
Your name:

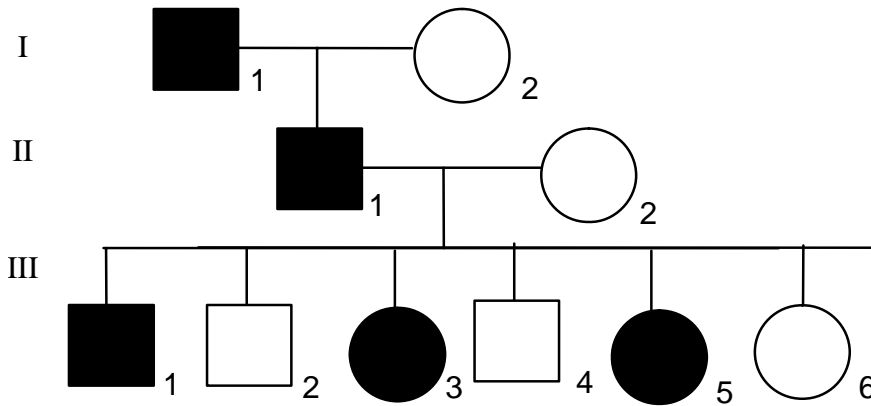
(4 points each). In each of the following there are two or more statements. One is true. Usually, it is taken directly from your textbook or from my lecture. In other cases, it might come from the scientific literature. Others have been modified so as to be untrue or misleading. Circle the letter next to the correct statement. In some cases, you might not know enough to be sure that statement is correct but you should be able to identify the others as bogus in some way.

1. a) Max homodimers are transcriptional **activators**, but the Myc-Max heterodimer activates a distinct set of genes involved in cell proliferation.
b) Max homodimers are transcriptional **repressors**, and is only when Myc and Max come together in a heterodimer that a transcriptional activator is formed.
2. a) **Epigenetics** is the study of heritable changes in gene expression that occur without a change in DNA sequence.
b) **Epistasis** is the study of heritable changes in gene expression that occur without a change in DNA sequence.
3. DNase I hypersensitivity sites are typically:
a) cell type-specific and within the coding region.
b) cell type-specific and intergenic.
c) found in all cells and within the coding region.
d) found in all cells and intergenic.
4. a) The **maintenance** methylase Dnmt1 exhibits predominantly hemimethylase activity.
b) The **de novo** methylase Dnmt1 exhibits predominantly hemimethylase activity.
5. a) Inhibitors of histone deacetylase **activate** the metallothionein I promoter.
b) Inhibitors of histone deacetylase **repress** the metallothionein I promoter.
6. a) The autoregulatory Sxl protein **promotes** the synthesis of more of itself through RNA splicing, resulting in a productive mRNA, thereby acting **as a self-reinforcing on/off switch**.
b) The autoregulatory Sxl protein **inhibits** the synthesis of more of itself through RNA splicing, resulting in less productive mRNA, thereby acting **to finely tune its level and activity**.
7. a) Nonsense mediated decay is a cellular mechanism of mRNA surveillance leading to degradation of **mRNAs** bearing premature stop codons.
b) Nonsense mediated decay is a cellular mechanism of mRNA surveillance leading to degradation of **proteins** truncated by premature stop codons.
8. a) Epigenetic imprints generally persist throughout the life of a mammal, but are erased during the passage of a gene through the germ line into the next generation.
b) Epigenetic imprints gradually decay during the life of a mammal and are rarely transmitted through the germ line into the next generation.
c) Epigenetic imprints generally persist throughout the life of a mammal, and are often transmitted through the germ line into the next generation.

Your name:

9. a) Autozygosity is a term used to refer to homozygosity **that results from mitotic recombination**.
 b) Autozygosity is a term used to refer to homozygosity **by descent from a common ancestor**.
10. a) Alleles at separate loci that are **associated with each other at a frequency significantly higher than expected by chance** are said to be in linkage disequilibrium.
 b) Alleles at separate loci that are **physically linked and common in a given population** are said to be in linkage disequilibrium in that population.
11. a) **Very few (less than 20) genes** that contribute to complex genetic traits have been identified and the prospects for identifying many more remain poor.
 b) **Dozens of genes** that contribute to complex genetic traits have been identified in the last few years by whole genome association mapping and our understanding of "genetic architecture" underlying these traits is rapidly improving.
13. a) In **forward** genetics, one starts with the mutant phenotype.
 b) In **reverse** genetics, one starts with the mutant phenotype.

This pedigree shows a family affected by an autosomal dominant genetic disease. Genotypes for three markers, A, B and C, are shown



The genotypes are:

I-1	A _{1,2} B _{1,2} C _{1,2}	III-1	A _{1,4} B _{1,4} C _{1,4}
I-2	A _{3,3} B _{3,3} C _{3,3}	III-2	A _{3,4} B _{3,4} C _{3,4}
II-1	A _{1,3} B _{1,3} C _{1,3}	III-3	A _{3,4} B _{1,4} C _{3,4}
II-2	A _{4,4} B _{4,4} C _{4,4}	III-4	A _{3,4} B _{3,4} C _{3,4}
		III-5	A _{3,4} B _{1,4} C _{1,4}
		III-6	A _{3,4} B _{3,4} C _{3,4}

Your name:

14. (9 points; three points per pair of markers) For each individual in the third generation (III-1, III-2, III-3, III-4, III-5 and III-6) indicate whether they are **recombinant**, **nonrecombinant** or **indeterminate** for each pair of **markers** in this pedigree (Fill in each of the 18 squares with **yes**, **no** or **maybe**).

individual	A B recombinant	B C recombinant	A C recombinant
III-1			
III-2			
III-3			
III-4			
III-5			
III-6			

15. (3 pt.) Considering only the data for marker locus A (i.e. ignoring B and C), what is the approximate lod score for linkage **between A and the disease gene** with $\theta = 0$?

16. (3 pt.) Considering only the data for marker locus **B**, what is the approximate lod score for linkage **between B and the disease gene** with $\theta = 0$?

17. (3 pt.) Considering only the data for marker locus **C**, what is the approximate lod score for linkage **between C and the disease gene** with $\theta = 0$?

18. (6 points) Other researchers in the field have reported that the disease gene is **not** present in an interval containing all three markers (A, B and C). Which of the following genetic maps is most likely, putting that information together with your pedigree (D stands for disease)?"

- a) ABCD b) DABC c) ACBD
 d) DACB d) BACD f) DBAC

19. (6 points). Do you expect Sxl to be subject to nonsense-mediated decay in *Drosophila* males? Explain.

20. (6 points). How are the phenomena you defined on the next page related to each other? Do they tend to be true of the same traits? What kind of traits? Give a few examples. [You may want to answer the questions on the next page first. If you need more space, use the back of this page, but be sure to tell us.]

Your name:

Define the following terms (4 points each):

incomplete penetrance

phenocopy

genetic heterogeneity

polygenic determination