

	UČNI NAČRT PREDMETA/COURSE SYLLABUS
Predmet	Humana genetika in genetski vzroki bolezni
Course title	Human Genetics and Genetic Causes of Diseases

Študijski program in stopnja Study programme and level	Študijska smer Study field	Letnik Academic year	Semester Semester
Zdravstvena nega / 2. stopnja	Ni smeri študija	2. letnik	3.
Nursing Care / 2 nd Cycle	No study field	2 nd year	3 rd

Vrsta predmeta/Course type modularni/module

Univerzitetna koda predmeta/University course code 2ZN 2 M5 UN1

Predavanja	Seminar	Sem. vaje	Lab. vaje	Teren. vaje	Samost. delo	ECTS
Lectures	Seminar	Tutorial	Laboratory work	Field work	Individ. work	
20		30			130	6

Nosilec predmeta/Lecturer: doc. dr. Nevenka Kregar Velikonja

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

Pogoji za vključitev v delo oz. za opravljanje študijskih obveznosti:	Prerequisites:
<ul style="list-style-type: none"> Vpis v drugi letnik študijskega programa. Študent mora pred izpitom pripraviti in predstaviti ter zagovarjati projektno/raziskovalno nalogo. 	<ul style="list-style-type: none"> A prerequisite for inclusion is enrolment in the second year of study. Student has to prepare, present and defend a project/research paper before the exam.

Vsebina:	Content (Syllabus outline):
<p><i>Dedna informacija v človeških celicah:</i></p> <ul style="list-style-type: none"> <i>Molekula DNK:</i> struktura in funkcija DNK, struktura in funkcija kromosomov, jedrna DNK, mitohondrijska DNK. <i>Geni:</i> struktura gena, eksoni in introni, aleli in genetska raznolikost. <i>Genom:</i> struktura genoma, kodirajoče regije proteinov in RNK, psevdogeni, repetitivne sekvence, transponibilni elementi, minisateliti in mikrosatelitne regije, polimorfizmi. <i>Mitoza:</i> podvajanje DNK pred celično delitvijo. 	<p><i>Genetic information in human cells:</i></p> <ul style="list-style-type: none"> <i>DNA molecule:</i> structure and function of DNA, chromosome structure and function, nuclear DNA, mitochondrial DNA. <i>Genes:</i> structure of genes, exons and introns, alleles and genetic diversity. <i>Genome:</i> genome structure, coding region of proteins and RNA, pseudogenes, repetitive sequences, transposable elements, minisatellites and microsatellite regions, polymorphisms.

- *Mejoza*: redukcijska delitev in nastanek spolnih celic, vezano dedovanje.
- *Izražanje genov*: transkripcija in translacija, mehanizmi regulacije izražanja genov, različno izražanje genov med celično diferenciacijo.
- *Mutacije*: genske, kromosomske, genomske; nastanek mutacij, mutageni dejavniki.

Oblike genetske nagnjenosti:

- *Monogenske lastnosti*:
 - Avtosomno dominantno dedovanje.
 - Avtosomno recesivno dedovanje.
 - Na kromosom X vezano recesivno dedovanje.
 - Na kromosom X vezano dominantno dedovanje.
 - Na kromosom Y vezano dedovanje.
 - Kodominantno dedovanje.
 - Mitohondrijsko dedovanje.
 - Posledice mutacij - monogenske bolezni.
 - Analiza rodovnikov in izračuni tveganja za dedovanje bolezni.
- *Bolezni, ki so posledica spremembe v številu in strukturi kromosomov.*
- *Multifaktorsko pogojene lastnosti*: tveganje za razvoj bolezni; interakcija genskih in okoljskih dejavnikov.
- *Somatske genetske bolezni*: kancerogeneza, dedne oblike rakavih bolezni, kriteriji za genetsko obremenitev z rakom v družini.
- *Netipični načini dedovanja*: mozaicizem, uniparentalna disomija, imprinting,
- *Mutacije v regijah, ki kodirajo molekule tRNA in rRNA*

Populacijska genetika in genetska raznolikost:

- *Genetski bazen in pogostost posameznih alelov.*
- *Hardy-Weinbergovo načelo.*
- *Vpliv migracij, mutacij in selekcije.*
- *Heterozigotična prednost in pogostost mutacij v populaciji*

