HITA / AABB Workshop
SNP ANALYSIS FOR IDENTIFICATION, ANCESTRY, GENEALOGY AND RELATIONSHIP ANALYSIS

Haploid Markers: mtDNA and Y-Chr

Michael Coble, PhD

National Institute of Standards and Technology

The 21st International Symposium on Human Identification
October 10, 2010

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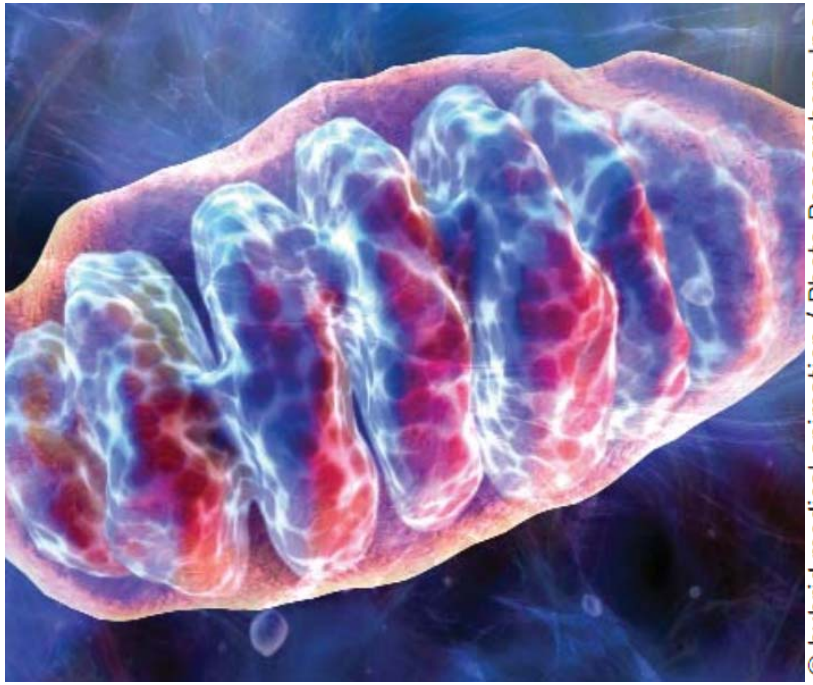
http://en.wikipedia.org/wiki/File:Paul_Gauguin_1891.png

Where Do We Come From? What Are We? Where Are We Going?

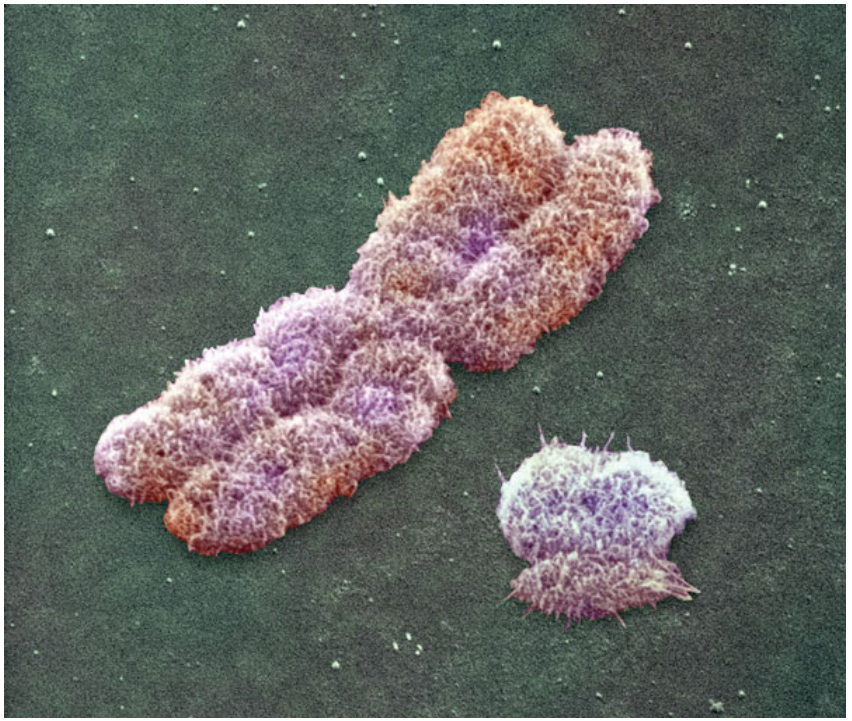
Paul Gauguin, 1897

http://en.wikipedia.org/wiki/File:Woher_kommen_wir_Wer_sind_wir_Wohin_gehen_wir.jpg

Where Do We Come From?



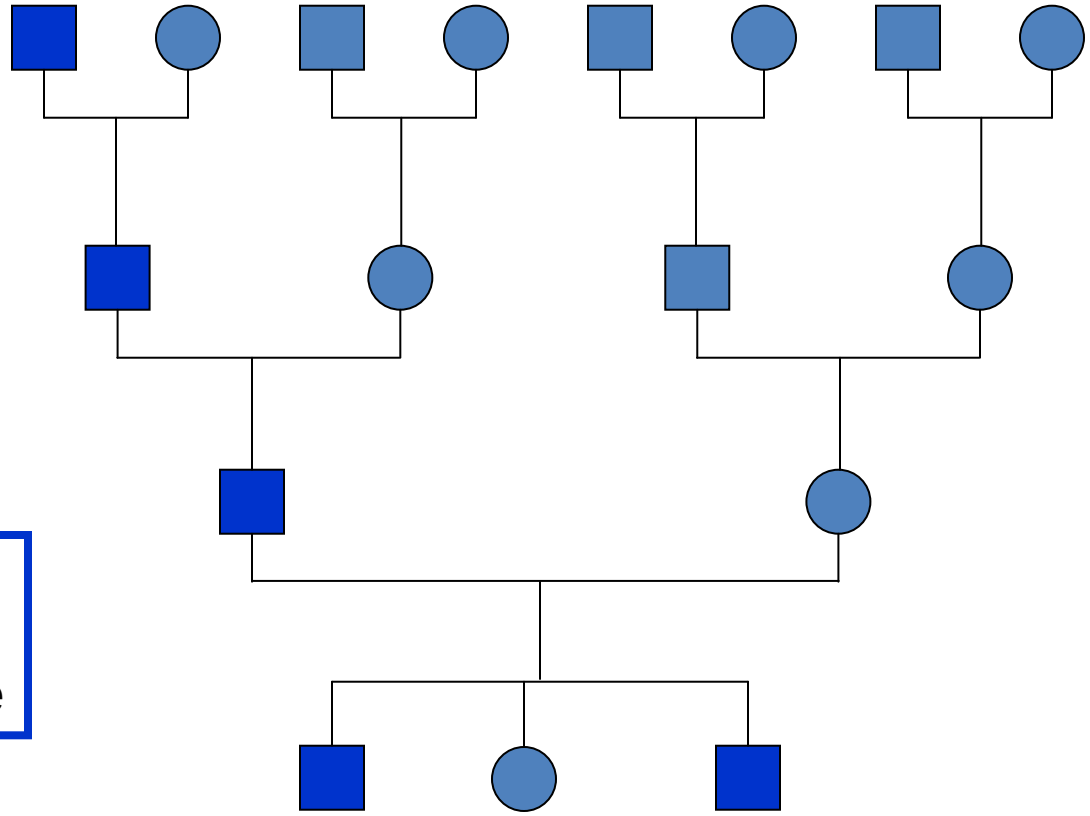
© hybrid medical animation / Photo Researchers, Inc.



http://www.nature.com/nature/journal/v423/n6942/fig_tab/423810a_F1.html

Haploid Markers – NO Recombination

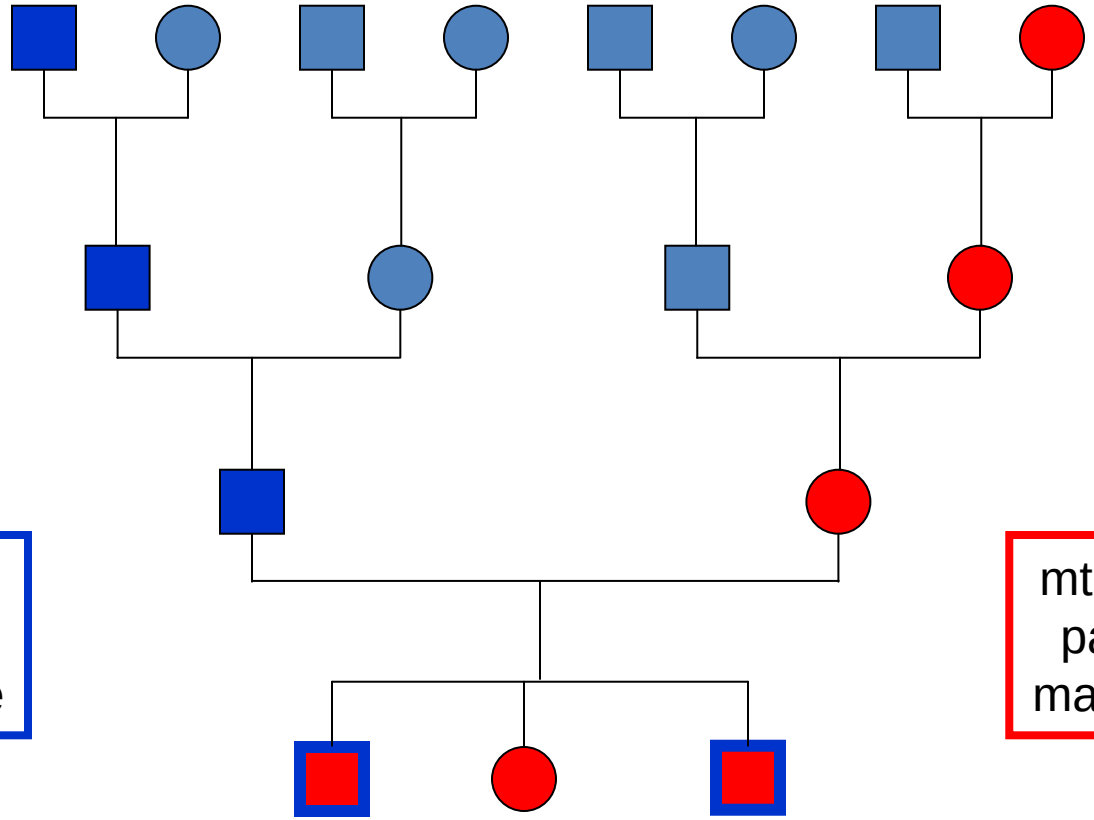
Haploid Markers



Y chromosome passed along paternal lineage

Autosomal DNA
1/8 from Great-grandparents

Haploid Markers



Y chromosome passed along paternal lineage

mtDNA genome passed along maternal lineage

Autosomal DNA
1/8 from Great-grandparents

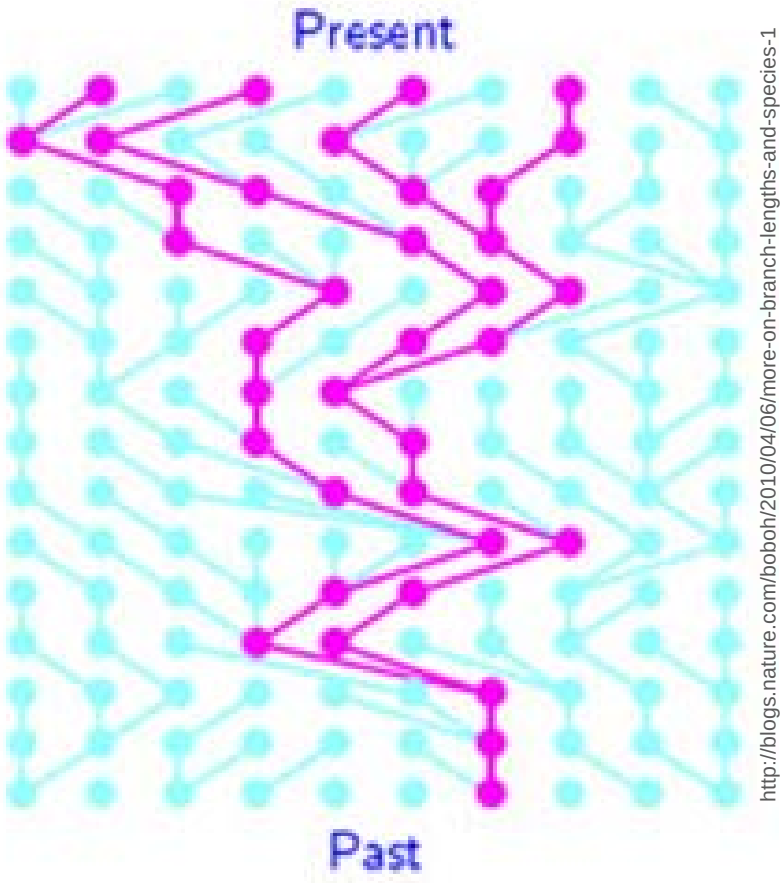
Some Nomenclature

- Haplotype – the mtDNA sequence variations within an individual (e.g. your HV1/HV2 type).
- Haplogroup – a group of related haplotypes. These form monophyletic clades on a phylogenetic tree.

