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Mitochondrial DNA variability in Poles and Russians

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SUMMARY

Mitochondrial DNA (mtDNA) sequence variation was examined in Poles (from the Pomerania-Kujawy region; $n = 436$) and Russians (from three different regions of the European part of Russia; $n = 201$), for which the two hypervariable segments (HVS I and HVS II) and haplogroup-specific coding region sites were analyzed. The use of mtDNA coding region RFLP analysis made it possible to distinguish parallel mutations that occurred at particular sites in the HVS I and II regions during mtDNA evolution. In total, parallel mutations were identified at 73 nucleotide sites in HVS I (17.8%) and 31 sites in HVS II (7.73%). The classification of mitochondrial haplotypes revealed the presence of all major European haplogroups, which were characterized by similar patterns of distribution in Poles and Russians. An analysis of the distribution of the control region haplotypes did not reveal any specific combinations of unique mtDNA haplotypes and their subclusters that clearly distinguish both Poles and Russians from the neighbouring European populations. The only exception is a novel subcluster U4a within subhaplogroup U4, defined by a diagnostic mutation at nucleotide position 310 in HVS II. This subcluster was found in common predominantly between Poles and Russians (at a frequency of 2.3% and 2.0%, respectively) and may therefore have a central-eastern European origin.

INTRODUCTION

Analysis of mitochondrial DNA (mtDNA) polymorphism has become a useful tool for human population and molecular evolution studies, allowing researchers to infer the pattern of female migrations and peopling of different regions of the world (Wallace, 1995). The use of the phylogeographic approach has allowed refinement of the analysis of maternal mtDNA lineages, suggesting the current model of complex demographic scenarios for European peopling (Richards *et al.* 2000). Although linguistic, anthropological and archaeological data, as well as classical genetic data, cover most of the Slavonic populations living in Europe, there are many unanswered questions about the origin and dispersal of Slavs.

Archaeological studies indicate that the Lusatian culture (1300 to 1100 B.C.) emerged in central Europe, and later spread over a region that reached from the central basin of the Oder River and the Bohemian mountain ridge, as far east as the Ukraine, and as far north as the shores of the Baltic Sea (Sedov, 1979; Šavli *et al.* 1996). Despite the divergent views on the ethnic affiliation of the Lusatian culture, it is often considered that this culture constituted the foundation of the historical development of the Proto-Slavs (Šavli *et al.* 1996). According to linguistic data, the split among Proto-Slavs, the bearers of the Lusatian culture, resulted in the three Slavonic language groups – Western, East-

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ern and Southern (Šavli *et al.* 1996). In the north, the Lusatian culture was succeeded by the Pomeranian culture, extending over the coastal region from the mouth of the Oder to the mouth of the Vistula. The Przeworsk group encompassed the southern parts of present-day Poland. In the 2nd and 3rd centuries A.D., this group spread northward into the swampy Pripet and united there with the Zarubincy culture. It has been suggested that out of this culture the Eastern Slavonic language group developed (Rybakov, 1981; Šavli *et al.* 1996). Archaeologists report that the Slavs invaded the Balkan peninsula as early as the 2nd century A.D., and since this settlement movement of the Southern Slavs gradually evolved (Sedov, 1979). All of these 'migration' hypotheses claim that modern Slavonic groups are the result of an admixture between pre-Slavonic European populations and Slavonic tribes, whose homeland was probably in central Europe (Sedov, 1979; Alekseeva & Alekseev, 1989). This theory also predicts that diverse modern Slavonic populations may have certain combinations of genetic markers derived from the gene pool of the assumed ancestral Proto-Slavonic population.

A high-resolution analysis of maternal mtDNA lineages appears to be a highly informative approach in the reconstruction of the past demographic events, when large enough samples are available (Helgason *et al.* 2000). MtDNA sequences can be used to create a detailed pattern of the spatially resolved distribution of maternal lineages in Slavonic populations, and to trace a number of shared maternal lineages unique for Slavonic groups, connecting them among themselves and to other neighbours such as present-day German and Finno-Ugric populations. However, the mtDNA data sets for Slavonic populations living in Southern, Central, and Eastern Europe are either incomplete or virtually non-existent for many regional groups of Slavs, especially for populations inhabiting the East European Plain. Population samples of Slavs have been analyzed in different ways: some covering only HVS I sequences, others also including coding-region RFLPs (Malyarchuk *et al.* 1995; Calafell *et al.* 1996; Orekhov *et al.* 1999; Richards *et al.* 2000; Tolk *et al.* 2000; Malyarchuk & Derenko, 2001). In addition, almost all of these mtDNA studies have not addressed specific questions about the origin and early dispersal of Slavs in Europe. To date, it is known that Slavonic populations sharing the same language group (such as Russians, Ukrainians, Bulgarians) display a large amount of interpopulation genetic variation (Malyarchuk & Derenko, 2001). Moreover, we have not found any specific combinations of unique mtDNA types that clearly distinguish Russians from Germans and the neighboring Eastern European populations.

To obtain a better characterization of Slavonic mtDNA variability, we present here mtDNA diversity data in Poles and Russians, based on the HVS I and HVS II sequences typed for the presence of major West Eurasian haplogroup-specific markers.

Materials and methods

Population samples

A population sample of 436 Poles from the Pomerania-Kujawy region of the northern part of Poland was studied. In addition, three population samples of Russians from the European region of Russia were analysed: 62 unrelated individuals from the south (Stavropol region), 76 from the centre (Orel region) and 63 from the east (Saratov region).

MtDNA analysis

DNA samples from the blood of individuals studied were used for mtDNA amplification and sequencing. PCR amplification of the entire noncoding region was performed using the primers L15926 and H00580. The temperature profile for 30 cycles of amplification was 94 °C for 20 sec, 50 °C for 30 sec, and 72 °C for 2.5 min (Thermal Cycler 9700; Perkin Elmer, USA). The resulting amplification product was diluted 1000-fold and 4 μl aliquots were added to an array of second-round, nested PCR reactions (32 cycles) to generate DNA templates for sequencing. The primer sets L15997/M13(−21)H16401 and M13(−21)L15997/H16401 were used to generate

both strands of the hypervariable segment I (HVS I). Similarly, the primer sets L00029/M13-(−21)H00408 and H00408/M13(−21)L00029 were used for hypervariable segment II (HVS II). Both primer sequences, and nomenclature, were used according to Sullivan *et al.* (1992). Negative controls were prepared for both the DNA extraction and the amplification process. PCR products were purified by ultrafiltration (Microcon 100; Amicon) and sequenced directly from both strands with the (−21)M13 primer using the BigDye Primer Cycle Sequencing Kit (Perkin Elmer) according to the manufacturer's protocol. Sequencing products were separated in a 4% PAGE gel on the ABI Prism™ 377 DNA Sequencer. Data were analyzed using DNA Sequencing Analysis and Sequence Navigator programs (Perkin Elmer). The nucleotide sequences obtained were compared with the Cambridge reference sequence (CRS; Anderson *et al.* 1981).

To determine the haplogroup status of the control region (CR) sequences, RFLP typing was performed by restriction endonuclease analysis of PCR amplified mtDNA fragments using the same primer pairs and amplification conditions as described by Torroni *et al.* (1996, 1997), Macaulay *et al.* (1999), and Finnilä *et al.* (2000). The samples were typed for a restricted set of RFLPs that were diagnostic of all major western Eurasian clusters, on the basis of the hierarchical mtDNA RFLP scheme (Macaulay *et al.* 1999).

To determine haplogroup H sequences, all samples were tested for 14766*Mse*I, 10394*Dde*I, and 7025*Alu*I. Samples lacking these three sites were assigned to cluster H. All non-H samples harboring −14766*Mse*I and −10394*Dde*I were tested for 15904*Mse*I, and samples with +15904*Mse*I site were classified as cluster pre-V, which is solely defined by the two CR mutations 16298C and 72C (Torroni *et al.* 2001). All non-H and non-pre-V samples (−14766*Mse*I and −10394*Dde*I) were determined as HV*.

All non-HV samples were tested for 12308*Hin*fI. Those with +12308*Hin*fI were assigned to clusters U and K, and were further determined as belonging to haplogroup K or to subgroups of the haplogroup U on the basis of the HVS I motif information (Richards *et al.* 1998; Macaulay *et al.* 1999). The phylogenetic status of subhaplogroup U4 was determined by RFLP screening of the 4643*Rsa*I site (Macaulay *et al.* 1999).

The remaining samples were tested for 13366*Bam*HI, 15606*Alu*I, 15925*Msp*I, and 12629*Ava*II. Those with +13366*Bam*HI, +15606*Alu*I, and −15925*MspI* were assigned to cluster T. The haplogroup T sequences lacking the 12629*Ava*II site were classified as T1, whereas those with +12629*Ava*II were declared as T*. The remaining samples were tested for 13704*Bst*OI, and those with −13704*Bst*OI and +10394*Dde*I were classified as J.

Further, mtDNAs were classified as follows: +14465*Acc*I to cluster X; −4529*Hae*II, +8249-*Ava*II, +16389*Bam*HI, and +10032*Alu*I to cluster I; +8249 *Ava*II and −8994*Hae*III to cluster W; +10394*Dde*I and +10397*Alu*I to cluster M. M-sequences were further classified as belonging to haplogroup C (−13259*Hin*cII, +13262*Alu*I), D (−5176*Alu*I), E (−7598*Hha*I), or G (+4830*Hae*II, +4831*Hha*I). The remaining control region sequences were assigned to certain haplogroups (such as R*, N1b, N1c, L3, pre-HV) on the basis of the HVS I motifs classification (Macaulay *et al.* 1999; Richards *et al.* 2000). Sequence classification into subhaplogroups was based on the HVS I motifs and nomenclature of Richards *et al.* (1998, 2000) and Macaulay *et al.* (1999).

Phylogenetic analysis

For phylogenetic analysis, all available published data on HVS I-RFLP mtDNA variability in West Eurasian populations were used (Richards *et al.* 2000). To classify the Slavonic mtDNA haplotype diversity, a phylogeographic approach, based on the phylogenetic analysis of the spatial distribution of mitochondrial haplotypes and haplogroups determined as a monophyletic clade, was performed (Richards *et al.* 1998). The phylogenetic relationships between mitochondrial haplotypes comprising various combinations of the HVS I and HVS II sequences and RFLPs were analyzed by the median-network method (Bandelt *et al.* 1995). To es-

timate the diversity of mtDNA haplotypes, the average number of transitions on the reconstructed phylogeny from ancestral type to each sample (ρ) was used, according to the methods of Forster *et al.* (1996).

