



UMF
UNIVERSITATEA DE
MEDICINĂ ȘI FARMACIE
IULIU HAȚIEGANU
CLUJ-NAPOCA



**Alianța Națională
pentru Boli Rare
România**



ASOCIAȚIA
PRADERWILLI
DIN ROMANIA

Scientific Program

**Inborn Errors of Metabolism
in child and adult pathology,
4th edition,**

with the session

**"At the crossroads between
metabolism and autoinflammatory disorders"**

**conference *with international
participation***

**29-30 June 2023,
Hybrid event, Cluj-Napoca, Romania**

USEFUL INFORMATION

Official languages: English, Romanian

The scientific secretariat is available according to the schedule below:

Thursday, 29 June: 08:00 - 18:00

Friday, 30 June: 08:00 - 19:00

Oral presentations will be given according to the scientific programme. All lecturers are kindly requested to keep to the time allocated for the presentation to ensure a smooth running of the event.

Posters will be presented during the scientific sessions according to the programme.

Certificates of participation will be issued on the last day of the event and during the next two weeks after the completion of the event, for all registered participants.

We remind you that this event has been credited with:

11 CME points by the Romanian College of Physicians

Organizers Contact:

Logistic Organizer: PROEVENTS & CONFERENCE

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Asociația
pentru inovare
în boli rare
inflamatorii,
metabolice,
genetice
INNOROG



Conference with international participation

Inborn Errors of Metabolism in child and adult pathology, 4th ed. with the session

"At the crossroads between metabolism and autoinflammatory disorders",

**29-30 JUNE 2023, Grand Hotel Napoca, Cluj-Napoca,
Hybrid event credited with CME points**

29 JUNE 2023

08.30 – 09.00 REGISTRATION

Chairs: Gabriella Horvath, Romana Vulturar

**09.00 – 09.15 Welcome, Inborn Errors of Metabolism in child and adult pathology, a short introduction,
Romana Vulturar, MD, PhD, Prof. Discipline of Cell and Molecular Biology,
"I. Hațieganu" University of Medicine and Pharmacy Cluj-Napoca, Romania**

**09.15 – 10.00 Treatable IEMs and App,
Gabriella Horvath, MD, PhD, Prof., Division of Biochemical Genetics,
Department of Pediatrics, University of British Columbia, BC Children's
Hospital, BC Children's Hospital Research Institute, Vancouver, Canada**

Scientific coordinators:

**Dr. Gabriella Horvath (Vancouver),
Dr. Laura Damian (Cluj-Napoca),
Dr. Romana Vulturar (UMF Cluj-Napoca)**

Local organizers:

**Dr. Romana Vulturar (UMF Cluj-Napoca),
Dr. Laura Damian (SCJU Cluj),
Dr. Adina Chiș (UMF Cluj-Napoca)**

29 JUNE 2023

- 10.00 – 10.20** **METABO^{MS} newborn extended screening project: first pilot program in Romania,**
Anca Dana Buzoianu, Rector, MD, PhD, Prof. “I. Hațieganu” University of Medicine and Pharmacy Cluj-Napoca, Romania, Department of Pharmacology, Toxicology and Clinical Pharmacology
Cristina-Adela Iuga, PhD, Prof. MedFUTURE Research Center for Advanced Medicine, “I. Hațieganu” University of Medicine and Pharmacy Cluj-Napoca, Romania
- 10.20 – 10.50** **Newborn screening in Vancouver,**
Bojana Rakic, MD, PhD, Department of Pathology and Laboratory Medicine, Faculty of Medicine, University of British Columbia, Vancouver, Canada
- 10.50 – 11.00** **Atypical absence epilepsy, ataxia and confusional states of genetic etiology; is it treatable or not?**
Mihaela Adela Vințan, MD, PhD, Lecturer Child Neurology Department, “I. Hațieganu” University of Medicine and Pharmacy Cluj-Napoca, Clinical Emergency Hospital for Children, Cluj-Napoca, Romania
- 11.00 – 11.20** **COFFEE BREAK, Poster viewing**
- 11.20 – 12.05** **Enzyme replacement therapy in lysosomal disorders,**
Ramona Salvarinova, MD, PhD, Prof. Division of Biochemical Genetics, Department of Pediatrics, University of British Columbia, BC Children's Hospital, BC Children's Hospital Research Institute, Vancouver, Canada
- 12.05 – 12.50** **Treatment in metabolic myopathies,**
Catherine Brunel, MD, PhD, Prof. Division of Biochemical Genetics, Department of Pediatrics, University of British Columbia, BC Children's Hospital, BC Children's Hospital Research Institute, Vancouver, Canada

