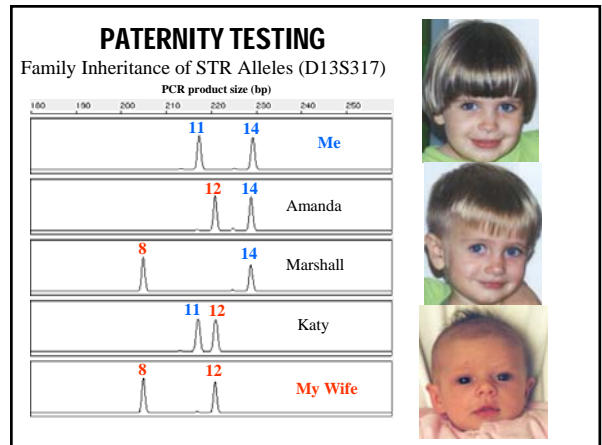
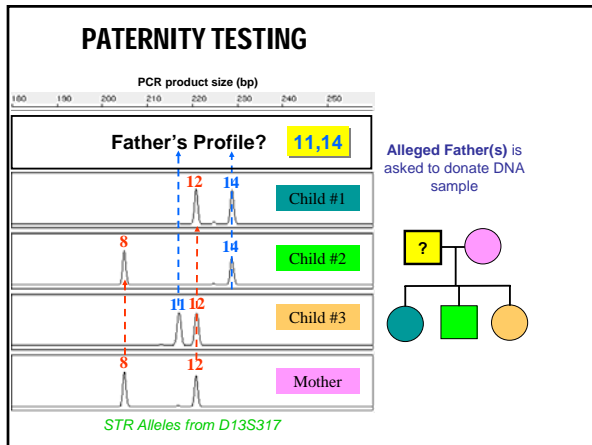

Other DNA Applications
 John M. Butler, Ph.D.
 National Institute of Standards and Technology
 Frankenmuth, MI
 May 4, 2007

Applications of Human Identity Testing

- Forensic cases -- **matching suspect with evidence**
- Paternity testing -- **identifying father**
- Missing persons investigations
- Military DNA “dog tag”
- Convicted felon DNA databases
- Mass disasters -- **putting pieces back together**
- Historical investigations and genetic genealogy

Involves generation of DNA profiles usually with the same core STR (short tandem repeat) markers and then MATCHING TO REFERENCE SAMPLE




Tsunami Survivor “Baby 81” Connected to His Parents with DNA

Wednesday, March 2, 2005 Posted: 9:27 AM EST (1427 GMT)

NEW YORK (AP) -- The parents of the infant tsunami survivor nicknamed “Baby 81” say they found it difficult to feel overjoyed about their reunion in the midst of so much tragedy.

The 4-month-old Sri Lankan baby and his parents, who were reunited after court-ordered [DNA tests proved their relationship](#), appeared on ABC’s “Good Morning America” Wednesday, a day after their 20-hour-long flight landed in New York.

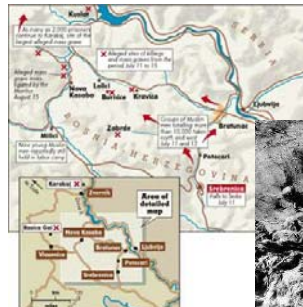

“Baby 81,” parents make TV appearance



<http://www.cnn.com/2005/US/03/02/baby.81.ap/index.html>

Identification of Remains from Former Yugoslavia

>90,000 family reference samples collected
>17,000 bones identified as of April 2007

DNA testing is performed on 100s of bones collected each week from mass graves in Bosnia and Croatia to help in the re-association of remains

Tomb of the Unknown Soldier



DNA History

- Armed Forces DNA Identification Laboratory (AFDIL) (Rockville, MD)
- In June 1998 AFDIL identified Michael J. Blassie as the Vietnam Unknown in the Tomb of the Unknown Soldier (located in Arlington National Cemetery)
- There will be no more "unknown" soldiers.

Butler, J.M. (2005) *Forensic DNA Typing, 2nd Edition*, Box 10.1, pp. 250-251

Armed Forces DNA Repository



>5 million bloodstain cards on file from members of U.S. military

Are being used to identify remains from combat casualties

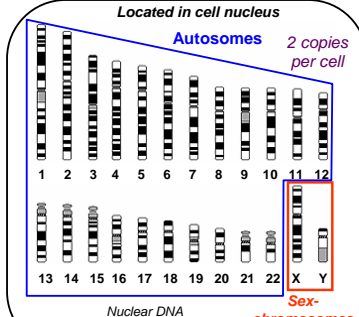
Located in Gaithersburg, Maryland

Human Genome

23 Pairs of Chromosomes + mtDNA

Located in cell nucleus


Autosomes 2 copies per cell



Nuclear DNA 3.2 billion bp

Sex-chromosomes X Y

Located in mitochondria (multiple copies in cell cytoplasm)



mtDNA 16,569 bp

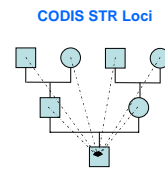
Mitochondrial DNA

100s of copies per cell

Butler, J.M. (2005) *Forensic DNA Typing, 2nd Edition*, Figure 2.3, ©Elsevier Science/Academic Press

