

23rd WMS Congress – 2018 - Program

Tuesday 2 October 2018

12:30-16:00	WMS Executive Board Meeting – Room: Lujan
14:00-18:00	Registration opens - Setting up of posters
16:00-18:00	Afternoon refreshments
18:00-18:45	Opening Ceremony – Auditorium
18:45-21:00	Networking Reception

Wednesday 3 October 2018

08:00	Conference desk opens
08:30-09:00	Congress opening - Message from the President Thomas Voit – Auditorium
09:00-10:30	Neuromuscular junction defects, Invited lectures (I.1-2) <i>Chairpersons: Thomas Voit and Marcelo Rugiero</i>
I.1	Age-related neuromuscular junction instability: causes and consequences <i>M. Ruegg; P. Castets; S. Lin; H. Brenner; D. Ham; M. Rich</i>
I.2	New genes and better treatment for congenital myasthenic syndromes <i>D. Beeson</i>
10:30-11:00	Morning refreshments, exhibition and posters
11:00-11:45	Myasthenia gravis; Invited lecture (I.3) - Auditorium <i>Chairpersons: Gisèle Bonne and Bruno Eymard</i>
I.3	From trial-and-error to trials without errors in myasthenia gravis <i>J. Verschuuren</i>
11:45-13:15	Selected oral presentations I - New genes, functions and biomarkers (O.1-6) <i>Chairpersons: Gisèle Bonne and Bruno Eymard</i>
O.1	Recessive mutations in <i>BET1</i> and <i>GOSR2</i> establish Q-SNARE Golgi-vesicle-transport genes as a cause for congenital muscular dystrophy with epilepsy <i>S. Donkervoort; Y. Hu; P. Shieh; J. Koliwer; L. Tsai; B. Cummings; M. Snyder; K. Chao; R. Kaur; D. Bharucha-Goebel; S. Iannaccone; D. MacArthur; A. Foley; M. Schwake; C. Bönnemann</i>
O.2	Mutations in fast skeletal troponin C (<i>TNNC2</i>) cause contractile dysfunction <i>M. van de Locht; J. Winter; S. Conijn; W. Ma; M. Helmes; T. Irving; S. Donkervoort; P. Mohassel; L. Medne; C. Quinn; O. Neto; S. Moore; A. Foley; N. Voermans; C. Bönnemann; C. Ottenheijm</i>
O.3	A patient-derived iPSC model reveals that genotoxic stresses can be risk factors by increasing the causative DUX4 expression in facioscapulohumeral muscular dystrophy (FSHD) <i>M. Sasaki-Honda; T. Jonouchi; M. Arai; A. Hotta; S. Mitsuhashi; I. Nishino; R. Matsuda; H. Sakurai</i>
O.4	Single-cell RNA-sequencing in facioscapulohumeral muscular dystrophy disease etiology and development <i>A. van den Heuvel; A. Mahfouz; S. Kloet; J. Balog; B. van Engelen; R. Tawil; S. Tapscott; S. van der Maarel</i>
O.5	Association of phosphorylated neurofilament heavy chain (pNF-H) with nusinersen treatment of SMA: analyses from the ENDEAR and CHERISH studies <i>B. Darras; R. Finkel; E. Mercuri; C. Sumner; M. Oskoui; E. Tizzano; M. Ryan; G. Zhao; M. Petrillo; C. Stebbins; W. Farwell</i>
O.6	Free Mg ²⁺ intramuscular concentration determined by combined ³¹ P and ¹ H NMR spectroscopy as a potential outcome measure in Duchenne muscular dystrophy <i>H. Reyngoudt; P. Carlier</i>

13:15-14:30

Lunch, exhibition and posters**Sponsor Meeting** – Room: Perdriel

14:30-16:00

Poster session 1: parallel sessions (P.1-97) – Poster area**Limb-girdle muscular dystrophy I (P.1-13)***Chairpersons: Bjarne Udd and Kevin Campbell*

- P.1 **Limb-girdle muscular dystrophy type 2L: clinical, neurophysiological, and imaging correlation in the first reported Brazilian cases**
A. Coimbra Neto; T. Leoni; T. Rosa; C. Iwabe-marchese; A. Martinez; A. Nucci; M. Franca Junior
- P.2 **ANOS - Three different phenotypes and a new histological pattern**
L. Gonzalez-Quereda; G. Garrabou; M. Rodriguez; P. Gallano; A. Sanchez; J. Grau; J. Milisenda
- P.3 **Phenotypic spectrum and muscle pathology in a Chinese cohort with ANO5 recessive mutations**
S. Luo; S. Cai; M. Gao; J. Xi; Z. Liu; D. Yue; J. Lu; C. Zhao
- P.4 **Axial muscular affection in patients with LGMD2L**
T. Khawajazada; J. de Stricker Borch; K. Rudolf; J. Dahlqvist; J. Vissing
- P.5 **Effect of MAPK Inhibition on the differentiation of Rhabdomyosarcoma cell line TE671 combined with CRISPR/Cas9 technology: an *in vitro* model for the study of human muscle diseases**
N. De Luna; X. Suárez-Calvet; M. Garicano; E. Fernández-Simón; R. Rojas-Garcia; J. Diaz-Manera; L. Querol; I. Illa; E. Gallardo
- P.6 **Functional recovery by readthrough therapy in a knock-in mouse model with nonsense dysferlin mutation**
J. Shin; K. Seo; J. Park; D. Kim
- P.7 **Proteomic investigation of muscle-derived proteomic biomarkers of dysferlinopathy**
H. Park; Y. Choi
- P.8 **Clinical outcome study for dysferlinopathy: three years of natural history data for clinical trial readiness**
M. James; A. Mayhew; R. Muni Lofra; M. Jacobs; A. Canal; T. Duong; R. Gee; M. Harman; S. Holsten; L. Lowes; E. Maron; B. Mendez; I. Pedrosa Belmonte; C. Sakamoto; C. Semplicini; C. Siener; S. Thiele; B. Vandervelde; K. Bushby; V. Straub
- P.9 **A comparison of the utility between three muscle strength assessment methods in dysferlinopathy**
N. Miller; L. Lowes; M. James; L. Alfano; A. Mayhew; E. Maron; R. Gee; M. Harman; T. Duong; B. Vandervelde; C. Siener; S. Thiele; B. Mendez; A. Canal; C. Sakamoto; S. Holsten; I. Pedrosa Belmonte; C. Semplicini; V. Straub
- P.10 **Imaging phenotype in dysferlinopathy and its relationship with disease duration and disability are unravelled by heatmaps and random forests**
D. Gómez-Andrés; J. Díaz; F. Munell; A. Sánchez-Montañez; I. Pulido-Valdeolivas; L. Suazo; C. Garrido; J. Bevilacqua
- P.11 **Clinical outcome study in dysferlinopathy: random forest approach to assess the relationship between baseline muscle MRI and longitudinal functional outcome measures**
J. Diaz-Manera; R. Fernández-Torron; M. James; A. Mayhew; M. Jacobs; S. Spuler; J. Day; K. Jones; D. Bharucha-Goebel; E. Salort-Campana; A. Pestronk; M. Walter; C. Paradas; T. Stojkovic; M. Mori-Yoshimura; E. Bravver; E. Pegoraro; J. Mendell; K. Bushby; V. Straub
- P.12 **Clinical outcome study in dysferlinopathy: medical comorbidities and polytherapy in a large population of dysferlinopathy patients**
R. Fernandez-Torron; J. Diaz-Manera; M. James; A. Mayhew; S. Spuler; J. Day; K. Jones; D. Bharucha-Goebel; E. Salort-Campana; A. Pestronk; M. Walter; C. Paradas; T. Stojkovic; M. Mori-Yoshimura; E. Bravver; E. Pegoraro; J. Mendell; Jain Consortium; K. Bushby; V. Straub
- P.13 **Rasch analysis of the individualised neuromuscular Quality of Life Questionnaire administered to patients with dysferlinopathy**
M. James; A. Mayhew; M. Jacobs; S. Spuler; J. Day; K. Jones; D. Bharucha-Goebel; E. Salort-Campana; A. Pestronk; M. Walter; C. Paradas; T. Stojkovic; M. Mori-Yoshimura; E. Bravver; J. Diaz Manera; E. Pegoraro; J. Mendell; K. Bushby; V. Straub
- Duchenne muscular dystrophy - clinical (P.14-30)**
Chairpersons: Alberto Dubrovsky and Jiri Vajsar
- P.14 **The profile of Duchenne muscular dystrophy patients younger than 10 years old from KUKAS registry, Turkey**
A. Karaduman; I. Alemdaroğlu Gürbüz; E. Acar Aslan; M. Güngör; N. Bulut; G. Aydin; Ö. Yilmaz; B. Talim; H. Topaloğlu
- P.15 **First report of natural history and survival in patients with Duchenne muscular dystrophy in Zimbabwe: a retrospective cohort study**
P. Karachunski; J. Dalton; R. Paulson; K. Mitchell; Z. Mugugunyeki; R. Machaka; J. Pazorora
- P.16 **Epidemiology, clinical and genetic features of Duchenne disease in Portugal: a multicentre retrospective study**
C. Garrido; F. Palavra; M. Cardoso; A. Sousa; R. Rocha; D. Alves; M. Santos; M. Vila Real; J. Vieira; T. Coelho; I. Fineza; T. Moreno; M. Santos
- P.17 **Reasons for first visit to neurologists in Chinese patients with dystrophinopathy: a survey study**
L. Wang; M. Xu; H. Li; C. Zhang

- P.18 **The importance of nutrition in Duchenne muscular dystrophy**
I. Verhaart; M. Fiorotto; A. De Luca; S. Wong; R. Quinlivan; Z. Davidson; L. van den Engel-Hoek; M. van Putten; N. de Roos; K. Kinnett; C. Saure; O. Dorchies; I. Roberts; M. Franken-Verbeek; F. De Angelis; N. Goemans; P. Furlong; J. Kuijer; A. Aartsma-Rus; E. Vroom
- P.19 **Prevalence of metabolic disorders in patients with Duchenne muscular dystrophy**
C. Saure; F. De Castro Perez; S. Monges; C. Caminiti
- P.20 **Cognitive performance in Duchenne muscular dystrophy**
M. Miranda; A. Yaeko; C. Sá; L. Grossklauss; F. Favero; M. Voos
- P.21 **Changes over years in the verbal IQ of patients with Duchenne muscular dystrophy**
H. Arahata; A. Miyoshi; A. Watanabe; Y. Kawano; A. Yamamoto; N. Sasagasako
- P.22 **Evaluation of methylphenidate in males with Duchenne muscular dystrophy and a comorbid attention deficit hyperactivity disorder: a preliminary study**
D. Hellebrekers; J. Lionarons; S. Klinkenberg; C. Faber; J. Hendriksen; J. Vles
- P.23 **Longitudinal follow-up of verbal working memory and processing speed in males with Duchenne muscular dystrophy**
D. Hellebrekers; N. Doorenweerd; D. Sweere; S. Kuijk; A. Aartsma-Rus; S. Klinkenberg; J. Vles; J. Hendriksen
- P.24 **Cognition and cerebral structural abnormalities in dystrophinopathies**
P. Tavares; G. Conte; S. Passos; T. Rezende; L. Souza; T. Rosa; S. Ciasca; A. Nucci; M. França Jr
- P.25 **Circadian rhythms in young boys with Duchenne muscular dystrophy**
R. Bendixen; A. Kelleher; N. Little; M. Feltman
- P.27 **Descriptive characteristics of males with Duchenne muscular dystrophy using insurance claims data**
J. Karafilidis; O. Mayer; B. Griffin; K. Higgins
- P.28 **The adult DMD patient. new challenges for an emerging phenotype**
A. Jáuregui; L. Mesa; J. Corderí; F. Chloca; D. Flores; A. Dubrovsky
- P.29 **Analysis of respiratory function of Duchenne muscular dystrophy with Chilaiditi syndrome**
M. Ogasawara; A. Ishiyama; E. Takeshita; Y. Shimizu-Motohashi; H. Komaki; M. Sasaki
- P.30 **Guidance regarding use of implantable cardioverter-defibrillators in Duchenne and Becker muscular dystrophy**
N. Kertesz; A. Kamp; W. Thompson; A. Barth; I. Law; S. Batlivala; N. Hanlon; A. Fournier; C. Spurney; M. Mori-Yoshimura; L. Markham; L. Cripe
- Duchenne muscular dystrophy - imaging and biomarkers (P.31-38)**
Chairpersons: Lee Sweeney and Pierre Carlier
- P.31 **Serum creatinine: a promising biomarker for distinguishing Duchenne muscular dystrophy from Becker muscular dystrophy in patients aged ≤ 3 Years**
L. Wang; M. Chen; R. He; Y. Sun; J. Yang; L. Xiao; J. Cao; H. Zhang; C. Zhang
- P.32 **Chemokine CXCL12 and osteopontin are highly expressed in Duchenne muscular dystrophy patients**
Y. Maeda; M. Ishizaki; Y. Nakajyo; Y. Yonemochi; T. Ueyama
- P.33 **Skeletal muscle T1 mapping correlates with MFM scores in dystrophinopathies**
L. Souza; P. Tavares; S. Passos; C. Iwabe-Marchese; T. Rosa; A. Nucci; M. França Jr; S. Dertkigil
- P.34 **MR biomarkers in imaging DMD clinical trial network**
S. Forbes; R. Willcocks; W. Triplett; H. Arora; W. Rooney; D. Wang; M. Daniels; E. Finanger; R. Finkel; G. Tennekoon; H. Sweeney; G. Walter; K. Vandenborne
- P.35 **Higher MRI muscle fat fraction at similar age is associated with earlier loss of ambulation in Duchenne muscular dystrophy**
K. Naarding; H. Reyngoudt; E. van Zwet; M. Hooijmans; B. Wong; C. Tian; I. Rybalsky; K. Shellenbarger; J. Le Louër; P. Carlier; H. Kan; E. Niks
- P.36 **Early impact on disease identified for Ezutromid using magnetic resonance spectroscopy (MRS) in Duchenne muscular dystrophy**
A. Heatherington; F. Muntoni; K. Vandenborne; G. Layton; D. Roblin & Imaging DMD Consortium; Phase Out DMD Study Group
- P.37 **Upper extremity quantitative muscle ultrasound is related to disease severity in boys with Duchenne muscular dystrophy**
W. Stuij; M. Jansen; I. de Groot
- P.38 **Description of Becker muscular dystrophy cardiomyopathy natural history by cardiac magnetic resonance imaging from the first to third decades provides insight for cardiac surveillance**
T. Johnston; K. Hor; M. Mah; L. Cripe
- Congenital myopathies, general and RYR1 (P.39-55)**
Chairpersons: Jorge Bevilacqua and Edmar Zanoteli
- P.39 **Congenital cytoplasmic body myopathy – a nosological clarification**
M. Schülke; W. Stenzel; M. Schwarz; H. Goebel
- P.40 **The distinct clinical phenotype of PIEZO2 loss of function**
D. Saade; M. Lee; D. Bharucha-Goebel; S. Donkervoort; S. Neuhaus; K. Alter; C. Zampieri; C. Stanley; J. Matsubara; A. Nickolls; A. Micheil Innes; J. Mah; C. Grosman; A. Nascimento; J. Colomer; F. Munell; G. Haliloglu; A. Foley; A. Chesler; C. Bönnemann

