Understanding Forensic DNA Testing:

Significance of DNA profiling in paternity cases

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Analysis For Life

2011

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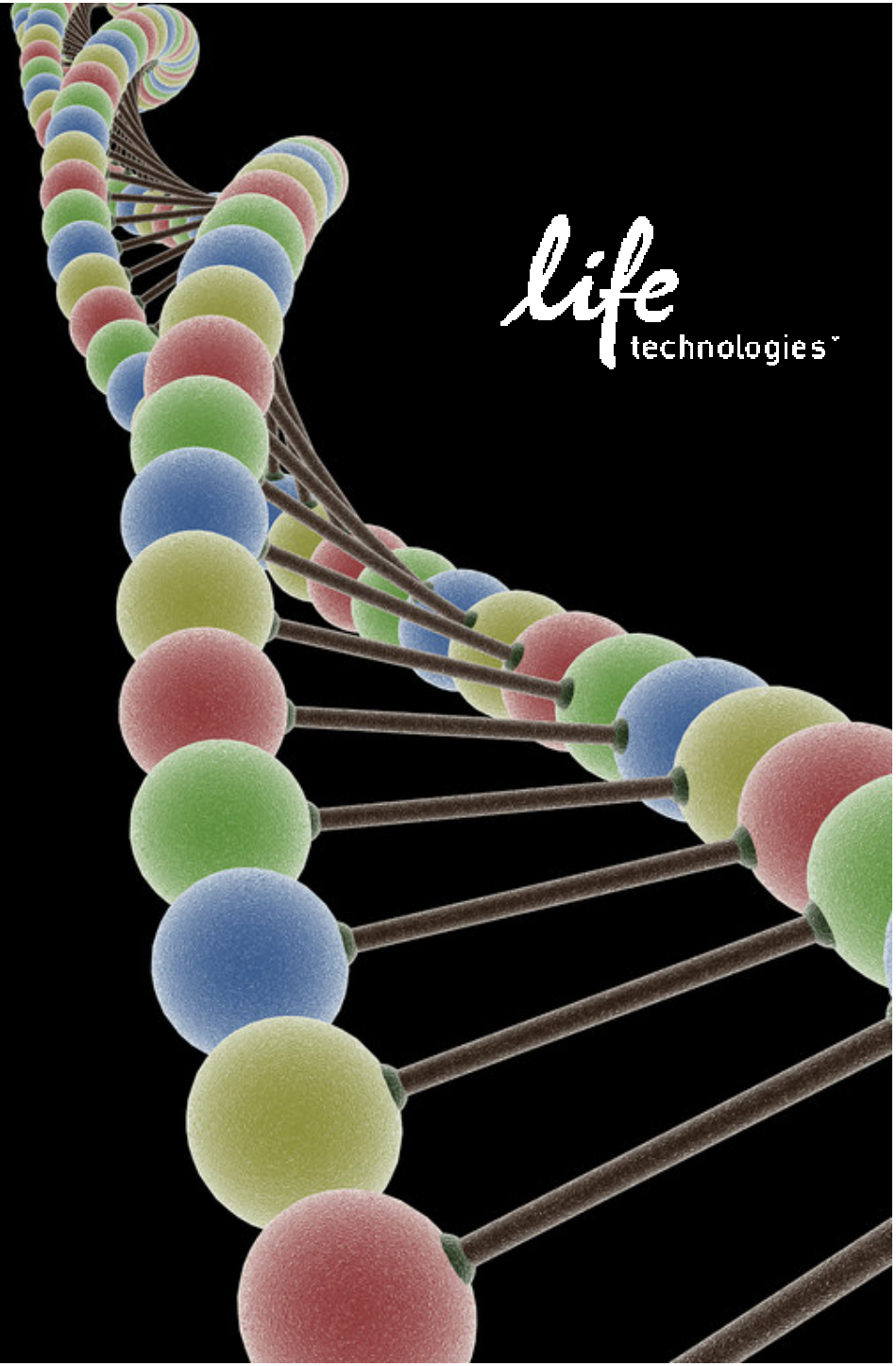
DNA Basic Genetics



What is DNA?

DNA is…

Deoxyribonucleic Acid
The inherited genetic material that makes us what we are

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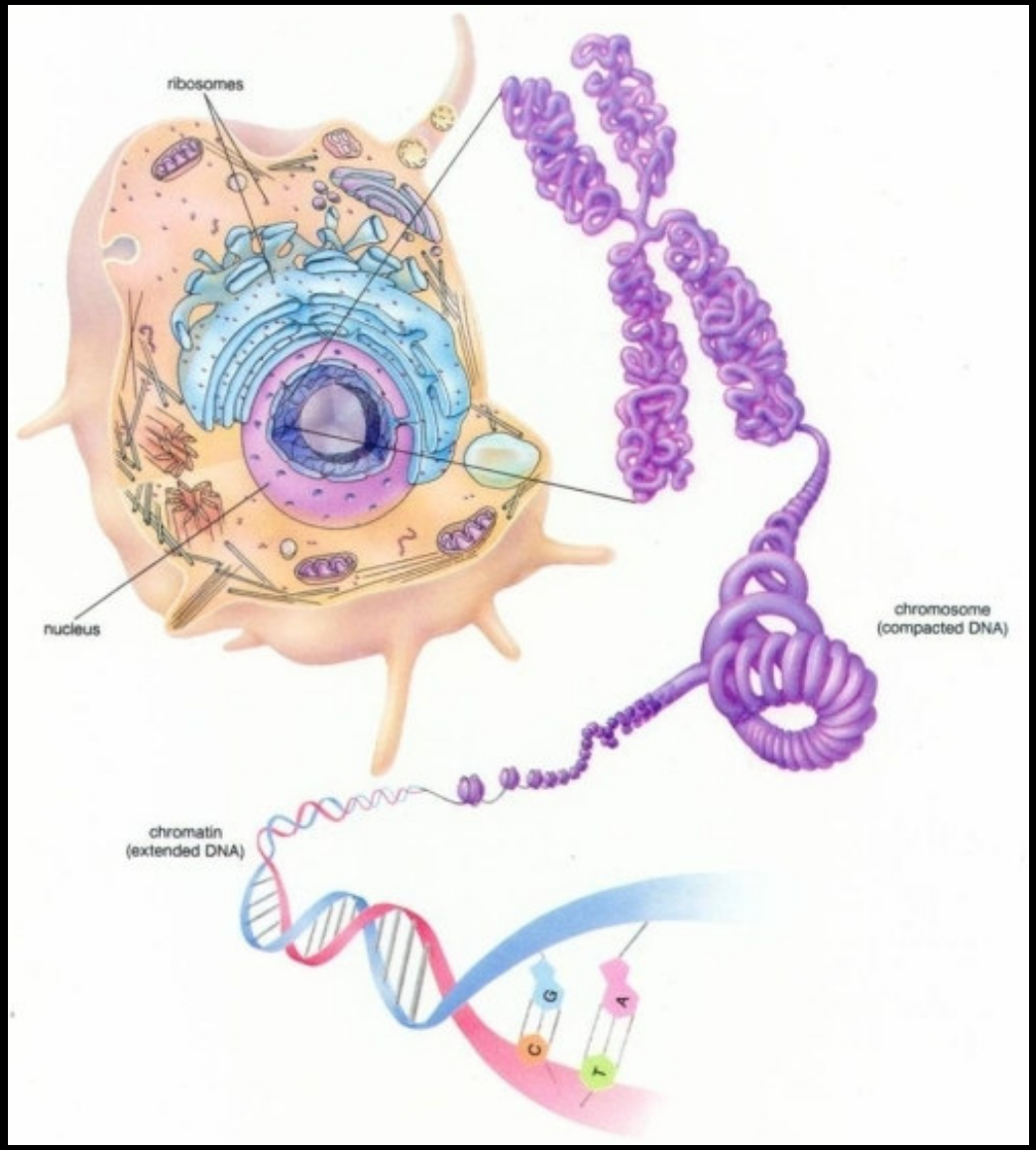
DNA in the Cell



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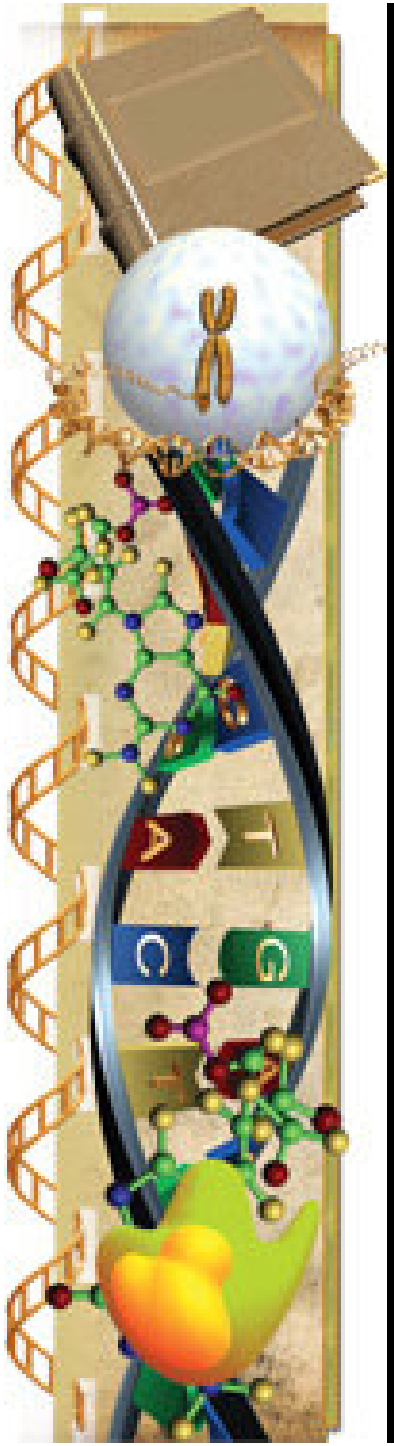

