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BIOLOGY

Mendelian Genetics & Inheritance Patterns

Vocabulary

Click on each word below to go to the definition.

amniocentesis
carpel
carrier
chorionic villus sampling
codominance
consanguineous
cross-pollination
dihybrid
dominant
F_1 generation
F_2 generation
fertilization
fetoscopy
gene therapy
genotype
heredity
heterozygous
homozygous
hybridization
incomplete dominance
inheritance
model organism
monohybrid
pedigree
P generation
phenotype
pleiotropy
polygenic
Punnett square
recessive
segregate
sex-linked
stamen
testcross
trait
trihybrid
true-breeding

Heredity Unit Topics

Click on the topic to go to that section

- **Mendelian Genetics**
- **Punnett Squares**
- **Non-Mendelian Inheritance**
- **Inherited Disorders**
- **Pedigrees**

Mendelian Genetics

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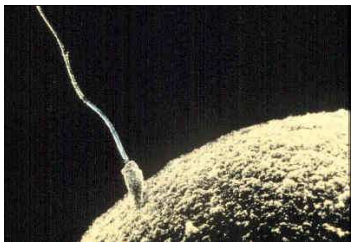
Sexual Reproduction

Most eukaryotes reproduce sexually, fusing two gametes to produce an offspring. The process of fusing gametes is called **fertilization**.

Sexual reproduction increases the genetic variation in a population by creating genetically unique individuals.



Fusion of haploid gametes during fertilization results in a diploid offspring.

Genetic Variation

If sexual reproduction creates genetically unique individuals, why do offspring still resemble their parents?

In 1865, an Austrian monk named Gregor Mendel provided part of the answer to this question when he announced that he worked out the rules of inheritance through a series of experiments on garden peas.

Gregor Mendel

Gregor Mendel was a monk who lived in an Austrian monastery. He had attended the University of Vienna where he studied the natural sciences, as well as physics and mathematics.

The monastery where he lived and worked was devoted to scientific teaching and research.

One important aspect of Mendel's work was the formation of an agricultural society emphasizing the importance of research that would help breeding programs become more efficient.

A Big Question

Think about the following question as we study what Mendel did:

What are the rules of inheritance that Mendel discovered?

Mendel's Experiments

Mendel's experiments with pea plants were aimed at addressing one of the most fundamental issues concerning heredity:

What are the basic patterns of heredity?

Important Words to Know

Heredity: Transmission of traits from parents to offspring.

Trait: Any characteristic of an individual such as height, eye color, etc.

Inheritance: Traits passed from parents to offspring.

Two Prevailing Original Hypotheses

At Mendel's time, there were 2 ideas to explain heredity:

- **Blending Inheritance**: This idea stated that genetic material from the two parents blends together. It was a widespread hypothesis with obvious short-comings.
  ex: a red flower and a white flower will produce a pink flower

- **Inheritance of Acquired Characteristics**: This idea stated that traits present in parents are modified, through use, and passed on to their offspring in the modified form. It was introduced as a theory by French naturalist Jean-Baptiste Lamarck in 1809.
  ex: A giraffe has a long neck because it's ancestors kept stretching their own necks out to reach the leaves in the trees, and the long neck trait was passed on.

Two Prevailing Original Hypotheses

Blending inheritance hypothesis and the inheritance of acquired characteristics hypothesis were supported by some of the greatest scientists of Mendel's time.

But were they correct?

What *are* the basic patterns of inheritance?

Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments. He was certainly not the first scientist to study how traits were inherited, but why was he successful when others failed?

Model Organisms

One of the most important reasons that Mendel was successful is that he chose to study an appropriate **model organism**. A model organism consists of individuals that are usually small, short lived, inexpensive to take care of, produce many offspring in a relatively short period of time, and can be easily manipulated experimentally.

What organisms can act as model organisms?

Mendel's Model Organism -The Pea Plant

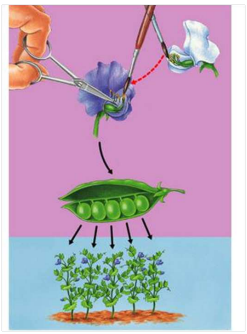
Mendel chose pea plants as his model organism because:

- There are **many varieties** with distinct inherited traits (such as color).
- **Controlled-matings** can be achieved with pea plants.
- Each pea plant has **both reproductive organs** - the pollen-producing organs (stamens) and ovule-producing organ (carpels).
- **Cross-pollination** (fertilization between different plants) can be achieved by dusting one plant with pollen from another

Controlled Breeding of The Pea Plant

In order to precisely control his breedings, Mendel would:

- Cut the stamens off the plant to insure no unwanted sperm would touch the carpels.
- Use a paintbrush to apply only the specific sperm to the carpels.



Mendel's Chosen Phenotypes

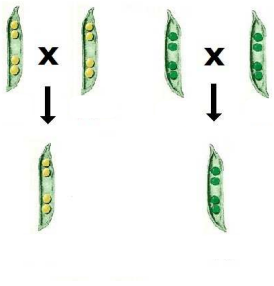
Seed		Flower	Pod		Stem	
Form	Cotyledons	Color	Form	Color	Place	Size
Grey & Round	Yellow	White	Full	Yellow	Axial pods, Flowers along	Long (6-7ft)
White & Wrinkled	Green	Violet	Constricted	Green	Terminal pods, Flowers top	Short (¾ -1ft)
1	2	3	4	5	6	7

Mendel chose to track 7 characteristics that varied in an "either-or" manner. There were two forms for each trait. These observable traits of an individual are known as the **phenotype.**

A Typical Experiment

In a typical experiment, Mendel started with two contrasting, **true-breeding** varieties.

True-breeding individuals will produce offspring identical to themselves when they self-pollinate or crossed to another member of the same population.



1 A plant with purple flowers is allowed to self-pollinate. Generation after generation, it produces purple flowers. This is an example of

- A hybridization.
- B incomplete dominance.
- C true-breeding.
- D the law of segregation.

2 Which hypothesis was published by Jean-Baptiste Lamarck

- A Evolution by Natural Selection
- B Inheritance of Acquired Characteristics
- C Blending Inheritance
- D Endosymbiosis

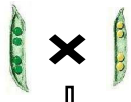
3 Pea plants were particularly well suited for use in Mendel's breeding experiments for all of the following reasons except that

- A peas show easily observed variations in a number of characters, such as pea shape and flower color.
- B it is possible to completely control matings between different pea plants.
- C it is possible to obtain large numbers of progeny from any given cross.
- D peas have an unusually long generation time.

Hybridization

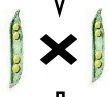
Mendel would take 2 true breeding individuals and cross-breed (hybridize) them.

