UČNI NAČRT PREDMETA / COURSE SYLLABUS	
Predmet:	OSNOVE GENETIKE
Course Title:	FUNDAMENTALS OF GENETICS

Študijski program in stopnja Study Programme and Level	Študijska smer Study Field	Letnik Academic Year	Semester Semester
UŠP Biokemija, 1. stopnja	**/**	**3.**	**5.**
USP Biochemistry, 1st Cycle	**/**	**3rd**	**5th**

Vrsta predmeta / Course Type: obvezni / Mandatory

Univerzitetna koda predmeta / University Course Code: BK134

Predavanja Lectures	Seminar Seminar	Vaje Tutorial	Klinične vaje Work	Druge oblike študija	Samost. delo Individual Work	ECTS
30	**/**	**25 SV + 20 LV**	**/**	**/**	**75**	**5**

Nosilec predmeta / Lecturer: doc. dr. Jernej Ogorevc / doc. dr. Jernej Ogorevc, Assistant Professor

Jeziki / Languages:

Predavanja / Lectures:	**slovenski / Slovenian**
Vaje / Tutorial:	**slovenski / Slovenian**

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

Študent oz. kandidat mora imeti predmet opredeljen kot študijsko obveznost.

Prerequisites:

The course has to be assigned to the student.

Vsebina:

PREDAVANJA
1. Uvod- zgodovina, evgenika, genetika danes
2. Osnovna in razširjena Mendelska genetika
3. Kromosomska teorija, citogenetika
4. Genetska rekombinacija, vezano dedovanje in kartiranje
5. Genske interakcije, komplementacija, penetranca, ekspresivnost
6. Osnove genomike – GWAS - študija primera
7. Epigenetika – dedovanje, učinki na izražanje genov, staranje
8. Uravnavanje genskega izražanja pri evkariontih in analitske metode za njegovo spremljanje

Content (Syllabus outline):

LECTURES
1. Introduction – history of genetics, eugenics
2. Mendelian genetics, other inheritance patterns
3. Chromosomal theory, citogenetics
4. Genetic recombination, linkage, gene mapping
5. Gene interactions, complementation, penetrance, expresivity
6. Basics of genomics – GWAS – Case study
7. Epigenetics – inheritance, effects on gene expression and ageing
8. Gene regulation in eukaryotes and analytical methods for monitoring gene expression

9. Dinamika in struktura evkariontskih genomov10. Razvojna genetika
11. Celična diferenciacija in reprogramiranje genoma
12. Populacijska in konzervacijska genetika
13. Genetika kvantitativnih lastnosti
14. Evolucijska genetika

SEMINARSKE VAJE
Osnovni principi dedovanja I, II
Spolno vezano in organelno dedovanje
Alelne interakcije
Genske interakcije
Vezani geni
Analiza genetske vezave in epigenetskega dedovanja
Populacijska genetika I, II
Kvantitativna genetika

LABORATORIJSKE VAJE

1. Analiza DNA sesalcev 1. del – izolacija, kvantifikacija, genetski označevalci
2. Analiza DNA sesalcev 2. del – analiza genetske variabilnosti; laboratorijske živali – ogled Centra
3. Analiza RNA sesalcev 1. del – izolacija, reverzna transkripcija, cDNA sinteza, analitika kvalitete RNA (RIN)
4. Analiza RNA 2. del – qPCR RT-PCR, izračuni_analiza diferencialnega izražanja
5. Karakterizacija matičnih celic / CRISPR/Cas – validacija sgRNA

9. Dynamics and structure of eukaryotic genomes
10. Developmental genetics
11. Cell differentiation and genome reprogramming
12. Population and conservation genetics
13. Quantitative genetics
14. Evolutionary genetics

CABINET TUTORIAL

1. Basic principles of heredity I, II
2. Sex-linked and organelle inheritance
3. Allele interactions
4. Gene interactions
5. Genetic linkage
6. Linkage analysis and epigenetic inheritance
7. Population genetics I, II
8. Quantitative genetics

