



UČNI NAČRT PREDMETA / COURSE SYLLABUS

Ime predmeta:	Pedriatrija s klinično genetiko
Course title:	Pediatrics with clinical genetics

Študijski program in stopnja Study programme and cycle	Študijska smer Study option	Letnik Year of study	Semester Semester
Splošna medicina, enovit magistrski študijski program		Peti	10.
General medicine, Uniform master's degree study program		Fifth	10th

Vrsta predmeta (obvezni ali izbirni) /
Course type (compulsory or elective)

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
45	75	AV LV RV	90		210	14

Nosilec predmeta / Course coordinator:

Red. prof. dr. Nataša Marčun Varda (nosilka)
Izr. prof. dr. Vojko Berce (sonosilec)

Jeziki /Languages:

Predavanja / Lectures: slovenski/slovene

Vaje / Tutorial: slovenski/slovene

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

Prerequisites for enrolling in the course or for performing study obligations:

Vsebina (kratek pregled učnega načrta):

Socialna in preventivna pediatrija: Morbiditeta pri novorojenčkih, dojenčkih, predšolskih, šolskih otrocih in mladostnikih. Smrtnost pri dojenčkih, neonatalna in perinatalna mortaliteta. Kongenitalne anomalije. Zgodnja detekcija in rehabilitacija prizadetih otrok in njihovo vključevanje v družbo. Pomen humanizacije v otroških bolnišnicah in dispanzerjih. Organizacija pediatrične dispanzerske in hospitalne mreže. Preventivni ukrepi v zgodnji otroški dobi – vakcinacije, preprečevanje rahitisa, sideropenične anemije, nasveti o pravilni prehrani, sistemsko odkrivanje presnovnih motenj, hipotireoze, fenilketonurije, galaktozemije,

Content (syllabus outline):

Social and Preventive Pediatrics: Morbidity in newborns, infants, preschool, school children and adolescents. Mortality in infants, neonatal and perinatal mortality. Congenital anomalies. Early detection and rehabilitation of disabled children and their inclusion in society. Meaning of humanisation in children hospitals and outpatient clinics. Organisation of pediatric outpatient clinics and hospital net. Preventive measures in early age - immunisations, prevention of rickets, sideropenic anemia, advising on healthy diet, systematic discovery of digestive disorders, hypotireosis, phenylketonuria, galactosemia, hip displasiae,

displazije kolka, cerebralne paralize, motenj vida, sluha, slabe drže in drugih nepravilnosti skeleta.

Rast in razvoj otroka: Razvojne faze v somatskem in psihomotoričnem razvoju otroka. Vrednotenje antropometričnih podatkov kot telesne teže, višine, obsega lobanje, površine telesa in ocena govora ter sluha. Ocena kostne starosti, razvoja zob, sekundarnih spolnih znakov, opredelitev in zdravljenje nizke in visoke rasti.

Neonatologija: Novorojenec, Nedonošenec. Prenošene. Zahiranec. Ocena vitalnosti novorojenca. Hiperbilirubinemija. Hemolitične bolezni novorojenca. Izmenjalna transfuzija. Fototerapija. Reanimacija asfiktičnega novorojenca. Respiratorni distress sindrom novorojenca. Poškodbe otrok ob porodu. Hemoragična bolezen novorojenca. Perinatalna simbioza otroka in matere.

Prehrana in presnova: Dojenje. Energijske in prehranske potrebe novorojenčka, dojenčka, predšolskega, šolskega otroka. Prilagojena mleka. Sestava jedilnikov. Distrofija, Podhranjenost in debelost. Avitaminoza. Deficiti mineralov. Akutni enterokolitis dojenčka, dehidracija, toksikoza. Rehidracija in dietna prehrana pri celiakiji, fenilketonuriji, galaktozemiji. Vrojene presnovne motnje.

Genetika: Rodovnik. Autosomno recesivne, dominantne, spolno vezane dedne bolezni. Sindrom Down, Sindrom Turner, Klinefelterjev sindrom. Teratogene nokse. Genetska prognoza in nasvet. Prenatalna detekcija genetskih bolezni.

