



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 Tyłki-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 diseases	17:15 - 17:30
Anna Wyszkon	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 Tyłki-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

2-6 July 2015, Białostrzegi "Promenade Center"



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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 Rumiantseva	Mucopolysaccharidoses: 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



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Ivanka Sinigerska	Biochemical diagnosis of mucopolysaccharidoses in Bulgaria-35 years experience	9:25 - 9:40
Rimante Cerkauskienė	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 Pawłęga	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 (<i>THRA</i>)	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 <i>SETBP1</i> 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 <i>PIGN</i> mutation	14:30 - 14:45
Krzysztof Szczaluba	<i>SETD5</i> 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 <i>GRIN2B</i> 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



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Session VIII		
Chairs:	Maurizio Scarpa, Gabor Linthorst	
Agnieszka Rózdzyńska-Swiątkowska	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 <i>ATP1A3</i> gene	11:20 - 11:40
Jacek Pilch	Alternating hemiplegia of childhood – a very rare or unrecognized disorder?	11:40 - 12:00
Agnieszka Stępień <i>K Guzek, A Kaufman</i>	Gross motor, fine motor and posture assessment in Alternating Hemiplegia of 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 Wrocław	12:30 - 12:50
Anna Tyłki 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	  	