BIOL2323: GENERAL GENETICS

STUDY GUIDE

Mendelian and “non-Mendelian“ Genetics

be prepared to...

- explain features of the experimental design that made Mendel’s experiments so successful
- set up monohybrid and dihybrid crosses
- know how to use a Punnett square
- know what a test cross is
- know what reciprocal crosses are
- know Mendel’s laws (uniformity, segregation, independent assortment)
- know the basic laws of probability (product, sum)
- know the basic genetic terminology: gene, allele, locus, phenotype, genotype, wild type, homozygous, heterozygous, recessive, dominant
- know symbols used in pedigree analysis
- determine if a disease is dominant, recessive, autosomal, or X-chromosomal using pedigree analysis
- explain what multifactorial/polygenic inheritance is
- know how to determine the probability for an individual to be affected by a heritable disorder
- know examples of major genetic disorders discussed in class
- explain heterogeneity and genetic complementation
- know the major extensions to Mendel: incomplete dominance, codominance, multiple alleles, epistasis, pleiotropy
- explain the ABO blood group system
- explain the Lyon hypothesis and Barr body formation
- explain why it comes to variable expressivity
- know what incomplete penetrance is

Chromosomes, mitosis, meiosis, germ cell formation

be prepared to...

- know the major sex chromosome systems
- know the basic mechanism of sex determination in humans and Drosophila
- explain what dosage compensation is and how dosage compensation works in humans and Drosophila
- explain the Lyon hypothesis and Barr body formation
- know how the sex-linked white gene is inherited in Drosophila
- know the main features of X-linked recessive disorders and examples for these disorders
- explain the different phases of the cell cycle and what happens during these phases
- know chromosome morphology and terminology
- explain the major steps of mitosis, where mitosis occurs and what the result of a mitotic cell division is
• explain the major steps of meiosis, where meiosis occurs and what the result of a meiotic cell divisions is
• explain structure and composition of the spindle apparatus
• know what the synaptonemal complex and crossing-over is
• explain how the random assortment of paternal and maternal chromosomes during meiosis supports Mendel’s law of the independent assortment of alleles
• explain the major steps in oogenesis and spermatogenesis in humans
• know what lampbrush chromosomes are
• explain why the risk for Down syndrome increased with the age of the mother

Gene linkage, recombination, gene mapping

be prepared to...

• explain how linked genes are transmitted and how linkage can be inferred by determining recombination frequencies (frequencies of parental and recombinant types)
• explain the key role that test crosses play in linkage analysis
• draw a genetic map from a set of experimentally determined recombination frequencies
• know how 2-point and 3-point test crosses are conducted and how map distances are calculated from the results
• explain chromosomal interference
• describe the life cycle of the baker’s yeast, Saccharomyces cerevisiae
• know what tetrads are and how they facilitate the analysis of meiotic products
• explain how linked genes behave in tetrad analysis

DNA

be prepared to...

• explain bacterial transformation and Avery’s experiments
• describe the Hershey/Chase experiment
• describe the chemical composition of DNA
• describe the major building blocks of DNA (sugars, bases, nucleosides, nucleotides etc.)
• know the chemical differences between DNA and RNA
• know the structural differences between DNA and RNA
• explain Chargaff’s rules
• explain the Double helix model
• know what denaturation and renaturation of DNA means
• explain the difference between conservative and semi-conservative DNA replication
• describe the Meselson/Stahl experiment
• know about origins and ARSs
• know the properties of DNA polymerases that “cause trouble” during replication
• describe how replication proceeds: continuous and discontinuous synthesis; Okazaki fragments; types and roles of DNA polymerases; role of ligase
• explain problems during DNA replication at the telomeres and how they are solved
Mutations

be prepared to...

- know the major categories and types of mutations
- elaborate on the frequency of mutations
- explain the Luria/Delbrück experiment
- know about tautomeric shifts and how they give rise to spontaneous mutations
- know the major repair mechanisms: proofreading function of DNA polymerase; mismatch repair; excision repair system; AP endonucleases
- explain how spontaneous lesions of the DNA are caused
- describe the Ames test
- describe how radiation can damage DNA: X-rays, UV and thymine dimers
- know what complementation groups are and how mutations can be assigned to complementation groups
- know about nutritional mutants and the “one enzyme, one protein hypothesis”
- determine the order of enzymes within a biosynthetic pathway by analyzing nutritional mutants
- know how the building blocks of proteins look like and how the chemical properties of proteins are determined
- explain protein structure (primary, secondary, tertiary, quarternary)
- know the differences between null, hypomorphic, hypermorphic, dominant negative and neomorphic mutations

Genetic code and transcription

be prepared to...