Endokrinologija: Motnje rasti. Konatalna hipotireoza, struma. Zgodnja in kasna puberteta. Diabetes insipidus. Hipoparatiroidizem. Kongenitalna adrenalna hiperplazija. Mb Addison. Mb Cushing. Feokromocitom. Hipogonadizem. Diferenciacija spola. Hipoglikemija, Diabetes mellitus. Tumorji endokrinih žlez.

Kardiologija: Prirojene srčne hibe. Motnje srčnega ritma. Vaskularne bolezni. Vzroki srčnih hib in patogeneza klinična slika in zdravljenje srčne insuficience pri otrocih.

Pulmologija: vzroki obstruktivne in restriktivne apnoe pri dojenčku. Subglotični laringitis, Bronhitis, pneumonia. Astma. Mukoviscidoza. Kronične pulmopatije.

Hematologija in onkologija: Sideropenična anemija. Megaloblastična in hemolitična anemija, Hiporegenerativna in druge anemije. Trombocitopenija. Levkemija, Limfom. Nevroblastom. Nefroblastom. Možganski tumorji in druge neoplazme v otroški dobi.

Revmatologija in imunologija: Akutna revmatska vročica. Kronični juvenilni revmatoidni artritis in najpogostejše druge kolagenoze v otroški dobi. Imunodeficienca.

cerebral palsy, vision, hearing disorders, bad posture and other skeletal abnormalities.

Growth and development of child: Developmental stages in somatic and psychomotor development of child. Evaluation of anthropometric data: body weight, height, head circumference, body surface area and assessment of speech and hearing. Assessment of body age, development of teeth, secondary sexual features, determination and treatment of short or high growth.

Neonathology: Newborn, premature. Postmature, Dysmature. Assessment of newborn's vitality: hyperbilirubinemia. Haemolytic disease of newborn. Exchange transfusion. Phototherapy. Reanimation of asphyxiated newborn. Respiratory distress syndrome in newborn. Birth trauma. Hemorrhagic disease in newborn. Perinatal symbiosis of mother and child.

Nutrition and metabolism: Breastfeeding: newborn's dieting and calorie needs of a newborn, an infant, a child and a preschool and school child. Adjusted milks. Composition of menus. Dystrophy. Malnutrition and obesity. Avitaminoses. Mineral deficit. Acute enterocolitis in infant, dehydration, toxicosis. Rehydration and diet in celiac disease, phenylketonuria, galactosemia. Congenital metabolic disorders.

Genetics: Family pedigree. Autosomal, recessive, dominant, sex linked hereditary diseases. Down's syndrome. Turner Syndrome, Klinefelter Syndrome, teratogen exposure. Genetic prognosis and counselling. Prenatal detection of genetic diseases.

Endocrinology: Growth disorders, congenital hypothyroidism, goitre. Early and late puberty. Diabetes insipidus. Hypoparathyroidism, congenital adrenal hyperplasia. Mb Addison. Mb Cushing. Feochromocytoma. Hypogonadism. Sex differentiation. Hypoglycemia, diabetes mellitus. Tumors of endocrine glands.

Cardiology: congenital heart anomalies. Heart rhythm disorders. Vascular diseases. Causes for congenital malformations and clinical picture of pathogenesis, treatment of heart insufficiency in children.

Pulmology: causes of obstructive and restrictive apnea in infant. Subglottic laryngitis, bronchitis, pneumonia. Asthma. Mucoviscidosis. Chronic pulmonary diseases.

Haematology and Oncology: sideropenic anaemia, megaloblastic and haemolytic anaemia, hyporegenerative and other anaemias. Thrombocytopenia. Leukemia. Limfom. Neuroblastoma. Brain tumors and other neoplasms in childhood.

Rheumatology and immunology: Acute rheumatic fever. Chronic juvenile rheumatoid arthritis and most often other collagenoses in childhood. Immunodeficiency.

Gastroenterologija: Bruhanje, Abdominalne bolečine pri otroku. Obstipacija. Akutna in kronična driska. Kongenitalne anomalije prebavil. Stenoza pilorusa. Ulkusna bolezen. Malabsorpcijski sindrom. Celiakija. Kronične vnetne bolezni črevesja. Megacolon. Apendicitis. Invaginacija črevesja. Hernija. Ileus. Hepatitis.

