Preliminary Full Programme

All times are in British Summer Time (BST)

Monday, 20 September 2021 – Industry Day

12:50-13:00	**President´s Introduction**
13:00-23:00	**WMS 2021 Parallel Industry Symposia**
	E-posters available to view on demand

Tuesday, 21 September 2021 – Pre-Congress Teaching Course

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

Wednesday, 22 September 2021 – Congress Day 1

14:00-14:15	**President's Welcome**
14:15-15:00	**Opening lecture - Why AI will be the Foundation of 21st Century Medicine** Topic: The use of machine learning/AI/applying smart tech to the NM field (1 x 30 mins pre-recorded presentation + 15 mins live Q&A) *Speaker: Brendan Frey* *Moderators: Jim Dowling* *Moderator: Kim Amburgey*
15:00-15:30	**Comfort break**
15:30-16:30	**Debate 1 - Pre-clinical research – Are large animal models necessary in translational research?** (60 min = 5 min moderator intro + 2 x 15 mins per person live debate + 25 mins Q&A/interaction with audience) *Speakers: Caroline Le Guiner, Annemieke Aartsma-Rus* *Moderator: Alan Beggs* *Moderator: TBC*
16:30-17:00	**Comfort break**
17:00-18:00	**Debate 2 – Man or machine? Who should evaluate patients in clinical trials?** (60 min = 5 min moderator intro + 2 x 15 mins per person live debate + 25 mins Q&A/interaction with audience)

	Speakers: Lindsay Alfano, Laurent Servais *Moderator: Jean-Yves Hogrel* *Moderator: Damien Bachasson*
18:00-18:30	**Comfort break**
18:30-19:30	**Debate 3 – Should neuromuscular diseases be reduced through pre-conception carrier testing?** (60 min = 5 min moderator intro + 2 x 15 mins per person live debate + 25 mins Q&A/interaction with audience) *Speakers: Yoram Nevo, Kathy Swaboda* *Moderator: Nathalie Goemans* *Moderator: Liesbeth De Waele*
19:30-19:45	**Comfort break**
19:45-20:45	**Social Activity**

Thursday, 23 September 2021 – Congress Day 2

14:00-15:00	**Clinical trial highlights – Selected oral presentations 1 (O.1-4)** (4 x 10 mins consecutive pre-recorded presentations + 20 mins live Q&A) *Moderators: Ulrike Schara-Schmidt & Giovanni Baranello* O.1 **ASPIRO gene therapy trial in X-linked myotubular myopathy (XLMTM): update on preliminary efficacy and safety Ffnding** P. Shieh; N. Kuntz; J. Dowling; W. Müller-Felber; A. Blaschek; C. Bönnemann; R. Foley; D. Saade; A. Sefarian; L. Servais; M. Lawlor; M. Noursalehi; S. Prasad; S. Rico; W. Miller O.2 **IGNITE DMD Phase I/II ascending dose study of SGT-001 microdystrophin gene therapy for DMD: 1.5-year functional outcomes update** V. Rao; B. Byrne; P. Shieh; S. Salabarria; J. Berthy; M. Corti; S. Redican; J. Lawrence; K. Brown; C. Shanks; S. Spector; P. Gonzalez; J. Schneider; C. Morris; C. Clary O.3 **A Phase 2 clinical trial evaluating the safety and efficacy of SRP-9001 for treating patients with Duchenne muscular dystrophy** J. Mendell; P. Shieh; Z. Sahenk; K. Lehman; L. Lowes; N. Reash; M. Iammarino; L. Alfano; B. Powers; J. Woods; C. Skura; H. Mao; L. Staudt; R. Potter; D. Griffin; S. Lewis; L. Hu; S. Upadhyay; T. Singh; L. Rodino-Klapac O.4 **RAINBOWFISH: A study of risdiplam in infants with presymptomatic spinal muscular atrophy (SMA)** L. Servais; M. Al-Muhaizea; M. Farrar; L. Nelson; A. Prufer; R. Finkel; Y. Wang; E. Zanoteli; M. El-Khairi; M. Gerber; K. Gorni; H. Kletzl; L. Palfreeman; R. Scalco; E. Bertini
15:00-15:30	**Breakout networking session – On the topic of selected oral presentations 1**
15:30-16:30	**Clinical research - Selected oral presentations 2 (O.5-8)** (4 x 10 mins consecutive pre-recorded presentations + 20 mins live Q&A) *Moderators: Jordi Dias & TBC*

O.5
A phase 2, randomized, double-blind, placebo-controlled, 48-Week study of the efficacy and safety of losmapimod in subjects with FSHD: ReDUX4
R. Tawil; K. Wagner; Behalf of the ReDUX4 Study Group

O.6
Pre-operative exercise and pyrexia as modifying factors in malignant hyperthermia
S. Riazi; L. van den Bersselaar; G. Islander; L. Heytens; M. Snoeck; A. Bjorksten; R. Gillies; G. Dranitsaris; A. Hellblom; S. Treves; N. Voermans; H. Jungbluth

O.7
Prospective natural history of upper limb disease evolution in Duchenne muscular dystrophy
C. Lilien; H. Reyngoudt; A. Seferian; T. Gidaro; M. Annoussamy; V. Chê; V. Decostre; I. Ledoux; J. Le Louër; E. Guemas; F. Muntoni; J. Hogrel; P. Carlier; L. Servais

O.8
Genotype-phenotype correlations in valosin containing protein disease: an international multicentric audit, the VCP International Study Group
M. Schiava; C. Ikenaga; T. Stojkovic; M. Caballero; I. Nishino; C. Paradas; A. Alonso-Jimenez; A. Kostera-Pruszczyk; F. Miralles Morell; J. De Bleecker; C. Domínguez-Gonzalez; G. Papadimas; K. Claeys; P. Laforet; A. Toscano; E. Pál; M. Farrugia; G. Tasca; C. Weihl; J. Diaz Manera

16:30-17:00 **Breakout networking session – On the topic of selected oral presentations 2**

16:30-17:30 **Poster session**
Autoimmune & Inflammatory NMD (EP.1-18)
EP.01
Autoimmune necrotizing myopathy with anti-signal recognition particle antibodies in the first year of life
A. Camacho; D. Ghandour; J. De Inocencio; A. Hernández Laín; O. Toldos; S. Vila; N. Núñez; R. Simón

EP.02
A window into intracellular events in myositis muscle through targeted proteomics
J. Peterson; R. Zahedi; M. Alamr; V. Leclair; J. DiBattista; K. Nagaraju; M. Hudson

EP.03
Clinicopathological findings of anti-mitochondrial antibody associated myositis
Y. Nishimori; S. Hayashi; S. Noguchi; I. Nishino

EP.04
Exploring the mechanism of myogenic and neurogenic changes on electromyography by quantifying muscle pathology in sporadic inclusion body myositis
N. Eura; T. Mano; A. Yamanaka; Y. Nishimori; T. Shiota; H. Nanaura; K. Sugie

EP.05
Clinicopathological characteristics of anti-TIF1-gamma antibody-positive dermatomyositis
A. Yamanaka; N. Eura; M. Yamaoka; M. Ozaki; T. Shiota; H. Nanaura; K. Sugie

