

Pathophysiology: Molecular Medicine Oral Presentations 2019/2020

HEMATO-ONCOLOGY AND IMMUNOLOGY

1. Role of fusion gene *bcr/ abl* in pathogenesis of leukemia
2. Fusion gene *bcr/ abl* and its products in diagnosis and therapy.
3. Etiology and pathogenesis of congenital and acquired methemoglobinemia (HbM, HbE, cytochrom b5 reductase)
4. Molecular mechanisms in pathophysiology of sickle cell disease (HbS)
5. Molecular mechanisms of alpha-thalassemia
6. Molecular mechanisms of beta-thalassemia
7. Tumor-suppressor genes and their role in cancer development.
8. Transcription factor defects in pathogenesis of acute myeloid leukemia.
9. Role of Bcl2 (B-cell leukemia/lymphoma 2) family proteins in the pathogenesis of non-Hodgkin lymphoma.
10. Transcription factor defects in pathogenesis of non-Hodgkin lymphoma.
11. Molecular mechanisms in the development of colorectal cancer.
12. Tissue macrophages and their role in tumor pathogenesis.
13. Regulation of erythropoietin gene expression
14. Angiogenic factors in the pathogenesis of solid tumors
15. Role of epigenetic modifiers (e.g. Tet, DNA methyltransferase, histone lysine N-methyltransferase) in tumor pathogenesis.
16. Telomerase and its role in tumor pathogenesis.
17. Causes and consequences of APC (activated protein C) resistance.
18. Targeted anticoagulation therapy (dabigatran, rivaroxaban, apixaban): mechanism and comparison to coumarin (warfarin)
19. Hematopoietic stem cell and bone marrow transplantation
20. Leukemia stem cell: its comparison to normal hematopoietic stem cell
21. Proteins p53, MDM2 in the cell cycle regulation and their role in tumorigenesis.
22. Proteasome system function and proteasome inhibitors in tumor therapy.
23. Molecular mechanisms of oncogene development.
24. Tumor suppressor genes and loss of heterozygosity: molecular mechanisms
25. Molecular mechanisms of retinoblastoma development: retinoblastoma (Rb) gene mutations and role of modifying genes
26. Tumor suppressor genes BRCA1, BRCA2: their function in the development of breast and ovarian cancer.

27. Paroxysmal nocturnal hemoglobinuria (PNH): molecular mechanism of disease.
28. Malignant transformation of hematopoietic cell: principals of leukemogenesis.
29. Cytokines and cytokine receptors in pathogenesis of congenital immunodeficiencies
30. Cytokines and cytokine receptors in myelopoiesis: possible therapeutical use in neutropenia
31. Cytokines and cytokine receptors in thrombopoiesis: possible therapeutical use in thrombocytopenia
32. Phosphatases in tumor pathogenesis.
33. TGF-beta signalling pathway in tumor pathogenesis.
34. Inhibition of angiogenesis in the therapy of solid tumors.
35. Biologic therapy and its use in malignant tumors
36. Biologic therapy and its use in autoimmune diseases.
37. Hereditary hemochromatosis.
38. Molecular mechanisms of hemophilia A
39. Molecular mechanisms and diagnosis of von Willebrand disease
40. Molecular mechanisms of thrombophilia
41. HLA antigens in the pathogenesis of diseases
42. Pathophysiology of graft versus host disease (GVHD)
43. Pathogenesis of Wilms tumor
44. Molecular mechanisms of porphyrias.
45. Molecular mechanism of cancer metastasis formation
46. Autoimmunity in disease pathogenesis: mechanisms of immune tolerance failure
47. Autoimmunity in disease pathogenesis: mechanisms of tissue damage
48. Role of immunity in transplantation
49. Neurotrophic factors and their receptors (Trk receptors) in tumor pathogenesis.
50. Congenital polycythemias.
51. Role of extracellular vesicles (microvesicles, exosomes) in pathogenesis of hematopoietic diseases.
52. Role of extracellular vesicles (microvesicles, exosomes) in pathogenesis of oncologic diseases.
53. Role of apoptosis in pathogenesis of hematooncologic diseses.
54. Membrane pattern recognition receptors (PRR) in pathogenesis of infectious and inflammatory diseases.
55. Intracellular pattern recognition receptors (PRR) in pathogenesis of of infectious and inflammatory diseases.

