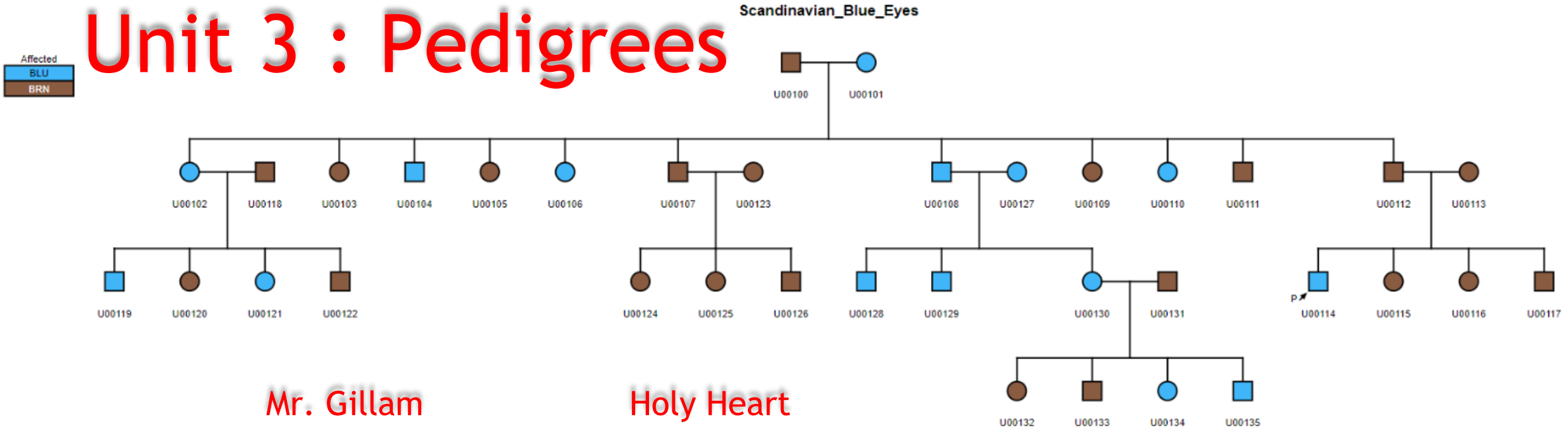
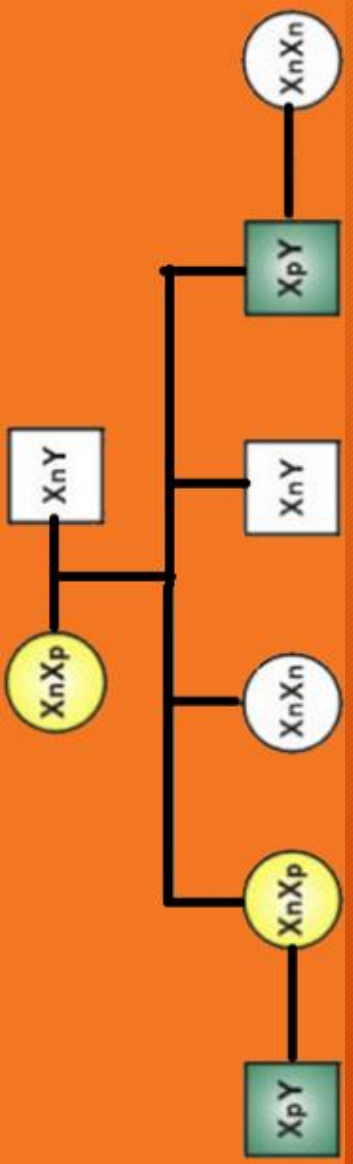
Unit 3 : Pedigrees



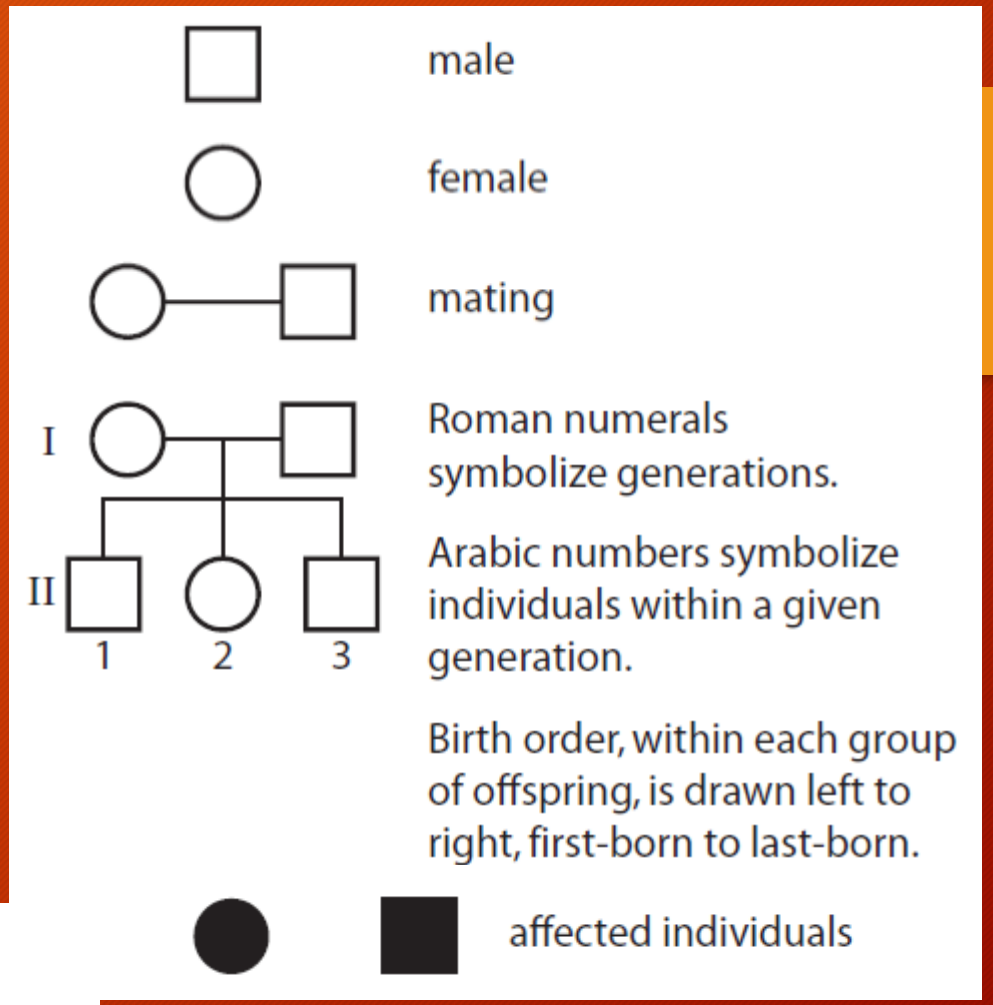
Mr. Gillam

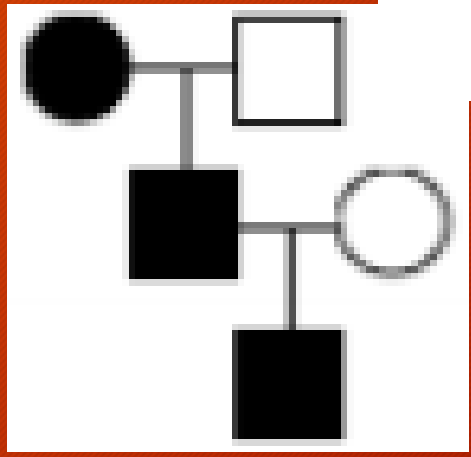
Holy Heart

What is a pedigree?



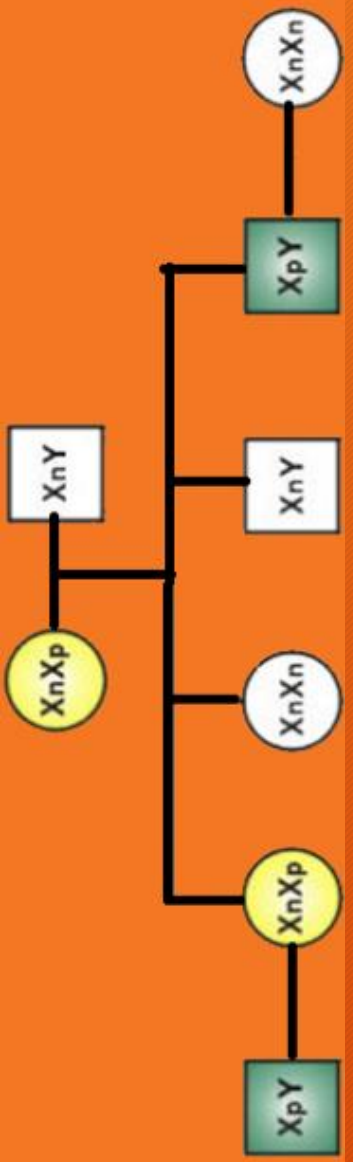
- **pedigree** flowchart that uses symbols to show inheritance of a trait within a biological family
- **Founding Parents** are the male and female at the very top of the pedigree.
- **Non-founding Parents:** one parent is from the original founders and the other is marrying into the family.





