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BIO SCI 97, GENETICS, FALL 2006 FINAL EXAMINATION

DO NOT OPEN EXAM UNTIL TOLD TO DO SO

BEFORE BEGINNING:

1. Write your **name** and **ID** number in the upper right corner of **this page** (and check the box if you're an extension student)
2. Write your **name** and **ID** number on your **Scantron** card, and bubble in the code box for your ID. Also, if you're an extension student, put an "X" in front of your written ID number, e.g X0698337
3. Write your exam version number (see top of this page) on the Scantron right after your name
4. Write your **name** and **ID** number on the answer sheet for Part II
5. Read the section below marked "very important" and sign the honesty pledge.
6. Bubble in your section and exam versions on the scantron as directed by the Professor

VERY IMPORTANT:

1. You must sit in your assigned seat unless given permission to do otherwise.
If you are not sitting in your assigned seat you may be given a zero on the exam.
2. This is a closed book test - no notes of any type are allowed.
3. Do not talk to another student. Do not look at another student's answers.
4. On desk - ID, Scantron, exam, answer sheet, pencil, calculator (calculator is optional). No notes, formulas, etc may be stored in calculator.
5. If you are seen with a cell phone or pager you may be given a zero on the exam.

I have read this page. I understand the rules and I pledge that I will not cheat on this exam or help others to do so.

_____ (sign your name here)

NOTES for all questions:

- Affected = has the disease
- Unaffected = does not have the disease (but may be a carrier if the disease is recessive)
- Normal = does not have the disease and is not a carrier.
- For all questions, assume any trait or disease mentioned has 100% penetrance, unless it is stated otherwise
- For inheritance questions, assume no new mutations, unless it is stated otherwise.
- All diseases mentioned are genetic diseases, unless it is stated otherwise.
- Pedigrees will only show you if someone is affected (black symbol) or unaffected (white symbol).
If someone is a heterozygous carrier of a recessive disease, they will be depicted as unaffected.

PART 1 – MULTIPLE CHOICE – ANSWER ON YOUR SCANTRON CARD

Choose the SINGLE best answer to each question. If none of the answers seem correct, or if more than one seems correct, choose the one that seems **most correct**. Mark your Scantron card with a dark PENCIL. Each question is worth one point unless otherwise noted.

1. A man smokes two packs of cigarettes per day from age 11, quits smoking at age 37, has his first child at age 39 and is diagnosed with malignant lung cancer at age 40. "I begged you not to smoke," says his wife, "and now you've probably given our baby your cancer". Her statement is:
 - A. reasonable, because cancer is a genetic disease
 - B. reasonable, due to the inheritance of acquired characteristics
 - C. reasonable, because the man probably had an inherited predisposition to cancer
 - D. not reasonable, because the carcinogenic mutations arose somatically and were not passed to the gametes
 - E. not reasonable, because the man stopped smoking before he got cancer, hence there is no connection between the smoking and the cancer

2. Which of the following statements is FALSE:
 - A. Cancer is a genetic disease of somatic cells
 - B. Somatic mutations are involved in many major diseases in the U.S., including cancer, heart disease, diabetes and schizophrenia
 - C. All the cells in a cancerous tumor have originated from a single progenitor cell
 - D. Mutations in several distinct genes are required for a normal cell to become cancerous
 - E. A predisposition to some types of cancer can also be inherited

3. Children and young adults get leukemia relatively frequently, whereas lung cancer is mainly a disease of older adults. The most likely explanation for this is:
 - A. Young adults smoke more than older adults
 - B. Increased risk of non-disjunction in older adults
 - C. Leukemia requires mutations in fewer distinct genes than does lung cancer
 - D. Children have fewer tumor suppressor genes
 - E. Higher mutation rate in children and young adults

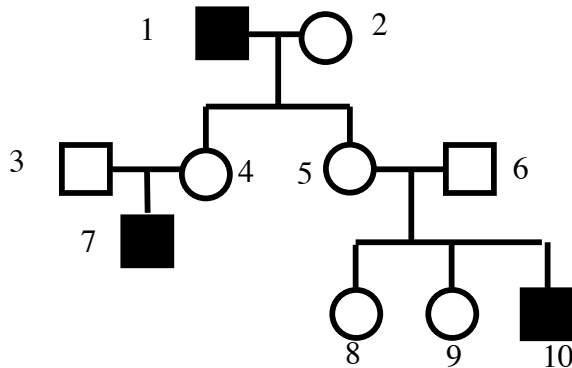
4. In the process of _____, bacterial DNA is transferred from one bacterial cell to another by a phage
 - A. transduction
 - B. transmutation
 - C. transformation
 - D. conjugation
 - E. transposition

