First Global and Fifth International Symposium Rare Disease Day 2025 - Warmia and Mazury, Olsztyn, Poland *„More than you can imagine – more than you can dream of!"*

Ladies and gentlemen,

on behalf of the Organizers, I have the pleasure to invite you to participate in the First Global and Fifth International Symposium Rare Diseases Day 2025 *„ More than you can imagine – more than you can dream of!"* 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 EURORDIS (rarediseaseday.org).

The Symposium will be dedicated to the rare neurodevelopmental and neurometabolic disorders manifested by brain malformations, paroxysmal events and sleep disorders. Among the invited speakers there are representatives of centers associated with European Reference Networks - ERN-EpiCare, ERN-RND and ERN-ITHACA, like also with International Consortium for Research on Alternating Hemiplegia of Childhood (IAHCRC). We will also host representatives of patients with alternating hemiplegia of childhood and Friedreich ataxia with their families.

All interested persons are cordially invited to participate in the First Global and Fifth International Symposium Rare Diseases Day 2025 *„ More than you can imagine – more than you can dream of!"*.

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 Clinical Pediatrics, Collegium Medicum,
University of Warmia and Mazury in Olsztyn

Date and time: **8 March 2025, 8:00 a.m. – 6:10 p.m. CET**

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

Application: **via e-mail to chorobyrzadkiewssd@gmail.com by 7 March 2025 (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 9 education points**

SYMPOSIUM PROGRAM

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

8:30 – 8:45 **Official welcome**
Krystyna Piskorz-Ogórek PhD
General Manager
Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland

8:45 – 9:00 **Rare Disease Day 2025 - Introduction**
Małgorzata Pawłowicz MD, PhD
Head, Department of Pediatric Neurogenetics and Rare Diseases – Member of ERN ITHACA
Regional Specialized Children's Hospital in Olsztyn, Olsztyn, Poland

Session I – Neurogenetics – new diseases, new phenotypes, new perspectives
"Dreams attract dreams" – Ralph Waldo Emerson

Session plan: 20-45 min. talks, plus 5-15 min. for discussion
Official language of session: English

9:00 – 9:30 **Keynote lecture: Early-onset hereditary spastic paraplegia: clinical and genetic study**
Prof. Alfons Macaya MD, PhD
Head, Pediatric Neurology Section
Professor of Pediatrics, Autonomous University of Barcelona, Spain
Hospital Universitari Vall d'Hebron, Barcelona – Member of ERN-RND, Spain

9:45 Joint photo of the Symposium participants on Teams Platform

9:45 – 10:25 **Keynote lecture: Deciphering alternating hemiplegia of childhood – classic and new phenotypes**
Dr. Eleni Panagiotakaki MD, PhD
Senior Consultant, Department of Paediatric Clinical Epileptology, Sleep Disorders and Functional Neurology, University Hospitals of Lyon, Lyon – Member of EpiCARE-ERN, Deputy Coordinator of IAHCRC, France

10:30 – 11:00 **Keynote lecture: *KAT6A* gene – from mouse models to clinical trials**
Dr. Sarah Donoghue BMedSci, MBBS, FRACP
Women's and Children's Hospital, North Adelaide, Australia
Department of Neurodevelopmental Disability and Rehabilitation, Murdoch Children's Research Institute, Melbourne, Australia

11:10 – 11:30 **Keynote lecture: Whole exome sequencing and discovery of novel disorders**
Prof. Rafał Płoski MD, PhD
Head, Department of Medical Genetics
Warsaw Medical University, Warsaw, Poland

11:40 – 11:45 Joint photo of the Symposium participants on Teams Platform

11:45 – 12:00 Short coffee break

Session II – Rare neurodevelopmental and neurometabolic diseases – interplay between genes, seizures, sleep and diet

"Whatever you intend to do, whatever you dream of, start acting." – Johann Wolfgang Goethe

Session plan: 30-45 min. talks, plus 5-10 min. for discussion
Official language of session: English