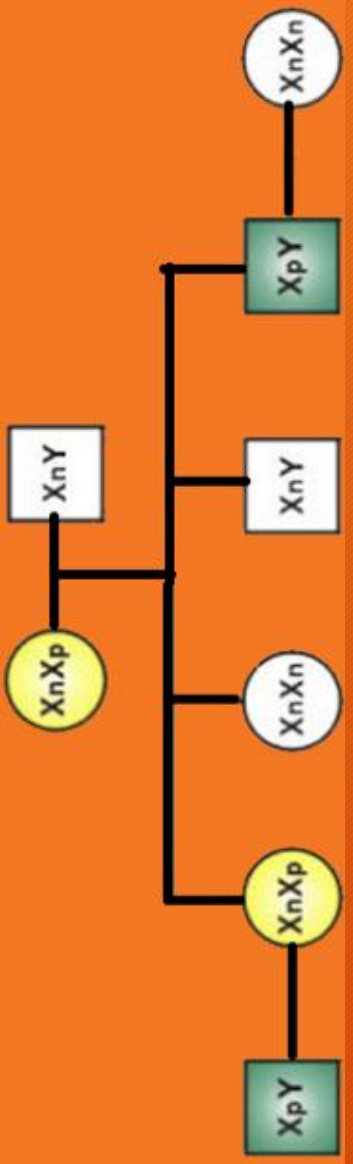
Non founding mother (married in)

Pedigrees Trace Inherited Genetic Disorders

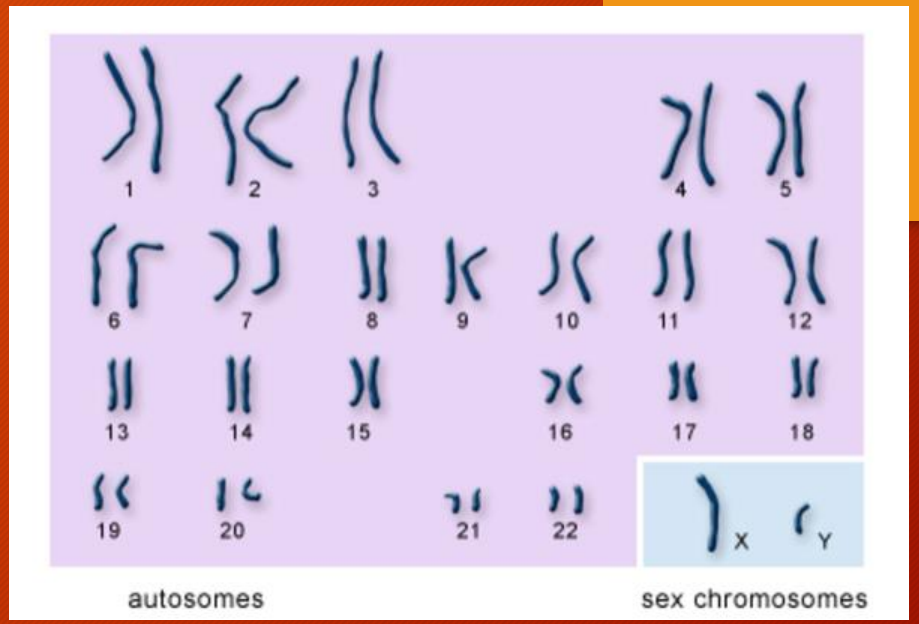


- A genetic disorder is an illness that is caused by changes to a person's genetic material. These changes can range from alterations of a single gene to changes to the structure and number of entire chromosomes.
- When geneticists want to learn about the inheritance of human traits, they collect as much information as possible about the history of a biological family and use this information to create a diagram called a pedigree.

Review of Terminology

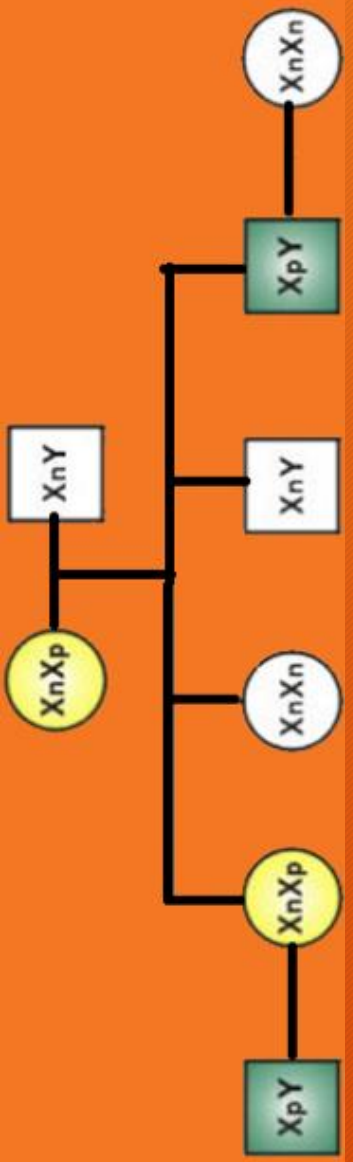


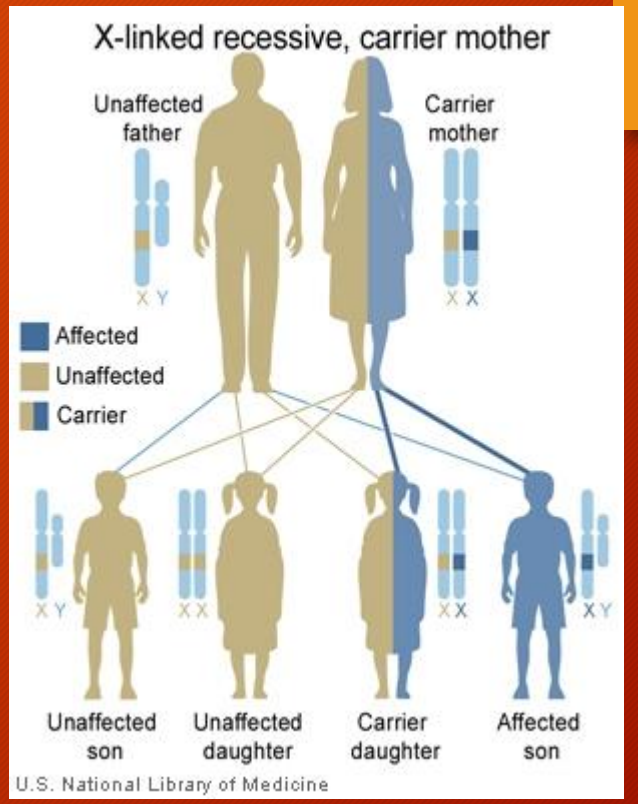
- **autosome** chromosome other than sex chromosome
- **sex chromosome** X or Y chromosome; determines genetic sex
- **Autosomal dominant** is a pattern of inheritance characteristic of some genetic diseases. Autosomal means that the gene in question is located on one of the numbered, or non-sex, chromosomes. Dominant means that a **single copy** of the disease-associated mutation is enough to cause the disease. i.e **Aa: the A carriers the disorder**
- **Autosomal recessive** is one of several ways that a trait, disorder, or disease can be passed down through families. An autosomal recessive disorder means **two copies** of an abnormal gene must be present in order for the disease or trait to develop.
- **a carriers the disorder**
- **i.e aa: has the disorder or Aa is a carrier of the disorder**

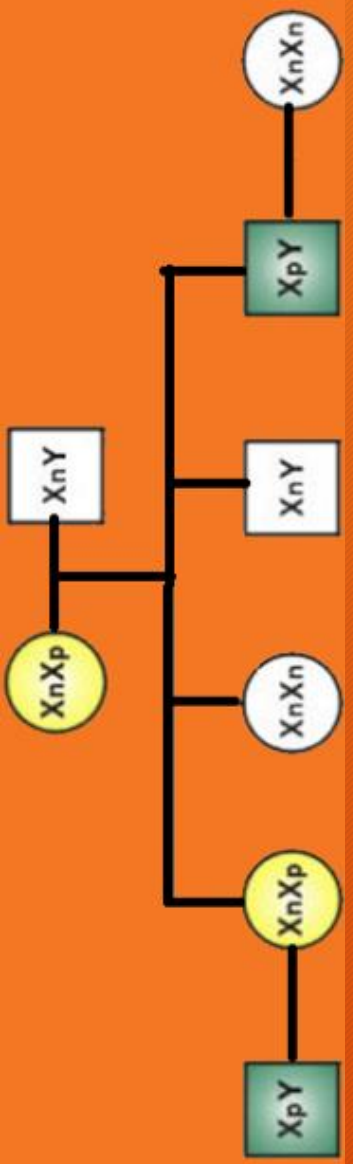


Review of Terminology

- **X-linked recessive** inheritance is a way a genetic trait or condition can be passed down from parent to child through mutations (changes) in a gene on the X chromosome.
- In **males** (who only have one X chromosome), a mutation in the copy of the gene on the **single X chromosome** causes the condition.
- **Females** would need **two copies** of the gene, one on each X chromosome.



