Second Global and Sixth International Symposium Rare Disease Day 2026
with Mini-Symposium 2026 Sleep Awareness Month for Europe – Olsztyn, Poland
„Your hardest times will become your strength - Move forward and look ahead!"

Ladies and gentlemen,

on behalf of the Organizers, I have the pleasure to invite you to participate in the Second Global and Sixth International Symposium Rare Diseases Day 2026 *„Your hardest times will become your strength - Move forward and look ahead!"* organized on the occasion of the Rare Disease Day and Sleep Awareness Month by Regional Specialized Children's Hospital in Olsztyn and the University of Warmia and Mazury in Olsztyn under auspices of EURORDIS (rarediseaseday.org) and European Sleep Research Society (ESRS).

The Symposium will be dedicated to the rare neurodevelopmental and neurometabolic disorders manifested by brain malformations, paroxysmal events, movement disorders and sleep pattern abnormalities. 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, Dravet syndrome and adrenomyeloneuropathy with their families.

All interested persons are cordially invited to participate in the Second Global and Sixth International Symposium Rare Diseases Day 2026 *„Your hardest times will become your strength - Move forward and look ahead!"*

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: **14 March 2026, 8:00 a.m. – 7:30 p.m. CET**

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

Application: **via e-mail to chorobyrzadkiewssd@gmail.com by 13 March 2026 (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**

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 2026 - 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 4P – how far are we?
"With hope in your heart, every mountain becomes a mere hill" – Reinhold Messner

Session plan: 20-45 min. talks, plus 5-10 min. for discussion
Official language of session: English

9:00 – 9:25 **Keynote lecture: Newborn screening in neurological conditions**
Dr. Marco Perulli MD, PhD
Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy

9:30 – 10:10 **Keynote lecture: Epilepsy vs. movement disorders – from theory to practice**
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

10:15 – 10:35 **Keynote lecture: Variants of unknown significance – challenges, opportunities and tips for interpretation**
Prof. Rafał Płoski MD, PhD
Head, Department of Medical Genetics
Warsaw Medical University, Warsaw, Poland

10:40 – 10:45 Joint photo of the Symposium participants on Teams Platform

10:45 – 11:00 Short coffee break

Session II – Rare neurodevelopmental and neurometabolic diseases – gamechangers in our practice
"Embrace the unknown – it's where the most profound discoveries await." – Ray Manzarek

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

11:00 – 11:25 **Molecular profiling of Polish pediatric patients with epilepsy – a single-center experience of the Children's Memorial Health Institute in Warsaw**
Beata Chałupczyńska MSc
Department of Medical Genetics
Children's Memorial Health Institute, Warsaw – Member of ERN-ITHACA, Poland

11:30 – 11:55 **When it's not cerebral palsy: lessons from five molecular diagnoses**
Amaia Lasa Aranzasti MD
Department of Clinical and Molecular Genetics, Vall d'Hebron Hospital Universitari, Vall d'Hebron Barcelona Hospital Campus, Barcelona – Member of ERN-ITHACA, Spain
Medicine Genetics Group, Vall d'Hebron Institut de Recerca (VHIR), Vall d'Hebron Hospital Universitari, Vall d'Hebron Barcelona Hospital Campus, Barcelona, Spain

12:00 – 12:30 **Keynote lecture: Rapid differential diagnosis in neurodevelopmental disorders with next-generation genetic and artificial intelligence approaches. A broad perspective and a new diagnostic guide for outpatient clinics.**
Prof. Serdar Ceylaner MD, PhD
Intergen Genetic and Rare Diseases Diagnosis and Research Center, Ankara, Türkiye
Department of Medical Genetics, Lokman Hekim University, Ankara, Türkiye

12:35 – 13:05 **Keynote lecture: Developmental abnormalities of cerebellum with cases presentation**
Prof. F. Müjgan Sönmez MD, PhD
Department of Pediatrics and Pediatric Neurology, Yuksek Ihtisas University, Ankara, Türkiye
National Delegate of AOCNA
Vice-President of Neuromuscular Research Association, Ankara, Türkiye
Board of Neuromuscular Research Center of Lokman Hekim University, Ankara, Türkiye

