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| **Naziv predmeta** | **Genetički pristup rijetkim i novim bolestima** |
| **Kod** | MFMI… | Godina studija | 2-6     . |
| **Nositelj/i predmeta** | Doc.dr.sc. Bernarda Lozić, dr.med | Bodovna vrijednost (ECTS) |      2 |
| Suradnici | Prof.dr.sc.Vjekoslav Krželj, dr.med.Doc.dr. sc.Branka Polić, dr.med.Doc.dr.sc.Sanja Lovrić Kojundžić, dr. med.Mr.sc. Maja Tomasović, dr. med.dr.sc. Maja Buljubašić | Način izvođenja nastave (broj sati u semestru) | P | S | V | T |  |
| 8 | 8 | 9 | 25 |
| Status predmeta |      Izborni | Postotak primjene e-učenja  |      0% |
| **OPIS PREDMETA** |
| Ciljevi predmeta |  |
| Uvjeti za upis predmeta i ulazne kompetencije potrebne za predmet | Položen ispit iz Medicinske genetike |
| Očekivani ishodi učenja na razini predmeta (4-10 ishoda učenja)  | 1. Razumijevanje etiologije/forme genetskog nasljeđivanja prema heredogramu2. Sažeti medicinsku dokumentaciju bolesnika s rijetkim i novim bolestima3. Izvođenje dodatne dijagnostike radi bolje procjene bolesnika s rijetkim i novim bolestima4. Objasnite i interpretirati genetske testove koji se koriste u dijagnostici rijetkih i novih bolesti. |
|  Sadržaj predmeta detaljno razrađen prema satnici nastave  | Predavanja (8 sati)1. Načini genetskog nasljeđivanja i procjena rizika ponavljanja (2h)2. Klinička evaluacija rijetkih multisistemskih i novih poremećaja (2h)3. Prikupljanje i pohrana bioloških uzoraka od bolesnika (2 sata)4. Objasnite genetički test i rizik za nasljeđivanje genetske bolesti (2h)Seminari (8 sati)1. Laboratorijske metode u genetici (2h)2. Klasična i molekularna citogenetika u kliničkoj praksi (2h)3. Laboratorijski molekularni testovi i sekvencioniranje egzoma u kliničkoj praksi (2h)4. Interpretacija nalaza genetičkih testova u nasljednim poremećajima (2h)Kliničke vježbe 9 sati:1. Klinički pregledi bolesnika s genetički dokazanim poremećajem (9 različitih slučajeva) |
| Vrste izvođenja nastave: | x predavanjax seminari i radionice x vježbe ☐ *on line* u cijelosti☐ mješovito e-učenje☐ terenska nastava | ☐ samostalni zadaci ☐ multimedija x laboratorij☐mentorski rad☐       (ostalo upisati) |
|
| Obveze studenata | Nazočnost na nastavi 80% predavanja, 90% seminari i 100% vježbe |
| Praćenje rada studenata *(upisati udio u ECTS bodovima za svaku aktivnost tako da ukupni broj ECTS bodova odgovara bodovnoj vrijednosti predmeta):* | Pohađanje nastave |       | 0,5 |  0,5    |  |       |
| Seminarski rad |       | 0,5 |  0,5     |       (Ostalo upisati) |       |
| Pismeni ispit |       | 1,0 |   1,0   |       (Ostalo upisati) |       |
|  |       |  |       |       (Ostalo upisati) |       |
|  |       |  |       |       (Ostalo upisati) |       |
| Ocjenjivanje i vrjednovanje rada studenata tijekom nastave i na završnom ispitu | Pisani ispit |
| Obvezna literatura (dostupna u knjižnici i putem ostalih medija) | - Nelson Textbook of Pediatrics, Edition 20th ed. Philadelphia: Saunders Elsevier, 2016. (X poglavlje) | **Broj primjeraka u knjižnici** | **Dostupnost putem ostalih medija** |
|  |       | *e- knjiga* |
| Dopunska literatura  | - Turnpenny P and Ellard S. Emery's Elements of Medical Genetics, 15th edition, Elsevier, 2017. (izabrana poglavlja – kopije poglavlja ce biti kopirane i podjeljene) |
| Načini praćenja kvalitete koji osiguravaju stjecanje utvrđenih ishoda učenja | -Analiza kvalitete nastave od strane studenata i nastavnika, -Analiza prolaznosti na ispitima, -Izvješća Povjerenstva za kontrolu provedbe nastave, -Izvaninstitucijska evaluacija (posjet timova za kontrolu kvalitete Nacionalne agencije za kontrolu kvalitete, uključenje u TEEP). |
| Ostalo (prema mišljenju predlagatelja) |       |

