



**Course:** CELL BIOLOGY WITH GENETICS

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**Department:** Medical Biology and Genetics, University of Rijeka, Medical Faculty

**Study program:** University Integrated Undergraduate and Graduate Study of Dental Medicine (in English)

**Course structure:** 90 hours: 30 lectures, 30 seminars, and 30 practicals (30L + 30S + 30 P)

**Study year:** first year

**Academic year:** 2024/25

## SYLLABUS

**Course description (a brief description of the course, general instructions, where and in what form the lessons are organized, necessary equipment, instructions for attendance and preparation for classes, student obligations, etc.):**

The focus of Cell Biology with Genetics is the study of the structure and function of the cell but also the functional interaction of the cell with its microenvironment. In this course, we will focus on Eukaryotic cell biology and will cover topics, such as membrane structure and composition, transport, and trafficking, the cytoskeleton and cell movement, the breakdown of macromolecules and the generation of energy, and the integration of cells into tissues.

We will also cover important cellular processes such as cell division, cell cycle regulation, signal transduction, apoptosis (programmed cell death), cancer cell biology, and the biology of reproduction. Furthermore, we will give insight into the fundamentals of molecular and systems biology, the flow of genetic information, human genome architecture, and the basics of medical genetics.

Throughout the semester we will attempt to relate defects in these various cellular processes to human diseases to help gain a better understanding of what happens when cells don't work properly.

Classes are centered on discussion-oriented lectures and seminars that encourage critical thinking and emphasize the significance of research as a tool for obtaining knowledge. The practical part exposes students to an overview of modern cell-related and molecular biology-related techniques and offers hands-on experience in classical cell biology experiments.

### GENERAL INSTRUCTIONAL AIMS

During the course, the students should acquire the following skills:

1. Oral and written communication,
2. The usage of information technology,
3. Evidence-based critical thinking and problem-solving
4. Individual and group work
5. Finding relevant scientific literature and acquiring scientific terminology



**SPECIFIC LEARNING OUTCOMES:** At the end of the course the student will be able to:

1. Describe the fundamental principles of cellular biology and apply them to current biological issues,
2. Understand how the cell structure relates to its functions,
3. Understand cell movement and how it is accomplished,
4. Understand how cells grow, divide, and die and how these processes are regulated,
5. Understand cell signaling and how it regulates cellular functions,
6. Understand how dysregulation of signaling processes leads to cancer and other diseases,
7. Interpret the behavior of cells in their microenvironment in multi-cellular organisms (i.e. a cell within its social context) with emphasis on cell-cell interactions, cell-extracellular matrix interactions,
8. Use the light microscope and prepare the slides,
9. Understand the main principles of Prokaryotic and Eukaryotic gene and genome organization, genome architecture, gene function, and regulation,
10. Understand the basic genetic mechanisms and the mechanisms of genome maintenance,
11. Classify chromosomal and gene mutations,
12. Understand basic and advanced cytogenetic and molecular-genetic methods in mutation detection and differential genetic diagnosis,
13. Calculate and interpret the recurrence risk for monogenic and polygenic human diseases,
14. Understand the methods and results of scientific research in the field of cell and molecular biology,
15. Integrate the knowledge of different educational units; acknowledge the interdisciplinary nature of the biomedicine field.

#### **COURSE ASSESSMENT TOOLS**

**(Important dates):**

**Midterm exam 1:** October 22, 2024; Tuesday, (28%)

**Midterm exam 2:** November 3, 2024; Friday, (28%)

Midterm exam 1 – retake: November 6, 2024

Midterm exam 2 - retake: November 21, 2024

**FINAL EXAM: November 8, 2024** – comprehensive (44%)

#### **CLASS FORMAT**

The course will be held in the autumn term between October 1<sup>st</sup> and November 8<sup>th</sup>, 2024 (5 weeks) and will consist of twenty-three-hour class sessions per week. The schedule for the lectures, seminars, and practicals along with the assigned lecturers and readings are listed in the tables titled “Course Teaching Plan” and „Course Schedule“ (below). Class session dates and times may vary with advance notice. Homework and pre-class assignments will be required for several classes, mostly seminars. Pre-class assignments will be posted on Merlin. It is advisable that students log into the course on Merlin and check for updates regularly.

All practicals will be held in the practicum of the Department of Medical Biology and Genetics (2nd Floor, east wing of the building).

#### **Assigned reading:**

1. Cooper, Geoffrey M; Hausman, Robert E. The Cell. A Molecular Approach. Sinauer Associates, Inc. Publishers Sunderland, Massachusetts U.S.A., Seventh Edition, ISBN 978-1-60535-290-9
2. Turnpenny, P; Ellard, S. Emery's ELEMENTS of MEDICAL GENETICS, Elsevier, 15th Edition, ISBN 978-0-7020-6685-6



**Optional/additional reading:**

1. Alberts B et al.: Molecular Biology of the Cell, Philadelphia, Sixth Edition, Garland Publ. Co, 2015., ISBN 978-0-8153-4464-3

**COURSE TEACHING PLAN:**

**The list of lectures (with topics and learning outcomes):**

**L1 Plan; literature**

- ✚ basic information about the course, and student obligations

**L2 Introduction to Cell Biology. Cell and Molecular Biology in Medicine**

- ✚ basics of cell and molecular biology
- ✚ hereditary basics of human diseases
- ✚ gene expression patterns in health and disease
- ✚ signaling networks
- ✚ intercellular and intracellular communication
- ✚ basics of systems biology and its application in general and dental medicine

**L3 Cell Origin and Evolution**

- ✚ to define the term evolution; distinguish between the standard and extended definitions
- ✚ to list the basic forces of evolution
- ✚ to describe the sequence of evolution (nuclear, physical, chemical, biological)
- ✚ to define the RNA world
- ✚ to explain the evolution of metabolism
- ✚ to explain the evolution of prokaryotes into eukaryotes (endosymbiotic theory)

