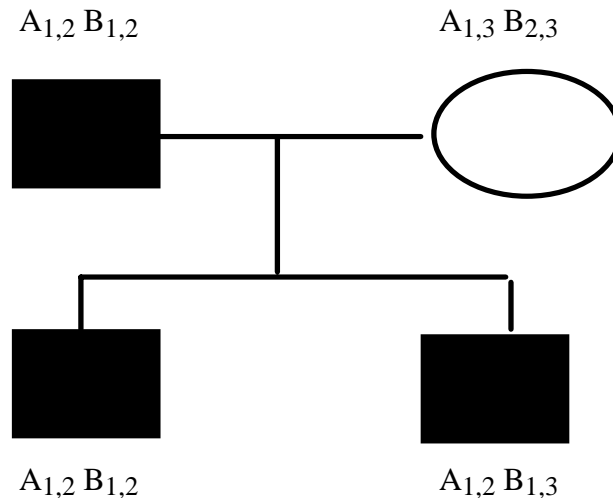


Homework due at the **beginning** of class on **Thursday, Nov. 20, 2008**.

**The exam will be Dec. 4. Homework will be returned on Nov. 25 and discussed Dec. 2.**

**Homework questions.** Please provide your answers on a separate sheet. These questions are mostly from lectures 15 and 16.

1. (2 points) Examine the following pedigree.



The  $A_1$  alleles in the two brothers are identical by state (this just means that they are both  $A_1$ ). The same is true of  $B_1$ . In either case can you infer that they are identical by descent? In other words, which alleles are identical by descent?

- You don't know about either  $A_1$  or  $B_1$ .
- You can conclude that  $A_1$  is identical by descent but don't know about  $B_1$ .
- You can conclude that  $B_1$  is identical by descent but don't know about  $A_1$ .
- You can conclude that both  $A_1$  and  $B_1$  are identical by descent.

(Questions 2-6): Consider two populations, 1 and 2, that differ at two unlinked loci, A and B. In each population a specific allele is fixed at each locus (*i.e.* all individuals in population 1 are homozygous for  $A_1$  and  $B_1$  -- they have the genotype  $A_{1,1} B_{1,1}$  -- while all individuals in population 2 have the genotype  $A_{2,2} B_{2,2}$ ).

First, you cross a single male from population 1 with a single female in population 2. Of course, all of the F1 progeny are heterozygous at both loci,  $A_{1,2} B_{1,2}$ .

- (1 point) Is locus A at Hardy-Weinberg equilibrium in the F1 generation (your answer would be the same for locus B)?
- (1 point) What is the expected frequency of each of the four possible gametes transmitted from the F1 to the F2. The possible genotypes are  $A_1 B_1$ ,  $A_1 B_2$ ,  $A_2 B_1$  and  $A_2 B_2$
- (1 point) Do the two alleles  $A_1$  and  $B_1$  show genetic association in this F1 generation? (consider the haplotypes that are transmitted by this F1 generation to the F2 generation).

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5. (1 point) What is the expected frequency of each of the nine possible genotypes in the F<sub>2</sub> progeny (assuming random mating among the F<sub>1</sub>)?

The possible genotypes are:

A<sub>1,1</sub> B<sub>1,1</sub> ; A<sub>1,1</sub> B<sub>1,2</sub> ; A<sub>1,1</sub> B<sub>2,2</sub> ;

A<sub>1,2</sub> B<sub>1,1</sub> ; A<sub>1,2</sub> B<sub>1,2</sub> ; A<sub>1,2</sub> B<sub>2,2</sub> ;

A<sub>2,2</sub> B<sub>1,1</sub> ; A<sub>2,2</sub> B<sub>1,2</sub> and A<sub>2,2</sub> B<sub>2,2</sub>?

6. (1 point) Is locus A at Hardy-Weinberg equilibrium in the F<sub>2</sub> generation (your answer would be the same for locus B)?

Later (questions 7-9), you allow a large and equivalent number of individuals from the two populations -- for example, 500 males and 500 females from population 1 and 500 males and 500 females from population 2 -- to mate at random (and they do mate at random).

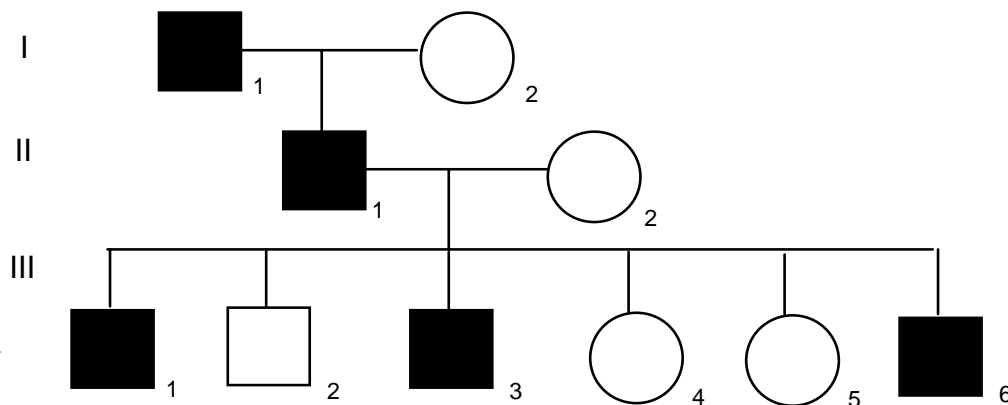
7. (1 point) Is locus A at Hardy-Weinberg equilibrium in the "G<sub>1</sub>" generation?

