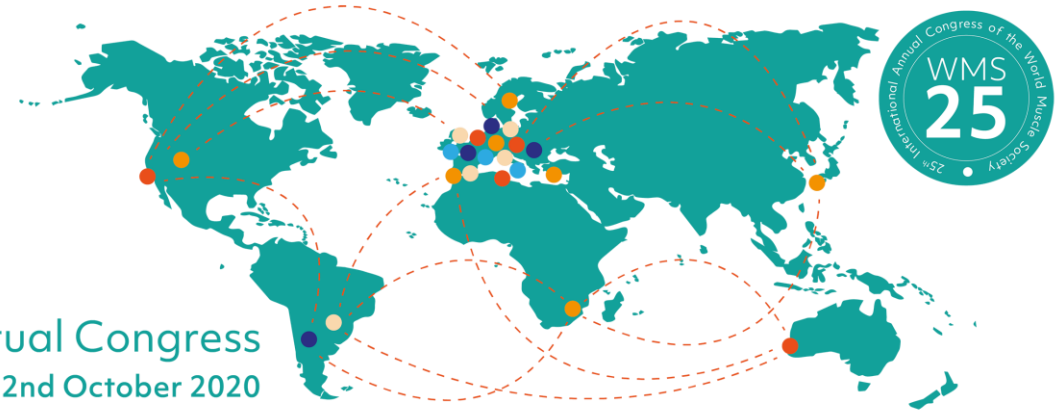

Preliminary Detailed Programme

All times are in British Summer Time (BST)

Monday, 28 September 2020

14:50 – 15:00	**President's Introduction**
15:00 – 16:30	**WMS Industry Symposia 1 and 2** *(2 x 90 minute parallel sessions incorporating live Q&A)*
16:30 – 17:00	**Comfort break**
17:00 – 18:30	**WMS Industry Symposia 3 and 4** *(2 x 90 minute parallel sessions incorporating live Q&A)*
18:30 – 19:00	**Comfort break**
19:00 – 20:30	**WMS Industry Symposia 5 and 6** *(2 x 90 minute parallel sessions incorporating live Q&A)*
	E-Posters available to view on demand

Tuesday, 29 September 2020

15:00 - 22:00	**Pre-Congress Teaching Course (congress registration required)** Congress Industry Symposia and all E-Posters available to view on demand

Wednesday, 30 September 2020

15:00-15:15	**President's Welcome**
15:15-15:45	**Opening lecture** **COVID-19 for NMD, telemedicine, changing practice in the pandemic situation** The COVID-19 pandemic has so far not been the largest, or the most lethal pandemic of the last 100 years, but the global response to it has been unprecedented, both in medical and in societal terms. It has led to a significant change in behaviour, specifically in the way medicine is practised, and its longer- term effects cannot yet be predicted confidently. The concerns for people with neuromuscular disease specifically are significant: in addition to potentially being a risk group for severe infection,

they are put at risk by an interruption to their access to treatment. The World Muscle Society is making an effort to help them face these challenges, by providing advice for patients and carers, trying to define best practice parameters, and suggest avenues to manage the future changes in medical practice. This lecture discusses the approach, its effects on patients, and potential future developments in telemedicine.

Speaker: Max Damian
Session Moderator: Jiri Vajsar

15:45-16:00 **Comfort break**

16:00-16:30 **Panel discussion 1**
Pre-clinical requirements for novel neuromuscular therapies

It is a unique time for neuromuscular disorders, as an unprecedented number of therapeutic candidates are entering clinical trial for a range of diseases. With this surge of potential treatments, the importance of pre-clinical models and rigor in pre-clinical drug development has never been more important. It is critical to evaluate study designs, predictive power, reliability and relevance of current animal models in assessing the efficacy and safety of emerging therapies. In this panel, we have assembled four world leading experts on the application of pre-clinical models to therapy development. We will present key ideas related to best practice and discuss important pitfalls that have plagued pre-clinical drug programs. We will also identify current gaps in the field and offer ideas for future improvements.

Speakers: Annamaria DeLuca, Kanneboyina Nagaraju, Dominic Wells, Jennifer Morgan
Session Moderator: Jim Dowling
Content Moderator: Grace Yoon

16:30-16:45 **Panel discussion 1 – Live Q&A**

16:45-17:15 **Comfort break, poster viewing, sponsors & exhibition, and chat room**

17:15-17:45 **Panel discussion 2**
Gene/ advanced/ new therapeutics

Gene and transcript directed therapies are rapidly becoming a reality and a promise for many neuromuscular disorders. Various such therapies are in the clinic, in clinical trials, or in advanced preclinical development. This panel will discuss opportunities and challenges of these therapies, with a focus on AAV mediated gene therapy. Determinants of success and potential failure will be discussed, touching on issues such as AAV capsids, biodistribution, therapeutic windows of opportunity and durability of expression, seropositivity and redosing, immune mediated toxicity and immunomodulation, as well as pathways and roadblocks for ultrarare applications. This will be on open discussion drawing on recent examples and experiences in the field of clinical neuromuscular gene therapy.

Speakers: Carsten Bönnemann, Francesco Muntoni, Barry Byrne, Ana Buj Bello
Session Moderator: Kathryn Wagner
Content Moderator: Chris Weihl

17:45-18:00 **Panel discussion 2 – Live Q&A**

18:15- 19:15 **WMS 25th Anniversary Quiz**

Thursday, 1 October 2020

15:00-16:00 **New genes in neuromuscular diseases – Selected oral presentations 1 (O.1-4)**
(4 x 10 mins consecutive presentations plus 20 mins live Q&A)

Muscle researchers have identified novel genes and their variants that may be causative to neuromuscular disorders, assess these with transgenic studies, sequencing, and phenotypic and molecular correlation, etc., and discuss future studies and outlook.

Session Moderator: Gisèle Bonne
Assistant Moderator: Hernán Gonorazky

O.01
Mutations in the SMPX gene cause the first X-linked recessive form of distal myopathy
M. Johari; M. Savarese; A. Vihola; M. Jokela; A. Torella; G. Piluso; P. Jonson; H. Luque; A. Magot; F. Magri; C. Kornblum; T. Stojkovic; N. Romero; P. Lahermo; K. Donner; V. Nigro; P. Hackman; B. Udd

O.02
A dominant variant in DNAJB4 causes a myofibrillar myopathy
M. Inoue; A. Iida; K. Watanabe; Y. Hosoi; H. Miyajima; S. Hayashi; Y. Inoue; T. Inoue; S. Noguchi; I. Nishino

O.03
Dominant mutations in Junctin are related to malignant hyperthermia and exertional heat stroke
Y. Endo; L. Groom; N. Kraeva; A. Celic; P. Hopkins; S. Riazi; R. Dirksen; J. Dowling

O.04
Heterozygous frameshift variants in hnRNPA2B1 cause a novel oculopharyngodistal muscular dystrophy
P. Mohassel; S. Donkervoort; H. Kim; A. Foley; X. Lornage; hnRNPA2B1 Study Group; N. Foulds; S. Hammans; T. Haack; J. Bohm; M. Tarnopolsky; V. Straub; J. Laporte; F. Muntoni; J. Taylor; C. Bönnemann

16:00-16:30 **Comfort break, poster viewing, sponsors & exhibition, and chat room**

16:30-17:30 **OMICs and AI approaches for muscle diseases - Selected oral presentations 2 (O.5-8)**
(4 x 10 mins consecutive presentations plus 20 mins live Q&A)

These presentations delve into new genetic, proteomic, and algorithmic approaches to evaluate neuromucsular disorders

Session Moderator: Werner Stenzel
Assistant Moderator: Louise Benarroch

O.05
Whole exome sequencing identifies compound heterozygous missense variants in the LOXL4 gene: a novel candidate cause of contractural myopathy
E. Cohen; I. Nelson; C. Gartioux; M. Beuvin; Z. Mezdari; F. Roth; R. Ben Yaou; S. Quijano-Roy; T. Stojkovic; R. Carlier; G. Bonne; V. Allamand

O.06
Haplotype and variant phasing of large muscle genes with linked-read sequencing
J. Lehtonen; M. Johari; H. Almusa; A. Lehesjoki; K. Wartiovaara; J. Saarela; P. Hackman; C. Wallgren-Pettersson; B. Udd; V. Lehtokari; K. Pelin; M. Savarese

O.07
Proteomic profiling in patients with Dermatomyositis
C. Preusse; A. Ross; D. Hathazi; A. Hentschel; H. Goebel; W. Stenzel

O.08
Deep convolutional-neural-network can differentiate twelve major muscular diseases better than human
Y. Kabeya; M. Okubo; S. Yonezawa; H. Nakano; M. Inoue; M. Ogasawara; Y. Saito; J. Tanboon; L. Indrawati; T. Kumutpongpanich; Y. Chen; R. Tokumasu; T. Iwamori; A. Takano; I. Nishino

17:30-19:30 **Comfort break, poster viewing and sponsors & exhibition**

17:30-19:30 **Poster session 1**
Autophagic myopathies / Myofibrillar Myopathies / Distal myopathies / Pompe disease (P.1-17)

P.01
Pre-clinical safety and efficacy of AT845, a muscle-directed AAV-based gene replacement therapy for Pompe disease
M. Eggers; C. Vannoy; J. Huang; P. Purushottoman; J. Brassard; J. Gray; M. Lawlor; C. Sadhu; F. Mavilio

P.03
NEO1/NEO-EXT studies: Safety and exploratory efficacy of repeat avalglucosidase alfa dosing after up to 6 years in late-onset Pompe disease (LOPD)
B. Schoser; R. Barohn; B. Byrne; O. Goker-Alpan; P. Kishnani; S. Ladha; P. Laforet; E. Mengel; L. Pena; S. Sacconi; V. Straub; J. Trivedi; P. Van Damme; A. van der Ploeg; J. Vissing; P. Young; K. Haack; I. Ivanina; Y. Wang; M. Dimachkie

P.04
Treatment outcome in classic-infantile Pompe disease: effects of dosing in clinical practice
I. Ditters; H. Huidekoper; M. Kruijshaar; A. Hahn; T. Mongini; F. Labarthe; M. Tardieu; J. van den Hout; A. van der Ploeg

P.05
Infantile onset Pompe disease: 10 years of experience in a pediatric reference centre for neuromuscular diseases.
C. Ortez; L. Carrera; J. Exposito; D. Natera; S. Zambudio; J. Colomer; A. Bazán; A. Pareja; E. Bobadilla; v. Sáez; J. Medina; C. Jou; A. Codina; J. Corbera; D. Yubero; L. Martorell; C. Jimenez - Mallebrera; A. Nascimento

P.06
Long-term effects of enzyme replacement therapy in an elderly cohort of late-onset Pompe disease patients
M. Winkler; C. von Landenberg; K. Kuchenbecker; J. Reimann; C. Kornblum

P.07
Efficacy and safety of avalglucosidase alfa in patients with late-onset Pompe disease: results from the phase 3 COMET trial
P. Clemens; The COMET Study Group

P.09
Chloroquine and hydroxychloroquine myopathy: Clinical spectrum and treatment outcomes
E. Naddaf; P. Pritikanta

P.10
Distal myopathy associated with two novel variants in SPTAN1
A. Meyer; W. Arnold; M. Waldrop; K. Flanigan

P.11
Pain characteristics and involvement of small epidermal nerve fibers in patients with late onset Pompe disease (LOPD)
E. Enax- Krumova; J. Görlach; A. Rosenbohm; K. Claeys; F. Montagnese; I. Schneider; D. Sturm; T. Nicoletto; T. Fangerau; A. Roth; J. Wanschitz; W. Löscher; A. Güttsches; S. Vielhaber; L. Zunk; H. Krämer-Best; B. Schoser; A. Hahn; A. Schänzer

P.12
Mutations in supervillin cause myopathy with myofibrillar disorganization and autophagic vacuoles
C. Hedberg-Oldfors

P.14
Autophagic vacuolar myopathy with AVSF (AVSF myopathy) including Danon disease and XMEA: A new clinical entity
K. Sugie; H. Komaki; T. Kurashige; D. Kaneda; N. Eura; T. Shiota; Y. Nishimori; N. Iguchi; H. Nanaura; T. Kiriyama; E. Mori; I. Nonaka; I. Nishino

P.16
Illuminating the role of ER stress in immune mediated necrotizing myopathy with respect to defective chaperone-assisted selective autophagy
N. Fischer; C. Preusse; Y. Allenbach; O. Benveniste; A. Roos; H. Goebel; W. Stenzel

P.17
Screening for Pompe disease using muscle section in Japan
Y. Saito; K. Nakamura; T. Fukuda; H. Sugie; S. Hayashi; S. Noguchi; I. Nishino

Congenital myopathies 1 – Nemaline *(P.18-30)*

P.18
KBTBD13, NEM6 congenital myopathy: deep muscle phenotyping reveals ring-rods fibers, nuclear clumps, and protein aggregates as new morphologic markers
K. Bouman; B. Küsters; J. De Winter; C. Gllet; E. Van Kleef; L. Eshuis; G. Brochier; A. Madelaine; C. Labasse; C. Boulogne; B. Van Engelen; C. Ottenheijm; M. Olive; N. Romero; N. Voermans; E. Malfatti

P.19
ACTA1-related myopathies: expanding the phenotype to include recognizable asymmetries of muscle involvement
A. Foley; S. Ferranti; D. Saade; P. Mohassel; S. Donkervoort; L. Medne; J. Dastgir; D. Bharucha-Goebel; K. Meilleur; M. Leach; M. Scavina; S. Yum; G. Tennekoon; C. Bönnemann

P.20
Mouse models of nemaline myopathy display structural and functional abnormalities of mitochondria
J. Tinklenberg; R. Slick; J. Sutton; M. Prom; E. Ott; S. Danielson; M. Vanden Avond; M. Beatka; H. Meng; M. Grzybowski; J. Heisner; J. Ross; J. Ochala; K. Nowak; L. Zhang; A. Geurts; D. Stowe; F. Montanaro; M. Lawlor

P.21
Ovine congenital progressive muscular dystrophy (OCPMD) is a model of TNNT1 nemaline myopathy
J. Clayton; E. McNamara; H. Goullee; S. Conijn; K. Muthsam; G. Musk; D. Coote; J. Kijas; A. Testa; R. Taylor; M. O'Hara; D. Groth; C. Ottenheijm; G. Ravenscroft; N. Laing; K. Nowak

P.22
Proteomic profiling in Nemaline myopathy to identify molecular phenotypes that contribute to disease heterogeneity
R. Slick; J. Tinklenberg; H. Meng; M. Beatka; M. Prom; E. Ott; F. Montanaro; L. Zhang; H. Granzier; E. Hardeman; A. Geurts; M. Lawlor

