Full Programme

Wednesday 2nd October

		Room: Concert Hall
07.30		**Registration desk opens** and setting up of posters
08.30 - 09.00		**Congress opening - Message from the President Thomas Voit**
09.00 - 10.00		**Metabolic disturbances in neuromuscular diseases** **Invited lectures (I.1-2)** **Chairpersons: Nicol Voermans and John Vissing**
09.00 - 09.30	I.1	**A journey through the growing world of muscle glycogenosis** Pascal Laforêt, Nord/Est/Ile-de-France Neuromuscular center, Raymond-Poincaré teaching Hospital, AP-HP, France
09.30 - 10.00	I.2	**Skeletal muscle lipid metabolism – diagnostics, pathophysiology and treatment options** Mette Cathrine Ørngreen, Department of Adolescent and Childhood Medicine and Copenhagen Neuromuscular Center, Copenhagen University Hospital, Rigshospitalet, Denmark
10.00 - 10.30		**Morning refreshments** – Concert Hall Foyer
10.30 - 11.30		**Metabolic disturbances in neuromuscular diseases** **Invited lectures (I.3-4)** **Chairpersons: Nicol Voermans and John Vissing**
10.30 - 11.00	I.3	**Glycosylation related myopathies** Dirk Lefeber, Radboudumc Expertise Centre for Disorders of Glycosylation, Translational Metabolic Laboratory, Department of Neurology, Radboud university medical center, The Netherlands
11.00 - 11.30	I.4	**Physical exercise training in patients with neuromuscular disorders** Tina Dysgaard Jeppesen, Copenhagen Neuromuscular Center, Denmark
11.30 - 11.45		Break
11.45 - 13.00		**Metabolic disturbances in neuromuscular diseases (O.1-3; O5-6)** **Parallel** oral presentations 1 **- Concert Hall** **Chairpersons: Cornelia Kornblum and Ros Quinlivan**
11.45 - 12.00	O.1	**Bi-allelic mutations in COX6A2 cause a striated muscle-specific cytochrome c oxidase deficiency.** M. Inoue; S. Uchino; A. Iida; S. Noguchi; S. Hayashi; T. Takahashi; K. Fujii; H. Komaki; E. Takeshita; I. Nonaka; T. Yoshizawa; L. Van Lommel; F. Schuit; Y. Goto; M. Mimaki; I. Nishino
12.00 - 12.15	O.2	**Growth differentiation factor 15 is a valuable biomarker of therapeutic response for TK2 deficient myopathy:** C. Domínguez-González; C. Badosa; M. Madruga-Garrido; I. Martí; C. Paradas; C. Ortez; J. Diaz-Manera; C. Blázquez-Bermejo; Y. Cámara; R. Martí; F. Mavillard Saborido; M. Martin; E. Martín-Hernández; R. Montero; R. Artuch; S. Kalko; A. Nascimento; C. Jimenez-Mallebrera
12.15 - 12.30	O.3	**MiR-379 link glucocorticoid treatment to mitochondrial dysfunction in Duchenne muscular dystrophy.** M. Sanson; E. Massourides; V. Mournetas; A. Vu Hong; P. Bénit; I. Barthélémy; S. Blot; V. Mouly; C. Pinset; P. Rustin; I. Richard; D. Israeli
12.30 - 12.45	O.5	**A new glycogen storage disorder caused by a dominant mutation in the glycogen myophosphorylase gene (PYGM).** A. Echaniz-Laguna; X. Lornage; E. Edelweiss; P. Laforêt; B. Eymard; J. Vissing; J. Laporte; J. Böhm
12.45 - 13.00	O.6	**Pre-clinical development of SPK-3006, an investigational liver-directed AAV gene therapy for the treatment of Pompe disease** S. Armour; J. Nordin; H. Costa Verdera; D. Cohen; P. Sellier; M. Crosariol; F. Collaud; C. Riling; W. Quinn III; H. Hanby; U. Cagin; F. Puzzo; V. Haurigot; G. Ronzitti; P. Colella; X. Anguela; F. Mingozzi
13.00 - 14.30		**Lunch, exhibition and posters** ■ Andersen Hall, ■ Lumbye Hall and ■ Gemyse
13.30 - 15.00		
15.15 - 16.45		**Poster session 1** Poster area ■ Andersen Hall, ■ Lumbye Hall and ■ Gemyse
16.15 - 16.45		Afternoon refreshments, exhibition and posters ■ Andersen Hall, ■ Lumbye Hall and ■ Gemyse
16.45 - 18.15		**Poster session 2** Poster area ■ Andersen Hall, ■ Lumbye Hall and ■ Gemyse

		Room: Axelborg Hall
11.45 - 13.00		**Molecular therapeutic approaches (O.7-11)** **Parallel o**ral presentations 2 - Axelborg Hall **Chairpersons: Gisèle Bonne and Carsten Bonneman**
11.45 - 12.00	O.7	**Correcting neuromuscular pathogenic variants with CRISPR-Cas9 technology** H. Best; K. Woodman; A. Lek; K. Koczwara; E. Xu; A. Aykanat; Y. Jiang; M. Saltzman; A. Beggs; M. Lek
12.00 - 12.15	O.8	**DUX4 mRNA silencing with CRISPR-Cas13 gene therapy as a prospective treatment for Facioscapulohumeral muscular dystrophy** A. Rashnonejad; G. Amini Chermahini; L. Wallace; S. Harper
12.15 - 12.30	O.9	**Dominant Collagen XII-related myopathy with a distal myopathy phenotype, amenable to treatment with allele-specific knockdown** P. Mohassel; T. Liewluck; Y. Hu; D. Ezzo; D. Saade; S. Neuhaus; V. Bolduc; Y. Zou; S. Donkervoort; L. Medne; C. Sumner; P. Dyck; K. Wierenga; R. Finkel; J. Chen; T. Winder; N. Staff; M. Koch; A. Foley; C. Bönnemann
12.30 - 12.45	O.10	**A novel target for splice-modulating therapies: a common pseudoexon-inducing mutation that causes a severe collagen VI-related muscular dystrophy** V. Bolduc; A. Foley; H. Solomon Degefa; A. Sarathy; S. Donkervoort; Y. Hu; H. Zhou; B. Cummings; M. Lek; O. Regev; C. Jimenez-Mallebrera; V. Allamand; A. Ferlini; S. Wilton; E. Hanssen; S. Lamandé; D. MacArthur; R. Wagener; F. Muntoni; C. Bönnemann
12.45 - 13.00	O.11	**Dystrophin gene codon usage lacks extreme codon bias and shows non-random codon distribution of disease-causing mutations** M. Fang; R. Rossi; C. Jiang; C. Yu; C. Flesia; W. Li; A. Ferlini
13:15		Lunch ■ Axelborg Hall
13.30 - 15.00		**Industry symposium 1**

Poster Session 1

15.15 - 16.45 Poster Area, locations: Lumbye Hall | Gemyse | Andersen Hall

	Inflammatory myopathies (P.01-21) Lumbye Hall
P.1	**Clinicopathological features of sporadic inclusion body myositis with anti- cytosolic 5'-nucleotidase 1A autoantibodies** S. Yamashita; N. Tawara; T. Ikeda; K. Hara; Z. Zhang; X. Zhang; T. Doki; Y. Ando
P.2	**Phase 2/3 study of Arimoclomol in sporadic inclusion body myositis: Study design** P. Machado; R. Barohn; M. McDermott; T. Blaetter; T. Lloyd; A. Shaibani; M. Freimer; A. Amato; E. Ciafaloni; T. Burns; T. Mozaffar; S. Gibson; M. Wicklund; D. Saperstein; T. Levine; C. Sundgreen; A. Aaes-Jørgensen; T. Liu; L. Herbelin; M. Hanna; M. Dimachkie
P.3	**Delayed diagnosis and treatment lead to worse outcome in immune mediated necrotizing myopathy** C. Phan; J. Kwan; A. Deboo; S. Biliciler
P.4	**Resistance exercises with blood flow restriction in patients with sporadic inclusion body myositis** C. Liang; M. Burk; A. Wall; H. Wong; L. Augustine; S. Bright; W. Brown; A. Harris; S. Holmes; S. Jeyalingam; S. Large; S. Mai; E. Raper; P. Wong; R. Davis; C. Sue; B. Lucas
P.5	**Feasibility and validation of modified oculobulbar facial respiratory score (mOBFRS) in sporadic inclusion body myositis** N. Araujo; M. Wencel; E. Medina; L. Zhang; D. Nguyen; A. Habib; T. Mozaffar; N. Goyal
P.6	**Muscle ultrasound in patients with Inclusion body myositis: Differentiating from mimics** K. Leeuwenberg; L. Christopher-Stine; J. Paik; E. Tiniakou; C. Mecoli; N. van Alfen; J. Doorduin; C. Saris; J. Albayda
P.7	**Severe axial and pelvifemoral muscle damage in immune-mediated necrotizing myopathy evaluated by whole-body MRI** O. Landon-Cardinal; C. Koumako; G. Hardouin; H. Reyngoudt; J. Boisserie; B. Granger; A. Rigolet; B. Hervier; N. Champtiaux; P. Guillaume-Jugnot; M. Vautier; P. Carlier; O. Benveniste; Y. Allenbach
P.8	**Interferon level assessed by ultrasensitive detection technology in myositis patients: a promising biomarker of disease activity in dermatomyositis and anti-synthetase syndrome** L. Bolko; C. Anquetil; S. Maillard; O. Landon-Cardinal; S. Toquet; K. Dorgham; N. Wesner; D. Amelin; G. Dzangue-Tchoupou; A. Llibre-Seradel; D. Duffy; P. Guillaume-Jugnot; A. Rigolet; B. Hervier; M. Vautier; N. Champtiaux; G. Gorochov; O. Benveniste; Y. Allenbach
P.9	**Vacuolar myopathy with monoclonal gammopathy and stiffness (VAMGS)** Y. Allenbach; E. Salort-Campana; E. Malfatti; B. Eymard; A. Rigolet; S. Attarian; A. Maues de Paula; S. Leonard-Louis; O. Benveniste; T. Stojkovic
P.10	**Growth differentiation factor-15 as a novel biomarker for idiopathic inflammatory myopathies** B. De Paepe; F. Verhamme; J. De Bleecker
P.11	**Clinicopathological difference between anti-SRP and anti-HMGCR myopathy** N. Eura; T. Shiota; M. Ozaki; N. Iguchi; Y. Uchihara; H. Nanaura; K. Fukushima; T. Kiriyama; T. Izumi; H. Kataoka; K. Sugie
P.12	**Active immunization mouse model of sporadic inclusion body myositis by cN1A peptides** N. Tawara; Z. Zhang; S. Yamashita; K. Hara; X. Zhang; T. Doki; S. Nakane; Y. Ando
P.13	**Treatment experience of Taiwanese patients with anti-HMGCR myopathy** W. Liang; C. Wang; W. Chen; S. Suzuki; I. Nishino; Y. Jong
P.14	**Immune-mediated neuromuscular complications of graft-versus-host disease** J. Saw; M. Sidiqi; M. Mauermann; H. Alkhateeb; E. Naddaf
P.15	**Myopathies featuring early or prominent dysphagia** J. Triplett; M. Pinto; M. Milone; T. Liewluck
P.16	**Cardiac and respiratory complications in necrotizing autoimmune myopathy** J. Triplett; C. Kassardjian; T. Liewluck; A. Tahir; S. Kopecky; M. Milone
P.17	**Xenograft model of sporadic inclusion body myositis** K. Britson; K. Russell; W. Tsao; J. Montagne; B. Larman; K. Wagner; L. Ostrow; T. Lloyd
P.18	**Comparing histological features and molecular gene expression in anti-Jo1-, anti-PL-7 and anti-PL-12 antibody-positive patients** C. Preusse; B. Paesler; T. Ruck; Y. Allenbach; O. Benveniste; N. Streichenberger; S. Meuth; W. Stenze
P.19	**Novel aspects in Immune-mediated necrotizing myopathy with pipestem capillaries** L. Schweizer; A. Schänzer; H. Goebel; W. Stenzel
P.20	**Expanding the myasthenia-myositis association spectrum: clinical, morphological and immunological data form a large case series** M. Garibaldi; L. Fionda; F. Vanoli; L. Leonardi; E. Bucci; S. Morino; G. Merlonghi; M. Lucchini; M. Mirabella; F. Andreetta; E. Pennisi; A. Petrucci; G. Antonini
P.21	**Genetic analysis of first-degree relatives with inclusion body myositis** S. Nicolau; Z. Niu; K. Ling; M. Milone

	Myotonic dystrophy (P.22-37) Lumbye Hall
P.22	**Subclinical myocardial dysfunction in patients with myotonic dystrophy type 1 and type 2 detected by cardiac magnetic resonance** C. von Landenberg; J. Luetkens; A. Isaak; A. Faron; D. Kuetting; C. Gliem; F. Schmeel; D. Dabir; D. Thomas; C. Kornblum
P.23	**Quality of life outcomes in patients with congenital and childhood-onset myotonic dystrophy** A. Kelder; B. Wong; P. Horn; I. Rybalsky; J. Bange; C. Tian
P.24	**A questionnaire for parents about raising children with congenital/childhood-onset myotonic dystrophy in Japan** M. Shichiji; K. Ishiguro; T. Sato; S. Nagata; K. Ishigaki
P.25	**A longitudinal study of autism spectrum disorders in children, adolescents and young adults with congenital and childhood myotonic dystrophy type 1** A. Ekström; G. Lindeblad; K. Sofou
P.26	**Strength training in myotonic dystrophy type 1: A promising therapeutic strategy** L. Hébert; M. Roussel; C. Gagnon; É. Duchesne
P.27	**Targeted delivery of oligonucleotide therapeutics to muscle reduces toxic DMPK RNA** M. Qatanani; T. Weeden; N. Hsia; C. Desjardins; S. Spring; B. Connolly; M. Yao; C. Stehman-Breen; R. Subramanian
P.28	**Tracking cognitive changes in DM1 in 5 years follow up study** E. Pinzan; V. Pegoraro; R. Marozzo; C. Angelini
P.29	**Patient reported outcome measures in myotonic dystrophy type 2** F. Montagnese; E. Rastelli; K. Stahl; N. Khizanishvili; B. Schoser
P.30	**Reliability of balance, function, and muscle strength measures in Myotonic Dystrophy Type 1** K. Knak; A. Sheikh; H. Andersen; N. Witting; J. Vissing
P.31	**Survey of the actual state of medical care of patients with myotonic dystrophy in Japan** T. Matsumura; M. Takahashi
P.32	**Perceived occupational competence, value of everyday activities, fatigue/daytime sleepiness and quality of life in adults with myotonic dystrophy type 1** U. Edofsson
P.33	**Quantified muscle testing: the right way to assess muscle strength impairments in DM1** L. Hébert; M. Roussel; É. Petitclerc; C. Gagnon; É. Duchesne
P.34	**Nuclear envelope abnormalities in myotonic dystrophy primary myoblasts** P. Meinke; S. Limmer; S. Hintze; B. Schoser
P.35	**Parental repeat length instability in myotonic dystrophy type 1 pre- and protomutations** I. Joosten; D. Hellebrekers; B. de Greef; H. Smeets; C. de Die-Smulders; C. Faber; M. Gerrits
P.36	**Genetic prevalence of myotonic dystrophy type 1: A population cohort study** R. Butterfield; C. Imburgia; D. Dunn; D. Duval; M. Feldkamp; R. Weiss; N. Johnson
P.37	**Onset and course of core symptoms in myotonic dystrophies** K. Stahl; M. Retzer; F. Dahlmanns; B. Schoser; S. Wenninger
	FSHD (P.38-51) Lumbye Hall
P.38	**Experiences with facial weakness in patients with facioscapulohumeral dystrophy: A qualitative study** E. Cup; S. Sezer; A. Lanser; K. Mul; J. Groothuis; B. Engelen; T. Satink; N. Voermans
P.39	**Genetic and epigenetic analysis of FSHD-linked 4q35 region in female Coats Disease patients** R. Fitzsimons; P. Jones; A. Fung; T. Jones
P.40	**Ophthalmological findings in facioscapulohumeral dystrophy** R. Goselink; V. Scheur; C. van Kernebeek; G. Padberg; S. van der Maarel; B. van Engelen; C. Erasmus; T. Theelen
P.41	**Extra-skeletal muscle manifestations of facioscapulohumeral muscular dystrophy** C. Kelly; J. Saw; P. Thapa; J. Mandrekar; E. Naddaf
P.42	**Discovery of novel small molecule treatment options for FSHD**M. Geese; M. Ermann; M. Schneider; S. Monecke; A. Kaever; S. Frankenreiter; M. Bayerlova; K. Schreiter; A. Dickie; P. Loke; T. James; A. Anighoro; R. Hirsch; S. Müller; J. De Maeyer
P.43	**Targeting DUX4 expression, the root cause of FSHD: Identification of a drug target and development candidate** O. Wallace; A. Accorsi; R. Barnes; A. Cacace; D. Cadavid; A. Chang; D. Eyerman; R. Gould; S. Kazmirski; J. Maglio; M. Mellion; P. Rahl; A. Robertson; A. Rojas; L. Ronco; N. Shen; L. Thompson; E. Valentine
P.44	**Safety and tolerability of losmapimod, a selective p38α/β MAPK inhibitor, for treatment of FSHD at its root cause** D. Cadavid; M. Mellion; O. Wallace; L. Ronco; D. Thompson; A. Rojas; M. Hage; R. Gould
P.45	**Patterns of muscle involvement, predictive characteristics, and meaningful change for functional motor tasks in facioscapulohumeral muscular dystrophy** J. Statland; K. Eichinger; M. Currence; M. McIntyre; N. Johnson; R. Tawil
P.46	**Clinical trial readiness to solve barriers to drug development in FSHD (ReSolve): protocol of a large, multi-center prospective study** S. LoRusso; N. Johnson; M. McDermott; K. Eichinger; R. Butterfield; K. Higgs; L. Lewis; K. Mul; B. Van Engelen; S. Sacconi; V. Sansone; E. Carraro; P. Shieh; K. Wagner; L. Wang; J. Statland; R. Tawil;

