Expert Masterclass on Duchenne Muscular Dystrophy

Thursday 16 – Friday 17 May 2019
Movenpick Hotel, Amsterdam City Centre, the Netherlands

Programme Management

Cathy Turner
TACT coordinator and Duchenne Liaison
John Walton Muscular Dystrophy Research Centre, Newcastle University, International Centre for Life,Newcastle upon Tyne, NE1 3BZ UK
T: +44 (0)191 241 8659 (direct)
T: +44 (0) 7976839553 (mobile)
E: catherine.turner@newcastle.ac.uk

Clare Bradley
DMD Education Coordinator
John Walton Muscular Dystrophy Research Centre, Newcastle University, International Centre for Life, Newcastle upon Tyne, NE1 3BZ UK
E: clare.bradley@newcastle.ac.uk

Accommodation and Meeting Venue

Mövenpick Hotel Amsterdam City Centre

Piet Heinkade 11

1019 BR Amsterdam

Netherlands

+31 (0) 20 519 1200

https://www.movenpick.com/en/europe/netherlands/amsterdam/hotel-amsterdam/overview/

Accommodation and Payment

Accommodation has been reserved for you according to your travel schedule and/or specific arrangements with masterclass organisers. Masterclass organisers will be in touch with details of your personal arrangements.

Your room and breakfast charges will be charged to the conference account where agreed, but we respectfully ask you to settle any incidental room charges yourself prior to departure.

Wireless Internet is available free of charge throughout the hotel.

Breakfast is served in the Silk Road Restaurant from 06.30.

Travelling to the hotel

The hotel is located near the central railway station, alongside the water's edge of the river IJ in the vibrant heart of Amsterdam and can be reached easily via multiple forms of transport.

From Amsterdam Airport Schiphol

The direct train from Schiphol Amsterdam Airport (AMS) to Central Station takes 20 minutes. Taxi to the hotel is around 30 minutes but is expensive and cannot be reimbursed other than in exceptional circumstances.

Amsterdam Central Station is walking distance from Mövenpick Hotel Amsterdam City Centre but the hotel also offers a complimentary shuttle bus. Please book your seats in advance at the concierge desk: +31 (0)20 519 1213.

By Tram

The "Muziekgebouw/Bimhuis" tram stop is in front of the hotel, which is adjacent to the Passenger Cruise Terminal.

By Car

Take the exit from the Eastern Highway (Ringweg) A10. At the end of the Piet Hein tunnel, turn right towards Central station. Follow the signs P-Piet Hein/City Centre. The Piet Hein parking garage offers electric car drivers the possibility to recharge their cars at charge points.

Parking

Q-Park Garage IJ Over Centrum: 200m from the hotel. Purchase a EUR 10 value card for 24 hours at the hotel reception (regular rate at EUR 40/day).

Underneath the hotel: There is a parking garage under the hotel (costs EUR 4.29 per hour, no discounts available). Therefore, we advise to park at Q-Park as described above.

Meals

Meals and refreshments will be provided throughout Thursday and until the end of the Masterclass on Friday. On Thursday evening, there is dinner for all delegates*, which will be served in the Silk Road restaurant at 19:00.

*please ensure you have sent information regarding any dietary requirements to: catherine.turner@newcastle.ac.uk or clare.bradley@newcastle.ac.uk

Hotel Check-Out

Please allow enough time to check-out prior to the start of the meeting on 17th May. Late checkout after 12 noon may incur a supplement, which will be charged to your extras account.

Attendance Certificates

We will email you an attendance certificate following the Masterclass.

Dress Code
The dress code for the meeting and dinner is business/casual.

Insurance
Please ensure you have adequate cover for personal possessions and any medical expenses.
TREAT-NMD cannot take responsibility for loss or damage to personal possessions during your stay or emergency medical costs.