P.23
Nutritional status and functioning of patients with nemaline myopathy and related disorders: A pilot study
V. Lehtokari; M. Tammepuu; M. Similä; S. Strang-Karlsson; S. Hiekkala; C. Wallgren-Pettersson

P.24
Respiratory muscle weakness in nemaline myopathy patients
E. van Kleef; C. Ottenheijm; M. Gaytant; W. de Weerd; B. Vosse; B. van Engelen; N. Voermans; J. Doorduin

P.25
Autosomal recessive congenital myopathy with splicing mutation (c.21522+3A>G) of NEB gene
J. Lee; H. Park; Y. Choi

P.26
A new intronic mutation in nebulin myopathy
N. Chrestian; N. Laflamme; Y. Labrie; N. Rioux; M. Dugas; S. Rivest; B. Lace

P.27
Asymmetric muscle weakness due to ACTA1 mosaic mutations
X. Lornage; H. Amthor; S. Quijano-Roy; R. Carlier; N. Monnier; N. Romero; J. Laporte; J. Bohm

P.28
Distinctive pathological features of inherited and sporadic late-onset nemaline myopathies
S. Nicolau; A. Dasgupta; D. Selcen; A. Engel; J. Doles; M. Milone

P.29
AAV gene therapy for TNNT1-associated Nemaline myopathy
E. D'Ambrosio; H. Gray-Edwards; H Grimason; L. Labdi; K. Mesa; M. Otero; M. Sena-Esteves; M. Otero

P.30
Multi exon skipping as a potential therapy for nemaline myopathy in Zebrafish
S. Viththiyapaskaran

Myasthenia & related disorders *(P.31-39)*

P.31
Identification of new recessive mutations in synaptotagmin-2 responsible for severe and early presynaptic forms of congenital myasthenic syndrome
S. Bauché; A. Sureau; D. Sternberg; J. Rendu; C. Buon; J. Messéant; M. Boëx; D. Furling; J. Fauré; X. Latypova; A. Bernabe Gelot; M. Mayer; F. Laffargue; M. Nougues; B. Fontaine; B. Eymard; A. Isapof; L. Strochlic

P.32
Treatment of metastatic urothelial cancer patients with myasthenia gravis with PD-1 inhibitors
A. Ishii; M. Yokoyama; H. Tsuji; Y. Fujii; A. Tamaoka

P.33
Clinical features and evolution of juvenile myasthenia gravis in an Israeli cohort
A. Kuzminsky; Y. Nevo; S. Aharoni; M. Rabie

P.34
Congenital myasthenic syndrome in two Egyptian siblings due to a novel low-expressor AChR epsilon subunit mutation
M. Gomez Garcia de la Banda; N. Fahmi; D. Sternberg; P. Blondy; S. Quijano-Roy; E. Malfatti

P.35
Improvement of strength and muscle MRI with dichlorphenamide in hypokalemic periodic paralysis myopathy: A case report
P. Mongiovi; S. Meyers; R. Griggs; K. Eichinger; E. Ciafaloni

P.36
Novel SCN4A mutations causing mild myotonic phenotype
J. Palmio; R. Mannikko; B. Udd

P.37
Autophagy affected in patients with hypokalemic periodic paralysis
T. Krag; S. Holm-Yildiz; N. Witting; J. Vissing

P.38
Autosomal recessive inheritance in hypokaliemic periodic paralysis due a homozygous SCN4A gene mutation
G. Remiche; L. Desmyter; M. Grenet; I. Vandernoot; O. Devuyst; G. Smits; V. Potmans

P.39
Free-living physical activity and sedentary behaviour in auto-immune myasthenia gravis: a cross-sectional study
S. Birnbaum; D. Bachasson; T. Sharshar; R. Porcher; J. Hogrel; P. Portero

DMD & BMD – Clinical *(P.40-60)*

P.40
A longitudinal study of respiratory function in adult patients with Becker muscular dystrophy
B. De Wel; S. Willaert; A. Nadaj-Pakleza; D. Testelmans; B. Buyse; K. Claeys

P.41
Development of cardiomyopathy, respiratory insufficiency, and loss of ambulation in Becker muscular dystrophy (BMD) - A systematic literature review
K. Johnston; R. Salhany; E. Ciafaloni; A. Mickle; S. Miller; K. Gooch

P.43
Patient and family centric care for Duchenne and Becker muscular dystrophy: towards the IPU (Integrated Practice Unit) model
B. Wong; T. Seckler; L. Dalla Pazze; R. Brown; L. Rhein; T. Flotte

P.44
Cardio-renal syndrome in Duchenne muscular dystrophy
B. Wong; C. Webb; M. Fahey; T. Meyer; K. Laraja; C. Kashtan; A. Salerno; K. Shellenbarger; N. Gupta

P.45
Prophylactic cardiac medication is associated with delayed left ventricular dysfunction and reduced death in childhood-onset dystrophinopathy
K. Mathews; S. Perlman; K. Conway; E. Ciafaloni; S. Thomas; J. Mann; P. Romitti

P.46
Bone health in Duchenne muscular dystrophy -an audit
N. Rajaraman; D. Parasuraman

P.47
Keeping a healthy weight is challenging in Duchenne muscular dystrophy: large influence of ambulatory level, limited influence of dietary intake
S. Houwen-van Opstal; A. de Baaij-Daalmeyer; D. Bot; M. Jansen; C. Dietvorst; K. Corbet; E. Cup; M. Willemsen; E. Niks; I. Groot

P.48
Facilitators and barriers to wearing hand orthoses by adults with Duchenne muscular dystrophy. A mixed method study design
S. Houwen- van Opstal; Y. van den Elzen; M. Jansen; M. Willemsen; E. Cup; I. Groot

P.49
Demographics of patients with nonsense mutation Duchenne muscular dystrophy receiving ataluren in the STRIDE Registry
F. Muntoni; E. Mercuri; F. Buccella; I. Desguerre; J. Kirschner; A. Nascimento Osorio; M. Tulinius; J. Jiang; A. Kristensen; S. Johnson; R. Able; P. Trifillis; C. Santos

P.50
Diffusion tensor MRI metrics reflect larger fibre sizes and heterogeneity of fibre size in Becker muscular dystrophy patients
D. Cameron; J. Burakiewicz; N. van de Velde; C. Baligand; T. Veeger; M. Hooijmans; J. Verschuuren; E. Niks; H. Kan

P.51
DMD Hub: demonstrating impact for DMD trials
E. Heslop; M. Guglieri; A. Irvin; C. Turner; B. Crow; E. George; A. Johnson; E. Crossley; F. Muntoni; V. Straub

P.52
The natural history of the cardiomyopathy of Duchenne and Becker muscular dystrophy
E. Nealon; B. Beckman; N. Kertesz; L. Cripe

P.53
Duchenne muscular dystrophy: does being a younger affected sibling confer long term improved outcomes through earlier diagnosis and management?
M. Main; J. Pisco-Domingos (Deceased); C. Rye; M. Scoto; F. Muntoni

P.54
Has the time to diagnosis of Duchenne muscular dystrophy improved? Data from the Muscular dystrophy surveillance, tracking, and research network
E. Ciafaloni; O. Fapo; K. Conway; N. Street; P. Romitti; C. Westfield; D. Fox; K. Matthews; J. Mann; S. Thomas; A. Soim; MD STARnet

P.55
Prognostic factors for loss of ability to rise from supine in Duchenne muscular dystrophy (DMD)
N. Goemans; C. McDonald; J. Signorovitch; G. Sajeev; M. Fillbrunn; H. Wong; E. Mercuri; K. Vandenborne; F. Muntoni; S. Ward; Investigators for PRO-DMD-01; The Imaging DMD study; iMDEX; cTAP

P.56
Disease attributes most important from a societal perspective: A case study involving Duchenne muscular dystrophy
S. Szabo; I. Audhya; D. Malone; P. Neumann; D. Feeny; M. Harwood; K. Gooch

P.57
Comparison of North Star Ambulatory Assessment scores in siblings with Duchenne muscular dystrophy
C. Rye; M. Main; J. Domingos; F. Muntoni

P.58
Hypermobility in young boys with Duchenne muscular dystrophy and its effect on attainment of North Star Ambulatory Assessment functional skills
L. Abbott; V. Selby; V. Ayyar Gupta; S. Wadsworth; A. Wolfe; M. Chesshyre; G. Baranello; M. Scoto; A. Manzur; M. Main; F. Muntoni

P.59
Functional progression in young DMD
V. Ayyar Gupta; L. Abbott; M. Chesshyre; M. Main; G. Baranello; M. Scoto; A. Manzur; F. Muntoni; U. Northstar Clinical Network

P.60
Involving families in the design of a weight management program for Duchenne muscular dystrophy- Supporting nutrition and optimising wellbeing program
N. Billich; P. Bray; H. Truby; M. Evans; B. Sowerby; K. de Valle; K. Carroll; D. Villano; M. Ryan; Z. Davidson

Muscle function & homeostasis / Molecular therapeutic approaches *(P.62-77)*

P.62
ASPIRO Gene Therapy Trial In X-Linked Myotubular Myopathy (XLMTM): Update on Safety And Efficacy
C. Bönnemann, P. Shieh, N. Kuntz, W. Müller-Felber, A. Blaschek, A. Reghan Foley, D. N. Saade, M. W. Lawlor, Laurent Servais, Weston Miller, M. Noursalehi, S. Prasad, S. Rico, J. Dowling

P.63
AAV.CAPN3 gene therapy for Limb-girdle muscular dystrophy type 2A (LGMD2A): Phenotypic improvements in the calpain3(CAPN3)-null mouse
B. Ozes; D. Murrey; M. Myers; L. Chen; K. Moss; A. Ridgley; C. Wier; Z. Sahenk

P.66
Cell sorting and characterization of urine-derived stem cells (USCs) using the novel technology Celector
M. Falzarano; N. Spedicato; A. Margutti; R. El Dani; R. Rossi; S. Zia; P. Reschiglian; B. Roda; A. Grilli; S. Bicciato; A. Ferlini

P.67
Skeletal muscle atlas: a tool for the muscle community
E. Lacène; M. Beuvin; T. Evangelista; N. Romero; B. Cadot

P.68
O-GlcNAcylation and its interplay with phosphorylation on desmin behaviour: focus on its partition and its interaction with alphaB-crystallin
C. Claeyssen; B. Bastide; C. Cieniewski-Bernard

P.69
Generation and characterization of Pax7-HA knock-in mice
S. Hayashi; Y. Inoue; H. Kosako; T. Inoue; S. Noguchi; I. Nishino

P.71
Role of caveolae in T-tubule biogenesis
E. Lemerle; S. Vassilopoulos

P.72
Importance of myofibre-type specific differences in disease manifestation for muscular dystrophies: focus on dysferlinopathy
E. Lloyd; G. Pinniger; R. Murphy; M. Watt; M. Grounds

P.73
Depletion of ATP limits membrane excitability of skeletal muscle by increasing both ClC1-open probability and membrane conductance
P. Leermakers; K. Dybdahl; K. Husted; A. Riisager; F. de Paoli; T. Pinos; J. Vissing; T. Krag; T. Pedersen

P.74
Studies on a potential interaction between nebulin alternative isoform S21b and ferritin light chain (FTL)
J. Laitila; M. Hanif; J. Sarparanta; J. Lehtonen; A. Khattab; M. Grönholm; C. Wallgren-Pettersson; K. Pelin

P.75
Studies of YBX3 and its variants in myogenesis
L. Sagath; J. Blondelle; J. Laitila; V. Lehtokari; K. Kiiski; M. Grönholm; C. Wallgren-Pettersson; S. Lange; K. Pelin

P.76
Validation of in vivo and ex vivo readouts in a murine model of hind limb unloading to optimize translational research on skeletal muscle atrophy
P. Mantuano; F. Sanarica; O. Cappellari; B. Boccanegra; N. Tarantino; E. Conte; M. De Bellis; G. Camerino; S. Pierno; A. De Luca

P.77
STIM1 heterozygous mutation pathological mechanism in tubular aggregate myopathy
A. Salvi; M. Bartoli

SMA - Clinical *(P.78-92)*

P.78
Burden of illness of spinal muscular atrophy type 1 (SMA1): an update
M. Droege; O. Dabbous; R. Arjunji; M. Gauthier-Loiselle; M. Cloutier; D. Sproule

P.79
Analysis of Cobb angle and clinical characteristics in children with spinal muscular atrophy who enrolled in CHERISH and SHINE
S. Dunaway Young; J. Montes; A. Glanzman; R. Gee; J. Day; R. Finkel; B. Darras; D. De Vivo; G. Gambino; R. Foster; J. Wong; B. Kandinov; Z. Berger

P.80
Distribution of weight and height in children and adolescents with spinal muscular atrophy: An observational retrospective study in the United States
B. Darras; S. Guye; J. Hoffart; S. Schneider; I. Gravestock; K. Gorni; S. Fuerst-Recktenwald; R. Scalco; R. Finkel; D. De Vivo

P.81
Nusinersen experience in later onset spinal muscular atrophy: A tertiary center
G. Öz Tunçer; F. Mıhçı; M. Dolu; Ü. Akça; S. Aydın; S. Türk; A. Aksoy; H. Taşdemir

P.83
A search for systemic manifestations of spinal muscular atrophy (SMA): A matched-cohort study using insurance claims data in the USA
J. Mouchet; M. Oskoui; S. Lipnick; S. Roumpanis; R. Naik; E. Regulier; R. Scalco; B. Darras

P.84
Magnetic resonance imaging of muscle and motor functional changes in patients with spinal muscular atrophy treated with nusinersen
Y. Shimizu-Motohashi; H. Yajima; A. Ishiyama; E. Takeshita; K. Mizuno; N. Sato; M. Sasaki; H. Komaki

P.85
Evolution at 18 months of SMA type 1 patients treated with nusinersen
S. De Lucia; A. Phelep; A. Seferian; P. Foyer; U. Walther-Louvier; J. Durigneux; C. Cances; J. Ropars; C. Vuillerot; L. Servais; O. Boespflug

P.86
Diminished muscle oxygen Uptake and fatigue during exercise in ambulatory spinal muscular atrophy patients
J. Montes; A. Goodwin; M. McDermott; D. Uher; F. Hernandez; K. Coutts; J. Cocchi; M. Hauschildt; K. Cornett; A. Rao; C. Garber; D. De Vivo