12:00 – 12:45 **Pontocerebellar hypoplasia - review with case presentations**

Prof. F. Müjgan Sönmez MD, PhD
Yuksek Ihtisas University, Department of Pediatrics and Pediatric Neurology, Ankara, Türkiye
National Delegate of AOCNA
Advisory Board Member of AOCNA 2025 Congress
Vice-President of Neuromuscular Research Association, Ankara, Türkiye
Board of Neuromuscular Research Center of Lokman Hekim University, Ankara, Türkiye

12:50 – 13:10 **Extending phenotype of *SLC10A7*-related neurodevelopmental disorders – a clinical case with stare blankly and sleep-like attacks**

Prof. F. Müjgan Sönmez MD and co-authors: Asburce Olgac, Selen Has Ozhan, Halil, Tuna Akar, Husamettin Sargın, Tahir Atik
Yuksek Ihtisas University, Department of Pediatrics and Pediatric Neurology, Ankara, Türkiye
National Delegate of AOCNA
Advisory Board Member of AOCNA 2025 Congress
Vice-President of Neuromuscular Research Association, Ankara, Türkiye
Board of Neuromuscular Research Center of Lokman Hekim University, Ankara, Türkiye

13:15 – 13:45 **Genetic evaluation and rare diseases are commonly associated with sleep disorders: a discussion on diagnostic algorithms based on case experiences.**

Prof. Serdar Ceylaner MD, PhD
Intergen Genetic and Rare Diseases Diagnosis and Research Center, Ankara, Türkiye
Lokman Hekim University, Department of Medical Genetics, Ankara, Türkiye

13:50 – 14:05 **Pyridoxine supplementation in *PACS2*-related encephalopathy: a case report of possible precision therapy**

Dr. Marco Perulli MD, PhD
Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy

14:10 – 14:40 **Neurometabolic and neurodevelopmental diseases specific for Roma ethnicity – clinical case series**

Dr. Mária Giertlová MD, PhD
Outpatient Service of Medical Genetics, Unilabs Slovakia Ltd, Košice, Slovakia
Outpatient Service of Medical Genetics, Children's Faculty Hospital, Banská Bystrica, Slovakia
Clinics of Neurology, Medical Faculty of P.J. Šafárik, Košice, Slovakia

14:45 – 15:10 **Neurodevelopmental disorders - the role of detailed genotype-phenotype correlations**
Dr. Agnieszka Madej-Pilarczyk MD, PhD
Head, Department of Medical Genetics
Children's Memorial Health Institute, Warsaw – Member of ERN-ITHACA, Poland

15:15 – 16:00 Lunch break

Session III – Alternating hemiplegia of childhood – challenging paroxysmal disease of childhood
"Reality is made of the same yarn as dreams" – Władysław Stanisław Reymont

Session plan: 20-30 min. talks, plus 5 min. for discussion,
Official language of session: English, Polish

16:00 – 16:20 **Sleep in alternating hemiplegia of childhood: international multicentre survey and sleep-EEG study**
Dr. Francesco Fortunato MD, PhD
Institute of Neurology, Magna Graecia University, Catanzaro, Italy

16:25 – 16:55 **Keynote lecture: Alternating hemiplegia of childhood as a progressive disease and early childhood is a particularly vulnerable period**
Prof. Mohamad A. Mikati MD
Wilburt C. Davison Distinguished Professor of Pediatrics
Professor of Neurobiology
Division of Pediatric Neurology and Developmental Medicine
Director, Pediatric Epilepsy Translational Research Laboratory
Duke University Medical Center, Durham, United States of America

17:00 – 17:15 **Alternating hemiplegia of childhood – perspective of patients and patients' families**
Representatives of the Polish Association for People with AHC

17:15 Joint photo of the Symposium participants on Teams Platform

Session IV – Neurometabolic pathways - the key to successful clinical practice
"The most beautiful dreams are those that come true" – Herbert George Wells

Session plan: 20-30 min. talks, plus 5-10 min. for discussion and interview
Official language of session: English, Polish

17:15 – 17:35 **Sleep disorders and their genotype-phenotype correlations in Angelman syndrome – how can theory change our practice?**
Dr. Małgorzata Pawłowicz MD, PhD
Department of Pediatric Neurogenetics and Rare Diseases, Regional Specialized Children's Hospital in Olsztyn, Olsztyn – Member of ERN ITHACA, Poland
Department of Clinical Pediatrics, Collegium Medicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland

