



UČNI NAČRT PREDMETA / COURSE SYLLABUS

Predmet:	Molekularna biologija z genetiko
Course title:	Molecular biology with genetics

Študijski program in stopnja Study programme and level	Študijska smer Study field	Letnik Academic year	Semester Semester
Splošna medicina General medicine		1	2

Vrsta predmeta / Course type

Univerzitetna koda predmeta / University course code:

Predavanja Lectures	Seminar Seminar	Sem. vaje Tutorial	Lab. vaje Laboratory work	Teren. vaje Field work	Samost. delo Individ. work	ECTS
30	15		15		90	5

Nosilec predmeta / Lecturer:

Red. prof. dr. Nadja KOKALJ VOKAČ

Jeziki /

Languages:

Predavanja / Lectures: Slovenščina/Slovene

Vaje / Tutorial: Slovenščina/Slovene

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

Prerequisites:

Vsebina:

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 kromosomi, 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 prokarioti in evkarioti.
8. DNA mutacije, popravljanje DNA napak.
9. RNA, transkripcija, translacija, vrste RNA, ribocimi.
10. Procesiranje proteinov, posttranskripcijske modifikacije proteinov.
11. Človeški genom, jedrni 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

Content (Syllabus outline):

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, Mendelian 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, ribosomes.
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

populacijami, polimorfizmi, genetsko neravnostezje.

16. Kompleksne bolezni, multifaktorsko dedvanje.

17. Citogenetika in molekularna genetika raka.

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.

populations, polymorphisms, linkage disequilibrium.

16. Complex diseases, multifactorial inheritance.

17. Cytogenetics and molecular genetic of cancer.

18. Genetics of immune system.

19. Development genetics.

20. Genetic diseases – selected cases.

21. Genomics, proteomics, farmacogenomics, personalized medicine.

22. Genetic counseling, 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. Alberts, Bray, Lewis, Raff, Roberts, Watson: Molecular Biology of the Cell, Garland Publishing, Inc, New York & London. 6th edition ISBN 9780815344322, 2014.
3. Tom Strachan, Judith Goodship & Patrick Chinnery, Genetics and genomics in medicine, Garland Sc, 978-0-8153-4480-3, 2014.
4. Lodish: Molecular Cell Biology, W.H. Freeman and Company, 6th ed., ISBN-10: 9780716776017 ISBN-13: 9780716776017, 2007.
5. Andrew Read and Dian Donnai: New Clinical Genetics, 3rd Ed., 2015, ISBN 9781907904677.

Dodatna literatura:

6. Mark F Sanders, John L Bowman, Genetic Analysis an integrated Approach, 2nd edition, ISBN-13: 978-0321948908 , 2014.
7. Edward S. Tobias, Michael Connor, Malcom Ferguson-Smith, Essential medical Genetics, 6th Edition, Wiley-Blackwell, ISBN 978-1-4051-6974-5, 2011.
8. Basiswissen Humangenetik: Schaaf, Zschocke, Springer Verlag, ISBN 978-3-642-28907-1, 2013.
9. Molekulare Genetik, Knippers et al., Georg Thieme Verlag, ISBN: 9783134770100, 2015.
10. Biologie für Mediziner, Hirsch-Kauffmann, Schweiger, Georg Thieme Verlag, ISBN 3-13-706502-X.
11. Christian P. Schaaf, Johannes Zschocke, Lorrain Potocki, Human Genetics: From Molecules to medicine., Wolters Kluwer/ Lippincott Williams & Wilkins, ISBN 978-1-60831-671-7, 2011.

Cilji in kompetence:

Cilj predmeta je, da študentu nudi znanje o zgradbi, organizaciji in funkciji informacijskih makromolekul, nosilcih dednega materiala ter o zakonitostih prenosa in ohranjanja genetske informacije s poudarkom na humanem genomu.

Premet ponuja študentu pregledna in nekatera poglobljena znanja o zgradbi, organizaciji ter delovanju eukariotskega 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 multifaktorskih boleznih, s kompleksno analizo delovanja in prenosa genetske informacije ter dedovanjem. Povdarek predavanj je na aplikativni vlogi genetike v sodobnih medicinskih tehnikah, diagnostiki in genski terapiji.