P.87
Shortening treatment initiation timeframe through prenatal/neonatal diagnosis of spinal muscular atrophy
B. Gross; S. Matesanz; E. Kichula

P.88
In-home body-weight support maximizes function after treatment in young children with spinal muscular atrophy
M. Iammarino; B. Powers; N. Miller; K. Shannon; L. Alfano; L. Lowes

P.89
Scoliosis in Spinal muscular atrophy type I in the nusinersen era
F. Al Amrani; R. Amin; J. Chiang; J. Boyd; J. Vajsar; J. Dowling; H. Gonorazky

P.90
Trunk involvement in spinal muscular atrophy type 2 and 3
R. Muni-Lofra; G. Coratti; D. Ramsey; D. Moat; J. Sodhi; M. James; A. D'amico; M. Scoto; M. Pane; E. Bertini; C. Marini-Bettolo; F. Muntoni; E. Mercuri; A. Mayhew

P.91
SMA carrier screening testing by droplet digital PCR reveals occurrence of 5 copy numbers of SMN1, substantiating potential 3+0 silent carrier status
S. Cook; N. Folch; L. Hasadsri; D. Oglesbee

P.92
Comparing motor and respiratory function in SMAType1 treated with Nusinersen using CHOP INTEND(CHOP) & Great Ormond Street Respiratory Score (GSR)
L. Edel; L. Abbott; E. Chan; M. Main; V. Robinson; P. Munot; A. Manzur; D. Ridout; G. Baranello; M. Scoto; F. Muntoni

Late Breaking Posters *- (LBP 1–14)*

LBP 1
From mouse- to patient-relevant models: Using CRISPR activation to upregulate a compensatory disease-modifying gene in LAMA2-CMD
Annie Arockiaraj, Jia Qi Cheng-Zhang, Salah Daghlas, Caleb Kim, Dwi Kemaladewi

LBP 2
COVID-19 Myopathy: Persistence of Viral Particles in the Skeletal Muscle
Jian-Qiang Lu, Katerina Gordon, Dubravka Dodig

LBP 3
Dominant mutations in ITPR3 cause Charcot-Marie-Tooth disease
Julius Rönkkö, Svetlana Molchanova, Anya Revah-Politi, Elaine Pereira, Mari Auranen, Jussi Toppila, Jouni Kvist, Anastasia Ludwig, Julika Neumann, Stephanie Humblet-Baron, Geert Bultynck, Adrian Liston, Anders Patau, Claudio Rivera, Matthew Harms, Henna Tyynismaa, Emil Ylikallio

LBP 4
Novel Epigenetic Small Molecule Approaches and Single-Cell Epigenetic Analysis for DMD
Lisa Maves, Jean-Baptiste Dupont, Shawn Luttrell, Melanie Morris, Arianna Gomez, Thao Pham, David Mack

LBP 5
HOPE-2 one-year results show clinically relevant improvements in upper limb &cardiac function in patients with later stage Duchenne Muscular Dystrophy
Linda Marban, Sigfried Rogy, Craig McDonald, Michelle Eagle, Richard Finkel, Cuixia Tian, Michael Taylor, Joanne Janas, Matthew Harmelink, Arun Varadhachary, Kan Hor, Oscar Henry Mayer, Pat Furlong, HOPE-2 Steering Committee

LBP 6
PRIME editing permits to introduce specific point mutations in the gene coding for dystrophin
Cedric Happi Mbakam, Joel Rousseau, Antoine Guyon, Guillaume Tremblay, Francis-Gabriel Bégin, Jacques P. Tremblay

LBP 7
Characterization of Novel Exon-51 Skipping Oligonucleotides that Safely Rescues Dystrophin Expression in a Severe Model of Duchenne Muscular Dystrophy
Charles ONeill, Maria Hedlund, Sundeep Chandra, M Benjamin Hock, Andrew Melton, Todd Oppeneer, Ganesh Cherala, Mika Aoyagi-Scharber, Ashley Frazer-Abel, Nicole Datson, Judith van Deutekom, Jukka Poulivali, Xiefan Fang, Sylvia Fong, David Jacoby, Shripad Bhagwat

LBP 8
A canine model for understanding the pharmacokinetic/pharmacodynamic relationship of muscle-targeted therapies
Robert W Grange, Eva R Chin, Jordan Klaiman, Darren Hwee, Bradley Morgan, Fady Malik, Martin Childers, SiWei Luo, David Mack

LBP 9
Constitutive Stim1 activation impairs myogenesis in Tubular Aggregate Myopathy: defective late differentiation of patient-derived myoblasts and potential druggable targets
Elena Conte, Giulia Maria Camerino, Alessandra Pannunzio, Mauro Coluccia1, Marina Mora, Lorenzo Maggi, Ornella Cappellari, Paola Imbrici, Annamaria De Luca, Antonella Liantonio

LBP 10
ACTN3 genotype influences skeletal muscle mass regulation and the response to dexamethasone
Jane Seto, Kelly Roeszler, Lyra Meehan, Harrison Wood, Chrystal Tiong, Lucinda Bek, Cheryl Lee, Paul Gregorevic, Peter Houweling, Professor Kathryn North

LBP 11
Congenital Muscular Dystrophy Type 1D (α-Dystroglycanopathy) in a Family of Labrador Retrievers with a Stop Codon Mutation in LARGE1 Gene
G. Diane Shelton, Kate M. Minor, Ling T. Guo, Steven G. Friedenberg, Jonah N. Cullen, Jeffrey M. Hord, David Venzke, Mary E. Anderson, Megan Devereaux, Caryl Handelman, Kevin P. Campbell, James R. Mickelson

LBP 12
Syndecan-4 Modulates Cell Polarity and Migration by Influencing Centrosome Positioning and Intracellular Calcium Distribution
Daniel Becsky, Kitti Szabo, Szuzina Gyulai-Nagy, Tamas Gajdos, Zsuzsa Bartos, Arpad Balind, Laszlo Dux, Peter Horvath, Miklos Erdelyi, Laszlo Homolya, Dr. Aniko Keller-Pinter

LBP 13
rAAVrh.74.MHCK7.micro-dystrophin gene therapy in children with Duchenne muscular dystrophy: A quantitative MR biomarker study
Rebecca J Willcocks, Sean C Forbes, Donovan J Lott, Claudia R Senesac, Alison M Barnard, Kelly J Lehman, Carrie E Nease, Kathleen Church, Zarife Sahenk, H Lee Sweeney, Louise R Rodino-Klapac, Glenn A Walter, Jerry R Mendell, Krista Vandenborne

LBP 14
High-fidelity modelling of skeletal muscle laminopathies using LMNA-mutant human iPS cells and bioengineered muscles for mutation-specific therapy development
Luca Pinton, Heather Steele-Stallard, Daniel Moore, Shilpita Sarcar, Tanel Ozdemir, Sara Martina Maffioletti, Jean-Marie Cuisset, Gisèle Bonne, Peter Steven Zammit, Francesco Saverio Tedesco

Poster session 2
Congenital myopathies 2 *(P.93-104)*

P.93
Novel TTN variants identified by whole exome sequencing in patients with prenatal onset titinopathy
A. Gruber; T. Moscarello; J. Machado; M. Kyriss

P.94
RYR1-related congenital myopathy: disease course in a large paediatric cohort of UK patients
M. Sa; D. Ridout; M. Distefano; M. Fernandez-Garcia; M. Main; R. Mein; R. Phadke; H. Jungbluth; F. Muntoni; A. Sarkozy

P.95
Novel mutation in KY gene causes a novel congenital myopathy with early contractures
H. Durmus; A. Cakar; Y. Parman

P.97
Valproic acid improves survival rate and skeletal muscle phenotype in mtm1 knockout mice
M. Ghahramani Seno; J. Volpatti; N. Sabha; C. Alper; E. Sarikaya; J. Dowling

P.98
A dog model for centronuclear myopathy carrying the most common DNM2 mutation
J. Bohm; I. Barthélémy; S. Blot; L. Tiret; J. Laporte

P.99
Phenotyping by NMR of a novel dog model of centronuclear myopathy
Y. Fromes; I. Barthelemy; L. Tiret; J. Boisserie; J. Le Louër; H. Reyngoudt; B. Marty

P.100
Clinical and genetic characterization of RYR1-related myopathy in a reference neuromuscular center in Chile
J. Jofre; B. Suarez; M. Martinez-Jalilie; M. Diemer; C. Hervias; G. Calcagno; X. Ortega; M. Palomino; X. Villanueva; S. Haro; S. Lillo; T. Vial; F. Fattori; E. Bertini; C. Castiglioni

P.102
miRNAs as biomarkers in myotubular myopathy
N. Maani; N. Sabha; D. Gustafson; A. Ramani; J. Fish; M. Alexander; J. Dowling

P.103
Congenital neuromuscular disease with uniform type 1 fiber due to RYR1 mutation is a de facto core myopathy
M. Ogasawara; M. Ogawa; I. Nonaka; S. Hayashi; S. Noguchi; I. Nishino

P.104
Titin related myopathy with ophthalmoplegia
H. Qashqari; H. Gonorazky; K. Amburgey; G. Yoon; L. Hazrati; J. Dowling

Hereditary neuropathies & ALS *(P.105-117)*

P.105
An unusual clinical presentation of CTBP1-related syndrome associating muscle dystrophy, cerebellar ataxia and paralytic ileus
E. El Houwayek; S. Coppens; A. Topf; J. Duff; J. Kaleeta; H. Kadhim; G. Remiche; V. Straub; N. Deconinck

P.106
Novel PLEKHG5 mutations in a lower motor neuron disease patient
L. Gonzalez-Quereda; P. Gallano; I. Pagola; M. Rodriguez; S. Bernal; L. Torné; I. Jericó
P.107
Review of ophthalmologic findings following intrathecal gene transfer for giant axonal neuropathy
D. Saade; D. Bharucha-Goebel; W. Zein; G. Norato; D. Rybin; K. Cheung; L. Charnas; E. Paredes; S. Inati; A. Foley; S. Gray; C. Bönnemann

P.108
First-in-human intrathecal gene transfer study for Giant Axonal Neuropathy: Three-year interim evaluation of safety and efficacy
D. Bharucha-Goebel; D. Saade; M. Jain; G. Norato; D. Rybin; K. Cheung; M. Waite; E. Paredes; A. Foley; T. Lehky; Y. Hu; R. Calcedo Del Hoyo; J. Chichester; S. Jacobson; A. Nath; L. Charnas; R. Samulski; S. Gray; C. Bönnemann

P.109
Expanding the phenotype of ATP7A-related copper transport diseases: Understanding the pathophysiology of neuropathy and copper metabolism
D. Natera; A. Sola; S. Boronat; P. Rego Sousa; C. Ortez; J. Arsmtrong; C. Jou; L. Carrera; J. Exposito; A. Codina; J. Colomer; C. Jimenez-Mallebrera; J. Hoenicka; F. Palau; A. Nascimento

P.110
Development and characterization of in vitro models to test the efficiency of gene therapy approaches in SOD1-linked Amyotrophic lateral sclerosis
M. Delamare; S. Elouej; A. Bigot; M. Cappella; M. Biferi

P.112
Description of muscular involvement in a NEFL-caused neurological disease
H. Kölbel; A. Henschel; A. Della Marina; A. Abicht; A. Sickmann; J. Weis; U. Schara; A. Roos

P.113
Clinical, genetic and neurophysiological characterization of children affected by hereditary neuropathies with clinical onset before 2 years
E. Bobadilla-Quesada; G. Nolasco; G. Salerno; A. Nascimento; L. Carrera-García; D. Natera-de Benito; J. Exposito; J. Colomer; A. Fronggia; V. Sáez; D. Yubero; L. Martorell; F. Bass; J. Medina; C. Ortez

P.114
DNAJB2 mutation causes dominant neuromyopathy
J. Sarparanta; P. Jonson; A. Vihola; H. Luque; J. Reimann; C. Kornblum; B. Udd

P.115
Analysis of off-target effects of antisense sequence inducing exon skipping in SOD1-linked amyotrophic lateral sclerosis
S. Elouej; M. Delamare; M. Cappella; M. Cohen-Tannoudji; S. Astord; T. Marais; B. Giroux; S. Pezet; M. Biferi

P.116
Determining the utility of the GAITRite walkway to assess foot drop in subjects with CMT1A
B. Powers; L. Alfano; N. Miller; M. Iammarino; K. Shannon; Z. Sahenk; L. Lowes

P.117
The relationship between upper and lower limb function in a cohort of children with Charcot-Marie-Tooth (CMT) disease
E. Milev; M. Laura; S. Rand; M. Walk-Ley; A. Wolfe; C. Rye; M. Reilly; F. Muntoni

DMD - Biomarkers & Outcomes measures *(P.118-134)*

P.118
Elevation of fast, but not slow TnI in plasma from Duchenne and Becker muscular dystrophy patients suggests differential fiber response to disease
K. Koch; B. Barthel; M. DuVall; D. Cox; M. Barbieri; E. Hoffmann; A. Russell

P.119
Identification of disease progression stages in patients with Duchenne muscular dystrophy using administrative claims data in the United States
Y. Zhong; J. Iff; D. Gupta; E. Tuttle; R. Schrader

P.120
Phenotype and genotype characterization as predictors of DMD 45 to 55 multi-exon skipping therapy
E. Gargaun; G. Bassez; K. Wahbi; R. Ben Yaou; M. Guibaud; G. Solé; V. Tiffreau; P. Laforet; M. Parent; M. Husson; A. Urtizberea; B. Eymard; A. Boland; J. Deleuze; D. Salgado; M. Khran; N. Levy; A. Blesius; F. Leturcq; F. Pietri-Rouxel

P.121
Lean body mass is associated with whole body bone mineral density and muscle function in steroid-naïve, ambulatory boys with DMD
L. Ward; S. Jackowski; J. Ma; M. Scharke; V. Konji; J. Jaremko; K. Koujok; M. Matzinger; N. Shenouda; S. Walker; H. McMillan; K. Siminoski; P. Bista; M. Mancini; J. Donovan

P.122
Translational implications of increased S100-beta and Tau5 proteins in dystrophic nerves of mdx mouse and rat models for Duchenne muscular dystrophy
V. Krishnan; L. Thanigaiarasu; M. Overzier; A. Aartsma-Rus; L. Bogdanik; C. Lutz; T. Larcher; C. Le Guiner; M. Grounds

P.123
Novel image analysis to assess molecular functionality of dystrophin in Duchenne muscular dystrophy clinical trials
D. Scaglioni; F. Catapano; M. Ellis; S. Torelli; D. Chambers; L. Feng; S. Husayni; J. Malhotra; S. Harriman; E. Koenig; A. Dugar; D. Steiner; J. Morgan; R. Phadke; F. Muntoni

