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Announcement of population data

Y-chromosome STR haplotype diversity in Brazilian populations

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Abstract

A sample of 198 unrelated males distributed among the five geopolitical regions in Brazil were typed for the minimal Y-STR haplotype composed of microsatellite loci DYS19, DYS385I/II, DYS389I, DYS389II, DYS390, DYS391, DYS392 and DYS393. Gene frequency data, gene diversity, haplotype diversity and power of discrimination were estimated. An AMOVA indicated that 99.97% of the haplotypic variation is found within regions and only a small 0.03% and non significant variation is found among the five regions (Fst = 0.00031, *P*-value = 0.43). This result suggests that a single national database of Y-STR haplotypes can be used in the quantitative assessment of matches in forensic casework in the Brazilian population. A significant haplotype diversity of 99.8% was found and 172 different haplotypes were observed in 198 chromosomes. Haplotype (14–11, 14–13–29–24–11–13–13) with five occurrences in 198 chromosomes was the most frequent in Brazil.

Keywords: Y-chromosome STR; Haplotypes; Brazilians; Population genetics

1. Population

DNA samples from 198 unrelated healthy males randomly sampled from paternity casework in populations from the five geopolitical regions in Brazil were studied: 25 from the North, 42 from the Northeast, 48 from the Central west, 36 from the South west and 47 from the South. All studies were carried out anonymously and participants provided written consent for the use of genetic data for statistical studies. All males were analyzed at the tetranucleotide microsatellite loci DYS19, DYS385I/II, DYS389I, DYS389II, DYS390, DYS391 and DYS393 and at the trinucleotide locus DYS392.

2. DNA extraction

DNA from different individuals was extracted from whole blood samples using GFX Genomic Blood Kit (Amersham-Pharmacia, Uppsala, SE) and quantified in agarose minigels.

3. PCR conditions

Locus information and PCR-primer sequences were obtained from Kayser et al. [1] or from the website: http://www.ystr.org. Forward primers were 5′ labeled with the following dyes: NED (DYS19, DYS389I and II), 6-FAM (DYS385I/II, DYS390 and DYS391), HEX (DYS392 and DYS393) for detection under virtual filter D with an ABI Prism 377-96 DNA sequencer. PCR conditions were 1.25 U Taq (Phoneutria), 1–10 ng genomic DNA, 10 mM Tris-HCl

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(PH 8.3), 200 μM dNTPs with 2 and 3 mM MgCl2 for the pentaplex and triplex reactions respectively in a total volume of 12.5 μl. Primer concentrations for the pentaplex (DYS19, DYS391, DYS392, DYS393 and DYS385I/II) and for the triplex (DYS19, DYS389 and DYS390) were as described by the PCR protocols in http://www.ystr.org. Cycle conditions used were as follows in a PE 9600 (GeneAmp PCR System): an initial pre-incubation at 95 °C for 10 min followed by 30 cycles of 94 °C for 1 min, 58 °C or 56 °C for 1 min for the pentaplex and triplex reaction respectively, 72 °C for 2 min, and a final extension step at 72 °C for 10 min.

4. STR typing

Locus ordering and allele designation is consistent with the database of the International Forensic Y-User Group (http://www.ystr.org). Fragment sizes were determined automatically using GeneScan Analysis 3.1 software (Applied Biosystems). Consistent allele designation and typing quality were assured by simultaneous electrophoretic analysis of at least two previously GEP-ISFG validated haplotyped samples. The allele nomenclature was as proposed by Kayser et al. [1].

5. Quality control

Proficiency testing of the GEP-ISFG-year 2001, 2002 and 2003 as well as successful typing of samples received from the Y-STR Haplotype Reference Database by the Institute of Legal Medicine, Humboldt-University, Berlin (http://www.ystr.org/europe).

6. Data analysis

Analyses were carried out with the Arlequin version 2.000 software (http://lgb.unige.ch/arlequin/) [2] following standard procedures.

7. Access to the data

Upon request by contacting dario@pos.ucb.br.

