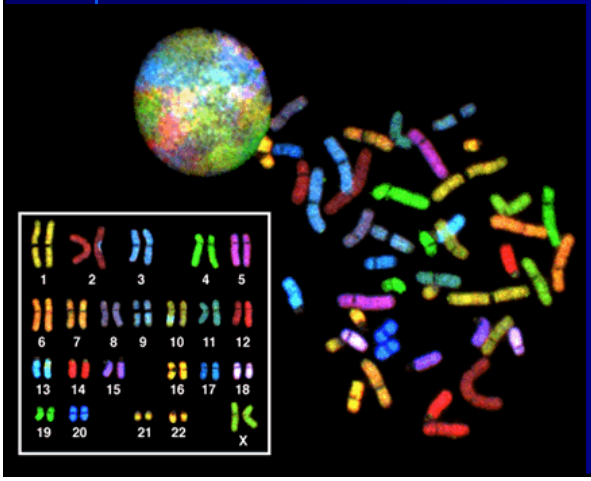
Heredity & Genetic Engineering

Human Chromosomes Review

- Human body cells, called somatic cells, have 46 chromosomes (diploid number)
- Gametes have 23 chromosomes (haploid number)
  - Zygote = fertilized egg is diploid.
- **Autosomes** – chromosome pairs 1-22 (44 total)
- **Sex chromosomes** – 23rd pair of unmatched chromosomes, determine sex
  - XX – female, XY – male

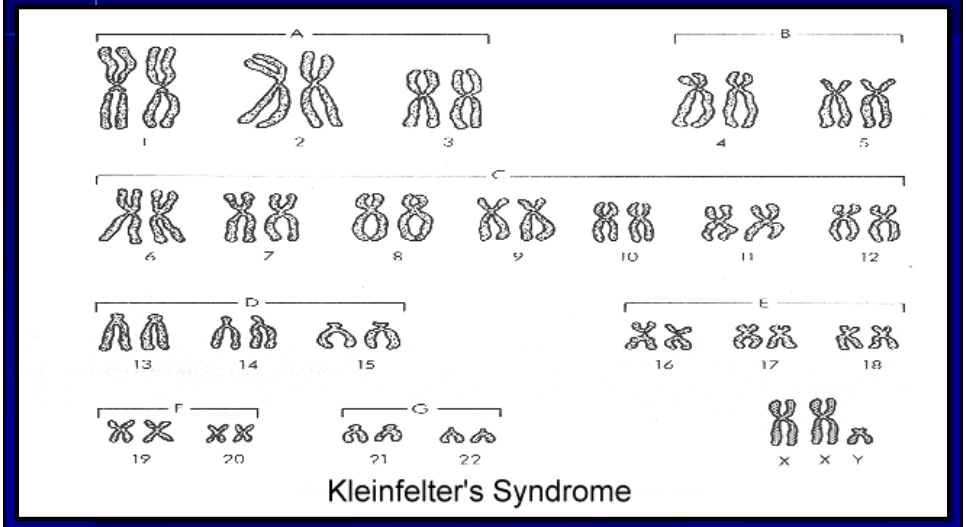
Karyotype

- photograph that shows the complete diploid set of chromosomes grouped together in pairs, arranged in order of decreasing size.



- **Monosomy-**missing a chromosome
- **Trisomy-** having an extra chromosome
- **genetic abnormalities** can be detected by looking at a karyotype of a person's chromosomes

What's Wrong with This Karyotype?



Kleinfelter's Syndrome

Tracking & Predicting Genetic Disorders

- **Human Genome (Karyotypes)**– complete set of genetic information; can be used to identify disorders.
- **Pedigree Charts** – shows relationships of traits within a family; how disorders could be inherited

Autosomal Disorders

- **Autosomal Recessive Gene Disorders**
  - Most numerous
  - Need 2 alleles for expression (aa)
  - **Carriers** – heterozygous (Aa)-normal
    - Have no sign of disorder
    - Offspring can inherit disorder
- **Autosomal Dominant Gene Disorders**
  - Only **1** allele for expression (AA or Aa)
  - 2 dominant alleles (AA) = usually **fatal**

