



OPIS PREDMETA / SUBJECT SPECIFICATION

Predmet:	FARMAKOGENOMIKA
Subject Title:	PHARMACOGENOMICS

Študijski program Study programme	Študijska smer Study field	Letnik Year	Semester Semester
BIOMEDICINSKA TEHNOLOGIJA		2	3 ali 4

Univerzitetna koda predmeta / University subject code:

Predavanja Lectures	Seminar Seminar	Sem. vaje Tutorial	Lab. vaje Lab. work	Teren. vaje Field work	Samost. delo Individ. work	ECTS
15	20		10		105	5

Nosilec predmeta / Lecturer:

Prof. dr. UROŠ POTOČNIK

Jeziki /
Languages:

Predavanja / Lecture: SLOVENSKI, ANGLEŠKI/ SLOVENIAN, ENGLISH
Vaje / Tutorial:

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

Prerequisites:

Kandidat mora doseči 300 ECTS na predhodnem študiju.

Graduate degree 300 ECTS

Vsebina:

DNA struktura in lastnosti, replikacija (prokarionti, eukarionti), rekombinacija DNA, DNA popravljalni mehanizmi, DNA mutacije
RNA struktura in lastnosti, vrste RNA mulekul in funkcije, transkripcija (prokarionti, eukarionti), postranskripcijske modifikacije
Struktura proteinov, sinteza proteinov, posttranslacijske modifikacije proteinov, zvižanje proteinov, transport proteinov
Regulacija proteinske sinteze: regulacija ekspresije genov pri prokariontih, pri bakteriofagih, pri evkariotskih organizmih (enoceličnih, multicelularnih, povezava z embrionalnim razvojem), regulacija na ravni translacije in posttranslacijska regulacija, Celični cikel, proliferacija, diferenciacija celic, apoptoza
Povezovanje celic v tkiva, komunikacija med celicami, signalne poti, receptorji, hormoni
Imunski sistem
Virusi, HIV, SARS, DNA diagnostika pri infekcijskih boleznih
Osnove dedovanja, kromosomska teorija dednosti, Mendlovo dedovanje, poligensko dedovanje
Projekt Humani genom, organizacija, struktura in funkcija genov in kromosomov (funkcionalna razporeditev), gensko mapiranje, mitohondrijski genom
Genetska raznolikost med posamezniki (individue): mutacije, polimorfizmi v DNA in v proteinih, uporaba polimorfizmov v medicinski

Contents (Syllabus outline):

DNA: structure, characteristics, replication (prokaryotes, eukaryotes), recombination, repair and mutations. RNA: structure, characteristics, types, function, transcription (prokaryotes, eukaryotes), posttranscriptional modification. Proteins: structure, synthesis, posttranslational modification, folding, transport. Regulation of protein synthesis, regulation of gene expression in prokaryotes, eukaryotes and in embryonic development, regulation of translation, posttranslational modifications. Cell cycle, proliferation, differentiation, apoptosis. Tissue organization, cell junctions, cell adhesion, cell to cell signalling, signal transduction pathways, receptors, hormones. Immune system. Viruses, HIV, SARS, DNA diagnosis of infection diseases. Chromosomal basis of heredity, Mendelian inheritance, polygenic inheritance. Human Genome Project, organization, structure and function of genes and chromosomes, gene mapping, mitochondrial genome. Genetic variation in individuals: mutations, DNA and protein polymorphisms, uses of polymorphisms in medical genetics, phenotype, genotype, allele frequency, haplotypes, haplotype blocks (HapMap project), the Hardy-Weinberg law, linkage analysis, linkage disequilibrium. Genetic variation in populations. Recombinant DNA technology, cloning.

genetiki; genska raznolikost med populacijami (populacijska genetika), fenotip, genotip, alelna frekvenca, haplotipi, haplotiski bloki (projekt HapMap), Hardy-Weinbergov zakon, analize genetske vezanost, vezavno neravnotežje (linkage disequilibrium)
 Rekombinantna DNA tehnologija, kloniranje človekovih genov
 Monogenske genetske bolezni, kompleksne genetske bolezni,
 Genetika raka: onkogeni, tumorsko zaviralni geni, dedne oblike, molekulska diagnostika in zdravljenje
 Preiskava DNA za tipizacijo tkiv in za osebno identifikacijo
 Genska tehnologija pri proizvodnji zdravil in diagnostičnih sredstev
 Gensko zdravljenje (genska terapija)
 Vloga molekularne biologije v sodobni družba: etični, sociološki in ekonomski vidiki

Genetic diseases with classical Mendelian and complex inheritance.
 Cancer genetics: oncogenes, tumour suppressor genes, hereditary cancers, molecular diagnostics and therapy.
 DNA analysis in forensics and bone marrow transplantation typing.
 Gene technology in drug discovery, production of drugs and diagnostics.
 Gene therapy.
 Molecular biology and society: ethical, social and economical issues.

Temeljni študijski viri / Textbooks:

1. B. ALBERTS et al.: *Molecular biology of the cell.*, 4th Ed., Garland Publish, Inc., New York, 2002
2. LODISH H., Baltimore D., Berk A., Zipursky S.L., Matsudaira P., Darnell J.: *Molecular Cell Biology*, 5th Ed., Scientific American Books, Freeman and Co., New York, 2004
3. Nussbaum RL, McInnes,RR, Huntington FW: *Thompson & Thompson Genetics in Medicine.*, 6th Ed, Saunders Co.,Philadelphia, 2001

Cilji:

Cilj predmeta je nuditi študentu poglobljeno znanje predvsem iz molekularnih procesov v celici, ki so povezani z nastankom bolezni pri človeku ter študenta usposobiti za raziskovalno delo na področju medicinske molekularne biologije.

Predvideni študijski rezultati:

Znanje in razumevanje:
 Študent bo znal povezati fenotipske značilnosti, predvsem bolezenska stanja z ustreznimi molekularnimi procesi.

Prenesljive/ključne spretnosti in drugi atributi:
 Študent bo znal načrtovati in izvesti lastni raziskovalni projekt in predstaviti rezultate v obliki ustne predstavitve in znanstvenega članka.

Metode poučevanja in učenja:

predavanja, seminar

Načini ocenjevanja:

Način (pisni izpit, ustno izpraševanje, naloge, projekt)
 ustni izpit, naloge

Objectives:

The students are provided with the advanced knowledge about molecular processes associated with human diseases and with the basic guidelines to conduct research projects in the field of medical molecular biology.

Intended learning outcomes:

Knowledge and Understanding:
 The student will be able to correlate phenotypes, particularly disease associated phenotypes with appropriate molecular processes.

Transferable/Key Skills and other attributes:
 The student will be able to design and conduct research project and present scientific results as oral presentation or/and scientific paper.

Learning and teaching methods:

lectures, seminar

Assessment:

Type (examination, oral, coursework, project):
 oral examination, coursework

Materialni pogoji za izvedbo predmeta :

Obveznosti študentov:

(pisni, ustni izpit, naloge, projekti)
 ustni izpit, naloge

Material conditions for subject realization

Students' commitments:

(written, oral examination, coursework, projects):
 oral examination, coursework