17:35 – 18:00 **Friedreich ataxia – new neurometabolic treatment: perspectives and challenges. Interview with patients' families**

Dr. Małgorzata Pawłowicz MD, PhD

Head, Department of Pediatric Neurogenetics and Rare Diseases, Regional Specialized Children's Hospital in Olsztyn, Olsztyn – Member of ERN ITHACA, Poland

Department of Clinical Pediatrics, Collegium Medicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland

18:00 – 18:10 **Concluding remarks and closing Symposium**

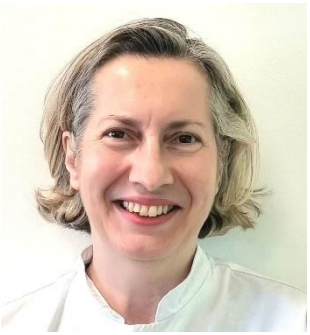
Dr. Małgorzata Pawłowicz MD, PhD

Head, Department of Pediatric Neurogenetics and Rare Diseases, Regional Specialized Children's Hospital in Olsztyn, Olsztyn – Member of ERN ITHACA, Poland

Department of Clinical Pediatrics, Collegium Medicum, University of Warmia and Mazury in Olsztyn, Olsztyn, Poland

SPEAKERS

Prof. Alfons Macaya is a Child Neurologist and Professor of Pediatrics in Barcelona, Spain. He trained in Pediatrics and Pediatric Neurology in Barcelona. He received his PhD title in 1992 after completing a research fellowship in Neuroscience at Columbia University, New York, which centered on excitotoxic injury to the newborn striatum and apoptosis of dopaminergic neurons. He is currently the head of department of Pediatric Neurology and Director of the Master in 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, Autonomous University of Barcelona. For the last 20 years his research has sought to improve diagnosis and treatment of various rare pediatric neurogenetic conditions, with particular interest in 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 a member of the Reference Network for Rare Neurological Diseases (ERN-RND) board. Prof. Macaya has authored over 200 peer-reviewed scientific articles, in the Neuroscience, Clinical Neurology, Genetics and Pediatrics fields (Scopus H-index 39).



Dr Eleni Panagiotakaki, senior Paediatric Neurologist – Epileptologist, is in charge, since 2008, of the Paediatric Clinical Epileptology unit, at University Hospitals of Lyon (HCL).

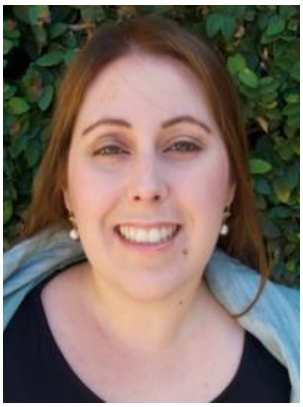
She received her MD degree from the University of Patras, Greece and her PhD in « Correlation of the genotype in Wilson's disease with the clinical and biochemical phenotype » from the Athens Medical School, where she also completed her Paediatric Training.

From 2004-2008, she worked as a Pediatric Neurologist at Robert Debré Hospital, in Paris. She was among the main partners of the ENRAH (European Network for Research on Alternating Hemiplegia, 2005-2011), and nEUroped projects (European Network of Rare Paediatric Neurological Diseases) financed respectively by the 6th and 7th Framework European programmes.

She is one of the founding members of the IAHCRC - International Consortium for Research on Alternating Hemiplegia of Childhood (iahcrc.net) created in 2014, and the leader of the phenotyping projects. She has recently (2022) been elected Deputy coordinator of the IAHCRC.

She is the Principal Investigator in Hospices Civils de Lyon in all studies concerning Alternating Hemiplegia of Childhood.

She is full member of EpiCARE-ERN, the European Reference Network for Rare and Complex Epilepsies (epi-care.eu).