Human Genome

~3 billion base pairs of DNA

30,000-35,000 genes

Population-each gene has multiple forms

Allelic variation-basis of forensic DNA typing

Dozens of polymorphic loci validated for forensic use

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DNA - Unique, Yet the Same

1. Of the 3,000,000,000 DNA bases, about 0.3% is not conserved:

~1 million bases

2. Forensic STR analysis looks at the length of 13-15 areas of DNA.

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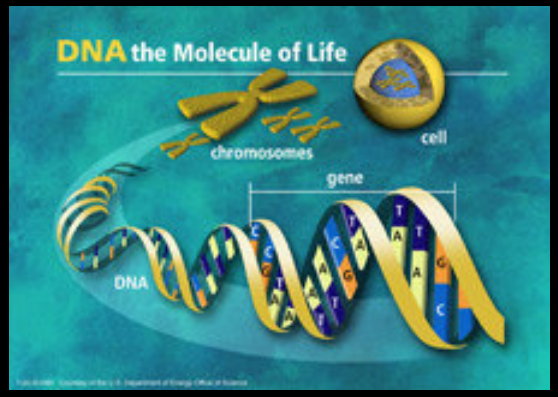
Characteristics of DNA

- DNA is inherited from parents (half from mother, half from father).
- No two individuals have the same DNA profile; except for identical twins.
- Genes have multiple forms. This variation is the basis of forensic DNA typing.



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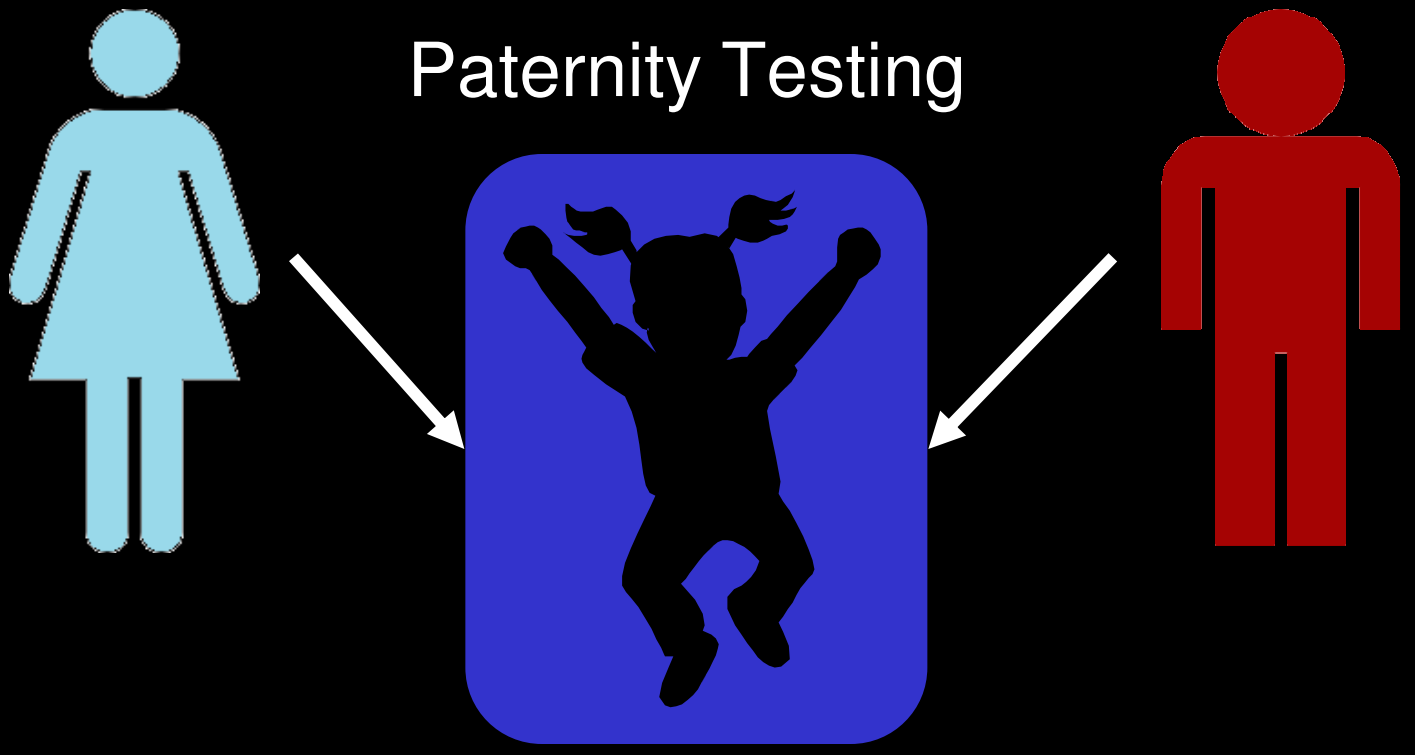
One set of 22 autosomes (plus X)

One set of 22 autosomes (plus X & Y)



Two alleles for each autosomal genetic marker

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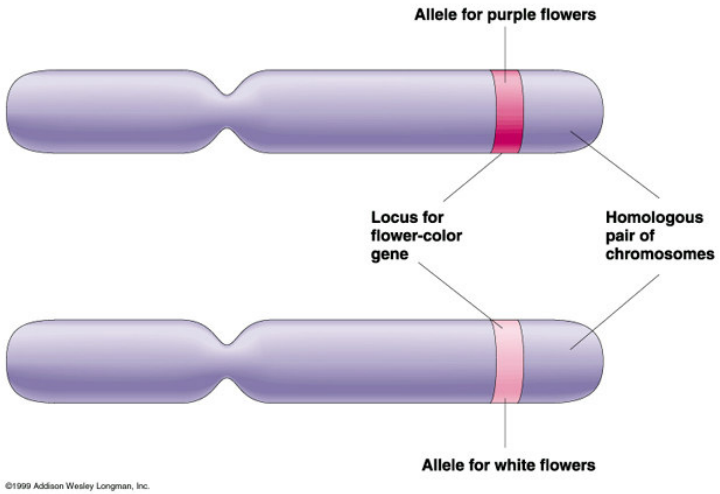
Allele- A variant of a gene or marker. In the context of microsatellite markers, two alleles will differ by the number of repeats present. For example, these are 4 different allele variants for a dinucleotide microsatellite marker.

Allele1	ACGT **CA CA CA CA CA CA** GGCGA
Allele2	ACGT **CA CA CA CA CA** GGCGA
Allele3	ACGT **CA CA CA CA** GGCGA
Allele4	ACGT **CA CA CA** GGCGA

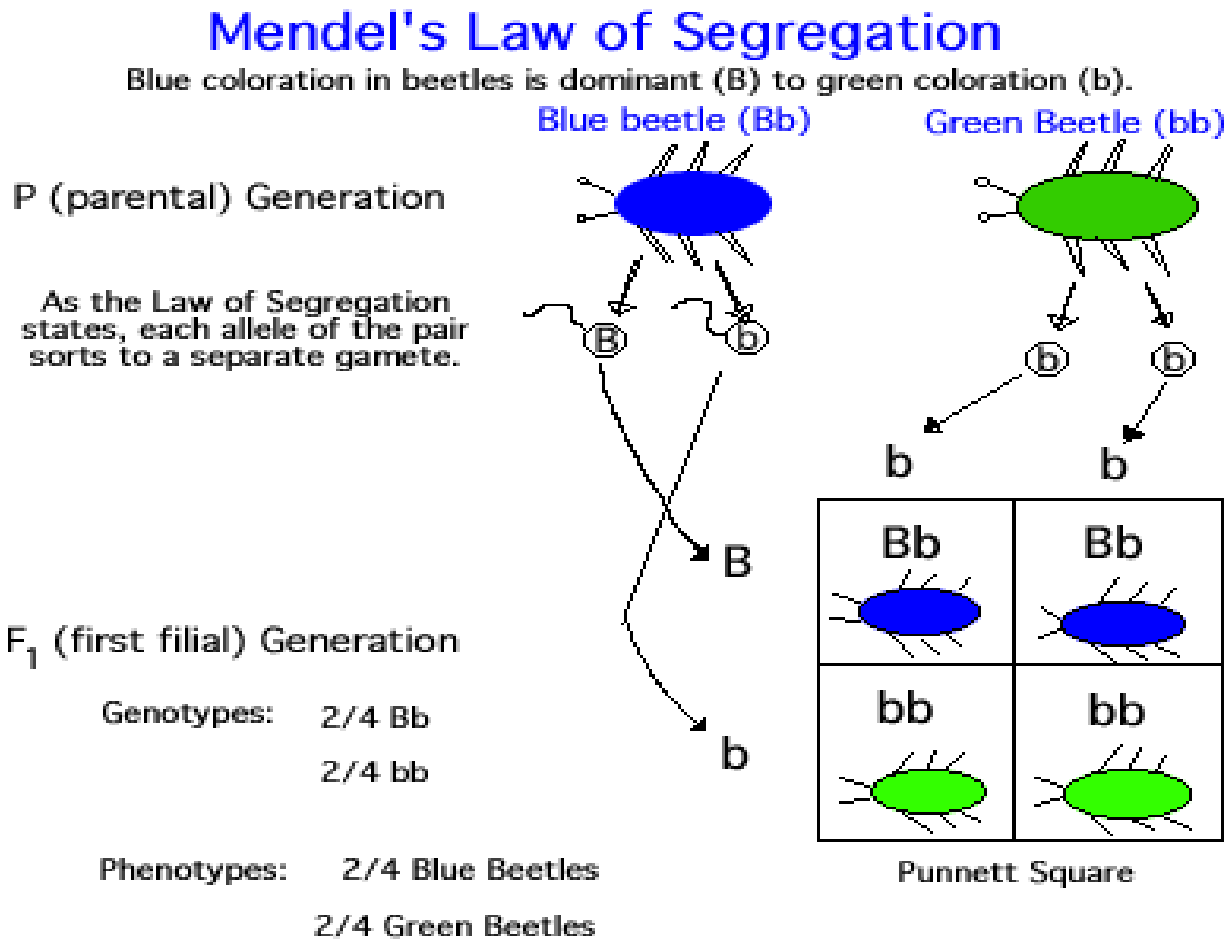


Genotype-What alleles an individual has for a particular marker or gene at a given locus.

