2025/2026

Univerza v Mariboru
Medicinska fakulteta

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:	**red. 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	• 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

• funkcijska genomika • transkriptomika • farmakogenomika in toksikologija • fiziološka genomika • 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	• pharmacogenomics and toxicology • 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 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. Genetics and genomics in medicine. Strachan, Tom; Lucassed, Anneke, 2nd ed. Boca Raton; Abingdon: CRC Press cop. 2023.
2. Human molecular genetics. Strachan Tom; Read, Andrew P., 5th ed. Boca Raton: CRC Press cop. 2019.
3. Ronald Cohn, Stephen Scherer and Ada Hamosh Thompson & Thompson Genetics and genomics in Medicine, 9the ed., Philadelphia : Elsevier ISBN: 9780323547628, 2024.
4. New clinical genetics : a guide to genomic medicine. Read, Andrew P. ; Donnai, Diane. 4th ed. - Banbury (UK) : Scion, cop. 2021
5. Jain K. K., Textbook of Personalized Medicine, 3rd ed., Springer; 2021

Dopolnilna literatura in viri/ Additional textbooks:

6. Lesk, Arthur: Introduction to genomics, 3rd ed. - New York : Oxford University Press, ISBN 978-0-19-875483-1; 0-19-875483-3; cop. 2017
7. 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:

Študenti bodo poglobili razumevanje načinov dedovanja, strukture in primerjave genov in genomov, genetske raznolikosti in genetskih napak povezanih z nastankom bolezni. Poudarek bo na prenosu novih znanj in dosežkov genomike, molekularane genetike in biomedicinske tehnologije v klinično prakso na področjih preprečevanja 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

Objectives and competences:

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

medicinske namene. Poudarek bo tudi na interpretaciji genetskih testov in genetskem svetovanju pri monogenskih in kompleksnih boleznih.	of genetic test and genetic counseling in monogenic and complex diseases will be discussed.

Predvideni študijski rezultati:	**Intended learning outcomes:**
Študent: * Pojasni zakonitosti prenosa genetske informacije med generacijami in delovanje genov ter celotnega genoma v zdravju in bolezni (npr. pri raku). * Analizira DNA-sekvence in poveže spremembe v genotipu s fenotipom * Pojasni dejavnike, ki vplivajo na frekvenco DNA polimorfizmov in genetsko raznolikost v različnih populacijah * Predlaga in pojasni napredne molekularne tehnike za določitev mutacij in genetske raznolikosti v molekularni evoluciji * Opiše razlike med eksperimentalni pristopi v DNA tehnologiji * Argumentira uporabnost različnih "omik" (genomika, transkriptomika, proteomika, epigenomika, metabolomika) v biomedicini in raziskovanju in jih primerjati Prenesljive/ključne spretnosti in drugi atributi: Študent zna ovrednostiti, katera metoda je najprimernejša za identifikacijo posameznega genetskega biooznačevalca.	The student: * Explains the principles of transmission of genetic information between generations and the function of genes and the whole genome in health and disease (e.g. cancer). * Analyses DNA sequences and relates changes in genotype to phenotype. * Explains the factors that influence the frequency of DNA polymorphisms and genetic diversity in different populations. * Proposes and explains advanced molecular techniques to identify mutations and genetic diversity in molecular evolution * Describes differences between experimental approaches in DNA technology * Discusses the applicability of different "omics" (genomics, transcriptomics, proteomics, epigenomics, metabolomics) in biomedicine and research and compare them Transferable/key skills and other attributes: The student will be able to evaluate which method is the most appropriate for the identification of a particular genetic biomarker.

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:		ACADEMIC OBLIGATIONS OF STUDENTS:
-študenti napišejo seminar na izbrano tematiko in ustno predstavijo seminar s kratkim predavanjem		-students should write an essay on selected topic and give oral presentation (seminar) -written exam

-pisni izpit 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:

GORENJAK, Mario, ZUPIN, Mateja, JEZERNIK, Gregor, SKOK, Pavel, POTOČNIK, Uroš. Omics data integration identifies ELOVL7 and MMD gene regions as novel loci for adalimumab response in patients with Crohn's disease. Scientific reports. 2021, vol. 11, str. 1-12, ilustr. ISSN 2045-2322. https://www.nature.com/articles/s41598-021-84909-z, https://doi.org/10.1038/s41598-021-84909-z, DOI: 10.1038/s41598-021-84909-z. [COBISS.SI-ID 54882051], [JCR, SNIP, WoS do 15. 4. 2023: št. citatov (TC): 6, čistih citatov (CI): 2, čistih citatov na avtorja (CIAu): 0,40, Scopus do 2. 6. 2023: št. citatov (TC): 7, čistih citatov (CI): 4, čistih citatov na avtorja (CIAu): 0,80]
kategorija: 1A1 (Z, A', A1/2);

