[Medicinski fakultet u Rijeci]

**Curriculum**
**2024/2025**

[Za kolegij]

# Medical genetics

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| Study program: | **Medical Studies in English (R)**<br>[Sveučilišni integrirani prijediplomski i diplomski studij] |
| Department: | **[Katedra za medicinsku biologiju i genetiku]** |
| Course coordinator: | **izv. prof. dr. sc. Pereza Nina, dr. med.** |
| Year of study: | **5** |
| ECTS: | **3** |
| Incentive ECTS: | **0 (0.00%)** |
| Foreign language: | **Possibility of teaching in a foreign language** |

## Course information:

Course Medical genetics is a mandatory course based on clinical reasoning in the fifth year of the University integrated undergraduate and graduate study of Medicine in English, and consists of 18 hours of lectures, 14 hours of seminars and 13 hourse of practicals, with a total of 45 hours (3 ECTS). The aim of the course is to describe and explain the comprehensive approach to a patient with a genetic disease or disorder or an increased risk for them, so that future physicians can apply the acquired knowledge, skills and attitudes in their own clinical practice.

**COURSE LEARNING OUTCOMES:**

Learning outcomes are determined and derived in accordance with the basic standards for the development of genetics competencies for health professionals in Europe, specifically physicians who are not specialists in medical genetics. These competencies are contained in the document Core competencies in genetics for health professionals in Europe (EuroGentest Project, https://www.eshg.org/index.php?id=139), which was accepted and approved by the Education Committee of the European society for human genetics in 2008.

*After passing the exam, the student will be able to:*

**A. COGNITIVE DOMAIN – KNOWLEDGE**

1. list and distinguish the types of genetic disorders as causes of diseases and medical conditions

2. list and compare the types and outcomes of genetic testing according to groups of indications

3. argue the advantages and limitations of genetic tests and methods used

4. calculate the recurrence risk of genetic disorders

5. distinguish categories of developmental anomalies and associate them to the corresponding causes

6. use the standardised terminology when describing dysmorphic features

7. distinguish the effects of gene variability on therapeutic outcome

**B.PSYCHOMOTORIC DOMAIN – SKILLS**

1. recognise patients with a genetic disease or disorder, as well as increased risk for them

2. choose the appropriate method of genetic testing according to indication and genetic cause of a disease or medical condition

3. basically interpret the findings of genetic testing

4. organise genetic care by referring patients to medical genetics specialists, as well as other appropriate specialists and experts

5. apply basic communication skills in the transmission of genetic information

6. search diagnostic and educational databases of genetic diseases and disorders (OMIM, GeneReviews, Orphanet, Human Phenotype Ontology, Face2Gene, PharmGKB)

**C. AFFECTIVE DOMAIN – VALUES AND ATTITUDES***

1. judge and identify the importance of making a genetic diagnosis

2. adopt the specificities of the approach to patients in medical genetics in relation to other medical specialisations

3. respect the importance of emotional, psychological, social and material consequences of genetic testing findings

4. apply the acquired knowledge and skills in providing the basic level of psychological and social support to patients and families affected by genetic disease or disorder

5. recognize the need for lifelong learning in medical genetics

**The course Medical Genetics places special emphasis on the development of the affective domain, as it is crucial that, in addition to the mentioned specific knowledge and skills, future physicians develop awareness for the multidimensionality of genetic diseases and disorders. They not only encompass the cellular level in a patient but are also reflected in all other, higher aspects of the patient's life, as well as those of their blood relatives and offspring. Genetics permeates all branches of medicine and all of humanity, from the structure and function of nuclear or mitochondrial DNA, a single cell, tissue, integrated organs, all the way to the psychological superstructure ofthe human body. As such, medical genetics is one of the most complex medical specialisations, as it requires the integration of all knowledge about the structuring and functioning of the human body and spirit.*

**COURSE CONTENT:**

With the aim of purposeful and meaningful realisation of learning outcomes, classes are organized in five large thematic units that answer specific questions related to approach to a patient with a genetic disease or disorder or increased risk for them:

**1. INDICATIONS FOR GENETIC TESTING**

(How to recognize a person with a genetic disorder?)

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|---|---|
| P1 | How to distinguish between genetic and non-genetic aetiology of diseases and medical conditions? |
| L2 | Types of genetic testing according to indications |
| L3 | The path to genetic diagnosis: care organization and diagnostic tools in medical genetics |

**2. TYPES OF GENETIC DISORDERS**

(How to choose the proper method of genetic testing and interpret the results at a basic level?)

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|---|---|
| **Disorders within genes** | |
| L4 | Spectrum of phenomena in autosomal dominant monogenic diseases |
| L5 | Autosomal recessive monogenic diseases: from patient to carrier |
| L6 | X-linked monogenic diseases and other rare types of inheritance |
| S1 | One disease – one monogenic cause |
| S2 | One disease – several possible monogenic causes: locus heterogeneity |
| S3 | One disease – several possible different causes: complex diseases |
| **Disorders of one part or entire chromosome** | |
| L7 | Deviations from the normal chromosome number |
| L8 | Balanced structural genomic rearrangements: translocations and inversions |
| L9 | Unbalanced structural genomic rearrangements: genomic disorders |
| S4 | Reproductive genetics I: fertility disorders and pregnancy complications |
| L10 | Prenatal screening and diagnostics of chromosomopathies |
| S5 | Reproductive genetics II: prenatal diagnostics |
| **Disorders of the (epi)genome** | |

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|---|---|
| L11 | Uniparental disomy and epimutations |

**3. THINKING IN PATTERNS OF CLINICAL FEATURES IN WIDE DIFFERENTIAL DIAGNOSIS**

(How to direct genetic testing in persons with multiple congenital anomalies with or without intellectual disabilities?)

