

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 is 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). Recently she chaired the TREAT-NMD Executive Committee. Thus far, she has published over 200 peer-reviewed papers and 11 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.

Drs Anita Atema | Nijmegen, The Netherlands

DRS ANITA ATEMA is an experienced and results-driven executive director and general manager with strong personal commitment to patient engagement and access. She has broad experience in various (bio/orphan) pharmaceutical companies in e.g. marketing, sales, medical affairs, market access, corporate affairs and patient advocacy.

Anita Atema worked for more than 25 years for various pharmaceutical companies such as Sandoz (now Novartis), Amgen, Roche and Celgene both at the local and the international level. She recently joined Jazz Pharmaceuticals, Nijmegen, The Netherlands, as the national

country manager. Anita specialized in the following areas: Hematology, Oncology, Inflammation & Immunology, Targeted Cancer Therapy (CAR-T) and Epigenetics. In the different positions she held, Anita was responsible for the successful launches of 5 (orphan) drugs in the Dutch market in more than 10 diseases by creating unique patient access solutions. Her additional international experience is the implementation of patient engagement in all phases of the day to day business (from clinical trials to market access to (launch) campaigns).



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.

Dr James Dowling | Toronto, Canada



DR JAMES DOWLING is a clinician-scientist focused on gene discovery and therapy development for childhood muscle diseases. Dr 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.

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. Bertrand Fontaine | Paris, France

PROF. BERTRAND FONTAINE is a medical researcher and neuro-myologist at the Salpêtrière Hospital in Paris and a Professor of Medicine at the Sorbonne University. Since October 2019, Bertrand is Medical and Scientific Director of the Institute of Myology, Paris, France.

Prof. Fontaine is one of the discoverers of the role of ion channels, molecules that transmit electrical signals, in neuromuscular diseases. Between 2003 and 2008 he was the Director of the Sorbonne-Université and the INSERM (U546) Research Institute. Since September 2018, he has been Director of the Myology

Research Center (UMR 974) and was appointed on July 11, 2019 as head of the Neuro-Myology Department, Institute of Myology.

Prof. Fontaine has received many awards for his outstanding medical research work and for his managerial activities. He is an appointed member of both national (France) and international Executive, Editorial and Scientific boards (e.g. International Committee of the American Academy of Neurology, National Institute of Health-USA) and is often asked for communications (interviews and publications) to the general public.



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. 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 Neuropedi-

atrics 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 EuroBio-Bank, a European (and Canadian) network of biobanks for rare disorders.

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 is Chair of TACT from November 2018 for the three years round and voted member of the Executive Committee of TREAT-NMD since 2020.



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.

Dr Carsten Bönnemann | Bethesda, 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 Neuro-

muscular and Neurogenetic Disorders of Childhood Section. Dr Bönnemann was a Pew Fellow in the Biomedical Sciences. He received the 2009 Derek Denny Brown Neurological Scholar Award. 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 many neuromuscular networks, such as the WMS, TREAT-NMD, ENMC and has an impressive track record of scientific publications, lectures and editorial work making progress in the research of neuromuscular patients.

Prof. Anders Oldfors | Gothenburg, Sweden

PROF. ANDERS OLDFORS studied medicine in Gothenburg, Sweden. After his PhD thesis on the perineurium in 1980 he did a postdoc with Michel Fardeau in Paris, 1981, learning muscle histopathology. During specialization in surgical and neuropathology, he initiated diagnostic service and research in muscle pathology in Gothenburg. The research was initially focused on mitochondrial diseases in an interdisciplinary network of pioneers. He started early to combine pathology and genetics in the lab to identify new diseases, mainly in the field of congenital myopathies such as myosin myopathies and glycogen storage diseases. Another longstanding and

still ongoing project concerns inflammatory myopathies especially inclusion body myositis. After organizing the 9th World Muscle Society (WMS) international congress in Gothenburg 2004, he has been a continuous member of the Executive Board of WMS and a member of the Editorial Board of Neuromuscular Disorders, the official journal of WMS. He also served for a period in the ENMC scientific committee and several other scientific priority committees. At present Anders Oldfors is Senior Professor at the University of Gothenburg and Consultant Pathologist at the Sahlgrenska University Hospital.



Prof. Markus Rüegg | Basel, Switzerland



PROF. MARCUS RÜEGG is Professor for Neuro-biology 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.

Prof. Ulrike Schara-Schmidt | Essen, Germany



PROF. ULRIKE SCHARA-SCHMIDT is Professor for Neuropaediatrics and Neuromuscular Diseases and is one of the leading national and international experts in these areas of clinical care and research. 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. She is president of the Society

for Neuropaediatrics, vice president of the German Brain Council, chair of the Ethics Committee of the Medical Faculty Duisburg-Essen and member of numerous national and international committees. She has also been a member of the ENMC Research Committee from 2012-2018. Prof. Schara-Schmidt and her team are involved in international studies.

