

# Evidence for mtDNA Admixture between the Finns and the Saami

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## Key Words

Population genetics · mtDNA haplotypes · Admixture

## Abstract

**Objectives:** The Finns, and to a more extreme extent the Saami, are genetic outliers in Europe. Despite the close geographical contact between these populations, no major contribution of Saami mtDNA haplotypes to the Finnish population has been detected. **Methods:** To examine the extent of maternal gene flow from the Saami into Finnish populations, we determined the mtDNA variation in 403 persons living in four provinces in central and northern Finland. For all of these samples, we assessed the frequencies of mtDNA haplogroups and examined sequence variation in the hypervariable segment I (HVS-I). The resulting data were compared with published information for Saami populations. **Results:** The frequencies of the mtDNA haplogroups differed between the populations of the four provinces, suggesting a distinction between northern and central Finland. Analysis of molecular variance suggested that the Saami deviated less from the population of northern Finland than from that of central Finland. Five HVS-I haplotypes, including that harboring the Saami motif and the Asian-specific haplogroup Z, were shared between the Finns

and the Saami and allowed comparisons between the populations. Their frequency was highest in the Saami and decreased towards central Finland. **Conclusions:** The high frequency of certain mtDNA haplotypes considered to be Saami specific in the Finnish population suggests a genetic admixture, which appears to be more pronounced in northern Finland. Furthermore, the presence of haplogroup Z in the Finns and the Saami indicates that traces of Asian mtDNA genotypes have survived in the contemporary populations.

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## Introduction

The oldest archeological evidence of settlement in Finland dates back approximately 9,000 years. The origin of these people is unknown, but they could have been ancestors of the Saami [1]. They subsisted on hunting, fishing and gathering, and probably came from the area of the Ural Mountains. The second wave of settlers arrived in the southern parts of Finland around 5,500 years before present. Permanent settlement extended across southern Finland and the coast and riversides of Ostrobothnia in the 16th century, but the forest areas of northern and eastern Finland still had no resident population [2]. There-

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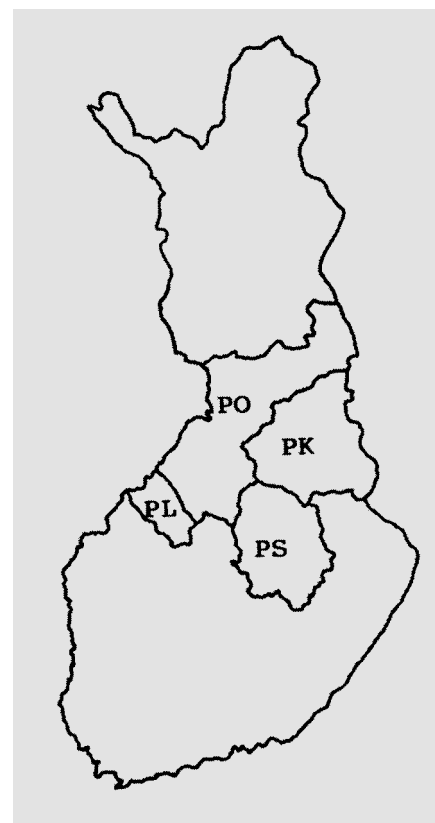
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after a major colonization of these areas began, with people mainly from southern Savo being directed to them by the Crown authorities.

The Finns are considered to be one of the outliers on the genetic map of Europe [3]. Geographic and cultural isolation has greatly shaped the Finnish gene pool. For example, certain recessive diseases are prevalent there even if they are infrequent elsewhere [2]. Furthermore, previous studies on the sequence variation in the Y chromosome and mitochondrial DNA (mtDNA) have shown high homogeneity in the Finnish population [4–8]. An Asian-specific polymorphism is quite frequent in the Finnish Y chromosome pool [8], whereas a clear West Eurasian pattern of polymorphisms has been detected in mtDNA [4–7]. About 40% of the Finns belong to mtDNA haplogroup H, which is also common among other Europeans [7, 9]. Other common haplogroups among the Finns include haplogroup U, with a frequency of 16%, haplogroup J, with a frequency of 14%, and haplogroup T, with a frequency of 6%. The remaining haplogroups (I, K, M, V, W, X) are less common, each with a frequency below 4% [7].

