ENMC MENTORS

Prof. Annemieke Aartsma-Rus | Leiden, The Netherlands

PROF. ANNEMIEKE AARTSMA-RUS is a Professor of Translational Genetics at the Department of Human Genetics of the Leiden University Medical Center. She played an important role in the development of the antisense mediated exon skipping therapy for Duchenne muscular dystrophy during her PhD research (2000-2004) at the Leiden University Medical Center (the Netherlands).
As of December 2007 she became leader of the "DMD exon skipgroup". Since 2013, she has a visiting professorship at the Institute of Genetic Medicine of Newcastle University (UK). She was President of the Oligonucleotide Therapeutics Society (2019-2021), vice-chair of COST Action "Delivery of antisense RNA therapies" and member of the Core Committee of the TREAT-NMD Advisory board for Therapeutics (TACT).
Thus far, she has published over 230 peer-reviewed papers and 12 book chapters, as well as 15 patents and has edited one book.
She has given many invited lectures at meetings, symposia and workshops as well as patient/parent organizations meetings, where she is known for her ability to present science in a clear and understandable way.
Annemieke has successfully applied for numerous grant applications and serves on many editorial and advisory boards. She collaborates with academia and industry.

Dr Alan Beggs | Boston, USA

DR ALAN H. BEGGS PhD is the Director of the Manton Center for Orphan Disease Research at Boston Children's Hospital and Sir Edwin & Lady Manton Professor of Pediatrics at Harvard Medical School. Following undergraduate studies at Cornell University, Dr Beggs obtained his PhD in Human Genetics at Johns Hopkins University, with subsequent post-doctoral fellowship training in medical and molecular genetics at Johns Hopkins and Boston Children's hospitals. He has general expertise in laboratory and clinical applications of genetics to human disease, and since 1992 has directed an independent research program in the Division of Genetics and Genomics. Over the years, he has used the toolset of human molecular genetics to study normal biology and pathophysiology of a variety of disorders including muscular dystrophies, cardiac arrhythmias, developmental brainstem defects, hereditary anemias, sudden infant death syndrome, and congenital myopathies. Dr Beggs has been a standing and ad hoc member of numerous NIH study sections and grant reviewer for the Muscular Dystrophy Association and March of Dimes. He is a member of several scientific advisory boards and boards of directors for nonprofit and commercial entities.

Prof. Olivier Benveniste | Paris, France

PROF. OLIVIER BENVENISTE has the great opportunity to control all the steps of the translational medicine process, from the immediate identification of myositis patients during their clinics or their hospitalization in his national reference centre for myositis, the in depth characterization of the patient' phenotype in a standardized database, their sampling and bio banking (muscle, PBMC, sera), fundamental researches (in his lab at Sorbonne University) from this biobank (in depth immunoprofiling of PBMC by CyTOF analyses, effect on muscle of myositis specific auto-antibodies, effect on muscle of type 1 interferons, research of new biomarker), the definition of the best outcome measures for clinical trials, the physiopathological "bench to the bedside" studies, the development of clinical academic and/or industrial clinical trials, as for example, the RAPAMI trial testing rapamycin (sirolimus) against placebo in inclusion body myositis.

Dr Carsten Bönnemann | Memphis, USA

DR CARSTEN BÖNNEMANN received his MD from Freiburg University, Germany. He completed pediatric training in Germany.
A residency in pediatric neurology at Harvard was followed by postdoctoral work with Dr Louis Kunkel at Children's Hospital Boston working on the molecular genetics of muscular dystrophy. In 2002 he joined the Children's Hospital of Philadelphia/University of Pennsylvania as Assistant Professor, and became Co-Director of the Neuromuscular Programme and Director of the Neurogenetics Clinic. He joined the National Institute of Neurological Disorders and Stroke (NINDS/NIH) in 2010 as Senior Investigator and Chief of the Neuromuscular and Neurogenetic Disorders of Childhood Section.
Currently Dr Bönnemann is the Chair of the Department of Genomic and Translational Neuroscience at the St. Jude Children's Research Hospital in Memphis. Research in Dr Bönnemann's laboratory revolves around molecular mechanisms underlying early onset muscle disease (congenital muscular dystrophies, congenital myopathies, and reducing body myopathy). Dr Bönnemann is involved in manyneuromuscular networks, such as the WMS,TREAT-NMD, ENMC and has an impressive track record of scientific publications, lectorates and editorial work making progress in the research of neuromuscular patients.

