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A concordance of nucleotide substitutions in the first and second hypervariable segments of the human mtDNA control region

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Abstract A new and easily accessible concordance of nucleotide substitutions in the hypervariable segments of the human mitochondrial DNA (mtDNA) control region has been constructed. The concordance indexes all population-specific mtDNA sequences in a standardized format. The first edition of the concordance includes 1,440 sequences representing 762 mtDNA types from over 65 populations for hypervariable region 1, and 520 sequences representing 260 mtDNA types from over 26 populations for hypervariable region 2. Investigators are invited to submit new sequences to the database, and details for doing so are given in the text.

Key words Human populations · Mitochondrial DNA · DNA sequencing · Population genetics · Anthropology

Introduction

The analysis of the hypervariable control region of human mitochondrial DNA (mtDNA) has proved to be a useful tool for forensic scientists [1], population and evolutionary biologists [2, 3], and anthropologists [4]. In each case, there is great interest in the degree of similarity between sequences obtained from different groups of individuals. This interest is particularly acute with the identification of unknown individuals, where a series of mitochondrial substitutions is often compared with groups of sequences from characterized populations that are, in some instances, marked by particular substitution motifs [5]. However, the number of individuals screened in recent years has greatly expanded and it has become increasingly difficult to access sequence data quickly and easily.

We have compiled a population-specific database of peer-reviewed mtDNA sequences and created a concordance from that database. A concordance is defined as "an alphabetical arrangement of principal words contained in a book, with citations of the passages in which they occur" [6]. We suggest the use of this term to identify a cross-referenced list of single nucleotide substitutions in the two hypervariable segments of the mtDNA control region. Concordances have been used for many years to process non-scientific information, and computers have been employed to create concordances since the late 1950s [7, 8].

The resulting reference list of base substitutions is in a standardized format arranged by site, and includes all the population-specific mtDNA types which share each individual substitution. In addition to the obvious advantages of such a reference list for the identification of mtDNA inclusions and exclusions in a given population, the data assembled are referenced to the publication in which they originally appeared. The concordance will be helpful in identifying population-specific variable sites, as well as potentially less informative sites, like those where parallel mutations occur. The concordance will be updated at regular intervals and is provided free of charge to interested researchers.

Materials and methods

A survey of sequences from the published literature, and the sequence databases EMBL and GenBank was conducted. Sequences attributable to specific populations and with at least 90% of a hypervariable region (or 50% for sequences from degraded biological samples, such as archaeological or forensic samples) were included in the survey. The sequences were concorded using the Oxford Concordance Program [9], and the results transformed into a Microsoft Excel [10] table. For each nucleotide substitution observed, the number of individuals of a given population with the particular substitution and the population frequency were computed and arranged in descending order of frequency. MtDNA types were organized in numerical order and the number of individuals sharing those types was recorded (the total number of indi-

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Table 1 Nucleotide substitutions in the human mtDNA hypervariable region 1 [bases 16,024 to 16,324] included in the concordance