EP.06
Granulomatous myositis, perimysial pathology and immune-mediated necrotizing myopathy in anti-PL7 antisynthetase syndrome
S. Souvannanorath; F. Cohen-Aubart; J. Authier

EP.07
Morphometrical quantification of histopathological parameters in SSc myopathy
L. Zaidan; N. Le Gouellec; N. Dognon; E. Hachulla; L. Mouthon; J. Authier

EP.08
Involvement of interferon-gamma in the physiopathology of inclusion body myositis
C. Hou; B. Periou; M. Gervais-Taurel; Y. Baba-Amer; F. Relaix; M. Bencze; J. Authier

EP.09
NanoString technology distinguishes anti-TIF-1g+ from anti-Mi-2+ dermatomyositis patients
C. Preusse; P. Eede; L. Heinzerling; K. Freitag; R. Koll; W. Froehlich; U. Schneider; Y. Allenbach; O. Benveniste; A. Schänzer; H. Goebel; W. Stenzel; J. Radke

EP.10
Dermatomyositis: muscle pathology according to antibody subtypes
J. Tanboon; M. Inoue; Y. Saito; S. Hayashi; S. Noguchi; N. Okiyama; M. Fujimoto; I. Nishino

EP.11
Racial disparities in skin tone representation of dermatomyositis rashes
S. Babool; S. Bhai; L. Christopher-Stine

EP.12
ER-stress and UPR-activation in immune-mediated necrotizing myopathy
C. Preusse; T. Marteau; N. Fischer; A. Hentschel; S. Lang; C. Dittmayer; U. Schneider; U. Schara-Schmidt; Y. Allenbach; O. Benveniste; H. Goebel; W. Stenzel; A. Roos

EP.13
Polymyositis-Mito and inclusion body myositis - shared T cell signatures may allow prognostic predictions
F. Kleefeld; C. Preusse; H. Goebel; K. Hahn; C. Dittmayer; W. Stenzel; A. Uruha

EP.14
Inflammatory features in sporadic late onset nemaline myopathy
J. Tanboon; A. Uruha; Y. Arahata; C. Dittmayer; L. Schweizer; H. Goebel; I. Nishino; W. Stenzel

EP.15
Severe Guillaine Barre syndrome associated with nelarabine with good prognosis
I. Hughes; E. Whitehouse; R. Wynn

EP.16
Myositis mimics: Not all that is inflamed is myositis
C. Sanderson; S. Bhai

EP.17
The impact of HTLV-I infection on clinical features of inclusion body myositis
S. Yamashita; K. Hara; N. Tawara

EP.18
Oculopharyngeal Muscular dystrophy patients with treatment-responsive, statin-associated autoimmune necrotic myopathy
K. Alrasheed; B. Brais; J. Schulz; T. Wein; J. Karamchandani; E. O'Ferrall

COVID-19 and neuromuscular diseases (EP.19-24)
EP.19
Facial Onset Acute Inflammatory Demyelinating Polyneuropathy Related to SARS-CoV-2
A. Alaamel; R. Şahin; M. Hashal; T. Taşkınoğlu; T. Özel; N. Şimşek Erdem; H. Uysal

EP.20
The change in weight gain during the coronavirus pandemic in children with Duchenne muscular dystrophy
Z. Alhaswani

EP.21
COVID-19 pandemia: Physical, psychological and social impact of lockdown on neuromuscular patients.
S. Delstanche; C. Bernar; L. Buscemi; C. Dubois; M. Duclos; L. Medard; L. Servais

EP.22
COVID-19 in children with neuromuscular disorders
D. Natera-de Benito; S. Aguilera-Albesa; L. Costa-Comellas; M. García-Romero; C. Miranda-Herrero; C. Ortez; L. Carrera-García; J. Expósito-Escudero; J. Rúbies Olives; O. García-Campos; E. Martínez del Val; J. Martinez Garcia; I. Medina Martinez; R. Cancho-Candela; M. Fernandez-Garcia; S. Pascual-Pascual; D. Gómez-Andrés; A. Nascimento

EP.23
Telemedicine tools to break down barriers in neuromuscular diseases: Clinical patient management system (CPMS) and telegenetics
F. Fortunato; M. Farnè; F. Bianchi; M. Neri; G. Siciliano; V. Sansone; A. Barp; E. Albamonte; G. Vita; A. Atalaia; T. Evangelista; F. Gualandi; A. Ferlini

EP.24
COVID-19 mitigation strategies for outcome measure assessments, training, and endpoint collection in clinic, clinical trials and natural history studies
M. James; L. Alfano; K. Rose; L. Lowes; M. Eagle

Congenital myopathies - Centronuclear myopathies (EP.25-31)
EP.25
Myotubular and centronuclear myopathy patient registry: Accelerating the pace of research and treatment
J. Bullivant; B. Porter; L. Murphy; L. Render; M. Bellgard; A. Lennox; M. Spring; A. Hollander; C. Bönnemann; H. Jungbluth; A. Buj-Bello; J. Dowling; C. Marini-Bettolo

EP.26
Leveraging natural history data in one- and two-arm hierarchical Bayesian studies of rare disease progression
A. Monseur; B. Carlin; B. Boulanger; A. Seferian; L. Servais; C. Freitag; L. Thielemans

EP.27
CPEO-like presentation of X-linked myotubular myopathy in an adult male
C. von Landenberg; M. Winkler; A. Abicht; D. Wolf; C. Kornblum; J. Reimann

EP.28
ASO-mediated Dnm2 knockdown ameliorates the centronuclear myopathy phenotype of Dnm2RW/+ mice in a dose-dependent manner after disease onset
M. Depla; A. Robé; S. Buono; C. Koch; M. Bitoun; S. Colombo; B. Cowling

EP.29
Multi-omics comparisons of different forms of centronuclear myopathies and the effects of several therapeutic strategies
S. Djeddi; D. Reiss; A. Menuet; S. Freismuth; J. de Carvalho Neves; S. Djerroud; X. Massana-Muñoz; A. Sosson; C. Kretz; W. Raffelsberger; C. Keime; O. Dorchies; J. Thompson; J. Laporte

EP.30
Statistical modelling of disease progression in a preclinical model of myotubular myopathy
S. Buono; A. Monseur; A. Menuet; A. Robé; C. Koch; J. Laporte; L. Thielemans; M. Depla; B. Cowling

EP.31
Carriers in XL-MTM: a spectrum extending from asymptomatic carriers to severely affected patients - Results of an international questionnaire study
F. Braun; S. Reumers; J. Spillane; J. Bohm; M. Pennings; M. Schouten; A. van der Kooi; A. Foley; C. Bönnemann; E. Kamsteeg; C. Erasmus; U. Schara-Schmidt; H. Jungbluth; N. Voermans

Congenital myopathies - Nemaline myopathies (EP.32-43)

EP.32

Respiratory management in nemaline myopathy due to mutations in the troponin-T type 1 (TNNT1) gene

A. Zambon; F. Abel; R. Phadke; L. Feng; A. Sarkozy; A. Manzur; F. Muntoni

EP.33

Severe forms of ACTA1-related nemaline myopathy: Reassessment of the morphological, clinical and molecular aspects

