Forensic DNA Testing: Beyond CSI

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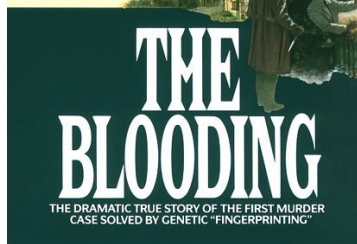Unfortunately, current DNA testing cannot be performed as quickly as a commercial break…

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

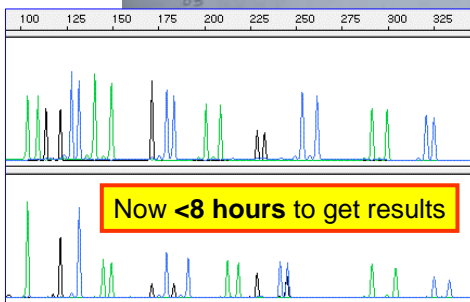**You never know the impact you can have on another person**

- 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
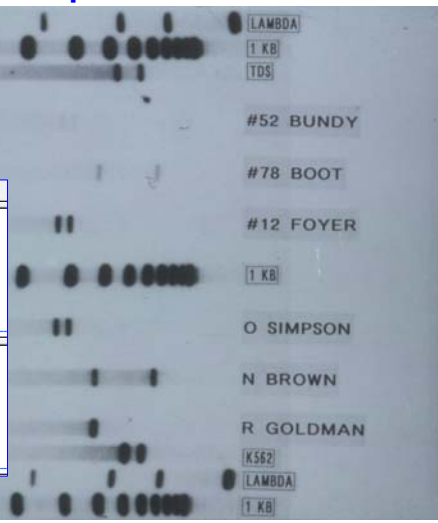

Progress Since 1995…

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**
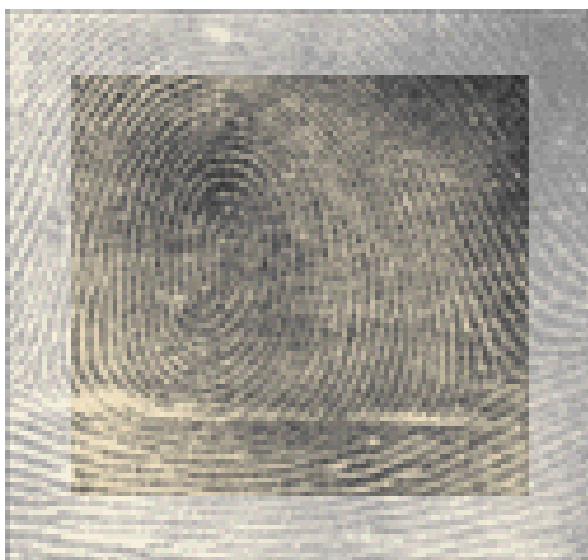

Crime Scene Evidence compared to **Suspect(s)** (Forensic Case)
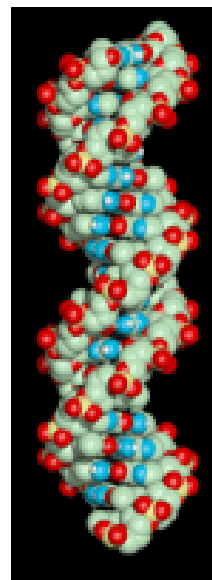**Child** compared to **Alleged Father** (Paternity Case)
Victim's Remains compared to **Biological Relative** (Mass Disaster ID)
Soldier's Remains compared to **Direct Reference Sample** (Armed Forces ID)

