Conference Programme

Thursady, June 27, 2019		
Lunch		***13:00 - 14:00***
Registration		*14:00 - 15:00*
Debate **„Do not miss a rare disease"** For patient organizations and members of the Federation of Patients with Rare Diseases of Central and Eastern Europe		
Teresa Matulka **Anna Tylki-Szymańska**	**Welcome and opening debate**	*15:00 - 15:10*
Teresa Matulka	**Presentation of the most important needs of patients with rare and ultra rare diseases**	*15:10 - 15:25*
Presentations of experts who will share their knowledge and experience		*15:25 - 16:00*
Patients with rare diseases - debate with health system experts 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 28, 2019		
Breakfast		***7:00 - 8:00***
Registration		*8:00 - 9:10*
Teresa Matulka **Anna Tylki-Szymańska**	**Welcome address**	*9:10 - 9:20*
Session I		
Chairs:	**Ekaterina Zakharova, Jakub Sikora**	***9:20 - 11:00***
Marc Dooms *Leuven, Belgium*	Rare Diseases in Medieval Europe	*9:20 - 9:35*
Olga Amaral *Porto, Portugal*	Not missing a rare disease: a matter of resilience and persistence	*9:35 - 9:50*
Mariusz Więckowski *Warsaw, Poland*	Changes in the OXPHOS proteome and alterations of mitochondrial parameters in fibroblasts of patients harboring defined mitochondrial disorders	*9:50 - 10:10*
Holger Prokisch *Munich, Germany*	The genetic landscape of paediatric mitochondrial diseases, a study of 1800 cases	*10:10 - 10:30*
David J. Begley *London, UK*	Delivery of ERT and other protien/peptide therapies across the blood-brain barrier: Are we targeting the right receptors for effective transcytosis?	*10:30 - 10:50*
	Discussion	*10:50 - 11:00*
Coffe break		***11:00 - 11:25***
Session II		
Chairs:	**Agnieszka Ługowska, Johannes Berger**	***11:25 - 13:00***
Jörn Oliver Sass *Bonn-Rhein-Sieg Univ, Germany*	Aminoacylases and Canavan Disease	*11:25 - 11:45*
Volkmar Gieselmann *Bonn, Germany*	Pathophysiology and treatment options for Metachromatic leukodystrophy	*11:45 - 12:05*
Barbara Oleksy *Warsaw, Poland*	A patient with leukoencephalopathy and temporal lobe cysts – a case report and differential diagnosis consideration	*12:05 - 12:20*
Hanna Mierzewska *Warsaw, Poland*	Leukodystrophy hypomyelinating	*12:20 - 12:35*
Jacek Pilch *Katowice, Poland*	Dysfunction of thyroid hormone transporter causes severe developmetal delay: A not identified disease, with potentially possible therapy	*12:35 - 12:50*
	Discussion	*12:50 - 13:00*
Lunch		***13:00 - 14:00***

Kontakt: 05-503 Głosków, ul. Radnych 9A, tel. (22) 757-81-29, fax (22) 757-81-97, kom. 601 30-04-52
Konto Bankowe: Bank Pekao S.A. Oddz. w Piasecznie, NR: 62 1240 6175 1111 0000 4568 0739
KRS: 0000060517, NIP: 123-09-99-634, Regon: 003448391

