

Algorithms in Genome Research

Winter 2025/2026

Exercises

Number 8, Discussion: 2026-January-9

1. Investigate: What is the difference between linkage and association?
2. 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—	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 for these regions.
 - (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?
3. Discuss: Can pedigree (family) information help in the analysis of whole-genome association studies?
4. Give general formulas for the following questions. If this is difficult, enumerate the solutions for small examples.
 - (a) For n biallelic sites, represented by the columns of a $k \times n$ binary haplotype matrix, how many *different* haplotype sequences (rows) are at most possible?
 - (b) If the haplotypes come in blocks of 10 sites each, 2 variants each, how does this decrease the number of *different* haplotype vectors?
 - (c) For l founder sequences and m recombination hot spots, how many haplotype vectors are possible (under the assumption that recombinations occur only at hot spots)?
 - (d) For k haplotype sequences, what is the maximum number $n_{\max}(k)$ of *different* configurations at segregating sites (columns) such that the four-gametes test does not fail?