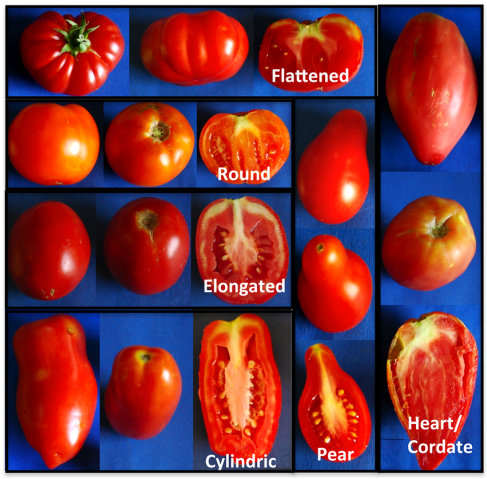
Bio393: Biomedical Genetics **Name:**_____
Midterm (5 pages, 64 points)

Question 1 (8 points):
Your farmer parents are so proud that their child goes to Northwestern that they want to make a purple pear-shaped tomato variety (to be named the *wildcat* strain). Pears come in six different shapes and ten different colors, as shown below. As a budding geneticist, you decide to help them out.



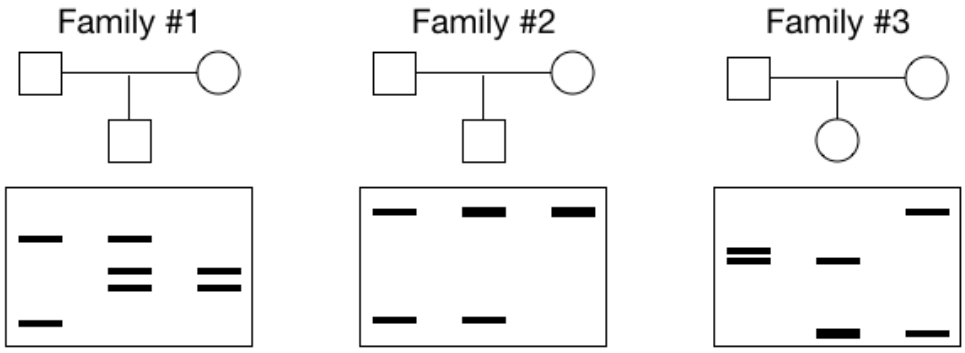
They order two true-breeding strains from an heirloom seed company: a round, purple variety and a pear, red variety. You cross the two strains to get all round, red tomatoes in the next generation. Just like peas, it's easy to self tomatoes, so you set up a large number of self crosses from that generation. In your entire garden, you only see tomatoes that are round or pear-shaped and red or purple. The majority of tomatoes are round and red. You search through 128 tomato plants to find two plants that have pear, purple tomatoes. When these pear, purple tomatoes are grown and selfed, they give rise to all pear, purple offspring. You've got your *wildcat* strain!

(a, 4 points): Describe the basic genetics of the color and shape traits.

(b, 4 points): Propose a hypothesis for how many genes are involved in the color and shape of tomatoes.

Question 2 (7 points):
Using linkage mapping, you determined the genetic distance between A and B is 100 cM. What is the minimum number of markers between A and B that you would need to make this estimate possible? Please explain your reasoning.

Question 3 (9 points):
You are interested in discovering genes that influence nondisjunction in human mothers. In order to select embryos with successful *in vitro* fertilization, clinics often look at the products of the union of sperm and egg. Some embryos have obvious visual defects and upon karyotype analysis have triploidy for one chromosome pair (an autosome). You obtain DNA from Mom, Dad, and the abnormal embryo to measure the genotype at a DNA marker on the triploid chromosome. The data from three trios (Mom, Dad, and embryo) are shown as agarose gels. Remember that Mom and Dad are diploid for the chromosome in question. The DNA bands show the different genotypes of the chromosomes by their different length products. Thicker bands show twice as much DNA as thinner bands.



Describe the nondisjunction in each family and any ambiguities.

Question 4 (22 points):

As an avid SCUBA diver, you are concerned about the loss of coral habitats caused by ocean acidification through rising carbon dioxide levels. You get a huge NSF grant to identify acid-tolerant staghorn coral. You can do coral crosses (once per year) and have a population of acid-sensitive coral. Coral are diploid, and this species is dioecious (males and females).

(a, 12 points) Write out a selection crossing scheme that will allow you to identify mutants with recessive acid resistance. You can use a mutagen. Describe which crosses are single-pair vs. bulk, when you apply the acidic ocean water selection, and what generation you score for your mutants.

(b, 10 points) A colleague in Australia finds a rare staghorn coral that seems to survive in the highly acidic ocean waters near his lab. His mutant has a dominant phenotype, and one of your mutants also has a dominant phenotype. You think his allele could be an allele of one of the same genes you mutated in your selection. Describe how would you determine if the Australian acid-tolerant true-breeding strain and your true-breeding strain are mutated in the same gene?

Question 6 (8 points):
You identify a very rare mutant in a screen for diatoms that fail to create radially symmetric shells. It has a dominant phenotype. You map the mutation to a chromosome and genomic interval where you have deficiencies (large deletions) and duplications. When you make heterozygotes with your mutant and either deficiencies or duplications, you get the same mutant phenotype with the same penetrance. Please explain how this rare mutation could affect gene function.

Question 7 (10 pts):
Your advisor would like you to isolate an amber suppressor (UAG nonsense codon suppressor) using the yeast *S. cerevisiae*. She gives you the following strains. Please select which strain would offer the most efficient way to screen for amber suppressor mutants and explain why. The *ACT1* and *IRA2* mutant phenotypes are different and do not interfere with each other.

act1-1	Amber nonsense allele of *ACT1*
ira2-87	Amber nonsense allele of *IRA2*
act1-1; ira2-87	*ACT1* and *IRA2* double mutant