


UČNI NAČRT PREDMETA / COURSE SYLLABUS

Predmet:	Orofacialna genetika
Course title:	Orofacial Genetics

Študijski program in stopnja Study programme and cycle	Študijska smer Study option	Letnik Year of study	Semester Semester
Dentalna medicina/Dental Medicine 2. stopnja/2nd cycle		3	6.

Vrsta predmeta / Course type

Obvezni/Compulsory

Univerzitetna koda predmeta / University course code:

Predavanja Lectures	Seminar Seminar	Vaje Tutorial	Klinične vaje Clinical training	Druge oblike študija Other forms of study	Samost. delo Individual work	ECTS
5	40				45	3

Nosilec predmeta / Lecturer:

Prof. dr. Danko Bakarčič/ Danko Bakarčič, DDM, PhD, Profesor

Jeziki /
Predavanja / Lectures: slovenščina/slovene

Languages:
Vaje / Tutorial:
Pogoji za vključitev v delo oz. za opravljanje študijskih obveznosti:
Prerequisites:

 Biologija
Morfologija zob (anatomija in fiziologija ustne votline)

 Biology
Tooth morphology (anatomy and physiology of oral cavity)

Vsebina:

Zgodovina in pomen orofacialne genetike. Epidemiologija kraniofacialnih malformacij in pogostnost genetskih bolezni v populaciji. Dismorfije kraniofacialnih struktur (minor in major anomalije). Geni in kromosomi kot nosilci dednosti (normalne in abnormalne strukture). Metode v genetiki: analiza družine, študije prebivalstva, dvojčki, kromosomi, dermatoglifi. Pregled in ocena kraniofacialne regije pri kraniofacialnih dismorfijah. Genetske nepravilnosti zob: anomalije števila, oblike, velikosti in strukture zoba. Najpogostejši kromosomski sindromi, ki zajemajo orofacialne strukture (Down, Fragilni X, Klinefelterjev, Turnerjev sindrom). Ektodermalna displazija: razvrstitev, diagnosticiranje in odkrivanje heterozigotov. Presnovne motnje in kraniofacialne strukture (mukopolisaharidoze, mukolipidoze, homocistinurija, Lesch-Nyhan sindrom). Genetske motnje paradontnih struktur. Nevrokutani sindromi in orofacialne strukture. Heiloshiza in razcepi ustnice in neba in sindromi z razcepi (Rovinova sekvenca, EEC sindrom, Vander-Woudov sindrom). Genetsko svetovanje v zobozdravstvu.

Content (Syllabus outline):

History and significance of orofacial genetics. Epidemiology of craniofacial malformations and frequency of genetic diseases in the population. Dysmorphia of craniofacial structures (minor and major anomalies). Genes and chromosomes as carriers of the inheritance (normal and abnormal structures). Methods in genetics: family analysis, population studies, twins, chromosomes, dermatoglyphs. Examination and evaluation of craniofacial region in cranial dysmorphia. Genetic anomalies of the teeth: anomalies in number, shape, size and structure of tooth. The most frequent chromosomal syndromes that affects orofacial structures (Down, fragile X, Klinefelter, Turner syndrome). Ectodermal dysplasia: classification, diagnosis and detection of heterozygotes. Metabolic disorders and craniofacial structures (mucopolysaccharidosis, mucillipidosis, homocystinuria, Lesch-Nyhan syndrome). Genetic Disorders of periodontal structures. Syndromes with cleft lips and palate (Robins sequence, EEC syndrome, Vander-Woude syndrome). Genetic counseling in dental medicine.



Temeljni literatura in viri / Readings:

- D. Bakarčić, N Ivančić Jokić. Osnove prevencije karijesa i parodontnih bolesi. Split: Redak; 2013.
- Škrinjarić I. Orofacijalna genetika. Školska knjiga, Zagreb, 2006
- Jurić H ur. *Dječja dentalna medicina*. Naklada Slap, Jastrebarsko, 2015:187-211.
- Zukanović A, Gržić R. *Stomatološko liječenje medicinski kompromitiranih pacijenata*. HKDM, Zagreb, 2012:319-41.

Cilji in kompetence:

Obvladovanje znanja, potrebnega za preprečevanje, diagnozo in zdravljenje genskih motenj. Študenti se morajo naučiti identificirati genske komponente v etiologiji bolezni za diagnozo, prognozo in racionalen pristop k zdravljenju.

Objectives and competences:

Mastering the knowledge necessary for the prevention, diagnosis and treatment of genetic disorders regarding orofacial structures. Students need to learn to identify genetic components in the etiology of the diseases for diagnosis, prognosis and rational approach to treatment.

