CONFERENCE · 6TH EDITION

Inherited Metabolic Diseases

in pediatric and adult pathology, 6th ed.

with the session

"At the crossroads between metabolism and autoinflammatory disorders"

Connecting the dots: bridging specialists and pathways

8-10 October 2026

Aula UMF Cluj (8 October - Opening session at 17.00)

Double Tree by Hilton Cluj City-Plaza Hotel (9 and 10 October)

Cluj-Napoca, Romania



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and Double Tree by Hilton Cluj City-Plaza Hotel (9 and 10 October) Cluj-Napoca, Romania

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
Department of Clinical Neurosciences, Lausanne University Hospital (CHUV), Lausanne, Switzerland;
Institut du Neurone (IdN), Montpellier University Hospital, Montpellier, France

Dr. Birgit Assmann
Department of General Pediatrics, Division of Neuropediatrics and Metabolic Medicine, University Hospital Heidelberg, Germany

Dr. Trine Tangeraas
Consultant in Pediatrics and Metabolic Medicine, Norwegian National Unit for Newborn Screening, Oslo University Hospital, Norway

Associate Prof. Dr. Cristina Belizna
Department of Vascular Medicine, Angers University Hospital (CHU Angers), Angers, France
University of Angers, Angers, France

Júlio César Rocha, PhD, Habil.
Clinical Nutritionist, Reference Centre for Inherited Metabolic Diseases, Unidade Local de Saúde de São José, Lisbon; Assistant Professor of Nutrition and Metabolism, NOVA Medical School, Lisbon, Portugal



https://www.proeventsinternational.com/event/inherited-metabolic-diseases-in-pediatric-and-adult-pathology/



Dear colleagues, dear participants,

It is a great honor and joy to welcome you warmly to our conference on rare genetic diseases with a particular focus on inherited metabolic diseases. Metabolism provides a unifying perspective from which to explore how genetic defects disrupt cellular function, linking energy production, molecular homeostasis and signaling to disease mechanisms across organs and systems. By bringing together advances in genetics, metabolism and cellular biology, we aim to translate this understanding into earlier diagnosis and more effective treatment. Building on previous editions, the session "*At the crossroads between metabolism and autoinflammatory disorders*" extends this perspective to the interplay between metabolic pathways, immune regulation and inflammation.

We meet again under the dome of a common desire: to bring light and solutions to a medical field of extraordinary complexity. Rare metabolic diseases and autoinflammatory pathologies represent real challenges of diagnosis and treatment, in which the boundaries between specialties disappear, leaving room for interdisciplinary collaboration.

This conference was designed as a dynamic space for dialogue, meant to unite specialists from various fields in child and adult pathology, researchers, biologists, nutritionists, students and patient associations. We aim to explore together the latest scientific discoveries, updated clinical guidelines and innovative therapeutic strategies that have the power to change destinies.

Beyond the academic presentations and scientific debates, the supreme value of this event lies in the community we build and strengthen. Every exchange of experience and every connection established here directly contributes to shortening our patients' diagnostic odyssey and improving their quality of life.

Thank you for your presence, for your dedication and for your tireless desire to excel for the benefit of patients with rare diseases. We wish you days full of inspiration and fruitful debates!

Prof. Dr. Romana Vulturar and Dr. Laura Damian

This event is accredited by the Romanian College of Physicians (CMR) for continuing medical education (CME).

LOCAL ORGANIZING COMMITTEE

Romana Vulturar, Laura Damian, Adina Chiș, Lorena Hanțig, Mădălina Rafa, Raul Gall

SCIENTIFIC COMMITTEE

Gabriella Horvath, Laura Damian, Romana Vulturar, Laura Cif, Birgit Assmann, Cecilia Lazea, Elena Neagu, Camelia Al-Khzouz, Cristina Belizna, Magdalena Budișteanu, Claudia Jurca, Alina Silaghi, Carolina Solomon, Júlio Rocha, Diana Miclea, Mădălina Leanca, Adina Chiș, Lucia Sur, Ștefan Popa, Adina Stan

8 October 2026 (Thursday),
Aula, "Iuliu Haţieganu" University of Medicine and Pharmacy, 23 Gheorghe Marinescu Street, Cluj-Napoca, Romania