P.124
mRNA in situ hybridization shows Dystrophin transcript subcellular localization imbalance in DMD cells
M. Falzarano; N. Spedicato; R. Rossi; R. El Dani; A. Margutti; J. Morgan; P. Ala; S. Torelli; F. Montanaro; F. Muntoni; A. Ferlini

P.126
Development of a regulatory-ready clinical trial simulation tool for Duchenne muscular dystrophy
J. Larkindale; S. Kim; K. LIngineni; J. Morales; S. Schmidt; D. Corey; K. Romero

P.127
Identification of biallelic DMD variants during maternal carrier testing for Becker muscular dystrophy
E. Ulm; C. Nagaraj; L. Dyer; K. Sund; C. Tian

P.128
Developing a behavioural assessment tool to monitor compliance with physical assessment in Duchenne muscular dystrophy
M. Main; K. Waters; L. Abbott; J. Longatto; M. Chesshyre; G. Baranello; F. Muntoni

P.129
Grip strength is a reliable outcome measure throughout the life span of patients with Duchenne muscular dystrophy
J. Hogrel; V. Decostre; I. Ledoux; M. de Antonio; E. Niks; I. de Groot; V. Straub; F. Muntoni; V. Ricotti; T. Voit; A. Seferian; T. Gidaro; L. Servais

P.130
Reliability and Construct Validity of the Duchenne video assessment
M. Contesse; A. Sapp; S. Apkon; Linda P. Lowes; L. Dalle Pazze; M. Leffler

P.131
A longitudinal study of creatine-phosphokinase and creatinine levels in Duchenne muscular dystrophy
A. Zygmunt; B. Wong; P. Horn; J. Lambert; J. Bange; I. Rybalsky; W. Chouteau; K. Shellenbarger; C. Tian

P.133
Increased physiological responses to threat in Duchenne muscular dystrophy: A potential CNS outcome measure
K. Maresh; A. Papageorgiou; D. Ridout; N. Harrison; W. Mandy; D. Skuse; F. Muntoni

P.134
The importance of data sharing for Duchenne muscular dystrophy
I. Verhaart; P. 't Hoen; M. Roos; E. Vroom

Limb Girdle Muscular Dystrophies *(P.135-156)*

P.135
The 7-year progression of proximodistal dysferlinopathy in another Acadian patient from the Maritimes
S. Bourque; D. Bourcier; J. Mamelona; J. Urtizberea; B. Brais; N. Crapoulet; A. Marrero

P.136
POMT2 homozygous mutation presenting as intellectual deficiency and isolated elevated CK in siblings
M. Masingue; B. Eymard; F. Leturcq

P.137
Treatment of aged mice and long-term durability of AAV-Mediated gene therapy in two mouse models of Limb girdle muscular dystrophy
E. Pozsgai; D. Griffin; E. Peterson; A. Kempton; O. Rogers; Y. Seo; L. Rodino-Klapac

P.138
Systemic dose-finding study with AAV-Mediated gamma-sarcoglycan gene therapy for treatment of muscle deficits in LGMD2C Mice
Y. Seo; A. Kempton; O. Rogers; S. Baine; S. Lewis; K. Adegboye; A. Haile; D. Griffin; E. Peterson; E. Pozsgai; L. Rodino-Klapac

P.139
Expression-functional correlation and validation of a surrogate marker for DAPC restoration in LGMD2E mouse model
E. Pozsgai; E. Peterson; A. Kempton; O. Rogers; Y. Seo; L. Rodino-Klapac

P.140
Systemic gene transfer with rAAVrh74.MHCK7.SGCB increased Beta-sarcoglycan expression in patients with Limb girdle muscular dystrophy type 2E (LGMD2E)
L. Rodino-Klapac; E. Pozsgai; S. Lewis; D. Griffin; A. Meadows; K. Lehman; K. Church; N. Miller; M. Iammarino; L. Lowes; J. Mendell

P.142
The age at loss of ambulation among patients with limb-girdle muscular dystrophy (LGMD) subtype 2: A systematic review
A. Cheung; I. Audhya; S. Szabo; M. Harwood; K. Gooch

P.143
Measuring motor function in LGMD2E: Determining which functional outcomes are the most responsive to change in a 1-year study
N. Miller; M. Iammarino; L. Alfano; B. Powers; K. Shannon; C. Nease; K. Lehman; J. Mendell; L. Lowes

P.144
Patient reported quality of life in limb girdle muscular dystrophy: A cross-sectional survey
N. Johnson; J. Statland; C. Weihl; K. Bates; A. Amato; P. Kang; L. Lowes; K. Mathews; T. Mozaffar; V. Straub; K. Wagner; C. Heatwole

P.145
Prevalence of CAPN3 mutations in Limb-girdle muscular dystrophy type 2A: from a multi-center analysis in China to a worldwide perspective
H. Zhong; Y. Zheng; P. Lin; Z. Zhao; J. Xi; W. Zhu; M. Yu; W. Zhang; H. Lv; C. Yan; J. Hu; Z. Wang; J. Lu; Y. Yuan; S. Luo

P.146
A multi-center LGMD2B-related DYSF mutation analysis in China
H. Zhong; M. Yu; P. Lin; Z. Zhao; J. Xi; W. Zhu; Y. Zheng; W. Zhang; H. Lv; C. Yan; J. Hu; Z. Wang; J. Lu; Y. Yuan; S. Luo

P.147
A simple and rapid neutrophils immunoassay for dysferlinopathies
D. Cox; D. Cox; M. Henderson; V. Straub; R. Barresi

P.148
Semiquantitative CT assessment of muscle involvement in limb girdle muscular dystrophy 2A and 2B with the wide range of duration
S. Kuru; T. Nakayama; A. Ishiyama

P.149
ANO5-related myopathy and muscle inflammation assessed by biopsy and MRI
S. Holm-Yildiz; N. Witting; K. Kass; T. Khawajazada; T. Krag; J. Vissing

P.150
Four individuals with a homozygous mutation in exon 1f of the PLEC gene and associated myasthenic features.
M. Mroczek; H. Durmus; A. Töpf; Y. Parman; V. Straub

P.153
Assessing muscle growth and function with a soluble activin type II b treatment in a LGMD2D mouse model
S. Engelbeen; A. Alqallaf; A. Palo; A. Aartsma-Rus; K. Patel; M. van Putten

P.154
Characterization of muscle biopsy inflammatory cells and complement C5b-9 in dysferlinopathy and other muscular dystrophies
N. Peyton; S. Moore; K. Jones

P.155
Clinical variability in LGMD2B: searching for modifier genes
L. Souza; J. Gurgel-Giannetti; G. Sampaio; J. Wang; M. Scliar; M. Zatz; M. Vainzof

P.156
Differences in demographic and phenotypic spectrum of genetically solved and unsolved cases in a large cohort of patients with limb girdle weakness
M. Mroczek; A. Töpf; S. Specht; K. Johnson; L. Philips; E. England; K. Chao; D. MacArthur; V. Straub

Muscle Imaging – MRI *(P.157-170)*

P.157
Sources of variability in vastus lateralis fat fraction measurements using MRI in clinical trials for Duchenne muscular dystrophy (DMD) therapy
M. Hammond; F. Roche; J. Mary; M. Berger; F. Vincent; S. Zabbatino; R. Scheyer; M. Peterson; S. Holland

P.158
The impact of muscle MRI in diagnostics of myopathies
D. Urban; M. Mohamed; A. Ludolph; J. Kassubek; A. Rosenbohm

P.159
Correlations between quantitative NMRI biomarkers for disease progression and disease activity in muscle of patients with dysferlinopathy
H. Reyngoudt; F. Smith; E. Caldas de Almeida Araujo; I. Wilson; R. Fernandez-Torron; M. James; U. Moore; B. Marty; L. Rufibach; H. Hilsden; H. Sutherland; J. Hogrel; T. Stojkovic; K. Bushby; V. Straub; P. Carlier; A. Blamire; Jain Foundation COS consortium

P.160
Correlations between 31P NMRS indices and NMRI biomarkers of disease progression in skeletal muscle of patients with dysferlinopathy
H. Reyngoudt; F. Smith; E. Caldas de Almeida Araujo; I. Wilson; R. Fernandez Torron; M. James; U. Moore; B. Marty; L. Rufibach; H. Heather; H. Sutherland; J. Hogrel; T. Stojkovic; K. Bushby; V. Straub; P. Carlier; A. Blamire; Jain Foundation COS consortium

P.161
Development of clinical trial simulation tools for Duchenne muscular dystrophy using magnetic resonance biomarkers
K. Vandenborne; S. Kim; R. Willcocks; J. Morales; K. Lingineni; A. Barnard; S. Schmidt; M. Daniels; W. Triplett; J. Larkindale; G. Walter; W. Rooney; D. MR Biomarker Steering Committe

P.162
The use of muscle MRI in the diagnosis of neuromuscular diseases
S. Xirou; C. Lopes; C. Bolano; J. Diaz Manera; V. Straub; C. Marini-Bettolo

P.163
Water T1 by Magnetic Resonance Fingerprinting for quantitative assessment of disease activity in neuromuscular disorders
B. Marty; H. Reyngoudt; E. Araujo; J. Boisserie; Y. Fromes; P. Carlier

P.164
Estimating lean thigh muscle volume using multifrequency serial bioelectrical impedance in patients with muscle atrophy and fatty degeneration
D. Bachasson; A. Carras Ayaz; A. Canal; J. Boisserie; J. Mosso; N. Jean-Amans; P. Carlier; E. Caldas; H. Reyngoudt; B. Marty; O. Benveniste; J. Hogrel

P.165
Quantitative ultrasound of muscle: a study in two different mouse disease models
S. Chrzanowski; A. Pigula Tresansky; J. Nagy; S. Rutkove; B. Anthony

P.166
Muscle MRI as biomarker of disease progression in Becker muscular dystrophy: a 2 year follow-up study
N. van de Velde; M. Hooijmans; Z. Koeks; I. Alleman; T. Veeger; A. Sardjoe-Mishre; E. van Zwet; J. Verschuuren; E. Niks; H. Kan

P.167
Last but not least: preserved thenar muscles in non-ambulant Duchenne muscular dystrophy patients
K. Naarding; K. Keene; A. Sardjoe Mishre; T. Veeger; N. Van de Velde; J. Verschuuren; M. Van der Holst; E. Niks; H. Kan

P.168
Broadening fibroadipose replacement pattern in patients with sarcoglycanopathies: towards whole body MRI approaches
L. Costa Comellas; Á. Sánchez-Montañez; L. Maggi; J. Díaz-Manera; A. Pichiecchio; A. D'Amico; M. Monforte; G. Brisca; N. Løkken; C. Marini-Bettolo; D. Vlodavets; M. C. Walter; V. Straub; S. Quijano-Roy; R. Yves-Carlier; J. Vissing; E. Mercuri; E. Bertini; D. Gómez-Andrés; F. Munell; G. Tasca

P.169
Muscular MRI patterns of limb girdle muscular dystrophies
I. Alawneh; H. Gonorazky

P.170
Association between the progression of muscle fat replacement and muscle architecture in Duchenne muscular dystrophy
T. Veeger; E. van Zwet; D. al Mohamad; K. Naarding; M. Hooijmans; E. Niks; J. de Groot; H. Kan

SMA: Registries, Biomarkers & Outcome measures *(P.171-191)*

P.171
Baseline plasma phosphorylated neurofilament heavy chain level predicts sitting in nusinersen-treated individuals with infantile-onset SMA
F. Muntoni; C. Sumner; T. Crawford; R. Finkel; E. Mercuri; Y. Liu; M. Petrillo; B. Kandinov; W. Farwell

P.172
SMArtCARE: Real-world-data collection of patients with spinal muscular atrophy
A. Pechmann; G. Bernert; T. Hagenacker; W. Müller-Felber; U. Schara; I. Schwersenz; M. Walter; H. Lochmüller; J. Kirschner

P.173
Switching between disease-modifying therapies in patients with spinal muscular atrophy: real-world data collected from the RESTORE Registry
L. Servais; J. Day; D. De Vivo; J. Kirschner; E. Mercuri; F. Muntoni; P. Shieh; E. Tizzano; I. Desguerre; S. Quijano-Roy; K. Saito; M. Droege; O. Dabbous; A. Shah; F. Anderson; R. Finkel

P.174
The Canadian neuromuscular disease registry: A national spinal muscular atrophy (SMA) registry for real world evidence
V. Hodgkinson-Brechenmacher; M. Oskoui; C. Campbell; J. Lounsberry; B. Brais; A. MacKenzie; H. McMillan; J. Vajsar; L. Korngut; C. CNDR Investigator Network

P.175
Emerging real-world use of nusinersen in adult patients with spinal muscular atrophy (SMA) in the US: A multi-site chart review study
A. Paradis; T. Cochrane; A. Khachatryan; M. Martinez

P.176
Longitudinal change over time on Timed Function Tests (TFTs) in ambulatory persons with SMA
K. Krosschell; L. Brown; K. Hoffman; C. Weigel; H. Munson; J. Bidwell; C. DiDonato; N. Kuntz; V. Rao

P.177
Efficacy evaluation of Nusinersen for spinal muscular atrophy type I and type II using bioelectrical impedance (BIA)
K. Ishiguro; T. Sato; M. Shichiji; K. Yuki; T. Murakami; N. Taniguchi; S. Nagata; K. Ishigaki

P.178
Development of motor function and changes in NFL in CSF in children with SMA treated with nusinersen
M. Tulinius; L. Alberg; I. Henriksson; L. Wahlgren; A. Kroksmark; I. Thuestad; K. Jahnke; E. Eklund; K. Blennow; H. Zetterberg

P.180
Identification of functional SMNs and suppressors of SMA causing mutations to define the critical function of SMN that causes SMA
A. Burghes; A. Blatnik III; C. Ruhno; V. McGovern; T. Le; V. Pessino; S. Driscoll; C. Iyer; K. Corlett; S. Likhite; B. Kaspar; S. Pfaff; U. Fischer

P.181
Improvements in the definition of biomarkers for Spinal Muscular Atrophy (SMA) type III and IV: a multimodal longitudinal study
G. Querin; J. Hogrel; R. Debs; V. Marchand-Pauvert; T. Stojkovic; A. Behin; P. Lafôret; F. Salachas; P. Bede; P. Pradat; T. Lenglet

P.182
ActiMyo®: Upper limb activity in non-ambulant patients with spinal muscular atrophy treated with Spinraza
C. Lilien; M. Annoussamy; M. Polleur; A. Seferian; O. Boespflug-Tanguy; K. Gorni; D. Eggenspieler; L. Servais

