Dr. Bertolotti

Chapter 14: Human Hereditary

Essential Question

How can we use genetics to study human inheritance?

How can karyotypes be used to help geneticists?

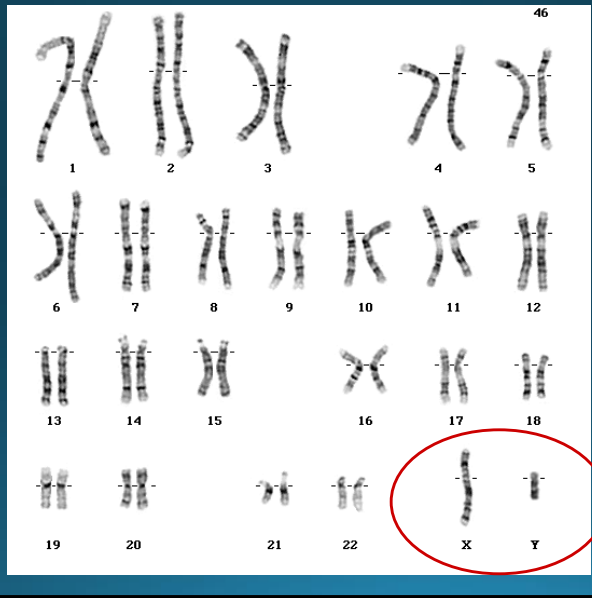
Human Hereditary

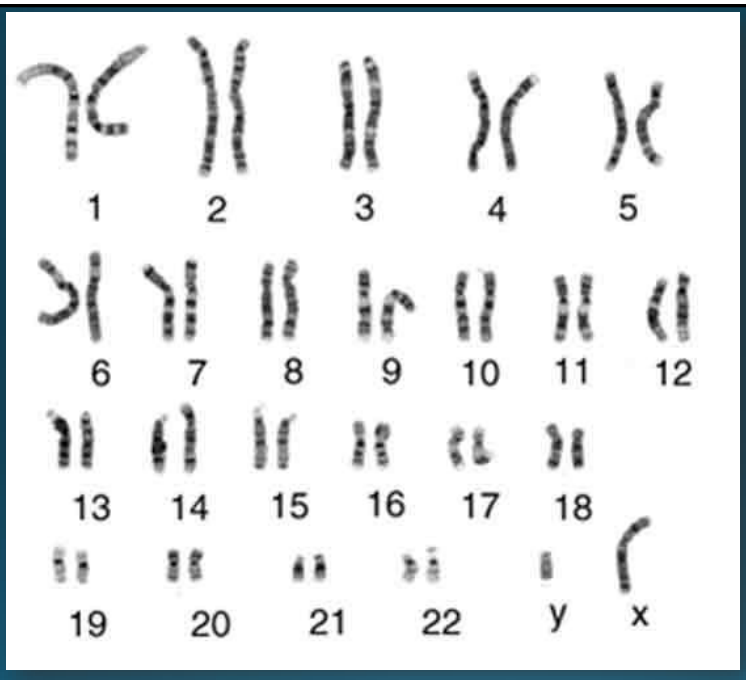
- A **Karyotype** is a photograph that shows the complete diploid set of chromosomes grouped together in pairs, arranged in order of decreasing size.
  - A **genome** is the full set of genetic information that an organism carries in its DNA.
- Humans contain 46 chromosomes
  - 2 of the 46 chromosomes are **sex chromosomes** because it determines the sex of an individual
    - Females have 2 copies of the X chromosome (XX)
    - Males are XY
  - The remaining 44 chromosomes are **autosomes** as it does not determine the sex of an individual

Genetics of sex

- Women & men are very different, but just a few genes create that difference
- In mammals = 2 sex chromosomes
  - X & Y
  - 2 X chromosomes = female: XX
  - X & Y chromosome = male: XY

