



WMS25: Virtual Congress 30th September - 2nd October 2020

Preliminary Detailed Programme All times are in British Summer Time (BST)

Monday, 28 September 2020

14:50 – 15:00	President's Introduction
15:00 – 16:30	WMS Industry Symposia 1 and 2 <i>(2 x 90 minute parallel sessions incorporating live Q&A)</i>
16:30 – 17:00	Comfort break
17:00 – 18:30	WMS Industry Symposia 3 and 4 <i>(2 x 90 minute parallel sessions incorporating live Q&A)</i>
18:30 – 19:00	Comfort break
19:00 – 20:30	WMS Industry Symposia 5 and 6 <i>(2 x 90 minute parallel sessions incorporating live Q&A)</i>
	E-Posters available to view on demand

Tuesday, 29 September 2020

15:00 - 22:00	Pre-Congress Teaching Course (congress registration required) Congress Industry Symposia and all E-Posters available to view on demand
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Wednesday, 30 September 2020

15:00-15:15	President's Welcome
15:15-15:45	Opening lecture COVID-19 for NMD, telemedicine, changing practice in the pandemic situation
	The COVID-19 pandemic has so far not been the largest, or the most lethal pandemic of the last 100 years, but the global response to it has been unprecedented, both in medical and in societal terms. It has led to a significant change in behaviour, specifically in the way medicine is practised, and its longer- term effects cannot yet be predicted confidently. The concerns for people with neuromuscular disease specifically are significant: in addition to potentially being a risk group for severe infection,

they are put at risk by an interruption to their access to treatment. The World Muscle Society is making an effort to help them face these challenges, by providing advice for patients and carers, trying to define best practice parameters, and suggest avenues to manage the future changes in medical practice. This lecture discusses the approach, its effects on patients, and potential future developments in telemedicine.

Speaker: Max Damian

Session Moderator: Jiri Vajsar

15:45-16:00

Comfort break

16:00-16:30

Panel discussion 1

Pre-clinical requirements for novel neuromuscular therapies

It is a unique time for neuromuscular disorders, as an unprecedented number of therapeutic candidates are entering clinical trial for a range of diseases. With this surge of potential treatments, the importance of pre-clinical models and rigor in pre-clinical drug development has never been more important. It is critical to evaluate study designs, predictive power, reliability and relevance of current animal models in assessing the efficacy and safety of emerging therapies. In this panel, we have assembled four world leading experts on the application of pre-clinical models to therapy development. We will present key ideas related to best practice and discuss important pitfalls that have plagued pre-clinical drug programs. We will also identify current gaps in the field and offer ideas for future improvements.

Speakers: Annamaria DeLuca, Kanneboyina Nagaraju, Dominic Wells, Jennifer Morgan

Session Moderator: Jim Dowling

Content Moderator: Grace Yoon

16:30-16:45

Panel discussion 1 – Live Q&A

16:45-17:15

Comfort break, poster viewing, sponsors & exhibition, and chat room

17:15-17:45

Panel discussion 2

Gene/ advanced/ new therapeutics

Gene and transcript directed therapies are rapidly becoming a reality and a promise for many neuromuscular disorders. Various such therapies are in the clinic, in clinical trials, or in advanced preclinical development. This panel will discuss opportunities and challenges of these therapies, with a focus on AAV mediated gene therapy. Determinants of success and potential failure will be discussed, touching on issues such as AAV capsids, biodistribution, therapeutic windows of opportunity and durability of expression, seropositivity and redosing, immune mediated toxicity and immunomodulation, as well as pathways and roadblocks for ultrarare applications. This will be an open discussion drawing on recent examples and experiences in the field of clinical neuromuscular gene therapy.

Speakers: Carsten Bönnemann, Francesco Muntoni, Barry Byrne, Ana Buj Bello

Session Moderator: Kathryn Wagner

Content Moderator: Chris Weihl

17:45-18:00

Panel discussion 2 – Live Q&A

18:15- 19:15

WMS 25th Anniversary Quiz

Thursday, 1 October 2020

15:00-16:00

New genes in neuromuscular diseases – Selected oral presentations 1 (O.1-4)

(4 x 10 mins consecutive presentations plus 20 mins live Q&A)

Muscle researchers have identified novel genes and their variants that may be causative to neuromuscular disorders, assess these with transgenic studies, sequencing, and phenotypic and molecular correlation, etc., and discuss future studies and outlook.

Session Moderator: Gisèle Bonne

Assistant Moderator: Hernán Gonorazky

O.01

Mutations in the SMPX gene cause the first X-linked recessive form of distal myopathy

M. Johari; M. Savarese; A. Vihola; M. Jokela; A. Torella; G. Piluso; P. Jonson; H. Luque; A. Magot; F. Magri; C. Kornblum; T. Stojkovic; N. Romero; P. Lahermo; K. Donner; V. Nigro; P. Hackman; B. Udd

O.02

A dominant variant in DNAJB4 causes a myofibrillar myopathy

M. Inoue; A. Iida; K. Watanabe; Y. Hosoi; H. Miyajima; S. Hayashi; Y. Inoue; T. Inoue; S. Noguchi; I. Nishino

O.03

Dominant mutations in Junctin are related to malignant hyperthermia and exertional heat stroke

Y. Endo; L. Groom; N. Kraeva; A. Celic; P. Hopkins; S. Riazzi; R. Dirksen; J. Dowling

O.04

Heterozygous frameshift variants in hnRNPA2B1 cause a novel oculopharyngodistal muscular dystrophy

P. Mohassel; S. Donkervoort; H. Kim; A. Foley; X. Lornage; hnRNPA2B1 Study Group; N. Foulds; S. Hammans; T. Haack; J. Bohm; M. Tarnopolsky; V. Straub; J. Laporte; F. Muntoni; J. Taylor; C. Bönnemann

16:00-16:30

Comfort break, poster viewing, sponsors & exhibition, and chat room

16:30-17:30

OMICs and AI approaches for muscle diseases - Selected oral presentations 2 (O.5-8)

(4 x 10 mins consecutive presentations plus 20 mins live Q&A)

These presentations delve into new genetic, proteomic, and algorithmic approaches to evaluate neuromuscular disorders

Session Moderator: Werner Stenzel

Assistant Moderator: Louise Benarroch

O.05

Whole exome sequencing identifies compound heterozygous missense variants in the LOXL4 gene: a novel candidate cause of contractural myopathy

E. Cohen; I. Nelson; C. Gartioux; M. Beuvin; Z. Mezdari; F. Roth; R. Ben Yaou; S. Quijano-Roy; T. Stojkovic; R. Carlier; G. Bonne; V. Allamand

O.06

Haplotype and variant phasing of large muscle genes with linked-read sequencing

J. Lehtonen; M. Johari; H. Almusa; A. Lehesjoki; K. Wartiovaara; J. Saarela; P. Hackman; C. Wallgren-Pettersson; B. Udd; V. Lehtokari; K. Pelin; M. Savarese

O.07

Proteomic profiling in patients with Dermatomyositis

C. Preusse; A. Ross; D. Hathazi; A. Hentschel; H. Goebel; W. Stenzel

O.08

Deep convolutional-neural-network can differentiate twelve major muscular diseases better than human

Y. Kabeya; M. Okubo; S. Yonezawa; H. Nakano; M. Inoue; M. Ogasawara; Y. Saito; J. Tanboon; L. Indrawati; T. Kumutpongpanich; Y. Chen; R. Tokumasu; T. Iwamori; A. Takano; I. Nishino

17:30-19:30

Comfort break, poster viewing and sponsors & exhibition

17:30-19:30

Poster session 1

Autophagic myopathies / Myofibrillar Myopathies / Distal myopathies / Pompe disease (P.1-17)

P.01

Pre-clinical safety and efficacy of AT845, a muscle-directed AAV-based gene replacement therapy for Pompe disease

M. Eggers; C. Vannoy; J. Huang; P. Purushottoman; J. Brassard; J. Gray; M. Lawlor; C. Sadhu; F. Mavilio

P.03

NEO1/NEO-EXT studies: Safety and exploratory efficacy of repeat avalglucosidase alfa dosing after up to 6 years in late-onset Pompe disease (LOPD)

B. Schoser; R. Barohn; B. Byrne; O. Goker-Alpan; P. Kishnani; S. Ladha; P. Laforet; E. Mengel; L. Pena; S. Sacconi; V. Straub; J. Trivedi; P. Van Damme; A. van der Ploeg; J. Vissing; P. Young; K. Haack; I. Ivanina; Y. Wang; M. Dimachkie

P.04

Treatment outcome in classic-infantile Pompe disease: effects of dosing in clinical practice

I. Ditters; H. Huidekoper; M. Kruijshaar; A. Hahn; T. Mongini; F. Labarthe; M. Tardieu; J. van den Hout; A. van der Ploeg

P.05

Infantile onset Pompe disease: 10 years of experience in a pediatric reference centre for neuromuscular diseases.

