

Brief History of Forensic DNA Typing

- 1980 - Ray White describes first polymorphic RFLP marker
- 1985 - Alec Jeffreys discovers multilocus VNTR probes
- 1985 - first paper on PCR
- 1988 - FBI starts DNA casework
- 1991 - first STR paper
- 1995 - FSS starts UK DNA database
- 1998 - FBI launches CODIS database

DNA Use in Forensic Cases

- Most are rape cases (>2 out of 3)
- Looking for match between evidence and suspect
- Must compare victim's DNA profile

Challenges

- Mixtures must be resolved
- DNA is often degraded
- Inhibitors to PCR are often present

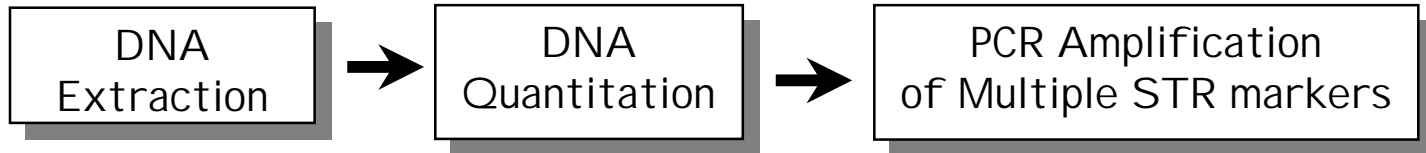
Human Identity Testing

- Forensic cases -- **matching suspect with evidence**
- Paternity testing -- **identifying father**
- Historical investigations
- Missing persons investigations
- Mass disasters -- **putting pieces back together**
- Military DNA “dog tag”
- Convicted felon DNA databases

