

	UČNI NAČRT PREDMETA/COURSE SYLLABUS
Predmet	Genetsko testiranje
Course title	Genetic Testing

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 UN3

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. Alenka Hodžić

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.
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Vsebina:

Content (Syllabus outline):

<ul style="list-style-type: none"> <i>Značilnosti genetskih testov:</i> osnovne karakteristike genetskih testov, analitična veljavnost, klinična veljavnost, klinična uporabnost, etična, socialna in pravna vprašanja. <i>Vrste genetskih testov (pristopi in metode genetskega testiranja in dela z vzorci):</i> <ul style="list-style-type: none"> simptomatsko, presimptomatsko in presejalno genetsko testiranje, citogenetika, molekularna genetika, molekularna citogenetika, sekvenciranje nove generacije, 	<ul style="list-style-type: none"> <i>Characteristics of genetic tests:</i> basic characteristics of genetic tests, analytical validity, clinical validity, clinical utility, ethical, legal and social issues. <i>Types of genetic testing (approaches and methods in genetic testing):</i> <ul style="list-style-type: none"> symptomatic, presymptomatic and screening genetics tests, cytogenetics, molecular genetics, molecular cytogenetics, next generation sequencing, types of samples, suitability of samples for specific testing,
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<ul style="list-style-type: none"> - tipi vzorcev, ustreznost vzorcev za specifično testiranje, priprava, procesiranje in hranjenje vzorcev. • <i>Neposredni in posredni genetski testi.</i> • <i>Prenatalna diagnostika:</i> metode prenatalne diagnostike, predimplantacijska genetska diagnostika, neinvazivna genetska diagnostika. • <i>Odkrivanje genetske nagnjenosti za razvoj bolezni, presejalni testi.</i> • <i>Genetsko testiranje za nediagnostične namene:</i> ugotavljanje starševstva, forenzika, ugotavljanje histokompatibilnosti. • <i>Etični, socialni in pravni vidiki (ELSI) genetskega testiranja:</i> pravilno podajanje informacij, zakonodaja in smernice na področju genetskega testiranja. <p><i>Seminarske vaje:</i> Predstavitev diagnostike konkretnih kliničnih primerov (citogenetska diagnostika, molekularno-genetska diagnostika, nova generacija sekvenciranja); interpretacija rezultatov diagnostičnih metod.</p>	<p>preparation, processing and storage of samples.</p> <ul style="list-style-type: none"> • <i>Direct and indirect genetic tests.</i> • <i>Prenatal diagnostics:</i> methods of prenatal diagnosis, pre-implantation genetic diagnostics, non-invasive genetic diagnostics. • <i>Detection of genetic predisposition for disease development, screening tests.</i> • <i>Genetic testing for non-diagnostic purposes:</i> paternity testing, forensics, determination of histocompatibility. • <i>Ethical, legal and social issues (ELSI) in genetic testing:</i> properly providing information, laws and recommendations for genetic testing. <p><i>Seminar tutorials:</i> Presentation of genetic diagnostics in examples of clinical cases (cytogenetic diagnostics, molecular genetic diagnostics, new generation sequencing); interpretation of the diagnostic methods results.</p>
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Temeljna literatura in viri/Readings:

Temeljna literatura/Basic literature

- Peterlin, B. in Writzl, K. (2003) *Humana genetika*. Ljubljana: Cankarjeva založba.
- Peterlin, B. (2014) *Genetika v ginekologiji*. Ljubljana: Univerzitetni klinični center, Klinični inštitut za medicinsko genetiko, Ginekološka klinika.

Priporočljiva literatura/Recommended literature

- Katsanis, S. H., & Katsanis, N. (2013). Molecular genetic testing and the future of clinical genomics. *Nature Reviews. Genetics*, 14(6), 415–426. <http://doi.org/10.1038/nrg3493>
- European Society of Human Genetics Center for Biomedical Ethics and Law - BOX 7001, KU Leuven, Kapucijnenvoer 35/3, Leuven 3000, Belgium. E-mail: Pascal.Borry@med.kuleuven.be. (2010). Statement of the ESHG on direct-to-consumer genetic testing for health-related purposes. *European Journal of Human Genetics*, 18(12), 1271–1273. <http://doi.org/10.1038/ejhg.2010.129>.