The true-breeding parents are known as the **P generation.**



P

The hybrid offspring of the P generation are called the **F_1 generation.**



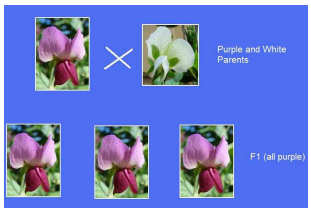
F_1

If F_1 individuals self-pollinate, the **F_2 generation** is produced.

F_2

Experiment # 1: Monohybrid Cross

Mendel crossed a true breeding pea plant having purple flowers with a true breeding pea plant having white flowers.This is called a **monohybrid** cross because the parent plants differ in only one trait, their flower color.



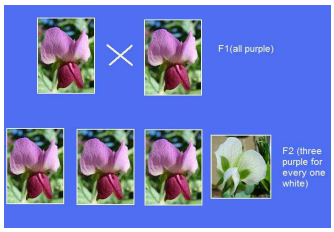
Observation:

All F_1 plants had purple flowers.

Monohybrid Cross

Mendel then mated two F_1 plants and this cross produced 929 F_2 plants.

Observation: 705 of the 929 plants had purple flowers and 224 had white flowers



(75% had purple flowers and 25% had white flowers)

Monohybrid Cross

Based on the results from this experiment, Mendel concluded that the "heritable factor" (now know as a gene) for white flowers did not disappear in the purple F_1 plants but instead that the purple-color factor was controlling flower color in the F_1 plants.

He also concluded that the F_1 plants must have carried two factors for the flower-color character, one for purple and one for white.

Is This a Pattern of Inheritance?

Mendel observed these same patterns of inheritance for the six other traits in pea plants. The F_1 generation all had the same phenotype which came from one of the parents.

In the F_2 generation 3/4 of the population would have the same phenotype as the F_1 generation and 1/4 had the phenotype of the other parent in the P_1 generation.

Mendel's Hypotheses

From these results, Mendel developed four hypotheses regarding inheritance:

Hypothesis # 1: Alleles

There are alternative forms of genes that account for variations in inherited characteristics - meaning there are two different versions of the gene for flower color in pea plants: one for purple and one for white.These alternative forms are now called **alleles**.

Mendel's Hypotheses

Hypothesis # 2: All Organisms have 2 alleles

For each characteristic, an organism inherits two alleles (one from each parent). The two alleles may be the same or they may be different.

An organism that has two identical alleles for a gene is **homozygous** for that gene.

An organism that has two different alleles for a gene is **heterozygous** for that gene.

Mendel's Hypotheses

Hypothesis # 3: Dominant and Recessive

If the two alleles of a pair are different (heterozygous), one determines the appearance and is called the **dominant allele**.

The other allele has no noticeable effect on the appearance and is called the **recessive allele**.

Mendel's Hypotheses

Hypothesis # 4: Law of Segregation

A gamete carries only one allele for each trait because allele pairs separate (segregate) from each other during meiosis. This is known as **The Law of Segregation.**

When sperm and egg unite at fertilization, each contributes its one allele, restoring the pair in the offspring.

Chromosomal Theory of Inheritance

Mendel published his results without knowing much about chromosomes or how they functioned.

It wasn't until about 1900 that biologists began to see parallels between Mendel's "heritable factors" and the behavior of chromosomes.

These observations began to give rise to the **chromosome theory of inheritance.**

Chromosome Theory of Inheritance

In 1903, two scientists, Walter Sutton and Theodor Boveri, independently linked Mendel's rules with the details of meiosis to formulate the **chromosome theory of inheritance.**

The theory states that...

* Meiosis causes the patterns of inheritance observed by Mendel in his pea plants.

* "Hereditary factors" called ***genes*** are located on chromosomes.

Allele Pairs

Remember from our study of mitosis and meiosis, that each diploid cell has two sets of homologous chromosomes.

It is on these homologous chromosomes that the allele pairs for each trait (Mendel's second hypothesis) are found.

4 Mendel referred to heritable characters that were passed from one generation to the next. What are these heritable characters known as today?

- A alleles
- B chromosomes
- C genes
- D heterozygotes

5 A genetic cross in which the parental organisms (P generation) differ in only one character is known as a _____ cross.

- A monohybrid
- B dihybrid
- C self
- D test

6 Alternate versions of a gene are called _____.

- A alleles
- B chromatids
- C heritable factors
- D heterozygotes

7 An organism that has two identical alleles for a gene is said to be _____ for that gene and is called a _____.

- A homozygous; heterozygote
- B homozygous; homozygote
- C heterozygous; homozygote
- D heterozygous; heterozygote

Punnett Squares

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Mendel's Segregation Model

Mendel's segregation model accounts for the 3:1 ratio he observed in the F_2 generation of his numerous crosses.

The possible combinations of sperm and egg can be shown using a **Punnett square**, a diagram for predicting the results of a genetic cross between individuals of known genetic makeup

An uppercase letter represents a dominant allele, and a lowercase letter represents a recessive allele.

P — dominant

p — recessive

Punnett Square

The diagram shows that each parental gamete carried two of the same alleles (for flower color), so the parental gametes in this particular cross are either PP or pp.

As a result of fertilization, the F_1 hybrid offspring each inherited one P and one p; therefore their **genotype** (genetic makeup) for this trait is Pp (heterozygous).

	w	w
W	Ww	Ww
W	Ww	Ww

Heterozygous Phenotype

The dominant allele has its full effect in the heterozygote, while the recessive allele has no effect on flower color in the heterozygote. Therefore, a Pp flower has a purple phenotype.

phenotype

genotype

PP or Pp

8 What color would the F_1 plants be in a cross between a purple (PP) and white (pp) flower?

- A white
- B purple
- C pink
- D 1/2 would be white and 1/2 would be purple

9 Why are all the F_1 plants purple when true breeding purple and white plants are crossed?

- A Purple is dominant to white flower color in pea plants.
- B This is a random event and is all due to chance.
- C The sperm with the allele for white flower color cannot fertilize the egg in pea plants.
- D All pea plants have purple flowers.

F_2 generation

	P	p
P	PP	Pp
p	Pp	pp

Mendel's hypotheses also explained the 3:1 ratio of purple to white flowers he observed in the F_2 generation.

Since the F_1 hybrids are Pp, 1/2 of their gametes will have the P allele and 1/2 will have the p allele.

Looking at Genotype

egg \ sperm	P	p
P	PP	Pp
p	Pp	pp

We can see that if a sperm cell carrying a P allele fertilizes an egg cell carrying a P allele, the offspring will be PP and therefore have purple flowers. This particular genotype will occur in 1/4 of the offspring.

Looking at Genotype

egg \ sperm	P	p
P	PP	Pp
p	Pp	pp

1/2 of the offspring will inherit one P allele and one p allele (Pp).

They will also have purple flowers since purple is dominant to white. (Typically, any color is dominant to white)

Looking at Genotype

egg \ sperm	P	p
P	PP	Pp
p	Pp	pp

The remaining 1/4 of the F_2 plants will inherit a p allele from each parent, giving it a genotype of pp and a phenotype of white flowers.