- P.41 **Dominant *TNNC2* mutations cause a distinct congenital myopathy with vocal cord paralysis, ophthalmoplegia and clinical improvement over time**
S. Donkervoort; P. Mohassel; N. Voermans; C. Quinn; M. van de Loch; J. de Winter; S. Conijn; M. Helmes; L. Medne; O. Lopes Abath Neto; S. Moore; C. Ottenheijm; A. Foley; C. Bönnemann
- P.42 **Congenital hyporegenerative microcytic anemia of unknown origin with XMEA-like muscle pathology**
J. Reimann; C. Scholtes; K. Cremer; S. Schoenberger; W. Kunz
- P.43 **Prevalence of cytoplasmic bodies in a large series of diagnostic paediatric muscle biopsies**
 M. Aizpurua; F. Cepas; A. Sarkozy; A. Manzur; F. Muntoni; C. Sewry; R. Phadke
- P.44 **Mutations in the myomaker gene causes Carey-Fineman-Ziter syndrome with muscle fiber hypertrophy**
C. Hedberg-Oldfors; C. Lindberg; A. Oldfors
- P.45 **A new congenital myopathy with multiple structured cores**
E. Malfatti; X. Lornage; J. Böhm; G. Brochier; R. Carlier; J. Laporte; M. Fardeau; N. Romero
- P.46 **Novel *ASCI* mutations causing prenatal-onset muscle weakness with arthrogryposis and congenital bone fractures**
J. Böhm; E. Malfatti; E. Oates; K. Jones; N. Romero; J. Laporte
- P.47 ***ASCI*-related myopathy is associated with defects in myoblast proliferation and muscle growth: defining the phenotypic spectrum and understanding the pathogenesis of an emerging congenital myopathy**
 I. Duband-Goulet; F. Catervi; E. Cabet; L. Davignon; C. Genetti; T. Gidaro; A. Kopari; S. Coppen; E. Pierce-Hoffman; A. Beggs; L. Servais; A. Ferreira
- P.48 **Hypercontractile congenital muscle stiffness**
C. Camelo; A. Da Silva; U. Reed; C. Bönnemann; E. Zanoteli
- P.49 **Redefining morphological spectrum of *RYR1* recessive myopathies**
M. Garibaldi; J. Rendu; J. Brocard; E. Iacene; M. Beuvin; G. Brochier; C. Labasse; A. Madelaine; E. Malfatti; J. Bevilacqua; F. Lubieniecki; S. Monges; A. Taratuto; I. Marty; N. Romero
- P.50 **Loss of FKBP12-*RYR1* binding *ex vivo* is a post-translational modification consistently evident across diverse ryanodine receptor 1-related myopathies**
J. Todd; J. Witherspoon; A. Kushnir; S. Reiken; M. Razaqyar; M. Shelton; I. Chrismer; C. Grunseich; A. Mankodi; C. Bönnemann; K. Meilleur
- P.51 **Familial variation in phenotype in *RYR1*-related myalgia-rhabdomyolysis syndrome**
N. Witting; T. Solheim; J. Dahlqvist; N. Poulsen; M. Duno; J. Vissing
- P.52 **Core and cytoplasmic bodies in a patient with asymptomatic hyperCKemia caused by a *RYR1* p.Arg163Cys mutation**
L. Gonzalez-Quereda; A. Pellisé; N. Vidal; M. Rodriguez; P. Gallano; M. Olivé
- P.53 **Clinical, genetic and pathological characterization of a wide paediatric cohort of patients with dominant and recessive *RYR1*-related myopathy**
M. Sa; M. DiStefano; R. Mein; R. Phadke; L. Feng; P. Munot; R. Quinlivan; A. Manzur; S. Robb; M. Main; C. Sewry; A. Sarkozy; F. Muntoni
- P.54 **Forced and slow vital capacities in *RYR1*-RM**
I. Chrismer; J. Witherspoon; B. Drinkard; M. Stockman; M. Shelton; A. Kuo; C. Allen; J. Todd; M. Jain; M. Meilleur
- P.55 **Historical perspective and proposal for a unified ryanodine receptor 1-related myopathies nomenclature**
T. Lawal; J. Todd; J. Witherspoon; C. Bönnemann; S. Hamilton; J. Dowling; K. Meilleur
- Congenital myasthenic syndromes and myasthenia (P.56-75)**
Chairpersons: Duygu Selcen and Ulrike Schara
- P.56 **Clinical manifestation and associated co-morbidities in patients with juvenile-onset myasthenia gravis: a retrospective study**
P. Karachunski
- P.57 **Clinical features in juvenile myasthenia gravis in an Argentinian cohort**
 M. García Erro; E. Cavassa; J. Muntadas; M. Pauni; G. Vázquez
- P.58 **Myasthenia gravis anti-MuSK (MuSK-MG): therapeutic experience in 27 patients**
 M. Rugiero; V. Salutto; V. Alvarez; M. Bettini; N. Genco; C. Mazia
- P.59 **Ocular vestibular evoked myogenic potentials in myasthenia gravis**
R. de Meel; K. Keene; M. Tannemaat; J. Verschuuren
- P.60 **Respiratory dysfunction in childhood myasthenia**
J. Vajsar; H. Katzberg; H. Qashqari; N. Chrestian; I. Narang
- P.61 **Pembrolizumab induced myasthenia gravis and necrotizing myopathy with severe respiratory failure**
M. Rugiero; M. Bettini; F. Silveira; F. Sosa Albacete; S. Christiansen
- P.62 **A case of clinically apparent myasthenia gravis after resection of non-myasthenic thymic cyst**
 S. Ho; J. So; D. Bae
- P.63 **Myasthenia gravis like syndrome after botulinum toxin type A injections for calf reduction**
J. So; S. Ho; D. Bae
- P.64 **Living with myasthenia gravis**
 E. Louet; S. Misdrahi; C. Orblin Bedos; S. Birnbaum; J.Y. Hogrel; P. Portero; B. Clair; B. Eymard; S. Demeret; G. Bassez; S. Berrih-Aknin; A. Jobic; P. Aegerter; P. Thoumie; T. Sharshar; M. Gargiulo; MGEX Study group

- P.65 **Muscular pathological features in Lambert-Eaton myasthenic syndrome**
Y. Zhang; R. Ban; H. Liu; C. Pu; Q. Shi
- P.66 **Congenital myasthenic syndromes: how do clinicians face diagnostic complexity and long-term prognosis in 2018?**
B. Eymard; D. Sternberg; M. Mayer; T. Stojkovic; E. Fournier; S. Nicole; A. Behin; P. Laforêt; L. Servais; S. Bauché; B. Fontaine; D. Hantaï; M. Fardeau; N. Romero
- P.67 **Congenital myasthenic syndromes due to impaired principal coupling pathway in the ϵ -subunit of muscle acetylcholine receptor**
X. Shen; D. Selcen; J. Brengman; S. Shen; H. Durmus; V. Preethish-Kumar; A. Yuceyar; S. Vengalil; A. Nalini; F. Deymeer; S. Sine; A. Engel
- P.68 **The p. N88K mutation in the *RAPSN* gene in Brazilian patients with congenital myasthenic syndrome**
E. Estephan; A. Zambon; P. Marchiori; A. Silva; C. Moreno; U. Reed; A. Töpf; H. Lochmüller; E. Zanoteli
- P.69 **New *AGRN* mutations in a patient with limb-girdle congenital myasthenic syndrome**
S. Coppens; G. Glibert; N. Deconinck
- P.70 **New homozygous mutation in *DPAGT1* gene leading to LG-CMS with tubular aggregates**
T. Gidaro; L. Vandenbrande; E. Malfatti; C. Labasse; P. Carlier; N. Romero; L. Servais; J. Böhm
- P.71 **Unexpected findings of congenital myasthenic syndromes by NGS testing using an extended gene panel on neuromuscular patients in Norway**
C. Jonsrud; P. Aden; G. Hansen; M. Mork; B. Nygård; T. Popperud; M. Rasmussen; N. Songstad; K. Ørstavik; T. Fagerheim
- P.72 **Clinical features of congenital myasthenic syndrome due to mutations in *COL13A1***
P. Rodríguez Cruz; J. Palace; D. Beeson
- P.73 **Characterization of an Indian congenital myasthenic syndrome cohort by whole exome sequencing**
S. Balaraju; A. Töpf; P. Veeramani; S. Vengalil; K. Polavarapu; S. Nashi; J. Kirschner; R. Horvath; N. Atchayaram; H. Lochmüller
- P.74 **Five years of salbutamol treatment in a girl with congenital myasthenic syndrome caused by mutations in *COL13A1***
F. Munell; D. Gomez-Andrés; L. Costa Comellas; A. Macaya; M. Gratacós; M. Dusl; J. Senderek; H. Lochmüller
- P.75 **Development of a home-based assessment tool for monitoring fluctuations in symptoms in the myasthenic population**
V. Selby; G. Ramdharry; M. Hanna; F. Muntoni
- SMA clinical data, outcome measures and registries (P.76-97)**
Chairpersons: Laurent Servais and Eduardo Tizzano
- P.76 **A prospective functional assessment in type 2 spinal muscular atrophy in the Spanish population. Importance of the age on disease progression rate**
D. Natera - de Benito; A. Frongia; M. Alarcón; A. Borrás; J. Armas; J. Exposito; L. Carrera; L. Martorell; D. Moya; N. Padros; S. Roca; M. Vigo; J. Medina; J. Colomer; C. Ortez; A. Nascimento
- P.77 **Two year longitudinal data of the European prospective natural history study of patients with type 2 and 3 spinal muscular atrophy**
A. Chabanon; M. Anoussamy; A. Daron; Y. Péréon; C. Cancès; C. Vuillerot; N. Goemans; J. Cuisset; V. Laugel; U. Schara; T. Gidaro; A. Seferian; L. Lowes; P. Carlier; JY. Hogrel; C. Czech; R. Hermosilla; O. Kwaja; L. Servais
- P.78 **Clinical and molecular features of proximal spinal muscle atrophy in Portugal: a multicentre retrospective study**
M. Oliveira Santos; C. Falcão de Campos; C. Garrido; I. Conceição; F. Palavra; L. Negrão; J. Pedro Vieira; C. Mendonça; T. Coelho; I. Fineza; M. Santos; T. Moreno
- P.79 **The relationship between function and muscle strength in the upper limb in a cohort of children with spinal muscular atrophy type II and III – a prospective study**
E. Milev; V. Selby; J. Reznik; R. Tillmann; M. Iodice; M. Scoto; JY. Hogrel; F. Muntoni
- P.80 **Survival and ventilation among those with type I spinal muscular atrophy: results from the 2017 Cure SMA membership survey**
L. Belter; J. Jarecki; C. Jones; A. Paradis; M. Jhaveri; S. Reyna; K. Hobby
- P.81 **Anthropometric and nutritional assessment in SMA type II and III**
C. Saure; F. de Castro Perez; S. Monges
- P.82 **Vitamin D status among patients with spinal muscular atrophy**
M. Martínez-Jalile; A. Lozano-Arango; C. Diemer; B. Suárez; K. Alvarez; C. Castiglioni
- P.83 **Longitudinal study of body composition and bone mass in spinal muscular atrophy type 2/3**
N. DiIorgi; E. Medone; G. Brigati; S. Notarnicola; C. Panicucci; C. Fiorillo; M. Pedemonte; C. Minetti; M. Maghnie; C. Bruno
- P.84 **Clinical discordance in spinal muscular atrophy siblings: the exception or the rule?**
S. Monges; J. Mozzoni; M. Franchi; S. Medrano; L. Gravina; H. Aráoz; F. de Castro; V. Aguerre; L. Alías; L. Chertkoff; E. Tizzano; S. Bernal
- P.85 **Cognitive performance of children with 5q-spinal muscular atrophy: a systematic review**
G. Polido; M. Miranda; N. Carvas Junior; F. Caromano; U. Reed; E. Zanoteli; M. Voos
- P.86 **Cognitive assessment in spinal muscular atrophy type 1-2 using eye tracking system**
L. Paternoster; S. Bajot; G. Deliens; N. Goemans; L. Servais; N. Deconinck
- P.87 **Use of the ACTIVE-mini for quantifying movement in infants with spinal muscular atrophy**
L. Nelson; L. Alfano; D. Chen; N. Miller; M. Dugan; S. Rust; E. Lin; S. Lin; S. Wang-Price; C. Swank; M. Thompson; L. Lowes