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Movenpick Hotel, Amsterdam City Centre, the Netherlands

Contact: Cathy Turner, TREAT-NMD Secretariat/Newcastle University, catherine.turner@ncl.ac.uk

Meeting objectives

- Provide a high-quality educational and interactive meeting for paediatric neurologists and other related specialities who may be diagnosing and/or managing the care of patients with Duchenne muscular dystrophy (DMD)
- Present the natural history of DMD and review the typical signs and symptoms of the disease
- Explain the genetics of DMD, and underline the importance of obtaining a mutation-specific diagnosis as early as possible
- Highlight the updated standards of care (2018) and discuss their implementation
- Give patient and parent perspectives on diagnosis, care and research in DMD

Meeting format

- The masterclass will be a mixture of lectures and breakout sessions, with a significant amount of time allocated to discussion
- Every delegate will be asked to download a messaging and polling app onto their phone or computer, with the aim of increasing engagement and interactivity at the meeting
- Reading will be provided in advance of the meeting and delegates are expected to prepare accordingly

Co-chairs

- Annemieke Aartsma-Rus, Leiden University Medical Center, the Netherlands, and John Walton Muscular Dystrophy Research Centre, Newcastle University, UK
- Nathalie Goemans, University of Leuven, Belgium
- Ros Quinlivan, MRC Centre for Neuromuscular Diseases, University College London, UK
- Elizabeth Vroom, World Duchenne Organization, the Netherlands

Additional speakers

- Maxime Arras, patient representative, Belgium
- Michela Guglieri, John Walton Muscular Dystrophy Research Centre, Newcastle University, UK
- Jana Haberlova, Charles University, Czech Republic
- Alex Johnson, Duchenne UK
- Erik Niks, Leiden University Medical Center, the Netherlands
- Elena Mazzone, Fondazione Telethon, Italy
- Cécile Ollivier, Aparito, the Netherlands
- Andoni Urtizberea, Paris Summer School of Myology, France
- Simon van Boxtel, patient representative
- Karim Wahbi, Cochin Hospital Paris, France

Meeting Details and Timings
Regardless of your time of arrival, please collect your delegate pack and name badge on Thursday morning from the TREAT-NMD conference desk in the hotel. Please wear your name badge throughout the conference including at dinner on Thursday night.

Day 1 – Thursday 16 May:
Chairs: Annemieke Aartsma-Rus (am) and Elizabeth Vroom (pm)

10:30-11:00	***Welcome coffee and registration***	
11:00–11:20	**Welcome and introduction to TREAT-NMD** • **Aims of the meeting**	Annemieke Aartsma-Rus
11:20-11:35	**Parent Perspective – Children** • **Behavioural issues and psychosocial impact**	Simon van Boxtel
11:35-12:00	**Parent Perspective – Adult** • **Experience in lower-income countries**	Elizabeth Vroom
12:00-12:15	**Introduction to DMD** • **Definitions** • **Variability of disease**	Nathalie Goemans
12:15–12:45	**Diagnosis** • **Clinical presentation** • **Communication** • **Neonatal screening** • **Tools to aid raised awareness**	Michela Guglieri and Alex Johnson
12:45–13:00	**Questions and Discussion**	
13:00–14:00	***Lunch***	
14:00–15:00	**Genetics** • **Explanation of genetics and mutation type** **Reference document:** **https://www.ncbi.nlm.nih.gov/pubmed/26754139** • **Need for sharing information** • **Explanation of diagnostic tools (NGS)** • **Genotype phenotype** • **Mutation specific therapies (brief)** • **Quiz**	Annemieke Aartsma-Rus
15:00–15:30	**Standards of Care (SoC)** • **Introduction** • **Steroid & management of chronic treatment** • **Emergency care** • **Introduction to break out**	Nathalie Goemans
15:30–15:50	***Coffee***	
15:50–16:30	**Breakout session (SoC)** • **Steroids**	3 groups, facilitated by Faculty
16:30-16:50	**Feedback and discussion**	
16:50-17:50	**Communication with parents** • **Identifying fake news from factual news** • **Delivering diagnosis appropriately** • **Answering parents' questions effectively and positively** • **Enabling parents to feel confident making decisions about their child's care** • **Managing parents' and carers' expectations**	Andoni Urtizberea

	• **Transition of care and autonomy for the child/adult – including the young adult in decision making as appropriate**	
17:50	**End of the day/wrap up**	Annemieke Aartsma-Rus
19:00	***Dinner***	

Day 2 – Friday 17 May
Chairs: Nathalie Goemans (am) and Ros Quinlivan (pm)