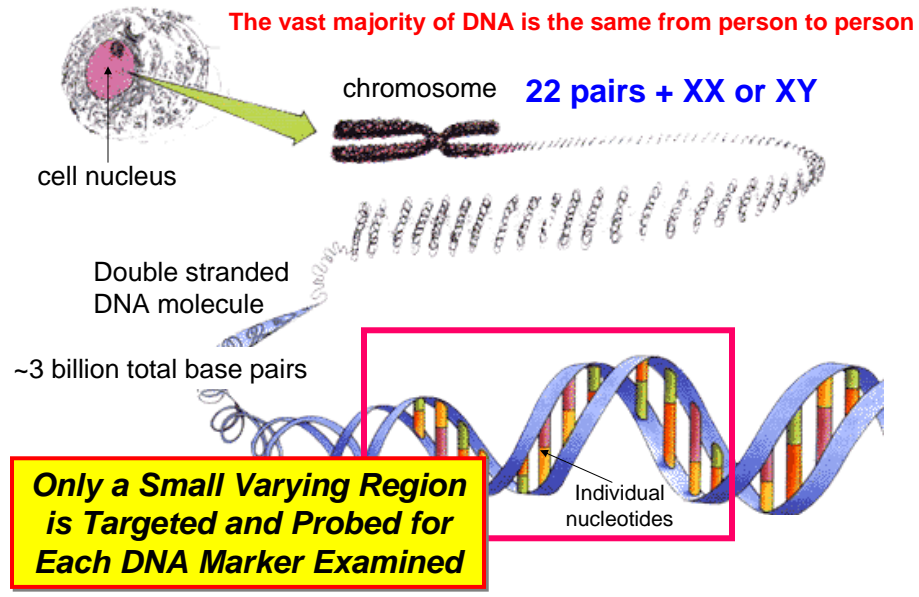---

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

chromosome **22 pairs + XX or XY**

cell nucleus

Double stranded
DNA molecule

~3 billion total base pairs

Individual
nucleotides

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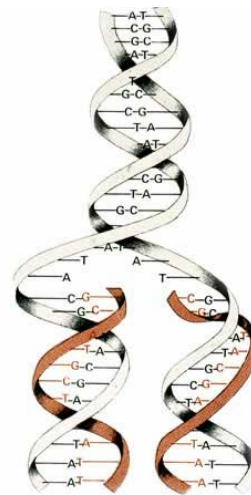is Targeted and Probed for
Each DNA Marker Examined*

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

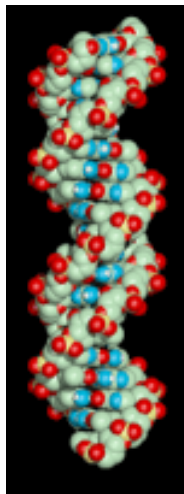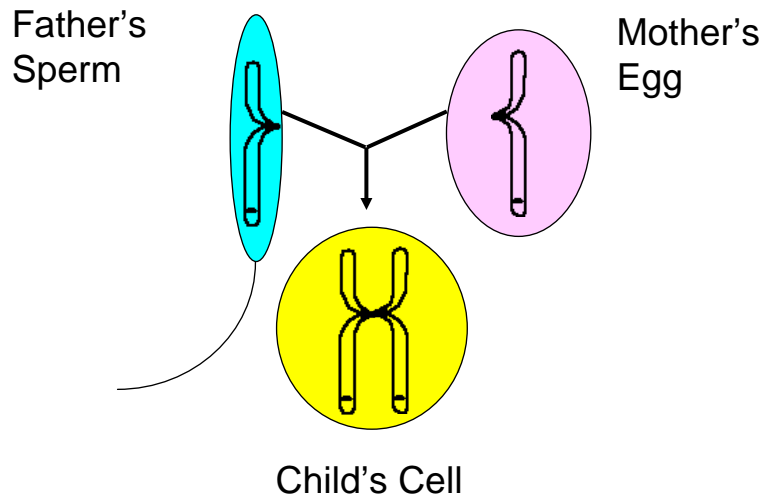| Printed Information	Genetic Information
Library	Body
Book	Cell
Chapter	Nucleus
Page Number	Chromosome
Line on Page	Locus (part of chromosome)
Word	Short DNA sequence
Letter	DNA nucleotides