- P.88 **Associations between NMR, electrophysiological, strength and function variables in SMA type 2 and 3**
J.Y. Hogrel; M. Annoussamy; A. Chabanon; A. Daron; Y. Péréon; C. Cances; C. Vuillerot; N. Goemans; J. Cuisset; V. Laugel; U. Schara; E. Gargaun; T. Gidaro; A. Seferian; S. Turk; R. Hermosilla; E. Fournier; P. Baudin; P. Carlier; L. Servais; Study Group
- P.89 **More than just fun and games: ACTIVE workspace volume video game quantifies meaningful change in function in individuals with spinal muscular atrophy**
 L. Alfano; N. Miller; M. Iammarino; M. Moore-Clingenpeel; S. Lowes; M. Dugan; M. Waldrop; K. Flanigan; G. Noritz; C. Tsao; S. Al Zaidy; J. Kissel; L. Lowes
- P.90 **The shifting landscape of SMA: development of a new mild mouse model to better understand disease in aging**
 M. Deguise; A. Beauvais; A. Tiernay; B. Paul; E. McFall; Y. De Repentigny; R. Kothary
- P.91 **Muscle imaging and function in patients with spinal and bulbar muscular atrophy**
J. Dahlqvist; S. Oestergaard; N. Poulsen; J. Vissing
- P.92 **Clinical and molecular characterization of non-5q spinal muscular atrophies**
 S. Calligaris; A. Guzmán; A. Savransky; C. Buompadre; F. Lubieniecki; C. Tesi Rocha; D. Tiziano; V. Fano; E. Tizzano; P. Gravina; S. Monges
- P.93 **Collaborative data collection by TREAT-NMD registries to support post-marketing surveillance in spinal muscular atrophy**
J. Bullivant; V. Hodgkinson-Brechenmacher; M. Rodrigues; Study Group; 2; V. Straub; H. Dawkins; C. Campbell; N. Goemans
- P.94 **Cure SMA membership: findings from the 2018 membership survey**
L. Belter; C. Jones; A. Paradis; M. Jhaveri; S. Reyna; K. Hobby; J. Jarecki
- P.95 **Economic burden of infant-onset (type 1) spinal muscular atrophy: a retrospective claims database analysis**
O. Dabbous; J. Seda; M. Droege; D. Sproule
- P.96 **Number needed to treat in spinal muscular atrophy type 1 with AVXS-101 relative to nusinersen**
O. Dabbous; M. Cloutier; A. Guerin; I. Pivneva; E. Wu; M. Droege; D. Sproule
- P.97 **Development of a decision-analytic model for the economic evaluation of newborn screening for spinal muscular atrophy**
T. Dangouloff; M. Hiligsmann; J. Caberg; F. Boemer; L. Servais
- 15:45-16:15 **Afternoon refreshments, exhibition and posters**
- 16:00-17:30 **Poster session 2: parallel sessions (P.98-195) – Poster area**
- LGMD autosomal recessive and dominant (P.98-110)**
Chairpersons: Isabelle Richard and Ivailo Tournev
- P.98 **AAV-mediated gene transfer of *FKRP* for therapy of LGMD2I**
 E. Gicquel; S. Brown; I. Richard
- P.99 **Limb-girdle muscular dystrophy 2Z in a Bulgarian family**
T. Chamova; A. Taneva; M. Gospodinova; D. Zlatareva; K. Johnson; A. Töpf; V. Straub; I. Tournev
- P.100 ***BVES* loss-of-function mutations in limb-girdle muscular dystrophy 2X with cardiac conduction disorders**
 I. Nelson; W. De Ridder; B. Asselbergh; B. De Paepe; M. Beuvin; R. Ben Yaou; A. Boland; J. Deleuze; T. Maisonobe; B. Eymard; J. De Bleecker; S. Symoens; R. Schindler; T. Brand; A. Töpf; K. Johnson; V. Straub; P. De Jonghe; J. Baets; G. Bonne
- P.101 **Recurrent rhabdomyolysis and subtle proximal weakness in two female siblings diagnosed with alpha sarcoglycanopathy and a review of the literature**
 H. Sampaio; M. Farrar; A. Al Safar
- P.102 **Limb girdle muscular dystrophy type 2A: divergent features of mitochondrial deficiencies associated with novel calpain-3 mutations**
R. El-Khoury; R. Sawaya; M. Lamaa; M. Ahdab-Barmada
- P.103 **A very early onset of calpainopathy (LGMD2A)**
R. Andrade; L. Lima; M. Melo; A. Miranda
- P.104 **Coalition to Cure Calpain 3: a patient organization committed to treating and ultimately curing limb girdle muscular dystrophy type 2A**
J. Levy; J. Boslego; M. Wrubel; L. Wrubel; M. Spencer
- P.105 **Recessive limb-girdle muscular dystrophies in a Brazilian tertiary hospital: epidemiological, clinical and genetic profile**
A. Coimbra Neto; T. Leoni; A. Martinez; A. Nucci; M. Franca Junior
- P.106 **Limb girdle muscular dystrophy: steps toward a comprehensive patient reported outcome**
 M. Hunter; A. Hatzipolakis; C. Heatwole; M. Wicklund; J. Statland; C. Weihl; N. Johnson
- P.107 **Limb-girdle muscular dystrophy in Taiwan: a referral center experience**
W. Liang; C. Wang; X. Tian; W. Chen; T. Kan; I. Nishino; L. Wong; Y. Jong
- P.108 **Functional characterization of *DNAJB6* J-domain mutations**
 P. Jonson; J. Sarparanta; S. Kawan; H. Luque; M. Jokela; B. Udd
- P.109 **Clinical presentation of a new transportinopathy phenotype in an Hungarian family LGMD D2**
C. Angelini; R. Marozzo; V. Pegoraro

- P.110 **Limb girdle muscular dystrophy type 1G caused by p.D378N mutation in *HNRPDL* gene with distal muscle weakness in a Chinese family**
Y. Da; Y. Sun; H. Chen; Y. lu; L. Di; X. Wen
- DMD clinical therapies I** (P.111-123)
Chairpersons: Francesco Muntoni and Nicolas Deconinck
- P.111 **Preservation of function over time as measured by North Star ambulatory assessment in boys with nonsense mutation Duchenne muscular dystrophy treated with ataluren**
C. McDonald; L. Wei; F. Jin; G. Elfring; P. Trifillis; M. Souza; J. McIntosh; S. Peltz; F. Muntoni
- P.112 **STRIDE™: A patient registry study examining the use of ataluren (Translarna™) in patients with nonsense mutation muscular dystrophy (nmdMD)**
F. Muntoni; I. Desguerre; M. Guglieri; E. Mercuri; A. Nascimento Osorio; J. Kirschner; M. Tulinius; F. Buccello; A. Delage; G. Elfring; C. Werner; T. Schilling; P. Trifillis
- P.113 **Long-term effect - four years - of ataluren in fourteen patients with nonsense mutation Duchenne muscular dystrophy**
C. Ortez; J. Medina; M. Vigo; O. Moya; N. Padros; D. Natera De Benito; L. Carrera; J. Colomer; I. Zschaek; C. Jimenez - Mallebrera; L. Solé; M. Cubells; C. Jou; A. Nascimento
- P.114 **Vamorolone as a replacement for corticosteroids in Duchenne muscular dystrophy: phase 2a results in 48 DMD boys**
K. Nagaraju; P. Clemens; J. Damsker; J. McCall; L. Mengle-Gaw; L. Conklin; E. Smith; D. Castro; J. Mah; C. McDonald; N. Kuntz; R. Finkel; M. Guglieri; M. Tulinius; Y. Nevo; M. Ryan; R. Webster; A. Smith; L. Morgenroth; E. Hoffman
- P.115 **Targeting NADPH oxidases in models of Duchenne muscular dystrophy**
H. Ismail; O. Dorchie; E. Gay; L. Neff; L. Scapozza
- P.116 **Edasalonexent, an NF- κ B inhibitor, slows disease progression over more than a year compared to control period in 4 to 7-year old patients with Duchenne muscular dystrophy**
F. Richard; K. Vandenborne; H. Sweeney; E. Finanger; G. Tennekoon; P. Shieh; R. Willcocks; G. Walter; W. Rooney; S. Forbes; W. Triplett; S. Yum; M. Mancini; J. MacDougall; A. Fretzen; P. Bista; A. Nichols; J. Donovan
- P.117 **Edasalonexent, an oral NF- κ B inhibitor, in development for treatment of Duchenne muscular dystrophy: the phase 3 POLARIS study design**
R. Finkel; C. McDonald; H. Sweeney; M. Mancini; J. MacDougall; J. Donovan
- P.118 **Assessing the safety and efficacy of MNK-1411 in patients with Duchenne muscular dystrophy in a multicenter, double-blind, placebo-controlled, multiple-dose study**
B. Due; P. Becker; T. Crawford
- P.119 **Tamoxifen in Duchenne muscular dystrophy: rationale and protocol for a multicentre, randomised, double-blind, placebo-controlled, phase 3 safety and efficacy 48-week trial**
S. Schmidt; P. Hafner; A. Orsini; D. Rubino-Nacht; O. Dorchie; A. Nascimento Osorio; U. Schara; S. Spinty; H. Topaloglu; D. Fischer
- P.120 **Epicatechin improves biomarkers of muscle growth and regeneration, oxidative stress, and NO reserve, and improves skeletal muscle exercise response in non-ambulatory DMD patients with presymptomatic cardiomyopathy**
C. McDonald; E. Henricson; Y. Dayan; A. Nicorici; E. Goude; F. Villareal; S. Dugar
- P.121 **Phase II study of TAS-205 in patients with Duchenne muscular dystrophy: subgroup analyses**
T. Nakayama; S. Kuru; H. Komaki; S. Takeda
- P.122 **A phase III clinical study assessing the efficacy and safety of idebenone in patients with Duchenne muscular dystrophy taking concomitant glucocorticoids (SIDEROS)**
G. Buyse; R. Drake; S. Hasham; R. Quinlivan; E. Mercuri; O. Mayer
- P.123 **A randomized, placebo-controlled, double-blind, phase 1b/2 study of the novel anti-myostatin adnectin RG6206 (BMS-986089) in ambulatory boys with Duchenne muscular dystrophy**
K. Wagner; B. Wong; B. Byrne; H. Sweeney; L. Jacobsen; G. Tirucherai; M. Rabbia; J. Dukart; H. Kletz; M. Krishnan; C. Bechtold
- DMD clinical therapies II** (P.124-135)
Chairpersons: Ketan Patel and Ronald Cohn
- P.124 **Eteplirsen is well tolerated in adults with mild or moderate renal impairment**
C. Mix; E. Naughton; S. Forte
- P.125 **Eteplirsen treatment attenuates respiratory decline in ambulatory and non-ambulatory patients with Duchenne muscular dystrophy**
N. Khan; L. Han; B. Kinane; H. Gordish-Dressman; L. Lowes; C. McDonald; CINRG DNHS Investigators
- P.126 **Respiratory function decline in eteplirsen-treated patients diverges from natural history comparators over time**
N. Khan; L. Han; B. Kinane; H. Gordish-Dressman; L. Lowes; C. McDonald
- P.127 **Identification of potent, muscle-targeting investigational stereo-pure oligonucleotides for exon 53 DMD therapy**
A. Durbin; C. Shivalila; N. Kothari; C. Rinaldi; G. McClorey; I. Antonijevic; M. Byrne; J. Davis II; L. Guo; N. Iwamoto; F. Liu; K. Longo; G. Lu; P. Nadella; E. Purcell-Estabrook; S. Standley; Z. Zhong; J. Zhang; M. Wood; C. Vargeese
- P.128 **A phase II, dose finding study to assess the safety, tolerability, pharmacokinetics, and pharmacodynamics of NS-065/NCNP-01 in boys with Duchenne muscular dystrophy** P. Clemens; V. Rao; A. Connolly; A. Harper; J. Mah; E. Smith; C. McDonald; L. Morgenroth; H. Osaki; E. Hoffman

- P.129 **A Japanese phase I/II study of NS-065/NCNP-01, exon 53 skipping drug, in patients with Duchenne muscular dystrophy - a dose-finding study**
H. Komaki; Y. Takeshima; T. Matsumura; S. Ozasa; M. Funato; Y. Egawa; S. Takeda
- P.130 **CRISPR/Cas9 and TALEN edit the DMD genetic mutation in golden retriever muscular dystrophy**
 S. Mata Lopez; C. Balog; S. Vitha; M. Bettis; H. Barnett; J. Krnegay; P. Nghiem
- P.131 **AAV-mediated gene transfer with GALGT2 in Duchenne muscular dystrophy: design of an ongoing phase I/II clinical trial**
K. Flanigan; L. Chicoine; J. Cheatham; S. Cheatham; T. Simmons; L. Lowes; M. Iammarino; N. Miller; L. Alfano; F. Rinaldi; M. Waldrop; D. Zygmunt; R. Xu; P. Martin
- P.132 **Identification of developmental myosin positive fibres acts both as a clinical biomarker for muscle disease and an important component of the process to confirm ezutromid target engagement**
J. Tinsley; F. Muntoni; G. Layton; C. Faelan; J. Patterson-Kane; A. Hetherington; K. Davies
- P.133 **The burden of participation in a clinical trial for boys with Duchenne muscular dystrophy**
 S. Geuens; J. Willen; C. Antonis; N. Goemans; L. De Waele
- P.134 **Comparing home-based respiratory function monitoring to hospital-based spirometry in Duchenne muscular dystrophy**
G. Buysse; T. Meier; M. Leinonen; S. Hasham; O. Mayer; T. Voit
- P.135 **DMD-HUB: expanding clinical trial capacity for Duchenne muscular dystrophy, 1 year on**
E. Heslop; M. Guglieri; K. Bushby; C. Turner; B. Crow; E. Crossley; A. Johnson; M. Scoto; F. Muntoni; V. Straub
- Congenital myopathies (CNM) (P.136-148)**
Chairpersons: Johann Böhm and Ronald Cohn
- P.136 **INCEPTUS pre-phase 1, prospective, non-interventional, natural history run-in study to evaluate subjects aged 3 years and younger with X-linked myotubular myopathy: preliminary findings**
P. Shieh; W. Müller-Felber; B. Smith; C. Bönnemann; J. Dowling; N. Kuntz; F. Muntoni; L. Servais; D. Bilder; T. Duong; R. Graham; M. Jain; M. Lawlor; V. MacBean; M. Noursalehi; T. Pitts; G. Rafferty; S. Rico; S. Prasad
- P.137 **Novel deep intronic mutations in MTM1 in X-linked myotubular myopathy**
T. Kumutpongpanich; S. Hayashi; A. Iida; M. Okubo; M. Matsuo; S. Noguchi; I. Nishino
- P.138 **The CHOP INTEND scale (Children's Hospital of Philadelphia infant test of neuromuscular disorders) in X-linked myotubular myopathy: content validity and psychometric performance**
 S. Rico; D. Bilder; T. Duong; E. James; M. Noursalehi; S. Bergman; G. Harding; S. Mannix; D. Phillips; C. Abel; S. Prasad
- P.139 **Mortality and respiratory support in X-Linked myotubular myopathy: The RECENSUS Study, an international, multicenter, retrospective medical record review**
 R. Graham; B. Byrne; S. de Chastonay; T. Haselkorn; I. Hughes; E. James; N. Kuntz; J. Simon; M. Yang; Z. Yu; S. Yum; S. Prasad; S. Rico; A. Beggs
- P.140 **Clinical changes over time in a European and North-american cohort of patients with X-linked myotubular myopathy**
M. Annoussamy; C. Lilien; T. Gidaro; V. Chê; U. Schara; A. D'Amico; J. Dowling; B. Darras; A. Daron; M. Mayer; A. Hernandez; C. Vuillerot; S. Fontaine; C. deLattre; R. Bellance; V. Biancalana; A. Buj-Bello; JY. Hogrel; H. Landy; L. Servais
- P.141 **High frequency of manifesting carriers in the recessive X-linked myotubular myopathy**
L. Souza; C. Almeida; L. Silva; R. Pavanello; J. Gurgel-Giannetti; E. Zanoteli; M. Zatz; P. Otto; M. Vainzof
- P.142 **X-linked myotubular myopathy with the presence of nemaline rods on muscle biopsy**
H. Gonorazky; K. Amburgey; C. Hawkins; J. Dowling
- P.143 **Myostatin as a novel blood-based biomarker for antisense oligonucleotide-mediated Dnm2 knockdown to treat myotubular myopathy in mice**
 S. Buono; C. Koch; A. Robé; C. Kretz; R. Gomez Oca; S. Guo; M. Depla; B. Monia; J. Laporte; L. Thielemans; B. Cowling
- P.144 **Targeting dynamin 2 rescues the three main forms of centronuclear myopathies**
 H. Tasfaout; S. Buono; I. Prokic; J. Ross; C. Kretz; S. Guo; P. Koebel; B. Monia; M. Bitoun; J. Ochala; J. Laporte; B. Cowling
- P.145 **Myotubular and centronuclear myopathy patient registry: accelerating the pace of research and treatment**
J. Bullivant; L. Murphy; K. Napier; L. Render; A. Hunter; M. Spring; A. Lennox; H. Lochmuller; M. Bellgard; C. Marini-Bettolo
- P.146 **Baseline characteristics of patients with centronuclear myopathy due to mutations in DNM2 gene enrolled in a European prospective natural history study**
M. Annoussamy; A. Grangé; C. Lilien; V. Chê; D. Duchêne; T. Gidaro; A. Behin; J. Baets; A. D'Amico; A. Daron; M. Bitoun; K. Paradis; L. Thielemans; S. Van Rooijen; L. Servais
- P.147 **Novel SPEG mutations in congenital myopathy without centralized nuclei**
X. Lornage; P. Sabouraud; B. Lannes; D. Gaillard; V. Biancalana; J. Böhm; J. Laporte
- P.148 **Centronuclear myopathy with BIN1-like myopathology and DNM2 mutation**
T. Rosa; J. Domingues; C. Martins Jr; J. Domingos; C. Iwabe-Marchese; E. Pacheco; L. Queiroz; M. França Jr; A. Nucci
- Inflammatory myopathies; (P.149-166)**
Chairpersons: Ichizo Nishino and Werner Stenzel
- P.149 **Characterization of a cohort of Chinese inflammatory myopathy patients**
Y. Luo; H. Duan; Q. Li; F. Bi
- P.150 **Clinical and muscle imaging follow-up in a cohort of patients with anti-SRP antibody myopathy**
 Y. Zhao; W. Zhang; Z. Wang; Y. Yuan