For the CR sequence sharing analysis, HVS I and HVS II haplotypes of Poles and Russians, as well as other European populations, were compared. Data from the following populations were used: 200 Southern Germans (Lutz *et al.* 1998); 101 Austrians (Parson *et al.* 1998); 150 Western Germans (Baasner *et al.* 1998; Baasner & Madea, 2000); 109 North-Western Germans (Pfeiffer *et al.* 1999); and 192 Finns (Finnilä *et al.* 2001*b*).

results and discussion

Sequence variability in Poles and Russians

In the present study, the nucleotide sequences of HVS I from position 15991 to 16400 and HVS II from position 20 to 420 have been determined in 436 Poles and 201 Russians. Comparison to the Cambridge reference sequence (Anderson *et al.* 1981) showed that 140 nucleotide sites were polymorphic in HVS I (34.2%) and 79 sites in HVS II (19.7%). Transitions and transversions were found at 136 nps in HVS I and at 73 nps in HVS II. For each hypervariable region, transitions predominate over transversions, being found with a ratio of 133:16 and 73:0 in HVS I and HVS II, respectively. Among the transitions, pyrimidine substitutions were observed with significantly higher frequency in HVS I (with a pyrimidine:purine ratio of 92:41), whereas in HVS II the pyrimidine:purine ratio was 42:31. Among the transversions in HVS I there is no predominating type: C→A transversions were found at 5 nps, A→C at 4 nps, C→G at 3 nps, A→T at 3 nps and G→C at one nucleotide position. It is interesting that multiple substitutions were found at 9 positions of HVS I – from C to T and A at np 16111; from C to T and A at np 16114; from G to A and C at np 16129; from C to T and G at np 16176; from C to T, G and A at np 16188; from C to T and G at np 16239; from A to G and T at np 16241; from A to G and C at np 16258; and from A to G, T and C at np 16318.

Point deletion and insertion events were observed both in HVS I and HVS II. In HVS I an insertion polymorphism was found at np 16193. The occurrence of such a type of polymorphism is probably due to instability in the homopolymeric tract between nps 16184 and 16193, which can be associated with a transition from T to C at np 16189 (Bendall & Sykes, 1995). Similarly, length polymorphism in the poly-C tract of HVS II at nps 303–315 was found in the majority of the mtDNA samples studied. In this tract, insertions of either one, two (at nps 309 and 315), or three (at np 309) C-residues were identified. In addition, insertions of single nucleotides were observed at nps 42 (+T), 60 (+T), 270 (+A) and 299 (+C). Deletions of nucleotides in the mtDNA control region appear to be rarer events, and they were found at nps 16073 (−C) and 16078 (−A) in HVS I, and at nps 249 (−A) and 315 (−C) in HVS II.

Heteroplasmic positions were clearly detected in four instances at nps 16093, 16231, 16325 and 72. The heteroplasmic status of these positions was confirmed several times by sequencing of both mtDNA strands.

The use of RFLP analysis for mtDNA coding regions amplified via PCR has allowed us to determine the exact phylogenetic status of HVS I and II sequences and distinguish independent (parallel) mutations occurring at particular sites during the evolution mtDNA lineages (Macaulay *et al.* 1999; Richards *et al.* 2000; Finnilä *et al.* 2001*b*; Malyarchuk & Derenko, 2001). As a result, we have identified a total of 73 hypervariable sites in HVS I (17.8%) and 31 hypervariable sites in HVS II (7.73%) at which more than one independent mutation is observed (Tables 1 and 2). However, in HVS II the number of parallel mutations is approximately 1.8 times as high as the corresponding HVS I value (279 and 155, respectively). Accordingly, the ratio of the average number of parallel mutations per site in HVS II (5.0) and HVS I (3.82) is 1.31. This estimate ranges from 1.17 in Russians to 1.34 in Poles. Therefore, as was suggested previously (Bandelt *et al.* 2000), although on average HVS II seems to be less

Table 1. *Parallel mutations detected in the mtDNA HVS I in Poles and Russians*

Position	Nucleotide change	Poles	*n*	Russians	*n*	Total
16051	A → G	H, U2, U5	3	H, U2	2	3
16069	C → T	J	1	K, J	2	2
16071	C → T	W	1	R*	1	2
16086	T → C	U*, I, X	3		0	3
16092	T → C	H, J1b, D	3	H, K, J*	3	5
16093	T → C	H, K, U4, U5, J1b, C, G	7	H, K, U2, U4, U5, J*, X	7	10
16126	T → C	(J*, T*)	1	U7, (J*, T*), pre-HV, pre-V, X, D	6	6
16129	G → A	H, U5, T*, T1, I, W, M*	7	H, U4, U1, I	4	9
16140	T → C	H, U5, T*	3		0	3
16145	G → A	J1b, J1a, N1b	3	U5, J*, J1b	3	5
16146	A → G	U8	1	T*	1	2
16148	C → T	H, M*	2	H, U7	2	3
16150	C → T	pre-V	1	U4	1	2
16153	G → A	H, pre-V	2	pre-V	1	2
16168	C → T	H, U3	2		0	2
16169	C → T	H, pre-V	2		0	2
16170	A → G	H, T1	2		0	2
16172	T → C	H, K, U5, J1b, T*, I	6	J1b, I	2	6
16179	C → T	H, U4, U5, U8, R*	5		0	5
16186	C → T	T1	1	J*, T1	2	2
16189	T → C	H, K, U4, U2, U5, J*, J1a, T*, T1, pre-V, X, C, D	13	H, U1, U2, U5, J*, T1, I, W, X, D	10	16
16192	C → T	H, K, U5, T*, W, M*	6	H, U5, J1b, W	4	7
16193	C → T	H, J2	2	H	1	2
16213	G → A		0	H, J*	2	2
16218	C → T	H, pre-V	2		0	2
16221	C → T	H, U4	2		0	2
16222	C → T	H, U5, J*, J1b	4	U5, J1b, T*	3	5
16223	C → T	U4, (I, N1b,N1c, W, X, L3, C, D, G, M*, E)	2	(I, W, X, D, G, M*)	1	2
16224	T → C	K	1	K, U1	2	2
16227	A → G	T*, R*, G	3	G	1	3
16231	T → C	H, J1a	2	H	1	2
16234	C → T	K, U4, U5, T*, C, G	6	HV*, G, M*	3	8
16239	C → T	H, U*	2		0	2
16241	A → G	J1a	1	X	1	2
16243	T → C	H	1	T1	1	2
16249	T → C	H, J*	2	H, U1	2	3
16256	C → T	H, K, U2, U5	4	H, U2, U5	3	4
16260	C → T	T	1	U5	1	2
16261	C → T	H, J1a, J1b	3	H, J*, J1b	3	4
16265	A → G	H, N1c	2	H	1	2
16266	C → T	H, U5, X, R*, D	5		0	5
16270	C → T	H, U5, pre-V	3	H, U5	2	3
16271	T → C	H, T*	2	T*	1	2
16274	G → A	H, K, X, R*	4	H	1	4
16278	C → T	H, J2, X, R*, G, E	6	H, X, G	3	6
16286	C → T	H, U5	2		0	2
16288	T → C	U5, C	2	H, U1	2	4
16290	C → T	H	1	H, J*	2	2
16291	C → T	H, K, U5, pre-V, M*	5	H, U5	2	5
16292	C → T	T*, W	2	U5, T*, W	3	3
16293	A → G	H, K, I	3	H, T1	2	4
16294	C → T	H, U4, U5, T	4	T, I	2	5
16295	C → T	W	1	HV*, W	2	2
16298	T → C	T*, pre-V, C, M*	4	pre-V	1	4
16300	A → G	X	1	H, X, M*	3	3
16304	T → C	H, T*	2	H, U5, T*, I	4	4
16309	A → G	U7, G	2	U7	1	2

Table 1. (*Cont.*)

Position	Nucleotide change	Poles	*n*	Russians	*n*	Total
16311	T → C	H, K, U*, U5, J*, T*, R*, HV*, pre-V, I, W	11	H, K, J*, J1a, HV*, I	6	12
16316	A → G		0	H, M*	2	2
16318	A → G	T*	1	H	1	2
16318	A → C	H	1	U7	1	2
16319	G → A	H, J*, J2, T1	4	H	1	4
16320	C → T	U*, U5, T*, W	4		0	4
16324	T → C	J*, T*	2	T*	1	2
16325	T → C	U5, W	2	H, U3, W	3	4
16342	T → C	H, U8	2		0	2
16343	A → G	H, U3	2	U3	1	2
16355	C → T	T*, HV*	2	R*	1	3
16356	T → C	H, U4	2	H, U4	2	2
16360	C → T	H	1	D	1	2
16362	T → C	H, U2, U5, U7, W, R*, D, G, E	9	H, U4, U2, U5, J*, T*, pre-HV, D, G, M*	10	14
16390	G → A	J*, N1b, X	3	U5	1	4
16391	G → A	I	1	H, I	2	2
16399	A → G	H, U5, T*	3	H, U5	2	3

Mutations are shown indicating positions relative to the revised CRS (Andrews *et al.* 1999). Haplogroup name denotes the presence of mutation occurring in the background of this haplogroup. A numeral (*n*) denotes the number of parallel mutations observed. Haplogroups, which have shared ancestry for a certain nucleotide variant, are shown in parentheses.

variable (per position) than HVS I, the homoplasic events are more numerous in HVS II, but concentrated at fewer sites – such as 146, 150, 152 and 195. These sites are at least as variable as the most variable positions (16093, 16189, 16311 and 16362) in HVS I.

The HVS I and HVS II regions differ slightly in the number of pyrimidine transitions at hypervariable sites, with a higher pyrimidine: purine ratio being found in HVS I (3.33 in total sample, 3.65 in Poles and 4.8 in Russians) in comparison with values for HVS II (2.52 in total sample, 2.68 in Poles and 2.63 in Russians).

The molecular instability of the polypyrimidine tract (C5)-T-(C4) located between nps 16184 and 16193 of the L-strand is one of the most studied manifestations of mtDNA hypervariability. It was found that a transition from T to C at np 16189 results in a continuous poly-C tract which may vary in length from 8 to 14 nucleotides (Bendall & Sykes, 1995; Marchington *et al.* 1996). Table 3 shows examples of variation in the tract length found in Polish and Russian mtDNAs. Another example of a hypervariable polypyrimidine sequence is a (C7)-T-(C5) tract starting at np 303 in HVS II. In comparison with the CRS (Anderson *et al.* 1981), insertion of an additional C residue at np 315 is common in Poles and Russians as well as in other population groups studied (Budowle *et al.* 1999). It is well established that both poly-C portions in this tract are very unstable; the length of the (C7)-sequences vary from 7 to 10 nucleotides and (C5)-sequences vary from 5 to 7 nucleotides (Torroni *et al.* 1994; Howell & Smejkal, 2001). The longest polypyrimidine tract, which was identified in Poles and Russians, was (C10)-T-(C6). In the present study, however, we have observed a poly-C tract with a total length of 13 C-residues, generated by a transition from T to C at np 310. This (C13)-sequence was observed in different mitochondrial haplogroups – H, U4, T, C and M* (see Appendix).

