Towards a Rare Brain Disease Ecosystem

Programme & Speakers

29 February 2024 | 09:30-14:00

University Foundation, Brussels & Virtual

Towards a Rare Brain Disease Ecosystem

Day: 29th February 2024 – Rare Disease Day

Duration: 09.30 - 14.00

Location: Francqui Room, University Foundation, Rue d'Egmont 11, 1000 Brussels, Belgium

Aim

The main purpose of Rare Disease Day is to raise awareness about rare diseases and their impact on the lives of those affected. In the continuity of the European Brain Council's (EBC) activities on rare brain diseases, EBC introduced the concept of a Rare Brain Disease Ecosystem to engage key stakeholders in the field to identify gaps, priorities, and projects in Europe. During this event, researchers, clinicians, persons living with brain disease and industry will share their perspectives and priorities in the rare brain disease research, care, and policy space, and will brainstorm about highly needed projects to be implemented.

Agenda

9.30 - 10.00	**Registration & Welcome Coffee**
Session 1:	**Welcome and Opening: The Need for a Rare Brain Disease Ecosystem**
10.00 - 10.05	**Welcome**
	Frédéric Destrebecq, European Brain Council
10.05 - 10.10	**Opening Address (video message)**
	MEP Stelios Kympouropoulos (GR, EPP)
10:10 - 10:25	**The MOONSHOT initiative**
	Magda Chlebus, European Federation of Pharmaceutical Industries and Associations (EFPIA)
Session 2:	**Patient and caregiver priorities**
10.25 - 10.35	**A personal testimony**
	Tim Buckinx, Epihunter

10:35 - 10:45	**Mental health in people living with a rare disease**
	Matt Bolz-Johnson, EURORDIS – Rare Diseases Europe
10.45 - 10.55	**The rare neurology charter**
	Orla Galvin, European Federation of Neurological Associations (EFNA)
Session 3:	**Industry innovation trends**
10.55 - 11.05	**Neuromyelitis Optica spectrum disorder (NMOSD) and Myasthenia Gravis (gMG)**
	Matthias Heck, Alexion Pharmaceuticals
11.05 - 11.15	**Amyotrophic Lateral Sclerosis, a rare neurodegenerative disease**
	Juan José Fernández Romero, Amylyx Pharmaceuticals
11.15 - 11.30	**Coffee Break**
Session 4:	**Clinical care pathways for rare disease in Europe: Panel Discussion**
11.30 - 12.10	*Moderator: Vinciane Quoidbach, European Brain Council*
	Paola Giunti, Ataxia UK: The case of ataxia
	Anita MacDonald, Birmingham Children's Hospital (UK): The case of phenylketonuria
	Giuseppe Turchetti, Scuola Superiore Sant'Anna (IT): Towards a digital care pathway?
Session 5:	**Unmet needs: Open Discussion**
12.10 - 12.50	*Moderator: Frédéric Destrebecq, European Brain Council*
	Anna Jansen, European Paediatric Neurology Society (EPNS)
	Michelangelo Mancuso, European Academy of Neurology (EAN)
	Interaction with the Audience: Q&A
12.50 - 13.00	**Wrap up and closing remarks**
	Frédéric Destrebecq, European Brain Council
13.00 - 14.00	**Networking Lunch**

Meet the speakers

Frédéric Destrebecq, Executive Director, European Brain Council

Frédéric Destrebecq is the Executive Director of the European Brain Council since October 2014. In this capacity, he is responsible for providing strategic direction and leadership while managing the day to day operations of EBC and its ongoing relationships with its member associations and other stakeholders, as well as representing the organisation in various European and national forums.

Fred holds a Master Degree in Political Science and International Relations from the Université Catholique de Louvain (Belgium). He also studied at the Institut d'Etudes Politiques (Paris) and University of Wales College (Cardiff), in the framework of the former EU Socrates exchange programme. Prior to EBC, Fred served the European Union of Medical Specialists (UEMS) as Chief Executive Officer, and previously as Director for European Affairs.

