

# Analysis of Human Traits

## HASPI Medical Biology Lab 22b

### Background/Introduction



### Using Statistics and Probability to Analyze Genetic Traits

The occurrence of natural selection is necessary in order for evolution to occur. Natural selection requires variation in genetic information as well as variation in how that information is expressed. For example, if an isolated human population had only one trait--blonde hair--every individual in that population will have blonde hair. Because there is no variation in genetic information or expression, natural selection cannot occur. If a mutation occurred in a single individual to produce brown hair, there would now be variation in the genetic information and expression: blonde and brown hair. If the environment were more conducive to brown-haired individuals, then brown hair would become more common. Traits that positively affect the survival of a population are more likely to be reproduced.

How can we quantitatively measure whether natural selection is occurring within a population? Statistics and probability can be used to support explanations that organisms with advantageous heritable traits tend to increase in proportion, as opposed to organisms that do not have the advantageous trait. There are several statistical and probability-based models that can be used to analyze and predict trait expression:

### Punnett Squares

A Punnett square is a chart used to predict the **genotype** (genetic characteristics) and the **phenotype** (physical characteristics) of the children in a cross between two parents. For our purposes, Punnett squares are capable of predicting the probability of offspring inheriting genetic traits, or conditions, from their parents. Genetic variations that cause these conditions can be found on autosomes (chromosomes that are not sex-linked) or on the sex chromosomes. Traits found on the autosomes can be dominant, recessive, or demonstrate more complex interactions. Traits found on the sex chromosomes are called sex-linked. The following examples demonstrate how to use a Punnett square to determine genotype and phenotype for autosomal, sex-linked, and two-factor crosses.

#### How to Perform a Punnett Square

Steps	Example
<b>Step 1.</b> Designate a letter to represent the condition. A capital letter represents the dominant trait, and a lowercase letter represents the recessive trait. If the traits are sex-linked, the capital or lowercase letter is represented as a subscript next to either the X or Y sex chromosome.	<b>Autosome Trait</b> <b>D</b> = dominant <b>d</b> = recessive  <b>Sex-linked Trait</b> <b>X<sup>D</sup></b> = dominant <b>X<sup>d</sup></b> = recessive
<b>Step 2.</b> Determine the genotypes of each parent. If a parent is <i>homozygous dominant</i> , he/she has two (same) dominant traits (DD); if the parent is <i>homozygous recessive</i> , he/she has two (same) recessive traits (dd); and if the parent is <i>heterozygous</i> (different), he/she has both a dominant and a recessive trait (Dd).	<b>Autosome Trait</b> <i>mom is heterozygous and dad is homozygous recessive</i> <b>Dd x dd</b>  <b>Sex-linked Trait</b> <i>mom is heterozygous and dad is recessive</i> <b>X<sup>D</sup>X<sup>d</sup> x X<sup>d</sup>Y</b>

Name(s):

Period:

Date:

**Step 3.** Draw the Punnett square. If we are only predicting the probability of one trait, it is a *monohybrid cross*. If we are predicting the probability of two traits, it is a *dihybrid cross*.

**Step 4.** Write the possible traits that each parent could pass to the offspring along the sides and top of the Punnett square.

**Step 5.** Fill each box of the Punnett square by recording the letters from above and to the left of each empty box. Uppercase letters are always placed before lowercase letters.

**Step 6.** Determine the probability of the offspring receiving a trait. List the possible genotypes of the offspring below the Punnett square. The genotype results can be written as fractions or percentages. Once the genotype is known, the phenotype of offspring can also be determined.

### Monohybrid Cross

Autosomal Trait  
 $Dd \times dd$

<b>d</b>	<b>d</b>
<b>D</b>	<b>Dd</b>
<b>d</b>	<b>dd</b>

2/4 offspring  $Dd = 50\%$   
 2/4 offspring  $dd = 50\%$

Sex-linked Trait  
 $X^DX^d \times X^dY$

<b>X<sup>d</sup></b>	<b>Y</b>
<b>X<sup>D</sup></b>	<b>X<sup>D</sup>X<sup>d</sup></b>
<b>X<sup>d</sup></b>	<b>X<sup>d</sup>X<sup>d</sup></b>

$\frac{1}{4}$  offspring  $X^DX^d = 25\%$   
 $\frac{1}{4}$  offspring  $X^dX^d = 25\%$   
 $\frac{1}{4}$  offspring  $X^DY = 25\%$   
 $\frac{1}{4}$  offspring  $X^dY = 25\%$

