Univerza v Mariboru

MEDICINSKA FAKULTETA

UČNI NAČRT PREDMETA / SUBJECT SPECIFICATION	
Predmet:	Molekularna biologija in genetika
Subject Title:	Molecular biology and genetics

Študijski program Study programme	Študijska smer Study field	Letnik Year	Semester Semester
Splošna medicina General medicine		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
30	15		15		90	**5**

Nosilec predmeta / Lecturer: Red.prof. dr. Nadja Kokalj Vokač

Jeziki / Languages:	**Predavanja / Lecture:**	Slovenščina / slovene
	Vaje / Tutorial:	Slovenščina / slovene

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

Vsebina:	**Content (Syllabus outline):**
1.Vloga molekularne biologije in genetike v medicini. 2. Struktura, morfologija in klasifikacija humanih kromosomov. 3. Osnove citogenetike, molekularna citogenetika, klinična citogenetika – sindromologija. 4. Spolni kormosomi, X inaktivacija napake spolnih kromosomov. 5. Dedovanje, Mendelejevi zakoni, dominantno, recesivno dedovanje, atipični vzorci dedovanja. 6. Struktura in lastnosti DNA molekule. 7. Podvajanje DNA: razlike med prokarionti in evkarionti. 8. DNA mutacije, popravljanje DNA napak. 9. RNA, transkripcija, translacija, vrste RNA, ribocimi. 10. Procesiranje proteinov, posttranskripcijske modifikacije proteinov. 11. Človeški genom, jederni genom, genetski kod, zgradba gena, genske družine, tandemske ponovitve, mitohondrijski genom. 12. Regulacija genske ekspresije. 13. Epigenetika, DNA metilacija. 14. Rekombinantna DNA tehnologija, genetski inžiniring, biotehnologija. 15. Virusi: zgradba, lastnosti, potek virusne infekcije. Prioni. 16. Genetska ranznolikost med posamezniki in populacijami, polimorfizmi, genetsko neravnostežje.	1. The role of molecular biology and genetics in medicine. 2. Structure, morphology and classification of human chromosomes. 3. The principles of cytogenetics, molecular cytogenetics, clinical cytogenetics (syndromology). 4. Sex chromosomes, X inactivation, sex chromosomes aberrations. 5. Inheritance, Mendelien laws, dominant, recessive single-gene inheritance, atypical patterns of inheritance. 6. Structure and features of DNA molecule. 7. DNA replication: prokaryotes, eukaryotes. 8. DNA mutations, repairing mechanisms. 9. RNA, transcription, translation, ribocimes 10. Protein processing, posttranslational modifications of proteins. 11. Human genome, nuclear genome, genetic code, structure of the gene, gene families, tandem repeats, mitochondrial genome. 12. Regulation of gene expression. 13. Epigenetics, DNA methylation. 14. Recombinant DNA technology, genetic engineering, biotechnology. 15. Viruses: structure, features, course of virus infection. Prions. 16. Genetic diversity between individuals and in populations, polymorphisms, linkage disequilibrium.

17. Kompleksne bolezni, multifaktorsko deodvanje. 18. Citogenetika in molekularna genetika raka. 19. Genetika imunskega sistema. 20. Razvojna genetika. 21. Izbrani primeri genetskih bolezni. 22. Genomika, proteomika, farmakogenomika. 23. Genetsko svetovanje, prenatalna diagnostika. 24. Genetika in družba.	17. Complex diseases, multifactorial inheritance. 18. Cytogenetics and molecular genetic of cancer. 19. Genetics of immune system. 20. Development genetics. 21. Genetic diseases – selected cases. 22. Genomics, proteomics, farmacogenomics. 23. Genetic counseling, prenatal diagnosis. 24. Genetic and society.

