PROGRAM

12th European Conference On Rare Diseases
"Living With A Rare Disease"
Spała 2014

Friday, July 04, 2014		
Breakfast		***7:30 - 8:30***
	Registration	*8:00 - 9:00*
Teresa Matulka **Anna Tylki-Szymańska** *Warsaw, Poland*	**Welcome Address**	*9:00 - 9:30*
Session I		
Chairs:	**Grzegorz Węgrzyn, Ekaterina Zakharova**	
Jörn Oliver Sass *Zürich, Switzerland*	Inborn Errors of Cobalamin Transport and Metabolism	*9:30-9:50*
Ladislav Kuchar *Prague, Czech Republic*	Tandem mass spectrometry: new possibilities for research and diagnosis of lysosomal storage disorders	*9:50-10:10*
Gajja S Salomons *Amsterdam, Netherlands*	Update on the D-2- and L-2-Hydroxyglutaric Acidurias	*10:10-10:30*
Johannes Berger *Vienna, Austria*	X-linked Adrenoleukodystrophy: From the basic research to possible therapeutic strategies.	*10:30-10:50*
Coffee break		***10:50-11:20***
Session II		
Chairs:	**Maciej Machaczka, Jana Ledvinova**	
Stefan Karlsson *Stockholm, Sweden*	Development of gene therapy in a mouse model for Gaucher disease	*11:20-11:50*
Hanna Rosenbaum *Haifa, Israel*	Gaucher disease: clinical course, diagnosis and innovation in treatment.	*11:50-12:10*
Jana Ledvinova *Prague, Czech Republic*	Diagnosis of lysosomal storage disorders in the Czech Republic	*12:10-12:30*
Nataliia Pichkur *Kyiv, Ukraine*	Hyperphenylalaninemias: biopterins deficiency	*12:30-12:45*
Lunch		***12:45-14:00***
Session III		
Chairs:	**Joern Olivier Sass, Johannes Berger**	
David J. Begley *London, UK*	Delivering enzyme replacement therapy for mucopolysaccharidosis and other lysosomal storage disorders to the CNS across the blood-brain barrier.	*14:00-14:20*
Ulrich Matzner *Bonn, Germany*	Advances in therapies for metachromatic leukodystrophy	*14:20-14:40*
Wioletta Kobiela *Gdansk, Poland*	Evaluation of the efficacy of different therapeutic approaches in the treatment of MPS I	*14:40-15:00*
Agnieszka Ługowska *Warsaw, Poland*	Does apolipoprotein E genotype and LRP1 polymorphisms influence the phenotype in patients with different clinical types of metachromatic leukodystrophy ?	*15:00-15:15*
Katarzyna Hetmańczyk *Warsaw, Poland*	Dipeptidyl peptidase-IV: a new biomarker for diagnosing of mucopolysaccharidoses – preliminary results	*15:15-15:25*

Karl-Eugen Mengel *Mainz, Germany*	Diagnosis, manifestation and management of GM1-Gangliosidosis in Germany	*15:25-15:45*
Shunji Tomatsu *Wilmington, USA,*	Newborn screening and diagnosis of mucopolysaccharidoses	*15:45-16:05*
Coffee break		***16:05 -16:30***
Session IV		
Chairs	**Michael Beck, Rafał Płoski**	
Grzegorz Węgrzyn *Gdansk, Poland*	Gene Expression-Targeted Isoflavone Therapy: Facts, Controversies and Further Possibilities	*16:30-16:50*
Maurizio Scarpa *Wiesbaden, Germany*	Pathophysiology of neurodegeneration in Lysosomal Storage Diseases and approaches to therapy of the CNS	*16:50-17:10*
Maciej Machaczka *Stockholm Sweden*	Thirty years' experience of allogeneic hematopoietic stem cell transplantation for inborn errors of metabolism in Sweden	*17:10-17:30*
Krzysztof Kałwak *Wroclaw, Poland*	Hematopoietic stem cell transplantation in metabolic disorders. What's new in the world and in Poland?	*17:30-17:50*
Grill-dinner		***20:00***
Saturday , July 5, 2014		
Breakfast		***7:30 - 8:30***
Session V		
Chairs	**Gajja Salomons, Agnieszka Ługowska**	
Rafał Płoski *Warsaw, Poland*	Whole exome sequencing for diagnosis of rare diseases - 1,5 year experience	*8:30-8:50*
Paulina Jędrak *Gdansk, Poland*	The significance of the level of mitochondrial DNA in cells of patients suffering from Huntington's disease	*8:50-9:10*
Barbara Zapała *Krakow, Poland*	A novel *TAZ* gene mutation and maternal mosaicism in Polish family with Barth syndrome	*9:10-9:30*
Hanna Mierzewska *Warsaw, Poland*	MRI in diagnostics of rare diseases with the nervous system involvement.	*9:30-9:50*
Coffee break		***9:50-10:20***
Session VI		
Chairs	**Maurizio Scarpa, Anna Tylki-Szymańska**	
Michael Beck *Mainz, Germany*	The Center for Lysosomal Storage Disorders at the University of Mainz (Villa Metabolica): A model for a Center of Rare Diseases	*10:20-10:50*
Christina Lampe *Wiesbaden, Germany*	Transition of Adolescents in Adulthood	*10:50-11:10*
Representatives of the state administration	Rare diseases in Poland	*11:10-11:50*
Roundtable meeting	Representatives of the state administration - representatives of patient organizations - *Knights' Hall*	*11:50 -13:00*
Arndt Rolfs *Rostock, Germany*	New insights in the diagnosis and treatment of Gaucher disease	*11:50-12:10*
Ivanka Sinigerska *Sofia, Bulgaria*	Biochemical Diagnosis of Lysosomal Storage Diseases in Bulgaria	*12:10-12:30*