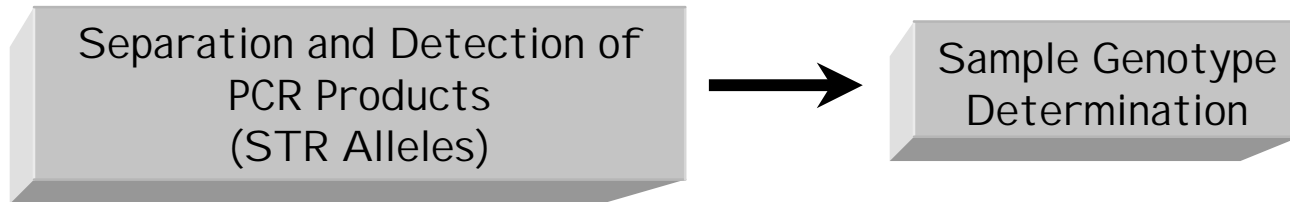
Steps in DNA Sample Processing

Sample Obtained from
Crime Scene or Paternity
Investigation

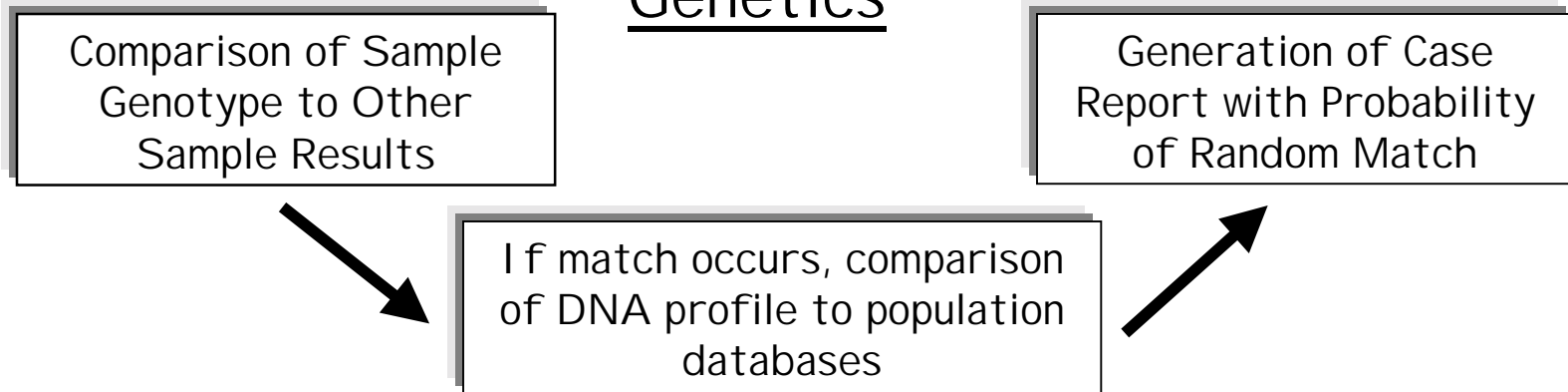
Biology



Technology



Genetics

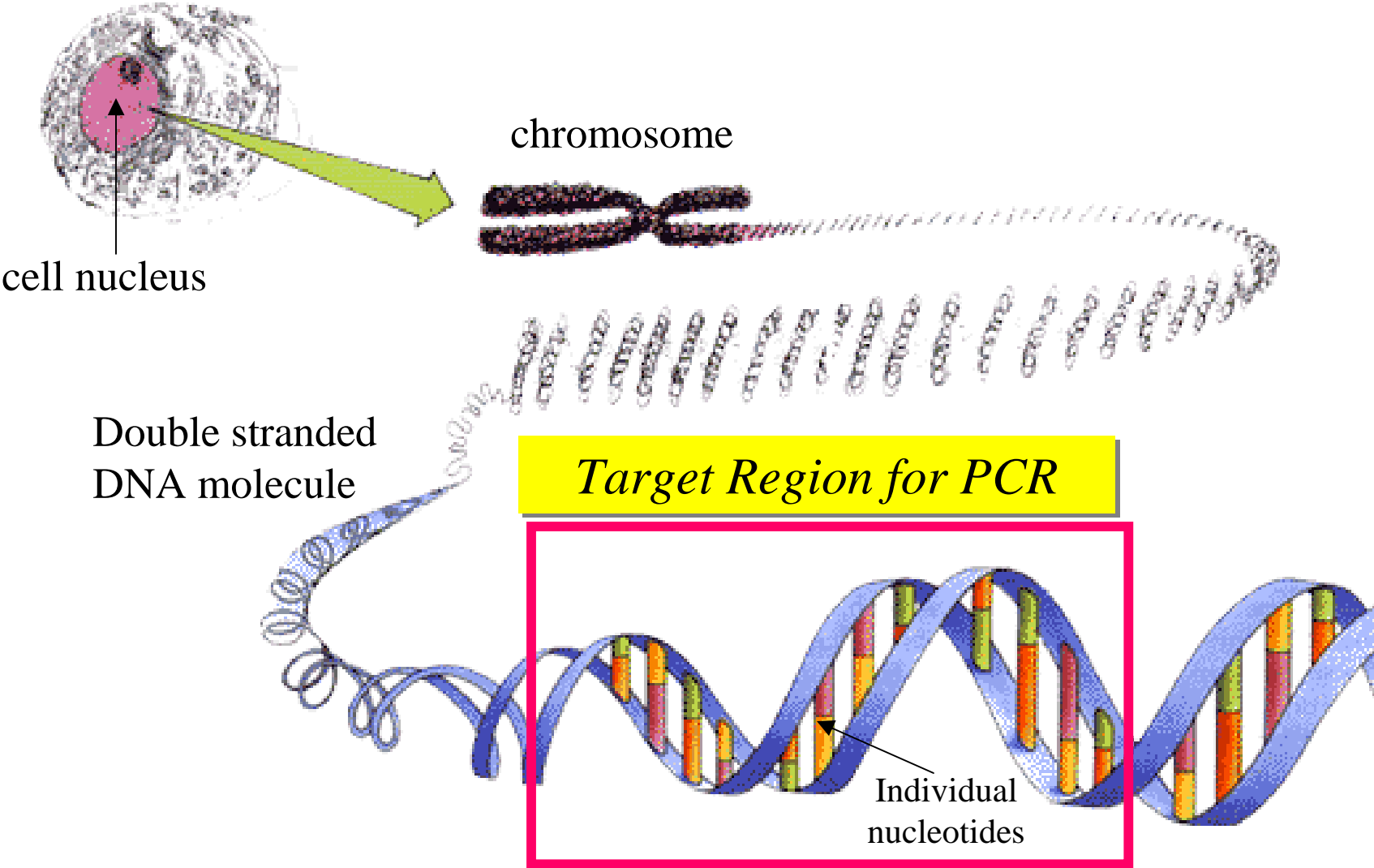


Sources of Biological Evidence

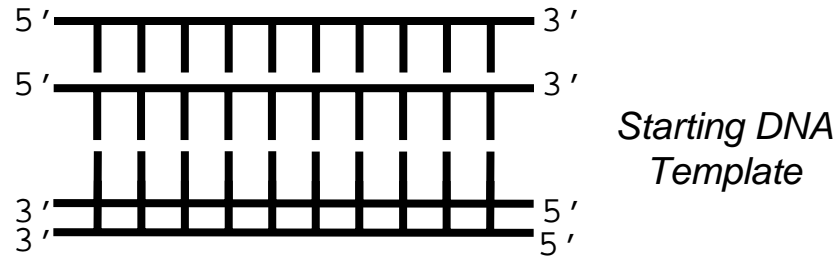
- **Blood**
- **Semen**
- **Saliva**
- **Urine**
- **Hair**
- **Teeth**
- **Bone**
- **Tissue**



