Univerza v Mariboru

Medicinska fakulteta

UČNI NAČRT PREDMETA / COURSE SYLLABUS	
Predmet:	**Molekularna biologija z genetiko**
Course title:	**Molecular Biology with Genetics**

Študijski program in stopnja Study programme and cycle	Študijska smer Study option	Letnik Year of study	Semester Semester
Dentalna medicina/Dental Medicine		**1**	**2.**
2. stopnja/2nd cycle			

Vrsta predmeta / Course type	Obvezni/ Compulsory
Univerzitetna koda predmeta / University course code:	

Predavanja Lectures	Seminar Seminar	Vaje Tutorial	Klinične vaje Clinical training	Druge oblike študija Other forms of study	Samost. delo Individual work	ECTS
30	15	15			90	5

Nosilec predmeta / Lecturer:	**Doc. dr. Špela Stangler Herodež**

Jeziki / Languages:	**Predavanja / Lectures:**	slovenščina/slovene
	Vaje / Tutorial:	slovenščina/slovene

Pogoji za vključitev v delo oz. za opravljanje študijskih obveznosti:	Prerequisits:

Vsebina:	Content (Syllabus outline):
1.Vloga molekularne biologije in genetike v medicini. 2. Struktura, morfologija in klasifikacija humanih kromosomov. 3. Osnove citogenetike, molekularna citogenetika, klinična citogenetika – sindromologija. 4. Spolni kormosomi, X inaktivacija napake spolnih kromosomov. 5. Dedovanje, Mendelejevi zakoni, dominantno, recesivno dedovanje, atipični vzorci dedovanja. 6. Struktura in lastnosti DNA molekule. 7. Podvajanje DNA: razlike med prokarionti in evkarionti. 8. DNA mutacije, popravljanje DNA napak. 9. RNA, transkripcija, translacija, vrste RNA, ribocimi. 10. Procesiranje proteinov, posttranskripcijske modifikacije proteinov. 11. Človeški genom, jederni genom, genetski kod, zgradba gena, genske družine, tandemske ponovitve, mitohondrijski genom. 12. Regulacija genske ekspresije. 13. Epigenetika, DNA metilacija. 14. Rekombinantna DNA tehnologija, genetski inženiring, biotehnologija. 15. Genetska raznolikost med posamezniki in populacijami, polimorfizmi, genetsko neravnotežje. 16. Kompleksne bolezni, multifaktorsko dedovanje. 17. Citogenetika in molekularna genetika raka.	1. The role of molecular biology and genetics in medicine. 2. Structure, morphology and classification of human chromosomes. 3. The principles of cytogenetics, molecular cytogenetics, clinical cytogenetics (syndromology). 4. Sex chromosomes, X inactivation, sex chromosomes aberrations. 5. Inheritance, Mendelien laws, dominant, recessive single-gene inheritance, atypical patterns of inheritance. 6. Structure and features of DNA molecule. 7. DNA replication: prokaryotes, eukaryotes. 8. DNA mutations, repairing mechanisms. 9. RNA, transcription, translation, ribocimes 10. Protein processing, posttranslational modifications of proteins. 11. Human genome, nuclear genome, genetic code, structure of the gene, gene families, tandem repeats, mitochondrial genome. 12. Regulation of gene expression. 13. Epigenetics, DNA methylation. 14. Recombinant DNA technology, genetic engineering, biotechnology. 15. Genetic diversity between individuals and in populations, polymorphisms, linkage disequilibrium. 16. Complex diseases, multifactorial inheritance. 17. Cytogenetics and molecular genetic of cancer.

18. Genetika imunskega sistema. 19. Razvojna genetika. 20. Izbrani primeri genetskih bolezni. 21. Genomika, proteomika, farmakogenomika, personalizirana medicina. 22. Genetsko svetovanje, prenatalna diagnostika. 23. Genetika in družba.	18. Genetics of immune system. 19. Development genetics. 20. Genetic diseases – selected cases. 21. Genomics, proteomics, farmacogenomics, personalized medicine. 22. Genetic counselling, prenatal diagnosis. 23. Genetic and society.

