health

close relatives

Genetic Genealogy

ethnicity

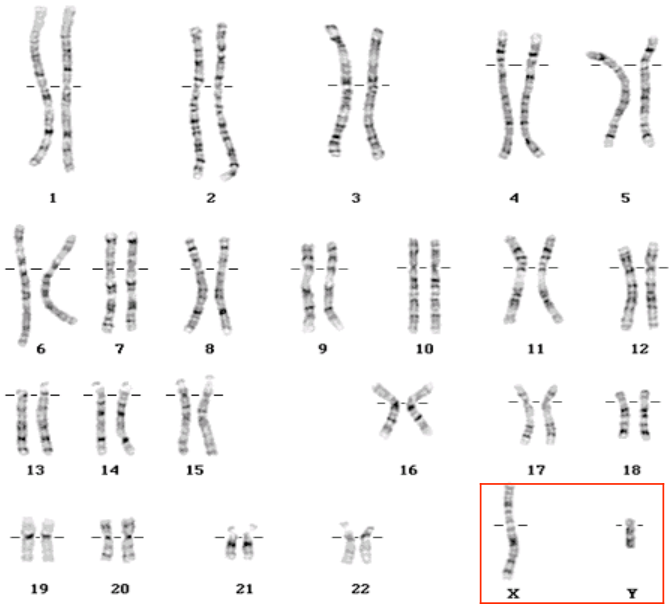
by Dirk Schweitzer, Ph.D.

2010-05-21

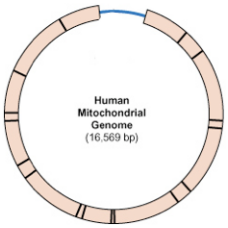
TEDx Lansing

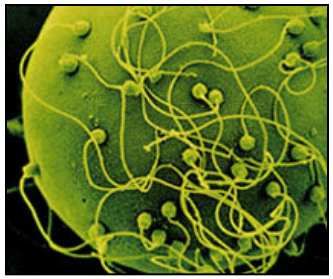
x = independently organized TED event

direct ancestry

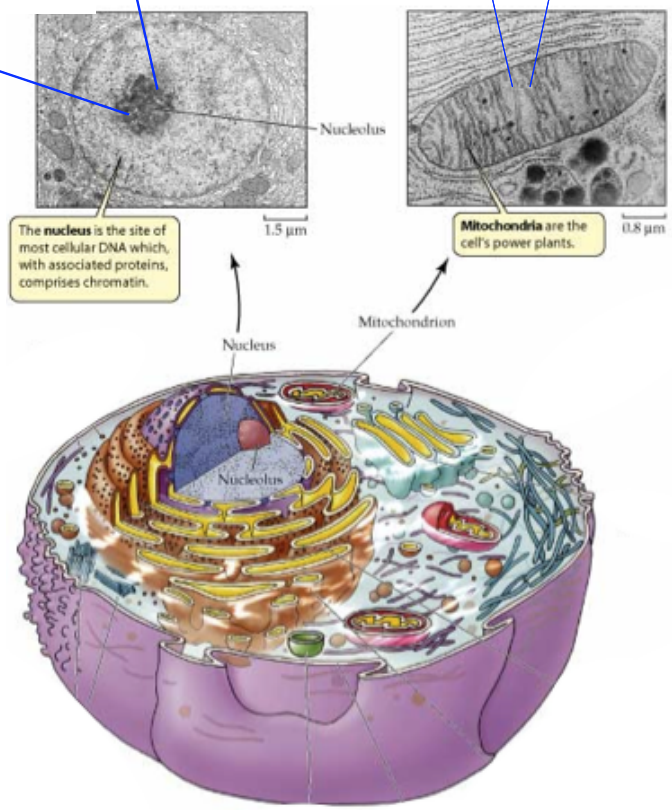


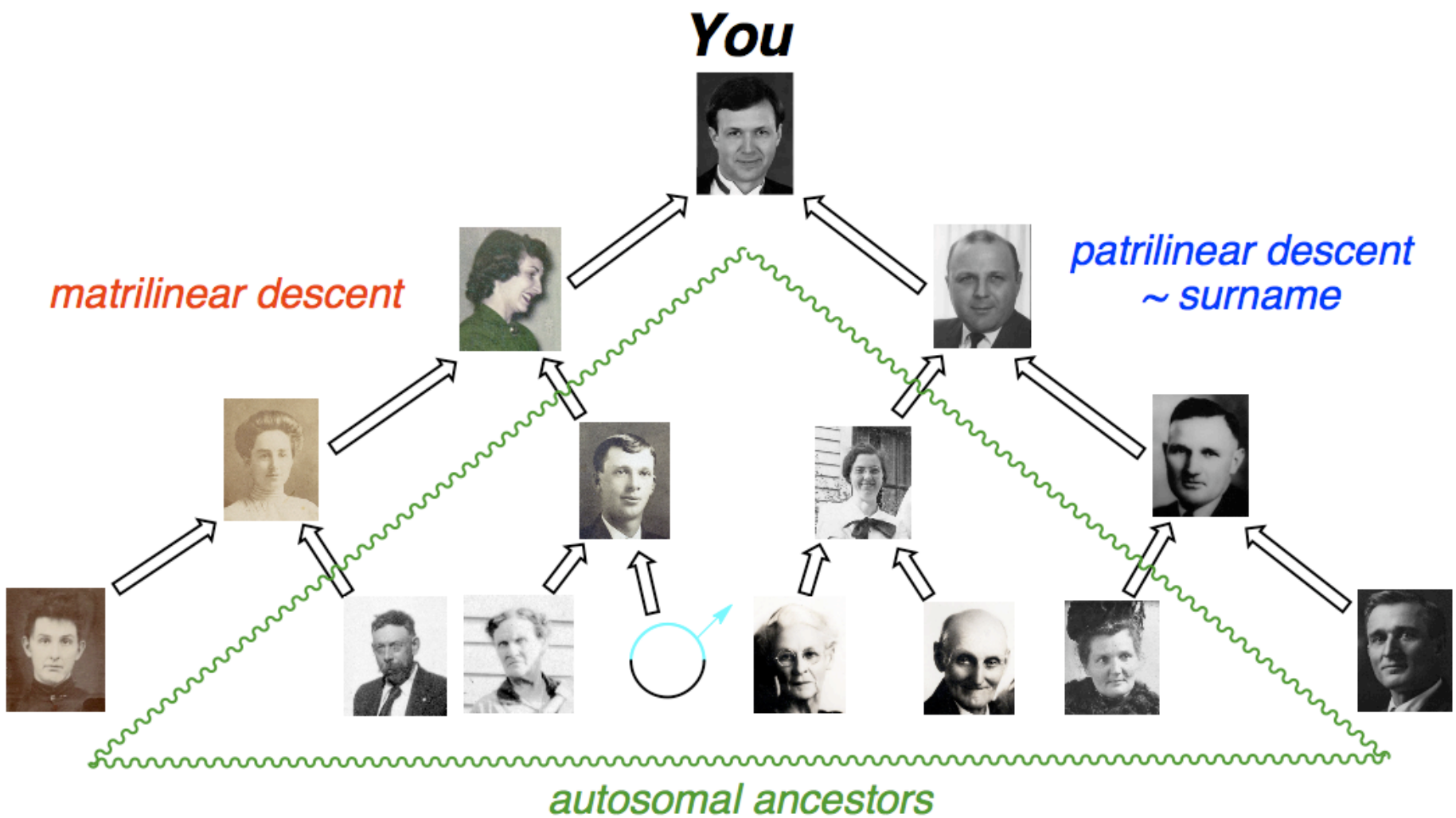
Sex Chromosomes.
The Y-chromsome is always inherited from a father to his sons = patrilinear descent.



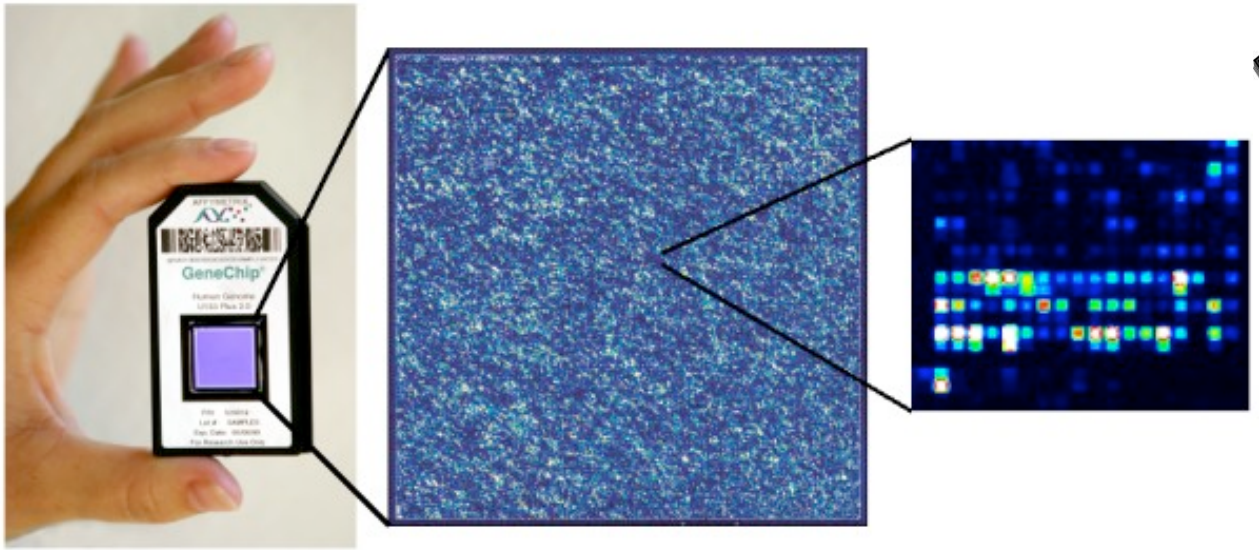


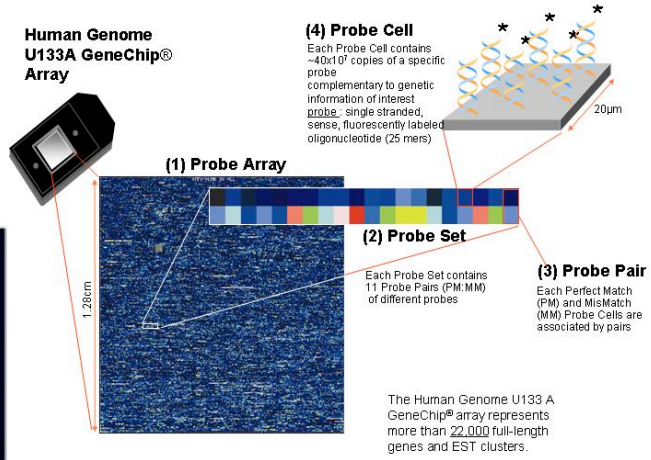
The mitochondria are always inherited from a mother to her children = matrilinear descent.