Allele1	ACGT **CA CA CA CA CA CA** GGCGA
Allele2	ACGT **CA CA CA CA CA** GGCGA
Allele3	ACGT **CA CA CA CA** GGCGA
Allele4	ACGT **CA CA CA** GGCGA

Homozygous- Both alleles for a marker/gene at a specific locus are identical.

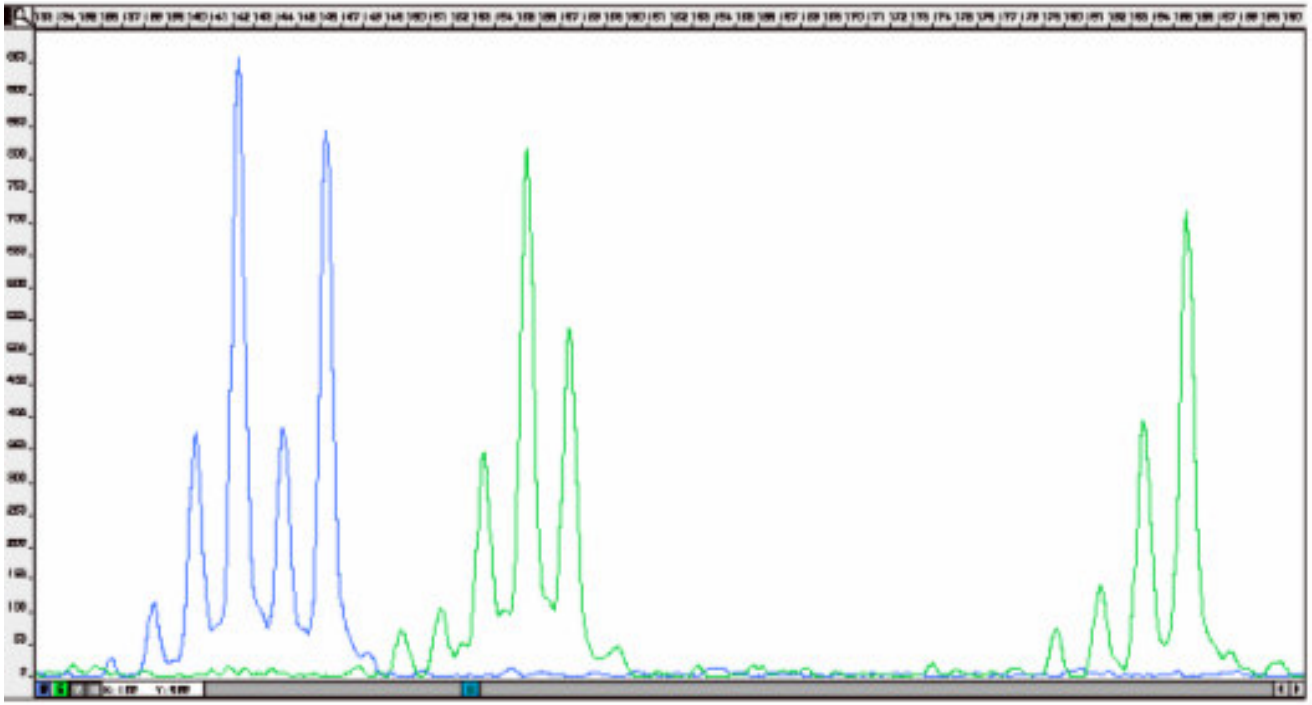
Heterozygous- Both alleles for a marker/gene at a specific locus are different.



Dinucleotide Repeats

5’ CACACACACACACACACACA 3’

5’ CACACACACACACACACACACACACACACACACACA 3’

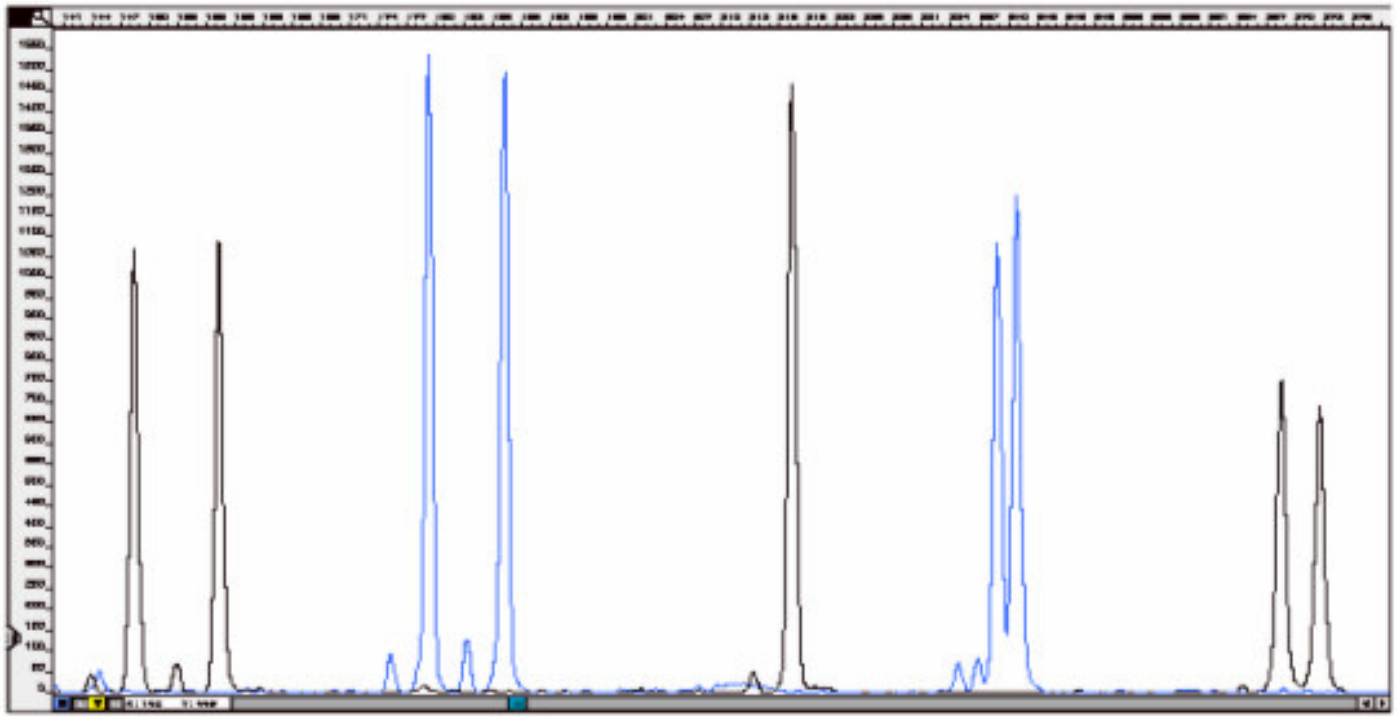


Heterozygote: **Heterozygote:** **Homozygote**

Tetranucleotide Repeats

5’TGAATGAATGAATGAATGAA 3’

5’TGAATGAATGAATGAATGAATGAATGAATGAA 3’



Heterozygote: **Heterozygote:** **Homozygote** **Heterozygote:** **Heterozygote:**

Tetranucleotide Repeats

5’TGAATGAATGAATGAATGAA 3’