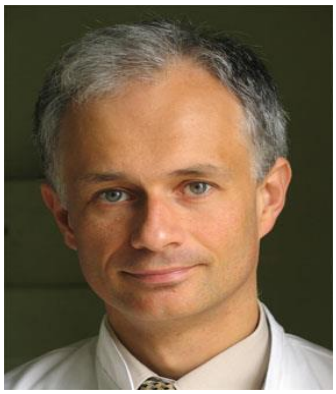


Dr. Sarah Donoghue is a general paediatrician who has a second fellowship in clinical genetics subspecialised metabolic medicine with Royal Australasian College of Physicians in December 2016. She cares for a wide range of both acute and chronic medical conditions in children from birth to adolescence.

Dr. Sarah Donoghue has particular interests in developmental delays/ regression, learning difficulties, autism spectrum disorders, genetic and metabolic conditions. Sarah has an interest in complex care co-ordination.

Dr. Sarah Donoghue was awarded her medical degree with honours in 2006 from the University of Tasmania. In addition to her training in genetics and metabolic medicine at the Royal Children's Hospital (RCH), she has participated in the Community Child Health Program conducted by RCH. Dr. Sarah Donoghue has also worked at Monash Medical Centre and trained in a number of specialist clinics for developmental disabilities and specific genetic syndromes (e.g., DiGeorge syndrome). Sarah commenced her PhD through the University of Melbourne in 2021 and is currently looking at whether chromatin disorders, in particular *KAT6A*, may be treatable. Her research aims to characterise changes in murine neural stem cells and human cortical neurons to find a biomarker to use in clinical trials.

Dr. Sarah Donoghue has publications in peer reviewed journals and has an interest in diagnostic methods to diagnose children with genetic and metabolic conditions. She was fortunate to train in genetics during the time research into the efficacy of exomes.



Prof. Rafał Płoski, Professor of Genetics, Head of the Department of Medical Genetics at the Warsaw Medical University. A graduate of the Medical Academy in Warsaw (1990). In 1995, after a 4-year stay in Norway, he obtained the title of 'Doctor of Medical Sciences' at the University of Oslo with a thesis titled "Genetic Predisposition to Juvenile Chronic Arthritis." From 1995 to 1997, he worked at the Laboratory of HLA of the Department of Pathophysiology and Immunology at the Institute of Rheumatology in Warsaw, serving successively as an assistant, assistant professor, and head of the laboratory.

From 1998 to 2005, he was employed at the Department of Forensic Medicine of the Medical University of Warsaw, where he established a laboratory for genetic research. From 2005 to 2006, he organized the Department of Medical Genetics at the Warsaw Medical University (ZGM) as the acting head. Since 2006, he has been the head of ZGM. In 2005, he obtained the postdoctoral degree in medical sciences with a thesis titled "Molecular Genetic Studies of the Polish Population and Their Application in Medical Sciences." In 2012, he was awarded the title of "Professor of Medical Sciences." Since 2009, he has been employed as a professor at the Warsaw Medical University.

In ZGM, Professor Płoski implements next-generation sequencing techniques for diagnostic and research purposes. His main area of interest is the development of personalized medicine based on exome and genome sequencing for diagnostics and the discovery of new human diseases.

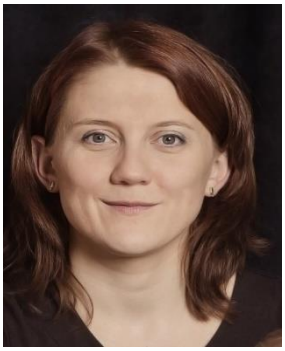
Professor Płoski is a specialist in laboratory medical genetics, forensic genetics, and is listed as an expert by the District Court in Warsaw in the field of genetics. He has published over 450 papers in the field of human genetics, with a total of over 8000 citations (H index = 45).

