



## UČNI NAČRT PREDMETA / SUBJECT SPECIFICATION

<b>Predmet:</b>	Izbrane vsebine in novosti v molekularni biologiji
<b>Subject Title:</b>	Selected topics and novelties in molecular biology

Študijski program Study programme	Študijska smer Study field	Letnik Year	Semester Semester
Spošna medicina General medicine – EMŠP		1	2

Univerzitetna koda predmeta / University subject code:

Predavanja Lectures	Seminar Seminar	Sem. vaje Tutorial	Lab. vaje Labor work	Teren. vaje Field work	Samost. delo Individ. work	ECTS
5	40				45	3

Nosilec predmeta / Lecturer:

Izred. prof. dr. Uroš Potočnik

Jeziki / Languages:	Predavanja / Lecture: Vaje / Tutorial:	Slovenski/Slovene
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Pogoji za vključitev v delo oz. za opravljanje študijskih obveznosti:

**Vsebina:**

DNA struktura in lastnosti, replikacija (prokarionti, eukarionti), rekombinacija DNA, DNA popravljalni mehanizmi, DNA mutacije, struktura in funkcija genov in kromosomov  
 RNA struktura in lastnosti, vrste RNA molekul in funkcije, transkripcija (prokarionti, eukarionti), postranskripcijske modifikacije  
 Struktura proteinov, sinteza proteinov, posttranslacijske modifikacije proteinov, zvijanje proteinov, transport proteinov  
 Regulacija proteinske sinteze: regulacija ekspresije genov pri prokariontih, pri bakteriofagih, pri evkariotskih organizmih (enoceličnih, multicelularnih, povezava z embrionalnim razvojem), regulacija na ravni translacije in posttranslacijska regulacija,  
 Embrionalni razvoj  
 Celični cikel, proliferacija, diferenciacija celic, apoptoza  
 Povezovanje celic v tkiva, komunikacija med celicami, signalne poti, receptorji, hormoni  
 Imunski sistem in avtoimunske bolezni  
 Virusi, HIV, SARS, DNA diagnostika pri infekcijskih boleznih  
 Molekularna patologija: molekularni mehanizmi vključeni v nastanek bolezni, od bolezni do gena metode in eksperimentalne tehnike v molekularni biologiji: izolacija bioloških materialov (DNA, RNA, proteini)

**Content (Syllabus outline):**

DNA structure and characteristics, replication (prokaryotes, eukaryotes), recombination, repair and mutations, structure and function of genes and chromosomes,  
 RNA structure characteristics: role of different types of RNA, transcription (prokaryotes, eukaryotes), post transcription modification  
 Protein structures, synthesis of proteins, translation, posttranslational modifications, protein folding, protein trafficking  
 Regulation of protein synthesis: transcriptional regulation of gene expression, regulation of translation, posttranslational regulation  
 Embryonic development  
 Cell division (meiosis, mitosis)  
 Cell cycle: proliferation, differentiation, apoptosis  
 Integration of cells into tissues, communication between cells, signal transduction, receptors, hormone signaling  
 Immune system  
 Viruses :HIV, SARS, Avian influence, DNA diagnostics and infection diseases  
 Molecular pathology: from disease to gene  
 Methods and experimental techniques in molecular biology: isolation of biological molecules (DNA, RNA, proteins) from clinical samples (blood, biopsy, tissue, resection specimens) and cell cultures; plasmid DNA

<p>proteinov) iz kliničnih vzorcev (kri, biopsije, tkivo-resektati) in celičnih kultur, izolacija plazmidne DNA, gelska elektroforeza, pomnoževanje DNA z verižno reakcijo z encimom polimerazo (PCR), analiza genske ekspresije z metodo PCR v realnem času (Taqman), hibridizacija odtisa (southern, northern, western), konstrukcija cDNA in genomske knjižnice, Rekombinantna DNA tehnologija, kloniranje človekovih genov</p> <p>Monogenske genetske bolezni, kompleksne genetske bolezni,</p> <p>Molekularna biologija raka: onkogeni, tumorsko zaviralni geni, dedne oblike, molekulska diagnostika in zdravljenje, biološka zdravila</p> <p>Vloga molekularne biologije v sodobni družbi: etični, sociološki in ekonomski vidiki</p>	<p>isolation, Polymerase Chain Reaction (PCR), gene expression analysis using Real time PCR (Taqman); hybridization and blotting (southern, western, northern); cDNA and genomic libraries</p> <p>Recombinant DNA technology, cloning of human genes</p> <p>Monogenic (Mendelian) and complex diseases</p> <p>Molecular biology of cancer: oncogenes, tumor suppressor genes, hereditary cancer, molecular diagnostics and treatment, biological drugs</p> <p>Molecular biology and society: ethical and economical aspects</p>
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**Temeljni literatura in viri / Textbooks:**