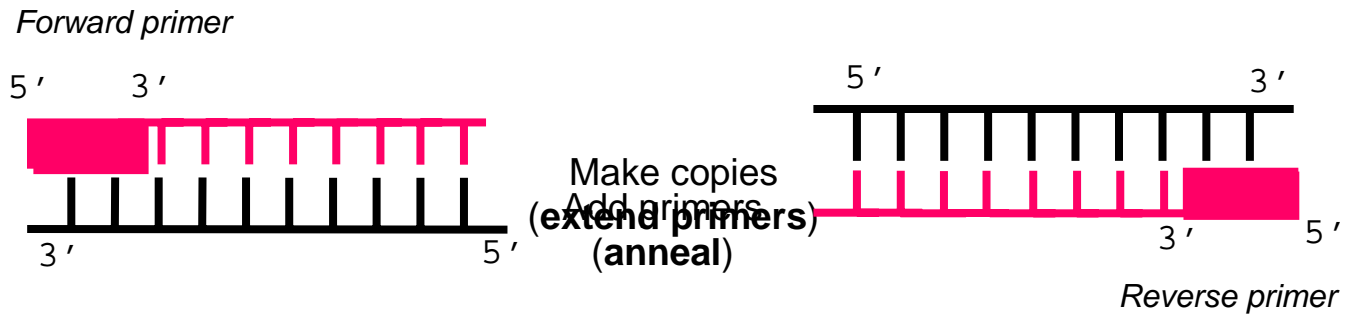
DNA in the Cell



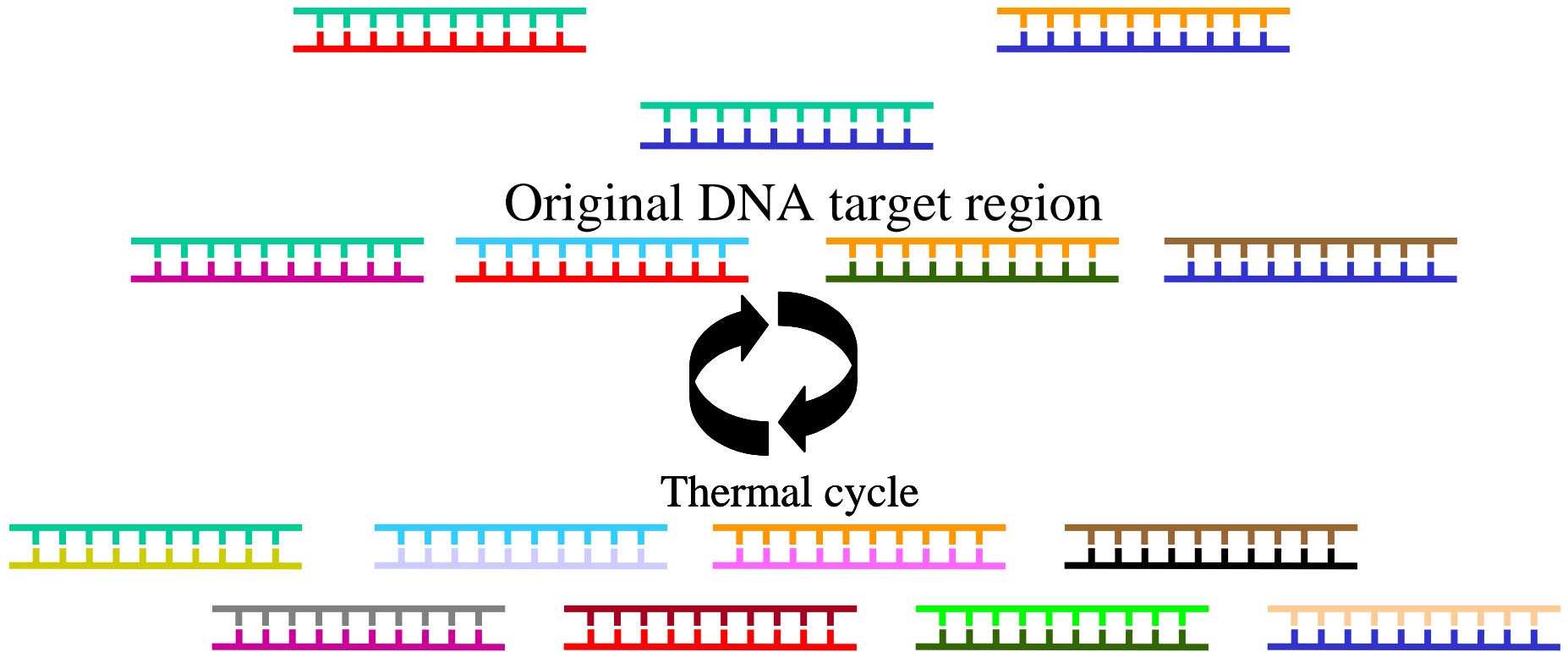
DNA Amplification with the Polymerase Chain Reaction (PCR)



Separate strands
(denature)

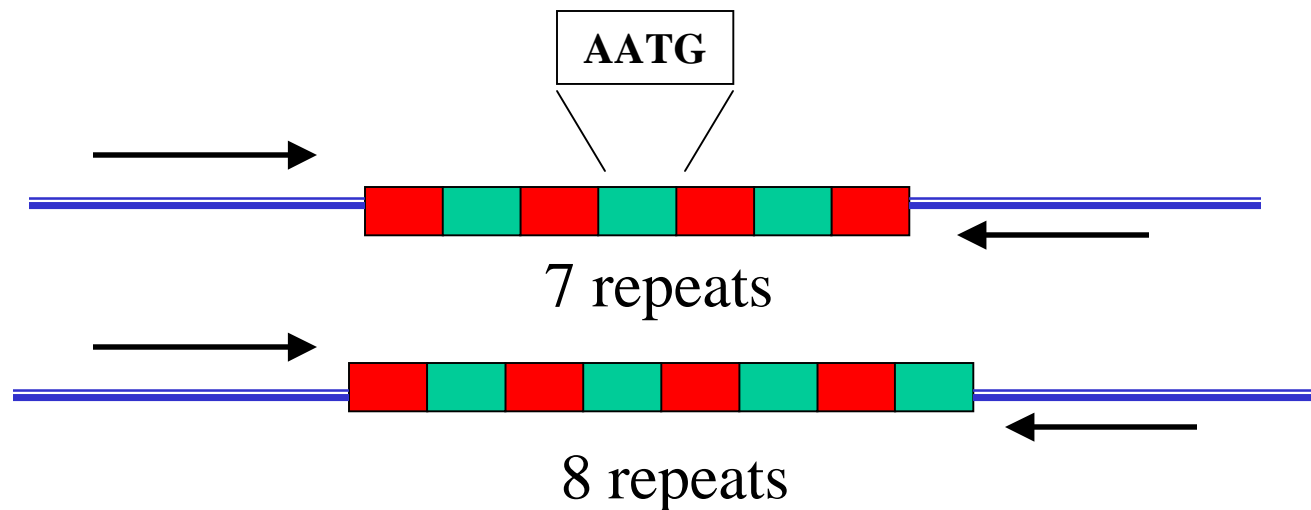


PCR Copies DNA Exponentially through Multiple Thermal Cycles



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

Short Tandem Repeats (STRs)