C. Ortez; L. Carrera; J. Exposito; D. Natera; S. Zambudio; J. Colomer; A. Bazán; A. Pareja; E. Bobadilla; v. Sáez; J. Medina; C. Jou; A. Codina; J. Corbera; D. Yubero; L. Martorell; C. Jimenez - Mallebrera; A. Nascimento

P.06

Long-term effects of enzyme replacement therapy in an elderly cohort of late-onset Pompe disease patients

M. Winkler; C. von Landenberg; K. Kuchenbecker; J. Reimann; C. Kornblum

P.07

Efficacy and safety of avalglucosidase alfa in patients with late-onset Pompe disease: results from the phase 3 COMET trial

P. Clemens; The COMET Study Group

P.09

Chloroquine and hydroxychloroquine myopathy: Clinical spectrum and treatment outcomes

E. Naddaf; P. Pritikanta

P.10

Distal myopathy associated with two novel variants in SPTAN1

A. Meyer; W. Arnold; M. Waldrop; K. Flanigan

P.11

Pain characteristics and involvement of small epidermal nerve fibers in patients with late onset Pompe disease (LOPD)

E. Enax- Krumova; J. Görlach; A. Rosenbohm; K. Claeys; F. Montagnese; I. Schneider; D. Sturm; T. Nicoletto; T. Fangerau; A. Roth; J. Wanschitz; W. Löscher; A. Güttsches; S. Vielhaber; L. Zunk; H. Krämer-Best; B. Schoser; A. Hahn; A. Schänzer

P.12

Mutations in supervillin cause myopathy with myofibrillar disorganization and autophagic vacuoles

C. Hedberg-Oldfors

P.14

Autophagic vacuolar myopathy with AVSF (AVSF myopathy) including Danon disease and XMEA: A new clinical entity

K. Sugie; H. Komaki; T. Kurashige; D. Kaneda; N. Eura; T. Shiota; Y. Nishimori; N. Iguchi; H. Nanaura; T. Kiriyaama; E. Mori; I. Nonaka; I. Nishino

P.16

Illuminating the role of ER stress in immune mediated necrotizing myopathy with respect to defective chaperone-assisted selective autophagy

N. Fischer; C. Preusse; Y. Allenbach; O. Benveniste; A. Roos; H. Goebel; W. Stenzel

P.17

Screening for Pompe disease using muscle section in Japan

Y. Saito; K. Nakamura; T. Fukuda; H. Sugie; S. Hayashi; S. Noguchi; I. Nishino

Congenital myopathies I – Nemaline (P.18-30)

P.18

KBTD13, NEM6 congenital myopathy: deep muscle phenotyping reveals ring-rods fibers, nuclear clumps, and protein aggregates as new morphologic markers

K. Bouman; B. Küsters; J. De Winter; C. Gllet; E. Van Kleef; L. Eshuis; G. Brochier; A. Madelaine; C. Labasse; C. Boulogne; B. Van Engelen; C. Ottenheijm; M. Olive; N. Romero; N. Voermans; E. Malfatti

P.19

ACTA1-related myopathies: expanding the phenotype to include recognizable asymmetries of muscle involvement

A. Foley; S. Ferranti; D. Saade; P. Mohassel; S. Donkervoort; L. Medne; J. Dastgir; D. Bharucha-Goebel; K. Meilleur; M. Leach; M. Scavina; S. Yum; G. Tennekoon; C. Bönemann

P.20

Mouse models of nemaline myopathy display structural and functional abnormalities of mitochondria

J. Tinklenberg; R. Slick; J. Sutton; M. Prom; E. Ott; S. Danielson; M. Vanden Avond; M. Beatka; H. Meng; M. Grzybowski; J. Heisner; J. Ross; J. Ochala; K. Nowak; L. Zhang; A. Geurts; D. Stowe; F. Montanaro; M. Lawlor

P.21

Ovine congenital progressive muscular dystrophy (OCPMD) is a model of TNNT1 nemaline myopathy

J. Clayton; E. McNamara; H. Goulee; S. Conijn; K. Muthsam; G. Musk; D. Coote; J. Kijas; A. Testa; R. Taylor; M. O'Hara; D. Groth; C. Ottenheijm; G. Ravenscroft; N. Laing; K. Nowak

P.22

Proteomic profiling in Nemaline myopathy to identify molecular phenotypes that contribute to disease heterogeneity

R. Slick; J. Tinklenberg; H. Meng; M. Beatka; M. Prom; E. Ott; F. Montanaro; L. Zhang; H. Granzier; E. Hardeman; A. Geurts; M. Lawlor

P.23

Nutritional status and functioning of patients with nemaline myopathy and related disorders: A pilot study

V. Lehtokari; M. Tammepuu; M. Similä; S. Strang-Karlsson; S. Hiekkala; C. Wallgren-Pettersson

P.24

Respiratory muscle weakness in nemaline myopathy patients

E. van Kleef; C. Ottenheijm; M. Gaytant; W. de Weerd; B. Vosse; B. van Engelen; N. Voermans; J. Doorduyn

P.25

Autosomal recessive congenital myopathy with splicing mutation (c.21522+3A>G) of NEB gene

J. Lee; H. Park; Y. Choi

P.26

A new intronic mutation in nebulin myopathy

N. Chrestian; N. Laflamme; Y. Labrie; N. Rioux; M. Dugas; S. Rivest; B. Luce

P.27

Asymmetric muscle weakness due to ACTA1 mosaic mutations

X. Lornage; H. Amthor; S. Quijano-Roy; R. Carlier; N. Monnier; N. Romero; J. Laporte; J. Bohm

P.28

Distinctive pathological features of inherited and sporadic late-onset nemaline myopathies

S. Nicolau; A. Dasgupta; D. Selcen; A. Engel; J. Doles; M. Milone

P.29

AAV gene therapy for TNNT1-associated Nemaline myopathy

E. D'Ambrosio; H. Gray-Edwards; H. Grimason; L. Labdi; K. Mesa; M. Otero; M. Sena-Esteves; M. Otero

P.30

Multi exon skipping as a potential therapy for nemaline myopathy in Zebrafish

S. Viththiyapaskaran

Myasthenia & related disorders (P.31-39)

P.31

Identification of new recessive mutations in synaptotagmin-2 responsible for severe and early presynaptic forms of congenital myasthenic syndrome

S. Bauché; A. Sureau; D. Sternberg; J. Rendu; C. Buon; J. Messéant; M. Boëx; D. Furling; J. Fauré; X. Latypova; A. Bernabe Gelot; M. Mayer; F. Laffargue; M. Nougues; B. Fontaine; B. Eymard; A. Isapof; L. Strohlic

P.32

Treatment of metastatic urothelial cancer patients with myasthenia gravis with PD-1 inhibitors

A. Ishii; M. Yokoyama; H. Tsuji; Y. Fujii; A. Tamaoka

P.33

Clinical features and evolution of juvenile myasthenia gravis in an Israeli cohort

A. Kuzminsky; Y. Nevo; S. Aharoni; M. Rabie

P.34

Congenital myasthenic syndrome in two Egyptian siblings due to a novel low-expressor AChR epsilon subunit mutation

M. Gomez Garcia de la Banda; N. Fahmi; D. Sternberg; P. Blondy; S. Quijano-Roy; E. Malfatti

P.35

Improvement of strength and muscle MRI with dichlorphenamide in hypokalemic periodic paralysis myopathy: A case report

