European Reference Network
for rare or low prevalence complex diseases
Network Neuromuscular Diseases (ERN EURO-NMD)

EURO-NMD | Building bridges and breaking barriers in rare neuromuscular diseases

EURO-NMD PATIENT REPRESENTATIVES

BOOKLET

OCTOBER 2023

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1. PATIENT REPRESENTATION AND PATIENT ADVISORY BOARD (PAB)

The **Patient Advisory Board** (PAB) aims to ensure true and equitable representation of the voice of patients within the EURO-NMD network so that EURO-NMD services can answer to the needs and expectations of rare neuromuscular disease patients and improve access to high quality diagnosis, care and treatment.

The PAB creates a bridge between the ERN and the rare neuromuscular patient community, by coordinating the participation of all patient representatives involved in EURO-NMD and liaising with its affiliated patient organisations. The Patient Advisory Board also nominates additional patient representatives to be part of each Specialist Group based on their expertise.

The PAB has established the Constitution and Rules of Procedure governing patient participation in EURO-NMD.

Membership of the Patient Advisory Board:

- Chair : François Lamy (AFM-Téléthon, France),
- Dimitrios Athanasiou (MDA-Hellas, World Duchenne Organization, Greece),
- Madelon Kroneman (Spierziekten Nederland, Dutch Patient Society of Neuromuscular Diseases),
- Massimo Marra (CIDP Italia ONLUS),
- Patricia Ann Melsom (Neuromuscular Disorders Association of Norway)
- Marisol Montolio (Duchenne Parent Project Spain),
- Michela Onali (Gli Equilibristi HIBM, Italy),
- Jean-Philippe Plançon (French Association against Peripheral Neuropathies, France - European Patient Organisation for Dysimmune and Inflammatory Neuropathies, EU),
- Evy Reviers (ALS Liga Belgium),

The members of the PAB commit to assist in the following:

- Governance of the ERN: The EURO-NMD Board includes the patient representatives that are part of the Patient Advisory Board. At least one representative of the PAB is also part of the Executive Committee (see *Board Terms of Reference*).

- Activity Groups and Advisory Boards: The PAB nominates additional patient representatives to be part of each Specialist Group based on their expertise.

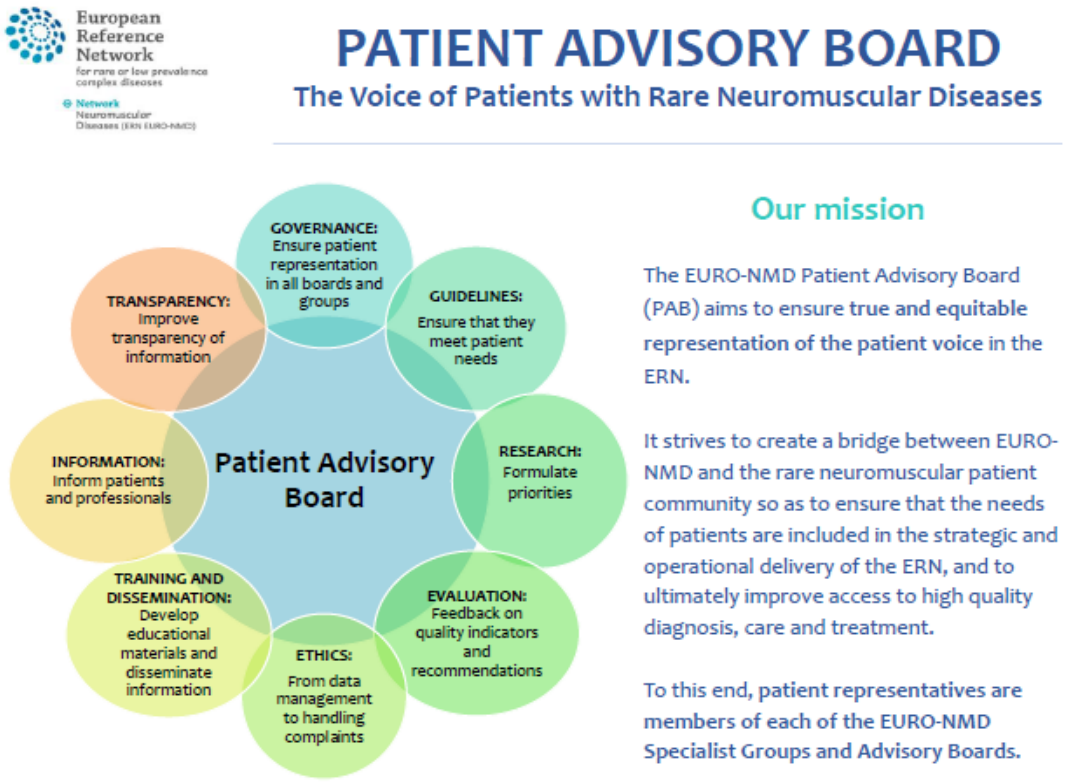
- Guideline development: PAB members are central to the development of clinical and specialised social services guidelines and outcome measures, ensuring these instruments meet patient needs. For this, they engage with patient representatives beyond EURO-NMD to appoint the best person for each task.

