Human Genetics: Karyotypes

Arranged by size in homologous pairs.

Show chromosomal abnormalities. (# or shape)

*Do not show genetic disorders.

46 TOTAL Chromosomes

23 homologous pairs

What is the difference between an Autosome and a Sex-chromosome?

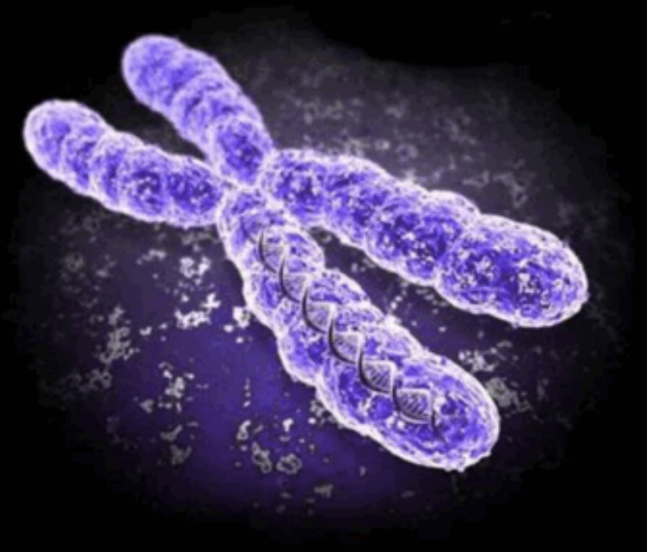
- Autosomes are the first 22 homologous pairs of human chromosomes that do not influence the sex of an individual.
- Sex Chromosomes are the 23rd pair of chromosomes that determine the sex of an individual.
  (Males XY, Females XX)

Genetic Diseases: Autosomal Traits

- Genes located on autosomes control autosomal traits and disorders.

2 Types of Traits:

- Autosomal Dominant
- Autosomal Recessive



Autosomal Recessive Traits

- In order to express the trait, [redacted] alleles must be present.
- What would be the genotype of an individual with an autosomal recessive trait? (A = dominant)

  [redacted]
- What would be the genotype of an individual without the autosomal recessive trait?

  [redacted]

  [redacted] – called a [redacted] because they carry the recessive allele and can pass it on to offspring, but they do not express the trait.

Autosomal Dominant Traits

- If [redacted] allele is present on the autosome, then the individual will express the trait.
- What would be the genotype of an individual with an autosomal dominant trait?

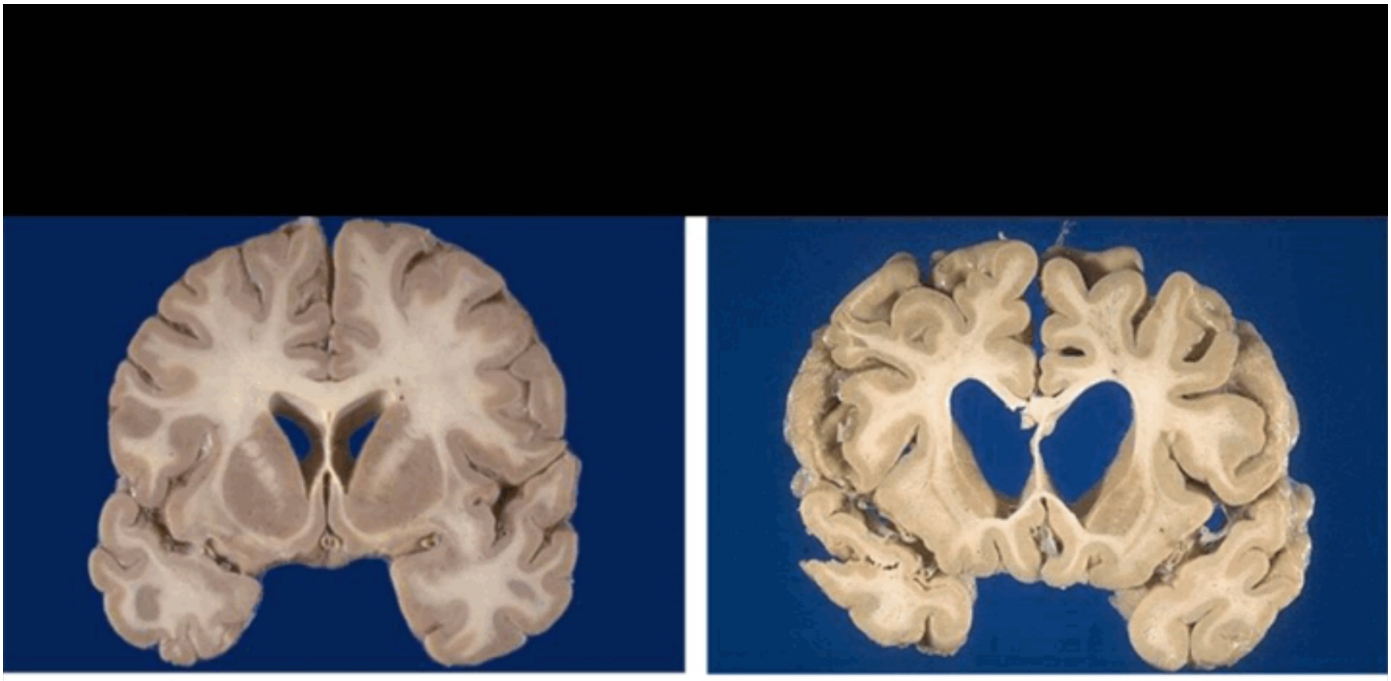
  [redacted] (Heterozygotes are affected)
- What would be the genotype of an individual without the autosomal dominant trait?