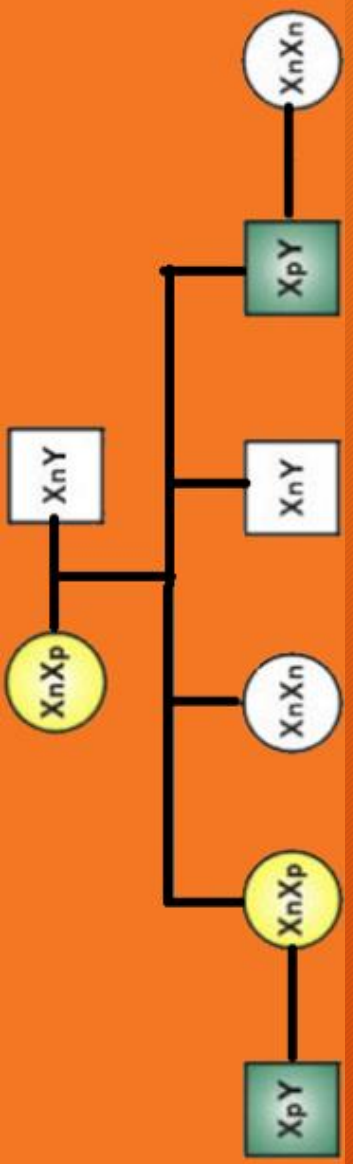




General Assumptions

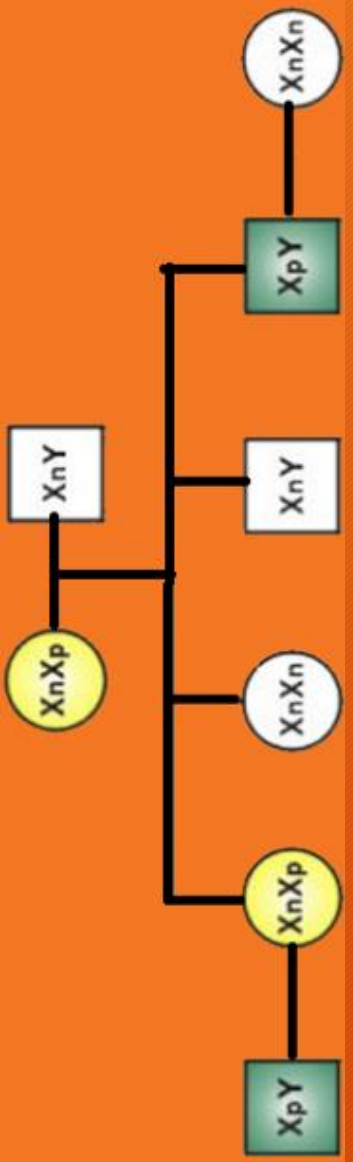
- In the problems that follow, you'll be reasoning about the mode of transmission of genetic traits that are controlled by **one gene**, with **two alleles**, a **dominant allele and a recessive allele.**
- We also make three simplifying assumptions:
- 1. An individual in the pedigree will have the disorder (**express the phenotype associated with a trait**) when the individual carries at least one dominant allele of a dominant trait, or two recessive alleles of a recessive a trait.
- 2. In each problem, the trait in question is rare in the general population. Assume for the purposes of these problems that **individuals who marry into the pedigree in the second and third generations are not carriers**. This does not apply to the founding parents – either or both of the individuals at the top of the pedigree could be carriers.
- 3. Not-Y-Linked. The causative genes in these problems may be autosomal or X-Linked, but are **not Y-linked**.

Analyzing a Human Pedigree

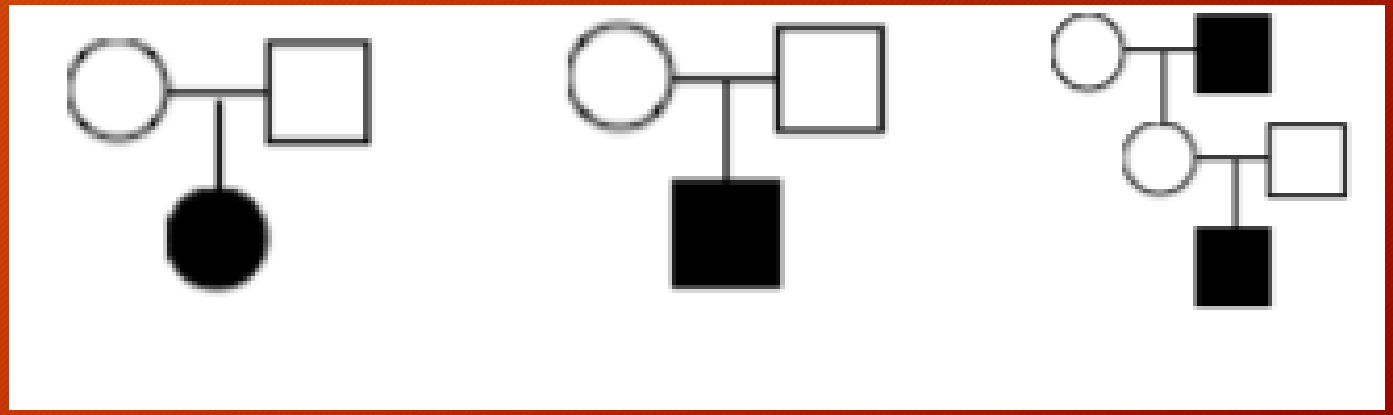


- **5 Key Clues**
- There are five things to remember in reasoning about pedigrees.
- (1) A unaffected female (white circle) or male (white square) individual cannot have any alleles of a dominant disorder. (because a single allele of a dominant trait causes an individual to be affected).
- (2) Individuals marrying into the family are assumed to not be carriers of recessive traits (because the trait is rare in the population)
- (3) An unaffected individual can be a carrier (have one allele) of a recessive trait. (because two alleles of a recessive trait are required for an individual to be affected)
- (4) When a trait is X-linked, a single recessive allele is sufficient for a male to be affected. (because the male only has one allele of an X-linked trait)
- (5) A father transmits his allele of X-linked genes to his daughters, but not his sons. A mother transmits an allele of X-linked genes to both her daughters and her sons.

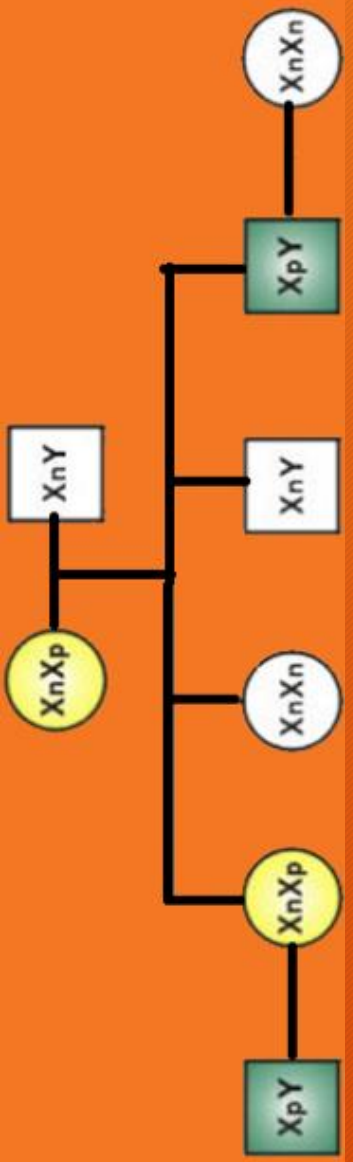
Patterns that Indicate A Recessive Trait



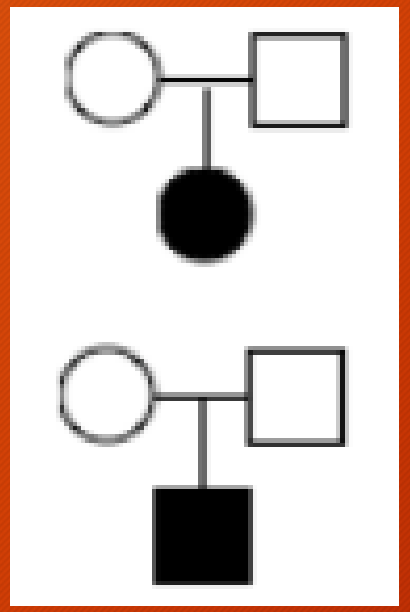
- **The disease must be recessive if any affected individual has 2 unaffected parents.**
- Since this is a genetic disease at least one parent must have an allele for the disease.
- If neither parent is affected, the trait cannot be dominant.

