Nominees for Academic Representative of the TREAT-NMD Executive Committee

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Nominees are listed in alphabetical order by surname.

Nominee Name: Virginia Arechavala-Gomeza

Organisation/Affiliation: Biocruces Bizkaia Health Research Institute

Please provide a summary of your neuromuscular career to date:
My first degree is Pharmacy and I am a PhD in Neurology (on the subject of amyotrophic lateral sclerosis, 2005). As a postdoctoral researcher, I contributed to the development of new treatments for neuromuscular disorders: exon skipping drugs for Duchenne muscular dystrophy such as eteplirsen and new dystrophin quantification methods that had been used in the evaluation of many DMD clinical trials. While most of my research career took place in the UK, I now lead the Neuromuscular Disorders Research Group at IIS Biocruces Bizkaia in Spain (www.arechavala-lab.com), where I have developed new methods to quantify muscle proteins in the preclinical development of new treatments. My current research interests include Duchenne muscular dystrophy, Myotonic Dystrophy and Collagen VI-related myopathies. I am extremely interested in the rapid application of new research developments into the clinic and I lead a network of stakeholders (researchers, clinicians, pharmaceutical companies and patients) interested in solving the problem of the poor delivery of RNA drugs to target tissues. This network currently includes more than 300 participants from 30 countries (www.antisenserna.eu) and it is funded until October 2022. You may find more about my academic publications here: https://tinyurl.com/wmlvsdb

Please describe your involvement in TREAT-NMD to date:
I have been a member of TREAT-NMD from its early days when I worked in the UK and I have continued my engagement after I returned home. I have actively participated in many TREAT-NMD activities: as a researcher, I have participated and also organised meetings sponsored by the network (such as a recent stakeholder meeting on the subject of dystrophin quantification, published here: https://tinyurl.com/vx6mjc4), I have also had the pleasure of participating as an active observer in a TACT meeting, and I have also contributed to communicate research activities in lay terms, translating to Spanish the DMD research overview: https://tinyurl.com/wukt8ng, amongst other contributions.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: I think my expertise may complement that of other members of the committee: I am, on one hand, a basic researcher, interested in thorough researched facts to advance treatment development. On the other hand, I am a pharmacist and I am thrilled to see science applied to treatments as rapidly as scientifically feasible. I also have extensive experience in science communication, which I do in Spanish and English, and many of the lectures and webinars I have given in Spanish are freely available in the web, http://www.arechavalalab.com/publications/dissemination/.

But the main reason that would make me a good candidate is that I am a passionate advocate for networking and a fan of the TREAT-NMD network, and I would love to contribute to make it as good as possible. Science in general, but in particular research in rare diseases cannot advance without collaboration and the participation of all stakeholders, (including patients) and I think TREAT-NMD represents the sort of collaboration that makes research advance for all.

Nominee Name: Jorge Alfredo Bevilacqua Rivas

Organisation/Affiliation: 1. Hospital Clínico Universidad de Chile. 2. Clínica Dávila Santiago, Chile

Please provide a summary of your neuromuscular career to date:
Adult Neurologist at the Hospital Clínico Universidad de Chile. Muscle Pathology training at the Hospital de Pediatría J. P. Garrahan, Buenos Aires, Argentina with Dr. AL Taratuto, trained in Electrophysiology EMG and NCS, Hospital Clínico Universidad de Chile, Diploma in Myology at the Université Paris 6 UPMC and Institute of Myology, Paris, France. Currently Full Professor of the Department of Neurology and Neurosurgery, Hospital Clínico Universidad de Chile and Head of the Neuromuscular Unit. Director of the Neuromuscular Disorders Fellowship Program of the University of Chile. Head of the Laboratory of Muscle Pathology at the Clínica Dávila, Santiago, Chile. Member of the Scientific Committee of the Sociedad Latinoamericana de Enfermedades Musculares (SOLANE); Founder Member of the Grupo Euro-Latinoamericano de Miología (GrELAM), and Founder Director/Coordinator of the Escuela Euro-Latinoamericana de Miología (EVELAM) is annually organized. Membre d'Honneur Étranger of the Societé Francaise de Neurologie (SFN), member of the American Association of Neuromuscular and Electrodiagnostic Medicine (AANEM), the American Academy of Neurology (AAN) and the World Muscular Society (WMS). Focused on the development of the clinical myology in Chile, and the implementation of imaging, electrophysiology, pathology and genetic diagnosis of muscle diseases. His basic research has been focused on limb girdle muscular dystrophies in particular dysferlinopathy; Pompe's disease, centronuclear myopathy, having support mainly from the Comisión Nacional de Ciencia y Tecnología de Chile. PI of the JAIN COS Dysferlinopathy Study, Chilean Site.

Please describe your involvement in TREAT-NMD to date:
Treat NMD Individual Member since 2017; Member of TREAT-NMD Education Committee.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: I feel that I could contribute with the LATAM vision to the TREAT-NMD network, which is in line with the objective of globalizing TREAT-NMD coverture. I have knowledge of the global scenario of the neuromuscular field, I have active collaborations with specialists in LATAM, EUROPE, and North America, as well as effective communication in Spanish, French and English. I am also currently involved in several different networks as the International Congress of Neuromuscular Disorders, SOLANE, EVELAM, etc. Finally, my academic background including my experience in teaching and research gives me a good perspective that may help to take adequate decisions for the benefit of the network and its members.

Nominee Name: Gillian Butler-Browne

Organisation/Affiliation: Institute of Myology

Please provide a summary of your neuromuscular career to date:
Gillian Butler-Browne began working on muscle at the Pasteur Institute in 1978. She characterizing the expression profile of different myosin isoforms during both human and rodent development and has published over 270 articles covering different aspects of muscle biology. She demonstrated the impact of the limit in proliferation of the precursor cells on the regenerative capacity of human skeletal muscle. Her group developed models to evaluate the regenerative potential of skeletal muscle in vitro by calculating the proliferative life span, measuring the telomeric erosion that accompanies cell divisions, and identified the involvement of a stress pathway in the proliferative arrest of muscle progenitors. A platform to produce immortalized muscle cell lines was the successful result of these findings. The in vivo regenerative potential of human muscle cells was evaluated by injection into muscles of immuno-deficient mice. A new dystrophic immunodeficient mouse model RAG/Dys was developed to do this as well as a novel human muscle xenograft model. More recently her group in collaboration with the clinical team of Prof Jean-Lacau-St Guily and Prof Sophie Perié completed a successful autologous myoblast transplantation clinical trial for Oculo-pharyngeal muscular dystrophy. A successful preclinical gene therapy approach for OPMD has been validated in her team. Gillian Butler-Browne retired in august 2018 from being director of the newly created Centre of Research in Myology situated within the Myology Institut in the GH Pitié-Salpetrière and is now an emerit scientific director within the research centre.

