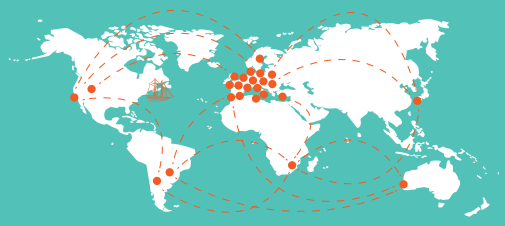

Programme Key:

I	**Invited Speaker**
O/LBO	**Selected Oral Presentation**
P/LBP	**Poster Presentation** (on display at the venue as a poster and e-poster and on the virtual platform)
VP	**Virtual Poster Presentation** (on display on the virtual platform and on e-poster boards at the venue)
FP	**Selected Flash Poster Presentation** (short flash talk in dedicated area + poster presentation as above)
LBP	**Late Breaking Poster** (on display at the venue as a poster and e-poster and on the virtual platform)
LSVP	**Late Submitted Poster** (only on display on the virtual platform and on e-poster boards at the venue)

WMS2022 Full Programme

Tuesday 11th October 2022

12:00-17:00	**WMS Executive Board Meeting** - Meeting Room 503
16:00-18:00	**Registration and Poster set up** - Ballroom Salon and Ballroom B1-B2
16:30-17:15	Tea and Coffee available
18:00-18:45	**Opening Ceremony** - Ballroom **Anything Is Possible** James Mullinger *Moderators: Jim Dowling and Jiri Vajsar*
18:45-21:00	**Networking Reception** - Ballroom Level

Wednesday 12th October 2022

06:30	**Conference desk opens and setting up of posters**	
07:00-08:30	**Symposium 1** - Ballroom	**Symposium 2** - Argyle
08:45-09:00	**Congress Welcome** - Message from the President - Ballroom	
09:00-10:30	**New Developments in Congenital Myopathies 1** Invited lectures (I.01-02) - Ballroom *Moderators: Sandra Donkervoort and Bjarne Udd*	
09:00-09:30	I.01 **Congenital myasthenic syndromes** H. Lochmüller	
09:30-10:00	I.02 **Titin The newly-emerged "titan" of the cardiac and skeletal muscle disease world** E. Oates	

	Selected oral presentations - New Developments in Congenital Myopathies 1 (O.01-02) - Ballroom *Moderators: Sandra Donkervoort and Bjarne Udd*
10:00-10:15	O.01 **Correlating molecular genetic testing for rare genetic variants with a broad clinicopathologic spectrum of congenital myopathies** D. Jayaraman; C. Genetti; A. Aykanat; W. Win; Z. Valivullah; E. O'Heir; B. Darras; R. Laine; A. O'Donnell-Luria; A. Beggs
10:15-10:30	O.02 **The phenotypic spectrum of DNM2-related centronuclear myopathy** L. Hayes; M. Perdomini; A. Aykanat; C. Genetti; H. Paterson; B. Cowling; C. Freitag; A. Beggs
10:30-11:00	Morning refreshments, exhibition and posters - Ballroom Salon and Pre Function Space
11:00-13:00	**New Developments in Congenital Myopathies 2** Invited lectures (I.03-04) - Ballroom *Moderators: Kristl Claeys and Andreea Seferian*
11:00-11:30	I.03 **Novel mechanisms and new therapies for congenital myopathies** J. Dowling
11:30-12:00	I.04 **Congenital myopathies in adulthood: the other end of the spectrum** N. Voermans
	Selected oral presentations - New Developments in Congenital Myopathies 2 (O.03-06) - Ballroom *Moderators: Kristl Claeys and Andreea Seferian*
12:00-12:15	O.03 **Nemaline myopathy type 6: from pathology to therapeutics** R. Galli; S. Shengyi; R. van der Pijl; H. Granzier; J. de Winter; C. Ottenheijm
12:15-12:30	O.04 **Improvement of muscle strength in a mouse model for recessive RYR1-related congenital myopathy treated with HDAC and inhibitors** A. Ruiz; S. Benucci; M. Franchini; S. Treves; F. Zorzato
12:30-12:45	O.05 **Silencing of the Ca2+ channel ORAI1 improves the multi-systemic phenotype of tubular aggregate myopathy and Stormorken syndrome in mice** R. Silva-Rojas; J. Laporte; J. Böhm
12:45-13:00	O.06 **Long term outcomes for X-Linked Myotubular Myopathy (XLMTM) with gene replacement therapy, resamirigene bilparvovec: preliminary results from ASPIRO** P. Shieh; N. Kuntz; J. Dowling; W. Müller-Felber; A. Blaschek; C. Bönnemann; R. Foley; D. Saade; A. Seferian; L. Servais; A. Bowden; M. Sarazen; J. Coats; N. Lakshman; C. Han; S. Prasad; S. Rico; W. Miller
13:00-14:30	Lunch, exhibition and posters - Ballroom Salon and Pre Function Space
13:30-14:00	**New WMS Members Event** - Meeting Room 503
14:30-16:00	**Poster session 1** (FP.01-11) - Flash Poster Presentation areas - Ballroom or Ballroom Salon (P.01-54) (VP.01-21) - Poster area - Ballroom B1-B2 ***(Please note that this session will not be live streamed for virtual delegates)***
14:30-14:40	**Congenital Myopathies** (FP.01-02) – Ballroom *Moderator: Vandana Gupta*
14:30-14:35	FP.01 **Novel disease pathways and therapeutic developments in Kelch-related congenital nemaline myopathy** A. Mansur; J. Casey; R. Joseph; J. Shi; E. Karimi; B. Tao; H. Granzier; V. Gupta
14:35-14:40	FP.02 **Mutation in *KBTBD13* causes stiffening of thin filaments in skeletal muscle** S. Conijn; R. Galli; M. van de Locht; W. Ma; T. Irving; J. de Winter; C. Ottenheijm
14:30-16:00	**Congenital Myopathies** (P.01-11) (VP.01-07) - Poster area - Ballroom B1-B2
	P.01 **A *KLHL40* 3'UTR splice-altering variant causes milder NEM8** L. Dofash; G. Monahan; E. Servián-Morilla; E. Rivas; F. Faiz; P. Sullivan; E. Oates; J. Clayton; R. Taylor; M. Davis; T. Beilharz; N. Laing; M. Cabrera-Serrano; G. Ravenscroft

P.02 **Mild nemaline myopathy 10 caused by a novel missense homozygous mutation in *LMOD3*: broadening the phenotype-genotype correlation**
A. Segarra-Casas; L. Gonzalez-Quereda; M. Caballero; M. Rodriguez; A. Vesperinas; R. Collet; J. Díaz-Manera; E. Gallardo; P. Gallano; M. Olive

P.03 **Myosin dysregulation in nemaline myopathy**
J. Laitila; N. Ranu; J. Mariano; C. Wallgren-Pettersson; N. Witting; J. Vissing; J. Vilchez; C. Fiorillo; E. Zanoteli; M. Auranen; M. Jokela; T. Beck; S. Larsen; A. Kontrogianni-Konstantopoulos; J. Ochala

P.04 **New developments and data highlights in the international myotubular and centronuclear myopathy patient registry**
S. McDonald; J. Bullivant; A. Lennox; A. den Hollander; C. Saegert; O. Lynch; D. Moat; R. Graham; U. Schara-Schmidt; C. Bönnemann; H. Jungbluth; A. Buj-Bello; J. Dowling; C. Marini-Bettolo

P.06 **Investigating the effect of a home-based training program on oxidative capacity in patients with truncating genetic variants in titin**
I. Flensted; C. Vissing; M. Stemmerik; S. Skriver; K. Axelsen; H. Bundgaard; J. Vissing

P.07 **Characterization of MRI brain abnormalities in X-linked myotubular myopathy**
L. Vogt; A. Marefi; K. Amburgey; N. Addour; E. Widjaja; C. Poulin; M. Oskoui; H. McMillan; N. Chrestian; C. Saint-Martin; M. Srour; J. Dowling

P.08 **Phase 1 open-label trial of Rycal S48168 (ARM210) for *RYR1*-related myopathies**
J. Todd; T. Lawal; I. Chrismer; A. Kokkinis; C. Grunseich; M. Jain; M. Waite; M. Barnes; V. Biancavilla; S. Pocock; K. Brooks; W. Reikof; M. Emile-Backer; A. Marks; Y. Webb; E. Marcantonio; A. Foley; K. Meilleur; C. Bönnemann; P. Mohassel

P.09 **Comprehensive database for *RYR1*-related disorders: concept and progress update**
T. Lawal; W. Riekhof; M. Goldberg; A. Kushnir; N. Terry; A. Marks; J. Todd

P.10 **Compound *CACNA1S* heterozygosity resulting in a novel phenotype of congenital myopathy and early onset periodic paralysis: report of two probands**
S. Aburahma; M. Shboul; S. Lucchiari; G. Comi; G. Meola; S. Pagliarani

P.11 **Clinical, pathological, imaging, and genetic characterization in a Taiwanese cohort with congenital myopathy**
W. Liang; C. Wang; W. Xao; W. Chen; I. Nishino; Y. Jong

VP.01 **Array comparative genomic hybridisation and droplet digital PCR uncover recurrent copy number variation of the titin segmental duplication region**
L. Sagath; V. Lehtokari; K. Pelin; K. Kiiski

VP.02 **Comprehensive characterization of early-onset skeletal muscle disease gene exon usage and splicing patterns across different developmental ages**
Z. Su; A. Smolnikov; A. Khazaal; M. Dinger; E. Oates

VP.03 **Diagnosing Titinopathy: lessons from a multi-omics pilot study**
Y. Zhang; L. Xu; Y. Lei; S. H.S. Chan; A. Javed

VP.04 **Ryanodine receptor - related disorders**
M. Marttila; Ö. Birsoy; V. Gupta; S. Amr; B. Funke; H. Hynes; C. Genetti; L. Swanson; P. Agrawal; H. Rehm; A. Beggs

VP.05 **Innervation defect: new pathomechanism of centronuclear myopathy?**
Y. Saito; S. Hayashi; S. Noguchi; I. Nishino

VP.06 **A new case of congenital myopathy related to *TNNC2***
B. Lace; N. Laflamme; S. Thonta Setty; N. Rioux; B. Ellezam; S. Rivest; N. Chrestian

VP.07 **Lived experience of functioning of patients with nemaline myopathy and related disorders in Finland**
V. Lehtokari; C. Wallgren-Pettersson; M. Similä; M. Tammepuu; S. Strang-Karlsson; S. Hiekkala

14:40-14:50 **DMD – Clinical** (FP.03-04) – Ballroom
Moderator: Michela Guglieri

14:40-14:45 FP.03 **The spine fracture burden in boys with DMD treated with the novel dissociative steroid vamorolone versus deflazacort and prednisone**
L. Ward; S. Jackowski; U. Dang; M. Scharke; J. Jaremko; K. Koujok; M. Matzinger; N. Shenouda; K. Siminoski; M. Leinonen; R. Rooman; S. Hasham; P. Clemens; M. McDermott; R. Griggs; M. Guglieri; E. Hoffman; The Ottawa Pediatr Bone Health; FOR DMD Invest Muscle Group; CINRG VBP15-002/003/LTE Invest

14:45-14:50 FP.04 **Three year natural history study in Becker muscular dystrophy in The Netherlands**
E. Schrama; Z. Koeks; N. van de Velde; M. Hooijmans; I. Alleman; J. Verschuuren; P. Spitali; H. Kan; E. Niks

14:30-16:00 **DMD – Clinical** (P.12-29) (VP.08-12) - Poster area - Ballroom B1-B2

P.12 **A case report of near normalization of serum creatine phosphokinase in a patient with Duchenne muscular dystrophy during acute pancreatitis**
A. Zygmunt; I. Rybalsky; L. Reebals; C. Tian

P.13 **What is the future for female patients with childhood onset symptomatic Duchenne muscular dystrophy?**
S. Houwen- van Opstal; R. Tak; M. Pelsma; F. van den Heuvel; H. Duyvenvoorde; E. Cup; A. Verrips; L. Sie; J. Vles; I. Groot; N.Voermans; M. Willemsen

P.14 **Analysis of the longitudinal CINRG Becker natural history study dataset**
P. Clemens; H. Gordish-Dressman; G. Niizawa; K. Gorni; M. Guglieri; A. Connolly; M. Wicklund; T. Bertorini; J. Mah; M. Thangarajh; E. Smith; N. Kuntz; C. McDonald; E. Henricson; S. Upadhyayula; B. Byrne; G. Manousakis; A. Harper; S. Iannaccone; U. Dang

P.15 **Symptomatic DMD carrier as a differential diagnosis in patients presenting asymmetrical limb weakness**
M. Cho; Y. Lee; A. Kim; J. Lee

P.16 **Parental illness intrusiveness, parental well-being and youth well-being in families confronted with Duchenne muscular dystrophy**
S. Prikken; S. Geuens; E. Gielis; N. Goemans; L. De Waele

P.17 **Importance of routine pulmonary check-up prior to ventilatory support in patients with Duchenne muscular dystrophy**
W. Choi; H. Cho; J. Lee; S. Kang; I. Kim; D. Kim; S. Pyo; C. Jang

P.18 **Comparison of the performance of the upper limb module in children with Becker muscular dystrophy, Duchenne muscular dystrophy and healthy controls**
A. Wolfe; M. Main; L. Abbott; N. Burnett; V. Selby; F. Muntoni

P.19 **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; A. Nascimento Osorio; M. Tulinius; S. Johnson; C. Werner; J. Jiang; J. Li; J. Jia; P. Trifillis; E. Mercuri

P.20 **Experiences of and perspectives on bullying in youth with myopathies**
N. Chatur; C. Ippolito; L. McAdam

P.21 **Comorbidity and leading causes of death in children and adolescents with Duchenne muscular dystrophy**
L. Wahlgren; M. Tulinius; A. Kroksmark; K. Sofou

P.22 **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; L. Morgenroth; H. Gordish-Dressman; S. Johnson; C. Werner; J. Jiang; J. Li; J. Jia; P. Trifillis; C. McDonald

P.23 **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; L. Morgenroth; H. Gordish-Dressman; S. Johnson; C. Werner; J. Jiang; J. Li; J. Jia; P. Trifillis; C. McDonald

P.24 **Consensus and collaboration approach to better defining and implementing harmonised standards of care across a healthcare system: examples from DMD care**
C. Turner; J. Bourke; A. Childs; V. Gowda; M. James; A. Johnson; A. Manzur; A. Mayhew; F. Muntoni; R. Quinlivan; S. Rodney; A. Sarkozy; V. Straub; S. Wong; M. Guglieri

P.25 **Patient-reported aspects of Becker muscular dystrophy from the Duchenne registry, a registry for Duchenne and Becker muscular dystrophy (BMD)**
J. Donovan; A. Bronson; N. Armstrong; A. Martin

P.26 **Motor and cognitive manifestations of young female carriers of Duchenne muscular dystrophy (DMD): a prospective natural history study**
M. Iammarino; N. Truba; L. Alfano; N. Reash; K. Ignasiak; L. Pietruszewski; A. Long; B. Sabo; K. Lehman; J. Mendell; L. Lowes;

P.27 **Spinal aspects in DMD patients on intermittent corticosteroid dosing - a single center study**
N. Ikelaar; M. Stoop; L. Blok; J. Bongers; Y. Meijer-Krom; P. Dibbets-Schneider; P. de Witte; E. Niks

P.28 **Introduction of 12 novel pathogenic *DMD* variants, associated phenotypes and studies of dystrophin and MAST1 abundances**
A. Gangfuss; K. Neuhoff; A. Hentschel; N. Kohlschmidt; H. Koelbel; U. Schara-Schmidt; A. Roos

P.29 **Accelerating clinical development of new therapeutics with patient data: evidence from the collaborative Trajectory Analysis Program (cTAP) in DMD**
C. McDonald; S. Ward; J. Signorovitch; F. Muntoni; N. Goemans; B. Wong; K. Vandenborne; A. Manzur; G. Sajeev; E. Mercuri

VP.08 **Interests and experiences of young adults with muscular dystrophy in receiving genetic information**
L. Hammond; K. Amburgey; D. Chitayat; S. Hewson; L. McAdam

VP.09 **Clinical characteristics and gene analysis of 10 rare cases with coexistence of double genetic diseases**
D. Tan; Y. Liu; D. Song; Y. Fan; H. Xiong

VP.10 **The occurrence of bowel symptoms in adults with Duchenne muscular dystrophy**
A. Pietrusz; M. Desikan; A. Emmanuel; R. Quinlivan

VP.11 **Shoulder subluxations and dislocations in non-ambulatory patients with Duchenne muscular dystrophy (DMD)**
C. Goncalves; R. Dos Santos; K. Shellenbarger; M. Kiefer; E. Shelton; Y. Mortimer; G. Gaebe; B. Wong

VP.12 **Covid-19 infections in an adult cohort of Duchenne muscular dystrophy attending the National Hospital for neurology and neurosurgery in London**
M. Desikan; F. Cruces; A. Pietrusz; R. Quinlivan

