

European Expert Masterclass on Congenital Myopathies

Virtual online event

Wednesday 23rd & Thursday 24th February 2022

Masterclass Programme

Masterclass Objectives

- Deliver high quality educational and interactive event for health care providers diagnosing or managing the care of patients with the Congenital Myopathies (CM) by providing information on the latest in diagnosis, natural history, and care discussions.
- Provide a platform for health care providers to share ideas, challenges, and successes in delivering optimal care to people with CM.
- Provide an overview of the current clinical trial landscape and the emerging implications on care.

Masterclass Delivery Format

Due to the situation with COVID-19 a decision has been made to deliver this expert European Masterclass virtually, as a wholly online event using our interactive web-conferencing platform, Whova. The format will involve lectures from expert speakers, two interactive workshops and opportunities for Q&A throughout. Sessions will be delivered to delegates in English.

Day One (Wednesday 23rd February, 2022)

- Day one of the masterclass will include a series of lectures delivered by a range of clinicians with expertise in CM. The masterclass will also include input from patients with CM who will discuss the impact of CM on their lives. Delegates will be given opportunity for Q&A throughout.

Day Two (Thursday 24th February, 2022)

- Day two will begin with a further number of speakers delivering a diverse range of lectures with opportunity for Q&A.
- Delegates will then be organised into one of two interactive workshops; these workshops will be delivered concurrently.

Chairs

- **Dr Anna Sarkozy, Great Ormond Street Hospital (GOSH), UK**
- **Dr Emily Oates, University of New South Wales, Australia**
- **Dr Nicol Voermans, Radboud University Medical Centre, Netherlands**
- **Dr Jim Dowling, Hospital for Sick Children, Canada**

PRE-RECORDED SESSIONS

These pre-recorded webinars will be made available to delegates in advance of the live masterclass.

PRE-RECORDED VIDEO - DURATION	PROGRAMME SESSION	SPEAKER DETAILS
30 mins	Physiotherapy	Tina Duong, USA Stanford University
30 mins	Genetics -Exome -Genome -Panel	Mark Davis, Australia PathWest Laboratory Medicine
30 mins	What is not CM?	TBC
30 mins	General care for patients with CM	Reghan Foley, USA National Institute of Neurological Disorders and Stroke, NIH

Masterclass (Day 1)

- Greenwich Mean Time, UK (GMT) – 09:00 – 14:05 (5 hours 5 minutes)
- Central European Time (CET) 10:00 – 15:05 (5 hours 5 minutes)

DAY 1 Wednesday 23 rd February 2022				
DURATION	GMT	CET	PROGRAMME SESSION	SPEAKER DETAILS
5 mins	09:00-09:05	10:00-10:05	Co-Chair Introduction & Welcome	Co-chairs
10 mins	09:05-09:15	10:05-10:15	Patient Perspectives <i>A patient/parental perspective of life with Congenital Myopathies</i>	TBC
10 mins	09:15-09:25	10:15-10:25	Patient Perspectives <i>A patient/parental perspective of life with Congenital Myopathies</i>	TBC
5 mins	09:25-09:30	10:25-10:30	Q & A	
25 mins	09:30 - 09:55	10:30-10:55	Genetic & Pathologic overview of CM	Johann Bohm, France French National Institute of Health and Medical Research (Inserm)
5 mins	09:55-10:00	10:55-11:00	Q & A	
25 mins	10:00 – 10:25	11:00-11:25	Muscle Biopsy based approach of CM	Edoardo Malfatti, France Reference Centre for Neuromuscular Diseases
5 mins	10:25-10:30	11:25-11:30	Q & A	
15 mins	10:30 - 10:45	11:30-11:45	BREAK	
25 mins	10:45-11:10	11:45-12:10	Thin Filament of Disease Mechanisms	Coen Ottenheijm, Netherlands Amsterdam University Medical Centre
5 mins	11:10 - 11:15	12:10-12:15	Q & A	
25 mins	11:15-11:40	12:15-12:40	Pathomechanisms 2: Excitation Contraction Coupling	Susan Treves, Switzerland Basel University Hospital
5 mins	11:40-11:45	12:40-12:45	Q & A	
15 mins	11:45-12:00	12:45-13:00	ACTA1 <i>(including 5 mins Q&A)</i>	Nigel Laing, Australia University of Western Australia
15 mins	12:00-12:15	13:00-13:15	TTN <i>(including 5 mins Q&A)</i>	Emily Oates, Australia University of New South Wales