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Dr Ana Buj-Bello | Paris, France

DR ANA BUJ-BELLO is Research Director at INSERM and head of the Integrare academic unit at Genethon, France. She received her degree in Medicine and Surgery from the University of Lleida, Spain, and a PhD in Neurosciences from the University of St Andrews, UK.
She also obtained a diploma in Myology from the University of Pierre et Marie Curie, France, and an EMBA at HEC Paris. Her research activities focus on developing AAV-based gene therapies for neuromuscular disorders, in particular congenital myopathies, with a major interest in clinical translation.
Her pioneering work on gene replacement therapy for myotubular myopathy led to the initiation of the first clinical trial in patients.
She received the Outstanding New Investigation award of the American Society of Gene and Cell Therapy (ASGCT) in 2015 and the Leon Baratz, Docteur Darolles award of the French National Academy of Medicine in 2024. She is involved in several research networks and professional associations, and served as a member of the ENMC Research Committee from 2020 to 2026.

Prof. James Dowling | Philadelphia, USA

PROF. JAMES DOWLING is a clinician-scientist focused on gene discovery and therapy development for childhood muscle diseases. Prof. Dowling received his B.Sc. and M.Sc. from Yale University and his MD/PhD from the University of Chicago. He did his residency in child neurology at Children's Hospital of Philadelphia and completed postdoctoral research at the University of Pennsylvania and Michigan. Before coming to Toronto, he was an assistant professor at the University of Michigan from 2009-2013. He worked at SickKids and became professor at the University of Toronto in 2020. Currently Prof. Dowling is the Director of the Penn Neurogenetics Therapy Center at Penn Medicine.
Jim Dowling's clinical expertise is in childhood neuromuscular disorders and he is considered one of the leading authorities on the diagnosis and management of congenital myopathies. His research examines questions of disease pathogenesis and therapy development for congenital myopathies and childhood muscular dystrophies. He has authored or co-authored more than 100 peer reviewed manuscripts and been fortunate to enjoy funding from several sources, including CIHR, NIH, MDA, and Genome Canada.

Prof. Ana Ferreiro | Paris, France

PROF. ANA FERREIRO is a Neuromuscular Neurologist and a clinical and translational research scientist. She is currently a Professor of Neurology at Sorbonne University, Head of the Neuromyology Department of the Pitié-Salpêtrière University Hospital, and responsible for an experimental research laboratory at the Myology Research Center (Institute of Myology), all in Paris. For almost 30 years, she has combined clinical care and research with basic, translational and therapeutic research on muscle diseases, with a particularly focus on congenital myopathies and desmin-related myopathies. Her work led to the phenotypical and molecular description of several entities (i.e. congenital titinopathies, recessive RYR1-related myopathies and desminopathies, SELENON-related myopathy) and to the identification of pathophysiological pathways, biomarkers and potentially therapeutic drugs, and was at the origin of the first clinical trials in congenital myopathies. Prof. Ferreiro was for 6 years (2019-2024) the Research Director of the European Neuromuscular Center (ENMC), served as an Executive Associate Editor for Neuromuscular Disorders and is currently a member of the Editorial board for several journals in the field.

Sarah Foye | Pine Brook, NJ, USA

SARAH FOYE, BS, is an occupational therapist and rare neuromuscular disease advocate, and the mother of an adult son with congenital titinopathy. She is the founder and president of Team Titin, Inc., a nonprofit organization serving individuals and families affected by titin (TTN)–related muscle and heart disorders. She considers her most valuable credentials to be "M.O.M."
Sarah serves on the Advisory Team of the Congenital Muscle Disease International Registry (CMDIR) and has been a patient representative at ENMC workshops, including the 219th ENMC International Workshop on Titinopathies and the 277th ENMC Workshop on congenital myopathies and diagnostic guidelines. She has contributed to multiple international scientific and patient-engagement initiatives, including SciFam conferences, PCORI engagement projects, and U.S. Department of Defense research review panels.
Her clinical background includes inpatient rehabilitation and pediatric occupational therapy, along with training in clinical medical ethics through the Rehabilitation Institute of Chicago and the University of Chicago. She is also an active legislative advocate through programs such as Rare Disease Legislative Advocates, the MDA Advocacy Institute, and the EveryLife Foundation Community Congress.
Sarah brings to mentoring a unique perspective that bridges families, clinicians, researchers, and policymakers. She is deeply committed to empowering the neuromuscular community through collaboration, advocacy, and compassionate leadership.

