UMF
UNIVERSITATEA DE MEDICINĂ ȘI FARMACIE
IULIU HAȚIEGANU
CLUJ-NAPOCA

SRGM
Societatea Romana de Genetica Medicala

ASOCIAȚIA
ȘANSA UNUI COPIL

Symposium
Inborn errors of metabolism in pediatric and adult pathology
with international participation

Bolile genetice de metabolism în patologia copilului și a adultului

Cluj-Napoca, Centrul Imogen, SCJU Cluj,
Str. Pasteur f.n.
(lângă Cl. Neurochirurgie)

7-8 June 2019, h 9.^{00}

Registration at: bgmorganizare@gmail.com

Manifestation accredited by the College of Physicians in Romania through EMC points.
Scientific coordinators:
Assoc. Prof. Gabriella Horvath (Vancouver), Dr. Paula Avram (Londra), Dr. Laura Damian (Cluj), Dr. Romana Vulturar (Dept. of Molecular Sciences. UMP Cluj, Imogen Cluj-Napoca)
Organizers:
Dr. Romana Vulturar (UMP Cluj-Napoca, Imogen, SCJU Cluj), Dr. Laura Damian (SCJU Cluj), Dr. Tunde Kovacs (UMP Cluj-Napoca, Imogen SCJU Cluj), Dr. Adina Chiș (Dept. of Molecular Sciences, UMP Cluj-Napoca).

7 June 2019

8.30-9,00 - ***Registration***

9,00-9,15 - ***Welcoming participants***, messages from UMP Cluj-Napoca, Imogen Center, Cluj County Hospital (Dr. Tunde Kovacs), Ministerial counselor for rare diseases (Mrs. Lidia Onofrei)

9,15-9,35 - Inborn errors of metabolism – a pathology affecting pediatric and adult patients: lessons to learn from several perspectives, *Romana Vulturar*, UMP Cluj-Napoca, Imogen – SCJU Cluj, Cognitive Neuroscience Laboratory- UBB Cluj-Napoca

9,35-10,05 - ***Metabolic emergencies in pediatrics,*** *Paula Avram*, Paediatric Intensive Care Consultant, Sheffield Children's Hospital, London, UK, Director Non-Governmental Organization *'Asociația 'Șansa unui copil'*

10,05-10,40 - ***Inborn errors of metabolism in adults,*** *Gabriela Horvath*, Clinical Assoc. Prof, Division of Biochemical Diseases - children and adults, Univ. of British Columbia, Vancouver, Canada

10,40-11,10 - ***Coffee break***

11,10-12,00 - ***At the interface of inborn errors of metabolism and rare auto-inflammatory disorders***, *Laura Damian*, Centre for Rare Musculoskeletal Auto-immune and Auto-inflammatory Diseases, SCJU Cluj

Awarding Mrs. Dr. Anca Cristea, a reference name in the introduction of diagnosis of rare diseases with immune mechanism in Romania

12,00-12,40 - ***Inborn errors of metabolism- implications in Obstetrics-Gynaecology***; ***link with the Materno-fetal medicine project***, *Gabriela Horvath*, Clinical Assoc. Prof, Division of Biochemical Diseases - children and adults, Univ. of British Columbia, Vancouver, Canada

12,40-13,30 - ***Lunch***

13,30-12,40 - ***Recommendations for the management of Tyrosinemia type I***

13,40 -14,00 - ***Early clinical manifestations in mucopolysaccharidosis,***

Camelia Al-Khzouz, Cecilia Lazea, Simona Bucerzan, Ioana Nașcu, Carmen Asăvoaie, Genetic Dept., Children Cluj County Emergency Hospital, UMP Cluj-Napoca

14,00-14,20 - ***Cardiac involvement and enzyme replacement therapy results in children with mucopolysaccharidosis,***

Cecilia Lazea, C. Al-Khzouz, S.Bucerzan, C. Asăvoaie, P.Grigoresc-Sido Ist Pediatric Clinic, UMP Cluj-Napoca

14,20-14,40 – Gaucher disease in Romanian patients,

Simona Bucerzan, Drugan C, Nascu I, Alkhzouz C, Crisan M, Zimmermann A, Lazea C, Lazar C, Popp RA, Miclea D, Asavoaie C, Grigorescu-Sido P, Genetic Dept., Children Cluj County Emergency Hospital, UMP Cluj-Napoca

7 June 2019

14,40 -15,10 - ***Genotype-phenotype evaluations in a group of Phenylketonuria Romanian patients. Is PKU a solved problem?***

Monica Mager (child neurologist –UMP Cluj-Napoca) R. Vulturar, S. Palade, C. Pantelimon

15,10-15,30 - ***Coffee break***

15,30-16,00 - ***Functional tests of variants of unknown significance in Inborn Errors of Metabolism,*** *Drd. Ana Pop,* Amsterdam UMC, VUmc, Department of Clinical Chemistry, Metabolic Unit, Amsterdam, The Netherlands

16,00-17,15 - ***Workshop, Case discussions***

1. A case of severe hypertrophic cardiomyopathy,
Cecilia Lazea, S. Oprita, C. Al-Khzouz, D. Miclea

2. „Spondylitis" and scleral pigmentation: alkaptonuria,
Bianca Jurjiu, M. Rinzis, A. Ungureanu, A. Iliescu, I. Felea, S. Rednic, R. Vulturar, L. Damian

3. Movement disorder and associated epileptic spasms- neurotransmitters deficiency?
Cristina Pantelimon, M. Mager

4. Type 1 glutaric aciduria- case presentation,
Monica Mager, C. Pantelimon, A. Resiga

5. Hepatocarcinoma with tumoral thrombosis invading the right athrium in a patient with hereditary hemochromatosis and cirrhosis,
Olga Orășan, A. Cozma, A. Sitar-Tăut

