

Y-chromosomal composition of mediaeval and contemporary populations in Norway and adjacent Scandinavian countries: Y-STR haplotypes and the rare Y-haplogroup Q

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Background

The project "Immigration and mobility in mediaeval and post-mediaeval Norway" is an international multidisciplinary research collaboration and is part of the Norwegian Research Council's ten-year research programme "SAMKUL" (Cultural conditions underlying social change).

It aims to combine historical information with archeological, osteological, isotopic and genetic data obtained from mediaeval (Norway-M) and post-mediaeval (Norway-PM) skeletal remains excavated in Trondheim, one of the mediaeval capitals of Norway.

The molecular genetic part of the project aims to investigate potential genetic similarities between the pre-modern sample and contemporary populations in Norway and neighboring European regions by analyzing Y-chromosomal markers.

The male-specific portion of the Y-chromosome is passed directly from fathers to male offspring. Typically, Short Tandem Repeats (STRs) and SNPs are typed when Y-chromosomal DNA is investigated for population genetic purposes.

Sets of Y-STRs can be combined to haplotypes (HTs), that can be directly compared between samples of different geographic origins. Genetic distances (e.g. Rst values) of the haplotypes between different populations may shed light on the phylogenetic and historic background of these populations.

Samples

Molecular genetic analyses were performed on 97 pre-modern human remains (see Figure 1) including molecular genetic sexing and Y-chromosomal DNA typing.

All samples were subjected to molecular genetic analyses of the sex using "Genderplex" consisting of two different regions of the amelogenin gene, SRY and four X-STR loci [Esteve et al. 2009 IJLM 123: 459-64]. From 90% of the extracted remains (n=87) sex assignment was possible. Of these, 49 (56.3%) were genetically assigned as males.

DNA extracts were subjected to Y-STR analysis using Yfiler Plus PCR Amplification Kit (Thermo Fisher Scientific) and/or PowerPlex Y23 System (Promega). From 35 samples at least 16 Y-STR markers included in the Yfiler-Kit were successfully typed (29 Norway-PM samples and 6 Norway-M samples). This set was used for AMOVA and network analysis (Network 5, fluxus-engineering.com) presented here. For YHRD searches additional samples with partial profiles were used (n=47).

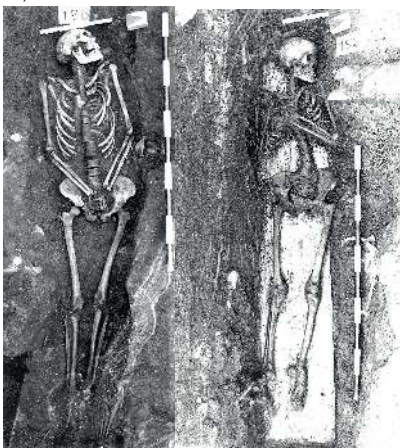


Figure 1: Examples of two individual graves. The skeletal remains were excavated in Trondheim, one of the mediaeval capitals of Norway. The photos were kindly provided by the Norwegian Directorate for Cultural Heritage.

Summary

- Medieval and post-mediaeval human remains excavated in Trondheim, Norway, were analysed for Y-STR haplotypes (HTs).
- The Y Chromosome Haplotype Reference Database (YHRD) provided contemporary HTs for AMOVA and database searches.
- Comparisons between pre-modern and contemporary HTs confirmed a high degree of similarity with present day Norwegians.
- One sample belonged to Y-HG-Q which is frequent in Native Americans but rare and less scientifically documented in Europe.
- SNP analysis proved its affiliation to HG Q-L804 a sister group to the main Native American HG Q-M3.
- Network analysis suggested that HG-Q exists in low frequencies in contemporary Norway.
- Our results give indications for a genetic continuity of the male Norwegian population during the last centuries.

Y-Haplogroup Q in historic and contemporary Norway

An assessment of the Y-chromosomal haplogroups (HGs) based on Y-HTs was performed as quality check (Athey's HG prediction tool). As expected most of the samples were attributable to the main European HGs I, R1a and R1b. However, one of the HTs seemed to be associated with HG-Q which is rare in Europe and hitherto little evaluated in this region. This general picture was also reflected by network analysis of Norway-PM and Norway-M HTs showing three clusters most probably attributable to the above mentioned European HGs. The sample assigned to HG-Q is clearly separated (see Figure 2). Further SNP analysis proved that this sample belongs to HG Q-L804 (Q1a2a1a2). This sub HG is a sister group to HG Q-M3 (Q1a2a1a1) the main subclade of Native Americans.