14:30-14:55 **Imaging and new tools and approaches for NMDs** (FP.05-09) – Ballroom Salon
Moderator: Jordi Díaz-Manera

14:30-14:35 FP.05 **From the Muscle Atlas to an AI-based diagnostic tool**
C. Meyer; E. Lacene; M. Beuvin; T. Evangelista; J. Laporte; A. Jeannin-Girardon; P. Collet; O. Poch; N. Romero; K. Chennen; B. Cadot

14:35-14:40 FP.06 **Use of an exercise challenge system to define a universal proteomic signature of muscle injury in a diverse set of adults with inherited myopathy**
M. Stemmerik; B. Barthel; N. Andersen; S. Skriver; A. Russell; J. Vissing

14:40-14:45 FP.07 **Experiences with systematic video recordings of individualized physiotherapy for 111 patients with neuromuscular disorders in a 7-year quality project**
A. Rosenberger; A. Lahelle

14:45-14:50 FP.08 **Evaluating the expression of spontaneous movements in infants with neuromuscular conditions using Prechtl's General Movements Assessment**
M. Iammarino; L. Pietruszewski; M. Wendland; N. Reash; O. Mogilnicki; K. Adderley; A. Long; L. Alfano; L. Lowes

14:50-14:55 FP.09 **Analysis of muscle MRI of a large cohort of chronic motor neuropathy/neuronopathy patients reveals characteristic features useful for diagnosis**
D. Esteller; J. Morrow; D. Reyes; G. Bisogni; M. Monforte; G. Tasca; A. Alangary; C. Marini Bettolo; M. Sabatelli; G. Ramdharry; J. Alonso Perez; J. Turon Sans; M. Guglieri; A. Rossor; M. Olive; E. Bertini; V. Straub; M. Reilly; R. Rojas Garcia; J. Díaz Manera

14:30-16:00 **Imaging and new tools and approaches for NMDs** (P.30-36a) - Poster area - Ballroom B1-B2

P.30 **Correlation of histopathological skeletal muscle biopsy features with quantitative muscle-MRI parameters**
A. Guettsches; R. Rehmann; A. Schreiner; M. Rohm; J. Forsting; M. Froeling; M. Tegenthoff; M. Vorgerd; L. Schlaffke

P.31 **A multiparametric quantitative NMR study at rest and during exercise in subjects between 22 and 65 years of age: Preliminary results**
A. Lopez Kolkovsky; B. Matot; P. Baudin; H. Reyngoudt; B. Marty; Y. Fromes

P.32 **Impact of age on muscle volume and T2-relaxation time during adulthood in mice using quantitative MRI**
B. Matot; E. Caldas de Almeida Araujo; P. Baudin; H. Reyngoudt; B. Marty; Y. Fromes

P.33 **Muscle ultrasound use in the initial diagnosis of childhood onset neuropathy and neuronopathy**
A. Zygmunt; S. Deng; S. Donkervoort; P. Mohassel; D. Bharucha-Goebel; D. Saade; S. Neuhaus; J. Dastgir; N. Acquaye; L. Hinkley; T. Lehky; A. Foley; C. Bönnemann

P.34 **MDA MOVR clinical data hub provides insights into adoption of approved therapies for neuromuscular disease**
S. Hesterlee; E. Kilroy; J. Waits

P.35 **Genetic newborn screening and digital technologies to accelerate rare disease diagnosis: the EU-IMI screen4Care project**
A. Ferlini; F. Fortunato; M. Farnè; R. Selvatici; C. Blankart; R. Röttger; J. Kirschner; J. Schenk; K. Zarakowska; J. Zschüntzsch; Y. Man; L. Goodman; E. Gross; N. Garnier

P.36 **Carrier frequency and genetic prevalence of autosomal recessive genetic neuromuscular disorders in Korea**
H. Lee; E. Oh; U. Yun; S. Lee; H. Park; Y. Choi

P.36a **"suMus", a novel digital system for arm movement metrics and muscle energy expenditure in neuromuscular diseases**
T. Gerhalter; C. Müller; E. Maron; A. Mähler; T. Schütte; M. Boschmann; R. Herzer; S. Spuler; E. Gazzerro

14:50-15:00 **SMA - Clinical & Biomarkers/Outcome Measures** (FP.10-11) – Ballroom
Moderator: Charlotte Lilien

14:50-14:55 FP.10 **Combination of BIO101 with antisense oligonucleotide therapy demonstrates synergistic beneficial effects in severe SMA-like mice**
C. Bezier; P. Nazari Hashemi; S. Cottin; R. Lafont; S. Veillet; F. Charbonnier; P. Dilda; M. Latil; O. Biondi

14:55-15:00 FP.11 **Impact of disease-modifying therapies on myostatin levels in SMA patients**
L. Mackels; V. Mariot; L. Servais; J. Dumonceaux

14:30-16:00 **SMA - Clinical & Biomarkers/Outcome Measures** (P.37-54) (VP.14-21) - Poster area - Ballroom B1-B2

P.37 **Outcomes in patients with spinal muscular atrophy and four or more *SMN2* copies treated with onasemnogene abeparvovec: Findings from RESTORE**
R. Finkel; K. Benguerba; A. Reid; D. Raju; E. Faulkner; N. LaMarca; L. Servais

P.38 **A novel splice site variant in a patient with spinal muscular atrophy and hypoplastic left heart syndrome**
C. Hedberg-Oldfors; E. Jennions; K. Visuttijai; J. Gudnason; A. Oldfors

P.39 **Altered functional connectivity in motor regions in children with spinal muscular atrophy**
N. Mugisha; H. Carlson; M. Brossard-Racine; A. Kirton; M. Oskoui

P.40 **The importance of bulbar/respiratory symptoms in spinal muscular atrophy: results from interviews with patients, caregivers and healthcare providers**
O. Gassner; S. Runge; J. Braid; C. Guittari; A. Hareendran; A. Skalicky; T. Perumal

P.41 **Adult SMA REACH: development and implementation data collection study in the UK Adult SMA population**
S. Segovia Simon; S. Fitzsimmons; L. Murphy; R. Muni-Lofra; C. Blewit; S. Adult SMA Reach Study Group; C. Marini Bettolo

P.42 **CuidAME: registry for longitudinal data collection of Spanish SMA patients**
S. Segovia-Simon; C. Nungo; J. Vazquez Costa; I. Pitarch; J. Caballero; C. Rodríguez Sánchez; S. Pascual; J. Expósito; C. Marco Cazcarra; M. Povedano; A. Pareja; M. Lopez Lobato; M. Álvarez; L. Costa; D. Gómez Andrés; F. Munell; A. Moreno; E. Martínez; A. Nascimento; C. CuidAME Study Group

P.43 **Evaluating knee ankle foot orthoses in children with type 1 spinal muscular atrophy**
A. Rohwer; M. Main; L. Abbott; A. Wolfe; M. Scoto; F. Muntoni

P.44 **Cost-effectiveness of spinal muscular atrophy newborn screening in Belgium**
T. Dangouloff; P. Thokala; A. Daron; S. Delstanche; L. Servais; M. Hiligsmann

P.45 **Adult SMA REACH: an integrated model to facilitate transition of data and longitudinal data collection of clinician and patient entered data**
S. Fitzsimmons; S. Segovia; L. Murphy; C. Blewitt; A. Mayhew; R. Muni-Lofra; C. Marini-Bettolo

P.46 **Magneto-inertial wearable device: upper limb trajectory identification in non-ambulant patients with spinal muscular atrophy**
C. Lilien; A. Tricot; M. Annoussamy; M. Polleur; L. Clavel; T. Terray; A. Guérin; D. Eggenspieler; D. Lozeve; L. Servais

P.47 **Adaptive test for neuromuscular disorders: design of a wheelchair-based assessment**
T. Duong; W. Tang; L. Nelson; D. Parker; A. Pasternak; S. Dunaway Young; R. Muni-Lofra; E. Maczek; J. Michell-Sodhi; D. Moat; S. Chatfield; P. Appleton; J. Day; A. Glanzman; A. Mayhew

P.48 **The impact of newborn screening and early therapy on the course of spinal muscular atrophy: a retrospective analysis of a single center experience**
P. Karachunski; M. Stark; K. McGrattan; C. Weigel; J. Ihinger; P. Kang

P.51 **Real-world experience after one year treating SMA children with risdiplam**
M. Gomez Garcia de la Banda; R. Garcia-Uzquiano; A. Benezit; J. Davion; I. Dabaj; H. Amthor; A. Bouadi; M. Spigarelli; C. Bocassin; S. Tirolien; M. Villart; L. Sonnet; L. Grimaldi-Bensouda; S. Quijano-Roy

P.52 **Determination of DLC1 isoform 1 (DLC1-i1) as a gene therapy for the treatment of spinal muscular atrophy**
T. Shi; B. Liao; W. Xiong; S. Chan; J. Liu; M. Cheung

P.53 **Muscle microRNAs in the cerebrospinal fluid predict clinical response to nusinersen therapy in type II and type III spinal muscular atrophy patients**
I. Magen; S. Aharoni; N. Yacovzada; I. Tokatly Latzer; C. Alves; L. Sagi; A. Fattal-Valevski; K. Swoboda; J. Katz; E. Bruckheimer; Y. Nevo; E. Hornstein

P.54 **Assessment of fine motor abilities using new touchscreen application, among children with spinal muscular atrophy (SMA) - a pilot study**
I. Klemm; A. Danial-Saad; Y. Nevo; I. Eshel; S. Aharoni

VP.13 **Updates on the development of the spinal muscular atrophy - person-reported outcome (SMA-PRO): a caregiver and self-proxy performance measure for child**
A. Pasternak; E. Maczek; M. Fragala-Pinkham; C. Dias; K. Nedeljkovic; J. Montes; A. Glanzman; S. Dunaway-Young; N. Dilek; B. Darras

VP.14 **Growth patterns in treated SMA children in the UK**
S. Raquq; G. Stimpson; M. Fewtrell; E. Cavalcante; F. Muntoni; G. Baranello

VP.15 **Clinical and genetic study of a spinal muscular atrophy family with variable phenotypes**
S. Liu; C. Wei; Y. Fan; Z. Jia; H. Xiong

VP.16 **Assessment of muscular, respiratory and cardiological function and the number of copies of the SMN2 gene in patients with spinal muscular atrophy (SMA)**
R. Escobar Cedillo; O. Hernandez Hernandez; A. Miranda Duarte; A. Luna Angulo; R. Coral Vazquez; F. Ramos Becerril; L. Qiuntanar Trejo; B. Gomez Díaz; R. Suarez Sanchez

VP.17 **Long-term effect of nusinersen treatment on motor, respiratory and bulbar function in children with SMA type 1 - a 3-year SMArtCARE registry study**
A. Pechmann; M. Behrens; G. Bernert; T. Hagenacker; W. Müller-Felber; U. Schara-Schmidt; I. Schwersenz; M. Walter; H. Lochmüller; J. Kirschner

VP.18 **Social communication skills in spinal muscular atrophy (SMA) type 1 children treated with approved disease-modifying therapies**
C. Brusa; H. Weststrate; E. Clark; E. Johnson; E. Barritt; M. Scoto; P. Munot; A. Manzur; F. Muntoni; G. Baranello

VP.19 **Rehab Robo: a high sensitivity patient outcome tracking and physical exercise tool for spinal muscular atrophy**
O. Kent; T. Roberts; B. Riello

VP.20 **Cathepsin D as biomarker in CSF of nusinersen-treated patients with spinal muscular atrophy**
D. Schorling; H. Kölbel; A. Hentschel; A. Pechmann; N. Meyer; B. Wirth; R. Rombo; A. SMArtCARE Consortium; A. Sickmann; J. Kirschner; U. Schara-Schmidt; H. Lochmüller; A. Roos

VP.21 **Baseline nutrition investigation in a Chinese cohort of pediatric patients with spinal muscular atrophy**
S. Li; S. Liu; Y. Wu; Y. Liu; D. Tan; Y. Fan; C. Wei; H. Xiong

15:45-16:15 Afternoon refreshments, exhibition and posters - Ballroom Salon and Pre Function Space

16:00-17:30 **Poster session 2**
(FP.12-24) - Flash Poster Presentation areas - Ballroom or Ballroom Salon
(P.55-118) (VP.22-56b) - Poster area - Ballroom B1-B2
(Please note that this session will not be live streamed for virtual delegates)

16:00-16:20 **DMD - Biomarkers/Outcome Measures** (FP.12-15) – Ballroom
Moderator: Linda Lowes

16:00-16:05 FP.12 **Application for primary endpoint qualification of the 95th centile of stride velocity (SV95C) in Duchenne muscular dystrophy**
M. Annoussamy; D. Eggenspieler; A. Seferian; E. Mercuri; V. Straub; F. Muntoni; M. Scoto; M. Poleur; A. Daron; N. Butoianu; A. Mirea; N. Goemans; S. Previtali; M. Tulinius; A. Nascimento; P. Heydemann; M. Panzara; T. Singh; P. Strijbos; L. Servais

16:05-16:10 FP.13 **Diffusion-tensor MRI captures increased diameter and size heterogeneity of skeletal muscle fibres in Becker muscular dystrophy, as verified by histology**
D. Cameron; T. Abbassi-Daloii; L. Heezen; N. van de Velde; Z. Koeks; T. Veeger; M. Hooijmans; J. Verschuuren; M. van Putten; A. Aartsma-Rus; V. Raz; P. Spitali; E. Niks; H. Kan

16:10-16:15 FP.14 **Dystrophin and satellite cell quantification in Duchenne and Becker muscular dystrophies**
S. Nicolau; T. Vetter; E. Frair; A. Bradley; K. Flanigan

16:15-16:20 FP.15 **AI-powered cell profiling enables the functional evaluation of therapies targeting muscle disorders in patient-derived myotubes**
B. Darimont; O. Lorintiu; T. Champetier; E. Duchemin-Pelletier; M. Flaender; V. Chapuis-Perrot; J. Young; C. Gaston; L. Griveau; M. Papin; P. Poydenot; E. Ventre; L. Selig

16:00-17:30 **DMD - Biomarkers/Outcome Measures** (P.55-73) (VP.22-28) - Poster area - Ballroom B1-B2

P.55 **A concisely recorded ambulatory assessment for enhancing real-world outcomes research in Duchenne muscular dystrophy: development and validation**
A. Mayhew; J. Signorovitch; V. Straub; C. Marini Bettolo; R. Muni-Lofra; A. Manzur; V. Ayyar Gupta; V. Selby; F. Muntoni

P.56 **Urine titin as a novel biomarker for Duchenne muscular dystrophy**
M. Ishii; M. Nakashima; H. Kamiguchi; N. Zach; R. Kuboki; R. Baba; T. Hirakawa; K. Suzuki; M. Quinton

P.57 **Longitudinal changes in fat fraction histograms using quantitative MRI in Duchenne muscular dystrophy**
H. Reyngoudt; P. Baudin; E. Caldas de Almeida Araujo; B. Wong; P. Carlier; B. Marty

P.58 **MRI muscle segmentation in Duchenne muscular dystrophy (DMD): stepwise region of interest (ROI) contractions to minimize fat fraction variability**
M. Hammond; S. Murthy; J. Harris; B. Luna; F. Roche; M. Berger; F. Vincent; S. Zabbatino; R. Scheyer; L. Heinichen; S. Holland

P.59 **Associations between body composition estimates and motor function in ambulatory individuals with Duchenne muscular dystrophy**
M. Kiefer; E. Townsend; C. Goncalves; K. Shellenbarger; B. Wong

P.60 **Is there correlation between North Star ambulatory assessments and performance upper limb module in ambulant boys with Duchenne muscular dystrophy?**
M. Main; H. Mallender; N. Burnett; A. Sarkosy; F. Muntoni

P.61 **Imaging Mass Cytometry reveals new clues to understand the pathogenesis of Becker muscular dystrophy**
P. Piñol; J. Verdú-Díaz; C. Lawless; E. Fernández-Simón; D. McDonald; C. Domínguez-Gonzalez; A. Hernández-Laín; P. Rushton; A. Bowey; R. Charlton; M. Henderson; X. Suárez-Calvet; A. Filby; J. Díaz-Manera

P.62 **Increased skeletal muscle extracellular volume fraction in patients with Becker muscular dystrophy assessed by quantitative magnetic resonance imaging**
B. Marty; P. Baudin; Y. Fromes; K. Wahbi; H. Reyngoudt

P.63 **Comparison of brain volume reduction in boys with Duchenne muscular dystrophy treated with different corticosteroid regimes**
S. Geuens; J. Van Dessel; J. Lemiere; E. Niks; N. Goemans; H. Kan; L. De Waele; N. Doorenweerd

P.64 **Validity of remote evaluation of the North Star Ambulatory Assessment in patients with Duchenne muscular dystrophy**
L. Lowes; M. Iammarino; N. Reash; K. Giblin; L. Hu; L. Yu; S. Wang; L. Alfano; J. Mendell