P.183
Patient reported health-related quality of life in pediatric patients with spinal muscular atrophy type 1, 2 and 3
S. Wagner; B. Wong; J. Lambert; P. Horn; J. Bange; I. Rybalsky; C. Tian

P.184
Disease and treatment burden of spinal muscular atrophy (SMA) on patients and caregivers in Canada
H. McMillan; B. Gerber; T. Cowling; W. Khuu; M. Mayer; J. Wu; B. Maturi; K. Klein-Panneton; C. Cabalteja; H. Lochmuller

P.185
A population-based study examining the epidemiologic burden, healthcare resource utilization and costs of spinal muscular atrophy in Alberta, Canada
G. Chen; B. Sharif; B. Gerber; M. Farris; T. Cowling; C. Cabalteja; J. Wu; B. Maturi; K. Klein-Panneton; J. Mah

P.186
"Registre- SMA France": a national registry of patients with spinal muscular atrophy (SMA)
M. Gomez-Garcia de la Banda; L. Grimaldi; J. Urtizberea; A. Behin; C. Vuillerot; P. Saugier-Veber; F. Audic; C. Barnerias; C. Cances; E. Campana-Salort; C. Spil; P. Laforet; V. Laugel; Y. Pereon; S. Sacconi; T. Stojkovic; C. Tard; B. Chabrol; I. Desguerre; S. Quijano-Roy

P.187
Systematic literature review of the economic burden and economic evaluations in spinal muscular atrophy
T. Dangouloff; L. Servais; M. Hiligsmann

P.188
ATEND: Development of a wheelchair based motor assessment
T. Duong; A. Pasternak; S. Dunaway Young; L. Nelson; R. Muni Lofra; T. Carry; D. Rome-Martin; E. Kichula; E. Maczek; G. Corrati; A. Glanzman

P.189
Diverse cohort of spinraza-treated spinal muscular atrophy patients at Mayo Clinic Rochester for theranostic biomarker discovery
S. Cook; N. Folch; L. Hasadsri; D. Oglesbee; N. Staff; D. Anderson; D. Haile; D. Selcen

P.190
Investigating temporal changes in percent predicted FVC and RULM score in non-ambulant SMA type III children
A. Wolfe; M. Scoto; E. Milev; R. Muni Lofra; A. Rohwer; R. Wake; A. Mayhew; C. Marini-Bettolo; F. Muntoni

P.191
Preliminary data for the cost-effectiveness assessment of the newborn screening for SMA in Belgium.
T. Dangouloff; L. Servais; M. Hiligsmann

Late Submitted Posters *(LSP 1-26*)

LSP 1
Characterization of R-DMDdel52, a preclinical rat model of Duchenne muscular dystrophy
Valentina Taglietti, Kaouthar Kéfi, Mathilde Rodrigues, Baptiste Périou, Bernadette Drayton, Peggy Lafuste, Frédéric Relaix

LSP 2
Persistent COUP-TFII Expression Underlies the Myopathy and Impaired Muscle Regeneration Observed in Resistance to Thyroid Hormone-Alpha
Paola Aguiari, Yan Yun Liu, Astgik Petrosyan, Sheue-yann Cheng, Gregory A Brent, Laura Perin, Anna Milanesi

LSP 3
CRISPR-Cas9 mediated tagging allows the detection of endogenous Gne in mice muscles and muscle cells
Avi Harazi, Nili Ilouz, Lena Yakovlev, Stella Mitrani-Rosenbaum

LSP 4
The complex role of syndecan-4 in skeletal muscle: myogenesis and oncogenesis
Kitti Szabo, Daniel Varga, Attila Gergely Vegh, Ning Liu, Lin Xu, Laszlo Dux, Miklos Erdelyi, Aniko Keller-Pinter

LSP 5
The effect of mutation in EMD gene on DNA-damage response and chromatin organization in cells from EDMD1 patients
Magdalena Machowska, Claudia Bearzi, Katarzyna Piekarowicz, Giovanna Lattanzi, Ryszard Rzepecki

LSP 6
Hierarchical Bayesian model of disease progression in centronuclear myopathy allows to demonstrate treatment efficacy with a small sample size
Arnaud Monseur, Bruno Boulanger, Leen Thielemans, Chris Freitag, Carlin Brad, Seferian M. Andreea, Laurent Servais, Nathis MTM study group

LSP 7
Clinical and genetic aspects of DNM2-related centronuclear myopathy; a retrospective, medical chart review to establish natural history
Asli Aykanat, Stéphanie Hoffmann, Casie Genetti, Khazal Paradis, Heather Paterson, Sundos Al-Husayni, Beiyu Lin, Leen Thielemans, Chris Freitag, Alan Beggs

LSP 8
Nusinersen treatment in SMA patients during COVID-19 pandemic
Karolina Aragon-Gawinska, Anna Potulska-Chromik, Anna Lusakowska, Anna Fraczek, Anna Kostera-Pruszczyk

LSP 10
Israeli SMA Registry 2020: Capturing real-life data
Aviva Fattal-Valevski, Irina E. Opincariu, Anat Weizman, Liora Sagie

LSP 11
Role of Syndecan-4 in myoblast metabolism and mitochondrial functions
Zoltán Márton Köhler, László Juhász, Tamás Gajdos, Miklós Erdélyi, György Trencsényi, László Dux, Anikó Keller-Pintér

LSP 12
SRF-mediated mechanotransduction is required for the response to exercise in both cancer patients and animal models
Medhi Hassani, Alezxandra Baccam, Alexandra Benoni, Caterina Gargano, Gabriela Salim de Castro, Joana Alves, Sara Chiappalupi, Viviana Moresi, Sergio Adamo, Francesca Riuzzi, Guglielmo Sorci, Maurizio Muscaritoli, Marilia Seelaender, Zhigang Xue, Zhenlin Li, Onnik Agbulut, Dario Coletti

LSP 13
Central core disease series disclosing pathogenicity and genetic inheritance of RYR1 variants
Ana Cotta, Lucas Santos Souza, Elmano Carvalho, Leticia Nogueira Feitosa, Antonio Lopes da-Cunha-Junior, Monica Machado Navarro, Julia Filardi Paim, Jaquelin Valicek, Miriam Melo Menezes, Simone Vilela Nunes, Rafael Xavier-Neto, Antonio Pedro Vargas, Reinaldo Issao Takata, Mariz Vainzof

LSP 14
Spinal Muscular Atrophy-III (SMA-III) Assessment in the Context of International Classification of Functioning, Disability and Health (ICF-DH): A Case Series
Kübra Sağır, Kubra Koce, Yunus Emre Tutuneken, Aysegul Asalioglu, Burcu Pamukcu, Gul Deniz Yılmaz Yelvar, Nurgul Elbasi, Yasemin Cirak, Habibe Serap Inal

LSP 15
8 years of experience in diagnosing hereditary neuromuscular disorders in Voronezh (Russia)
Sergei Aleksandrovich Kurbatov

LSP 16
Nusinersen – Significant Results in Early Initiation in Spinal Muscular Atrophy Type II
Evelina Carapancea, Daniela Vasile, Eugenia Roza, Oana Vladacenco, Diana Epure, Mrs Raluca Ioana Teleanu

LSP 17
Lysosomal degradation of GMPPB is associated with limb- girdle muscular dystrophy type 2T
Wotu Tian, Xinghua Luan, Li Cao

LSP 18
Hereditary motor-sensory neuropathy and girdle-limb muscular dystrophy in the Moscow region
Olga Sidorova, Sergey Kotov, Elena Borodataya, Mark Bunak, Alexander Polyakov, Yuriy Filyushkin

LSP 20
Evaluation of Children with Neuromuscular Diseases in terms of Life Quality
Merve Feyza Yüksel, Ömer Bektaş, Süleyman Şahin, Miraç Yıldırım, Serap Teber

LSP 21
Limb-Girdle Muscular Dystrophy (LGMD) Assessment in the Context of International Classification of Functioning Disability and Health (ICF-DH): A Case Series
Kubra Koce, Burcu Pamukcu1, Aysegul Asalioglu, Yunus Emre Tutuneken, Kubra Sagir, Nurgul Elbasi, Gul Deniz Yılmaz Yelvar, Yasemin Cirak, Habibe Serap Inal

LSP 22
Activity and participation assessment in people with muscular dystrophies: a systematic review of validated instruments
Keysy Andrade, Luana Soares, Vinícius Oliveira, Hércules Leite, Thais Gaiad

LSP 23
Congenital disorder of glycosylation type 1T with a novel truncated homozygous mutation in PGM1 gene
Wotu Tian, Xinghua Luan, Li Cao

LSP 24
New phenotype of DCTN1-related spectrum: early-onset dHMN plus congenital foot deformity
Wotu Tian, Xinghua Luan, Li Cao

LSP 25
Role of Stimulated single fiber electromyography (SSFEMG) in early diagnosis of Lambert-Eaton myasthenic syndrome (LEMS)
Ayatallah Farouk Hussein

LSP 26
Emery-Dreifuss muscular dystrophy caused by a mutation in the lamin A/C gene identified by exome sequencing: case report from Indonesia
Kristy Iskandar, Sunartini, Farida Niken, Rizki Amalia, Guritno Adistyawan, Poh San Lai

Poster session 3

Congenital muscular dystrophies *(P.192-203)*

P.192
Gene therapy approach for LAMA2-related muscular dystrophy using linker proteins
J. Reinhard; S. Lin; K. McKee; P. Yurchenco; M. Ruegg

P.193
Collagen VI-related myopathy. Clinical and genetic findings in a large Chilean Cohort
B. Suarez; J. Jofre; M. Martinez-Jalilie; M. Diemer; X. Ortega; T. Vial; S. Lillo; M. Haro; G. Calcagno; M. Palomino; C. Hervias; C. Castiglioni

P.194
Diet, motor activity & daily activity limitations in individuals with SELENON (SEPN1) - Related myopathy
J. Prystupa; R. Alvarez; C. Genetti; E. Weller; S. Liu; B. Moghadaszadeh; E. Troiano; A. Beggs

P.195
The efficacy of steroid therapy for patients with Fukuyama congenital muscular dystrophy
T. Murakami; T. Sato; M. Adachi; M. Shichiji; K. Ishiguro; Y. Kihara; S. Nagata; K. Ishigaki

P.196
Effective pseudo-exon skipping of a COL6A1 intronic mutation in cultured muscle interstitial fibroblasts from a novel humanized mouse model
V. Bolduc; F. Guirguis; J. Cheng; L. Garrett; C. Bönnemann

P.197
Increasing allele selectivity of small interfering RNAs to target a dominant-negative glycine substitution causing a collagen VI-related dystrophy
A. Sarathy; V. Bolduc; C. Bönnemann

P.198
TRAPPC11-related muscular dystrophy with hypoglycosylation of alpha-dystroglycan in skeletal muscle and brain
P. Munot; N. McCrea; S. Torelli; A. Manzur; C. Sewry; D. Chambers; L. Feng; P. Ala; I. Zaharieva; N. Ragge; H. Roper; T. Marton; P. Cox; M. Milev; M. Sacher; W. Liang; S. Maruyama; I. Nishino; R. Phadke; F. Muntoni

P.201
Clinical and molecular spectrum associated with COL6A3 c.7447A>G variant: elucidating its role in CollagenVI-related myopathies
R. Villar Quiles; S. Donkevoort; A. de Becdelievre; V. Allamand; V. Jobic; J. Urtizberea; G. Sole; A. Furby; M. Cerino; E. Campana-Salort; A. Magot; A. Ferreiro; B. Eymard; C. Bönnemann; P. Richard; C. Metay; T. Stojkovic

P.202
Long-term motor function and pulmonary function in COL6-related dystrophies are associated with the maximal motor ability achieved
D. Natera-de Benito; A. Reghan-Foley; C. Domínguez-González; C. Ortez; M. Jain; A. Mebrahtu; S. Donkervoort; Y. Hu; M. Fink; P. Yun; T. Ogata; J. Medina; J. Díaz-Manera; L. Carrera-García; J. Expósito-Escudero; M. Olivé; J. Colomer; C. Jiménez-Mallebrera; C. Bönnemann; A. Nascimento

P.203
Recessive variants in COL25A1 are responsible for arthrogryposis multiplex congenita with an ocular congenital cranial dysinnervation disorder
D. Natera-de Benito; I. Zaharieva; V. Pini; A. Manzur; P. Munot; S. Parker DiTroia; S. Di Gioia; J. Jurgens; B. Barry; E. England; D. Ledoux; A. O´Donell-Luria; D. MacArthur; L. Feng; R. Phadke; A. Sarkozy; E. Engle; F. Muntoni

DMD - Animal models & Preclinical treatment *(P.204-222)*

P.204
Patient-derived induced pluripotent stem cells differentiated cardiomyocytes as platform for disease modelling For X-linked dilated cardiomyopathy
S. Zhu; M. Deng; A. Law; E. Poon; J. Lo; R. Liang; A. Kwong; K. Ng; H. Tse; G. Chan; S. Chan

P.206
Evaluation of the lipid-binding and stability properties of recombinant dystrophin spectrin-like repeat constructs
G. Cooper-Olson; R. Potter; L. Rodino-Klapac

P.207
AAV.U7 technology for two mutational hotspots of the DMD gene (~6%) results in efficient exon skipping, protein restoration and force improvement
N. Wein; T. Simmons; D. Rajakumar; D. Lesman; D. Li; C. Gaffney; R. Rafferty; N. Huang; Y. Rodriguez; C. Young; M. Spencer; K. Flanigan

P.208
Inhibition of NF-kB signaling by edasalonexent prevents the development of DMD-associated cardiomyopathy in mdx:Utrn+/- mice
J. Huang; T. Gemelli; P. Bista; T. Tassin; X. Jiang; J. Shelton; A. Nichols; J. Donovan; P. Mammen

P.211
Src tyrosine kinase as potential target in Duchenne muscular dystrophy: assessment of a novel dasatinib formulation in the mdx mouse model
F. Sanarica; P. Mantuano; B. Boccanegra; O. Cappellari; E. Conte; G. Camerino; A. Cutrignelli; N. Denora; A. Mele; M. De Bellis; A. De Luca

P.212
Systemic delivery of PPMO restores the full-length dystrophin protein in the Dup2 Mouse
L. Gushchina; K. Grounds; E. Frair; N. Huang; F. Schnell; G. Hanson; T. Simmons; N. Wein; K. Flanigan

