

Your name: _____

Define the following terms (5 points each):

OMIM

Online Mendelian Inheritance in man, a database of human genetic diseases and the genes that are responsible.

twin spot

A pair of clones (of mitotic cells) each of which is homozygous for one of two homologs (distal to a point of recombination). Twin spots are derived from the reciprocal products of a single mitotic recombination event. They are normally found adjacent to one another and can be distinguished by recessive cell autonomous markers.

RefSeq

A special type of NCBI accession number for protein and nucleotide sequences. These accessions are non-redundant (one per protein or mRNA isoform) and are actively curated by NCBI staff.

codominant alleles

Alleles whose phenotypes are observed in the presence of other alleles ("alternative traits are both visible in the F1 hybrid"); this depends on the phenotype. Examples include antigenic variation (and almost any molecular phenotype). Codominance is distinct from semidominance, which case the heterozygote has an intermediate phenotype.

template

The template nucleic acid (RNA or DNA) is complementary to the nucleic acid being synthesized, and provides information that directs its synthesis.

Questions 1-4 are worth 5 points each. They concern two recessive mouse mutations, *clubpaw1* (*cp1*) and *clubpaw2* (*cp2*), that show the identical phenotype (fused digits on the paw). You first want to test whether these mutations are in the same gene. You cross *cp1* homozygotes with *cp2* homozygotes. For each of the four cases below, predict the frequency of mice with fused digits (assuming simple Mendelian ratios).

- 1) If *cp1* and *cp2* complement, you expect **0** % of the F1 mice to have fused digits.
- 2) If *cp1* and *cp2* do not complement, you expect **100** % of the F1 mice to have fused digits.
- 3) If *cp1* and *cp2* complement, you expect **43.75%** (7/16) of the F2 mice to have fused digits. This is the standard ratio for a two-hybrid cross.
(If the two genes are closely linked you would expect 50%)

Your name: _____

4) If *cp1* and *cp2* do not complement, you expect **100** % of the F2 mice to have fused digits.

[If you confused complementation with noncomplementation we gave you some partial credit]

5. (5 points) Three **wild-type** alleles, 1, 2 and 3, are present in a population in Hardy-Weinberg equilibrium and no other alleles are present at appreciable frequencies. Homozygotes for allele 1 represent 16% of the population, and homozygotes for allele 2 represent 9% of the population. What fraction of the population is homozygous for allele 3?

9%

6. (5 points) Allele 3 confers a semidominant increased risk of early-onset Alzheimer's disease. What fraction of the population shows the intermediate level of risk associated with **heterozygosity** for allele 3?

42%

7. (5 points) Here is the sequence of the template strand of a DNA fragment:

GTCGCCGTGCAATGATGTAGGCGACTATGGTTGA

Applying the standard **convention** for polarity (which end is 5' and which is 3'), which of the following would be the complementary, nontemplate, strand?

a) **TCAACCATAGTCGCCTACATCATTGCACGGCGAC**

Problems 8-10 (4 points each). In each of the following there are two or more statements. One is true (generally, it is taken directly from your textbook) and the others have been modified so that to be untrue or misleading. Circle, check or otherwise designate the correct statement. Ambiguous marks (checking both, placing a mark between the two statements, etc.) will be considered wrong.

8. **c** The **primer** DNA provides a terminus with a free **3'-OH** to which nucleotides are added during DNA synthesis.
9. **b** The same mutant allele can be dominant with respect to some traits and recessive with respect to others.
10. **a** The probability of two independent events occurring together is the **product** of the probabilities that each event will occur by itself.

Consider two genes: *ABC1* and *DEF2*.

When a wild-type (*ABC1* and *DEF2*) strain is crossed to a doubly mutant strain (*abc1* and *def2*) and sporulated, the following tetrads are observed:

83 tetrads with two *ABC1 DEF2* spores and two *abc1 def2* spores.

79 tetrads with two *ABC1 def2* spores and two *abc1 DEF2* spores.

Your name: _____

no tetrads of any other type.

11. (5 points) Are these two genes linked to each other (yes or no)? **no**

12. (5 points) Are these genes linked to the centromere (i.e. is *ABC1* centromere-linked? Is *DEF2* centromere-linked?) Either or both can be true. Select **one** of the following:

c Both are centromere-linked, but they are on different chromosomes (not linked to each other).

13. (10 points) Which of the following are true? None, all or any number can be true. (circle the letter corresponding to each that is true; this is really 5 true/false questions)?

T a Experimental recombination frequencies between two genes are never greater than 50%

T b The two terms centimorgan and map unit are interchangeable.

T c When two genes are linked parental ditypes will outnumber nonparental ditypes.

T d When two genes are unlinked the number of parental ditypes will be approximately the same as the number of nonparental ditypes.

F e When two genes are linked but 60 cM. or farther apart the number of parental ditypes will be approximately the same as the number of nonparental ditypes.

14. (3 points) Consider the following two possible alignments. *yfg* (your favorite gene) differs from each of the other two sequences by three substitutions in the region of alignment shown. To which sequence (1 or 2) is *yfg* more likely to be related? Explain.

1	RQNSRW
<i>yfg</i>	VQQLRW
2	IQNLKW

sequence 2. The reason is that V to I and R to K are conservative changes while V to R and L to S are not.

15. (5 points) The five amino acids E, F, G, N and W are pictured below. Which is which (put a number besides each of the five letters)?

E – glutamate glu **1**

F – phenylalanine phe **4**

G – glycine gly **2**

N – asparagine asp **3**

W – tryptophan trp **5**