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D1S1171: a new highly variable short tandem repeat polymorphism located on chromosome 1

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Abstract

This study reports the evaluation of the short tandem repeat (STR) locus D1S1171 (GDB: 312934) for forensic purposes, which was investigated by PCR amplification and native polyacrylamide gel electrophoresis in 141 unrelated Austrians. No deviations from Hardy–Weinberg expectations were observed. The mean exclusion chance (MEC) was 0.677, the discriminating power (DP) was 0.951 and the observed heterozygosity rate was 0.853. An allelic ladder consisting of 10 sequenced alleles (96–132 bp) was constructed. Sequence analysis revealed a GAAA repeat motif. According to the number of tetranucleotide repeats, the smallest allele was designated 9 and the largest allele 18.

Keywords: STR; PCR; Sequencing; D1S1171

1. Introduction

Individual differences are based on repetitive DNA sequences in the human genome. In case of short tandem repeat loci (STRs) these sequences consist of di- to pentameric repeats [1,2] with fragment lengths usually smaller than 300 bp. These properties make STRs highly suitable for forensic purposes such as stain analysis and paternity testing. As these loci are abundant in the human genome [3] and only a small number has been evaluated up to now, the main interest should focus on the evaluation of new, more efficient STRs. The aim of this study was to determine the forensic parameters of the

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Table 1
Tabular summary of the alleles (9–18) found at the D1S1171 locus

Alleles	Sequence	Length (bp)	Frequency (*f*)
9	5′ -CAAAAAA-$(GAAA)_9$-3′	96	0.04
10	5′ -CAAAAAA-$(GAAA)_{10}$-3′	100	0.07
11	5′ -CAAAAAA-$(GAAA)_{11}$-3′	104	0.17
12	5′ -CAAAAAA-$(GAAA)_{12}$-3′	108	0.23
13	5′ -CAAAAAA-$(GAAA)_{13}$-3′	112	0.38
14	5′ -CAAAAAA-$(GAAA)_{14}$-3′	116	0.52
15	5′ -CAAAAAA-$(GAAA)_{15}$-3′	120	0.36
16	5′ -CAAAAAA-$(GAAA)_{16}$-3′	124	0.14
17	5′ -CAAAAAA-$(GAAA)_{17}$-3′	128	0.06
18	5′ -CAAAAAA-$(GAAA)_{18}$-3′	132	0.03

The repeat region sequence structure of each allele is shown in the middle, the fragment length and frequency are given on the right.

tetrameric repeat locus D1S1171 (GDB ID 312934) which have not been investigated so far.

2. Materials and methods

DNA was extracted from blood samples of 141 unrelated Austrians as described [4]. Primers were selected from the GDB entry ID GDB 312934. Amplification and typing was performed according to Reichenpfader et al. [5]. Sequencing was done on the capillary electrophoresis system ABI Prism 310 Genetic Analyzer. Statistical analysis was performed as described [6].

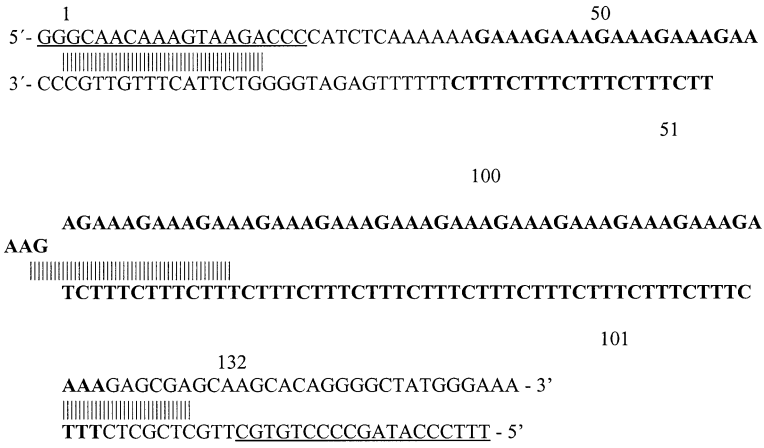

Fig. 1. Consensus sequence of allele 18 at the D1S1171 locus. Repeat region in bold print, primer sequences underlined.

3. Results

Table 1 summaries the characteristic features of D1S1171 with its hypervariable region consisting of a varying number of tetranucleotide GAAA repeats which were therefore used for allelic designation from 9 to 18. With the 10 sequenced alleles with fragment lengths from 96 to 132 bp, an allelic ladder was constructed. The consensus sequence of D1S1171 is shown in Fig. 1. A total of 34 genotypes was found in the 141 Austrian probands tested. No significant deviations from Hardy–Weinberg expectations were observed. The heterozygosity rate was 0.853, the mean exclusion chance (MEC) was amounted to 0.677 and the discriminating power (DP) was 0.951.

4. Discussion

The fragment length of D1S1171 distinctly below 150 bp should render this locus as suitable for typing severely degraded stains. The polymorphism of the locus proposed in this study is higher than that of other STRs commonly used in forensic practice [2,7]. From the uninterrupted repeat length of this locus, it can be concluded that the mutation rate should be comparable to that of other STRs [8]. Since typing the D1S1171 locus proved to be easy and reliable, we suggest this locus as a useful tool for both paternity testing and stain analysis.

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