

Godišnja i izborna skupština Sekcije za bolesti metabolizma
Šibenik, 11.10.2025.

Izvješće o radu Sekcije za razdoblje od 02/2021. do 10/2025.

Danijela Petković Ramadža
Tamara Žigman

Kongres „Europediatrics”

Zagreb, 9.10.2021.

Simpozij:

Inborn errors of metabolism (SSIEM)

Chairs: Christian Staufner (Heidelberg, Germany), Martin Magner (Prague, Czech Republic), Ivo Barić (Zagreb, Croatia), Ellen Crushell (Dublin, Ireland)

ABC of inherited metabolic disorders, Ellen Crushell (Dublin, Ireland)

Genetic diseases underlying fever related liver dysfunction, Christian Staufner (Heidelberg, Germany)

Rhabdomyolysis - mind inherited metabolic diseases, Danijela Petković Ramadža (Zagreb, Croatia)

Lysosomal diseases: Past, present and future, Martin Magner (Prague, Czech Republic)

ROUND TABLE: Hyperammonemia: A clinical approach with case presentations

Chairs: Ivo Barić (Zagreb, Croatia), Tamara Žigman (Zagreb, Croatia)



Stručni sastanci Sekcije - online

- **Gangliozidi i neurometaboličke bolesti, 25.1.2022.**

(predavači: prof. Marija Heffer, Danijela Petković Ramadža, Goran Krnjak, Tamara Žigman)

- **Porodična hiperkolesterolemija – uvođenje i provedba obveznog probira pri upisu u školu, 25.1.2023.**

(predavači: Ivo Barić, Željko Reiner, Danijela Petković Ramadža, Tamara Žigman)

- **Epilepsija u djece zbog nasljednih bolesti metabolizma, 14.2.2024.**

(predavači: Nina Barišić, Ivo Barić, Tamara Žigman, Gordana Mustać, Ivana Kolić, Danijela Petković Ramadža)

- Sastanak organiziran u suradnji s Hrvatskim društvom za dječju neurologiju

Prosječno 80-ak sudionika na sastancima

Recordati Rare Diseases Course INBORN ERRORS OF METABOLISM: FACTS, DOUBTS AND FUTURE OF NUTRITION THERAPY

- Zadar, 2.- 4.6.2022.
- Znanstveni odbor: Ivo Barić, Julio Cesar Rocha, Valentina Rahelić





Recordati
Rare Diseases
Fondation d'entreprise

WITH THE SUPPORT OF: REFERRAL CENTRE FOR GENETICS, METABOLIC DISEASES AND NEWBORN SCREENING OF THE MINISTRY OF HEALTH, REPUBLIC OF CROATIA (DEPARTMENT OF PAEDIATRICS, UNIVERSITY HOSPITAL CENTRE ZAGREB); CROATIAN PAEDIATRIC SOCIETY, SECTION FOR METABOLIC DISEASES AND THE CROATIAN SOCIETY OF NUTRITIONISTS AND DIETITIANS

PROGRAMME

2 June

14:00 Start of the course

Session I Meet participants

Welcome and introduction
(Ivo Barić, Zagreb, Júlio César Rocha, Lisbon, Valentina Rahelić, Zagreb)

The field of inherited metabolic diseases (Ivo Barić, Zagreb)

Role of dietitians in care of patients with inherited metabolic diseases – round table (Júlio César Rocha, Lisbon and Valentina Rahelić, Zagreb)

Session II Low protein diets throughout life cycle – part 1

Phenylketonuria: prototype of inborn errors of metabolism and their treatment (Jiri Zeman, Prague)

Nutrition in phenylketonuria: what is beyond phenylalanine levels? (Júlio César Rocha, Lisbon)

Challenges in nutritional phenylketonuria treatment - interactive session and patients' experience (Charlotte Ellerton, London)

Food replacements - now and beyond (Anita MacDonald, Birmingham)

Dinner at the hotel

Note: Participants are encouraged to prepare case reports which were difficult or illustrative in terms of nutritional management.

