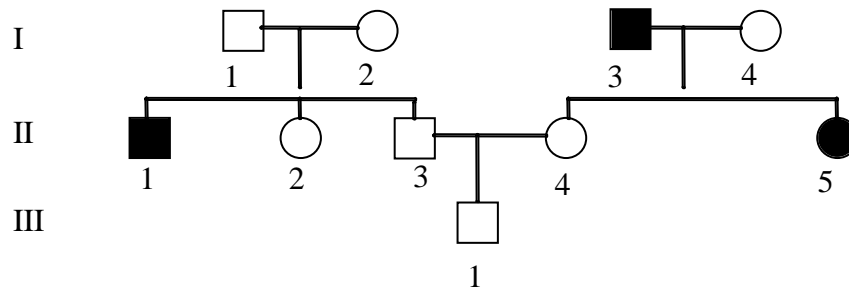


**Appendix A. Examples of homework, quiz and exam questions that address Learning Goal 1.**

**A. Examples of homework questions (Learning Goal 1)**

**1. Shared questions on homeworks for both majors and non majors.**



1. If the pedigree above is for an autosomal recessive characteristic, which individuals are *definitely* heterozygous?

- A. I-1, I-2, I-4, II-2, and II-3 only
- B. I-1, I-2, and I-4 only
- C. I-1 and I-II only
- D. I-1, I-2, I-4, II-4 only
- E. I-1, I-2, I-4, II-4, III-1 only

*Answer: D*

2. Could the characteristics followed in the pedigree be caused by an X-linked recessive allele?

- A. Yes, all individuals fit the X-linked recessive inheritance pattern.
- B. No, the offspring of I-1 and I-2 contradict an X-linked recessive inheritance.
- C. No, the offspring of I-3 and I-4 contradict an X-linked recessive inheritance.
- D. No, the offspring of II-3 and II-4 contradict an X-linked recessive inheritance.

*Answer: A*

3. Menkes Syndrome is a rare X-linked disease that affects copper metabolism. Treatment with copper injections may improve the life of a person with Menkes disease but there is no newborn screening for this disorder. Most children born with this disease die before they turn 10.

Lawrence and Natasha do not have Menkes syndrome. They have 2 sons: Cale and Reid. Cale has Menkes syndrome. Cale inherited the mutation in the MK gene that causes Menkes syndrome from:

- A. His mother
- B. His father
- C. Both parents
- D. Could be either parent- it cannot be determined

*Answer: A*

4. Assuming meiosis is normal, what is the probability that Reid is a carrier for Menkes Syndrome?

- A. 0%
- B. 25%
- C. 50%
- D. 75%
- E. 100%

*Answer: A*

5. Lawrence and Natasha have a daughter named Lilly. Lilly marries Steve. Steve does not have Menkes Syndrome. They have a daughter named Jessica. What is the chance that Jessica is a carrier for Menkes Syndrome?

- A. 1
- B. 1/2
- C. 1/4
- D. 1/16
- E. 0

*Answer: C*

## **2. Majors only homework questions**

1. Sickle cell anemia, Tay-Sachs disease, cystic fibrosis, and phenylketonuria are all inherited in an autosomal recessive manner.

Let's say a man named Jake: Is a carrier for sickle cell anemia; Is a carrier for Tay-Sachs disease; Does not have cystic fibrosis nor is a carrier for cystic fibrosis; Has phenylketonuria

Let's say a woman named Eleanor: Has sickle cell anemia; Is a carrier for Tay-Sachs; Is a carrier for cystic fibrosis; Has phenylketonuria.

If Jake and Eleanor have a baby boy named Tim what is the chance that he will be a carrier for sickle cell anemia, have Tay-Sachs, be a carrier for cystic fibrosis, and have phenylketonuria?