Haplogroup diversity in Poles and Russians and notes to mtDNA classification

The analysis of HVS I and II variability, in combination with RFLP typing of the coding region haplogroup-diagnostic sites, in a total sample of 637 Polish and Russian individuals,

Table 2. *Parallel mutations detected in the mtDNA HVS II in Poles and Russians*

Position	Nucleotide change	Poles	*n*	Russians	*n*	Total
64	C → T	H	1	pre-HV	1	2
73	G → A	H, HV*, U5, I, N1c	5	H, HV*, pre-HV	3	6
93	A → G	H, HV*, pre-V	3	H, pre-V	2	3
143	G → A	H, U4, W	3		0	3
146	T → C	H, K, U4, U5, J*, J1b, T*, W, X	9	H, K, U7, T*, HV*, R*	6	12
150	C → T	H, K, U*, U3, U5, J1a, J2, T*, HV*, W, D	11	H, U1, U3, U5, J1a, R*	6	13
151	C → T	H, K, pre-V, L3	4	H, U7, pre-HV	3	6
152	T → C	H, K, U2, U3, U4, U5, U7, J*, J1a, J2, T*, T1, I, N1b, W, X, L3, C, G	19	H, K, U4, U2, U3, U5, U7, T*, T1, pre-HV, R*	11	21
153	A → G	X	1	X, M*	2	2
182	C → T	H, X	2		0	2
189	A → G	K, pre-V, N1c, W	4	H, W	2	5
194	C → T	pre-V, W, R*	3	W	1	3
195	T → C	H, K, U4, U3, J1a, T*, T1, pre-V, N1c, W, X, R*, L3	13	H, U4, U1, U5, U7, J1a, T1, pre-V, W, X	10	16
198	C → T	H, X	2	U4, U5, pre-HV	3	5
199	T → C	H, T, I, W	4	T, I, X	3	5
200	A → G	U5, HV*	2	H	1	3
204	T → C	H, U4, U5, (I, N1c, W)	4	H, K, U5, J1a, (I, W), X	6	7
207	G → A	H, T*, (I, N1c, W)	3	H, (I, W)	2	3
210	A → G	J*, N1c	2		0	2
215	A → G	H, K, U4, J1a	4	U4, W	2	5
228	G → A	H, U2, U4, J*, pre-V	5	J*, pre-V	2	5
236	T → C	H	1	U4	1	2
239	T → C	H, I	2	H	1	2
240	A → G	T	1	H	1	2
250	T → C	K, I	2	I	1	2
263	G → A		0	H, T*	2	2
279	T → C	H, T*	2		0	2
295	C → T	H, J, R*	3	J	1	3
310	T → C	U4, T*, C, M*	4	H, U4	2	5
319	T → C	H, J1a, T*	3		0	3
385	A → G		0	U1, T1	2	2

Mutations are shown indicating positions relative to the HVS II sequence that differs from the revised CRS at np 73. For further information, see footnote to Table 1.

Table 3. *Instability of the polypyrimidine tracts in HVS I and II regions in Poles and Russians*

Nucleotide sequence	Nucleotide changes	Length of polypyrimidine tracts
HVS I, nps 16180-16193		
AAAACCCCCTCCCC	CRS	C5-T-C4
AAAACCCCCCCCCC	16189 T → C	C10
AAAACCCCCCCCCCC	16193 + C	C11
AAAACCCCCCCCCCCC	16193 + 2C	C12
AAACCCCCCCCCCC	16183 A → C	C11
AAACCCCCCCCCCCC	16183 A → C, 16193 + C	C12
AACCCCCCCCCCCC	16182 A → C, 16183 A → C	C12
AACCCCCCCCCCCCC	16182 A → C, 16183A → C, 16193 + C	C13
HVS II, nps 300-315		
AAACCCCCCCTCCCCC	CRS	C7-T-C5
AAACCCCCCCTCCCCCC	315 + C	C7-T-C6
AAACCCCCCCCTCCCCCC	309 + C, 315 + C	C8-T-C6
AAACCCCCCCCCTCCCCCC	309 + 2C, 315 + C	C9-T-C6
AAACCCCCCCCCCTCCCCCC	309 + 3C, 315 + C	C10-T-C6
AAACCCCCCCTCCCCCCC	315 + 2C	C7-T-C7
AAACCCCCCCCCCCCCC	310 T → C	C13
AAACCCCCCCCCCCC	310 T → C, 315 − C	C12

Table 4. *Haplogroup distributions* (*no. of individuals and % values in parentheses*) *in Poles and Russians*

Haplogroup	Poles (436)	Russians (201)
H	197 (45.18)	85 (42.29)
HV*	4 (0.92)	4 (1.99)
pre-V	21 (4.82)	11 (5.47)
pre-HV	0	1 (0.50)
J	34 (7.80)	16 (7.96)
T*	41 (9.40)	18 (8.96)
T1	9 (2.06)	4 (1.99)
K	15 (3.44)	6 (2.99)
U1	0	2 (1.00)
U2	4 (0.92)	3 (1.49)
U3	2 (0.46)	2 (1.00)
U4	22 (5.05)	7 (3.48)
U5	38 (8.72)	21 (10.45)
U7	1 (0.23)	1 (0.50)
U8	2 (0.46)	0
U*	1 (0.23)	0
I	8 (1.83)	5 (2.49)
W	16 (3.67)	4 (1.99)
X	8 (1.83)	7 (3.48)
N1b	1 (0.23)	0
N1c	1 (0.23)	0
R*	2 (0.46)	1 (0.50)
L3	1 (0.23)	0
M	8 (1.83)	3 (1.49)

allowed detection of 455 different mitochondrial haplotypes (see Appendix): 329 haplotypes among 436 Poles and 158 haplotypes among 201 Russians. This high resolution ensured that only 32 shared HVS I and II haplotypes were found between Poles and Russians.

In order to determine the phylogenetic status of HVS I and II sequences, restriction analysis of the coding regions was performed. As a result, it was found that mitochondrial haplotypes in Poles and Russians are clustered, according to the mtDNA classification (Macaulay *et al.* 1999; Richards *et al.* 2000; Torroni *et al.* 2001), into haplogroups H, pre-V, HV*, pre-HV, U, K, J, T, I, X, N, R*, M, L3 and their subgroups (Table 4). The Polish and Russian samples studied are characterized by a similar pattern of mtDNA haplogroup distributions. Comparison between them did not reveal statistical differences ($\chi^2 = 23.33$, 23 d.f., $p = 0.44$).

The main mitochondrial haplogroup of the Polish and Russian sequences is group H, which is the most frequent haplogroup in Europe and also common in the Near East (Richards *et al.* 1998, 2000). Haplogroup H comprises the majority of the Russian (42.3%) and Polish (45.2%) samples. This haplogroup has been difficult to subdivide based on HVS I and II variation alone. A number of major clades within haplogroup H were revealed on the basis of high-resolution mtDNA analysis and complete or partial mtDNA sequencing (Macaulay *et al.* 1999; Torroni *et al.* 1999; Finnilä *et al.* 2001*b*). Based on mtDNA HVS I and II sequencing data in Poles and Russians, only three clearly defined H-subgroups, which are characterized by HVS I and II motifs 16362-239, 16293-16311-195 and 16162-73, respectively, were found. A transition, T → C at np 239, appears to be a relatively stable marker for H-16362 sequence types, being found only sparsely on the background of haplogroup I (Table 2). Variant 73G is a relatively stable marker for those HVS I types which distinguish themselves from the CRS by a variant 16162G. However, HVS I sequence types determined by motif 16293, 16311 exhibit diagnostic variant 195C in a position that seems to be hypervariable (Table 2).

The node designated as HV* (Richards *et al.* 1998, 2000) is highly important in mtDNA phylogeny because two of the most frequent haplogroups in Europe, H and pre-V, descend from it. The haplogroup HV*, rare in European populations, was identified in Polish and Russian samples with low frequency (1% and 2%, respectively). However, these sequences are heterogeneous and belong to various HV*-subgroups, determined by variants 16067T, 16311C or 73G. A single sequence type 16126C-16362C, which was found among Russians, is a member of cluster pre-HV (Richards *et al.* 2000).

Haplogroup pre-V sequences, defined by the CR motif 16298-72 (Torroni *et al.* 2001), were present in Poles and Russians at frequencies of 4.8% and 5.5%, respectively. All of these samples harbored a full CR motif of haplogroup pre-V, with the exception of three Polish individuals who do not have a marker at np 72, but belong nevertheless to the haplogroup pre-V, being found in association with RFLP variant

+15904*Mse*I. In addition, the HVS I haplogroup pre-V sequences lacking 16298C variant were found in one Polish and one Russian individual. It may be noted that haplogroup pre-V frequencies in Poles and Russians correspond to those observed in other Western, Central and Northern European populations (Table 1 in Torroni *et al*. 2001).

Phylogenetic studies have shown that haplogroups J and T stem from a common node which is distinguished from the ancestral node R* by polymorphisms at nps 4216, 11251, 15452 and 16126 (Macaulay *et al*. 1999; Finnilä *et al*. 2001*b*). Both haplogroups are widely distributed in European populations as well as in the Polish and Russian samples presented here. Haplogroup T represents 11.5% of the Polish and 11% of the Russian mtDNAs, and includes two distinct subgroups, T* and T1, distributed among Poles and Russians with equal frequencies. On the basis of HVS I motifs, subhaplogroup T* may be further differentiated into several subclusters; but their phylogenetic reliability appears to be ambiguous due to the influence of several unstable nucleotide positions, such as 16296, 16292 and, possibly, 16304 (Malyarchuk & Derenko, 1999; Richards *et al*. 2000; Finnilä & Majamaa, 2001).

Haplogroup J sequences in Poles and Russians are characterized by similar frequencies of 7.8% and 8%, respectively. Based on HVS I polymorphisms, this haplogroup can be divided into four subgroups, determined by motifs 16069-16126 (J*), 16069-16126-16145-16222-16261 (J1b), 16069-16126-16145-16231-16261 (J1a), and 16069-16126-16278 (J2) (Richards *et al*. 1998). Based on HVS II mutations, haplogroup J is characterized by marker variant 295T and can be further divided into subgroups, represented by additional motifs 185-228 (for J*), 242 (for J1b), 150-195 (for J1a), and 150 (for J2). Phylogenetic analysis of the complete mtDNA sequences revealed a subdivision of the haplogroup J into two major subclusters. One is determined by polymorphism at np 3010 and combines subgroups J* and J1b. The second subcluster is defined by polymorphisms at nps 7476 and 15257, and consists of J1b and J2 (Finnilä & Majamaa, 2001). Therefore, comparison of the mtDNA coding region with non-coding region variability data suggests that parallel mutations at nps 16145, 16172 and 16261 should be assumed to obtain a concordant network for haplogroup J (Finnilä & Majamaa, 2001). In addition, np 16222 has undergone two independent mutations, being found in J1b and J* Polish sequences (Table 1). Similarly, identical HVS I sequence types 16069-16126-16311 were observed in Poles and Russians as belonging to two different subgroups, J* and J1a, based on HVS II information. Subgroup J* was found to be predominant among Poles and Russians and was present at population frequencies of 4.6% and 5.5%, respectively. The remaining J-subclusters were found with lower frequencies, ranging from 0.9% for J1a and J2 and 1.4% for J1b in Poles, and from 0.5% for J1a and 2% for J1b in Russians.