Objectives and competences:

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 genome with special attention to human genome. The student gets the knowledge of major techniques used for genome 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.

2018/2019

Predvideni študijski rezultati:

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 alelna 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.).
9. Zna izračunati tveganje za kompleksne bolezni.

Intended learning outcomes:

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 electrophoresis.
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.
8. Student knows how to determine the phase of binding alleles, calculate the frequency of recombination and the probability of linkage disequilibrium (Calculate the value of the LOD.).
9. Knows how to calculate the risk for complex diseases.

Metode poučevanja in učenja:

Predavanja : 50%
Laboratorijske vaje: 25%
Seminarji: 25%

Learning and teaching methods:

Lectures : 50%
Laboratory work : 25%
Seminary work: 25%

Načini ocenjevanja:

Delež (v %) /
Weight (in %)

Assessment:

Način (pisni izpit, ustno izpraševanje, naloge, projekt)
Seminarji
Kolokvij iz vaj
Pisni izpit.
(Opravljene naloge pri vajah in seminarjih so pogoj za pristop k pisnemu izpitu.)

ŠTUDIJSKE OBVEZNOSTI ŠTUDENTOV
POGOJI ZA PRISTOP K POSAMEZNEMU PREVERJANJU ZNANJA
30 ur predavanj je razdeljenih med štiri 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

10 %
20 %
70 %

Type (examination, oral, coursework, project):
Seminary work
Laboratory work examination
Written examination
(Laboratory and seminary work project is condition for approaching to written exam.)

REQUIREMENTS FOR ACCESS TO INDIVIDUAL KNOWLEDGE CHECKING:
30 hours of lectures are divided between four 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

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 seminarских 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 VAJE 15 ur vaj je razporejenih v 5 sklopov: •Vsi študenti si morajo preskrbeti Navodila za vaje, ki jih lahko kupijo ali si sposodijo v knjižnicah. •Na vajo se je potrebno predhodno pripraviti, Z vsemi vajami študent lahko pridobi 10 točk. •Pri vsaki vaji je potrebno oddati poročilo. Vsaka uspešno (Ne samo opravljena!) 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 napisane 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.	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 LABORATORY WORK 15 hours of laboratory work is arranged in 5 sets. 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
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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 pravih odgovorov pri nalogah, ocena poročila in/oz. ocena sodelovanja v diskusiji. Teme seminarjev so zajete tudi pri pisnem izpitu, zato je poznavanje vsebin seminarjskih tem nujno za dobro opravljen izpit. Seminarjska naloga pomeni 10% končne ocene pri predmetu. Prisotnost na seminarjih je obvezna in se preverja.		they study the notes of the lectures and the relevant chapters of the mentioned accompanying literature. 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.
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Reference nosilca / Lecturer's references:

