

## **Expert Masterclass on Duchenne Muscular Dystrophy** (An educational program organised and funded by PTC Therapeutics)

Thursday 3 – Friday 4 May 2018  
Melia Castilla Madrid, Spain

### **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, as well as the current management guidelines
- Explain the genetics of DMD, and underline the importance of obtaining a mutation-specific diagnosis as early as possible
- Discuss the region-specific differences and similarities in the diagnosis pathway of DMD, and share best practice in order to improve patient outcomes
- Assess the effectiveness of different measurements for accurately determining the progression of disease in the clinic, and discuss regional differences and challenges in the management of patients with DMD
- Understand the importance of physiotherapy in the management of DMD
- Understand the importance of outcome measures to follow disease progression and assess therapies
- Provide an overview of existing therapies and those that are currently in late stage clinical development for the treatment of DMD
- Consider how the involvement of patient advocacy groups with industry and regulators may improve patient access to treatment

### **Meeting format**

- The masterclass will be a mixture of lectures and workshop 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. The app will ensure that discussions revolve around the topics of most interest to you and your colleagues, as well as allowing results of key questions taken at the meeting to be displayed

### **Co-chairs**

- **Annemieke Aartsma-Rus**, Leiden University Medical Center, Leiden, the Netherlands, and John Walton Muscular Dystrophy Research Centre, Newcastle University, Newcastle, UK
- **Jordi Díaz Manera**, Unitat de Malalties Neuromusculars, Servei de Neurologia, Hospital de la Santa Creu i Sant Pau de Barcelona, Spain

**Day 1 – Thursday 3 May**

12:30–13:30	<b>Arrivals and welcome lunch with Caregiver Masterclass delegates</b>	
13:30–13:40	<b>Welcome and programme overview</b>	Jordi Díaz Manera and Annemieke Aartsma-Rus
13:40–13:50	<b>Overview of TREAT-NMD</b>	<b>Presentation</b> by Annemieke Aartsma-Rus (10 minutes)
13:50–14:20	<b>The true impact of DMD: A parent's outlook</b> <ul style="list-style-type: none"> <li>○ Day-to-day care for a patient with DMD; how disease affects their ability to participate in educational and social activities, and their psychological state of mind; practical considerations for the family; support needed from healthcare systems</li> </ul>	<b>Presentation</b> by Filippo Buccella (15 minutes) <b>Discussion</b> (15 minutes)
14:20–15:20	<b>Introduction to DMD and current management guidelines</b> <ul style="list-style-type: none"> <li>○ Natural history; signs and symptoms; complications (cardiac, respiratory, bone, etc.)</li> <li>○ Outline the new international Standards of Care/guidelines for the management of DMD (and what has been updated since the 2010 guidelines), including brief overview of steroids and existing mutation-specific treatments for DMD, ventilation and physiotherapy</li> </ul>	<b>Presentation</b> by Nathalie Goemans (40 minutes) <b>Discussion</b> (20 minutes)
15:20–15:40	<b>Coffee</b>	
15:40–16:40	<b>The genetics of DMD and the importance of genetic testing in obtaining a mutation-specific diagnosis of DMD</b> <ul style="list-style-type: none"> <li>○ DMD gene in relation to dystrophin function, different mutation types and relation to diagnostic tests (MLPA vs exon sequencing vs full gene sequencing), importance of genetic confirmation of mutation, genetic testing vs muscle biopsy, importance of carrier assessment and early diagnosis</li> </ul>	<b>Presentation</b> by Annemieke Aartsma-Rus (30 minutes) <b>Discussion</b> (30 minutes)
16:40–17:50	<b>Group discussion: Diagnostics in practice</b> <ul style="list-style-type: none"> <li>○ How is genetic diagnosis done in your institute?</li> <li>○ Is small mutation analysis available? If not, how do you proceed when no deletions/duplications are found?</li> <li>○ How do you think diagnostics could be improved in your institute/country?</li> </ul>	<b>Discussion</b> facilitated by Annemieke Aartsma-Rus (70 minutes)
17:50–18:00	<b>Day 1 summary and close</b>	Jordi Díaz Manera
Evening	<b>Dinner</b>	

**Day 2 – Friday 4 May**

08:20–08:25	<b>Welcome back</b>	Annemieke Aartsma-Rus
08:25–10:50	<p><b>Practical management of DMD</b></p> <ul style="list-style-type: none"> <li>○ Summary of outcome measures used in DMD and the effectiveness of different measurements for accurately determining the progression of disease in the clinic</li> <li>○ Introduce concepts of new tools used to measure outcomes in DMD e.g. muscle MRI</li> <li>○ How to implement the standards of care guidelines into the day-to-day multidisciplinary clinic</li> <li>○ Brief overview of speaker's experience of best practice in paediatric patient management in the clinic in Spain</li> <li>○ Brief overview of speaker's experience in the management of adults with DMD in Spain and with using new outcome measures</li> </ul> <p><b>Breakout session:</b></p> <ul style="list-style-type: none"> <li>○ Region-specific workshops discussing the delegates' patient management practices (including hurdles, opportunities and examples of overcoming challenges that others can learn from)</li> </ul>	<p><b>Presentation</b> by Imelda de Groot (30 minutes)</p> <p>Andrés Nascimento Osorio (10 minutes)</p> <p>Jordi Díaz Manera (15 minutes)</p> <p><b>Breakout/discussion</b> facilitated by Imelda de Groot, Andrés Nascimento, Jordi Díaz Manera (60 minutes' breakout; 30 minutes' plenary discussion)</p>
10:50–11:10	<b>Coffee</b>	
11:10–12:10	<p><b>The importance of physiotherapy in the management of DMD</b></p> <ul style="list-style-type: none"> <li>○ Overview of physiotherapy assessments and management of DMD in the child and young adult, including the role of physiotherapists in the diagnosis of DMD</li> </ul> <p><b>Practical group session:</b></p> <ul style="list-style-type: none"> <li>○ Demonstrating stretches for the ambulant and non-ambulant child with DMD, and management of respiratory complications in the patient</li> </ul>	<p><b>Presentation</b> by Marina di Marco (30 minutes)</p> <p><b>Group practical/discussion</b> (30 minutes)</p>
12:10–13:10	<b>Lunch</b>	
13:10–14:00	<p><b>Therapies for DMD</b></p> <ul style="list-style-type: none"> <li>○ Overview of existing therapies and those that are currently in late stage clinical development for the treatment of DMD</li> </ul>	<p><b>Presentation</b> by Annemieke Aartsma-Rus (30 minutes)</p> <p><b>Discussion</b> (20 minutes)</p>
14:00–14:30	<b>Ensuring access to treatments for patients with DMD in Europe</b>	<p><b>Presentation</b> by Dimitrios Athanasiou (15 minutes)</p> <p><b>Discussion</b> (15 minutes)</p>
14:30–14:45	<b>Coffee</b>	
14:45–15:20	<p><b>Fighting for the patient with DMD: A patient advocacy group outlook</b></p> <ul style="list-style-type: none"> <li>○ The challenges that patient advocacy groups face in supporting patients with DMD, including a summary of the success stories and challenges discussed by the caregivers at the Caregiver Masterclass</li> </ul>	<p><b>Presentation</b> by Dimitrios Athanasiou (15 minutes)</p> <p><b>Discussion</b> (20 minutes)</p>
15:20–15:30	<b>Day 2 summary and meeting close</b>	Annemieke Aartsma-Rus