## Topic 3: Mendelian Genetics

### Learning Objective

EVO-2.A: Explain how shared, conserved, fundamental processes and features support the concept of common ancestry for all organisms.
IST-1.I: Explain the inheritance of genes and traits as described by Mendel's laws.

### Essential Knowledge

EVO-2.A.1: DNA and RNA are carriers of genetic information.
EVO-2.A.2: Ribosomes are found in all forms of life.
EVO-2.A.3: Major features of the genetic code are shared by all modern living systems.
EVO-2.A.4: Core metabolic pathways are conserved across all currently recognized domains.
IST-1.I.1: Mendel's laws of segregation and independent assortment can be applied to genes that are on different chromosomes.
IST-1.I.2: Fertilization involves the fusion of two haploid gametes, restoring the diploid number of chromosomes and increasing genetic variation in populations by creating new combinations of alleles in the zygote—

a. Rules of probability can be applied to analyze passage of single-gene traits from parent to offspring.
b. The pattern of inheritance (monohybrid, dihybrid, sex-linked, and genetically linked genes) can often be predicted from data, including pedigree, that give the parent genotype/phenotype and the offspring genotypes/phenotypes.

### Vocabulary/Review Questions

| | | | |
|---|---|---|---|
| True Breeding | P Generation | $F_1$ Generation | $F_2$ Generation |
| Punnett Squares | Monohybrid Cross | Dihybrid Cross | Phenotype |
| The Multiplication Rule | Law of Segregation | Law of Independent Assortment | The Addition Rule |
| Recessive | Alleles | Heterozygous | Genotype |
| Dominant | Homozygous | Pedigrees | |

1. What could have happened if Mendel did not study pea plants (i.e. if he only had roses to study)?
2. Describe how a test cross can be used to determine whether a dominant trait is homozygous or heterozygous.
3. Differentiate between the law of segregation and the law of independent assortment.
4. A plant heterozygous for seed shape (Ss) is crossed with a plant that is homozygous recessive for seed shape (ss). What percentage of the offspring will be heterozygous?
5. Two plants that are heterozygous for color and seed shape (RrWw= red flowers and wrinkled seeds) were crossed. Draw and complete the punnett square. If there were 600 offspring total, how many would be white with smooth seeds?
6. What type of cross would give a 3:1 ratio? What about a 9:3:3:1 ratio?
7. In a cross between AaBBCcDD x AabbCcDd what is the probability that the offspring will be AaBbccDd?
8. *Be able to read/create pedigrees*

## Topic 4: Non-Mendelian Genetics

### Learning Objective

IST-1.J.1: Explain deviations from Mendel's model of the inheritance of traits.

### Essential Knowledge

IST-1.J.1: Patterns of inheritance of many traits do not follow ratios predicted by Mendel's laws and can be identified by quantitative analysis, where observed phenotypic ratios statistically differ from the predicted ratios—

a. Genes that are adjacent and close to one another on the same chromosome may appear to be genetically linked; the probability that genetically linked genes will segregate as a unit can be used to calculate the map distance between them.

IST-1.J.2: Some traits are determined by genes on sex chromosomes and are known as sexlinked traits. The pattern of inheritance of sex-linked traits can often be predicted from data, including pedigree, indicating the parent genotype/phenotype and the offspring genotypes/phenotypes.
IST-1.J.3: Many traits are the product of multiple genes and/or physiological processes acting in combination; these traits therefore do not segregate in Mendelian patterns.

### Vocabulary/Review Questions

| | | | |
|---|---|---|---|
| Incomplete dominance | Polygenic inheritance | Multiple alleles | Epistasis |
| X-linked | Codominance | Y-linked | Hemizygous |
| Barr body | Recombinants | Linkage map | Chi square |

1. If a red flower is crossed with a white flower, and the resulting offspring are pink, what is the easiest way to explain these results?
2. If a red flower is crossed with a white flower, and the resulting offspring are both red and white, what is the easiest way to explain these results?
3. A woman with type B blood has a child with a man who has type A blood. With this limited information, what are possible genotypes for the woman? For the man?
4. Why are there very few Y-linked disorders?
5. Circle the correct option: if an x-linked trait is due to a recessive allele, females will only express the trait if they are homozygous / heterozygous.
6. Color blindness is a common, recessive X-linked disorder. Normal vision (XN) is dominant to color blindness (XC). If a colorblind male has a child with a woman who is a carrier for the disorder, what is the probability that their child will be color blind?

## Common Ancestry

_____ and _____ carry genetic information.

The genetic code is shared by:

## Gregor Mendel

Mendel was an Austrian _____ who experimented on _____ plants and discovered the basic principles of _____.