6. The rhythm disorders and carnitine deficiency,
Simona Căinap, B. Lucian, G. Tita, G. Cismaru, T. Pop

7. Dopa –sensitive dystonia?- case presentation,
Monica Mager, C. Pantelimon, A. Resiga

8. Malignant HTA –where to proceed with the investigations? Simona Căinap, B. Simionescu, G. Tita, O. Fufezan

17,15-17,30 - ***Eurordis, Holistic care of patients with rare diseases, Achieving Holistic Person-centred care to leave no one behind***, *Dorica Dan,* APWR president, ARCrare, Board member - Eurordis, CES member, CNBR, Centrul NoRo coordinator

19,00 - ***Dinner***

8 June 2019

8.30-9,00 - ***Registration***, ***Welcoming participants***

9,00-9,30 - ***Deep Brain Stimulation in pediatric movement disorders, focus on inborn errors of metabolism***, *Laura Cif*, Université de Montpellier, France

9,30-10,00 - ***Organic acidurias***, *Paula Avram,* Paediatric Intensive Care Consultant, Sheffield Children's Hospital, London, UK, Director Non-Governmental Organization *'Asociația 'Șansa unui copil'*

10,00-10,40 - ***Screening and recommandations in treatable metabolic disorders***, *Gabriella Horvath*, Clinical Assoc. Prof, Division of Biochemical Diseases - children and adults, Vancouver, Canada

10,40-11,10 - ***Expanded Newborn screening: implications and goals, simple or complex?*** *Isabela Țârcomnicu,* Cytogenomic Medical Laboratory, București

11,10-11,40 - ***Coffee break***

11,40-12,15 - ***Urea cycle disorders: from diagnosis to recent advances***, *Emanuela Manea,* Paediatric Consultant with interest in Metabolic disorders, Great Ormond Street Hospital for children, London, UK

12,15-12,30 - ***Treatment in hyperammonemias***

12,30-13,00 - ***Approach to neurometabolic disorders manifesting with epilepsy and movement disorders,*** *Gabriella Horvath,* Clinical Assoc. Prof, Division of Biochemical Diseases - children and adults, Vancouver, Canada

13,00-14,00 – Lunch

14,00-14,20 - ***From Bedside to Bench and Beyond: our experience in diagnostic of small molecules defects***, ***including NMR spectroscopy method,***

Romana Vulturar, A. Nicolescu, A. Chiș, C. Deleanu, UMP Cluj, Biospectroscopy Group Inst. of Macromolec Chemistry Iasi, Romanian Acad Inst. of Biochemistry, Bucharest

14,20-14,40 - ***X-linked hypophosphatemic rickets – diagnosis and current standard of care***, *Daniela Iacob*, IIIrd Pdiatric Clinic, University of Medicine and Pharmacy Cluj-Napoca

14,40-15,00 - ***Multidisciplinary approach to early diagnosis of rare diseases. Case report,*** *Bogdan Chiș*, Adult IInd Medical Clinic, SCJU Cluj

15,00-15,10 - ***Deficiency of alpha – manosidase, a treatable disease?***

Romana Vulturar, UMP Cluj-Napoca, Imogen – SCJU Cluj, Cognitive Neuroscience Laboratory- UBB Cluj-Napoca

8 June 2019

15,10 -16,00 - Workshop, Case discussions

1. ***Videocapilaroscopy- the diagnostic key in a rare pathology,***
Ileana Filipescu, S. Rednic, S.-P. Simon, C. Pamfil, L. Muntean, L. Damian
2. ***A case of recurrent stress fractures: hypophosphatasia***,
Bianca Balan, P. Vele, S.-P. Simon, C. Bolоșiu, D. Fodor, L. Damian
3. ***Refsum disease – a chameleon in clinical practice*** , Anca Moldovan, I. Felea
4. ***Severe hypercalcemia – a cause of hypotonia in infancy,***
Simona Căinap, O. Fufezan, E. Francesco
5. ***Abdominal pain and psychosis: lupus or homocysteinemia?***
Agnes Ungureanu, L. Damian, B. Jurjiu, L. Bene, S. Rednic
6. ***Rare causes of choreoathetosis***,
Ioana Stanescu, N. Radics, M. Patiu, L. Damian,
7. ***The eye of a perfect storm: acute motor axonal neuropathy in hypobetalipoproteinemia***
Laura Damian, V. Todea, F. Moldovan, M. M. Tamas, G. Gușetu, S. Rednic, R. Vulturar
8. ***Inaugural hemophagocytic syndrome- in search for etiology***,
Lucia Herbel, M. Lupse, L. Urian, A. Bojan, O. Antal, M. Sparchez, C. Lazăr, R. Vulturar, L. Damian
9. ***Amyloid angiopathy – a metabolic disorder?***
Adina Stan, C. Cismaru, L. Damian
10. ***Recurrent rhabdomyolysis- which is the diagnostic approach?***
Laura Damian, I. Kacso, L. Rogojan, L. Bene, D. Miclea, S. Rednic, R. Vulturar
11. ***Real or induced Wilson Disease – in a 20 years old woman***,
Adela Sitar-Tăut, A. Cozma, O. Orășan, A. Fodor, R. Suharoschi, S. Secară, I. Popovici, V. Negrean, D. Sâmpelean
12. ***STH deficiency, nephrocalcinosis, polyuric-polydipsic syndrome***,
Daniela Iacob, Rodica Corneanu.

18,00 - Dinner