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 2022

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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 in the Network, and liaising with its affiliated patient organisations. The Patient Advisory Board also endorses additional patient representatives to join Specialist Groups based on their expertise.

The PAB has established its own Constitution and Rules of Procedure.

Membership:

Members of the PAB include those elected via EURORDIS who constitute the European Patient Advocacy Group (ePAG) for EURO-NMD. Members from umbrella organisations (e.g. Spierziekten Nederland) have also been invited to join the Patients Advisory Board to ensure a proper representation of the neuromuscular patient community among the PAB.

ePAG representatives:

- François Lamy (AFM-Téléthon, France),
- Dimitrios Athanasiou (MDA-Hellas),
- Massimo Marra and Patrizia Garzena (alternate) (CIDP Italia ONLUS),
- 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),

Other NMD patient representatives:

- Madelon Kroneman (Spierziekten Nederland, Dutch Patient Society of Neuromuscular Diseases).

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

- Governance of the ERN: The EURO-NMD Board will include the patient representatives that are part of the Patient Advisory Board. One representative of the PAB will also be part of the Executive Committee (see *Board Terms of Reference*).

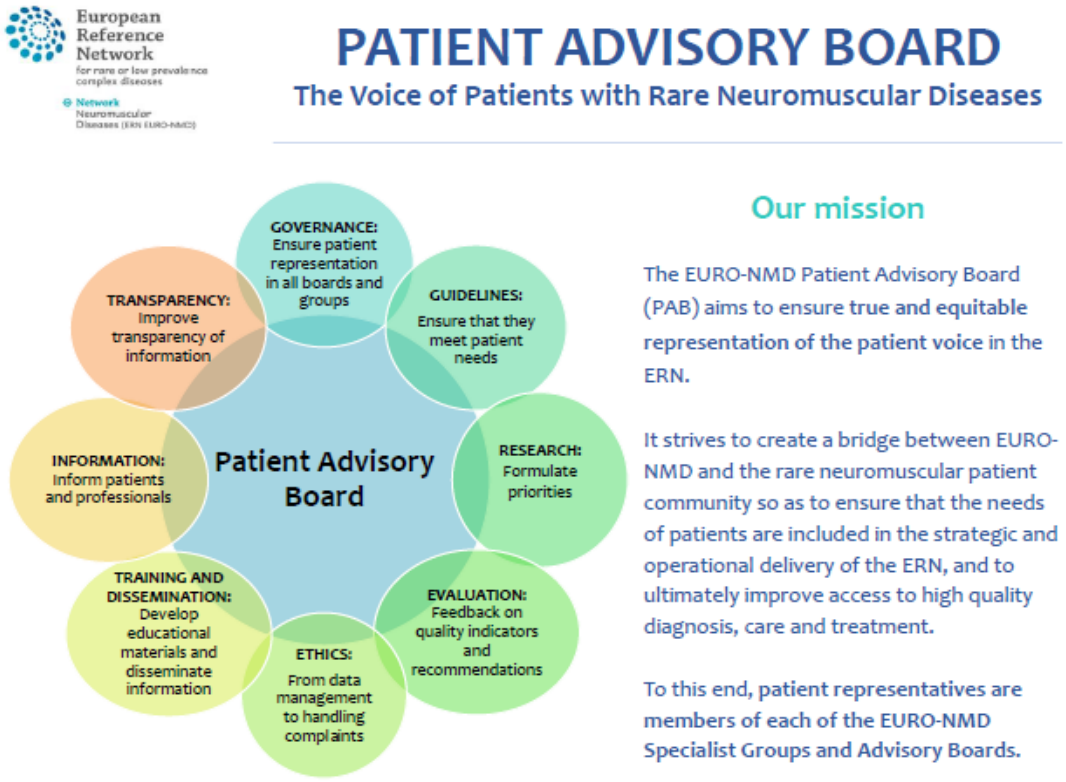
- Activity Groups and Advisory Boards: The PAB will nominate patient representatives to be part of each Activity Group and Advisory Board.

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

- Ethics: PAB members will be involved in ethics activities. The PAB will appoint one patient representative as the chair of the Ethics Committee.