Genotype vs. Phenotype

Because an organism's appearance does not always reveal its genetic composition, we need to distinguish between an organism's expressed, or physical traits (**phenotype**), and its genetic makeup (**genotype**).

In Mendel's pea plants, flower color phenotype was purple or white

but there are three different genotypes PP, Pp, pp

Genotype vs. Phenotype

In this flower color example, *PP* and *Pp* plants have the same phenotype but different genotypes.

phenotype

genotype

PP or Pp

Genotype vs. Phenotype

Complete the chart below for the flowers we just discussed.

Genotype	Phenotype
	Purple
	Purple
pp	

10 Red is dominant over white in this plant. In reference to the parent plants below, Parent Plant A has ____flowers.

- A red
- B white

flower genotype	
parent plant A Rr	parent plant B RR

11 Red is dominant over white in this plant. In reference to the parent plants below, Parent Plant B has ____ flowers.

- A red
- B white

flower genotype	
parent plant A Rr	parent plant B RR

12 The plants below can produce a plant with red flowers.

- True
- False

flower genotype	
parent plant A Rr	parent plant B RR

13 The probability that the parent plants A and B will produce a plant with red flowers is _____%.

flower genotype	
parent plant A Rr	parent plant B RR

14 The probability that the parent plants A and B will produce a plant with white flowers is _____ %.

flower genotype	
parent plant A Rr	parent plant B RR

Determining Genotype from Phenotype

How can we tell the genotype of an individual with the dominant phenotype? In Mendel's pea plants those with PP or Pp both appeared purple.

So if we have a pea plant with purple flowers, how do we determine which genotype the plant has?

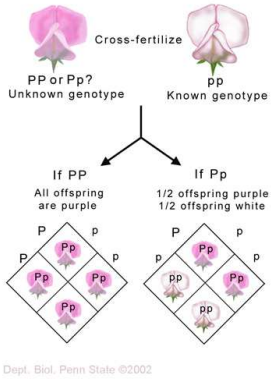
PP ?

Pp ?

Testcross

A **testcross** involves breeding the individual whose genotype we are trying to determine with a homozygous recessive individual.

If any offspring display the recessive phenotype, the parent must be heterozygous.



15 What is the ratio of purple to white flowers Mendel observed in the F_2 generation when he crossed two F_1 hybrids?

- A 1:1
- B 2:1
- C 3:1
- D 4:1

16 An organism's expressed, or physical, traits is known as its:

- A genome
- B genotype
- C phenome
- D phenotype

17 Crossing an individual that is homozygous recessive with an organism of unknown genotype that exhibits a dominant phenotype is known as a _____.

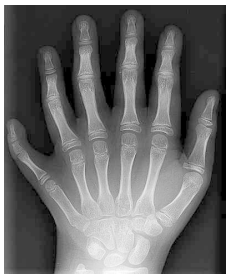
- A hybrid cross.
- B heterozygous cross.
- C test cross.
- D unknown cross.

Dominant ≠ Common

Just because an allele is dominant, does not mean that it is more common than its recessive counterpart in a population.

For example, 1 out of 400 babies in the United States is born with extra fingers or toes (polydactyl). The allele for this unusual trait is dominant to the allele for the more common trait of five digits per appendage.

In this example, the recessive allele is far more prevalent than the dominant allele in the population.



18 Whether an allele is dominant or recessive depends on:

- A how common the allele is, relative to other allleles.
- B whether it is inherited from the mother or the father.
- C whether it or another allele determines the phenotype when both are present.
- D which chromosome it is on.

Law of Segregation... and Beyond

Remember Mendel's 4th Hypothesis?

Law of Segregation: A sperm or egg carries only one allele for each trait because allele pairs separate from each other during gamete formation.

Mendel did not stop there...

He devised a second law of inheritance by observing two traits at a time (e.g. seed color and seed shape.) Mendel wanted to see if these different traits affected each other or not, in other words, did they behave independently of each other or not?

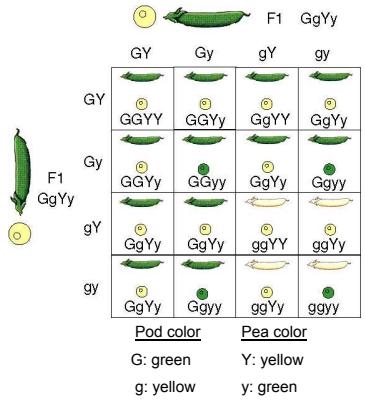
Dihybrid Cross

Mendel performed **dihybrid crosses**, crossing true-breeding parents differing in two characteristics to determine whether the characteristics are transmitted to the offspring together or independently. For example a pea plant with yellow pods and yellow seeds (ggYY) crossed with a pea plant with green pods and green seeds (GGyy).

Such a cross produces an F_1 generation which is heterozygous for both characteristics (GgYy).

Dihybrid Cross

Crossing the F_1 generation produced a 9:3:3:1 ratio of phenotypes and a genotypic ratio of 1:2:2:4:1:2:1:2:1.



The Law of Independent Assortment

Using a dihybrid cross, Mendel developed his second law of inheritance.

The law of independent assortment states that each pair of alleles segregates independently of other pairs of alleles during gamete formation.

This law of independent assortment applies only to genes on different, nonhomologous chromosomes. Genes located near each other on the same chromosome tend to be inherited together.

The Law of Independent Assortment

The alleles of one gene segregate into gametes independently of another gene's alleles.

Think of this like a coin toss:
If you toss a coin, what is the probability of it coming up heads?

Now, if you toss a second coin, what is the probability of it coming up heads?

When tossing a coin, the outcome of one toss has no impact on the outcome of the next toss!

Rules of Probability

Mendel's laws of segregation and independent assortment reflect the rules of probability.

Remember from math.....

The probability scale ranges from 0 to 1.
An event that is certain to occur has a probability of 1.
An event that is certain not to occur has a probability of 0.

The probabilities of all possible outcomes for an event must add up to 1.

Using Probability Rules When Solving Genetics Problems

There are two rules of probability which are helpful when solving problems in genetics:

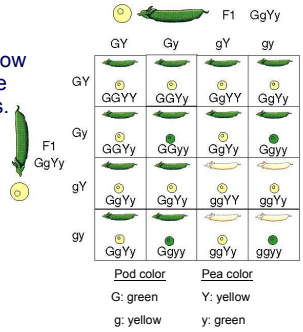
The Multiplication Rule

The Addition Rule

The Multiplication Rule

The **multiplication rule** states that the probability that two or more independent events will occur together is the product of their individual probabilities

For example: You would use the multiplication rule if you want to know the probability that a plant will have *both* yellow seeds and yellow pods.



