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Polymorphism of SE 33 locus in a population sample from Upper Silesia (southern Poland)

Summary

In the paper the authors present the results of population research on locus SE 33 in a representative sample of 1315 unrelated individuals (males and females) of European origin living in a southern part of Poland (Upper Silesia).

Keywords locus SE 33, Upper Silesia, population genetics, statistical analysis

Introduction

Locus SE 33 (ACTBP2) is known to be the most polymorphic [2, 3] and differentiated locus of all STR markers used in forensic genetics practice [1]. Due to its high PD, SE 33 locus is among 8 other STR loci in the German DNA database [6]. However, this locus has not been involved in ESS [7] yet, which can result from its high molecular weight and chance to undergo degradation. Its advantages include big number of alleles in the ladder and a small chance for occurrence of stutters, whereas its drawback lies in long amplicons, which can present analytical problems while working with degraded DNA [8]. Locus SE 33 occurs in many kits commercially used for forensic purposes.

The aim of the research was: to study the distribution of allele frequencies for locus SE 33 in a population sample from Upper Silesia (southern Poland), show their genetic balance with Hardy-Weinberg equilibrium as well as to calculate and compare statistical parameters which could allow for assessing the usefulness of this genetic marker for forensic purposes.

Materials and methods

The population sample consisted of 1315 unrelated individuals (males and females) living in Upper Silesia (southern Poland). Total DNA was extracted from whole blood or buccal swabs using a Blood Mini or Sherlock AX set (A&A Biotechnology) as well as an EZ 1 Tissue DNA kit (Qiagen, Germany) [9] in robotic station Biorobot EZ 1 (Qiagen, Germany) [10]. Concentration of human DNA was measured using a NanoDrop ND-1000 spectrometer (ThermoFisher Scientific TK, Biotechnology, USA). PCR reactions were conducted according to the recommendations of the manufacturer of the AmpFISTR®SEfiler Plus™PCR amplification kit (Applied Biosystems, USA) [11] and Power Plex ESX 17 kit (Promega, Germany) on Gene Amp®PCR System 2700 and 8700 (Applied Biosystems, USA) [12]. PCR products were separated and detected on the 3130 AB1 Prism Genetic Analyzer (Applied Biosystems, USA). DNA growth standards were the GeneScan-500 LIZ marker (Applied Biosystems, USA) and the CC5ILS-500 marker (Promega, Germany). The obtained data were analyzed with GeneMapper®IDX v. 3.2 computer software (applied Biosystems, USA). Allelic ladders were provided by the manufacturers of the kits. Genotypes were determined for all individuals. The χ^2 and exact tests as implemented in TEPGA computer software were used to test for significant deviations from Hardy-Weinberg equilibrium. FatRec software was applied for calculation of the forensically relevant statistical parameters (PD, Ht_{obs}, Ht_{exp}, PM, MEC, PIC and MEP). Interpopulation comparisons were made by the locus-by-locus AMOVA test implemented in the Arlequin v. 3.01 package [15]. P and F_{ST} values were calculated on the basis of 10 1000 allele permutations. Diagrams were done using StatSoftware STATISTICA 10.The examined population sample was compared with a population sample from northern Poland [16, 17] and a population sample from the Podlasie area [18].

Results and discusion

The distribution of allele frequencies of locus Se 33 for the examined population from Upper Silesia (n = 1315) as well as populations from northern Poland (n = 1007, 2012 and n = 255, 2008) [16, 17] and the Podlasie area is presented in Table 1. 47 alleles were found in the examined population sample and respectively 46 and 37 alleles in a population of northern Poland [16, 17] and also 35 alleles in a population sample from the Podlasie area. The most frequent alleles in the examined population sample were respectively: 28.2 (f = 0, 0867), 27.2 (f = 0.0757), 17 (f = 0,0696), 29.2 (f = 0,0665) and 19 (f = 0,0654). Alleles 7.3 (f = 0,0004), 8 (f = 0,0004), 9 (f = 0,0008), 16,3 (f = 0,0004), 35 (f = 0,0011), 37 (f = 0,0011) and 41 (f = 0,0004) rarely occurred in the examined individuals. Some of these alleles have not been described in literature yet. Alleles: 7.3, 12.2, 13.2, 14.2, 15.2, 16.3, 17.3, 19.2, 23, 28, 31, 32,33,34 and 41 which are no in allelic ladders for a Power Plex ESX 17 and AmpFISTR® SEfilerPlus™PCR amplification kit were determined on the basis of precise measurements of the size of DNA fragments without their sequencing. PowerPlex ESX17 kit was used for typing. In case of out ladder alleles and allele 22 (only in the ladder for a PowerPlexESX17 kit) and allele 35.2 (only in the ladder for an AmpFISTR® SEfilerPlus™PCR amplification kit), the obtained results were verified using a SEfiler kit (Applied Biosystems, USA). The performed χ^2 and exact tests did not indicate deviation from Hardy-Weinberg equilibrium (χ^2 = 834,51003, df = 108, p = 1) in the examined population sample. High values of statistical parameters such as PIC (0.9451), Ht_{obs} (0.948)0, PD (0.9947), PE (0.8941), MEC (0.8939) and low values of PM (0.0053) confirmed the usefulness of locus SE 33 for forensic purposes, especially in genetic identification examinations of biological evidence including mixtures as well as in consanguinity examinations.

Values of F_{st} and P for separate alleles of locus SE 33 in the examined population sample and 4 other population samples [16, 17, 18] are presented in Table 2. Comparison of these values for the above mentioned population samples [16, 17, 18] is shown in Table 3. There was no deviation between the

Table 1

Allele frequencies and basic statistical parameters for locus SE33 in four population samples from different part of Poland

Allele	Upper Silesia		Northern Poland		Northern Poland		Podlasie	
N		1315		1007		255		220
4.2	–	–	–	–	–	–	–	–
6.3	–	–	–	–	–	–	–	–
7.3	0.0004	1	–	–	–	–	–	–
8	0.0004	1	–	–	–	–	–	–
9	0.0008	2	–	–	–	–	–	–
10	–	–	–	–	–	–	–	–
11	–	–	–	–	–	–	0.0023	1
11.2	–	–	–	–	0.0020	1	–	–
12	0.0023	6	0.0036	6	0.0059	3	0.0136	6
12.2	0.0011	3	0.0005	1	–	–	–	–
13	0.0095	25	0.0070	14	0.0098	5	0.0023	1
13.2	0.0008	2	0.0015	3	0.0020	1	0.0023	1
14	0.0475	125	0.0318	64	0.0235	12	0.0273	12
14.2	0.0027	7	0.0070	14	–	–	–	–
15	0.0365	96	0.0422	85	0.0510	26	0.0500	22
15.2	0.0008	2	0.0010	2	–	–	–	–
15.3	–	–	0.0005	1	0.0020	1	–	–
16	0.0487	128	0.0501	101	0.0431	22	0.0477	21
16.2	–	–	0.0010	2	–	–	–	–
16.3	0.0004	1	–	–	–	–	–	–
17	0.0696	183	0.0775	156	0.0529	27	0.0705	31
17.3	0.0004	1	–	–	–	–	0.0023	1
18	0.0646	170	0.0546	110	0.0667	34	0.0432	19

18.3	–	–	–	–	–	–	0.0023	1
19	0.0654	172	0.0650	131	0.0804	41	0.0818	36
19.2	0.0057	15	0.0050	10	0.0059	3	0.0091	4
20	0.0529	139	0.0546	110	0.0588	30	0.0432	19
20.2	0.0057	15	0.0119	24	0.0020	1	0.0091	4
21	0.0205	54	0.0223	45	0.0294	15	0.0341	15
21.1	–	–	–	–	0.0020	1	–	–
21.2	0.0186	49	0.0209	42	0.0137	7	0.0182	8
22	0.0049	13	0.0094	19	0.0039	2	0.0068	3
22.2	0.0338	89	0.0283	57	0.0275	14	0.0341	15
23	0.0019	5	0.0015	3	0.0020	1	–	–
23.2	0.0373	98	0.0397	80	0.0412	21	0.0227	10
24	–	–	0.0030	6	–	–	0.0023	1
24.2	0.0395	104	0.0482	97	0.0373	19	0.0341	15
25	–	–	0.0005	1	–	–	–	–
25.2	0.0437	115	0.0407	82	0.0510	26	0.0432	19
26	–	–	0.0015	3	–	–	–	–
26.2	0.0521	137	0.0586	118	0.0608	31	0.0432	19
27	–	–	0.0005	1	–	–	–	–
27.2	0.0757	199	0.0765	154	0.0667	34	0	0
28	0.0004	1	0.0015	3	–	–	0.0841	37
28.2	0.0867	228	0.0755	152	0.0922	47	0.0841	37
29	–	–	0.0015	3	–	–	–	–
29.2	0.0665	175	0.0611	123	0.0745	38	0.0523	23
30	–	–	0.0005	1	–	–	0.0023	1
30.2	0.0487	128	0.0427	86	0.0373	19	0.0636	28
30.3	–	–	0.0005	1	–	–	–	–
31	0.0004	1	0.0015	3	0.0020	1	–	–
31.2	0.0179	47	0.0248	50	0.0196	10	0.0273	12
32	0.0011	3	–	–	0.0020	1	–	–
32.2	0.0137	36	0.0114	23	0.0118	6	0.0227	10
33	0.0027	7	0.0020	4	–	–	0.0091	4
33.2	0.0068	18	0.0045	9	0.0118	6	0.0045	2
34	0.0027	7	0.0035	7	0.0020	1	0.0023	1
34.2	0.0027	7	0.0015	3	0.0020	1	–	–
35	0.0011	3	–	–	–	–	–	–
35.2	0.0027	7	0.0020	4	0.0020	1	–	–
36	0.0004	1	–	–	0.0020	1	0.0023	1
37	0.0011	3	–	–	–	–	–	–
39	–	–	–	–	–	–	–	–
41	0.0004	1	–	–	–	–	–	–
42	–	–	–	–	–	–	–	–

Ht	0.9480	0.9380	0.9410	0.9490
PD	0.9947	0.9940	0.9910	0.9900
PM	0.0053	0.0060	0.0090	0.0100
MEC	0.8939	–	–	–
MEP	0.8941	–	–	–
PIC	0.9451	0.9500	0.9400	0.9500

Table 2

Values of FST (below diagonal) and P (above diagonal) for allele frequencies (AMOVA test). P > 0,05 in bold

	Katowice (N = 1315)	Gdańsk 2008 (N = 255)	Gdańsk 2012 (N = 1007)	Białystok (N = 220)
Katowice	–	0.53965	0.26473	0.00000
Gdańsk 2008	0.00009	–	0.39917	0.00000
Gdańsk 2012	0.00008	0.00005	–	0.00000
Białystok	0.00684	0.00590	0.00662	–

examined population sample, a population sample of northern Poland [16, 17] and a population of northern Poland with n = 1262. However, a statistically significant deviation was found between the examined population sample and a population sample from the Podlasie area [18].

In already published papers on polymorphism of locus SE 33 [16], statistically significant deviations between a population sample from northern Poland [16, 17] and a population sample from the Podlasie area [18] were described. It is known that the population of Poland shows a high homogeneity for most autosomal STR loci. Local data bases concerning allele frequencies can be useful for statistical analysis. The fact that allele 28 is most frequent (f = 0.0841) in a population sample of the Podlasie area seems to be astonishing. In the examined population samples from other parts of Poland this allele was rare or did not occur at all (for Fig. 1 Two-dimensional diagram of relative distances between population samples for allele frequencies of locus SE33 [multi-dimensional scaling on the basis of the values calculated using an AMOVA test] and Fig. 2 Dimensional diagram of relative distaces between population samples for allele frequencies of locus SE33 [multi-dimensional scaling on the basis of the values calculated using an AMOVA test]; see Polish version).

Table 3

Values of FST (below diagonal) and P (above diagonal) for allele frequencies (AMOVA test). P > 0,05 in bold

	Katowice (N = 1315)	Gdańsk (N = 1262)	Białystok (N = 220)
Katowice	–	0.33947	0.00000
Gdańsk	0.00004	–	0.00000
Białystok	0.00684	0.00647	–

Conclusions

The examined population sample from Upper Silesia (southern Poland) is found to be in agreement with Hardy-Weinberg equilibrium.

Statistical parameters for locus SE 33 in the examined population show that this marker can be useful for forensic purposes.

Except Podlasie, the population of Poland shows homogeneity with reference to alleles for locus SE 33.

In case of population studies on multi-allelic loci, the size of the examined group should be big.

Source

Figs. 1–2: authors
Tabs. 1–3: own elaboration

Translation *Kornelia Drożdziok, Jadwiga Kabiesz, Marcin Tomsia, Krzysztof Rębała*