Learning objectives in Genetics

IV Semester, Module N14 „Environment and Health“

1. The molecular basis of genetic disease
2. The effect of mutation on protein function
3. Allelic and locus heterogeneity
4. Modifier genes
5. Hematological genetics and disorders
6. Structure and function of hemoglobin
7. Developmental expression of globin genes and globin switching
8. Locus control region
9. Major classes of hemoglobin structural variants
10. Hb Kempsey
11. Hb Hammersmith
12. Hb Hyde Park
13. Hemoglobinopathies
14. Sickle cell anemia
15. Hereditary persistence of fetal hemoglobin
16. Role of modifier genes on the expression of HbF (BCL11A and MYB)
17. Thalassemia: α and β thalassemia; molecular and biochemical basis, etiology, pathogenesis, phenotype, management, inheritance risk
18. Genetics of metabolic disorders
19. Principles of biochemical genetic diseases due to mutations in different classes of proteins
20. Aminoacidopathies
21. Hyperphenylalaninemias
22. allelic and locus heterogeneity in hyperphenylalaninemias
23. lysosomal storage diseases
24. Tay-Sachs disease- molecular and biochemical basis, etiology, pathogenesis, phenotype, management, inheritance risk

25. Gaucher disease - molecular and biochemical basis, etiology, pathogenesis, phenotype, management, inheritance risk
26. Loss of protein function due to impaired binding or metabolism of cofactors – homocystinuria - molecular and biochemical basis, etiology, pathogenesis, phenotype, management, inheritance risk
27. Mutations of an enzyme inhibitor: α1-antitrypsin deficiency - molecular and biochemical basis, etiology, pathogenesis, phenotype, management, inheritance risk
28. Loss of glycosylation - I-cell disease - molecular and biochemical basis, etiology, pathogenesis, phenotype, management, inheritance risk
29. Dysregulation of a biosynthetic pathway - acute intermittent porphyria - molecular and biochemical basis, etiology, pathogenesis, phenotype, management, inheritance risk
30. Defects in receptor proteins
31. Familial hypercholesterolemia, genes associated with familial hypercholesterolemia
32. Classes of mutations in the LDL receptor
33. The PCSK9 protease and it's link with LDL cholesterol
34. Cystic fibrosis - molecular and biochemical basis, etiology, pathogenesis, phenotype, management, inheritance risk
35. CFTR gene and protein
36. Genetics of cystic fibrosis
37. Mutations in the CFTR Polypeptide
38. A cystic fibrosis genocopy
39. Mutations in the epithelial sodium channel gene SCNN1
40. Disorders of structural proteins
41. Duchenne, Becker, and other muscular dystrophies - molecular and biochemical basis, etiology, pathogenesis, phenotype, management, inheritance risk
42. Collagenopathies - osteogenesis imperfecta - molecular and biochemical basis, etiology, pathogenesis, phenotype, management, inheritance risk
43. Neurodegenerative disorders
44. Alzheimer Disease - etiology, pathogenesis, phenotype, management, inheritance risk
45. Mitochondrial diseases - etiology, pathogenesis, phenotype, management, inheritance risk
46. MELAS

47. MERF
48. Leigh syndrome
49. Kearns-Sayre syndrome (KSS)
50. Progressive sensorineural deafness
51. Diseases due to the expansion of unstable repeat sequences; the pathogenesis of diseases due to unstable repeat expansions
52. Huntington disease - etiology, pathogenesis, phenotype, management, inheritance risk
53. Fragile X syndrome - etiology, pathogenesis, phenotype, management, inheritance risk
54. Fragile X tremor/ataxia syndrome
55. Friedreich ataxia - etiology, pathogenesis, phenotype, management, inheritance risk
56. Myotonic dystrophy 1 and 2 - etiology, pathogenesis, phenotype, management, inheritance risk