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Research article

New autosomal STR loci☆

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Abstract

Additional STR loci can be beneficial for a number of human identity, forensic casework, and DNA database applications. The marker selection and characterization process applied at NIST in developing these new loci and assays are described along with concordance testing results from non-overlapping PCR primers. A 23plex for simultaneous amplification of 22 autosomal STR loci and an amelogenin sex-typing assay is also demonstrated.

Keywords: STR; Short tandem repeat; NIST; D10S1248; D2S441; D22S1045; Multiplex assays; MiniSTRs; Degraded DNA; Forensic DNA typing

1. Introduction

At the U.S. National Institute of Standards and Technology (NIST), we are characterizing additional autosomal and Y-chromosome STR loci that have a number of potential uses. In casework, additional information can be obtained from degraded DNA samples using miniSTR systems [1,2]. For identity testing work, kinship analysis, missing persons/mass disaster sample testing, and complex paternity testing can benefit by additional genetic markers [3]. More loci can help resolve relatives in growing national DNA databases to avoid adventitious matches. For example, although the U.K. National DNA Database started with 6 STR loci (SGM loci), it quickly expanded to 10 STRs (SGM Plus kit), and a future pan-European database is expected to include more than 10 STRs [4].

2. Materials and methods

This work represents an extension of initial studies begun by Coble and Butler [2] and continued by Hill et al. [5].

3. Results and Discussion

We have set about finding loci with narrow allele ranges, moderate to high heterozygosities, and clean flanking sequences that can be used in miniSTR assays [2,5]. The selected loci were characterized by examining the variation in ~660 U.S. population samples coming from African American, Caucasian, and Hispanic groups. Chromosomal positions were precisely defined, allelic ladders constructed, and standard samples were sequenced and genotyped to provide reference repeat calibration [6].

With the defined allele ranges characterized, a multiplex assay was developed that is capable of amplifying 22 of the 26 autosomal STRs and small amelogenin X–Y products for sex-typing purposes. This 23plex, dubbed the "Autoplex", uses five-dye chemistry to keep all PCR products under 400 bp in size (Fig. 1). Comparison of allele calls between the miniSTR assays and the Autoplex found full concordance in 99.80% of the 14,058 genotypes evaluated. This is similar to the 99.74%

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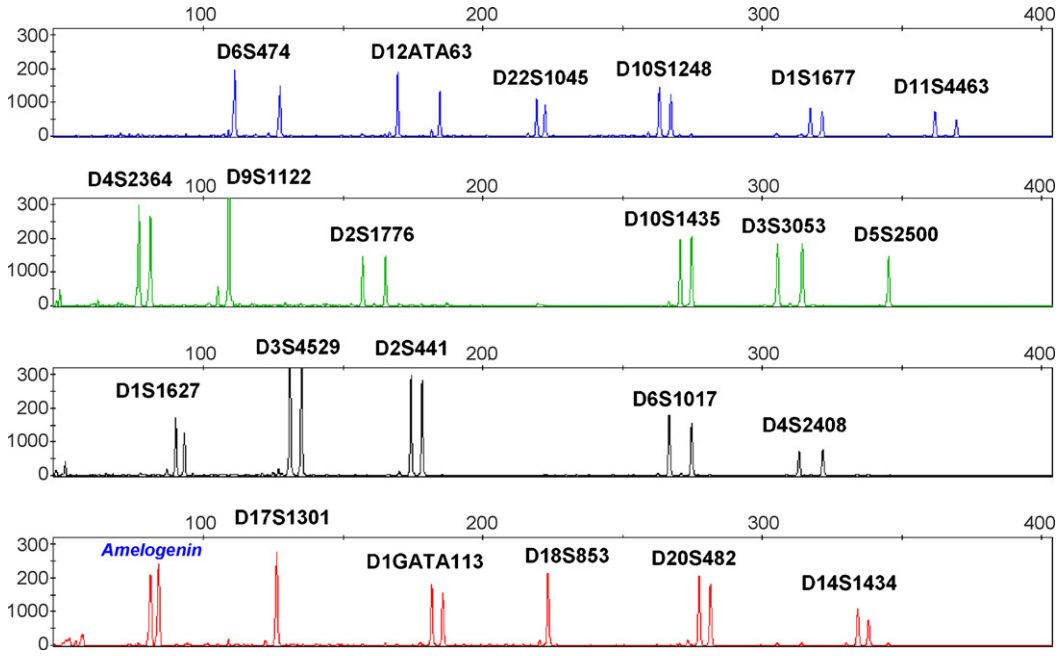

Fig. 1. Results from a 23plex amplification including 22 autosomal STRs and amelogenin for sex typing.

concordance rate found when evaluating the Identifiler kit versus MiniFiler kit allele calls [7].

Thus far, three of the 26 autosomal STR loci – D10S1248, D2S441, and D22S1045 – have been recommended for extending the core European loci [4]. We plan to continue to make information on these new loci available on the STRBase website at http://www.cstl.nist.gov/biotech/strbase/newSTRs.htm.

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Conflict of interest

None.

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