D13S317
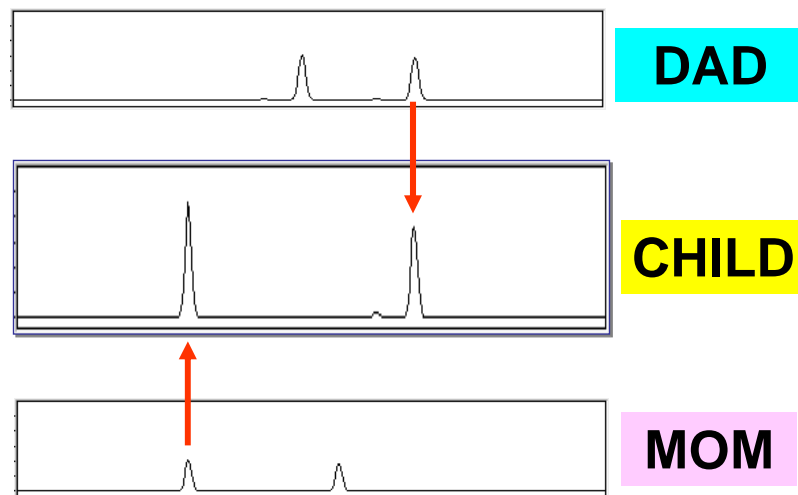
Characteristics of DNA

- 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
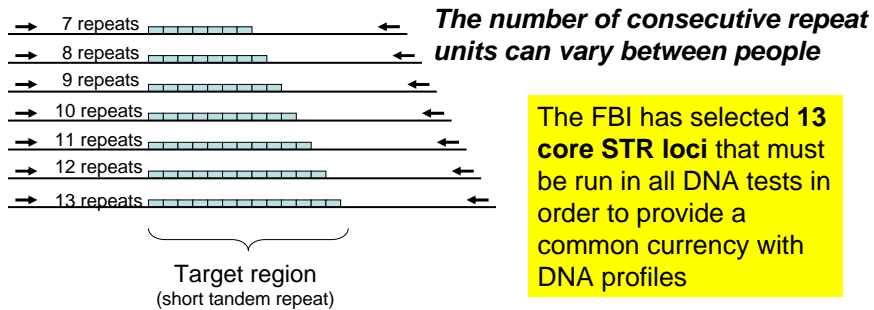
Inheritance Pattern of DNA Profiles

DAD

CHILD
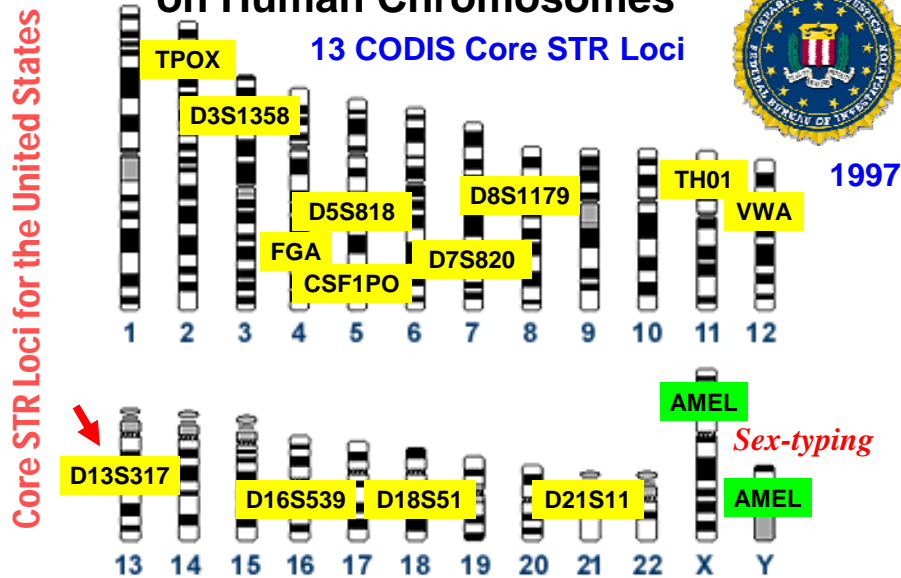
MOM

Short Tandem Repeat (STR) Markers

An accordion-like DNA sequence that occurs between genes

TCCCAAGCTCTTCCTCTTCCCTAGATCAATACAGACAGAAGACA
GGTG**GATAGATAGATAGATAGATAGATAGATAGATAGATAGA
TAGATA**TCATTGAAAGACAAAACAGAGATGGATGATAGATACAT
GCTTACAGATGCACAC

