

Pathophysiology: Molecular Mechanisms of Disorders

Oral Presentations

SUGGESTED OUTLINE OF PRESENTATION

- Brief introduction (clinical and/or experimental context of the presentation)
- Description and explanation of molecular mechanisms that are involved in the etiopathogenesis of presented disorder
- Short summary (e.g. implications for diagnosis, therapy, prognosis, etc.)
- Overview of used literature and other sources
- Powerpoint or PDF presentation (max. duration 7 minutes, i.e. ~ 7 slides) [if the topic is extensive – select only part of the topic, so the presentation makes sense and molecular mechanisms can be clearly described]

HEMATO-ONCOLOGY AND IMMUNOLOGY

1. Fusion gene *bcr/ abl* in the pathogenesis of leukemia (CML, AML, ALL)
2. *Bcr-abl* in diagnosis and therapy of leukemia
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 factors in the pathogenesis of leukemia (e.g. MLL–AF9, RUNX1–ETO, PML–RAR α)
9. Transcription factors in the pathogenesis of breast cancer (e.g. SIX1, RUNX2, FOXO)
10. Role of Bcl2 (B-cell leukemia/lymphoma 2) family proteins in the pathogenesis of non-Hodgkin lymphoma.
11. Transcription factors in pathogenesis of non-Hodgkin lymphoma (e.g. MYC)
12. Estrogen receptor in the etiopathogenesis of breast cancer.
13. Molecular mechanisms in the development of colorectal cancer.
14. Tissue macrophages and their role in tumor pathogenesis.
15. Angiogenic factors in the pathogenesis of solid tumors
16. Role of epigenetic modifiers (e.g. Tet, DNA methyltransferase, histone-lysine N-methyltransferase) in tumor pathogenesis.
17. Telomerase and its role in tumor pathogenesis.
18. Causes and consequences of APC (activated protein C) resistance.
19. Targeted anticoagulation therapy (dabigatran, rivaroxaban, apixaban): mechanism and comparison to coumarin (warfarin)

20. Hematopoietic stem cells and bone marrow transplantation
21. Leukemia / tumor stem cells
22. MDM2 in the cell cycle regulation and its role in tumorigenesis.
23. Proteasome system function and proteasome inhibitors in tumor therapy.
24. Tumor suppressor genes and molecular mechanisms of loss of heterozygosity in tumor cells.
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 signaling pathway in tumor pathogenesis.
34. Inhibition of angiogenesis in the therapy of solid tumors.
35. Tumor cell surface antigens as targets in the therapy of tumors
36. Targeting of immune-checkpoint molecules for the therapy of tumors
37. Immune cells and immune signaling molecules as targets in therapy of autoimmune diseases.
38. Hereditary hemochromatosis.
39. Molecular mechanisms of hemophilia A
40. Molecular mechanisms and diagnosis of von Willebrand disease
41. Molecular mechanisms of thrombophilia
42. HLA antigens in the pathogenesis of diseases
43. Pathophysiology of graft versus host disease (GVHD)
44. Molecular mechanisms of porphyrias.
45. Molecular mechanism of cancer metastasis formation
46. Autoimmunity in disease pathogenesis: mechanisms of immune tolerance failure
47. Role of immunity in transplantation
48. Neurotrophic factors and their receptors (Trk receptors) in tumor pathogenesis

49. Congenital polycythemias
50. Role of extracellular vesicles (microvesicles, exosomes) in pathogenesis of hematopoietic diseases.
51. Role of extracellular vesicles (microvesicles, exosomes) in pathogenesis of oncologic diseases.
52. Membrane pattern recognition receptors (PRR) in pathogenesis of infectious and inflammatory diseases.
53. Intracellular pattern recognition receptors (PRR) in pathogenesis of infectious and inflammatory diseases.
54. Congenital malformations of lymphatics: etiology and pathogenesis

ENDOCRINOLOGY AND METABOLISM

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

73. Congenital defects of thyroid gland function.
74. Insulin receptors in pathogenesis of diseases.
75. Molecular mechanisms in the etiopathogenesis of achondroplasia

NEUROLOGY

76. Tau protein and its role in the pathogenesis of neurodegenerative diseases.
77. Alfa-synuclein and its role in the pathogenesis of neurodegenerative diseases.
78. Amyloid beta A4 protein (APP) and its paralog amyloid-like protein 1 (APLP) and their role in the pathogenesis of neurodegenerative diseases.
79. Pathogenesis of sclerosis multiplex: molecular mechanisms of demyelization.
80. Neurotrophic factors and their receptors (Trk receptors) in the pathogenesis of neurologic diseases.
81. Molecular and genetic mechanism of Parkinson disease
82. Molecular and genetic mechanism of Huntington disease (chorea)

RESPIRATORY SYSTEM

83. Molecular mechanisms of α_1 -antitrypsin deficiency associated lung disease
84. Molecular mechanism of cystic fibrosis.
85. Genetic disorders causing surfactant dysfunction
86. Etiology and pathogenesis of newborn respiratory distress syndrome
87. Etiology and pathogenesis of chronic bronchitis
88. Etiology and pathogenesis of emphysema
89. Etiology and pathogenesis of heritable pulmonary hypertension
90. Persistent pulmonary hypertension of the newborn (PPHN)
91. Etiology and pathogenesis of idiopathic pulmonary fibrosis

CARDIOVASCULAR DISEASES

92. Etiology and pathogenesis of hypertrophic cardiomyopathy.
93. Molecular mechanisms of dilated cardiomyopathy
94. Molecular mechanisms of endothelial activation and its consequences.
95. Hormonal and cytokine changes in cardiac failure
96. Molecular mechanisms in arrhythmias associated with genetic defects of ion channels.
97. Endocrine and paracrine factors in pathogenesis of atherosclerosis
98. Tyrosine kinase receptors in angiogenesis and vasculogenesis.

