Rieder - Genetics
Worksheet 6: **HALLOWEEN EDITION!** 
Due: 10/24/17 at the beginning of class

Name: _____

I consulted/worked with: _____

This homework was (circle all that apply):

Stupid Fun Too easy Too difficult Educational

Other: _____ Helpful Not helpful

I will not accept late homework. Special exceptions will be made **only** in the event of illness or if you contact me at least 24 hours ahead of when the assignment is due (at my discretion).

POINTS: / 50

MUMMIES
When archaeologists uncovered the mummy of Egyptian Pharaoh Akhenaten (also named Amenhotep IV), whose consort was the famous queen Nefertiti and whose son was the famous Tutankhamun, they suggested, based on his tall, thin stance and elongated limbs, that he suffered from Marfan's Syndrome.

1. Marfan's is an **autosomal dominant** genetic disease. If Akhenaten was **heterozygous** for the **allele** that causes Marfan's, what percentage of his children are predicted to have Marfan's if Nefertiti did not suffer from the disease? How did you arrive at this percentage (1 pt)?

2. Pharaoh Akhenaten had 6 daughters and 3 sons. Is the allele that causes Marfan's more likely to be inherited by one sex over the other? How did you arrive at this conclusion (1 pt)?

3. Are there **carriers** of the Marfan's allele? Why/why not (1 pt)?

VAMPIRES

Historians speculate that the legend of vampires originated from the genetic disease porphyria. Porphyria sufferers have a **mutation** in one of the genes involved in heme synthesis-- heme carries oxygen in your blood, so porphyria patients have extremely low levels of both heme and oxygen in their bloodstreams.

Modern treatments for porphyria include intravenous blood transfusions, but ancient remedies *could* (speculation) have involved porphyria patients actually drinking human blood! Porphyria sufferers also have skin that is sensitive or blisters in the sunlight, causing them to avoid the daylight. The phenomenon when a mutation in one gene causes a variety of symptoms that may seem unrelated is called **pleiotropy**. The **pleiotropic** symptoms of porphyria are very vampyric!

Many genes contribute to heme synthesis, and mutations in any ONE of them can lead to porphyria symptoms. Because these genes are located on different chromosomes, mutations that cause porphyria may be inherited in an **autosomal dominant**, **autosomal recessive**, or **X-linked dominant** manner, depending on the location of the gene and nature of the allele.

Vlad, a porphyria sufferer from Transylvania, has porphyria symptoms that result from a mutation in a heme synthesis **X-linked** gene. His mutation is **dominant**.

4. What percentage of Vlad's daughters will inherit the porphyria allele and what percentage will be affected by porphyria symptoms (2 pts)?

5. What percentage of Vlad's sons will inherit the porphyria allele and how many will be affected by porphyria symptoms (2 pts)?

6. Vlad's vampyric friend Morticia suffers from porphyria caused by a mutation in a different gene than Vlad's. She has an **autosome-linked recessive** mutation, which results in the p allele. The **dominant** allele for this locus is P. What are all the possible **diploid genotypes** of Morticia's parents (2 pts)?

Parent 1	Parent 2
_____	_____
_____	_____
_____	_____

Another vampyric genetic disease is xeroderma pigmentosum, which causes extreme sensitivity to sunlight. Sufferers have mutations in genes that are involved in repairing damaged DNA. We will discuss this in more detail in class, but sunlight, specifically ultraviolet light, can cause **DNA damage**. As we learned in a recent presentation on melanoma, DNA damage can lead to cancer, so this is why it's important to wear sunscreen!

7. Mutations in genes involved in **DNA repair** result in sufferers being unable to repair sunlight-induced DNA damage. Most sufferers in the US have mutations in their *XPC*, *ERCC2*, or *POLH* genes. Do you think these mutations are **gain-of-function** or **loss-of-function** mutations and why (1 pt)?