Dr. MEP Stelios Kympouropoulos (EPP, GR)

Dr. MEP Stelios Kympouropoulos is a Greek psychiatrist and politician and has been serving as a Member of the European Parliament since 2019. He joined the European People's Party. He is a member of the Committee on Employment and Social Affairs (EMPL), Committee on Petitions (PETI). He is also a Member of the newly established Subcommittee on Public Health (SANT). In addition, he is the EPP coordinator in COVI Committee; a substitute member of the ENVI Committee; REGI Committee and the Subcommittee on Human Right; and the co-chair of the Disability Intergroup of the European Parliament. He holds a master in "Mental Health Promotion - Prevention of Psychiatric Disorders" and a BA in Medicine from the Department of Medicine of the National and Kapodistrian University of Athens. In 2013, he pioneered the organization of the first Greek delegation to the pan-European event "Freedom Drive", which became the springboard for the creation of the first Independent Living Organization of Greece, "I-Living!", which is an urban non-profit company run exclusively by individuals with a disability.

Magda Chlebus, Executive Director Scientific & Regulatory Affairs, European Federation of Pharmaceutical Industries and Associations (EFPIA)

Magda Chlebus is Executive Director Scientific & Regulatory Affairs at the European Federation of Pharmaceutical Industries and Associations (EFPIA), representing the R&D-based pharmaceutical industry in Europe.

Magda and her team are in charge of following policy and legislative developments that influence the research and regulatory environments for the healthcare industry in Europe. This includes public private collaborations (inter alia the Innovative Medicines Initiative and Innovative Health Initiative), enabling and sensitive technologies and the interface between new science and technology and regulation.

She joined EFPIA in 1995. Her experience covers public and government affairs mainly at EU level, on a range of legislative and non-legislative files in the area of research, development and access to medicines and enabling technologies.

Magda, a Polish national, holds a Master Degree in Applied Linguistics from the University of Warsaw.

Tim Buckinx, Founder & CEO, Epihunter

With more than a decade steering global digital initiatives at Bose, Tim Buckinx is no stranger to the power of technology. But it's his personal journey as a father to a son living with refractory epilepsy that truly ignited his passion. Driven by this deep personal connection, Tim founded epihunter, a trailblazing digital therapeutics company aimed at transforming lives.

His vision? To leverage the untapped potential of digital technology in diminishing the everyday challenges posed by neurological disorders. Imagine glasses, which don't cure bad vision but drastically reduce its impact, enabling people to engage fully with the world. Now, what if digital solutions could do the same magic for those grappling with brain disorders? Tim Buckinx believes they can—and he's on a mission to prove it.

Matt Bolz-Johnson, Patient advocate, EURORDIS

Matt Bolz-Johnson has worked at EURORDIS since 2014 as a patient advocate. Matt has over 15 years experience working in the rare diseases field specialising in the selection and approval of expert centres and development of clinical networks in England as well as supporting the development of the European Reference Networks in the European Union.

Matt has also been a lead and researcher on advance therapies in EURORDIS Rare Impact initiative. This patient-led initiative assessed the challenges and proposed actionable solutions to improve patient access to advanced therapy medicinal products (ATMPs) in Europe.

Previously, Matt worked in NHS, England as a hospital manager and commissioner and specialised in rare disease and highly specialised healthcare.

Orla Gavin, Executive Director, European Federation of Neurological Associations

Dr. Orla Galvin came to patient advocacy with a PhD in Medicine and background in drug discovery and design in both academic and industry environments. Transitioning to patient advocacy work as Director of Research and Policy, Orla led high impact, multi-stakeholder socio-economic studies, patient preference studies, and accessibility studies across the globe assessing both rare and common conditions.

Orla is an internationally invited speaker to EU Parliament, WHO-forums, research and clinical learned societies, patient organisations, and industries on topics such as:

- Patient and public involvement in advocacy, research and policy,
- Research in advocacy and policy/evidence-based advocacy,
- Education in advocacy,
- Generation of real-world data,
- Patient reported outcomes, and
- Health economics.