**L4 Tools of Cell Biology**

- ✚ to define the construction and functional differences between light and electron microscopy
- ✚ to describe the types of light microscopes and their application
- ✚ to describe functional differences and application of fluorescent and confocal microscopy
- ✚ to describe the basics of tissue preparation methods for light and electron microscopy
- ✚ to describe the methods of metal shadowing for high-resolution TEM
- ✚ to describe the methods of freeze-fracture and ice-sublimation for high-resolution TEM

**L5 Compartmentalization of Eukaryotic Cells**

- ✚ to define cellular compartments, their position, morphology and main functions
- ✚ to list the advantages of the eukaryotic organization type

**L6 Structure of The Plasma Membrane**

- ✚ to describe the main structural features of biological membranes and the plasma membrane
- ✚ to explain the structure, roles, and distribution of membrane lipids, proteins, and sugars
- ✚ to explain the organization, function, and dynamics of lipid rafts
- ✚ to describe structural features of highly specialized membranes: the inner mitochondrial membrane

**L7 Membrane Transport**

- ✚ to define basic principles of passive and active transport
- ✚ to define the differences in transport proteins' function: carrier proteins and channel proteins
- ✚ to describe differences between ATP-driven and ion gradient-driven active transport



- ✚ to describe endocytosis, exocytosis, and transcytosis; differentiate between constitutive and regulated endo- and exocytosis

### L8 Bioenergetics

- ✚ to describe the production of metabolic energy in the cell (mitochondria, chloroplasts)
- ✚ to describe and differentiate between an anaerobic and aerobic respiration
- ✚ to describe the role of the inner mitochondrial membrane
- ✚ to describe the process of chemiosmotic coupling during energy production
- ✚ to define the term photosynthesis
- ✚ to differentiate between reactions in the light and reactions in the dark during photosynthesis

### L9 Cytoskeleton, the Extracellular Matrix, and Cell-Cell Interactions

- ✚ to recognize the structure, distribution, dynamics, and various roles of cytoskeletal elements in eukaryotic cells
- ✚ to understand cytoskeletal structural and functional integrity
- ✚ to describe the integrity of the extracellular matrix, plasma membrane, and cytoskeleton
- ✚ to describe the role and main components of ECM (structural proteins, polysaccharide gel, adhesion proteins)
- ✚ to describe the types, roles, and major proteins of intercellular junctions

### L10 Regulation of the Cell Cycle

- ✚ to define cell cycle phases and control points
- ✚ to define key proteins that induce cell cycle progression and their roles
- ✚ to define protein complexes: start kinase and mitosis-promoting factor (MPF)
- ✚ to explain the mechanism of activation and deactivation of regulatory complex MPF (G2 checkpoint)
- ✚ to describe the sequence of events induced by start kinase in yeast (G1), and MPF in cells of higher eukaryotes (G2)
- ✚ to explain the difference between the standard cell cycle in somatic cells and cell cycle of the embryonic cells

### L11 The Basics of Cell Signaling

- ✚ to explain bacterial quorum sensing
- ✚ to differentiate between direct signaling and signaling by secreted molecules
- ✚ to describe common signaling pathways, signal molecules, and their receptors
- ✚ to define first and second messengers

### L12 Protein Sorting and Transport in Eukaryotic cell; Endocytic and Secretion pathways. Vesicular transport. Biogenesis of the lysosomes and peroxisomes

- ✚ to define protein sorting and the role of signal sequences
- ✚ to differentiate between three basic mechanisms of protein transport in a eukaryotic cell
- ✚ to describe the basics of gated, transmembrane, and vesicular transport
- ✚ to associate protein sorting mechanisms with lysosome and peroxisome biogenesis

### L13 Basics of Human Cytogenetics and Numerical Aberrations of Human Chromosomes

- ✚ to explain what is cytogenetics
- ✚ to describe cytogenetic methods (cell culture, banding techniques, fluorescent in situ hybridization, molecular karyotypization)
- ✚ to describe somatic and dental anomalies associated with the major chromosomal syndromes (autosomal and gonosomal aneuploidies)



#### **L14 Eukaryotic Gene Organization**

- ✚ to describe eukaryotic gene organization: transcription unit and regulatory sequences (proximal-distal)
- ✚ to define types and positions of proximal regulatory sequences (promotor sequences and termination signal)
- ✚ to define types and positions of distal regulatory sequences (enhancers and silencers) and their mechanism of interaction with promoter
- ✚ to define a role for specific transcription factors (proteins activators and repressors)
- ✚ to explain features and the role of intron sequences in gene function
- ✚ to explain the transcriptional complexity of eukaryotic genes
- ✚ to explain the origin of multigene families

#### **L15 Genome Organization in Prokaryotes and Eukaryotes. The Human Genome**

- ✚ to compare the size and organization of prokaryotic and eukaryotic genomes
- ✚ to describe mitochondrial genome organization
- ✚ to explain variability in the size of Eukaryotic genomes
- ✚ to define C-value
- ✚ to explain the general human genome organization (chromosomes, karyotype)
- ✚ to list the types of DNA sequences found within the human genome
- ✚ to describe the position of highly-repetitive and moderately-repetitive DNA sequences within the human genome and their possible roles
- ✚ to explain the mechanisms of DNA transposition and retrotransposition

#### **L16 Human Genome Variation and Pharmacogenetics**

- ✚ to differentiate between qualitative (SNPs) and quantitative variants
- ✚ to describe the main types of structural genome variation
- ✚ to explain the features of single nucleotide polymorphisms (SNPs)
- ✚ to list several examples of SNPs and their phenotypic effects
- ✚ to define pharmacogenetics and describe the application of SNP analyses
- ✚ to describe the application of genetic association studies
- ✚ to describe copy number variations (CNVs) and genome research associated with this type of variation