Doug Wallace

Antonio Torroni

Coalescence Tree



http://blogs.nature.com/boboh/2010/04/06/more-on-branch-lengths-and-species-1

"Mitochondrial Eve"

"Y-Adam"

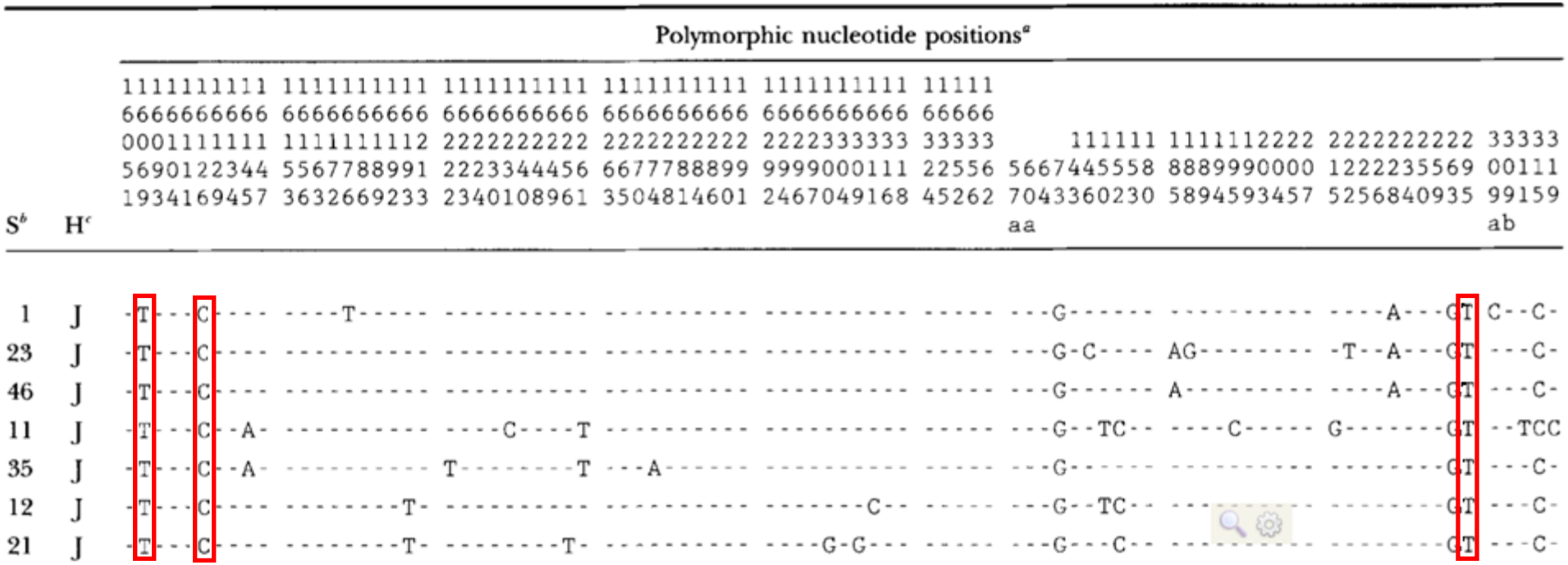
Classification of European mtDNAs From an Analysis of Three European Populations

Antonio Torroni,*,† Kirsi Huoponen,* Paolo Francalacci,‡ Maurizio Petrozzi,† Laura Morelli,‡ Rosaria Scozzari,†,§ Domenica Obinu,‡ Marja-Liisa Savontaus and Douglas C. Wallace***

**Department of Genetics and Molecular Medicine, Emory University School of Medicine, Atlanta, Georgia 30322, †Dipartimento di Genetica e Biologia Molecolare, Universita' di Roma "La Sapienza," 00185 Rome, Italy, ‡Istituto di Antropologia, Universita' di Sassari, 07100 Sassari, Italy, §Centro di Genetica Evoluzionistica, C.N.R., Rome, Italy and **Department of Medical Genetics, University of Turku, 20520 Turku, Finland*

Manuscript received March 12, 1996
Accepted for publication August 26, 1996

Genetics (1996)

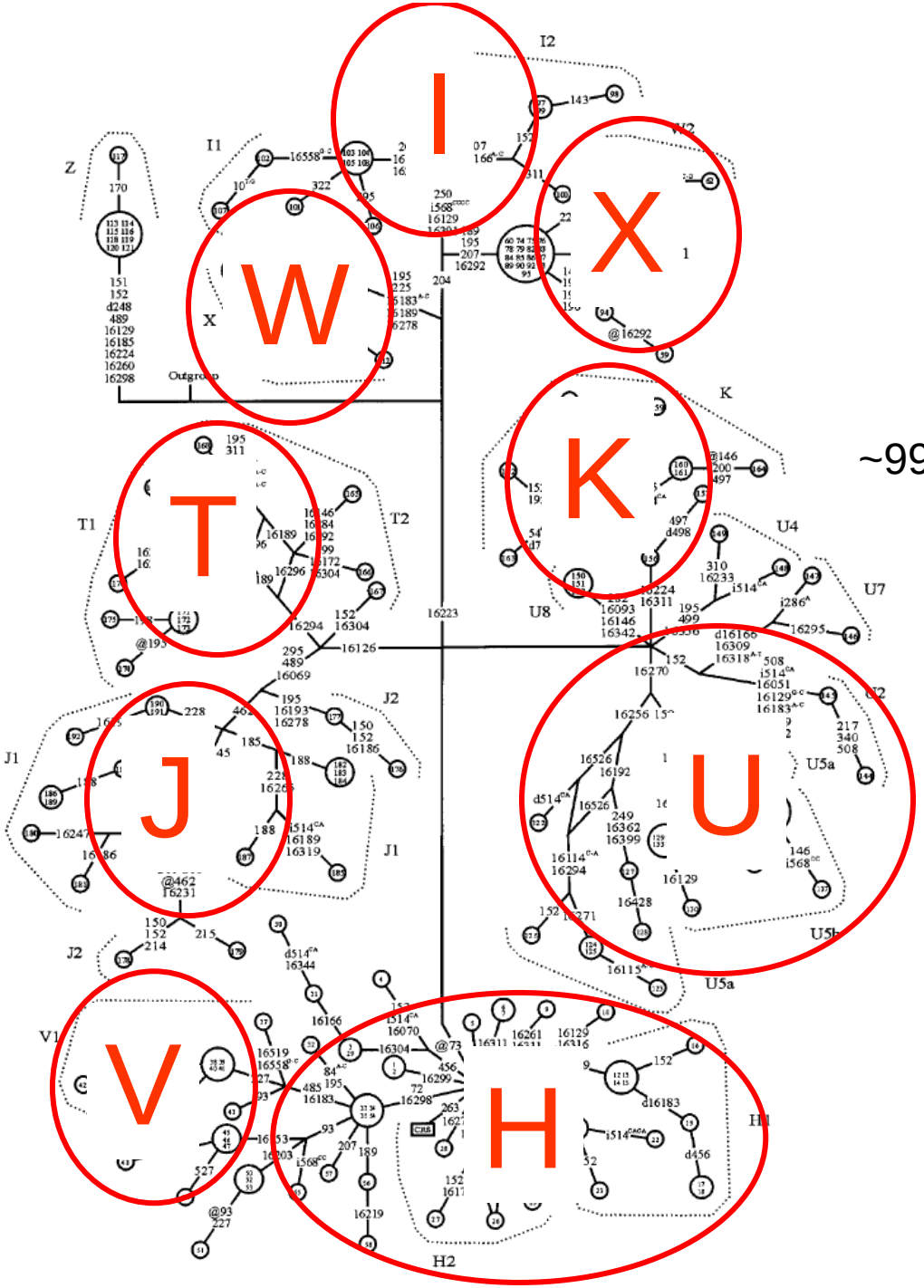


Polymorphic nucleotide positions[a]

S[b]	H[c]	1111111111 6666666666 0001111111 5690122344 1934169457	1111111111 6666666666 1111111112 5567788991 3632669233	1111111111 6666666666 2222222222 2223344456 2340108961	1111111111 6666666666 2222222222 6677788899 3504814601	1111111111 6666666666 2222333333 9999000111 2467049168	11111 66666 33333 22556 45262	111111 5667445558 7043360230 aa	1111112222 8889990000 5894593457	2222222222 1222235569 5256840935	33333 00111 99159 ab
1	J	-T---C---- ----T----- ---------- ---------- ---------- -----						---G------	----------	----A---GT	C--C-
23	J	-T---C---- ---------- ---------- ---------- ---------- -----						---G-C----	AG--------	-T--A---GT	---C-
46	J	-T---C---- ---------- ---------- ---------- ---------- -----						---G------	A---------	----A---GT	---C-
11	J	-T---C--A- ---------- ----C----T ---------- ---------- -----						---G--TC--	----C-----	G-------GT	--TCC
35	J	-T---C--A- ---------- T--------T ---A------ ---------- -----						---G------	----------	--------GT	---C-
12	J	-T---C---- --------T- ---------- ---------- -------C-- -----						---G--TC--	----------	--------GT	---C-
21	J	-T---C---- --------T- --------T- ---------- ----G-G--- -----						---G---C--	----------	--------GT	---C-

16069 C-T
16126 T-C
295 C-T

Torroni *et al.* ***Genetics*** (1996)