P.47	**Design of a biomarker of DUX4 activity to evaluate losmapimod treatment effect in FSHD Phase 2 trials** L. Ronco; D. Cadavid; A. Chang; M. Mellion; A. Rojas; N. Shen; R. Tawil; S. Tapscott; L. Wang; O. Wallace
P.48	**The role of Dnmt3b in DUX4 repression in transgenic mice** L. Bouwman; B. den Hamer; P. Verveer; L. Lerink; L. Daxinger; S. van der Maarel; J. de Greef
P.49	**Translating DUX4-targeted RNAi-based gene therapy for FSHD** L. Wallace; G. Amini Chermahini; A. Fowler; M. Guggenbiller; K. Kazimir; C. Cash; S. Knoblaugh; S. Harper
P.50	**An in-situ hybridization-based method for detecting DUX4 RNA expression in vitro** G. Amini Chermahini; A. Rashnonejad; S. Harper
P.51	**Identification of the DUX4-targeted miRNome from a library of 1,881 natural human miRNAs** N. Saad; S. Harper
	Mitochondrial diseases (P.52-63) — **Gemyse**
P.52	**Systematic review of the effect of ketogenic or high fat diet in mitochondrial diseases** A. Wegberg van; H. Zweers-van Essen; J. Smeitink; C. Saris ; M. Janssen
P.53	**Potentially confounding variables of mitochondria biomarker GDF-15** A. Ishii ; S. Nohara; F. Yamamoto; H. Tsuji; S. Yatsuga; Y. Koga; A. Tamaoka
P.54	**Defects in iron-sulphur cluster assembly proteins ISCU and FDX2 cause characteristic mitochondrial myopathy** C. Thomsen ; J. Gurgel-Giannetti; Y. Sunnerhagen; A. Giannetti; F. Kok; M. Vainzof; A. Oldfors
P.55	**Sensory ataxic neuropathy with dysarthria and ophthalmoparesis: Seven case reports** H. Karasoy ; R. Tuncel; O. Ekmekci; A. Yuceyar; F. Bademkiran; H. Onay
P.56	**Role of NcoR1 and PGC-1 for mitochondrial dysfunction in skeletal muscle of ovariectomized mice** Y. Kim; T. Ha; J. Ahn
P.57	**Isolated mitochondrial myopathy is common presentation in A3243G-related mitochondrial diseases** K. Ji ; C. Yan
P.58	**No effect of resveratrol supplementation in patients with mitochondrial myopathy - a randomized, double-blind, placebo-controlled, cross-over study** N. Løkken ; T. Khawajazada; J. Storgaard; D. Raaschou-Pedersen; M. Ørngreen; J. Vissing
P.59	**The mutations in mtDNA encoded NADH dehydrogenase subunits genes as common causes of MELAS** K. Ji ; C. Yan
P.60	**A retrospective study of the combination of pyrimidine nucleos(t)ides in patients with thymidine kinase 2 (TK2) deficiency** J. Quan; C. Domínguez-González; C. Paradas; M. Madruga-Garrido; A. Nascimento Osorio; F. Munell; H. Mandel; T. Falik-Zaccai; M. Ginsberg; G. Tal; C. Garone; E. Barca; T. Moors; M. Hirano
P.61	**Continuous renal replacement therapy in children with mitochondrial diseases** H. Lee ; Y. Lee
P.62	**Muscle multiple mitochondrial DNA deletions: A genetic biomarker to detect nuclear-gene mutations in mtDNA maintenance disorders?** P. Serrano-Lorenzo ; I. Hidalgo; A. González-Quintana; J. Docampo; J. Sánchez-Zapardiel; G. Amate; A. Delmiro; J. Arenas; C. Domínguez-González; A. Blázquez-Encinar; M. Martín
P.63	**Chronic progressive external ophthalmoplegia (CPEO) and CPEO-plus cohort of 54 patients from the Netherlands** R. Schoenaker ; E. Kamsteeg; R. Rodenburg; B. Van Engelen; M. Jansen; C. Saris
	Pompe disease and metabolic disorders (PO.64-75) — **Andersen Hall**
P.64	**Deep clinic and histopathologic phenotyping in a cohort of 17 patients with GYG1-related polyglucosan body myopathy** E. Malfatti ; G. Marrosu; T. Stojkovic; S. DiMauro; C. Dominguez-Gonzalez; A. Hernandez-Lain; P. Van den Bergh; F. Petit; A. Oldfors; J. Vissing; P. Laforêt
P.65	**A prospective 4 years follow up longitudinal study of quantitative muscle MRI in a large cohort of patients with late onset Pompe disease** C. Nuñez-Peralta; J. Alonso-Pérez; S. Segovia; J. Llauger; P. Montesinos; I. Belmonte; I. Pedrosa; E. Montiel; A. Alonso-Jimenez; J. Sánchez-González; I. Illa; J. Díaz-Manera
P.66	**Symptomatology and prevalence of Pompe disease in patients with proximal muscle weakness and high CK levels** J. Teodoro; M. Silva; L. Zimerman; K. Turke; L. Silva ; D. Feder; A. Carvalho
P.67	**Overexpression of a constitutively active glycogen synthase in the GSL30 mouse leads to a polyglucosan myopathy** C. Massey ; D. Wells; R. Piercy
P.68	**Breed specifics of muscle function changes in the Austrian Haflinger and Austrian Noriker horses with polysaccharide storage myopathy (PSSM)** R. Zsoldos ; N. Khayatzadeh; H. Soelkner; T. Licka
P.69	**NEO1 and NEO-EXT Studies: Exploratory efficacy of repeat avalglucosidase alfa dosing for up to 5 years in participants with late-onset Pompe disease (LOPD)** B. Schoser ; R. Barohn; B. Byrne; O. Goker-Alpan; P. Kishnani; S. Ladha; P. Laforêt; K. Mengel; L. Pena; S. Sacconi; V. Straub; J. Trivedi; P. Van Damme; A. van der Ploeg; J. Vissing; P. Young; K.A. Haack; K. Liu; P. Fraser; M. Dimachkie
P.70	**Discontinuation of enzyme replacement therapy in adults with Pompe disease: evaluating motives and clinical course** H. Van Kooten; L. Harlaar; N. Van der Beek; P. Van Doorn; A. Van Der Ploeg; E. Brusse

P.71	**The emerging phenotype in long-term enzyme-treated classic infantile Pompe patients: severe distal muscle weakness** J. van den Dorpel ; E. Poelman; L. Harlaar; H. Kooten; L. van der Giessen; P. van Doorn; A. van der Ploeg; J. van den Hout; N. van der Beek
P.72	**Optimizing long-term outcome in classic infantile Pompe patients: effects of higher dosing and immunomodulation** E. Poelman; J. van den Dorpel ; M. Hoogeveen-Westerveld; J. van den Hout; L. van der Giessen; N. van der Beek; W. Pijnappel; A. van der Ploeg
P.73	**Correlation of gluteus medius muscle activities at walk and trot with myopathy changes on biopsies of the exact same locations in horses without clinical signs of type 1 polysaccharide storage myopathy** T. Licka ; R. Van den Hoven; U. Schröder; C. Hahn; R. Zsoldos
P.74	**Uptake of recombinant human GAA in a primary muscle cell differentiation system** S. Limmer ; S. Hintze; P. Meinke; B. Schoser
P.75	**Targeted population screening of late onset Pompe disease in unspecified myopathy patients with 8 neuromuscular centers in Korea** J. Lee ; J. Shin; D. Kim; K. Kim; B. Kim; J. Sung; T. Nam; Y. Park; J. Park; S. Kim; Y. Choi
	Myofibrillar and autophagic myopathies (P.76-84) — **Andersen Hall**
P.76	**Clinical features of X-linked myopathy with excessive autophagy (XMEA) in Japan: A nationwide survey** K. Sugie ; H. Komaki; T. Kurashige; A. Ohkuma; N. Eura; T. Shiota; N. Iguchi; H. Nanaura; T. Abe; I. Nonaka; I. Nishino
P.77	**Repurposing of metformin identified as a potential therapy in models of BAG3 myofibrillar myopathy** A. Ruparelia; E. McKaige; C. Williams; K. Schulze; M. Fuchs; V. Oorschot; E. Lacene; M. Mirella; E. Baxter; Y. Torrente; G. Ramm; T. Stojkovic; J. Lavoié; R. Bryson-Richardson
P.78	**Sarcomeric pathology induced by homozygous expression of the myofibrillar myopathy - associated p.W2711X filamin C mutant** J. Schuld; Z. Orfanos; F. Chevessier; A. Unger; G. Kirfel; P. van der Ven; W. Linke; C. Clemen ; D. Fürst; R. Schröder
P.79	**Malignant cardiac phenotype after pressure overload in autosomal-dominant desminopathies: lessons from heterozygous p.R349P desmin knock-in mice** F. Stöckigt; L. Eichhorn; T. Beiert; V. Knappe; T. Radecke; M. Steinmetz; C. Berwanger; G. Nickenig; V. Peeva; W. Kunz; C. Clemen ; R. Schröder; J. Schrickel
P.80	**Imbalances in protein homeostasis caused by mutant desmin** C. Clemen ; L. Winter; A. Unger; C. Berwanger; M. Spörrer; W. Linke; R. Schröder
P.81	**Mutation in Z-disk associated protein filamin C (p.Ala2430Val) causes myofibrillar hypertrophic cardiomyopathy** A. Schänzer ; E. Schuhmann; D. Zengeler; J. Schuld; L. Gulatz; G. Maroli; S. Gräf; U. Ahting; A. Hahn; A. Nemeth; D. Fürst; S. Rupp; P. van der Ven
P.82	**First clinical and neuropathological description of a myofibrillar myopathy with congenital onset based on a homozygous recessive FLNC mutation** H. Kölbel ; A. Roos; K. Nolte; K. Johnson; A. Töpf; L. Kollipara; W. Kress; P. van der Ven; V. Straub; J. Weis; D. Fürst; U. Schara
P.83	**Vacuolar myopathy with valosin containing protein (VCP)-positive intranuclear and cytoplasmic inclusions: report of two cases with early and late childhood-onset disease** R. Phadke ; A. Dean; M. Evans; A. Parker; D. Maxwell; C. Sewry; A. Sarkozy; F. Muntoni
P.84	**Impaired cargo-selective autophagy due to altered signaling causes the Z-disc myofibrillar disintegration in myofibrillar myopathy due to LDB3 p.A165V mutation in a knock-in mouse model** P. Pathak ; Y. Blech-Hermoni; K. Subedi; J. Mpamugo; C. Obeng-Nyarko; A. Mankodi
	Lipid myopathies (P.85-89) — **Andersen Hall**
P.85	**TANGO2-related disorder: Spectrum of mutations and clinical phenotype characterized by recurrent rhabdomyolysis and metabolic crisis** D. Natera-de Benito; L. Carrera; C. Ortez; N. Juliá; M. O C´ allaghan; V. Delgadillo; J. Eiris; A. Garcia; C. Jou; C. Jiemenez- Mallebrera; A. Codina; J. Pijuan; J. Colomer; L. Martorell; J. Exposito; D. Yubero; J. Hoenicka; C. Arjona; F. Palau; A. Nascimento
P.86	**Depends on how you look: hereditary spastic paraplegia, leukodystrophy or brain iron accumulation** N. Eroglu-Ertugrul; H. Topaloˇglu
P.87	**Carnitine deficiency in patients with neuromuscular diseases on long-term tube feeding** K. Ueno ; H. Takada; H. Matsuo; S. Kuru; K. Goto; T. Mitsui; M. Ishizaki; S. Sugimoto; K. Ogata; T. Matsumura; S. Suwazono; H. Furuya; A. Watanabe; Y. Kawano; A. Yamamoto; N. Sasagasako; H. Arahata
P.88	**Lipid storage myopathies due to ETFDH and PNPLA2 mutations: MRI and microRNA role as biomarkers** C. Angelini ; V. Pegoraro; R. Marozzo; S. Missaglia; D. Tavian; D. Tavian
P.89	**Infantile-onset lipid storage myopathy** L. Indrawati ; S. Noguchi; J. Tanboon; M. Ogasawara; Y. Saito; T. Kumutpongpanich; M. Inoue; M. Okubo; T. Fukuda; H. Sugie; Y. Goto; A. Iida; S. Hayashi; I. Nishino
	CMT and neuropathy (P.90-96) — **Andersen Hall**
P.90	**Reduced power and manual dexterity have an effect on fine motor function in children with CMT1A** J. Longatto ; M. Main; F. Muntoni
P.91	**Optimization of AAV-mediated gene therapy for SOD1-linked ALS** M. Biferi ; M. Cohen-Tannoudji; T. Marais; B. Giroux; S. Astord

P.92	**Feasibility and validation of modified oculobulbar facial respiratory score (mOBFRS) in amyotrophic lateral sclerosis** N. Araujo ; M. Wencel; E. Medina; L. Zhang; D. Nguyen; A. Habib; T. Mozaffar; N. Goyal
P.93	**Small fiber neuropathy underlying auto inflammatory syndromes in children** I. Shinkarevsy; I. Nevo; L. Harel; G. Amarilyo; A. Dori; n. Agmon-Levin; L. Kachko; R. Dabby; M. Rabie; S. Aharoni
P.94	**Peripheral myelin protein 2 – a novel mutation causing Charcot-Marie-Tooth neuropathy in a Bulgarian family** T. Chamova ; P. Palaima; V. Mitev; C. Van Broeckhoven; K. Peeters; A. Jordanova; I. Tournev
P.95	**Walking speed in Charcot-Marie-Tooth disease: a marker of disease progression during childhood and adolescence** G. Acsadi ; T. Wren; K. Pierz; S. Ounpuu
P.96	**Clinical and genetic aspects in Korean patients with hereditary spastic paraplegia** H. Kim ; Y. Choi; H. Park
	Myotonia (P.97-101) — **Andersen Hall**
P.97	**KCNJ2 channelopathy causes life-threatening arrhythmia and periodic paralysis in children with Andersen-Tawil syndrome** D. Vlodavets ; M. Shkolnikova; V. Bereznitskaya; Z. Romantsova; R. Ildarova; A. Monakhova; O. Shidlovskaya; I. Shulyakova; S. Artemieva; E. Litvinova; E. Belousova
P.98	**RCT of 2mg bumetanide for hypokalaemic periodic paralysis** R. Scalco ; J. Morrow; A. Manole; I. Skorupinska; F. Ricciardi; E. Matthews; M. Hanna; D. Fialho
P.99	**A multiplex in-situ hybridisation and immunohistochemical (ISH-IHC) assay to study developmental changes in relation to fibre type and sodium channels in human skeletal muscle and their contribution to disease severity** E. Matthews ; D. Chambers; L. Feng; F. Launchbury; A. Richard-Londt; F. Muntoni; M. Hanna; R. Phadke
P.100	**Pseudo-increment at repetitive nerve stimulation (Arzel's sign): a new tool for Brody myopathy diagnosis** M. Masingue ; M. Arzel; D. Sternberg; T. Stojkovic; A. Behin; G. Bassez; S. Vicart; Y. Péréon; T. Kuntzer; B. Eymard; E. Fournier
P.101	**Defining the phenotype of Andersen-Tawil syndrome: The largest reported series** V. Vivekanandam ; L. Germain; I. Skorupinska; K. Seutterlin; D. Fialho; D. Kozyra; R. Sud; S. Holmes; N. James; M. Hanna; E. Matthews
	E-Posters - Mitochondrial / Metabolic diseases – case reports (EP.01-13) — **Lumbye Hall**
EP.01	**Becker muscular dystrophy with acid α-glucosidase pseudodeficiency** A. Ishiyama ; Y. Oitani; M. Kosuga; E. Takeshita; Y. Shimizu-Motohashi; H. Komaki; I. Nishino; T. Okuyama; M. Sasaki
EP.02	**Deep sequencing of mitochondrial DNA and functional characterization of a novel POLG mutation in a patient with autosomal recessive progressive external ophthalmoplegia** C. Hedberg-Oldfors ; B. Macao; S. Basu; J. Uhler; D. Erdinc; B. Peter; C. Lindberg; E. Larsson; M. Falkenberg; A. Oldfors
EP.03	**Mevalonate kinase deficiency – a mimic of mitochondrial myopathy in clinic and muscle biopsy** C. von Landenberg; C. Kornblum; J. Reimann
EP.04	**A case of late onset multiple acyl-CoA dehydrogenase deficiency with novel ETFDH mutation** J. Shin ; Y. Hong; J. Sung
EP.05	**New POLG mutation causing autosomal dominant PEO, proximal and distal myopathy and respiratory failure** L. Gonzalez Mera ; L. Carreño-Gago; A. Pellisé; I. Paramonov; E. García-Arumí; M. Olive
EP.06	**Adult-onset recurrent rhabdomyolysis due to variants in LPIN1** T. Minton; N. Forrester; S. Al Baba; K. Urankar; S. Brady
EP.07	**Impaired lipolysis in propionic acidemia – a case story** J. Storgaard ; K. Madsen; N. Loekken; J. Vissing; G. van Hall; A. Lund; M. Oerngreen
EP.08	**Highly asymmetrical distribution of muscle wasting correlated with heteroplasmy level in a patient suffering from a mitochondrial myopathy with single deletion of mitochondrial DNA** M. Masingue ; B. Eymard; N. Romero; B. Rucheton
EP.09	**Hypersomnia-featured very long-chain acyl-coenzyme a dehydrogenase deficiency caused by a homozygous R428H mutation of ACADVL** Y. DA ; Y. lu; M. Pang; X. Shen
EP.10	**A progressive infantile myopathy case with TK2-related mitochondrial DNA depletion syndrome: correlation with muscle pathology** J. Shin ; S. Kim; Y. Park; A. Ko; J. Kong; S. Nam
EP.11	**Impaired fat oxidation during exercise in long-chain acyl-CoA dehydrogenase deficiency patients and effect of IV-glucose** K. Madsen ; M. Stemmerik; A. Buch; N. Nielsen; A. Lund; J. Vissing
EP.12	**Mitochondrial myopathy and progressive external ophthalmoplegia associated with novel mutations m.5669G > A and m.5701delA in MT-TN** K. Visuttijai ; C. Hedberg-Oldfors; U. Lindgren; Ó. Elíasdóttir; S. Nordström; A. Oldfors
EP.13	**Long-lasting corticosteroids-induced improvement in inclusion body myositis with prominent mitochondrial changes** C. Damien ; N. Mavroudakis; H. Kadhim; G. Remiche

