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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) *Chairpersons: Thomas Voit and Marcelo Rugiero*
	I.1	**Age-related neuromuscular junction instability: causes and consequences** M. Ruegg; P. Castets; S. Lin; H. Brenner; D. Ham; M. Rich
	I.2	**New genes and better treatment for congenital myasthenic syndromes** D. Beeson
10:30-11:00		**Morning refreshments, exhibition and posters**
11:00-11:45		**Myasthenia gravis;** Invited lecture (I.3) - Auditorium *Chairpersons: Gisèle Bonne and Bruno Eymard*
	I.3	**From trial-and-error to trials without errors in myasthenia gravis** J. Verschuuren
11:45-13:15		**Selected oral presentations I - New genes, functions and biomarkers** (O.1-6) *Chairpersons: Gisèle Bonne and Bruno Eymard*
	O.1	**Recessive mutations in *BET1* and *GOSR2* establish Q-SNARE Golgi-vesicle-transport genes as a cause for congenital muscular dystrophy with epilepsy** 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
	O.2	**Mutations in fast skeletal troponin C (*TNNC2*) cause contractile dysfunction** 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
	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)** M. Sasaki-Honda; T. Jonouchi; M. Arai; A. Hotta; S. Mitsuhashi; I. Nishino; R. Matsuda; H. Sakurai
	O.4	**Single-cell RNA-sequencing in facioscapulohumeral muscular dystrophy disease etiology and development** A. van den Heuvel; A. Mahfouz; S. Kloet; J. Balog; B. van Engelen; R. Tawil; S. Tapscott; S. van der Maarel
	O.5	**Association of phosphorylated neurofilament heavy chain (pNF-H) with nusinersen treatment of SMA: analyses from the ENDEAR and CHERISH studies** B. Darras; R. Finkel; E. Mercuri; C. Sumner; M. Oskoui; E. Tizzano; M. Ryan; G. Zhao; M. Petrillo; C. Stebbins; W. Farwell
	O.6	**Free $Mg2^{+}$ intramuscular concentration determined by combined 31P and 1H NMR spectroscopy as a potential outcome measure in Duchenne muscular dystrophy** H. Reyngoudt; P. Carlier

https://doi.org/10.1016/j.nmd.2018.06.017
0960-8966

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 **ANO5 - 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-Montáñez; 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. Quinlivian; 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. Grosmann; 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 Locht; 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 *ASCC1* 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 ***ASC1*-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. Koparir; S. Coppen; E. Pierce-Hoffman; A. Beggs; L. Servais; A. Ferreiro

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. lacene; 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; JY. 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. Rodriguez 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 clincal 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. Borras; 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. Annoussamy; A. Daron; Y. Péréon; C. Cances; 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. Baijot; 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**
JY. 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

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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

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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^{TM}: A patient registry study examining the use of ataluren (Translarna^{TM}) 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. Zschaeck; C. Jimenez - Mallebrera; L. Solé; M. Cubells; C. Jou; A. Nascimento

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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. Dorchies; E. Gayi; L. Neff; L. Scapozza

P.116 **Edasalonexent, an NF-κB Inhibitor, slows disease progression over more than a year compared to control period in 4 to 7-year old patients with Duchenne muscular dystrophy**
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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**
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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**
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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

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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

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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**
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P.131 **AAV-mediated gene transfer with *GALGT2* in Duchenne muscular dystrophy: design of an ongoing phase I/II clinical trial**
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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

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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

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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

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K. Aragon-Gawinska; A. Seferian; L. Vanden Brande; A. Daron; A. Ulinici; N. Deconinck; M. Annoussamy; C. Vuillerot; C. Cances; 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

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A. Tagiyev; N. Bulut; G. Aydin; I. Alemdaroğlu Gürbüz; N. Eroğlu; O. Yilmaz; G. Haliloğlu; A. Karaduman; H. Topaloğlu

