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 Predmetni učitelj, 2. stopnja		3.	5.
Unified master's study program »Subject Teacher«, 2nd cycle		3.	5.

Vrsta predmeta / Course type Obvezni / Obligatory

Univerzitetna koda predmeta / University course code:

Predavanja **Lectures**	**Seminar** **Seminar**	**Vaje** **Tutorial**	**Klinične vaje** **work**	**Druge oblike študija**	**Samost. delo** **Individ. work**	**ECTS**
15	15		15			**3**

Nosilec predmeta / Lecturer: doc. dr. Danijela Krgović, univ. dipl. biokem.

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

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

Vsebina:

1. Struktura, morfologija in klasifikacija humanih kromosomov.
2. Osnove citogenetike, molekularna citogenetika, klinična citogenetika – sindromologija.
3. Dedovanje, Mendelejevi zakoni, dominantno, recesivno dedovanje, atipični vzorci dedovanja.
4. Struktura in lastnosti DNA molekule.
5. DNA mutacije, popravljanje DNA napak.
6. Človeški genom, jederni genom, genetski kod, zgradba gena, genske družine, tandemske ponovitve, mitohondrijski genom.
7. Genetska raznolikost med posamezniki in populacijami, polimorfizmi, genetsko neravnostežje.
8. Kompleksne bolezni, multifaktorsko dedovanje.
9. Somatske genetske bolezni: Citogenetika in molekularna genetika raka.
10. Izbrani primeri genetskih bolezni.
11. Genetsko svetovanje, prenatalna diagnostika, preimpantacijska genetika, genska terapija.
12. Genetsko testiranje v medicini.
13. Preprečevanje in zdravljenje gentskih bolezni.
14. Etična vprašanja genetike v medicini.

Content (Syllabus outline):

1. Structure, morphology and classification of human chromosomes.
2. The principles of cytogenetics, molecular cytogenetics, clinical cytogenetics (syndromology).
3. Inheritance, Mendelien laws, dominant, recessive single-gene inheritance, atypical patterns of inheritance.
4. Structure and features of DNA molecule.
5. DNA mutations, repairing mechanisms.
6. Human genome, nuclear genome, genetic code, structure of the gene, gene families, tandem repeats, mitochondrial genome.
7. Genetic diversity between individuals and in populations, polymorphisms, linkage disequilibrium.
8. Complex diseases, multifactorial inheritance.
9. Somatic genetic diseases: Cytogenetics and molecular genetic of cancer.
10. Genetic diseases – selected cases.
11. Genetic counseling, prenatal diagnosis, preimplantation genetics, gene therapy.
12.Genetic testing in medicine.
13. Genetic disease prevention and treatment
14. 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 , ISBN: 9781437706963 8th ed. 2016.
2. Passarge E.: Color Atlas of Genetics, 2017, ISBN: 9783132414402, 5th ed.
3. Borut Peterlin, Karin Writzl: Humana genetika, Cankarjeva založba, 1 izdaja, 1 natis, ISBN 961-231-351-2, 2003.

Cilji in kompetence:

Študenti se pri predmetu seznanijo z možnimi načini analize genoma in določevanjem genetskih napak. Pri predmetu so predstavljene zakonitosti monogenskega dedovanja in predstavljene bolezni vezane na napake v posamičnih genih. Predmet študente 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

Objectives and competences:

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.

