

PHS 555: Section on Statistical Genetics

(due 25 January, 2017)

Homework 1: Linkage Analysis

Problem 1 – Mendel's First Laws

Suppose that there are two individuals, whose genotypes are ABC|abc and abc|abc at three genes A, B, and C, respectively. These two individuals are crossed to generate 20 progeny with the following genotypes:

Progeny	Gene A	Gene B	Gene C
1	Aa	Bb	Cc
2	Aa	bb	cc
3	Aa	Bb	Cc
4	Aa	bb	cc
5	Aa	Bb	Cc
6	Aa	Bb	Cc
7	Aa	Bb	Cc
8	Aa	Bb	cc
9	Aa	Bb	Cc
10	Aa	Bb	Cc
11	aa	Bb	Cc
12	aa	bb	Cc
13	aa	bb	Cc
14	aa	bb	Cc
15	aa	bb	cc
16	aa	bb	Cc
17	aa	bb	Cc
18	aa	bb	cc
19	aa	bb	cc
20	aa	Bb	Cc

This is a backcross design. Address the following questions:

Question 1: Does the segregation of gene A follow Mendel' First Law?

Question 2: Does the segregation of gene B follow Mendel' First Law?

Question 3: Does the segregation of gene C follow Mendel' First Law?

Problem 2 – Linkage analysis

Based on the above data, address the following questions:

Question 4: What is the recombination rate between genes A and B?

Question 5: Is the linkage between genes A and B significant at the 5% significance level?

Question 6: What is the recombination rate between genes A and C?

Question 7: Is the linkage between genes A and C significant at the 5% significance level?

Question 8: What is the recombination rate between genes B and C?

Question 9: Is the linkage between genes B and C significant at the 5% significance level?

Problem 3 – Gene order determination

Using the above data, determine the most likely order based on the estimated likelihood values.

Question 10: What is an optimal gene order among genes A, B and C?

Problem 4 – Map function

After an optimal order is determined, address the following questions:

Question 11: What are the commonly used map functions?

Question 12: What is a map distance between marker A and B?

Question 13: What is a map distance between marker A and B?

Question 14: What is a map distance between marker A and B?

Problem 5 – Marker analysis in the F2

Two genes A and B form nine genotypes in the F2 family, with observations as follows:

	BB	Bb	bb
AA	n22=20	n21=17	n20=3
Aa	n12=20	n11=49	n10=19
aa	n02=3	n01=21	n00=19

Using this data, address the following questions:

Question 15: Does the segregation of gene A follow Mendel' First Law?

Question 16: Does the segregation of gene B follow Mendel' First Law?

Question 17: What are the expected frequencies of each genotype in terms of the recombination fraction, r ?

Question 18: Are these two markers linked?

Question 19 (optional): What is the recombination rate between these two markers?

All 18 required questions are weighted equally.