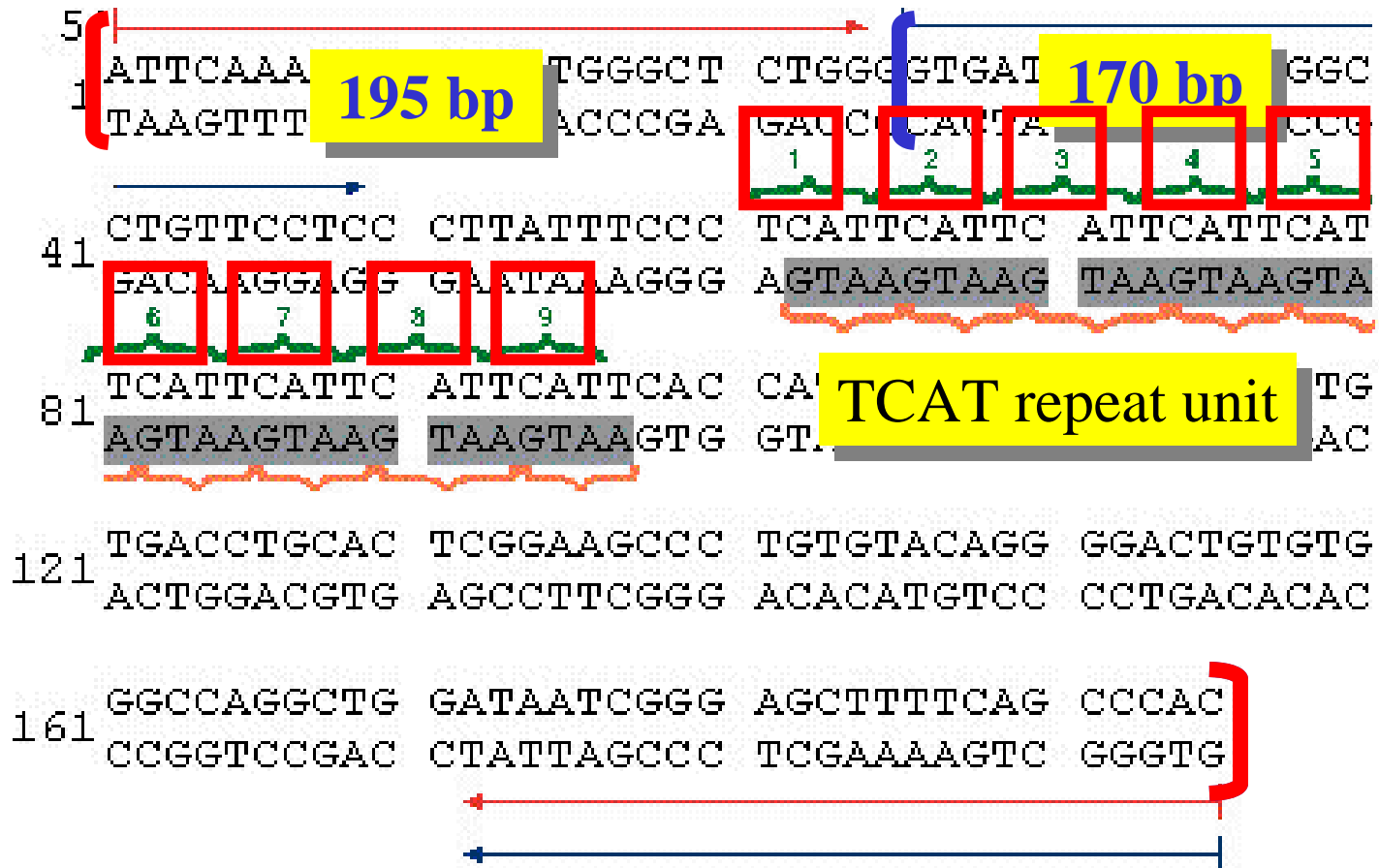


the repeat region is variable between samples while the flanking regions where PCR primers bind are constant

Homozygote = both alleles are the same length

Heterozygote = alleles differ and can be resolved from one another

HUMTH01 Sequence from GenBank (Accession D00269)



Different primer sets produce different PCR product sizes for the same STR allele



