


Y-Chromosome and Mitochondrial DNA Analysis

Lineage Markers in Human Identity Testing

NEAFS 2006 Workshop
Rye Brook, NY
November 1, 2006



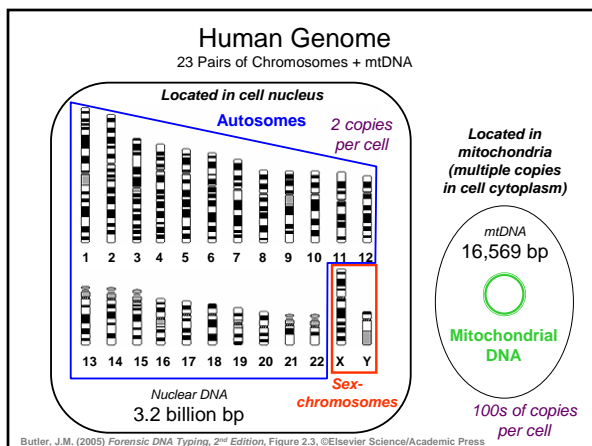
Northeastern Association
of
Forensic Scientists

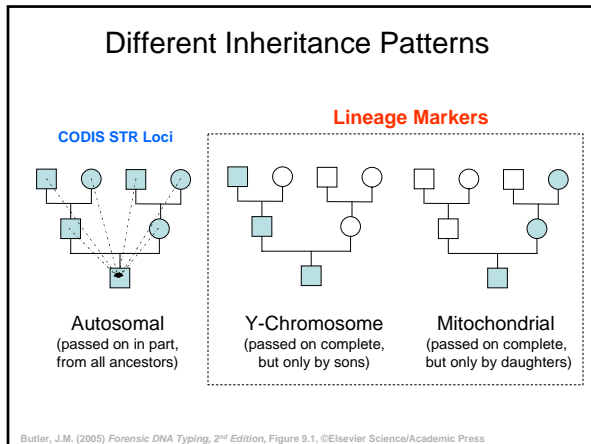
Dr. John M. Butler
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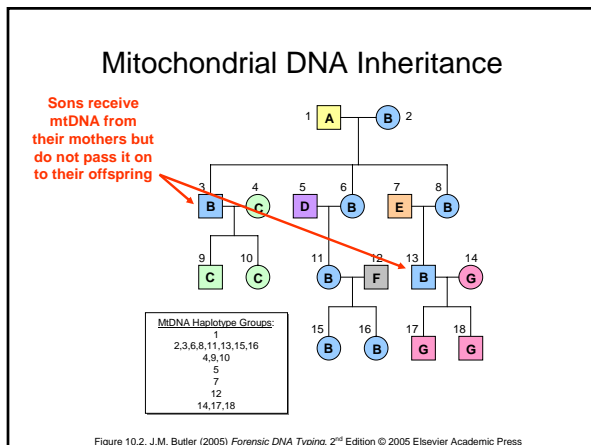
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Presentation Outline

- Autosomal STRs versus Y-STRs and mtDNA
- Advantages and disadvantages of lineage markers
- Work with lineage markers outside of the forensic arena (e.g., genetic genealogy)







Role of Y-STRs and mtDNA Compared to Autosomal STRs

- **Autosomal STRs provide a higher power of discrimination and are the preferred method whenever possible**
- **Due to capabilities for male-specific amplification**, Y-chromosome STRs (**Y-STRs**) can be useful in extreme female-male mixtures (e.g., when differential extraction is not possible such as fingernail scrapings)
- **Due to high copy number**, mitochondrial DNA (**mtDNA**) may be the only source of surviving DNA in highly degraded specimens or low quantity samples such as hair shafts