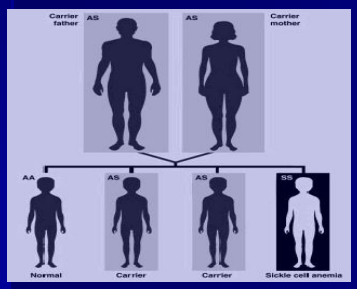
Autosomal Recessive Gene Disorders

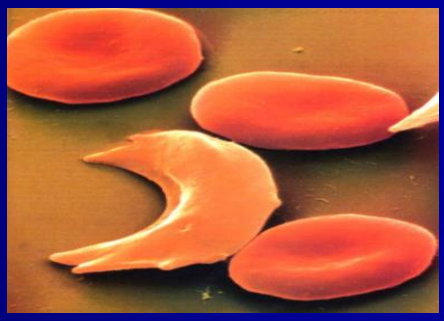
- **Phenylketonuria (PKU)** – lack enzyme to break down phenylalanine (amino acid that forms proteins) in milk & foods → can lead to mental retardation, seizures, and other serious medical problems
  - Diagnosed early- on a strict diet can lead normal life
- **Tay-Sachs** – lack enzyme to break down lipids- deterioration of mental and physical abilities
  - Commences around 6 months old and causes death by age 4; No cure or treatment
- **Cystic Fibrosis** – produce too much mucus in lungs, pancreas, liver & intestines (digestive tract)
  - Mutation in gene for a certain protein
  - Lung infections, sinus infections, poor growth and infertility

Codominant Recessive **Gene** Disorders

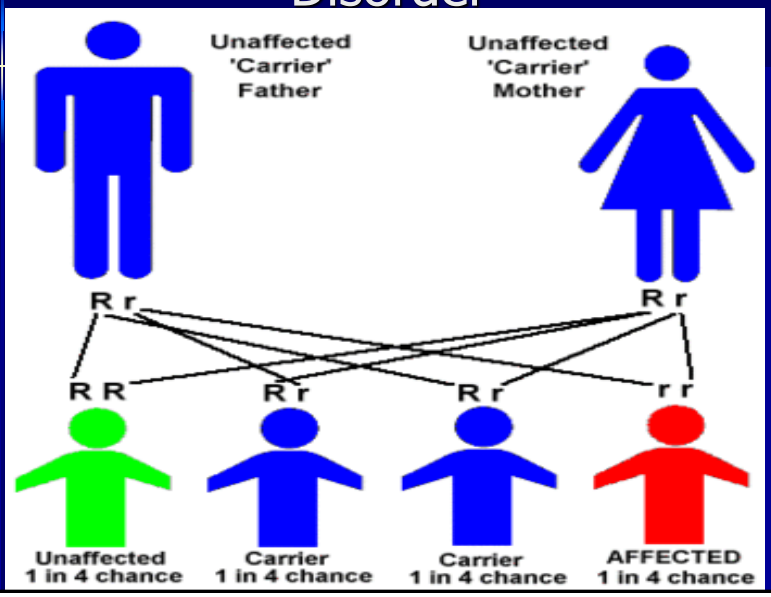
Sickle –cell anemia

- **Carriers** (both normal hemoglobin & sickle-celled hemoglobin is expressed –Ss)
  - show some symptoms – trouble w/ exercising or heavy activity because they aren't getting enough oxygen throughout their body
  - Carriers are immune to **malaria**
- Sufferers – ss – usually fatal





Autosomal Recessive Gene Disorder



Autosomal Dominant Gene Disorders

- **Achondroplasia** – form of **dwarfism**
  - AA = **dead**; Aa = **dwarf**, aa = **normal**
  - If both parents of a child have achondroplasia, and both parents pass on the mutant gene, then it is very unlikely that the homozygous child will live past a few months of its life.
- **Huntington's Disease** – neurodegenerative- affects **nervous** system and muscle coordination
  - Mutation in "Huntingtin" gene
  - Any child of an affected person typically has a 50% chance of inheriting the disease