- Ethics: PAB members are involved in ethics activities.

- Research: PAB members contribute to the definition of research priority areas based on what is important to patients and families and will ensure that patients are embedded in the research activities of the network.

- Evaluation: PAB members play a key role in the evaluation of the network, ensuring feedback based on patient experience and evaluating ERN performance by reviewing quality indicators and making recommendations.

- Training and dissemination: PAB members will commit to disseminate information about the network and its initiatives to their organisations and to other patient groups not currently part of the PAB. They will develop educational materials for patients and families and healthcare professionals and participate in teaching activities.



2. EURO-NMD PAB MEMBERS

FRANÇOIS LAMY

Chair of the Patient Advisory Board
Member of the Executive Committee
Member of the Muscle Diseases Specialist Group
Member of the Genetics Specialist Group

François Lamy is the father of a 15-year-old boy with Duchenne Muscular Dystrophy. He has been a member of the Board of Directors of the AFM-Téléthon since 2012, and currently serves as its Vice-President in charge of the Téléthon. François Lamy also serves on the Board of Directors of the Centre for the Study of Stem Cells (I-Stem CECS).

DIMITRIOS ATHANASIOU

Member of the Patient Advisory Board
Member of the Educational Board

When his son was diagnosed with Duchenne Muscular Dystrophy, Dimitrios Athanasiou became a fulltime international patient advocate in Duchenne and in rare diseases. Dimitrios Athanasiou initiated the Duchenne Patent Group and served as BoD consultant in MDA HELLAS in Greece, he was also a former BoD of EPF and PDCO EMA Committee Member. Currently, he is a Board Member of the World Duchenne Organization (WDO), a European Medicines Agency Patient Expert in Duchenne Muscular Dystrophy, and the Chair of Rare Diseases Greece He is also a member of EURORDIS, a EUPATI fellow, and a member of EUPATI's Course Committee. He established the Greek EUPATI National Liaison Team.

MADELON KRONEMAN

Member of the Patient Advisory Board
Member of the Muscle Diseases Specialist Group

Madelon Kroneman was diagnosed with Myofibrilar Myopathy (Desminopathy). Her disease started at the age of 33, almost 30 years ago. She is presently a member of the diagnosis working group "Muscular dystrophies and distal myopathies" of Vereniging Spierziekten Nederland, a Dutch neuromuscular patient organisation. She chaired this working group between 2002 and 2016. She is involved in the scientific developments for muscle related diseases and is involved in reviewing scientific research concerning muscular diseases for research requiring the involvement of members of the patient association. She is member of the INPIRE-NMD group, that promotes social and psychological support for patients with neuromuscular

diseases. She participated in several ENMC workshops. She completed the EUPATI training for patient representatives. Professionally, she is involved in health systems research.

MASSIMO MARRA

Member of the Patient Advisory Board
Member of the Peripheral Nerve Diseases Specialist Group

Massimo Marra suffers from rare peripheral neuropathy disease. He founded the Italian Association against Dysimmune Neuropathies in 2012 because there were problems with access to treatment in Italy. He promotes several information and awareness campaigns, awards and grants for young researchers and research on quality of life. He is interested in patient self-monitoring projects, wearable technology and registry. He believes that technology allows people to be informed, and that being informed is essential to exercise one's rights. Technology enables rights.

PATRICIA ANN MELSOM

Member of the Patient Advisory Board

Patricia was diagnosed with Central Core Myopathy shortly after the birth of her daughter, who also has the same disorder. Patricia also has a son and a grandchild with Central Core Myopathy. She is at present president, chairperson of the board and founding member of The Neuromuscular Disorders Association of Norway (Foreningen for muskelsyke - FFM). She is in addition a member of the reference group of The National Advisory Unit on Rare Disorders (Nasjonal kompetansetjeneste for sjeldne diagnoser -NKSD) and member of the national ethics committee for clinical trials for medicines and medical equipment (Komiteen for klinisk utprøving av legemidler og medisinsk utstyr REK KULMU A). Patricia is a retired journalist and editor specialising in medical science, health policy and ethics.