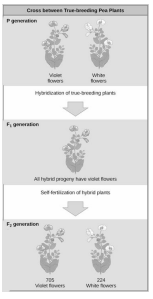
## Pea Plant Traits

Mendel only tracked characteristics that came in **two** distinct forms:

Examples:

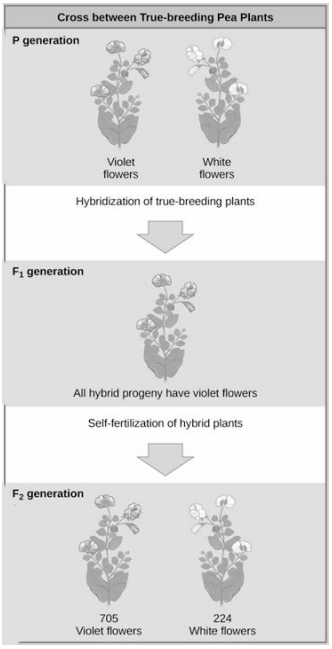
True breeding:

Example:




➔ Questions?
➔ Textbook chapters/pages to review

## Generations



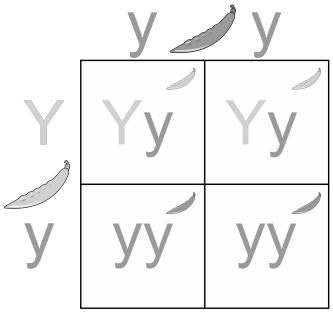

P generation:

$F_1$ generation:

$F_2$ generation:

## Punnett Squares

Punnett squares:




## Genetics Vocabulary

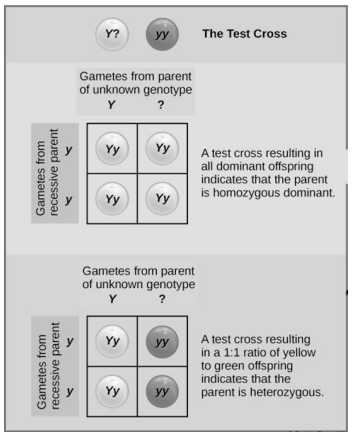
Homozygous:

➔ Questions?
➔ Textbook chapters/pages to review

Heterozygous:

Genotype:

Phenotype:




**Testcross**

Testcrosses help to determine:

**Principles of Heredity**

Mendel's experiments allowed him to develop <u>two</u> fundamental _____.

1. The law of _____

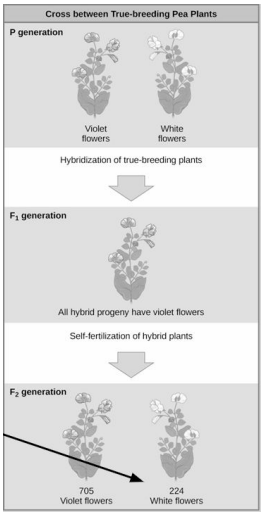
2. The law of _____

**Discoveries**

Mendel noticed that the cross between _____ and _____ true breeding pea plants

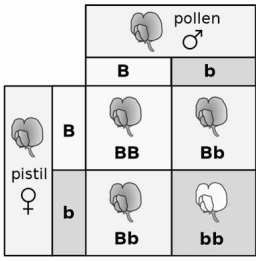
produced only _____ $F_1$ offspring

Did the white characteristic disappear?




➔ Questions?
➔ Textbook chapters/pages to review

**Dominant vs Recessive**




Mendel performed the same crosses for each of the _____ characteristics of pea plants and found the same result:

**Mendel's Model**

To explain the ___:___ ratio he observed in the _____ generation, Mendel created a model with four concepts:

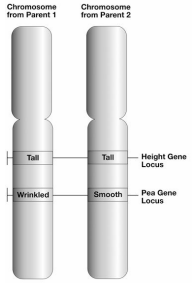
1.

2.

3.

4.

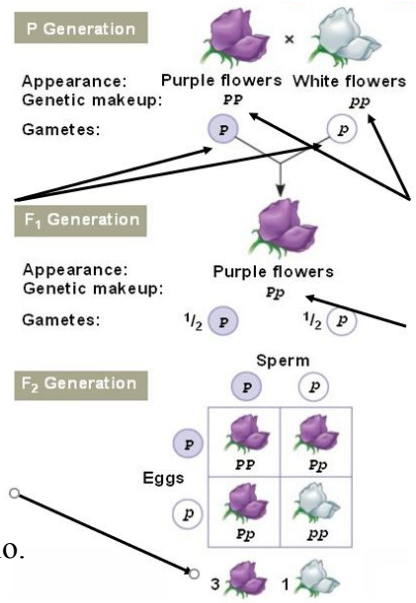
**Alleles: A Closer Look**




➔ Questions?
➔ Textbook chapters/pages to review

**The Law of Segregation**

Each _____ for the P generation will contain one _____ for flower color.




