	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 / 2nd Cycle	No study field	2nd year	3rd

Vrsta predmeta/Course type	modularni/module
Univerzitetna koda predmeta/University course code	2ZN 2 M5 UN1

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

Nosilec predmeta/Lecturer:	izr. prof. 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:**
• Vpis v drugi letnik študijskega programa. • Študent mora pred izpitom pripraviti in predstaviti ter zagovarjati projektno/raziskovalno nalogo.	• 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):**
Dedna informacija v človeških celicah: • *Molekula DNK:* struktura in funkcija DNK, struktura in funkcija kromosomov, jedrna DNK, mitohondrijska DNK. • *Geni:* struktura gena, eksoni in introni, aleli in genetska raznolikost. • *Genom:* struktura genoma, kodirajoče regije proteinov in RNK, psevdogeni, repetitivne sekence, transponibilni elementi, minisateliti in mikrosatelitne regije, polimorfizmi. • *Mitoza:* podvajanje DNK pred celično delitvijo.	*Genetic information in human cells:* • *DNA molecule: structure and function of DNA, chromosome structure and function, nuclear DNA, mitochondrial DNA.* • *Genes: structure of genes, exons and introns, alleles and genetic diversity.* • *Genome: genome structure, coding region of proteins and RNA, pseudogenes, repetitive sequences, transposable elements, minisattiles 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 dismay, 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.*

delovanja genov in proteinov, interakcija proteinov. • *Personalizirana medicina.* *Seminarske vaje:* • *Praktični primeri za podkrepitev in razumevanje vsebine predavanj – izdelava portfolia.* • *Predstavitve primerov dednih bolezni.*	*Genomics:* • *The field exploring gene transcription and translation as well as interaction that occur between genes and proteins, and in protein-protein interactions.* • *Personalised medicine.* *Seminar tutorials:* • *Practical examples for supporting and understanding the content of lectures - creating a portfolio.* • *Presentation of cases of hereditary diseases.*

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:	**Objectives and competences:**
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,	*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

• 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.	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:	Intended learning outcomes:
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.	*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:	Learning and teaching methods:
• *predavanja* z aktivno udeležbo študentov (razlaga, diskusija, vprašanja, primeri, reševanje problemov), • *seminarske vaje:* priprava, predstavitev in uspešen zagovor projektne/raziskovalne naloge, • *konzultacije.*	• *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:**
Načini: • izpit • izdelava, predstavitev in zagovor projektne/raziskovalne naloge Ocenjevalna lestvica: ECTS.	 50 % 50 %	Types: • exam • preparation, presentation and defence of the project/research paper Grading scheme: ECTS.