Temeljni literatura in viri / Readings:

1. Thompson & Thompson : Genetics in Medicine, W.B.Saunders Company., 6the ISBN 0-7216-0244-4 and 7the ed. ISBN: 9781416030805, 2007 , ISBN: 9781437706963 8th ed. 2016.
2. Robert L. Nussbaum, Roderick R. McInnes, Huntington F. Willard: Thompson and Thompson Genetics in Medicine with Access 8th. Elsevier, 2015, ISBN 9781437706963.
3. Tom Strachan, Judith Goodship & Patrick Chinnery,Genetics and genomics in medicine, Garland Sc, 978-0-8153-4480-3, 2014.
4. Stangler Herodež Š., Erjavec Škerget A., Zagorac A., Kokalj-Vokač N.: Navodila za vaje iz molekularne biologije in genetike: skripta. Maribor: Medicinska fakulteta, 2012.

Vsakoletne dopolnitve skripte so na voljo v knjižnici MF.

Dopolnilna literatura:

1. Kumar Mahato Niladri, Pal GP: Genetics in Dentistry, 2010, ISBN 9788184489415, 10.5005/jp/books/11353
2. Agnes Bloch-Zupan Heddie Sedano Crispian Scully 2012: Dento/Oro/Craniofacial Anomalies and Genetics, **ISBN:** 9780323282239, 1st Edition.
3. Andrew Read and Dian Donnai: New Clinical Genetics, 3nd Ed., 2015, ISBN 9781907904677.

Cilji in kompetence:	**Objectives and competences:**
Cilj predmeta je, da študentu nudi znanje o zgradbi, organizaciji in funkciji informacijskih makromulekul, nosilcih dednega materiala ter o zakonitostih prenosa in ohranjanja genetske informacije s poudarkom na humanem genomu. Predmet ponuja študentu pregledna in nekatera poglobljena znanja o zgradbi, organizaciji ter delovanju evkariontskega genoma s poudarkom na humanem genomu. Študenta seznani z možnimi načini analize genoma in ugotavljanja genetskih napak. Študenta seznani z vlogo genetskih faktorjev pri vzrokih humanih bolezni ter prispevku pri multifaktorjalnih boleznih, s kompleksno analizo delovanja in prenosa genetske informacije ter dedovanjem. Poudarek predavanj je na aplikativni vlogi genetike v sodobnih medicinskih tehnikah, diagnostiki in genski terapiji.	The goal of the subject is to offer the student knowledge of the structure and function of informative macromolecules. Of principal interest are the macromolecule complexes of DNA, RNA and protein and the processes of replication, transcription and translation and technologies involved in manipulating these molecules. The courses offer to students overview and some selected topics about organization, function and structure of eukaryotic genom with special attention to human genom. The student gets the knowledge of major techniques used for genom analysis, and mutation analysis. The student is introduced to the role of genetic factors in etiology of human hereditary disorders, the contribution to multifactorial diseases and understanding the complex information of functioning and transmission of genetic information. Special attention is given to diagnostic techniques, recombinant DNA technology and gene therapy.

Predvideni študijski rezultati:	**Intended learning outcomes:**
Znanje in razumevanje: 1. Zna narisati družinsko drevo. 2. Zna ločiti strukturno od numerične kromosomske spremembe. 3. Zna interpretirati rezultat molekularne kariotipizacije. 4. Zna izolirati DNA molekulo iz periferne krvi in določiti njeno koncentracijo. 5. Zna izvesti verižno reakcijo s polimerazo ter določiti produkt z gelsko elektroforezo. 6. Zna določiti gensko mutacijo z metodo alelno specifične verižne reakcije s polimerazo. 7. Zna določiti frekvenco alelov v populaciji. 8. Zna določiti fazo vezave alelov, izračunati frekvenco rekombinacije in verjetnost vezave dveh lokusov (Izračunati vrednost LOD.).	Knowledge and understanding: 1. Knowledge of drawing a family pedigree. 2. Knowledge of differences between structural and numerical chromosome aberration. 3. Knowledge of interpreting the results of molecular kariotyping. 4. Knowledge of isolation of DNA molecules from peripheral blood and measuring its concentration. 5. Knowledge of polymerase chain reaction and determination of the product on gel electroforesis. 6. Student knows how to determine the genetic mutation by the method of allele-specific polymerase chain reaction 7. Student knows how to determine the frequency of alleles in a population.

