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New sequence data of allelic variants at the STR loci ACTBP2 (SE33), D21S11, FGA, vWA, CSF1PO, D2S1338, D16S539, D18S51 and D19S433 in Caucasoids

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Abstract. On the occasion of a study on mutation rates of human STR loci, numerous alleles have been sequenced. Several new sequence or length variant alleles were found.

Keywords: Sequence data; Sequence variation; Sequencing; Variant alleles

1. Introduction

This study reports sequence data of rare and new sequence or length variant alleles which have been found in course of a study on mutation rates of human STR loci. These variant alleles are briefly discussed, especially with regard to further investigations.

2. Materials and methods

DNA was extracted from blood or buccal swab samples of Caucasoids living in Austria and Switzerland and from one sample which was derived from a black individual. The same primers were used for PCR and sequencing, which was carried out in both directions (GenBank accession nos. M84567, M64982, G07925, G08036 and G08202).

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SE33 alleles were designated according to the ACTBP2 nomenclature recommendations of GEDNAP [1].

3. Results and discussion

While the SE33 alleles 7.3, 12.1, 13, 19.2, 20, 25.2, 26.2, 27.2, 29.2, 30.2, 31.2, 33, 33.2 and 34.2 represent the same type of variation already known for this locus [2,3], the alleles 11.2, 12.2, 13.2, 16.2, 16.3 and 18.3 represent a new type of allelic sequence structure lacking the $(AAAG)_3$ AG structure in the 5′ flanking region (Table 1). Additionally, the alleles 16.3 and 18.3 exhibited a doubled G-$(AAAG)_3$ structure in the 3′ flanking region, which is unique so far.

The sequence structure of three variant D21S11 alleles 29, 29.2 and 30 found in this study (Table 2) has not been reported so far, the other variant alleles have already been observed in Asian populations [4], but not in Caucasoid individuals until now. Further sequence or length variant alleles at the FGA and the vWA locus are given in Table 2, including another rare variant vWA allele 15 [5] and a new variant vWA allele 16; it exhibited a C → T transition at position 3 in the 3′ flanking region leading to 13 instead of 11 consecutive TCTA repeats. This type of variation has only been reported for black populations so far in alleles 12–14 [6].

At the CSF1PO locus a rare length variant allele 10.3 was found: $(AGAT)_5$ GAT $(AGAT)_5$. A D16S539 allele 9 and an allele 10 exhibited an A → C transversion at position 16 in the 3′ flanking region, a D18S51 allele 14 showed a G → A transition at position 18 in 5′ flanking region (TTCT strand).

Table 1
Sequence structure of new HumACTBP2 (SE33) alleles

Allele name	Sequence structure	Length in bp	No. of alleles
7.3[a]	$(AAAG)_{11}$	216	1
11.2[b]	$(AAAG)_{15}$	231	2
12.1[c]	$(AAAG)_{12}$	234	1
12.2[b]	$(AAAG)_{16}$	235	2
13	$(AAAG)_{13}$	237	1
13.2[bd]	$(AAAG)_{17}$	239	1
16.2[b]	$(AAAG)_{20}$	251	1
16.3[b]	$(AAAG)_{17}$	252	1
18.3[b]	$(AAAG)_{19}$	260	1
19.2[e]	$(AAAG)_9$ AAAAAG $(AAAG)_9$	263	1
20[e]	$(AAAG)_{20}$	265	1
25.2	$(AAAG)_8$ AAAAAG $(AAAG)_{16}$	287	2
26.2	$(AAAG)_{12}$ AAAAAG $(AAAG)_{13}$	291	1
27.2	$(AAAG)_{14}$ AAAAAG $(AAAG)_{12}$	295	1
29.2	$(AAAG)_{10}$ AAAAAG $(AAAG)_{18}$	303	1
30.2	$(AAAG)_{10}$ AAAAAG $(AAAG)_{20}$	307	1
30.2	$(AAAG)_8$ AAAAAG $(AAAG)_{21}$	307	1
31.2	$(AAAG)_8$ AAAAAG $(AAAG)_{22}$	311	1
33	$(AAAG)_8$ AAAAAG $(AAAG)_{13}$ AAAAAG $(AAAG)_9$	317	1
33.2	$(AAAG)_{12}$ AAAAAG $(AAAG)_{20}$	319	1
34.2	$(AAAG)_{13}$ AAAAAG $(AAAG)_{20}$	323	1