P.213
Absence of toxicity with intravenous dosing of the Exon 2-skipping AAV9.U7-ACCA vector in non-human primates
L. Gushchina; E. Frair; N. Rohan; A. Bradley; T. Simmons; H. Chavan; M. Waldrop; N. Wein; K. Flanigan

P.214
CRISPR-Cas9 genome editing rescues dystrophin expression in a dog DMD model with a mutation in the N-terminal mutation hotspot
R. Maruyama; K. Lim; Q. Nguyen; M. Tsoumpra; S. Takeda; Y. Aoki; T. Yokota

P.217
Enhancing the rigor and reproducibility of preclinical efficacy drug trials for DMD using reduced number of wild type and mdx control mice
H. Gordish-Dressman; K. Uaesoontrachoon; A. Mackinnon; A. Mullen; E. Hoffman; K. Nagaraju

P.218
A novel in-frame deletion of exons 52-55 DMD mouse model preserves muscle function
M. Kustermann; T. Wong; E. Ivakine; R. Cohn

P.219
Antisense oligonucleotide-mediated knockdown of IGFBP3 to increase IGF-1 signaling in dystrophic muscle
A. Yavas; M. van Putten; E. Niks; A. Aartsma-Rus

P.220
Targeted genome editing in vivo corrects a Dmd duplication restoring wild-type dystrophin expression
E. Maino; D. Wojtal; S. Evagelou; A. Farheen; T. Wong; K. Lindsay; O. Scott; A. Chiodo; M. Schneeweiss; M. Rok; E. Ivakine; R. Cohn

P.221
Edasalonexent maintains bone density and bone strength in the mdx mouse model of Duchenne muscular dystrophy
I. Boraschi-Diaz; D. Rauch; O. Kiraly; P. Bista; A. Nichols; F. Rauch

P.222
Skeletal and cardiac muscle dysfunction in the D2-mdx mouse model of Duchenne muscular dystrophy: a comparison with BL10-mdx mice using TREAT-NMD SOPs
P. Mantuano; F. Sanarica; A. Mele; O. Cappellari; B. Boccanegra; E. Conte; M. De Bellis; G. Camerino; A. De Luca

FSHD / OPMD / Myotonic dystrophy *(P.223-241)*

P.223
Specific biopsy findings: a key factor in the diagnosis of OPMD presenting with isolated dropping head syndrome
M. Masingue; M. Bisciglia; A. Chanut; C. Labasse; C. Metay; T. Evangelista

P.224
A homozygous nonsense variant in LRIF1 associated with facioscapulohumeral muscular dystrophy
D. Sikrova; K. Hamanaka; S. Mitsuhashi; H. Masuda; Y. Sekiguchi; A. Sugiyama; K. Shibuya; R. Lemmers; R. Goossens; M. Ogawa; K. Nagao; C. Obuse; S. Noguchi; Y. Hayashi; S. Kuwabara; J. Balog; I. Nishino; S. van der Maarel

P.225
CRISPR-Cas9 mediated removal of an intronic SMCHD1 mutation supresses DUX4 expression in FSHD myocytes in vitro
R. Goossens; M. van den Boogaard; R. Lemmers; J. Balog; P. van der Vliet; I. Willemsen; J. Schouten; I. Maggio; N. van der Stoep; R. Hoeben; S. Tapscott; N. Geijsen; M. Gonçalves; S. Sacconi; R. Tawil; S. van der Maarel

P.226
A Phase 2, randomized, placebo-controlled, 24-Week study of the efficacy and safety of losmapimod in treating subjects with FSHD: ReDUX4
M. Mellion; R. Tawil; L. Ronco; A. Rahilly; A. Rojas; A. Odueyungbo; K. Wagner; J. Statland; L. Wang; A. Genge; S. Gibson; N. Goyal; J. Hamel; N. Johnson; H. Lochmuller; S. LoRusso; A. Pestronk; S. Sacconi; P. Shieh; D. Cadavid; R. Study Group

P.228
Need for therapy and effect of symptomatic treatment in myotonic disorders: the Myotonia observation survey of patient access to therapy (MyoPath)
A. Zozulya-Weidenfeller; U. Nowak; A. Whiting; A. Kole; P. von Gallwitz

P.229
Open-Label study of losmapimod evaluating safety, tolerability, and changes in biomarker and clinical outcome assessments in subjects with FSHD1
M. Mellion; J. Kools; K. Mul; L. Ronco; A. Odueyungbo; K. Marshall; B. van Engelen; D. Cadavid

P.230
New insights from post-hoc analyses of the OPTIMISTIC trial into the relation of the DM1-Activ-c questionnaire with other commonly used outcome measures
D. van As; K. Okkersen; B. van Engelen; G. Bassez; B. Schoser; G. Gorman; P. 't Hoen

P.231
A biomarker of DUX4 activity to evaluate losmapimod treatment effect in FSHD Phase 2 trials
L. Ronco; K. Wagner; D. Cadavid; A. Chang; M. Mellion; A. Robertson; S. Raines; J. Chadchankar; A. Rojas; P. Shieh; N. Shen; J. Statland; S. Tapscott; R. Tawil; B. van Engelen; L. Wang; N. Johnson; O. Wallace

P.232
Single-cell transcriptomes in facioscapulohumeral muscular dystrophy
L. Hayward; D. Guo; K. Wagner; O. King; C. Emerson. Jr

P.234
Exercise-induced aerobic, functional and cellular adaptations in patients with myotonic dystrophy type 1
A. Mikhail; M. Tarnopolsky

P.235
Development and evaluation of a whole-body MRI imaging protocol and analysis algorithms to measure changes in skeletal muscle in FSHD
M. Mellion; S. Raines; P. Widholm; O. Dahlqvist Leinhard; D. Cadavid

P.236
Spectrum and impact on quality of life of gastrointestinal disorders in myotonic dystrophy type 1
G. Balloy; P. Derkinderen; A. Emonet; Y. Pereon; A. Magot

P.237
Survey of patient care situations for congenital/childhood onset myotonic dystrophy in Japan -- Nationwide patient survey
M. Shichiji; Y. Kihara; K. Ishiguro; T. Sato; M. Takahashi; H. Komaki; H. Takada; S. Kuru; T. Matsumura; K. Ishigaki

P.238
Colorectal cancer occurs as the extramuscular manifestation of elderly FSHD patients
T. Kurashige; H. Morino; H. Ueno; T. Murao; T. Watanabe; T. Hinoi; I. Nishino; H. Maruyama; T. Torii

P.239
Quantitative muscle magnetic resonance imaging in patients with myotonic dystrophy type 1
B. Marty; J. Le Louër; A. Canal; J. Hogrel; M. Gyenge; N. Jebrouni; H. Reyngoudt; G. Bassez

P.240
Targeting unfolded protein response to resolve aggresomes accumulation in oculopharyngeal muscular dystrophy
A. Boulinguiez; F. Roth; J. Dhiab; M. Bui; T. Evangelista; N. Romero; E. Negroni; J. Lacau St Guily; V. Mouly; G. Butler-Browne; C. Trollet

P.241
Genitourinary and lower gastrointestinal signs and symptoms in patients with myotonic dystrophy type 1: a systematic review
I. Fisette-Paulhus; C. Gagnon; M. Morin; L. Girard-Côté

Outcome measures *(P.242-252)*

P.242
Utility of the neuromuscular gross motor outcome to quantify function in neuromuscular disorders
L. Alfano; N. Miller; M. Iammarino; B. Powers; K. Shannon; A. Connolly; M. Waldrop; R. Shell; C. Tsao; K. Flanigan; J. Mendell; L. Lowes

P.243
First step evaluation of a myasthenic syndrome specific patient reported home-based assessment for monitoring physical symptoms
V. Selby; G. Ramdharry; F. Muntoni

P.245
Rasch analysis of the North Star Assessment for limb-girdle-type muscular dystrophies across subtypes
L. Alfano; M. James; N. Miller; R. Muni-Lofra; M. Iammarino; D. Moat; B. Powers; J. Sodhi; M. McCallum; K. Shannon; M. Eagle; A. Mayhew; L. Lowes

P.246
Restorable workspace: A conceptual 3D upper extremity physical function measure for muscle strength and joint contractures
T. Ogata, P. Mohassel; A. Gravunder; M. Jain; T. Bulea; D. Damiano; A. Foley; C. Bönnemann

P.247
Validation and trial studies using the Motor Function Measure (MFM) as an outcome measure: a systematic review
S. Ribault; P. Rippert; D. Vincent Genod; A. Barrière; A. Berruyer; C. Garde; M. Bernard; G. Bertrand; M. Tinat; P. Crépin; M. Naffrechoux; A. Allara; D. Morel; L. Le Goff; C. Vuillerot

P.248
Documenting the psychometric properties of the SARA to advance trial readiness in autosomal recessive spastic ataxia of Charlevoix-Saguenay
D. Bourcier; M. Bélanger; I. Côté; B. Brais; M. Synofzik; J. Brisson; X. Rodrigue; M. Gagnon; J. Mathieu; C. Gagnon

P.249
ActiMyo® from the patients, families and caregivers' perspectives: An international cross-sectional survey on patients with a neuromuscular disease
M. Annoussamy; D. Eggenspieler; P. Furlong; L. Servais

P.250
Relationship between hand strength and function in patients with Duchenne muscular dystrophy or spinal muscular atrophy
V. Decostre; M. de Antonio; L. Servais; J. Hogrel

P.251
The influence of limb girdle muscle weakness on gait parameters: a pilot study across 8 subtypes
B. Powers; L. Alcock; M. Iammarino; M. James; N. Miller; H. Hilsden; K. Shannon; L. Lowes; L. Alfano

P.252
Expansion of normative reference values for the 100 meter timed test for broad use across neuromuscular diseases
M. Iammarino; N. Miller; L. Alfano; B. Powers; K. Shannon; A. Connolly; M. Waldrop; C. Tsao; Z. Sahenk; K. Flanigan; J. Mendell; L. Lowes

SMA – Therapy *(P.253-279)*

P.253
Clinical Development of SRK-015, a Fully Human Anti-proMyostatin Monoclonal Antibody, for the Treatment of Later-Onset Spinal Muscular Atrophy
A. Place; D. Barrett; S. Cote; G. Nomikos; R. Iarrobino; C. Yung

P.254
Nusinersen in adolescents and young adults with SMA: Longitudinal experience from an expanded cohort of CS2/CS12 and SHINE participants
B. Darras; J. Day; K. Swoboda; C. Chiriboga; S. Iannaccone; D. De Vivo; N. Deconinck; R. Finkel; M. Tulinius; K. Saito; J. Montes; R. Foster; D. Ramirez-Schrempp; B. Kandinov; J. Wong; W. Farwell

P.255
One-time administration of AVXS-101 intrathecal (IT) for spinal muscular atrophy in the phase 1 study (STRONG): safety report
D. Chand; R. Finkel; J. Day; B. Darris; N. Kuntz; A. Connolly; C. Zaidman; T. Crawford; R. Butterfield; P. Shieh; G. Tennekoon; J. Brandesma; S. Iannaccone; M. Meriggioli; S. Tauscher-Wisniewski; J. Shoffner; F. Ogrinc; S. Kavanagh; D. Feltner; J. Mendell

P.256
Intravenous (IV) onasemnogene abeparvovec for spinal muscular atrophy (SMA): integrated safety report
D. Chand; R. Finkel; E. Mercuri; R. Masson; J. Parsons; A. Kleyn; M. Menier; K. Montgomery; D. Sproule; S. Reyna; D. Feltner; S. Tauscher-Wisniewki; J. Mendell

P.257
Longer-term treatment with nusinersen: Results in later-onset spinal muscular atrophy from the SHINE study
E. Mercuri; B. Darras; C. Chiriboga; M. Farrar; J. Kirschner; N. Kuntz; G. Acsadi; M. Tulinius; J. Montes; G. Gambino; R. Foster; D. Ramirez-Schrempp; J. Wong; B. Kandinov; W. Farwell

P.258
Onasemnogene aveparvovec gene therapy for spinal muscular atrophy type 1 (SMA1): Phase 3 study update (STR1VE-EU)
E. Mercuri; G. Baranello; R. Masson; O. Boespflug-Tanguy; C. Bruno; S. Corti; A. Daron; N. Deconinck; M. Scoto; L. Servais; V. Straub; F. Ogrinc; H. Ouyang; D. Sproule; S. Reyna; S. Tauscher-Wisniewki; F. Baldinetti; D. Chand; D. Feltner; A. Lavrov; F. Muntoni

P.259
FIREFISH Part 1: 24-month safety and exploratory outcomes of risdiplam (RG7916) in infants with Type 1 spinal muscular atrophy (SMA)
G. Baranello; O. Bloespflug-Tanguy; B. Darras; J. Day; N. Deconinck; A. Klein; R. Masson; E. Mercuri; A. Dodman; M. El-Khairi; M. Gerber; K. Gorni; H. Kletzl; R. Scalco; L. Servais

P.260
Onasemnogene abeparvovec gene therapy in presymptomatic spinal muscular atrophy (SMA): SPR1NT study update
K. Strauss; M. Farrar; K. Swoboda; K. Saito; C. Chiriboga; R. Finkel; S. Iannaccone; J. Krueger; J. Kwon; H. McMillan; L. Servais; J. Mendell; J. Parsons; M. Scoto; P. Shieh; C. Zaidman; M. Schultz; F. Ogrinc; S. Group; F. Muntoni

P.261
Long-term follow-up of onasemnogene abeparvovec gene therapy in spinal muscular atrophy type 1 (SMA1)
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. Joshi; D. Sproule; M. Meriggioli; D. Feltner; S. Al-Zaidy

P.262
Value of onasemnogene abeparvovec in spinal muscular atrophy type 1: improvements in motor function, ventilation-free survival, and hospitalizations
O. Dabbous; S. Reyna; D. Feltner; F. Ogrinc; M. Menier; H. Ouyang; M. Droege; M. Bischof; N. LaMarca; R. Arunji

P.263
SUNFISH Part 1: 24-month safety and exploratory outcomes of risdiplam (RG7916) treatment in patients with Type 2 or 3 spinal muscular atrophy (SMA)
J. Day; G. Baranello; O. Boespflug-Tanguy; S. Borell; N. Goemans; J. Kirschner; R. Masson; M. Pera; L. Servais; S. Fuhrer; M. Gerber; K. Gorni; H. Kletzl; C. Martin; R. Scalco; H. Staunton; W. Yeung; E. Mercuri

P.264
Longer-term effects of nusinersen on motor function outcomes based on age at treatment initiation
R. Finkel; J. Kirschner; E. Mercuri; D. De Vivo; E. Bertini; R. Foster; G. Gambino; D. Ramirez-Schrempp; R. Chin; B. Kandinov; W. Farwell