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| L12 | Classifications and causes of congenital anomalies |
| L13 | Basics of clinical dysmorphology |
| S6 | Genetics of mental development: developmental delay, intellectual disabillity and autism spectrum disorders |
| P2 | How to recognize patterns of multiple congenital anomalies? |

**4. PERSONALIZED MEDICINE**

(How to approach each patient individually?)

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|---|---|
| L14 | Practical pharmacogenomics |
| L15 | Gene therapy in clinical practice |
| S7 | Interpretation of results in pharmacogenomics |
| P3 | How to organize comprehensive patient care? |

**5. COMMUNICATION SKILLS IN MEDICAL GENETICS**

(How to convey genetic information to a patient?)

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|---|---|
| L16 | Specific challenges in genet(h)ic counseling |
| L17 | Basic communication skills in medical genetics |
| P4 | Simulation of clinical reasoning in medical genetics |
| P5 | How to convey genetic information to a patient? |

*The concept of individual forms of teaching is based on the following:*

*Lectures – From clinical picture to genetic causes and diagnostic methods*

*Seminars - From recognising the indication to interpreting the findings of genetic testing*

*Practicals - Implementation of acquired competencies in one's own clinical practice*

**COURSE CONCEPT:**

Course Medical genetics is a course of clinical reasoning, the foundations of which are based on stories about patients, from whom it all begins, specifically on those examples of genetic diseases or disorders that physicians most often encounter after graduation (examples from cardiogenetics, neurogenetics, oncogenetics, pediatric genetics, gynecology, etc.). With the aim of acquiring specific knowledge, skills and values / attitudes in medical genetics, all forms of teaching (lectures, seminars and practicals) are conducted through active learning techniques, encouraging the development of open, analytical and critical thinking. All material will be presented through case analyses (case-based learning), problem-based and experiential learning in a way that reflects the authentic form of action in clinical practice, in which the physician first meets the patient, not their molecular structure. This approach to learning and teaching, reverse to the classical way, encourages simpler and more meaningful mastery and application of basic theoretical knowledge, as well as thinking about the patient in patterns of clinical features in making a working / clinical diagnosis and directing genetic testing. Therefore, lectures, seminars and exercises will

be held in an interactive environment.

## List of assigned reading:

1. Pereza N. Handbook with case reports in Medical genetics. First edition. 2021. Rijeka: University in Rijeka, Faculty of Medicine, Department of medical biology and genetics. Available at: https://repository.medri.uniri.hr/en/islandora/object/medri%3A4811

2. Materials from lectures

## List of optional reading:

1. Turnpenny P, Ellard S. Emery's elements of medical genetics. 14. ed. 2011. Philadelphia: Elsevier/Churchill Livingstone.

2. Read A, Donnai D. New Clinical Genetics. Third edition. 2015. Banbury, UK: Scion Publishing Limited.

## Curriculum:

### [Vježbe] list (with titles and explanation):

**Practical 1. How to distinguish between genetic and non-genetic aetiology of diseases and medical conditions?**

Identify individuals with indications for genetic testing (b1). Conclude and argue on the justification of the indication for genetic testing using anamnesis and clinical evaluation data (a2, b1). Draw and interpret family tree (b1).

**Practical 2. How to recognize patterns of multiple congenital anomalies?**

Recognize and describe dysmorphic features (a5, a6, b1). Search OMIM, GeneReviews, Orphanet and Face2Gene databases (b6). Apply standardised nomenclature for dysmorphic features during dysmorphologic examination (a5, a6, b1). Select the appropriate genetic testing method (a1-3, b2). Basically interpret the findings of genetic testing (a1-3, b3). Associate genotype with phenotype (b3). Apply acquired knowledge to predict reproductive outcome and recurrence risk (a1-4, b3). Assess the complexity of diagnosis in diseases with wide differential diagnosis (a3, b3, c1, c2).

**Practical 3. How to organize comprehensive patient care?**

Integrate the acquired knowledge and skills in the course into the basic interpretation of genetic testing findings (a1-5, b1-b4, b6). Organize genetic care by referring the patient to medical genetics specialists as well as other appropriate specialists and experts (c4). Assess the importance of electronic databases in lifelong learning in medical genetics (c5).

**Practical 4. Simulation of clinical reasoning in medical genetics**

Integrate the acquired knowledge, skills and attitudes in the course into conducting the integrative approach the patient with genetic disorder or disease, or an increased risk for them (a1-a7, b1-b6, c1- 5).

**Practical 5. How to convey genetic information to a patient?**

Integrate the acquired knowledge and skills in the course into a conversation with a patient (a1-a5, a7, b1-b5, c1-6). Apply basic principles of genetic counseling and communication skills in conversation with a patient (b5, c2). Help patients make informed decisions and choices about further care in an understandable, clear and sensitive way (b5, c2-4). Recognise and appreciate the emotional, social and material impact of genetic diagnosis on the patient and thier family (c4, c5).