- Research: PAB members will 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 will play a key role in network evaluation, 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 Board
Member of the Muscle Diseases Specialist Group
Member of the Genetics Specialist Group
Contact: flamy@afm-telethon.fr

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 research. 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
Contact: dathax@gmail.com

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 founded the Parent Project of MDA HELLAS in Greece. Currently, he is a Board Member of the World Duchenne Organization (WDO), a European Medicines Agency Patient Expert in Duchenne Muscular Dystrophy, and co-chairs EFGCP's Children's Medicine Working Party (CMWP). He is DIA's EuroMeeting 2017 Program Committee Member. 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.

PATRIZIA GARZENA

Member of the Patient Advisory Board (alternate to Massimo Marra)
Contact: garzena.lavoro@gmail.com

Patrizia Garzena was diagnosed with chronic inflammatory demyelinating polyneuropathy in 2016 whilst she was living in Sweden. Having moved back to Italy, in 2017 she joined CIDP Italia APS, the Italian non-profit Association for patients with Dysimmune Neuropathies. She is part of EPODIN's Board of Directors, the European Patient Organization for Dysimmune and Inflammatory Neuropathies.

MADELON KRONEMAN

Member of the Patient Advisory Board
Member of the Ethics Committee
Member of the Muscle Diseases Specialist Group
Contact: madelonkroneman@gmail.com

Madelon Kroneman was diagnosed with Myofibrilar Myopathy (Desminopathy). Her disease started at the age of 33. 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 now the editor of the working group newsletter. She also follows scientific developments and is involved in reviewing scientific research concerning muscular diseases, both for funding bodies and for research requiring the involvement of members.

MASSIMO MARRA

Member of the Patient Advisory Board
Member of the Peripheral Nerve Diseases Specialist Group
Contact: cidpitaliaonlus@gmail.com

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 has promoted 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.

MARISOL MONTOLIO

Member of the Patient Advisory Board
Member of the Muscle Diseases Specialist Group
Member of the Research Board
Contact: research@duchenne-spain.org

With more than 15 years of research experience, Marisol Montolio is the Scientific Director 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 at the University of Barcelona.

MICHELA ONALI

Member of the Patient Advisory Board
Member of the Muscle Diseases Group
Member of the Research Board
Member of the Educational Board
Contact: michela.onali@gmail.com

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
Contact: jean-philippe.plancon@epodin.org

Jean-Phillipe 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

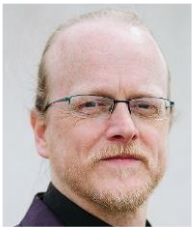
Member of the Patient Advisory Board
Chair of the Ethics Committee
Member of the Executive Committee
Member of the Motor Neuron Diseases Specialist Group
Contact: evy@als.be

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

PETER ASHLEY



Member of the Muscle Diseases Specialist Group

Contact: pete@cmmd.uk

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 carers. 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.

PATRICIA BLOMKWIST-MARKENS

Member of the Peripheral Nerve Diseases Group

Contact: gbsliaison@gmail.com

After her recovery from a severe case of Guillain-Barré syndrome (GBS) in 1990, Patricia Blomkwist-Markens founded the Dutch GBS|CIDP support group (which later merged with Spierziekten Nederland, the Dutch neuromuscular patient organization). In 1991, she was appointed as volunteer (liaison) of the GBS|CIDP Foundation International and some years later as Regional Director, responsible for the Foundation's volunteers outside of the United States and Canada. Since 2005, she has been a member of the Board of Directors of the Foundation and in recent years, Vice-President for International Activities. As patient representative, Patricia has been involved in several workshops of the European Neuromuscular Center, has co-authored publications and is currently a member of the Task Forces for the development of EAN/PNS Guidelines for GBS and CIDP.

