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 and Exhibition area open*

Tuesday, 21 September 2021 PRE-CONGRESS TEACHING COURSE

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

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: Vandana Gupta*
16:30-17:00	**Comfort break**
17:00-18:00	**Debate 2 – (Wo)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, Damjan Osredkar* *Moderator: Nathalie Goemans* *Moderator: Liesbeth De Waele*
19:30-19:45	**Comfort break**
19:45-20:45	**Live Fun Social Hour**

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)

In the field of neuromuscular disorders there is a continuously growing knowledge of genetic background, pathophysiology, molecular mechanisms, and therapeutic options. Translational medicine, from basic science to international clinical trials in humans are necessary to bring therapeutic options to patients around the world. In this session international trials for gene therapy (replacement and regulation of transcription), as well as pharmacotherapy, will be introduced and interim analyses will be published.

Moderators: Ulrike Schara-Schmidt & Giovanni Baranello

O.1

ASPIRO gene therapy trial in X-linked myotubular myopathy (XLMTM): update on preliminary efficacy and safety findings

P. Shieh; N. Kuntz; J. Dowling; W. Müller-Felber; A. Blaschek; C. Bönnemann; R. Foley; D. Saade; A. Seferian; 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.3a

2.5-years of vamorolone treatment in Duchenne muscular dystrophy: Results of an open label long-term extension

E. Hoffman; U. Dang; P. Clemens; H. Gordish-Dressman; B. Schwartz; L. Mengle-Gaw; M. Leinonen; E. Smith; D. Castro; N. Kuntz; R. Finkel; M. Tulinius; Y. Nevo; M. Ryan; R. Webster; J. van den Anker; L. Ward; J. Damsker; C. McDonald; M. Guglieri; J. Mah

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 **Clinical trial highlights breakout session**

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)

Neuromuscular translational research is a continuing evolving field.

In this session, the importance of well-characterised natural history, including genotype-phenotype correlations and outcome measures for rare neuromuscular conditions will be discussed as a fundamental milestone to design clinical trials aiming to bring therapeutic options to patients.

Furthermore in this session it will be discussed how improving the knowledge of genetics, pathophysiology and molecular mechanisms or rare neuromuscular conditions can play a role in preventing mortality and morbidity through modifying guidelines.

Moderators: Jordi Dias & Mariacristina Scoto

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 **Clinical research breakout session**

16:30-18:30 **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* lossof-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 leads to myopathy in homozygotes; 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
P. Mantuano; B. Boccanegra; F. Sanarica; E. Conte; A. Mele; M. De Bellis; O. Cappellari; A. De Luca

EP.81
Enhanced exon skipping and Dystrophin production in a mouse model of Duchenne muscular dystrophy with EEV-PMO treatment
N. Kreher; X. Li; M. Kheirabadi; K. Kamer; P. Dougherty; W. Lian; C. Waters; N. Gao; S. Peddigari; A. Stadheim; M. Dhanabal; N. Sethuraman; Z. Qian

EP.82
Detailed natural history study of the D2-mdx and BL10-mdx models for Duchenne muscular dystrophy
M. van Putten; C. Tanganyika-de Winter; K. Putker; S. Engelbeen; D. Van de Vijver; M. Verhaeg; M. Overzier; A. Aartsma-Rus

EP.83
Creation of Becker muscular dystrophy murine models to predict the efficacy of therapy for patients with Duchenne muscular dystrophy
A. Polikarpova; I. Galkin; D. Korshunova; V. Skopenkova; M. Dzhenkova; D. Tsvirkun; A. Shmidt; A. Khamatova; V. Loginov; A. Deikin; M. Bardina; T. Egorova

EP.84
Cholesterol metabolism is a potential therapeutic target in Duchenne muscular dystrophy
A. Vu Hong; F. Amor; G. Corre; M. Sanson; L. Suel; S. Blaie; L. Servais; T. Voit; I. Richard; D. Israeli

EP.85
Oestrogen and HPA/renin-angiotensin-aldosterone pathways are associated with the behavioural sensitivity to stress in dystrophin-deficient mice
A. Lindsay; A. Trewin; P. Della Gatta; C. Laird; A. Russell

EP.86
Identifying heart and skeletal muscle homing peptides to improve the uptake of PMOs for Duchenne muscular dystrophy therapy
A. Schneider; S. Jirka; C. Tanganyika-de Winter; H. Mei; J. Boom; A. Aartsma-Rus

EP.87
Antisense oligonucleotide-mediated knockdown of IGF-binding proteins to increase IGF-1 signaling
A. Yavas; M. van Putten; E. Niks; A. Aartsma-Rus

EP.88
Inflammasome inhibitors for the treatment of muscular dystrophies
N. Dubuisson; M. Abou-Samra; M. Davis; L. Noel; C. Selvais; S. Brichard

EP.89
TRPC1 and TRPC3 involvement in DMD physiopathology and as potential targets for treatment in complement to rAAV-microdystrophin
A. Creisméas; C. Gazaille; A. Bourdon; A. Lafoux; M. Allais; V. Le Razavet; M. Ledevin; T. Larcher; G. Toumanianz; I. Anegon; O. Adjali; C. Huchet; C. Le Guiner; B. Fraysse

EP.90
A novel AAV8-based gene therapy for Duchenne muscular dystrophy: Preclinical studies in the *mdx* Mouse
S. Kim; N. Buss; C. Qiao; H. Patel; L. Yang; K. Elliott; R. Qian; L. Ye; M. Fiscella; O. Danos

EP.91
Oral treatment with AdipoRon attenuates cardiomyopathy in an aged mouse model for Duchenne muscular dystrophy
M. Abou-Samra; A. Marino; C. Selvais; N. Dubuisson; L. Noel; C. Beauloye; S. Horman; S. Brichard

EP.92
Growth hormone secretagogues in Duchenne muscular dystrophy: preclinical proof-of-concept in vivo and in vitro assays
P. Mantuano; B. Boccanegra; E. Bresciani; F. Sanarica; A. Mele; M. De Bellis; O. Cappellari; A. Liantonio; S. Denoyelle; A. Torsello; A. De Luca

EP.93
The DLK1-DIO3 cluster miRNAs regulate mitochondrial functions in the dystrophic muscle in DMD
A. Vu Hong; N. Bourg-Alibert; P. Sanatine; J. Poupiot; K. Charton; E. Gicquel; M. Spinazzi; I. Richard; D. Israeli

EP.94
Efficient modification of DMD gene using PRIME editing
C. Happi Mbakam; J. Rousseau; G. Tremblay; J. Tremblay

EP.95
Downregulation of the genetic modifier *PITPNA* as means of therapy in Duchenne muscular dystrophy
M. Lambert; Y. Zhang; J. Spinazzola; J. Widrick; J. Conner; L. Kunkel

EP.96
Micro-dystrophin gene therapy delivery and therapeutic plasma exchange in nonhuman primates
E. Peterson; R. Potter; D. Griffin; S. Lewis; E. Pozgai; A. Meadows; L. Rodino-Klapac

DMD - clinical care (EP.97-111)

EP.97
Renal failure in an adult cohort of Duchenne muscular dystrophy attending the National hospital for neurology and neurosurgery in London
M. Desikan

EP.98
Clinical and molecular characterisation of unusual DMD mutations
S. Nicolau; T. Vetter; A. Bradley; E. Frair; R. Weiss; K. Flanigan

EP.99
Respiratory function and sleep-disordered breathing in Duchenne muscular dystrophy
A. Zambon; A. Laverty; M. Riley; D. Ridout; A. Manzur; F. Abel; F. Trucco; F. Muntoni

EP.100
Growing into adulthood with Duchenne muscular dystrophy: Complaints, secondary diagnoses, and their impact on social participation
S. Houwen; L. Heutinck; M. Jansen; Y. Krom; E. Cup; J. Hendriksen; M. Willemsen; J. Verschuuren; E. Niks; I. De Groot

EP.101
Appendicular lean mass index (ALMI) changes in patients with dystrophinopathies
B. Wong; S. Summer; P. Horn; M. Rutter; C. Tian; I. Rybalsky; K. Shellenbarger; H. Kalkwarf

EP.102
Clinical characteristics of early-onset cardiomyopathy in patients with Duchenne muscular dystrophy
W. Jinnai; Y. Shimizu-Motohashi; H. Komaki; E. Takeshita; A. Ishiyama; K. Segawa; M. Sasaki

EP.103
DMD Care UK: implementation of the latest Duchenne standards of care in the UK healthcare setting. How we are overcoming the hurdles
C. Turner; A. Childs; A. Johnson; A. Manzur; R. Quinlivan; A. Sarkozy; J. Wong; M. Guglieri

EP.104
The effect of corticosteroid on pulmonary function in non-ambulant adults with Duchenne muscular dystrophy
A. Pietrusz; M. Guglieri; R. Astin; M. Desikan; R. Muni-Lofra; A. Mayhew; K. Waller; S. Chapman; G. Ramdharry; R. Quinlivan

EP.105
Hepatic parenchymal changes and hepatomegaly in Duchenne muscular dystrophy
M. Naume; C. Høi-Hansen; A. Born; L. Borgwardt; M. Jørgensen; D. Stærk; M. Ørngreen

