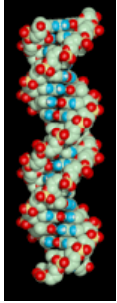
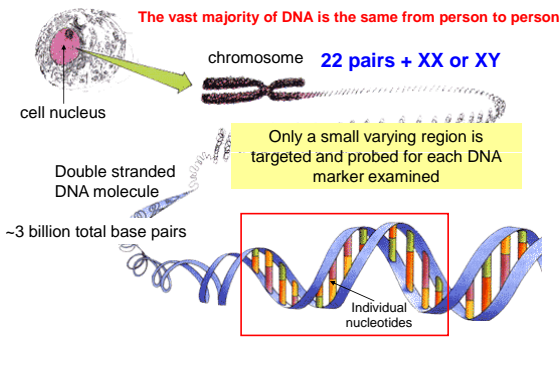


General Characteristics of Genomic DNA



- Each person has a unique DNA profile (except identical twins)
- Each person's DNA is the same in every cell (DNA from skin cells will match DNA from blood cells)
- An individual's DNA profile remains the same throughout life
- Half of your DNA comes from your mother and half from your father

DNA in the Cell



Forensic DNA Testing

Probe subsets of genetic variation in order to differentiate between individuals

DNA typing must be done efficiently and reproducibly (information must hold up in court)

Typically, we are not looking at genes – little/no information about race, predisposition to disease, or phenotypic information (eye color, height, hair color) is obtained

Short Tandem Repeat (STR) Markers

An accordion-like DNA sequence that occurs between genes

TCCCAAGCTCTTCTCTCCCTAGATCAATACAGACAGAAGACAGGTGG
 ATAGATAGATAGATAGATAGATAGATAGATAGATAGATATCATTGA
 AAGACAAAACAGAGATGGATGATAGATACATGCTTACAGATGCACAC

= 11 GATA repeats ("11" is all that is reported)

The number of consecutive repeat units can vary between people

The FBI has selected **13 core STR loci** that must be run in all DNA tests in order to provide a common currency with DNA profiles

Position of Forensic STR Markers on Human Chromosomes

Core STR Loci for the United States

13 Core U.S. STR Loci

1997

Sex-typing

Sources of Biological Evidence

- Blood
- Semen
- Saliva
- Urine
- Hair
- Teeth
- Bone
- Tissue

Blood Sample

Only a very small amount of blood is needed to DNA

best results with >100 cells, but DNA profiles can be recovered from fewer cells

Applications

- Forensic cases: matching suspect with evidence
- Paternity testing: identifying father
- Missing persons investigations
- Military DNA "dog tag"
- Convicted offender DNA databases
- Mass fatalities
- Historical investigations
- Genetic genealogy
- DNA as a biometric tool

DNA Testing Requires a Reference Sample

A DNA profile by itself is fairly useless because it has no context...

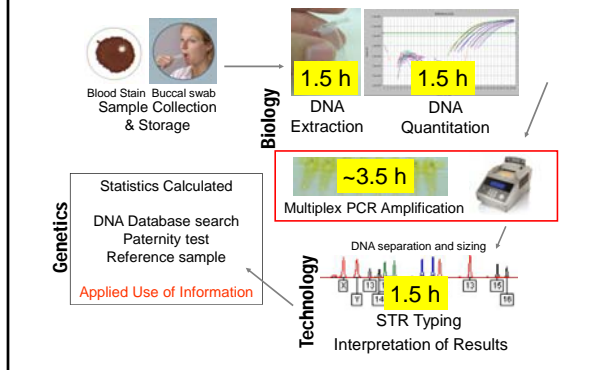


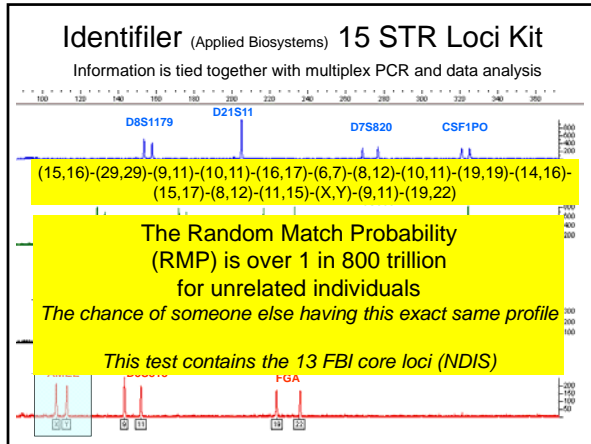
DNA analysis for identity only works by comparison – **you need a reference sample**