C. Labasse; G. Brochier; J. Rendu; J. Bohm; S. Monges; S. Quijano-Roy; H. Amthor; L. Servais; A. Madelaine; E. Lacène; M. Bui; S. Coppens; V. Biancalana; F. Lubieniecki; N. Laing; A. Taratuto; A. Buj-Bello; T. Evangelista; J. Laporte; N. Romero

EP.34

3'UTR variant in KLHL40 causes nemaline myopathy

L. Dofash; F. Faiz; E. Servián-Morilla; E. Rivas; P. Sullivan; E. Oates; J. Clayton; R. Taylor; M. Davis; N. Laing; M. Cabrera-Serrano; G. Ravenscroft

EP.35

Cross-Sectional phenotypic and genotypic analysis of a large cohort of patients with nemaline myopathies

L. Perry; R. Phadke; R. Mein; Y. Clinch; S. Robb; P. Munot; L. Feng; C. Sewry; A. Manzur; R. Quinlivan; M. Scoto; G. Baranello; F. Muntoni; A. Sarkozy

EP.36

Food consumption, nutrition and functioning of patients with nemaline myopathy and related disorders in Finland

V. Lehtokari; M. Similä; M. Tammepuu; S. Hiekkala; S. Strang-Karlsson; C. Wallgren-Pettersson

EP.37

ACTA1 congenital myopathy: clinical and genetic spectrum of 5 chilean newborns

R. Erazo-Torricelli; A. Gallardo; E. Bertini; F. Fattori; A. Zakharova; C. Arce; E. Alcalde; J. Carrasco; P. Gómez

EP.38

Generation of induced pluripotent stem cell lines from a 4-month-old severe nemaline myopathy patient with a dominant ACTA1 c.553C>A (p.R183S) variant

J. Clayton; C. Scriba; N. Romero; E. Malfatti; S. Saker; T. Larmonier; K. Nowak; G. Ravenscroft; N. Laing; R. Taylor

EP.39

Adult-onset nemaline myopathy due to a novel homozygous variant in the TNNT1 gene

C. Fuenmayor-Fernández de la Hoz; A. Hernández-Laín; A. Arteche López; A. Hernández Voth; M. Olivé; C. Domínguez-González

EP.40

Neuromuscular junction defects in ACTA1-related nemaline myopathy

C. Bogni; E. Girard; K. Poulard; G. Brochier; E. Errazuriz-Cerda; J. Cosette; C. Labasse; A. Madelaine; A. Lia Taratuto; N. Messaddeq; L. Schaeffer; N. Romero; A. Buj-Bello

EP.41

Characterising myosin function in nemaline myopathy

J. Laitila; T. Beck; K. Pelin; C. Wallgren-Pettersson; J. Ochala

EP.42

A ddPCR method for the detection of copy number variations in the nebulin triplicate region

L. Sagath; V. Lehtokari; C. Wallgren-Pettersson; K. Pelin; K. Kiiski

EP.43
Utilization of RNA sequencing to diagnose and to provide mechanistic insight in NEB-related myopathy
S. Silverstein; S. Syeda; A. Foley; K. Meilleur; M. Leach; P. Uapinyoying; K. Chao; S. Donkervoort; C. Bönnemann

Congenital myopathies (EP.44-53)
EP.44
Electrophysiological findings in patients with congenital myopathies
R. Escobar Cedillo; B. Estrada Cortes; M. Castillo Herrera; N. Hernandez Valadez; F. Lona Pimentel; E. Malfatti; A. Hernandez; A. Cedeño; B. Vargas; A. Miranda; N. Olamendi

EP.45
Three cases of SEPN1-related myopathy
S. Lee; H. Park; Y. Choi

EP.46
Diagnosing pathogenic mutations for congenital myopathy and cardiomyopathy patients
M. Marttila; V. Gupta; Ö. Birsoy; S. Amr; B. Funke; H. Hynes; C. Genetti; L. Swanson; P. Agrawal; H. Rehm; A. Beggs

EP.47
A novel compound heterozygous mutation in the PYROXD1 gene in a patient with congenital myopathy
C. Ucar; M. Yildirim; O. Bektas; M. Altıntas; Y. Sayar; S. Teber

EP.48
Transcriptional analysis of muscles from patients with congenital myopathies
C. Bachmann; N. Kruijt; L. van den Bersselaar; K. Bouman; M. Fernandez; F. Muntoni; H. Jungbluth; N. Voermans; F. Zorzato; S. Treves

EP.49
Establishment and characterization of zebrafish models for CACNA1S congenital myopathy
Y. Endo; E. Pannia; S. Wang; L. Groom; R. Dirksen; J. Dowling

EP.50
Congenital myopathy secondary to CACNA1S mutation in two pediatric chilean patients
R. Erazo-Torricelli; C. Arce; E. Alcalde; A. Urtizberea; K. Reinbach; E. Bertini; J. Carrasco; p. Gomez

EP.51
Treatment of an animal model carrying recessive RYR1 mutations with inhibitors targeting epigenetic enzymes
A. Ruiz; C. Bachmann; S. Benucci; M. Franchini; S. Treves; F. Zorzato

EP.52
Characterization of a novel mouse model carrying the homozygous p.F4976L RyR1 mutation, identified in a severely affected child
S. Benucci; M. Franchini; A. Ruiz; C. Bachmann; L. Ruggiero; L. Santoro; F. Zorzato; S. Treves

EP.53
Continuous disease manifestations in patients with RYR1-related malignant hyperthermia and exertional rhabdomyolysis and the effects on daily life
L. van den Bersselaar; N. Kruijt; S. Buckens; L. Joosten; G. Scheffer; L. van Eijk; J. Kusters; B. van Engelen; N. van Alfen; S. Riazi; S. Treves; H. Jungbluth; E. Kamsteeg; M. Snoeck; N. Voermans

Collagen related muscle diseases (EP.54-63)
EP.54
Establishment of a col6a2 disease model in zebrafish
N. Odenthal; V. Vedder; M. Behrensen; F. Haarich; J. Erdmann

EP.55
The Global registry for COL6-related muscle diseases
L. Imber; A. Blain; V. Straub

EP.56
Collagen VI-related neuropathy
K. Arntzen; K. Müller; S. Løseth; S. Mellgren; A. Bågenholm; H. Halvorsen; E. Buvang; G. Hoem; C. Jonsrud; M. Van Ghelue

EP.57
Identifying preclinical outcome measures of a novel humanized knock-in mouse modeling a collagen VI deep intronic pathogenic variant
F. Guirguis; V. Bolduc; J. Cheng; L. Garrett; C. Bönnemann

EP.58
Expanding the spectrum of recessive collagen XII-related EDS/myopathy overlap disorder caused by biallelic variants in COL12A1
R. McCarty; Study Group

EP.59
Genetic etiology of retractile myopathies in a cohort of 80 children under 11 years following NGS analysis
C. Metay; V. Jobic; A. Isapof; J. Cuisset; C. Barnerias; S. Whalen; F. Demurger; J. Melki; F. Jobic; A. Afenjar; I. Desguerre; K. Benistan; Y. Elaribi; A. Ferreiro; V. Laugel; M. Nougues; A. Benezit; J. Davion; S. Quijano; P. Richard