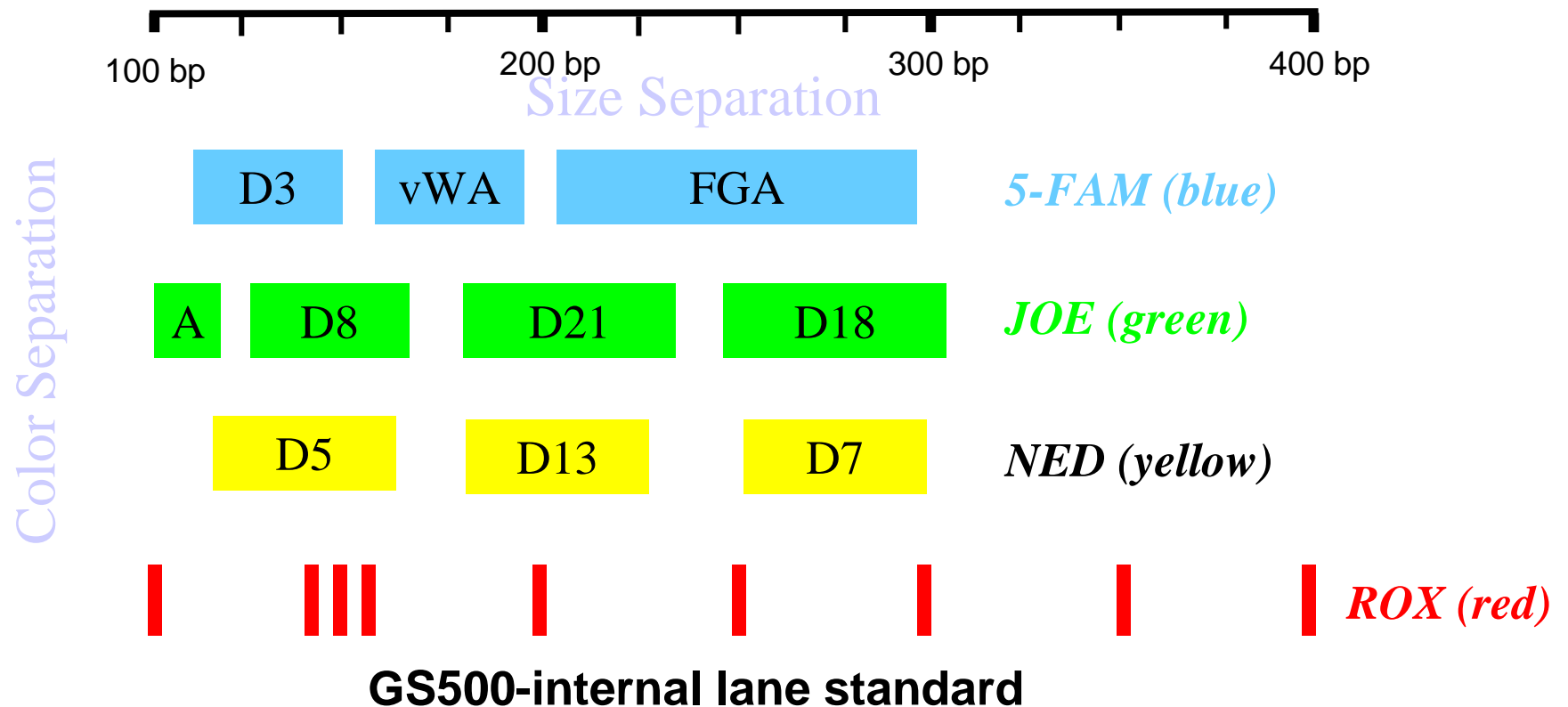
Multiplex PCR

- Over 10 Markers Can Be Copied at Once
- Sensitivities to levels less than 1 ng of DNA
- Ability to Handle Mixtures and Degraded Samples
- Different Fluorescent Dyes Used to Distinguish STR Alleles with Overlapping Size Ranges

An Example Forensic STR Multiplex Kit

AmpFISTR® Profiler Plus™

Kit available from PE Biosystems (Foster City, CA)