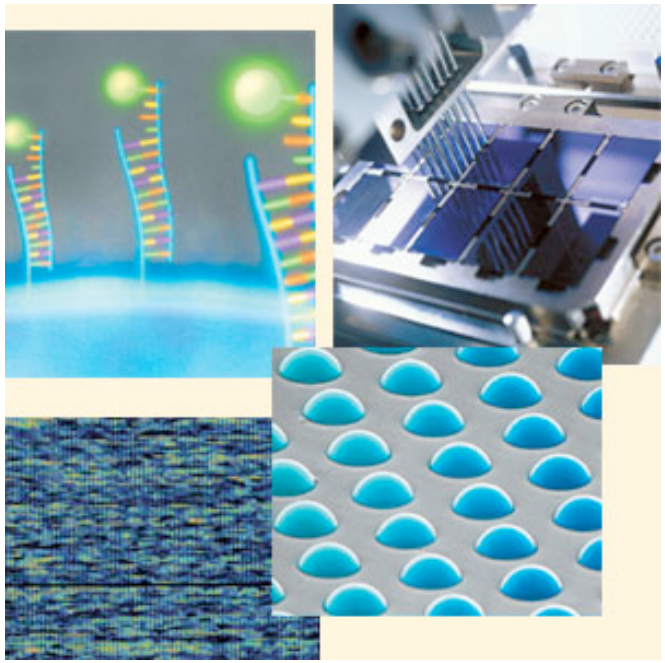



You
matrilinear descent
patrilinear descent
~ surname
autosomal ancestors

Using gene-chip / DNA-microarray technology:







~ 600,000 investigated positions
on the genome provide:

- health information,
- ethnicity estimate,
- identification of close relatives, &
- direct male & female ancestry.

Show results for Dirk Schweitzer

See new and recently updated reports »

Disease Risks (79)

Elevated Risks	Your Risk	Average Risk
Restless Legs Syndrome	2.5%	2.0%
Exfoliation Glaucoma	2.2%	0.7%
Ulcerative Colitis	1.4%	0.8%
Multiple Sclerosis	0.8%	0.3%

more »

Decreased Risks	Your Risk	Average Risk
Type 2 Diabetes	18.3%	23.7%

more »

multi-factorial diseases

See all 79 risk reports...

Carrier Status (24)

Familial Hypercholesterolemia Type B new	Variant Absent
Familial Mediterranean Fever new	Variant Absent
Phenylketonuria new	Variant Absent
Alpha-1 Antitrypsin Deficiency	Variant Absent
Bloom's Syndrome	Variant Absent
BRCA Cancer Mutations (Selected)	Variant Absent
Canavan Disease	Variant Absent
Cystic Fibrosis	Variant Absent

See all 24 carrier status...

Mendelian disorders are caused by a single deleterious mutation.

Traits (33)

Alcohol Flush Reaction	Does Not Flush
Bitter Taste Perception	Unlikely to Taste
Earwax Type	Wet
Eye Color	Likely Blue
Lactose Intolerance	Likely Tolerant

See all 33 traits...

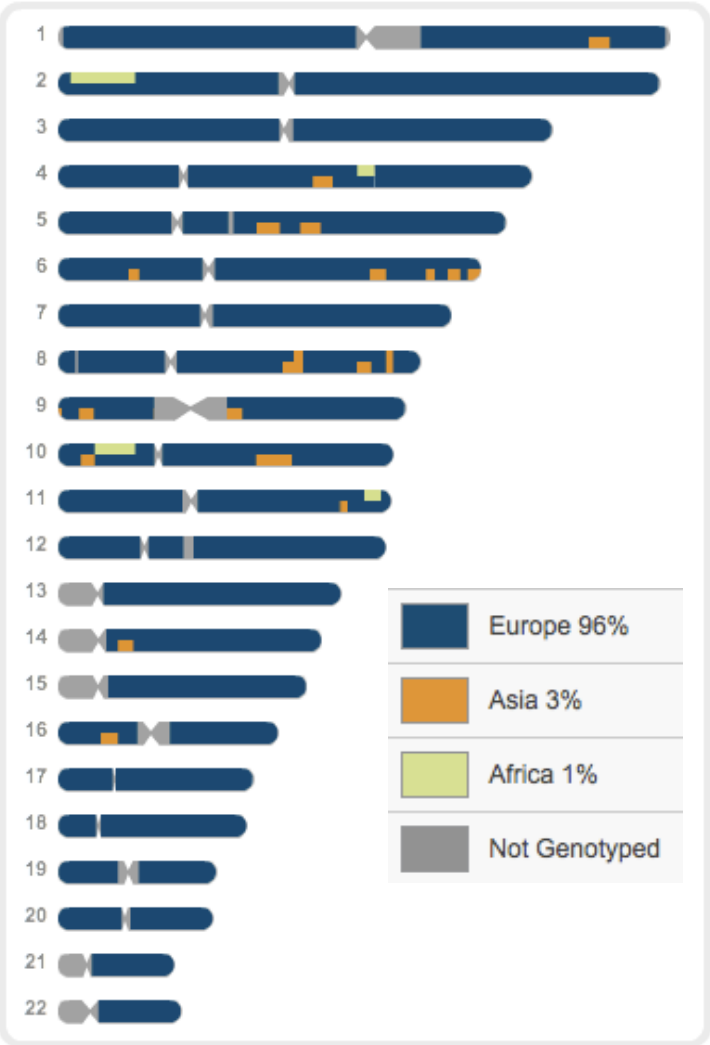
Drug Response (15)

Clopidogrel (Plavix®) Efficacy	Reduced
Warfarin (Coumadin®) Sensitivity	Increased
Abacavir Hypersensitivity	Typical
Alcohol Consumption, Smoking and Risk of Esophageal Cancer	Typical
Fluorouracil Toxicity	Typical

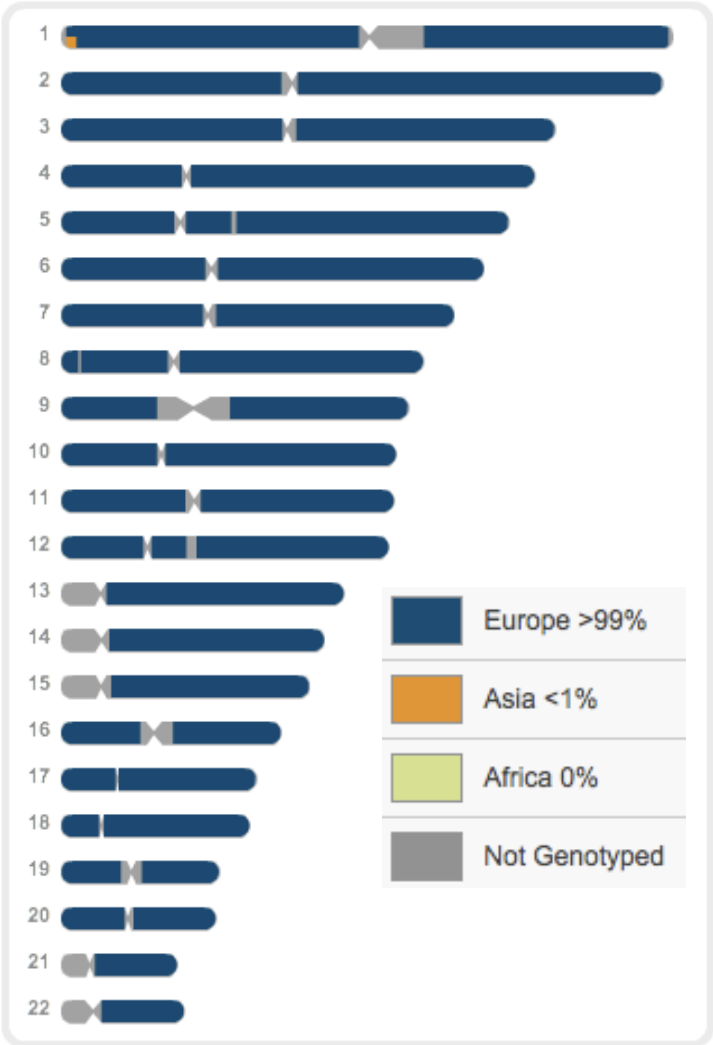
See all 15 drug response...

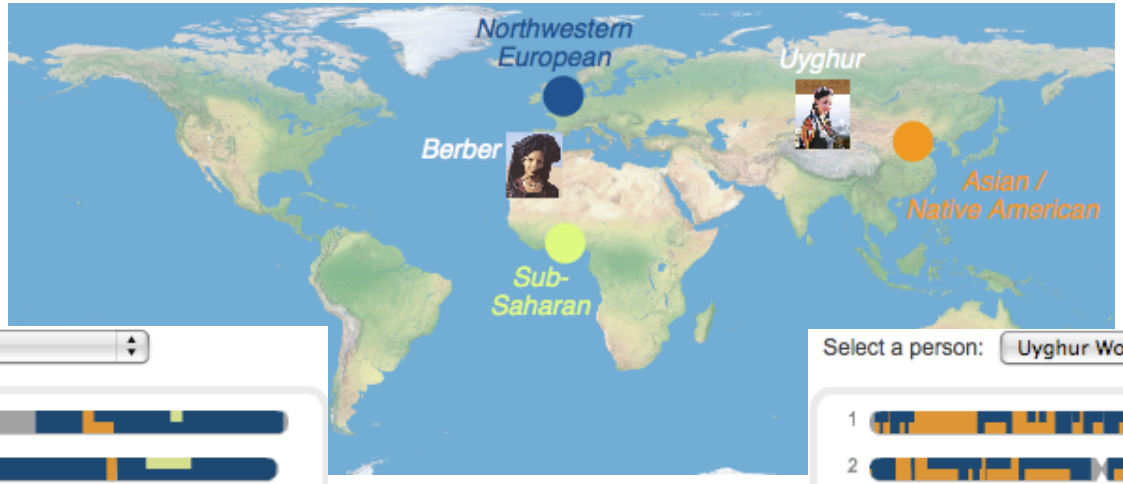
Epigenetic diseases are not detected.
http://en.wikipedia.org/wiki/Epigenetic#Epigenetic_effects_in_humans

