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| | 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 Lectures | Sem. vaje Tutorial | Kab. vaje Cabinet tutorial | Lab. vaje Laboratory work | Teren. vaje Field work | Samost. delo Individ. work | ECTS |
|--------------------------------------|-------------------------------------|---|--|---|---|-------------|
| 30 | 30 | | | | 180 | 8 |

Nosilec predmeta/Lecturer: prof. dr. Borut Peterlin

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

| Pogoji za vključitev v delo oz. za opravljanje študijskih obveznosti: | Prerequisites: |
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| Vpis v drugi letnik študijskega programa. | The prerequisite for inclusion is enrolment in the second year of study. |

| Vsebina: | Content (Syllabus outline): |
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| <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 | <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, |

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| <p>citogenetika, sekvenciranje nove generacije,</p> <ul style="list-style-type: none"> • tipi vzorcev, ustreznost vzorcev za specifično testiranje, priprava, procesiranje in hranjenje vzorcev. <ul style="list-style-type: none"> • <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> Predstavitve diagnostike konkretnih kliničnih primerov (citogenetska diagnostika, molekularno-genetska diagnostika, nova generacija sekvenciranja); interpretacija rezultatov diagnostičnih metod.</p> | <ul style="list-style-type: none"> • types of samples, suitability of samples for specific testing, preparation, processing and storage of samples. • <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 predstavitve 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,
- razvije kritično presojo o medicinski uporabnosti genetskih testov,
- se usposobi za osnovno interpretacijo rezultatov genetskih testov in kritično

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,
- recognition of critical judgment on the medical relevance of genetic tests,

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| presajo o medicinski uporabnosti genetskih testov. | <ul style="list-style-type: none"> 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:

- *predavanja* z aktivno udeležbo študentov (razlaga, diskusija, vprašanja, primeri, reševanje problemov, e-učenje),
- *seminarske vaje*: priprava, predstavitev in uspešen zagovor projektne/raziskovalne naloge, portfolio (reševanje problemov, študije primera, kritično presojanje, diskusija, projektno delo).

Learning and teaching methods:

- *lectures* with active student participation (explanation, discussion, questions, examples, problem solving, e-learning),
- *seminar tutorial*: preparation, presentation and a successful defence of a project/research paper, portfolio (problem solving, case studies, methods of critical thinking, discussion, project work).

| 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 raziskovalne naloge – 100 % ocene; • če študent ni 100 % udeležen na predavanjih in vajah: <ul style="list-style-type: none"> - izpit – 70 % ocene, - priprava, predstavitev in zagovor raziskovalne naloge – 30 % ocene. <p>Ocenjevalna lestvica: ECTS.</p> | <p>100 %</p> <p>ali /or</p> <p>70 %</p> <p>30 %</p> | <p>Types:</p> <ul style="list-style-type: none"> • 100 % attendance at lectures and tutorials: preparation, presentation and defence of project paper – 100 % of the grade; • if the students' attendance at lectures and tutorials is not 100%: <ul style="list-style-type: none"> - exam - 70% of the grade, - preparation, presentation and defense of the research paper – 30% of the grade. <p>Grading scheme: ECTS.</p> |