



14th International
Rare Diseases Conference
“Rare Diseases - our task”
July 1-3, 2016

Conference Programme

Thursday, June 30, 2016

Lunch		13:00 - 14:00
Registration		14:00 - 15:00
Session for patient organizations and members of Federation of Rare Diseases Patients in Central and Eastern Europe		
Teresa Matulka Anna Tylki-Szymańska	Welcome and opening	15.00 - 15.10
Presentations of experts who will share their knowledge and experience		15.10 - 16.00
Who will take care of us? Patients with rare diseases debate - with experts from the health care system, government officials and representatives of patients' organizations		16.00 - 17.00
Questions and answers		17.00 - 18.00
Hotelu Allianz	Welcome dinner for invited conference guests	19.00 - 22:00

Friday, July 01, 2016

Breakfast		7:00 - 8:30
Registration		8:00 - 9:00
Teresa Matulka Anna Tylki-Szymańska	Welcome address	9:00 - 9:10
Session I		
Chairs:	Johannes Berger, Ekaterina Zakharova	
Marc Dooms , <i>Leuven, Belgium</i>	Rembert Dodoens (1517-1585): Pioneer in Rare Diseases and Orphan	9:10 - 9:25
Timothy Cox <i>Cambridge, UK</i>	Gaucher disease: a treasure-house of knowledge	9:25 - 9:50
Ségolène Aymé , <i>Paris France</i>	State of Play of research and health services for rare diseases in Europe	9:50-10:10
Marc Dooms , <i>Leuven, Belgium</i>	Cross-Border Healthcare and Rare Diseases	10:10-10:30
Olga Amaral <i>Porto, Portugal</i>	Foreseen accomplishments through precision cellular and molecular biology	10:30-10:45
Coffee break		10:45-11:10
Session II		
Chairs:	Ségolène Aymé, Jörn Oliver Sass	
Sylvia Stockler <i>Vancouver, Canada</i>	Diagnostic approach to neurodevelopmental disabilities (NDD): from a traditional metabolic to an -omics perspective	11:10-11:50
Henk Blom <i>Freiburg, Germany</i>	Homocystinuria and epigenetic modifications.	11:50-12:15



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Marie T. Vanier <i>Lyon, France</i>	Niemann-Pick disease type C (NP-C) and Acid Sphingomyelinase deficiencies (ASMD): evolution of laboratory diagnostic strategies and the place of plasma biomarkers profiles	12:20-12:40
Ladislav Kuchař <i>Praha, Czech Republic</i>	Differential screening of Niemann-Pick diseases type A/B and type C: benefits of co-measurement of lysosphingomyelin and its 509 analogue by LC-MS/MS	12:40-13:00
Lunch		13:00-14:00

Session III

Chairs:	Marie-T. Vanier, Henk Blom	
Jörn Oliver Sass <i>Bonn-Rhein-Sieg, Germany</i>	5-oxoprolinuria (pyroglutamic aciduria) and OPLAH mutations	14:00-14:25
Johannes Berger, <i>Vienna, Austria</i>	Comparative thoughts on Metachromatic Leukodystrophy and X-linked Adrenoleukodystrophy	14:25-14:50
Tomasz Kmieć <i>Warsaw, Poland</i>	Analysis of phenotype and genotype of PKAN and MPAN types in NBIA group (formerly Hallervorden-Spatz disease) in Poland	14:50-15:10
David J. Begley <i>London, UK</i>	Blood-Brain Barrier Changes in MPSIIIA Mice.	15:10-15:35
Shunji Tomatsu <i>Wilmington, USA</i>	Neonatal cellular and gene therapies for mucopolysaccharidoses: the earlier the better?	15:35-16:00
Coffee break		16:00-16:30

Session IV

Chairs:	David J. Begley, Hanna Mierzweska	
Sara Mole <i>London, UK</i>	Batten disease – an update	16:30 -16:50
Tomasz Kmieć <i>Warsaw, Poland</i>	Case of 10-years old girl with very slow progressive disturbance of walk and visual difficulties with variant neuronal ceroidlipofuscinosis type 2 (vNCL2)	16:50 -17:05
Małgorzata Bednarska-Makaruk <i>Warsaw, Poland</i>	The tripeptidyl peptidase 1 (TPP1) deficiency in 36-years old patient with cerebellar-extrapyrmidal syndrome and dilated cardiomyopathy"	17:05 -17:20
Katarzyna Hetmańczyk <i>Warsaw, Poland</i>	Deterioration of visual acuity as the first sign in 5 yrs old boy with ceroidlipofuscinosis type 3"	17:20 -17:35
Izabela Michałus <i>Białystok, Poland</i>	Hypophosphatasia – clinical features and new methods of treatment	17:35 -17:50
Grill-dinner		20:00

Saturday, July 2, 2016

Breakfast	7:00 - 8:30
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Session V

Chairs:	Timothy Cox, Zita Krumina	
Sylvia Stockler <i>Vancouver, Canada</i>	Diagnosis and Treatment of Cerebral Creatine Deficiency Disorders	8:35 – 9:10
Hanna Mierzweska <i>Warsaw, Poland</i>	Diseases with basal ganglia lesions visible in neurovisual examinations The	9:10 - 9:25
Dariusz Rokicki <i>Warsaw, Poland</i>	Specificity of MR imaging in diagnosis of inborn neurodegeneration disorders	9:25 - 9:40