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Prof. Cynthia Gagnon | Montreal, Canada

PROF. CYNTHIA GAGNON is a senior career-award researcher specializing in adult genetic neuromuscular disorders. She holds a professorial appointment at the School of Rehabilitation at the University of Sherbrooke. She is the scientific director of the Groupe de recherche interdisciplinaire sur les maladies neuromusculaires (GRIMN) and is a researcher at the Centre de recherche Charles-Le Moyne-Saguenay-Lac-St- Jean sur les innovations en santé. She trained as an occupational therapist at McGill University. She has a doctoral degree in experimental medicine from Laval University and pursued a postdoctoral fellowship in program evaluation at Montreal University. Her work aims at improving clinical care and speeding up trial readiness in the most prevalent neuromuscular diseases in Canada. Her main interest is to document the natural history of the disease through an interdisciplinary perspective to be able to document the progression of the disease and to identify significant predictor and explanatory factors related to participation in daily activities and social roles of patients such as work and autonomous living. Her other interest is to define the best outcome measures to assess potential therapeutic targets such as muscle strength, fatigue or cognitive functions. She also works on developing knowledge translation strategies related to rare diseases to ensure effective and just-in-time knowledge translation to the interdisciplinary team through different strategies including wiki, articles, clinical practice guidelines to improve clinical care for patients and their families. She is involved in several international projects in relation to myotonic dystrophy type 1 (DM1), oculopharyngeal muscular dystrophy (OPMD) and autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS).

Prof. Heinz Jungbluth | London, United Kingdom

PROF. HEINZ JUNGBLUTH is Professor of Paediatric Neurology at King's College London and Consultant Paediatric Neurologist at the Evelina Children's Hospital, Guy's & St Thomas' NHS Foundation Trust, London, UK, with more than 25 years clinical experience in Paediatric Neurology. His main research interest is in early-onset neuromuscular and neurodevelopmental disorders. He has been particularly interested in the clinical and genetic characterization of the congenital myopathies, in particular those affecting excitation-contraction coupling due to mutations in the skeletal muscle ryanodine receptor (RYR1) gene, and related episodic disorders such as malignant hyperthermia (MH) and (exertional) rhabdomyolysis (ERM). Other neuromuscular interests include congenital myasthenic syndrome, juvenile myasthenia gravis as well as muscle magnetic resonance imaging. He and his team have also introduced the concept of congenital disorders of autophagy, a novel class of inborn neuromuscular and neurometabolic conditions linking aberrant neurodevelopment with common adult-onset neurodegenerative disorders such as dementia, Parkinson's disease and ALS.
He has published widely and is a member of national and international expert consortia concerned with improving the care and developing therapies for early-onset neuromuscular and neurological disease.

Prof. Eric P. Hoffman | New York, USA

PROF. ERIC P. HOFFMAN is Professor of Pharmaceutical Sciences and Associate Dean for Research, Binghamton University – SUNY, and has co-founded and holds management positions in three academic spin-off companies focused on neuromuscular disease (CEO of ReveraGen BioPharma; Vice President of AGADA BioSciences; board member of TRiNDS LLC). Prof. Hoffman received his PhD in Drosophila molecular genetics from Johns Hopkins University and transitioned to human molecular genetics as post-doctoral fellow with Louis Kunkel at Boston Children's Hospital and Harvard Medical School working on the identification of the Duchenne muscular dystrophy gene and dystrophin protein.
He has held faculty positions at Harvard Medical School (1988-1990), University of Pittsburgh (1990-1998), George Washington University and Children's National Medical Center (1998-2016). He co-founded the Cooperative International Neuromuscular Research Group (CINRG) and has helped lead drug development programs of viltolarsen (exon skipping), and vamorolone. He is an inventor on over 20 patents and has authored over 500 publications.

