2021/2022

Univerza v Mariboru

Medicinska fakulteta

UČNI NAČRT PREDMETA / SUBJECT SPECIFICATION	
Predmet:	**Izbrane vsebine in novosti v genetiki in genomiki medicine**
Subject 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
Dentalna medicina/Dental Medicine 2. stopnja/2nd cycle		**1**	**1 ali 2**

Vrsta predmeta / Course type Izbirni/Elective

Univerzitetna koda predmeta / University subject code:

Predavanja Lectures	Seminar Seminar	Vaje Tutorial	Klinične vaje Clinical training	Druge oblike študija Other forms of study	Samost. delo Individual work	ECTS
5	**40**				**45**	**3**

Nosilec predmeta / Lecturer: **red. prof. dr. Uroš Potočnik**

Jeziki / Languages:
Predavanja / Lecture: slovenščina/slovene
Vaje / Tutorial:

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

Prerequisits:

Vsebina:

- genetika in genomika
- genomika in odkrivanje novih zdravil
- asociacijske študija v celotnem genomu (GWAs) za odkrivanje novih genov povezanih s kompleksnimi boleznimi (avtoimunske bolezni, rak, astma, sladkorna…)
- sodobne tehnologije v genomiki: nanotehnologija, mikromreže (biočipi), tehnologija za avtomatsko gensko tipizacije enonukleotidnih polimorfizmov v celotnem genomu (SNPov), sekvenciranje naslednje generacije, masna spektroskopija visoke resolucije (nHPLC-HRMS), Maldi-TOF, 2-D elektroforeza
- funkcijska genomika
- transkriptomika

Content (Syllabus outline):

- genetics and genomics
- genomics in drug discovery
- Genome wide association study (GWAs) in common complex diseases (autoimmune diseases, cancer, asthma, diabetes…)
- modern genomic technologies: nanotechnology, microarrays (Biochips), whole genome genotyping of Single nucleotide Polymorphisms (SNPs), next generation sequencing (NGS), high resolution mass spectometrs (nHPLC-HRMS), Maldi-TOF, 2-D electrophoresis
- functional genomics
- transcriptomics
- pharmacogenomics and toxicology, personalized medicine

– farmakogenomika in toksikologija, personalizirana medicina – fiziološka genomika – primerjalna (komparativna) genomika – metagenomika v medicini – 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), integracija podatkov-omik; napovedni modeli, sistemska biologija in sistemska medicina; – projekt humani genom projekt HapMap, projekt 1000 genomov – populacijska genetika:velikost in struktura populacije, naravni izbor, mutacije, genetski zdrs, genski pretok, parjenje v sorodstvu; molekularna evolucija, molekularna ura, nastanek genomov – interptacija podatkov sekvenciranja naslednje generacije za postavitev diagnoze – novi pristopi in uspešni primeri genske terapije – etika v genomiki – Genetika bolezni zob in dlesni s primeri (parodontitisa (paradontoze), karies)	– physiological genomics – comparative genomics – genomics of microorganisms – proteomics – bioinformatics: human genome databases, bioinformatics tools (genome sequence analysis, design and segregation analysis of family tree data, statistical genetics) integration od-omic data; prediction models Human genome project, HapMap projects, 1000 genome projects – Population genetics: size and structure of population, natural selection, mutations, genetic drift, gene flow, inbreeding, molecular evolution, molecular clocks, evolution of genomes – From NGS data to diagnosis – New developments and successful examples in gene therapy – Ethics in genomics – Genetics of dental and gum pathology including periodontitis and dental caries

Temeljni literatura in viri / Textbooks:

1. Tom Strachan, Judith Goodship & Patrick Chinnery,Genetics and genomics in medicine, Garland Sc, ISBN 9780815344803, 2014
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:

1. Lesk, Arthur: Introduction to genomics, 3rd ed. - New York : Oxford University Press, ISBN 978-0-19-875483-1; 0-19-875483-3; cop. 2017
2. 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:	**Objectives:**
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	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

tehnologij molekularne genetike in genomike v medicinske namene. Povdarek bo tudi na interpretaciji genetskih testov in genetskem svetovanju pri monogenskih in kompleksnih boleznih.	discussed. Examples of interpretation of genetic test and genetic counselling in monogenic and complex diseases will be discussed.