	E-Posters – Inflammatory myopathies and muscular dystrophies – case reports (EP.14-28) Andersen Hall
EP.14	**Acute muscle weakness in paediatric age masking an underlying limb girdle muscular dystrophy type 2I** A. Silwal ; F. Muntoni; R. Mein; A. Manzur
EP.15	**Valosin-containing protein-related myopathy and Meige's syndrome: just a coincidence or not?** C. Papadopoulos ; E. Malfatti; E. Anagnostou; M. Savarese; B. Udd; G. Papadimas
EP.16	**Factors associated with postoperative surgical complications in myotonic dystrophy type 1** J. Park ; D. Kim
EP.17	**Vaccination as possible trigger of anti-srp mediated necrotizing myopathy - a case report** J. Cavalcanti; W. Lima Junior; K. Turke; L. Silva ; A. Carvalho
EP.18	**Mutations in TRAPPC11 are associated with Rett-like syndrome in absence of significant muscle involvement** S. Vila-Bedmar ; P. Quijada-Fraile; F. Martinez-Azorin; A. Hernández-Lain; R. Cancho-Candela; N. Núñez-Enamorado; A. Camacho-Salas
EP.19	**Anti-HMGCR positive necrotizing myopathy presenting in childhood** M. Rasmussen ; A. Selvaag; S. Wallace; E. Kirkhus; E. Merckoll; E. Antal; J. Pahnke; B. Udd
EP.20	**ANO5 -related myopathy: report of the first Greek patients** M. Svingou; K. Kekou; C. Papadopoulos; S. Xirou; E. Kararizou; G. Papadimas
EP.21	**Bcl-2-associated athanogene-3 (BAG3) myopathy in an ethnic-Indian Malaysian patient** K. Goh ; S. Low; K. Mun; C. Tan; N. Shahrizaila; I. Nishino; K. Wong
EP.22	**Anti-HMGCR antibody in asymptomatic patients with high CK – Case report** I. Soares; V. Comprido; B. Hsu; K. Turke; L. Silva ; D. Feder; A. Carvalho
EP.23	**A novel LMNA mutation identified in an Argentinian patient with autosomal dominant Emery Dreifuss muscular dystrophy phenotype.** C. Azcona; M. Bettini; C. Belziti; M. Brogger; M. Rugiero
EP.24	**Recessive mutations in CAV3 – a new differential diagnosis of early-onset neuromuscular disorders.** M. Fernández-García ; T. Kerr; P. Johns; M. McEntagart; R. Phadke; V. Gowda; H. Jungbluth; E. Wraige
EP.25	**A limb girdle muscular dystrophy phenotype with mutations in ISPD and TTN** A. Aksoy; Ö. Yayıcı Köken ; B. Çavdarlı; B. Talim; D. Yüksel
EP.26	**Phenotype may predict the clinical severity of facioscapulohumeral muscular dystrophy** Y. Liu; D. Yue; W. Zhu ; J. Li; S. Cai; S. Luo; J. Xi; J. Lin; J. Lu; L. Zhou; Z. Liang; J. Lu; C. Zhao
EP.27	**Sporadic late-onset nemaline myopathy: an unusual case misdiagnosed as immune-mediated necrotizing myopathy** D. Pehl ; J. Prudlo; H. Goebel; W. Stenzel
EP.28	**Immune checkpoint inhibitors (ICIs)-related ocular myositis** M. Garibaldi ; F. Calabrò; G. Merlonghi; S. Pugliese; L. Cristiano; T. Tartaglione; A. Petrucci

16.15-16.45 Afternoon refreshments, exhibition and posters - ■ Andersen Hall ■ Lumbye Hall and ■ Gemyse

Poster Session 2

16.45 - 18.15 Poster Area, locations: Lumbye Hall | Gemyse | Andersen Hall

	Congenital myopathies: centronuclear and others (P.103-114) Lumbye Hall
P.103	**STIM1 over-activation generates a multi-systemic phenotype affecting skeletal muscle, spleen, eye, skin, bones, and the immune system in mice** R. Silva-Rojas; S. Treves; J. Laporte; J. Böhm
P.104	**Skeletal muscle reduction of Dnm2 with antisense oligonucleotides in myotubular myopathy** S. Buono; A. Robé; R. Gomez Oca; C. Koch; S. Guo; M. Depla; B. Monia; J. Laporte; L. Thielemans; B. Cowling
P.105	**INCEPTUS pre-phase 1, prospective, non-interventional, natural history run-in study to evaluate subjects aged 4 years and younger with X-linked myotubular myopathy (XLMTM)** L. Servais ; P. Shieh; J. Dowling; N. Kuntz; W. Müller-Felber; B. Smith; C. Bönnemann; F. Muntoni; D. Bilder; T. Duong; R. Graham; M. Jain; M. Lawlor; V. MacBean; M. Noursalehi; T. Pitts; G. Rafferty; S. Rico; S. Prasad
P.106	**Mutation-specific therapy for X-linked myotubular myopathy** S. Hayashi ; S. Noguchi; T. Kumutpongpanich; A. Iida; M. Okubo; M. Matsuo; I. Nishino
P.107	**Clinical changes over time in patients with centronuclear myopathy due to mutations in DNM2 gene enrolled in a European prospective natural history study** M. Annoussamy; J. Baets; W. De Ridder; D. Duchêne ; A. Grangé; C. Lilien; V. Chê; T. Gidaro; A. Seferian; A. Behin; N. Voermans; M. Bitoun; J. Hogrel; C. Freitag; K. Paradis; L. Thielemans; S. Van Rooijen; L. Servais
P.108	**Recessive MYH7 -related myopathy in two families** S. Beecroft ; M. van de Locht; J. de Winter; C. Ottenheijm; C. Sewry; S. Mohammed; M. Ryan; I. Woodcock; L. Sanders; R. Gooding; M. Davis; E. Oates; N. Laing; G. Ravenscroft; C. McLean; H. Jungbluth
P.109	**Congenital myopathy in patients with Kabuki and Au-Kline syndromes - double trouble or expansion of the phenotypes?** A. Sarkozy ; M. Fernandez-Garcia; A. Manzur; R. Mein; I. Bodi; R. Phadke; E. Wraige; C. Deshpande; S. Holder; J. Hurst; M. Gautel; H. Jungbluth; F. Muntoni
P.110	**Clinical, morphological and genetic data in Italian patients with fiber-type-disproportion** L. Maggi ; M. Verardo; A. Rubegni; C. Bruno; A. Lupica; A. Berardinelli; M. Ripolone; G. Vattemi; L. Ruggiero; A. D'Amico; C. Rodolico; M. Sciacco; V. Nigro; F. Santorelli; P. Tonin; C. Fiorillo; M. Mora
P.111	**Modelling autosomal dominant centronuclear myopathy in zebrafish** S. Coppens ; E. Dupuis; P. Gillotay; S. Costagliola
P.112	**Two murine models for tubular aggregate myopathy with mutations in Stim1 and Orai1** M. Ogasawara ; M. Ogawa; Y. Endo; J. Lee; Y. Inoue; T. Inoue; I. Nishino; S. Noguchi
P.113	**Phenotype, genetics and natural history in 131 SEPN1 -related myopathy patients: towards clinical trial readiness** R. Villar-Quiles ; M. von der Hagen; S. Quijano-Roy; V. Gonzalez; S. Donkervoort; M. de Visser; A. Fidzianska; D. Orlikowski; N. Goemans; M. Mayer; L. Merlini; N. Romero; M. Fardeau; H. Topaloglu; C. Métay; P. Richard; B. Estournet; C. Bönnemann; U. Schara; A. Ferreiro
P.114	**Clinical, histological, and genetic characterization of PYROXD1 -related myopathy** X. Lornage ; V. Schartner; I. Balbueno; T. Willis; A. Echaniz-Laguna; E. Malfatti; S. Scheidecker; V. Biancalana; C. Sewry; N. Romero; J. Laporte; J. Böhm
	McArdle disease (P.115-124) Lumbye Hall
P.115	**Natural history of McArdle disease in a cohort of 220 patients** R. Quinlivan ; A. Pietrusz; C. Pizzamiglio; j. Pattni; O. Mahroo; K. Khan; P. Elliott; M. Patasin; S. Chatfield; A. Lucia; j. Zugaza; F. Llavero; R. Godfrey
P.116	**The existence of the 'Third Wind' phenomenon in McArdle disease** R. Godfrey ; J. Burman; G. Lees; R. Scalco; S. Chatfield; J. Pattni; A. Wakelin; R. Quinlivan
P.117	**Rhabdomyolysis due to unaccustomed exercise: experiences from a multidisciplinary clinic** S. Chatfield ; J. Pattni; R. Godfrey; R. Quinlivan
P.118	**Descriptions of pain in McArdle disease** S. Chatfield ; J. Pattni; S. Booth; R. Godfrey; R. Quinlivan
P.119	**Analysis of the structural and metabolic consequences of McArdle disease using the murine model** A. Real-Martínez; A. Brull; J. Huerta; M. Villarreal-Salazar; G. Tarrasó; A. Lucia; M. Martin; J. Arenas; A. Andreu; G. Nogales-Gadea; J. Vissing; T. Krag; N. de Luna; T. Pinós
P.120	**EUROMAC: a European registry for patients with McArdle disease and other very rare muscle glycogenoses.** T. Pinós ; R. Scalco; A. Lucia; M. Martin; A. Martinuzzi; A. Toscano; B. San Millan; N. Voermans; P. Laforet; M. Vorgerd; S. Sacconi; H. Durmus; B. Kierdaszuk; G. Siciliano; C. Bruno; A. Wakelin; R. Quinlivan; J. Vissing; R. Martí; E. EUROMAC Consortium
P.121	**Development of a rowing ergometer protocol to test whole body VO2peak in McArdle disease** R. Godfrey ; J. Newbury; S. Chatfield; J. Pattni; A. Wakelin; R. Quinlivan
P.122	**Feasibility open label trial shows no effect of sodium valproate for McArdle disease** R. Scalco; M. Stemmerik; N. Løkken; C. Vissing; K. Madsen; Z. Michalak; R. Godfrey; J. Pattni; J. Holton; G. Samandouras; P. Bassett; R. Haller; J. Vissing; R. Quinlivan
P.123	**Frequency of coronary artery disease in people with McArdle disease** A. Pietrusz ; R. Scalco; C. Pizzamiglio; K. Savvatis; T. Gkosios; P. Elliott; R. Godfrey; J. Pattni; S. Chatfield; M. Patasin; R. Quinlivan

P.124	**Is the expression of muscle glycogen phosphorylase tissue-specific? New perspectives on McArdle disease** F. Francisco Llavero Bernal ; A. Alazne Arrazola Sastre; M. Miriam Luque Montoro; R. Renata Scalco; C. Chiara Pizzamiglio; J. Jatin Pattni; S. Sherryl Chatfield; R. Ros Quinlivan; M. Miguel A. Martin; J. Joaquín Arenas; A. Alejandro Lucia; J. José L Zugaza
	Muscle homeostasis (P.125-138) **Lumbye Hall**
P.125	**Protein homeostasis in LGMD2I** V. Franekova ; G. Leivseth; O. Nilssen
P.126	**SRK-015, a Fully human monoclonal antibody inhibiting myostatin activation, offers sustained target engagement across multiple species, including humans** A. Kalra ; S. Cote; D. Barrett; K. Long; M. Kavosi; S. Wawersik; H. Faulds; E. Treece; R. Iarrobino; Y. Chyung
P.127	**KER-050, a novel muscle anabolic, functions as a ligand trap that binds myo-catabolic TGF β ligands and has reduced binding affinity for BMP9, a critical vascular remodeling ligand** E. Lema ; N. Kondratiev; R. Nathan; T. Backus; f. Fisher; J. Seehra; J. Lachey
P.128	**2, 6-Dimethoxy-1, 4-benzoquinone increases skeletal muscle mass through Akt/mTOR signaling pathway** H. Son; Y. Jang; J. Ahn; C. Jung; T. Ha
P.129	**Latent myostatin specific elimination by sweeping antibody® is a novel therapeutic approach to improve muscle strength** H. Muramatsu
P.130	**Myoscreen TM , a drug discovery platform for disease modeling and the screening of compounds restoring muscle health** M. Flaender ; J. Young; E. Duchemin-Pelletier; O. Lorintiu; P. Poydenot
P.131	**Cytoglobin modulates skeletal muscle regeneration by targeting canonical Wnt signaling** S. Singh; J. Haung; D. Rivas; T. Gemelli; M. Weiler; T. Tassin; E. Chung; S. Goetsch; J. Shelton; J. Richardson; J. Schneider; P. Mammen
P.132	**Dissection of UBA5 dysfunction in nervous system disorders** R. Serrano; V. Oorschot; G. Ramm; R. Bryson-Richardson
P.133	**A novel CaV β1 isoform connecting voltage sensing with muscle mass homeostasis** M. Traore; C. Gentil; C. Benedetto; J. Hogrel; P. De la Grange; S. Benkhelifa-Ziyyat; L. Julien; M. lemaitre; A. Ferry; S. Falcone
P.134	**Physical and transcriptional characterization of human urinary stem cell populations** M. Falzarano; N. Spedicato; A. Margutti; R. El Dani; R. Rossi; F. Gualandi; R. Selvatici; M. Fang; Z. Lu; S. Zia; P. Reschiglian; B. Roda; A. Grilli; S. Bicciato; A. Ferlini
P.135	**Chemochine CXCL12 as an intrinsic modulator of skeletal muscle mass in healthy human** Y. Maeda
P.136	**DNAJB6 integrates sarcomere protein homeostasis and myogenic signaling in coordinated skeletal muscle stress response** A. Findlay ; R. Bengoechea; S. Pittman; C. Weihl
P.137	**ETS1 facilitates muscle satellite cell proliferation and stemness perpetuation** T. Kumutpongpanich ; H. Shinichiro; S. Noguchi; N. Ichizo
P.138	**C-terminal titin fragments in mouse muscles** J. Sarparanta ; A. Vihola; R. Singh; I. Richard; B. Udd
	DMD clinical and biomarkers (P.139-152) **Lumbye Hall**
P.139	**Paediatric use of tamoxifen: good safety profile for boys with Duchenne muscular dystrophy and X-linked myotubular myopathy** O. Dorchies ; L. Neff; E. Gayi; C. Baulet; S. Fabien; R. de Mestral; P. Hafner; D. Fischer; T. Dor; L. Scapozza
P.140	**RNA-Seq shows an absence of off-target splicing effects in AAV9-U7snRNA Mediated Skipping of DMD Exon 2** K. Flanigan; N. Wein; L. Gushchina ; M. Waldrop; R. Weiss
P.141	**PPMO-mediated skipping therapy of duplicated exon 2 in the DMD gene** L. Gushchina ; K. Grounds; H. Huang; E. Frair; F. Schnell; G. Hanson; T. Simmons; N. Wein; K. Flanigan
P.142	**Very low residual dystrophin levels mitigate dystrophinopathy towards Becker muscular dystrophy** Y. de Feraudy ; R. Ben Yaou; K. Wahbi; F. Leturcq; H. Amthor
P.143	**Correlation between dystrophin espression and clinical phenotype using high-throughput digital immunoanalysis in Duchenne and Becker muscular dystrophy patients** S. Torelli ; D. Scaglioni; V. Sardone; J. Domingos; A. Jones; M. Ellis; D. Eastwood; L. Feng; F. Leturcq; R. Ben Yaour; E. Ricci; M. Beuvin; G. Bonne; C. Sewry; G. Tasca; R. Phadke; J. Morgan; F. Muntoni
P.144	**A cross sectional and longitudinal miRNA profiling study identified a set of novel free-circulating and exosomal miRNAs dysregulated in plasma from Duchenne muscular dystrophy (DMD) patients** F. Catapano; D. Scaglioni ; K. Maresh; V. Selby; P. Ala; J. Domingos; V. Ricotti; L. Phillips; M. van der Holst; L. Servais; A. Seferian; i. de Groot; y. Krom; T. Voit; j. Verschuuren; E. Niks; V. Straub; J. Morgan; F. Muntoni
P.145	**Optimisation of a high–throughput digital script for multiplexed immunofluorescent analysis of sarcolemmal dystrophin and associated protein complex (DPC) and myofibre regeneration in entire transverse sections of muscle biopsies in Duchenne muscular dystrophy (DMD)** D. Scaglioni ; M. Ellis; F. Catapano; S. Torelli; D. Chambers; L. Feng; C. Sewry; J. Morgan; F. Muntoni; R. Phadke
P.146	**Novel high-throughput digital analysis to quantify the amount of functional sarcolemmal dystrophin and myofibre regeneration in Duchenne muscular dystrophy (DMD) clinical trial samples (exon 53 skipping with golodirsen)** D. Scaglioni ; F. Catapano; M. Ellis; S. Torelli; D. Chambers; L. Feng; D. Frank; A. Nair; S. Harriman; A. Dugar; J. Morgan; R. Phadke; F. Muntoni