"European" Brazilian

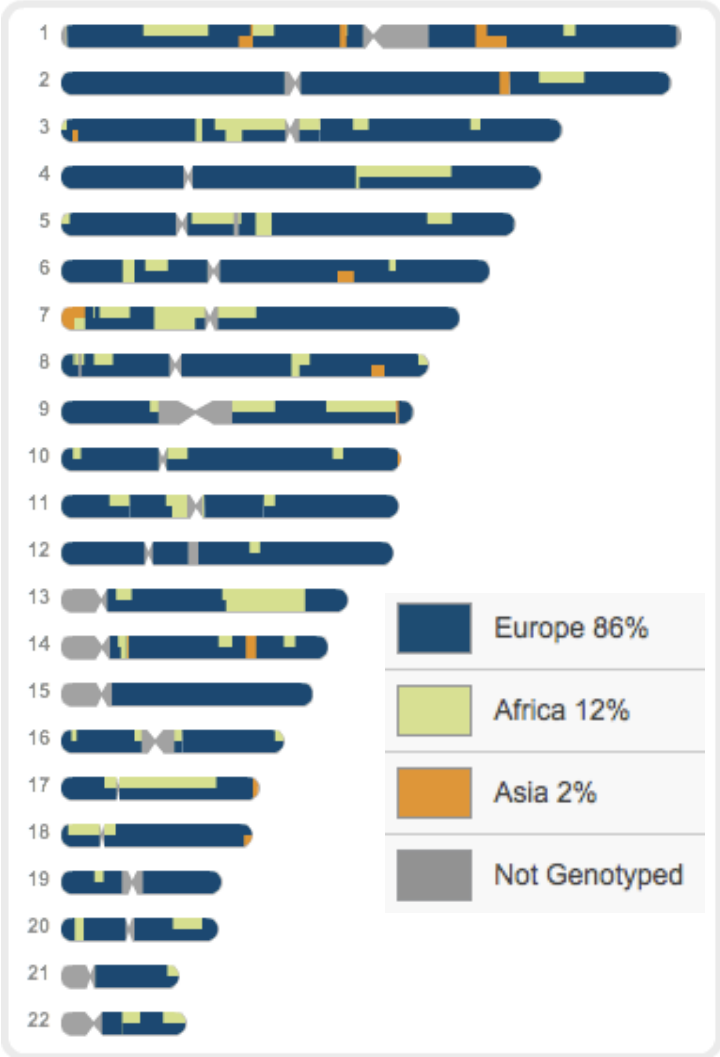


"European" USA citizen

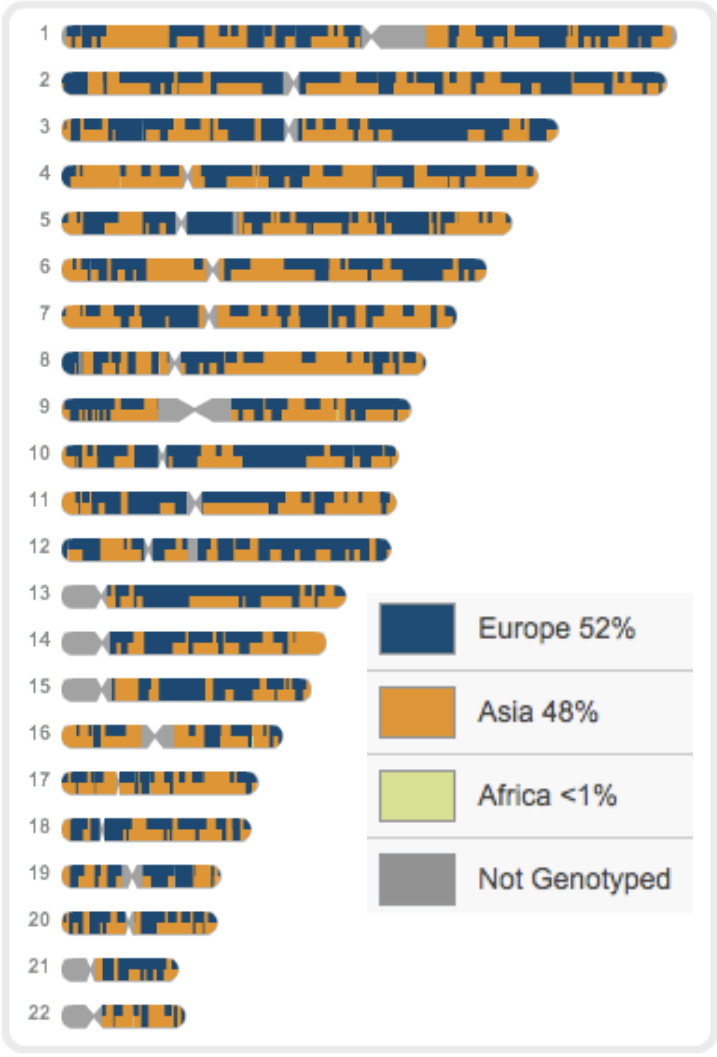


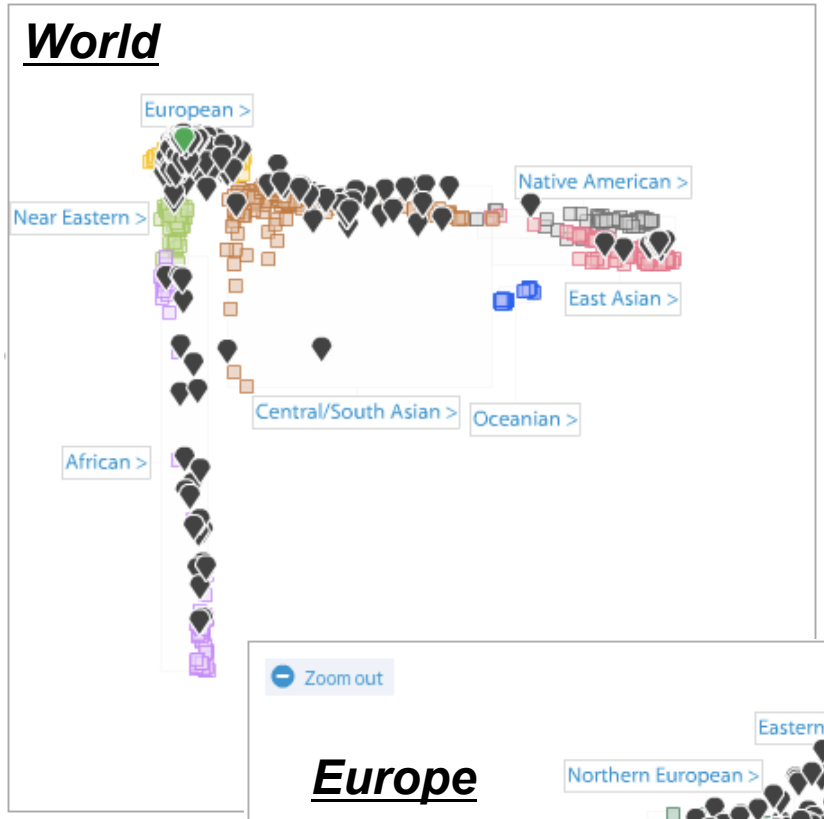


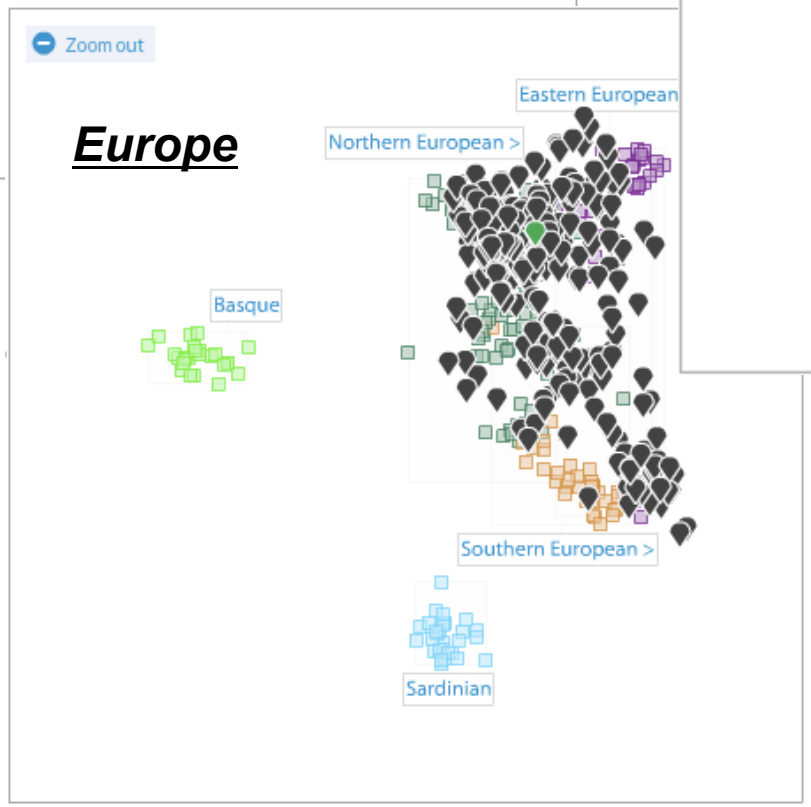
Select a person: Berber Woman

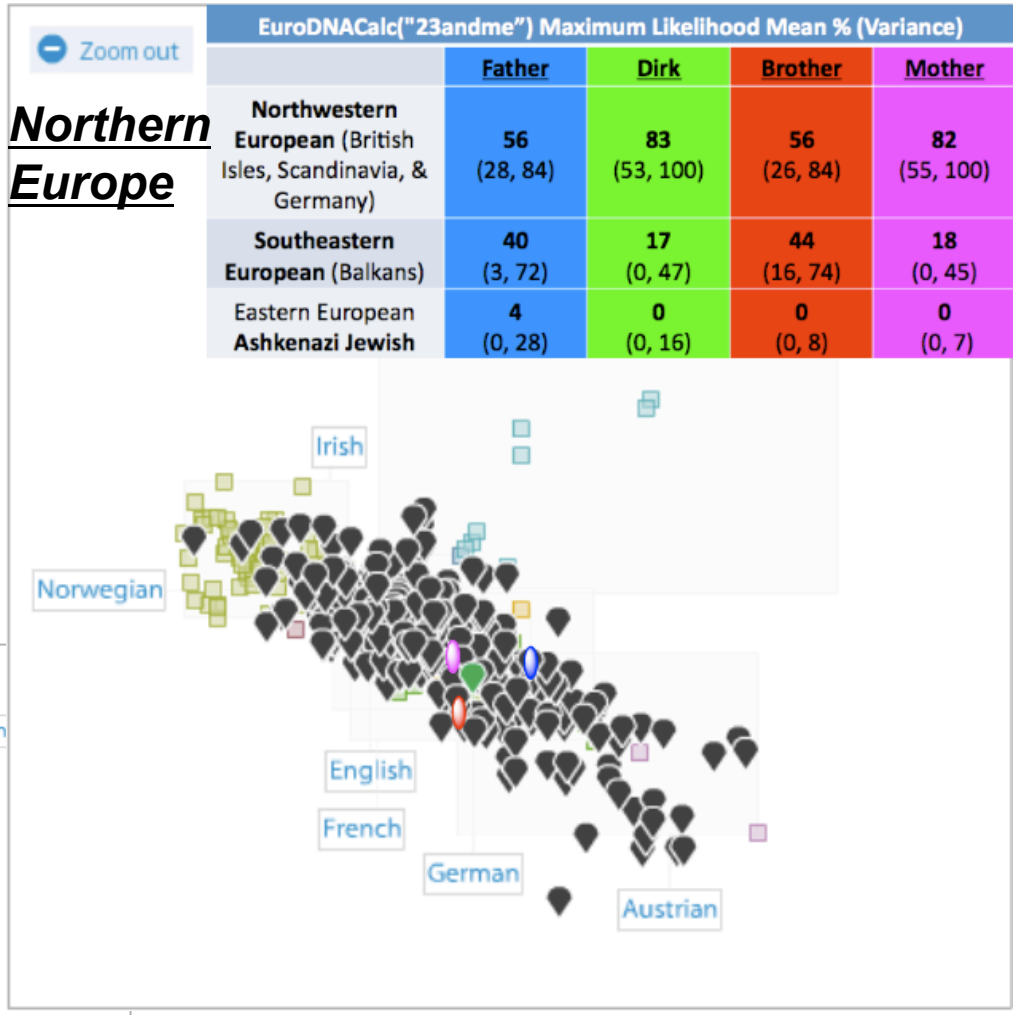


Select a person: Uyghur Woman



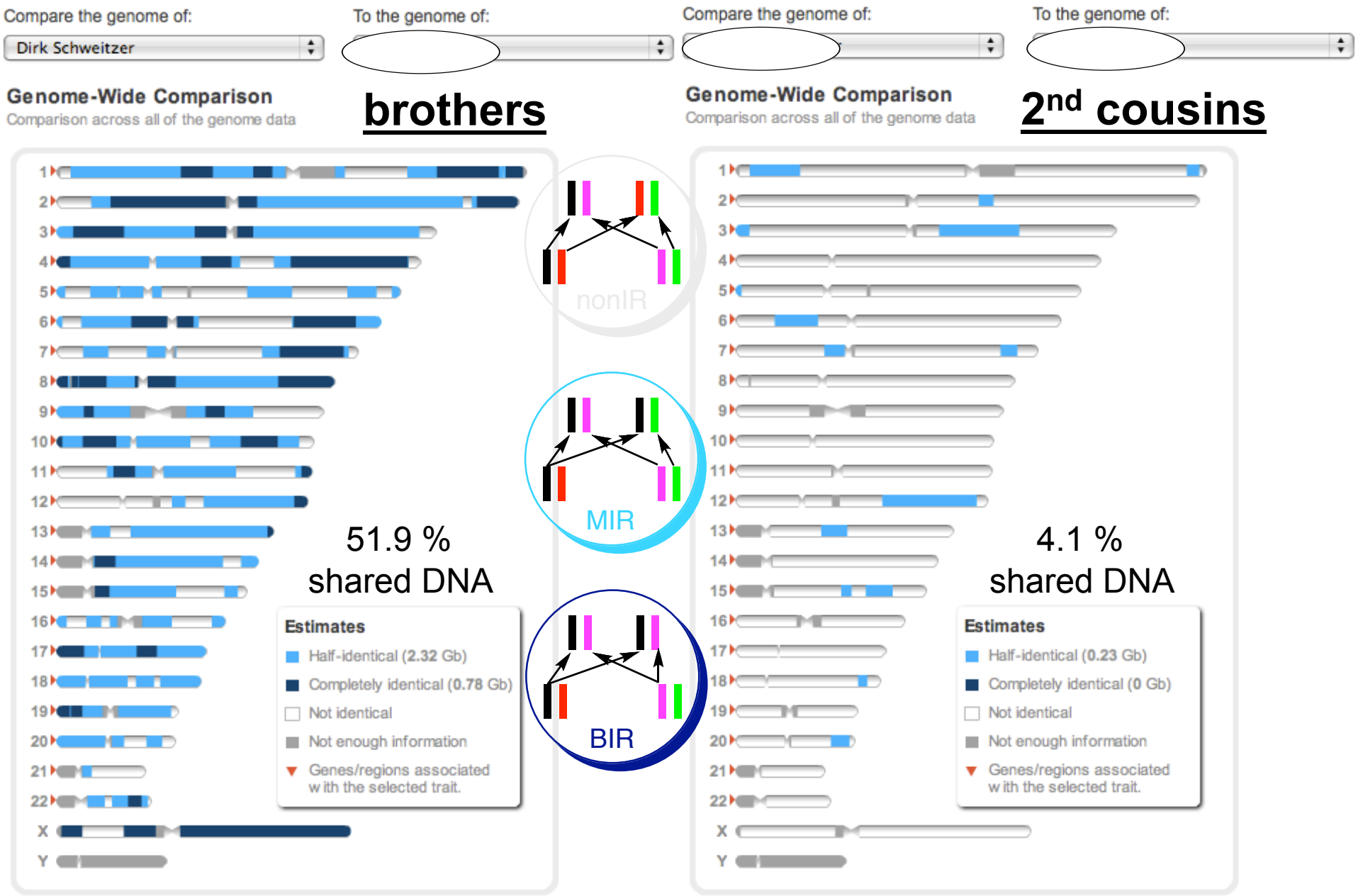






	EuroDNACalc("23andme") Maximum Likelihood Mean % (Variance)			
	Father	**Dirk**	**Brother**	**Mother**
Northwestern European (British Isles, Scandinavia, & Germany)	**56** (28, 84)	**83** (53, 100)	**56** (26, 84)	**82** (55, 100)
Southeastern European (Balkans)	**40** (3, 72)	**17** (0, 47)	**44** (16, 74)	**18** (0, 45)
Eastern European **Ashkenazi Jewish**	**4** (0, 28)	**0** (0, 16)	**0** (0, 8)	**0** (0, 7)

EuroDNACalc
Price *et al. PLoS Genetics*, **2008**, *4(1)*, e236
"We constructed a panel of 300 validated autosomal SNPs that are highly informative for distinguishing these ancestries."
http://dienekes.blogspot.com/2008/06/euro-dna-calc-11-released.html