FRANKENSTEIN

8. Even though Dr. Frankenstein's monster, Frank, is sewn together from various people's body parts, Frankenstein knew he had to carefully match body part blood type or Frank could suffer from organ rejection, so Frank the monster has blood type A-. His consort, Bride of Frank (BOF), has blood type B+. What are the possible **diploid blood genotypes** and all possible **haploid gamete genotypes** of Frank and Bride of Frank (3 pts)?

Frank diploid genotypes:	Resulting Frank haploid gametes:
_____	_____
_____	_____

BOF diploid genotypes:	Resulting BOF haploid gametes:
_____	_____
_____	_____
_____	_____
_____	_____

9. Is it possible that Frank and BOF's son will have blood type AB+ (1 pt) (yes/no)?_____

10. Is it possible that Frank and BOF's son will have blood type O- (1 pt) (yes/no)?_____

Monsters like Frank with type A- blood have antibodies in their blood **plasma** (the cell free portion of the blood) against both the B molecule and the Rhesus (Rh) molecule. People like BOF with type B+ blood have blood plasma antibodies against the A molecule. **Antibodies** are molecules produced by our immune systems that attack cells or bacteria/viruses that the body recognizes as "foreign."

Individuals with blood type O- are considered the "universal blood *cell* donors" because red blood cells from O- individuals can be donated to people of any blood type (O- red blood cells do not have any A, B, or Rh molecules on them, so a recipient's plasma antibodies will not recognize them as "foreign"). People with blood type AB+ are the "universal blood *cell* recipients" because they do not have plasma antibodies against A, B, or Rh molecules, so will not recognize any donated blood cell as "foreign," regardless of its molecules.

11. Which parent(s) (Frank, BOF) can donate blood cells to their son if his blood type is AB+ (1 pt)?

12. Which parent(s) (Frank, BOF) can donate blood cells to their son if his blood type is O- (1 pt)?

13. Which parent(s) (Frank, BOF) can donate blood cells to their son if his blood type is A+ (1 pt)?

When you donate blood, your blood is often spun (in a centrifuge, just like in the Hershey/Chase blender experiment!) to separate the heavier red blood cells from the lighter plasma. These two components can be given to recipients separately. *Plasma* donation is different than cell donation; individuals with blood type AB+ are considered the "universal plasma donor" because their *plasma* contains no antibodies against A, B, or Rh molecules.

14. Which parent(s) (Frank, BOF) can donate *plasma* to their son if his blood type is AB+ (1 pt)?

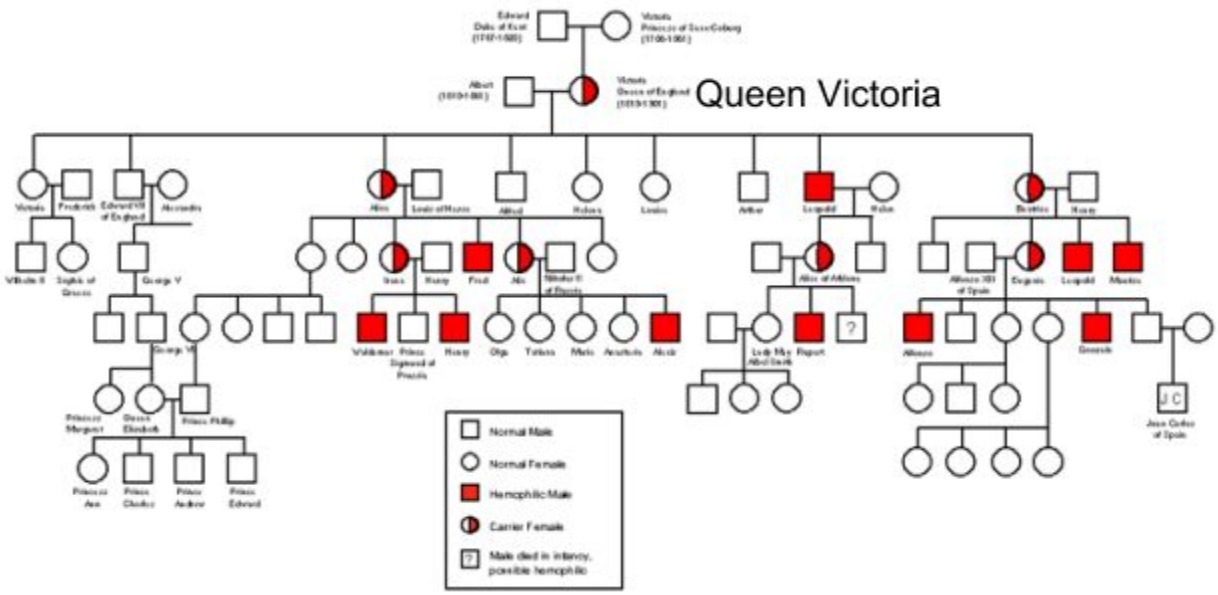
15. Which parent(s) (Frank, BOF) can donate *plasma* to their son if his blood type is O- (1 pt)?