EP.106
Modelling growth pattern in boys with Duchenne muscular dystrophy
G. Stimpson; S. Raquq; M. Fewtrell; A. Manzur; A. Sarkozy; V. Ayyar Gupta; D. Ridout; F. Muntoni; G. Baranello

EP.107
Current situation of medical care in female carriers of DMD
J. Exposito-Escudero; A. Camacho; A. Amado; J. Fernandez; A. Molina; V. Cantarin; L. Carrera; D. Natera-De Benito; C. Ortez; A. Nascimento

EP.108
Utilisation of NIV in patients with DMD in a UK cohort
F. Cruces; M. Borghi; M. Desikan; R. Quinlivan; F. Crummy; R. Astin

EP.109
Duchenne muscular dystrophy and sleep: Patients and primary caregivers
Ö. Yayıcı Köken; P. Gültutan; E. Gürkaş; A. Değerliyurt

EP.110
Ambulatory performance of Duchenne muscular dystrophy patients on intermittent corticosteroid treatment – a single center study
N. Ikelaar; M. van der Holst; Y. Meijer - Krom; J. van Egmond - van Dam; N. van de Velde; N. Doorenweerd; E. Niks

EP.111
Evolution of scoliosis management in Duchenne muscular dystrophy
L. Wahlgren; S. Nordström; A. Kroksmark; M. Tulinius; K. Sofou

DMD / BMD - genetics (EP.112-117)

EP.112
Genotype-phenotype correlation for cognitive and neurodevelopmental comorbidities in Duchenne and Becker muscular dystrophy
J. Collyer; T. Whitaker; E. Caron

EP.113
Lack of correlation between *DMD* exon 2 duplication splicing choices and phenotype severity
M. Mietto; M. Fabris; R. Bonaccorso; S. Previtali; A. Zambon; F. Gualandi; R. Rossi; M. Falzarano; A. Ferlini

EP.114
Theragnosis for Duchenne muscular dystrophy
M. Carcione; L. Luce; C. Mazzanti; L. Mesa; A. Dubrovsky; J. Corderí; F. Giliberto

EP.115
RNA-seq analysis for Dystrophinopathy
M. Okubo; S. Noguchi; S. Hayashi; H. Komaki; I. Nishino

EP.116
Establishing clinical and genetic characteristics in young, glucocorticoid-naïve boys with Duchenne muscular dystrophy: the FOR DMD study
M. Schiava; R. Amos; H. VanRuiten; M. McDermott; W. Martens; S. Gregory; A. Mayhew; E. McColl; R. Tawil; T. Willis; K. Bushby; R. Griggs; Guglieri M & the FOR DMD group

EP.117
Whole genome sequencing and RNA analysis allow genetic diagnosis of DMD atypical mutations
R. Selvatici; R. Rossi; M. Fang; J. Lu; V. Sansone; M. Falzarano; F. Gualandi; L. Bello; E. Pegoraro; A. Ferlini

DMD / BMD - outcome measures (EP.118-132)

EP.118
The TREAT NMD DMD Global Registry development
J. Mckenna; A. Ambrosini; C. Campbell; N. Goemans; M. Heidemann; A. Martin; S. Segovia; C. Turner; I. Zito

EP.119
Evidence synthesis in rare diseases: A natural history model for Duchenne muscular dystrophy
J. Broomfield; M. Hill; M. Guglieri; M. Crowther; J. Larkindale; J. Godfrey; F. Chandler; K. Abrams

EP.120
Industry and patient advocacy led co-development of a novel digital endpoint for Duchenne muscular dystrophy
O. Anyanwu; M. Stoodley; E. Davies; D. Lewi; E. George; D. Bull; E. Crossley; A. Johnson

EP.121
A collaborative national Duchenne muscular dystrophy (DMD) registry for real world evidence
V. Hodgkinson-Brechenmacher; A. McCormick; J. Sheriko; J. Lounsberry; H. Osman; N. Worsfold; C. Campbell; J. Mah; L. McAdam; K. Selby; L. Korngut; N. CNDR Investigator Network

EP.122
Prognostic factors for pulmonary milestones in Duchenne muscular dystrophy (DMD)
N. Goemans; J. Signorovitch; G. Sajeev; B. Wong; C. Tian; C. McDonald; E. Mercuri; E. Niks; J. Freimark; M. Jenkins; C. Xu; S. Ward

EP.123
Functional trajectories of upper limb and pulmonary function before and after loss of ambulation in Duchenne muscular dystropwhy
N. Goemans; J. Signorovitch; C. McDonald; E. Mercuri; E. Niks; B. Wong; G. Sajeev; M. Fillbrunn; E. Yim; I. Dieye; S. Ward

EP.124
Actimyo®: Normative data in a non-controlled environment
M. Poleur; A. Ulinici; A. Daron; O. Schneider; F. Dal Farra; M. Demonceau; M. Annoussamy; D. Vissière; D. Eggenspieler; L. Servais

EP.125
Minimal detectable changes in functional measures in Duchenne muscular dystrophy (DMD): A study of multiple centers, networks and trial arms
F. Muntoni; J. Signorovitch; G. Sajeev; N. Done; Z. Yao; N. Goemans; C. McDonald; E. Mercuri; E. Niks; B. Wong; L. Servais; V. Straub; I. de Groot; C. Tian; A. Manzur; K. Vandenborne; I. Dieye; H. Lane; S. Ward

EP.126
DMD Hub: Preparing the field for gene therapy trials
E. Heslop; C. Turner; E. George; A. Irvin; A. Robertson; E. Crossley; A. Johnson; R. Fischer; H. Peay; F. Muntoni; V. Straub; M. Guglieri

EP.127
The Dutch multicenter Duchenne and Becker register: facilitation of trial readiness and effective use of patient data
Y. Meijer-Krom; N. van de Velde; N. Ikelaar; H. van der Holst; J. Verschuuren; E. Vroom; A. Horemans; J. Hendriksen; S. Houwen-van Opstal; I. de Groot; R. Snijder; E. Niks

EP.128
Incorporating treatment advances and a multidisciplinary approach with Duchenne muscular dystrophy: A study of online education impact on clinical care
C. Drexel; J. Brandsema; E. del Nido; W. Turell

EP.129
Value of global longitudinal strain for identification and monitoring of left ventricular dysfunction in Becker muscular dystrophy
N. van de Velde; T. Gegenava; Z. Koeks; S. Butscher; A. Roest; J. Bax; D. Atsma; P. Spitali; N. Ajmone Marsan; E. Niks

EP.130
Minor cognitive impairments in adult males with Becker muscular dystrophy
Z. Koeks; D. Hellebrekers; N. vd Velde; I. Alleman; P. Spitali; H. van Duyvenvoorde; J. Verschuuren; J. Hendriksen; E. Niks

EP.131
Epidemiology of Becker muscular dystrophy in the Netherlands
E. Schrama; Z. Koeks; N. van de Velde; H. Kan; Y. Krom; H. van Duyvenvoorde; E. Niks

EP.132
Double the trouble: familial hyperlipidemia and Becker muscular dystrophy with a hemizygous nonsense mutation in the dystrophin gene
Y. Sayar; M. Yildirim; O. Bektas; N. Yeniay Sut; S. Teber

DMD - brain (EP.133-138)

EP.133
Expression and localization of dystrophin isoforms transcripts in human adult control brain areas
M. Falzarano; R. Rachele; M. Mietto; F. Fortunato; R. Selvatici; P. Spitali; F. Montanaro; F. Muntoni; A. Ferlini

EP.134
Regulation of the alternative splicing of Dp71, the major brain dystrophin transcript, by splicing factors
C. Wood; K. Anthony

EP.135
In-depth investigation in the murine brain of the consequences of a lack of one or multiple dystrophin isoforms
M. Verhaeg; K. Adamzek; D. Van De Vijver; K. Putker; S. Engelbeen; E. Suidgeest; M. Overzier; L. van der Weerd; A. Aartsma-Rus; M. Putten

EP.136
Determinants of reduced cerebral blood flow in Duchenne muscular dystrophy
N. Ikelaar; R. Hendriksen; J. van Dijk; F. Kerkhof; R. Reijntjes; A. Roest; M. van Osch; H. Kan; R. Thijs; N. Doorenweerd; E. Niks

EP.137
Preliminary results of longitudinal brain volume analyses in adolescents with Duchenne muscular dystrophy
N. Doorenweerd; M. Hoegen; E. Broek; K. Hollingsworth; C. Marini-Bettolo; J. Hendriksen; E. Niks; V. Straub; H. Kan

EP.138
Dp71 and cognitive impairment in patients with Duchenne muscular dystrophy
K. Iskandar; B. Indraswari; A. Triono; E. Herini; Sunartini

DMD - treatment (EP.139-152)

EP.139
Phase 1/2a trial of SRP-9001 in patients with Duchenne muscular dystrophy: 3-year safety and functional outcomes
J. Mendell; Z. Sahenk; K. Lehman; C. Nease; L. Lowes; N. Reash; M. Iammarino; L. Alfano; J. Vaiea; S. Lewis; K. Church; R. Shell; R. Potter; D. Griffin; E. Pozsgai; M. Hogan; L. Rodino-Klapac

