

Human Problem Set
Due May 1, 2009

1. Compare and contrast the following pairs of terms in less than 3 sentences.

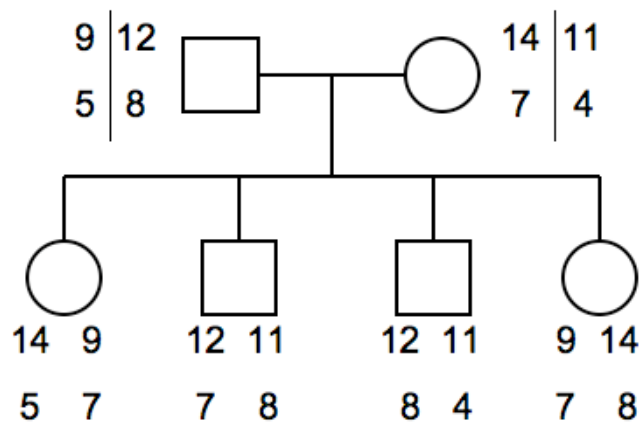
- A. Genetic linkage vs linkage disequilibrium
- B. Haplotype vs SNP
- C. Linkage study vs association study

2. You are analyzing linkage of two STR (simple tandem repeat) markers on Chromosome 16, D16S539 and D16S476. You do PCR analysis using samples from 2 families to determine the number of repeats at each loci, shown below on the pedigrees. Note that the two alleles for D16S539 are the top two numbers for each individual, and the alleles of D16S476 are the bottom two numbers. The vertical line indicates the phase (e.g. for the father of family A, the allele with 9 repeats at D16S539 is on the same chromosome as the allele with 5 repeats at D16S476).

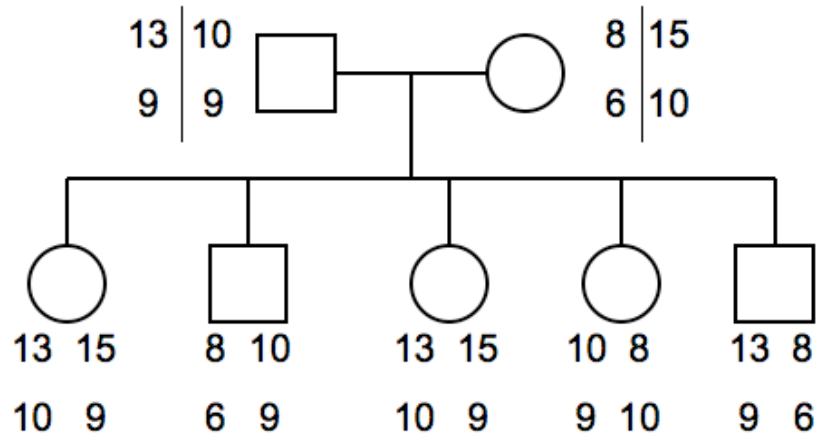
For all the children in both families, write the genotypes with the phase (if it can be determined), and label the meioses as recombinant or non-recombinant. Write out the formula for the LOD score for each family (you don't have to solve it). Assume

$$\hat{\theta} = \frac{\# \text{ recombinant alleles}}{\# \text{ meioses}}$$

Family A:



Family B:



C: When you incorporate the data from both families, what is the LOD score between these two marker loci? (You don't have to give me a number for the LOD score—you can express it in a formula or tell me how you would determine it).