THE ALLELIC DISTRIBUTION OF THE Y CHROMOSOMAL STR LOCI IN AN ESTONIAN POPULATION SAMPLE

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ABSTRACT: Six Y chromosomal short tandem repeat (STR) loci, DYS19, DYS388, DYS390, DYS391, DYS392 and DYS393 were analysed in a reference group of Estonian population consisting of 107 unrelated Estonians. The primers and conditions for PCR amplification were chosen according to the literature references. Detection of alleles was performed by polyacrylamide gel electrophoresis, using silver staining for allele visualisation. Allelic ladders for each STR were used, to ensure correct typing of the alleles. Allele frequencies, haplotype frequencies and gene diversity values were calculated. Allelic frequencies of Estonian population sample were compared to those of some other populations.

KEY WORDS: Y chromosome; Short tandem repeat (STR); Estonians.

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INTRODUCTION

Different types of polymorphisms have been found in the Y chromosome, including single nucleotide polymorphisms, duplications/deletions, insertions, complex rearrangements, satellite sequences like major, mini- and micro-satellites [10].

The Y chromosome analysis has been widely used to describe movements of populations all over the world [4]. Recent studies have revealed that the mutation rates of Y chromosomal STRs appear to be orders of magnitude higher than that of the SNPs [3]. In the investigation of male expansion in the world, the SNPs are indispensable. Due to the high mutation rate and specific mutational processes the STRs are valuable in distinguishing between recently diverged populations.

Another field exploiting Y chromosome polymorphisms is that of the forensic and paternity analysis which concentrates on individuals instead of populations.

The aim of the present study was to analyse Y chromosome STRs in an Estonian population sample primarily for forensic casework validation.

Estonians belong to the Finno-Ugric language group. There are only 5 populations in Europe whose mother tongue is not an Indo-European language. These are Basques and the speakers of Finno-Ugric languages: Hungarians, Saami, Finns and Estonians. In the biological history of Basques and Finns the genetic founder effects have played an important role and the genetic diversity of the Y chromosome is drastically reduced in these populations. It has been suggested that the same reduction would also occur in an Estonian population [14].

To understand the biological history of the Estonian Y chromosome, a brief overview of Estonian history would be valuable.

The oldest settlement in Estonia was a Mesolithic one – 7500 years BC. The ethnic identity of those inhabitants is unknown. About at the end of the third millennium BC the ancestors of Baltic people moved in. The population of Estonia at that time was very small – only a few thousands.

At the beginning of the 13th century the population of Estonia was relatively numerous, 150 000–180 000. After the German and Danish conquest the ethnic structure of Estonia changed. Before the conquest the number of non-Estonians in Estonia had been small, the non-Estonians being mostly the members of several Finnish tribes. After the conquest Germans settled mostly in towns, Swedes in the Estonian islands and the coastal areas. In some parts of South Estonia Latgalians infiltrated.

After the Livonian war in the 16th century Estonia was divided between Sweden, Denmark and Poland. In the next hundred years Estonia went under the Swedish rule. Warfare, epidemics and famine diminished the Estonian population, but soon a rapid population growth took place. A number of immigrants came in. Most of them were Finns, Russians and Latvians. It is estimated that in 1695 the Estonian population was 400 000.

The next great famine, the Northern War and the plague diminished the number of inhabitants in Estonia again. As a result of the Northern war Estonia was included in the Russian Empire for two hundred years. Estonia was an independent state between the First and the Second World War.

During the Soviet occupation the ethnic structure of Estonia was dramatically changed. The number of Estonians decreased, a mass migration from Russia and other countries led to an increase in the overall population.

The present Estonian population is about 1.5 million. About 1 million of them are Estonians, the remaining are Russians, Ukrainians, Belorussians and others.

The history allows us to believe that there a gene inflow into the Estonian gene pool has occurred throughout the centuries.