MARIA BORRELL

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

Contact: mbpbcn@yahoo.com

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

Contact: presidente@apn.pt

Joaquim Brites has a 29-year-old son with Duchenne Muscular Dystrophy. Joaquim Brites has been involved with APN, a Portuguese neuromuscular association, for more than 20 years. He was a member of APN's Board of Directors and became its President in 2012. APN focuses on direct support for neuromuscular disease patients and their families. Joaquim Brites is also the President of the General Assembly of the Portuguese Alliance of Associations of Rare Diseases since 2012 and represents Portuguese neuromuscular patients, in several European organisations. He represents Portugal in the Rare Diseases Platform ("Plateforme Maladies Rares") in France.

FERNANDA DE ANGELIS

Member of the Muscle Diseases Specialist Group

Contact: f.deangelis@parentproject.it

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

Contact: ingrid.de.groot@upcmail.nl

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.

EMMA DEL-REY

Member of the Mitochondrial Diseases Specialist Group

Contact: emma.delrey31@gmail.com

Emma Del-Rey has a daughter who was diagnosed with Mitochondrial Disease. Since 2008, she is the President of AMMI, a French Mitochondrial Disease Organisation. She organises meetings for the regional delegate and national, provides information to hospitals and families, and organises events to raise funds for scientific projects and help affected families. She is a member of International Mito Patients (IMP), part of the board since 2011, and is in charge of international project for mitochondrial patients.

YASEMIN ERBAS

Member of the Motor Neuron Diseases Specialist Group

Contact: Yasemin.Erbas@sma-europe.eu

Yasemin Erbas has a son with Spinal Muscular Atrophy (SMA) type 1. She is a patient representative for the Belgian-Flemish association Spierziekten Vlaanderen, co-founder of SMA Belgium, and vice-president of SMA Europe, the umbrella of European SMA patient and research organizations. For SMA Europe, she advices EMA on scientific protocols and she is the co-lead of the evidence generation workgroup. She is a strong believer of supporting advocacy work with patient-centric research. She has a PhD in Psychology from the KU Leuven, Belgium, and currently works as an Assistant Professor in Developmental Psychology at Tilburg University, The Netherlands.

MARGUERITE FRICONNEAU

Member of the Neuromuscular Junction Defects Specialist Group

Contact: mfriconneau@afm-telethon.fr

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.

JEREMIE GAUTREAU

Member of the Peripheral Nerve Diseases Specialist Group

Contact: Jeremie.gautreau@neuropathies-peripheriques.org

Jérémie Gautreau suffers from Chronic Inflammatory Demyelinating Polyneuropathy. He works as a Health and Safety Consultant at the GHBS (Hospitals of South Britanny) to improve working conditions for hospital personnel and is the Secretary of the AFNP- French Association against Peripheral Neuropathies. The AFNP helps patients with their neuropathy and works to raise awareness about these diseases. Jérémie values the importance of helping others and has been a volunteer firefighter for 10 years. Trained in the domain of patient therapeutic education, he works on the development of a therapeutic education program to improve patients' quality of life.

SYLVIE GENET

Member of the Muscle Diseases Specialist Group

Contact: sgenet@afm-telethon.fr

Sylvie Genet, born in 1960, mother and grand-mother, FSHD, ex-engineer, ex-teacher, on medical leave before retirement. She has been a member of AFM-Téléthon for around 20 years, is active in the patient support group for FSHD (scientific and medical news, social networks). She would like to share widely (patients, doctors, scientists) the expertise on FSHD she acquired during these years of volunteering.

FILIPPO GENOVESE

Member of the Peripheral Nerve Diseases Specialist Group

Contact: filippo@acmt-rete.it

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 Group

Contact: csma@ukr.net

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 more than 15 years of public experience 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

Contact: amejat@afm-telethon.fr

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 as International Scientific Affairs manager.

CAROLINA NAVALON-MARTINEZ

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

Contact: cnavalon@artrogriposis.org

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.