EP.60
Muscle ultrasound in COL6-related muscular dystrophy: Patterns and progression
S. Syeda; M. Mohammed; A. Foley; S. Donkervoort; D. Saade; S. Neuhaus; P. Mohassel; D. Bharucha-Goebel; M. Leach; M. Fink; J. Dastgir; C. Bönnemann

EP.61
Skeletal dysplasia, abnormal collagen, and a COL11A1 gene variant: report of one case
A. Cotta; A. Cunha-Jr; E. Carvalho; J. Valicek; B. Cordeiro; A. Reis

EP.62
Increasing allele selectivity of small interfering RNAs to target a dominant-negative glycine substitution causing a collagen VI-related dystrophy
A. Sarathy; A. Brull; V. Bolduc; G. Chen; R. McCarty; C. Bönnemann

EP.63
Neonatal clinical features distinguishing COL6-related dystrophy and TTN-related myopathy
M. Mohammed; S. Syeda; A. Foley; S. Donkervoort; S. Neuhaus; D. Saade; P. Mohassel; D. Bharucha-Goebel; M. Leach; K. Meilleur; M. Fink; S. Iannaccone; C. Konersman; C. Bönnemann

Congenital muscular dystrophies (EP.64-70)
EP.64
Brain image phenotypes and developmental milestones in Fukuyama congenital muscular dystrophy
Y. Shimizu-Motohashi; N. Sato; E. Takeshita; A. Ishiyama; M. Mori-Yoshimura; Y. Oya; I. Nonaka; K. Maruo; H. Komaki; M. Sasaki

EP.65
Evaluation of the levels of essential trace elements in patients with Fukuyama congenital muscular dystrophy
K. Ishiguro; T. Sato; M. Shichiji; Y. Kihara; T. Murakami; S. Nagata; K. Ishigaki

EP.66
Identification of lamin A interactors in healthy and congenital muscular dystrophy immortalized myoblasts
E. Storey; S. Khilar; I. Holt; S. Shirran; G. Morris; H. Fuller

EP.67
Congenital muscular dystrophy (head drop) due to LMNA mutation: description of 5 Chilean patients
R. Erazo-Torricelli; A. Urtizberea; A. Cobo; P. Richard; M. Schultz

EP.68
Therapeutic effect of linker protein-mediated gene therapy in a mouse model for LAMA2-related muscular dystrophy
J. Reinhard; S. Lin; M. Rüegg

EP.69
LAST STRONG: LAMA2 and SELENON To study trial readiness, outcome measures and natural history
K. Bouman; J. Groothuis; J. Doorduin; N. van Alfen; F. Udink ten Cate; F. van den Heuvel; R. Nijveldt; W. van Tilburg; S. Buckens; A. Dittrich; J. Draaisma; M. Janssen; E. Kamsteeg; E. van Kleef; J. Smeitink; F. van Tienen; H. Smeets; B. van Engelen; C. Erasmus; N. Voermans

EP.70
Exploring the role of genetic modifiers in a mild LAMA2-RD case associated with a LAMA2 loss-of-function mutation
V. Pini; B. Weisburd; V. Ganesh; S. Di Troia; F. Catapano; S. Aguti; E. Busch-Nentwich; F. Muntoni

Distal myopathies (EP.71-76)
EP.71
Long-term evaluation parameters and complications in GNE myopathy: a five-year observational follow-up natural history study
M. Mori-Yoshimura; Y. Yajima; A. Kimura; K. Segawa; Y. Oya; K. Mizuno; S. Noguchi; I. Nishino; Y. Takahashi

EP.72
Isoform specific variant in MLIP as a potential cause of adult-onset distal myopathy
J. Mezreani; F. Martin; S. Audet; V. Triassi; J. Charbonneau; E. Bareke; A. Laplante; B. Brais; E. O'Ferrall; J. Karamchandani; M. Tetreault

EP.73
Effects of SMPX on stress granule dynamics
J. Sarparanta; P. Jonson; H. Luque; A. Vihola; B. Udd

EP.74
GNE pathogenic variant p.D207V rarely develops myopathy in homozygote; GNE might not be the only pathogenic determinant of GNE myopathy
W. Yoshioka; K. Sonehara; A. Iida; Y. Oya; T. Kurashige; M. Okubo; M. Ogawa; F. Matsuda; K. Higasa; M. Mori-Yoshimura; H. Nakamura; S. Hayashi; Y. Okada; S. Noguchi; I. Nishino

EP.75
Functional validation of a novel variant of the SPTAN1 gene identified in a family with distal motor myopathy with nerve involvement
S. Elouej; I. Nelson; E. Cohen; R. Ben Yaou; A. Isapof; O. Dubourg; N. Romero; G. Bonne; M. Biferi; T. Stojkovic

EP.76

Mutations in the prion-like domain of heterogenous nuclear ribonuclearprotein A1 (HNRNPA1) cause proximal and distal myopathies

M. Johari; S. Rusanen; M. Savarese; P. Jonson; J. Sarparanta; A. Vihola; I. Paramonov; L. Matalonga; A. Topf; M. Christine; M. Fradin; F. Letournel; P. Marcorelles; A. Nadaj; M. Spinazzi; P. Hackman; B. Udd

DMD - animal models (EP.77-96)

EP.77

Subcellular organization of skeletal muscle dystrophin is restored by tricyclo-DNA mediated exon skipping in the DmdEGFP-mdx mouse model

A. Morin; O. Petrova; M. Petkova; T. Tensorer; T. Manoliu; I. Richard; L. Garcia; M. Schuelke; C. Laplace-Builhé; A. Goyenvalle; A. Stantzou; H. Amthor

EP.78

BCAA supplementation does not improve muscle function in mdx and D2-mdx mouse models for Duchenne muscular dystrophy

S. Engelbeen; C. Tanganyika-de Winter; D. Van De Vijver; M. Holierhoek; A. Yavas; S. Kooijman; A. Aartsma-Rus; M. van Putten

EP.79

Deletion of Exons 6 and 7 using AAV-CRISPR restores dystrophin expression and ameliorates dystrophic phenotype in DMD mouse model

T. Egorova; A. Polikarpova; I. Savchenko; S. Vassilieva; Y. Ivanova; V. Skopenkova; M. Dzhenkova; D. Tsvirkun; A. Shmidt; A. Khamatova; V. Loginov; O. Velyaev; A. Deykin; V. Soldatov; M. Bardina

EP.80

"Of Mice and Measures": a natural history study of the D2-mdx mouse model to improve translational research for Duchenne muscular dystrophy

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M. Falzarano; R. Rachele; M. Mietto; F. Fortunato; R. Selvatici; P. Spitali; F. Montanaro; F. Muntoni; A. Ferlini

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C. Chiriboga; C. Bruno; T. Duong; D. Fischer; J. Kirschner; E. Mercuri; M. Gerber; K. Gorni; H. Kletzl; I. Carruthers; C. Martin; F. Warren; M. Scoto

