

Answer all questions. Show your work clearly. You may be eligible for partial credit even if your answer is wrong.

1. (20 points) There are several recessive mutants in *Drosophila*, “mutant d” is *dumpy*, “mutant e” is *ebony* and “mutant f” is *forked* wing. A wild type **F1 female** was crossed to a **dumpy, ebony, forked wing male**. The following progeny were produced:

- What were the phenotypes of the inbred parents of the **F1 female**?
- Draw the metaphase I chromosomes of the F1 female with the genes marked in the appropriate positions.
- In your picture above (part b) indicate the distances between your genes.

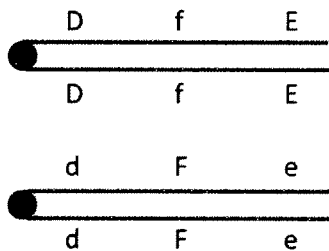
**Be Careful!! The gametes are not ordered for you; put the correct complimentary classes together.**

same same diff.

D	E	F	2	DCO	F is “odd man out” so it is in the middle.
D	E	f	424	NCO	
D	e	F	44		
D	e	f	37		
d	E	F	33		
d	E	f	36		
d	e	F	421	NCO	
d	e	f	3		
			1000		

A) Forked was crossed to dumpy ebony to produce the F1 female.

B) Take the two NCO gametes and put the genes in the correct order (F in the middle) and you have the two chromosomes of the F1 female.



C) Distance between D - F is:  $44 + 36 + 2 + 3 = 85 / 1000 = 0.085 = 8.5\%$

Distance between F - E is:  $37 + 33 + 2 + 3 = 75 / 1000 = 0.075 = 7.5\%$

Extra credit (6 points) What is interference and why does it occur? How much is there in this cross?

Interference refers to the phenomenon of often finding fewer double crossovers than expected. It is detected when viewing markers that are close together because one crossover often physically interferes with a neighboring one.

For question 1, observed DCO = 5; expected DCO =  $(0.085)(0.075)(1000) = 6.4$

$I = 1 - 5/6.4 = 0.22$ ; about 22% of the DCO are missing due to interference.

2. (18 points) In *Drosophila*, two mutations, **Bristle** (*B*) and **twisted** (*t*), are linked on chromosome 2. Bristle is a dominant allele and lethal in the homozygous state and twisted is recessive. A **Bristle, twisted individual** was crossed to an **inbred wildtype (not bristle and not twisted)**. Show the genotypes of this cross.

**Bb tt x bb TT**

You choose a **Bristle female** from the progeny of the cross above and want to map the genes in that female. Using the allele notation above, **give the genotypes of this Bristle female and the genotype and phenotype of the male** you would chosen as a mate for this mapping experiment.

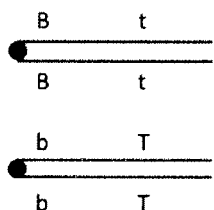
**The female Bristle female would be:**

**Bb Tt**

**To map it is best to cross to a homozygous recessive for the traits of interest so, the male would be a not bristle twisted:**

**bb tt**

If these two genes are 34 map units apart, **draw the chromosome of the female** of the above cross as they would appear in Metaphase 1 of meiosis with ALL alleles in the right positions. **Give genotype, phenotype and expected frequencies of the progeny of this cross.**