OPENING session	
17.00-17.15	Welcome, Introduction, The Fascination of Rare Diseases: why Metabolic Disorders Matter and What They Teach Us **Romana Vulturar, Laura Damian** *Department of Cell and Molecular Biology, "I. Haţieganu" University of Medicine and Pharmacy Cluj-Napoca, Romania; Department of Rheumatology, Center for Rare Musculoskeletal Autoimmune and Autoinflammatory Diseases, Emergency Clinical County Hospital Cluj, Romania*
17.15- 17.45	Autophagy in Inherited Metabolic Diseases, an Emerging Disease Mechanism **Gabriella Horvath** *Division of Biochemical Genetics, Department of Pediatrics, University of British Columbia, BC Children's Hospital, BC Children's Hospital Research Institute, Vancouver, Canada*
17.45-18.15	Dopamine Transporter Deficiency Syndrome: the dilemma of too much and inefficient dopamine in the same time **Birgit Assmann** *Centre for Child and Adolescent Medicine, Department of General Pediatrics, Division of Neuropediatrics and Metabolic Medicine, University Hospital Heidelberg, Germany*
18.15-18.45	Approaching Complex Movement Disorders in Genetic and Neurometabolic Diseases: Toward More Objective Assessments **Laura Cif** *Department of Clinical Neurosciences, Lausanne University Hospital (CHUV), Lausanne, Switzerland; Institut du Neurone (IdN), Montpellier University Hospital, Montpellier, France*
19.00	***Welcome Refreshments*** ***Foyer – 'Iuliu Haţieganu' Auditorium***

9 October 2026 (Friday),
Double Tree by Hilton Cluj City-Plaza Hotel, 9-13, Sindicatelor St, Cluj-Napoca
Speakers are kindly requested to keep strictly within their allocated presentation time!

SESSION I — INHERITED METABOLIC DISEASES: CARDIOVASCULAR AND LIPID INVOLVEMENT, **Chairs: Cecilia Lazea, Cristina Belizna**	
8.30-8.45	Ventricular preexcitation in inherited metabolic diseases **Cecilia Lazea** *1st Pediatrics Clinic, Emergency Pediatric Clinical Hospital, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*

8.45-09.00	Vascular complications in inborn metabolic diseases **Cristina Belizna** *Department of Vascular Medicine, Angers University Hospital (CHU Angers); University of Angers, France*
9.00-09.15	Diagnosis and therapeutic approach in primary systemic carnitine deficiency **Andra Giorgiana Zaruha**, E. Neagu, G. Nicolae, C.I. Raita, B. Pavlov, C. Bercu, M. Leanca, R. Petcu, M. Bejan, A. Mirea *"Robănescu-Pădure" National Clinical Center for Children Neuro-Psychomotor Rehabilitation, Bucharest, Romania*
9.15-9.35	SOBI SYMPOSIUM — Genetic testing in familial chylomicronemia syndrome **Adela Chiriță** *Department of Medical Genetics, "Victor Babeș" University of Medicine and Pharmacy, Timișoara; "Louis Țurcanu" Emergency Clinical Hospital for Children, Timișoara, Romania*
SESSION II — INHERITED METABOLIC DISEASES: FROM NEUROLOGICAL MANIFESTATIONS TO THERAPEUTIC ADVANCES **Chairs: Adina Stan, Romana Vulturar**	
9.35-9.50	Neurologic manifestations in rare inborn metabolic diseases **Adina Stan** *Department of Neurology, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*
9.50-10.10	From Control to Optimisation in the Nutritional Management of Inherited Metabolic Disorders **Júlio Rocha** *Reference Centre for Inherited Metabolic Diseases; NOVA Medical School, University NOVA de Lisboa, Portugal*
10.10-10.30	PTC SYMPOSIUM — Advancement in PKU management: mechanism of action and clinical data of sepiapterin **Romana Vulturar** *Department of Cell and Molecular Biology, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*
10.30-10.50	PTC SYMPOSIUM — Sepiapterin and dietary management in PKU patients: guidelines and real practice **Meda Ada Bugi** *"Louis Țurcanu" Clinical Emergency Hospital for Children, Timișoara, Romania*
10.50-11.05	***COFFEE BREAK & POSTER VIEWING***
SESSION III — METABOLISM, IMMUNE DYSREGULATION AND NEUROLOGICAL DISEASE: FROM MECHANISMS TO CLINICAL PRACTICE **Chairs: Gabriella Horvath, Laura Cif**	
11.05-11.30	Immune dysregulation in inborn errors of metabolism **Gabriella Horvath** *Division of Biochemical Genetics, Department of Pediatrics, University of British Columbia, BC Children's Hospital, BC Children's Hospital Research Institute, Vancouver, Canada*

