Molecular Cell Biology and Genetics
Sample Questions for Exam 1 and part of Exam2 Material:

1. Which of the following amino acids has a side chain R group that is most likely to be positively charged at physiological pH values?
   A. Proline
   B. Glutamate
   C. Tyrosine
   D. Arginine
   E. Cysteine

2. Which of the following is a derived amino acid?
   A. Hydroxyproline
   B. Isoleucine
   C. Glutamine
   D. Asparagine
   E. Cysteine

3. The lowering of the pH of the blood within rapidly metabolizing tissues has what effect on Hb?
   A. Increases the Hill coefficient for oxygen association-dissociation
   B. Increases the P_{50} value for Hb binding to oxygen
   C. Causes Hb to dissociate into α and β dimers
   D. Increases the conversion of Hb from the T conformation to the R conformation
   E. Causes the pKa of His-146 to decrease

4. Which statement best explains the positive cooperativity observed in the binding of O_2 to hemoglobin?
   A. The binding of O_2 induces the dissociation of tetrameric hemoglobin into two αβ-dimers that have a higher affinity for O_2.
   B. The binding of O_2 to one subunit induces a conformational change that is transmitted to neighboring subunits through their contacts.
   C. The binding of O_2 to one subunit helps recruit DPG to the DPG binding site.
   D. The binding of O_2 to one subunit causes a localized increase in pH, which increases the affinity of the remaining subunits for O_2.
   E. The binding of O_2 to one subunit induces an R to T conformational transition in that subunit.

5. Which statement describes a feature of protein secondary structure?
   A. Amino acids found in regions of regular secondary structure always have phi (φ) bond angles that are equal to their psi (ψ) bond angles.
   B. Every third amino acid residue in an α-helix is either proline or hydroxyproline.
   C. A peptide bond in the α-helix is hydrogen bonded to a peptide bond above and below it in the helix.
   D. β-structure is unlikely to be observed in globular proteins because β-strands mostly form flat, sheet-like structures.
6. Glucokinase and hexokinase both catalyze the conversion of glucose to glucose 6-phosphate. Glucokinase has a $K_m$ for glucose of $3 \times 10^{-3} \text{ M}$ and a $V_{\text{max}}$ of $10 \text{ mM/sec}$. Hexokinase has a $K_m$ of $3 \times 10^{-6} \text{ M}$ and a $V_{\text{max}}$ of $5 \text{ mM/sec}$. Which statement best describes the rate with which glucose is converted to glucose 6-phosphate by each of the enzymes?

A. At a glucose concentration of $3 \times 10^{-3} \text{ M}$, both enzymes convert glucose to glucose 6-phosphate at the same rate.

B. Doubling the glucose concentration from $3 \times 10^{-3} \text{ M}$ to $6 \times 10^{-3} \text{ M}$ nearly doubles the rate by which both enzymes convert glucose to glucose 6-phosphate.

C. At a glucose concentration of $3 \times 10^{-3} \text{ M}$, adding a competitive inhibitor of hexokinase at a concentration equal to its $K_i$ reduces its rate by a factor of 2.

D. At a glucose concentration of $3 \times 10^{-3} \text{ M}$, adding a noncompetitive inhibitor to both enzymes reduces the rate of glucokinase reaction but not that of hexokinase.

Questions 7 through 9 refer to the figure below, which shows the reaction of an enzyme in the absence and presence of various inhibitors.

7. Which line best describes the activity of an enzyme in the absence of an inhibitor?

8. Which line best describes the activity of the same enzyme in the presence of a competitive inhibitor?

9. Which line best describes the activity of the same enzyme in the presence of a noncompetitive inhibitor?

10. Pepsin, a proteolytic enzyme found in the stomach, contains an active site aspartate residue. The β-carboxylic acid side chain group must be in its unprotonated (basic) form for the enzyme to be catalytically active. The $pK_a$ of the β-carboxylic acid group is 3.5. At pH 4.5, what is the approximate activity of the enzyme?

A. 1% of its maximum potential activity

B. 10% of its maximum potential activity

C. 50% of its maximum potential activity

D. 90% of its maximum potential activity

E. 99% of its maximum potential activity
11. Some members of a family are carriers of the following mutation in the β-globin gene. Which of the following techniques would most likely detect the presence of a carrier of the defective gene?

![Diagram of β-globin gene with IVS-1 and IVS-2 introns]

A. Fluorescence in situ hybridization (FISH)
B. Quantitative reverse transcriptase linked PCR (qRT-PCR) to measure the level of β-globin mRNA
C. Southern blotting using β-globin cDNA as a probe
D. Sequencing β-globin cDNAs from each family member
E. Western blotting using an antibody to the TATA binding protein

12. The following alleles are found for a given locus in two individuals identified as subject A and subject B

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<tr>
<th>Subject A</th>
<th>AGCTTGGTCC (GTA)$_8$ ACTGATAGCG</th>
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<td>AGCTTGGTCC (GTA)$_{18}$ ACTGATAGCG</td>
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<th>Subject B</th>
<th>AGCTTGGTCC (GTA)$_{13}$ ACTGATAGCG</th>
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<td>AGCTTGGTCC (GTA)$_{26}$ ACTGATAGCG</td>
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Which combination of techniques would you most likely use to distinguish between DNA samples from subject A and Subject B based on an analysis of the given locus?