Predvideni študijski rezultati:

Znanje in razumevanje:

- Opisati zgodovino in pomen orofacialne genetike.
- Opredeliti malformacije dedne etiologije.
- Določiti sindrome.
- Definirati sindrome z orodontalnimi anomalijami.
- Opisati epidemiologijo kraniofacialnih malformacij.
- Razlikovati pogostnost genskih bolezni v populaciji.
- Opisati dismorfije kraniofacialnih struktur (manjših in glavnih anomalij),
- Razlikovati genske anomalije zob: anomalije števila, oblike, velikosti in strukture.
- Opisati in razlikovati simptomatologijo orofacialne regije pri motnjah rasti.
- Opisati in razlikovati simptomatologijo pri najpogostejših sindromih različnih genov z manifestacijami v orofacialni regiji.
- Opisati najpogostejše kromosomske sindrome, ki zajemajo orofacialne strukture (Down, Fragilni X, Klinefelterjev, Turner sindrom).
- Opisati ektoermalne displazije (razvrstitev, diagnosticiranje in odkrivanje heterozigotov).
- Opisati metabolične motnje kraniofacialne strukture (mukopolisaharidoza, mukolipidoza, homocistinurija, Lesh-Nyhanov sindrom.)
- Opisati genske motnje paradontalnih struktur
- Opisati in razlikovati cepitev ustnic, čeljusti in neba.
- Razlikovati sindrome (Rovinovo zaporedje, sindrom EEC, Vander-Woudeov sindrom).
- Razlikovati kombinacije, pri katerih se srečujemo s sindromi z orodontalnimi anomalijami.
- Opisati pregled in oceno kraniofacialne regije pri kraniofacialni dismorfijah.
- Razlikovati metode v genetiki (družinska analiza, populacijske študije, dvojčki, kromosomi, dermatoglify).
- Opraviti genetsko svetovanje v dentalni medicini.

Intended learning outcomes:

Knowledge and understanding:

- describe the history and significance of orofacial genetics
- define malformations of hereditary etiology
- define syndromes
- define syndromes with orodontal anomalies
- describe epidemiology of craniofacial malformations
- distinguish the frequency of genetic diseases in the population
- describe the dysmorphism of craniofacial structures (minor and major anomalies)
- distinguish genetic anomalies of the teeth: anomalies of the number, shape, size and structure of the teeth.
- describe and differentiate the symptomatology of the growth disorders in orofacial region
- describe and distinguish symptomatology in the most common syndromes with manifestations in the orofacial region
- describe the most frequent chromosomal syndromes that affect theoretic structures (Down, fragile X, Klinefelter, Turner syndrome).
- describe ectodermal dysplasia (classification, diagnosis and detection of heterozygous)
- describe the metabolic disorders of the craniofacial structure (mucopolysaccharidosis, mucillipidosis, homocystinuria, Lesch-Nyhan syndrome)
- describe genetic disorders of periodontal structures
- describe and distinguish differences between the cleft lips and the palate.
- distinguish syndromes (Robins sequence, EEC syndrome, Vander-Woude syndrome)
- distinguish combinations that is encountered with orodontal anomalies
- describe the examination and evaluation of the craniofacial region in craniofacial dysmorphia
- distinguish methods in genetics (family analysis, population studies, twins, chromosomes, dermatoglyphs)



	• perform counseling in dental medicine
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Metode poučevanja in učenja:

Predavanja
Seminarji

Learning and teaching methods:

Lectures
Seminars

Načini ocenjevanja:

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

Pisni izpit

Delež (v %) /**Weight (in %) /**

100 %

Assessment:

Type (examination, oral, coursework, project):

Final written examination

Reference nosilca / Lecturer's references:

1. Cicvarić O, Gržić R, Šimunović-Erpušina M, Simonić-Kocijan S, Bakarčić D, Ivančić Jokić N. Association of Masticatory Efficiency and Reduced Number of Antagonistic Contacts Due to Extraction, Changing Dentition or Malocclusion in Children. Dent. J. 2023, 11, 64.
2. Orlić E, Špalj S, Ivančić Jokić N, Bakarčić D, Cicvarić O, Gržić R. Pandemic financial stress in dental medicine in Croatia. Dent J 2023;11,9.
3. Ivančić Jokić N, Cicvarić O, Gavić L, Dujmović M, Bakarčić D. Maternal Dental Anxiety and Early Childhood Caries Development. Int J Clin Dent 2022; 15(4):685-91.
4. Ivančić Jokić N, Krištić J, Cicvarić O, Šimunović-Erpušina M, Stanfel D, Bakarčić D. Preschool teachers' knowledge and attitudes about dental trauma in Rijeka, Croatia: a cross-sectional study. J Oral Res 2021; 10(4):1-7.
5. Ivančić Jokić N, Bakarčić D, Cicvarić O, Šimunović-Erpušina M, Zukanović A, Hefler G, Nastić V. Znanja, navike i stavovi medicinskih sestara / tehničara Kliničkog bolničkog centra Rijeka o oralnom zdravlju Sestrinski glasnik 2021;26:19-23