Momochromosomal Identical Region (MIR)
Bichromosomal Identical Region (BIR)

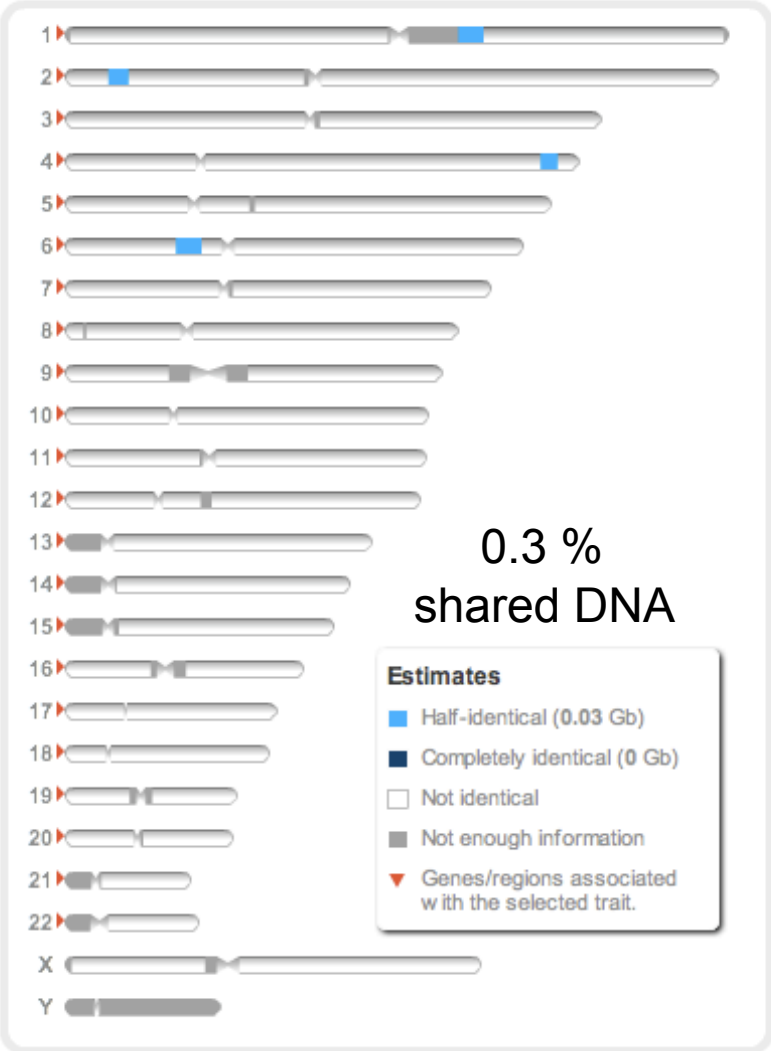
more distant relatives

Compare the genome of:

To the genome of:

Genome-Wide Comparison

Comparison across all of the genome data

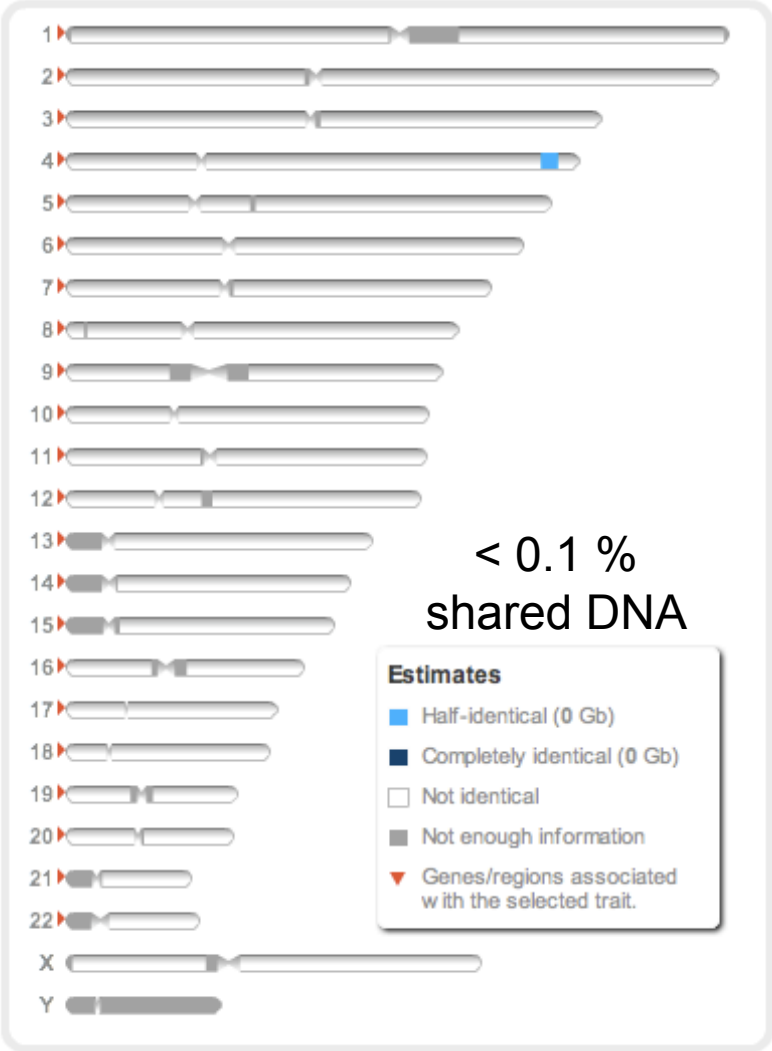


Compare the genome of:

To the genome of:

Genome-Wide Comparison

Comparison across all of the genome data

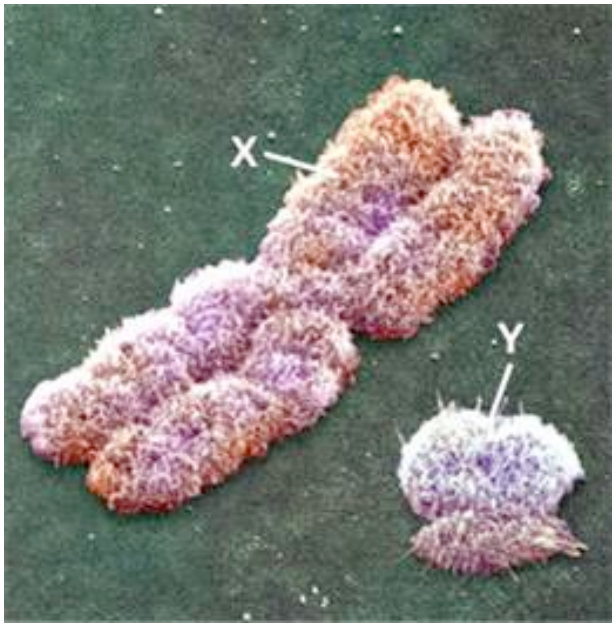


relationship to another person (# of generations to the common ancestor)	average % of shared/identical DNA expected (non-inbred population)
parent or sibling (1)	50
uncle / aunt / half-sibling (1)	25
cousin / half-uncle/aunt (2)	12.5
2nd cousin (3)	3.13
2nd cousin - once removed	1.56
3rd cousin (4)	0.78
4th cousin (5)	0.20
5th cousin (6)	0.05
6th and higher cousins	0.05, if shared/identical DNA inherited

Relatives whose common ancestor with one lived up to 5 generations ago are correctly identified.

6th and higher cousins likely do not share DNA, and are undistinguishable from a 5th cousin.