MARIE-CHRISTINE OUILLADE

Member of the Ethics Committee

Contact: marie-christine.ouillade@wanadoo.fr

Marie-Christine Ouillade has a daughter who has been diagnosed with Spinal Muscular Atrophy type II. Marie-Christine Ouillade has been actively involved in the activities of the AFM-Téléthon since 1996, and became a member of its Board of Directors in 2006, as well as a member of the Board of Directors of Genethon since 2007. She is also a member of the TREAT-NMD Project Ethics Council since 2007, a Board Member of the International Rare Diseases Research Consortium since 2011, and the former President of SMA Europe. Between 2006 and 2012, she was a member of the Steering Committee of the French Biomedicine Agency. In addition, Marie-Christine Ouillade chairs the steering committee of the SMA NBS Alliance, advocating for inclusion of SMA into newborn screening panel.

JACQUES SALAMA

Member of the Neuromuscular Imaging Specialist Group

Contact: jsalama@hotmail.fr

Jacques Salama is the father of a young man with Duchenne Muscular Dystrophy. After his son was diagnosed with DMD in 1990, he joined the AFM-Téléthon. He later became its General Delegate for scientific policy, and joined the Board of Directors of Genethon in 1996. In 1999, he became the General Secretary of the Institute of Myology in France, an institute dedicated to research, care and education on muscle function and muscle diseases. There, he worked closely with the scientists developing medical imaging applied to muscle diseases. To this day, he appreciates the value of these new techniques of imaging to provide the possibility of non-invasive tools for diagnosis and follow-up of emerging therapies; they are of fundamental importance for the future development of drugs.

SANDRINE SEGOVIA-KUENY

Member of the Neurophysiology Specialist Group

Contact: ssegovia-kueny@afm-telethon.fr

Sandrine Segovia-Kueny is the Medical Director at AFM-Téléthon. She is a doctor of medicine, graduate of the University of Nice, France, and holds an Executive Advanced Master in Strategy and Management of Health Industries from the ESSEC Business School, France. Sandrine Segovia-Kueny also brings 15 years of experience in the public sector, including several years as a Deputy General Director in the Health Regional Agency in Lille, as an Environmental and Public Health Director in the Health Regional Agency in Orleans, as an Health

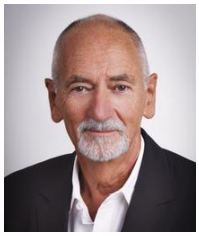
Advisor in the French Ministry for sustainable development and a Medical Advisor in the General Health Direction in Paris. Sandrine Segovia-Kueny is also a Board and an Advisory Committee member in various public organisations.

BORYANA STOYANOVA

Member of the Mitochondrial Diseases Specialist Group
Contact: info@mitobg.com

Boryana Stoyanova is a mother of a 21-year-old boy who was diagnosed at the age of 14 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. She played an active role so that the association becomes a member of International Mito Patients and the National Patients Organization in Bulgaria. She is in charge of creating social media content for the website and the Facebook page of the association and took part in the organization of Light Up for Mito in September 2021 (for the first time in Bulgaria). In December 2021, Boryana was elected as secretary of the Rare Disease and Transplantations Section of the National Patients Organization in Bulgaria and currently is part of the Advisory Board of EmpoweRARE2021 project in Bulgaria. Boryana is a lawyer with over 20 years of experience in commercial and corporate law.

DANIEL TANESSE



Member of the Peripheral Nerve Diseases Specialist Group
Contact: tanesse@free.fr

Daniel Tanesse and his wife both suffer from Charcot-Marie-Tooth (CMT). Daniel Tanesse first joined Charcot-Marie-Tooth France as a Regional Delegate in 1995. He later became its Referent for Congress and now serves as its Vice-President. He is also one of the founders of the CMT-Europe Consortium, which aims, *inter alia*, at supporting all European CMT associations, enhancing patient representation, and helping researchers and laboratories to find volunteers for future therapeutics trials. In November 2017, he was elected Chairman of the European CMT Federation. He has himself been and still is involved in therapeutics trials for CMT: as a member of the ethical committee for the acid ascorbic trial (2004), as an investigator for the PXT-3003 trial, and as a scientific and technical correspondent with the candidate drug called IFB-088 from InFlectisBioScience laboratory.