8. Results

The 198 complete nine-locus haplotypes were obtained and subject to analysis (Table 1). The average gene diversity (GD) at the nine STR loci, corresponding to the power of paternity exclusion (PE) [3] was 63.7%. Loci DYS385I/II and DYS389II were the most informative with overall Brazilian population GD of 70% and 90.5% respectively, and loci DYS391, DYS393 and DYS389I the least informative, below the 55% range (Table 2). In the total sample, 172 different haplotypes in 198 individuals were observed and out of the 172, a total of 152 were observed only once (Table 3). A remarkable haplotype diversity of 99.8% was found when all

Table 1
Observed Y chromosome haplotype distribution in the five geopolitical regions in Brazil

DYS19	DYS385I/II	DYS389I/I	DYS389I/II	DYS390	DYS391	DYS392	DYS393	*N*
North								
14	11,14	14	29	24	11	13	13	1
14	11,12	13	29	24	10	13	13	1
14	11,14	13	29	24	10	13	13	2
14	11,14	13	29	23	11	13	14	1
14	14,14	12	28	22	10	11	14	1
14	11,14	14	30	24	11	13	13	1
14	11,15	13	30	24	11	13	13	1
16	17,18	13	30	21	10	11	14	1
16	16,16	13	30	21	10	11	15	1
14	11,14	14	31	24	11	13	13	1
13	14,17	14	30	24	10	14	13	1
16	15,15	14	30	24	11	12	14	1
14	11,14	14	30	22	11	13	13	1
14	11,11	14	30	23	11	13	13	1
14	13,15	13	30	23	10	11	11	1
15	11,14	13	30	24	10	13	13	1
15	11,14	13	30	24	10	11	13	1
14	12,16	13	30	24	11	13	14	1
16	11,11	14	32	24	10	11	13	1
14	12,15	13	30	25	11	13	13	1
14	13,16	13	30	22	10	11	12	1
14	13,14	12	28	22	10	11	13	1

Table 1 (*Continued*)

DYS19	DYS385I/II	DYS389I/I	DYS389I/II	DYS390	DYS391	DYS392	DYS393	*N*
15	12,17	12	28	24	10	11	12	1
14	16,18	13	30	24	10	12	12	1
Northeast								
13	12,18	12	29	24	10	11	12	1
13	11,15	13	29	24	11	14	13	1
15	13,15	14	30	24	11	13	13	1
14	11,14	14	30	24	11	13	13	1
15	11,12	13	28	22	11	11	13	1
14	11,14	13	29	24	11	13	13	1
15	11,14	13	30	24	10	13	13	1
13	17,18	12	29	24	10	11	13	1
15	13,13	14	32	22	10	11	14	1
15	12,14	12	26	24	11	13	13	1
12	11,12	14	30	25	11	13	13	1
14	11,14	12	29	25	11	13	13	1
14	11,15	13	29	25	10	13	13	1
15	13,14	13	29	24	11	13	13	1
14	11,17	14	30	24	11	13	13	1
15	12,15	13	29	23	10	11	13	1
13	15,18	13	29	24	10	14	13	1
15	13,17	12	29	25	11	11	12	1
14	11,14	13	29	24	10	13	13	1
13	17,18	13	30	22	10	11	13	1
14	11,14	12	28	24	11	13	13	1
13	16,18	13	30	24	10	11	13	1
14	11,14	15	31	24	11	13	13	1
13	12,13	13	30	21	11	13	13	1
16	12,14	13	30	26	11	12	13	1
14	11,14	13	29	24	11	13	12	1
14	11,14	13	29	24	11	13	14	1
13	13,14	14	30	24	9	12	14	1
14	11,14	14	30	25	11	13	13	1
14	14,17	13	29	24	10	11	12	1
15	13,18	13	29	23	10	11	12	1
14	14,16	13	30	24	10	11	12	1
14	13,16	13	29	23	10	11	12	1
14	11,15	13	30	23	11	13	13	1
15	11,15	13	29	24	11	13	13	1
15	16,16	13	29	23	10	12	13	1
13	16,16	13	31	25	10	11	13	1
14	11,13	13	29	24	11	13	13	1
15	14,17	12	28	23	10	11	12	1
14	10,14	14	30	24	11	13	14	1
14	11,14	13	30	23	11	14	13	1
13	16,16	14	31	23	10	11	13	1
Central west								
14	11,14	13	30	24	11	14	13	1
14	11,11	12	28	24	11	13	13	1
15	11,16	13	29	24	12	13	13	1
14	11,16	13	29	24	11	13	13	1
13	17,17	12	29	24	10	11	13	2
15	16,17	13	31	21	10	11	13	1
14	14,16	13	29	24	11	11	12	1
14	12,14	13	29	23	11	13	13	1
14	11,14	13	29	25	11	13	13	1
14	11,15	13	29	24	11	13	13	2
15	12,14	14	30	23	10	13	13	1
14	13,15	12	28	23	10	11	13	1

