Instructions

- To help you navigate these slides, you should set your viewer to display thumbnails of these slides. On many viewers, this can be done by pressing the F4 key.

- Viewing will be easier if you maximize the window.

- The slides should be viewed in order because a slide may provide information that is needed to understand the next or subsequent slides. It is important that you understand each slide before advancing to the next because concepts build upon previous concepts.

- This slide presentation is intended to be a lecture. It might be helpful to take notes as you progress through the slides.

Autosomal Traits

- The words "autosomal dominant" refer to dominant genes located on the autosomes (chromosomes that are not the sex chromosomes). Similarly, autosomal recessive genes are recessive genes located on the autosomes.

- Upper case letters are used to represent autosomal dominant genes and lower case letters are used to represent autosomal recessive genes as shown below.

A = dominant
a = recessive

Genotype	Phenotype
AA	dominant
Aa	dominant
aa	recessive

Notice from the table above that AA individuals and Aa individuals will both have the dominant phenotype but aa individuals will have the recessive phenotype.

X-linked Recessive

- X-linked recessive genes are recessive genes found on the X chromosome.

- Upper case and lower case superscripts are used to represent dominant and recessive alleles as shown below.

X^A = dominant
X^a = recessive

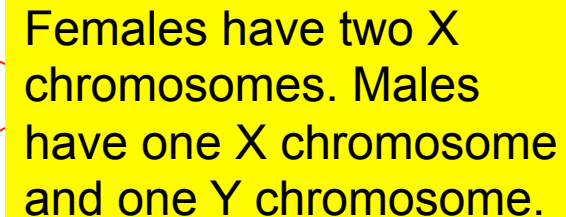<u>Females</u>
$X^A X^A$ – will have the dominant phenotype
$X^A X^a$ – will have the dominant phenotype
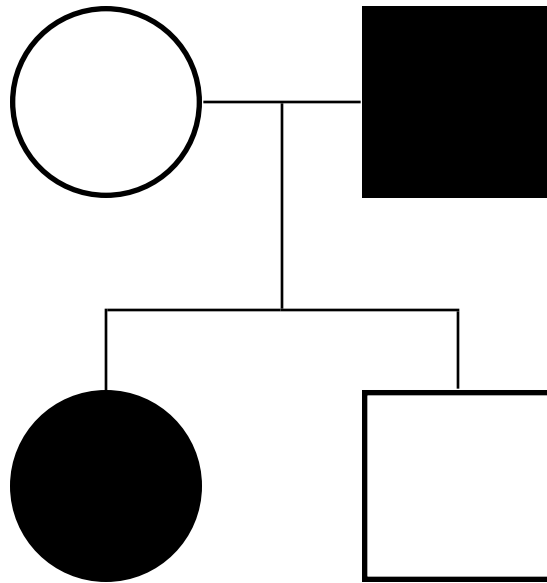$X^a X^a$ – will have the recessive phenotype

<u>Males</u>
$X^A Y$ – will have the dominant phenotype
$X^a Y$ – will have the recessive phenotype

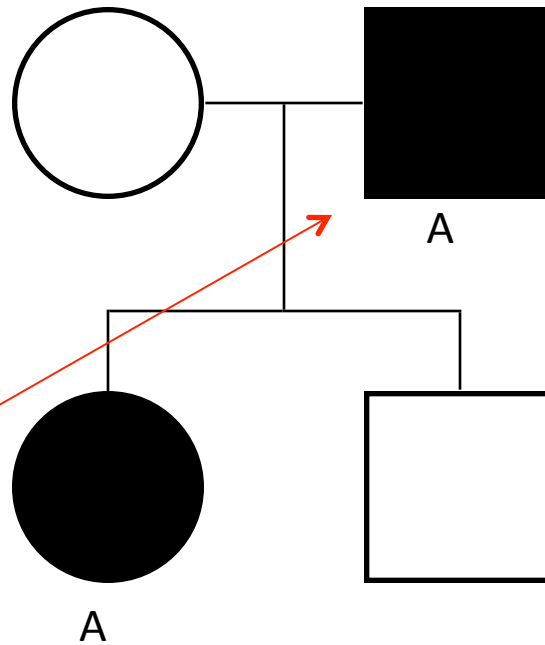Females have two X chromosomes. Males have one X chromosome and one Y chromosome.

Example 1 - Can this pedigree be for a trait that is autosomal dominant?

The answer to this question is on the next several slides.

Example 1 - Can this pedigree be for a trait that is autosomal dominant?
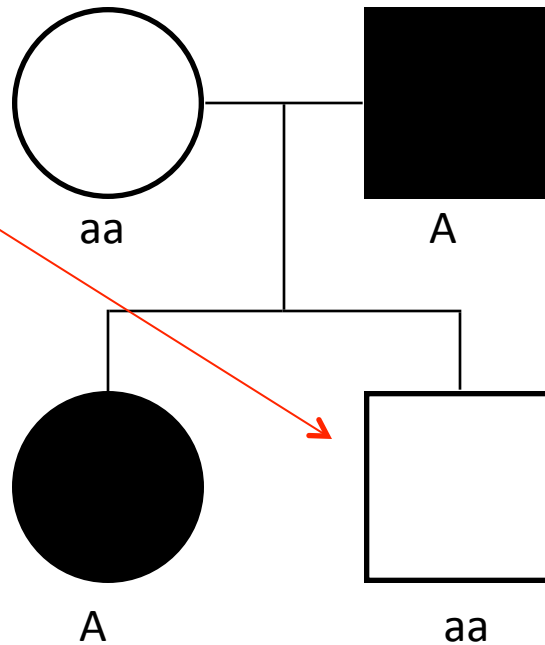
A = dominant
a = recessive

If the trait is dominant, then these individuals must have at least one "A" allele.

A

A

Example 1 - Can this pedigree be for a trait that is autosomal dominant?
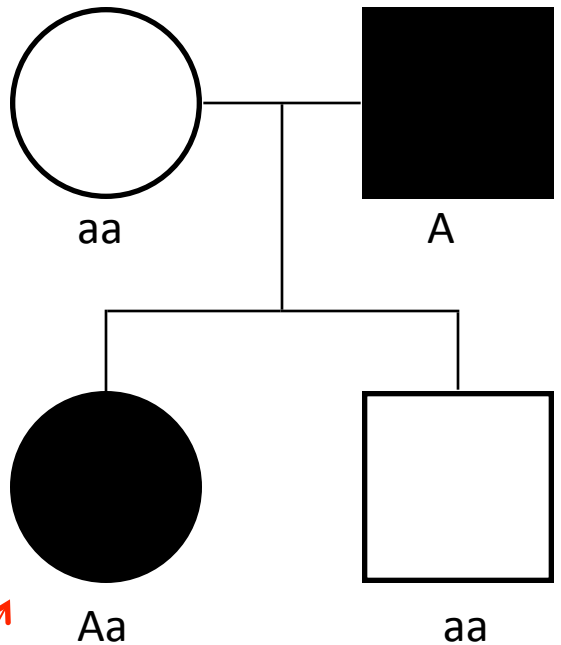
A = dominant
a = recessive

These individuals have the recessive phenotype. Therefore, their genotype is "aa."

aa

A

A

aa

Example 1 - Can this pedigree be for a trait that is autosomal dominant?
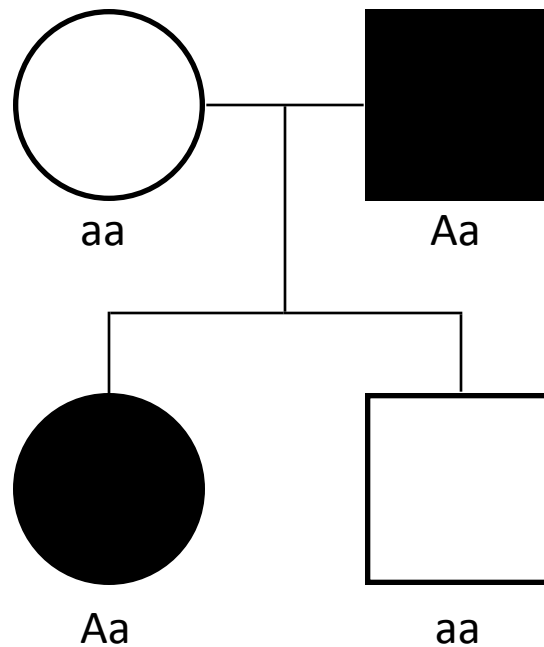
A = dominant
a = recessive

This individual can only produce "a" gametes. Therefore all of her offspring must have an "a."

aa   A

Aa   aa

This individual must have an "a" allele from her mother.

Example 1 - Can this pedigree be for a trait that is autosomal dominant?

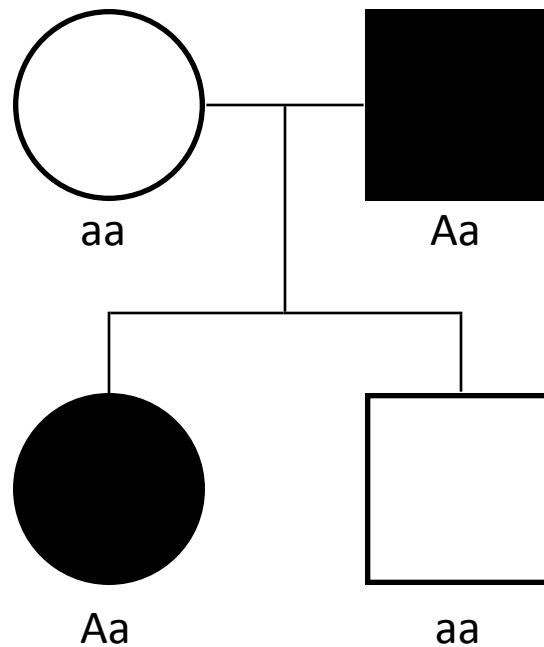A = dominant
a = recessive

aa

Aa

Aa

aa

This individual is "aa." He received one "a" from each parent. Therefore, his father must have an "a" allele.

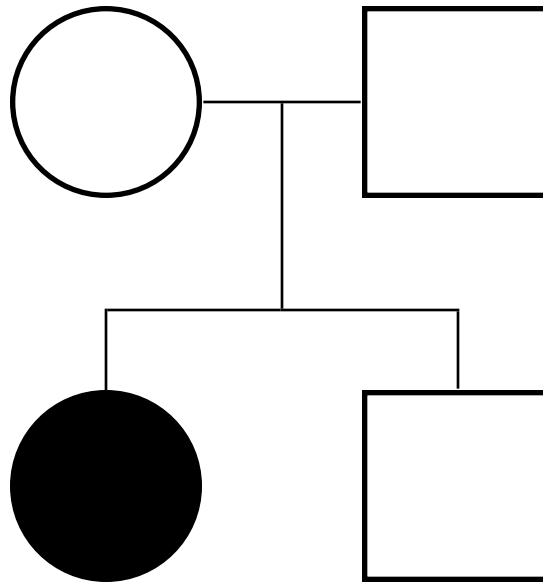# Example 1 - Can this pedigree be for a trait that is autosomal dominant?

A = dominant
a = recessive

This pedigree can therefore be for a trait that is autosomal dominant.

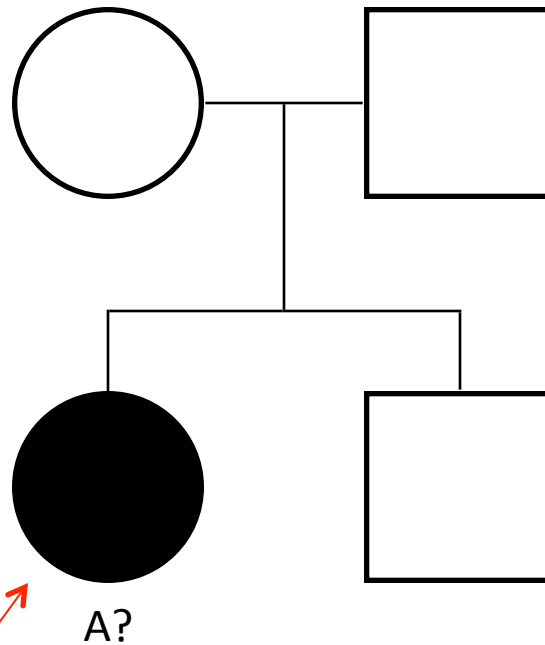# Example 2 - Can this pedigree be for a trait that is autosomal dominant?

A = dominant
a = recessive

The answer to this question is on the next several slides.