Pea color - Yy x Yy = 75% yellow

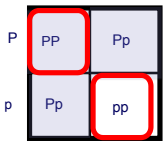
Pod color - Gg x Gg = 25% yellow

0.75 x 0.25 = 0.1875 = 3/16

The Rule of Addition

The rule of addition states the probability that an event can occur in two or more alternative ways is the sum of the separate probabilities of the different ways.

For example: You would use the addition rule if you want to know the probability of a plant (produced from a monohybrid cross) being homozygous dominant for flower color or homozygous recessive for flower color.



25% PP + 25% pp = 50% homozygous

19 If we toss two coins at the same time, what is the chance that both coins will land heads up?

20 In an F_1 cross between two pea plants that are heterozygous (Pp) for purple flowers and heterozygous for pea color (Gg), what is the probability that a particular offspring of this cross will have the recessive genotype for both traits (white flowers, yellow seeds)?

21 Mendel stated that each pair of alleles segregates independently of other pairs of alleles during gamete formation.
What is this law known as?

- A Law of Segregation.
- B Law of Independent Assortment.
- C Law of Probability.
- D Law of Pea Plant Genetics.

22 An organism heterozygous for two characteristics is a _____.

- A dihybrid
- B monohybrid
- C homozygote
- D double dominant

Trihybrid Crosses

We can use the rules of probability to solve complex genetics problems.

If we crossed two organisms both having the genotype AaBbCc, what is the probability that an offspring of this cross will have the genotype aabbcc? Since each allele pair assorts independently, we can treat this trihybrid cross as three separate monohybrid crosses:

Aa X Aa: Probability of aa offspring = _____

Bb X Bb: Probability of bb offspring = _____

Cc X Cc: Probability of cc offspring = _____

aabbcc

Because the segregation of each allele pair is an independent event, we can use the multiplication rule to calculate the probability of an offspring being aabbcc.

What is the probability?

23 If you cross two F_1 individuals both having the genotype AaBbCc, what is the probability that an F_2 offspring will have the genotype AabbCc?

24 A plant with genotype AABbCC is crossed with a plant having
genotype AaBbCc. What is the probability of an offspring
having the genotype AABBCC?

Non-Mendelian Inheritance

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Not Always an Easy Relationship

The relationship between genotype and phenotype is rarely as simple as in the pea plant characters Mendel studied.

Many heritable characteristics are not determined by only one gene with two alleles; however, the basic principles of segregation and independent assortment still apply even to more complex patterns of inheritance.

When Mendel Doesn't Work

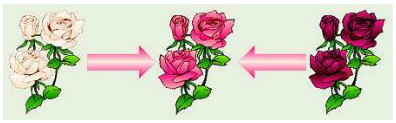
Inheritance of characters for a single gene may deviate from simple Mendelian patterns in the following 3 situations:

1. When alleles are not completely dominant or recessive.
2. When a gene has more than two alleles.
3. When a gene produces multiple phenotypes.

Complete vs. Incomplete Dominance

What we have seen until now when studying Mendel's work is **complete dominance.** This occurs when the phenotypes of the heterozygote (Pp) and dominant homozygote (PP) are identical.

In **incomplete dominance**, the phenotype of F_1 hybrids is somewhere between the phenotypes of the two parental varieties.



Blending vs. Incomplete Dominance

This third phenotype results from the heterozygote having less red pigment than the red homozygote.

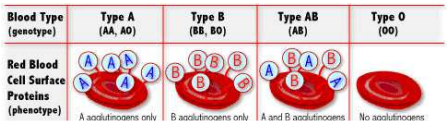
This is NOT blending. If blending occurred you would never be able to retrieve the red and white traits from the pink hybrids.

25 In snapdragons, heterozygotes have pink flowers whereas homozygotes have red or white flowers. When plants with red flowers are crossed with plants with white flowers what percentage of the offspring will have pink flowers?

Codominance

In **codominance**, two dominant alleles affect the phenotype in separate, distinguishable ways. One of the best examples of this is the ABO blood group phenotypes in humans. This involves a single gene with three different alleles. (Therefore, this is a trait that also has multiple alleles).

These three alleles, in different combinations, produce four phenotypes.



The Relationship Between Dominant and Recessive Alleles

So what is the relationship between dominant and recessive alleles?

A dominant allele does not subdue a recessive allele; alleles don't interact.

Alleles are simply variations in a gene's nucleotide sequence.

For any characteristic, the dominance or recessiveness in relationships of alleles depend on the level at which we examine the phenotype.

Pleiotropy

Pleiotropy is a situation in which a single gene may affect many phenotypic characteristics.

Pleio = more than one

-tropy = affecting or pertaining to

An example of pleiotropy in humans in **sickle cell disease**.

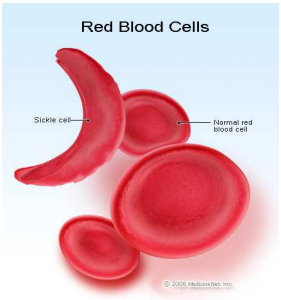
Sickle Cell Disease

This is a disease characterized by many diverse symptoms such as:

Abnormal red blood cells leading to physical weakness and anemia.

Clumping of cells and clogging of small blood vessels leading to pain and fever, possible brain damage, and possible heart failure.

Accumulation in the spleen of abnormally shaped red blood cells leading to spleen damage.



Sickle Cell Disease

All of the possible phenotypic effects of sickle cell disease are caused by the presence of a single allele when it is present on both homologous chromosomes.

The direct effect of sickle cell allele is to make red blood cells produce abnormal hemoglobin molecules, which tend to link together and crystallize.

This, in turn, leads to abnormally shaped red blood cells, which take on a crescent shape.

26 People who carry the sickle-cell trait

- A are heterozygous for the sickle-cell allele.
- B are usually healthy.
- C produce normal and abnormal hemoglobin.
- D all of the above

27 In sickle cell disease, which of the following is the direct effect of the sickle cell allele?

A abnormally shaped red blood cells

B accumulation of sickled cells in the spleen

C red blood cells produce abnormal hemoglobin molecules

D heart failure or brain damage

Polygenic Inheritance

Another variation from Mendel's laws is that a single character may be influenced by many genes. Remember, Mendel studied characters that were classified as "either-or", such as purple or white flowers. Mendel's pea plants were either tall or short, there was no in-between.

It does not always work that way in genetics. Many characteristics vary over a large scale in a population and are determined by many genes.

poly = many

genic = genes

Now, if you look around the room, what do you notice? Can you name examples of polygenic inheritance?

Human Height

What governs height in humans is actually the additive effects of two or more genes on a single phenotypic characteristic (in this case height).

Polygenic inheritance is the opposite of pleiotropy, in which a single gene affects several characteristics.

Skin Color in Humans

Skin color in humans is controlled by *at least* three genes that are inherited separately.