#### **L17 The Structure and Topological Organization of Chromatin**

- ✚ to differentiate between features of euchromatin and heterochromatin
- ✚ to define a nucleosome and a chromatosome
- ✚ to describe 30nm chromatin fiber organization
- ✚ to describe the assembly of hypothetical 300nm chromatin fiber: the role of flexible chromosome scaffold (the structure made of the nuclear lamina and nonhistone nuclear proteins i.e. nuclear matrix proteins) and MARs/SARs chromosomal regions
- ✚ to explain chromatin remodeling

#### **L18 The Nuclear Envelope and Chromosome Territories; The Nuclear Bodies**

- ✚ to describe nuclear envelope, nuclear lamina and nuclear pore structure
- ✚ to define nuclear localization signals and describe basics of gated transport
- ✚ to describe structural and functional compartmentalization of interphase nucleus
- ✚ to define chromosome territories, interchromatin domains, and processes that take place there
- ✚ to describe nucleolar structure and function
- ✚ to describe ribosomal structure and biogenesis



#### **L19 Basic Genetic Mechanisms: DNA Replication**

- ✚ to list enzymes associated with DNA replication
- ✚ to list features of the DNA polymerase enzyme
- ✚ to explain basic principles (semiconservative replication), and the processes of DNA replication initiation and elongation
- ✚ to explain telomere replication

#### **L20 Molecular Basis of Gene Mutations**

- ✚ to differentiate between chromosomal and gene mutations, somatic and germinative mutations, induced or spontaneous mutations
- ✚ to describe base substitution examples, and frame-shift mutations
- ✚ to associate specific mutations with human diseases
- ✚ to describe dynamic mutations and to associate them with hereditary diseases
- ✚ to describe the examples of spontaneous mutations
- ✚ to define some physical, chemical, and biological mutagens and to describe DNA damages induced by these agents

#### **L21 Monogenic and Polygenic Human Disorders**

- ✚ to define monogenic and polygenic diseases
- ✚ to list and explain the types of inheritance in monogenic diseases
- ✚ to describe the examples of non—Mendelian inheritance
- ✚ to list the examples of multifactorial diseases
- ✚ to explain the recurrence risk for monogenic and polygenic disorders
- ✚ to describe the methods of genetic and genomic studies for candidate gene research in multifactorial diseases

#### **L22 DNA Repair**

- ✚ to list and describe the main mechanisms of DNA repair
- ✚ to list different types of DNA excision repair
- ✚ to list enzymes involved in DNA repair and explain their roles
- ✚ to differentiate between DNA repair mechanisms of single-stranded and double-stranded DNA lesions

#### **L23 Programmed Cell Death**

- ✚ to differentiate between apoptosis and necrosis
- ✚ to explain the biological relevance of apoptosis
- ✚ to describe morphologic events during apoptosis
- ✚ to list regulatory proteins of apoptosis

#### **L24 Regulation of Transcription; mRNA processing**

- ✚ to describe transcription in Prokaryotes and Eukaryotes
- ✚ to describe mRNA processing during transcription
- ✚ to describe a spliceosome
- ✚ to define a role for RNA polymerase and transcription factors

#### **L25 Posttranscriptional Control of Gene Expression**

- ✚ to describe the structure of mature mRNA (define the role for UTRs)
- ✚ to describe the main mechanisms of post-transcription control: gene silencing and mRNA editing
- ✚ to describe a role for ncRNA (miRNAs, gRNAs) in transcription regulation



**L26 mRNA translation. Posttranslational Modifications of Proteins. Protein Degradation: Ubiquitin-Proteasome Pathway.**

- ✚ to describe the structure of tRNA
- ✚ to define the ribosomal decoding center and peptidyl transferase center
- ✚ to describe translational initiation, elongation, and termination
- ✚ to describe the types of posttranslational protein modifications (adding of small chemical groups, complex molecules, or enzymatic cleavage), and roles for these modifications

**L27 The Development and Causes of Cancer**

- ✚ to define process of oncogenesis/cancerogenesis
- ✚ to describe phases in tumor development
- ✚ to differentiate between exogenic and endogenic cancerogens
- ✚ to describe the features of tumor cells

**L28 Molecular Oncogenesis: Abnormal Cell Cycle in Malignancy**

- ✚ to define genomic instability in cancer
- ✚ to define proto-oncogenes, oncogenes, and tumor-suppressor genes
- ✚ to describe the main mechanisms (mutations) that can convert a proto-oncogene into an oncogene
- ✚ to describe the role of epigenetic markers in tumor-suppressor genes

**L29 Tools of Molecular Biology I**

- ✚ to list and describe molecular genetic techniques and explain their application: recombinant DNA technology, polymerase chain reaction (PCR), restriction fragment length polymorphism (RFLP)

**L30 Tools of Molecular Biology II**

- ✚ to list and describe molecular genetic techniques and explain their application: DNA hybridization technique, DNA microchips, genome/exome sequencing

**The list of seminars with descriptions (Learning outcomes):**

**S1 Cell chemistry. Three Domains of Life. Prokaryotic and Eukaryotic Cells.**

- ✚ to explain the cell theory
- ✚ to describe the basic chemistry of macromolecules
- ✚ to describe how noncovalent chemical bonds contribute to the shape, stability, and interactions between macromolecules
- ✚ to describe the basic organization of prokaryotic and eukaryotic cells
- ✚ to differentiate between the three domains of life

**S2 Transport of Small Molecules and Macromolecules**

- ✚ to explain the basic principles of active and passive transport
- ✚ to explain the differences between simple and facilitated diffusion
- ✚ to explain the differences between active transport processes driven by ATP hydrolysis or ionic gradient
- ✚ to explain the differences between carrier proteins and channel proteins, and their roles in passive and active transport
- ✚ to explain the role of ionic pumps
- to recognize the role of membrane transport processes and to associate abnormalities in cell membrane transport with diseases
- ✚ to describe endocytosis, exocytosis, and transcytosis; to differentiate between different types of endocytosis



### **S3 The Structure and Function of Nucleic Acids**

- ✚ to define the terms: nucleotide, nucleoside, purine, pyrimidine,  $\alpha$ -helix,, 3' and 5' ends
- ✚ to describe the structure of DNA and RNA molecules (components and chemical bonds within and between polynucleotide chains)
- ✚ to explain different RNA conformations
- ✚ to define the ribonucleoprotein complex
- ✚ to explain the genetic code