Dr Selma Tromp | Nieuwegein, The Netherlands

DR SELMA TROMP is Neurologist / Clinical Neurophysiologist at the Leiden University Medical Center, The Netherlands. She worked since 2011 at St. Antonius Hospital, Nieuwegein, the Netherlands, where she was responsible for the education of residents and medical management of the department. After her PhD, Dr Tromp started her career as Neurologist / Clinical Neurophysiologist at the departments of Clinical Neurophysiology and Neurology of the Academic Hospital Maastricht, the Netherlands. Dr Tromp has held several board functions.

From 2013, Dr Tromp was member / chair of the educational committee of the Dutch Society for Clinical Neurophysiology; from 2009-2017, she was member of the board of the Dutch Society for Neurology and in the last three years she was the Chair of this board. From Nov 2017-2020, Dr Tromp was member of the Supervisory Board of the Elisabeth-TweeSteden Hospital, Tilburg, the Netherlands. Currently, she is member of the board of the Dutch Federation of Medical Specialists, with her special focus on quality of care.



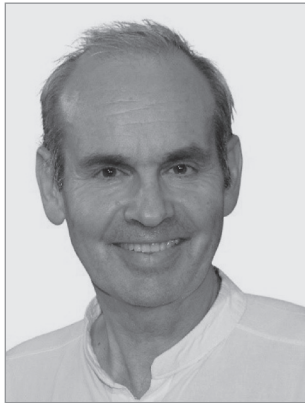
Prof. Bert Smeets | Maastricht, The Netherlands



PROF. BERT SMEETS is Professor in Clinical Genomics with a focus on Mitochondrial Diseases. He is an internationally distinguished 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 Netherlands, 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. His research is funded by a broad variety of national and international funding agencies and he published over 225 original research articles, reviews and book chapters (H-Index 49).

Prof. Andoni Urtizberea | Paris, France



PROF. ANDONI URTIZBEREA is a physician trained in Paris University and certified both in paediatrics and physical medicine and rehabilitation. After graduating in parallel from the Institut d' Etudes Politiques de Paris in 1987, he served many years as Medical Director of the AFM-Telethon and then as General Delegate of the Institut de Myologie of Paris (1993-2000). As Scientific Director of the ENMC (1999-2005) and together with AFM's support, he contributed to the establishment of many worldwide networks in myology, an emerging discipline in the medical field. He served until December 2019 as a part-time clinical myologist in Hendaye, France (APHP)

and as deputy coordinator of the French Neuromuscular Network (FILNEMUS) in Marseilles. Over the past twenty years, he headed various worldwide educational events dedicated to myology (in France, Russia, Latin America and, more recently in the Middle-East). He is a regular visiting professor in various countries and a consultant for many pharma involved in the field. Ideally located at the intersection of industry, patient advocacy groups and academia, his main objective is to raise more awareness about these rare conditions notably in emerging countries and more specifically in the context of novel cutting-edge therapies.

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.



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 honorary clinical appointments with the Newcastle upon Tyne Hospitals NHS Foundation Trust and the North Tees and Hartlepool NHS Foundation Trust. Volker 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). Volker 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. He is PI for a number of natural history and interventional trials in DMD, LGMD, Pompe disease, spinal muscular atrophy and other NMDs. Volker recently chaired the European MYO-MRI COST Action to develop applications of MR imaging and spectroscopy techniques in neuromuscular disease. He is part of the steering committee of the MRC Centre for neuromuscular diseases and is now President of the World Muscle Society. He is author on > 300 peer-reviewed publications.

Dr Carina Wallgren-Pettersson | Helsinki, Finland



DR CARINA WALLGREN-PETTERSSON is a Medical Doctor specialised in Medical Genetics, working as a Principal Investigator at The Folkhälsan Institute of Genetics and Department of Medical and Clinical Genetics, University of Helsinki, Biomedicum Helsinki. She coordinates the scientific efforts of the Research Group on Nemaline Myopathy and Related Disorders, together with her Co-PI Katarina Pelin, PhD. Carina's research has concentrated on NMDs, mainly on nemaline myopathy and previously also on myotubular myopathy. She initiated the ENMC Interna-

tional Consortium on Myotubular Myopathy in 1993 and coordinated its research activities until January 2000. The consortium organised altogether 9 workshops on nemaline myopathy (NM) and 7 on myotubular myopathy. Carina Wallgren-Pettersson initiated the informal network called the Finnish Neuromuscular Club and was one of the initiators of the Finnish network on muscle disorders in children and, most recently, the European Reference Network NMD-FIN Consortium, now accepted as part of EURO-NMD.