Nefrologija: Uroinfekt. Obstruktivska uropatija. Glomerulonefritis. Nefrotski sindrom. Prirojene tubulopatije. Malformacije uropoetskega trakta, spolnih žlez in spolovil.

Pedopsihiatrija: Anoreksija nervosa. Bulimija. Psihoza. Avtizem. Tentamen suicida. Depresivna stanja v otroški dobi. Šolska fobija. Spolna zloraba.

Nevrologija: Zvišan intrakranialni pritisk pri otroku. Cerebralna paraliza. Možganski tumorji. Glavobol. Metabolne in heredodegenerativne bolezni živčevja. Vročinski krči in druge konvulzije. Epilepsija. Meningitis in encefalitis. Polinevritis. Fakomatoze. Progresivna mišična distrofija. Kongenitalna anomalija živčevja.

Ostala področja pediatrije: ustne in kožne bolezni pri otrocih. Bolezni skeleta. Parazitoze. Nesreče. Intoksikacije in druga nujna stanja v pediatriji. Otroška kirurgija.

Gastroenterology: Vomiting, abdominal pain in child, Opistipation, Enterocolitis, Congenital anomalies of digestive tract. Stenosis pylorus. Ulcer disease. Malabsorption syndrome. Celiac disease. Chronic inflammatory intestinal diseases. Megacolon. Appendicitis. Intestinal invagination. Hernia. Ileus. Hepatitis.

Nefrology: Uroinfect. Obstructive uropathy. Glomerulonephritis. Nephritic syndrome. Congenital tubulopathies. Malformations of uropoetic tract, sexual glands and genitals.

Pedopsychiatry: Anorexia nervosa. Bulimia. Psychosis. Tentamen suicidii. Depressive moods in childhood. School phobia. Sexual abuse.

Neurology: hypercranial pressure in child. Cerebral palsy. Brain tumors. Headache. Metabolic and hereditary degenerative diseases of the nerves. Fever seizures and other convulsions. Epilepsy. Meningitis and encephalitis. Polyneuritis. Facomatosis. Progressive muscular dystrophy. Congenital anomaly of nerves.

Other pediatrics fields: oral and skin diseases in children. Skeletal diseases. Parasitosis. Accidents. Intoxications and other emergency conditions in paediatrics. Pediatric surgery.

Temeljni literatura in viri / Reading materials:

- Kržišnik C., Pediatrija, Ljubljana, 2014
- Mardešič D., Pediatrija, Šolska knjiga, Zagreb, 2003.
- Kliegman R., St. Geme J., Nelson Textbook of Pediatrics 21th edition, W.B. Saunders Company, Philadelphia, London, Toronto, Montreal, Sydney, Tokyo 2019
- Berhman R.E., Kliegman R. Nelson: **Essentials of Pediatrics**, 8th edition. Saunders, Philadelphia 2006.
- Srečanja pediatrov v Mariboru z mednarodno udeležbo, SB (UKC) Maribor, Maribor, [od l. 1995 dalje](#).
- Izbrana poglavja iz pediatrije, Medicinska fakultete Univerze v Ljubljani, [od l. 1991 dalje](#)

Cilji in kompetence:

Cilj tega predmeta je pripraviti študente na samostojno delo z bolnimi otroki.

Objectives and competences:

The objective of this course is to prepare students to work independently with pediatric patients.

Predvideni študijski rezultati:

Znanje in razumevanje:

Po zaključku tega predmeta bo študent sposoben samostojno pristopiti k bolnemu otroku, obvladal bo odnos z njim in starši in svojci, znal bo vzeti anamnezo, pregledati bolnega otroka, ovrednotiti diagnostične metode, predpisati ustrezno terapijo.

Prenesljive/ključne spretnosti in drugi atributi:
Študent se bo naučil tudi praktičnih opravil, ki so potrebna pri delu z bolnimi otroki:

jemanje krvi, dajanje terapije, opravljanje osnovnih funkcionalnih preiskav.

Intended learning outcomes:

Knowledge and Understanding:

On completion of this course student will be able to approach patient independently, and communicate accordingly with the patient and his family members, he will take history of illness, examine the patient, and evaluate diagnostic methods to be applied and to prescribe appropriate therapy.