1. B. ALBERTS et al.: Molecular biology of the cell., 5th Ed., Gerland Publish, Inc., New York, 2008
2. EPSTEIN RJ: Human molecular biology, An Introduction to the Molecular Basis of Health and Disease; Cambridge University Press, Cambridge, 2002
3. LODISH H., Baltimore D., Berk A., Zipursky S.L., Matsudaira P., Darnell J.: Molecular Cell Biology, 5th Ed., Scientific American Books, Freeman and Co., New York, 2004 STRACHAN T and READ AP: Human Molecular genetics, Gerland Publish, Inc., New York, 3rd ed., 2004 Liciano J. (ed.): Pharmacogenomics, The Search for Individualized Therapies, John Wiley&Sons, 2002 R.J.M

**Cilji:**

Predmet bo nudil študentom poglobitev razumevanja bistvenih molekularnih in bioloških procesov v celici, tkivih, organih in celotnem organizmu. Poseben povidarek bo na razumevanju patoloških sprememb v molekularnih procesih pri nastanku, razvoju in zdravljenju bolezni. Predstavljene bodo osnovne metode in eksperimentalne tehnike v molekularni biologiji in molekularni patologiji ter njihova uporaba pri raziskavah in preiskavah molekularnih označevalcev v diagnostiki, prognozi, načrtovanju novih zdravil in individualiziranem zdravljenju

**Objectives:**

Student will have deep understanding of molecular and biological processes in cells, tissues, organs and whole human organism during health and disease. The focus will be on molecular mechanisms during disease development and treatment. Student will learn most important molecular biology and molecular pathology laboratory methods for diagnostics, biomarker discovery, novel drug development and individualized treatment based on patients genetic makeup.

**Predvideni študijski rezultati:****Intended learning outcomes:****Znanje in razumevanje:**

- osnovnimi molekularnimi in biološkimi procesi v celici, tkivih, organih in celotnem organizmu v zdravju in bolezni

Prenesljive/ključne spretnosti in drugi atributi: laboratorijske metode in eksperimenti v biomedicini

**Knowledge and Understanding:**

- molecular and biological processes in cells, tissues, organs and whole human organism during health and disease

Transferable/Key Skills and other attributes: laboratory methods and experimental techniques in biomedicine

**Metode poučevanja in učenja:****Learning and teaching methods:**

- Predavanje
- Seminar

- Lectures
- seminar

**Načini ocenjevanja:**

Delež (v %) / Weight (in %)

**Assessment:**

<p>Način (pisni izpit, ustno izpraševanje, naloge, projekt) seminar Izpit</p>	<p><b>40 %</b> <b>60 %</b></p>	<p>Type (examination, oral, coursework, project): seminar Exam</p>
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**Reference nosilca / Lecturer's references:**

JOSTINS, Luke, MITROVIČ, Mitja, POTOČNIK, Uroš, et al. Host-microbe interactions have shaped the genetic architecture of inflammatory bowel 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 15. 1. 2014: št. citatov (TC): 131, čistih citatov (CI): 130, normirano št. čistih citatov (NC): 52, Scopus do 8. 1. 2014: št. citatov (TC): 168, čistih citatov (CI): 167, normirano št. čistih citatov (NC): 266]; 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. 1. 2014: št. citatov (TC): 124, čistih citatov (CI): 124, normirano št. čistih citatov (NC): 34, Scopus do 15. 1. 2014: št. citatov (TC): 139, čistih citatov (CI): 139, normirano št. čistih citatov (NC): 38]; 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