11.30-11.55	Genetic causes of syndromic movement disorders involving autoimmunity: an overview **Laura Cif** *Department of Clinical Neurosciences, Lausanne University Hospital (CHUV), Lausanne, Switzerland; Institut du Neurone (IdN), Montpellier University Hospital, Montpellier, France*
11.55-12.10	Aicardi–Goutières syndrome: when type I interpheron shapes the neurological picture **Mădălina Leanca,** Elena Neagu, Daniela Vasile, Raluca Petcu, Silvia Shelby, Andrada Mirea *"Robănescu-Pădure" National Clinical Center for Children Neuro-Psychomotor Rehabilitation, Bucharest*
12.10-12.30	When steroids may reverse brain atrophy: a novel autoinflammatory encephalopathy due to PTPN1 haploinsufficiency **Birgit Assmann** *Centre for Child and Adolescent Medicine, Department of General Pediatrics, Division of Neuropediatrics and Metabolic Medicine, University Hospital Heidelberg, Germany*
12.30-12.35	**Discussions**
12.35-12.45	Metabolic imbalance and inflammatory mechanisms in pediatric migraine **Mihaela Adela Vințan** *Child Neurology Department, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania; Clinical Emergency Hospital for Children, Cluj-Napoca*
12.45-13.00	From Molecular Diagnosis to Family Counseling: Navigating Uncertainty and Recurrence Risk **Iulia-Elena Simina** *Victor Babeș University of Medicine and Pharmacy, Department of Genetics, Center of Genomic Medicine, Timișoara, Romania*
13.00-13.15	From patients to flies: personalized Drosophila models in metabolic disease research **Levente Kovacs** *Babeș-Bolyai University, Cluj-Napoca, Romania; Institute for Research, Development and Innovation in Applied Natural Sciences*
13.15-14.15	***LUNCH***
SESSION IV — WHEN TIME MATTERS: EARLY RECOGNITION AND THERAPEUTIC APPROACHES IN RARE GENETIC DISEASES **Chairs: Gabriella Horvath, Elena Neagu**	
14.15-14.45	Early recognition of hyperammonemia-a life-threatening emergency **Trine Tangeraas** *National Newborn Screening Unit, Division of Pediatric and Adolescent Medicine, Oslo University Hospital, Oslo, Norway*

14.45-15.15	IMMEDICA SYMPOSIUM — Urea cycle Disorders: A Therapeutic Approach **Gabriella Horvath** *Division of Biochemical Genetics, Department of Pediatrics, University of British Columbia, BC Children's Hospital, BC Children's Hospital Research Institute, Vancouver, Canada*
15.15-15.45	ROCHE SYMPOSIUM — Spinal muscular atrophy - a rare genetic disease treatable systematically **Elena Neagu** *Genetics Laboratory, National University Center for Children Neurorehabilitation "Robănescu-Pădure", Bucharest, Romania*
15.45-16.00	NOVARTIS SYMPOSIUM — Early diagnosis in Spinal muscular atrophy: the critical role of genetic testing in optimizing prognosis I **Monica Mager** *Department of Pediatric Neurology, Cluj Children's Emergency Clinical Hospital; Department of Neurology and Child Neurology, "Iuliu Haţieganu" University of Medicine and Pharmacy Cluj-Napoca, Romania*
16.00-16.15	NOVARTIS SYMPOSIUM — Early diagnosis in Spinal muscular atrophy: the critical role of genetic testing in optimizing prognosis II **Romana Vulturar** *Department of Neurology and Child Neurology, Department of Cell and Molecular Biology, "Iuliu Haţieganu" University of Medicine and Pharmacy Cluj-Napoca, Romania*
16.15-16.25	Improving outcome for an adolescent with Tay-Sachs disease: a case study **Elena Neagu**, Silvia Shelby, A.G. Zaruha, C. Bercu, A. Grigoriu, M. Leancă, D. Vasile, R. Petcu, M. Bejan, A. Mirea *National University Center for Children Neurorehabilitation "Robănescu-Pădure", Bucharest, Romania*
16.25-16.40	***COFFEE BREAK & POSTER VIEWING***
SESSION V — INHERITED STORAGE DISEASES ACROSS THE LIFESPAN **Chairs: Crina Rusu, Camelia Al-Khzouz**	
16.40-16.55	Liver Involvement in Glycogen Storage Diseases: Clinical Spectrum and Principles of Management **Tudor L. Pop**, Alina Grama *2nd Pediatric Discipline, "Iuliu Haţieganu" University of Medicine and Pharmacy Cluj-Napoca, 2nd Pediatric Clinic, Center of Expertise in Pediatric Liver Rare Diseases, Clinical Emergency Hospital for Children Cluj-Napoca*
16.55-17.10	Update on the management of Fabry disease **Crina Rusu** *Department of Nephrology, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania; Clinical Nephrology and Dialysis Department, Cluj County Emergency Clinical Hospital*
17.10-17.25	Diagnosis and management of type 1 Gaucher disease in children **Camelia Al-Khzouz** *1st Pediatrics Clinic, Emergency Pediatric Clinical Hospital, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*

