

Lecture for Wednesday


Dr. Prince

DNA PROFILING

Like your SS Number, your DNA is your identity

- **DNA profiling** uses DNA to identify a particular individual
 - Compares genetic markers from noncoding regions that show variation between individuals

1 DNA isolated


Crime scene


Suspect 1


Suspect 2

2 DNA of selected markers amplified



3 Amplified DNA compared

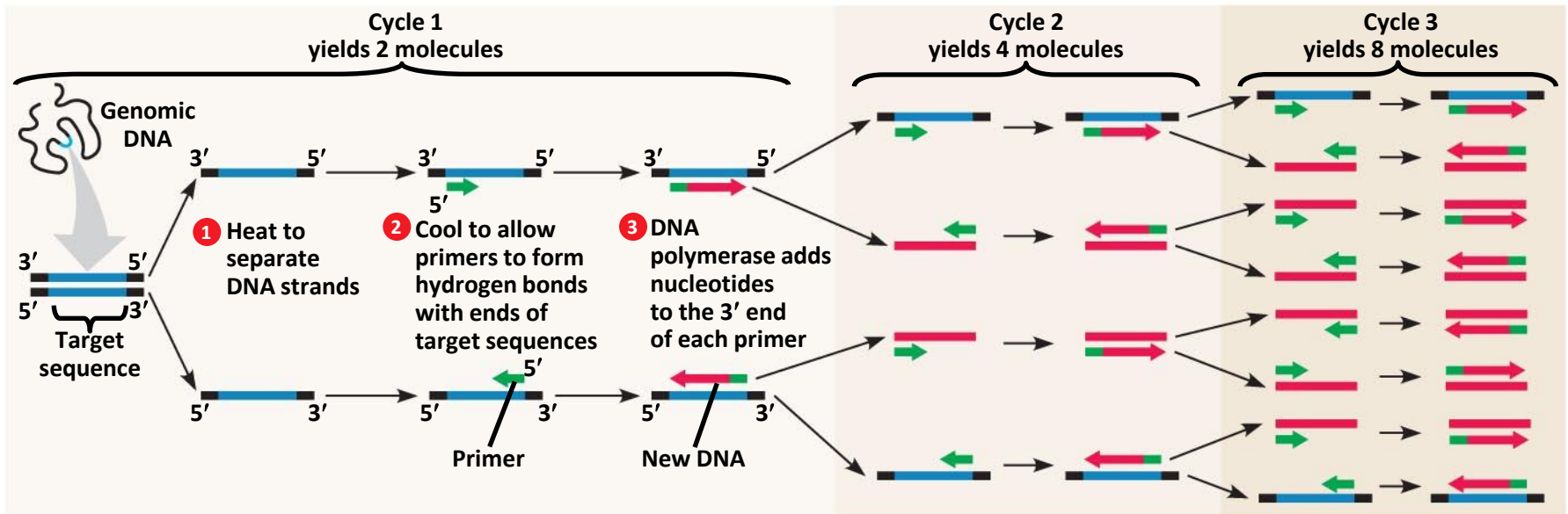


PCR

- **Polymerase chain reaction (PCR)** is a method of amplifying a segment of DNA
- Relies upon a pair of short DNA molecules that bind to sequences at each end (**primers**) and serve as a starting point for DNA replication

Advantages of PCR

- Can amplify DNA from a small sample
- Results are obtained rapidly
- Reaction is highly sensitive, copying only the target sequence