Please describe your involvement in TREAT-NMD to date:
None

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee. I have worked together with many of the charitable organisations supporting neuromuscular disease throughout my career. I have an extensive knowledge of both basic science as well as neuromuscular disease physiopathology and different treatment approaches. I am very motivated to participate to the executive committee and since being retired have more time for such actions.

Nominee Name: Kristl Claeys

Organisation/Affiliation: University Hospitals Leuven (UZ Leuven) and University of Leuven (KU Leuven)

Please provide a summary of your neuromuscular career to date:
I am a neurologist (MD, PhD) specialized in neuromuscular disorders and working at the Department of Neurology at University Hospitals Leuven, Leuven, Belgium, where I am affiliated with the largest Neuromuscular Reference Centre (NMRC) in Belgium. I am appointed as associate professor at the Department of Neurosciences at KU Leuven. After graduating, I further specialized at the Institute of Myology at the Salpétrière Hospital in Paris, France, between 2007 and 2010. At the Institute of Myology, I was affiliated with the Neuromuscular Reference Centre and the Neuromuscular Morphology Unit. I obtained the Diploma of Myologist at the University Pierre et Marie Curie in Paris in 2008. Between 2010 and 2016, I worked as a neuromuscular specialist at the Department of Neurology and the Institute of Neuropathology at the RWTH University Hospital in Aachen, Germany. Since 2016, I am heading the Laboratory for Muscle Diseases and Neuropathies at the Department of Neurosciences at KU Leuven. My research topics are clinical research, genetics and translational medicine in neuromuscular disorders, with a focus on hereditary muscle diseases and neuropathies. I am an author on a large number of publications in international peer-reviewed journals and wrote several book chapters, on diverse topics in the field of neuromuscular disorders. I am currently Chair of the Belgian-Dutch Neuromuscular Study Group, Vice-Chair of the Belgian Neurological Society (BNS), Chair of the subgroup Neuromuscular diseases of the Flemish Network for Rare Diseases (VNZZ) and Board member of the European Reference Network for Rare NeuroMuscular Diseases (ERN Euro-NMD).

Please describe your involvement in TREAT-NMD to date:
I am a member of the TREAT-NMD Alliance Neuromuscular Network since 2012 and I am participating in The Care and Trial Site Registry (CTSR) of TREAT-NMD since 2013. In Belgium, I am actively involved in the Belgian Neuro-Muscular Disease Registry (BNMDR) in which clinical data from all neuromuscular patients included in the neuromuscular reference centres in Belgium are collected and particularly data on DMD and SMA patients are transferred to TREAT-NMD for further study. A further indirect link with TREAT-NMD is the fact that I am a Board member of the European Reference Network for rare neuromuscular diseases (ERN Euro-NMD) since 2016.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: Since I am a neurologist with specific expertise and many years of experience in neuromuscular diseases, and since I am working in a national clinical neuromuscular reference centre and in an academic setting, I have a very good view on both clinical care and needs of the patients, as well as on ongoing and planned clinical trials and translational research in neuromuscular disorders. I am an active member of several international and national societies that involve neuromuscular patients. In the past years, I have built a broad network of national and international collaborations, both for my clinical work as well as for my research. Since neuromuscular diseases are rare, it is of the utmost importance to collaborate with many colleagues in the field. My collaborations resulted in solving diagnoses in many patients and in a large number of important publications in the field of neuromuscular disorders. As an Academic Executive Committee member of TREAT-NMD, I would like to actively promote and contribute in advancing diagnosis, care and treatment for people with neuromuscular diseases all around the world. Furthermore, I will represent and promote TREAT-NMD at appropriate conferences and events, take responsibility for delivering certain areas of the TREAT-NMD Strategic Plan & Action Plan that are situated within my expertise, and provide advice, opinion and expertise on relevant issues.

Nominee Name: Paula Clemens

Organisation/Affiliation: University of Pittsburgh

Please provide a summary of your neuromuscular career to date:
I have 30 years experience as a physician-scientist in neuromuscular disease with a focus on the muscular dystrophies. I have performed translational research both at the bench and in the clinic. I am the medical director of the CINRG group that conducts multi-center muscular dystrophy clinical and translational research.

Please describe your involvement in TREAT-NMD to date:
CINRG has a collaboration with TREAT-NMD, by participating in the registries and through interactions at meetings. I have served as a TACT reviewer when needed.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee:
Extensive experience in global neuromuscular research that provides a perspective with a historical foundation and a deep working knowledge of neuromuscular research and clinical care.

Nominee Name: Annamaria De Luca

Organisation/Affiliation: Unit of Pharmacology, Department of Pharmacy and Drug Sciences, University of Bari Aldo Moro, Italy

Please provide a summary of your neuromuscular career to date:
My career has been devoted to pre-clinical research in neuromuscular disorders, mainly muscular dystrophies and inherited channelopathies. In this general frame, I also dedicated part of my research to other pathophysiological conditions of neuromuscular system, such as muscle atrophy and cachexia, muscle plasticity and aging, and the role of inflammation in muscle degeneration / repair in both acute and chronic conditions. Being a pharmacologist, the main aim of my research is to clarify pathogenic mechanisms that can represent druggable targets. Accordingly, a great part of my research has been focused on assessing the in vitro action of novel and repurposed compounds in modulating ion channel biophysics (in particular voltage gated chloride, sodium and calcium channels). The main aim was to identify molecules with greater tissue selectivity and then a better efficacy/safety ratio, or able to exert mutation-specific actions so to pave the way to personalized therapies of channelopathies. In parallel, I performed several studies for testing, both in vivo and ex vivo, the efficacy of drugs able to target disease-related molecular pathways by use of proper animal models of muscle disorders, with particular focus on Duchenne muscular dystrophy. I established one of the first core for pre-clinical assessment of pathology progression and drug efficacy using a wide array of in vivo and ex vivo tests on disease-related readouts parameters and had fruitful collaborations over time with both academic groups and private companies interested in developing orphan drugs for neuromuscular diseases.

