UČNI NAČRT PREDMETA / COURSE SYLLABUS	
Ime predmeta:	Izbirne vsebine in novosti v genetiki in genomiki v medicini
Course title:	Selected topics and novelties in genetics and genomics in medicine

Študijski program in stopnja **Study programme and cycle**	**Študijska smer** **Study option**	**Letnik** **Year of study**	**Semester** **Semester**
Splošna medicina, enovit magistrski študijski program		Prvi	2.
General medicine, Uniform master's degree study program		First	2nd

Vrsta predmeta (obvezni ali izbirni) / **Course type (compulsory or elective)**	izbirni elective
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**
		AV	**LV**	**RV**				
5	40						45	3

Nosilec predmeta / Course coordinator:	**Prof. dr. Uroš Potočnik**
Jeziki /Languages: **Predavanja / Lectures:**	slovenski/slovene
Vaje / Tutorial:	slovenski/slovene

Pogoji za vključitev v delo oz. za opravljanje študijskih obveznosti:	**Prerequisites for enrolling in the course or for performing study obligations:**

Vsebina (kratek pregled učnega načrta):	**Content (syllabus outline):**
• genetika in genomika • genomika in odkrivanje novih zdravil • genomika raka • asociacijske študija za odkrivanje novih genov povezanih s kompleksnimi boleznimi (avtoimunske bolezni, rak, astma, sladkorna…) • tehnologije v genomiki: nanotehnologija, mikromreže (biočipi), tehnologija za avtomatsko gensko tipizacije enonukleotidnih polimorfizmov v celotnem genomu (SNPov), sekvenciranje celotnih genomov, Maldi-TOF, 2-D elektroforeza • funkcijska genomika • transkriptomika • farmakogenomika in toksikologija • fiziološka genomika	• genetics and genomics • genomics in drug discovery • cancer genomics • disease association study in common complex diseases (autoimmune diseases, cancer, asthma, diabetes…) • genomic technologies: nanotechnology, microarrays (Biochips), whole genome genotyping of Single nucleotide Polymorphisms (SNPs), Maldi-TOF, 2-D electrophoresis • functional genomics • transcriptomics • pharmacogenomics and toxicology • physiological genomics • comparative genomics • genomics of microorganisms

• primerjalna (komparativna) genomika • mikrobna genomika • proteomika • bioinformatika: podatkovne zbirke v genetiki in genomiki, programska orodja (za urejanje, analizo in poravnavo nukleotidnih zaporedij, za risanje in segregacijsko analizo družinskih dreves, za statistično genetiko) • projekt humani genom in projekt HapMap • genetske razlike med posamezniki • populacijska genetika: velikost in struktura populacije, naravni izbor, mutacije, genetski zdrs, genski pretok, parjenje v sorodstvu; molekularna evolucija, molekularna ura, nastanek genomov • genetski testi v diagnostiki bolezni • genska terapija • etika v genomiki	• proteomics • bioinformatics: human genome databases, bioinformatics tools (genome sequence analysis, design and segregation analysis of family tree data) • statistical genetics and disease association analysis • Human genome and HapMap projects • Genetic diversity among individuals • Population genetics: size and structure of population, natural selection, mutations, genetic drift, gene flow, inbreeding, molecular evolution, molecular clocks, evolution of genomes • Genetics in diagnosis • Gene therapy • Ethics in genomics

Temeljni literatura in viri / Reading materials:

1. Tom Strachan, Judith Goodship & Patrick Chinnery: Genetics and genomics in medicine, Garland Sc, ISBN 978-0-8153-4480-3, 2015
2. Nussbaum, Robert L., 1950-; McInnes, Roderick R.; Willard, Huntington F.: Thompson & Thompson Genetics in Medicine, 8the ed., Philadelphia : Elsevier., ISBN: 978-1-4377-0696-3, 2016.

Dopolnilna literatura in viri/ Additional textbooks:

3. Lesk, Arthur: Introduction to genomics, 3rd ed. - New York : Oxford University Press, ISBN 978-0-19-875483-1; 0-19-875483-3; cop. 2017
4. Maitland-van der Zee, Anke-Hilse, Daly, Anne: Pharmacogenetics and individualized therapy; New Jersey : John Wiley & Sons, ISBN 978-0-470-43354-6, 2012

Cilji in kompetence:	**Objectives and competences:**
Študenti bodo poglobili razumevanje načinov dedovanja, strukture in primerjave genov in genomov, genetske raznolikosti in genetskih napak povezanih z nastankom bolezni. Povdarek bo na prenosu novih znanj in dosežkov genomike, molekularane genetike in biomedicinske tehnologije v klinično prakso na področjih preprečavenja in diagnosticiranja bolezni, načrtovanju in uporabi molekularnih in bioloških zdravil ter individualiziranemu zdravljenju na osnovi genetskih testov. Študentom bodo predstavljene možnosti, prednosti, omejitve, tveganja in etični vidiki uporabe tehnologij molekularne genetike in genomike v medicinske namene. Povdarek bo tudi na interpretaciji genetskih testov in genetskem svetovanju pri monogenskih in kompleksnih boleznih.	Student will deeply understand how genes works in health and disease The focus will be on structure of genes and genomes, genetic diversity and mutation in human genome associated with disease. The focus will be on how can we transfer human genome discoveries and use of genomic technologies into clinical practice for disease prevention, diagnosis, development of novel molecular targeted biological drugs and individualized treatment based on patients genetic and gene expression profiles. Possibilities, advances, limitations, ethical issues and potential risks using genomics in biomedicine will be discussed. Examples of interpretation of genetic test and genetic counseling in monogenic and complex diseases will be discussed.