Sex-linked Genes & Disorders

- Other traits on the **X or Y chromosome**
  - **Most Sex-linked disorders are on the X chromosome**

- **Recessive** traits on the **X chromosome**:
  - **Color-blindness, hemophilia** (blood doesn't clot correctly)**, muscular dystrophy** (**MD-** skeletal muscle weakness, defects in muscle proteins & death of muscle cells & tissue)
  - Tends to be passed from **mother to son-** more likely to occur in males than females
    - Because males only have one X- if they get "infected X" from mom then they have disorder
    - Daughters have to get 2 "infected x's" to express disorder

- Could a daughter be color-blind? If so, how?
- *Father has to be color-blind, mother is a carrier, and daughter receives both infected X chromosomes*

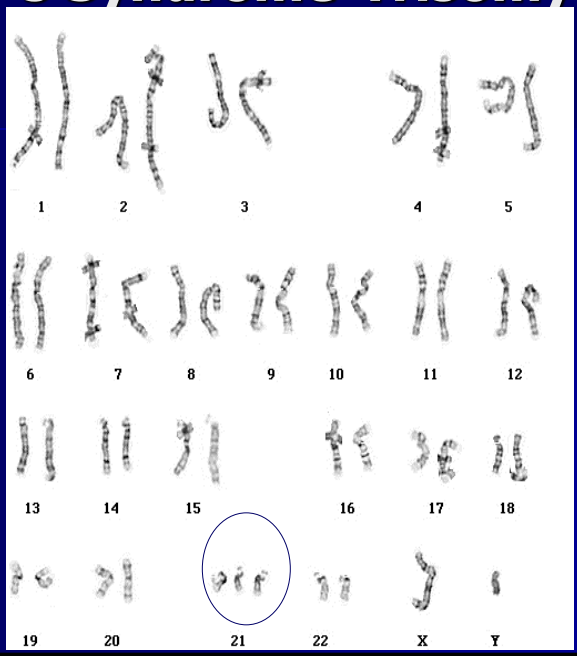
Chromsomal **Mutation** Disorders

- **Nondisjunction** is the failure of chromosomes to divide properly during **meiosis** -error in meiotic cell division
  - It results in **extra chromosomes** or loss of **chromosomes**.

Nondisjunction of Autosomes (chromosomes 1-22)

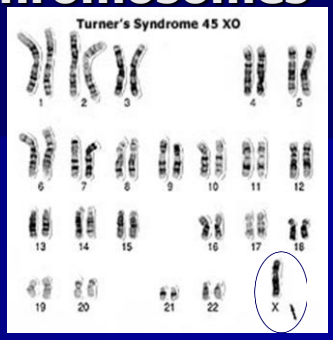
- Down's Syndrome – **extra** 21st chromosome, called trisomy 21
  - Most common chromosome abnormality in humans
  - Delay in cognitive ability and physical growth & a particular set of facial characteristics
- Cat Cry Syndrome – **deletion** of the 5th (or part of the **5th chromosome**)
  - Affected children have "cat-like" cry- about 1/3 of children lose the cry by age 2
  - Problems with larynx and nervous system → can lead to intellectual disability

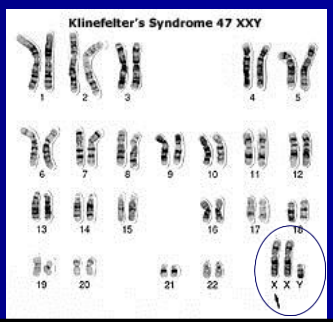
Down's Syndrome-Trisomy 21



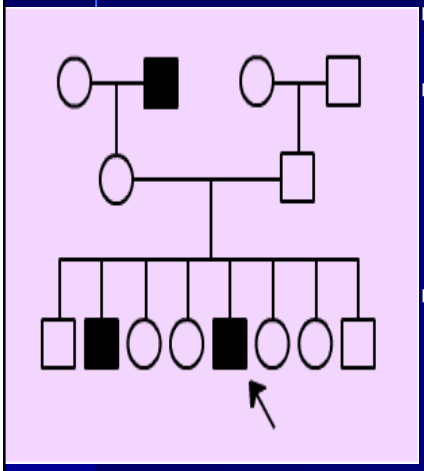
Nondisjuntion of Sex Chromosomes

- **Turner's Syndrome** – **45X** female with 1 X chromosome, female is sterile
- **Klinefelter's Syndrome** – **47XXY**, male with an extra X chromosome, cannot reproduce
- **Supermale** – **47XYY**, male with extra Y chromosome, usually very tall