MARISOL MONTOLIO

Member of the Patient Advisory Board
Member of the Muscle Diseases Specialist Group
Member of the Research Board

With more than 20 years of research experience, Marisol Montolio is the Director of the Research Department and Technology Department of the Duchenne Parent Project Spain and Curator of the Patient Registry, promoting scientific research in Duchenne and Becker Muscular Dystrophy. Marisol Montolio is a Biologist and has a PhD in Neuroscience from the University of Barcelona. She has worked in the identification of drugs for neurological diseases, including genetic and

degenerative diseases, and later acquired extensive experience in coordinating and executing research collaborations between academic groups, private companies, foundations and associations on the research of rare diseases. She is also adjunct professor and the Scientific Director of the Chair UB rare diseases at the University of Barcelona.Dr Marisol Montolio is in the External Scientific Board of the Hospital Sant Joan de Déu, Barcelona and the Chair of the Scientific Directors Committee in World Duchenne Organization.

MICHELA ONALI

Member of the Patient Advisory Board
Member of the Muscle Diseases Group
Member of the Research Board
Member of the Educational Board

Michela Onali's advocacy work for GNE Myopathy began in 2014 while living in Canada, allowing her to develop a broad knowledge and have a direct look into the reality and challenges faced by the RD community not only in Europe but also in US and Canada, from national health policies to R&D and approaches to rare diseases in general. After moving back to Europe, she completed the EURORDIS Summer and Winter Schools, the Nutrimet School on Metabolomics and the EUPATI training course. She is involved in working groups aimed at implementing patient engagement in research development and decision-making. Michela Onali advocates to facilitate dialogue, report on patients' needs, guarantee effective and clear information, and to define research priorities while promoting the fundamental role of Natural History Studies and FAIR data in clinical trial design and therapy development.

JEAN-PHILIPPE PLANÇON

Member of the Patient Advisory Board
Chair of the Educational Board
Member of the Executive Committee
Member of the Peripheral Nerve Diseases Specialist Group

Jean-Philippe Plançon has been living with a rare peripheral neuropathy since 2000. He founded the French Association against Peripheral Neuropathies in 2006 and co-founded the French Alliance of Rare Peripheral Neuropathies Patients Associations. At the European level, he is the chair of EPODIN, the European Patient Organisation for Dysimmune and Inflammatory Neuropathies. Administrator of the Rare Diseases French Alliance, graduated in the fields of rare diseases, education, ethics, public health, sciences and public affairs, he advocates at the national and European level to improve quality of life of patients within several expert working-groups and to health policy makers.

EVY REVIERS

Member of the Patient Advisory Board
Member of the Motor Neuron Diseases Specialist Group

Evy Reviers is the daughter of a patient living with Amyotrophic Lateral Sclerosis (ALS). As Chief Executive Officer since 2006 and Chairwoman since 2021 of ALS Liga Belgium, Evy Reviers performs the general management of the organisation and coordinates the support to Belgian ALS patients. She has obtained several priority procedures to increase the quality of life of ALS patients in Belgium. At the European level, she is the Chairwoman of EUpALS, the European Organisation for Professionals and Patients with ALS, and defends the rights of ALS patients in scientific advice procedures at EMA. She is also a member of TRICALS, Project MinE, the Rare Diseases Organisation Belgium, the Drug Information Association, and EURORDIS. Since December 2017, she is a re-elected member of the Board of Directors of the International Alliance of ALS/MND Associations.

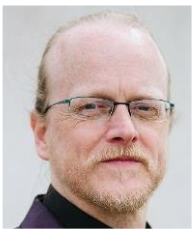
3. OTHER EURO-NMD PATIENT REPRESENTATIVES

ARABELA ACALINEI

Member of the Peripheral Nerve Diseases Working Group

Arabela Acălinei is President of Neuro Move CMT Association in Romania and President of EAMDA - European Alliance of Neuromuscular Disorders Associations. She is also a Board Member of the European CMT Federation and their Eastern Europe Ambassador. Arabela is a patient with Charcot-Marie-Tooth type 1A disease and has 12 more people in her family with the same condition, her 2 sons included. That is the reason she become a passionate patient advocate for people living with CMT and similar neuromuscular conditions. She is a Romanian-English teacher, a translator, and a trainer. She has a degree in Project management, but also in Career Counselling for people with disabilities. She is EURORDIS alumni, EUPATI fellow and a member of EURORDIS HTA Task Force.

