

# Major genomic mitochondrial lineages delineate early human expansions

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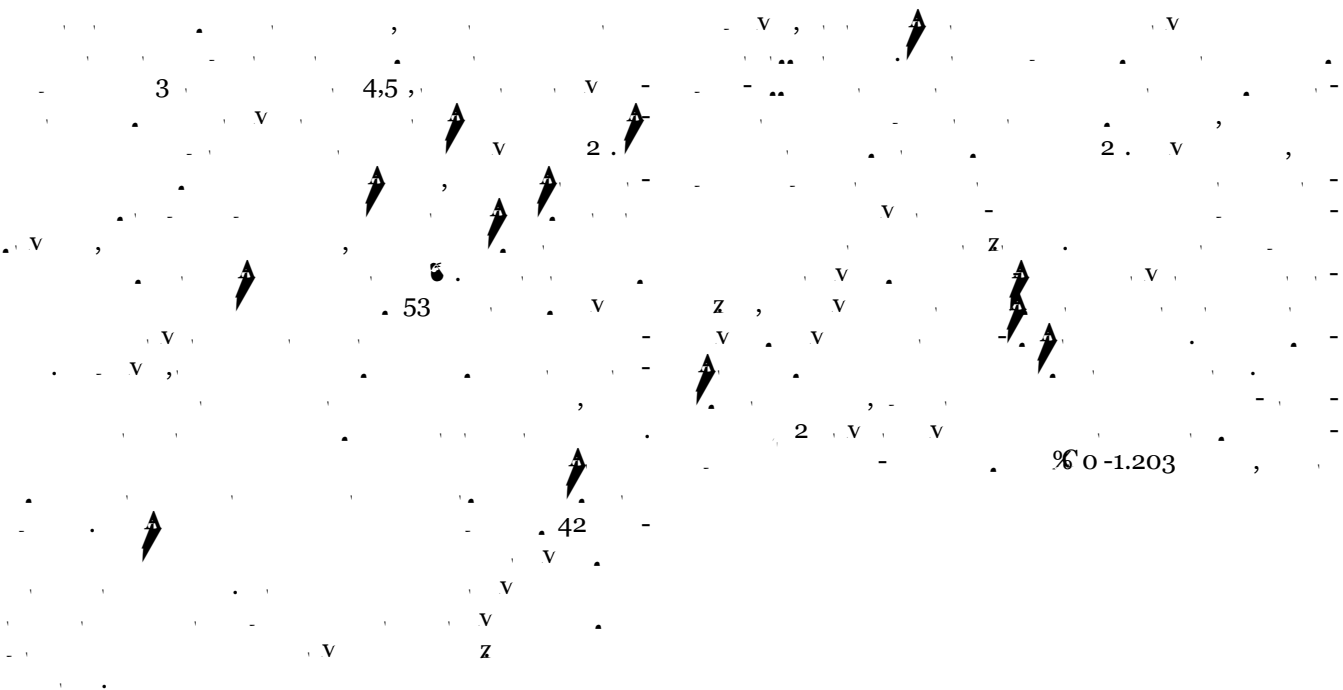
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## Abstract

**Background:** The phylogeographic distribution of human mitochondrial DNA variations allows a genetic approach to the study of modern *Homo sapiens* dispersals throughout the world from a female perspective. As a new contribution to this study we have phylogenetically analysed complete mitochondrial DNA(mtDNA) sequences from 42 human lineages, representing major clades with known geographic assignation.

**Results:** We show the relative relationships among the 42 lineages and present more accurate temporal calibrations than have been previously possible to give new perspectives as how modern





**Results and Discussion**

42

(.1) 150,

152,303 155

124 v (1,122 (15,44

35 (2 %)

45. 40 (11%)

Table 1: HVS I motifs

Sample	HVS I motif	Haplogroup	Origin	Ref. <sup>a</sup>
K	145 224 311	K	Iberian	1
U7	248 318T	U7	Iberian	1
U3 <sub>1</sub>	343 356 390	U3	Canarian	1
U3 <sub>2</sub>	343 390	U3	Moroccan	1
U2 <sub>1</sub>	051 092 129C 189 362 368	U2	Jordanian	1
U2 <sub>2</sub>	051 129C 189 319 362	U2	Iberian	1
U2	051 189 234 294	U2	Jordanian	1
U5b	189 192 270	U5b	Berber	1
U5a	093 153 256 270 311 399	U5a1a	Swede	2
U6	172 219	U6	Moroccan	1
H <sub>1</sub>		H	Mauritanian	1
H <sub>F</sub>	093 183d 189	H		3
RCRS		H	European	4
H <sub>2</sub>		H	Iberian	1
V	298	V	Berber	1
HV	278 311	HV	Jordanian	1
T5	126 153 189 294	T5	Moroccan	1
T1	126 163 186 189 294	T1	Iberian	1
J1b	069 126 145 222 261	J1b	Moroccan	1
J2	069 126 193 300	J2	Iberian	1
B	136 183C 189 217 284	B	Japanese	5
I	129 148 223 391	I	Iberian	1
I <sub>F</sub>				



