

## Biology: Student Work for Hemophilia and PKU CR Prompt

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- The difference between sex chromosomes and autosomal chromosomes is that a sex chromosome is the last two chromosomes that decides your gender and an autosomal chromosome is the other 44 chromosomes that you get from your parents.
- Females are less likely to inherit Hemophilia because their sex chromosomes has 2 X's while males have one X and one Y. Since Hemophilia is a human X-linked recessive disorder it would be less likely that both of the X-chromosomes (that a female has) carries Hemophilia. If she has one X with Hemophilia she doesn't have Hemophilia but she will be a carrier of Hemophilia. While males have one X. If that one X chromosome inherits Hemophilia the male automatically has Hemophilia since it is an X linked disorder, and not a Y linked disorder.
- Since males and females have the same amount of autosomal chromosomes, the chances of a male having PKU is the same as a female.

## Assessments

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A genetically viable human being has 23 pairs of chromosomes. This total of 46 genetic carriers gives us great versatility in both function and form. However, as with every machine, we are not all perfect. There are several cases in which abnormalities occur, and these can be traced back to our chromosomes. One must first understand that each chromosome has another that pairs with it. We inherit one for each pair from our two parents, yielding the two. The first 22 pairs are known as ~~the~~ autosomal chromosomes, while the remaining pair is our sex chromosomes that determine gender. In males, the first sex chromosome is labeled X and the second is labeled Y. In females, they are known as X<sub>1</sub> and X<sub>2</sub>. Females pass down an X chromosome while males may pass down either a X or a Y. Diseases that are sex-linked, such as hemophilia, are known as such due to their genetic location being on a sex chromosome. The rest are found on autosomal chromosomes which determine characteristics ranging from eye color to hair color. One must completely comprehend this unique differentiation in order to further study human genetics and the errors that may occur.

As with any genetic disease, the first step in identifying is finding its chromosomal location. ~~Hemophilia is a sex-link~~ In the case of hemophilia, the disease where blood loses its ability to clot, ~~it~~ it is a sex-linked trait. Hemophilia is recessive, found on the X chromosome, and is inherited from a parent. Due to the fact that hemophilia may be passed from either parent, carriers (and not just those with the disease) should be taken into account. Since females have two X chromosomes, both of their parents must carry the disease. In males, only one recessive allele from the mother must be passed down to see the disease (the dad will pass

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the Y chromosome.) Because of this, females have less of a chance to inherit the disease; both, the X chromosome of the dad and one of the chromosomes of the mom must carry the ~~gene~~ allele, while only one of the female parents X chromosomes needs to be passed down for a male offspring to have the disease.

Others diseases that involve <sup>autosomal</sup> chromosome genes, such as phenylketonuria (PKU) are indiscriminate from gender. The most relevant fact that supports this is that PKU is not sex-linked and is recessive. Therefore, both of the alleles on the chromosomes belonging to a pair have to carry the disease for its symptoms to appear. Analyzing the fact that there are 2 parents, each with a pair of their own chromosomes (a total of 4), and that only 2 will be passed down, we know the result for the offspring is all up to chance. There is no influence from gender and reduced or pair variation. The offspring will only get the disease if one parent passes down a ~~gene~~ allele with PKU and the other parent does the same, regardless of whether the offspring is male or female.

# Assessments

## Student Work for Hemophilia and PKU CR Prompt

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• Sex chromosomes are completely different from autosomal chromosomes. The sex chromosomes are decided from the gender of the human being. It depends on whether it is a boy or a girl. An autosomal chromosomes doesn't have anything to do with the gender of a human being.

• Females are less likely to develop hemophilia than Males because Females are recessive compared to the Males who are dominant. Hemophilia is a recessive disorder. According to my data, boys have a 3:1 ratio of developing hemophilia compared to girls.

	$X^H$	$Y$
$X^H$	$X^H X^H$	$X^H Y$
$X^h$	$X^H X^h$	$X^h Y$

3:1 • Females and Males have an equal chance of developing PKU because it is not a sex related disorder. It doesn't matter on the gender of the person.

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A sex chromosome is something that is carried solely on the X or Y chromosome and an autosomal chromosome is carried on ~~either~~ both the X or Y chromosome.