Table 1 (*Continued*)

DYS19	DYS385I/II	DYS389I/I	DYS389I/II	DYS390	DYS391	DYS392	DYS393	*N*
14	10,14	14	30	22	10	13	12	1
14	12,13	14	30	24	10	14	13	1
15	13,18	13	29	23	10	11	12	1
14	11,14	13	29	22	10	13	13	1
15	13,18	13	30	23	11	11	12	1
17	17,19	13	30	21	10	11	15	1
13	16,16	14	33	24	10	11	12	1
14	11,14	13	29	24	10	13	14	1
13	13,14	13	29	24	9	11	13	1
14	12,14	13	29	25	10	11	14	1
13	17,18	13	30	24	10	11	13	1
15	14,14	12	30	23	10	12	14	1
13	14,14	14	30	24	10	11	12	1
16	13,17	12	28	25	10	11	12	1
17	12,17	13	30	24	11	11	13	1
14	11,14	13	30	24	10	13	13	1
13	13,14	14	30	24	9	11	13	1
14	11,14	13	29	23	11	13	13	1
14	11,14	14	31	25	10	13	12	1
14	11,14	13	29	24	11	13	12	2
15	11,14	13	29	24	11	13	13	1
14	13,17	13	31	23	10	11	12	1
13	17,18	13	31	25	10	11	13	1
13	16,19	13	30	24	10	11	13	1
14	13,13	14	30	23	10	11	12	1
14	11,14	13	29	25	11	12	13	1
14	13,17	13	30	23	10	11	12	1
16	14,15	12	29	21	10	11	14	1
16	12,12	13	28	23	10	11	13	1
16	12,12	13	28	23	11	11	13	1
14	14,18	13	31	22	10	11	12	1
13	16,18	13	30	24	10	11	13	1
14	11,15	14	30	25	11	13	13	1
Southeast								
14	11,14	13	30	23	11	13	13	1
13	16,17	13	30	23	10	11	13	1
15	13,15	13	30	23	10	11	12	1
13	12,12	13	30	24	10	11	13	1
14	13,15	12	28	22	10	11	13	1
14	11,12	12	29	24	11	14	13	1
14	11,14	13	29	24	11	13	13	2
15	13,15	12	30	21	10	11	14	1
15	11,14	13	29	24	11	13	13	1
15	13,13	14	32	22	10	11	14	1
14	13,14	12	28	22	10	11	13	1
13	11,14	13	30	26	11	13	13	1
15	11,16	13	30	24	11	13	13	1
15	10,19	14	30	22	10	14	13	1
14	12,15	13	29	25	10	13	13	1
15	13,18	13	31	24	10	11	12	1
15	14,14	12	29	22	10	11	14	1
14	11,15	14	30	24	11	14	13	1
14	11,12	12	28	24	10	13	13	1
15	15,18	13	31	23	10	11	14	1
14	11,15	13	29	25	11	13	11	1
13	19,20	12	29	24	10	11	13	1
14	12,18	12	28	24	10	11	12	1
14	13,14	13	29	22	10	11	13	1

Table 1 (*Continued*)