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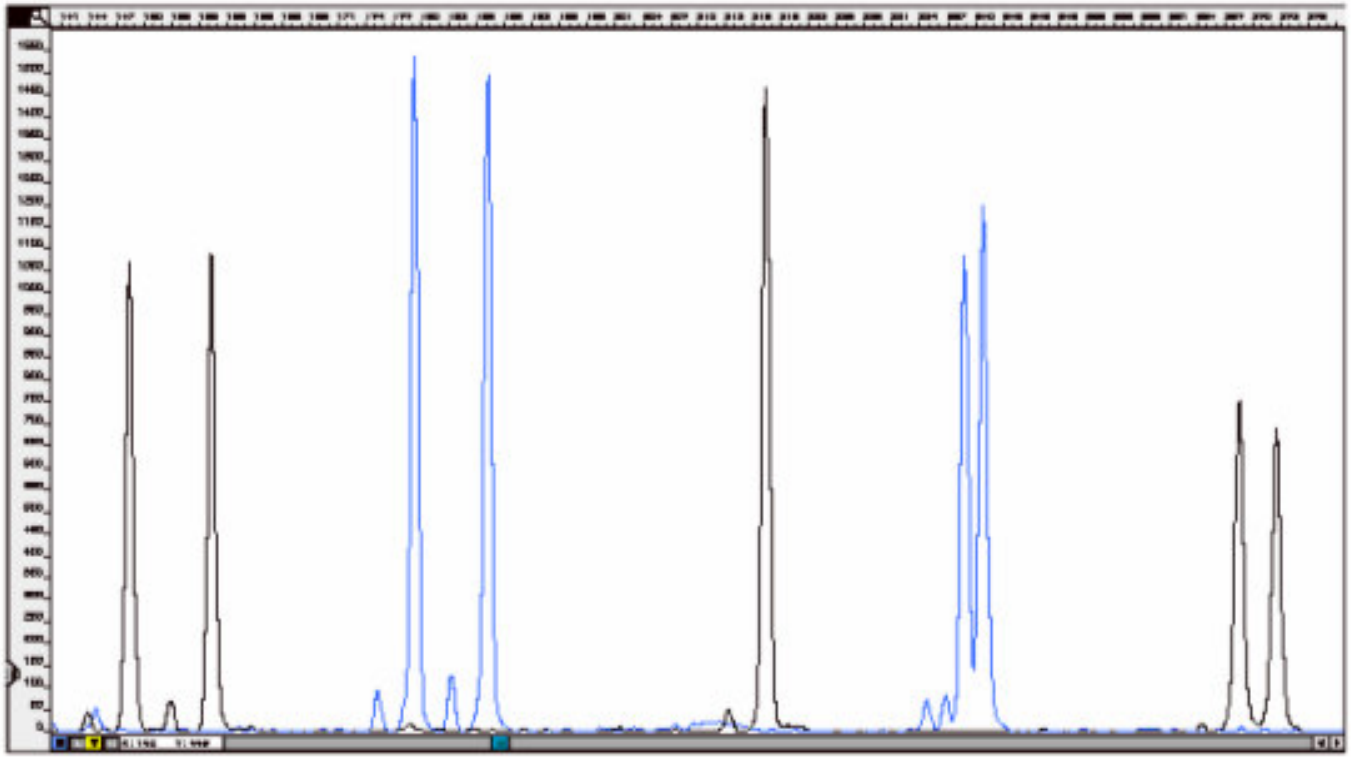

Heterozygote: **Heterozygote:** **Homozygote** **Heterozygote:** **Heterozygote:**

Genetic Analyzer [Capillary Electrophoresis]

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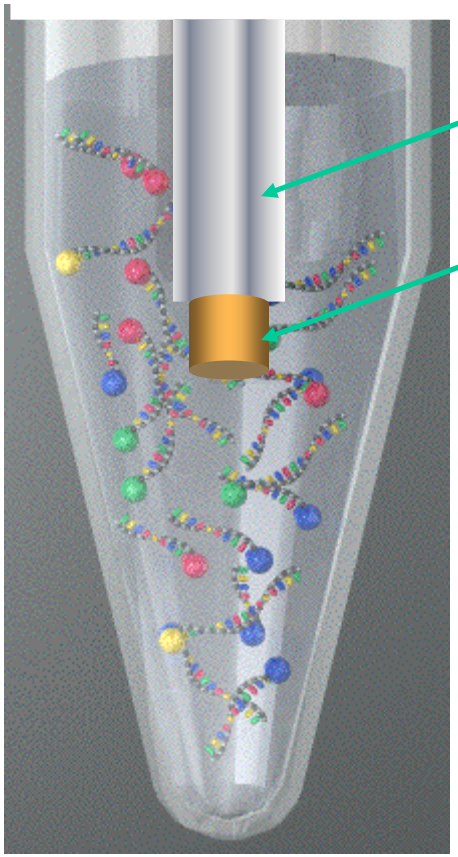
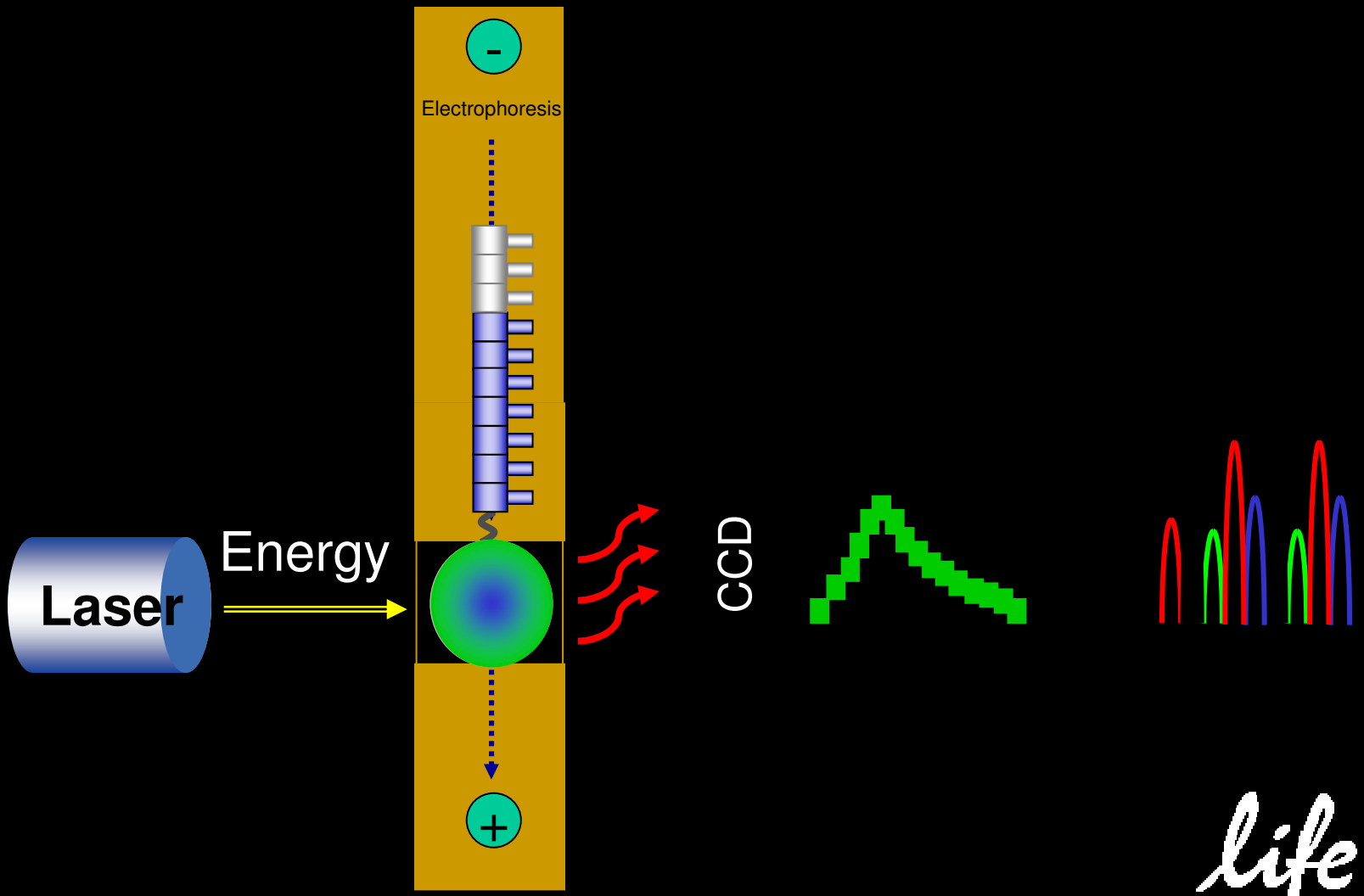
Electrokinetic Injection



- Capillary and electrode (cathode) are placed into the sample
- Voltage is applied
- Negatively-charged DNA enters the capillary as it migrates toward the postively-charged electrode (anode) at the other end of the capillary

Fluorescent Signal Detection



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Forensic Science:

the application of natural sciences to matters of the law.

