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
18:00-18:45	**Opening Ceremony** - Ballroom **Anything Is Possible** James Mullinger *Moderators: Jim Dowling and Volker Straub*
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
08:40-09:00	**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
14:30-14:50	**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.05 **MicroRNAs as biomarkers for myotubular myopathy** N. Maani; D. Gustafson; N. Sabha; A. Ramani; J. Fish; M. Alexander; J. Dowling
	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

14:30-14:50 **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.50 **Characterization of brain white matter pathology in spinal muscular atrophy: a pilot study**
A. Oliveira-Carneiro; M. Oskoui; H. Carlson; M. Brossard-Racine; A. Kirton; N. Mugisha

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.22-56a) - Poster area - Ballroom B1-B2
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.099-118) (VP.50-56a) - 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.111 **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

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

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: Montse Olivé 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: Montse Olivé 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	
13:15-14:30	Lunch, exhibition and posters - Ballroom Salon and Pre Function Space	
13:15-14:45	***Neuromuscular Disorders* Editorial Board Meeting** - Meeting Room 603-604	

14:30-17:30	**Poster viewing** - Poster area - Ballroom B1-B2
17:30-20:00	**Reception (pre-registration required)** - Pier 21

Friday 14th October 2022

06:45	**Registration desk opens**	
07:00-8:30	**Symposium 7** - Ballroom	**Symposium 8** - Argyle
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 **RNA editing by recruiting endogenous ADAR using long RNAs to correct glycine substitutions in COL6-RD** A. Brull; L. Trank; V. Bolduc; C. Bönnemann	
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. Grigenrath; 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
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.151 **Myopathy caused by mutations in the *HNRNPA1* gene**
P. Hackman; S. Välipakka; P. Jonson; J. Sarparanta; A. Vihola; M. Johari; M. Savarese; B. Udd

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.182 **The first case of autosomal recessive limb-girdle muscular dystrophy-5 in Korea**
U. Yun; H. Park

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: Biomarkers or functional outcomes?** - 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) - Poster area - Ballroom B1-B2

16:00-16:20 **Congenital muscular dystrophies** (FP.37-40) – Ballroom
Moderator: Francesco Muntoni

16:00-16:05 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

16:05-16:10 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

16:10-16:15 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

16:15-16:20 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

16:00-16:10 **DMD – Preclinical** (FP.41-42) – Ballroom Salon
Moderator: Thomas Krag

16:00-16:05 FP.41 **Duchenne muscular dystrophy functional muscle organoid-on-a-chip for potential therapies evaluation**
J. Fernández-Costa; A. Tejedera-Villafranca; J. Ramón-Azcón

16:05-16:10 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

16:10-16:20 **Myotonic Dystrophy** (FP.43-44) – Ballroom Salon
Moderator: Benedikt Schoser

16:10-16:15 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

16:15-16:20 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
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** - Ballroom *Moderators: Ichizo Nishino and Lindsay Alfano*
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