Haplogroup U and K sequences, which are defined by a variant +12308*Hin*fI, were found in 19.5% of the Polish mtDNAs and in 20.0% of the Russian mtDNAs. Of these, haplogroup K sequences are relatively rare both in Poles and in Russians (3.4% and 3.0%, respectively). On the contrary, haplogroup U itself is widely distributed in Slavonic populations and is represented by subgroups U*, U1, U2, U3, U4, U5, U7 and U8.

U5, the most frequent and ancient subgroup of haplogroup U in Europe (Torroni *et al*. 1996; Richards *et al*. 1998, 2000), is represented in Poles and Russians by two main subgroups, U5a and U5b. On the basis of complete mtDNA sequence variation in Finns, Finnilä *et al*. (2000) found mutations at np 14793 and at nps 7768 and 14182 were shared, correspondingly, by subgroups U5a (with HVS I motif 16192-16256-16270) and U5b (with HVS I motif 16189-16192-16270). This subdivision of haplogroup U5 is confirmed by HVS II sequence data, since all of U5b-sequences which were observed in Poles and Russians have an additional marker mutation at np 150 (Appendix). Although HVS II site 150 has undergone multiple mutations on the background of different mitochondrial lineages (Table 2), it nonetheless seems to be fixed for subgroup

Table 5. *U4a sequence types distribution in different populations*

HVS I sequence	HVS II sequence	Coding region markers	Sample origin
CRS	73 195 263 310	4646 12308	Poles[1]
CRS	73 152 195 263 310	4646 12308	Russians[1]
CRS	66 73 195 263 310 315D	4646 12308	Poles[1]
CRS	73 195 263 310	ND	Austrians[2]
16129 16362	73 195 263 310	4646 12308	Russians[1]
16189	73 195 263 310	4646 12308	Poles[1]
16294	73 195 263 310	4646 12308	Poles[1]
16294	ND	12308	Nenets[3]
16263	73 195 263 310	4646 12308	Russians[1]
16356	73 143 195 263 310	4646 12308	Poles[1]
16356	73 195 263 310 315D	ND	Germans[4]
16223 16356	73 195 263 310	4646 12308	Poles[1], Finns[5]

Data from the following studies were analyzed: [1] Present study, [2] Parson *et al.* 1998, [3] Saillard *et al.* 2000, [4] Baasner & Madea, 2000, [5] Finnilä *et al.* 2001*a*. ND, not determined.

U5b. The distribution of the subgroup U5a and U5b frequencies in Poles and Russians is approximately equal, with the U5a subgroup prevailing over U5b – 5.3% and 3.4% in Poles, and 7.5% and 3% in Russians.

U4 (with CR motif 16356-195) is the next relatively frequent subgroup in the populations studied, being found at a frequency of 5% in Poles and 3.5% in Russians. Phylogeographic studies revealed that two major founder clusters characterize U4, determined by HVS I motifs 16356 and 16134-16356 (Richards *et al.*1998, 2000); it was also suggested that the latter subgroup appears to be specific for Central and Eastern European populations. In this study, 16134-16356 sequences with low frequencies of 1.4% in Poles and 0.5% in Russians were observed. Perhaps more importantly, among Poles and Russians 14 HVS I sequences which belong to haplogroup U (+12308*Hin*fI) have been identified, but they do not share any mutations with subgroup-specific polymorphisms within haplogroup U (Table 5). All of these sequence types, as well as some members of subgroup U4, are characterized by HVS II motif 73-310. These samples were tested for the presence of a U4-diagnostic site +4643*Rsa*I and it was found that all of them belong to the U4-subgroup. In accordance with the style of established mtDNA nomenclature (Richards *et al.* 1998; Macaulay *et al.* 1999) we designated U4-sequences with the 310C variant in HVS II as belonging to clade U4a. Analysis of the published HVS I and II data allowed us to reveal U4a sequence types, although at a low frequency, in populations of Finno-Ugric-speaking Finns and Nenets, and German-speaking populations of Austrians and Germans (Table 5). Nevertheless, the current data on population distribution of U4a sequences led us to assume that the majority of them are characteristic for Poles and Russians, where this U4-subcluster was found with a frequency of 2.3% and 2.0%, respectively.

The geographic picture of the U4a sequence distribution remains unclear, since many published population data on the HVS I and II variability appear to be insufficient to determine an exact phylogenetic status of the CR sequences (such as CRS-73, for instance) without the support of coding-region sites. This study has observed CRS-73 sequences belonging to haplogroups H and HV*. Therefore, additional detailed studies are required to elucidate the origin and diversification of the U4a subcluster in Europe. In addition, phylogenetic relationships between control region sequences belonging to the U4 subgroup remain ambiguous, and therefore, the branching order of these sequence types cannot be resolved (Figure 1). The median network demonstrates that two possible phylogenetic directions are possible. The first scenario suggests that mutation at np 310 appeared later than marker mutation at np 16356, and further diversification of the U4a occurred after back-mutation at np 16356. On the contrary, the second scenario suggests that mutation at np 310 outstripped change at np 16356, and hence, the U4a subcluster may be

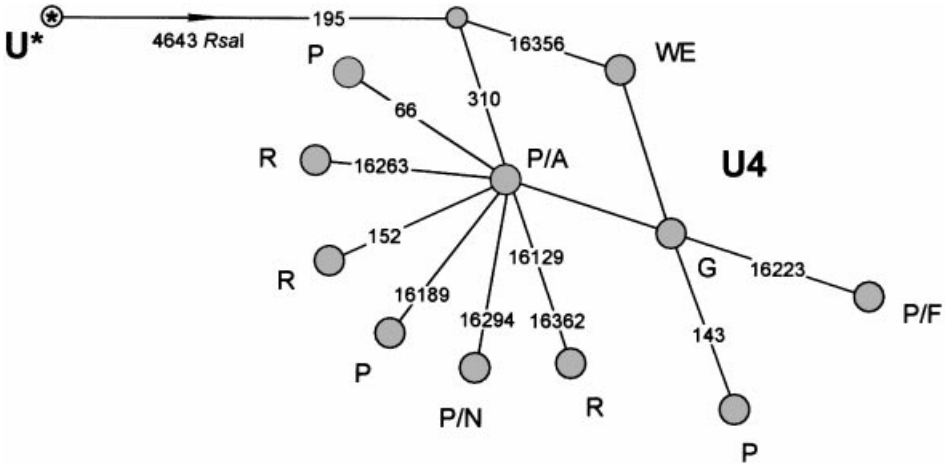

Fig. 1. Schematic phylogenetic network of the subhaplogroup U4 sequence types. The node U*, labelled by an asterisk (*), is defined by the RFLP variant +12308*Hin*fI and 73G variant in the HVS II, in comparison with the revised CRS (Andrews *et al.* 1999). The deletion event at np 315 was not considered. Any diversity within the node defined by 16356 variant alone is not shown. Reticulation in the network indicates ambiguity in the topology. RFLP variant is shown with the arrow pointing in the direction of a site gain. The nodes in the network represent the haplotypes found in populations (Table 5) as well as hypothetical intermediate haplotypes (empty nodes). Labelled nodes are U4a haplotypes observed in Poles (P), Russians (R), Germans (G), Austrians (A), Finns (F), Nenets (N), or in West Eurasians (WE).

considered as an ancestral state for the U4-phylogeny. Unfortunately, with the data on nucleotide stability in HVS I and II regions, we were unable to resolve this inconsistency due to an almost equal instability of nps 16356 and 310 (Tables 1 and 2). According to our data, the variant 16356C appeared twice and independently in the background of haplogroups U and H, but the variant 310C occurred independently in the background of haplogroup U, and rarely in association with H, T*, C, M* lineages. In addition, diversity estimates calculated for the two subsets of U4, with and without the 310C variant, gave similar values of $\rho = 0.929$ for U4a and $\rho = 1.083$ for the remaining U4-HVS I sequences found in Poles and Russians.

Besides subgroups U5 and U4, several minor U-subclusters were found in Polish and Russian mtDNA pools. Subgroup U1 with HVS I motif 16189-16249 (Macaulay *et al.* 1999), accompanied by variant 285T in HVS II, was present at a frequency of 1.0% in Russians. Subgroup U2 sequences, characterized by HVS I and II motif 16051-16129C-152-217-340, were observed at low frequencies, both in Poles (0.9%) and in Russians (1.5%). U3-sequences with CR motif 16343-150 appear to be rare in Poles and Russians, being found at frequencies of 0.5% and 1.0%, respectively. Similarly, U7 sequences with CR motif 16309-16318T/C-152 were present in the populations studied at low frequency (less than 1.0%). The U8-subgroup (Finnilä *et al.* 2001*b*), which is defined by motif 16342-282, was observed only in Poles at a frequency of 0.5%.

The remaining haplogroups I, W, X, N1b, N1c observed in Poles and Russians belong to the macro-haplogroup N, which also encompasses all aforementioned clusters of haplogroups (HV, JT, UK) as members of the macro-haplogroup R (Macaulay *et al.* 1999; Richards *et al.* 2000). Haplogroup I, characterized by CR motif 16129-16223-16391-199-204-250, occurred in Poles and Russians at a frequency of 1.8% and 2.5%, respectively. N1b and N1c sequences are defined by tentative HVS I motifs 16145-16176G-16223 and 16223-16265, correspondingly (Richards *et al.* 2000), and were found as individual haplotypes in Poles. Haplogroup W sequences (CR motif 16223-16292-189-204-207) were observed in Poles and Russians at frequencies of 3.7% and 2.0%. Topology of the phylogenetic network of haplogroups I and W was resolved based on mtDNA variability in the coding region, with the exception of reticulation composed of polymorphisms at nps 1719 and 8251 (Finnilä *et al.* 2001*b*). According to the phylogeny suggested by

Finnilä *et al.* (2001*b*), variant 204C appears to be ancestral for the IW branch, but various 207A originated twice as a parallel mutation in haplogroups W and I. However, population data on HVS I and II variation presented here demonstrated that combination of the variants 204C-207A is characteristic for mtDNA sequences from haplogroups W, I and N1c, implying that this motif may be considered as ancestral.