The Saami are a small ethnic group living in the northern parts of Finland, Sweden, Norway and northwestern Russia. Genetically they are the most unique population in Europe and are sharply differentiated from an otherwise homogeneous set of European populations [3, 10–13]. In particular, Saami mtDNA show restricted sequence variation, with many of them being characterized by a motif defined by the nucleotide variants 16144T→C, 16189T→C and 16270C→T that belong to subcluster U5 [10, 14]. Because of the high frequency of these U5 mtDNA in Saami populations (32–52%) and their near absence in other European populations [10], it is thought that they originated in the Saami, with the series of mutations defining U5 being called the ‘Saami motif’. The majority of the remaining Saami mtDNAs exhibited a high frequency of the 16298T→C nucleotide variant.

Previous studies have failed to demonstrate any major contribution of the Saami mtDNA haplotypes to the Finnish population, although the settlers and the original population probably lived in close contact during the colonization of Finland and members of the original population probably acculturated with the settlers [1]. Signs of such contacts would be most probable in regions that have been colonized by the Finns during recorded history. To identify the maternal genetic contribution of the Saami to the Finnish population, we collected samples from four neighboring provinces in central and northern



**Fig. 1.** Map of Finland showing the provinces of Northern Ostrobothnia (PO), Kainuu (PK), Northern Savo (PS) and Central Ostrobothnia (PL).

Finland, areas that differ in their population history but are currently devoid of a Saami population. For all of these samples, we assessed the frequencies of mtDNA haplogroups and examined sequence variation in the hypervariable segment I (HVS-I). The resulting data were compared with published information for Saami populations.

## Subjects and Methods

### *Subjects and Samples*

A total of 403 samples from healthy blood donors (242 men, 161 women) were obtained at the Finnish Red Cross offices in the capitals of the provinces of Northern Ostrobothnia, Kainuu, Northern Savo and Central Ostrobothnia (fig. 1). Because the population of Finland is concentrated in the southern part of the country, the provinces of Northern Ostrobothnia and Kainuu will represent northern Finland, while the provinces of Northern Savo and Central Ostrobothnia will represent the central part. The province of Lapland, lying north of Northern Ostrobothnia (fig. 1), was not used as a

source of samples because it was not possible to exclude blood donors who might have immediate maternal ancestors in the Saami population.

The mean age of the donors was  $41 \pm 12$  years (range 18–64 years). The donors and their mothers were required to be free of the common manifestations of mitochondrial diseases, such as diabetes mellitus, hearing impairment and neurological ailments. In addition, it was required that the donors and their mothers had been born in the same province. After obtaining this information, the samples were anonymized. The generation time in the population of the provinces of Northern Ostrobothnia and Kainuu has recently been calculated to be 28.4 years [15], suggesting that the mothers of the blood donors had been born between the years 1906 and 1952. The research protocol was approved by the Ethics Committee of the Medical Faculty, University of Oulu, and the Ethics Committee of the Finnish Red Cross.

Previously published Saami sequences were obtained from the EMBL data library under accession numbers X90001–X90115 [10].

#### *Analysis of mtDNA Haplogroups and Haplotypes*

Total DNA was isolated from blood cells using the QIAamp Blood Kit (Qiagen, Hilden, Germany), and mtDNA haplogroups were determined by restriction fragment analysis to identify the most informative polymorphic sites [7], except that 12308A→G was detected by *Dde*I digestion of a fragment amplified in the presence of a mismatched (underlined nucleotide) forward primer 12279–12307 (5'-AAC AGC TAT CCA TTG GTC TTA GGC CCT AA-3'). The cleavage was observed when 12308A→G was present. The mtDNA haplogroups (H, I, J, K, M, T, U, V, W, X) and clusters of related haplogroups (HV, TJ, UK, WIX) were defined according to the published criteria [7, 9].

The polymorphism 9477G→A, determining haplogroup U5 [14], was detected by conformation-sensitive gel electrophoresis (CSGE). The DNA fragment spanning between nts 9211 and 9595 was first amplified and a portion of the PCR product was then taken for heteroduplex formation, where an unknown fragment was mixed with the corresponding fragment harboring the polymorphic variant 9477A. Mixtures of the amplified fragments were denatured at 95 °C for 5 min and the heteroduplexes subsequently allowed to anneal at 68 °C for 30 min. CSGE was carried out as described earlier [14].