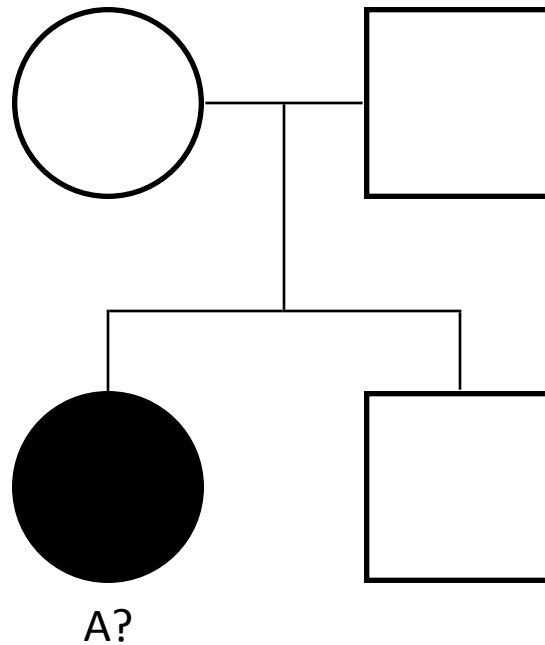# Example 2 - Can this pedigree be for a trait that is autosomal dominant?

A = dominant
a = recessive

A?

If the trait is dominant, then this individual must have at least one "A" allele.

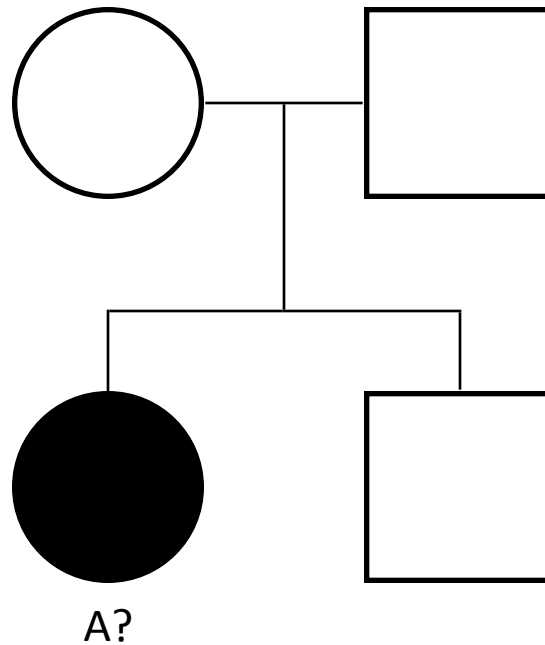# Example 2 - Can this pedigree be for a trait that is autosomal dominant?

A = dominant
a = recessive

The "A" allele must have come from one of her parents. If one of the parents has an "A" allele, then the parent must have the trait. The pedigree shows that they do not have the trait, therefore, this pedigree is not possible; the trait cannot be autosomal dominant.

A?

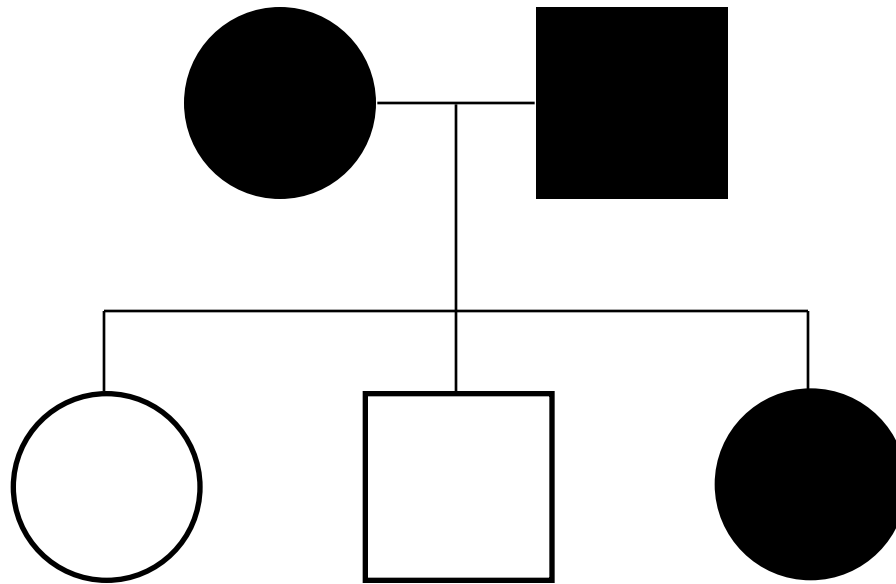# Example 2 - Can this pedigree be for a trait that is autosomal dominant?

A = dominant
a = recessive

A?

From example 2, we can conclude that if a person has a dominant trait, at least one of the parents must have the trait.

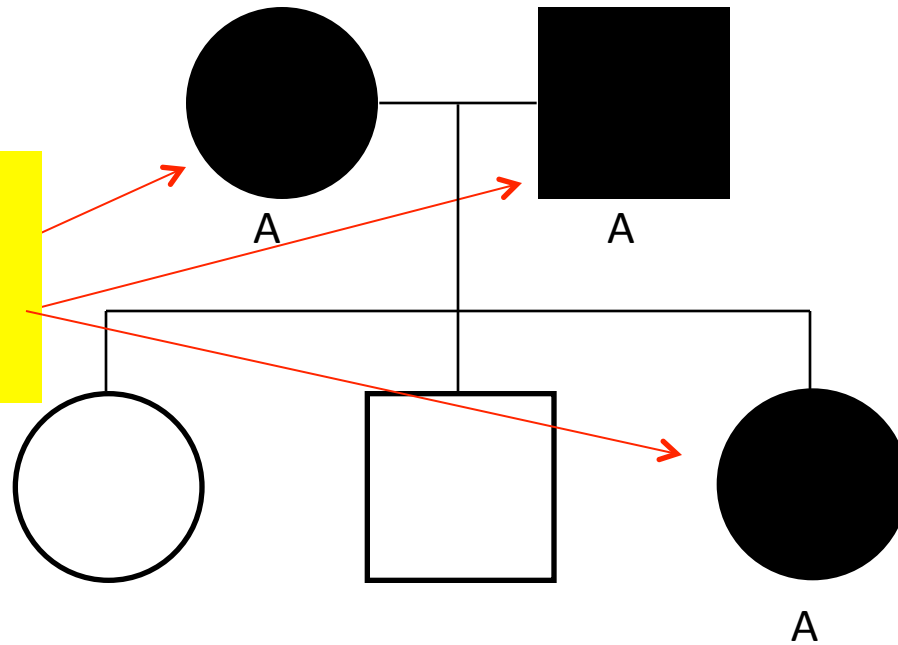# Example 3 - Can this pedigree be for a trait that is autosomal dominant?

A = dominant
a = recessive

Example 3 - Can this pedigree be for a trait that is autosomal dominant?
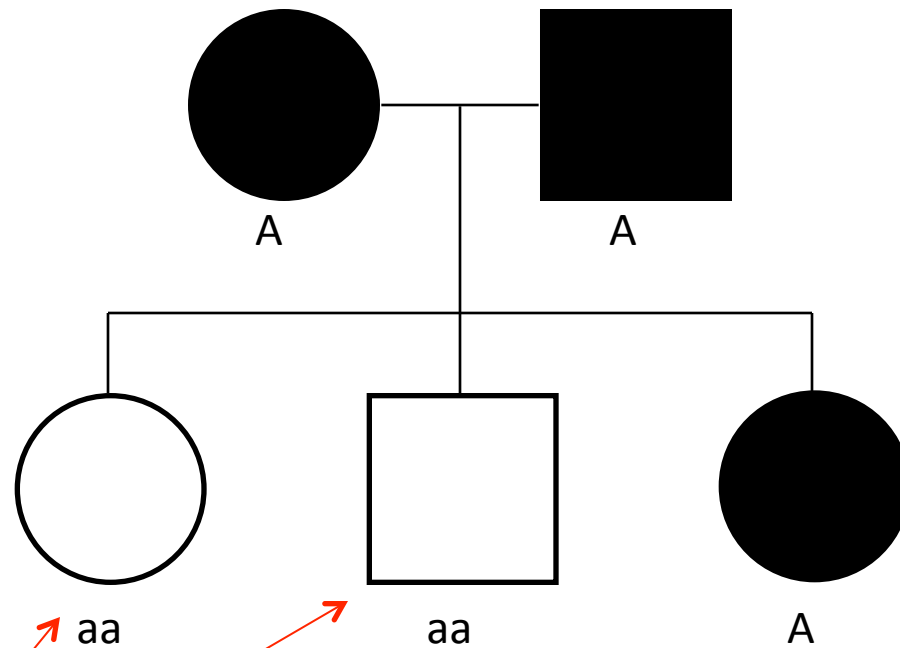
A = dominant
a = recessive

If the trait is dominant, then these individuals must have at least one "A" allele.

A
A
A

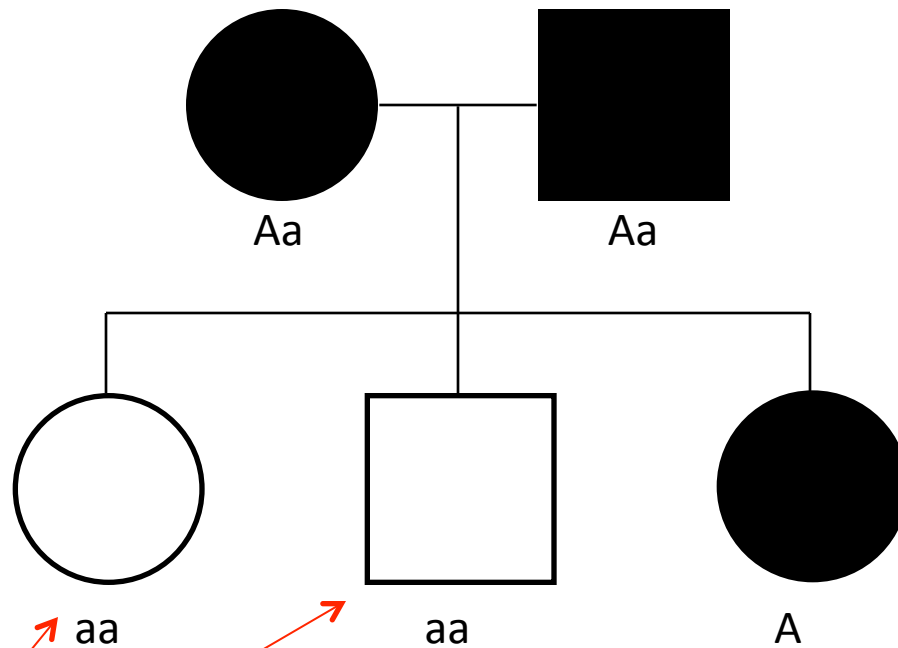# Example 3 - Can this pedigree be for a trait that is autosomal dominant?

A = dominant
a = recessive

aa

aa

A

These individuals have the recessive phenotype so they must be "aa."

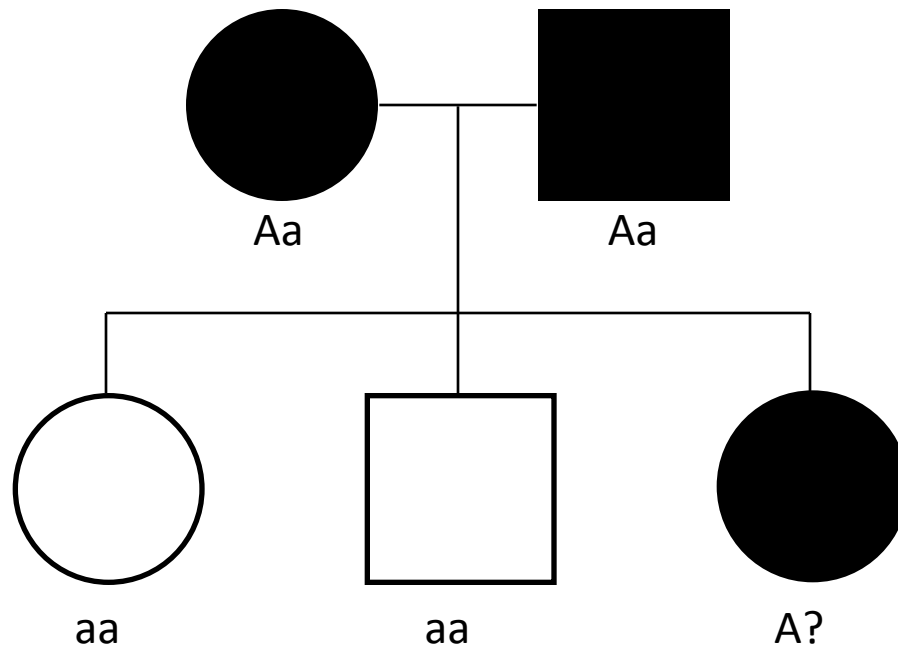# Example 3 - Can this pedigree be for a trait that is autosomal dominant?

A = dominant
a = recessive

Aa   Aa

aa   aa   A

They each received one "a" from each parent. Therefore, their parents must each have an "a" allele.