Please describe your involvement in TREAT-NMD to date:
Since the early beginning of TREAT-NMD, I have been involved in all the activities related to implementation of pre-clinical studies in rare neuromuscular disorders. In fact, for more than 10 years now, I have been active in the TREAT-NMD work package aimed at establishing guidelines and standard operating procedures (SOPs) for getting robust preclinical data when assessing the natural history course and testing therapeutics in animal

models of muscular dystrophies. The key objective was and still is to enhance reliability and reproducibility of results so to boost bench to bedside transfer of results. In this frame, I have been actively participating to several meetings and workshops organized by TREATNMD, and in collaboration with other international networks. I have been directly involved in writing and upgrading SOPs that are published on TREAT-NMD website and also active in meetings involving multiple stakeholders, such as regulatory and funding agencies, patients advocacy groups and scientific journal editors in order to find a large consensus on best guidelines for translational research. As pre-clinical expert and pharmacologist I have also been involved in the TREAT-NMD Advisory Committee for Therapeutics (TACT) as reviewer since its institution in 2009; I then became part of the Core Committee and I have the privilege nowadays to be TACT Chair, being in office since November 2018.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: My candidature to the Executive Committee is motivated by my long involvement in the TREAT-NMD activities, mostly those related to reinforce the pre-clinical studies in translational research of rare neuromuscular conditions. Over the years, I had the chance to appreciate the multidisciplinary nature of TREAT-NMD and the efforts made to get the best from the exchange of different expertise. My key attribute of value to the Executive Committee would reside in my personal experience in the field that led me to acquire the attitude of conjugating basic pre-clinical scientific research, as academic pharmacologist, with the prioritization of best candidates for potential clinical use. I am well aware of every single step for the risky process of therapy discover in rare diseases and of the importance to support the independent and science-based thinking as a fundamental value to address the efforts of clinicians, patients associations and industries. I further implemented this attitude during my experience in the frame of TACT, which is indeed a model for providing a multidisciplinary and independent guidance to academic or private companies for de-risking projects of therapy development. I wish I can offer my personal experience to TREAT-NMD as a further bridge in the virtuous dialog between the various stakeholders in the pure interest of patients that urgently need effective and safe therapies. This goal requires a great sense of teamwork for the common cause, which has an immense value for the entire community.

Nominee Name: Jordi Díaz-Manera

Organisation/Affiliation: The John Walton Muscular Dystrophy Research Centre, University of Newcastle

Please provide a summary of your neuromuscular career to date:
I did my training in Neurology at the Hospital de la Santa Creu I Sant Pau (HSCSP) in Barcelona from 2001 to 2005. I have been involved in the follow-up of patients with neuromuscular disorders my whole career. During the first years, I worked in the Myasthenia Gravis (MG) Clinics, where we described the utility of rituximab as a treatment for drug-resistant patients. From 2008 to 2010, I did a predoctoral stay in Milan at the Stem Cell Research Institute of the San Rafaelle Hospital with Prof. Giulio Cossu. During that period, I expanded my knowledge of basic research in muscular dystrophies. From 2010 to 2020 I coordinated the Muscle Team at HSCSP where I was involved in several clinical and basic research projects. I have an especial interest in the application of muscle imaging to the study of neuromuscular diseases. In this sense, we have used semi-quantitative and quantitative muscle MRI sequences to study patients with different muscular disorders. I have collaborated with several Spanish patient associations, being part of their scientific committees or giving talks at their meetings. I have coordinated the Neuromuscular Diseases Study Group of the Spanish Society of Neurology and I have been part of the directive committee of the Catalan Society of Neurology. In both societies I have promoted the development of standard of care guidelines for patients with neuromuscular diseases. I have recently moved from Barcelona to Newcastle where I have been appointed as Professor of Neuromuscular Disorders at the University of Newcastle.

Please describe your involvement in TREAT-NMD to date:
I joined TREAT-NMD several years ago because I was interested in being part of the International Neuromuscular community. I think TREAT-NMD is a perfect platform to interact with other colleagues, patients association representatives and members of the medical industry. I fully agree with the main mission of TREAT-NMD which is to unite all these stakeholders and providing an infrastructure to accelerate research and therapy

development. I have attended some of the TREAT-NMD conferences and I have also participated as member of the faculty in some of the TREAT-NMD Masterclasses. I am the responsible for TREAT-NMD of the Spanish Neuromuscular registries. I am at present cochair of the Limb Girdle Muscle Dystrophy (LGMD) Task Force of the TREAT-NMD with Dr. Michela Guglieri. We have just started to organize the taskforce that will include several working groups such as registries, education, standards of care, outcome measures and developing countries.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee:
Personally, I consider TREAT_NMD an excellent platform for the different stakeholders involved in the follow-up and care of patients with neuromuscular diseases to work together with the common goal of improving the treatment and quality of life of patients with these diseases. In this sense, I feel fully qualified to be part of the Executive Committee, since I am a person who knows deeply the situation of patients with these diseases, the needs that the clinical teams have for the diagnosis and follow-up of these patients and the new opportunities that recent scientific developments offer us. I have a long experience in being part of the organising committees of scientific associations or patient associations. I consider myself to be an enthusiastic, responsible, hard-working person with a deep spirit of collaboration. I believe that the only way we can defeat these diseases is through initiatives such as TREAT-NMD that enable interaction between patients, clinicians and the pharmaceutical industry. I also believe that my role within the organizational structure of TREAT-NMD can be clearly linked to an educational role, in which I have long experience. I think it is very important that at this time TREAT-NMD plays a key role in raising awareness of the clinical features and diagnostic process of neuromuscular diseases in the medical community. I also have a clear idea of the responsibility that TREAT-NMD has as an entity that should help to develop certain areas of knowledge in developing countries.