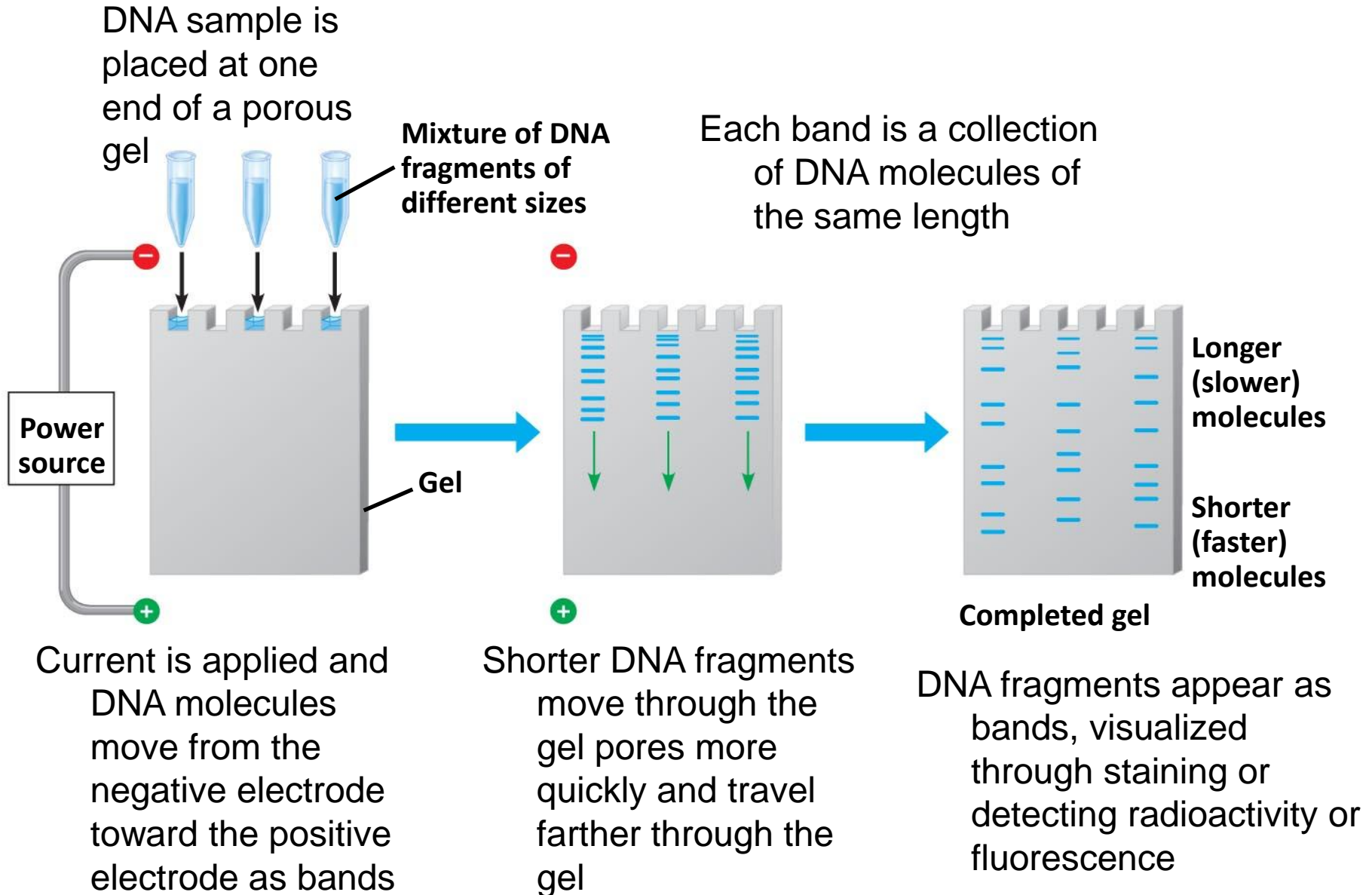


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Repeated cycle of steps for PCR

- Sample is heated to separate DNA strands
- Sample is cooled and primer binds to specific target sequence
- Target sequence is copied with heat-stable DNA polymerase

Gel electrophoresis separates DNA molecules based on size



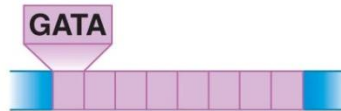
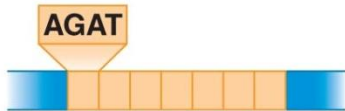
STR

- **Short tandem repeats (STRs)** are commonly used in DNA profiling
 - STRs are short DNA sequences that are repeated many times in a row at the same location
 - **STR analysis** compares the lengths of 13 different STR sequences at specific regions of the genome as they can differ between individuals

