International Symposium Rare Disease Day 2024 - Warmia and Mazury, Olsztyn

„We are the 300 million – we are all fighters!"

Ladies and gentlemen,

on behalf of the Organizers, I have the pleasure to invite you to participate in the International Symposium Rare Diseases Day 2024 *„We are the 300 million – we are all fighters!"* 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 rare neurodevelopmental disorders manifested by paroxysmal events. Among the invited speakers there are representatives of centers associated with three European Reference Networks - ERN-EpiCare, ERN-ITHACA and ERN-RND. We will also host representatives of patients with alternating hemiplegia of childhood and their families.

All interested persons are cordially invited to participate in the International Symposium Rare Diseases Day 2024 *„We are the 300 million – we are all fighters!"*.

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: **24 February 2024, 8:00 a.m. – 6:10 p.m. CET**

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

Application: **via e-mail to chorobyrzadkiewssd@gmail.com by 23 February 2024 (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 2024 - 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 - when diagnosis and follow-up goes beyond the clinical exome

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

9:00 – 9:45 **Keynote lecture: Epileptic and developmental encephalopathies**
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

10:00 – 10:20 **Keynote lecture: New neurodevelopmental disorders**
Prof. Rafał Płoski MD, PhD
Head, Department of Medical Genetics
Warsaw Medical University, Warsaw, Poland

10:30 – 11:10 **Keynote lecture: Alternating hemiplegia of childhood – (Natural) history and future perspectives**
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

11:25 – 11:55 **Keynote lecture: Non-stationary outcome of alternating hemiplegia of childhood into adulthood**
Dr. Marco Perulli MD, PhD
Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy

5 min. Joint photo of the Symposium participants on Teams Platform, short break

Session II – Paroxysmal events in rare neurodevelopmental diseases – when clinic meets genetics

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

12:00 – 12:45 **Vitamin-dependent encephalopathies with case presentations**
Prof. F. Müjgan Sönmez MD, PhD
Retired Lecturer, Department of Child Neurology, KTU Medical Faculty, Ankara, Turkey
Vice-President of Neuromuscular Research Association, Ankara, Turkey
Board of Neuromuscular Research Center of Lokman Hekim University, Ankara, Turkey

12:50 – 13:20 **Periodic diseases and their triggering relationships with each other**
Prof. Serdar Ceylaner MD, PhD
Intergen Genetic and Rare Diseases Diagnosis and Research Center, Ankara, Turkey
Lokman Hekim University, Department of Medical Genetics, Ankara, Turkey

13:25 – 13:45 **Genetic heterogeneity of epilepsies or "epilepsies beyond the NGS panel"**
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

13:50 – 14:10 **Data reanalisis in epileptic encephalopathies – collection of cases**
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

10 min. Short refresh break

Session III – Developmental brain malformations - a lesson that needs to be learned

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

14:20 – 14:50 **Neuroimaging and exome sequencing applied to fetal neurology**
Dr. Nerea Maiz Elizaran MD, PhD
Research coordinator, Maternal Fetal Medicine Specialist, Obstetrics Department
Hospital Universitari Vall d'Hebron, Barcelona, Spain

14:55 – 15:15 **CASP2-related intellectual disability and lissencephaly – case presentation**
Eyyüp Üçtepe MD
Acıbadem Labgen Genetic Diagnosis Center, Ankara, Turkey

15:20 – 15:40 **The wide and growing range of laminopathies – Polish perspective**
Dr. Agnieszka Madej-Pilarczyk MD, PhD
Head, Department of Medical Genetics
Children's Memorial Health Institute, Warsaw – Member of ERN-ITHACA, Poland

15:40 – 16:00 **Genetics of human brain development – time for new classification of neurodevelopmental disorders?**
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

15 min. Short refresh break

Session IV – Alternating hemiplegia of childhood - underestimated paroxysmal disease of childhood

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

16:15 – 16:40 **Alternating hemiplegia of childhood – the clinical landscape**
Dr. Francesco Fortunato MD, PhD
Institute of Neurology, Magna Graecia University, Catanzaro, Italy