17.25-17.55	SANOFI SYMPOSIUM — Over 20 years of Cerezyme experience in Gaucher disease: evidence on bone manifestation control and outcomes **Bogdan Chiș** *2nd Medical Department, Emergency Clinical County Hospital Cluj-Napoca; Faculty of Medicine, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*
17.55-18.10	Neuronal ceroid lipofuscinosis in a child from a consanguineous family with three affected members **Alexandru Bolunduț,** Lucia Sur *Department of Mother and Child, Pediatrics I, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania; 1st Pediatric Clinic, Emergency Clinical Hospital for Children, Cluj-Napoca*
SESSION VI — IMAGING IN RARE METABOLIC DISEASES **Chairs: Carolina Solomon, Laura Damian**	
18.10-18.25	Imaging in rare neuromuscular diseases: a practical approach **Carolina Solomon** *Department of Radiology, Emergency Clinical County Hospital Cluj, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*
18.25-18.40	Imaging in rare diseases affecting the salivary glands **Manuela Lenghel,** Marcela Iojiban *Department of Radiology and Medical Imaging, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*
18.40-18.55	***COFFEE BREAK & POSTER VIEWING***
SESSION VII- DIAGNOSTIC AND THERAPEUTIC CHALLENGES IN RARE METABOLIC DISEASES **Chairs: Diana Miclea, Romana Vulturar**	
18.55-19.10	Liver biopsy in chronic liver disease: diagnostic pitfalls and the need to exclude inherited metabolic disorders **Ștefan Popa** *2nd Department of Internal Medicine, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania; 2nd Medical Clinic, Cluj-Napoca*
19.10-19.20	The challenges of genetic diagnosis for rare and ultra-rare diseases: clinical cases **Silvia Shelby**, Elena Neagu *"Robănescu-Pădure" National Clinical Center for Children Neuro-Psychomotor Rehabilitation, Bucharest, Romania*
19.20-19.30	Metabolic Evaluation in Autism Spectrum Disorder **Diana Miclea** *Department of Mother and Child, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*
19.30-19.40	Alkaptonuria: from Musculoskeletal Symptoms in a Disease-Modifying Treatment **Elena Rezuș, Laura Damian** *Department of Rheumatology, Center for Rare Musculoskeletal Autoimmune and Autoinflammatory Diseases, Emergency Clinical County Hospital Cluj, Romania*

19.40-20.00	ASTRA ZENECA SYMPOSIUM — Hypophosphatasia: From Early Diagnosis to Targeted Treatment **Romana Vulturar** *Department of Cell and Molecular Biology, " Iuliu Haţieganu" University of Medicine and Pharmacy Cluj-Napoca, Romania*
20.00	***DINNER***

10 October 2026 (Saturday), Double Tree by Hilton Cluj City-Plaza Hotel, 9-13, Sindicatelor St, Cluj-Napoca

Speakers are kindly requested to keep strictly within their allocated presentation time!

