



Inherited Metabolic Diseases in pediatric and adult pathology,
focus on therapy, 5th ed.,
with the session

"At the crossroads between metabolism and autoinflammatory disorders"

28-29 June 2024, Grand Hotel Napoca, Cluj-Napoca, Romania

Event credited with continuing medical education (EMC) points

Distinguished International Speakers

Prof. Dr. Gabriella Horvath

Division of Biochemical Diseases, BC Children's Hospital,
University of British Columbia, Vancouver, Canada

Dr. Doc. Laura Cif

Unit of Functional Neurosurgery, Centre Hospitalier Universitaire Montpellier, France;
Lausanne University Hospital, Switzerland

Dr. Birgit Assmann

Heidelberg University, Medical Faculty Heidelberg,
Center for Pediatric and Adolescent Medicine
Department I, Division of Pediatric Neurology and Metabolic Medicine, Germany

Prof. Dr. Anna Lehman

Medical Director of the Adult Metabolic Diseases Clinic at Vancouver General Hospital and Associate
Professor of Medical Genetics at the University of British Columbia, Vancouver, Canada

Speakers	
Camelia AL-KHZOUZ , MD, PhD, Associate Professor, calkhuzouz@umfcluj.ro	1 st Pediatrics Clinic, Emergency Pediatric Clinical Hospital, "Iuliu Hațieganu" University of Medicine and Pharmacy Cluj-Napoca, Romania
Birgit ASSMANN , MD, PhD, birgit.assmann@med.uni-heidelberg.de	Heidelberg University, Medical Faculty Heidelberg, Center for Pediatric and Adolescent Medicine Department I, Division of Pediatric Neurology and Metabolic Medicine, Germany
Andreea BADEA , MD, PhD, Assistant Professor andreea.stefan24@gmail.com	"Alex. Obregia" Clinical Hospital of Psychiatry Bucharest, Carol Davila University of Medicine and Pharmacy, Bucharest, Romania
Diana BÂRCĂ , MD, Assistant Professor diana_barca@yahoo.com	Alex. Obregia Clinical Hospital of Psychiatry Bucharest, Carol Davila University of Medicine and Pharmacy, Bucharest, Romania

Corina BOCȘA , MD, PhD ibocsa@yahoo.com	2 nd Internal Medicine Department, "Iuliu Hațieganu" University of Medicine and Pharmacy, Interservisan Medical and Surgical Center Cluj-Napoca, Romania
Magdalena BUDIȘTEANU , MD, PhD, Associate Professor, magda_efrim@yahoo.com	V. Babeș National Institute of Pathology, Bucharest, "Alexandru Obregia", Clinical Hospital of Psychiatry Bucharest, Titu Maiorescu University, Faculty of Medicine, Bucharest, "Carol Davila" University of Medicine and Pharmacy, Bucharest, Romania
Meda Ada BUIGI , Dietitian, Assistant professor, PhD St, buiji.ada@umft.ro	"Louis Țurcanu" Clinical Emergency Hospital for Children Timișoara, Romania
Laura CIF , Dr. Doc., MD, PhD, a-cif@chu-montpellier.fr	Unit of Functional Neurosurgery, Unit Movement Disorders, Unité "Pathologies Cérébrales Résistantes", Department of Neurosurgery, University Hospital, CHRU, Montpellier University Hospital, France, Lausanne University Hospital, Switzerland
Bogdan CHIȘ , MD, PhD, Lecturer, bogdan_a_chis@yahoo.com	2 nd Medical Department, Emergency Clinical County Hospital, Cluj-Napoca, Faculty of Medicine, "Iuliu Hațieganu" University of Medicine and Pharmacy Cluj-Napoca
Laura DAMIAN , MD, PhD ldamian.reumatologie@gmail.com	Department of Rheumatology, Center for Rare Musculoskeletal Autoimmune and Autoinflammatory Diseases, Emergency Clinical County Hospital Cluj, Romania
Dorica DAN , Psychologist, dorica.dan@eurordis.org	Vice-President of EURORDIS-Rare Diseases Europe, Chair of the Romanian Prader Willi Association, President of the National Alliance for Rare Diseases (ANBRARO)
Dan DÎRZU , MD, PhD, dirzudan@gmail.com	Department of Anesthesia and Intensive Care, Orthopedic clinic, Emergency Clinical County Hospital Cluj-Napoca, "Regina Maria" Hospital Cluj-Napoca, Romania
Mihaela FARCĂU , MD, PhD, mihaelafarcu@yahoo.com	Department of Pediatrics, "Regina Maria" Hospital Cluj-Napoca, Romania
Ileana FILIPESCU , MD, PhD, Lecturer, ileana_nicoara@yahoo.com	Rheumatology Department, Emergency Clinical County Hospital Cluj, "Iuliu Hațieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania
Simona GRAD , MD, Associate Professor, Simona.Grad@umfcluj.ro	2 nd Department of Internal Medicine, "Iuliu Hațieganu" University of Medicine and Pharmacy, Cluj-Napoca; 2 nd Medical Department, Emergency Clinical County Hospital, Cluj-Napoca, Romania
Gabriella HORVATH , MD, PhD, Professor, ghorvath@cw.bc.ca	Division of Biochemical Genetics, Department of Pediatrics, University of British Columbia, BC Children's Hospital, BC Children's Hospital Research Institute, Vancouver, Canada
Dana IACOB , MD, PhD, Associate Professor, Daniela.Iacob@umfcluj.ro	Department of Pediatrics, "Iuliu Hațieganu" University of Medicine and Pharmacy Cluj-Napoca, Clinical Hospital for Pediatric Emergencies, Cluj-Napoca, Romania
Silvia IANCU , MD, PhD, silviastefaniaiancu@gmail.com	Clinical Center of Diabetes, Nutrition and Metabolic Diseases, Cluj-Napoca, Romania