  [redacted]

Sex Linked

- Carried on the X chromosome and are usually recessive. NOT AUTOSOMAL!
- Men are more likely to be affected.
- Females can be carriers but NOT men!
- What would be the genotype of an individual with a sex linked disorder?

Name	Type of Inheritance	Description
Hemophilia	Sex-linked recessive	Inability to clot blood
Huntington's Disease	Autosomal dominant	Breaks down certain areas of the brain
Cystic Fibrosis	Autosomal recessive	Accumulation of thick mucus in lungs & digestive tract
Tay Sach's Disease	Autosomal recessive	Degeneration of brain and spinal cord. Fatal by two years of age
Phenylketoneuria (PKU)	Autosomal recessive	Unable to break down phenylalanine (a.a. in food/drinks)=Nervous system damage. Controlled with diet
Sickle-Cell Anemia	Autosomal co-dominant	Abnormal RBCs, slows blood flow, results in tissue damage


WT
HD

Chromosomal Disorders:

Down Syndrome

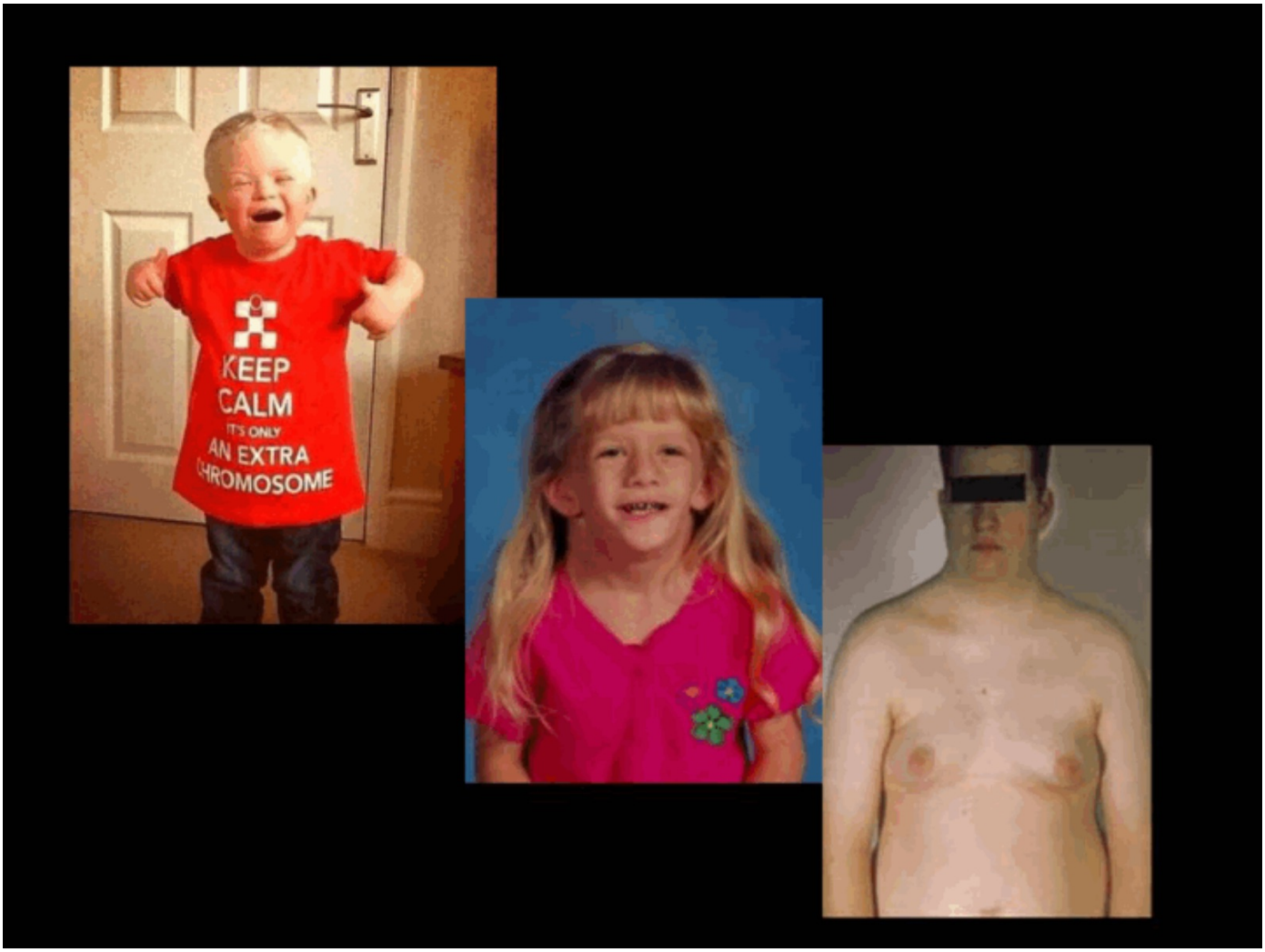