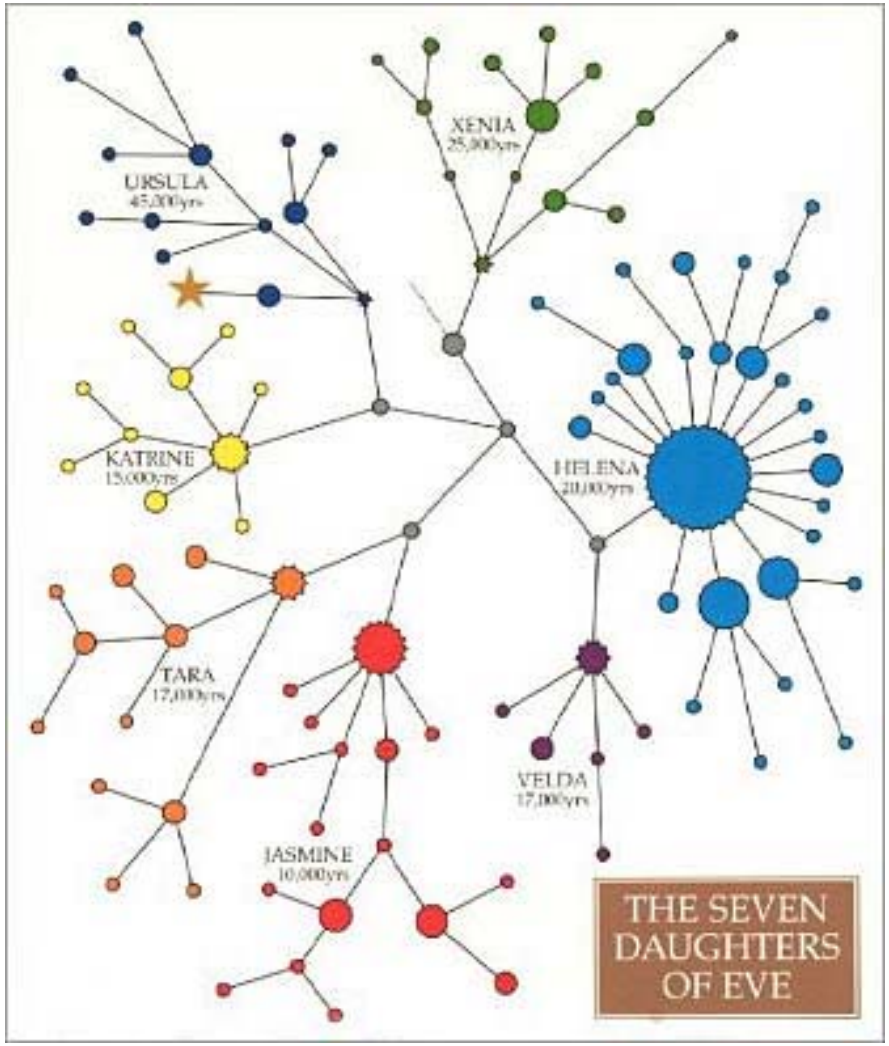


121 Finnish Samples – entire control region sequence data shown here (Finnila *et al.* 2001)

~99% of Caucasians can be classified into one of these 9 haplogroups

Finnila *et al.* (2001)

The 7 Daughters of Eve

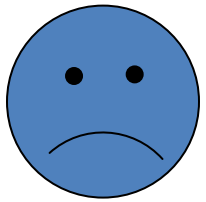


Haplogroup H = Helena
Haplogroup V = Velda
Haplogroup T = Tara
Haplogroup J = Jasmine
Haplogroup U = Ursula
Haplogroup K = Katrine
Haplogroup X = Xenia

(I and W – omitted)

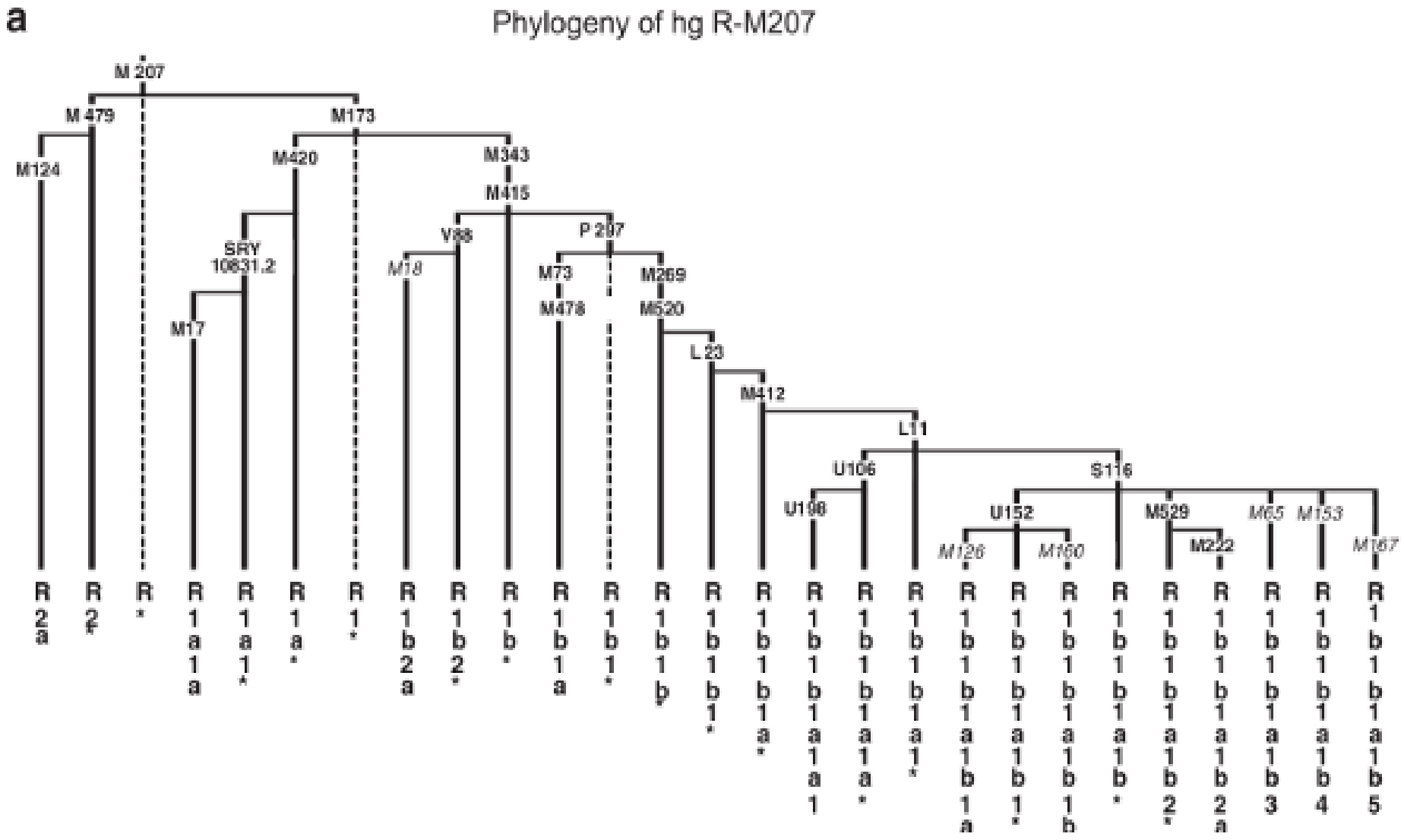
Me →

16189 T-C
16192 C-T
16270 C-T
73 A-G
150 C-T
263 A-G
315.1 C



No Clan Mother

Y-SNP Haplogroups



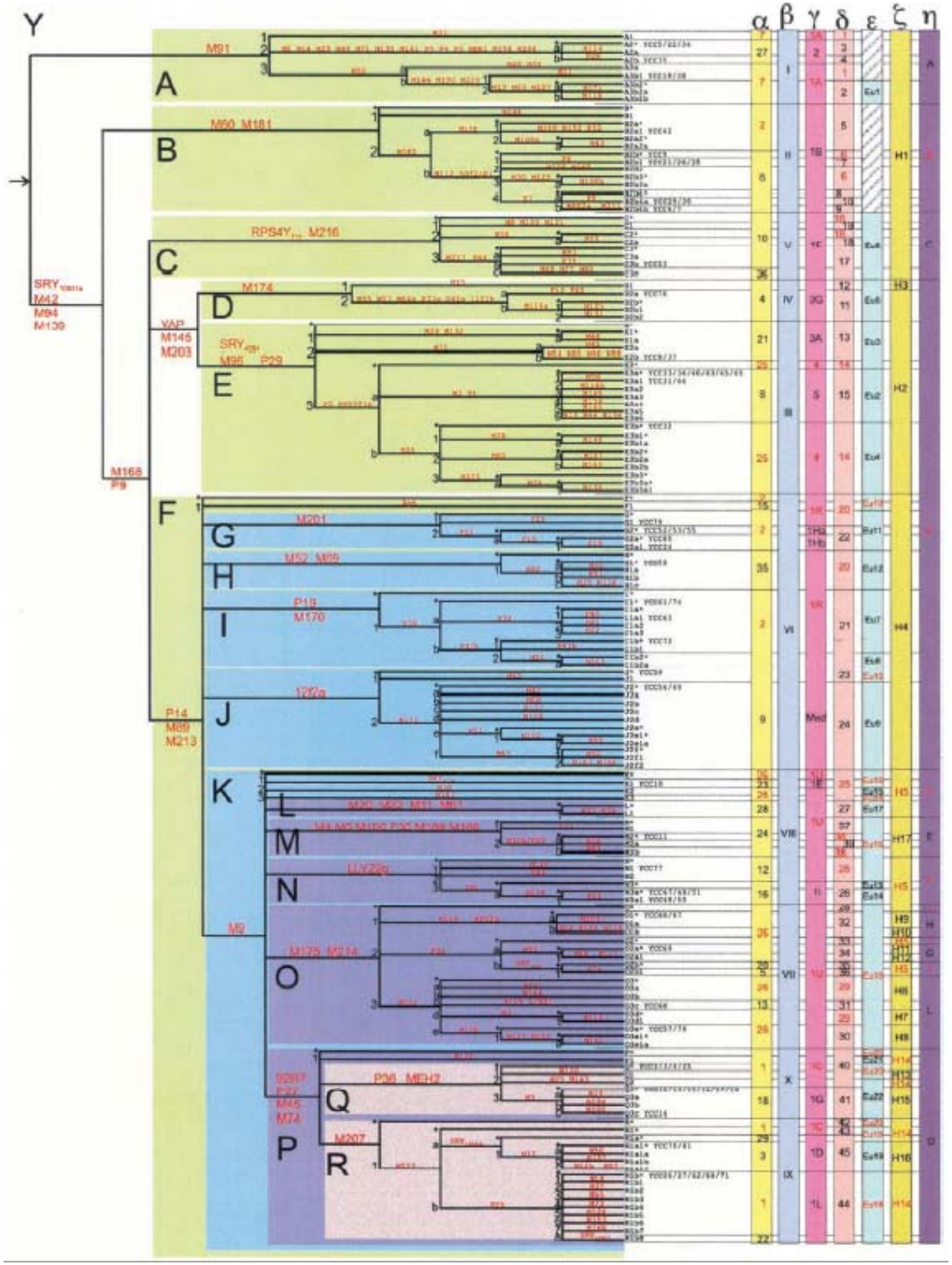
http://blogs.discovermagazine.com/gnxp/2010/09/a-tale-of-y-chromosomes-and-tea-leaves/

Resource

A Nomenclature System for the Tree of Human Y-Chromosomal Binary Haplogroups

The Y Chromosome Consortium[1]

Genome Research (2002) 12: 339-348.


Y
α
β
γ
δ
ε
ζ
η
A
B
C
D
E
F
G
H
I
J
K
L
M
N
O
P
Q
R
M91
M60 M181
RPS4Y711 M216
M174
SRY10831a
M42
M94
M139
YAP
M145
M203
SRY2814
M96 P29
M168
P9
M201
M52 M89
P19
M170
122a
P14
M69
M213
M9
M175 M214
LLY22g
P36 MEH2
92R7
P37
M45
M74
M207
H1
H2
H3
H4
H5
H6
H7
H8
H9
H10
H11
H12
H13
H14
H15
H16
H17

What Are We?
(Why are we here?)

MtDNA SNPs

- As a screening tool for sorting hair evidence or to eliminate multiple suspects.