### Dihybrid Cross

Autosomal Traits  
 $DdHh \times DdHh$

<b>DH</b>	<b>Dh</b>	<b>dH</b>	<b>dh</b>	
<b>DH</b>	<b>DDHH</b>	<b>DDHh</b>	<b>DdHH</b>	<b>DdHh</b>
<b>Dh</b>	<b>DDHh</b>	<b>DDhh</b>	<b>DdHh</b>	<b>Ddh<sub>h</sub></b>
<b>dH</b>	<b>DdHH</b>	<b>DdHh</b>	<b>ddHH</b>	<b>ddHh</b>
<b>dh</b>	<b>DdHh</b>	<b>Ddh<sub>h</sub></b>	<b>ddHh</b>	<b>ddhh</b>

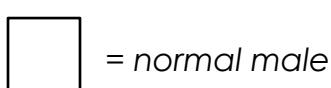
$\frac{1}{16}$  DDHH = 6.25%  
 $\frac{1}{16}$  DDhh = 6.25%  
 $\frac{1}{16}$  ddHH = 6.25%  
 $\frac{1}{16}$  ddhh = 6.25%

$\frac{2}{16}$  DDHh = 12.5%  
 $\frac{2}{16}$  DdHH = 12.5%  
 $\frac{2}{16}$  Ddhh = 12.5%  
 $\frac{2}{16}$  ddHh = 12.5%  
 $\frac{4}{16}$  DdHh = 25%

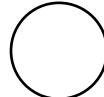
## Pedigrees

A pedigree is a diagram that uses symbols to represent family and genetic relationships. Pedigrees make it easier to visualize inheritance patterns within a family, and can be used to determine how a trait, or condition, is inherited. The following symbols and lines are used to represent individuals and relationships within a pedigree.

### Common Pedigree Symbols



= normal male

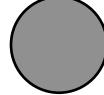


= normal female

= parents



= afflicted male



= afflicted female

= generation



= carrier male



= carrier female



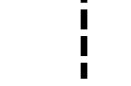
= siblings



= deceased before reproducing male



= deceased before reproducing female



= adoption

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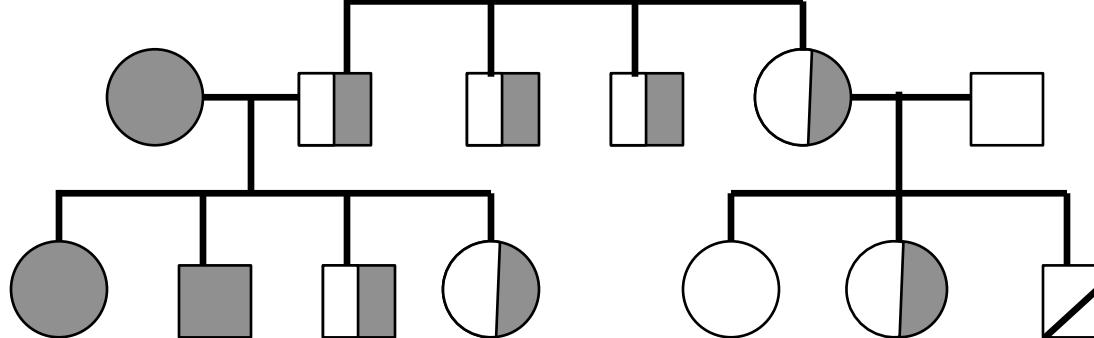
A pedigree is particularly useful when determining how genetic diseases are inherited within a family. Individuals who know their chances of having children that may inherit a difficult disease have the chance to make more informed decisions in their choices to have children. The following is an example of a simple pedigree with a mother who is afflicted with a genetic disease, and a father who is normal over three generations.

### Example Pedigree

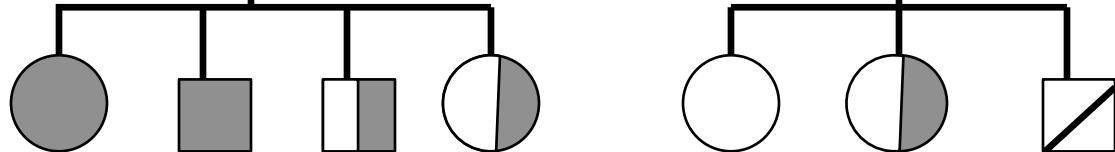
Generation I



Generation II



Generation III



### Hardy-Weinberg Equilibrium

The Hardy-Weinberg formula is used to analyze traits in populations, and detect whether a change has occurred in the frequency of those traits. In this way, the Hardy-Weinberg formula is capable of determining whether evolution has occurred. A change in the gene frequencies of a population over time can be used to determine whether the traits in a population are changing. If there is little or no change, the population is in equilibrium and no evolution is occurring.