_____ plants will have two identical alleles.

_____ generation are all hybrids: Pp

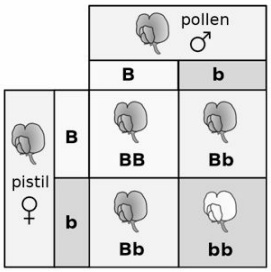
_____ generation:

Pp x Pp produces __:__ ratio.

**Monohybrid Crosses**

Example:

Monohybrid crosses:




**The Law of Independent Assortment**

Mendel's second principle is the law of independent assortment:

This law only applies to

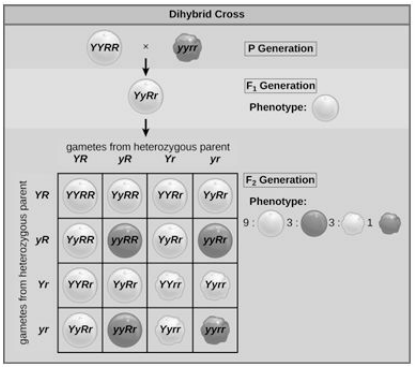
1.

OR

2.

➔ Questions?
➔ Textbook chapters/pages to review

## Dihybrid Crosses




The law of independent assortment was determined by doing crosses between plants that were true breeding for _____ traits, which produced $F_1$ hybrids known as _____

Example:

Dihybrid cross:

Produces a ___:___:___:___ phenotypic ratio.

## How to Solve Genetics Problems

1.

2.

3.

4.

## Practice Problems

1. A tall plant (TT) is crossed with a short plant (tt). What percentage of the offspring will be tall?



➔ Questions?
➔ Textbook chapters/pages to review

2. Round seed shape is dominant to wrinkled seed shape. A plant that is heterozygous for the round seed shape is crossed with a plant that is homozygous for round seed shape. What percent of the offspring will be homozygous dominant?

3. In cats, short hair is dominant to long hair. A true-breeding short haired cat is crossed with a cat that is heterozygous for the trait. What percentage of the offspring will have long hair?

4. Purple (P) is dominant to white (p) flowers. In a homozygous dominant purple plant, what gametes would be produced? What about in a plant that is heterozygous?



➔ Questions?
➔ Textbook chapters/pages to review

5. In pea plants, purple flower color is dominant to white flower color, and round pods are dominant to wrinkled pods. If a true breeding purple flowered round pod plant is crossed with a true breeding white flowered wrinkled pod plant, what will the resulting $F_1$ generation be? Hint: don't do a Punnett square.

6. In pea plants, purple flower color is dominant to white flower color, and round pods are dominant to wrinkled pods. If a plant that is heterozygous for both traits is self crossed, what will the phenotypic ratios of the $F_1$ generation be?

## Mendelian Genetics

1. What is meant by the term "true breeding?"

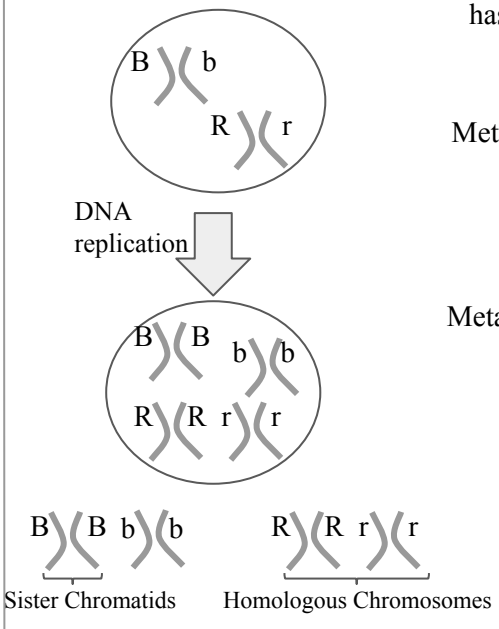
2. Differentiate between the P, $F_1$, and $F_2$ generation.

