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TECHNICAL NOTE

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A very long ACTBP2 (SE33) allele

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Abstract Analysing a buccal swab we found a long allele in the STR system ACTBP2. For confirmation we sequenced the isolated PCR product and found a sequence structure common in alleles of type III. Based on the repeat array the new allele is assigned as allele "49".

Keywords ACTBP2 · Long STR variant · Sequence structure type III · DNA data base

Introduction

Since the first publications in the early 1990s, the STR system ACTBP2 has been intensively investigated in Germany [2, 3, 4, 6, 7]. Considering the high sequence and length variability of this system "out of ladder" alleles can occur but might not be detected.

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Materials and methods

Genomic DNA was extracted from a buccal swab using the Chelex protocol [8]. PCR products of ACTBP2 were generated using singleplex PCR [8] or the multiplex kits genRES MPX-1, genRES MPX-2 (Serac/genFor, Bad Homburg, Germany) and PowerPlex® ES system (Promega, Mannheim, Germany).Sequencing was carried out with PCR products eluted from polyacryamide gels (QIAEX II gel extraction kit, Qiagen, Hilden, Germany) using the BigDye Terminator kit v2.0 (Applied Biosystems, Weiterstadt, Germany).

Results and discussion

Analysing the DNA from the buccal swab with singleplex PCR or the multiplex kits we found a peak at the size of 368 bp (Fig. 1). Additional we found a second peak at 364 bp which was smaller and can be regarded as stutter band. We sequenced the 368 bp PCR product and found the following structure of the variable region:

– $(AAAG)_{14}$ AAAAAG $(AAAG)_{16}$ AAAAAG $(AAAG)_{16}$.

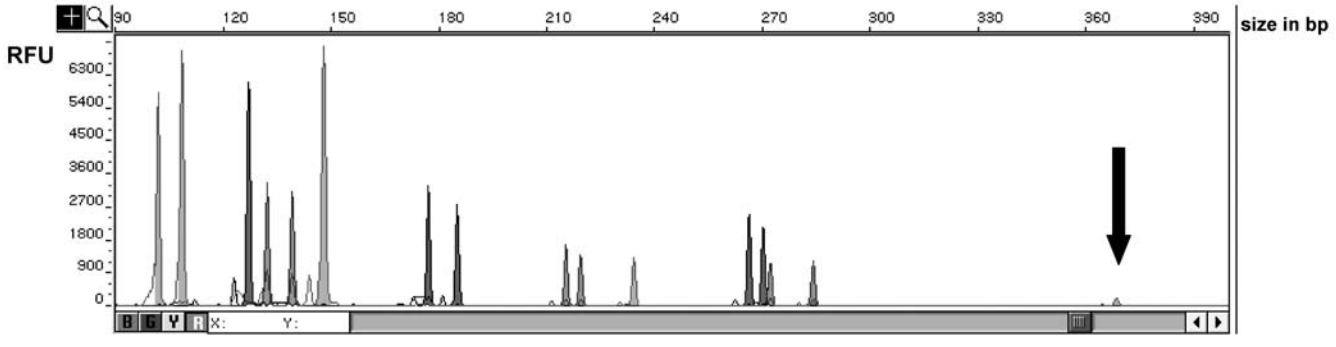

Fig. 1 The electropherogram shows a DNA profile with the SE33 allele 49 (*black arrow*). The DNA profiles were generated with genRES MPX2 kit amplifying the STR systems TH01, vWA, SE33, FIBRA, D21S11, D3S1358, D8S1179, D18S51 and Amelogenin (*RFU* relative fluorescent unit, *bp* basepair)

This typical sequence structure for ACTBP2 alleles greater than 32 (Type III sequence structure [3, 4]) proves that this peak is a real ACTBP2 allele. According to the nomenclature the detected allele is assigned as ACTBP2 allele "49" [5]. Until now ACTBP2 alleles have been reported up to allele 44 [1]. As most of the commercially available PCR kits for STR typing contain allelic ladders which lack such long alleles, determination and identification is more difficult especially when the possibility of long variants is not considered.

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