SESSION VIII — AUTOINFLAMMATORY DISEASES: AT THE BORDER BETWEEN INHERITED METABOLIC DISORDERS AND IMMUNITY ERRORS **Chairs: Cristina Pamfil, Ciprian Jurcuţ**	
9.00-9.15	Age-related presentations of autoinflammatory diseases **Laura Damian** *Department of Rheumatology, Center for Rare Musculoskeletal Autoimmune and Autoinflammatory Diseases, Emergency Clinical County Hospital Cluj*
9.15-9.35	Somatic mutations in autoinflammatory diseases **Ciprian Jurcuţ** *Department of Internal Medicine, Carol Davila Central Military Emergency Hospital, Bucharest, Romania*
9.35-9.50	Pregnancy in autoinflammatory diseases **Cristina Pamfil** *Department of Rheumatology, Center for Rare Musculoskeletal Autoimmune and Autoinflammatory Diseases, Emergency Clinical County Hospital Cluj*
9.50-10.05	Clinical and paraclinical aspects of the interaction between metabolic diseases and autoinflammatory syndromes in pediatrics **Mirela Crişan** *Pediatrics I, Emergency Clinical Hospital for Children, Cluj-Napoca, Romania*
10.05-10.25	SOBI SYMPOSIUM (ANAKINRA) — Cryopyrinopathies in the Era of Treatable Rare Diseases **Laura Damian** *Department of Rheumatology, Center for Rare Musculoskeletal Autoimmune and Autoinflammatory Diseases, Emergency Clinical County Hospital Cluj*
SESSION IX — HYPERINFLAMMATION AND IMMUNE DYSREGULATION: CLINICAL CHALLENGES AND COMPLICATIONS **Chairs: Mihaela Lupşe, Laura Urian**	
10.25-10.40	Infectious Complications of Biologic Therapies **Mihaela Lupşe** *Department of Infectious Diseases, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*

10.40-10.55	Hyperinflammation and hemophagocytic syndrome: a hematologist's perspective **Laura Urian** *Department of Hematology, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania; Department of Hematology, "Prof. Dr. Ion Chiricuţă" Oncology Institute, Cluj-Napoca*
10.55-11.10	Hyperinflammation and hemophagocytic syndrome: a rheumatologist's perspective **Maria Magdalena Tămaş** *Department of Rheumatology, Center for Rare Musculoskeletal Autoimmune and Autoinflammatory Diseases, Emergency Clinical County Hospital Cluj*
11.10-11.25	***COFFEE BREAK & POSTER VIEWING***
SESSION X — LIPID METABOLISM AT THE CROSSROADS OF METABOLIC AND IMMUNE DYSREGULATION Chairs: Ina Kacso, Alina Silaghi	
11.25-11.40	Lipodystrophies: a practical approach to diagnosis, metabolic implications and treatment strategies **Alina Silaghi,** Laura Damian, Ina Kacso *Department of Endocrinology, Clinical County Hospital Cluj-Napoca, Romania*
11.40-11.55	Lipodystrophies in autoimmune rheumatic diseases **Ileana Nicoară** *Rheumatology Department, Emergency Clinical County Hospital Cluj, "Iuliu Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*
11.55-12.05	**Discussions:** Ina Kacso, Laura Damian, Alina Silaghi**,** Ileana Nicoară
12.05-12.25	ASTRA ZENECA SYMPOSIUM — Lysosomal Acid Lipase Deficiency (LAL-D) **Tudor L. Pop** *2nd Pediatric Discipline, "I. Haţieganu" University of Medicine and Pharmacy Cluj-Napoca, 2nd Pediatric Clinic, Clinical Emergency Hospital for Children Cluj-Napoca*
SESSION XI — RARE AND COMPLEX DISEASES: HEALTH AND SOCIETY Chairs: Dorica Dan, Magdalena Budişteanu	
12.25-12.40	A rare case of dystonia in a child with a new THAP1 gene variant **Magdalena Budişteanu** *Victor Babeş National Institute of Pathology, Bucharest, Romania; Psychiatry Research Laboratory, Prof. Dr. Alexandru Obregia Clinical Hospital of Psychiatry, Bucharest; Faculty of Medicine, Titu Maiorescu University, Bucharest, Romania*
12.40-12.50	PROFILE-PD: developing capacity in Parkinson's disease research and advocacy among students and early career professionals **Darius Lotrean,** Romana Vulturar *Faculty of Medicine, "I.Haţieganu" University of Medicine and Pharmacy, Cluj-Napoca, Romania*
12.50-13.00	Phenylketonuria: An Integrated Approach from the Physician's and Parent's Perspectives **Sorina Cezara Coste** *Patient Association PKU Life Romania, Oradea, Bihor County, Romania*

13.00-13.15	Role of patients' partnership in advancing knowledge in rare diseases **Dorica Dan** *President, Romanian National Alliance for Rare Diseases (ANBRaRo); Director, NoRo Centre, Zalău, Romania*
13.15-13.30	ROUND TABLE — Patients' pathways in rare metabolic diseases in Romania
13.30-13.40	Discussion. Poster Evaluation and Awards. Conference Closing.
13.40	***LUNCH***

Speakers	
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3. Diaz GA et al. Hepatology. 2013; 57:2171–2179. COM-007739 Data pregătirii: septembrie 2026

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