

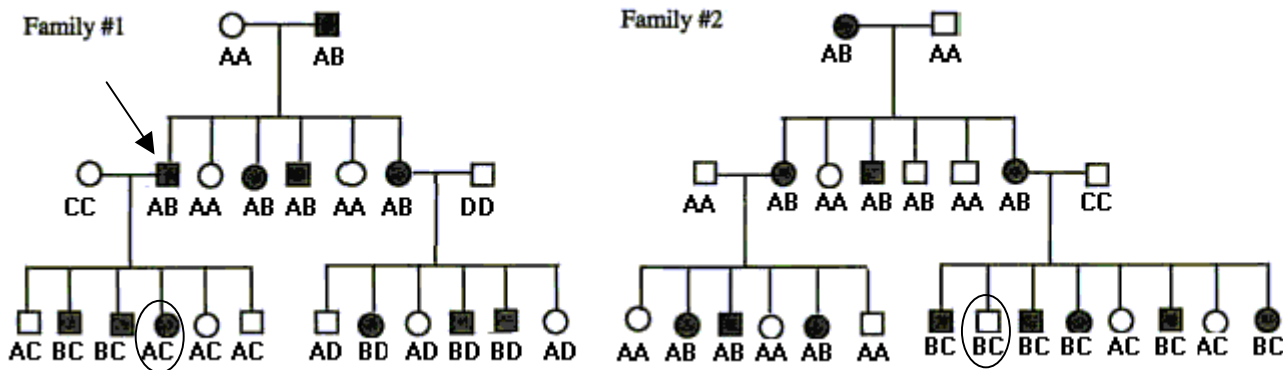
Biology 305 Genetics  
 Problem Set 4

Name \_\_\_\_\_

By printing your name above you acknowledge that while there may have been group discussions of these problems, the work below is yours.

I want to encourage you to work together on these problems, but make sure you understand how to solve each problem on your own. Show all of your work. No credit will be given for answers without work. If a question requires you to determine a genotype show how you obtained it. Please write neatly. If I can't read it, I can't grade it. The homework is due Monday, April 5th before class begins. If it is handed in after class has started there is a 5-point deduction and a further 5 points for each day it is late.

You are studying a dominantly inherited Hobbit syndrome that produces excessively hairy feet. You have begun to examine the segregation of this dominant trait with a microsatellite marker (with alleles A, B, C, D) in the following two human pedigrees. Individuals with the Hobbit mutation are indicated with filled squares and circles. Alleles at the marker locus are indicated by the letters under each individual.



A. What does it mean for a marker locus to be informative when looking for linkage in a pedigree?

**It means that you can identify non-recombinants and recombinants in the progeny.**

B. How many individuals in Family 1 can be considered in calculating LOD scores?

**12, we can't count the second generation because we don't know whether they are recombinants or non-recombinants. The father at the top of the pedigree has the trait and the A and B alleles. But we do not know which allele he got from his affected parent so the A or B allele could be linked in his chromosomes. But, we do know that the male with the arrow got B and the Disease from his father and thus the two are linked in his DNA, thus we can use his progeny.**

C. How many individuals in Family 2 can be considered in calculating LOD scores?

**14 for the same reason as above.**

D. Which allele at the marker locus is possibly linked (in phase) with the dominant Hobbit mutation? **B**

E. Are any individuals in the pedigrees recombinant? If so, circle them

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Hereditary elliptocytosis is an autosomal dominant anemia in which red blood cells have an abnormal elliptical shape and a shortened life span, which results from the increase destruction of these cells in the spleen. You have two families which are transmitting the trait. You have calculated the following LOD scores in the two families in a linkage analysis between the alleles of the Rh Blood Group locus on chromosome 1 and the elliptocytosis locus.

Recombination Distance ( $\Theta$ )	Family 1 LOD Scores	Family 2 LOD Scores
0.05	4.74	-2.9
0.10	5.44	-2.00
0.15	5.35	-1.73
0.20	4.89	-1.34
0.25	4.2	-1.12
0.30	3.39	-0.91
0.35	2.49	-0.30
0.40	1.54	-0.02
0.50	0	0

- A. What can you conclude about the linkage of the Rh blood locus and the elliptocytosis locus in both families? (Be sure to address whether the two loci are linked and the most probable map distance in both families)

Family one Answer:

**The Rh locus appears to be linked at 10 map units away. The LOD score at 10 map units is the highest significant score above 3.**

Family two Answer:

**The Rh locus appears to be unlinked in Family 2 because all of the LOD scores are negative.**

- B. Propose a genetic explanation for the two different results you have found with these two families

**Locus Heterogeneity: the two families have the same phenotype, but it is due to mutations in different genes in each family.**