GASTROINTESTINAL SYSTEM

99. Molecular mechanism of liver regeneration and fibrosis
100. Mechanism of cholera toxin and pertussis toxin effects on enterocytes.
101. Adenomatous polyposis coli gene and its role in familial adenomatous polyposis.
102. Molecular mechanism of Wilson disease
103. Molecular mechanisms of congenital defects of bile production.
104. Molecular mechanisms of bilirubin metabolism defects.
105. Molecular mechanisms of α_1 -antitrypsin deficiency associated liver disease

BONE, JOINTS, AND CONNECTIVE TISSUE

106. Molecular mechanism of hereditary myopathies
107. Molecular mechanism of osteogenesis imperfecta
108. Molecular mechanism of Marfan and Ehlers-Danlos syndromes

UROGENITAL SYSTEM

109. Etiology and pathogenesis of autosomal dominant tubulointerstitial kidney diseases (ADTKD)
110. Etiology and pathogenesis of polycystic kidney disease (PKD/PCKD)
111. Molecular pathophysiology of IgA nephropathy
112. Molecular mechanisms in the etiology and pathogenesis of nephrotic syndrome
113. Pathophysiology of Alport syndrome

EXPERIMENTAL MEDICINE AND NEW DIAGNOSTIC APPROACHES

114. Use of PCR in identification of known mutations (RFLP, ARMS)
115. Use of PCR in therapy efficacy monitoring.
116. Methods of DNA sequencing and their use in diagnostic process (Sanger sequencing, “next generation” sequencing, mass spectroscopy sequencing)
117. Principles of Southern and northern blotting and their use in diagnosis of diseases
118. DNA fingerprinting and its use in diagnostics
119. Principle of RNAseq and its use in medical research: perspectives in diagnosis
120. Bio-chip technologies in medical research: perspectives in diagnosis

121. Experimental methods of functional inactivation of the genes
122. Principals of RNA-interference (RNAi) and its use in biomedical research
123. Use of transgenic organisms in biomedical research
124. Tumor biomarkers: mechanism of production and application possibilities
125. Inflammation biomarkers: mechanism of production and application possibilities
126. Cloning (experimental, therapeutic, reproductive): principals and significance.
127. Stem cell types and their potential in disease therapy
128. Embryonic stem cells (ESC) definition and function.
129. Mesenchymal stromal (stem) cells (MSC) definition, function, and possible therapeutic use.

GENERAL MECHANISMS

130. Molecular mechanism of hypoxia sensing (HIF) and its involvement in etiopathogenesis of diseases
131. Role of hypoxia in disease pathogenesis
132. Molecular mechanism of hypoxia-inducible genes regulation.
133. Role of adhesion molecules in leukocyte migration and pathogenesis of inflammation
134. Mediators of inflammatory response.
135. Mitochondrial diseases
136. Regulation of acute phase proteins production in infection and inflammation.
137. Function of acute phase proteins in infection and inflammation.
138. Oxidative stress in disease pathogenesis and effects of antioxidants
139. Apoptosis triggered by extrinsic (receptor) apoptotic pathway activation and its role in pathogenesis of diseases.
140. Deregulation of cell cycle in disease pathogenesis
141. Cell aging: telomeres and telomerase.
142. Type of receptors and their proximal signaling mechanisms
143. Cytokines and cytokine receptors: JAK/STAT signaling in disease etiopathogenesis
144. G-protein receptors (GPCR) mechanism of activation and signaling
145. Adaptor molecules in signal transduction (e.g. cAMP, cGMP, Ca²⁺, DAG, IP₃)
146. Protein kinases: classification and functions
147. Phospholipase C and its second messengers, DAG, IP₃, Ca²⁺, principal of signaling pathway function (calmoduline, protein-kinase C, etc.)

148. Mitosis blockers: mechanism of action and their use in therapy
149. Copy-number variations (CNV) in pathogenesis of diseases.
150. mTOR in the regulation of cell metabolism and its role in pathologic conditions.
151. mikroRNA its function and role in pathogenesis of diseases.
152. rRNA its function and role in pathogenesis of diseases.
153. lncRNA its function and role in pathogenesis of diseases.
154. piRNA its function and role in pathogenesis of diseases.
155. snRNA a snoRNA its function and role in pathogenesis of diseases.
156. DNA methylatiion in the regulation of gene expression and in pathogenesis of diseases.
157. Histone deacetylation ce in the regulation of gene expression and in pathogenesis of diseases.
158. Transcription factors and their role in pathogenesis of non-malignant diseases.
159. Proteostasis and its role in disease etiopathogenesis
160. Chaperons in etiology and pathogenesis of diseases
161. Protein folding defects and the role of chaperons in this process.
162. Endoplasmic reticulum stress and unfolded protein response in disease pathogenesis
163. Failure of protein quality control mechanisms in the pathogenesis of diseases
164. Role of signaling peptide in protein translocation to ER: general consequences of signal peptide pathologic variants.
165. Role of MAP kinases in cell proliferation and disease etiopathogenesis
166. Pathogenesis of acute high-altitude illness