15th International Rare Diseases Conference

Rare Diseases - Open Your Heart and Mind

29 June – 2 July 2017

Conference Programme

Thursday, 29 June 2017		
Lunch		***13:00 - 14:00***
Registration		*14:00 - 15:00*
Debate „Rare diseases - caring for patients" 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*
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*
Holy Mass in intention of patients with rare diseases and their families		*18:00 - 19:00*
Hotel Rewita	Welcome dinner for invited conference guests	***19:00 - 22:00***
Friday, 30 June 2017		
Breakfast		***7:00 - 8:30***
Registration		*8:00 - 9:10*
Teresa Matulka **Anna Tylki-Szymańska**	**Welcome address**	*9:10 - 9:20*
Session I		
Chairs:	**Ségolène Aymé, Jörn Oliver Sass**	*9:20 - 11:00*
Timothy M Cox *Cambridge, UK*	**Developing Gene Therapy for GM2 gangliosidosis**	*9:20 - 9:40*
Volkmar Gieselmann *Bonn, Germany*	**Advancing treatment options for metachromatic leukodystrophy.**	*9:40 - 10:00*
Grzegorz Węgrzyn *Gdansk, Poland*	**The past, present and future of genistein in treatment of lysosomal storage diseases**	*10:00 - 10:20*
Marc Dooms *Louven, Belgium*	**From Promising Molecules to Orphan Drugs: Early Clinical Drug Development**	*10:20 - 10:40*
Robert Dobrovolny *Prague, Czech Republic*	**Induced Pluripotent Stem Cells As A Model To Study Pathogenesis Of Lysosomal Disorders**	*10:40 - 11:00*
Coffee break		***11:00 - 11:30***
Session II		
Chairs:	**Ekaterina Y Zakharova, Grzegorz Węgrzyn**	*11:30 - 13:00*
Jörn Oliver Sass *Bonn-Rhein-Sieg,Germany*	**Clinical presentation and Outcome in a Series of 32 Patients with 2-Methylacetoacetyl-Coenzyme A Thiolase (β-Ketothiolase, MAT)**	*11:30 - 11:50*
Galina V Boydakova *Moscow, Russia*	**What does biochemical test results really mean for physician?**	*11:50 - 12:10*
Jörn Oliver Sass *Bonn-Rhein-Sieg, Germany*	**Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency: Clinical Presentation and Outcome in a Series of 37 Patients**	*12:10 - 12:30*
Filip Majer *Prague, Czech Republic*	**Multi-exon deletions in the LAMP2 gene – a potential confounder to both targeted and NGS-based identification of Danon disease heterozygous female patients**	*12:30 - 12:50*
Lunch		***13:00 - 14:00***
Session III		