Prof. Cornelia Kornblum | Berlin, Germany

PROF. CORNELIA KORNBLUM is an associate professor of neurology at the Center for Neurology, University Hospital Bonn. She headed the Department of Neuromuscular Diseases until December 2025. Since January 2026, she has run the Department of Neurology at the Theodor-Wenzel-Werk in Berlin while continuing her teaching activities in Bonn. She received her Ph.D. (Promotion) 1999 from the University Medical School Bonn and specialized in neurology in 2003. From 2004 to 2005, she worked as a postdoctoral researcher at the Life & Brain Center in Bonn, where she focused on mitochondrial genetics. In 2008, she received the venia legendi in neurology for her habilitation thesis on mitochondrial diseases. In 2014, she was appointed associate professor. Cornelia's research focuses on brain involvement in neuromuscular diseases, with a particular emphasis on mitochondrial disorders and myotonic dystrophies. She is a member of several national and international scientific societies. From 2017 to 2025, she represented the University Hospital Bonn as a healthcare provider in the European Reference Network (ERN) EURO-NMD and has since continued her work as a co-chair of the EURO-NMD mitochondrial working group. She served as a member of the ENMC Research Committee until 2026.

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Prof. Teerin Liewluck | Rochester, USA

PROF. TEERIN LIEWLUCK is Professor of Neurology at the Department of Neurology, Mayo Clinic, Rochester, Minnesota, USA. He received his MD from Mahidol University in Thailand and completed postdoctoral research with Prof. Ichizo Nishino at the National Center of Neurology and Psychiatry in Tokyo, Japan. He subsequently completed neurology residency training at the University of Miami, Florida, followed by fellowship training in neuromuscular disorders with Prof. Andrew G. Engel at Mayo Clinic and in Clinical Neurophysiology (EMG).
Prof. Liewluck's clinical and research work focuses on the clinical, pathological, and electrophysiological characterization of muscle diseases, including both acquired and hereditary myopathies. His particular interests include monoclonal gammopathy–associated myopathies, inclusion body myositis, immune mediated necrotizing myopathy, and immune checkpoint inhibitor–related myositis. In 2024, he described a novel subtype of autoimmune myopathy termed immune-mediated megaconial myopathy. He served as Chair of the ANA Neuromuscular Disease Special Interest Group in 2023, Vice Chair of the AAN Neuromuscular Section from 2023 to 2025, and Vice Chair of the IMACS Scientific Committee from 2023 to 2025. He currently chairs the AANEM Abstract Review Committee, is an Associate Board Member of IMyoS, and serves on the UCNS Clinical Neuromuscular Pathology Examination Committee.

Prof. Hanns Lochmüller | Ottawa, Canada

PROF. HANNS LOCHMÜLLER is a Professor of Neurology at the University of Ottawa Faculty of Medicine and The Ottawa Hospital Department of Medicine. He is a neurologist and clinical academic specializing in genetic neuromuscular disorders and rare disease. Hanns is Senior Scientist at the Children's Hospital of Eastern Ontario (CHEO) Research Institute. Hanns trained as a neurologist in Munich, Germany and in Montreal, Canada. From 2007 to 2017, he held the chair of experimental myology at the Institute of Genetic Medicine at Newcastle University in the UK. He continues to hold a scientific appointment at the Department of Neuropediatrics and Muscle Disorders of the Medical Center – University of Freiburg in Germany and as visiting scientist at the Centro Nacional de Análisis Genómico (CNAG), Centre for Genomic Regulation, Barcelona in Spain.
In addition to his scientific and clinical research interests, he is internationally active in rare disease science policy and research collaborations.
He chaired the IRDIRC and TREAT-NMD. He initiated and coordinated the highly successful "RD-Connect" international infrastructure for rare disease data and biosample sharing and analysis, MD-net and EuroBioBank, a European (and Canadian) network of biobanks for rare disorders (https://lochmullerlab.org/).