- P.151 **Spontaneous recovery in a child with anti-HMGCR autoimmune necrotizing myopathy**
B. Suárez; A. Lozano; A. Díaz; X. Ortega; J. Diaz; G. Calcagno; C. Hervias; J. Bevilacqua; C. Castiglioni
- P.152 **Early-onset anti-HMGCR myopathy associated with muscle mitochondrial alterations and calpain-3 deficiency**
M. Villanova; Y. Ivanovic-Barbeito; N. Romero; J. Nectoux; T. Mongini; F. Leturcq; E. Malfatti
- P.153 **Tight association between microinfarction and capillary MAC deposition in dermatomyositis**
M. Inoue; A. Uruha; J. Charuel; L. Musset; S. Suzuki; M. Kuwana; T. Mimori; I. Nishio
- P.155 **Brachio-cervical myopathy as the clinical presentation of scleroderma. Case series**
A. Alonso-Jimenez; X. Suárez-Calvet; I. Castellví; E. Gallardo; J. Diaz-Manera
- P.156 **Brachio-cervical inflammatory myopathy with lymphoid follicle-like structures in a patient with scleroderma**
A. Silva; M. Vianna; R. Mendonça; E. Zanoteli
- P.157 **Dysfunctional T cells in immune mediated necrotizing myopathy, inclusion body myopathy and immune toxicity related myopathy**
S. Knauß; C. Preuß; N. Fischer; Y. Allenbach; H. Radbruch; V. Matyash; M. Endres; H. Goebel; O. Benveniste; W. Stenzel
- P.158 **Kv1.3⁺ cells in blood and muscle from patients with sporadic inclusion body myositis**
T. Mozaffar; G. Coulis; J. Kastenschmidt; M. Wencel; S. Villalta
- P.159 **Feasibility and validation of modified oculobulbar facial respiratory score (mOBFRS) in sporadic inclusion body myositis**
N. Goyal; M. Wencel; N. Araujo; E. Medina; L. Zhang; D. Nguyen; T. Mozaffar
- P.160 **A bioinformatics approach to define the aggregation capacity of the myofiber proteome in inclusion body myositis**
C. Wehl; P. Ciryam; A. Guttsches; K. Marcus; R. Morimoto; M. Vendruscolo; R. Kley
- P.161 **A rare case of distal myositis in a patient with thymoma**
J. Shin; A. Lee; S. Choi; Y. Hong; J. Sung
- P.162 **IgG-4 related myositis - a new entity among inflammatory myopathies**
V. Casteleyn; H. Radbruch; H. Goebel; U. Schneider; W. Stenzel
- P.163 **Myositis and fasciitis due to disseminated histoplasmosis**
A. Silva; M. Vianna; H. Esteves; F. Comello; E. Zanoteli
- P.164 **Predictive value of cerebral 18F-FDG PET for diagnosing macrophagic myofasciitis: an individual SVM-based approach**
P. Blanc-Durand; A. Van Der Gucht; E. Guedj; M. Abulizi; M. Aoun Sebaiti; E. Itti; F. Authier
- P.165 **Sporadic late-onset nemaline myopathy**
E. Naddaf; M. Milone; A. Kansagra; F. Buadi; T. Kourelis

SMA therapies I (P.167-182)

Chairpersons: Eugenio Mercuri and Arthur Burghes

- P.167 **Evaluating the respiratory health of children with spinal muscular atrophy type 1 on nusinersen under the Expanded Access Program (EAP)**
L. Edel; F. Muntoni; V. Robinson; C. Grime; F. Abel; A. Manzur; P. Munot; M. Scoto; E. Chan
- P.168 **Treatment by nusinersen in spinal muscular atrophy type 1 patients older than 7 months - 14 months follow-up**
K. Aragon-Gawinska; A. Seferian; L. Vanden Brande; A. Daron; A. Ulinici; N. Deconinck; M. Annoussamy; C. Vuillerot; C. Cancès; J. Ropars; M. Chouchane; Z. Balintova; S. Modrzejewska; I. Cuppen; I. Hughes; M. Illingworth; C. Marini-Bettolo; K. White; M. Scoto; T. Gidaro; L. Servais
- P.169 **Nusinersen experience in spinal muscular atrophy type 1: two-year results of 21 patients**
A. Tagiyev; N. Bulut; G. Aydin; I. Alemdaroğlu Gürbüz; N. Eroğlu; O. Yılmaz; G. Haliloğlu; A. Karaduman; H. Topaloğlu
- P.170 **Interim report on the safety and efficacy of longer-term treatment with nusinersen in infantile-onset spinal muscular atrophy: results from the SHINE study**
D. Castro; M. Farrar; R. Finkel; M. Tulinius; K. Krosschell; K. Saito; Y. Zhang; I. Bhan; W. Farwell; S. Reyna
- P.171 **Transforaminal intrathecal delivery of nusinersen using cone-beam computed tomography in children with spinal muscular atrophy: results of technical success and safety**
S. Apkon; J. Weaver; N. Natarajan; D. Shaw; K. Koo; G. Shivaram; E. Monroe
- P.172 **Retrospective study in children with SMA treated by intrathecal Nusinersen**
M. Gomez Garcia de la Banda; B. Doré; A. Bénézit; I. Dabaj; B. Mbieleu; S. Pruvost; A. Felter; T. Thiry; S. Tirolien; P. Dupont; C. Bocassin; A. Essid; L. Durigneux; C. Cancès; L. Servais; I. Haegy; J. Bergounioux; I. Desguerre; R. Carlier; S. Quijano-Roy
- P.173 **Spinraza experience in Dallas**
L. Nelson; M. Valle; D. Forrest; E. Klingman; T. Ramm; A. Farrow-Gillespie; T. Spain; D. Castro; S. Iannaccone
- P.174 **Lumbar catheter placement for nusinersen administration in a SMA2 patient with spinal deformities and previous spinal surgery**
R. Mendonça; A. Silva; O. Velasco; D. Cardeal; U. Conti-reed; E. Zanoteli
- P.175 **Intrathecal nusinersen in spinal muscular atrophy: case series in Argentina**
G. Vazquez; E. Cavassa; M. Pinto; S. Jaimovich; A. Scheitschnaider; F. Dallesandro; S. Duhalde; V. Aguerre; C. Routaboul
- P.176 **Experience using Spinraza to treat adults with spinal muscular atrophy**
J. Day; C. Wolford; C. Macpherson; K. Hagerman; S. Paulose; M. Zeineh; W. Martens; M. McDermott; B. Darras; D. De Vivo; Z. Zolkipli Cunningham; R. Finkel; J. Sampson; T. Duong

- P.177 **AVXS-101 phase 1 gene therapy clinical trial in spinal muscular atrophy type 1: event-free survival and achievement of developmental milestones**
J. Mendell; S. Al-Zaidy; R. Shell; W. Arnold; L. Rodino-Klapac; T. Prior; L. Lowes; L. Alfano; K. Berry; K. Church; J. Kissel; S. Nagendran; J. L'Italien; D. Sproule; C. Wells; A. Burghes; K. Foust; K. Meyer; S. Likhite; B. Kaspar
- P.178 **AVXS-101 phase 1 gene therapy clinical trial in spinal muscular atrophy type 1: improvement in respiratory and bulbar function reduces frequency and duration of hospitalizations compared to natural history**
R. Shell; S. Al-Zaidy; W. Arnold; L. Rodino-Klapac; T. Prior; K. Kotha; G. Paul; L. Lowes; L. Alfano; K. Berry; K. Church; J. Kissel; S. Nagendran; F. Ogrinc; D. Sproule; C. Wells; K. Meyer; S. Likhite; B. Kaspar; J. Mendell
- P.179 **AVXS-101 trial experience: CHOP-INTEND effectively quantifies early, rapid, and sustained improvements that precede subsequent milestone achievement but is not sensitive to continued advances in motor function in infants with SMA type 1**
L. Lowes; L. Alfano; M. Iammarino; N. Miller; M. Menier; J. Cardenas; D. Sproule; S. Nagendran; S. Al-Zaidy; J. Mendell
- P.180 **AVXS-101 phase 1 gene replacement therapy clinical trial in spinal muscular atrophy type 1: patients treated early with the proposed therapeutic dose were able to sit unassisted at a younger age**
L. Alfano; L. Lowes; S. Al-Zaidy; R. Shell; W. Arnold; L. Rodino-Klapac; T. Prior; K. Berry; K. Church; J. Kissel; S. Nagendran; J. L'Italien; D. Sproule; C. Wells; A. Burghes; K. Foust; K. Meyer; S. Likhite; B. Kaspar; J. Mendell
- P.181 **AVXS-101 gene replacement therapy for spinal muscular atrophy type 1: pivotal study (STRIVE) update**
J. Day; D. Feltner; F. Ogrinc; T. Macek; C. Wells; L. Muehring; J. L'Italien; D. Sproule; S. Nagendran; B. Kaspar; J. Mendell
- P.182 **Early diagnosis and speed to effect in infant-onset spinal muscular atrophy**
O. Dabbous; M. Droege; D. Feltner; A. Novack; M. Menier; D. Ryman; D. Sproule
- Mitochondrial diseases (P.183-195)**
Chairpersons: Anders Oldfors and Rahul Phadke
- P.183 **MELAS and reversible cerebral vasoconstriction**
Y. Zhao; C. Yan
- P.184 **Hypercapnic respiratory failure is common presentation in A3243G-related mitochondrial myopathy**
Y. Zhao; C. Yan
- P.185 **MTTL 3243A>G mutation in a patient with a pure mitochondrial myopathy mimicking a distal myopathy, a striking uncommon phenotype**
A. Berardo; R. Reisin; G. Tasca; B. Udd
- P.186 **Muscle histopathology in infantile DNMLL-related mitochondrial epileptic encephalopathy is key for clinical diagnosis**
E. Bertini; D. Verrigni; D. Battaglia; A. Torracco; L. Figa Talamanca; R. Carozzo; D. Diodato; A. D'Amico; L. Papetti; D. Ghezzi; A. Ardisson; C. Lamperti; A. Legati; P. Goffrini
- P.187 **Mitochondrial disorders with polymerase gamma 1 (POLG1) mutations: a study from tertiary referral centre**
N. Gayathri; P. Bindu; P. Govindaraj; C. Shwetha; K. Chetan; S. Deepha; N. Madhu; A. Taly
- P.188 **Novel POLG mutations and variable clinical phenotypes in 5 Chinese patients with mitochondrial diseases**
Z. Wang; X. Zhao; J. Liu; W. Zhang; Y. Yuan
- P.189 **A novel case of MSTO1 gene related congenital muscular dystrophy with cerebellar involvement**
D. Ardichi; I. Zaharieva; A. Sarkozy; R. Phadke; C. Deshpande; I. Bodi; A. Siddiqui; J. U-King-Im; H. Jungbluth; F. Muntoni
- P.190 **Novel variant of GOSR2 gene in a patient presenting with mitochondrial myopathy and epilepsy**
M. Arroyo; B. Wong; C. Fuller; E. Schorry; E. Ulm; C. Tian
- P.191 **Single muscle fiber analysis of extraocular and skeletal muscles in a CPEO patient harboring a pathogenic point mutation in the MT-TN gene**
E. Schlapakow; V. Peeva; M. Jeub; B. Wabbels; G. Zsurka; W. Kunz; C. Kornblum
- P.192 **Severe isolated mitochondrial myopathy in childhood**
M. Loos; H. Ar oz; F. Lubieniecki; A. Taratuto; R. Caraballo; L. Chertkoff; S. Monges
- P.193 **Unexpected genetic diagnosis of mitochondrial disease in three consanguineous Turkish families**
A. Topf; Y. Oktay; S. Balaraju; E. Yilmaz; E. S nmezler; A. Yaramis; S. G ng r; S. Laurie; S. Beltran; I. Gut; H. Lochm ller; S. Hiz; R. Horvath
- P.194 **Deficiency of the iron-sulphur cluster assembly protein ISCU causes impaired biogenesis or stability of respiratory chain complex I, II and IV in muscle**
C. Thomsen; Y. Sunnerhagen; A. Oldfors
- P.195 **A novel multiplex chromogenic immunoassay for evaluating mitochondrial respiratory chain complex I and complex IV defects in diagnostic muscle biopsies**
D. Chambers; A. Kumar; L. Feng; I. Hargreaves; A. Lam; A. Manzur; F. Muntoni; C. Sewry; J. Poulton; R. Phadke