3. What is the difference between an organism's genotype and an organism's phenotype?

4. What is stated by the laws of segregation and independent assortment?

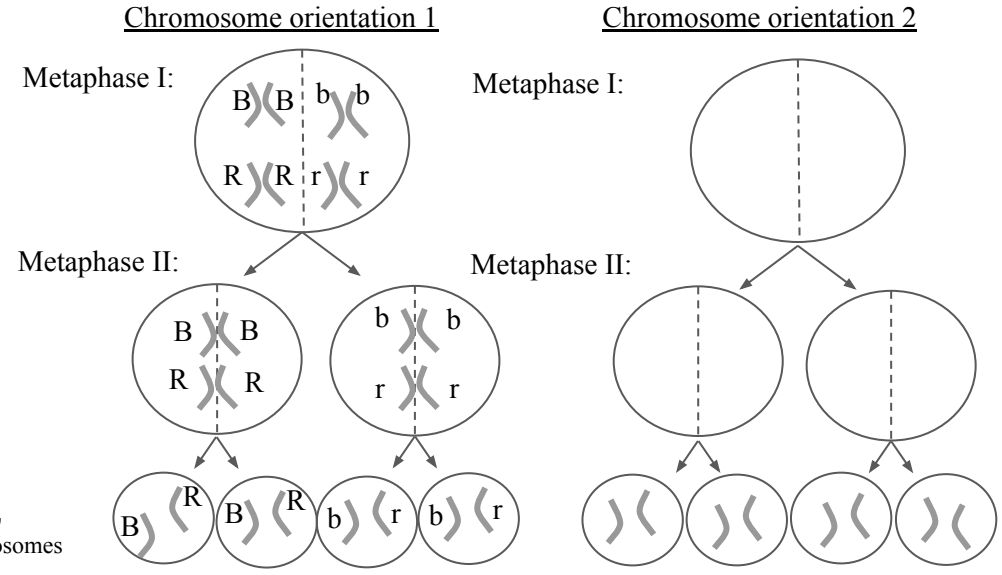
5. To help with the concept of independent assortment, let's look at an example of a plant that is heterozygous for two traits. One trait (B) is for a blue flower color. The second trait (R) is for round seeds. The plant's genotype therefore is BbRr. During meiosis, we will see the law of independent assortment in action. Let's take a look inside the plants cell.

a) Remember, during interphase the chromosomes are replicated to produce identical sister chromatids

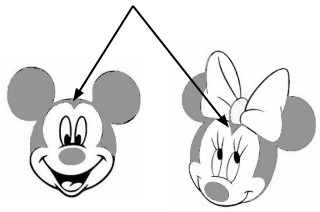
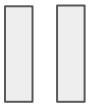


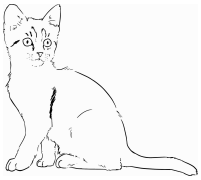
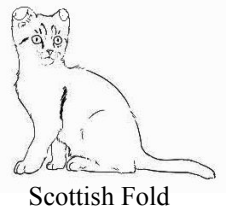
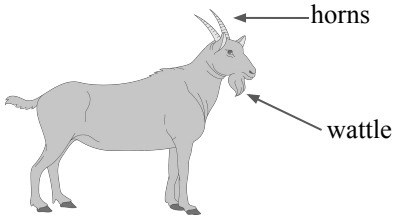

b) Next, during meiosis, specifically metaphase I, the homologs will randomly line up at the metaphase plate. The first chromosome orientation has been done for you.

c) There is another way the homologs can align at the metaphase plate. Determine the second orientation and fill in the subsequent stages




d) At the end of meiosis the BbRr plant produces four unique gametes: _____

| | |
|---|---|
| 1. The trait for a widow's peak is dominant to having a straight hairline. Mickey and Minnie mouse both have widow's peaks. If both Mickey and Minnie are heterozygous for the trait, what is the chance that their children will also have a widow's peak?<br>  | Show work here: |
| 2. Dimples are a dominant trait. If a man who is homozygous dominant for dimples has a child with a woman who is heterozygous for the trait, what will the genotypic ratios of the child be? | Show work here: |
| 3. Freckles are clusters of cells that overproduce melanin (the pigment that gives skin color). Having freckles is dominant to not having freckles. If two people who are heterozygous for freckles has a child, what would the genotypic and phenotypic ratios of the child be? | Show work here: |
| 4. Marfan syndrome is an autosomal dominant disease that affects connective tissue in many parts of the body. Connective tissue provides strength, flexibility, and support to bones, ligaments, and muscles. People suffering from Marfan syndrome experience heart issues and eye sight problems. A couple is interested in having a child, but suspect that one of them has the disease. They are worried about passing the disease to their children, so they go to a genetic counselor for advice. Use the image below to help you determine the parents' genotypes. Then, determine the probability that, if they have a child, the child will have Marfan syndrome<br>  <br>Affected parent<br>  <br>Unaffected parent | Show work here: |