- Have an extra 21st chromosome (47 total).
- Trisomy 21; Short stature, some level of mental impairment, often heart defects, flat facial features.

Turner's Syndrome

- Females with one X chromosome. (45 total)
- A female who lacks ovaries and has underdeveloped sex characteristics

Klinefelter

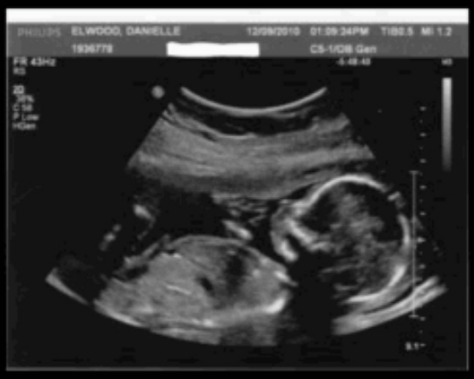
- Males with an extra X chromosome. (47 total)
- Males who are often sterile, some mental impairments, sometimes female characteristics.


KEEP
CALM
IT'S ONLY
AN EXTRA
CHROMOSOME

Detection of Chromosomal Disorders

In order to detect a chromosomal disorder several steps are taken:

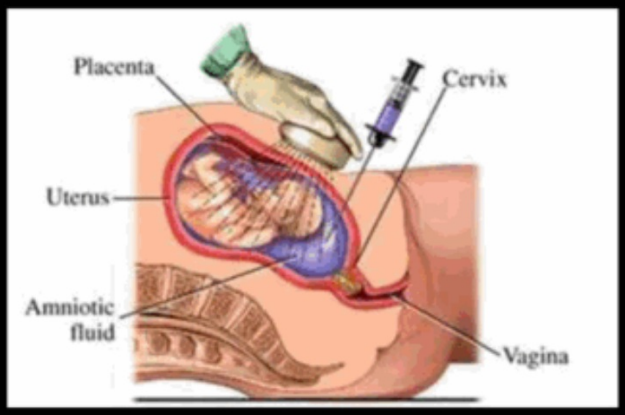
1) An ultrasound is done around 15-18 weeks.
2) An amniocentesis is done around 18 weeks.
3) A karyotype is done using the information obtained during the amniocentesis

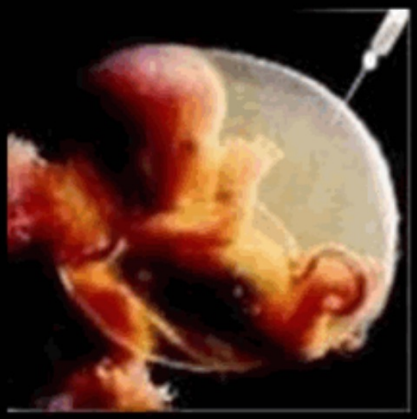


Amniocentesis

A small sample of the amniotic fluid (fluid filled sac around the baby) is taken.

