Name: _____ Date: _____ Block: _____

# All About Genetics Webquest

This webquest will guide you through the "Big Ideas" of Genetics, from Mendel's first pivotal experiments to practicing some of the more recent advances in genetic research on your own! Please record your answers to each section in the space provided.

**I. INTRODUCTION**: CRASH COURSE: HEREDITY

- View the YouTube video Heredity: Crash Course Biology #9 at http://www.youtube.com/watch?v=CBezq1fFUEA. Answer the questions below.

1. What is HEREDITY? Who first studied heredity in a scientific way?

2. What is the difference between a GENE and an ALLELE?

3. What does DOMINANCE refer to in genetics?

4. What is the difference between HETERZYGOUS and HOMOZYGOUS?

5. Explain the difference between AUTOSOMES and SEX CHROMOSOMES.

**II. MENDEL'S GENETICS:** Mendel is often called the "Father of Modern Genetics". Check out the basic ideas behind his experiments at http://anthro.palomar.edu/mendel/mendel_1.htm. Pay close attention how Mendel designed his experiments. Most of Mendel's early work in genetics can be summarized in two major principles, the Law of Segregation and the Law of Independent Assortment. Answer the questions below.

*HINT: If you don't understand an underlined term, click on it for a short definition!*

1. Name SEVEN traits that Mendel found that are passed from one generation to another without being changed or blended (they appear in only one of two forms).

2. What are TWO reasons that Mendel chose to use pea plants for his experiments?

3. Summarize in your own words the TWO major principles derived from Mendel's work.

4. After you have finished learning about Mendel, click on "Practice Quiz" and quiz yourself. Then click "next topic" on the bottom of the page and learn about Punnett Squares and the Probability of Inheritance.

**III. PROBABILITY OF INHERITANCE** Carefully read over "Probability of Inheritance" at http://anthro.palomar.edu/mendel/mendel_2.htm. The Punnett Square is an important tool used to determine the PROBABILITY of having offspring with a particular genotype.

1. What is the difference between GENOTYPE and PHENOTYPE in an organism? Give an example of each.

2. Punnett squares plot the (circle one) GENOTYPES / PHENOTYPES of the parents.

3. If two people who are both carriers for a genetically inherited fatal recessive disease decide to become parents, what will be the odds that their children will also be carriers? Show your work with your Punnett Square.

a. 1 out of 4
b. 2 out of 4
c. 3 out of 4
d. 4 out of 4

3. After you have finished learning about Punnett Squares, complete the "Practice Quiz". Then answer the questions provided on your answer sheet. Finally click "next topic" on the bottom of the page and learn about the exceptions to Mendel's genetics.

**IV. EXCEPTIONS TO SIMPLE INHERITANCE:** Not every genetic trait follows the rules of basic dominance/recessiveness that Mendel described! There are some important exceptions to his "rules". Find out about them at http://anthro.palomar.edu/mendel/mendel_3.htm.

1. What does it mean if a trait is POLYGENIC? List three traits that are polygenic in humans.

2. "Intermediate Expression" is also known as CODOMINANCE (this is the term our textbook uses). What is different about heterozygous offspring if the trait is codominant rather than completely dominant (like Mendel's pea plants)?

3. What does it mean to be CODOMINANT? How does human blood type illustrate codominance?

4. What are "MULTIPLE ALLELE Series" traits? How do human blood types demonstrate multiple alleles?

**V. GENETIC DISEASES:** Genetic diseases and disorders can be classified based on their causes. A disorder where there is a mutation in the DNA sequence of a single gene is called a point mutation (or a single gene disorder). Chromosomal mutations, however, involve deletions, additions, or alterations of entire chromosomes (or very large portions of them).

Check out the website, "Your Genes, Your Health" from Cold Spring Harbor Laboratory at http://www.ygyh.org/. Take the time to learn about some of the more prominent genetic disorders, selecting them from the menu at the left. Answer the questions below.

1. HUNTINGTON DISEASE: (Circle one) This disease is DOMINANT / RECESSIVE.

   - How many copies of the gene most offspring inherit to inherit the disease? _____

2. SICKLE CELL DISEASE:
   - What are the symptoms of sickle cell disease?

   - Why is it considered beneficial to be HETEROZYGOUS for the disease?

   - Does someone who is heterozygous for the disease show all of the symptoms?

DOWN SYNDROME:

- How does the inheritance of Down Syndrome differ from that of Huntington Disease or Sickle Cell?

- What type of genetic disorder would this be classified as?