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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**
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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. Ardicli; 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. Aráoz; 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. Yılmaz; E. Sönmezler; A. Yaramis; S. Güngör; S. Laurie; S. Beltran; I. Gut; H. Lochmüller; 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

18:00-19:30 **Symposium 1**

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. Ferreiro

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. Hourdé; 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. Bönnemann; J. Dowling; M. Lawlor; W. Müller-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. Bönnemann

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 **Microutrophin delivery shows phenotype improvement in mdx mice**
T. Egorova; D. Vlodavets; 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 DMDmdx 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 (Bl10/*mdx*) mouse models**
L. Dalle Pazze; 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. Vlodavets; A. Deikin

P.204 **R-DMDdel52, a novel preclinical rat model of Duchenne muscular dystrophy**
M. Goddard; B. Drayton; F. Piétri-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 sarconeos (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 Koeveringe; 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 **Unsolicited 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. Freilinger; R. Bittner; G. Bernert

Congenital myopathies: nemaline and titinopathies (P.225-244)
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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. Goullee; 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. Trangulao; 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. Iannaconne; 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. Grosmann; 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)
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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

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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

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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

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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

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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**
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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

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T. Crawford; C. Sumner; M. Petrillo; C. Stebbins; W. Farwell

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E. Sneszhko; P. Baudin; P. Carlier

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B. Marty; P. Carlier

P.294 **Muscle biopsy in the study of muscle disease in pediatric population**
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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. Escolar; M. Hermreck; H. Semus; D. Hood; A. Jackson

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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

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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. Alemdaroğlu 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

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R. Bendixen; A. Kelleher; M. Feltman; N. Little

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E. Henricson; R. Abresch; A. Bagley; E. Goude; C. Owens; L. Williams; C. McDonald

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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

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C. Iwabe-Marchese; N. Morini Jr; c. Sanches; T. Rosa

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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. Artilheiro; E. Oliveira; N. Carvas Junior; F. Favero; F. Caromano; C. Sá; M. Voos

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F. Munell; D. Gomez-Andres; A. Sanchez-Montañez; L. Costa Comellas; S. Ferrer-Aparicio; P. Romero; S. Quijano-Roy; M. Olivé

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S. Noguchi; M. Ogawa; I. Nishino

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P.331 **Longitudinal changes of motor outcome measures in individuals with *COL6*-RDs and *LAMA2*-RD**
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P. Massaro Sanchez; A. Savransky; C. Rugilo; S. Monges

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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

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I. Öncel; Q. Loic; G. Haliloğlu; J. Melki; H. Topaloğlu

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S. Ho; D. Bae

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O. Ekmekci; M. Argin; H. Karasoy

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S. Cao; S. Parelkar; H. Ko; P. Thompson; L. Hayward

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N. Goyal; M. Wencel; N. Araujo; E. Medina; D. Nguyen; L. Zhang; T. Mozaffar

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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

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S. Rico; B. Schoser; D. Dimmock; M. Lawlor; E. James; M. Morton; S. Prasad

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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

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J. Corderí; F. Chloca; A. Jáuregui; D. Flores; A. Dubrovsky

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S. Wenninger; E. Greckl; H. Babacic; B. Schoser

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V. Decostre; P. Laforêt; K. Kachetel; 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

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JY. Hogrel; C. Chéraud; I. Ledoux; G. Ollivier; R. Ben Yaou; F. Leturcq; A. Behin; T. Stojkovic; B. Eymard; P. Laforet

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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**
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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**
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N. Saad; M. Al-Kharsan; S. Garwick-Coppens; N. Pyne; S. Harper

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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

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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

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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. Selvativci; C. Trabanelli; 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 Ehuletche; 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; l. Zhiyuan; A. Grilli; S. Bicciato; 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 *Chairpersons: Kathryn Swoboda and Werner Stenzel*
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 *Chairpersons: Valeria Ricotti and Carsten Bönnemann*
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**