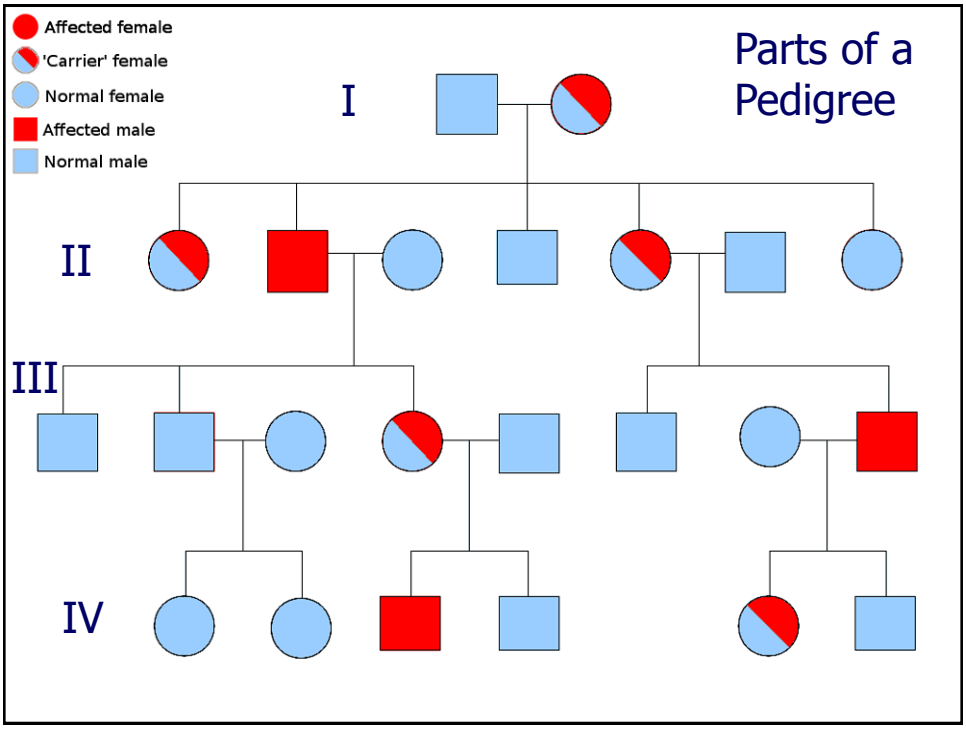




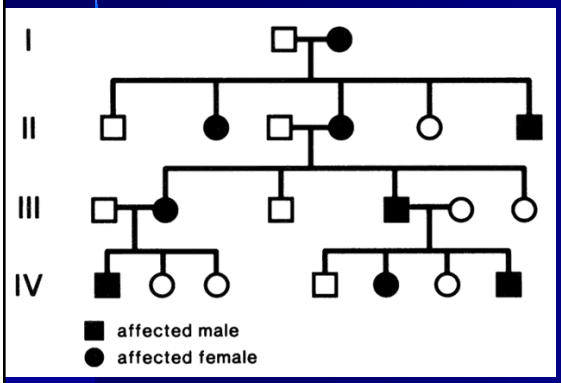
Pedigrees



- Pedigrees study how a **trait** is passed from one generation to the next.
- Infers **genotypes** of family members
- Disorders can be carried on…
  - **Autosomes** (1-22 pairs of chromosomes)
  - **Sex Chromosomes** (X or Y)
  - **Number** of **Chromosomes** (either X>46>X)
- Keep in mind: traits are influenced heavily by non-**genetic** factors or **environmental** factors
  - Nutrition
  - Exercise
  - Toxins (mutagens)
  - Disease



