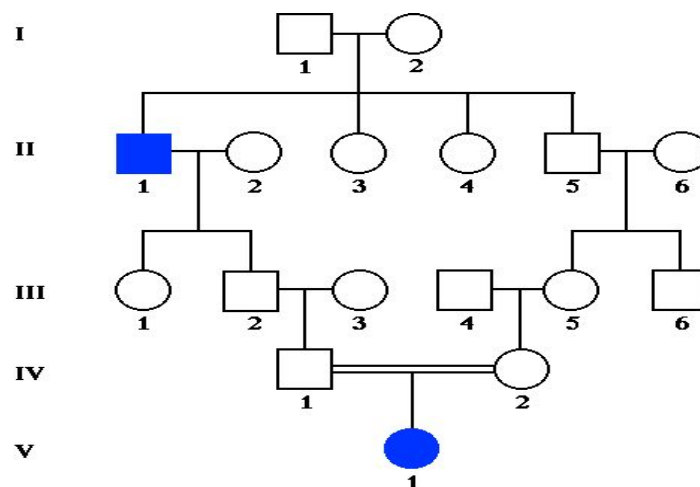
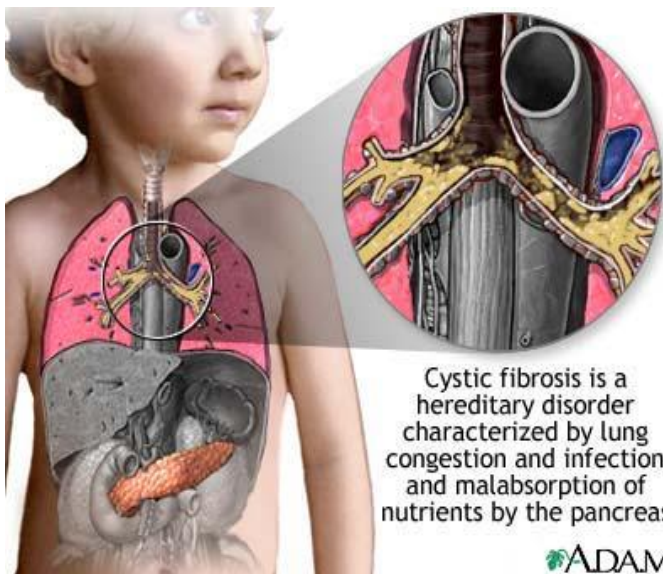
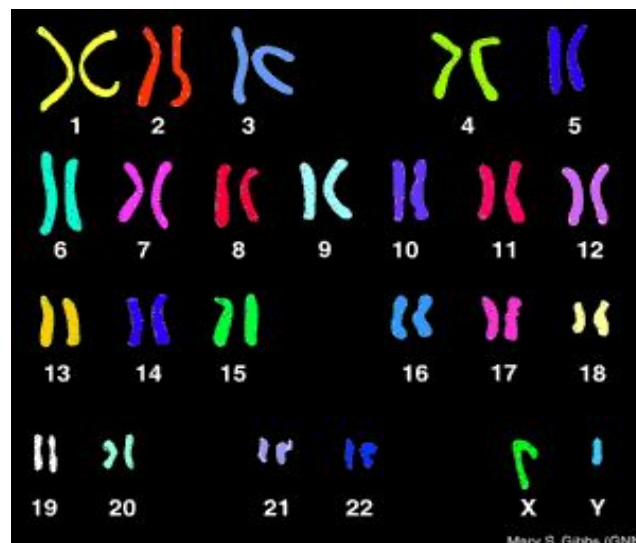


Human Heredity Notes

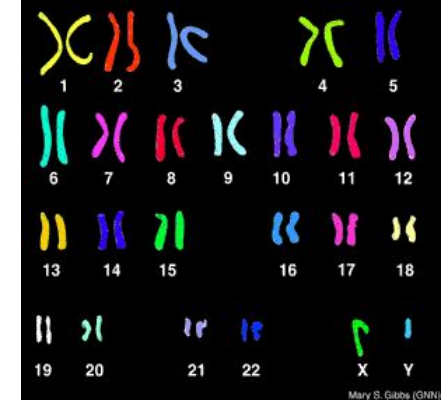


Chapter 14

Page 391

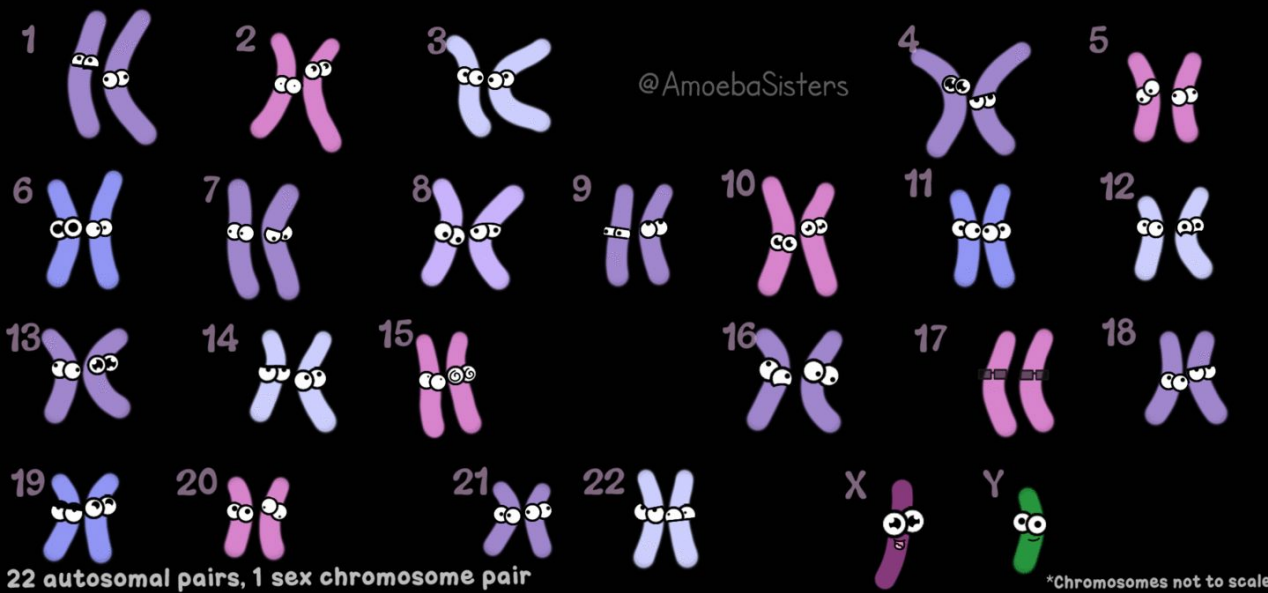


Genome



- The full set of genetic information that an organism carries in its DNA
- Scientists look at chromosomes to study the genome
- Chromosomes are best seen in metaphase (mitosis) since they are in the middle of the cell
- The chromosomes are cut and placed into a picture called a karyotype

HUMAN KARYOTYPE: 46 CHROMOSOMES*

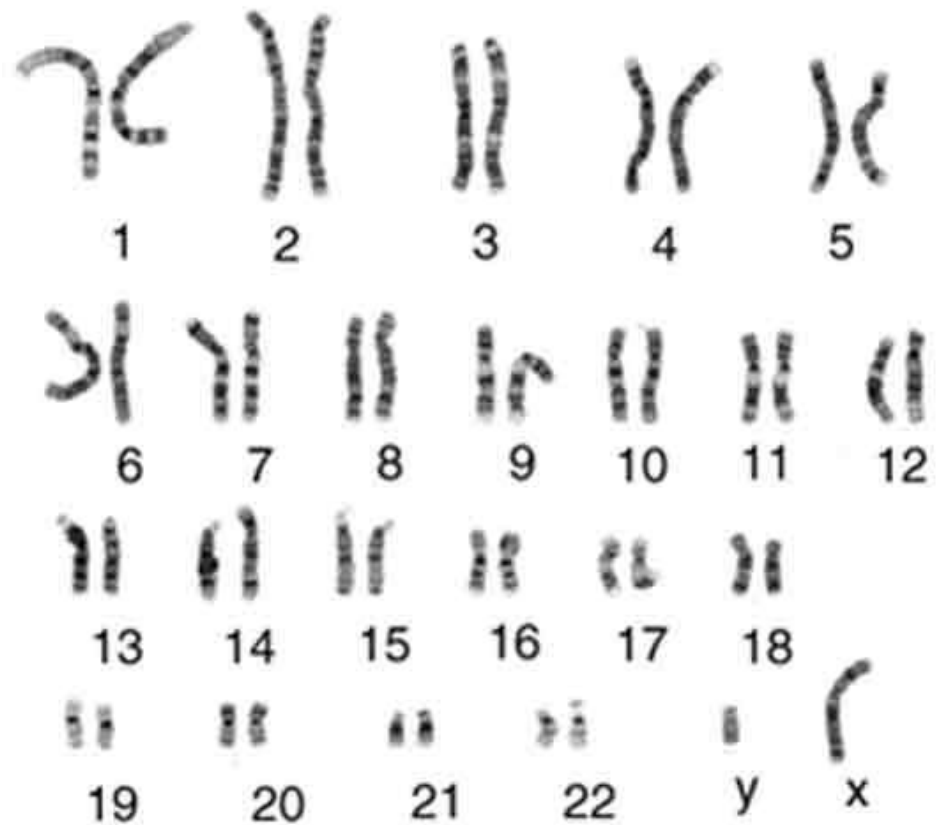


Karyotype

- A diagram that shows the complete diploid set of chromosomes grouped in pairs
- Arranged in decreasing size
- Humans have 46 total or 23 pairs
- Chromosomes are aligned in pairs
- The last set or the 23rd pair are called the sex chromosomes