13:10 – 13:30 **Disorders of creatine metabolism with cases presentation**
Prof. F. Müjgan Sönmez MD, PhD and co-authors: Halil Ibrahim Aydin
Department of Pediatrics and Pediatric Neurology, Yuksek Ihtisas University, Ankara, Türkiye
Department of Pediatrics, Sections of Inborn Errors of Metabolism, Faculty of Medicine, Başkent University, Ankara, Türkiye

13:35 – 13:55 **Atypical Zellweger syndrome: *PEX16* gene variant**
Associate Prof. Dilek Çavuşoğlu MD, PhD
Department of Pediatric Neurology, Faculty of Medicine, Afyonkarahisar Health Sciences University, Afyonkarahisar, Türkiye

14:00 – 14:25 **Keynote lecture: Beyond the White: the main pediatric leukodystrophies**
Dr. Caroline Sevin MD, PhD
Service de Neurologie Pédiatrique, Centre de Référence des Leucodystrophies, Centre de Référence Maladies Métaboliques, Hôpital Bicêtre, Paris – Member of ERN-RND, France

14:30 Joint photo of the Symposium participants on Teams Platform

14:30 – 14:55 **Hypomyelination leukodystrophy 14 (gene UFM1) – frequent neurodegenerative disorder in Slovak Roma population**
Dr. Mária Giertlová MD, PhD
Department of Clinical Neurosciences
Center of Clinical and Preclinical Research MEDIPARK and Department of Neurology, Faculty of Medicine, P. J. Šafárik, Košice, Slovakia

Department of Medical Genetics, Children's University Hospital, Banská Bystrica, Slovakia

15:00 – 15:15 Lunch break

Session III – Challenges in neurodevelopmental disorders
"With each challenge, we carve our path to triumph." – William Ernest Henley

Session plan: 30-50 min. talks, plus 5-10 min. for discussion
Official language of session: English, Polish

15:15 – 16:05 **Keynote lecture: The genetics and genetic workup of developmental epileptic encephalopathies**
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

16:15 – 17:00 **Keynote lecture: Newborn screening: more than 60 years of saving babies' lives**
Dr. Iveta Sosova PhD, DABMGG, FACMG
Alberta Precision Laboratories Genetics and Genomics Edmonton, Canada

17:05 Joint photo of the Symposium participants on Teams Platform

17:05 – 17:30 **Neurodevelopmental disorders beginning in childhood and adolescence – the perspective of adult patients**
Representatives of patients with alternating hemiplegia of childhood, Dravet syndrome and adrenomyeloneuropathy with their families

Session IV – Mini-Symposium 2026 Sleep Awareness Month for Europe
"Dream big, act small – every action counts." – Robin Sharma

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

17:30 – 18:10 **Orofacial aspects and sleep disorders in rare diseases**
Dr. Stanimira I. Sparreboom-Kalaykova, DDS, MSc, PhD
Department of Dentistry
Radboud University Medical Center, Nijmegen, The Netherlands

18:15 – 18:45 **Sleep apnea in children: A clinical study linking physiopathological changes and sleep apnea risk**
Dr. Rita Castanheira Simões
Hospital CUF Cascais, Clínica CUF S. Domingos Rana, Lisboa, Portugal

Dr. Carlos Sousa, LMD, MClinDent, PGCert, PGDip, FHEA
Clinical Director and CEO of Clínica Ferreira Borges, Lisboa, Portugal
Hospital CUF Cascais, Hospital CUF Sintra, Lisboa, Portugal

18:50 – 19:25 **Sleep, nutrition and screen time - some genetic and neuroscientific aspects**

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

19:25 – 19:30 **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

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 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).

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 10,500 citations (H index = 50).

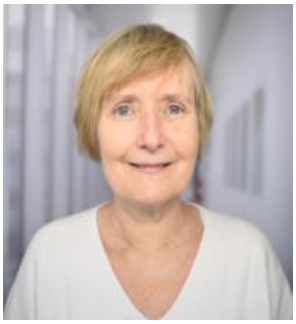
Beata Chałupczyńska, master in biology, is a laboratory diagnostician with specialization in medical molecular genetics, professionally affiliated with the Children's Memorial Health Institute for ten years. Her main area of interest is the diagnostics of neurodevelopmental disorders and epilepsy. She is an active participant in interdisciplinary meetings of the ERN ITHACA and ERN EpiCARE networks at Children's Memorial Health Institute.

