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

APRIL 2025

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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),
- **Arabela Acalinei** (Neuro Move CMT Association of Romania - European Alliance of Neuromuscular Disorder Associations EAMDA, EU),
- **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, European organisation for professionals and people with ALS – EupALS, EU).

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

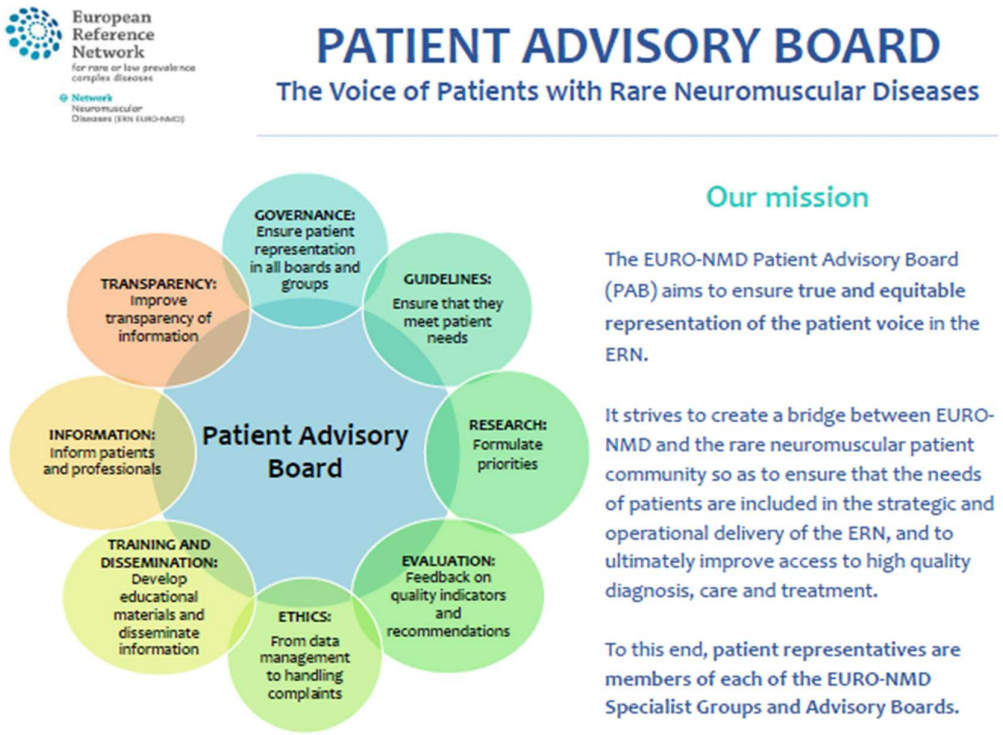
patient needs. For this, they engage with patient representatives beyond EURO-NMD to appoint the best person for each task.

- 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
Member of the Registry Steering Committee

François Lamy is the father of a boy with Duchenne Muscular Dystrophy. He has been elected as a member of the Board of Directors of the AFM-Téléthon since 2012, and currently serves as its Vice-President in charge of Téléthon. François Lamy also serves on the Board of Directors of the Center for the Study of Stem Cells (I-Stem CECS) and GenoTher (French biocluster dedicated to Gene Therapy).

ARABELA ACALINEI

Member of the Patient Advisory Board
Member of the Peripheral Nerve Diseases Working Group
Member of the Inter-ERN Gene Therapy Working Group

Arabela Acălinei serves as the President of Neuro Move CMT Association in Romania and President of EAMDA - the European Alliance of Neuromuscular Disorders Associations. Since 2020, she is a Board Member of the European CMT Federation, serving as their Eastern Europe Ambassador. Furthermore, since 2024, she is a Board Member of EFNA – the European Federation of Neurological Associations. 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. Dealing with such a disease, ignited her passion for patient advocacy, particularly for individuals living with CMT and similar neuromuscular disorders. In her professional life, Arabela is a Romanian/English teacher, translator, and professional trainer. Her educational background also includes trainings in Project Management and Career Counseling for people with disabilities. Furthermore, Arabela Acălinei is a EURORDIS Winter and Summer School alumni, EUPATI fellow and a member of EURORDIS HTA Task Force. In July 2023, she graduated the 5th Neuromuscular Translational School, organized in Leiden, Netherlands by EURO-NMD and TREAT-NMD. Beyond her advocacy and professional pursuits, she is passionate about reading, learning, traveling, and connecting to people.