- **Crime Scene Evidence** compared to **Suspect(s)** (Forensic Case)
- **Child** compared to **Alleged Father** (Paternity Case)
- **Victim's Remains** compared to **Biological Relative** (Mass Disaster ID)
- **Soldier's Remains** compared to **Direct Reference Sample** (Armed Forces ID)

Steps in Forensic DNA Analysis

Usually 1-2 day process (a minimum of ~8 hours)





Kinship Testing

- DNA profiles can also be used to evaluate the probability of a specific familial relationship
- As a familial relationship becomes more distant, the ability of DNA to confirm the likelihood of that relationship decreases
 1. Parent-offspring
 2. Siblings
 3. Half siblings = uncle/nephew = grandchild
 4. Cousins

Complex Kinship Testing

Requirements:

- Genotypes of individuals being tested
- Allele frequencies for the loci involved in the testing
- A Hypothesis!

- Basic statistical equations are known
- Difficult to identify distant relationships
- Discriminatory power comes from multiple family members and the use of informative markers

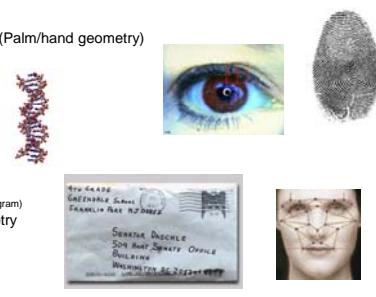
The statistical power for complex kinship testing significantly decreases compared to one-to-one matching

DNA as a Biometric

Current Biometrics

Some commonly measured features

- Physical
 - Fingerprints (Palm/hand geometry)
 - Iris, retinal
 - Face
 - Odor/scent
 - DNA?
- Behavioral
 - Gait
 - Voice
 - Vein (IR thermogram)
 - Hand geometry
 - Handwriting



Characteristics of a Biometric

- Universality
 - each person should have the characteristic
- Uniqueness
 - is how well the biometric separates individuals from another
- Permanence
 - measures how well a biometric resists aging and variance over time
- Collectability
 - ease of acquisition for measurement

Jain, A. K., Ross, Anuj, Prabhakar, Sath (January 2004), "An introduction to biometric recognition", IEEE Transactions on Circuits and Systems for Video Technology 14(1): 4-20

Characteristics of a Biometric

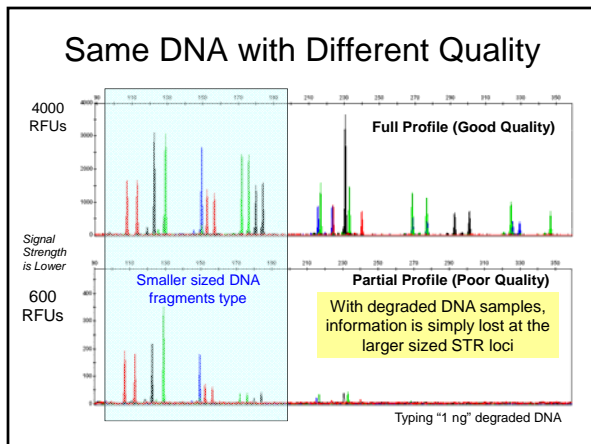
(practical considerations)

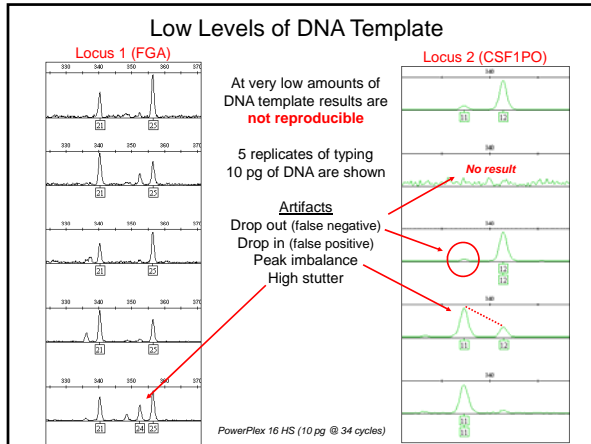
- Performance
 - accuracy, **speed**, and robustness of technology used
- Acceptability
 - degree of approval of a technology
- Circumvention
 - ease of use of a substitute