STR site 1

STR site 2

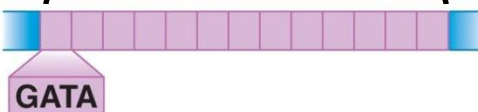
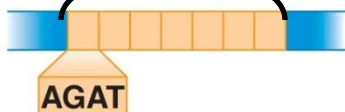
Crime scene DNA



Number of short tandem repeats match

Number of short tandem repeats do not match

Suspect's DNA



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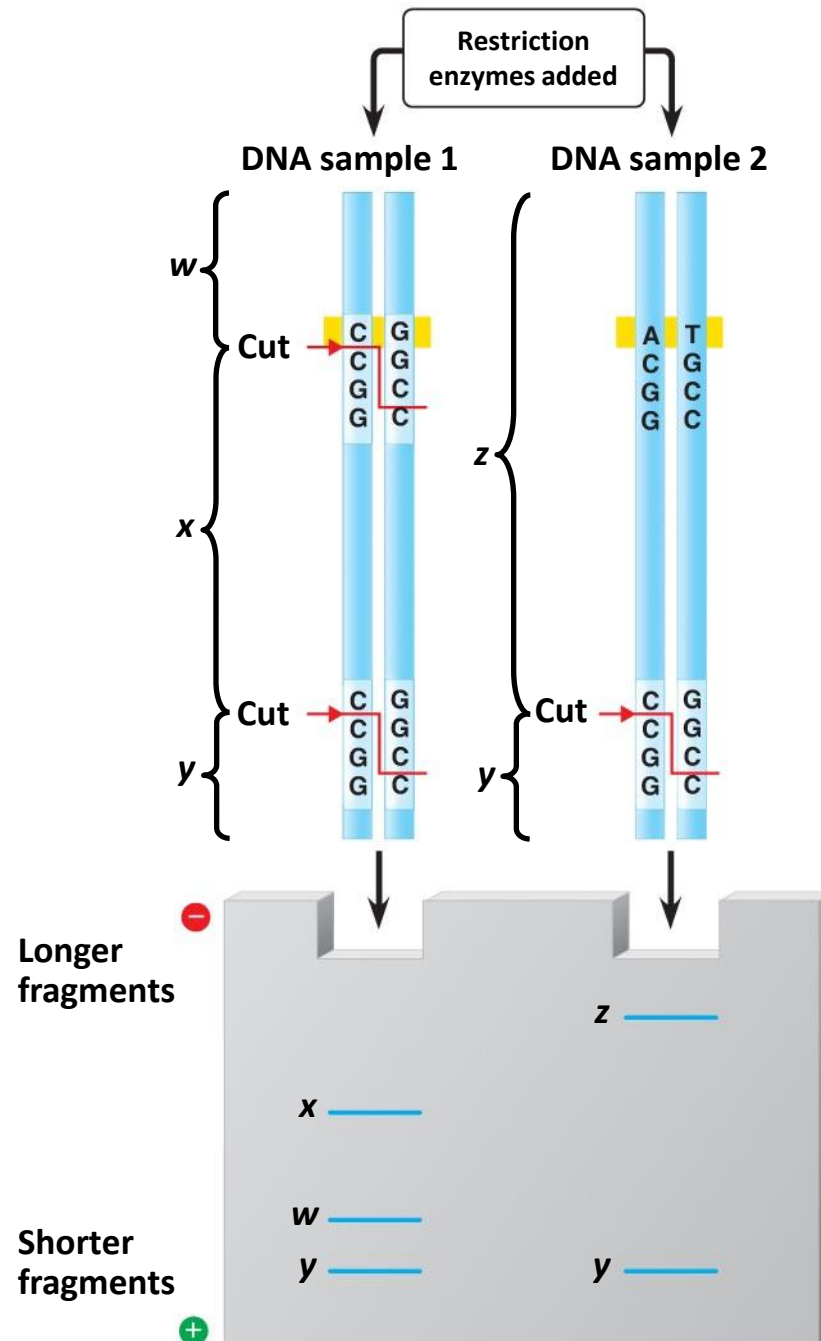
DNA profiling provides evidence

- Forensics
 - Evidence to show guilt or innocence
- Establishing family relationships
 - Paternity analysis
- Identification of human remains
 - September 11, 2001, attack on the World Trade Center
- Species identification
 - Evidence for sale of products from endangered species

RFLP

Restriction fragment length polymorphism (RFLP) is a variation in the size of DNA fragments due to a SNP that alters a restriction site.