Network analysis was applied for detecting HTs similar to the HG-Q sample in different contemporary populations of Norway stored in the YHRD. For this analysis the HT of the Q-L804 sample was added to the queried YHRD population sample. Again, this HT lies always outside the main clusters. However, as shown in the network for the population sample "Central Norway" (n=285) there is evidence for similar HTs in contemporary Norway which may be considered as HG-Q candidate samples (see Figure 3).

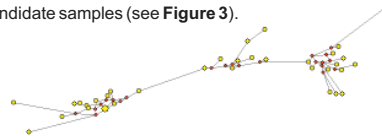


Figure 2: Network of Norway-M and Norway-PM with at least 16 typed Y-STRs. The sample belonging to HG Q-L804 is shown in blue.

Searching Y-Haplotypes in contemporary populations

A detailed comparison between mediaeval/post-mediaeval and contemporary Y-chromosomes was performed by searching the obtained haplotypes (HTs) in the Y Chromosome Haplotype Reference Database (YHRD: <https://yhrd.org>) comprising 154,329 haplotypes from 991 populations in 129 countries at the time of query (Release 50). YHRD searches of the pre-modern haplotypes yielded full matches and neighbor-matches differing at only one allele from the query HT. Table 1 shows the number of the historic Norwegian samples that match at least one HT of the whole YHRD and of the YHRD-Norway subsample. Table 2 shows the same for the neighbor matches.

Table 1

Samples	n	% (all)	%(of matching samples)
Total	47	100	
matching in YHRD	30	64	100
matching YHRD Norway	22	47	73

Table 2

Samples	n	% (all)	%(of matching samples)
Total	47	100	
matching in YHRD	43	91	100
matching YHRD Norway	35	74	81

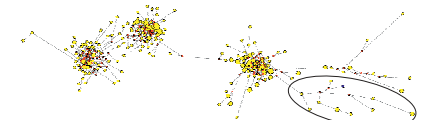
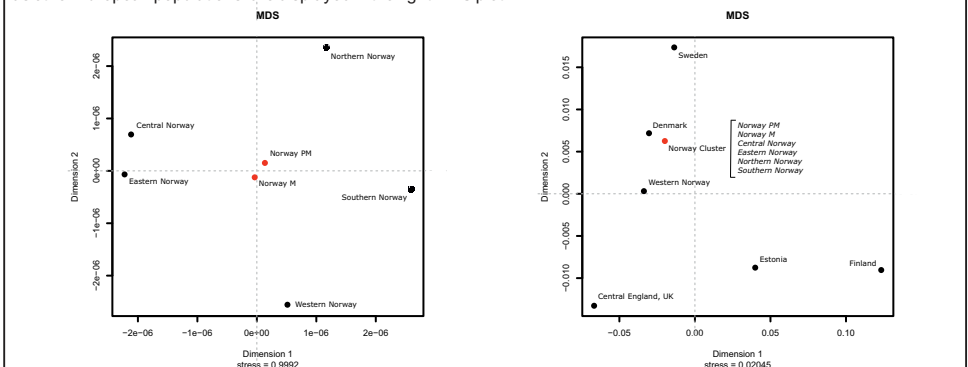


Figure 3: Network of the contemporary YHRD population "Central Norway". The historic Q-L804 sample was added (blue) and samples considered as HG Q candidates are encircled.

AMOVA

AMOVA was performed using the YHRD online tool on pairwise Rst values to create the corresponding MDS plots. The pre-modern HTs were compared to contemporary populations from Scandinavian and other European populations.

Both pre-modern populations showed had small genetic distances to contemporary Norwegians from different regions of the country (left MDS plot). Almost all Norwegian population samples showed no significant genetic inter-population variation in a separate AMOVA analysis. The distances between this "Norway Cluster" (Rst threshold 0.01) and other Scandinavian as well as other European populations are displayed in the right MDS plot.



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