#### The Hardy-Weinberg Formula(s)

$$p + q = 1$$

$$p^2 + 2pq + q^2 = 1$$

**p** = frequency of dominant allele

**q** = frequency of recessive allele

**$p^2$**  = percentage of homozygous dominant

**$q^2$**  = percentage of homozygous recessive

**$2pq$**  = percentage of heterozygous

Name(s):

Period:

Date:

## Hardy-Weinberg Example Problems

Example Problem 1	Example Problem 2
<p>16% of the Alaskan population has a recessive condition that makes the individuals sensitive to sunlight. Determine how many individuals in the population are heterozygous for this condition.</p> <p><b>1. Calculate the frequency of a single recessive allele (q)</b></p> <p>If <math>q^2 = 16\% = 0.16</math>, then <math>q = \mathbf{0.4}</math> (square root of 0.16)  <i>The frequency of the recessive allele as a percentage is 40% (<math>0.4 \times 100 = 40\%</math>)</i></p> <p><b>2. Calculate the frequency of a single dominant allele (p)</b></p> <p>Since <math>q = 0.4</math>, use the <math>p + q = 1</math> equation  <math>p + 0.4 = 1 \rightarrow p = \mathbf{0.6}</math>  <i>The frequency of the dominant allele as percentage is 60%</i></p> <p><b>3. Calculate the frequency of the remaining genotypes (homozygous dominant and heterozygous)</b></p> <p><u>homozygous dominant</u>  if <math>p = 0.6</math> then <math>p^2 = 0.6 \times 0.6 = \mathbf{0.36}</math></p> <p><u>heterozygous</u>  if <math>p = 0.6</math> and <math>q = 0.4</math>, then  <math>2pq = 2(0.6)(0.4) = \mathbf{0.48}</math></p> <p>The homozygous dominant frequency as a percentage is: ____%; Heterozygous frequency as a percentage is: ____%</p> <p>Check your answers by adding your <math>p^2</math>, <math>2pq</math>, and <math>q^2</math> values. (Remember: <math>p^2 + 2pq + q^2 = 1</math>)  Your values should equal 1.</p>	<p>A condition causing extremely brittle nails is recessive. In a village population, it is observed that 654 people have normal nails, and 86 people have brittle nails. How many people are homozygous dominant, heterozygous, and homozygous recessive?</p> <p><b>1. Calculate the frequency of a single recessive allele (q)</b></p> <p>There are 86 people with the condition, which can be used to determine <math>q^2</math>:  <math>86/740 \text{ total} = 0.12 \text{ (or } 12\%) = q^2</math>  if <math>q^2 = 0.12</math>, then <math>q = \mathbf{0.35}</math> (square root of 0.12)</p> <p><b>2. Calculate the frequency of a single dominant allele (p)</b></p> <p>Since <math>q = 0.35</math>, use the <math>p + q = 1</math> equation  <math>p + 0.35 = 1 \rightarrow p = \mathbf{0.65}</math></p> <p><b>3. Calculate the frequency of the remaining genotypes (homozygous dominant and heterozygous)</b></p> <p><u>homozygous dominant</u>  if <math>p = 0.65</math> then <math>p^2 = 0.65 \times 0.65 = \mathbf{0.42}</math>  42% of the village population = <math>0.42 \times 740 = 310</math> individuals</p> <p><u>heterozygous</u>  if <math>p = 0.65</math> and <math>q = 0.35</math>, then  <math>2pq = 2(0.65)(0.35) = \mathbf{0.46}</math>  46% of the village population = <math>0.46 \times 740 = 340</math> individuals</p> <p>Number of homozygous recessive individuals is: _____</p>

## The Chi-Square Test

While predicting the outcome of genetic crosses through probability is useful, it is only meaningful to science when the data is compared to actual test results. If the test results do not match the probability exactly, how can we determine whether the data collected from an experiment is correct? We use a “goodness of fit” test, called the chi-square test, which is a mathematical way to determine if the collected results are close enough to the expected results to be significant (low probability of being wrong/by chance). The chi-square formula is:

$$X^2 = \sum \frac{(obs - exp)^2}{exp}$$

### FORMULA KEY

<b>X</b> = is the Greek letter chi	<b>Σ</b> = sum	<b>obs</b> = observed results	<b>exp</b> = expected results
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Use the following steps to conduct a chi-square test on expected and actual results of a genetic cross. An example of each step has been provided for two parents who are heterozygous for brown eyes (Bb x Bb).