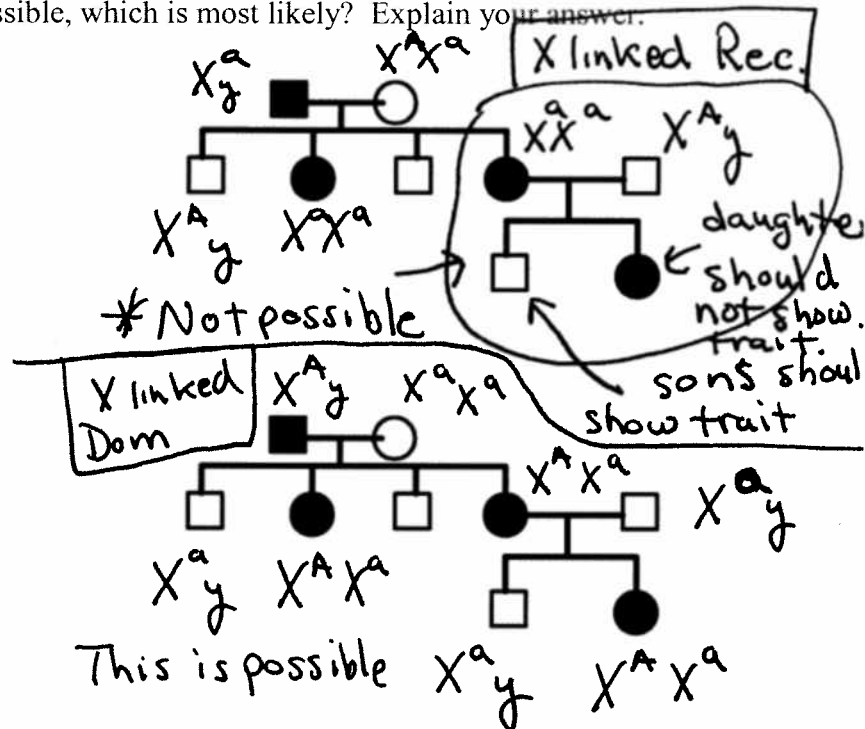
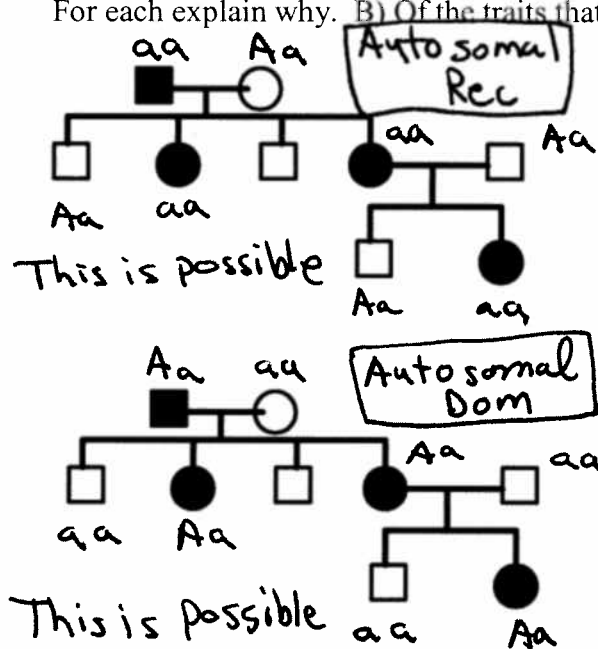
The gametes from the female are shown below. The male contributes a bt gamete:

Female gametes	Freq	male gametes	Progeny Genotype	Progeny Phenotype
<b>B T</b>	17%	<b>bt</b>	<b>Bb Tt</b>	<b>Bristle not twist</b>
<b>B t</b>	33%	<b>bt</b>	<b>Bb tt</b>	<b>Bristle twist</b>
<b>b T</b>	33%	<b>bt</b>	<b>bb Tt</b>	<b>not bristle not twist</b>
<b>b t</b>	17%	<b>bt</b>	<b>bb tt</b>	<b>not bristle twist</b>

3. (12 points) Recombination for a given length of DNA can vary across species, individuals, chromosome and regions of a chromosome. Briefly explain two examples discussed in class that demonstrate this variation in recombination rates. **There are many more than two examples in the answer below.**

While recombination is somewhat random, there are large differences between species, individuals and positions on a chromosome. There are hot and cold spots of recombination. Among species, humans average around 1% recombination for every 1 million base pairs. Yeast at the other extreme has about 1% for every 4000 bp. Differences among individuals can be even more dramatic. Male *Drosophila* have no recombination (during meiosis, homologous chromosomes line up but they do not recombine), while females do have recombination (about 1% for 50,000 bp). In humans chromosome do recombine in meiosis in males but at a much lower rate (but not zero) than females. Thus recombination maps based on male meiosis are shorter. Within individuals there are many hot and cold spots for recombination. Most of the Y chromosome in human males does not recombine with the X, but the pseudoautosomal region (PAR) at the end of the Y always has a crossover. Thus two genes just a short distance away in the PAR will look nearly unlinked while two genes far apart on the non-recombining part of the Y will look completely linked. Other regions of chromosomes have low recombination like the centromeres and telomeres.

4. (18 points) Consider the following pedigree involving a rare single human trait (the same pedigree has been repeated four times to give you working space to answer the question). A) Which mode(s) of inheritance **can be excluded** (autosomal dominant, autosomal recessive, X linked dominant, X linked recessive or Y linked)? For each explain why. B) Of the traits that are possible, which is most likely? Explain your answer.



Can not be Y linked because some females have the trait.