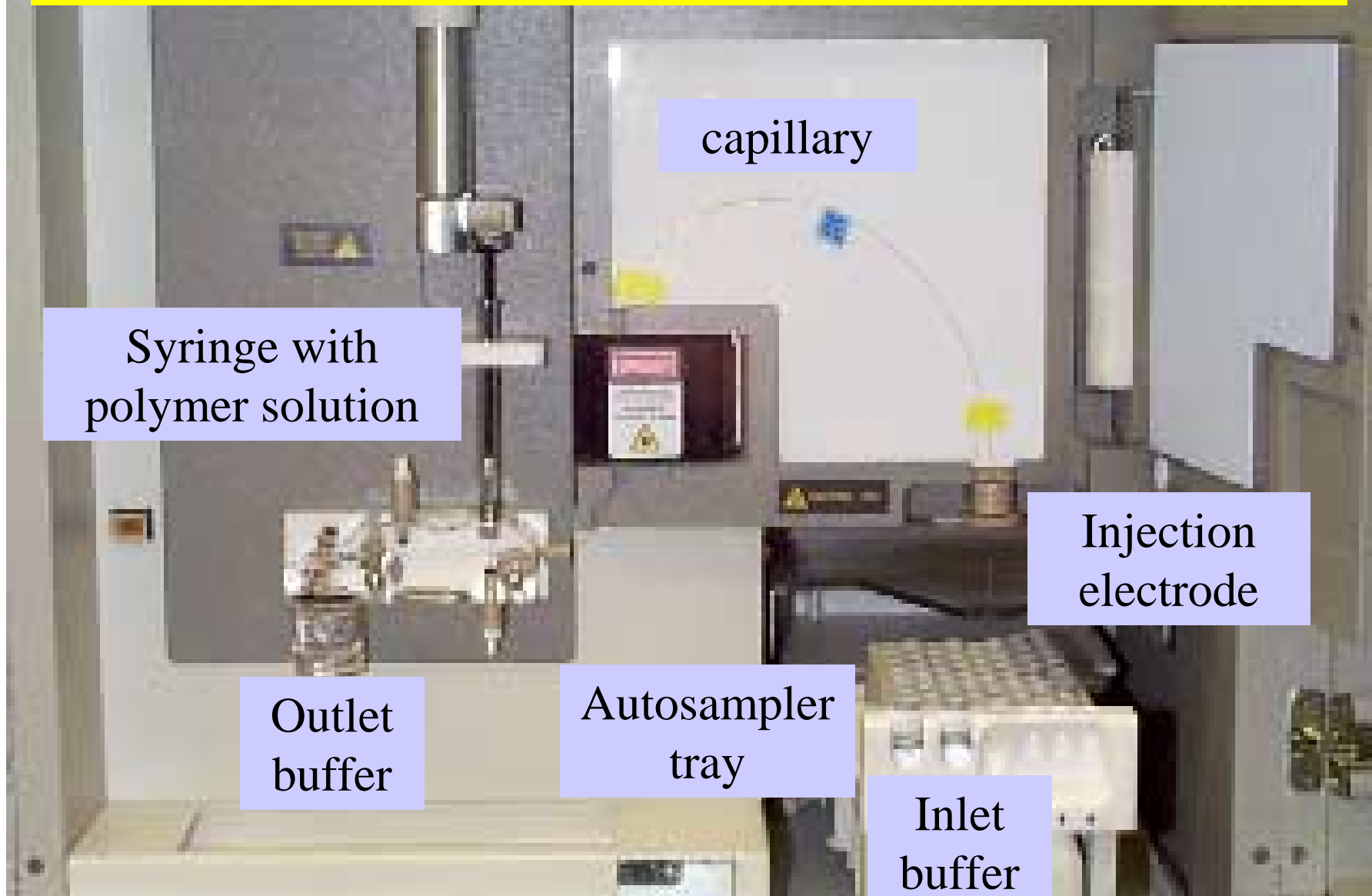


9 STRs amplified along with sex-typing marker amelogenin in a single PCR reaction

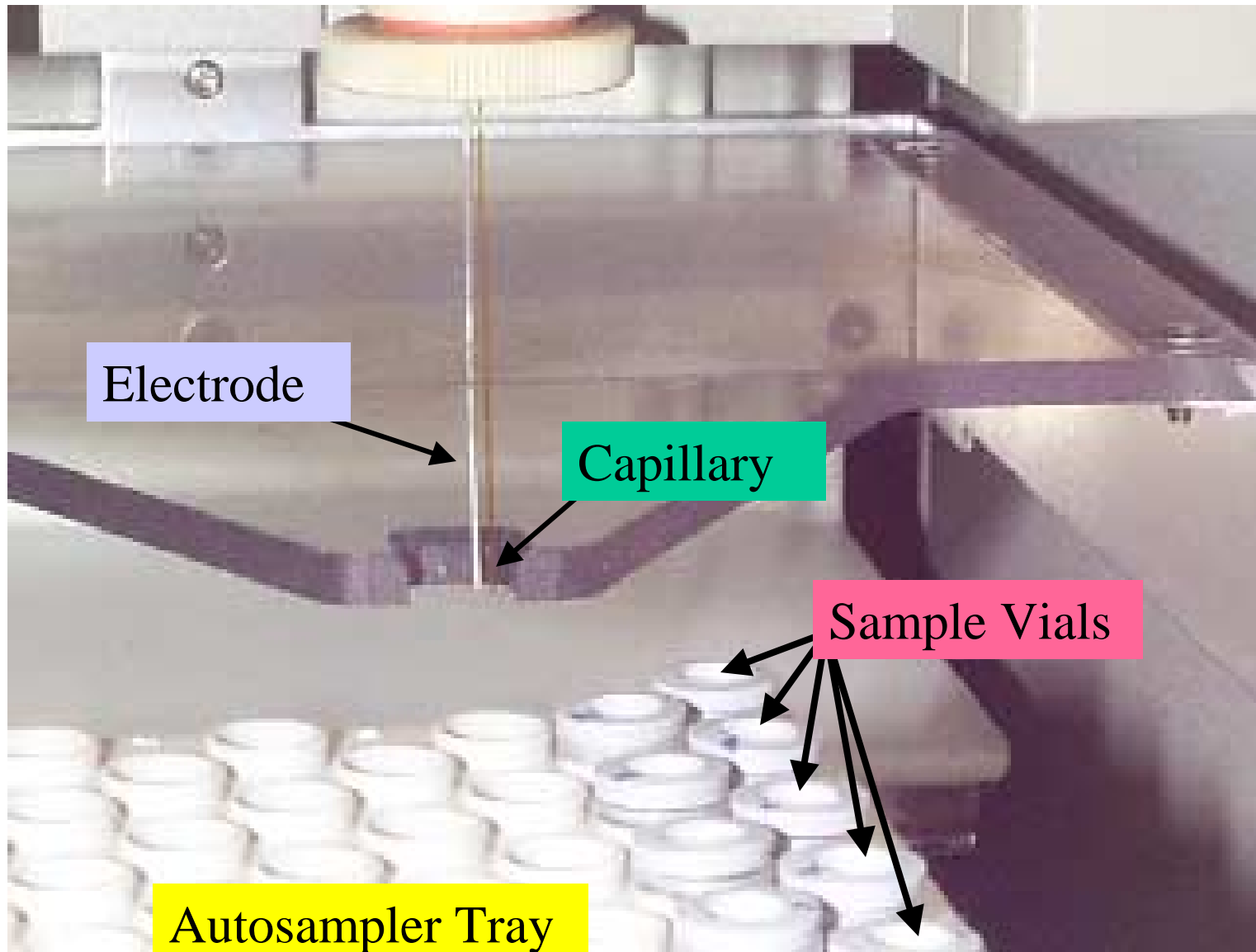
Available Kits for STR Analysis

- Kits make it easy for labs to just add DNA samples to a pre-made mix
- 13 CODIS core loci
 - Profiler Plus and COfiler (PE Applied Biosystems)
 - PowerPlex 1.1 and 2.1 (Promega Corporation)
- Increased power of discrimination
 - CTT (1994): 1 in 410
 - SGM Plus™ (1999): 1 in 3 trillion
 - PowerPlex™ 16 (2000): 1 in 2×10^{17}

ABI Prism 310 Genetic Analyzer



Close-up of ABI Prism 310 Sample Loading Area



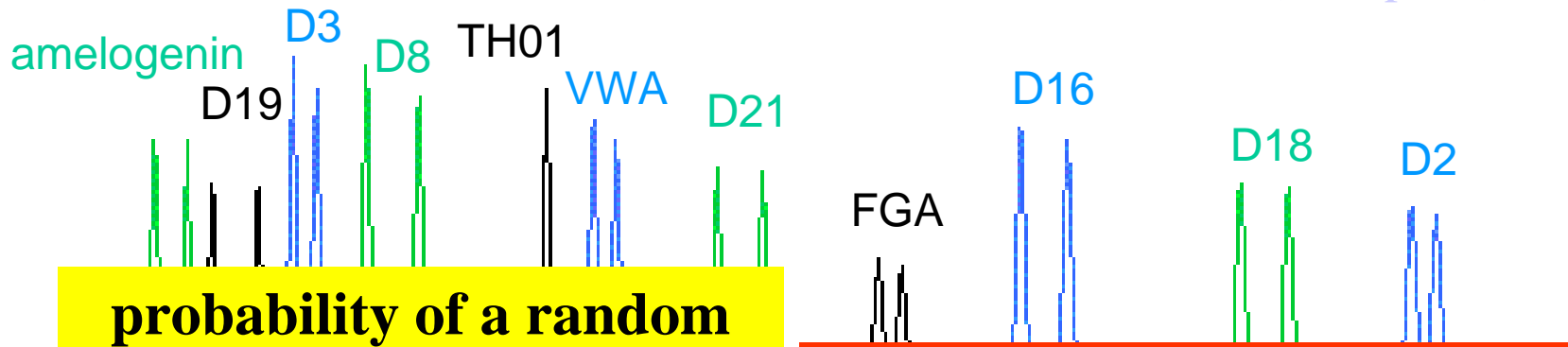
See Technology section for more information on CE