EP.280
Pooled safety data from the risdiplam clinical trial development program
G. Baranello; L. Servais; E. Bertini; C. Chiriboga; B. Darras; J. Day; N. Deconinck; D. Fischer; N. Goemans; J. Kirschner; A. Klein; R. Masson; M. Mazurkiewicz-Bełdzińska; Y. Wang; S. Bader-Weder; K. Gorni; B. Jaber; T. McIver; R. Scalco; E. Mercuri

EP.281
Branaplam in type 1 spinal muscular atrophy: second and third part of a phase II study
S. Jevtic; D. Carr; A. Dobrzycka-Ambrozewicz; K. Kotulzca-Jozwiak; O. Lvova; T. Pervunina; Y. Petryaikina; I. Shishimorov; D. Vlodavets; S. Nally; H. Ramos; B. Borowsky

EP.282
Preserved swallowing function in infants who initiated nusinersen treatment in the presymptomatic stage of SMA: results from the NURTURE study
V. Sansone; K. Swoboda; D. De Vivo; E. Bertini; W. Hwu; C. Makepeace; J. Bohn; R. Chin; S. Raynaud; A. Paradis

EP.283
Nusinersen in children with spinal muscular atrophy (SMA) who received onasemnogene abeparvovec: Design of the phase 4, open label respond study
C. Proud; J. Parsons; J. Brandsema; R. Finkel; K. Swoboda; R. Foster; C. Makepeace; A. Paradis; Z. Berger; K. Somera-Molina

EP.284
SUNFISH Part 2: 24-month efficacy and safety of risdiplam in patients with Type 2 or non-ambulant Type 3 spinal muscular atrophy (SMA)
A. Nascimento; J. Day; N. Deconinck; E. Mazzone; M. Oskoui; K. Saito; C. Vuillerot; G. Baranello; O. Boespflug-Tanguy; N. Goemans; J. Kirschner; A. Kostera-Pruszczyk; L. Servais; M. Gerber; K. Gorni; C. Martin; R. Scalco; H. Staunton; W. Yeung; E. Mercuri

EP.285
Nusinersen dosing patterns in US patients with spinal muscular atrophy (SMA) type 1: Findings from a retrospective claims database analysis
M. Gauthier-Loiselle; M. Cloutier; W. Toro; A. Patel; S. Shi; M. Davidson; M. Bischof; N. LaMarca; O. Dabbous

EP.286
Phosphorylated neurofilament heavy chain level is associated with future motor function in nusinersen-treated individuals with infantile-onset SMA
T. Crawford; K. Swoboda; C. Sumner; M. Farrar; F. Muntoni; R. Finkel; E. Mercuri; J. Sohn; M. Petrillo; R. Chin; X. Jiang; A. Paradis

EP.287
Evaluation of biomarkers and clinical outcome measures during 22 months of nusinersen treatment in 16 adult SMA 3-4 patients
B. De Wel; M. De Schaepdryver; K. Poesen; K. Claeys

EP.288
Experience of nusinersen treatment to adult patients with spinal muscular atrophy
T. Saito; Y. Matsuoka; H. Odani; T. Matsumura; H. Nishio

EP.289
Routine practices in use of onasemnogene abeparvovec (OA) in older patients with spinal muscular atrophy (SMA): Early findings from RESTORE
L. Servais; D. De Vivo; J. Kirschner; E. Mercuri; F. Muntoni; E. Tizzano; S. Roy; K. Saito; M. Menier; N. LaMarca; F. Anderson; O. Dabbous; R. Finkel

EP.290
Newborn screening (NBS) for spinal muscular atrophy (SMA) in the United States (US): Early findings from the RESTORE registry
L. Servais; D. De Vivo; J. Kirschner; E. Mercuri; F. Muntoni; E. Tizzano; S. Roy; K. Saito; M. Menier; N. LaMarca; F. Anderson; O. Dabbous; R. Finkel

EP.291
Real-world treatment patterns and outcomes in patients with spinal muscular atrophy (SMA): Updated findings from the RESTORE Registry
L. Servais; J. Day; D. De Vivo; E. Mercuri; F. Muntoni; P. Shieh; E. Tizzano; I. Desguerre; K. Saito; M. Menier; N. LaMarca; F. Anderson; O. Dabbous; R. Finkel

EP.292
Real-world outcomes of onasemnogene abeparvovec (OA) alone or with prior nusinersen in pediatric SMA: Interim analysis of a US chart review
O. Dabbous; M. Yang; M. Georgieva; W. Toro; N. LaMarca; A. Patel; N. Minkoff; C. Carley; J. Zhu; A. Anderson; E. Wu

EP.293
CT-guided nusinersen injection techniques in patients with SMA
S. Bensoussan; L. Sagi; Y. Fainmesser; A. Weizmann; A. Shtamler; I. Opincariu; V. Drory; A. Fattal-Valevski

EP.294
Determination of cerebrospinal fluid and serum neurofilament (pNF-H) concentration in type 2 and type 3 SMA children under Nusinersen treatment
S. Trifunov; L. Carrera; J. Exposito Escudero; A. Codina; D. Natera-de Benito; C. Ortez; J. Medina; S. Bernal; L. Alias; C. Badosa; S. Torres; D. Alcolea; A. Nascimento; C. Jimenez-Mallebrera

Neuromuscular junction related disorders (EP.295-300)
EP.295
Brachio-cervical inflammatory myopathy associated with myasthenia gravis
S. Souvannanorath; T. Nordine; T. Gendre; J. Lefaucheur; V. Plante-Bordeneuve; J. Authier

EP.296
Effect of ipidacrine on the decrement of the M-response in a patient with generalized myasthenia gravis
O. Sidorova; S. Kotov; M. Khirbek; A. Kotov

EP.297
Adding persistent gaze to orthoptic measurements in myasthenia gravis as a sensitive tool to identify involved extra-ocular muscles
K. Keene; J. Beenakker; I. Notting; J. de Nie; J. Verschuuren; H. Kan; M. Tannemaat

EP.298
Juvenile myasthenia gravis and determining the causes of delays to diagnosis: A ten-year experience
O. Abdul Hamid; E. Caron

EP.299
Paediatric myasthenia gravis: A cohort of 20 years in a referral neuromuscular center.
C. Ortez; M. Botelli; J. Exposito; L. Carrera; D. Natera De Benito; G. Nolasco; J. Colomer; A. Nascimento

EP.300
A case of congenital myasthenic syndrome diagnosed at adolescent period
S. Sahin; M. Yildirim; O. Bektas; M. Yuksel; A. Kartal; S. Teber

New genes and diseases (EP.301-305)
EP.301
The alpha2-subunit of the AP2 clathrin adaptor as the causal gene in an atypical myopathy with granulofilamentous inclusions
G. Moulay; I. Nelson; J. Lainé; E. Cohen; M. Lemaître; K. Mamchaoui; L. Julien; G. Brochier; M. Beuvin; R. Ben Yaou; E. Malfatti; C. Fardeau; M. Fardeau; N. Romero; M. Bitoun; T. Stojkovic; G. Bonne; S. Vassilopoulos