P.265

Escalating dose and randomized, controlled study of high-dose nusinersen in SMA; study design and updated enrollment for the DEVOTE Study

R. Finkel; J. Day; M. Ryan; E. Mercuri; D. De Vivo; S. Pascual Pascual; J. Montes; J. Gurgel-Giannetti; N. Mitchell-Sweeney; R. Foster; P. Sun; D. Ramirez-Schrempp; B. Kandinov; W. Farwell

P.266

Nusinersen in infantile-onset spinal muscular atrophy: results from longer-term treatment from the open-label SHINE extension study

R. Finkel; D. Castro; M. Farrar; M. Tulinius; K. Krosschell; K. Saito; G. Gambino; R. Foster; D. Ramirez-Schrempp; J. Wong; B. Kandinov; W. Farwell

P.267

Minimally invasive fusionless surgery offers satisfactory early and definitive treatment for scoliosis in SMA children: Our experience over a decade

M. Gaume; E. Saudeau; M. Gomez Garcia de la Banda; V. Azzi-Salameh; C. Barnerias; A. Benezit; I. Dabaj; A. Essid; C. Gitiaux; I. Haegy; B. Mbieleu; R. Sauvagnac; D. Verollet; R. Carlier; J. Bergounioux; I. Desguerre; V. Topouchian; S. Quijano-Roy; L. Miladi; C. Glorion

P.268

Nusinersen effect in infants who initiate treatment in a presymptomatic stage of SMA: NURTURE results

T. Crawford; M. Ryan; J. Kirschner; R. Finkel; K. Swoboda; D. De Vivo; E. Bertini; W. Hwu; R. Foster; D. Ramirez-Schrempp; R. Chin; W. Farwell

P.269

Impact of Continued Nusinersin treatment on Caregiver Experience and Health-Related Quality of Life in Later-onset SMA: Results From the SHINE Study

J. Montes; D. Krasinski; R. Foster; G. Gambino; J. Wong; B. Kandinov; A. Paradis; N. Johnson

P.270

Myostatin dynamics in health and disease: Pharmacologic effects of SRK-015, a highly selective monoclonal antibody inhibitor of myostatin activation

K. Long; S. Cote; S. Wawersik; S. Study Group

P.271

FIREFISH Parts 1 and 2: 12-month pooled safety and efficacy outcomes of risdiplam (RG7916) in infants with Type 1 spinal muscular atrophy (SMA)

L. Servais; O. Bloespflug-Tanguy; B. Darras; J. Day; N. Deconinck; A. Klein; R. Masson; M. Mazurkiewicz-Bełdzińska; E. Mercuri; K. Rose; D. Vlodavets; H. Xiong; E. Zanoteli; A. Dodman; M. El-Khairi; M. Gerber; K. Gorni; H. Kletzl; R. Scalco; G. Baranello

P.273

Jewelfish: Safety and pharmacodynamic data in non-naïve patients with spinal muscular atrophy receiving treatment with risdiplam (RG7916)

C. Chiriboga; C. Bruno; T. Duong; D. Fischer; J. Kirschner; E. Mercuri; M. Gerber; K. Gorni; H. Kletzl; T. McIver; R. Scalco; F. Warren; M. Scoto

P.274

RAINBOWFISH: A study of risdiplam (RG7916) in infants with presymptomatic spinal muscular atrophy (SMA)

L. Servais; E. Bertini; M. Al-Muhaizea; L. Nelson; A. Prufer; D. Vlodavets; Y. Wang; E. Zanoteli; L. Burke; M. El-Khairi; K. Gorni; H. Kletzl; M. Gerber; R. Scalco; R. Finkel

P.276

Monitoring Chilean SMA patients treated with nusinersen. A single-center experience

C. Castiglioni; M. Martinez-Jalilie; M. Diemer; G. Calcagno; C. Hervias; J. Jofre; B. Suarez; M. Palomino; S. Lillo; M. Haro; E. Muñoz; A. Chahin

P.278
Use of motor function measure-20 (MFM-20) to monitor SMA type 1 and 2 patients under nusinersen
L. Le Goff; A. Seferian; A. Phelep; P. Rippert; M. Mathieu; C. Cances; C. de Lattre; J. Durigneux; G. Gousse; S. Quijano-Roy; C. Sarret; L. Servais; C. Vuillerot

P.279
Efficacy and safety of nusinersen treated adult patients with spinal muscular atrophy (SMA) types 2-3-4
B. De Wel; K. Claeys

Poster session 4

DMD - Therapy *(P.280-294)*

P.280
Systemic gene transfer with rAAVrh74.MHCK7.micro-dystrophin in patients with Duchenne muscular dystrophy
J. Mendell; Z. Sahenk; K. Lehman; C. Nease; L. Lowes; N. Miller; M. Iammarino; L. Alfano; J. Vaiea; S. Al-Zaidy; S. Lewis; K. Church; R. Shell; R. Potter; D. Griffin; E. Pozsgai; M. Hogan; L. Rodino-Klapac

P.281
Investigating the effect of DMD non-sequential splicing on exon skipping strategies
R. Goossens; N. Verwey; F. Schnell; A. Aartsma-Rus

P.282
Edasalonexent treatment in young boys with Duchenne muscular dystrophy is associated with age-normative growth and normal adrenal function
E. Finanger; R. Finkel; G. Tennekoon; K. Vandenborne; L. Sweeney; P. Shieh; S. Yum; M. Mancini; J. MacDougall; J. Donovan

P.283
Long-term safety and efficacy of golodirsen in male patients with Duchenne muscular dystrophy amenable to Exon 53 skipping
F. Muntoni; L. Servais; V. Straub; M. Guglieri; A. Dugar; M. Whalen-Kielback; D. Steiner; E. Koenig; T. Feng; X. Wang; E. Mercuri

P.284
ATL1102 treatment improves PUL2.0 in non-ambulant boys with Duchenne muscular dystrophy compared to a natural history control
G. Tachas; N. Desem; P. Button; G. Coratti; M. Pane; E. Mercuri;

P.285
In non-ambulatory DMD patients, one-year treatment benefit of idebenone in respiratory function extrapolates to 3-year delay in start of assisted vent
G. Buyse; T. Voit; C. McDonald; H. Gordish-Dressman; E. Henricson; T. Serjesen; G. Bernert; M. D'Angelo; M. Leinonen

P.286
In the global phase 3 polarisDMD trial for edasalonexent, standardized outcome measure training produces excellent test-retest variability in NSAA
M. Eagle; J. MacDougall; M. Mancini; J. Donovan

P.287
Accelerating the development of personalized DMD gene therapies
M. Rok; E. Ivakine; R. Cohn

P.288
Casimersen treatment in eligible patients with Duchenne muscular dystrophy: Safety, tolerability, and pharmacokinetics over 144 weeks of treatment
N. Kuntz; K. Wagner; L. East; S. Upadhyay; B. Han; E. Koenig; D. Steiner; P. Shieh

P.289
Open-label evaluation of eteplirsen in males with DMD amenable to exon 51 Skipping: PROMOVI
E. Koenig; P. Shieh; H. Abdel-Hamid; A. Connolly; C. McDonald; D. Steiner; J. Malhotra; N. Khan; W. Hu; B. Han; E. Ciafaloni

P.290
Delay in Duchenne muscular dystrophy progression with eteplirsen: attenuation of pulmonary decline and projected freedom from continuous ventilation
J. Iff; C. Gerrits; E. Birk; E. Tuttle; Y. Zheng; E. Henricson; C. McDonald

P.291
Real-world evidence of eteplirsen treatment effects on Duchenne muscular dystrophy related health outcomes using claims data in the United States
J. Iff; E. Tuttle; C. Gerrits; D. Gupta; Y. Zhong

P.292
Ataluren delays loss of ambulation and decline in pulmonary function in patients with nonsense mutation Duchenne muscular dystrophy
C. McDonald; F. Muntoni; M. Rance; J. McIntosh; J. Jiang; A. Kristensen; V. Penematsa; F. Bibbiani; E. Goodwin; H. Gordish-Dressman; L. Morgenroth; R. Able, P. Trifillis; M. Souza; M. Tulinius

P.293
Safety and efficacy of teriparatide treatment for severe osteoporosis in patients with Duchenne muscular dystrophy
N. Nasomyont; C. Keefe; C. Tian; L. Hornung; J. Khoury; J. Tilden; P. Hochwalt; E. Jackson; I. Rybalsky; B. Wong; M. Rutter

P.294
Does steroid therapy influence pulmonary function decline in adults with Duchenne muscular dystrophy after loss of ambulation?
A. Pietrusz; R. Astin; G. Ramdharry; R. Quinlivan; M. Desikan

Autoimmune myopathies *(P.295-312)*

P.295
Analysis of treatment and outcome of 81 patients with idiopathic inflammatory myopathy
N. Eura; A. Yamanaka; T. Shiota; Y. Nishimori; N. Iguchi; M. Ozaki; H. Nanaura; N. Iwasa; T. Kiriyama; T. Izumi; H. Kataoka; K. Sugie

P.296
The ProDERM study: A randomized, double-blinded, placebo controlled trial of IVIG (Octagam 10%) in patients with dermatomyositis
R. Aggarwal; C. Charles-Schoeman; J. Schessl; Z. Bata-Csorgo; M. Dimachkie; Z. Griger; S. Moiseev; C. Oddis; E. Schiopu; J. Vencovsky; I. Beckmann; E. Clodi; T. Levine; ProDERM Investigators

P.297
A case of Juvenile HMGCR antibody myositis presenting as limb girdle
N. Chrestian; N. Rioux; J. Proulx-Gauthier; B. Ellezam; Y. Labrie; S. Rivest; B. Lace

P.298
Anti-HMGCR myopathy overlaps with dermatomyositis: A distinct subtype of IIM
Y. Hou; K. Shao; Y. Yan; T. Dai; W. Li; Y. Zhao; G. Norman; C. Yan

P.299
Distinct disease-activity markers between inflammatory and dystrophic myopathies revealed by the multi-exponential behaviour of water T2-relaxation
E. Caldas de Almeida Araujo; B. Marty; P. Carlier; P. Baudin; H. Reyngoudt

P.300
TSP-1 mediates muscle damage in brachio-cervical inflammatory myopathy and systemic sclerosis
X. Suárez-Calvet; J. Alonso-Pérez; A. Carrasco-Rozas; E. Fernández-Simón; P. Piñol-Jurado; I. Castellvi; C. Zamora; L. Martínez-Martínez; A. Alonso-Jiménez; D. Castillo; E. Gallardo; I. Illa; J. Díaz-Manera

P.301
Optimizing hand-function patient outcome measures for inclusion body myositis
A. Lin; C. Siener; A. Faino; M. Seiffert; C. Weihl; L. Wang

P.302
Autoantibodies targeting membrane repair proteins represent a novel pathologic mechanism in idiopathic inflammatory myopathy
K. McElhanon; N. Young; J. Hampton; B. Paleo; T. Kwiatkowski; E. Beck; A. Capati; K. Jablonski; T. Gurney; M. Lopez; R. Aggarwal; C. Oddis; W. Jarjour; N. Weisleder

P.303
Mitochondrial DNA rearrangements and point mutations in inclusion body myositis
A. Oldfors; C. Hedberg-Oldfors; S. Basu; U. Lindgren; C. Lindberg; E. Larsson; M. Falkenberg

P.305
Clinical onset of sporadic inclusion body myositis after years of immunosuppressive therapy
M. Winkler; C. von Landenberg; C. Kornblum; J. Reimann

P.306
Clinical correlates of KLRG1+ T cells in inclusion body myositis
N. Goyal; S. Greenberg; J. Cauchi; N. Araujo; V. Li; M. Wencel; T. Irani; L. Wang; G. Coulis; A. Villalta; T. Mozaffar

P.308
Characteristics of chest CT-scan patterns of idiopathic inflammatory myopathies: association with myositis-specific autoantibodies
K. Mariampillai; A. Laporte; Y. Allenbach; O. Benveniste; P. Grenier; S. Boussouar

P.309
Immune mediated necrotising myopathy: a treatable condition missed in the Arab and African regions
R. El Sherif; M. Gamal; I. Nishino

P.310
Myopathy in scleroderma/systemic sclerosis patients: From a histopathological point of view
Y. Chen; M. Inoue; M. Ogasawara; Y. Saito; L. Indrawati; J. Tanboon; T. Kumutpongpanich; M. Okubo; W. Yohioka; S. Hayashi; S. Noguchi; I. Nishino

P.311
Myostatin in inflammatory idiopathic myopathies: a marker of activity and muscle recovery?
A. Mahoudeau; S. Maillard; C. Anquetil; N. Tawara; D. Amelin; Y. Allenbach; O. Benveniste

P.312
"Amyopathic" MDA5-positive dermatomyositis with severe lung involvement presents with myositic morphological features
B. Englert; W. Stenzel

Mitochondrial diseases & Metabolic myopathies *(P.313-324)*

P.313
Design of a phase 3 prospective clinical study for the treatment of thymidine kinase 2 deficiency (TK2d)
Q. Abu Ali; C. Domínguez-González; B. Cohen; A. Berardo; R. Haas; C. Konersman; R. McFarland; G. D'Souza; J. Quan; B. Thompson; M. Hirano

P.314
Mitochondrial DNA mutant load in A3243G mutation and clinical correlation
H. Lee; Y. Lee

P.315
Collaborative model for diagnosis and treatment of very rare diseases: experience in Spain with thymidine kinase 2 deficiency
C. Paradas; C. Domínguez-González; M. Madruga-Garrido; M. Hirano; I. Martí; F. Munell; A. Nascimento; M. Olivé; J. Quan; D. Sardina; R. Martí

P.316
MT1621 for thymidine kinase 2 deficiency (TK2d): mechanism of action is via mitochondrial DNA incorporation
L. Tsuruda; D. Kenny; C. Blázquez-Bermejo; A. Karlsson; R. Martí; Y. Cámara

P.317
A novel m.9143T>C variant -- intercurrent severe lactate acidosis and immunodeficiency as an extension of the phenotypic spectrum in MT-ATP6 mutations
D. Urban; L. Scholle; M. Wagner; A. Ludolph; A. Rosenbohm

P.318
RBCK1-related disease: A rare multisystem disorder with polyglucosan storage, autoinflammation, recurrent infections, skeletal, and cardiac myopathy
R. Phadke; C. Hedberg-Oldfors; R. Scalco; D. Lowe; M. Ashworth; M. Novelli; R. Vara; A. Merwick; H. Amer; R. Sofat; M. Sugarman; A. Jovanovic; M. Roberts; V. Nakou; A. King; I. Bodi; H. Jungbluth; A. Oldfors; E. Murphy

