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Research article

Unusual FGA and D19S433 off-ladder alleles and other allelic variants at the STR loci D8S1132, vWA, D18S51 and ACTBP2 (SE33)

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Abstract

A very short FGA allele *14 and a long D19S433 allele *19.2 were detected and sequenced, as well as the new D8S1132 alleles *12.1, *14 and *15.1. Further new sequence data (vWA allele *18.3, D18S51 allele *11.2, SE33 alleles *24.2, *32, *34 and *37, including the rare variant allele *13.2) are described.

Keywords: FGA; D19S433; D8S1132; vWA; D18S51; ACTBP2 (SE33); STR; Sequence

1. Introduction

In the course of population genetic studies and other investigations, a series of new and rare variant alleles have been found and sequenced. Some of the allelic variants have already been observed, but sequence data have not been published before.

2. Materials and methods

Blood or buccal swab samples were taken from Caucasoids living in Austria and Switzerland. DNA was extracted with Chelex or with the Qiamp DNA Mini Kit (Qiagen). Multiplex PCR was carried out with the Powerplex 16 System (Promega) or with the AmpFlSTR Identifiler PCR Amplification kit on an ABI Prism 310 Genetic Analyzer (Applied Biosystems) according to the manufacturer's instructions. The alleles were separated, purified and reamplified with unlabelled primers, sequenced with the Big Dye Terminator Cycle Sequencing Ready Reaction Kit on an ABI Prism 310 Genetic Analyzer (Applied Biosystems). The same primers were used for PCR and sequencing [1–5], which was carried out in both directions (GenBank accession nos. M64982, G08036, G08685, M25858, X91254 and V00481). SE33 alleles were designated according to the ACTBP2-nomenclature recommendations [6].

3. Results and discussion

A very short off-ladder FGA allele, found in a Swiss Caucasoid individual, was difficult to attribute to a locus in the Powerplex 16 System (Promega) as its amplicon size was between the expected allelic size ranges for the TPOX and the FGA locus. Both loci showed apparent homozygosity and the difference in relative size was 22.88 bp to the longest TPOX and 8.32 bp to the shortest FGA ladder allele, respectively. Therefore a TPOX allele >*13 (*18.3?) or an FGA allele <*16 (*14?) was assumed. Heterozygosity was observed after singleplex PCR at the FGA locus and sequencing exhibited an FGA allele *14 (164 bp) which differed by 2 CTTT repeats

Table 1
Sequence structure of the new length variant D8S1132 alleles *12.1, *14 and *15.1 and some new sequence variant alleles *20–*24 and *26

Allele name	Sequence structure	Length (bp)
*12.1	$(TCTA)_{11}$ TCTG TCTA	119
*14	$(TCTA)_6$ TCA $(TCTA)_8$	126
*15.1	$(TCTA)_{14}$ TCTG TCTA	131
*20	$(TCTA)_8$ TCA $(TCTA)_{10}$ TCTG TCTA	150
*21	$(TCTA)_{11}$ TCA $(TCTA)_{10}$	154
*21	$(TCTA)_{11}$ TCA $(TCTA)_8$ TCTG TCTA	154
*23	$(TCTA)_{12}$ TCA $(TCTA)_{11}$	162
*24	$(TCTA)_{12}$ TCA $(TCTA)_{12}$	166
*26	$(TCTA)_{11}$ TCA $(TCTA)_{15}$	174

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Table 2
Sequence structure of new length or sequence variant alleles

Allele name	5′ flanking region				Central region				3′ flanking region					No. of alleles	Length (bp)
	AAAG	AG	AAAG	AG	AAAG	AAAAAG	AAAG	AAAAAG	AAAG	G	AAGG	AAAG/ANAG	G	AAGG	AG		
13.2[a]	2	1	0	0	17	0	0	0	0	1	0	3	0	0	1	1	239
24.2	2	1	3	1	9	1	14	0	0	1	1	2	0	0	1	1	283
32	2	1	3	1	11	1	9	1	9	1	1	2	0	0	1	1	313
34	2	1	3	1	11	1	11	1	9	1	1	2	0	0	1	1	321
37	2	1	3	1	12	1	11	1	11	1	1	2	0	0	1	1	333

[a] Already observed in black individuals.

(8 bp) from the common allele *16 [1]: $(TTTC)_3$ TTTT TTCT $(CTTT)_6$ CTCC $(TTCC)_2$.

An analogous off-ladder allele, which differed in relative size by 8.4 bp from the longest D19S433 ladder allele *17.2 and by 10.81 bp from the shortest vWA ladder allele *11, was observed at the D19S433 locus using the AmpFlSTR Identifiler PCR Amplification kit. As heterozygosity was found at the vWA locus, a D19S433 allele >*17.2 (*19.2?) was suggested. Sequencing exhibited a D19S433 allele *19.2 (172 bp) showing a 2 bp deletion (AG) in an AAAG repeat (leading to an incomplete AA).

At the D8S1132 locus the new alleles *12.1, *14 and *15.1 have been found and sequenced (Table 1). In common D8S1132 alleles, an incomplete TCA repeat interrupts the stretch of $(TCTA)_n$ repeats, which has been considered to be conserved in previous studies [4].This incomplete repeat was missing in the new variant alleles *12.1 and *15.1. Further new sequence variant alleles *21–*24 and *26 are also shown in Table 1.

A new allele was also found at the vWA locus: the allele *18.3 (157 bp) exhibited an incomplete TCA repeat, which is very uncommon for vWA alleles [5]: TCTA $(TCTG)_4$ $(TCTA)_{11}$TCA $(TCTA)_2$.

Furthermore, the new length variant D18S51 allele *11.2 was investigated, which differed from the common allele *12 by a 2 bp deletion (AG) in the 3′ flanking region (allelic length 280 bp). As the position of the deletion is within an [$(AG)_3$ A]—structure (positions 108–114 of the 3′ flanking region) of the common D18S51 alleles (GenBank accession nos. X91254 and AP001534), the exact position of the deletion could not be assigned.

Some new variant alleles, either in sequence or in length, have been identified at the HumACTBP2 locus including the alleles *24.2, *32, *34 and *37. The rare variant allele *13.2 was found in a Caucasoid individual (Table 2).

For alleles within the size range of the allelic ladder, intrapolation can easily be performed to obtain the correct allelic designations, like for the vWA, D18S51 and SE33 alleles of this study. But in rare cases, extremely short or long alleles outside the common allelic size range can be difficult to be attributed to a locus in case of multiplex PCR testing. Amplification of individual loci can be helpful in these cases. Since extrapolation of relative size outside of the size range of the allelic ladders can give erroneous results in capillary electrophoresis, sequencing should be performed to obtain the correct allelic designation and exact length in bp in these cases.

Conflict of interest

None.

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