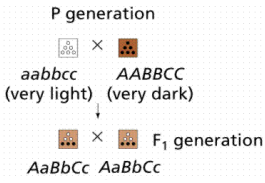
For simplicity, we will stick with three genes in our example and these three genes A, B, and C. Each of these genes has two alleles (a "dark skin" allele and another allele.)

The "dark skin" allele for each gene (A,B, or C) contributes one "unit" of darkness to the phenotype and is incompletely dominant to the other allele (a, b, or c).

Skin Color in Humans

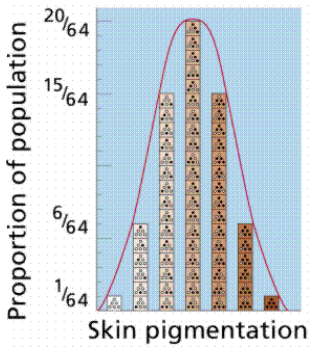
A person who is genotype AABBCC would be very dark while a person with genotype aabbcc would be very light.

A heterozygote (AaBbCc) would have a shade of skin that is intermediate to the first two.



Since these alleles have an additive effect on the phenotype, a person who is AaBbCc would have the same skin color as any other genotype that has only three dark-skin alleles (for example: AABbcc).

Skin Color in Humans



Skin-Color Phenotypes

In the last example we saw how three different genes could produce seven different skin-color phenotypes in humans.

However, if we look at a real human population, we might see many more than just seven shades of skin color. Why is this?

Heredity and Environment

Many characteristics result from a combination of heredity and environment.

From the previous example, skin color in humans is determined not only by the specific allele combination of three genes, but also by exposure to the sun.

Another example is seen in trees. A tree's phenotype is genetically determined but its environment will influence the phenotype as well. For example, exposure to wind and sun will affect leaf size, shape and color.

28 Hydrangea plants of the same genotype are planted in a large flower garden. Some of the plants produce blue flowers and others pink flowers. This can be best explained by:

- A environmental factors such as soil pH.
- B the allele for blue hydrangea being completely dominant.
- C the alleles being codominant.
- D the fact that a mutation has occurred.

29 Which of the following is an example of polygenic inheritance?

- A pink flowers in snapdragons
- B the ABO blood groups in humans
- C Huntington's disease in humans
- D skin pigmentation in humans

Inherited Disorders

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Inherited Disorders

We have seen how errors in transcription and translation could lead to mutations. Here we will see how these may be passed on to offspring.

Genetic disorders can exist in every type of plant and animal. There are thousands of genetic disorders in humans alone.

Some of these disorders are inherited as dominant or recessive traits controlled by a single gene. All of these disorders show simple inheritance patterns just like the traits in Mendel's pea plants.

Recessive Disorders

Many genetic disorders are inherited in a recessive manner.

Recessively inherited disorders show up only in individuals homozygous for the allele. **Carriers** are heterozygous individuals who carry the recessive allele but are phenotypically normal

Recessively Inherited Disorders

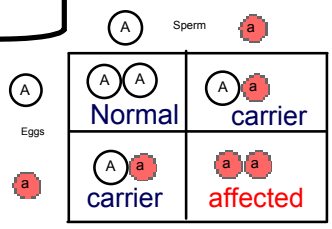
These recessively inherited disorders range in severity from relatively mild to life threatening. An example of a recessive disorder that is relatively mild is **albinism**, in which there is a lack of pigmentation in the skin.

An example of a life-threatening disorder is cystic fibrosis, which involves excessive secretion of a very thick mucus from the lungs, pancreas and other organs.

Aa (X) Aa

Using Mendel's laws, we can predict the percentage of affected children likely to result from a mating between two carriers.

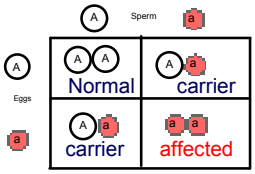
Parents: Normal Aa X Normal Aa



Genotypic and Phenotypic Ratios

Genotypically, 1/4 of the offspring have the chance of exhibiting the particular disorder. 1/2 of the offspring will be carriers for the trait, while 1/4 will be normal. (Ratio of 1:2:1)

Phenotypically, there is a 3/4 chance that each child will not exhibit the disorder. (Ratio of 3:1)



Remember, this is the chance for each baby. This does not mean for all the children in the family.

Inbreeding

Matings between relatives can increase the probability of the appearance of a genetic disease. Such matings are called **consanguineous** matings (inbreeding).

con = with
sangui = blood
-ous = pertaining to

The Fugates of Kentucky

Methemoglobinemia is a recessive disorder characterized by the presence of a higher than normal level of a chemical that blocks oxygen absorption to red blood cells, causing tissues to turn blue.

The Fugates of Hazard, Kentucky were a group of people who carried the gene for this recessive disorder. Additionally, they had numerous consanguineous marriages, resulting many children who were tinted blue!

Examples of Recessive Disorders

Albinism: Lack of pigmentation in skin, hair, and eyes.

Cystic fibrosis: Excessive mucus in lungs, digestive tract and liver. Increases susceptibility to infections. Death occurs in early childhood unless treated.

Phenylketonuria (PKU): Accumulation of the amino acid phenylalanine in blood, which can lead to developmental and/or intellectual disabilities.

Sickle cell disease: Abnormal hemoglobin leads to abnormally shaped red blood cells. Can damage many tissues.

Tay-Sachs Disease: Accumulation of lipid (fat) in brain cells leading to developmental and/or intellectual disabilities, blindness and death in early childhood.

Dominant Disorders

Some human disorders are due to dominant alleles.

One example is **achondroplasia**, a form of dwarfism that is lethal when homozygous for the dominant allele.

The late actor David Rappaport lived with achondroplasia.

Huntington's Disease

Huntington's disease is a dominant degenerative disease of the nervous system.

The disease has no obvious phenotypic effects until about 35 to 40 years of age. This is beneficial for the genes, as by that point, most people with the disease will have unknowingly passed the genes on to their offspring.

30 In order for a child to inherit and exhibit a recessive disorder, both parents must be carriers of the recessive allele.

- True
- False

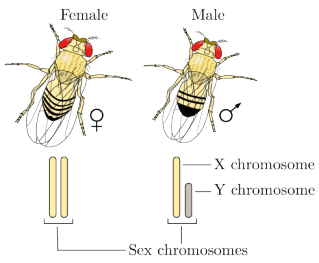
31 Huntington's disease is caused by a dominant allele. If one of your parents has the disease, what is the probability that you, too, will have the disease?

- A 1
- B 3/4
- C 1/2
- D 1/4

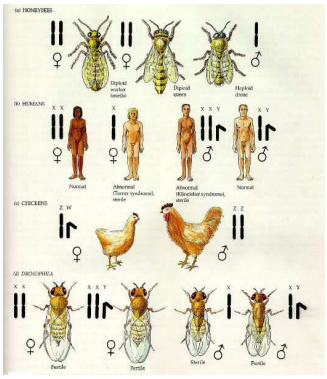
Sex Determination

In humans and other animals, as well as some plants, there is a chromosomal basis of sex determination. An organism's sex is an inherited phenotypic character determined by the presence or absence of certain chromosomes.

