International Congress Series 1288 (2006) 240–242

www.ics-elsevier.com

Y-chromosome variation in Swedish, Saami and Österbotten male lineages

A. Karlsson [a,*], A. Götherström [b], T. Wallerström [c], G. Holmlund [a]

[a] *The National Board of Forensic Medicine, Department of Forensic Genetics, University Hospital, SE-581 85 Linköping, Sweden*
[b] *Department of Evolutionary Biology, Uppsala University, Sweden*
[c] *Department of Archaeology and Ancient History, University of Lund, Sweden*

Abstract. We have analysed 383 unrelated males from Sweden ($n=305$), Österbotten, in Finland ($n=40$) and a Saami population ($n=38$). All samples were typed for 16 Y-chromosomal biallelic markers and nine Y-chromosomal short tandem repeats (Y-STRs). Analysis of both haplogroup frequencies and haplotypes revealed a minor stratification within Sweden. The northern Swedish region Västerbotten differed significantly ($P<0.05$) from other Swedish regions both comparing haplogroup frequencies and pairwise R_{ST} values. A flow of Y-chromosomes into Västerbotten from mainly Saami and Finnish populations, in higher degree than to other Swedish regions, could explain this stratification.

Keywords: Y-chromosome; Y-SNP; Y-STR; Sweden

1. Introduction

Data available on Y-chromosomes in Sweden, based on a limited number of markers, indicate little or no variation between different regions [1,2]. To further investigate the Y-chromosomal variation in Sweden, we have used additional markers and methods well suited for comparing the information given by Y-chromosomes from different geographical regions.

2. Materials and methods

305 unrelated males from seven geographically different regions in Sweden were collected for this study [1]. For reference, 38 male samples from a Swedish Saami

* Corresponding author. Tel.: +46 13223036; fax: +46 13136005.
E-mail address: andreas.karlsson@rmv.se (A. Karlsson).

0531-5131/
doi:10.1016/j.ics.2005.09.107

Table 1
Primers used for typing of biallelic markers were designed using Assay Design Software (Biotage)

Marker	Forward primer[a] (5′→3′)	Reverse primer[a] (5′→3′)	Sequencing primer (5′–3′)
M26[b]	*CCAAGATTCAATTTTTTTCTGAATTA	TGACGAAATCTGCAGCAAAAAT	GGCCATTCAGTGTTCTC
M35[b]	*ATGGTCCCTTTCTATGGATAGCA	TGAACAACTAATCCATGCAGACTT	TCGGAGTCTCTGCCTGT
M78[b]	*CGACATGAACACAAATTGATACAC	GCAAGTACTATGACCAGCTTATTTTG	GAAATATTTGGAAGGGC
M89[b]	CCTGGATTCAGCTCTCTTCCTAAG	*TGCAACTCAGGCAAAGTGAGA	CCTAAGGTTATGTACAAAAA
M170[b]	GTTTTCATATTCTGTGCATTATACAA	*AGGTCCTCATTTTACAGTGAGACA	TTACTTAAAAATCATTGTTC
M269[b]	CACAATAGAAGGGGAATGATCAGGGT	*GTGTGCCTTCTGAGGCACATATGAT	TGATCAGGGTTTGGTTAA
M201[b]	*ATCTAATAATCCAGTATCAACTGAGG	CGACGTATCTGAGGTTCAAATC	ACTAAGTACCTATTACGAAA
M223[b]	*GCTGGAGTCTGCACATTGAT	AGGCAAGTATGCCGCTATAAA	AGATGATGCAATTTATTT
M253[b]	GATGCTCAGCTAACTAGTCCTGTTTA	*CATTCAATGAAGAACCTGGAGA	TTGTTGATAGATAGCAAGTT
M9[b]	*TCAGGACCCTGAAATACAGAACT	TTGAAGCTCGTGAAACAGATTAG	AACATGTCTAAATTAAAGA
TAT[b]	GACTCTGAGTGTAGACTTGTGA	*CTTGCTGTGCTCTGAAATATTAAATTAA	TCTGAGTGTAGACTTGTGAA
M17[b]	GTGGTTGCTGGTTGTTACGG	*AGCTGACCACAAACTGATGTAGA	TGCTGGTTGTTACGG
SRY[b] 10831	*TCATTCAGTATCTGGCCTCTTG	CACCACATAGGTGAACCTTGAA	GGTGAACCTTGAAAATGTT
92R7[b]	*TGCATGAACACAAAAGACGTA	GCATTGTTAAATATGACCAGC	TGACCAGCAAAGACAA
YAP[c]	GGACTAGCAATAGCAGGGGAAGATA	TTCCAGGGCCAACTCCAA	–
12f2[d]	TGAGAACCACAAATTGGATGC	CTGAGGCTGTTGGTTCACTG	–

[a] The asterisk (*) represents the biotin molecule needed for the Pyrosequencing assay.

[b] Reaction was carried out in a volume of 25 μl with final concentrations as follows: 1× PCR buffer II (Applied Biosystems), 3.75 mM $MgCl_2$, 200 μM each of dATP, dCTP, dGTP, 400 μM of dUTP, 0.016% BSA, 2% glycerol, 400 nM of each primer, 0.05 U of uracil–DNA glycosylase (USB corporation), 1.25 U of Ampli*Taq* Gold DNA polymerase (Applied Biosystems) and 10 ng template DNA. The amplification programme was: 10 min at 37 °C, 5 min at 95 °C followed by 50 cycles of 1 min at 94 °C, 1 min at 56 °C, 1 min at 72 °C with a final extension for 10 min at 72 °C and 10 min at 94 °C.