Int J Legal Med (2006) 120: 372–376
DOI 10.1007/s00414-006-0085-y

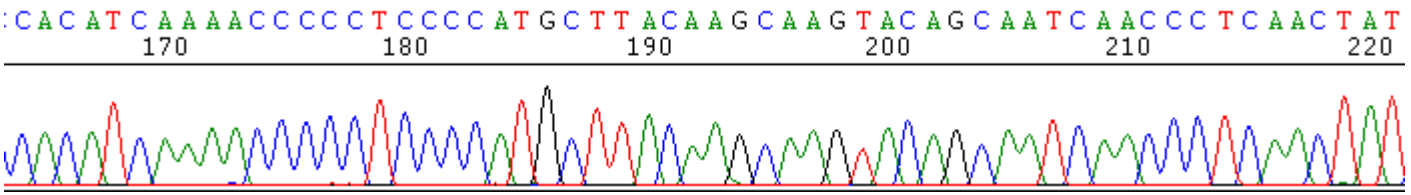
CASE REPORT

Reinhard Szibor · Ines Plate · Herrmann Schmitter ·
Holger Wittig · Dieter Krause

Forensic mass screening using mtDNA

Disadvantages to Sequencing

- Expensive
  - Primarily due to intensive labor in data analysis
- Error potential with more data to review
- Most information is not used



Review forward and reverse sequences across 610 bases only to report…

263G, 315.1C

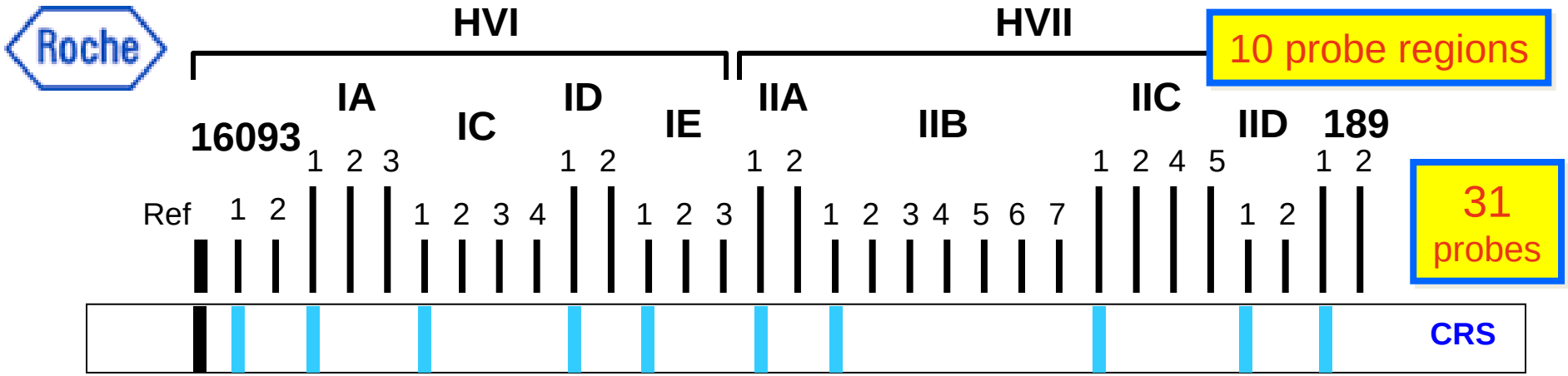
Most common type: found in ~7% of Caucasians…

Advantages to Screening Methods

- Rapid results
- Aids in exclusion of non-matching samples
- Less labor intensive
- Usually less expensive
- Permits more labs to get involved in mtDNA

Sequencing is necessary to certify that every position matches between a question and a known sample.

Screening assays are essentially a presumptive test prior to final confirmatory DNA sequencing.



16093

16093 1	A T **T** T C
16093 2	. . **C** . .

16126 16129

IA 1	T G **T** A C **G** G T
IA 2	. . **C** . . . . .
IA 3	. . . . . **A** . .

16304 16309 16311

IC 1	A G **T** A C A T **A** G **T** A C
IC 2	. . **C** . . . . .
IC 3	. . . . . **C** . .
IC 4	. . . . . **G** . . . .

16362

ID 1	C G **T** C C
ID 2	. . **C** . .

16270 16278

IE 1	C A **C** T A G G A T A **C** C A
IE 2	. . . . . **T** . .
IE 3	. . **T** . . . . .

73

IIA 1	G T **A** G T
IIA 2	. . **G** . .

18 SNPs

146 150 152

IIB 1	C C **T** C A T **C** C **T** A T
IIB 2	. . **C** . . . . .
IIB 3	. . . . . **C** . .
IIB 4	. . **C** . . . . **C** . .
IIB 5	. . . . . **T** . . . .
IIB 6	. . . . . **T** . **C** . .
IIB 7	. . **C** . . . **T** . **C** . .

189 195 198 200

IIC 1	G A **A** C A T A C **T** T A **C** T **A** A A
IIC 2	. . . . . **C** . . . . .
IIC 4	. . . . . **C** . . **T** . . . .
IIC 5	. . **G** . . . . . **G** . .

247

IID 1	T T **G** A A
IID 2	. . **A** . .

189

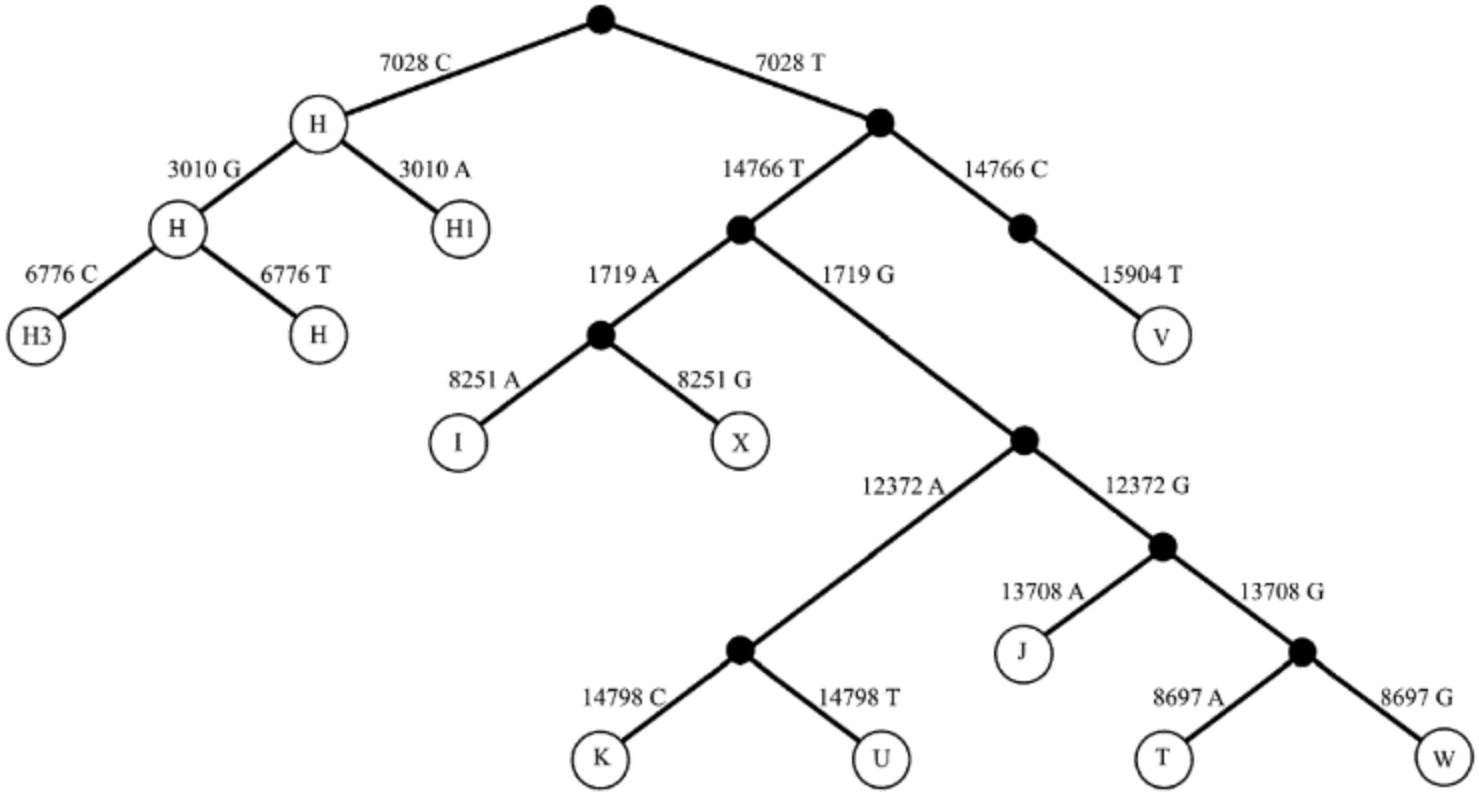
189 1	G A **A** C A
189 2	. . **G** . .

Int J Legal Med (2003) 117 : 291–298
DOI 10.1007/s00414-003-0395-2

ORIGINAL ARTICLE

Anita Brandstätter · Thomas J. Parsons · Walther Parson

Rapid screening of mtDNA coding region SNPs for the identification of west European Caucasian haplogroups



Available online at www.sciencedirect.com
ScienceDirect
Forensic Science International: Genetics 2 (2008) 61–68
FSI GENETICS
www.elsevier.com/locate/fsig

Identification of West Eurasian mitochondrial haplogroups by mtDNA SNP screening: Results of the 2006–2007 EDNAP collaborative exercise

Walther Parson [a,*], Liane Fendt [a], David Ballard [b], Claus Børsting [c], Bernd Brinkmann [d], Ángel Carracedo [e], Mónica Carvalho [f], Michael D. Coble [g], Francisco Corte Real [f], Stijn Desmyter [h], Berit M. Dupuy [i], Cheryl Harrison [b], Carsten Hohoff [d], Rebecca Just [g], Tanja Krämer [j], Niels Morling [c], Antonio Salas [e], Hermann Schmitter [k], Peter M. Schneider [j], Marie-Luise Sonntag [k], Peter M. Vallone [l], Anita Brandstätter [a]

Table 3
Detailed summary of the obtained results

	Laboratory											
	01	02	03	04	05	06	07	08	09	10	11	12
Electrophoresis	Ok	Ok	Ok	Profiles low	Peak imbalances	Ok	Electrophoresis problems reported	Ok	Ok	Profiles low	No raw data	Ok
Buccal Swab												
01	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	One marker ambiguous	Ok
02	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Typing correct; Clerical error
03	Ok	Ok	Ok	Ok	Two markers drop out; macrobranch identified	Ok	Ok	Ok	Ok	Two markers wrong basecall annotation	Three markers ambiguous	Ok
04	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	One marker ambiguous	Ok
05	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Ok	Two markers wrong basecall annotation	One marker ambiguous	Ok
Hair shaft												
01	Ok	Ok	Ok	Ok	Seven markers drop out; macrobranch identified	Ok	One marker drop out	Ok	Ok	10 markers drop out; no haplogroup identified	One marker ambiguous	Ok
02	Ok	Ok	Ok	Ok	Six markers drop out; no haplogroup identified	Ok	Ok	Ok	Ok	Two markers drop out; no haplogroup identified	Ok	Typing correct; Clerical error
03	Ok	Ok	Ok	Ok	Five markers drop out; macrobranch identified	Ok	Ok	Ok	Ok	10 markers drop out; no haplogroup identified	One marker ambiguous	Ok
04	Ok	Ok	Ok	Ok	Nine markers drop out; no haplogroup identified	Ok	Two markers drop out	Ok	Ok	Total drop out of all markers	One marker ambiguous	Ok
05	Ok	Ok	Ok	Ok	Nine markers drop out; no haplogroup identified	Ok	Ok	Ok	Ok	Total drop out of all markers	Ok	Ok

Comments were partially from the labs or added after inspection of the raw data.

