

Biostatistics 666

Problem Set 1

Due Tuesday, January 24, 2005

1. Consider a population where allele frequencies differ between the sexes. Assume that there are equal numbers of males and females and that genotypes occur in Hardy-Weinberg proportions within each sex. Focus on a single di-allelic marker in this population. The marker has allele frequency  $p_M = p + \Delta$  in males and  $p_F = p - \Delta$  in females, where  $p = (p_F + p_M)/2$ .
  - a) Calculate offspring genotype frequencies after one generation of random mating.
  - b) How do genotype frequencies differ from those expected under Hardy-Weinberg equilibrium?
  - c) How many additional generations are required before Hardy-Weinberg equilibrium is reached?
2. In a sample of 80 individuals, 75 homozygotes for allele A, 2 homozygotes for allele B and 3 heterozygotes were observed. Conditional on the number of observed A and B alleles, answer the following questions:
  - a) What is the probability of this particular sample configuration?
  - b) What is the probability of observing an equal or greater number of heterozygotes?
  - c) What is the probability of observing a smaller number of heterozygotes?
  - d) What is the chi-squared statistic for Hardy-Weinberg equilibrium?
3. Consider two loci in disequilibrium in a large population. Assume that the recombination fraction between the two loci is 0.01. In how many generations do you expect the disequilibrium coefficient  $D$  to be halved?
4. Consider the following set of haplotype frequencies:
$$p_{AB} = 0.3, p_{Ab} = 0.2; p_{aB} = 0.1; p_{ab} = 0.4$$
  - a) Calculate  $D$ ,  $D'$  and  $\Delta^2$  between the two markers.
  - b) What is the probability that allele A will be present in a chromosome that carries allele b?
  - c) What is the maximum possible value of  $r^2$  for this marker pair?

