PROGRAM

13th International Conference On Rare Diseases
"Rare Diseases Crossing Borders Together"

Thursday, July 02, 2015		
Lunch		***13:00 - 14:00***
	Registration	*14:00 - 16:00*
Inauguration of the 25th anniversary of the Association of Mucopolysaccharidosis (MPS) and Rare Diseases		
Teresa Matulka **Anna Tylki-Szymańska** **Barbara Czartoryska**	Opening ceremony	*16:00 - 16:30*
Teresa Matulka	Summary of 25th Anniversary of the Association of Mucopolysaccharidosis (MPS) and Rare Diseases	*16:30 - 17:00*
Marek Michalak Minister	Address from Ombudsman for Children,	*17:00 - 17:15*
Jarosław Waligóra European Commission	EU policy on rare diseseaes	*17:15 - 17:30*
Anna Wyszkoni	Singing	*17:30 - 18:00*
Volkmar Gieselmann	Evaluation of treatment options in mouse models of leukodystrophies	*18:00 - 18:20*
	Holy Mass in intention of patients with rare diseases and their families	*18:30*
Welcome dinner, Allianz Hotel		***20:00***
Friday, July 03, 2015		
Breakfast		***7:00 - 8:30***
	Registration	*8:00 - 9:00*
Teresa Matulka **Barbara Czartoryska** **Anna Tylki-Szymańska**	Welcome address	*9:00 - 9:10*
Session I		
Chairs:	**Arndt Rolfs, Ekaterina Zakharova**	
Ségolène Aymé, MD, PhD	Rare diseases in Europe: Achievements and remaining challenges	*9:10 - 9:30*
Jörn Oliver Sass	Inborn errors of ketone body utilization revisited	*9:30 - 9:50*
Ronald J.A. Wanders	Mitochondrial fatty acid oxidation deficiencies.	*9:50 - 10:10*
Maurizio Scarpa	The biological clock and the molecular basis of LSDS	*10:10 - 10:30*
David J. Begley	Transport to the CNS via lipoprotein receptors: a mechanism for targeting ERT to the brain?	*10:30 - 10:50*
Coffee break		*10:50 - 11:15*
Session II		
Chairs:	**Johannes Berger, Agnieszka Ługowska**	
Marie-T. Vanier	Niemann-Pick Disease type C: Update on laboratory diagnosis and therapeutic approaches (*25min*)	*11:15 - 11:40*
Cristin Davidson	Cyclodextrin as a therapy for Niemann-Pick type C disease	*11:40 - 12:00*

PROGRAM

13th International Conference On Rare Diseases
"Rare Diseases Crossing Borders Together"

Natalija Pichkur	Niemann-Pick disease spectrum of phenotypes	*12:00 - 12:20*
Arndt Rolfs	New biomarkers in the early diagnostics and monitoring of LSDs	*12:20 - 12:40*
Ekaterina Zakharova	Pitfalls of laboratory testing of inherited diseases	*12:40 - 13:00*
Lunch		***13:00 - 14:00***
Session III		
Chairs:	**Marie-T. Vanier, Jörn Oliver Sass**	
Ronald Wanders	Peroxisomal disorders	*14:00 - 14:20*
Johannes Berger	Pathophysiology of clinical heterogeneity in X-linked adrenoleukodystrophy	*14:20 - 14:40*
Eduard Paschke	GM1-Gangliosidosis and Morquio B: Pathobiochemistry and possible treatment strategies	*14:40 - 15:00*
Olga Amaral	Genetic diversity of Lysosomal Storage Disorders in Portugal	*15:00 - 15:20*
Jakub Sikora	Danon disease: LAMP2 protein content screening or high throughput sequencing	*15:20 - 15:40*
Coffee break		***15:40 - 16:00***
Session IV		
Chairs:	**Zita Krumina, Maciej Machaczka**	
Ursula Plöckinger	Emerging adults - problems in transition after transfer	*16:00 - 16:20*
Marija Jakutovič	The cardiovascular system in lysosomal storage disorders	*16:20 - 16:40*
Natalija Rumiantsava	Mucolipidoses: clinical data, genetic counseling, possibility of diagnostics and prevention in Belarus	*16:40 - 17:00*
Jaroslav Čermák	Paroxysmal nocturnal hemoglobinuria – a rare disorder of hematopoiesis	*17:00 - 17:20*
Maciej Machaczka	Hemophagocytic lymphohistiocytosis in children and adults	*17:20 - 17:40*
Agnieszka Ługowska	Metabolic Laboratory at the Department of Genetics, Institute of Psychiatry and Neurology – 48 years of activity in diagnostics of inherited metabolic diseases	*17:40 - 18:00*
Grill-dinner		***20:00***
Saturday , July 4, 2015		
Breakfast		***7:00 - 8:30***
Session V		
Chairs:	**Eduard Paschke, Olga Amaral**	
Gabor Linthorst	Fabry disease: challenges in diagnosis and treatment.	*8:35 - 8:55*
Cristina Drugan	Gaucher disease in Romanian patients: clinical spectrum and genetic analysis	*8:55 - 9:10*
Assel Tulebayeva	The frequency, structure and clinical features of mucopolysaccharidosis in Kazakhstan Republic	*9:10 - 9:25*

PROGRAM

13th International Conference On Rare Diseases
"Rare Diseases Crossing Borders Together"