Nominee Name: James Dowling

Organisation/affiliation: Hospital for Sick Children

Please provide a summary of your neuromuscular career to date:
I have been involved in clinical care and research related to neuromuscular disease for the majority of my career. After my MD, PhD at the University of Chicago (PhD with Elaine Fuchs in cell biology), I pursued a child neurology residency at Children's Hospital of Philadelphia, where I received mentorship from Richard Finkel and Carsten Bonnemann. I then did a neuromuscular medicine fellowship under the guidance of Eva Feldman at University of Michigan. After completing my training, I started at University of Michigan my independent research program and clinical practice, both of which were focused on paediatric neuromuscular disease. I then moved to Hospital of Sick Children, where I have been since 2013 and am currently a senior clinician-scientist. Clinically, I direct our neuromuscular program, including leading patient care, education, and clinical research. My area of special interest is congenital muscle disease, where I am a recognized expert in clinical care and treatment. In terms of research, I have an active research team focused on gene discovery and therapy development for congenital myopathies. My group has published extensively in terms of genetic causes of neuromuscular diseases, establishment and study of new models of disease, and identification and translation of therapies. Outside of SickKids, I have been very involved in the broader neuromuscular community. I am an Executive Board member of the World Muscle Society, and am co-organizer of the upcoming annual meeting. I serve as chair of the Canadian Paediatric Neuromuscular Group, and am a core member of TACT.

Please describe your involvement in TREAT-NMD to date:
My main involvement with TREAT NMD has come through my participation in TACT (TREAT NMD Advisory Committee for Therapeutics). I began as an ad hoc reviewer, both for pre-clinical and clinical assessment, and in 2018 became a standing member of the core committee. I also have been involved in the registry efforts of TREAT NMD. This has primarily come via my participation with the MTM registry. I am currently co-chair of the steering committee for the registry. I have been involved with the registry since its design and inception. Of note, I am also involved with registry efforts within Canada through the Canadian Neuromuscular Disease Registry. I also participate in several initiatives affiliated with TREAT NMD. One is connect4children. I am a content expert for neuromuscular diseases for this program. Another is NMD4C, a newly established Canada wide network

project that will affiliate with TREAT NMDs broader international programs. I am one of the co-PIs of this project.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee:
I believe there are several reasons why I would be a good candidate for the executive committee. Foremost is that I have experience and expertise across the broad spectrum of research and care for neuromuscular diseases. This experience spans basic, fundamental discovery science, where I have worked with various models including patient cells, C. elegans, zebrafish, and mice, and am familiar with mechanistic studies and multi-omics approaches. On the clinical side, I have led both observational and interventional clinical trials, and am involved in clinical care for the full breadth of neuromuscular disorders. Therefore, I feel that I bring a unique collection of comprehensive and complementary knowledge. Second, I have exposure to multiple layers of care, advocacy, and administration, having worked on initiatives including TACT, MTM registry, and as an advisor to organizations such as Muscular Dystrophy Association, Muscular Dystrophy Canada, and RYR1 Foundation. Third, I have demonstrated international leadership in the field, including as chair of the Canadian Paediatric Neuromuscular Group and as a member of the WMS Executive. Lastly, I would consider myself someone with high levels of energy and enthusiasm, and who is willing to apply my skills and knowledge to the team setting.

Nominee Name: Alessandra Ferlini

Organisation/Affiliation: UNIVERSITY HOSPITAL ST. ANNA, FERRARA, ITALY

Please provide a summary of your neuromuscular career to date:
I am (primary) a medical geneticists and (secondary) a neurologist. My job is either about clinics (genetic testing, genetic counselling and some experience in clinical trials for DMD boys based on antisense oligonucleotides) and on translational research. Main focus has always been on neuromuscular diseases. Research fields are gene discovery using omics, drug delivery by nanovechicles, biomarker discovery, RNA studies on DMD and Collagen VI myopathies to better understand the pathomechanisms of these diseases in terms of muscle impairment causes. I was fortunate to have funding for my research via European Union (7 funded projects, 2 as a Coordinator), and other foundations as Telethon and AFM. All these granted projects were related to neuromuscular diseases. I am deeply involved also in the diagnostics of these diseases and with my team in Ferrara we are heavily working on genetic definition of neuromuscular disorders, from DNA to RNA profiling. I believe that my dual engagement (diagnostic and research) has been very important in my career and has contributed to my enthusiasm in studying these rare disorders. My role is Associate Professor, Head of the Medical Genetics Unit at the University of Ferrara (Italy) but also Honorary Visting Professor at UCL, London.

Please describe your involvement in TREAT-NMD to date:
I was involved in the TREAT-NMD, and member of the PEC (Project Ethical Committee). I participated to many conferences contributing as a speaker or via poster presentations.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: The TREAT-NMD has already done an outstanding work for the neuromuscular community. I think that it might be important now to increase multidisciplinarity and networking activities with other rare diseases communities with countries that are not already involved in the TREAT-NMD. Important points in my view might be finding collaborative schemes with Industries and Funding Agencies (as for example European Research Council for outstanding

research applications, or others). And finally reinforce the liaison with European Reference Networks to facilitate their heavy work. Therefore, a candidate who is happy to put effort in these activities might be appropriate.

Nominee Name: Arnaldo José Godoy

Organisation/Affiliation: Sao Caetano do Sul University, Medical Course, Campus Sao Paulo, Brazil

Please provide a summary of your neuromuscular career to date:
In 2004, the mother of a Duchenne Muscular Dystrophy (DMD) patient, living in a city located in Sao Paulo State, Brazil, decided to create an Association of Parents and Friends of Muscular Dystrophy (MD) Patients. That Association asked the Director of the Medical Course where I worked if patients could be seen at the University. I was chosen to create and coordinate an Outpatient Clinic for MD Patients. A few years later more than one hundred DMD boys and more than thirty Becker MD patients had been seen for me, nutritionists, orthopaedist, cardiologist, physical therapist and dentist. In 2007 I was a fellow at the National Children's Hospital, in Washington D.C. at Prof. Eric Hoffman's lab. In 2009 I moved to Sao Paulo City to work at Sao Paulo University in the biggest MD Clinic of the country. In 2011 I became sub-investigator of the first clinical trial for DMD patients in Brazil (exon skipping). During the last few years I have seen MD patients at a Private Clinic and I have given several lectures to medical students about muscular dystrophies.