EP.140
Long-term efficacy and safety of viltolarsen in the treatment of Duchenne muscular dystrophy
P. Clemens; A. Connolly; A. Harper; J. Mah; C. McDonald; V. Rao; E. Smith; C. Zaidman; T. Nakagawa; E. Hoffman

EP.141
Updated demographics and safety data from patients with nonsense mutation Duchenne muscular dystrophy receiving ataluren in the STRIDE Registry
F. Muntoni; F. Buccella; I. Desguerre; J. Kirschner; E. Mercuri; A. Nascimento Osorio; M. Tulinius; S. Johnson; C. Werner; A. Kristensen; J. Jiang; J. Li; P. Trifillis; C. Santos

EP.142
Age at loss of ambulation in patients with DMD from the STRIDE Registry and the CINRG Natural history study: a matched cohort analysis
E. Mercuri; F. Muntoni; F. Buccella; I. Desguerre; J. Kirschner; A. Nascimento Osorio; M. Tulinius; S. Johnson; C. Werner; A. Kristensen; J. Jiang; J. Li; P. Trifillis; C. Santos; C. McDonald

EP.143
Assessment of patients on Eteplirsen (Exondys 51) therapy using real world data
A. Zygmunt; I. Rybalsky; W. Chouteau; P. Horn; C. Tian

EP.144
Pulmonary function in patients with Duchenne muscular dystrophy from the STRIDE Registry and CINRG Natural History Study: a matched cohort analysis
M. Tulinius; F. Buccella; I. Desguerre; J. Kirschner; E. Mercuri; F. Muntoni; A. Nascimento Osorio; S. Johnson; C. Werner; A. Kristensen; J. Jiang; J. Li; P. Trifillis; C. Santos; C. McDonald

EP.145
Evaluation of DMD transcripts after golodirsen treatment of MyoD-converted fibroblasts from 4053-101 clinical trial patients
R. Rossi; M. Moore; S. Torelli; P. Ala; F. Catapano; R. Phadke; J. Morgan; J. Malhotra; F. Muntoni

EP.146
scAAV.U7snRNA-mediated therapy: prolonged dystrophin expression and muscle function correction in adult Dup2 mice
L. Gushchina; A. Bradley; T. Vetter; E. Frair; C. Bellinger; T. Simmons; N. Rohan; N. Wein; K. Flanigan

EP.147
2.5-years of vamorolone treatment in Duchenne muscular dystrophy: Results of an open label long-term extension
E. Hoffman; U. Dang; P. Clemens; H. Gordish-Dressman; B. Schwartz; L. Mengle-Gaw; M. Leinonen; E. Smith; D. Castro; N. Kuntz; R. Finkel; M. Tulinius; Y. Nevo; M. Ryan; R. Webster; J. van den Anker; L. Ward; J. Damsker; C. McDonald; M. Guglieri; J. Mah

EP.148
Monitoring treatment-benefit in patients with Duchenne muscular dystrophy treated with ataluren
E. Michael; K. Sofou; L. Wahlgren; A. Kroksmark; M. Tulinius

EP.149
Delay in Duchenne muscular dystrophy progression with eteplirsen: Longer time to loss of ambulation versus standard of care
J. Iff; G. Bungey; A. Paine; B. Han; H. Gordish-Dressman; E. Henricson; C. McDonald

EP.150
Casimersen in patients with Duchenne muscular dystrophy amenable to exon 45 skipping: Interim results from the Phase 3 ESSENCE trial
S. Iannaccone; H. Phan; V. Straub; F. Muntoni; E. Koenig; J. Malhotra; B. Han; E. Darton; E. Mercuri

EP.151
Evaluation of total binding antibodies against rAAVrh74 in patients with Duchenne muscular dystrophy
D. Griffin; N. Goedeker; C. Zaidman; S. Dharia

EP.152
ENDEAVOR: A gene delivery study to evaluate the safety of and expression from SRP-9001 in Duchenne muscular dystrophy
C. Zaidman; C. Proud; C. McDonald; K. Giblin; L. Collins; S. Wang; S. Upadhyay; S. Lewis; J. Malhotra; D. Griffin; R. Potter; M. Guridi; L. Rodino-Klapac; J. Mendell

DMD - biomarkers (EP.153-161)

EP.153
Platelet Derived Growth Factor-AA correlates with muscle function tests and quantitative muscle magnetic resonance in dystrophinopathies
A. Alonso-Jiménez; E. Fernández-Simón; D. Natera-De Benito; C. Ortez; C. Jimenez-Mallebrera; A. Nascimento; C. García; E. Montiel; I. Belmonte; I. Pedrosa; P. Piñol-Jurado; A. Carrasco-Rozas; X. Suárez-Calvet; C. Nuñez-Peralta; J. Llauger; P. Montesinos; J. Alonso-Pérez; E. Gallardo; I. Illa; J. Díaz-Manera

EP.154
Spatially mapping molecular markers of histopathological characteristics in Duchenne muscular dystrophy mouse models
L. Heezen; A. Mahfouz; A. Aartsma-Rus; P. Spitali

EP.155
A comparative myopathologic analysis reveals progressive muscle stem cells senescence in Duchenne muscular dystrophy (DMD)
N. Cardone; V. Taglietti; K. Kefi; P. Periou; C. Gitiaux; M. Traverso; C. Panicucci; S. Baratto; J. Authier; C. Bruno; P. Lafuste; C. Fiorillo; F. Relaix; E. Malfatti

EP.156
Expression of Periostin in DMD patients and mdx mice
C. Preusse; T. Ruck; D. Cengiz; A. von Moers; A. Hentschel; H. Lochmüller; U. Schara-Schmidt; A. Sickmann; A. Gangfuß; A. Förster; S. Meuth; H. Goebel; W. Stenzel; A. Roos

EP.157
Digital quantitative analysis of dystrophin and utrophin expression in muscle biopsies from female carriers of dystrophinopathy
F. Catapano; M. Ellis; S. Torelli; D. Chambers; T. Evangelista; F. Leturcq; D. Natera-de Benito; C. Jimenez-Mallebrera; C. Marini-Bettolo; R. Charlton; W. Stenzel; C. Dittmayer; A. Schänzer; D. Hilton; J. Lilleker; F. Roncaroli; A. Sarkozy; F. Muntoni; R. Phadke

EP.158
Robust spectral confocal microscopy dystrophin automatic quantification method
A. Anna Codina; M. Mònica Roldán; C. Carlos Ortez; L. Leslie Mataolonga; D. Daniel Cuadras; D. Daniel Natera; J. Juan Ramon Corbera; L. Laura Carrera; J. Jesica María Exposito; J. Jesús Márquez; C. Cecilia Jimenez; J. Josep Porta; A. Andres Nascimento; C. Cristina Jou

EP.159
Automated immunofluorescence analysis for sensitive and precise dystrophin quantification in muscle biopsies
T. Vetter; A. Bradley; E. Frair; S. Nicolau; K. Flanigan

EP.160
Quantification of dystrophin and mini dystrophin by mass spectrometry: Application in gene therapy development for Duchenne muscular dystrophy
V. Farrokhi; J. Walsh; J. Palandra; J. Brodfuehrer; T. Caiazzo; J. Owens; M. Binks; S. Neelakantan; F. Yong; H. Neubert

EP.161
A novel cell-based screening assay for the identification of utrophin-upregulating compounds
P. Soblechero-Martín; A. López-Martínez; V. Arechavala-Gomeza

FSHD (EP.162-173)

EP.162
Launch of a national registry for facioscapulohumeral muscular dystrophy in Japan
T. Matsumura; H. Nakamura; I. Nishino; M. Sasaki-Honda; T. Suzuki; H. Komaki

EP.163
Characterising FSHD and supporting national and international research projects: Eight years of the UK FSHD patient registry
B. Porter; R. Orrell; A. Graham; S. Watt; P. Lunt; F. Norwood; M. Roberts; T. Willis; E. Matthews; R. Muni-Lofra; C. Marini-Bettolo

EP.164
The assessment of cognitive and psychological status in an adult-onset FSHD sample: a pilot study
F. Torri; G. Spadoni; C. Bettio; R. Tupler; G. Ricci; G. Siciliano

EP.165
Systemic delivery of antisense oligonucleotides targeting DUX4; a promising therapeutic strategy for facioscapulohumeral muscular dystrophy
L. Bouwman; B. den Hamer; A. van den Heuvel; M. Franken; M. Jackson; C. Dwyer; S. Tapscott; F. Rigo; S. van der Maarel; J. de Greef

EP.166
Respiratory function in facioscapulohumeral muscular dystrophy: How does it progress in five years?
S Teeselink; S. Vincenten; N. Voermans; J. Groothuis; J. Doorduin; P. Wijkstra; C. Horlings; B. van Engelen; K. Mul

EP.167
Longitudinal clinical course of early onset facioscapulohumeral muscular dystrophy
K. Tabata; E. Takeshita; H. Komaki; N. Sumitomo; Y. Shimizu-Motohashi; T. Saito; A. Ishiyama; E. Nakagawa; I. Nishino; M. Sasaki

EP.168
An open-label study of Losmapimod to evaluate the safe subjects with FSHD1
J. Kools; N. Voermans; K. Mul; B. van Engelen; L. Ronco; J. Jiang; J. Shoskes; M. Mellion; K. Marshall; D. Cadavid

