

ePoster number	Presenting author	Poster title	Presentation order	ePoster screen	Theme	Moderator
1.009eP	Svetlana Gorokhova	Studying CAPN3 variant co-inheritance patterns to gain insight into genetic landscape of calpainopathy	1	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.010eP	Reoto Ueda	Comparison of muscle involvement in anoctaminopathy and dysferlinopathy	2	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.011eP	Raehan Kusumah	Calpainopathy: clinical, electrophysiological, and genetic characterization of three Indonesian patients	3	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.012eP	Toshiki Ochi	Genotype-phenotype correlation in calpain 3-related myopathy: a case series of early-onset and late-onset LGMD	4	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.013eP	Pengfei Lin	Novel POMT2 variants associated with limb-girdle muscular dystrophy R14: genetic, histological and functional studies	5	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.014eP	Özlem Hergüner	A retrospective analysis of patients with sarcoglycanopathy from a single tertiary center	6	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.015eP	Özlem Hergüner	Beyond limb-girdle weakness: a single center experience with calpainopathy	7	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.016eP	Atsuko Tsuneyama	A 48-year-old man with limb-girdle muscular dystrophy type D1 presenting with neurogenic changes and fasciculations	8	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.017eP	Meirong Liu	Clinical and genetic analysis of LGMD R1 caused by novel non-canonical CAPN3 splicing mutations	9	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.018eP	Ayşe Aksoy	HMGR-related limb-girdle muscular dystrophy: case report	10	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.019eP	Kjell Arne Arntzen	FKRP-related limb-girdle muscular dystrophy R9 caused by a novel intronic variant	11	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.020eP	Emilie Groulx Boivin	Homozygous 13q12.12 contiguous gene deletion causing concurrent LGMD2C and ARSACS: a blended neuromuscular disorder	12	Screen 1	LGMD - Clinical care and case reports	Jan De Bleecker
1.021eP	Carla Florencia Bolaño Diaz	Longitudinal behaviour of NSAD and muscle fat fraction as monitoring outcome measures in LGMD R2-DYSF related	1	Screen 2	LGMD - Imaging and outcome measures	Coen Ottenheijm
1.022eP	Heather Gordish-Dressman	Clinical trial readiness in dysferlinopathy from the 10-year, multicenter Jain clinical outcome study	2	Screen 2	LGMD - Imaging and outcome measures	Coen Ottenheijm
1.023eP	Sajin Mathew	Systematic literature review of clinical outcomes and disease burden in limb-girdle muscular dystrophy subtype 2	3	Screen 2	LGMD - Imaging and outcome measures	Coen Ottenheijm
1.024eP	Corrado Angelini	Clinical protocol proposal for LGMD-D2: validation in a patient's series.	4	Screen 2	LGMD - Imaging and outcome measures	Coen Ottenheijm
1.025eP	Chand Nagpal	Muscle MRI patterns in dysferlinopathy patients: a case series	5	Screen 2	LGMD - Imaging and outcome measures	Coen Ottenheijm
1.026eP	Anne Katrine Kjemsø Jensen	Alpha-dystroglycan glycosylation in heterozygous FKRP carriers of LGMDR9	6	Screen 2	LGMD - Pathophysiology of NMDs	Coen Ottenheijm
1.027eP	Weiyan Hou	Pathological and transcriptomic comparison of Dysferlinopathy and Immune-mediated necrotizing myopathy	7	Screen 2	LGMD - Pathophysiology of NMDs	Coen Ottenheijm
1.028eP	Jinhong Shin	Calcium modulation improves motor function in a mouse model of dysferlinopathy	8	Screen 2	LGMD - Pathophysiology of NMDs	Coen Ottenheijm
1.029eP	Carlos Sanchez Riera	HUMAN IPS-DERIVED CARDIOMYOCYTES TO STUDY BETA-SARCOGLYCANPATHY	9	Screen 2	LGMD - Pathophysiology of NMDs	Coen Ottenheijm
1.030eP	Marielena Schiava	Longitudinal comparison of creatinine- and cystatin C-based eGFR in LGMDR2	10	Screen 2	LGMD - Treatments	Coen Ottenheijm
1.031eP	Gaoyuan Li	Correction of aberrant splicing caused by intronic CAPN3 mutations using RNA-targeted therapeutic strategies in mice	11	Screen 2	LGMD - Treatments	Coen Ottenheijm
1.059eP	Milla Laarne	Application of nanopore sequencing for the diagnosis of muscle disorders	12	Screen 2	Novel molecular genetic approaches for NMD	Coen Ottenheijm
