

BMB 526 - Exam II November 29, 2006

You may use this grid to record your answers for comparison with the posted answer keys.

Version of the Exam: 1A

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| 1. _____ | 8. _____ | 15. _____ | 22. _____ | 29. _____ |
| 2. _____ | 9. _____ | 16. _____ | 23. _____ | 30. _____ |
| 3. _____ | 10. _____ | 17. _____ | 24. _____ | 31. _____ |
| 4. _____ | 11. _____ | 18. _____ | 25. _____ | 32. _____ |
| 5. _____ | 12. _____ | 19. _____ | 26. _____ | 33. _____ |
| 6. _____ | 13. _____ | 20. _____ | 27. _____ | 34. _____ |
| 7. _____ | 14. _____ | 21. _____ | 28. _____ | |

BMB 526 Exam II - November 29, 2006

NAME (print clearly) _____ **CHM** **COM** (circle one)

BEFORE you begin the exam, please complete the following information on your response sheet:

- (a) your name and signature**
 - (b) your student number (PID)**
 - (c) your college --- in the area under SECTION: mark 001 for CHM student
mark 002 for COM student**
 - (d) your version of the exam is 1A --- mark this in the area under FORM**
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This exam contains 34 questions. For each question, please mark the letter on the response sheet that corresponds to the best available answer.

When you leave the exam room, please turn in your RESPONSE SHEET and your EXAM to the proctors. Once you exit the exam room, please DO NOT re-enter until the end of the exam period. To reduce the noise entering the exam room, please do not remain in the hall outside the exam room.

Answer Keys will be posted at the Angel website following the exam.

You have 65 minutes to complete this exam.

Do well and good luck.

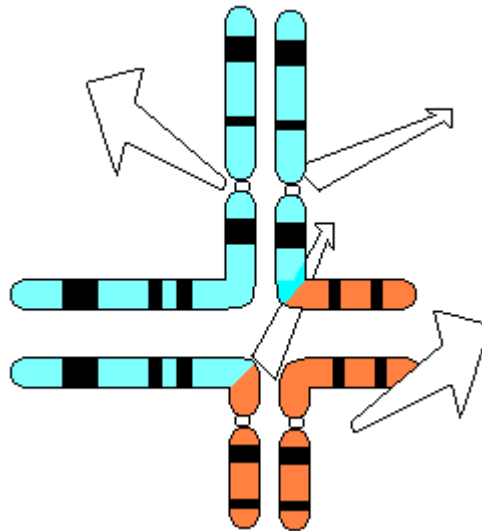
- 1) In regulating gene expression, different sigma factors are used:
 - A. to initiate transcription of different sets of genes.
 - B. to turn on or to turn off expression of the *lac* operon, depending on the availability of glucose or lactose.
 - C. for each of the three eukaryotic RNA polymerases.
 - D. to bind to different subunits of RNA polymerase.
 - E. to select different 5'-splice sites for alternative splicing events.

- 2) Which of the following events would occur at the *E. coli lac* operon when the glucose concentration of the growth medium is low and the lactose concentration is high?
 - A. The *lac* repressor will bind to the operator sequence.
 - B. Ribosomes will bind to the promoter sequence.
 - C. The transcribed RNA will feedback inhibit the transcription of the structural genes of *lac* operon by RNA polymerase.
 - D. The CAP protein, bound with cAMP, will recruit RNA polymerase to the promoter.
 - E. An alternative sigma factor will recognize sequences upstream of each open frame (ORF) in the operon.

- 3) If a mutation occurred in the gene encoding the glucocorticoid receptor, such that the receptor protein no longer could bind zinc atoms, which activity of the receptor would be most directly affected?
 - A. ligand (hormone) binding
 - B. DNA binding
 - C. cytoplasmic anchorage
 - D. nuclear localization
 - E. targeting of the receptor to the plasma membrane

- 4) The gene encoding the β subunit of hemoglobin (β -globin) resides on human chromosome #11. A patient of yours has a mutation in one allele of his two β -globin genes; the defect involves the deletion of an enhancer controlling expression of the affected β -globin gene, leading to lower levels of β -globin protein expression. Choose the **CORRECT** statement.
 - A. The reduced levels of β -globin protein in the patient's red cells reflect reduced half-life of the protein.
 - B. The β -globin mRNAs in the patient's reticulocytes will not be translated effectively by ribosomes.
 - C. Abnormal (truncated) and full-length versions of the β -globin protein will coexist in the patient's red cells.
 - D. The β -globin mRNAs isolated from the patient will be normal in length and intron/exon structure.
 - E. The reduced levels of β -globin protein in the patient's red cells reflect reduced half-life of β -globin mRNA.