### **S4 Cell Signaling in Tooth Development**

- ✚ to describe different stages of tooth development and connect them with the appropriate time period of prenatal and postnatal development
- ✚ to explain the role of signal centers, as well as specific signaling pathways and molecules, during the odontogenesis process
- ✚ to explain the role of programmed cell death (apoptosis) in the odontogenesis process
- ✚ to connect the disorders of the specific signaling pathways in odontogenesis with the occurrence of congenital developmental anomalies of teeth

### **S5 Problems: Protein and Lipid Sorting, Transport and Other Topics of Cell Biology**

- ✚ to define the basic organization and function of biological membranes
- ✚ to distinguish eukaryotic cell compartments, their morphology, and functions
- ✚ to understand vesicular transport and protein sorting
- ✚ to describe the nuclear organization

### **S6 Mechanisms of Aneuploidy and Polyploidy**

- ✚ to distinguish the types of numerical chromosomal aberrations in humans
- ✚ to learn the origin and causes of chromosomal nondisjunction
- ✚ to recognize common chromosomal disorders associated with numerical aberrations
- ✚ to learn to write a karyotype associated with numerical aberrations according to ISCN
- ✚ to distinguish methods of numerical chromosomal aberration analysis
- ✚ to associate karyotypes with specific phenotypes

### **S7 Human Chromosomal Rearrangements I**

- ✚ to distinguish the types of chromosomal rearrangements
- ✚ to explain the causes of chromosomal rearrangements
- ✚ to distinguish between balanced and unbalanced chromosomal rearrangements
- ✚ to describe the chromosome nomenclature according to ISCN
- ✚ to associate the karyotype with a specific phenotype
- ✚ to learn principles of genetic counseling for a carrier of chromosomal rearrangements

### **S8 Human Chromosomal Rearrangements II**

- ✚ to describe causes of reciprocal and Robertsonian translocations
- ✚ to associate chromosomal rearrangement with a distinct phenotype
- ✚ to differentiate between balanced and unbalanced chromosomal rearrangements
- ✚ to differentiate between bivalents, trivalent, and quadrivalent
- ✚ to explain types of chromosomal disjunction in carriers of chromosomal rearrangements, and mechanisms of unbalanced and balanced gamete formation





### **S9 Human Fertilization and Early Embryonic Development**

- ✚ to discuss the sequential nature of fertilization in which ordered changes in the gametes “drive” the process of fertilization toward completion
- ✚ to explain the role of specialized sperm and egg surface structures in fertilization
- ✚ to explain the current state of knowledge about sperm-egg membrane fusion and how sperm components are incorporated into the egg
- ✚ to describe how polyspermy is prevented and the fertilized egg is activated for development

### **S10 Problems: Mendelian and Non-Mendelian Inheritance**

- ✚ to define basic genetic terms and explain Mendel’s laws of inheritance
- ✚ to interpret the results of different types of crosses
- ✚ to differentiate monogenic and polygenic human disorders
- ✚ to differentiate polygenic and multifactorial traits
- ✚ to interpret non-Mendelian inheritance

### **S11 Genetic Abnormalities of Orofacial Structures**

- ✚ to distinguish abnormalities of the teeth number, shape, structure, and time of teeth eruption including abnormalities of other orofacial structures
- ✚ to define basic terms describing orofacial abnormalities
- ✚ to acknowledge the possible associations between genetic mutations and orofacial abnormalities
- ✚ to acknowledge variable modes of inheritance for orofacial features

### **S12 Problems: The Flow of Genetic Information - from DNA to Protein**

- ✚ to define enzymes and mechanisms of DNA replication, DNA transcription, and protein synthesis on ribosomes (translation)
- ✚ to differentiate between mechanisms of DNA transcription in bacteria and eukaryotic cells
- ✚ to define and explain the mechanisms of RNA processing (pre-mRNA splicing) and posttranslational modifications of proteins

### **S13 Epigenetics: DNA Methylation, Noncoding RNAs, Genomic Imprinting**

- ✚ to define epigenetics and epigenetic mechanisms
- ✚ to define the significance of epigenetic modifications
- ✚ to understand the mechanisms and impact of genomic imprinting
- ✚ to describe the clinical consequences of genomic imprinting

### **The list of practicals with descriptions (Learning outcomes):**

#### **P1 Basics of Light Microscopy**

- ✚ to show, name, and describe the parts of a compound microscope
- ✚ to adjust the microscope for observing objects under given magnifications
- ✚ to determine the size of an object and field of view under the light microscope
- ✚ to use a light microscope individually
- ✚ to describe basic cytological and histological methods of cell and tissue sampling for microscopic slide preparation in medicine



### **P2 Prokaryotic cell**

- ✚ to recognize basic staining techniques used in prokaryotic cell analysis
- ✚ to recognize the shape and organization of bacteria under the light microscope

### **P3 Eukaryotic cell**

- ✚ to define the morphological characteristics of eukaryotic cells visible under a light microscope
- ✚ to define differences in the morphology of plant and animal cells
- ✚ to explain the plasmolysis and deplasmolysis in plant cells

### **P4 Mitosis in Plant and Animal Cells**

- ✚ to define the cell cycle and mitosis
- ✚ to describe, find and recognize the stages of mitosis on a microscope slide of onion root
- ✚ to find and recognize the stages of mitosis on a microscope slide of a fish blastula
- ✚ to recognize and describe parts and functions of a mitotic spindle during the cell cycle
- ✚ to describe differences in cytokinesis between animal and plant cells

### **P5 Meiosis and Gametogenesis**

- ✚ to analyze the histological transverse section of the human testis
- ✚ to identify the morphology and location of male germinal epithelium, Sertoli, and Leydig cells
- ✚ to analyze the histological transverse section of the female ovary
- ✚ to distinguish different positions and sizes of the follicles
- ✚ to determine the parts of the Graafian follicle
- ✚ to identify the corpus luteum
- ✚ to associate stages of gametogenesis with appropriate chromosome number