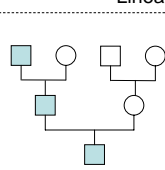
Different Inheritance Patterns

CODIS STR Loci

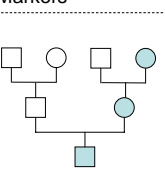


Autosomal
(passed on in part, from all ancestors)

Lineage Markers




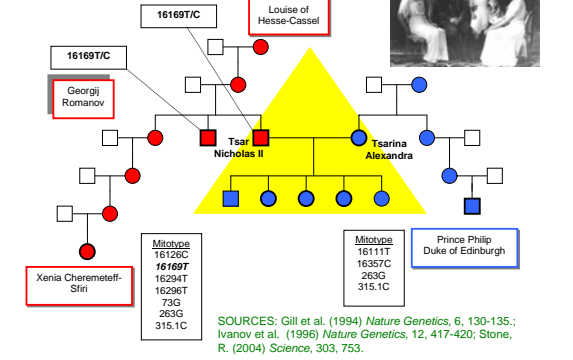
Y-Chromosome
(passed on complete, but only by sons)



Mitochondrial
(passed on complete, but only by daughters)

Butler, J.M. (2005) *Forensic DNA Typing, 2nd Edition*, Figure 9.1, ©Elsevier Science/Academic Press

The Romanovs (Last Russian Czar)

16169T/C

16169T/C

16169T/C

Mitotype
16126C
16169T
16294T
16296T
73G
263G
315.1C

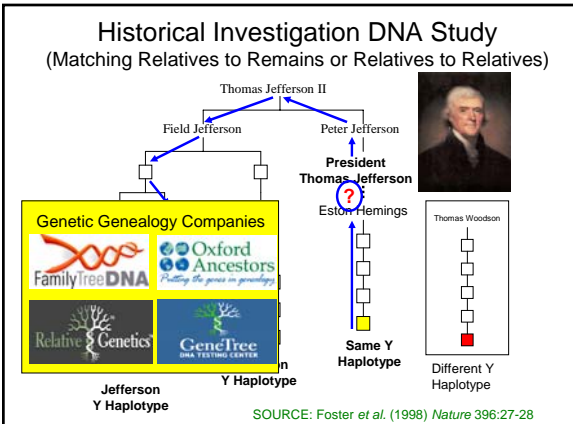
Mitotype
16111T
16357C
263G
315.1C

SOURCES: Gill et al. (1994) *Nature Genetics*, 6, 130-135.; Ivanov et al. (1996) *Nature Genetics*, 12, 417-420; Stone, R. (2004) *Science*, 303, 753.

D.N.A. Box 10.2, J.M. Butler (2005) *Forensic DNA Typing, 2nd Edition* © 2005 Elsevier Academic Press

Historical Investigation DNA Study

(Matching Relatives to Remains or Relatives to Relatives)



Thomas Jefferson II

Field Jefferson

Peter Jefferson

President Thomas Jefferson

Eston Hemings

Thomas Woodson

Genetic Genealogy Companies

- FamilyTreeDNA
- Oxford Ancestors
- RelativeGenetics
- GeneTree

Jefferson Y Haplotype

Same Y Haplotype

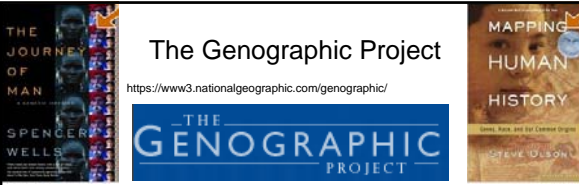
Different Y Haplotype

SOURCE: Foster et al. (1998) *Nature* 396:27-28

Butler, J.M. (2001) *Forensic DNA Typing, Figure 17.4*, ©Academic Press


The Genographic Project

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



- Different populations carry distinct markers. Following them through the generations reveals a genetic tree on which today's many diverse branches may be followed ever backward to their common African root
- Our genes allow us to chart the ancient human migrations from Africa across the continents
- 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

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

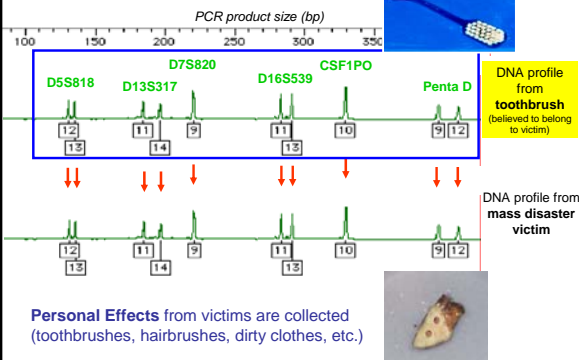


World's first genealogy driven DNA testing company
FamilyTreeDNA

© 1997 Randy Glasbergen, 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."