Females are less likely to inherit hemophilia because it is a recessive trait on the X chromosome and the chances that they will have a recessive trait on both X's is very small. Boys, however, only have one X chromosome so they have a greater chance.

Females and Males are equally likely to get PKU because it can be carried on the X and Y chromosomes and is not sex-linked.

## Assessments

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### Student Work for Hemophilia and PKU CR Prompt

Student E, Page 1 of 1

- Sex chromosomes are opposite of autosomal chromosomes.
- Why females are less likely to inherit hemophilia than males?
  - b/c if a female has an  $X^h$  and  $X^H$  she has a 50% chance, but all men need is one  $X^h Y$  and they will have it: since they don't have 2 X chromosomes like women.
- Why males & females are equally likely to inherit PKU?
  - because they have  $X^h$  and  $X^H$  she ~~has~~ still has a 50-50 chance but since  $X^h$  is recessive they are just as likely as males ( $X^h Y$ ).

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With Hemophilia its transferred through the X sex cell. The females have two X sex cells <sup>(XX)</sup> so they have twice as many chances to get the Dominate trait of not getting Hemophilia but the males sex cells are (X Y) so they only have  $\frac{1}{2}$  as many chances to get hemophilia or not get it. In PKU it doesn't matter what your sex is because it isn't transferred or inherited through your X sex cell. Females could have a P, p and not get PKU but males could also have P, p. It is all differred by how many chances you get from getting a Dominate trait to be able to not get these disorders.

## Assessments

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### Student Work for Hemophilia and PKU CR Prompt

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Males are more likely to contract hemophilia because they have only one X. The Female has two Xs and have a greater chance of becoming negative for hemophilia because the disease is recessive.

PKU has equal inheritance because it isn't a sex-linked disease. So both male and female have an equal chance of contracting the disease.

The difference between an autosomal and X-linked is that X-linked is another term for sex-linked, or a gene that's only inherited through intercourse.

## Biology: Hemophilia and PKU Constructed-Response Articulations

### Response A—Anchor Score 3

This response demonstrates a competent knowledge and understanding of the subject matter. The content of the response is generally correct, but lacks thoroughness. The response shows an understanding of autosomal chromosomes and sex chromosomes by addressing the fact that sex chromosomes determine gender and implying that there are 46 chromosomes in humans and 44 are autosomal. Conceptually, the response sufficiently explains why females are less likely to inherit a recessive trait carried on the X chromosome. *Since Hemophilia is a human X-linked recessive disorder it would be less likely that both of the X chromosomes (that a female has) inherits Hemophilia... While males have one X, if that one X chromosome carries hemophilia the male automatically has Hemophilia...* The response shows some general understanding related to the inheritance of a recessive autosomal chromosomes disorder. *Since males and females have the same amount of autosomal chromosomes, the chances of a male having PKU is the same as a female.* Further explanation of why this is so would have enhanced this response. The response is mostly clear and sometimes uses appropriate scientific terminology.

### Response B—Anchor Score 4

This response demonstrates a thorough knowledge and understanding of the subject matter. The content of the response contains elaboration and demonstrates a thorough understanding of scientific concepts with no misconceptions. The response correctly addresses the basic difference between sex and autosomal chromosomes. *The first twenty-two pairs are known as autosomal chromosomes, while the remaining pair is our sex chromosomes that determine gender.* Conceptually, the examinee logically explains why an X (sex)-linked recessive disorder is less likely to be inherited in females than males. *Because of this, females have less of a chance to inherit the disease; both, the X chromosome of the dad and one of the chromosomes of the mom must carry the disease, while only one of the female parent's X chromosomes needs to pass down for a male offspring to have the disease.* Continuing, the examinee correctly contrasts how the autosomal recessive disorder PKU is inherited. *There is no influence from gender on the autosomal trait, PKU. The offspring will only get the disease if one parent passes down an allele with PKU and the other does the same.* This implies some understanding that females and males both have an alternative version of an autosomal trait that could be inherited. The response is clearly communicated and uses appropriate scientific terminology.