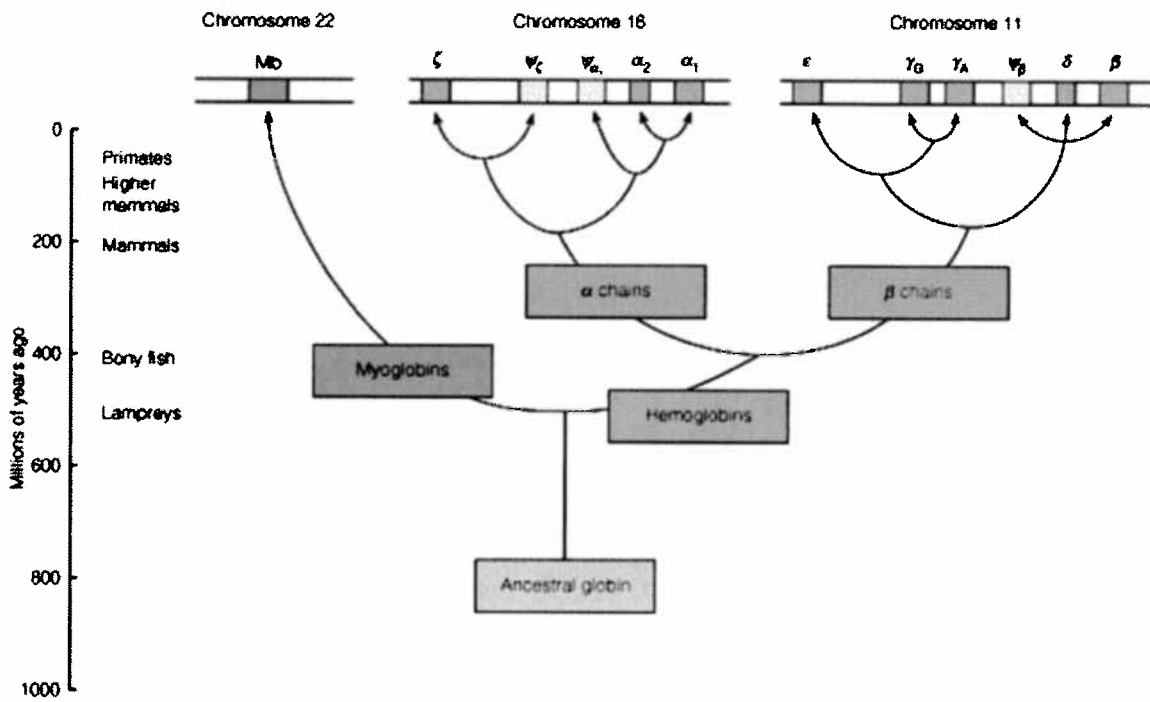
5. (16 points) How is sex determined in Drosophila? Be clear and precise and mention any critical genes involved.

Sex is determined by the ratio of X chromosome to sets of autosomes (or the ploidy level).

If the  $X/A = 1 = \text{female}$ ;  $X/A = 1/2 = \text{male}$ .

In typical diploids there are two sets of autosomes so for a typical female the  $X/A$  ratio would be  $2/2 = 1$  and for a typical male  $X/A = 1/2 = 1/2$

The *sisterless* (*sis*) genes on the X chromosome act as enhancers for the key *Sxl* gene and essentially "count" the number of X chromosomes in an individual; a female will have twice as many enhancers as a male. The *deadpan* (*dpn*) gene on an autosome "counts" the number of autosome sets. This *dpn* gene product acts as a repressor of *Sxl*. Females have more enhancer/repressor (i.e. higher  $X/A$  ratio) gene products and *Sxl* gene is turned on. This gene in turn turns on *transformer* which turns on *doublesex* resulting in female phenotype.



6. (16 points) We have been talking about different kinds of chromosomal changes in class. A) What kind of chromosomal changes must have occurred during the evolutionary history of globins shown above (be specific about the kind of change)? B) Are all the genes shown on the human chromosomes at the top of the figure functional? C) Are these genes different? D) Explain what they do and why we do or do not need them.

A) The ancestral globin gene has been duplicated many times. Copies have also been translocated or moved to different chromosomes.

B) Most of these genes are functional but a few (three) are pseudo genes ( $\Psi$  in the figure above) and are not functional; they were duplicated but a mutation later occurred to render it non-functional.

C) The protein products of all of these genes carry oxygen, but these genes are turned on at different times and in different tissues and have different functions. For example myoglobin gene is expressed in muscles, while some hemoglobins are expressed in embryonic tissues or in fetal liver or in adult bone marrow.

D) These different hemoglobins (always composed of 2 polypeptides from the alpha group and 2 from the beta group), have different affinities for oxygen and the fetal ones have a stronger affinity which allows them to capture oxygen from the mother's adult blood hemoglobins.

7. (10 points) Rainbow the cat is a calico. Carbon Copy was cloned from Rainbow and is the world's first cloned cat. Does Carbon Copy look like Rainbow? Explain why or why not.

Carbon Copy (CC) does not look like Rainbow. CC is not calico because the nucleus that came from Rainbow that was placed in an embryo to "start" CC was from a somatic cell. This somatic cell had one inactive X (apparently the X carrying the yellow allele for fur color) chromosome and that X was never reactivated (as it normally would be in a naturally formed embryo). Thus CC had no patches of yellow fur, only the gray-black patches. Even if the X had been reactivated CC probably would not look exactly like Rainbow since X inactivation is random and the same patches would not have been yellow or black.