Pathobiochemistry 2011/2012

Exam Questions

Section I: "Metabolites and enzymes"

1. Basic characterization of IEM.
2. Pathogenetic mechanisms of IEM.
3. IEMs of small molecules.
4. IEMs of complex molecules.
5. Classification of lysosomal storage disorders and pathogenic mechanisms.
6. Mucopolysaccharidoses and glycoproteinoses.
7. Lipidoses and deficiencies of hydrolases activators.
8. Peroxisomal disorders.
9. Mitochondrial disorders caused by deficiencies of enzymes in respiratory chain and citric acid cycle.
10. Mitochondrial disorders caused by mutations in mitochondrial DNA.
11. Disorders of mitochondrial beta oxidation of fatty acids.
12. Starvation and disorders of ketone bodies production.
13. Liver glycogenoses.
14. Muslce glycogenoses and M. Pompe.
15. Hereditary disorders of galactose and fructose metabolism.
16. Hereditary disorders of protein glycosylation (CDG syndromes).
17. Disorders or aromatic and branched-chain amino acids.
18. Urea cycle disorders.
19. Dietary and genetic disorders of folate, cobalamine, and sulfur amino acid metabolism.
20. Disorders of amino acid metabolism and of creatine synthesis.
21. Disorders of uric acid metabolism.
22. Disorders of purine and pyrimidine metabolism
23. Hepatic porphyrias.
24. Cutaneous porphyrias.
25. Methods for diagnosis of IEM.
26. Neonatal and selective screening for IEM.
27. Treatment of IEMs affecting small molecules – principles and examples.
28. Treatment of IEMs affecting complex molecules – principles and examples.

Section II: "Metabolism of information"

29. Mechanism of tumor disease formation – overview.
30. Physical factors participating in tumor induction.
31. Chemical carcinogenesis.
32. Viral carcinogenesis.
33. Mechanisms of tumor transformation.
34. Disorders of cell signaling pathways resulting in uncontrolled proliferation of tumor cells.
35. Disorders of apoptotic signaling pathways in tumor cells.
36. Disorders of DNA repair mechanisms in tumor cells.
37. Molecular mechanisms of neovascularization and ways of their medical modification.

38. Angiogenesis and neovascularization (differences, medical modification).
39. Molecular mechanisms of metastases formation, ways of medical modification.
40. Selection of resistant tumor clones, medical modification.
41. Tumor microenvironment: relationships among transformed cells and tumor stroma.
42. Tumor stroma as a target for therapy.
43. Pathology of signaling cascades regulating cellular proliferation: concept and examples.
44. Targeted therapy: examples of therapeutical intervention at the molecular level in oncology.
45. Hereditary cancer syndromes and sporadic tumor diseases.
46. Techniques for analysis of mutations in inherited predispositions to cancer.
47. Analysis of somatic mutations and microsatellite markers in sporadic tumors.
48. Possibilities of detection of minimal residual disease.
49. Purpose and types of anti-cancer treatment.
50. Types of chemotherapeutics, their undesirable effects.
51. Biochemical principles of chemotherapy and radiotherapy.
52. Biochemical principles of hormonal and targeted therapy.
53. Description and role of tumor markers in anti-cancer treatment.
54. Sensitivity and specificity of tumor markers, examples.
55. Cancer- and tissue-specific tumor markers, examples.
56. Tumor markers – application and interpretation: screening, monitoring, diagnosis.

Section III: "The inner environment and limits of its maintenance"

57. Metabolic acidosis, its causes and consequences.
58. Metabolic alkalosis, its causes and consequences.
59. Combined disorders of acid-base equilibrium.
60. Relations between acid-base equilibrium and concentration of ions. Changes in ionogram in disorders of acid-base equilibrium. Changes in acid-base equilibrium in disorders of ion metabolism.
61. Principal reactive oxygen and nitrogen species: properties, reactions, main sources in the body, role in pathogenesis.
62. Physiological role of reactive oxygen species in metabolism: tissue hormones, phagocyte weapons, hydroxylases, redox signaling.
63. Lipid peroxidation as an example of oxidative damage to biomolecules. Significance of transition metals (iron, copper) in pathobiochemistry of reactive oxygen species.
64. Antioxidant defense of human body.
65. Biochemical basis of ageing. Radical/mitochondrial theory, ageing as catabolic failure, relationship to chronic inflammation.
66. Role of mitochondria in cell death (apoptosis and necrosis) and physiological ageing.
67. What a cell needs to become immortal? Autophagy, Hayflick limit, telomerase.
68. Difference between average life expectancy and maximum lifespan. Role of genes, theory of antagonistic pleiotropy, present possibilities how ageing can be affected by lifestyle: caloric restriction, physical activity, diet composition.
69. Metabolic syndrome and insulin resistance – characteristic, cause and implication, possible therapeutic approach.
70. Formation of AGEs, interaction AGE - RAGE, potential mechanisms to reduce formation/effect of AGEs.
71. Mechanisms of hyperglycemia-induced tissue damage.

72. Carbonyl stress, its role in pathogenesis of long-term diabetic complications, atherosclerosis and renal failure.
73. Role of LDL in atherosclerosis.
74. Role of HDL in atherosclerosis.
75. Explain the biochemical processes during initial stages of atherosclerosis.
76. Role of monocytes/macrophages, endothelium, smooth muscle cells and T lymphocytes in atherosclerosis.
77. Rules of protein folding.
78. Role of chaperones, proteasomes and lysosomes in the cell.
79. Mechanism of prion diseases.
80. Origins of pathological conformation of proteins and examples of clinical consequences.
81. Endoplasmic reticulum stress.
82. Metabolic alterations in cell during anoxia, ischemia, and postischemic reperfusion.
83. Excitotoxicity in pathogenesis of CNS disorders.
84. General mechanisms of neuronal cell death in neurodegenerative diseases.