Predvideni študijski rezultati:	**Intended learning outcomes:**
Znanje in razumevanje: Delovanje genov in celotnega genoma v zdravju in bolezni Prenesljive/ključne spretnosti in drugi atributi: laboratorijske metode in eksperimenti v biomedicini	Knowledge and Understanding: The role of genes and whole genome in health and disease Transferable/Key Skills and other attributes: laboratory methods and experimental techniques in biomedicine

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

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

Reference nosilca / Lecturer'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. [Print ed.], 2012, vol. 491, no. 7422, str. 119-124, doi: 10.1038/nature11582. [COBISS.SI-ID 512230968], [JCR, SNIP, WoS do 5. 6. 2016: št. citatov (TC): 945, čistih citatov (CI): 940, čistih citatov na avtorja (CIAu): 46.32, normirano št. čistih citatov (NC): 374, Scopus do 5. 5. 2016: št. citatov (TC): 986, čistih citatov (CI): 979, čistih citatov na avtorja (CIAu): 48.25, normirano št. čistih citatov (NC): 1558], SCI impact factor=36.28

CLEYNEN, Isabelle, BOUCHER, Gabrielle, JOSTINS, Luke, SCHUMM, Philip L., ZEISSIG, Sebastian, AHMAD, Tariq, ANDERSEN, Vibeke, ANDREWS, Jane M, ANNESE, Vito, BRAND, Stephan, et al., MITROVIČ, Mitja (sodelavec pri raziskavi), POTOČNIK, Uroš (sodelavec pri raziskavi), et al. Inherited determinants of Crohn's disease and ulcerative

colitis phenotypes : a genetic association study. **The Lancet**, ISSN 1474-547X. [Online ed.], 2016, vol. 387, iss. 10014, str. 156-167. http://www.thelancet.com/journals/lancet/article/PIIS0140-6736(15)00465-1/abstract, doi: 10.1016/S0140-6736(15)00465-1. [COBISS.SI-ID 512567352], [JCR, SNIP, WoS do 1. 6. 2016: št. citatov (TC): 3, čistih citatov (CI): 3, čistih citatov na avtorja (CIAu): 0.13, normirano št. čistih citatov (NC): 1, Scopus do 1. 6. 2016: št. citatov (TC): 4, čistih citatov (CI): 4, čistih citatov na avtorja (CIAu): 0.17, normirano št. čistih citatov (NC): 1];
SCI impact factor= 45.22

HUANG, Hailiang, FANG, Ming, JOSTINS, Luke, UMIĆEVIĆ MIRKOV, Maša, BOUCHER, Gabrielle, ANDERSON, Carl A., ANDERSEN, Vibeke, CLEYNEN, Isabelle, CORTES, Adrian, CRINS, François, et al., MITROVIČ, Mitja (sodelavec pri raziskavi), POTOČNIK, Uroš (sodelavec pri raziskavi), et al. Fine-mapping inflammatory bowel disease loci to single-variant resolution. **Nature**, ISSN 1476-4687. [Online ed.], 2017 http://www.nature.com/nature/journal/vaop/ncurrent/full/nature22969.html?WT.feed_name=subjects_computational-biology-and-bioinformatics, doi: 10.1038/nature22969. [COBISS.SI-ID 512723768],
SCI impact factor= 38.14

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 6. 6. 2016: št. citatov (TC): 280, čistih citatov (CI): 279, čistih citatov na avtorja (CIAu): 12.61, normirano št. čistih citatov (NC): 77, Scopus do 6. 5. 2016: št. citatov (TC): 299, čistih citatov (CI): 297, čistih citatov na avtorja (CIAu): 13.42, normirano št. čistih citatov (NC): 82]
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, issue 6, 523-529. 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 4. 2. 2016: št. citatov (TC): 6, čistih citatov (CI): 5, čistih citatov na avtorja (CIAu): 1.67, normirano št. čistih citatov (NC): 2, Scopus do 4. 12. 2015: št. citatov (TC): 8, čistih citatov (CI): 7, čistih citatov na avtorja (CIAu): 2.33, normirano št. čistih citatov (NC): 3]; SCI impact factor= 5.513

KODER, Silvo, REPNIK, Katja, FERKOLJ, Ivan, PERNAT DROBEŽ, Cvetka, SKOK, Pavel, WEERSMA, Rinse K., POTOČNIK, Uroš. Genetic polymorphism in ATG16L1 gene influences the response to adalimumab in Crohn's disease patients. Pharmacogenomics, ISSN 1462-2416, 2015, vol. 16, no. 3, str. 191-204, doi: 10.2217/pgs.14.172. [COBISS.SI-ID 512474168], [JCR, SNIP, WoS do 25. 6. 2017: št. citatov (TC): 9, čistih citatov (CI): 7, čistih citatov na avtorja (CIAu): 1.00, Scopus do 27. 6. 2017: št. citatov (TC): 9, čistih citatov (CI): 7, čistih citatov na avtorja (CIAu): 1.00]; SCI impact factor= 2.71