PETER ASHLEY



Member of the Muscle Diseases Specialist Group

Peter represents Euro-DyMA, the federation of European Myotonic Dystrophy support groups. Euro-DyMA brings the point of view of the patients and families to European institutions, health professionals and drug developers. It identifies areas where care or research efforts should be intensified to harmonise knowledge in Europe. Peter is the Chair of Cure Myotonic Dystrophy UK (Cure DM), a charity that facilitates research, raises awareness and provides support to those with DM, their families and their careers. He is a member of many panels and groups in the National Health Service and other healthcare organisations and is the vice-chair of the Muscular Dystrophy UK Lay Research Panel. Peter is an Engineer and Management Consultant with over 20 years' experience in business management systems, change management, quality systems and regulatory approvals before his son was diagnosed with congenital DM1 and his focus turned to advocacy and the voluntary sector.

İPEK BADIRGALI

Member of the Muscle Diseases Specialist Group
Member of the Genetics Specialist Group
Member of the Motor Neuron Diseases Specialist Group

İpek Badırgalı received her BSc. in chemical engineering at Ege University, followed by master's degree in marketing at Galatasaray University. She continues her education with a Master at Universita Cattolica, in Patient Advocacy field.
She has worked in managerial positions at the sales, corporate communications, marketing, and business development departments, during her professional life. After her niece Ece was

diagnosed with SMA, she became acquainted with the field of SMA and rare diseases. She worked as Board Member at Turkish Neuromuscular Disorders Association, KASDER. She made patient advocacy at policy making level and pioneered the healthcare system change in SMA disease management in Türkiye. She carries out active roles in the international associations and worked in the PARADIGM project of EURORDIS. She currently works as the secretariat at the European Alliance of Neuromuscular Disorders Associations (EAMDA). İpek Badırgalı is also the founder of the Ecelereumutol platform, which aims to raise awareness in SMA. She is the author of the book Ece'nin Küçük Hayalleri together with her niece Ece.

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MARIA BORRELL

Member of the Research Board
Member of the Muscle Diseases Specialist Group

Maria Borrell has a daughter who was diagnosed with Collagen VI Muscular Dystrophy at the age of 3. She is also a senior investigator in the cardiovascular field in Barcelona. Prior appointments include a postdoctoral position the results of which led to a Phase II clinical trial in French Huntington Disease patients. She has also worked in the identification of drugs for cardiovascular diseases in Germany and in the United States. She is an active member of prestigious national and international societies in cardiology. She has been collaborating with EURORDIS for several years now and has participated in several Meetings and Scope actions to Strengthen Collaborations for Operating Pharmacovigilance in Europe. She was also involved in the PARADIGM Program (Patients Active in Research and Dialogues for an Improved Generation of Medicines) that allows structured patient engagement throughout three key decision-making points of the development of medicinal products: the research priority setting; the design of clinical trials and the early dialogues with regulators from the different European countries. She is an active member of ASEM (Spanish Muscular Association) and Fundació Noelia (Col VI MD association).

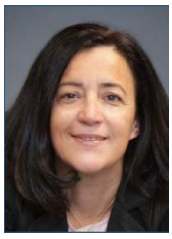
JOAQUIM BRITES

Member of the Muscle Diseases Specialist Group
Member of the Motor Neuron Diseases Specialist Group

Joaquim Brites is the President of APN (Portuguese Neuromuscular Association) since 2012. Born in Leiria, Portugal, he is 62 years old and has one of his children, a boy 31 years old, with Duchenne Muscular Dystrophy. He was also President of the General Assembly of the Portuguese Alliance of Rare Disease Associations between 2012 and 2021. He is a representative of the Portuguese Neuromuscular Patients in several European Organizations. He was a member of the Ad Hoc Committee and of the Installing Committee of the New National Representative Organization of the Rare Disease Associations. He is also a Member of the Patient Advisory Board of the European Patient Advocacy Group (ePAG), of EURORDIS. Member representing Portugal,

of the "Plateforme Maladies Rares", in France. Member of TREAT NMD, in individual name and in representation of APN. Member of the ERN-NMD - European Reference Network for Neuromuscular Diseases (Vice Chair of the Muscle Diseases Group). Member of the Commission for Inclusion Policies for People with Disabilities, of the Ministry of Labor and Social Security. Founding Member of the Platform SIP-PT (Societal Impact of Pain - Portugal). Member of the Mission Structure for the Promotion of Accessibilities, representing motor disability. Member of the Monitoring Committee of the National Strategy for the Inclusion of People with Disabilities. Member of the National Health Council - representing APN.