DYS19	DYS385I/II	DYS389I/I	DYS389I/II	DYS390	DYS391	DYS392	DYS393	*N*
13	14,15	14	31	24	9	11	13	1
14	11,15	15	31	24	10	13	13	1
13	16,16	14	33	24	10	11	12	1
14	11,14	13	29	22	11	13	12	2
14	11,14	13	29	24	12	13	13	1
14	13,18	13	31	23	10	11	12	1
14	12,16	13	30	23	10	11	12	1
14	12,14	13	31	24	11	13	13	1
15	13,14	11	27	22	10	11	13	1
14	12,14	13	29	22	11	13	13	1
South								
14	13,18	13	30	23	11	11	12	1
15	11,14	13	29	24	11	13	13	1
14	11,15	13	29	25	11	14	14	1
14	14,16	14	30	24	10	11	12	1
14	13,17	13	29	23	10	11	12	1
13	16,16	13	30	24	10	11	12	2
14	11,13	14	30	24	11	13	14	1
15	13,17	13	29	23	10	13	11	1
16	14,14	13	33	24	11	11	13	1
15	11,14	14	30	24	11	13	13	1
14	12,14	13	29	24	10	13	13	2
14	14,17	13	30	24	10	11	12	1
14	11,14	13	30	24	10	13	13	1
14	13,17	13	32	23	10	11	12	1
14	12,14	14	32	24	11	14	13	1
14	11,14	13	29	24	11	13	13	2
14	14,14	13	30	22	10	11	14	1
14	11,14	13	29	24	11	13	14	1
17	10,14	13	31	25	10	11	13	1
14	11,13	13	29	24	10	13	13	1
14	11,14	14	30	24	11	14	12	1
14	14,19	12	28	23	11	11	12	1
14	11,15	14	30	24	11	13	11	1
16	14,14	11	29	22	10	11	14	1
14	11,14	12	27	24	10	13	13	1
14	11,14	13	30	24	11	13	13	1
14	12,15	13	28	24	11	13	13	1
13	13,17	13	28	24	10	16	13	1
13	15,17	12	31	23	10	11	14	1
14	12,16	14	30	24	10	13	14	1
13	16,18	12	29	24	10	11	13	1
14	11,14	13	29	25	11	13	13	1
14	12,14	13	29	24	11	13	13	1
14	13,14	14	31	24	11	13	13	1
14	11,16	14	30	24	12	13	13	1
15	12,12	13	28	24	10	14	15	1
14	11,13	12	28	23	10	11	13	1
14	11,15	14	31	23	11	13	13	1
15	11,14	13	30	25	10	13	13	1
14	11,14	13	29	23	11	13	13	1
14	12,14	13	29	24	11	13	15	1
14	11,14	13	29	25	10	14	13	1
15	11,14	13	29	24	11	14	13	1
15	12,15	13	29	24	11	13	13	1

Table 2
Allele and DYS385I/II haplotype frequencies and gene diversities (GD) at nine Y chromosome STR loci in the five geopolitical regions and the total Brazilian population sample studied

Locus	North	Northeast	Central west	Southeast	South	Brazil
N	25	42	48	36	47	198
DYS19						
12	0,000	0,024	0,000	0,000	0,000	0,005
13	0,040	0,238	0,208	0,167	0,106	0,162
14	0,680	0,429	0,521	0,556	0,681	0,566
15	0,120	0,286	0,146	0,278	0,149	0,197
16	0,160	0,024	0,083	0,000	0,043	0,056
17	0,000	0,000	0,042	0,000	0,021	0,015
GD	0,496	0,676	0,655	0,586	0,501	0,611
DYS389I						
11	0,000	0,000	0,000	0,028	0,021	0,010
12	0,120	0,167	0,146	0,222	0,106	0,152
13	0,560	0,595	0,667	0,583	0,660	0,621
14	0,320	0,214	0,188	0,139	0,213	0,207
15	0,000	0,024	0,000	0,028	0,000	0,010
GD	0,570	0,572	0,498	0,590	0,507	0,548
DYS389II						
26	0,000	0,024	0,000	0,000	0,000	0,005
27	0,000	0,000	0,000	0,028	0,021	0,010
28	0,120	0,071	0,104	0,083	0,106	0,101
29	0,200	0,452	0,417	0,361	0,404	0,384
30	0,600	0,357	0,354	0,278	0,319	0,364
31	0,040	0,071	0,104	0,167	0,085	0,096
32	0,040	0,024	0,000	0,028	0,043	0,025
33	0,000	0,000	0,021	0,028	0,021	0,015
GD	0,582	0,657	0,679	0,755	0,714	0,700
DYS390						
21	0,080	0,024	0,063	0,028	0,000	0,035
22	0,160	0,071	0,063	0,278	0,043	0,111
23	0,120	0,190	0,250	0,167	0,191	0,192
24	0,600	0,548	0,479	0,444	0,638	0,540
25	0,040	0,143	0,146	0,056	0,021	0,086
26	0,000	0,024	0,000	0,028	0,106	0,035
GD	0,592	0,637	0,679	0,693	0,543	0,649
DYS391						
9	0,000	0,024	0,042	0,028	0,000	0,020
10	0,600	0,429	0,583	0,583	0,489	0,530
11	0,400	0,548	0,354	0,361	0,489	0,434
12	0,000	0,000	0,021	0,028	0,021	0,015
GD	0,480	0,515	0,533	0,529	0,522	0,530
DYS392						
11	0,360	0,357	0,542	0,500	0,319	0,419
12	0,080	0,071	0,042	0,000	0,000	0,035
13	0,520	0,500	0,375	0,417	0,532	0,465
14	0,040	0,071	0,042	0,083	0,128	0,076
15	0,000	0,000	0,000	0,000	0,000	0,000
16	0,000	0,000	0,000	0,000	0,021	0,005
GD	0,592	0,612	0,562	0,569	0,598	0,601
DYS393						
11	0,040	0,000	0,000	0,028	0,043	0,020
12	0,120	0,190	0,292	0,222	0,191	0,212
13	0,600	0,714	0,604	0,639	0,574	0,626

