Problem set 3

Problems 14.8, 14.9, 14.10, 14.12

1. The normal sequence of a certain *Drosophila* chromosome is ABC*DEFGHI, where the * represents the centromere. Aberrant chromosomes with the structures shown below were isolated. In parts a) and b), give the correct term for each type of rearrangement and show how each rearranged chromosome would pair with its normal homolog during meiosis I (label the chromosomes in your drawings).

a) ABC*DHGFEI

Region EFGH inverted-paracentric inversion.

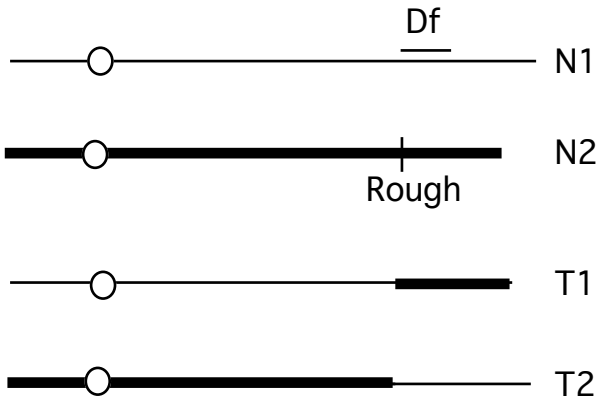
b) ABC*DEFEFGHI

EF duplicated-tandem duplication

c) Draw the meiotic products resulting from of a single crossover between F and G of the paired homologs of part a).

Centromeres connected by a dicentric bridge containing DEFGHD. Acentric fragment containing IHFRI.

2. The structures of two *Drosophila* autosomes (N1 and N2) and two reciprocal translocation chromosomes are illustrated below.



The circles represent the centromeres. The position of the wild-type Rough gene on chromosome N2 and the position of a small deficiency (*Df*) of chromosome N1 are shown. Mutations in Rough (ro^-) lead to a recessive adult rough eye phenotype. Individuals that are homozygous for the deficiency die as embryos. Assume that the Rough gene and the *Df* are very close to the translocation

breakpoints and that there is essentially no recombination between the breakpoint and Rough and between the breakpoint and the *Df*.

a) You cross T1/N1*(Df)*; T2/N2*(ro⁻)* females x T1/N1*(Df)*; T2/N2*(ro⁻)* males . What is the frequency of dead embryos produced in this cross?
b) What is the frequency of adults with rough eyes produced in the above cross?

In flies heterozygous for the reciprocal translocation chromosomes, alternate and adjacent-1 segregation patterns will generate dour types of gametes in equal proportions.

Adjacent: N1(df) T2 and N2(ro) T1
Alternate: N1(Df) N2(ro) and T1 T2

The easiest way to solve this problem, believe it or not, is with a Punnett square. You should get 5/16 live and 11/16 dead with no Rough eyed progeny. (This assumes that the T1T2/T1T2 homozygotes are alive. If the breakpoints in the translocations disrupt an essential gene, then 12/16 progeny would die.)

		Male gametes			
		N1(df) T2	**N2(ro) T1**	**N1(Df) N2(ro)**	**T1 T2**
Female gametes	**N1(df) T2**	**N1(df) T2 N1(df) T2 (dead)**	**N2(ro) T1 N1(df) T2 (wild type)**	**N1(Df) N2(ro) N1(df) T2 (dead)**	**T1 T2 N1(df) T2 (dead)**
	N2(ro) T1	**N1(df) T2 N2(ro) T1 (wild type)**	**N2(ro) T1 N2(ro) T1 (dead)**	**N1(Df) N2(ro) N2(ro) T1 (dead)**	**T1 T2 N2(ro) T1 (dead)**
	N1(Df) N2(ro)	**N1(df) T2 N1(Df) N2(ro) (dead)**	**N2(ro) T1 N1(Df) N2(ro) (dead)**	**N1(Df) N2(ro) N1(Df) N2(ro) (dead)**	**T1 T2 N1(Df) N2(ro) (wild type)**
	T1 T2	**N1(df) T2 T1 T2 (dead)**	**N2(ro) T1 T1 T2 (dead)**	**N1(Df) N2(ro) T1 T2 (wild type)**	**T1 T2 T1 T2 (wild type)**

3. Black body (b), reduced bristles (rb) and purple eyes (p) all map within a 5 mu interval on a *Drosophila* autosome. rb maps between b and p. A friend crosses