VICTORIA CASTILLO SANCHEZ



Member of the Genetics Specialist Group

Victoria Castillo is Pablo´s mum. Pablo was born in 2002 with clubbed feet, a dislocated left hip, one extended knee and flexed the other. After a few months, he was diagnosed into the Arthrogryposis group. In 2014 she became part of the Spanish Arthrogryposis Multiplex Congenita (AMC) association, and, in 2019 became delegate for the Catalan delegation of the association. Since 2021, she has been collaborating with an international group of professionals for a patient registry of AMC, and with an international consortium. Looking for alliances with other support groups, they had their first contacts with the Netherlands and France. She is looking for a conservative approach to treatment.

FERNANDA DE ANGELIS

Member of the Muscle Diseases Specialist Group

Fernanda De Angelis is a biologist with a PhD in genetics and molecular biology with more than 10 years of research experience mostly spent on the development of therapeutic strategies for Duchenne and Becker muscular dystrophy. Since 2011, she is a member of the Italian Parent Project Scientific Office where she works on the promotion of scientific research related to DMD/BMD, dissemination of research results to patients and their family and in the management of the Italian DMD/BMD Patient Registry.

INGRID DE GROOT

Member of the Muscle Diseases Specialist Group

Ingrid de Groot (1970, The Netherlands) was diagnosed with dermatomyositis in 2006. She is chair of the Dutch Myositis Working Group for Spierziekten Nederland, the Dutch patient organisation for neuromuscular diseases. She is also involved in several national and international projects regarding either myositis or neuromuscular and

rheumatic diseases in general, like reviewing scientific research for funding bodies, as patient research partner of study groups in myositis (OMERACT, IMACS), member of EULAR (European League against Rheumatism) Study group for Collaborative Research and speaker on patient advocacy (Treat NMD). Ingrid has assisted patients in other countries to set up a myositis working group. She has a particular interest in research and development of outcome measures. She also was recently invited to be the first patient on the Medical Advisory Board of the Myositis Association (world's largest patient association for myositis) and in that position will focus on research and education regarding psychosocial support and exercise and rehabilitation.

EMMA DEL-REY

Member of the Mitochondrial Diseases Specialist Group

Emma Del-Rey is the president of the AMMi association (association against mitochondrial diseases). It is a national organisation and represents French patients. She has been involved in mitochondrial disease since her daughter was diagnosed 16 years ago. She's 19 years old today. She is at IMP board (international mito-patient), in the Genomit working group for the patient database, at Eurordis, rare disease alliance and in contact with the reference centers in France.

MARGUERITE FRICONNEAU

Member of the Neuromuscular Junction Defects Specialist Group

Marguerite Friconneau was diagnosed with Myasthenia Gravis in 1985. She has been elected as a member of the Board of Directors of the AFM-Téléthon since 2007. She has also been a member of the French patient group on Myasthenia Gravis since 1987, and chaired that group from 1994 to 2009. The Myasthenia Gravis patient group is dedicated to patient care and advocacy with the scientific and medical communities as well as public authorities. Marguerite Friconneau was actively involved in the design of the French National Guidance Guide called PNDS in 2015.

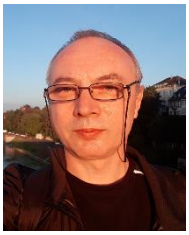
FILIPPO GENOVESE

Member of the Peripheral Nerve Diseases Specialist Group

Filippo Genovese is the vice-president of the European CMT Federation and board member of ACMT-Rete per la malattia di Charcot-Marie-Tooth OdV, an Italian PAG of CMT patients and a founder member of the Federation. He has a scientific background (Ph.D. in Pharmaceutical Sciences, working in the field of Mass Spectrometry/Proteomics) and he lends

a hand to both ACMT-Rete and ECMTF with informatics too (website, social media, communication). He suffers from Charcot-Marie-Tooth 1X, but he does not live the relationship with the disease as a condemnation but as a part of him. It was not easy to metabolize it and it is not easy at certain times when the difficulties related to the disease add up to everything else. He tries to convey this energy and turn it into a stimulus to advocate for a better quality of life for people with a neuromuscular condition.