Haplogroup X was found in Poles and Russians at a frequency of 1.8% and 3.5%, respectively. This haplogroup, rare in Europe, is determined by CR motif 16189-16223-16278-153-195-225 and further subdivided into two clusters defined by mutations at nps 226 and 227. Interestingly, both in Poles (0.5%) and in Russians (1.5%) several sequence types without HVS II diagnostic mutations at nps 153, 195, 225 were observed. Several rare X-HVS I sequences defined by variants 16248T and 16266T-16274A were previously revealed in southern West Eurasian populations (Richards *et al.* 2000). In addition, X-HVS I sequences determined by variant 16241G, rare in Russians (1%), were described recently among Gypsies at a frequency of 2.2% (Gresham *et al.* 2001).

The remaining CR sequences found in Poles and Russians were classified as belonging to the East Eurasian macro-haplogroup M. Both macro-haplogroups M and N coalesce to the African cluster L3, which is considered as the most recent ancestor of all Eurasians (Quintana-Murci *et al.* 1999; Ingman *et al.* 2000). M-haplogroups such as C, D, E, G and Z are very rare in western European populations. We have observed members of the haplogroups C, D, E, G and M* in Poles and Russians at a frequency of 1.8% and 1.5%, respectively. However, diversity of the M-CR sequence types was high, both in Poles and in Russians. Haplogroup C sequences defined by CR motif 16223-16298-16327-249D were present in Poles. Haplogroup C sequences were previously also described at low frequency in Russian populations (Orekhov *et al.* 1999; Malyarchuk *et al.* 2001). In addition, haplogroup Z sequences were revealed in Russians at a frequency of 1.3% (Orekhov *et al.* 1999; Malyarchuk & Derenko, 2001). Interestingly, both haplogroup C and Z sequences are characterized by the deletion of an adenine residue at np 249 (variant 249D). According to the phylogenetic data based on variation in the complete mtDNA sequences, both haplogroups C and Z have shared polymorphisms at nps 4715, 7196CA, and 8584 (Finnilä *et al.* 2001*b*; Maca-Meyer *et al.* 2001) and should be considered as sister haplogroups (Kivisild *et al.* 2001). Haplogroup Z sequences were found in many Siberian/Central Asian populations (Kolman *et al.* 1996; Derenko & Shields, 1997; Schurr *et al.* 1999; Derenko *et al.* 2000) as well as in Saami (Sajantila *et al.* 1995). The Saami gene pool is also characterized by the presence of the D-lineage with motif 16126-16136-16189-16223-16360-16362, found at a low frequency of 4.7% (Delghandi *et al.* 1998). In the present study, an identical sequence type was found among Russians. A similar CR sequence type, observed in Poles, belongs to the 16189-subcluster of haplogroup D. In addition, both Polish and Russian samples are characterized by the presence of the Saami-specific U5b-motif (16144-16189-16270) found at a frequency of 0.5% in Poles and 1.5% in Russians. The presence of the Saami-specific mtDNAs from haplogroups D and U5b, as well as haplogroup Z sequences, in the mitochondrial gene pool of Russians was considered as a consequence of local Finno-Ugric tribe assimilation by Slavs during their movement to the north of Eastern Europe, a trend suggested previously by anthropologists (Alekseeva, 1973).

The remaining M-sequences in Poles and Russians were identified as belonging to haplogroups G, E and M*. In the case of haplogroup G, both Russian and Polish sequence types had both G and E specific RFLPs (+4830*Hae*II/+4831*Hha*I for G and −7598*Hha*I for E); the latter marker originated on the background of haplogroup G due to mutation at np 7600, which gives a similar E-specific RFLP pattern (Kivisild *et al.* 2001).

Therefore, the results of mtDNA variation study demonstrated that all major West Eurasian haplogroups and their subgroups were

Table 6. *The frequency of shared haplotypes found in Poles* (*POL*) *and in Russians* (*RUS*) *in comparison with Germans* (*GER*) *and Finns* (*FIN*)

HG	HVS I sequence	HVS II sequence	POL (436)	RUS (201)	GER (560)	FIN (192)
H	CRS	263	9.4	8.0	8.9	0
H	CRS	146 195 263	0.2	0.5	0.2	0
H	CRS	152 263	1.4	1.0	2.7	0
H	CRS	195 263	0.2	0.5	0.5	0
H	16093	263	0.5	2.5	0.2	0
H	16129	263	0.7	1.0	0.2	0
H	16274	146 263	0.2	0.5	0	0
H	16304	263	1.6	1.5	1.1	1.0
H	16051 16162 16259	73 263	0.2	0.5	0	0
H	16189 16356	263	0.5	0.5	0.4	0
H	16080 16189 16356	263	0.2	0.5	0	0
H	16189 16356 16362	263	0.7	0.5	0.4	0
H	16311	263	0.7	0.5	0.9	1.0
H	16278 16293 16311	195 263	0.2	2.0	0	0
H	16354	263	0.5	2.5	0	0
H	16362	239 263	1.4	2.0	0.5	0
HV*	CRS	73 263	0.5	0.5	?	0
K	16224 16311	73 146 152 263	0.7	1.5	0.5	2.6
U4	16093 16356	73 195 215 263	0.2	0.5	0	0
U5	16192 16256 16270	73 263	1.0	0.5	0.7	0
U5	16192 16256 16270 16399	73 152 263	0.2	0.5	0	0
U5	16192 16222 16256 16270 16399	73 263	0.2	0.5	0.2	0
U5	16256 16270 16399	73 263	0.2	1.5	0.5	0
U5	16256 16270 16399	73 152 263	0.2	1.0	0	0
J*	16069 16126	73 185 263 295	0.7	0.5	0.2	0
J*	16069 16126	73 185 228 263 295	0.2	0.5	1.1	0
J*	16069 16126 16311	73 185 263 295	0.2	0.5	0	0
J1b	16069 16126 16145 16172 16222 16261	73 242 263 295	0.7	1.5	0.2	0
T*	16126 16294 16296	73 263	0.5	1.0	1.3	0
T*	16126 16294 16296 16304	73 263	1.6	3.0	1.4	0
T1	16126 16163 16186 16189 16294	73 152 195 263	1.2	1.0	0.7	1.6
pre-V	16298	72 263	2.1	1.5	1.3	2.1
I	16129 16172 16223 16311 16391	73 199 203 204 250 263	0.2	0.5	0.4	3.1
X	16189 16223 16255 16278	73 153 195 225 227 263	0.2	0.5	0.2	0.5

HG denotes mitochondrial haplogroup. A question mark (?) denotes that haplogroup affiliation of the CR sequence type cannot be determined without additional coding-region markers.

detected in Poles and Russians. It was also found that the East Asian admixture in Poles and Russians appears to be insignificant (less than 2.0%).

MtDNA haplotypes and subclusters shared between Poles and Russians

It has been suggested, by means of phylogenetic analysis (Comas *et al.* 1997; Richards *et al.* 1998; Simoni *et al.* 2000), that European populations demonstrate limited genetic differentiation and do not exhibit any obvious geographic patterns. However, the study by Helgason *et al.* (2000) indicated that European populations contain a large number of closely related mtDNA lineages, many of which have not yet been sampled in the current comparative data set. This means that geographic patterns of mtDNA variation may exist at the level of individual lineages or lineage subclusters.

In the present study, a high level of mtDNA diversity in Poles and Russians sharing the same language group has been found. In order to

Table 7. *The frequency of shared HVS I subclusters found in Poles* (*POL*) *and in Russians* (*RUS*) *in comparison with Germans* (*GER*) *and Finns* (*FIN*)

HG	HVS I subclusters	POL (436)	RUS (201)	GER (560)	FIN (192)
H	16129, 16129-16316	0.7	2.5	0.7	0.5
H	16256, 16256-16352, 16256-16319	0.2	2.5	0.4	0
H	16291	0.2	0.5	0.4	0
H	16278-16293-16311	0.7	1.5	0.2	0
H	16092-16140-16265-16293-16311	2.8	0.5	0.2	0
H	16192-16304-16311	0.2	1.0	0.5	0
H	16189-16356	1.6	0.5	0.4	0
H	16080-16189-16356	0.5	1.0	0.2	0
H	16189-16356-16362	1.2	1.0	1.1	0
H	16354	0.7	4.5	0	1.0
pre-V	16153-16298	0.5	1.0	0.2	2.6
J*	16069-16126-16311	0.2	0.5	0	1.0
U2	16051-16129C-16189-16256	0.5	0.5	0.2	0
U4	16093-16356	0.2	0.5	0	0
U4a	CRS, 16356	2.3	2.0	0.4	0.5
U5	16192-16222-16256-16270-16399	0.2	1.0	0.2	0
U5	16192-16256-16270-16291-16399	0.9	0.5	0.5	0
W	16223-16292-16325	0.7	0.5	0	0

HG denotes mitochondrial haplogroup.

investigate genetic similarity between them at the level of shared mtDNA haplotypes and their subclusters, the total number of CR sequence types has been reduced by means of removing polymorphic variants at unstable sites (such as A → C transversions at nps 16182 and 16183) and insertions of additional C residues in the HVS I and II poly-C tracts. As a result, out of 297 and 142 CR sequence types observed in Poles and Russians, respectively, 34 are shared between these population samples. Table 6 shows the distribution of shared haplotypes between Poles and Russians in comparison with Germans and Finns. The latter populations were selected in accordance with their geographic proximity and the historical evidence concerning their participation in the formation of modern Poles and Russians. The results of this analysis indicate that only a small fraction of the CR haplotypes (10 out of 34 haplotypes) appear to be actually shared between Poles and Russians, not being found in German and Finnish gene pools. These haplotypes belong to five different haplogroups – H, HV*, U4, U5, and J*. It should be noted, however, that the majority of these haplotypes belong to subclusters which can be found in common among many West Eurasian populations.

In order to identify subclusters of CR haplotypes which are specific mainly for Poles and Russians, the distribution of haplotypes that differ by the fewest number of base substitutions in Poles and Russians and their neighbours, Germans and Finns, were analyzed. Although almost all of the mtDNA subclusters observed in Poles and Russians can be accounted for in many European populations, this analysis allowed us to reveal at least 16 subclusters of relatively rare haplotypes which have a preferential distribution among Poles and Russians (Table 7). In Russians, H-subclusters determined by HVS I motifs 16129 (and 16129-16316), 16256 (and 16256-16352, 16256-16319), and 16354 were found at a relatively high frequency, ranging from 2.5 % to 4.5 %. In Poles, sequence types with motif 16092-16140-16265-16293-16311 occurred at the frequency of 2.8 %. Similar haplotypes, defined by motif 16293-16278-16311, were also common between Russians and Poles (1.5 % and 0.7 %, respectively). However, it is known that the subcluster of H-haplotypes determined by motif 16293-16311 has a pan-European distribution (Richards & Macaulay, 2000). Moreover, the highest frequency (6.1 %) of this H-subcluster was found in Estonians (Tambets *et al*. 2000). Another H-subcluster, defined by motif

16189-16356 and its branches 16080-16189-16356 and 16189-16356-16362, was found frequently in Poles (3.3%) as well as in Russians (2.5%) and Germans (1.7%). A relatively high occurrence of H1-sequences determined by motif 16192-16304-16311 is characteristic for Russians in comparison to Poles and Germans, but another H1-branch (with motif 16294-16304) is clearly common between Germans and Poles.