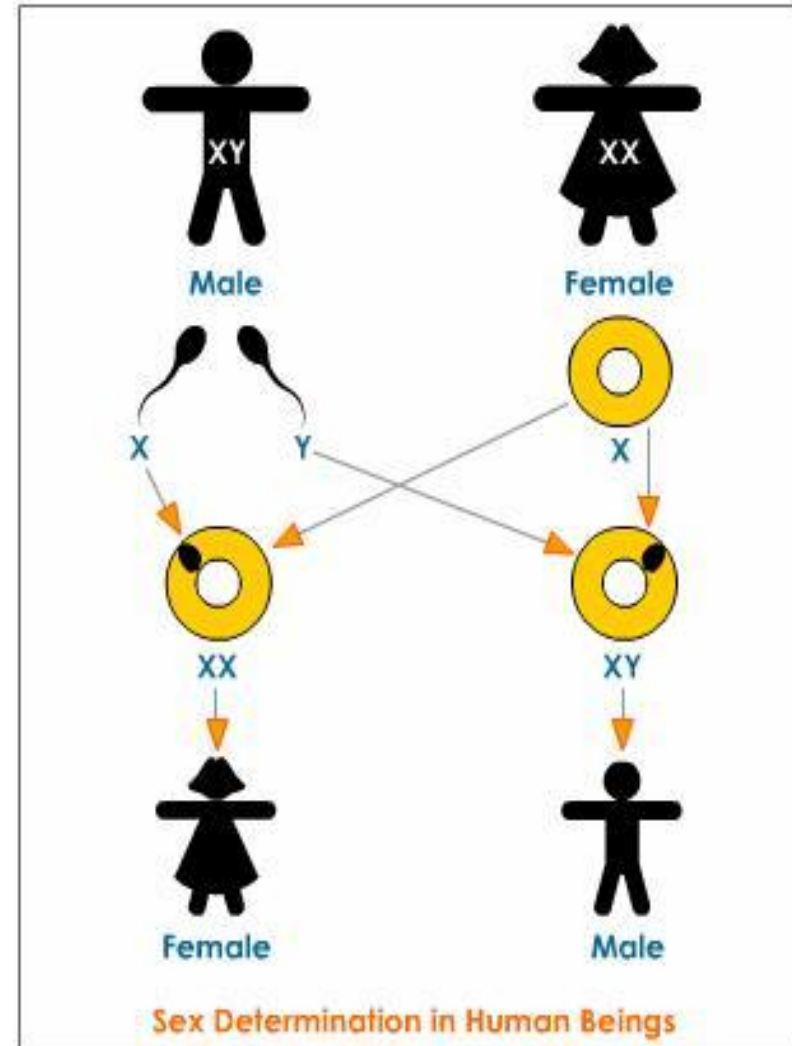
Karyotype

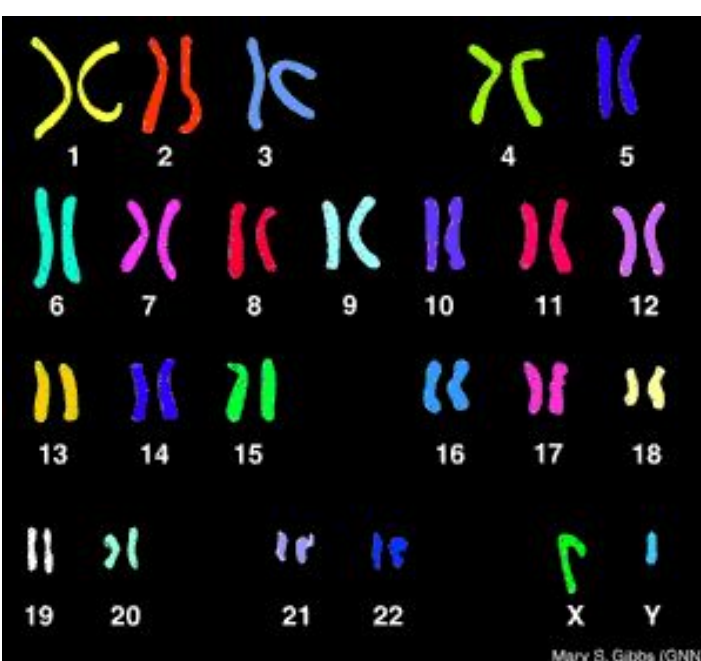
- 23rd pair is the **sex chromosomes**
 - Males = **XY**
 - Females = **XX**
- Sets **#1-22** are called **autosomes**
 - These determine the rest of the **traits**



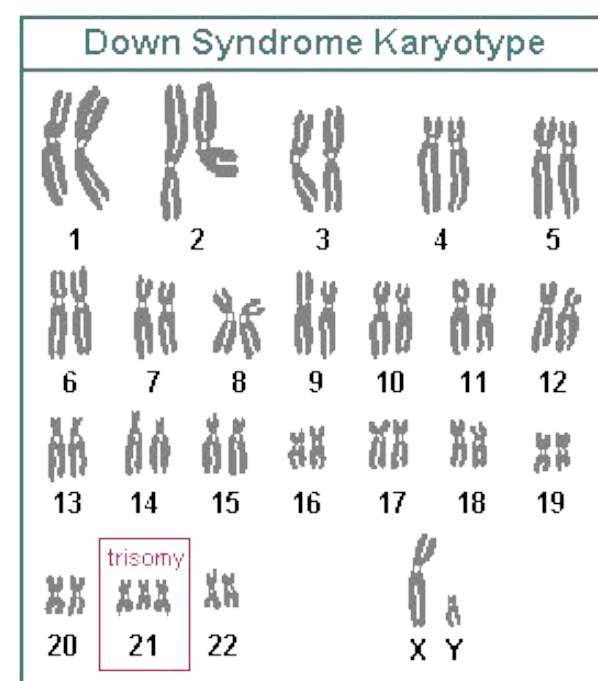
Sex Determination

- Gender is determined by the sex chromosomes (#23 pair)
- Female = XX
- Male = XY
- Males determine gender of baby
 - Why?

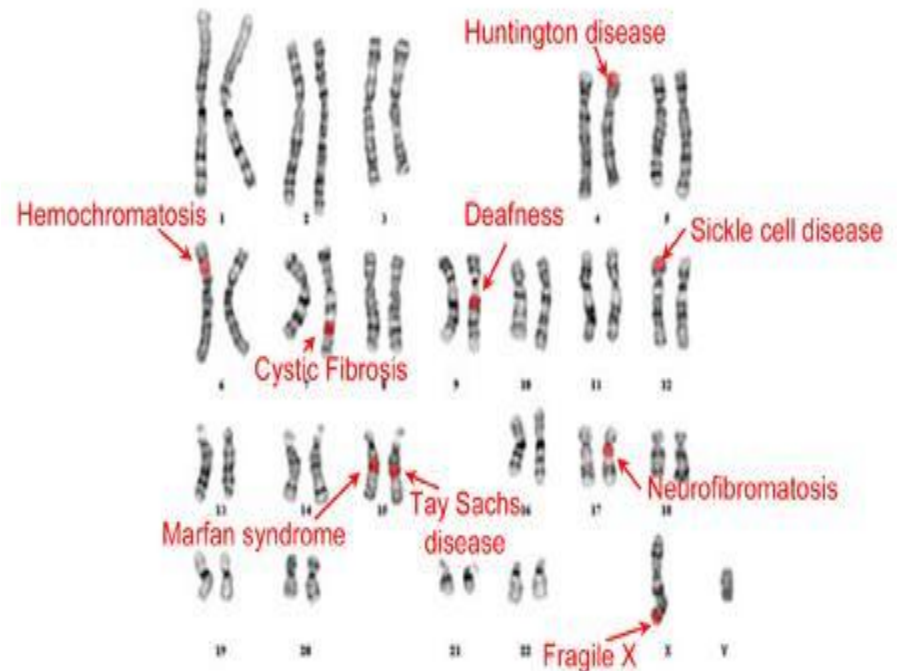
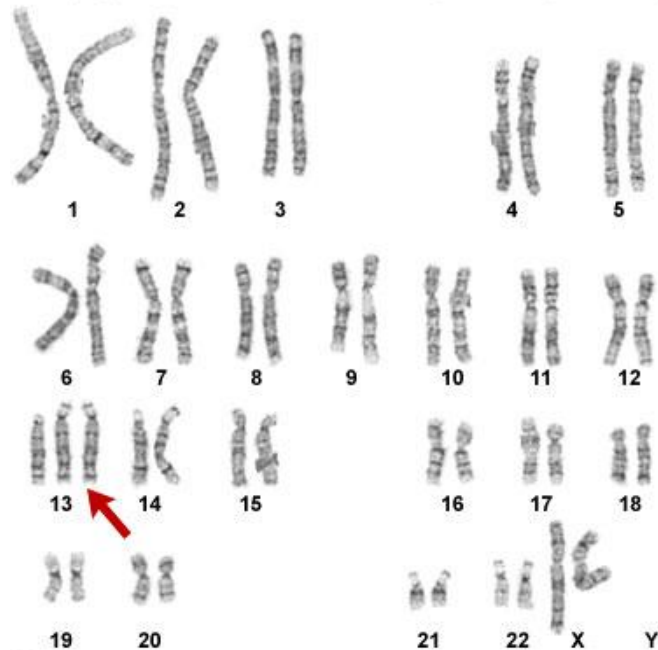