1.089eP	Lisa Edel	Functional Outcomes in Infants with Spinal Muscular Atrophy (SMA) Type 1: Motor, Respiratory, and Bulbar Domain	1	Screen 3	SMA - Imaging and outcome measures	Julian Sanchez-Dal Cin
1.090eP	Felipe Franco Da Graca	Facial nerve diameter as a potential biomarker in adults with spinal muscular atrophy	2	Screen 3	SMA - Imaging and outcome measures	Julian Sanchez-Dal Cin
1.091eP	Emer O'Reilly	Correlating EK2 and RULM to assess upper limb function in SMA	3	Screen 3	SMA - Imaging and outcome measures	Julian Sanchez-Dal Cin
1.092eP	Allison Grangé	User experience in a home-based exergame in youth with spinal muscular atrophy: preliminary findings from a pilot study	4	Screen 3	SMA - Imaging and outcome measures	Julian Sanchez-Dal Cin
1.093eP	Toshio Saito	Evaluation of motor function in patients with spinal muscular atrophy using finger movements	5	Screen 3	SMA - Imaging and outcome measures	Julian Sanchez-Dal Cin
1.094eP	Claudia Mazzà	Smartphone-based two- versus six-minute walk test for remote assessment of gait	6	Screen 3	SMA - Imaging and outcome measures	Julian Sanchez-Dal Cin
1.095eP	Meghan Moore Burk	Translation and cross-cultural adaption of the functional ability scale for evolving spinal muscular atrophy (EVO-FAST)	7	Screen 3	SMA - Imaging and outcome measures	Julian Sanchez-Dal Cin
1.096eP	Sneha Banavara Shyamprasad	Telemedicine enabled continuum of care in Spinal Muscular Atrophy	8	Screen 3	SMA - Imaging and outcome measures	Julian Sanchez-Dal Cin
1.097eP	Julie Bohill	The integration of PROMs and real-world data: a holistic approach to characterise disease burden and treatment outcomes	9	Screen 3	SMA - Imaging and outcome measures	Julian Sanchez-Dal Cin
1.098eP	Ayşe Ipek Polat	Investigation of long non-coding RNA profiles derived from peripheral blood mononuclear cells in spinal muscular atrophy	10	Screen 3	SMA - Pathophysiology	Julian Sanchez-Dal Cin
1.099eP	Marie Machiels	Neurofilament study: assessment of neurofilament levels in premature newborns	11	Screen 3	SMA - Pathophysiology	Julian Sanchez-Dal Cin
1.100eP	Jing Marantz	Clinical pharmacology of apitegromab in spinal muscular atrophy	12	Screen 3	SMA - Treatments	Julian Sanchez-Dal Cin
1.037eP	Tarun Arora	Standing balance improvements with additional sensory input in Charcot-Marie-Tooth	1	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.038eP	Evelin Milev	The progression of ankle range of motion in paediatric Charcot Marie Tooth (CMT): implications for function and quality of life	2	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.039eP	Aysylu Murtazina	Targeted search for the second variant in patients with SORD-neuropathy	3	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.040eP	Can Ebru Kurt	Alpha-synuclein expression in skeletal muscle of amyotrophic lateral sclerosis patients: an immunohistochemical study	4	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.041eP	Serkan Kirik	ALS2-related pediatric motor neuron disease in six children	5	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.042eP	Yigit Can Guldiken	A pilot study on AI vs. physician-led counseling for newly diagnosed ALS and GBS patients	6	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.043eP	Didem Ardiçli	Childhood chronic inflammatory demyelinating polyneuropathy: a single-center case series	7	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.044eP	Camilla Hagedorn Thomsen	Rethinking relapse detection in neuropathy: integrating patient-reported outcomes and objective functional tests	8	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.045eP	Deniz Yüksel	Retrospective evaluation of pediatric Guillain-Barré syndrome according to 2023 european academy of neurology	9	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.046eP	Mahima Paul	Enhancing gait mechanics in paediatric Charcot Marie Tooth with lightweight carbon fiber ankle foot orthosis	10	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.047eP	Deniz Yüksel	RCC1-related acute-onset axonal neuropathy mimicking Guillain-Barré syndrome in two infants	11	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos
1.048eP	Deniz Yüksel	Clinical and genetic characteristics of pediatric hereditary neuropathies: a single-center experience	12	Screen 4	Neurogenic Disorders - Clinical care and case reports	Michelle Lorentzos

1.034eP	Heike Kölbel	Multilocus pathogenic variation in a pediatric patient: a quadruple molecular diagnosis shaping a complex phen	1	Screen 5	Multiorgan syndromes	Stephan Zuchner