Chairs:	**Zita Krumina, Timothy Cox**	*14:00 - 15:55*
Shunji Tomatsu *Wilmington, USA*	**Mucopolysaccharidoses Update: impact to the bone**	*14:00 - 14:20*
Arndt Rolfs *Rostock, Germany*	**Global diagnosis of 270 Morquio A patients based on a dry blood spot assay: a two years study**	*14:20 - 14:40*
Ekaterina Y Zakharova *Moscow, Russia*	**One drop of blood- revolution for diagnosis of IMD**	*14:40 - 14:55*
Saskia B. Wortmann *Salzburg, Austria*	**Pyrimidine biosynthesis pathway: a new treatable inborn error of metabolism and a non disease causing benign orotic aciduria**	*14:55 - 15:15*
Jakub Sikora *Prague,Czech Republic*	**Lysosomal storage in valvular interstitial cells is a dominant feature of aortic valve pathology in acid sphingomyelinase deficiency**	*15:15 - 15:30*
Olga Amaral *Porto, Portugal*	**Rare diseases - Open your heart and keep an open mind**	*15:30 - 15:45*
Karolina Pierzynowska *Gdansk, Poland*	**Can many different neurodegenerative diseases be treated with a single drug?**	*15:45 - 15:55*
Coffee break		***15:55 - 16:20***
Session IV		
Chairs:	**Saskia Wortmann, Volkmar Gieselmann**	*16:20 - 18:00*
Ségolène Aymé *Paris, France*	**New developments in research and health care organization for rare diseases in Europe**	*16:20 -16:40*
Sara Mole *London, UK*	**Batten disease – working towards therapy**	*16:40 -16:55*
Sander F. Garrelfs *Amasterdam The Netherlands*	**Clinical characterisation of Primary Hyperoxaluria by using the OxalEurope collaborative database"**	*16:55 - 17:10*
Anna Wilbik *Eindhoven, The Netherlands*	**Data analytics in healthcare applications**	*17:10 - 17:25*
Marc Dooms *Louven, Belgium*	**Good off-label use practices for rare diseases**	*17:25 - 17:40*
Joanna Brokowska *Gdansk, Poland*	**Induction of the autophagy process by isothiocyanates in the light of potential therapy of Huntington's disease**	*17:40 - 17:50*
Lidia Gaffke *Gdańsk, Poland*	**Changes in the cytoskeleton in Sanfilippo and Huntinton diseases, and their correction by genistein**	*17:50 - 18:00*
Grill-dinner		***20:00***
Saturday, 1 July 2017		
Breakfast		***7:00 - 8:45***
Session V		
Chairs:	**Sara Mole, Jakub Sikora**	*9:00 - 10:45*
Hanna Rosenbaum *Haifa, Israel*	**IGG4 related sclerosing disease –a rare disorder with common multisystemic symptoms.**	*9:00 - 9:20*
Maciej J Machaczka *Stockholm ,Sweden*	**Therapy response and long-term outcome of adults with hematological malignancy-associated Hemophagocytic Lymphohistiocytosis**	*9:20 - 9:40*
Alena Lapatsentava *Minsk, Belorussia*	**Progressive mesenteric lymphadenopathy in child with Gaucher disease despite enzyme therapy. Case report.**	*9:40 - 9:55*
Assel Tulebayeva *Almaty, Kazakhstan*	**Cardiovascular morphology in children with mucopolysaccharidosis in the Republic of Kazakhstan.**	*9:55 - 10:10*
Dariusz Rokicki *Warsaw, Poland*	**Liver transplantation in urea cycle disorders – dilemmas and problems on the example of Polish patients**	*10:10 - 10:25*
Tomasz Kmieć *Warsaw, Poland*	**Multi target treatment of PKAN-related dystonia with subthalamic or pallidal nucleus deep brain stimulation**	*10:25 - 10:45*
Coffee break		***10:45 - 11:15***
Session VI		