The mtDNA sequence between nts 16024 and 16400 in HVS-I was determined by automated sequencing (ABI PRISM™ 377 Sequencer using the Dye Terminator Cycle Sequencing Ready Kit, Perkin Elmer, Foster City, Calif., USA) after treatment with exonuclease I and shrimp alkaline phosphatase [16]. Each unique mtDNA sequence was defined as a haplotype.

#### *Summary Statistics and Interpopulation Analysis*

Differences in mtDNA haplogroup frequencies between the populations were evaluated using the exact test of population differentiation [17] as implemented in ARLEQUIN 2.0 [18] or, if applicable, by the  $\chi^2$  test or Fischer's exact test. Sequence data on the mtDNA HVS-I were used to calculate molecular diversity indices within populations by means of the ARLEQUIN 2.0 software package [18]. Analysis of molecular variance (AMOVA) was used to detect variance among and within populations. The HVS-I sequence spanning the nts 16024–16400 was used for the Finnish populations, whereas comparisons between the Finns and Saami were based on the nts 16024–16383 sequence.

## **Results**

### *Frequencies of mtDNA Haplogroups in Four Regions in Finland*

Determination of the frequencies of mtDNA haplogroups in individuals from central and northern Finland showed haplogroups H and U to be the most common mtDNA genotypes, with frequencies of 40 and 28%, respectively (table 1). Haplogroup W was the third most common, while the remaining ones were considered rare, being present in less than 6%. In addition, macrohaplogroup M was present in 2.5% of the Finnish populations, based on the presence of the *Dde*I site at nt 10934 and *Alu*I site at nt 10397. The haplogroup of one sample could not be determined.

The frequencies of the haplogroups differed from those reported previously for the Finns ( $p = 0.04$ ; exact test of population differentiation) [7], as also did the frequencies of the five mtDNA haplogroup clusters HV, UK, TJ, WIX and M ( $\chi^2$  test,  $p = 0.006$ ). Analysis at the province level (table 2) showed that the samples from the two provinces in northern Finland did not differ from each other, nor did those from the two provinces in central Finland. However, significant differences in haplogroup frequencies were observed between all pairs of one northern and one central province. Haplogroups that were more frequent in the northern provinces included U, V, T and M, while haplogroups H, J and I were more frequent in the central provinces (table 1).

### *Sequence Variation in the HVS-I in Northern and Central Finland*

Sequencing of the HVS-I in the mtDNA of the 403 samples revealed 103 haplotypes characterized by 82 polymorphic sites and including 77 transitions, 8 transversions and 2 single base deletions (see Appendix). The standard indices of genetic heterogeneity (table 3) were quite similar to those previously reported for the Finns [19]. The distribution of pairwise differences in the samples from the four provinces was unimodal, with a mode value between 3 and 5 and with raggedness values between 0.0122 and 0.0157. AMOVA in the HVS-I segment suggested differences between the populations of the two northern provinces and those of the two central provinces (table 4).

Three HVS-I haplotypes were the most common among Finnish populations. These included haplotype H/ht1 (10%), which is identical to the Cambridge reference sequence (CRS) [22], haplotype W/ht1 (8.9%), and haplotype U/ht1 (7.4%), which contains the Saami motif (see Appendix). Haplotypes U/ht6 and U/ht7 also harbored

**Table 1.** Mitochondrial DNA haplogroup frequencies in the four populations

	Northern Ostrobothnia	Kainuu	Northern Savo	Central Ostrobothnia	Total	Finland <sup>1</sup>
<i>Haplogroup</i>						
H	34	37 (36)	53	39	163 (40)	20 (41)
V	6	9	4	3	22 (5.5)	2 (4.1)
U	34	36 (35)	17	25	112 (28)	8 (16)
K	3	2	4	3	12 (3.0)	2 (4.1)
T	8	1	1	0	10 (2.5)	3 (6.1)
J	2	1	6	9	18 (4.5)	7 (14)
W	8	10	6	13	37 (9.2)	2 (4.1)
I	0	2	7	5	14 (3.5)	1 (2.0)
X	1	0	0	3	4 (1.0)	2 (4.1)
M	3	6	1	0	10 (2.5)	1 (2.0)
Unknown	0	0	1	0	1 (0.2)	1 (2.0)
Total	99	104	100	100	403 (100)	49 (99.5)
<i>Cluster</i>						
HV	40	46 (44)	57	42	185 (46)	22 (45)
UK	37	38 (37)	21	28	124 (31)	10 (20)
TJ	10	2	7	9	28 (6.9)	10 (20)
WIX	9	12	13	21	55 (14)	5 (10)
M	3	6	1	0	10 (2.5)	1 (2.0)
Unknown	0	0	1	0	1 (0.2)	1 (2.0)
Total	99	104	100	100	403 (100)	49 (99)

The absolute and relative frequencies are identical with the exception of the relative frequencies shown in parentheses.