EP.169
Interaction between mesenchymal stem cells and myoblasts in pathophysiology of FSHD
O. Serbina; Y. Vassetzky; E. Kiseleva

EP.170
Use of snRNA-seq to characterize the skeletal muscle microenvironment during pathogenesis in FSHD
A. Raman; A. Accorsi; B. Riehle; M. Mellion; L. Ronco; A. Rojas; C. Moxham

EP.171
Radiological and circulating biomarkers in Facioscapulohumeral muscular dystrophy: a longitudinal study
S. Bortolani; N. Mosca; M. Monforte; T. Tartaglione; E. Ragozzino; E. Ricci; G. Tasca

EP.172
Atypical phenotypes and diagnostic delay in FSHD
A. Alobaidan; E. Bugiardini; J. Morrow; S. Shah; S. MacDonald; A. Carr; P. Machado; R. Quinlivan; C. Turner; T. Yousry; M. Parton; M. Hanna

EP.173
Facioscapulohumeral muscular dystrophy myopathologic dissection in 22 patients shows highly variable morphologic findings
L. Hubregtse; K. Bouman; S. Lassche; B. Küsters; B. Periou; J. Authier; N. Voermans; E. Malfatti

LGMD (EP.174-195)

EP.174
Transportinopathy the link between LGMD and HIV-1: Cytokine analysis of LGMD-D2 patients from two different families
C. Angelini; M. Lopez Huertas; S. Rodriguez-Mora; J. Alcami

EP.175
Natural history and phenotypic spectrum of LGMD-D1: Implications for clinical Trials
S. Robinson; S. Poelker; M. Seiffert; C. Weihl; A. Findlay

EP.176
DNAJB6 isoform specific knockdown: Mechanistic insights and therapeutic potential for LGMDD1
A. Findlay; M. Paing; J. Daw; S. Pittman; R. Bengoechea; T. Chou; C. Weih

EP.177
Clinical and genetic spectrum of a large cohort of delta-sarcoglycan muscular dystrophy
J. Alonso-Pérez; L. González-Quereda; C. Bruno; C. Panicucci; A. Alavi; E. Zanoteli; N. Muelas; J. Vilchez; E. Dourado; N. Kadem; M. Umair; M. Guglierir; C. Marini-Bettolo; V. Straub; J. Diaz- Manera

EP.178
Global FKRP Registry - the research database for limb girdle muscular dystrophy R9 (2I)
L. Murphy; L. Alfano; K. Brazzo; N. Johnson; J. Laurent; K. Mathews; S. Thiele; J. Vissing; M. Walter; L. Woods; V. Straub

EP.179
Substantial improvement of shoulder function with a new physiotherapy approach in children with Limb girdle muscular dystrophy
M. Pelsma; J. Ijspeert; Y. Veenhuizen; S. Houwen - van Opstal; H. Braakman

EP.180
Gait patterns in patients with dysferlinopathy when walking barefoot, in socks and shod: Preliminary findings from the COSII study
M. James; L. Alcock; V. Straub; A. Mayhew

EP.181
Gait patterns in patients with dysferlinopathy when walking at preferred and fast walking speeds: Preliminary findings from the COSII study
M. James; L. Alcock; V. Straub; A. Mayhew

EP.182
Quality of life in dysferlinopathy can be good despite poor function
U. Moore; M. James; S. Spuler; J. Day; K. Jones; D. Bharucha-Goebel; E. Salort-Campana; A. Pestronk; M. Walter; C. Paradas; T. Stojkovic; M. Mori Yoshimura; E. Bravver; E. Pegoraro; J. Mendell; K. Bushby; V. Straub; A. Mayhew

EP.183
Mutation profiling and immunohistochemical characterization of Japanese patients with sarcoglycanopathy
Y. Saito; S. Hayashi; S. Noguchi; I. Nishino

EP.184
Treat NMD LGMD global registry network development
S. Segovia; A. Ambrosini; C. Campbell; J. Diaz-Manera; S. Study Group; M. Guglieri

EP.185
Safety -sarcoglycan expression, and functional outcomes from systemic gene transfer of rAAVrh74.MHCK7.hSGCB in LGMD2E/R4
L. Rodino-Klapac; E. Pozsgai; S. Lewis; D. Griffin; A. Meadows; K. Lehman; K. Church; N. Reash; M. Iammarino; L. Lowes; E. Koenig; S. Neuhaus; X. Li; J. Mendell

EP.186
Muscle inflammatory infiltrates composition and MHCI expression in alpha and gammasarcoglycanopathies
C. Panicucci; S. Baratto; R. Lizzia; P. Tonin; A. D'Amico; G. Tasca; M. Traverso; C. Fiorillo; S. Previtali; E. Pegoraro; C. Bruno

EP.187
Clinical and genetic characteristics of LGMDR1 patients in the north of England
K. Waller; M. Schiava; C. Marini-Bettolo; V. Straub; A. Mayhew; R. Muni-Lofra; M. James; J. Hudson; J. Diaz-Manera; M. Guglieri

EP.188
Elevation of fast but not slow muscle fiber injury biomarkers after exercise in Becker and LGMDR9 muscular dystrophies
M. Stemmerik; N. Andersen; A. Russell; B. Barthel; M. Madden; S. Skriver; J. Vissing

EP.189
Two-years prospective natural history study in 29 adult LGMDR12 patients: Clinical and radiological outcome measures
B. De Wel; L. Huysmans; R. Peeters; A. D'Hondt; V. Goosens; S. Ghysels; K. Byloos; G. Putzeys; J. De Bleecker; F. Maes; P. Dupont; K. Claeys

EP.190
Treatment of the Sgca-null mouse with a second-generation tyrosine kinase inhibitor (TKI)
P. Piñol-Jurado; X. Suárez-Calvet; M. Borrell-Pagès; E. Fernández-Simón; V. López Díaz; E. Gallardo; L. Badimon; J. Díaz-Manera

EP.191
Selective P2X7 antagonism to dampen inflammation in alpha-sarcoglycan-deficient muscular dystrophy
C. Panicucci; L. Raffaghello; E. Principi; S. Baratto; S. Pintus; F. Antonini; G. Del Zotto; A. Benzi; S. Bruzzone; C. Minetti; E. Gazzerro; C. Bruno

EP.192
Predicting loss of ambulation in LGMDR9
L. Coffey; M. Zimmerman; S. Mockler; K. Mathews

EP.193
Prospective clinical trial readiness in LGMDR9 FKRP-related muscular dystrophy: A GRASP consortium study
M. Iammarino; L. Alfano; M. James; T. Mozaffar; K. Mathews; C. Weihl; D. Leung; J. Statland; P. Kang; M. Wicklund; L. Lowes; J. Vissing; J. Diaz Manera; V. Ganesh; B. Holmberg; E. DeSpain; K. Bates; D. Sproule; N. Johnson; GRASP Consortium

EP.194
Using qualitative interviews to select movement tasks in the development of the limb-girdle video assessment
S. Bolding; M. Leffer; K. Davis; A. Burns; M. Contesse

EP.195
The most common CAPN3 disease causing variant in nothern-eastern europe population or controversial coincidence
B. Lace; J. Stavusis; I. Micule; V. Kenina; G. Taurina; A. Zdanovica; M. Mroczek; B. Burnyte; I. Inaskina

Pompe disease (EP.196-203)

EP.196
Cipaglucosidase alfa-miglustat versus alglucosidase alfa-placebo in late-onset Pompe disease (LOPD): PROPEL study subgroup analyses
B. Schoser; D. Bratkovic; B. Byrne; P. Laforet; T. Mozzaffar; J. Diaz-Manera; A. van der Ploeg; M. Roberts; A. Toscano; H. Jiang; S. Sitaraman; S. Kuchipudi; M. Goldman; J. Castelli; P. Kishnani

EP.197
Antibodies against recombinant human alpha-glucosidase do not affect clinical outcome in childhood onset Pompe disease
H. van Kooten; I. Ditters; M. Hoogeveen-Westerveld; J. van den Hout; P. van Doorn; W. Pijnappel; A. van der Ploeg; N. van der Beek

EP.198
Large-scale validation of the Rasch-built Pompe activity scale (R-PAct) across twelve countries
H. van Kooten; M. Horton; S. Wenninger; B. Schoser; C. Lefeuvre; P. Laforêt; S. Segovia; J. Díaz Manera; K. Claeys; T. Mongini; O. Musumeci; A. Toscano; T. Hundsberger; E. Brusse; I. Merkies; P. van Doorn; A. van der Ploeg; N. van der Beek; R-PAct Study Group

EP.199
Spanish Pompe registry: Analysis of the 100 patients included
D. Reyes-Leiva; A. Nascimento; N. Muelas; C. Domínguez-González; C. Paradas; M. Olivé; J. Grau; M. Barba-Romero; M. Gómez-Caravaca; J. Diaz-Manera; S. Spanish Pompe Study Group

EP.200
Patient relevant change in forced vital capacity % predicted in late onset Pompe disease (LOPD) in the COMET trial
K. Berger; P. DasMahapatra; E. Baranowski; K. An Haack; S. Sparks; N. Thibault; C. Gwaltney; C. Ivanescu; G. Flore; L. Pollissard; A. Hamed