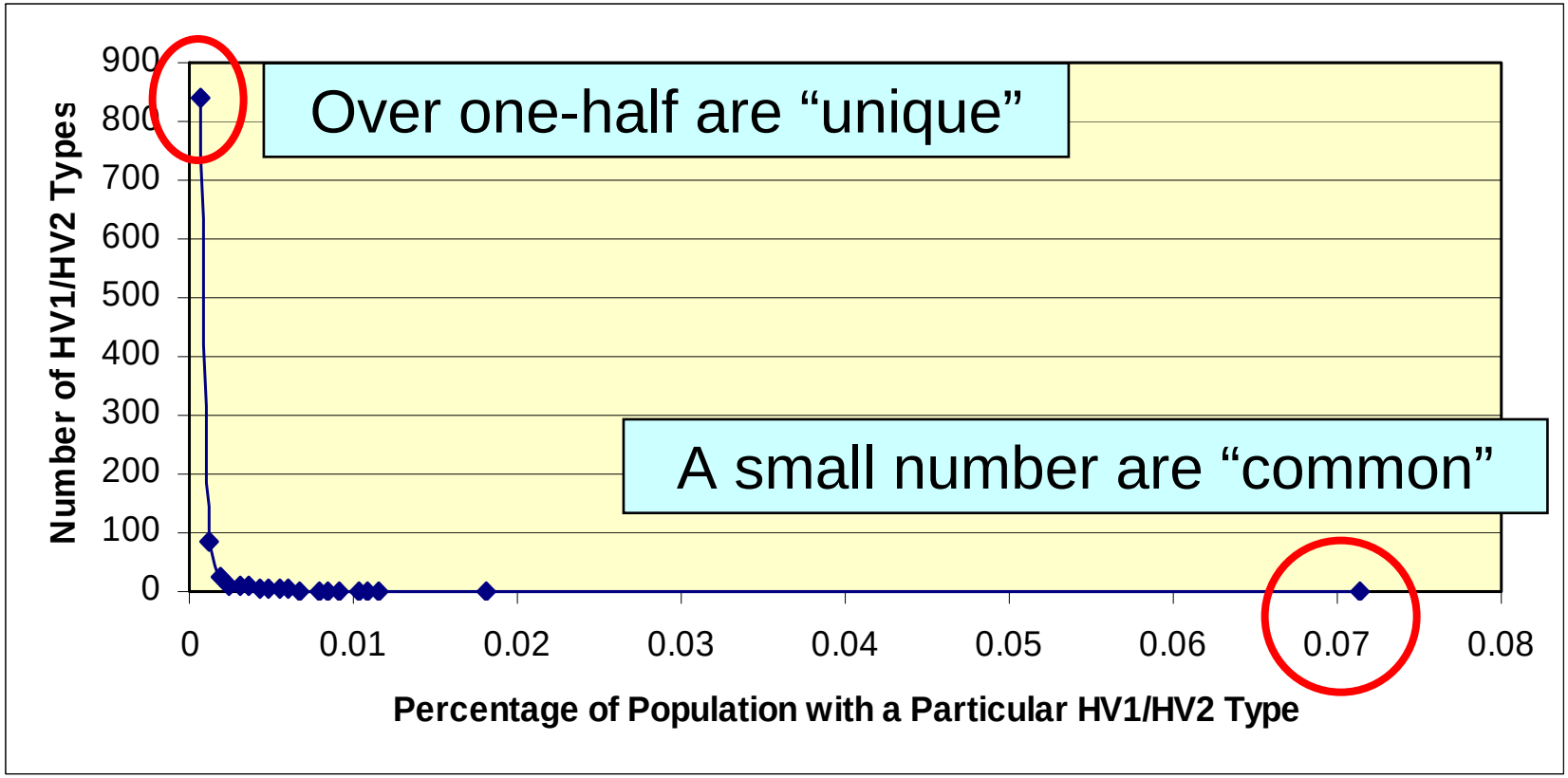
What Are We?
(Why are we here?)

MtDNA SNPs

- As a screening tool for sorting hair evidence or to eliminate multiple suspects.
- To increase the power of discrimination for common mtDNA haplotypes that match control region sequence data.

mtDNA Population Distribution

Caucasians (n=1665)



263 A-G, 315.1C

Coble *et al.* (2004)

“Singletons”

Table 1
Some haplotype populations sample statistics.

	Sample size	Singletons	Singleton proportion
	$n-1$	$\alpha-1$	$\kappa=\alpha/n$
Mitotyping technologies			
mtDNA	2000	~1200	~60%
mtDNA AFDIL			
All races	7867	4941	0.63
Caucasians	1219	750	0.62

Brenner (2010) *FSI-Genetics* **4:** 281-291

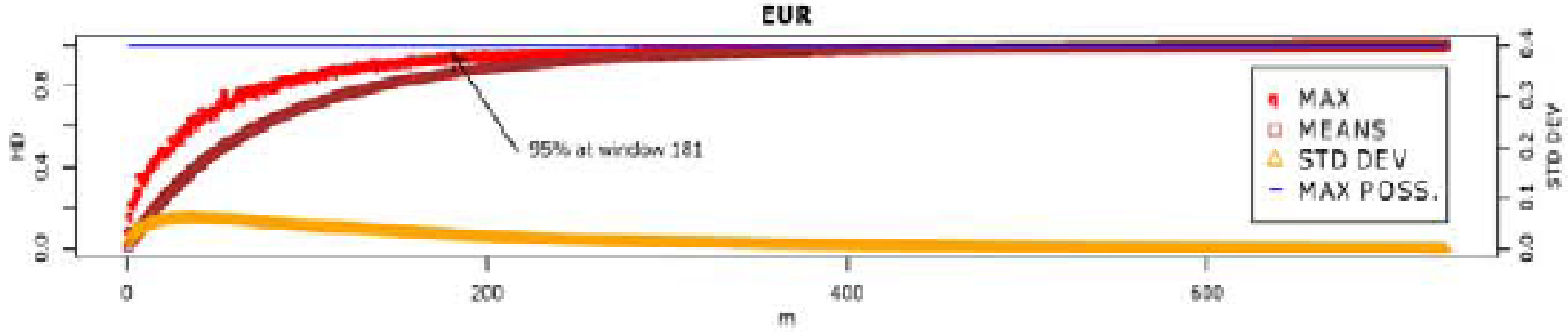
OPEN ACCESS Freely available online

PLoS one

A Reduced Number of mtSNPs Saturates Mitochondrial DNA Haplotype Diversity of Worldwide Population Groups

Antonio Salas[1]*, **Jorge Amigo**[1,2]

1 Unidade de Xenética, Departamento de Anatomía Patolóxica e Ciencias Forenses, and Instituto de Medicina Legal, Facultade de Medicina, Universidade de Santiago de Compostela, Galicia, Spain, **2** Grupo de Medicina Xenómica, Universidade de Santiago de Compostela, Galicia, Spain



			EUR		
Study	n° SNPs	HD	N	H	HD
Brandstätter et al. (2003)	16	0.7153	15	0.0647	0.9155
Vallone et al. (2004)	11	0.7508	11	0.0416	0.8242
Quintáns et al. (2004)	17	0.7446	16	0.0416	0.9075
Umetsu et al. (2005)	36	0.9453	21	0.0600	0.8975
Grignani et al. (2005)	16	0.5214	13	0.0370	0.6030
Brandstätter et al. (2006)	45	0.7313	34	0.0878	0.8803
Wiesbauer et al. (2006)	10	0.4848	10	0.0370	0.8424
Lee et al. (2006)	22	0.9225	7	0.0208	0.5636
Álvarez-Iglesias et al. (2006)	32	0.8630	13	0.0393	0.5692
Coble et al. (2004)	59	0.8889	57	0.1894	0.9463
Endicott et al. (2006)	20	0.0666	1	0.0046	0.0046
Köhnmenn et al. (2008)	22	0.8504	22	0.0993	0.9428
Wu et al. (2008)	10	0.8171	9	0.0208	0.5736
Watkins et al. (2008)	32	0.7736	21	0.0439	0.7730
Rosa et al. (2008)	19	0.8394	18	0.0600	0.8978
Present study	9/22/11/10/10	0.9950	88	0.4111	0.9909
Maximum possible values	–	0.9998	–	0.8545	0.9989

Salas and Amigo (2010) *PLoS One* 5(5): e10218

The Nature of mtDNA SNPs

		Polymorphic nucleotide positions[a]									
		1111111111	1111111111	1111111111	1111111111	1111111111	11111				
		6666666666	6666666666	6666666666	6666666666	6666666666	66666				
		0001111111	1111111112	2222222222	2222222222	2222333333	33333	111111	1111112222	2222222222	33333
		5690122344	5567788991	2223344456	6677788899	9999000111	22556	5667445558	8889990000	1222235569	00111
		1934169457	3632669233	2340108961	3504814601	2467049168	45262	7043360230	5894593457	5256840935	99159
S[b]	H[c]							aa			ab
1	J	-T---C----	----T-----	----------	----------	----------	-----	---G------	----------	----A---GT	C--C-
23	J	-T---C----	----------	----------	----------	----------	-----	---G-C----	AG--------	-T--A---GT	---C-
46	J	-T---C----	----------	----------	----------	----------	-----	---G------	A---------	----A---GT	---C-
11	J	-T---C--A-	----------	----C----T	----------	----------	-----	---G--TC--	----C-----	G-------GT	--TCC
35	J	-T---C--A-	----------	T--------T	---A------	----------	-----	---G------	----------	--------GT	---C-
12	J	-T---C----	--------T-	----------	----------	-------C--	-----	---G--TC--	----------	--------GT	---C-
21	J	-T---C----	--------T-	--------T-	----------	----G-G---	-----	---G---C--	----------	--------GT	---C-

Haplogroup <u>SNPs</u>
16069 C-T
16126 T-C
295 C-T

Sub-Haplogroup <u>SNPs</u>
16193 C-T (J1/J2)
185 G-A (J1/J3)

<u>Hot Spots</u>
152 T-C

<u>Private Mutations</u>
222 C-T

Table 2. Excerpt of the data in Table S1 showing the top five mtSNP that are shared between the top 15 mtSNPs in the three main continental groups.

Position	AFR	AFR-AM	ASI	EUR	HIS	ALL	rCRS	Variant	MapLocus	MR
16519	1	1	1	1	1	1	T	C	MT-DLOOP1	1
152	2	3	4	2	5	2	T	C	MT-DLOOP2	2
16189	3	7	2	6	14	3	T	C	MT-DLOOP1	6
16129	6	115	3	8	6	4	G	A	MT-DLOOP1	7
195	7	44	9	14	0	8	T	C	MT-DLOOP2	5

MR (mutational ranking) column refers to the position of these variants in the list of relative site-specific mutation rates as reported in [29]. Other legends are as in Table S1.

doi:10.1371/journal.pone.0010218.t002

Salas and Amigo (2010) *PLoS One* 5(5): e10218

What Are We?
(Why are we here?)

MtDNA SNPs

- As a screening tool for sorting hair evidence or to eliminate multiple suspects.
- To increase the power of discrimination for common mtDNA haplotypes that match control region sequence data.

