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Syllabus for practical semester exam for Medical students

2020/2021

1. Building a pedigree
2. Genetic anamnesis
3. Determining type of inheritance and risk calculation in AR, AD, XD, XR, mitochondrial inheritance
4. Diagnosis, treatment and prevention of beta-thalassemia major. Heterozygous carrier screening - beta-thalassemia minor
5. Diagnosis, treatment and prevention of cystic fibrosis
6. Diagnosis, treatment and prevention of PKU
7. Diagnosis, treatment and prevention of galactosemia
8. Diagnosis, treatment and prevention of hereditary hypercholesterolemia
9. Diagnosis, treatment and prevention of Huntington's disease
10. Diagnosis, treatment and prevention of myotonic dystrophy
11. Diagnosis, treatment and prevention of *FMR1* associated diseases
12. Diagnosis, treatment and prevention of OI type I and type II
13. Diagnosis, treatment and prevention of Marfan syndrome
14. Diagnosis, treatment and prevention of SMA
15. Diagnosis, treatment and prevention of Duchenne / Becker muscular dystrophy
16. Diagnosis, treatment and prevention of mitochondrial diseases - LHON
17. Balanced karyotype - Robertsonian and reciprocal translocations. Associated risks. Risk estimation.
18. Diagnosis, treatment and prevention of Down syndrome. Cytogenetic forms and recurrence risk assessment
19. Diagnosis, treatment and prevention of Patau syndrome
20. Diagnosis, treatment and prevention of Edwards syndrome
21. Diagnosis, treatment and prevention of Turner syndrome and its variants
22. Diagnosis, treatment and prevention of Klinefelter syndrome
23. Diagnosis, treatment, risk assessment and prevention of Prader-Willi syndrome
24. Diagnosis, treatment, risk assessment and prevention of Angelman syndrome
25. Diagnosis, treatment, risk assessment and prevention of DiGeorge syndrome
26. Diagnosis, treatment, risk assessment and prevention of Williams-Boyren syndrome
27. Diagnosis, treatment, risk assessment and prevention of Familial adenomatous polyposis
28. Diagnosis, treatment, risk assessment and prevention of Lynch syndrome
29. Diagnosis, treatment, risk assessment and prevention of BRCA 1 and 2 associated inherited breast and ovarian cancer
30. Genetic markers for targeted therapy in breast cancer
31. Genetic markers for targeted therapy in CRC
32. Genetic Markers for Targeted Therapy at the NSCLC



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33. Genetic markers for targeted therapy in CML
34. Indications and prenatal and pre-implantation diagnosis

1. Genomic medicine, 2016, Sofia, Toncheva, D., Ganey V. ISBN 978-619-183-043-5
2. Medical genetics. Practical course for medical students, Toncheva, D., Hadjidekova S., 2020, Publisher: ARSO
3. Emery's Elements of Medical genetics, 14th ed. ISBN: 978-0702066856
4. Genetics home reference - <https://ghr.nlm.nih.gov/>

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HEAD OF THE DEPARTMENT OF MEDICAL GENETICS
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