1.035eP	Gökür Haliloğlu	Neonatal-onset myotonia further expanding the spectrum of SCN1A-related arthrogryposis multiplex congenita	2	Screen 5	Multiorgan syndromes	Stephan Zuchner
1.049eP	Abraham Al Jody	Diagnostic course of chronic inflammatory demyelinating polyneuropathy spectrum with distal predominant we	3	Screen 5	Neurogenic Disorders - Clinical care and case reports	Stephan Zuchner
1.050eP	Malin Eriksen	Evaluating muscle changes in persons with Charcot Marie Tooth disease using electrical impedance myography	4	Screen 5	Neurogenic Disorders - Clinical care and case reports	Stephan Zuchner
1.051eP	Sookin Moon	Creatine kinase elevation mimicking myopathy in amyotrophic lateral sclerosis: a case of clinical diagnostic chall	5	Screen 5	Neurogenic Disorders - Clinical care and case reports	Stephan Zuchner
1.052eP	Yigit Can Guldiken	C9orf72 hexanucleotide repeat expansion presenting as progressive muscular atrophy with frontotemporal dem	6	Screen 5	Neurogenic Disorders - Clinical care and case reports	Stephan Zuchner
1.053eP	Mustafa Cam	A case atypical Guillain-Barre syndrome	7	Screen 5	Neurogenic Disorders - Clinical care and case reports	Stephan Zuchner
1.054eP	Mustafa Cam	Advanced age Guillain Barre syndrome case	8	Screen 5	Neurogenic Disorders - Clinical care and case reports	Stephan Zuchner
1.055eP	Pola Gaid	Quantitative ALS: development of a novel, physician-reported, ALS outcome measure	9	Screen 5	Neurogenic disorders - Imaging and outcome measures	Stephan Zuchner
1.056eP	Joo Hye Sung	Muscle ultrasound feature-based predictive modeling for amyotrophic lateral sclerosis staging: machine learnin	10	Screen 5	Neurogenic disorders - Imaging and outcome measures	Stephan Zuchner
1.057eP	Camila Gonzalez Barral	Continuous real-world assessment of ambulation in amyotrophic lateral sclerosis using digital health technology	11	Screen 5	Neurogenic disorders - Imaging and outcome measures	Stephan Zuchner
1.058eP	Jean-yves Hogrel	Accelerating ALS trials through muscle-centered assessment	12	Screen 5	Neurogenic disorders - Imaging and outcome measures	Stephan Zuchner
1.060eP	Erin Welsh	Healthcare utilization and associated barriers among adults living with spinal muscular atrophy in the United Sta	1	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.061eP	Hacer Durmus Tekce	Real-world treatment with risdiplam in adults with spinal muscular atrophy (SMA): a single center experience	2	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.062eP	Charlotte Lilien	Advancing care in spinal muscular atrophy through patient-centred, motor-focused rehabilitation: Prospective i	3	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.063eP	Min Tsui Ong	Priorities in information sharing and management in spinal muscular atrophy (PRISM-SMA), a mixed methods st	4	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.064eP	Chamindra Laverty	The Natural History and Treatment Effects of SMA Patients with ≥4 SMN2 copies in the United States	5	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.065eP	Andres Eduardo Nascimento Osorio	Impact of early treatment initiation on ambulatory outcomes in SMA type 3a: insights from Spanish and Italian r	6	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.066eP	Esra Sarigecili	Case Report: Phenotypic diversity in a family with SMN1 homozygous deletion and 4 copies of SMN2	7	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.067eP	Sema Saltik	Evaluation of motor function in patients with SMA types 2 and 3 receiving disease-modifying therapy: a single-ce	8	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.068eP	Katie Obrien	Prospective study protocol investigating nutrition, bulbar and motor function in infants with spinal muscular atr	9	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.069eP	Sara Loprieno	Beyond the floor effect: long-term respiratory stabilization with risdiplam in severe adult SMA type 2, a single ce	10	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.070eP	Ayşe Nur Özdamar Acarlı	The rarest of the rare: a case series of spinal muscular atrophy with lower extremity predominance type 1 (SMA	11	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.071eP	Anna Lemska	Clinical characteristics and treatment outcomes in patients with spinal muscular atrophy and four SMN2 copies	12	Screen 6	SMA - Clinical care and case reports	Tamara Dangouloff