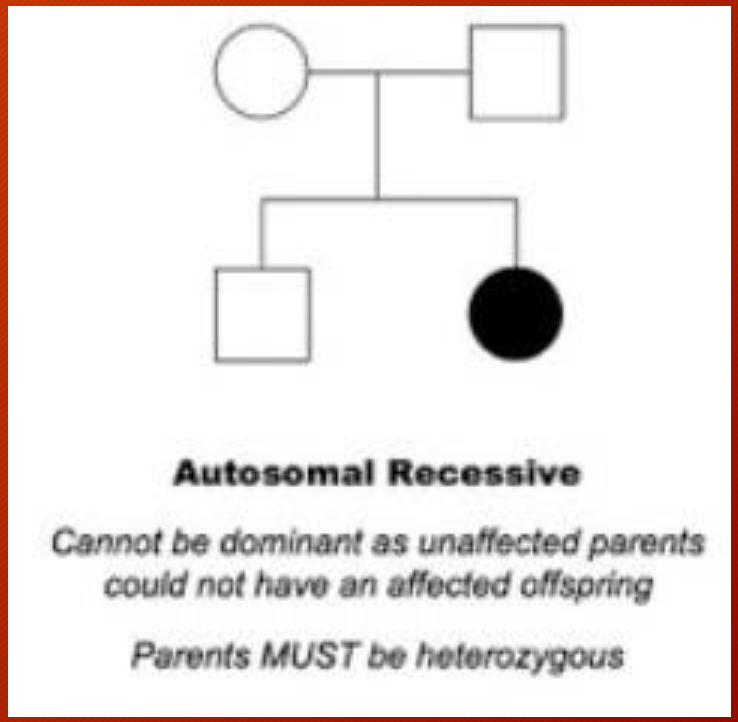


Patterns that Indicate An Autosomal Recessive Trait

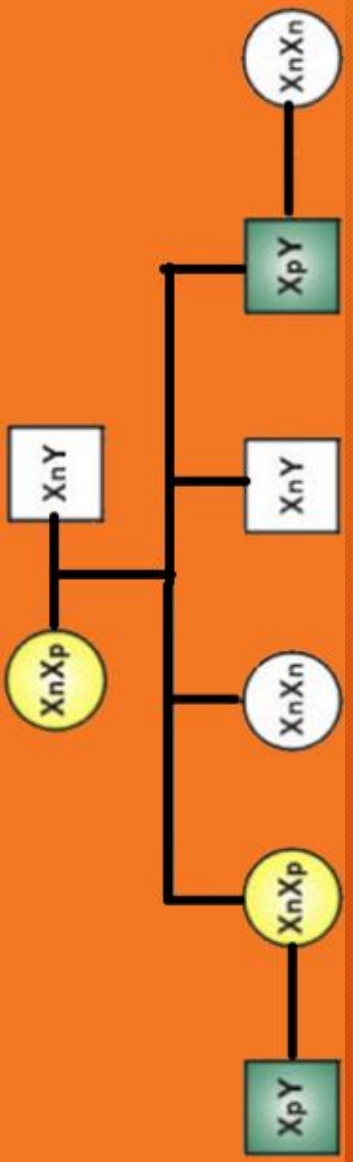


- If any affected daughter has 2 unaffected parents the disease must be autosomal recessive and not x linked.
- An affected individual must inherit a recessive allele from both parents, so both parents must have an allele.
- If the father had a recessive X-linked allele, he would have to be affected (since he only has one X-linked allele).

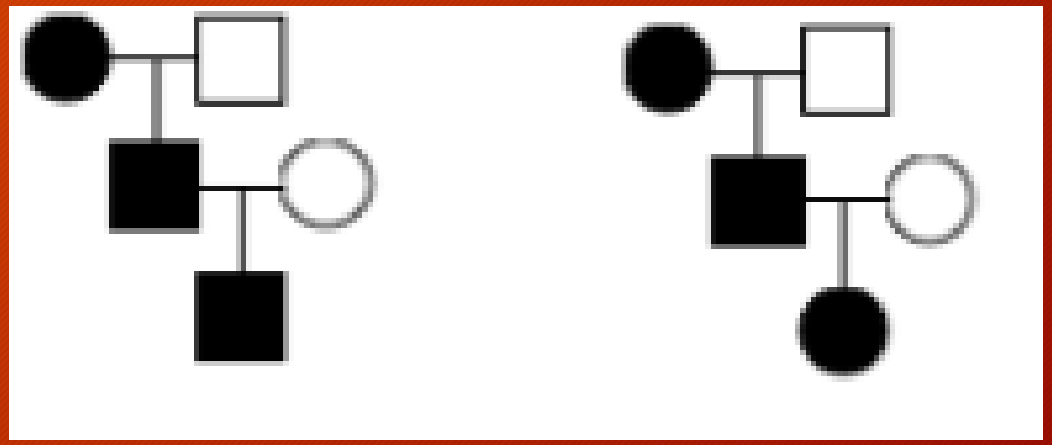




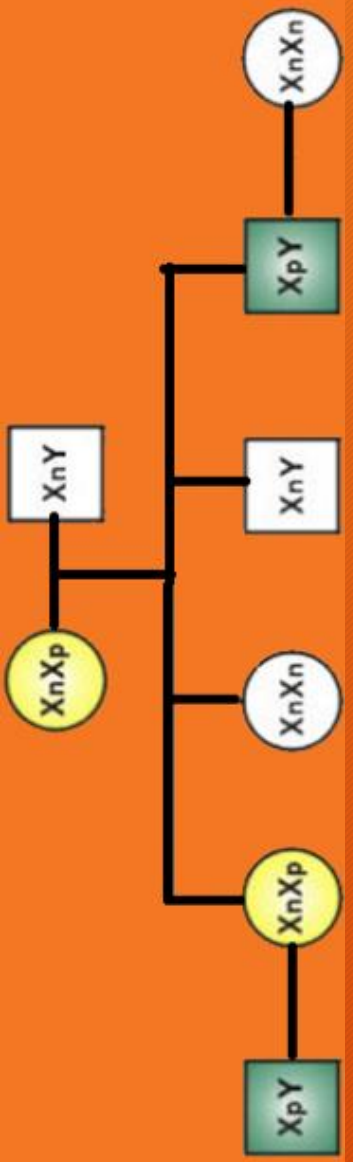
Patterns that Indicate a Dominant Trait



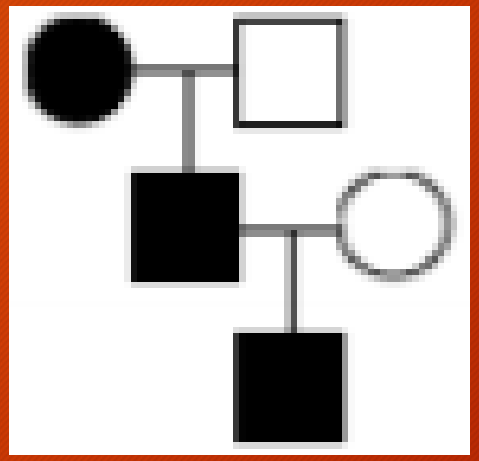
- **The disease must be dominant if every affected child of founding parents has an affected parent.**
- The unaffected mother, who is marrying in, does not carry an allele for the disease; so the affected child inherits an allele only from the affected father.
- No child could be affected by a single autosomal recessive allele, or X-linked recessive allele, so the trait is dominant.

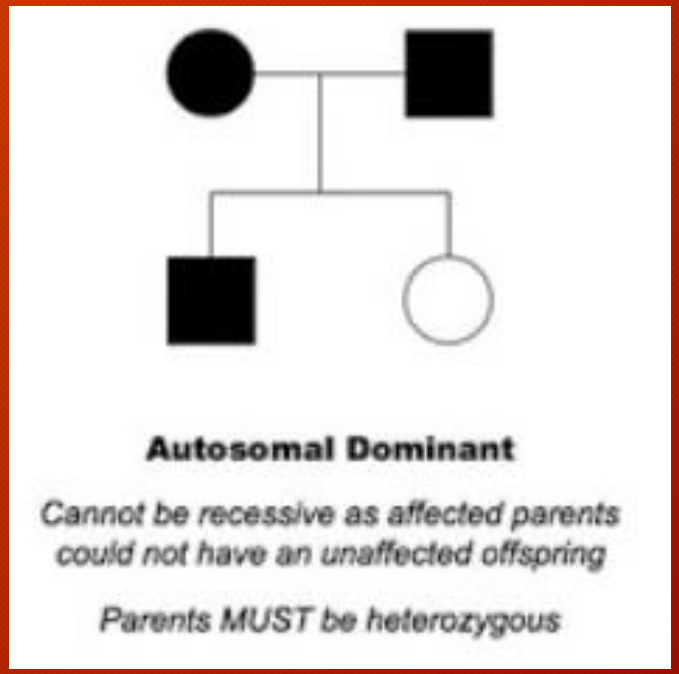


Patterns that Indicate an Autosomal Dominant Trait

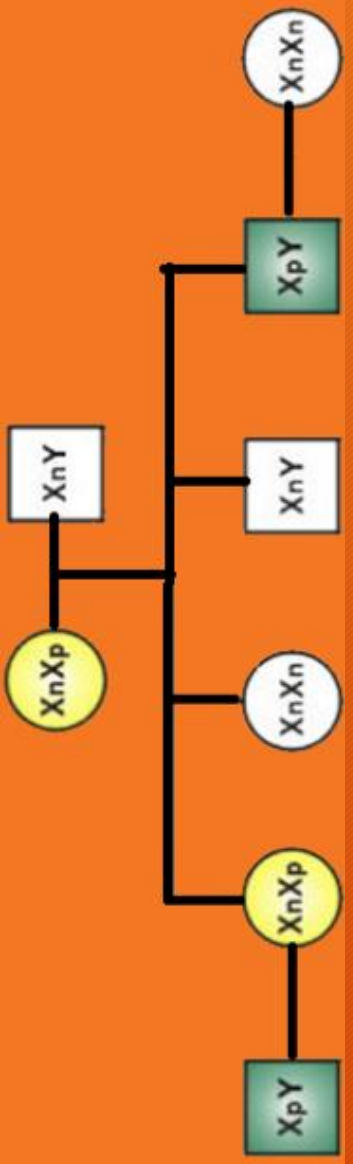


- When an affected son of non-founding parents has an affected father the disease must be autosomal dominant.
- A father does not transmit X-linked alleles to a son, so the disease cannot be X-linked dominant.