LABORATORY PRACTICAL

1. Mammalian DNA analysis Part 1 - isolation, quantification, genetic markers
2. Mammalian DNA analysis Part 2 - genetic variability analysis; Laboratory animals - tour of the Center
3. Mammalian RNA analysis Part 1 - isolation, reverse transcription, cDNA synthesis, RNA quality analysis (RIN)
4. RNA analysis Part 2 - qPCR, calculations_analysis of differential expression
5. Stem-cell characterization / CRISPR/Cas based validation of sgRNA

Temeljna literatura in viri / Readings:

- Anthony Griffiths; John Doebley; Catherine Peichel; David A. Wassarman: Introduction to Genetic Analysis, 12th Edition, 2020
- znanstveni članki

Cilji in kompetence:

Študenti bodo razumeli, kako se kvalitativne in kvantitativne lastnosti prenašajo med generacijami, ob tem pa bodo znali analizirati različna križanja oziroma rodovnike. Poglobili bodo razumevanje konceptov naravne in umetne selekcije, delovanja različnih učinkov

Objectives and Competences:

Understanding how qualitative and quantitative traits are transmitted through generations, getting to know how experimental genetic crosses and pedigrees are analysed. They will deepen their comprehension of basic concepts of natural and artificial selection, gene actions,

genov (dominantnost, recesivnost, nad-dominanca, aditivnost), interakcije med aleli in geni, mehanizem genetske rekombinacije, epigenetskega dedovanja in sodobnih genomskih pristopov. Obvladali bodo osnovna statistična orodja v klasični, populacijski in kvantitativni genetiki in pristope h genskemu kartiranju. Hkrati bodo znali na primerih razložiti uporabnost genskih markerjev in njihove lastnosti. Razumeli bodo, kakšne so možnosti analize genoma na ravni celice in organizma, ter kako genetsko opišemo populacije. Spoznali bodo osnovne principe genetike celične diferenciacije in reprogramiranja. Vedeli bodo, zakaj je pomembno ohranjanje genskih virov in kako je to mogoče doseči. Študentje bodo kompetentni pri izbiri optimalnega genetskega modela za določeno znanstveno vprašanje.	gene interactions, mechanisms of meiotic recombination, epigenetic inheritance and current genomics approaches. Students will become competent in applying basic statistical methods in classical, population and quantitative genetic problems and understand the main characteristics of genetic and biochemical markers and their suitability for various applications. They will gain basic insights into genetics of cell differentiation and reprograming. They should develop a genome-wide understanding of underlying biochemical processes at a cell, tissue, organismal or population level. Students should be able to identify and select the most appropriate genetic model for a particular scientific question under study.

Predvideni študijski rezultati:	**Intended Learning Outcomes:**
<u>Znanje in razumevanje</u> Študentje bodo pridobili znanje različnih genetskih konceptov in razumevanja elementov determinacije fenotipa, načinov dedovanja, kvalitativne in kvantitativne genetike, epigenetike in reprogramiranja in se naučili analizirati genetska križanja in rodovnike. Pridobili bodo kompetence za samostojno uporabo sodobnih bioinformacijskih orodij za analizo dednih dejavnikov v bioloških sistemih. Osvojili bodo "genetski" način razmišljanja pri obravnavanju fenotipa in dednih bolezni ter možnosti uporabe pri svojih projektih iz ožjega področja biokemije.	<u>Knowledge and Comprehension</u> Students should be able to acquire knowledge of a wide spectrum of genetic concepts in determination of phenotype, modes of inheritance of qualitative and quantitative traits, epigenetic and reporgramming principles and learn how to analyse genetic crosses and pedigrees. Competences gained cover a comprehensive understanding of genetic concepts and development of genetic approach to thinking about the biochemical and biological questions, phenotype and inherited diseases.
<u>Uporaba</u> Uporabnost statističnih metod in modelov v biologiji. Računanje alelnih frekvenc in drugih genetskih lastnosti populacije. Analiza poskusnih križanj ali obstoječih rodovnikov za posamezne dedno prenosljive lastnosti. Integriranje klasične mendelske genetike z molekularnimi in biokemijskimi mehanizmi.	<u>Application</u> Ability to use state of the art statistical tools will enable students to analyse genetic mapping problems, use of genetic markers and how to characterise populations with genetic means. Students should be able to integrate classical genetic knowledge with modern biochemistry and molecular biology.
<u>Refleksija</u> Tudi biološki procesi (dedovanje) so podvrženi matematičnim zakonitostim. Populacije imajo	<u>Analysis</u> Students should be able to reflect how inheritance is linked to biochemical and