JAVIER TORRAS

Member of the Muscle Diseases Specialist Group
Contact: torrasjavier@gmail.com

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
Contact: isatudo@yahoo.com

Isabela Tudorache has more than 15 years of experience in social support activities for disadvantaged groups, in collaboration with government and non-government organizations. Her work included: delivering strategies of social inclusion for various vulnerable groups, combating human trafficking, child abandonment prevention, counselling and also implementing EU-funded projects and programs to address poverty and the disability in the region. In March 2004, together with another Duchenne parent, she founded Parent Project Romania - Association for Research and Support in Muscular Dystrophy, and, in time, she received membership of UPPMD, the TREAT-NMD Global Database Oversight Committee, and ANBRaRo – the nation organization of rare diseases. Her current role is to establish partnerships between stakeholders in order to improve the life expectancy and the quality of life for all affected by DMD in Romania, to provide advocacy for research and implementation of international standards of care for DMD/DMB and other NMD's in Romania.

DIANA VAN DER MEIJ-KIM

Member of the Muscle Diseases Specialist Group
Contact: dvdmeij@yahoo.co.uk

Diana van der Meij-Kim has a son who was diagnosed with FSHD at the age of 10. She subsequently took an active position in promoting the research for FSHD and within a year she became a member of the FSHD working group of the Dutch patient organisation VSN and a Board Member of the Dutch FSHD Foundation (the latter of which she is no longer a Board member). As a member of Dutch patient organisations, Diana van der Meij-Kim joined FSHD Europe and became its President in 2016.

KATIE WALLER

Member of the Mitochondrial Diseases Specialist Group
Contact: katie@thelilyfoundation.org.uk

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

Contact: gerardwellenberg1@gmail.com

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

Board/Committee	Chair(s)	Patient representatives
Executive Board	Teresinha Evangelista (Institute of Myology)	François Lamy (AFM-Téléthon) Evy Reviers (ALS Liga Belgium) Jean-Philippe Plançon (French Association against Peripheral Neuropathies)
Ethics Committee	Evy Reviers (ALS Liga Belgium)	Madelon Kroneman (Spierziekten Nederland) Marie-Christine Ouillade (AFM-Téléthon) Carolina Navalon (Spanish Patients´ Association affected by Arthrogryposis Multiplex Congenita)
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)
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)
Muscle Diseases	Marianne de Visser	François Lamy (AFM-Téléthon) Marisol Montolio (Duchenne Parent Project Spain) Joaquim Brites (Associacao Portuguesa de Neuromusculares) Gerard Wellenberg (Dutch MD1 Action Team) Diana van der Meij-Kim (FSHD Europe) Madelon Kroneman (Spierziekten Nederland) Javier Torras (ASEM Catalunya) Fernanda de Angelis (Parent Project Italy)

		Ingrid de Groot (Spierziekten Nederland) Michela Onali (Gli Equilibristi HIBM) Sylvie Genet (AFM-Telethon) Peter Ashley (Euro-DyMA) Maria Borrell (ASEM) Carolina Navalon (Spanish Patients´ Association affected by Arthrogryposis Multiplex Congenita)
Peripheral Nerve Diseases	Davide Pareyson	Jean-Philippe Plançon (French Association against Peripheral Neuropathies) Daniel Tanesse (CMT-France) Massimo Marra (CIDP Italia) Patricia Blomkwist-Markens (Spierziekten Nederland) Filippo Genovese (ACMT-Rete per la malattia di Charcot-Marie-Tooth OdV) Jérémie Gautreau (French Association against Peripheral Neuropathies)
Neuromuscular Junction Defects	Hanns Lochmüller	Marguerite Friconneau (AFM-Téléthon)
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)
Motor Neuron Diseases	Leonard van den Berg	Evy Reviers (ALS liga Belgium) Yasemin Erbas (SMA Europe) Vitaliy Matyushenko (Children with SMA, Ukraine)
Neuromuscular Imaging	Pierre Carlier	Jacques Salama (Institute of Myology)
Neurophysiology	Camiel Verhamme Peter van den Bergh	Sandrine Segovia-Kueny (AFM-Téléthon)
Neuromuscular Pathology	Montse Olive Plana	
Genetics	Alessandra Ferlini	Alexandre Méjat (AFM-Téléthon) François Lamy (AFM-Téléthon)