Univerza v Mariboru

Fakulteta za naravoslovje in matematiko

UČNI NAČRT PREDMETA / COURSE SYLLABUS	
Predmet:	**HUMANA GENETIKA**
Course title:	**HUMAN GENETICS**

Študijski program in stopnja Study programme and level	Študijska smer Study field	Letnik Academic year	Semester Semester
Enovit magistrski študijski program druge stopnje Predmetni učitelj	/	3	5
Five-year master's degree program Subject Teacher	/		

Vrsta predmeta / Course type Obvezni / Obligatory

Univerzitetna koda predmeta / University course code:

Predavanja Lectures	Seminar Seminar	Vaje Tutorial	Lab. vaje Laboratory work	Terenske vaje Field work	Samost. delo Individ. work	ECTS
15	15		15		45	3

Nosilec predmeta / Lecturer: Prof.dr.Nadja Kokalj Vokač

Jeziki / Languages:	
Predavanja / Lectures:	slovenski / slovene
Vaje / Tutorial:	slovenski / slovene

Pogoji za vključitev v delo oz. za opravljanje študijskih obveznosti:	Prerequisits:
Jih ni.	None

Vsebina:	Content (Syllabus outline):
2. Struktura, morfologija in klasifikacija humanih kromosomov. 3. Osnove citogenetike, molekularna citogenetika, klinična citogenetika – sindromologija. 4. Dedovanje, Mendelejevi zakoni, dominantno, recesivno dedovanje, atipični vzorci dedovanja. 5. Struktura in lastnosti DNA molekule. 6. DNA mutacije, popravljanje DNA napak. 7. Človeški genom, jederni genom, genetski kod, zgradba gena, genske družine, tandemske ponovitve, mitohondrijski genom. 8. Genetska raznolikost med posamezniki in populacijami, polimorfizmi, genetsko neravnostežje. 9. Kompleksne bolezni, multifaktorsko dedovanje.	2. Structure, morphology and classification of human chromosomes. 3. The principles of cytogenetics, molecular cytogenetics, clinical cytogenetics (syndromology). 4. Inheritance, Mendelien laws, dominant, recessive single-gene inheritance, atypical patterns of inheritance. 5. Structure and features of DNA molecule. 6. DNA mutations, repairing mechanisms. 7. Human genome, nuclear genome, genetic code, structure of the gene, gene families, tandem repeats, mitochondrial genome. 8. Genetic diversity between individuals and in populations, polymorphisms, linkage disequilibrium. 9. Complex diseases, multifactorial inheritance.

10. Somatske genetske bolezni: Citogenetika in molekularna genetika raka. 11. Izbrani primeri genetskih bolezni. 12. Genetsko svetovanje, prenatalna diagnostika, preimpantacijska genetika, genska terapija. 13. Genetsko testiranje v medicini. 14. Preprečevanje in zdravljenje gentskih bolezni. 15. Etična vprašanja genetike v medicini.	10. Somatic genetic diseases: Cytogenetics and molecular genetic of cancer. 11. Genetic diseases – selected cases. 12. Genetic counseling, prenatal diagnosis, preimplantation genetics, gene therapy. 13.Genetic testing in medicine. 14. Genetic disease prevention and treatment 15. Etical issues in medical genetics.

Temeljni literatura in viri / Readings:

1. Thompson & Thompson : Genetics in Medicine, W.B.Saunders Company., 6the ISBN 0-7216-0244-4 and 7the ed. ISBN: 9781416030805, 2007.
2. Borut Peterlin, Karin Writzl: Humana genetika, Cankarjeva založba, 1 izdaja, 1 natis, ISBN 961-231-351-2, 2003.
3. Vogel, Motulsky, Human Genetcs, Springer. 4rd ed. ISBN 978-3-540-37653-8, 2009.

Cilji in kompetence:	**Objectives and competences:**
Predmet študenta seznani z možnimi načini analize genoma in ugotavljanja genetskih napak. Študenta seznani z zakonitostmi monogenskega dedovanja in boleznimi vezanimi z napakami posamičnih genov. Študenta seznani z vlogo genetskih faktorjev pri vzrokih humanih bolezni ter prispevku k multifaktorskim boleznim, 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 courses offer to students the knowladge of major techniques used for genom analysis, and mutation analysis. The student is introduced to Mendelien laws, single-gene inheritance and diseases linked to monogenic disorders. The student is also 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:**
Znanje in razumevanje: Pozna pravila in vrste monogenskega dedovanja. Pozna primere bolezni, ki so posledica sprememb v enem genu. Pozna normalni človeški kariotip, mehanizme kromosomskih napak, ki se pojavljajo v humanem genomu in posledične sindrome. Ve kaj je gen, kako nastajajo mutacije, kako se popravljajo in kako jih določamo. Pozna tehnike določanja kromosomskih in genski sprememb, ki se uporabljajo v medicinski genetski diagnostiki. Razume, kaj je razlika med mutacijo in polimorfizmom ter pozna genetske variabilnosti med posamezniki in človeškimi populacijami. Pozna razloge za nastanek kompleksnih bolezni. Pozna osnovne mehanizme genetike raka. Pozna načine testiranja genetskih bolezni.	**Knowledge and understanding:** • Knowledge of Mendelien laws, single-gene inheritance and diseases linked to monogenic disorders. • Knowledge of human karyotype, mechanisms of chromosome rearrangements in human genome and the syndromes as their consequences. • Understanding of the gene, mutations, repairing mechanisms and methods of detection. • Knowledge of techniques used in medical genetic diagnostics. • Understanding the differences between mutation and polymorphism and knowledge about genetic variations in individuals and human populations. • Understanding of complex diseases. • Understanding of principal mechanisms of cancer genetics. • Knowledge of genetic tests.