P.147	**Muscle biopsies in clinical trials for Duchenne muscular dystrophy – patients' and caregivers' perspective** I. Verhaart; A. Johnson; S. Thakrar; E. Vroom; F. De Angelis; F. Muntoni; A. Aartsma-Rus; E. Niks
P.148	**Long-term natural history data in Duchenne muscular dystrophy ambulant patients with mutations amenable to skip exons 44, 45, 51 and 53** C. Brogna; G. Coratti; M. Pane; V. Ricotti; S. Messina; C. Bruno; A. Berardinelli; F. Ricci; R. Battini; L. Bello; E. Pegoraro; G. Baranello; S. Previtali; L. Politano; G. Comi; A. D'Amico; E. Bertini; F. Muntoni; N. Goemans; E. Mercuri
P.149	**The MYODA operational seamless clinical trial design phase I to III: a new approach for rare diseases to evaluate the safety, efficacy, pharmacokinetics and pharmacodynamics of BIO101 (MAS Activator) in paediatric patients with a genetically confirmed diagnosis of Duchenne muscular dystrophy (DMD)** M. Chabane ; W. Dioh; P. Dilda; R. Lafont; s. Veillet; t. Voit; S. Agus
P.150	**Consistency of 48-week changes in North Star Ambulatory Assessment (NSAA) between Duchenne muscular dystrophy (DMD) natural history data and clinical trial placebo arms, after adjustment for prognostic factors** N. Goemans ; B. Wong; F. Muntoni; C. McDonald; E. Mercuri; A. Mazur; J. Signorovitch; G. Sajeev; H. Wong; I. Hussain; M. Jenkins; S. Ward
P.151	**Motor performances in exon-2 duplication of the dystrophin gene** A. Zambon; E. Ferlini; E. Albamonte; G. Baranello; E. Bertini; A. D'Amico; G. D'Angelo; C. Fiorillo; S. Messina; S. Messina; M. Moggio; T. Mongini; M. Natali Sora; M. Pane; A. Pini; L. Politano; F. Ricci; G. Vita; E. Mercuri; G. Comi; S. Previtali
P.152	**A longitudinal study of miRNA biomarkers in paediatric Duchenne muscular dystrophy patients** S. Trifunov; C. Badosa; D. Natera de Benito; J. Exposito Escudero; C. Ortez; J. Medina; L. Carrera; A. Nascimento; C. Jimenez-Mallebrera
	Congenital myopathies: nemaline myopathies (P.154-166) — Gemyse
P.154	**Mouse models of nemaline myopathy display structural and functional abnormalities of mitochondria** J. Tinklenberg ; R. Slick; M. Vanden Avond; M. Beatka; M. Prom; E. Siebers; H. Meng; M. Grzybowski; J. Heisner; J. Ross; J. Ochala; K. Nowak; L. Zhang; A. Geurts; D. Stowe; F. Montanaro; M. Lawlor
P.155	**Proteomic profiling in nemaline myopathy to identify molecular phenotypes** R. Slick ; J. Tinklenberg; M. Vanden Avond; M. Beatka; H. Meng; E. Seibers; A. Geurts; H. Granzier; E. Hardeman; L. Zhang; F. Montanaro; K. Nowak; M. Lawlor
P.156	**Unusual respiratory pattern in children with NEB -related core-rod myopathy** V. Aguerre; V. Leske; M. de Castro; J. Mozzoni; M. Guerdile; P. Gravina; X. Lornage; J. Böhm; N. Romero; J. Laporte; S. Monges
P.157	**L-tyrosine in nemaline Myopathy – what do we know?** S. Weiss ; T. Birsak; A. Ille; A. Kellersmann; M. Gosk-Tomek; R. Bittner; W. Schmidt; G. Bernert
P.158	**Congenital-onset hypertrophic cardiomyopathy and skeletal myopathy with nemaline rods and actin filament aggregates due to likely pathogenic recessive variants in CFL2** R. Phadke ; A. Childs; A. Manzur; P. Munot; R. Mein; A. Sarkozy; C. Sewry; F. Muntoni
P.159	**Disease spectrum in nemaline myopathy due to LMOD3 mild founder mutation in Austria and southern Germany** S. Weiss ; U. Schatz; J. Zschoke; S. Wenninger; B. Schoser; S. Rudnik-Schoeneborn; W. Muss; A. Schossig; W. Schmidt; R. Bittner; G. Bernert; M. Baumann
P.160	**The spectrum of disease-causing and normal variation in the nebulin gene** V. Lehtokari ; L. Sagath; K. Kiiski; C. Wallgren-Pettersson; K. Pelin
P.161	**ADSSL1 myopathy is a fatigability disease presenting both nemaline bodies and lipid droplets in skeletal muscles – a study of 57 Japanese cases** Y. Saito ; S. Noguchi; A. Nishikawa; S. Hayashi; A. Iida; I. Nishino
P.162	**Novel Kbtbd13 R408C -knockin mouse model phenocopies NEM6 myopathy** J. de Winter ; M. Yuen; R. Van der Pijl; F. Li; S. Shengyi; S. Conijn; M. Van de Locht; S. Bogaards; E. van Kleef; R. Bryson-Richardson; K. Campbell; W. Ma; T. Irving; E. Malfatti; H. Granzier; B. van Engelen; N. Voermans; C. Ottenheijm
P.163	**Mutations in fast skeletal troponin C (TNNC2) cause contractile dysfunction** M. van de Locht ; J. de Winter; S. Conijn; W. Ma; M. Helmes; T. Irving; S. Donkervoort; P. Mohassel; L. Medne; C. Quinn; O. Neto; S. Moore; A. Foley; J. Pinto; N. Voermans; C. Bönnemann; C. Ottenheijm
P.164	**Expression of alternative nebulin isoforms containing super repeat S21a or S21b in skeletal muscle** J. Laitila ; C. Sewry; L. Lam; I. Holt; G. Morris; C. Wallgren-Pettersson; C. Pelin
P.165	**Update on functional studies of YBX3 variants associated with nemaline myopathy** L. Sagath ; J. Laitila; V. Lehtokari; K. Kiiski; M. Grönholm; C. Wallgren-Pettersson; K. Pelin
P.166	**Retrospective longitudinal study of patients with NEB-related nemaline myopathy in the United Kingdom** C. Brusa ; D. Steel; R. Mein; A. Manzur; S. Robb; P. Munot; E. Wraige; R. Quinlivan; M. Main; C. Sewry; L. Feng; R. Phadke; H. Jungbluth; A. Sarkozy; F. Muntoni
	Limb girdle muscular dystrophies (P.167-188) — Andersen Hall
P.167	**TRAPPC11-opathies: An emerging phenotype of muscle-eye-brain-liver** E. Bobadilla ; N. Julia; C. Jimenez - Mallebrera; C. Ortez; L. Carrera; D. Natera De Benito; J. Exposito; J. Colomer; C. Jou; A. Codina; D. Yubero; A. Nascimento
P.168	**Clinical spectrum and histopathological characterization of alpha-dystroglycanopathies** E. Bobadilla ; A. Codina; C. Jou; D. Natera; L. Carrera; C. Ortez; D. Yubero; L. Martorell; L. Gonzalez; P. Gallano; J. Exposito; J. Corbera; D. Itzep; J. Colomer; C. Jimenenez-Mallebrera; A. Nascimento

P.169	**Sarcoglycanophathies: experience of a terciary centre** C. Garrido ; A. Sousa; M. Cardoso; R. Taipa; E. Vieira; A. Gonçalves; M. Melo Pires; R. Santos; T. Coelho; M. Santos
P.170	**Follow-up of motor and cardiopulmonary function, joint mobility and extra-muscular manifestations in limb girdle muscular dystrophies** K. Sofou ; J. Weichbrodt; A. Kroksmark; M. Tulinius
P.171	**A clinical outcome study for dysferlinopathy: biobanking samples collected through a collaborative international multi site study** H. Hilsden ; U. Moore; D. Cox; J. Day; K. Jones; D. Bharucha-Goebel; A. Pestronk; M. Walter; T. Stojkovic; E. Bravver; J. Mendell; L. Rufibach; C. Paradas; J. Diaz-Manera; E. Pegoraro; V. Straub
P.172	**Identification and characterisation of CAPN3 splicing defect mutations in unexplained cases of LGMD patients from the MYO-SEQ project** J. Duff; A. Topf ; D. Cox; S. Specht; M. Mroczek; E. England; K. Chao; D. MacArthur; M. MYO-SEQ Consortium; V. Straub
P.173	**Ketogenic diet ameliorates dysferlinopathy phenotype in Dysf -/-mice by promoting mitochondrial function** C. Astorga; C. Basualto-Alarcon; P. Caviedes; J. Bevilacqua ; J. Cárdenas
P.174	**Establishing divergent phenotypes in limb girdle muscular dystrophies** M. Iammarino ; N. Miller; L. Alfano; K. Lehman; L. Rodino-Klapac; J. Mendell; L. Lowes
P.175	**Detection and interpretation of variants in dystroglycanopathy-causing genes in a cohort of 1,566 patients with unexplained limb-girdle muscle weakness** A. Topf ; A. Casasus; R. Barresi; K. Johnson; M. Mroczek; J. Duff; L. Phillips; E. England; L. Xu; E. Valkanas; D. MacArthur; V. Straub
P.176	**Limb-girdle muscular dystrophies (LGMD) C, D and E: long term clinical follow up, anatomopathological and molecular genetic study of in a cohort of 35 pediatric patients** C. Ortez ; A. Martinez; L. Carrera; D. Natera; J. Exposito; J. Colomer; C. Jou; A. Codina; C. Jimenez - Mallebrera; P. Gallano; L. González; D. Yubero; A. Nascimento
P.177	**Measuring what matters in dysferlinopathy – Linking functional ability to patient reported outcome measures** A. Mayhew ; M. James; H. Hilsden; H. Sutherland; M. Jacobs; S. Spuler; J. Day; K. Jones; D. Bharucha-Goebel; E. Salort-Campana; A. Pestronk; M. Walter; C. Paradas; T. Stojkovic; M. Mori-Yoshimura; E. Bravver; J. Diaz Manera; E. Pegoraro; J. Mendell; L. Rufibach; V. Straub
P.178	**A novel mutation in TNPO3 causes congenital limb girdle myopathy with slow progression** A. Vihola ; J. Palmio; O. Danielsson; S. Penttilä; D. Louiselle; S. Pittman; C. Weihl; B. Udd
P.179	**European collaboration on clinical and genetic heterogeneity of sarcoglycanopathies** J. Alonso-Perez ; A. Nascimiento-Osorio; C. Ortez-Gonzalez; M. Guglieri; V. Straub; B. Schoser; B. Udd; A. Damico; J. Haberlova; C. Dominguez-Gonzalez; G. Tasca; A. Lopez-Munain; R. Fernandez-Torron; M. Bela; H. Kinga; C. Semplicini; E. Pegoraro; I. Richard; J. Diaz-Manera
P.180	**Paraspinal muscle affection in limb-girdle muscular dystrophy type 2I patients** K. Revsbech ; T. Khawajazada; J. Borch; K. Rudolf; A. Sheikh; J. Dahlqvist; N. Løkken; N. Witting; J. Vissing
P.181	**Focal muscle contraction of biceps brachii - a clinical sign highly specific to dysferlinopathy** R. El Sherif ; R. Hussein; M. Gamal; I. Nishino
P.182	**Autosomal recessive limb-girdle and Miyoshi muscular dystrophies in the Netherlands: the clinical and molecular spectrum of 244 patients** L. ten Dam ; W. Frankhuizen; W. Linssen; C. Straathof; E. Niks; C. Faber; J. Fock; J. Kuks; e. Brusse; I. de Coo; N. Voermans; A. Verrips; J. Hoogendijk; W. van der Pol; D. Westra; M. de Visser; A. van der Kooi; H. Ginjaar
P.183	**Functional progression in dyferlinopathy: results of a 3-year natural history study** M. Jacobs ; M. James; A. Mayhew; H. Hilsden; H. Sutherland; S. Spuler; J. Day; K. Jones; D. Bharucha-Goebel; E. Salort-Campana; A. Pestronk; M. Walter; C. Paradas; T. Stojkovic; M. Mori-Yoshimura; E. Bravver; J. Diaz Manera; E. Pegoraro; j. Mendell; L. Rufibach; V. Straub
P.184	**Clinical outcome study for dysferlinopathy: a longitudinal examination of the upper limb involvement using physiotherapy outcome measures and T1w MRI** M. James ; R. Fernández-Torrón; A. Mayhew; L. Alfano; R. Muni-Lofra; T. Duong; E. Maron; M. Hutchence; B. Vandervelde; B. Mendez; S. Holsten; C. Sakamoto; I. Pedrosa Belmonte; S. Thiele; A. Canal; C. Semplicini; C. Seiner; L. Lowes; V. Straub; J. Diaz-Manera
P.185	**The clinical outcome study for dysferlinopathy: quantitative MRI and physiotherapy outcomes to capture disease progression** M. James ; F. Smith; H. Reyngoudt; I. Wilson; A. Mayhew; R. Fernández-Torrón; E. Araujo; T. Stojkovic; A. Blamire; P. Carlier; V. Straub
P.186	**Two patients with PURA syndrome in a large cohort of patients with unexplained muscle disease** M. Mroczek ; A. Töpf; D. Zafeiriou; A. Roos; E. Bartels; N. Kohlschmidt; J. Duff; V. Straub
P.187	**CAPN3 c.598_612delTTCTGGAGTGCTCTG: another CAPN3 dominant variant?** M. Mroczek ; A. Töpf; J. Duff; R. Barresi; J. Hudson; E. England; K. Chao; D. MacArthur; V. Straub
P.188	**The clinical outcome study for dysferlinopathy: pregnancy in dysferlinopathy** U. Moore ; M. James; M. Jacobs; A. Mayhew; S. Spuler; J. Day; D. Bharucha-Goebel; T. Stojkovic; J. Mendell; V. Straub

	Outcome measures (P.189-205) Andersen Hall
P.189	**Measuring fatigability during real-life assessment in spinal muscular atrophy** L. Servais ; T. Gidaro; A. Seferian; E. Gasnier; A. Daron; A. Ulinici; M. Annoussamy; M. Grelet; D. Vissiere
P.190	**Feasibility, reliability and convergent validity for digital biomarkers captured via a smartphone application (app) to assess motor behaviors in individuals with spinal muscular atrophy (SMA) in the JEWELFISH trial** D. Fischer; S. Ewing ; D. Wolf; G. Pointeau; Y. Zhang; F. Lipsmeier; Y. Qian; I. Eng; R. Salazar; S. Dunaway Young; J. Sprengel; C. Czech; C. Gossens; M. Lindemann
P.191	**Maximal stride velocity detects positive and negative changes over 6- month-time period in ambulant patients with Duchenne muscular dystrophy** L. Servais ; T. Gidaro; A. Seferian; E. Gasnier; A. Daron; A. Ulinici; M. Grelet; D. Vissiere
P.192	**The 100-meter timed test: responsiveness to change, predicting loss of ambulation, and data-driven phenotypes** L. Alfano ; N. Miller; M. Iammarino; M. Moore-Clingenpeel; M. Waldrop; K. Flanigan; J. Mendell; L. Lowes
P.193	**The neuromuscular gross motor outcome as an outcome measure in spinal muscular atrophy** L. Alfano ; N. Miller; M. Iammarino; L. Lowes
P.194	**Activity monitoring in Duchenne and limb girdle muscular dystrophies: A longitudinal natural history study** N. Miller ; L. Alfano; M. Iammarino; M. Moore-Clingenpeel; L. Lowes
P.195	**Physical activity monitoring using wrist-worn accelerometer in the assessment of patients with myositis** O. Landon-Cardinal; D. Bachasson; P. Guillaume-Jugnot; M. Vautier; N. Champtiaux; B. Hervier; A. Rigolet; O. Benveniste; J. Hogrel; Y. Allenbach
P.196	**Estimating clinically meaningful change thresholds in the NORTH STAR ambolatory assessment (NSAA) and four-stairclimb (4SC) in Duchenne muscular dystrophy (DMD)** B. Wong ; J. Signorovitch; H. Staunton; R. Ong; M. Rabbia; G. Sajeev; Z. Yao; I. Dieye; S. Ward
P.197	**Use of the D3-creatine dilution test as a non-invasive and accurate measurement of total body muscle mass in Duchenne muscular dystrophy** E. Smith ; M. Shankaran; M. Hellerstein; K. Brown; C. Morris; V. Ricotti; W. Evans
P.198	**Dynamic arm study: quantitative description of upper extremity function and activity of people with spinal muscular atrophy** M. Janssen; L. Peeters; I. de Groot
P.199	**Accelerometer prediction equations in boys with Duchenne muscular dystrophy: importance of disease-specific equations** R. Bendixen ; A. Hartman; N. Little; M. Feltman
P.200	**Feasibility and baseline values of continuous movement measurement in patients with centronuclear myopathy by using ActiMyo®** M. Annoussamy ; E. Gasnier; J. Baets; U. Schara; A. Grangé; C. Lilien; V. Chë; D. Duchêne; T. Gidaro; A. Seferian; A. Hernandez; C. de Lattre; A. D'Amico; A. Behin; M. Grelet; J. Hogrel; H. Landy; A. Buj-Bello; C. Freitag; L. Servais
P.201	**Home-based video assessment of the quality of movement of patients with Duchenne: scoring scale development** M. Leffler ; M. Contesse; L. Lowes; L. Dalle Pazze; T. Seckler; C. McSherry
P.202	**A composite prognostic score for time to loss of walking ability in Duchenne muscular dystrophy (DMD)** N. Goemans ; J. Signorovitch; G. Sajeev; M. Fillbrunn; H. Wong; S. Ward; C. McDonald; E. Mercuri
P.203	**Towards high-resolution clinical digital biomarkers for Duchenne muscular dystrophy** V. Ricotti ; B. Kadirvelu; V. Selby; T. Voit; A. Faisal
P.204	**Full-body behaviour analytics reveals DMD disease state within the first few steps of the 6-minute-walk test** V. Ricotti ; B. Kadirvelu; S. Rabinowicz; V. Selby; T. Voit; A. Faisal
P.205	**Daily life digital biomarkers for longitudinal monitoring of Duchenne muscular dystrophy with wearable sensors** V. Ricotti ; B. Kadirvelu; C. Auepanwiriyakul; S. Zeng; V. Selby; T. Voit; A. Faisal
	E-Posters – Muscle imaging (EP.29-38) Lumbye Hall
EP.29	**Strength and quality of lower extremity muscles in patients with Myotonic dystrophy - an MRI study** C. Steenkjaer ; R. Mencagli; M. Vaeggemose; H. Andersen
EP.30	**Usefulness of extraocular muscle imagery in the diagnosis of myasthenia gravis and chronic progressive external ophthalmoplegia** M. Lepetit; F. Toulgoat; S. Wiertlewski; Y. Pereon; A. Magot
EP.31	**Characteristic findings of skeletal muscle MRI in caveolinopathies** K. Ishiguro ; T. Nakayama; M. Yoshioka; T. Murakami; S. Kajino; M. Shichiji; T. Sato; N. Fukuyo; S. Kuru; M. Osawa; S. Nagata; M. Okubo; N. Murakami; Y. Hayashi; I. Nishino; K. Ishigaki
EP.32	**Quantitative assessment of muscle involvement in limb girdle muscular dystrophy 2Aand 2B** T. Nakayama; A. Ishiyama; S. Kuru
EP.33	**Quantitative analysis of muscle bundles of patients with myotonic dystrophy type 1 (DM1), using CT impairment ratio** T. Nakayama ; S. Kuru
EP.34	**The companion analysis of MR and ultrasound images could suggest small-size pathological findings** T. Kurashige ; T. Kanbara-Murao; T. Iryo; T. Watanabe; T. Sugiura; T. Mukai; H. Ueno; H. Maruyama; T. Torii