VITALIY MATYUSHENKO



Member of the Motor Neuron Diseases Specialist Group

Vitaliy Matyushenko is the President of Ukrainian Charitable Foundation "Children with spinal muscular atrophy" since it was founded in 2004. He has a daughter who has been diagnosed with Spinal Muscular Atrophy type II. Vitaliy is a physicist but has almost 20 years in activities related to patient organisations both at the national and international levels. For the past years, he has organized different initiatives promoting the rare disease at national level such as legal initiatives, workshops on standards of care, the National conferences on SMA, national and international meetings, trainings. In 2008 he received membership the TREAT-NMD Global Database Oversight Committee and he plays most valuable role as curator of the Registry for SMA in Ukraine.

ALEXANDRE MÉJAT

Member of the Research Board
Member of the Genetics Specialist Group

Molecular and cellular biologist by training, Alexandre Méjat is also affected by a Bethlem myopathy. He led a research team dedicated to Emery-Dreyfuss muscular dystrophy for 8 years. He was also member of the Board of Directors of AFM-Téléthon and the I-Stem institute. Alexandre Méjat is currently the Chair of the European NeuroMuscular Centre (ENMC) and he joined the Scientific Direction of AFM-Téléthon in 2018, where he currently holds the position of International Scientific Networks Manager.

CAROLINA NAVALON-MARTINEZ

Member of the Education Board
Member of the Research Board
Member of the Muscle Diseases Specialist Group

Carolina Navalon-Martinez was born with a congenital disorder, Arthrogryposis Multiplex Congenita, a rare neuromuscular syndrome. As an adult she was diagnosed with two autoimmune disorders, Axial Spondyloarthritis and Sjögren's syndrome, the latter also a rare disease. She is a Board Member of the Spanish Patients´ Association affected by Arthrogryposis Multiplex

Congenita (AMC) as well as Board Member of the Catalan League for inflammatory rheumatic diseases. She attended the Eurordis Summer School Spanish version on Medicines Research and Development in 2018, becoming a Patient Advocate after completing the course. Main liaison for the Creation of the I Spanish Patient Registry for AMC Arthrogryposis (Paediatric Hospital Sant Joan de Deu Barcelona (Spain); Shriners Hospital Canada; Ciberer, the Center for Biomedical Network Research on Rare Diseases in Spain). Collaboration with Shriners Hospital USA & Canada for AMC Arthrogryposis International Patient Registry: assistance on definition of Common Data Elements, data sharing accessibility, Pre-Delphi (2021-2022) from the patient´s and patient representative perspective. Next objective: the creation of the I European Alliance for Arthrogryposis organised by Patient Representatives.

ALEJANDRA PÉREZ DEL REAL

Member of the Educational Board
Member of the Peripheral Nerve Diseases Specialist Group
Member of the Research Board

Alejandra Pérez del Real has had ITP (a rare haematological disorder) since toddler and developed CIDP (a rare dysimmune inflammatory neuropathy) in 2006. Co-founder and president of GBS|CIDP España- Polineuropatias Inmunomedidas, she is also a member of EPODIN and the GBS|CIDP Foundation International committee, an entity for which she has been a liaison since 2008. An expert patient in several research projects, she is keen on the fields of neuroimmunology and peripheral neuropathies, looking for ways to expand PREMS and PROMS to enhance research and patient empowerment. She is also a member of the Spanish panel for plasma self-sufficiency and the Peripheral Nerve Society.

SILKE SCHLÜTER

Member of the Muscle Diseases Specialist Group

Silke Schlüter (Germany, 1975) was diagnosed with an autoimmune hepatitis and liver cirrhosis in 1990. The diagnosis polymyositis was made in 2012. This was replaced by the diagnosis of overlap-myositis 2015. Silke is the chair of the German Myositis-Group within the Deutsche Gesellschaft für Muskelkranke e.V. - DGM (german society for neuromuscular diseases) and federal board member of this DGM. She is also board member of the German "MYOSITIS NETZ e.V." and the "International Myositis Society – iMyoS" and she is an ePAG at ERN ReCONNET. As a member of the MYOSITIS NETZ she is involved in the development of SOPs (Standard Operating Procedure). She is co-author of several, national and international editorials, guidelines, and textbooks in the field of myositis. She is listed as a patient representative for national and international projects dealing with either myositis or neuromuscular and rheumatic diseases in general (ENMC, IMACS). Silke has a particular interest in disease

management, mental health, and the development of standardized myositis criteria for diagnosis and treatment.