Karyotype examples

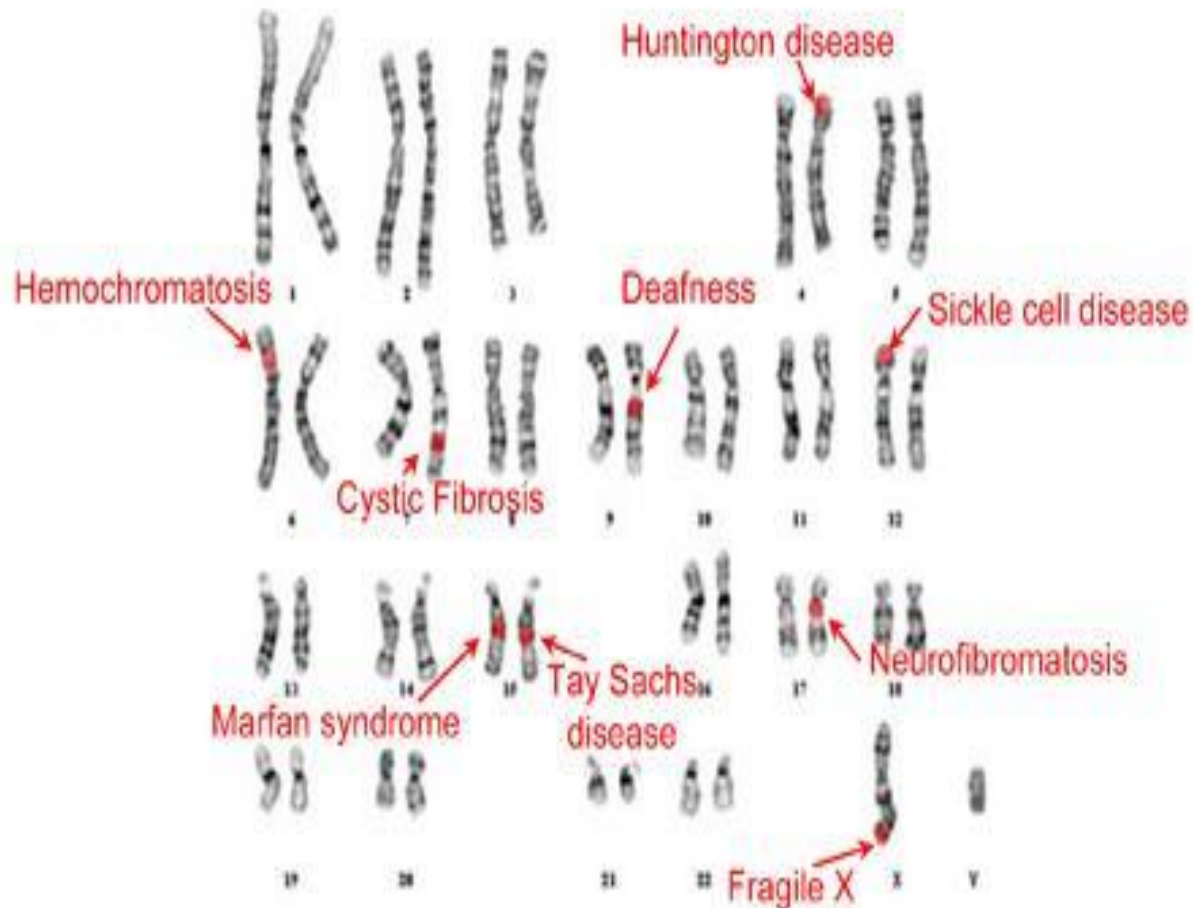


Karyotype from a female with Patau syndrome (47,XX,+13)



Karyotype Journal

- Answer all questions into your journal books



CARRIER

- an individual who is heterozygous for a trait
ex: Yy, Tt, Rr, Bb
- Has 1 dominant allele that covers 1 recessive allele
 - Doesn't show the recessive trait since the dominant has overpowered it
 - They are "carrying" the trait to possibly pass it on to the next generation/offspring

Sex-Linked Traits

- Traits controlled on the “X” chromosome
- Males are most affected the most
 - Since they have only 1 X-chromosome
- Females less likely to get it
 - Have 2 X-chromosomes
- Examples: color blindness, hemophilia
- *How do you do a Punnett square properly??*
- *Let's practice* 😊

Practice of Sex-linked Traits

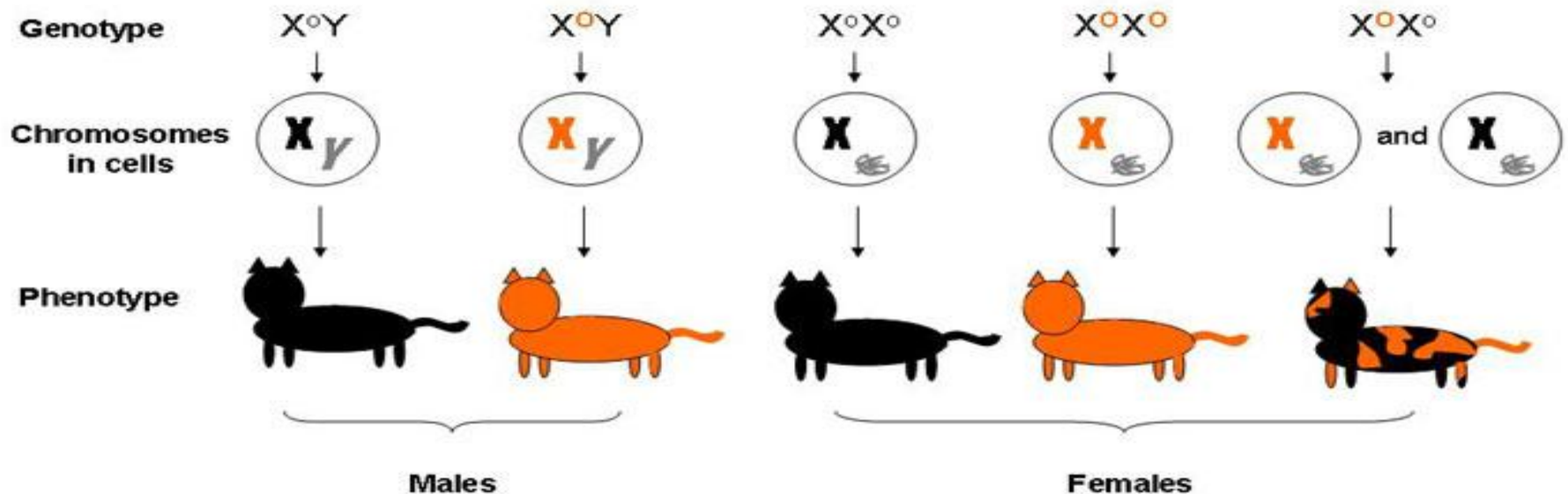
- Where do males get their X chromosome from?
- How do you write the genotype for a female with a sex-linked trait?
- How do you write the genotype of a male with a sex-linked trait?
- Cross a color blind male to a carrier female.
 - List the genotypes (male/female & colorblind)

Chromosome inactivation

- Female have 2 doses since they have two X-chromosomes
- X is necessary for development in males/females
- One X-chromosome will stop functioning
 - This creates a Barr body
- The “Y” chromosome continues to work determining male characteristics

Calico Cats

- This can affect the coat color of cats
- Example of calico cats
 - **Black color**= black on the X (either male or female)
 - **Orange color**= orange on the X (either male or female)
 - **Calico**= has 1 black and 1 orange
 - can only be a female cat

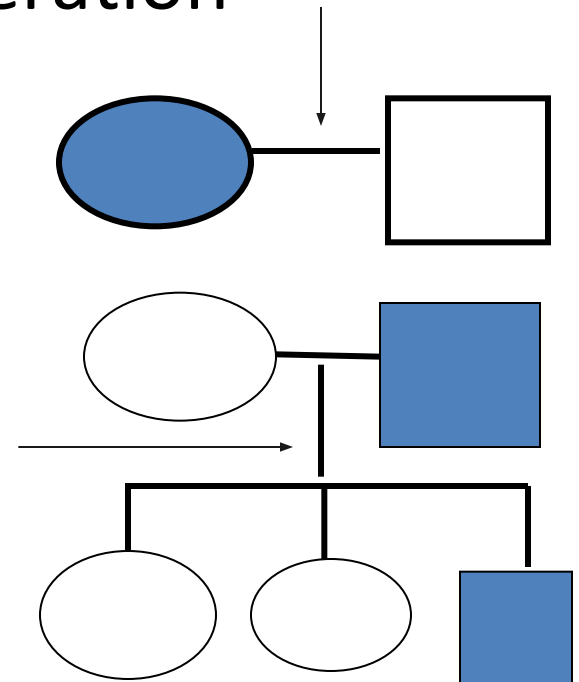
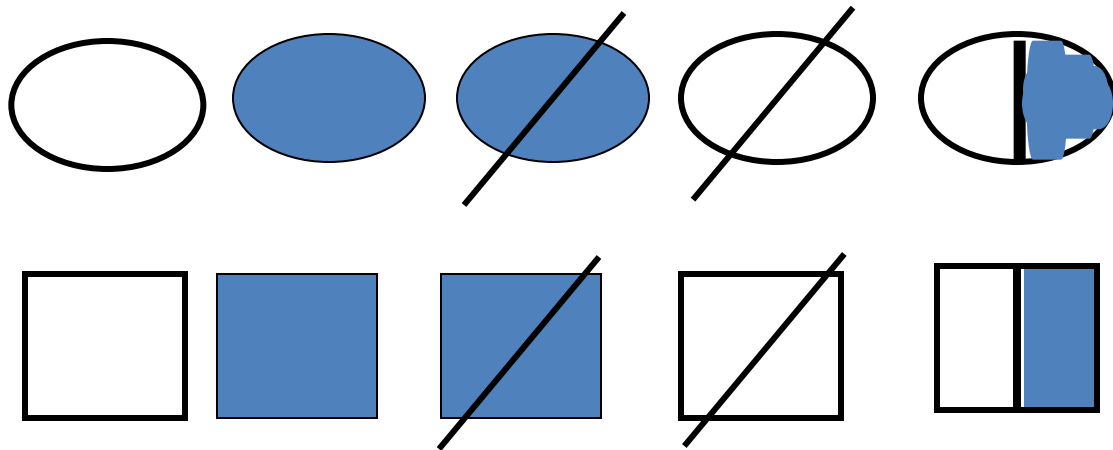