Y-SNPs

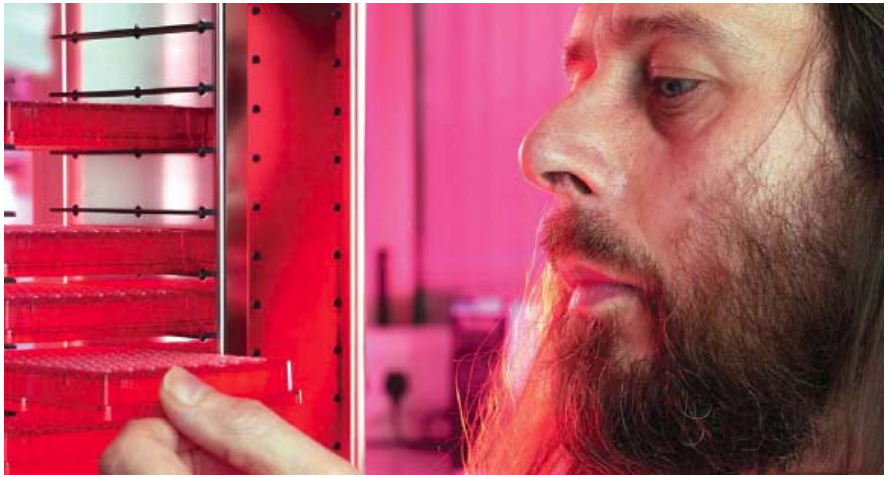
- No real forensic use…

Am. J. Hum. Genet. 72:717–721, 2003

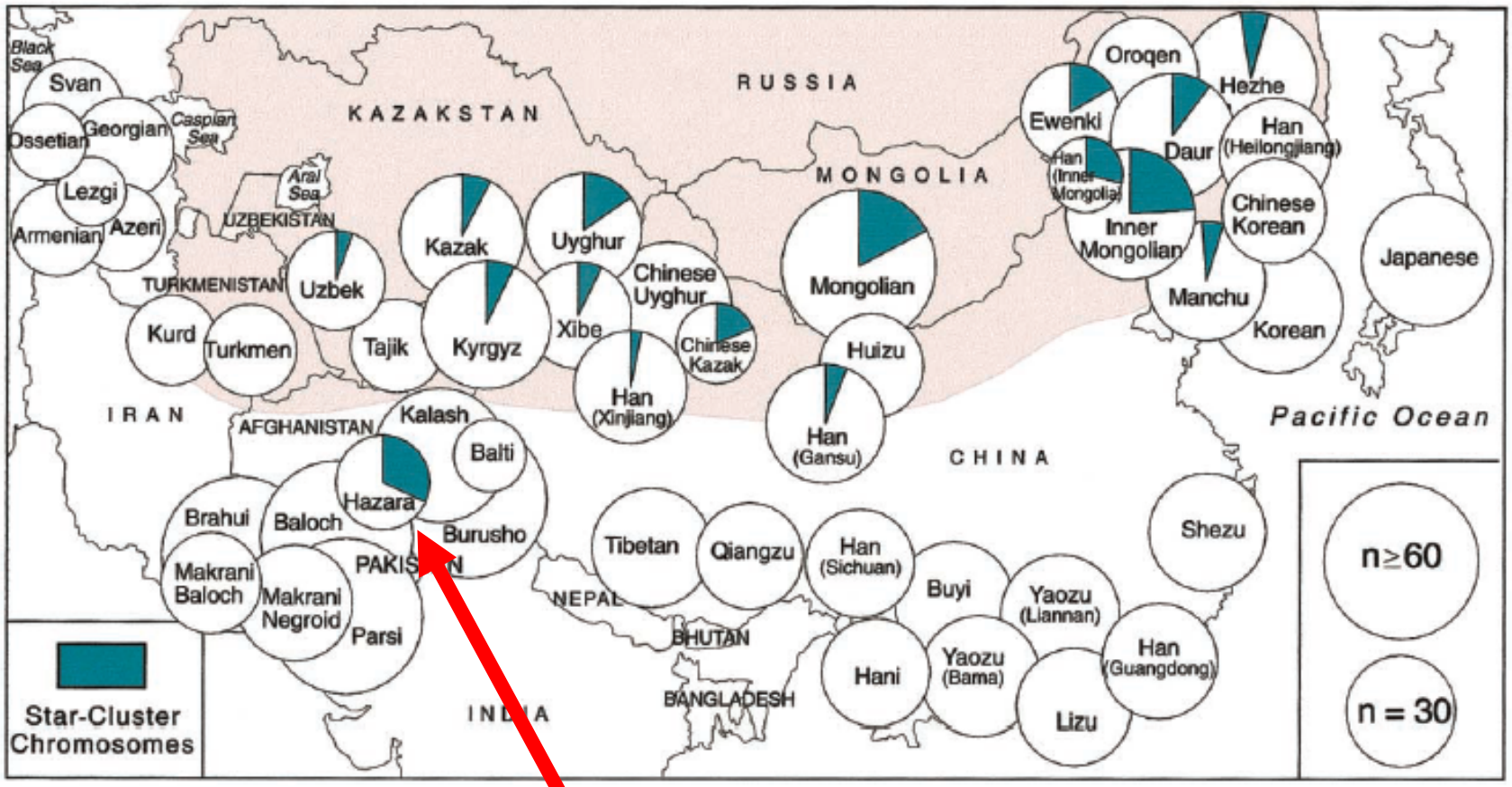
Report

The Genetic Legacy of the Mongols

Tatiana Zerjal,[1] Yali Xue,[1,2] Giorgio Bertorelle,[3] R. Spencer Wells,[4] Weidong Bao,[1,5] Suling Zhu,[1,5] Raheel Qamar,[1,6] Qasim Ayub,[1,6] Aisha Mohyuddin,[1,6] Songbin Fu,[2] Pu Li,[2] Nadira Yuldasheva,[4,7] Ruslan Ruzibakiev,[7] Jiujin Xu,[5] Qunfang Shu,[5] Ruofu Du,[5] Huanming Yang,[5] Matthew E. Hurles,[8] Elizabeth Robinson,[1,*] Tudevdagva Gerelsaikhan,[1,†] Bumbein Dashnyam,[9] S. Qasim Mehdi,[5] and Chris Tyler-Smith[1]



Chris Tyler-Smith


Black Sea
Svan
Georgian
Ossetian
Caspian Sea
Lezgi
Armenian
Azeri
Aral Sea
UZBEKISTAN
TURKMENISTAN
Uzbek
Kurd
Turkmen
Tajik
Kazak
Kyrgyz
Uyghur
Xibe
Chinese Uyghur
Chinese Kazak
Han (Xinjiang)
KAZAKSTAN
RUSSIA
MONGOLIA
Mongolian
Huizu
Han (Gansu)
Oroqen
Hezhe
Ewenki
Daur
Han (Heilongjiang)
Han (Inner Mongolia)
Inner Mongolian
Chinese Korean
Manchu
Korean
Japanese
Pacific Ocean
CHINA
IRAN
AFGHANISTAN
Kalash
Balti
Hazara
Brahui
Baloch
Burusho
Makrani Baloch
Makrani Negroid
Parsi
PAKISTAN
Tibetan
Qiangzu
Han (Sichuan)
Buyi
Yaozu (Liannan)
Shezu
NEPAL
BHUTAN
Hani
Yaozu (Bama)
Han (Guangdong)
Lizu
BANGLADESH
INDIA
Star-Cluster Chromosomes
n≥60
n = 30

washingtonpost.com

Tom Robinson

DNA Shows Man a Descendant of Genghis Khan

By JILL LAWLESS
The Associated Press
Tuesday, June 6, 2006; 8:30 PM

LONDON -- Tom Robinson had long wondered about his family tree. He never suspected its roots might lie in the Mongolian steppe.

The Florida accountant knew that his great, great-grandfather had come to the United States from England _ but beyond that his research drew a blank. So he turned to the burgeoning field of "bioarchaeology," having his DNA tested to see what it revealed about his origins.

“I haven’t done any conquering, per se.”

The New York Times

In the Body of an Accounting Professor, a Little Bit of the Mongol Hordes

By NICHOLAS WADE
Published: June 6, 2006

Oxford Ancestors, the world's foremost and leading company in ancestral DNA analysis has uncovered the first American descendent of the great warlord **Genghis Khan**... **Tom Robinson**, Associate Professor of Accountancy and professional investment consultant, of Miami, Florida, USA.

It turns out that **Dr Robinson** is a direct descendent of **Genghis**, and he is the first American to find this out through a genetic test. His Y-Chromosome bears an astonishing seven out of nine possible genetic markers identical to **Genghis Khan's** (as DNA mutates over generations, two altering DNA markers is a remarkably low number for a period stretching over 700 years).

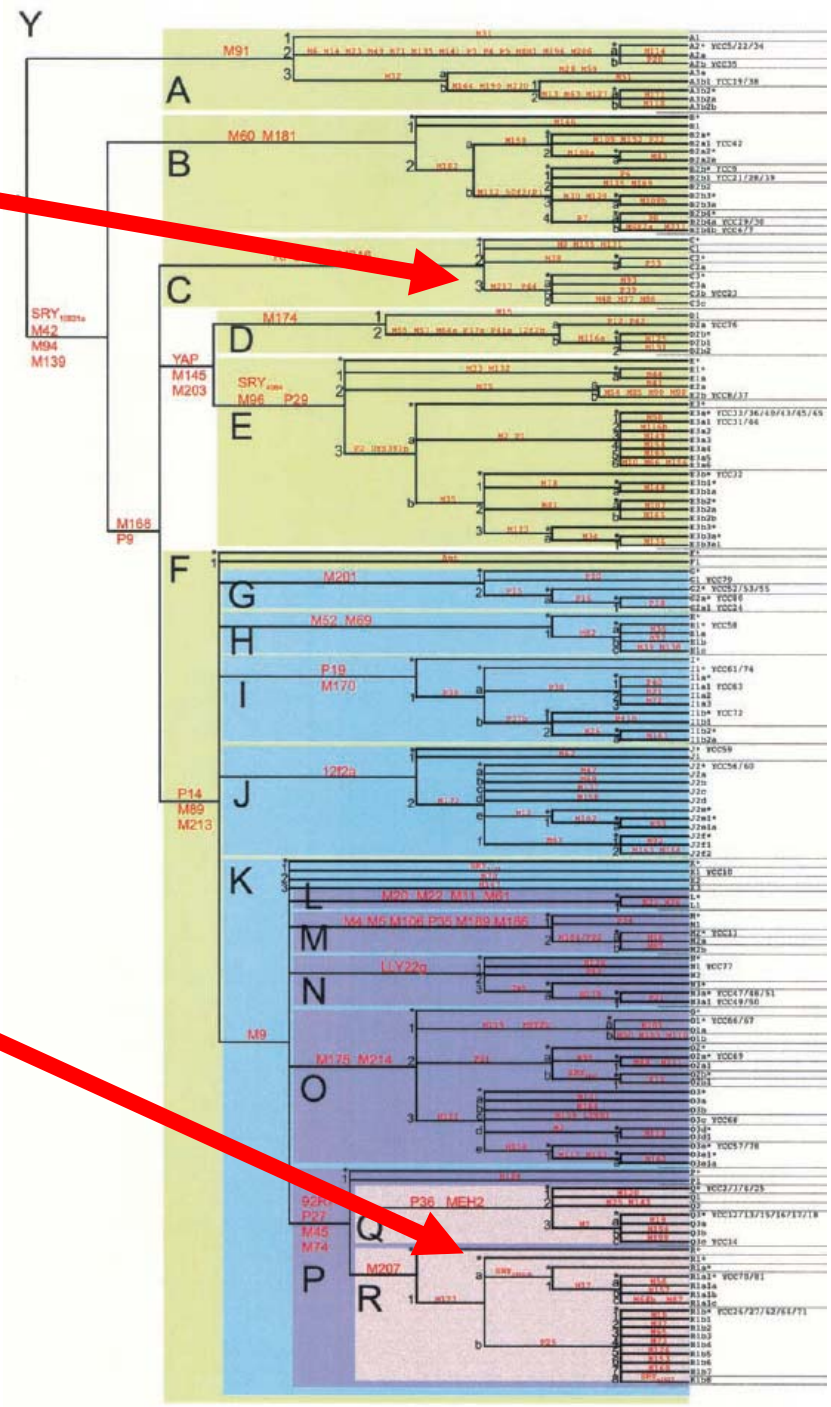
Marker	Oxford Ancestors Robinson	Mongolian Benchmark
DYS19	16	16
DYS390	25	25
DYS391	10	10
DYS392	11	11
DYS393	13	13
DYS389I	13	13
DYS389II	31	29
DYS425	12	12
DYS426	12	11

Ghengis Khan – is thought to have belonged to the Asian YHg - C3

Additional testing by Family Tree DNA found that Tom Robinson belonged to Y-haplogroup R1a (W. European)

“A match at 10 fast-mutating sites is outvoted by a discrepancy at one slow-mutating site.”

