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No obvious geographical Y-chromosome gradient in the Swedish population

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Abstract. We have analysed 305 Y-chromosomes from seven regions in Sweden to find out if there is population stratification. The Y-STR-minimal haplotype markers including DYS385 a and b were analysed and haplogroups were defined by the SNPs TAT, 92R7, M9, M17 and SRY1532. The STRs were analysed on an ABI377 and the SNPs by Pyrosequencing. Y-haplogroups 1, 2, 3, 16 and 26 were found in approximately equal frequencies in the sub-populations. Also, the gene diversity within the STR-haplotypes, analysed by the Markov chain algorithm, showed no differences. Thus, the whole database can be used for frequency calculations.

Keywords: Y-STR; Y-SNP; Haplotypes; Haplogroups; Population

1. Introduction

The Y-STR databases, including regional data, give invaluable means for the calculation of match probabilities. However, there is an apparent risk for an overestimation of the significance of a match, if there are no data on the genetic stratification of the population.

2. Materials and methods

2.1. Sub-populations and blood samples

Seven regions, shown in Fig. 1, were selected according to the demographic history of the Swedish people [1,2].

Three-hundred-five blood samples were collected, of which 109 were men from paternity cases, with known place of birth and typical Swedish names. The rest (196) were from blood donors having given their written consent and questioned about their origin.

2.2. Y-chromosome markers

The STR-markers DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393 and DYS385 were analysed as described earlier [3]. The DYS385 was separated into DYS385a and DYS385b according to Kittler et al. [4]. All fragment lengths were analysed on an ABI377.

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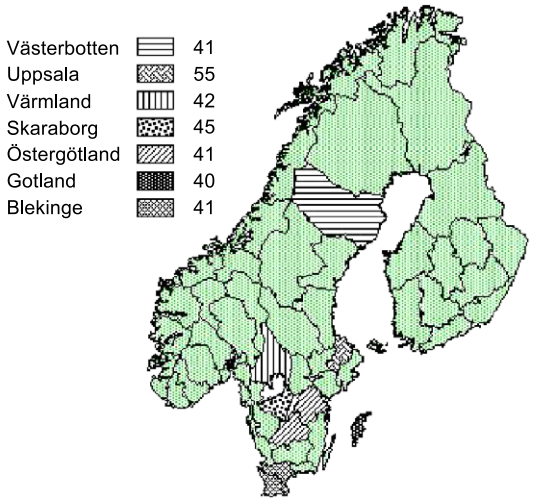

Fig. 1. Map of Sweden showing the number and the geographical origin of the Y-chromosomes.

The SNPs TAT, 92R7, M9, M17 and SRY1532 (=SRY10831) were amplified according to Thomas et al. [5]. For the Pyrosequencing [6] one primer was biotinylated and the amplicons purified by streptavidine bead binding filters. The Pyrosequencing strategy is shown in Table 1. In 96 samples, SNP-detection was also done with Denaturing High-Performance Liquid Chromatography (DHPLC) [7].

2.3. Statistical analyses

Arlequin [8] was used for population comparisons based on haplogroup data. The analysis is based on a Markov chain algorithm to estimate the gene diversity for the STRs within the haplogroups for each population.

3. Results

Twenty-eight Y-STR-haplotypes were found more than once. The most frequent haplotype, 14,12,28,23,10,11,13,14a,14b (16/305, 0.0524), was evenly distributed in six of the seven sub-populations, but missing in Östergötland. The other relatively frequent haplotypes (4-7/305) were also evenly distributed. For the more rare haplotypes (2-3/305), however, the distribution differs in that they are found within the same region, which shows relatedness between the men. Separating DYS385 into a and b elevated the power of discrimination from 75% to 79%.