Prof. Annamaria De Luca | Bari, Italy

PROF. ANNAMARIA DE LUCA is Full Professor of Pharmacology specialized in pre-clinical in vivo and ex vivo studies of inherited and acquired neuromuscular disorders.
In this general frame, she has a long lasting experience in preclinical research on pharmacological treatments for DMD and muscle channelopathies and collaborates with leading scientists in the field. She developed multidisciplinary assays for mdx mice and largely contributed to standardization of methods used to test pathology progression and drug efficacy, especially muscle functionality and electrophysiology. Annamaria has a profound knowledge of mouse physiology and pathology as well as of pharmacodynamics and pharmacokinetic topics for classical and innovative approaches, and hence of the critical points that make out the quality of a study in translational research of novel therapeutics. The results of her research led to registration of novel and repurposed drugs as orphan drugs, for various muscle disorders.
Annamaria De Luca is actively engaged in the pre-clinical work within TREAT-NMD and started working at the harmonization of preclinical studies on mdx before TREAT-NMD was launched. Since 2008, she has been involved in the TREAT-NMD Advisory Committee of Therapeutics (TACT), first as pharmacologist and pre-clinical expert, since 2015 as member of the Core committee. Annamaria has been Chair of TACT in the 2018-2022 round and voted member of the TREAT-NMD executive committee since 2020. Nowadays she is nominated component of the Neuromuscular Disease Advisory Committee (NMDAC) of TREAT-NMD and Member for Italy of the Governing Board of ERDERA.

Dr Andrew Mammen | Bethesda, USA

DR ANDREW MAMMEN is currently Associate Director of the Johns Hopkins Myositis Center and Investigator of the National Institutes of Health (NIH) in Bethesda, USA.
After obtaining his MD and PhD at Johns Hopkins, Dr Mammen completed his neurology residency and neuromuscular fellowship at the same institution. He co-founded the Johns Hopkins Myositis Center in 2007, where he and his colleagues discovered a novel form of autoimmune myopathy associated with statin use and autoantibodies recognizing HMG-CoA reductase, the pharmacologic target of statins. In 2014, Dr Mammen moved to the NIH, where he is an Investigator and Leader of the Muscle Disease Unit. His laboratory focusses on understanding the pathological mechanisms underlying disease in dermatomyositis, polymyositis, immune-mediated necrotizing myopathy, and inclusion body myositis. In addition to seeing myositis patients at the NIH Clinical Center, he maintains an appointment as Adjunct Professor of Neurology and Medicine at Hopkins, where he continues to see patients at the Myositis Center.

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Dr Thomas Meier | Basel, Switzerland

DR THOMAS MEIER is a dynamic life-science entrepreneur who established Santhera Pharmaceuticals as successful Biotech/Specialty Pharma company in Switzerland. In the past 20 years he held executive manager positions (CSO and CEO) at Santhera and successfully executed merger/acquisitions, product licensing and turn-around situations. He also is co-founder of SEAL Therapeutics, developing an innovative gene therapy approach for LAMA2-related muscular dystrophy.
In 2020 Dr Meier became Managing Partner at Viopas Venture Consulting. Currently Thomas holds Board positions and advises several private and public pharma/biotech companies. Dr. Meier has a PhD in Biology and is lecturer in neurosciences at the Biozentrum, University of Basel. He is an internationally recognized scientist with track record in clinical research of neuromuscular and other orphan diseases.

Prof. Maria Pennuto | Padova, Italy

PROF. MARIA PENNUTO is a Professor of Molecular Biology at the Department of Biomedical Sciences of the University of Padova and a Group Leader at the Veneto Institute of Molecular Biology (VIMM). After graduating from the University of Rome "La Sapienza", she obtained her Ph.D. in Cellular and Molecular Biology at the University of Milan, Italy, followed by post-doctoral training on neurological diseases at San Raffaele Scientific Institute, and at the NINDS, NIH in the USA, and Staff Scientist position at the Neurology Department of the University of Pennsylvania. In 2009 she returned to Italy to direct a research group working on the pathogenesis of motor neuron and neuromuscular diseases. Prof Pennuto identified several drugs with therapeutic potential that led to two phase II clinical trials. She authored more than 80 peer-reviewed papers, and her research is supported by National and International grant agencies. Prof Pennuto mentored several students (BS, MS, and PhD) and post-doctoral fellows, five of which obtained a position as independent investigators in academia or the industry in Italy and abroad.