Taking into account the data presented in Tables 6 and 7, one can conclude that we were not able to find any specific combinations of unique mtDNA haplotypes and their subclusters clearly distinguishing Poles and Russians, as Slavonic-speaking populations, from the neighboring European populations such as Germans and Finns. This trend was also noted in a previous study on the HVS I-RFLP variation in Russians in comparison with Western and Eastern European populations (Malyarchuk & Derenko, 2001). One possible exception is subgroup U4a. This subgroup comprises 10 (2.3%) out of 436 Poles, 4 (2.0%) out of 201 Russians, 2 (0.4%) out of 560 Germans (Parson *et al.* 1998; Baasner & Madea, 2000) and 1 (0.25%) out of 403 Finns (Finnilä *et al.* 2001*a*). Given the relatively high frequency and diversity of U4a among Poles and Russians and its low frequency in the neighbouring German and Finnish populations, one can suggest a central-eastern European origin of U4a. It is possible that the subsequent dispersal of this mtDNA subgroup in Eastern European populations was due to Slavonic migrations. Undoubtedly, to elucidate the origin of the U4a subgroup, additional analysis is required followed by a much more extensive sampling of Slavonic and other European populations.

CONCLUSION

Analysis of mtDNA variation, performed by means of sequencing two hypervariable segments and assaying of haplogroup-diagnostic polymorphisms in the coding region, appears to be an effective genetic tool for inferring the genetic history of populations (Macaulay *et al.* 1999). In the present study, we have found that Poles and Russians are characterized by the same West Eurasian mtDNA haplogroups which describe at least 95% of mtDNA variations in Europe and the Near East (Torroni *et al.* 1996; Richards *et al.* 1998, 2000). Although there is a good correspondence between CR sequences and RFLPs grouping into monophyletic mtDNA clusters, and this looks to be strongest when only HVS I sequences are used in comparison (Bandelt *et al.* 2000); the addition of the HVS II sequences may be extremely useful in the resolution of the phylogenetic relationships among some uncertain mitochondrial lineages. A good example of this is subgroup U4a which does not have a certain HVS I motif, but can be recognized on the basis of HVS II information.

Despite the high level of mtDNA variation in Poles and Russians, both populations exhibit a similar pattern of mtDNA haplogroup distribution, which is also typical for many European populations studied. Moreover, the analysis of distribution of CR haplotypes and subclusters shared between Poles and Russians has shown that both Slavonic populations share them mainly with Germans and Finns. These data allow us to suggest that Europeans, despite their linguistic differences, originated in the common genetic substratum which predates the formation of the most modern European populations. It seems that considerable genetic similarity between European populations, which has been revealed by mtDNA variation studies, was further accelerated by a process of gene redistribution between populations due to the multiple migrations occurring in Europe during the past millenia (Sykes, 1999; Helgason *et al.* 2000; Richards *et al.* 2000). As for Slavonic-speaking populations, the evidence from the present study and our previous work (Malyarchuk & Derenko, 2001) suggests that the assumption of a common central European origin of Slavs should be tested with additional studies of mtDNA and Y chromosome variability in Slavonic populations inhabiting different regions of Europe.

APPENDIX

mtDNA haplotypes and their distribution in Polish and Russian populations

HVS I (minus 16000)	HVS II	HG	POL	RUS
CRS	263 309.1 315.1	H	12	8
CRS	263 315.1	H	22	5
CRS	263 309.1 309.2 315.1	H	7	3
CRS	263 309.1 315.1 337	H	1	
CRS	143 228 263 309.1 315.1	H	1	
CRS	146 263 309.1 315.1	H	2	
CRS	146 152 263 309.1 315.1	H		1
CRS	146 263 309.1 309.2 315.1	H	1	
CRS	146 195 263 309.1 315.1	H	1	1
CRS	150 263 315.1	H	1	
CRS	93 152 263 309.1 315.1	H	1	
CRS	152 263 315.1	H	4	1
CRS	152 263 309.1 315.1	H	2	
CRS	152 263 309.1 309.2 315.1	H		1
CRS	152 195 263 309.1 315.1	H	1	
CRS	152 199 263 309.1 309.2 315.1	H	1	
CRS	152 204 309.1 309.2 315.1	H		1
CRS	186 263 315.1	H	1	
CRS	186 263 309.1 315.1	H	1	
CRS	195 263 315.1	H	1	
CRS	195 263 309.1 309.2 315.1	H		1
CRS	199 263 309.1 315.1	H	1	
CRS	228 263 309.1 315.1	H	1	
CRS	262 263 309.1 315.1	H	1	
CRS	263 269 315.1	H	1	
CRS	263 315.1 357	H	1	
CRS	73 146 263 309.1 309.2 315.1	H	1	
CRS	73 182 263 309.1 315.1	H	1	
066 172 218 318AC 328CA	195 198 263 309.1 309.2 315.1	H	1	
093	263 309.1 315.1	H	2	4
093	263 315.1	H		1
093 129 316	73 263 315.1	H		1
093 129 189 193.1 316	207 263 309.1 309.2 315.1	H		1
095	93 263 309.1 315.1	H	1	
111 189 193.1	152 182 263 309.1 309.2 315.1	H	1	
114 270	146 263 309.1 315.1	H	1	
129	263 315.1	H	1	
129	263 309.1 315.1	H	2	1
129	N/A	H		1
129 210	263 309.1 309.2 315.1	H		1
148 247	263 315.1	H	1	
168	152 263 309.1 315.1	H	1	
176	195 263 315.1	H		1
179	263 309.1 315.1	H	1	
183 239CG	152 263 309.1 315.1	H	1	
209	263 315.1	H	1	
209	42.1 146 182 215 263 309.1 315.1	H	1	
111 209 218	263 309.1 315.1	H	1	
222	263 315.1	H	1	
231C/T	93 263 309.1 315.1	H		1
189 291	263 309.1 315.1	H		1
235 291	263 309.1 315.1	H	1	
239	263 315.1	H	1	
244 399	207 263 309.1 309.2 315.1	H	1	
250	263 309.1 309.2 315.1	H	1	
256	200 263 315.1	H		1
256	263 309.1 315.1	H		1
256 352	263 309.1 315.1	H		2
261	263 309.1 315.1	H	1	
261	152 263 309.1 315.1	H		1
265	152 263 309.1 309.2 315.1	H		1
266	152 263 315.1	H	1	
270	315.1	H		1
274	146 263 315.1	H	1	
274	146 263 309.1 315.1	H		1
274	73 263 315.1	H	1	
278	263 309.1 315.1	H	1	
286 311	143 152 263 309.1 315.1	H	1	
304	263 309.1 315.1	H	4	2
304	263 315.1	H	3	1
304	146 263 309.1 315.1	H		1
304	152 263 309.1 315.1	H	1	
304	195 263 315.1	H		1
304	199 263 309.1 309.2 315.1	H	1	
093 304	263 309.1 309.2 315.1	H	1	
111CA 304	263 309.1 315.1	H	1	
153 304	263 315.1	H	1	
172 304	263 315.1	H	1	
213 304	263 309.1 315.1	H		1
243 304	263 315.1	H	1	
271 304	263 309.1 309.2 315.1	H	1	
092 294 304	263 315.1	H	1	
294 304	263 315.1	H	1	
294 304	236 263 315.1	H	1	
192 304	195 240 263 315.1	H		1
192 304 311	263 309.1 309.2 315.1	H		1
167 192 304 311	263 309.1 315.1	H	1	
162	73 263 309.1 315.1	H	2	

APPENDIX (*cont.*)

HVS I (minus 16000)	HVS II	HG	POL	RUS
162 209	73 199 263 309.1 315.1	H	1	
162 258AC	73 263 309.1 315.1	H		1
051 162 259	73 263 315.1	H	1	1
051 162 291 304	73 263 315.1	H	1	
188CA	263 315.1	H	1	
188CG	263 315.1	H	1	
188	263 315.1	H	1	
188	263 309.1 309.2 315.1	H	1	
188CG 189	263 309.1 309.2 315.1	H	1	
189	152 263 309.1 315.1	H	1	
183C 189 193.1	263 309.1 315.1	H	1	
093C/T 183C 189 193.1	263 309.1 309.2 315.1	H	1	
189 193.1	152 263 309.1 315.1	H	1	
189 193.1	263 315.1	H	1	
189 193.1 193.2	263 315.1	H	1	
188 189 356	263 315.1	H	1	
169 183C 189 356	263 315.1	H	1	
189 193.1 356	152 263 315.1	H	2	
189 193.1 356	146 263 309.1 309.2 315.1	H	1	
080 189 356	263 309.1 315.1	H		1
080 183C 189 356	151 263 309.1 309.2 315.1	H		1
080 189 193.1 356	263 309.1 315.1	H	1	
080 189 193.1 193.2 231 356	146 263 309.1 309.2 315.1	H	1	
189 356	263 309.1 309.2 315.1	H		1
183C 189 356	263 309.1 309.2 315.1	H	1	
183C 189 356	263 315.1	H	1	
183C 189 193.1 356 360	263 315.1	H	1	
093 183C 189 356 360	263 315.1	H	1	
183C 189 356 362	263 315.1	H	1	
182C 183C 189 356 362	263 315.1	H	1	
092 183C 189 193.1 356 362	263 315.1	H	1	
189 193.1 356 362	263 309.1 309.2 315.1	H		1
189 356 362	263 315.1	H	1	
189 290 291 311 356 362	64 263 315.1	H	1	
189 318 356	263 315.1	H		1
311	263 315.1	H	3	1
311	152 263 315.1	H	1	
311	144 195 263 309.1 315.1	H	1	
311	195 263 309.1 315.1	H	1	
311 391	263 309.1 309.2 315.1	H		1
093 311	263 315.1	H		1
093 311	131 152 263 309.1 315.1	H		1
157 311	263 309.1 315.1	H	1	
221 311	263 309.1 315.1	H	1	
249	152 263 309.1 315.1	H		1
249 311	263 309.1 315.1	H	1	
042 288 290 311	263 309.1 315.1	H		1
319	263 309.1 315.1	H	1	
148 256 319	146 189 193 263 309.1 309.2 315.1	H		1
265 352	263 309.1 315.1	H	1	
278 293	195 263 315.1	H	1	
278 293 311	195 263 309.1 315.1	H	1	2
278 293 311	195 263 315.1	H		2
209 278 293 311 319	195 215 263 309.1 309.2 315.1	H	1	
129 293	263 315.1	H	1	
170 293	263 315.1	H	1	
293 311	143 195 263 309.1 315.1	H	1	
293 311	195 263 315.1	H	2	
293 311	195 263 309.1 315.1	H	1	
092	195 263 309.1 315.1	H	1	
092	263 309.1 315.1	H	1	
092 183 293 311	195 263 315.1	H	1	
092 140 265 293 311	195 263 315.1	H	8	
092 140 265 270 293 311	195 263 309.1 315.1	H	1	
092 140 265 293 311	195 204 263 315.1	H	1	
092 140 189 193.1 265 293 311	195 263 315.1	H		1
342	263 309.1 315.1	H	1	
070	152 263 315.1	H	2	
070 343	152 263 309.1 309.2 315.1	H	1	
304 343	151 263 279 309.1 309.2 315.1	H	1	
354	263 309.1 315.1	H	1	4
354	263 309.1 309.2 315.1	H	1	1
167 354	263 309.1 309.2 315.1	H	1	
193 354	127 152 263 309.1 309.2 315.1	H		1
354 399	150 263 310	H		1
354 399	N/A	H		1
399	146 263 309.1 309.2 315.1	H		1
362	239 263 315.1	H	1	
362	239 263 309.1 315.1	H	4	2
362	239 258 263 309.1 309.2 309.3 315.1 319	H	1	
362	239 263 309.1 309.2 309.3 315.1 319	H	1	
362	239 263 309.1 309.2 315.1	H	1	2
182C 183C 189 362	146 239 263 309.1 309.2 309.3 315.1	H		1
193 219 362	93 204 239 263 309.1 315.1	H	1	
193 219 319 362	204 239 263 315.1	H	1	
300 325C/T 362	239 263 309.1 315.1	H		1
137AT 176 256 311	73 152 263 295 309.1 315.1	H	1	
CRS	72 263 309.1 309.2 315.1	pre-V		1
380	72 263 315.1	pre-V	1	