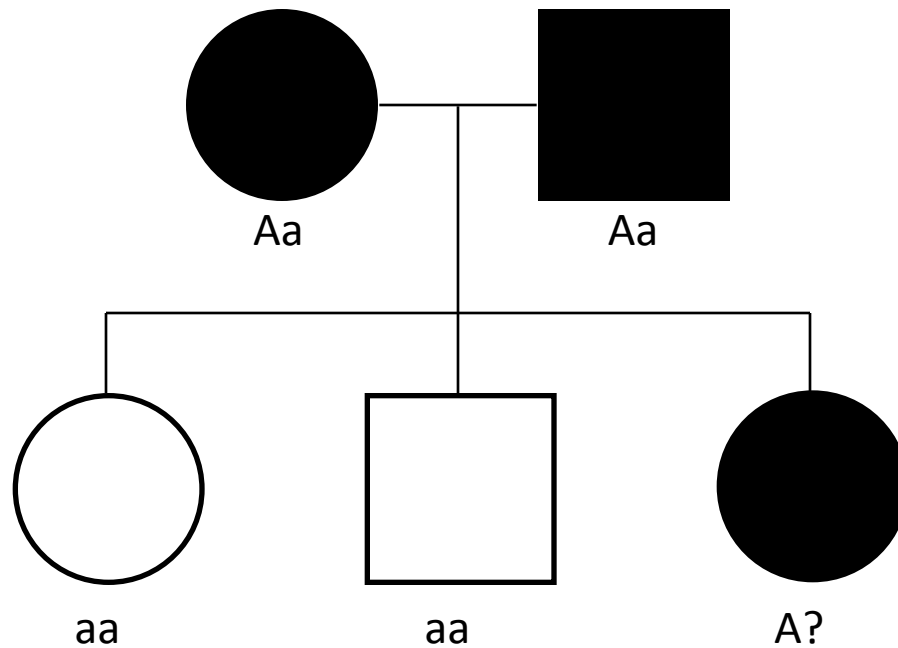# Example 3 - Can this pedigree be for a trait that is autosomal dominant?

A = dominant
a = recessive

Aa

Aa

aa

aa

A?

This individual has the trait so she must have at least one "A." The other allele could be "A" or it could be "a."

Example 3 - Can this pedigree be for a trait that is autosomal dominant?
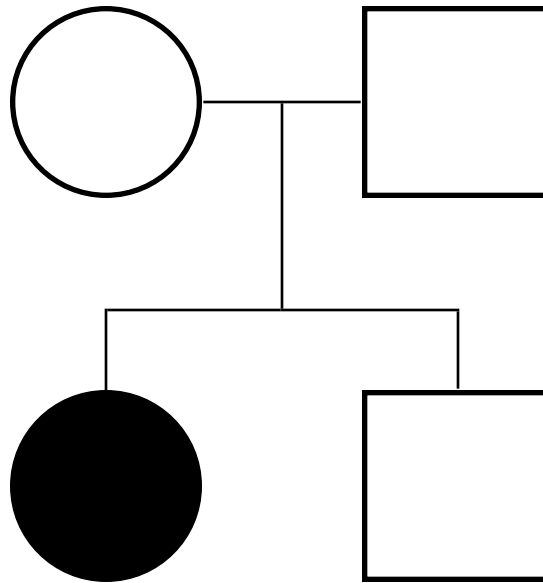
A = dominant
a = recessive

From example 3, we can conclude that if two parents have a dominant trait, their children might or might not have the trait.

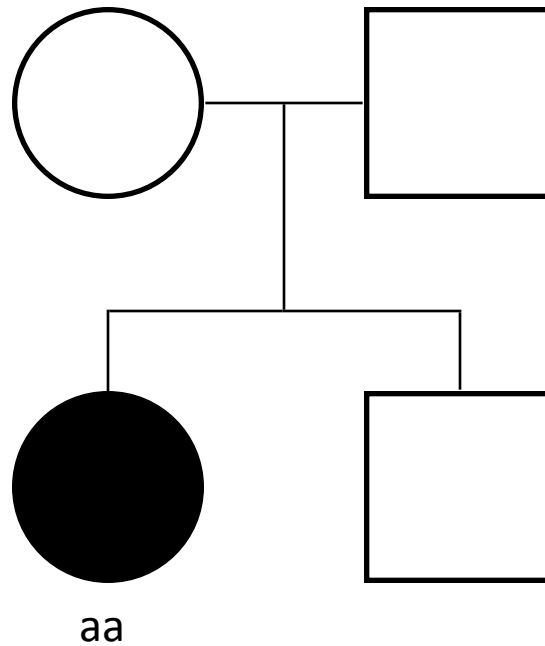# Example 4 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

The answer to this question is on the next several slides.

A = dominant
a = recessive

aa

This individual has the recessive phenotype so she must be "aa."
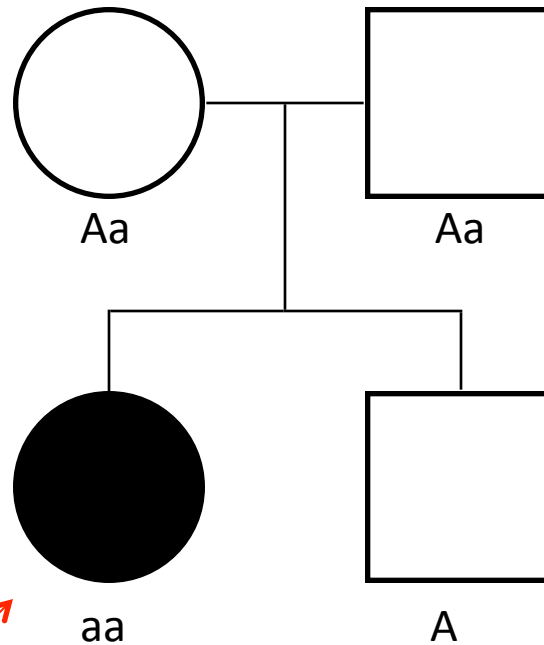
A = dominant
a = recessive
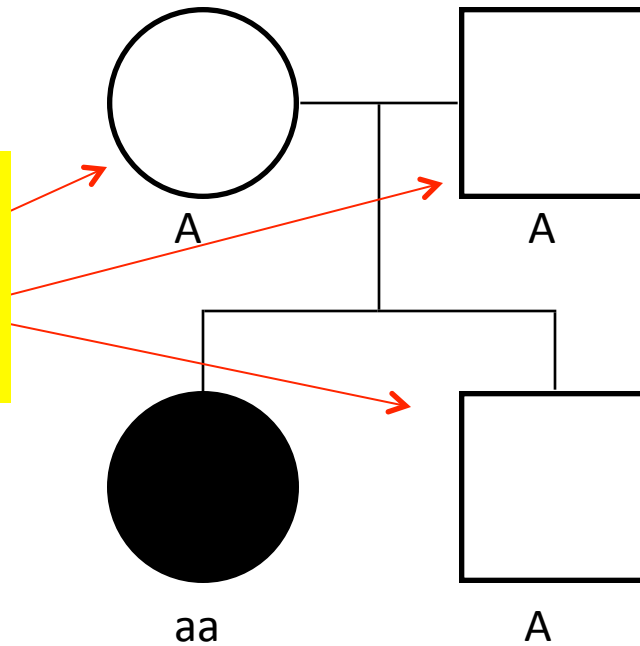
These individuals have the dominant phenotype so they must have at least one "A" allele.

A

A

aa

A

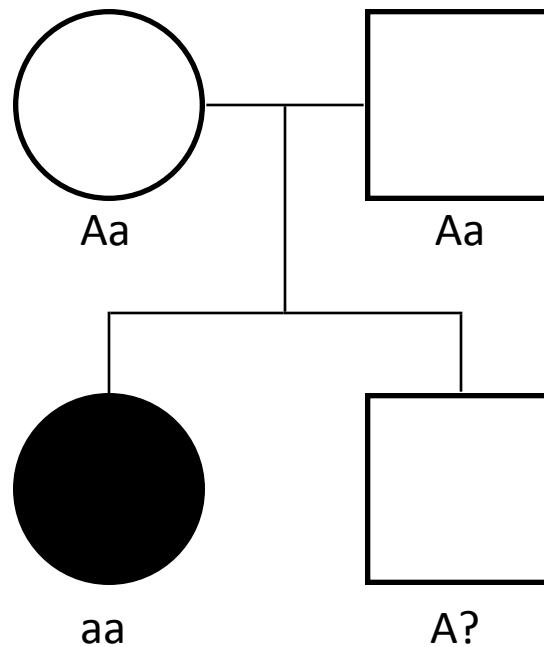# Example 4 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

Aa   Aa

aa   A

This individual has two "a" alleles.
One comes from each parent.
Therefore each parent must have at least one "a" allele.

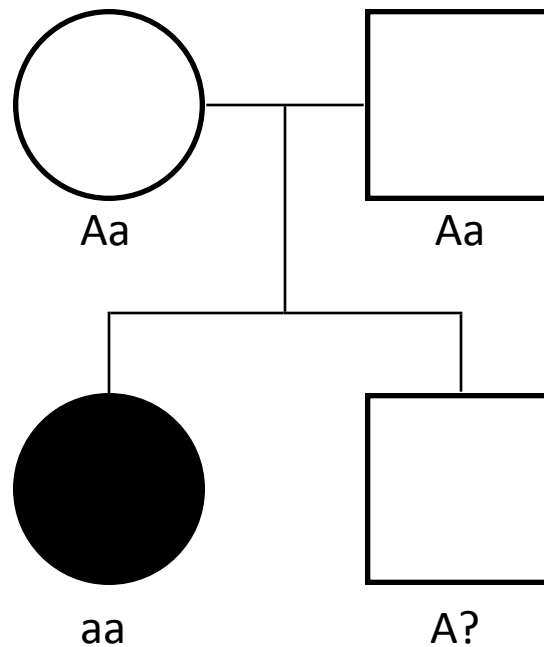# Example 4 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

Aa   Aa

aa   A?

This individual has the dominant trait so he must have at least one "A." The other gene could be "A" or it could be "a."

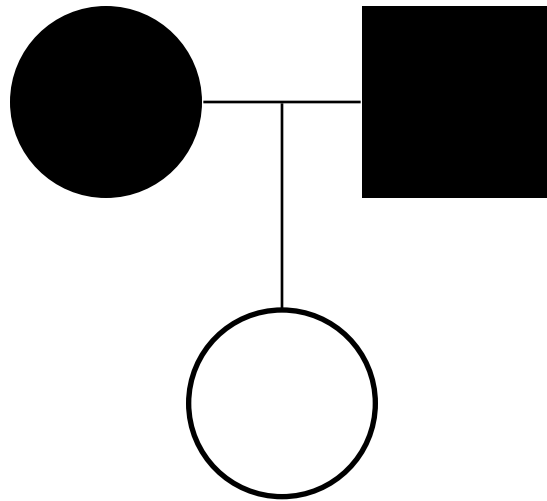# Example 4 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

Aa   Aa

aa   A?

From example 4 we can conclude that this pedigree can be for a trait that is autosomal recessive. A person that has an autosomal recessive trait can have two parents that do not have the trait. This is not true for dominant traits (example 2).

Example 5 - Can this pedigree be for a trait that is autosomal recessive?
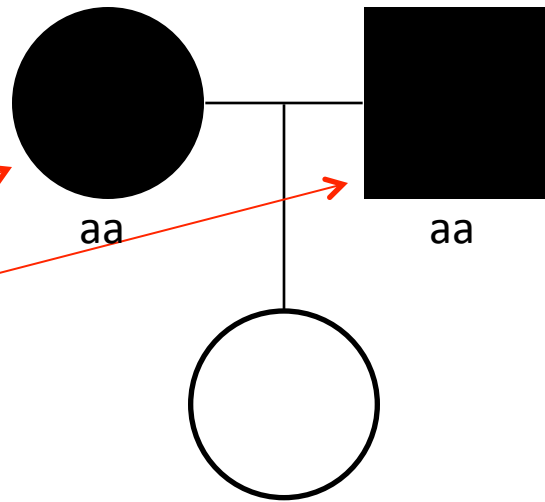
A = dominant
a = recessive

The answer to this question is on the next several slides.

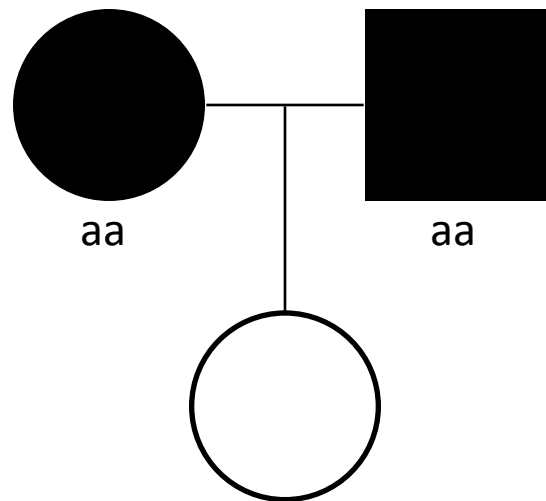# Example 5 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

These individuals have the recessive phenotype so they must be "aa."
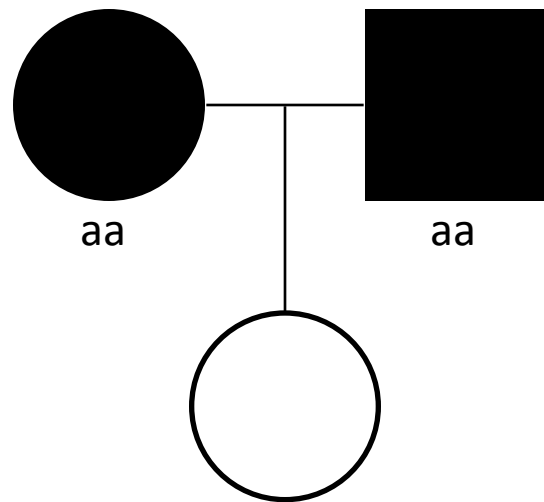
aa   aa

A = dominant
a = recessive

aa   aa

An "aa" individual can only produce "a" gametes. Therefore, this individual must be "aa." It is not possible for her to have the dominant phenotype as shown. This pedigree is not possible.