- discover one's biological identity, if one was adopted
- reconnect with close relatives, or their descendants, with whom contact was lost during recent historical events

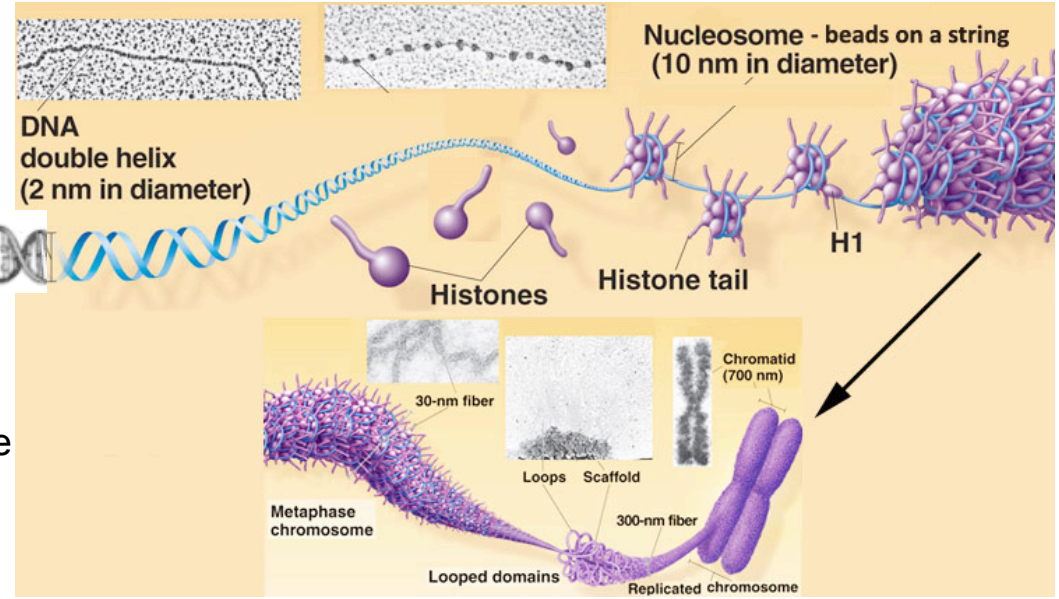


noncoding
antisense
template
transcribed

-

+

coding
sense
non-template
strand



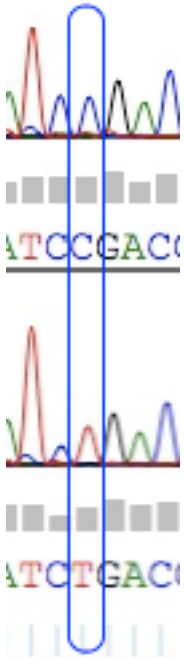
Y-chromosome = patrilinear descent

+ strand

mutated (ChrY:6919957, V22): C

SNP

=> "Haplogroup"



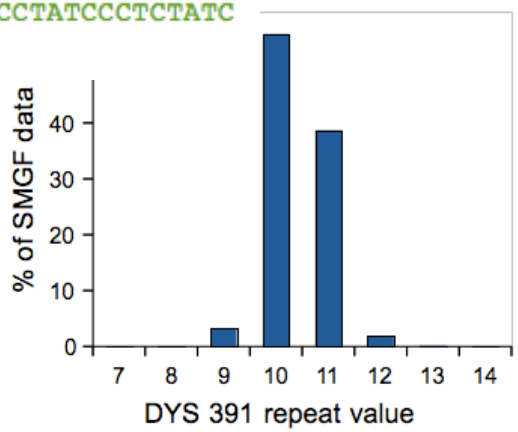
reference: T

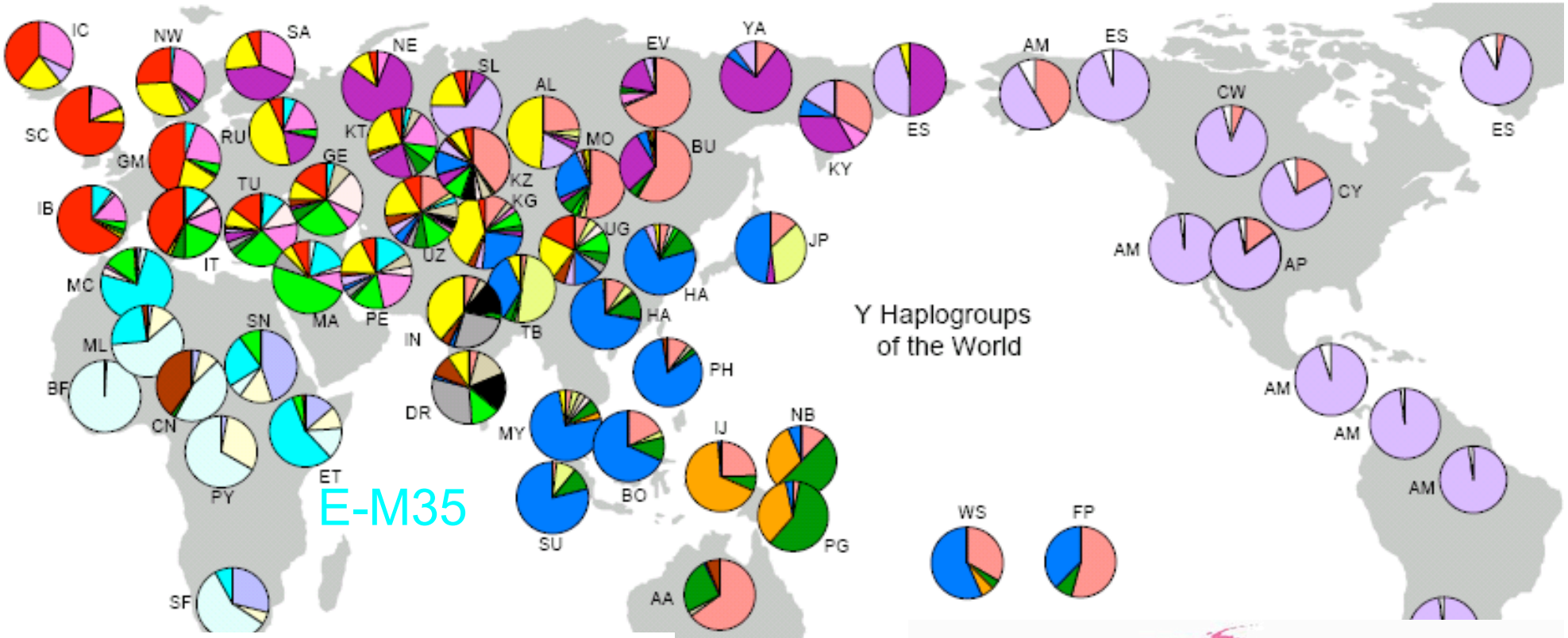
string/series of marker repeat values

=> "Haplotype"

DYS391 (CEPH 1358-01)
Example Sequence:
TTCATCATACACCCATATCTGTCTGTCTG/TCTA/TCTA/
TCTA/TCTA/TCTA/TCTA/TCTA/TCTA/TCTA/TCTA/
TCTA/TCTGCCTATCTGCCTGCCTACCTATCCCTCTATC

11 repeats 107 bp (TCTA)11




Y Haplogroups
of the World
E-M35
IC
NW
SA
NE
SL
EV
YA
AM
ES
ES
SC
GM
RU
KT
AL
MO
BU
KY
CW
CY
IB
TU
GE
KZ
KG
UG
JP
AM
AP
MC
IT
UZ
HA
HA
ML
SN
MA
PE
IN
TB
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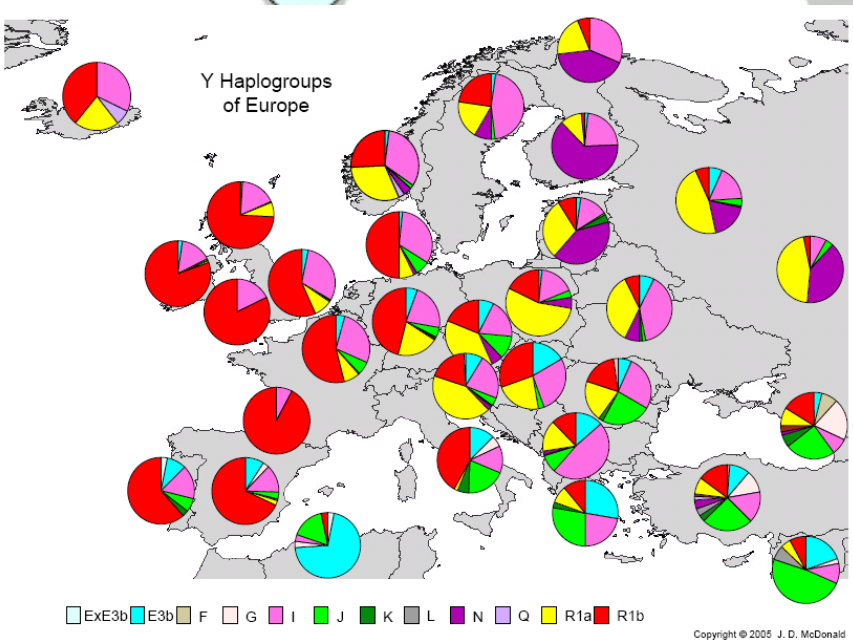
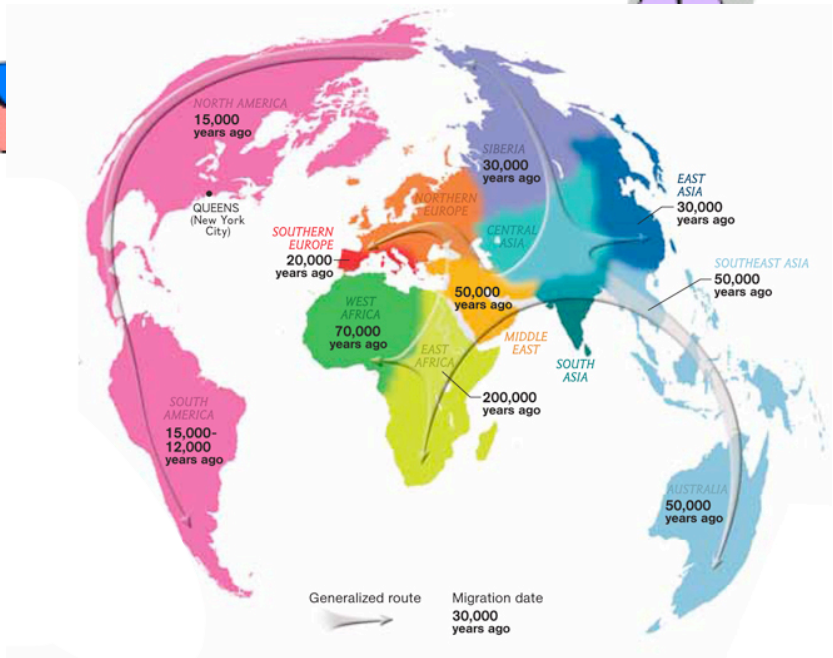
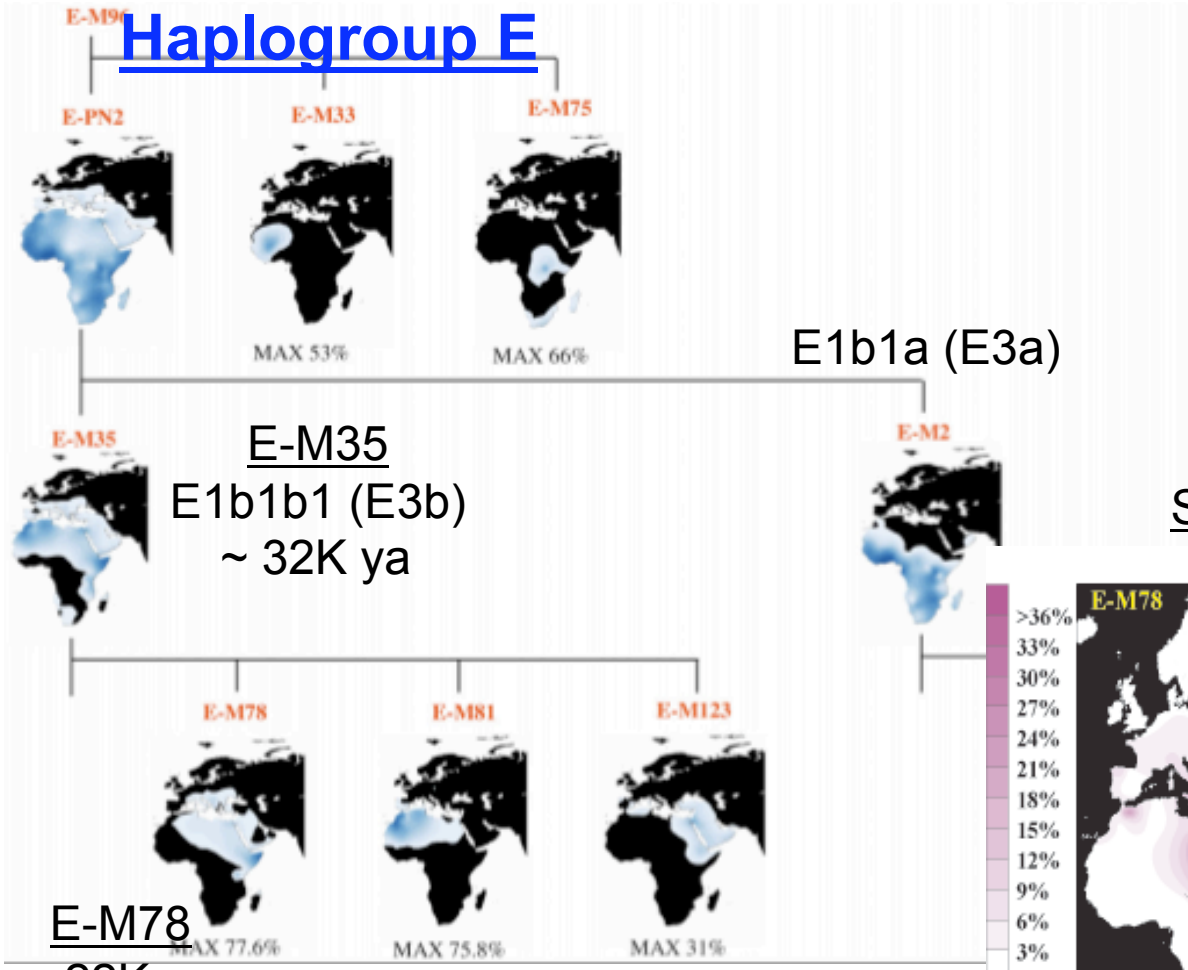
WS
FP
SF
AA