Prof. F. Müjgan Sönmez, Professor of Pediatrics and Pediatric Neurology. Graduated from Hacettepe University Medical School MD degree. In 1988 started to pediatrics training in Karadeniz Technical University (KTU), Faculty of Medicine, Department of Pediatrics, Trabzon, Türkiye. In 1994, 1996 and 2002: became Assistant Professor, Associate Professor and Professor of Pediatrics in KTU, respectively. Completed Child Neurology Fellowship in Hacettepe University Medical Faculty , Ankara, Türkiye in 1998. Founded the Child Neurology Department, Pediatric EEG Laboratory and Pediatric Video-EEG Monitorisation Unite and the Sleep Laboratory with Departments of Neurology, Chest Disease, Psychiatry and Ear-nose and Throath in KTU. Also, founded The Trabzon Branch of Neuromuscular Disorder Association of Türkiye and Chief of the branch between 2000-2012. Organized "IX National Neuromuscular Diseases Symposium and 10th National Turkish Child Neurology Congress and head of the Congress. She gives lectures as honorable lecturer in different universities in Ankara and Istanbul. She is president of Turkish Neuromuscular Research Association and the Board-Member of Neuromuscular Research Center of Lokman Hekim University in Ankara and coordinator of the developmental screening test. She has been working as a Professor in Yuksek Ihtisas University, Pediatrics and Pediatric Neurology Departments , Ankara, Türkiye since November 6, 2024. She is also National Delegate of AOCNA and Advisory Board Member of AOCNA 2025 Congress. Prof. Sonmez has published > 100 research papers from the area of Child Neurology which have been cited > 2000 times, (H-index is 24 in Researchgate). Her research interests are epilepsy, neurometabolic, neurogenetic, sleep and rare disorders and vitamin D associated neurological disorders. Member of Undiagnosed Diseases Network International (UDNI).

Prof. Serdar Ceylaner is a medical doctor - medical geneticist who focuses on rare and undiagnosed diseases for both diagnosis and scientific studies. He is the director, partner and founder of Intergen Genetics and Rare Diseases Diagnostic and Research Center and Lokman Hekim University, Department of Medical Genetics. Prof. Dr. Serdar Ceylaner is also: • European Union of Medical Specialists (UEMS), Vice President of the Rare and Undiagnosed Diseases Committee, • UEMS -Department of Medical Genetics- Turkish representative, • UEMS- Medical Genetics European Exam Committee member, • UEMS- Rare Disease European Exam Committee Chair, • UDNI- Undiagnosed Disease Network International Member, • Rare Diseases International (RDI)- CGN4RD Member. Prof. Serdar Ceylaner is the former president of the Turkish Medical Genetics Association and was a board member between 2009-2017. He was the founder of the Genetics Department of Zekai Tahir Burak Women's Health and Training Hospital between 1997 and 2017.

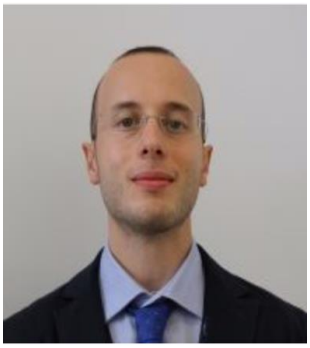
Prof. Serdar Ceylaner has focused on genetics and rare diseases and studies in this field for 30 years. Undiagnosed diseases, medical complications, and intensive care unit patients are the main research areas in recent years. He has experience in more than 250 international publications, 10 book chapters, more than 500 conferences, and more than 50 scientific projects.

Dr. Marco Perulli is a pediatric neurologist specializing in epilepsy, neonatal neurology and rare genetic conditions. During his PhD in neuroscience he was visiting research fellow at UCL in London. He currently has a post-doc research position at Fondazione Policlinico Universitario A. Gemelli IRCCS in Rome and he also works as a consultant pediatric neurologist.