5. Gene therapy is most likely to work against which class of genetic diseases?
 - A. dominant
 - B. recessive
 - C. multifactorial
 - D. infectious
 - E. somatic

6. The human *POLR2A* gene, encoding the largest subunit of RNA polymerase II, is an essential gene. There are no known human diseases associated with mutations in *POLR2A*. This is most likely because defective alleles of *POLR2A* are embryonic lethal when homozygous. Estimate the percentage chance that you are a carrier of a recessive allele of *POLR2A*.
 - A. virtually no chance at all
 - B. great than A, but less than 0.1%
 - C. greater than B, but less than 1%
 - D. greater than C, but less than 10%
 - E. between 10% and 100%

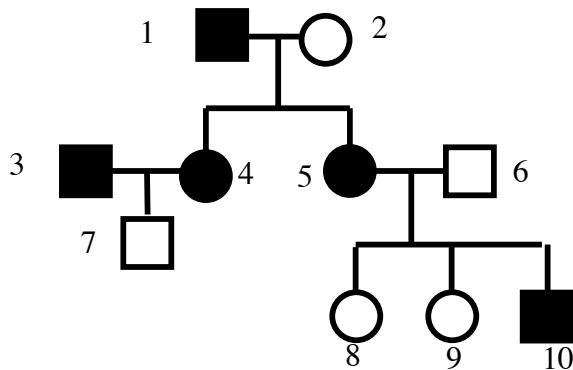
7. Which of the following statements is false?
- Current U.S. policy on cloning is that reproductive cloning should be banned but therapeutic (or research) cloning should not be.
 - There is no verified claim of the reproductive cloning of a human being.
 - There reproductive cloning of several different mammalian species has been achieved.
 - There is currently no federal law banning genetic discrimination.
 - More than one of the above statements (A-D) is false.

8-10. The pedigree shows the inheritance of a genetic disease in three generations of a family



8. Based on the pedigree, the mode of inheritance of this disease could be
- Autosomal recessive
 - X-linked recessive
 - Autosomal dominant
 - X-linked dominant
 - A or B
9. Assume that #7 was an affected female, instead of an affected male as shown. Given that assumption, then the mode of inheritance of this disease is most likely...
- Autosomal recessive
 - X-linked recessive
 - Autosomal dominant
 - X-linked dominant
 - A or B
10. Again, assume that #7 was an affected female, instead of an affected male as shown. What is the probability that #8 is heterozygous for the disease-causing allele?
- 100%
 - 67%, or 2/3
 - 50%, or 1/2
 - 25%, or 1/4
 - 0%
11. Which one of the following statements (A-D) are TRUE?. If you think more than one is true, pick E.
- Forced sterilization of American citizens judged to be genetically "inferior" has never been practiced in the U.S.
 - Most Americans routinely eat food containing genetically engineered crops.
 - Anyone who is a carrier of a deleterious recessive allele (one that would cause death or disease if homozygous) should not have children.
 - Germline gene therapy is routinely practiced in the U.S., although it is not always covered by medical insurance.
 - More than one of the above statements (A-D) is true.

12-14. The pedigree shows the inheritance of a genetic disease in three generations of a family



12. Based on the pedigree, the mode of inheritance of this disease could be
- Autosomal
 - X-linked
 - Y-linked
 - A or B
 - A or C
13. Based on the pedigree, the mode of inheritance of this disease could be
- Dominant
 - Recessive
 - A or B
 - None of the above are consistent with the pedigree
14. Assume that #10 marries an unaffected woman, and they have an unaffected daughter. Given that assumption, then the mode of inheritance of this disease is most likely...
- Autosomal recessive
 - X-linked recessive
 - Autosomal dominant
 - X-linked dominant
 - C or D
15. A single mutation occurs in the coding region of a gene and results in a non-functional gene product (protein). The type of mutation that most likely occurred (because it's the type most likely to result in a non-functional gene product) is:
- Missense or silent
 - Missense or nonsense
 - Missense or frameshift
 - Nonsense or frameshift
 - Silent or nonsense
16. The big problem with deaminated cytosine in DNA is that...
- It base-pairs with guanine
 - It base-pairs with adenine
 - There is no way for the cell to repair it
 - It will block replication and transcription
 - It depletes the cellular pool of uracil

17. In a certain species of slugs, angry, controlled by the Ang locus, is the dominant phenotype (genotype AA or Aa); the recessive phenotype is meek (genotype aa). Also in these slugs, beauty, controlled by the But gene, is the dominant phenotype (genotype BB or Bb); the recessive phenotype is ugly (genotype bb). Ang and But are linked. For an experiment you want to do, you need a lot of doubly-heterozygous beautiful angry slugs with the recessive alleles in trans. You will get these by crossing two slugs from a set of true breeding lab strains you have. What cross will you perform? (You may assume there is no recombination in these slugs).