Sex chromosomes



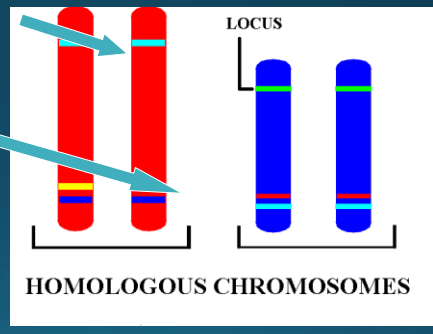


A KARYOTYPE

- Males and females are born in a roughly 50:50 ratio because of the segregation during meiosis
  - All human egg cells carry a single X chromosome (23, X)
  - Half of all sperm cells carry an X chromosome (23,X) and half carry a Y chromosome (23,Y)
- 23 pairs of **homologous chromosomes**— one from Mom and one from Dad that are very similar in structure and function.

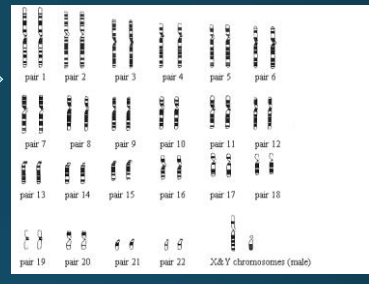
Homologous Chromosomes

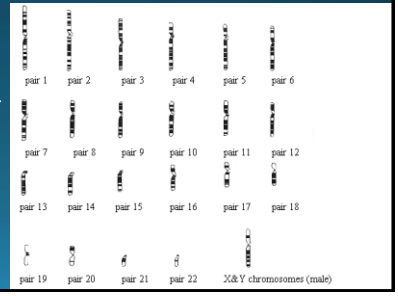
- Mom contributes one and Dad contributes other of each pair of homologous chromosomes.
- This means each somatic cell has two copies of each chromosome.



2 Types of Cells

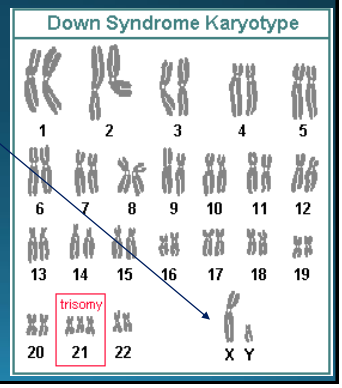
- Body cells
  - called **somatic cells**
  - diploid→ chromosomes are in pairs (46=23 pairs for humans)
  - 1 set from mom, 1 set from dad
- Reproductive cells
  - egg & sperm
  - called **gametes**
  - haploid →no pairs (only 23 single chromosomes)



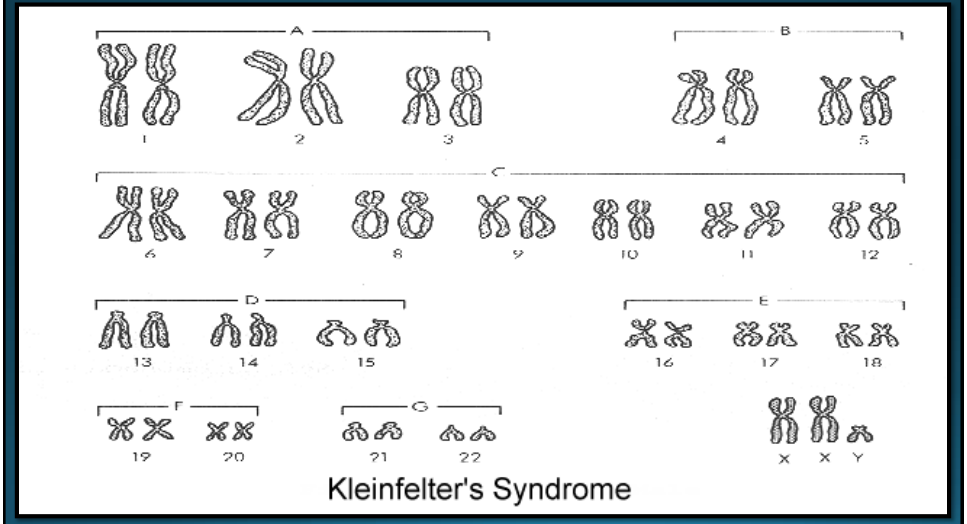


What can be determined from looking at a karyotype?