EP.302
Peptidylarginine deiminase 2 (PAD2) gene variant leading to abnormal myelination, and motor dysfunction
G. Eser; G. Celik; S. Ceylaner; T. Tuncali; Z. Yapici; H. Topaloglu

EP.303
Neonatal-lethal dilated cardiomyopathy due to a homozygous LMOD2 donor splice-site variant
M. Yuen; L. Worgan; J. Iwanski; C. Pappas; H. Joshi; J. Churko; S. Arbuckle; E. Kirk; T. Roscioli; C. Gregorio; S. Cooper

EP.304
Early onset lower limp assimmetry: A new clinical phenotype of calsequestrin mutation
C. Angelini; V. Nigro

EP.305
Homozygous WASHC4 variant in two sisters causes a syndromic phenotype with skeletal muscle involvement
A. Gangfuß; A. Czech; A. Hentschel; U. Münchberg; R. Horvath; A. Töpf; E. O'Heir; H. Lochmüller; F. Stehling; C. Kiewert; A. Sickmann; A. Kuechler; K. Frank; H. Kölbel; J. Christiansen; U. Schara-Schmidt; A. Roos

New insights into cellular or muscle function (EP.306-311)
EP.306
Human muscle pathology is associated with altered phosphoprotein profile of mitochondrial proteins in the skeletal muscle
B. Sunitha; M. Kumar; N. Gowthami; S. Unni; N. Gayathri; T. Keshava Prasad; A. Nalini; K. Polavarapu; S. Vengalil; K. Veeramani Preethish; B. Padmanabhan; M. Srinivas Bharath

EP.307
Role of cAMP-dependent pathways on mRNA levels of myosin heavy chain class II and interleukin-6 in murine myocytes
J. Yamaji; R. Hiroshima; Y. Mori

EP.308
Isolation of human fibroadipogenic progenitors and satellite cells from frozen muscle biopsies
X. Suárez-Calvet; E. Fernández-Simón; P. Piñol-Jurado; J. Alonso-Pérez; A. Carrasco-Rozas; C. Lleixà; S. López-Fernández; G. Pons; L. Soria; A. Bigot; I. Illa; E. Gallardo; J. Jaiswal; J. Díaz-Manera

EP.309
Generation and characterization of a Pax7-HA knock-in mouse line
S. Hayashi; Y. Inoue; H. Kosako; T. Inoue; S. Noguchi; I. Nishino

EP.310
Molecular and cellular differences between skeletal muscles revealed by large-scale RNA expression profiling of six muscles across 20 young adults
T. Abbassi-Daloii; S. El Abdellaoui; L. Voortman; D. Meuffels; E. van Arkel; V. Raz; P. 't Hoen; H. Kan

EP.311
Molecular and genetic characterization of the role of the Krüppel-like transcription factor Klf4 in skeletal muscle stem cells
P. Geara; L. Machado; F. Relaix; P. Mourikis

Application of next generation technologies (EP.312-321)
EP.312
Whole exome sequencing and whole Genome sequencing re-analysis to identify the genetic causes of undiagnosed neugomuscular phenotypes
R. Selvatici; M. Neri; F. Fortunato; R. Rossi; A. D'Amico; E. Bertini; M. Pane; E. Mercuri; S. Fini; S. Bigoni; F. Gualandi; A. Ferlini

EP.313
The utility of RNA-sequencing in the study of muscle biology and neuromuscular disease
A. Smolnikov; E. Oates; M. Wilkins

EP.314
Determining the contribution of RFC1 repeat expansions to disease in an Australasian neurological disease cohort shows increased genetic heterogeneity
C. Scriba; F. Faiz; P. Lamont; N. Laing; S. Beecroft; M. Davis; G. Ravenscroft

EP.315
Use of NGS for diagnosis of asymptomatic hyperckemia in childhood
P. Marti; N. Muelas; I. Pitarch; A. Casasus; I. Azorin; R. Vilchez; M. Nieto; J. Vilchez

EP.316
Diagnostic yield of targeted next-generation sequencing panels in muscular dystrophies
B. Cavdarli; A. Ceylan; B. Arslan Satılmış; Ö. Yayici Köken; C. Semerci Gündüz; H. Topaloglu

EP.317
Genetic algorithm in etiological research of hereditary peripheral neuropathies
G. Ceylan; E. Habiloglu; B. Cavdarli; E. Tuncez; S. Bilen; Ö. Yayıcı Köken; C. Semerci Gündüz

EP.318
Next-generation sequencing panel approach for the molecular diagnosis of neuromuscular diseases in Argentinian paediatric patients
E. Foncuberta; F. Lubieniecki; E. Cavassa; F. Garcia; N. Piergrossi; G. Veneruzzo; P. Gravina; S. Monges

EP.319
The Treatabolome flags treatable genes and variants: an emerging concept.
A. Atalaia; R. Thompson; L. Matalonga; C. Hernandez-Ferrer; A. Corvo; L. Carmody; B. Zurek; R. Ben Yaou; R. Horvath; H. Graessner; O. Riess; P. Robinson; H. Lochmuller; S. Beltran; G. Bonne; Treatabolome Project Group

EP.320

AI-based processing of patient voice in rare neuromuscular disorders: Understanding patient experience and early disease detection

I. Efimenko; M. Samsonov; A. Paleeva; S. Kurbatov; I. Stanovaya; O. Germanenko; S. Bortkevicha; K. Demina; S. Efimenko; A. Ivankov; V. Khoroshevsky; E. Krotova; A. Leonova; S. Mikhailov; A. Sutormina; E. Smirnova; A. Tsvetkova; O. Ramzaitseva; O. Pozdnyakova; M. Lutoshkina

EP.321

Revolutionizing drug discovery in genetically defined muscle disease using single-cell and high dimensional datasets

L. Rojas; A. Ergun; A. Accorsi; C. Sartain; A. Raman; R. Lieberman; E. Valentine; M. Wertz; E. Townsend; G. Wilson; D. Plioudakis; O. Wallace; S. Silver; C. Moxham

Outcome measures (EP.322-327)

EP.322

Relevance of maximal performance measurement during real-life ambulation in neuromuscular diseases: patients' and caregivers' perspectives

M. Annoussamy; D. Eggenspieler; E. Camino; B. Porter; J. Bullivant; L. Imber; L. Murphy; Z. Hughes; W. Hughes; L. Servais

EP.323

Wearable technology allows faster and more accurate detection of functional changes in Duchenne muscular dystrophy than traditional hospital-based tests

M. Annoussamy; D. Eggenspieler; P. Strijbos; M. Rabbia; A. Tricot; D. Vissière; L. Servais

EP.324

TREAT-NMD remote outcome measures group -- consensus guidelines and recommendations

L. Lowes; L. Alfano; R. Bendixen; E. Hardy-Bosson; M. James; K. Laubscher; M. Leffler; C. Lilien; A. Mayhew; S. Segovia Simon; J. Wallace; M. Jain

EP.325

Remote testing: is it worth the effort? Feasibility in the clinical and research setting

B. Powers; N. Reash; M. Iammarino; K. Shannon; M. Waldrop; A. Connolly; K. Flanigan; C. Tsao; J. Mendell; L. Lowes; L. Alfano