16. Which parent(s) (Frank, BOF) can donate *plasma* to their son if his blood type is A+ (1 pt)?

BLOOD

Hemophilia is genetic disorder in which the blood does not clot properly. This was a disease rampant in European royal families of the 1800s-1900s, beginning with Queen Victoria. There are two forms of hemophilia, A and B, caused by mutations in two different genes. However, the two genes are located on the same chromosome and have similar **inheritance patterns**.

Below is a **pedigree**, a "family tree" that can help us parse out the inheritance pattern for genetic diseases. Male members are squares, while female members are circles. Marriages between two individuals are drawn as horizontal lines between individuals, and their children are branched below them. Those unaffected by the disease (hemophilia) are uncolored, those affected are colored red, and individuals thought to be carriers are half colored:



17. Based on the pedigree, on what chromosome do you think genes involved in hemophilia A and B are carried and why? Is this hemophilia allele **dominant** or **recessive** (2 pts)?

WEREWOLVES

The genetic disease that leads to Ambras syndrome causes sufferers to grow hair all over their bodies. This has earned Ambras syndrome the nickname "werewolf syndrome."

Ambras is often caused by mutations in the gene *TRPS1*, which encodes a **transcription factor** that controls the expression of many other genes (another transcription factor we discussed in class is the *SRY* gene, located on the male Y-chromosome and responsible for

"maleness" in humans). The *TRPS1* **locus** is on human chromosome 8 (an **autosome**) and mutations that cause Ambras are inherited in a **dominant** manner.

TRPS1 regulates the expression of genes important for hair and bone development. One important gene regulated by the TRPS1 **transcription factor** is *Sox9*, which *itself* encodes a **transcription factor** that regulates genes active in hair follicle cells.

Below is a diagram of the *Sox9* gene:

18. In what region of the *Sox9* gene do you expect the TRPS1 **transcription factor** to bind and how will this affect expression of the SOX9 protein (2 pts)?

WITCHES AND WIZARDS

In the fictional Harry Potter world, wizards and witches are in the minority, compared to non-magic muggles. The magic allele (m) is **recessive** to the muggle allele (M).