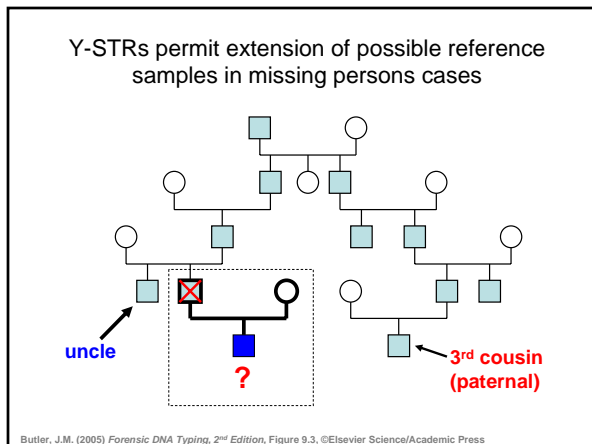
Value of Y-Chromosome Markers

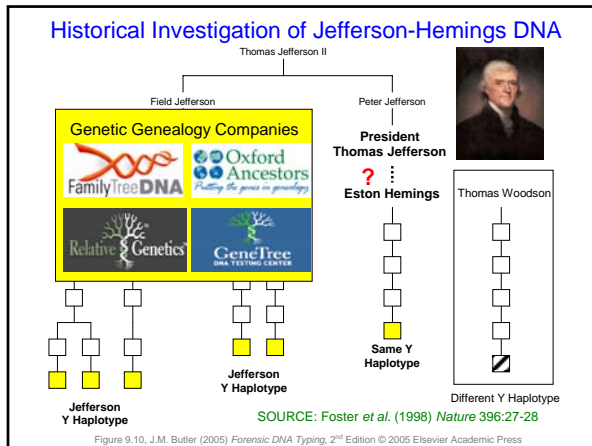
J.M. Butler (2005) *Forensic DNA Typing, 2nd Edition*; Table 9.1

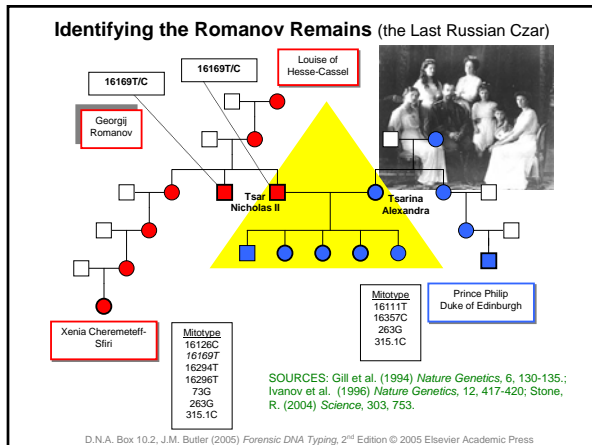
<u>Application</u>	<u>Advantage</u>
Forensic casework on sexual assault evidence	Male-specific amplification (can avoid differential extraction to separate sperm and epithelial cells)
Paternity testing	Male children can be tied to fathers in motherless paternity cases
Missing persons investigations	Patrilineal male relatives may be used for reference samples
Human migration and evolutionary studies	Lack of recombination enables comparison of male individuals separated by large periods of time
Historical and genealogical research	Surnames usually retained by males; can make links where paper trail is limited

Lineage Markers: Y-STRs and mtDNA

<u>Advantages</u>	<u>Disadvantages</u>
<ul style="list-style-type: none"> Extend possible reference samples beyond a single generation (benefits missing persons cases and genetic genealogy) Family members have indistinguishable haplotypes unless mutations have occurred 	<ul style="list-style-type: none"> Lower power of discrimination due to no genetic shuffling with recombination Family members have indistinguishable haplotypes unless mutations have occurred







Genetic Genealogy Companies

The rapidly growing field of genetic genealogy is expanding the number of Y-STR loci in use.

Y-STR Loci in Use for Genetic Genealogy

- **Family Tree DNA** (12, 37, or 67 loci) – DYS19, DYS385 a/b, DYS388, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS426, DYS438, DYS439, DYS447, DYS448, DYS449, DYS454, DYS455, DYS456, DYS459 a/b, DYS464 a/b/c/d, DYS436, DYS442, DYS460, GATA-H4, YCA II a/b, DYS456, DYS570, DYS576, DYS607, DYS724 a/b (CDY a/b), DYS395S1a/b, DYS406S1, DYS413 a/b, DYS425, DYS436, DYS444, DYS446, DYS450, DYS472, DYS481, DYS487, DYS490, DYS492, DYS511, DYS520, DYS531, DYS534, DYS537, DYS557, DYS565, DYS568, DYS572, DYS578, DYS590, DYS594, DYS617, DYS640, DYS641
- **Relative Genetics** (18, 26, or 43 loci) – DYS19, DYS385 a/b, DYS388, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS426, DYS438, DYS439, DYS447, DYS448, DYS460, YCAII a/b, GATA-H4, DYS393, DYS437, DYS454, DYS455, DYS461, DYS462, GGAAT1B07, GATA-A10, DYS635, DYS441, DYS442, DYS444, DYS445, DYS446, DYS449, DYS452, DYS456, DYS458, DYS459 a/b, DYS463, DYS464 a/b/c/d
- **Ethnoancestry** (18 loci) –DYS481, DYS487, DYS490, DYS494, DYS505, DYS522, DYS531, DYS533, DYS549, DYS556, DYS575, DYS578, DYS589, DYS594, DYS636, DYS638, DYS641, DYS406S1 + Y-SNPs
- **Oxford Ancestors** (10 loci) – DYS19, DYS388, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS425, DYS426
- **GeoGene** (6 loci) – DYS19, DYS388, DYS390, DYS391, DYS392, DYS393 + Y-SNPs