Ciprian JURCUȚ , MD, PhD, cjurcut@gmail.com	2 nd Internal Medicine, Dr. Carol Davila Central Military Emergency Hospital, Bucharest, Romania
Kinga KOZMA , MD, PhD, kozmaiki@gmail.com	Regional Center for Medical Genetics Bihor, Clinical Emergency County Hospital Oradea, Faculty of Medicine and Pharmacy, University of Oradea, Romania
Cecilia LAZEA , MD, PhD, Associate Professor, cicilazearo@yahoo.com	1 st Pediatrics Clinic, Emergency Pediatric Clinical Hospital, "Iuliu Hațieganu" University of Medicine and Pharmacy Cluj-Napoca, Romania
Anna LEHMAN , MD, PhD, Professor, alehman@cw.bc.ca	Adult Metabolic Diseases Clinic at Vancouver General Hospital, Medical Genetics - University of British Columbia, Vancouver, Canada
Monica Alina MAGER , MD, PhD, Assistant Professor, maberecki@yahoo.com	Department of Pediatric Neurology, Cluj Children's Emergency Clinical Hospital, Department of Neurology and Child Neurology, University of Medicine and Pharmacy "Iuliu Hațieganu" Cluj-Napoca, Romania
Alma MANIU , MD, PhD, Professor, aurelia.maniu@umfcluj.ro	Department of Otolaryngology, "Iuliu Hațieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania
Mircea Milaciu , MD, PhD, Lecturer, mircea_milaciu@yahoo.com	4th Department-Internal Medicine, 4th Medical Discipline, Faculty of Medicine, "Iuliu Hațieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania
Maria PUIU , MD, PhD, Professor, maria.puiu@gmail.com	Victor Babeș University of Medicine and Pharmacy, Department of Genetics, Center of Genomic Medicine, Timișoara, Romania
Iulia-Elena SIMINA , MD, PhD, siminaiuliaelena@gmail.com	Victor Babeș University of Medicine and Pharmacy, Department of Genetics, Center of Genomic Medicine, Timișoara, Romania
Siao-Pin SIMON , MD, PhD, Associate Professor, simonsiao-pin@gmail.com	Rheumatology Department, Emergency Clinical County Hospital Cluj, "Iuliu Hațieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania
Andreea Simona SCROBOTE , MD, andreea84scrobote@yahoo.com	"Alessandrescu Rusescu" Institute for Mother and Child Health, Pediatric Department, Bucharest, Romania
Carolina SOLOMON , MD, PhD, Associate Professor, carolinasolomon12@gmail.com	Emergency Clinical County Hospital Cluj, Department of Radiology, "Iuliu Hațieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania
Mihaela STĂNCULETE , MD, PhD, Associate Professor, mihaela.fadgyas@umfcluj.ro	Clinical Section of Psychiatry II, Emergency Clinical County Hospital Cluj, Department of Neurosciences, Discipline of Psychiatry, University of Medicine and Pharmacy "Iuliu Hațieganu" Cluj-Napoca
Ioana STĂNESCU , MD, PhD, Associate Professor, ioanastanescu@umfcluj.ro	Department of Neurology I - Clinical Recovery Hospital, University of Medicine and Pharmacy "Iuliu Hațieganu" Cluj-Napoca
Svetlana ENCICA , MD, PhD s_encica@yahoo.com	Department of Morphopathology, "Niculae Stancioiu" Heart Institute, Cluj-Napoca, Romania
Laura ȚIBURCĂ , MD, PhD St. laura_tiburca@yahoo.com	Department of Rheumatology, Oradea County Emergency Clinical Hospital, Faculty of Medicine and Pharmacy, University of Oradea, Romania
Kinga Cristina SLĂVEȘCU , MD, PhD, Lecturer, slavescu.kinga@yahoo.com	Emergency Clinical Children's Hospital Cluj-Napoca, "Iuliu Hațieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania

Lucia Maria SUR , MD, Lecturer, surlucia@gmail.com	Pediatric Clinic I - Emergency Clinical Hospital for Children, Cluj-Napoca, University of Medicine and Pharmacy "Iuliu Hațieganu" Cluj-Napoca, Romania
Lăcrămioara VINTER , lacrimioara.lacrimioara.vinter@gmail.com	President of the Patient Association PKU Life Romania, Oradea, Bihor county, Romania
Mihaela Adela VINȚAN , MD, PhD, Assistant Professor, mihaela.vintan@umfcluj.ro	Department of Pediatric Neurology, Cluj Children's Emergency Clinical Hospital, Department of Neurology and Child Neurology, University of Medicine and Pharmacy "Iuliu Hațieganu" Cluj-Napoca, Romania
Romana VULTURAR , MD, PhD, Professor, romanavulturar@gmail.com	Department of Cell and Molecular Biology, "Iuliu Hațieganu" University of Medicine and Pharmacy Cluj-Napoca, Romania

28 June 2024 (Friday)

Session CHALLENGES IN THE MANAGEMENT OF INHERITED METABOLIC DISEASES (I)	
9.00-9.20	Introduction, Inherited Metabolic Diseases in pediatric and adult pathology Romana Vulturar, Laura Damian
9.20-9.35	Cardiac Manifestations in Inherited Metabolic Diseases Cecilia Lazea
9.35-9.50	Early Clinical Manifestations in Lysosomal Storage Diseases Camelia Al-Khzouz
9.50-10.05	BH ₄ Deficiency, cases from Cluj Children's Emergency Hospital, Romania Lucia Maria Sur, A. Bolunduț, K. Slăvescu, I. Badiu, A. Trifa, M. Mager
10.05-10.20	Carpal Tunnel Syndrome in Hunter's disease Corina Bocșa
10.20-10.35	Pain in Patients with Genetic Metabolic Diseases Dan Dîrzu
10.35-10.50	Discussions
10.50-11.20	COFFEE BREAK, Posters viewing
Session TREATABLE INHERITED METABOLIC DISEASES	
11.20-12.00	Treatable Inherited Metabolic Diseases Gabriella Horvath
12.00-12.40	Treatable Causes of Encephalopathy Birgit Assmann
12.40-13.20	Recognizing and Treating Dystonic Crisis and Status Dystonicus Laura Cif
13.20-14.20	LUNCH
Session CHALLENGES IN THE MANAGEMENT OF INHERITED METABOLIC DISEASES (II)	
14.20-14.35	Movement disorders in Genetic Metabolic Diseases in Adult Pathology Ioana Stănescu
14.35-14.50	Psychiatric Presentations of Inherited Metabolic Diseases Mihaela Stănculete

14.50-15.20	SANOFI SYMPOSIUM 14.50-15.05 Gaucher Disease: The Experience of the Cluj-Napoca Center Bogdan Chiş 15.05-15.20 Novelties in the Treatment of Niemann-Pick Type A/B and B Disease Iulia-Elena Simina
15.20-15.35	Treatment Options for Leigh Disease Monica Alina Mager
15.35-15.50	Arginino-succinic aciduria, Case Presentation Kinga Kozma
15.50-16.05	Therapeutic Principles and Nutritional Behavioral Challenges in Phenylketonuria Meda Ada Buigi
16.05-16.20	Our Experience in the Investigation of IMD Involving Small Molecule Defects Romana Vulturar, A. Nicolescu, C. Deleanu, A. Chiş, A. Bashimov, L. Damian
16.20-16.45	<i>COFFEE BREAK, Posters viewing</i>
Session METABOLIC RED FLAGS FOR ADULT RHEUMATOLOGISTS	
16.45-17.00	Rheumatological Features of Adult-onset Metabolic Diseases Laura Damian
17.00-17.15	Imaging in Metabolic Diseases Carolina Solomon
17.15-17.30	Capillaroscopy as a Diagnostic Means in Rare Disease Ileana Filipescu
17.30-17.45	Thrombosis in VEXAS Syndrome Siao-Pin Simon
17.45-18.00	Metabolomics Data in Ankylosing Spondylitis Laura Ţiburcă
18.15	<i>GUIDED TOUR of the Old Center of Cluj, presenting the rich history and vibrant culture of Cluj-Napoca.</i>