Genomika:

- *Proučevanje delovanja genov na ravni transkripcije in translacije; interakcija*

- *Mitosis: duplication of DNA before cell division.*
- *Meiosis: formation of reproductive cells, linked inheritance.*
- *Gene expression: transcription and translation, regulation of gene expression, different expression of genes during cell differentiation.*
- *Mutations: gene, chromosomal, genomic mutations; mutagenic factors.*

Forms of genetic predisposition:

- *Monogenic traits*:
 - Autosomal dominant inheritance.
 - autosomal recessive inheritance.
 - X-linked recessive inheritance.
 - X-linked dominant inheritance.
 - Y-linked inheritance.
 - Co-dominant inheritance.
 - Mitochondrial inheritance.
 - Consequences of mutations - monogenic diseases.
 - Family history and pedigree analysis; risk analysis for inheritance of the disease.
- *Diseases resulting from a change in the number and structure of chromosomes.*
- *Multifactorial inheritance*: risk of disease development; interaction of gene and environmental factors.
- *Somatic genetic diseases*: carcinogenesis, hereditary forms of cancer, criteria for genetic load with cancer in the family.
- *Untypical ways of inheritance*: mosaicism, uniparental disomy, imprinting,
- *Mutations in regions that encode the tRNA and rRNA molecules*

Population genetics and genetic diversity:

- *Genetic pool and allele frequency.*
- *Hardy-Weinberg Equilibrium.*
- *The impact of migration, mutations and selection.*
- *Heterozygous advantage and frequency of mutations in the population.*

<p><i>delovanja genov in proteinov, interakcija proteinov.</i></p> <ul style="list-style-type: none"> • <i>Personalizirana medicina.</i> <p><i>Seminarske vaje:</i></p> <ul style="list-style-type: none"> • <i>Praktični primeri za podkrepitev in razumevanje vsebine predavanj – izdelava portfolia.</i> • <i>Predstavitve primerov dednih bolezni.</i> 	<p><i>Genomics:</i></p> <ul style="list-style-type: none"> • <i>The field exploring gene transcription and translation as well as interaction that occur between genes and proteins, and in protein-protein interactions.</i> • <i>Personalised medicine.</i> <p><i>Seminar tutorials:</i></p> <ul style="list-style-type: none"> • <i>Practical examples for supporting and understanding the content of lectures - creating a portfolio.</i> • <i>Presentation of cases of hereditary diseases.</i>
---	---

Temeljna literatura in viri/Readings:

Temeljna literatura/Basic literature

- Peterlin, B. in Writzl, K. (2003). *Humana genetika*. Ljubljana, Cankarjeva Založba.
- Schaefer, G.B., Thompson, J.N. (2014). *Medical genetics: an integrated approach*. New York: McGraw-Hill Education
- Stušek, P. in Vilhar, B. (2011). *Biologija celice in genetika*. Ljubljana: DZS. Izbrana poglavja.

Priporočljiva literatura/Recommended literature

- Wilson G.N. (2000). *Clinical genetics: a short course*. New York, Willey-Liss Inc.
- Jorde, L.B., Carey, J.C., Bamshad, M.J. (2010). *Medical genetics*. Philadelphia: Mosby Elsevier.

Cilji in kompetence:

Učna enota prispeva predvsem k razvoju naslednjih splošnih in specifičnih kompetenc:

- celovito kritično razmišljanje, sposobnost analize, sinteze in predvidevanja rešitev s področij zdravstvenih ved, zdravstvene nege in medicine; družboslovnih in humanističnih ved, naravoslovno-matematičnih ter drugih ved (interdisciplinarnost),
- obvladovanje raziskovalnih metod, postopkov, procesov in tehnologije,
- sposobnost kreativne uporabe znanja pri delu v kliničnem okolju,
- sposobnost reševanja kompleksnih problemov v kliničnem okolju z multidisciplinarnim pristopom: analiza situacije, načrtovanje ukrepov, izvedba načrta, vrednotenje in samovrednotenje,

Objectives and competences:

The learning unit mainly contributes to the development of the following general and specific competences:

