16th International Conference on Rare Diseases «Understanding rare diseases»

June 28th – July 1st 2018

Conference Programme

Thursday, June 28 th 2018		
Lunch		***13:00 - 14:00***
Registration		*14:00 - 15:00*
Debate „ Understanding rare diseases" for patient organizations and members of Federation of Rare Diseases Patients in Central and Eastern Europe		
Teresa Matulka	**Welcome and opening**	*15:00 - 15:10*
Presentations of experts who will share their knowledge and experience		*15:10 - 16:00*
Debate - with experts from the health care system, government officials and representatives of patients' organizations		*16:00 - 17:00*
Discussion		*17:00 - 18:00*
Welcome dinner for invited conference guests		***19:00 - 22:00***
Friday, June 29th 2018		
Breakfast		***7:00 - 8:00***
Registration		*8:00 - 9:20*
Teresa Matulka **Anna Tylki-Szymańska**	**Welcome address**	*9:20 - 9:30*
Session I		
Chairs:	**Małgorzata Krajewska-Walasek, Jakub Sikora**	***9:30 -11:00***
Timothy M. Cox	What it means to be Rare	*9:30 - 9:50*
Ségolène Aymé	Treating genetic diseases: lessons from experience and prospects	*9:50 - 10:10*
Maurizio Scarpa	The European Reference Network for Rare Hereditary Metabolic Diseases, MetabERN	*10:10 - 10:40*
Marc Dooms	Information needs of physicians regarding the diagnosis of rare diseases	*10:40 - 11:00*
Coffee break		***11:00 - 11:20***
Session II		
Chairs:	**Marie T. Vanier, Timothy M. Cox**	***11:20 - 13:00***
Mariusz Więckowski	What can we foretell from the mitochondrial parameters	*11:20 - 11:45*
Małgorzata Wiweger	Zebrafish models for (ultra)rare diseases	*11:45 - 12:05*
David J. Begley	New Insights into Transcytosis at the BBB: Implications for ERT Delivery to the CNS	*12:05 - 12:25*
Lars Schlotawa	Current aspects of Multiple Sulfatase Deficiency: understanding a very rare disease and efforts towards a treatment.	*12:25 - 12:45*
Joanna K. Purzycka-Olewiecka	Women are Rare	*12:45 - 13:00*
Lunch		***13:00 - 14:00***

Session III		
Chairs:	**Ekaterina Zakharova, Marc Dooms**	*14:00 - 15:30*
Timothy M. Cox	Sphingolipids in health and disease	*14:00 - 14:20*
Marie T. Vanier	Niemann-Pick disease type C: an update	*14:20 - 14:50*
Jakub Sikora	Acid sphingomyelinase deficiency causes variable cerebellar pathology that can be ameliorated by N-butyl-deoxynojirimycin in the mouse model	*14:50 - 15:10*
Mia Horovitz	Gaucher disease and its association with Parkinson's disease	*15:10 - 15:35*
Agnieszka Ługowska	Gene expression profile in patients with Gaucher disease: preliminary results of a microarray study	*15:35 - 15:50*
Coffee break		***15:50 - 16:10***
Session IV		
Chairs:	**Ségolène Aymé, Maurizio Scarpa**	*16:10 – 17:40*
Shunji Tomatsu	Gene therapy for Mucopolysaccharidoses	*16:10 - 16:35*
Assel Tulebayeva	Aortic root dilatation in patients with Hunter syndrome	*16:35 - 16:50*
Natalia Trofimova	Genetic heterogeneity of patients with mucopolysaccharidosis I and IIIA types in Ukraine	*16:50 - 17:05*
Galina Baydakova	The ethnic and regional features of lysosomal storage diseases in Russia	*17:05 - 17:25*
Oksana Barvinska	Mutation analysis of *IVD*, *GCDH* and *MUT* genes in patients with organic acidurias from Ukraine	*17:25 - 17:45*
Ana Skaricic	Expanded newborn screening in Croatia – a 9 months pilot project experience	*17:45 - 18:00*
Grill-dinner		***20:00***
Saturday, June 29th 2018		
Breakfast		***7:00 - 8:45***
Session V		
Chairs:	**Sylvia Stockler, Mariusz Więckowski**	*9:00 - 10:45*
Saskia B. Wortmann	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?	*9:00 - 9:20*
Holger Prokisch	Probing the non-coding variation in Mendelian diseases	*9:20 - 9:40*
Rafał Płoski	Whole exome and whole genome sequencing for delineation of novel monogenic diseases in human	*9:40 - 10:00*
Ekaterina Zakharova	From biochemical tests for Inherited metabolic disorders to DNA analysis and *vice versa*	*10:00 - 10:20*
Jörn Oliver Sass	Human D-glycerate kinase and D-glyceric aciduria	*10:20 - 10:45*
Coffee break		***10:45 - 11:00***
Session VI		
Chairs:	**Zita Krumina, Jörn Oliver Sass**	*11:00 – 13:05*
Sylvia Stockler	Morquio B Disease: A journey towards a better understanding of an ultra rare disease.	*11:00 - 11:35*
Nataliia Mytcyk	Features of spectrum and regional prevalence of mutations in *GLB1* gene in Ukrainian patients with GM1-gangliosidosis	*11:35 - 11:50*
Alena Lapatsentava	Diagnosis and treatment of Gaucher's disease in the Republic of Belarus	*11:50 - 12:05*
Nataliia Olkhovitz	Molecular genetics characteristic of Gaucher disease in Ukraine	*12:05 - 12:25*
Svetlana Volgina	Tuberous Sclerosis Complex Diagnostic Criteria Update	*12:25 - 1245*
Natallia Rumiantsava	Fryns syndrome in Belarus: clinical and genetic characterization, differential diagnostics	*12:45 - 13:05*
Lunch		***13:05 - 14:00***