3 June

Session III Low protein diets throughout life cycle – part 2

Overview of organic acidurias (Daniela Karal, Innsbruck)

Nutritional management of organic acidurias (Carmen Rohde, Leipzig)

Clinical spectrum of urea cycle disorders (Ivo Barić, Zagreb)

Nutritional management of urea cycle disorders (Carmen Rohde, Leipzig)

Clinical and biochemical monitoring of metabolic diseases treatable with low protein diets (Jiri Zeman, Prague)

Multidisciplinary approach in treatment of severe ornithine transcarbamylase deficiency - interview with a mother (Moderators: Ivo Barić and Valentina Rahelić, Zagreb)

Session IV Carbohydrates and fats

Liver glycogenoses (Danijela Petković, Zagreb)

Nutritional management of glycogenoses (Alice Dianin, Verona)

How restrictive can we be? (Ivo Barić, Valentina Rahelić, Zagreb)

Fatty acid oxidation defects - clinical aspects (Carlo Dionisi Vici, Rome)

Nutritional management of fatty acid oxidation defects - recommendations and open questions (Monika Jörg-Streller, Innsbruck)

Case report (s) (Monika Jörg-Streller, Innsbruck)

Familial Hypercholesterolemia - clinical approach (Albert Wiegman, Amsterdam)

Recommended medical nutrition therapy of hypercholesterolemia (Hülya Gökmen Özel, Ankara)

Unusual patient- Case report

Hypertriglyceridemia - clinical approach (Albert Wiegman, Amsterdam)

Medical nutrition therapy of hypertriglyceridemia (Hülya Gökmen Özel, Ankara)

Dinner with a guided tour of Zadar

4 June

Session V Ketogenic diet and special treats

Who needs ketogenic diet? (Daniela Karal, Innsbruck)

Ketogenic diet in practice (Nikola Mesarić, Zagreb)

Food supplements in inborn errors of metabolism – do we pay enough attention? (Anita MacDonald, Birmingham)

Pregnancy related challenges in inborn errors of metabolism (Elaine Murphy, London)

Debates (each 15 minutes)

- How to prevent tube feeding dependence and aversion to food in inborn errors of metabolism - tricks, tips and facts (Moderators: Danijela Petković Ramadža, Zagreb and Charlotte Ellerton, London)
- Do we need at all a diet for hyperuricemia? (Moderators: Nikola Mesarić, Zagreb and Jiri Zeman, Prague)
- Specific parenteral nutrition for inborn errors of metabolism (Moderators: to be confirmed)

How to educate our patients? (Anita MacDonald, Birmingham)

A family's perspective - (Moderators: Elaine Murphy, London and Julio Cesar Rocha, Lisbon)

13:30 End of the course

Kongres Hrvatskog pedijatrijskog društva Rovinj 2023.

- Predavanje ispred Sekcije - profesor Barić "Pet godina proširenog novorođenačkog probira u Hrvatskoj- što smo postigli?"
- Usmeno priopćenje: Klara Miljanić
- 7 poster prezentacija



Tečaj Pedijatrija danas 2023.

Mini simpozij: HIPERKOLESTEROLEMIJA U DJECE

- Zagreb (Medicinski fakultet), 9.12.2023.
- Moderatorice: Tamara Žigman i Danijela Petković Ramadža
- Predavači: Ivo Barić, Danijela Petković Ramadža, Katja Dumić Kubat i Valentina Rahelić
- Panel rasprava