Please describe your involvement in TREAT-NMD to date:
In 2009, thanks to the father of a DMD patient, I went to Newcastle University to meet Dr. Volker Straub and Dr. Kate Bushby. In 2010, with the support of TREAT-NMD I organised an International Symposium in Sao Paulo City, with speakers from different regions of The United States and from several European countries. After that meeting Brazil was one of the chosen countries for a DMD clinical trial (exon skipping). Later, I visited Freiburg and Paris to attend TREAT-NMD courses. In 2019 I went to Amsterdam to attend the DMD Expert Masterclass. Nowadays I am helping TREAT-NMD staff to organise the 2020 DMD Expert Masterclass in Sao Paulo City, at the university where I work.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: I have worked as a university teacher, in different medical courses of my country, for the past 25 years. To teach is one of my passions. I love to create courses, to organise clinics and help students to do research and present their data at international meetings. I would be very happy working with other Committee members to promote training courses about muscular dystrophies and other neuromuscular disorders, for physicians, physical therapists and many health professionals. It would be wonderful to give support for those interested in establishing clinics for neuromuscular diseases' patients with international standards of care. I never get tired when performing educational tasks or seeing patients. I can help with the organisation of meetings and travel frequently to visit institutions willing to establish clinics or to give training courses.

Nominee Name: Erik K. Henricson, PhD, MPH

Organisation/Affiliation: Department of Physical Medicine & Rehabilitation and Clinical and Translational Research Center, University of California Davis

Please provide a summary of your neuromuscular career to date:
Dr. Henricson is an Assistant Professor and Associate Director for Clinical Research for the Neuromuscular Research Center at the UC Davis Department of Physical Medicine and Rehabilitation, and Director of the Regulatory Knowledge and Support section of the University's Clinical and Translational Science Center in Sacramento, California. He is ViceChair of the University's biomedical IRB, and Chair of the Nutritional and Behavioural IRB. Dr. Henricson is a founding member of the Cooperative International Neuromuscular Research Group (CINRG), and served as Co-PI or Co-investigator on many of the CINRG clinical trials conducted in the early 2000's, including the group's multicenter randomized study of daily vs. high-dose weekly Prednisone in boys with Duchenne muscular dystrophy. He is Co-Chair of the UC Davis/CINRG Duchenne Natural History Study (DNHS), and has collaborated widely with colleagues in academia and industry on use of DNHS data in areas including clinical endpoint development, biomarker discovery, person-reported outcomes and disease progression modelling. Recently, Dr. Henricson has been Co-PI of a series of clinical trials of (+)-epicatechin in patients with Duchenne and Becker muscular dystrophy. He is actively pursuing research in measurement of community mobility, including use of wearable devices, and development of the person-reported Duchenne Muscular Dystrophy Lifetime Mobility Scale (DMD-LMS).

Please describe your involvement in TREAT-NMD to date:
Dr. Henricson has participated in several TREAT-NMD-sponsored projects and events. At UC Davis, he has served as a co-investigator for the FOR-DMD steroid optimization trial. He has participated in TREAT-NMD workshops for upper limb outcome measure development, the Rasch analysis workshop for person-reported outcome measure development, and in workshops with the TREAT-NMD-associated Collaborative Trajectory Analysis Project (C-TAP) group. His work with the CINRG Duchenne Natural History Study has been shared at

numerous TREAT-NMD and used in the preparation of best-practice white papers and practice guidelines.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: Dr. Henricson has a long history of multidisciplinary team science collaboration with experts from clinical, basic, and measurement science disciplines represented in the neuromuscular disease community. He brings an appreciation and understanding of the need to link quantitative laboratory and clinical measures of disease with community-based and person reported outcomes in order to describe the impact of therapeutic interventions on day-today wellness and life satisfaction of patients and their families. In addition, as an IRB Chair he has extensive experience in scientific, regulatory and ethical review and design of clinical research projects in a variety of adult and paediatric patient populations.

Nominee Name: Anri Human

Organisation/Affiliation: Sefako Makgatho Health Sciences University

Please provide a summary of your neuromuscular career to date:
I have been a qualified physiotherapist for 16 years, however my journey with NMD only began about eight years ago when I first met a real-life hero, a teenager living with Duchenne. At this stage, I already completed my Master's degree on bronchiolitis in infants, but due to the zeal of a senior physiotherapist and the courage of this particular teenager, I was inspired to become involved in children with NMD. As an academic, I attempted to find clinically relevant solutions through extensive reading, asking questions and performing research. I am currently finalising my PhD thesis titled: "The effect of inspiratory muscle training on clinical outcomes and health-related quality of life in children with neuromuscular disease and respiratory muscle weakness." Over the past few years I have learned and grown immensely from my interaction with patients and their families. I have witnessed tremendous courage, dedication and commitment and I truly believe that together we can find a cure, but until we do, we can improve the quality of life of all children across the world!

Please describe your involvement in TREAT-NMD to date:
My involvement with TREAT-NMD started when I presented at the ICNMD congress in Austria during 2018. I met two lovely ladies (Anne Oyewole and Joanne Lee) at the TREATNMD stand who provided valuable information and encouraged me to become a member of TREAT-NMD. I signed up for the newsletter and applied to become part of the scientific committee that was involved in organising the TREAT-NMD congress hosted in Leiden during 2019. As a member of this scientific committee, I had the privilege to work alongside numerous experts in the field of NMD as well as present and chair the session on adults with progressive NMD. In addition, I was privileged to attend the Duchenne expert masterclass hosted in the Netherlands (Amsterdam, 2019), which was an unforgettable experience (my reflection has been included in the February issue of the TREAT-NMD newsletter). Lastly, I am also part of and coordinating a project to translate the SMA International Standards of Care guidelines from English into Afrikaans, for use in the South African context.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: I might have less experience in NMD compared to others that have served on this Executive

committee, but I trust my enthusiasm can compensate for lack of seniority I have had exposure in various fields of physiotherapy, clinically and within academia, and have been working as a full-time lecturer since 2008. I completed a postgraduate certificate in higher education (cum laude) in 2011 and have a passion for all components of higher education: teaching and learning, research and service-learning (community engagement). Over the past 12 years, I presented at numerous local and international congresses/workshops and supervised research projects for undergraduate as well as postgraduate physiotherapy students. My awards include best poster prizes at the WCPT international congress; SMU Research Day (2017); ICNMD in Austria (2018); winner of the 3MT competition at UCT (2017) as well as national 3MT winner and People's choice awards (2018). I serve on various committees within the South African Society of Physiotherapy, the International Confederation of Cardiorespiratory Physical therapists, Muscular Dystrophy Foundation of South Africa (Gauteng branch) and am a director of the Myalgic Encephalomyelitis Chronic Fatigue Syndrome Association (SA). Particularly, over the past five years, I have been involved in social responsibility and community projects to raise awareness about NMD. In addition, I hope to take hands with my colleagues and be part of a collaborative effort between Africa and Europe, to share ideas, experiences and expertise in order to improve the lives of all our patients.