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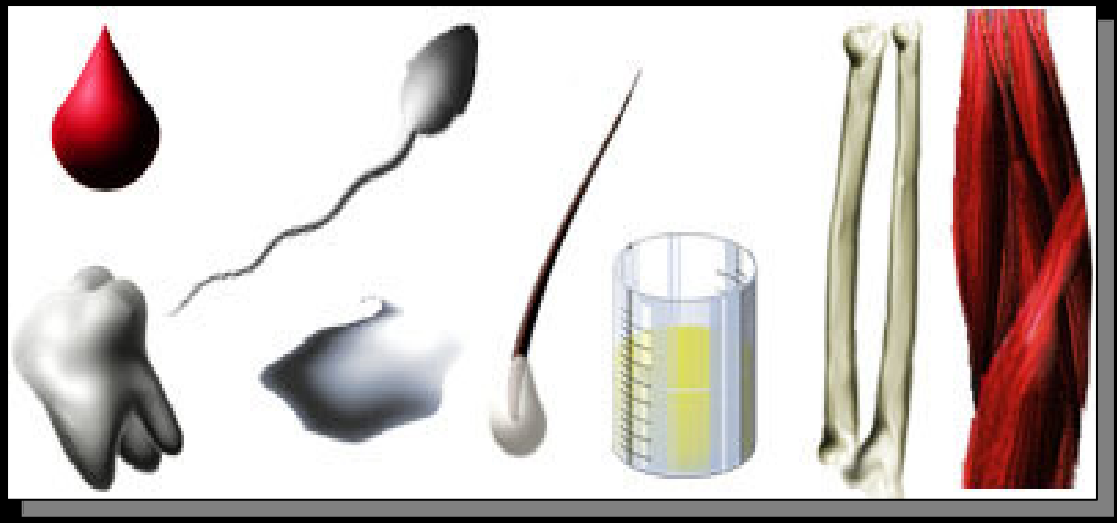
Sources of Biological Evidence

- Blood
- Semen
- Saliva
- Urine
- Hair
- Teeth
- Bone
- Tissue
- All cells – except RBC



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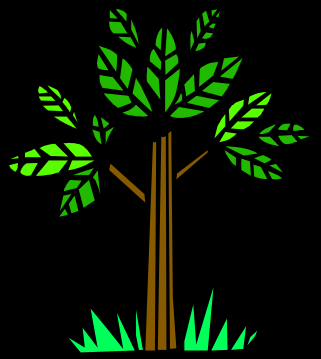
Other Possible items for DNA Testing:

1. cigarette butts

2. gloves, bandanas, ski masks, baseball caps
   general clothing



3. condoms (inside vs. outside)

4. stains on furniture, pillows, sheets

5. hair clips, lipsticks

6. letters, envelopes, and stamps

7. plant and animal sources of evidence

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Locard's Principle of Exchange

Anytime there is contact between two

surfaces, there will be a mutual exchange of matter across the contact boundary

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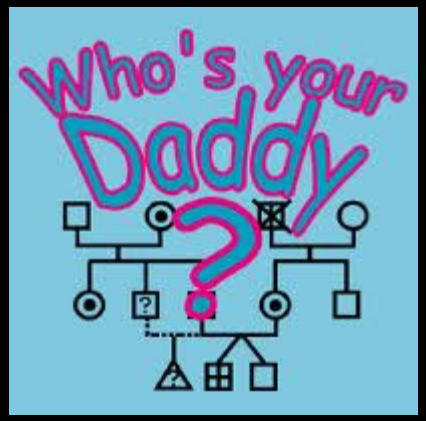
Forensic DNA Paternity Testing



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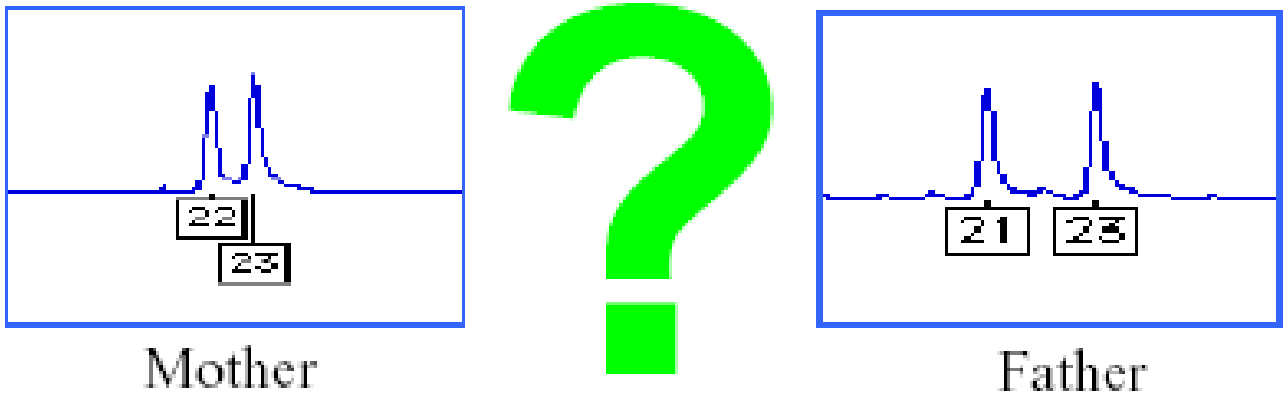
Paternity Testing



Mother

Father

Child

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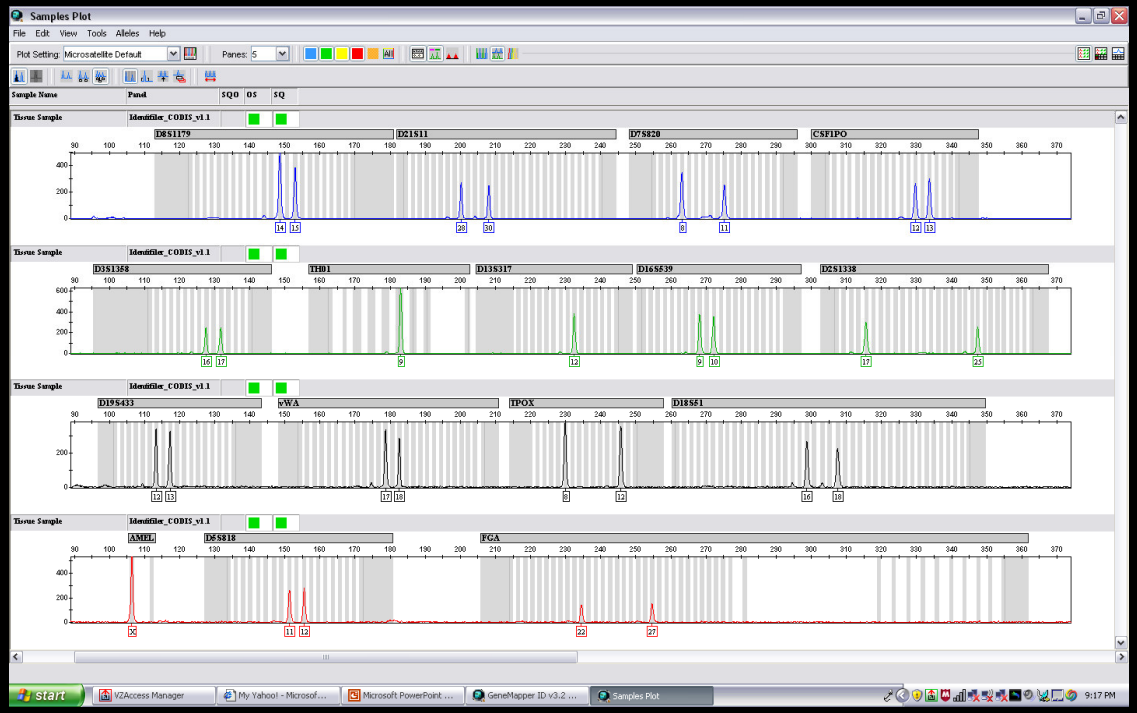
How do we go from this . . .

. . . To this?