08:30–09:30	**Physiotherapy & Orthopaedics SoC** • **Care & assessments** • **Adult and emergency** • **PUL and North Star**	Elena Mazzone
09:30-10:15	**Respiratory SoC**	Jana Haberlova
10:15–10:45	***Coffee***	
10:45–11:25	**Cardiac SoC** • **Becker** • **Adults and carriers** • **Emergency**	Karim Wahbi
11:25–11:55	**Adult Duchenne Management including Becker** • **Introduction**	Ros Quinlivan
11:55-12:45	**Breakout** • **Adult DMD and Becker Management**	**Facilitated by Faculty**
12:45-13:00	**Feedback and discussion**	
13:30-13:45	***Lunch***	
13:45-14:00	**Reaching a consensus on SoC**	Erik Niks
14:00-14:40	**Emerging therapies – Bench to Bedside** • **Biomarkers** • **Regulatory & HTA** • **Financial implications**	Michela Guglieri and Cécile Ollivier
14:40-15:10	**Emerging therapies - Therapeutics in trials** • **Dystrophin restoration** • **Secondary pathology**	Annemieke Aartsma-Rus
15:10-15:25	**Clinical trial burden – patient perspective**	Maxime Arras
15:25	**Meeting wrap up and close**	Annemieke Aartsma-Rus
15:30	***Coffee available***	

Reading List

In advance of the Masterclass, we ask that you review the information in the following links/publications:

Publication on the genetic diagnosis of Duchenne muscular dystrophy: https://www.ncbi.nlm.nih.gov/pubmed/26754139

The 2018 Standards of Care publication from the Lancet which can be accessed here: http://www.treat-nmd.eu/care/dmd/diagnosis-management-DMD/

The DMD Guide for Families, based on the Lancet papers above: http://www.treat-nmd.eu/care/dmd/family-guide/translations/

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Co-Chairs

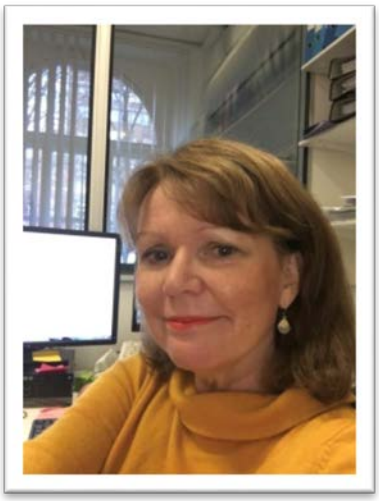
Annemieke Aartsma-Rus

Annemieke Aartsma-Rus 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, Department of Human Genetics (the Netherlands). As of December 2007 she became leader of the "DMD exon skip group" and is professor at the Department of Human Genetics. Since 2013 she has had a visiting professorship at the Institute of Genetic Medicine of Newcastle University (UK). In 2015 she was selected as the most influential scientist in Duchenne muscular dystrophy in the past 10 years (2006-2015) by Expertscape based on contributions to the understanding and treatment of Duchenne muscular dystrophy. In 2011 she received the Duchenne Award from the Dutch Duchenne Parent Project in recognition of this work and her dedication to the Duchenne field. In 2013 she was elected a member of the junior section of the Dutch Royal Academy of Sciences, which consists of what are considered the top 50 scientists in the Netherlands under 45.

Nathalie Goemans

Nathalie Goemans is a paediatrician and child neurologist, with certification in rehabilitation medicine. She has been a staff member at the University Hospitals Leuven since 1987. Currently she is head of clinic within the department of Pediatrics (Child Neurology) and head of the Neuromuscular Reference Centre for Children at the University Hospitals Leuven. She is assistant professor at the Faculty of Medicine, University of Leuven, Belgium. She is actively involved in national and international research networks, with a particular interest in the multidisciplinary aspects and the dissemination of standards of care in neuromuscular disorders and the development of international registries. Research activities involve translational and clinical research in various neuromuscular disorders, the development of new therapies for Duchenne muscular dystrophy and spinal muscular atrophies and the research and development of outcome measures to assess interventions in these diseases.