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

→ 7 repeats ←
→ 8 repeats ←
→ 9 repeats ←
→ 10 repeats ←
→ 11 repeats ←
→ 12 repeats ←
→ 13 repeats ←

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

Target region
(short tandem repeat)

Position of Forensic STR Markers on Human Chromosomes

Core STR Loci for the United States

13 CODIS Core STR Loci

1997

TPOX

D3S1358

D5S818

FGA

CSF1PO

D8S1179

D7S820

TH01

VWA

1 2 3 4 5 6 7 8 9 10 11 12

AMEL

Sex-typing

D13S317

D16S539   D18S51

D21S11

AMEL

13 14 15 16 17 18 19 20 21 22 X Y

National DNA Index System (NDIS)

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

**FBI
LABORATORY**

http://www.fbi.gov/hq/lab/codis/index1

Combined DNA Index System (CODIS)

Launched in October 1998
Used for linking s̶... with repeat offenders
Convicte̶... samples along with a missing persons

...R markers
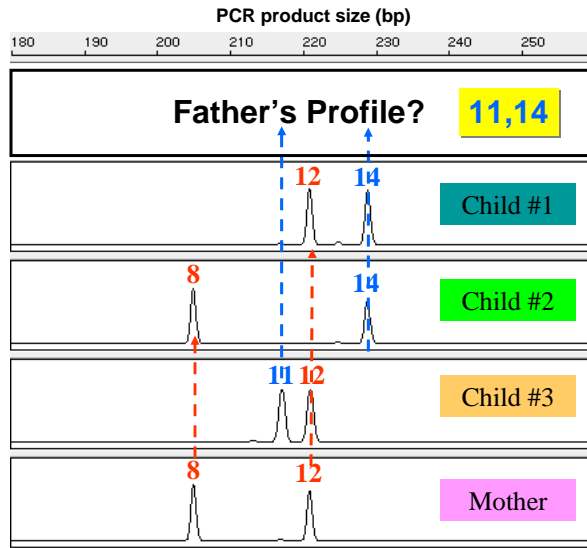...nvestigations aided nationwide as of March 2009
Contains more than 7.0 million DNA profiles

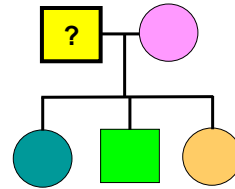*FBI requires labs putting samples into the CODIS database to run the NIST DNA standard*

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.

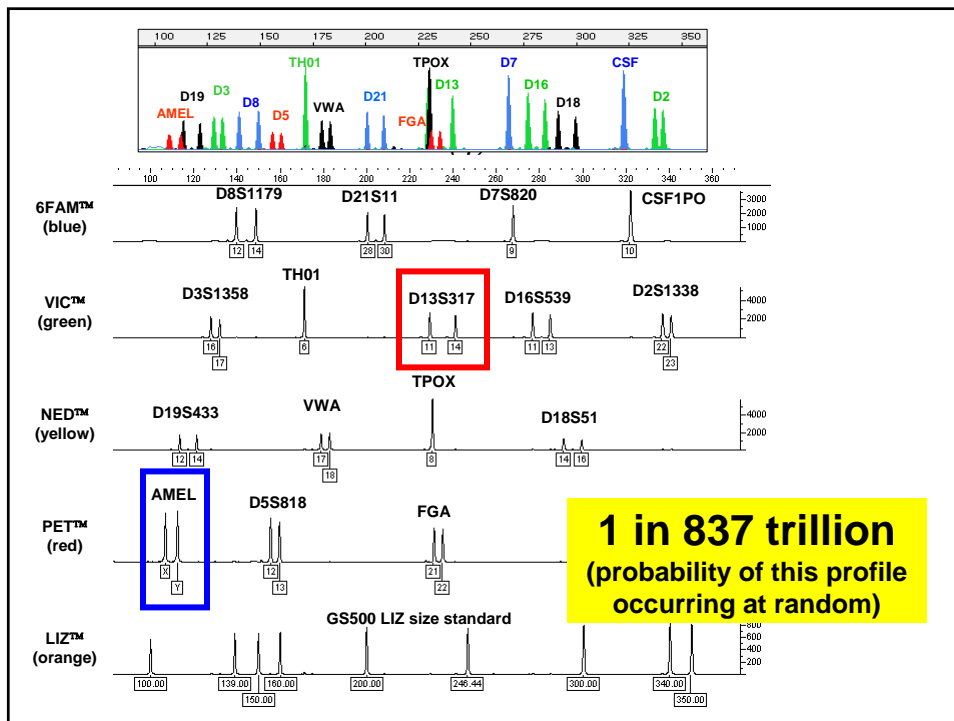- Develop new DNA tests which are more rapid and efficient than those currently used.