EP.201
Living with Pompe disease in the UK: Characterizing the patient journey and burden on physical, emotional and social quality of life
A. Muir; K. Odedra; N. Johnson; L. Bashorum; A. Patmore; P. Slade; J. Lewthwaite

EP.202
Safety of home-based infusion of alglucosidase alfa in late-onset Pompe disease: 13 years of experience from the Erasmus MC university medical center
I. Ditters; H. van Kooten; N. van der Beek; J. van den Hout; H. Huidekoper; A. van der Ploeg

EP.203
Mobility, usual activities and EQ-5D visual analogue score improvement with Avalglucosidase alfa in Late-onset Pompe disease during the COMET trial
L. Pollissard; P. DasMahapatra; E. Baranowski; K. An Haack; T. Zhou; N. Thibault; J. Msihid; A. Hamed

Metabolic myopathies (EP.204-210)

EP.204
No effect of resveratrol on exercise capacity in patients with fatty acid oxidation disorders
J. Storgaard; N. Loekken; K. Madsen; P. Laforêt; N. Voermans; v. Gerrit; J. Vissing; M. Oerngreen

EP.205
High prevalence of paraspinal involvement in McArdle disease
R. Álvarez-Velasco; C. Nuñez-Peralta; J. Alonso-Pérez; D. Reyes-Leiva; E. Gallardo; J. Llauger; J. Díaz-Manera; I. Illa; M. Olivé

EP.206
No effect of oral ketone ester supplementatin on exercise capacity in patients with McArdle disease: a randomized placebo-controlled cross-over study
N. Løkken; J. Storgaard; K. Revsbech; N. Voermans; G. Van Hall; J. Vissing; M. Ørngreen

EP.207
Clinical findings of late-onset long-chain fatty acid oxidation disorders overlap with neuromuscular conditions in a sponsored genetic testing program
D. Marsden; V. Rangel Miller; T. Chettiath; B. Johnson; S. Sarafrazi; A. Willcock; N. Miller

EP.208
Natural history of McArdle disease: A single centre study of a cohort of 220 patients
C. Pizzamiglio; O. Mahroo; K. Khan; M. Patasin; R. Quinlivan

EP.209
Automated image-analysis method showed positive correlation between body mass index and skeletal muscle fiber size in human
B. Periou; C. Hou; X. Decrouy; E. Malfatti; F. Relaix; J. Authier

EP.210
A rare cause of exercise-induced rhabdomyolysis: Very long chain acyl-CoA dehydrogenase deficiency
N. Yeniay Sut; M. Yildirim; O. Bektas; S. Sahin; S. Teber

Mitochondrial diseases (EP.211-218)

EP.211
The analysis of mitochondrial respiration and membrane potential in skeletal muscles of Huntington disease patients
S. Kopishinskaia; P. Pchelin; M. Korotysh; T. Kovaleva; S. Svetozarskiy; S. Nikitin; I. Mukhina

EP.212
Preferent diaphragmatic involvement in TK2 deficiency: an autopsy case study
C. Domínguez-González; S. Laine-Menéndez; A. Delmiro; I. García-Consuegra; M. Fernández-de la Torre; A. Hernández-Laín; J. Sayas; C. de Fuenmayor; M. Martin; M. Morán

EP.213
Plasma lactate responses during and after submaximal handgrip exercise are not diagnostically helpful in mitochondrial myopathy
N. Loekken; S. Vinther Skriver; T. Khawajazada; J. Storgaard; J. Vissing

EP.214
Early onset hereditary neuropathies related to mitochondrial dynamics in pediatric patients
L. Carrera; L. Cantarero; G. Nolasco; J. Pijuan; D. Natera; B. Estevez; J. Expósito; J. Colomer; C. Ortez; F. Palau; J. Hoenicka; A. Nascimento

EP.215
Use of self-directed home video capture for assessing muscle weakness in primary mitochondrial myopathy: A feasibility study
A. Burns; M. Leffler; A. Sapp; A. Allred; E. Kelly; S. Doyle; T. Uz; A. Karaa

EP.216
Predictors of clinical outcome and survival in mitochondrial disease patients in intensive care unit
H. Lee; Y. Lee

EP.217
Comparison between pediatric and adult onset MELAS: three autopsy cases
A. Ishii; S. Ouchi; R. Matsuoka; A. Tamaoka; M. Noguchi

EP.218
Adult-onset Leigh syndrome / lactic aidosis and stroke-like episodes overlap syndrome due to m9176T>C mutation with cortical involvement
M. Lee; J. Lee; H. Lee

Channelopathies and related disorders (EP.220-228)

EP.220
Repetitive nerve stimulation decrement as a discrimination tool for hereditary myotonic syndromes
S. Kurbatov; E. Ivanova; N. Ryadninskaya; I. Kuznetsova; V. Fedotov; S. Bardakov; V. Zabnenkova; O. Shchagina; A. Polyakov

EP.221
Diagnostic delay and atypical phenotypes in a French cohort of Andersen-Tawil syndrome
R. Villar Quiles; G. Tredez; D. Sternberg; N. Romero; T. Evangelista; P. Laforêt; P. Cintas; G. Sole; S. Sacconi; S. Bendahhou; M. Arzel-Hezode; E. Fournier; B. Fontaine; T. Stojkovic; S. Vicart

EP.222
The Impact of non-dystrophic Myotonia on patients and caregivers quality of life (IMPACT) survey results
J. Diaz-Manera; C. Hewamadduma; G. Meola; F. Montagnese; U. Nowak; R. Pleticha; S. Sacconi; P. von Gallwitz; A. Zozulya-Weidenfeller

EP.223
Development of new riluzole analogs as use-dependent sodium channel blockers for the treatment of non-dystrophic myotonias
C. Altamura; A. Farinato; M. Cavalluzzi; N. Tarantino; M. Carratù; S. Pierno; G. Lentini; J. Desaphy

EP.224
Repurposing of niflumic acid for ClC-1 mutation-driven pharmacological approach in myotonia congenita
C. Altamura; E. Conte; G. Camerino; F. Girolamo; C. Campanale; M. Carratù; P. Imbrici; J. Desaphy

EP.225
Autosomal dominant early onset vacuolar myopathy with granular material, without periodic paralysis, associated with c.1583C>A CACNA1S gene mutation
M. Bisciglia; H. Kadhim; S. Lecomte; I. Vandernoot; L. Desmyter; G. Remiche

EP.226
Muscle fat fraction, cross-sectional area and contractility in patient with skeletal muscle sodium channel disorders
J. Pedersen; L. Jakobsen; S. Skriver; M. Stemmerik; J. Vissing

EP.227
A novel compound heterozygous mutation in the CLCN1 gene in a patient with congenital myotonia
A. Kartal; M. Yildirim; O. Bektas; C. Ucar; S. Teber

EP.228
Efficacy and safety of mexiletine in non-dystrophic myotonias: a randomized, double-blind, placebo-controlled, cross-over study
S. Vicart; J. Franques; F. Bouhour; A. Magot; Y. Pereon; S. Sacconi; A. Nadaj-Pakleza; A. Behin; N. Zahr; M. Hezode; E. Fournier; C. Payan; L. Lacomblez; B. Fontaine

Myotonic dystrophy (EP.229-238)

EP.229
Characterising myotonic dystrophy (DM) and supporting national and international research projects: Nine years of the UK DM patient registry
B. Porter; C. Turner; D. Monckton; M. Bowler; M. Roberts; M. Rogers; M. Rose; R. Orrell; J. Donachie; D. Williams; M. Hamilton; C. Hewamadduma; J. Sodhi; C. Marini-Bettolo

EP.230
Comparing patient-reported swallowing symptoms and videofluoroscopy findings in patients with Myotonic dystrophy type 1 (DM1)
J. Allen; G. Ramdharry; R. Astin; C. Turner; C. Smith

EP.231
Predictors of dysphagia and aspiration in Myotonic dystrophy type 1 (DM1)
J. Allen; G. Ramdharry; R. Astin; C. Turner; C. Smith

EP.232
Understanding dysphagia in patients living with Myotonic dystrophy type 1 (DM1)
J. Allen; G. Ramdharry; R. Astin; C. Turner; C. Smith

EP.233
The FORCE™ platform achieves durable knockdown of toxic human nuclear DMPK RNA and correction of splicing in the hTfR1|DMSXL mouse model
S. Zanotti; T. Picariello; N. Hsia; T. Weeden; R. Russo; L. Schlaefke; M. Yao; A. Wen; S. Hildebrand; J. Najim; Q. Qui; M. Quinn; M. Qatanani; R. Subramanian; O. Beskrovnaya

EP.234
Liver dysfunction and dyslipidemia in myotonic dystrophy type 1: A cross-sectional study using the national registry of Japan
M. Hama; R. Horie; T. Kubota; T. Matsumura; E. Kimura; H. Nakamura; M. Takahashi; H. Takada

EP.235
MyoScreen, a robust muscle-based assay platform validated for screening pharmacologicals against Myotonic dystrophy
M. Flaender; J. Young; E. Duchemin-Pelletier; O. Lorintiu; L. Compere; T. Champetier; E. Ventre