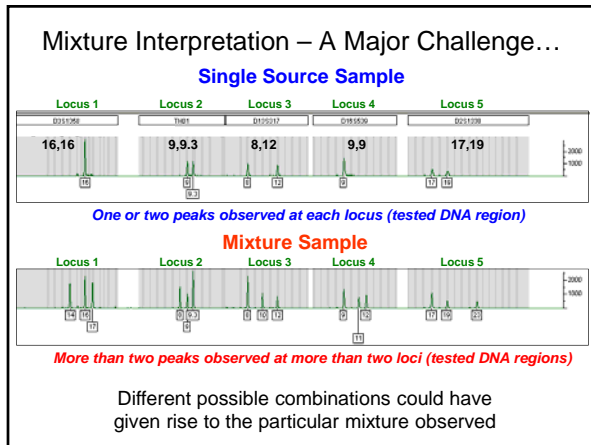
Jain, A. K., Ross, Anuj; Pabhanikar, Salf (January 2004). "An introduction to biometric recognition". IEEE Transactions on Circuits and Systems for Video Technology 14(1): 4-20

DNA Typing as a Biometric

<p><i>Advantages</i></p> <ul style="list-style-type: none">• High level of accuracy (Gold Standard)• Solid foundation of Forensic DNA Testing (pop stats, molecular biology, court acceptance, protocols, training, education)• Kinship determination (unique to DNA)• Potential use for:<ul style="list-style-type: none">– Phenotype (traits: eye/hair color)– Biogeographical Ancestry	<p><i>Challenges</i></p> <ul style="list-style-type: none">• Expensive• Time consuming• Sample collection (invasive, stability issues)• Technical expertise required for analysis• Low level template, mixtures, PCR inhibition• Policy/Privacy/Ethical issues
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Interest in Rapid DNA Typing

- DoD (field testing, rapid intelligence, mass fatalities)
- DHS (kinship determination, border security, immigration)
- DoJ (law enforcement, initial information)
- Industry (security, authentication)

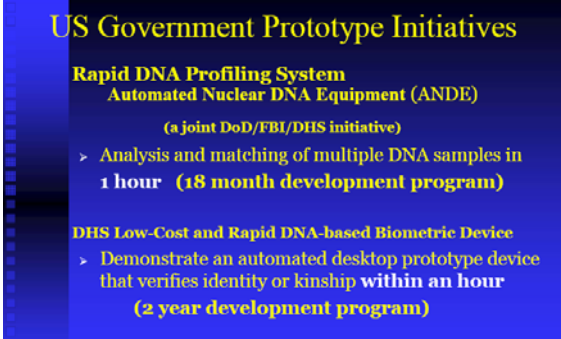
- Each customer will have specific requirements
 - sample input
 - information output
 - degrees of 'accuracy'

The time required for generating a STR profile will have to be significantly reduced

Goals for Rapid DNA Typing Systems

- Develop an **integrated system** capable of performing DNA testing in less than **1 hour**
- Little user interaction (or experience)
- Rugged
- Robust **Swab in...answer out**
- Simple data interpretation
- 4-16 samples per run
- Disposable chips (with reagents on board)

ANDE (Automated Nuclear DNA Equipment)



US Government Prototype Initiatives

Rapid DNA Profiling System
Automated Nuclear DNA Equipment (ANDE)
(a joint DoD/FBI/DHS initiative)

- Analysis and matching of multiple DNA samples in **1 hour (18 month development program)**

DHS Low-Cost and Rapid DNA-based Biometric Device

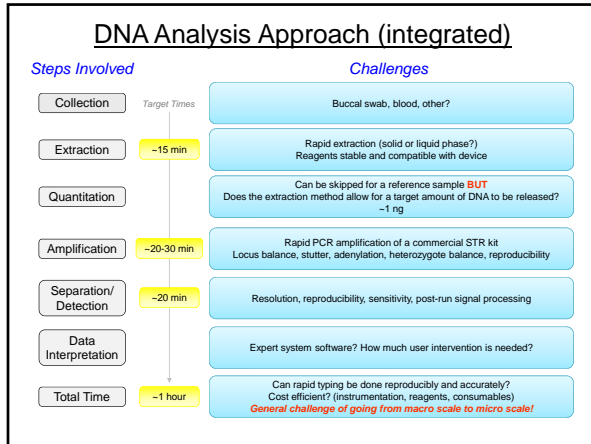
- Demonstrate an automated desktop prototype device that verifies identity or kinship **within an hour (2 year development program)**