When the sample is taken there are cells from the baby in the fluid. The DNA from those cells is extracted and a karyotype is made.

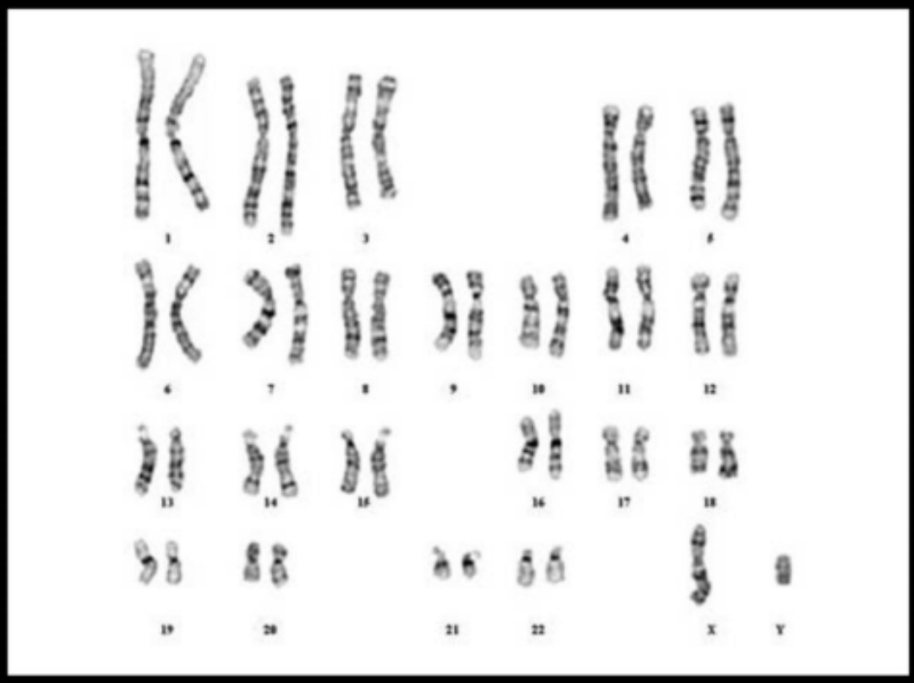




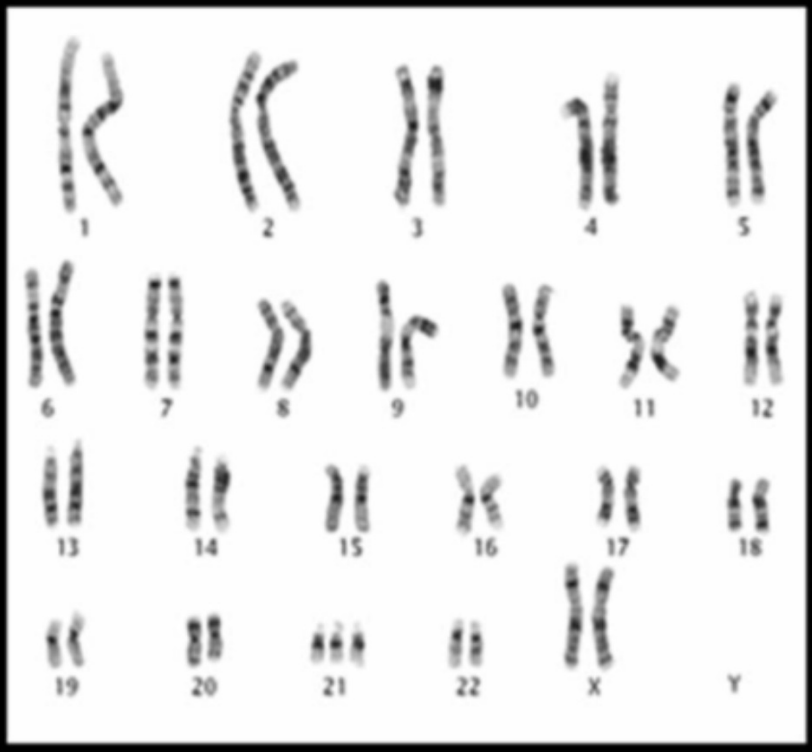
How to read a karyotype:

1) Count the total # of chromosomes. There should be only 46. Indicate any extra or shortages in chromosome #.
2) Identify the gender by the 23rd pair.
3) Do they have a chromosomal disease? It is possible to have a genetic disease and not be indicated on a karyotype. They only show chromosome disorders.

