OPIS PREDMETA / SUBJECT SPECIFICATION	
Predmet:	**Genetika**
Subject Title:	**Genetics**

Študijski program Study programme	Študijska smer Study field	Letnik Year	Semester Semester
BIOMEDICINSKA TEHNOLOGIJA/BIOMEDICAL TECHNOLOGY 3. stopnja/3rd Degree		1	1/2

Vrsta predmeta / Course type Temeljni/Basic

Univerzitetna koda predmeta / University subject code: 1005

Predavanja Lectures	Seminar Seminar	Sem. vaje Tutorial	Lab. vaje Lab. work	Teren. vaje Field work	Samost. delo Individ. work	ECTS
20	40		15		195	9

Nosilec predmeta / Lecturer:	Prof. dr. Nadja KOKALJ VOKAČ Prof. dr. Peter DOVČ Prof. dr. Damjan GLAVAČ

Jeziki / Languages:	**Predavanja / Lecture:**	Slovenščina / Slovene Angleščina / English
	Vaje / Tutorial:	

Pogoji za vključitev v delo oz. za opravljanje študijskih obveznosti:	**Prerequisites:**
Kandidat mora imeti pred vpisom ustrezno znanje iz naravoslovnih ved z ustreznega področja na nivoju univerzitetnega študija.	Prior to entering, the candidate for postgraduate program must have an appropriate knowledge and understanding of bioscience (biology, chemistry, physics, mathematics) on the university level.

Vsebina:	**Contents (Syllabus outline):**
Vloga genetike v medicini. Struktura genoma in njen pomen za medicinsko genetiko. Izbrani primeri monogenskih in poligenskih bolezni. Genetika raka. Nove metode molekularne diagnostike. Nekodirajoča DNA in njen pomen za medicinsko genetiko. Primeri uporabe asociacijske analize na celotnem genomu (ang. GWAS) npr. pri ALS (amiotrofični lateralni sklerozi). Dolga nekodirajoča RNA (ang. lncRNA) pri raku. Pomen in vloga mikroRNA pri boleznih. Izbrani primeri farmakogenetike /farmakogenomike. Različne ravni uravnavanja izražanja genov (transkripcija, stuktura kromatina, ranslacija, procesiranje RNA, RNA inerferenca, razvojno uravnavanje genske ekspresije). Pomen metilacije pri raku in metode za njeno določanje. Odstopanja od Menedlovih principov dedovanja in populacijska genetika Analiza genoma in odkrivanje vzročnih regij za fenotipske lastnosti (poligenske lastnosti)	The role of genetics in medicine. The structure of the genome and its significance for medical genetics. Selected cases of monogenic and polygenic diseases. Genetics of cancer. New methods of molecular diagnostics. Non-coding DNA and its significance for medical genetics. Examples of the use of GWAS-related association assay, e.g. in ALS (amyotrophic lateral sclerosis). Long non-coding RNA (angina lncRNA) in cancer. The importance and role of microRNA in diseases. Selected examples of pharmacogenetics / pharmacogenomics. Different levels of gene expression control (transcription, chromatin structure, translation, RNA processing, RNA inertia, developmental regulation of gene expression). The importance of methylation in cancer and methods for its determination. Deviations from Mendel's principles of inheritance and population genetics

Nove tehnologije za študij transkriptiona, transkriptomika posameznih celic Tehnologije, ki omogočajo gensko zdravljenje (različne oblike kloniranja,matične celice, celična terapija) Strategije odkrivanja vzročnih lokusov za recesivne bolezni (regije homozigotnosti) Genetsko ozadje procesov staranja Mitohondrijske mutacije in z njimi povzročene bolezni. Interakcija mikrobioma z genomom gostitelja Odkrivanje in mapiranje strukturnih genomskih sprememb. Potek obravnave bolnika z genetsko boleznijo v ambulanti za genetsko svetovanje in genetska diagnostika. Strukturne genomske variabilnosti in njihov pomen v razvoju in nevropsihiatričnih stanjih.	Genome analysis and detection of causal genomic regions for phenotypic traits (polygenic) New transcriptomic technologies, transcriptomics of single cells Gene therapy technologies (various cloning strategies, stem cells, cell therapy) Strategies for detecting causative loci for recessive diseases (regions of homozygosity) Genetic background of aging processes Mitochondrial mutations and diseases caused by them. Interaction of microbial with host genome Detection and mapping of structural genomic mutations. The course of treatment of a patient with a genetic disorder in a clinic for genetic counseling and genetic diagnostics. Structural genomic variability and their importance in development and neuropsychiatric conditions.

Temeljni študijski viri / Textbooks:

Predavanja
Članki za seminarje
Učbeniki:
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.
Andrew Read and Dian Donnai: New Clinical Genetics, 3nd Ed., 2015, ISBN 9781907904677.
G. Bradley Schaefer, James N. Thompson, Jr. Medical Genetics: An Integrated Approach. McGraw-Hill Education Press, 2014.

