

ePoster number	Presenting author	Paper title	Presentation order	ePoster screen	Theme	Moderator
4.001eP	David Zocche	Molecular spectrum and exploratory genotype-phenotype correlations in a prospective natural history cohort of nemaline myopathy	1	Screen 1	CMyo - Clinical care and case reports	Jan Kirschner
4.002eP	Ella Nippala	Investigating genetically unresolved cases of congenital fibre-type disproportion by a combination of nanopore sequencing and fibre-type analysis	2	Screen 1	CMyo - Clinical care and case reports	Jan Kirschner
4.003eP	Minobu Shichiji	Two boys with X-linked myotubular myopathy presenting with peliosis hepatis after infancy	3	Screen 1	CMyo - Clinical care and case reports	Jan Kirschner
4.04eP	Merve Yavuz	Marinesco-Sjögren syndrome: a case report of three siblings due to a novel mutation in the SIL1 gene	4	Screen 1	CMyo - Clinical care and case reports	Jan Kirschner
4.005eP	Jorge Alonso-Pérez	Novel Biallelic CRELD1 variants associated with early-onset myopathy and epilepsy.	5	Screen 1	CMyo - Clinical care and case reports	Jan Kirschner
4.006eP	Sibel Öz Yildiz	Phenotypic variability in COL12A1-related myopathy: a case series highlighting genotype-phenotype correlations	6	Screen 1	CMyo - Clinical care and case reports	Jan Kirschner
4.007eP	Hilde Braakman	Risk of acute liver failure in myopathic patients following therapeutic acetaminophen doses: a review of underrecognized risks	7	Screen 1	CMyo - Clinical care and case reports	Jan Kirschner
4.008eP	Emna Farhat	Dusty core myopathy related to heterozygous RYR1 gene mutation: description of a first north African family	8	Screen 1	CMyo - Clinical care and case reports	Jan Kirschner
4.009eP	Byeong Joo Choi	A Korean case of childhood-onset MEGF10-related myopathy with novel compound heterozygous variants	9	Screen 1	CMyo - Clinical care and case reports	Jan Kirschner
4.010eP	Vilma-Lotta Lehtokari	Nemaline cardiomyopathy caused by a missense variant in ACTA1 with a RYR2 variant as a possible genetic modifier	10	Screen 1	CMyo - Clinical care and case reports	Jan Kirschner
4.011eP	Kara Farstad-O'Halloran	In vitro characterisation of mesoporous silica nanoparticles (MSNs) as drug delivery agents in patient-derived models of RYR1-related myopathy	11	Screen 1	CMyo - Clinical physiopathology	Jan Kirschner
4.012eP	Kathryn Yammine	Gene replacement therapy for ADSS1 myopathy: preclinical investigation in mice	12	Screen 1	CMyo - Clinical physiopathology	Jan Kirschner
4.015eP	Hirofumi Teranishi	The epidemiology of congenital myasthenic syndromes in Japan: an exploration of diagnosed and genetic prevalence	1	Screen 2	Congenital myasthenic syndrome	Göknur Haliloğlu
4.016eP	Akram Alnajati	Heterozygous DOK7-related congenital myasthenic syndrome responsive to salbutamol: a case series from Iraq	2	Screen 2	Congenital myasthenic syndrome	Göknur Haliloğlu
4.017eP	Ercan Demir	Epidermolysis bullosa simplex with muscular dystrophy and myasthenia: a case report with plectinopathy	3	Screen 2	Congenital myasthenic syndrome	Göknur Haliloğlu
4.028eP	Josefine Jul Jarbæk Nielsen	Statin use preceding myasthenia gravis diagnosis: a nationwide case-control study in Denmark	4	Screen 2	Myasthenia gravis	Göknur Haliloğlu
4.029eP	Arnaud Jacquier	Live cell-based diagnostic assay for the detection of low-affinity anti-AChR antibodies in autoimmune myasthenia gravis in France	5	Screen 2	Myasthenia gravis	Göknur Haliloğlu
4.030eP	Hamdi Necdet Karli	Clinical and demographic characteristics of patients with very late onset myasthenia gravis	6	Screen 2	Myasthenia gravis	Göknur Haliloğlu
4.031eP	Lukas Theissen	A simplified murine model of experimental autoimmune myasthenia gravis (EAMG)	7	Screen 2	Myasthenia gravis	Göknur Haliloğlu
4.032eP	Madeleine Rzepa Hopkirk	A national study on juvenile myasthenia gravis in Denmark: incidence rate, onset and treatment	8	Screen 2	Myasthenia gravis	Göknur Haliloğlu
4.033eP	Ahmad Elmesallami	Clinical spectrum, management and outcomes of myasthenia gravis in Oman: a retrospective observational cohort study in a single tertiary center	9	Screen 2	Myasthenia gravis	Göknur Haliloğlu
4.034eP	Patimat Isabekova	Very late-onset myasthenia gravis: A Single-Center Experience	10	Screen 2	Myasthenia gravis	Göknur Haliloğlu
