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LETTER TO THE EDITOR

Nomenclature discrepancies in the HPRTB short tandem repeat

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Dear Sir,

Unfortunately, forensic literature comprising typing data for the X-chromosomal STR locus HPRTB exhibits a discrepancy with regard to the nomenclature. The aim of this letter is to address the problem and to suggest an agreement to avoid further confusion.

After automated complete sequencing of the HPRT gene [6], the first descriptions of the tetranucleotide repeat polymorphism at the HPRT locus were published in 1991. Hearne and Todd [11] described the STR as a coding strand sequence $(TCTA)_n$ and Edwards et al. [7] assigned the same repeat to the complementary strand resulting in the $(AGAT)_n$ sequence. As a consequence, the outcome is a different number of repeats depending on whether the coding or the complementary strand is used in the allele designation (Fig. 1). This fact was communicated by J. M. Butler and D. J. Reeder in the Short Tandem Repeat DNA internet data base http://www.cstl.nist.gov/biotech/strbase/str_hprt.htm.

The use of $(AGAT)_n$ for allele designation yields one repeat less than the alternative motif and was commonly used during the early years of HPRTB typing. Counting the $(AGAT)_n$ repeats was also performed in the GenePrint® STR Systems (Promega, Madison, WI, USA) as well as some papers of Szibor and colleagues [5, 22, 23]. Consequently, in the early years of HPRTB typing, many population studies were published which all referred to the AGAT repeat counting [5, 8, 13, 16, 22]. Although Mertens et al. [12] chose the coding strand for allele designation, they wrongly defined the STR as $(ATCT)_n$ which resulted in the same repeat number as described for the $(AGAT)_n$ repeat counting. Similarly, in the HPRTB ladders of the Argus X-UL® and Argus X-8® kits (Biotype AG, Dresden, Germany), the nomenclature of the alleles is based on the coding strand sequence, but the repeat was wrongly defined as $(CTAT)_n$ [3] which again resulted in a loss of one repeat compared with the correct $(TCTA)_n$ counting mode. Moreover, the repeat allele design according to Mertens et al. [12] and Becker et al. [3] accidentally was in agreement with the traditional HPRTB nomenclature based on the $(AGAT)_n$ repeat counts defined by Edwards et al. [7]. Hence, the confusion was not obvious. Due to the fact that actually the Argus X-UL® and Argus X-8® are the only X-chromosomal STR kits commercially available, some population studies had already been performed [14, 24–26] and they were also in accordance with the nomenclature according to Edwards et al. [7]. Likewise, the recommendation to use cell line DNA as a standard in forensic DNA typing [23] listed the results of the traditional HPRTB typing. This bore the following HPRTB

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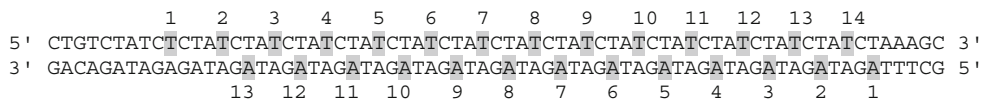

Fig. 1 Coding and complementary strands of the HPRTB repeat sequence

pattern for the standards as: K562*13, NA9947A*14/14, NA9948*14, and NA3657*13. There are some examples in which this recommendation was consulted in designing homemade multiplex kits and consequently the population data considered the traditional repeat counting mode [18–21].

In contrast, several authors of some recent papers, e.g. [1, 4, 9, 10, 15, 17, 27], used the allele designation according to Hearne and Todd [11] in order to meet exactly the ISFG nomenclature recommendations [2]. As a result, actually two different allele designation modes exhibiting one repeat difference are currently used, and the population data of different studies become comparable only after revision of the HPRTB data.

Although the latter counting mode follows better logical aspects, we recommend conserving the AGAT nomenclature according to Edwards et al., since it is the most employed one, and has already been included in widely used commercial kits. This proposal also meets the ISFG recommendations [2], as which are as follows: "If a nomenclature is already established in the forensic field but not in accordance with the aforementioned guideline, the nomenclature shall be maintained to avoid unnecessary confusion".

In the light of the present recommendation, HPRTB allelic ladders should be calibrated with cell line DNA genotypes as described above [23].

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