Thursday 4 October 2018

- 07:00- Conference desk opens
- 07:30-09:00 **Symposium 2**
- 09:00-10:30 **Mitochondrial diseases I**; Invited lectures (I.4-5) – Auditorium
Chairpersons: Cornelia Kornblum and John Vissing
- I.4 **Skeletal muscle manifestations in mitochondrial disease**
P. Mishra
- I.5 **Perturbed mitochondrial homeostasis in the pathogenesis of mitochondrial disorders**
M. Zeviani; A. Dogan; M. Sanchez; R. Cerutti; C. Beninca; C. Viscomi
- 10:30-11:00 **Morning refreshments, exhibition and posters**
- 11:00-12:00 **Mitochondrial diseases II**; Invited lectures (I.6-7) - Auditorium
Chairpersons: Mariz Vainzof and Kevin Flanigan
- I.6 **Deoxynucleoside therapy for mitochondrial DNA depletion disorders**
M. Hirano; C. Lopez-Garcia; X. Rosales; K. Engelstad; J. Uddin; C. Dominguez; C. Paradas; R. Marti; C. Garone
- I.7 **Development of genetic therapy for mtDNA diseases**
S. Bacman; C. Pereira; U. Zekonyte; T. Arguello; S. Williams; J. Stewart; D. Jantz; C. Moraes
- 12:00-13:30 **Selected oral presentations II - new insights into cellular functions (O.7-12) – Auditorium**
Chairpersons: Mariz Vainzof and Kevin Flanigan
- O.7 **Mitochondrial dysfunction triggers a pro-survival adaptive response through distinct DNA methylation of nuclear genes**
L. Mayorga; B. Salassa; C. Garcia Samartino; M. Loos; H. Eiroa; P. Romano; M. Roque
- O.8 **SEPN1-related myopathy is a systemic metabolic disease: selenoprotein N maintains endoplasmic reticulum-mitochondrial interaction and regulates mitochondrial bioenergetics**
A. Filipe; E. Varone; S. Arbogast; A. Chernorudskiy; D. Pozzer; R. Villar; C. Dill; S. Dudhal; S. Fumagalli; M. De Simoni; M. Giovarelli; C. De Palma; P. Pinton; C. Giorgi; E. Clementi; S. Missiroli; S. Boncompagni; E. Zito; A. Ferreira
- O.9 **Dysfunctional mitophagy: a potential therapeutic target in inclusion body myositis**
S. Brady; E. Wang; J. Carver; M. Hofer; D. Hilton; D. Hilton-Jones; C. Fratter; J. Poulton
- O.10 **The nuclear-cytoskeleton connection and nuclear positioning during muscle formation**
P. Gimpel; W. Roman; Y. Lee; B. Burke; E. Gomes; B. Cadot
- O.11 **Secretion of toxic exosomes by muscle cells of ALS patients**
I. Le Gall; O. Lucas; S. Roquevière; V. Mariot; J. Dumonceaux; G. Ouandaogo; G. TRANE study; F. Ratti; A. Mejat; A. Durieux; J. Gonzales De Aguilar; C. Martinat; S. Knoblach; C. Raoul; W. Duddy; P. Pradat; S. Duguez
- O.12 **Eccentric contraction causes loss of microtubule lattice organization in mdx skeletal muscle expressing mini- or micro-dystrophin**
D. Nelson; A. Lindsay; D. Lowe; J. Ervasti
- 13:30-14:00 **Lunch, exhibition and posters**
- 14:00-15:30 **Symposium 3**
- 15:30-18:00 **Poster viewing session– Poster area**

Friday 5 October 2018

- 07:30- Conference desk open
- 07:30-08:30 **NMD Editorial Board Meeting – Room: Agrelo**
- 08:30-10:00 **New therapeutic approaches**; Invited lectures (I.8-9) – Auditorium
Chairpersons: Nathalie Goemans and Benedikt Schoser
- I.8 **Emerging therapeutic approaches for facioscapulohumeral muscular dystrophy (FSHD)**
S.Q. Harper
- I.9 **Treatment of Duchenne muscular dystrophy: current efforts, bottlenecks and future prospects.**
E. Mercuri
- 10:00-10:30 **Morning refreshments, exhibition and posters**
- 10:30-12:30 **Selected oral presentations III - New therapeutic approaches and their readout (O.13-20) – Auditorium**
Chairpersons: Nathalie Goemans and Benedikt Schoser
- O.13 **Genome editing for Duchenne muscular dystrophy**
C. Gersbach; C. Nelson; J. Robinson-Hamm; J. Kwon; V. Gough; M. Gemberling

- O.14 **A mutation-independent approach via transcriptional upregulation of a disease modifier gene rescues muscular dystrophy *in vivo***
D. Kemaladewi; P. Bassi; K. Lindsay; S. Erwood; E. Hyatt; K. Place; R. Marks; K. Gawlik; M. Durbeej; E. Ivakine; R. Cohn
- O.15 **Humoral and cell mediated immune response to new dystrophin after morpholino-induced exon skipping therapy in dystrophin-deficient *mdx* mice**
K. Nagaraju; M. Vila; J. Novak; J. Boehler; M. Hogarth; A. Zhang; T. Kinder; D. Mazala; M. Benny Klimek; A. Fiorillo; J. van den Anker; Y. Hathout; E. Hoffman; T. Partridge
- O.16 **Myostatin expression is a reliable and quantifiable biomarker to monitor dose-dependent drug response in muscular dystrophy**
J. Dumonceaux; V. Mariot; C. Le Guiner; I. Barthelemy; C. Hourd e; M. Montus; S. Blot; T. Voit
- O.17 **ASPIRO phase 1/2 gene therapy trial In X-linked myotubular myopathy: preliminary safety and efficacy findings**
N. Kuntz; P. Shieh; B. Smith; C. B nnemann; J. Dowling; M. Lawlor; W. M ller-Felber; M. Noursalehi; S. Rico; L. Servais; S. Prasad
- O.18 **Significantly reduced muscle damage and inflammation observed in Duchenne muscular dystrophy patients following ezutromid treatment**
F. Muntoni; G. Layton; I. Bhattacharya; K. Vandenborne; C. Faelan; A. Heatherington; D. Roblin; J. Tinsley; Imaging DMD Consortium & Phase Out DMD Study Group; K. Davies
- O.19 **First-in-human intrathecal gene transfer study for giant axonal neuropathy: review of safety, immunologic responses and interim analysis of efficacy**
D. Saade; D. Bharucha-Goebel; M. Jain; M. Waite; G. Norato; K. Cheung; A. Foley; A. Soldatos; D. Rybin; T. Lehky; H. Ying; M. Whitehead; R. Calcedo Del Hoyo; S. Jacobson; E. Leibovitch; A. Nath; J. Grieger; R. Samulski; S. Gray; C. B nnemann
- O.20 **Results from ATB200-02: first-in-human, open-label, phase 1/2 study of ATB200 co-administered with AT2221 for Pompe disease**
B. Schoser; D. Bratkovic; B. Byrne; P. Clemens; T. Geberhiwot; O. Goker-Alpan; P. Kishnani; X. Ming; T. Mozaffar; P. Schwenkreis; K. Sivakumar; A. van der Ploeg; J. Wright; F. Johnson; S. Sitaraman; J. Barth; S. Sathe; M. Roberts
- 12:30-14:00 **Lunch, exhibition and posters**
- 14:00-15:30 **Symposium 4**
- 15:30-17:00 **Poster session 3: parallel sessions (P.197-298) – Poster area**
- DMD treatment, animal models (P.197-211)**
Chairpersons: Thomas Crawford and Craig McDonald
- P.197 **Antisense PMO treatment improves muscle recovery from fatigue after a novel *in situ* dynamic muscle contraction protocol in *mdx* mice**
W. Eilers; K. Foster
- P.198 **Stabilised helical peptide-PMO conjugates improve dystrophin exon skipping in the heart of *mdx* mice**
W. Eilers; A. Gadd; H. Foster; K. Foster
- P.199 **Microtrophin delivery shows phenotype improvement in *mdx* mice**
T. Egorova; D. Vlodayets; A. Starikova; A. Polikarpova; A. Smidt; N. Trushkin; E. Luckina; S. Vassilieva; E. Usachev; A. Deikin
- P.200 **Assessment of efficacy of a rAAV9-mini-dystrophin gene therapy candidate (PF-06939926) administered to aged DMD*mdx* rats**
C. Le Guiner; P. Moullier; M. McIntyre; T. Larcher; O. Adjali; A. LaFoux; G. Toumaniantz; J. Owens; X. Xiao; M. Binks; G. LaRosa; R. Samulski
- P.201 **Early insights from 'Of Mice and Measures', a collaborative project to improve models and methods for preclinical research in Duchenne muscular dystrophy, and its first focus on the D2. B10-Dmd^{mdx}/J (D2/*mdx*) and C57BL/10ScSn-Dmd^{mdx}/J (B110/*mdx*) mouse models**
L. Dalle Pазze; H. Gordish-Dressman
- P.202 **Low Kindlin-2 levels in patients and mouse model of Duchenne muscular dystrophy**
J. Konikov-Rozenman; N. Yanay; M. Rabie; Y. Nevo
- P.203 **Exons 6 and 7 skipping test on new murine model of Duchenne muscular dystrophy**
T. Egorova; D. Reshetov; A. Polikarpova; S. Vassilieva; D. Vlodayets; A. Deikin
- P.204 **R-DMDdel52, a novel preclinical rat model of Duchenne muscular dystrophy**
M. Goddard; B. Drayton; F. Pi tri-Rouxel; F. Relaix
- P.205 **Effect of PDE5 inhibition on the post-contractile MRI blood-oxygenation-level-dependent (BOLD) effect in skeletal muscle of dystrophic mice**
S. Forbes; A. Batra; C. Baligand; K. Vandenborne; G. Walter
- P.206 **Effects of sarcoeos (API BIO101) on *in vivo* and *in vitro* models of Duchenne muscular dystrophy**
P. Dilda; M. Serova; S. On; B. Didry-Barca; M. Latil; S. Veillet; R. Lafont

- P.207 **Full-length but not truncated osteoprotegerin binds directly to muscle cells and increases rapidly dystrophic muscle force**
A. Boulanger Piette; L. Marcadet; D. Hamoudi; S. Bossé; A. Argaw; J. Frenette
- P.208 **Therapeutic benefits of intravenous cardiac progenitor cell and exosome-based therapies in a mouse model of Duchenne muscular dystrophy**
R. Rogers; M. Fournier; L. Sanchez; M. Aminzadeh; E. Marban
- P.209 **Dystrophin expression in the rat urinary bladder**
 J. Lionarons; G. Hoogland; R. Hendriksen; C. Faber; D. Hellebrekers; G. Van Koeveeringe; S. Schipper; J. Vles
- P.210 **Dystrophin expression in the rat intestine**
 J. Lionarons; R. Slegers; J. Hendriksen; C. Faber; G. Hoogland; J. Vles
- P.211 **Myostatin inhibition and growth factor treatment of pre- and post-disease onset mdx mice does not improve the phenotype coherently**
 T. Nielsen; C. Hjortkaer; T. Pinos; J. Vissing; T. Krag
- Duchenne muscular dystrophy - genetics (P.212-224)**
Chairpersons: Ieke Ginjaar and Alessandra Ferlini
- P.212 **Mutational spectrum of the DMD gene in pediatric patients from an Argentinian referral center**
 M. Foncuberta; F. Lubieniecki; L. Gravina; L. González Quereda; P. Gallano; L. Chertkoff; S. Monges
- P.213 **The DMD Italian network: reporting 2127 genetic diagnoses of referred dystrophinopathies, reflections and impact on care and personalized therapies**
 M. Neri; A. Mauro; F. Gualandi; C. Bruno; F. Santorelli; S. Tedeschi; A. D'Amico; E. Giardina; M. Castori; M. Cau; C. Scuderi; V. Sansone; S. Messina; E. Pegoraro; L. Politano; E. Bertini; G. Comi; V. Nigro; E. Mercuri; A. Ferlini
- P.214 **Genetic profile of Chilean patients with Duchenne muscle dystrophy**
S. Lara; V. Saez; P. Santander; G. Fariña; M. Troncoso; G. Legaza
- P.215 **Genetic modifiers of Duchenne muscular dystrophy**
L. Schottlaender; J. Domingos; L. D'Argenzio; I. Zaharieva; P. Ala; A. Manzur; J. Bourke; J. Morgan; F. Muntoni
- P.216 **Genetic carrier screening for Duchenne muscular dystrophy: the outcome of over forty years of genetic counselling on disease incidence in New South Wales, Australia**
H. Sampaio; D. Kariyawasam; M. Buckley; D. Mowat; J. Robinson; P. Taylor; K. Jones; M. Farrar
- P.217 **Duchenne and Becker muscular dystrophy carrier mothers: characterization of skeletal and cardiac muscle compared to healthy controls**
S. Al Zaidy; E. Camino; N. Miller; K. Lehman; L. Lowes; L. Alfano; M. Iammarino; J. Alexander; L. Cripe; K. Hor; M. Mah; J. Mendell
- P.218 **Heart and skeletal muscle affection in female carriers of a dystrophin gene mutation**
T. Solheim; F. Fornander; R. Møgelvang; N. Poulsen; A. Andersen; A. Eisum; M. Duno; H. Bundgaard; J. Vissing
- P.219 **Duchenne and Becker muscular dystrophy carriers: emerging evidence for a clinically important cardiomyopathy**
 M. Mah; L. Cripe; S. Al-Zaidy; E. Camino; M. Slawinski; J. Jackson; J. Mendell; K. Hor
- P.220 **A neuropsychological and neuroimaging study of female carriers of DMD mutations**
 S. Passos; P. Tavares; T. Rezende; L. Souza; T. Rosa; C. Iwabe-Marchese; A. Nucci; M. França Jr
- P.221 **Unsolicted findings in the DMD gene; what are the implications?**
H. van Duyvenvoorde; D. van Heusden; M. Hoffer; H. Ginjaar
- P.222 **Influence of the intronic breakpoint of the DYS 45-55 exon deletion on the clinical phenotype**
 J. Poyatos; C. Gomis; N. Muelas; P. Marti; J. Vilchez
- P.223 **Small mutation detection in the DMD gene by whole exome sequencing of Argentine dystrophinopathy children**
 L. Luce; M. Carcione; C. Mazzanti; L. Mesa; A. Dubrovsky; F. Giliberto
- P.224 **Whole-genome sequencing reveals a complex intra-chromosomal rearrangement disrupting the dystrophin gene due to an intronic 0.5 Mb-insertion in a boy suffering from Duchenne muscular dystrophy**
A. Ille; W. Schmidt; M. Gosk-Tomek; S. Weiss; M. Freilingner; R. Bittner; G. Bernert
- Congenital myopathies: nemaline and titinopathies (P.225-244)**
Chairpersons: Ana Lia Taratuto and Ana Ferreira
- P.225 **Clinical, genetic and neuropathological heterogeneity in a pediatric cohort with nemaline myopathy**
 J. Martins; J. Oliveira; R. Taipa; C. Garrido; M. Melo Pires; M. Santos
- P.226 **Functional nebulin studies for assessment of pathogenicity**
J. Lehtonen; S. Sofieva; J. Laitila; C. Wallgren-Pettersson; M. Grönholm; K. Pelin; V. Lehtokari
- P.227 **Unraveling muscle slowness in NEM6 myopathy: a key role for the skeletal muscle thin filament**
J. de Winter; J. Molenaar; M. van Willigenburg; S. Conijn; S. Lassche; T. Irving; K. Campbell; B. Van Engelen; N. Voermans; C. Ottenheijm
- P.228 **Severe nemaline myopathy manifesting as 'Amish phenotype' related to homozygous mutation in TNNT1**
 A. D'Amico; F. Fattori; C. Fiorillo; M. Verardo; M. Catteruccia; E. Bellacchio; M. Moggio; C. Bruno; E. Bertini
- P.229 **Genetics and modeling of TNNT1 genetic variants in nemaline myopathy**
C. Konersman; A. Aykanat; E. Troiano; A. Beggs
- P.230 **Clinical, genetic and pathological characterization of a wide cohort of UK patients with NEB gene related nemaline myopathy**
 D. Steel; A. Sarkozy; R. Mein; R. Phadke; C. Sewry; F. Muntoni