Ros Quinlivan



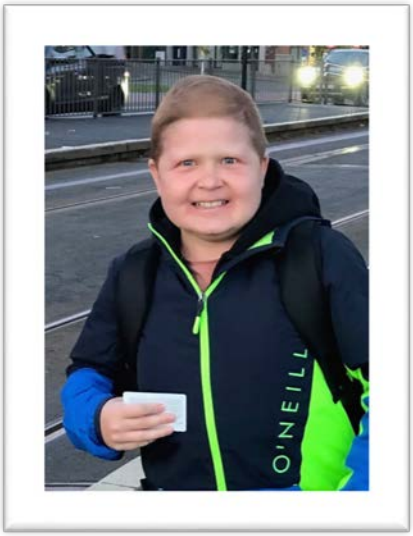
Ros Quinlivan studied at University College London. She has been working in the field of neuromuscular disease since 1990. She trained in paediatrics and developed an interest in neuromuscular disorders (NMD) during her time at Evelina Children's Hospital where she studied Duchenne and Becker muscular cardiomyopathy. She was director of the Centre for Inherited Neuromuscular Disease at RJAH Hospital in Oswestry from 2002-2010. She then moved to London to take up a post leading for transition of young people with NMD from Great Ormond Street Hospital to The National Hospital for Neurology and Neurosurgery. Currently, she cares for young adults with inherited NMD in the adult service. She also leads the North Star clinical network for adult DMD in the UK. She was a PI for the Ataluren trials and is currently a PI for the Sideros trials. She is joint co-ordinating editor for the Cochrane Neuromuscular Disease Group and has recently published a systematic review of cardiac treatment for DMD and BMD.

Elizabeth Vroom

Elizabeth Vroom is founder and president of the Duchenne Parent Project Netherlands and Chair and co-founder of the World Duchenne Organisation (formerly UPPMD). She is the mother of a son with Duchenne muscular dystrophy. She is a chair of the ethics board and member of the Executive Committee of TREAT-NMD and currently serves on several advisory boards regarding care, research, ethics, development of new medicines and regulatory issues in the Netherlands as well as internationally. She is involved in several publications regarding Standards of Care and drug development for Duchenne muscular dystrophy, concerning development of outcome measures and role of biomarkers. She is co-author of a handbook on psychosocial aspects of DMD. She participates in several EU funded projects, such as Asterix, Adapt Smart, Vision-DMD and Share4Rare.

Additional Speakers

Maxime Arras



Maxime is a resident of Mortsel, Belgium. He is 20 years of age.

He is currently a student at Antwerp KdG College, where he is in his first year studying for a BA in Marketing.

Maxime has Duchenne muscular dystrophy and was part of an exon skipping clinical trial for 7 years. He also has FDA & EMA testimonial experience.

Michela Guglieri

Michela Guglieri is a Senior Lecturer and Honorary Consultant in Human Genetics at the John Walton Muscular Dystrophy Research Centre in Newcastle, UK where she leads the clinical research team. Michela has developed a great interest in translational research, drug development and natural history studies in neuromuscular diseases, and specifically in Duchenne muscular dystrophy. Michela is the PI in several clinical trials in DMD. She is the Chief Medical Coordinator of the FOR DMD study; a large international study aiming to **F**ind the **O**ptimum **C**orticosteroid **R**egime for **DMD** and Study Chair of the Vision DMD, Phase IIb study of vamorolone in DMD. Michela is a member of the DMD national patient registry, the North Star Consortium and the CINRG group. She is involved in advisory boards of pharmaceutical companies developing drugs for neuromuscular disease. Michela has been involved as clinical expert in NICE and EMA consultations. She has developed an educational program in collaboration with the RCPCH to improve early recognition of DMD and other neuromuscular conditions in children.

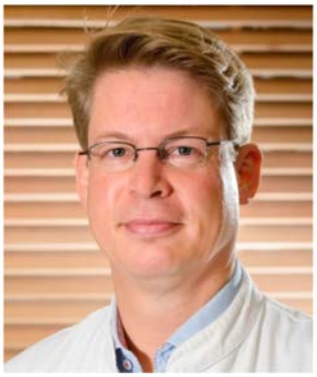
Jana Haberlova

Jana was trained as an adult neurologist, and has now been working for 20 years as a paediatric neurologist and electrophysiologist. Her main interests are neuromuscular disorders in children. During her training, she underwent a repeated short stay visit in MRC Centre for Neuromuscular Diseases in London, and for one year she worked in the John Walton Muscular Dystrophy Research Centre in Newcastle. Now she is back in Prague, where she set up the Neuromuscular Centre for Children in the University Hospital Motol in Prague.
She is involved in the clinical research and clinical trials with new therapies, especially in DMD and in SMA.
She is a Research Secretary of the Czech Paediatric Neurology Society, and a member of TREAT-NMD and WMS.