P. Mongiovi; S. Meyers; R. Griggs; K. Eichinger; E. Ciafaloni

P.36

Novel SCN4A mutations causing mild myotonic phenotype

J. Palmio; R. Mannikko; B. Udd

P.37

Autophagy affected in patients with hypokalemic periodic paralysis

T. Krag; S. Holm-Yildiz; N. Witting; J. Vissing

P.38

Autosomal recessive inheritance in hypokaliemic periodic paralysis due a homozygous SCN4A gene mutation

G. Remiche; L. Desmyter; M. Grenet; I. Vandernoot; O. Devuyt; G. Smits; V. Potmans

P.39

Free-living physical activity and sedentary behaviour in auto-immune myasthenia gravis: a cross-sectional study

S. Birnbaum; D. Bachasson; T. Sharshar; R. Porcher; J. Hogrel; P. Portero

DMD & BMD – Clinical (P.40-60)

P.40

A longitudinal study of respiratory function in adult patients with Becker muscular dystrophy

B. De Wel; S. Willaert; A. Nadaj-Pakleza; D. Testelmans; B. Buyse; K. Claeys

P.41

Development of cardiomyopathy, respiratory insufficiency, and loss of ambulation in Becker muscular dystrophy (BMD) - A systematic literature review

K. Johnston; R. Salhany; E. Ciafaloni; A. Mickle; S. Miller; K. Gooch

P.43

Patient and family centric care for Duchenne and Becker muscular dystrophy: towards the IPU (Integrated Practice Unit) model

B. Wong; T. Seckler; L. Dalla Pазze; R. Brown; L. Rhein; T. Flotte

P.44

Cardio-renal syndrome in Duchenne muscular dystrophy

B. Wong; C. Webb; M. Fahey; T. Meyer; K. Laraja; C. Kashtan; A. Salerno; K. Shellenbarger; N. Gupta

P.45

Prophylactic cardiac medication is associated with delayed left ventricular dysfunction and reduced death in childhood-onset dystrophinopathy

K. Mathews; S. Perlman; K. Conway; E. Ciafaloni; S. Thomas; J. Mann; P. Romitti

P.46

Bone health in Duchenne muscular dystrophy -an audit

N. Rajaraman; D. Parasuraman

P.47

Keeping a healthy weight is challenging in Duchenne muscular dystrophy: large influence of ambulatory level, limited influence of dietary intake

S. Houwen-van Opstal; A. de Baaij-Daalmeyer; D. Bot; M. Jansen; C. Dietvorst; K. Corbet; E. Cup; M. Willemsen; E. Niks; I. Groot

P.48

Facilitators and barriers to wearing hand orthoses by adults with Duchenne muscular dystrophy. A mixed method study design

S. Houwen- van Opstal; Y. van den Elzen; M. Jansen; M. Willemsen; E. Cup; I. Groot

P.49

Demographics of patients with nonsense mutation Duchenne muscular dystrophy receiving ataluren in the STRIDE Registry

F. Muntoni; E. Mercuri; F. Buccella; I. Desguerre; J. Kirschner; A. Nascimento Osorio; M. Tulinius; J. Jiang; A. Kristensen; S. Johnson; R. Able; P. Trifillis; C. Santos

P.50

Diffusion tensor MRI metrics reflect larger fibre sizes and heterogeneity of fibre size in Becker muscular dystrophy patients

D. Cameron; J. Burakiewicz; N. van de Velde; C. Baligand; T. Veeger; M. Hooijmans; J. Verschuuren; E. Niks; H. Kan

P.51

DMD Hub: demonstrating impact for DMD trials

E. Heslop; M. Guglieri; A. Irvin; C. Turner; B. Crow; E. George; A. Johnson; E. Crossley; F. Muntoni;

V. Straub

P.52

The natural history of the cardiomyopathy of Duchenne and Becker muscular dystrophy

E. Nealon; B. Beckman; N. Kertesz; L. Cripe

P.53

Duchenne muscular dystrophy: does being a younger affected sibling confer long term improved outcomes through earlier diagnosis and management?

M. Main; J. Pisco-Domingos (Deceased); C. Rye; M. Scoto; F. Muntoni

P.54

Has the time to diagnosis of Duchenne muscular dystrophy improved? Data from the Muscular dystrophy surveillance, tracking, and research network

E. Ciafaloni; O. Fapo; K. Conway; N. Street; P. Romitti; C. Westfield; D. Fox; K. Matthews; J. Mann; S. Thomas; A. Soim; MD STARnet

P.55

Prognostic factors for loss of ability to rise from supine in Duchenne muscular dystrophy (DMD)

N. Goemans; C. McDonald; J. Signorovitch; G. Sajeev; M. Fillbrunn; H. Wong; E. Mercuri; K. Vandenberg; F. Muntoni; S. Ward; Investigators for PRO-DMD-01; The Imaging DMD study; iMDEX; cTAP

P.56

Disease attributes most important from a societal perspective: A case study involving Duchenne muscular dystrophy

S. Szabo; I. Audhya; D. Malone; P. Neumann; D. Feeny; M. Harwood; K. Gooch

P.57

Comparison of North Star Ambulatory Assessment scores in siblings with Duchenne muscular dystrophy

C. Rye; M. Main; J. Domingos; F. Muntoni

P.58

Hypermobility in young boys with Duchenne muscular dystrophy and its effect on attainment of North Star Ambulatory Assessment functional skills

L. Abbott; V. Selby; V. Ayyar Gupta; S. Wadsworth; A. Wolfe; M. Chesshyre; G. Baranello; M. Scoto; A. Manzur; M. Main; F. Muntoni

P.59

Functional progression in young DMD

V. Ayyar Gupta; L. Abbott; M. Chesshyre; M. Main; G. Baranello; M. Scoto; A. Manzur; F. Muntoni; U. Northstar Clinical Network

P.60

Involving families in the design of a weight management program for Duchenne muscular dystrophy- Supporting nutrition and optimising wellbeing program

N. Billich; P. Bray; H. Truby; M. Evans; B. Sowerby; K. de Valle; K. Carroll; D. Villano; M. Ryan; Z. Davidson

Muscle function & homeostasis / Molecular therapeutic approaches (P.62-77)

P.62

ASPIRO Gene Therapy Trial In X-Linked Myotubular Myopathy (XLMTM): Update on Safety And Efficacy

C. Bönnemann, P. Shieh, N. Kuntz, W. Müller-Felber, A. Blaschek, A. Reghan Foley, D. N. Saade, M. W. Lawlor, Laurent Servais, Weston Miller, M. Noursalehi, S. Prasad, S. Rico, J. Dowling

P.63

AAV.CAPN3 gene therapy for Limb-girdle muscular dystrophy type 2A (LGMD2A): Phenotypic

improvements in the calpain3(CAPN3)-null mouse

B. Ozes; D. Murrey; M. Myers; L. Chen; K. Moss; A. Ridgley; C. Wier; Z. Sahenk

P.66

Cell sorting and characterization of urine-derived stem cells (USCs) using the novel technology Celector

M. Falzarano; N. Spedicato; A. Margutti; R. El Dani; R. Rossi; S. Zia; P. Reschiglian; B. Roda; A. Grilli; S. Bicciato; A. Ferlini

P.67

Skeletal muscle atlas: a tool for the muscle community

E. Lacène; M. Beuvin; T. Evangelista; N. Romero; B. Cadot

P.68

O-GlcNAcylation and its interplay with phosphorylation on desmin behaviour: focus on its partition and its interaction with alphaB-crystallin

C. Claeysen; B. Bastide; C. Cieniewski-Bernard

P.69

Generation and characterization of Pax7-HA knock-in mice

S. Hayashi; Y. Inoue; H. Kosako; T. Inoue; S. Noguchi; I. Nishino

P.72

Importance of myofibre-type specific differences in disease manifestation for muscular dystrophies: focus on dysferlinopathy

E. Lloyd; G. Pinniger; R. Murphy; M. Watt; M. Grounds

P.73

Depletion of ATP limits membrane excitability of skeletal muscle by increasing both ClC1-open probability and membrane conductance

P. Leermakers; K. Dybdahl; K. Husted; A. Riisager; F. de Paoli; T. Pinos; J. Vissing; T. Krag; T. Pedersen

P.74

Studies on a potential interaction between nebulin alternative isoform S21b and ferritin light chain (FTL)