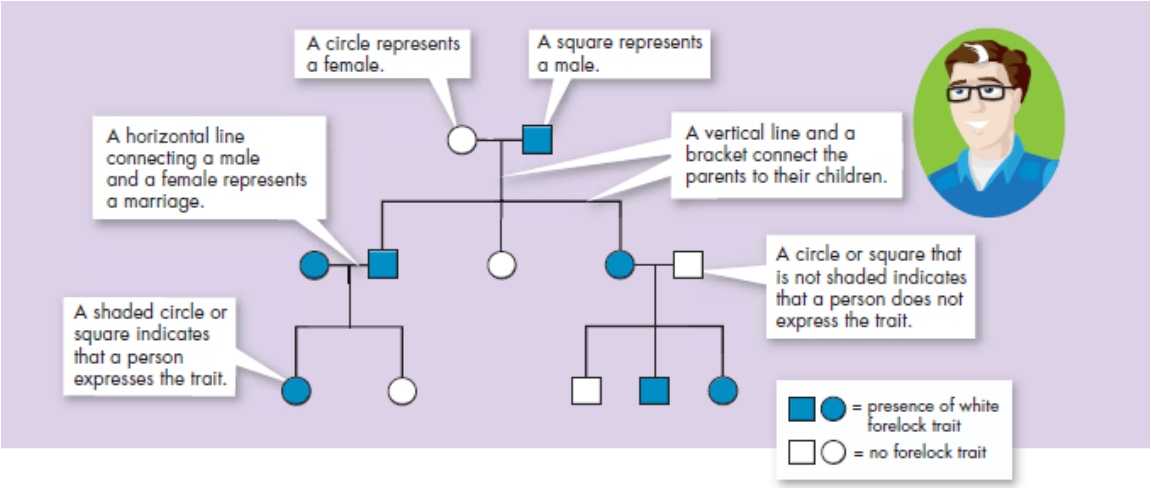
TAY-SACH'S DISEASE:

- How is Tay-Sach's disease inherited?

- Would this be considered a (circle one) DOMINANT or RECESSIVE disease?
- What are the symptoms of early-onset Tay-Sach's?

## VI. PEDIGREES

A **pedigree** is a visual chart that depicts a family history or the transmission of a specific trait. They can be interesting to view and can be important tools in determining patterns of inheritance of specific traits. Pedigrees are used primarily by genetic counselors when helping couples decide to have children when there is evidence of a genetically inherited disorder in one or both families. They are also used when trying to determine the predisposition of someone to carry a hereditary disease for example, familial breast cancer.



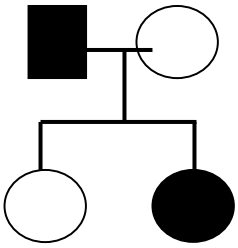

**The Components of a Pedigree:**

**<u>Squares</u> are used to indicate males in a family.** 

**<u>Circles</u> are used to indicate females.** 

**If the individual is "affected" by the trait (dominant or recessive) we <u>darken</u> the shape.**

**A <u>line</u> between a male and a female indicates a marriage or union.**
**A <u>line</u> drawn down from the marriage line indicates offspring.**



**Sometimes, you will see some shapes filled in only <u>half way</u> –**
**this notation indicates a *hybrid* (heterozygous) or *carrier* of the trait.**

**Analyzing Simple Pedigrees:**
A pedigree is just like a family tree except that it focuses on a specific genetic trait. A pedigree usually only shows the phenotype of each family member. With a little thought, and the hints below, you may be able to determine the genotype of each family member as well!

Hints for analyzing pedigrees:
1) If the individual is homozygous recessive, then both parents MUST have at least one recessive allele (parents are heterozygous or homozygous recessive).

2) If an individual shows the dominant trait, then at least one of the parents MUST have the dominant phenotype. This one will be pretty obvious when you look at the pedigree.

3) If both parents are homozygous recessive, then ALL offspring will be homozygous recessive.

**NOTE:** In a pedigree, the trait of interest can be *dominant or recessive*. The majority of harmful genetic conditions are only seen when an individual is homozygous recessive - examples of conditions caused by **recessive** alleles include cystic fibrosis (a disease of the secretory glands, including those that make mucus and sweat), albinism (a lack of pigmentation), and phenylketonuria (a metabolic disorder). Some genetic conditions are caused by **dominant** alleles (and may therefore be expressed in homozygous dominant or heterozygous individuals)- examples of conditions caused by dominant alleles include polydactyly (presence of extra fingers), achondroplasia (a type of dwarfism), and neurofibromatosis (a nervous disorder).