Radka Tincheva *Sofia, Bulgaria*	Healthcare of children with treatable rare diseases in Bulgaria"	*12:30-12:50*
Chris Hendriksz *Manchester, UK*	Enzyme Replacement therapy for Morquio (MPS IVA)	*12:50-13:10*
Lunch break		***13:00 - 14:00***
Session VII **Dysmorphology meeting**		
Chairs	**Małgorzata Krajewska-Walasek, Ewa Obersztyn**	
Aleksandra Jezela-Stanek *Warsaw, Poland*	What dysmorphologists should keep in mind during evaluation of patients with blepharophimosis and mental disability?	*14:10-14:30*
Małgorzata Krajewska-Walasek *Warsaw, Poland*	Genitopatellar syndrome contra Ohdo-like /SBBYS syndrome: two disorders – one gene. Understanding clinical similarities and differences according to known *KAT6B* defects	*14:30-14:50*
Robert Śmigiel *Wroclaw, Poland*	Ohdo syndrome caused by mutation in *KAT6B* gene - case report and phenotype analysis on the basis of the literature	*14:50-15:05*
Jacek Pilch *Katowice, Poland*	Microdeletion 2q31.1-32.2 in a boy with blepharophimosis and mental retardation – a phenotype analysis	*15:05-15:20*
Coffee break		***15:20 - 15:40***
Moderators: Małgorzata Krajewska-Walasek, Robert Śmigiel	Presentations of unknown cases	*15:40 -17:50*
Gala Dinner		***20:00***
Sunday , July 6, 2014		
Breakfast		***7:30 – 8:30***
Session VIII		
Chairs	**Zita Krumina, Anna Latos-Bieleńska**	
Zita Krumina *Riga, Latvia*	Difficulties in diagnosis of rare genetic disorders in Latvia	*9:10-9:30*
Julia Zakharchuk *Chelyabinsk, Russia*	The clinical case of MPS VI type Maroteaux – Lamy	*9:30-9:50*
Aleksandra Żurowska *Gdansk, Poland*	Do we have a Wonder Drug for atypical hemolytic-uremic syndrome ?	*9:50-10:10*
Olga Cirstea *Chisinau, Moldova*	Wegener's Granulomatosis in children: Case report and the need for a multidisciplinary approach	*10:10-10:30*
Svetlana Volgina *Kazan, Russia*	Fabry disease in the children: from the onset of clinical symptoms until diagnosis (speech in Russian)	*10:30-10:50*
Coffee break		***10:50 - 11:20***
Session IX		
Arunas Valiulis *Vilnius, Lithuania*	Alpha-1 Antitrypsin Deficiency Network as Pattern of Management of Rare Disease in Central - Eastern Europe"	*11:20-11:40*
Anna Tylki-Szymańska *Warsaw, Poland*	Inborn errors of metabolism – manifestation in newborns	*11:40-12:00*
Ekaterina Zakharova *Moscow, Russia*	Undiagnosed patient	*12:00-12:20*
Teresa Matulka **Anna Tylki-Szymańska** *Warsaw, Poland*	Summary of the conference	*12:20-13:00*

Lunch	***13:00 - 14:00***
Medical consultations	*9:00 - 16:30*
Holy Mass in intention of patients with rare diseases and their families	*17:00 -18:00*
Dinner	***19:00***
Monday, July 7, 2014	
Breakfast	***8:00 -9:00***
Transfer to the airport   	

Session for Patients, Parents and Care givers

Saturday , July 5, 2014		
Breakfast		***8:30 - 9:30***
Session for Patients, Parents and Care givers		
Chairs	**Anna Pruszyńska, Paulina Zalewska**	
Barbara Czartoryska *Warsaw, Poland*	The role of the laboratory in the diagnosis of lysosomal storage diseases	*9:30-10:00*
Jolanta Wierzba Gdańsk, *Poland*	Why children are unique genetic.	*10:00-10:30*
Coffee break		***11:00 - 12:00***
Jolanta Marucha *Warsaw, Poland*	The role of physiotherapy in progressing dysfunction of musculoskeletal system in patients with selected metabolic diseases	*12:00-12:30*
Beata Prorok *Łódź, Poland*	Palliative care of patients with rare diseases	*12:30-13:00*
Lunch		***14:00 - 15:00***
Maciej Machaczka *Stockholm Sweden*	Karolinska experience in allogeneic hematopoietic stem cell transplantation for inherited metabolic disorders	*15:00 - 15:30*
Tylki Szymańska Anna *Warsaw, Poland*	Living with a rare disease	*15:30 - 16:00*
Summary and the end of the conference		*16:00 - 16:20*
Medical consultations, optical examinations		*16:20 - 18:00*
Gala Dinner		*20:00*
Monday, July 7, 2014		
Breakfast		***8:00 -9:00***
Medical consultations - *paediatric, metabolic, genetic, neurological, psychological, orthopaedic and genetic oncology*		***9:00-16:00***