DIMITRIOS ATHANASIOU

Member of the Patient Advisory Board
Member of the Registry Steering Committee

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
Member of the Multidisciplinary Management and Care Working Group
Member of the Inter-ERN Gene Therapy Working 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
Member of the Neuromuscular Pathology Specialist Group

Massimo Marra, following a personal experience, founded the Italian Association Against Dysimmune Neuropathies in 2012 to address issues related to access to treatment in Italy. Since then, he has been actively involved in promoting information and awareness campaigns, supporting awards and grants for young researchers, and advancing research on improving patients' quality of life.

Massimo is particularly interested in the importance of listening to the needs of patients and their families, as well as exploring how and what can be done to provide effective solutions.

PATRICIA MELSOM

Member of the Patient Advisory Board
Member of the Multidisciplinary Management and Care Working Group

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 a founding member of The Neuromuscular Disorders Association of Norway (Foreningen for muskelsyke – FFM), has served as President in several periods and is at present project developer and policy advisor. 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
Member of the Working Group on Postgraduate Curriculum for NMDs

Jean-Philippe Plançon was born in 1969 and has been living with a rare immune-mediated neuropathy for 25 years. He graduated in the field of public health, education, rare diseases, advocacy, business and corporate strategy. In 2006 Jean-Philippe founded *Association Française contre les Neuropathies Périphériques* (AFNP), a patient organization supporting people affected by rare neuromuscular diseases such as Guillain-Barré syndrome, CIDP, MMN, Lewis-Sumner syndrome, Miller Fisher and other related disorders including painful neuropathies. Since 2019, Jean-Philippe is the president of the European Patient Organization for Dysimmune and Inflammatory Neuropathies (EPODIN) and he is also the vice-president of the French Rare Diseases Alliance, composed of 240 rare diseases patient associations. After having been a member of several expert committees of the French National Agency for the Safety of Medicines and Health Products (ANSM), since 2021 Jean-Philippe is vice-chairman of the Board of Directors of this agency.

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 2007 and Chairwoman since 2021 of ALS Liga Belgium, Evy Reviers performs the general management of the organisation and stand up for the voice of people living with ALS in Europe and on international scale. 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, patient representative at EMA, and EURORDIS. She has served already for several terms as a member of the Board of Directors of the International Alliance of ALS/MND Associations and is elected again at the 2024 AGM.

3. OTHER EURO-NMD PATIENT REPRESENTATIVES.

LUTGARDE ALLARD

Member of the Neuromuscular Junction Defects Diseases Specialist Group

Lutgarde was born in 1959 and was diagnosed with Myasthenia Gravis in March 1994. She was educated to be a nurse and worked as a civil servant from 1980 to 2010. She was a member of the Flemish Myasthenia Gravis organisation from 1994 to 2023. Lutgarde has been a patient advocate since 1998 and a contact person for other Myasthenia Gravis patients. Since May 2023, she is a member of the board of the European Myasthenia Gravis Association (EuMGA) and its treasurer. In 2023, she attended an in-depth training on patient advocacy organised by Patvocates and took an active role in the development of the EURO-NMD patient journey on Myasthenia Gravis.

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.

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 63 years old, married and has two children, one of them, a boy 32 years old, with Duchenne Muscular Dystrophy. An active Patient Advocate in Neuromuscular Disorders and rare diseases, he was President of the General Assembly of the Portuguese Alliance of Rare Disease Associations between 2012 and 2021 and was a member of the Ad Hoc Committee and of the Installing Committee of the New National Representative Organization of the Rare Disease Associations in Portugal. Currently, he still represents Portugal in the Rare Diseases Platform in France and remains a representative of the Portuguese neuromuscular patients in several European organizations. He is a member of several EURO-NMD working groups, and a member of Treat-NMD, both as an individual and in representation of APN. At the national level, he is a member of the Commission for Inclusion Policies for People with

Disabilities of the Ministry of Labor and Social Security, a founding Member of the Platform SIP-PT (Societal Impact of Pain - Portugal), a member of the Mission Structure for the Promotion of Accessibilities, representing motor disability, a member of the Monitoring Committee of the National Strategy for the Inclusion of People with Disabilities, and a member of the National Health Council, representing APN.