| | |
|---|---|
| 5. TRPV4 is a gene in cats that encodes for calcium permeable ion channels. The Scottish Fold is a breed of cats that has a mutated dominant TRPV4 gene. Mutations to this dominant gene cause developmental issue in bones and cartilages. One copy of the mutated dominant gene causes the the signature "fold" in the ears of the Scottish Fold breed. Two copies of the dominant gene cause a condition known as osteochondrodysplasia (crippling effect) and causes significant issues to the tail, limbs, and paws of the cat. Imagine you are a part of the local government and have been tasked with creating a set of rules for breeders of Scottish Fold cats. 1) Determine all possible gene combinations and their resulting phenotypes. 2) Determine the acceptable genotypes that breeders can cross so that no cats will be born with osteochondrodysplasia.<br>  <br>Normal (straight) ears<br>  <br>Scottish Fold | Show work here: |
| 6. Goats can either be pulled or horned. The pulled, or no horn, trait (P) is dominant to the horned (p) trait. Another trait in goats are wattles, which are hair covered structures on their necks. Even though most goats do not have wattles, having wattles (W) is dominant to not having wattles (w). A breeder crosses a pulled, wattled male goat with a pulled, wattled female goat. Some of the offspring have horns and no wattles. 1) Knowing this, what are the genotypes of the male and female parental goats? 2) Perform a cross of the parental genotypes to find the phenotypic ratios of the $F_1$ generation.<br>  <br> | Show work here: |
| 7. In pea plants, round seeds (R) are dominant to wrinkled seeds (r), and yellow seeds (Y) are dominant to green seeds (y). If you were to cross a Rryy plant with a rrYy plant, what proportion of the offspring would have wrinkled, yellow seeds? What proportion would have round, green seeds? | Show work here: |

8. In pea plants, purple (P) flower color is dominant to white (p) flowers, and yellow (Y) seeds are dominant to green (y) seeds. Flower color and seed color are inherited independently. A cross is done between two plants of unknown genotypes and the results of the $F_1$ generation are analyzed.

| Purple/yellow | Purple/green | White/yellow | White/green |
|---|---|---|---|
| 0 | 0 | 321 | 101 |

You notice right away that all offspring are white. You then notice that the yellow: green ratio is approximately 3:1. Knowing this, determine the parental genotypes.

Show work here:

9. A second cross is done between two plants of unknown genotypes (possessing the same traits as in problem #8). The results of the $F_1$ generation are in the table below.

| Purple/yellow | Purple/green | White/yellow | White/green |
|---|---|---|---|
| 155 | 146 | 52 | 49 |

The results for this appear to be more complicated, but you know that the ratios between purple:white flowers and yellow:green seeds are the key to solving the parental genotypes. Use problem 8 to help you if you get stuck.

Show work here:

10. In guinea pigs, a black coat (B) is dominant to an albino coat (b), and short hair (H) is dominant to long hair (h). A BbHh guinea pig is crossed with a bbHH guinea pig. If 30 offspring are produced, how many would be expected to be a) black with short hair, b) albino with short hair, and c) albino with long hair?

Show work here:

**Laws of Probability**

The laws of segregation and independent assortment reflect rules of probability

The multiplication rule:

Example: If you flip a coin twice, what is the probability that it will land heads up both times?

Example: What is the probability of having 3 girls in a row?

**Practice Problems:**

1. Tallness is dominant to dwarfism in pea plants. Plants that are heterozygous for height are crossed. What is the probability that the offspring will be homozygous dominant?

2. In a cross between AABBCc x AaBbCc what is the probability that the offspring will be AaBbcc?

****The rule of multiplication allows you to not do a massive Punnett square! Instead, work through each letter at a time in the initial cross.*

**Laws of Probability**

The addition rule:

Example: what is the chance of rolling a dice and it lands on a 1 or 6?