Examples:



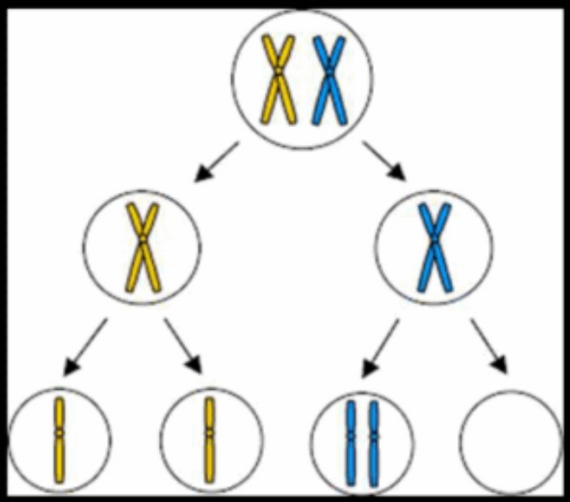
Examples:



How does this happen?

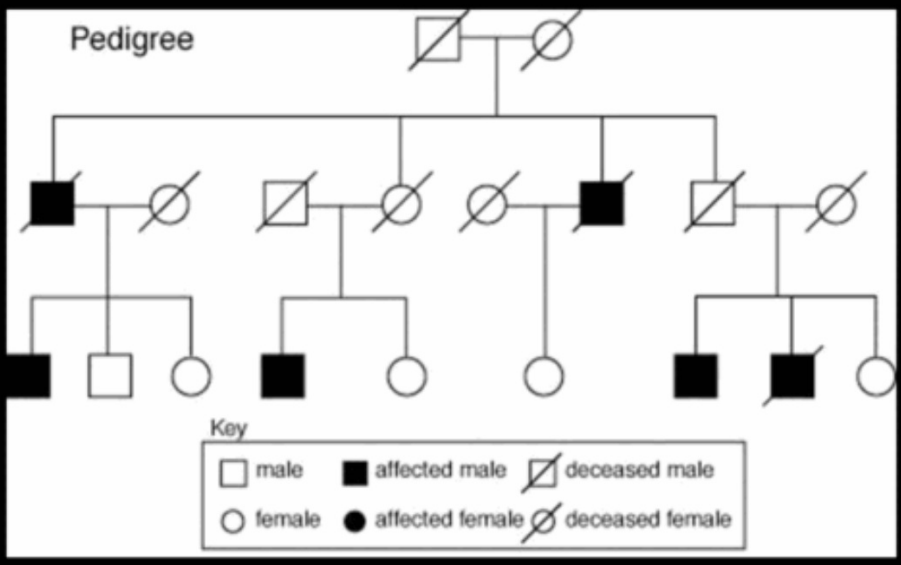
Nondisjunction

- Cause aneuploidy (wrong #) chromosomal disorders.
- Chromosomes do not separate properly during meiosis!



Pedigree Analysis

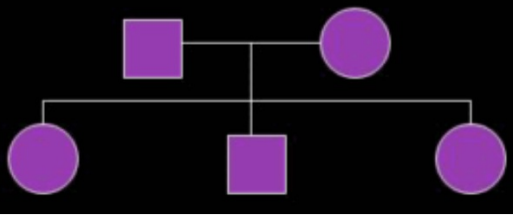
- A pedigree shows the relationship between parents and children over the generations and how a trait is passed down from one generation to the next.



How to Construct a Pedigree?

- A Pedigree is a visual showing the pattern of inheritance for a trait. (Family tree)

- Symbols and Rules:
- Male = Female =
- Affected = Unaffected = Carrier =
- Link parents together with a line and then make a vertical line to connect to offspring.