EP.35	**Patterns of muscle involvement in SMA patients** C. Brogna; L. Cristiano; T. Verdolotti; A. Pichiecchio; C. Cinnante; A. Berardinelli; V. Sansone; E. Albamonte; L. Sconfienza; G. Comi; M. Pera; M. Garibaldi; G. Antonini; T. Tartaglione; M. Pane; E. Mercuri
EP.36	**PATCHS MRI score correlates with clinical severity in facioscapulohumeral muscular dystrophy** Y. Liu; D. Yue; W. Zhu ; J. Li; S. Cai; S. Luo; J. Xi; J. Lin; J. Lu; L. Zhou; Z. Liang; J. Lu; C. Zhao
EP.37	**Whole-body MRI pattern in pediatric patients with sarcoglycanopathies** L. Costa Comellas ; J. Escudero Fernandez; M. Alvarez Molinero; S. Ferrer Aparicio; P. Romero Duque; A. Sanchez-Montañez; D. Gómez-Andrés; A. Macaya Ruiz; F. Munell
EP.38	**Muscle MRI in myotonic dystrophy type 1 (DM1)** M. Garibaldi ; E. Bucci; L. Cristiano; S. Morino; L. Fionda; F. Vanoli; L. Leonardi; E. Pennisi; T. Tartaglione; A. Petrucci; G. Antonini
	E-Posters – Clinical trial readiness (EP.39-50) — **Andersen Hall**
EP.39	**The development of a weak cough pathway for children and young people with neuromuscular disorders: a literature review and pilot study.** C. Frimpong-Ansah ; J. Watson; A. Majumdar; A. Prendiville
EP.40	**The strategic role of the research nurse: No research nurse, no trial** M. Cubells; A. Rodriguez ; L. Sole; A. Nascimento
EP.41	**Project HERCULES: could international collaboration on value assessment change clinical practice?** K. Abrams; J. Carlton; F. Chandler; E. Crossley ; M. Crowther; S. Ghosh; J. Godfrey; M. Guglieri; M. Hill; A. Johnson; J. Mumby-Croft; P. Powell; D. Rowan; F. Woodcock
EP.42	**Health utility index scores in patients with spinal muscular atrophy: findings from the 2019 cure SMA community update survey** L. Belter ; R. Cruz; J. Jarecki
EP.43	**Review of the regional orthotic services for children with neuromuscular disorders** Z. Alhaswani ; R. Rabb; H. McMurchie; H. Roper; D. Parasuraman
EP.44	**Improving healthcare professionals' capacity for facilitating self-determination among children with neuromuscular conditions: assessing the need** L. McAdam ; K. Schultz; K. Bell; P. Sparling; C. Campbell; A. McPherson; S. Kingsnorth; D. Greenspoon
EP.45	**Neuromuscular disease services crossing boundaries: a multi-disciplinary network approach in the United Kingdom.** A. Merrison ; S. Brady; N. Grose; J. Ashworth; A. Majumdar
EP.46	**MuscleViz: free open-source software for muscle weakness visualization** J. Wittenbach; B. Cocanougher; P. Yun; A. Foley ; C. Bönnemann
EP.47	**Sharing evidence-based physiotherapy for neuromuscular patients through a digital learning platform: Description of a service development project** A. Rosenberger ; I. Lund; H. Hæstad
EP.48	**Informing paediatric clinical research participants: an innovative approach** B. Crow ; C. Turner; D. Athanasiou; E. Vroom; V. Straub; R. Bendixen; J. Haberlova; P. Clemens; M. Guglieri
EP.49	**The muscular dystrophy association neuromuscular observational research (MOVR) Data Hub** G. Pavlath ; M. MOVR Team
EP.50	**A service evaluation of orthotic provision for neuromuscular patients at the John Walton neuromuscular research centre** D. Moat ; M. McCallum; M. James; J. Sodhi; C. Hall; C. Marini-Bettolo; R. Muni-Lofra; A. Mayhew
	E-Posters – Outcome measures (EP.51-55) — **Andersen Hall**
EP.51	**Expert advice to improve clinical trials: from TACT to ACT** J. Lee ; V. Straub; C. Turner; A. de Luca
EP.52	**Overnight pulse oximetry for respiratory progression screening in a neuromuscular service** M. McCallum; A. Mayhew; D. Moat; J. Sodhi; M. James; S. Specht; M. Guglieri; V. Straub; C. Marini-Bettolo; R. Muni-Lofra
EP.53	**Evaluation of the reliability and validity of the facioscapulohumeral composite outcome measure (FSH-COM) in children with facioscapulohumeral muscular dystrophy (FSHD): Study protocol** K. de Valle; I. Woodcock ; M. Ryan; K. Carroll; F. Dobson; J. McGinley
EP.54	**Assessment of trunk muscle strength in patients with muscular dystrophies using stationary and hand-held dynamometry: a test-retest reliability study** K. Rudolf ; A. Sheikh; K. Knak; N. Witting; J. Vissing
EP.55	**A critical review of tools assessing health related quality of life, activity of daily living and caregiver burden in SMA** S. Messina; A. Frongia; L. Antonaci; M. Pera; G. Coratti; M. Pane; A. Mayhew; A. Pasternak; M. Civitello; R. Finkel; F. Muntoni; E. Mercuri

18.30 - 20.00 Industry symposium 2 ■ Axelborg Hall

Full Programme

Thursday 3rd October

		Room: Concert Hall
07.00		**Registration desk opens**
08.30 - 09.30		**Extra-muscular manifestations in NMD (I.5-6)** **Invited lectures** **Chairpersons: Nathalie Goemans and Francesco Muntoni**
08.30 - 09.00	I.5	**Combining genetics, neuropsychology and neuroimaging to better understand the brain involvement in Duchenne muscular dystrophy** Nathalie Doorenweerd, Leiden University Medical Center, The Netherlands and Newcastle University, United Kingdom
09.00 - 09.30	I.6	**Arthrogryposis multiplex congenita; new genes and old acquaintances** Markus Schülke-Gerstenfeld, Department of Neuropediatrics and NeuroCure Clinical Research Center, Charité-Universitätsmedizin, Berlin, Germany
09.30 - 10.00		**Morning refreshments** – Concert Hall Foyer
10.00 - 11.00		**Extra-muscular manifestations in NMD (I.7-8)** **Invited lectures** **Chairpersons: Mariz Vainhof and Chris Weihl**
10.00 - 10.30	I.7	**Improved survival of patients with neuromuscular conditions by cardiac preventive treatments** Karim Wahbi, Cochin Hospital, Paris Descartes University, Myology Institute, Paris
10.30 - 11.00	I.8	**Reversible formation of TDP-43 assemblies during skeletal muscle regeneration** Bradley Olwin, University of Colorado Boulder, United States of America
11.00 - 11.15		Break
11.15 - 12.30		**Extramuscular manifestation in NMD (O.12-16)** Parallel oral Presentations 3 – Concert Hall **Chairpersons: Anne Connolly and Yoram Nevo**
	O.12	**Contribution of cardiac defects to spinal muscular atrophy pathology: a human tissue study** C. Alves ; R. Garner; F. Nery; J. Siranosian; A. Johnstone; K. Swoboda
	O.13	**Nemaline myopathy patients with mutations in KBTBD13 display a cardiac phenotype** J. de Winter ; K. Bouman; M. van den Berg; J. Strohm; J. Jongbloed; W. van der Roest; J. van Wijngaarden; J. Timmermans; E. Kamsteeg; B. Van Engelen; R. Van der Pijl; H. Granzier; K. van Spaendonck-Zwarts; N. Voermans; C. Ottenheijm
	O.14	**B3GNT4 deficiency: A new α-dystroglycanopathy causing late-onset progressive brain atrophy and muscular dystrophy** J. Vissing ; A. Töpf; V. Straub; T. Krag
	O.15	**MRI as a tool to indicate early diaphragmatic weakness in adult patients with Pompe disease** L. Harlaar ; P. Ciet; G. van Tulder; A. Pittaro; H. van Kooten; N. van der Beek; E. Brusse; P. Wielopolski; M. de Bruijne; A. van der Ploeg; H. Tiddens; P. van Doorn
	O.16	**Diagnosis of fetal akinesia and arthrogryposis by panel sequencing and functional genomics** G. Ravenscroft ; J. Clayton; F. Faiz; D. Milnes; R. Cincotta; P. Moon; P. McGrath; A. Colley; K. Chao; N. Laing; M. Davis
12.30 - 14.00		**Lunch, exhibition and posters** ■ Andersen Hall, ■ Lumbye Hall and ■ Gemyse
13.00		
13.30 - 15.00		
15.00 - 18.00		**Poster viewing session and afternoon refreshments** ■ Andersen Hall, ■ Lumbye Hall and ■ Gemyse

		Room: Axelborg Hall
11.15 - 12.30		**New genes and diseases (O.17-21)** Parallel oral Presentations 4 - Axelborg Hall **Chairpersons: Carina Wallgreen-Petterson and Nigel Laing**
	O.17	**Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy** G. Yoon ; M. Estañ; E. Fernandez-Nuñez; M. Zak; M. Esteban; S. Donkervoort; C. Hawkins; J. Caparros-Martin; D. Saade; Y. Hu; V. Bolduc; K. Chao; G. Otaify; S. Temtamy; M. Aglan; M. Issa; C. Bönnemann; P. Lapunzina; V. Ruiz-Perez
	O.18	**Recessive mutations in the myosin chaperone UNC-45B impair muscle myofibrillar integrity, manifesting as progressive myopathy with eccentric cores** S. Donkervoort ; Y. Hu; X. Lornage; C. Kutzner; M. Mroczek; S. Neuhaus; N. Kuntz; A. Töpf; S. Monges; F. Lubieniecki; K. Chao; J. Böhm; N. Romero; V. Straub; J. Laporte; A. Foley; C. Ottenheijm; T. Hoppe; C. Bönnemann
	O.19	**PAX7 deficiency causes mild congenital myopathy with rigid spine and respiratory insufficiency** H. Amthor ; A. Marg; H. Escobar; S. Grunwald; E. Metzler; J. Kieshauer; E. Malfatti; D. Mompoint; S. Quijano-Roy; R. Carlier; S. Spuler
	O.20	**Mutations in ACTN2 gene cause a novel form of adult-onset distal myopathy** M. Savarese ; J. Palmio; J. Poza; J. Weinberg; M. Olive; A. Cobo; A. Vihola; P. Jonson; J. Sarparanta; F. García-Bragado; J. Urtizberea; P. Hackman; B. Udd
	O.21	**Immune checkpoint inhibitor-associated myositis: a new entity with a poor prognosis** C. Anquetil ; J. Salem; B. Lebrun-Vignes; D. Johnson; A. Mammen; W. Stenzel; S. Louis-Léonard; O. Benveniste; J. Moslehi; Y. Allenbach
12.30 - 14.00		**NMD Editorial Board Meeting** ■ Bokssalen at Axelborg Hall
13.00		Lunch ■ Axelborg Hall
13.30 - 15.00		**Industry symposium 3**

Full Programme

Friday 4th October

		Room: Concert Hall
08.00		**Registration desk opens**
08.30 - 10.00		**Advances in the treatment of neuromuscular disorders (I.9-11)** Invited lectures **Chairpersons: Annemieke Rus and Volker Straub**
08.30 - 09.00	I.9	Muscle MRI as an outcome measure **Jordi Díaz Manera, Hospital de la Santa Creu i Sant Pau de Barcelona, Spain**
09.00 - 09.30	I.10	Gene variant databases: driving research, diagnosis, treatment and interaction **Johan den Dunnen, University of Leiden, The Netherlands**
09.30 - 10.00	I.11	AAV vectors and the host immune system: a complicated relationship **Federico Mingozzi, Genethon & INSERM U951**
10.00 - 10.30		**Morning refreshments** – Concert Hall Foyer
10.30 - 12.30		**Pre clinical approaches and early clinical results (O.22-29)** Parallel oral presentations 5 – Concert Hall **Chairpersons: Maggie Walter and Ichizo Nishino**
	O.22	**First-in-human study of ATB200/AT2221 in patients with Pompe disease: 24-month functional assessment results from the ATB200-02 trial** B. Schoser ; P. Kishnani; D. Bratkovic; B. Byrne; P. Clemens; O. Goker-Alpan; X. Ming; M. Roberts; P. Schwenkreis; K. Sivakumar; A. van der Ploeg; S. Sitaraman; J. Barth; H. Lagast; T. Mozaffar
	O.23	**A 10-year prospective study on the effects of enzyme replacement therapy in adult Pompe patients** L. Harlaar ; J. Hogrel; B. Perniconi; M. Kruijshaar; D. Rizopoulos; N. Taouagh; A. Canal; E. Brusse; P. van Doorn; A. van der Ploeg; P. Laforêt; N. van der Beek
	O.24	**Biodistribution of onasemnogene abeparvovec (AVXS-101) DNA, mRNA, and SMN protein in human tissue** B. Kaspar; G. Thomsen; C. Hsieh; J. Do; S. Solano; B. Chu; B. Barkho; M. Fugere; P. Kaufmann ; K. Foust; A. Kaspar; J. L'Italien; D. Sproule; D. Feltner; W. Chung; A. Burghes; V. McGovern; R. Hevner; M. Conces; J. Mendell
	O.25	**Phase 1 clinical trial of losmapimod in FSHD: safety, tolerability and target engagement** M. Mellion ; L. Ronco; D. Thompson; M. Hage; S. Brooks; E. van Brummelen; L. Pagan; U. Badrising; B. Van Engelen; G. Groeneveld; D. Cadavid
	0.26	**The α-actinin-3 deficiency is related to early onset of dilated cardiomyopathy in Duchenne muscular dystrophy patients** M. Nagai ; H. Awano; T. Yamamoto; R. Bo; H. Nishio; M. Matsuo; K. Iijima
	O.27	**Ultra-exome: a new tool to solve the unsolved NMD** A. Torella; M. Onore; F. Del Vecchio Blanco; F. Musacchia; G. Piluso; V. Nigro
	O.28	**Safety and tolerability of suvodirsen (WVE-210201) in patients with Duchenne muscular dystrophy: Results from a Phase 1 clinical trial** K. Wagner ; H. Phan; L. Servais; T. Gidaro; L. Cripe; M. Eagle; F. Muntoni; V. Straub; P. Lonkar; S. Schmitz; S. Lake; X. Hu; M. Panzara
	O.29	**Engineering stem cell fate determinants for myogenic cell therapies: Notch and PDGF signalling promote stemness and migration of muscle satellite cells and human iPS cell derived myogenic progenitors** M. Gerli; L. Moyle; G. Ferrari; H. Sakai; P. Ala; P. DeCoppi; S. Tajbakhsh; G. Cossu; F. Muntoni; F. Tedesco
12.30 - 14.30		**Lunch, exhibition and posters** ■ Andersen Hall, ■ Lumbye Hall and ■ Gemyse
13.00		
13.15 - 14.45		
15.00 - 16.15		**Poster session 3** ■ Andersen Hall, ■ Lumbye Hall and ■ Gemyse

		Room: Axelborg Hall
10.30 - 12.30		**Diagnostic and experimental treatment approaches (O.30-37)** Parallel oral Presentations - Axelborg Hall **Chairpersons: Sabrina Sacconi and Peter Van den Bergh**
	O.30	**Dysregulation of cytokines and calcium dominate the molecular pathology of sporadic inclusion body myositis** M. Johari ; M. Savarese; A. Vihola; J. Palmio; M. Jokela; J. Buzkova; L. Pihlajamaki; A. Wartiovaara; P. Hackman; B. Udd
	O.31	**Proteomic profiling unravels a key role of CD74, CD163 and STAT1 in sporadic inclusion body myositis** C. Preusse ; A. Roos; D. Hathazi; H. Goebel; W. Stenzel
	O.32	**Genome editing of expanded CTG repeats within the human DMPK gene reduces nuclear RNA foci in muscle of DM1 mice** M. Lo Scrudato; K. Poulard; C. Sourd; S. Tomé; A. Klein; G. Corre; A. Huguet; D. Furling; G. Gourdon; A. Buj-Bello
	O.33	**Subcutaneous immunoglobulin in myasthenia gravis: results of a North American open label study** M. Pasnoor; V. Bril; T. Levine; J. Trivedi; N. Silvestri; M. Phadnis; D. Saperstein; S. Nations; H. Katzberg; g. Wolfe; L. Herbelin; K. Higgs; A. Heim; J. Statland; R. Barohn; M. Dimachkie
	O.34	**Leiomodin-3 (LMOD3) deficiency affects contractile function and structure of fast muscle fibres** M. Yuen ; L. Schultz; R. Mayfield; S. Shen; S. Conijn; J. de Winter; S. Bogaards; R. van der Pijl; M. Meskovic; I. De Vries; H. Granzier; C. Gregorio; C. Ottenheijm
	O.35	**Disruption of bioavailability of TGF α in collagen VI-related muscular dystrophy** P. Mohassel ; J. Rooney; M. Nalls; P. Yun; D. Sleboda; T. Roberts; Y. Zou; C. Bönnemann
	O.36	**Defective lysosome homeostasis during autophagy causes skeletal muscle disease** M. McGrath ; M. Eramo; R. Gurung; A. Sriratana; S. Feeney; S. Gehrig; G. Lynch; M. Lazarou; C. McLean; C. Mitchell
	O.37	**AI-based muscle histopathologist can differentiate major muscular dystrophies better than human** Y. Kabeya ; T. Iwamori; S. Yonezawa; Y. Takeuchi; H. Nakano; Y. Nagisa; M. Okubo; M. Inoue; R. Tokumasu; I. Ozawa; A. Takano; I. Nishino
13.00		Lunch ■ Axelborg Hall
13.15 - 14.45		**Industry symposium 4**