4.035eP	Patimat Isabekova	Predictors of Adverse Outcomes in Myasthenia Gravis: A Single-Center Cohort	11	Screen 2	Myasthenia gravis	Göknur Haliloğlu
4.036eP	Patimat Isabekova	Juvenile Myasthenia Gravis: an unusual presentation	12	Screen 2	Myasthenia gravis	Göknur Haliloğlu
4.037eP	Niklas Huntemann	Target antigen switch from CD19 to BCMA in sequential bispecific T-cell engager therapy for refractory Myasthenia gravis-Lambert-Eaton overlap syndrome	1	Screen 3	Myasthenia gravis	Jean-yves Hogrel
4.039eP	Nikoletta Nikolenko	Magnetoencephalography and cognition in myotonic dystrophy type 1	2	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.040eP	Eva Coll Liesa	Comparison between long-read genome sequencing technologies and the gold-standard small pool PCR in the study of myotonic dystrophy type 1	3	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.041eP	Hüseyin Per	Retrospective evaluation of clinical findings, prognosis, electrophysiological studies, and genetic results in patients with myotonia	4	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.042eP	Aaron Novack	HARMONIA study design: a global phase 3 trial assessing the efficacy and safety of z-basivarsen in myotonic dystrophy type 1 (DM1)	5	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.043eP	Nikoletta Nikolenko	Patient-reported experiences, preferences and challenges within palliative and end-of-life care for myotonic dystrophy: a UK community survey	6	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.044eP	Paul Streck	Data from the first cohort of FREEDOM2-DM1 multiple-ascending dose study of PGN-EDODM1 support continued development	7	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.045eP	Guillaume Bassez	Developing a disease progression model for individuals living with myotonic dystrophy type 1	8	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.046eP	José Verdú Díaz	Longitudinal muscle MRI as an outcome measure in myotonic dystrophy type 1: quantitative fat fraction, STIR biomarkers and radiomics	9	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.047eP	Julie Bohill	The UK myotonic dystrophy patient registry: empowering clinical research and patient voice with an effective translational research tool	10	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.048eP	Fatma Munevver Gokyigit	Clinical and therapeutic evaluation of patients with myotonic disorders: a single-center retrospective study	11	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.049eP	Wander Jacco Adrian Aarts	Apathy in myotonic dystrophy type 1: prevalence, subtype differences and clinical correlates	12	Screen 3	Myotonic disorders and Periodic Paralysis	Jean-yves Hogrel
4.050eP	Ridhi Parasrampur	Pharmacokinetic and pharmacodynamic translation of ENTR-601-44, an investigational therapy for patients with exon 44 skip-amenable DMD	1	Screen 4	Myotonic disorders and Periodic Paralysis	Antonio Lauletta
4.051eP	Yoshihiko Inuzuka	Clinical burden of myotonic dystrophy type 1: targeted review of mortality/morbidity and associated multisystemic impacts	2	Screen 4	Myotonic disorders and Periodic Paralysis	Antonio Lauletta
4.052eP	Najoua El Boujnouni	Mitochondrial dysfunction in myotonic dystrophy type 1	3	Screen 4	Myotonic disorders and Periodic Paralysis	Antonio Lauletta
4.053eP	Julia Gusatovic	Initial MRI and real-life wearable data from a 2-year natural history study of myotonic dystrophy type 1	4	Screen 4	Myotonic disorders and Periodic Paralysis	Antonio Lauletta
4.054eP	Dong Won Kwack	Early severe cardiac involvement in Myotonic Dystrophy type 1: case series	5	Screen 4	Myotonic disorders and Periodic Paralysis	Antonio Lauletta
4.055eP	David Bovenkerk	Atrial and ventricular pacing proportions in patients with myotonic dystrophy type 1	6	Screen 4	Myotonic disorders and Periodic Paralysis	Antonio Lauletta
4.056eP	Basil Van Veen	Liver enzyme profiles and fibrosis assessment in myotonic dystrophy type 1: a multicenter observational study	7	Screen 4	Myotonic disorders and Periodic Paralysis	Antonio Lauletta