1. Use a Punnett square to determine the expected phenotype results.

**Phenotype**

3 brown eyes ( $3/4 = 0.75$ )  
1 blue eyes ( $1/4 = 0.25$ )

	B	b
B	BB	Bb
b	Bb	bb

2. Add up the total number of observed results from the collected data.

Collected Data		
Brown Eyes	Blue Eyes	Total Results
155	61	216

3. Now determine what the expected phenotype results would be with the total number of observed results calculated.

Expected Results	Brown Eyes	Blue Eyes
	0.75	0.25
Total Results	216	
Expect Phenotype Results	$0.75 \times 216 = 162$	$0.25 \times 216 = 54$

4. Compare the expected and observed results (collected from experiment) using the chi-square test.

Brown Eyes	Blue Eyes
$X^2 = (\text{obs} - \text{exp})^2 / \text{exp}$	
$X^2 = (155 - 162)^2 / 162$	$X^2 = (61 - 54)^2 / 54$
$X^2 = (-7)^2 / 162$	$X^2 = (7)^2 / 54$
$X^2 = 49 / 162$	$X^2 = 49 / 54$
$X^2 = 0.30$	$X^2 = 0.91$
$X^2 = 0.30 + 0.91 = 1.21$	

5. Compare the calculated chi-square value to Table 1 below. The degrees of freedom (df) is simply the number of phenotype options minus 1. In a monohybrid cross, the df = 1 and in a dihybrid cross, the df = 3. A P-value of 0.05 means that the observed and expected results did not deviate from each other more than 5%.

Table 1. Significance of Chi-Square Values	
Degrees of Freedom (df)	$X^2$ at a P-value of 0.05
1	3.84
2	5.99
3	7.81
4	9.49
5	11.07

Results
$X^2 = 1.21$
There are only 2 phenotypes so the df = 1, which makes the significant chi value 3.84.
Since the chi value you calculated (1.21) is less than 3.84, the expected ratio of a genetic cross is correct. If the chi value you calculated happened to be greater than 3.84, then the expected ratio would be incorrect and there are likely additional factors going on to create genetic variation for this gene.

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# Analysis of Human Traits

## HASPI Medical Biology Lab 22b



### Purpose

When focusing on genetics to understand evolution, probability and concepts of statistics are used to support explanations that organisms with an advantageous heritable trait tend to increase, in proportion to organisms lacking this trait. Human genetic diseases caused by mutations can appear to go against evolutionary theory, but not all mutations are completely detrimental. Some traits produced by these mutations could even be considered advantageous in certain environments. In this activity, you will have the opportunity to use common statistic analysis models--Punnett squares, pedigrees, the Hardy-Weinberg formula, and the chi-square test--to analyze human traits and hypothesize whether some of the traits could relay an advantage.

### Materials

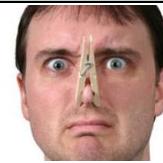
Calculator

### Directions

For each statistical analysis, a description and directions are provided followed by practice problems with real human mutations.

#### 1. What's That Smell?

Trimethylaminuria is an *autosomal recessive* condition that causes a pungent smell. Individuals with this condition cannot break down a compound known as trimethylamine, whose smell has been described as rotting eggs, rotten fish, garbage, or even urine. Trimethylamine builds up in urine, breath, and sweat creating a strong smell. Approximately 200,000 people in the United States have trimethylaminuria.



[http://www.lineaysalud.com/images/stories/sindromedeolorapescado\(1\).jpg](http://www.lineaysalud.com/images/stories/sindromedeolorapescado(1).jpg)

1a. Use a Punnett square to determine whether two individuals who are heterozygous for the trimethylaminuria trait could have a child with trimethylaminuria.

1b. Use a Punnett square to determine the chances of a man with trimethylaminuria and a woman who is normal (TT) having a child with trimethylaminuria?

1c. Could this trait possibly be advantageous in a specific environment?  
Explain your answer.

Name(s):

Period:

Date:

## 2. Origins of the Werewolf Myth

Hypertrichosis, also known as "werewolf syndrome," is a genetic condition that causes excessive body hair growth. Those who have this condition exhibit hair on the face, shoulders, and ears. There are two types of hypertrichosis – the most severe form is *hypertrichosis lanuginose* which is an *autosomal dominant* condition, while generalized hypertrichosis is less severe and sex-linked.



[http://cdn2.top10hut.com/wp-content/uploads/2013/02/1326947669\\_03657000.jpg](http://cdn2.top10hut.com/wp-content/uploads/2013/02/1326947669_03657000.jpg)

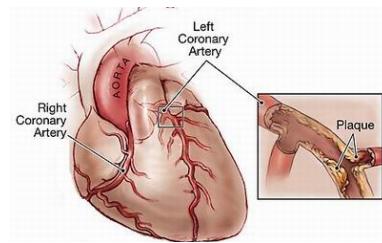
2a. A mother is heterozygous for hypertrichosis lanuginose. What are the chances of her having a child with hypertrichosis lanuginose if the father does not have the condition? Use a Punnett square to explain your answer.

2b. A man has generalized hypertrichosis and has married a woman who does not carry the trait. He is worried about his children having the trait. What are the chances of them having children with generalized hypertrichosis?

2c. Could this trait possibly be advantageous in a specific environment?  
Explain your answer.