P.65 **Consistency of changes in %-predicted forced vital capacity between real-world data and trial placebo arms in ambulatory Duchenne muscular dystrophy**
N. Goemans; C. McDonald; F. Muntoni; J. Signorovitch; G. Sajeev; N. Done; A. Manzur; B. Wong; C. Tian; E. Mercuri; C. He; D. Peterson; H. Akbarnejad; S. Ward; PRO-DMD-01 study investigators; cTAP

P.66 **Predicting trajectories of ambulatory function in Duchenne muscular dystrophy (DMD)**
F. Muntoni; J. Signorovitch; N. Goemans; A. Manzur; N. Done; G. Sajeev; E. Niks; L. Servais; V. Straub; I. de Groot; S. Ward; C. McDonald

P.67 **Development and evluation of a time to event endpoint for clinical trials in Duchenne muscular dystrophy (DMD)**
C. McDonald; F. Muntoni; J. Marden; N. Goemans; A. Gomez-Lievano; A. Zhang; S. Ward; J. Signorovitch

P.68 **Validation of a composite prognostic score for time to loss of ambulation in Duchenne muscular dystrophy**
C. McDonald; H. Gordish-Dressman; J. Signorovitch; G. Sajeev; M. Fillbrunn; M. Frean; S. Ward; N. Goemans; K. Vandenborne; E. Mercuri; F. Muntoni; .. Investigators for CINRG-DNHS; .. The PRO-DMD-01 Study; .. The Imaging DMD Study; .. The iMDEX Study; .. Collaborative Trajectory Analy

P.69 **Minimum clinically important difference in magnetic resonance biomarkers in DMD**
R. Willcocks; A. Barnard; S. Forbes; W. Triplett; J. Brandsema; E. Finanger; W. Rooney; D. Wang; D. Lott; C. Senesac; G. Walter; H. Sweeney; K. Vandenborne

P.70 **Rasch analysis of the PROMIS parent proxy item banks administered to caregivers of patients with Duchenne muscular dystrophy**
L. Lowes; N. Reash; L. Alfano; M. Iammarino; C. LeReun; I. Audhya; K. Gooch

P.71 **Vamorolone has less impact than daily prednisone or deflazacort on height and body mass index in patients with Duchenne muscular dystrophy (DMD)**
L. Ward; V. Rao; M. Leinonen; M. Guglieri; P. Clemens; R. Griggs; J. Mah; R. Finkel; N. Goemans; V. Straub; E. Smith; J. Haberlova; A. Childs; G. Baranello; E. Niks; P. Shieh; E. Hoffman; the VISION-DMD Investigators; FOR-DMD and CINRG investigator

P.72 **Motor delays are present in most boys with dystrophinopathies in infancy**
L. Lowes; N. Reash; N. Iammarino; M. Waldrop; K. Flanigan; C. Tsao; J. Mendell; A. Connolly; L. Alfano

P.73 **Structural damage in dystrophinopathies: a multimodal neuroimaging study**
M. Rabelo de Brito; T. Rezende; C. Iwabe; G. Conte; F. Franco da Graça; A. Nucci; F. Cendes; M. França Jr

VP.22 **Dystrophin transcript profile in urinary stem cells allows to study the impact of missense mutations**
M. Mietto; M. Neri; F. Ricci; R. Rossi; A. Margutti; V. Nagliati; R. Selvatici; A. Ferlini; M. Falzarano

VP.23 **Survey about the prevalence of urinary symptoms and abnormal renal function in adults with Duchenne muscular dystrophy (DMD)**
A. Pietrusz; M. Desikan; K. Koutroutsos; R. Quinlivan

VP.24 **T Cell-mediated immune response to dystrophin in Duchenne muscular dystrophy - A natural history study**
K. Anthony; P. Ala; F. Catapano; J. Meng; J. Domingos; M. Perry; V. Ricotti; K. Maresh; L. Phillips; V. Straub; M. Guglieri; L. Servais; A. Seferian; S. De Lucia; I. de Groot; Y. Krom; J. Verschuuren; E. Niks; T. Voit; J. Morgan; F. Muntoni

VP.25 **Muscle MRI in female carriers of Duchenne muscular dystrophy**
Y. Sun; B. Wong; K. Shellenbarger

VP.26 **Potential of the MyoSuit, a lightweight wearable lower-limb cable-actuated exoskeleton in patients with neuromuscular disorders: preliminary findings**
R. Feigean; C. Afroun; E. Gasnier; O. Benveniste; J. Hogrel; G. Bassez; D. Bachasson

VP.27 **Potential of the Keeogo+, a lightweight wearable powered assistive exoskeleton in patients with neuromuscular disorders: preliminary findings**
R. Feigean; C. Afroun; E. Gasnier; O. Benveniste; G. Bassez; J. Hogrel; D. Bachasson

VP.28 **Psychometric properties of muscle strength assessment by hand-held dynamometry in healthy adults: a reliability study**
M. Morin; L. J. Hébert; M. Perron; E. Petitclerc; S. Lake; E. Duchesne

16:00-16:20 **Metabolic Myopathies** (FP.16-19) – Ballroom Salon
Moderator: John Vissing

16:00-16:05 FP.16 **The effects of resistance exercise training on mitochondrial myopathy patients**
V. Di Leo; J. Newman; C. Lawless; F. Robertson; Y. Levy; J. Ochala; S. Pickett; G. Hudson; G S. Gorman; H A. Tuppen; A E. Vincent; O M. Russell

16:05-16:10 FP.17 **Long-term follow-up of cipaglucosidase alfa/miglustat in ambulatory patients with Pompe disease: An open-label phase I/II study (ATB200-02)**
B. Schoser; P. Kishnani; D. Bratkovic; P. Clemens; O. Goker-Alpan; X. Ming; M. Roberts; M. Vorgerd; K. Sivakumar; A. van der Ploeg; M. Goldman; J. Wright; F. Holdbrook; V. Jain; S. Sitaraman; Y. Wasfi; T. Mozaffar; B. Byrne

16:10-16:15 FP.18 **Modified ketogenic diet in patients with McArdle disease: a double-blind, placebo-controlled, cross-over study**
N. Løkken; M. Nielsen; M. Stemmerik; C. Ellerton; M. Macrae; K. Revsbech; B. Krett; G. Beha; R. Quinlivan; J. Vissing

16:15-16:20 FP.19 **Quantification of glycogen distribution in late-onset Pompe patients using 7 Tesla C13 NMR spectroscopy**
G. Beha; M. Stemmerik; V. Boer; A. Marsman; L. Jacobsen; E. Petersen; J. Vissing

16:00-17:30 **Metabolic Myopathies** (P.74-86) (VP.29-37) - Poster area - Ballroom B1-B2

P.74 **Using high-field magnetic resonance spectroscopy to measure muscle glycogen in patients with McArdle disease**
M. Stemmerik; G. Beha; V. Boer; A. Marsman; L. Jacobsen; E. Petersen; J. Vissing

P.75 **GM2 Gangliosidoses patient journey: results from interviews with late-onset GM2 patients and frontline treaters**
M. Lopshire; A. Flores; J. Burns; R. Gould; I. Batsu

P.76 **Gene variant and neuromuscular findings from a long-chain fatty acid oxidation disorder gene panel program**
D. Marsden; V. Miller; P. Baker II; O. Japalaghi; N. Longo; H. McLaughlin; K. Simmons; J. Yong; N. Miller

P.77 **Clinical presentation of two Korean patients with adolescent-onset very-long-chain acyl-CoA dehydrogenase deficiency**
H. Lee; U. Yun; H. Park; Y. Choi

P.78 **Skeletal myopathy or cardiomyopathy in glycogenin-1 deficiency - Two sides of the coin**
K. Visuttijai; C. Hedberg-Oldfors; N. Bermingham; D. Costello; E. Englund; O. Braun; A. Oldfors

P.79 **Late onset oculopharyngeal muscular dystrophy in a *POLG1*-related progressive external ophthalmoplegia (PEO), a diagnostic challenge**
G. Remiche; H. Kadhim; S. Lecompte; M. Meneri; D. Ronchi; G. Comi; S. Seneca; S. Capiau; H. Stepman; G. Smits

P.80 **Respiratory chain dysfunction in dermatomyositis is associated with mitochondrial DNA depletion**
C. Hedberg-Oldfors; U. Lindgren; K. Visuttijai; S. Roos; C. Thomsen; A. Oldfors

P.81 **Nutritional status and bone health in pediatric patients with low skeletal muscle mass**
M. Naume; C. Høi-Hansen; A. Born; M. Hørby; L. Borgwardt; J. Vissing; D. Stærk; M. Ørngreen

P.82 **Case-Control cardiopulmonary exercise testing for patients with neuromuscular disease**
T. Duong; D. Parker; V. Stevens; S. Dunaway Young; W. Tang; J. Myers; E. Ashley; M. Wheeler; J. Christle

P.83 **Molecular pathology of human PPP1R21 deficiency**
N. Meyer; N. Kohlschmidt; H. Lochmüller; U. Schara; L. Hannappel; A. Grüneboom; A. Schänzer; A. Hentschel; A. Gangfuss; A. Roos

P.84 **Gait analysis of patients with Pompe disease using a portable system**
M. Claramunt; S. Idelssonhn; M. James; M. Corti; V. Anton; B. Byrne; J. Díaz Manera

P.85 **Analysis of Juvenile onset Pompe disease patients included in the Spanish Pompe Registry**
R. Martínez Marín; J. Sánchez Caro; D. Reyes Leiva; A. Nascimento; N. Muelas; C. Dominguez; C. Paradas; M. Olivé; S. Pascual Pascual; M. Barba Romero; M. Gomez; M. Usón; R. Blanco; J. Barcena Llona; A. López de Munuain; A. Gutiérrez; A. Colomé; F. Pla-Junca; S. Segovia Simón; J. Díaz Manera

P.86 **Spanish Pompe registry: update of the 122 patients included**
R. Martínez Marín; D. Reyes Leiva; A. Nascimento; N. Muelas; C. Dominguez; C. Paradas; M. Olivé; J. Grau; M. Barba Romero; S. Pascual Pascual; R. Blanco Lago; M. Usón; A. Gutiérrez; J. Barcena Llona; A. Colomé; A. López de Munuain; F. Pla-Junca; S. Segovia Simón; J. Díaz Manera

VP.29 **Safety analysis of home-based enzyme replacement therapy with alglucosidase alfa in Pompe disease; a prospective study**
I. Ditters; A. van der Ploeg; N. van der Beek; J. van den Hout; H. Huidekoper

VP.30 **Neurofilament light as a biomarker for involvement of the brain in classic infantile Pompe patients**
M. Mackenbach; J. van den Dorpel; N. van der Beek; E. Willemse; D. Rizopoulos; C. Teunissen; A. van der Ploeg; J. van den Hout

VP.31 **10-year course of treatment with enzyme replacement therapy for childhood-onset Pompe disease**
A. Ishii; N. Mamada; H. Tsuji; A. Tamaoka

VP.32 **Living with Pompe disease in the UK: characterising the patient journey; burden on physical and emotional quality of life; and impact of COVID-19**
A. Muir; D. Hughes; L. Bashorum; V. Buxton; N. Johnson; G. McCaughey; P. Slade; N. Patel

VP.33 **Quantification of the burden, unmet needs, management, and COVID 19 impact of living with Pompe disease in the UK: results of an online patient survey**
V. Buxton; A. Muir; N. Johnson; G. McCaughey; P. Slade; D. Hughes; N. Patel

VP.34 **Two-year follow-up of muscle strength and function in patients with glycogen storage disease type IIIa**
V. Decostre; M. Masingue; P. Laforêt; R. Ben Yaou; P. Labrune; J. Hogrel

VP.35 **Muscle involvement in a Chinese patient with *TRNT1*-related disorder**
C. Wei; H. Xiong

VP.36 **Hypothyroidism impairs skeletal muscle regeneration through dysregulation of MuSCs cell cycle**
P. Aguiari; V. Villani; Y. Liu; G. Brent; L. Perin; A. Milanesi

VP.37 **Sleep deprivation induces aging like changes in antigravity muscles of young adult male wistar rats**
B. Sharma; A. Roy; J. Banerjee; K. Deepak; T. Nag; R. Netam; N. Akhtar; H. Mallick

16:20-16:30 **Immune mediated - and NMJ-related NMDs** (FP.20-21) – Ballroom
Moderator: Werner Stenzel

16:20-16:25 FP.20 **Serum metabolomics differentiates treatment response of Myasthenia Gravis clinical outcome measures**
H. Kaminski; L. Yaoxiang; M. Cheema; G. Wolfe; L. Kusner; I. Aban; P. Sikorski

16:25-16:30 FP.21 **Congenital myasthenic syndrome with Desmin aggregates: a novel association in recessive desminopathies due to a recurrent intronic *DES* mutation**
K. Polavarapu; R. Thompson; L. Matalonga; B. Nandeesh; S. Vengalil; V. Preethish-Kumar; S. Laurie; A. Nalini; H. Lochmüller

16:00-17:30 **Immune mediated - and NMJ-related NMDs** (P.87-98) (VP.38-49) - Poster area - Ballroom B1-B2

P.87 **Muscle cramps may be a clue for *GFPT1* gene related congenital myasthenic syndrome**
Ö. Yayıcı Köken; G. Öz Tunçer; B. Cavdarli; A. Ceylan; A. Aksoy; H. Topaloğlu

P.88 **Long-term follow-up of patients with chronic inflammatory demyelinating polyradiculoneuropathy (CIDP)**
L. Baisier; F. Daems; P. De Jonghe; J. Baets; A. Alonso-Jiménez

P.89 **Immune-mediated necrotizing myopathy associated with anti-SRP Antibodies: three cases in Korea**
H. Lee; U. Yun; S. Lee; H. Park; Y. Choi

P.90 **Understanding the role of *GFPT1* in congenital myasthenic syndromes**
S. Holland; D. O'Neil; S. Spendiff; H. Lochmuller

P.91 **Double seropositive inflammatory myositis with anti-PL-7 and anti-Mi-2 antibodies**
M. Lee; H. Im; M. Yoon; J. Lee

P.92 **LUMINESCE: Phase 3 study of satralizumab, a therapeutic recycling antibody targeting the IL-6 receptor, in patients with generalised myasthenia gravis**
H. Kaminski; C. Zhao; G. Meyer zu Horste; K. O'Connor; G. Klingelschmitt; P. Krumova; S. Bolt; I. Vodopivec; H. Murai

P.93 **Effect of rapamycine on quantitative MRI outcome measures in inclusion body myositis**
H. Reyngoudt; D. Bachasson; J. Hogrel; P. Baudin; Y. Allenbach; P. Carlier; O. Benveniste; B. Marty

P.94 **Clinical and pathologic features of clinically diagnosed inclusion body myositis (IBM) patients in Korea**
M. Kang; D. Kim; J. Shin

P.96 **Clinical characteristics of patients with seronegative myasthenia gravis**
R. Andersen; K. Axelsen; J. Vissing; N. Witting

P.97 **Clinical differences between ocular and generalized myasthenia gravis**
K. Axelsen; R. Andersen; J. Vissing; N. Witting

P.98 **Calculating the genetic prevalence of congenital myasthenic syndromes based on data from genomic databases**
R. Thompson; S. Rodger; K. Polavarapu; S. Laurie; L. Matalonga; S. Beltran; H. Lochmüller

VP.38 **A double-blinded, randomized, placebo-controlled phase II study of FcRn antagonist batoclimab in Chinese generalized myasthenia gravis patients**
C. Yan; R. Duan; H. Yang; H. Li; Z. Zou; H. Zhang; H. Zhou; X. Li; H. Zhou; L. Jiao; J. Chen; J. Yin; Q. Du; M. Lee; Y. Chen; X. Chen; C. Zhao

VP.39 **A new era for gMG management: Impact of continuing education on improving diagnosis and classification of gMG patients**
C. Drexel; E. Bixler; K. Kowalski; S. Masterson; J. Howard

VP.40 **Exploring barriers and facilitators to physical exercise in autoimmune myasthenia gravis: the MYaEX study**
S. Birnbaum; A. Archer; C. Stalens; J. Lejeune; J. Hogrel

VP.41 **Congenital myasthenic syndrome: natural history of an Italian cohort of patients**
A. Gallone; A. Pugliese; R. Brugnoni; I. Tramacere; S. Bonanno; M. Garibaldi; C. Rodolico; L. Maggi

VP.42 **Clinical characteristics, molecular genetics and long-term clinical outcomes in 43 patients with congenital myasthenia syndrome due to *RAPSYN* mutation**
S. Ramdas; P. Munot; P. Rodríguez Cruz; S. Alabaf; S. Robb; S. Jayawant; D. Beeson; H. Jungbluth; J. Palace

VP.43 **Immune-mediated necrotizing myopathies: clinical-serological features of a large Italian cohort of patients**
S. Bonanno; L. Maggi; -. IMNM Italian Study Group

VP.44 **Juvenile Anti-PM Scl75 myositis with necrosis, phagocytosis, and endomysial fibrosis**
A. Cotta; E. Carvalho; A. Cunha Jr.; J. Valicek; M. Navarro; J. Paim; M. Lima; A. Cauhi; M. Quintero; A. Reis