9. Zna izračunati tveganje za kompleksne bolezni.	8. Student knows how to determine the phase of binding alleles, calculate the frequency of recombination and the probability of linkage disequlibrium (Calculate the value of the LOD.). 9. Knows how to calculate the risk for complex diseases.

Metode poučevanja in učenja:	Learning and teaching methods:
Predavanja Seminarji Vaje (laboratorijske)	Lectures Seminars Tutorial (laboratory)

Načini ocenjevanja:	Delež (v %) / Weight (in %)	Assessment:
Način (pisni izpit, ustno izpraševanje, naloge, projekt)		Type (examination, oral, coursework, project):
Seminarji	10 %	Seminary work
Kolokvij iz vaj	20 %	Laboratory work examination
Pisni izpit	70 %	Written examination
(Opravljene naloge pri vajah in seminarjih so pogoj za pristop k pisnemu izpitu.)		(Laboratory and seminary work project is condition for approaching to written exam)
ŠTUDIJSKE OBVEZNOSTI ŠTUDENTOV POGOJI ZA PRISTOP K POSAMEZNEMU PREVERJANJU ZNANJA 30 ur predavanj je razdeljenih med tri predavatelje, ki samostojno pripravijo izpitna vprašanja na svoje teme. Udeležba na predavanjih je zelo priporočljiva, ker se predavanja, seminarji in vaje prepletajo in dopolnjujejo. Na izpitu se zahteva znanje pridobljeno pri vseh treh oblikah poučevanja. Izpit lahko študenti opravijo s sprotnimi kolokviji, kar je zelo priporočljivo, saj tekom predavanj utrdijo znanje, ki je potrebno za razumevanje naslednjih predavanj ter pripravo seminarjev. Sprotni kolokviji so v treh sklopih. Za priznavanje pisnega izpita, morajo biti vsi kolokviji pozitivni. Kolokviji se pišejo samo na enem roku. Študenti, ki opravijo pozitivno vse tri kolokvije, seminarske naloge in kolokvij iz vaj, se prijavijo na prvi izpitni rok v juniju, kjer se jim prizna opravljen izpit. Študenti, ki niso opravljali sprotnih kolokvijev ali katerega od njih niso opravili pozitivno, pristopijo h končnemu preverjanju znanja po opravljenem kolokviju iz vaj in oddanih seminarskih nalogah, ki je v obliki pisnega izpita. Pri pisnem izpitu je možno doseči 70 točk. Vsaj 35 točk pri pisnem delu izpita je potrebnih, da se upoštevajo še točke dosežene pri vajah in seminarjih. Minimalno skupno število doseženih točk za opravljen izpit iz predmeta je 60 od 100 možnih točk. Končna ocena predmeta je sestavljena iz : 70% - pisni izpit 20% - laboratorijske vaje 10% - seminarska naloga		REQUIREMENTS FOR ACCESS TO INDIVIDUAL KNOWLEDGE CHECKING: 30 hours of lectures are divided between three lecturers who independently prepare examination questions on their topics. Attendance at lectures is highly recommended because the lectures, seminars and tutorials intertwined and are complementary. On examination the knowledge acquired in all three forms of teaching is required. Students can pass the exam with colloquia which is highly recommended, because during the lectures consolidate knowledge is necessary for understanding the following lectures and seminar preparation. Colloquia are divided in three sets. For the recognition of written examination must all be positive. Colloquia are written only in one period of time! Students who pass the three colloquia, laboratory and seminary work can apply for the first examination period in June, where their exam is recognized as final examination. Students who are not engaged in ongoing colloquia or any of them have not been performed positively, accede to the final examination after completion of the laboratory and seminary work. At final exam is possible to achieve 70 points. At least 35 points in the written part of the exam is necessary to take into account the points achieved in tutorials and seminars. Minimum number of points scored by examinations of the course is 60 out of 100 possible points. Final evaluation of the course consists of: 70% - written exam 20% - laboratory work 10% - seminary work
VAJE 15 ur vaj je razporejenih v 5 sklopov:		LABORATORY WORK 15 hours of laboratory work is arranged in 5 sets.

