International Symposium Rare Disease Day 2021 - Warmia and Mazury, Olsztyn
„Long diagnosis, misdiagnosis, or no diagnosis ... – Rare diseases under the radar"

Ladies and gentlemen,

on behalf of the Organizers, I have the pleasure to invite you to participate in the International Symposium Rare Diseases Day 2021 *"Long diagnosis, misdiagnosis, or no diagnosis – Rare diseases under the radar."* organized on the occasion of the Rare Disease Day by Regional Specialized Children's Hospital in Olsztyn and the University of Warmia and Mazury in Olsztyn under auspices of Eurocordis (rarediseaseday.org).

The Symposium will be dedicated to the use of multi-omnics methods, including next-generation sequencing (NGS), in the diagnosis of rare diseases, such as also in discovering and describing new rare diseases. They will be discussed the possibility of personalized therapies in patients with selected rare neuronal channelopathies. Among the invited speakers there are representatives of centers associated with three European Reference Networks - ERN-RND, ERN-ITHACA, ERN-CRANIO. We will also host small patients of our Hospital diagnosed with congenital neuronal channelopathies at various stages of implementing personalized therapy.

All interested persons are cordially invited to participate in the International Symposium Rare Diseases Day 2021 *"Long diagnosis, misdiagnosis, or no diagnosis – Rare diseases under the radar."*

Małgorzata Pawłowicz MD, PhD
Coordinator of Department of Pediatric Neurogenetics and Rare Diseases
Regional Specialized Children's Hospital in Olsztyn

DETAILS OF THE EVENT

Organisers: **Department of Pediatric Neurogenetics and Rare Diseases, Regional Specialized Children's Hospital in Olsztyn**
Department of Human Physiology and Pathophysiology, Collegium Medicum, University of Warmia and Mazury in Olsztyn

Date and time: **27 February 2021, 8:30 a.m. – 4:45 p.m.**

Symposium Venue: **online, Microsoft Teams platform**

Application: **via e-mail to chorobyrzadkiewssd@gmail.com by 26 February 2021 (in the e-mail, please provide your name and surname, e-mail address, academic degree, license number in the case of doctors) - the number of places is limited**

Participation fee: **free of charge**

Confirmation of participation: **personal certificate, award of 6 education points**

SYMPOSIUM PROGRAM

8:30 – 9:00 **Symposium registration – Teams platform login**

9:00 – 9:15 **Official welcome**
Krystyna Piskorz-Ogórek PhD
General Manager
Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland

9:15 – 9:30 **Rare Disease Day 2021 „Rare diseases under the radar" - introduction**
Małgorzata Pawłowicz MD, PhD
Head, Department of Pediatric Neurogenetics and Rare Diseases
Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland

Session I - Multi-omics studies for undiagnosed rare diseases
"Without knowing neither the direction nor the destination, you will not get a ticket." Les Brown

Session plan: 30-40 min. talks, plus 5-10 min. for discussion
Official language of session: English

09:30 **Special talk: Undiagnosed Rare Disease Project of Catalonia (URD-Cat Project)**
Prof. Alfons Macaya MD, PhD
Head, Pediatric Neurology Section
Professor of Pediatrics, Autonomous University of Barcelona
Hospital Universitari Vall d'Hebron, Barcelona, Spain

10:15 **Whole-exome and whole-genome sequencing in the diagnosis and therapy of new human diseases**
Prof. Rafał Płoski MD, PhD
Head, Department of Medical Genetics
Warsaw Medical University, Warsaw, Poland

Session II – Rare congenital neuronal and peripheral channelopathies
"You can get lost from any good path. You can turn back from any wrong path." Aldona Różanek

Session plan: 20-25 min. talks, plus 5-10 min. for discussion
Official language of session: Polish

11:00 **Coded time and destination – ion channel functions in early brain development and neurodevelopmental disorders related to their dysfunctions**
Małgorzata Pawłowicz MD, PhD
Head, Department of Pediatric Neurogenetics and Rare Diseases
Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland
Assistant Professor, Department of Human Physiology and Pathophysiology, Collegium Medicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland

11:30 **Keto-tracking - an interdisciplinary team and its tasks in implementing a ketogenic diet in patients with rare congenital neuronal channelopathies**
Adrianna Różniecka Msc
Coordinating nurse, Department of Pediatric Neurogenetics and Rare Diseases, Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland
Department of Nursing, School of Public Health, Collegium Medicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland

11:50 **Decoded pathways - implementation of a ketogenic diet in patients with rare congenital neuronal channelopathies from the point of view of a dietitian**
Anna Hinburg PhD, Monika Szostek Msc
Nutrition Department
Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland

12:15 **Neurotoxin-induced plasticity of sensory and sympathetic neurons - a new paradigm in the treatment of peripheral channelopathies?**
Prof. Mariusz Majewski PhD
Head, Department of Human Physiology and Pathophysiology, Collegium Medicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland

12:45 – 13:00 Coffee break

Session III – Clinical cases presentation, interviews with patients' families with rare congenital channelopathies
"There will come a time when you find that everything is over. This will be the beginning." Louis L'Amour

Session plan: 10-15 min. talks, plus 10-15 min. for discussion and interview
Official language of session: Polish

13:00 **In search of lost time: pathogenic variants in the *SCN1A* gene - clinical cases presentation and interview with families**
Małgorzata Pawłowicz MD, PhD, Agnieszka Dutka MD
Department of Pediatric Neurogenetics and Rare Diseases, Department of Pediatric Neurology, Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland
Department of Human Physiology and Pathophysiology, Collegium Medicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland

13:30 **In search of the lost gene: pathogenic variants in the *KCNA1* gene - clinical case presentation and interview with the family**
Małgorzata Pawłowicz MD, PhD
Department of Pediatric Neurogenetics and Rare Diseases, Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland
Department of Human Physiology and Pathophysiology, Collegium Medicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland

14:00 **In search of the lost phenotype: pathogenic variants in the *CACNA1A* gene - clinical case presentation and family interview**
Oliwia Czyżniewska, Małgorzata Pawłowicz MD, PhD
Student Research Group of Clinical Genetics, Department of Human Physiology and Pathophysiology, Collegium Medicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland
Department of Pediatric Neurogenetics and Rare Diseases, Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland

14:00 – 14:30 Lunch break

Session IV – Rare acquired neuronal channelopathies
"Your "now" is not your "always"" John Green

Session plan: 20-25 min. talks, plus 5-10 min. for discussion
Official language of session: Polish

14:30 **Rare autoimmune channelopathies: a concise review**
Julia Kulczycka MD, PhD
Senior Assistant, Autoantibody Diagnostics Laboratory
University Clinical Centre of Gdansk, Gdansk, Poland

15:00 **Neuromyelitis optica in the theory and practice**
Aleksandra Melnyk MD, Agnieszka Dutka MD
Department of Pediatric Neurology
Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland
Department of Clinical Pediatrics, Collegium Biomedicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland

Session V – "Neurosurgical channelopathies" in rare diseases
"The difficulties overcome are the opportunities won." Winston Churchill

Session plan: 20-25 min. talks, plus 5-10 min. for discussion
Official language of session: Polish

15:30 **Spine malformations coexisting with congenital craniofacial malformations - diagnostic possibilities and neurosurgical treatment**
Prof. Dawid Larysz MD, PhD
Professor, Department of Clinical Pediatrics, Collegium Biomedicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland

16:00 **Modern operational methods in rare craniofacial malformations**
Krzysztof Dowgierd MD, PhD
Head, Center of Craniofacial Malformation – Associate Member ERN Cranio
Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland

16:30 – 16:45 **Concluding remarks and closing Symposium**
Krystyna Piskorz-Ogórek PhD, Małgorzata Pawłowicz MD, PhD
Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland

SPEAKERS

Dr. Alfons Macaya is a Child Neurologist and Professor of Pediatrics in Barcelona, Spain. He received his MD degree from Autonomous University of Barcelona (UAB) and then trained in Pediatrics and then Pediatric Neurology at Vall d'Hebron Children's Hospital. He was next (1990-1993) a post-doc research fellow at the Neuroscience Institute, Columbia University, New York, NY. His Ph D thesis dealt with excitotoxic injury to the newborn striatum and apoptosis of dopaminergic neurons.

Dr. Macaya is currently the head of department of Pediatric Neurology at Vall d'Hebron Children's Hospital in Barcelona and the coordinator of the Pediatric Neurology Research Group at the Vall d'Hebron Research Institute, UAB. Over the last two decades he has devoted his research efforts to the field of pediatric neurogenetics, with a particular interest in rare neurological diseases and the molecular basis of neurological paroxysmal disorders, including developmental epilepsies, migraine and movement disorders.