## 3. Blood Type and Heart Disease Risks

Recent research has proposed a link between heart disease and blood types. Individuals with Type O blood have the lowest risk, Type A had a 6% increase in risk, Type B had a 15% increase in risk, and Type AB had a 23% increase in risk. There are actually 3 traits, or alleles, that contribute to blood type – A ( $i^A$ ), B ( $i^B$ ), and O ( $i$ ). Since there are more than two alleles, blood types are considered to have multiple alleles. We can only inherit two alleles, and how these alleles combine determine our blood type. Both the A allele and the B allele are dominant, and they both show up in the phenotype. This is called codominance.



[http://www.medicalook.com/diseases\\_images/coronary\\_heart\\_disease.jpg](http://www.medicalook.com/diseases_images/coronary_heart_disease.jpg)

3a. A couple both have Type AB blood. They are worried that all of their children will also be Type AB and have a high risk for heart disease. What percentage of their children could have Type AB?

3b. A child has Type O blood. What blood types could the child's parents have?

3c. Could this trait possibly be advantageous in a specific environment?  
Explain your answer.

Name(s):

Period:

Date:

#### 4. A Real Life Genetic Superhero?

Two relatively new and rare mutations could be capable of making a genetic superhero. The first mutation is *autosomal dominant* and impacts the LRP5 protein, which regulates bone mineral density. The mutation causes the protein to increase the density of bones making them incredibly strong. The second mutation is *autosomal recessive* and impacts the myostatin protein, which limits muscle growth. Individuals with this mutation are capable of drastically increasing muscle growth and strength. The combination of incredibly strong bones and strength in a single individual could be amazing!



[http://1.bp.blogspot.com/\\_Dg1MLpkMYL0/TSuWc2fmlZI/AAAAAAAAGA/RyZ7tih38vM/s1600/blog68.jpg](http://1.bp.blogspot.com/_Dg1MLpkMYL0/TSuWc2fmlZI/AAAAAAAAGA/RyZ7tih38vM/s1600/blog68.jpg)

4a. Complete a dihybrid Punnett square to determine if any of the children from the following couple could create an individual with increased bone mineral density and increased muscle growth/strength.

Increased bone density = **B**

Normal bone density = **b**

Normal muscle growth/strength = **M**

Increased muscle growth/strength = **m**

The mother's genotype is **BbMm**,  
and the father's genotype is **bbMm**.

4b. Could a father with increased bone density and normal muscle growth, and a mother with increased bone density and normal muscle growth, produce a child with increased bone density and increased muscle growth? Explain your answer using a Punnett square.

4c. Could these traits possibly be advantageous in a specific environment?  
Explain your answer.

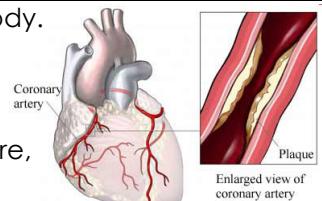
Name(s):

Period:

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## 5. Apolipoprotein AI-Milano

Apolipoprotein AI-Milano is a mutation in a protein that carries cholesterol within the body. Individuals who have this mutation have less cholesterol in the bloodstream and have a significantly reduced risk of coronary heart disease, which is caused by plaque buildup in the arteries (atherosclerosis). In fact, to date none of the individuals with this mutation have exhibited any signs or symptoms of coronary heart disease. While still rare, 3.5% of Limone sul Garda, a small village in Italy, carry this mutation.



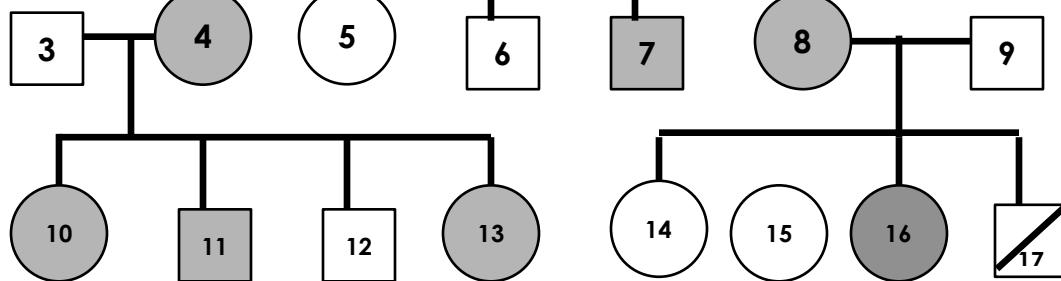
<http://www.thewellingtoncardiacservices.com/images/Heart/Coronary-Artery-Disease2.jpg>

The following pedigree displays the inheritance of the Apolipoprotein AI-Milano mutation in a family from Limone sul Garda.

### Generation I



### Generation II



5a. From looking at the pedigree, is the Apolipoprotein AI-Milano mutation autosomal dominant, autosomal recessive, or sex-linked? Explain your answer.