<sup>1</sup> Data from Torroni et al. [7].

**Table 2.** Exact test of population differentiation

	1	2	3	4	5
1		-	+	+	+
2	0.17±0.01		+	+	+
3	<0.0001	<0.0001		-	-
4	0.0005±0.0002	0.0007±0.0005	0.18±0.02		-
5	0.03±0.01	0.002±0.002	0.14±0.01	0.10±0.01	

Populations: 1 = Northern Ostrobothnia; 2 = Kainuu; 3 = Northern Savo; 4 = Central Ostrobothnia; 5 = Finland [7]. Below the diagonal: actual significance values; above the diagonal: + = significant difference (significance level = 0.05).

the Saami motif, making its frequency 8.7%, the highest observed outside the Saami population. However, the frequency of the Saami motif differed between northern (12.3%) and central (5.0%) Finland ( $p = 0.01$ , Fischer's exact test). In addition, the partial Saami motif 16189T→C and 16270C→T was found at a frequency of 7.4% (8.4% in the northern provinces and 6.5% in the

central provinces). Altogether, 16270C→T was found in 82% of the samples belonging to haplogroup U. This substitution has been used to define subcluster U5, but we found that an additional 11 samples belonged to this subcluster when the 9477G→A polymorphism was used as well (see Appendix) [14]. Thus subcluster U5 was found at a frequency of 26% in the Finnish population.

**Table 3.** mtDNA HVS-I sequence diversity in central and northern Finland

Population	Samples	Lineages	Samples with shared lineages	Samples with private lineages	Variable sites	Gene diversity	$\Theta_\pi$ (SD)	$\Theta_k$ (95% CI)	$\Theta_s$ (SD)	Private lineages %
Northern Ostrobothnia	99	48	48	24	59	0.973±0.006	4.80 (2.62)	36.1 (24.1–53.9)	11.42 (3.10)	44
Kainuu	104	39	59	13	43	0.955±0.008	4.66 (2.55)	22.2 (14.7–33.3)	8.05 (2.26)	31
Northern Savo	100	41	56	19	52	0.950±0.011	4.14 (2.30)	25.5 (16.8–38.2)	10.04 (2.76)	39
Central Ostrobothnia	100	43	46	24	47	0.964±0.008	4.61 (2.53)	28.1 (18.6–42.1)	9.08 (2.53)	42
Finns <sup>1</sup>	176	74	n.a.	n.a.	64	0.957	3.61 (2.04)	47.6 (34.7–64.9)	11.14 (2.78)	38
Saami <sup>1</sup>	115	25	n.a.	n.a.	29	0.815	3.79 (2.13)	9.6 (5.9–15.0)	5.45 (1.61)	40

<sup>1</sup> Data from Helgason et al. [19] that are based on sequence data from Sajantila et al. [10] for the Saami and from Pult et al. [5], Sajantila et al. [10], Richards et al. [20] and Kittles et al. [21] for the Finns. n.a. = not applicable.

**Table 4.** AMOVA results for the populations of the four provinces in central and northern Finland

Populations	Variance		$\Phi_{ST}$	p value
	among populations	within populations		
1-2-3-4	0.70	99.30	0.00699	0.006
1-2	-0.06	100.06	-0.00061	0.48
3-4	0.27	99.73	0.003	0.19
1-3	0.59	99.41	0.006	0.06
1-4	0.69	99.31	0.0069	0.05
2-3	1.51	98.49	0.015	0.003
2-4	1.18	98.82	0.012	0.014

Populations: 1 = Northern Ostrobothnia; 2 = Kainuu; 3 = Northern Savo; 4 = Central Ostrobothnia.

Another interesting feature of the Finnish mtDNA haplotypes was the findings that 9 out of the 10 samples that had been tentatively assigned to macrohaplogroup M exhibited a motif characterized by 16129G→A, 16185C→T, 16223C→T, 16224T→C, 16260C→T and 16298T→C. The same motif has been observed in mtDNAs from populations inhabiting northeastern Asia and has been taken to define haplogroup Z [23]. The presence of 16298T→C was intriguing, because this polymorphism is commonly used as evidence of haplogroup V. However, we found that 31% of the Finnish samples harboring 16298T→C in fact belonged to haplogroup Z.