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Nataliia Pichkur Kyiv, Ukraina	Manifestation of Fanconi Renal Syndrom in the Inherited Metabolic Diseases	9:40- 10:00
Natalia Rumiantseva, Minsk, Belarus	Genetic disorders manifested by exocrine pancreatic insufficiency: clinical characterization and genetic counseling	10:00 – 10:15
Adam Golda Gliwice, Poland	Cardiological aspects of mucopolysaccharidoses	10:15 -10:30
Assel Tulebayeva Almaty, Kazakhstan	Multifactorial causes of respiratory dysfunction in children with mucopolysaccharidosis	10:30-10:45
Nesrin Karabul Bochum, Germany	New developments in treatments of LSDs – Chaperone therapy	10:45-11:00
Coffee break		11:00-11:20
Session VI		
Chairs:	Sylvia Stockler, Johannes Berger,	
Ekaterina Y Zakharova Moscow, Russia	Diagnostic scenarios for inherited metabolic diseases	11:20 - 11:40
Jörn Oliver Sass Bonn-Rhein-Sieg, Germany	Inborn Errors of Metabolism: Laboratory Curiosity or Real Disease?	11:40 - 12:10
Nataliia Olkhovych Kyiv, Ukraina	The Pseudodeficiency of Lysosomal Enzymes	12:10 - 12:40
		12:40 - 13:00
Lunch		13:00 - 14:00
Session VII Dysmorphology meeting		
Moderators:	Ewa Pronicka, Anna Tyłki-Szymańska	
Saskia B. Wortmann Salzburg, Austria	3-methylglutaconic aciduria - your key to diagnosis Whole exome sequencing for diagnosis of rare neurological diseases in the Polish population – novel mutations, genes, diseases	14:00- 14:15
Rafal Płoski Warsaw, Poland	Whole exome sequencing for diagnosis of rare neurological diseases in the Polish population – novel mutations, genes, diseases	14:15-14:30
Robert Śmigiel Wrocław, Poland	NGS application in <i>post mortem</i> diagnosis – cases analysis	14:30-14:45
Jacek Pilch Katowice, Poland	Severe, paroxysmal, familial myoglobinuria caused by mutation in <i>LPIN1</i> gene. Whether must be lethal?	14:45-15:00
Dorota Karczmarewicz Warsaw Poland	Orphanet in everyday clinical practice	15:00-15:15
Lunch		15:15-15:45
Dysmorphology meeting (continued)		
Moderators:	Ewa Obersztyn, Robert Śmigiel	15:45 -17:45
Holy Mass in intention of patients with rare diseases and their families		18.00
Gala Dinner		20:00
Sunday , July 3, 2016		
Breakfast		7:30 – 8:30



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Session VIII

Moderators:	Marc Dooms, Saskia Wortman	15:00 -18:00
Arndt Rolfs <i>Rostock, Germany</i>	TBD	9:00 - 9:20
Grzegorz Węgrzyn <i>Gdańsk, Poland</i>	Flavonoids in lysosomal storage diseases	9:20 - 9:40
Karolina Pierzynowska <i>Gdańsk, Poland</i>	Genistein-mediated lysosome stimulation as a novel approach for the treatment of Huntington's disease	9:40 - 9:55
Anna Tylki Szymańska <i>Warszawa, Poland</i>	Transaldolase (TALDO) deficiency	9:55 -10:10
Maciej Machaczka <i>Stockholm Sweden</i>	The megaloblastic anaemia in the course of Lesch-Nyhan syndrome	
Svetlana Volgina <i>Kazan Russia</i>	Rett syndrome in children. Case report	10:25 - 10:40
Coffee break		10:40 - 11:00

Session IX

Chairs:	Zita Krumina, Grzegorz Węgrzyn	
Ivanka Sinigerska <i>Sofia, Bulgaria</i>	Chitotriosidase – a useful biomarker in the diagnostic approach to LSD	11:00-11:20
Elzbieta Szczepanik <i>Warsaw, Poland</i>	Glucose transporter deficiency type 1 – rare but treatable cause of refractory epilepsy and neurodevelopmental disorders.	11:20-11:40
Agnieszka Ługowska <i>Warsaw, Poland</i>	Laboratory diagnostics of mucopolysaccharidoses	11:40-12:00
Hanna Mierzewska <i>Warsaw, Poland</i>	4H syndrome – POLR3A and POLR3B genes deficits..	12:00-12:20
Anna Jakubiuk-Tomaszuk <i>Białystok, Poland</i>	Clinical evolvement of leukodystrophy - hypomyelination with atrophy of the basal ganglia and cerebellum - a clinical report of the 18-years old boy.	12:20-12:40
Anna Tylki Szymańska Teresa Matulka	Closing remarks and summary,	12:40-13:00
Lunch		13:00 - 14:00
Medical consultations		14:30-17:00
Consultations of unknown cases		14:30-17:00
Dinner		18:00

Monday, July 4, 2016

Breakfast		7:00 -8:30
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**Session for Patient Organizations
and members of the Federation of Patients with Rare Diseases in Central and Eastern Europe**

	The benefits of the cooperation of patients organisations - exchange of experience	10:00 - 12:00
Teresa Matulka	Closing remarks and summary	

Transfer to the airport	 	
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