Chapter 7  DNA Profiling

By the end of this chapter you will be able to:

7.1 Explain how DNA can be important to criminal investigations.

7.2 Explain how crime-scene evidence is collected for DNA analysis.

7.3 Describe how crime-scene evidence is processed to obtain DNA.

7.4 Explain what a short tandem repeat (STR) is, and explain its importance to DNA profiling.
Chapter 7  DNA Profiling

By the end of this chapter you will be able to:

7.5  Explain how law-enforcement agencies compare new DNA evidence to existing DNA evidence.

7.6  Describe the use of DNA profiling using mtDNA and Y STRs to help identify a person using the DNA of family members.

7.7  Compare and contrast a gene and a chromosome, and an intron and an exon.
Chapter 7
Vocabulary

- allele
- chromosome
- Combined DNA Index System (CODIS)
- DNA fingerprint (profile)
- electrophoresis
- exon
- gene
- genome
- intron

- karyotype
- polymerase chain reaction (PCR)
- polymorphism
- primer
- restriction enzyme
- restriction fragment
- short tandem repeats (STR)
Introduction

- Mid-1980s
  - DNA fingerprinting dramatically changed forensic science and the ability of law enforcement to link perpetrators with crime scenes.

- Early 1990s
  - The US military began collecting and storing blood samples of soldiers

- 1994
  - DNA Identification act
Introduction (continued)

- 1998
  - National DNA Index System (NDIS)
- CODIS (The Combined DNA Index System)
  - Collects, analyzes, and communicates criminal DNA information
- By 2014, CODIS had:
  - 11.1 million offenders its DNA profiles
  - 1.9 million arrestee DNA profiles
    - More than 257,000 hits assisting in more than 246,000 investigations
o Watson and Crick developed double helix model of DNA in 1953

o Sir Alec Jeffreys processed the first DNA used in a criminal case

o Colin Pitchfork was the first man convicted using DNA

o **Write this on the back of your notes.**
What is DNA?

- DNA deoxyribonucleic acid
  - The genetic material of all living things
  - With the exception of red blood cells, all human cells contain DNA.
- All the DNA found in human cells makes up the human genome.

Figure 7-1 A DNA nucleotide consists of a deoxyribose sugar, a phosphate group, and one of four nitrogenous bases.
Figure 7-2  Base pairs on the double helix of DNA.
Chromosomes

- The 46 chromosomes found in human body cells are composed of tightly coiled DNA.
- Most males have one X and one Y sex chromosomes (XY).
- Most females have two X chromosomes (XX).
- Half of your DNA is inherited from your mother and the other half is inherited from your father.
Figure 7-3  In this karyotype of human chromosomes, note the 22 homologous pairs of autosomes followed by the two sex chromosomes.
Genes

- A gene is a segment of DNA in a chromosome that serves in the production or regulation of proteins.
- Genes average about 3000 base pairs, but they can be composed of many thousands of base pairs.
Figure 7-4  The coding region of DNA known as a gene includes both exons and introns. Only the exons will be expressed. The introns will be deleted.
Collection and Preservation of DNA Evidence

- Using polymerase chain reactions (PCR), trace samples of DNA evidence can be amplified so that adequate amounts of DNA evidence are available for testing.
- Avoiding contamination when collecting, preserving, and identifying DNA evidence is critical.
- DNA evidence should be kept dry and cool during transportation and storage.
Forensic DNA and Personal Identification

- DNA profiling is the most useful tool for law enforcement since the development of fingerprinting.
- Scientists use polymorphisms to distinguish one person from another.
- Because 99% of all human DNA is the same, scientists only need to examine the 1% region of variability.
Early DNA Fingerprinting Using Gel Electrophoresis

- In DNA fingerprinting, DNA is isolated and cut using restriction enzymes, creating fragments of DNA called restriction fragments.
- Each person's length and number of DNA restriction fragments differs.
- Gel electrophoresis has been replaced by the use of STR analysis, which analyzes shorter pieces of DNA.
Gel Electrophoresis