**Practice Problems:**

1. In a cross between AABBCc x AaBbCc, what is the probability that the offspring will be AaBbcc or AABBCC?

| | A | A |
|---|---|---|
| A | | |
| a | | |

| | B | B |
|---|---|---|
| B | | |
| b | | |

| | C | c |
|---|---|---|
| C | | |
| c | | |

➔ Questions?
➔ Textbook chapters/pages to review

## Using Probability in Genetics

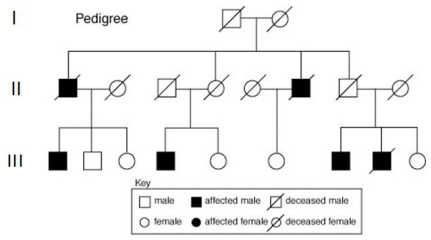
| | |
|---|---|
| 1. Let's say you roll a dice (standard sides 1-6), what is the probability it will land on a 2 or a 5? To solve this, will you use the addition rule or multiplication rule? Why? | Show work here: |
| 2. The genotype of a particular plant is AaBbCcDd. If this plant was self-crossed, what is the probability that the offspring would have the following genotypes:<br>a) AaBBCCDD<br>b) aaBBccdd<br>c) AaBbCcDd<br><br>(Hint: don't do a tetrahybrid cross, do monohybrid crosses for each gene then use the multiplication rule.) | Show work here: |
| 3. Approximately 4000 years ago, a small number of people settled in areas of Finland and became separated from the rest of the population. These people reproduced, but due to the low number of people, it caused a loss of genetic diversity in the subsequent offspring, which caused many disorders to arise. These disorders are collectively known as Finnish heritage diseases. This event was so significant that even today, one in five Finnish people carry at least one gene related to a Finnish heritage disease. A man and a woman, both of Finnish heritage are aware of this, so they see a genetic counselor. They are interested in having a child, but fear they may pass on a disease. They have their DNA analyzed and it comes back that they are both carriers for the recessive disease known as megaloblastic anemia, a type of anemia common in Finnish descent. Thankfully, if they have an affected child, it is treatable.<br>a) What is the probability that, if they have a child,. it will have megaloblastic anemia?<br>b) Let's say they decide to have three children total. What is the probability that all three children would have the disease?<br>c) What is the probability that, if they have three children, none of them will have the disease? | Show work here: |

| | |
|---|---|
| 4. Cystic fibrosis is an autosomal recessive disease that affects the lungs and digestive system. A young couple named Sara and Daniel are interested in having a child. Neither Sara or Daniel nor either of their parents have cystic fibrosis, but Sara and Daniel both have siblings who have the disease.<br>a) What are the genotypes of both Sara and Daniel's parents?<br>b) What is the probability that Sara and Daniel are carriers for the disease? (Do the parental Punnett square and remember they DON'T have the disease,)<br>c) Using the probability from part b, what is the chance that, if they are both carriers, their child will have the disease? | Show work here: |
| 5. A plant with the genotype BbCc is crossed with a plant that has the genotype BBcc. What is the chance that the offspring will be either BbCc or BBcc? | Show work here: |
| 6. There is a couple named Louise and Robert. They want to have exactly four children. Use the multiplication and addition rules to calculate the probability that Louise and Robert will have two girls and two boys in <u>any</u> order. (Hint: set up a table to help you determine all the different combinations possible.) | Show work here: |

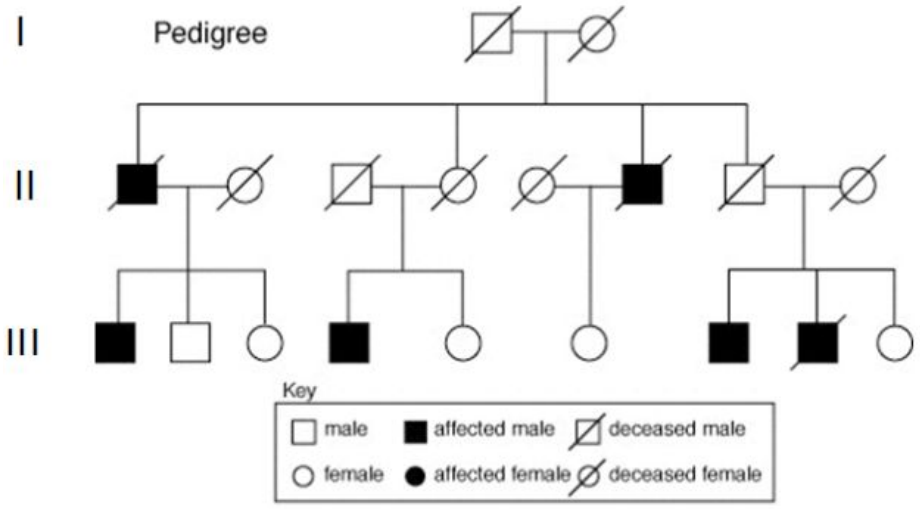
## Pedigrees

Many human traits follow Mendelian patterns of genetics

Pedigrees:




## Reading Pedigrees




If a trait is _____, one parent must have the trait.

If a trait is X-linked, then _____ are more commonly affected than _____.