### [Predavanja] list (with titles and explanation):

**L1 Syllabus. Active learning model based on case analysis, problem solving, and experiential learning.**

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**Lecture 2. Types of genetic testing according to indications**

List the types of genetic disorders (a1). List, compare and distinguish types of genetic testing according to groups of indications: diagnostic, predictive, carrier testing, population screening, pharmacogenomic (a2). Compare the aims and outcomes of the types of genetic testing according to the groups of indications (a2, c1). Judge and determine the importance of making a genetic diagnosis (c1). Define and distinguish between direct to consumer genetic testing and genetic testing as part of genetic counseling (a2, c2). Compare the terms genetic susceptibility and genetic determinism (a1). Associate the types of genetic testing according to indications with specific examples of diseases and conditions (a2, b1).

**Lecture 3. The path to genetic diagnosis: care organization and diagnostic tools in medical genetics (online tutorial)**

Describe and compare the organization of medical genetics in the Republic of Croatia and the world (b4, c2). Describe the role of medical genetics specialists and non-medical genetics specialists in the approach to a patient with a (possible) genetic disease / disorder (c2). Define the concept of genetic literacy and judge its importance in medicine (c5). List, distinguish and access diagnostic and educational databases of genetic diseases (OMIM, GeneReviews, Orphanet, Human Phenotype Ontology, Face2Gene, PharmGKB) (b6, c5).

**Lecture 4. Spectrum of phenomena in autosomal dominant monogenic diseases**

Repeat the basic principles / criteria of autosomal dominant inheritance (a1, a4, b1). Define and divide sequence variants as the cause of genetic diseases (a1). Explain specific genetic phenomena that affect the clinical picture of autosomal dominant diseases: penetrance continuum, variable expressivity, zygosity, anticipation, later age of onset (a1, b1). Define and distinguish between locus and allelic genetic heterogeneity (a1). Evaluate the importance of determining genotype-phenotype correlation (a4, b3). Distinguish genetic testing methods according to the type of causal sequence variant and mode of inheritance: next generation sequencing (whole genome sequencing, whole exome sequencing, clinical exome sequencing), Sanger sequencing and polymerase chain reaction variants (a3).

**Lecture 5. Autosomal recessive monogenic diseases: from patient to carrier**

Repeat the basic principles/criteria of autosomal recessive inheritance (a1, a4, b1). Explain the influence of sequence variants population grouping on genetic testing (a1, b2, b3). Distinguish the choice of genetic testing methods according to the groups of indications (a2, a3, b2). Define extended genomic screening for carriers (b2). Explain the mechanism of neoplasm development in autosomal dominant inherited forms of cancer (a1).

**Lecture 6. X-linked monogenic diseases and other rare types of inheritance**

Repeat the basic principles / criteria of X-linked inheritance (a1, a4, b1). Explain the differences in the expression of X-linked diseases between women and men (a1). List X-linked diseases lethal to males (a1, a4). Explain the influence of X chromosome inactivation on the clinical picture of X-linked diseases (a1, b3). List other rare forms of inheritance (mitochondrial diseases, Y-linked inheritance) (a1).

**Lecture 7. Deviations from the normal chromosome number**

Repeat the definitions of polyploidy and aneuploidy (a1). List and describe the clinical picture of polyploidies and the most common aneuploidies (b1). Associate polyploidies, constitutional and mosaic aneuploidies with the appropriate mechanism of occurrence and recurrence risk (a1, a4, b1). Distinguish methods of genetic testing to determine numerical chromosome disorders: GTG-method, fluorescent in-situ hybridization, quantitative fluorescent polymerase chain reaction, array comparative genomic hybridization (a3).

**Lecture 8. Balanced structural genomic rearrangements: translocations and inversions**

Define structural variations (a1). List and describe the differences between balanced structural genome rearrangements: translocations and inversions (a1). Explain the concept carriers and their possible reproductive and non-reproductive consequences (a1, a4, b1). Compare genetic testing methods for unbalanced and balanced genome rearrangements (a2, a3, b2).

**Lecture 9. Unbalanced structural genomic rearrangements: genomic disorders**

Define copy number variations (a1). List and describe the differences between the types of genomic disorders: recurrent and sporadic microdeletion / microduplication syndromes (a1). Explain the genotypephenotype correlation, i.e. different influence of the types of genomic disorders on the clinical picture and recurrence risk (a1, a4, b1). List and describe the clinical picture of the most common genomic disorders syndromes (b1). Distinguish genetic testing methods for genomic disorders (a3).

**Lecture 10. Prenatal screening and diagnostics of chromosomopathies**

List and distinguish the types of prenatal screening with regard to indications, period and objectives of implementation (a1, a2, b2). Explain the advantages and limitations of the types of prenatal screening (a3). Describe and explain invasive methods of prenatal diagnosis (b2). Judge the importance of conducting diagnostic testing in case of a positive prenatal screening test (a3, a4, b2, b3, c1).

**Lecture 11. Uniparental disomy and epimutations**

Define uniparental disomy and epimutations (a1). State the mechanism of occurrence of uniparental disomy in the context of calculating the recurrence risk (a1, a4). Describe the clinical picture of diseases resulting from uniparental disomy and epimutation (b1). List methods for genetic testing (a2, a3).