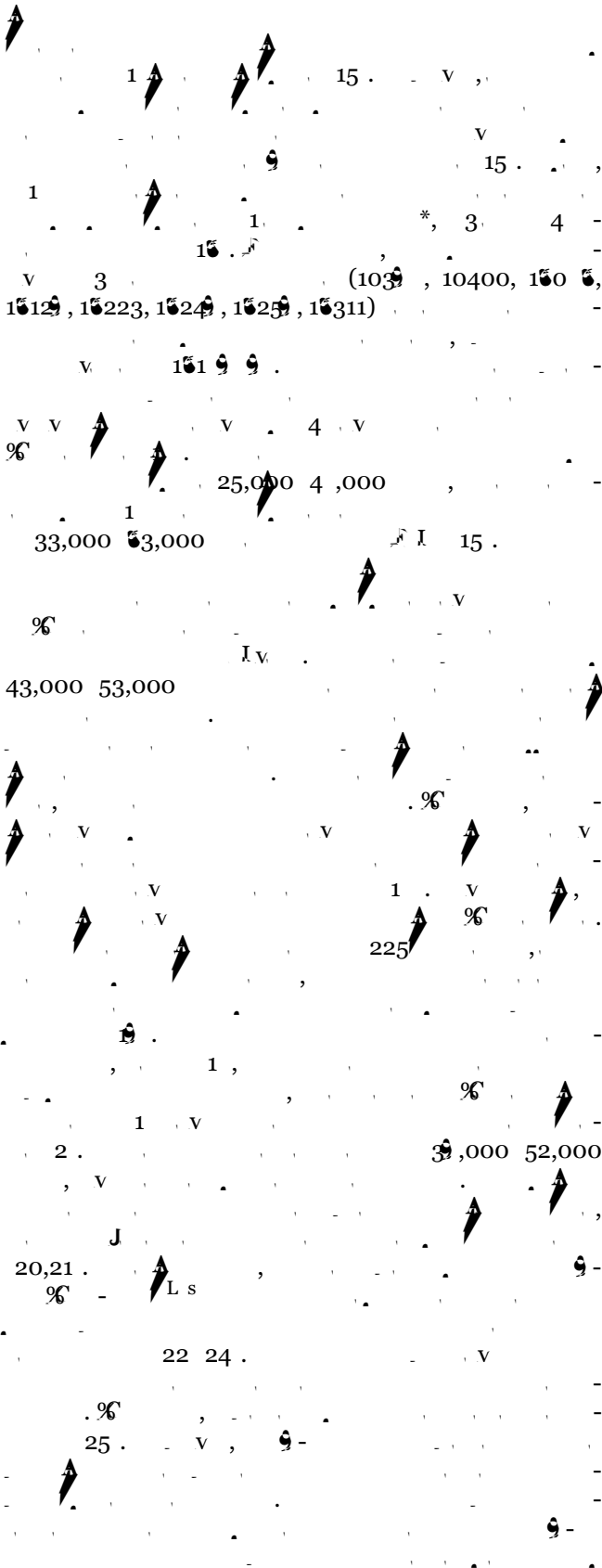


Table 2: Oligonucleotide pairs used in the amplification and sequencing

Name	CRS reference	Sequence (5'-3')	Fragment size (pb)	Annealing temp.(°C)
L16340	(16318-16340)	AGCCATTTACCGTACATAGCACA	681	52
H408	(429-408)	TGTTAAAAGTGCATACCGCCA		
L382	(362-382)	CAAAGAACCCTAACACCAGCC	603	56
H945	(964-945)	GGGAGGGGGTGATCTAAAC		
L923	(902-923)	GTCACACGATTAACCCAAGTCA	607	56
H1487	(1508-1487)	GTATACTTGAGGAGGGTGACGG		
L1466	(1445-1466)	GAGTGCTTAGTTGAACAGGGCC	629	58
H2053	(2073-2053)	TTAGAGGGTTCTGTGGGCAA		
L2025	(2004-2025)	GCCTGGTGATAGCTGGTTGTCC	609	52
H2591	(2612-2591)	GGAACAAGTGATTATGCTACCT		
L2559	(2538-2559)	CACCGCCTGCCAGTGACACAT	591	56
H3108	(3128-3108)	TCGTACAGGGAGGAATTTGAA		
L3073	(3051-3073)	AAAGTCTACGTGATCTGAGTTC	640	52
H3670	(3690-3670)	GGCGTAGTTTGAGTTTGATGC		
L3644	(3625-3644)	GCCACCTCTAGCCTAGCCGT	623	58
H4227	(4247-4227)	ATGCTGGAGATTGTAATGGGT		
L4210	(4189-4210)	CCACTCACCTAGCATTACTTA	625	55
H4792	(4813-4792)	ACTCAGAAGTGAAAGGGGGCTA		
L4750	(4729-4750)	CCAATACTACCAATCAATACTC	599	52
H5306	(5327-5306)	GGTGATGGTGGCTATGATGGTG		
L5278	(5259-5278)	TGGGCCATTATCGAAGAATT	593	58
H5832	(5851-5832)	GACAGGGGTTAGGCCTCTTT		
L5781	(5762-5781)	AGCCCCGGCAGGTTTGAAGC	626	58
H6367	(6387-6367)	TGGCCCCTAAGATAGAGGAGA		
L6337	(6318-6337)	CCTGGAGCCTCCGTAGACCT	601	58
H6899	(6918-6899)	GCACTGCAGCAGATCATTTC		
L6869	(6850-6869)	CCGGCGTCAAAGTATTTAGC	578	58
H7406	(7427-7406)	GGGTTCTTCGAATGTGTGGTAG		
L7379	(7358-7379)	AGAAGAACCCTCCATAAACCTG	580	56
H7918	(7937-7918)	AGATTAGTCCGCCGTAGTCG		
L7882	(7861-7882)	TCCCTCCCTTACCATCAAATCA	506	56
H8345	(8366-8345)	TTTCACTGTAAAGAGGGTTGG		
L8299	(8280-8299)	ACCCCCTCTAGAGCCACTG	603	56
H8861	(8882-8861)	GAGCGAAAGCCTATAATCACTG		
L8799	(8779-8799)	CTCGGACTCCTGCCTCACTCA	638	58
H9397	(9416-9397)	GTGGCCTTGGTATGTGCTTT		
L9362	(9342-9362)	GGCCTACTAACCAACACACTA	609	56
H9928	(9950-9928)	AACCACATCTACAAAATGCCAGT		
L9886	(9865-9886)	TCCGCCAACTAATATTTCACTT	617	56
H10462	(10481-10462)	AATGAGGGGCATTTGGTAAA		
L10403	(10383-10403)	AAAGGATTAGACTGAACCGAA	612	56
H10975	(10994-10975)	CCATGATTGTGAGGGGTAGG		
L10949	(10930-10949)	CTCCGACCCCTAACCAACCC	617	58
H11527	(11546-11527)	CAAGGAAGGGGTAGGCTATG		
L11486	(11467-11486)	AAAAC TAGGCGGTATGGTA	629	56
H12076	(12095-12076)	GGAGAATGGGGATAGGTGT		
L12028	(12008-12028)	GGCTCACTCACCCACACATT	615	58
H12603	(12623-12603)	ACGAACAATGCTACAGGGATG		
L12572	(12553-12572)	ACAACCCAGCTCTCCCTAAG	591	56
H13124	(13143-13124)	ATTTTCTGCTAGGGGGTGG		
L13088	(13068-13088)	AGCCCTACTCCACTCAAGCAC	618	58
H13666	(13685-13666)	AGGGTGGGGTTATTTTCGTT		
L13612	(13593-13612)	AAGCGCCTATAGCACTCGAA	614	56
H14186	(14206-14186)	TGGTTGAACATTGTTTGG		
L13612	(13593-13612)	AAGCGCCTATAGCACTCGAA	614	56