It involves a comparison of sizes of fragments by gel electrophoresis



Single nucleotide polymorphism (SNP) is a variation at one base pair



GENOMICS

Genomics the study of whole genomes

“Immortal Thread – It’s *^%# hot here.”

- **Genomics** is the study of an organism’s complete set of genes and their interactions
- The Human Genome Project
- Evolutionary relationships can be elucidated
 - Genomic studies showed a 96% similarity in DNA sequences between chimpanzees and humans
 - Functions of human disease-causing genes have been determined by comparisons to similar genes in yeast

TABLE 12.17

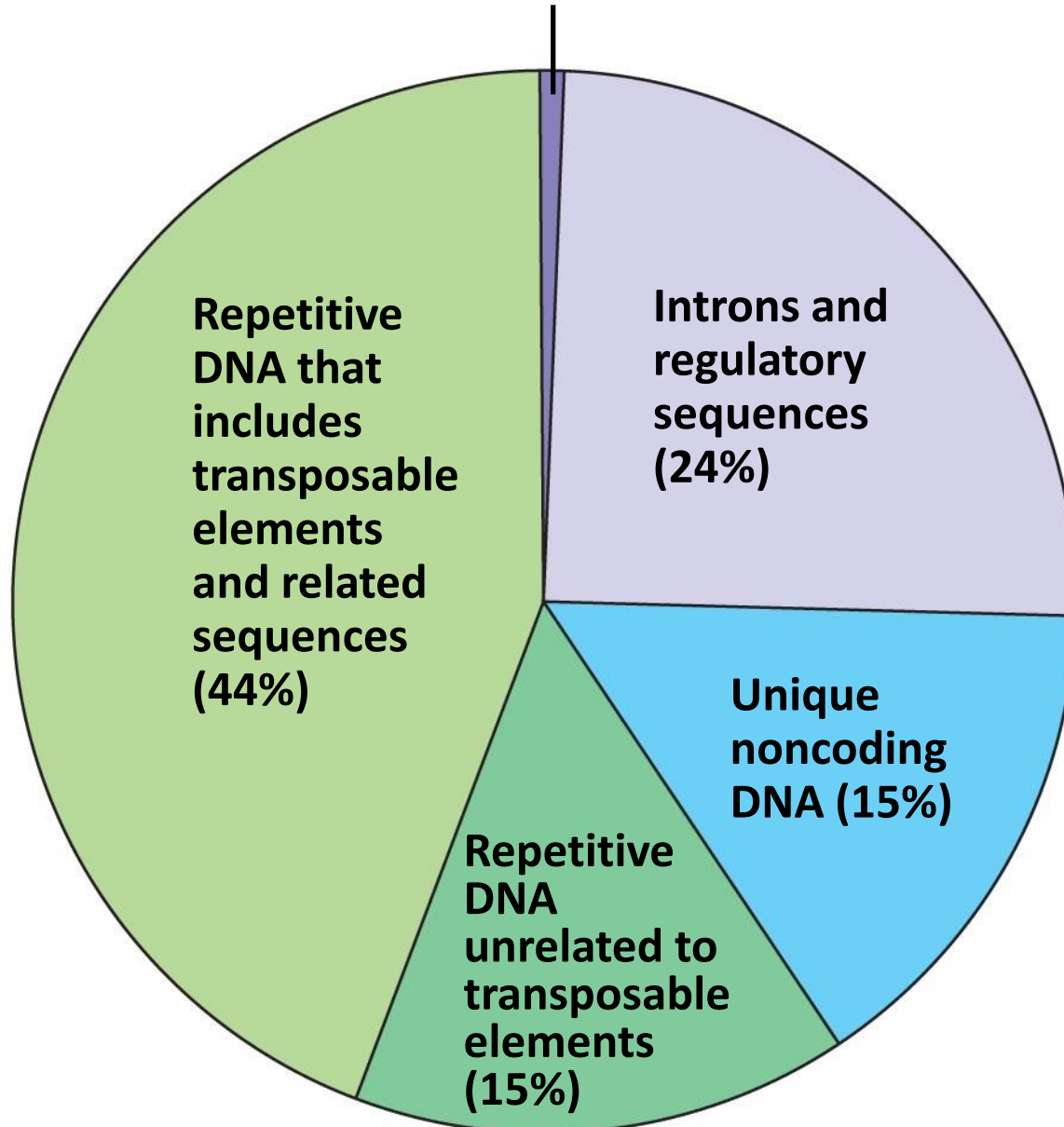
SOME IMPORTANT COMPLETED GENOMES

Organism	Year Completed	Size of Genome (in Base Pairs)	Approximate Number of Genes
<i>Haemophilus influenzae</i> (bacterium)	1995	1.8 million	1,700
<i>Saccharomyces cerevisiae</i> (yeast)	1996	13 million	6,200
<i>Escherichia coli</i> (bacterium)	1997	4.6 million	4,400
<i>Caenorhabditis elegans</i> (nematode)	1998	97 million	19,000
<i>Drosophila melanogaster</i> (fruit fly)	2000	180 million	13,700
<i>Arabidopsis thaliana</i> (mustard plant)	2000	118 million	25,500
<i>Mus musculus</i> (mouse)	2001	2.6 billion	22,000
<i>Oryza sativa</i> (rice)	2002	430 million	60,000
<i>Homo sapiens</i> (humans)	2003	3.2 billion	21,000
<i>Rattus norvegicus</i> (lab rat)	2004	2.8 billion	25,000
<i>Pan troglodytes</i> (chimpanzee)	2005	3.1 billion	22,000
<i>Macaca mulatta</i> (macaque)	2007	2.9 billion	22,000

The Human Genome Project

- Goals of the **Human Genome Project (HGP)**
 - To determine the nucleotide sequence in the human genome
 - To identify the location and sequence of every human gene
- Revealed that most of the human genome does not consist of genes
- Humans have 21,000 genes in 3.2 billion nucleotide pairs