## Scoring Articulations

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### Response C—Anchor Score 1

This response demonstrates a minimal knowledge and understanding of the subject matter. Even though the examinee addresses each bullet, the content of the response is mostly incorrect. *Females are less likely to develop hemophilia than males because females are recessive compared to the males who are dominant.* The response demonstrates little or no evidence of understanding of scientific concepts and contains misconceptions. *According to my data, boys have a 3:1 ratio of developing hemophilia compared to girls.* The examinee does not adequately use scientific terminology to communicate understanding, and the reader must interpret the response because it is unclear. *The sex chromosomes are decided from the gender of the human being. It depends on whether it is a boy or a girl.* This response contains little, if any, correct content or correct conceptual understanding.

### Response D—Anchor Score 2

This response demonstrates a basic knowledge and understanding of the subject matter. The content of the response is partially correct. The response reveals a partial understanding of autosomal chromosomes and sex chromosomes by addressing the bullet in regard to sex-linked and not sex-linked traits instead of sex chromosomes and autosomal chromosomes. The response partially explains why females are less likely to inherit a recessive trait carried on the X chromosome, however the explanation of why males are more likely to inherit sex-linked traits is not complete. *Females are less likely to inherit hemophilia because it is a recessive trait on the X chromosome and the chances that they will have a recessive trait on both Xs is very small. Boys, however, only have one X chromosome so they have a greater chance.* The response shows some significant misconceptions related to the inheritance of an autosomal trait being carried on X and Y chromosomes. *Females and males are equally likely to get PKU because it can be carried on the X and Y chromosomes and is not sex-linked.* The response is sometimes clear and sometimes uses appropriate scientific terminology.

### Response E—Anchor Score 1

This response demonstrates a minimal knowledge and understanding of the subject matter. The content of the response is mostly incorrect. The examinee does not correctly describe the difference between sex chromosomes and autosomal chromosomes, nor does the examinee correctly denote that PKU is carried on autosomal chromosomes, though they do correctly state that males do not have 2 X chromosomes. There is little evidence of understanding of scientific concepts. *Why males & females are equally likely to inherit PKU? –because they have  $X^h$  and  $X^{H^1}$  she still has a 50-50 chance but since  $X^h$  is recessive they are just as likely as males ( $X^hY$ ).* Additionally, the prompt clearly states that hemophilia is a recessive disorder, but the examinee incorrectly writes, *but all men need is one  $X^{H^1}Y$  and they will have it (hemophilia).* The response demonstrates little correct use of scientific terminology to communicate understanding.

## Response F—Anchor Score 2

This response demonstrates a basic knowledge and understanding of the subject matter. The content of the response is partially correct. The response does not directly address the first bullet, but despite using the word “cell” for “chromosome” the examinee correctly communicates that females have two X sex chromosomes and males have one X (XY), and that, *In PKU it doesn't matter what your sex is because it isn't transferred or inherited through your X sex cell.* Conceptually, the examinee correctly distinguishes between sex chromosomes and autosomal chromosomes, and provides an adequate, though incomplete, explanation for why females are less likely to inherit hemophilia. *The females have two X sex cells (XX) so they have twice as many chances to get the Dominant trait of not getting Hemophilia but the males sex cells are (XY) so they only have 1/2 as many chances to get hemophilia or not get it.* The examinee correctly states that females and males could both have P and p and not get PKU, which demonstrates some understanding that females and males both have two versions of an autosomal trait. The response is sometimes clear and sometimes uses appropriate scientific terminology.

## Response G—Anchor Score 1

This response demonstrates a minimal knowledge and understanding of the subject matter. The content of the response is mostly incorrect, or a repetition of the prompt material. The examinee does correctly state that males have one X (chromosome) and females have two Xs (chromosomes), but does not provide any conceptual understanding to state why this difference leads to males being more likely to have hemophilia than females, which is simply provided in the prompt material. Additionally, the response demonstrates little or no evidence of understanding of scientific concepts when discussing PKU or the difference between autosomal and sex chromosomes. The PKU statement is primarily a repeat of the prompt material, except for minimal understanding that PKU is not a sex-linked disease. *PKU has equal inheritance because it isn't a sex-linked disease. So both male and female have an equal chance of contracting the disease.* The response demonstrates little correct use of scientific terminology to communicate understanding.