Region/ Country	Population	Identification code	Number of individuals	Number of mtDNA types	Hypervariable region 1 positons used	Reference
America, Central	Boruca	CA1	2	2	16,039–16,324	[15]
	Guaymi	CA2	1	1	16,039–16,324	[15]
	Maya	CA3	4	4	16,039–16,324	[15]
America, North	Apache	NA1	1	1	16,039–16,324	[15]
	Bella Coola	NA2	4	4	16,039–16,324	[15]
	Dogrib	NA3	2	2	16,039–16,324	[15]
	Haida	NA4	1	1	16,039–16,324	[15]
	Navajo	NA5	2	2	16,039–16,324	[15]
	Nuu-Chah-Nulth	NA8	63	25	16,024–16,324	[16]
	Ojibwa	NA6	3	3	16,039–16,324	[15]
	Pima	NA7	3	3	16,039–16,324	[15]
	Windover	WIN	14	11	16,151–16,317	[17]
	Yakima	NA9	41	16	16,024–16,324	[18]
America, South	Kraho	SA1	3	3	16,039–16,324	[15]
	Makiritare	SA2	1	1	16,039–16,324	[15]
	Mataco	SA3	3	3	16,039–16,324	[15]
	Ticuna	SA4	3	3	16,039–16,324	[15]
	Yanomama	SA5	3	3	16,039–16,324	[15]
Argentina	Mapuche	SA6	39	13	16,050–16,324	[19]
Australia	Aborigine	AUS	1	1	16,025–16,324	[14]
Botswana	Herero	HRO	28	7	16,025–16,324	[14]
	!Kung	VRK, VSA, VZH	26	15	16,025–16,324	[14]
Central African Republic	Biaka Pygmy	VBP	17	7	16,025–16,324	[14]
China	H'mong	VDH	1	1	16,025–16,324	[14]
	Southern	VCH	10	10	16,025–16,324	[14]
	Chinese	VAB, VHD, VUC	5	5	16,025–16,324	[14]
	Chinese	CCH	2	2	16,048–16,324	[20]
Colombia	Colombian	CMM	6	3	16,024–16,324	[34]
Costa Rica	Huetar	CRH	27	7	16,024–16,324	[21]
Estonia	Estonian	EST	28	23	16,024–16,324	[22]
Finland	Finnish	FIN	50	33	16,024–16,324	[22]
	Karelian	KAR	83	41	16,024–16,324	[22]
	Saami	SAA	114	24	16,024–16,324	[22]
Guatemala	[Quiche?]	GUA	30	16	16,024–16,324	[23]
Iceland	Icelandic	ICS	39	26	16,024–16,324	[22]
India	Havik	INH	48	37	16,048–16,324	[24]
	Kadar	INA	7	2	16,048–16,324	[24]
	Mukri	INM	43	13	16,048–16,324	[24]
Indonesia	Hiri, Ternate, Alor, Flores, Roti, and Timor	IND	27	20	16,025–16,324	[25]
	Indonesian	VIN	1	1	16,025–16,324	[14]
Italy	Sardinian	SAR	69	46	16,043–16,324	[26]
Ivory Coast	African	ICH	1	1	16,048–16,324	[20]
Japan	Japanese	JPO	18	18	16,025–16,324	[27]
	Japanese	JPH	11	8	16,048–16,324	[20]
	Japanese	VJP	1	1	16,025–16,324	[14]

Table 1 (continued)

Region/ Country	Population	Identification code	Number of individuals	Number of mtDNA types	Hypervariable region 1 positons used	Reference
Korea	Korean	KOR	4	4	16,039–16,324	[15]
	Korean	KOH	1	1	16,048–16,324	[20]
Middle East	Israeli Arab, Saudi Arabian Bedouin, Yemenite Jew	DME	42	37	16,043–16,324	[26]
Nigeria	Yoruban	VNY	14	12	16,025–16,324	[14]
Panama	Kuna	PAK	63	5	16,024–16,324	[28]
	Ngöbé	KNG	46	6	16,024–16,324	[29]
Papua New Guinea	Aua, Bam, East Sepik and Milne Bay Provinces, Manus, Wokeo	VWE	6	3	16,025–16,324	[14]
	Central Province	VCP	2	2	16,025–16,324	[14]
	East New Britain	ENB	2	2	16,025–16,324	[14]
	Eastern Highlands	VEH, VNG	2	2	16,025–16,324	[14]
	Gulf Province	VGP	2	2	16,025–16,324	[14]
	Northern Province	VNP	2	2	16,025–16,324	[14]
	Southern Highlands	VSH	4	4	16,025–16,324	[14]
Philippines	Filipino	VKP	1	1	16,025–16,324	[14]
	Filipino	PHH	1	1	16,048–16,324	[20]
Polynesia	Tongan	VSP	1	1	16,025–16,324	[14]
Russia	Tsar Nicholas, Tsarina Alexandra, Dr. Botkin, Servants 1–3	ROM	6	6	16,025–16,324	[1]
	Volga-Finnic	VOL	33	22	16,024–16,324	[22]
Siberia and the Russian Far East	Altai	SI4	17	12	16,024–16,324	[18]
	Evenk	SI1	11	10	16,039–16,324	[30]
	Nivkh	SI2	2	2	16,039–16,324	[30]
	Udegey	SI3	3	2	16,039–16,324	[30]
Samoa	Samoan	SAM	24	5	16,025–16,324	[25]
Spain	Basque	BAS	45	26	16,024–16,324	[31]
Switzerland	Swiss	SWI	74	41	16,024–16,324	[32]
Taiwan	Han	HAN	6	6	16,039–16,324	[15]
	Taiwanese	VTA	1	1	16,025–16,324	[14]
Tanzania	Hazda	HDZ	17	4	16,025–16,324	[14]
Tyrolean Alps	"Ice Man"	TIM	1	1	16,056–16,324	[33]
United Kingdom	British	RC1	100	62	16,025–16,324	[13]
Zaïre	Zaïrese	ZAH	1	1	16,048–16,324	[20]
	Mbuti Pygmy	VPG	20	11	16,025–16,324	[14]
Totals ≥ 65			1,440	762		

Table 2 Nucleotide substitutions in the human mtDNA hypervariable region 2 [base 63 to 322] included in the concordance