There are two chromosomal sex determination systems, XY and ZW.



Sex Determination in Various Species



Sex-linked Genes

The sex chromosomes have genes for many characteristics unrelated to sex. A gene located on eithersex chromosome is called a **sex-linked gene**.

Sex-linked genes follow specific patterns of inheritance. Many sex-linked disorders are located on the X chromosome. Typically these disorders are inherited through the mother, as she gives an X chromosome to any child she has. Sex-linked disorders show up in males more often than females because they only have 1 copy of an X chromosome.

X-linked Disorders

Some disorders caused by recessive alleles on the X chromosome in humans:

Color blindness

Duchenne muscular dystrophy

Hemophilia

Male pattern baldness

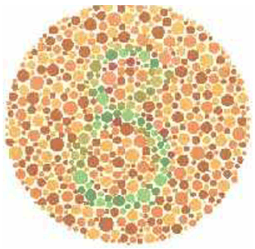
Sex-linked
H = normal & h = hemophilia
Cross: XX^h x X^hY

	X	X^h
X^h	X^hX	X^hX^h
Y	XY	X^hY

Genotypic ratio: 1:1:1:1
(X^hX =25% X^hX^h=25% XY=25% X^hY=25%)

Phenotypic ratio: 1:1:1:1
Female carrier =25% Female hemophilia =25%
Male normal =25% Male hemophilia =25%

32 What number is inside the circle?



33 X-linked disorders show up more often in males because in terms of sex chromosomes:

- A they are likely to inherited the recessive allele from their f
- B they only inherit one X-chromosome
- C the Y-chromosome from their mother carries the recessiv
- D the father can pass the damaged allele on

34 In reference to the parental genotypes below, _____% of children are likely to be male.

- A 100
- B 25
- C 50
- D cannot be determined

father	mother
XY	X^hX

35 In reference to the parental genotypes below, _____% of children are likely to be female.

- A 100
- B 50
- C 25
- D cannot be determined

father	mother
XY	X^hX

36 In reference to the parental genotypes below, there is _____ likelihood that a child will have a hemophilia phenotype.

- A no
- B some

father	mother
XY	X^hX

37 In reference to the parental genotypes below, there is _____ likelihood that a female child will have a hemophilia phenotype.

- A no
- B some

father	mother
XY	X^hX

38 In reference to the parental genotypes below, there is _____ likelihood that a male child will have a hemophilia phenotype.

- A no
- B some

father	mother
XY	X^hX

39 The likelihood that a male child with hemophilia will be born is _____ %.

father	mother
XY	X^hX

40 An X-linked recessive trait cannot go from father to son.

- True
- False

Genetic Counselors

Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease.

Using family histories, genetic counselors help couples determine the odds that their children will have genetic disorders. For a growing number of diseases, tests are available that identify carriers and help define the odds more accurately.

Fetal Testing

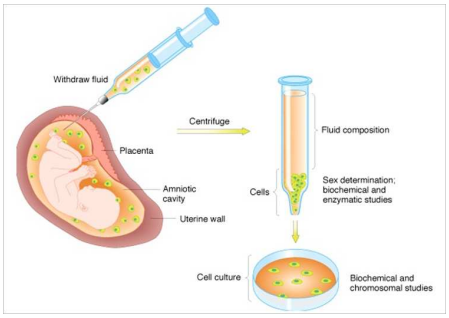
There are several technologies available that can provide insight into potential genetic defects.

Parents can be tested to see if they are carriers of a certain allele.

Amniocentesis

Amniocentesis is a procedure performed between weeks 14 and 20 of pregnancy in which a needle is inserted into the mother's uterus and some amniotic fluid is extracted.

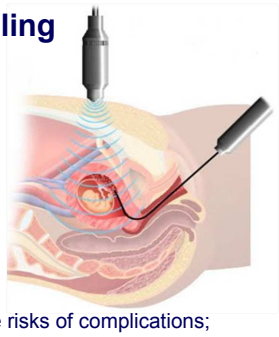
Fetal cells found within this fluid can be tested for genetic disorders.



Click here to see an video of amniocentesis

Chorionic Villus Sampling

In **chorionic villus sampling** (CVS), a sample of the placenta is removed and tested. Results can be obtained within 24 hours; therefore, much quicker than amniocentesis. Another advantage is that it can be performed as early as week 8 of pregnancy.



Both amniocentesis and CVS pose some risks of complications; therefore, they are generally only done in situations where the risk of genetic disorder is relatively high.

Ultrasound and Fetoscopy

Other techniques, such as **ultrasound** and **fetoscopy**, allow a doctor to examine a fetus directly for anatomical deformities *in utero*. Ultrasound imaging uses sound waves to produce a picture of the fetus. The procedure is non-invasive and has no known risks.

Fetoscopy involves the insertion of a long, thin tube containing a scope and fiber optics into the uterus. It can provide highly detailed images of the fetus but does have some risk involved. Fetoscopy can also be used to do fetal surgery *in utero*.

Click here to see an video of fetoscopy

Common Genetic Tests

Some genetic disorders can be detected at birth by simple tests that are now routinely performed in most hospitals in the United States.

One of the most common tests is PKU screening. **Phenylketonuria** is a recessively inherited disorder that occurs in about one out of every 10,000 births in the United States. Children with this disorder cannot properly break down the amino acid **phenylalanine**, which may lead to developmental and/or intellectual disabilities.

If this is detected in a newborn, the baby is placed on a special diet low in phenylalanine and this will usually prevent developmental disabilities.

41 Which of the following techniques involves the preparation of a karyotype?

- A amniocentesis
- B chorionic villus sampling
- C fetoscopy
- D A and B only

42 Which prenatal test allows a doctor to remove a sample from the placenta?

- A amniocentesis
- B fetoscopy
- C chorionic villus sampling
- D ultrasound

43 Which technique is not known to have any associated risks?

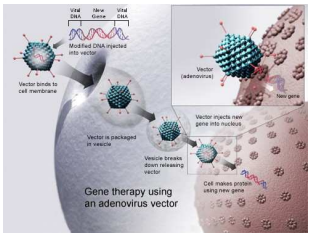
- A amniocentesis.
- B chorionic villus sampling.
- C ultrasound.
- D fetoscopy.

Biotech: Gene Therapy

Gene therapy is a technique for correcting defective genes responsible for disease development.

It is a unique procedure because it can cure ailments at the genetic level, rather than just treat the symptoms of the ailment.

In gene therapy, therapeutic DNA is placed inside a body via a vector. Once inside, the DNA is expressed by the body, producing proteins that treat the targeted disease.