- 5) Suppose a patient carries a mutation in the DNA that codes for the 3'-untranslated region of the mRNA for transferrin receptor (T_fR), such that the Iron Responsive Element (IRE) no longer binds Iron Responsive Factor (IRF). Under conditions of low iron, what will be the **PRIMARY** problem arising from this mutation?
- Decreased transcription of the mRNA for T_fR .
 - Altered processing of the hnRNA for T_fR .
 - Increased susceptibility of the T_fR mRNA to nuclease degradation.
 - Altered subcellular distribution of the T_fR polypeptide.
 - Altered subunit association of the T_fR polypeptide.
- 6) Which of the following molecules can covalently modify and unpack nucleosomes?
- sigma factor
 - iron responsive factor
 - tryptophan operon
 - cyclic AMP (cAMP)
 - histone acetyl transferase
- 7) Which chromosomal aberration produces the following synaptic configuration during Meiosis I?



- Inversion
- Duplication
- Translocation
- Deletion
- None of the above

- 8) The percentage that most closely approximates the percent of infants born with a congenital anomaly that will require medical attention at some point within the first 5 years of life is:
- A. .01%
 - B. .1%
 - C. 3-6%
 - D. 10-15%
 - E. None of the above
- 9) What fraction most closely approximates the frequency of spontaneous abortions that have chromosomal aberrations?
- A. Less than .1%
 - B. 66%
 - C. 5%
 - D. 20%
 - E. None of the above
- 10) Which statement about most cases of Down's syndrome is FALSE?
- A. The frequency increases with advancing maternal age.
 - B. Meiotic Non-disjunction of chromosome #21 can be a cause.
 - C. Most affected individuals have Trisomy for Chromosome 21.
 - D. One does not need to get a karyotype if your patient has the classic phenotype of Down Syndrome.
 - E. None, all statements are true.
- 11) Which statement concerning Klinefelter's syndrome is FALSE:
- A. Behavioral abnormalities can be found in affected individuals.
 - B. Two Barr bodies would be evident on Karyotype.
 - C. Infertility can be a sole reason for clinical presentation.
 - D. Tall stature and small testicles in a pre-pubertal teen male could be a sign of Klinefelters Syndrome.
 - E. None, all statements are true.
- 12) Which of the following is not an indication for chromosome analysis:
- A. Previous child with a heritable chromosome abnormality.
 - B. Both parents have bipolar affective disorder (manic depression).
 - C. Child has microcephaly with developmental delay.
 - D. Child with multiple congenital anomalies.
 - E. All of the above.

13) Leading known genetic cause of mental retardation in humans:

- A. Fragile X syndrome
- B. XXY
- C. Down Syndrome
- D. Prader-Willi Syndrome
- E. Turner Syndrome

14) Which of the following is **TRUE** about chromosome alterations?

- A. Carriers of a Paracentric Inversion primarily face the prospect of giving birth to chromosomally unbalanced offspring.
- B. Inversions are primarily responsible for non-disjunction events.
- C. A chromosomally unbalanced offspring derived from a parent carrying a balanced translocation is caused by recombination events between chromosomes during meiosis.
- D. Abnormal children having a deletion of a chromosome segment may have sibs that have partial trisomy or monosomy for chromosome segments as well.
- E. None of the above is true.

15) Which of the following items is TRUE when distinguishing Edwards Syndrome (Trisomy 18) from Patau Syndrome (Trisomy 13):

- A. Trisomy 18 is more frequent than Trisomy 13.
- B. Mental retardation is only a feature of Trisomy 18.
- C. High lethality within first year of life occurs only in Trisomy 13.
- D. Congenital cardiac malformations are only present in Trisomy 18.
- E. Increased risk with advanced paternal age occurs only with Trisomy 13.

16) Some individuals affected by Prader-Willi syndrome can show each of the following findings or symptoms (the latter at some point during their development) **EXCEPT**:

- A. Hypotonia (decreased muscle tone).
- B. Hyperphagia (Excessive eating).
- C. Deletion of chromosome bands encompassing chromosome 15q11.
- D. Uniparental disomy for paternal information at chromosome 15q11.
- E. Gross deletions of the same chromosomal region as noted in some individuals affected by Angelman syndrome.

17) Regarding individuals affected by Fragile X Syndrome, which of the following is FALSE?

- A. Such an individual carries an expanded CGG trinucleotide repeat in the Fragile X gene as the most likely cause for his/her phenotype.
- B. Females have a 50% chance of passing on the trait to their offspring.
- C. Unaffected female premutation carriers could pass on an expanded allele to their offspring, resulting in serious cognitive impairment in that offspring.
- D. Males carrying premutation size Fragile X alleles do not suffer from any medical problems.
- E. Females having the same size expansion as an affected male may have less cognitive involvement as a result of random X-inactivation.

18) Which of the following is FALSE regarding the trinucleotide repeat expansions that cause the neurodegenerative disorder, Huntington Disease?