Prof. Gina Ravenscroft | Nedlands, Australia

PROF. GINA RAVENSCROFT is a Senior Principal Research Fellow at the University of Western Australia and leads the Rare Disease Genetics and Functional Genomics group at the Harry Perkins Institute of Medical Research. She has been awarded >$29M in competitive grant funding, including three NHMRC Fellowships (ECF, CDF1, EL2). Her research focuses on disease gene discovery and characterisation for neuromuscular and neurogenetic diseases that present across the lifespan. Recently her team have identified and characterised novel repeat expansion disorders. In addition, she also works closely with the Neurogenetic Unit, PathWest to implement new diagnostic tests for these disorders. She is on the scientific advisory boards for A Foundation Building Strength for Nemaline Myopathy and the FSHD Global Research Foundation and is Secretary of the World Muscle Society (WMS) and a member of the WMS Scientific Programme Committee. In 2026 she chairs the Local Organising Committee for the joint AAN-AOMC congress in Perth. Gina's research and advocacy have been recognised through awards including AIPS Tall Poppy Award, WMS Young Myologist of the Year, Business News Perth Intrapreneur of the Year and the WA Premier's Mid-Career Scientist Award. She is passionate about supporting women in STEM, mothers in STEM and EMCRs.

Prof. Markus Rüegg | Basel, Switzerland

PROF. MARCUS RÜEGG is Professor for Neurobiology at the Biozentrum, University of Basel, Switzerland. He studied Biochemistry and Neurobiology in Zurich and Stanford and was co-founder and member of the management of MyoContract, now called Santhera Pharmaceuticals, a biotech company that dedicated to develop therapies for neuromuscular diseases.
Prof. Rüegg is an internationally recognized expert in neuromuscular research and has published numerous scientific papers in the field of neuromuscular research. In recent years, his work has also been devoted to the study of therapeutic interventions in mouse models for congenital muscular dystrophy. Prof. Rüegg is also partner of the network TREAT-NMD and he has been an active member of the ENMC Research Committee for six years.

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Prof. Ulrike Schara-Schmidt | Essen, Germany

PROF. ULRIKE SCHARA-SCHMIDT is professor for neuropaediatrics and neuromuscular diseases. She heads the Department of Neuropaediatrics and the Centre for Neuromuscular Diseases in Children and Adolescents at the Children's Hospital of the University Hospital Essen.
Prof. Dr. Schara-Schmidt is one of the leading national and international experts in the field of neuropaediatrics / neuromuscular diseases. She is vice president of the Society of neuropediatrics and board member of the German Brain Council, chair of the Ethics Committee of the Medical Faculty Duisburg-Essen and member of numerous national and international committees. Prof. Schara-Schmidt and her team are involved in international studies.

Prof. Bert Smeets | Maastricht, The Netherlands

PROF. BERT SMEETS is Professor in Clinical Genomics with a focus on Mitochondrial Diseases. He is an internationally dis-tin¬guished clinical molecular geneticist. He founded the Genome Center Maastricht and established a Master Programme in Systems Biology at Maastricht University (Maastricht, NL).
Prof. Smeets studied Molecular Biology at the University of Nijmegen, where he did a PhD on myotonic dystrophy. For 10 years he worked in Nijmegen, and since 1995 at Maastricht University, The Nether-lands, where he became Professor in 2010, combining research with genetic testing services.
His research concentrated on the genomics of mitochondrial disorders. His research involves identifying genetic defects by next-generation sequencing, studying the pathophysiology and mtDNA bottleneck mechanism in cell lines, iPSC and zebrafish models, developing new treatment options (compounds or autologous stem cells) and preventing the transmission of mitochondrial diseases, either by prenatal diagnosis, preimplantation genetic diagnosis or whole exome preconception screening. The focus of his current research is the development of and autologous stem cell therapy for genetic and non-genetic muscle disease. This is currently in a Phase II clinical trial for patients with an mtDNA mutation. To accelerate the road to the patient eh established a company Milocron Therapeutics to achieve this. His research is funded by a broad variety of national and international funding agencies.