Pedigree

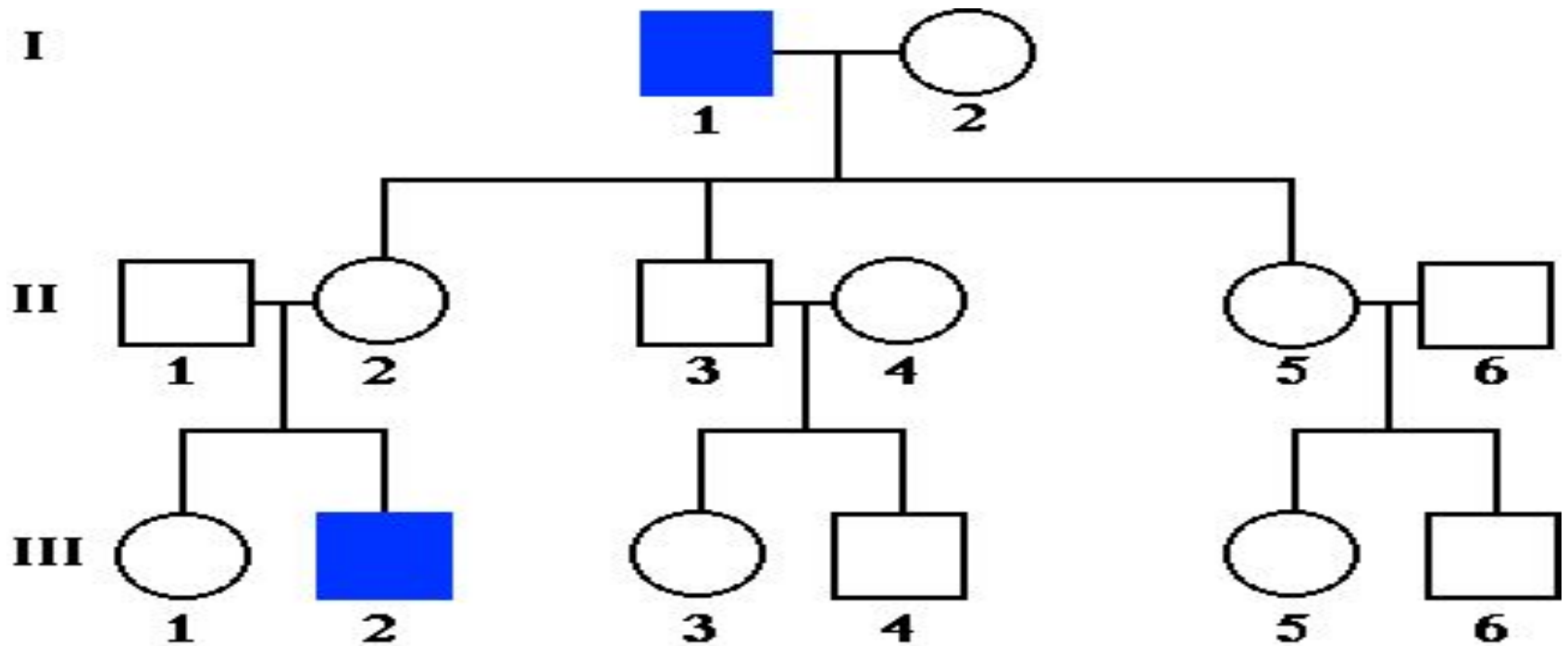
- A chart that shows the pattern of inheritance for a particular trait within a family
- **Dominant traits:** every generation (Male/female)
- **Recessive traits:** skips a generation
- **Autosomal traits:** trait is on chromosome #1-22
- **Sex-linked traits:** on the X chromosome (male)

Pedigree

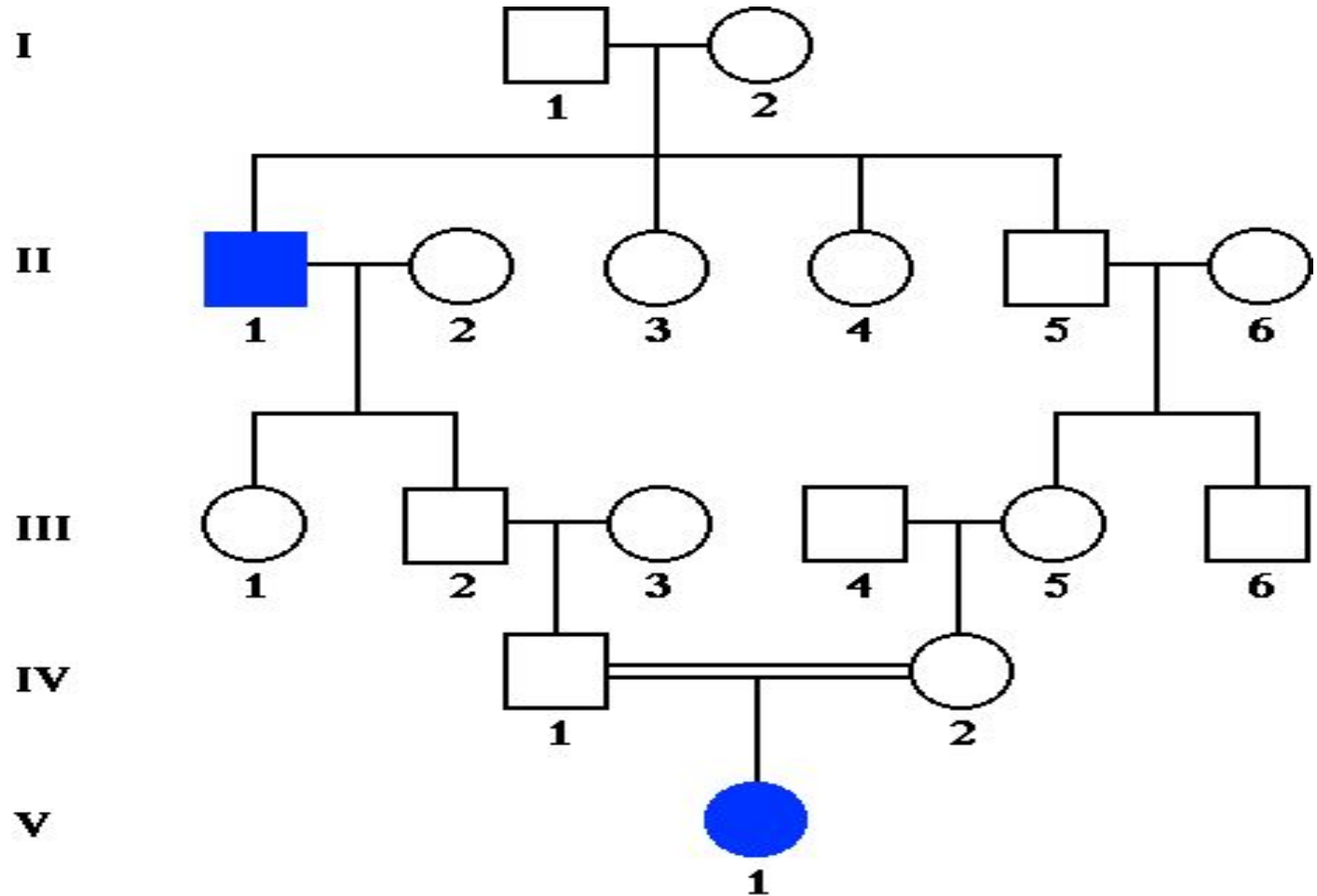
- What does each represent?
- **Roman numerals** stand for the generation number
- **Numbers** stand for the number of an individual in a particular generation



