Forensic DNA Testing:
Beyond CSI

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Unfortunately, current DNA testing cannot be performed as quickly as a commercial break…

Real labs have better lighting but fewer instruments. The instruments on CSI are real – they just do not collect data as quickly as shown on TV.
The Beginning of My Interest in Forensic DNA Typing

• At the age of 15, I decided to pursue a career in forensic science

• This book was given to me in 1990 by Dave Feldman, the Acting Police Chief for Fort Collins, Colorado

• It describes the first use of DNA (in 1986) to solve a double rape-homicide case in England

You never know the impact you can have on another person

Progress Since 1995…

O.J. Simpson DNA testing was performed with RFLP

Almost 8 weeks needed to get results

Now <8 hours to get results
We are finding new ways to use DNA…

Applications of Forensic DNA Typing

- 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

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

Methods for Human Identification

Fingerprints have been used since 1901

DNA since 1986
DNA in the Cell

The vast majority of DNA is the same from person to person.

- **22 pairs + XX or XY**
- **~3 billion total base pairs**
- **Double stranded DNA molecule**

Only a Small Varying Region is Targeted and Probed for Each DNA Marker Examined

Information Storage

You know that no two people share the same fingerprint, but did you know that the cells that make up your body also have a unique fingerprint unlike anyone else’s? Your cells contain a complex molecule that we call DNA. Unless you have an identical twin, no one else has DNA just like yours.

Scientists can analyze DNA. If a criminal leaves DNA at a crime scene, police can use it to prove who committed the crime. At NIST, we help crime labs analyze DNA accurately. We make DNA standards so crime labs can tell if their results are right.

Text Storage is by the order of letters, words and paragraphs

DNA Storage is by the order of nucleotides, genes and chromosomes
Identification of Information

<table>
<thead>
<tr>
<th>Printed Information</th>
<th>Genetic Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Library</td>
<td>Body</td>
</tr>
<tr>
<td>Book</td>
<td>Cell</td>
</tr>
<tr>
<td>Chapter</td>
<td>Nucleus</td>
</tr>
<tr>
<td>Page Number</td>
<td>Chromosome</td>
</tr>
<tr>
<td>Line on Page</td>
<td>Locus (part of chromosome)</td>
</tr>
<tr>
<td>Word</td>
<td>Short DNA sequence</td>
</tr>
<tr>
<td>Letter</td>
<td>DNA nucleotides</td>
</tr>
</tbody>
</table>

Characteristics of DNA

- Each person has a unique DNA profile (except identical twins).
- Each person's DNA is the same in every cell.
- An individual’s DNA profile remains the same throughout life.
- Half of your DNA comes from your mother and half from your father.
Our DNA Comes from our Parents

Father’s Sperm

Mother’s Egg

Child’s Cell

Inheritance Pattern of DNA Profiles

DAD

CHILD

MOM
Short Tandem Repeat (STR) Markers

An accordion-like DNA sequence that occurs between genes

TCCTGAATCTTCCTCTTCCTCCCTAGATCAATACAGACAGACA
GGTGATGATGATGATGATGATGATGATGATGATGATGATGAT
TAGATATCATGAAAGACAAAACAGAGATGGATGATAGATACAT
GCTTACAGATGACAC

= 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

The target region (short tandem repeat)

Position of Forensic STR Markers on Human Chromosomes

13 CODIS Core STR Loci

D13S317 D16S539 D18S51 D21S11 AMEL

AMEL

FTX VWA

Core STR Loci for the United States

1997

Sex-typing
Combined DNA Index System (CODIS)

Launched in October 1998 and now links all 50 states
Used for linking serial crimes and unsolved cases with repeat offenders
Convicted offender and forensic case samples along with a missing persons index
Requires 13 core STR markers
More than 86,000 investigations aided nationwide as of March 2009
Contains more than 7.0 million DNA profiles

No names are associated with DNA profiles uploaded to NDIS
If my profile was entered for searching:
16,17-17,18-21,22-12,14-28,30-14,16-12,13-11,14-14-9,9-9,11-6-8,8-10,10

FBI requires labs putting samples into the CODIS database to run the NIST DNA standard

The Role of NIST Scientists

• **Develop DNA standards** so that laboratories around the world may compare their results.
• **Conduct tests of laboratories** around the world to insure accurate results in DNA testing.
• **Develop new DNA tests** which are more rapid and efficient than those currently used.
Paternity Testing

PCR product size (bp)

Father’s Profile? 11,14

Child #1

Child #2

Child #3

Mother

Alleged Father(s) is asked to donate DNA sample

STR Alleles from D13S317

1 in 837 trillion (probability of this profile occurring at random)
We got him!

Saddam Hussein’s capture was verified with DNA testing conducted in Rockville, MD at the Armed Forces DNA Identification Laboratory.

DNA Profile

Saddam was known to have many “stunt doubles” that acted as decoys for his own safety.

Biological Relatives Served as References

Captured December 13, 2003

Matching Y-STR Haplotype Used to Confirm Identity

(along with allele sharing from autosomal STRs)

Is this man really Sadaam Hussein?

Killed July 22, 2003

Uday and Qusay Hussein

Mystery Solved: The Identification of the Two Missing Romanov Children Using DNA Analysis

Michael D. Coble 1*, Odile M. Lorelle 1*, Mark J. Wadhams 1, Suni M. Edson 1, Kerry Maynard 1, Carina E. Meyer 1, Harald Niederstätter 2, Cordula Berger 2, Burkhard Berger 2, Anthony B. Falsetti 3, Peter Gill 4,5, Walther Parson 2, Louis N. Finelli 1

1 Armed Forces DNA Identification Laboratory, Armed Forces Institute of Pathology, Rockville, Maryland, United States of America, 2 Institute of Legal Medicine, Innsbruck Medical University, Innsbruck, Austria, 3University of Florida, Gainesville, Florida, United States of America, 4Department of Pure and Applied Chemistry, University of Strathclyde, Glasgow, United Kingdom, 5 Institute of Forensic Medicine, University of Oslo, Oslo, Norway

The Romanovs – Russia’s Royal Family (1913)

Photo taken five years before they were killed
Future Predictions

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

Ancestry DNA Tests (Using Lineage Markers)

Y-Chromosome Tests, which examine genetic traits passed on from father to son, provide only a partial picture of the past paternal lineage.

Mitochondrial DNA (mtDNA) Tests, which examine genetic traits passed on from mother to children, provide only a partial picture of the past maternal lineage.

The genetic signatures of many ancestors (and people alive years ago without living descendants) are never seen when testing modern DNA samples.