5b. What are the genotypes of individuals 1 and 2? Explain your answer.

5c. How many of the children between individuals 1 and 2 should carry the Apolipoprotein AI-Milano mutation? How many actually do carry the mutation according to the pedigree?

5d. How many of the children between individuals 3 and 4 should carry the Apolipoprotein AI-Milano mutation? How many actually do carry the mutation according to the pedigree?

5e. How many of the children between individuals 8 and 9 should carry the Apolipoprotein AI-Milano mutation? How many actually do carry the mutation according to the pedigree?

5f. Explain why the number of children predicted to carry the Apolipoprotein AI-Milano mutation is different than the actual number of children produced by this family.

5g. Could this trait possibly be advantageous in a specific environment? Explain your answer.

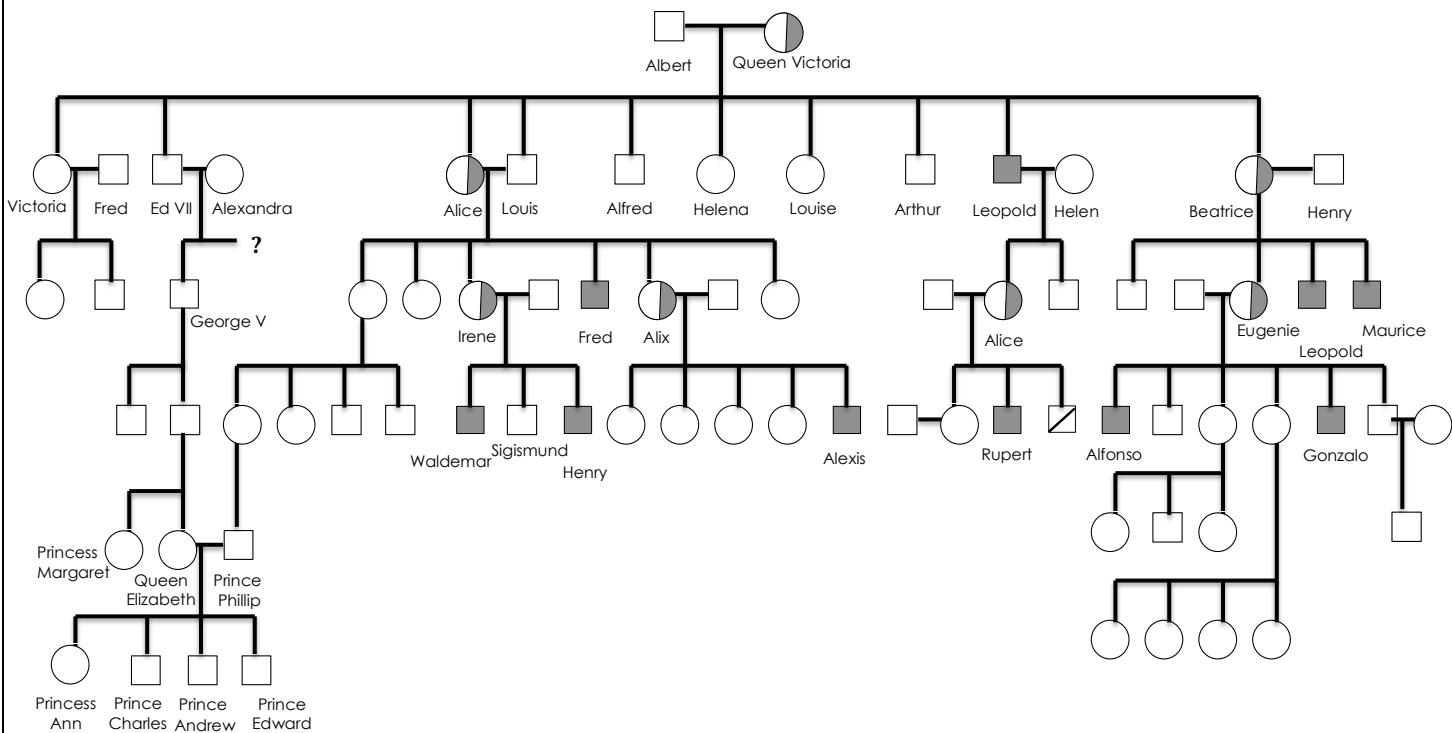
## 6. Hemophilia and The Royals

Hemophilia is a sex-linked condition in which individuals are missing or have low levels of clotting factors in the blood. Clotting factors and platelets are needed in order to stop bleeding when a cut or bruise occurs. A lack of clotting factors causes uncontrolled external and internal bleeding following injury, and can even lead to death. Hemophilia spread through the royal families of Europe and Russia in large part by Queen Victoria, who was a carrier, and three of her children – Alice, Beatrice, and Leopold.