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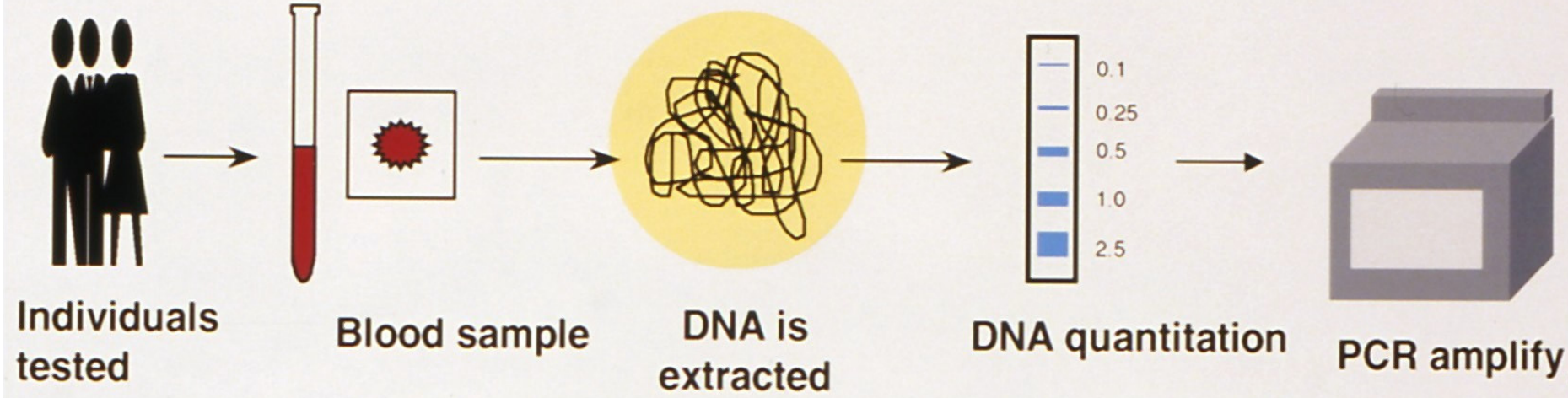
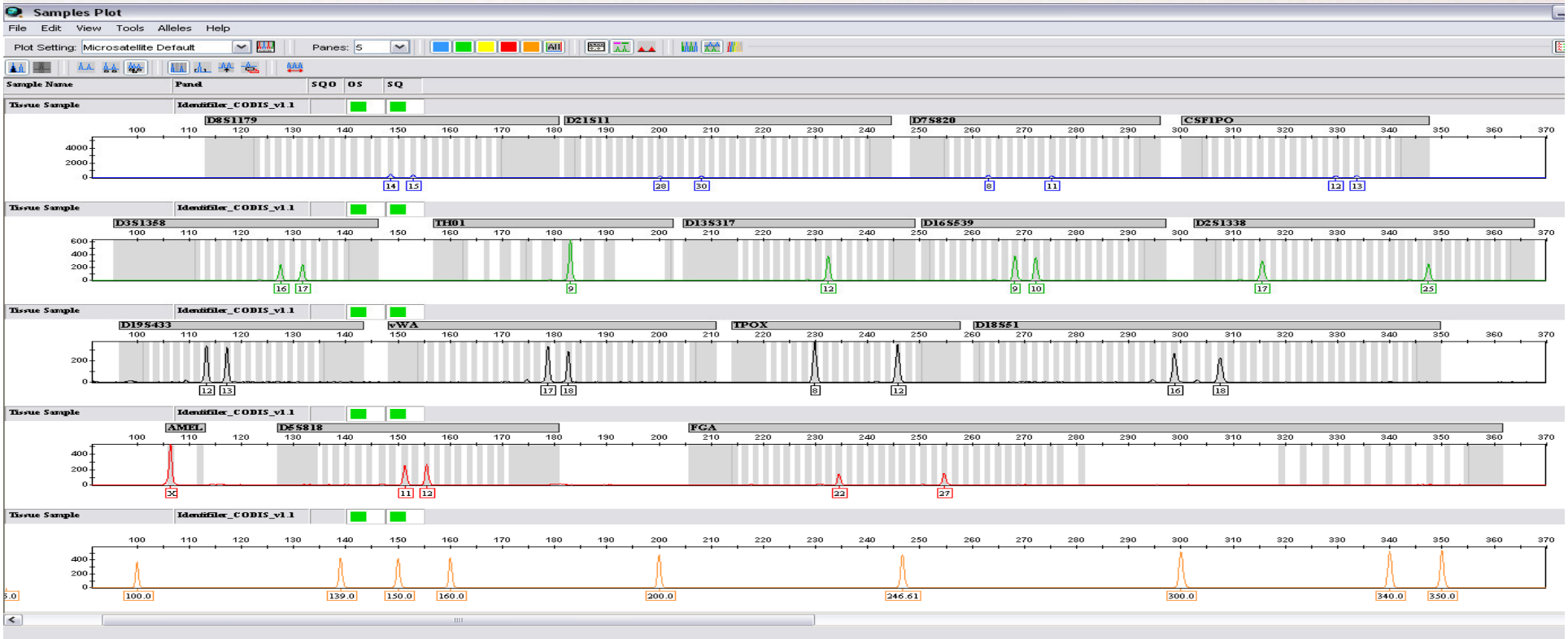
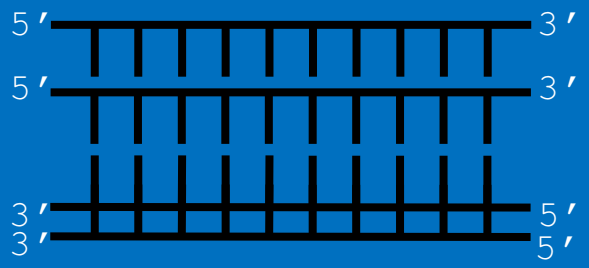
DNA analysis using STRs and the DNA Database





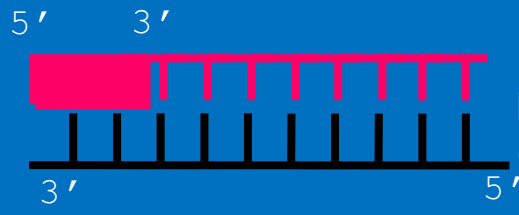
DNA Amplification with the Polymerase Chain Reaction (PCR)



Starting DNA Template

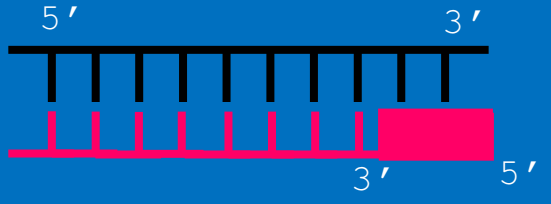
Separate strands (denature)

Forward primer



Make copies (extend primers)

Add primers (anneal)



Reverse primer

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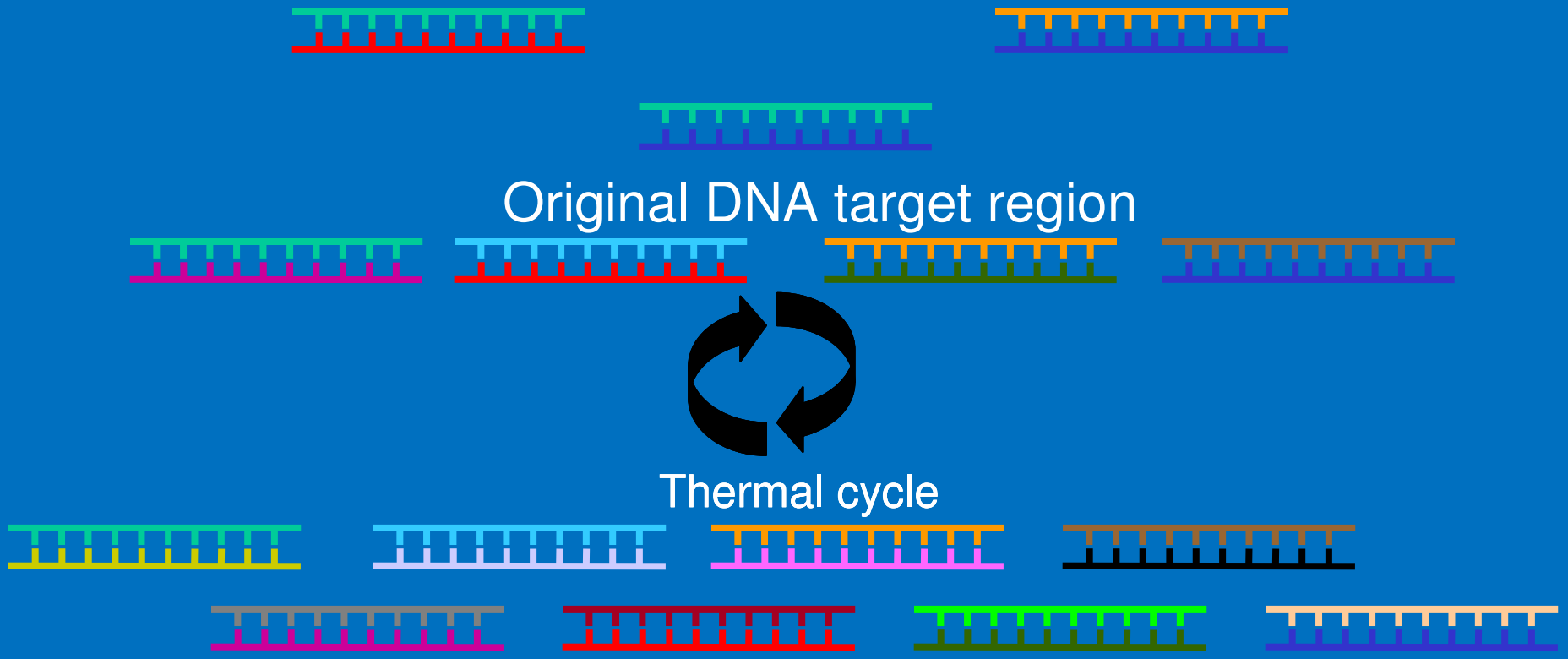
PCR Copies DNA Exponentially through Multiple Thermal Cycles



In 32 cycles at 100% efficiency, 1.07 billion copies of targeted DNA region are created

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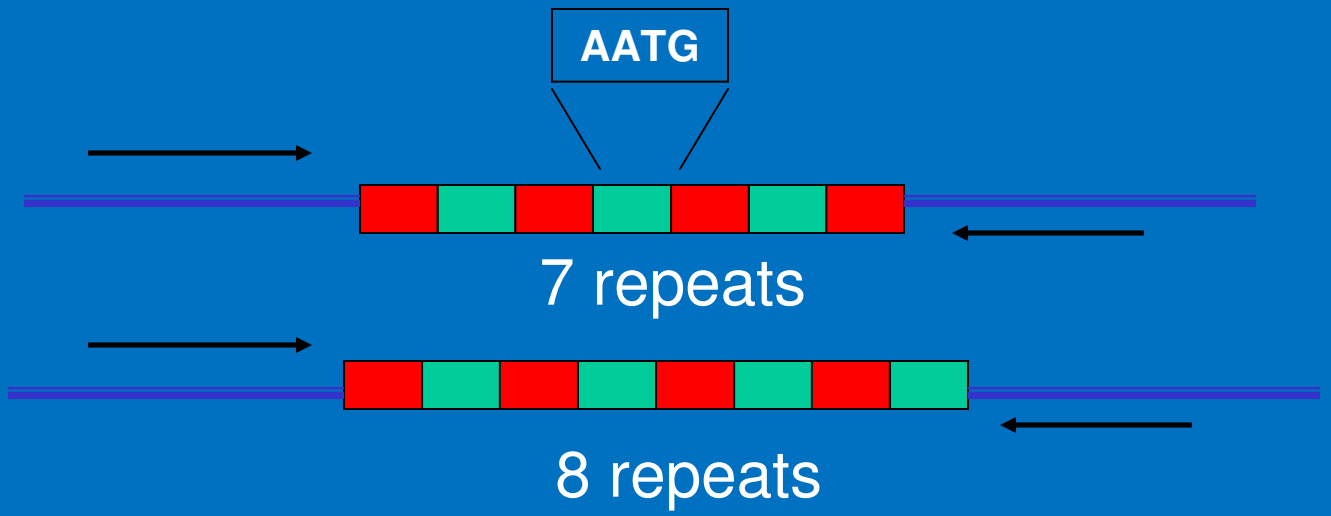
Short Tandem Repeats (STRs)



Repeat number varies between alleles. PCR primers bind to flanking regions that are constant.