- comprehensive critical thinking, the ability to analyse, synthesise and predict solutions in the field of nursing care, humanities, educational, social, organisational, natural mathematical and other sciences (interdisciplinarity),
- mastering research methods, procedures, processes and technology,
- the ability to creatively use knowledge in the clinical environment,
- the ability to solve complex issues in the clinical environment using the multidisciplinary approach: analysis of the situation, planning the measures, implementing the plan, evaluating and self-evaluating,
- professional communication with experts from other scientific fields and

<ul style="list-style-type: none"> • profesionalna komunikacija s strokovnjaki drugih znanstvenih področij in usposobljenost za delovanje v medpoklicnih timih, • vsestransko in sistematično obravnavo pacienta glede na relevantne fizične, psihične, socialne, kulturne, duhovne in družbene dejavnike, • varovanje pacientovega dostojanstva, zasebnosti in zaupnosti podatkov, • uvajanje in izvajanje sodobnih metod dela v zdravstveni negi in sistemu zdravstvenega varstva, • razumevanje in prepoznavanje značilnosti dednih bolezni ter obvladovanje postopkov obravnave pacientov v procesu genetskega svetovanja. 	<ul style="list-style-type: none"> • qualification to work in interprofessional teams, • comprehensive and systematic treatment of the patient with regard to relevant physical, psychological, social, cultural, spiritual and social factors, • protection of patients' dignity, privacy and data confidentiality, • introduction and implementation of contemporary work methods in nursing care and the healthcare system, • understanding and identifying the characteristics of inherited diseases as well as the management of patients in process of genetic counselling.
---	--

Predvideni študijski rezultati:

Študent/študentka:

- pozna molekularne osnove dedovanja,
- razume mehanizme izražanja genov,
- pozna načine dedovanja,
- pozna različne mutacije in razume vpliv mutagenih dejavnikov,
- pozna terminologijo, ki se uporablja v humani genetiki,
- pozna osnovne metode, ki se uporabljajo v genetski diagnostiki,
- razume pomen populacijske genetike,
- razume pomen genomike,
- se usposobi za izdelavo rodovnikov in pripravo družinske anamneze,
- se usposobi za obravnavo pacientov v procesu genetskega svetovanja.

Intended learning outcomes:

Students:

- know the molecular basis of inheritance,
- understand the mechanisms of expression of genes,
- know the ways of inheritance,
- know various mutations and understand the influence of mutagenic factors,
- know the terminology used in human genetics,
- know the basic methods used in genetic diagnostics,
- recognise the importance of population genetics,
- recognise the importance of genomics,
- develop skills for preparation of pedigrees and family history,
- develop skills to deal with patients in the genetic counselling process.

Metode poučevanja in učenja:

- *predavanja* z aktivno udeležbo študentov (razlaga, diskusija, vprašanja, primeri, reševanje problemov),
- *seminarske vaje:* priprava, predstavitev in uspešen zagovor projektne/raziskovalne naloge,
- *konzultacije.*

Learning and teaching methods:

- *lectures* with active student participation (explanation, discussion, questions, examples, problem solving);
- *tutorial:* preparation, presentation and a successful defence of a project/research paper,
- *consultations.*

Načini ocenjevanja:	Delež (v %) Weight (in %)	Assessment:
<p>Načini:</p> <ul style="list-style-type: none"> • 100 % udeležba na predavanjih in vajah: priprava, predstavitev in zagovor projektne/raziskovalne naloge – 100 % ocene; • če študent ni 100 % udeležen na predavanjih in vajah: <ul style="list-style-type: none"> - izpit – 60 % ocene, - priprava, predstavitev in zagovor projektne/raziskovalne naloge – 40 % ocene. <p>Ocenjevalna lestvica: ECTS.</p>	<p>100 %</p> <p>ali / or</p> <p>60 %</p> <p>40 %</p>	<p>Types:</p> <ul style="list-style-type: none"> • 100% attendance at lectures and tutorials: preparation, presentation and defence of project/research paper – 100 % of the grade; • if the students' attendance at lectures and tutorials is not 100%: <ul style="list-style-type: none"> - exam - 60% of the grade, - preparation, presentation and defense of the project/research paper – 40% of the grade. <p>Grading scheme: ECTS.</p>