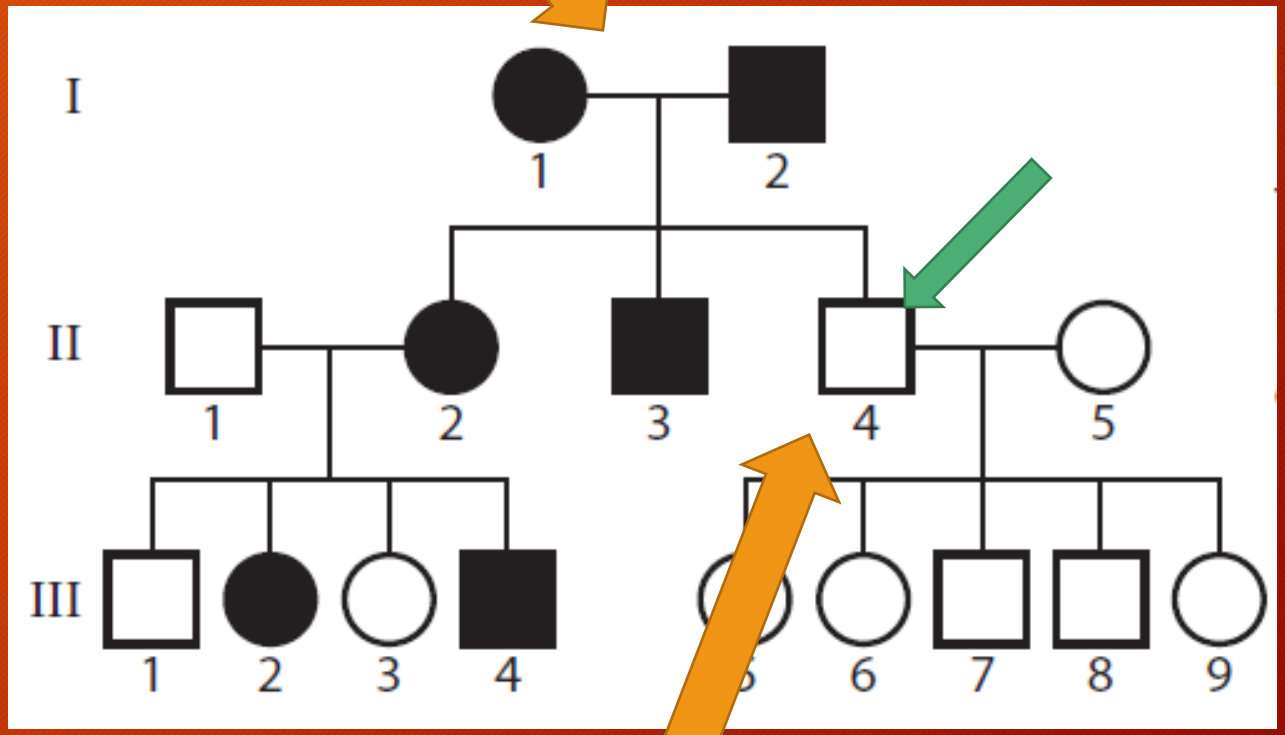
Lets try some



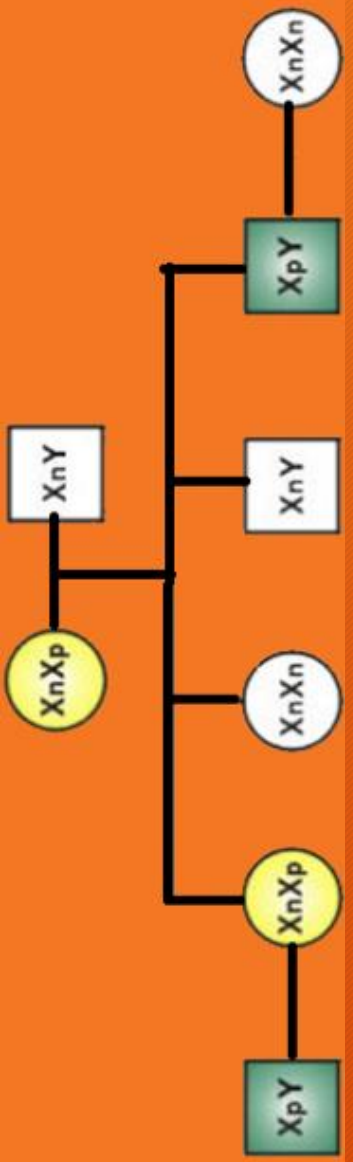
- What type of inheritance pattern is shown by this pedigree?

- It is autosomal dominant

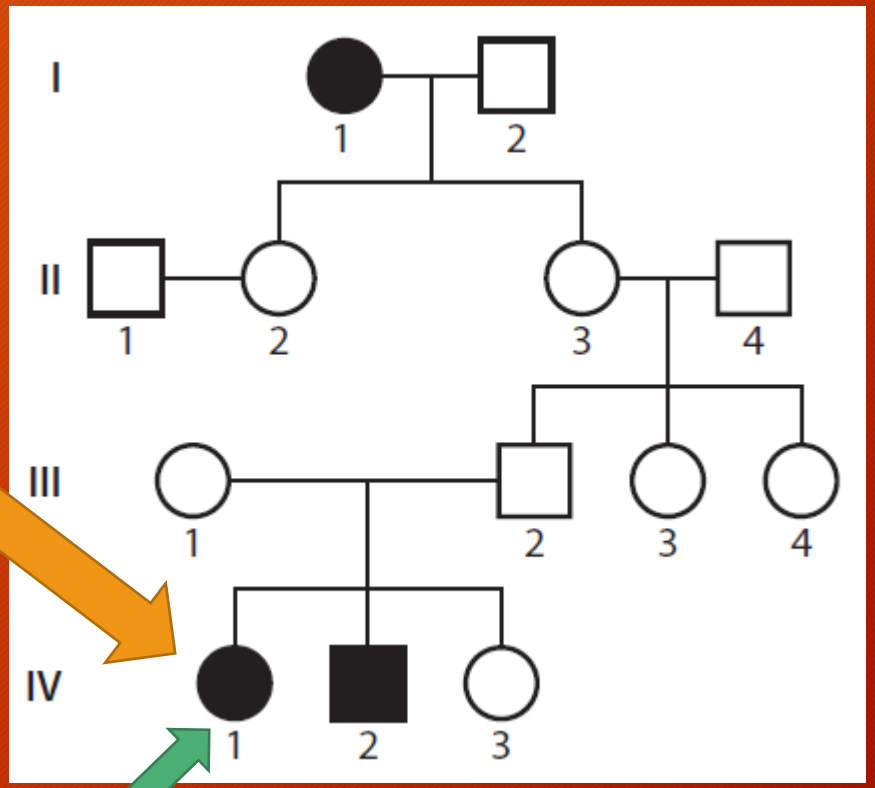
- Both parents have it but one child does not so it cannot be recessive.



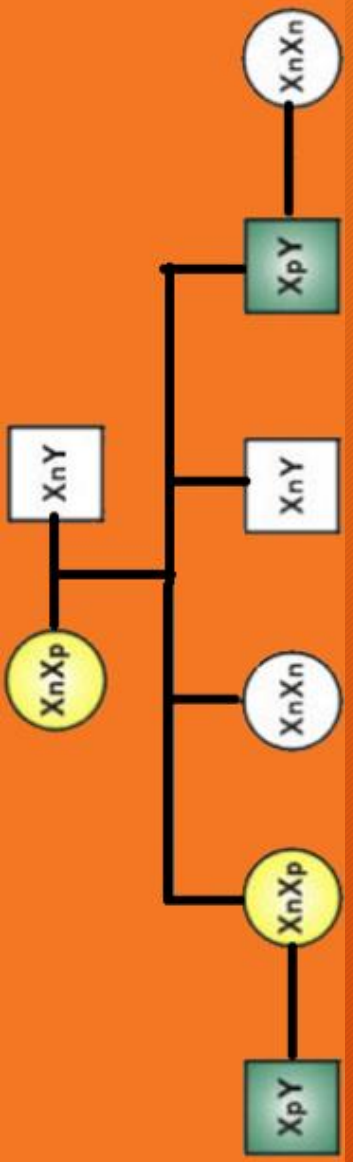
- This male does not have it and his X chromosome had to come from his mother so it cannot be x linked.



- What type of inheritance pattern is shown by this pedigree?