Homozygote = Two copies of same allele.

Heterozygote = Two different alleles.

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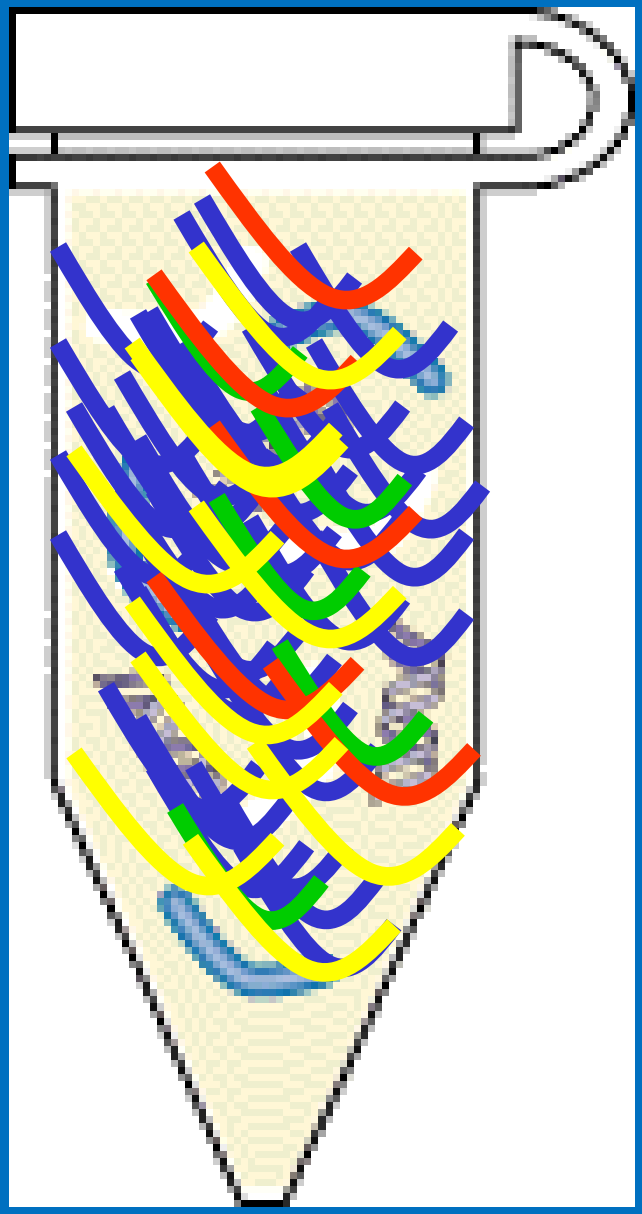

Multiplex PCR

- 15 STR Markers Can Be Amplified in 1 reaction.
- Sensitivity = less than 250 pg of DNA.
- Ability to Handle Mixtures and Degraded Samples.
- Different Fluorescent Dyes Used to Distinguish STR Alleles with Overlapping Size Ranges.

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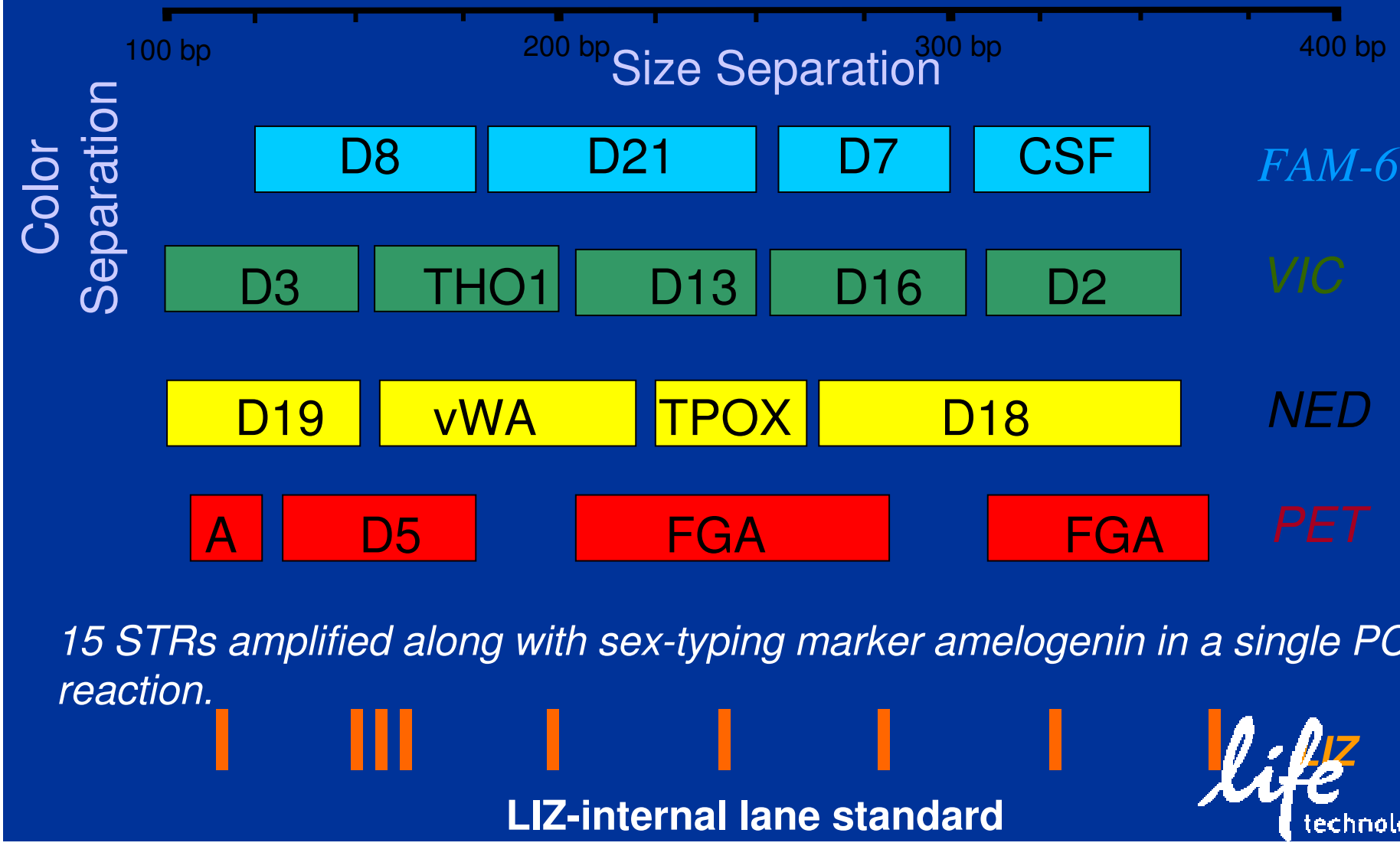
Example of Forensic STR Multiplex Kit

AmpFISTR® Identifiler™

Kit available from PE Biosystems (Foster City, CA)



15 STRs amplified along with sex-typing marker amelogenin in a single PCR reaction.

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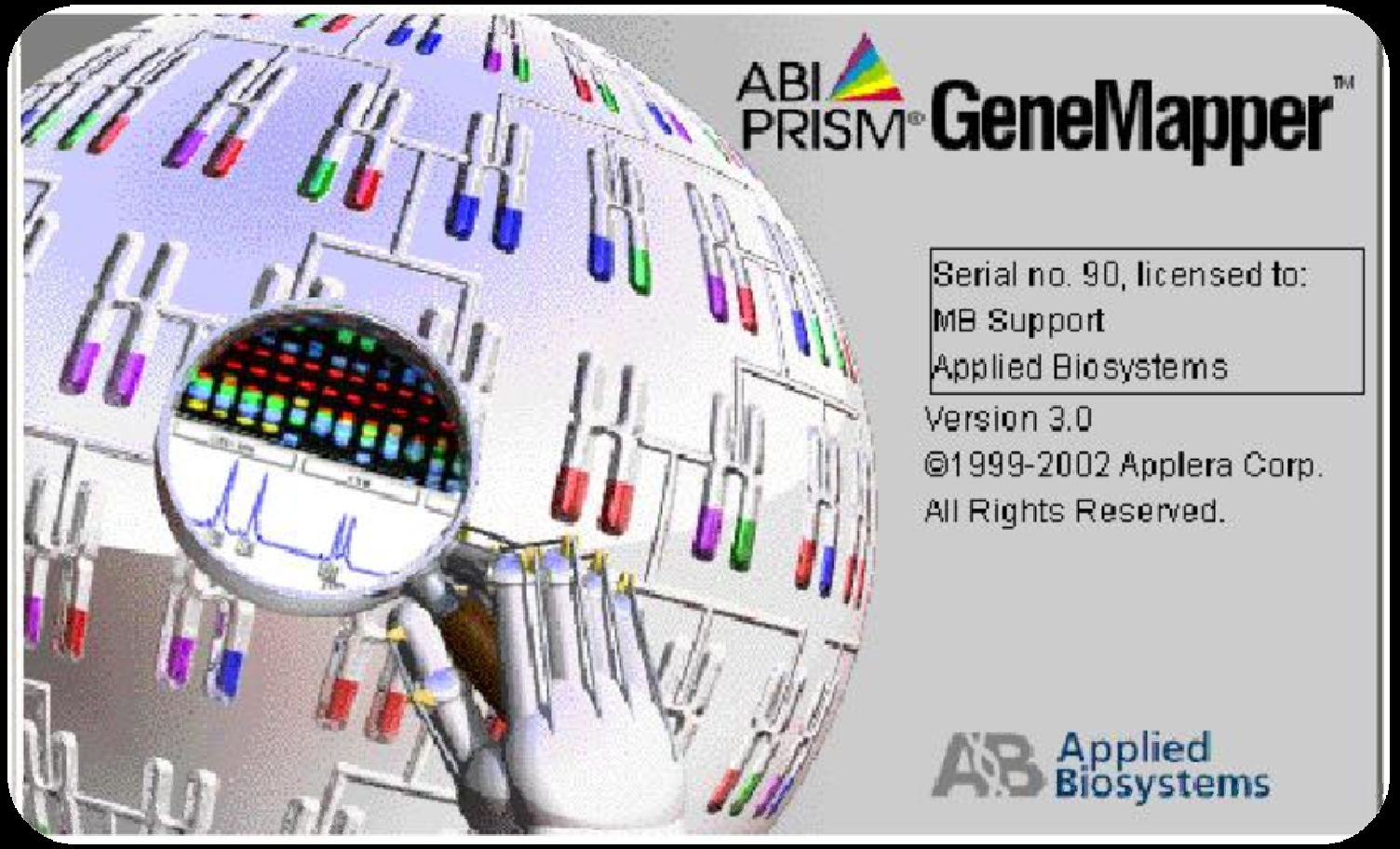
GeneMapper Software