BORYANA STOYANOVA

Member of the Mitochondrial Diseases Specialist Group

Boryana Stoyanova is a mother of a 22-year-old boy who was diagnosed with mitochondrial disease – Kearns-Sayre Syndrome. In December 2019, she became one of the founders of the National Association of Patients with Mitochondrial Diseases in Bulgaria and was elected as Board member of the same. Boryana is involved in different activities of the association like raising awareness about mitochondrial diseases, translating in Bulgarian language and disseminating information materials among the mito community, participation in the worldwide annual initiative Light Up for Mito, registration of 3 mitochondrial diseases in the national register of rare disease etc., She also contributes to the communication with International Mito Patients. In April 2023 she was elected as Board member of Rare Disease Bulgaria, national umbrella organization in the area of rare disease. Boryana is a lawyer with over 20 years of experience in commercial and corporate law as well as compliance specialist.

EVA STUMPE

Member of the Motor Neuron Diseases Specialist Group

Eva is mother of 2 adult kids. Her 28-year-old daughter Sarah is living with SMA II. Eva is a long-time patient contact person within the German DGM e.V., is member of the leadership team of Initiative SMA and is treasurer and board member of SMA Europe e.V. Eva is a trained patient advocate (alumni of the 3rd cohort of the EUPATI Patient Expert Training Course and the EURORDIS Summer and Winter School). On a professional level, she works as a lawyer, supports the family business company and is member of a local housing board.

JAVIER TORRAS

Member of the Muscle Diseases Specialist Group

Javier Torras suffers from Becker Muscular Dystrophy. He was diagnosed when he was 14 years old because his 5 years older brother, with more severe symptoms, was diagnosed. In his case, the symptoms of this disease have been mild and he is lucky to say that, despite starting to have some difficulties, he can still walk fairly well at 51 years old and have an almost normal life. His disease has not impacted much in his life as he has been able to study a university degree, work normally, get married and build a family. He joined ASEM about 15 years

ago to receive information about his disease and be up to date on all investigations and possible treatments. He decided to take a more active role in their activities last year, and becoming a patient representative in the EURO-NMD European Reference Network is a good way to do so.

ISABELA TUDORACHE

Member of the Educational Board
Member of the Muscle Diseases Specialist Group

Isabela Tudorache is the mother of a young Duchenne Muscular Dystrophy man, aged 25. The main objective of her activity was to increase the life expectancy for all DMD/MBD children in her country, through legislative and educational interventions, to overcome the fatalistic attitude towards diseases with limited life span, and changing the mentality and perception towards disability in her country, She is the founder of the Parent Project for Research and Assistance in Muscular Dystrophy in Romania, a Eupati fellow, Project Manager and counselor for people with disabilities. She has PhD in Philosophy and Sociology, and several trainings in psychotherapy and project management. She received membership of WDO, the TREAT-NMD Global Database Oversight Committee, and ANBRaRo.

KATIE WALLER

Member of the Mitochondrial Diseases Specialist Group

Katie Waller is a registered paediatric nurse with significant experience in the field of clinical research. As a research nurse, she has acted as lead nurse on several academic and commercial clinical trials with patients with neuromuscular diseases including Duchenne Muscular Dystrophy, FSHD and Becker Muscular Dystrophy, and she has had experience in providing clinical care to patients with Spinal Muscular Atrophy, Limb Girdle Muscular Dystrophies and other rare neuromuscular diseases. Her joint passion for clinical research and the importance of patient engagement has led to her taking on the role of Science and Patient Engagement Officer for the Lily Foundation. The Lily Foundation is the UK's leading mitochondrial disease charity and the largest charitable funder of mitochondrial research in Europe. The mission of the Lily Foundation is to improve the lives of people affected by mitochondrial diseases, while working towards a future where mitochondrial diseases can be effectively treated or cured. The Lily Foundation are also represented on the Board of IMP (International Mito Patients) and are proud to participate in many other European and global collaborations.

GERARD WELLENBERG

Member of the Muscle Diseases Specialist Group

Within Gerard Wellenberg's family, the hereditary muscular disease Myotonic Dystrophy type-1 (MD1) has been diagnosed in 2001. From 2001 on, he lost one

niece and one brother. His mother passed away in 1991 at the age of 58. Two other family members still suffer MD1. Gerard Wellenberg has more than 35 years of experience in the field of human and animal disease surveillance, control and eradication programs, epidemiology and the management of many research projects in the Netherlands and abroad. He has been active in the field of MD1 since 2012. He is the Chairman of the Dutch MD1 Action Team, which is involved in fundraising activities and raises Dutch citizens' awareness on the importance of MD1 research and on the impact of MD1 within families.