- **1. Sex of the individual.**
  - 2 types of chromosomes
    - Autosomes (1-22)
    - Sex chromosomes (X or Y).
  - XX→female XY→male
  - What sex?
- **2. Chromosomal mutations**
  - Monosomy-one less
  - Trisomy--extra
  - Deletions-piece missing



What's Wrong with This Karyotype?



QUESTION AND ANSWER

How can karyotypes be used to help geneticists?

What patterns of inheritance do human traits follow?

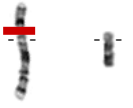
Sex linked genes

- **Sex linked genes** are genes located on the X or Y chromosome
  - Many are found on the X chromosome
  - Because males have just 1 X chromosome, all X linked alleles are expressed in males even if they are recessive
  - A defective version of an X chromosome produces color blindness in males
  - Hemophilia is also a sex linked disorder caused by 2 recessive alleles
    - It is common in males
    - The protein necessary for blood clotting is absent
  - Duchenne Muscular Dystrophy is a sex linked disorder that results in progressive weakening and loss of skeletal muscle

Sex-linked traits

- Sex chromosomes have other genes on them, too
  - especially the X chromosome
  - hemophilia in humans
    - blood doesn't clot
  - Duchenne muscular dystrophy in humans
    - loss of muscle control
  - red-green color blindness
    - see green & red as shades of grey