More Y-STRs Provides Greater Confidence in Matching Results Between Potential Relatives



Meaning of matches for same or similar surname



Probability for Most Recent Common Ancestor (MRCA)

Number of markers	50% probability that the MRCA was no longer than this number of generations	90% probability that the MRCA was no longer than this number of generations	95% probability that the MRCA was no longer than this number of generations
10 of 10	16.5	56	72
11 of 12	17	39	47
12 of 12	7	23	29
23 of 25	11	23	27
24 of 25	7	16	20
25 of 25	3	10	13
35 of 37	6	12	14
36 of 37	4	9	10
37 of 37	2	5	7



FamilyTree DNA
<http://www.familytreedna.com>

- Y-STR typing and mtDNA sequencing performed in Mike Hammer's lab at the University of Arizona

9/26/2006: Family Tree DNA announced that their DNA databases contain **111,139** records: **3547** SURNAME PROJECTS, **55,610** unique surnames, **77,543** Y-DNA records in the database, **20,531** distinct 12-marker haplotypes, **25,921** distinct 25-marker haplotypes, **19,840** distinct 37-marker haplotypes, **33,596** mtDNA records in the database

10/24/2006: Family Tree DNA hits new milestones solidifying its leadership with the largest DNA databases in the field of Genetic Genealogy with **114,856** records: **3608** SURNAME PROJECTS, **56,441** unique surnames, **79,970** Y-DNA records in the database, **20,929** distinct 12-marker haplotypes, **26,577** distinct 25-marker haplotypes, **20,460** distinct 37-marker haplotypes, **34,886** mtDNA records in the database

In approximately one month, 3,717 new records were added including 1,290 mtDNA, 398 12-marker, 656 25-marker, 620 37-marker Y-STR haplotypes

www.Ysearch.org

Address: http://www.ysearch.org/

Navigation: CREATE A NEW USER, EDIT AN EXISTING USER, ALPHABETICAL LIST OF LAST NAMES, SEARCH BY LAST NAME, SEARCH FOR GENETIC MATCHES, SEARCH BY HAPLOGROUP, RESEARCH TOOLS, STATISTICS

Welcome

Much has happened since Y-DNA testing first became available commercially through FamilyTreeDNA in February of 2000. Many thousands of people have tested to find family connections as well as family origins. Since then, other labs have entered the market, and the number of tested individuals is growing as the use of DNA is becoming more and more accepted as an important tool for family research, enhancing traditional genealogy research methods.

In order to allow people that have tested with the different companies to make their results available for comparison, FamilyTreeDNA is offering Ysearch as a free public service. We have added several tools that allow you to compare side-by-side different users - the [YsearchCompare](#) - as well as generate a [Genetic Distance™ Report](#) and many other features, including the upload of GEDCOM files.

GEDCOM Update (what is GEDCOM?):
If you've already created an account, please upload your GEDCOM

What next?	Size of the database?
<ul style="list-style-type: none"> • Create a new user • Search for genetic matches • Search by last name • Edit an existing user 	<ul style="list-style-type: none"> • Surname Count: 18665 • Unique Haplotypes: 25,254 • Number of Records: 33471 • FamilyTreeDNA - 30641 • Relative Genetics - 643 • Other Services - 195 © 2005 - 2006

www.mitoSearch.org

Address: http://www.mitoSearch.org/

Navigation: Create A New User, Edit An Existing User, Search For Genetic Matches, Search By Haplogroup, Research Tools, Statistics

Welcome

With thousands of people that have tested their mtDNA with different companies, MitoSearch.org is brought to you as a free public service that allows individuals that have tested with those companies to make their results available for comparison.

We have added tools that allow you to compare side-by-side different users - the [mitoSearchCompare](#) as well as many other features.