EP.236
Clinical improvement of DM1 patients after cognitive behavioural therapy is reflected in diseaserelevant gene expression changes in blood
R. van Cruchten; B. van Engelen; P. 't Hoen; D. van As; The OPTIMISTIC consortium; The ReCognitION consortium

EP.237
A systematic review and meta-analysis of cancer risk in myotonic dystrophy: Consistent with a general increased risk of cancer
K. McConville; C. McShane; R. Fallis; K. McConville; L. Anderson

EP.238
Masseter muscle volume as a disease marker in adult-onset myotonic dystrophy type 1
A. Oliwa; C. Hocking; M. Hamilton; J. McLean; S. Cumming; B. Ballantyne; R. Jampana; The Scottish DM1 Consortium; C. Longman; D. Monckton; M. Farrugia

Motor neuron disorders and neuropathies (EP.239-247b)

EP.239
Charcot Marie Tooth disease caused by compound heterozygous GDAP1 variants: A case report
A. Zygmunt; C. Thomas; C. Tian

EP.240
Charcot-Marie-Tooth disease, clinical and genetic features in a pediatric canadian cohort
A. Alemán; N. Alharthi; K. Amburgey; J. Vajsar; G. Yoon; M. Bouchard; J. Dowling; H. Gonorazky

EP.241
A compound heterozygous mutation in the B4GALNT1 gene causes axonal Charcot-Marie-Tooth disease
H. Park; S. Lee; Y. Choi

EP.242
Fraction ratio of muscle thickness as ultrasonographic biomarker in patients with amyotrophic lateral sclerosis
E. Fages Caravaca; J. Ríos-Diaz; J. Martinez Payá; A. Rios -Cortés; E. del Baño Aledo; P. García Torres; O. Meca Birlanga; E. Kanh Mesía

EP.243
Young-onset SOD1-ALS Caused by a novel homozygous mutation, a family report
N. Fahmy; K. Müller; P. Andersen; S. Marklund; A. Ludolph; N. Hamdi

EP.244
Clinical characteristics of SOD1 and c9ORF72 positive ALS: Findings from the Canadian Neuromuscular Disease Registry
V. Hodgkinson-Brechenmacher; J. Lounsberry; A. Abrahao; T. Benstead; A. Breiner; H. Briemberg; A. Genge; I. Grant; S. Kalra; A. Marrero; R. Massie; G. Matte; C. O'Connell; G. Pfeffer; K. Schellenberg; C. Shoesmith; S. Taylor; A. Izenberg; W. Johnston; L. Korngut

EP.245
Characterization of a novel form of ALS associated with changes in the sphingolipid metabolism
M. Lone; T. Hornemann

EP.246
Kennedy disease in two sisters with biallelic CAG expansions of the androgen receptor gene
K. Müller; M. Nebuchenykh; Ø. Nilssen; M. Van Ghelue; S. Løseth; C. Jonsrud; G. Hoem; K. Arntzen

EP.247a
Debilitating oste-ochondral manifestations in a patient with congenital insensitivity to pain with anhidrosis (CIPA) due to NTRK1 mutation
X. Li; V. Li; J. Sun; Y. Jiang; S. Chan; E. Kuong

EP.247b
Enterovirus infection with isolated progressive motor neuron disease
N. Alharthi; H. Qashqari; A. Aleman; H. Gonorazky

SMA clinical data (EP.248-254a)

EP.248
Longitudinal developmental profile of newborns and toddlers treated for spinal muscular atrophy
M. Ngawa; F. Dal Farra; A. Marinescu; L. Servais

EP.249
ANCHOVY: A retrospective cohort study of the natural history of type 1 spinal muscular atrophy (SMA) using medical record data
E. Zanoteli; C. Cances; D. Vlodavets; G. Comi; R. Masson; M. Mazurkiewicz-Bełdzińska; K. Saito; A. Dodman; M. El-Khairi; K. Gorni; I. Gravestock; J. Hoffart; R. Scalco; B. Darras

EP.250
Description of cardiac involvement in 5q SMA pediatric patients
S. van Otterdijk; H. Kölbel; A. Schönecker; L. Modler; A. Della Marina; U. Neudorf; U. Schara

EP.251
Feeding and swallowing problems in SMA: a pilot observational study testing the usefulness of the IOPI pressure measurement system
S. Ben Mechri; N. Deconinck

EP.252
Minimally invasive fusionless surgery for scoliosis in spinal muscular atrophy: Long-term follow up results in a series of 59 children
S. Quijano-Roy; M. Gaume; E. Saudeau; M. Gomez-Garcia de la Banda; V. Azzi-Salameh; B. Mbieleu; D. Verollet; C. Barnerias; A. Benezit; I. Dabaj; A. Essid; I. Doehring; H. Amthor; A. Oulhissane; R. Carlier; I. Desguerre; J. Bergounioux; C. Glorion; L. Miladi

EP.253
The international spinal muscular atrophy registry (ISMAR): Baseline characteristics
J. Montes; G. Coratti; M. Scoto; J. Balashkin; M. Pera; S. Samsuddin; W. Martens; A. Bozzardi; A. Rodriguez; M. Civitello; M. Madden; B. Lings; A. Rohwer; S. Hall; Z. Zolkipli; J. Day; B. Darras; D. De Vivo; F. Muntoni; R. Finkel; E. Mercuri

EP.254
Newborn screening for spinal muscular atrophy in the world today
T. Dangouloff; E. Vrscaj; L. Servais; D. Osredkar

EP.254a
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

SMA outcome measures and registries (EP.255-267)

EP.255
Development of CHOP-INTEND test subgroups in spinal muscular atrophy type 1
A. Erden; E. Acar Arslan; M. Emirzeoglu; N. Yıldız; E. Gülnur

EP.256
Investigation of motor functions items in children with spinal muscular atrophy type 2
A. Erden; E. Acar Arslan; M. Emirzeoğlu; N. Yıldız; E. Gülnur

EP.257
Prediction of loss of ambulation in spinal muscular atrophy type IIIA using ten-meter walk test results
A. Wolfe; C. Rye; M. Scoto; M. Main; F. Muntoni

EP.258
Health care resource utilization in pediatric patients with spinal muscular atrophy: Interim analysis of a US retrospective chart review study
O. Dabbous; M. Yang; M. Georgieva; W. Toro; N. LaMarca; A. Patel; N. Minkoff; C. Carley; J. Zhu; A. Anderson; E. Wu

EP.259
CuidAME: a new registry for longitudinal data collection of Spanish SMA patients
S. Segovia; J. Exposito; C. Ñungo; J. Vazquez; I. Pitarch; J. Caballero; S. Pascual; A. Moreno; E. Martinez; S. SMA Study Group; A. Nascimento

EP.260
Real-world data on access to SoC for the SMA population in the UK
R. Muni-Lofra; L. Murphy; K. Adcock; M. Farrugia; J. Irwin; J. Lilleker; J. McConville; A. Merrison; M. Parton; L. Ryburn; M. Scoto; C. Marini-Bettolo; A. Mayhew

EP.261
Improving the implementation of spinal muscular atrophy (SMA) standards of care (SoC) internationally – the SMA SoC for all platform
C. Lilien; G. Baranello; G. Coratti; L. Edel; O. Germanenko; M. Krstic; M. Mazurkiewicz-Bełdzińska; S. Ray; A. Shatillo; J. Taytard; D. Vlodavets; C. Vuillerot; L. Cruz; G. Tachibana; C. Viljoen; L. Servais

EP.262
The Canadian neuromuscular disease registry: A national spinal muscular atrophy (SMA) registry for real world evidence
V. Hodgkinson-Brechenmacher; M. Oskoui; B. Brais; C. Campbell; H. Gonorazky; J. Lounsberry; A. MacKenzie; H. McMillan; J. Vajsar; L. Korngut; C. CNDR Investigator Network

EP.263
TREAT-NMD core dataset for SMA – An important tool for post marketing surveillance
J. Das; V. Hodgkinson; M. Rodrigues; J. Bullivant; H. Walker; V. Straub; C. Campbell; M. Guglieri; A. Ambrosini

EP.264
Evaluating perceived fatigue within an adult SMA population
L. Belter; J. Jarecki

EP.265
Longitudinal follow-up of adult patients with spinal muscular atrophy undergoing nusinersen treatment using an innovative magneto-inertial device
M. Poleur; M. Annoussamy; D. Eggenspieler; S. Delstanche; A. Daron; L. Servais

EP.266
Assessment of current best-practices to triage and expedite incoming referrals for the evaluation of spinal muscular atrophy
M. Curry; R. Cruz; L. Belter; M. Schroth; J. Jarecki

EP.267
"Registre- SMA France": one year after launching the National French registry on SMA
M. Gomez Garcia de la Banda; L. Grimaldi-Bensouda; J. Urtizberea; A. Behin; C. Vuillerot; P. Saugier-Veber; F. Audic; C. Barnerias; C. Cances; E. Campana-Salort; C. Espil; P. Laforet; V. Laugel; Y. Pereon; S. Sacconi; T. Stojkovic; C. Tard; B. Chabrol; I. Desguerre; S. Quijano-Roy

SMA - treatment (EP.268-294)