vlogi genetike v sodobnih medicinskih tehnikah, diagnostiki in genski terapiji.	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. • Seznani se z genetskim svetovanjem in načini preprečevanja genetskih bolezni ter gensko terapijo. • Seznani se z etičnimi problemi v medicinski gentiki.	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. • Understanding of genetic testing, preventing of genetic diseases and gene therapy. • Understanding the ethical problems in medical genetics.
Prenesljive/ključne spretnosti in drugi atributi: 1. Razume zakone Mendelejevega dedovanja in prenos bolezni vezanih na monogensko dedovanje. 2. Pozna, kako sestaviti kariogram po ISCN nomenklaturi. 3. Zna ločiti strukturno od numerčne kromosomske spremembe. 4. Zna izolirati DNA molekulo iz periferne krvi in določiti njeno koncentracijo. 5. Pozna postopek verižne reakcije s polimerazo ter določitve produktov z gelsko elektroforezo	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. KRGOVIĆ, Danijela, GORENJAK, Mario, RIHAR, Nika, OPALIČ, Iva, STANGLER HERODEŽ, Špela, GREGORIČ KUMPERŠČAK, Hojka, DOVČ, Peter, KOKALJ-VOKAČ, Nadja. Impaired neurodevelopmental genes in Slovenian autistic children elucidate the comorbidity of autism with other developmental disorders. Frontiers in molecular neuroscience. Jun. 2022, vol. 15, str. 1-17, ilustr.
2. GREGORIČ KUMPERŠČAK, Hojka, KRGOVIĆ, Danijela, DROBNIČ RADOBULJAC, Maja, ŠENICA, Nina, ZAGORAC, Andreja, KOKALJ-VOKAČ, Nadja. CNVs and chromosomal aneuploidy in patients with early-onset schizophrenia and bipolar disorder : genotype-phenotype associations. Frontiers in psychiatry. 12. Jan. 2021, vol. 11, str. 1-16, ilustr. ISSN 1664-0640.
3. STANGLER HERODEŽ, Špela, MARČUN-VARDA, Nataša, KOKALJ-VOKAČ, Nadja, KRGOVIĆ, Danijela. De novo KMT2D heterozygous frameshift deletion in a newborn with a congenital heart anomaly. Balkan journal of medical genetics. 2020, vol. 23, issue 1, str. 83-90, ilustr. ISSN 2199-5761.
4. 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. 2018, vol. 39, iss. 11, str. 1650-1659, ilustr. ISSN 1098-1004.
5. ZAGRADIŠNIK, Boris, KRGOVIĆ, Danijela, STANGLER HERODEŽ, Špela, ZAGORAC, Andreja, ČIZMAREVIČ, Bogdan, KOKALJ-VOKAČ, Nadja. Identification of genomic copy number variations associated with specific clinical features of head and neck cancer. Molecular cytogenetics. [Online ed.]. 2018, vol. 11, str. [1]-9. ISSN 1755-8166.
6. KRGOVIĆ, Danijela, KOKALJ-VOKAČ, Nadja, ZAGORAC, Andreja, GREGORIČ KUMPERŠČAK, Hojka. Rare structural variants in the DOCK8 gene identified in a cohort of 439 patients with neurodevelopmental disorders. Scientific reports. 21. 6. 2018, [vol.] 8, str. 1-7. ISSN 2045-2322.
7. GREGORIČ KUMPERŠČAK, Hojka, KRGOVIĆ, Danijela, KOKALJ-VOKAČ, Nadja. Specific behavioural phenotype and secondary cognitive decline as a result of an 8.6 Mb deletion of 2q32.2q33.1. JIMR on-line. 2016, vol. 44, issue 2, str. 395-402, ilustr. ISSN 1473-2300.
8. KRGOVIĆ, Danijela, BLATNIK, Ana, BURMAS, Ante, ZAGORAC, Andreja, KOKALJ-VOKAČ, Nadja. A

coalescence of two syndromes in a girl with terminal deletion and inverted duplication of chromosome 5. BMC medical genetics. 2014, [vol.] 15, str. 1-9, ilustr. ISSN 1471-2350.
9. 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. [Print ed.]. 2013, vol. 524, no. 2, str. 386-389, ilustr. ISSN 0378-1119.
10. VERGULT, Sarah, KRGOVIĆ, Danijela, LOEYS, Bart, LYONNET, Stanislas, LIEDÉN, Agne, ANDERLID, Britt-Marie, SHARKEY, Freddie, JOSS, Shelagh, MORTIER, Geert, MENTEN, Björn. Nasal speech and hypothyroidism are common hallmarks of 12q15 microdeletions. EJHG. European journal of human genetics. 2011, vol. 19, [no. 10], str. 1032-1037. ISSN 1018-4813.
11. 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. [Online ed.]. 2011, [vol.] 4, 17. ISSN 1755-8166.
12. 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. Jul. 2016, vol. 90, issue 1, str. 99-101, ilustr. ISSN 1399-0004.