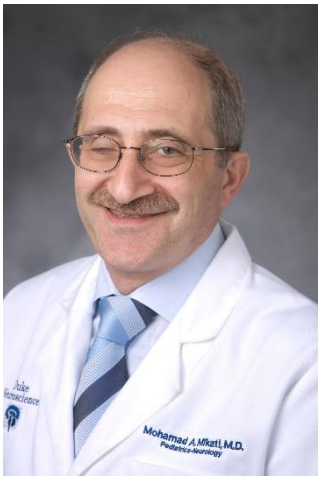


Dr. Mária Giertlová is a medical doctor graduated from the Faculty of Medicine at U.P. Šafárik in Košice (2006) and completed PhD in pharmacology (2010). Clinical geneticist since 2016, will complete her specialisation as a clinical geneticist in 2020. She focuses on genetic diagnosis of rare diseases with special interest in neurogenetics, neurodevelopmental and neurometabolic disorders. She is interested in the implementation of genomic analysis in clinical practice and the collaboration of clinical and scientific institutions. Overall, due to the significant Roma population in Slovakia and their social and ethnical exclusion, she has a passion for studying rare genetic diseases in the Roma population. She works at the Medical Genetics Outpatient Clinic in Košice (Unilabs Slovakia LtD) and at the Children's Faculty Hospital in Banská Bystrica. Due to her interest in rare disease research, she also works as a scientist at the Clinic of Neurology of the P.J. Šafárik Medical Faculty in Košice (since 2023) and has a scientific cooperation with the Department of Paediatrics and Inherited Metabolic Disorders, First Faculty of Medicine, Charles University and the General University Hospital in Prague. She is a member of the National Society of Medical Genetics and a regional expert in medical genetics (Košice region).

Dr. Agnieszka Madej-Pilarczyk specialist in clinical genetics and internal medicine, Head of the Department of Medical Genetics, Children's Memorial Health Institute (CMHI) in Warsaw. Formely she was employed in the Neuromusuclar Unit, Mossakowski Medical Research Centre, Polish Academy of Sciences and in the Department of Endocrinology, Medical University of Warsaw. She is a co-author of 55 papers, majority of them are devoted to laminopathies. Other areas of scientific interests: genetic aspets of rare diseases, including among others dysmorphic syndromes, intellectual disability, muscular dystrophies, myopathies, collagenopathies and rare forms of spinal muscular atrophy. Information scientist in Orphanet Poland. Participant of the Orphanet projects OD4RD (2023-2024) and OD4RD2 (2022-2025). Substitute representative of the CMHI in the Board of ERN-ITHACA Network.



Dr. Francesco Fortunato graduated cum laude with honorable mention in Medicine and Surgery from Magna Graecia University, Catanzaro, Italy on 23/07/2015. He completed the residency training in Neurology with laude in November 2020, discussing a experimental thesis entitled "Perampanel as first add-on choice in the treatment of mesial temporal lobe epilepsy: an observational real life study." He also earned cum laude, in July 2023, a PhD in "Biomarkers of clinical and complex diseases- Analysis of imaging data, neurophysiological biosignals and molecular profiling for the identification of biomarkers applied to Neurosciences", discussing the following experimental thesis: "Circulating micro-RNAs as potential novel diagnostic biomarkers to predict drug resistance in temporal lobe epilepsy: a pilot study". Dr. Fortunato did one-year research fellowship in genetics of the epilepsies at Queen Square Institute of Neurology, National Hospital for Neurology and Neurosurgery, Queen Square, London, UK, and Chalfont Centre for Epilepsy, under the supervision of Prof. Sanjay M. Sisodiya and Prof. Simona Balestrini. Dr. Francesco Fortunato works as post-doc research fellow and consultant neurologist at Department of Medical and Surgical Sciences, Magna Graecia University of Catanzaro. He is also active member of the "Promoting Internationalization Committee", Department of Medical and Surgical Sciences, Magna Graecia University of Catanzaro. Dr. Fortunato is co-author of 40 international publications with an H-index of 11 (source Scopus, 20/11/2023). He is an active member of many national and international scientific societies and study groups such as Epi 25 Collaborative Group, on the behalf of NIH USA Genome Institute (NHGRI) and International League Against Epilepsy (ILAE); and Enhancing Neuroimaging Genetics through Meta-Analysis (ENIGMA)- Epilepsy Working Group on the behalf of the University of Southern California (USC) USA, co-chairs Prof. Sanjay M. Sisodiya and Prof. Carrie McDonald. He also joined from 2023 as an active member the IAHCRC International Consortium for the Research on Alternating Hemiplegia of Childhood Duke University, on the behalf of School of Medicine, Durham, NC, USA (current scientific coordinator Prof. Mohamad Mikati). Dr. Fortunato's main research interests are clinical, neuroimaging and genetic aspects of the epilepsies.