29 JUNE 2023

- 12.50 – 13.20 Dietary management of small molecule disorders in children,**
Gloria Ho - Registered dietitian Vancouver Hospital, Canada
- 13.20 – 14.30 LUNCH BREAK**
- 14.30 – 14.45 The complex management of a rare disorder, pyridoxal 5'-phosphate dependent developmental and epileptic encephalopathy - case report,**
Mihaela Adela Vințan, Alina Grama, Mariela Militaru, Mihai Militaru, Carmen Culcitchi, Dan Deleanu, Diana Barca,
MD, PhD, Lecturer, Child Neurology Department, “I. Hațieganu” University of Medicine and Pharmacy Cluj-Napoca, Children's Clinical Emergency Hospital Cluj Napoca, Romania
- 14.45 – 15.00 Two Cases of 6-Pyruvoyl Tetrahydropterin Synthase Deficiency,**
Lucia Maria Sur, Alexandru Bolunduț, Monica Mager, Adrian Trifa,
Dana T. Anton-Păduraru,
MD, PhD, Lecturer, University of Medicine and Pharmacy “I. Hațieganu” Cluj-Napoca, Clinical Emergency Hospital for Children, Cluj-Napoca, Romania
- 15.00 – 15.10 SANOFI SYMPOSIUM**
Acid sphingomyelinase deficiency insights and a new hope in the first therapy to treat it,
Iulia-Elena Simina, Simona Cerbu, Irina Sarău, Maria Puiu,
MD, PhD, Assis. Prof. “Victor Babeș” University of Medicine and Pharmacy Timișoara, Romania
- 15.10 – 15.45 Dietary management of small molecule disorders in adults,**
Jennifer Branov, Registered dietitian Vancouver Hospital, Canada
- 15.45 – 16.00 Neurological implications in cystinosis,**
Vitalie Văcăraș, Cristina Jucan, Dragoș Mihai Corcan
MD, PhD, Assoc. Prof. Neuroscience Department, “I. Hațieganu” University of Medicine and Pharmacy Cluj-Napoca, Romania

29 JUNE 2023

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- 16.00 – 16.15** **Lessons learned: our experience in diagnostic and evaluation of small molecule defects in child and adult pathology,**
Romana Vulturar, Adina Chiş, Alina Nicolescu, Călin Deleanu, Olga Orăşan, Cecilia Lazea, Melinda Baizat, Ligia Blaga, Alina Grama, Tudor Pop, Gabriella Horvath *MD, PhD, Prof. Discipline of Cell and Molecular Biology, “I. Haţieganu” University of Medicine and Pharmacy Cluj-Napoca, Romania*
- 16.15 – 16.35** **COFFEE BREAK, Poster viewing**
- 16.35 – 17.50** **Workshop with case presentations and discussions (cases will illustrate presentation, diagnostic work-up and treatment)**
coordinated by **Gabriella Horvath** and speakers online: Dr. Haifa Al-Sudairy, Dr. Deepa Subramoniam, Dr. Felix Levesque, Dr. Brandon Chalazan, Dr. Hamad Althiyab - Fellows in Biochemical Genetics Department, Vancouver, Canada
- 17.50** **Discussions, closure of the 1st day**

30 JUNE 2023

Chairs: Gabriella Horvath, Birgit Assmann

09.00 – 09.10 Introduction, Welcome

09.10 – 10.00 Treatable Inborn Errors of Metabolism in adults,
Gabriella Horvath, MD, PhD, Prof. Division of Biochemical Genetics,
Department of Pediatrics, University of British Columbia, BC Children's
Hospital, BC Children's Hospital Research Institute, Vancouver, Canada

10.00 – 10.30 Inborn errors of metabolism in adults,
Anna Lehman, MD, PhD, Adult Metabolic Diseases Clinic at Vancouver
General Hospital, Medical Genetics - University of British Columbia,
Vancouver, Canada

10.30 – 10.45 Rare causes of stroke – Fabry disease,
Adina Stan, MD, PhD, Lecturer, Neuroscience Department, “I. Hațieganu”
University of Medicine and Pharmacy Cluj-Napoca, Romania

10.45 – 11.00 Spastic paraparesis – a challenging diagnosis,
Hanna Dragoș, MD, Adina Stan, MD, PhD
Neuroscience Department, “I. Hațieganu” University of Medicine and Pharmacy
Cluj-Napoca, Romania