- A. They are considered to be gain-of function mutations.
- B. Expansion of the trinucleotide repeat is more likely to occur when the mutation is transmitted through the mother, than through the father.
- C. Trinucleotide expansion in the huntingtin gene results in an expanded polyglutamine tracts in the huntingtin protein.
- D. They contrast with some other trinucleotide disorders, in that trinucleotide expansions in the latter can occur in non-protein coding regions of genes.
- E. A larger size of an expansion generally correlates with an earlier age for onset of Huntington symptoms.

19) Of the following, which is false regarding the etiology of many congenital anomalies/birth defects?

- A. some are due to a detectable chromosome abnormality.
- B. some are caused by being part of a previously defined association.
- C. some are caused by being part of a multiple congenital anomaly syndrome.
- D. some are detected in isolation, and not associated with other anomalies.
- E. most are present as an isolated finding, and thus are very unlikely to recur in a family.

20) Which of the following is **least** likely to increase the recurrence risk of trisomy 13 in a family?

- A. Advanced paternal age.
- B. Father carries a 13/18 Robertsonian translocation.
- C. Mother carries a 13/14 Robertsonian translocation.
- D. Germline mosaicism in the mother, in which some cells are 47,XX+13.
- E. None of the above.

21) Of the following chromosome abnormalities involving a portion of the terminal p- arm of a given chromosome, which abnormality is **most** likely to cause the most severe disease in a carrier of the abnormality?

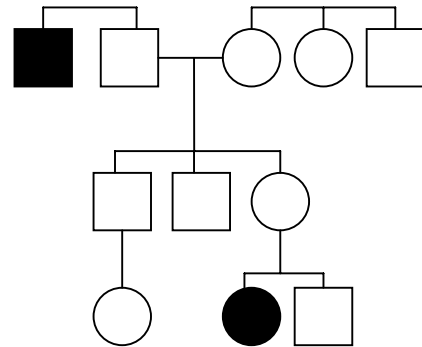
- A. Duplication of the region.
- B. Deletion of the region.
- C. Pericentric inversion of the region.
- D. Paracentric inversion of the region.
- E. A balanced translocation involving the region.

22) If a male child with developmental delay is your patient, an initial investigation into the potential genetic causes for this symptom should include:

- A. SMAC-20
- B. Electrolyte panel
- C. Chromosome analysis
- D. Fragile X DNA analysis
- E. C and D

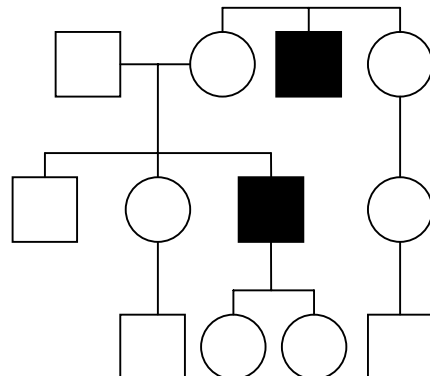
23) Please indicate the most likely mode of inheritance for the following pedigree:

- A. Autosomal dominant
- B. Autosomal recessive
- C. X-linked dominant
- D. X-linked recessive
- E. Mitochondrial



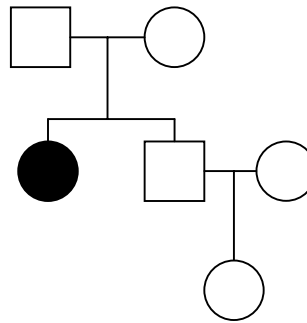
24) The following pedigree represents a family with an x-linked recessive condition. Please indicate the obligate carriers:

- A. I-2, III-2 and III-3
- B. I-2 and II-2
- C. I-2, II-2, III-2 and III-3
- D. II-2, III-2 and III-3
- E. I-2, I-4, II-2 and II-4



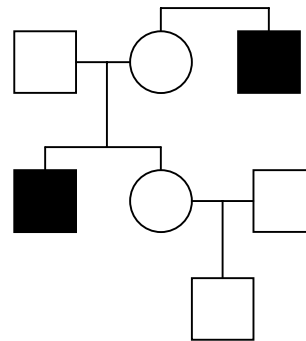
- 25) Given the following pedigree for an autosomal recessive condition, what is the chance that individual III-1 is a carrier? We can assume that individual II-3 is not a carrier of this condition.

- A. 0
 B. 1 / 4
 C. 1 / 3
 D. 1 / 2
 E. 2 / 3



- 26) The following pedigree represents a family with Duchenne muscular dystrophy. Individual II-2 just had a baby boy. What is the chance that he will have Duchenne muscular dystrophy?