J. Laitila; M. Hanif; J. Sarparanta; J. Lehtonen; A. Khattab; M. Grönholm; C. Wallgren-Pettersson; K. Pelin

P.75

Studies of YBX3 and its variants in myogenesis

L. Sagath; J. Blondelle; J. Laitila; V. Lehtokari; K. Kiiski; M. Grönholm; C. Wallgren-Pettersson; S. Lange; K. Pelin

P.76

Validation of in vivo and ex vivo readouts in a murine model of hind limb unloading to optimize translational research on skeletal muscle atrophy

P. Mantuano; F. Sanarica; O. Cappellari; B. Boccanegra; N. Tarantino; E. Conte; M. De Bellis; G. Camerino; S. Pierno; A. De Luca

P.77

STIM1 heterozygous mutation pathological mechanism in tubular aggregate myopathy

A. Salvi; M. Bartoli

SMA - Clinical (P.78-92)

P.78

Burden of illness of spinal muscular atrophy type 1 (SMA1): an update

M. Droege; O. Dabbous; R. Arjunji; M. Gauthier-Loiselle; M. Cloutier; D. Sproule

P.79

Analysis of Cobb angle and clinical characteristics in children with spinal muscular atrophy who enrolled in CHERISH and SHINE

S. Dunaway Young; J. Montes; A. Glanzman; R. Gee; J. Day; R. Finkel; B. Darras; D. De Vivo; G. Gambino; R. Foster; J. Wong; B. Kandinov; Z. Berger

P.80

Distribution of weight and height in children and adolescents with spinal muscular atrophy: An observational retrospective study in the United States

B. Darras; S. Guye; J. Hoffart; S. Schneider; I. Gravestock; K. Gorni; S. Fuerst-Recktenwald; R. Scalco; R. Finkel; D. De Vivo

P.81

Nusinersen experience in later onset spinal muscular atrophy: A tertiary center

G. Öz Tunçer; F. Mıhçı; M. Dolu; Ü. Akça; S. Aydın; S. Türk; A. Aksoy; H. Taşdemir

P.83

A search for systemic manifestations of spinal muscular atrophy (SMA): A matched-cohort study using insurance claims data in the USA

J. Mouchet; M. Oskoui; S. Lipnick; S. Roumpanis; R. Naik; E. Regulier; R. Scalco; B. Darras

P.84

Magnetic resonance imaging of muscle and motor functional changes in patients with spinal muscular atrophy treated with nusinersen

Y. Shimizu-Motohashi; H. Yajima; A. Ishiyama; E. Takeshita; K. Mizuno; N. Sato; M. Sasaki; H. Komaki

P.85

Evolution at 18 months of SMA type 1 patients treated with nusinersen

S. De Lucia; A. Phelep; A. Seferian; P. Foyer; U. Walther-Louvier; J. Durigneux; C. Cancès; J. Ropars; C. Vuillerot; L. Servais; O. Boespflug

P.86

Diminished muscle oxygen Uptake and fatigue during exercise in ambulatory spinal muscular atrophy patients

J. Montes; A. Goodwin; M. McDermott; D. Uher; F. Hernandez; K. Coutts; J. Cocchi; M. Hauschildt; K. Cornett; A. Rao; C. Garber; D. De Vivo

P.87

Shortening treatment initiation timeframe through prenatal/neonatal diagnosis of spinal muscular atrophy

B. Gross; S. Matesanz; E. Kichula

P.88

In-home body-weight support maximizes function after treatment in young children with spinal muscular atrophy

M. Iammarino; B. Powers; N. Miller; K. Shannon; L. Alfano; L. Lowes

P.89

Scoliosis in Spinal muscular atrophy type I in the nusinersen era

F. Al Amrani; R. Amin; J. Chiang; J. Boyd; J. Vajsar; J. Dowling; H. Gonorazky

P.90

Trunk involvement in spinal muscular atrophy type 2 and 3

R. Muni-Lofra; G. Coratti; D. Ramsey; D. Moat; J. Sodhi; M. James; A. D'amico; M. Scoto; M. Pane; E. Bertini; C. Marini-Bettolo; F. Muntoni; E. Mercuri; A. Mayhew

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SMA carrier screening testing by droplet digital PCR reveals occurrence of 5 copy numbers of SMN1, substantiating potential 3+0 silent carrier status

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Comparing motor and respiratory function in SMA Type1 treated with Nusinersen using CHOP INTEND(CHOP) & Great Ormond Street Respiratory Score (GSR)

L. Edel; L. Abbott; E. Chan; M. Main; V. Robinson; P. Munot; A. Manzur; D. Ridout; G. Baranello; M. Scoto; F. Muntoni

Late Breaking Posters - (LBP 1–14)

LBP 1

From mouse- to patient-relevant models: Using CRISPR activation to upregulate a compensatory disease-modifying gene in LAMA2-CMD

Annie Arockiaraj, Jia Qi Cheng-Zhang, Salah Daghlas, Caleb Kim, Dwi Kemaladewi

LBP 2

COVID-19 Myopathy: Persistence of Viral Particles in the Skeletal Muscle

Jian-Qiang Lu, Katerina Gordon, Dubravka Dodig

LBP 3

Dominant mutations in ITPR3 cause Charcot-Marie-Tooth disease

Julius Rönkkö, Svetlana Molchanova, Anya Revah-Politi, Elaine Pereira, Mari Auranen, Jussi Toppila, Jouni Kvist, Anastasia Ludwig, Julika Neumann, Stephanie Humblet-Baron, Geert Bultynck, Adrian Liston, Anders Patau, Claudio Rivera, Matthew Harms, Henna Tyynismaa, Emil Ylikallio

LBP 4

Novel Epigenetic Small Molecule Approaches and Single-Cell Epigenetic Analysis for DMD

Lisa Maves, Jean-Baptiste Dupont, Shawn Luttrell, Melanie Morris, Arianna Gomez, Thao Pham, David Mack

LBP 5

HOPE-2 one-year results show clinically relevant improvements in upper limb & cardiac function in patients with later stage Duchenne Muscular Dystrophy

Linda Marban, Sigfried Rogy, Craig McDonald, Michelle Eagle, Richard Finkel, Cuixia Tian, Michael Taylor, Joanne Janas, Matthew Harmelink, Arun Varadhachary, Kan Hor, Oscar Henry Mayer, Pat Furlong, HOPE-2 Steering Committee

LBP 6

PRIME editing permits to introduce specific point mutations in the gene coding for dystrophin

Cedric Happi Mbakam, Joel Rousseau, Antoine Guyon, Guillaume Tremblay, Francis-Gabriel Bégin, Jacques P. Tremblay

LBP 7

Characterization of Novel Exon-51 Skipping Oligonucleotides that Safely Rescues Dystrophin Expression in a Severe Model of Duchenne Muscular Dystrophy

Charles O'Neill, Maria Hedlund, Sundeep Chandra, M Benjamin Hock, Andrew Melton, Todd Oppeneer, Ganesh Cherala, Mika Aoyagi-Scharber, Ashley Frazer-Abel, Nicole Datson, Judith van Deutekom, Jukka Poulivali, Xiefan Fang, Sylvia Fong, David Jacoby, Shripad Bhagwat

LBP 8

A canine model for understanding the pharmacokinetic/pharmacodynamic relationship of muscle-targeted therapies

Robert W Grange, Eva R Chin, Jordan Klaiman, Darren Hwee, Bradley Morgan, Fady Malik, Martin Childers, SiWei Luo, David Mack

LBP 9

Constitutive Stim1 activation impairs myogenesis in Tubular Aggregate Myopathy: defective late differentiation of patient-derived myoblasts and potential druggable targets

Elena Conte, Giulia Maria Camerino, Alessandra Pannunzio, Mauro Coluccia1, Marina Mora, Lorenzo Maggi, Ornella Cappellari, Paola Imbrici, Annamaria De Luca, Antonella Liantonio

LBP 10

ACTN3 genotype influences skeletal muscle mass regulation and the response to dexamethasone
Jane Seto, Kelly Roeszler, Lyra Meehan, Harrison Wood, Chrystal Tiong, Lucinda Bek, Cheryl Lee, Paul Gregorevic, Peter Houweling, Professor Kathryn North