MATERIALS AND METHODS

In the experimental part of the current study we analysed blood samples, obtained from 107 unrelated Estonian individuals. Samples were collected on FTA bloodstain cards. DNA was purified according to the standard procedure. DNA was amplified in singleplex PCRs in previously published conditions [11, 12, 15].

We studied the distribution of four tetranucleotide STRs: DYS19, DYS390, DYS 391 and DYS393, and two trinucleotide STRs: DYS388 and DYS392.

Macrophor electrophoresis apparatus (Pharmacia LKB Biotechnology AB) was used for the separation of the amplified fragments on polyacrylamide gels, followed by visualisation by silver staining. Allelic ladders for each STR were used, to ensure correct typing of the alleles. The allelic ladder for DYS19 locus was derived in the course of the study, allelic ladders for the other loci were kindly delivered by Peter de Knijff (Leiden University).

Allele frequencies, haplotype frequencies and genetic diversity values were calculated for all loci. Statistical analyses of samples were carried out using the ARLEQUIN version 1.1 software package.

RESULTS AND DISCUSSION

The allele frequencies of the Y-STRs are shown in the Table I. The genetic diversities of the Y-STR loci is also presented.

Locus DYS19. 5 alleles altogether alleles were detected. The allele 14 was the most frequently represented one.

Locus DYS388. Only 3 alleles altogether were observed. The most frequent allele was 12. This locus has the lowest importance for the forensic purpose, as the genetic diversity for this locus is only 0.35.

Locus DYS390. 6 alleles altogether were observed. The allele 23 was the most frequently represented one. This locus has the highest genetic diversity, 0.72.

Locus DYS391. 4 alleles altogether were found. The most common allele was 11.

Locus DYS392. 5 alleles altogether were found. The most frequently represented allele was 11 but the frequency of the 14th allele is also high. No other European population has been reported to have such a high frequency of allele 14. The most common alleles in European populations are 11 and 13 [12]. In contrast, native Americans appear to have alleles containing over 13 repeats [1]. The allele 12 is rare in any European population [2, 5, 6] including Estonian population (Figure 1). The mutation mechanism of this lo-

cus discussed in the literature apparently does not follow the common single-repeat mutation model [1].

TABLE I. ALLELE FREQUENCIES AT SIX MICROSATELLITE LOCI AND GENETIC DIVERSITY VALUES FOR ANALYSED LOCI IN AN ESTONIAN POPULATION SAMPLE

Allele	DYS19	DYS388	DYS390	DYS391	DYS392	DYS393
9	–	–	–	0.019	–	–
10	–	–	–	0.430	–	–
11	–	–	–	0.533	0.486	–
12	–	0.794	–	0.019	0.028	0.019
13	0.019	0.056	–	–	0.056	0.542
14	0.458	0.150	–	–	0.393	0.411
15	0.271	–	–	–	0.037	0.028
16	0.234	–	–	–	–	–
17	0.019	–	–	–	–	–
22	–	–	0.084	–	–	–
23	–	–	0.421	–	–	–
24	–	–	0.168	–	–	–
25	–	–	0.262	–	–	–
26	–	–	0.056	–	–	–
27	–	–	0.009	–	–	–
Genetic diversity	0.67	0.35	0.72	0.54	0.61	0.54

Locus DYS393. 4 alleles altogether were observed. The alleles 13 and 14 were most frequently represented. The allele 14 does not have such a high frequency in any other European population. The frequency of the 13th allele is lower than in any other European population [8, 9, 13] (Figure 2). These data suggest the possible population-specific mutation occurrence in an Estonian population.

The genetic diversity values for these loci do not support the presence of founder effects in the Estonian population.

The Y-haplotypes were constructed on the basis of the six analysed loci. For 107 unrelated males, we observed 50 haplotypes. 32 haplotypes were unique, 5 haplotypes occurred twice, 4 combinations three times, 2 haplotypes four times, 2 haplotypes five times, 1 haplotype six times, three haplotypes seven times and the most frequent haplotype was found eight times. The most frequent haplotype was 14-12-23-11-14-14 (DYS19-DYS388-DYS390-DYS391-DYS392-DYS393).

