



## Population genetics of Y chromosomal STR haplotypes in Japanese population

C. Hashimoto\*, S. Nakamura, C. Sasaki, H. Takeshita, T. Nagai,  
H. Sugie, M. Furukawa, K. Kurihara

*Department of Legal Medicine, Kitasato University School of Medicine, Sagamihara 228-8555, Japan*

---

**Abstract.** Five Y chromosomal short tandem repeat (STR) loci DYS389I, DYS389II, DYS439, DYS438 and DYS392 have been analysed to obtain the allele and haplotype frequencies data in 129 unrelated Japanese male individuals living in Kanagawa by means of multiplex PCR with Y-PLEX™ 5 Primer Mix, followed by capillary electrophoresis using the ABI PRISM 310 Genetic Analyzer. The gene diversity values ranged from 0.555 (DYS439) to 0.737 (DYS389II). A total of 72 haplotypes was detected, of which 51 haplotypes were unique and the others occurred at least two times. The most common haplotype (DYS389I, DYS389II, DYS439, DYS438, DYS392) was 14–30–12–13–13 with a frequency of 10%, and the haplotype diversity was calculated as 0.969. The combined resolutions of the 5 Y STR multiplex system using the Y-PLEX™ 5 was applicable to routine case work. © 2003 Elsevier B.V. All rights reserved.

*Keywords:* Y chromosome; STR; Gene diversity; Haplotype diversity; Japanese population

---

### 1. Introduction

Short tandem repeat (STR) polymorphisms are powerful tools for human identification, paternity analysis and genetic mapping. We have already analysed the AmpFISTR Profiler Plus STR multiplex system and the 18 STR loci on the X chromosome in the Japanese population [1,2].

Recently, Y chromosomal STRs have been extensively investigated in forensic science for application in male identification and paternity testing. In particular, the Y chromosomal STRs are useful for analysis of stains in forensic investigations when a male suspect is involved, as is the case in most violent crimes, including sexual offenses. A number of Y STR multiplex assays have also been developed [3,4].

---

\* Corresponding author. Tel./fax: +81-42-778-9026.

*E-mail address:* mm02035r@st.kitasato-u.ac.jp (C. Hashimoto).

Table 1  
Y STR consensus structures and allele ranges

STR marker	Dye label	Repeat motif	Allele size range
DYS389I	FAM	TCTG/TCTA	243–260
DYS389II	FAM	TCTG/TCTA	361–385
DYS439	HEX	GATA	234–254
DYS438	TAMRA	TTTTC	131–158
DYS392	TAMRA	TAT	247–262

In the present investigation, we analysed 5 STR loci *DYS389I*, *DYS389II*, *DYS439*, *DYS438* and *DYS392* to obtain the allele and haplotype frequencies in a Japanese population living in Kanagawa.

## 2. Materials and methods

EDTA-treated blood samples were collected from 129 unrelated Japanese male individuals living in Kanagawa, and genomic DNA was isolated by using the DNA IQ™ System (Promega). Five Y chromosomal STRs analysed in the present study were indicated in Table 1. Multiplex PCR amplification of these 5 STR loci was performed using the Y-PLEX™ 5 Primer Mix (Reliagene Technologies, Inc.) according to the user's manual provided by the manufacturer. PCR products of 1 µl were mixed with 0.5 µl of GeneScan-500 [ROX] size standard (PE Applied Biosystems) and 12 µl of Hi-Di formamide. These samples were denatured at 95 °C for 3 min, then chilled on ice. The fluorescently labeled amplified fragments were electrophoresed using the ABI PRISM 310 Genetic Analyzer in a 47 cm, 50 µm i.d. capillary, filled with Performance Optimized Polymer 4 (POP4, PE Applied Biosystems) at 15 kV for 24 min at 60 °C, and analysed