<http://diseasespictures.com/wp-content/uploads/2012/10/hemophilia-2.jpg>

The following pedigree shows how hemophilia was carried through Queen Victoria's family.



6a. Did any females in Queen Victoria's line inherit hemophilia? If yes, who?

6b. Did any males in Queen Victoria's line inherit hemophilia? If yes, who?

6c. Why is there a difference in the number of females/males that inherited this trait?

6d. If Maurice or Leopold were to marry a normal female (no hemophilia), what are the chances their children would inherit hemophilia? What are the chances their grandchildren would inherit hemophilia?

6e. Could this trait possibly be advantageous in a specific environment? Explain your answer.

Name(s):

Period:

Date:

## 7. The Blue People

Methemoglobinemia is an *autosomal recessive* condition that causes a deficiency in the protein that allows red blood cells to use glucose. The function of red blood cells is to carry oxygen, and when they are carrying oxygen they are red in color. The deficiency of the protein causes the red blood cells to carry oxygen differently and turn blue in color. This creates a blue skin color, which is often worse under stressful conditions. Methemoglobinemia has been found in Native Americans, Eskimos, and a small population in Kentucky.

[http://trialx.com/g/Congenital\\_Methemoglobinemia-4.jpg](http://trialx.com/g/Congenital_Methemoglobinemia-4.jpg)



7a. Using the following information, create a pedigree for the family to determine how methemoglobinemia was inherited through a family. Carrie and her husband Mark are both healthy, but they have just had a son, Jathan, born with methemoglobinemia. Neither Carrie nor Mark had ever heard of the disease. Carrie knows very little about her mother's extended family in Kentucky. Carrie's mother, Sandra, passed away when she was young, and Carrie moved with her father, Michael, to California. She has decided to visit her Kentucky relatives to understand how her son may have inherited methemoglobinemia. Visiting with her great grandmother, Vivian, gave her the following information:

Vivian's mother and father, Mary and Luke, looked normal and never turned blue. Vivian and her two sisters, Anna and Jemma, also looked normal, but their brother Jeb always turned slightly blue when he was worried. Jeb and Anna never had any children. Jemma married Tom from down the road and they had five children: two boys and three girls. All of Jemma and Tom's children looked normal. Vivian married Sam from across the river. They had two children, Robert and Sandra, who were both normal. Robert died when he was 8 from pneumonia. Sandra had one child, Carrie.

Using this information, create a pedigree of Carrie's family and relatives to determine how her son ended up with methemoglobinemia.

7b. Could this trait possibly be advantageous in a specific environment? Explain your answer.

Name(s):

Period:

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### 8. Backwards or Forwards Evolution: Uner Tan Syndrome

Uner Tan Syndrome is an extremely rare *autosomal recessive* condition in which individuals travel on all fours, speak primitively, and have varying levels of mental retardation. It was only recently discovered and is localized in western Asia. While there have been multiple accounts of Uner Tan Syndrome, currently only a single family in Turkey is exhibiting symptoms of this genetic condition.



<http://blogs.plos.org/neuroanthropology/files/2010/09/turkey-walking-on-all-fours-1.jpg>

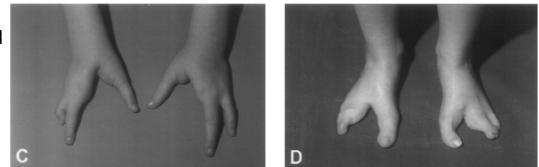
8a. A hidden population has been discovered in Turkey. Researchers have calculated that 22% of the population have Uner Tan Syndrome. Using the Hardy-Weinberg formulas, calculate what percentage of the population are heterozygous carriers for this condition. Show your work.

8b. Could this trait possibly be advantageous in a specific environment? Explain your answer.

### 9. Human Claws: Ectrodactyly

Ectrodactyly is an *autosomal dominant* disorder that causes a cleft, or split, where the middle finger or middle toe should form. This creates a limb, or limbs, that appear to form a claw, which is why this condition is also known as "lobster claw syndrome." Some individuals also suffer from hearing loss. Approximately 1 in 90,000 births results in a child with this condition.

<http://withfriendship.com/images/i/44490/with-ectrodactyly-usually.jpg>



9a. The town of Freunberg has a population of 3,523. Approximately 652 individuals in this population have extreme ectrodactyly, which is a homozygous dominant condition. Using the Hardy-Weinberg formulas, calculate what percentages of the population are homozygous dominant, heterozygous, and homozygous recessive. Show your work.

9b. Could this trait possibly be advantageous in a specific environment? Explain your answer.