**Lecture 12. Classifications and causes of congenital anomalies**

Define and categorise congenital anomalies according to the number of affected body regions and clinical significance (a5, b1). Distinguish between structural and functional developmental anomalies (a5, a6). Distinguish between types of isolated (malformation, dysplasia, disruption and deformation) and multiple congenital anomalies (syndrome, sequence) (a5). List the groups of causes and associate them with the corresponding group of congenital anomalies (a1, a5, c5).

**Lecture 13. Basics of clinical dysmorphology**

Define the most common dysmorphic features of the head, neck, torso and extremities (a5, a6). Describe the

dysmorphic examination (a5, a6). List the dysmorphic features of the most common syndromes that occur because of genetic and chromosomal disorders (a5, b1).

**Lecture 14. Practical pharmacogenomics**

Define pharmacogenomics and explain its association with pharmacodinamics and pharmacokynetics (a7). Explain the relevance of the implementation of pharmacogenetics and pharmacogenomics to clinical practice (a2, a3, c1). List and argument the basic indications for pharmacogenomic testing according to areas of clinical medicine (b1).

**Lecture 15. Gene therapy in clinical practice**

Define gene therapy, gene editing and silencing (a7, c5). List selected examples of gene therapy in clinical practice and genetic diseases for which it is applied (a7, b1). Define criteria for rare diseases (a1). Search the Orphanet database for orphan drugs (b6). Give examples of genetic diseases for which orphan drugs are used (a7, b1).

**Lecture 16. Specific challenges in genet(h)ic counseling**

Identify and explain the specificities of genetic medicine in relation to other medical specialisations (b4, c1-5). Recognise the importance of the emotional, psychological, social, and material consequences of genetic testing findings (c2-4). Explain the ethical, legal, and social implications of genetic information (c1-5). Recognize the need for collaboration with other specialists and experts (b4, c5).

**Lecture 17. Basic communication skills in medical genetics**

State and describe the basic principles important for communication with patients in medical genetics (b5, c2, c3).

**[Seminari] list (with titles and explanation):**

**Seminar 1. One disease – one monogenic cause**

Make a working / clinical diagnosis based on the anamnesis and findings of clinical evaluation (a1, b1). Conclude and argue the type of inheritance using anamnesis and family tree (a1, b1). Determine the cause of a working / clinical diagnosis using OMIM and GeneReviews databases (b6). Select the appropriate genetic testing method (a1-3, b2). Basically interpret the findings of genetic testing (a1-3, b3). Associate genotype with phenotype (b3). Apply the acquired knowledge to determine the recurrence risk (a1-4, b3).

**Seminar 2. One disease – several possible monogenic causes: locus heterogeneity**

Distinguish the influence of genetic factors on the occurrence of monogenic, polygenic and complex diseases (a1). Basically interpret the predictive values of genetic testing (a1-3, b3). Evaluate the need for genetic testing for complex diseases (a1, a4, c1). Evaluate the importance of evidence-based medicine in everyday clinical practice (c4, c5).

**Seminar 3. One disease – several possible different causes: complex diseases**

Distinguish the influence of genetic factors on the occurrence of monogenic, polygenic and complex diseases (a1). Basically interpret the predictive values of genetic testing (a1-3, b3). Evaluate the need for genetic testing for complex diseases (a1, a4, c1). Evaluate the importance of evidence-based medicine in everyday clinical practice (c4, c5).

**Seminar 4 Reproductive genetics I: fertility disorders and pregnancy complications**

Causally associate balanced genome rearrangements and numerical chromosome changes with selected reproductive disorders (a1, b1). Select appropriate genetic testing methods for infertility, primary amenorrhea, and recurrent miscarriages (a1-3, b2). Basically interpret the findings of genetic testing (a1-3, b3). Apply acquired knowledge to predict reproductive outcome and recurrence risk (a1-4, b3). Determine the importance of making a genetic diagnosis in carriers of balanced genome rearrangements due to possible subjection to assisted reproduction or prenatal diagnostics (a4, b3, c1, c2)

**Seminar 5. Reproductive genetics II: prenatal diagnostics**

Conclude and identify indications for prenatal diagnosis: amniocentesis and chorionic villus sampling (a2, a3, b1, b2). Distinguish the aims and outcomes of genetic testing methods used in prenatal diagnosis (a3, b2). Select the appropriate genetic testing method (a1-3, b2). Basically interpret the findings of genetic testing (a1-3, b3). Apply acquired knowledge to predict reproductive outcome and recurrence risk (a1-4, b3).

**Seminar 6. Genetics of mental development: developmental delay, intellectual disabillity and autism**

**spectrum disorders**

List the genetic causes of intellectual and developmental delay and autistic spectrum disorders (a1, b1). Describe the clinical picture of intellectual and developmental delay and autism spectrum disorders and associate with appropriate causes (b1). Select the appropriate genetic testing method (a1-3, b2). Basically interpret the findings of genetic testing (a1-3, b3). Apply the acquired knowledge to calculate the recurrence risk (a1-4, b3).

**Seminar 7. Interpreting the results of pharmacogenomics**

Search the pharmGKB base (b6, c5). Direct pharmacogenomic testing (a1-3, b1, b2). Basically interpret the findings of pharmacogenomic testing (a1-3, b2, c1, c3). Adjust therapy according to interpreted pharmacogenomic findings (b4, c1).

## Student obligations:

All information regarding the course, as well as materials from the lectures will be available on the Merlin elearning platform. Students should visit the mentioned platform regularly in order to be informed in a timely manner of any facts or changes concerning the course. Furthermore, students should regularly fulfill the obligations related to course attendance and active participation in classes.

**COURSE ATTENDANCE:**

Classes are organized according to the schedule published on the Merlin e-learning platform and INP application. Attendance of lectures, seminars, practicals and midterm exams is mandatory and attendance records are kept separately for each student. All of the previously mentioned types of classes start at the exact time according to the specified schedule and being late is treated as absence from class. Entries / exits during classes are not allowed. A student may justifiably miss up to 30% of the hours provided separately for practicals, seminars and lectures, solely for health reasons, which is confirmed by a medical certificate (including absences from midterm exams). If a student is unjustifiably absent from more than 30% of class hours for each class type (5 hours of lectures, 4 hours of seminars, 4 hours of exercises), the student cannot continue to attend the course and loses the opportunity to take the final exam (0 ECTS, grade F).

**ACTIVE PARTICIPATION IN CLASSES:**

Since the course is implemented through forms of active learning, students must have and use the Handbook with case reports from Medical Genetics in all forms of classes (lectures, seminars and practicals) (either in electronic or printed version). During certain classes, students will independently use information technology, including active search and use of genetic electronic databases freely available on the Internet. Therefore, it is recommended to use smart phones in contact form, especially during seminars and practicals.

## Exam (exam taking, description of the written/oral/practical part of the exam, point distribution, grading criteria):

Student assessment is carried out according to the current regulations at the University of Rijeka, and according to the Ordinance on student grading at the Medical Faculty in Rijeka (adopted by the Faculty Assembly of the Medical Faculty in Rijeka). Assessment is carried out using ECTS grading system (% / A-F) and a numerical grading system (1-5). Student progress will be evaluated and graded during classes and at the final exam. Out of a total of 100 credits, the student can achieve a maximum of 70 credits (70%) during the course in two written midterm exams, and a maximum of 30 credits (30%) in the final, structured oral exam.

### I. DURING COURSE - MIDTERM EXAMS (TOTAL MAXIMUM OF 70 CREDITS):

During the course, the acquired knowledge from lectures, seminars and practicals will be assessed by two midterm exams in the form of a written test with multiple choice questions (Midterm exams I and II). At each midterm exam, the criterion for obtaining credits is at least 50% of correctly answered questions. Passed midterm exams are not transferable and are valid for the current academic year. Midterm exams I and II will be conducted onsite at the Faculty of Medicine in Rijeka.

Midterm exam I includes teaching units L2-P10, S1-5 and P1, has 40 questions and carries up to 40 credits. Writing time is 50 minutes. The number of correctly answered questions is converted into credits as follows:

| Number of correct answers | 0-19 | 20 | 21 | 22 | 23 | 24 | 25 | 26 | 27 | 28 | 29 | 30 | 31 | 32 | 33 | 34 | 35 | 36 | 37 | 38 | 39 | 40 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Credits | 0 | 20 | 21 | 22 | 23 | 24 | 25 | 26 | 27 | 28 | 29 | 30 | 31 | 32 | 33 | 34 | 35 | 36 | 37 | 38 | 39 | 40 |

Midterm exam II includes teaching units L11-17, S6-7 and P2-5, has 30 questions and carries up to 30 credits. Writing time is 40 minutes. The number of correctly resolved questions is converted into credits as follows:

| Number of correct answers | 0-14 | 15 | 16 | 17 | 18 | 19 | 20 | 21 | 22 | 23 | 24 | 25 | 26 | 27 | 28 | 29 | 30 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Credits | 0 | 15 | 16 | 17 | 18 | 19 | 20 | 21 | 22 | 23 | 24 | 25 | 26 | 27 | 28 | 29 | 30 |

*Midterm repetition*

Midterm exam repetitions can be accessed by students who:

• did not collect 35 credits during the classes, i.e. did not pass the midterm exam I and/or II, and/or

• were justifiably absent during classes during the midterm I and/or II exams (due to illness, with a medical certificate), and/or

• have passed the midterm exam I and/or II, but are not satisfied with the achieved credits

In case of midterm I and/or II repetition, the final credits are those that the student achieves on the repeated midterm exam. Midterm exam repetitions can be accessed only once for each midterm exam. Repetitions are held live after the completion of regular classes in two terms, in each of which only one midterm is retaken:

Midterm I repetition – in agreement with students ----- Midterm II repetition – in agreement with students

The students can apply for repetitions of midterm exams via the e-mail address of the Secretary of the Department of medical biology and genetics no later than two days before the repetition of the midterm exams.

### II. FINAL EXAM (TOTAL MAXIMUM OF 30 EVALUATION POINTS):

The final exam cannot be accessed by students who:

• finally achieve less than 35 credits after having taken the repetition of the midterm exam I and/or II, and / or

• have 30% or more of unjustified absences from classes.

Such a student is graded with an F (unsuccessful), cannot obtain ECTS credits or take the final exam, i.e. must re-enroll the course in the following academic year.

The final exam can be accessed by

• achieved ≥35 credits (50% or more of the possible 70 credits) during classes, and

• do not have more than 30% of justified absences from classes.

The final exam is a structured oral exam in the form of a patient management problem that will be held onsite at the Faculty of Medicine in Rijeka and consists of four patient cases that will examine the integration of knowledge, skills and values / attitudes (monogenic disorder, chromosomal disorder, dysmorphic syndrome, pharmacogenomics). The final exam is passed if the student achieves 15 out of 30 credits.

Success in the structured oral exam is evaluated and converted into credits in the following way :

| | Credits | |
|---|---|---|
| **Grade** | **Case 1-3** | **Case 4** |
| the answer meets the minimum criteria | 4 | 3 |
| averagely good answer | 5 | 4 |
| very good answer | 6-7 | 5 |
| excellent answer | 8 | 6 |

**III. FINAL GRADE:**

The final grade is the sum of the credits collected during the course and at the final exam. Assessment within the ECTS system is carried out according to the achieved final success in the following way:

| **Percentage of assessed credits** | **ECTS grade** | **Numerical grade** |
|---|---|---|
| 90 - 100 | A | excellent (5) |
| 75 - 89,9 | B | very good (4) |
| 60 – 74,9 | C | good (3) |
| 50 - 59,9 | D | sufficient (2) |
| 0 - 49,9 | F | insufficient (1) |

## Other notes (related to the course) important for students:

**COMMUNICATION WITH TEACHERS:**

Teachers are available daily during working hours via e - mail addresses (available on the webpage of the Faculty of Medicine in Rijeka) for all questions concerning the course. Consultations are possible by appointment and can be conducted live or through the online platform MS Teams.

**ACADEMIC INTEGRITY:**

It is expected that the teacher will respect the Code of Ethics of the University of Rijeka, and the students the Code of Ethics for students of the University of Rijeka .

## COURSE HOURS 2024/2025

Medical genetics

| **[Predavanja]**<br>(Place and time or group) | **[Vježbe]**<br>(Place and time or group) | **[Seminari]**<br>(Place and time or group) |
|---|---|---|
| **07.01.2025** | | |
| L1 Syllabus. Active learning model based on case analysis, problem solving, and experiential learning.:<br>• [P01] (09:00 - 09:45) [328]<br>  ○ MG_400<br><br>Lecture 2. Types of genetic testing according to indications:<br>• [P01] (12:30 - 13:15) [328]<br>  ○ MG_400<br><br>Lecture 3. The path to genetic diagnosis: care organization and diagnostic tools in medical genetics (online tutorial):<br>• [Katedra za medicinsku biologiju i genetiku - Praktikum] (13:15 - 14:00) [328]<br>  ○ MG_400 | Practical 1. How to distinguish between genetic and non-genetic aetiology of diseases and medical conditions?:<br>• [P01] (10:00 - 12:15) [328]<br>  ○ MG PGI<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (10:00 - 12:15) [330]<br>  ○ MG PGIII<br>• [P03 - INFORMATIČKA UČIONICA] (10:00 - 12:15) [332]<br>  ○ MG PGII | |
| izv. prof. dr. sc. Dević Pavlić Sanja, dipl. sanit. ing. [330] · dr.sc. Mladenić Tea, mag. biotech. in med. [332] · izv. prof. dr. sc. Pereza Nina, dr. med. [328] | | |
| **08.01.2025** | | |
| Lecture 4. Spectrum of phenomena in autosomal dominant monogenic diseases:<br>• [P01] (08:15 - 09:00) [328]<br>  ○ MG_400<br><br>Lecture 5. Autosomal recessive monogenic diseases: from patient to carrier:<br>• [P01] (09:15 - 10:00) [328]<br>  ○ MG_400<br><br>Lecture 6. X-linked monogenic diseases and other rare types of inheritance:<br>• [P01] (10:15 - 11:00) [330]<br>  ○ MG_400 | | Seminar 1. One disease – one monogenic cause:<br>• [P15 - VIJEĆNICA] (11:30 - 13:00) [330]<br>  ○ MG SGI<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (11:30 - 13:00) [328]<br>  ○ MG SGII |
| izv. prof. dr. sc. Dević Pavlić Sanja, dipl. sanit. ing. [330] · izv. prof. dr. sc. Pereza Nina, dr. med. [328] | | |
| **09.01.2025** | | |