Region/Country	Population	Identification code	Number individuals types	Number of of mtDNA	Hypervariable region 2 positions used	Reference
Argentina	Mapuche	SA6	39	18	63–322	[19]
Australia	Aborigine	AUS	1	1	63–322	[14]
Botswana	Herero	HRO	28	5	63–322	[14]
	!Kung	VRK, VSA, VZH	26	13	63–322	[14]
Central African Republic	Biaka Pygmy	VBP	17	14	63–322	[14]
China	Southern	VCH	10	10	63–322	[14]
	Chinese	VAB, VHD, VUC	5	5	63–322	[14]
Costa Rica	Huetar	CRH	27	7	63–322	[21]
India	Havik	INH	48	26	63–322	[24]
	Kadar	INA	7	2	63–322	[24]
	Mukri	INM	43	9	63–322	[24]
Indonesia	Hiri, Ternate, Alor, Flores, Roti, and Timor	IND	27	12	63–322	[25]
	Indonesian	VIN	1	1	63–322	[14]
Japan	Japanese	JPO	18	16	63–322	[27]
	Japanese	VJP	1	1	63–322	[14]
Nigeria	Yoruban	VNY	14	12	63–322	[14]
Panama	Kuna	PAK	3	3	63–322	[28]
	Ngöbé	KNG	15	10	63–322	[29]
Papua New Guinea	Aua, Bam, East Sepik and Milne Bay Provinces, Manus, Wokeo	VWE	6	2	63–322	[14]
	Central Province	VCP	2	2	63–322	[14]
	East New Britain	ENB	2	2	63–322	[14]
	Eastern Highlands	VEH, VNG	2	2	63–322	[14]
	Gulf Provice	VGP	2	2	63–322	[14]
	Northern Province	VNP	2	2	63–322	[14]
	Southern Highlands	VSH	4	4	63–322	[14]
Philippines	Filipino	VKP	1	1	63–322	[14]
Polynesia	Tongan	VSP	1	1	63–322	[14]
Russia	Tsar Nicholas, Tsarina Alexandra, Dr. Botkin, Servants 1–3	ROM	6	5	63–322	[1]
Samoa	Samoan	SAM	24	3	63–322	[25]
Tanzania	Hazsa	HDZ	17	4	63–322	[14]
Taiwan	Taiwanese	VTA	1	1	63–322	[14]
United Kingdom	British	RC1	100	52	63–322	[13]
Zaïre	Mbuti Pygmy	VPG	20	12	63–322	[14]
Totals ≥ 26			520	260		

Table 3 Excerpt from the concordance, showing data for mtDNA hypervariable region 2.

93[A]	Papua New Guinean [1:0.05], British [1:0.01]	
	72[C] 93[A] 195[C] 263[G] 309.1[C] 315.1[C]	RC1[1]
	74[G] 78[C] 93[A] 147[C] 264[G] 315.1[C]	VSH[1]
94[G]	Biaka Pygmy [7:0.41], Mbuti Pygmy [6:0.30], Hazda [5:0.29], Papua New Guinean [1:0.05], Japanese [1:0.05], Tongan [1]	
	64[T] 94[G] 133[G] 237[C] 248[A] 264[G] 309.1[C] 309.2[C] 315.1[C]	VPG[1]
	64[T] 94[G] 186[A] 190[G] 201[G] 237[C] 248[A] 264[G] 315.1[C]	VSP[1]
	64[T] 94[G] 237[C] 248[A] 264[G] 309.1[C] 309.2[C] 315.1[C]	VPG[3]
	64[T] 94[G] 237[C] 248[A] 264[G] 309.1[C] 315.1[C]	VPG[2]
	74[G] 94[G] 213[C] 264[G] 315.1[C]	VWE[1]
	74[G] 94[G] 96[C] 147[C] 247[C] 264[G] 315.1[C] [N]	HDZ[5]
	74[G] 94[G] 96[C] 153[C] 183[T] 187[A] 190[C] 237[C] 248[A] 264[G] 299[G] 315.1[C] 316[A]	VBP[1]
	74[G] 94[G] 96[C] 183[T] 187[A] 190[C] 197[C] 237[C] 248[A] 264[G] 299[G] 309.1[C] 315.1[C] 316[A]	VBP[2]
	94[G] 151[T] 264[G] 309.1[C] 315.1[C]	VJP[1]
	94[G] 153[C] 190[G] 237[C] 248[A] 264[G] 309.1[C] 315.1[C]	VBP[3]
	94[G] 153[C] 190[G] 237[C] 248[A] 309.1[C] 315.1[C] 316[A]	VBP[1]
96[C]	Hazda [5:0.29], Biaka Pygmy [4:0.24]	
	74[G] 94[G] 96[C] 147[C] 247[C] 264[G] 315.1[C] [N]	HDZ[5]
	74[G] 94[G] 96[C] 153[C] 183[T] 187[A] 190[C] 197[C] 237[C] 248[A] 264[G] 299[G] 315.1[C] 316[A]	VBP[1]
	74[G] 94[G] 96[C] 183[T] 187[A] 190[C] 197[C] 237[C] 248[A] 264[G] 299[G] 309.1[C] 315.1[C] 316[A]	VBP[2]
	74[G] 96[C] 183[T] 187[A] 190[C] 197[C] 237[C] 248[A] 264[G] 299[G] 315.1[C]	VBP[1]

