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, *Leuven, Belgium*	Rembert Dodoens (1517-1585): Pioneer in Rare Diseases and Orphan	*9:10 - 9:25*
Timothy Cox *Cambridge, UK*	Gaucher disease: a treasure-house of knowledge	*9:25 -9:50*
Ségolène Aymé, *Paris France*	State of Play of research and health services for rare diseases in Europe	*9:50-10:10*
Marc Dooms, *Leuven, Belgium*	Cross-Border Healthcare and Rare Diseases	*10:10-10:30*
Olga Amaral *Porto, Portugal*	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 *Vancouver, Canada*	Diagnostic approach to neurodevelopmental disabilities (NDD): from a traditional metabolic to an -omics perspective	*11:10-11:50*
Henk Blom Freiburg, Germany	Homocystinuria and epigenetic modifications.	*11:50-12:15*

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Marie T. Vanier *Lyon, France*	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 *Praha, Czech Republic*	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 Bonn-Rhein-Sieg,Germany	5-oxoprolinuria (pyroglutamic aciduria) and OPLAH mutations	*14:00-14:25*
Johannes Berger, *Vienna, Austria*	Comparative thoughts on Metachromatic Leukodystrophy and X-linked Adrenoleukodystrophy	*14:25-14:50*
Tomasz Kmieć *Warsaw, Poland*	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 *London, UK*	Blood-Brain Barrier Changes in MPSIIIA Mice.	*15:10-15:35*
Shunji Tomatsu *Wilmington, USA*	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 Mierzewska**	
Sara Mole *London, UK*	Batten disease – an update	*16:30 -16:50*
Tomasz Kmieć *Warsaw, Poland*	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 *Warsaw, Poland*	The tripeptidyl peptidase 1 (TPP1) deficiency in 36-years old patient with cerebellar-extrapyramidal syndrome and dilated cardiomyopathy"	*17:05 -17:20*
Katarzyna Hetmańczyk *Warsaw, Poland*	Deterioration of visual acuity as the first sign in 5 yrs old boy with ceroidlipofuscinosis type 3"	*17:20 -17:35*
Izabela Michałus *Białystok, Poland*	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***
	Session V	
Chairs:	**Timothy Cox, Zita Krumina**	
Sylvia Stockler *Vancouver, Canada*	Diagnosis and Treatment of Cerebral Creatine Deficiency Disorders	*8:35 – 9:10*
Hanna Mierzewska *Warsaw, Poland*	Diseases with basal ganglia lesions visible in neurovisual examinations The	*9:10 - 9:25*
Dariusz Rokicki Warsaw, Poland	Specificity of MR imaging in diagnosis of inborn neurodegeneration disorders	*9:25 - 9:40*

Nataliia Pichkur Kyiv, Ukraina	Manifestation of Fanconi Renal Syndrom in the Inherited Metabolic Diseases	*9:40- 10:00*
Natallia Rumiantsava, *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 Errorrs 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 Tylki-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 *post mortem* diagnosis – cases analysis**	*14:30-14:45*
Jacek Pilch *Katowice, Poland*	Severe, paroxysmal, familial myoglobinuria caused by mutation in *LPIN1* 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 *Rostock, Germany*	TBD	*9:00 - 9:20*
Grzegorz Węgrzyn *Gdańsk, Poland*	Flavonoids in lysosomal storage diseases	*9:20 - 9:40*
Karolina Pierzynowska *Gdańsk, Poland*	Genistein-mediated lysosome stimulation as a novel approach for the treatment of Huntington's disease	*9:40 - 9:55*
Anna Tylki Szymańska *Warszawa, Poland*	Transaldolase (TALDO) deficiency	*9:55 -10:10*
Maciej Machaczka *Stockholm Sweden*	The megaloblastic anaemia in the course of Lesch-Nyhan syndrome	
Svetlana Volgina *Kazan Russia*	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 *Sofia, Bulgaria*	Chitotriosidase – a useful biomarker in the diagnostic approach to LSD	*11:00-11:20*
Elzbieta Szczepanik *Warsaw, Poland*	Glucose transporter deficiency type 1 – rare but treatable cause of refractory epilepsy and neurodevelopmental disorders.	*11:20-11:40*
Agnieszka Ługowska *Warsaw, Poland*	Laboratory diagnostics of mucopolysaccharidoses	*11:40-12:00*
Hanna Mierzewska *Warsaw, Poland*	4H syndrome – POLR3A and POLR3B genes deficits..	*12:00-12:20*
Anna Jakubiuk-Tomaszuk, *Białystok, Poland*	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***
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