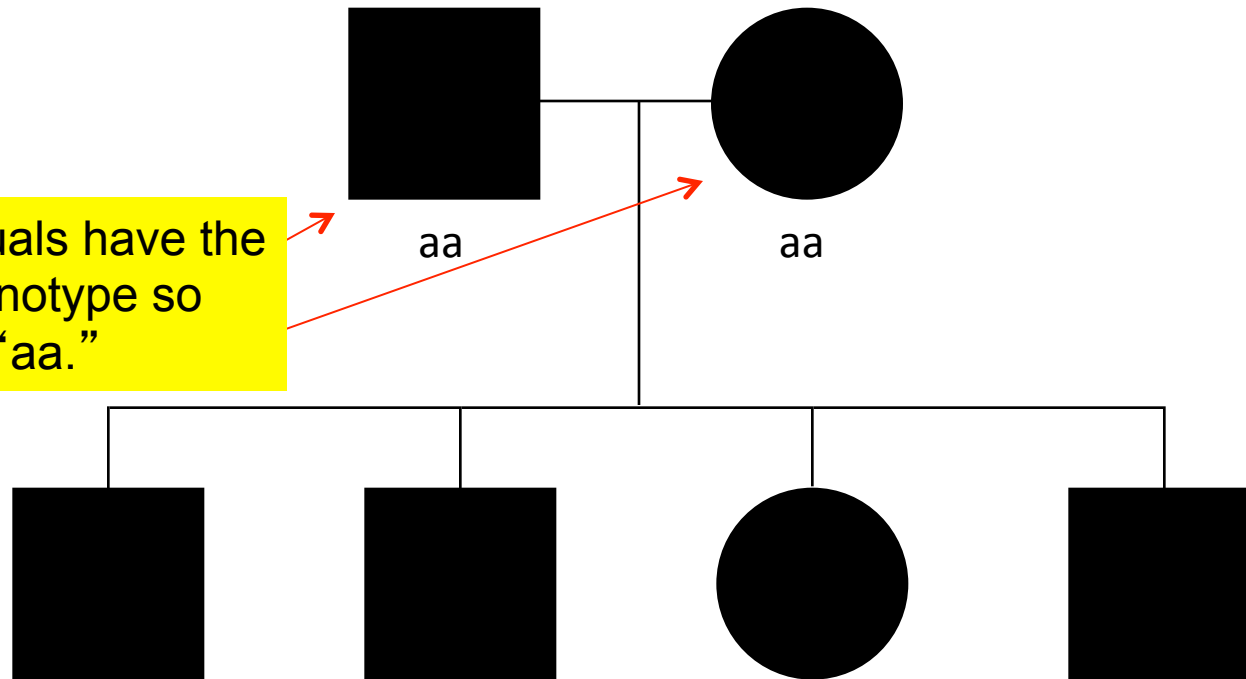# Example 5 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

aa   aa

From example 5 we can conclude that if two individuals have the recessive phenotype, the children must have the phenotype.

A = dominant
a = recessive

These individuals have the recessive phenotype so they must be "aa."

aa   aa

A = dominant
a = recessive

aa   aa

aa   aa   aa   aa

An "aa" individual can only produce "a" gametes. Therefore, all of the children must be "aa" and have the recessive phenotype.

Example 5 (continued) - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

aa   aa

aa   aa   aa   aa

From example 5 we can conclude that if two individuals have the recessive phenotype, all of the children must have the phenotype.

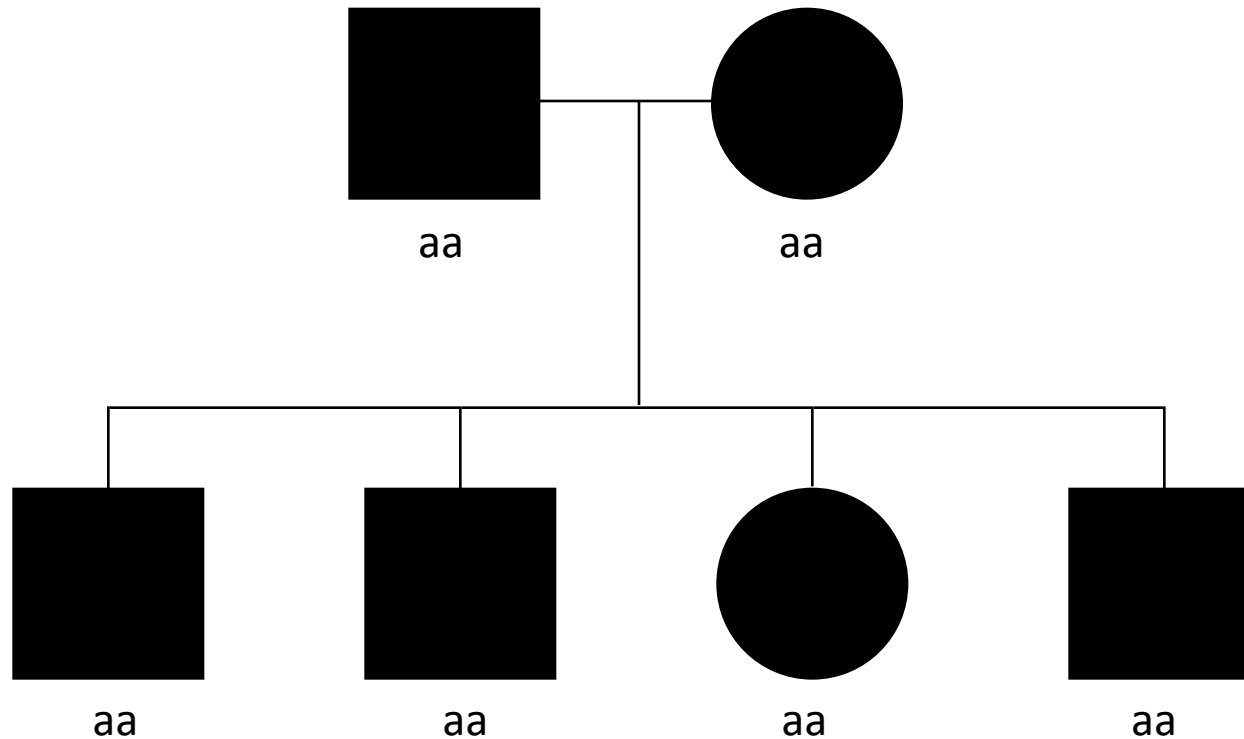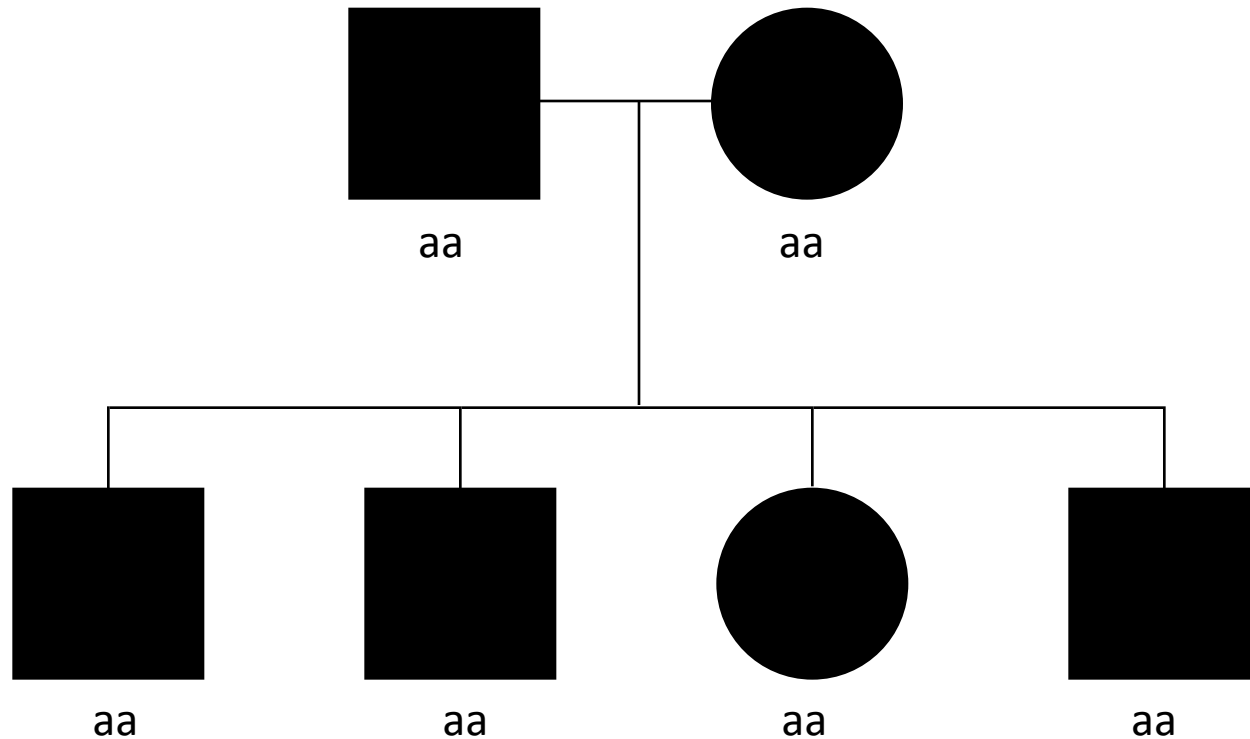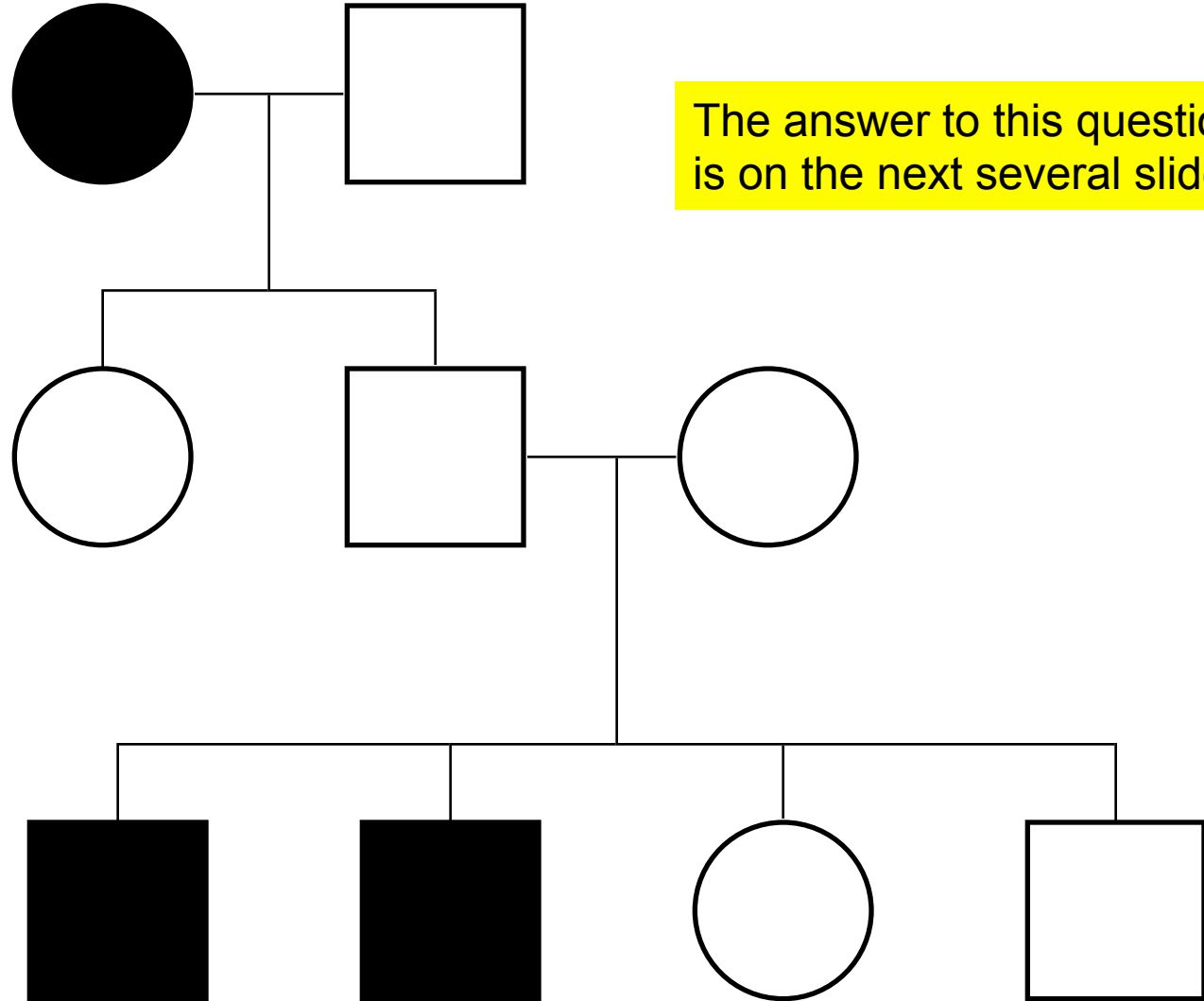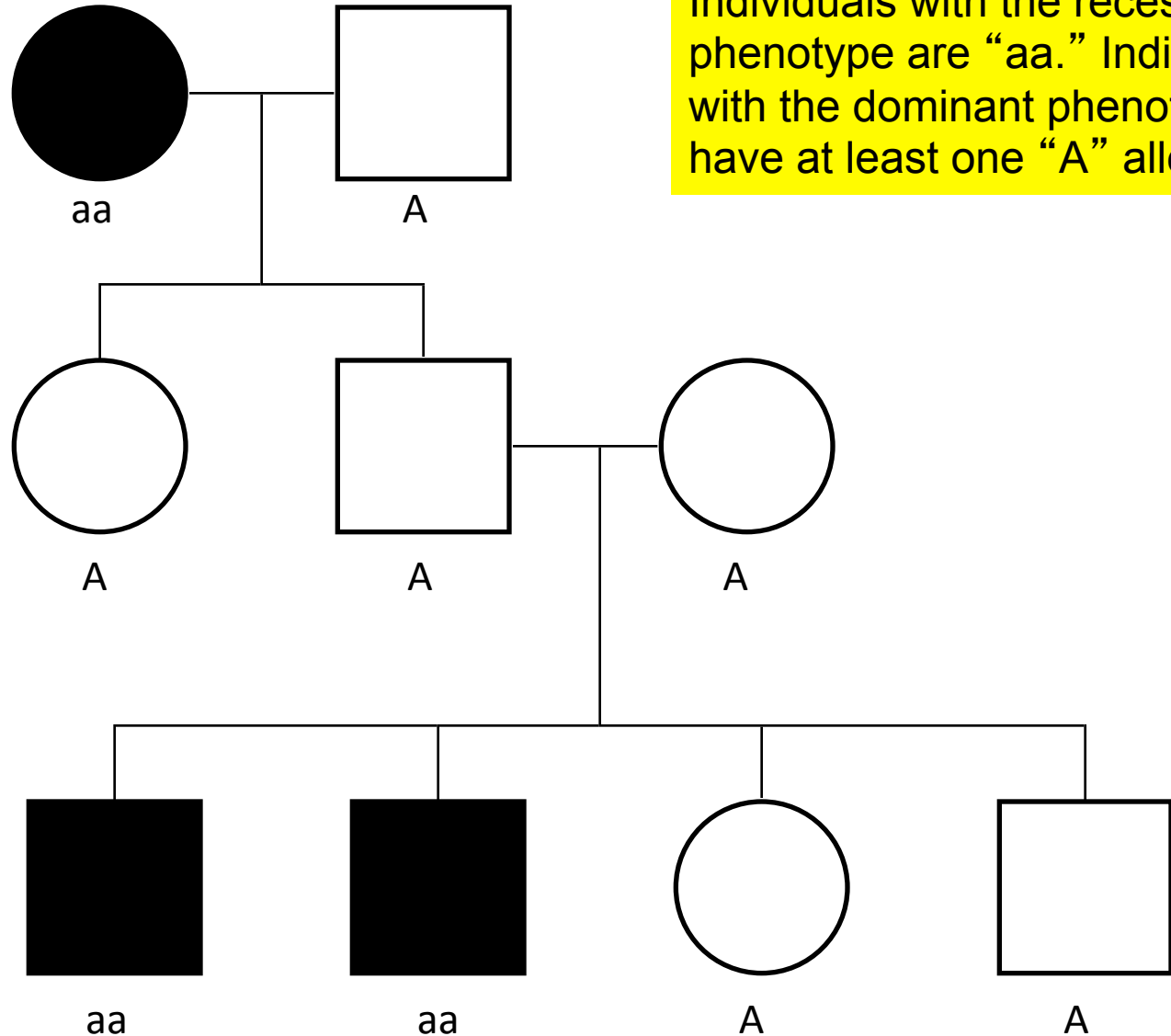# Example 6 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