[c] The PCR conditions were the same as above except that 500 μM each of dATP, dCTP, and dGTP, and 1 mM of dUTP were used and 200 nM of each primer. Cycling conditions deviated in that 40 cycles were used and the annealing temperature was at 60 °C.

[d] Expand Long Template PCR System (Roche) was used. 10 ng template DNA was added to 1× PCR buffer 3, 500 μM of each dNTP, 80 nM of each primer, 2 units of polymerase mix and water to a final volume of 25 μl. The PCR programme was: 2 min at 94 °C followed by 35 cycles of 15 s at 94 °C, 30 s at 56 °C, 6 min at 68 °C with a final extension for 7 min at 68 °C.

population (Jokkmokk nomads) and 40 samples from Österbotten in Finland were also analysed.

Sixteen different biallelic markers were used in order to define the major male lineages. Markers M9, TAT, 92R7, M17, M35, M78, M89, M201, M170, M26, M223, SRY10831, M253, and M269 were typed using Pyrosequencing technique, while YAP and 12f2 were typed using agarose gel electrophoresis (details are shown in Table 1). An additional nine STRs (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a and DYS385b) were typed as described elsewhere [3].

Measures of genetic variation, and associated probability values, were estimated using ARLEQUIN v 2.0 [4]. Principal component analyses (PCA), based on haplogroup frequencies and pairwise R_{ST} values, were performed using MATLAB 7.0 software (MathWorks).

3. Results

Thirteen different haplogroups were found among the Swedish, Saami and Österbotten males. Haplogroup I1a* was the most common in Sweden while N3 was the most frequent haplogroup in both the Österbotten and the Saami lineages.

A PCA on haplogroup frequencies revealed that Västerbotten tend to cluster outside other Swedish regions and also closer to the Saami population (Fig. 1a). An exact test for population differentiation confirmed that Västerbotten significantly differed ($P<0.01$) from all the other Swedish regions combined when haplogroup frequencies were compared.

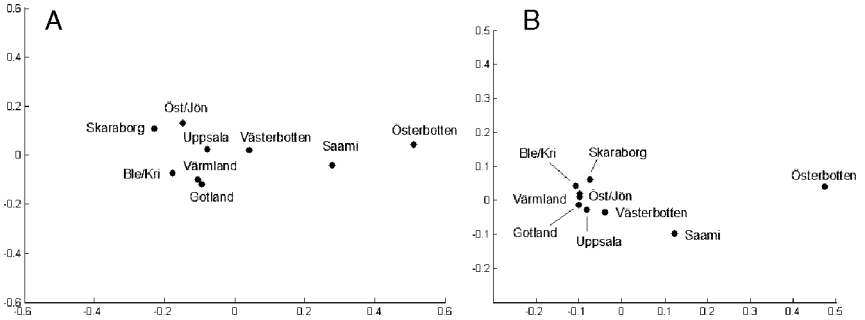

Fig. 1. Principal component analysis of the genetic relations between seven Swedish regions, a Saami population and a Österbotten population: (A) relationships based on Y haplogroup frequencies; (B) plot based on pairwise R_{ST} values obtained from nine Y-STRs. Ble/Kri=Blekinge/Kristianstad; Öst/Jön=Östergötland/Jönköping.

Fig. 1b shows a PCA on pairwise R_{ST} values based on Y-chromosome haplotype data from nine Y-STRs. A minor difference was revealed when Västerbotten was compared with haplotypes from all the other Swedish regions ($R_{ST}=0.02$, $P<0.05$). This was also true for Skaraborg ($R_{ST}=0.02$, $P<0.05$).

4. Discussion

Y-chromosomal markers used in this study revealed some minor differences among seven Swedish regions. Västerbotten significantly differed from other Swedish regions both comparing haplogroups and haplotypes.

Analysis showed that the high frequencies of N3 and I1c, and a higher exchange of Y-chromosomes from Saami and Finnish populations, can partly explain why Västerbotten clustered outside other Swedish regions.

Acknowledgements

We thank M. Hedman and L. Beckman for the collection of Österbotten and Saami samples, respectively. This study was partly funded by The Board for Research and Development, The National Board of Forensic Medicine, Sweden (grant no. 55217), Swedish National Laboratory of Forensic Science and Forensic Science Centre at Linköping University.

References

[1] G. Holmlund, et al., No obvious geographical Y-chromosome gradient in the Swedish population, in: C. Doutremépuich, N. Morling (Eds.), Progress in Forensic Genetics 10, International Congress Series, vol. 1261, 2004, pp. 357–359.
[2] L. Roewer, et al., Signature of recent historical events in the European Y-chromosomal STR haplotype distribution, Hum. Genet. 116 (2005) 279–291.
[3] G. Holmlund, et al., Y-chromosome STR haplotypes in Sweden. Forensic Sci. Int. (in press).
[4] P. Schneider, D. Roesseli, L. Excoffier, ARLEQUIN, version 2.000: A Software for Population Genetics Data Analysis, Genetics and Biometry Laboratory, University of Geneva, Geneva, 2000.