VP.45 **Clinical features of anti-mitochondrial M2 antibody-positive myositis: case series of 17 patients**
A. Nagai; T. Nagai; H. Yaguchi; S. Fujii; K. Horiuchi; S. Ura; S. Shirai; I. Iwata; M. Matsushima; T. Anzai; I. Yabe

VP.46 **Dermatomyositis-specific autoantibodies and muscle MRI findings**
H. Une; S. Hayashi; S. Noguchi; I. Nishino

VP.47 **Interleukin 31 (IL-31) inhibition as a trigger for an immune-mediated myopathy?**
M. Winkler; K. Kappes-Horn; J. Reimann

VP.48 **Clinicopathological characteristics of 105 patients with idiopathic inflammatory myopathy based on muscle specific antibodies**
A. Yamanaka; N. Eura; T. Shiota; M. Yamaoka; Y. Nishimori; N. Iguchi; M. Ozaki; H. Nanaura; N. Iwasa; T. Kiriyama; T. Izumi; H. Kataoka; K. Sugie

VP.49 **Total sleep deprivation leads to changes in neuromuscular junction of soleus muscle in male wistar rats**
B. Sharma; A. Roy; A. Singh; M. Tripathi; J. Banerjee; R. Netam; N. Akhtar; T. Nag; K. Deepak; H. Mallick

16:30-16:45 **SMA – Therapy** (FP.22-24) – Ballroom
Moderator: Nathalie Goemans

16:30-16:35 FP.22 **Results from the end of Part A of the ongoing 3-part DEVOTE study to explore higher doses of nusinersen in SMA**
J. Day; R. Finkel; S. Pascual Pascual; M. Ryan; E. Mercuri; D. De Vivo; J. Montes; J. Gurgel-Giannetti; G. Gambino; C. Makepeace; R. Foster; V. Irzhevsky; Z. Berger

16:35-16:40 FP.23 **Bulbar function for patients with spinal muscular atrophy type 1 following onasemnogene abeparvovec**
K. McGrattan; R. Shell; R. Hurst-Davis; S. Dunaway Young; E. O'Brien; A. Lavrov; S. Wallach; N. LaMarca; S. Reyna; B. Darras

16:40-16:45 FP.24 **RAINBOWFISH: Preliminary efficacy and safety data in risdiplam-treated infants with presymptomatic spinal muscular atrophy (SMA)**
R. Finkel; M. Farrar; D. Vlodavets; E. Zanoteli; M. Al-Muhaizea; L. Nelson; A. Prufer; L. Servais; Y. Wang; C. Fisher; M. Gerber; K. Gorni; H. Kletzl; L. Palfreeman; R. Scalco; E. Bertini

16:00-17:30

SMA – Therapy (P.99-118) (VP.50-56b) - Poster area - Ballroom B1-B2

P.99 **Suboccipital puncture for administration of Nusinersen. Description of 2 cases**
A. Alonso-Jiménez; M. Niekel; J. Baets

P.100 **Rationale/design of the phase 3b ASCEND study of investigational higher dose nusinersen in participants with SMA previously treated with risdiplam**
B. Darras; T. Hagenacker; R. Finkel; E. Mercuri; J. Montes; N. Kuntz; M. Farrar; V. Sansone; Z. Berger; D. MacCannell; C. Shen; A. Paradis; J. Bohn; J. Wagner; K. Somera-Molina

P.101 **Baseline characteristics/initial safety in RESPOND: phase 4 study of nusinersen in children with SMA who previously received onasemnogene abeparvovec**
C. Proud; J. Parsons; R. Masson; J. Brandsema; R. Finkel; K. Swoboda; E. Finanger; Y. Liu; C. Makepeace; A. Paradis; Z. Berger; J. Wagner; K. Somera-Molina

P.102 **Apitegromab in SMA: An analysis of multiple efficacy endpoints in the TOPAZ extension study**
T. Crawford; B. Darras; J. Day; D. Barrett; G. Song; J. O'Neil; N. Kertesz; S. Bilic; J. Patel; G. Nomikos; Y. Chyung

P.103 **Onasemnogene abeparvovec (OA) treatment outcomes by patient weight at infusion: Initial findings from the RESTORE registry**
L. Servais; K. Benguerba; D. De Vivo; J. Kirschner; E. Mercuri; F. Muntoni; C. Proud; E. Tizzano; S. Quijano-Roy; K. Saito; D. Raju; N. LaMarca; R. Sun; F. Anderson; E. Faulkner; R. Finkel

P.104 **Treatments and outcomes for patients with spinal muscular atrophy (SMA) type 2: findings from RESTORE registry**
L. Servais; K. Benguerba; D. De Vivo; J. Kirschner; F. Muntoni; C. Proud; E. Tizzano; S. Quijano-Roy; I. Desguerre; K. Saito; D. Raju; N. LaMarca; R. Sun; F. Anderson; E. Faulkner; R. Finkel

P.105 **Safety and effectiveness of onasemnogene abeparvovec (OA) alone or with other disease-modifying therapies (DMTs): findings from RESTORE**
L. Servais; K. Benguerba; D. De Vivo; J. Kirschner; F. Muntoni; C. Proud; E. Tizzano; K. Saito; D. Raju; N. LaMarca; R. Sun; F. Anderson; E. Faulkner; R. Finkel

P.107 **Nusinersen effects on SMA-related fatigue: clinical and neuromuscular jitter follow-up in a late-onset patient**
F. Franco da Graca; C. Iwabe; M. França Jr

P.108 **Clinical and electrophysiological evaluation of fatigue in adult patients with spinal muscular atrophy (SMA)**
C. Iwabe; F. Franco da Graca; A. Nucci; M. França Jr

P.109 **FIREFISH Parts 1 and 2: 36-month safety and efficacy of risdiplam in Type 1 spinal muscular atrophy (SMA)**
L. Servais; G. Baranello; O. Boespflug-Tanguy; J. Day; N. Deconinck; A. Klein; R. Masson; M. Mazurkiewicz-Bełdzińska; E. Mercuri; K. Rose; D. Vlodavets; H. Xiong; E. Zanoteli; M. El-Khairi; M. Gerber; K. Gorni; H. Kletzl; L. Palfreeman; A. Dodman; E. Gaki; B. Darras

P.110 **JEWELFISH: 24-month safety and pharmacodynamic data in non-treatment-naïve patients with spinal muscular atrophy (SMA)**
C. Chiriboga; C. Bruno; T. Duong; D. Fischer; J. Kirschner; M. Scoto; E. Mercuri; M. Gerber; K. Gorni; H. Kletzl; I. Carruthers; C. Martin; T. Gidaro; F. Muntoni

P.112 **Impact of nusinersen on respiratory progression in paediatric patients with spinal muscular atrophy type 2 and non-ambulant type 3**
F. Trucco; H. Weststrate; D. Ridout; M. Scoto; A. Rohwer; G. Coratti; M. Main; M. Pane; S. Messina; A. D'Amico; C. Bruno; D. De Vivo; B. Darras; J. Day; G. Baranello; V. Sansona; E. Bertini; R. Finkel; E. Mercuri; F. Muntoni

P.113 **Safety update: Risdiplam clinical trial development program**
C. Chiriboga; L. Servais; G. Baranello; B. Darras; J. Day; N. Deconinck; M. Farrar; R. Finkel; E. Bertini; J. Kirschner; R. Masson; M. Mazurkiewicz-Bełdzińska; D. Vlodavets; S. Bader-Weder; K. Gorni; B. Jaber; T. McIver; G. Papp; R. Scalco; E. Mercuri

P.114 **SUNFISH parts 1 and 2: 3-year efficacy and safety of risdiplam in types 2 and 3 spinal muscular atrophy (SMA)**
J. Day; N. Deconinck; E. Mazzone; A. Nascimento; M. Oskoui; K. Saito; C. Vuillerot; G. Baranello; O. Boespflug-Tanguy; N. Goemanns; J. Kirschner; A. Kostera-Pruszczyk; L. Servais; J. Braid; M. Gerber; K. Gorni; C. Martin; R. Scalco; W. Yeung; E. Mercuri

P.115 **Functional follow-up of patients with spinal muscular atrophy treated post-symptomatically with spinraza: Clinical trial versus real life**
C. Lilien; A. Marinescu; K. Aragon-Gawinska; N. Deconinck; L. de Waele; T. Duong; N. Goemans; L. Szabo; L. Médard; L. Servais

P.116 **Evaluating 2-3 year responses to disease modifying treatment in adults with spinal muscular atrophy**
T. Duong; W. Tang; S. Dunaway Young; D. Parker; C. Wolford; J. Sampson; J. Day

P.117 **Collection of real-world evidence of nusinersen treatment for SMA patients through a national registry: description of the paediatric cohort in the UK**
A. Rohwer; M. Main; S. Wadsworth; A. Wolfe; M. Madden; E. Cavalcante; S. Samsuddin; P. Munot; A. Manzur; G. Baranello; M. Scoto; F. Muntoni

P.118 **The effect of nusinersen on function and muscle strength in the upper limb in a cohort of children with spinal muscular atrophy (SMA) type 2 and 3**
E. Milev; A. Wolfe; M. Main; G. Baranello; M. Scoto; F. Muntoni

VP.50 **Management of spinal muscular atrophy in the preterm infant: A case study**
E. Nigro

VP.51 **Impact of nusinersen on caregiver experience and health-related quality of life (HRQoL) when initiated in the presymptomatic stage of SMA in NURTURE**
J. Kirschner; T. Crawford; M. Ryan; R. Finkel; K. Swoboda; D. De Vivo; E. Bertini; H. Hwu; V. Sansone; A. Pechmann; J. Montes; D. Krasinski; R. Chin; Z. Berger; C. Zhu; S. Raynaud; A. Paradis; N. Johnson

VP.52 **Identification of a novel cytokine profile in serum and CSF of pediatric and adult SMA patients and its modulation upon nusinersen treatment**
S. Bonanno; P. Cavalcante; E. Salvi; E. Giagnorio; C. Malacarne; M. Cattaneo; F. Andreetta; A. Venerando; V. Pensato; C. Gellera; R. Zanin; C. Dosi; R. Masson; R. Mantegazza; L. Maggi; S. Marcuzzo

VP.53 **Image-guided nusinersen intrathecal injections in SMA patients: a single centre experience**
H. Weststrate; L. Davies; F. Robertson; F. Muntoni; A. Rennie; A. Rose; A. Manzur; G. Baranello; P. Munot; S. Craig; M. Scoto

VP.54 **Amifampridine safety and efficacy in spinal muscular atrophy ambulatory patients: a randomized placebo-controlled, crossover, phase 2 trial**
S. Bonanno; R. Giossi; R. Zanin; V. Porcelli; C. Iannacone; G. Baranello; G. Ingenito; S. Iyadurai,; z. Stevic; S. Peric; L. Maggi

VP.55 **Fatigue, pain, breathing, voice, fatigability, sleep, rest and vulnerability as meaningful outcomes in SMA care: the patients´and caregivers' voice**
M. Povedano; J. Vázquez-Costa; I. Pitarch; M. López-Lobato; J. Medina; J. Fernández-Ramos; M. Lafuente-Hidalgo; R. Rojas-García; J. Caballero-Caballero; I. Málaga; J. Eirís; M. De Lemus; M. Cattinari; M. Madruga-Garrido; M. Branas; R. Cabello-Moruno; P. Díaz-Abós; A. Terrancle; J. Maurino; P. Rebollo

VP.56 **Perception of treatment efficacy among pediatric neurologists caring for patients with spinal muscular atrophy**
G. Saposnik; A. Camacho; P. Díaz-Abós; M. Brañas; V. Sánchez-Menéndez; R. Cabello-Moruno; M. Terzaghi; J. Mauriño; I. Malaga

VP.56a **Real-world assessment of onasemnogene abeparvovec treatment in patients with spinal muscular atrophy: RESTORE/post-marketing surveillance in Japan**
K. Saito; R. Nagao; K. Tsuchida; R. Teshima; K. Kawase

VP.56b **Nusinersen in children and adults with spinal muscular atrophy in Argentina: real world experience**
A. Dubrovsky; L. Mesa; J. Corderi; D. Flores; M. Morosini; C. Bolaño; A. Jauregui; L. Pirra; G. Vazquez; F. Chloca

17:30-19:00	**Symposium 3** - Ballroom	**Symposium 4** - Argyle

Thursday 13th October 2022

07:00	**Conference desk opens**	
07:30-09:00	**Symposium 5** - Ballroom	**Symposium 6** - Argyle
09:15-10:45	**Neuropathies and Non-5q Motor Neuron Disease 1;** Invited lectures (I.05-06) - Ballroom *Moderators: Bernard Brais and Peter Van den Bergh*	
09:15-09:45	I.05 **Preclinical testing of emerging therapies for inherited peripheral neuropathies** R. Burgess	
09:45-10:15	I.06 **Late onset forms of inherited neuropathies** M. Auer-Grumbach	
	Selected oral presentations **Neuropathies and Non-5q Motor Neuron Disease 1** (O.07-08) - Ballroom *Moderators: Bernard Brais and Peter Van den Bergh*	
10:15-10:30	O.07 **TDP43 accumulates in intramuscular nerve bundles of ALS patients** T. Kurashige; H. Morino; T. Murao; Y. Izumi; T. Sugiura; K. Kuraoka; H. Kawakami; T. Torii; H. Maruyama	
10:30-10:45	O.08 **Biallelic variants in the mitochondrial form of phosphoenolpyruvate carboxykinase (*PCK2*) cause a recessive form of Charcot-Marie-Tooth disease** N. Sondheimer; A. Aleman; J. Cameron; H. Gonorazky; N. Sabha; P. Oliveira; A. Wahedl; D. Wang; K. Amburgey; M. Shy; J. Dowling	
10:45-11:15	Morning refreshments, exhibition and posters - Ballroom Salon and Pre Function Space	
11:15-13:15	**Neuropathies and Autosomal Dominant LGMD;** Invited lectures (I.07-08) - Ballroom *Moderators: Carmen Paradas Lopez and Jodi Warman-Chardon*	
11:15-11:45	I.07 **Clinical trial readiness across the lifespan in Charcot-Marie-Tooth disease** K. Eichinger	
11:45-12:15	I.08 **Motor neuron disease caused by excess sphingolipid synthesis** P. Mohassel; T. Dunn; C. Bönnemann	
	Selected oral presentations **Neuropathies and Autosomal Dominant LGMD** (O.09-12) - Ballroom *Moderators: Carmen Paradas Lopez and Jodi Warman-Chardon*	
12:15-12:30	O.09 **Gene therapy of spinal muscular atrophy with progressive myoclonic epilepsy (SMA-PME)** J. Denard; M. Marinello; V. Latournerie; D. Bonnin; M. Derome; S. Martin; J. Medin; A. Buj Bello	
12:30-12:45	O.10 **First-in-human intrathecal gene transfer study for giant axonal neuropathy: preliminary review of long-term efficacy and safety** D. Bharucha-Goebel; D. Saade; J. Todd; G. Norato; M. Jain; M. Waite; D. Armao; A. Foley; T. Lehky; G. Averion; Y. Hu; P. Mohassel; A. Hoke; T. DeLong; N. Acquaye; L. Hinkley; J. Chichester; C. Mendoza; A. Soldatos; S. Gray; C. Bönnemann	
12:45-13:00	O.11 **Identification of a novel heterozygous *DYSF* variant in a large family with a dominantly-inherited dysferlinopathy** C. Folland; R. Johnsen; A. Botero Gomez; D. Trajanoski; M. Davis; U. Moore; V. Straub; R. Barresi; M. Guglieri; H. Hayhurst; A. Schaefer; N. Laing; P. Lamont; G. Ravenscroft	
13:00-13:15	O.12 **Novel functional test to distinguish between variants causing dominant and recessive forms of calpainopathy** A. Salvi; S. Courrier; M. Cerino; N. Da Silva; M. Krahn; M. Bartoli; S. Gorokhova	
13:15-13:30	Lunch, exhibition and posters - Ballroom Salon and Pre Function Space	
13:15-14:45	***Neuromuscular Disorders* Editorial Board Meeting** - Meeting Room 603-604	

13:30-15:00	**Symposium 8** - Ballroom **Change to programme**
15:00-18:00	**Poster viewing** - Poster area - Ballroom B1-B2
18:00-20:00	**Reception (pre-registration required)** - Pier 21