The answer to this question is on the next several slides.

Example 6 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
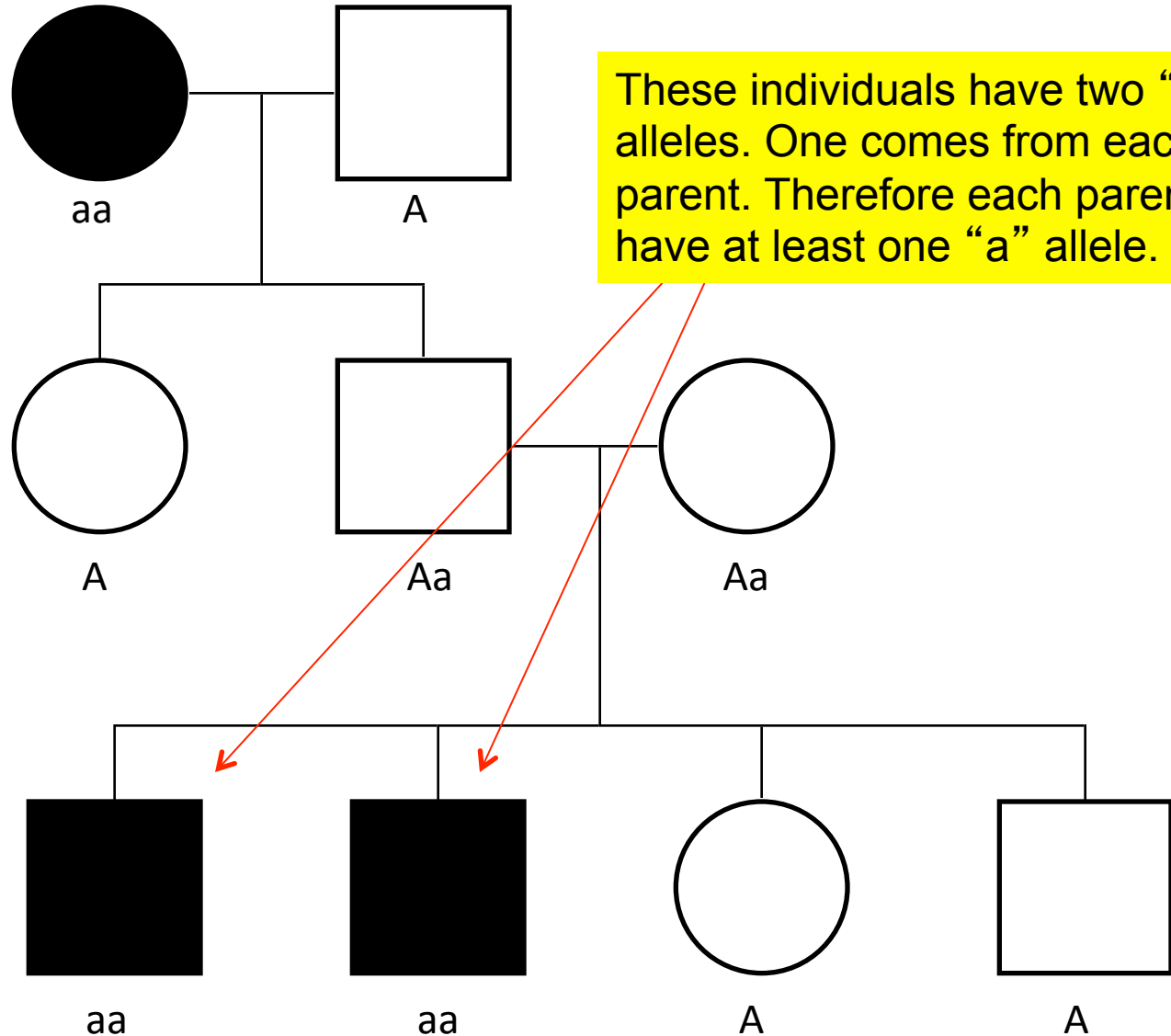a = recessive

Individuals with the recessive phenotype are "aa." Individuals with the dominant phenotype have at least one "A" allele.

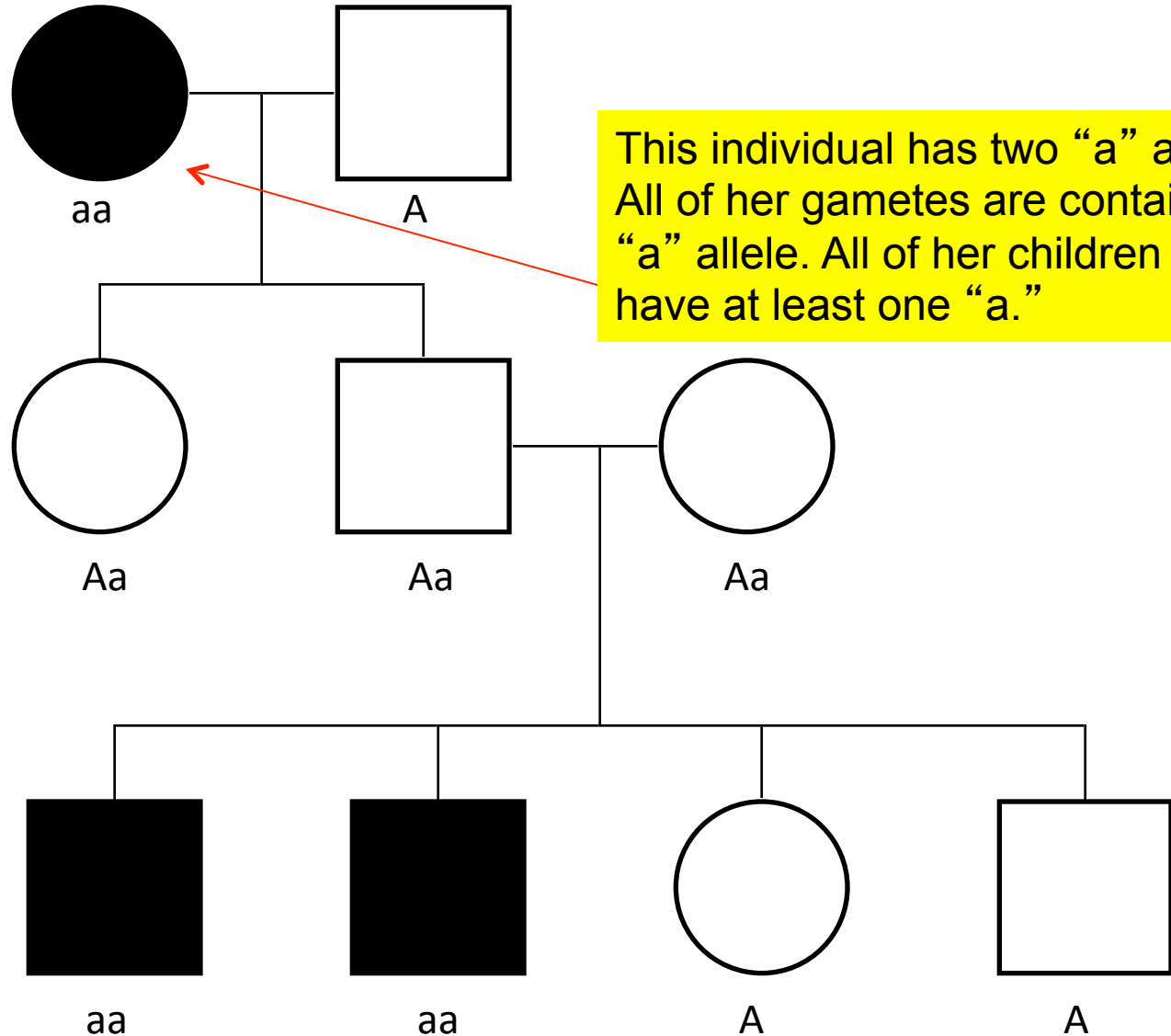# Example 6 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

These individuals have two "a" alleles. One comes from each parent. Therefore each parent must have at least one "a" allele.