20 mins	12:15-12:35	13:15-13:35	BREAK	
15 mins	12:35-12:50	13:35-13:50	NEB <i>(including 5 mins Q&A)</i>	Carina Wallgren Petterson, Finland University of Helsinki
25 mins	12:50-13:15	13:50-14:15	RYR1 <i>(including 5 mins Q&A)</i>	Heinz Jungbluth, UK Guys & St Thomas' NHS Trust
15 mins	13:15-13:30	14:15-14:30	MTM1 <i>(including 5 mins Q&A)</i>	Laurent Servais, France University of Oxford
15 mins	13:30-13:45	14:30-14:45	SEPN <i>(including 5 mins Q&A)</i>	Ana Ferreira, France French National Institute of Health and Medical Research (Inserm)
15 mins	13:45-14:00	14:45-15:00	DNM2 <i>(including 5 mins Q&A)</i>	Jim Dowling, Canada Hospital for Sick Children
5 mins	14:00-14:05	15:00-15:05	Close of day one	Co-Chairs

Virtual Masterclass (Day 2)

- British Summer Time (BST) – 11:00– 16:30 (5 hours 30 minutes)
- Central European Time (CET) 12:00 – 17:30 (5 hours 30 minutes)

DAY 2 Thursday 24th February 2022

DURATION	GMT	CET	PROGRAMME SESSION	SPEAKER
5 mins	11:00-11:05	12:00-12:05	Co-Chair Introduction & Welcome	Co-chairs
25 mins	11:05-11:30	12:05-12:30	Clinical Approach to CM	Heinz Jungbluth, UK Guys & St Thomas' NHS Trust
5 mins	11:30-11:35	12:30-12:35	Q & A	
25 mins	11:35-12:00	12:35-13:00	Adult presentation and consideration of CM.	Nicol Voermans, Netherlands Radboud University Medical Centre
5 mins	12:00-12:05	13:00-13:05	Q & A	
25 mins	12:05 - 12:30	13:05-13:30	Diagnostic approach to CM including clinical/genetic imaging	TBC
5 mins	12:30-12:35	13:30-13:35	Q & A	
25 mins	12:35-13:00	13:35 - 14:00	BREAK	
25 mins	13:00-13:25	14:00 - 14:25	The genetically undiagnosed patient	TBC

5 mins	13:25-13:30	14:25 – 14:30	Q & A	
25 mins	13:30-13:55	14:30 - 14:55	Emerging Therapies -Mechanistic view	Jocelyn Laporte, France French National Institute of Health and Medical Research (Inserm)
5 mins	13:55 - 14:00	14:55 - 15:00	Q & A	
25 mins	14:00-14:25	15:00 - 15:25	Emerging Therapies -In trial & Trial Readiness	Payam Mohassel, USA National Institute of Health (NIH)
5 mins	14:25-14:30	15:25 - 15:30	Q & A	
1 hour 55 mins (a short break will be included during the workshop)	14:30-16:25	15:30 - 17:25	<p>Workshop 1: Care Considerations.</p> <p>Workshop hosts:</p> <ul style="list-style-type: none"> -A. Reghan Foley, NIH, USA -Marion Main, GOSH, UK -Juan Kaski, GOSH, UK -Federica Trucco, UK -Sandra Donkovort, USA -Anna Sarkozy, GOSH, UK <p>The workshop will cover the following themes:</p> <ul style="list-style-type: none"> -Diagnostics -General Neuromuscular Management -Physiotherapy -Cardiology -Respiratory -Genetic Counselling 	<p>Workshop 2: Trial readiness, registries and data.</p> <p>Workshop hosts:</p> <ul style="list-style-type: none"> - Payam Mohassel, NIH, USA -Laurent Servais, Inserm, France -Tina Duong, Stanford, USA <p>The workshop will cover the following themes:</p> <ul style="list-style-type: none"> -Standardised Assessment and outcomes -What is Trial Readiness (TREAT-NMD) -Drug development process -Registries and common data elements -Need for natural history data
5 mins	16:25-16:30	17:25-17:30	Closing of Masterclass	Co-chairs