Prof. Werner Stenzel | Berlin, Germany

PROF. WERNER STENZEL is a Neuropathologist and Myopathologist, and board certified in general Neurology and in Neuropathology. Currently, he is the head of the neuromuscular diagnostic and research unit in Berlin at the Charité Hospital. He also acts as co-Director of the German reference center of Neuromuscular diseases. His primary research interest is in juvenile and adult forms of myositis and inflammatory nerve pathologies. His team study pathogenesis of the different subentities of myositis, pathogenicity of autoantibodies, cancer association and the role of different immune cells. They also use new techniques for ultrastructural myopathology in diagnostic routine as well as for research questions related to the identification and characterization of morphological features of new entities. More recently they integrate multi-omics approaches with their morphological data. Prof. Stenzel is serving on the editorial of several journals in the field of Neurology and Neuropathology as well as Neuromuscular diseases. Many scientists and medical doctors have been in his laboratory in the past years, and it has always been a great pleasure to work with people from all over the world and learn about their different aims and needs to achieve the next steps in their respective careers.

Prof. Volker Straub | Newcastle, UK

PROF. VOLKER STRAUB is the Deputy Dean, Harold Macmillan Professor of Medicine and Professor of Neuromuscular Genetics at the Institute of Translational and Clinical Research at Newcastle University, United Kingdom. He is the Director of the university's John Walton Muscular Dystrophy Research Centre and holds an honorary clinical appointment with the Newcastle upon Tyne Hospitals NHS Foundation Trust. He was trained as a paediatric neurologist at the University of Düsseldorf and the University of Essen in Germany. He wrote his PhD thesis on Duchenne muscular dystrophy (DMD) and worked as a postdoctoral research fellow in Dr Kevin Campbell's laboratory at the Howard Hughes Medical Institute at the University of Iowa in Iowa City, Iowa, USA, on limb girdle muscular dystrophies (LGMD). He was the co-founder of the EU FP6 funded network of excellence for genetic neuromuscular diseases, TREAT-NMD, which he coordinated together with Kate Bushby. In 2019, he established TREAT-NMD Services Ltd., the business arm of the alliance, and is one of the directors of the enterprise. Volker represents the UK on the Scientific Committee of the European Cooperation in Science and Technology (COST). He is PI for several natural history and interventional trials in DMD, LGMD, Pompe disease, spinal muscular atrophy and other NMDs. He is a current Executive Board member and past-president of the World Muscle Society and an author on > 500 peer-reviewed publications.

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Prof. Vincent Timmerman | Antwerp, Belgium

PROF. VINCENT TIMMERMAN is full professor at the University of Antwerp (UA). His Peripheral Neuropathy Research Group started in 1996 with mapping and identifying genes associated with inherited peripheral neuropathies. They study the effect of mutations on the normal functioning of genes in neuronal and non-neuronal cell lines, including neurons differentiated from human induced pluripotent stem cells (iPSC). They gained knowledge with diverse gene cloning technologies and have phenotyped -in house made- mouse and Drosophila models for peripheral neuropathies. So far, Prof. Timmerman supervised 10 postdocs and 15 PhD students (6 on-going PhDs). He is a lecturer and supervisor of bachelor/master students at the UA. Further, he became a partner in the UA Research Excellence Center (OEC µNeuro) on: 'Multidimensional analysis of the nervous system in health and disease'. He was also an active partner in the FP7 Neuromics, H2020 Solve-RD project, SAB member of the MRC Center for Neuromuscular Diseases and member of the Inherited Neuropathy Consortium - Rare Disease Clinical Research Consortium (USA). Further, he was the secretary of the CMT and related disease consortium (CMTR) within the Peripheral Nerve Society, and currently member of the PNS program committee. He is a reviewer for several granting organizations in Europe.

Dr Pannie Trifillis | Whitehouse Station, USA

DR PANNIE TRIFILLIS Ph.D. is a global medical affairs leader who is passionate about discovering, developing and commercializing therapies for rare diseases with high unmet need, and providing access to treatment globally for every patient who can benefit.