Table 1
Polymorphisms and *primer-sites* used for Pyrosequencing of Y-SNPs

Marker	Sequence
TAT	CTGAGACTCACATCTGAACACTTAAGT T/C CAACAAAATTAAATTATAAAGT
92R7	TGCATGAACACAAAAGACGTAGAAG T/C TTGTCTTTGCTGGTCATATTTAACAATGC
M9	AAAGAAACGGCCTAAGATGGTTGAAT G/C CTCTTTATTTTTCTTTAATTTAGACATGTTCAA
M17	ACCAACGACCAACAATGCCCG G/- AAAAAAATTCACTTAAAACCCCAAACAATTC
SRY1532	GAAGACTGAAAAAGTGTGTCA G/A ATTGTAAAAGTTCCAAGTGG

Table 2
Y-STR gene diversity within each haplogroup in the sub-populations

Haplogroup	Västerbotten	Uppsala	Värmland	Skaraborg	Östergötland	Gotland	Blekinge
2	0.9532	0.9717	0.9625	0.9667	0.9641	0.9267	0.9640
3	(1)	0.8650	0.8101	0.7619	0.8415	0.8099	0.8650
1	0.8822	0.8959	0.8653	0.9222	0.8946	0.8431	0.8657
16	0.8657	(1)	(1)			0.7636	
26	0.7627	0.7636	0.6829	(2)			(2)

The standard error varied between 0.008 and 0.0161.
(1) Not calculated, only two haplotypes; (2) not calculated, only one haplotype.

The haplogroups were assigned according to Rosser et al. [9] and Weale et al. [10]. The relative haplogroup frequencies in the whole population were for hg3 0.118, hg2 0.600, hg1 0.190, hg16 0.049 and hg26 0.043. There were no significant differences between the geographical regions although hg16 is missing in Östergötland, Skaraborg and Blekinge and hg26 in Värmland, Gotland and Östergötland. Interestingly, hg3 is about half as frequent as in other European populations. SRY1532 is closely associated with DYS385a-11 and DYS385b-14 as shown by Kittler et al. [4]. Only minor gene diversity differences between the sub-populations were found, as shown in Table 2.

4. Conclusion

There were no statistical differences between the sub-populations, regarding both haplotype and haplogroup distributions. The Swedish population is accordingly not stratified and the complete database can be used for Y-chromosome STR-haplotype-frequency calculations.

There can, however, be some risk for an overestimation of match probabilities for the more rare haplotypes since they can locally be more frequent than in the whole population.

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References

[1] S. Erixon, Atlas över svensk folkkultur. Uppsala, 1957.
[2] S. Erixon, Svensk byggnadskultur. Lund, 1982.
[3] G. Holmlund, et al., Y-chromosome STR haplotypes in a Swedish population, in: B. Brinkmann, A. Carracedo (Eds.), Progress in Forensic Genetics, vol. 9, Elsevier, Amsterdam, 2003, pp. 343–348.
[4] R. Kittler, et al., Apparent intrachromosomal exchange of the Y chromosome explained by population history, Eur. J. Hum. Genet. 11 (4) (2003) 304–314.
[5] M.G. Thomas, N. Bradman, H.M. Flinn, High throughput analysis of 10 microsatellite and 11 diallelic polymorphisms on the human Y-chromosome, Hum. Genet. 105 (1999) 577–581.
[6] M. Ronaghi, et al., Real-time DNA sequencing using detection of pyrophosphate release, Anal. Biochem. 242 (1996) 84–89.
[7] P.A. Underhill, et al., Detection of numerous Y chromosome biallelic polymorphisms by denaturing high-performance liquid chromatography, Genome Res. 7 (1997) 996–1005.
[8] Arlequin, http://www.antrhro.unige.ch/arlequin.
[9] H.Z. Rosser, et al., Y-chromosomal diversity in Europe is clinal and influenced primarily by geography, rather than by language, Am. J. Hum. Genet. 67 (2000) 1526–1543.
[10] M.E. Weale, et al., Armenian Y chromosome haplotypes reveal strong regional structure within a single ethno-national group, Hum. Genet. 109 (2001) 659–674.