LBP 11

Congenital Muscular Dystrophy Type 1D (α -Dystroglycanopathy) in a Family of Labrador Retrievers with a Stop Codon Mutation in LARGE1 Gene

G. Diane Shelton, Kate M. Minor, Ling T. Guo, Steven G. Friedenberg, Jonah N. Cullen, Jeffrey M. Hord, David Venzke, Mary E. Anderson, Megan Devereaux, Caryl Handelman, Kevin P. Campbell, James R. Mickelson

LBP 12

Syndecan-4 Modulates Cell Polarity and Migration by Influencing Centrosome Positioning and Intracellular Calcium Distribution

Daniel Becsky, Kitti Szabo, Szuzina Gyulai-Nagy, Tamas Gajdos, Zsuzsa Bartos, Arpad Balind, Laszlo Dux, Peter Horvath, Miklos Erdelyi, Laszlo Homolya, Dr. Aniko Keller-Pinter

LBP 13

rAAVrh.74.MHCK7.micro-dystrophin gene therapy in children with Duchenne muscular dystrophy: A quantitative MR biomarker study

Rebecca J Willcocks, Sean C Forbes, Donovan J Lott, Claudia R Senesac, Alison M Barnard, Kelly J Lehman, Carrie E Nease, Kathleen Church, Zarife Sahenk, H Lee Sweeney, Louise R Rodino-Klapac, Glenn A Walter, Jerry R Mendell, Krista Vandeborne

LBP 14

High-fidelity modelling of skeletal muscle laminopathies using LMNA-mutant human iPS cells and bioengineered muscles for mutation-specific therapy development

Luca Pinton, Heather Steele-Stallard, Daniel Moore, Shilpita Sarcar, Tanel Ozdemir, Sara Martina Maffioletti, Jean-Marie Cuisset, Gisèle Bonne, Peter Steven Zammit, Francesco Saverio Tedesco

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Phenotyping by NMR of a novel dog model of centronuclear myopathy

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Clinical and genetic characterization of RYR1-related myopathy in a reference neuromuscular center in Chile

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H. Zhong; M. Yu; P. Lin; Z. Zhao; J. Xi; W. Zhu; Y. Zheng; W. Zhang; H. Lv; C. Yan; J. Hu; Z. Wang; J. Lu; Y. Yuan; S. Luo

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Correlations between quantitative NMRI biomarkers for disease progression and disease activity in muscle of patients with dysferlinopathy

H. Reygoudt; F. Smith; E. Caldas de Almeida Araujo; I. Wilson; R. Fernandez-Torron; M. James; U. Moore; B. Marty; L. Rufibach; H. Hilsden; H. Sutherland; J. Hogrel; T. Stojkovic; K. Bushby; V. Straub; P. Carlier; A. Blamire; Jain Foundation COS consortium

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Development of clinical trial simulation tools for Duchenne muscular dystrophy using magnetic resonance biomarkers

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Estimating lean thigh muscle volume using multifrequency serial bioelectrical impedance in patients with muscle atrophy and fatty degeneration

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Muscle MRI as biomarker of disease progression in Becker muscular dystrophy: a 2 year follow-up study

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Broadening fibroadipose replacement pattern in patients with sarcoglycanopathies: towards whole body MRI approaches

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Association between the progression of muscle fat replacement and muscle architecture in Duchenne muscular dystrophy

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Baseline plasma phosphorylated neurofilament heavy chain level predicts sitting in nusinersen-treated individuals with infantile-onset SMA

F. Muntoni; C. Sumner; T. Crawford; R. Finkel; E. Mercuri; Y. Liu; M. Petrillo; B. Kandinov; W. Farwell

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SMARtCARE: Real-world-data collection of patients with spinal muscular atrophy

A. Pechmann; G. Bernert; T. Hagenacker; W. Müller-Felber; U. Schara; I. Schwersenz; M. Walter; H. Lochmüller; J. Kirschner

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L. Servais; J. Day; D. De Vivo; J. Kirschner; E. Mercuri; F. Muntoni; P. Shieh; E. Tizzano; I. Desguerre; S. Quijano-Roy; K. Saito; M. Droege; O. Dabbous; A. Shah; F. Anderson; R. Finkel

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The Canadian neuromuscular disease registry: A national spinal muscular atrophy (SMA) registry for real world evidence

V. Hodgkinson-Brechenmacher; M. Oskoui; C. Campbell; J. Lounsberry; B. Brais; A. MacKenzie; H. McMillan; J. Vajsar; L. Korngut; C. CNDR Investigator Network

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Emerging real-world use of nusinersen in adult patients with spinal muscular atrophy (SMA) in the US: A multi-site chart review study

A. Paradis; T. Cochrane; A. Khachatryan; M. Martinez

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Longitudinal change over time on Timed Function Tests (TFTs) in ambulatory persons with SMA

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Efficacy evaluation of Nusinersen for spinal muscular atrophy type I and type II using bioelectrical impedance (BIA)

K. Ishiguro; T. Sato; M. Shichiji; K. Yuki; T. Murakami; N. Taniguchi; S. Nagata; K. Ishigaki

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Development of motor function and changes in NFL in CSF in children with SMA treated with nusinersen

M. Tulinius; L. Alberg; I. Henriksson; L. Wahlgren; A. Kroksmark; I. Thuestad; K. Jahnke; E. Eklund; K. Blennow; H. Zetterberg

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Identification of functional SMNs and suppressors of SMA causing mutations to define the critical function of SMN that causes SMA

A. Burghes; A. Blatnik III; C. Ruhno; V. McGovern; T. Le; V. Pessino; S. Driscoll; C. Iyer; K. Corlett; S. Likhite; B. Kaspar; S. Pfaff; U. Fischer

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Improvements in the definition of biomarkers for Spinal Muscular Atrophy (SMA) type III and IV: a multimodal longitudinal study

G. Querin; J. Hogrel; R. Debs; V. Marchand-Pauvert; T. Stojkovic; A. Behin; P. Lafôret; F. Salachas; P. Bede; P. Pradat; T. Lenglet

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ActiMyo®: Upper limb activity in non-ambulant patients with spinal muscular atrophy treated with Spinraza

C. Lilien; M. Annoussamy; M. Polleur; A. Seferian; O. Boespflug-Tanguy; K. Gorni; D. Eggenpieler; L. Servais

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Patient reported health-related quality of life in pediatric patients with spinal muscular atrophy type 1, 2 and 3

S. Wagner; B. Wong; J. Lambert; P. Horn; J. Bange; I. Rybalsky; C. Tian

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Disease and treatment burden of spinal muscular atrophy (SMA) on patients and caregivers in Canada

H. McMillan; B. Gerber; T. Cowling; W. Khuu; M. Mayer; J. Wu; B. Maturi; K. Klein-Panneton; C. Cabalteja; H. Lochmuller

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A population-based study examining the epidemiologic burden, healthcare resource utilization

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G. Chen; B. Sharif; B. Gerber; M. Farris; T. Cowling; C. Cabalteja; J. Wu; B. Maturi; K. Klein-Panneton; J. Mah

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"Registre- SMA France": a national registry of patients with spinal muscular atrophy (SMA)

M. Gomez-Garcia de la Banda; L. Grimaldi; J. Urtizberea; A. Behin; C. Vuillerot; P. Saugier-veber; F. Audic; C. Barnerias; C. Cances; E. Campana-Salort; C. Spil; P. Laforet; V. Laugel; Y. Pereon; S. Sacconi; T. Stojkovic; C. Tard; B. Chabrol; I. Desguerre; S. Quijano-Roy

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Systematic literature review of the economic burden and economic evaluations in spinal muscular atrophy

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ATEND: Development of a wheelchair based motor assessment

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Diverse cohort of spinraza-treated spinal muscular atrophy patients at Mayo Clinic Rochester for theranostic biomarker discovery

S. Cook; N. Folch; L. Hasadsri; D. Oglesbee; N. Staff; D. Anderson; D. Haile; D. Selcen

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Investigating temporal changes in percent predicted FVC and RULM score in non-ambulant SMA type III children

A. Wolfe; M. Scoto; E. Milev; R. Muni Lofra; A. Rohwer; R. Wake; A. Mayhew; C. Marini-Bettolo; F. Muntoni

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Preliminary data for the cost-effectiveness assessment of the newborn screening for SMA in Belgium.