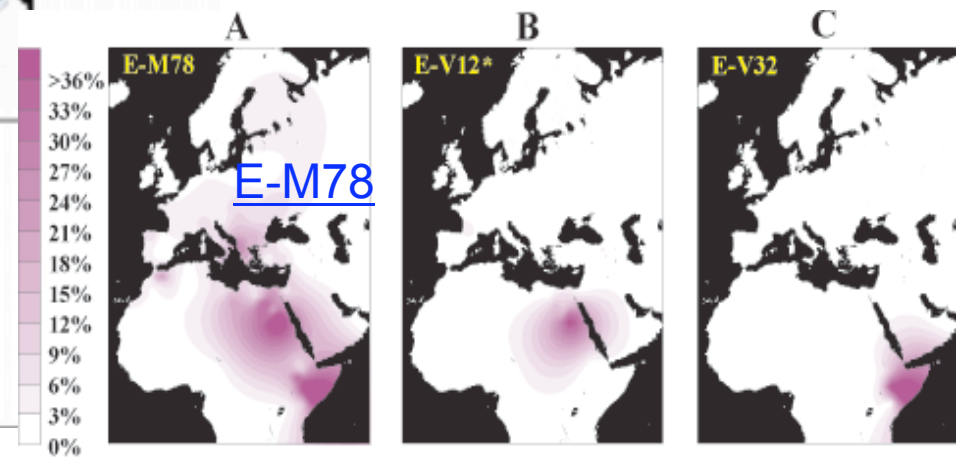

Y Haplogroups
of Europe
ExE3b
E3b
F
G
I
J
K
L
N
Q
R1a
R1b
Copyright © 2005 J. D. McDonald


NORTH AMERICA
15,000
years ago
QUEENS
(New York
City)
SIBERIA
30,000
years ago
EAST
ASIA
30,000
years ago
NORTHERN
EUROPE
CENTRAL
ASIA
SOUTHERN
EUROPE
20,000
years ago
SOUTHEAST ASIA
50,000
years ago
50,000
years ago
WEST
AFRICA
70,000
years ago
MIDDLE
EAST
EAST
AFRICA
SOUTH
ASIA
200,000
years ago
SOUTH
AMERICA
15,000-
12,000
years ago
AUSTRALIA
50,000
years ago
Generalized route
Migration date
30,000
years ago

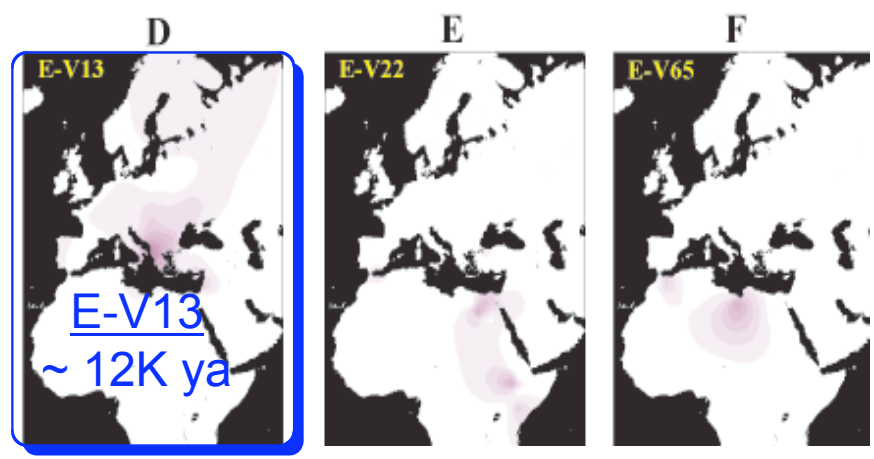
Haplogroup E



Subhaplogroups below E-M78

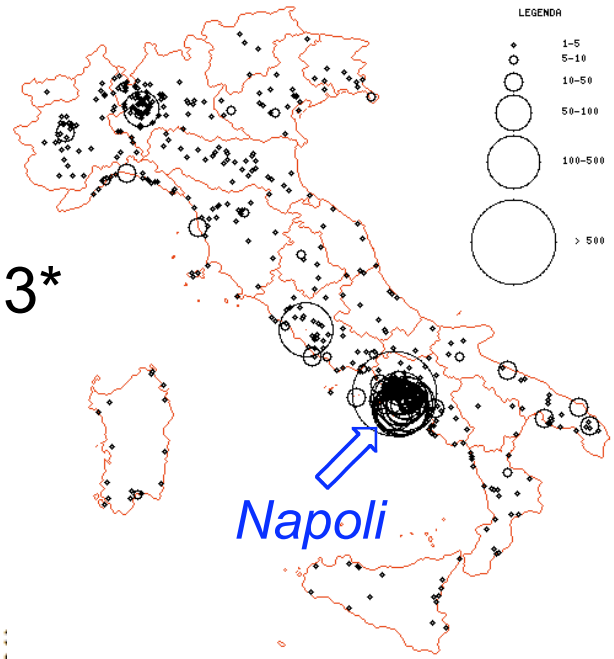


So far, 9 geographically more localized subhaplogroups below E-M35-M78-V13 are known. ====>



Gargiulo

Y-DNA = E-M35-M78-V13*

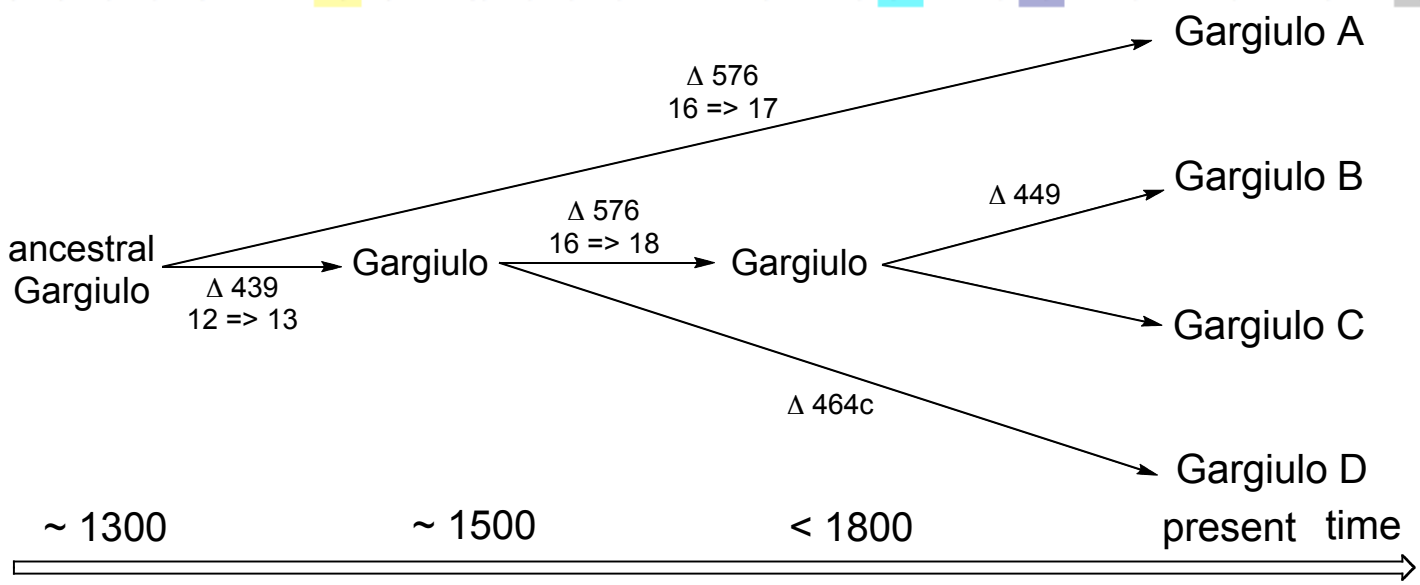


Haplotype

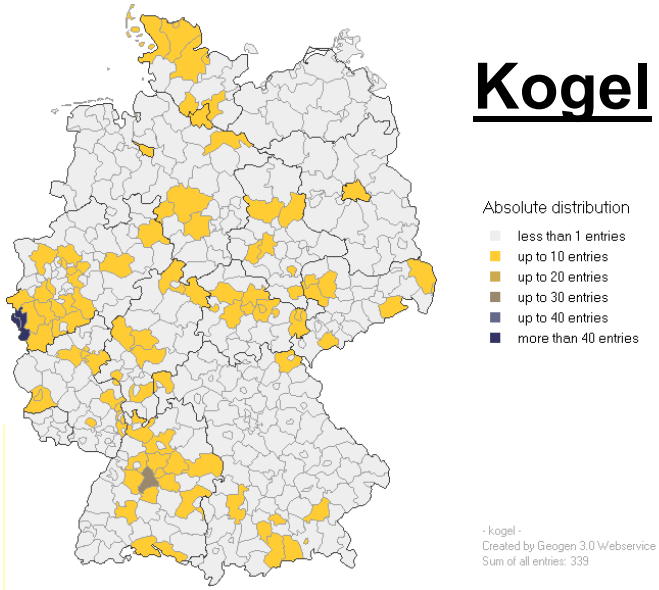
Cognome: GARGIULO
Presente in 543 comuni