- A technique used to separate DNA fragments
- An electrical current is moved through a gel substance causing molecules to sort by size
- The smaller, lighter molecules will move farthest on the gel
- After developing, the fragments can be visualized for characterization
Short Tandem Repeats (STRs)

- The repeating bases in STRs are shorter than the repeating bases used in DNA fingerprinting.
- STRs can be more easily recovered from the often degraded and limited quantities of DNA typically found in evidence.
- The FBI uses 13 core STRs for identification of Americans.
**Figure 7-5** A law-enforcement officer taking precautions to avoid contaminating the DNA evidence he is collecting.
Inheritance of STRs

- A child inherits one allele from each parent for each STR locus.
- A homozygous genotype occurs when one individual has two alleles that are the same for a specific STR.
- A heterozygous genotype occurs when a person has two different alleles for a particular STR.
Figure 7-6  You can locate each of the 13 core STRs used by the FBI in this image. AMEL on the X and Y chromosomes is not an STR but an indicator of sex.
DNA STR Profiles

- An individual's DNA STR profile is unique.
- DNA STR analysis is performed using automated machines and computers.
- Adding fluorescent dyes to the PCR reaction makes it possible to identify different STR markers.
**Figure 7-7** Inheritance of STRs through independent assortment.

- **Mother** with two different STR alleles per locus: (9, 12)
- **Father** with two different STR alleles per locus: (14, 15)

**Gametes**
- 9
- 12
- 14
- 15

**Offspring**
- 9,14
- 9,15
- 12,14
- 12,15

Possible genotypes in offspring.
Figure 7-8 Steps in STR typing.

STR Allele Frequencies

- An allele frequency is a calculation of how often a particular allele appears within a given population.
- Allele frequency =
  
  \[
  \frac{\text{the number of times an allele is observed in a given population}}{\text{the total alleles observed in the population}}
  \]
Figure 7.9 Crime-scene STR profile comparing evidence DNA to the DNA of two suspects. Three different STR markers are analyzed: D3S1358, VWA, and FGA. Note that Suspect 1 was excluded, while Suspect 2’s STR profile was consistent with the STR profile of the evidence DNA.

Y STR and mtDNA Analyses

- Y STRs
  - Trace ancestry through the male line
- mtDNA
  - Only mothers pass on mitochondrial DNA
- mtDNA and Y STRs
  - Can only yield class evidence
- Fourth amendment rights may become a concern as DNA technology advances.
Uses of DNA Testing

- To identify potential suspects
- To exonerate individuals
- To identify crime and casualty victims
- To establish paternity
- To match organs
Possible Outcomes

- **Match** – The DNA profile appears the same. Lab will determine the frequency.

- **Exclusion** – the genotype comparison shows profile differences that can only be explained by the two samples originating from different sources.

- **Inconclusive** – the data do not support a conclusion as to whether the profiles match.
DNA is a nucleic acid that contains the genetic information necessary for a cell to replicate and make proteins. The code of DNA is found within the sequence of nitrogenous bases.

DNA sequences are unique to each individual (except an identical twin). The variations within noncoding parts of the DNA molecule are the basis for forensic identification.

DNA analysis can help solve crimes and exonerate the falsely accused.
Using PCR amplification, minute amounts of DNA evidence can be used to solve crimes.

DNA contains within its noncoding regions many repeated sequences, including STRs, which vary in number among individuals; these differences are used to produce a DNA profile of a person.

DNA profiling has dramatically improved over the past 25 years due to improvements in biotechnology, computers, and automated processing of DNA. STR analysis has replaced gel electrophoresis in forensics work.
Summary (continued)

- DNA profiling enables us to determine whether DNA samples came from the same person or different persons, or to establish kinship.
- Analyses of hypervariable base sequences of mtDNA in noncoding regions can help identify people through their maternal relatives.
- CODIS and the NDIS have helped to prevent and solve crimes by improving communication among law enforcement agencies at the local, state, and national levels.