LIESBET CASIER

Member of the Motor Neuron Diseases Specialist Group

Liesbet Casier has been with the ALS Liga Belgium since 2020. With a background in psychology, she is responsible for the support and guidance of people living with Amyotrophic Lateral Sclerosis (ALS), their loved ones and healthcare professionals in Belgium. In 2023, she became an ALS casemanager, a position framed within a government project that is committed to deploying specialized knowledge to support people in complex care situations with a goal of better quality of life.
She also started using the accumulated knowledge and expertise to work structurally for a better life for people living with ALS, by advocating before political bodies, participating in projects at the national and European level (within different working groups) as a member of INARC (International Network for ALS Research and Care) and by actively working on education for people who come into contact with ALS from different perspectives.

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.

LISE CONNELLY

Member of the Muscle Diseases Specialist Group

Lise Connelly is married with 3 children, where her youngest has the same diagnose as her, Central Core disease mith RyR1. She is involved in work on exercise for children and adults with NMD, rehabilitation, educational needs of children and young people,

and ethics. She is also involved in a multi-centre research project to develop a new muscle biopsy test for malignant hyperthermia in NMD.

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.

EMMA DEL-REY

Member of the Mitochondrial Diseases Specialist Group

Emma Del-Rey is the president of the AMMi association (French 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 centres 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 President of the European CMT Federation and Chief Scientific Officer 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.

BINE HAASE

Member of the Muscle Diseases Specialist Group

Bine Haase is a passionate advocate for individuals living with facioscapulohumeral muscular dystrophy (FSHD), a neuromuscular disease she inherited from her mother. Born in 1978, Bine has been actively involved in the FSHD community for many years. Since 2015, she has served on the board of the FSHD group within the DGM e.V. (Deutsche Gesellschaft für Muskelkranke – German Society for Muscle Diseases). Her commitment to the cause extends to the European level, where she has been a board member of FSHD Europe since 2016. Furthermore, Bine represents the patient perspective at prominent organizations, including EURORDIS (Rare Diseases Europe), EUPATI (European Patients' Academy on Therapeutic Innovation), and the EMA (European Medicines Agency), ensuring that the voices of those affected by FSHD are heard in crucial discussions about research, treatment, and policy.

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 joined the Scientific Department of AFM-Telethon in 2018 where he currently holds the position of Head of International Scientific Networks. He is also member of the Board of the European NeuroMuscular Centre (ENMC) and EURORDIS Rare Diseases Europe and is involved in the European Rare Diseases Research Alliance (ERDERA) and International Rare Diseases Research Consortium (IRDiRC).

CAROLINA NAVALON-MARTINEZ

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

Carolina Navalon-Martinez is a Barcelona-based Patient Advocate representing several associations focusing on: Rare Neuromuscular Disorders, Rheumatological and Autoimmune Illnesses, and Sports & Disability. Carolina 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 and Vice-President of the Spanish Patients´ *Association for Arthrogryposis Multiplex Congenita (AMC España*). She is also a Board Member of the C*atalan League for inflammatory rheumatic diseases*. Furthermore, she is also the founding member and President of the Association *Blue Health SUP Terapéutico*, an NGO advocating for the establishment of therapeutic aquatic sports programs. She attended the Eurordis Summer School (Spanish version) on Medicines Research and Development in 2018, becoming a Patient Advocate after completing the course. She has also been an EMA adviser.She is the main liaison for the creation of the 1st Spanish Patient Registry for AMC Arthrogryposis (joint project with *Paediatric Hospital Sant Joan de Deu Barcelona, Shriners Hospital Montreal-Canada and Ciberer*, the *Center for Biomedical Network Research on Rare Diseases in Spain*). She is part of ongoing working groups with international experts worldwide to create an AMC Arthrogryposis International Patient Registry and an AMC Alliance. She has assisted as a Patient Expert Advocate on the definition of the Common Data Elements, data sharing accessibility, Pre-Delphi (2021-2022) from the patient´s and patient representative perspective, which lead to the publishing of the scientific article ¨*Common data elements for arthrogryposis multiplex congenita: An international framework*¨.