Copyright 2000 LABO
http://www.labo.net/

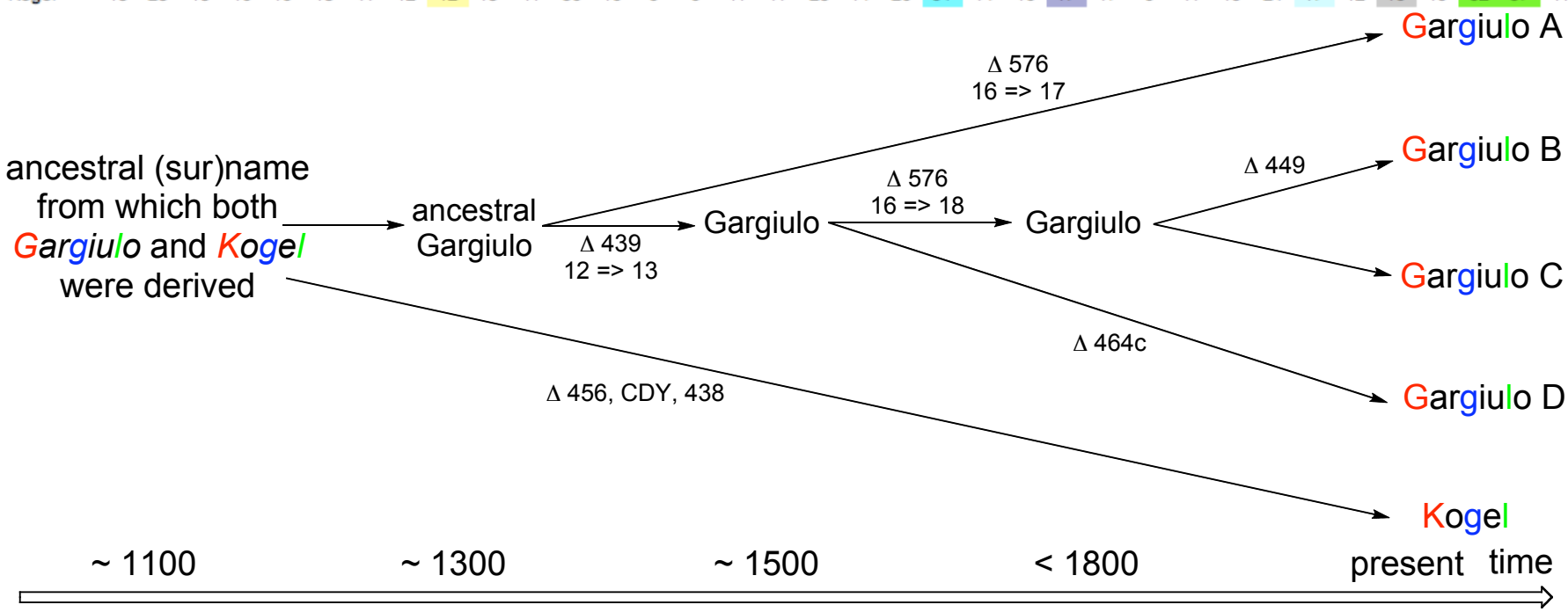
#	Family Name	393	390	19	391	385a	385b	426	388	439	389-1	392	2	8	a	b	5	4	7	7	8	9	a	b	c	464d	460	GATA H4	YCAII a	YCAII b	456	607	576	570	CDY a	CDY b	442	438
A	Gargiulo	13	23	13	10	16	18	11	12	12	13	11	30	16	9	9	11	11	25	14	20	31	14	16	17	17	9	11	19	21	15	12	17	19	30	34	11	10
B	Gargiulo	13	23	13	10	16	18	11	12	13	13	11	30	16	9	9	11	11	25	14	20	30	14	16	17	17	9	11	19	21	15	12	18	19	30	34	11	10
C	Gargiulo	13	23	13	10	16	18	11	12	13	13	11	30	16	9	9	11	11	25	14	20	31	14	16	17	17	9	11	19	21	15	12	18	19	30	34	11	10
D	Gargiulo	13	23	13	10	16	18	11	12	13	13	11	30	16	9	9	11	11	25	14	20	31	14	16	16	17	9	11	19	21	15	12	16	19	30	34	11	10



Kogel

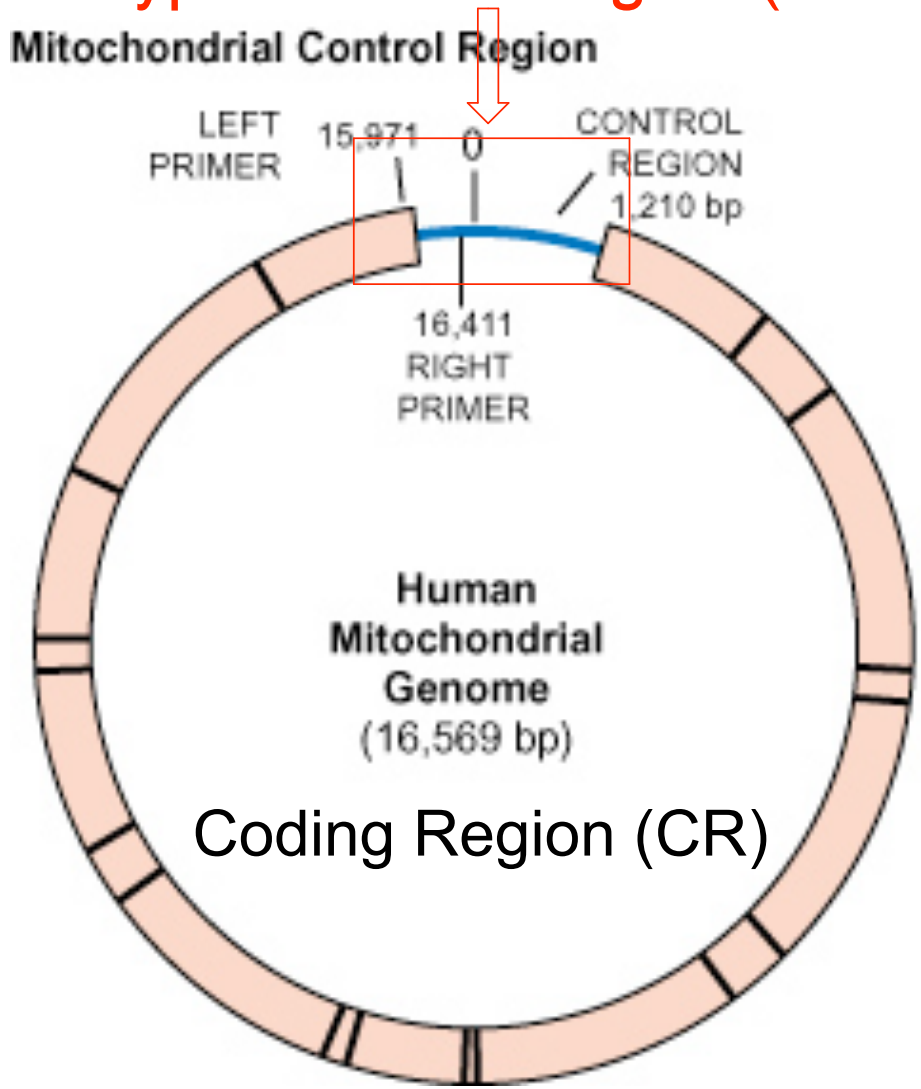


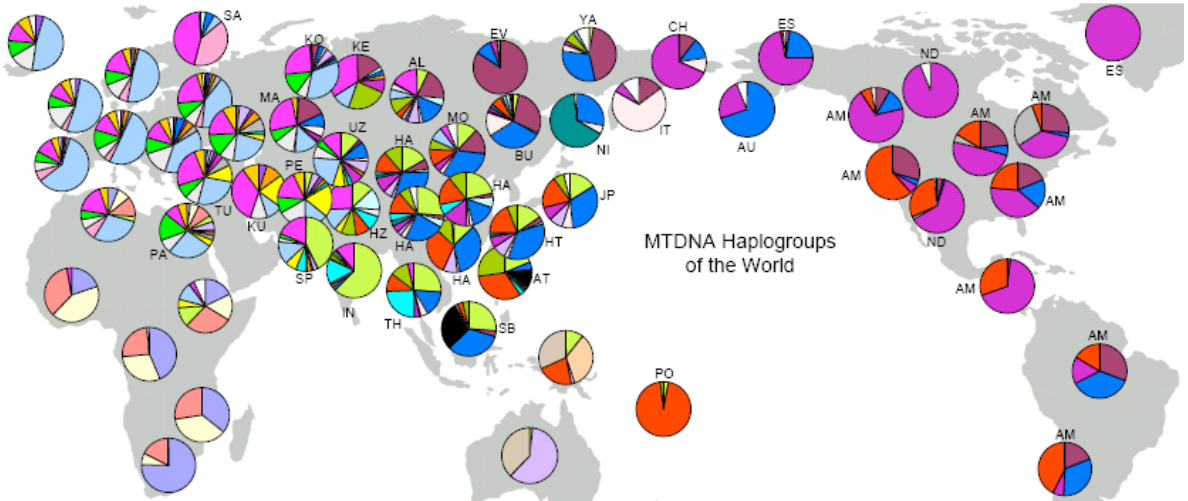
#	Family Name	393	390	19	391	385a	385b	426	388	9	1	2	2	8	a	D	5	4	7	7	8	9	a	D	c	d	450	GATA H4	YCAII a	YCAII b	456	607	576	570	CDY a	CDY b	442	438
A	Gargiulo	13	23	13	10	16	18	11	12	12	13	11	30	16	9	9	11	11	25	14	20	31	14	16	17	17	9	11	19	21	15	12	17	19	30	34	11	10
B	Gargiulo	13	23	13	10	16	18	11	12	13	13	11	30	16	9	9	11	11	25	14	20	30	14	16	17	17	9	11	19	21	15	12	18	19	30	34	11	10
C	Gargiulo	13	23	13	10	16	18	11	12	13	13	11	30	16	9	9	11	11	25	14	20	31	14	16	17	17	9	11	19	21	15	12	18	19	30	34	11	10
D	Gargiulo	13	23	13	10	16	18	11	12	13	13	11	30	16	9	9	11	11	25	14	20	31	14	16	16	17	9	11	19	21	15	12	16	19	30	34	11	10
	Kogel	13	23	13	10	16	18	11	12	12	13	11	30	16	9	9	11	11	25	14	20	31	14	16	17	17	9	11	19	21	17	12	16	19	32	37	11	11



mitochondrial DNA = matrilinear descent

Hypervariable Region (HVR)





AL	Altaians	KO	Komi
AT	Aboriginal Taiwanese	KU	Kurds
		MA	Mansi
AU	Aleuts	MO	Mongols
AM	Amerinds	ND	Na-Dene
BU	Buryats	NI	Nivkhs
CH	Chukchi	PA	Palestine+Egypt
ES	Eskimo	PE	Persians (Iran)
EV	Evenks	PO	Polynesians
HA	Han Chinese	SA	Saami
HT	Han Taiwanese	SB	Sabah (Borneo)
HZ	Hazara	SP	South Pakistan
IN	India	TH	Thailand
IT	Itelmen	TU	Turks
JP	Japanese	UZ	Uzbeks
KE	Kets	YA	Yakuts

