18th International Conference on Rare Diseases

„What's new in rare diseases"

35th anniversary of the Association

June 27 - 29, 2025, Serock near Warsaw

Programme

Friday, June 27, 2025		
	Registration	*10:00 - 13:00*
Lunch		**13:00 - 14:00**
Inauguration of the 35th anniversary of the Polish Association of Patients with Mucopolysaccharidosis and Rare Diseases		
Teresa Matulka **Anna Tylki-Szymańska**	Opening ceremony	14:30 - 15:00
Anna Tylki-Szymańska	Introduction	15:00 - 15:20
Teresa Matulka	Summary of 35th anniversary of the Polish Association of Patients with Mucopolysaccharidosis and Rare Diseases	15:20 - 15:40
	Acknowledgments	15:40 - 17:00
Kim Angel **Executive Director**	Building Bridges, Transforming Lives: The IMPSN's Role in Worldwide MPS and Related LSD Advocacy	17:00 - 17:15
Lut de Baere **President**	„BOKS on a wave" Belgian Patient Organization for Rare Metabolic Diseases (BOKS)	17:15 - 17:30
Samat Ramazanov **Head of Foundation**	Overview of Zhana Omyr Patient's Organization's work in Kazakhstan	17:30 - 17:45
Alev Şaylan **Board Member**	Latest Updates on Rare Diseases in Türkiye and works of the MPS-LH Association (MPSTURK)	17.45 – 18.00
Grill-dinner		***19:00***
Saturday, June 28, 2025		
Breakfast		**8:00 - 9:00**
Session I		
Chairs:		
Ségolène Aymé **MD, PhD**	Achievements and remaining challengges in the field of rare diseases	9:00 - 9:20
Maurizio Scarpa **MD, PhD**	Revolutionizing Rare Disease Diagnosis with Artificial Intelligence	9:20 - 9:40
Marc Dooms **Pharm, PhD**	Orphan Devices	9:40 - 10:00
Sebastian Wardak **MD**	WES isn't as daunting as it seems – Diagnostic experience of MedGen Medical Center	10:00 - 10:20
Ekaterina Zakharova **MD, PhD**	Inherited Metabolic Disorders: How to Defeat the Dragon of Diagnostic Uncertainty	10:20 - 10:40
Jakub Sikora **MD, PhD**	Genetic Causes and Therapy of Rare Hereditary Amyloid A amyloidosis	10:40 - 11:00
Coffe break		**11:00 - 11:30**
Session II		
Chairs:		

Timothy M. Cox MD, PhD	Hereditary Fructose Intolerance - outcomes, expectations and a future	11:30 - 11:50
Mariusz Więckowski MD, PhD	Basic research for understanding of the pathomechanisms of rare and ultra-rare diseases such as NBIA and PACS2 Syndrome	11:50 - 12:10
Aneta Szulc MSc	Changes in Gene Expression Profiles in Cellular and Animal Models of Huntington's Disease	12:10 -12:25
Jörn Oliver Sass Prof. Dr. rer.nat.	Inborn Errors of Ketogenesis (Ketone Body Synthesis)	12:25-12:45
Ewa Ziółkowska PhD	Identifying and treating pathological changes outside the central nervous system in multiple forms of Batten disease	12:45-13:05
Lunch		**13:05 - 14:00**

Session III

Chairs:		
Assel Tulebayeva MD, PhD	Epidemiology of mucopolysaccharidoses in Kazakhstan	14:00 - 14:20
Lena Sagi-Dain MD	Genetic screening and testing in Israel - lessons learned	14:20 - 14:40
Anna Tylki Szymańska MD, PhD	Effects of *GBA1* Variants and Prenatal Exposition on the Glucosylsphingosine (Lyso-Gb1) Levels in Gaucher Disease Carriers	14:40 - 15:00
Ladislav Kuchař PhD	Fabry disease and AGALopathy: diagnostics, differentiation of X-linked phenotypes in females and therapy monitoring utilizing lysoGb3, enzyme activity and molecular genetics	15:00 - 15:20
Zbigniew Żuber MD, PhD	Rare diseases in rheumatology	15:20 - 15:40
Coffe break		**15:40 - 16:10**

Session IV

Chairs:		
Patryk Lipiński MD, PHD	*ATP6AP1*-congenital disorder of glycosylation: 2025 update	16:10 - 16:30
Agnieszka Ługowska PhD	Oncological Aspects of Lysosomal Storage Diseases	16:30 - 16:50
Grazina Kleinotienė MD	A diagnosis challenge: thrombocytopenia, systemic lupus erythematosus, and a genetic variant of Bernard-Soulier syndrome	16:50 - 17:05
Zita Krumina MD, PhD	Disease associated with *RINT1* gene. Case report	17:05 - 17:20
Weronika Wójtowicz MSc	The Hidden Burden: PTSD Symptoms and Quality of Life in Caregivers of Children with Rare Genetic Conditions	17:20 - 17:40
Edyta Radzanowska-Alenowicz, MSc	The Role of the Rehabilitation Manager in the Support System for People with Rare Diseases - A Profession of the Future or a Real Necessity?	17:40 - 17:55
Gala dinner		***19:00***

Sunday, June 29, 2025

Breakfast		**8:00 - 9:00**

Session V

Chairs:		
Shunji Tomatsu MD, PhD	Gene Therapy for Mucopolysaccharidosis IVA	9:00 - 9:20
Grzegorz Węgrzyn PhD	Mucopolysaccharidoses – monogenic diseases with a complex molecular mechanism	9:20 - 9:40
Karolina Pierzynowska PhD	Disruptions of Pathways Leading to Ferroptosis in a Mouse Model of Mucopolysaccharidosis Type I	9:40 - 9:55
Estera Rintz PhD	Immune response and autophagy in Mucopolysaccharidosis IIIB Mouse Model after resveratrol treatment	9:55 - 10:10
Karolina Wiśniewska MSc	Identification of Genetic, Biochemical, and Cellular Factors Affecting the Development of Sanfilippo Syndrome	10:10 - 10:25
Coffe break		**10:25 - 11:00**

Session VI

Chairs:		
Magdalena Żabińska MSc	The Role of The GPER Receptor in Mucopolysaccharidosis Type I	11:00 - 11:15
Zuzanna Cyske MSc	Understanding MPS-plus: What Do We Know About the Molecular Basis of the Disease?	11:15 - 11:30
Agnieszka Różdżyńska-Świątkowska, MD	Body Height of MPS I and II Patients after Hematopoietic Stem Cell Transplantation: The Impact of Dermatan Sulphate	11:30 - 11:50
Zbigniew Szymczak MD	Possibilities of surgical treatment for advanced degenerative changes of the hip joints in a patient with mucopolysaccharidosis IVA - a case report	11:50 - 12:10
Mariana Ryznychuk MD	Mucolipidosis type II: a case report	12:10 - 12:30
Anna Tylki-Szymańska Teresa Matulka	**Closing the conference**	12:30 – 13:00
Lunch		**13:00 - 14:00**
	Session for patients	
	Medical consultation - unexplained cases	14:00- 17:00
Dinner		**18:00**
	Monday, June 30, 2025	
Breakfast		**8:00 - 9:00**
Transfer	  	

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Uniwersytecki Szpital Dziecięcy w Krakowie

ul. Wielicka 265, 30-663 Kraków

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