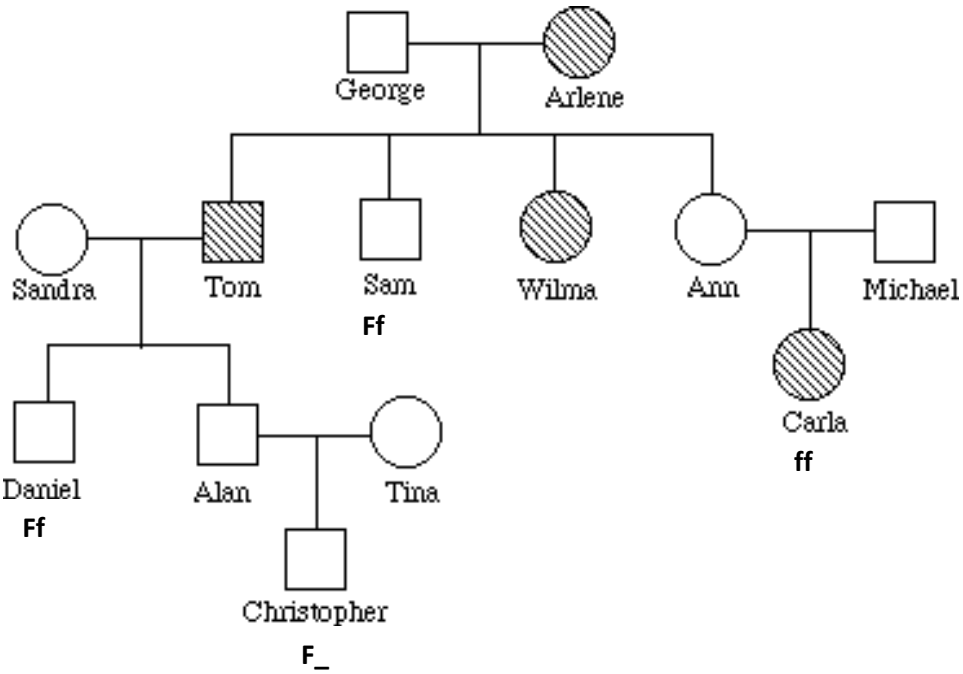
**Human Pedigrees:**

1. **Tracing the path of an autosomal recessive trait** - Falconi anemia

**Forms of the trait:**

- The **dominant** form is <u>normal</u> bone marrow function - in other words, no anemia.
- The **recessive** form is Falconi anemia. Individuals affected show slow growth, heart defects, possible bone marrow failure and a high rate of leukemia.

A **typical pedigree** for a family that carries Falconi anemia. Note that carriers are **not** indicated with half-colored shapes in this chart.




Analysis Questions:
To answer questions #1-5, use the letter "f" to indicate the recessive Falc oni anemia allele, and the letter "F" for the normal allele.

1. What is Arlene's genotype?_____
2. What is George's genotype?_____
3. What are Ann & Michael's genotypes?_____
4. Most likely, Sandra's genotype is _____

5. List three people from the chart (other than George) who are most likely *carriers* of Falconi anemia.

_____

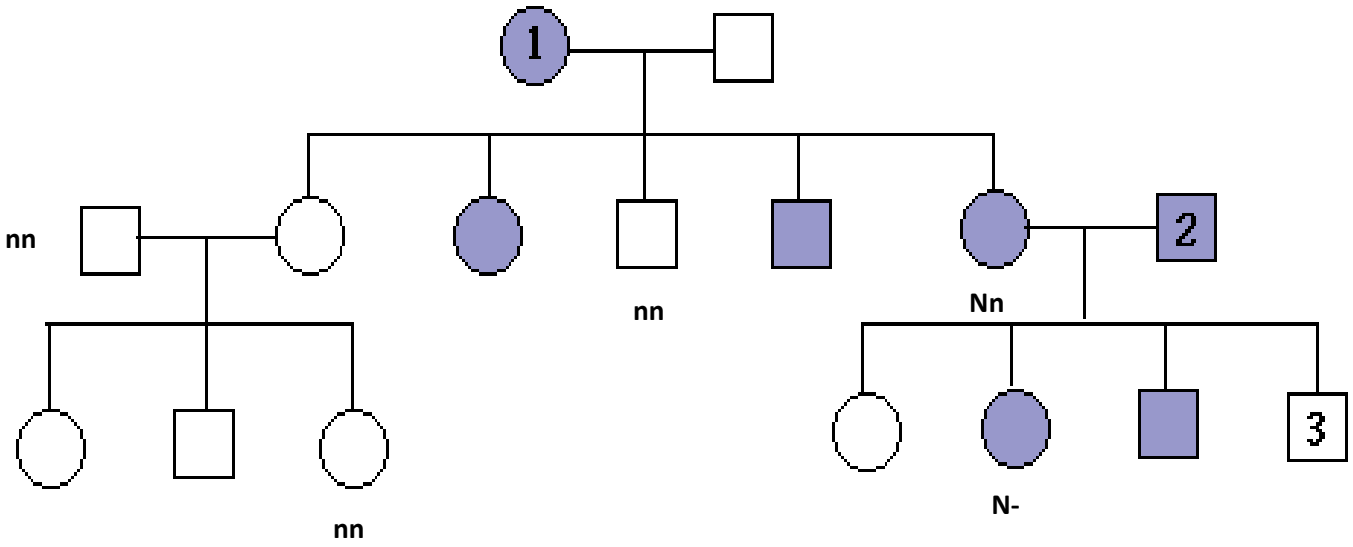
**Source:** *www.aurora-schools.org/.../**Studyingpedigrees**JR10-...*