Temeljni literatura in viri / Textbooks:

1. Thompson & Thompson : Genetics in Medicine, W.B.Saunders Company., 6the ISBN 0-7216-0244-4 and 7the ed. ISBN: 9781416030805, 2007.
2. Alberts, Bray, Lewis, Raff, Roberts, Watson: Molecular Biology of the Cell, Garland Publishing, Inc,New York London. Edition: 5,ISBN 0815341113, 9780815341116, 2007.
3. Lodish: Molecular Cell Biology, W.H.Freeman and Company, 6th ed., ISBN-10: 9780716776017 ISBN-13: 9780716776017, 2007.
4. Strachan T, Read AP: Human Molecular Genetics 2, BIOS Scientific Publishers Limited. 3rd ed., ISBN-13: 9780471330615 ISBN: 0471330612, 2003.

Dodatna literatura:

5. Vogel Motulsky, Human Genetcs, Springer. 4rd ed. ISBN 978-3-540-37653-8, 2009.
6. Lewin B: Genes VIII, Oxford University press. ISBN 0-13-123924-4., 2004.
7. Basiswissen Humangenetik: Schaaf, Zschocke, Springer Verlag, ISBN 978-3-540-71222-0
8. Molekulare Genetik, Knippers et al., Georg Thieme Verlag, ISBN 3-13-477005-9
9. Biologie für Mediziner, Hirsch-Kauffmann, Schweiger, Georg Thieme Verlag, ISBN 3-13-706502-X

Cilji:	**Objectives:**
Cilj predmeta je, da študentu nudi znanje o zgradbi, organizaciji in funkciji informacijskih makromulekul, nosilcih dednega materiala ter o zakonitostih prenosa in ohranjanja genetske informacije s poudarkom na humanem genomu. Premet ponuja študentu pregledna in nekatera poglobljena znanja o zgradbi, organizaciji ter delovanju prokariontskega in eukariontskega genoma. Študenta seznani z možnimi načini analize genoma in ugotavljanja genetskih napak.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. Povdarek predavanj je na aplikativni vlogi genetike v sodobnih medicinskih tehnikah, diagnostiki in genski terapiji.	The goal of the subject is to offer the student knowledge of the structure and function of informative macromoleculs. Of principal interest are the macromolecule complexes of DNA, RNA and protein and the processes of replication, transcription and translation and technologies involved in manipulating these molecules. The courses offer to students overvew and some selected topis about organization, function and structure of eukaryotic and prokaryotic genom with special attantion to human genom. The student gets the knowladge of major techniques used for genom analysis, and mutation analysis. The student is introduced to the role of genetic factors in etiology of human heriditary disorders, the contribution to multifactorial diseases and understanding the comlex information of functioning and transmission of genetic information. Special attantion is given to diagnostic techniques, recombinant DNA technology and gene therapy.

Predvideni študijski rezultati:	**Intended learning outcomes:**
Prenesljive/ključne spretnosti in drugi atributi: 1. Zna sestaviti kariogram po ISCN nomenklaturi. 2. Zna ločiti strukturno od numerčne kromosomske spremembe. 3. Zna izolirati DNA molekulo iz periferne krvi in določiti njeno koncentracijo. 4. Zna izvesti verižno reakcijo s polimerazo ter določiti produkt z gelsko elektroforezo.	Transferable/Key Skills and other attributes: 1. Knowledge of makeing karyotype to ISCN nomenclature. 2. Knowledge of differences between structural and numerical chromosome aberration. 3. Knowledge of isolation of DNA molecules from periferal blood and messuring its concentration. 4. Knowledge of polymerase chain reaction and determination of the product on gel electroforesis.

