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| **Medical Biology and Genetics - Part II: GENETICS** | |
| **1** | **History of genetics**; Model organisms in genetic analysis. |
| **2** | **Mendelian genetics**; Mendel's laws; Examples in human genetics |
| **3** | **Allelic heterogeneity and allelic interactions**; Examples and relevance in human disorders |
| **4** | **Basic types and relevance of DNA polymorphisms**; Examples |
| **5** | **Basic types of mutations distinguished by cell type** having a mutation, relevance for human diseases; Explain the difference between the terms "mutation" and "polymorphism" |
| **6** | **Basic types of mutations distinguished by the change in DNA sequence** and consequences on the expression and primary structure of the encoded protein, relevance for human diseases |
| **7** | **Basic types of mutations distinguished by an impact on function of the encoded protein,** relevance for human diseases; Haploinsufficiency |
| **8** | **Basic types of mutations distinguished by location of a mutation within a gene,** relevance for human diseases |
| **9** | **Epigenetics**, types of epigenetic changes, biological mechanisms and relevance of epigenetic changes to human biology and diseases; Examples |
| **10** | **Chromosome structure, classification and nomenclature;** Basic methods of chromosome analysis, karyotyping; Karyotype designation |
| **11** | **Genomic mutations, causes and consequences in human** |
| **12** | **Numerical chromosomal mutations,** types, sources, consequences, examples |
| **13** | **Structural chromosomal mutations,** types, sources, consequences, examples |
| **14** | **Nondisjunction** and its consequences |
| **15** | **Germinal and somatic karyotypic changes**, examples and biological explanation of resulting human syndromes |
| **16** | **Gene linkage and its use for gene mapping** |
| **17** | **Gene interactions**, types, examples; Modifier genes - examples in human genetics or animal models |
| **18** | **Meiosis,** its course and relevance; Describe meiosis using the variables C and n |
| **19** | **Genetic determination of sex**, basic molecular processes and their potential disturbances |
| **20** | **Gene dosage compensation,** its relevance and basic molecular mechanisms |
| **21** | **Gametogenesis,** comparison of spermatogenesis and oogenesis. **Fertilization,** basic molecular processes; What are the main differences in the contribution of egg and sperm to the zygote? Why parthenogenesis is not possible in mammals? |
| **22** | **Monogenic diseases,** criteria for distinguishing basic types of inheritance in pedigrees |
| **23** | **Sex-related inheritance;** Types, pedigrees, examples |
| **24** | **Dominant diseases**, examples of pedigrees and types of mutations that may underlie them |
| **25** | **Recessive diseases**, examples of pedigrees and types of mutations that may underlie them; Inbreeding and its genetic consequences. |
| **26** | **Cystic fibrosis**, genetics and molecular biology |
| **27** | **Penetrance and expressivity**, examples and possible underlying mechanisms |

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| **28** | **Basic principles of pedigree drawing and its significance in medicine**, distinguishing features of basic inheritance patterns; **Locus heterogeneity** - how could it be identified in human genetic diseases? Cite examples |
| **29** | **Risk assessment and genetic prognosis**; In what circumstances would genetic prognosis be preferred over DNA diagnostics? |
| **30** | **Population genetics**; Hardy-Weinberg law and processes changing allelic frequencies |
| **31** | **Complex diseases and heritability,** examples; Phenocopy and genotype - environment interactions including examples; Twin studies and their importance in medical genetics |
| **32** | **Genetic predisposition to cancer;** Benign and malignant tumours and their characteristics; Acquired capabilities of malignant tumours (= Hallmarks of Cancer) |
| **33** | **Tumour suppressor genes** and molecular alterations found in them in tumours; Examples; Loss of heterozygosity, its causes and consequences |
| **34** | **Oncogenes, proto-oncogenes** and types of mutations found in them in tumours; Examples; Philadelphia chromosome |
| **35** | **Mendelian and non-Mendelian inheritance** - comparison; Examples of non-Mendelian inheritance - in pedigrees, diseases/syndromes |
| **36** | **Immunogenetics and MHC antigens** (basic two classes and their comparison - structure, function...) and their interactions; Their genes and haplotypes; Consequences of allogeneic transplantations |
| **37** | **T- and B-lymphocyte interaction** during the antibody synthesis and its malfunction during the genetic diseases of immunity; Antigen presentation (MHC restriction) |
| **38** | **Antibodies** - basic structure, their production and function, and principles of generation of antibody diversity; Monoclonal and polyclonal antibodies and their use in medicine |
| **39** | **Blood group systems,** their genetic determination, comparison, examples and their clinical relevance; Basic immunologic differences between blood group systems AB0 and Rh; Bombay phenotype |
| **40** | **Fragile X syndrome and Huntington disease** |
| **41** | **Structure of haemoglobin, globin genes**, their organization and regulation, regulation of haemoglobin synthesis during development, role of chaperone AHSP, basic classification of hemoglobinopathies |
| **42** | **Diseases due to structural haemoglobin changes** - examples, genetics and molecular biology |
| **43** | **Thalassaemias** - classification, genetics, types of mutations, and molecular biology |
| **44** | **Pharmacogenetics;** Xenobiotic metabolism and its phases; Enzymes; Personalized medicine; Examples |
| **45** | **Pharmacogenetics; Warfarin metabolism;** Enzymes; Drug interactions |
| **46** | **Unstable repeat disorders** - examples, genetics and molecular biology; Genetic anticipation |
| **47** | **Polycystic kidney disease** - genetics and molecular biology |
| **48** | **PCR method;** Definition, types, particular steps; Results; Use in human genetics / medicine |
| **49** | **Repetitive sequences**, microsatellites and their use in DNA diagnostics/forensic medicine |
| **50** | **DNA diagnostics**. Basic types and their advantages and disadvantages, sources of errors |