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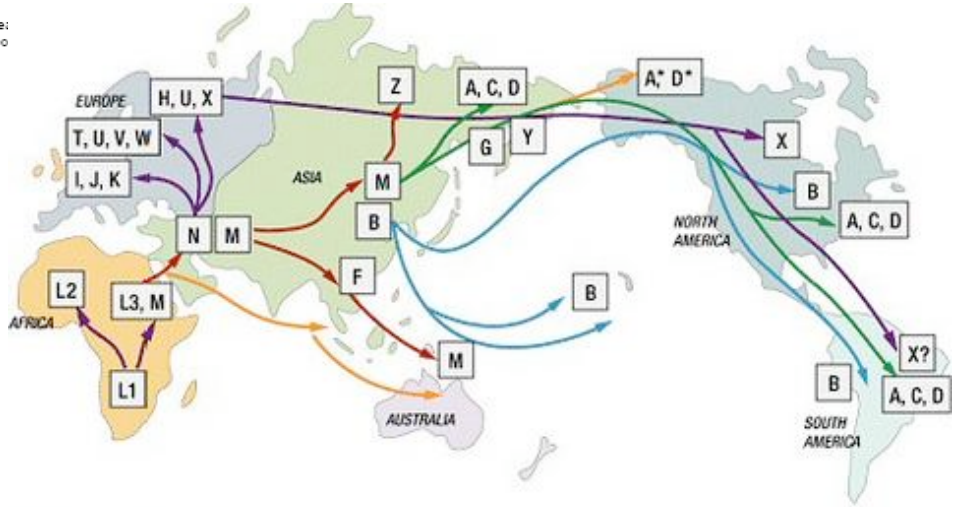
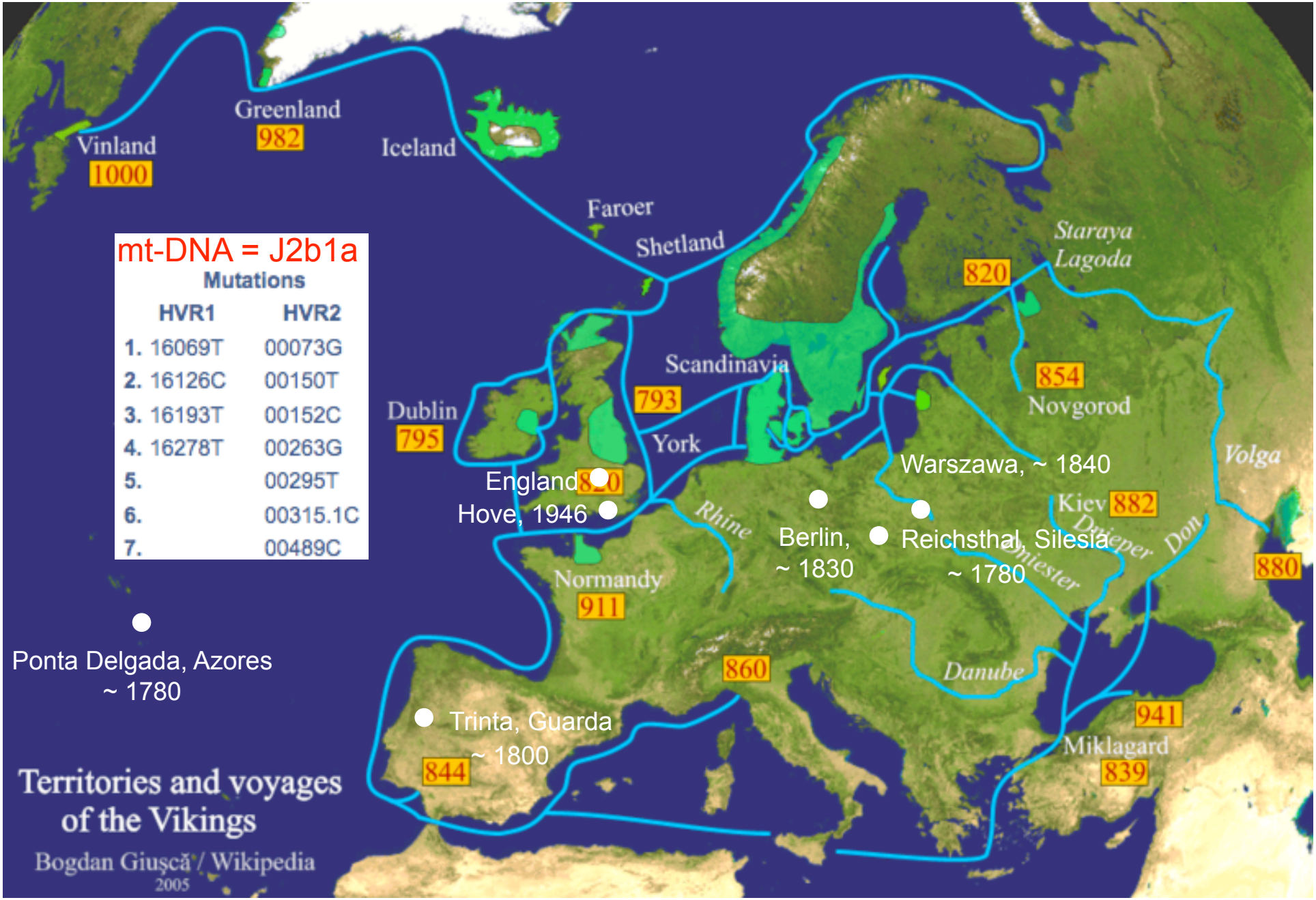
L1 L2 L3 M C Z D G E
Q N I W A X Y R B
F HV H V P J T U K Other

Specific tribes or locations are shown at left. Unlabelled pies are for general population in the are: African, American, and especially Polynesian areas are very large. The data in this chart is suppo to represent the situation before the recent European expansion beginning about 1500 AD. Assignments in Australia are somewhat iffy.



EXPANSION TIMES *(years ago)*	
Africa	120,000 - 150,000
Out of Africa	55,000 - 75,000
Asia	40,000 - 70,000
Australia/PNG	40,000 - 60,000
Europe	35,000 - 50,000
Americas	15,000 - 35,000
Na-Dene/Esk/Aleuts	8,000 - 10,000

FamilyTreeDNA
mtDNA Migrations Map



mt-DNA = J2b1a

Mutations

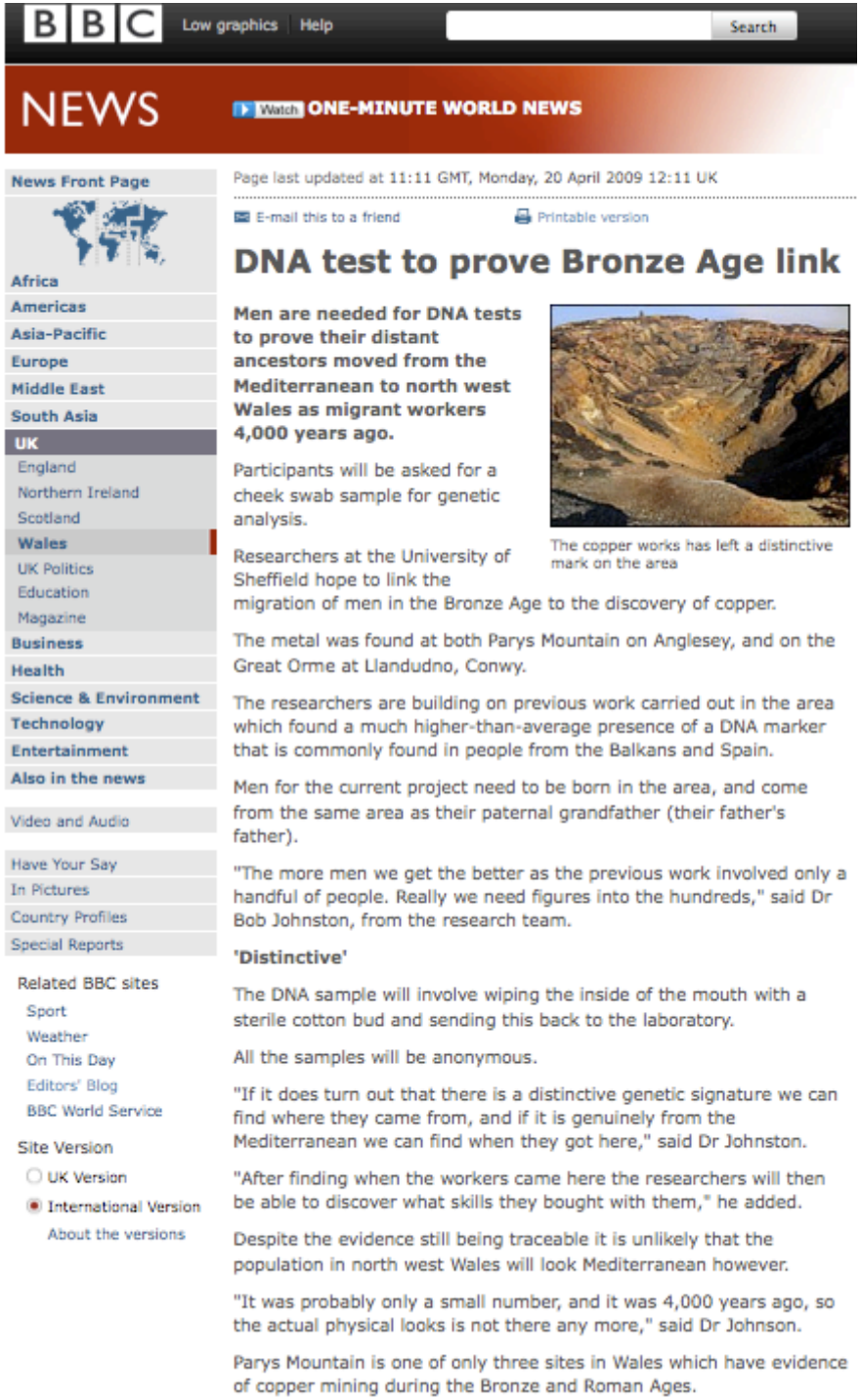
	HVR1	HVR2
1.	16069T	00073G
2.	16126C	00150T
3.	16193T	00152C
4.	16278T	00263G
5.		00295T
6.		00315.1C
7.		00489C

Be a part of cutting-edge research!

http://www.shef.ac.uk/archaeology/research/copper-mines/index.html

http://archiver.rootsweb.ancestry.com/th/read/GENEALOGY-DNA/2009-04/1240332305

*"We are following up on the Weale study (Mol. Biol. Evol. **2002**, 19(7) 1008-1021) which reported a much higher than average number of E3b individuals in Abergele. We are interested in the possibility that these may be linked to the Bronze age copper mines nearby, but obviously this is just one possibility. The first step is to see if we can replicate the findings of the 2002 study in a much larger sample."*



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DNA test to prove Bronze Age link

Men are needed for DNA tests to prove their distant ancestors moved from the Mediterranean to north west Wales as migrant workers 4,000 years ago.

Participants will be asked for a cheek swab sample for genetic analysis.

The copper works has left a distinctive mark on the area

Researchers at the University of Sheffield hope to link the migration of men in the Bronze Age to the discovery of copper.

The metal was found at both Parys Mountain on Anglesey, and on the Great Orme at Llandudno, Conwy.

The researchers are building on previous work carried out in the area which found a much higher-than-average presence of a DNA marker that is commonly found in people from the Balkans and Spain.

Men for the current project need to be born in the area, and come from the same area as their paternal grandfather (their father's father).

"The more men we get the better as the previous work involved only a handful of people. Really we need figures into the hundreds," said Dr Bob Johnston, from the research team.

'Distinctive'

The DNA sample will involve wiping the inside of the mouth with a sterile cotton bud and sending this back to the laboratory.

All the samples will be anonymous.

"If it does turn out that there is a distinctive genetic signature we can find where they came from, and if it is genuinely from the Mediterranean we can find when they got here," said Dr Johnston.

"After finding when the workers came here the researchers will then be able to discover what skills they bought with them," he added.

Despite the evidence still being traceable it is unlikely that the population in north west Wales will look Mediterranean however.

"It was probably only a small number, and it was 4,000 years ago, so the actual physical looks is not there any more," said Dr Johnson.

Parys Mountain is one of only three sites in Wales which have evidence of copper mining during the Bronze and Roman Ages.

Considerations:

- a family member may not be biologically related to you (social ancestry =/= biological ancestry)
- discover unexpected health issues
- errors in disease risk predictions (validity)
- unauthorized use of your data / data-security
- technical measurement errors = accuracy of the data

Organizations whose Test Results were shown:

THE GENOGRAPHIC PROJECT

https://genographic.nationalgeographic.com/

FamilyTreeDNA
HISTORY UNEARTHED DAILY

http://www.familytreedna.com/

23andMe

https://www.23andme.com/

Discussion of Results:

DNA-Forums
A Genetic Genealogy Community

http://dna-forums.com/