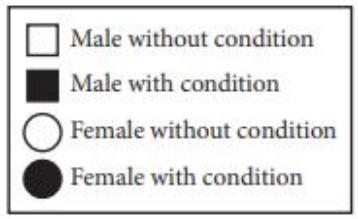
➔ Use this space to reflect on Topic 3
➔ Textbook chapters/pages to review

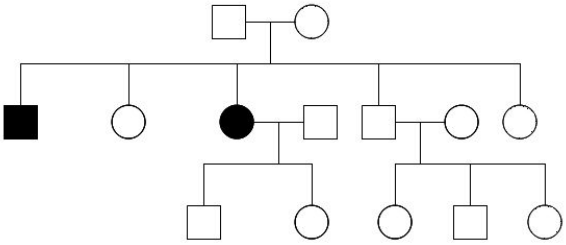
## Practice: Analyzing Pedigrees

1. The pedigree to the right is tracking a disease through three generations. Is this disease autosomal recessive or autosomal dominant? Why?

Could this disease be X-linked? Why or why not?

Pedigree:

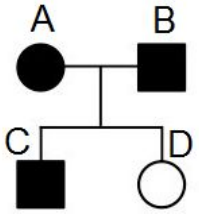


2. Look at the pedigree to the right. Is this disease autosomal recessive or autosomal dominant? Why?

What are the possible genotypes for individuals A-D?

Pedigree:

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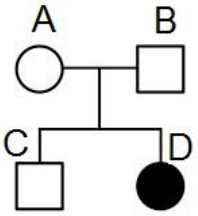
Pedigree:



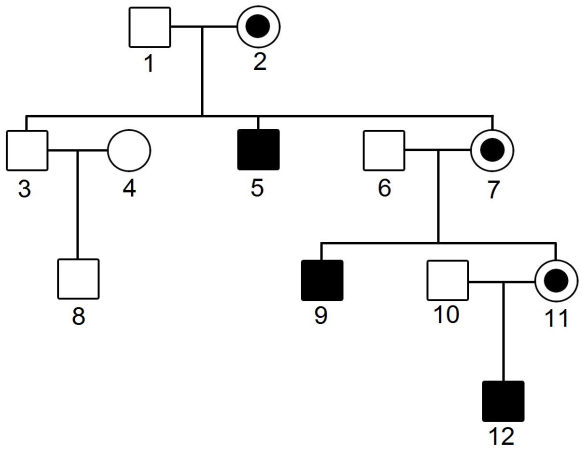
*Come back to this page after you have learned about non-Mendelian genetics*

4. Analyze the pedigree to the right. Notice the dots in the center of some individuals? That means they are carriers for the disease. With this information, is this disease likely autosomal dominant, autosomal recessive, X-linked recessive, or X-linked dominant? Why?

What are the likely genotypes of individuals 1 and 2?

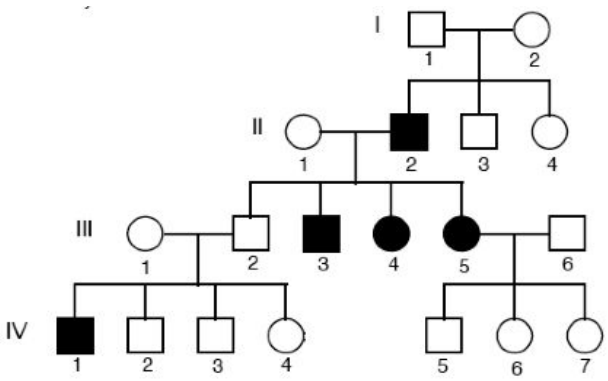
What are the likely genotypes of individuals 10 and 12?

Pedigree:



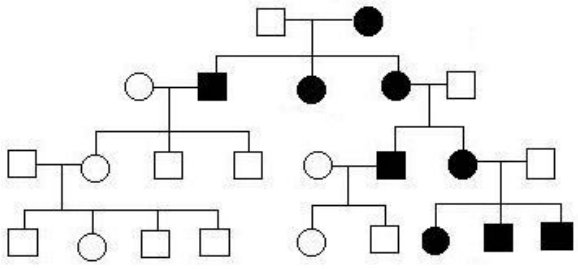

5. Using the pedigree to the right, explain how the DNA of individual 4 from generation III has DNA from both individuals 1 and 2 in generation I.

Pedigree:




6. The pedigree to the right depicts the inheritance of a disorder through mitochondrial DNA. Why is it that affected males produce no offspring with the disorder?

Pedigree:



## It Started with One

Do your genes matter to anyone besides you? Do they affect the rest of the population? Well, as it turns out, they might have more of an effect than you imagine.

Huntington's disease (HD) is a rare, **autosomal dominant** neurodegenerative disease caused by a type of mutation known as a triplet expansion. The triplet expansion occurs on a mutated *HTT* gene, where a triplet of nucleotides (CAG) is repeated several times. The number of repeats also plays an important role in the age of onset of the disease; individuals who have 6-35 CAG repeats will be unaffected, while people with 40 or more CAG repeats are guaranteed to develop symptoms of the disease.