Dr. Chris Tyler-Smith



Haplogroup Predictor

Y Haplogroup Prediction from Y-STR Values

What's NEW?

Home | Instructions | Conventions | ISOGG Y Tree

01 Jan 2009
Changed I2a2 to I2a1 to match ISOGG-2009

09 Jun 2008
Added C3 and G1 to beta
Changed to ISOGG-2008 Names

01 Feb 2008
Updated database

Which One Are You In?

Haplogroup G2c
Haplogroup H
Haplogroup I1

NOTE: A "batch" version of this program is now available for application to large numbers of haplotypes. Please CLICK HERE for instructions

Acknowledgement

The haplogroup program has been recoded in a very efficient manner by Doug McDonald, and his considerable contributions are gratefully acknowledged.

Click your program choice:

21-Haplogroup Program → FTDNA Order | Numeric Order

23-Haplogroup Beta Program → FTDNA Order | Numeric Order

Prediction of Y-chromosome haplogroup from Y-STR values is easy and fast with this program. You may choose a version of the program with the markers in FTDNA order or numerical order by clicking the button of your choice above. First time users should read the Instructions page first (see tabs above). Some companies use different standards for some markers, so read the "Conventions" page (see tabs above) to see which standards (nomenclature) are used in this program. Markers for which there are nomenclature issues are colored pink on the data entry page. An example of the data entry screen is shown below.

There are links on the right to two articles describing the "fitness score" calculations and the Bayesian probability calculations.

Click here for article that explains fitness scores in the Journal of Genetic Genealogy, 1:1-7 (2005)

Click here for article that explains the Bayesian approach to probabilities in the *Journal of Genetic Genealogy*, 2:34-39 (2006)

Click the ISOGG link above for a detailed Y phylogenetic tree and descriptions of the haplogroups with references and resources.

http://www.hprg.com/hapest5/

Enter any combination of one or more markers, *or* use the string entry form below

Area Selection: Equal priors

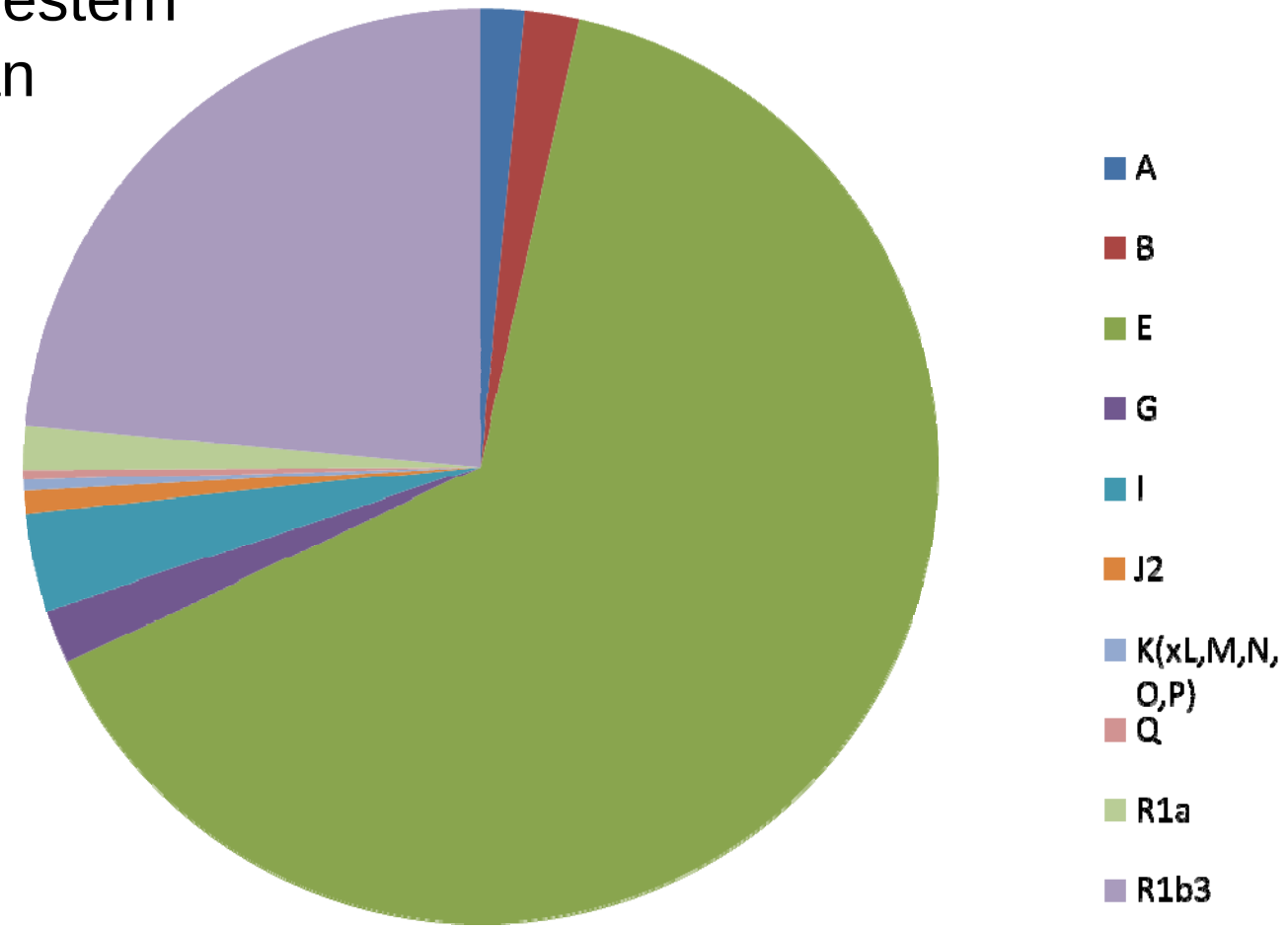
Reset

19	385a	385b	388	389|1	389|2	390	391
0	0	0	0	0	0	0	0
392	393	395a	395b	406	413a	413b	425
0	0	0	0	0	0	0	0
426	436	437	438	439	441	442	444
0	0	0	0	0	0	0	0
445	446	447	448	449	450	452	454
0	0	0	0	0	0	0	0
455	456	458	459a	459b	460	461	462
0	0	0	0	0	0	0	0
463	464a	464b	464c	464d	472	481	485
0	0	0	0	0	0	0	0
487	490	492	495	505	508	511	520
0	0	0	0	0	0	0	0
522	531	532	533	534	537	540	556
0	0	0	0	0	0	0	0
557	565	568	570	572	576	578	590
0	0	0	0	0	0	0	0
594	607	617	635C4	640	641	643	1B07
0	0	0	0	0	0	0	0
A10	CDYa	CDYb	H4	YCAIIa	YCAIIb		
0	0	0	0	0	0		

Results Table

Haplo-group	Fitness score	Proba-bility (%)
E1b1a	-	-
E1b1b	-	-
G2a	-	-
G2c	-	-
H	-	-
I1	-	-
I2a (xI2a1)	-	-
I2a1	-	-
I2b (xI2b1)	-	-
I2b1	-	-
J1	-	-
J2a1b	-	-
J2a1h	-	-
J2a1 x J2a1-bh	-	-
J2b	-	-
L	-	-
N	-	-
Q	-	-
R1a	-	-
R1b	-	-
T	-	-

~25% Western European



259 self-described African Americans

Lao et al. Human Mutation in press

Where Are We Going?

- Limitations of present SNP testing…
  - Allele Specific Primer Extension assays are not in a "kit" format for rapid validation and QC.

Where Are We Going?

- Limitations of present SNP testing…

Int J Legal Med (2007) 121:493–499
DOI 10.1007/s00414-007-0177-3

TECHNICAL NOTE

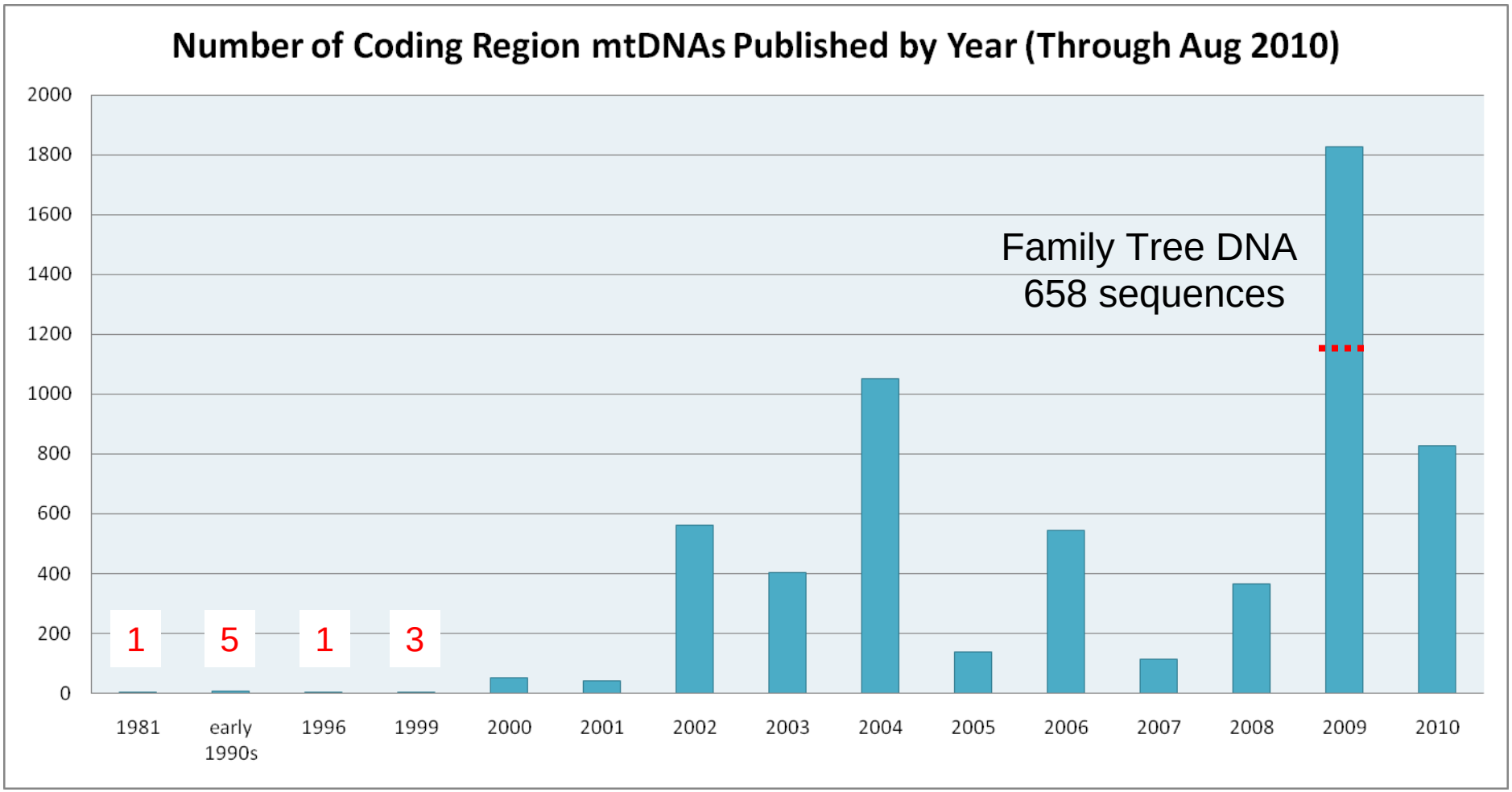
First successful assay of Y-SNP typing by SNaPshot minisequencing on ancient DNA

C. Bouakaze · C. Keyser · S. Amory · E. Crubézy · B. Ludes

Where Are We Going?