4. OVERVIEW OF PATIENT PARTICIPATION TO EURO-NMD WORKING GROUPS AND CONTACT DETAILS

Advisory Boards	Chair(s)	Patient representatives
Executive Board	Teresinha Evangelista (Sorbonne University – Pitié Salpêtrière Hospital – APHP)	François Lamy (AFM-Téléthon) Jean-Philippe Plançon (French Association against Peripheral Neuropathies)
Educational Board	Jean-Philippe Plançon (French Association against Peripheral Neuropathies) Sabrina Sacconi	Dimitrios Athanasiou (MDA Hellas) Isabela Tudorache (Parent Project Romania) Michela Onali (Gli Equilibristi HIBM) Carolina Navalon (Spanish Patients´ Association affected by Arthrogryposis Multiplex Congenita) Alejandra Perez del Real (GBS|CIDP España-Polineuropatias Inmunomedidas)
Research Board	Hanns Lochmüller	Marisol Montolio (Duchenne Parent Project Spain) Alexandre Méjat (AFM-Téléthon) Maria Borrell (ASEM) Michela Onali (Gli Equilibristi HIBM) Carolina Navalon (Spanish Patients´ Association affected by Arthrogryposis Multiplex Congenita) Alejandra Perez del Real (GBS|CIDP España-Polineuropatias Inmunomedidas)
Diseases working Group (WG)	**Chair(s)**	**Patient representatives**
Muscle Diseases	Marianne de Visser	François Lamy (AFM-Téléthon) Madelon Kroneman (Spierziekten Nederland) Michela Onali (Gli Equilibristi HIBM) Silke Schlüter (Deutsche Gesellschaft für Muskelkranke e.V - DGM) Marisol Montolio (Duchenne Parent Project Spain) Joaquim Brites (Associacao Portuguesa de Neuromusculares) Gerard Wellenberg (Dutch MD1 Action Team) Javier Torras (ASEM Catalunya) Fernanda de Angelis (Parent Project Italy) Ingrid de Groot (Spierziekten Nederland) Peter Ashley (Euro-DyMA) Maria Borrell (ASEM)

		Carolina Navalon (Spanish Patients´ Association affected by Arthrogryposis Multiplex Congenita) Ipek Badirgali (EAMDA) Isabela Tudorache (Parefnt Project Romania)
Peripheral Nerve Diseases	Davide Pareyson	Jean-Philippe Plançon (French Association against Peripheral Neuropathies) Massimo Marra (CIDP Italia) Alejandra Perez del Real (GBS|CIDP España-Polineuropatias Inmunomedidas) Filippo Genovese (ACMT-Rete per la malattia di Charcot-Marie-Tooth OdV) Arabela Acilanei (Neuro Move CMT Association, Romania)
Neuromuscular Junction Defects	Hanns Lochmüller	Marguerite Friconneau (AFM-Téléthon)
Motor Neuron Diseases	Jan Kirschner Vincenzo Silani	Evy Reviers (ALS liga Belgium) Eva Stumpe (SMA Europe) Vitaliy Matyushenko (Children with SMA, Ukraine) Ipek Badirgali (EAMDA) Joaquim Brites (Associacao Portuguesa de Neuromusculares)
Mitochondrial Diseases	Cornelia Kornblum Michelangelo Mancuso	Emma Del-Rey (French Association for Mitochondrial Diseases) Boryana Stoyanova (National Association of Patients with Mitochondrial Diseases, Bulgaria) Katie Waller (The Lily Foundation)
Crosscutting Specialist Groups	**Chair(s)**	**Patient representatives**
Neuromuscular Pathology	Montse Olive Plana	
Neuromuscular Imaging	Pierre Carlier	
Neurophysiology	Camiel Verhamme Peter van den Bergh	
Multidisciplinary Management and Care	Marianne Nordstrøm Andreas D. Rosenberger Hanne Ludt Fossmo Kristin Ørstavik	
Genetics	Alessandra Ferlini	Alexandre Méjat (AFM-Téléthon) François Lamy (AFM-Téléthon)

	Victoria Castillo Sanchez (Spanish Patients´ Association affected by Arthrogryposis Multiplex Congenita) Ipek Badirgali (EAMDA)