### **P6 Human Chromosomes**

- ✚ to determine types of a G-banding metaphase spread of human chromosomes
- ✚ to define the term karyotyping; define and describe the human karyotype
- ✚ to explain the Fluorescent In Situ Hybridization (FISH) method and the application of FISH probes in the detection of numerical chromosomal aberrations

### **P7 Genomic DNA Extraction**

- ✚ to describe and explain the phases of genomic DNA isolation
- ✚ to calculate the concentration and purity of isolated DNA

### **P8 The Relationship Between Chromatin Structure and Transcriptional Activity**

- ✚ to distinguish euchromatin and heterochromatin appearance and features
- ✚ to associate the degree of chromatin decondensation/condensation with transcriptional activity/inactivity
- ✚ to explain the appearance of Barr body, polytene chromosomes, and lampbrush chromosomes
- ✚ to define endoduplication, and endomitosis
- ✚ to define somatic polyploidy

### **P9 Genotoxicity of Materials in Dental Medicine**

- ✚ to recognize the main types of dental materials
- ✚ to explain the term „biocompatibility“ and the adverse effects caused by biomaterials
- ✚ to understand the principles behind the micronucleus test and comet assay
- ✚ to explain the application of the micronucleus test and comet assay in testing the genotoxicity of dental materials



#### **P10 Patterns of Disease Inheritance**

- ✚ to recognize the symbols in genealogy and to draw family trees
- ✚ to reveal the pattern of inheritance based on the family tree (to explain the criteria for a pattern of inheritance)

#### **P11 Population Genetics**

- ✚ to describe basic characteristics of the human population (genetic structure, allele, and genotype frequencies, population genetic equilibrium through the Hardy-Weinberg principle)
- ✚ to describe the differences between qualitative and quantitative hereditary traits.
- ✚ to calculate allele and genotype frequencies using the Hardy-Weinberg equation
- ✚ to explain the Hardy-Weinberg equation and its application with examples of human diseases

#### **P12 Genetic Counselling: problems**

- ✚ to create a pedigree chart based on given information; determine the type of inheritance;
- ✚ to recognize chosen monogenic diseases according to the symptoms of the proband;
- ✚ to determine a recurrence risk of diseases when taking into account the type of inheritance;
- ✚ to explain the clinical expression of the disease and relate it to specific genes/mutations

#### **P13 Tumor Cell Biology: Molecular Oncogenesis in Clinical Practice**

- ✚ to describe the differences in morphology and intracellular structures of tumor cells compared to normal, healthy cells
- ✚ to compare the nuclear-cytoplasmic ratio in tumor cells and normal cells

#### **P14 Tools of Molecular Genetics**

- ✚ to describe and explain the polymerase chain reaction (PCR) and its application
- ✚ to describe the mechanism and application of restriction endonucleases
- ✚ to explain the principle of gel electrophoresis
- ✚ to define restriction fragment length polymorphism (RFLP) and its application
- ✚ to describe the application of recombinant DNA technology in medicine

#### **Students' obligations:**

##### *1. ATTENDANCE*

Students are advised to attend all classes to avoid missing out on the material presented in class. In return, the students can benefit from each other's contribution in class discussions. In case of absence from any class, the students are required to cover the material missed and inquire about any announcements made during their absence. **STUDENTS WHO ATTENDED LESS THAN 70% OF LECTURES, SEMINARS, OR PRACTICALS HAVE FAILED THE COURSE.**

##### *2. PARTICIPATION*

###### *Lectures:*

Students should listen to the lectures and take detailed notes. They should be prepared to participate by taking occasional quizzes and by asking questions.

###### *Seminars:*

Obligatory preparation and active participation are required (assignment readings and working tutorials will be set before seminars). Working in small groups is an important aspect of seminar activity. Students are encouraged to prepare questions and actively engage with the lecturer in order to reach a conceptual understanding of the topic.

###### *Online forum & homework:*



The online forum is an example of an individual students' out-of-class activity. Several research assignments or take-home assignments (homework) may be given during the semester at *Merlin*. This activity may bring students 2 additional credits.

*Practicals:*

Students generally do not need to prepare for practicals. They need drawing equipment (a pencil, crayons, a rubber, etc.) and a white (laboratory) coat.

3. EXAMINATION

Midterm exams and the final exam are obligatory. Midterm exams 1 and 2 are scheduled for the 3rd and 5<sup>th</sup> week.

Students arriving more than 10 minutes late for an exam will not be allowed to take the exam. In addition, under no circumstances will students be able to take an exam once other students have completed the exam and left the room.

**Assessment (exams, description of written / oral / practical exam, the scoring criteria):**

1. ASSESSMENT OF STUDENTS' WORK (EXAMS)

Students may obtain a total of 100 credits: a maximum of 56 credits during the course and a maximum of 44 credits on the final exam (**Table 1**). Students must gain a minimum of 28 credits to be allowed to take the final exam. Those students who did not gain the required 28 credits (because of illness or other relevant reasons) will be given the opportunity to obtain the required credits after classes are over, but before taking the final exam. The latest grading scale is valid. The grading scale is valid for the current academic year.

Table 1. Distribution of Credits in the Cell Biology with Genetics Course

Activity	Max. Credits
Attendance (lectures, practicals, individual and group work in seminars)	0
Activity in online forum	0
Midterm exams 1 and 2	56 (2 x 28)
Final exam (written and oral part)	44 (14 + 30)
Total	100

Throughout the course, students have two OBLIGATORY in-class midterm exams consisting of 40 multiple-choice questions each. Each correct answer is worth 0.7 credits (Table 2). Therefore, each midterm exam is worth max. 28 credits, and two midterm exams together are worth max. 56 credits. The final exam, taking place after classes are over, consists of a written and oral part. The written part has 20 multiple-choice questions. Each correct answer is worth 0.7 credits (20x0.7=14). Each midterm exam takes about 60 minutes to complete while the written part of the final exam takes about 30 minutes to complete. The distribution of credits for midterm exams is presented in Table 2.