- A. 0
- B. 1/6
- C. 1/8
- D. 1/16
- E. 1/32
- F. 1/56
- G. 1

*Answer: D*

2. You have decided to develop a new designer breed of dog so you mate a male Bull Mastiff with a female Papillon. Later you find out that:

- a. The Bull Mastiff has an autosomal dominant form of blindness. "R" represents the mutant allele of a rhodopsin gene and "r" represents the normal allele of the rhodopsin gene. There only needs to be a mutation in one copy of the rhodopsin gene in order to be blind.
- b. The papillon has an autosomal recessive disease known as Black Hair Follicular Dysplasia. "M" represents the normal allele melanocyte stimulating receptor gene and "m" represents the mutant allele of the melanocyte stimulating receptor gene. There must be a mutation in both

copies of the melanocyte stimulating receptor gene in order to have Black Hair Follicular Dysplasia. If the Bull Mastiff is RrMm, what is the chance that their puppies will be blind and have Black Hair Follicular Dysplasia?

- A. 0%
- B. 25%
- C. 50%
- D. 75%
- E. 100%

*Answer: B*

3. You mate two of the offspring that are not blind and do not have Black Hair Follicular Dysplasia together. What is the chance that the puppies will have Black Hair Follicular Dysplasia?

- A. 0%
- B. 25%
- C. 50%
- D. 75%
- E. 100%

*Answer: B*

4. One of the male puppies mates with his Papillon mother and they have 12 puppies. All of the puppies have Black Hair Follicular Dysplasia. Six puppies are also blind. What was the phenotype of the male puppy?

- A. Black Hair Follicular Dysplasia and blindness
- B. Black Hair Follicular Dysplasia only
- C. Blindness only
- D. Normal

*Answer: A*

5. You are studying fruit flies that have a metabolic disorder. An enzyme assay is available to determine the level of the enzyme in tested flies. Flies that are EE are scored as 1.0 (100% activity), flies that are Ee are scored as 0.5 (50% activity), and flies that are ee are scored as 0.0 (0% activity). Two Ee flies mate and you test the enzyme activity of all of their progeny. Here are the results:

<u># of flies</u>	<u>enzyme activity</u>
29	1.0
68	0.5
23	0.0

For this cross, how many flies do you expect would have an enzyme activity of 0.5?

- A. 70
- B. 60
- C. 50
- D. 40
- E. 30
- F. 20

*Answer: B*

6. What is your Chi-Square statistic and conclusion for this analysis?
- A. 1.64; there is no significant difference between observed and expected values
  - B. 1.64; there is a significant difference between observed and expected values
  - C. 2.73; there is no significant difference between observed and expected values
  - D. 2.73; there is a significant difference between observed and expected values
  - E. 3.12; there is no significant difference between observed and expected values
  - F. 3.12; there is a significant difference between observed and expected values

Answer: C

### 3. Non majors only homework questions

1. Say that Jade is a carrier for phenylketonuria and also a carrier for sickle cell anemia (these genes are on different chromosomes). Her genotype for these two genes is: PpAa. Her husband is a carrier for phenylketonuria, but is not a carrier for the sickle cell anemia allele (and does not have the disease). What is the probability they will have a child who has phenylketonuria and is also a carrier for sickle cell anemia?