Friday 14th October 2022

06:45	**Registration desk opens**	
07:00-8:30	**Symposium 7** - Ballroom	Please note that **Symposium 8** has been rescheduled to **Thursday 13 October 13:30-15:00**. Lunch will be available in the Ballroom during this session
08:45-10:15	**The Development of Therapeutic Approaches 1;** Invited lectures (I.09-10) - Ballroom *Moderators: John Brandsema and Grace Yoon*	
08:45-09:15	I.09 **Induced pluripotent stem cells for modeling neuromuscular disorders: development of disease-specific assays, live cells functional testing and drug design** M. Delourme, C. Laberthonnière, S. Testa, L. Caron, F. Magdinier	
09:15-09:45	I.10 **Gene therapy and other novel treatment approaches for CMT** D. Pareyson	
	Selected oral presentations **The Development of Therapeutic Approaches 1** (O.13-14) - Ballroom *Moderators: John Brandsema and Grace Yoon*	
09:45-10:00	O.13 **Givinostat in DMD: results of the Epidys Study** Mercuri E, Vilchez J, Boespflug-Tanguy O, Zaidman C, Mah J, Goemans N, Müller-Felber W, Niks E, Munell F, Schara U, Bertini E, Comi G, Mathews K, Servais L, Vandenborne K, Cazzaniga S, Coceani N, Bettica P, McDonald C	
10:00-10:15	O.14 **EEV-Conjugated PMO results in nuclear foci reduction and aberrant splicing correction in myotonic dystrophy cell and animal models** M. Girgenrath; N. Estrella; A. Hicks; X. Shen; M. Wysk; M. Kheirabadi; M. Streeter; W. Lian; N. Liu; S. Blake; C. Brennan; N. Li; V. Batagui; K. Oye; N. Gao; D. Wang; Z. Qian; N. Sethuraman	
10:15-10:45	Morning refreshments, exhibition and posters - Ballroom Salon and Pre Function Space	
10:45-12:45	**The Development of Therapeutic Approaches 2;** Invited lectures (I.11-12) - Ballroom *Moderators: Maryam Oskoui and Jiri Vajsar*	
10:45-11:15	I.11 **Directed evolution of a family of AAV capsid variants enabling potent muscle-directed gene delivery across species** S. Tabebordbar; K. Lagerborg; S. Ye; A. Stanton; E. King; L. Tellez; A. Krunnfusz; S. Tavakoli; J. Widrick; K. Messemer; E. Troiano; B. Moghadaszadeh; B. Peacker; K. Leacock; N. Horwitz; A. Beggs; A. Wagers; P. Sabeti	
11:15-11:45	I.12 **Modulating muscle stem cells to enhance regeneration to ameliorate DMD disease progression** M. Rudnicki	
	Selected oral presentations **The Development of Therapeutic Approaches 2** (O.15-18) - Ballroom *Moderators: Maryam Oskoui and Jiri Vajsar*	
11:45-12:00	O.15 **Muscular MRI pattern recognition for muscular dystrophies: The era of artificial intelligence beyond a systematic review** I. Alawneh; H. Gonorazky; S. Alawneh	

12:00-12:15	O.16 **Single cell RNA sequencing study of FAPS obtained from muscle samples of DMD patients reveals new pathogenic pathways of the muscle degeneration process** X. Suarez Calvet; E. Fernandez Simon; P. Pinol Jurado; A. Unsworth; J. Alonso Perez; M. Schiava; R. Queen; S. Lopez Fernandez; G. Pons; I. Mathews; P. Rushton; D. Cox; A. Bowey; M. Henderson; R. Charlton; C. Ortez; D. Natera; C. Jimenez Mallebriera; A. Nascimento; J. Díaz Manera
12:15-12:30	O.17 **Mining extracellular vesicles for novel RNA-based therapeutic agents in Duchenne muscular dystrophy** R. Rogers; A. Rannou; J. Alfaro; L. Sanchez; E. Marbán
12:30-12:45	O.18 **Preliminary results from MLB-01-003: an open label phase 2 study of BBP-418 in patients with Limb-girdle muscular dystrophy type 2I** A. Harper; R. Langeslay; H. Rodriguez; A. Hutchaleelaha; K. Kelley; M. Lynn; D. Sproule
12:45-14:30	Lunch, exhibition and posters - Ballroom Salon and Pre Function Space
13:00-14:00	**Sponsor Meeting** - Meeting Room 503
14:30-16:00	**Poster session 3** (FP.25-36) - Flash Poster Presentation areas - Ballroom or Ballroom Salon (P.119-183) (VP.58-71) - Poster area - Ballroom B1-B2 ***(Please note that this session will not be live streamed for virtual delegates)***
14:30-14:50	**DMD - Trials & Treatments** (FP.25-28) - Ballroom *Moderator: Kevin Flanigan*
14:30-14:35	FP.25 **Contracture management in ambulant boys with Duchenne muscular dystrophy (DMD)** D. Moat; M. McCallum; R. Muni-Lofra; K. Wong; J. Michell-Sodhi; M. James; D. Michura; M. Richardson; G. Carden; C. Hall; K. Frith; S. Fitzimmons; C. Marini-Bettolo; A. Mayhew
14:35-14:40	FP.26 **Early effect of corticosteroids on functional outcomes in young patients with Duchenne Muscular dystrophy within the first 18 months of treatment** S. Marianela; J. Broomfield; K. Abrams; M. McDermott; W. Martens; S. Gregory; A. Mayhew; C. McDonald; R. Griggs; M. Guglieri
14:40-14:45	FP.27 **Results of a double-blind cross-over trial of vamorolone in DMD: a safer alternative to corticosteroids** E. Hoffman; M. Guglieri; P. Clemens; S. Perlman; E. Smith; I. Horrocks; R. Finkel; J. Mah; N. Deconinck; N. Goemans; J. Haberlova; V. Straub; A. Harper; R. Webster; H. McMillan; G. Baranello; S. Spinty; A. Childs; K. Selby; J. Vilchez-Padilla; E. Niks
14:45-14:50	FP.28 **IGNITE DMD phase I/II study of SGT-001 microdystrophin gene therapy for DMD: Long-term outcomes and expression update** R. Donisa Dreghici; S. Redican; J. Lawrence; K. Brown; F. Wang; J. Gonzalez; J. Schneider; C. Morris; P. Shieh; B. Byrne
14:30-16:00	**DMD - Trials & Treatments** (P.119-134a) (VP.58-60) - Poster area - Ballroom B1-B2
	P.119 **Home infusion for antisense oligonucleotide therapy** E. Romano; Ç. Yanar Ayanoglu; T. Coskun; G. Eser; H. Topaloğlu
	P.120 **Unlocking the potential of oligonucleotide therapeutics for Duchenne muscular dystrophy through enhanced delivery** M. Mellion; J. McArthur; A. Holland; S. Gunnoo; S. Ching; R. Johnson; C. Irwin; P. Lonkar; S. Bracegirdle; N. Svenstrup; J. Goyal; C. Godfrey; J. Larkindale
	P.121 **Dnm2 reduction combined with dystrophin re-expression ameliorates the myopathic phenotype observed in the D2-mdx model of Duchenne muscular dystrophy** A. Menuet; S. Buono; A. Robé; S. Chhor; L. Eyler; J. Becker; S. Colombo; B. Cowling
	P.122 **Comparative safety and efficacy of different corticosteroid regimens in boys with Duchenne muscular dystrophy: results of a randomized controlled trial** M. Guglieri; M. McDermott; K. Bushby; K. Hart; R. Tawil; W. Martens; B. Herr; E. McColl; C. Speed; J. Wilkinson; J. Kirschner; W. King; M. Eagle; M. Brown; W. Willis; R. Griggs
	P.123 **A Phase I/II study of NS-089/NCNP-02, Exon 44 skipping drug, in patients with Duchenne muscular dystrophy** H. Komaki; E. Takeshita; K. Kunitake; Y. Shimizu-Motohashi; M. Sasaki; C. Yonee; S. Maruyama; E. Hida; D. Matsubara; T. Hatakeyama; Y. Muashige; Y. Aoki

P.124 **EDG-5506 targets fast skeletal myosin and reduces muscle damage biomarkers in a phase 1 trial in Becker muscular dystrophy (BMD)**
J. Donovan; N. Kilburn; G. Gordon; B. Barthel; M. DuVall; A. Bronson; A. Russell; C. Sherman; M. Evanchik

P.125 **DMD Hub: A UK network enabling trials in Duchenne muscular dystrophy**
E. Heslop; P. Cammish; M. McNiff; K. Pegg; A. Irvin; E. Reuben; A. Johnson; A. Gaeta; C. Turner; R. Fischer; H. Peay; F. Muntoni; A. Childs; V. Straub; M. Guglieri

P.126 **Genotype-unmatched controls are feasible for drug development in Duchenne muscular dystrophy (DMD)**
F. Muntoni; J. Signorovitch; M. Frean; M. Fillbrunn; G. Sajeev; S. Ward; C. McDonald; N. Goemans; E. Niks; B. Wong; L. Servais; V. Straub; I. de Groot; M. Chesshyre; C. Tian; A. Manzur; E. Mercuri; A. Aartsma-Rus; Study Groups

P.127 **A multi-disciplinary, independent expert approach to improve translational research in NMDs at all stages of the pipeline: developments in the TACT model**
C. Turner; A. Aartsma-Rus; D. Allison; A. De Luca; J. Lee; L. Robertson; V. Straub

P.128 **Integrated analyses of data from clinical trials of delandistrogene moxeparvovec in DMD**
C. Zaidman; P. Shieh; C. Proud; C. McDonald; J. Day; S. Mason; M. Guridi; L. Hu; L. Yu; C. Reid; E. Darton; C. Wandel; J. Richardson; J. Malhotra; T. Singh; L. Rodino-Klapac; J. Mendell

P.129 **One-year data from ENDEAVOR, a phase 1b trial of delandistrogene moxeparvovec in boys with DMD**
C. Zaidman; C. Proud; C. McDonald; S. Mason; M. Guridi; S. Wang; C. Reid; E. Darton; C. Wandel; S. Lewis; J. Malhotra; D. Griffin; R. Potter; L. Rodino-Klapac; J. Mendell

P.130 **RGX-202: an investigational AAV8 gene therapy coding for a novel microdystrophin as a treatment for Duchenne muscular dystrophy**
N. Dastgir; P. Falabella; C. Qiao; S. Kim; N. Buss; M. Fiscella; S. Pakola; O. Danos

P.131 **Building a FORCETM platform-based DMD franchise for the treatment of individuals with mutations amenable to exon skipping**
C. Desjardins; R. Venkatesan; E. O'Donnell; J. Hall; R. Russo; S. Spring; K. Tang; J. Davis; T. Weeden; S. Zanotti; O. Beskrovnaya

P.132 **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; D. Wolf; J. Malhotra; R. Chu; E. Darton; E. Mercuri

P.133 **Daily regimens of prednisone, deflazacort and vamorolone improve motor function similarly in patients with Duchenne muscular dystrophy**
C. McDonald; E. Henricson; M. Leinonen; A. Linden; M. Guglieri; P. Clemens; R. Griggs; P. Shieh; S. Horrocks; J. Mah; R. Finkel; N. Goemans; V. Straub; M. Ryan; H. McMillan; S. Spinty; E. Hoffman

P.134 **Real-world outcomes of exon skipping therapy use in patients with Duchenne muscular dystrophy: experience at a single, large tertiary care center**
A. Yaworski; T. Duong; J. Low; R. Gee; K. Watson; M. Buu; B. Kaufman; J. Klotz; J. Day; J. Guzman; C. Tesi Rocha

P.134a **Phase 1/2a trial of delandistrogene moxeparvovec in patients with DMD: 4-year update**
J. Mendell; Z. Sahenk; K. Lehman; L. Lowes; N. Reash; M. Iammarino; L. Alfano; S. Lewis; K. Church; R. Shell; R. Potter; D. Griffin; E. Pozsgai; M. Hogan; L. Hu; S. Mason; E. Darton; L. Rodino-Klapac

VP.58 **Golodirsen induced DMD transcripts localization and dystrophin production in 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

VP.59 **A single-arm, open-label, multicenter study of tranilast for advanced heart failure in patients with muscular dystrophy**
T. Matsumura; H. Hashimoto; M. Sekimizu; A. Saito; M. Asakura; K. Kimura; Y. Iwata

VP.60 **Every breath counts! Inspiratory muscle training in children with neuromuscular diseases: a cross-over randomised controlled trial**
A. Human; L. Corten; E. Lozano-Ray; B. Morow

14:30-14:40 **FSHD** (FP.29-30) – Ballroom Salon
Moderator: Carmen Paradas

14:30-14:35 FP.29 **AAV-CRISPR-Cas13 gene therapy for FSHD: DUX4 gene silencing efficacy and immune responses to Cas13b protein**
A. Rashnonejad; G. Amini-Chermahini; N. Taylor; A. Fowler; E. Kraus; O. King; S. Harper

14:35-14:40 FP.30 **TREAT-NMD FSHD Global Registry Network: a collaboration of neuromuscular and FSHD patient registries**
B. Porter; N. Bennett; D. Allison; C. Campbell; M. Guglieri; A. Ambrosini; R. Tupler

14:30-16:00 **FSHD** (P.135-144b) (VP.61-62) - Poster area - Ballroom B1-B2

P.135 **Safety and tolerability of losmapimod for the treatment of FSHD**
J. Shoskes; V. Ramana; M. Mellion

P.136 **Design of Reach: Phase 3 randomized, double-blind, placebo-controlled, 48-week study of the efficacy and safety of losmapimod in FSHD**
R. Tawil; J. Han; L. Wang; J. Vissing; B. van Engelen; J. Statland; M. Mellion; J. Shoskes; C. Morabito; J. Jiang; J. Webster

P.137 **Reachable workspace to evaluate efficacy of losmapimod in subjects with FSHD in two phase 2 studies**
R. Tawil; ReDUX4 Study Group

P.138 **Annualized rates of change from a phase 2, randomized, double-blind, placebo-controlled, 48-week study of losmapimod in subjects with FSHD: ReDUX4**
R. Tawil; ReDUX4 Study Group

P.139 **A cross sectional study of genetically confirmed cohort of facioscapulohumeral muscular dystrophy (FSHD) in the Indian population**
V. Vishnu; R. Lemmers; E. Bugiardini; A. Reyaz; S. Efthymiou; S. van der Maarel; R. Bhatia; R. Pitceathly; P. Srivastava; M. Hanna

P.140 **Understanding the patients' journey pre- and post-diagnosis of facioscapulohumeral muscular dystrophy (FSHD): a real-world retrospective data analysis**
C. Konersman; K. Munoz; R. Brook; N. Kleinman; K. DiTrapani; B. McEvoy; A. Peters; C. Chen; M. Stahl

P.141 **Manoeuvre study design: a study of GYM329 (RO7204239) in patients with facioscapulohumeral muscular dystrophy (FSHD)**
J. Vissing; K. Eichinger; J. Morrow; J. Statland; G. Tasca; A. Dodman; B. Jaber; H. Kletzl; T. McIver; R. Scalco; W. Yeung; E. Gaki; K. Wagner

P.142 **Improving FSHD RNAi gene therapy using myotropic MyoAAVs**
L. Wallace; T. Riley; M. Guggenbiller; G. Amini Chermahini; S. Harper

P.143 **Investigation of human bone marrow mesenchymal stem cell-derived extracellular vesicles as therapeutic agents for Facioscapulohumeral muscular dystrophy**
L. Wallace; S. Harper; N. Saad

P.144 **Developing Cas13-ADAR-mediated *DUX4* mRNA editing as a prospective therapy for FSHD**
N. Saljoughian; L. Rizzotto; Y. Sezgin; H. Faraji; L. Wallace; M. Naeimi Kararoudi; D. Palmieri; S. Harper

P.144a **Muscle ultrasound in an open-label study of Losmapimod in subjects with FSHD1**
J. Kools; N. Voermans; K. Mul; J. Jiang; J. Shoskes; K. Marshall; M. Mellion; B. van Engelen; M. Karllson

P.144b **Feasibility of measuring functional performance of FSHD patients using wearable sensors to quantify physical activity**
J. Kools; N. Voermans; K. Mul; M. Mellion; J. Jiang; J. Shoskes; K. Marshall; D. Jackson; Y. Zhao; A. Tarachandani; J. Figueredo; D. Eggenspieler; B. van Engelen

VP.61 **An AAV-shRNA *DUX4*-based therapy to treat Facioscapulohumeral muscular dystrophy (FSHD)**
V. Mariot; E. Sidlauskaite; L. Le Gall; E. Corbex; J. Dumonceaux

VP.62 **Dux4 expression turn on the myogenic program in MSC**
O. Serbina; E. Kiseleva; Y. Vassetzky

14:40-14:50 **Other myopathies & muscular dystrophies** (FP.31-32) – Ballroom Salon
Moderator: Gisèle Bonne

14:40-14:45 FP.31 ***ANXA11* related adult-onset muscular dystrophy in Greek families**
M. Johari; G. Papadimas; C. Papadopoulos; S. Xirou; M. Savarese; P. Hackman; B. Udd

14:45-14:50 FP.32 ***BAG3* p.P209L variant leads to changes in nuclear and actomyosin dynamics and impairment of the transmission of mechanical signals**
R. Robertson; M. Dicaire; J. Lavoie; B. Brais

14:30-16:00 **Other myopathies & muscular dystrophies** (P.145-158) (VP.65-70) - Poster area - Ballroom B1-B2

P.145 **Identification of potential genetic modifiers underlying phenotypic variability in a French family with striated muscle laminopathies**
L. Benarroch; A. Bertrand; M. Beuvin; I. Nelson; N. Naouar; F. Simonet; C. Dina; C. Pionneau; J. Schott; R. Ben Yaou; G. Bonne