29 June 2024 (Saturday)

Session CENTRAL NERVOUS SYSTEM INVOLVEMENT, CHALLENGES IN THE MANAGEMENT OF INHERITED METABOLIC DISEASES (III)	
9.00-9.20	Neurometabolic and Genetic Movement Disorders Case Presentations: Symptomatic and Disease-specific Treatments Laura Cif
9.20-9.45	Workshop with cases of Inherited Metabolic Diseases Gabriella Horvath
9.45-10.00	Epileptic encephalopathy caused by mutations of CAD gene – a rare, treatable neurometabolic condition Magdalena Budişteanu, D. Bârcă, D. Surlică, A. Glangher, C. Iliescu
10.00-10.15	Neuronal Ceroid-lipofuscinoses in Pediatric Age - the Experience of an Expertise Center in Rare Diseases Diana Bârcă, A. Dică, D. Craiu, C. Iliescu

10.15-10.30	Mitochondrial Myopathies - Approach and Modern Treatment Options Andreea Badea, D. Bârcă, C. Moțoescu
10.30-10.45	Incorporating Ketogenic Diet into Treatment of Mitochondrial Encephalopathy; the Story of SLC25 Family Transporters and Brain Disorders Mihaela Adela Vințan, D. Bârcă, D. Craiu
10.45-11.00	Challenges in the Management of Methylmalonic Aciduria Patients Andreea Simona Scrobote, Roxana Minciună
11.00-11.30	COFFEE BREAK, Posters viewing
Session “AT THE CROSSROADS BETWEEN METABOLISM AND AUTOINFLAMMATORY DISORDERS”	
11.30-11.50	NOVARTIS SYMPOSIUM: The Spectrum of Mevalonate Kinase Deficiency Laura Damian
11.50-12.05	A Challenging Diagnostic Journey in Two Brothers Mihaela Farcău
12.05-12.20	Mevalonate Kinase Deficiency and Other Rare Diseases that Mimic Inflammatory Bowel Disease Simona Grad
12.20-12.35	The Ear in Autoinflammatory Diseases Alma Maniu
12.35-12.50	Somatic Mutations in Autoinflammatory Diseases Ciprian Jurcuț
12.50-13.00	Discussions
13.00-14.15	LUNCH
Session CHALLENGES IN THE MANAGEMENT OF INHERITED METABOLIC DISEASES (IV)	
14.15-14.30	Phenylketonuria in Children in Cluj-Napoca Regional Center: from Screening to Monitoring Kinga Cristina Slăvescu, L.M. Sur, I. Badiu, C. Costache
14.30 – 14.45	Challenges in the Care of Adult PKU Patients Silvia Iancu
14.45-15.00	Carbaglu in the Treatment of NAGS Deficiency & Organic Acidemias (Recordati Rare Diseases) Romana Vulturar
15.00-15.30	Treatment with Enzyme Replacement Therapy Anna Lehman
Session INHERITED METABOLIC DISEASES: HEALTH AND SOCIETY	
15.30-15.45	Expanding Neonatal Screening: From Suspicion to Confirmed Diagnosis and Integrated Management Maria Puiu, I.E. Simina
15.45-16.00	Expanding Neonatal Screening in Romania from the Patients' Perspective Dorica Dan

16.00-16.15	Evolution and Current Needs in the Treatment of Phenylketonuria in Romania Lăcrimioara Vinter
16.15-16.45	<i>COFFEE BREAK, Posters viewing</i>
Session RARE SYSTEMIC DISEASES ON THE BORDER OF INHERITED METABOLIC DISEASES	
16.45-17.00	Cardiovascular Morphopathology in a Case of Ehlers-Danlos Syndrome Svetlana Encica Department of Morphopathology, "Niculae Stancioiu" Heart Institute, Cluj-Napoca, Romania
17.00-17.15	Proteus Syndrome Dana Iacob , Simona Șenilă, Carolina Solomon, Mihaela Mureșan Department of Pediatrics, "Iuliu Hațieganu" University of Medicine and Pharmacy Cluj-Napoca, Clinical Hospital for Pediatric Emergencies, Cluj-Napoca, Romania
17.15-17.30	Macrodystrophia Lipomatosa Mircea Milaciu , Angela Cozma, Laura Damian 4 th Department-Internal Medicine, 4 th Medical Discipline, Faculty of Medicine, "Iuliu Hațieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania
17.30-17.45	Discussions, Posters Awards. Closing the event.

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