Poster Session 3

15.00 - 16.15 Poster Area, locations: Lumbye Hall | Gemyse | Andersen Hall

	Clinical studies and quality of life (P.207-228) Andersen Hall
P.207	**Cognitive assessment in spinal muscular atrophy type 1-2 using eye tracking system: results of a prospective multicenter study and comparison with age matched control and Down syndrome cohorts** L. Paternoster; S. Baijot; G. Deliens; M. Kissine; N. Goemans; S. Paquay; L. Servais; N. Deconinck
P.208	**Improving recognition of spinal muscular atrophy: a retrospective case note review** M. Guglieri; H. van Ruiten ; M. Scoto; A. Manzur; P. Munot; F. Muntoni; A. Alanzi; C. Brusa; C. Marini-Bettolo; S. Specht; V. Straub
P.209	**Event-free survival and motor milestone achievement following onasemnogene abeparvovec and nusinersen interventions contrasted to natural history for spinal muscular atrophy t1 (SMA1) patients** O. Dabbous ; D. Sproule; D. Feltner; F. Ogrinc; M. Menier; M. Droege; B. Maru; F. Khan; R. Arjunji
P.210	**Burden of illness of spinal muscular atrophy type 1 (SMA1)** M. Droege; O. Dabbous ; R. Arjunji; J. Seda; M. Gauthier-Loiselle; M. Cloutier; D. Sproule
P.211	**Pilot study of genetic newborn screening for spinal muscular atrophy in Germany: clinical results after more than a year** W. Mueller-Felber ; H. Kölbel; O. Schwartz; A. Blaschek; B. Olgemüller; E. Harms; W. Röschinger; J. Durner; D. Gläser; S. Burggraf; U. Nennstiel; B. Wirth; U. Schara; M. Becker; K. Vill
P.212	**Mortality in patients with spinal muscular atrophy over the last 10 years: the UK experience** S. Specht; M. Scoto; A. Childs; G. Eglon; L. Hastings; K. Pysden; S. Manning; C. Jimenez; P. Munot; C. Brusa; M. Turner; M. Guglieri; A. Manzur; F. Muntoni; V. Straub; C. Marini-Bettolo
P.213	**Ambulation status, role participation and caregiver assistance among individuals with spinal muscular atrophy type III: Results from the 2018 cure SMA membership survey** L. Belter ; J. Jarecki; R. Cruz; C. McCarthy O'Toole; K. O'Brien; S. Reyna; S. Hall; A. Paradis
P.214	**Presymptomatic spinal muscular atrophy: reality or myth?** L. Lowes ; L. Alfano; D. Chen; L. Nelson; N. Miller; M. Iammarino; M. McColly; J. Mendell; S. Rust
P.215	**Utility of functional outcomes in adults with spinal muscular atrophy** L. Alfano; N. Miller; M. Iammarino; L. Lowes
P.216	**SMN genes molecular testing in a cohort of 1546 subjects tested for genetic diagnosis and trial enrollment** A. Margutti; A. Venturoli; M. Neri; F. Fortunato; F. Gualandi; P. Rimessi; A. Ferlini
P.217	**Patterns of disease progression in patients with spinal muscular atrophy type 3 using the motor function measure** A. Seferian ; M. Annoussamy; A. Chabanon; C. Cances; N. Goemans; A. Daron; U. Schara; Y. Pereon; J. Cuisset; M. El-Khairi; T. Seabrook; C. Vuillerot; L. Servais
P.218	**Utility based health related quality of life in children and adolescents with spinal muscular atrophy** D. Love; R. Hicks; Y. Wei; E. ZapataAldana; S. AlMobarak; C. Campbell
P.219	**Bone mass, nutrition and motor function in children and adolescents with spinal muscular atrophy type II and III** A. Kroksmark ; A. Ekelund; J. Weichbrodt; A. Ekström; A. Söderpalm
P.220	**The revised Hammersmith scale (RHS) for spinal muscular atrophy: longitudinal trajectories in a large international cohort of patients with type 2 and 3 SMA** D. Ramsey ; M. Scoto; A. Mayhew; R. Muni Lofra; M. Main; E. Milev; E. Mazzone; J. Montes; A. Glanzman; A. Pasternak; T. Duong; M. Civitello; G. Coratti; V. Straub; J. Day; B. Darras; D. De Vivo; R. Finkel; E. Mercuri; F. Muntoni
P.221	**Trajectories of disease progression in ambulant and non ambulant SMA: 12 month follow-up** E. Mercuri ; G. Coratti; M. Pera; M. Scoto; N. Goemans; M. Pane; A. D'amico; V. Sansone; S. Messina; A. Nascimiento; E. Bertini; R. Finkel; B. Darras; D. De Vivo; J. Day; F. Muntoni
P.222	**Long term progression in type II spinal muscular atrophy: a retrospective observational study** E. Mercuri ; S. Lucibello; M. Pera; S. Carnicella; G. Coratti; R. De Sanctis; E. Mazzone; N. Forcina; L. Fanelli; G. Norcia; L. Antonaci; A. Frongia; M. Pane
P.223	**Respiratory function in SMA type 2 and non-ambulant SMA type 3, longitudinal data from the international SMA consortium (iSMAc)** F. Trucco ; D. Ridout; R. Finkel; E. Mercuri; M. Scoto; F. Muntoni; International SMA consortium
P.224	**Supportive thoraco-lumbar-sacral orthosis (TLSO) provision for spinal muscular atrophy (SMA) type 1 children treated with nusinersen** L. Abbott ; M. Main; A. Manzur; M. Scoto; F. Muntoni
P.225	**Reducing the diagnosis time of neonatal screening by optimizing the screening process: the southern Belgian experience** T. Dangouloff ; F. Boemer; J. Caberg; S. Di Fiore; P. Beckers; S. Marie; L. Marcelis; L. Servais
P.226	**Longitudinal study of the natural history of spinal muscle atrophy type 2 and 3** J. Exposito ; D. Natera-de Benito; L. Carrera; a. Frongia; m. Alarcón; A. Borras; J. Armas; L. Martorell; O. Moya; N. Padros; S. Roca; M. Vigo; J. Medina; J. Colomer; C. Ortez; A. Nascimento
P.227	**Secondary clinical outcomes of spinal surgery and satisfaction in patients with spinal muscular atrophy (SMA) II and non-ambulant III** C. Brusa ; J. De Graaf; A. Manzur; M. Main; E. Milev; M. Iodice; D. Ramsey; S. Tucker; T. Ember; R. Nadarajah; F. Muntoni; M. Scoto

P.228	**General movements assessment in infants with spinal muscular atrophy: a pilot study** B. Yardımcı Lokmano˘glu; N. Bulut; G. Aydın ; G. Sırtba¸s; F. Kabakçı; D. Porsnok; A. Mutlu; I. Gürbüz; H. Topaloglu; O. Yılmaz; A. Karaduman
	Congenital myopathies: RYR1 and Titin (P.229-243) — **Lumbye Hall**
P.229	**Electron microscopy characterisation of triads in RYR1 rhabdomyolysis-myalgia syndrome** T. Evangelista ; C. Labasse; M. Beuvin; G. Brochier; A. Madelaine; J. Pastor; N. Romero
P.230	**Phenotype and pathological variability in RYR1 -related myopathy with compound heterozygous variants in Japan** A. Ozaki ; A. Ishiyama; E. Takeshita; Y. Shimizu-Motohashi; H. Komaki; S. Noguchi; I. Nishino; M. Sasaki
P.231	**Paternal uniparental disomy of chromosome 19 unmasking a recessive variant in RYR1 in a case of congenital core** M. Davis ; F. Faiz; K. Woodward; J. Clayton; T. Robertson; D. Azmanov; A. Cairns
P.232	**Hereditary myopathy with early respiratory failure in chinese population** W. Zhu ; S. Luo; Z. Wang; J. Lin; J. Xi; D. Yue; J. Lu; C. Zhao
P.233	**The phenotypical spectrum associated with the recurrent RYR1 c.12861_ 12869dup9 (p.T4288_A4290dup) mutation** M. Fernandez-Garcia ; E. Wraige; A. Radunovic; K. Knop; K. Flanigan; A. Sarkozy; F. Muntoni; H. Zhou; S. Treves; S. Riazi; N. Voermans; H. Jungbluth
P.234	**Disruptive recessive TTN missense mutations cause a wide range of clinico-pathological features** M. Rees ; A. Fukuzawa; R. Nikoopour; A. Kho; J. Qi; M. Fernandez-Garcia; E. Wraige; S. Cirak; M. Pfuhl; J. Fluss; F. Muntoni; A. Sarkozy; R. Schröder; N. Voermans; E. Wilichowski; E. Mathews; E. Oates; A. Ferreiro; H. Jungbluth; M. Gautel
P.235	**Clinico-pathological characterisation of CACNA1S related congenital myopathy in children** M. Pal-Magdics; A. Sarkozy; G. McCullagh; R. Mein; A. Childs; C. Longman; C. DeVile; R. Phadke; F. Muntoni; E. Matthews; P. Munot
P.236	**Myofibres with subsarcolemmal rims and/or central aggregates of mitochondria (SRCAM) are prevalent in congenital titinopathies** R. Phadke ; A. Sarkozy; E. Oates; R. Mein; I. Bodi; L. Feng; A. Manzur; N. Thomas; M. Illingworth; I. Mazanti; S. Ellard; C. Sewry; M. Gautel; H. Jungbluth; F. Muntoni
P.237	**Functional analyses and phenotype-genotype correlation studies in patients suspected of titinopathy** R. Juntas Morales ; A. Perrin; U. Walther-Louvier; P. Cintas; G. Sole; C. Espil; C. Cances; D. Renard; M. Arne-Bes; V. Rigau; A. Maues de Paula; E. Uro-coste; M. Martine Negrier; N. Leboucq; D. Thorel; C. Theze; H. Pegeot; M. Koenig; M. Cossee
P.238	**The Dubowitz Neuromuscular Centre experience in TTN gene analysis in UK patients with congenital myopathies** A. Sarkozy ; R. Mein; E. Oates; I. Zaharieva; H. Jungbluth; A. Manzur; S. Robb; P. Munot; L. Feng; C. Sewry; R. Phadke; F. Muntoni
P.239	**Therapy development for RYR1 related myopathies** P. Onofre-Oliveira ; S. Brennan; J. Volpatti; M. Garcia-Castaneda; A. Michelucci; L. Groom; N. Sabha; R. Dirksen; J. Dowling
P.240	**ASC-1 related myopathy: phenotypic spectrum and pathophysiology of an emerging congenital myopathy** R, Villar-Quiles ; I. Duband-Goulet; E. Cabet; L. Davignon; C. Genetti; T. Gidaro; A. Koparir; A. Yuksel; S. Coppens; N. Deconinck; E. Pierce-Hoffman; R. Juntas-Morales; M. Cossée; L. Servais; M. Olivé; A. Beggs; J. Böhm; A. Ferreiro
P.241	**Congenital titinopathy as a cause of severe to profound congenital weakness and early death** S. Coppens ; N. Deconinck; R. Phadke; C. Sewry; H. Kadhim; C. Tay; M. Bakshi; D. de Silva; N. Thomas; S. Park; C. French; M. Ward; Y. Arens; A. Manzur; G. Ravenscroft; N. Laing; E. Kamsteeg; M. Davis; F. Muntoni; E. Oates
P.242	**Phenotypic variability in a cohort of patients with TTN -related congenital myopathy** D. Natera de Benito ; C. Jou; C. Jimenez-Mallebrera; C. Ortez; A. Codina; L. Carrera Garcia; J. Exposito Escudero; J. Colomer Oferil; F. Palau; A. Nascimento
P.243	**Dusty core disease (DuCD): a novel morphological hallmark for RYR1 recessive myopathies** M. Garibaldi ; J. Rendu; J. Brocard; E. Lacene; J. Faurè; G. Brochier; M. Beuvin; C. Labasse; A. Madelaine; E. Malfatti; J. Bevilacqua; F. Lubieniecki; S. Monges; A. Taratuto; J. Laporte; I. Marty; G. Antonini; N. Romero
	Distal myopathies (P.244-257) — **Lumbye Hall**
P.244	**SYNE1 cerebellar ataxia "plus" syndrome: characterizing the skeletal muscle and cardiopulmonary phenotype** D. Saade ; E. Drakou; S. Donkervoort; B. Monteleone; C. Brasington; E. Wiggs; T. Lehky; A. Foley; C. Bönnemann
P.245	**Morphological, ultrastructural and western blot analysis in adult and child with PLEC1 -related myopathy** M. Beuvin; E. Lacène; C. Labasse; G. Brochier; A. Madelaine; R. Ben Yaou; T. Stojkovic; G. Sole; R. Juntas-Morales; M. Martin-Negrier; F. Duval; I. Nelson; J. Nectoux; F. Leturcq; M. Cossée; D. Sternberg; B. Eymard; G. Bonne; T. Evangelista; N. Romero
P.246	**Drosophila model of myosin myopathy rescued by overexpression of a TRIM-protein family member** H. Tajsharghi ; M. Dahl-Halvarsson; M. Olive; M. Pokrzyw; K. Ejeskär; R. Palmer; A. Uv
P.247	**Capturing disease progression in oculopharyngeal muscular dystrophy (OPMD)** R. Kroon ; J. Kalf; B. de Swart; C. Horlings; B. van Engelen
P.248	**CRISPR-Cas9 tagging allows the detection of endogenous gne in mice** A. Harazi ; L. Yakovlev; S. Mitrani-Rosenbaum
P.249	**Protective pathways in the kidney of Gne M743T/M743T mutated knock in mice are related to muscle genes** H. Benyamini; Y. Kling; L. Yakovlev; Z. Argov; I. Sela; S. Mitrani-Rosenbaum