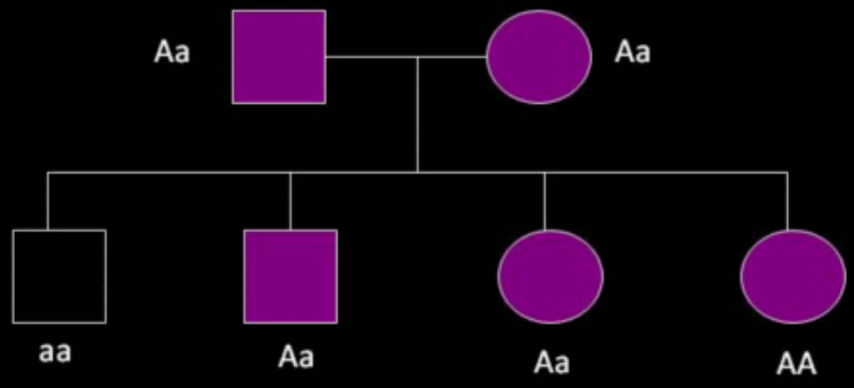


Autosomal Dominant Pedigree

- Draw a Pedigree showing a cross between Heterozygous parents that have 2 boys and 2 girls. (Show all possibilities)

<u>Genotypes of Affected and Unaffected</u>:

- AA and Aa = Affected aa = Unaffected



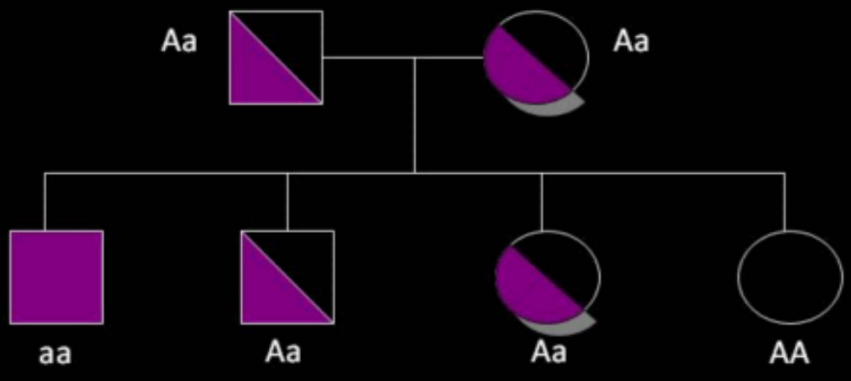
Autosomal Recessive Pedigree

- Draw a Pedigree showing a cross between Heterozygous parents that have 2 boys and 2 girls. (Show all possibilities)

<u>Genotypes of Affected and Unaffected</u>:

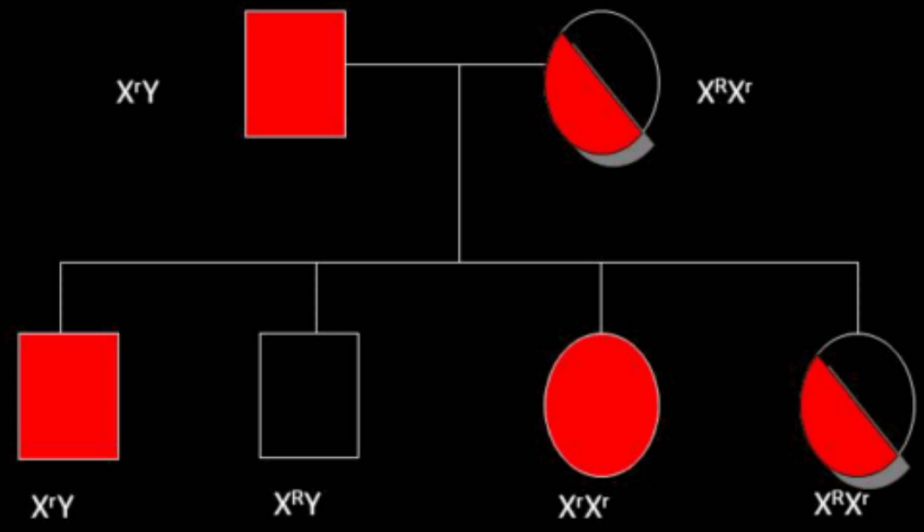
- AA=Unaffected Aa=Carrier aa=Affected

If 2 unaffected people have an affected child it's recessive.