16:45 – 17:15 **Alternating hemiplegia of childhood – update and Polish perspective**
Dr. Aleksandra Gergont MD, PhD
Department of Pediatric and Adolescent Neurology, Jagiellonian University, Cracow, Poland
Department of Pediatric Neurology, University Children's Hospital, Cracow, Poland

17:15 – 17:40 **Alternating hemiplegia of childhood – perspective of patients and patients' families**
Representatives of the Polish Association for People with AHC

17:45 – 18:00 **Disorders mimicking alternating hemiplegia of childhood – challenging differential diagnosis**
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

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

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).

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).

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. 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.

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, Turkey. 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, Turkey 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 Turkey 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 has been retired lecturer since 2016. She gives lectures as honorable lecturer in different universities in Ankara and Istanbul. She is vice 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. Prof. Sonmez has published > 100 research papers from the area of Child Neurology

which have been cited > 1800 times, (H-index is 36.59 in Researchgate). Her research interests are epilepsy, neurometabolic, neurogenetic, sleep and rare disorders and vitamin D associated neurological disorders.

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. 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. 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-).

Dr. Nerea Maiz Elizaran is a specialist in Obstetrics and Gynecology, experienced in Fetal Medicine. She joined the Fetal Medicine team at Vall d'Hebron University Hospital in Barcelona (Spain) in 2016. She is also Research coordinator of the Obstetrics Department. Since 2020 she is also a Professor at the Universitat de Vic-Universitat Central de Catalunya (UVIC-UCC). Since 2013 she is a member of the board of Ultrasound section of the Spanish Society of Obstetrics and Gynecology (SESEGO).

Prior experience: In 2003 she joined the Institut Universitari Dexeus, where she started her sub-specialization and her passion for Fetal Medicine was awakened. Between 2006 and 2009 she worked as a Research Fellow at Kings College Hospital in London, under the supervision of Professor Nicolaides, which was a turning point in her career. During this period, she sub-specialized in Fetal Medicine and Therapy and trained as a researcher, carrying out the work that led to the defense of her doctoral thesis at the Universitat de Barcelona in 2010. Between 2011 and 2016 she worked at Cruces University Hospital (Biscay), where she set up the Fetal Medicine and Therapy Unit. Since 2006 she has participated as a teacher in multiple training courses, both nationally and internationally in Latin America and Europe. She was an associate professor in the subject of Obstetrics at the University of the Basque Country between 2012 and 2016.

Research: Her research career began in London in 2006, and later she studied a Master's degree in Research Methodology at the Autonomous University of Barcelona from 2009-2013. She is the author or co-author of more than 100 articles in international journals.

Dr. Eyyüp Üçtepe is a physician in Acıbadem Labgen Genetic Diagnosis Center in both patient care and research. His main interest is to move towards early diagnosis and more effective treatment of children with rare diseases that will improve their quality of life and that of their families. He and his colleagues showed for the first time *CASP2* gene and intellectual disability association.

Prior training and jobs: *Degree in Medicine: Ankara University Faculty of Medicine (2004-11); *Residency in Medical Genetics: Turgut Ozal University Faculty of Medicine (Ankara) (2012-16); *Physician Specialist in Medical Genetics: Ankara Dışkapı Yıldırım Beyazıt Education and Research Hospital (2016-17); *Physician Specialist in Medical Genetics:

Ankara Genetic Diagnosis Center (2019-2020). Director in Training in Acıbadem Labmed Ankara Tissue Typing Laboratory (2020-2023); Director in Acıbadem Labmed Ankara Tissue Typing Laboratory (2023); *Physician Specialist in Acıbadem Labgen Genetic Diagnosis Center (2023-24). Areas of research interest: data mining in rare diseases, novel gene-disease association in neurodevelopmental disorders, bioinformatics.

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. 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 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 – 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. 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), Polish Genetic Society (PTG). 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 of the Warmian-Masurian Voivodeship, awarded by patients.

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.