Nominee Name: Linda P Lowes

Organisation/Affiliation: Center for Gene Therapy at Nationwide Children's Hospital; Associate Professor in Pediatrics, The Ohio State University

Please provide a summary of your neuromuscular career to date:
As a principal investigator specializing in outcome measures research at the Center for Gene Therapy at Nationwide Children's hospital, I have had the privilege to work on many first in human trials such as the spinal muscular atrophy trial that led to the FDA approval of Zolgensma. As part of this team I have contributed to protocol development, data collection and interpretation, manuscript preparation and presentation. I have also worked on several industry trials and provide outcome measures consultation and training for numerous pharmaceutical companies. Part of this work involves traveling to sites around the world to establish inter-rater reliability with other clinical evaluators as I have been designated as one of the "gold standard" evaluators. This has allowed me to meet colleagues in over 15 countries and evaluate patients together to document our reliability. These interactions have given me insight into successes and issues facing our field. In addition to research, I provide clinical care to patients with diverse diagnoses through our neuromuscular disorders clinics. Although based at Nationwide Children's Hospital, this includes caring for both adults and children.

Please describe your involvement in TREAT-NMD to date:
I have had the opportunity to work with TREAT-NMD in several ways. I have served as a reviewer in the TACT program, lectured at a TREAT-NMD masterclass and most recently served on the planning committee for the TREAT-NMD conference. I also serve as a member of the limb girdle muscular dystrophy advisory board.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: I believe my background as a physical therapist could bring a unique perspective to the

TREAT-NMD executive committee. I addition to serving as a principal investigator on several multi-site natural history studies, I provide training to other clinical evaluators world-wide. My focus on outcome measures and natural history studies makes me uniquely qualified to assist with one of TREAT-NMD main initiatives; developing collaborative registries. Nationwide Children's hospital sees a diverse patient population of adults and children which has allowed me to interact with numerous other TREAT-NMD members. I believe I would be able to represent the wide range of interests and specialties that comprise the organization. As a member of the Center for Gene therapy I have also had the opportunity to interact with scientists working in all phases of research, including basic scientists, regulatory specialists, clinical trial investigators and industry. I look forward to the opportunity to contribute to the success and growth of this important organization.

Nominee Name: Pradeep P.A. Mammen, MD

Organisation/Affiliation: UT Southwestern Medical Center

Please provide a summary of your neuromuscular career to date:
I am a clinician-scientist with clinical expertise in advanced heart failure, ventricular assist devices and heart transplantation. Secondary to additional training I received in molecular cardiology, I have developed a unique interest as well as expertise in the care of patients who develop a genetic form of cardiomyopathy (esp. neuromuscular-associated cardiomyopathies). In fact in 2010, I became the Founding Medical Director of the UT Southwestern Neuromuscular Cardiomyopathy Clinic, which has become one of the largest adult clinics in the world and currently actively follows 700 patients to date. We are utilizing this clinic as a platform for translational studies focused on novel therapies directed towards muscular dystrophy patients. These studies are aimed at improving both the overall and cardiovascular care we provide to this unique patient population. In addition to a very active clinical practice I also run a molecular laboratory dedicated to muscular dystrophy, which is funded by the NIH and various pharmaceutical companies. In fact in the Fall of 2015 the NIH awarded UT Southwestern one of six Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Centers in the country, for which Dr. Eric Olson, Chair of the UT Southwestern Department of Molecular Biology, and I are the Co-Directors of this Center. The core mission of the Center is to rapidly develop genome editing into a viable and effective therapy for patients with Duchenne muscular dystrophy. Since 2017, I have co-authored with Dr. Olson or been the senior author on 8 papers related to muscular dystrophy.

Please describe your involvement in TREAT-NMD to date:
Although I have attended TREAT-NMD meetings in the past, I have not been involved to date in the leadership of the organization per se. With that stated, I am very involved in various activities related to a variety of muscular dystrophies. In fact, I played an important role in a recently published consensus paper (published in the Journal of the American Heart

Association February of 2020) related to the cardiovascular care of myotonic dystrophy patients. I am often asked to speak on the cardiovascular issues facing many patients with neuromuscular disorders, in particular by organizations like PPMD and CureDuchenne. Finally, for the past four years as the Co-Director of the UT Southwestern Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center, I have organized and chaired the Annual UT Southwestern Wellstone Duchenne/Becker Muscular Dystrophy Clinical Symposium, which attracts 100-120 participates each year. This Clinical Symposium provides state-of-the-art research updates and well as standard of care practices for Duchenne and Becker muscular dystrophy patients.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: I believe I bring a very unique perspective to the table when it comes to the care of muscular dystrophy patients. Not only do I run one of the largest Adult Neuromuscular Cardiomyopathy Clinics in the United States dedicated to the cardiovascular needs of muscular dystrophy patients (700 patients and growing), I am also actively involved in novel research related to muscular dystrophy. As the Co-Director of the UT Southwestern Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center (MDCRC), I not only have my finger on the pulse of muscular dystrophy patients, but I also serve a significant role in the scientific investigation involving therapies for muscular dystrophy. My work positions me perfectly to succeed at functionally integrating the many individuals, including physicians, nurses, scientists and support staff, required to manage a complex entity like the UT Southwestern Wellstone MDCRC.

Nominee Name: Kanneboyina Nagaraju

Organisation/Affiliation: School of Pharmacy and Pharmaceutical Sciences, Binghamton University-SUNY

Please provide a summary of your neuromuscular career to date:
I am an immunologist with specific expertise in skeletal muscle inflammation. My research focuses on understanding how skeletal muscle contributes to the initiation and perpetuation of inflammation in autoimmune and genetic neuromuscular diseases. I extensively used mouse models to delineate the pathological mechanisms underlying muscle damage and weakness in inflammatory muscle diseases. Series of experiments done in our group for the last decade have identified several non-immune mechanism, as well as innate immune mechanisms controlling metabolism in skeletal muscle. I have co-authored several textbook chapters on autoimmune muscle diseases and has over 150-refereed publications in neuromuscular diseases. I also led international efforts to define reliable and sensitive tests for drug efficacy in mouse models of neuromuscular diseases and currently directs a robust basic and translational research program for neuromuscular disorders.