EP.268
In vivo effects of Sarconeos (API BIO101) on a mouse model of severe spinal muscular atrophy
C. Bezier; P. Dilda; R. Lafont; S. Veillet; F. Charbonnier; M. Latil; O. Biondi

EP.269
Scientific rationale for a higher dose of nusinersen
M. Ryan; R. Finkel; S. Pascual Pascual; J. Day; E. Mercuri; D. De Vivo; J. Montes; J. Gurgel-Giannetti; D. MacCannell; Z. Berger

EP.270
Proteins associated with the sarcomere and costamere in hearts are dysregulated in two mouse models of spinal muscular atrophy
S. Owen; D. Šoltić; S. Synowsky; E. Crompton; R. Yáñez-Muñoz; B. Schneider; H. Shorrock; S. Shirran; T. Gillingwater; M. Bowerman; H. Fuller

EP.271
Part A results from the ongoing DEVOTE study to explore higher-dose nusinersen in SMA
E. Mercuri; R. Finkel; J. Day; S. Pascual Pascual; M. Ryan; D. De Vivo; J. Montes; J. Gurgel- Giannetti; G. Gambino; R. Nuzzo; C. Makepeace; S. Garafalo; Z. Berger

EP.272
Motor function and safety profile following gene replacement therapy with Omnasemnogene-Abeparvovec- report of 10 patients
J. Friese; J. Kirschner

EP.273
Long-term follow-up (LTFU) of onasemnogene abeparvovec gene therapy in spinal muscular atrophy (SMA)
J. Mendell; R. Finkel; E. Mercuri; K. Strauss; J. Day; A. Kleyn; D. Chand; S. Tauscher-Wisniewski; M. Meriggioli

EP.274
Onasemnogene Abeparvovec Gene Therapy in Presymptomatic spinal muscular atrophy (SMA): SPR1NT study update in children with 3 Copies of SMN2
K. Strauss; F. Muntoni; M. Farrar; K. Saito; J. Mendell; L. Servais; H. McMillan; K. Swoboda; J. Kwon; C. Zaidman; C. Chiriboga; S. Iannaccone; J. Krueger; J. Parsons; P. Shieh; S. Kavanagh; D. Chand; S. Tauscher-Wisniewski; T. Macek

EP.275
Pharmacokinetics and safety of treatment with paracetamol in children and adults with spinal muscular atrophy
M. Naume; K. Revsbeck; T. Krag; J. Vissing; M. Møller; C. Høi-Hansen; A. Born; H. Holst; S. Haslund-Krog; P. Jensen; M. Ørngreen

EP.276
Bulbar function evolution in SMA type 1 children treated with nusinersen
H. Weststrate; G. Stimpson; L. Thomas; M. Scoto; A. Manzur; P. Munot; M. Main; E. Johnson; F. Muntoni; G. Baranello; E. Conway

EP.277
Insights into the potential pharmacological effects of apitegromab in health and disease: Data from preclinical and clinical studies
A. Place; D. Barrett; S. Cote; G. Nomikos; G. Song; S. Bilic; A. Kalra; M. Sadanowicz; J. O'Neil; R. Iarrobino; N. Kertesz; Y. Chyung

EP.278
FIREFISH Parts 1 and 2: 24-month safety and efficacy of risdiplam in type 1 spinal muscular atrophy (SMA)
R. Masson; O. Boespflug-Tanguy; B. Darras; J. Day; N. Deconinck; A. Klein; M. Mazurkiewicz-Bełdzińska; E. Mercuri; K. Rose; L. Servais; D. Vlodavets; H. Xiong; E. Zanoteli; A. Dodman; M. El-Khairi; E. Gaki; M. Gerber; K. Gorni; H. Kletzl; G. Baranello

EP.279
JEWELFISH: Safety, pharmacodynamic and exploratory efficacy data in non-naïve patients with spinal muscular atrophy (SMA) receiving risdiplam
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 nonambulant 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 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 TREATNMD 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 SPG15
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. 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.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

Late Breaking Posters - (LBP 1–25)

LBP.01
Tandem duplication in the DMD gene in Labrador Retrievers with a mild clinical phenotype
G.D. Shelton; K.M. Minor; N.M. Vieira; L.M. Kunkel; S.G. Friedenberg: J.N. Cullen; L.T. Guo; M. Zatz; J.R. Mickelson

LBP.02
Allele-specific disruption of a dominant COL6A1 mutation restores collagen VI in the extracellular matrix of patients' fibroblasts
A. López-Márquez; M. Morín; S. Fernández-Peñalver; C. Badosa; A. Hernández-Delgado; D. Natera-de Benito; C. Ortez; A. Nascimento; D. Grinberg; S. Balcells; M. Roldán; M.Á. Moreno-Pelayo; C. Jiménez-Mallebrera

LBP.03
Linker Protein repair of Lama2-deficiency by AAV somatic gene therapy
P. Yurchenco; K. McKee

LBP.04
Perivascular and endomysial macrophages expressing VEGF and CXCL12 promote angiogenesis in anti-HMGCR immune-mediated necrotizing myopathy
F. Girolamo

LBP.05
Notch and PDGF signalling regulate differentiation and migration of human iPS cell-derived myogenic progenitors: implication for advanced skeletal muscle therapies
G. Ferrari; S. Choi; L. Moyle; K. Mackinlay; N. Naouar; C. Wells; F. Muntoni; F.S. Tedesco

LBP.06
ANTXR2 as a novel player in skeletal muscle stem cells
S. Metti; L.J. Bracq; F.G van der Goot2, P. Bonaldo

LBP.07
The discovery of an epigenetic signature for Duchenne muscular dystrophy using genome-wide DNA methylation analysis
L. Schreyer; J. Reilly; J. Kerkhof; M.A. Levy; J. Hu; M. Hnaini; B. Sadikovic; C. Campbell

LBP.08
Vamorolone versus corticosteroid real-world experience: Comparisons of 2-year treatment period with NorthStar UK Network and CINRG Duchenne natural history study
J. Mah; P. Clemens; M. Guglieri; E. Smith; R. Finkel; M. Tulinius; Y. Nevo; M. Ryan; R. Webster; D. Castro; N. Kuntz; C. McDonald; G. Stimpson; D. Ridout; V. Ayyar-Gupta; A. Manzur; F. Muntoni; L. Ward; H. Gordish-Dressman; E. Hoffman; U. Dang

LBP.09
Thigh muscle phenotyping for limb girdle muscular dystrophies R1 and R2 with quantitative fat fractions and radiomics
H. Zhong; J. Li; D. Yue; M. Gao; J. Xi; W. Zhu; J. Dong; Y. Chen; L. Luo; X. Yu; S. Tan; X. Zheng; J. Díaz-Manera; C. Zhao; Q. Ke; Z. Liang; S. Luo

LBP.10
Apitegromab in SMA: an analysis of multiple endpoints and PD relationships to efficacy in the TOPAZ trial
T. Crawford; B. Darras; J. Day; G. Nomikos; G. Song; A. Place; D. Barrett; S. Bilic; J. O'Neil; N. Kertesz; S. Cote; J. Patel; Y. Chyung

LBP.11
Vamorolone versus placebo and prednisone in Duchenne muscular dystrophy: Results from a 24-week double-blind randomized trial
M. Guglieri; P. Clemens; S. Perlman; E. Smith; I. Horrocks; R. Finkel; M. Monduy; N. Deconinck; N. Goemans; J. Haberlova; M. Katsalouli; S. Spinty; A.M. Childs; G. Baranello; V. Straub; J. Vilchez-Padilla; A. Nascimento; J. Damsker; M. James; E. Hoffman

LBP.12
Brain Dp140 alters glutamatergic transmission and social behaviour in Duchenne muscular dystrophy mouse model
Y. Hashimoto; H. Kuniishi; K. Sakai; Y. Fukushima; X. Du; K. Yamashiro; K. Hori; N. Motohashi; M. Imamura; M. Hoshino; M. Yamada; T. Araki; H. Sakagami; S. Takeda; K. Itaka; N. Ichinohe; F. Muntoni; M. Sekiguchi; Y. Aoki

LBP.13
ASPIRO gene replacement therapy trial with resamirigene bilparvovec in XLMTM: pathologic findings in three deceased study participants
M. Lawlor; P. Shieh; C.G. Bönnemann; W. Müller-Felber; J.J. Dowling; B. Schoser; M. Margeta; H. Meng; A.R. Foley; D. N. Saade; A. Blaschek; S. Prasad; S. Rico; M. Murtagh; N. Bachtell; W. Miller; ASPIRO Study Group

LBP.14
Objective evaluation of clinical actionability for genes involved in myopathies: 34 promising genes
M. Vecten; E. Pion; R.J. Morales; D. Sternberg; J. Rendu; T. Stojkovic; C.A. Bourdain; C. Metay; I. Richard; L. Villard; M. Cerino; M. Milh; S. Gorokhova; N. Levy; X. Martin; G. Bonne; V. Biancalana; F. Petit; A. Perrin; P. Laforet; M. Bartoli; M. Cossee; M. Krahn