X X



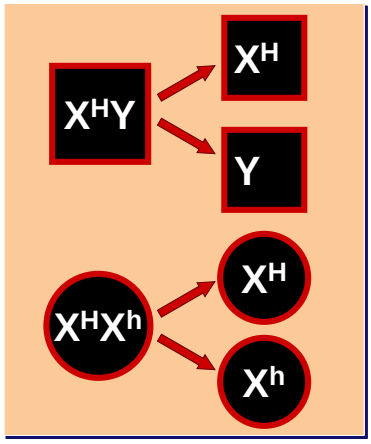
X Y

Sex-linked traits

sex-linked recessive

X^HY x X^HX^h

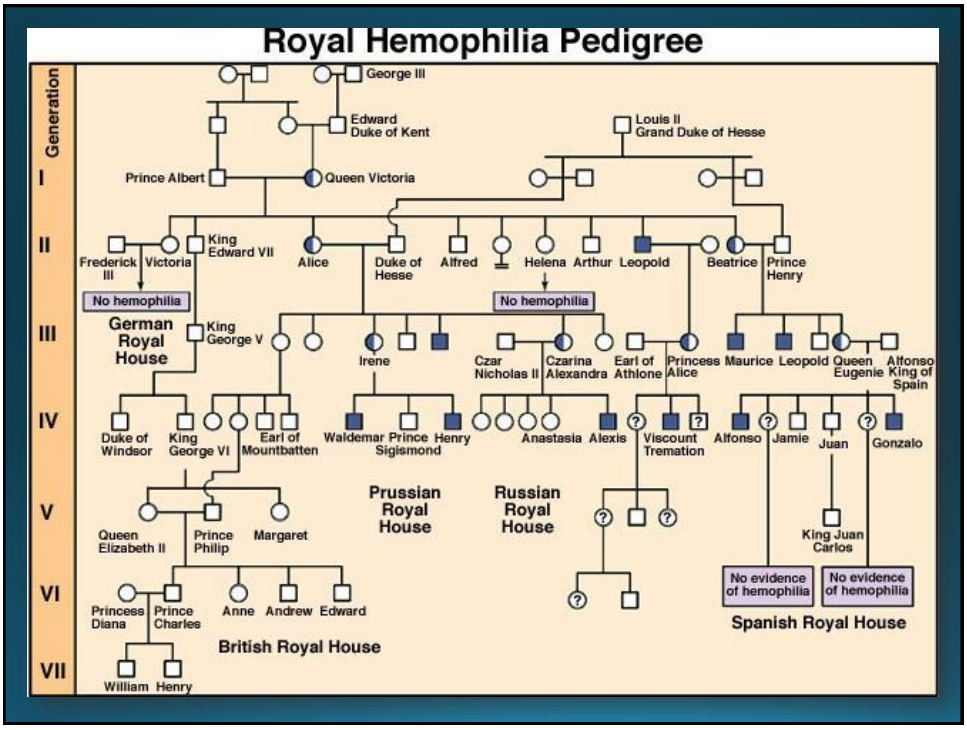
2 normal parents, but mother is carrier



male / sperm

female / eggs	X^H	Y
X^H	X^HX^H	X^HY
X^h	X^HX^h	X^hY

Queen Victoria and Descendants



QUESTION AND ANSWER

What patterns of inheritance do human traits follow?

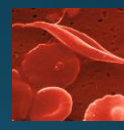
How do small changes in DNA molecules affect human traits?

From molecule to phenotype

- Changes in a gene's DNA sequence can change proteins by altering their amino acid sequences, which may directly affect one's phenotype.

Disorders caused by individual genes

- **1. Sickle cell Disease**
  - Sickle cell disease is caused by a codominant allele in which a small change in the DNA of a single gene affects the structure of a protein
  - Is characterized by the bent and twisted shape of red blood cells

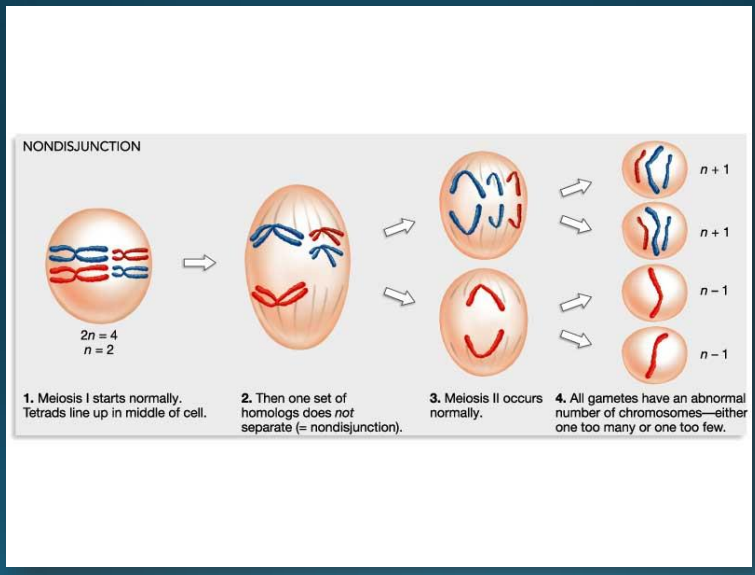


- **2. Cystic Fibrosis**
  - Cystic fibrosis or CF is a common genetic disease caused by a recessive allele on chromosome 7
  - Individuals affected produce a heavy mucus that makes it hard for them to breathe

Chromosomal disorders

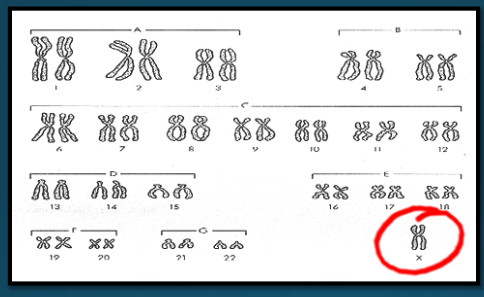
- If nondisjunction occurs during meiosis, gametes with an abnormal number of chromosomes may result, leading to a disorder of chromosome numbers.
- The most common error in meiosis is **nondisjunction** which is an error in meiosis in which homologous chromosomes fail to separate
  - ***If nondisjunction occurs, abnormal numbers of chromosomes may find their way into gametes, and a disorder of chromosome numbers may result***

