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Polymorphism at three STR loci on chromosome 21 (D21S1411, D21S1414, and D21S1435) in Croatia

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Abstract. In order to increase the number of STR loci for forensic DNA analysis as well as for prenatal diagnosis of Down syndrom in the Croatian population, we have studied three tetranucleotide STR loci on chromosome 21 (D21S1411, D21S1414, and D21S1435). One hundred and fifty-six unrelated healthy individuals were tested in this study. The results clearly demonstrated that the most polymorphic locus (20 different alleles) was D21S1414 with PIC value of 0.9279, while at D21S1435 locus seven different alleles were observed with PIC value of 0.7706. The third locus, D21S1411, also showed great polymorphism with 15 distinct alleles, PIC value 0.8920.

Keywords: STR; Prenatal diagnostics; Down syndrome

1. Introduction

Short tandem repeat loci have been used as routine genetic markers for population genetics, forensics and paternity testing and for following chimerism after bone marrow transplantation. Recently, many laboratories are developing protocols for prenatal diagnosis of trisomies such are Down's (chromosome 21), Edward's (chromosome 18), or Pataouv's syndrome (chromosome 13) [1]. Namely, classical cytogenetic methods consume 3–4 weeks to obtain results, while application of STR loci in prenatal diagnosis would be helpful because the results can be available after 3–4 days [2]. However, in order to be useful in prenatal diagnosis, any of the STR loci have to display a significant degree of polymorphism within population. For that reason, we examined the allele distribution of three STR loci (D21S1411, D21S1414, and D21S1435) on chromosome 21 in our population.

2. Materials and methods

Blood samples were collected from 156 unrelated healthy individuals living in Zagreb. DNA was isolated using NucleoSpin kit (Macherey-Nagel, Duren, Germany);

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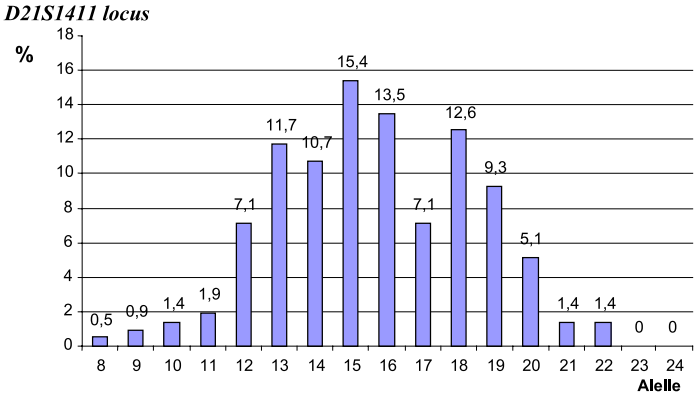
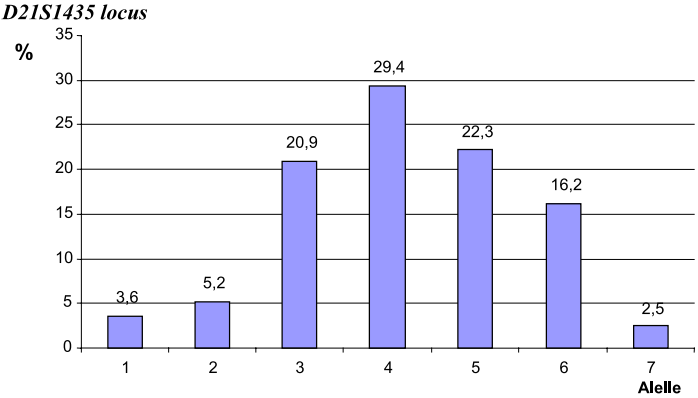
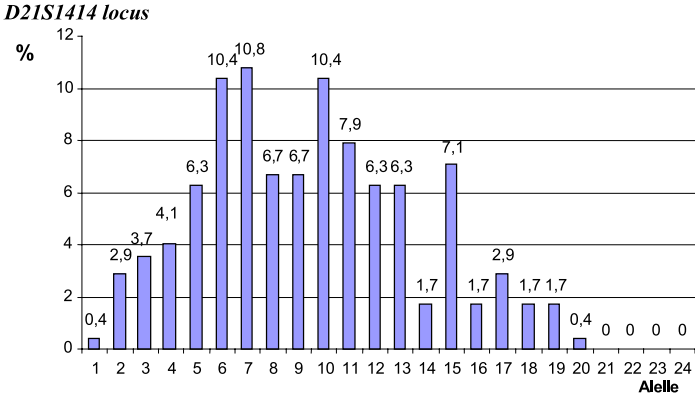


*Legende: Alelles **8**-208bp; **9**-212bp; **10**-216bp; **11**-220bp; **12**-224bp; **13**-228bp; **14**-232bp; **15**-236bp; **16**-240bp; **17**-244bp; **18**-248bp; **19**-252bp; **20**-256bp; **21**-260bp; **22**-264bp; **23**-268bp; **24**-272bp.*



*Legende: Alelles **1**-163bp; **2**-167bp; **3**-171bp; **4**-175bp; **5**-179bp; **6**-183bp; **7**-187bp.*



*Legende: Alelle**1**-325bp; **2**-327bp; **3**-329bp; **4**-331; **5**-333; **6**-335; **7**-337; **8**-339; **9**-341; **10**-343; **11**-345; **12**-347; **13**-349; **14**-351; **15**-353; **16**-355; **17**-357; **18**-359; **19**-361; **20**-363; **21**-365; **22**-367; **23**-369; **24**-371.*

Fig. 1. Distribution of alleles at D21S1411, D21S1435 and D21S1414 loci in the Croatian population ($N = 156$).

Table 1
Statistical parameters for D21S1411, D21S1435 and D21S1414 loci

LOCUS	HET_{obs}	HET_{exp}	PIC
D21S1411	0.850467	0.992463	0.891951
D21S1435	0.758242	0.964932	0.770623
D21S1414	0.766666	0.928681	0.927856

HET_{obs}—observed heterozygosity; HET_{exp}—expected heterozygosity; PIC—polymorphism information content.

after isolation DNA samples were amplified in three separate PCR reactions [3]. Electrophoresis was run on 6% polyacrylamide gel in automated laser fluorescence sequencer (ALFexpress, Pharmacia Biotech, Uppsala, Sweden). Allele size determination was performed using Allele Locator program and for standard we used two internal size markers and one commercial. Statistical analysis included the estimation of allele and genotype frequencies, observed (OH) and expected (EH) heterozygosity and polymorphic information content (PIC). The Hardy–Wienberg equilibrium was verified using the χ^2-test.

3. Results and discussion

In this study we present data concerning three STR loci on chromosome 21. The observed allele frequencies are presented in Fig. 1. Alleles at D21S1414 locus have a simple dinucleotide repeat structure, while alleles at the D21S1411 and D21S1435 loci consisted of tetranucleotide repeat units. At D21S1411 and D21S1435 loci we did not observe intermediate alleles. Some statistical parameters are summarised in Table 1. No deviation from Hardy–Weinberg equilibrium was observed. The highest PIC value was observed for D21S1414 locus (0.9278). Our results show that the observed heterozygosity of three STR loci is higher than 75%. It means that these three STR loci have a sufficiently high level of informativeness in the Croatian population and, for that reason, can be applied as genetic markers in prenatal diagnostics as well as in forensics.

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