1. Determine if the trait is dominant or recessive.



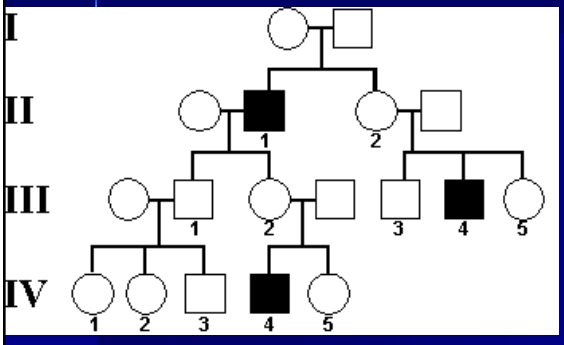
Recessive:

- If the trait skips a generation
- If affected individual has normal parents or vice versa

Dominant:

- If the trait appears in every generation

1. Determine if the trait is autosomal or sex-linked.



Females tend to "carry" a trait and affect their sons.
Females get the trait from an affected father or carrier/affected mother
Affected males got it from their mother and give it to their daughters to "carry."

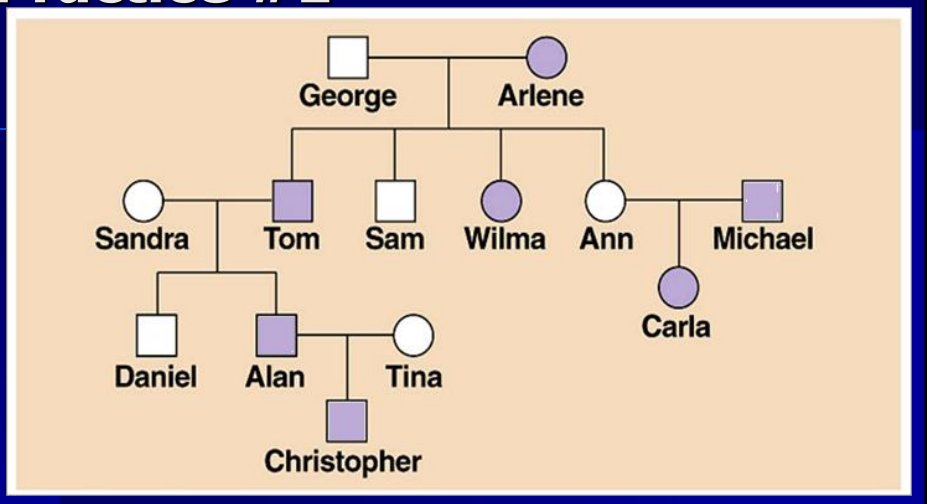
Autosomal

- If the trait affects males and females equally

Sex-Linked:

- If the trait affects one sex more than the other *(especially males)*

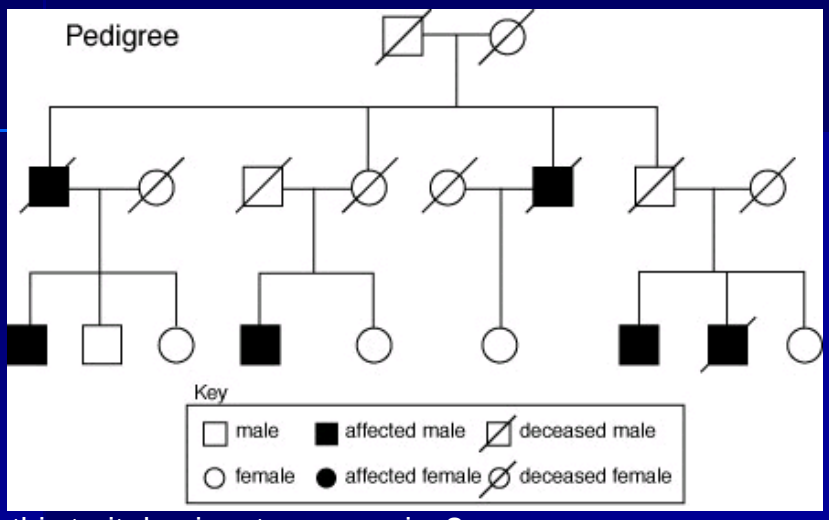
Practice #1



Is this trait dominant or recessive?
Is this trait Autosomal or Sex-linked?

