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Define the following terms (5 points each):

blastp

BLink

codominant alleles

interference

1. (6 points) Explain loss of heterozygosity, including possible causes, mechanisms and consequences.

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Mams is a grandmother. She had 10 children, all with the same husband! Now, those children are all grown and each has had 10 children of their own (and with a single spouse in each case). Her 10 children are going to throw a party for her to celebrate this accomplishment (the 100<sup>th</sup> grandchild was born recently, and all 110 descendants are healthy). The celebration will be paid for by Guinness, which publishes the "Guinness Book of World Records" (there were several records broken here).

A representative from Guinness comes to you, as a genetics expert, with questions related to the fact that Mams has a genetic disease. She has hereditary hemochromatosis, an autosomal recessive condition causing an abnormal buildup of iron in the body.

2) (4 points) Two of her grandchildren (first cousins) are in love. If they marry and have a child, what is the risk that their child will inherit hereditary hemochromatosis? For this question, assume that the unrelated, unaffected parents of the cousins are not carriers.

3) (4 points) Hereditary hemochromatosis is not rare. In fact, 1/400 people are affected. Assuming Hardy-Weinberg equilibrium, what fraction of unaffected people do you expect to be carriers?

4) (4 points) In view of this number, what was the risk to each of Mam's grandchildren? I'm asking you to calculate the probability that one of her grandchildren would have hereditary hemochromatosis based solely on the fact that their grandmother was homozygous. In this case, however, the unrelated, unaffected, parent of the grandchild could be a carrier (with the probability calculated in problem 3).

5) (7 points) Despite the risk that you just calculated, **none** of the 100 grandchildren has hereditary hemochromatosis. The doctors have examined each one and they are quite sure. What the woman from Guinness really wants to know is "How likely is that?" What answer would you give her?

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(4 points each). In the following questions, one of the statements is true.

Circle either **a** or **b** (or **c**), next to the correct statement. Ambiguous marks (circling both, placing a mark between the two statements, etc.) will be considered wrong.

6. **a** A particular mutant allele is either dominant or recessive for all traits that it affects.  
**b** The same mutant allele can be dominant with respect to some traits and recessive with respect to others.
7. **a** The two terms centimorgan and map unit are interchangeable.  
**b** The two terms centimorgan and map unit refer to different units.
8. **a** In tetrad analysis, when two genes are **linked** the number of parental ditypes will be approximately equal to the number of nonparental ditypes.  
**b** In tetrad analysis, when two genes are **unlinked** the number of parental ditypes will be approximately equal to the number of nonparental ditypes.
9. **a** When two genes are unlinked the relative number of tetratypes and nonparental ditypes will be independent of their distance(s) from the centromere.  
**b** When two genes are unlinked the relative number of tetratypes and nonparental ditypes will depend on their distance(s) from the centromere.
10. **a** When two genes are linked, parental ditypes will significantly outnumber nonparental ditypes, even if the genes are over 80 cM. apart.  
**b** When two genes are linked, parental ditypes will usually outnumber nonparental ditypes, but distant genes (over 50 cM.) will appear to be unlinked.  
**c** When two genes are linked, the number of parental ditypes will be approximately equal to the number of nonparental ditypes, whatever the distance between the genes.
11. **a** The rate of recombination between two loci is a characteristic property of those loci. The expected rate is the same in all strains and in both sexes.  
**b** The rate of recombination between two loci is a varies somewhat between strains and can be particularly different between the two sexes.

12. (18 points) Label **each** of the following six statements as

**1)** true of DNA polymerase, **2)** true of RNA polymerase, **3)** true of both or **4)** true of neither.

- \_\_\_\_\_ **a.** Requires a primer whose 3' end is base-paired to the template.
- \_\_\_\_\_ **b.** Requires a template.
- \_\_\_\_\_ **c.** Uses dCTP as a substrate.
- \_\_\_\_\_ **d.** Can have (usually has) a 3'-5' exonuclease "proofreading" activity.
- \_\_\_\_\_ **e.** Adds nucleotides to the 5' end of a polynucleotide chain in accordance with basepairing to a template strand.
- \_\_\_\_\_ **f.** Adds nucleotides to the 3' end of a polynucleotide chain in accordance with basepairing to a template strand.

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13. (4 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? Answer 1 or 2 and explain.

your favorite gene (the query)      **ELVIS**  
alignment to hit 1                      **DIVLS**

your favorite gene (the query)      **ELVIS**  
alignment to hit 2                      **LIVES**

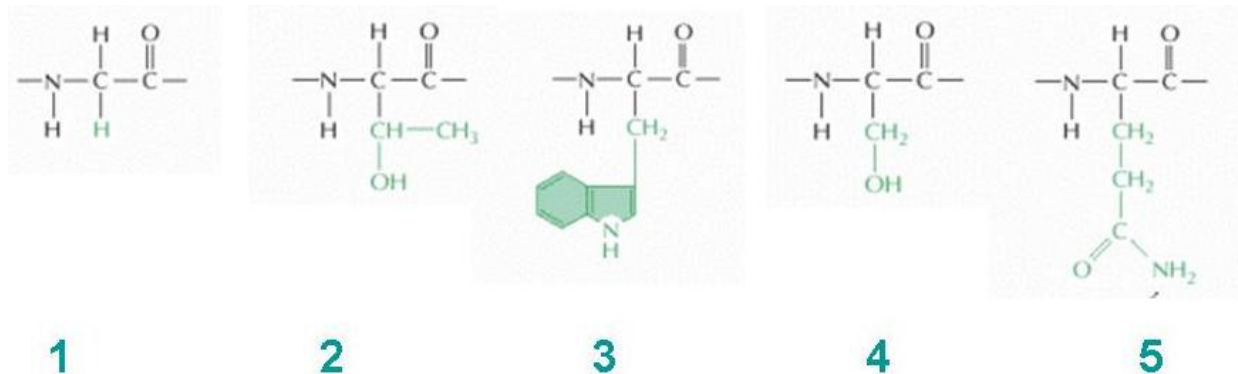
14. (4 points) Consider the following two possible alignments. This time, yfg (your favorite gene) differs from each of the other two sequences by only two substitutions in the region of alignment shown. To which sequence (3 or 4) is yfg more likely to be related? Answer 3 or 4 and explain.

your favorite gene                      **WHACK**  
alignment to hit 3                      **WHICH**

your favorite gene                      **WHACK**  
alignment to hit 4                      **SHARK**

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

N  
S  
G  
T  
W



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