- P.231 **The clinical, genetic, and pathological findings in a Chinese cohort of patients with hereditary nemaline myopathy**
Z. Wang; Z. Hu; W. Zhang; H. Lv; M. Yu; Y. Yuan
- P.232 **Core and rod myopathy due to a novel mutation in BTB domain of *KBTBD13* gene presenting as LGMD**
M. Garibaldi; F. Fattori; C. Bortolotti; G. Brochier; C. Labasse; M. Verardo; E. Bertini; E. Pennisi; C. Paradas; N. Romero; G. Antonini
- P.233 **Comparison of new mouse models with different variants in the nebulin gene**
J. Laitila; E. McNamara; H. Goulee; C. Wingate; M. Lawlor; J. Ross; J. Ochala; L. Griffiths; G. Ravenscroft; C. Sewry; N. Laing; C. Wallgren-Pettersson; K. Pelin; K. Nowak
- P.234 **Proteomic profiling in nemaline myopathy to identify disease subclass biomarkers**
E. Siebers; J. Tinklenberg; H. Meng; S. Ayres; M. Vanden Avond; R. Slick; K. Nowake; H. Granzier; E. Hardeman; F. Montanaro; M. Lawlor
- P.235 **Recessive congenital fiber type disproportion caused by *TPM3* mutation**
C. Moreno; E. Estephan; O. Abath Neto; C. Camelo; A. Silva; U. Reed; C. Bönnemann; E. Zanoteli
- P.236 **Congenital fiber type disproportion with mutations in tropomyosin 3 (*TPM3*) gene presenting as respiratory failure**
D. Namgung; J. Lee; W. Kim; Y. Choi
- P.237 **Congenital fatal cap-rod myopathy due to a de novo autosomal dominant pathogenic *ACTA1* variant**
R. Phadke; B. Herron; D. Hurrell; S. Craig; B. Kelly; A. Sarkozy; C. Sewry; F. Muntoni; V. McConnell
- P.238 **The international database of titin gene variations and their phenotypes**
P. Hackman; M. Savarese; C. Bönnemann; A. Ferreiro; A. Beggs; J. Dawson; R. Thompson; T. Evangelista; H. Lochmüller; J. Nikodinovic Glumac; H. Jungbluth; S. Foye; B. Udd
- P.239 **Congenital titinopathy: severe and atypical presentations**
E. Oates; K. Jones; S. Coppens; N. Deconinck; G. Ravenscroft; H. Luk; M. Bakshi; J. Pinner; N. Foulds; M. Illingworth; N. Thomas; S. Ellard; I. Mazanti; S. Cooper; F. Muntoni; M. Davis; N. Laing
- P.240 **Distal upper limb onset myopathy in the first Chilean case reported with titinopathy**
L. González-Quereda; M. Fuentealba; J. Díaz; A. Tringulao; P. Gallano; J. Bevilacqua
- P.241 **Loss of sarcomeric scaffolding as a common baseline histopathologic lesion in titin-related myopathies**
E. Malfatti; R. Avila-Polo; X. Lornage; I. Nelson; J. Nectoux; J. Bohm; C. Hedberg-Oldfors; B. Eymard; S. Monges; F. Lubieniecki; G. Brochier; A. Madelaine; C. Labasse; A. Taratuto; B. Udd; F. Leturcq; G. Bonne; A. Oldfors; J. Laporte; N. Romero
- P.242 **Neonatal presentations of recessive *TTN*-related myopathy: an emerging distinct clinical phenotype**
S. Neuhaus; S. Donkervoort; M. Leach; S. Iannaccone; C. Konersman; D. Saade; A. Foley; C. Bönnemann
- P.243 **Taking on the titin: semitendinosus muscle involvement as a diagnostic marker of early onset recessive *TTN*-related myopathy**
S. Neuhaus; L. Hayes; D. Saade; S. Donkervoort; P. Mohassel; J. Dastgir; D. Bharucha-Goebel; M. Leach; C. Vuillerot; S. Iannaccone; C. Grossmann; A. Beggs; A. Foley; C. Bönnemann
- P.244 **A ddPCR method for the analysis of copy number variation in the segmental duplication regions of the sarcomeric giants nebulin and titin**
L. Sagath; V. Lehtokari; C. Wallgren-Pettersson; K. Pelin; K. Kiiski
- Myofibrillar and distal myopathies (P.245-254)**
Chairpersons: Montse Olivé and Hans-Hilmar Goebel
- P.245 **Assessment of new lectin-based protocols for the diagnosis of GNE myopathies**
Y. Parkhurst; R. Barresi
- P.246 **GNE myopathy in Chinese population: hotspot and novel mutations**
Y. Chen; J. Xi; W. Zhu; J. Lin; S. Luo; D. Yue; S. Cai; C. Sun; C. Zhao; S. Mitsuhashi; I. Nishino; M. Xu; J. Lu
- P.247 **Clinical and genetic profiles of GNE myopathy in Korean patients**
J. Shin; Y. Park; J. Lee; D. Kim
- P.248 **A novel exon 1 deletion mutation in the *GNE* gene in a GNE myopathy patient**
J. Miao; X. Liu; F. Su; X. Wei; Z. Kang; Y. Gao; X. Yu
- P.249 **A novel mutation in *MYH7* giving rise to different phenotypes in a mother and her daughter**
K. Orstavik; V. Almaas; M. Rasmussen; C. Jonsrud; S. Jensen; S. Loseth; T. Leren
- P.250 **The clinical, myopathological characteristics of a Chinese cohort of myofibrillar myopathy: a retrospective study**
Y. Luo; Q. Li; H. Duan; F. Bi; H. Yang
- P.251 **Family with a new mutation in the *DES* gene of autosomal recessive transmission**
P. Marti; N. Muelas; I. Azorin; J. Vilchez
- P.252 **A case report: a heterozygous deletion (2791_2805 del) in exon 18 of the *FLNC* gene causing filamin C-related myofibrillar myopathies**
J. Miao; X. Wei; Z. Kang; Y. Gao; X. Yu
- P.253 **Severe intestinal pseudo-obstruction in a R405W desmin knock-in model: a new phenotype leads to light smooth muscle involvement in myofibrillar myopathies**
E. Cabet; D. Delacour; C. Hakibilen; F. Delort; S. Pichon; P. Vicart; A. Ferreiro; A. Lilienbaum
- P.254 ***PYROXD1* mutations cause recessive adult-onset slowly progressive LGMD**
J. Palmio; M. Sainio; S. Välipakka; M. Jokela; M. Auranen; A. Paetau; S. Huovinen; H. Lapatto; E. Ylikallio; B. Udd; H. Tyynismaa

SMA therapies II and biomarkers (P.255-266)*Chairpersons: Susan Iannaccone and Richard Finkel*

- P.255 **SUNFISH Part 1: RG7916 treatment results in a sustained increase of SMN protein levels and the first clinical efficacy results in patients with type 2 or 3 SMA**
E. Mercuri; G. Baranello; J. Kirschner; L. Servais; N. Goemans; M. Carmela Pera; J. Buchbjerg; G. Armstrong; H. Kletzl; M. Gerber; C. Czech; Y. Cleary; K. Gorni; O. Khwaja
- P.256 **A study of RG7916 in infants with pre-symptomatic spinal muscular atrophy**
E. Bertini; J. Day; M. Al Muhaizea; H. Xiong; L. Servais; A. Prufer; J. Buchbjerg; G. Armstrong; K. Gorni; O. Khwaja
- P.257 **JEWELFISH: RG7916 increases SMN protein in patients with SMA that have previously received therapies targeting SMN2 splicing**
C. Chiriboga; E. Mercuri; D. Fischer; D. Kraus; N. Thompson; G. Armstrong; H. Kletzl; M. Gerber; Y. Cleary; T. Bergauer; K. Gorni; O. Khwaja
- P.258 **FIREFISH Part 1: early clinical results following a significant increase of SMN protein in SMA type 1 babies treated with RG7916**
G. Baranello; L. Servais; J. Day; N. Deconinck; E. Mercuri; A. Klein; B. Darras; R. Masson; H. Kletzl; Y. Cleary; G. Armstrong; T. Seabrook; C. Czech; M. Gerber; K. Gelblin; K. Gorni; O. Khwaja
- P.259 **SMN protein levels before and after treatment with RG7916 in type 1, 2 and 3 SMA patients compared to healthy subjects**
H. Kletzl; C. Czech; Y. Cleary; S. Sturm; A. Günther; G. Baranello; E. Mercuri; L. Servais; J. Day; N. Deconinck; A. Klein; B. Darras; R. Masson; J. Kirschner; N. Goemans; M. Pera; C. Chiriboga; D. Fischer; K. Gorni; O. Khwaja
- P.260 **A long-term, open-label, follow-up study of olesoxime in patients with type 2 or non-ambulatory type 3 SMA who participated in a placebo-controlled phase 2 trial**
F. Muntoni; S. Fuerst-Recktenwald; E. Bertini; E. Mercuri; J. Kirschner; C. Reid; A. Lusakowska; G. Comi; J. Cuisset; J. Ives; W. van der Pol; C. Vuillerot; K. Gorni; P. Fontoura
- P.261 **Safety and efficacy of the oral splice modulator branaplam in type 1 spinal muscular atrophy**
N. Deconinck; A. Born; G. Baranello; E. Bertini; H. Jullien de Pommerol; B. Gomez Mancilla; N. Goemans; R. Pingili; J. Praestgaard; R. Roubenoff; U. Schara
- P.262 **Phosphorylated neurofilament heavy chain (pNF-H) levels in infants and children with SMA: evaluation of pNF-H as a potential biomarker of SMA disease activity**
T. Crawford; C. Sumner; R. Finkel; D. De Vivo; M. Oskoui; E. Tizzano; G. Zhao; M. Petrillo; C. Stebbins; W. Farwell
- P.263 **Neurofilament light chain as a potential biomarker in spinal muscular atrophy**
H. Jullien de Pommerol; A. Kieloch; D. Leppert; T. Peters; D. Theil; M. Valentin; E. Voltz
- P.264 **Circulating microRNAs as biomarkers in Spinraza treated SMA patients**
I. Zaharieva; M. Lauffer; E. Bollen; K. Aragon-Gawinska; L. Servais; M. Scoto; H. Zhou; F. Muntoni
- P.265 **Patients with spinal muscular atrophy without cardiac disease show elevated cardiac troponin T**
A. Ille; A. van Egmond-Fröhlich; S. Weiss; M. Gosk-Tomek; M. Foedinger; S. Peithner; G. Bernert
- P.266 **Neurofilament as a potential biomarker for spinal muscular atrophy: rationale based on animal and human studies**
T. Crawford; C. Sumner; M. Petrillo; C. Stebbins; W. Farwell

Metabolic myopathies I (P.267-277)*Chairpersons: Ros Quinlivan and Corrado Angelini*

- P.267 **Local experience of hyperCKaemia in a multidisciplinary neuromuscular clinic**
H. Sampaio; M. Farrar; A. Al Safar
- P.268 **Reduced skeletal muscle fat oxidation during exercise in an adult with LPINI-deficiency**
D. Raaschou-Pedersen; K. Madsen; M. Stemmerik; A. Eisum; J. Vissing
- P.269 **Neutral lipid storage disease with myopathy: clinical and genetic spectrum in a large cohort of Chinese patients**
Y. Yuan; C. Yan; C. Zhao; J. Hu; C. Zhang
- P.270 **Lipid storage disorder-Proteomic analysis of skeletal muscle mitochondria**
N. Gayathri; B. Debashree; K. Manish Kumar; T. Keshava Prasad; N. Archana; C. Rita; A. Nalini; P. Bindu; M. Srinivas Bharath
- P.271 **MicroRNA dysregulation and signalling in lipid storage myopathies**
C. Angelini; R. Marozzo; V. Pegoraro
- P.272 **No effect of triheptanoin on exercise performance in patients with McArdle disease - a double blind placebo-controlled crossover study**
K. Madsen; P. Laforêt; A. Buch; M. Stemmerik; S. Hatem; D. Raaschou-Pedersen; N. Poulsen; M. Atencio; C. Ottolenghi; C. Jardel; R. Quinlivan; F. Mochel; J. Vissing
- P.273 **Delineating the phenotypic spectrum of PGKI-associated phosphoglycerate kinase deficiency: the French experience**
A. Echaniz-Laguna; Y. Nadjar; A. Béhin; V. Biancalana; M. Piraud; P. Laforêt
- P.274 **Heterozygous mutation in ISCU associated with recurrent rhabdomyolysis**
C. Gitiaux; S. Gobin-Limballe; I. Desguerre; C. Barnerias; P. De Lonlay; F. Authier
- P.275 **A case of late-onset multiple acyl-coenzyme A dehydrogenase deficiency in a young female of Turkish descent**
D. Pehl; A. von Renesse; L. Harms; H. Goebel; M. Schuelke; W. Stenzel
- P.276 **Electron transfer flavoprotein-ubiquinone oxidoreductase defect and FAD homeostasis in riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency**
J. Xu; D. Li; J. Lv; X. Xu; B. Wen; P. Lin; F. Liu; K. Ji; J. Shan; W. Li; Y. Zhao; J. Pok; C. Yan