Dr. Amaia Lasa Aranzasti is a clinical geneticist in the Clinical Genetics Team at Vall d'Hebron Barcelona Hospital since 2020 in both patient care and research. Her main areas of interest are clinical and diagnostic aspects of genetically determined rare diseases with special interest in prenatal genetic diagnosis and neurogenetic diseases.
Prior training and jobs: *Degree in Medicine: University of Basque Country (2009-15); *Medical internal resident in pediatrics: Donostia University Hospital (Basque Country 2016-20); * University expert in medical genetics: Universidad Rey Juan Carlos Madrid (2018) *Subspecialty in Clinical Genetics: Vall d´Hebron Barcelona Hospital (2020). *Master in Healthcare genetics: Universidad Autónoma de Barcelona UAB (2020-2022) *European Certificate in Medical Genetics and Genomics (2022). * Doctoral Student Universidad Autónoma de Barcelona UAB (2021-).

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.

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 vise president of Turkish Neuromuscular Research Association and the Board-Member of Neuromuscular Research Center of Lokman Hekim University in Ankara and coordinator of the Yalaz developmental screening test and coordinator of the Neurocutaneous disorders study group of Turkish Child Neurology Society.
She has been working as a Professor in Yuksek Ihtisas University, Pediatrics and Pediatric Neurology Departments , Ankara, Türkiye since November 6, 2024. She was National Delegate of AOCNA between 2021-2025 and Advisory Board Member of AOCNA 2025 Congress. She is also Member of Undiagnosed Diseases Network International (UDNI). Prof. Sonmez has published >100 research papers
from the area of Child Neurology which have been cited >2000 times, (H-index is 26 in Researchgate). Her research interests are epilepsy, neurometabolic, neurogenetic, sleep and rare disorders and vitamin D associated neurological disorders.

Prof. Dilek Çavuşoğlu, born in Tarsus, he received his medical training at Ege University Faculty of Medicine 2001-2008 years. In 2008, he worked as a general practitioner in Van/Erciş. Between 2009 and 2013, he worked as a patient alongside patients in the Department of Child Health and Diseases at İzmir Tepecik Training and Research Hospital. In 2013, he worked as a specialist in Child Health and Diseases in Van. Between 2014 and 2017, he received training in Pediatric Neurology at the Department of Child Health and Diseases, İzmir Katip Çelebi University Faculty of Medicine. In 2017, he started working as a Pediatric Neurologist at Afyon Kocatepe University. In 2022, he earned the title of Associate Professor of Pediatric Neurology. Since 2021, he has been serving as the head of the Department of Pediatric Neurology at Afyonkarahisar Health Sciences University Faculty of Medicine.



Dr. Caroline Sevin is a pediatric neurologist based at Hôpital Bicêtre in Le Kremlin-Bicêtre, France, where she works in the Pediatric Neurology Department and the Reference Center for Leukodystrophies. She is affiliated with Université Paris-Saclay and focuses on the diagnosis, management, and research of rare neurological diseases, particularly leukodystrophies and other inherited white matter disorders.
Dr. Sevin's clinical work involves the multidisciplinary care of children with complex neurogenetic conditions. As part of the French Reference Center for Leukodystrophies, she participates in the coordination of care pathways, genetic counseling, and long-term management strategies for affected patients and their families. Her responsibilities include neurological assessments, diagnostic evaluations, and contributions to therapeutic planning.

She is involved in several clinical and translational research projects aimed at improving understanding of the natural history, biomarkers, and potential treatments for leukodystrophies. Dr. Sevin also contributes to national and European research networks focused on rare neurological disorders. Her work includes participation in clinical trials, international registries, and collaborative studies exploring novel therapeutic approaches, including gene and enzyme replacement therapies.
In addition to her clinical and research responsibilities, Dr. Sevin takes part in the education and supervision of medical students, neurology residents, and fellows in pediatric neurology. She also contributes to professional training and public awareness initiatives related to leukodystrophies and pediatric neurometabolic diseases.