Nondisjunction



- **Extra copies**
  - Example: Down Syndrome
  - An individual with Down Syndrome has 3 copies of chromosome 21
  - Produces mild to severe retardation and increased susceptibility to many diseases
- **Sex chromosome disorders**
  - Are disorders that occur among sex chromosomes,
    - Example: Turners syndrome and Klinefelter's syndrome

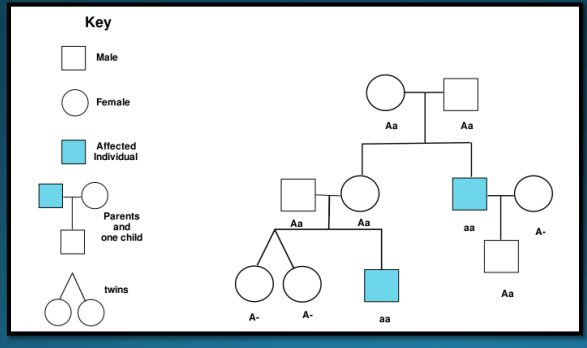
- In Turner's syndrome, a female inherits only 1 X chromosome and therefore are sterile. Nondisjunction is the source of error.



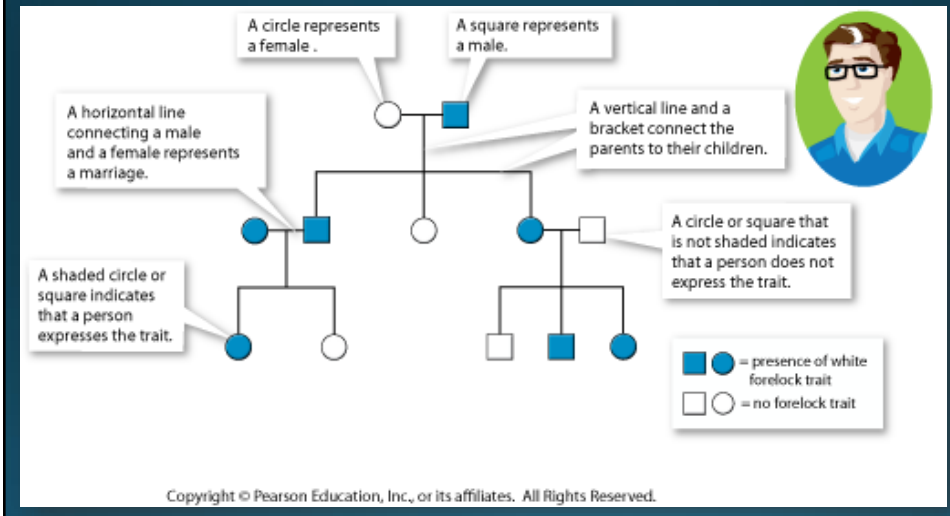
- In males, Klinefelter's syndrome is marked by possessing an extra X chromosome which prevents the individual from reproducing. Nondisjunction is the source of error.

Human Pedigrees

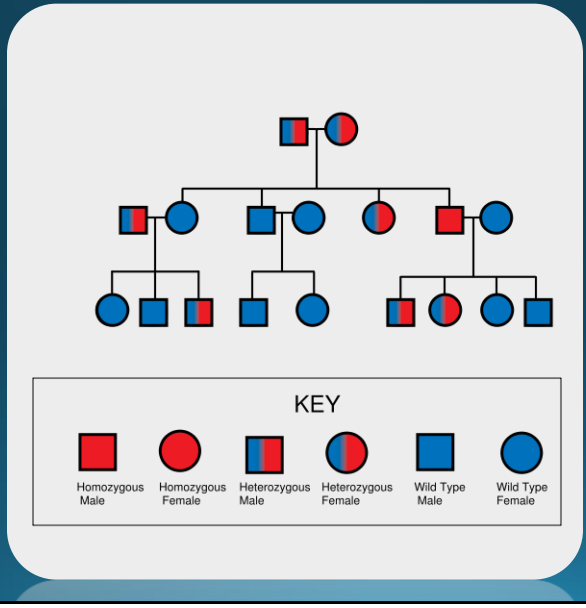
- A **pedigree chart** shows the relationships within a family
  - The information gained from pedigree analysis makes it possible to determine the nature of genes and alleles associated with inherited human traits



Pedigree chart



Autosomal recessive



QUESTION AND ANSWER

How do small changes in DNA molecules affect human traits?