appendix (*cont.*)

HVS I (minus 16000)	HVS II	HG	POL	RUS
298	72 263 315.1	pre-V		1
298	72C/T 195 263 309.1 315.1	pre-V		1
298	72 195 263 309.1 309.2 315.1	pre-V		1
298	72 263 309.1 315.1	pre-V	6	2
298	72 263 309.1 309.2 315.1	pre-V	3	
298	72 195 228 263 309.1 315.1	pre-V		1
104CA 298	72 151 228 263 309.1 315.1	pre-V	1	
126 298	72 263 309.1 315.1	pre-V		2
150 298	42.1 72 263 309.1 309.2 315.1	pre-V	1	
169 298	72 263 309.1 315.1	pre-V	1	
216 298	263 309.1 309.2 315.1	pre-V	2	
218 298	72 189 194 263 309.1 309.2 315.1	pre-V	1	
270 298	72 263 315.1	pre-V	1	
298 311	195 263 309.1 309.2 315.1	pre-V	1	
153 298	72 93 263 309.1 315.1	pre-V	1	
153 298	72 93 195 263 309.1 315.1	pre-V		1
153 298	72 93 195 263 315.1	pre-V		1
153 189 298	72 93 195 263 309.1 315.1	pre-V	1	
291 298	72 93 195 263 309.1 309.2 315.1	pre-V	1	
CRS	73 263 315.1	HV*		1
CRS	73 263 309.1 315.1	HV*	2	
067 355	150 200 263 309.1 315.1	HV*	1	
295	146 263 315.1	HV*		1
295	263 309.1 309.2 315.1	HV*		1
311	93 263 309.1 309.2 315.1	HV*	1	
234 311	263 315.1	HV*		1
126 362	60.1 64 151 152 197 198 263 309.1 309.2 315.1	pre-HV		1
069 126	73 146 185 188 222 228 263 295 315.1	J*	2	
069 126	73 185 188 228 263 295 309.1 315.1	J*	3	
069 126	73 185 263 295 315.1	J*	1	
069 126	73 185 263 295 309.1 315.1	J*	1	1
069 126	73 185 263 295 309.1 309.2 315.1	J*	1	
069 126	73 146 185 188 228 263 295 315.1	J*	1	
069 126	73 185 228 263 295 315.1	J*	1	1
069 126	73 185 210 228 263 295 315.1	J*	1	
063 069 126	73 228 263 295 315.1	J*	1	
069 078D 126	73 185 188 228 263 295 315.1	J*		1
069 126 189 193.1	73 185 228 263 295 309.1 315.1	J*	1	
069 126 145 183C 189	73 185 228 263 295 315.1	J*		1
069 126 213	73 146 185 188 228 263 295 315.1	J*		1
069 126 222	73 185 228 263 295 315.1	J*	1	
069 126 230	73 185 228 263 295 315.1	J*		1
069 126 249	73 185 188 228 263 295 315.1	J*	1	
069 092 126 261	73 185 228 263 295 315.1	J*		1
069 093 126 290	73 185 188 228 263 295 309.1 315.1	J*		1
069 126 311	73 185 263 295 315.1	J*	1	1
069 126 319	73 152 185 228 263 295 315.1	J*	1	
069 126 319	73 185 228 263 295 315.1	J*	1	
069 126 186 362	73 185 188 228 263 295 309.1 309.2 315.1	J*		1
069 126 324 366	73 185 188 228 263 295 309.1 315.1	J*	1	
069 126 366	73 185 188 228 263 295 315.1	J*		1
069 126 366 390	73 185 188 263 295 309.1 315.1	J*	1	
069 126 145 172 222 261	73 242 263 295 309.1 315.1	J1b		1
069 126 145 172 222 261	73 242 263 309.1 315.1	J1b	1	
069 126 145 172 222 261	73 242 263 295 315.1	J1b	3	2
069 092 126 145 172 222 261	73 146 242 263 295 315.1	J1b	1	
069 093 126 145 172 222 261	73 242 263 295 309.1 309.2 315.1	J1b	1	
069 126 145 172 192 222 261	73 242 263 295 315.1	J1b		1
069 126 145 231 261	73 150 152 195 215 263 295 309.1 315.1 319	J1a	2	
069 126 145 189 193.1 231 261	73 150 152 195 215 263 295 309.1 315.1 319	J1a	1	
069 126 311	73 150 195 204 263 295 309.1 315.1	J1a		1
069 126 241	150 195 263 295 315.1	J1a	1	
069 126 193 278	73 150 263 295 309.1 315.1	J2	1	
069 126 193 278	73 150 152 263 295 309.1 315.1	J2	1	
069 126 193 278 319	73 150 263 295 309.1 315.1	J2	1	
069 126 278	73 263 315.1	J2	1	
126 294	73 152 263 315.1	T*	1	
126 185 294	73 152 263 309.1 315.1	T*		1
126 192 292 294	73 146 152 263 279 309.1 315.1	T*	1	
126 146 284 292 294 296	73 263 315.1	T*		2
126 294 296	73 263 315.1	T*	1	1
126 294 296	73 263 309.1 315.1	T*	1	1
126 209 294 296	73 263 315.1	T*	1	
126 294 296 301	73 263 310	T*	1	
126 294 296 320	73 263 315.1	T*	1	
126 171 294 296 324	73 263 315.1	T*		1
126 294 296 324	73 152 263 309.1 315.1	T*	1	
126 294 304	73 263 315.1	T*		2
126 294 304	73 263 309.1 309.2 315.1	T*	1	
126 294 304	73 263 309.1 315.1	T*	1	
126 294 304 355	73 152 263 309.1 315.1	T*	3	
126 172 192 294 304	73 263 309.1 315.1	T*	1	
126 172 189 214 294 304	73 263 309.1 315.1	T*	1	
126 222 294 304	73 263 309.1 315.1	T*		1
126 294 296 304	73 146 263 309.1 315.1	T*	1	
126 294 296 304	73 150 263 315.1	T*	1	
126 294 296 304	73 152 263 315.1	T*	2	
126 294 296 304	73 263 315.1	T*	2	4
126 294 296 304	73 263 309.1 315.1	T*	4	2
126 294 296 304	73 263 309.1 309.2 315.1	T*	1	

APPENDIX (*cont.*)

HVS I (minus 16000)	HVS II	HG	POL	RUS
126 294 296 304	73 207 263 309.1 309.2 315.1	T*	1	
294 296 304	73 263 315.1	T*	2	
126 129 294 296 304	73 263 315.1	T*	1	
126 294 296 304 318	73 150 263 309.1 315.1	T*	1	
126 234 294 296 304	73 152 263 315.1	T*	1	
126 172 227 294 296 304	73 263 315.1	T*	1	
126 271 294 296 304	73 263 315.1	T*	1	
271 294 304	73 315.1	T*		1
126 260 294 296 304	41 73 263 309.1 315.1 319	T*	1	
126 294 296 304 362	73 263 315.1	T*		1
126 294 296 304 362	73 146 199 263 315.1	T*		1
126 294 296 304 399	73 263 315.1	T*	1	
126 147 294 296 297 304	73 263 309.1 315.1	T*	1	
126 147 189 294 296 297 304	73 240 263 309.1 309.2 315.1	T*	1	
126 140 183C 189 193.1 294 296 311	73 195 263 309.1 315.1	T*	1	
126 182C 183C 189 294 296 298	73 195 263 309.1 309.2 315.1	T*	1	
126 294 296 298	73 199 263 309.1 315.1	T*	1	
126 129 163 189 243 294	73 263 309.1 315.1 385	T1		1
126 163 186 189 294	73 152 195 263 309.1 315.1	T1	3	1
126 163 186 189 294	73 152 195 263 315.1	T1	2	
126 163 186 189 294	73 195 263 315.1	T1	1	
126 163 186 189 294	73 152 195 263 309.1 309.2 315.1	T1		1
126 163 170 186 189 294	73 152 195 263 315.1	T1	1	
126 186 189 209 294	73 152 195 263 309.1 315.1	T1	1	
126 163 186 189 293 294	73 152 195 263 315.1	T1		1
126 163 186 189 294 319	73 263 309.1 315.1	T1	1	
224 311	73 204 263 315.1	K		1
224 311	73 263 309.1 315.1	K	1	
224 311	73 195 263 315.1	K	1	
224 311	73 146 195 263 315.1	K	1	
224 311	73 146 152 263 315.1	K	2	3
224 311	73 146 152 263 309.1 309.2 315.1	K	1	
093 224 311	73 247 250 263 315.1	K	1	
093 192 224 311	73 150 195 263 315.1	K	1	
093 224 256 311	73 263 309.1 315.1	K	1	
224 274 311	73 195 263 315.1	K	1	
224 291 311	73 263 309.1 309.2 315.1	K	1	
224 293 311	73 263 309.1 315.1	K	1	
189 192 224 311	73 195 263 309.1 309.2 315.1	K	1	
093 183C 189 193.1 224 311	73 189 195 215 263 315.1	K	1	
224 311 368	73 146 152 263 309.1 315.1	K		1
069 092 224 311	73 146 152 263 315.1 324	K		1
172 189 234 311	73 151 195 263 315.1	K	1	
086 239 311 320	73 150 263 315.1	U*	1	
129 183C 189 224 249 288	73 150 195 263 285 315.1 385	U1		1
182C 183C 189 249	73 263 285 309.1 315.1	U1		1
051 129C 183C 189 193.1 209 362	73 217 228 263 315.1 318 340	U2	1	
051 129C 183C 189 193.1 362	73 152 217 263 309.1 309.2 315.1 340	U2	1	
051 093 129C 182C 183C 189 193.1 362	73 152 217 263 309.1 309.2 315.1 340	U2		1
051 093 129C 183C 189 362	73 152 217 263 309.1 315.1	U2		1
051 129C 189 256	73 152 217 263 315.1 340	U2	1	
051 129C 189 209 256	73 152 217 263 315.1 340	U2	1	
129C 183C 189 256	73 152 217 263 315.1 340	U2		1
343	73 150 152 263 315.1	U3		1
343	73 150 179 195 263 315.1	U3	1	
168 343	73 150 152 263 309.1 315.1	U3	1	
325 343	73 150 263 315.1	U3		1
CRS	73 195 263 310	U4a	3	
CRS	73 152 195 263 310	U4a		1
CRS	66 73 195 263 310 315D	U4a	1	
129 362	73 195 263 310	U4a		1
189	73 195 263 310	U4a	1	
263	73 195 263 310	U4a		2
294	73 195 263 310	U4a	1	
356	73 143 195 263 310	U4a	1	
223 356	73 195 263 310	U4a	3	
356	73 146 195 228 263 315.1	U4*	1	
356	73 146 195 263 309.1 315.1	U4*	1	
356	73 195 263 309.1 315.1	U4*	1	
356	73 195 236 263 309.1 315.1	U4*		1
093 356	73 195 215 263 309.1 315.1	U4*	1	1
134	73 152 195 263 296 315.1	U4*	1	
134 356	73 152 195 263 309.1 315.1	U4*	3	
134 150 356	73 152 195 198 263 309.1 309.2 315.1	U4*		1
134 221 234 356	73 152 195 263 309.1 315.1	U4*	1	
134 207 356	73 152 195 204 263 315.1	U4*	1	
179 356	73 195 263 309.1 315.1	U4*	1	
179 356	73 195 263 315.1	U4*	1	
270	73 263 315.1	U5a	1	
192 256 270	73 263 315.1	U5a	4	1
093 189 192 256 270	73 263 315.1	U5a	1	
192 256 270 399	73 152 263 315.1	U5a	1	1
192 256 270 399	73 195 198 204 263 315.1	U5a		1
192 222 256 270 399	73 263 309.1 315.1	U5a	1	1
192 222 256 270 390 399	73 204 263 309.1 315.1	U5a		1
192 256 270 291 399	73 263 315.1	U5a	1	
093 192 256 291 399	73 263 315.1	U5a	1	
172 192 256 270 291 399	73 200 263 315.1	U5a	1	
192 256 270 304 399	73 263 315.1	U5a		1
192 256 270 304 399	73 263 309.1 315.1	U5a		1
129 256 270 311 399	73 146 263 299.1 309.1 315.1	U5a	1	

