AAFS 2009 CRIMINALISTICS Session I:
Envisioning the Future

Forensic DNA:

Perspectives on Progress in a Rapidly Growing Field

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NIST and NIJ Disclaimer

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Presentation Outline

- Historical Perspective
  - where have we come in the past 25 years with DNA?
- Current conditions:
  - applications, advantages, and advocates of DNA
- Impact of DNA database growth
- Future predictions

Value of a Historical Review

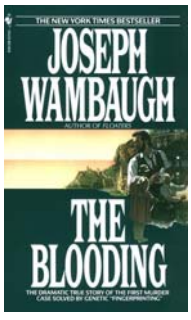
"If you want to understand today, you have to search yesterday."

– Attributed to Pearl Buck
(http://www.quotegarden.com/history.html)

Pearl Buck

The Nobel Prize in Literature 1938

Lessons from the First Case Involving DNA Testing

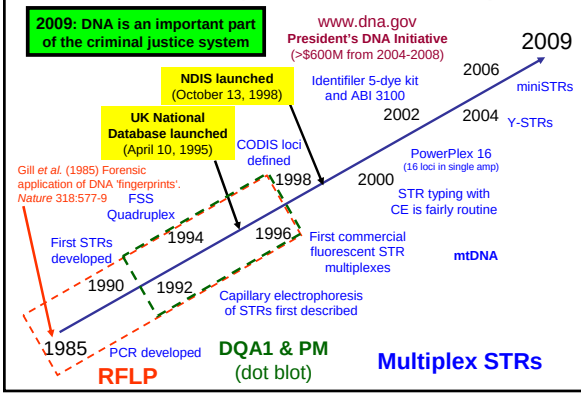


Describes the first use of DNA (in 1986) to solve a double rape-homicide case in England; about 5,000 men asked to give blood or saliva to compare to crime stains

- Connection of two crimes (1983 and 1986)
- Use of "DNA database" to screen for perpetrator (DNA only done on 10% with same blood type as perpetrator)
- Exoneration of an innocent suspect
- DNA was an investigative tool – did not solve the case by itself (confession of an accomplice)

A local baker, Colin Pitchfork, was arrested and his DNA profile matched with the semen from both murders. In 1988 he was sentenced to life for the two murders.

Historical Perspective on DNA Typing



Stages of Forensic DNA Progression

Stages	Time Frame	Description
Exploration	1985-1995	Beginnings, different methods tried (RFLP and early PCR)
Stabilization	1995-2005	**Standardization to STRs**, selection of core loci, implementation of Quality Assurance Standards
Growth	2005-2009	Rapid growth of DNA databases, extended applications pursued
Sophistication	*The Future*	*Expanding tools available, confronting privacy concerns*

AAFS 2009 Topics Regarding Forensic DNA

- Improved DNA extraction
- Predicting hair color and ancestry with SNPs
- X-chromosome STRs
- **Familial searching**
- Y-STRs and mixtures
- **Low level DNA samples**
- miniSTRs
- DNA screening assays
- Optimizing database labs
- Microfluidic biochip systems
- Use with property crimes
- Recovery from handguns
- DNA from IEDs
- Expert systems
- Automation with robotics
- DNA quantitation – qPCR
- PCR directly from blood
- mtDNA
- RNA
- Non-human DNA (dogs & cows)
- **Mixture interpretation**

Applications for DNA Testing

- Crime solving – **matching suspect with evidence…**
- Accident victims – **after airplane crashes…**
- Soldiers in war – **who is the "unknown" soldier…**
- Paternity testing – **who is the father…**
- Immigration testing **– are two people related…**
- Missing persons investigations – **whose remains…**
- Convicted felons databases – **cases solved…**

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

Advantages for STR Markers

- Small product sizes are generally **compatible with degraded DNA** and PCR enables recovery of information from **small amounts of material**
- Numerous alleles per locus aid **mixture interpretation**
- Multiplex amplification with fluorescence detection enables **high power of discrimination** in a single test
- Commercially available in an **easy to use kit format**
- Uniform set of **core STR loci** provide capability for national (and international) sharing of criminal DNA profiles