LBP.15
Safety, pharmacokinetics, and preliminary efficacy of Rycal S 48168 (ARM210) for RYR1-related myopathies: a phase one, open-label dose-escalation trial
J.J. Todd; T. Lawal; I. Chrismer; A. Kokkinis; C. Grunseich; M. Jain; M. Waite; V. Biancavilla; M. Barnes; S. Pocock; K. Brooks; M. Emile-Backer; Y. Webb; E.E. Marcantonio; A.R. Foley; K. Meilleur; C. Bönnemann; P. Mohassel

LBP.16
Characterizing novel exploratory biomarkers in a longitudinal natural history study in patients with Types 2 and 3 SMA
N. Hellbach; T.M. Karrer; M.L. Moal; N.M.P. King; W.E. Glaab; A. Daron; M. Annoussamy; Y. Pereon; C. Cances; A.M. Seferian; N. Goemans; V. Laugel; U. Schara; K. Gorni; T. Seabrook; C. Czech; L. Servais; T.S. Zabka on behalf of the NatHis-SMA Study Group

LBP.17
Pathogenic variants in slow skeletal troponin I (TNNI1) cause skeletal muscle contractile dysfunction
S. Donkervoort; M. van de Locht; G. Ravenscroft; S. Conijn; J. Reunert; O.L.A. Neto; C. McLean; S. McModie; V. Bolduc; J. de Winter; A. Viaene; Y. Hu; S. Neuhaus; L. Medne; M. Davis; A.R. Foley; T. Marquardt; N. Laing; C. Bönnemann; C. Ottenheijm

LBP.18
Comparison of North Star Ambulatory Assessment score change in nmDMD patients receiving ataluren: STRIDE Registry vs phase 3 clinical trial
F. Muntoni; M. Tulinius; F. Buccella; I. Desguerre; J. Kirschner; A. Nascimento Osorio; S. Johnson; C. Werner; J. Jiang; J. Li; P. Trifillis; C.L. Santos; E. Mercuri

LBP.19
Associations between dystrophin genotype and ambulatory outcomes in DMD: implications for trials of genotype-targeted therapies
F. Muntoni; J. Signorovitch; G. Sajeev; H. Lane; M. Jenkins; I. Dieye; S. Ward; C. McDonald; N. Goemans; E. Niks; B. Wong; L. Servais; V. Straub; I.J.M. De Grooot; C. Tian; A. Manzur; E. Mercuri

LBP.20
ATL1102 treatment in non-ambulant boys with DMD modulates Latent TGF-beta-binding protein 4, a disease genetic modifier of DMD, and CXCL16
G. Tachas; C. Mueller; I. Woodcock; M. Ryan; N. Desem

LBP.21
Pulmonary function decline analysis in non-ambulatory patients with DMD: ataluren Study 019 compared with the CINRG Duchenne natural history study
M. Tulinius; F. Muntoni; V. Penematsa; J. Jiang; A. Kristensen; E. Goodwin; H. Gordish-Dressman; L. Morgenroth; C. Werner; J. Li; P. Trifillis; C.M. McDonald

LBP.22
Safety, tolerability, and pharmacokinetics of eteplirsen in patients 6–48 months old with Duchenne muscular dystrophy amenable to exon 51 skipping
E. Mercuri; A.M. Seferian; L. Servais; N. Deconinck; H. Stevenson; L. East; W. Zhang; S. Upadhyay; F. Muntoni

LBP.23
Longitudinal evaluation of Creatine Kinase, Creatine/Creatinine ratio and Myostatin and associations with disease severity and disease progression in Becker muscular dystrophy
N. van de Velde; Z. Koeks; M. Signorelli; N. Verwey; M. Overzier; J. Bakker; G. Sajeev; J. Signorovitch; J. Verschuuren; K. Brown; P. Spitali; E. Niks

LBP.24
Duchenne muscular dystrophy patients lacking Dp140 and Dp71 and mouse models lacking Dp140 have a more severe motor phenotype
M. Chesshyre; D. Ridout; Y. Hashimoto; Y. Ookubo; S. Torelli; K. Maresh; V. Ricotti; L. Abbott; V.A. Gupta; M. Main; M. Scoto; G. Baranello; A. Manzur; Y. Aoki; F. Muntoni

LBP.25
SPTLC2 as a cause of childhood-onset amyotrophic lateral sclerosis
S. Syeda; S. Donkervoort; P. Mohassel; A. Lakhotia; J.E. Galarza-Brito; M.C. França; P. Munot; A. Majumdar; A. Asamoah; K.E. Jackson; P. B. Shieh; T.L. Winder; K.R. Chao; M. Lone; S.D. Gupta; K. Gable; F. Muntoni; A.R. Foley; T. Hornemann; T.M. Dunn; C.G. Bönnemann

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)
Investigators report a new monogenic cause for childhood-onset ALS with associated therapeutic implications, a novel chaperonopathy manifesting as a myopathy with early respiratory failure, the use of tissue and single cell transcriptomics for studying differential gene expression across muscles, and the strengths and weaknesses of RNA sequencing for evaluating variants in titin.
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 **New genes, new techniques in neuromuscular disorders breakout session**

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)
This is a very exciting time for development of therapy for neuromuscular disorders, which makes this session very interesting. Generation of pre-clinical data in relevant models of diseases is very valuable and is indicating which avenues some therapy will take before going to clinic. This year, this session is highlighting four talks: one investigating overexpression of a genetic modifier (jagged1) to treat DMD, one looking at inhibiting RhoA pathway to diminish fibrosis in DMD, one exploring a low dose of AAV to treat Fukutin-related-protein deficiencies and finally a talk reporting the utilization of allele specific gene editing to treat COL6A1 associated disorders.

Moderators: Nicolas Wein & Adjali Oumeya

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 Rhokinase 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 **Pre-clinical developments in neuromuscular disorders breakout session**

Friday, 24 September 2021 **CONGRESS DAY 3**

13:30-14:30 **General Assembly (WMS Member congress delegates only)**

15:00-15:45 **Poster highlights (PH 1-6)**
(6 x 5 mins consecutive pre-recorded presentations + 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.
Moderators: Johann Böhm & Jantima Tanboon

PH 1
GNE pathogenic variant p.D207V rarely leads to myopathy in homozygotes; 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

PH 2
scAAV.U7snRNA-mediated therapy: prolonged dystrophin expression and muscle function correction in adult Dup2 mice
L. Gushchina; A. Bradley; T. Vetter; E. Frair; C. Bellinger; T. Simmons; N. Rohan; N. Wein; K. Flanigan

PH 3
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

PH 4
Efficacy and safety of mexiletine in non-dystrophic myotonias: a randomized, double-blind, placebo-controlled, cross-over study
S. Vicart; J. Franques; F. Bouhour; A. Magot; Y. Pereon; S. Sacconi; A. Nadaj-Pakleza; A. Behin; N. Zahr; M. Hezode; E. Fournier; C. Payan; L. Lacomblez; B. Fontaine

PH 5
Longitudinal developmental profile of newborns and toddlers treated for spinal muscular atrophy
M. Ngawa; F. Dal Farra; A. Marinescu; L. Servais

PH 6
Efficient modification of DMD gene using PRIME editing
C. Happi Mbakam; J. Rousseau; G. Tremblay; J. Tremblay

15:45-16:15 **Poster highlights breakout session**

16:15-17:00 **Late breaking News Session (LBO 1-3)**
(3 x 10 mins consecutive pre-recorded presentations + 15 mins live Q&A)
One of the most exciting sessions of the annual WMS Congress, providing 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.
Moderators: Gisèle Bonne & Carsten Bönnemann

LBO 01
Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy
M. Taniguchi-ikeda; E. Bonora; S. Chakrabarty; G. Kellaris; J. Tanboon; I. Nishino; T. Toda; Y. Goto; I. Nonaka; N. Katsanis; F. A. M. Duijkers; R. De Giorgio;

LBO 02
Biallelic loss-of-function OBSCN variants predispose individuals to severe, recurrent rhabdomyolysis
M. Cabrera-Serrano; L. Caccavell; M. Savarese; A. Vihola; M. Jokela; M. Johari; T. Capiod; M. Madrange; E. Bugiardini; S. Brady; R. Quinlivan; A. Merve; R. Scalco; D. Hilton-Jones; H. Houlden; H. Aydin; S. Ceylaner; J. Vockley; R. Taylor; H. Goullee; E. Ylikallio; M. Auranen; H. Tyynismaa; B. Udd; A. Forrest; M. Davis; D. Bratkovic; N. Manton; T. Robertson; P. McCombe; N. Laing; L. Phillips; P. de Lonlay; G. Ravenscroft;

LBO 03
HOPE-2 Multi-center Randomised Clinical Trial of Intravenous Human Cardiosphere-Derived Cells for Late-Stage Duchenne Muscular Dystrophy
C. McDonald; E. Marbán; S. Hendrix; N. Hogan; R. R. Smith; M Eagle; R. Finkel; C. Tian; J. Janas; M. Harmelink; A. Varadhachary; M. Taylor; K. Hor; O. Mayer; E. Henricson; P. Furlong; D. Ascheim; S. Rogy; P. Williams; L. Marbán.

17:15-18:15 **Prize presentations:** *Johann Böhm*
Close of Congress: *Volker Straub*
Introduce 2022 and presentation of flag: *Jim Dowling and Jiri Vajsar*