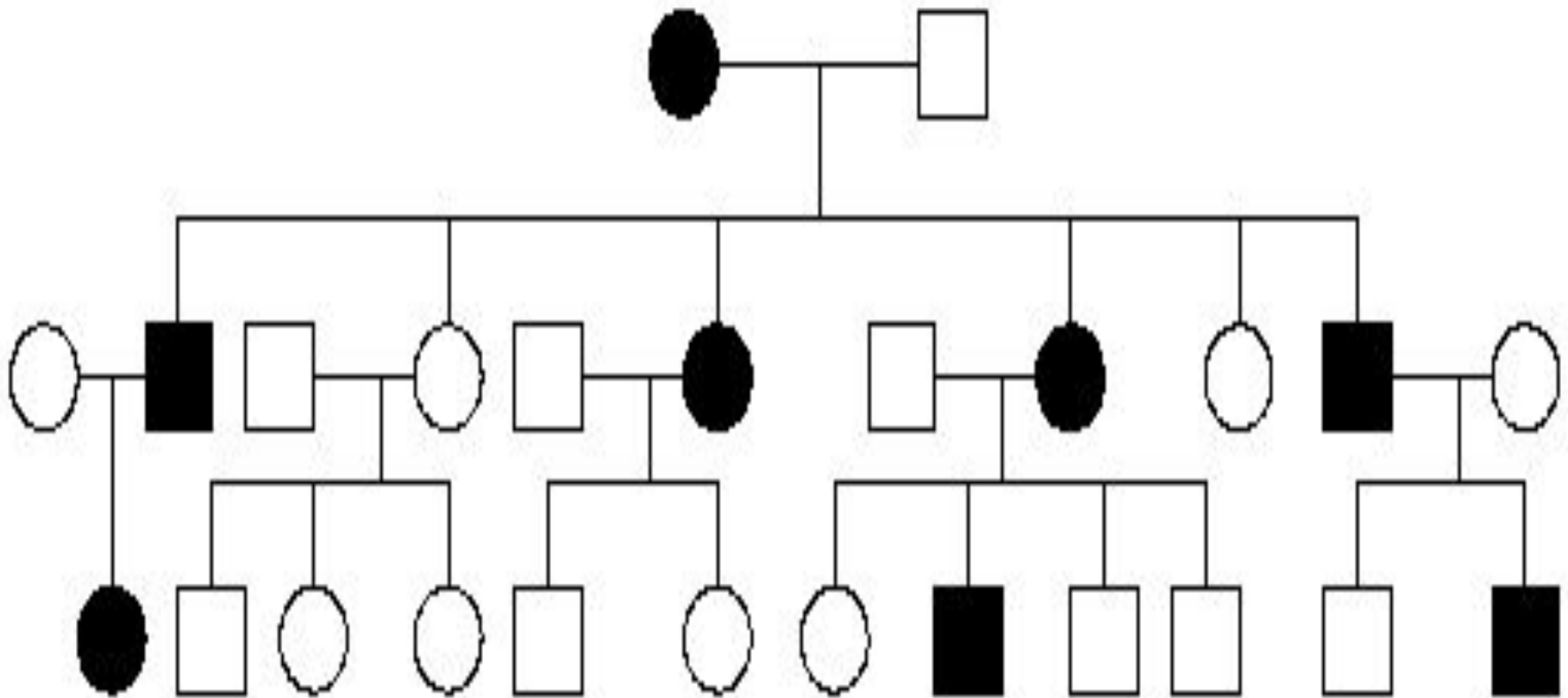
What type of Pedigree?



What type of pedigree?

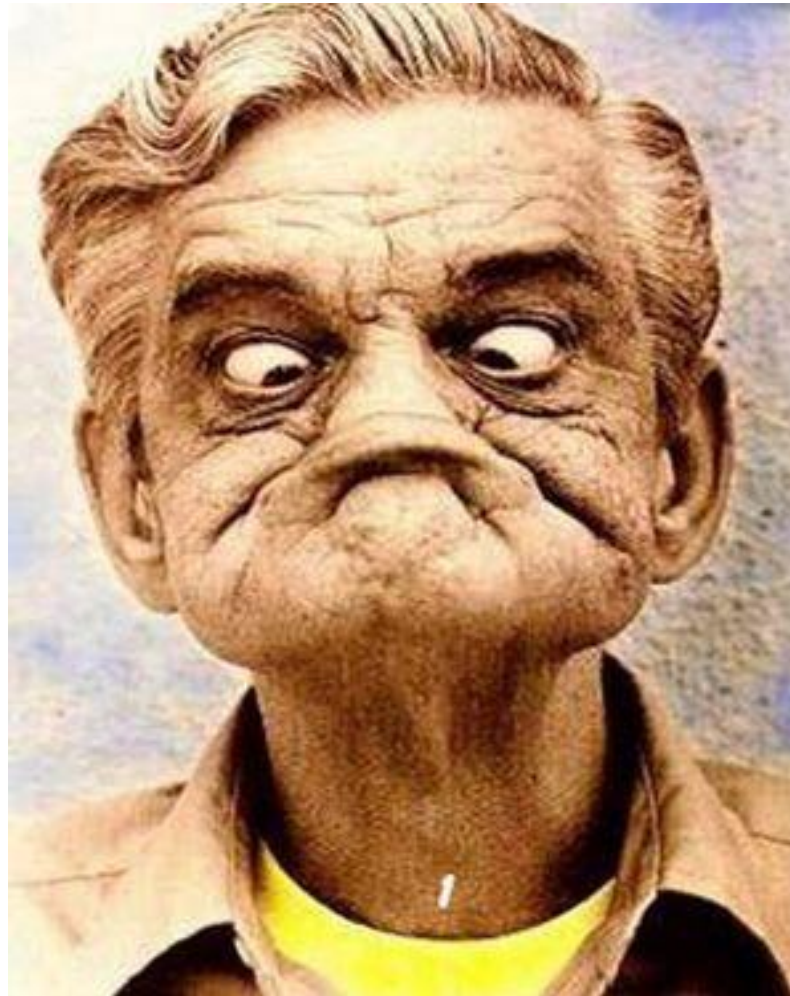


What type of Pedigree?



I AM MY OWN GRANDPA

- https://www.youtube.com/watch?v=zelsxXDy_jlc



Amoeba Sisters

- <https://www.youtube.com/watch?v=Gd09V2AkZv4>

–Questions

- A couple with the ability to taste PTC have 2 grown sons and 1 grown daughter. The sons have the ability to taste PTC. Their daughter is a PTC non-taster. She married a PTC non-taster man, and they have 2 sons. What is the genotype of the grandsons?

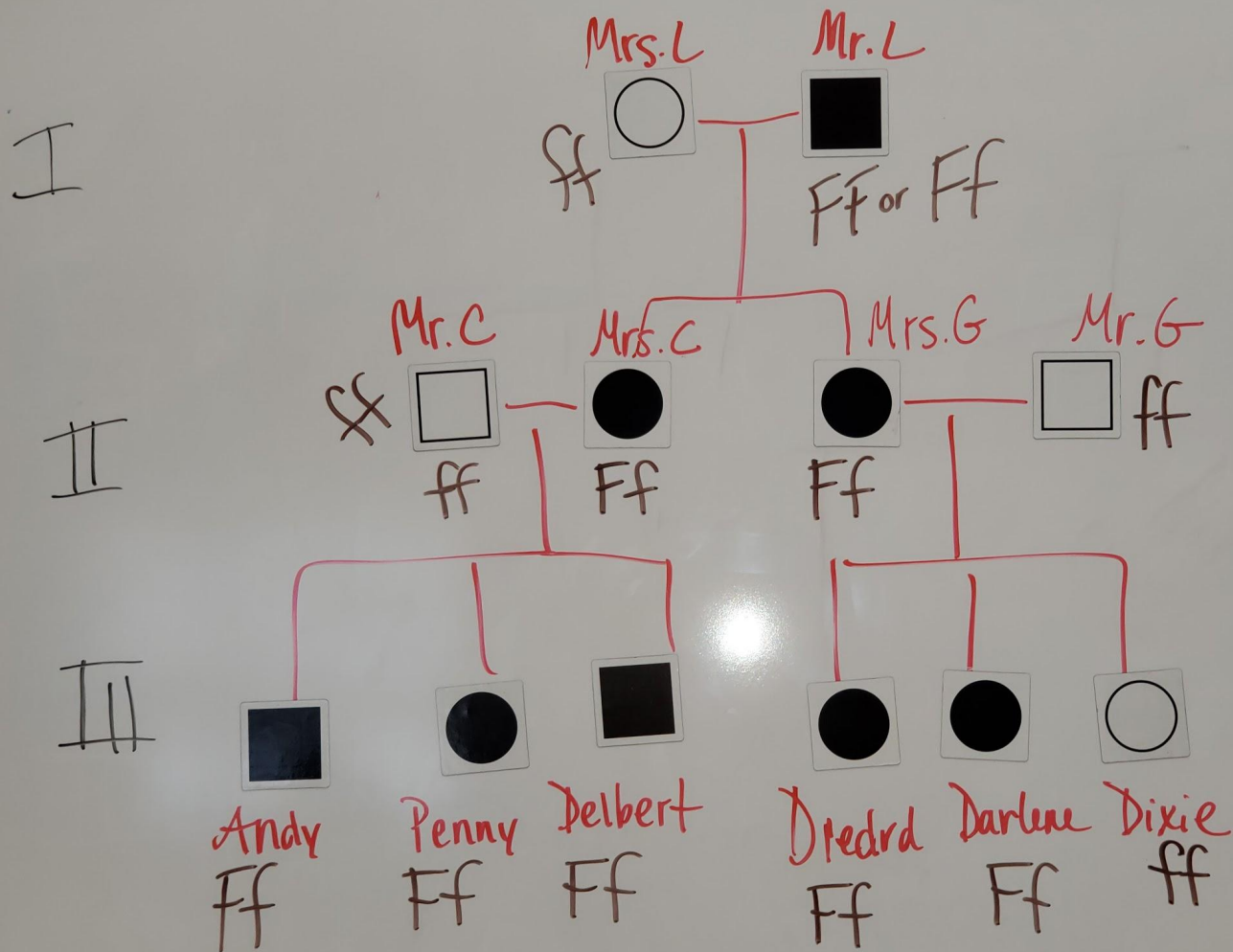
Pedigree Practice in Journals Pg 108

- Draw a pedigree showing a married couple with 4 kids, all boys?
- How would you draw a married couple with 2 kids, the daughter is married and has a son, the older brother is not married?
- How would you draw a family that has 2 boys and 2 girls, the girls are carriers, and only the boys are married? All of the 2 granddaughters (cousins) are carriers just like their grandmother.

