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Penta-, nona- and decaplex Y-STR typing systems: a comparative study

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Abstract

The analysis of Y chromosome-specific short tandem repeat (Y-STR) haplotypes allows patrilineage tracking and complements the highly robust autosomal STR systems used in forensic and kinship identification. Since 1994, in our lab, the use of the Y-STR Y27H39 (DYS19) was implemented. Later on, increasing availability of Y-STRs has broadened the patrilineage tracking potential. A complete nonaplex Y-STR set, including DYS385, 389 I and II, 390, 391, 392, 393 and DYS19, was investigated in 1995, provided information for the Charite Reference Database from Argentina and supported forensic casework investigations. For forensic purposes, we selected a pentaplex including: DYS19, DYS390, DYS391, DYS392 and DYS393. More recently, a decaplex, including DYS435, 436, 437, 438, 439, A7.1, H4, DYS19, DYS391 and DYS392, became available. In order to evaluate the penta-, nona- and decaplex Y-STR systems, unrelated Argentine males were analyzed (500, 100 and 104, respectively) by using an automated platform (ABI 310). For rapid forensic investigations, in which autosomal STR analysis is required, the pentaplex might be considered; if more informative data is required, nonaplex should be selected, especially due to worldwide internet supported Y-STR reference databases. It is suggested that part of the decaplex Y-STR markers should be included in the Charite Database in order to stimulate the systems use.

Keywords: Y-STR multiplexes; Argentina population

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1. Introduction

One of the latest developments introduced in the field of genetic markers suitable for forensic identification by DNA typing was that provided by the Y chromosome-specific short tandem repeats (Y-STRs) introduced by Roewer et al. [1]. Males commit most violent crimes; accordingly, the identification of a particular Y-STR haplotype might contribute to the identification of the perpetrator established by autosomal STR typing. In 1995, a sample of 100 unrelated males inhabiting Argentina were typed with a set of nine Y-STRs (2, www.ystrbase.charite.de). The local Y-STR haplotype reference database and the development of software that allowed the estimation of the frequency of a complete or partial Y-STR haplotype in our population [3] allows the inclusion of this value in the statistical evaluation of paternity indexes or in likelihood ratios. The aim of this work is to compare three different multiplex Y-STR systems; penta- [4], nona- [2] and decaplex [5] were compared in the population of Argentina.

2. Materials and methods

2.1. Samples

Blood samples were obtained from volunteer unrelated males from different areas of Argentina.

2.2. PCR reactions

Pentaplex: One injection of amplicons combined from a duplex and a triplex reaction, comprising duplex: DYS392 and DYS393, and triplex: DYS19, DYS390 and DYS391. Cycling conditions (for both multiplexes): 95 °C, 11 min, 1 cycle; 94 °C, 1 min; 59 °C, 1 min; 72 °C, 1 min, 28 cycles, 60 °C, 45 min, 1 cycle.

Nonaplex: One injection of two multiplex amplicons. A triplex DYS385, DYS389-I/II, DYS390. Cycling conditions: 95 °C, 7 min; 95 °C, 30 s, 60 °C, 1 min, 72 °C, 20 s, 29 cycles, 72 °C, 10 min. Quadraplex: DYS394, DYS391, DYS392, DYS393. Cycling conditions: 95 °C, 7 min; 95 °C, 30 s, 59 °C, 1 min, 72 °C, 20 s, 26 cycles, 72 °C, 10 min.

Table 1
Comparison between three Y-STR multiplexes

	Pentaplex	Nonaplex	Decaplex
Population size (*N*)	508	100	104
Discriminative power (%)	97.09	99.88	99.79
Frequency (most frequent haplotype) (%)	13.97	2.0	2.8
Number of unique haplotypes	93/508; 18.3%	88/100; 88%	86/104; 82.7%
Number of amplifications	2	2	1
Number of injections (ABI 310)	1	1	1

