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Medical genetics syllabus for pharmacy students 2020/2021

1. Cellular and molecular basis of inheritance. Organization of the human genome. Nuclear and mitochondrial DNA.Epigenome.
2. Hereditary information. Gene structure and function. Regulation of gene function.
3. Chromosomal organization of the hereditary material. Human karyotype.
4. Chromosomal aberrations.
5. Chromosomal diseases – Down syndrome, Edwards syndrome.
6. Chromosomal diseases- Klinefelter syndrome, Turner syndrome.
7. Patterns of inheritance. Criteria of autosomal – dominant inheritance. Osteogenesis imperfect. Marfan syndrome. Familial hypercholesterolemia.
8. Patterns of inheritance. Criteria of autosomal – recessive inheritance. Cystic fibrosis. Beta-thalassemia. Phenylketonuria.
9. Patterns of inheritance. Criteria of sex-linked inheritance. Haemophilia A and B. Muscular dystrophy type Duchenne/Becker.
10. Mutations and mutagenesis. Mutational potential of drugs.
11. Types of gene mutations. Role of mutagensesis in the etiology of human pathology.
12. Methods for genetic analysis.
13. Genetic regulation of the cell cycle.
14. Protooncogenes - normal function and mechanisms of activation.
15. Tumor-suppressor genes - normal function and mechanisms of inactivation and their role in familial cancer.
16. Apoptosis – mechanisms and factors of the programmed cell death.
17. Congenital anomalies and dysmorphic syndromes. Teratogenic factors. Teratogenic potential of drugs.
18. Pharmacogenetics. Definition. Pharmacogenetic defects. Principal pathogenetic mechanisms of adverse drug reactions.
19. Pharmacogenetic defects. Adverse drug reactions, caused by glucose-6-phospate dehydrogenase deficiency.
20. Pharmacogenetic defects. Adverse drug reactions, caused by atypical butyrilcholinesterase. Malignant hyperthermia.
21. Pharmacogenetic defects. Adverse drug reactions, caused by defects of enzymes, involved in glutathion synthesis, methemoglobinreductase defect, abnormal hemoglobins, catalase deficiency.
22. Polymorphic pharmacogenetic defects. Genetic variations of alcoholdehydrogenase, aldehyddehydrogenase and N-acetyltransferase (NAT2).TPMT.
23. Genetic polymorphism of cytochrome P450 monooxygenases and drug metabolism. CYP2C9, CYP2D6, CYP3A4, CYP2C19.
24. Conventional therapy for genetic diseases.
25. Gene therapy for monogenic diseases – principles, possibilities, application. CRISPR gene editing system.
26. Antisense gene therapy and tumor-suppressor gene therapy - principals, possibilities, application. mRNA based anti-tumor vaccines.

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27. Target therapy for oncological diseases- principles, possibilities, application. Orphan drugs in medicine.
28. Target therapy for breast cancer.
29. Target therapy for lung cancer.
30. Target therapy for colon cancer
31. Target therapy for chronic myelogenic leukemia.
32. Medical-genetic counseling – organization, goals, indications. Genetic risk and genetic prognosis.
33. Prevention of hereditary diseases – prenatal diagnosis – indications, approaches, methods.
34. Prevention of hereditary diseases – general and selective screening – principles and methods.

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HEAD OF THE DEPARTMENT OF MEDICAL GENETICS
PROF. SAVINA HADZHIDEKOVA, MD, PhD

LITERATURE:

1. Genomic medicine, 2016, Sofia, Toncheva, D., Ganev V.
2. Medical genetics workbook for students in Pharmacy, D.Toncheva, S.hadzhidekova, R.Staneva, Simel pres, Sofia, 2018 ISBN:978-619-183-065-7,
3. Emery's Elements of Medical genetics,15th ed.
4. Genetics home reference - https://ghr.nlm.nih.gov/