1. GREGORIČ KUMPERŠČAK, Hojka, KRGOVIĆ, Danijela, **KOKALJ-VOKAČ, Nadja**. Specific behavioural phenotype and secondary cognitive decline as a result of an 8.6 Mb deletion of 2q32.2q33.1. *JIMR on-line*, ISSN 1473-2300, 2016, vol. , no. , str. 1-8, ilustr. <http://imr.sagepub.com/content/early/2016/01/22/0300060515595651.full.pdf+html>, doi: [10.1177/0300060515595651](https://doi.org/10.1177/0300060515595651). [COBISS.SI-ID [5609791](https://www.cobiss.si/id/5609791)], [JCR, SNIP, WoS do 25. 4. 2016: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0, Scopus do 11. 4. 2016: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0] IF = 2,14
2. MUJEZINOVIĆ, Faris, KRGOVIĆ, Danijela, BLATNIK, Ana, ZAGRADIŠNIK, Boris, VIPOTNIK-VESNAVER, Tina, ČAKŠ GOLEC, Tina, TUL, Nataša, **KOKALJ-VOKAČ, Nadja**. Simpson-Golabi-Behmel syndrome : a prenatal diagnosis in a foetus with GPC3 and GPC4 gene microduplications. *Clinical genetics*, ISSN 0009-9163, 2016, vol. , no. , str. [1-3], ilustr. <http://onlinelibrary.wiley.com/doi/10.1111/cge.12725/epdf>, doi: [10.1111/cge.12725](https://doi.org/10.1111/cge.12725). [COBISS.SI-ID [5610815](https://www.cobiss.si/id/5610815)], [JCR, SNIP, Scopus do 14. 3. 2016: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0] IF = 3,93
3. KRGOVIĆ, Danijela, BLATNIK, Ana, BURMAS, Ante, ZAGORAC, Andreja, **KOKALJ-VOKAČ, Nadja**. A coalescence of two syndromes in a girl with terminal deletion and inverted duplication of chromosome 5. *BMC medical genetics*, ISSN 1471-2350, 2014, [Vol.] 15, str. 1-9, ilustr. <http://www.biomedcentral.com/1471-2350/15/21>, doi: [10.1186/1471-2350-15-21](https://doi.org/10.1186/1471-2350-15-21). [COBISS.SI-ID [4929087](https://www.cobiss.si/id/4929087)], IF = 2,536.
4. **KOKALJ-VOKAČ, Nadja**, ČIZMAREVIČ, Bogdan, ZAGORAC, Andreja, ZAGRADIŠNIK, Boris, LANIŠNIK, Boštjan. An evaluation of SOX2 and hTERC gene amplifications as screening markers in oral and oropharyngeal squamous cell carcinomas. *Molecular cytogenetics*, ISSN 1755-8166. [Online ed.], Jan. 2014, vol. 7, str. [1]-8, ISSN 1755-8166. [Online ed.], Jan. 2014, vol. 7, str. [1]-8, ilustr. <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3900261/pdf/1755-8166-7-5.pdf>, doi: [10.1186/1755-8166-7-5](https://doi.org/10.1186/1755-8166-7-5). [COBISS.SI-ID [4897599](https://www.cobiss.si/id/4897599)], IF = 2,36
5. MACEDONI-LUKŠIČ, Marta, KRGOVIĆ, Danijela, ZAGRADIŠNIK, Boris, **KOKALJ-VOKAČ, Nadja**. Deletion of the last exon of SHANK3 gene produces the full Phelan-McDermid phenotype : a case report. *Gene*, ISSN 0378-1119. [Print ed.], 2013, vol. 524, no. 2, str. 386-389, ilustr. <http://www.sciencedirect.com/science/article/pii/S0378111913004642>, doi: [10.1016/j.gene.2013.03.141](https://doi.org/10.1016/j.gene.2013.03.141). [COBISS.SI-ID [721836](https://www.cobiss.si/id/721836)], IF = 2,196
6. KRGOVIĆ, Danijela, MARČUN-VARDA, Nataša, ZAGORAC, Andreja, **KOKALJ-VOKAČ, Nadja**. Submicroscopic interstitial deletion of chromosome 11q22.3 in a girl with mild mental retardation and facial dysmorphism: Case report. *Molecular cytogenetics*, ISSN 1755-8166. [Online ed.], 2011, [Vol.] 4, 17. <http://www.molecularcytogenetics.org/content/pdf/1755-8166-4-17.pdf>, doi: [doi:10.1186/1755-8166-4-17](https://doi.org/10.1186/1755-8166-4-17).

7. ŽERJAVIČ, Katja, ZAGRADIŠNIK, Boris, LOKAR, Lidija, GLASER, Marjana, **KOKALJ-VOKAČ, Nadja**. The association of the JAK2 46/1 haplotype with non-splanchnic venous thrombosis. Thrombosis research, ISSN 0049-3848. [Print ed.], 2013, vol. , no. , str. <http://www.sciencedirect.com/science/article/pii/S0049384813002727>, doi: 10.1016/j.thromres.2013.06.021. [COBISS.SI-ID 4724799], IF = 3,133.