- A. 0
 B. 1 / 4
 C. 1 / 3
 D. 1 / 2
 E. 2 / 3

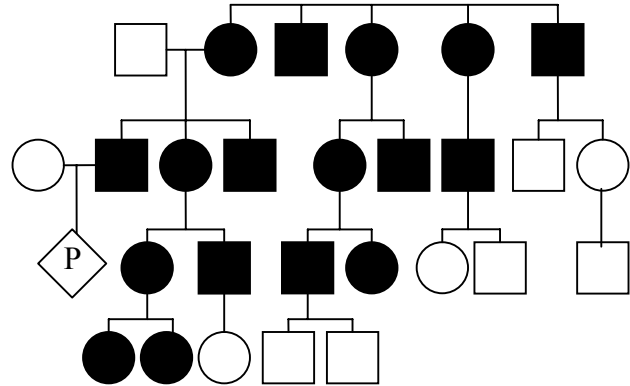


- 27) You have a 6-year-old female patient who you are seeing for the first time. She was adopted and her family history is unknown. She has microcephaly, seizures, and severe developmental delay. Throughout the appointment, you notice that she is making unusual repetitive movements with her hands, that you would describe as 'hand wringing' and 'hand washing'. Upon reviewing her developmental history, her mother reports that she seemed to be developing normally until around her first birthday, at which time she starting losing developmental skills. Her head circumference had also been on the normal growth curve as an infant. What is your suspected diagnosis?

- A. Marfan syndomre
 B. MELAS
 C. Neurofibromatosis
 D. Rett syndrome
 E. Tuberous sclerosis

28) Individual II-1 and II-2 are pregnant with their first child. Based on the most likely mode of inheritance, what is the chance that this child will be affected by the condition present in his father?

- A. 0
- B. 1 / 4
- C. 1 / 3
- D. 3 / 4
- E. 1



29) A 37-year-old male who works in a manufacturing plant has recently developed hearing loss. You find out he also has a family history of hearing loss. Other individuals in his family have undergone genetic testing and have a known mutation in a hearing loss-related gene. You test your patient and he does not have the mutation that is in his family. How could this be explained?

- A. Incomplete dominance
- B. Reduced penetrance
- C. Variable expressivity
- D. Phenocopy
- E. Co-dominance

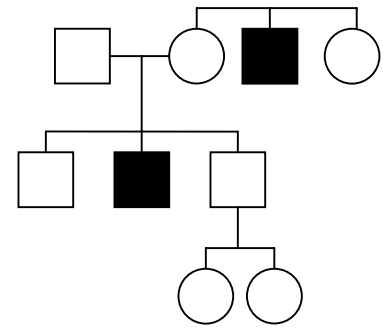
30) An autosomal dominant condition has a penetrance of 80%. What is the chance that the child of an affected individual will also express the condition?

- A. 40%
- B. 45%
- C. 50%
- D. 60%
- E. 100%

31) You have a 12-year-old patient with tuberous sclerosis. He has multiple hypopigmented macules, facial angiofibromas, a shagreen patch, seizures and mental retardation. During your evaluation, you examine both of his parents and find that his father has a few hypopigmented macules and a periungual fibroma. Therefore, you also diagnose him with tuberous sclerosis. What factor would explain the presentation in this family?

- A. Reduced penetrance
- B. Locus heterogeneity
- C. Variable expressivity
- D. De novo mutation
- E. Epistasis

- 32) You have thoroughly examined each individual in this family, and individual I-2 has no clinical features of this condition. Which of the following choices is a *highly unlikely* explanation for I-2 being unaffected.



- A. This is a sex-limited condition that is only expressed in males.
- B. This is an autosomal dominant condition with reduced penetrance.
- C. This is an X-linked recessive condition and the mother is an unaffected carrier.
- D. This is an autosomal dominant condition, and individual I-2 has germline mosaicism for the mutation.
- E. This is an autosomal recessive condition and I-2 and her partner I-1 are both carriers.
- 33) Which of the following statements is FALSE regarding X-linked inheritance and X-inactivation:
- A. A female who is a carrier of an X-linked recessive disease may show some features of the condition.
- B. A female who is a carrier of an X-linked dominant condition may have a very mild condition or show no features at all.
- C. A female who is homozygous recessive for an X-linked recessive condition will always be affected.
- D. A male who is hemizygous recessive for an X-linked recessive condition will always be affected.
- E. A male who is hemizygous recessive for an X-linked recessive condition may not be affected.
- 34) Which of the following statements is TRUE regarding mosaicism:
- A. Somatic mosaicism occurs only in the germ cells of an individual.
- B. Individuals with somatic mosaicism have a more severe expression of a condition than an individual with a mutation in all of their cells.
- C. Somatic mosaicism is not inherited from a parent.
- D. Individuals with somatic mosaicism have no increased risk of passing on the mutation to their children.
- E. Individuals with somatic mosaicism will never express the condition themselves, but may pass it on to their children.