XI European Conference on Rare Diseases

"Rare Diseases - upcoming challenges"

Friday, June 28, 2013		
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:	**Volkmar Gieselmann, Agnieszka Ługowska**	
Segolene Aymé Paris, France	Improvement of the quality of care for patients with rare diseases: where do we stand in Europe ?	9:30 - 9:50
Thomas Kolter Bonn, Germany	Biochemical Principles of sphingolipidoses	9:50 - 10:10
Aldona Dembińska-Kieć Cracow, Poland	Progress in markers of inherited diseases. Lipidomics and epigenetics	10:10 - 10:30
Jörn Oliver Sass Zürich, Switzerland	Inborn errors of ketone body metabolism	10:30 - 10:50
	Discussion	10:50 - 11:00
Coffee break		*11:00 - 11:20*
Session II		
Chairs:	**Marie T. Vanier, Grzegorz Węgrzyn**	
Volkmar Gieselmann Bonn, Germany	Metachromatic leukodystrophy: lessons from animal models	11:20 - 11:40
Johannes Berger Vienna, Austria	Molecular mechanisms in X-linked adrenoleukodystrophy	11:40 - 12:00
David J. Begley London, UK	New prospects for delivering enzyme replacement therapy (ERT) across the blood-brain barrier in neurodegenerative lysosomal storage disorders.	12:00 - 12:20
Ben J.H.M. Poorthuis Amsterdam, the Netherlands	Globotriaosylsphingosine (lysoGb3) as a biomarker for Fabry disease	12:20 - 12:40
Ekaterina Zakharova Moscow, Russia	Diagnostic clues for inherited metabolic diseases	12:40 - 13:00
	Discussion	13:00 - 13:05
Lunch		*13:05 - 14:00*
Session III		
Chairs:	**Jörn Oliver Sass, Catherine Caillaud**	
Marie T. Vanier Lyon, France	Niemann-Pick disease type C: update on epidemiology, diagnostic strategies and management	14:00 - 14:20
Catherine Caillaud Paris, France	GM2 gangliosidoses : from diagnosis to future therapies	14:20 - 14:40
Maurizio Scarpa Padova, Italy	Pathophysiology of neurodegeneration in lysosomal storage diseases	14:40 - 15:00
Debora Elstein Jerusalem, Israel	Gaucher disease	15:00 - 15:20
Christina Lampe Mainz, Germany	Morquio A syndrome - clinical trials and results	15:20 - 15:40
Agnieszka Ługowska Warsaw, Poland	Mutations in the gene for beta-glucocerebrosidase are more frequent in patients with early onset Parkinson's disease than in control individuals	15:40 – 15:55
	Discussion	15:55 -16:00
Coffee break		*16:00 -16:20*
Session IV		

Chairs	**Olga Amaral, Zita Krumina**	
Jiri Zeman Prague, Czech Republic	Mitochondrial medicine: from symptoms to diagnosis	16:20 - 16:40
Linda De Meirleir Brussels, Belgiun	New phenotypes in mitochondrial diseases	16:40 - 17:00
Leyla S.Namazova-Baranova Moscow, Russia	Managing children with rare deseases - Russian experience"	17:00 - 17:20
Olga Amaral Porto, Portugal	An account of the Portuguese experience with lysosomal storage disorders	17:20 - 17:35
Zita Krumina Riga, Latvia	Rare diseases in Latvia	17:35 - 17: 50
Wiesława Radziewicz Vilnius, Lithuania	Haemophilia patients – rehabilitation challenges	17:50 - 18:00
Grill-dinner		*19:00*
Saturday, 29 June, 2013		
Breakfast		*7:30 - 8:30*
	Registration	8:00 - 9:00
Grzegorz Węgrzyn Gdańsk, Poland	Second Day Welcome Address	9:00 - 9:10
Session V		
Chairs	**David Begley, Johannes Berger**	
Shunji Tomatsu Wilmington, USA	Diagnosis and Treatments for Mucopolysaccharidoses	9:10 - 9:30
Grzegorz Węgrzyn Gdańsk, Poland	Molecular Mechanisms of Gene Expression-Targeted Isoflavone Therapy for Lysosomal Storage Diseases	9:30 - 9:50
Magdalena Narajczyk, Gdańsk, Poland	Electron microscopy methods used in biomarker analysis of mucopolysaccharidoses – a group of lysosomal storage disorders	9:50 - 10:10
Maciej Machaczka Stockholm, Sweden	Special features of Gaucher disease in Sweden	10:10 - 10:25
Cristina Drugan Cluj-Napoca, Romania	Fabry disease in Romanian patients: clinical spectrum and genetic analysis	10:25 -10:40
	Discussion	10:40 -10:45
Coffee break		*10:45 - 11:00*
Session VI		
Chairs	**Jolanta Wierzba, Teresa Matulka**	11:00 - 13:00
Liesbeth Siderius Amsterdam, The Netherlands	Rare diseases; common medial and social indicators	11:00 - 11:20
Igor Radziewicz-Winnicki Warsaw, Poland	Deputy Minister of Health	11:20 - 11:40
Krzysztof Łanda Warsaw, Poland	Foundation Watch Health Care	11:40 - 12:00
	Discussion	12:00 - 13:00
Lunch break		*13:00 - 14:00*
Session VII Dysmorphology meeting		
Chairs	**Małgorzata Krajewska-Walasek, Ewa Obersztyn**	
Aleksandra Jezela-Stanek Warsaw, Poland	Ciliopathies - whether we are able to keep up with the progress in molecular etiology?	14:00 - 14:30
Anna Latos-Bieleńska Poznań, Poland	Sensenbrenner syndrome (Cranioectodermal dysplasia, CED) – a genetically heterogenous ultra rare disease with unusual phenotype	14:30 - 14:45
Anna Jakubiuk-Tomaszuk Białystok, Poland	Metabolic disorders in Alström syndrome in 3 year old girl – case report.	14:45 - 15:00
Monika Kugaudo Warsaw, Poland	Acrocallosal syndrome (ACLS) as an example of ciliopathy	15:00 - 15:15