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).

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 has more than 320 peer reviewed publications, 420 abstracts 42 chapters one book and two booklets. He also has more than 15,156 citations in the literature with an h-index of 69 and an i-10index of 244 (Google Scholar database Feb 9 2026). 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. Iveta Sosova is a laboratory biochemical geneticist with the Alberta Newborn Screening and Biochemical Genetics Laboratories in Edmonton, Canada, and the head of the Biochemical Genetics Laboratory. She holds board certification from the American Board of Medical Genetics and Genomics in Clinical Biochemical Genetics. She is also an associate clinical professor in the Dept of Lab Medicine and Pathology at the University of Alberta where she contributes to academic teaching and clinical research.

Originally from Slovakia, she earned her doctoral degree in Biochemical Sciences from the National Autonomous University of Mexico (UNAM) in Mexico City in 2002. In 2015, she completed a three-year Canadian College of Medical Geneticists post-graduate fellowship training program in biochemical genetics at the University of Calgary. Since 2015, she has been working as a biochemical geneticist in Edmonton. She is particularly interested in newborn screening, mitochondrial metabolism, and mitochondrial disorders.

Dr. Rita Castanheira Simões is a Portuguese dentist specialized in Orthodontics and Dental Sleep Medicine. She graduated in Dental Medicine from the Instituto Universitário Egas Moniz in 2013 and began developing a focused interest in Dental Sleep Medicine in 2016. In 2017, she expanded her expertise by attending the World Sleep Congress in Prague, further deepening her knowledge in the field of sleep disorders.

In 2018, Dr. Simões completed her specialization in Orthodontics. Driven by a growing interest in pediatric care, she pursued advanced training in Functional Orthopedics of the Jaws at the Wilma Simões European Institute, graduating in 2021. This additional qualification strengthened her clinical focus on early intervention and craniofacial development in children.

In 2024, she began a postgraduate program in Sleep Medicine at the Católica Medical School. That same year, in collaboration with Dr. Carlos Sousa, she initiated a research project entitled "Linking Physiopathological Changes and Sleep Apnea Risk in Children," reflecting her commitment to advancing scientific knowledge in pediatric sleep-disordered breathing.

Dr. Rita Castanheira Simões is a qualified dentist in Dental Sleep Medicine, certified by the SEMDES - Spanish Society of Dental Sleep Medicine and the EADSM - European Academy of Dental Sleep Medicine. Her clinical practice integrates orthodontics, functional jaw orthopedics, and sleep medicine, with a particular dedication to improving pediatric airway health and overall quality of life.

Dr. Carlos Sousa is a dentist and orthodontist based in Portugal with nearly two decades of clinical experience across Portugal and the United Kingdom. He holds a Master's degree in Orthodontics and Dentofacial Orthopaedics from BPP University (London), a Postgraduate Diploma in Clinical Education from King's College London, and a Postgraduate degree in Prosthodontics and Implantology from Egas Moniz. He is currently completing a Postgraduate programme in Sleep Medicine at the Faculty of Medicine, Universidade Católica de Lisboa. His extensive training includes specialised courses in Snoring and Sleep Apnoea Diagnosis and Treatment, Temporomandibular Dysfunction and Orofacial Pain, Functional Orthopaedics of the Jaws (Instituto Wilma Simões, Lisbon), Orthognathic Surgical Orthodontics (Instituto Formedika, Madrid), and a Postgraduate programme in Aligner Orthodontics (CESPU, Oporto). He has completed multiple additional trainings in bone-anchored maxillary expansion, temporary anchorage devices, and orthodontic mini-implant anchorage systems. He is Clinical Director and CEO of Clínica Ferreira Borges and works in private hospitals including Hospital CUF Cascais and Hospital CUF Sintra. A Fellow of the Higher Education Academy (FHEA) and member of the European Orthodontic Society, Dr Sousa combines his broad orthodontic and multidisciplinary training with a dedicated focus on Dental Sleep Medicine, with a particular interest in paediatric airway management and early intervention strategies.

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.