He is the recipient of the award to "Excellence in Research" by the Barcelona Medical College in 2018. He is also a past-president of the Spanish Pediatric Neurology Society (2016-2018) and currently a member of its board. He also acts as liaison of the European Pediatric Neurology Society (EPNS) and the European Reference Network for Rare Neurological Diseases (ERN-RND), also being a board member of the latter. He was a member of the EPNS Committee of National Advisors for training and education of Pediatric Neurology in Europe. He serves at the editorial boards of several pediatric and pediatric neurology journals and is an active reviewer for national/international research funding agencies. He is the director of the "Master in Pediatric Neurology" at the UAB.

Dr. Macaya has authored over 170 peer-reviewed scientific articles, in the Neuroscience, Clinical Neurology, Genetics and Pediatrics fields. He authored or co-authored 75 of these publications in 2016-2020 and directed nine doctoral theses.

Dr. Macaya's lab experimental approaches currently include analysis of NGS and phenomics (multi-omics) data from patients with rare neurological diseases and development of in vitro cell models to explore disease-causality of novel genetic variants and gene pathways, including ongoing work with neuronal transdifferentiation.

Prof. Rafał Płoski, Professor of Genetics, the Head of Medical Genetics Department of Warsaw Medical University. Graduated Warsaw Medical Academy (1990) and got his PhD at the University of Oslo (1995). In his lab Prof. Płoski has introduced next generation NGS for diagnostic and research use. The main current focus of his activity is whole exome sequencing on Illumina HiSeq platform for the diagnosis of human monogenic disorders as well as genomewide methylation analysis using NGS. Prof. Płoski is a specialist in laboratory medical genetics and laboratory forensic genetics; he also serves as an expert witness in human genetics at Regional Court of Justice in Warsaw. Prof. Płoski has published > 350 research papers from the area of human genetics which have been cited > 6000 times, (Prof. Płoski's Hirsh Index is 37). His current research concerns searching for novel monogenic diseases in human using approaches including, among others, studies of disease discordant monozygotic twins, mapping of break point regions in symptomatic balanced chromosomal translocations as well as development of novel bioinformatic tools for the analysis of WES data.

Dr. Małgorzata Pawłowicz pediatrician, pediatric neurologist, currently undergoing specialist training in clinical genetics. Her doctoral thesis in the field of pediatric diabetology on the genotype-phenotype correlation in the group of children and adolescents with newly diagnosed type 1 diabetes was distinguished as important for the development of the Pomeranian Voivodeship in the InnoDoktorant scholarship competition organized by the Pomeranian Voivodeship and the European Union. Since her medical studies, she has been fascinated by the idea of personalized medicine. She implements modern procedures from 4P medicine in her research and clinical work. Currently, the Coordinator of the Department of Pediatric Neurogenetics and Rare Disease in the Regional Specialized Children's Hospital in Olsztyn - a center applying to the European Reference Network for rare congenital malformation and syndromes with intellectual and other neurodevelopmental disorders (ERN-ITHACA). In the diagnostic and therapeutical fields of neurogenetic diseases, Dr. Małgorzata Pawłowicz co-works with leading national centers: the Department of Medical Genetics of the Medical University of Warsaw and the Department of Medical Genetics of the Memorial Institute - Children's Health Center in Warsaw. Member of the European Pediatric Neurology Society (EPNS). Leader of several research and research&development projects financed by national and European research funds. Currently also an assistant professor at the Department of Human Physiology and Pathophysiology, Collegium Biomedicum, University of Warmia and Mazury in Olsztyn.

Adrianna Rożniecka Msc, graduate of the Faculty of Medical Sciences at the University of Warmia and Mazury in the field of Nursing (2016). During third-degree studies in medicine at the University of Warmia and Mazury in Olsztyn and specialization training in pediatric nursing. Coordinating nurse, Department of Pediatric Neurogenetics and Rare Diseases of the Regional Specialized Children's Hospital in Olsztyn. Assistant at the Department of Nursing at the School of Public Health, Collegium Medicum, University of Warmia and Mazury in Olsztyn. She is preparing a doctoral dissertation on the assessment of the quality of life of children and adolescents with primary headaches.