aa

A

A

Aa

Aa

aa

aa

A

A

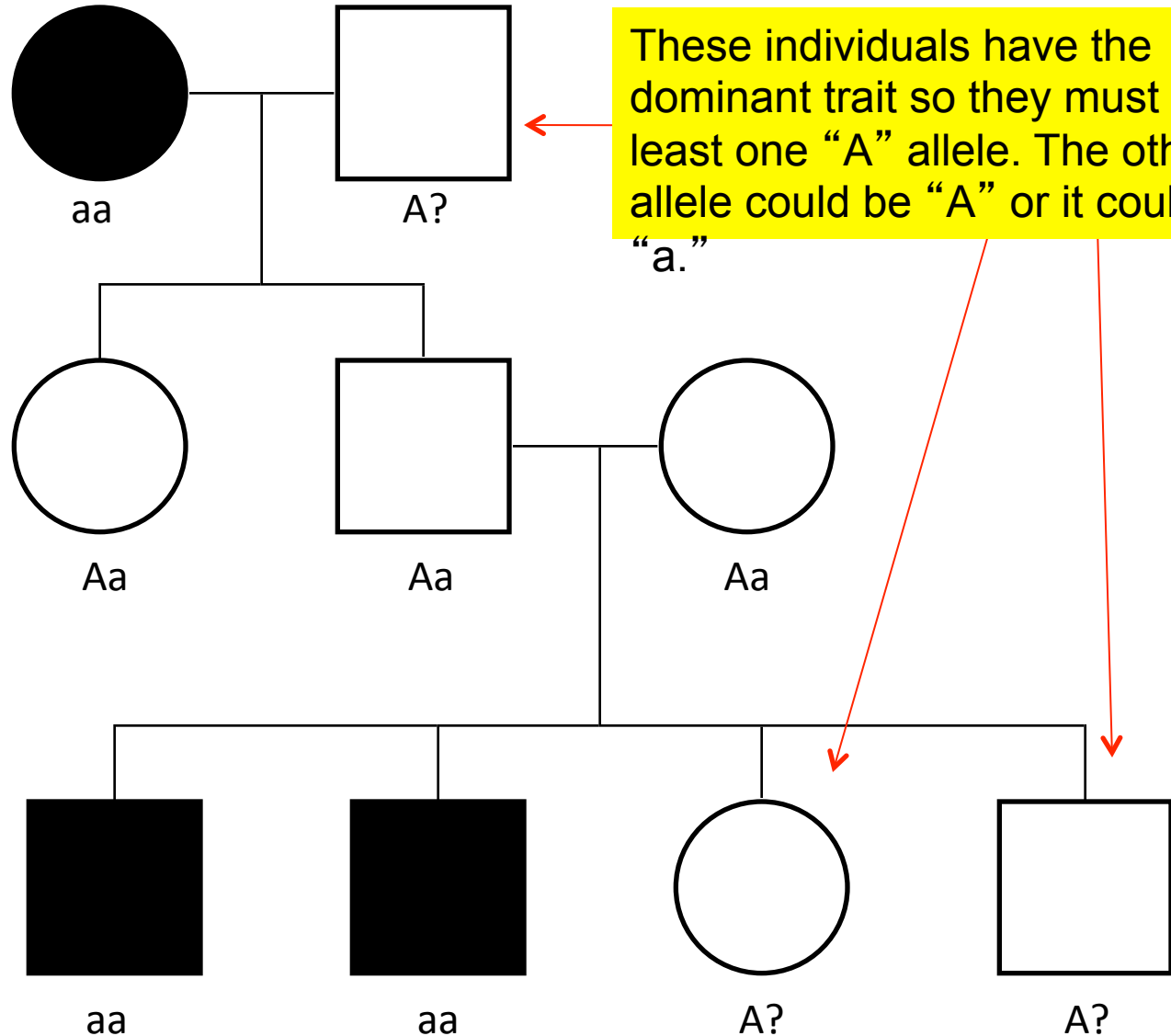# Example 6 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

aa

A

This individual has two "a" alleles. All of her gametes are contain one "a" allele. All of her children must have at least one "a."

Aa

Aa

Aa

aa

aa

A

A

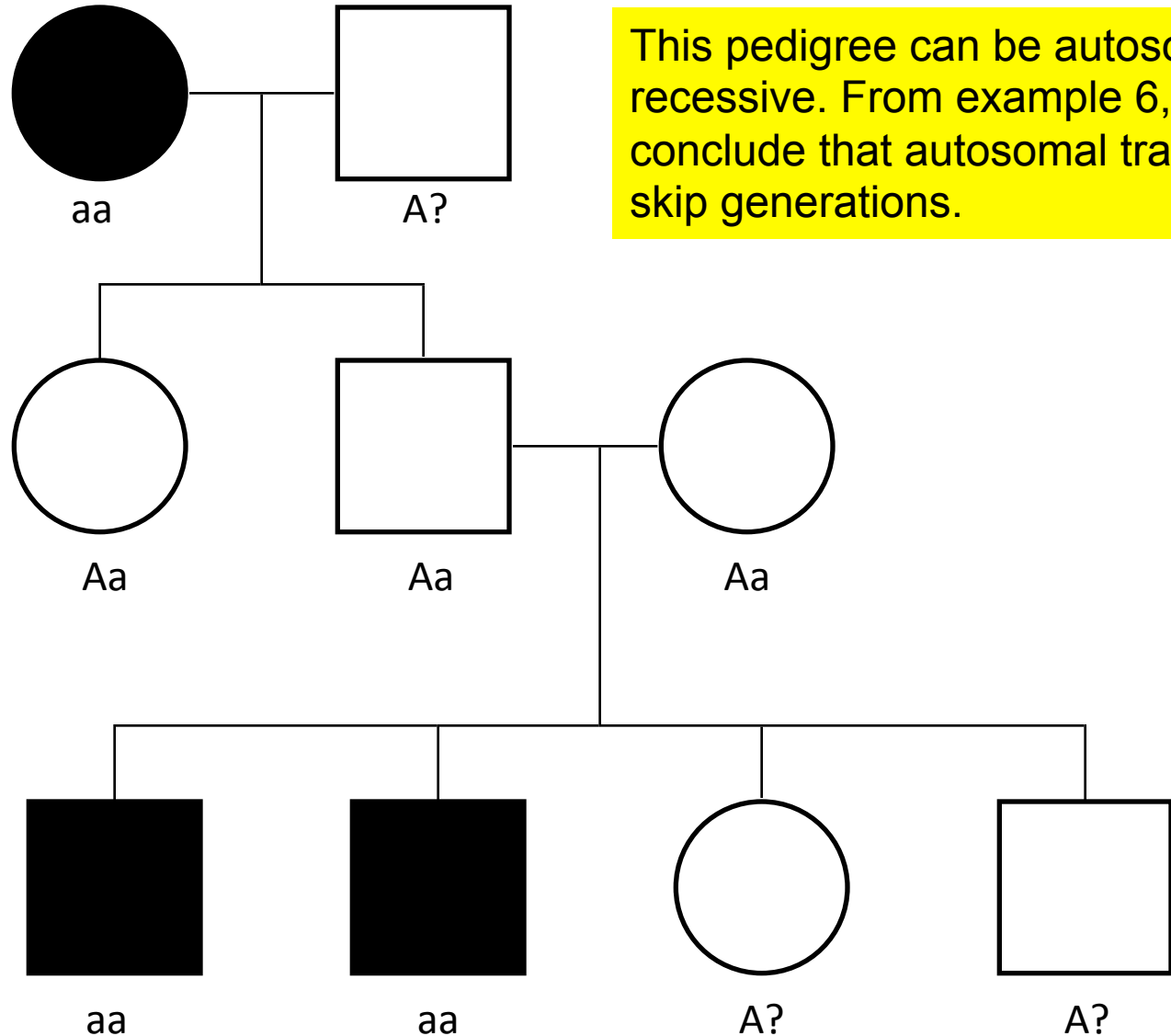# Example 6 - Can this pedigree be for a trait that is autosomal recessive?

A = dominant
a = recessive

These individuals have the dominant trait so they must have at least one "A" allele. The other allele could be "A" or it could be "a."

aa

A?

Aa

Aa

Aa

aa

aa

A?

A?

Example 6 - Can this pedigree be for a trait that is autosomal recessive?
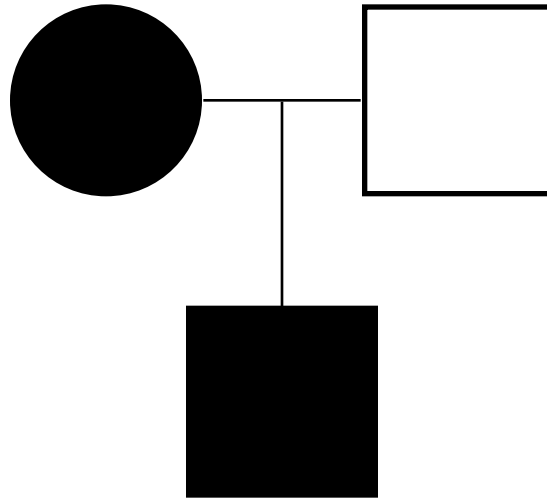
A = dominant
a = recessive

This pedigree can be autosomal recessive. From example 6, we can conclude that autosomal traits can skip generations.

aa

A?

Aa

Aa

Aa

aa

aa

A?

A?

Example 7 - Can this pedigree be for a trait that is X-linked recessive?
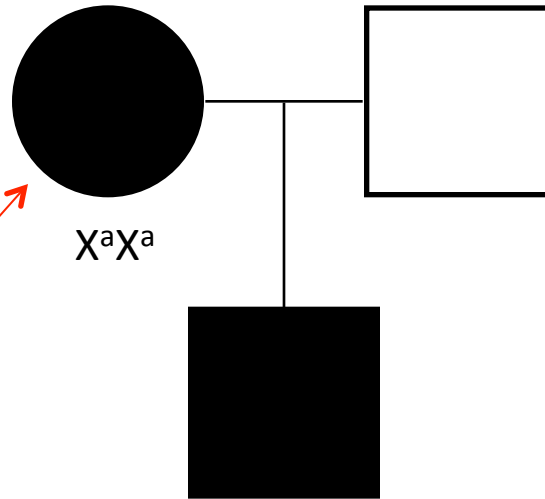
X^A = dominant
X^a = recessive

The answer to this question is on the next several slides.

Example 7 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
X^a = recessive

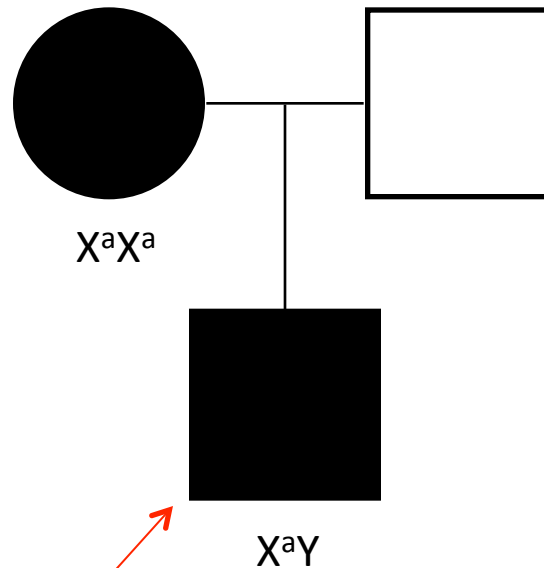$X^a X^a$

Females have two X chromosomes (XX). This individual has the recessive phenotype, so her genotype is $X^a X^a$.

Example 7 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
X^a = recessive

$X^a X^a$

$X^a Y$

Males have one X chromosome and one Y chromosome (XY). This individual has the recessive phenotype, so his genotype is $X^a Y$.

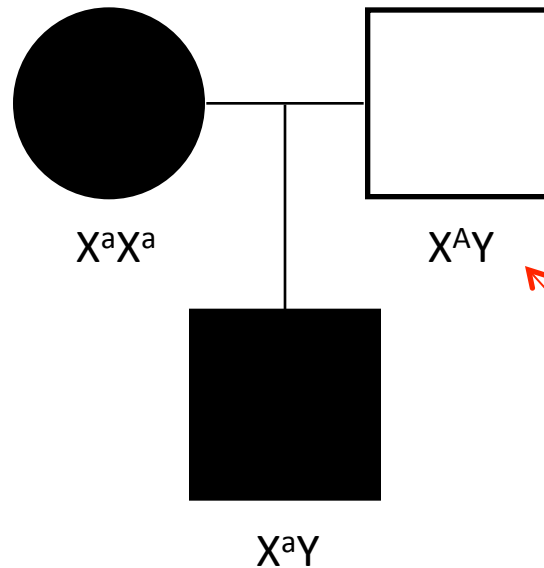# Example 7 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
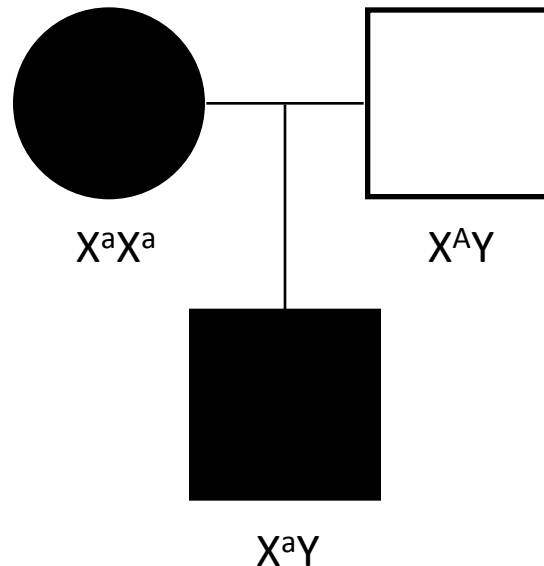X^a = recessive

X^aX^a

X^AY

X^aY

The father has the dominant phenotype, so his genotype is X^AY.