Sex-Linked Recessive Pedigree

- Draw a Pedigree showing a cross between a affected male and a Carrier Female.
- Genotypes of Parents:
- Male = X^rY Female = X^RX^r

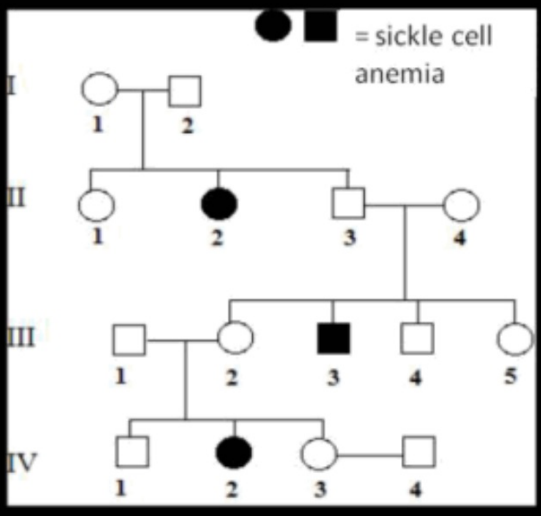


Pedigree Analysis

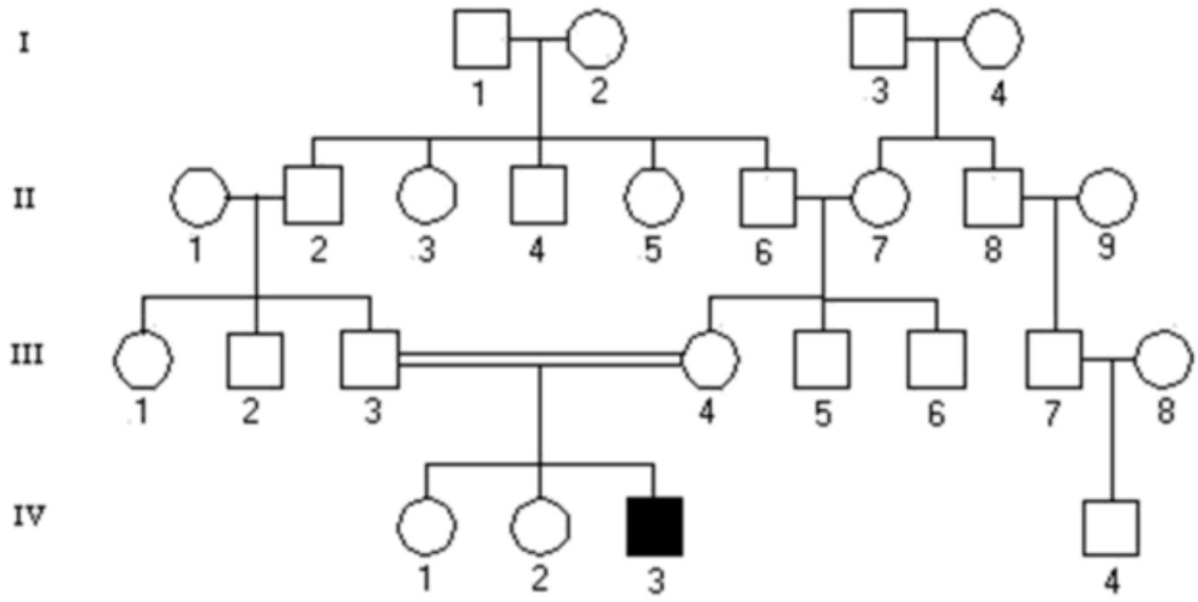
Generations are indicated with Roman numerals (I, II, III) and individuals within generations are marked with arabic numbers (1, 2, 3, 4).

Pedigree Questions: this pedigree doesn't indicate carriers

1. What sex is individual I-2?
2. How many children are in the 2nd generation from the union of I-1 and I-2?
3. What are their sexes?
4. Which individual was married in generation 2?
5. How many daughters are in generation 3?
6. How many sons are in generation 4?
7. List the 3 individuals who were afflicted with sickle cell anemia?
8. Were individuals I-1 and I-2 carriers of sickle cell?
9. How do you know? (Explain your answer to #8)
10. List another carrier of sickle cell anemia.