A. Reverse transcription of an mRNA sample followed by Southern Blotting
B. PCR amplification of a cDNA sample followed by electrophoresis
C. mRNA isolation followed by Northern blotting
D. PCR amplification of genomic DNA followed by electrophoresis
E. Electrophoresis of a protein sample followed by Western blotting

13. Which general transcription factor contains both a helicase subunit and a protein kinase subunit?

A. TFIIA
B. TFIIB
C. TFIID
D. TFIIF
E. TFIIH
14. Which statement BEST describes the properties of a promoter?
A. A promoter consists of a symmetrical arrangement of short DNA sequences that recruit RNA polymerase to transcribe DNA in either of two directions
B. A promoter is able to directly bind an RNA polymerase unaided and with high affinity to transcribe a given gene
C. TATA binding protein is present only in transcription initiation complexes that assemble on promoters that contain a TATA box sequence.
D. The nature of the promoter determines which of the three RNA polymerases will be recruited to transcribe a particular gene

15. Recruiting which protein(s) to a region of DNA is most likely to convert the region into heterochromatin?
A. HAT enzymes
B. HpaII enzymes
C. Swi/Snf protein
D. LCR binding proteins
E. HDAC enzymes

16. Which statement best describes a characteristic of DNA methylation?
A. DNA methylation usually activates gene expression
B. A HDAC protein removes methylated sites in newly synthesized DNA
C. A possible site for DNA methylation is the cytosine in the sequence ACTAT
D. CG islands are extensively methylated in normal cells
E. A possible site for DNA methylation is the cytosine in the sequence ATCGT

17. Which statement best describes a feature of alternative splicing?
A. The mRNA for every gene expressed in a eukaryotic cell is subject to alternative splicing
B. Alternative splicing is facilitated by antisense RNAs that bind and inactivate specific intron-exon junctions
C. Alternative splicing controls sex determination in humans
D. Alternative splicing may help in the establishment of neuronal circuits in the brain of Drosophila (fruit flies)

18. Which of the following is most likely to increase the rate of translation of an mRNA?
A. The loss of the 5’ N7-methylguanine cap
B. The loss of the 3’ polyA(+) tail
C. The phosphorylation of the translation initiation factor, eIF2
D. The phosphorylation of eIF4E-BP, an eIF4E-binding protein
E. The presence of a 5’ untranslated region that contains multiple start and stop codons
19. Which chromosomal abnormality is most likely to require FISH for its diagnosis?
   A. A Robertsonian Translocation  
   B. A Monosomy  
   C. A Trisomy  
   D. A pericentric inversion  
   E. A microdeletion

20. A female is an unbalanced carrier of a duplication of part of the q arm (long arm) of one X chromosome. What X inactivation pattern would you expect in this patient?
   A. Random X inactivation  
   B. Nonrandom X inactivation where the normal X is preferentially inactivated  
   C. Nonrandom X inactivation where the abnormal X is preferentially inactivated  
   D. Inactivation of just the duplicated region on the abnormal X-chromosome  
   E. X-inactivation pattern depends upon the tissue you are analyzing.
21. A retarded young man with small hands, feet and genitals is grossly overweight. His parents indicate that he is continually hungry, always eating, and hoarding and stealing food. He and his parents underwent genetic testing. In the linked image above, we see the results of a PCR analysis of two microsatellite markers (highly polymorphic short tandem repeat) on chromosome 1 and two on chromosome 15 from all three. What disorder is the son likely to have and what is its likely cause?
A. Prader-Willi syndrome/microdeletion
B. Prader-Willi syndrome/uniparental disomy
C. Angelman syndrome/microdeletion
D. Angelman syndrome/uniparental disomy
E. The results show no evidence of a genetic disorder

22. Tay-Sachs is an autosomal recessive disease that causes cherry-red spots in the eye, “startle” responses in infancy, neurodegeneration, and death. What is the risk that the grandmother of an affected child is a carrier of the abnormal allele?
A. 100%
B. 67%
C. 50%
D. 25%
E. Virtually 0
23. With regard to the following pedigree, which of the following is the most likely mode of inheritance?

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

24. Autosomal recessive conditions are best characterized by which of the following statements?
A. They are often associated with deficiency in an enzyme activity
B. The two abnormal alleles always contain the same inactivating mutation
C. The resulting phenotypes are more variable than observed for autosomal dominant conditions
D. Most persons do not carry any abnormal recessive genes
E. Affected individuals are likely to have affected offspring
Huntington disease (HD) is a neurodegenerative disorder characterized by a degeneration of the striatum and cortex of the brain. Patients show motor abnormalities, personality changes, a gradual loss of higher brain functions, and eventually die. Below is a family pedigree and an accompanying Southern blot that was obtained with genomic DNA that was probed with a DNA complementary to the gene for the Huntingtin protein. Based on the linked image above, which statement is most likely to be CORRECT?

A. HD is an autosomal recessive disease
B. The huntingtin gene is likely to be located on the X-chromosome
C. A likely cause of HD is an unstable triplet repeat in the HD gene
D. The pattern of bands in the blot is indicative of abnormal splicing of the HD mRNA
E. In most individuals, the region of the HD gene detected by the DNA probe is distributed on six different restriction fragments
**Answers**

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* For the same enzyme, line A would result from the presence of a mixed inhibitor (one that decreases Vmax and increases Km).