P.250	**Phenotypic variability of GNE myopathy: stratification for clinical trial readiness** Y. Park; D. Kim; Y. Choi; J. Shin
P.251	**Canary Islands: Clinical data from a long homogenous cohort of OPMD patients with (GCN)15 expansion** J. Alonso-Perez ; J. Leon-Hernandez; A. Gutierrez-Martinez; D. Mendoza-Grimon; H. Perez-Perez; I. Hadjigeorgiou; F. Monton-Alvarez; J. Diaz-Manera
P.252	**LGMD, exercise intolerance, ptosis, ophthalmoplegia and dermatologic features: the phenotypic pleiotropy of plectinopathies in 8 French families** R. Ben Yaou ; T. Stojkovic; M. Cerino; F. Duval; R. Juntas-Morales; I. Nelson; M. Beuvin; E. Lacene; D. Sternberg; J. Nectoux; M. Martin-Negrier; M. Bartoli; M. Cossee; F. Leturcq; G. Sole; M. Krahn; N. Romero; B. Eymard; G. Bonne
P.253	**GNE myopathy in Turkey: clinical features and novel mutations** H. Durmus ; S. Ceylaner; F. Deymeer; Y. Parman; P. Oflazer-Serdaroglu
P.254	**The clinical and genetic spectrum of a UK cohort of paediatric and adult patients with MYH7 gene related skeletal myopathies** S. Pula ; K. Brennan; M. Bertoli; R. Quinlivan; A. Manzur; S. Robb; R. Mein; P. Munot; M. Main; L. Feng; R. Barresi; T. Polvikoski; C. Sewry; R. Phadke; V. Straub; F. Muntoni; A. Sarkozy
P.255	**Possible efficacy of rapamycin on myodegeneration in VCPDM mouse model** K. Hara ; S. Yamashita; X. Zhang; N. Tawara; T. Tanaka; Z. Zhang; Y. Matsuo; Y. Ando
P.256	**Steroid treatment may change natural history in young children with LMNA mutations and dropped head syndrome** M. Gomez Garcia de la Banda ; D. Natera-De Benito; I. Dabaj; R. Ben Yaou; C. Ortez; K. Wahbi; S. Maldonado; N. Clarke; R. Carlier; J. Colomer; G. Bonne; A. Nascimento; M. Monges; S. Quijano-Roy
P.257	**Analysis of 34 Korean patients of GNE mutations associated distal myopathy with rimmed vacuoles** Y. La ; J. Lee; H. Park; Y. Choi
	DMD clinical (P.258-268) — Gemyse
P.258	**Predictors of cardiac fibrosis in Duchenne and Becker muscular dystrophy carriers** M. Mah ; M. Slawinski; L. Cripe; E. Camino; S. Al-Zaidy; S. Hassan; J. Jackson; L. Lowes; M. Iammarino; N. Miller; L. Alfano; K. Lehman; J. Mendell; K. Hor
P.259	**Feasibility of osteoporosis clinical trials in Duchenne muscular dystrophy: a survey of the opinion of families, young adults and neuromuscular clinicians** M. Guglieri ; S. Wong; S. Joseph; N. Capaldi; M. Di Marco; J. Dunne; I. Horrocks; V. Straub; S. Ahmed
P.260	**Lean muscle mass changes in patients with Duchenne and Becker muscular dystrophies** B. Wong ; S. Summer; P. Horn; M. Rutter; I. Rybalsky; C. Tian; K. Shellenbarger; S. Hu; J. Bange; H. Kalkwarf
P.261	**Consistent long-term effect of idebenone in reducing respiratory function decline in advanced patients with Duchenne muscular dystrophy** G. Buyse ; L. Servais; O. Mayer; C. McDonald; T. Voit; E. Mercuri
P.262	**Crossing thresholds and changing rates of respiratory function decline are predictive of clinical outcomes in Duchenne muscular dystrophy** C. McDonald ; H. Gordish-Dressman; M. Leinonen; O. Mayer
P.263	**Juvenile Duchenne muscular dystrophy patients with abnormal Q wave are at risk for early onset of cardiac dysfunction** T. Yamamoto ; H. Awano; K. Fukuoka; M. Nagai; T. Imanishi; M. Matsuo; K. Iijima; J. Saegusa
P.264	**SYROS study – long-term reduction in rate of respiratory function decline in patients with Duchenne muscular dystrophy (DMD) treated with idebenone** L. Servais ; O. Mayer; C. McDonald; C. Straathof; U. Schara; T. Voit; E. Mercuri; G. Buyse
P.265	**Integrating clinical outcome measures, MRI and circulating biomarkers in subjects with Becker muscular dystrophy: a prospective longitudinal study** Z. Koeks; N. van de Velde; N. Verwey; V. Ricotti; K. Brown; G. Sajeev; J. Signorovitch; P. Spitali; H. Kan; E. Niks
P.266	**Bone mass acquisition and determinants of bone mineral density and fragility fractures in DMD patients** C. Panicucci ; N. Di Iorgi; G. Brigati; S. Notarnicola; C. Fiorillo; M. Pedemonte; C. Minetti; M. Maghnie; C. Bruno
P.267	**Modifiers of respiratory and cardiac function in the Italian Duchenne muscular dystrophy network and CINRG Duchenne natural history study** L. Bello ; G. D'Angelo; C. Bruno; A. Berardinelli; G. Comi; A. D'Amico; G. Astrea; L. Politano; G. Baranello; V. Sansone; S. Previtali; G. Vita; T. Mongini; A. Pini; E. Mercuri; C. Calore; A. Vianello; E. Hoffman; C. McDonald; E. Pegoraro
P.268	**Does 6-minute walk test distance correlate with motor function assessment and timed tests in ambulant boys with Becker muscular dystrophy?** L. Abbott ; V. Selby; A. Wolfe; M. Main; A. Manzur; F. Muntoni
	SMA biomarkers (P.269-277) — Andersen Hall
P.269	**Urinary titin is elevated in spinal muscular atrophy** H. Awano ; M. Nagai; T. Shirakawa; K. Osawa; T. Lee; Y. Takeshima; H. Nishio; M. Matsuo; K. Iijima
P.270	**Association between plasma phosphorylated neurofilament heavy chain and efficacy endpoints in the nusinersen NURTURE study** F. Muntoni ; C. Sumner; B. Darras; T. Crawford; R. Finkel; E. Mercuri; D. De Vivo; M. Oskoui; E. Tizzano; M. Ryan; Y. Liu; M. Petrillo; C. Stebbins; E. Koenig; S. Fradette; W. Farwell
P.271	**Potential translation of neurofilament light chain (NfL) as a safety biomarker for neurotoxicity in spinal muscular atrophy** D. Theil; H. Jullien de Pommerol ; J. Kuhle; D. Brees; E. Tritto; M. Valentin; A. Hartmann

P.272	**Cardiac troponin T (cTnT) as a highly sensitive parameter for spinal muscular atrophy (SMA) in a floppy infant** T. Birsak ; A. Ille; A. van Egmond-Froehlich; S. Weiss; M. Gosk-Tomek; A. Kellersmann; M. Foedinger; S. Peithner; G. Bernert
P.273	**Serum neurofilament light chain in type 1 spinal muscular atrophy: 30 months data from first part of a branaplam phase II study** T. Peters ; N. Deconinck; M. Valentin; A. Kieloch; D. Theil; H. Jullien de Pommerol
P.274	**Serum neurofilament light chain in type 1 spinal muscular atrophy: Second part of a branaplam phase II study** T. Peters ; S. Jevtic; D. Carr; A. Kieloch; M. Valentin; A. Dobrzycka-Ambrozevicz; K. Kotulska-Jó´zwiak; I. Litvinenko; O. Lvova; T. Pervunina; E. Petryaikina; I. Shishimorov; D. Vlodavets; A. Kakarieka
P.275	**Determination of protein markers in skeletal muscle of SMA type 3 patients** H. Kölbel ; A. Gangfuß; L. Kollipara; R. Horvarth; A. Roos; U. Schara
P.276	**Childhood-onset spinal muscular atrophy or juvenile amyotrophic lateral sclerosis without pontocerebellar hypoplasia caused by a novel VRK1 splice variant** M. Sedghi; A. Moslemi; M. Olive; M. Etemadifar; B. Ansari; J. Nasiri; I. Emrahi; H. Mianesaz; N. Laing; H. Tajsharghi; H. Tajsharghi
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EP.92	**Contribution of the lower limbs in maintaining sitting balance in individuals with DMD: a pilot study** L. Peeters; M. Gameren; I. de Groot
EP.93	**Electrical impedance myography goes global: collaborative efforts to advance a promising preclinical and clinical tool for the development of future DMD therapies** M. van Putten; P. Mantuano; A. Aartsma-Rus; A. De Luca; M. Leitner; L. Dalle Pazze ; J. Nagy; M. Mortreux; C. Semple; S. Rutkove
EP.94	**Reliability and validity of trunk control measurement scale in Duchenne muscular dystrophy** N. Bulut; B. Yardımcı Lokmanoğlu; G. Aydın; F. Kabakçı; M. Atcı; I. Gürbüz; H. Topaloglu; A. Karaduman; O. Yılmaz
EP.95	**Patient perception of outcome measures for non-ambulant Duchenne muscular dystrophy patients** K. Naarding ; M. Van der Holst; N. Van de Velde; P. Van Schaik-Bank; H. Kan; E. Niks

	E-Posters – Next generation sequencing (EP.96-103) Lumbye Hall
EP.96	**"Double trouble" in a large cohort of patients with unexplained muscle weakness** A. Topf ; K. Johnson; M. Mroczek; L. Phillips; J. Duff; E. Valkanas; E. England; D. MacArthur; V. Straub; MYO-SEQ consortium
EP.97	**Targeted sequencing reveals rare variants in non-coding regions of CAPN3 in Polish limb-girdle muscular dystrophy patients.** A. Macias; J. Fichna; M. Topolewska; M. Redowicz; A. Kaminska; C. Zekanowski; A. Kostera-Pruszczyk
EP.98	**Next-generation sequencing-based molecular diagnosis of neuromuscular patients: results of three years experience** M. Van Ghelue ; T. Fagerheim; G. Hansen; B. Nygård; K. Arntzen; K. Ørstavik; M. Rasmussen; Ø. Nilssen; C. Jonsrud
EP.99	**Genetic diagnosis of congenital neuromuscular disorder patients using next-generation sequencing** S. Jang ; A. Cho; B. Lim; J. Lee; S. Kim; W. Kim; M. Kim; M. Seong; S. Park; K. Kim; J. Chae
EP.100	**Impact of NGS on hyperCKemia diagnosis** P. Marti; N. Muelas; I. Azorin; R. Vilchez; J. Vilchez
EP.101	**Clinical, histopathological and molecular characterization of a cohort of Spanish patients with congenital myopathy** D. Natera de Benito ; C. Jou; C. Jimenez-Mallebrera; C. Ortez; A. Codina; L. Carrera Garcia; J. Exposito Escudero; J. Colomer Oferil; F. Palau; A. Nascimento
EP.102	**Genetic diagnosis in large Japanese cohort using targeted re-sequencing system** M. Okubo ; A. Iida; M. Inoue; Y. Saito; M. Ogasawara; S. Hayashi; S. Noguchi; I. Nishino
EP.103	**Genetic and phenotypic characterisation of inherited myopathies in a tertiary neuromuscular centre** E. Bugiardini ; A. Khan; R. Phadke; D. Lynch; A. Cortese; L. Feng; Q. Gang; A. Pittman; J. Morrow; C. Turner; A. Carr; R. Quinlivan; A. Rossor; J. Holton; M. Parton; J. Blake; M. Reilly; H. Houlden; E. Matthews; M. Hanna

16.00 - 16.30 **Afternoon refreshments, exhibition and poster**s – ■ Andersen Hall ■ Lumbye Hall and ■ Gemyse

Poster Session 4

16.15 - 17.30 Poster Area, locations: Lumbye Hall | Gemyse | Andersen Hall

	SMA treatments (P.348-373) Andersen Hall
P.348	**Intrathecal administration of nusinersen in children: usefulness of neuroimaging in patients with abnormal spinal anatomy** L. Carrera-García ; N. Natera De Benito; J. Muchart; J. Lázaro; J. Expósito; J. Colomer; C. Ortez; A. Nascimento
P.349	**Onasemnogene abeparvovec gene-replacement therapy (GRT) for spinal muscular atrophy type 1 (SMA1): pivotal phase 3 study (STR1VE) update** J. Day; C. Chiriboga; T. Crawford; B. Darras; R. Finkel; A. Connolly; S. Iannaccone; N. Kuntz; L. Pena; M. Schultz; P. Shieh; E. Smith; D. Feltner; F. Ogrinc; H. Ouyang; T. Macek; E. Kernbauer; D. Sproule ; Authorship Truncated; J. Mendell
P.350	**Onasemnogene abeparvovec gene-replacement therapy (GRT) in pre-symptomatic spinal muscular atrophy (SMA)** M. Schultz; K. Swoboda; M. Farrar; H. McMillan; J. Parsons; E. Kernbauer; M. Farrow; F. Ogrinc; S. Kavanagh; D. Feltner; B. McGill; S. Spector; J. L'Italien; D. Sproule ; K. Strauss
P.351	**Gene-replacement therapy (GRT) in spinal muscular atrophy type 1 (SMA1): long-term follow-up from the onasemnogene abeparvovec phase 1/2a clinical trial** J. Mendell; R. Shell; K. Lehman; M. McColly; L. Lowes; L. Alfano; N. Miller; M. Iammarino; K. Church; F. Ogrinc; H. Ouyang; E. Kernbauer; S. Shah; J. L'Italien; D. Sproule ; D. Feltner; S. Al-Zaidy
P.352	**Interim report on the safety and efficacy of longer-term treatment with nusinersen in later-onset spinal muscular atrophy (SMA): results from the SHINE study** J. Kirschner ; B. Darras; M. Farrar; E. Mercuri; C. Chiriboga; N. Kuntz; P. Shieh; M. Tulinius; J. Montes; S. Reyna; G. Gambino; R. Foster; I. Bhan; J. Wong; W. Farwell
P.353	**FIREFISH Part 1: 16-month safety and exploratory outcomes of risdiplam (RG7916) treatment in infants with type 1 spinal muscular atrophy (SMA)** G. Baranello; L. Servais; J. Day; N. Deconinck; E. Mercuri; A. Klein; B. Darras; R. Masson; H. Kletzl; Y. Cleary; M. El-Khairi; T. Seabrook; C. Czech; M. Gerber; C. Nguyen; K. Gelblin; K. Gorni
P.354	**Treatment effects of nusinersen in longstanding adult 5q-SMA type 3 - a prospective observational study over 10 months** M. Walter ; J. Stauber; M. Hiebeler; S. Thiele; E. Greckl; A. Pechmann; J. Kirschner; B. Schoser
P.355	**Investigating an in-home body-weight support harness system to maximize treatment benefit in spinal muscular atrophy** M. Iammarino ; L. Alfano; N. Miller; L. Lowes
P.356	**Nusinersen in infants who initiate treatment in a presymptomatic stage of spinal muscular atrophy: interim results from the phase 2 NURTURE study** M. Ryan ; D. De Vivo; E. Bertini; W. Hwu; T. Crawford; K. Swoboda; R. Finkel; J. Kirschner; N. Kuntz; J. Parsons; R. Butterfield; H. Topaloglu; T. Ben Omran; V. Sansone; Y. Jong; F. Shu; R. Foster; I. Bhan; S. Fradette; W. Farwell
P.357	**Type I spinal muscular atrophy patients treated with AVXS-101 have greater health outcome improvements and lower use of ventilatory support, hospitalization, and associated costs contrasted to those treated with nusinersen** R. Arjunji; R. Dean; I. Jensen; B. Miller; M. Menier; D. Sproule; D. Feltner; M. Droege; F. Khan; O. Dabbous
P.358	**The value of AVXS-101 gene-replacement therapy (GRT) for spinal muscular atrophy type 1 (SMA1): improved survival, pulmonary and nutritional support, and motor function with decreased hospitalization** O. Dabbous ; D. Sproule; D. Feltner; M. Droege; F. Khan; R. Arjunji
P.359	**The beneficial effect of nusinersen on the breathing pattern of SMA type 1 children** A. LoMauro; R. Masson; C. Mastella; A. Aliverti; G. Baranello
P.360	**Body composition predicts clinical response in SMA patients treated with nusinersen** G. Baranello ; R. De Amicis; C. Mastella; A. Leone; R. Masson; R. Zanin; M. Arnoldi; K. Alberti; A. Foppiani; A. Battezzati; S. Bertoli
P.361	**Language development in spinal muscular atrophy (SMA) type 1 children treated with nusinersen** C. Brusa ; M. Scoto; A. Manzur; M. Main; F. Muntoni; F. Vargha-Khadem; G. Baranello
P.362	**RAINBOWFISH: A study of risdiplam (RG7916) in newborns with pre-symptomatic spinal muscular atrophy (SMA)** E. Bertini; J. Day; M. Muhaizea; H. Xiong; L. Servais; A. Prufer; M. Tichy; W. Yeung; K. Gorni
P.363	**JEWELFISH: Safety and pharmacodynamic data in patients with spinal muscular atrophy (SMA) receiving treatment with risdiplam (RG7916) that have previously been treated with nusinersen** C. Chiriboga; E. Mercuri; D. Fischer; J. Day; D. Kraus; W. Yeung; H. Kletzl; M. Gerber; Y. Cleary; K. Gorni
P.364	**Experiences from a 12 month period of treatment with nusinersen on motor function in Swedish patients with spinal muscular atrophy** L. Wahlgren ; I. Henriksson; L. Alberg; A. Kroksmark; M. Tulinius
P.365	**Clinical outcomes in patients with spinal muscular atrophy type 1, 2 or 3 after 1 year of nusinersen therapy** M. Waldrop ; L. Lowes; J. Toops; L. Alfano; N. Miller; M. Iammarino; K. Kotha; G. Paul; M. Moore-Clingenpeel; C. Tsao; G. Noritz; R. Shell; K. Flanigan
P.366	**Nusinersen improves motor function in ambulatory SMA III patients** T. Birsak ; A. Ille; S. Weiss; A. Kellersmann; M. Gosk-Tomek; A. van Egmond-Froehlich; Y. Lechner; I. Maier; W. Schmidt; R. Bittner; G. Bernert