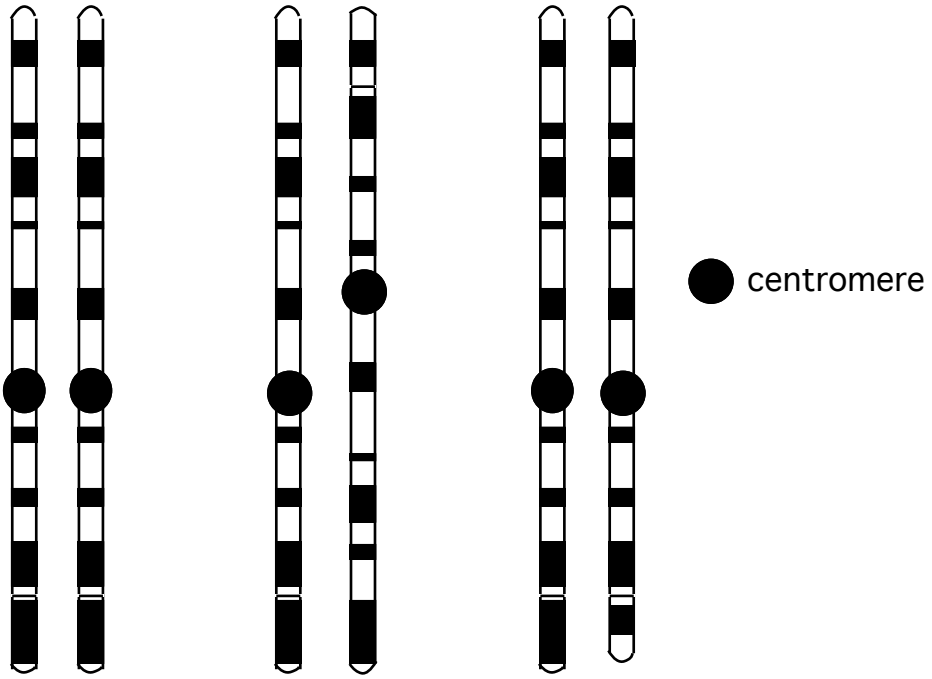
a wild-type female to a black body purple eyed male (b p/b p) and 109 of the F1s are wild type and the other 98 have black body and purple eyes. The friend suspects that the mutant chromosome in the female could contain two point mutations in the b and p genes, could be a deletion that removes the two genes or could be an inversion has breakpoints within the b and p genes. What do you think? How would you distinguish between these possibilities?

The three possibilities are: (1) point mutations in b and p; (2) deletion that removes b and p; (3) inversion with break points in b and p. (1) is unlikely because we would expect b and p recombinants in the F1. You can distinguish between 2 and 3 by crossing the P0 female with rb/rb males. A deletion would produce 50% progeny with reduced bristles, whereas an inversion would produce only wild-type progeny.

4. Test crosses show that a female *Drosophila* is heterozygous for mutations in two closely linked autosomal genes, *black body*, which results in a black colored body, and *short bristles*, which results in shortened bristles along the body. The genes are separated by six map units. What cross would you conduct to determine whether your strain carries a deletion that removes both genes or two independent point mutations in the *black body* and *short bristles*genes. You only have the heterozygous female and a doubly mutant strain carrying independent mutations in the *short bristles* and *black body* genes. Describe the results of the cross if the mutation is a deletion and if it is a double mutant.

Cross the heterozygote to the homozygous double mutant and look for recombinants.The presence of bb or sb animals in the offspring would indicate a double mutant.

5. Abe and Claris Norman have been trying to have children for the last few years and this has resulted in two miscarriages and then recently, an abnormal child that died shortly after birth. At the birth of the child, the Normans met with a genetic counselor who took tissue samples from the child before it died. The counselor prepared metaphase chromosome spreads of the sample and found cytogenetic differences between the child's chromosome 22 pair. The counselor then prepared metaphase chromosomes from both parents. The chromosome 22 spreads for both parents and the child are shown below.



Claris Norman Abe Norman child

a) Do the parents have an abnormal chromosome 22, and if so which parent(s) and what is the nature of the abnormality (be specific)?

Abe has a pericentric inversion

b) How did the child's abnormal chromosome arise?

When Abe's chromosomes paired they formed an inversion loop. Crossing over produced the partially duplicated, partially deleted chromosome that his child inherited.

c) Why is the child abnormal?

The deleted a duplicated portions of chromosome 22 resulted in genic imbalance.

d) Do you think this couple is capable of having additional abnormal children? Why ?

Yes, if crossing over occurs in the loop.

e) Do you think this couple is capable of having normal children? Why?

Yes, if crossing over occurs outside the loop.

6. An adult woman has the 14:21 Robertsonian translocation that we discussed in class. Assuming that alternate and both types of adjacent segregation patterns occur with equal frequency, what is the probability that she will give birth to a normal child? What is the likelihood that she will have a miscarriage?

The probabilities are predicted in Figure 14.22 of your textbook. Alternate segregation will produce balanced gametes and result in normal children. Adjacent-1 segregation will produce unbalanced gametes, but one will result in a live birth, but the child will have Down syndrome. The remaining adjacent-1 gamete and the adjacent-2 gametes will be unbalanced and will result in miscarriages. Thus, she has a 2/3 chance of giving birth to a normal child and 50% of her pregnancies will result in miscarriages.