- P.277 **A curable myopathy manifesting as exercise intolerance and respiratory failure**
A. Silva; R. Mendonça; D. Soares; D. Callegaro; V. Caldas; M. Carvalho; E. Zanoteli
- Registries and care of neuromuscular disorders (P.278-298)**
Chairpersons: Helen Roper and Tahseen Mozaffar
- P.278 **How can we ensure children with neuromuscular conditions achieve personally meaningful futures?**
L. McAdam; D. Greenspoon; K. Bell; K. English; S. Keenan; A. McPherson
- P.279 **A four-year review of a Canadian pediatric neuromuscular clinic**
K. Amburgey; H. Gonorazky; J. Dowling
- P.280 **Registry of neuromuscular genetic disorders in Russia**
S. Artemyeva; D. Vlodayets; A. Monakhova; E. Melnik; I. Shulyakova; O. Shidlovskaya; E. Belousova; D. Reshetov
- P.281 **Participation and its determinants in children with neuromuscular disease**
C. de Montferrand; M. Morard; C. Pons-Becmeur; J. Ropars; P. Rippert; C. Vuillerot
- P.282 **Drop-out in longitudinal natural history studies in neuromuscular diseases: rates and main rationale**
M. Annoussamy; D. Ho; A. Seferian; T. Gidaro; k. Aragon; L. Vanden Brande; L. Servais
- P.283 **The development of a Brazilian Portuguese version of the activity limitations scale (ACTIVLIM)**
M. Voos; D. Almeida; A. Silva; P. Santos; U. Reed; E. Zanoteli
- P.284 **Quantifying activity changes of neuromuscular patients using the ACTIVLIM questionnaire: a 5-years longitudinal study**
 C. Bleyenheuft; N. Goemans; S. Wanyama; P. Van Damme; J. De Bleecker; R. Van Coster; P. De Jonghe; D. Beysen; P. Van den Bergh; S. Paquay; L. Servais; A. Maertens de Noordhout; J. Haan; L. De Meirleir; G. Remiche; N. Deconinck; BNMDR study group; C. Arnould
- P.285 **Validation of the Brazilian Portuguese version of the motor function measure - short form (MFM-20) for neuromuscular diseases in children from two to seven years old**
 A. Pedrosa; V. Van der Linden; C. Iwabe-Marchese; M. Voos; E. Zanoteli; J. Teixeira; E. Araújo; U. Reed
- P.286 **A new minimally invasive fusionless technique that avoid vertebral arthrodesis for neuromuscular scoliosis**
 M. Gaume; R. Sauvagnac; S. Quijano-Roy; V. Azzi-Sallameh; I. Dabaj; A. Bénézit; B. Mbieleu; D. Verollet; A. Essid; I. Haegy; J. Bergounioux; I. Desguerre; L. Miladi; C. Glorion
- P.287 **Mechanically assisted cough - how to keep it simple**
S. Zacher; A. van Egmond-Fröhlich; S. Weiss; B. Guenther
- P.288 **Diagnosis, management and outcome of severe congenital onset neuromuscular disorders in a series of 50 infants**
M. Sa; R. Biancheri; N. McCrea; R. Phadke; M. Pitt; A. Manzur; P. Munot
- P.289 **Patients experience of diagnosis of a genetic muscle disorder**
M. Rodrigues; R. Roxburgh; G. O'Grady; G. Poke; A. Theadom
- P.290 **Audit of impact of the quality of the pre-test information on the outcome of muscle biopsy assessment**
K. Urankar; A. Kanagasabai; S. Brady
- P.291 **Clinicopathological study for ultrasound-guided biopsy cases using linear probe**
T. Kurashige; T. Kanbara; N. Sumi; S. Tasaka; T. Sugiura; H. Maruyama; T. Torii
- P.292 **Skeletal muscle NMR image automatic segmentation using convolutional neural network**
 E. Sneszhko; P. Baudin; P. Carlier
- P.293 **A novel, ultrafast and robust NMR imaging approach to evaluate disease activity and chronic degenerative changes in skeletal muscle using an optimal fingerprinting radial sequence**
 B. Marty; P. Carlier
- P.294 **Muscle biopsy in the study of muscle disease in pediatric population**
 R. Escobar; R. Gejman; C. Jaque; M. Beytia; D. Avila; J. Casar; O. Trujillo; R. Fadic
- P.295 **HyperCKemia asymptomatic or oligosymptomatic in an Argentinian neuropaediatric cohort**
 M. Garcia Erro; E. Cavassa; J. Muntadas; M. Pauni; G. Vazquez
- P.296 **ICD code refinement for Duchenne/Becker muscular dystrophy**
 A. Kennedy; R. Valdez; C. Westfield; J. Bolen; K. Kinnett; D. Perez; P. Furlong
- P.297 **Treatment responsive outcome measures in mouse models of neuromuscular disease**
A. Mullen; K. Uaesoontrachoon; S. Srinivassane; M. Moraca; W. Ross; A. MacKinnon; C. Bell; E. Gillis; J. Rowsell; M. Malbasic; M. Barkhouse; J. Warford; D. Shala; E. Hoffman; K. Nagaraju
- P.298 **Development of a microRNA-155 inhibitor as a therapeutic for neuroinflammatory and neurodegenerative diseases**
D. Escobar; M. Hermreck; H. Semus; D. Hood; A. Jackson

17:00-18:30

Poster session 4: parallel sessions (P.299-387) – Poster area**Duchenne muscular dystrophy - physiotherapy (P.299-322)***Chairpersons: Linda Lowes and Imelda de Groot*

- P.299 **Minimal detectable change in the North Star ambulatory assessment (NSAA) in Duchenne muscular dystrophy (DMD)**
F. Muntoni; A. Manzur; A. Mayhew; NorthStar Clinical Network; J. Signorovitch; G. Sajeev; Z. Yao; I. Dieye; M. Jenkins; S. Ward
- P.300 **Prognostic factors for changes in 4-stair climb ability in patients with Duchenne muscular dystrophy**
N. Goemans; B. Wong; J. Signorovitch; G. Sajeev; M. Jenkins; I. Dieye; Z. Yao; I. Hossain; S. Ward

- P.301 **Home based movement monitoring allows pivotal trials in DMD with ten times less patients than classical outcomes measures**
L. Servais; E. Gasnier; M. Grelet; T. Gidaro; A. Seferian; D. Vissieres
- P.302 **Genetic association study of articular range of motion in the CINRG Duchenne natural history study**
T. Duong; L. Bello; E. Henricson; E. Hoffman; C. McDonald; H. Gordish-Dressman; CINRG Investigators
- P.303 **Development of a conversion method to enable an accurate PUL v.2 score from PUL v.1.2 data in a cohort of Duchenne muscular dystrophy patients**
 V. Selby; V. Ricotti; A. Mayhew; D. Ridout; J. Pitchforth; E. Niks; L. Servais; I. de Groot; V. Straub; E. Mercuri; F. Muntoni
- P.304 **Relationship of lower extremity strength and range of motion on timed function tests in Duchenne muscular dystrophy**
T. Duong; H. Gordish-Dressman; M. Pavlvolgyi; M. Fensterwald; C. McDonald; E. Henricson; CINRG Investigators
- P.305 **Disease progression in arm versus leg muscles in Duchenne muscular dystrophy**
 H. Arora; R. Willcocks; S. Forbes; W. Triplett; W. Rooney; D. Wang; M. Daniels; E. Finanger; G. Tennekoon; J. Brandsema; H. Sweeney; G. Walter; K. Vandeborne
- P.306 **Utility of the Bayley-III, North Star Ambulatory Assessment, and 100-meter timed test in quantifying gross motor delay in very young boys with Duchenne muscular dystrophy**
 N. Miller; L. Alfano; M. Iammarino; M. Dugan; M. Moore-Clingenpeel; S. Al-Zaidy; C. Tsao; M. Waldrop; K. Flanigan; J. Mendell; L. Lowes
- P.307 **Relationships between hand strength and function in non-ambulant patients with Duchenne muscular dystrophy or spinal muscular atrophy**
 V. Decostre; M. Anoussamy; M. De Antonio; A. Canal; L. Servais; JY. Hogrel
- P.308 **Determination of the minimal clinically important difference (MCID) for clinical trial outcome measures in Duchenne muscular dystrophy**
J. Pitchforth; J. Domingos; M. Iodice; A. Mayhew; F. Muntoni
- P.309 **The effects of trunk and lower extremity flexibility on lumbar lordosis in children with Duchenne muscular dystrophy**
 L. Akkurt; G. Aydin; I. Alemdaroglu Gürbüz; A. Karaduman; H. Topaloğlu; Ö. Yilmaz
- P.310 **The comparison of children with Duchenne muscular dystrophy and healthy peers in terms of pulmonary and upper extremity functions**
N. Bulut; G. Aydin; i. Alemdaroglu Gürbüz; A. Karaduman; H. Topaloğlu; O. Yilmaz
- P.311 **The effect of kinesiology taping on balance in Duchenne muscular dystrophy**
G. Aydin; I. Alemdaroglu Gürbüz; N. Bulut; A. Karaduman; H. Topaloğlu; O. Yilmaz
- P.312 **Factors influencing spontaneous maximal stride speed in individual Duchenne muscular dystrophy boys**
C. Lilien; M. Grelet; E. Gasnier; T. Gidaro; A. Seferian; A. Rigaud; D. Vissière; L. Servais
- P.313 **Use of a powered arm support devices for upper limb function in non-ambulatory men with Duchenne muscular dystrophy**
R. Bendixen; A. Kelleher; M. Feltman; N. Little
- P.314 **Kinematic/behavioural fingerprints in Duchenne muscular dystrophy and their clinical applications**
V. Ricotti; S. Haar; V. Selby; T. Voit; A. Faisal
- P.315 **Stride to height ratio as a new ambulatory outcome measure in Duchenne muscular dystrophy**
 E. Henricson; R. Abresch; A. Bagley; E. Goude; C. Owens; L. Williams; C. McDonald
- P.316 **Utility of ACTIVE workspace volume as a clinically meaningful measure of functional capacity in individuals with neuromuscular disease**
M. Iammarino; L. Alfano; N. Miller; M. Dugan; M. Moore-Clingenpeel; S. Al-Zaidy; C. Tsao; M. Waldrop; K. Flanigan; L. Rodino-Klapac; J. Mendell; L. Lowes
- P.317 **Clinically meaningful change on the 100-meter timed test in neuromuscular diseases**
 L. Alfano; N. Miller; M. Iammarino; K. Berry; M. Moore-Clingenpeel; M. Dugan; S. Al-Zaidy; C. Tsao; L. Rodino-Klapac; M. Waldrop; K. Flanigan; J. Mendell; L. Lowes
- P.318 **Insights from a multisite study utilizing dedicated technology to assess electrical impedance myography as an outcome measure for Duchenne muscular dystrophy**
 C. Zaidman; K. Kapur; B. Darras; B. Wong; M. Yang; M. Leitner; L. Dalle Pазze; M. Buck; L. Freedman; S. Rutkove
- P.319 **Can we use elastic bandage in children with Duchenne muscular dystrophy by therapy taping methods? Pilot study**
C. Iwabe-Marchese; N. Morini Jr; c. Sanches; T. Rosa
- P.320 **Trunk movement and muscle activity in children with Duchenne muscular dystrophy when performing daily activities**
 L. Peeters; I. Kingma; J. van Dieën; I. de Groot
- P.321 **Bringing the spoon to the mouth or the mouth to the spoon? The analysis of compensatory movements of simulated feeding in Duchenne muscular dystrophy: a case-control study**
 M. Artilhiero; E. Oliveira; N. Carvas Junior; F. Favero; F. Caromano; C. Sá; M. Voos
- P.322 **Development of dynamic trunk and head supportive devices for children with neuromuscular disorders**
 L. Peeters; M. Mahmood; S. Verros; H. Koopman; I. de Groot; Symbionics working group
- Congenital muscular dystrophies (P.323-334)**
Chairpersons: Haluk Topaloğlu and Soledad Monges
- P.323 **Review of the natural history of mental development in Fukuyama congenital muscular dystrophy patients, based on a written questionnaire from their families**
M. Shichiji; K. Ishigaki; T. Sato; A. Yamashita; S. Nagata

- P.324 **Clinical characteristics and long-term course of selenoprotein N1 related myopathy in a large multi-centric cohort**
A. Silwal; M. Scoto; D. Ridout; A. Schmidt; A. Laverty; M. Henriques; L. D'Argenzio; M. Main; R. Mein; A. Sarkozy; A. Manzur; F. Al-Ghamdi; C. Genetti; F. Abel; G. Haliloglu; D. Ardicli; H. Topaloğlu; A. Beggs; F. Muntoni
- P.325 **Severe loss of semimembranosus muscle bulk is an early phenomenon in SEPNI-related muscle disorders: toward early recognition of early-onset muscle disorders by imaging**
F. Munell; D. Gomez-Andres; A. Sanchez-Montañez; L. Costa Comellas; S. Ferrer-Aparicio; P. Romero; S. Quijano-Roy; M. Olivé
- P.326 **Minimal clinically important difference for the Motor Function Measure in patients with congenital muscular dystrophy and congenital myopathy**
L. Le Goff; M. Fink; G. Norato; P. Rippert; K. Meilleur; R. Foley; M. Jain; M. Waite; S. Donkervoort; C. Bönnemann; C. Vuillerot
- P.327 **A model for dominant-mutated collagen VI-related disorder and allele-specific gene silencing therapy**
S. Noguchi; M. Ogawa; I. Nishino
- P.328 **Pneumothorax in Ullrich congenital muscular dystrophy**
Y. Arahata; A. Ishiyama; M. Ogawa; S. Noguchi; R. Tanaka; E. Takeshita; Y. Shimizu-Motohashi; H. Komaki; Y. Saito; I. Nishino
- P.329 **CALLISTO: a phase I open-label, sequential group, cohort study of pharmacokinetics and safety of omigapil in LAMA2 and COL6-related dystrophy patients**
A. Foley; M. Leach; G. Averion; Y. Hu; P. Yun; S. Neuhaus; D. Saade; C. Arevalo; M. Fink; J. DeCoster; C. Mendoza; O. Mayer; R. Hausmann; D. Petraki; K. Cheung; C. Bönnemann
- P.330 **Long-term motor function in collagen VI-related myopathies is associated with the maximal motor ability achieved: a classification proposal**
D. Natera - de Benito; R. Foley; J. Diaz-Manera; M. Fink; A. Mebrahtu; J. Minal; S. Donkervoort; Y. Hu; D. Cuadras; L. Carrera-García; M. Alarcón; J. Milisenda; R. Dominguez; M. Olive; C. Jou; J. Colomer; C. Jimenez-Mallebrera; C. Bönnemann; A. Nascimento
- P.331 **Longitudinal changes of motor outcome measures in individuals with COL6-RDs and LAMA2-RD**
M. Jain; K. Meilleur; G. Norato; M. Waite; M. Leach; S. Donkervoort; A. Foley; C. Bönnemann
- P.332 **Dynamic breathing MRI: A promising biomarker of diaphragmatic function in COL6-related dystrophy patients and LAMA2-related dystrophy patients**
P. Yun; G. Norato; N. Hsieh; R. Zhu; J. Dastgir; M. Leach; S. Donkervoort; J. Yao; A. Arai; C. Bönnemann; A. Foley
- P.333 **Laminin $\alpha 1$ chain overexpression has potentially broad therapeutic spectrum for LAMA2-CMD**
K. Gawlik; V. Harandi; R. Cheong; A. Petersen; M. Durbej
- P.334 **Recessive loss-of-function mutations in ITGA7 cause cardiac arrhythmia with or without structural cardiomyopathy and respiratory muscle weakness**
E. Bugiardini; R. Phadke; R. Maas; A. Pittman; B. Kusters; J. Morrow; M. Parton; A. Nunes; M. Akhtar; P. Syrris; L. Lopes; T. Fotelonga; H. Houlden; P. Elliott; M. Hanna; J. Raaphorst; D. Burkin; E. Matthews
- CMT and neurogenic disease (P.335-347)**
Chairpersons: Michael Shy and Peter Van den Bergh
- P.335 **Guillain-Barré syndrome subtype diagnosis: a prospective multicentric European study**
P. Van den Bergh; F. Piéret; J. Woodard; S. Attarian; A. Grapperon; G. Nicolas; M. Brisset; J. Cassereau; Y. Rajabally; V. Van Parijs; D. Verougstraete; P. Jacquerye; J. Raymackers; C. Redant; C. Michel; E. Delmont
- P.336 **Imaging findings in patients with Guillain-Barré syndrome**
P. Massaro Sanchez; A. Savransky; C. Rugilo; S. Monges
- P.337 **Giant axonal neuropathy presenting as CMT2: results from the NIH natural history study**
D. Saade; D. Bharucha-Goebel; G. Norato; A. Foley; M. Waite; M. Jain; S. Debs; R. Vasavada; C. Nichols; R. Kaur; S. Donkervoort; S. Neuhaus; Y. Hu; T. Lehky; S. Gray; M. Fink
- P.338 **X-linked Charcot-Marie-Tooth case with a novel variant in GJB1**
J. Lee; J. Shin
- P.339 **Preliminary phase 2 results for ACE-083, local muscle therapeutic, in patients with CMT1 and CMTX**
M. Shy; D. Herrmann; F. Thomas; C. Quinn; J. Statland; D. Walk; N. Johnson; S. Subramony; C. Karam; T. Mozaffar; S. D'Eon; B. Miller; C. Glasser; M. Sherman; K. Attie
- P.340 **Distal arthrogryposis, peripheral neuropathy and an autosomal dominant pedigree leading to a diagnosis of TRPV4-path**
I. Öncel; Q. Loic; G. Haliloglu; J. Melki; H. Topaloğlu
- P.341 **Case report: CMT2D with intermediate pattern. an expanding phenotype?**
E. Estephan; P. Sampaio; F. Souza; M. Rocha; E. Zanoteli; W. Marques Jr
- P.342 **Molecular characterization in Charcot-Marie-Tooth in Argentina: 121 case series**
E. Cavassa; M. Muntadas; M. Pauni; M. Garcia Erro; G. Vazquez
- P.343 **Colchicine induced neuromyopathy in a patient using concomitant diuretics**
S. Ho; D. Bae
- P.344 **Lipodystrophy and muscle hypertrophy**
O. Ekmekci; M. Argin; H. Karasoy
- P.345 **Serum auto-antibody positivity induced by intravenous immunoglobulin (IVIG) infusion**
R. Al-dahhak; J. Conway; L. Bostan-Shirin