We hope you enjoy the use of mitoSearch. Should you have any questions or suggestions, please do not hesitate to [contact us](#).

What next?


- Create a new user
- Search for genetic matches
- Ysearch Users - Create a mitoSearch account

Home Create a New User Edit an Existing User Search for Genetic Matches Search by Haplogroup Research Tools Statistics
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Genetic Genealogy

http://www.isogg.org

"The mission of the International Society of Genetic Genealogy is to advocate for and educate about the use of genetics as a tool for genealogical research, and promote a supportive network for genetic genealogists."



Famous DNA

<http://www.isogg.org/famousdna.htm>

Jesse James
 In 1995, the infamous outlaw, Jesse James' body was exhumed for DNA testing. Samples of hair and other fragments from his first burial site at his home were also recovered and sampled, along with mtDNA samples extracted from two living James' relatives. All samples resulted in a perfect match, thus concluding that the body in Mt. Olivet Cemetery, is indeed that of Jesse James.

Name	mtDNA Haplogroup	mtDNA Sequence
Jesse James	T2	16126C, 16274A, 16294T, 16296T, 16304C

Jesse James mtDNA Results

Stone et al. (2001) J. Forensic Sci. 46(1):173-176

LAST WORD SOCIETY
 Anne C. Stone,¹ Ph.D.; James E. Starrs,² LL.M.; and Mark Stenking,¹ Ph.D.

Mitochondrial DNA Analysis of the Presumptive Remains of Jesse James*

Nucleotide Position	1	11	111	1111
Sample	6	4	4	4

Reference	T	G	C	T
C	C	A	T	T
F	C	A	T	T
H-1	C	A	T	T
H-2	C	A	T	T
RJ	C	A	T	T
MN	C	A	T	T

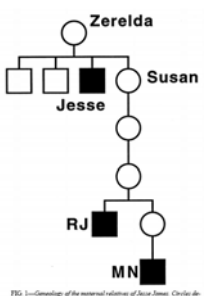



FIG. 3.—Genealogy of the maternal relatives of Jesse James. Circle denotes female and square denotes James male. RJ is a great-grandson and MN is a great-great-grandson of Jesse's sister Susan, and they both are expected to have the same mtDNA sequence as Jesse James.

DNA Results from Some Famous People

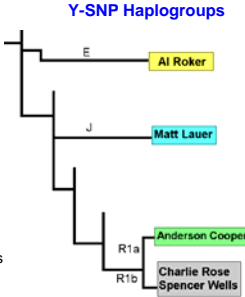


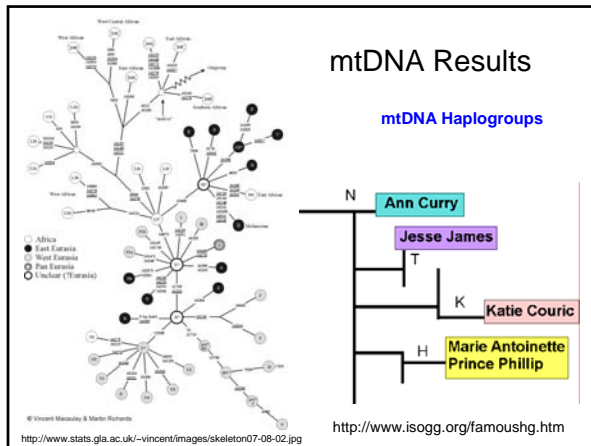
Famous Haplogroups

<http://www.isogg.org/famoushg.htm>

The following haplogroups for the hosts of the "Today Show" were aired 18 Nov 2005 during an interview with Dr. Spencer Wells. The Y-haplogroups for Dr. Wells and PBS host, Charlie Rose, were aired on the 23 Jan 2006 segment of the "Charlie Rose Show". Anderson Cooper's haplogroup was referenced on 21 Feb 2006 "Anderson Cooper 360" segment.

<http://msnbc.msn.com/id/10095659/>





Ancient DNA

<http://www.isogg.org/ancientdna.htm>

Cheddar Man

In 1903, skeletal remains were found in a cave in Cheddar, England. The remains of a 23 year-old man, who was killed by a blow to the face, were discovered to be at least 9,000 years old. Ninety-four years after the discovery of "Cheddar Man", scientists were able to extract mitochondrial DNA from his tooth cavity.

Bryan Sykes, and his team at Oxford University distributed DNA test kits to local Cheddar schools, and a match was found to a local schoolteacher, Adrian Targett.