Alex Johnson

Alex founded the charity Joining Jack with her husband, ex-rugby league player Andy Johnson, following their son Jack's diagnosis with Duchenne Muscular Dystrophy in 2011. The rugby community came together to help the family create international awareness for Duchenne with innovative campaigns such as #Link4DMD. The charity has raised more than £2 million to spend on translational research and clinical trials. Alex has subsequently joined the board of United Parent Project (UPPMD) and has helped to organise international awareness events like World Duchenne Awareness Day. She has represented the Duchenne community by speaking at the Houses of Parliament as well as international conferences, workshops and meetings with the FDA, EMA, MHRA, NICE. Alex has also completed training at a Eurordis summer school to become a trained patient advocate and is a member of the MHRA patient group consultative forum.

Erik Niks



Erik Niks is consultant neurologist and paediatric neurologist at the Leiden University Medical Center, the Dutch Expert Center for Duchenne and Becker muscular dystrophies, and acquired neuromuscular junction diseases. After finishing his neurology training, he did a fellowship in myology funded by the Dutch Prinses Beatrix Fonds, which included an honorary fellowship at Dubowitz Neuromuscular Centre at Great Ormond Street Hospital, London in 2010. He has a particular interest in Duchenne and Becker muscular dystrophy and myasthenia gravis. He performed clinical, immunological, and epidemiological studies in seronegative and MuSK antibody positive myasthenia gravis in the Netherlands as part of his PhD. He is principal investigator at the LUMC for clinical trials in Duchenne and Becker, and coordinates the multidisciplinary care for children with neuromuscular diseases and is involved in international imaging and biomarker studies in these conditions. Erik is board member of the Duchenne Centre Netherlands and coordinator of the national biobank for Duchenne and Becker muscular dystrophy.

Elena Mazzone

Elena is a Physical Therapist who has 20 years of clinical experience with neuromuscular patients. For the past 10 years she has been working at the Catholic University in Rome, Italy in an internationally recognized neuromuscular centre as coordinator for therapists in national research networks and clinical PT.

At present, she is a master trainer in NMD therapeutic clinical trials and on different advisory boards. She has collaborated and played a key role in the development of specific outcome measures in SMA and DMD such as: the RULM, CHOP INTEND, Revised Hammersmith Scale, Performance of Upper limb, PROM and collected natural history data.

She has had many articles published on the value of standardized assessments in neuromuscular disorders and their application as clinical outcome measures in clinical trials.

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Cécile Ollivier

Cécile Ollivier is a senior health engineer with an MSc in international drug development and registration. She has extensive experience in global drug development in paediatric and rare disease. Prior to joining Aparito, Cécile was a scientific officer in the Paediatric office and the Science and Innovation office at the European Medicines Agency (EMA) for over 10 years. She provided technical and regulatory expert guidance on the design, conduct and interpretation of paediatric developments across multiple therapeutic areas. She has reviewed over 200 paediatric investigation plans (PIPs) and paediatric scientific advice (SA) across all phases of the development. Cécile has been recognised for her work leading and developing the EMA paediatric extrapolation strategy at EU and global level.

Andoni Urtizberea

Jon Andoni Urtizberea is head of the internationally renowned Paris Summer School of Myology and a consultant at the neuromuscular reference centre at Hendaye Hospital in the French Basque region, which is part of the large Assistance publique - Hôpitaux de Paris (AP-HP) hospitals group. In Hendaye he is responsible for a clinic exclusively dedicated to neuromuscular patients.

Andoni's special interest is in neuromuscular training and education and international networking and he has close links with specialists across the world. The annual Paris Summer School of Myology, which he established in 1997 together with Michel Fardeau, has trained 400 students from 55 nationalities in 10 years and is recognised as the leading specialist school for myology.

Simon van Boxtel
And
Esther Bruggink

Simon van Boxtel is a photographer in the world of classical music.

Esther Bruggink is a visual artist. She also works in the Rijksmuseum as a museum docent and museum guide.

They are parents of 12 year old twin brothers, Ilya and Milan, who are both affected by Duchenne muscular dystrophy.

Ilya also suffers from ASS and Milan from ASS, OCD and ADHD.

Karim Wahbi

Karim Wahbi is Associate Professor of Cardiology at the University of Paris Descartes, France and head of the reference centre for neuromuscular cardiomyopathies in Cochin Hospital, Paris. He received his medical degree from the University of Tours and completed his cardiology internship and residency at the University of Paris Descartes.

Karim specialises in the clinical management of patients with cardiomyopathies with a specific expertise in neuromuscular diseases such as dystrophinopathies. Key areas of clinical and fundamental research for Dr Wahbi are prevention of heart failure and sudden death and the study of molecular mechanisms underlying cardiomyopathy in patients with neuromuscular conditions.


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