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| | | Seminar 2. One disease – several possible monogenic causes: locus heterogeneity:<br>• [P08] (08:15 - 10:30) [328]<br>  ○ MG SGI<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (08:15 - 10:30) [330]<br>  ○ MG SGII<br><br>Seminar 3. One disease – several possible different causes: complex diseases:<br>• [P08] (10:45 - 11:30) [2299]<br>  ○ MG SGI<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (10:45 - 11:30) [2773]<br>  ○ MG SGII |
| izv. prof. dr. sc. Dević Pavlić Sanja, dipl. sanit. ing. [330] · izv. prof. dr. sc. Pereza Nina, dr. med. [328] · doc. dr. sc. Saftić Martinović Lara, mag. pharm. inv. [2299] · Stanković Matić Ivana, dr.med. [2773] | | |
| **10.01.2025** | | |
| Lecture 7. Deviations from the normal chromosome number:<br>• [P02] (08:15 - 09:00) [328]<br>  ○ MG_400<br><br>Lecture 8. Balanced structural genomic rearrangements: translocations and inversions:<br>• [P02] (09:15 - 10:00) [328]<br>  ○ MG_400<br><br>Lecture 9. Unbalanced structural genomic rearrangements: genomic disorders:<br>• [P02] (10:15 - 11:00) [328]<br>  ○ MG_400 | | Seminar 4 Reproductive genetics I: fertility disorders and pregnancy complications:<br>• [P07] (11:30 - 13:00) [332]<br>  ○ MG SGI<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (11:30 - 13:00) [317]<br>  ○ MG SGII |
| dr.sc. Mladenić Tea, mag. biotech. in med. [332] · izv. prof. dr. sc. Pereza Nina, dr. med. [328] · prof. dr. sc. Starčević Čizmarević Nada, dipl. ing. [317] | | |
| **13.01.2025** | | |
| Lecture 10. Prenatal screening and diagnostics of chromosomopathies:<br>• [P01] (08:15 - 09:00) [326]<br>  ○ MG_400<br><br>Lecture 11. Uniparental disomy and epimutations:<br>• [P01] (09:15 - 10:00) [330]<br>  ○ MG_400 | | Seminar 5. Reproductive genetics II: prenatal diagnostics:<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (10:15 - 11:45) [332]<br>  ○ MG SGII<br>• [P04] (10:15 - 11:45) [326]<br>  ○ MG SGI |
| izv. prof. dr. sc. Dević Pavlić Sanja, dipl. sanit. ing. [330] · dr.sc. Mladenić Tea, mag. biotech. in med. [332] · izv. prof. dr. sc. Vraneković Jadranka, mag. educ. biol. et chem. [326] | | |
| **14.01.2025** | | |

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| Lecture 12. Classifications and causes of congenital anomalies:<br>• [P01] (08:15 - 09:00) [328]<br>  ◦ MG_400<br><br>Lecture 13. Basics of clinical dysmorphology:<br>• [P01] (09:15 - 10:00) [328]<br>  ◦ MG_400 | | Seminar 6. Genetics of mental development: developmental delay, intellectual disabillity and autism spectrum disorders:<br>• [P04] (10:15 - 11:45) [317]<br>  ◦ MG SGI<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (10:15 - 11:45) [330]<br>  ◦ MG SGII |
| izv. prof. dr. sc. Dević Pavlić Sanja, dipl. sanit. ing. [330] · izv. prof. dr. sc. Pereza Nina, dr. med. [328] · prof. dr. sc. Starčević Čizmarević Nada, dipl. ing. [317] | | |
| **15.01.2025** | | |
| | Practical 2. How to recognize patterns of multiple congenital anomalies?:<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (08:15 - 09:45) [330]<br>  ◦ MG PGI<br>• [P03 - INFORMATIČKA UČIONICA] (08:15 - 09:45) [332]<br>  ◦ MG PGII<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (10:00 - 11:30) [2299]<br>  ◦ MG PGIII | |
| izv. prof. dr. sc. Dević Pavlić Sanja, dipl. sanit. ing. [330] · dr.sc. Mladenić Tea, mag. biotech. in med. [332] · doc. dr. sc. Saftić Martinović Lara, mag. pharm. inv. [2299] | | |
| **16.01.2025** | | |
| Lecture 14. Practical pharmacogenomics:<br>• [P01] (09:15 - 10:00) [328]<br>  ◦ MG_400<br><br>Lecture 17. Basic communication skills in medical genetics:<br>• [P01] (10:15 - 11:00) [2829]<br>  ◦ MG_400 | Practical 3. How to organize comprehensive patient care?:<br>• [P04] (12:45 - 14:15) [2299]<br>  ◦ MG PGII<br>• [P06] (12:45 - 14:15) [332]<br>  ◦ MG PGIII<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (13:00 - 14:30) [330]<br>  ◦ MG PGI | Seminar 7. Interpreting the results of pharmacogenomics:<br>• [P04] (11:15 - 12:45) [330]<br>  ◦ MG SGI<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (11:30 - 13:00) [332]<br>  ◦ MG SGII |
| nasl. doc. dr. sc. Babić Božović Ivana, dr. med. [2829] · izv. prof. dr. sc. Dević Pavlić Sanja, dipl. sanit. ing. [330] · dr.sc. Mladenić Tea, mag. biotech. in med. [332] · izv. prof. dr. sc. Pereza Nina, dr. med. [328] · doc. dr. sc. Saftić Martinović Lara, mag. pharm. inv. [2299] | | |
| **17.01.2025** | | |