Ivanka Sinigerska	Biochemical diagnosis of mucopolysaccharidoses in Bulgaria-35 years experience	*9:25 - 9:40*
Rimante Cerkauskiene	Centre of Pediatric Rare Diseases – Lithuanian experience	*9:40 - 9:55*
Gabor Linthorst	Inborn errors of metabolism from an adult perspective	*9:55 - 10:10*
Mohamad Mikati	The knock-in mouse model of AHC and Clinical implications	*10:10 - 10:30*
Igor Radziewicz-Winnicki/ Tomasz Pawlęga	Address of Polish Ministry of Health	*10:30 - 11:00*
Coffee break		***11:00 - 11:20***
Session VI		
Chairs:	**Małgorzata Krajewska-Walasek, Rafał Płoski**	
Anna Tylki Szymańska, ***R. Acuna-Hidalgo, M. Krajewska-Walasek***	Thyroid hormone resistance syndrome due to mutations in the thyroid hormone receptor alpha gene (*THRA*)	*11:20 - 11:40*
Peter Robinson	Human Phenotype Ontology and clinical diagnostics	*11:40 - 12:10*
Elżbieta Ciara, Magdalena Pelc	Application of the next generation sequencing to identification of the molecular background of genetically heterogeneous disorders such as RASopathies	*12:10 - 12:40*
Tomasz Gambin	Modern methods of novel Mendelian disease genes discovery using whole exome sequencing data	*12:40 - 13:00*
Lunch break		***13:00 - 14:00***
Session VII **Dysmorphology meeting**		
Moderators:	**Małgorzata Krajewska-Walasek, Robert Śmigiel**	*15:50 - 18:00*
Rocío Acuña-Hidalgo	Germline *SETBP1* mutations in Schinzel-Giedion syndrome	*14:00 - 14:15*
Kamila Czerska	Exome sequencing in medical diagnostics – so far experience	*14:15 - 14:30*
Aleksandra Jezela-Stanek	Novel mutations and clinical delineation of congenital disorder of glycosylation (CDG) caused by *PIGN* mutation	*14:30 - 14:45*
Krzysztof Szczałuba	*SETD5* gene mutation in two siblings discordant for ID/DD but consistent with the clinical diagnosis of oral-facial-digital spectrum	*14:45 - 15:00*
Robert Śmigiel	Next Generation Sequencing as a diagnostic method in searching the cause of severe epileptic encephalopathy – case report with *GRIN2B* mutation.	*15:00 - 15:15*
Małgorzata Krajewska-Walasek	Orphanet Europe - Polish team activity	*15:15 - 15:30*
Coffee break		***15:30 - 15:50***
Dysmorphology meeting (continued)		*15:50 - 18:00*
Moderators:	**Małgorzata Krajewska-Walasek, Robert Śmigiel**	*15:50 - 18:00*
Gala Dinner		***20:00***
Sunday , July 5, 2015		
Breakfast		***7:30 - 8:30***

PROGRAM

13th International Conference On Rare Diseases
"Rare Diseases Crossing Borders Together"

Session VIII		
Chairs:	**Maurizio Scarpa, Gabor Linthorst**	
Agnieszka Różdżyńska-Świątkowska	Anthropologist's contribution supporting diagnosis of rare diseases	*9:00 - 9:20*
Shunji Tomatsu	Therapies for the bone in mucopolysaccharidoses	*9:20 - 9:40*
Hanna Mierzewska	Alexander disease – in our own observation	*9:40 - 9:55*
Katarzyna Hetmańczyk	Chitotriosidase activity in patients with various types of dementia	*9:55 - 10:10*
Natalija Olkhovych	The chitotriosidase Activity in diagnostics of Lysosomal Storage Diseases	*10:10 - 10:25*
Gabor Linthorst	Inborn errors of metabolism from an adult perspective	*10:25 - 10:40*
Coffee break		***10:40 - 11:00***
Session IX		
Chairs:	**Zita Krumina , Aleksandra Gergont**	
Allison Brashear	ATP1A3: A spectrum or distinct phenotypes?	*11:00 - 11:20*
Aleksandra Gergont	Alternating hemiplegia of childhood and *ATP1A3* gene	*11:20 - 11:40*
Jacek Pilch	Alternating hemiplegia of childhood – a very rare or unrecognized disorder?	*11:40 - 12:00*
Agnieszka Stępień *K Guzek, A Kaufman*	Gross motor, fine motor and posture assessment in Alternating Hemiplegia od Childhood (AHC) individuals.	*12:00 - 12:20*
Svetlana Volgina	Lipoprotein Lipase Deficiency in an Infant. Case report	*12:20 - 12:30*
Krzysztof Kałwak	What's new in HSCT for metabolic disorders? Presentation of the first patient with MPS1H, who has been successfully transplanted in Department of Pediatric Hematology/Oncology and BMT in Wroclaw	*12:30 - 12:50*
Anna Tylki Szymańska, Teresa Matulka	Closing remarks and summary,	*12:50 - 13:00*
Lunch		***13:00 - 14:00***
	Medical consultation	*14:30 - 17:00*
	Consultation of unknown cases	*14:30 - 17:00*
Dinner		***18:00***
Monday, July 6, 2015		
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 health care system - European examples	*10:00 - 11:00*
	The benefits of the cooperation of patients organisations - exchange of experience	*11:00 - 12:00*
Teresa Matulka	Closing remarks and summary,	
Transfer to the airport