Session VII **Dysmorphology meeting** **Małgorzata Krajewska-Walasek**		
Chairs:	**Dariusz Rokicki, Jennifer Castañeda**	***14:00 - 16:00***
Małgorzata Krajewska-Walasek	Introduction to issue: Agenesis of the corpus callosum - a clinical approach to diagnosis	*14:00 - 14:20*
Elżbieta Jurkiewicz	Corpus callosum - without secrets?	*14:20 - 15:00*
Krzysztof Szczałuba	Advances in the genetic diagnosis and neuroimaging of mid-/hindbrain malformations with special emphasis on Joubert syndrome	*15:00 - 15:25*
Robert Śmigiel	Mowat-Wilson syndrome – clinical and molecular analysis of Polish patients	*15:25 - 15:45*
Jacek Pilch	Hypoplasia of the corpus callosum in a 16,5-year-old boy with 13q22-q32 deletion: phenotype-genotype analysis	*15:45 - 16:00*
Coffee break		***16:00 - 16:15***
Dysmorphology meeting		
Chairs:	**Robert Śmigiel, Krzysztof Szczałuba, Małgorzata Krajewska-Walasek**	***16:15 -18:00***
Olga Khurs	Angelman syndrome in infants: presentation of three cases	*16:15 - 16:30*
Irina Matusik	Cri-du-Chat syndrome: de novo and inherited monosomy 5p in infants	*16:30 - 16:45*
	Unknown cases	*16:45 - 18:00*
Gala Dinner		***20:00***
Sunday, July 1st 2018		
Breakfast		***7:30 - 8:45***
Session VIII		
Chairs:	**Saskia B. Wortmann, David J. Begley**	***9:00 – 11:00***
Marc Dooms	Compounded medication for patients with rare diseases	*9:00 - 9:20*
Zita Krumina	The stem cell transplantation in patient with Alpha mannosidosis. Case report.	*9:20 - 9:40*
Elżbieta Szczepanik	Efficacy and safety of intrathecal infusion of autologic adipose derived regenerative cells (ADRC) in an autoimmune determined refractory epilepsy in children	*9:40 - 10:00*
Natalia Pechatnikova	Success experience of the treatment severe respiratory insufficiency in child with Pompe disease with high dose of Alglucosidase alfa	*10:00 - 10:20*
Anna Tylki-Szymańska	Enzyme replacement therapy in lysosomal storage diseases	*10:20 - 10:40*
Teresa Matulka **Anna Tylki-Szymańska**	Closing remarks and summary	*10:40 - 11:00*
Coffee break		***11:00 - 11:30***
Medical consultations		
Medidal consultations		*11:30 - 13:00*
Lunch		***13:00 - 14:00***
Medical consultations		*14:00 - 17:00*
Dinner		***18:00 - 19:00***
Monday , July 1st 2018		
Breakfast		***7:00 - 8:30***

Transfer

Patronage

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Marek Michalak

Rzecznik Praw Pacjenta

RZECZNIK PRAW OBYWATELSKICH

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Dziecka im. Jana Pawła II

The organizers

Choroby Rzadkie

Cooperation

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