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.

ECE SOYER DEMIR

Member of the Motor Neuron Diseases Specialist Group

Ece Soyer Demir was born in Eskişehir in 1989. She graduated as a primary school teacher from the Faculty of Education at Eskişehir Osmangazi University in 2010. She completed her master's degree in the field of "Educational Measurement and Evaluation." Throughout her career, she has worked in various cities across Turkey.

After her son, Çağan Meriç, was diagnosed with SMA, she became one of the founders of the Turkey SMA Foundation, where she currently serves as the Vice Chair of the Board of Directors. She contributed to the SMA Family Information Book, which was prepared as a guide for SMA patients and their families, by writing the sections titled "Advice from Experienced Caregivers" and "Our Rights in Education." After its publication, the book was translated into two different languages. She compiled her memories with her son, Çağan Meriç, into a book titled "SMA – A Hope That Keeps People Alive," published by Doğan Publishing, aiming to guide newly diagnosed families and raise awareness about SMA.

She has actively engaged in advocacy efforts for the inclusion of SMA carrier screening and newborn screening in national screening programs. To raise awareness about the disease and its preventability, she has participated as a speaker in numerous national seminars and conferences. She also took part in the documentary film "I Am SMA" to highlight the challenges faced by patients and their families. A mother of one, Ece Soyer Demir dreams of a world where all children have an equal and fair chance to pursue their dreams and where their voices are heard.

BORYANA STOYANOVA

Member of the Mitochondrial Diseases Specialist Group

Boryana Stoyanova is a mother of a 24-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 54 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 18 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 4 years ago, 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.

MAYA BONARIA UCCHEDDU

Member of the Neuromuscular Junction Defects Diseases Specialist Group

Maya is a patient living with Myasthenia Gravis (MG), a pharmacist, and a freelance digital artist with a master's degree in pharmaceutical chemistry and technology. Maya co-owns and manages a para-pharmacy in Italy. She advocates for MG patients at national and international levels, serving as the chairperson for the MG patient advocacy committee in the European MG Association (EuMGA) and as a member of the Italian association AIM (Associazione Italiana Miastenia e Malattie Immunodegenerative). She successfully completed the "MG Advocate Development Program" and is attending the Eurordis "School on Medicines Research & Development".

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

Advisory Boards	Chair(s)	Patient representatives
Executive Board	Teresinha Evangelista	François Lamy (AFM-Téléthon) Jean-Philippe Plançon (French Association against Peripheral Neuropathies)
Educational Board	Jean-Philippe Plançon	Isabela Tudorache (Parent Project Romania) Michela Onali (Gli Equilibristi HIBM) 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)
Patient Advisory Board	François Lamy	Arabela Acalinei (EAMDA) Dimitrios Athanasiou (MDA-Hellas, WDO) Madelon Kroneman (Spierziekten Nederland) 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, EPODIN) Evy Reviers (ALS Liga Belgium)
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) Peter Ashley (Euro-DyMA)

		Maria Borrell (ASEM) Carolina Navalon (Spanish Patients´ Association affected by Arthrogryposis Multiplex Congenita) Ipek Badirgali (EAMDA) Isabela Tudorache (Parent Project Romania) Lise Connelly (Neuromuscular Disorders Association of Norway) Bine Haase (DGM)
Peripheral Nerve Diseases	Davide Pareyson Kleopas Kleopa	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 Lorenzo Maggi	Marguerite Friconneau (AFM-Téléthon) Lutgarde Allard (EuMGA) Maya Uccheddu (Associazione Italiana Miastenia e Malattie Immunodegenerative – AIM)
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) Liesbet Casier (ALS Liga Belgium) Ece Soyer Demir (Turkey SMA Foundation)
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	Edoardo Malfatti	Massimo Marra (CIDP Italia)
Neuromuscular Imaging	Pierre Carlier	
Neurophysiology	Camiel Verhamme Shahram Attarian	
Multidisciplinary Management and Care	Marianne Nordstrøm Andreas D. Rosenberger Hanne Ludt Fossmo	Patricia Melsom (Neuromuscular Disorders Association of Norway) Madelon Kroneman (Spierziekten Nederland)

	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)