Chairs:	**Olga Amaral, Maciej Machaczka**	*11:15 - 12:50*
Ksenija Fumić *Zagreb, Croatia*	**Laboratory diagnosis of inborn errors of metabolism in Croatia**	*11:15 - 11:30*
Marina Szlago *Buenos Aires, Argentina*	**Inherited errors of metabolism (IEM). From clinics to diagnosis: an Argentinian experience**	*11:30 - 11:45*
Irina Naumchik *Minsk, Belarus*	**Management of healthcare for patients with rare diseases in Belarus**	*11:45 - 12:00*
Natalija Pichkur *Kiev, Ukraine*	**Diagnostics and management of MPS patients in Ukraine.**	*12:00 - 12:20*
Olga Riga *Kharkiv, Ukraine*	**Rare Diseases in Young Children and Pediatrics Palliative Care in Kharkiv region of Ukraine**	*12:20 - 12:35*
Sanija Perić *Zagreb, Croatia*	**Rare Disease Croatia – connection between patients and health care and social services**	*12:35 - 12:50*
Lunch		***13:00 - 14:00***
Session VII **Dysmorphology meeting - Genomic imprinting as a cause of diseases/ Prof Małgorzata Krajewska-Walasek**		
Moderators:	**Agnès Linglart, Krystyna Chrzanowska**	*14:00 - 16:00*
Dorota Jurkiewicz *Warsaw, Poland*	**Molecular mechanisms leading to imprinting disorders**	*14:00 - 14:15*
Agnès Linglart *Paris, France*	**Pseudohypoparathyroidism, recent advances in diagnosis and care**	*14:15 - 14:45*
Krystyna Chrzanowska *Warsaw, Poland*	**Strategy for investigation, diagnosis and management of Silver-Russell syndrome: presentation of the first international consensus recommendations**	*14:45 - 15:05*
Ewa Obersztyn *Warsaw, Poland*	**Prader- Willi syndrome - the present times and hopes for the future. Diagnostics, clinics and therapeutic prospects.**	*15:05 - 15:25*
Agnieszka Pollak *Warsaw, Poland*	**Challenges in molecular diagnostics of *GNAS* locus**	*15:25 - 15:40*
Agata Skórka *Warsaw, Poland*	**Three sporadic cases of Beckwith-Wiedemann syndrome due to three novel mutations in *CDKN1C* gene**	*15:40 - 15:50*
Tatiana Demidovich *Minsk, Belarus*	**Beckwith-Wiedemann syndrome in Belarusian patients.**	*15:50 - 16:00*
Coffee break		***16:00 - 16:15***
Dysmorphology meeting		
Moderators:	**Ewa Obersztyn, Robert Śmigiel, Małgorzata Krajewska-Walasek**	*16:15 - 18:00*
Małgorzata Krajewska-Walasek *Warsaw, Poland*	**Clinical utility of whole-exome sequencing in diagnosing patients with non-specific intellectual disability with or without dysmorphic features (so called „unknown cases")**	*16:15 - 16:35*
Rafał Płoski *Warsaw, Poland*	**Search for novel disease genes by precise breakpoint detection in *de novo* balanced chromosomal translocations carriers**	*16:35 - 16:50*
Karolina Matuszewska *Poznan, Poland*	**Phenotype of two Polish patients with mutation in *MAGEL2* gene, identified by *next-gen*eration sequencing**	*16:50 - 17:05*
Dorota Karczmarewicz *Warsaw, Poland*	**Orphanet and Orphanet Poland**	*17:05 - 17:20*
	Presentations of unknown and known cases (for each case 5 minutes)	*17:20 - 18:00*
Gala Dinner		***20:00***
Sunday, 2 July 2017		
Breakfast		***7:30 – 8:45***
Session VIII		
Chairs:	**Assel Tulebayeva, Marc Dooms**	*9:00 - 10:50*
Hanna Mierzewska *Warsaw, Poland*	**Atypical fetal onset Alpers syndrome due to *PARS2* Gene mutations**	*9:00 – 9:20*
Agnieszka Ługowska *Warsaw, Poland*	**Multiple Sulfatase Deficiency (MSD) in a young patient with leukodystrophy and transient signs of ichtyosis.**	*9:20 - 9:40*
Svetlana Volgina *Kazan, Russia*	**Congenital multiple tumors of the glomus in a child (a case report).**	*9:40 - 10:00*
Zita Krumina *Riga, Latvia*	**Diagnostic difficulties in clinical practise. Case report.**	*10:00 - 10:15*
Olga Zobikova *Minsk, Belarus*	**Carvajal Syndrome. Case report.**	*10:15 - 10:30*
Volha Smirnova *Mogilev, Belarus*	**Medium-chain acyl-CoA dehydrogenase deficiency in the Mogilev region of the Republic of Belarus**	*10:30 - 10:50*
Coffee break		***10:50 - 11:20***

Session IX		
Chairs:	**Svetlana Volgina , Grzegorz Węgrzyn**	*11:20 - 12:00*
Agnieszka Rusińska *Łodz, Poland*	**Recent advances in the molecular diagnostics and treatment of osteogenesis imperfecta in children – experience *of the Department***	*11:20 - 11:40*
Anna Tylki-Szymańska *Warsaw, Poland*	**Alpha-mannosidosis - lysosomal disease with immune deficiency**	*11: 40 - 12:00*
Izabela Michalus *Łodz, Poland*	**Rare, genetically conditioned forms of rickets - differential diagnosis and treatment.**	*12:00 - 12:20*
Anna Tylki Szymańska Teresa Matulka	**Closing remarks and summary.**	*12: 20 - 12:30*
Lunch		***13:00 - 14:00***
Medical consultations		*14:30 - 17:00*
Consultations of unknown cases		*14:30 - 17:00*
Dinner		***18:00 - 19:00***
Integration meeting *"Konferencje"* building		*19:00 - 23:00*
Monday, 3 July 2017		
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	  	

Patronate

15th International Conference on Rare Diseases "Rare Diseases - Open Your Heart and Mind."

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