



VII International Scientific Conference

“Research brings hope to people living with a rare disease”

March 17, 2017, Crowne Plaza Hotel, M.K.Čiurlionio str.84, Vilnius

www.rarediseases.lt

Programme

8:00 – 9:00 Registration

Plenary session (in English)

Chairpersons: E. Jurevičienė, MD, assoc. prof. R. Čerkauskienė

9:00 – 9:15	Opening speech	Prof. A. Utkus Prof. V. Usonis Assoc. prof. R. Čerkauskienė
9:15 – 9:30	European reference networks - new possibilities to share, care and cure	J. Januševičienė
9:30 – 9:45	An overview screening and treatment in Lithuania	Prof. A. Laucevičius
9:45 – 10:00	Rare diseases: together we are stronger	E. Jurevičienė , MD Assoc.prof. R.Čerkauskienė B. Tumienė, MD
10:00 – 10:50	LSD treatment: an Update	Prof. M. Beck
10:50 – 11:30	Clinical studies in LAL-D	B. Skölvling, MD, PhD
11:30 – 12:00 Coffee & Tea		

Plenary session (in Lithuanian)
Chairpersons: prof. Ž. Petrulionienė, prof. A. Utkus

12:00 – 12:20	Molecular Testing in Dyslipidemias	Prof. A.Utkus
12:20 – 12:40	Familial Homozygous and Heterozygous Hypercholesterolemia: a Review	Prof. Ž. Petrulionienė
12:40 -13:00	Innovations in Severe Hyperlipidemia Treatment: Statins, Combined Preparations, PCSK9 Inhibitors	Assoc. prof. V. Dzenkevičiūtė
13:00-13:20	Severe Hyperlipidemia: Screening and Diagnostic Algorithm	S. Kutkienė, MD
13:20 – 13:40	Biochemical testing in lipid disorders	D.Karčiauskaitė, MD, PhD
13:40 – 14:00	Discussion	
14:00 – 15:00 Lunch		
Part A, (in Lithuanian) Chairpersons: prof. A. Jankauskienė, prof. M. Miglinas.		
15:00 – 15:25	Apolipoprotein L1-Associated Nephropathy	Prof. M. Miglinas
15:25 – 15:50	Relation Between Obesity and Renal Diseases in Childhood	Prof. A. Jankauskienė
15:50 – 16:15	Fatty Acids: Metabolism and Rare Disorders	B. Skerlienė, MD, PhD
16:15 – 16:40	Causes of dyslipidemia in childhood: experience of a single center	V.Černiauskienė, MD M.Jakutovič, MD Assoc.prof. R.Čerkauskienė
16:40 – 17:05	Influence of Microbiota on Lipid Metabolism	Assoc. prof. V. Urbonas
17:05 – 17:30	Discussion	

B dalis rezidentų ir doktorantų sekcija (anglų kalba)

Moderatoriai: prof. M. Beck, prof. dr. A.Utkus

15:00 – 15:15	Diagnostic challenge: two cases of necrotizing encephalopathy in childhood	<u>Dovilė Jonuškaitė</u> , Rūta Praninskienė, Gintaras Michailovas, Rimantė Čerkauskienė, Eugen Boltshauser
15:15 – 15:30	Differential diagnosis of bowel obstruction in a case of systemic sclerosis	<u>Eglė Dieninytė</u> , Rita Rugienė
15:30 – 15:45	Upshaw – Schulman syndrome in a posttransplant patient	<u>Ernesta Mačionienė</u> , Marius Miglinas
15:45 – 16:00	Gender related prophylaxis of life threatening ventricular arrhythmias according to genetic type of long QT syndrome	Dovilė Jančauskaitė, <u>Lina Gedvilaitė</u> , Jūratė Barysienė, Živilė Katliorienė, Violeta Mikštienė
16:00 – 16:15	Acute kidney injury in extremely low birth weight infant with the nephrolithiasis	Rasa Garunkštienė, Rimutė Vaitkevičienė, Ieva Paulavičienė, Nijolė Drazdienė, Rimantė Čerkauskienė
16:15 – 16:30	Autoimmune polyendocrine syndrome with recurrent serositis	<u>Paulius Kalibatas</u> , Skaistė Sendžikaitė, Rita Sudikienė
16:30 – 16:45	Homozygous familial hypercholesterolemia – case report	<u>Eglė Skiauterytė</u> , Žaneta Petrulionienė, Antanas Griškevičius, Sandra Kutkienė
16:45 – 17:00	BCG-associated osteomyelitis in a 12 month-old girl	<u>Lina Radevičiūtė</u> , Inga Ivaškevičienė, Elena Sučilienė, Šarūnas Bernotas, Virgilijus Urbanavičius, Vytautas Usonis
17:00 - 17:15	Niemann-Pick Type C1 – neurological manifestation of a lysosomal storage disease	<u>Agnė Ulytė</u> , R. Kaladytė-Lokominienė, A. Matulevičienė, L. Kupčinskas, L. Dvorakova, M. Hnizdova Bouckova, H. Klimentova, H. Vlaskova, V. Kozich, H.Y. Wu, K. Brammeier, H. Church
17:15 – 17:30	Discussion	
17:30 – 18:30	Poster presentation	

INVITED SPEAKERS

Prof. M. Beck

University Medical Center, Johannes
Gutenberg-University of Mainz, Germany

Assoc. prof. R. Čerkauskienė

Head of Coordinating Centre for Rare
Children's Diseases, Children's Hospital,
Affil. of VUH SK

V. Černiauskienė, MD

Doctor pediatric cardiologist, Children's
Hospital, Affil. of VUH SK

Assoc. prof. V. Dzenkevičiūtė

Cardiology and Angiology Department's
doctor cardiologist, VUH SK

M. Jakutovič, MD

Doctor pediatric cardiologist, Children's
Hospital, Affil. of VUH SK

Prof. A. Jankauskienė

Head of Pediatric centre, Children's
Hospital, Affil. of VUH SK

J. Januševičienė

Director of Health care resources and
innovation management
department, Ministry of Health of the
Republic of Lithuania

E. Jurevičienė, MD

Managing director of VUH SK, Head of
Coordinating Centre for Adult's Rare
Diseases

D. Karčiauskaitė, MD, PhD

Centre's for Laboratory Medicine doctor,
VUH SK

S. Kutkienė, MD

Doctor cardiologist, Cardiology and
Angiology Department, VUH SK

Prof. A. Laucevičius

Head of Clinic of Cardiovascular diseases,
Vilnius University Faculty of Medicine

Prof. M. Miglinas

Head of Nephrology Department,
VUH SK

Prof. Ž. Petrulionienė

Head of I Cardiology Department, Vilnius
University, Centre of Cardiology and
Angiology, VUH SK

B. Skerlienė, MD, PhD

Doctor of rare pediatric diseases,
Children's Hospital, Affil. of VUH SK

B. Skölvig, MD, PhD

Medical Affairs Manager „Metabolics“,
Sweden

B. Tumienė, MD

Coordinator for Rare Disease
Competence Centres, VUH SK

Assoc. prof. V. Urbonas

Doctor pediatric gastroenterologist,
Children's Hospital, Affil. of VUH SK

Prof. V. Usonis

Head of Clinic of Children's Diseases,
Vilnius University Faculty of Medicine

Prof. A. Utkus

Dean of the Faculty of Medicine, Vilnius
University, Head of Centre for Medical
Genetics, VUH SK