4.057eP	Leandre La Fontaine	Development of the Rasch-built Myotonic Dystrophy type 2 activity and participation scale (DM2-Activ)	8	Screen 4	Myotonic disorders and Periodic Paralysis	Antonio Lauletta
4.058eP	Rebekka Bernhard	Hyperkalemic periodic paralysis: cross-sectional quantification of muscle fat replacement using MRI	9	Screen 4	Myotonic disorders and Periodic Paralysis	Antonio Lauletta
4.059eP	Annica Enegaard	The effect of progressive resistance training in persons with hypokalemic periodic paralysis	10	Screen 4	Myotonic disorders and Periodic Paralysis	Antonio Lauletta

4.060eP	Charlotte Handberg	Understanding barriers to participate in specialized neuromuscular rehabilitation experienced by people with neuromuscular diseases	1	Screen 5	Registries, networks and care	Gauthier Remiche
4.061eP	Emma Heslop	Exploring patient and research site perspectives on the implementation of home nursing in Duchenne muscular dystrophy clinical trial protocols	2	Screen 5	Registries, networks and care	Gauthier Remiche
4.062eP	Lucy Hickson	Diagnostic journeys in genetic muscle disorders: a scoping review of delays and barriers to genetic diagnosis	3	Screen 5	Registries, networks and care	Gauthier Remiche
4.063eP	Elizabeth Lynne Wood	Impact of local neuromuscular clinic access on patient travel distance, costs, and work loss: A case for a community clinic's role in multidisciplinary care	4	Screen 5	Registries, networks and care	Gauthier Remiche
4.065eP	Sara Loprieno	Transitioning from pediatric to adult care in neuromuscular diseases: a management model in the Tuscany region, Italy	5	Screen 5	Registries, networks and care	Gauthier Remiche
4.066eP	Edoardo Malfatti	ERN EURO-NMD AI PathologyHub: Supporting the biological classification of H&E-stained muscle biopsy slides in neuromuscular disease	6	Screen 5	Registries, networks and care	Gauthier Remiche
4.067eP	Jennifer Levy	The LGMD2A/calpainopathy registry: a patient-powered natural history study and trial recruitment tool	7	Screen 5	Registries, networks and care	Gauthier Remiche
4.068eP	Himshweta Balwani	From fragmentation to collaboration: building a multidisciplinary neuromuscular ecosystem in India	8	Screen 5	Registries, networks and care	Gauthier Remiche
4.069eP	Bayram Sirri	Evaluation of trunk control in neuromuscular diseases: a systematic review of measurement properties of trunk-specific instruments	9	Screen 5	Registries, networks and care	Gauthier Remiche
4.072eP	Elizabeth Lynne Wood	Leveraging partnerships between small and large care centers to provide telehealth consultation improves access and equity	10	Screen 5	Registries, networks and care	Gauthier Remiche
4.064eP	Julie Bohill	The UK facioscapulohumeral muscular dystrophy patient registry: a powerful tool to support clinical research and patient voice in the translational research pa	1	Screen 6	Registries, networks and care	Rhonda Taylor
4.070eP	Brett Kinsler	Relative Patient Burden in Patient-Reported Outcomes for Neuromuscular Clinical Trials: Incorporating Relevance and Ease of Completion Beyond Time Alone	2	Screen 6	Registries, networks and care	Rhonda Taylor
4.071eP	Masatoshi Ishizaki	Educational booklet for health management of caregivers of muscular dystrophy patients in Japan	3	Screen 6	Registries, networks and care	Rhonda Taylor
4.073eP	Sneha Banavara Shyamprasad	Diagnostic odyssey and its impact on caregivers in a progressive neuromuscular disorder: A qualitative study with quantitative insights	4	Screen 6	Registries, networks and care	Rhonda Taylor
4.074eP	Esther Brusse	Lifestyle interventions in muscle disease: a systematic review of exercise and dietary programs in myopathies	5	Screen 6	Registries, networks and care	Rhonda Taylor
4.075eP	Catherine Turner	A multi-disciplinary education programme for healthcare professionals in care and research for Duchenne muscular dystrophy: Duchenne Education in Care an	6	Screen 6	Registries, networks and care	Rhonda Taylor
4.077eP	Takahiro Nakayama	The promotion of health and employment support for the patients with muscular dystrophies	7	Screen 6	Registries, networks and care	Rhonda Taylor
4.078eP	Julie Bohill	Global FKRP registry: data trends and highlights from over 1,000 participants	8	Screen 6	Registries, networks and care	Rhonda Taylor