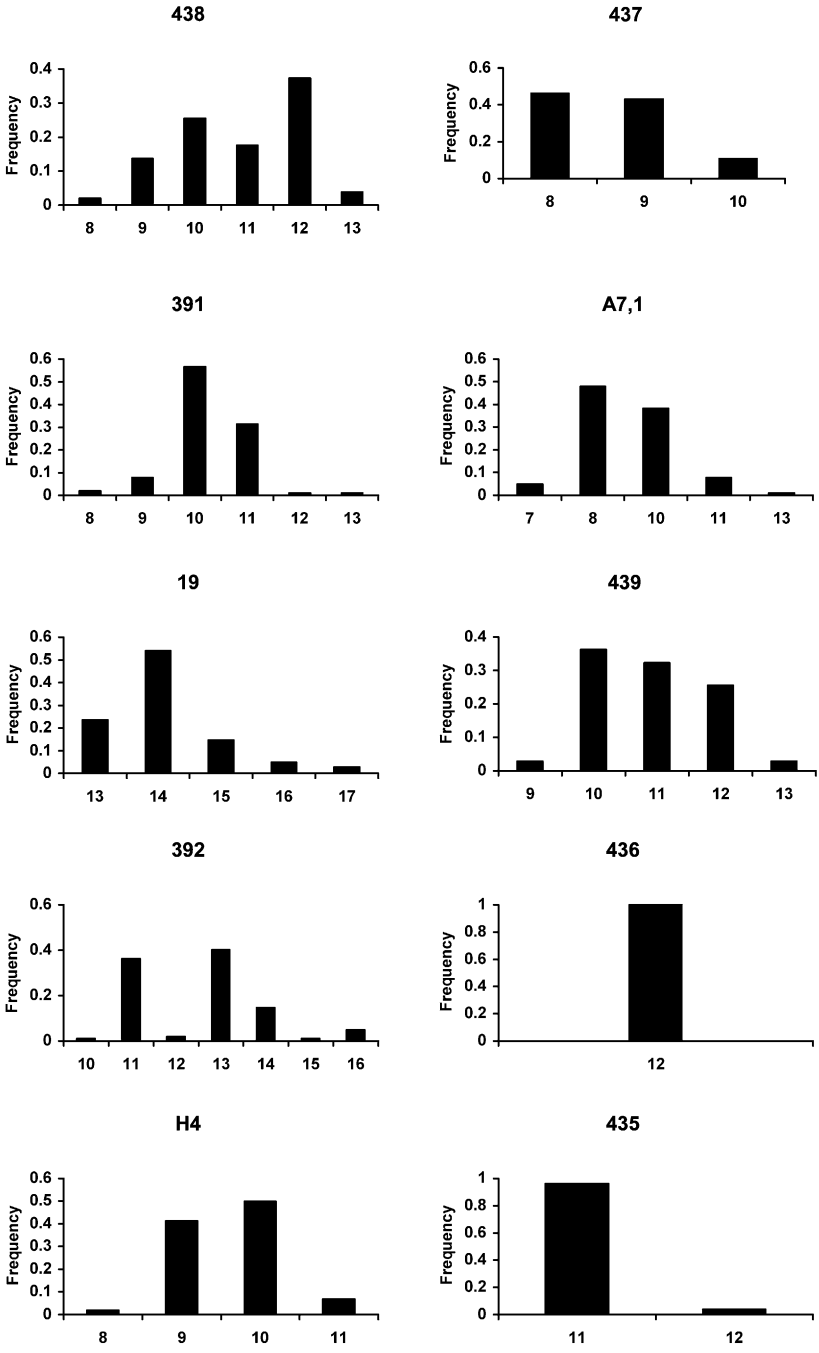

Fig. 1. Allele frequency distribution of decaplex system in Argentina population.

Decaplex single reaction: DYS435, DYS436, DYS437, DYS438, DYS439, A7.1, H4, DYS19, DYS391, DYS392. Cycling conditions: 95 °C, 7 min, 94 °C, 1 min, 55 °C, 1 min., 72 °C, 1 min, 25 cycles; 60 °C, 45 min.

2.3. Detection systems

All multiplexes were analyzed with capillary electrophoresis using an ABI 310.

2.4. Statistical evaluation

Discriminative power and genetic diversity were determined in accordance with Edwards et al. [6].

3. Results and discussion

The genetic attributes in the population of Argentina, for three different Y-STR multiplex systems, are summarized in Table 1. The discriminative power of the pentaplex, nonaplex and decaplex was 97.00%, 99.87% and 99.79%, respectively. Although nonaplex and decaplex systems are the most discriminative, the decaplex is the most sensitive and rapid (only one PCR reaction is required). However, its haplotype frequency distribution (Fig. 1) is not as varied as that found in the nonaplex system; this observation could be explained by the fact that in our population, extremely low levels of polymorphism were observed in both DYS436 and DYS435 (in the latter system, 100 samples exhibited the allele 11 and only 4 samples the allele 12).

In our lab, the five Y-STR system of the pentaplex, combined with more than 13 autosomal STRs, has been successfully used for routine forensic casework. In more than 500 cases, mostly rape cases, corpse identification and paternity testing, typing of a pentahaplotype allowed the determintion of patrilineage and confirmation of identity.

For rapid forensic investigations, in which autosomal STRs are required, the pentaplex might be considered; if more informative data is required, nonaplex should be selected, specially due to the worldwide internet supported Y-STR reference database [7,8]. Inclusion of part of the decaplex Y-STR markers in the Charite database may be recommended in order to broaden the patrilineage tracking potential.

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