- A. 1/16
- B. 1/8
- C. 1/4
- D. 1/2
- E. 2/3
- F. 3/8
- G. 9/16

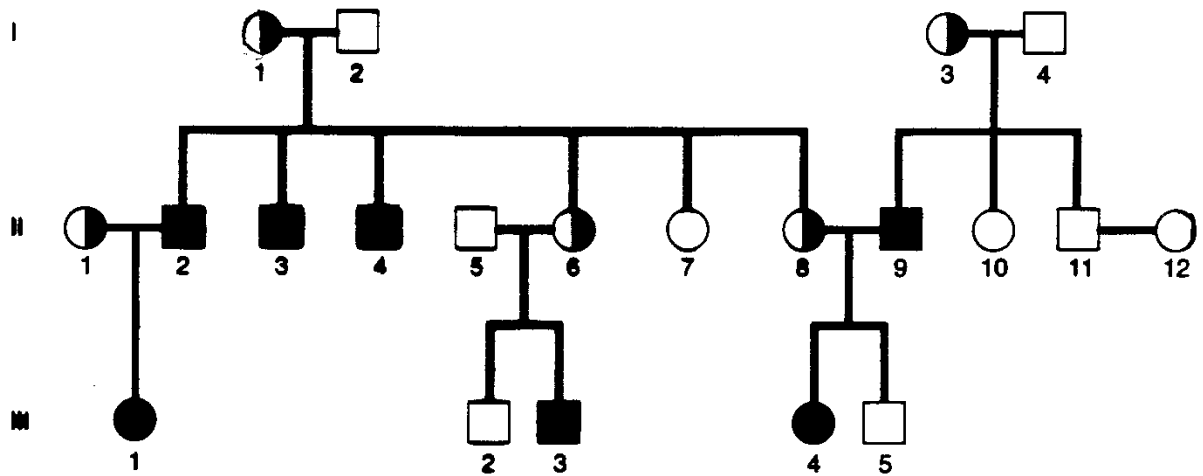
Answer: B

2. What is the most likely mode of inheritance shown in the pedigree below? All carriers are known and indicated.

- A. autosomal recessive
- B. autosomal dominant
- C. x-linked recessive
- D. x-linked dominant
- E. mitochondrial

Answer: C

3. Explain your answer to the above question. *this trait cannot be dominant, otherwise there would not be any carriers. This trait cannot be autosomal recessive, because individuals in the first generation (#2 and #4) are not carriers; thus offspring could not have disease. It can only be X linked recessive.*



4. Amelogenesis imperfecta (call the gene *a*) is X-linked recessive and causes abnormal tooth enamel. Tom has this trait, and Amy is a carrier. With reference only to the X and Y chromosomes, what are Amy's possible gametes? What are Tom's possible gametes? Please list them IN ORDER (Amy's, then Tom's)

Amy: *X<sup>a</sup>* and *X<sup>A</sup>*

Tom: *X<sup>a</sup>* and *Y*

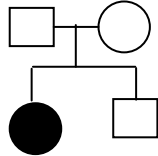
5. If Tom and Amy have a daughter, what is the probability that their daughter will inherit the condition (have defective tooth enamel)?

- A. 0%
- B. 25%
- C. 50%
- D. 75%
- E. 100%

Answer: C

### B. Shared quiz questions (Learning Goal 1)

1. This pedigree shows a family with a rare genetic disease. There could be unmarked carriers, but only those marked actually have the disease. Where is the mutant gene located?



- A. on an autosome
- B. on the X chromosome
- C. could be on either the X chromosome or an autosome

Answer: A

2. Let's say fur color in rabbits is represented by F/f and ear color is represented by E/e. A male rabbit has white fur and pink ears: FfEe. A female rabbit has brown fur and pink ears: ffEE. What is the chance that one of their offspring will have white fur and pink ears?

- A. 0
- B. 1/4
- C. 1/2
- D. 3/4
- E. 1

Answer: C

3. Renpenning Syndrome is an X-linked recessive disease. A woman is a carrier of the mutation for this disease. She marries a man who does not show symptoms of Renpenning Syndrome. They have one son together. What is the probability that the son has Renpenning Syndrome?

