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Allele distribution at two STR loci (D15S642 and D15S659) in the Croatian population

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Abstract. Population studies of two short tandem repeats (STR) loci (D15S642 and D15S659) were carried out in a sample of 130 unrelated healthy individuals. After PCR amplification, samples were run on 6% polyacrylamide gel in automated sequencer (ALFexpress). Twelve different alleles were identified at D15S642 locus and 11 alleles at D15S659 locus. The most frequent alleles at D15S642 were allele 2 (16.7%), allele 8 (16.3%) and allele 9 (14.0%), while the most frequent genotype was 2-2. Among 11 different alleles at D15659, the most frequent were allele 9 (22.1%), allele 3 (19.1%) and allele 8 (18.4%). Genotype 9-8 showed the highest frequency (9.6%) at D15S659 locus. The observed heterozygosities for these two loci were 0.81818 for D15S642 and 0.83088 for D15S659. The PIC was calculated as follows: 0.88 for D15S642 and 0.83 for D15S659. No significant deviations from Hardy–Weinberg equilibrium could be observed for these systems. The results indicate that these two loci are useful genetic markers for paternity testing as well as for prenatal or postnatal diagnosis. © 2005 Elsevier B.V. All rights reserved.

Keywords: STR; Chromosome 15; Croatian population

1. Introduction

Short tandem repeats (STR) markers are widely applied for forensic identification, population genetic studies, paternity testing and for following chimerism after bone marrow transplantation. STR markers are also used in prenatal and postnatal diagnosis of chromosomal aneuploidies and uniparental disomy (UPD). Namely, molecular genetic analysis of STRs located on chromosome 15 from the proband and both parents is used in most laboratories to trace the transmission of chromosome 15 from each parent to the

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child. This is performed in order to determine whether the child demonstrates normal biparental inheritance or has only maternal (Prader-Willi syndrome) or paternal (Angelman syndrome) markers [1].

To be useful in diagnostic testing for UPD of chromosome 15, any of the STR loci have to display a significant degree of polymorphism within population. For that reason, we examined the allele distribution of two STR loci (D15S642 and D15S659) on chromosome 15 in our population.

2. Materials and methods

Blood samples were obtained from 130 unrelated healthy individuals living in Zagreb. DNA was isolated using NucleoSpin blood kit (Machery-Nagel, Duren, Germany), and amplified in two separate PCR reactions [2]. The amplified products were separated on a 6% polyacrylamide gel using the automated laser fluorescence sequencer (ALFexpress, Pharmacia Biotech, Uppsala, Sweden). The size of alleles was determined using Allele Locator program. Allele and genotype frequencies for each STR locus were determined by direct counting. Hardy–Weinberg equilibrium (HWE) was tested using the Chi-squared testing method. The polymorphism information content (PIC), observed (HET_{obs}) and expected (HET_{exp}) heterozygosity were calculated as proposed by Gourraud [3].

3. Results and discussion

Typing for D15S642 and D15S659 loci revealed 12 and 11 different alleles, respectively. Fragment lengths ranged from 192 bp to 222 bp for D15S642 and 168 bp to 208 bp for D15S659. The allele frequencies observed as well as statistic parameters for these two STR loci are shown in Tables 1 and 2.

Table 1
Allele frequencies ($n = 130$) and statistic parameters for locus D15S642

Allele	Frequency
1	0.015
2	0.167
3	0.095
4	0.072
5	0.045
6	0.057
7	0.110
8	0.163
9	0.140
10	0.800
11	0.027
12	0.030
HET_{obs}	0.81818
HET_{exp}	0.87603
PIC	0.88371

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

Table 2
Allele frequencies ($n = 130$) and statistic parameters for locus D15S659

Allele	Frequency
1	0.004
2	0.026
3	0.191
4	0.158
5	0.001
6	0.059
7	0.015
8	0.184
9	0.221
10	0.110
11	0.026
HET_{obs}	0.83088
HET_{exp}	0.82265
PIC	0.83239

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

Table 3
The most frequent genotypes for loci D15S642 and D15S659

Locus	Genotype	Frequency
D15S642	2-2	0.068
	2-9	0.053
	3-8	0.053
	7-8	0.045
	8-8	0.038
D15S659	8-9	0.095
	4-9	0.088
	3-9	0.081
	3-4	0.081
	3-8	0.066

The observed heterozygosities, which ranged from 0.81818 (D15S642) to 0.83088 (D15S659) are close to those expected. The five most frequent genotypes for each locus are shown in Table 3. No significant deviation from Hardy–Weinberg equilibrium was detected. The results indicate that these two loci are useful genetic markers in prenatal and postnatal diagnosis of Prader-Willi/Angelman syndrome as well as for personal identification and paternity testing in the Croatian population.

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