Table 2. Midterm examination – result evaluation

Percentage (%)	No. of correct answers	Credits
90 - 100	36 - 40	25,2 – 28,0
75 - 89,9	30 - 35	21,0 – 24,5
60 – 74,9	24 – 29	16,8 – 20,3
50 – 59,9	20 - 23	14,0 – 16,1
0 – 49,9	0 - 19	0

Exams will be based on the content of the lectures, seminars, practicals and assigned reading; however, the material covered in class will be emphasized. The final exam will be comprehensive.

If students, by taking two midterm exams, gain a total of 28.0 credits, they are allowed to take the final exam,

In the written part of the final exam, which weighs max.14 credits, the students must give a correct answer to at least 50% of multiple-choice questions (i.e 10 questions) to pass. The written part of the final exam is a prerequisite for the oral part. In case the student passes the oral examination, he/she may gain min. 12 (40%) and max. 30 credits. The distribution of credits on the final exam is presented in Tables 3 and 4. Ten questions will be asked; each answer is worth from 0 to 3.0 credits.

Table 3. Final Exam (written part) – result evaluation

Percentage (%)	No. of correct answers	Credits
91 - 100	19 - 20	13,3 – 14,0
81 - 90	17 - 18	11,9 – 12,6
71 – 80	15 - 16	10,5 – 11,2
61 – 70	13 - 14	9,1 – 9,8
51– 60	11 - 12	7,7 – 8,4
<b>50</b>	<b>10</b>	<b>7,0</b>
< 50	< 10	0

Table 4. Final Exam (oral part) – result evaluation

Grade	Credits
sufficient	12,0 - 30,0
insufficient	< 12,0



**FINAL GRADING CRITERIA:**

A (5) – 90-100%, B (4) – 75-89.99%, C (3) – 60 – 74.99%, D (2) – 50 – 59.99%, F and FX (1) – <50%

The final grading is presented in Table 5.

Table 5. Final Grading

	No. of correct answers	Credits	Percentage (%)	Final Grade
1. Midterm exam	40	28,0	40	
2. Midterm exam	30	21,0	30	
Final exam – written	15	10,5	15	
Final exam - oral	-	22,0	-	
<i>Total</i>		<i>81,5 / 100</i>		
<b>Final Grade</b>				<b>B</b>

Students will have the opportunity to prepare a written essay or a PowerPoint presentation on a topic approved by the course coordinator. The topic must be related to the course content. This activity IS NOT obligatory; however, it may bring students up to 8 additional credits. Furthermore, students will have the opportunity to take some learning activities on Merlin; this additional effort might bring up to 4 additional credits.



Table 6. Summary of the Course Activities and Grading

Activity	Detailed Activity		Max. Grade Points
Attendance (lectures)	- active participation, - discussion		0
Attendance (practicals)	- slide preparation, - using a light microscope - assignment and problem-solving	- individual work to pursue students' competence	0
Attendance (seminars)	- individual and group work, problem-solving, results presentation	- obligatory preparation for the class (reading or research assignment)	0
Midterm exam 1	- objective evaluation of knowledge using multiple-choice questions	- includes content from L1-L16 S1 – S8, P1 – P6	28
Midterm exam 2	- objective evaluation of knowledge using multiple-choice questions	- includes content from L17-L30 S9 - S13, P7 - P14	28
Final exam	- written and oral examination	includes content L1 – L30, P1 - P14, S1 - S13 (comprehensive)	44
Essay writing or PowerPoint presentation	- presentation of scientific knowledge and results – NOT OBLIGATORY	- competence in reading and presenting scientific content - the ability to present scientific content briefly and clearly	8
Activities in Merlin	- lectures, dictionary	- learning through problem assignment and term description	4
<b>Total</b>			<b>100 +12</b>

**Other important information regarding to the course:**

All information regarding lectures, reading assignments and homework will be posted on the Merlin website which may be entered on <https://moodle.srce.hr/2024-2025/>.

Correspondence: For questions or concerns, please feel free to send us a message by email or by using the Merlin website and we will do our best to respond within 24 to 48 hours. Only students who are registered for the course will have access to the Merlin website protected under a password. The password will be given by the course coordinator. If you cannot access the website, inform Prof. Buretić-Tomljanović at [alenabt@uniri.hr](mailto:alenabt@uniri.hr). Students are expected to check their Merlin accounts frequently for important course updates/information.

Office visits: If you want to speak with us during office hours, please let us know by email or in class.

Academic policies: As a student enrolled in this course and at the University of Rijeka you should be familiar with the policies that govern the institution's academic processes. For example, Academic Dishonesty, Enrollment Status, and Grades and Grading. Please read the Undergraduate Academic Policies at [www.uniri.hr](http://www.uniri.hr), [www.medri.hr](http://www.medri.hr) and <http://medical-studies-in-english.com/>.

Academic dishonesty by students enrolled in undergraduate and graduate courses and programs offered by the Department of Biology and Medical Genetics will not be tolerated. Academic dishonesty includes, but is not limited to:

1. Obtaining assistance from another individual during an examination.
2. Giving assistance to another individual during an examination.