Dr. Anna Hinburg clinical dietician. She obtained the professional qualifications of a dietician in 2002. A graduate of the Faculty of Food Sciences at the University of Warmia and Mazury in Olsztyn, specialization: food technology and human nutrition. PhD in food technology and nutrition. She deals with nutritional education in the Regional Specialized Children's Hospital in Olsztyn. She is a member of the Polish Society of Clinical Nutrition for Children.

Co-author of the textbooks "Selected health education programs in pediatrics" and "Pediatrics and pediatric nursing", publications on the nutrition evaluation of overweight and obese children and adolescents and the impact of the application of nutritional recommendations on body composition and biochemical parameters in obese adult population.

Monika Szostek Msc, dietician of the Regional Specialized Children's Hospital in Olsztyn. At the Hospital, she cooperates with metabolic, endocrinological and diabetic outpatient clinics. She completed 3-year BA studies at the University of Warmia and Mazury in Olsztyn. She continued her master's studies at the Medical University of Gdańsk and the Warsaw University of Life Sciences. During her master's studies, she gained experience during an internship at the Regional Rehabilitation Hospital for Children in America.

Agnieszka Dutka MD, pediatrician, pediatric neurologist. A graduate of the Faculty of Medicine of the Medical Academy in Gdańsk (2004). She completed her specialization training in pediatrics (2012) and pediatric neurology (2016) at the Regional Specialized Children's Hospital in Olsztyn. Currently an assistant at the Department of Pediatric Neurology of the Regional Specialized Children's Hospital in Olsztyn. Co-author of the chapter "Central nervous system malformations" in polish "Standards of diagnostic and therapeutic treatment in diseases of the nervous system of children and adolescents", edited by prof. Barbara Steinborn.

Dr. Julia Kulczycka, graduate of the Faculty of Medicine at the Medical Academy in Gdańsk, specialist in the field of Laboratory Medical Immunology, Senior Assistant at the Laboratory of Clinical Immunology and Transplantology at University Clinical Centre in Gdańsk. The area of interest is the diagnosis of autoimmune and allergic diseases. Lecturer of the Medical University of Gdańsk.

Aleksandra Melnyk MD, graduate of the Faculty of Medicine at the Medical Academy in Gdańsk, specialist in pediatrics and pediatric neurology, deputy head of the Department of Pediatric Neurology of the Regional Specialized Children's Hospital in Olsztyn. The area of interest is inflammatory and demyelinating diseases of the central nervous system. Lecturer of the Department of Clinical Pediatrics, Collegium Biomedicum, University of Warmia and Mazury in Olsztyn. Co-author of the chapter "Central nervous system malformations" in polish "Standards of diagnostic and therapeutic treatment in diseases of the nervous system of children and adolescents", edited by prof. Barbara Steinborn.

Prof. Dawid Larysz, specialist in neurosurgery, neuropsychologist, speech therapist. He graduated from the Faculty of Medicine in Katowice, Medical University of Silesia in Katowice. In 2013, he obtained professor title based on the monograph "Assessment of the results of treatment of isolated craniosynostosis in children, taking into account clinical, biomechanical and neurodevelopmental aspects". Additionally, he defended his master's thesis "Assessment of neurodevelopmental disorders in children treated for isolated premature atresia of cranial sutures" at the Faculty of History and Pedagogy of the University of Opole and completed postgraduate studies in molecular biology at the Jagiellonian University. Currently a professor at the Department of Clinical Pediatrics at the University of Warmia and Mazury in Olsztyn, consultant in the field of neurosurgery at the Oncology Center in Gliwice. Head of the Center for Treatment of CNS Disorders and Support for Child Development "Kangaroo" in Katowice. Since 2011, he has been a member of the interdisciplinary surgical team at the Center of Craniofacial Malformation of the Regional Specialized Children's Hospital in Olsztyn. Scientific interests are concentrated in three areas: 1) multi-disciplinary diagnostics and therapy of children with skull defects - he was the first in Poland to introduce minimally invasive, endoscopic methods of treatment of children with craniosynostoses into neurosurgical practice, in cooperation with engineers from the Silesian University of Technology in Katowice he developed and introduced into everyday practice clinical methods of preoperative planning and postoperative evaluation, neurosurgical procedures in virtual reality, including procedures using distraction osteogenesis methods. 2) molecular biology of skull defects and primary neoplasms of the central nervous system, 3) minimally invasive endoscopic methods of surgical treatment of tumors of the skull base and the use of modern microneurosurgical methods for the treatment of primary glial tumors of the brain.