



RARE DISEASES MALTA

Seminar on Rare Diseases and European Reference Networks

Under the distinguished patronage of
His Excellency George Vella, President of Malta
Tuesday 28th February 2023, University Campus, Valletta

Programme

09:00 - 09:30	Registration
09:30 - 09:40	Opening speech by Hon Mr Chris Fearne, Minister for Health
09:40 - 10:00	Introduction by Dr James Clark
10:00 - 10:10	Agenda of the Czech Republic Presidency
10:10 - 10:30	Presentation by Professor Alexis Arzimanoglou
10:30 - 11:00	Coffee break
11:00 - 11:20	Presentation by Professor Holm Graessner
11:20 - 11:40	Presentation by Ms Michelle Battye
11:40 - 11:50	Presentation by a parent of a child with a rare disease
11:50 - 12:10	Presentation by Dr Maryanne Caruana
12:10 - 12:30	Presentation by Ms Daniela Carabott Pawley
12:30 - 13:20	Lunch break
13:20 - 13:40	Presentation by Dr Nicola Dingli
13:40 - 13:50	Video of patient
13:50 - 14:00	Contribution by the Chief Medical Officer, Mr Walter Busuttil



Speaker Biographies

James Clark

Dr James Clark graduated Doctor of Medicine and Surgery in 1995 and specialised in General Medicine and Gastroenterology. He also read for a Masters degree in Patient Safety and Quality Improvement at Imperial College London. Dr Clark is the ERN representative for Mater Dei Hospital which is Malta's National Coordination Hub for all 24 European Reference Networks.

Alexis Arzimanoglou

Professor Alexis Arzimanoglou is the Coordinator of the European Reference Network for Rare and Complex Epilepsies ERN EpiCARE, currently also chairing the 24 ERN Coordinators Group. He is a Child Neurologist, Director of the Paediatric Clinical Epileptology, Sleep Disorders and Functional Neurology Department, University Hospitals of Lyon, France and Scientific Coordinator of the Epilepsy Unit at San Juan de Déu Hospital, Universitat de Barcelona, Spain. Elected member of the current ILAE-Europe Commission, Editor-in-Chief Emeritus of the ILAE journal Epileptic Disorders and Associate Editor of the European Journal of Paediatric Neurology. He received the EPNS Aicardi Award for Excellence in Paediatric Neurology, the ILAE Ambassador award and the European Epilepsy Education award and is an Honorary member of the Academia Iberoamericana de Neurologia Pédiatrica.

Holm Graessner

Professor Holm Graessner has graduated in Biomedical Engineering, Electrical Engineering and Cybernetics, German Language and Literature, Philosophy as well as Business Administration. He received his PhD "Summa cum laude" in 2004 and, then, he obtained his MBA degree in 2008.

He has been Managing Director of the Rare Disease Centre, since 2010, at the University and University Hospital Tübingen, Germany. www.zse-tuebingen.de He is Coordinator of the European Reference Network for Rare Neurological Diseases (ERN-RND). www.ern-rnd.eu. Together with Olaf Riess, he coordinates the H2020 Solve-RD project on "Solving the unsolved rare diseases". www.solve-rd.eu. In 2021, together with colleagues from Tübingen, Heidelberg and Leiden, HG founded the 1 Mutation 1 Medicine initiative (1M1M) that develops mutation-specific therapies for patients with severely debilitating or life-threatening (SDLT) rare neurological diseases.

Michelle Battye

Ms Michelle Battye is responsible for all operational aspects of the network, reporting to the HCP Network Coordinator Representative, Prof. Wout Feitz. Her main focus is on ensuring the network is active, working together as a team, and that it is delivering. She oversees ERN eUROGEN's coordination and governance, dissemination and collaboration, evaluation and monitoring, virtual consultations, registry and other digital activities, and training and education. She is responsible for drafting multiple European Commission (EC) grant proposals to secure ERN funding, managing and reporting on the grants, and fostering relationships with the EC. She also helps out with the EC ERN Monitoring Working Group (which ERN eUROGEN leads for all ERNs).

Ms Battye has a background in European project management, European policy coordination, European partnerships, conference management, and training, and has worked for the European Commission, UK Government, and UK National Health Service (NHS).

Maryanne Caruana

Dr. Maryanne Caruana is a consultant cardiologist at Mater Dei Hospital in Malta, where she runs the Adult Congenital Heart Disease (ACHD) and Aortopathy services. She obtained an MD from the University of Malta in 2002, and, after completing her basic medical and higher specialist cardiology training in Malta, she furthered her training in ACHD in tertiary centres in London and Bristol in the UK. In November 2018, she was conferred a Ph.D. by the University of Malta following her studies on the adult congenital heart disease population in Malta. She has been a focal point for the Heritable Thoracic Aortic Disease Working Group within VASCERN in Mater Dei Hospital since 2020. She is also the current president of Beating Hearts Malta, a voluntary organisation for adults and children with congenital heart disease.

Daniela Carabott Pawley

Ms Daniela Carabott Pawley graduated as a physiotherapist in 2013 from the University of Malta. In 2022 she obtained a M.Sc in Physiotherapy from the same university. She has joined the public sector in 2013, and continues to further her professional development in Paediatric rehabilitation and acute care. Her other main area of interest, is the role of physiotherapy in the management of non-communicable diseases across all ages. She has presented her work in this area, during the 4th European congress of the ER-WCPT held in Liverpool (2016). This is her first ERN exchange. She has previously participated in the Leonardo da Vinci programme for vocational and education training (Saint Luc, Belgium 2014) and also had the opportunity to join the Traditional Chinese Medicine Summer school program (Shanghai, China 2018). Besides her professional career Daniela is also an avid lover of performing arts and is a freelance stage actress.

Nicola Dingli

Dr Nicola Dingli graduated as a doctor in medicine and surgery from the University of Malta in 2005. She subsequently went on to specialise in adult neurology and clinical neurophysiology, reading for a Masters degree at University College London. Dr Dingli works as a consultant neurologist at Mater Dei Hospital and has a special interest in movement disorders. She is also actively involved in teaching, holding teaching positions at University of Malta, Queen Mary University, Malta Campus as well as MCAST.