Metode poučevanja in učenja:	Learning and teaching methods:
Predavanja : 80%. Laboratorijske vaje: 20%.	Lectures : 80% Courswork : 20%

Načini ocenjevanja:	Delež (v %) / Weight (in %)	Assessment:
Seminarji Kolokvij iz vaj. Pisni izpit. (opravljene naloge pri vajah so pogoj za pristop k pisnemu izpitu)	**10 %** **20 %** **70 %**	Seminary work Courswork examination Written examination (Courswork project is condition for approach to written examination)

Reference nosilca / Lecturer's references:

MACEDONI-LUKŠIČ, Marta, KRGOVIĆ, Danijela, ZAGRADIŠNIK, Boris, KOKALJ-VOKAČ, Nadja. Deletion of the last exon of SHANK3 gene produces the full Phelan-McDermid phenotype : a case report. Gene, ISSN 0378-1119. [Print ed.], 2013, vol. 524, no. 2, str. 386-389, ilustr. http://www.sciencedirect.com/science/article/pii/S0378111913004642, doi: 10.1016/j.gene.2013.03.141. [COBISS.SI-ID 721836], [JCR, SNIP, WoS do 16. 9. 2013: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0, Scopus do 15. 6. 2013: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0]

ŽERJAVIČ, Katja, ZAGRADIŠNIK, Boris, LOKAR, Lidija, GLASER, Marjana, KOKALJ-VOKAČ, Nadja. The association of the JAK2 46/1 haplotype with non-splanchnic venous thrombosis. Thrombosis research, ISSN 0049-3848. [Print ed.], 2013, vol. , no. , str. http://www.sciencedirect.com/science/article/pii/S0049384813002727, doi: 10.1016/j.thromres.2013.06.021. [COBISS.SI-ID 4724799], [JCR, SNIP, WoS do 7. 10. 2013: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0, Scopus do 21. 9. 2013: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0]

ERJAVEC ŠKERGET, Alenka, STANGLER HERODEŽ, Špela, ZAGORAC, Andreja, ZAGRADIŠNIK, Boris, MUJEZINOVIĆ, Faris, KOKALJ-VOKAČ, Nadja. Slovenian five-year experiences with rapid prenatal diagnosis of common chromosome aneuploidies using quantitative fluorescence polymerase chain reaction. Genetic testing and molecular biomarkers, ISSN 1945-0265, 2013, vol. , no. , str. http://online.liebertpub.com/doi/abs/10.1089/gtmb.2013.0082, doi: 10.1089/gtmb.2013.0082. [COBISS.SI-ID 4738623], [JCR, SNIP, WoS do 7. 10. 2013: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0, Scopus do 23. 9. 2013: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0]

KRGOVIĆ, Danijela, MARČUN-VARDA, Nataša, ZAGORAC, Andreja, KOKALJ-VOKAČ, Nadja. Submicroscopic interstitial deletion of chromosome 11q22.3 in a girl with mild mental retardation and facial dysmorphism: Case report. Molecular cytogenetics, ISSN 1755-8166. [Online ed.], 2011, [Vol.] 4, 17. http://www.molecularcytogenetics.org/content/pdf/1755-8166-4-17.pdf, doi: doi:10.1186/1755-8166-4-17. [COBISS.SI-ID 4029247], [JCR, SNIP, WoS do 5. 11. 2012: št. citatov (TC): 0, čistih citatov (CI): 0, normirano št. čistih citatov (NC): 0, Scopus do 27. 11. 2013: št. citatov (TC): 2, čistih citatov (CI): 2, normirano št. čistih citatov (NC): 1]

KOKALJ-VOKAČ, Nadja, KODRIČ, Tatjana, ERJAVEC ŠKERGET, Alenka, ZAGORAC, Andreja, TAKAČ, Iztok. Screening of TERC gene amplification as an additional genetic diagnostic test in detection of cervical preneoplastic lesions. Cancer genetics and cytogenetics, ISSN 0165-4608. [Print ed.], 2009, vol. 195, no. 1, str. 19-22. [COBISS.SI-ID 3500607], [JCR, SNIP, WoS do 17. 1. 2013: št. citatov (TC): 3, čistih citatov (CI): 3, normirano št. čistih citatov (NC): 1, Scopus do 6. 8. 2013: št. citatov (TC): 7, čistih citatov (CI): 7, normirano št. čistih citatov (NC): 2]