Table 2 (*Continued*)

Locus	North	Northeast	Central west	Southeast	South	Brazil
14	0,200	0,095	0,083	0,111	0,149	0,121
15	0,040	0,000	0,021	0,000	0,043	0,020
GD	0,582	0,445	0,543	0,529	0,608	0,548
DYS385I/II						
10,14	0,000	0,024	0,021	0,000	0,021	0,015
10,19	0,000	0,000	0,000	0,028	0,000	0,005
11,11	0,080	0,000	0,021	0,000	0,000	0,015
11,12	0,040	0,048	0,000	0,056	0,000	0,025
11,13	0,000	0,024	0,000	0,000	0,064	0,020
11,14	0,360	0,262	0,229	0,222	0,298	0,268
11,15	0,040	0,095	0,063	0,083	0,064	0,071
11,16	0,000	0,000	0,042	0,028	0,021	0,020
11,17	0,000	0,024	0,000	0,000	0,000	0,005
12,12	0,000	0,000	0,042	0,028	0,021	0,020
12,13	0,000	0,024	0,021	0,000	0,000	0,010
12,14	0,000	0,048	0,063	0,056	0,106	0,061
12,15	0,040	0,024	0,000	0,028	0,043	0,025
12,16	0,040	0,000	0,000	0,028	0,021	0,015
12,17	0,040	0,000	0,021	0,000	0,000	0,010
12,18	0,000	0,024	0,000	0,028	0,000	0,010
13,13	0,000	0,024	0,021	0,028	0,000	0,015
13,14	0,040	0,048	0,042	0,083	0,021	0,045
13,15	0,040	0,024	0,021	0,083	0,000	0,030
13,16	0,040	0,024	0,000	0,000	0,000	0,010
13,17	0,000	0,024	0,063	0,000	0,085	0,040
13,18	0,000	0,024	0,042	0,056	0,021	0,030
14,14	0,040	0,000	0,042	0,028	0,064	0,035
14,15	0,000	0,000	0,021	0,028	0,000	0,010
14,16	0,000	0,024	0,021	0,000	0,021	0,015
14,17	0,040	0,048	0,000	0,000	0,021	0,020
14,18	0,000	0,000	0,021	0,000	0,000	0,005
14,19	0,000	0,000	0,000	0,000	0,021	0,005
15,15	0,040	0,000	0,000	0,000	0,000	0,005
15,17	0,000	0,000	0,000	0,000	0,021	0,005
15,18	0,000	0,024	0,000	0,028	0,000	0,010
16,16	0,040	0,071	0,021	0,028	0,043	0,040
16,17	0,000	0,000	0,021	0,028	0,000	0,010
16,18	0,040	0,024	0,021	0,000	0,021	0,020
16,19	0,000	0,000	0,021	0,000	0,000	0,005
17,17	0,000	0,000	0,042	0,000	0,000	0,010
17,18	0,040	0,048	0,042	0,000	0,000	0,025
17,19	0,000	0,000	0,021	0,000	0,000	0,005
19,20	0,000	0,000	0,000	0,028	0,000	0,005
GD	0,842	0,898	0,917	0,910	0,872	0,905

five regions together were analyzed, resulting in an estimate of combined power of individual discrimination of 87% (Table 4). In a recent study, a high haplotype diversity (99.14%) was found for a different set of eight Y-STR loci in a population sample of 67 Brazilian Caucasians [4].