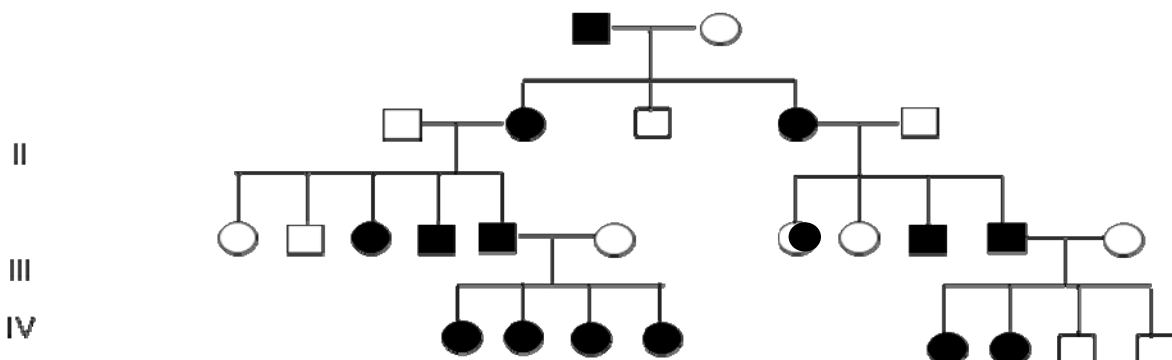
- A. 0
- B. 1/4
- C. 1/2
- D. 3/4
- E. 1

Answer: C

4. What mode of inheritance is **not** possible in the pedigree shown below? There could be carriers that are unmarked.

- A. autosomal recessive
- B. autosomal dominant
- C. x-linked recessive
- D. x-linked dominant
- E. all of the above are possible

Answer: C



5. A man has the X-linked recessive trait of color blindness. A woman is a carrier for the trait (one normal X chromosome and one X chromosome with the color blindness allele). They have a daughter who ends up with only one X chromosome rather than two. The daughter has only one X because the X chromosomes failed to separate properly in meiosis when her mother

produced an egg. The daughter still received an X chromosome from her father. What is the chance that the daughter will have the mutation that causes colorblindness?

- A. 0%
- B. 50% (1/2)
- C. 75% (3/4)
- D. 100% (1)

Answer: D

### C. Shared exam questions (Learning Goal 1)

1. A person heterozygous for an autosomal dominant trait that causes excess body hair has children with a person who has normal body hair. What is the probability that their first offspring will have excess body hair?

- A. 100%
- B. 50%
- C. 25%
- D. 0%

Answer: B

2. Sally and Tom just found out that they are going to have a baby! Sally knows that her mother is a carrier for the autosomal disease Alper's syndrome. Sally's father is not a carrier. There is no history of this disorder in Tom's family, so assume no one is a carrier. Hint: draw the pedigree to answer this question! What is the chance that Sally and Tom's baby will be a carrier for Alper's syndrome?

- A. 0%
- B. 25%
- C. 50%
- D. 75%
- E. 100%

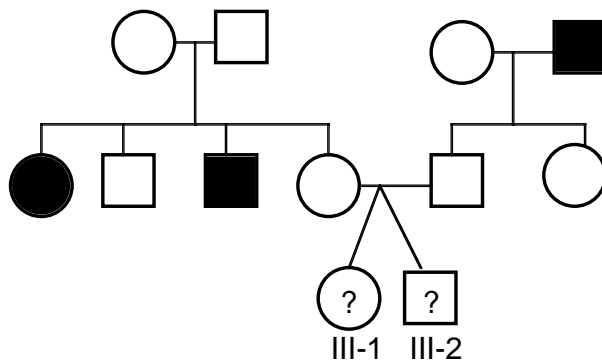
Answer: B

3. Below is a pedigree of a family that has a newly characterized disease. What is the mode of inheritance? There may be carriers that are not marked.

What is the mode of inheritance?

- A. Autosomal recessive
- B. Autosomal dominant
- C. X-linked recessive
- D. X-linked dominant
- E. More than one of the above is possible

Answer: A



4. Hemophilia is an X-linked recessive disease. If a woman who is heterozygous for the disease has kids with a man who has the disease, the probability ratios for their kids' possible phenotypes should be:

A. 3 normal females: 1 hemophiliac male

B. 1 normal female: 1 hemophiliac female: 1 normal male: 1 hemophiliac male

C. All males and females should be normal

D. All males and females should be hemophiliac

*Answer: B*