

	UČNI NAČRT PREDMETA/COURSE SYLLABUS
Predmet	Medicinska genetika v klinični medicini
Course title	Medical Genetics in Clinical Medicine

Študijski program in stopnja Study programme and level	Študijska smer Study field	Letnik Academic year	Semester Semester
Zdravstvena nega / 2. stopnja	Ni smeri študija	1. / 2. letnik	2. / 3.
Nursing Care / 2 nd Cycle	No study field	1 st / 2 nd year	2 nd / 3 rd

Vrsta predmeta/Course type izbirni/elective

Univerzitetna koda predmeta/University course code 2ZN I_2 IP5

Predavanja Lectures	Sem. vaje Tutorial	Kab. vaje Cabinet tutorial	Lab. vaje Laboratory work	Teren. vaje Field work	Samost. delo Individ. work	ECTS
25	30				125	6

Nosilec predmeta/Lecturer: doc. dr. Karin Writzl

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

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

Vsebina:	Content (Syllabus outline):
<ul style="list-style-type: none"> • <i>Genetika v ginekologiji in porodništvu.</i> Genetski vzroki neplodnosti, spontanih splavov, razvojnih nepravilnosti pri plodu. • <i>Genetika v pediatriji.</i> Klinična slika pogostejših genetskih bolezni v otroški dobi, terapija genetskih bolezni, genetsko svetovanje družini. • <i>Kardiogenetika.</i> Genetski vzroki bolezni srca in ožilja, možnosti genetskih preiskav, genetsko svetovanje. • <i>Onkogenetika.</i> 	<ul style="list-style-type: none"> • <i>Genetics in gynaecology and obstetrics.</i> Genetic causes of infertility, miscarriages, foetal structural abnormalities. • <i>Genetics in paediatrics.</i> The clinical presentation of common genetic conditions in the paediatric period, therapy of genetic conditions, genetic family counselling. • <i>Cardiogenetics.</i> Inherited cardiovascular conditions, genetic testing and genetic counselling.

<p>Onkološko genetsko svetovanje in testiranje.</p> <ul style="list-style-type: none"> • Gensko zdravljenje • Komunikacija z drugimi zdravstvenimi delavci pri obravnavi preiskovancev / družine z genetsko boleznijo. <p>Multidisciplinarnost obravnave bolnikov z genetskimi boleznimi.</p> <p>Seminarske vaje: Predstavitve primerov genetskih bolezn</p>	<ul style="list-style-type: none"> • <i>Oncogenetics.</i> Genetic counselling and testing for hereditary cancer. • <i>Gene therapy</i> • <i>Communication with other health professionals in treating individuals / families with genetic diseases.</i> Multidisciplinary care of patients with genetic disorders. <p>Seminar tutorials: Presentations of genetic diseases</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. Writzl, K., Rudolf, G., Volk, M., Lovrečič, L., Maver, A. in Geršak, K. (2014) *Genetika v ginekologiji*. Ljubljana: Univerzitetni klinični center, Klinični inštitut za medicinsko genetiko, Ginekološka klinika.
- Writzl K. (2015) *Kako se deduje? Opisi načinov dedovanja na primerih nevroloških bolezn*. Univerzitetni klinični center, Klinični inštitut za medicinsko genetiko, Ginekološka klinika.

Priporočljiva literatura/Recommended literature

- Read, A. in Donnai, D. (2015). *New Clinical Genetics, 3rd edition*. Scion Publishing LTD.
- Izbrani članki iz zbornikov znanstvenih srečanj, izbrani članki iz domačih in tujih znanstvenih revij s področja klinične genetike, izbrane internetne strani.

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,

Objectives and competences:

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

- qualification for recognizing 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

<ul style="list-style-type: none"> • 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. 	<p>relevant physical, psychological, social, cultural, spiritual and social factors,</p> <ul style="list-style-type: none"> • 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, organizing and analyzing nursing care.
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Predvideni študijski rezultati:

Intended learning outcomes:

<p>Študent/študentka:</p> <ul style="list-style-type: none"> • razume genetske mehanizme in načine dedovanja pogostejših genetskih bolezni, • pozna klinični potek pogostejših genetskih bolezni, ki se pojavljajo v različnih življenjskih obdobjih (prenatalno obdobje, pediatrija, dedni raki, bolezni v odraslem obdobju). • razume psihosocialni in etični vidik genetskega testiranja in pomen multidisciplinarnega delovanja. 	<p>Students:</p> <ul style="list-style-type: none"> • recognise the importance of genetic mechanisms and inheritance patterns of common genetic disorders, • know the clinical presentation of a range of common genetic conditions that appear in different periods of life (prenatal, paediatrics, inherited cancers, cardiovascular disorders in adult life), • know the impact of psychosocial and ethical issues during genetic testing and the importance of collaborative multidisciplinary work.
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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, portfolio (reševanje problemov, študije primera, kritično presojanje, diskusija, refleksija izkušenj, vrednotenje, projektno delo, timsko delo). 	<ul style="list-style-type: none"> • <i>lectures</i> with active student participation (explanation, discussion, questions, examples, problem solving), • <i>seminar tutorial</i>: preparation, presentation and a successful defence of a project/research paper, portfolio (problem solving, case studies, methods of critical thinking, discussion, reflection of experience, evaluation, project work, team work).
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Načini ocenjevanja:	Delež (v %) Weight (in %)	Assessment:
<p>Načini:</p> <ul style="list-style-type: none"> • 100 % udeležba na predavanjih in vajah: priprava, predstavitve 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, predstavitve 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>