– Vsi študenti si morajo priskrbeti Navodila za vaje, ki jih lahko kupijo ali si sposodijo v knjižnici. – Na vajo se je potrebno predhodno pripraviti. Z vsemi vajami študent lahko pridobi 10 točk. – Pri vsaki vaji je potrebno oddati poročilo. Vsaka uspešno opravljena vaja se točkuje z dvema točkama. Eno točko se pridobi za uspešno izvedbo vaje, drugo pa za izpolnjeno poročilo. – Ob koncu vaj je OBVEZNI kolokvij, ki se točkuje z maksimalno 10 točkami. – Skupno torej lahko študent pridobi na vajah 20 točk. – Uspešno opravljene vaje zahtevajo minimalno 12,5 točk. – Uspešno opravljene vaje so pogoj za pristop k izpitu. – Kolokvij se opravlja takoj po končanih vajah in izjemoma pred jesenskim izpitnim obdobjem (konec avgusta). – Študenti se morajo pred pričetkom vaj seznaniti z navodili za opravljanje vaj in varno delo (nevarnosti pri delu, delo s kemikalijami in infektivnim materialom), ki so napisana na koncu Navodil za vaje. SEMINARJI 15 ur seminarjev se opravlja v treh skupinah, pri štirih predavateljih. Vsak predavatelj pripravi seminarske teme, ki so objavljene v datumskem razporedu. Študenti se predhodno pripravijo na seminar tako, da preštudirajo snov po zapiskih in ustrezna navedena poglavja po priloženi literaturi. Seminarske teme se navezujejo na področje orofacialne genetike. Profesor izvede krajše uvodno predavanje, ki mu sledijo naloge in diskusija. Študenti rešujejo računske in problemske naloge v času seminarjev ali dobijo naloge na predavanjih in jih individualno ali po dva skupaj rešijo do seminarske ure, kjer se naloge predstavi in o njih diskutira. Študenti oddajo v pisni obliki poročilo, povzetek diskusije oz. rešene problemske naloge. Oceno seminarja sestavlja število pravilnih odgovorov pri nalogah, ocena poročila in/oz. ocena sodelovanja v diskusiji. Teme seminarjev so zajete tudi pri pisnem izpitu, zato je poznavanje vsebin seminarskih tem nujno za dobro opravljen izpit. Seminarska naloga pomeni 10% končne ocene pri predmetu. Prisotnost na seminarjih je obvezna in se preverja.		All students must provide instructions for exercises that can be bought or borrowed in the library of Medical faculty. It is necessary to be prepared in advance for the course. With all laboratory work the student can obtain 10 points. Each course is required to submit a report. Each successfully completed course is scored by 2 points. One point is obtained for the successful implementation of the exercise, the other for the completed report. At the end of the courses is mandatory colloquium, which counts with a maximum of 10 points. The total, therefore, a student can obtain in laboratory work is 20 points. Completion of assignments require a minimum of 12.5 points. Completion of assignments are a prerequisite for the written exam. The colloquium is carried out immediately after completion of courses and exceptionally before the autumn exam period (end of August). Students should be aware of before starting the courses with instructions for performing exercises and work safely (hazard at work, working with chemicals and infective material), which are written at the end of the Instructions for laboratory work. SEMINARY WORK 15 hours of seminars are carried out in three groups with four professors. Each lecturer will prepare seminar topics, which are published in the schedule date range. Students are conditioned to the seminar so that they study the notes of the lectures and the relevant chapters of the mentioned accompanying literature. Seminar topics are related to the field of orofacial genetics. The professor conducted a short introductory lecture, followed by tasks and discussion. Students solve computational tasks and problem during seminars or receive tasks in class and individual or two together to solve seminar problems, Students submitting in writing a report, or a summary of the discussion. problem solved tasks. Assessment of the seminar consists of the number of correct answers in the tasks, evaluation reports and / or. rating participate in the discussion. Topics seminars are also covered in the exam, so mastering the contents of this seminar is essential for good exam. Coursework represents 10% of final grade for the course. Attendance at seminars is mandatory and shall be checked.

