

Algorithms in Genome Research  
Winter 2011/2012

Exercises

Number 11, Discussion: 2012 February 3

1. Remember and explain a few of the terms (hopefully) mentioned in class:
  - genotype, haplotype
  - segregating site
  - genealogy
  - infinite sites model
  - four-gametes test
2. Give general formulas for the following questions. If this is difficult, enumerate the solutions for small examples.
  - (a) For  $k$  markers of two alleles each, how many different haplotype vectors are possible?
  - (b) If the haplotypes come in blocks of 10 sites each, how does this decrease the number of different haplotype vectors?
  - (c) For  $m$  founder sequences and  $l$  recombination hot spots, how many haplotype vectors are possible (under the assumption that recombinations only occur at hot spots)?
  - (d) For  $k$  individuals, what is the maximum number of different segregating sites such that the four-gametes test does not fail?
3. Let the following haplotype matrix be given (circles represent the presence of a mutant allele):

	1	2	3	4	5	6	7	8	9	10
A	—O—	————	—O—	—O—	—O—	————	————	—O—	—O—	—
B	————	————	—O—	————	————	————	—O—	————	————	————
C	—O—	—O—	————	—O—	—O—	————	—O—	—O—	—O—	—
D	————	————	—O—	————	————	————	————	—O—	————	————
E	—O—	—O—	————	————	—O—	————	————	—O—	————	————
F	————	————	————	—O—	————	————	—O—	————	—O—	————

- (a) Find the maximal regions around each segregating site, not violating the four-gametes test.
- (b) Draw the local trees of the segregating sites.
- (c) Assume that individuals  $C$  and  $E$  are the cases, the other the controls. Which of the segregating sites show highest evidence for association with the disease?