

**UČNI NAČRT PREDMETA / COURSE SYLLABUS**
**Predmet:** Populacijska genetika

**Course title:** Population Genetics

Študijski program in stopnja Study programme and level	Študijska smer Study field	Letnik Academic year	Semester Semester
Ekologija z naravovarstvom, 1. stopnje		3	zimski ali letni
Ecology with nature protection, 1st. degree			

**Vrsta predmeta / Course type**

Izbirni/optional

**Univerzitetna koda predmeta / University course code:**

Predavanja Lectures	Seminar Seminar	Sem. vaje Tutorial	Lab. vaje Laboratory work	Teren. vaje Field work	Samost. delo Individ. work	ECTS
30		15			135	180/6

**Nosilec predmeta / Lecturer:** Uroš POTOČNIK

**Jeziki /**
**Languages:**
**Predavanja /** Slovenski /Slovenian

**Lectures:**
**Vaje / Tutorial:** Slovenski /Slovenian

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

-Jih ni

-No

**Vsebina:**
**Content (Syllabus outline):**

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| <ul style="list-style-type: none"> <li>• Osnove molekularne genetike: DNA struktura in lastnosti, replikacija (prokarioti, eukarioti), rekombinacija DNA, popravljalni mehanizmi DNA, mehanizem nastanka mutacij DNA, organizacija, struktura in funkcija genov, struktura genoma (rastlinski, živalski in človeški), transkripcija, translacija, regulacija genske ekspresije</li> <li>• Osnove dedovanja, kromosomska teorija dednosti, Mendlovo dedovanje, poligencko dedovanje</li> <li>• Gensko mapiranje, mitohondrijski genom</li> <li>• Mutacije, polimorfizmi v DNA in v proteinih, fenotip, genotip, alelna frekvanca, haplotipi, haplotiski bloki (projekt HapMap), Hardy-Weinbergov zakon, analiza genetske vezave, vezavno neravnotežje (linkage disequilibrium)</li> <li>• Velikost in struktura populacije</li> <li>• Naravni izbor, mutacije, genetski zdrs, genski pretok, parjenje v sorodstvu</li> <li>• Molekularna evolucija, molekularna ura, nastanek genomov, genetika ogroženih vrst</li> <li>• Kvantitativna genetika</li> <li>• Genetsko testiranje posameznikov in populacije: metode genske tipizacije in določanja mutacij, genski testi v medicini (monogenske genetske bolezni, kompleksne genetske bolezni), preiskava DNA za tipizacijo tkiv in za osebno identifikacijo (forenzika)</li> <li>• Vloga molekularne in populacijske genetike v sodobni družbi: etični, sociološki in ekonomski vidiki</li> </ul> | <ul style="list-style-type: none"> <li>• Basic molecular genetics: DNA structure and characteristics, replication (prokaryotes, eukaryotes), recombination, repair and mutations, organization, structure and function of genes and chromosomes, genome structure (plant, animal, human), transcription (prokaryotes, eukaryotes), translation, regulation of gene expression</li> <li>• Chromosomal basis of heredity, Mendelian inheritance, polygenic inheritance</li> <li>• Gene mapping, mitochondrial genome</li> <li>• Mutations, polymorphisms, phenotype, genotype, allele frequency, haplotypes, haplotype blocks (HapMap project), the Hardy-Weinberg law, linkage analysis, linkage disequilibrium</li> <li>• Size and structure of population</li> <li>• Natural selection, mutations, genetic drift, gene flow, inbreeding</li> <li>• Molecular evolution, molecular clocks, how genomes evolve, conservation genetics</li> <li>• Quantitative traits</li> <li>• Gene testing in individuals and populations: mutation detection and genotyping methods, genetic testing in medicine (genetic diseases with classical Mendelian and complex inheritance), DNA analysis in forensics and bone marrow transplantation typing</li> <li>• Molecular and population genetic and society: ethical, social and economical issues</li> </ul> |
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**Temeljni literatura in viri / Readings:**

- Hedrick, P.W.2004: Genetics of Populations. Jones & Bartlett Publishers, Sudbury, Inc., 3rd ed.
- Klug, M., M. R. Cummings, 2003: Genetics: A Molecular Perspective. Pearson Education, Inc. New Jersey.
- Strachan, T., A. P. READ, 2004: Human Molecular genetics. Gerland Publish, Inc., New York, 3rd ed.

**Cilji in kompetence:**

- Študenti bodo seznanjeni z osnovnimi koncepti populacijske genetike.
- Poudarek v razumevanju genetske raznolikosti populacije in evolucijsko pomebnih genov bo na interpretaciji novih informacij, pridobljenih z modernimi pristopi molekularne genetike kot so sekvenciranje celotnih genomov in primerjalna genomika.

**Objectives and competences:**

- Students will be provided with basic population genetics principles.
- The focus will be on new molecular data including genome projects that compare population samples to identify patterns of genetic diversity and genes that have been under selection which helps to understand molecular evolution.

**Predvideni študijski rezultati:**

Znanje in razumevanje:

- Zakonitosti prenosa genetske informacije med generacijami
- Povezave med genotipom in fenotipom
- Dejavniki, ki vplivajo na frekvenco DNA polimorfizmov in genetsko raznolikost v različnih populacijah
- Vloga mutacij in genetske raznolikosti v molekularni evoluciji

Prenesljive/ključne spretnosti in drugi atributi:

- Pristopi in orodja statistične genetike

**Intended learning outcomes:**

Knowledge and Understanding:

- Principals of heredity and transfer of genetic information between generations
- Correlations genotype-phenotype
- Factors that influence frequency of DNA polymorphisms and genetic diversity in different populations
- The role of mutations and genetic diversity in evolution

Transferable/Key Skills and other attributes:

- Approaches and program tools for statistical genetics

**Metode poučevanja in učenja:**

- Predavanja
- Seminarske vaje

**Learning and teaching methods:**

- Lectures
- Tutorial

Delež (v %) /

**Načini ocenjevanja:**

Weight (in %)    **Assessment:**

• Pisni izpit	50	• Written examination
• Ustni izpit	50	• Oral examination

**Reference nosilca / Lecturer's references:**

- JOSTINS, Luke, MITROVIČ, Mitja, POTOČNIK, Uroš et al.. Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease. *Nature (Lond.)*, 2012, vol. 491, no. 7422, str. 119-124, doi: 10.1038/nature11582.
- Rivas MA et al. Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. *Nat Genet*. 2011 Oct 9;43(11):1066-73.
- Liu JZ et al. Dense genotyping of immune-related disease regions identifies nine new risk loci for primary sclerosing cholangitis. *Nat Genet*. 2013 Apr 21. doi: 10.1038/ng.2616
- BERCE, Vojko, PINTO KOZMUS, Carina, POTOČNIK, Uroš. Association among ORMDL3 gene expression, 17q21 polymorphism and response to treatment with inhaled corticosteroids in children with asthma. *Pharmacogenomics j.* , 2012 (Print), str. 1-7.
- Repnik, Katja, Potocnik, Uros. Haplotype in the IBD5 region is associated with refractory Crohn's disease in Slovenian patients and modulates expression of the SLC22A5 gene. *J Gastroenterol*, 2011 Sep;46(9):1081-91.