Preko 100 prijavljenih sudionika na tečaju Pedijatrija danas

40. Proljetna pedijatrijska škola BOLESTI METABOLIZMA

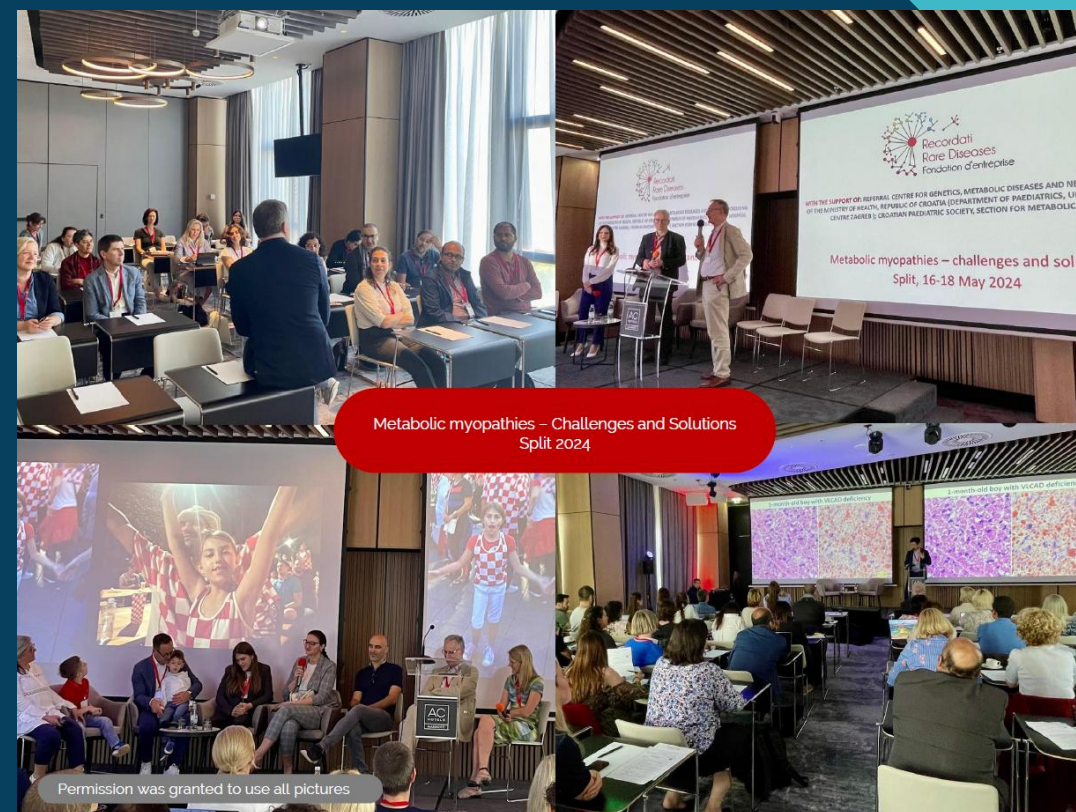
- Split, 17. i 18.4.2024.
- Moderatori: Ivo Barić, Silvija Pušeljić i Danijela Petković Ramadža
- 21 predavanje
- 7 inozemnih predavača (eksperata)



Recordati rare diseases course

METABOLIC MYOPATHIES - CHALLENGES AND SOLUTIONS

- Split, 16.-18.5.2024.
- Znanstveni odbor: Ivo Barić, Danijela Petković Ramadža, Hanns Lochmuller



Tečaj Nasljedne metaboličke bolesti 2024.

BOLESTI KOSTIJU I POREMEĆAJI PROMETA MINERALA U DJECE

- NOVE SPOZNAJE I MOGUĆNOSTI LIJEČENJA

- Zagreb (Medicinski fakultet), 27.9.2024.
- Voditelji: Ivo Barić i Danijela Petković Ramadža
- Tajnik: Ivan Tečer
- U okviru tečaja održan Simpozij Sekcije
- 15 predavača
- 40 polaznika



Interdisciplinarni stručni sastanak KETOGENA DIJETA- KOME TREBA I KAKO JE PROVODITI?