What techniques are used to study human DNA?

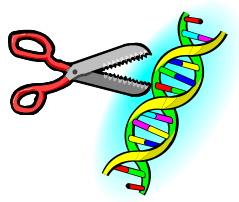
Manipulating DNA

- Scientists use their knowledge of the structure of DNA and its chemical properties to study and change DNA molecules. Different techniques are use to extract DNA from cells, to cut DNA into smaller pieces, to identify the sequence of bases in a DNA molecule, and to make unlimited copies of DNA

Tools of molecular biology

- **Genetic engineering** is the process of making changes in the DNA code of living organisms
- *DNA can be manipulated by*:
  - A) DNA extraction
  - B) Cutting DNA using **restriction enzymes** which are enzymes that cut DNA at a specific sequence of nucleotides
  - C) Making copies of genes/DNA using polymerase chain reaction (PCR)

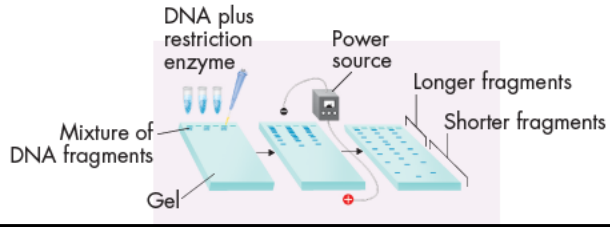
Cutting DNA