- explain the central dogma of molecular biology
- explain the theoretical possibilities to encode 20 amino acids using a 4-letter code
- explain the phenomenon of intragenic suppression and how it was used to prove the existence of a triplet code
- know the experimental approaches that were used to crack to the genetic code
- know the start codon and the amino acid it encodes as well as the stop codons
- explain and work with the properties of the genetic code: degenerate, nonoverlapping, commaless, universal
- to explain what an open reading frame is
- give examples of exceptions from the rule that the genetic code is universal
- explain the spatial and temporal organization of RNA and protein synthesis in prokaryotes and eukaryotes
- know the different types of RNA and their functions
- describe the structure of a eukaryotic gene using the appropriate terminology
- describe the 3 phases of transcription
- explain how the primary transcript is processed in a eukaryotic nucleus
- explain how alternative splicing can be used to generate different products from a single gene
- describe the basic structure of tRNA and how tRNAs bind the appropriate amino acid
describe what the nucleolus is and how the composition and structure of ribosomes looks like
explain the difference in translation initiation between prokaryotes and eukaryotes
give an overview over the process of translation
give examples of posttranslational modifications of proteins

Recombinant DNA technology

be prepared to...

describe the steps of a typical cloning experiment
know the meaning of the terms “restriction” and “modification” in bacteria
describe how restriction enzyme recognition sites look like and which kinds of DNA ends are produced by restriction enzymes
calculate the frequency of restriction sites
explain what partial digests are good for
explain the properties and uses of the major cloning vectors plasmid, phage, and YAC
describe the basic selection procedures for transformants and transformants that carry recombinant plasmid
know how genomic and cDNA libraries are obtained and what the major differences between these two types of libraries are

Direct detection of genotype and genome analysis

be prepared to...

define the terms genome and genotype
give a picture of the size and composition of the human genome
give an overview over the major DNA polymorphisms, their frequency and their use as DNA markers
know how micro- and minisatellites arise
explain what SNPs and RFLPs are and how they can be detected
explain how DNA fingerprinting works
explain how positional cloning works
how Southern and northern blots are performed
explain what cytogenetic, linkage, physical, and sequence maps are and how they contribute to the analysis of genomes
explain what FISH is
explain the importance of sequence similarities between genomes and the different types of homologies that can be detected
know the major model organisms and to explain the advantages of model organism research

Chromatin and chromatin structure

be prepared to...

define the term chromatin and to describe its composition
know the general properties of histones and the different types of histones
give examples of nonhistone chromatin proteins
describe the structure of the nucleosome and of the 11 nm fiber
describe higher orders of chromatin structure
know the degree of condensation that is required to pack the DNA into a nucleus
describe the major differences between heterochromatin and euchromatin
distinguish between constitutive and facultative heterochromatin and to give examples of each type of heterochromatin
explain the molecular mechanism of Barr body formation
explain how polytene chromosomes are formed, where they are found, and what their basic structural features are
describe our current knowledge about yeast centromeres and the centromeres of higher eukaryotes

Chromosome mutations

be prepared to...

define the different types of structural chromosome rearrangements, explain how they are derived and how they impact viability and fertility of an organism
describe the two major types of transposable elements and the mechanism they use for transposition
explain the life cycle of a retrovirus
describe how transposable elements can be used for mutagenesis and as transformation vectors
define the terms monosomy and trisomy, to describe how they arise, and to give examples of viable human monosomies and trisomies

Gene regulation

be prepared to...

give an overview over the processes that can be targeted to control gene expression
explain the basic principles of transcriptional control using the lac operon of E. coli as an example
explain the terms negative and positive control, enzyme induction, allosteric control
explain what transcription factors do and how they look like
explain how enhancers and silencers are believed to work and what their basic properties are
explain the difference between a transcriptional activator and a repressor
explain how enhancers can direct when and where a gene is expressed
explain what coactivators and corepressor are and how they can act
explain the role that histone modifications play in transcriptional control
explain the impact that heterochromatin has on gene expression
describe how steroid hormones and nuclear receptors control gene expression

Developmental genetics

be prepared to...

explain what a master regulatory gene is
describe the function of eyeless/pax-6
explain the difference between maternal and zygotic genes
• describe how the major body axes are determined in Drosophila
• explain the mechanism of action of the dorsal protein
• describe the major classes of developmental genes that direct Drosophila embryogenesis and how they act
• describe the function of homeotic selector genes
• explain the conservation of developmental processes using the Hox gene complex as an example