



Univerza v Mariboru

Fakulteta za naravoslovje
in matematiko

UČNI NAČRT PREDMETA / COURSE SYLLABUS

Predmet:	HUMANA GENETIKA
Course title:	HUMANE 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:

Prof.dr.Nadja Kokalj Vokač

**Jeziki /
Languages:**

**Predavanja /
Lectures:**

slovenski / Slovenian

Vaje / Tutorial:

slovenski / Slovenian

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

Jih ni.

Prerequisites:

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, jedrni genom, genetski kod, zgradba gena, genske družine, tandemske ponovitve, mitohondrijski genom.
7. Genetska raznolikost med posamezniki in populacijami, polimorfizmi, genetsko neravnostezje.
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 genetskih 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, Mendelian 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. Ethical issues in medical genetics.

Temeljni literatura in viri / Readings:

1. Thompson & Thompson : Genetics in Medicine, W.B.Saunders Company., 6th ISBN 0-7216-0244-4 and 7th ed. ISBN: 9781416030805, 2007 , ISBN: 9781437706963 8th ed. 2016.
2. Passarge E.: Color Atlas of Genetics, 2017, ISBN: 9783132414402, 5th ed.
2. Borut Peterlin, Karin Writzl: Humana genetika, Cankarjeva založba, 1 izdaja, 1 natis, ISBN 961-231-351-2, 2003.

Cilji in kompetence:

Predmet študenta seznaniti z možnimi načini analize genoma in ugotavljanja genetskih napak. Študenta seznaniti z zakonitostmi monogenskega dedovanja in boleznimi vezanimi z napakami posamičnih genov. Študenta seznaniti z vlogo genetskih faktorjev pri vzrokih humanih bolezni ter prispevku k

Objectives and competences:

The courses offer to students the knowledge of major techniques used for genom analysis, and mutation analysis. The student is introduced to Mendelian 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 hereditary

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.

disorders, the contribution to multifactorial 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.

Predvideni študijski rezultati:

Znanje in razumevanje:

- Pozna pravila in vrste monogenega 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.
- Seznan se z genetskim svetovanjem in načini preprečevanja genetskih bolezni ter gensko terapijo.
- Seznan se z etičnimi problemi v medicinski genetiki.

Prenosljive/ključne spretnosti in drugi atributi:

1. Razume zakone Mendelejevega dedovanja in prenos bolezni vezanih na monogeno 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.

Intended learning outcomes:

Knowledge and Understanding:

- Knowledge of Mendelian 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.

Transferable/Key Skills and other attributes:

1. Understanding of Mendelian 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.

5. Pozna postopek verižne reakcije s polimerazo ter določitve produktov z gelsko elektroforezo.

4. Knowledge of isolation of DNA molecules from periferal blood and measuring its concentration.
5. Understanding of polymerase chain reaction and determination of the product on gel electroforesis.

Metode poučevanja in učenja:

- Predavanja: 33,3%
- Laboratorijske vaje : 33,3%
- Individualno delo: 33,3%

Learning and teaching methods:

- Lectures
- Laboratory excersises
- Individual work

Delež (v %) /

Načini ocenjevanja:

Weight (in %)

Assessment:

<ul style="list-style-type: none"> • Ocena iz laboratorijskega dela in zaključni kolokvij. • Ocena iz seminarske naloge. • Pisni izpit 	<p>10%</p> <p>20%</p> <p>70%</p>	<ul style="list-style-type: none"> • Assessment of laboratory work • Assessment of seminary work. • Written exam
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Reference nosilca / Lecturer's references:

1. 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, ISSN 0009-9163, 2016, vol. , no. , str. [1-3], ilustr. <http://onlinelibrary.wiley.com/doi/10.1111/cge.12725/epdf>
2. 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, ISSN 1098-1004, 2018, vol. , no. , f. 1-35. <https://onlinelibrary.wiley.com/doi/abs/10.1002/humu.23610>, <https://doi.org/10.1002/humu.23610>, doi: 10.1002/humu.23610. [COBISS.SI-ID 6435647]
3. 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, ISSN 2045-2322, 21. 6. 2018, [Vol.] 8, str. 1-7. <https://www.nature.com/articles/s41598-018-27824-0>, doi: 10.1038/s41598-018-27824-0. [COBISS.SI-ID 6377279] <https://doi.org/10.1038/s41598-018-27824-0>
4. 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, ISSN 1473-2300, 2016, vol. , no. , str. 1-8, ilustr. <http://imr.sagepub.com/content/early/2016/01/22/0300060515595651.full.pdf+html>, doi: 10.1177/0300060515595651. [COBISS.SI-ID 5609791],
5. KRSTANOSKI, Zoran, KOKALJ-VOKAČ, Nadja, ZAGORAC, Andreja, POSPIHALJ, Boris, MUNDA, Miha, DŽEROSKI, Sašo, GOLOUH, Rastko. TMRSS2:ERG gene aberrations may provide insight into pT stage in prostate cancer. BMC urology. 2016, vol. 16, str. 1-6, ilustr. ISSN 1471-2490.

<https://bmcurol.biomedcentral.com/articles/10.1186/s12894-016-0160-8>, DOI: 10.1186/s12894-016-0160-8. [COBISS.SI-ID 29626407]