3. The unauthorized use of study material or textbooks during an examination.
4. Changing answers on a test after it has been returned and then submitting it for regrading.
5. Plagiarizing written assignments. Plagiarizing includes but is not limited to a) copying laboratory reports from previous years, b) copying or paraphrasing reports, term papers, or those prepared by other students, c) unauthorized collaboration in the preparation of reports, term papers, or theses, and d) use of another author's materials without appropriate acknowledgment through quotation and citation, e) using AI while preparing the reports or presentations without claiming it.
6. Attempting to bribe or otherwise induce an instructor to alter either a grade or examination score





## COURSE SCHEDULE (Autumn Semester; for the academic year 2024/2025)

1st week					
TUESDAY	Oct. 1, 2024.	11:15-14:00	L1, L2, L4	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library
		14:15-15:45	P1	Asst. Prof. Sanja Dević Pavlič, PhD	Dept. of Biology, practical room
WEDNESDAY	Oct. 2, 2024	11:15-14:00	L5, L6, L7	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library
		14:15-15:45	P2	Lara Saftić Martinović, PhD	Dept. of Biology, practical room
THURSDAY	Oct. 3, 2024	9:15-11:00	L3, L8	Prof. Saša Ostojić, MD, PhD	Dept. of Biology, library
		11:15-13:00	L9, L10	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library
		13:15-14:45	S1	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library
FRIDAY	Oct. 4, 2024	08:45-10:15	P3	Tea Mladenić, Assistant	Dept. of Biology, practical room
		10:45-12:30	L11, L12	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library
		12:45-14:15	S2	Prof. Jadranka Vraneković, PhD	Dept. of Biology, library
2nd week					
TUESDAY	Oct. 8, 2024	10:15-11.45	S3	Prof. Nada Starčević Čizmarević, PhD	Dept. of Biology, library
		12:15-13:00	L13	Prof. Jadranka Vraneković, PhD	Dept. of Biology, library
		13:30-15:00	P4	Prof. Nada Starčević Čizmarević, PhD	Dept. of Biology, practical room
WEDNESDAY	Oct. 9, 2024	8:15-10:00	L14, L16	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library
		10:30-12:00	P5	Asst. Prof. Sanja Dević Pavlič, PhD	Dept. of Biology, practical room
		12:15-13:45	P6	Prof. Jadranka Vraneković, PhD	Dept. of Biology, practical room
THURSDAY	Oct. 10, 2024	9:15-10:00	L15	Prof. Saša Ostojić, MD, PhD	Dept. of Biology, library
		10:15-11:00	L17	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library
		11:15-12:00	L18	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library
		12:30-14:00	S5	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library



FRIDAY	Oct. 11, 2024	09:15-10:45	S4	Asst. Prof. Sanja Dević Pavlič, PhD	Dept. of Biology, library
		11:15-12:45	P7	Prof. Nada Starčević Čizmarević, PhD	Dept. of Biology, practical room
		13:15-14:45	P8	Prof. Jadranka Vraneković, PhD	Dept. of Biology, practical room
3 <sup>rd</sup> week					
TUESDAY	Oct. 15, 2024	9:15-10:45	S6	Prof. Nada Starčević Čizmarević, PhD	Dept. of Biology, library
		11:15-12:45	S7	Prof. Jadranka Vraneković, PhD	Dept. of Biology, library
		13:15-14:45	S8	Prof. Jadranka Vraneković, PhD	Dept. of Biology, library
WEDNESDAY	Oct. 16, 2024	09:15-11:00	L19 L20	Prof. Nada Starčević Čizmarević, PhD	Dept. of Biology, library
		11:15-12:00	L24	Prof. Jadranka Vraneković, PhD	Dept. of Biology, library
		12:15-13:00	L25	Prof. Jadranka Vraneković, PhD	Dept. of Biology, library
THURSDAY	Oct. 17, 2024	9:15-10:45	S9	Asst. Prof. Sanja Dević Pavlič, PhD	Dept. of Biology, library
		11:15-12:00	L21	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library
		12:15-13:00	L22	Asst. Prof. Sanja Dević Pavlič, PhD	Dept. of Biology, library
			P9	Asst. Prof. Magda Trinajstić Zrinski, DMD, PhD	online (MS Teams)
FRIDAY	Oct. 18, 2024	9:15-10:45	S10	Asst. Prof. Sanja Dević Pavlič, PhD	Dept. of Biology, library
		11:15-12:45	P10	Asst. Prof. Sanja Dević Pavlič, PhD	Dept. of Biology, practical room
			S11-1	Prof. Alena Buretić-Tomljanović, PhD	online (Merlin)
4 <sup>th</sup> week					
TUESDAY	Oct. 22, 2024	<b>8:45-10:00</b>	<b>1<sup>st</sup> Midterm</b>	Prof. Alena Buretić-Tomljanović, PhD	online (Merlin, MS Teams)
		10:15-11:00	L23	Prof. Jadranka Vraneković, PhD	online (Merlin)
		12:30-14:00	P11	Asst. Prof. Sanja Dević Pavlič, PhD	Dept. of Biology, practical room
WEDNESDAY	Oct. 23, 2024	9:15-10:45	P12	Lara Saftić Martinović, PhD	Dept. of Biology, library
		11:15-12:00	L26	Prof. Jadranka Vraneković, PhD	Dept. of Biology, library
		12:15-13:00	S11-2	Prof. Alena Buretić-Tomljanović, PhD	Dept. of Biology, library



THURSDAY	Oct.24, 2024	9:15-10:00	L27	Prof. Saša Ostojić, MD, PhD	Dept. of Biology, library
		10:15-11:45	S12	Prof. Nada Starčević Čizmarević, PhD	Dept. of Biology, library
		12:15-13:00	L28	Prof. Alena Buretić- Tomljanović, PhD	Dept. of Biology, library
		13:15-14:45	S13	Lara Saftić- Martinović, PhD	Dept. of Biology, library
FRIDAY	Oct.25, 2024	10:15-12:00	L29 L30	Prof. Nada Starčević Čizmarević, PhD	Dept. of Biology, practical room
		12:15-13:45	P14	Prof. Nada Starčević Čizmarević, PhD	Dept. of Biology, practical room
		14:15-15:45	P13	Tea Mladenić, Assistant	Dept. of Biology, practical room
5 <sup>th</sup> week					
TUESDAY	Nov 5, 2024			<b>2nd Midterm Exam</b>	online (Merlin, MS Teams)
FRIDAY	Nov 8, 2024			<b>FINAL EXAM</b>	online/onsite