PATERNITY TESTING

STR Alleles from D13S317

1 in 837 trillion
(probability of this profile occurring at random)
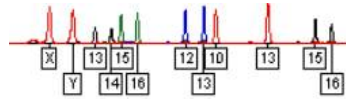
December 14, 2003

"We got him!"

DNA Profile

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

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

Source: www.cnn.com; *The Scientist* Dec 19, 2003

Biological Relatives Served as References

Captured December 13, 2003

Matching Y-STR Haplotype Used to Confirm Identity

(along with allele sharing from autosomal STRs)

Uday and Qusay Hussein

Is this man really Sadaam Hussein?

Killed July 22, 2003

Butler, J.M. (2005) *Forensic DNA Typing, 2nd Edition,* Box 23.1, p. 534

Article published March 11, 2009

http://www.plosone.org

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

Michael D. Coble[1,9]*, Odile M. Loreille[1,9], Mark J. Wadhams[1], Suni M. Edson[1], Kerry Maynard[1], Carna 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, 3 University of Florida, Gainesville, Florida, United States of America, 4 Department of Pure and Applied Chemistry, University of Strathclyde, Glasgow, United Kingdom, 5 Institute of Forensic Medicine, University of Oslo, Oslo, Norway
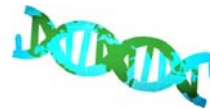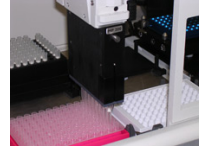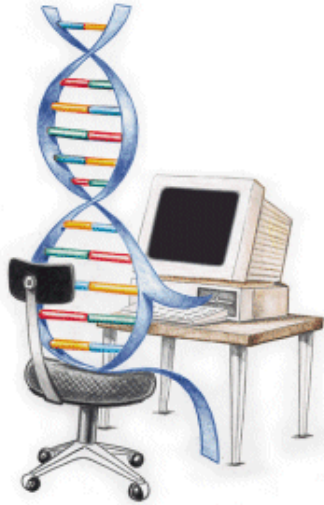
The Romanovs – Russia's Royal Family (1913)
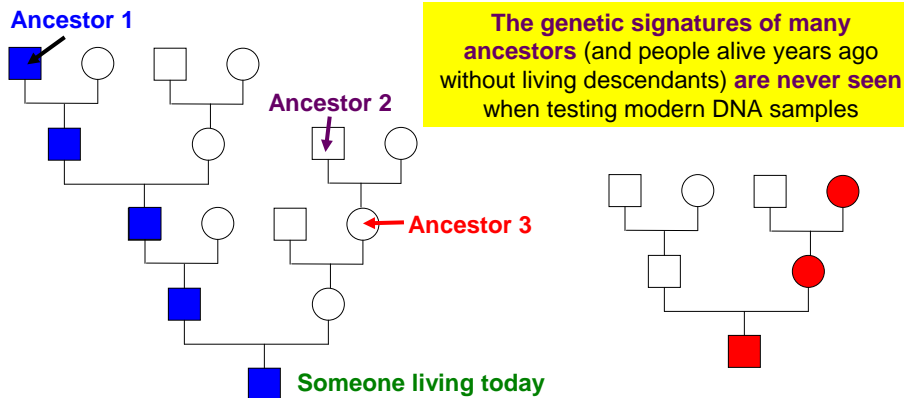

Photo taken five years before they were killed

Future Predictions

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

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

Ancestry DNA Tests (Using Lineage Markers)

Ancestor 1

Ancestor 2

Ancestor 3

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

Someone living today

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**