H14186	(14206-14186)	TGGTTGAACATTGTTTGG		
L14125	(14104-14125)	TCTTTCTTCTCCACTCATCC	602	58

### Conclusions

The present study has shown that the mitochondrial DNA (mtDNA) haplotypes of the studied populations are highly diverse. The haplotypes were found to be distributed in a wide geographic area, including Europe, Africa, and Asia. The results of the present study are in agreement with previous studies that have shown a high degree of genetic diversity in the mtDNA of human populations. The present study also shows that the mtDNA haplotypes of the studied populations are highly similar to those of other human populations. This suggests that the mtDNA haplotypes of the studied populations are of African origin. The present study also shows that the mtDNA haplotypes of the studied populations are highly similar to those of other human populations. This suggests that the mtDNA haplotypes of the studied populations are of African origin.

### Materials and Methods

#### Lineages

The mtDNA haplotypes of the studied populations were determined by sequencing the D-loop region of the mtDNA. The sequences were then compared to those of other human populations using the HaploTree software. The results of the present study are shown in Figure 1. The present study also shows that the mtDNA haplotypes of the studied populations are highly similar to those of other human populations. This suggests that the mtDNA haplotypes of the studied populations are of African origin.

#### Complete mtDNA sequences

The complete mtDNA sequences of the studied populations were determined by sequencing the entire mtDNA. The sequences were then compared to those of other human populations using the HaploTree software. The results of the present study are shown in Figure 2. The present study also shows that the mtDNA haplotypes of the studied populations are highly similar to those of other human populations. This suggests that the mtDNA haplotypes of the studied populations are of African origin.

#### Statistic analyses

The genetic diversity of the mtDNA haplotypes of the studied populations was determined using the Haplotype Diversity (Hd) and Nucleotide Diversity (π) indices. The results of the present study are shown in Table 1. The present study also shows that the mtDNA haplotypes of the studied populations are highly similar to those of other human populations. This suggests that the mtDNA haplotypes of the studied populations are of African origin.

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3.5 // v  
95% V  
1. × 10<sup>-8</sup>  
53

#### Accession numbers

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