Pedigree Lab

- Open up journals books (page 108/109) and let's practice making some pedigree's

Pedigree #2



Homework

- Figure out Rudolph's family history of red-noses
- Since a Red nose is recessive and Black noses are dominant
 - Red Noses =
 - Black Noses=

To do

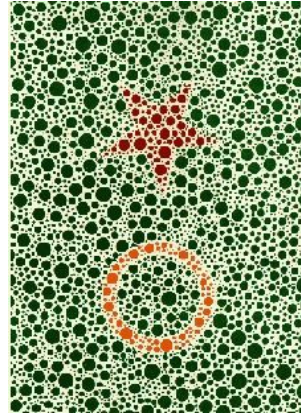
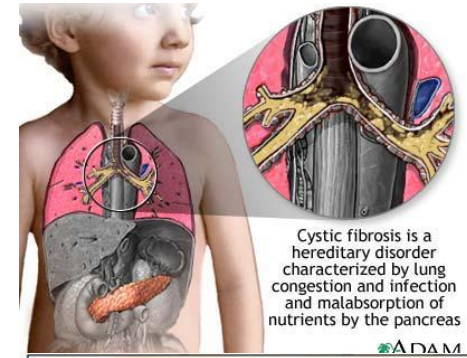
1. Complete the Symbol column (circles or squares)
2. Complete the pedigree on the back of his Family History (there should be 4 generations)
3. Don't forget each reindeer's name

Pedigree Review Quiz

- Answer the questions onto page 110

Genetic Disorders

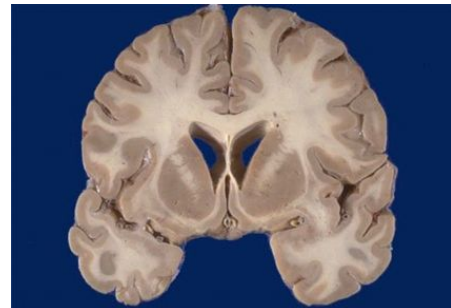
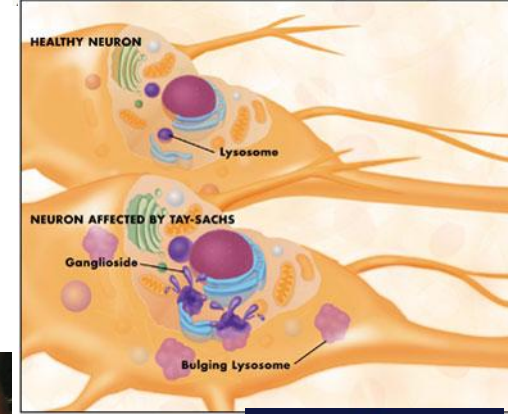
- Cystic Fibrosis
- Albinism
- Tay-Sachs Disease
- Huntington's Disease
- Achondroplasia
- Sickle cell anemia disease
- Hemophilia
- Color Blindness



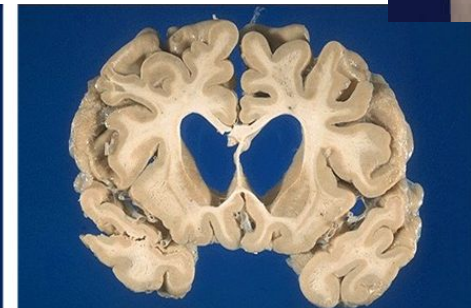
Normal red blood cell



Sickled red blood cell



WT



HD

Do you Remember?

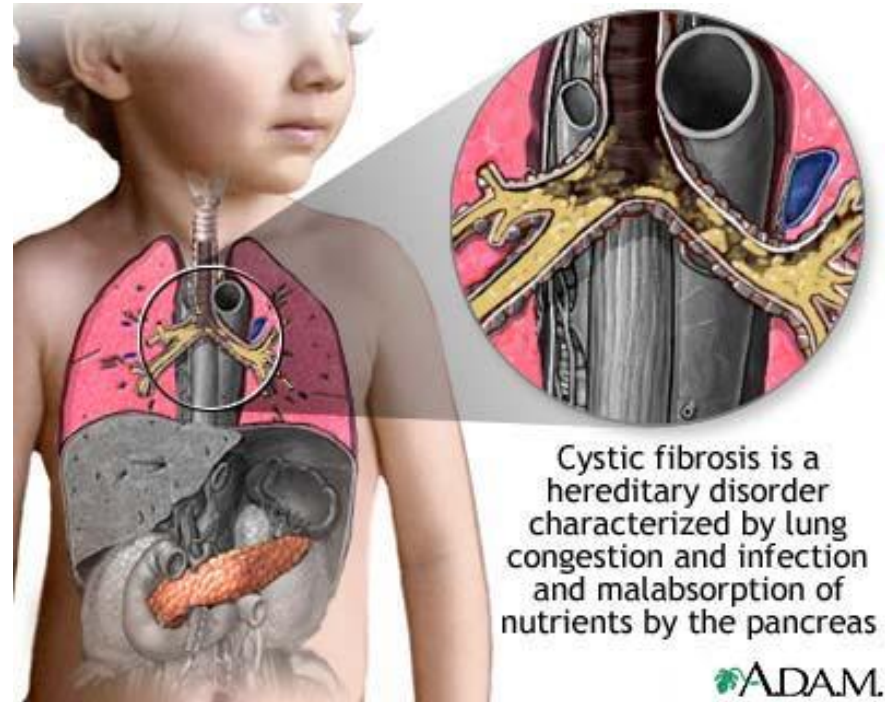
- What does it mean to have an autosomal trait?
- What does it mean to have a sex-linked trait?
- What is a dominant trait mean?
- What does a recessive trait mean?

DNA changes = human traits affected

- Changes in a gene's DNA sequence can change proteins by altering their sequences which directly affects their phenotype

Cystic Fibrosis

- Caused by a defective gene that codes for a membrane protein
- Effect= Excessive mucus produced in lungs
- No cure
- Treatment includes drugs to clean out mucus
- Occurs primarily in the white population
- **Recessive**
- **Autosomal**

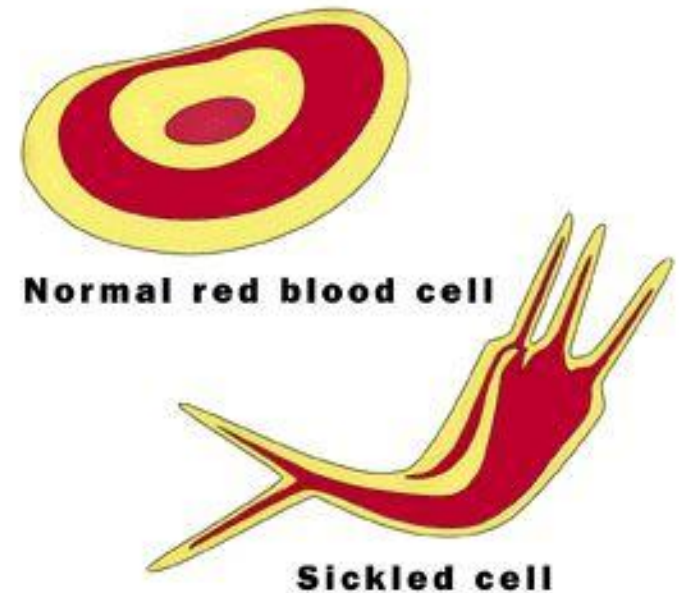


Cystic fibrosis is a hereditary disorder characterized by lung congestion and infection and malabsorption of nutrients by the pancreas

 ADAM.

Sickle Cell Anemia Disease

- Cause= defective membrane protein
- Effect= RBC become sickle shaped
- No cure
- Treatment: blood transfusions
- Occurs usually in the African American population
- **Recessive**
- **Autosomal**



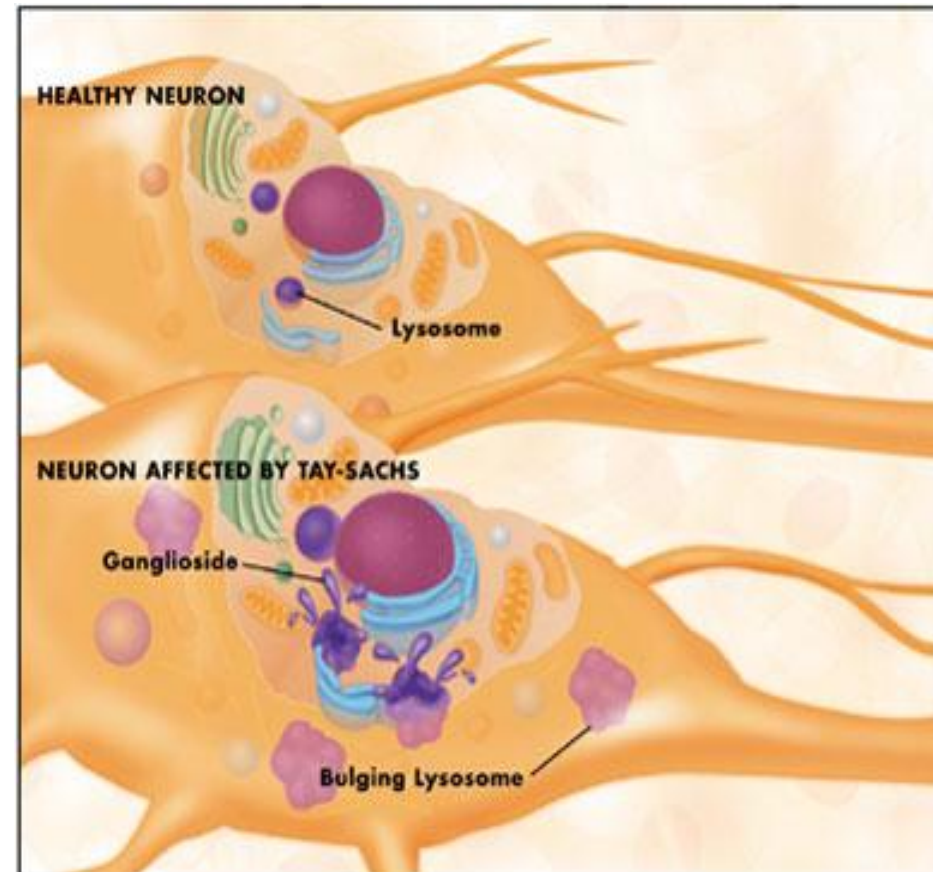
Huntington's Disease

- Cause= extra protein build up in brain
- Effect= Nervous system deteriorates during middle age
- No cure or treatment
- Death in middle age
- **Dominant**
- **Autosomal**