The discrimination capacity of the combination of the six Y chromosomal loci was 46.7%, which is quite a low one. Additional markers have to be in-

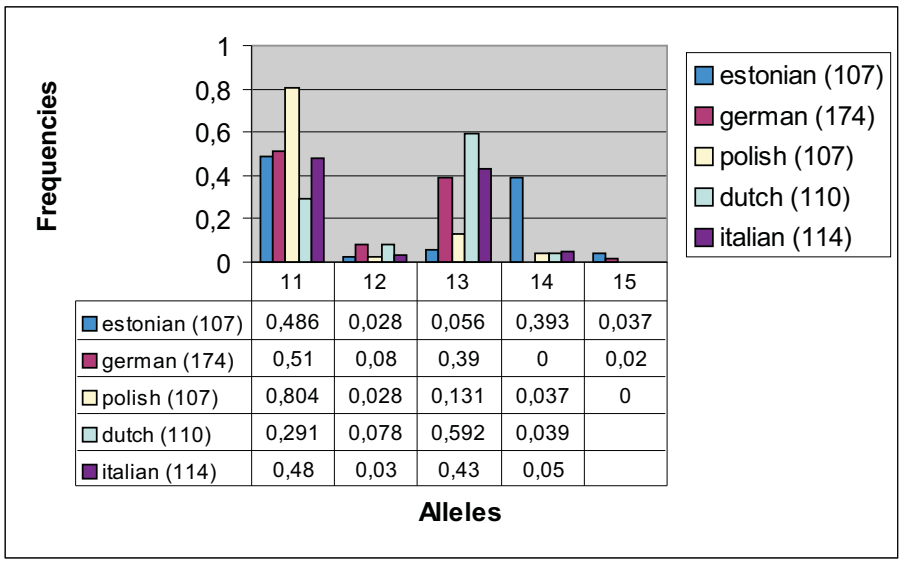

Fig. 1. DYS392 allelic distribution in different populations.

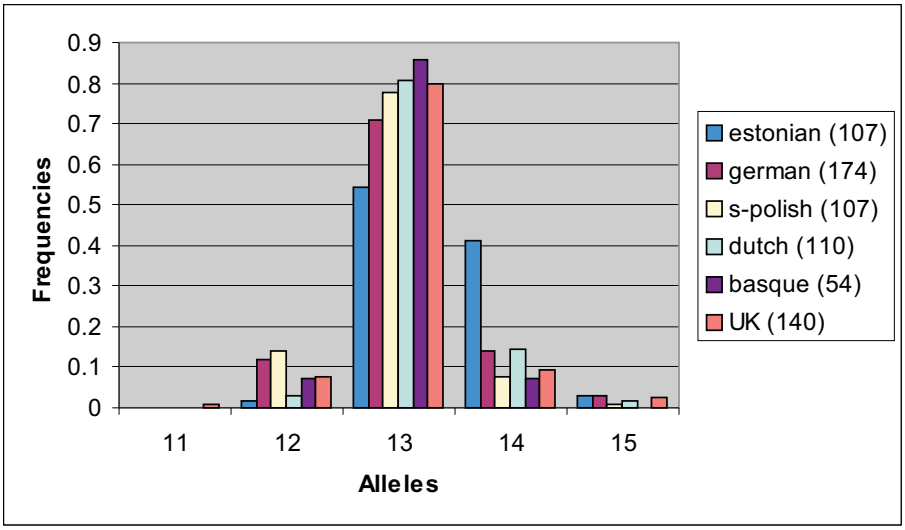

Fig. 2. DYS393 allelic distribution in different populations.

cluded to increase the individualisation power of the Y-chromosomal system.

In forensic practice the Y-chromosomal marker system consisting of six loci would be quite useful for excluding suspects. However, more markers

have to be included into the Y-chromosomal system for routine casework to increase discrimination power. Results of the present study revealed quite a high genetic diversity of Y chromosomal loci in the Estonian population.

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