Transferable/Key Skills and other attributes:

The student will also acquire necessary practical knowledge: blood taking, giving therapy, to perform basic functional examinations.

Metode poučevanja in učenja:

- predavanja,
- seminarji,
- vaje.

Learning and teaching methods:

- Lectures,
- Seminars,
- Practical training in the clinical environment

Delež (v %) /
Share (in %)

Načini ocenjevanja:**Assessment methods:**

• pisni izpit (opravljeni seminarji, opravljen preizkus praktične usposobljenosti sta pogoja za pristop k pisnemu izpitu)	40 %	• Examination (Completed seminars and Completed practical skills examination are condition for approach to examination)
• ustno izpraševanje POGOJI ZA PRISTOP K POSAMEZNEMU PREVERJANJU ZNANJA - Pred izpitom morajo študenti obvezno opraviti klinične vaje in seminarje (pozitivno mnenje asistentov) - Obvezna 50 % udeležba na predavanjih - pisni izpit: 50 vprašanj (možen eden ali več pravih odgovorov) - opravljeni seminarji in opravljen preizkus praktične usposobljenosti sta pogoja za pristop k pisnemu izpitu)	60 %	• Oral examination. REQUIREMENTS FOR ACCESS TO INDIVIDUAL KNOWLEDGE CHECKING Before taking the exam it is obligatory for students to complete clinical practice and coursework (positive opinion of the assistants). Obligatory 50% attendance at lectures. Written exam: 50 questions (one or more correct answers are possible). Completed coursework and completed test of practical qualification are requirements for access to the written exam.

Reference nosilca / Course coordinator's references:**NATAŠA MARČUN VARDAR:**

1. MARČUN-VARDA, Nataša, MOČNIK, Mirjam. Pulse wave velocity, central haemodynamic parameters, and markers of kidney function in children. *Kidney & blood pressure research*, ISSN 1423-0143. [Online ed.], 2022, vol. 47, no. 1, str. 43-49. <https://www.karger.com/Article/FullText/519340>, <https://doi.org/10.1159/000519340>, doi: [10.1159/000519340](https://doi.org/10.1159/000519340). [COBISS.SI-ID 81633795].
2. MOČNIK, Mirjam, MARČUN-VARDA, Nataša. Lipid biomarkers and atherosclerosis - old and new in cardiovascular risk in childhood. *International journal of molecular sciences*, ISSN 1422-0067, 2023, vol. 24, issue 3, [article no.] 2237, str. [1]-17, ilustr. <https://www.mdpi.com/1422-0067/24/3/2237>, <https://doi.org/10.3390/ijms24032237>, doi: [10.3390/ijms24032237](https://doi.org/10.3390/ijms24032237). [COBISS.SI-ID 139222787].
3. HERTIŠ PETEK, Tjaša, PETEK, Tadej, MOČNIK, Mirjam, MARČUN-VARDA, Nataša. Systemic inflammation, oxidative stress and cardiovascular health in children and adolescents : a systematic review. *Antioxidants*, ISSN 2076-3921, 2022, vol. 11, issue 5, str. [1]-43, ilustr. <https://www.mdpi.com/2076-3921/11/5/894>, <https://doi.org/10.3390/antiox11050894>, doi: [10.3390/antiox11050894](https://doi.org/10.3390/antiox11050894). [COBISS.SI-ID 106260483].
4. RADULOVIC, Živa, POLAK ZUPAN, Zarja, TOMAZINI, Aljoša, MARČUN-VARDA, Nataša. Vitamin D in pediatric patients with obesity and arterial hypertension. *Scientific reports*, ISSN 2045-2322, 2021, [Vol.] 1, str. 1-8. <https://www.nature.com/articles/s41598-021-98993-8>, <https://doi.org/10.1038/s41598-021-98993-8>, doi: [10.1038/s41598-021-98993-8](https://doi.org/10.1038/s41598-021-98993-8). [COBISS.SI-ID 79144963].
5. GOLOB JANČIČ, Sonja, MOČNIK, Mirjam, ŠVIGELJ, Marjetka, MARČUN-VARDA, Nataša. Body composition and cardiovascular risk factors in a paediatric population. *Children*, ISSN 2227-9067, May 2022, vol. 9, issue 5, str. [1]-11,

ilustr. <https://www.mdpi.com/2227-9067/9/5/603>, <https://doi.org/10.3390/children9050603>, doi: [10.3390/children9050603](https://doi.org/10.3390/children9050603). [COBISS.SI-ID [105834755](https://www.cobiss.si/en/record/105834755)].
, doi: [10.3390/metabo11080551](https://doi.org/10.3390/metabo11080551). [COBISS.SI-ID [74162179](https://www.cobiss.si/en/record/74162179)].