T. Dangouloff; L. Servais; M. Hiligsmann

Late Submitted Posters (LSP 1-26)

LSP 1

Characterization of R-DMDdel52, a preclinical rat model of Duchenne muscular dystrophy

Valentina Taglietti, Kaouthar Kéfi, Mathilde Rodrigues, Baptiste Périou, Bernadette Drayton, Peggy Lafuste, Frédéric Relaix

LSP 2

Persistent COUP-TFII Expression Underlies the Myopathy and Impaired Muscle Regeneration Observed in Resistance to Thyroid Hormone-Alpha

Paola Aguiari, Yan Yun Liu, Astgik Petrosyan, Sheue-yann Cheng, Gregory A Brent, Laura Perin, Anna Milanese

LSP 3

CRISPR-Cas9 mediated tagging allows the detection of endogenous Gne in mice muscles and muscle cells

Avi Harazi, Nili Ilouz, Lena Yakovlev, Stella Mitrani-Rosenbaum

LSP 4

The complex role of syndecan-4 in skeletal muscle: myogenesis and oncogenesis

Kitti Szabo, Daniel Varga, Attila Gergely Vegh, Ning Liu, Lin Xu, Laszlo Dux, Miklos Erdelyi, Aniko Keller-Pinter

LSP 5

The effect of mutation in EMD gene on DNA-damage response and chromatin organization in cells from EDMD1 patients

Magdalena Machowska, Claudia Bearzi, Katarzyna Piekarowicz, Giovanna Lattanzi, Ryszard Rzepecki

LSP 6

Hierarchical Bayesian model of disease progression in centronuclear myopathy allows to demonstrate treatment efficacy with a small sample size

Arnaud Monseur, Bruno Boulanger, Leen Thielemans, Chris Freitag, Carlin Brad, Seferian M. Andreea, Laurent Servais, Nathis MTM study group

LSP 7

Clinical and genetic aspects of DNMT2-related centronuclear myopathy; a retrospective, medical chart review to establish natural history

Asli Aykanat, Stéphanie Hoffmann, Casie Genetti, Khazal Paradis, Heather Paterson, Sundos Al-Husayni, Beiyu Lin, Leen Thielemans, Chris Freitag, Alan Beggs

LSP 8

Nusinersen treatment in SMA patients during COVID-19 pandemic

Karolina Aragon-Gawinska, Anna Potulska-Chromik, Anna Lusakowska, Anna Fraczek, Anna Kostera-Pruszczyk

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Israeli SMA Registry 2020: Capturing real-life data

Aviva Fattal-Valevski, Irina E. Opincariu, Anat Weizman, Liora Sagie

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Role of Syndecan-4 in myoblast metabolism and mitochondrial functions

Zoltán Márton Köhler, László Juhász, Tamás Gajdos, Miklós Erdélyi, György Trencsényi, László Dux, Anikó Keller-Pintér

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SRF-mediated mechanotransduction is required for the response to exercise in both cancer patients and animal models

Medhi Hassani, Alezandra Baccam, Alexandra Benoni, Caterina Gargano, Gabriela Salim de Castro, Joana Alves, Sara Chiappalupi, Viviana Moresi, Sergio Adamo, Francesca Riuzzi, Guglielmo Sorci, Maurizio Muscaritoli, Marilia Seelaender, Zhigang Xue, Zhenlin Li, Onnik Agbulut, Dario Coletti

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Central core disease series disclosing pathogenicity and genetic inheritance of RYR1 variants

Ana Cotta, Lucas Santos Souza, Elmano Carvalho, Leticia Nogueira Feitosa, Antonio Lopes da-Cunha-Junior, Monica Machado Navarro, Julia Filardi Paim, Jaquelin Valicek, Miriam Melo Menezes, Simone Vilela Nunes, Rafael Xavier-Neto, Antonio Pedro Vargas, Reinaldo Issao Takata, Mariz Vainzof

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Spinal Muscular Atrophy-III (SMA-III) Assessment in the Context of International Classification of Functioning, Disability and Health (ICF-DH): A Case Series

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8 years of experience in diagnosing hereditary neuromuscular disorders in Voronezh (Russia)

Sergei Aleksandrovich Kurbatov

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Nusinersen – Significant Results in Early Initiation in Spinal Muscular Atrophy Type II

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Lysosomal degradation of GMPPB is associated with limb-girdle muscular dystrophy type 2T
Wotu Tian, Xinghua Luan, Li Cao

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Hereditary motor-sensory neuropathy and girdle-limb muscular dystrophy in the Moscow region
Olga Sidorova, Sergey Kotov, Elena Borodataya, Mark Bunak, Alexander Polyakov, Yuriy Filyushkin

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Evaluation of Children with Neuromuscular Diseases in terms of Life Quality
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New phenotype of DCTN1-related spectrum: early-onset dHMN plus congenital foot deformity
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Role of Stimulated single fiber electromyography (SSFEMG) in early diagnosis of Lambert-Eaton myasthenic syndrome (LEMS)
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Emery-Dreifuss muscular dystrophy caused by a mutation in the lamin A/C gene identified by exome sequencing: case report from Indonesia
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Poster session 3

Congenital muscular dystrophies (P.192-203)

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Collagen VI-related myopathy. Clinical and genetic findings in a large Chilean Cohort
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Effective pseudo-exon skipping of a COL6A1 intronic mutation in cultured muscle interstitial fibroblasts from a novel humanized mouse model
V. Bolduc; F. Guirguis; J. Cheng; L. Garrett; C. Bönnemann

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Increasing allele selectivity of small interfering RNAs to target a dominant-negative glycine substitution causing a collagen VI-related dystrophy
A. Sarathy; V. Bolduc; C. Bönnemann

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TRAPPC11-related muscular dystrophy with hypoglycosylation of alpha-dystroglycan in skeletal muscle and brain
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Long-term motor function and pulmonary function in COL6-related dystrophies are associated with the maximal motor ability achieved
D. Natera-de Benito; A. Reghan-Foley; C. Domínguez-González; C. Ortez; M. Jain; A. Mebrahtu; S. Donkervoort; Y. Hu; M. Fink; P. Yun; T. Ogata; J. Medina; J. Díaz-Manera; L. Carrera-García; J. Expósito-Escudero; M. Olivé; J. Colomer; C. Jiménez-Mallebrera; C. Bönnemann; A. Nascimento

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D. Natera-de Benito; I. Zaharieva; V. Pini; A. Manzur; P. Munot; S. Parker DiTroia; S. Di Gioia; J. Jurgens; B. Barry; E. England; D. Ledoux; A. O'Donnell-Luria; D. MacArthur; L. Feng; R. Phadke; A. Sarkozy; E. Engle; F. Muntoni

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Patient-derived induced pluripotent stem cells differentiated cardiomyocytes as platform for disease modelling For X-linked dilated cardiomyopathy
S. Zhu; M. Deng; A. Law; E. Poon; J. Lo; R. Liang; A. Kwong; K. Ng; H. Tse; G. Chan; S. Chan

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G. Cooper-Olson; R. Potter; L. Rodino-Klapac

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N. Wein; T. Simmons; D. Rajakumar; D. Lesman; D. Li; C. Gaffney; R. Rafferty; N. Huang; Y. Rodriguez; C. Young; M. Spencer; K. Flanigan

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Inhibition of NF- κ B signaling by edasalonexent prevents the development of DMD-associated cardiomyopathy in mdx:Utrn \pm mice

J. Huang; T. Gemelli; P. Bista; T. Tassin; X. Jiang; J. Shelton; A. Nichols; J. Donovan; P. Mammen

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Src tyrosine kinase as potential target in Duchenne muscular dystrophy: assessment of a novel dasatinib formulation in the mdx mouse model

F. Sanarica; P. Mantuano; B. Boccanegra; O. Cappellari; E. Conte; G. Camerino; A. Cutrignelli; N. Denora; A. Mele; M. De Bellis; A. De Luca

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Systemic delivery of PPMO restores the full-length dystrophin protein in the Dup2 Mouse
L. Gushchina; K. Grounds; E. Frair; N. Huang; F. Schnell; G. Hanson; T. Simmons; N. Wein; K. Flanigan