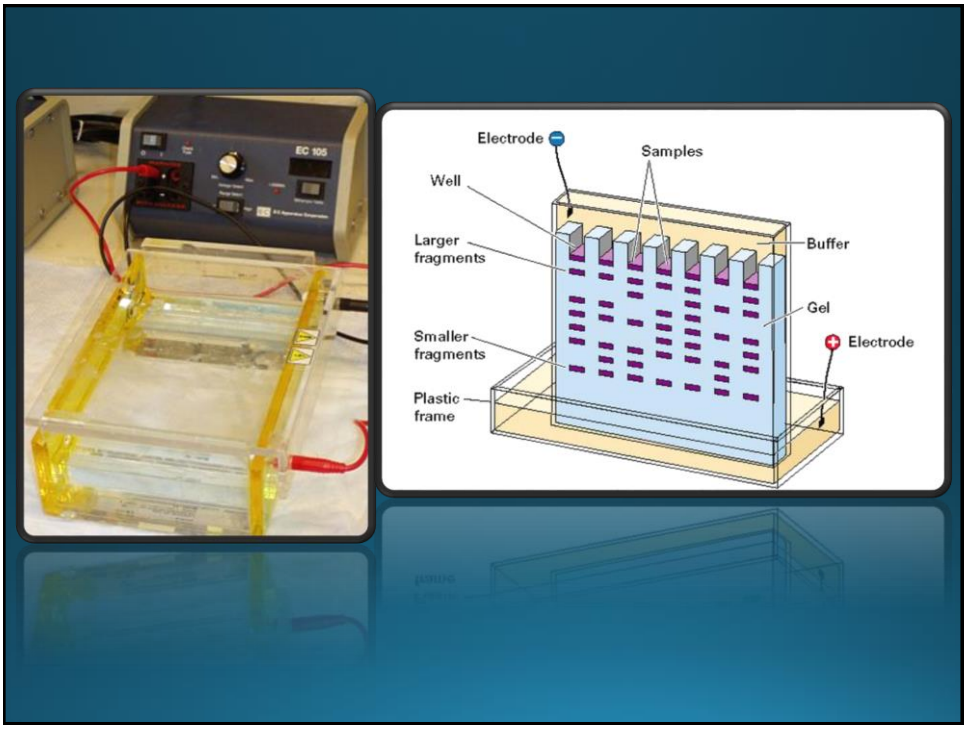


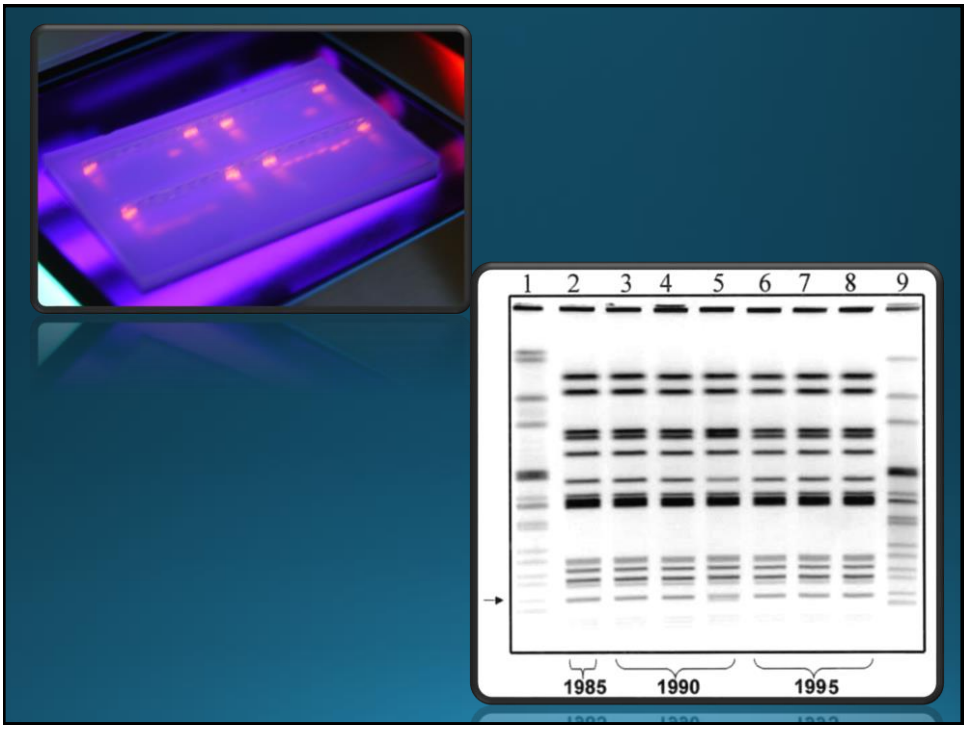
- DNA "scissors"
  - enzymes that cut DNA
  - **restriction enzymes**
    - used by bacteria to cut up DNA of attacking viruses
    - EcoRI, HindIII, BamHI
  - cut DNA at specific sites
    - enzymes look for specific base sequences

GTAACGAATTCACGCTT
CATTGCTTAAGTGCGAA

- C) Separating DNA by **gel electrophoresis** which is a procedure used to separate and analyze DNA fragments at one end of a porous gel and applying an electric voltage to the gel
  - The smaller the DNA fragment, the faster and further it moves
  - Can be used to locate and identify 1 particular gene and compare genomes

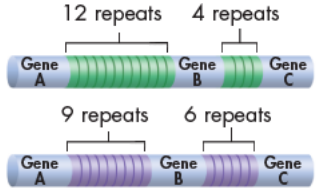



Electrode
Samples
Well
Larger fragments
Buffer
Gel
Smaller fragments
Electrode
Plastic frame