Name(s):

Period:

Date:

## 10. Sickle Cell and Malaria Resistance

Sickle cell anemia is an autosomal recessive condition that causes red blood cells to collapse and appear curved in shape. This shape limits the ability of red blood cells to carry oxygen, and individuals with sickle cell anemia experience varying degrees of pain, shortness of breath, and blood flow blockage. Sickle cell is an interesting disease because, while it is recessive, individuals who are heterozygous experience mild symptoms of sickle cell anemia. This is called incomplete dominance and occurs because half of the red blood cells are created with the normal allele, and are normal, while the other half of the red blood cells are created with the sickle cell allele, and are sickle-shaped. Even more interesting is the fact that individuals who are heterozygous for sickle cell anemia are protected from malaria, a serious infectious disease passed by mosquitoes and caused by the Plasmodium parasite. Malaria infections are very common in areas of Africa, and from recent research it is estimated that 10-40% of the populations in these areas carry the sickle cell allele.

<https://www.sciencenews.org/sites/default/files/14307> ; [http://compulenta.computerra.ru/upload/iblock/88b/o\\_plasmodium%20falciparum.jpg](http://compulenta.computerra.ru/upload/iblock/88b/o_plasmodium%20falciparum.jpg)

**Sickle Red Blood Cell**



**Plasmodium Parasite**



10a. In Kenya, approximately 16% of the population has sickle cell anemia (homozygous recessive). What percentage of the population will have resistance to malaria from the sickle cell allele? Show your work.

10b. A genetic research project in Ethiopia determined that 68% of the population carries the normal allele for red blood cells. What percentage of the population carries the sickle cell allele? Show your work.

10c. What percentages of the population are homozygous dominant, heterozygous, and homozygous recessive?

10d. Could this trait possibly be advantageous in a specific environment? Explain your answer.

Name(s):

Period:

Date:

## 11. The Dominance of Marfan Syndrome

Marfan Syndrome is an *autosomal dominant* condition that affects connective tissues within the body. Due to the fact that connective tissues are present throughout the body, the mutation causes a large variety of signs and symptoms. A few common signs include disproportionate limbs, tall and slender build, protruding breastbone, crowded teeth, scoliosis, heart murmurs, and nearsightedness. Approximately 1 in 10,000 births result in a child with Marfan Syndrome.



<http://i.imgur.com/3fXmSxt.jpg>

11a. In a research project, children of individuals that were known to be heterozygous for Marfan Syndrome were tested for the disease. The following observed results were collected:

Marfan Syndrome (Mm)	Normal (mm)
378	412

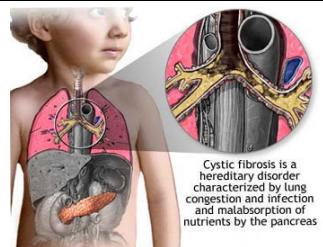
Perform a chi-square test to determine whether these results support that Marfan Syndrome is a dominant disease. Show your work.

11b. Could this trait possibly be advantageous in a specific environment? Explain your answer.

## 12. Is Cystic Fibrosis Really Recessive?

Cystic fibrosis is an *autosomal recessive* disease caused by a mutation in a common cell membrane protein. This mutation creates a problem with how substances move into and out of cells, causing a buildup of substances such as sweat and mucus in body tissues and organs. A common symptom of cystic fibrosis is an overabundance of mucus in the lungs. The life expectancy for individuals with cystic fibrosis was historically very low (early 20s), but has improved due to medical discoveries.

Approximately 1 in 3,000 births results in a child with cystic fibrosis.



<http://medicalstuttering.com/wp-content/uploads/2013/05/Cystic-fibrosis.jpg>

12a. In a 2008 study, there were 30,000 people in the United States with cystic fibrosis. The approximate total population of the United States at the time was 305,000,000. Perform a chi-square test to determine whether these results support that cystic fibrosis is a recessive disease. Show your work.

12b. Could this trait possibly be advantageous in a specific environment? Explain your answer.

Name(s):

Period:

Date:

### 13. A New Dwarfism

Dwarfism is a condition characterized by individuals who are short in stature caused by a mutation that slows or delays growth. Technically, dwarfism is defined as a height of less than 147 cm. There are actually different dwarfism mutations. Achondroplasia is an *autosomal dominant* condition, while diastrophic dysplasia (DTD) is an *autosomal recessive* condition. Approximately 70% of all dwarfism is caused by achondroplasia.



<http://geneticmutationruebe3.wikispaces.com/file/view/little-people-big-world31.jpg/309905494/little-people-big-world31.jpg>

13a. A new form of dwarfism has been discovered in a population in a remote area of Australia. Out of a population of 980, researchers counted 72 individuals with dwarfism. Using these values and the chi-square test, determine whether the condition is dominant or recessive. (HINT: You will need to compare your results to phenotype ratios for a dominant condition AND a recessive condition.) Show your work and explain your answer.

13b. Could this trait possibly be advantageous in a specific environment? Explain your answer.

Name(s):

Period:

Date: