



Univerza v Mariboru
University of Maribor

Medicinska fakulteta
Faculty of Medicine



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:

Prerequisites:

Vsebina:

- 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 kromosomi, 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, popraviljanje DNA napak.
9. RNA, transkripcija, translacija, vrste RNA,

Content (Syllabus outline):

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, Mendelian 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, ribosomes
10. Protein processing, posttranslational

ribocimi. 10. Procesiranje proteinov, posttranskripcijske modifikacije proteinov. 11. Človeški genom, jedrni genom, genetski kod, zgradba gena, genske družine, tandemske ponovitve, mitohondrijski genom. 12. Regulacija genske ekspresije. 13. Epigenetika, DNA metilacija. 14. Rekombinantna DNA tehnologija, genetski inženiring, biotehnologija. 15. Virusi: zgradba, lastnosti, potek virusne infekcije. Prioni. 16. Genetska raznolikost med posamezniki in populacijami, polimorfizmi, genetsko neravnostezje. 17. Kompleksne bolezni, multifaktorsko dedovanje. 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.	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. 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.
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Temeljni literatura in viri / Textbooks:

1. Thompson & Thompson : Genetics in Medicine, Sixth Edition, W.B.Saunders Company, 2001.
2. Strachan T, Read AP: Human Molecular Genetics 2, BIOS Scientific Publishers Limited, 2000.
- 3.. Alberts, Bray, Lewis, Raff, Roberts, Watson: Molecular Biology of the Cell, 4rd Ed., Garland Publishing, Inc, New York & London, 2002.
4. Lodish: Molecular Cell Biology, W.H.Freeman and Company, 5th Ed. 2003.
5. Cox & Sincler: Molecular Biology in Medicine, Blackwell Science, 1997.
6. Lewin B: Genes VII, Oxford University press, 2000.
7. Voet D and Voet JG: Fundamentals of Biochemistry, John Wiley & Sons, 1999.

Cilji:

Cilj predmeta je, da študentu nudi znanje o zgradbi, organizaciji in funkciji informacijskih makromolekul, 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 prokariotskega in eukariotskega 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 multifaktorskih boleznih, s kompleksno analizo delovanja in prenosa genetske informacije ter dedovanjem. Povdarek

Objectives:

The goal of the subject is to offer the student knowledge of the structure and function of informative macromolecules. 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 overview and some selected topics about organization, function and structure of eukaryotic and prokaryotic genome with special attention to human genome. The student gets the knowledge of major techniques used for genome analysis, and mutation analysis. The student is introduced to the role of genetic factors in etiology of human hereditary disorders, the contribution to multifactorial

predavanj je na aplikativni vlogi genetike v sodobnih medicinskih tehnikah, diagnostiki in genski terapiji.	diseases and understanding the complex information of functioning and transmission of genetic information. Special attention is given to diagnostic techniques, recombinant DNA technology and gene therapy.
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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 making karyotype to ISCN nomenclature. 2. Knowledge of differences between structural and numerical chromosome aberration. 3. Knowledge of isolation of DNA molecules from peripheral blood and measuring its concentration. 4. Knowledge of polymerase chain reaction and determination of the product on gel electrophoresis.
Metode poučevanja in učenja:		Learning and teaching methods:
Predavanja : 80%. Laboratorijske vaje: 20%.		Lectures : 80% Coursework : 20%
Načini ocenjevanja:	Delež (v %) / Weight (in %)	Assessment:
Seminarji Kolokvij iz vaj. Pisni izpit. (opravljen naloge pri vajah so pogoj za pristop k pisnemu izpitu)	10 % 20 % 70 %	Seminary work Coursework examination Written examination (Coursework project is condition for approach to written examination)