Małgorzata Krajewska-Walasek Warsaw, Poland	Nicolaides-Baraitser syndrome or Coffin-Siris syndrome?: that is the question!	15:15 - 15:45
Ewa Obersztyn Warsaw, Poland	Nicolaides-Baraitser syndrome: a photo-essay on a natural history of a 25-year old female patient	15:45 - 16:00
Coffee break		*16:00 - 16:20*
Robert Śmigiel Wrocław, Poland	Coffin-Siris syndrome caused by *SMARCA4* gene mutation – case presentation and literature review	16:20 - 16:35
Cases presentation	„Unkown cases"	16:35 -18:00
Gala Diner		*20:00*
Sunday, 30 June, 2013		
Breakfast		*7:30 – 9:00*
Session VIII		
Chairs	**Ekaterina Zakharowa, Tomasz Kmieć**	9:00 - 9:10
Karl Eugen Mengel Mainz, Germany	Outcomes with ERT in infantile Pompe disease	9:10 - 9:30
Svetlana Volgina, Kazan, Russia	Art diagnosis and treatment of Fabry disease	9:30 - 9:50
Krzysztof Kałwak, Wrocław, Poland	Haematopoietic stem cell transplantation in metabolic diseases - worldwide recommendations and single centre own experience	9:50 - 10:10
Tomasz Wierzba Gdańsk, Poland	Functional analysis of cardiovascular system In patients with Williams syndrome	10:10 - 10:30
Anna Tylki-Szymańska Warsaw, Poland	Metabolic diseases in newborns	10:30 - 10:50
	Discussion	10:50 - 11:00
Coffee break		*11:00 - 11:20*
Session IX		
Chairs	**Maciej Machaczka, Anna Tylki-Szymańska**	
Małgorzata Musielak Warsaw, Poland	The role of enzymatic assay in Krabbe disease diagnostics	11:20 - 11:35
Jolanta Marucha Warsaw, Poland	Altered spine structure and biomechanical properties in patients with mucopolysaccharidosis	11:35 - 11:55
Tomasz Kmieć Warsaw, Poland	TIRCON - An EU-funded rare disease project dedicated to NBIA	11:55 - 12:10
Zbigniew Szymczak Warsaw, Poland	Injections of hyaluronic acid in patients with arthritis in mucopolysaccharidosis Type Iva - case study	12:10 - 12:25
Wioletta Żuraw Warsaw, Poland	Intravenous infusions in patients on ERT, analysis on patients with MPSII	12:25 - 12:40
Maciej Borowiec Łódź, Poland	Diagnosis and differentiation of rare monogenic forms of diabetes	12:40 - 12:55
	Discussion	12:55 - 13:00
Lunch		*13:00 - 14:00*
	Medical consultations	14:00 - 16:30
Holy Mass in intention of patients with rare diseases and their families		17:00 -18:00
Diner		*19:00*
Monday, 1 July, 2013		
Breakfast		*8:00 -9:00*
Transfer to the airport