

Learning objectives in Genetics

IV Semester, Module N13 „Organism and Ecosystem“

1. The relative contributions of genes and environment to complex disease
2. Familial aggregation and correlation
3. Concordance and discordance
4. Gaussian plot
5. The normal distribution
6. Twin studies
7. Case-control studies
8. Characteristics of inheritance of complex diseases.
9. Examples of common multifactorial diseases with a genetic contribution
10. Multifactorial congenital malformations
11. Neuropsychiatric disorders
12. Coronary artery disease
13. Digenic inheritance
14. gene-environment interactions in venous thrombosis
15. Type 1 diabetes mellitus
16. Alzheimer disease
17. Methods of risk calculation in genetic diseases
18. The Hardy-Weinberg law
19. Allele and genotype frequencies in populations
20. The Hardy-Weinberg law in autosomal and X-linked diseases
21. Factors that disturb Hardy-Weinberg equilibrium
22. Stratification
23. Exceptions to large populations with random mating
24. The factors affecting constant allele frequency
25. Ethnic differences in the frequency of various genetic diseases

26. Founder effect
27. Gene flow
28. Heterozygote advantage
29. Drift versus heterozygote advantage
30. Ancestry informative markers
31. Population genetics and race
32. Methods of prenatal diagnosis and indications for prenatal diagnosis by invasive and noninvasive testing
33. Prenatal screening
34. Laboratory studies in prenatal diagnosis – cytogenetic, biochemical and fetal genome analysis
35. Genetic counseling for prenatal diagnosis and screening
36. Personalized medicine
37. Screening for genetic susceptibility to disease
38. Genetic screening in populations
39. Newborn screening
40. Heterozygote screening
41. Pharmacogenomics
42. Adverse drug reactions
43. Variation in drug metabolism
44. Cytochrome p-450 variants
45. Strategies used in genetic epidemiology
46. Personalized genomic medicine.
47. Family history in risk assessment
48. Indications for genetic counseling
49. Determining and managing the risk for disease recurrence
50. Psychological aspects
51. Genetic counseling for complex disorders and consanguinity
52. Molecular and genome-based diagnostics – gene panels, exome sequencing; variant

interpretation

53. Ethical and social issues in genetics and genomics
54. Principles of biomedical ethics
55. Ethical dilemmas arising in medical genetics
56. Privacy of genetic information