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Review article

Identification, characterization and forensic application of novel Y-STRs

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Abstract

Y-chromosomal polymorphic STRs are a powerful tool for forensic and evolutionary studies. Within the last decade, a series of Y-STR systems have been developed and demonstrated to be suitable for a variety of forensic applications including sexual assault cases and paternity testing. This review describes our recent studies on novel male-specific Y-STRs, involving identification, development of a multiplex-PCR system, population study and forensic application.

Keywords: Forensic science; Y chromosome; Short tandem repeat; Multiplex polymerase chain reaction; Population study; Stains

1. Introduction

Among human chromosomes, the Y chromosome is unique in that most of it, excluding the pseudoautosomal region, does not participate in meiotic recombination and is inherited unaltered from father to son [1,2]. For this reason, analyses of human Y-linked polymorphic markers have been proposed as highly helpful tools for tracing human migration and evolution through male lineage as well as for forensic studies. Within the last decade, it has been demonstrated that these Y-specific markers, especially highly polymorphic Y-specific short tandem repeats (STRs), are suitable for forensic applications including cases of sexual assault and paternity cases where the putative father or other relatives are lacking, [3–5] and a series of Y-STR multiplex systems have been developed [6–10]. Of the various Y-STRs systems described so far, that consisting of nine loci—DYS19, DYS390, DYS391, DYS392, DYS393, DYS389I/II, and DYS385I/II—is best characterized and the haplotype data are continuously available online (Y-STR Haplotype Reference Database; http://www.ystr.org).

Recently, we have identified five additional and novel male-specific and polymorphic Y-STRs through a search of sequence database information and developed a multiplex system consisting of these STRs. In this article, we focus on our recent studies of these Y-STRs—DYS 441, DYS442, DYS443, DYS444 and DYS445—including identification, characterization and haplotype analysis, development of a multiplex-PCR system and forensic application.

2. Identification of male-specific and polymorphic Y-STRs

The Y chromosome consists of three regions: two pseudoautosomal regions (PARs) located on the most distal short arm (PAR1) and at the tip of the long arm (PAR2), a heterochromatic region, and the reminder of the chromosome (the non-recombining region, NRY) (Fig. 1). During male meiosis, the PAR1 and sometimes the PAR2 undergo pairing and meiotic exchange with the X chromosome [11]. The heterochromatic region is composed of highly repeated DNA families (DYZ1 and DYZ2) and considerable individual variation exists with regard to its length. The Y-STRs that are suitable for forensic application lie within the NRY since this region is transmitted unaltered from father to son. However, the NRY contains many blocks of X–Y homologous or Y-autosome homologous sequences [2], and thus these regions must be excluded for male-specific identification.

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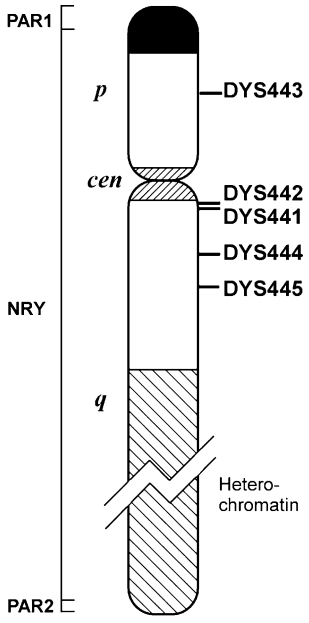

Fig. 1. A schematic diagram of the Y chromosome showing the positions of the five Y-STRs.

As a first step to identify novel male-specific and polymorphic Y-STRs, sequence database information (http://www2.ebi.ac.uk/genomes/mot/) was screened for DNA sequences on the Y chromosome that had been mapped in proximity to Y-specific regions within the NRY, and tetranucleotide microsatellites containing a stretch of nine or more repeat units were selected. After designing PCR primers and optimizing the PCR conditions for specific amplification of these microsatellites, male-specificity, existence of polymorphism and a regular mode of inheritance were tested by amplifying female DNA samples as negative controls, by analyzing the length of PCR products amplified from more than 10 male DNA samples, and by analyzing father/son combinations from routine paternity cases, respectively [12,13]. Some of the microsatellites that we located from the database were male-specific but not polymorphic, at least in the Japanese population, and therefore these STRs were excluded from our system. The positions of the five novel male-specific and polymorphic Y-STRs on the Y chromosome which we identified are shown in Fig. 1.

3. Development of a multiplex-PCR system

In order to utilize these STRs more availably in real forensic casework, a sensitive and simultaneous typing method using a single PCR and electrophoretic run has been anticipated, and therefore we developed a new multiplex-PCR system consisting of these five STRs. In certain situations such as cases involving old stains or highly decomposed body remains, it has been difficult to obtain

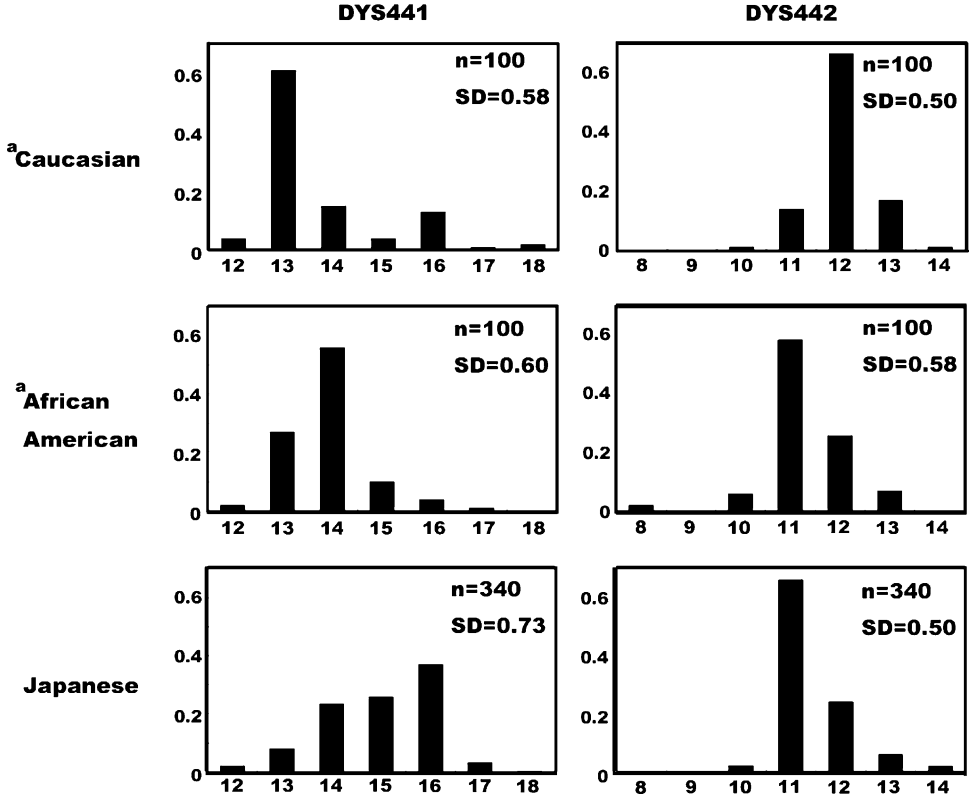

Fig. 2. Distributions of allele frequencies of DYS441 and DYS442 in Caucasian, African American and Japanese populations. SD: STR diversity. [a]Hanson EK and Ballantyne J, personal communication.