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Forensic value of the multicopy Y-STR marker DYS464☆

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Abstract. The tetranucleotide Y-chromosome short tandem repeat (Y-STR) marker, DYS464, first reported by Redd et al. [Forensic Sci. Int. 130 (2002) 97] appears to be the most polymorphic Y-STR marker discovered to date. A single primer pair can generate up to four distinct peaks over an allele range of 9–20 repeats. Allele calls can be made based on peaks that are present (conservative approach; C-type) or a combination of alleles and peak height ratios (expanded typing method; E-type). We have observed 113 C-types and 179 E-types in 679 males from three US populations.

Keywords: Y-STR; Y-chromosome; DYS464; Multicopy loci; DNA typing

1. Introduction

DYS464 occurs at least four times in the highly palindromic region near the center of the long arm of the Y-chromosome [1–3]. In forensic casework applications where the amount of typable DNA material may be limited, the use of highly polymorphic markers is advantageous in order to limit the number of markers needed to distinguish unrelated individuals.

2. Materials and methods

A total of 679 unique male DNA samples from three different US populations [4] were typed with DYS464 along with 21 additional Y-chromosome short tandem repeat (Y-STR) markers [3]. Only the DYS464 results are described in this report. Two novel DYS464 primer pairs, which differ from those originally described by Redd et al. [2], were used.

☆ Points of view are those of the authors and do not necessarily represent the position of the US Departments of Justice or Defense. Certain commercial equipment, instruments, and materials are identified in order to specify experimental procedures as completely as possible. In no case does such identification imply a recommendation or endorsement by the National Institute of Standards and Technology, nor does it imply that any of the materials, instruments, or equipment identified are necessarily the best available for the purpose.

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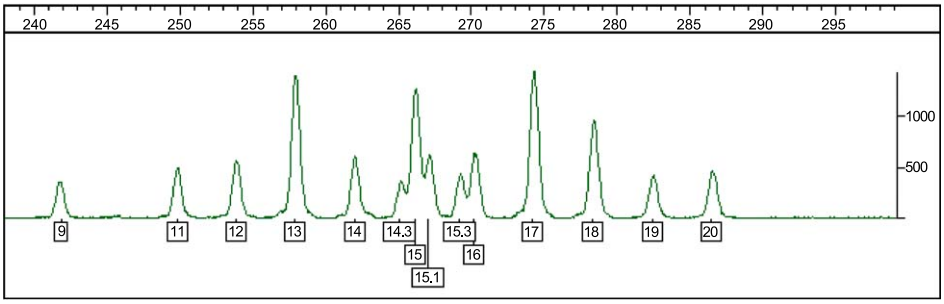

Fig. 1. NIST allelic ladder for DYS464 produced from eight different DNA samples. The variant alleles 14.3, 15.1, and 15.3 are helpful as a tool for measuring single-base resolution in electrophoretic systems.

The primers **VIC**-CTTTGGGCTATGCCTCAGTTT and GCCATACCTGGGTAACAGAGAGAC produce green-labeled amplicons in the size range of 242–286 bp for DYS464 alleles 9–20, while the primers **6FAM**-AGTTTACGAGCTTTGGGCTATG and GTGGCAAGATCTCATTTCTTCAA generate blue dye-labeled polymerase chain reaction (PCR) products that are 327–367 bp in size. PCR conditions are as previously described for the Y-STR 20plex [5]. An allelic ladder was created for DYS464 (Fig. 1), which contains all of the major alleles as well as single-base variants observed in our population study [3].

3. Results and discussion

We observed 179 expanded types with DYS464 in 679 male samples from three different US population sets: 265 African–Americans, 262 Caucasians, and 152 Hispanics. The addition of peak height information (E-type) to just allele calls (C-type) can result in an expansion in the number of types observed (e.g., Fig. 2). The most common types are 15,15,17,17, which occurs in 10.6% of this data set, and 15,15,16,17, which occurs in 7.5%. All other DYS464 types are found at less than the 5% level, with over half of the observed types occurring only once (92 of 179 observed). If the DYS464 data are collapsed to C-types (conservative typing method), then 113 allele calls are observed in these 679 males. By way of comparison to other DNA typing markers in the same data set, DYS385 exhibited only 56 different types and FGA, the single best autosomal STR examined, had only 78 different genotypes. Furthermore, the four single-copy Y-STRs DYS19, DYS391, DYS392, and DYS393, when combined, only produced 93 different

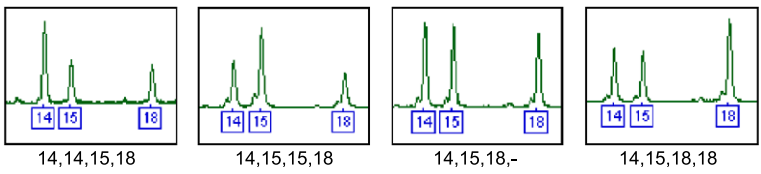

Fig. 2. Example of four samples with the same conservative DYS464 type, 14,15,18, which can be separated from one another by considering their expanded type through peak height variation.

haplotypes. Thus, DYS464 is more polymorphic than DYS385, the previously considered best multicopy Y-STR, and four single-copy Y-STRs in combination. A human Y-chromosome DNA profiling standard, NIST Standard Reference Material (SRM) 2395, is available and contains DYS464 information on its five male samples (see http://www.nist.gov/srm). This SRM will enable laboratories worldwide to accurately calibrate their DYS464 typing results.

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