Using Personal Effects to Identify Remains



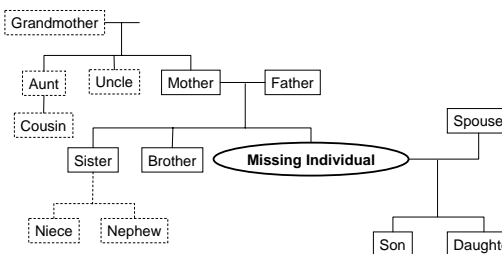
PCR product size (bp)

DNA profile from toothbrush (believed to belong to victim)

DNA profile from mass disaster victim

Personal Effects from victims are collected (toothbrushes, hairbrushes, dirty clothes, etc.)

Kinship DNA Testing

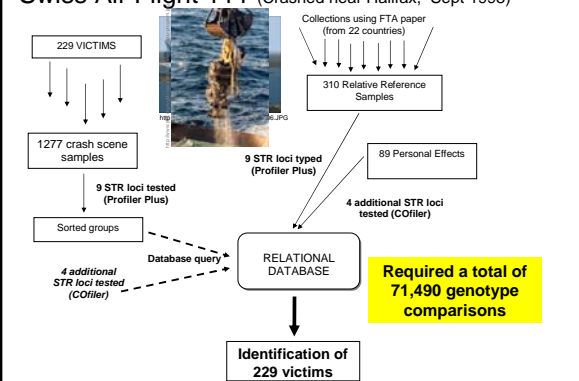


Grandmother, Aunt, Uncle, Mother, Father, Cousin, Sister, Brother, Missing Individual, Spouse, Niece, Nephew, Son, Daughter

Samples that would be valuable for the maternally transmitted mitochondrial DNA are indicated in dashed boxes.

Figure 24.2, J.M. Butler (2005) Forensic DNA Typing, 2nd Edition © 2005 Elsevier Academic Press

Swiss Air Flight 111 (Crashed near Halifax, Sept 1998)



229 VICTIMS

1277 crash scene samples

9 STR loci tested (Profiler Plus)

Sorted groups

4 additional STR loci tested (COfiler)

RELATIONAL DATABASE

Database query

Identification of 229 victims

Required a total of 71,490 genotype comparisons

Figure 24.3, J.M. Butler (2005) Forensic DNA Typing, 2nd Edition © 2005 Elsevier Academic Press

Identifying Victims of Mass Disasters

Butler, J.M. (2005) Forensic DNA Typing, 2nd Edition, Chapter 24

POLICY FORUM

EPIDEMIOLOGY


DNA Identifications After the 9/11 World Trade Center Attack


Leticia C. Bressler, Juan E. Bailey-Wilson, Jack Ballantyne, Howard Baum, Frederick B. Bisher, Charles Brenner, Bruce Budowle, John M. Butler, George Carmody, P. Michael Comasally, Barry D. Coleman, Arthur Eisenberg, Lisa Forman, Kenneth K. Kidd, Bruce L. Leland, Steven Hingrad, Thomas J. Harston, Elizabeth Fugh, Robert Shaler, Stephen T. Sherry, Amanda Soccer, Anna Walsh

Science (2005) 310: 1122-1123

Largest Forensic Case in History

~20,000 bone fragments were processed
>6,000 family reference samples and personal effects samples were analyzed





STRBase

Short Tandem Repeat DNA Internet Database
<http://www.cstl.nist.gov/biotech/strbase>

<u>General Information</u>	<u>Forensic Interest Data</u>	<u>Supplemental Info</u>
•Intro to STRs (downloadable PowerPoint)	•FBI CODIS Core Loci	•Reference List >2500
•STR Fact Sheets	•DAB Standards	•Technology Review
•Sequence Information	•NIST SRMs 2391	•Addresses for Scientists
•Multiplex STR Kits	•Published PCR Primers	•Links to Other Web Sites
•Variant Allele Reports	•Y-Chromosome STRs	•DNA Quantitation
•Training Slides	•Population Data	•mtDNA
	•Validation Studies	•New STRs
	•miniSTRs	•Forensic SNPs

New information is added regularly...

Conclusions

- This is an exciting time to be involved in forensic DNA testing
- However, it is a little scary because technology is advancing so rapidly on some fronts
- Thus, training for both the scientific and legal communities is vital to make the most effective use of the wonderful power of DNA technology

Thank you for your attention...

Questions?

Our publications and presentations are available at:
<http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm>

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