<http://biometrics.org/bc2009/presentations/wednesday/McCurdy%20BrA%20Wed%201040-1055.pdf>

Rapid DNA Typing Systems Under Development

- Systems are currently under development and are not yet commercially available
- Network Biosystems (Woburn, MA)
<http://www.netbio.com>
- MicroLab Diagnostics and Lockheed Martin (Charlottesville, VA)
<http://www.microlabdiagnostics.com>
- Microchip Biotechnologies, Inc. (Pleasanton, CA)
<http://www.microchipbiotech.com>
- Forensic Science Service (UK)
<http://www.forensic.gov.uk/>

Use of DNA as a Biometric Tool, American Academy of Forensic Science, Feb 22, 2010, Seattle, WA
<http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm>




- ### NIST Efforts with DNA Biometrics
- **Developing rapid PCR protocols**
 - Evaluating kinship analysis software
 - Support for other rapid DNA efforts
 - Designing standards materials for device testing
 - Preparing to test prototype rapid DNA devices

Recent Work with Rapid PCR


At NIST we are working on new PCR methods to reduce the time for PCR down to 20 minutes

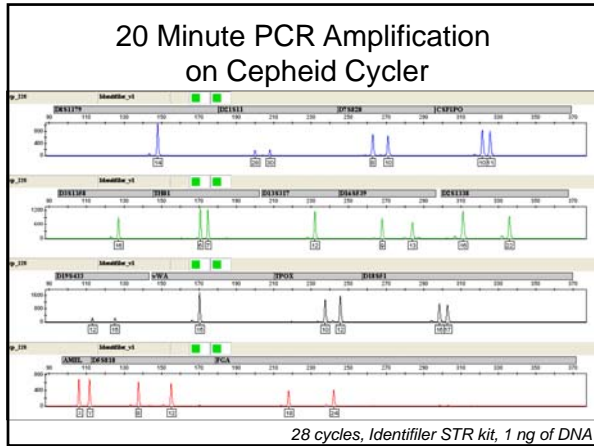
Polymerase Chain Reaction (PCR) is a means to create billions of exact copies of the human genome – necessary/essential for DNA typing

~3.5 h → ~20 min?



Multiplex PCR Amplification





Rapid PCR Article

ARTICLE IN PRESS

Forensic Science International: Genetics

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

Short communication
Demonstration of rapid multiplex PCR amplification involving 16 genetic loci[®]
Peter M. Vallone^{a,*}, Carolyn R. Hill, John M. Butler
^a National Institute of Standards and Technology, Biomedical Science Division, 370 Research Drive, Mail Stop 4711, Gaithersburg, MD 20899-4711, United States

Vallone, P.M., Hill, C.R., Butler, J.M. (2008) Demonstration of rapid multiplex PCR amplification involving 16 genetic loci. *FSI Genetics* 3(1): 42-45.

Rapid PCR Amplification of STR Typing Kits 20th Annual International Symposium on Human Identification (Promega Meeting) October 14, 2009, Las Vegas, NV

Rapid Amplification of Commercial STR Typing Kits, International Society of Forensic Genetics (ISFG), September 16, 2009, Buenos Aires, Argentina

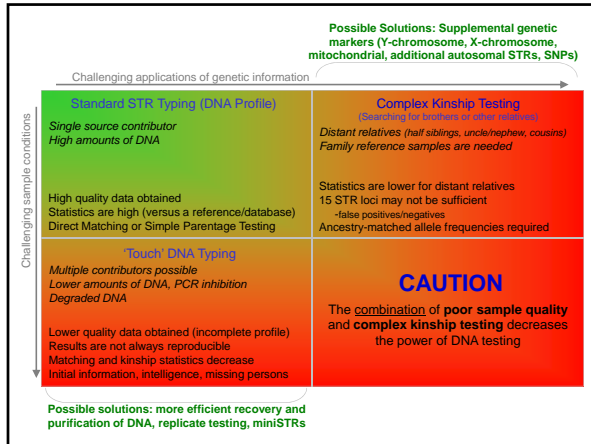
Use of DNA as a Biometric Tool, American Academy of Forensic Science, Feb 22, 2010, Seattle, WA

<http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm>

- ### Future Directions
- Functional prototypes should be available for testing in the next 12-18 months

 - 3-4 year horizon until concordance testing and validation

 - Further education on the **strengths and limitations** of DNA



Thank you for your attention!

Questions?
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 301-975-4872

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 Dr. Kristen Lewis
 Becky Hill

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