

Event programme

15th UK C.005 Day 1 –

15th April 2022

Poster viewing:

Introduction
Professor Michael Hanna

Session 1:

Professor Mary Reilly Professor Volker Straub

Development and Assessment of Novel biomarkers for Charcot-Marie-Tooth Disease
Professor John Svaren

Update on methods development in rare disease diagnostics for Mendelian myopathies
Dr Vijay Ganesh

The relevance of Smart Technologies for patient reported outcomes/remote assessments
Dr Elin Haf Davies

Flash poster sessions
Dr Anna Sarkozy Professor Rita Horvath

Loss of function mutations in DNAJB4 cause a myopathy with early respiratory failure
Dr Ana Topf

REN001, a novel peroxisome proliferator-activated receptor delta (PPAR δ) agonist, improves gait in patients with primary mitochondrial myopathy after 12 weeks
Dr Lisa Alcock

A Cross-sectional Analysis Of Spinal MRI Findings In UK hATTR
Dr Luke O' Donnell

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Day 2 – Weds 27th April 2022

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