ENDOCRINOLOGY AND METABOLISM

56. Molecular mechanisms and origins of male pseudo-hermaphroditism
57. PTH receptor mutation – defects of bone metabolism.
58. Molecular mechanisms of multiple endocrine neoplasia syndrome 1 (MEN1 syndrome)
59. Molecular mechanisms of multiple endocrine neoplasia syndrome 2 (MEN 2 syndrome)
60. Receptor and post receptor mechanisms of resistance to insulin.
61. Molecular mechanisms of congenital obesity (leptin, POMC, MC4, PPAR-gamma, and others)
62. Molecular mechanisms of hypercellular and hypertrophic obesity.
63. Endocrine function of fat tissue.
64. Molecular mechanism of congenital hyperlipoproteinemia.
65. Regulation of Langerhans islets beta-cells and their genetic defects.
66. Signaling pathways activated by insulin and glucagon in the pathogenesis of diabetes mellitus type II.
67. Receptor mediated endocytosis defect in pathogenesis of hypercholesterolemia.
68. Molecular mechanisms in pathogenesis of congenital forms of diabetes mellitus.
69. Molecular mechanism of diabetes mellitus type 1 pathogenesis.
70. Molecular mechanisms in the regulation of appetite: orexigenic and anorexigenic factors.
71. Molecular mechanisms of adrenogenital syndromes
72. Congenital defects of thyroid gland function.
73. Insulin receptors in pathogenesis of diseases.
74. Receptor mediated apoptosis in the pathogenesis of autoimmune diseases.

NEUROLOGY

75. Molecular mechanisms of Alzheimer disease
76. Apoptosis in the pathogenesis of neural system diseases
77. Molecular mechanisms of transmissible spongiform encephalopathy (TSE).
78. Tau protein and its role in the pathogenesis of neurodegenerative diseases.
79. Alfa-synuclein and its role in the pathogenesis of neurodegenerative diseases.

80. Amyloid beta A4 protein (APP) and its paralog amyloid-like protein 1 (APLP) and their role in the pathogenesis of neurodegenerative diseases.
81. Pathogenesis of sclerosis multiplex: molecular mechanisms of demyelization.
82. Neurotrophic factors and their receptors (Trk receptors) in the pathogenesis of neurodegenerative diseases.
83. Neurotrophic factors and their receptors (Trk receptors) in the pathogenesis of neurologic developmental defects.
84. Molecular and genetic mechanism of Parkinson disease
85. Molecular and genetic mechanism of Huntington disease (chorea)

RESPIRATORY SYSTEM

86. Diseases associated with α_1 -antitrypsin deficiency
87. Molecular mechanism of cystic fibrosis.

CARDIOVASCULAR DISEASES

88. Etiology and pathogenesis of hypertrophic cardiomyopathy.
89. Molecular mechanisms of dilated cardiomyopathy
90. Molecular mechanisms of endothelial activation and its consequences.
91. Hormonal and cytokine changes in cardiac failure
92. Molecular mechanisms in arrhythmias associated with genetic defects of ion channels.
93. Endocrine and paracrine factors in pathogenesis of atherosclerosis
94. Tyrosine kinase receptors in angiogenesis and vasculogenesis.
95. Role of extracellular vesicles (microvesicles, exosomes) in endothelial damage.

GASTROINTESTINAL SYSTEM

96. Molecular mechanism of liver regeneration and fibrosis
97. Cytochrome P450 (CYP) polymorphism - pharmacogenetics
98. Mechanism of cholera toxin and pertussis toxin effects on enterocytes.
99. Adenomatous polyposis coli gene and its role in familial adenomatous polyposis.
100. Molecular mechanism of Wilson disease
101. Molecular mechanisms of hereditary hemochromatosis
102. Molecular mechanisms of congenital defects of bile production.