VOJKO BERCE:

1. BERCE, Vojko, TOMAZIN, Maja, GORENJAK, Mario, BERCE, Tadej, LOVRENČIČ, Barbara. The usefulness of lung ultrasound for the aetiological diagnosis of community-acquired pneumonia in children. *Scientific reports*, ISSN 2045-2322, 2019, [Vol.] 9, str. [1]-10, ilustr. <https://www.nature.com/articles/s41598-019-54499-y>, doi: [10.1038/s41598-019-54499-y](https://doi.org/10.1038/s41598-019-54499-y). [COBISS.SI-ID [6865727](https://www.cobiss.si/en/record/6865727)].
2. BERCE, Vojko, TOMAZIN, Maja, JERELE, Erika, CUGMAS, Maša, BERCE, Maša, GORENJAK, Mario. Comparison of the effectiveness of penicillin and broad-spectrum β -lactam antibiotics in the treatment of community-acquired pneumonia in children. *Archives of Medical Science*, ISSN 1896-9151, 2020, vol. , issue , str. [1]-8, ilustr. <https://doi.org/10.5114/aoms.2020.98198>, <https://www.termedia.pl/Journal/-19/pdf-41583-10?filename=Comparison%20of%20the%20effectiveness.pdf>, doi: [10.5114/aoms.2020.98198](https://doi.org/10.5114/aoms.2020.98198). [COBISS.SI-ID [28762883](https://www.cobiss.si/en/record/28762883)].
3. BERCE, Vojko, MARHOLD, Teja, DELOPST, Vid. The impact of comorbidities on the severity of atopic dermatitis in children. *Postępy dermatologii i alergologii*, ISSN 2299-0046, 2022, vol. 39, [no.] 4, str. 697-703. <https://www.termedia.pl/The-impact-of-comorbidities-on-the-severity-of-atopic-ndermatitis-in-children,7,44920,1,1.html>, <https://doi.org/10.5114/ada.2021.108426>, doi: [10.5114/ada.2021.108426](https://doi.org/10.5114/ada.2021.108426). [COBISS.SI-ID [78409219](https://www.cobiss.si/en/record/78409219)].
4. KING, Charlotte, MCKENNA, Amanda, FARZAN, Niloufar, VIJVERBERG, Susanne J, SCHEE, Marc P. van der, MAITLAND-VAN DER ZEE, Anke-Hilse, ARIANTO, Lambang, BISGAARD, Hans, BØNNELYKKE, Klaus, BERCE, Vojko, POTOČNIK, Uroš, REPNIK, Katja, et al. Pharmacogenomic associations of adverse drug reactions in asthma : systematic review and research prioritisation. *The pharmacogenomics journal*, ISSN 1473-1150, Oct. 2020, vol. 20, issue 5, str. 621-628. <https://www.nature.com/articles/s41397-019-0140-y.pdf>, <https://doi.org/10.1038/s41397-019-0140-y>, doi: [10.1038/s41397-019-0140-y](https://doi.org/10.1038/s41397-019-0140-y). [COBISS.SI-ID [6917951](https://www.cobiss.si/en/record/6917951)].
5. BERCE, Vojko, KOREN, Brigita, HOJNIK, Tina. Clinical and laboratory characteristics of children with anaphylaxis. *Indian journal of pediatrics*, ISSN 0973-7693, 2022, [v tisku][1 str.]. <https://doi.org/10.1007/s12098-022-04389-5>, <https://link.springer.com/article/10.1007/s12098-022-04389-5>, doi: [10.1007/s12098-022-04389-5](https://doi.org/10.1007/s12098-022-04389-5). [COBISS.SI-ID [127960835](https://www.cobiss.si/en/record/127960835)].