Pedigrees

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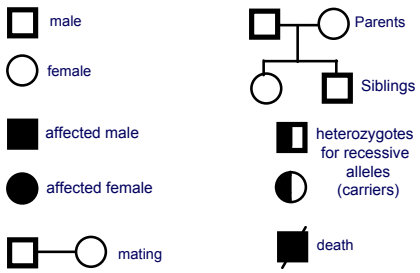
Pedigrees

One way geneticists can determine if offspring are going to inherit a genetic trait or a genetic disease is by creating a **pedigree** and analyzing the patterns.

A pedigree is similar to a family tree except that it also shows whether each person in the tree possess a specific trait, is a carrier for the trait, or does not possess the alleles for the trait.

Important Symbols



Autosomal Traits

If the trait being looked at is an autosomal trait then both the males and females can exhibit the trait.

If the trait is **autosomal dominant** then every individual with the phenotype for that trait will have a parent with the phenotype for that trait.

If the trait is **autosomal recessive** then the individual with the phenotype for that trait can have one, two, or neither parent exhibit the phenotype for that trait.

Heterozygous vs. Homozygous

If an individual has a homozygous dominant or heterozygous genotype, then they will exhibit the dominant phenotype.

If an individual is homozygous recessive, then they will exhibit the recessive phenotype.

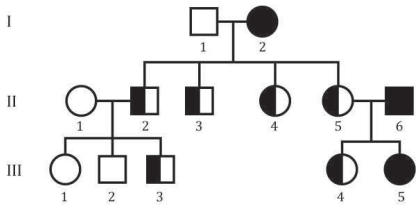
If two individuals are heterozygous for a recessive trait, they will not show the trait phenotypically but they can produce offspring that are homozygous recessive and do exhibit the trait.

44 Which of the following terms is used interchangeably with the term *trait*?

- A genotype
- B allele
- C phenotype
- D gene

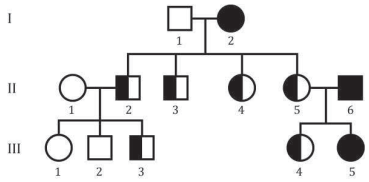
45 Referring to the pedigree chart below, in generation I, which individual is affected?

- A 1
- B 2



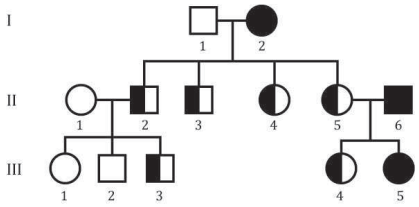
46 Referring to the pedigree chart below, what were the results of the mating of generation I?

- A 1 unaffected female, 2 carrier females, 2 carrier males, 1 affected male
- B 2 carrier males, 2 carrier females
- C 1 unaffected, 5 affected
- D all affected



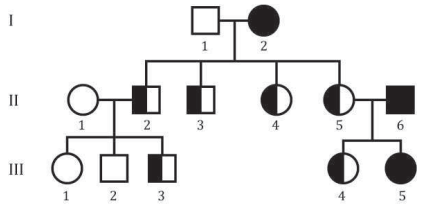
47 Referring to the pedigree chart below, in generation III, who is a carrier for the allele?

- A 1,2,3,4,5
- B 3,4,5
- C 3,4
- D only individual 5



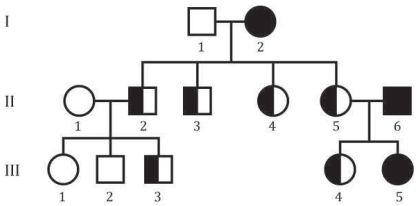
48 Referring to the pedigree chart below, in generation II, who exhibits the phenotype?

- A 1 only
- B 2,3,4,5
- C 6 only
- D 2,3,4,5,6



49 Referring to the pedigree chart below, in generation III, who exhibits the phenotype?

- A 1,2,3
- B 3,4,5
- C only 5
- D 1,2

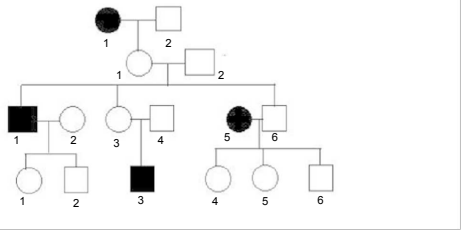


Autosomal Recessive Trait

Not all pedigree charts show the heterozygous genotype. If only the individuals who express the trait are indicated, you must infer (based on expression of the trait in later generations) who is a carrier.

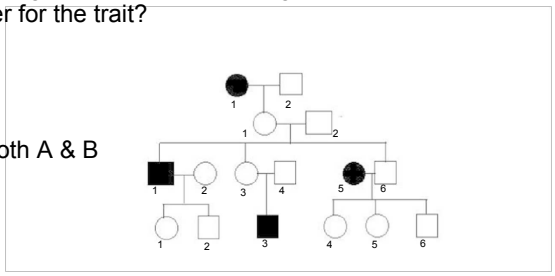
50 Referring to the chart below, in generation I, who exhibits the phenotype?

- A 1
- B 2



51 Referring to the chart below, in generation II, who is a carrier for the trait?

- A 1
- B 2
- C Both A & B



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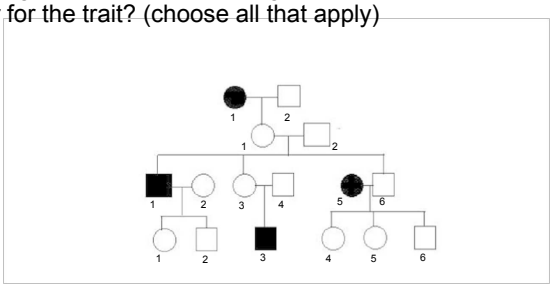
52 Referring to the chart below, in generation III, who is a carrier for the trait? (choose all that apply)

- ☐ A 1
- ☐ B 2
- ☐ C 3
- ☐ D 4
- ☐ E 5
- ☐ F 6



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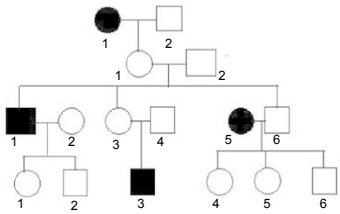
53 Referring to the chart below, in generation IV, who is a carrier for the allele? (choose all that apply)

- ☐ A 1
- ☐ B 2
- ☐ C 3
- ☐ D 4
- ☐ E 5
- ☐ F 6



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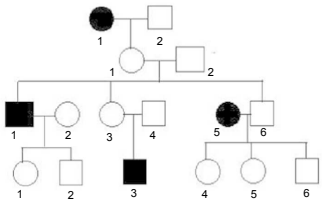
54 Referring to the chart below, in generation II, who exhibits the phenotype?