viduals sharing each mtDNA type is not always specified in the original papers; in such instances, we have simply reported each mtDNA type as a single individual). Frequencies were not calculated for sample sizes less than ten. The following changes to published works were adopted in order to standardize the sequence information:

1. Hypervariable region 1 and 2 were treated as separate mtDNA types, to allow the calculation of frequencies.
2. The numbering system of the published mtDNA reference sequence [11] was adopted for all sequences.
3. All sequence data included were within the hypervariable regions defined by Vigilant et al. [12]. Sequence information outside the defined regions was not included.

Results and discussion

Tables 1 and 2 list the sequences for the mtDNA hypervariable regions 1 and 2 included in the first edition of the concordance. Each population was assigned a three letter population code. Codes originally ascribed by an investigator were retained unless they represented a duplication of a pre-existing code. Original codes prefixed by less than three letters were retained, generally with the addition of the author's initial. Also listed are the number of individuals and the number of mtDNA types in each population. These may deviate from the published data, depending on the length of sequence provided by the investigator and whether each mtDNA types was attributed unambiguously to a given individual or group of individuals in the original study. The length of sequences used to determine each mtDNA types is indicated in the concordance by the position of the first and last nucleotide of each DNA segment. Only those sequences which were population-specific and concentrated on hypervariable regions 1 and 2 were included in the concordance, but a wider set will eventually be covered.

A sample entry of the concordance to hypervariable region 2 is provided in Table 3. For sites where more than one substitution was observed, each substituted base is listed alphabetically. Below each substitution heading is a list of the known populations in which the substitution was observed. After each population name is a set of bracketed numbers. The first shows the number of individuals exhibiting the substitution, followed by a colon, followed by the frequency of the substitution in that population. Each substitution heading is followed by a list of the mtDNA types with that substitution. Beside each mtDNA type are the population codes with the number of individuals of each population [bracketed number] with that mtDNA type. In the example provided in Table 3, an A occurs at position 93 of hypervariable region 2. Substitutions other than A have not been reported. This substitution was observed in two populations, British [13] and Papua New Guineans [14]. The substitution was observed in two mtDNA types, one in each population, and each of these types was observed in only one individual. Thus, the single individual repre-

sents 1% in the sample from Britain, but 5% of the sample from Papua New Guinea. In the next entry, a G at position 94, the population frequency in Tongans was not calculated, because there were less than 10 individuals in the Tonga sample.

Compilation of the database brought to light many inconsistencies in the reporting of sequences. It may be noticed, for example, that the number of individuals reported in original publications is not always as described here. This is due to the fact that while all mtDNA types, or lineages, may have been reported in a publication, the total number of individuals sharing each mtDNA type is not always specified. In such instances, we have simply reported each mtDNA type as a single individual. This obviously underestimates the total number of individuals represented. In addition, the numbering system was standardized to that of the published human mtDNA reference sequence [11], and sequences reported with other numbering systems were altered to comply with this standard. Ambiguous and undetermined nucleotide positions in published sequences are provided under their mtDNA type to denote the possibility of additional substitutions associated with that type. In all cases the nomenclature of the International Union of Pure and Applied Chemistry (IUPAC) has been used.

The database is lodged at the University of Cambridge Computing Service and will be updated at regular intervals to include additional published sequence data. The concordance is as complete as possible given the database presently in the public domain, but we welcome comments from users and invite the submission of new peer-reviewed sequences, or sequences from bona fide forensic mtDNA data banks, for inclusion in future editions of the concordance. The latest version of the concordance will be available via electronic mail as a 'uuencoded' or 'binhexed' file by e-mailing to JLD1@cam.ac.uk. Plans are currently being made to provide anonymous FTP access to the concordance. MtDNA sequences for inclusion in the concordance should be submitted in a simple text format readable by Microsoft Word, like those automatically produced by automated DNA sequencers.

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