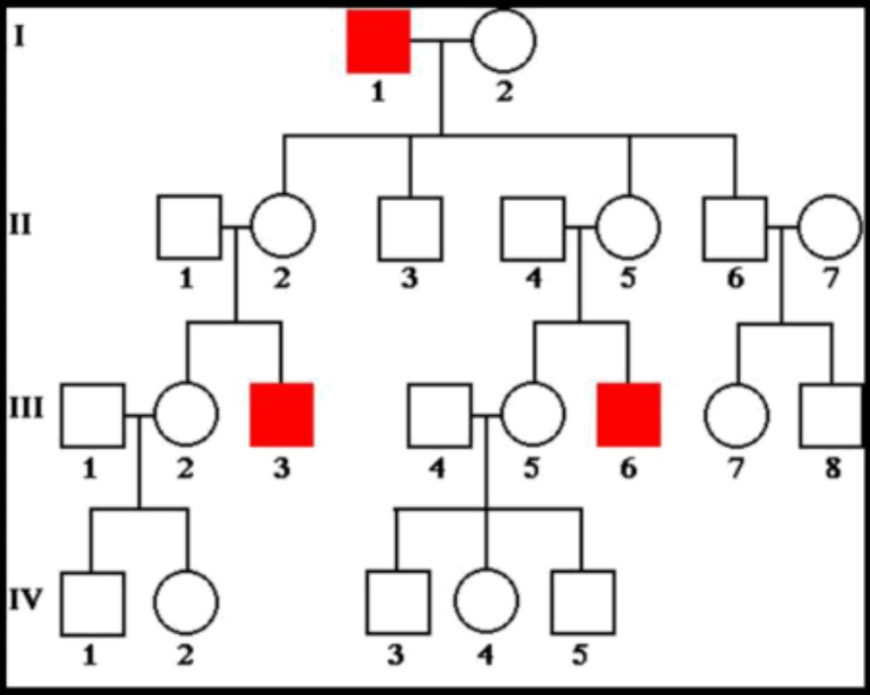
What type of Inheritance?
How many generations?
What is genotype of Individual?



What type of Inheritance?

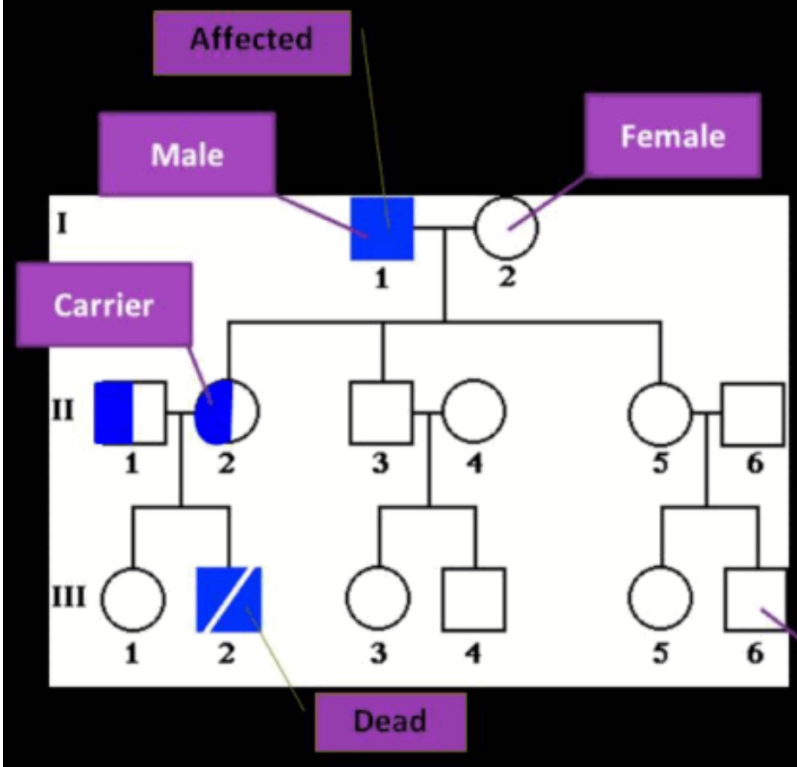
What is genotype of Individual I-1?

How many generations?



What is a pedigree? What does a pedigree look like?

- **Definition:** a graphic representation of genetic inheritance used by geneticists to map genetic traits



- Generations in separate rows indicated by Roman numerals (I, II, III…)
- Individuals within one generation indicated by Arabic numerals (1, 2, 3…)
- Parents connected by horizontal lines
- Offspring connected by vertical lines