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A SNP-STR locus within the HLA class II region: Sequence and population data of D6S2822

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Abstract. Population genetics and sequence structure of the tetranucleotide short tandem repeat locus D6S2822 were investigated in an Austrian Causasoid population sample in order to reveal information about its usefulness for HLA haplotyping, linkage analyses and forensic testing. Seven different sized alleles additionally showing sequence polymorphism could be detected. As for allele distribution and statistic parameters D6S2822 turned out to be a moderately repetitive and informative marker. Due to the A/G SNP adjacent to the repeat region this locus might also be of interest for phylogenetic studies.

Keywords: D6S2822; STR; Sequence; Population data; HLA

1. Introduction

Polymorphic STR markers within the HLA region can be used for a better characterization of HLA haplotypes, determination of disease associations, recombination point mapping or even for forensic testing if no linkage disequilibrium exists between STR alleles and other markers tested.

In this study, population genetics and sequence structure of the tetranucleotide repeat locus D6S2822 (M2_4_25; GATA129G03; GenBank G10435; UniSTS 239167, 464402, 464403) [1,2] situated nearby the HLA class II region (6p21.3) were investigated in an Austrian Causasoid population sample of 153 unrelated individuals in order to reveal its characteristics.

2. Materials and methods

PCR amplification was performed on a GeneAmp 9700 PCR System (Applied Biosystems, Foster City, USA) in 15 μl volume using described primers [2] (forward

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Table 1
Sequence structure of D6S2822 SNP-STR polymorphism

Allele designation	Sequence structure		Length (bp)	No. of sequenced alleles
	Position 64–67 5′-FR	Repeat region		
10	GGGG	$(TATC)_9$ (CATC)	189	1
11	GGGG	$(TATC)_{10}$ (CATC)	193	2
11	GAGG	$(TATC)_{10}$ (CATC)	193	2
12	GGGG	$(TATC)_{11}$ (CATC)	197	2
12	GAGG	$(TATC)_{11}$ (CATC)	197	9
13	GGGG	$(TATC)_{12}$ (CATC)	201	2
13	GAGG	$(TATC)_{11}$ $(CATC)_2$	201	4
14	GGGG	$(TATC)_{13}$ (CATC)	205	1
14	GAGG	$(TATC)_{13}$ (CATC)	205	3
14	GAGG	$(TATC)_{12}$ $(CATC)_2$	205	3
15	GAGG	$(TATC)_{14}$ (CATC)	209	1
15	GAGG	$(TATC)_{13}$ $(CATC)_2$	209	3
16	GAGG	$(TATC)_{14}$ $(CATC)_2$	213	1

primer 6-FAM labelled) in a modified protocol (95 °C 11 min for 1 cycle; 94 °C 20 s, 61 °C 1 min, 72 °C 1 min for 28 cycles; 60 °C 45 min for 1 cycle). Typing of the amplification products in comparison with a locus-specific allelic ladder containing the most common alleles as well as cycle sequencing of selected alleles applying BigDye chemistry according to the manufacturer's protocol (Applied Biosystems, Foster City, USA) were carried out using denaturing capillary electrophoresis on an ABI Prism® 310 Genetic Analyzer.

3. Results and discussion

Sequencing a total of 34 alleles from the population study and further samples, which were not included into frequency data, revealed 7 different sized alleles ranging from 189 to 213 bp and showing a $(TATC)_{9–14}$ $(CATC)_{1–2}$ repeat pattern. No incomplete repeats were found (Table 1).

Table 2
Allele frequencies and further statistic parameters of D6S2822

Allele designation[a]	Allele frequency
11	0.026
12	0.261
13	0.510
14	0.183
15	0.017
16	0.003
Rate of heterozygosity	0.595
Power of exclusion	0.285
Polymorphism information content	0.580
Power of discrimination	0.816
Typical paternity index	1.230

[a] The rare allele 10 was only found once in the additional samples and thus not included into frequency data.

Additionally, 17 bp upstream from the repeat region (position 65 of the allele, situated in the 5′-flanking region), an A/G SNP with the major allele A and the minor allele G (76.5% and 23.5% of all sequenced alleles, respectively), was observed (Table 1).

The resulting allele frequencies as well as further statistic data are shown in Table 2. No deviation from Hardy-Weinberg equilibrium could be detected ($0.4 < p < 0.5$). Furthermore no mutations were observed in 263 meioses within 71 Austrian Caucasoid families.

4. Conclusion

The tetranucleotide repeat locus D6S2822 showed an interesting sequence structure and an average allele distribution. Apart from its applications concerning the HLA system, due to its SNP nearby the repeat region, it might be a candidate for phylogenetic investigations.

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References

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