Matthias Heck, Attorney-at-law, Head of EU Government Affairs & Strategic Alliance Development, Alexion Astra Zeneca Rare Disease

Based in Brussels, Matthias heads up Alexion's EU Government Affairs and Strategic Alliance Development. Matthias is blessed to be working with the rare disease community as part of this role, particularly in therapy areas of Hematology, Nephrology, and Neurology. Against the backdrop of the revision of the EU pharmaceutical legislation, Matthias leverages his practical experience in funding negotiations across diverse EU markets, including AMNOG, as well as from leading engagement on framework conditions from a policy perspective at national and European levels. Next to company experience, Matthias' has served as Head of Brussels office of the German Pharmaceutical Industry Association as well as Legal Counsel of the European Confederation of Pharmaceutical Entrepreneurs (EUCOPE). He regularly writes and speaks on life sciences topics.

Juan Jose Fernandez Romero, Lead of Government Affairs - EMEA, Amylyx Pharmaceuticals

I am the Lead of Government Affairs - EMEA at Amylyx Pharmaceuticals. I am contributing to Amylyx's mission of exploring the potential of its therapies and advancing our pipeline in an effort to support our mission of one day ending the suffering caused by neurodegenerative diseases. More specifically, I am working closely with the community of people who live with amyotrophic lateral sclerosis (ALS). I have a background in European policies and public affairs, having worked in patient organisations, medicines regulators, and the Spanish diplomatic corps.

Vinciance Quoidbach, Research Project Manager, European Brain Council

Vinciane Quoidbach is Research Project Manager – Public Health and Policy for the Value of Treatment for Brain Disorders Study at the European Brain Council.

Vinciane holds a Master Degree in Hospital and Healthcare Management from Solvay Brussels School of Economics and Management and a Postgraduate Diploma in Public Health Management from Manchester Business School. She also holds a Master Degree in Political Sciences and European Studies from University of Antwerp. Prior to this position, Vinciane worked in both public and private sector, at national and international level. She served the Belgian Federal Public Service Health, and the Federal Minister of Health Cabinet as Advisor in charge of public health policy and regulations development in the area of Cancer, Chronic Diseases and Rare Diseases as well as the Biopharmaceutical Company AbbVie as Health Initiatives Manager. She also worked for the World Health Organization in Geneva, Swizterland as Technical Officer, Non communicable diseases and mental health cluster and the United Nations Development Programme in Pretoria, South Africa as Technical Officer, Social development department).

Vinciane is also a staff member at the Belgian Centre for Evidence-Based Medicine (CEBAM) at KULeuven.

Paola Giunti, Professor, University College London

Paola is Professor at UCL and Head at the Ataxia Centre at the Department of Clinical and Movement Neurosciences, UCL Queen Square Institute of Neurology. She studied Medicine specializing in Neurology and gained her PhD at the University of Rome, "Sapienza", in Italy. During 1992-1995 she was a Research Fellow at the Institute of Neurology UCL, Queen Square in Department of Clinical Neurology, working in the team of Prof Anita Harding. In 2004 she was successful in gaining a Fellowship from Ataxia UK and was made a Senior Clinical Associate in the Department of Clinical Neurology at Queen Square. In 2005 the Ataxia Centre, a model of translational research, she established was awarded a Centre of Excellence by Ataxia UK. This award, the first of this kind, was instrumental to open up other three centres in the UK based on her model. The worldwide recognition of her successes made the National Ataxia Foundation in the USA to award her centre as an Ataxia Centre Excellence one of few outside the USA. She authored more than 200 high Impact factor scientific papers (Nature Medicine, The Lancet, The Lancet of Neurology, and others). She serves on several National and International Steering Committees. In 2016 she was endowed with the Health Professional Award (Neurology Advocacy Awards) by the European Federation of Neurological Associations (EFNA). In 2023 she was presented with the brand-new Ataxia UK Excellence in Ataxia Research and Care Award.

Anita MacDonald, Consultant Dietitian in Inherited Metabolic Disorders, Birmingham Children's Hospital

Dr Anita MacDonald OBE is Consultant Dietitian in Inherited Metabolic Disorders at Birmingham Children's Hospital, and an Honorary Professor in Dietetics at Plymouth University, UK. Although she semi-retired 8 years ago, she is even more involved in PKU work, concentrating solely on this group as well as doing some voluntary work for the National Society for PKU (NSPKU).

Her involvement in inherited metabolic disorders (IMD) has spanned almost all her working life (45 years).

