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Population genetics of seven Y-STR loci in the Pomerania–Kujawy region of Poland

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Abstract

We have performed haplotyping of seven Y-STRs (DYS19, DYS389, DYS390, DYS391, DYS392, DYS393 and DYS385). We have found 199 different haplotypes in 277 unrelated males from Pomerania–Kujawy region of Poland. Among the haplotypes found, 164 (59.2%) were unique in our population sample, while 35 occurred more than once. Interpopulation comparisons were also performed and showed significant differences in haplotype frequencies between our population and other populations of Western and Southern European origin, while haplotype distributions in populations from other regions of Poland, as well as Central and Eastern Europe, were similar.

Keywords: STR; Y chromosome; Haplotype; Database; Population

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1. Introduction

Microsatellite loci localized on the Y chromosome are useful tools for both forensic identification and molecular anthropology. However, due to the Y chromosomes' uniparental mode of transmission, a haplotype frequency calculation requires a large number of individuals to be haplotyped. This requires the creation of a database large enough to properly assess haplotype frequencies. Such a database was created and is accessible by the Internet (http://ystr.charite.de) [1]. The aim of this study was to assess variability of Y-STR markers in the population of Pomerania–Kujawy region of Poland and compare the results with other populations of European origin.

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2. Materials and methods

The population investigated comprised 277 healthy, unrelated males, originating from the Pomerania–Kujawy region of Poland (city of Bydgoszcz and its surroundings). DNA was isolated from the whole blood using standard organic extraction method. DNA quantitation was performed by UV absorbance measurement. DNA samples were diluted to a concentration of 5 ng/μl. The loci under investigation, giving nine Y-STR products with seven primer pairs, were amplified in three PCR reactions (four PCR products from

Table 1
The 35 most common haplotypes and their frequencies in the Pomerania–Kujawy population

No.	Haplotype	Frequency
1	16-13-29-25-10-11-13-11,14	0.0433
2	17-13-30-25-10-11-13-10,14	0.0325
3	16-13-30-25-10-11-13-11,14	0.0253
4	16-13-30-25-11-11-13-11,14	0.0217
5	15-13-29-23-11-14-14-11,14	0.0181
6	15-13-30-24-11-11-13-11,14	0.0181
7	17-13-29-25-10-11-13-10,14	0.0181
8	15-13-30-25-11-11-13-12,14	0.0144
9	16-13-30-25-10-11-13-10,14	0.0144
10	15-13-30-25-11-11-13-11,14	0.0108
11	16-13-29-24-10-11-13-11,14	0.0108
12	16-13-30-24-11-11-13-14,15	0.0108
13	16-13-31-25-11-11-13-11,15	0.0108
14	17-13-31-25-10-11-13-10,14	0.0108
15	14-12-28-22-11-12-13-14,14	0.0072
16	14-13-29-23-11-13-13-11,14	0.0072
17	14-13-29-24-10-13-13-11,14	0.0072
18	14-13-29-25-11-13-13-11,14	0.0072
19	14-13-30-24-10-13-12-11,14	0.0072
20	14-13-30-24-11-13-12-11,14	0.0072
21	15-12-28-22-10-11-13-13,14	0.0072
22	15-13-29-23-10-14-14-11,14	0.0072
23	15-13-29-25-10-11-13-11,14	0.0072
24	15-13-30-26-11-11-13-11,15	0.0072
25	16-13-29-24-10-11-13-11,15	0.0072
26	16-13-29-25-11-11-13-11,14	0.0072
27	16-13-29-25-11-11-13-11,15	0.0072
28	16-13-30-24-10-11-13-11,14	0.0072
29	16-13-30-25-11-11-13-11,15	0.0072
30	16-13-31-23-11-11-13-14,15	0.0072
31	16-14-30-25-10-11-13-11,14	0.0072
32	17-13-30-25-10-11-13-10,11	0.0072
33	17-13-30-25-10-11-13-10,13	0.0072
34	17-13-31-24-11-11-13-11,14	0.0072
35	17-13-32-24-11-11-13-14,15	0.0072

Haplotypes are presented in a simplified form, as allele names only. Alleles are listed according to the following order: DYS319, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385. Full list of Y-STR haplotypes observed in our population sample is available from the corresponding author.