Please describe your involvement in TREAT-NMD to date:
I have closely interacted with TREAT-NMD since its inception. I collaborated with TREATNMD to setup standard operating procedures of preclinical assessments in neuromuscular disease models. I also closely worked to jump start TACT committee. I served as preclinical core member on TACT committee and continue to serve the TACT committee as an extended committee member. I also involved in organizing mock TACT reviews in collaboration with TACT secretariat.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: I am well suited to serve on the executive committee because one of my academic goals not only do basic and translational research but also to setup teaching and research infrastructure

for NMD research and patients community. In this aspect my goals are closely aligned to TREAT-NMD's mission to advance treatments and care for patients with rare NMDs. In addition, I have developed close working relation with many neuromuscular disease stake holders in academic, industry and patient advocacy organizations. I have worked closely with TREAT-NMD leaders as well as members of the secretariat to further TREAT-NMD mission.

Nominee Name: Terence Partridge

Organisation/Affiliation: Genetic Medicine Center, Children's National Medical Center, Washington DC, USA

Please provide a summary of your neuromuscular career to date:
I have worked on muscular dystrophy research since 1970, and was instrumental in development and validation of the concept of myoblast transplantation and more recently in the demonstration of the applicability of exon skipping for DMD.

Please describe your involvement in TREAT-NMD to date:
I joined TREAT-NMD only a few years ago and have become increasingly impatient with the quality of the preclinical research that is being translated into clinical trials on the basis of insufficiently thoroughly validated data. I wish to work to increase pressure on this aspect of research which is gathering an increasingly bad reputation.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee. I have a long record of moving observational research into reproducible quantitative measures and would work to encourage and disseminate this approach more widely within the neuromuscular research community, in the hope that this will reduce the frequency of disappointing and dead-end preclinical research projects and aid their application to clinical trials.

Nominee Name: Antonio Toscano

Organisation/Affiliation: Neurology and Neuromuscular Diseases Unit, University of Messina, Italy

Please provide a summary of your neuromuscular career to date:
Antonio Toscano, M.D., born 16/12/1956 in Messina, Italy. Since 2009, Full Professor of Neurology, University of Messina, Italy. 1986-1987 Postdoctoral fellow, University of London, UK at "The National Hospital for Nervous Diseases", London, working with Dr. J. A Morgan-Hughes and with J.B. Clark on "Mitochondrial disorders". European Neurological society's involvement: Chairman and speaker for several years for the ENS Neuromuscular Teaching Courses. 2014-2018 Member of EAN SC. 2016-2020 Chairman of EAN panel on Muscle disorders. Since 2016 Director of the ERN-NMD Center for Rare Neuromuscular Diseases of Messina. Since 2017 Chairman of the organization of the "Italian Neuromuscular Day" (contemporarily in 16 Italian cities).Since 2018, Dean of the Faculty of Medicine of the University of Messina. Member of scientific Italian and International Societies (SIN, AIM, ASNP, ENS, EAN, WMS, EPOC, Solane). 2009-2012 President Italian Association Myology (AIM).Since 2009 Member World Pompe Global Advisory Board. Since 2011 Member TREAT-NMD Task Force. 2014-2018 Member European Academy of Neurology - EAN SC. Since 2015 Member European Pompe Consortium (EPOC) Advisory Board. Since 2019 Member EAN Programme Committee. Member Editorial Boards of Neuromuscular disorders, Journal of Neuromuscular diseases, Neurological Sciences. Prof. Toscano scientific activity was mainly developed in neuromuscular disorders, especially for Metabolic myopathies as muscle glycogenoses (particularly Pompe disease), lipid storage and mitochondrial myopathies, Genetic myopathies (limb-girdle myopathies, channelopathies, cardiomyopathies, congenital myasthenias), Peripheral neuropathies (disimmune and genetic neuropathies). He is author of over 220 publications in indexed journals in the neuromuscular diseases field

Please describe your involvement in TREAT-NMD to date:
In 2011, I have been involved in the TREAT-NMD task force. Because of this, I have participated to some European meetings organized by TREAT-NMD. I have also sustained the manifestations organized by TREAT-NMD in Italy.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee:
I think that I may contribute to TREAT-NMD objectives because, since at least 30 years, I have accumulated a quite relevant experience in the neuromuscular diseases (NMDs) field, also been in touch with the main Neuromuscular groups in every part of the world, attending meetings and often presenting new data on different kinds of neuromuscular disorders, giving talks, proposing and participating to several educational activities, collaborating for new developments in the neuromuscular fields in terms of European and extra-European initiatives in favour of patients with NMDs, promoting and participating to several clinical and therapeutic trials. I have also organized several International and National meetings of great success and I suppose that this is also important for the TREAT-NMD organization. Because of this large experience, matured in the very specific field of NMDs and also in the neurological rare disorders, I suppose that I could be able to represent TREAT-NMD in dedicated National and International events. Key attributes: large experience in the NMDs field, member and/or leader of several scientific societies, very good knowledge of National and International groups involved in the field, long educational practice, invited speakers in the main countries of the world, continuous research work with relevant results and publications, joint activities with patients associations

Nominee Name: Arun Shastry

Organisation/Affiliation: Dystrophy Annihilation Research Trust

Please provide a summary of your neuromuscular career to date:
Dr.Arun Shastry completed his PhD on the topic "Immunogenetic markers in autoimmune diabetes". He was awarded Guest Research Scholarship from Swedish Institute to do research In Karolinska Institute Sweden (2004-2006). He obtained the CSIR Senior Research Fellowship award (2008-2010). Dr. Arun Shastry joined Dystrophy Annihilation Research Trust, Bangalore to set up India's first Research and Development facility dedicated in finding a cure for Duchenne Muscular Dystrophy (DMD) in 2013. He has established the in vitro pre-clinical research department by creating Cell Lines of Duchenne Muscular Dystrophy patients. In 2017, Hanugen Therpaeutics Private Limited was started which specialized in customized Antisense Oligonucleotide (AON's) Synthesis. Hanugen has an inhouse Oligosynthesis facility and Organic Chemistry lab. The lab has the ability to perform oligo-synthesis and in vitro testing to show proof of concept of exon skipping. They have obtained similar results in concordance with principals from Murdoch University, Australia and Leiden University medical Center, Netherlands. The team is setting up in-vivo platforms for studying long-term efficacy and toxicity of AON's in its potential as a therapy for DMD patients. Dr. Arun recently led the first investigator-led clinical study on Exon Skipping in the country which to the best of our knowledge is also the first double-exon skipping in the world.