Example 7 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
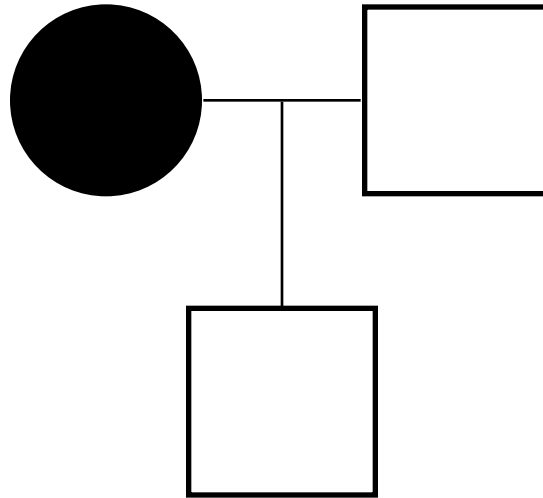X^a = recessive

X^aX^a X^AY

X^aY

The son inherited his Y chromosome from his father and the X^a gene from his mother. This pedigree can be for a trait that is X-linked recessive.

Example 8 - Can this pedigree be for a trait that is X-linked recessive?
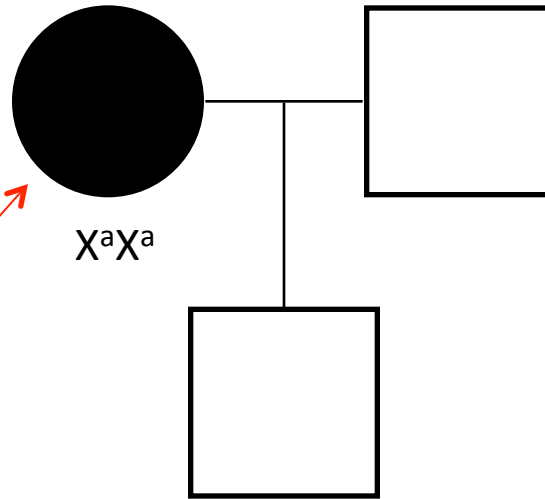
X^A = dominant
X^a = recessive

The answer to this question is on the next several slides.

Example 8 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
X^a = recessive

X^aX^a

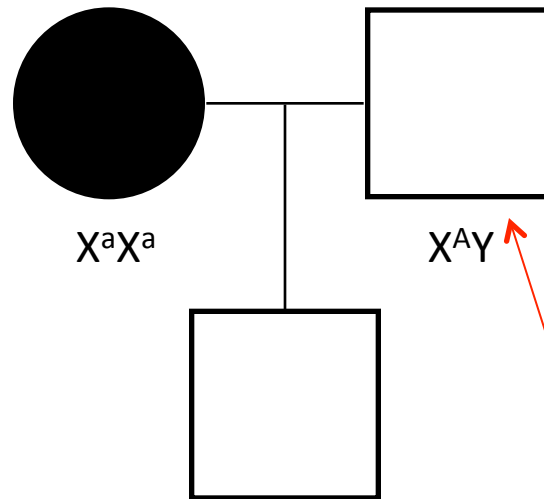Females have two X chromosomes (XX). This individual has the recessive phenotype, so her genotype is X^aX^a.

Example 8 - Can this pedigree be for a trait that is X-linked recessive?
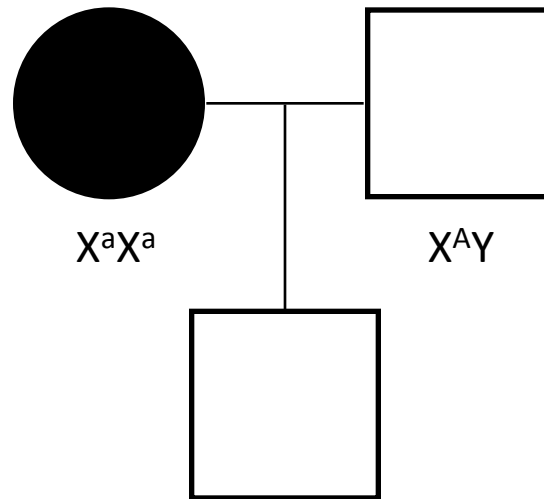
X^A = dominant
X^a = recessive

X^aX^a

X^AY

Males have one X chromosome and one Y chromosome (XY). These individual have the dominant phenotype, so their genotype must be X^AY. (continued, next slide)

Example 8 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
X^a = recessive

X^aX^a X^AY

The son can only receive a Y chromosome from his father. If he were to inherit the X chromosome, he would be female (XX). Males inherit their X chromosome from their mother. All of the gametes produced by the mother in this example are X^a. If the son's genotype were X^aY, he would have the recessive phenotype. This pedigree is not possible because he has the dominant phenotype. The trait cannot X-linked recessive.

Example 8 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
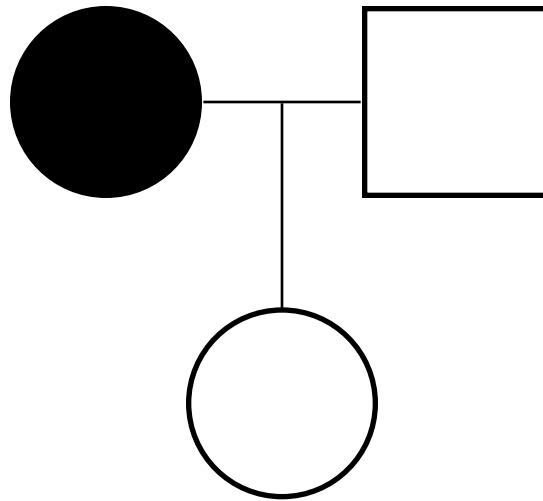X^a = recessive

X^aX^a X^AY
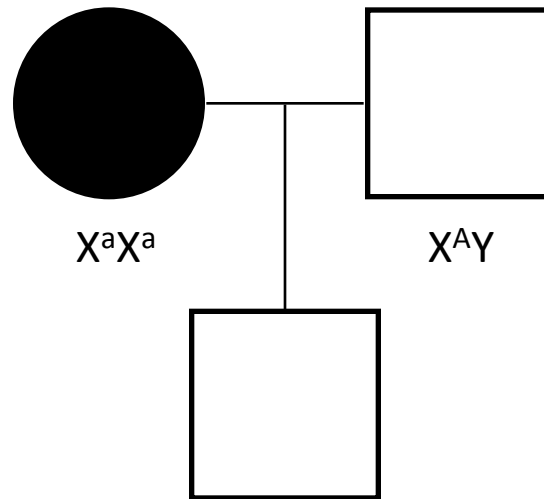
From example 8, we can conclude that if a female has an X-linked recessive trait, her sons will have the trait.

Example 9 - Can this pedigree be for a trait that is X-linked recessive?
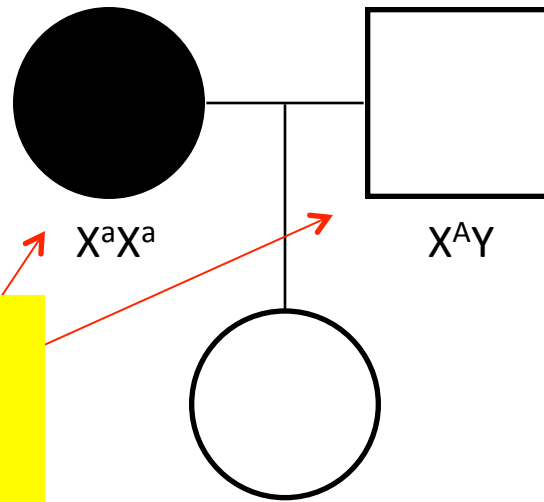
X^A = dominant
X^a = recessive

The answer to this question is on the next several slides.

Example 9 - Can this pedigree be for a trait that is X-linked recessive?
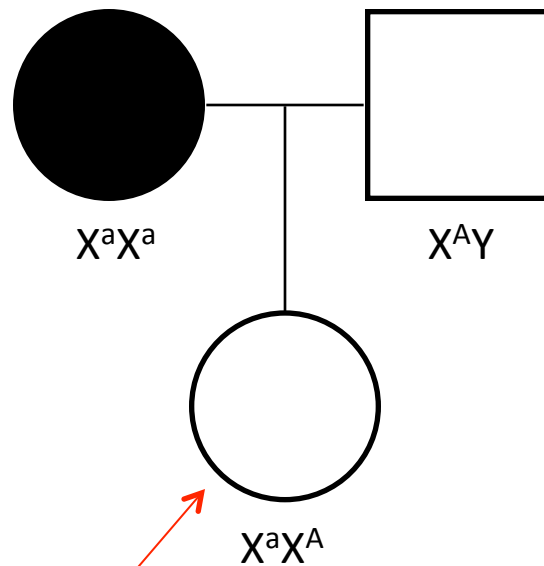
X^A = dominant
X^a = recessive

X^aX^a

X^AY

As discussed in examples 7 and 8, females with the recessive phenotype are X^aX^a. Males with the dominant phenotype are X^AY.

Example 9 - Can this pedigree be for a trait that is X-linked recessive?

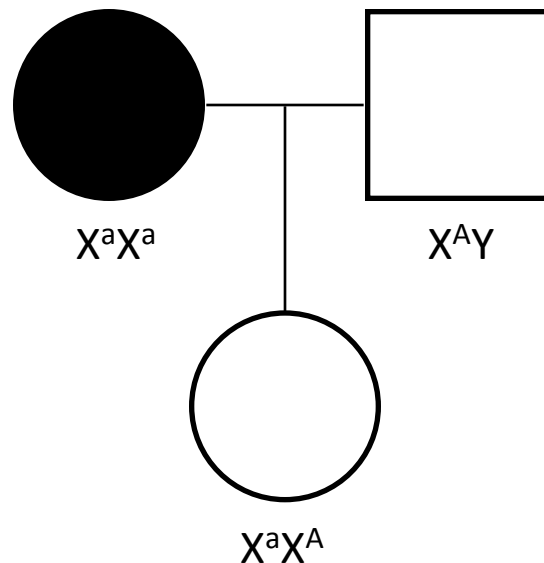X^A = dominant
X^a = recessive

X^aX^a X^AY

X^aX^A

The daughter received an X chromosome from each parent. Her mother's gametes are all have the X^a allele and her father's X chromosome has the X^A allele. Therefore, her genotype is X^aX^A and she has the dominant phenotype.

Example 9 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
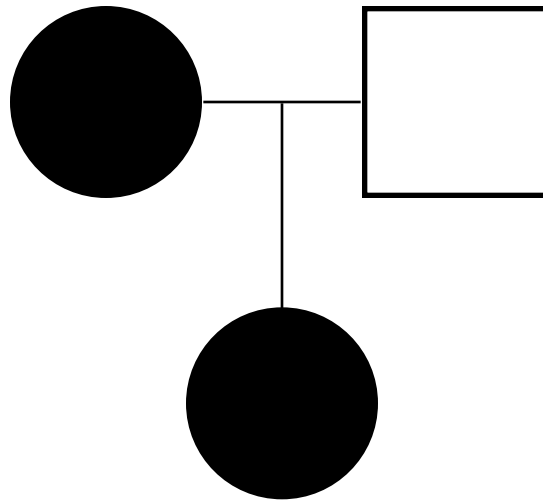X^a = recessive

$X^a X^a$

$X^A Y$

$X^a X^A$

In example 8 we concluded that all of the sons of a female with an X-linked recessive trait will have the recessive phenotype. In the present example (example 9), we conclude that the daughters might not have the trait.

Example 10 - Can this pedigree be for a trait that is X-linked recessive?
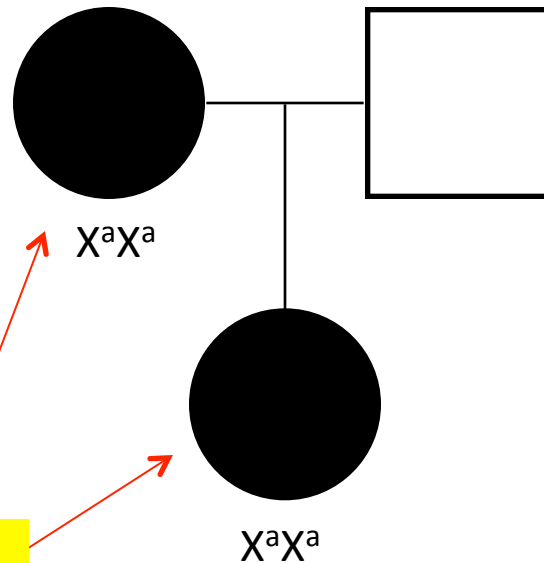
X^A = dominant
X^a = recessive

The answer to this question is on the next several slides.