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L. Gushchina; E. Frair; N. Rohan; A. Bradley; T. Simmons; H. Chavan; M. Waldrop; N. Wein; K. Flanigan

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CRISPR-Cas9 genome editing rescues dystrophin expression in a dog DMD model with a mutation in the N-terminal mutation hotspot

R. Maruyama; K. Lim; Q. Nguyen; M. Tsoumpra; S. Takeda; Y. Aoki; T. Yokota

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A novel in-frame deletion of exons 52-55 DMD mouse model preserves muscle function

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Antisense oligonucleotide-mediated knockdown of IGFBP3 to increase IGF-1 signaling in dystrophic muscle

A. Yavas; M. van Putten; E. Niks; A. Aartsma-Rus

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Targeted genome editing in vivo corrects a Dmd duplication restoring wild-type dystrophin expression

E. Maino; D. Wojtal; S. Evagelou; A. Farheen; T. Wong; K. Lindsay; O. Scott; A. Chiodo; M. Schneeweiss; M. Rok; E. Ivakine; R. Cohn

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Skeletal and cardiac muscle dysfunction in the D2-mdx mouse model of Duchenne muscular dystrophy: a comparison with BL10-mdx mice using TREAT-NMD SOPs

P. Mantuano; F. Sanarica; A. Mele; O. Cappellari; B. Boccanegra; E. Conte; M. De Bellis; G. Camerino; A. De Luca

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A homozygous nonsense variant in LRIF1 associated with facioscapulohumeral muscular dystrophy

D. Sikrova; K. Hamanaka; S. Mitsuhashi; H. Masuda; Y. Sekiguchi; A. Sugiyama; K. Shibuya; R. Lemmers; R. Goossens; M. Ogawa; K. Nagao; C. Obuse; S. Noguchi; Y. Hayashi; S. Kuwabara; J. Balog; I. Nishino; S. van der Maarel

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CRISPR-Cas9 mediated removal of an intronic SMCHD1 mutation suppresses DUX4 expression in FSHD myocytes in vitro

R. Goossens; M. van den Boogaard; R. Lemmers; J. Balog; P. van der Vliet; I. Willemsen; J. Schouten; I. Maggio; N. van der Stoep; R. Hoeben; S. Tapscott; N. Geijsen; M. Gonçalves; S. Sacconi; R. Tawil; S. van der Maarel

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A Phase 2, randomized, placebo-controlled, 24-Week study of the efficacy and safety of losmapimod in treating subjects with FSHD: ReDUX4

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A. Mikhail; M. Tarnopolsky

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Spectrum and impact on quality of life of gastrointestinal disorders in myotonic dystrophy type 1

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Survey of patient care situations for congenital/childhood onset myotonic dystrophy in Japan -- Nationwide patient survey

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Rasch analysis of the North Star Assessment for limb-girdle-type muscular dystrophies across subtypes

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Restorable workspace: A conceptual 3D upper extremity physical function measure for muscle strength and joint contractures

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The influence of limb girdle muscle weakness on gait parameters: a pilot study across 8 subtypes

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Expansion of normative reference values for the 100 meter timed test for broad use across neuromuscular diseases

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Clinical Development of SRK-015, a Fully Human Anti-proMyostatin Monoclonal Antibody, for the Treatment of Later-Onset Spinal Muscular Atrophy

A. Place; D. Barrett; S. Cote; G. Nomikos; R. Iarrobino; C. Yung

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Nusinersen in adolescents and young adults with SMA: Longitudinal experience from an expanded cohort of CS2/CS12 and SHINE participants

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One-time administration of AVXS-101 intrathecal (IT) for spinal muscular atrophy in the phase 1 study (STRONG): safety report

D. Chand; R. Finkel; J. Day; B. Darris; N. Kuntz; A. Connolly; C. Zaidman; T. Crawford; R. Butterfield; P. Shieh; G. Tennekoon; J. Brandesma; S. Iannaccone; M. Meriggioli; S. Tauscher-Wisniewski; J. Shoffner; F. Ogrinc; S. Kavanagh; D. Feltner; J. Mendell

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Intravenous (IV) onasemnogene abeparvovec for spinal muscular atrophy (SMA): integrated safety report

D. Chand; R. Finkel; E. Mercuri; R. Masson; J. Parsons; A. Kleyn; M. Menier; K. Montgomery; D. Sproule; S. Reyna; D. Feltner; S. Tauscher-Wisniewski; J. Mendell

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Longer-term treatment with nusinersen: Results in later-onset spinal muscular atrophy from the SHINE study

E. Mercuri; B. Darras; C. Chiriboga; M. Farrar; J. Kirschner; N. Kuntz; G. Acsadi; M. Tulinius; J. Montes; G. Gambino; R. Foster; D. Ramirez-Schrempp; J. Wong; B. Kandinov; W. Farwell

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Onasemnogene aveparvovec gene therapy for spinal muscular atrophy type 1 (SMA1): Phase 3 study update (STRIVE-EU)

E. Mercuri; G. Baranello; R. Masson; O. Boespflug-Tanguy; C. Bruno; S. Corti; A. Daron; N. Deconinck; M. Scoto; L. Servais; V. Straub; F. Ogrinc; H. Ouyang; D. Sproule; S. Reyna; S. Tauscher-Wisniewski; F. Baldinetti; D. Chand; D. Feltner; A. Lavrov; F. Muntoni

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FIREFISH Part 1: 24-month safety and exploratory outcomes of risdiplam (RG7916) in infants with Type 1 spinal muscular atrophy (SMA)

G. Baranello; O. Bloespflug-Tanguy; B. Darras; J. Day; N. Deconinck; A. Klein; R. Masson; E. Mercuri; A. Dodman; M. El-Khairi; M. Gerber; K. Gorni; H. Kletzl; R. Scalco; L. Servais

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Onasemnogene abeparvovec gene therapy in presymptomatic spinal muscular atrophy (SMA): SPRINT study update

K. Strauss; M. Farrar; K. Swoboda; K. Saito; C. Chiriboga; R. Finkel; S. Iannaccone; J. Krueger; J. Kwon; H. McMillan; L. Servais; J. Mendell; J. Parsons; M. Scoto; P. Shieh; C. Zaidman; M. Schultz; F. Ogrinc; S. Group; F. Muntoni

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Long-term follow-up of onasemnogene abeparvovec gene therapy in spinal muscular atrophy type 1 (SMA1)

J. Mendell; R. Shell; K. Lehman; M. McColly; L. Lowes; L. Alfano; N. Miller; M. Iammarino; K. Church; F. Ogrinc; H. Ouyang; E. Kernbauer; S. Joshi; D. Sproule; M. Meriggioli; D. Feltner; S. Al-Zaidy

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Value of onasemnogene abeparvovec in spinal muscular atrophy type 1: improvements in motor function, ventilation-free survival, and hospitalizations

O. Dabbous; S. Reyna; D. Feltner; F. Ogrinc; M. Menier; H. Ouyang; M. Droege; M. Bischof; N. LaMarca; R. Arunji

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SUNFISH Part 1: 24-month safety and exploratory outcomes of risdiplam (RG7916) treatment in patients with Type 2 or 3 spinal muscular atrophy (SMA)

J. Day; G. Baranello; O. Boespflug-Tanguy; S. Borell; N. Goemans; J. Kirschner; R. Masson; M. Pera; L. Servais; S. Fuhrer; M. Gerber; K. Gorni; H. Kletzl; C. Martin; R. Scalco; H. Staunton; W. Yeung; E. Mercuri

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Longer-term effects of nusinersen on motor function outcomes based on age at treatment initiation

R. Finkel; J. Kirschner; E. Mercuri; D. De Vivo; E. Bertini; R. Foster; G. Gambino; D. Ramirez-Schrempp; R. Chin; B. Kandinov; W. Farwell

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Escalating dose and randomized, controlled study of high-dose nusinersen in SMA; study design and updated enrollment for the DEVOTE Study

R. Finkel; J. Day; M. Ryan; E. Mercuri; D. De Vivo; S. Pascual Pascual; J. Montes; J. Gurgel-Giannetti; N. Mitchell-Sweeney; R. Foster; P. Sun; D. Ramirez-Schrempp; B. Kandinov; W. Farwell

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Nusinersen in infantile-onset spinal muscular atrophy: results from longer-term treatment from the open-label SHINE extension study

R. Finkel; D. Castro; M. Farrar; M. Tulinius; K. Krosschell; K. Saito; G. Gambino; R. Foster; D. Ramirez-Schrempp; J. Wong; B. Kandinov; W. Farwell

P.267

Minimally invasive fusionless surgery offers satisfactory early and definitive treatment for scoliosis in SMA children: Our experience over a decade

M. Gaume; E. Saudeau; M. Gomez Garcia de la Banda; V. Azzi-Salameh; C. Barnerias; A. Benezit; I. Dabaj; A. Essid; C. Gitiaux; I. Haegy; B. Mbieleu; R. Sauvagnac; D. Verollet; R. Carlier; J.

