mtDNA sequences in the Norwegian Saami and main populations

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INTRODUCTION

The mitochondrial DNA has several properties useful for the reconstruction of human population history. The origin of the Saami people is still not known, even if much have been done to unravel the secrets of the past. One hypothesis is that Saami are the ancient Nordic people, another that they have a strong Mongolian component among their ancestors, and a third that they are mainly of European descent (for a comprehensive review, see Eriksson 1988). In a recent report, Sajantila et al. (in press 1995) show that Saami are distinct from other Europeans by characteristic patterns in mtDNA D-loop sequence, and they conclude that the Saami seem to have a long history distinct from other European populations.

A considerable proportion of Saami live in Norway, with Karasjok and Kautokeino as two main, geographically separated communities (Fig. 1). We have a rather large, 25 years old material of serum samples from Karasjok and Kautokeino.

The aim of the present study was to find out if these samples could be used as a source for mtDNA sequencing. Having shown that this is indeed the case, we have embarked on a study of D-loop variation in Norwegian Saami and main populations. This is a preliminary report of this study.

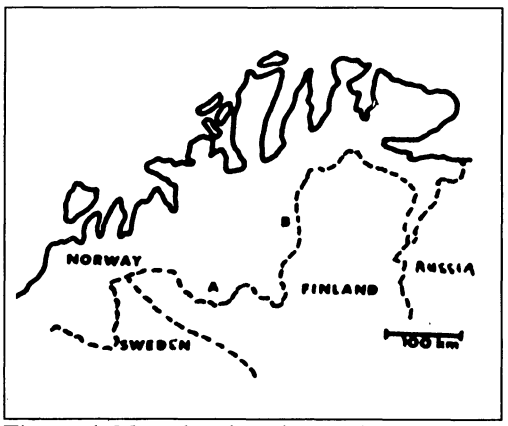

Figure 1 Map showing the northern parts of Norway. The letters indicate the administrative centers of Kautokeino (A) and Karasjok (B).

MATERIALS

The Saami material consists of serum from 201 Saami from the Karasjok and Kautokeino communities. 159 samples (92 from Karasjok, 67 from Kautokeino) were collected in 1970. The criteria as to the Saami origin and unrelatedness are described by the collector (Teisberg 1971). In 1989 another 42 samples from Karasjok were collected. 20 of these are from individuals which may be related to/identical with persons in the old material.

The material from the main Norwegian population consists of blood samples from 30 unrelated individuals from all over Norway. They all have four Norwegian grandparents.

METHODS

DNA extraction: Serum samples were spun in an Eppendorf centrifuge 12000 rpm, 30 min. The pellets were digested overnight by proteinase K, and DNA extracted by phenol/chloroform, pelleted by 100% ethanol and resuspended in 50 microliter TE buffer. Blood samples (EDTA) were extracted by the salting out method (Miller et al. 1988).

Primers and PCR: Amplification conditions for the 9 bp deletion: Hertzberg et al. 1989. **Electrophoretic procedures:** 6% PAGE, ABI 373A Sequencer, Software: 673 Genescan, 373 Sequencer, and Sequence Navigator. **Sequencing reactions:** PRISM™ Solid Phase Sequenase Dye Terminator DNA Sequencing Kit (Perkin Elmer).

RESULTS AND DISCUSSION

Table 1: Distribution of mtDNA sequences in 30 main population Norwegians.

HAPLOTYPES	16113	16129	16145	16162	16171	16185	16189	16192	16213	16215	16221	16223	16224	16256	16260	16263	16270	16271	16275	16278	16291	16292	16293	16298	16304	16311	16362	16391
Anderson-81	G	G	G	A	A	C	T	C	G	A	C	C	T	C	C	T	C	T	G	C	C	C	A	T	T	T	T	G
Norwegians:																												
No. of obs																												
6																												
2																									C			
1									A																C			
2													C													C		
1										G			C													C		
1							C						C										G			C		
1		A			G							T											C			C	A	
2																							G			C		
1							C										T											
1							C										T											
1												T								T							C	
1		A				T						T	C		T									C				
1												T										T						
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1								T						T			T				T							
1				G																								
1			A																									
1																C												
1							C											C										
1																								C				
1											T								A									
TOTAL:30																												

Table 2: Distribution of mtDNA sequences in Norwegian Saami populations.

	HAPLOTYPES		16013	16126	16136	16144	16148	16149	16154	16162	16189	16223*	16256	16261	16270	16294	16298	16335	16358	16360	16362*	16400
		Anderson-81	A	T	T	G	A	C	G	A	T	C	C	C	C	C	T	A	A	C	T	C
SAAMI	SAAMI	Obs. in																				
KARASJOK	KAUTOKEINO	NORWEGIANS																				
No. of obs	No. of obs	No. of obs																				
50	13	1															C					
41	29	0				C					C				T							
7	2	0				C	T				C				T							
19	17	0				C	T				C				T			G				
1	0	0				C	T				C	T										
5	0	0		C	C						C	T								T	C	
5	0	0								G												
1	0	0								G	C											
3	0	0										T										
0	1	0		C					A							T						
0	2	0											T		T							
1	0	6																				
TOTAL:133	TOTAL:64	TOTAL:7																				

PCR products were obtained from all samples. All 201 Saami lacked the 9 bp deletion commonly seen in Asians (Hertzberg et al. 1989).

mtDNA sequences (15997-16401, primers included) for 197 individuals (four are still not typed) are depicted in Tables 1 and 2 for the main population and the Saami, respectively. Ordinarily, about 350 bp could be read. In the 30 individuals from the main population, the Cambridge sequence (Anderson 1981) was experienced in 20 % (6 individuals), otherwise no particular sequence was seen more than twice (7%), a total of 22 different sequences being observed. In the 197 Saami, only twelve different sequences were observed. 57% of the Saami shared a main theme with C in positions 16144 and 16189, and T in 16270 (Motif A, not seen in the main population), while another 31% shared a sequence differing from the Cambridge sequence only by a C in position 16298 (Motif B, seen once in the main population). These findings are in good accordance with those of Sajantila et al. (in press 1995). In the present material, motif A is divided in two groups by a A->T transversion in 16148, and a further subdivision by a A->G transition in 16335 was also observed. While Motif A thus includes several variants, Motiv B does not. These findings might indicate that Motif B represents an admixture of a population with a mtDNA sequence closely related to the most common European sequence into a population already exhibiting multiple variants of Motif A.

There are significant differences in sequence distribution between the Kautokeino and Karasjok Saami subpopulations (p=0.005). In particular, Kautokeino Saami have a higher proportion of Motif A and a lower proportion of Motif B than Saami from Karasjok. Moreover, the number of different mtDNA lineages may seem to be even less in Kautokeino than in Karasjok.

Comparing the Saami mtDNA sequences with those of other populations (see e.g. Stoneking et al. 1991), no obvious close relationships are found. The C in 16298 as well as the C in 16189 are each found in one of four sequences characteristic for Amerind populations, but with several site differences elsewhere (Ginther et al. 1993).

The remarkable differences in sequence distributions between Saami and neighbouring populations have already proved that mtDNA is a particularly efficient tool for population studies in arctic north-western Europe. We have started studies of ancient bones which hopefully will add valuable contributions to the knowledge of the history of people living in these areas.

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