



**UČNI NAČRT PREDMETA / SUBJECT SPECIFICATION**

<b>Predmet:</b>	Izbirne vsebine in novosti v genetiki in genomiki medicine
<b>Subject Title:</b>	Selected topics and novelties in genetics and genomics in medicine

Študijski program Study programme	Študijska smer Study field	Letnik Year	Semester Semester
Splošna medicina General medicine - EMŠP		1	2

Univerzitetna koda predmeta / University subject code:

Predavanja Lectures	Seminar Seminar	Sem. vaje Tutorial	Lab. vaje Labor work	Teren. vaje Field work	Samost. delo Individ. work	ECTS
5	40				45	3

Nosilec predmeta / Lecturer:

Jeziki / Languages:   
 Predavanja / Lecture:  
 Vaje / Tutorial:

Pogoji za vključitev v delo oz. za opravljanje študijskih obveznosti:   
 Prerequisites:

Vsebina:

- genetika in genomika
- genomika in odkrivanje novih zdravil
- genomika raka
- asociacijske študije za odkrivanje novih genov povezanih s kompleksnimi boleznimi (avtoimunske bolezni, rak, astma, sladkorna...)
- tehnologije v genomiki: nanotehnologija, mikromreže (biočipi), tehnologija za avtomatsko gensko tipizacijo enonukleotidnih polimorfizmov v celotnem genomu (SNPov), sekvenciranje celotnih genomov, Maldi-TOF, 2-D elektroforeza
- funkcijska genomika
- transkriptomika
- farmakogenomika in toksikologija
- fiziološka genomika

Content (Syllabus outline):

- genetics and genomics
- genomics in drug discovery
- cancer genomics
- disease association study in common complex diseases (autoimmune diseases, cancer, asthma, diabetes...)
- genomic technologies: nanotechnology, microarrays (Biochips), whole genome genotyping of Single nucleotide Polymorphisms (SNPs), Maldi-TOF, 2-D electrophoresis
- functional genomics
- transcriptomics
- pharmacogenomics and toxicology
- physiological genomics
- comparative genomics
- genomics of microorganisms
- proteomics
- bioinformatics: human genome

- primerjalna (komparativna ) genomika
- mikrobna genomika
- proteomika
- bioinformatika: podatkovne zbirke v genetiki in genomiki, programska orodja (za urejanje, analizo in poravnavo nukleotidnih zaporedij, za risanje in segregacijsko analizo družinskih dreves, za statistično genetiko)
- projekt humani genom in projekt HapMap
- genetske razlike med posamezniki
- populacijska genetika: velikost in struktura populacije, naravni izbor, mutacije, genetski zdrs, genski pretok, parjenje v sorodstvu; molekularna evolucija, molekularna ura, nastanek genomov
- genetski testi v diagnostiki bolezni
- genska terapija
- etika v genomiki

- databases, bioinformatics tools (genome sequence analysis, design and segregation analysis of family tree data)
- statistical genetics and disease association analysis
  - Human genome and HapMap projects
  - Genetic diversity among individuals
  - Population genetics: size and structure of population, natural selection, mutations, genetic drift, gene flow, inbreeding, molecular evolution, molecular clocks, evolution of genomes
  - Genetics in diagnosis
  - Gene therapy
  - Ethics in genomics

#### Temeljni literatura in viri / Textbooks:

1. STRACHAN T and READ AP: Human Molecular genetics, Garland Publish, Inc., New York, 3rd ed., 2004
2. BROWN MS: Essentials of medical genomics, John Wiley&sons, Inc., Hoboken, New Jersey, 2003
3. Nussbaum RL, McInnes,RR, Huntington FW: Thompson & Thompson Genetics in Medicine., 6th Ed, Saunders Co.,Philadelphia, 2001
4. Philip Benfey,: Genomics , Prentice Hall, Inc., New Jersey , 2005
5. Liciano J. (ed.): Pharmacogenomics, The Search for Individualized Therapies, John Wiley&Sons, 2002R.J.M

#### Cilji:

Študenti bodo poglobili razumevanje načinov dedovanja, strukture in primerjave genov in genomov, genetske raznolikosti in genetskih napak povezanih z nastankom bolezni. Povdarek bo na prenosu novih znanj in dosežkov genomike, molekularne genetike in biomedicinske tehnologije v klinično prakso na področjih preprečavanja in diagnosticiranja bolezni, načrtovanju in uporabi molekularnih in bioloških zdravil ter individualiziranemu zdravljenju na osnovi genetskih testov. Študentom bodo predstavljene možnosti, prednosti, omejitve, tveganja in etični vidiki uporabe tehnologij molekularne genetike in genomike v medicinske namene. Povdarek bo tudi na interpretaciji genetskih testov in genetskem svetovanju pri monogenih in kompleksnih boleznih.

#### Predvideni študijski rezultati:

#### Objectives:

Student will deeply understand how genes works in health. and disease The focus will be on structure of genes and genomes, genetic diversity and mutation in human genome associated with disease. The focus will be on how can we transfer human genome discoveries and use of genomic technologies into clinical practice for disease prevention, diagnosis, development of novel molecular targeted biological drugs and individualized treatment based on patients genetic and gene expression profiles. Possibilities, advances, limitations, ethical issues and potential risks using genomics in biomedicine will be discussed. Examples of interpretation of genetic test and genetic counseling in monogenic and complex diseases will be discussed.

#### Intended learning outcomes:

Znanje in razumevanje:  
 Delovanje genov in celotnega genoma v zdravju in bolezni

Prenosljive/ključne spretnosti in drugi atributi:

laboratorijske metode in experimenti v biomedicini

Knowledge and Understanding:

- The role of genes and whole genome in health and disease

Transferable/Key Skills and other attributes: laboratory methods and experimental techniques in biomedicine

**Metode poučevanja in učenja:**

**Learning and teaching methods:**

- Predavanja
- Seminar

- Lectures
- seminar

**Načini ocenjevanja:**

Delež (v %) /  
 Weight (in %)

**Assessment:**

Način (pisni izpit, ustno izpraševanje, naloge, projekt) seminar Izpit	<b>40 %</b> <b>60%</b>	Type (examination, oral, coursework, project): seminar Examination
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