- Limitations of present SNP testing…
  - Allele Specific Primer Extension assays are not in a "kit" format for rapid validation and QC.
  - What SNPs to test? Ideally, targeting private mutations are the most forensically important – but the Y-chr is 58million bp!
  - Statistics for matching haplotypes – Using the whole genome can "downgrade" your stats…

mitoGenomics



Contents lists available at ScienceDirect

Forensic Science International: Genetics

journal homepage: www.elsevier.com/locate/fsig

Short Communication

mtGenome reference population databases and the future of forensic mtDNA analysis

Jodi A. Irwin[a,*], Walther Parson[b], Michael D. Coble[a,1], Rebecca S. Just[a]

[a] Armed Forces DNA Identification Laboratory, Armed Forces Institute of Pathology, 1413 Research Blvd., Rockville, MD 20850, USA
[b] Institute of Legal Medicine, Innsbruck Medical University, Müllerstrasse 44, Austria

Journal of Human Genetics 54, 174-181 (March 2009)

Median network analysis of defectively sequenced entire mitochondrial genomes from early and contemporary disease studies

Hans-Jürgen Bandelt, Yong-Gang Yao, Claudio M Bravi, Antonio Salas and Toomas Kivisild

The American Journal of Human Genetics *85*, 929–945,

LETTERS TO THE EDITOR

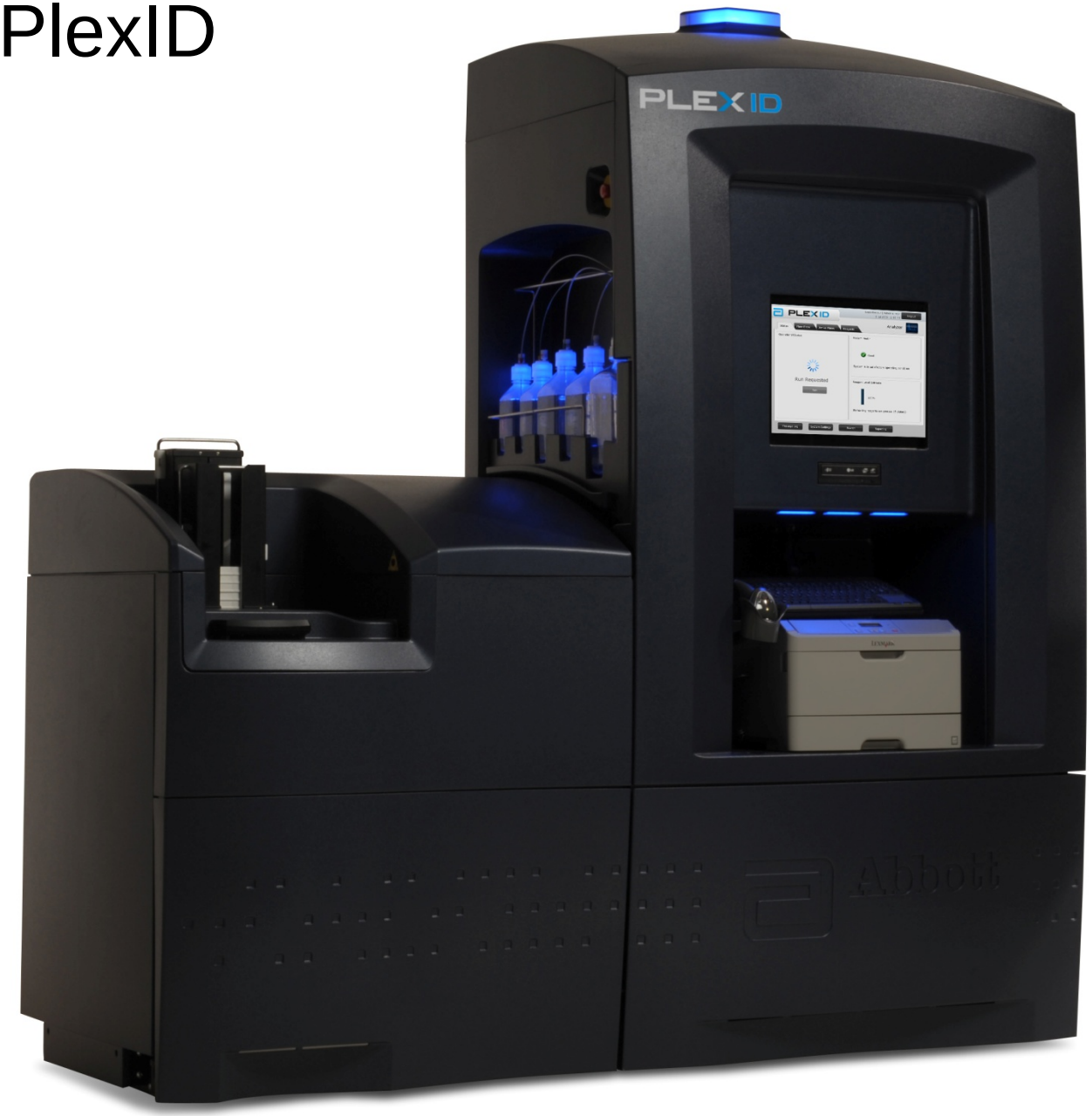
mtDNA Data Mining in GenBank Needs Surveying

Yong-Gang Yao,[1] Antonio Salas,[2] Ian Logan,[3] and Hans-Jürgen Bandelt[4,*]

Where Are We Going?

- Technology improvements for rapid, high-throughput SNP testing.

Abbott PlexID



ABI Solid

Roche 454

Ilumina
Solexa

Forensic Science International: Genetics 4 (2010) 59–61

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journal homepage: www.elsevier.com/locate/fsig

Review

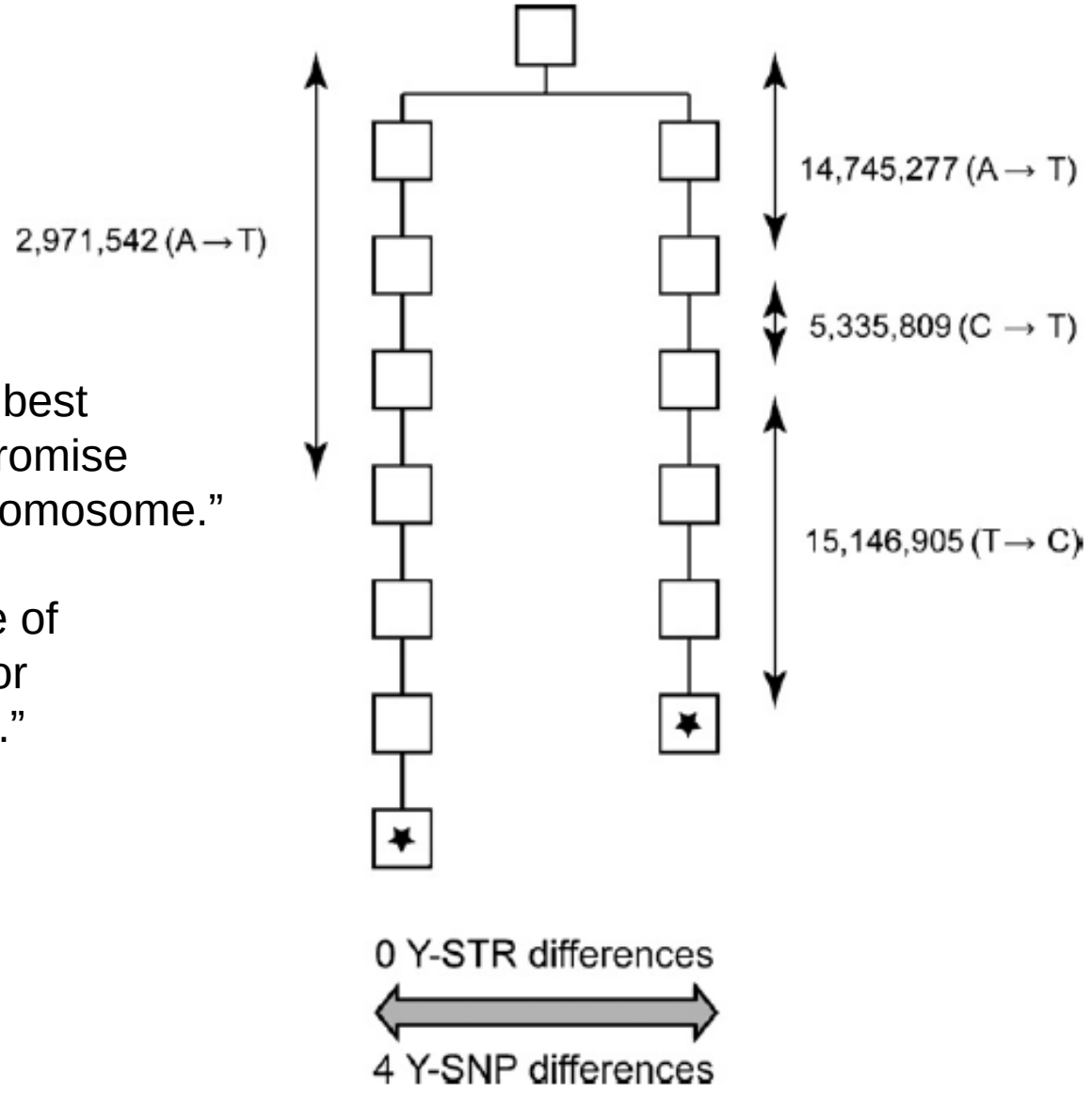
The hare and the tortoise: One small step for four SNPs, one giant leap for SNP-kind

Yali Xue, Chris Tyler-Smith *

The Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Hinxton, Cambs CB10 1SA, UK

"Y-SNPs therefore now offer the best resolution of Y haplotypes and promise to distinguish almost every Y chromosome."

"This work illustrates the promise of current sequencing technology for forensically relevant applications."



Y. Xue, C. Tyler-Smith / Forensic Science International: Genetics 4 (2010) 59–61

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journal homepage: www.elsevier.com/locate/fsig

Commentary

A comment on "The hare and the tortoise: One small step for four SNPs, one giant leap for SNP-kind"

António Amorim [a,b,*]

[a] IPATIMUP, Instituto de Patologia e Imunologia Molecular da Universidade do Porto, Portugal
[b] Faculdade de Ciências, Universidade do Porto, Portugal

Interpretation of the evidence
Legal and ethical constraints

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journal homepage: www.elsevier.com/locate/fsig

Discussion

Response to the comment on "The hare and the tortoise: One small step for four SNPs, one giant leap for SNP-kind"

Yali Xue, Chris Tyler-Smith *

The Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Hinxton, Cambs. CB10 1SA, UK

"We agree with Amorim that caution is needed, but now is the time to begin both research exploration of the forensic potential of these technologies and discussions of their application."

Conclusions

- Haploid SNPs can be useful for sorting, screening, and increasing discrimination of highly challenged samples where nDNA is not reliably amplified.
- It is important to realize the limitations of your sample!!!
- New technologies can increase the use of haploid markers for forensic investigations.

Acknowledgements

NIST Applied Genetics

The NIST Human Identity Project Team

(Forensic DNA & DNA Biometrics)

Funding from the **National Institute of Justice (NIJ)** through the NIST Office of Law Enforcement Standards and the **FBI S&T Branch** through the NIST Information Access Division

…Bringing traceability and technology to the scales of justice…


John Butler


Erica Butts


Dave Duewer


Becky Hill


Margaret Kline


Kristen O'Connor


Jan Redman


Pete Vallone

Current and Former AFDIL Research


Jodi Irwin


Rebecca Just


Odile Loreille


Tom Parsons

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