- A. Cross a beautiful angry slug with an ugly meek slug
- B. Cross an ugly angry slug with a beautiful meek slug
- C. Cross an ugly angry slug with a beautiful angry slug
- D. Cross an ugly angry slug with an ugly meek slug
- E. Cross beautiful angry slug with a beautiful meek slug

18 & 19.

THIS PROBLEM IS WORTH 2 POINTS – FILL IN BOTH NUMBERS ON YOUR SCANTRON

You want to clone an insert containing a gene-of-interest into a plasmid vector. Which of the following is TRUE?

- A. Both the insert and plasmid must be cut with the same restriction enzyme.
- B. Complementary sticky ends from different DNA molecules cannot base pair.
- C. After ligation, the next step is transposition into a bacterial host.
- D. A replication origin is required in both the plasmid and the insert.
- E. More than one of the above statements (A-D) is true.

20 & 21.

THIS PROBLEM IS WORTH 2 POINTS – FILL IN BOTH NUMBERS ON YOUR SCANTRON

Autism is a multifactorial threshold trait with a broad-sense heritability near 1.0. Autistic individuals have an impaired ability to interact with other people. Sally has a half brother who is autistic, but she is not, and no-one else in her family is. Sally is concerned about her risk of having an autistic child. Which of the following statements is the most correct:

- A. Sally has a 1/3 chance of being a carrier for autism
- B. Since environmental factors play a major role in the development of autism, Sally should probably leave home
- C. Since autism is multifactorial, Sally should be concerned about the percentage of alleles she shares with her brother, which is 25%
- D. Sally's risk of being autistic would have been higher had she and her brother been twins
- E. More than one of the above statements is true.

22 & 23.

THIS PROBLEM IS WORTH 2 POINTS – FILL IN BOTH NUMBERS ON YOUR SCANTRON

The test cross $ab/ab \times Ab/aB$ is performed. The following number of progeny of each genotype are obtained: 67 AaBb, 436 Aabb, 417 aaBb, 80 aabb.

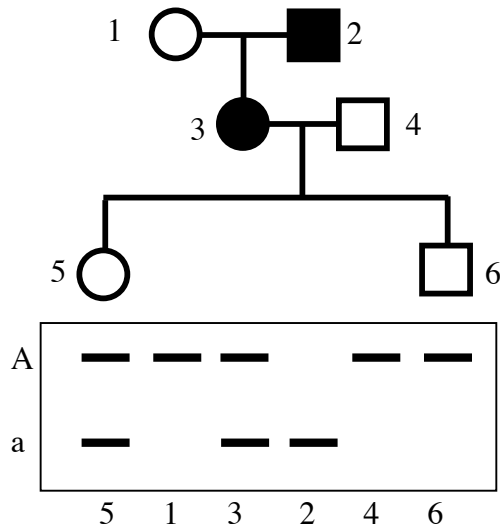
What is the approximate distance (in map units) between the two genes in question?

- A. 25 centiMorgans (cM)
- B. 20 cM
- C. 15 cM
- D. 10 cM
- E. 5 cM

24 & 25.

THIS PROBLEM IS WORTH 2 POINTS – FILL IN BOTH NUMBERS ON YOUR SCANTRON

The pedigree shows the transmission of an autosomal dominant disease with a late age of onset in three generations of a family. A marker that is tightly linked to the disease locus has restriction fragment length polymorphism alleles A and a, yielding the bands in the gel below the pedigree.



Is it likely that #5 will get the disease?

Is it likely that #6 will get the disease?

- | | |
|-------------|----------|
| A. #5 = YES | #6 = YES |
| B. #5 = YES | #6 = no |
| C. #5 = no | #6 = YES |
| D. #5 = no | #6 = no |

PART II - Short Answer Questions – Not Scantron – Write on Part II Answer Sheet

20 points total. Questions are worth 1 point each unless stated otherwise. Answers that are not clearly and carefully written will be marked wrong. Answers may be fractions or decimals. Decimal answers must be written to three decimal places. Do NOT, for example, round 0.667 up to 0.7.