[a] Loss of G $(AAAG)_3$ in the 3′ flanking region. [b] Loss of $(AAAG)_3$ AG in the 5′ flanking region. [c] Insertion of a single A after the first of 12 AAAG repeats. [d] Black individual. [e] A → T transversion at position 17 in the 5′ flanking region.

Table 2
Sequence structure of new and rare variant Caucasoid D21S11, FGA and vWA alleles

Locus	Allele name	Sequence structure	Length in bp	No. of alleles
D21S11	29[a]	$(TCTA)_6$ $(TCTG)_5$ [...] $(TCTA)_{10}$	221	1
	29	$(TCTA)_4$ $(TCTG)_7$ [...] $(TCTA)_{10}$	221	1
	29.2	$(TCTA)_5$ $(TCTG)_6$ [...]$(TCTA)_9$ TA TCTA	223	1
	30[a]	$(TCTA)_5$ $(TCTG)_6$ [...]$(TCTA)_{11}$	225	1
	30	$(TCTA)_4$ $(TCTG)_6$ [...]$(TCTA)_{12}$	225	1
	31[a]	$(TCTA)_4$ $(TCTG)_6$ [...]$(TCTA)_{13}$	229	2
	32[a]	$(TCTA)_5$ $(TCTG)_6$ [...]$(TCTA)_{13}$	233	2
	35.2[a]	$(TCTA)_5$ $(TCTG)_6$ [...]$(TCTA)_{15}$ TA TCTA	247	1
FGA	22[b]	$(TTTC)_3$ TTTTTTCT $(CTTT)_{14}$ CTCC $(TTCC)_2$	196	1
	24.1[c]	$(TTTC)_3$ TTTTTTCT $(CTTT)_{16}$ CTCC $(TTCC)_2$	205	1
	26	$(TTTC)_3$ TTTTTTCT $(CTTT)_{16}$ GTTT CTTT CTCC $(TTCC)_2$	212	1
	27	$(TTTC)_3$ TTTTTTCT $(CTTT)_{17}$ GTTT CTTT CTCC $(TTCC)_2$	216	1
vWA	14	TCTA $(TCTG)_3$ $(TCTA)_{10}$	138	2
	15[d]	TCTA TCTG TCTA $(TCTG)_4$ $(TCTA)_3$ TCCA $(TCTA)_3$TCCA	142	1
	16[e]	TCTA $(TCTG)_4$ $(TCTA)_{11}$	146	1

[a] Already observed in Asian individuals. [b] T → C at position 3 in the 3′ flanking region. [c] Insertion of T following the position 35 in the 5′ flanking region. [d] T → C transition at position 7 in the 3′ flanking region (leading to 3 consecutive TCCA repeats). [e] C → T transition at position 3 in the 3′ flanking region (leading to 13 consecutive TCTA repeats).

All out of four D19S433 alleles sequenced differed from the GenBank sequence (accession no. G08036) by a T → A transversion at position 8 of the 3′ flanking region. Eleven out of fourteen D2S1338 alleles sequenced revealed a T → G transition at position 35 in the 3′ flanking region, which is in contrast to the GenBank sequence (accession no. G08202).

The new SE33 alleles observed in this study might give a further hint how the complex sequence structure at this locus might have evolved. Other allelic variants might have implications on mutation analysis of STR loci, in order define the maternal or paternal origin of a mutation.

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