Session III		
Chairs:	**Małgorzata Krajewska-Walasek, Jörn Oliver Sass**	*14:00 - 16:00*
Jakub Sikora *Prague, Czech Republic*	LAMP2 deficiency (Danon disease) is an underdiagnosed X-linked cardiomyopathy – overview and pitfalls to clinical and laboratory diagnostics	*14:00 - 14:20*
Ekaterina Zakharova *Moscow, Russia*	Diagnosis of inherited metabolic diseases – the anadventure quest from symptoms to NGS	*14:20 - 14:40*
Galina Baydakova *Moscow, Russia*	Not missing a rare disease: a matter of resilience and persistence	*14:40 - 15:00*
Rafał Płoski *Warsaw, Poland*	Whole exome sequencing for diagnosis and discovery of novel rare diseases	*15:00 - 15:15*
Patryk Lipiński *Warsaw, Poland*	Congenital disorder of deglycosylation caused by mutations in NGLY1 gene	*15:15 - 15:30*
Anna Tylki-Szymańska *Warsaw, Poland*	Glycosylation disorders - ATP6AP1 deficiency - analysis of the progressive disease process	*15:30 - 15:50*
	Discussion	*15:50 - 16:00*
Coffe break		***16:00 - 16:25***
Session IV		
Chairs:	**Olga Amaral, David Begley**	*16:25 - 18:20*
Martina Zivna *Prague, Czech Republic*	Genetic, molecular and clinical aspects of Autosomal Dominant Tubulointerstitial Kidney Disease (ADTKD)	*16:25 - 16:45*
Lars Schlotawa *Goettingen, Germany*	A meta-analysis of published patient data improves the natural disease history in Multiple Sulfatase Deficiency	*16:45 - 17:05*
Mariusz Kujawa *Gdansk, Poland*	Evaluation of early changes in the course of alkaptonuria in children	*17:05 - 17:25*
Yuliya Nikalayeva *Gomel, Belarus*	Unexpected coma in newborn. Case report of citrullinemia type 1	*17:25 - 17:40*
Iryna Zhauniaronak *Minsk, Belarus*	Dynamics of clinical manifestations of the deficiency of long-chain 3-hydroxyacyl-CoA-dehydrogenase fatty acids (clinical case)	*17:40 - 17:55*
Nataliia Samonenko *Kyiv, Ukraine*	Enzyme replacement therapy in Ukraine.	*17:55 - 18:10*
	Discussion	*18:10 - 18:20*
Grill-dinner		***20:00***
Saturday, June 29, 2019		
Breakfast		***7:00 - 8:45***
Session V		
Chairs:	**Assel Tulebayeva, Holger Prokisch**	*9:00 - 11:00*
Sofia Pchelina *St. Petersburg, Russia*	Molecular basis of Parkinson's disease linked to mutations in the glucocerebrosidase gene	*9:00 - 9:20*
Johannes Berger *Vienna, Austria*	Macrophage dysregulation in X-linked adrenoleukodystrophy prevents self-limitation of brain lesions	*9:20 - 9:40*
Timothy M. Cox *Cambridge, UK*	Hereditary Fructose intolerance – Slavery and Sugar and its Medical Consequences	*9:40 - 10:00*
Magdalena Podlacha *Gdansk, Poland*	Therapeutic effects of genistein on cognitive abnormalities in Huntington's disease	*10:00 - 10:15*
Estera Rintz *Gdansk, Poland*	A new look at treatment of Huntington's disease - therapy with the use of genistein	*10:15 - 10:30*
Marc Dooms *Leuven, Belgium*	Patient Involvement in the Lifecycle of Medicines: the Gap Between Theory and Practice	*10:30 - 10:50*
	Discussion	*10:50 - 11:00*
Coffe break		***11:00 - 11:25***

Session VI		
Chairs:	**Hanna Mierzewska, Jacek Pilch**	*11:25 - 13:10*
Shunji Tomatsu *Wilmington, USA*	Development of AAV Gene Therapy for Morquio A Syndrome	*11:25 - 11:45*
Agnieszka Ługowska *Warsaw, Poland*	Mucolipidosis III (pseudo-Hurler polydystrophy) in an adult patient with cardiomyopathy and osteo-skeletal disorder	*11:45 - 12:00*
Ewa Kaczorowska *Gdansk, Poland*	Persistence of fetal hemoglobin in patients with intellectual disability, congenital malformations and dysmorphic features - Dias-Logan syndrome.	*12:00 - 12:15*
Katarzyna Niepokój *Warsaw, Poland*	Hearing loss - the most common disease of the senses, and yet a rare disease	*12:15 - 12:30*
Aleksandra Kuźniar-Pałka *Warsaw, Poland*	Epilepsy in patients with Trisomy 21	*12:30 - 12:45*
Aksana Prybushenia *Minsk, Belarus*	Fetal tumors: prenatal diagnosis and genetic counselling	*12:45 - 13:00*
	Discussion	*13:00 - 13:10*
Lunch		***13:10 - 14:00***

Session VII
Dysmorphology meeting

RASopathies as an example of disorders of the RAS / MAPK pathway - presentation of clinical features and molecular basis in Polish patients with Noonan, Costello and cardio-facial-skin syndromes
Prof. Małgorzata Krajewska-Walasek

Chairs:	**Robert Śmigiel, Ewa Obersztyn**	*14:00 - 16:00*
Małgorzata Krajewska-Walasek *Warsaw, Poland*	The RASopathies, an example of RAS/MAPK pathway disturbances – updated review of clinical and molecular results of Polish Noonan syndrome (NS) and NS-related patients	*14:00 - 14:15*
Magdalena Pelc *Warsaw, Poland*	The RASopathies: consequences of Ras/MAPK pathway dysregulation	*14:15 - 14:35*
Monika Gos *Warsaw, Poland*	New possibilities in the molecular diagnosis of RASopathies	*14:35 - 15:00*
Natalia Braun-Walicka *Warsaw, Poland*	Research on a pathogenesis of cognitive impairments in Noonan syndrome using modern psychological tools and neuroimaging techniques – an attempt of correlation with RAS/MAPK signaling pathway genes mutations	*15:00 - 15:15*
Anna Wałdoch *Gdansk, Poland*	Cardiac problems in the Noonan syndrome - own experience	*15:15 - 15:30*
Ewa Obersztyn *Warsaw, Poland*	RASopathies - unusual findings or expanding the phenotype and overlapping syndromes	*15:30 - 15:45*
Dorota Wicher *Warsaw, Poland*	Molecular variants in *KMT2D* and *SOS1* genes – disease causing variants or accidental findings?	*15:45 - 16:00*
Coffe break		***16:00 - 16:15***