Reference nosilca / Lecturer's references:

VOKAČ, Damijan, STANGLER HERODEŽ, Špela, KRGOVIĆ, Danijela, KOKALJ-VOKAČ, Nadja. The role of next-generation sequencing in the management of patients with suspected non-ischemic cardiomyopathy after syncope or termination of sudden arrhythmic death. *Genes*. Jan. 2024, vol. 15, issue 1, str. [1]-11, ilustr. ISSN 2073-4425. https://doi.org/10.3390/genes15010072, https://www.mdpi.com/2073-4425/15/1/72, DOI: 10.3390/genes15010072. [COBISS.SI-ID 181229059], [JCR, SNIP, WoS, Scopus]
projekt: Financer: University Medical Centre Maribor, grant number IRP-2015/01-07
projekt: P4-220 Primerjalna genomika in genomska biodiverziteta [P4-0220]; financer: ARIS

RIHAR, Nika, KRGOVIĆ, Danijela, KOKALJ-VOKAČ, Nadja, STANGLER HERODEŽ, Špela, ZORC, Minja, DOVČ, Peter. Identification of potentially pathogenic variants for autism spectrum disorders using gene-burden analysis. *PloS one*. 2023, vol. 18, iss. 5, [article no.] e0273957, str. 1-17, ilustr. ISSN 1932-6203. https://journals.plos.org/plosone/article?id=10.1371/journal.pone.0273957, Repozitorij Univerze v Ljubljani – RUL, DOI: 10.1371/journal.pone.0273957. [COBISS.SI-ID 152010499], [JCR, SNIP, WoS, Scopus do 1. 3. 2024: št. citatov (TC): 1, čistih citatov (CI): 1, čistih citatov na avtorja (CIAu): 0.17]

KRGOVIĆ, Danijela, GORENJAK, Mario, RIHAR, Nika, OPALIČ, Iva, STANGLER HERODEŽ, Špela, GREGORIČ KUMPERŠČAK, Hojka, DOVČ, Peter, KOKALJ-VOKAČ, Nadja. Impaired neurodevelopmental genes in Slovenian autistic children elucidate the comorbidity of autism with other developmental disorders. *Frontiers in molecular neuroscience*. Jun. 2022, vol. 15, str. 1-17, ilustr. ISSN 1662-5099. https://doi.org/10.3389/fnmol.2022.912671, https://www.frontiersin.org/articles/10.3389/fnmol.2022.912671/full, DOI: 10.3389/fnmol.2022.912671. [COBISS.SI-ID 112881155], [JCR, SNIP, WoS do 17. 7. 2023: št. citatov (TC): 1, čistih citatov (CI): 1, čistih citatov na avtorja (CIAu): 0.13, Scopus do 22. 7. 2023: št. citatov (TC): 1, čistih citatov (CI): 1, čistih citatov na avtorja (CIAu): 0.13]
projekt: Z3-9294 Advanced genomic analyses of Slovenian children with autistic spectrum disorders; financer: ARRS
projekt: P4-0220 Comparative genomics and genome biodiversity; financer: ARRS