**Table 5.** Frequencies of mtDNA haplotypes that are common to the Finnish and Saami populations

Haplotype	Central Finland %	Northern Finland %	Saami %
V/ht1	0.5	1.5	3.5
V/ht3	1.0	3.0	36
U/ht1	4.0	10.8	22
U/ht12	0	1.0	2.6
M/ht1	0.5	3.4	4.3
H/ht1	13	8.4	0.9
H/ht12	1.5	1.0	0.9
H/ht19	3.5	0.5	0.9
U/ht4	0	4.9	0.9
U/ht18	0.5	0	0.9

#### *Admixture between the Saami and Finnish Populations in Northern Finland*

HVS-I sequences from 403 Finns were compared to those from 115 Saami [10]. The Saami HVS-I sequences contained 26 haplotypes, of which 10 were common to both the Saami and Finnish populations (table 5). However, 5 of these were rare in the Saami, whereas haplotypes V/ht1, V/ht3, U/ht1, U/ht12 and M/ht1 were more prevalent (see Appendix). Interestingly, the frequencies of these haplotypes differed between the Saami and the Finns in a manner that suggested a geographical gradient (table 5). Their combined frequencies were 68% in the Saami, 20% in northern Finland and 6.0% in central Finland ( $p < 0.001$  for the difference,  $\chi^2$  test).

**Table 6.** AMOVA results for the populations of central and northern Finland and the Saami

Populations	Variance		$\phi_{ST}$	p value
	among populations	within populations		
1-2	0.93	99.07	0.00934	0.001
1-3	11.46	88.54	0.11464	<0.001
2-3	7.00	93.00	0.07000	<0.001

Populations: 1 = Central Finland; 2 = Northern Finland; 3 = Saami.

These five haplotypes harbored sequence signatures that are common among the Saami but less frequent or absent elsewhere in Europe [9, 10]. Haplotype U/ht1 was characterized by the Saami motif, while haplotype U/ht12 harbored a partial motif lacking 16144T→C. The 16298T→C polymorphism has been reported to occur at a high frequency in the Saami, and haplotypes V/ht1 and V/ht3 appeared to contribute to this. Interestingly, haplotype M/ht1, which is known to represent haplogroup Z, was found in 5 Saami mtDNAs, and a nearly identical HVS-I was found in 1 sample.

AMOVA suggested that whereas variance due to variation among samples was low in northern and central Finland, a remarkably high variance was observed between the Saami and Finnish samples (table 6). Interestingly, the variance due to variation among populations was slightly lower between the northern Finland material and the Saami than between central Finland and the Saami (table 6).

## Discussion

The frequencies of the mtDNA haplogroups were found to differ significantly from those reported earlier for the Finns [7, 10], the most conspicuous difference being the high frequency of haplogroup U. The finding may be due to a true difference in haplogroup frequencies between southern and northern Finland. The majority of the Finns live in the southern part of the country, suggesting that, in the case of random ascertainment, the samples in the two previous studies [7, 10] represent southern Finland. Indeed, the analysis of haplogroup frequencies between the populations of four neighbouring provinces in

this study and the results of AMOVA suggested differences in the genetic structure of the populations of northern and central Finland. The subjects had been enrolled on the basis of their province of residence, but it was also required that their mothers should have been born in the same province, to ensure that the samples represented the ancestral population of the respective provinces. We estimated that the mothers of the subjects had been born between the years 1906 and 1952. The majority of the population was rural and the average migration rate had been low during the period concerned [24]. In fact, 96% of the Finns in 1880 were living in the same province where they had been born, and the corresponding percentage in 1920 was 95% in the provinces of Northern Ostrobothnia and Kainuu and slightly lower in Central Ostrobothnia and Northern Savo [1].

The population of northern Finland differed markedly from other European populations in the frequency of haplogroup U. In fact, in this population, subcluster U5 comprised 92% of mtDNAs from this haplogroup (see Appendix). By contrast, subcluster U5 appears at much lower frequencies in Europe as a whole, varying in frequency from 0% in Western European countries to 53% in the Saami [12]. On average, the frequency of this subcluster is twice as high in Finland as in Europe as a whole [12], but it appears to be even higher in northern Finland. However, this comparison of frequencies is subject to an unknown error due to the fact that U5 was previously defined by the presence of 16270C→T [see 12]. Further examination of these HVS-I sequences indicates that a certain portion of subcluster U5 has experienced a back mutation at this site [14], resulting in approximately 10% of the Finnish haplotypes lacking the 16270C→T polymorphism.