P.320
Analysis of morbidity and mortality in untreated patients with thymidine kinase 2 deficiency
C. Domínguez-González; C. Garone; G. D'Souza; B. Thompson; M. Morton; J. Quan; M. Hirano

P.321
Mitochondrial myopathy in X-linked creatine transporter deficiency due to a novel mutation of SLC6A8 gene
M. Bisciglia; A. Busson; M. Marangoni; P. David; I. Vandernoot; A. Michotte; G. Remiche

P.322
Defects in mucolipin-1 (mucolipidosis type IV) cause dystrophic changes in human muscle
A. Zambon; A. Lemaigre; R. Phadke; S. Grunewald; A. Sarkozy; E. Clement; F. Muntoni

P.323
A genetic basis is identified in 74% cases of paediatric hyperCKaemia without weakness
W. Wong; S. Bryen; A. Bournazos; S. Bommireddipall; L. Waddell; M. Menezes; R. Webster; M. Davis; C. Liang; S. Cooper; K. Jones

P.324
Characterization of mitochondrial respiratory chain complexes in reversible infantile respiratory chain deficiency
S. Roos; C. Hedberg-Oldfors; K. Visuttijai; G. Kollberg; C. Lindberg; N. Darin; A. Oldfors

New genes and diseases / NGS & related techniques *(P.326-334)*

P.326
Whole exome sequencing identifies two novel candidate genes and extends the diagnostic spectrum of patients with neuromuscular diseases
R. Rossi; M. Falzarano; M. Pinotti; D. Balestra; M. Neri; F. Fortunato; E. Mercuri; M. Pane; F. Gualandi; R. Selvatici; A. Ferlini

P.327
Whole exome and whole genome sequencing for the genetic diagnosis of dystrophinopathies
R. Selvatici; M. Fang; M. Falzarano; F. Gualandi; S. Delin; S. Bensemmane; A. Shatillo; L. Bello; E. Pegoraro; A. Ferlini

P.328
Neuromuscular disease variant of unknown significance (VUS) resolution using muscle biopsy evaluation: The Iowa experience
K. Jones; S. Moore

P.329
Whole exome sequencing in the pediatric neuromuscular clinic
A. Meyer; C. Cottrell; S. Reshmi; R. Pfau; K. Lee; M. Mathew; D. Corsmeier; V. Jayaraman; A. Dave-Wala; S. Hashimoto; T. Matthews; D. Mouhlas; M. Stein; M. Waldrop; K. Flanigan

P.330
An approach to prioritizing novel disease gene candidates and investigating their role in muscle biology and disease
H. Goullee; J. Clayton; R. Taylor; N. Laing; G. Ravenscroft; A. Forrest

P.331
RNA-Seq is useful in research of neuromuscular disorders
M. Johari; M. Savarese; P. Jonson; S. Koivunen; B. Udd; P. Hackman

P.332
Role of whole exome sequencing in identifying rare genetic variants in a cohort of patients presenting with congenital myopathy
A. Aykanat; C. Genetti; W. Win; Z. Valivullah; E. O'Heir; B. Darras; R. Laine; A. O'Donnell-Luria; A. Beggs

P.333
Progressive proximal muscle weakness, myopathic muscle biopsy and a novel candidate gene
S. Puusepp; T. Reimand; S. Pajusalu; C. Bruels; C. Bönnemann; K. Chao; S. Coppens; S. Donkervoort; J. Goodrich; P. Kang; P. Mohassel; L. Pais; T. Siddique; D. Vargas-Franco; M. Wojcik; W. Stenzel; K. Ounap

P.334
Introme identifies non-canonical splice-altering variants in neuromuscular patients resulting in multiple new genetic diagnoses
P. Sullivan; C. Mayoh; M. Wong-Erasmus; V. Gayevskiy; S. Beecroft; M. Pinese; E. Oates; M. Cowley

Registries, Care, Quality of Life, management of NMD *(P.335-349)*

P.335
Cardiac involvement in Duchenne and Becker muscular dystrophy
G. Öz Tunçer; I. Sahin; Ü. Akça; A. Aksoy

P.336
Novel and more sensitive criteria for identifying chronic respiratory failure in progressive neuromuscular disease
O. Mayer

P.337
Variability of cardiac function in patients with Duchenne muscular dystrophy experiencing sudden onset chest pain and electrocardiographic changes
A. Yamamoto; H. Komaki; K. Segawa; Y. Shimizu-Motohashi; E. Takeshita; A. Ishiyama; N. Sumitomo; E. Nakagawa; M. Sasaki

P.338
The "Hospital goes to School": A psychoeducational program to raise awareness about neuromuscular disorders in schools to prevent social exclusion
I. Zschaeck; A. Colomer; D. Natera De Benito; L. Carrera; J. Exposito; C. Ortez; J. Colomer; A. Nascimento

P.340
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.341
Use of the assessment of caregiver experience with neuromuscular disease (ACEND with SMA) - a caregiver experience from a single center
L. Brown; K. Hoffman; K. Krosschell; C. Weigel; C. Blomgren; H. Munson; J. Bidwell; N. Kuntz; V. Rao

P.342
Life expectancy and causes of death in patients with Duchenne muscular dystrophy
L. Wahlgren; A. Kroksmark; M. Tulinius; K. Sofou

P.344
The rare disease cures accelerator- Data and analytics platform: Value for drug development in muscle diseases
J. Larkindale; V. Boulanger; P. Gavin; R. Liwski; K. Romero; M. Campbell

P.345
Rasch analysis of the pediatric quality of life inventory 4.0 generic core scales administered to patients with Duchenne muscular dystrophy
E. Landfeldt; J. Iff; E. Henricson

P.346
Child-to-adult healthcare transition for inherited muscle diseases: a single-center study in suburbs of Tokyo, Japan
K. Ogata; T. Murakami; K. Yatabe; M. Suzuki; I. Nonaka; T. Tamura

P.347
Illness identity in young adults with neuromuscular disorders
S. Geuens; K. Leyen; J. Willen; V. Maenen; J. Lemiere; N. Goemans; L. De Waele; K. Luyckx

P.348
NMD4C: A new neuromuscular disease network for Canada
K. Amburgey; J. Dowling; J. Warman Chardon; R. Kothary; B. Stead-Coyle; B. Brais; C. Campbell; C. Gagnon; H. McMillan; K. Selby; L. Korngut; M. Oskoui; R. Amin; P. Esler; N. Worsfold; T. Buffone; D. Wojtal; H. Osman; H. Lochmuller

P.349
Subspecialty healthcare utilization in pediatric patients with muscular dystrophy in the United States
S. Matesanz; J. Edelson; K. Iacobellis; E. Mejia; J. Brandsema; C. Wittlieb-Weber; H. Griffis; O. Okunowo; K. Lin

19:30-20:30

From the spinal cord to the muscle - Selected oral presentations 3 (O.9-12)
(4 x 10 mins consecutive presentations plus 20 mins live Q&A)

Molecular and clinical assessments of spinal cord disease, sarcoglycanopathy, and dysferlinopathy are discussed, along with genetic therapy for the latter.

Session Moderator: John Vissing
Assistant Moderator: Mo Zhao

O.09
Spinal cord MRI for early detection of presymptomatic pathology in c9orf72 mutation carriers: A longitudinal neuroimaging study
G. Querin; P. Bede; M. El Mendili; M. Péllegrini-Issac; D. Rinaldi; M. Catala; D. Saracino; F. Salachas; A. Camuzat; V. Marchand-Pauvert; J. Cohen-Adad; O. Colliot; I. Le Ber; P. Pradat; x. PREV-DEMALS Study Group

O.10
European collaboration on the clinical and genetic spectrum of sarcoglycan-deficient muscular dystrophy
J, Alonso-Pérez; L, Gonález-Quereda; C, Semplicini; P, Gallano; E, Pegoraro; A, Nascimento; C, Ortez; M, Devisser; A, Van der Krooi; C, Garrido; M, Santos; M, Guglieri; V, Straub; U, Schara; S, Sarcoglycan Study group; J, Díaz-Manera

O.11
Limb girdle muscular dystrophy R2 and Miyoshi myopathy are not distinct clinical phenotypes in dysferlinopathy
U. Moore; H. Gordish; J. Díaz Maneraz; M. James; A. Mayhew; M. Guglieri; S. Spuler; J. Day; K. Jones; D. Bharucha-Goebel; E. Salort-Campana; A. Pestronk; M. Walter; C. Paradas; T. Stojkovic; M. Yoshimura; E. Bravver; E. Pegoraro; J. Mendell; V. Straub

O.12
Midi-Dysferlin gene therapy for dysferlinopathies
O. Ballouhey; S. Courrier; M. Krahn; N. Levy; M. Bartoli

20:30-21:00

Comfort break, poster viewing, sponsors & exhibiton, and chat room

21:00–22:00

Highlights across Myology - Selected oral presentations 4 (O.13-16)
(4 x 10 mins consecutive presentations plus 20 mins live Q&A)

From basic science to clinical research, investigators use a variety of approaches to assess NM disease, including calcium studies, gene editing, large-scale electron microscopy, and pre-conception carrier screening.

Session Moderator: Mariz Vainzof
Assistant Moderator: Nicolas Chrestian

O.13
Altered calcium handling in a zebrafish model of SELENON congenital muscular dystrophy
M.Wright

O.14
Gene editing using CRISPR/CAS9 in an animal model of nemaline myopathy
N. Sabha; H. Gonorazky; S. Khattak; S. Viththiyapaskaran; N. Bhambra; H. Granzier; J. Dowling

O.15
Large-scale electron microscopy reveals capillary pathology in muscle samples of patients with systemic sclerosis
C. Dittmayer; E. Siegert; A. Uruha; H. Goebel; W. Stenzel

O.16
Results of a preconception carrier screening trial including severe neuromuscular disorders
S. Edwards; R. Ong; M. Davis; R. Allcock; G. Androga; B. Kamien; K. Harrop; G. Ravenscroft; M. Fietz; N. Pachter; J. Beilby; N. Laing

Friday, 2 October 2020

15:00-15:45

Poster highlights (PH1-6)
(6 x 5 mins consecutive presentations plus 15 mins live Q&A)

This is one of the most popular sessions of the annual WMS Congress. The session moderators, with support from the programme committee, select 6 posters for short oral presentations that they feel are of particular interest to the neuromuscular community. The poster may address controversial or polarising topics or topics that aren't regularly covered in plenary lectures, but are nevertheless scientifically sound and relevant.

Session Moderators: Nicol Voermans and Ichizo Nishino

PH 1
Increasing allele selectivity of small interfering RNAs to target a dominant-negative glycine substitution causing a collagen VI-related dystrophy
A. Sarathy; V. Bolduc; C. Bönnemann

PH 2
A homozygous nonsense variant in LRIF1 associated with facioscapulohumeral muscular dystrophy
D. Sikrova; K. Hamanaka; S. Mitsuhashi; H. Masuda; Y. Sekiguchi; A. Sugiyama; K. Shibuya; R. Lemmers; R. Goossens; M. Ogawa; K. Nagao; C. Obuse; S. Noguchi; Y. Hayashi; S. Kuwabara; J. Balog; I. Nishino; S. van der Maarel

PH 3
Introme identifies non-canonical splice-altering variants in neuromuscular patients resulting in multiple new genetic diagnoses
P. Sullivan; C. Mayoh; M. Wong-Erasmus; V. Gayevskiy; S. Beecroft; M. Pinese; E. Oates; M. Cowley

PH 4
COVID-19 Myopathy: Persistence of Viral Particles in the Skeletal Muscle
Jian-Qiang Lu, Katerina Gordon, Dubravka Dodig

PH 5
Illuminating the role of ER stress in immune mediated necrotizing myopathy with respect to defective chaperone-assisted selective autophagy
N. Fischer; C. Preusse; Y. Allenbach; O. Benveniste; A. Roos; H. Goebel; W. Stenzel

PH 6
An approach to prioritizing novel disease gene candidates and investigating their role in muscle biology and disease
H. Goullee; J. Clayton; R. Taylor; N. Laing; G. Ravenscroft; A. Forrest

15:45-16:15

Comfort break, poster viewing, sponsors & exhibition, and chat room

16:15-17:00

Late Breaking News Session (LBO 1-3)
(3 x 10 mins consecutive presentations plus 15 mins live Q&A)

One of the most exciting sessions of the annual WMS Congress, this sessions provides a platform for still unpublished, high quality and high impact research findings that are of great interest to all congress participants. Presentations often focus on new disease causes, pathomechanisms and therapies.

Session Moderators: Laurent Servais and Kevin Flanigan

LBO 1
Pathogenic variants in TNNC2 cause congenital myopathy due to an impaired force response to calcium
Martijn van de Locht, Sandra Donkervoort, Josine de Winter, Stefan Conijn, Benno Kusters, Ying Hu, Reghan Foley, Gwimoon Seo, Darren Hwee, Thomas Irving, Weikang Ma, Henk Granzier, Kalyan Immadisetty, Peter Kekenes-Huskey, José Pinto, Nicol Voermans, Carsten Bönnemann, Coen Ottenheijm

LBO 2

A novel form of muscular dystrophy associated with mutations in JAG2

Sandra Coppens, Sanna Puusepp, Katrin Õunap, Alison Barnard, Sandra Donkervoort, Reza Maroofian, Henry Houlden, Nicolas Deconinck, Fowzan Alkuraya, Hessa Alsaif, Erica Macke, Anna Lusakowska, Andreas Hahn, Volker Straub, Ana Töpf, Catheline Vilain, Marie Rivera-Zengotita, Carsten Bönnemann, Glenn Walter, Peter Kang

LBO 3

Expression of apparent full-length dystrophin in skeletal muscle in a first-in-human gene therapy trial using the scAAV9.U7-ACCA vector

Dr. Megan Waldrop, Dr. Michael Lawlor, Dr. Tatyana Meyers Vetter, Emma Frair, Margaret Beatka, Dr. Hui Meng, Megan Iammarino, Brenna Powers, Johan Harris, Maryann Kaler, Dr. Tabatha Simmons, Dr. Nico Wein, Dr. Kim McBride, Dr. Kevin Flanigan

17:00-17:15 **Comfort break, poster viewing, sponsors & exhibition**

17:15-18:15 **Prize presentations:** *Nicol Voermans*

Close of conference: *Volker Straub*

Introduce 2021 and presentation of flag: *Jana Haberlová*