Bergounioux; I. Desguerre; V. Topouchian; S. Quijano-Roy; L. Miladi; C. Glorion

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Nusinersen effect in infants who initiate treatment in a presymptomatic stage of SMA: NURTURE results

T. Crawford; M. Ryan; J. Kirschner; R. Finkel; K. Swoboda; D. De Vivo; E. Bertini; W. Hwu; R. Foster; D. Ramirez-Schrempp; R. Chin; W. Farwell

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Impact of Continued Nusinersin treatment on Caregiver Experience and Health-Related Quality of Life in Later-onset SMA: Results From the SHINE Study

J. Montes; D. Krasinski; R. Foster; G. Gambino; J. Wong; B. Kandinov; A. Paradis; N. Johnson

P.270

Myostatin dynamics in health and disease: Pharmacologic effects of SRK-015, a highly selective monoclonal antibody inhibitor of myostatin activation

K. Long; S. Cote; S. Wawersik; S. Study Group

P.271

FIREFISH Parts 1 and 2: 12-month pooled safety and efficacy outcomes of risdiplam (RG7916) in infants with Type 1 spinal muscular atrophy (SMA)

L. Servais; O. Bloespflug-Tanguy; B. Darras; J. Day; N. Deconinck; A. Klein; R. Masson; M. Mazurkiewicz-Beldzińska; E. Mercuri; K. Rose; D. Vlodayets; H. Xiong; E. Zanoteli; A. Dodman; M. El-Khairi; M. Gerber; K. Gorni; H. Kletzl; R. Scalco; G. Baranello

P.273

Jewelfish: Safety and pharmacodynamic data in non-naïve patients with spinal muscular atrophy receiving treatment with risdiplam (RG7916)

C. Chiriboga; C. Bruno; T. Duong; D. Fischer; J. Kirschner; E. Mercuri; M. Gerber; K. Gorni; H. Kletzl; T. McIver; R. Scalco; F. Warren; M. Scoto

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RAINBOWFISH: A study of risdiplam (RG7916) in infants with presymptomatic spinal muscular atrophy (SMA)

L. Servais; E. Bertini; M. Al-Muhaizea; L. Nelson; A. Pruffer; D. Vlodayets; Y. Wang; E. Zanoteli; L. Burke; M. El-Khairi; K. Gorni; H. Kletzl; M. Gerber; R. Scalco; R. Finkel

P.276

Monitoring Chilean SMA patients treated with nusinersen. A single-center experience

C. Castiglioni; M. Martinez-Jalilie; M. Diemer; G. Calcagno; C. Hervias; J. Jofre; B. Suarez; M. Palomino; S. Lillo; M. Haro; E. Muñoz; A. Chahin

P.278

Use of motor function measure-20 (MFM-20) to monitor SMA type 1 and 2 patients under nusinersen

L. Le Goff; A. Seferian; A. Phelep; P. Rippert; M. Mathieu; C. Cances; C. de Lattre; J. Durigneux; G. Gousse; S. Quijano-Roy; C. Sarret; L. Servais; C. Vuillerot

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Efficacy and safety of nusinersen treated adult patients with spinal muscular atrophy (SMA) types 2-3-4

B. De Wel; K. Claeys

Poster session 4

DMD - Therapy (P.280-294)

P.280

Systemic gene transfer with rAAVrh74.MHCK7.micro-dystrophin in patients with Duchenne muscular dystrophy

J. Mendell; Z. Sahenk; K. Lehman; C. Nease; L. Lowes; N. Miller; M. Iammarino; L. Alfano; J. Vaiea; S. Al-Zaidy; S. Lewis; K. Church; R. Shell; R. Potter; D. Griffin; E. Pozsgai; M. Hogan; L. Rodino-Klapac

P.281

Investigating the effect of DMD non-sequential splicing on exon skipping strategies

R. Goossens; N. Verwey; F. Schnell; A. Aartsma-Rus

P.282

Edasalonexent treatment in young boys with Duchenne muscular dystrophy is associated with age-normative growth and normal adrenal function

E. Finanger; R. Finkel; G. Tennekoon; K. Vandenborne; L. Sweeney; P. Shieh; S. Yum; M. Mancini; J. MacDougall; J. Donovan

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Long-term safety and efficacy of golodirsen in male patients with Duchenne muscular dystrophy amenable to Exon 53 skipping

F. Muntoni; L. Servais; V. Straub; M. Guglieri; A. Dugar; M. Whalen-Kielback; D. Steiner; E. Koenig; T. Feng; X. Wang; E. Mercuri

P.284

ATL1102 treatment improves PUL2.0 in non-ambulant boys with Duchenne muscular dystrophy compared to a natural history control

G. Tachas; N. Desem; P. Button; G. Coratti; M. Pane; E. Mercuri; M. Mercuri

P.285

In non-ambulatory DMD patients, one-year treatment benefit of idebenone in respiratory function extrapolates to 3-year delay in start of assisted vent

G. Buyse; T. Voit; C. McDonald; H. Gordish-Dressman; E. Henricson; T. Serjesen; G. Bernert; M. D'Angelo; M. Leinonen

P.286

In the global phase 3 polarisDMD trial for edasalonexent, standardized outcome measure training produces excellent test-retest variability in NSAA

M. Eagle; J. MacDougall; M. Mancini; J. Donovan

P.287

Accelerating the development of personalized DMD gene therapies

M. Rok; E. Ivakine; R. Cohn

P.288

Casimersen treatment in eligible patients with Duchenne muscular dystrophy: Safety, tolerability, and pharmacokinetics over 144 weeks of treatment

N. Kuntz; K. Wagner; L. East; S. Upadhyay; B. Han; E. Koenig; D. Steiner; P. Shieh

P.289

Open-label evaluation of eteplirsen in males with DMD amenable to exon 51 Skipping: PROMOVI

E. Koenig; P. Shieh; H. Abdel-Hamid; A. Connolly; C. McDonald; D. Steiner; J. Malhotra; N. Khan; W. Hu; B. Han; E. Ciafaloni

P.290

Delay in Duchenne muscular dystrophy progression with eteplirsen: attenuation of pulmonary decline and projected freedom from continuous ventilation

J. Iff; C. Gerrits; E. Birk; E. Tuttle; Y. Zheng; E. Henricson; C. McDonald

P.291

Real-world evidence of eteplirsen treatment effects on Duchenne muscular dystrophy related health outcomes using claims data in the United States

J. Iff; E. Tuttle; C. Gerrits; D. Gupta; Y. Zhong

P.292

Ataluren delays loss of ambulation and decline in pulmonary function in patients with nonsense mutation Duchenne muscular dystrophy

C. McDonald; F. Muntoni; M. Rance; J. McIntosh; J. Jiang; A. Kristensen; V. Penematsa; F. Bibbiani; E. Goodwin; H. Gordish-Dressman; L. Morgenroth; P. Trifillis; M. Souza; M. Tulinus

P.293

Safety and efficacy of teriparatide treatment for severe osteoporosis in patients with Duchenne muscular dystrophy

N. Nasomyont; C. Keefe; C. Tian; L. Hornung; J. Khoury; J. Tilden; P. Hochwalt; E. Jackson; I. Rybalsky; B. Wong; M. Rutter

P.294

Does steroid therapy influence pulmonary function decline in adults with Duchenne muscular dystrophy after loss of ambulation?

A. Pietrusz; R. Astin; G. Ramdharry; R. Quinlivan; M. