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Population genetic study of the three minisatellites loci: D7S21, D12S11 and D5S110 in Poland

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Abstract. The VNTR loci: D7S21, D12S11 and D5S110 are the highly polymorphic markers of the human genome. The efficiency of forensic evaluation for these loci in the population of Poland was compared with similar data for other world populations. The combined values of PD and PE for the three-locus profile in the investigated population were calculated to be at 99.99997% and 99.996% respectively. The values of forensic efficiency indicators received for D7S21, D12S1 and D5S110 minisatellite markers in the population of Poland confirm their extreme polymorphism and high suitability for research as shown in investigation of all other populations. This adjudicates that RFLP markers such as these investigated are invaluable help in resolving paternity determination cases, where the DNA profile of the defendant is not available as well as in analyses of the relationship between any given people.

Keywords: D7S21; D12S11; D5S110; Poland; Forensic evaluation; Interpopulation comparison

1. Introduction

Loci: D7S21, D12S11, D5S110 with repeat unit between 20 and 53 base pair and chromosomal localisation: 7p22pter, 12q24.3 and distal 5p respectively were discovered by Wong et al. [1] and by Armour et al. [2]. These markers belong to hypervariable regions, so-called HVR, which are presented in human DNA in a large number of allelic forms [3]. Our practise indicates that they are an invaluable help in resolving the most

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difficult cases of kinship analysis [4]. A comprehensive database of these regions has not been set up for the Polish population. In the following work efficiency of forensic evaluation for these three VNTR markers for the population of Poland was compared against similar data for other world populations.

2. Materials and methods

The material used for this research was blood samples of 300 non-related people directly involved in the disputed paternity cases. DNA was restricted with Hinf I enzyme, distributed in 0.7% agarose in TBE buffer, vacuum transfered onto a Hybond N+ membrane, hybridized with alkaline phosphatase conjugating single locus probes: MS31 (locus D7S21), MS43A (locus D12S11), MS621 (locus D5S110) and MW100 (Ladder) [5], detected with the use of Lumi Phos 530 substrate and Hyperfilm™ ECL and analysed with software BIO1D in comparison to the NICE™ DNA Analysis Ladder. For the measured DNA fragments binned into steps of 4.6% fragment size (the maximum testing error for the laboratory) the following parameters characterising usefulness and proof value of the investigated markers were calculated: heterozygosity—*H*, discrimination index—DI, power of discrimination—PD, power of exclusion—PE, paternity index—PI, polymorphism information content—PI and mutation rate.

3. Results

The parameters elucidating the polymorphism and usefulness for paternity testing of the investigated loci: D7S21, D12S11 and D5S110 in Poland are shown in the Table 1. The combined values of PD and PE for the three-locus profile in the investigated population were calculated to be at 99.99997% and 99.996% respectively. The high values of the observed heterozygosity 0.96 for each locus were adjacent to the analogical values in other populations, as is shown in Table 2. Also, the accessible data of the discrimination index (DI) value are displayed in the Table 2. The mutation rates was 0.12% (D12S21), 0.57% and 0.78% (D5S110) (data not presented).

4. Discussion

The analysis of forensic efficiency parameters of the minisatellite markers: D7S21, D12S1, D5S110 in Poland proved them to be very useful in forensic practise, as it was indicated earlier for other world populations. The cumulated value for the three locus profile in Poland equals 3.6×10^{-8}, i.e. the certain three-locus profile will appear on average once for every 28 million in the population. Previously, the uniqueness of the

Table 1
The forensic efficiency parameters: $H_{exp,obs}$—expected, observed heterozygosity, PIC—polymorphism information content, PD—power of discrimination, $PE_{exp,obs}$—expected, observed power of exclusion and PI_{av}—average paternity index of the investigated loci in a sample population of Poland

Locus	$H_{exp.}$	H_{obs}	PIC	PD	PI_{av}	PE_{exp}	PE_{obs}
D7S21	0.9398	0.9567	0.9348	0.9927	11.26	0.8748	0.9737
D12S11	0.9370	0.9567	0.9317	0.9921	11.27	0.8692	0.9615
D5S110	0.9465	0.9633	0.9422	0.9942	14.26	0.8888	0.9643
Total				0.9999997	1810.26	0.9982	0.99996

Table 2
The comparison of coefficients of polymorphism: heterozygosity—*H* and discrimination index—DI in different populations

Population/locus	D7S21 *H* (DI)	D12S11 *H* (DI)	D5S110 *H* (DI)
Poland	0.96 (3.7×10^{-3})	0.96 (3.7×10^{-3})	0.96 (2.6×10^{-3})
Germany	0.95 (4.1×10^{-3})	0.96 (3.6×10^{-3})	–
Austria	0.93 (1.0×10^{-2})	0.90 (1.8×10^{-2})	–
England	0.97 (1.8×10^{-3})	0.96 (3.1×10^{-3})	–
Italy	–	0.96	–
Spain	0.93	0.91	–
Turkey	0.95	0.86	–
Denmark	0.94	0.91	–
Finland	0.95	–	–
USA	–	–	0.97
Mexico	–	0.98 (2.0×10^{-4})	–
New Zealand	–	– (1.1×10^{-2})	– (5.0×10^{-3})
Asia	0.94 (9.0×10^{-3})	0.87 (8.0×10^{-3})	–
Afro-Caribbean	0.95 (4.0×10^{-3})	0.95 (3.0×10^{-3})	–
Average H (DI)	0.95 (3.8×10^{-3})	0.93 (5.7×10^{-3})	0.96 (5.1×10^{-3})

three-locus SLP profile in different populations was proven by Gill et al. [6]. In practice, both Lambert et al. [7] in Great Britain and Sharma et al. [8] in the USA did not observe any occurrence of an identical three-locus profile while analysing 16 and 69 thousands ones respectively. The research at our laboratory has proved Henke and Fimmers' [9] observations that two matches between the child and the alleged father in hypervariable minisatellite regions such as D7S21, D12S11 and D5S110 establish kinship between the subjects. This adjudicates that RFLP markers such as these investigated are invaluable help in resolving paternity determination cases, where the DNA profile of the defendant is not available as well as in analyses of the relationship between any given people.

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