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| **NAME OF THE COURSE** | **Genetic Approaches to Rare and Novel Diseases**  |
| **Code** | MFMI… | Year of study | 2- 6 |
| Course teacher | Assoc.prof. Bernarda Lozić, MD, PhD | Credits (ECTS) | 2 |
| Associate teachers | prof. Vjekoslav Krželj, MD, PhDAssoc. Prof. Branka Polić, MD, PhDAssoc. Prof. .prof. Sanja Lovrić Kojundžić, MD, PhDMaja Tomasović, MD, MScMaja Buljubašić, PhD | Type of instruction (number of hours) | L | S | E | T |
| 8 | 8 | 9 | 25 |
| Status of the course | Elective course | Percentage of application of e-learning | 0% |
| **COURSE DESCRIPTION** |
| Course enrolment requirements and entry competences required for the course | Completion of Medical Genetics Course |
| Learning outcomes expected at the level of the course (4 to 10 learning outcomes) | * Understanding of etiology/forms of genetic inheritance according the pedigree of family
* Summarizing the patient’s medical history with rare and novel disease
* Perform additional diagnostic evaluation in patients with rare and novel

 diseases* Explain and interpret the genetic tests used in the diagnosis of rare and novel diseases
 |
|  Course content broken down in detail by weekly class schedule (syllabus) | Lectures (8 hours)1. Modes of genetic inheritance and estimate the recurrence risk (2h)
2. Clinical evaluation of multisystemic rare and novel disase (2h)
3. Collection and storage of biological samples from the affected patients (2h)
4. Explain genetic test and the risks of hereditary genetic disorder (2h)

Seminars (8 hours)1. Laboratory genetic studies (2h)2. Classical and molecular cytogenetics in clinical practice (2h)3. Laboratory molecular tests and exome sequencing in clinical practice(2h)4. Review of genetic tests in hereditary disorders (2h)Practice (9 hours)1. Problem exercise
 |
|  Format of instruction | ☒ lectures☒ seminars and workshops☒ exercises ☐ *on line* in entirety☐ partial e-learning☐ field work | ☐ independent assignments☐ multimedia x laboratory☐ work with mentor☐       (other) |
|
| Student responsibilities | In accordance to Rules of studying and Deontological code for USSM students. |
| Screening student work *(name the proportion of ECTS credits for each* *activity so that the total number of ECTS credits is equal to the ECTS value of the course)* | Class attendance | 0,5 | Research |       | Practical training |       |
| Experimental work |       | Report |       |       (Other) |       |
| Essay |       | Seminar essay | 0,5 |       (Other) |       |
| Tests |       | Oral exam |       |       (Other) |       |
| Written exam | 1,0 | Project |       |       (Other) |       |
| Grading and evaluating student work in class and at the final exam | Written exam (10 MCQ) |
| Required literature (available in the library and via other media) | **Title** | **Number of copies in the library** | **Availability via other media** |
| - Nelson Textbook of Pediatrics, Edition 20th ed. Philadelphia: Saunders Elsevier, 2016. (selected chapters of Part X- Human genetics) |       |   YES    |
| - Materials from the lectures |       |       |
|       |       |       |
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| Optional literature (at the time of submission of study programme proposal) | - Turnpenny P and Ellard S. Emery's Elements of Medical Genetics, 15th edition, Elsevier, 2017. (students will get copiesof selected chapters) |
| Quality assurance methods that ensure the acquisition of exit competences | * Teaching quality analysis by students and teachers
* Exam passing rate analysis
* Committee for control of teaching reports
* External evaluation
 |
| Other (as the proposer wishes to add) |       |