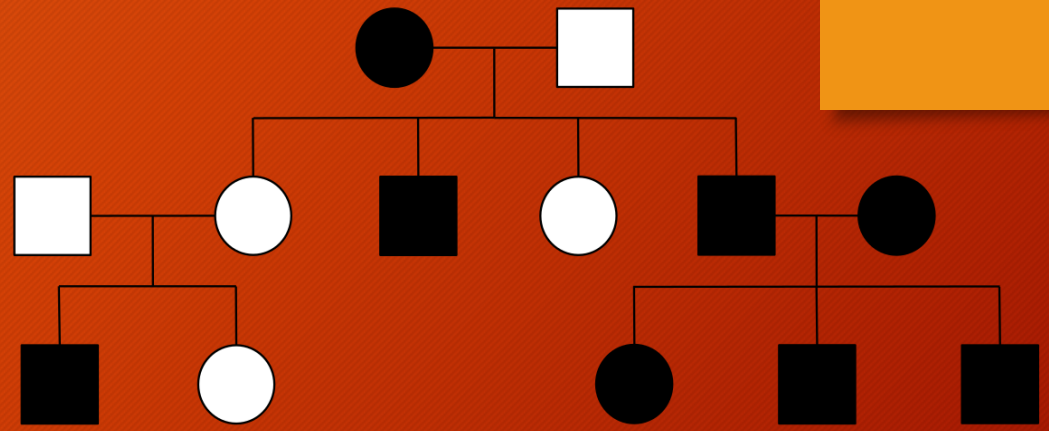


- Daughter and son have it but neither parent does so it has to be recessive
- It is autosomal recessive
- This female has the disorder and neither her father or mother has it, if it was X linked her father would have to have it, so it cannot be X linked

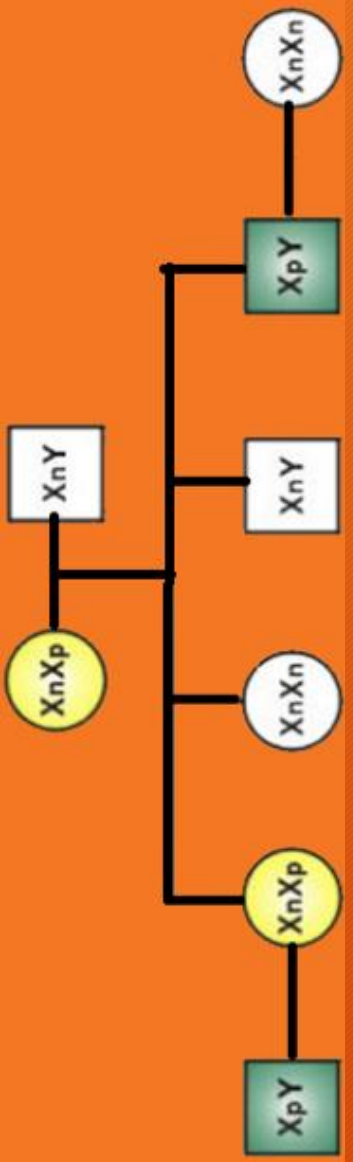


- What type of inheritance pattern is shown by this pedigree?

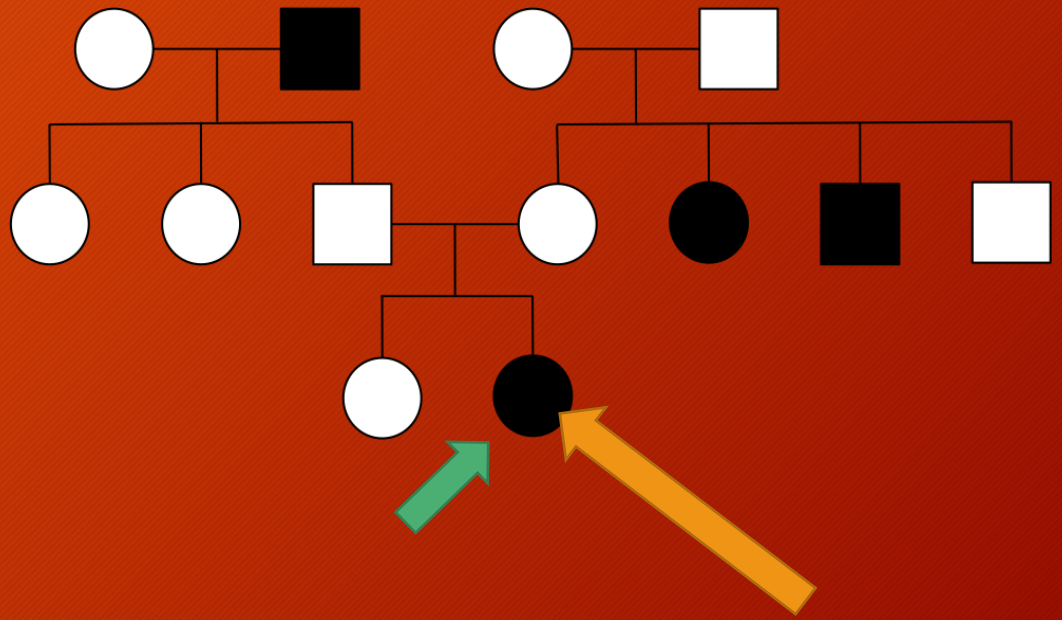
- It is sex linked recessive



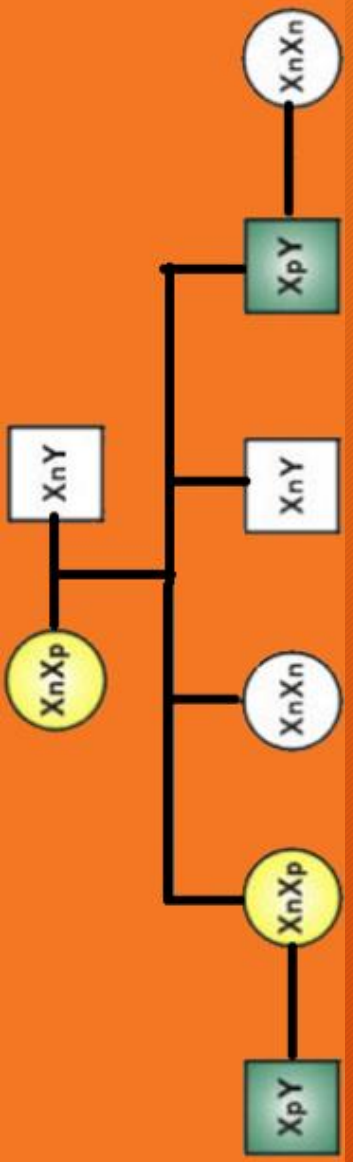
- Son has it but neither parent does and the father is marrying from outside so cannot be a carrier. This means its X linked



- What type of inheritance pattern is shown by this pedigree?

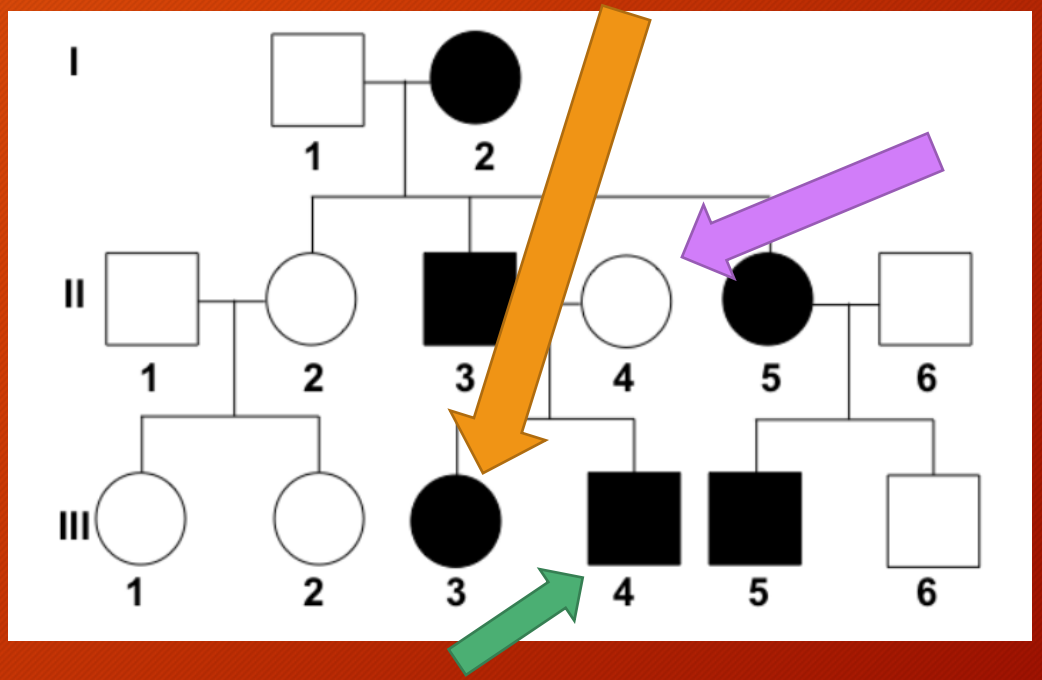


- It is autosomal recessive
- This female has the disorder and her father does not so it cannot be x linked
- Daughter has it but neither parent does so it has to be recessive