DYS19, DYS389I/II, DYS390 loci; three PCR products from DYS 391, DYS392, DYS393; two PCR products from DYS385). Primer concentrations were as follows—Reaction I: DYS319 0.32 μM, DYS390 0.32 μM, DYS389I/II 0.4 μM; Reaction II: DYS391 0.4 μM, DYS392 0.96 μM, DYS393 0.24 μM; Reaction III: DYS385 0.12 μM. Both primer sequences and amplification conditions (except for primer concentrations) were used according to Kayser et al. [2]. One microliter of PCR product was mixed with 1.5 μl of Dextran Blue loading solution (Promega) and 0.5 μl of fluorescent ladder (CXR) 60–400 bp (Promega) and loaded onto gel. The samples were run on a 5% polyacrylamide/bisacrylamide (19:1), 6 M urea, on an ABI 377 DNA sequencer. Electrophoresis results were analyzed using GeneScan Analysis 3.7 and Genotyper 1.2 software (ABI). Previously haplotyped DNA samples were used as controls for correct haplotyping.

3. Results

We have performed haplotyping of seven Y-STRs (giving nine PCR products): DYS19, DYS389I/II, DYS390, DYS391, DYS392, DYS393 and DYS385 (the so-called minimal haplotype). We have found 199 different haplotypes in 277 unrelated males from the Pomerania–Kujawy region of Poland. Among the haplotypes found 164 (59.2%) were unique in our population sample, 35 occurred more than once and seven of them occurred at least five times (see Table 1). The most frequent haplotypes were DYS19 16, DYS389I 13, DYS389II 29, DYS390 25, DYS391 10, DYS392 11, DYS393 13, DYS385 11,14, present at the frequency of ca. 0.04 (12 counts in 277 males) in our population and at the frequency of 0.009 in YHRD database. Haplotype frequency estimation, calculated using the Frequency Calculator on the YHRD Web page gave for this haplotype the following results: posterior frequency distribution: mean=7.6×10^{-03}, mode=7.3×10^{-03}, trace haplotype frequency: mean=7.9×10^{-03}, mode=7.6×10^{-03}.

Interpopulation comparisons were also performed for the most frequent haplotypes observed in our population (frequency at least 0.0072%, i.e. two counts in 277 individuals). We have noticed that 28 out of 35 such haplotypes tend to cluster in Eastern and Central part of Europe (including Germany), being scarcely represented or absent in other populations of European origin. On the contrary, the haplotype that is considered to be the most frequent in the YHRD database (14-13-29-24-11-13-13-11,14) occurred only once in our population sample. Moreover, we have observed that among 35 haplotypes which occurred more than once in our sample, one seemed to be specific for Polish subpopulations (14-12-28-22-11-12-13-14,14, frequency 0.0072), while two other haplotypes (16-13-31-23-11-11-13-14,15 and 17-13-30-25-10-11-13-10,13, both at frequency 0.0072) were observed in the Pomerania–Kujawy subpopulation exclusively and, thus, might be specific for this region.

4. Discussion

Y chromosome microsatellite (Y-STRs) population data from various parts of the world, especially from many populations of European origin, reveal the potential of Y-

STR polymorphisms for the identification of male DNA, including the possibility of defining the geographical region from which an individual may originate. These possibilities are becoming more promising as more Y-STR haplotypes are collected and thus Y-STR haplotype geographical distribution is better understood. Our study of 277 male DNA samples, with 199 distinct haplotypes (71.8%), revealed significant level of Y-STR polymorphism in the population of Pomerania–Kujawy region of Poland. Such a level of polymorphism is of great use for forensic purposes as well as for interpopulation comparisons. We have found that the most frequent haplotypes in our sample seem to be frequent, or at least present, in subpopulations of Poland and other countries of Eastern Europe, as well as in Germany and Scandinavian populations, while they are virtually absent in other populations of European origin. On the contrary, the haplotype that is very frequent in Western European populations is rather rare in all Polish populations investigated up to now. Thus, our results suggest that the observed interpopulation differences in Y-STR haplotypes distribution could be of geographical rather than of ethnical nature. Additionally, we have identified three haplotypes which seem to be potential markers of an individual's origin, as they are present exclusively in Polish subpopulations, including two haplotypes present in the Pomerania–Kujawy subpopulation only.

Additional experiments are required to fully assess the Y-STR polymorphism in our subpopulation as well as in other Polish subpopulations. Statistical calculations are also necessary, which will allow for precise estimation of the inter- and intrapopulation level of genetic diversity. Such experiments and calculations are under way.

References

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