2. **Tracing the path of an autosomal dominant trait**: Neurofibromatosis

Forms of the trait:

- The dominant form is neurofibromatosis, caused by the production of an abnormal form of the protein neurofibromin. Affected individuals show spots of abnormal skin pigmentation and non-cancerous tumors that can interfere with the nervous system and cause blindness. Some tumors can convert to a cancerous form.
- The recessive form is a <u>normal</u> protein - in other words, no neurofibromatosis.

A typical pedigree for a family that carries neurofibromatosis is shown below. Note that carriers are not indicated with half-colored shapes in this chart. Use the letter "N" to indicate the dominant neurofibromatosis allele, and the letter "n" for the normal allele.




Analysis Questions:

1. Is individual #1 most likely homozygous dominant or heterozygous? Explain how you can tell.

2. What is the genotype of individual #3?

3. Can you be sure of the genotypes of the affected siblings of individual #3? Explain.

## VII. BLOOD TYPING

**Remember, earlier in this webquest we learned that multiple alleles can influence a person's blood type…**

Human red blood cells carry markers on their cell surface. These markers are antigens. We call these markers antigens because they will cause an immune response and produce antibodies. A "B" red blood cell (a red blood cell carrying the B antigen) will be destroyed by the B antibody. Similarly, a red blood cell carrying the A antigen will be destroyed by the A antibody. RBC is short for Red Blood Cell.

People that are blood type B carry antibodies to A. This is why we cannot give Type A blood to a Type B person. The A antibodies present in the patient's own blood will attack the donated Type A blood. This is the same for people with Type A blood. These people make antibodies that will attack Type B blood. Therefore we cannot give Type B blood to a Type A person.

There are two more variations of human blood type when considering the ABO blood system: Type AB and Type O. People with Type AB, the codominant trait, blood have A and B antigens on their own blood cells, so they can't have antibodies to either. If they did, those antibodies would constantly attack the person's own red blood cells. So Type AB people have no antibodies present in their blood and can receive blood of any blood type. These people are called ***universal recipients***.

Type O blood does not have the A or B antigen present on the red blood cells. Because of this reason, Type O blood can be given to any person. People with Type O blood are therefore called ***universal donors***. Because there are no antigens present on the donated red blood cells, patients that receive this blood will not have a reaction to the donated red blood cells.

Here is a chart to help you remember the important parts:

| Blood Type | Genotype | | Can Receive Blood From: |
|---|---|---|---|
| A | $i^A i$<br>$i^A i^A$ | AA<br>AO | A or O |
| B | $i^B i$<br>$i^B i^B$ | BB<br>BO | B or O |
| AB | $i^A i^B$ | AB | A, B, AB, O |
| O | $i\,i$ | OO | O |

**Source:** **http://www.biologycorner.com/anatomy/blood/notes_bloodtype.html**

**Complete the blood typing activity food at http://www.zerobio.com/drag_gr11/pedigree/pedigree4.htm**

Follow the directions on the screen to complete the activity. Answer the questions below.

1. Type the cross for the parents (father x mother) in generation 1.

2. What blood type can the parents in generation 1 NOT produce?

3. What is the best way to write the genotype of the son with type A blood in generation 2?

4. What other blood type can be produced in generation 3?

5. Could the husband of the daughter in generation 2 have blood type B and still produce a type O child? Explain.

6. Click the "next" arrow at the bottom right of the screen. Complete the quiz and record your quiz score in the answer packet attached.

Quiz Score: _____/10

## CONGRATULATIONS! YOU ARE FINISHED!!!!!!!!!

**PA Biology Standards:**

| | |
|---|---|
| **BIO.B.1.2.2** | Explain the functional relationships among DNA, genes, alleles, and chromosomes and their roles in inheritance. |
| **BIO.B.2.1.1** | Describe and/or predict observed patterns of inheritance (i.e., dominant, recessive, co-dominance, incomplete dominance, sex-linked, polygenic, and multiple alleles). |
| **BIO.B.2.1.2** | Describe processes that can alter composition or number of chromosomes (i.e., crossing-over, nondisjunction, duplication, translocation, deletion, insertion, and inversion). |
| **BIO.B.2.3.1** | Describe how genetic mutations alter the DNA sequence and may or may not affect phenotype (e.g., silent, nonsense, frame-shift). |
| **BIO.B.2.4.1** | Explain how genetic engineering has impacted the fields of medicine, forensics, and agriculture (e.g., selective breeding, gene splicing, cloning, genetically modified organisms, gene therapy). |