11.00 – 11.20 COFFEE BREAK, Poster viewing

30 JUNE 2023

Chairs: Laura Cif, Birgit Assmann

- 11.20 – 12.05 Movement disorders in Inborn Errors of Metabolism,**
Birgit Assmann, MD, PhD, Neuropediatrics and Metabolic Medicine Section,
Heidelberg, Germany
- 12.05 – 12.15 Developmental and epileptic encephalopathy associating epilepsy,
dyskinesia and ASD in a child with ALG1 mutation (CDG-Ik),**
Mihaela Adela Vințan, MD, PhD, Lecturer, Child Neurology Department, “I.
Hațieganu” University of Medicine and Pharmacy Cluj-Napoca, Children's Clinical
Emergency Hospital Cluj Napoca, Romania
- 12.15 – 13.00 Treatment of dystonia with Deep Brain Stimulation,**
Laura Cif, MD, PhD, Consultant in the Unit of Functional Neurosurgery Unit in
Movement Disorders, Unité “Pathologies Cérébrales Résistantes”, Department of
Neurosurgery, University Hospital, CHRU, Montpellier University Hospital
Montpellier, France
- 13.00 – 13.20 Workshop with cases who had Deep Brain Stimulation presented
from Vancouver,**
Gabriella Horvath, MD, PhD, Prof. Division of Biochemical Genetics, Department
of Pediatrics, University of British Columbia, BC Children's Hospital, BC Children's
Hospital Research Institute, Vancouver, Canada
- 13.20 – 14.45 LUNCH BREAK**

30 JUNE 2023

Chairs: Laura Damian, Gabriella Horvath

14.45 – 15.00 Session THE PATIENT’S VOICE,

The need for the extension of neonatal screening from the patients’ perspective,
Dorica Dan, President of Romanian Prader Willi Association, president of Romanian National Alliance for Rare Diseases

“AT THE CROSSROADS BETWEEN METABOLISM AND AUTOINFLAMMATORY DISORDERS”,

session coordinated by **Dr. Laura Damian and Dr. Romana Vulturar**

15.00 – 15.20

The Mentors in Rare Disease Award 2023

Honouring **Assoc. Prof. Dr. Ioan Parasca**

Laudatio: **Prof. Dr. Simona Rednic**

1st Part

Chairs: Simona Rednic, Călin Lazăr

15.20 – 15.40

At the interface between metabolism and autoinflammatory diseases,

Laura Damian, MD, PhD, Department of Rheumatology, Emergency Clinical County Hospital Cluj, Romania

15.40 – 16.00

SOBI SYMPOSIUM: Interleukin-1 as a therapeutic target: focus on pediatric inflammatory diseases,

Mihaela Spârchez, MD, PhD, Lecturer, “I. Hațieganu” University of Medicine and Pharmacy Cluj-Napoca, Romania, IInd Pediatric Clinic, Clinical Emergency Hospital for Children, Cluj-Napoca, Romania

16.00 – 16.20

The deficiency of adenosine deaminase 2 (DADA2),

Ciprian Jurcuț, MD, PhD, Department of Internal Medicine, Carol Davila Central Military Emergency Hospital, Bucharest, Romania

16.20 – 16.40

The mevalonate kinase deficiency,

Alina Mara, MD, Astra Sibiu Polyclinic, Romania

16.40 – 17.00

COFFEE BREAK, Posters viewing

30 JUNE 2023

2nd Part

Chairs: Mihaela Spârchez, Ciprian Jurcuț

17.00 – 17.20 ***NOVARTIS SYMPOSIUM***

Anti-interleukin-1 therapy in rare systemic autoinflammatory diseases,

Laura Damian MD, PhD, Department of Rheumatology, Emergency Clinical County Hospital Cluj, Romania

17.20 – 17.40 **Recurrent fever in inherited errors of metabolism,**

Cecilia Lazea MD, PhD, Assoc. Prof., “I. Hațieganu” University of Medicine and Pharmacy Cluj-Napoca, Ist Pediatric clinic, Clinical Emergency Hospital for Children, Cluj-Napoca, Romania

17.40 – 18.00 **Colchicine in autoinflammatory diseases,**

Ileana Filipescu MD, PhD, Lecturer, “I. Hațieganu” University of Medicine and Pharmacy Cluj-Napoca, Department of Rheumatology, Emergency Clinical County Hospital Cluj, Romania

18.00 – 18.20 ***Metabolic mimics of juvenile idiopathic arthritis,***

Maria-Magdalena Tămaș MD, PhD, Lecturer, “I. Hațieganu” University of Medicine and Pharmacy Cluj-Napoca, Department of Rheumatology, Emergency Clinical County Hospital Cluj, Romania

18.20 **Discussions, posters evaluation and awards, closure.**

We thank to our partners