Predvideni študijski rezultati:	**Intended learning outcomes:**
Znanje in razumevanje:	Knowledge and understanding:

Delovanje genov in celotnega genoma v zdravju in bolezni Prenesljive/ključne spretnosti in drugi atributi: laboratorijske metode in experimenti v biomedicini	The role of genes and whole genome in health and disease. Transferable/Key Skills and other attributes: laboratory methods and experimental tecniques in biomedicine

Metode poučevanja in učenja:	**Learning and teaching methods:**
• Predavanja • Seminar	• Lectures • seminar

Načini ocenjevanja:	Delež (v %) / Share (in %)	**Assessment methods:**
Način (pisni izpit, ustno izpraševanje, naloge, projekt)		Method (written or oral exam, coursework, project):
seminar	**40%**	seminar
pisni izpit	**60%**	written examination
ŠTUDIJSKE OBVEZNOSTI ŠTUDENTOV: -študenti napišejo seminar na izbrano tematiko in ustno predstavijo seminar s kratkim predavanjem -pisni izpit		ACADEMIC OBLIGATIONS OF STUDENTS: -students should write an essay on selected topic and give oral presentation (seminar) -written exam
POGOJI ZA PRISTOP K POSAMEZNEMU PREVERJANJU ZNANJA: Opravljen seminar je pogoj za pristop k pisnemu izpitu.		REQUIREMENTS FOR ACCESS TO INDIVIDUAL KNOWLEDGE CHECKING: Students should complete seminar in order to approach to the written exam.

Reference nosilca / Course coordinator's references:

JOSTINS, Luke, MITROVIČ, Mitja, POTOČNIK, Uroš, et al. Host-microbe interactions have shaped the genetic architecture of inflammatorybowel disease. Nature, ISSN 0028-0836, 2012, vol. 491, no. 7422, str. 119-124, doi: 10.1038/nature11582. [COBISS.SI-ID 512230968], [JCR, SNIP, WoS do 22. 10. 2014: št. citatov (TC): 354, čistih citatov (CI): 353, čistih citatov na avtorja (CIAu): 17.40, normirano št. čistih citatov (NC): 140, Scopus do 22. 10. 2014: št. citatov (TC): 395, čistih citatov (CI): 392, čistih citatov na avtorja (CIAu): 19.32, normirano št. čistih citatov (NC): 624. SCI impact factor=36.28

RIVAS, Manuel A, MITROVIČ, Mitja, POTOČNIK, Uroš, et al. Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. Nature genetics, ISSN 1061-4036, 2011, vol. 43, no. 11, str. 1066-1073, doi: 10.1038/ng.952. [COBISS.SI-ID 15421974], [JCR, SNIP, WoS do 14. 10. 2014: št. citatov (TC): 180, čistih citatov (CI): 180, čistih citatov na avtorja (CIAu): 8.14, normirano št. čistih citatov (NC): 50, Scopus do 22. 10. 2014: št. citatov (TC): 192, čistih citatov (CI): 191, čistih citatov na avtorja (CIAu): 8.63, normirano št. čistih citatov (NC): 53

SCI impact factor=35.53

LIU, Jimmy Z, MITROVIČ, Mitja, POTOČNIK, Uroš, et al. Dense genotyping of immune-related disease regions identifies nine new risk loci for primary sclerosing cholangitis. Nature genetics, ISSN 1061-4036, 79 str., ilustr. http://www.nature.com/ng/journal/vaop/ncurrent/full/ng.2616.html, doi: 10.1038/ng.2616. [COBISS.SI-ID 512280376], [JCR, SNIP, WoS do 18. 12. 2013: št. citatov (TC): 9, čistih citatov (CI): 9, normirano št. čistih citatov (NC): 2, Scopus do 1. 1. 2014: št. citatov (TC): 12, čistih citatov (CI): 12, normirano št. čistih citatov (NC): 3];SCI impact factor=35.53

BERCE, Vojko, PINTO KOZMUS, Carina, POTOČNIK, Uroš. Association among ORMDL3 gene expression, 17q21 polymorphism and response to treatment with inhaled corticosteroids in children with asthma. Pharmacogenomics journal, ISSN 1470-269X, Dec. 2013, vol. 13, iss. 6. http://www.nature.com/tpj/journal/vaop/ncurrent/full/tpj201236a.html, doi: 10.1038/tpj.2012.36. [COBISS.SI-ID 4406079], [JCR, SNIP, WoS do 23. 12. 2013: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0, Scopus do 23. 10. 2013: št. citatov (TC): 3, čistih citatov (CI): 3, normirano št. čistih citatov (NC): 1] ;SCI impact factor=5.13

REPNIK, Katja, POTOČNIK, Uroš. Haplotype in the IBD5 region is associated with refractory Crohn's disease in Slovenian patients and modulates expression of the SLC22A5 gene. Journal of gastroenterology, ISSN 0944-1174, 2011, vol. 46, no. 9, str. 1081-1091, doi: 10.1007/s00535-011-0426-6. [COBISS.SI-ID 15110422], [JCR, SNIP, WoS do 5. 4. 2012: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0, Scopus do 24. 11. 2011: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0];SCI impact factor=4.16