**9.26**

Results

SE33 STR locus polymorphism can be detected at different frequencies in different societies. Very high degree of heterozygosity, power of discrimination and power of exclusion in SE33 STR locus were reported in other population analyses as U.S, Germany and Portugal [6,14,15]. In order to compare the accuracy and consistency of the data obtained, a global comparison was made with data from different geographies and different ethnic groups.

In our study, 47 different alleles belonging to the SE33 locus were detected. Among all the alleles, the allel 18 was the most common with 8.7%. The least common allele was 7,10,16.3,17.2,23,24,29, 34.2,35.2,37 alleles with a frequency of 0.1% among all alleles.

A total of 209 different genotypes were seen, including 196 heterozygous and 13 homozygous genotypes. In this study we demonstrate that SE33 is one of the best STR for forensic identification purposes because forensic statistics parameters; Power of discrimination (PD):0.992, Matcing probabilty (MP):0.008, Polymorphism information content(PIC):0.95, Power of exclusion (PE):0.89, Typical paternity index (TPI):9.26.

Findings from different countries around the world

Power of discrimination of SE33 STR loci in İndia population determined as 0.990 [16]. In another study; in the Pakistani population; Forensic statistics parameters for the SE33 STR locus were Power of discrimination (PD):0.991, Power of exclusion (PE):0.769, Polymorphism information content(PIC):0.95 which Forensic statistics parameters for SE33 STR locus was determined [17].

In the study conducted in the Jordanian population; they was determined power of discrimination (PD) value as 0.983 and the rates of heterozygot observed as 0.881 by Al-Eitan et al [18].

Frequency study in the German population, the power of discrimination was found 0.987 and the observed heterozygosity was found 0.951 for SE33 [19]. In 175 Japanese individuals, forensic parameter data of the SE33 locus were reported as power of discrimination (PD): 0.9872, Polymorphic Information Content (PIC): 0.9331, Typical paternity index (TPI): 6.7308 and observed Homozygosity (H): 0.0743 [20]. In the Korean population, for 990 healthy individuals, SE33 was determined to be the most informative locus compared to other STR loci used in forensic identification [21]. In addition, forensic parameter data belonging to SE33 were determined as observed heterozygosity (Hobs): 0.9475, Probability of Matching (PM): 0.0080 and polymorphic information content (PIC): 0.8930 [21]. According to the data of 543 Bahrain individuals, SE33 was the most informative region compared to other STR loci in the study because Power Discrimination (PD) was determined as 0.9937 [22].

As stated above , our results are similar to the population studies and results obtained on SE33 around the world. Even when looking at forensic parameters such as Power of Discrimination, Matching of Probabilty, Polymorphic Information Content, Typical Paternity Index and heterozygous rates, higher values were detected.

Population data for other STR loci

In a study conducted in the European population, they was determined power of discrimination (PD): 0.977 for D1S1656 STR locus; 0.977 for D12S391 STR locus; 0.971 for D21S2055 STR locus by Iyavoo et al. [23]. Heterozygot rate values for the same loci were found to be 0,900, 0,896, 0.875 respectively [23]. In the same study in South Asia population, they was determined power of discrimination (PD): 0.972 for D1S1656 STR locus; 0.963 for D12S391 STR locus; 0.980 for D21S2055 STR locus . And than Heterozygot rate values for the same loci were found to be 0.888, 0.859, 0.901 respectively [23].

In a study conducted in the European population, they was determined power of discrimination (PD): 0.9613 for D18S51 STR loci; 0.9322 for D2S441 STR loci; 0.9349 for D91S2157 STR loci [24,25]. Considering the data of the present loci (D1S1656, D12S391, D21S2055, D18S51, D2S441, D9S2157), the values we obtained for the SE33 locus are quite sufficient for identification.

Some population studies on the SE33 STR locus in Turkey are as follows

Data for SE33 (ACTBP2) have been published from 1995 in Southern Turkey: the discrimination index (or Power of Discrimination) is 0.99 observed heterozygosity is 0.91 and Mean exclusion Chance (MEC) or Power of Exclusion (PE) is 0.89 [26]. Population data obtained from 147 individuals in Adana city of Turkey have indicated that the power of discrimination (PD):0.9773, observed heterozygosity: 0.8535 and MEC(or Power of Exclusion) 0.7991 [27]. The results of the study conducted with samples obtained from the South East region of the country; Population (n): 257, PD: 0.992 PM: 0.008 PIC: 0.95 PE: 0.841 TPE: 6.43 observed heterozygosity 92.2 [28].

Discussion

As stated before, the forensic parameters we obtained in our study; Population (n): 500 PD: 0.992 PM: 0.008 PIC: 0.95 PE: 0.89 TPE: 9.26 heterozygote is 94.6. Compared to the studies mentioned above (Alper B et al. 1995, Lászik A et al. 2001, Bozman N et al. 2018, especially studies with the Turkish population), we supported our study with a larger population number. Bozman N. et al. (2018) and our study's (PD), (PM) and (PIC) parameters have the same values. However, the typical paternity index (TPI) and heterozygosity parameters were found to be higher in our study.

In addition, the sensitivity to select unrelated individuals and the fact that the selected individuals come from cities with high immigration (Izmir, Manisa, Denizli, Aydın and Balıkesir) are the factors that make our study stronger. Therefore, the calculated forensic parameters and observed alleles are capable of reflecting the Turkish population homogeneously and widely. Obtaining the data from the accredited institution that cooperates with GEDNAP makes the results reliable.

Conclusion

In our data, it was presented that the SE33 STR locus is a powerful tool that provides the forensic parameters required for the identification of individuals. As a result of the data we have obtained, we report that the SE33 can be used safely in identification.

Conflict of interests
The authors declare that there is no conflict of interest in the study.

Financial Disclosure
The authors declare that they have received no financial support for the study.

Ethical approval
This research study was conducted retrospectively from the data obtained for forensic identification. The Health Sciences Ethics Committee of Manisa Celal Bayar University was consulted, who determined that our study did not require ethical approval.

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