Example 10 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
X^a = recessive

X^aX^a

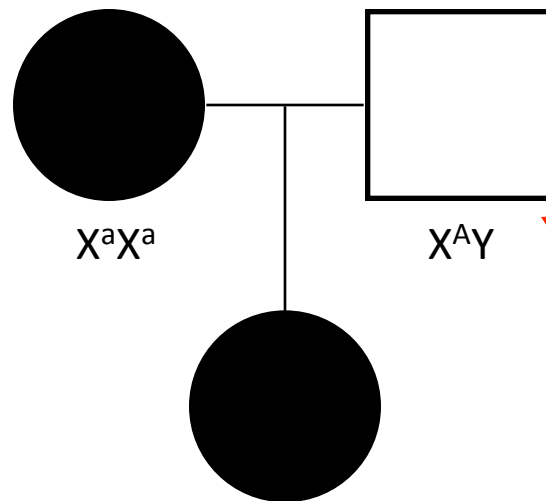X^aX^a

Females with an X-linked recessive phenotype are X^aX^a.

Example 10 - Can this pedigree be for a trait that is X-linked recessive?

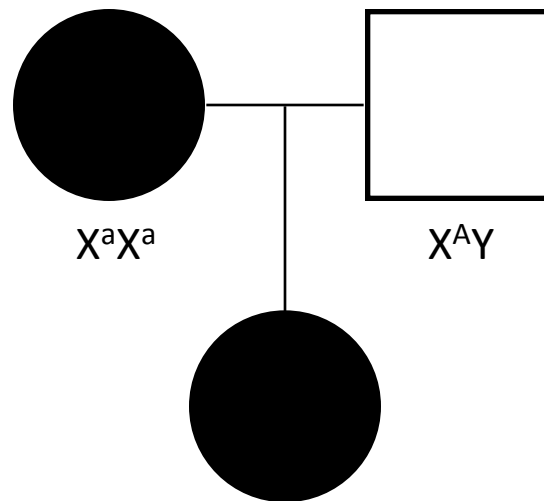X^A = dominant
X^a = recessive

X^aX^a

X^AY

Males with the dominant phenotype are X^AY.

Example 10 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
X^a = recessive

X^aX^a X^AY

This male can produce two kinds of gametes: X^A and Y. All of his daughters will receive the X^A allele. If the daughter has an X^A allele, she will have the dominant phenotype like her father. However, the daughter in this pedigree has the recessive phenotype. The trait in this pedigree therefore cannot be X-linked recessive.

Example 10 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
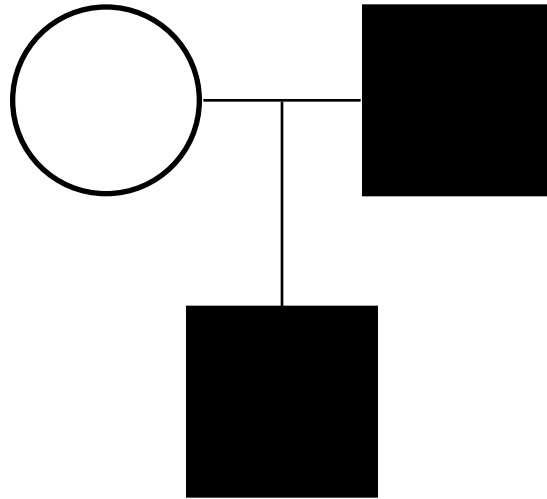X^a = recessive

X^aX^a X^AY
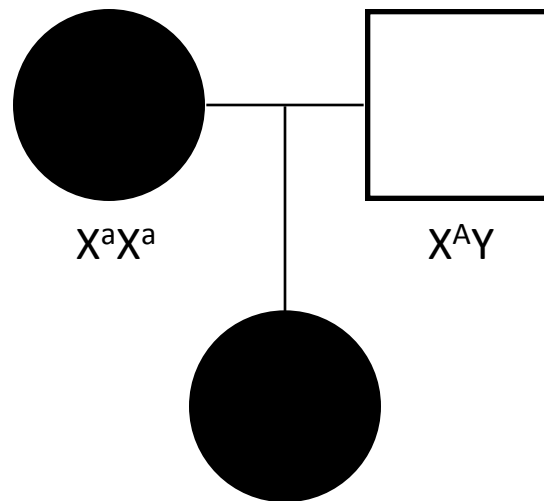
From example 10 we can conclude that if a female has an X-linked recessive trait, her father must also have the trait. If the father had the trait (X^aY), she would inherit an X^a allele from him and she would have the trait.

Example 11 - Can this pedigree be for a trait that is X-linked recessive?
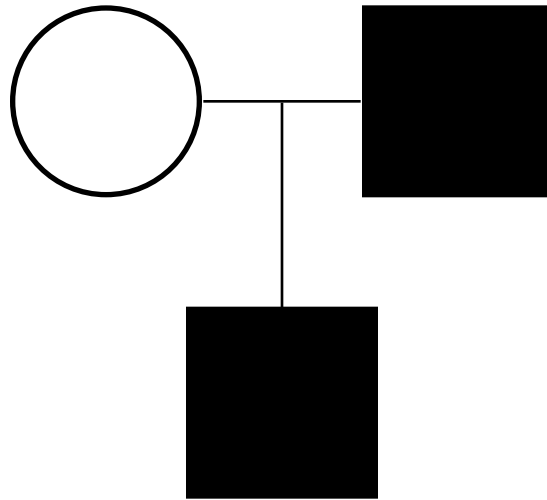
X^A = dominant
X^a = recessive

The answer to this question is on the next several slides.

Example 11 - Can this pedigree be for a trait that is X-linked recessive?
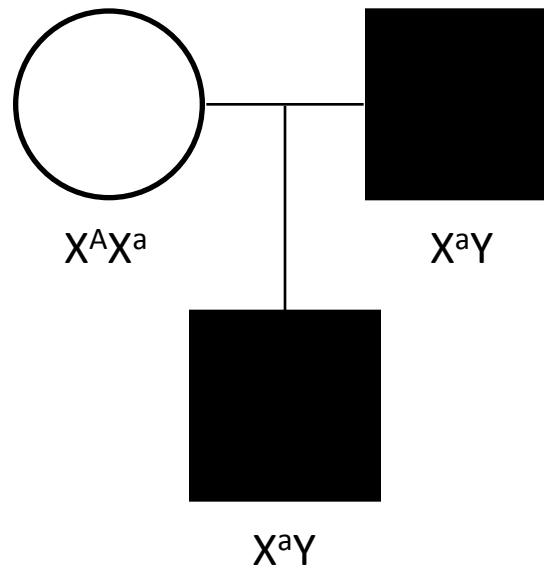
X^A = dominant
X^a = recessive

A father cannot pass an X-linked trait to his son because he gives his son the Y chromosome. At first glance, it appears that this pedigree cannot be for a trait that is X-linked recessive because the father appears to have given his son an X chromosome.

Example 11 - Can this pedigree be for a trait that is X-linked recessive?

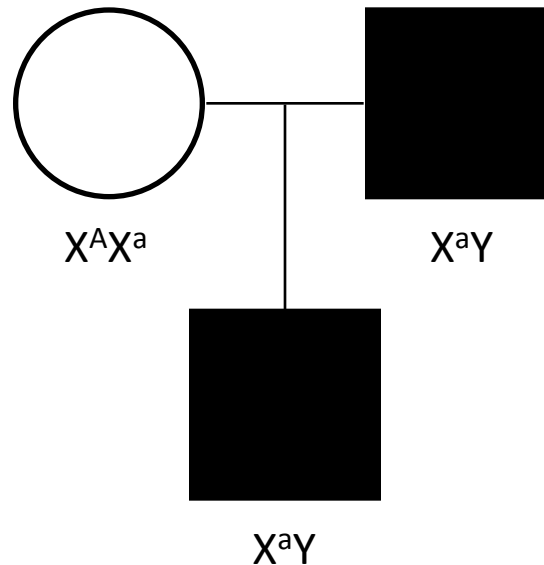X^A = dominant
X^a = recessive

$X^A X^a$

$X^a Y$

$X^a Y$

However, it is possible that the son received the X^a allele from his mother, not his father. The pedigree can therefore be for an X-linked recessive trait.

Example 11 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
X^a = recessive
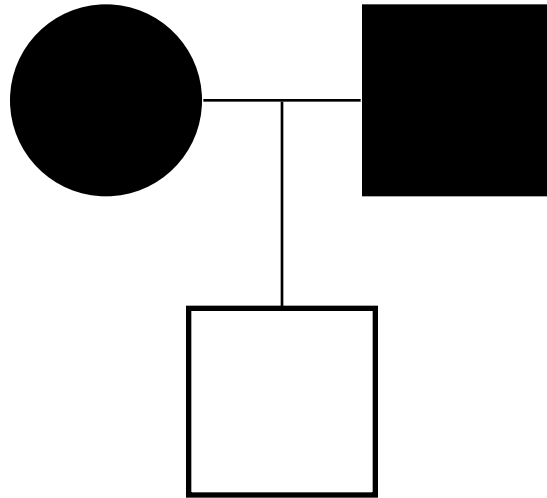

X^AX^a X^aY

X^aY

From example 11, we can conclude that father-to-son transmission of X-linked recessive traits does not occur. Keep in mind, however, that the pedigree above is possible because the son inherited the trait from his mother.

Example 12 - Can this pedigree be for a trait that is X-linked recessive?
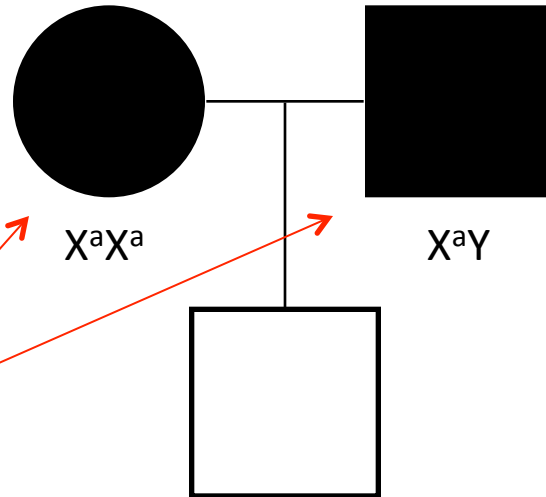
X^A = dominant
X^a = recessive

The answer to this question is on the next several slides.

Example 12 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
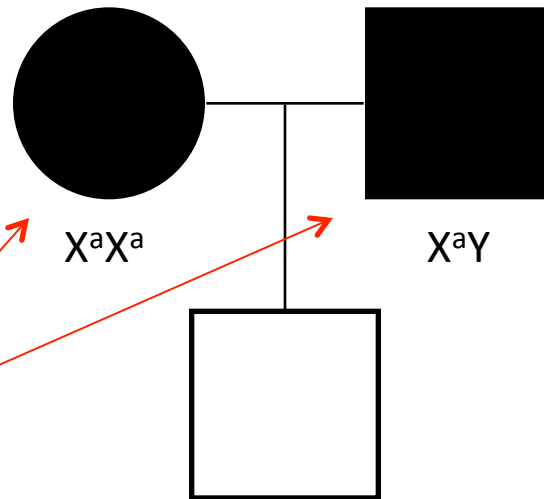X^a = recessive

$X^a X^a$

$X^a Y$

These individuals have the recessive phenotype. They carry only the recessive alleles.

Example 12 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
X^a = recessive

X^aX^a

X^aY

The female's gametes will all carry the X^a allele. One half of the male's gametes will carry X^a, the other half will carry the Y chromosome.

All of the children will have the recessive trait. Females will receive one X^a allele from each parent and will be X^aX^a. Males will receive one X^a allele from their mother and the Y chromosome from their father. Their genotype will be X^aY. The pedigree above is not possible because the male child does not have the trait.

Example 12 - Can this pedigree be for a trait that is X-linked recessive?

X^A = dominant
X^a = recessive

X^aX^a X^aY
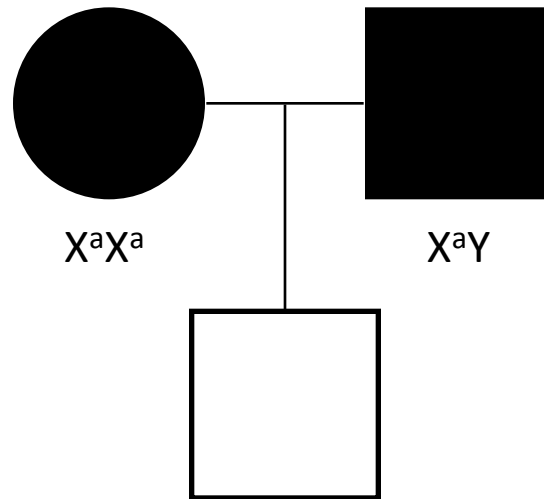
From example 12 we can conclude that if two parents have an X-linked recessive trait, all of their children will have the trait. This is the same conclusion that we had from example 5.