HD is unique in that the symptoms and onset of the disease begin in adulthood, and usually appear around the age of 30-45; this means that many people who have the disease have already had kids and passed on the genetic trait before their symptoms even appeared. Symptoms of HD include: a loss of motor control, altered personality, and a decline in overall cognitive function. The disease is fatal and there are no current treatments to slow the progression of the disease.

A small, rural fishing village in the state of Zulia, Venezuela has the highest concentration of people suffering from Huntington's Disease in the world. In 1979, scientists caught wind of the alarming rates of HD in this area. Several research teams were sent to meet with the people in the village and take blood samples to examine their DNA. The data that was collected and continues to be collected from the people of this village has led to astounding advancements of our knowledge of the disease, which will hopefully one day lead to better treatments, prevention, and maybe even a cure.

Scientists were able to obtain over 4,000 blood samples for DNA analysis as well as an immense amount of clinical data (i.e. family history, symptoms, etc.). Using this data they were able to determine the source of the devastating genetic mutation, a single woman who helped found the village ten generations prior. Currently there are over 18,000 individuals living in this area of Venezuela who have Huntington's Disease. Examine Figures 1 and 2 below then answer the corresponding questions..

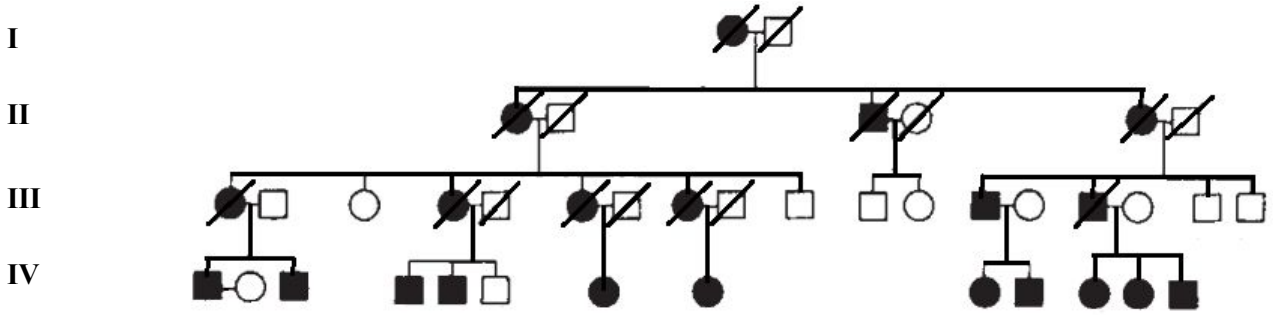



**Fig. 1** Pedigree depicting four generations of a Venezuelan Huntington's Disease family. Circles represent females and squares represent males. A shaded symbol means they are affected with the disease. A slashed symbol means the individual has died.

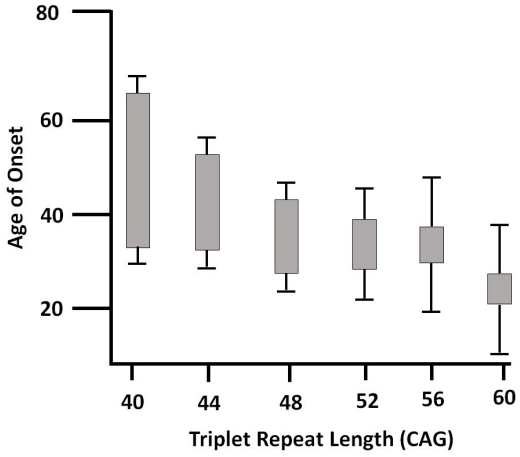



**Fig. 2** Box plot representing the age of onset vs. the length of the triplet expansion. The larger the boxes, the more data that fell within that range.

**Analysis Questions**

1. If one person with the trait for Huntington's disease (Hh) has a child with a person who does not have the trait for Huntington's disease, what is the chance their child will have the disease? *Show your work in the space below.*

2. What is the chance that these parents (from problem 1) will have two children who do not have the disease? *Show your work in the space below.*

3. If two people who carry the trait for Huntington's disease (Hh) have a child, what is the chance their child will have the disease? What is the chance their child will be genotypically normal? *Show your work in the space below.*

*Questions 4-6 refer to Figure 1.*

4. How could you tell that HD is an autosomal dominant disease just by looking at the pedigree?

5. In the first generation, what is the genotype of the father?

6. In the second generation, what is the genotype of the son?

*Questions 7-8 refer to Figure 2.*

7. What happens to the age of onset as the triplet expansion lengthens?

8. Why might the error bars be larger for the data including triplet repeats of 56-60?