Human Identity Testing with Multiplex STRs

AmpFlSTR® SGM Plus™ kit

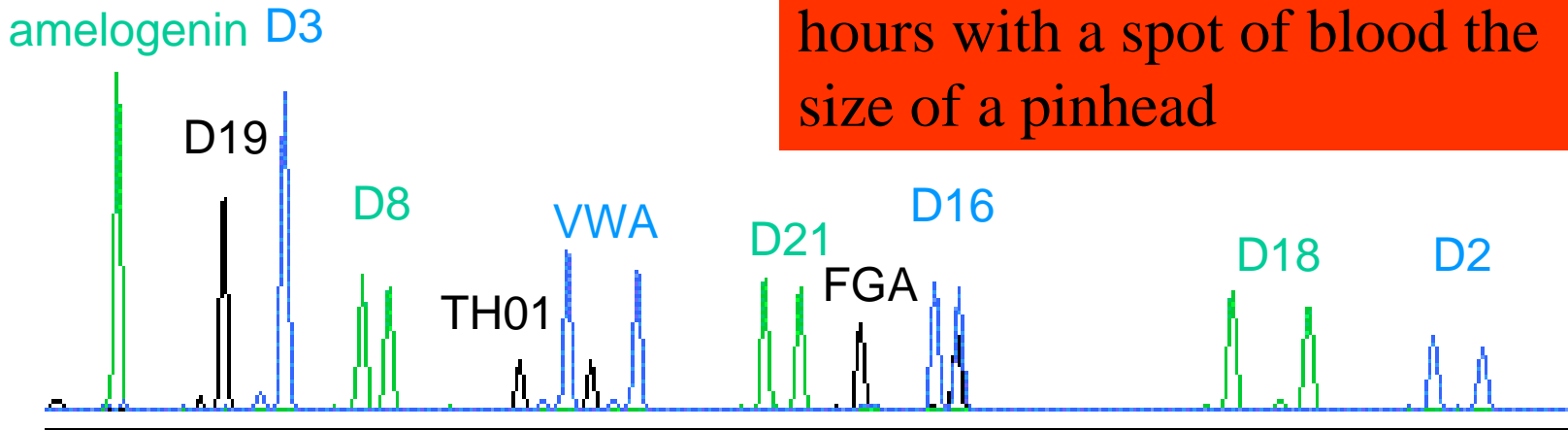


Two different individuals



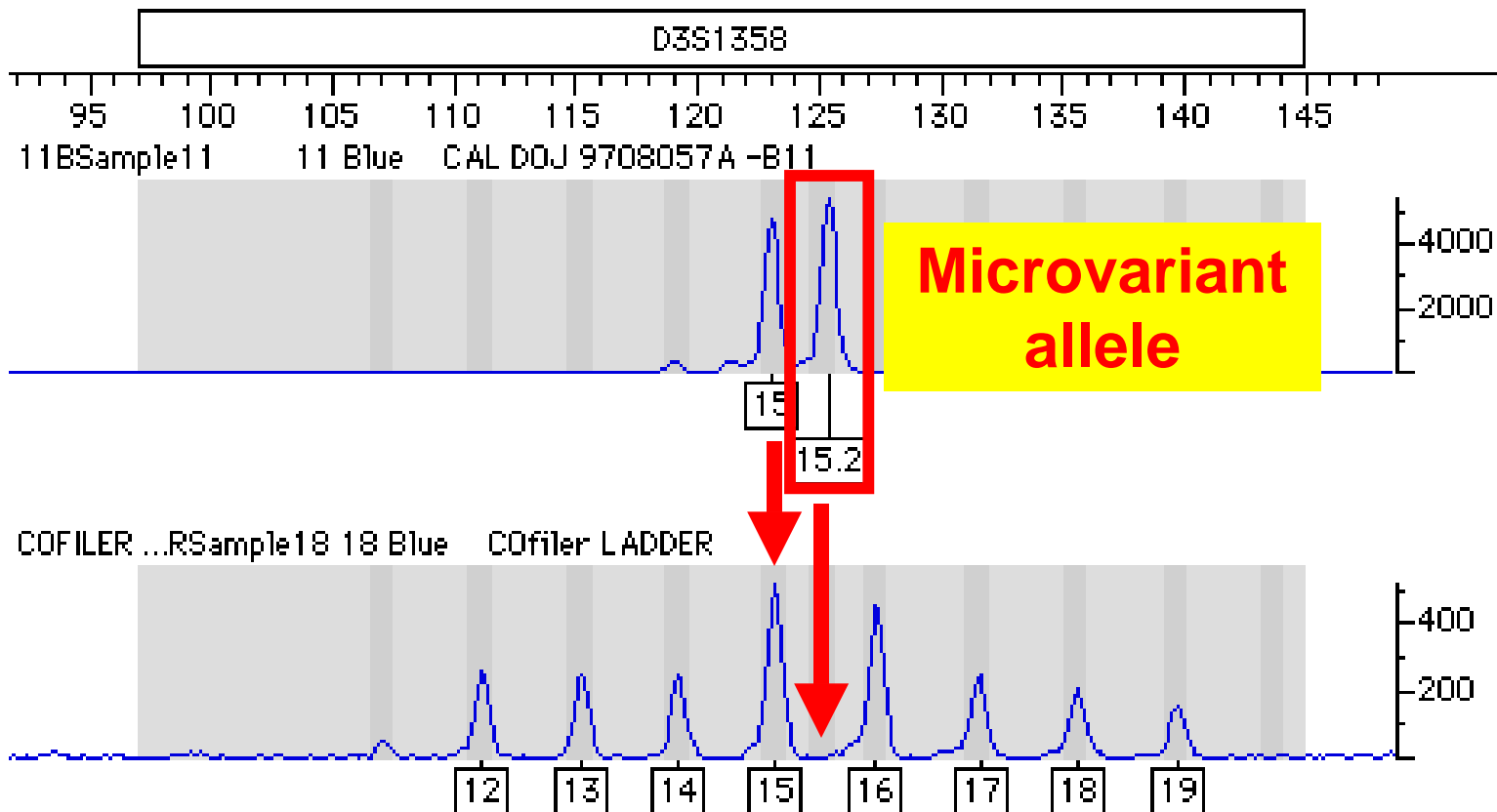
probability of a random match: ~1 in 3 trillion