Name	mtDNA Haplogroup	mtDNA Sequence
Cheddar Man	U5a	16192T, 16270T

The New York Times
nytimes.com

March 24, 1997

Tracing Your Family Tree to Cheddar Man's Mum
By SARAH LYALL

Until several weeks ago, Adrian Targett, a high school history teacher, didn't appear to have much in common with Cheddar Man, a 9,000-year-old pile of bones at the Natural History Museum in London.

Sure, Mr. Targett had heard of Cheddar Man, and had even visited the cave in this quaint Somerset village where his skeleton was found in 1903. But after a seemingly quixotic experiment in which scientists compared Cheddar Man's DNA to that of 20 local residents, Mr. Targett recently received a wholly unexpected piece of news: He is, it seems, related to Cheddar Man on his mother's side.

"I'm thinking of writing to the Marquess of Bath, who owns these caves, and saying, 'I'd like my cave back,'" Mr. Targett, 42, said over a meat pie and a pint in the local pub recently, considering the implications of having such a venerable relative. "All those times I'd visited this cave before, and I'd never realized I was going home."

<http://query.nytimes.com/gst/fullpage.html?res=9807EED8133BF937A15750CDA961958260&sec=health&pagewanted=print>

Genghis Khan's Y-Chromosome Profile

Zerjal et al. (2003) *Am. J. Hum. Genet.* 72:717-721

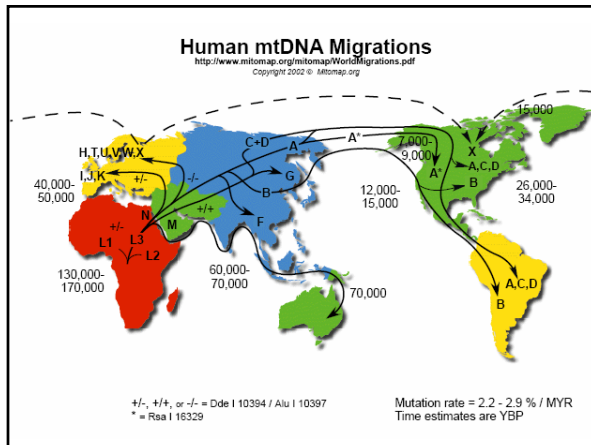
The Genetic Legacy of the Mongols

Tatiana Zerjal,¹ Yali Xue,^{1,2} Giorgio Bertorelle,³ R. Spencer Wells,⁴ Weidong Bao,^{1,5} Sufang Zhu,^{1,6} Raheel Qamar,^{1,6} Qasim Ayub,^{1,6} Aisha Mohyuddin,^{1,6} Songbin Fu,⁷ Pu Li,² Nadira Yuldasheva,^{4,7} Ruslan Ruzibakiev,⁷ Jiujun Xu,⁷ Qunfang Shu,⁷ Ruofu Du,⁷ Huanming Yang,² Matthew E. Hurles,⁸ Elizabeth Robinson,^{1,2} Tudevagva Gerelsaikhan,^{1,4} Bumbein Dashnyam,⁹ S. Qasim Mehdi,² and Chris Tyler-Smith¹

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DYS 393	DYS 390	DYS 19/394	DYS 391	DYS 385a	DYS 385b	DYS 426	DYS 388	DYS 439	DYS 389.1
13	25	16	10	12	13	11	14	10	13
DYS 392	DYS 389.2	DYS 458	DYS 459a	DYS 459b	DYS 455	DYS 454	DYS 447	DYS 437	DYS 448
11	29	18	8	8	11	12	26	14	22
DYS 449	DYS 464a	DYS 464b	DYS 464c	DYS 464d					
27	11	11	12	16					

Haplogroup: C3
Last name: Genghis Khan deduced Y-DNA Profile <http://www.ysearch.org>



The Genographic Project

<https://www3.nationalgeographic.com/genographic/>

- Funded \$50 million for 5 years by IBM and National Geographic
- Will gather and run DNA samples from ~100,000 people around the world with Y-SNPs and mtDNA
- For U.S. participants, Mike Hammer's lab is running 12 Y-STRs or sequencing mtDNA HV1

Perhaps the Real Reason Some Genetic Genealogy Is Performed...



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E-mail: randy@glasbergen.com

"You don't look anything like the long haired, skinny kid I married 25 years ago. I need a DNA sample to make sure it's still you."