Cilji in kompetence:

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

- usposobljenost za prepoznavanje potreb po zdravstveni negi, določitev negovalnih diagnoz, načrtovanje in izvajanje kakovostne in varne zdravstvene nege ter vrednotenje doseženih ciljev,
- avtonomnost pri sprejemanju odločitev v procesu zdravstvene nege,
- usposobljenost za komuniciranje v domačem in mednarodnem okolju,
- zavezanost profesionalni etiki, sposobnost etičnega odločanja in ravnanja v primeru etičnih dilem v zdravstveni negi,
- elaboriranje in predstavitev individualno ali timsko pridobljenih dognanj,
- vsestransko in sistematično obravnavo pacienta glede na relevantne fizične, psihične, socialne, kulturne, duhovne in družbene dejavnike,
- aktivno promoviranje zdravja, ocenjevanje tveganja za nastanek bolezni in skrb za varnost ter zdravje ljudi,
- varovanje pacientovega dostojanstva, zasebnosti in zaupnosti podatkov,
- primerno komuniciranje v negovalnem, zdravstvenem, multidisciplinarnem timu, razvoj in vzdrževanje profesionalnih medsebojnih odnosov med zaposlenimi, pacienti, njihovimi družinami, skupinami in skupnostjo,
- planiranje, organiziranje in analiza storitev zdravstvene nege.

Objectives and competences:

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

- qualification for recognising the nursing care needs, setting nursing diagnoses, planning and implementation of quality and safe nursing care, and evaluating the objectives achieved,
- autonomy in decision-making in the process of nursing care,
- the ability to communicate in the local and international environment,
- commitment to professional ethics, the ability to ethically decide and act in the event of ethical dilemmas in nursing care,
- elaboration and presentation of individual and teamwork findings,
- comprehensive and systematic treatment of the patient with regard to relevant physical, psychological, social, cultural, spiritual and social factors,
- active promotion of health, risk assessment and care for the safety and health of people,
- protection of patients' dignity, privacy and data confidentiality,
- appropriate communication in a nursing, health or multidisciplinary team, the development and maintenance of good mutual relations among employees, patients, their families, groups and the community,
- planning, organising and analysing nursing care services.

Predvideni študijski rezultati:

Študent/študentka:

- pozna osnove pristopov testiranja v genetskih laboratorijih in praktično uporabo v klinični praksi,
- razume značilnosti, specifičnosti in pomen genetskega testiranja v klinični praksi, pa tudi etične, legalne in socialne implikacije genetskega testiranja,

Intended learning outcomes:

Students:

- know the basics of testing approaches in genetic laboratories and practical use in clinical practice,
- recognise the characteristics, specificity and importance of genetic testing in clinical practice, as well as the ethical, legal and social implications of genetic testing,

<ul style="list-style-type: none"> • razvije kritično presojo o medicinski uporabnosti genetskih testov, • se usposobi za osnovno interpretacijo rezultatov genetskih testov in kritično presojo o medicinski uporabnosti genetskih testov. 	<ul style="list-style-type: none"> • recognition of critical judgment on the medical relevance of genetic tests, • develop skills for basic interpretation of the results of genetic tests and a critical assessment of the medical applicability of genetic tests.
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Metode poučevanja in učenja:

Learning and teaching methods:

<ul style="list-style-type: none"> • <i>predavanja</i> z aktivno udeležbo študentov (razlaga, diskusija, vprašanja, primeri, reševanje problemov), • <i>seminarske vaje</i>: priprava, predstavitev in uspešen zagovor projektne/raziskovalne naloge, • <i>konzultacije</i>. 	<ul style="list-style-type: none"> • <i>lectures</i> with active student participation (explanation, discussion, questions, examples, problem solving); • <i>tutorial</i>: preparation, presentation and a successful defence of a project/research paper, • <i>consultations</i>.
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Načini ocenjevanja: Delež (v %) Weight (in %) **Assessment:**

<p>Načini:</p> <ul style="list-style-type: none"> • izpit • izdelava, predstavitev in zagovor projektne/raziskovalne naloge <p>Ocenjevalna lestvica: ECTS.</p>	<p>50 %</p> <p>50 %</p>	<p>Types:</p> <ul style="list-style-type: none"> • exam • preparation, presentation and defence of the project/research paper <p>Grading scheme: ECTS.</p>
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