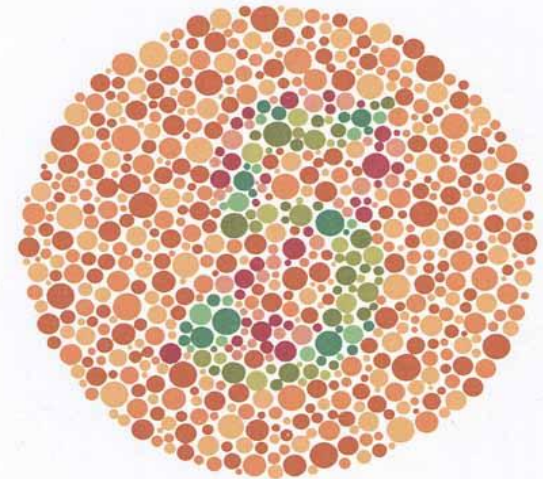
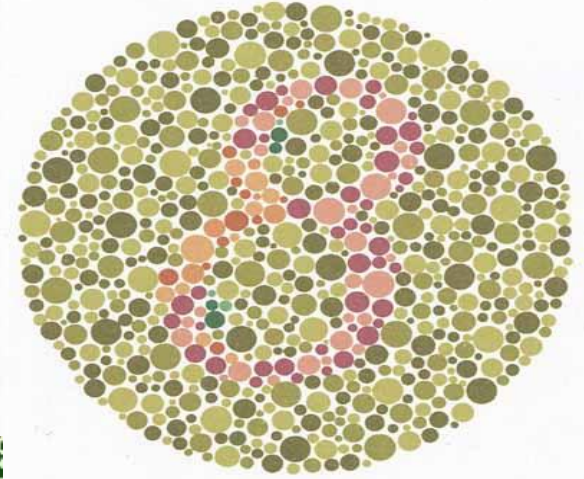
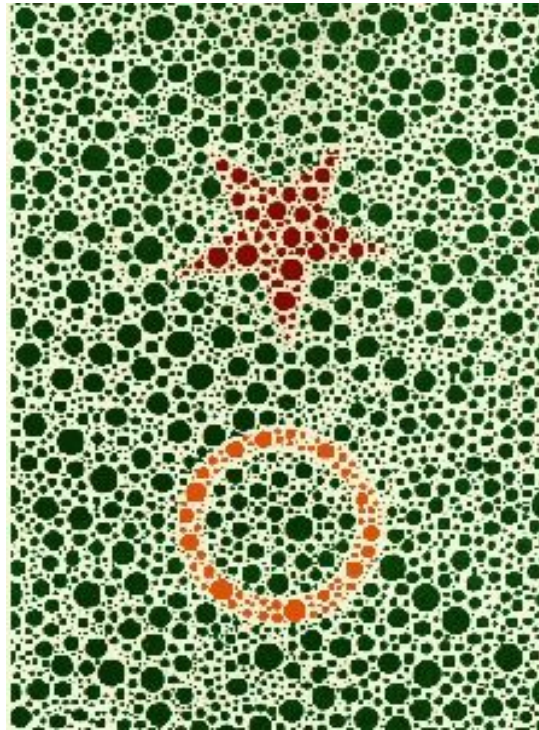
Tay-Sachs Disease

- Absence of a gene to break down fatty substances in brain
- **Effect**= Build-up of fatty deposits on brain
- Mental disabilities
- No cure or treatment
- Death by age 5
- Occur primarily in the Jewish population
- **Recessive**
- **Autosomal**

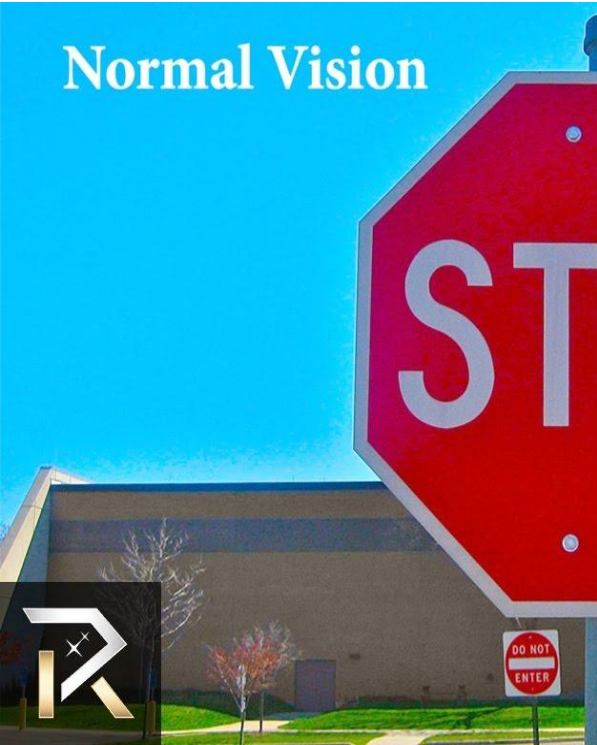


Color Blindness

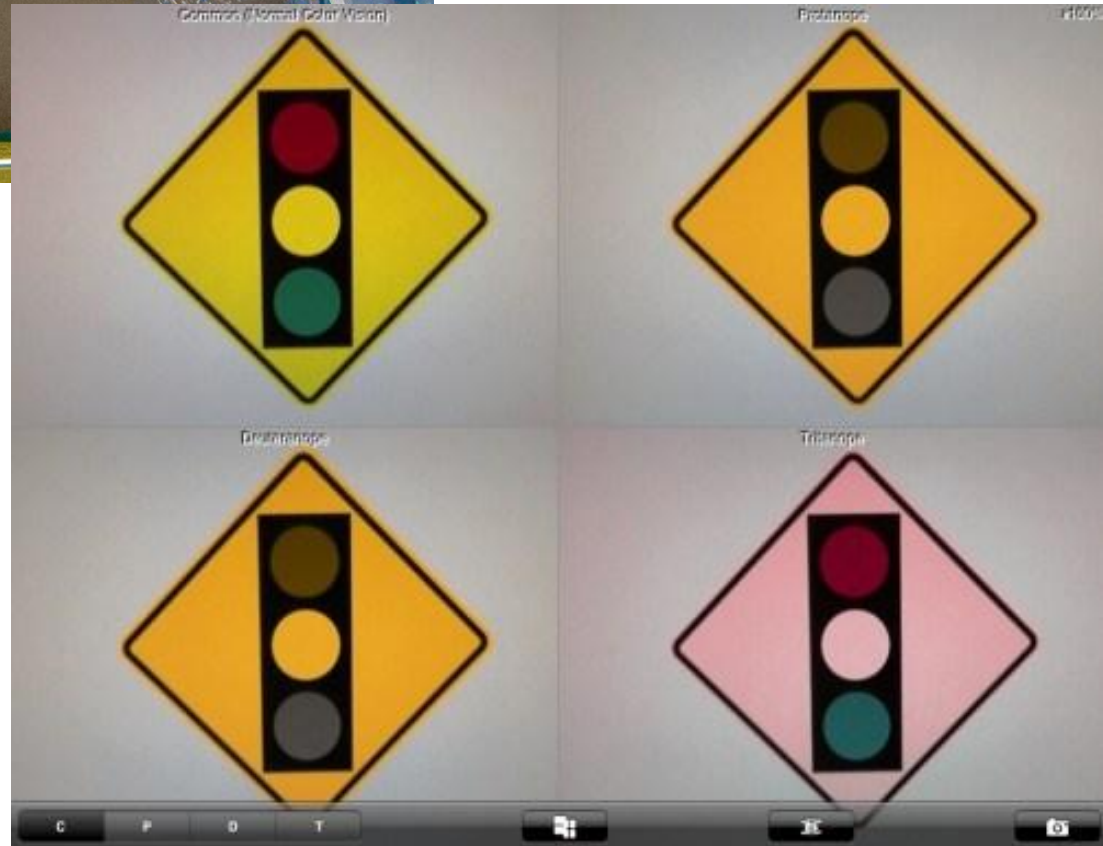
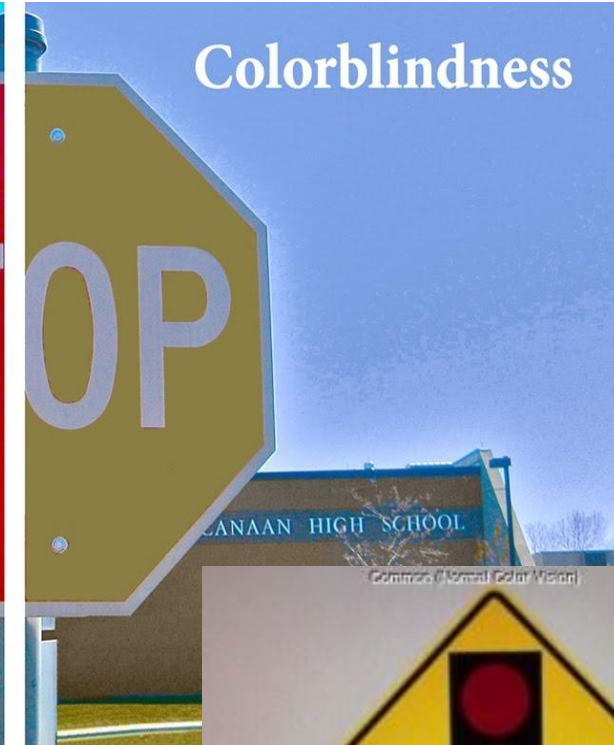
- Defect in cones of the eye
- Effect= Cannot see colors
- Occurs mostly in males
- **Recessive**
- **Sex-linked**