Prof. Mohamad A. Mikati M.D., is the Wilburt C. Davison Professor of Pediatrics, Professor of Neurobiology, and Chief of the Division of Pediatric Neurology. Dr. Mikati's clinical research has centered on characterization and therapy of pediatric epilepsy and neurology syndromes, describing several new pediatric neurological entities with two carrying his name (POSSUM syndromes # 3708 and 4468), developing novel therapeutic strategies for epilepsy and related disorders particularly Alternating Hemiplegia of Childhood, and applying cutting edge genetic and Magnetic Resonance Imaging techniques to drug resistant pediatric epilepsy. In the laboratory he has elucidated mechanisms of seizure related neuronal injury, particularly those related to the ceramide pathway, and demonstrated neuroprotective effects of several agents including erythropoietin. Most recently he has concentrated his laboratory research on the pathophysiology of ATP1A3 dysfunction in the brain as model for epilepsy and of Alternating Hemiplegia of Childhood. He has more than 290 peer reviewed publications, 400 abstracts 41 chapters one book and two booklets. He also has more than 10,497 citations in the literature with an h-index of 58 and an i-10index of 190. Dr. Mikati has written chapters on epilepsy and related disorders in the major textbooks of Pediatrics and Pediatric Neurology including

Swaiman's Pediatric Neurology and Nelson's Pediatrics. Before joining Duke in 2008 he had completed his M.D. and Pediatric training at the American University of Beirut, his Neurology at the Massachusetts General Hospital, his Neurophysiology at Boston Children's Hospital and had been on the Faculty at Harvard as Director of Research in the Epilepsy Program at Boston Children's Hospital and then as Professor and Chairman, Department of Pediatrics, Founder and Director of the Adult and Pediatric Epilepsy Program at the American University of Beirut. Dr. Mikati has had several international leadership roles including being President of the Union of the Middle Eastern and Mediterranean Pediatric Societies, on the Standing Committee of the International Pediatric Association (IPA), Chair of the Strategic Advisory Group on Early Childhood Development of the IPA, Officer of the International Child Neurology Association, Consultant to UNICEF, WHO, and the American Board of Pediatrics. He was selected to organize and chair the American Epilepsy Society's Merritt-Putnam Symposium and was one of only two Pediatric Neurologists, initially chosen worldwide, on the WHO advisory committee for the International Classification of Disease. He has received several national and international honors including, among others, Merritt Putnam American Epilepsy Society Fellowship Award, Harvard Community Health Plan Peer recognition Award, Debs Research Award, Hamdan Award for contributions to Medicine, Hans Zellweger Award for contributions to Pediatric Neurology, Patient Choice Award and the Michael Frank Award for research and lifetime contributions to the field of Pediatric Neurology.

Dr. Małgorzata Pawłowicz pediatrician, pediatric neurologist, clinical geneticist and dietician, currently undergoing specialist training in metabolic pediatrics. 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 Region 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 – full member of the European Reference Network for rare congenital malformation and syndromes with intellectual and other neurodevelopmental disorders (ERN-ITHACA) and the Coordinator of the Expert Center for Rare Diseases established by the Minister of Health of the Republic of Poland as part of National Plan for Rare Diseases. Dr. Małgorzata Pawłowicz is appointed for the term of office 2024-2029 as a consultant of the Warmia and Mazury Region in the field of clinical genetics. 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), European Sleep Research Society (ESRS), American Academy of Sleep Medicine (AASM), Polish Genetic Society (PTG), Scientific Council of the Polish Association For People With AHC ahc-pl. Leader of several research and research & development projects financed by national and European research funds. Currently also the assistant professor and coordinator of clinical genetics at the Department of Clinical Pediatrics, Collegium Medicum, University of Warmia and Mazury in Olsztyn. Winner of the Hippocrates Award - Neurologist of the Year 2023 and 2024 of the Warmia and Mazury Region, awarded by patients. In 2024 awarded the Bronze Cross of Merit by the President of the Republic of Poland for her contribution to the development of the Warmia and Mazury Region.