- Assign genotypes to the pedigree to show the inheritance pattern.

Practice #2



Is this trait dominant or recessive?
Is this trait Autosomal or Sex-linked?

- Assign genotypes to the pedigree to show the inheritance pattern.

Manipulating DNA

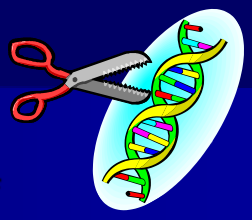
- Scientists use their knowledge of the structure of DNA and its chemical properties to study and change DNA molecules. Different techniques are use to extract DNA from cells, to cut DNA into smaller pieces, to identify the sequence of bases in a DNA molecule, and to make unlimited copies of DNA

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)

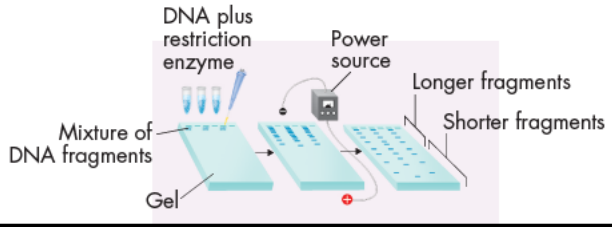
Cutting DNA

- 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



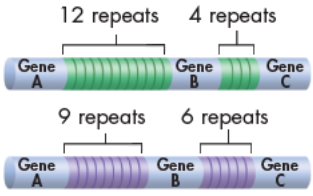
GTAACGAATTCACGCTT
CATTGCTTAAGTGCGAA

- 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



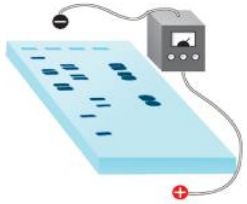
Personal Identification

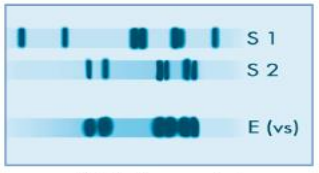
- No individual is exactly like any other genetically—except for identical twins, who share the same genome.
- Chromosomes contain many regions with repeated DNA sequences that do not code for proteins. These vary from person to person. Here, one sample has 12 repeats between genes A and B, while the 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 (analyzes sections of hair, blood, sperm or skin tissue)



Personal Identification

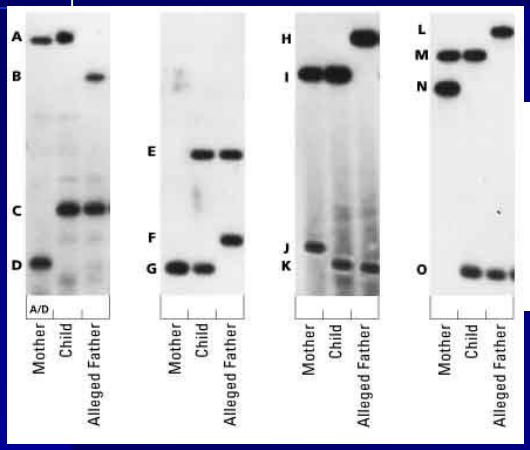
- In DNA fingerprinting, restriction enzymes first cut a small sample of human DNA into fragments containing genes and repeats. Note that the repeat fragments from these two samples are of different lengths.
- Next, gel electrophoresis separates the restriction fragments by size.
- A DNA probe then detects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