4.079eP	Miriam Rodrigues	TREAT-NMD Global registry network: an insight into a large global neuromuscular patient dataset	9	Screen 6	Registries, networks and care	Rhonda Taylor
4.080eP	Julie Bohill	Myotubular and centronuclear myopathy: analysis of registry data to understand current nutritional practices and explore potential associations between liver	10	Screen 6	Registries, networks and care	Rhonda Taylor
4.076eP	Michael Bond	From user centred design to participatory co-design in neuromuscular assistive technologies	1	Screen 7	Registries, networks and care	Volunteer
4.081eP	Rebecca Leary	PaLaDin – Improving Decision Making in Neuromuscular Diseases (NMDs). What are the benefits of using the Interactium data platform?	2	Screen 7	Registries, networks and care	Volunteer
4.082eP	Michael Bond	Duchenne UK connect: a digital patient platform and app supporting trial readiness, research participation and care planning in Duchenne muscular dystrophy	3	Screen 7	Registries, networks and care	Volunteer
4.083eP	Michio Kobayashi	Webinar on the health of caregivers for patients with muscular dystrophy	4	Screen 7	Registries, networks and care	Volunteer
4.084eP	Emily Reuben OBE	HERCULES use of lived experience to inform a disability-based model of the cost of Duchenne muscular dystrophy (DMD)	5	Screen 7	Registries, networks and care	Volunteer
4.085eP	Sajin Mathew	Long-term survival, quality-adjusted life-years, and economic burden in limb-girdle muscular dystrophy type 2I/R9: a health outcomes model	6	Screen 7	Registries, networks and care	Volunteer
4.086eP	Geir Hevnskjel Ringvold	"Neuromuscular disorders in a nutshell": an innovative way of visualizing clinical summaries for rare neuromuscular diseases	7	Screen 7	Registries, networks and care	Volunteer
4.087eP	Nicol Voermans	FSHD Europe: Strengthening the European FSHD ecosystem: bridging clinical care, research, and patient-centered outcomes	8	Screen 7	Registries, networks and care	Volunteer
4.088eP	Jacopo Luca Casiraghi	Sexual health in neuromuscular disease: knowledge, attitudes and practices of a multidisciplinary team: the E.M.B.R.A.C.E. study	9	Screen 7	Registries, networks and care	Volunteer
4.013eP	Cara Timpani	AI-driven drug repurposing for ADSS1 myopathy using cross-cohort skeletal muscle transcriptomic signatures	1	Screen 8	CMYo - Clinical physiopathology	Grace Yoon
4.014eP	Dean Campelj	Characterization of adenylosuccinate synthase 1 deficiency in mouse models of inherited myopathy	2	Screen 8	CMYo - Clinical physiopathology	Grace Yoon
4.018eP	Mary Foltz	Defining MRI-based biomarkers for separating muscle fat neogenesis and atrophy in FSHD	3	Screen 8	Muscle imaging	Grace Yoon
4.019eP	Yukako Yae	Muscle imaging features in tubular aggregate myopathies associated with STIM1 and ORAI1 variants	4	Screen 8	Muscle imaging	Grace Yoon
4.020eP	Yoshihiko Saito	MRI-Based Assessment of Masticatory Muscle Involvement in Hereditary Myopathies	5	Screen 8	Muscle imaging	Grace Yoon
4.021eP	Max Hahn	High-throughput 3D light-sheet microscopy of intact mouse hindlimbs enables skeletal muscle morphometrics in rodent myopathy models	6	Screen 8	Muscle imaging	Grace Yoon
4.022eP	Cecilie Aronsen Haestad	Quantitative muscle ultrasound as a new imaging technique: steps toward clinical implementation in Norway	7	Screen 8	Muscle imaging	Grace Yoon
4.023eP	Jens Reimann	How useful is muscle MRI in the assessment and differential diagnosis of recessive myotonia congenita (Becker)?	8	Screen 8	Muscle imaging	Grace Yoon
4.024eP	Maja Olsen	Evaluation of 2-point, 3-point and 6-point Dixon MRI for quantification of muscle fat replacement in patients with muscular dystrophy	9	Screen 8	Muscle imaging	Grace Yoon
4.025eP	Jenni Mäki-Jouppila	Orchidectomy-induced osteosarcopenia in aged rats: a preclinical model for muscle research	10	Screen 8	Muscle imaging	Grace Yoon
4.026eP	Jenni Mäki-Jouppila	Non-invasive imaging techniques for the analysis of muscle, bone, and fat tissues in ovariectomized mice	11	Screen 8	Muscle imaging	Grace Yoon
4.027eP	Ryoko Inutsuka	Atypical skeletal muscle MRI pattern involving the iliopsoas and the short head of the biceps femoris in Stormorken syndrome	12	Screen 8	Muscle imaging	Grace Yoon