103. Molecular mechanisms of bilirubine metabolism defects.

BONE, JOINTS, AND CONNECTIVE TISSUE

104. Molecular mechanism of hereditary myopathies
105. Molecular mechanism of osteogenesis imperfecta
106. Molecular mechanism of Marfan and Ehlers-Danlos syndromes

UROGENITAL SYSTEM

107. Pathophysiology of autosomal dominant hereditary interstitial nephropaties
108. Molecular mechanism of kidney tubulopathias
109. Molecular pathophysiology of IgA nephropathy

EXPERIMENTAL MEDICINE AND NEW DIAGNOSTIC APPROACHES

110. Use of PCR in identification of known mutations (RFLP, ARMS)
111. Use of PCR in therapy efficacy monitoring.
112. Methods of DNA sequencing and their use in diagnostic process (Sanger sequencing, “next generation” sequencing, mass spectroscopy sequencing)
113. Principals of Southern and northern blotting and their use in diagnosis of diseases
114. DNA fingerprinting a its use in diagnostics
115. Principle of RNAseq a its use in medical research: perspectives in diagnosis
116. Bio-chip technologies in medical research: perspectives in diagnosis
117. Experimental methods of functional inactivation of the gene
118. Principals of RNA-interference (RNAi) and its use in biomedical research
119. Use of transgenic organisms in biomedical research
120. Tumor biomarkers: mechanism of production and application possibilities
121. Inflammation biomarkers: mechanism of production and application possibilities
122. Cloning (experimental, therapeutic, reproductive): principals and significance.
123. Potential of stem cells in disease therapy
124. Embryonic stem cells (ESC) definition and function.
125. Mesenchymal stromal (stem) cells (MSC) definition, function, and possible therapeutic use.

GENERAL MECHANISMS

126. Molecular mechanism of hypoxia sensing and function of hypoxia inducible factor (HIF)

127. Role of hypoxia in disease pathogenesis
128. Hypoxia inducible genes and molecular mechanism of hypoxia inducible genes regulation.
129. Role of adhesion molecules in leukocyte migration and pathogenesis of inflammation
130. Mediators of inflammatory response.
131. Mitochondrial diseases
132. Regulation of acute phase proteins production in infection and inflammation.
133. Function of acute phase proteins in infection and inflammation.
134. Oxidative stress in disease pathogenesis and effects of antioxidants
135. Mechanism of apoptosis triggered by extrinsic (receptor) apoptotic pathway activation.
136. Deregulation of cell cycle in disease pathogenesis
137. Cell aging: telomeres and telomerase.
138. Type of receptors and their proximal signaling mechanisms
139. Cytokines and cytokine receptors: JAK/STAT signaling
140. G-protein receptors (GPCR) mechanism of activation and signaling
141. Adaptor molecules in signal transduction (e.g. cAMP, cGMP, Ca²⁺, DAG, IP₃)
142. Protein kinases: classification and function.
143. Phospholipase C and its second messengers, DAG, IP₃, Ca²⁺, principal of signaling pathway function (calmoduline, protein-kinase C, etc.)
144. Mitosis blockers: mechanism of action and their use in therapy
145. Copy-number variations (CNV) in pathogenesis of diseases.
146. mTOR in the regulation of cell metabolism and its role in pathologic conditions.
147. miRNA its function and role in pathogenesis of diseases.
148. rRNA its function and role in pathogenesis of diseases.
149. lncRNA its function and role in pathogenesis of diseases.
150. piRNA its function and role in pathogenesis of diseases.
151. snRNA a snoRNA its function and role in pathogenesis of diseases.
152. DNA methylation in the regulation of gene expression and in pathogenesis of diseases.
153. Histone deacetylation in the regulation of gene expression and in pathogenesis of diseases.
154. Therapeutic targeting of epigenome.
155. Transcription factors and their role in pathogenesis of non-malignant diseases.

156. Chaperons in protein transport and diseases associated with defective transport function of chaperons.
157. Protein folding defects and the role of chaperons in this process.
158. Role of signaling peptide in protein translocation to ER: general consequences of signal peptide pathologic variants.
159. Role of MAP kinases in cell proliferation
160. Function of p53 in gene repair and apoptosis.
161. Pathogenesis of acute high-altitude illness.
162. Endoplasmic reticulum stress in disease pathogenesis.