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POPULATION DATA

Frequency data for 17 Y-chromosomal STRs and 19 Y-chromosomal SNPs in the Tyrolean district of Reutte, Austria

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Abstract We established a data set of 17 Y-STRs of 261 males from the Tyrolean district of Reutte. In total we observed 228 different haplotypes, 203 of which were unique and 25 occurred between two and four times. The haplotype diversity was 0.9987 and the discrimination capacity was 0.8736. Further, samples were typed with a selection of 19 Y-SNPs to establish the haplogroup background. Data are available in the Y chromosome haplotype reference database under accession number YA003715.

Keywords Y-STR · Y-SNPs · Y chromosome · Population data · Forensic science

The Tyrolean district of Reutte (Austria) is 1,237 km² in size and has 37 villages with a total of 31,711 inhabitants (Census 2011, Statistik Austria, http://www.statistik.at/). It is located in the northern slopes of the Central Alps and borders to Bavaria (Germany) to the north and the Austrian federal state of Vorarlberg to the west. To the south and east, this region is geographically structured by deep cutting valleys and massive mountain ranges resulting in relatively restricted historic connections to the remaining regions in Tyrol. The geographic isolation to the southeast is still evident in the dialect that originates from Western Upper German dialects, in contrast to the remaining regions in Tyrol, especially the eastern part of the Inn valley where descendants of Eastern Upper German dialects (e.g., Bavarian) are spoken [1]. Blood samples were collected from healthy adult men under written consent (ethical committee of Innsbruck Medical University UN2598, session number 241/4.5) during the regular blood donation cycle of the blood bank of Innsbruck. DNA was extracted from whole blood using the Nexttec Genomic DNA Isolation Kit (Biozym Scientific GmbH, Hessisch Oldendorf, Germany). DNA extracts were amplified using the AmpFISTR Yfiler PCR amplification kit (Life Technologies (LT), Carlsbad, CA, USA) using the manufacturer's protocol. The amplification products were separated on an ABI PRISM 3100 Genetic Analyzer (AB), and data were analyzed using Genotyper (v. 3.7, LT) and GeneMarker HID (v. 1.70, SoftGenetics, State College, PA, USA). The haplotype diversity (H) was calculated using the formula $H=N\ /\ (N-1)\times(1-\sum p_i^2)$, where Σ stands for the sum over $i=1$ to the total number of different haplotypes, p_i is the frequency of the ith haplotype and N is the number of individuals in the sample [2]. The discrimination capacity (DC) was calculated as the ratio between the number of different haplotypes and the total number of specimens in the population sample. Analysis of molecular variance and the genetic distance between populations (F_{st}) were estimated using Arlequin ver. 3.5 [3]. The Y-STR haplotypes based on the minimal haplotype loci (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, and DYS385a/b) were compared to

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2,881 haplotypes from the following nine populations contained in the Y chromosome haplotype reference database (YHRD, http://www.yhrd.org/) [4]: Brescia, Italy (N=106, YA003295); Lombardy, Italy (N=182, YA003068 and YA003095); Udine, Italy (N=47, YA003721); Veneto, Italy (N=120, YA003094 and YA003091); Verona, Italy (N=153, YA003327); Salzburg, Austria (N=176, YA003408); Stuttgart, Germany (N=1122, YA003031); Munich, Germany (N=281, YA002896); and Freiburg, Germany (N=433, YA003038); and their MDS plots were calculated. To correct for multiple comparisons the method outlined in [5] was employed. Furthermore, samples were typed for 19 single nucleotide polymorphisms (Y-SNPs) including loci M9 (rs3900), M17 (rs3908), M45 (rs2032631), M78, M89 (rs2032652), M96 (rs9306841), M170 (rs2032597), M173 (P241, rs2032624), M201 (rs2032636), M223, M253 (rs9341296), M269 (rs9786153), M304 (rs13447352), M343 (rs9786184), P15, P37, SRY_{10831} (SRY_{1532}, rs2534636), U106 (M405, S21, rs16981293), and U152 (S28, rs1236440) using a single nucleotide primer extension approach as outlined in [6]. Our sample set, comprising 261 male individuals from Reutte, yielded 228 different 17 locus Y-STR haplotypes, 203 of which were unique (Table S1). The two most frequent Y-STR haplotypes were found four times each (1.53 %), and four haplotypes occurred three times (1.15 %, Table S2). Allele frequencies and diversity indices are given in Table S3. The H values without and with DYS385 were 0.9987±0.0005 (DC=0.8621) and 0.9999±0.0000 (DC=0.8736), respectively. Our Y-STR population sample was compared to the neighboring datasets accessible via the YHRD (within a geographical distance of 250 km). It clustered closer to the populations from Munich and Salzburg (Fig. S1 and Table S4) than to the southwestern German populations (Freiburg and Stuttgart), which share the same linguistic root (Western Upper German dialect). By analysis of 19 Y-chromosomal SNPs, 258 of the 261 samples gave successful results and were assigned to 16 different haplogroups (Tables S1 and S5) with a major proportion (n=54, 20.9 %) belonging to haplogroup R-U106, followed by haplogroups R-M269 (n=35, 13.6 %), R-U152 (n=32, 12.4 %), and I-M253 (n=27, 10.5 %). The R1b clade, comprising of haplogroups R-M269, R-U106, and R-U152, accounted for 46.9 % of the Y chromosomes. The results of our analyses demonstrate that the investigated population sample is representative for the West Eurasian gene pool.

This paper follows the updated recommendations of the DNA commission of the International Society of Forensic Genetics on the use of Y-STRs in forensic analysis [7] and the guidelines for publication of population data requested by the journal [8, 9].

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