P.146 **Characterising the molecular consequences of *LMNA*-related congenital muscular dystrophy in patient myoblasts**
E. Storey; I. Holt; S. Owen; S. Synowsky; S. Shirran; G. Morris; H. Fuller

P.147 **Deflazacort treatment in *LMNA*-related congenital muscular dystrophy: an ongoing Italian cohort pilot study**
G. Ricci; L. Maggi; A. D'Amico; C. Fiorillo; E. Schirinzi; A. Pini; E. Pegoraro; E. Bertini; P. Bernascono; G. Lattanzi; A. Lo Gerfo; G. Siciliano

P.148 **Genotype-phenotype correlations in human diseases caused by mutations of LINC complex-associated genes: a systematic review and meta-summary**
E. Storey; H. Fuller

P.149 **Differential expression of intermediate filament proteins; Lamins A/C and Desmin within and between adult skeletal muscles**
E. Shaqoura; E. McCallion; H. Fuller; M. Bowerman

P.150 **RNA Sequencing confirms the pathogenicity of a novel *FLH1* deletion**
H. Kushlaf; C. Nagaraj; C. Tian

P.152 **The novel *ANXA11* variant p.Asp40Ile in a childhood-onset oculopharyngeal muscular dystrophy shows the pathogenic relevance of Asp40 in ANXA11 disorders**
D. Natera-de Benito; J. Olival; C. Garcia-Cabau; A. Codina; M. Roldan; J. Expósito-Escudero; C. Batlle; L. Carrera-García; C. Ortez; C. Jou; X. Salvatella; F. Palau; A. Nascimento; J. Hoenicka

P.153 **Pilot trial of sialyllactose in patients with *GNE* myopathy**
Y. Park; J. Choi; L. Kim; J. Shin

P.154 **A novel *TIA1* frameshift variant in a dominant myopathy family**
J. Sarparanta; P. Jonson; A. Vihola; H. Luque; S. Brady; B. Udd

P.155 **Recurring homozygous *ACTN2* variant (p.Arg506Gly) cause a recessive, adult-onset myofibrillar myopathy**
S. Donkervoort; P. Mohassel; M. O'Leary; T. Hartley; T. Mozaffar; M. Saporta; D. Dyment; C. Austin-Tse; S. Verma; K. Hurth; J. Warman-Chardon; A. O'Donnell-Luria; C. Bönnemann

P.156 **Novel repeat expansions in *PLIN4* in two Spanish families suffering from autosomal dominant distal myopathy with unique pathological features**
M. Olive; I. Stevanovski; L. González Quereda; G. Morris; A. Segarra-Casas; B. Rodriguez-Santiago; P. Gallano; R. Alvarez; A. Vesperinas; B. San Millan; C. Navarro; G. Ravenscroft; I. Illa; I. Deveson; E. Gallardo

P.157 **Dominant *HSPB6* mutation in a myopathy patient**
J. Sarparanta; P. Jonson; A. Vihola; H. Luque; A. Vainio; R. Villar-Quiles; T. Stojkovic; N. Romero; B. Eymard; B. Udd

P.158 **Is there a myopathic component in Urofacial (Ochoa) syndrome?**
G. Remiche; L. Desmyter; I. Vandernoot; H. Kadhim; S. Coppens; A. Herbaut

VP.65 **Screening of small molecules for activation of GNE protein carrying non-catalytic site mutation based on molecular docking simulation**
W. Yoshioka; K. Yamamoto; S. Hayashi; M. Sekijima; I. Nishino; S. Noguchi

VP.66 **CRISPR/Cas9-targeted single molecule long-read sequencing reveals allelic microheterogeneity of triplet repeat expansion in oculopharyngodistal myopathy**
N. Eura; S. Noguchi; M. Ogasawara; A. Iida; S. Hayashi; I. Nishino

VP.67 **A novel adult-onset vacuolar myopathy caused by a large expansion of the *PLIN4* gene - clinical, histological and imaging data**
L. Maggi; S. Gibertini; E. Iannibelli; A. Gallone; C. Bragato; S. Bonanno; F. Blasevich; R. Mantegazza; M. Mora; A. Ruggieri

VP.68 ***ACTN2*: Mutation update**
J. Ranta-aho; M. Olivé; G. Roticiani; M. Vandroux; C. Dominguez; M. Johari; A. Torella; J. Böhm; J. Turon; V. Nigro; P. Hackman; J. Laporte; B. Udd; M. Savarese

VP.69 **Natural history of Tibial muscular dystrophy**
V. Kuusinen; M. Savarese; N. Sandholm; P. Hackman; B. Udd

VP.70 **OPALE: a patient registry for laminopathies and emerinopathies in France**
R. Ben Yaou; F. Anselme; A. De Sande-Giovannoli; E. Campanna- Salort; P. Charron; C. Chikhaoui; I. Jeru; F. Labombarda; F. Leturcq; S. Quijano-Roy; C. Stalens; P. Richard; C. Vigouroux; G. Bonne; K. Wahbi

14:50-15:10 **LGMD** (FP.33-36) - Ballroom
Moderator: Nicholas Johnson

14:50-14:55 FP.33 **Ataluren treatment in 30-week-old dysferlinopathy mouse with nonsense mutation**
K. Seo; D. Kim; H. Lee; J. Shin

14:55-15:00 FP.34 **Clinical outcome study of dysferlinopathy: correlation between MRI fat fraction in lower limbs and clinical outcome assessments over a 3 year period**
F. Smith; H. Reyngoudt; J. Díaz Manera; M. James; I. Wilson; E. Caldas de Almeida Araujo; C. Bolano Diaz; H. Gordish Dressman; L. Rufibach; A. Mayhew; K. Jones; E. Salort Campana; M. Walter; T. Stojkovic; M. Yoshimura; E. Pegoraro; J. Mendell; V. Straub; A. Blamire; P. Carlier

15:00-15:05 FP.35 **Myostatin concentration is unreliable as a biomarker of disease progression in dysferlinopathy**
U. Moore; E. Fernandez Simon; J. Day; K. Jones; D. Bharucha-Goebel; A. Pestonk; M. Walter; C. Paradas; T. Stojkovic; E. Bravver; E. Pegoraro; J. Mendell; M. Guglieri; V. Straub; J. Díaz Manera

15:05-15:10 FP.36 **Genetic variants in DTNA cause a mild dominantly inherited muscular dystrophy**
A. Nascimento; C. Bruels; A. Codina; J. Milisenda; C.Li; L. Carrera-García; E. Estrella; J. Pijuan; J. Expósito-Escudero; S. Stafki; L. Martorell; H. Lidov; C. Ortez; F. Palau; B. Darras; C. Jou; L. Kunkel; J. Hoenicka; P. Kang; D. Natera-de Benito

14:30-16:00 **LGMD** (P.159-183) (VP.71) - Poster area - Ballroom B1-B2

P.159 **Correlation of clinical outcome parameters in patients with LGMDR1 with quantitative muscle MRI of the leg muscles**
A. Guettsches; J. Forsting; M. Rohm; R. Rehmann; M. Froeling; M. Vorgerd; L. Schlaffke

P.160 **Clinical outcome assessments in limb girdle muscular dystrophy R1/2A**
S. Poelker; S. Study Group

P.161 **The molecular landscape of *CAPN3* mutations in limb-girdle muscular dystrophy: experience of Tertiary Center from Turkey**
Ö. Yayıcı Köken; A. Ceylan; T. Esen; C. Semerci Gündüz

P.162 **Clinical outcome study of dysferlinopathy: Performance of upper limb entry item to predict forced vital capacity in dysferlinopathy (LGMDR2)**
M. James; H. Gordish Dressman; H. Hilsden; L. Rufibach; A. Human; T. Duong; E. Maron; B. DeWolf; K. Rose; C. Siener; S. Thiele; N. Sánchez-Aguilera Práxedes; A. Canal; S. Holsten; C. Sakamoto; I. Pedrosa-Hernández; L. Bello; L. Alfano; L. Pax Lowes; V. Straub; A. Mayhew

P.163 **Quantitative MRI in upper limb muscle of patients with dysferlinopathy: preliminary baseline results of the natural history study Jain COS2**
H. Reyngoudt; F. Smith; I. Wilson; E. Caldas de Almeida Araujo; B. Marty; P. Baudin; J. Díaz-Manera; L. Rufibach; H. Gordish Dressman; H. Hilsden; H. Sutherland; G. Querin; T. Stojkovic; V. Straub; P. Carlier; A. Blamire

P.164 **Clinical outcome study of dysferlinopathy 2: characterising involvement of the intrinsic muscles of the hand in LGMDR2**
M. James; A. Mayhew; H. Gordish-Dressman; L. Rufibach; K. Wong; W. Roper; S. Holsten; L. Lowes; T. Duong; C. Yochai; A. Zabala Pardo; Y. Ogasawara,; K. Rudolph; S. Alarcon; J. Weber; E. Montiel Morillo; I. Pedrosa- Hernandez; S. Birnbaum; J. Rojas Rojas; J. Day; V. Straub

P.165 **Clinical outcome study of dysferlinopathy: lower limb water T2 predicts functional decline in patients with dysferlinopathy**
U. Moore; E. Caldas de Almeida Araujo; H. Reyngoudt; H. Gordish-Dressman; F. Smith; J. Wilson; M. James; A. Mayhew; L. Rufibach; T. Stojkovic; A. Blamire; V. Straub; P. Carlier; J. Díaz Manera

P.166 **Clinical outcome study of dysferlinopathy: the impact of lower limb orthoses on gait - a longitudinal single case study**
M. James; L. Alcock; K. Wong; M. Richardson; V. Straub; A. Mayhew

P.167 **Clinical outcome study of dysferlinopathy: gait analysis of siblings and phenotype variation**
M. James; L. Alcock; K. Wong; M. Richardson; V. Straub; A. Mayhew

P.168 **Observational study: the quality of life in patients with alpha-sarcoglycan, beta-sarcoglycan and gamma-sarcoglycan gene mutation**
B. Vola; Y. Torrente; M. Cerletti; R. Maggi; C. Sanchez Riera; C. Paniga

P.169 **Nintedanib improves muscle function and reduces fibrosis in a murine model of alpha-sarcoglycanopathy**
J. Alonso-Perez; X. Suarez-Calvet; A. Carrasco-Rozas; P. Piñol-Jurado; E. Fernández-Simón; M. Borrel-Pages; L. Wollin; M. Olivé; J. Díaz-Manera

P.170 **Safety, β-Sarcoglycan Expression, and Functional Outcomes From Systemic Gene Transfer of Bidridistrogene Xeboparvovec in Limb-Girdle Muscular Dystrophy Type 2E/R4**
L. Rodino-Klapac; E. Pozsgai; S. Lewis; D. Griffin; A. Meadows; K. Lehman; K. Church; N. Reash; M. Iammarino; B. Sabo; L. Alfano; L. Lowes; S. Neuhaus; X. Li; J. Mendell

P.171 **Detection of alpha-dystroglycan glycation in muscle biopsies using a multiplexed western blot method**
H. Rodriguez; T. Rajasingham; A. Ji; E. Huang; U. Sinha; D. Sproule

P.172 **AAV-mediated strategy for *TCAP* gene correction as a new treatment for LGMDR7/LGMD2G dystrophy**
L. Gushchina; B. Bradley; K. Terry; S. Casey; B. Petrykowski; J. Lay; E. Frair; T. Vetter; N. Rohan; G. Cox; S. Wolfe; C. Emerson; K. Flanigan

P.173 **Preliminary natural history quantitative MRI data in lower limb muscle and heart of patients with limb-girdle muscular dystrophy type R9**
H. Reyngoudt; Y. Fromes; M. Granier; P. Baudin; G. Querin; V. Straub; T. Stojkovic; S. Olivier; J. Vissing; B. Marty

P.174 ***TRIM32* related muscular dystrophy mimicking inflammatory myopathy: clinical and histopathological features in two siblings**
R. Orbach; L. Ostrow; R. Roda

P.175 **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; K. Ørstavik; V. Straub

P.176 **Evaluation of thigh muscle fat fraction with quantitative MRI in 24 adult LGMDR12 patients over 2 years of follow-up**
B. De Wel; L. Huysmans; R. Peeters; V. Goosens; S. Ghysels; K. Byloos; G. Putzeys; A. D'Hondt; J. De Bleecker; P. Dupont; F. Maes; K. Claeys

	P.177 **Analysis of the proximo-distal gradients of fat replacement along the length of thigh muscles in LGMDR12 patients** B. De Wel; L. Huysmans; F. Maes; P. Dupont; K. Claeys P.178 **Clinical classification of variants in the valosin containing protein gene associated with multisystem proteinopathy** J. Díaz Manera; M. Schiava; C. Ikenaga; T. Stojkovic; I. Nishino; S. Nair; G. Manousakis; C. Quinn; Z. Sahenk; M. Monforte; A. Oldfords; E. Pal; B. Velez Gomez; J. de Bleecker; M. Farrugia; M. Harms; S. Ralston; J. Sotoca Fernandez; J. Bevilacqua; C. Weihl & the VCP M Study Group P.179 **Clinical trial readiness and validation of onsite and remote evaluation in valosin containing protein-associated multisystem proteinopathy** L. Alfano; A. Peck; M. Iammarino; S. Patel; N. Reash; M. Almomen; J. Mendell; B. Sabo; A. Long; L. Pietruszewski; L. Lowes; N. Peck P.180 **Diagnostic range of targeted next-generation sequencing in a single center experience with limb-girdle muscular dystrophy** S. Lee; S. Kim; H. Park; K. Park; Y. Choi P.181 **Limb girdle muscle dystrophy: a Brazilian cohort on ICGNMD study** P. Tomaselli; R. Frezatti; C. Record; R. Pitceathly; H. Houlden; M. Hanna; M. Reilly; W. Marques Jr; C. Sobreira P.183 **A case of limb-girdle muscular dystrophy D2 with *TNPO3* mutation in Korea** J. Lee; H. Park; Y. Choi VP.71 **Simultaneous texture and relaxation estimation in skeletal muscle in cystinosis patient compared to healthy control with high-field MRI** B. Sveinsson; R. Sadjadi
15:30-16:00	Afternoon refreshments exhibition and posters - Ballroom Salon and Pre Function Space
16:00-17:00	**Debate Clinical Trials: Should biomarkers replace outcome measures?** - Ballroom Speakers: Nicol Voermans, Radboud University Medical Center, Netherlands Jim Dowling, Hospital for Sick Children, Canada *Moderators: Meredith James, John Walton Muscular Dystrophy Research Centre, UK* *Gina Ravenscroft, Harry Perkins Institute Of Medical Research - UWA, Australia*
17:00-18:00	**Poster session 4** (FP.37- 44) - Flash Poster Presentation areas - Ballroom or Ballroom Salon (P.184 -224) (VP.72-93) (LBP.01-03) (LSVP.01-37) - Poster area - Ballroom B1-B2 ***(Please note that this session will not be live streamed for virtual delegates)***
17:00-17:20	**Congenital muscular dystrophies** (FP.37-40) – Ballroom *Moderator: Francesco Muntoni*
17:00-17:05	FP.37 **CDP-ribitol prodrug treatment ameliorates ISPD-deficient muscular dystrophy mouse model** H. Tokuoka; R. Imae; H. Nakashima; H. Manya; C. Masuda; S. Hoshino; K. Kobayashi; D. Lefeber; R. Matsumoto; T. Okada; T. Endo; M. Kanagawa; T. Toda
17:05-17:10	FP.38 **Linker protein-mediated gene therapy ameliorates muscle and nerve pathology in mouse models for LAMA2-related congenital muscular dystrophy** J. Reinhard; S. Lin; M. Ruegg
17:10-17:15	FP.39 **An international retrospective early natural history study of *LAMA2*-related dystrophies** R. Orbach; J. Park; L. Hinkley; N. Acquaye; R. Alvarez; G. Dziewczapolski; C. Bönnemann; A. Foley
17:15-17:20	FP.40 **A cross-sectional study on *LAMA2*-related muscular dystrophy and *SELENON*-related myopathy, the first results of the LAST STRONG Study** K. Bouman; J. Groothuis; J. Doorduin; N. van Alfen; F. Udink ten Cate; F. van den Heuvel; R. Nijveldt; A. Dittrich; J. Draaisma; M. Janssen; B. van Engelen; C. Erasmus; N. Voermans

17:00-18:00

Congenital muscular dystrophies (P.184-190) (VP.72-81) - Poster area - Ballroom B1-B2

P.184 **Defining the pathological natural history of *LAMA2* muscular dystrophy**
A. Hopp; K. Jones; H. Meng; E. Ott; N. Basuni; S. Axon; T. Moors; S. Moore; M. Lawlor

P.185 **Evaluation of brain microstructure in LAMA2 related muscular dystrophy by NODDI: a pilot study**
G. Oz Tuncer; B. Genc; S. Aydin; K. Aslan; M. Ceyhan Bilgici; A. Aksoy