Results obtained in less than 5 hours with a spot of blood the size of a pinhead

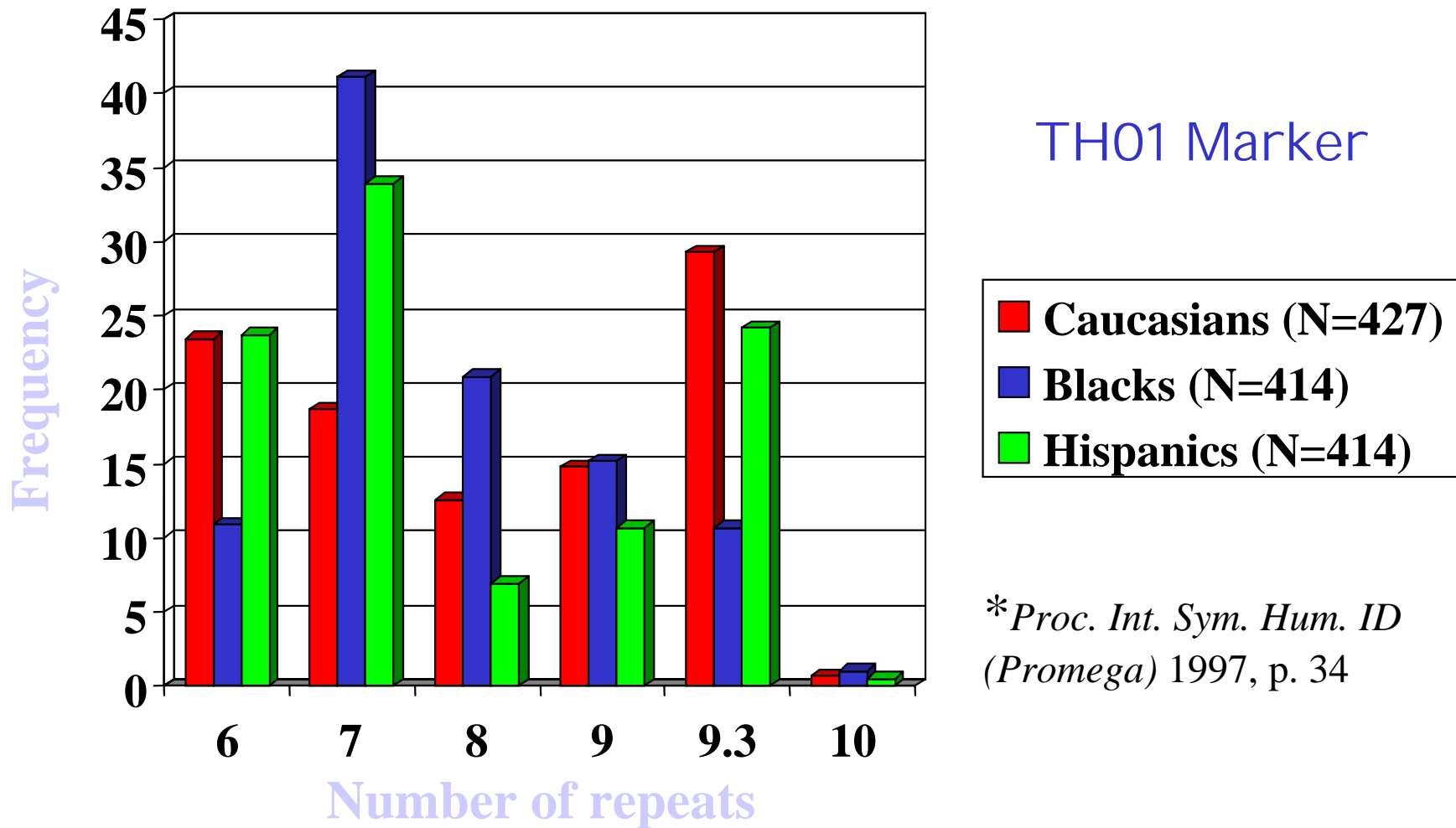


Simultaneous Analysis of 10 STRs and Gender ID

STR genotyping is performed by comparison of sample data to allelic ladders



STR Allele Frequencies



FBI's CODIS DNA Database

Combined DNA Index System

- Used for linking serial crimes and unsolved cases with repeat offenders
- Launched October 1998
- Links all 50 states
- Requires >4 RFLP markers and/or 13 core STR markers
- Current backlog of >600,000 samples



13 CODIS Core STR Loci with Chromosomal Positions