P**lease describe your involvement in TREAT-NMD to date:**
Dr. Arun has been very active in dispensing information about the work done by TREATNMD with families who have been affected by Neuromuscular Disorders in India. He has taken part in the summer translation school organized by Dr. Andoni in Paris and when he returned to our lab he ensured that all of us researchers were up to date on the latest techniques and innovations in physiotherapy and disease management.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: I believe Dr. Arun Shastry will be an excellent candidate for Executive Committee because of his long and illustrious involvement with DMD. He is also the Chief Scientific Officer of the

premier lab in India which is doing path-breaking research in to genetic research. India has one of the (if not THE) largest cohorts of Neuromuscular Disorder patients in the World, with the number of children suffering from DMD alone outnumbering many other countries clubbed together. He will be an excellent representative of this immense group and with his vast experience and contacts he will be able to give invaluable advice and a whole different slant of thinking to TREAT-NMD.

Nominee Name: Gabriele Siciliano

Organisation/Affiliation: Department of Clinical and Experimental Medicine, University of Pisa

Please provide a summary of your neuromuscular career to date:
Since 1993 he is responsible for the Unit of Neuromuscular Diseases and for the Laboratory of Molecular Diagnosis in the former Department of Neuroscience, now Department of Clinical and Experimental Medicine - Neurological Clinics - University of Pisa, with main interests in clinical, laboratory and molecular aspects of acquired and neurogenic disorders of muscle diseases, conducted, since 1986, in collaboration with national and international scientific partners. In particular, he has studied several issues on muscle fatigue and exercise in normal and myopathic subjects, motorneuron output evaluation and subsarcolemmal excitability in myotonic syndromes and muscular dystrophies, metabolic performance in glycogen storage diseases and mitochondrial myopathies, peripheral neuropathies and motor neurone diseases. Also he has been engaged in epidemiological and therapeutic studies, epidemiological surveys, as well as studies on the diagnostic usefulness of various laboratory investigation procedures and validation of clinical assessment scales. More recently, Prof. Gabriele Siciliano has been largely involved in studies in the fields of investigation on molecular pathways in the pathophysiology of the neuromuscular disorders, as well as searching for their phenotypic determinants and reliable biomarkers suitable in defining natural history of the illness and designing proper outcome measures for clinical trials. All these activities have led to over 300 full scientific papers, dealing with the above indicated arguments, funded projects from Institutional and Charity bodies, responsibility in research evaluation committees for public and private funding research bodies, participation to national and international clinical trials performed according to GCP (last GCP training certificate obtained in 23 January 2020).

Please describe your involvement in TREAT-NMD to date:
Prof. Gabriele Siciliano has been involved in clinical and laboratory activities, in the field of neuromuscular disorders, aimed to exchange experience and data with other partners within TREAT-NMD consortium. These activities, conducted in collaboration with national and international scientific partners, have been aimed to harmonize protocols of clinical assessment of neuromuscular patients, especially in the field of muscle fatigue and exercise

intolerance investigation, get epidemiological data, evaluate the diagnostic usefulness of some diagnostic investigations and define molecular pathways in the pathophysiology of these diseases, as well as to search for their phenotypic determinants and reliable biomarkers suitable in defining natural history of the illness and designing proper outcome measures for clinical trials. All these activities have been conducted within a shared view of the matter with the most accredited scientific societies in the field, among which the World Muscle Society, European Academy of Neurology and Associazione Italiana di Miologia, and patients organization, leading to a scientific production of more than 300 full papers on this topic, in specialized cited international journals, under the responsibility of Patients organization, Institutional and Charity bodies, and participation to research evaluation committees for public and private funding research bodies.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: Long career activity and experience in the field of clinical and laboratory issues within the neuromuscular diseases, strong commitment to work in team collaboration, confidence in multidisciplinary collaboration to increase knowledge in the field, faith in translational approach to get relevant insights in the pathophysiology of these disease to improve the efficacy of any treatment, working within the Scientific Community, Institutional bodies and deputies to increase the possibility to care and cure.

Nominee Name: H Lee Sweeney

Organisation/Affiliation: University of Florida College Medicine

Please provide a summary of your neuromuscular career to date:
H. Lee Sweeney, Ph.D., is the Thomas H. Maren endowed Professor of Pharmacology & Therapeutics at the University of Florida, and Director of the UF Myology Institute. Dr. Sweeney earned a bachelor's degree in biochemistry at the Massachusetts Institute of Technology, and received a master's degree in physiology and a doctorate in physiology and biophysics from Harvard University. He moved to Southwestern Medical School in Dallas for his postdoctoral work, before joining the faculty at the University of Texas at Austin as an Assistant Professor. He moved from Austin to the University of Pennsylvania where he served as Chairman of the Department of Physiology at the University of Pennsylvania School of Medicine from 1999-2013. Much of Dr. Sweeney's research program is translational in focus, and has produced highly cited research on inherited forms of cardiovascular disease, and on the skeletal and cardiac aspects of muscular dystrophy. He was elected as a Fellow of the American Heart Association in 2001 and a fellow of the Biophysical Society in 2018. He has been Director of a Paul Wellstone Muscular Dystrophy Cooperative Center first at Penn and currently at UF since 2005. Dr. Sweeney is actively developing therapeutics for rare diseases that include both small molecule and gene therapy approaches. He serves as a scientific advisor to Parent Project Muscular Dystrophy and as a consultant to a number of industry therapeutic development efforts for Duchenne muscular dystrophy and Spinal Muscular Atrophy.

Please describe your involvement in TREAT-NMD to date:
I have served multiple times as a TACT reviewer and have participated in TREAT-NMD meetings.

Why do you think you would be a good candidate for the Executive Committee? Please also include key attributes you feel would be of value to the Executive Committee: I think that my experience, my role in organizing US meetings, as well as my being well informed about many of the therapeutic developments ongoing in the US would bring valuable insights to the TREAT-NMD Executive Committee. I would also like to become

involved with TACT again, as it has been a number of years since I served as a reviewer for them.