P.186 **Evaluating the feasibility of functional outcomes and biomarkers in young patients with laminin alpha2-related dystrophies performed in clinic or remote**
L. Alfano; M. Jain; A. Foley; N. Reash; M. Iammarino; L. Lowes; A. Long; V. Kent; L. Hinckley; N. Acquaye; C. Bönnemann; A. Connolly

P.187 **SelN expression in activated satellite cells following muscle injury**
M. Wright

P.188 **Early growth and metabolic abnormalities in zebrafish and cellular models of *SELENON*-related myopathy**
P. Barraza-Flores; B. Moghadaszadeh; B. Mitchell; E. Troiano; A. Mansur; V. Gupta

P.189 **Using in situ hybridization to delineate collagen VI genes' expression patterns in skeletal muscles of wild-type and COL6-related dystrophies mice**
F. Guirguis; H. Zhou; V. Bolduc; F. Muntoni; C. Bönnemann

P.190 **Congenital muscular dystrophy associated to conserved oligomeric Golgi complex subunit 1 homozygous mutation**
J. Balkenhol; P. Araneda; B. Suarez; J. Jofre; M. Martinez-Jalilie; M. De la Fuente; F. Fattori; E. Bertini; M. Serrano; C. Castiglioni

VP.72 **Diversity of splice-acting variants in the *COL6A1, COL6A2* and *COL6A3* genes associated with collagen VI-related dystrophies**
V. Bolduc; R. McCarthy; Y. Hu; S. Silverstein; P. Uapinyoying; S. Donkervoort; A. Foley; C. Bönnemann

VP.73 **Characteristics of cardiac dysfunction in patients with Fukuyama congenital muscular dystrophy**
K. Ishiguro; T. Sato; M. Shichiji; Y. Kihara; T. Murakami; S. Nagata; K. Ishigaki

VP.74 **Uniparental disomy unmasks a homozygous mutation of *POMGNT1* in a case of muscle-eye-brain disease**
Y. Liu; H. Xiong

VP.75 ***Jab1* deletion in muscle lineage causes a muscular dystrophy that resembles *LAMA2* disease**
E. Porrello; M. Molina; M. Lorenzetti; S. Previtali

VP.76 **Natural history of a novel mouse model for *LAMA2*-related congenital muscular dystrophy**
D. Tan; Q. Shen; Y. Liu; L. Xu; H. Zhang; H. Xiong

VP.77 **Muscle transcriptomic study of a novel *LAMA2*-related congenital muscular dystrophy mouse model**
D. Tan; H. Zhang; H. Xiong

VP.78 **Clinical and genetic study of *LAMA2*-related muscular dystrophy patients with seizures**
X. Huang; H. Yang; D. Tan; L. Ge; Y. Fan; X. Chang; Z. Yang; H. Xiong

VP.79 **Challenges in genetic diagnosis of *LAMA2*-MD - when the pieces do not fit**
A. Goncalves; C. Garrido; E. Vieira; M. Oliveira; M. Pinto; R. Taipa; I. Carrilho; M. Santos; R. Santos

VP.81 **In *vivo* modulation of novel genetic modifiers for *LAMA2-RD***
V. Pini; B. Weisburd; M. Merteroglu; I. Sealy; R. White; E. Busch-Nentwich; F. Muntoni

17:00-17:10 **DMD – Preclinical** (FP.41-42) – Ballroom Salon
Moderator: Thomas Krag

17:00-17:05 FP.41 **Duchenne muscular dystrophy functional muscle organoid-on-a-chip for potential therapies evaluation**
J. Fernández-Costa; A. Tejedera-Villafranca; J. Ramón-Azcón

17:05-17:10 FP.42 **Correction of point mutations in the DMD gene using the prime editing**
C. Happi Mbakam; J. Rousseau; J. Tremblay

17:00-18:00 **DMD – Preclinical** (P.191-200) (VP.82-86) - Poster area - Ballroom B1-B2

P.191 **RKER-065, a novel muscle and bone anabolic, increased muscle, grip strength and trabecular bone in a mouse model of Duchenne muscular dystrophy**
R. Nathan; C. Materna; D. Welch; T. Nurse; E. Lema; A. Gudelsky; C. Tseng; F. Fisher; J. Seehra; J. Lachey

P.192 **High-throughput screening of antifibrotic and antiadipogenic drugs using human FAP cells**
E. Fernandez Simon; I. Matthews; P. Piñol Jurado; X. Suarez Calvet; D. Cox; V. Justian; A. Carrasco Rozas; A. Bowey; P. Rushton; S. Lopez Fernandez; J. Díaz Manera

P.193 **Phosphoryl guanidine-containing backbone modifications enhance exon skipping, dystrophin restoration and survival in a severe mouse model for DMD**
X. Hu; P. Kandasamy; G. McClorey; M. Shimizu; N. Kothari; N. Iwamoto; M. Byrne; F. Liu; K. Longo; J. Oieni; C. Shivalila; C. Rinaldi; H. Yang; M. Wood; C. Vargeese

P.194 **Development of a novel, EEV-Conjugated PMO for Duchenne muscular dystrophy**
N. Kreher; A. Kumar; A. Hicks; S. Peddigari; X. Li; A. Pathak; M. Kheirabadi; K. Kamer; N. Estrella; P. Dougherty; W. Lian; N. Liu; N. Gao; D. Wang; M. Streeter; M. Dhanabal; Z. Qian; M. Girgenrath; N. Sethuraman

P.195 **Quantitative skeletal muscle MRI of golden retriever muscular dystrophy dogs**
B. Marty; I. Barthélémy; X. Cauchois; P. Baudin; Y. Fromes; S. Blot; H. Reyngoudt

P.196 **Rebuilding muscle in Duchenne by correcting stem cell polarity**
R. Mitchell; S. Narayan; F. Gleeson; M. Rudnicki

P.197 **Evaluating pharmacology and efficacy of delandistrogene moxeparvovec in young and aged DMDMDX rats**
R. Potter; C. Wier; G. Cooper-Olson; E. Wheeler; E. Anderbery; A. Kempton; L. Clements; K. Adegboye; A. Haile; E. Peterson; L. Rodino-Klapac

P.198 **Durable AOC mediated exon 44 skipping in non-human primate muscle tissue and dystrophin protein restoration in DMD patient derived skeletal muscle cells**
G. Karamanlidis; U. Etxaniz; M. Missinato; M. Díaz; R. Bhardwaj; O. Tyaglo; K. Lemoine; I. Marks; T. Albin; L. Leung; P. Kovach; A. Anderson; M. Cochran; H. Huan; H. Younis; M. Flanagan; A. Levin

P.200 **Automated quantification of dystrophin immunofluorescence in human and mouse muscle sections**
T. Vetter; A. Bradley; S. Nicolau; E. Frair; L. Gushchina; K. Flanigan

VP.82 **PK/PD modelling to inform clinical development of an adeno-associated virus gene transfer therapy for Duchenne muscular dystrophy**
L. East; R. Potter; J. Snedeker; A. Haile; C. Wier; L. Rodino-Klapac

VP.83 **Ion channels and myogenesis in Duchenne muscular dystrophy: Electrophysiological profile of wild-type and dystrophic myoblasts and myocytes**
A. Cerchiara; P. Imbrici; S. Cirmi; D. Wells; A. De Luca; O. Cappellari

VP.84 **Growth hormone secretagogues in Duchenne muscular dystrophy: a preclinical evaluation of potential benefits on muscle function and morphology**
P. Mantuano; B. Boccanegra; O. Cappellari; S. Cirmi; E. Bresciani; E. Conte; A. Mele; M. De Bellis; S. Denoyelle; A. Torsello; A. Liantonio; A. De Luca

VP.85 **C-Path's Duchenne Regulatory Science Consortium: Accelerating drug development for Duchenne muscular dystrophy**
R. Belfiore-Oshan; V. Aggarwal; S. Sivakumaran; D. Corey; C. Ollivier; K. Romero; K. Vandenborne; S. Kim; J. Morales; K. Lingineni; T. Martinez

VP.86 **Effect of a chronic treatment with L-citrulline on funtional, histological and molecular readouts of dystrophic mdx mouse model**
L. Tulimiero; B. Boccanegra; P. Mantuano; S. Cirmi; M. De Bellis; O. Cappellari; A. De Luca

17:10-17:20 **Myotonic Dystrophy** (FP.43-44) – Ballroom Salon
Moderator: Benedikt Schoser

17:10-17:15 FP.43 **Congenital myotonic dystrophy patients exhibit unique patterns of transcriptomic dysregulation independent of CTG repeat expansion**
N. Johnson; K. Bates; M. Provenzano; M. Hale

17:15-17:20 FP.44 **Exploring the role of MuscleBlind-Like proteins in the regulation of CaVB1 isoform expression in adult skeletal muscle**
A. Vergnol; A. Sureau; M. Traore; G. Gourdon; D. Furling; F. Pietri-Rouxel; S. Falcone

17:00-18:00 **Myotonic Dystrophy** (P.201-213a) (VP.88-92) - Poster area - Ballroom B1-B2

P.201 **A case of periodic paralysis attending with myotonia and family screening**
G. Oz Tuncer; M. Yıldız; N. Randa; S. Peynir; S. Aydin; H. Türker; A. Aksoy

P.202 **Mitochondrial dysfunction in myotonic dystrophy type 1 patients**
V. Di Leo; C Lawless; M P. Roussel; G S. Gorman; O M. Russell; H A. Tuppen; E Duchesne; A E. Vincent

P.203 **Biomarkers for central nervous system involvement in Myotonic dystrophy type 1**
A. Varghese; S. Spendiff; A. Ross; H. Lochmüller

P.204 **Blood based biomarker discovery in DM1**
D. van As; R. van Cruchten; J. Glennon; B. van Engelen; P. 't Hoen

P.205 **Utility of electrical bioimpedance as a biomarker in myotonic dystrophy type I**
S. Kapetanovic Garcia; P. Rodrigo Armenteros; A. Monzon Mendiola; O. Monasterio Jimenez; M. Ponce; J. Rekondo; L. Varona Franco; N. Iglesias Hernandez

P.206 **Diffusion tensor imaging (mDTI) in myotonic dystrophy type 1 and type 2**
A. Guettsches; R. Rehmann; C. Schneider-Gold; M. Rohm; J. Forsting; M. Froeling; M. Vorgerd; L. Schlaffke

P.207 **Muscle magnetic resonance imaging in Myotonic dystrophy type 1: Longitudinal study for 5 years**
J. Park; Y. Lim; M. Kang; J. Shin; D. Kim

P.208 **The neurocognitive phenotype of childhood Myotonic dystrophy type 1: A multicenter pooled analysis**
D. Sweere; S. Moelands; S. Klinkenberg; L. Leenen; J. Hendriksen; H. Braakman

P.209 **Prevalence of healthcare conditions and services used by patients with myotonic dystrophy (DM) pre- and post-diagnosis: A real-world data analysis**
J. Day; K. Munoz; R. Brook; B. McEvoy; L. Tai; K. DiTrapani; N. Kleinman; C. Chen; M. Stahl

P.210 **A phase 1/2 clinical trial evaluating the safety and pharmacokinetics of AOC 1001 in adults with myotonic dystrophy type 1: MARINA study design**
N. Johnson; J. Day; J. Hamel; J. Statland; S. Subramony; W. Arnold; C. Thornton; M. Wicklund; P. Soltanzadeh; B. Knisely; V. Goel; K. DiTrapani; C. Chen; K. Clark; A. Peters; C. Heusner; H. Younis; L. Tai; E. Ackermann

P.211 **TREAT-NMD myotonic dystrophy Global Registry Network: providing data in congenital myotonic dystrophy to support FDA regulatory decision making**
N. Bennett; C. Campbell; B. Porter; E. Ashley; B. Esparis; D. Allison; M. Guglieri; A. Ambrosini; T. Stevenson; K. Cumming; C. Marini-Bettolo; M. Rodrigues; V. Hodgkinson; L. Korngut; R. Forbes; M. Ryan; M. Snape; S. Evans; J. Horrigan; S. Peric; R. Roxburgh

P.212 **TREAT-NMD Myotonic dystrophy (DM) Global Registry Network: an update in 2022**
N. Bennett; R. Roxburgh; B. Porter; H. Walker; D. Allison; E. Ashley; B. Esparis; C. Campbell; M. Guglieri; A. Ambrosini; S. Peric

P.213 **TREAT-NMD myotonic dystrophy global registry network: an international collaboration in myotonic dystrophy type 2**
S. Peric; B. Porter; N. Bennett; D. Allison; E. Ashley; B. Esparis; C. Campbell; M. Guglieri; A. Ambrosini; R. Roxburgh

P.213a **Multimodal fusion of neuroimaging and neuropsych data: a machine learning approach to study brain alterations linked with cognitive domains in DM1**
T. Kamali; D. Parker; G. Deutsch; J. Sampson; J. Day; J. Wozniak

VP.88 **Characterisation of cell culture models of myotonic dystrophy type I by In-Cell Western technology and digital droplet PCR**
A. López-Martínez; P. Soblechero-Martín; C. Catalli; A. Jauregui-Barrutia; S. Kapetanovic-Garcia; G. Nogales-Gadea; V. Arechavala-Gomeza

VP.89 **Skeletal muscle after a single bout of eccentric exercise in myotonic dystrophy type 1: a complete proteomic analysis**
M. Roussel; L. Hébert; C. Gagnon; E. Duchesne

VP.90 **Is an early diagnosis of congenital and childhood forms of myotonic dystrophy type 1 possible? Clinical and electromyographic description of case series**
S. Kurbatov; V. Kenis; M. Savina; I. Kleimenova; Y. Kryukov; N. Priymak; A. Kokorina; N. Ryadninskaya; I. Kuznetsova; O. Shchagina; A. Poliakov; S. Efimenko

VP.91 **A preliminary assessment of the psychometric properties of the congenital myotonic dystrophy type 1 rating scale (CDM1-RS) in a phase 2/3 study**
N. Nikolenko; J. Horrigan; M. Snape; A. Veerapandiyan; S. Evans; H. Lochmüller

VP.92 **Autonomic symptoms are frequent in myotonic dystrophy type 1**
S. Alonso; N. Alvarez; N. Amaral; G. Morís

17:00-18:00 **Neuropathies and Non-5q Motor Neuron Diseases** (P.214-224) (VP.93) - Poster area - Ballroom B1-B2

P.214 **Utility of ENMG in children with rare genetic neurogenic disorders: a case series**
G. Dufort; C. Nguyen

P.215 **Non 5Q SMA: a Brazilian cohort study**
R. Frezatti; P. Tomaselli; C. Record; R. Pitceathly; H. Houlden; M. Hanna; M. Reilly; W. Marques Junior

P.216 **Two interesting and unique cases of acquired neuropathies in pediatrics**
G. Arbour; S. Perreault; N. Prudhomme; P. Teira; R. Scott; C. Nguyen

P.217 **Cervical spinal cord MRI parameters as predictors of early degeneration in asymptomatic *C9Orf72* carriers: a longitudinal study**
G. Querin; D. Saracino; D. Rinaldi; F. Salachas; V. Marchand-Pauvert; J. Cohen Adad; I. Le Ber; P. Pradat

P.218 **Moving along the ALS-bvFTD spectrum: longitudinal changes in MEG-based brain network topology of ALS patients with cognitive/behavioural impairment**
R. Govaarts; E. Scheijbeler; E. Beeldman; M. Fraschini; A. Griffa; M. Engels; A. van der Kooi; Y. Pijnenburg; M. de Visser; C. Stam; J. Raaphorst; A. Hillebrand

P.219 **Novel clinical phenotype of early-onset amyotrophic lateral sclerosis and frontotemporal dementia with *SNCB* mutation**
H. Lee; M. Lee; J. Lee

P.220 **Neuropsychological functioning in CMT type 2Z: a case report of language deficits masquerading as intellectual disability**
A. Miele; M. Yang; S. Apkon; C. Silver; M. Gibbons; M. Gibbons; H. Foster; A. Ballard

P.221 **Improvement in respiratory and bulbar function in two patients with SMALED2B**
A. Meyer; S. Hickey; S. Nicolau; M. Waldrop

P.222 **Wearable inertial sensors for longitudinal follow-up of patients with amyotrophic lateral sclerosis**
M. Poleur; M. Annoussamy; L. Clavel; L. Buscemi; S. Delstanche; D. Eggenspieler; A. Maertens de Noordhout; O. Bouquiaux; I. Lievens; L. Servais

P.223 **Electrophysiologic evidence of *MORC2* pathogenic variant with motor neuron involvement: a case report**
A. Mekmangkonthong; G. Yoon; A. Aleman; F. Paiz; H. Gonorazky

P.224 **Non-5q spinal muscular atrophy in twin sisters with SPG11/CMT2X associated spatacsin gene mutation**
G. Siciliano; G. Ricci; F. Torri; A. Govoni; A. Trabacca

VP.93 **A novel variant of *DYNC1H1* mutations in spinal muscular atrophy lower extremity predominant in an Indonesian patient: A case report**
K. Iskandar; Gunadi; G. Ivana; A. Triono; E. Herini