EP.326

Serum neurofilament light chain analysis in giant axonal neuropathy

D. Bharucha-Goebel; D. Saade; E. Paredes; Y. Hu; A. Saxena; P. McCoy; G. Averion; M. Jain; G. Norato; K. Cheung; M. Waite; R. Foley; T. Lehky; S. Gray; C. Bönnemann

EP.327

The relationship between illness perceptions and quality of life outcomes in adolescents with neuromuscular disorders

S. Geuens; J. Willen; S. Prikken; N. Goemans; K. Luyckx; L. De Waele

Imaging (EP.328-339)

EP.328

The effect of chronic corticosteroid use in boys with Duchenne muscular dystrophy on brain structure: a pilot T1-weighted MR imaging study

S. Geuens; C. Sleurs; J. Lemiere; M. Verly; E. Niks; N. Goemans; H. Kan; L. De Waele; N. Doorenweerd

EP.329

MRI of muscles and mitochondrial disorders in the face-scapulo-shoulder muscular dystrophy (FSCMD)

S. Kotov; O. Sidorova; M. Bunak; E. Borodataya; I. Vasilenko; Y. Filyushkin

EP.330
Whole body MRI quantitative muscle analysis to evaluate efficacy of Losmapimod in a phase 2 placebo-controlled study in subjects with FSHD (ReDUX4)
M. Mellion; ReDUX4 Study Grp

EP.331
Quantitative muscle analysis in FSHD using Whole-Body MRI: Composite muscle measurements for cross-sectional analysis
M. Mellion; P. Widholm; M. Karlsson; A. Ahlgren; O. Dahlqvist-Leinhard; R. Tawil; K. Wagner; J. Statland; L. Wang; P. Shieh; B. van Engelen; D. Cadavid; L. Ronco; A. Odueyungbo; J. Han; M. Hatch

EP.332
Patients with McArdle disease have increased fat replacement of paraspinal muscles on MRI: A European multicenter study
N. Løkken; K. Revsbech; L. Jacobsen; A. Martinuzzi; A. Toscano; M. Martin; J. Díaz Manera; C. Stefan; C. Domínguez-González; G. Brondani; O. Musumeci; C. Merino-Sanchez; C. Nuñez; P. Montesinos; F. Granata; T. Khawajazada; J. Vissing

EP.333
Muscle MRI characteristic pattern for late-onset TK2 deficiency diagnosis
C. Dominguez-Gonzalez; R. Fernandez-Torron; U. Moore; B. Velez; J. Alonso Perez; L. Gonzalez-Mera; M. Olivé; J. García García; G. Morís; J. León Hernández; N. Muelas; C. de Fuenmayor Fernández de la H; M. Martín; J. Díaz-Manera; C. Paradas

EP.334
Using MRI to review contractile properties and fat replacement of muscles in patients with myotonia congenita
L. Jacobsen; J. Pedersen; S. Skriver; M. Stemmerik

EP.335
Reducing the minimum detectable change in vastus lateralis fat fraction using MRI in clinical trials for Duchenne's muscular dystrophy (DMD) therapy
M. Hammond; F. Roche; J. Harris; B. Luna; J. Mary; M. Berger; F. Vincent; S. Zabbatino; L. Heinichen; R. Scheyer; S. Holland

EP.336
Quantitative MRI, strength and function of the upper extremity flexor muscles in non-ambulant DMD patients: an 18-month follow-up analysis
A. Prins; K. Naarding; M. Holst, van der; J. Verschuuren; E. Niks; H. Kan

EP.337
Different approaches to analyze muscle fat replacement with dixon MRI
A. Alonso-Jiménez; C. Nuñez-Peralta; J. Alonso-Pérez; C. García; E. Montiel; I. Belmonte; I. Pedrosa; P. Montesinos; S. Segovia; J. Díaz-Manera

EP.338
Fat fraction histogram metrics assessed by quantitative MRI demonstrate differences between neuromuscular diseases
H. Reyngoudt; P. Baudin; E. Araujo; P. Carlier; B. Marty

EP.339
Longitudinal assessment of muscle ultrasound in oculopharyngeal muscular dystrophy
R. Kroon; J. Kalf; B. de Swart; B. van Engelen; C. Horlings

Registries and care of NMD (EP.340-347)
EP.340
Improving and de-risking clinical trials in neuromuscular disease: over 10 years of the TREAT-NMD Advisory Committee for Therapeutics (TACT)
C. Turner; C. Csimma; A. De Luca; J. Lee; K. Wagner; D. Wells; V. Straub

EP.341
TREAT-NMD global registries platform
C. Ogden; S. Simon; J. McKenna; S. Cardiff; J. Wilkins; B. Watling; J. Bullivant; J. Das; B. Leary; C. Turner; B. Tye; M. Fowler; P. Owens; L. Braithwaite; S. Woods; D. Osredkar; B. Palmafy; T. Chamora; M. Guglieri; C. Campbell; A. Ambrosini

EP.342
Uniting the Canadian NMD community: One year of the neuromuscular disease network for Canada (NMD4C)
E. Beattie; J. Dowling; J. Warman Chardon; R. Kothary; S. Lintern; R. Amin; T. Buffone; B. Brais; C. Campbell; C. Gagnon; H. Gonorazky; J. Karamchandani; L. Korngut; H. McMillan; M. Oskoui; H. Osman; K. Selby; D. Wojtal; N. Worsfold; H. Lochmüller

EP.343
ERN EURO-NMD registry hub
A. Atalaia; S. Bakker; C. D'Angelo; E. Sakellariou; N. van Lin; G. Bassez; C. Eng; F. Lamy; M. Frenkian; E. Vroom; D. Athanasiou; H. Lochmuller; P. 't Hoen; A. Tassoni; T. Evangelista

EP.344
The international GNE myopathy patient registry
L. Imber; V. Straub

EP.345
The EURO-NMD registry project: how to establish the datasets for the first European registry for all neuromuscular diseases
C. D'Angelo; A. Atalaia; D. Jaeger; N. van Lin; P. 't Hoen; T. Evangelista; A. Tassoni; H. Lochmuller

EP.346
The Egyptian neuromuscular registry
R. El Sherif; M. Gamal; A. Hanafy

EP.347
Development of a standard of care for patients with valosin-containing protein associated multisystem proteinopathy
M. Korb; A. Peck; K. Berger; M. James; N. Ghoshal; E. Healzer; C. Henchcliffe; S. Khan; P. Mammen; S. Patel; G. Pfeffer; S. Ralston; B. Roy; B. Seeley; A. Swenson; T. Mozaffar; C. Weihl; V. Kimonis; L. Alfano

Other NMDs (EP.348-362)
EP.348
Can immunohistochemistry help narrow the differential diagnosis of myopathies with rimmed vacuoles?
E. Lacene; A. Chanut; T. Stojkovic; P. Laforet; C. Metay; I. Nelson; G. Brochier; C. Labasse; A. Madelaine; N. Romero; T. Evangelista