- ○ A both
- ○ B none



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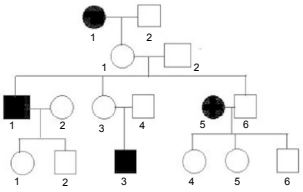
55 Referring to the chart below, in generation III, who exhibits the phenotype? (choose all that apply)

- ☐ A 1
- ☐ B 2
- ☐ C 3
- ☐ D 4
- ☐ E 5
- ☐ F 6



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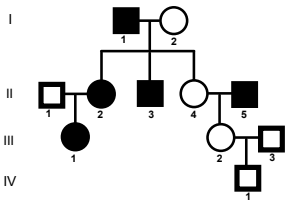
56 Referring to the autosomal dominant pedigree chart below, in generation 1 who exhibits the phenotype? (choose all that apply)

- ☐ A 1
- ☐ B 2



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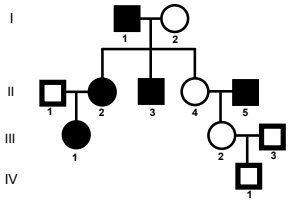
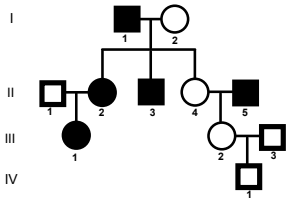
57 Referring to the autosomal dominant pedigree chart below, who is heterozygous for the allele? (choose all that apply)

- ☐ A I-1
- ☐ B II-2
- ☐ C II-3
- ☐ D II-5
- ☐ E III-1



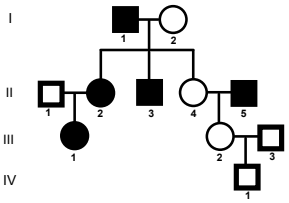
58 Does the individual in generation 4 carry the allele?

- ○ Yes
- ○ No



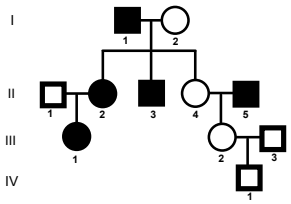
59 Referring to the autosomal dominant pedigree chart below, In generation 2 who exhibits the phenotype? (choose all that apply)

- ☐ A 1
- ☐ B 2
- ☐ C 3
- ☐ D 4
- ☐ E 5



60 Referring to the autosomal dominant pedigree chart below, In generation 3 who exhibits the phenotype? (choose all that apply)

- ☐ A 1
- ☐ B 2
- ☐ C 3



61 If an individual does not exhibit a recessive trait, but one of their parents does, you can assume:

- ○ A the individual is a carrier
- ○ B the individual is not a carrier
- ○ C the individual did not get the allele
- ○ D you can assume nothing without knowing the phenotype of the other parent

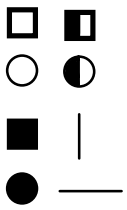
62 When analyzing a pedigree, two individuals who do NOT exhibit a recessive trait have a child who does exhibit the trait. With this information you can assume:

- ○ A one parent was a carrier
- ○ B both parents were carriers
- ○ C the child will not pass the trait on
- ○ D a mutation must have occurred

Practice Pedigree

Problem: Progressive retinal atrophy (PRA) causes blindness in many breeds of dog. This trait is autosomal recessive.

A breeder has a female golden retriever whose mother had PRA. The dog's father was not a carrier for the trait. The breeder mates the dog with an unaffected male. They have a litter of 6 puppies, 4 males, 2 females. Draw a pedigree for the 3 generations of dogs, using all possible genotypes from the mating.



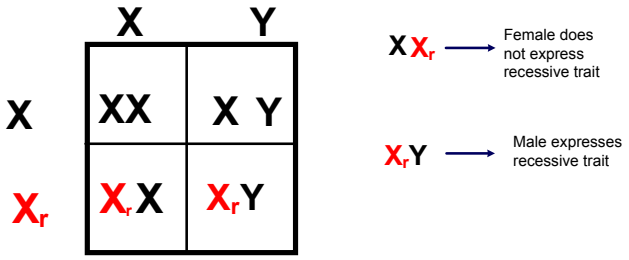
Sex-Linked Trait

If a trait is sex-linked it is usually only seen in males because most sex-linked traits are recessive and located on the X chromosome. Therefore a male receiving an X chromosome with the recessive allele will exhibit the trait.

A female receiving one X chromosome with the recessive allele is a carrier but will not exhibit the trait because she also has a X chromosome with the dominant allele.

A female will only exhibit the trait if she receives two X chromosomes with the recessive allele. If a sex-linked trait is carried on the Y chromosome, only males can exhibit the trait. This is pretty rare.

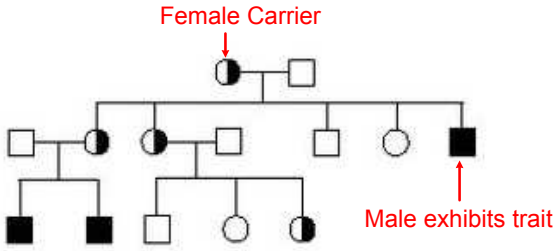
Sex-Linked Trait



63 Most sex-linked traits are recessive and located on the ____ chromosome, which means they are usually only exhibited by

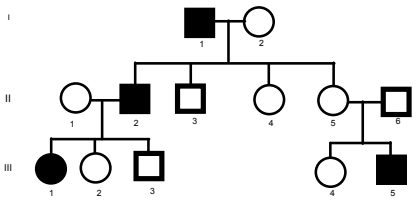
- A X, females
- B X, males
- C Y, females
- D Y, males

Pedigree Chart: X-Linked Recessive Trait



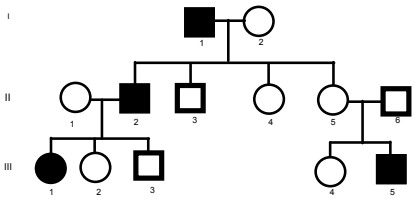
64 In the X-linked recessive trait pedigree below, what is the genotype of female I-2?

- A homozygous recessive
- B heterozygous
- C homozygous dominant
- D more information is needed



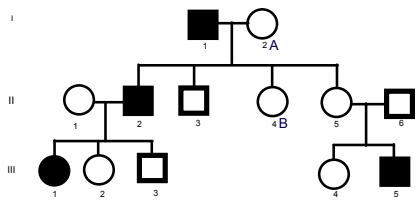
65 If female II-4 were to mate with an individual who does not carry the X-linked trait what is the probability their son would exhibit the trait?

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



66 If female 4B were to mate with an individual who does not carry the X-linked trait, what is the probability their daughter would be heterozygous?

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



67 Hemophilia is a rare genetic disorder that results in abnormal blood clotting and has afflicted many members of the royal family in Russia and England. The pedigree chart below traces inheritance of hemophilia in the famous Romanov family of Russia. What inheritance pattern does hemophilia appear to follow?

- A auto. dominant
- B auto. recessive
- C X-linked recessive
- D Y-linked recessive