19. Hermoine is a witch born to muggle parents. What must be Hermoine's parents **diploid genotypes** (1 pt)?

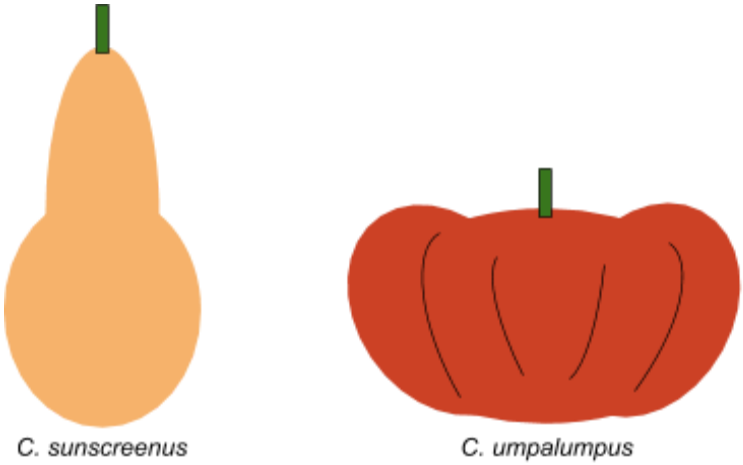
Mother: _____ Father: _____

20. Do you think the M/m gene **locus** is located on an **autosome** or **sex-chromosome**? Why (1 pt)?

21. The Malfoys and other "death eaters" believe that "pure-blood" wizards and witches are superior to "half-blood" or "muggle-born" wizards and witches. Pure-bloods are **true-breeding**, just like Mendel's peas. If the death eaters succeeded in creating a pure-blood race, what would be the prevalence of the M allele in this population (1 pt)?

PUMPKINS

Cucurbita is the genus that includes squash, gourds, and pumpkins. All species of *Cucurbita* are **diploid** and have 20 chromosomes (40 total chromosomes in a **diploid** cell), although today we will simplify to two. Because species have the same number of chromosomes, they can easily **hybridize** with each other. Take, for example, these two **true breeding** *Cucurbita* species:



C. sunscreenus *C. umpalumpus*

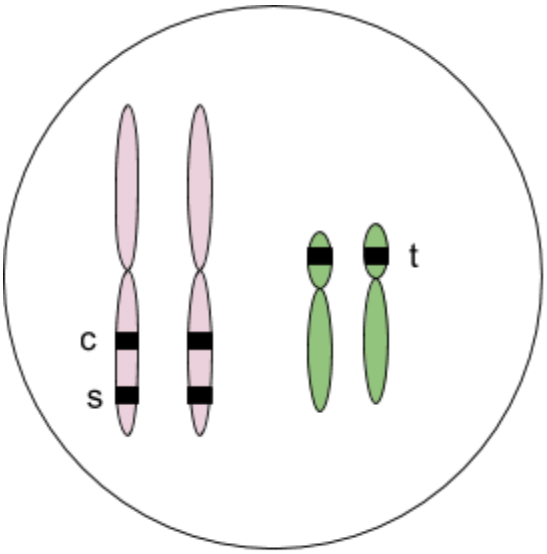
C. sunscreenus has alleles that give it a smooth (t), pale skin (c) and elongated shape (s).

C. umpalumpus has alleles that give it ribbed (T), deep orange skin (C) and a squat shape (S).

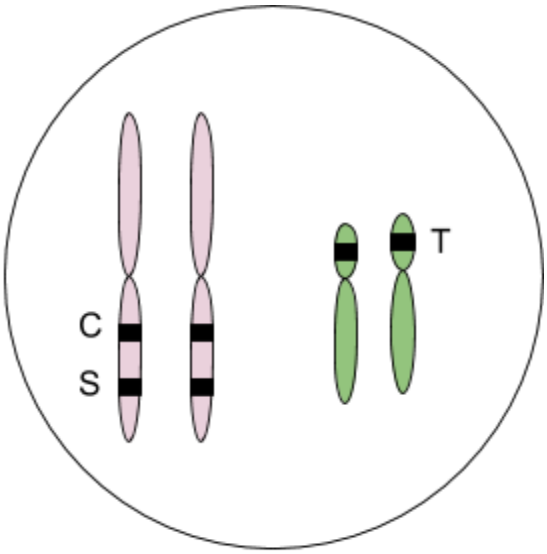
The ribbed, deep orange, and squat alleles are all **dominant** to the smooth, pale, and elongated shape alleles.

The genes that control skin color and body shape are located on the **same chromosome** (they are ***linked***), while the gene that controls skin texture is located on a different chromosome (it is ***unlinked*** to the genes that control skin color and body shape).

Below are the **diploid genotypes** of both species (we are simplifying to just two chromosomes):



C. sunscreenus: ccsstt



C. umpalumpus: CCSSTT

22. What will be the **genotype** and **phenotype** of the **hybrid F1** offspring between *C. sunscreenus* and *C. umpalumpus* (1 pt)?