Dr MacDonald obtained her PhD in phenylketonuria (PKU) in 1999. She has directly cared for over 400 patients with PKU. She has always been actively involved in PKU research, supervises PhD students, Master students and lectures worldwide on PKU. She has around 450 publications – many are research publications on PKU.

She is a member of the European PKU Guidelines group (which is trying to standardise PKU care across Europe), is a member of ESPKU Scientific Advisory Committee, and member of the UK NSPKU Medical Advisory Panel.

The retirement slippers remain well and truly in their box!

Giuseppe Turchetti, Co-Founder of the Institute of Management of the Scuola Superiore Sant'Anna (SSSA - Pisa)

Prof. Giuseppe Turchetti received his Laurea Degree in Economics from the University of Pisa. He received his PhD in Management from the Scuola Superiore Sant'Anna in Pisa, where he is professor of Economics and Management of Innovation and Healthcare. Fulbright Scholar, he spent several years for research in USA as Visiting Scholar at The Wharton School of the University of Pennsylvania (Philadelphia), at the Illinois Institute of Technology (Chicago) and at the Kellogg School of Management of the Northwestern University (Chicago). He is Co-Founder of the Institute of Management of the Scuola Superiore Sant'Anna (SSSA - Pisa), of the Research Center on Technologies and Services for the Support of Longevity (SSSA - Pisa), of the Research Center on European Transplantation Management (SSSA - Pisa), of the Center of Excellence ENDOCAS (on Computer Assisted Surgery) of the University of Pisa. His main research interests are in the fields of the: organisation, financing and evaluation of healthcare services and health technologies, of the management of innovation, organization and commercialization of medical technologies (pharma, medical devices, e-health), strategic management and marketing in the healthcare sector. His research addresses many therapeutic areas and the wide field of rare diseases. He is working on/coordinating several national and international projects in the area of healthcare technologies and management. He collaborates in several projects within the ERN Program (on a regular basis with ERN ReCONNET), European Commission. He is author/editor of twenty books and of three hundred scholarly papers and book chapters..

Anna Jansen, Paediatric Neurologist, Antwerp University Hospital

Prof Anna Jansen is a paediatric neurologist at Antwerp University Hospital. Her clinical work focuses on rare neurogenetic disorders including tuberous sclerosis complex (TSC) and brain malformations. She runs a multidisciplinary TSC clinic and has participated in several clinical trials in TSC. Her clinical work is complemented by teaching in paediatric neurology and preventive medicine. She is involved in translational research on the genetics of brain development and in multi-stakeholder participatory research that focuses on empowering families living TSC. Together with Prof. Petrus de Vries she led the TANDem project, an international participatory mobile-health project to empower families and individuals who live with TSC around the world.

Prof Jansen is affiliated to the Translational Neurosciences Research Group at the University of Antwerp and to the Neurogenetics and Mental Health and Wellbeing Research Groups at Vrije Universiteit Brussel. Her work is supported by a Senior Clinical Investigator Fellowship from the Research Foundation Flanders (FWO). She heads the Advocacy and Collaboration Committee of the European Paediatric Neurology Society and serves as a scientific advisor to be-TSC, the Belgian TSC advocacy association.

Michelangelo Mancuso, Co-chair of the EAN Coordinating Panel on Rare Neurological Diseases, European Academy of Neurology

Professor Mancuso is the head of the Centre of Neurogenetics and expertise for mitochondrial diseases and rare diseases at the Neurological Clinic of the University Hospital of Pisa (Orphanet Center EUGTIT247621).

Scientific and research activity of Professor Michelangelo Mancuso has mainly been conducted in the field of mitochondrial, neurogenetics and stroke. As a whole result, he published more than 300 full papers on peer-rewieved Life Science/Current Contents cited scientific journals. According to Scopus, Dr Mancuso's articles have more than 18000 citations, and the H-Index is 54.

Mancuso is the co-chair of the EAN Coordinating Panel on Rare Neurological Diseases, Past-Coordinator of the Neurogenetics Group of the Italian Society of Neurology and past-chair of the Neurogenetics Panel of the European Academy of Neurology. Recently, Prof Mancuso has been nominated co-chair of the mitochondrial working group of the ERN NMD, and chair of the inter-ERN mitochondrial working group.