Advocates for DNA Funding and Expansion

Debbie Smith

Victim

Mitch Morrissey

Prosecutor

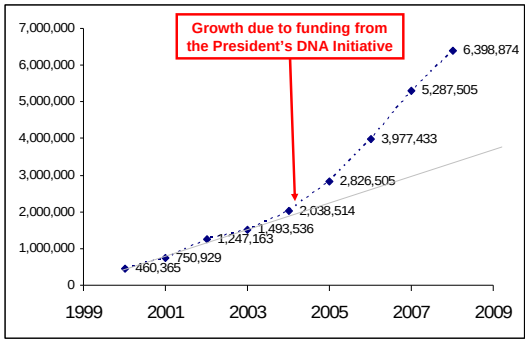
Kirk Bloodsworth

Exoneree
(Innocence Project)

Growth of DNA Databases

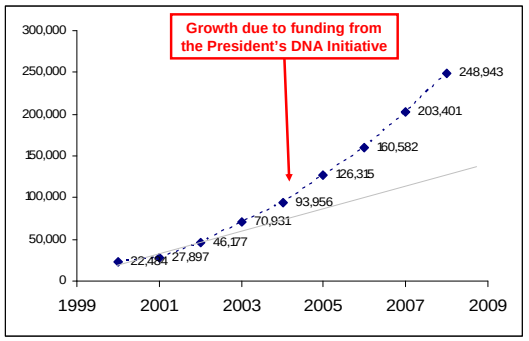
- Have benefited from significant federal funding over the past five years
- Expanded laws now enable more offenders to be included
- Have effectively locked technology with core STR markers used to generate DNA profiles that now number in the millions

Numbers of **Offenders** in U.S. National DNA Database



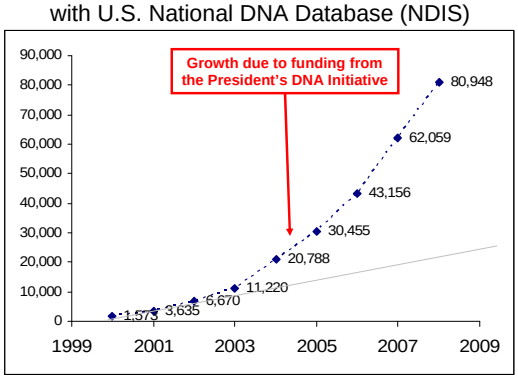
Source: FBI Laboratory's CODIS Unit

Numbers of **Forensic Samples** in U.S. National DNA Database



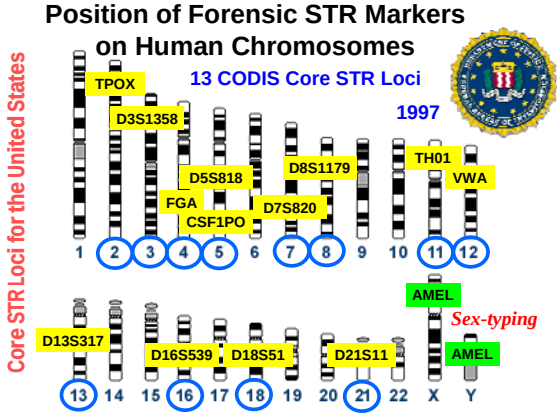
Source: FBI Laboratory's CODIS Unit

Numbers of **Investigations Aided** with U.S. National DNA Database (NDIS)



Source: FBI Laboratory's CODIS Unit

Position of Forensic STR Markers on Human Chromosomes



Possible scenarios for extending sets of genetic markers to be used in national DNA databases

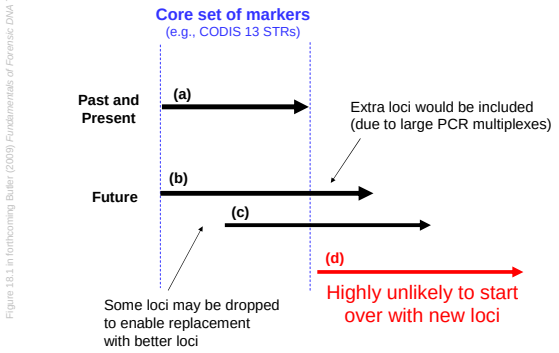


Figure 18.1 in forthcoming Butler (2009) *Fundamentals of Forensic DNA Typing*

Additional STR Loci in the Future?