Full diploid genotype: _____

Phenotype: _____

23. What will be the **haploid gamete genotypes** produced by the F1 hybrid offspring (4 pts)? Here you should apply the principles of **linkage** as well as Mendel's laws of **Segregation** and **Independent Assortment**!

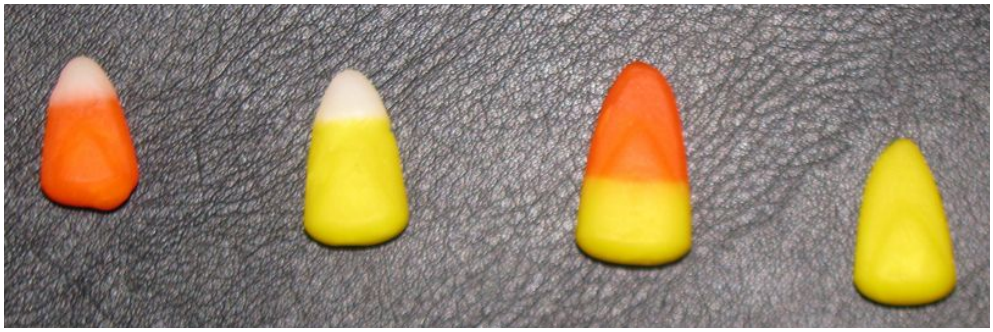
24. The hybrid F1 offspring now **selfs**. DRAW and fill out a Punnett Square below to reveal the **genotypes** of the **F2** generation (3 pts):

25. What is the distribution (as a fraction of total offspring) of all **phenotypes** of the **F2** generation? Do you see trait combinations you did not see in the parental (*C. sunscreenus* and *C. umpalumpus*) or F1 hybrid generation (2 pt)?

CANDY CORN

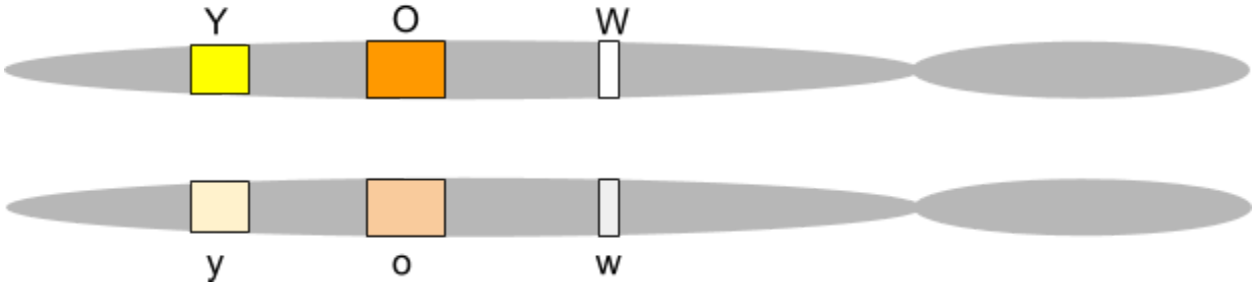
There are only maybe 12 ingredients in candy corn. Such a simple candy… candy corn only has one chromosome (the physical location of those 12 genes)! Today we are only concerned with the three pigment-producing genes. A **wild-type** candy corn has the **phenotype** below:

However, we occasionally see other color combinations of candy corn:



Because the candy corn **genome** consists of only one chromosome, we know that the variation in phenotypes cannot be due to Mendel's law of **Independent Assortment**, as we saw with the *Cucurbita* species above. Instead, this variation is due to **crossing over** during **meiosis I**, which removes **linkage** on the same chromosome.