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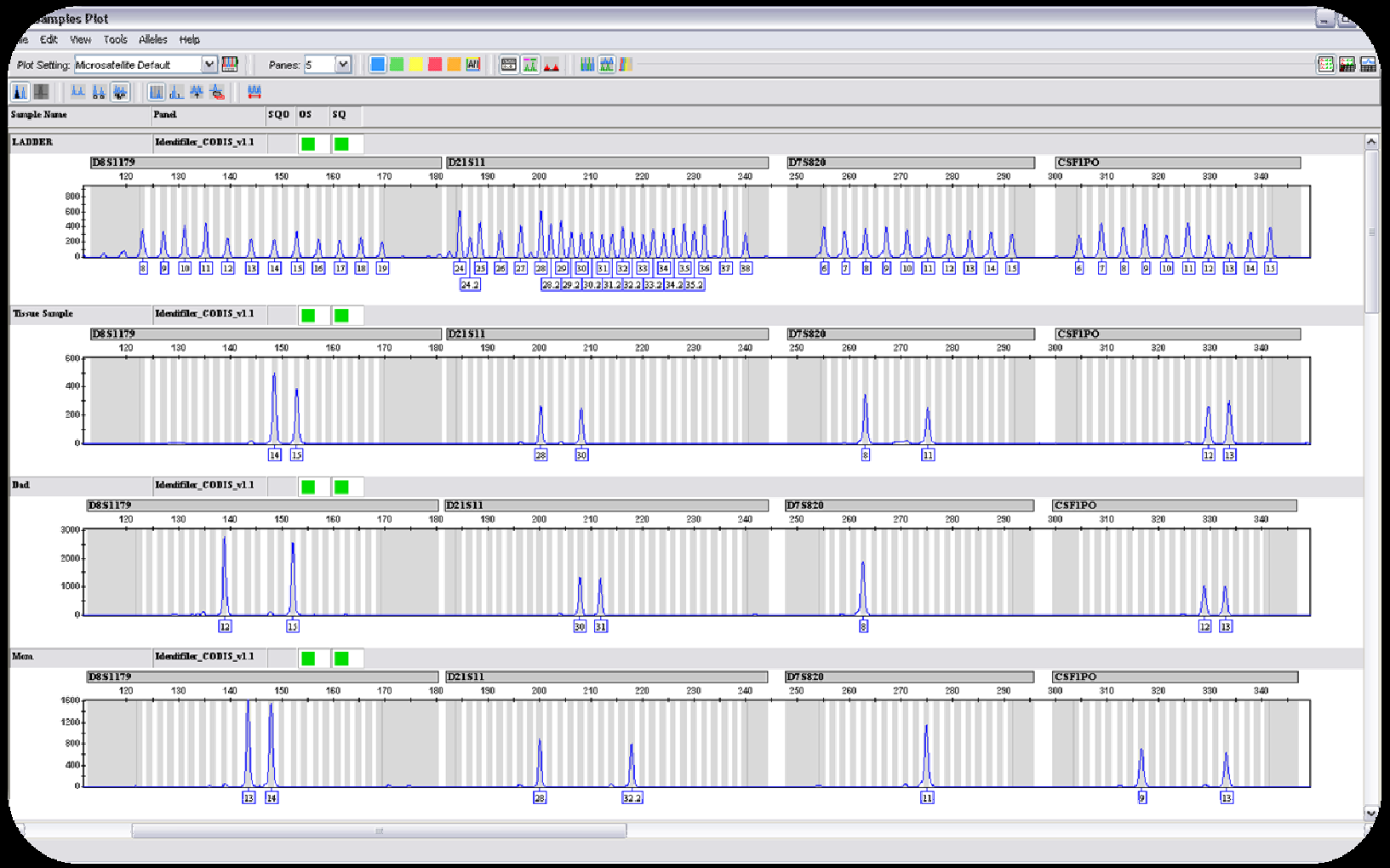
Forensic DNA Analysis



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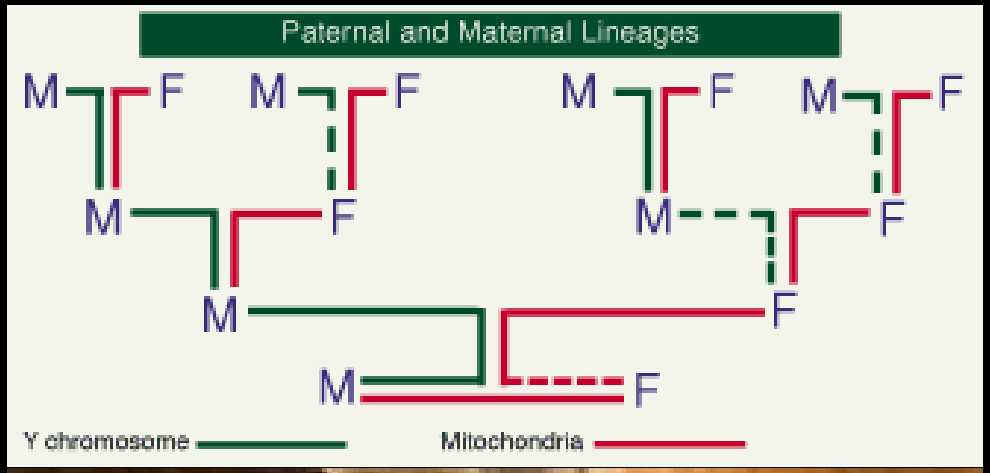
Y-DNA Typing



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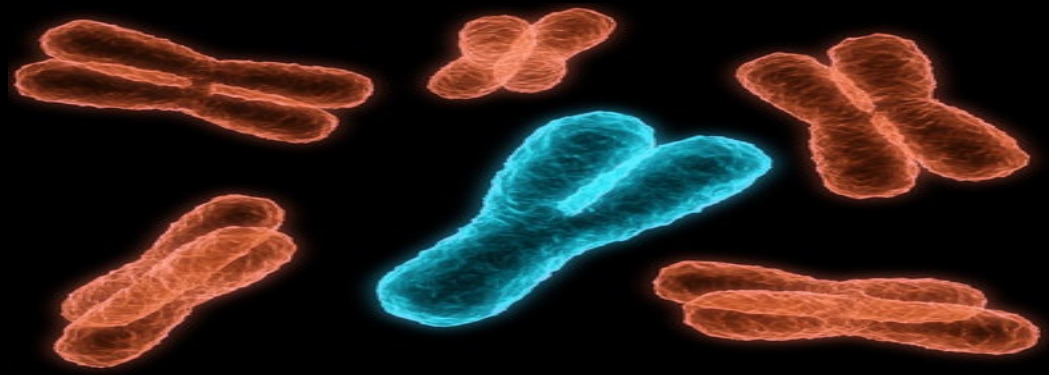
Y Chromosome Testing



- Although 17 loci are typed Paternal inheritance.
- Detects male component of a mixture.
- Less discriminating than standard DNA testing. Statistics = counting method (linkage).
- Important for detecting the semen donor in sexual assault mixtures.

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Modern Use of Y-STR Testing

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

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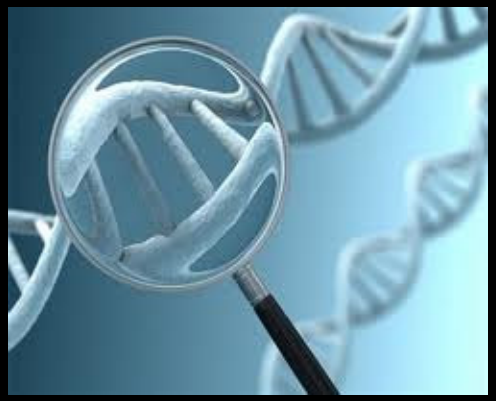
Any Questions…



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