STANGLER HERODEŽ, Špela, KOKALJ-VOKAČ, Nadja, ROŠKAR, Zlatko, DREISINGER, Mojca. Natančnost in hitrost poročanja o molekularnem odzivu pri KML bolnikih z in vitro diagnostičnim testom Xpert BCR-ABL = Accuracy and speed of molecular response reporting with Xpert BCR-ABL Ultra in vitro diagnostic test in CML patients. *Acta medico-biotechnica : AMB*. [Tiskana izd.]. 2021, vol. 14, [no.] 1, str. 49-55. ISSN 1855-5640. https://journals.um.si/index.php/amb/article/view/1428, https://dk.um.si/IzpisGradiva.php?id=83671, Digitalna knjižnica Slovenije - dLib.si, Digitalna knjižnica Univerze v Mariboru – DKUM, DOI: 10.18690/actabiomed.215. [COBISS.SI-ID 68177923]

STANGLER HERODEŽ, Špela, MARČUN-VARDA, Nataša, KOKALJ-VOKAČ, Nadja, KRGOVIĆ, Danijela. De novo KMT2D heterozygous frameshift deletion in a newborn with a congenital heart anomaly. *Balkan journal of medical genetics*. 2020, vol. 23, issue 1, str. 83-90, ilustr. ISSN 2199-5761. https://content.sciendo.com/view/journals/bjmg/23/1/article-p83.xml, https://doi.org/10.2478/bjmg-2020-0008, DOI: 10.2478/bjmg-2020-0008. [COBISS.SI-ID 28473347], [JCR, SNIP, WoS do 23. 1. 2024: št. citatov (TC): 8, čistih citatov (CI): 8, čistih citatov na avtorja (CIAu): 2.00, Scopus do 3. 1. 2024: št. citatov (TC): 7, čistih citatov (CI): 7, čistih citatov na avtorja (CIAu): 1.75]

JANČAR, Maja, KRGOVIĆ, Danijela, GORENJAK, Mario, STANGLER HERODEŽ, Špela, KOKALJ-VOKAČ, Nadja. The use of bioinformatic programming tool to analyse the data obtained with next-generation sequencing = Uporaba bioinformatskih orodij za analizo podatkov, dobljenih z metodo sekvenciranja nove generacije. V: DOVČ, Peter (ur.), ZORC, Minja (ur.). *Book of proceedings : 8th Colloquium on Genetics : 28th September 2020, online event*. [Ljubljana]: Genetic Society Slovenia, 2020. Str. 24-29. https://sgd.si/docs/PROCEEDINGS_SGD_2020.pdf. [COBISS.SI-ID 33197827]

STANGLER HERODEŽ, Špela. Določanje nukleotidnega zaporedja odsekov DNA v ginekološki onkologiji z naslednjo generacijo sekvenciranja = Determination of DNA nucleotide variations in gynecological oncology with next-generation sequencing. V: TAKAČ, Iztok (ur.), LUKANOVIČ, Adolf (ur.), ANZELJC, Veronika. *Pregled sodobne ginekologije in porodništva pred epidemijo covida-19 leta 2020 = A state-of-the-art 2020 review on obstetrics and gynecology in the pre-Covid-19 era*. 1. izd. Maribor: Univerza v Mariboru, Univerzitetna založba: = University of Maribor, University Press, 2021. Str. 663-669, ilustr. ISBN 978-961-286-532-0. [COBISS.SI-ID 102192899]

STANGLER HERODEŽ, Špela. Molekularnogenetske preiskave v ginekološki onkologiji. V: TAKAČ, Iztok (ur.), ARKO, Darja. *Ginekološka onkologija*. 1. izd. Maribor: Univerzitetna založba Univerze, 2020. Str. 271-278. ISBN 978-961-286-330-2. [COBISS.SI-ID 512976184]