- Only 1.5% of the DNA codes for proteins, tRNAs, or rRNAs
- The remaining 88.5% of the DNA contains
 - Control regions (promoters and enhancers)
 - Unique noncoding DNA
 - Repetitive DNA in centromeres, telomeres, and dispersed throughout the genome

**Exons (regions of genes coding for protein
or giving rise to rRNA or tRNA) (1.5%)**



The whole-genome shotgun method of sequencing a genome can provide a wealth of data quickly

- Three stages of the Human Genome Project
 - A low-resolution linkage map was developed using RFLP analysis of 5,000 genetic markers
 - A physical map was constructed from nucleotide distances between the linkage-map markers
 - DNA sequences for the mapped fragments were determined
 - Restriction enzymes were used to produce fragments that were cloned and sequenced
 - Computer analysis assembled the sequence by aligning overlapping regions



Chromosome

**Chop up with
restriction enzyme**



DNA fragments

**Sequence
fragments**



**Align
fragments**



**Reassemble
full sequence**

AATCGGACGATTAATGTA

Proteomics is the scientific study of the product of gene expression

– **Proteomics**

- Studies the proteome, the complete set of proteins specified by a genome
- Investigates protein functions and interactions
- The human proteome may contain 100,000 proteins

Genomes hold clues to the evolution of humans

- Comparisons of human and chimp genomes
 - Differ by 1.2% in single-base substitutions
 - Differ by 2.7% in insertions and deletions of larger DNA sequences
 - Human genome shows greater incidence of duplications
 - Genes showing rapid evolution in humans
 - Genes for defense against malaria and tuberculosis
 - Gene regulating brain size
 - *FOXP2* gene involved with speech and vocalization