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| Lecture 16. Specific challenges in genet(h)ic counseling:<br>• [P02] (08:15 - 09:45) [150]<br>  ◦ MG_400<br><br>Lecture 15. Gene therapy in clinical practice:<br>• [P02] (10:00 - 10:45) [330]<br>  ◦ MG_400 | Practical 4. Simulation of clinical reasoning in medical genetics:<br>• [P06] (11:00 - 12:30) [330]<br>  ◦ MG PGI<br>• [P15 - VIJEĆNICA] (11:00 - 12:30) [328]<br>  ◦ MG PGII<br>• [P07] (11:00 - 12:30) [332]<br>  ◦ MG PGIII<br><br>Practical 5. How to convey genetic information to a patient?:<br>• [P06] (12:45 - 14:15) [2773]<br>  ◦ MG PGI<br>• [P15 - VIJEĆNICA] (12:45 - 14:15) [328]<br>  ◦ MG PGII<br>• [P07] (12:45 - 14:15) [150]<br>  ◦ MG PGIII<br>• [P09 - NASTAVA NA ENGLESKOM JEZIKU] (14:15 - 15:45) [2773] [150] [328]<br>  ◦ MG PGI<br>  ◦ MG PGII<br>  ◦ MG PGIII | |
| izv. prof. dr. sc. Dević Pavlić Sanja, dipl. sanit. ing. [330] · dr.sc. Mladenić Tea, mag. biotech. in med. [332] · prof. dr. sc. Ostojić Saša, dr. med. [150] · izv. prof. dr. sc. Pereza Nina, dr. med. [328] · Stanković Matić Ivana, dr.med. [2773] | | |

### List of lectures, seminars and practicals:

| [PREDAVANJA] (TOPIC) | Number of hours | Location |
|---|---|---|
| L1 Syllabus. Active learning model based on case analysis, problem solving, and experiential learning. | 1 | [P01] |
| Lecture 2. Types of genetic testing according to indications | 1 | [P01] |
| Lecture 3. The path to genetic diagnosis: care organization and diagnostic tools in medical genetics (online tutorial) | 1 | [Katedra za medicinsku biologiju i genetiku - Praktikum] |
| Lecture 4. Spectrum of phenomena in autosomal dominant monogenic diseases | 1 | [P01] |
| Lecture 5. Autosomal recessive monogenic diseases: from patient to carrier | 1 | [P01] |
| Lecture 6. X-linked monogenic diseases and other rare types of inheritance | 1 | [P01] |
| Lecture 7. Deviations from the normal chromosome number | 1 | [P02] |
| Lecture 8. Balanced structural genomic rearrangements: translocations and inversions | 1 | [P02] |
| Lecture 9. Unbalanced structural genomic rearrangements: genomic disorders | 1 | [P02] |
| Lecture 10. Prenatal screening and diagnostics of chromosomopathies | 1 | [P01] |
| Lecture 11. Uniparental disomy and epimutations | 1 | [P01] |
| Lecture 12. Classifications and causes of congenital anomalies | 1 | [P01] |
| Lecture 13. Basics of clinical dysmorphology | 1 | [P01] |
| Lecture 14. Practical pharmacogenomics | 1 | [P01] |
| Lecture 15. Gene therapy in clinical practice | 1 | [P02] |
| Lecture 16. Specific challenges in genet(h)ic counseling | 2 | [P02] |

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| Lecture 17. Basic communication skills in medical genetics | 1 | [P01] |

| [VJEŽBE] (TOPIC) | Number of hours | Location |
|---|---|---|
| Practical 1. How to distinguish between genetic and non-genetic aetiology of diseases and medical conditions? | 3 | [P01]<br>[P03 - INFORMATIČKA UČIONICA]<br>[P09 - NASTAVA NA ENGLESKOM JEZIKU] |
| Practical 2. How to recognize patterns of multiple congenital anomalies? | 2 | [P03 - INFORMATIČKA UČIONICA]<br>[P09 - NASTAVA NA ENGLESKOM JEZIKU] |
| Practical 3. How to organize comprehensive patient care? | 2 | [P04]<br>[P06]<br>[P09 - NASTAVA NA ENGLESKOM JEZIKU] |
| Practical 4. Simulation of clinical reasoning in medical genetics | 2 | [P06]<br>[P07]<br>[P15 - VIJEĆNICA] |
| Practical 5. How to convey genetic information to a patient? | 4 | [P06]<br>[P07]<br>[P09 - NASTAVA NA ENGLESKOM JEZIKU]<br>[P15 - VIJEĆNICA] |

| [SEMINARI] (TOPIC) | Number of hours | Location |
|---|---|---|
| Seminar 1. One disease – one monogenic cause | 2 | [P09 - NASTAVA NA ENGLESKOM JEZIKU]<br>[P15 - VIJEĆNICA] |
| Seminar 2. One disease – several possible monogenic causes: locus heterogeneity | 3 | [P08]<br>[P09 - NASTAVA NA ENGLESKOM JEZIKU] |
| Seminar 3. One disease – several possible different causes: complex diseases | 1 | [P08]<br>[P09 - NASTAVA NA ENGLESKOM JEZIKU] |
| Seminar 4 Reproductive genetics I: fertility disorders and pregnancy complications | 2 | [P07]<br>[P09 - NASTAVA NA ENGLESKOM JEZIKU] |
| Seminar 5. Reproductive genetics II: prenatal diagnostics | 2 | [P04]<br>[P09 - NASTAVA NA ENGLESKOM JEZIKU] |
| Seminar 6. Genetics of mental development: developmental delay, intellectual disabillity and autism spectrum disorders | 2 | [P04]<br>[P09 - NASTAVA NA ENGLESKOM JEZIKU] |
| Seminar 7. Interpreting the results of pharmacogenomics | 2 | [P04]<br>[P09 - NASTAVA NA ENGLESKOM JEZIKU] |

**EXAM DATES (final exam):**

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| 1. | 19.05.2025. |
| 2. | 05.07.2025. |
| 3. | 02.09.2025. |
| 4. | 16.09.2025. |