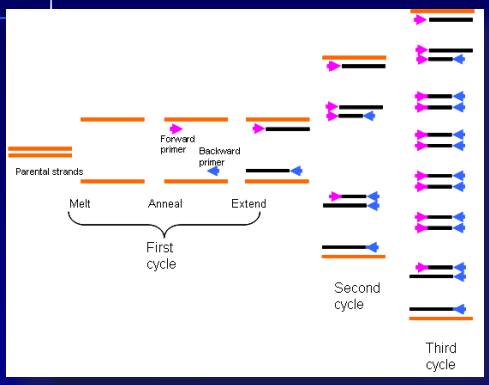
DNA fingerprint

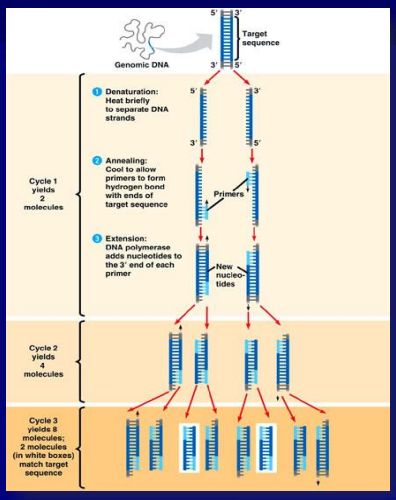
FORENSIC ANALYSIS



- **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)

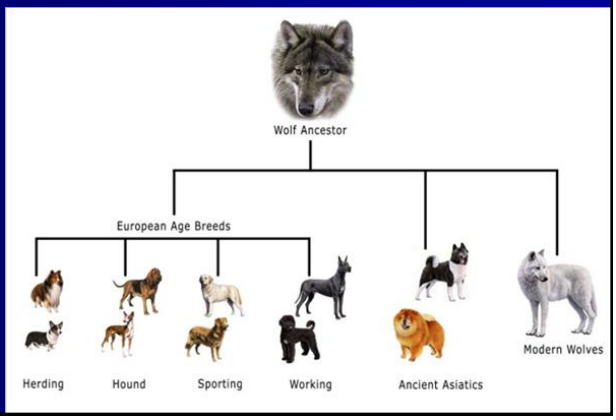




- **Selective breeding** is the method of breeding that allows only those individual organisms with desired characteristics to produce the next generation.
  - Humans use selective breeding, which takes advantage of naturally occurring genetic variation, to pass wanted traits on to the next generation of organisms.
  - Example: Dog breeds, development of corn, etc.
- **Hybridization** is a breeding technique that involves crossing dissimilar individuals to bring together the best traits of both organisms
  - Hybrids are often better than their parents

Selective Breeding

- First true "dog" is a species of gray wolf- most breeds of dog today are only a couple hundred years old
- Dogs were selectively bred for particular traits and behaviors
- Through selective breeding the dog has developed into hundreds of breeds



- **Inbreeding** is the continued breeding of individuals with similar characteristics to maintain the desired characteristics of a line of organisms
  - Helps to ensure that the characteristics that make each breed unique will be preserved
  - Most members of a breed are genetically similar and so the probability of a genetic defect is higher in this population, ex. Joint deformities in German Shepherds

Transgenic Organisms

- Transgenic means an object contains genes from another foreign organism.
- A gene from one organism can be inserted into the genetic makeup of another organism to "correct" or change an organism's traits.

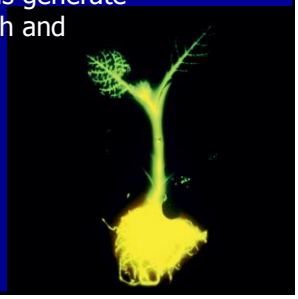
Transforming Bacteria

- Recombinant DNA (small piece of targeted DNA) is used to **transform** bacteria.
  - The DNA is joined to a small circular bacterial DNA molecule known as a **plasmid**.
- Recombinant DNA is possible because DNA molecules from all organisms share the same chemical structure.
- If transformation is successful, the **recombinant** DNA (TARGETED DNA STRAND) is integrated into one of the chromosomes of the bacterial cell

Uses of Recombinant DNA

- **Recombinant human insulin**
  - A form of insulin made from recombinant DNA that is identical to human insulin
  - Used to treat diabetics who are allergic to preparations made from beef or pork insulin (pigs & cattle)
  - We can now use bacteria
- **Recombinant human growth hormone (HGH)**
  - Administered to patients whose pituitary glands generate insufficient quantities to support normal growth and development.
- **Recombinant DNA in Plants**

- This plant was grown from a tobacco cell transformed with the firefly luciferase (causes bioluminescence)gene.