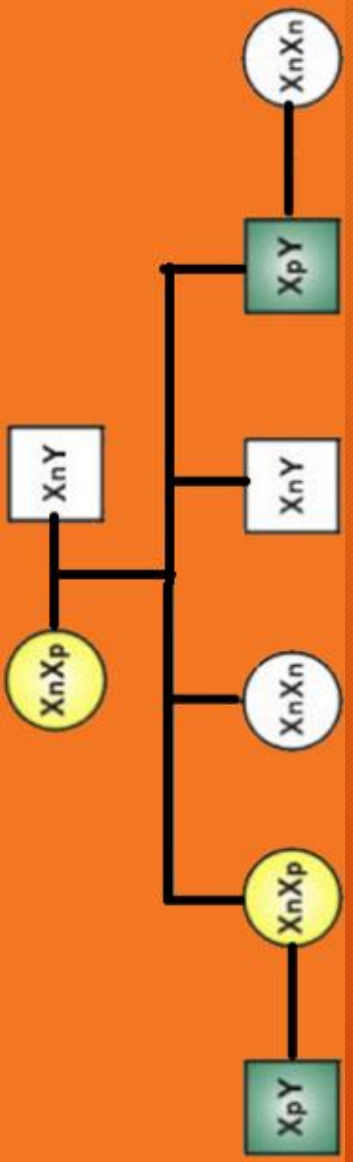
- What type of inheritance pattern is shown by this pedigree?
- Dominant traits will never skip a generation
- It is autosomal dominant

- Cannot be X linked because this female has it and her mother cant be a carrier because she is marrying into the family



- An affected son who has a father with the disorder and the mother once again cannot be a carrier means that it is dominant

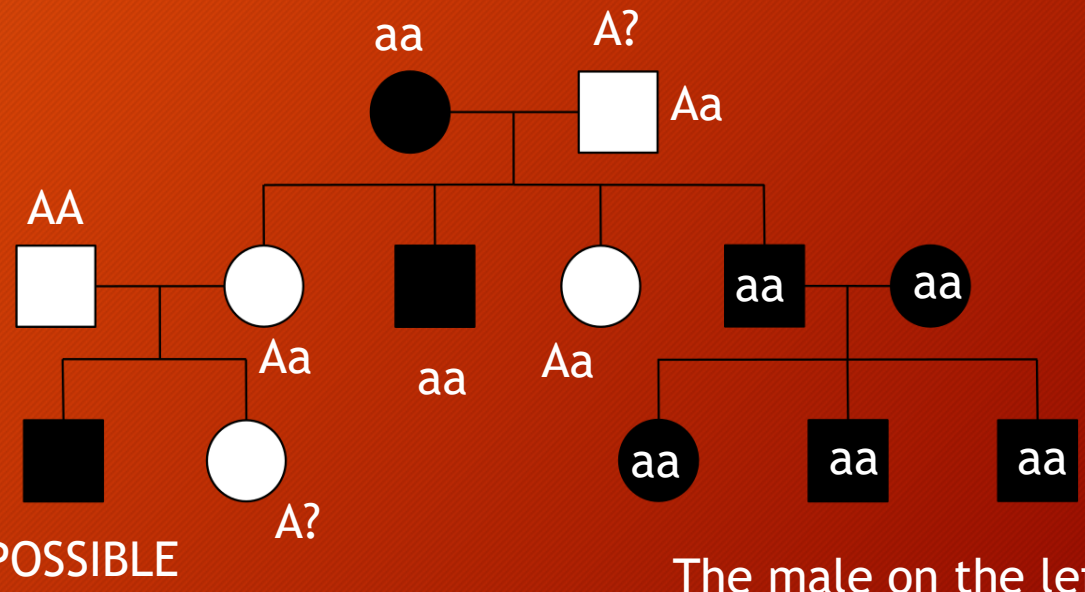
All else fails try them by writing possibilities on them.



We are going to trial and error autosomal recessive – this is the one from earlier that was sex linked recessive

Since the female is aa the father has to be Aa

If possible start with the parents or fill in the ones you know. In this case we will start with the parents One has to be aa and the other has to be A? because it does not have the disorder



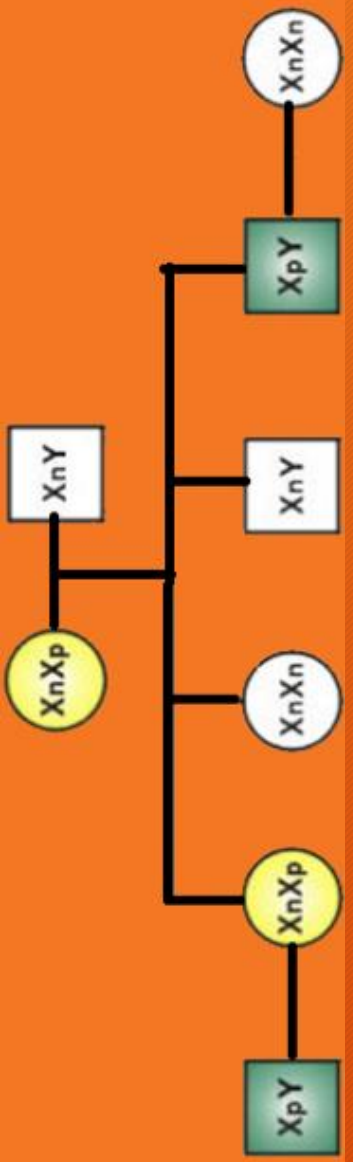
IMPOSSIBLE

It cant be autosomal recessive!

Now we work downwards from there.

The male on the left has to be AA because he is marrying in so cannot be a carrier

All else fails try them by writing possibilities on them.



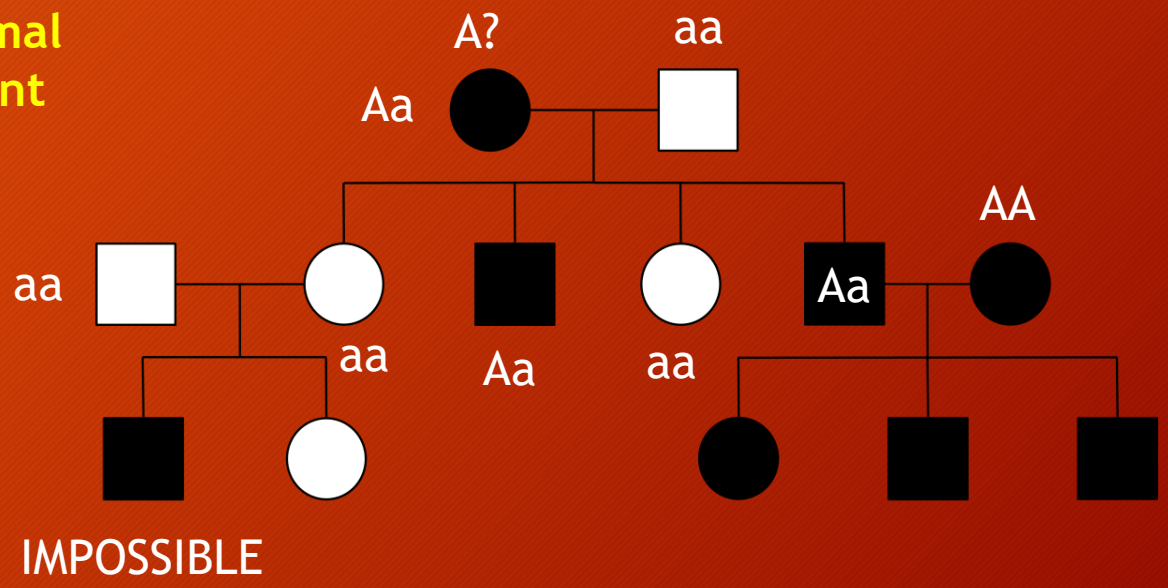
In this case
A = disorder
A = no disorder
If possible start with the parents or fill in the ones you know. In this case we will start with the parents but we are unsure of one. One has to be aa and the other has to be A? because it has the disorder

Now we work downwards from there.

We are going to trial and error autosomal dominant – this is the one from earlier that was sex linked recessive

It cant be autosomal dominant

After completing their first child we know that the male parent has to be Aa. So we write that in

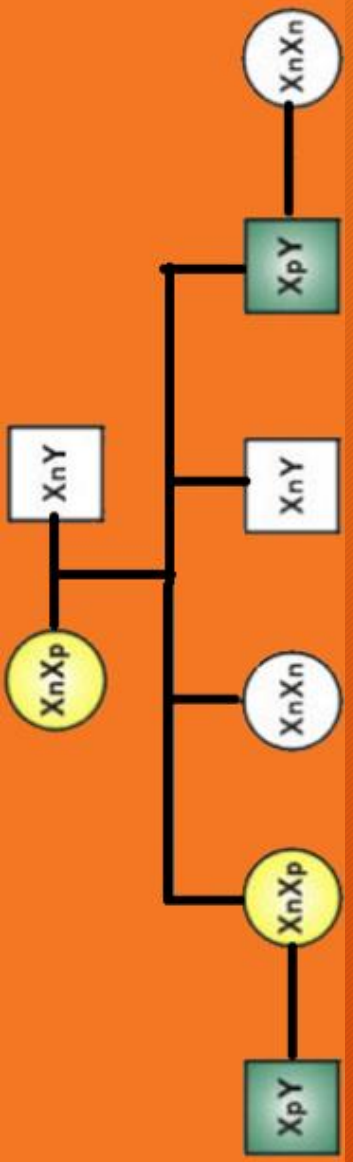


IMPOSSIBLE

This next female has to be AA because the rules of pedigrees say they are not allowed to be carriers. Same for the female on the left has to be aa