svoje skupne genetske lastnosti. Dedne bolezni se dedujejo predvidljivo. Za ohranjanje genetskih lastnosti ni dovolj ohranjanje biotopa. Genetske lastnosti dinamične populacije so odvisne tudi od njene velikosti. Naravna in umetna selekcija imata enako genetsko osnovo a različne cilje. Razvijanje "genetskega" načina razmišljanja pri obravnavanju biokemijskih procesov ter fenotipa ter kritično vrednotenje genetskih konceptov.	biological processes and mathematical/statistical concepts – what constitutes common genetic architecture of a population and why can we predict inheritance patterns of next generations. How natural and artificial selection have common genetic basis but different aims. Ability to reflect common grounds of biochemistry and genetics.
Prenosljive spretnosti Statistična analiza populacij in dedovanja. Predstavitve strokovnih vsebin na osnovi angleškega izvirnika; uporaba terminološkega slovarja. Razumevanje etičnih vidikov povezanih s sodobno genetiko je lahko koristno pri evalvaciji širših bioetičnih problemov v bioznanostih.	Skill-transference Ability Use of genetic and statistical analyses in problem solving. Use of foreign contemporary literature and genetic terminology to enhance understanding in other fields of life sciences. Students will also gain insight into ethical, legal and social issues surrounding modern genetics.

Metode poučevanja in učenja:	**Learning and Teaching Methods:**
Predavanja, laboratorijske in seminarske vaje; vmesna kratka preverjanja znanja (kvizi); individualno reševanje problemov doma	Lectures, laboratory and seminar practicum; short quizzes; take-home individual problem solving exercises.

Načini ocenjevanja:	Delež (v %) / Weight (in %)	**Assessment:**
Dva kolokvija ali končni pisni izpit		Two partial exams or final written exam

Reference nosilca / Lecturer's references:

- **OGOREVC, Jernej**, SIMČIČ, Mojca, ZORC, Minja, ŠKRJANC, Monika, DOVČ, Peter. TLR2 polymorphism (rs650082970) is associated with somatic cell count in goat milk. *PeerJ*, ISSN 2167-8359, 31. jul. 2019, vol. 7, str. 1-9, e-7340, ilustr. https://peerj.com/articles/7340.pdf, doi: 10.7717/peerj.7340. [COBISS.SI-ID 4274568].

- FORSTNERIČ, Vida, OVEN, Irena, **OGOREVC, Jernej**, LAINŠČEK, Duško, PRAZNIK, Arne, LEBAR, Tina, JERALA, Roman, HORVAT, Simon. CRISPRa-mediated FOXP3 gene upregulation in mammalian cells. *Cell & bioscience*, ISSN 2045-3701, 21. Nov. 2019, vol. 9, art. 93, str. 1-12, ilustr. https://cellandbioscience.biomedcentral.com/articles/10.1186/s13578-019-0357-0, doi: 10.1186/s13578-019-0357-0. [COBISS.SI-ID 4332936].

- **OGOREVC, Jernej**, DOVČ, Peter. Expression of estrogen receptor 1 and progesterone receptor in primary goat mammary epithelial cells. *Animal science journal*, ISSN 1740-0929, 2016, vol. 87, no. 12, str. 1464-1471. http://onlinelibrary.wiley.com/doi/10.1111/asj.12553/abstract;jsessionid=88B56CB81EBED8B3D29F8D35AB4186E1.f03t02, doi: 10.1111/asj.12553. [COBISS.SI-ID 3713928]