Normal Vision



Colorblindness



Hemophilia

- Gene defect with clotting blood
- Effect= Blood does not clot and can increase bleeding
- No cure
- Treatment: must take clotting factor VIII daily
- Effects males mostly
- **Recessive**
- **Sex-linked**



Achondroplasia

- A defective gene that affects bone growth
- Results in dwarfism
- Anyone
- **Dominant**
- **Autosomal**



Genetic Advantages

- Affects those homozygous for sickle cell
 - **NN** = normal cells
 - **SS** = sickle cells
 - **SN** = both sickle and normal cells together

Heterozygote is more resistant to malaria

Genetic Advantages

- Cystic Fibrosis heterozygote

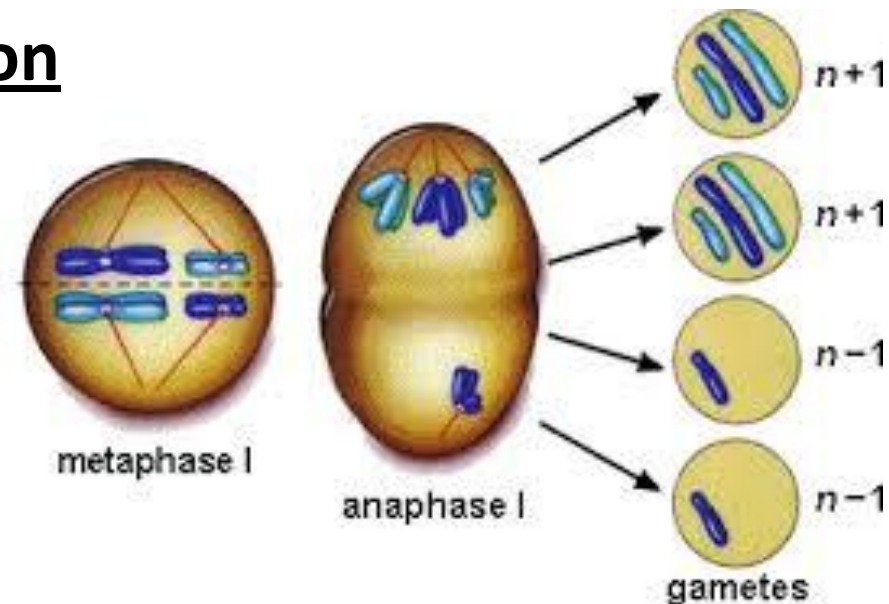
CC = normal

cc = has Cystic Fibrosis

Cc = normal but resistant to typhoid fever

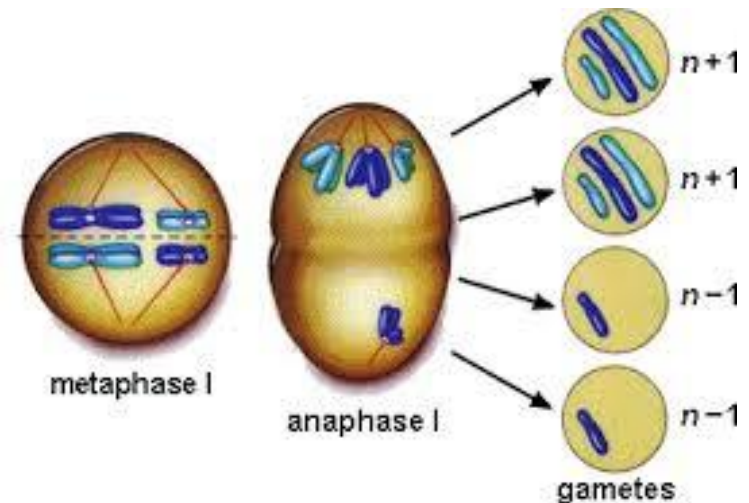
Chromosomal Disorders

- Means an error in **Meiosis**
- Each human **gamete** should have **23** chromosomes
- But what happens if the **homologous** chromosomes do **NOT** separate correctly
 - This is called **Non-disjunction**



Non-disjunction

- A gamete with an abnormal number of chromosomes leading to a disorder of chromosome number
- The gamete will have 1 copy of a chromosome
 - Example: Turner's Syndrome
 - A female with only 1 X-chromosome



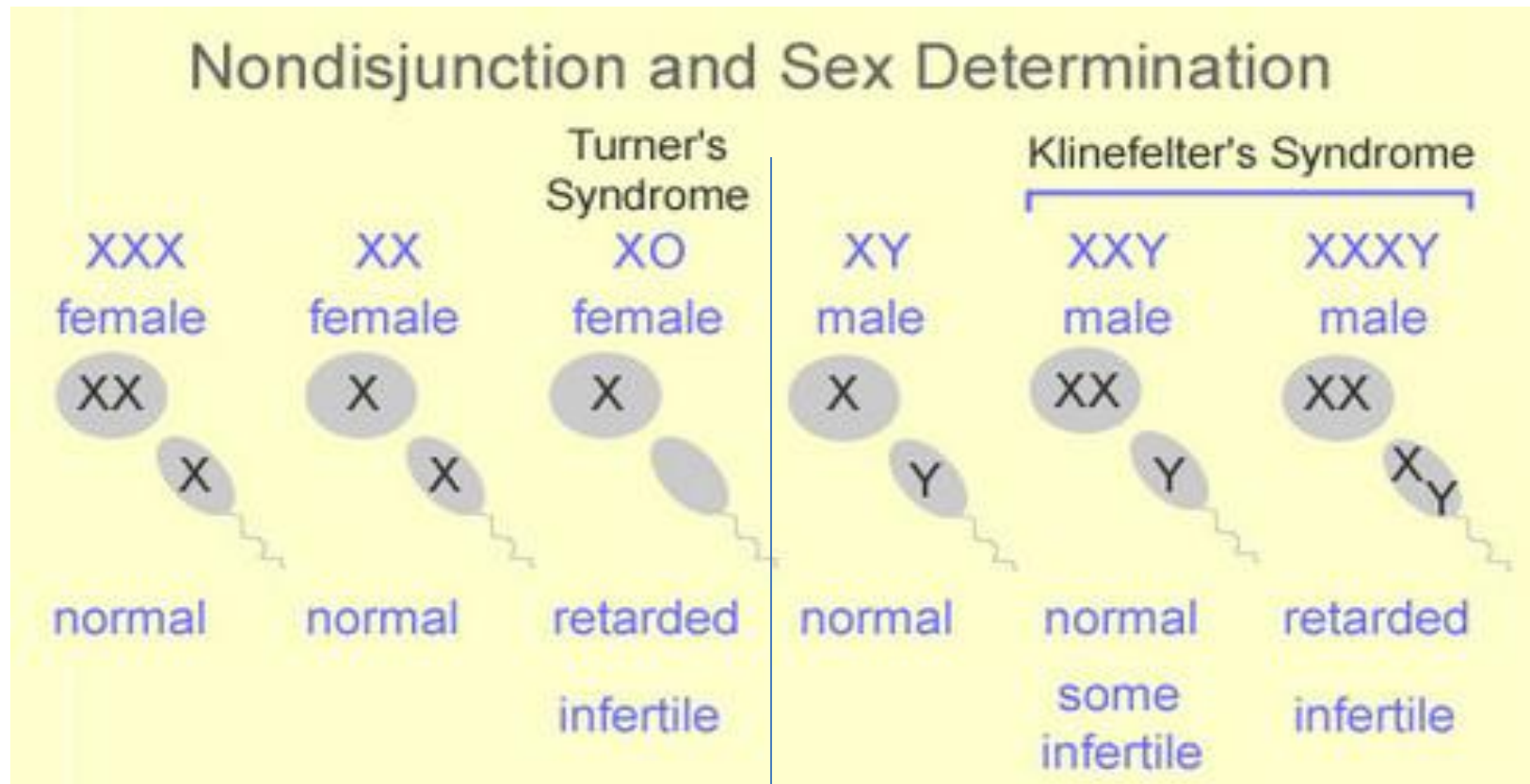


Down Syndrome



- Gamete could have 3 copies of a chromosome
- This is called Trisomy
- Example: Down Syndrome
 - 3 copies of #21 chromosome
- Example: Klinefelter's Syndrome
 - A male who has XXY sex chromosomes

Non-disjunction in sex chromosomes



Studying the Human Genome

- **Human Genome Project**

- 13 year international effort
- Sequence all of the 3 billion base pairs of Human DNA
- This would identify ALL human genes
- Completed in 2003
- Helps us to locate diseases and disorders
- Someone can know their exact genetic make-up and what they can pass on to their offspring

Human Genome