- Zagreb (KBC Zagreb), 5.10.2024.
- Voditelji: Ivo Barić i Danijela Petković Ramadža
- Predavači: Ivo Barić, Nataša Nenadić Baranašić, Nikola Mesarić, Dunja Piljac
- Degustacija pripravaka za ketogenu dijetu
- Sudionici: liječnici, nutricionisti, roditelji i pacijenti, predstavnici farmaceutskih firmi



iNTD Forum

- Zagreb, **16.5.2025.**
- Sekcija za bolesti metabolizma - suorganizator
- Naši članovi predavači i sudionici



 **SAVE THE DATE!** 

RARE NEUROTRANSMITTER DISEASES (RND)
RESEARCHERS FORUM

**CONNECTING THE NETWORK:
FROM BASIC RESEARCH AND CLINICAL
PHENOTYPES TO PATIENT VOICE**

16th May 2025, Hotel Movenpick, Zagreb, Croatia
Registration [HERE!](#)

The RND Researchers Forum is

- Supported by the **iNTD International Working Group on Neurotransmitter-Related Disorders**
- In collaboration with the **Croatian Pediatric Society, Section for Metabolic Diseases**

MAY 2025

MONDAY	TUESDAY	WEDNESDAY	THURSDAY	FRIDAY	SATURDAY	SUNDAY
12	13	14	15	16	17	18

Chan Zuckerberg Initiative 

This Forum has been made possible by funding from Chan Zuckerberg Initiative.



Simpozij PORODIČNA HIPERKOLESTEROLEMIJA U DJECE: DOSADAŠNJA ISKUSTVA I NOVOSTI U PROVOĐENJU NACIONALNOG PROBIRA

- Zagreb (Škola narodnog zdravlja), 6.6.2024.
- Voditelji: Ivo Barić i Danijela Petković Ramadža
- Tajnica: Katarina Stojanović
- Predavači: Ivo Barić, Tamara Žigman, Silvija Pušeljić, Višnja Tokić Pivac, Ivan Pećin, Valentina Rahelić, Danijela Petković Ramadža, Kristina Crkvenac Gornik, Katarina Stojanović
- Dvije panel rasprave
- 35 sudionika (pedijatri i specijalisti školske medicine)



Nagrada „Dijagnoza godine”

- 2022. – dr. Luka Brajković (KBC Split)
- 2023. – dr. Dijana Mlinar (OB Koprivnica)
- 2024. – dr. Sara Dejanović Bekić (KBC Zagreb)



Suradnja sa Savezom za rijetke bolesti i udruženjima pacijenata

- Udruga za pomoć obiteljima s PKU
- Udruga oboljelih od Gaucherove bolesti
- Niemann Pick Hrvatska
- Udruge Hrabriša i Bromologos
- European Galactosemia Society
- International NPD Association



10 INPDA Meeting Argentina&Brazil 9/2025



EGS/Galnet Meeting 11/2023



5. Regionalni sastanak pacijenata s Gaucherovom bolešću 10/2023



8. Nacionalna konferencija o rijetkim bolestima 9/2025



Regionalni sastanak pacijenata s rijetkim bolestima 9/2024

Ostale aktivnosti

Suradnja s Hrvatskim zavodom za zdravstveno osiguranje:

- Dopuna indikacije i smjernice pri propisivanju pripravaka za niskoproteinsku prehranu (niskoproteinski pripravci se mogu izdavati u ljekarnama na recept)
- Stručna mišljenja vezana uz smjernicu za primjenu lijekova za rijetke bolesti

Suradnja s Ministarstvom zdravstva:

- Povjerenstvo za novorođenački probir (Ivo Barić, Ksenija Fumić, Danijela Petković Ramadža)
- Povjerenstvo za nacionalni probir na porodičnu hiperkolesterolemiju (Ivo Barić)

Nacionalno povjerenstvo za rijetke bolesti (Ivo Barić)

- Izrada Nacionalnog programa za rijetke bolesti

Članstvo

- **30 članova** (liječnici, biokemičari, dijetetičari, medicinske sestre)
- Nema članarine (članstvo u HPD-u)