Seznani se z genetskim svetovanjem in načini preprečevanja genetskih bolezni ter gensko terapijo. Seznani se z etičnimi problemi v medicinski gentiki. **Prenesljive/ključne spretnosti in drugi atributi:** 1. Pozna, kako 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. Pozna postopek verižne reakcije s polimerazo ter določitve produktov z gelsko elektroforezo.	• Understanding of genetic testing, preventing of genetic diseases and gene therapy. • Understanding the ethical problems in medical genetics. **Transferable/Key Skills and other attributes:** 1. Understanding of Mendelien laws, single-gene inheritance and diseases linked to monogenic disorders. 2. Understanding how to make karyotype according to ISCN nomenclature. 3. Knowledge of differences between structural and numerical chromosomal aberrations. 4. Knowledge of isolation of DNA molecules from periferal blood and messuring its concentration. 5. Understanding of polymerase chain reaction and determination of the product on gel electroforesis.
Metode poučevanja in učenja:	**Learning and teaching methods:**
• Predavanja: 33,3% • Laboratorijske vaje : 33,3% • Individualno delo: 33,3%	• Lectures • Laboratory excersises • Individual work

Načini ocenjevanja:	Delež (v %) / Weight (in %)	**Assessment:**
• Ocena iz laboratorijskega dela in zaključni kolokvij. • Ocena iz seminarske naloge. • Pisni izpit	10% 20% 70%	• Assessment of laboratory work • Assessment of seminary work. • Written exam

Reference nosilca / Lecturer's references:

1. KRGOVIC, Danijela, MARCUN-V ARDA, Natasa, ZAGORAC, Andreja, KOKALJ-VOKAC, Nadja. Submicroscopic interstitial deletion of chromosome 11q22.3 in a girl with mild mental retardation and facial dysmorphism: Case report. *Molecular cytogenetics.* [Online ed.], 2011, [Vol.] 4, 17. http://www.molecularcytogenetics.org/content/pdf/1755-8166-4-17.pdf.doi: doi:lO.1186/1755-8166-4-17. [COBISS.SI-ID 4029247]
2. ZTAKAC, Iztok, ARKO, Darja, KODRIC, Tatjana, POLJAK, Mario, ZAGORAC, Andreja, ERJAVEC SKERGET, Alenka, KOKALJ-VOKAC, Nadja. Human telomerase gene amplification and high-risk human papillomavirus infection in womeu with cervical intra-epithelial neoplasia. *J. into med. res.,* 2009, vol. 37, no. 5, str. 1588-1595. http://www.jimronline.net/content/fuI1l2009/93/1251.pdf.[COBISS.SI-ID 3455295]
3. ERJAVIC, Katja, ZAGRADISNIK, Boris, STANGLER HERODEZ, Spela, LOKAR, Lidija, GLASER, Marjana, KOKALJ-VOKAC, Nadja. Is the JAK2 . V617F mutation a hallmark for a different forms of thrombosis? *Acta Haematol.,* 2010, vol. 124, no. 1, str. 49-56, doi: 10.1159/000314645. [COBISS.SI-ID 3691839]
4. KOKALJ-VOKAC, Nadja, KODRIC, Tatjana, ERJAVEC SKERGET, Alenka, ZAGORAC, Andreja, TAKAC, Iztok. Screening ofTERC gene amplification as an additional genetic diagnostic test in detection of cervical preneoplastic lesions. *Cancer genet. cytogenet ..* [Print ed.], 2009, vol. 195, no. 1, str. 19-22. [COBISS.SIID 3500607]
4. KOKALJ-VOKAC, Nadja, ZAGORAC, Andreja, ERJAVEC SKERGET, Alenka, ROSKAR, Ziatko, PODGORNIK, Helena, CERNELC, Peter. Der(I;16) (ql0;pl0) in acute myeloid leukemia: the first female case described. *Acta . haematol. (Online),* 2008, letn. 119, st. 1, str. 54-56, doi: 10.1159/000115965. [COBISS.SI-ID 24117977]
5. TAKAC, IZlok, ARKO, Darja, KODRlC, Taljana, POLJAK, Mario, ZAGORAC, Andreja, ERJAVEC SKERGET, Alenka, KOKALJ-VOKAC, Nadja. Human lelomerase gene amplification and high·risk human papiUomavirus infection in women with cervical intraepilhelial neoplasia. *J. into med. res.,* 2009, vol. 37, no. 5, sIr. 1588-1595. http://www.jimronline.neticonlentifull/2009/93/125I.pdf.[COBISS.SI-ID 3455295]