- Will be needed for more complex kinship analyses and extended applications
  - Example: Y-STRs needed for familial searching
- Immigration testing needs more than 13 STRs
- Larger DNA databases will require more loci

Going Beyond the Core Competencies of Forensic DNA Testing

Core Competency

Standard STR Typing (DNA Profile)

Sufficient DNA quantity (ng)

Direct Matching (or Parentage)

Challenging kinship search questions →

Familial Searching Attempts (fishing for brothers or other relatives)

Solution: Additional Markers (Y-chromosome, more STRs) and Multiple Reference Samples

Lower amounts of DNA being tested ↓

Touch DNA Attempts (poor quality, mixtures, low-level stochastic effects)

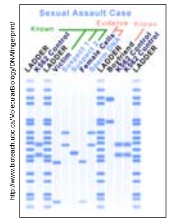
Solution: Replicate Testing

Be very cautious when outside the box…

Figure 18.3 in forthcoming Butler (2009) *Fundamentals of Forensic DNA Typing*

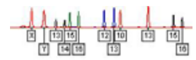
The DNA Field Moves Forward…

The Past	The Present	The Future
RFLP	STRs	



http://www.biotech.ubc.ca/MolecularBiology/DNAfingerprint/

RFLP



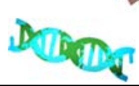
STRs

http://www.marnastungare.com/publications/genetic/dna.gif

Future Predictions

http://www.marnastungare.com/publications/genetic/dna.gif

- **More Automation**
- **Expert Systems**
- **Animal & Plant DNA**
- **Portable Devices**
- **Estimation of Physical Characteristics and Sample Ethnicity**



STRs vs SNPs Article

Butler et al. (2007) STRs vs SNPs: thoughts on the future of forensic DNA testing. *Forensic Science, Medicine and Pathology* 3:200-205.

Forensic Sci Med Pathol (2007) 3:200–205
DOI 10.1007/s12024-007-0018-1

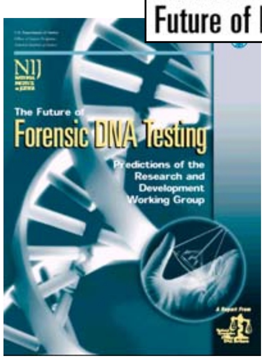
ORIGINAL PAPER

STRs vs. SNPs: thoughts on the future of forensic DNA testing

John M. Butler · Michael D. Coble ·
Peter M. Vallone

- **SNPs are unlikely to replace STRs** for routine forensic DNA testing due to challenges with high-level multiplexing and mixture detection/interpretation
- Most likely use of SNPs will be as ancestry-informative markers (AIMs) **for sample ethnicity estimation**

National Commission on the Future of DNA Evidence



- Report published in Nov 2000
- Asked to estimate where DNA testing would be 2, 5, and 10 years into the future

Conclusions

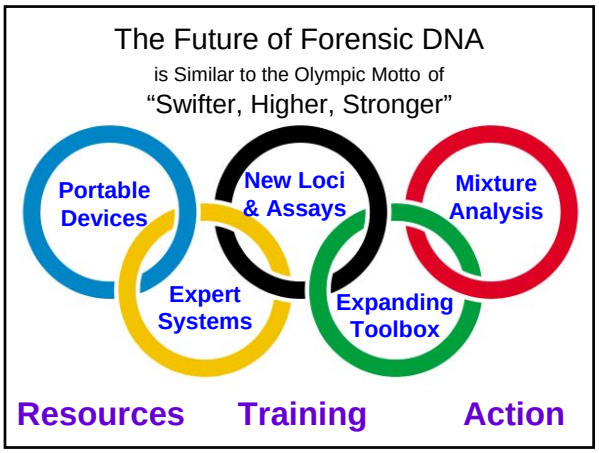
STR typing is here to stay for a few years because of DNA databases that have grown to contain millions of profiles

http://www.ojp.usdoj.gov/nij/pubs-sum/183697.htm

Summary of NIJ-Funded Research

9 areas of funded research

http://www.dna.gov/research/


The Future of Forensic DNA
is Similar to the Olympic Motto of
"Swifter, Higher, Stronger"
Portable Devices
New Loci & Assays
Mixture Analysis
Expert Systems
Expanding Toolbox
Resources
Training
Action


Thank you for your attention…
Our publications and presentations are available at:
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