- P.346 **Small molecule image-based screen identifies modulators of PML nuclear body phenotype in ALS models**
S. Cao; S. Parelkar; H. Ko; P. Thompson; L. Hayward
- P.347 **Feasibility and validation of modified oculobulbar facial respiratory score (mOBFRS) in amyotrophic lateral sclerosis**
N. Goyal; M. Wencel; N. Araujo; E. Medina; D. Nguyen; L. Zhang; T. Mozaffar
- Metabolic myopathies II (P.348-358)**
Chairpersons: Antonio Toscano and Claudio Bruno
- P.348 **Identification of late-onset Pompe disease with nationwide high-risk screening study in Japan**
K. Ogata; M. Kosuga; E. Takeshita; T. Matsumura; K. Ishigaki; S. Ozasa; H. Arahata; K. Sugie; T. Takahashi; S. Kuru; M. Kobayashi; H. Takada; A. Hattori; M. Takahashi; N. Tanaka; T. Kimura; M. Funato; T. Okuyama; H. Komaki
- P.349 **Late-onset Pompe disease: still a missing diagnosis?**
M. Oliveira Santos; I. Conceição
- P.350 **A novel hybrid promote directing AAV-mediated expression of acid alpha-glucosidase to liver, muscle and CNS yields optimized outcomes in a mouse model of Pompe disease**
G. Heffner; L. James; C. Chaivorapol; M. Mei; T. Stinchcombe; J. Ton; B. Schoser; H. Meng; M. Lawlor; S. Prasad; J. Gray
- P.351 **Exploring study design and endpoint selection to evaluate safety, preliminary efficacy, and dose selection of AAV8 gene therapy in patients with infantile and late onset Pompe disease**
S. Rico; B. Schoser; D. Dimmock; M. Lawlor; E. James; M. Morton; S. Prasad
- P.352 **A novel recombinant human acid alpha-glucosidase, ATB200, co-administered with a pharmacological chaperone, leads to greater substrate reduction and improvement in Pompe disease-relevant markers compared to alglucosidase alfa in *Gaa* KO mice**
Y. Lun; S. Xu; R. Soska; A. Nair; M. Frascella; A. Garcia; A. Ponery; J. Feng; C. Della Valle; R. Gotschall; H. Do; K. Valenzano; R. Khanna
- P.353 **Dissecting late onset Pompe disease outcomes. What are we measuring?**
J. Corderf; F. Chloca; A. Jáuregui; D. Flores; A. Dubrovsky
- P.354 **Safety and efficacy of recurrent inspiratory muscle training in late onset Pompe disease**
S. Wenninger; E. Greckl; H. Babacic; B. Schoser
- P.355 **Glycogen storage disease type IV: a wide clinical range of neuromuscular phenotypes**
H. Kölbl; A. Della Marina; O. Kaiser; F. Stehling; J. Weis; A. Abicht; U. Schara
- P.356 **Lower limb muscle strength and function in a cross-sectional study of patients with glycogen storage disease type IIIa**
V. Decostre; P. Laforêt; K. Kachel; A. Canal; G. Ollivier; F. Petit; P. Labrune; JY. Hogrel
- P.357 **Expression analysis of glycogenin-1 and glycogenin-2 in patients with glycogen storage disease XV**
K. Visuttijai; C. Thomsen; C. Hedberg-Oldfors; A. Oldfors
- P.358 **The diagnostic value of hyperCKemia induced by the non-ischemic forearm exercise test**
JY. Hogrel; C. Chéraud; I. Ledoux; G. Ollivier; R. Ben Yaou; F. Leturcq; A. Behin; T. Stojkovic; B. Eymard; P. Laforet
- FSHD/OPMD/EDMD/DM1 (P.359-371)**
Chairpersons: Julie Dumonceaux and Scott Harper
- P.359 **Evaluation of dynamic movement orthoses (DMO) as a means to relieve pain and fatigue in patients with facioscapulohumeral muscular dystrophy**
P. Drivsholm; L. Busk; T. Nybro; U. Werlauff
- P.360 **Longitudinal MR evaluation of inflammatory lesions in muscle of patients with facioscapulohumeral muscular dystrophy**
J. Dahlqvist; N. Poulsen; S. Oestergaard; F. Fornander; A. Eisum; C. Thomsen; J. Vissing
- P.361 **Estimating thigh muscle volume using bioelectrical impedance analysis with reference to contractile muscle volume assessed by nuclear magnetic resonance imaging**
D. Bachasson; J. Mosso; B. Marty; P. Carlier; JY. Hogrel
- P.362 **Regulation of facioscapulohumeral muscular dystrophy candidate protein DUX4**
J. Eidahl; L. Zhang; M. Hoover; O. Branson; M. Freitas; S. Harper
- P.363 **Progressive myopathy in a new mouse model of facioscapulohumeral muscular dystrophy facilitates development of targeted molecular therapies**
C. Giesige; L. Wallace; K. Heller; J. Eidahl; A. Fowler; N. Pyne; N. Saad; M. Alkharsan; A. Rashnonejad; G. Chermahini; J. Domire; D. Mukweyi; S. Garwick-Coppens; S. Guckes; K. McLaughlin; L. Rodino-Klapac; S. Harper
- P.364 **Natural microRNAs as potential modifiers of DUX4 toxicity in facioscapulohumeral muscular dystrophy**
N. Saad; M. Al-Kharsan; S. Garwick-Coppens; N. Pyne; S. Harper
- P.365 **Results for a dose-escalation phase 2 study to evaluate ACE-083, a local muscle therapeutic, in patients with facioscapulohumeral muscular dystrophy**
J. Statland; E. Bravver; C. Karam; L. Elman; N. Johnson; N. Joyce; J. Kissel; P. Shieh; L. Korngut; C. Weihl; R. Tawil; A. Amato; C. Campbell; A. Genge; G. Manousakis; A. Leneus; B. Miller; M. Sherman; C. Glasser; K. Attie
- P.367 **BB-301: a single "silence and replace" AAV-based vector for the treatment of oculopharyngeal muscular dystrophy**
V. Strings-Ufombah; A. Malerba; S. Harbaran; F. Roth; O. Cappellari; N. Nguyen; S. Kao; K. Takahashi; C. Kloth; P. Roelvink; G. Dickson; C. Trollet; D. Suhy
- P.368 **Clinical characteristics of 4 patients with childhood-onset reducing body myopathy in Japan**
A. Ishiyama; S. Kusabiraki; M. Inoue; Y. Oya; H. Miyahara; E. Takeshita; Y. Motohashi; H. Komaki; M. Sasaki; I. Nishino

- P.369 **A Chinese case of the early onset recessive Emery-Dreifuss-like phenotype without cardiomyopathy**
L. Wang; H. Li; C. Zhang
- P.370 **Diffusion tensor imaging and voxel based morphometry correlates with the CTG repeats and motor function in adult onset myotonic dystrophy type 1**
J. Park; S. Hwang; Y. Chang; H. Song
- P.371 **Do scientific publications fit with DM1 individuals expectations? - A systematic review and comparative study**
M. De Antonio; C. Dogan; D. Hamroun; A. Geille; B. Eymard; Study Group; G. Bassez
- Next generation sequencing and experimental myology (P.372-387)**
Chairpersons: Silvère van der Maarel and Conrad Weihl
- P.372 **Next generation sequencing: new phenotype-genotype correlations**
R. Juntas Morales; C. Cances; P. Cintas; D. Renard; G. Sole; C. Espil; F. Rivier; U. Walther Louvier; E. Uro-Coste; A. Perrin; N. Leboucq; V. Rigau; M. Arne-Bes; F. Duval; B. Acket; D. Peyroulan; C. Theze; H. Pegeot; M. Cossee
- P.373 **The NIH-NNDCS/CMG integrated clinico-genomic approach to undiagnosed pediatric neuromuscular patients in the NGS era**
S. Donkervoort; A. Foley; D. Saade; S. Neuhaus; P. Mohassel; D. Bharucha-Goebel; K. Chao; B. Cummings; M. Lek; D. MacArthur; C. Bönnemann
- P.374 **Identification of gene mutations in patients with primary periodic paralysis using targeted next-generation sequencing**
S. Luo; M. Xue; J. Sun; K. Qiao; J. Song; s. Cai; W. Zhu; L. Zhou; J. Xi; J. Lu; X. Ni; T. Dou; C. Zhao
- P.375 **Application of next-generation sequencing using customized targeted gene panel for neuromuscular disorders in South Korea**
J. Lee; H. Shin; S. Lee; J. Choi; S. Kim; Y. Choi
- P.376 **International DMD: a project devoted to dystrophin mutation identification by NGS technology in eastern Europe and northern African countries**
R. Selvatici; C. Trabaneli; S. Neri; R. Rossi; S. Fini; P. Rimessi; A. Venturoli; M. Neri; F. Gualandi;
A. Ferlini
- P.377 **The latin America experience with a next generation sequencing genetic panel for recessive limb-girdle muscular weakness**
T. Almeida; M. Guecaimburu Ehuleteche; A. Perna; J. Bevilacqua; A. Dubrovsky; M. Franca; S. Vargas; M. Hegde; K. Claeys; V. Straub; N. Daba; R. Faria; A. Loaeza; V. Luccerini; M. Periquet; S. Sparks; N. Thibault; R. Araujo
- P.378 **Clinical nonsense mutations in neuromuscular disorders**
E. Zapata-Aldana; C. Campbell
- P.379 **Transcriptome analysis of trans-differentiated myotubes for the diagnosis of neuromuscular disorders**
H. Gonorazky; S. Naumenko; D. Kao; P. Mashouri; A. Ramani; K. Mathews; M. Tarnopolsky; S. Moore; M. Brudno; J. Dowling
- P.380 **The complexity of splicing pattern in human adult skeletal muscles: a key to understanding genotype-phenotype correlations**
M. Savarese; P. Jonson; S. Huovinen; L. Paulin; P. Auvinen; B. Udd; P. Hackman
- P.381 **Muscle satellite cells and impaired late stage regeneration in different murine models of muscular dystrophies**
A. Ribeiro Junior; R. Ishiba; S. Alcântara Fernandes; C. Freitas Almeida; D. Ayub Gerrieri; A. Fernandes dos Santos; I. Santos; E. Souza; M. Vainzof
- P.382 **Klf5 has essential roles in myoblast differentiation and survival during fetal muscle development**
S. Hayashi; Y. Oishi; I. Manabe; I. Nishino
- P.383 **Cullin-3 is required for normal skeletal muscle development**
J. Blondelle; K. Tallapaka; P. Shapiro; M. Ghassemian; J. Singer; S. Lange
- P.384 **RNAseq in urine-derived stem cells identified the expression of 308 neuromuscular gene transcripts**
M. Falzarano; H. Osman; R. Rossi; R. Selvatici; M. Neri; F. Gualandi; M. Fang; I. Zhiyuan; A. Grilli; S. Biciato; A. Ferlini
- P.385 **Physician-Level muscle disease classifier for computer-aided diagnostics with neural networks**
Y. Kabeya; Y. Takeuchi; H. Nakano; M. Okubo; M. Inoue; R. Tokumasu; I. Ozawa; A. Takano; T. Iwamori; S. Yonezawa; I. Nishino
- P.386 **A versatile, modular digital script for automated high-throughput multiparametric myofibre analysis in brightfield and epifluorescent paradigms**
M. Ellis; V. Sardone; S. Torelli; S. Saeed; A. Sigurta; N. Hill; D. Scaglioni; L. Feng; C. Sewry; M. Singer; F. Muntoni; R. Phadke
- P.387 **Nonsense mutation induced exon skipping in Becker muscular dystrophy**
M. Okubo; S. Noguchi; S. Hayashi; M. Matsuo; I. Nishino
- 20:00-00:00 **Networking dinner**

Saturday 6 October 2018

08:00-	Conference desk opens
08:30-10:30	Poster Highlights – Auditorium <i>Chairpersons: Kathryn Swoboda and Werner Stenzel</i>
10:00-10:30	Morning refreshments, exhibition and posters
10:30-11:30	WMS General Assembly - Auditorium
11:30-13:00	Late breaking session - Auditorium <i>Chairpersons: Valeria Ricotti and Carsten Bönnemann</i>
13:00-13:30	Prize giving and welcome to the 24th WMS Congress Handover of the WMS flag and close of congress – Auditorium
13:30-14:30	Lunch and end of congress