Cilji:	**Objectives:**
Premet ponuja študentu pregledna in nekatera poglobljena znanja o zgradbi, organizaciji ter delovanju prokariontskega in eukariontskega genoma. 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 subject offers a review and some extensive knowledge of the structure, organization and action of the prokaryotic and eukaryotic genome. It will acquaint the student with the role of genetic factors in human diseases and with their contribution to multifactorial diseases, with a complex analysis of the action and transfer of genetic information and inheritance. The stress is on the applicative role of genetics in modern medical techniques, diagnosis and gene therapy.

Predvideni študijski rezultati:	**Intended learning outcomes:**
Znanje in razumevanje: Študent si poglobi znanje o genetskih mehanizmih ter razširi poznavanje uporabe genetskih tehnologij v medicini in biotehnologiji. Praktična znanja pridobi pri individualnem delu ob izdelavi raziskovalnega (doktorskega) projekta v genetskem laboratoriju, kjer se seznani z osnovnimi tehnikami genetske analize. **Prenesljive/ključne spretnosti in drugi atributi:** Ob izdelavi raziskovalnega (doktorskega) projekta se nauči uporabljati metode ter rezultate genetskih analiz v različne aplikativne namene na področju medicinske genetske diagnostike.	**Knowledge and Understanding**: The student acquires knowledge of genetic mechanisms and a broader knowledge of the use of genetic technologies in medicine and biotechnology. He/she acquires practical knowledge in individual work in preparing a research (doctoral) project in the genetic lab, where he/she becomes acquainted with the basic techniques of genetic analysis. **Transferable/Key Skills and other attributes:** In preparing the research (doctoral) project the student learns to use the methods and results of genetic analyses for various applicative purposes in the field of medical genetic diagnosis.

Metode poučevanja in učenja:	Teaching and learning methods:
• predavanja • seminarji • laboratorijske vaje	• Lectures • Seminars • laboratory work

Načini ocenjevanja:	Delež (v %) / Weight (in %)	Assessment:
pisni izpit, seminarske naloge	**60 %** **40 %**	Written examination Seminar work

Reference nosilca / Lecturer's references:

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. [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

RIGGS, Erin R., NELSON, Tristan, MERZ, Andrew, ACKLEY, Todd, BUNKE, Brian, COLLINS, Christin D., COLLINSON, Morag N., FAN, Yao-Shan, GOODENBERGER, McKinsey L., GOLDEN, Denae M., KRGOVIĆ, Danijela, **KOKALJ-VOKAČ, Nadja**, et al. Copy number variant discrepancy resolution using the ClinGen dosage sensitivity map results in updated clinical interpretations in ClinVar. Human mutation, ISSN 1098-1004, 2018, vol. , no. , f. 1-35. https://onlinelibrary.wiley.com/doi/abs/10.1002/humu.23610, https://doi.org/10.1002/humu.23610, doi: 10.1002/humu.23610. [COBISS.SI-ID 6435647] IF = 4,71

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YANG, Bin, CUI, Leilei, PEREZ-ENCISO, Miguel, TRASPOV, Aleksei, CROOIJMANS, Richard, ZINOVIEVA, Natalia, SCHOOK, Lawrence B., ARCHIBALD, Alan, GATPHAYAK, Kesinee, KNORR, Christophe, TRIANTAFYLLIDIS, Alex, ALEXANDRI, Panoraia, SEMIADI, Gono, HANOTTE, Olivier, DIAS, Deodália, **DOVČ, Peter**, UIMARI, Pekka, IACOLINA, Laura, SCANDURA, Massimo, GROENEN, Martien, HUANG, Lusheng, MEGENS, Hendrik-Jan. Genome-wide SNP data unveils the globalization of domesticated pigs. Genetics selection evolution, ISSN 1297-9686. [Online ed.], 2017, vol. 49, no. 71, str. 1-15, ilustr. https://gsejournal.biomedcentral.com/articles/10.1186/s12711-017-0345-y, doi: 10.1186/s12711-017-0345-y. [COBISS.SI-ID 4038024],

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DELSER, Pierpaolo Maisano, RAVNIK-GLAVAČ, Metka, GASPARINI, Paolo, **GLAVAČ, Damjan,** MEZZAVILLA, Massimo. Genetic landscape of Slovenians : past admixture and natural selection pattern. Frontiers in genetics, ISSN 1664-8021, Nov. 2018, vol. 9, str. 1-8, ilustr. https://www.frontiersin.org/articles/10.3389/fgene.2018.00551/full, doi: 10.3389/fgene.2018.00551. [COBISS.SI-ID 34067673]