1 2 3 4 5 6 7 8 9
1985
1990
1995

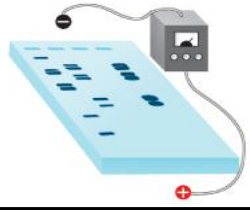
Personal Identification

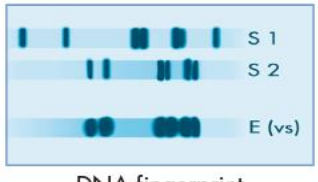
- No individual is exactly like any other genetically—except for identical twins, who share the same genome.
- Chromosomes contain many regions with repeated DNA sequences that do not code for proteins. These vary from person to person. Here, one sample has 12 repeats between genes A and B, while the second has 9 repeats between the same genes.
- **DNA fingerprinting** can be used to identify individuals by analyzing these sections of DNA that may have little or no function but that vary widely from one individual to another.



Personal Identification

- In DNA fingerprinting, restriction enzymes first cut a small sample of human DNA into fragments containing genes and repeats. Note that the repeat fragments from these two samples are of different lengths.
- Next, gel electrophoresis separates the restriction fragments by size.
- A DNA probe then detects the fragments that have highly variable regions, revealing a series of variously sized DNA bands.

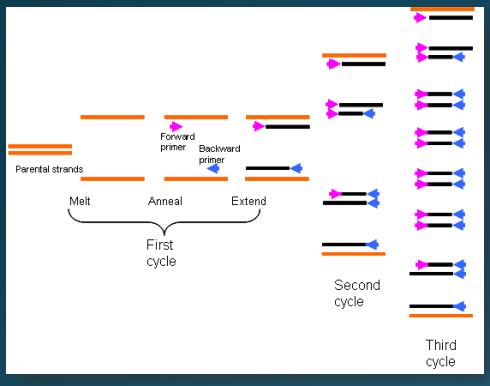


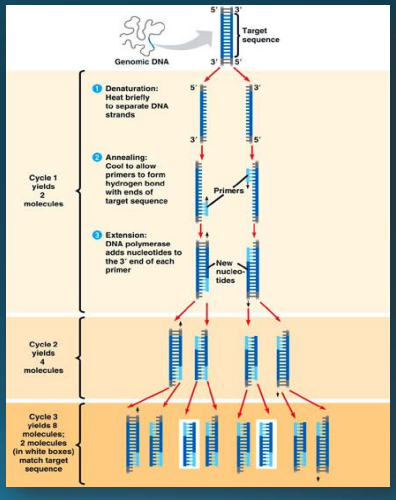


DNA fingerprint

- **Polymerase chain reaction (PCR)** is a technique that allows molecular biologists to make many copies of a particular gene
  - The first step in using the polymerase chain reaction method to copy a gene is to heat a piece of DNA, which separates its two strands. Then, as the DNA cools, primers bind to the single strands. Next, DNA polymerase starts copying the region between the primers. These copies can serve as templates to make still more copies.

Polymerase Chain Reaction (PCR)





- **Polymerase chain reaction (PCR)** is a technique that allows molecular biologists to make many copies of a particular gene
  - The first step in using the polymerase chain reaction method to copy a gene is to heat a piece of DNA, which separates its two strands. Then, as the DNA cools, primers bind to the single strands. Next, DNA polymerase starts copying the region between the primers. These copies can serve as templates to make still more copies.

QUESTION AND ANSWER

What techniques are used to study human DNA?

- **The Human Genome project** is an ongoing 13-year international effort to analyze all 3 billion base pairs of human DNA and identifying all human genes.
  - The Human Genome Project pinpointed genes and associated particular sequences in those genes with numerous diseases and disorders. It is also identified about three million locations where single-base DNA differences occur in humans.
- In gene therapy, an absent or faulty gene is replaced by a normal, working gene
- **Epigenetics**:
  - As an organism grows and develops, carefully orchestrated chemical reactions activate and deactivate parts of the genome at strategic times and in specific locations. Epigenetics is the study of these chemical reactions and the factors that influence them.
  - Research into epigenetics has shown that environmental factors affect characteristics of organisms. These changes are sometimes passed on to the offspring.

Essential Question

How can we use genetics to study human inheritance?