In her 30-year career, Dr. Trifillis has launched five rare disease drugs globally. More impactfully, she has brought two rare disease drugs, Translarna and Evrysdi, from a concept idea in the lab through clinical trials to patients living with Duchenne Muscular Dystrophy and Spinal Muscular Atrophy, respectively.
As the first employee at PTC Therapeutics Inc. and during her 25-year tenure there, she played pivotal roles in research, strategic alliance management, and global medical affairs. Her expertise spans diverse therapeutic areas, and she has fostered collaborations between biopharma companies, patient advocacy groups, and health care professionals. Dr. Trifillis currently serves as Vice President and Head of Medical Affairs at Skyhawk Therapeutics, Inc.
Born in Cyprus, Dr. Trifillis went to the USA as a Fulbright Scholar where she earned her BA in Biology and Chemistry from Wellesley College, MA. She subsequently earned a PhD in Molecular Biology and Human Genetics from the University of Pennsylvania.

Prof. Bjarne Udd | Helsinki, Finland

PROF. BJARNE UDD, born in Finland, received his MD from the University of Berne, Switzerland in 1975, and completed Residency training in Neurology at the University Hospital in Umeå, Sweden, and the University Hospital in Helsinki, Finland. He received his PhD degree from Helsinki University in 1992 with the thesis on a new type of muscular dystrophy, causing the distal Tibial muscular dystrophy (TMD) in heterozygotes and severe LGMD R10 in homozygote patients. With academic positions (docent) at Tampere University from 1997 he was appointed Professor of Neurology in Neuromuscular Disorders in 2007 and the Head of the Neuromuscular Research Center, Tampere University Hospital. He is currently PI of the neuromuscular research group in Helsinki, at the Folkhälsan Research Center. Prof. Udd is member of many international research boards in Neuromuscular diseases. His research focuses on very rare neuromuscular diseases: clinical and molecular genetic research on muscular dystrophies, in particular: distal myopathies, rimmed vacuolar myopathies, limb-girdle dystrophies and myotonic dystrophy type 2. Since the first ever description of human titinopathy the research efforts have resulted in more than 25 primary descriptions of new muscle diseases many with previously unknown myopathy genes.

Prof. Mariz Vainzof | São Paulo, Brazil

PROF. MARIZ VAINZOF is Professor of Genetics, Head of Laboratory at the Human Genome Research Center, University of Sao Paulo (USP), São Paulo, in Brazil. She graduated in Biology, at the Instituto de Biociências (IB-USP) in 1977, obtained a MSc in genetics in the Department of Biology in 1983 and a Ph.D. in the Department of Genetics in 1989. She was trained in the Department of genetics, The Hospital for Sick Children, Toronto, Canada. Since more than 30 years, Prof. Mariz Vainzof is Head of the muscle protein and Comparative Histopathology at the Human Genome and Stem Cell Research Center, IB-USP.
Prof. Mariz Vainzof has an extensive list of scientific publications, books and has won many awards for her research activities. She is member of the International Consortium on Nemaline Myopathy, International Standard of Care Committee for Congenital Myopathies, and several other international and national organizations involved in muscle research.
Last but not least, Mariz is highly involved with the Muscle World Society, being at the moment part of the International Congresses program committee.

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Prof. Kathryn Wagner | Baltimore, USA

PROF. KATHRYN WAGNER received her MD and PhD in molecular neuroscience from the Johns Hopkins School of Medicine. Her PhD thesis was on the original cloning and characterization of the 87kD protein later named dystrobrevin. Following residency training in neurology and clinical fellowships in neuromuscular and neurogenetic disorders, Kathryn was a postdoctoral fellow in molecular biology where she described the loss of myostatin in the mdx model of DBMD and in an otherwise healthy human infant. She continued this work, elucidating the role of myostatin and related signaling molecules in muscle regeneration in her own laboratory at Hopkins and is currently Professor Emerita of Neurology and Neuroscience at the Johns Hopkins School of Medicine. She was the founding director of the Center for Genetic Muscle Disorders at the Kennedy Krieger Institute where she cared for pediatric and adult individuals with neuromuscular disorders and conducted investigator and industry initiated clinical trials. In order to advance drug development for rare disorders, she transitioned to industry where she served as Vice President, Global Head of Neuromuscular Diseases at Roche leading late-stage programs in DMD, SMA, gMG and FSHD, obtaining multiple regulatory approvals. She subsequently served as Vice President, Global Head of Translational Medicine, Neuroscience at Novartis where she directed early development of a broad portfolio. Currently, Prof Wagner is the Chair of the Board of Trustees of TREAT-NMD and an independent consultant to nonprofits and biotechnology companies.