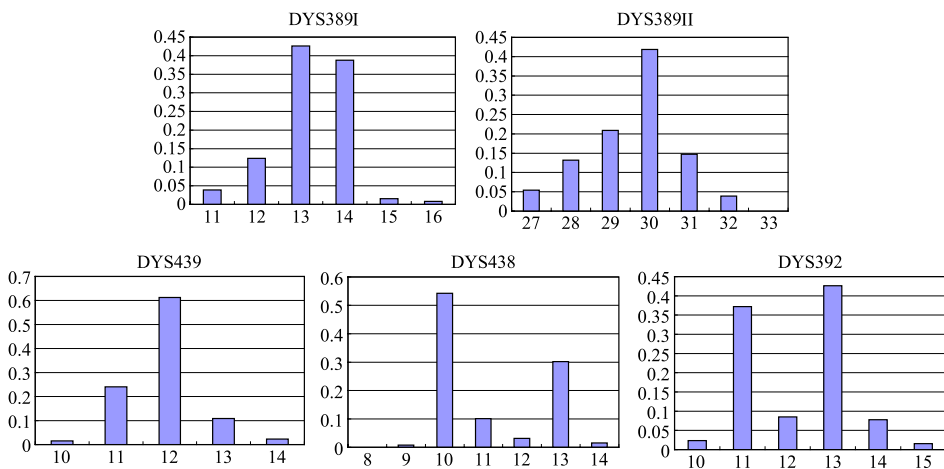


Fig. 1. The distribution of allele frequencies of Y-PLEX™ 5 loci in the Japanese population.

Table 2  
Y-PLEX™ 5 gene and haplotype diversities (%) in Japanese population

STR marker	Gene diversity
DYS389I	65.1
DYS389II	73.8
DYS439	55.5
DYS438	60.3
DYS392	66.6
Haplotype diversity	96.9

using GeneScan Analysis Software ver. 3.1.2. The Y-PLEX™ 5 Allelic Ladder was used as a reference for allele designation.

### 3. Results and discussion

In 129 Japanese male subjects, a total of 6 alleles for DYS389I, 6 alleles for DYS389II, 5 alleles for DYS439, 6 alleles for DYS438, and 6 alleles for DYS392 were observed. The distributions allele frequencies of Y-PLEX™ 5 loci in Japanese population were indicated in Fig. 1. The most common alleles were 13 (42.6%), 30 (41.9%), 12 (61.2%), 10 (54.3%), and 13 (42.6%) for DYS389I, DYS389II, DYS439, DYS438, and DYS392, respectively. The gene diversity values ranged from 0.555 (DYS439) to 0.737 (DYS389II). A total of 72 haplotypes was detected, of which 51 haplotypes were unique and the others occurred at least two times. The most common haplotype (DYS389I, DYS389II, DYS439, DYS438, DYS392) was 14–30–12–13–13 with a frequency of 10%, and the haplotype diversity was calculated as 0.969. Y-PLEX™ 5 gene and haplotype diversities in Japanese population were presented in Table 2. From these results, the five Y STR multiplex system using the Y-PLEX™ 5 was applicable for routine case work.

### References

- [1] S. Nakamura, T. Nagai, H. Matsui, M. Tabuchi, H. Sugie, M. Furukawa, H. Imai, K. Kurihara, Genetic analysis of AmpF1 STR Profiler Plus Loci in Japanese, *Prog. Forensic Genet.* 8 (2000) 236–238.
- [2] H. Matsushita, S. Nakamura, T. Nagai, M. Nakamura, H. Sugie, M. Furukawa, T. Komuro, K. Kurihara, Allele frequencies of dinucleotide repeat marker loci on the X chromosome in the Japanese population, *Forensic Sci. Int.* 129 (2002) 134–136.
- [3] M. Prinz, K. Boll, H. Baum, B. Shaler, Multiplexing of Y chromosome specific STRs and performance for mixed samples, *Forensic Sci. Int.* 85 (1997) 209–218.
- [4] J.M. Butler, R. Schoske, P.M. Vallone, M.C. Kline, A.J. Redd, M.F. Hammer, A novel multiplex for simultaneous amplification of 20 Y chromosome STR markers, *Forensic Sci. Int.* 129 (2002) 10–24.