The results of AMOVA suggested that variance due to variation among populations was slightly lower between the northern Finland population and the Saami than between that of central Finland and the Saami. Ten out of the 103 Finnish and 26 Saami haplotypes were shared, giving a similar estimate for the frequency of shared haplotypes as has been reported previously [11]. Five of the 10 haplotypes occurred at a frequency >1% in the Saami and, interestingly, each of them was more frequent in northern Finland than in central Finland. One of these harbored the Saami motif [10]. Haplotypes with this motif have been thought to be restricted to the Saami and have been observed at a frequency of 58% in the Norwegian Saami [25] and 34% in the Finnish and Swedish Saami [10], but the motif is rare elsewhere in northern Europe and absent in other parts of Europe [9, 10]. We

found the frequency of this motif to be 12.3% in the samples from northern Finland and 5% in the samples from central Finland (see Appendix), and it has been reported in 1 out of 32 samples (3%) representing the entire Finnish population [11]. The Saami motif has been found at a frequency of 1.0% among Russians [26] and 2.3% among Norwegians [27], but it was not detected in a small sample of Swedes [7], although 3197T→C, a polymorphism suggestive of subcluster U5 [14], has been found at a frequency of 11% [28].

The Saami are considered genetic outliers in Europe [3, 29], and therefore the high frequency of the Saami motif in northern Finland is interesting, as it suggests admixture between the Saami and Finnish settlers during the colonization of Finland. On the basis of the differences in haplotype frequencies between northern and central Finland and on the assumption that the acculturation of Saami women to an agricultural lifestyle was more probable than that of Finnish women to a nomadic lifestyle, it may be said that the flow of mtDNA has been from the Saami to the Finns. However, gene flow has probably been reciprocal, and an opposite flow may be suggested by the observation of three closely related European-specific haplotypes H/ht1, H/ht12 and H/ht19 in the Saami and by the presence of haplotype V/ht3 that is widely distributed in European populations [21].

In addition to the Saami motif, we found haplotypes V/ht1, V/ht3 and M/ht1 to occur at a higher rate in northern Finland than in central Finland. The 16298C→T polymorphism has been used to define haplogroup V in European populations [7, 9, 12], but we found that it was also present in samples belonging to macrohaplogroup M. In fact, this macrohaplogroup accounted for 9.4% of the Saami samples harboring 16298T→C and 31% of the Finnish samples harboring 16298C→T (see Appendix). Both haplogroup V and macrohaplogroup M showed a geographical variation in frequency similar to that observed for the Saami motif. Interestingly, 9 out of the 10 Finnish samples and 5 out of the 6 Saami samples belonging to macrohaplogroup M exhibited the HVS-I motif of haplogroup Z. Haplogroup Z mtDNAs have recently been identified in two Kamchatka populations, with a frequency of 6.5% in the Itel'men and 5.4% in the Koryaks [23]. It has also been found with a frequency of 26% in the Evens population of northeastern Asia [30] and with a frequency of 1.9% in Mongolians [31], but it is absent in many other east Asian populations [23]. Interestingly, it is present in 4.3% of the Saami [10, 11] and 3.9% of the population of northern Finland, and it has also been detected in one sample representing the Norwegians [25].

The European pattern of mtDNA haplotypes is evident in the Finns, whereas the Saami are quite distinct from all other European populations [10, 12]. Certain haplotypes considered to be Saami-specific had high frequencies in the Finns, suggesting a genetic admixture. This maternal genetic contribution appeared to have been more pronounced in northern Finland. Previous data have shown a high frequency of Asian Y chromosome haplotypes in the Finns and Saami [8], whereas the mtDNA haplotypes in the Finns have been considered distinctively European. The presence of haplogroup Z in the Finns and Saami evidently indicates that traces of Asian mtDNA genotypes have survived in the contemporary populations, but the discrepancy between the high frequency of Asian-specific haplotypes of the Y chromosome and the rarity of Asian-specific mtDNA haplotypes may suggest that the Y chromosome and mtDNA are subject to differential drifting that has led to an almost complete loss of Asian-specific mtDNA haplotypes.

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