Uses of genetic engineering

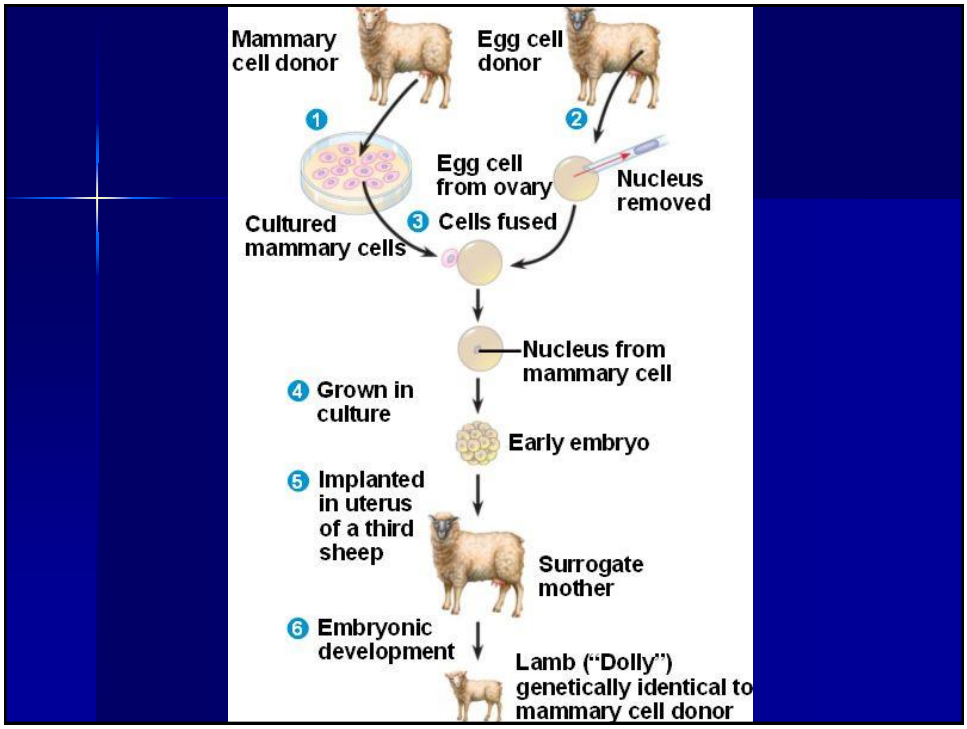
- Genetically modified organisms (GMO)
  - enabling plants to produce new proteins
    - **Protect crops from insects:** BT corn
      - corn produces a bacterial toxin that kills corn borer (caterpillar pest of corn)
    - **Extend growing season:** fishberries
      - strawberries with an anti-freezing gene from flounder
    - **Improve quality of food:** golden rice
      - rice producing vitamin A improves nutritional value

Cloning

- **Clone** is a member of a population of genetically identical cells produced from a single cell.
  - Researchers are hoping that cloning could possibly help endangered species.
- Controversy: Cloned animals may suffer from genetic defects and health problems.

Dolly The Sheep

- First mammal to be cloned from an adult somatic (body) cell
- Used the process of nuclear transfer
  - where the cell nucleus from an adult cell is transferred into an unfertilized **oocyte** (developing egg cell) that has had its nucleus removed
- Born on July 5th, 1996 and she lived until the age of six, at which point she died from a progressive lung disease.


Mammary cell donor
Egg cell donor
1
2
Egg cell from ovary
Nucleus removed
Cultured mammary cells
3 Cells fused
Nucleus from mammary cell
4 Grown in culture
Early embryo
5 Implanted in uterus of a third sheep
Surrogate mother
6 Embryonic development
Lamb ("Dolly") genetically identical to mammary cell donor