A **wild-type diploid** candy corn has the following chromosomes:



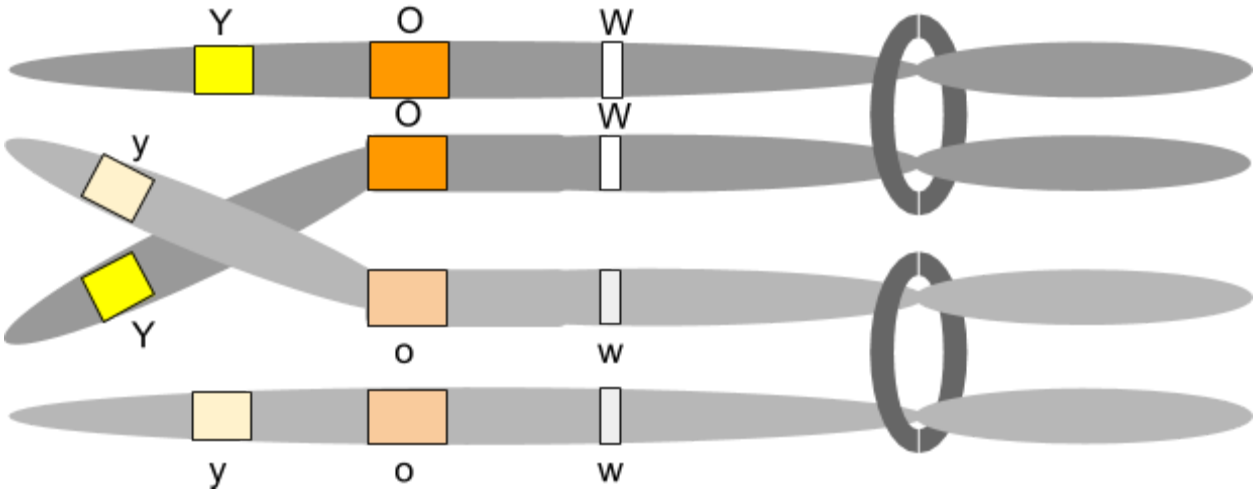
The Y, O and W alleles produce yellow, orange, and white pigment, respectively, and are **dominant** to the **loss-of-function** alleles y, o, and w, none of which produce any pigment. The wild-type candy corn is **heterozygous** at all of these **loci** and has the **diploid genotype:**

YyOoWw

Wild-type candy corn is not **true-breeding**. Every now and then a yyooww (no pigment **phenotype**) individual is produced, but it is removed from the population before packaging by sharp-eyed candy sorters!

How, then, do we genetically produce varieties with non-wild-type color combinations? Take, for example, this individual:

To have a **phenotype** lacking yellow pigment ("yellowless"), this corn's **diploid genotype** must be yy at the yellow **locus**, even though it still carries at least one dominant allele at the O and W loci. This occurred due to a **crossover** (also called a **chimera**) during **meiosis I** in one of the yellowless corn's parents:



Notice that, above, **DNA polymerase** has synthesized **sister chromatids**, which remain connected at the **cetromere**. During **meiosis I**, **homologs** align next to each other before they are separated into two cells. While they are aligned, homologs can **crossover**, exchanging genetic material and unlinking the Y allele from the O and W alleles.

26. What are the **haploid genotypes** of the 4 **different** gametes that will be produced after **meiosis** is complete in the above cell (2 pts)?

27. One of the gametes from question 27 will fuse to a gamete with the **haploid genotype** yow to produce the yellowless candy corn pictured above. Which gamete from #26 should you choose and what is the resulting **diploid genotype** of the yellowless individual (2 pts)?

28. Which gamete from #26 will fuse to a gamete with the haploid genotype yow to produce the a candy corn with the following non-wild-type **phenotype** (1pt)?

29. Draw the **meiosis I** chromosome **crossover(s)** that must occur to obtain **gametes** that, upon fertilization by a yow gamete, will give rise to the following individual. Label the **loci** of all the genes with specific **alleles** (4 pts)**.**

30. Draw the **meiosis I** chromosome **crossover(s)** that must occur to obtain **gametes** that, upon fertilization by a yow gamete, will give rise to the following individual. Label the **loci** of all the genes with specific **alleles** (4 pts).

Extra credit! (1 pt)

(not one of mine, but still great)

What genetics-themed pumpkin should I carve this year? Your answer should be:

a) Based on something we have discussed in class
b) Related to genetics
c) Awesome
d) Practical for my modest carving skills

I will carve the winner!