8. (1 point) What is the expected frequency of each of the four possible gametes transmitted from the G<sub>1</sub> to the G<sub>2</sub>. The possible genotypes are A<sub>1</sub> B<sub>1</sub>, A<sub>1</sub> B<sub>2</sub>, A<sub>2</sub> B<sub>1</sub> and A<sub>2</sub> B<sub>2</sub>

9. (1 point) Do the two alleles A<sub>1</sub> and B<sub>1</sub> show genetic association in this G<sub>1</sub> generation?

This pedigree shows a family affected by an autosomal dominant genetic disease.

Genotypes for three linked markers, A, B and C, are shown



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The genotypes are:

I-1        A<sub>1,2</sub> B<sub>1,2</sub> C<sub>1,2</sub>

I-2        A<sub>3,3</sub> B<sub>3,3</sub> C<sub>3,3</sub>

II-1       A<sub>1,3</sub> B<sub>1,3</sub> C<sub>1,3</sub>

II-2       A<sub>4,4</sub> B<sub>4,4</sub> C<sub>4,4</sub>

III-1      A<sub>1,4</sub> B<sub>1,4</sub> C<sub>1,4</sub>

III-2      A<sub>3,4</sub> B<sub>3,4</sub> C<sub>3,4</sub>

III-3      A<sub>1,4</sub> B<sub>3,4</sub> C<sub>3,4</sub>

III-4      A<sub>3,4</sub> B<sub>3,4</sub> C<sub>1,4</sub>

III-5      A<sub>3,4</sub> B<sub>3,4</sub> C<sub>3,4</sub>

III-6      A<sub>1,4</sub> B<sub>1,4</sub> C<sub>1,4</sub>

10. (1 point) Indicate the phase of alleles in individual II-1 by showing his haplotypes. There are four possibilities. They are

a) A<sub>1</sub> B<sub>1</sub> C<sub>1</sub> / A<sub>3</sub> B<sub>3</sub> C<sub>3</sub>

b) A<sub>1</sub> B<sub>1</sub> C<sub>3</sub> / A<sub>3</sub> B<sub>3</sub> C<sub>1</sub>

c) A<sub>1</sub> B<sub>3</sub> C<sub>3</sub> / A<sub>3</sub> B<sub>1</sub> C<sub>1</sub>

d) A<sub>1</sub> B<sub>3</sub> C<sub>1</sub> / A<sub>3</sub> B<sub>1</sub> C<sub>3</sub>

11. (1 point) What is the order of these three markers? (ABC, ACB or BAC)?

12. (2 points) Ignoring all of the other loci, calculate a lod score for linkage to of the disease to A with  $\theta = 0$

13. (2 points) Now, assume that this disease is only 75% penetrant. What is the lod score for linkage to A with  $\theta = 0$  under this revised model?

14. (1 point) Ignoring all other loci, what value of  $\theta$  would give the highest lod score for linkage of the disease to B?

15. (1 point) What is the value of that maximal lod score (for linkage of the disease to B at the value of  $\theta$  that gives the highest possible lod score)?

16. (1 point) Ignoring all other loci, what value of  $\theta$  would give the highest lod score for linkage of the disease to C?

17. (1 point) What is the value of that maximal lod score (for linkage of the disease to C at the value of  $\theta$  that gives the highest possible lod score)?

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### Study material.

This study guide covers material for the third exam:

Transcription, post-transcriptional gene expression, gene mapping, positional cloning, LOD scores, complex traits and types of allele.

This material was given in lectures Oct. 28 through Nov. 18. See the syllabus for readings.

### Be able to define, discuss, and explain the following concepts:

promoter	enhancer	core promoter
transcription factor	maternal effect genes	gap genes
epigenetics	nucleosomes	DNase sensitivity
DNase hypersensitivity	locus control region	core histones
mediator	DNA hemimethylase	<i>de novo</i> methylation
heterochromatin	euchromatin	position effect variegation
dosage compensation	X-inactivation	Barr bodies
Xist RNA	roX RNA	imprinting
macronucleus	micronucleus	RNA polymerase IV
polyadenylation signal	polyadenylation site	spliceosome
U1 snRNP	U2 snRNP	RNA editing
pan editing	Ran GTPase	importins
exportins	IRES	eIF2
eIF4	aminoacyl tRNA	peptidyl tRNA
NMD	exosome	EJC
RNAi	dicer	RISC
miRNA	siRNA	ubiquitin
FISH	STS	radiation hybrid mapping
informative meioses	LOD scores	autozygosity
haplotype	allelic association	linkage disequilibrium
complex traits	phenocopy	penetrance
QTL mapping	transmission disequilibrium test	sib pair analysis
null	amorph	haploinsufficient
allelic series	hypomorph	hypermorph
antimorph	neomorph	

Know features associated with active and inactive chromatin.

Be familiar with sex determination in *Drosophila melanogaster*.

Be able to explain the A, P and E sites of the ribosome.

Be able to summarize the key differences between prokaryotic and eukaryotic translation.

Be able to distinguish linkage maps, physical maps and cytological maps and know methods that allow each type of map to be correlated with the others.

Be able to look at a pedigree and identify recombinants and nonrecombinants, alleles identical by descent and informative vs. uninformative meioses. Be able to describe or recognize pedigrees showing maternal imprinting, paternal imprinting and anticipation.

Be familiar with the definition of a LOD score and how it might change with different models.