

# Portal:Questions for final examination in pathobiochemistry (1.LF, GM)

## Section I: Metabolites and enzymes

1. Basic characteristics of IEM
2. Pathogenic 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. Muscle glycogenoses and M.Pompe
15. Hereditary disorders of galactose and fructose metabolism
16. Hereditary disorders of protein glycosylation (CDG syndromes)
17. Disorders of 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 IEMs
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

1. Mechanism of tumor disease formation – overview
2. Physical factors participating in tumor induction
3. Chemical carcinogenesis
4. Viral carcinogenesis
5. Mechanisms of tumor transformation
6. Disorders of cell signaling pathways resulting in uncontrolled proliferation of tumor cells
7. Disorders of apoptotic signaling pathways in tumor cells
8. Disorders of DNA repair mechanisms in tumor cells
9. Molecular mechanisms of neovascularization and ways of their medical modification
10. Angiogenesis and neovascularization (differences, medical modification)
11. Molecular mechanisms of metastases formation, ways of medical modification
12. Selection of resistant tumor clones, medical modification
13. Tumor microenvironment: relationships among transformed cells and tumor stroma
14. Tumor stroma as a target for therapy
15. Pathology of signaling cascades regulating cellular proliferation: concept and examples
16. Targeted therapy: examples of therapeutical intervention at the molecular level in oncology
17. Hereditary cancer syndromes and sporadic tumor diseases
18. Techniques for analysis of mutations in inherited predispositions to cancer
19. Analysis of somatic mutations and microsatellite markers in sporadic tumors
20. Possibilities of detection of minimal residual disease
21. Purpose and types of anti-cancer treatment
22. Types of chemotherapeutics, their undesirable effects
23. Biochemical principles of chemotherapy and radiotherapy
24. Biochemical principles of hormonal and targeted therapy
25. Description and role of tumor markers in anti-cancer treatment
26. Sensitivity and specificity of tumor markers, examples
27. Cancer-and tissue-specific tumor markers, examples
28. Tumor markers - application and interpretation: screening, monitoring, diagnosis

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

1. Metabolic acidosis, its causes and consequences
2. Metabolic alkalosis, its causes and consequences
3. Combined disorders of acid-base equilibrium
4. 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.
5. Principal reactive oxygen and nitrogen species: properties, reactions, main sources in the body, role in pathogenesis
6. Physiological role of reactive oxygen species in metabolism: tissue hormones, phagocyte weapons, hydroxylases, redox signaling
7. Lipid peroxidation as an example of oxidative damage to biomolecules. Significance of transition metals (iron, copper) in pathobiochemistry of reactive oxygen species.
8. Antioxidant defense of human body
9. Biochemical basis of ageing. Radical/mitochondrial theory, ageing as catabolic failure, relationship to chronic inflammation
10. Role of mitochondria in cell death (apoptosis and necrosis) and physiological ageing
11. What a cell needs to become immortal? Autophagy, Hayflick limit, telomerase
12. 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.
13. Metabolic syndrome and insulin resistance – characteristic, cause and implication, possible therapeutic approach
14. Formation of AGEs, interaction AGE -RAGE, potential mechanisms to reduce formation/effect of AGEs
15. Mechanisms of hyperglycemia-induced tissue damage
16. Carbonyl stress, its role in pathogenesis of long-term diabetic complications, atherosclerosis and renal failure
17. Role of LDL in atherosclerosis
18. Role of HDL in atherosclerosis
19. Explain the biochemical processes during initial stages of atherosclerosis
20. Role of monocytes/macrophages, endothelium, smooth muscle cells and T lymphocytes in atherosclerosis
21. Rules of protein folding
22. Role of chaperones, proteasomes and lysosomes in the cell
23. Mechanism of prion diseases
24. Origins of pathological conformation of proteins and examples of clinical consequences
25. Endoplasmic reticulum stress
26. Metabolic alterations in cell during anoxia, ischemia, and postischemic reperfusion
27. Excitotoxicity in pathogenesis of CNS disorders
28. General mechanisms of neuronal cell death in neurodegenerative diseases