26. (2 points) Which of the following are true about viruses (your may choose more than one answer)
- Common antibiotics are not effective against them.
 - They are dependent upon the host protein synthesis machinery.
 - They have their own proteins but do not have their own genome.
 - They typically have acquired multiple antibiotic resistance genes.
 - They are not cells, nor are they composed of cells.
 - None of the above (A-E) are true.

27-30. Match the following diseases to all true facts about their genetics by writing in the appropriate letter(s) from the table. For each disease, there is at least one true fact and no more than 4 true facts. You may use the items of genetic information more than once.

Example: Cystic Fibrosis **A R L** (autosomal recessive loss-of-function)

- 27. Huntington's Disease
- 28. Hemophilia
- 29. Sickle-Cell Anemia
- 30. Severe Combined Immunodeficiency (ADA gene)

GENETIC FACTS

- A. Autosomal
- X. X-linked

- D. Dominant
- R. Recessive

- G. Gain-of-function
- L. Loss of function

- I. An infectious disease, not a genetic disease
- H. Associated with heterozygote superiority
- T. In 1990, it became the first gene to be approved for use in gene therapy
- O. Late age of onset

31. a. Are dominant alleles usually gain-of-function (G) or loss-of-function (L)?
b. Are recessive alleles usually gain-of-function (G) or loss-of-function (L)?

32-37. A population consisting of 50 men and 50 women at Hardy Weinberg equilibrium take a 3-hour cruise on a tiny yacht that gets shipwrecked in a big storm, leaving them stranded on a desert island. Of the 100 people stranded on the desert island, 36 of them (18 men and 18 women) are affected with an autosomal recessive condition called "coconutitis" that is controlled by a single gene with two alleles, **C** (dominant, wild-type) and **c** (recessive, coconutitis-causing). Sadly, there's not much else to eat on the island but coconuts...

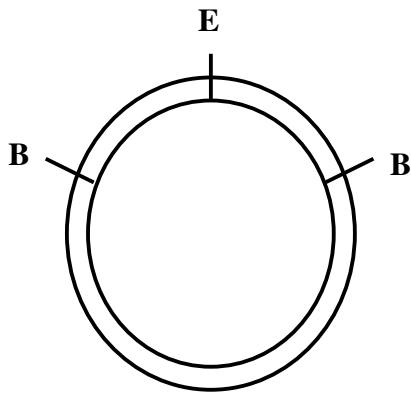
32. a. What is the frequency of the **C** allele?
b. What is the frequency of the **c** allele?
33. a. What is the frequency of heterozygous **Cc** individuals in this population?
b. How many people is this total?

34-36. Tragically, during a big coconut feast, all 36 of those affected with coconutitis die horrid deaths. Questions 34-36 refer to the 64 individuals remaining in the "post-feast" population (i.e., those people alive after the feast).

34. a. What is the frequency of the **C** allele?
b. What is the frequency of the **c** allele?
35. What is the frequency of heterozygous **Cc** individuals in this population?

36. Is the the “post-feast” population in Hardy Weinberg equilibrium (yes or no).
37. With no rescue in sight, the post-feast population undergoes one round of random mating. Each couple has two kids, producing a new generation of 64 kids.
- What is the frequency of kids affected with coconutitis?
 - How many kids total in the new generation are affected with coconutitis? (HINT: if you did the problem correctly, this should be a whole number between 0 and 64)

38-40.



The figure to the left shows a 10 kb plasmid molecule containing a single cleavage site for the EcoRI (E) restriction endonuclease and two BamHI (B) cleavage sites, located 2 kb to either side of the EcoRI site.

38. If this plasmid is cut with BamHI, what size band(s) will be observed on a gel?

39. If this plasmid is cut with EcoRI, what size band(s) will be observed on a gel?

40. The plasmid is cut with EcoRI and a 3 kb fragment is inserted into the EcoRI site. The insert does not contain any BamHI sites. If the new recombinant DNA molecule is cut with BamHI, what size band(s) will be observed on a gel.

41. A multifactorial trait is one for which the variation in the trait is typically... (choose none, one or more than one):
- determined by the combined effect of multiple genes
 - determined by the variance in the environment
 - determined by genetic drift
 - mitochondrially inherited
 - determined by somatic mutations

42-44 are true-false questions.

42. Recombination mistakes usually result in point mutations
43. In the U.S., it is illegal to sell cloned or genetically modified pets.
44. The rarer an autosomal recessive disease, the higher proportion of recessive alleles that will be found in carriers.

HAPPY HOLIDAYS!