9. Other remarks

An AMOVA indicated that 99.97% of the haplotypic variation is found within geopolitical regions and only 0.03% between regions. No significant differentiation for the Y-STR haplotype was seen among populations from the five geopolitical regions based on an exact test and bootstrap resampling (Fst = 0.00031; *P*-value = 0.43), corroborating previous reports that also found no significant differentiation based on 12 unique event polymorphisms [5]. This result indicates that a single national database of Y-STR haplotypes can be used in the quantitative assessment of matches in forensic casework in Brazilian populations. The haplotype (14–11–14–13–29–24–11–13–13, frequency = 0.025) was the most frequent in the Brazilian population sample

Table 3
Nine (DYS19, DYS385I/II, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393) Y-STR locus haplotype counts in the five geopolitical regions and in the total Brazilian population sample. Listed only the haplotypes observed twice or more

Haplotype	North	Northeast	Central west	Southeast	South	Brazil
14-11,14-13-29-24-11-13-13	0	1	0	2	2	5
15-11,14-13-29-24-11-13-13	0	0	1	1	1	3
14-11,14-13-29-24-10-13-13	2	1	0	0	0	3
14-11,14-13-29-24-11-13-12	0	1	2	0	0	3
14-11,14-14-30-24-11-13-13	1	1	0	0	0	2
15-11,14-13-30-24-10-13-13	1	1	0	0	0	2
14-13,14-12-28-22-10-11-13	1	0	0	1	0	2
15-13,13-14-32-22-10-11-14	0	1	0	1	0	2
13-16,18-13-30-24-10-11-13	0	1	1	0	0	2
14-11,14-13-29-24-11-13-14	0	1	0	0	1	2
15-13,18-13-29-23-10-11-12	0	1	1	0	0	2
13-17,17-12-29-24-10-11-13	0	0	2	0	0	2
14-11,14-13-29-25-11-13-13	0	0	1	0	1	2
14-11,15-13-29-24-11-13-13	0	0	2	0	0	2
13-16,16-14-33-24-10-11-12	0	0	1	1	0	2
14-11,14-13-30-24-10-13-13	0	0	1	0	1	2
14-11,14-13-29-23-11-13-13	0	0	1	0	1	2
14-11,14-13-29-22-11-13-12	0	0	0	2	0	2
13-16,16-13-30-24-10-11-12	0	0	0	0	2	2
14-12,14-13-29-24-10-13-13	0	0	0	0	2	2
Haplotypes with single occurrence	20	33	35	28	36	152
Total	25	42	48	36	47	198

Table 4
Summary of Y-chromosome minimal STR haplotype information based on nine STR loci in the five geopolitical regions of Brazil

Number of individuals with same haplotype	Brazil	North	Northeast	Central west	Southeast	South
5	1					
3	3					
2	16	1		3	2	3
1	152	23	42	42	32	41
Number of different haplotypes (*H*)	172	24	42	45	34	44
Number of individuals (*N*)	198	25	42	48	36	47
Power of individual discrimination (*H*/*N*)	0.87	0.96	1.00	0.94	0.94	0.94
Haplotype diversity	0.998	0.997	0.955	0.933	0.963	0.935

analyzed (Table 3). The same haplotype was the most frequent in a worldwide population sample of 20,669 haplotypes involving a set of 181 populations, with 513 matches found (frequency = 0.0248), and also the most frequent, with 408 matches (frequency = 0.029), in an European population sample of 13,990 haplotypes (Y-STR database at http://www.ystr.org). It was also the most frequent in populations from Central Portugal [6] and Spain [7,8]. On the other hand this same haplotype was not found (0 matches) in an African population sample of 328 haplotypes in a set of 4 populations. This observation is in line with previous reports based on biallelic Y polymorphisms [5], of strong directional mating between European males and Amerindian and African females in the formation of the current Brazilian population. This paper follows the guidelines for publication of population data requested by the journal [9].

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