EP.349
GFPT1-related myopathy overlapping with autophagic vacuolar myopathy. A case report
L. Costa-Comellas; M. Alvarez-Molinero; D. Gómez-Andrés; M. Gratacós Viñola; E. Martínez-Saez; A. Sánchez-Montañez; K. Chao; S. Donkervoort; C. Bönnemann; M. Olivé; F. Munell

EP.350
A study of LINC complex proteins reveals temporal emerin and SUN2 expression changes during myoblast differentiation
E. Storey; I. Holt; G. Morris; H. Fuller

EP.351
Painful spasms during childhood: Stiff-person syndrome
A. Curado; M. Rosário; C. Gonçalves; D. Silva; I. Conceição; J. Coelho; T. Santos; T. Moreno

EP.352
Venglustat, a novel brain-penetrant glucosylceramide synthase inhibitor, for GM2 gangliosidosis and related diseases: Phase 3 AMETHIST trial design
M. Petrovic; R. Zheng; C. Canabarro; P. Minini; I. Batsu

EP.353
Spinal surgery in neuromuscular patients: the experience of a UK tertiary paediatric centre between the years of 2003-2021
I. Hughes; E. Whitehouse; H. Dashti

EP.354
An integrated modelling methodology for estimating global incidence and prevalence of hereditary spastic paraplegia subtypes SPG4, SPG11, SPG7 and SPG11
G. Vander Stichele; A. Dürr; G. Yoon; R. Schüle; C. Blackstone; G. Esposito; C. Buffel; I. Oliveira; C. Freitag; S. van Rooijen; S. Hoffmann; L. Thielemans; B. Cowling

EP.355
Comprehensive morphometric assessment of skeletal muscle development from birth to 18 years
T. Evangelista; M. Kandji; M. Lacene; A. Chanut; M. Thao BUI; R. Marty; L. Buffat; K. Knobloch; B. B. Rudkin; N. B Romero

EP.356
The phenotypic heterogeneity of autosomal recessive Desmin myopathies
M. Keogh; A. Topf; C. Marini-Bettolo; J. Hudson; J. Colomer; A. Nascimento; M. Oliver; R. Alvarez; H. Durmus; S. Nafissi; A. Bastian; J. Vissing; N. Witting; J. Diaz-Manera; V. Straub

EP.357
Nuclear envelope involvement in BAG3 myofibrillar myopathy
R. Robertson; M. Dicaire; N. Chabaytah; J. Lavoie; B. Brais

EP.358
Oculopharyngeal Muscular dystrophy patients with treatment-responsive, statin-associated autoimmune necrotic myopathy
K. Alrasheed; K. Alrasheed; B. Brais; B. Brais; J. Schulz; J. Schulz; T. Wein; T. Wein; J. Karamchandani; J. Karamchandani; E. O'Ferrall; E. O'Ferrall

EP.359
Novel dominant distal titinopathy phenotype associated with Copy Number Variation
A. Perrin; R. Juntas Morales; F. Chapon; C. Thèze; D. Lacourt; P. Pégeot; E. Uro-Coste; D. Giovannini; N. Leboucq; M. Mallaret; V. Rigau; K. Gaudon; P. Richard; M. Koenig; C. Métay; M. Cossée

EP.360
Is7 domain of titin is essential for cardiac function in mice
A. Biquand; S. Spinozzi; P. Tonino; J. Strom; J. Cosette; Z. Elbeck; R. Knöll; H. Granzier; W. Lostal; I. Richard

EP.361
Early presentation of cardiac dysfunction in Salih myopathy
P. Anandakrishnan; J. Sivabalakrishnan

EP.362
Expanding the clinical phenotype of recessive PIEZO2 mutations
A. Camacho; J. Quesada; A. Hernández Laín; C. Alonso; S. Vila; N. Núñez; R. Simón

18:30-19:30

New genes, new techniques in neuromuscular disorders - Selected oral presentations 3 (O.9-12)
(4 x 10 mins consecutive pre-recorded presentations + 20 mins live Q&A)
Moderators: Reghan Foley & Bernard Brais

O.9
Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis
P. Mohassel; S. Donkervoort; M. Lone; M. Nalls; K. Gable; S. Gupta; A. Foley; SPTLC1 Study Group; T. Hornemann; T. Dunn; C. Bönnemann

O.10
Loss of function mutations in DNAJB4 cause a myopathy with early respiratory failure
A. Topf; R. Bengoechea; J. Duff; R. Charlton; M. Mroczek; S. Kapetanovic Garcia; C. Dominguez; A. Alsaman; A. Findlay; G. Ravenscroft; C. Weihl; V. Straub

O.11
Transcriptomic profiling of paired normal skeletal muscle using bulk RNAseq and snRNAseq
S. Nieves-Rodriguez; F. Barthelemy; J. Woods; E. Douine; R. Wang; A. Huang; M. Miceli; S. Nelson

O.12
Muscle biopsy and RNAseq in the diagnosis of titin related diseases
M. Savarese; M. Johari; A. Vihola; H. Luque; M. Iacomino; P. Hackman; B. Udd

19:30-20:00 **Breakout networking session – On the topic of selected oral presentations 3**

20:00–21:00 **Pre-clinical developments in neuromuscular disorders - Selected oral presentations 4 (O.13-16)**
(4 x 10 mins consecutive pre-recorded presentations + 20 mins live Q&A)
Moderators: Nicolas Wein & TBC

O.13
Overexpression of JAG1 a therapeutic modifier for Duchenne muscular dystrophy
Y. Zhang; M. Lambert; J. Widrick; J. Conner; J. Spinazzola; L. Kunkel

O.14
PDGF-AA enhances skeletal muscle fibrosis in Duchenne muscular dystrophy through Rho-kinase pathway
E. Fernández Simón; X. Suárez Calvet; A. Carrasco-Rozas; P. Piñol Jurado; S. López Fernández; C. de la Torre; J. Bech Serra; N. de Luna; E. Gallardo Vigo; J. Díaz Manera

O.15
Unprecedented low dose of AAV-mediated gene transfer corrects the pathology in a model for Fukutin-related-protein deficiencies
E. Gicquel; M. Faivre; S. Brown; L. Buscara; N. Daniele; E. Thevenot; I. Richard

O.16
Allele-specific gene editing inactivates a dominant-negative, disease-causing, single nucleotide variant in COL6A1 through non-homologous end joining
V. Bolduc; K. Sizov; P. Uapinyoying; E. Esposito; A. Brull; G. Chen; A. Sarathy; K. Johnson; C. Bönnemann

21:00-21:30 **Breakout networking session – On the topic of selected oral presentations**

Friday, 24 September 2021

13:30-14:30	**General Assembly (WMS Members only)**
15:00-15:45	**Poster highlights** (6 x 5 mins consecutive pre-recorded presentations + 15 mins live Q&A) *Moderators: Johann Böhm & Jantima Tanboon*
15:45-16:15	**Breakout networking session – On the topic of poster highlights**
16:15-17:00	**Late breaking news** (3 x 10 mins consecutive pre-recorded presentations + 15 mins live Q&A) *Moderators: Gisèle Bonne & Carsten Bönnemann*
17:15-18:15	**Prize presentations:** Johann Böhm **Close of Congress:** Volker Straub **Introduce 2022 and presentation of flag:** Jiri Vajsar