Dysmorphology meeting

Chairs:	**Jolanta Wierzba, Aleksandra Jezela-Stanek**	*16:15 - 18:00*
Paweł Własienko *Warsaw, Poland*	Prenatal diagnosis of Noonan syndrome - the importance of ultrasound assessment and indications for the analysis of RAS/MAPK gene mutation	*16:15 - 16:30*
Aleksandra Jezela-Stanek *Gdansk, Poland*	What's new about cardio-facio-cutaneous syndrome since 2015?	*16:30 - 16:45*
Agata Skórka *Gdansk, Poland*	A story about phenotypic spectrum of Polish patients with Costello syndrome finished with a RASopathy surprise	*16:45 - 17:00*
Robert Śmigiel *Wroclaw, Poland*	Phenotypic differential diagnosis of RASopathies in clinical geneticist practice	*17:00 - 17:15*
Krystyna Chrzanowska *Warsaw, Poland*	Changes in the cell cycle as a new aspects in the pathogenesis of mucopolysaccharidoses	*17:15 - 17:30*
Magdalena Kalwas-Śliwińska *Warsaw, Poland*	ORPHANET - why is it worth to know it better?	*17:30 - 17:45*
	Discussion	*17:45 - 18:00*
Gala dinner		***20:00***

Sunday, June 30, 2019		
Breakfast		***7:30 - 9:00***
Session VIII		
Chairs:	**Gražina Kleinotiene, Timothy M. Cox**	***9:30 - 12:10***
Assel Tulebayeva *Almaty, Kazakhstan*	The sleep-disordered breathing in patients with mucopolysaccharidosis in the Republic of Kazakhstan	*9:30 - 9:45*
Grzegorz Węgrzyn *Gdansk, Poland*	Transcriptomic analysis in all types of mucopolysaccharidosis reveals complexity of disturbances in cellular processes	*9:45 - 10:05*
Joanna Brokowska *Gdansk, Poland*	Changes in the cell cycle as a new aspects in the pathogenesis of mucopolysaccharidoses	*10:05 - 10:20*
Zuzanna Cyske *Gdansk, Poland*	Changes in the cytoskeleton in mucopolysaccharidoses	*10:20 - 10:35*
Karolina Pierzynowska *Gdansk, Poland*	Disturbances in the effectiveness of the autophagy process as a new aspect of mucopolysaccharidosis pathogenesis: transcriptomic and cellular studies	*10:35 - 10:50*
Coffe break		***10:50 - 11:20***
Lidia Gaffke *Gdansk, Poland*	Changes in the vacuolar transport as a unknown aspect of mucopolysaccharidosis patogenesis	*11:20 - 11:35*
Svetlana Volgina *Kazan, Russia*	Diagnosis of glycosylphosphatidylinositol biosynthesis defect 11	*11:35 - 11:50*
	Discussion	*11:50 - 12:00*
Anna Tylki Szymańska Teresa Matulka	**Closing remarks and summary**	*12:00 - 12:10*
Lunch		***13:00 - 14:00***
Medical consultation		*14:00 - 17:00*
Consultations of unexplained cases		*14:00 - 17:00*
Dinner		***18:00 - 19:00***
Integration meeting "Café"		*19:00 - 23:00*
Monday, July 1, 2019		
Breakfast		***7:00 - 9:00***
Transfer	  	

Credit points:

According to the Minister of Health of October 6th, 2004 related with completing training requirement for doctors and dentists, a participant is entitled to get **33 credit points**.

During XVII International Rare Diseases Conference «Don't miss a rare disease» laboratory diagnosticians will be entitled to **6 credit points** awarded by National Chamber of Laboratory Diagnosticians (KIDL).

W imieniu Komitetu Naukowego, organizacyjnego i członków Stowarzyszenia Chorych na MPS i Choroby Rzadkie serdecznie zapraszamy do udziału w konferencji wszystkich tych, którzy chcą zrozumieć choroby rzadkie oraz poznać tych niezwykle rzadkich pacjentów.

Nie przegapcie nas, naszych trosk, naszych obaw i naszego lęku o przyszłość naszych niezwykle rzadkich dzieci, gdyż nie ma większej tragedii dla rodziców od nieuleczalnej choroby dziecka.

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Kontakt: 05-503 Głosków, ul. Radnych 9A, tel. (22) 757-81-29, fax (22) 757-81-97, kom. 601 30-04-52
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