**List of lectures, seminars, and practicals:**

	LECTURES (Topic)	Teaching Hours	Lecture Room
L1	Plan; literature	1	Dept. of Biology
L2	Introduction to Cell Biology. Cell and Molecular Biology in Medicine	1	Dept. of Biology
L3	Cell Origin and Evolution	1	Dept. of Biology
L4	Tools of Cell Biology	1	Dept. of Biology
L5	The Compartmentalization of Cells	1	Dept. of Biology
L6	Structure of The Plasma Membrane	1	Dept. of Biology
L7	Membrane Transport	1	Dept. of Biology
L8	Bioenergetics	1	Dept. of Biology
L9	Cytoskeleton, the Extracellular Matrix, and Cell-Cell Interactions	1	Dept. of Biology
L10	Regulation of the Cell Cycle	1	Dept. of Biology
L11	The Basics of Cell Signaling	1	Dept. of Biology
L12	Protein Sorting and Transport in Eukaryotic cell; Endocytic and Secretion pathways. Vesicular transport. Biogenesis of the lysosomes and peroxisomes	1	Dept. of Biology
L13	Basics of Human Cytogenetics and Numerical Aberrations of Human Chromosomes	1	Dept. of Biology
L14	Eukaryotic Gene Organization	1	Dept. of Biology
L15	Genome Organization in Prokaryotes and Eukaryotes. The Human Genome	1	Dept. of Biology
L16	Human Genome Variation and Pharmacogenetics	1	Dept. of Biology
L17	The Structure and Topological Organization of Chromatin	1	Dept. of Biology



L18	The Nuclear Envelope and Chromosome Territories; The Nuclear Bodies	1	Dept. of Biology
L19	Basic Genetic Mechanisms: DNA Replication	1	Dept. of Biology
L20	Molecular Basis of Gene Mutations	1	Dept. of Biology
L21	Monogenic and Polygenic Human Disorders	1	Dept. of Biology
L22	DNA Repair	1	Dept. of Biology
L23	Programmed Cell Death	1	online
L24	Regulation of Transcription; mRNA processing	1	Dept. of Biology
L25	Posttranscriptional Control of Gene Expression		Dept. of Biology
L26	mRNA translation. Posttranslational Modifications of Proteins. Protein Degradation: Ubiquitin-Proteasome Pathway.	1	Dept. of Biology
L27	The Development and Causes of Cancer	1	Dept. of Biology
L28	Molecular Oncogenesis: Abnormal Cell Cycle in Malignancy	1	Dept. of Biology
L29	Tools of Molecular Biology I	1	Dept. of Biology
L30	Tools of Molecular Biology II	1	Dept. of Biology
<b>TOTAL TEACHING HOURS</b>		<b>30</b>	

	Seminars (Topics)	Teaching Hours	Lecture Room
S1	Cell chemistry. Three Domains of Life. Prokaryotic and Eukaryotic Cells.	2	Dept. of Biology
S2	Transport of Small Molecules and Macromolecules	2	Dept. of Biology
S3	The Structure and Function of Nucleic Acids.	2	Dept. of Biology
S4	Cell Signaling in Tooth Development	2	Dept. of Biology
S5	Problems: Protein and Lipid Sorting, Transport and Other Topics of Cell Biology	2	Dept. of Biology
S6	Mechanisms of Aneuploidy and Polyploidy	2	Dept. of Biology
S7	Human Chromosomal Rearrangements I	2	Dept. of Biology
S8	Human Chromosomal Rearrangements II	2	Dept. of Biology
S9	Human Fertilization and Early Embryonic Development	2	Dept. of Biology
S10	Problems: Mendelian and Non-Mendelian Inheritance	2	Dept. of Biology
S11	Genetic Abnormalities of Orofacial Structures	2	online + Dept. of Biology
S12	Problems: The Flow of Genetic Information - from DNA to Protein	2	Dept. of Biology
S13	Epigenetics: DNA Methylation, Noncoding RNAs, Genomic Imprinting	2	Dept. of Biology
	I. Midterm exam	2	online
	II. Midterm exam	2	online
<b>TOTAL TEACHING HOURS</b>		<b>30</b>	



	Practicals (content)	Teaching Hours	Lecture Room
P1	Basics of Light Microscopy.	2	Dept. of Biology
P2	Prokaryotic cell.	2	Dept. of Biology
P3	Eukaryotic cell.	2	Dept. of Biology
P4	Mitosis in Plant and Animal Cells.	2	Dept. of Biology
P5	Meiosis and Gametogenesis.	2	Dept. of Biology
P6	Human Chromosomes	2	Dept. of Biology
P7	Genomic DNA Extraction	2	Dept. of Biology
P8	The Relationship Between Chromatin Structure and Transcriptional Activity	2	Dept. of Biology
P9	Genotoxicity of Materials in Dental Medicine	2	<i>online</i>
P10	Patterns of Disease Inheritance	2	Dept. of Biology
P11	Population Genetics	2	Dept. of Biology
P12	Genetic Counselling: problems	2	Dept. of Biology
P13	Tumor Cell Biology: Molecular Oncogenesis in Clinical Practice	2	Dept. of Biology
P14	Tools of Molecular Genetics	2	Dept. of Biology
P15	Consultations	2	Dept. of Biology
<b>TOTAL TEACHING HOURS</b>		<b>30</b>	

	FINAL EXAM DATES
1.	November 8, 2024
2.	November 22, 2024
3.	February 7, 2025
4.	June 27, 2025
5.	September 19, 2025
1 <sup>st</sup> Midterm Retake	November 6, 2024
2 <sup>nd</sup> Midterm Retake	November 21, 2024
1 <sup>st</sup> Midterm Retake	September 17, 2025
2 <sup>nd</sup> Midterm Retake	September 18, 2025

	Lectures	Seminars	Practicals	Total
Total number	30	30	30	90
On-line	1	5	2	8
Percentage	3,3	16,7	6,7	8,9



**Sveučilište u Rijeci**  
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**Sveučilište u Rijeci • Fakultet dentalne medicine**  
**University of Rijeka • Faculty of Dental Medicine**

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