P.367	**Comparative analysis of single stranded rAAV vectors expressing human SMN1 in correcting spinal muscular atrophy in mice** M. Marinello; J. Denard; V. Latournerie; D. Bonnin; J. Cosette; S. Martin; A. Buj-Bello
P.368	**Spinraza treatment decreases the hospitalization of ventilated SMA patients** G. Acsadi
P.369	**Nusinersen treatment in type 1 spinal muscular atrophy: real-life clinical experience in Portugal** J. Ribeiro ; A. Lomba; F. Palavra; C. Garrido; T. Moreno; M. Santos; J. Vieira; I.Fineza
P.370	**BIO101 demonstrates combined beneficial effects on muscle and motor neurons in a mouse model of severe spinal muscular atrophy** M. Latil ; C. Bézier; S. Cottin; R. Lafont; S. Veillet; P. Dilda; F. Charbonnier; O. Biondi
P.371	**Nusinersen treatment in spinal muscular atrophy: the experience of Bambino Gesù children's hospital** A. D'Amico ; M. Catteruccia; C. Cherchi; M. Chiarini; G. Colia; A. Bonetti; A. Carlesi; M. Rollo; A. Longo; F. Nicita; T. Corsetti; R. Bianchi; R. Cutrera; E. Bertini
P.372	**CSF miRNAs as biomarkers to indicate the clinical response to Spinraza treatment for patients with spinal muscular atrophy** B. Doreste; M. Sanson; I. Zaharieva; M. Scoto; F. Muntoni; H. Zhou
P.373	**Longitudinal changes in electrophysiological findings caused by nusinersen administration in patients with spinal muscular atrophy type 2 or type 3** Y. Arahata ; A. Ishiyama; E. Takeshita; Y. Shimizu-Motohashi; H. Komaki; M. Sasaki
	Myasthenia gravis (P.374-380) — **Lumbye Hall**
P.374	**Surrogacy prevents recurrence of fetal acetylcholine receptor inactivation syndrome** D. Nguyen; S. Botez; C. Nguyen
P.375	**Does rhythmic auditory stimulation influence walking speed in the 6-minute walk test in patients with myasthenia gravis?** L. Andersen ; N. Witting; J. Vissing
P.376	**Effect of low-dose rituximab treatment on T- and B-cell lymphocyte imbalance in refractory myasthenia gravis** S. Jing; J. Lu; J. Song; S. Luo; L. Zhou; C. Quan; J. Xi; C. Zhao
P.377	**Myasthenia gravis anti-MuSK in Argentina: demographic, clinical findings and therapeutic response in a cohort of 56 patients** M. Rugiero ; M. Bettini; V. Alvarez; d. Genco; V. Salutto; C. Mazia; M. Figueredo; H. Gomez; E. Fulgenzi; L. Pirra; M. De Rosa; A. Dubrovsky; R. Reisin; J. Muntadas; M. Nogues
P.378	**A complex movement disorder associated with myasthenic features: a novel phenotype caused by a homozygous NGLY1 mutation** D. Jacquier ; J. Good; B. Laubscher; D. Mercati; E. Roulet-Perez; T. Kuntzer; B. Royer-Bertrand; L. Mittaz-Crettol; H. Fostad; A. Superti-Furga; A. Klein
P.379	**Fetal Acetylcholine Receptor Inactivation Syndrome (FARIS): a potentially treatable autoimmune disorder mimicking a wide range of genetic neuromuscular conditions** M. O'Rahelly; M. Fernandez-Garcia ; A. Hahn; C. Nguyen; D. Kim; S. Byun; H. Koelbel; U. Schara; M. Henrich; J. Leslie; B. Eymard; M. Chouchane; K. Roefke; A. Thieme; P. Van den Bergh; S. Paquay; C. Schneider-Gold; A. Vincent; N. Allen; H. Jungbluth
P.380	**Bi-allelic loss of function mutations in SYT2 cause a congenital onset severe presynaptic myasthenic syndrome** S. Donkervoort ; P. Mohassel; L. Laugwitz; E. Kamsteeg; K. Chao; C. Verschuuren-Bemelmans; V. Horber; J. Fock; N. Voermans; Y. Hu; M. Snyder; S. Iannaccone; H. Lochmüller; T. Haack; A. Foley; R. Horvath; C. Bönnemann
	Disorders of the extracellular matrix (P.381-387) — **Lumbye Hall**
P.381	**Sequencing the fibroblasts Col6A1-3 cDNAs versus gene panel genomic DNA in the diagnostic of COLVI related myopathies** T. Stojkovic; A. de Becdelievre; S. Quijano-Roy; V. Jobic; C. Ledeuil; C. Gartioux; V. Allamand; A. Ferreiro; A. Behin; P. Laforet; B. Eymard; P. Richard; C. Metay
P.382	**Recessive COL12A1 loss of function EDS/myopathy overlap syndrome: confirmation and expansion of a consistently severe phenotype** S. Neuhaus ; C. Konersman; D. Saade; S. Donkervoort; S. Ceulemans; P. Magoulas; A. Skalsky; J. Friedman; D. Malicki; M. Bainbridge; C. Shimul; S. Nahas; D. Dimmock; S. Kingsmore; J. Gleeson; T. Lotze; A. Foley; C. Bönnemann
P.383	**Antisense oligonucleotides therapy for COLVI-related congenital muscular dystrophy** S. Aguti; V. Bolduc; A. Sarathy; Y. Zou; C. Bönnemann; F. Muntoni; H. Zhou
P.384	**Single cell RNA-seq unravels the molecular pathomechanism for collagen VI-related disorders in murine model** S. Noguchi ; M. Ogawa; I. Nishino
P.385	**Automated diagnosis of collagen VI related muscular dystrophies using advanced image analysis and machine learning.** M. Roldán; A. Bazaga; C. Badosa; J. Porta; C. Jimenez-Mallebrera
P.386	**Genome and transcriptome analysis of COLVI genes and characterization of a new promising cellular model** R. Rossi; C. Trabanelli; M. Falzarano; P. Sabatelli; A. Grilli; S. Bicciato; A. D'Amico; A. Ferlini ; F. Gualandi
P.387	**A novel in-situ hybridisation (ISH) assay mapping the in-frame pseudoexon 11 (pE11) expression in cultured dermal fibroblasts (CDF) and skeletal muscle in patients with severe collagen VI disease due to a deep intronic mutation in COL6A1** M. Beck ; S. Aguti; P. Ala; A. Richard-Loendt; D. Chambers; D. Scaglioni; D. Ardicli; L. Feng; R. Mein; H. Zhou; C. Sewry; A. Sarkozy; S. Torelli; F. Muntoni; R. Phadke

	Registries (P.388-399)	Lumbye Hall
P.388	**The RESTORE registry: a resource for measuring and improving spinal muscular atrophy (SMA) outcomes** L. Servais; J. Day; D. De Vivo; J. Kirschner; E. Mercuri; F. Muntoni; P. Shieh; E. Tizzano; M. Droege; O. Dabbous ; F. Khan; F. Anderson; R. Finkel	
P.389	**Global FKRP registry** L. Murphy ; J. Laurent; K. Mathews; J. Stevenson; S. Thiele; J. Vissing; M. Walter; L. Woods; V. Straub	
P.390	**Pulmonary function in patients with Duchenne muscular dystrophy from the STRIDE registry and the CINRG natural history study: a matched cohort analysis** M. Tulinius ; F. Buccella; I. Desguerre; J. Kirschner; E. Mercuri; F. Muntoni; A. Nascimento Osorio; A. Delage; J. Zhu; A. Kristensen; P. Trifillis; C. Santos; C. McDonald	
P.391	**The role of registry in care and treatment of rare disorders: Polish registry of SMA patients** A. Lusakowska ; A. Kaminska; P. Dziala; K. Janiszewska; P. Grochowski; A. Kostera-Pruszczyk	
P.392	**SMArtCARE - Real-world-data collection of patients with spinal muscular atrophy** A. Pechmann ; G. Bernert; U. Schara; I. Schwersenz; M. Walter; H. Lochmüller; J. Kirschner	
P.393	**New advances in the neuromuscular diseases registry in Russia** D. Vlodavets ; A. Monakhova; O. Shidlovskaya; I. Shulyakova; S. Artemieva; E. Melnik; E. Litvinova; T. Okhapkina; T. Egorova; O. Germanenko; D. Reshetov; E. Belousova	
P.394	**Coalition to cure calpain 3: a patient organization committed to treating and ultimately curing limb girdle muscular dystrophy type 2A** J. Levy ; J. Boslego; M. Wrubel; L. Wrubel; M. Spencer	
P.395	**Spanish Pompe registry: baseline characteristics of first 49 patients with adult onset of Pompe disease** J. Alonso-Perez ; S. Segovia-Simon; C. Dominguez-Gonzalez; M. Olive-Plana; D. Mendoza-Grimon; R. Fernandez-Torron; A. Lopez-Munain; J. Muñoz-Blanco; A. Ramos-Fransi; M. Almendrote; I. Illa-Sendra; J. Diaz-Manera	
P.396	**The DM-Scope registry: an innovative framework for myotonic dystrophy translational resarch** M. De Antonio; D. Hamroun; M. Gyenge; B. Eymard; J. Puymirat; C. Gagnon; F. Myotonic Dystrophy Study group; G. Bassez	
P.397	**Adult NorthStar clinical network collaboration to improve standards of care in adults with Duchenne muscular dystrophy living in the UK** A. Pietrusz ; M. Desikan; C. Marini-Bettolo; R. Quinlivan	
P.398	**Usefullness of R-Pact scale for the follow-up of patients with late-onset Pompe disease: results from the French Pompe registry** C. Lefeuvre ; F. Bouhour; A. Nadaj-Pakleza; S. Sacconi; E. Salort-Campana; G. Sole; C. Tard; M. De Antonio; N. Taouagh; D. Hamroun; P. Laforet; a. French pompe study group	
P.399	**The international spinal muscular atrophy (SMA) registry: longitudinal collection and refinement of outcome measures for spinal muscular atrophy** M. Scoto ; F. Muntoni; S. Hall; S. Eaton; A. Rashid; J. Avendano; S. Samsuddin; J. Balashkina; R. Finkel; E. Mercuri	
	E-Posters – Neuropathies / ALS – case reports (EP.104-119)	**Andersen Hall**
EP.104	**Spinal muscular atrophy overlapping syndrome – "double trouble" or a chance for better outcome?** A. Fraczek; A. Potulska-Chromik; M. Bednarska-Makaruk; A. Sulek; E. Obersztyn; N. Braun-Walicka; B. Ryniewicz; A. Kostera-Pruszczyk	
EP.105	**Unilateral calf hypertrophy due to focal myositis induced by S1 radiculopathy** O. Ekmekci ; T. Duksal; M. Baklacı; M. Argın; H. Karasoy	
EP.106	**Hypertrophic neuropathy of the sciatic nerve** U. Yis ; M. Arslan; H. Guleryuz	
EP.107	**Transthyretin amyloidosis with variant Ile93Val mutation** W. Kim ; Y. La; Y. Choi	
EP.108	**Expanding phenotype of EGR2 gene related Charcot-Marie-Tooth disease type 1D** A. Silwal ; A. Manzur; M. Main; R. Harrison; F. Muntoni; A. Sarkozy	
EP.109	**Establishing a new psychology service for people living with amyotrophic lateral sclerosis** A. Merrison ; S. Brady; H. Madden; J. Ashworth	
EP.110	**Clinical and neurophysiological outcome of a patient with predicted type 1 SMA presymptomatically treated with Nusinersen** M. Alvarez Molinero ; D. Gómez-Andrés; M. Gratacós Viñola; E. Toro Tamargo; L. Costa Comellas; L. Gonzalez Gardó; I. Iglesias; A. Macaya; F. Munell	
EP.111	**Identification of novel biallelic mutations in SPTBN4 in a child with NEDHND featuring a spinal muscular atrophy phenotype** C. Castiglioni ; R. Kelly; M. Heather; J. Jofre; B. Suarez; W. Langley; P. Nagy; F. Fattori; E. Bertini	
EP.112	**Congenital sensory neuropathy as a new associated finding in COG6 ang PIGG neurological disorders** L. CostaComellas ; D. Gómez Andrés; M. Alvarez-Molinero; M. Gratacós Viñola; A. Sanchez-Montáñez; S. Ferrer Aparicio; P. Romero Duque; A. Marcè-Grau; A. Macaya; F. Munell	
EP.113	**Infantile-onset CMT2Z is caused by two MORC2 gene mutations and is associated with a distinct phenotype** G. Stettner ; U. Knirsch; W. Berger; U. Graf; B. Hendriks; R. Seidl; G. Bernert; J. Behunova; F. Laccone; S. Weiss	
EP.114	**A CMT family with AD and AR inheritance of a MME variant** G. Ravenscroft; R. Ong; N. Laing; P. Lamont	
EP.115	**DNAH10 : a new candidate gene for ALS** L. Silva ; R. Pamphlett; Y. Bing; D. Christofolini; A. Carvalho	

EP.116	**Genetic neuropathies presenting with CIDP-like features in childhood – 3 novel cases and review of the literature.** M. Fernández-García ; M. Kinali; A. Clarke; P. Fallon; V. Gowda; E. Wraige; H. Jungbluth
EP.117	**Charcot-Marie-Tooth neuropathy, intellectual disability, intractable epilepsy, aggressiveness, and biallelic MCM3AP variants in two sibs** S. Puusepp ; K. Reinson; S. Pajusalu; E. Oiglane-Shlik; P. Ilves; M. Wojcik; K. Ounap
EP.118	**Triple trouble or normal aging?** A. Alonso-Jimenez ; H. Goebel; C. Dittmayer; W. Stenzel
EP.119	**A novel mutation in VAPB causing spinal muscular atrophy, Finkel type in Finland** M. Jokela ; S. Penttilä; S. Huovinen; S. Sandell; O. Kuismin; B. Udd
	E-Posters – Early onset muscle disease – case reports (EP.120-133) **Lumbye Hall**
EP.120	**ADSSL1 distal myopathy presenting with a more rapid progressive course in patient with trisomy 21** L. Medne ; M. Santi; C. Skraban; J. Berkowitz; J. Brandsema; E. Dechene; E. Denenberg; C. Bonnemann; A. Santani; B. Banwell
EP.121	**Carey-Fineman-Ziter syndrome: a MYMK -related myopathy mimicking brainstem dysgenesis** A. Camacho ; B. Menéndez; S. Alvarez; R. Pérez; B. Gil-Fournier; S. Ramiro-León; A. Hernández-Laín; S. Vila; R. Núñez; R. Simón
EP.122	**Recurrent pneumothorax in an adult case of Ulrich congenital muscular dystrophy-case report and review of literature** M. Desikan ; R. Quinlivan; R. Astin
EP.123	**Selenoprotein-related congenital myopathy in two siblings** O. Herguner ; S. Bilge; N. Ozcan; G. Gul Mert; D. Ozcanyuz; F. Incecik; S. Altunbasak
EP.124	**Myopathies presenting with head drop: clinical spectrum and treatment outcomes** R. Alhammad; E. Naddaf
EP.125	**SCN4A mutation causes episodic myotonia and persistent paralysis with internalized blood vessels in skeletal muscle** J. Zheng; Q. Wang; S. Zhang; Y. Zhao; W. Li; C. Yan
EP.126	**Congenital fiber type disproportion related to novel autosomal dominant mutation in TNNT1** F. Fattori ; A. D'Amico; M. Verardo; E. Bellacchio; T. Brizzi; A. Fiumara; C. Rodolico; M. van de Locht; C. Ottenheijm; E. Bertini
EP.127	**Ullrich congenital muscular dystrophy in a boy with 21q22.3 deletion: A revisited diagnosis** P. Simsek Kiper; S. Oguz; G. Utine; M. Kasifoglu; G. Haliloglu
EP.128	**Measuring motor function response to treatment in DOK7 congenital myasthenic syndrome** R. Kennedy ; M. Ryan; K. de Valle; K. Carroll
EP.129	**A probable new pathogenic variant in RYR1 gene? - 3 cases reports** M. Koch; E. Perrone; L. Silva ; A. Carvalho
EP.130	**Congenital myopathy due to TPM2 gene mutation associated to CMT1A in two siblings** M. Bisciglia ; J. Rendu; N. Romero; E. Lacene; G. Brochier; B. Eymard; S. Leonard Louis
EP.131	**Novel ACTA1 mutation causes late-onset nemaline myopathy with fuzzy-dark cores** M. Garibaldi ; F. Fattori; E. Bucci; G. Merlonghi; L. Fionda; F. Vanoli; L. Leonardi; S. Morino; A. Micaloni; S. Raffa; E. Bertini; E. Pennisi; G. Antonini
EP.132	**Central core myopathy in Chinese patients with Nav1.4 p.R675Q mutation** J. Sun; S. Luo ; M. Gao; K. Qiao; H. Lv; C. Zhao
EP.133	**Biallelic mutations in tenascin X, TNXB cause slowly progressive asymmetric myopathy with mild joint dislocations and connective tissue alterations** M. Brisset ; C. Metay; N. Romero; R. Carlier; C. Badosa; C. Marques; C. Jimenez-Mallebrera; P. Laforet; E. Malfatti
	19.00 Networking dinner – Langelinie Pavillonen Buses will depart from Tivoli, H.C. Andersens Boulevard at 17:30 hrs.

Full Programme

Saturday 5th October

		Room: Concert Hall
08.00		**Registration desk opens**
8.30 - 9.00	**I.12**	**Keynote lecture (I.12)** **Ethomic Digital Biomarkers** Aldo Faisal, Imperial College London, UK **Chairpersons: Lindsay Alfano and Thomas Voit**
9.00 - 10.30		**Poster Highlights** **Chairpersons: Kathryn Swoboda and Werner Stenzel**
10.30 - 11.00		**Morning refreshments** – Concert Hall Foyer
11.00 - 12.00		**WMS General Assembly**
12.00 - 13.00		**Lunch** - Concert Hall Foyer
13.00 - 14.00		**Selected oral presentations 7 - Clinical trial highlights (O.38-42)** **Chairpersons: Valeria Ricotti and Haluk Topaloglu**
	O.38	**Systemic gene transfer with rAAVrh74.MHCK7. SGCB increased β-sarcoglycan expression in patients with limb girdle muscular dystrophy type 2E** L. Rodino-Klapac; E. Pozsgai ; S. Lewis; D. Griffin; A. Meadows; K. Lehman; K. Church; N. Miller; M. Iammarino; L. Lowes; J. Mendell
	O.39	**ASPIRO phase 1/2 gene therapy trial in X-linked motubular myopathy (XLMTM): update on preliminary safety and efficacy findings** J. Dowling ; P. Shieh; N. Kuntz; C. Bönnemann; W. Muller-Felber; M. Lawlor; L. Servais; B. Smith; M. Noursalehi; S. Rico; S. Prasad
	O.40	**Intrathecal administration of onasemnogene abeparvovec gene-replacement therapy (GRT) for spinal muscular atrophy type 2 (SMA2): Phase 1/2a study (STRONG)** R. Finkel; J. Day; B. Darras; N. Kuntz; A. Connolly; T. Crawford; R. Butterfield; P. Shieh; G. Tennekoon; S. Iannaccone; F. Ogrinc; S. Kavanagh; E. Kernbauer; J. Whittle; J. L'Italien; B. Kaspar; D. Sproule ; S. Spector; D. Feltner; J. Mendell
	O.41	**Sunfish part 1: 18-month safety and exploratory outcomes of risdiplam (RG7916) treatment in patients with type 2 or 3 spinal muscular atrophy (SMA)** E. Mercuri; G. Baranello; J. Kirschner; L. Servais; N. Goemans; M. Pera; M. Tichy; W. Yeung; H. Kletzl; M. Gerber; C. Czech; M. Annoussamy; Y. Cleary; K. Gorni
	O.42	**Treatment of young boys with Duchenne muscular dystrophy with the NF- κB Inhibitor edasalonexent showed a slowing of disease progression as assessed by MRI and functional measures** R. Finkel ; K. Vandenborne; H. Sweeney; E. Finanger; G. Tennekoon P. Shieh; R. Willcocks; G. Walter; W. Rooney; S. Forbes; W. Triplett; S. Yum; M. Mancini; J. MacDougall; A. Fretzen; P. Bista; A. Nichols; J. Donovan
14.00 - 15.30		**Late breaking session** **Chairpersons: Montse Olive Plana and Bjarne Udd**
15.30 - 16.30		**Prize giving and welcome to the 25th WMS Congress** Handover of the WMS flag and close of congress