appendix (*cont.*)

HVS I (minus 16000)	HVS II	HG	POL	RUS
192 256 270 286 320 399	73 183 263 315.1	U5a	2	
076 192 256 270 399	73 263 309.1 315.1	U5a		1
145 189 192 256 270 399	73 195 263 309.1 315.1	U5a		1
256 270	73 263 309.1 315.1	U5a	1	
256 270	73 263 315.1	U5a	1	
256 270 399	73 263 315.1	U5a	1	
256 270 399	73 263 309.1 315.1	U5a		3
256 270 399	73 152 263 309.1 315.1	U5a	1	2
051 256 270 399	73 263 309.1 315.1	U5a	1	
256 270 362 399	73 152 204 263 309.1 315.1	U5a	1	
256 260 270 291 399	73 263 309.1 315.1	U5a		1
256 270 291 294 399	73 263 309.1 315.1	U5a	1	
114CA 192 256 270 294	73 263 309.1 315.1	U5a	1	
114CA 192 256 270 294	263 309.1 315.1	U5a	1	
189 270	73 150 263 315.1	U5b		1
189 270 291	73 150 152 263 315.1	U5b	1	
093 189 270	73 150 152 263 315.1	U5b	2	
093 189 270	73 150 152 263 270.1 315.1	U5b		1
093 189 193.1 270	73 150 263 309.1 315.1	U5b	1	
179 189 193 193.1 270	73 150 263 315.1	U5b	1	
183C 189 193.1 270 286	73 150 152 263 315.1	U5b	1	
093 182C 183C 189 193.1 270	73 150 152 263 315.1 315.2	U5b	1	
140 174 183C 189 193.1 270 288 311	73 150 263 309.1 315.1	U5b	1	
093 258 270 292 362	73 150 263 309.1 315.1	U5b		1
189 325	73 150 152 263 315.1	U5b	1	
189 217 234 270 398	73 150 263 315.1	U5b	1	
189 192 270 398	73 150 263 315.1	U5b	2	
189 270 398	73 150 263 315.1	U5b	1	
144 189 270	73 150 263 315.1	U5b		2
144 189 266 270	73 150 263 292 315.1	U5b	1	
144 183C 189 193.1 241AT 270	73 150 263 315.1	U5b	1	
144 189 193.1 270	73 150 152 243 263 315.1	U5b		1
309 318AT 362	60 73 152 263 315.1	U7	1	
073D 126 148 309 318AC	73 146 151 152 195 263 315.1	U7		1
146 342	73 263 282 309.1 315.1	U8	1	
179 342	73 263 282 309.1 315.1	U8	1	
179 187 227 245 266 274 278 362	73 194 195 246 315.1	R*	1	
071 355 357	73 81 146 150 152 263 283 309.1 315.1	R*		1
311	73 263 295 315.1	R*	1	
129 223 391	73 152 199 204 207 250 263 315.1	I	1	
129 223 391	73 152 199 204 207 239 250 263 309.1 309.2 315.1	I	1	
129 223 391	73 152 199 204 207 250 263 309.1 309.2 315.1	I	1	
086 129 223 391	73 152 199 204 207 239 250 263 315.1	I	1	
129 223 304 391	73 199 204 250 263 315.1	I		1
129 223 311	199 204 250 263 315.1	I	1	
129 223 311 391	73 199 204 250 263 309.1 315.1	I		1
129 172 223 311 391	73 199 203 204 250 263 315.1	I		1
129 172 223 311 391	73 199 203 204 250 263 309.1 315.1	I	1	
129 172 223 293 311 391	73 199 203 204 250 263 309.1 315.1	I	1	
129 172 223 311 391	73 199 204 250 263 315.1	I	1	
129 172 189 223 311 391	73 199 250 263 309.1 315.1	I		1
129 172 223 294 311 391	73 199 203 204 250 263 309.1 315.1	I		1
145 176G 223 390	73 152 263 315.1	N1b	1	
201 223 265	75 189 195 204 207 210 263 309.1 315.1	N1c	1	
223 292	73 189 194 195 204 207 263 309.1 315.1	W		1
223 292	73 189 194 195 199 204 207 263 309.1 315.1	W	1	
223 292	73 146 195 204 207 263 309.1 315.1	W	1	
223 292	73 143 189 194 195 204 207 263 315.1	W	2	
223 292	73 119 189 195 204 207 263 315.1	W	1	
223 292 311	73 119 152 189 195 204 207 263 315.1	W	1	
223 292 311	73 189 195 204 207 263 309.1 315.1	W	1	
223 292 362	73 189 194 195 204 207 263 309.1 315.1	W	1	
223 292 362	73 189 194 195 207 263 315.1	W	2	
071 129 223 292	73 150 189 194 195 199 204 207 263 309.1 315.1	W	1	
178 223 292	73 189 194 195 204 207 263 315.1	W		1
223 292 295	73 119 152 189 195 207 263 315.1	W	1	
192 223 292 325	73 189 194 195 204 207 263 309.1 315.1	W	1	
192 223 292 325	73 189 194 195 204 207 263 315.1	W	1	
192 223 325	73 189 194 195 204 207 215 263 309.1 315.1	W		1
223 292 325	73 189 194 195 207 263 309.1 315.1	W	1	
189 223 292 295	73 189 195 204 207 263 315.1	W		1
223 292 320	73 189 195 204 207 263 315.1	W	1	
223 278	73 153 195 225 226 263 309.1 315.1	X	1	
189 223 278	73 153 195 225 226 263 309.1 315.1	X		1
183C 189 223 278	73 146 153 195 198 225 226 309.1 315.1	X	1	
183C 189 223 255 278	73 153 195 225 227 263 315.1	X		1
183C 189 223 255 278	73 153 195 225 227 263 309.1 315.1	X	1	
183C 189 223 255 278 300	73 153 195 225 227 263 315.1	X		2
086 182C 183C 189 223 255 278 300	73 153 195 225 227 263 315.1	X	1	
108 189 193.1 193.2 223 255 278	73 153 195 225 263 315.1	X	1	
183C 189 223 255 278 344	73 153 195 225 227 263 309.1 315.1	X	1	
189 223 248 278	73 153 195 199 263 309.1 315.1	X		1
126 183C 189 223 241 278	73 263 315.1	X		1
093 189 193.1 223 241 278	73 195 204 263 315.1	X		1
183C 189 223 266 274 278 390	73 146 152 182 195 263 309.1 309.2 315.1	X	1	
183C 189 193.1 223 266 274 278	73 146 152 195 263 309.1 315.1	X	1	
124 223	73 151 152 195 263 315.1	L3	1	
093 223 234 288 298 327	73 249D 263 315.1	C	2	
093 223 234 288 298 327	54 73 249D 263 315.1	C	1	
189 193.1 223 288 298 327	73 152 249D 263 310	C	1	
126 136 182C 183C 189 193.1 223 360 362	73 150 263 309.1 309.2 315.1	D		1

APPENDIX (*cont.*)

HVS I (minus 16000)	HVS II	HG	POL	RUS
092 102 164 182C 183C 189 193.1 223 266 362	42.1 73 150 263 309.1 309.2 315.1	D	1	
223 227 234 278 362	73 263 309.1 315.1	G		1
093 209 223 227 234 278 309 362	73 152 263 315.1	G	1	
129 148 192 223 291 298	73 263 310	M*	1	
223 234 300 316 362	73 153 263 315.1	M*		1
223 278 362	73 260 263 309.1 315.1	E	1	

Sample codes: POL, Poles; RUS, Russians. Mutations are shown indicating positions relative to the CRS (Anderson *et al*. 1981). The nucleotide positions in HVS I and II sequences correspond to transitions; transversions are further specified. Haplogroup names (HG) are given in capital letters according to the mtDNA classification (Macaulay *et al*. 1999; Richards *et al*. 2000). The presence of insertions or deletions is referred by .1, .2 and .3 or D, respectively, following the nucleotide position.

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