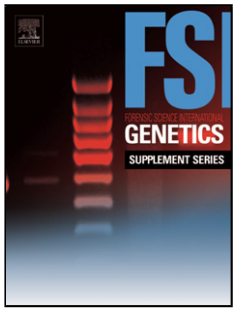

Title: Comparison of Y-chromosomal haplogroup predictors

Authors: Barbora Emmerova, Edvard Ehler, David Comas, Jitka Votrubova, Daniel Vanek

PII: S1875-1768(17)30164-6
DOI: http://dx.doi.org/10.1016/j.fsigss.2017.09.025
Reference: FSIGSS 1291

To appear in:

Received date: 1-9-2017
Accepted date: 11-9-2017

Please cite this article as: {http://dx.doi.org/

Comparison of Y-chromosomal haplogroup predictors

Barbora Emmerova[1,2], Edvard Ehler[1,3], David Comas[4], Jitka Votrubova[1], Daniel Vanek[1,5]

[1] Forensic DNA Service, Prague, Czech Republic
[2] Charles University in Prague, Faculty of Science, Prague, Czech Republic
[3] Charles University in Prague, Faculty of Education, Prague, Czech Republic
[4] Institut de Biologia Evolutiva (CSIC-UPF), Barcelona, Spain
[5]Charles University in Prague, 2nd Faculty of Medicine, Prague, Czech Republic

*Corresponding author: Daniel Vanek, Forensic DNA Service, Janovskeho 18, 170 00 Prague 7, Czech Republic, Tel.: +420 603 979 915; E-mail: daniel.vanek@fdnas.cz

Abstract

We used Y-chromosome DNA typing data from 342 unrelated individuals from the Czech Republic to obtain the Y-STR haplotypes and haplogroup-defining single nucleotide polymorphisms (SNPs). The resulting Y-STR haplotypes were subsequently entered into 5 different Y-haplogroup predictors (Vadim Urasin, Nevgen, Hapest, Jim Cullen, and Felix Immanuel), and the results were compared. We also evaluated the influence of the number of STRs used (12 vs. 19 loci) on the accuracy of the Y-haplogroup predictions.

Keywords: Y-chromosome; haplotype; haplogroup; predictors; SNP; STR

Material and methods

DNA typing

We used Y-chromosome DNA typing data from 342 unrelated individuals from the Czech to obtain Y-STR haplotypes and haplogroup-defining SNPs. The samples were genotyped for the non-recombining region of the Y-chromosome using TaqMan Real-Time PCR Assays (Thermo Fisher Scientific, USA) in a hierarchical manner for a set of variable numbers of SNP markers and for six indels that were amplified in a single multiplexed assay (termed the Multiplex-2 and including M91, M139, M60, M186, M175, and M17) that was run on an ABI3100 (Thermo Fisher Scientific, USA). All of the samples were first typed for M168, M89, M45, M9, M207, and the markers of the Multiplex-2. All individuals were derived from M168. Individuals ancestral to M89 were then typed for M174 and M96, and those derived from M96 were further typed for P147, M33, P177, P2, M2, M35, M215, M78, M81, V12, V13, V65 and M123. The individuals derived from M89 and ancestral to M9 were typed for M201, M69, M170 and M304. Those individuals derived from M170 were then typed for M253, L22, P215, P37.2, M26, M423 and M223. Those derived from M304 were typed for M267, M172, M410 and M102. Those derived from M410 were typed for M67 and M92, and those derived from M102 were typed for M241. The individuals derived from M201 were typed for P15 and P287. The individuals derived from M9 were typed for M207. The individuals derived from M207 were first typed for M269, and those derived from M269 were then typed for L23, P311, P312, M153, SRY2627, U106, L48, L2, L20, and U152. Those derived from M207 and ancestral to M269 were typed for M173, SRY10831.2, M343, M18, M335 and P297. The individuals derived from M17 (in the Multiplex-2) were typed for M458. The individuals derived from M9 but not from any of the previously described assays were typed for M20, N-LLY22 and M70.

All individuals were typed for a set of 19 STRs using two different multiplexes, i.e., 17 STRs were amplified with the AmpFlSTR Yfiler PCR Amplification kit (Thermo Fisher Scientific, USA), and two additional STRs, DYS426 and DYS388, were amplified in a separate PCR multiplex reaction.