1.072eP	Vanessa Christie-Brown	SMA Care UK: optimising bone health for individuals with SMA across the UK	1	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.073eP	Vanessa Christie-Brown	SMA Care UK: management of hips in SMA in the DMT era - UK multidisciplinary consensus recommendations	2	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.074eP	Anna Lia Frongia	Rehabilitation needs and care models in spinal muscular atrophy: a real-world analysis from an Italian communi	3	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.075eP	Charlotte Lilien	Developing exercise guidelines for spinal muscular atrophy: An SMA Care UK initiative	4	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.076eP	Tanitnun Paprad	Real-world Outcomes of Early Gene Therapy for SMA Following Newborn Screening.	5	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.077eP	Svitlana Kormoz	Nationwide newborn screening for spinal muscular atrophy during active military conflict: the Ukrainian implem	6	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.078eP	Julie Bohill	Adult SMA REACH: characterisation of adult patients living with SMA at start of treatment	7	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.079eP	Min Tsui Ong	The use of spinal bracing in severe spinal muscular atrophy: a multicentre audit	8	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.080eP	Julie Bohill	Adult SMA REACH: a comparative UK real world data study to assess the safety and efficacy profiles of nusiners	9	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.081eP	Julie Bohill	UK real-world data collection study to analyse respiratory trajectories in SMA patients undergoing treatment wi	10	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.082eP	Revital Lavi	Motor trajectories after risdiplam following gene therapy in SMA type 1	11	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.083eP	Begimay Zholdasbekova	Motor outcomes of nusinersen therapy in spinal muscular atrophy type 2: analysis of the national cohort from K	12	Screen 7	SMA - Clinical care and case reports	Margherita Milone
1.032eP	Berta Estévez-Arias	Divergent limb phenotypes associated with PITX1 alterations: from lower limb malformations to Liebenberg syn	1	Screen 8	Multiorgan syndromes	TBC
1.033eP	Berta Estévez-Arias	Expanding the phenotypic spectrum of GLDN-related disorders: survival beyond the neonatal period and genoty	2	Screen 8	Multiorgan syndromes	TBC
1.036eP	Nicolas Chrestian	A case of severe neonatal hypotonia with IPEX myositis cured after hematopoietic stem cell transplantation	3	Screen 8	Multiorgan syndromes	TBC
1.084eP	Gauri Khandekar	Changes in functional mobility following generic risdiplam spinal muscular atrophy: a 6 month observational stu	4	Screen 8	SMA - Clinical care and case reports	TBC
1.085eP	Yun Jeong Lee	Phenotypic discordance and near-presymptomatic gene therapy in dizygotic twins with SMA Type 1: evidence fo	5	Screen 8	SMA - Clinical care and case reports	TBC
1.086eP	Deniz Yüksel	Beyond the classical phenotype: a homozygous ASAH1 variant presenting with juvenile-onset myoclonic epileps	6	Screen 8	SMA - Clinical care and case reports	TBC
1.087eP	Milos Brkusanin	When spinal muscular atrophy is there but the test says no: a case of somatic mosaicism leading to false-negati	7	Screen 8	SMA - Clinical care and case reports	TBC
1.088eP	Phisitphon Ounchokdee	"Double-trouble" with spinal muscular atrophy: diagnostic complexity and therapeutic implications	8	Screen 8	SMA - Clinical care and case reports	TBC
1.101eP	Stephanie Fradette	Phase 1 interim results evaluating salanersen safety, tolerability, pharmacokinetics, and exploratory efficacy for	9	Screen 8	SMA - Treatments	TBC
1.102eP	Claudia Chiriboga	Final results from the JEWELFISH study: 5-year risdiplam treatment in non-treatment-naïve individuals with spin	10	Screen 8	SMA - Treatments	TBC
1.103eP	Stephanie Fradette	The SOLAR study: phase 3 study to evaluate the efficacy and safety of salanersen in participants aged 15–60 yea	11	Screen 8	SMA - Treatments	TBC
1.104eP	Crystal Proud	STELLAR phase 3 studies to evaluate the efficacy and safety of salanersen in infants with spinal muscular atroph	12	Screen 8	SMA - Treatments	TBC