ERNANDEZ-PACHECO, Natalia, VIJVERBERG, Susanne J, HERRERA-LUIS, Esther, LI, Jiang, SIO, Yang Yie, GRANELL, Raquel, CORRALES, Almudena, MAROTEAU, Cyrielle, LETHEM, Ryan, PEREZ-GARCIA, Javier, REPNIK, Katja, GORENJAK, Mario, BERCE, Vojko, POTOČNIK, Uroš, et al. Genome-wide association study of asthma exacerbations despite inhaled corticosteroids use. European respiratory journal. [Online ed.]. 2021, vol. 57, no. 5, str. [1]-16, ilustr. ISSN 1399-3003. https://erj.ersjournals.com/content/early/2020/11/11/13993003.03388-2020, DOI: 10.1183/13993003.03388-2020. [COBISS.SI-ID 42912771], [JCR, SNIP, WoS do 31. 3. 2023: št. citatov (TC): 10, čistih citatov (CI): 7, čistih citatov na avtorja (CIAu): 0,45, Scopus do 25. 5. 2023: št. citatov (TC): 11, čistih citatov (CI): 8, čistih citatov na avtorja (CIAu): 0,51]
financer: ARRS, Programi, P3-0067, SI, Farmakologija in farmakogenomika; MIZŠ, C3330-16-500106, SyPharmPedia
kategorija: 1A1 (Z, A'', A', A1/2)

HERRERA-LUIS, Esther, GORENJAK, Mario, POTOČNIK, Uroš, et al. Multi-ancestry genome-wide association study of asthma exacerbations. Pediatric allergy and immunology. June 2022, vol. 33, iss. 6, 17 str. ISSN 1399-3038. https://onlinelibrary.wiley.com/doi/epdf/10.1111/pai.13802, DOI: 10.1111/pai.13802. [COBISS.SI-ID 111186947], [JCR, SNIP, WoS do 7. 4. 2023: št. citatov (TC): 3, čistih citatov (CI): 3, čistih citatov na avtorja (CIAu): 0,17, Scopus do 26. 5. 2023: št. citatov (TC): 5, čistih citatov (CI): 5, čistih citatov na avtorja (CIAu): 0,28]
kategorija: 1A1 (Z, A', A1/2);

JEZERNIK, Gregor, GORENJAK, Mario, POTOČNIK, Uroš. MIF variant rs755622 is associated with severe Crohn's disease and better response to anti-TNF adalimumab therapy. Genes. 2023, vol. 14, issue 2, [article no.] 452, str. [1]-15, ilustr. ISSN 2073-4425. https://www.mdpi.com/2073-4425/14/2/452, https://doi.org/10.3390/genes14020452, DOI: 10.3390/genes14020452. [COBISS.SI-ID 141538563], [JCR, SNIP, WoS, Scopus]
financer: ARRS, Programi, P3-0427, SI, Sistemski pristopi k raziskavam človeškega genoma za personalizirano medicino kroničnih imunskih bolezni; ARRS, Projekti, J3-9258, SI, Molekularno genetski biooznačevalci in mehanizmi neodzivnosti na biološko zdravljenje z anti-TNF bolnikov s kroničnimi imunskimi boleznimi
kategorija: 1A2 (Z, A1/2);

GORENJAK, Mario, JEZERNIK, Gregor, KRUŠIČ, Martina, SKOK, Pavel, POTOČNIK, Uroš. Identification of novel loci involved in adalimumab response in Crohn's disease patients using integration of genome profiling and isoform-level immune-cell deconvoluted transcriptome profiling of colon tissue. Pharmaceutics. [Online ed.]. Sep. 2022, vol. 14, issue 9, str. 1-16, ilustr. ISSN 1999-4923. https://doi.org/10.3390/pharmaceutics14091893,

https://www.mdpi.com/1999-4923/14/9/1893, DOI: 10.3390/pharmaceutics14091893. [COBISS.SI-ID 120609795], [JCR, SNIP, WoS, Scopus]
financer: ARRS, Programi, P3-0427, SI, Sistemski pristopi k raziskavam človeškega genoma za personalizirano medicino kroničnih imunskih bolezni; ARRS, Projekti, J3-9258, SI, Molekularno genetski biooznačevalci in mehanizmi neodzivnosti na biološko zdravljenje z anti-TNF bolnikov s kroničnimi imunskimi boleznimi
kategorija: 1A1 (Z, A', A1/2);