17:00-18:00

Late Breaking News
(LBP.01-03) Poster area - Ballroom B1-B2

LBP.01 **Efficacy and safety of Avalglucosidase Alfa in participants with late-onset Pompe Disease after 145 weeks' treatment during the COMET trial**
Schoser B, Kishnani P, Kushlaf H, Ladha S, Mozaffar T, Straub V, Toscano A, van der Ploeg A, Clemens P, Day J, Illarioshkin S, Roberts M, Attarian S, Carvalho G, Erdem-Özdamar S, Goker-Alpan O, Kostera-Pruszczyk A, Díaz-Manera J, Dimachkie M, on behalf of the COMET Investigator Group

LBP.02 **Biallelic MYO15B variants are associated with a congenital core myopathy with rigid spine, and PAX7 deficiency**
Onnée M, Taglietti V, Cecconi B, de Becdelièvre A, Bastu S, Villanova M, Authier F, Relaix F, Fanen P, Funalot B, Malfatti E

LBP.03 **Biallelic variants in ASCC3 expand the phenotypic spectrum of congenital myasthenic syndromes**
Johari M, Herman I, Polavarapu K, Nalini A, Lupski J, Töpf A, Villanova M, Tasca G, Mercuri E, Pegoraro E, Sivasankar M, Straub V, Malfatti E, Lochmüller H, Calame D, Ravenscroft G[1]

Late Submitted Abstracts
(LSVP.01-37) Virtual platform and E-Posters only

LSVP.01 **BGE-117 accelerates recovery from eccentric muscle injury and increases voluntary activity levels**
Pan Y, Wang Y, Cowdin S, Banicki M, Morgen E, Rebo J

LSVP.02 **A novel antisense oligonucleotide targeting DUX4 strongly prevents motor function decline in an FSHD model ACTA1-MCM/FLExDUX4 mice.**
Kakimoto T, Ogasawara A, Ishikawa K, Kurita T, Yoshida K, Harada S, Inoue Y, Kumagai S, Sasaki T, Aihara H

LSVP.03 **Patient reported outcome measures in adult spinal muscular atrophy: connecting the dots**
Slayter J, Casey L, O'Connell C

LSVP.04 **TRPML1 in muscle biogenesis and disease**
Syeda A, Sun X, Wang P, Sao Q, Xu M, Dong X

LSVP.05 **Endogenous Human SMN1 Promoter-driven Gene Replacement Improves the Efficacy and Safety of AAV9-mediated Gene Therapy for SMA in mice**
Xie J, Xie Q, Ma H, Chen X, Zhu Y, Ma Y, Jalinous L, Su Q, Tai P, Gao G

LSVP.06 **Real-world experience of delivering gene-therapy with Onasemnogene Abeparvovec (Zolgensma®) for children with Spinal Muscular Atrophy (SMA) type-1 in the UK**
Gowda V, Jungbluth H, Standing E, Sheehan J, Hughes I, Mccullagh G, Atherton M, Ong M, Majumdar A, Murugan A, Dhawan A, Muntoni F, Baranello G, Scoto M, Manzur A, Munot P, Abbot L, Horrocks I, Tirupathi S, Wraige E

LSVP.07 **Biomechanical properties of skeletal muscle: the impact of dysferlinopathy, ageing, and muscle type**
Lloyd E[1,2], Hepburn M[3,4], Li J[3,4,5], Mowla A[3,4], Choi Y[1], Hwang Y[6], Jackaman C[2], Kennedy B[3,4,5], Grounds M[1]

LSVP.08 **AOC 1020: An Antibody Oligonucleotide Conjugate (AOC) in Development for the Treatment of FSHD**
Malecova B, Sala D, Melikian G, Erdogan G, Johns R, Jordan M, Hartmann M, Arias D, Battacharya A, Doppalapudi R, Huang H, Flanagan M, Levin A

LSVP.09 **Results of a 4-year Viltolarsen extension study of functional and safety outcomes**
Clemens P, Rao V, Connolly A, Harper A, Mah J, McDonald C, Smith E, Zaidman C, Hoffman E

LSVP.10 **Consequences of the lack of one or multiple dystrophin isoforms on cognition in prednisolone treated mdx mouse models**
Verhaeg M, Van De Vijver D, Tanganyika-de Winter C, Stan T, Mastenbroek L, Aartsma-Rus A, van Putten M

LSVP.11 **Endolysosomal cation channel TRPML3 in X-linked myotubular myopathy**
Xu M, Zhong X, Dong X

LSVP.12 **Preliminary safety and outcomes from a phase I/IIa trial of IGHMBP2 gene replacement in SMARD1/CMT2S**
Waldrop M, Kaler M, Brown K, Das S, Meyer A, Iammarino M, Likhite S, Cox G, Burghes A, Sierra Delgado A, Alfano L, Lowes L, Paul G, Shell R, Connolly A, Flanigan K, Meyer K

LSVP.13 **Human cardiosphere-derived cell therapy in Duchenne Muscular Dystrophy: a new treatment approach to target skeletal and cardiac muscle pathogenesis: HOPE-2-OLE**
McDonald C, Hendrix S, Wassom M, Mallinckrodt C, Eagle M, Harmelink M, Varadhachary A, Steigler P, Marban L, Paulson D

LSVP.14 **Nonparametric analysis of forced vital capacity in the COMET trial demonstrates superiority of avalglucosidase alfa vs alglucosidase alfa**
Diaz-Manera J, Boentert M, Salort Campana E, Attarian S, Dimachkie M, Periquet M, Thibault N, Miossec P, Wang Y, Berger K

LSVP.15 **Avalglucosidase Alfa Improves Motor and Respiratory Function in Late-onset Pompe Disease: A COMET Win-Ratio Analysis**
Diaz-Manera J, Ishak J, Caro J, Hamed A, Riou França L, Thibault N, Shukla P, Berger K, Dimachkie M, Boentert M

LSVP.16 **A new score combining CMAP amplitudes and motor score is predictive of motor outcome after Onasemnogene Abeparvovec in SMA1 infants**
Barrois R, Barnerias C, Quijano-Roy S, Deladrière E, Leloup-Germa V, Tervil B, Audic F, Cances C, Cintas P, Davion J, Espil-Taris C, Manel V, Pereon Y, Piarroux , Vuillerot C, Walther-Louvier U, Desguerre I, Gitiaux C

LSVP.17 **Results from 96 Weeks Open-Label Extension of a Phase 2 Trial of Losmapimod in Subjects with FSHD: ReDUX4**
Wang L, Han J, Shoskes J, Dunn J, Jiang J, Tawil R

LSVP.18 **Bulbar Function in Children with Two or Three SMN2 Copies Who Received Onasemnogene Abeparvovec Presymptomatically for Spinal Muscular Atrophy (SMA)**
Shell R, McGrattan K, Hurst-Davis R, Dunaway Young S, Baranello G, Lavrov A, O'Brien E, Wallach S, LaMarca N, Reyna S, Darras B

LSVP.19 **Indirect treatment comparison of three enzyme replacement treatments for late-onset Pompe disease: a network meta-analysis with patient-level and aggregate data**
Fu S, Hummel N, Mozaffar T, Castelli J, Fox B, Keyzor I, Raza S, Sitaraman S, Shohet S

LSVP.20 **Repurposing Tamoxifen to Cure Muscle Disease**
Wang P, Alia S, Xu M, Dong X

LSVP.21 **Real-world Outcomes of Nusinersen or Onasemnogene Abeparvovec (OA) Monotherapy, or Nusinersen Switching to OA in SMA Patients Aged ≥6 Months**
Dabbous O, Yang M, Georgieva M, Toro W, LaMarca N, Patel A, Anderson A, Akbarnejad H, Reyna S

LSVP.22 **Estimating Health State Utilities in Duchenne Muscular Dystrophy (DMD) using the EQ5D and Health Utilities Index (HUI).**
Audhya I, Szabo S, Bever A, O'Sullivan F, Malone D, Feeny D, Neumann P, Iannaccone S, Gooch K

LSVP.23 **DMD patients treated by SGT-001 Microdystrophin Gene Therapy improve in the objective endpoint of spontaneous walking velocity**
Servais L, Gonzalez P, Morris C, Shieh P, Byrne B, Annoussamy M, Donisa Dreghici R

LSVP.24 **First case of Complex Allele identified in LAMA2 Gene: an Egyptian case report**
Elsayed N, Fahmy N, Abdulhady H, Pereira C, Bertoli-Avella A, Béroud C

LSVP.25 **Pharmacodynamic noninferiority study comparing subcutaneous injections of Efgartigimod PH20 with intravenous infusions of Efgartigimod: results of phase 3 ADAPTsc study**
Casey J, Li G, Vu T, Bril V, Margania T, Korobko D, Smilowski M, Banaszkiewicz K, Liu L, Steeland S, Noukens J, Van Hoorick B, Podhorna J, Li Y, Utsugisawa K, Wiendl H, De Bleecker J, Mantegazza R, Howard, Jr J

LSVP.26 **Ataluren preserves upper limb function in nmDMD patients from Study 041, a phase 3 placebo-controlled trial, and the STRIDE Registry**
McDonald C, Mercuri E, Muntoni F, Gordish-Dressman H, Morgenroth L, Dutra de Resende M, Zhou S, Neeharika M, Haginoya K, Ramos-Platt L, Williams P, Penematsa V, Chou C, Lin M, Johnson S, Werner C, Trifillis P

LSVP.27 **Creation of novel BMD murine model with deletion of exons 8-50 to predict the efficacy of gene therapy for DMD**
Polikarpova A, Galkin I, Korshunova D, Savchenko I, Vassilieva S, Velyaev O, Dzhenkova M, Bardina M, Egorova T

LSVP.28 **Myofiber characteristics in Kbtbd13R408C-knockin mouse model to understand disease onset in NEM6 myopathy**
Baelde R, Galli R, Fortes Monteiro A, Ottenheijm C, de Winter J

LSVP.29 **Factor levels in spinal muscular atrophy**
Acar Arslan E, Yıldız N, Bahadır A, Erden A, Özkan Kart P, Kaya G

LSVP.30 **Survival in Eteplirsen-Treated vs Duchenne Muscular Dystrophy (DMD) natural history patients: an indirect treatment comparison using real-world data**
Iff J, Done N, Tuttle E, Zhong Y, Wei F, Darras B, McDonald C, Mercuri E, Muntoni F

LSVP.31 **Interim analysis of EVOLVE: a long-term observational study evaluating Eteplirsen, Golodirsen, or Casimersen in routine clinical practice**
Ricchetti-Masterson K, Santra S, Hornibrook S, Byrne B, Kumar A, Mathews K, Ramos-Platt L, Waldrop M, Zaidman C, Sehinovych I, Miller D, McDonald C

LSVP.32 **Restoration of full-length dystrophin expression through exon skipping in Duchenne muscular dystrophy patients with single exon duplications**
Nicolau S, Malhotra J, Iammarino M, Reash N, Lowes L, Flanigan K

LSVP.33 **Tamoxifen in Duchenne muscular dystrophy: A multicentre, randomised, double-blind, placebo-controlled, phase 3 safety and efficacy 48-week trial**
Henzi B, Schmidt S, Nagy S, Rubino-Nacht D, Schaedelin S, Putananickal N, Hafner P, Dorchies O, Fischer D, TAMDMD Study Group

LSVP.34 **TPNO3-related LGMD-D2: study of micro-RNA-206, inflammatory cytokines, clinical,MRI and QOL status in two families in view of drug treatment.**
Pegoraro V, Angelini C, Costa R, Cenacchi G, Alcami P

LSVP.35 **Analysis of vector shedding following treatment with delandistrogene moxeparvovec, an investigational rAAVrh74-based gene therapy for DMD**
Malhotra J, Lewis S, Zhang X, Asher D, Hu L, East L, Potter R, Rodino-Klapac L

LSVP.36 **A Phase 2 clinical trial evaluating the safety and efficacy of delandistrogene moxeparvovec in patients with DMD**
Mendell J, Shieh P, Sahenk Z, Lehman K, Lowes L, Reash N, Iammarino M, Alfano L, Sabo B, Woods J, Skura C, Mao H, Staudt L, Potter R, Griffin D, Lewis S, Hu L, Singh T, Rodino-Klapac L

	LSVP.37 **Safety and efficacy of ataluren in nmDMD patients from Study 041, a phase 3, randomized, double-blind, placebo-controlled trial** McDonald C, Wu S, Gulati S, Komaki H, Ruiz-Garcia M, Kostera-Pruszczyk A, Vlodavets D, Chae J, Jong Y, Karachunski P, Statland J, Lorentzos M, Penematsa V, Chou C, Lin M, Werner C, Trifillis P, Gordon G, Klein M
19:00-00:00	**Networking Dinner (pre-registration required)** - Halifax Convention Centre, Convention Level

Saturday 15th October 2022

08:00	**Conference desk opens**
8:30-9:00	**Keynote Lecture** (I.15) - Ballroom **Neuromuscular Disorders with Founder Effects in French Canada: why, where and how they contribute to the NMD field** Bernard Brais, Montreal Neurological Institute and Hospital, Canada *Moderators: Volker Straub and Teresinha Evangelista*
9:00-10:30	**Poster Highlights** - Ballroom *Moderators: Johann Böhm and Laurent Servais*
10:30-11:00	Morning refreshments, exhibition and posters - Ballroom Salon and Pre Function Space
11:00-12:00	**WMS General Assembly & Election Presentation** - Ballroom
12:00-13:30	**Late Breaking News** (LBO.01-09) Ballroom *Moderators: Ichizo Nishino and Lindsay Alfano*
12:00-12:10	LBO.1 **Recessive truncating variants in JPH1 cause a congenital myopathy with ocular and bulbar involvement** Topf A, Johari M, Duff J, Folland C, Marti P, Muelas N, Harris E, Robertson T, Cairns A, Marini-Bettolo C, Vilchez J, Ravenscroft G, Straub V
12:10-12:20	LBO.2 **Microvasculopathy in SMA is driven by a reversible autonomous endothelial cell defect** Zhou H, Hong Y, Scoto M, Thomson A, Pead E, MacGillivray T, Hernandez-Gerez E, Catapano F, Meng J, Zhang Q, Hunter G, Shorrock H, Baranello G, Howell K, Gillingwater T, Brogan P, Thompson D, Parson S, Muntoni F
12:20-12:30	LBO.3 **Biallelic variants in HMGCS1 are a novel cause of rare rigid spine syndrome** Dofash L, Haywood J, Rivas E, Wyllie J, Soares da Costa T, Clayton J, Taylor R, Groth D, Paradas C, Fiorillo C, Scala M, Donkervoort S, Bönnemann C, VanNoy G, Mangilog B, Pais L, Nishino I, Laing N, Cabrera-Serrano M, Ravenscroft G
12:30-12:40	LBO.4 **Fetal therapy for spinal muscular atrophy: a case report** Finkel R, Hughes S, Parker J, Civitello M, Johnson L
12:40-12:50	LBO.5 **Deep mutational scanning and high throughput functional testing to resolve variants of unknown significance in sarcoglycanopathies** Weihl C, Li C, Haller G
12:50-13:00	LBO.6 **Replacement kinetics of the giant muscle protein nebulin are slow and further reduced by a frequently observed mutation in Neb** Bogaards S, Yuen M, Onderwater Y, Clara C, Galli R, Vizoso M, Conijn S, Peters E, Nahidiazar L, Jalink K, van Rheenen J, Granzier H, Ottenheijm C
13:00-13:10	LBO.7 **Repeat expansions in the 5'UTR of ABCD3 cause oculo-pharyngo-distal myopathy with possible sex-biased penetrance** Cortese A, Beecroft S, Cabrera-Serrano M, Curro R, Facchini S, Stevanovski I, Chintalaphani S, Delatycki M, Storey E, Stojkovic T, Houlden H, Kennerson M, McLean C, Deveson I, Laing N, Lamont P, Lockhart P, Fahey M, Bugiardini E, Ravenscroft G

13:10-13:20	LBO.8 **The transcriptomic profile of muscle biopsies from patients with immune checkpoint inhibitor-induced myopathy.** Quintana A, Pinal-Fernandez I, Liewluck T, Milisenda J, Pak K, Casal-Dominguez M, Milone M, Shelly S, Matas-Garcia A, Garrabou G, Padrosa J, Ros J, Trallero-Araguás E, Walitt B, Zhao C, Swift S, Rajan A, Maria Grau J, Selva-O'Callaghan A, Mammen A
13:20-13:30	LBO.9 **Restoring DLC1 isoform 1 expression instead of SMN is a more effective gene therapy for spinal muscular atrophy** Shi T, Liao B, Chang H, Xiong W, Chan S, Liu J, Cheung M
13:30-13:50	**Prize Giving** - Ballroom *Presenter: Johann Böhm*
13:50-14:00	**Introduction to the WMS 2023 Congress, Charleston, USA** Handover of the WMS flag and close of congress - Ballroom *Volker Straub, Lindsay Alfano and Chris Weihl*
14:00-15:00	Homeward lunch - Ballroom Salon and Pre Function Space