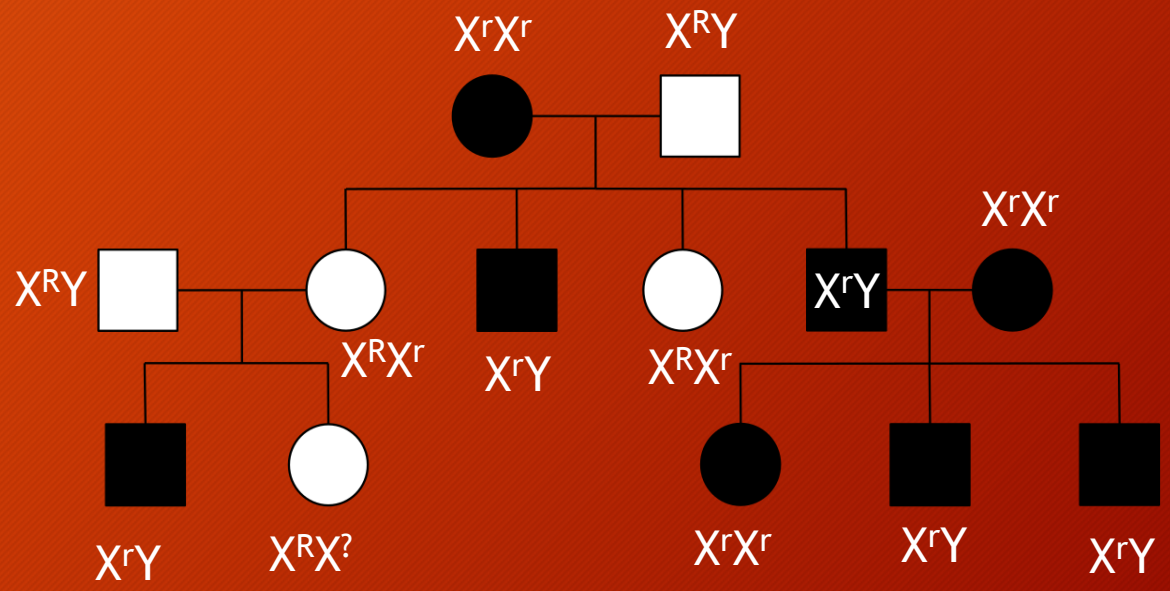
All else fails try them by writing possibilities on them.

We are going to trial and error sex linked – this is the one from earlier that was sex linked recessive



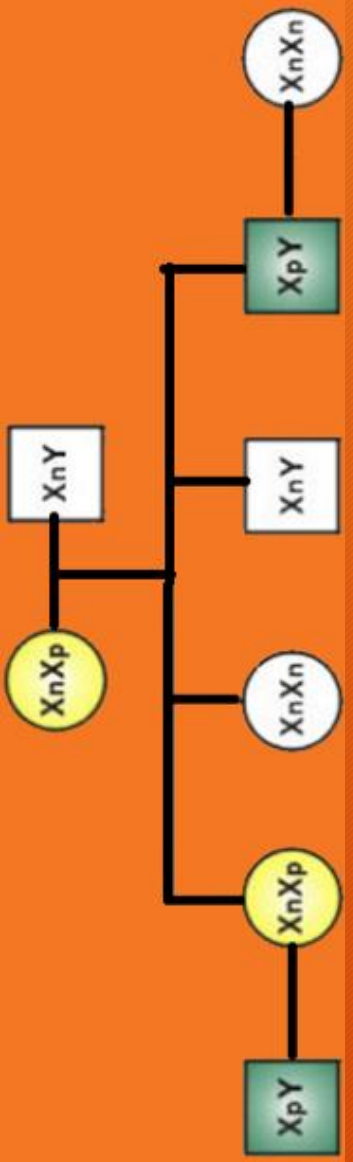
In this case
X^R = normal
X^r = disorder
Y = normal male
If possible start with the parents or fill in the ones you know. In this case we will start with the parents but we are unsure of the female.
The male has to be X^RY because he does not have the disorder and the female is X^rX^r because she has the disorder.

Now we work downwards from there.

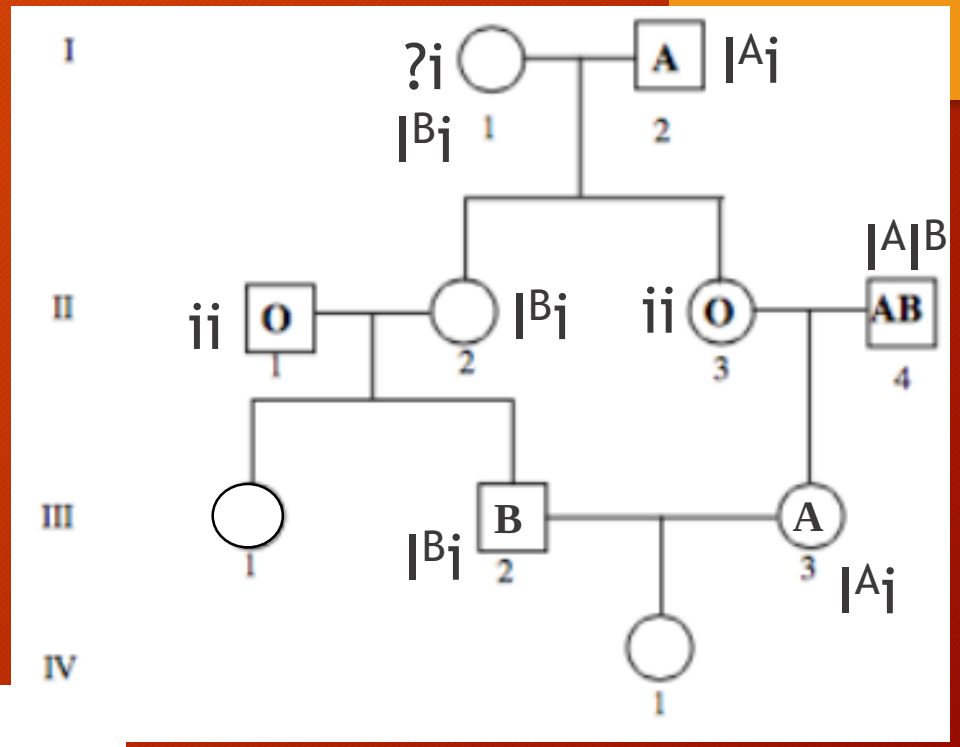


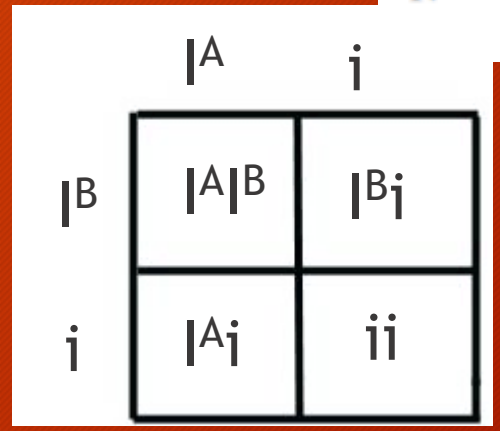
Everything worked! It is sex linked recessive

Blood Types can also be shown in pedigrees. In order to do blood types you will have to work them out.



- What is the probability that a IV-1 will have blood type AB?
- As you can see we do not know the parents, so the best way to start is with O blood type that has to be ii and AB that has to be I^AI^B
- Now know that both of the parents had at least one i.
- Now we look at the B. It has to be I^Bi because of the O above it. Which means that person II-2 has to be I^Bi so I-1 has to be I^Bi as well
- Now female III-3 has to be I^Ai and we can complete a Punnett square.

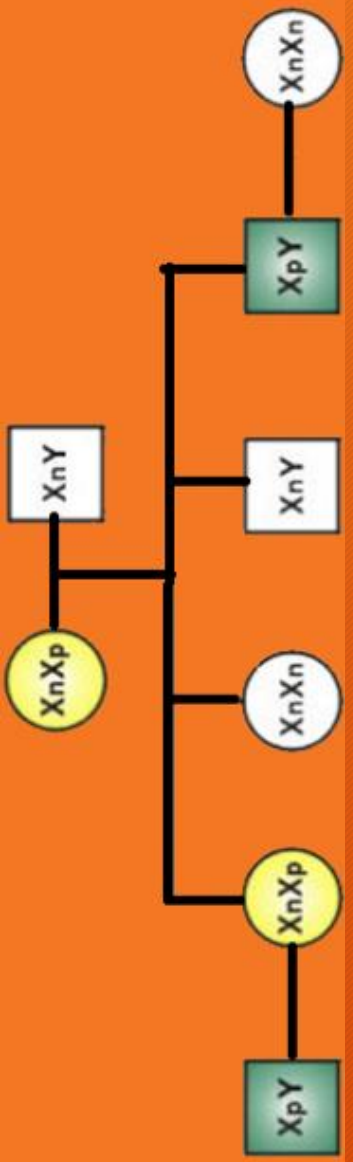




- There is a 25% chance that person IV-1 will have AB blood type

Activity 14.5

- Analyzing Pedigrees



Exit Card #7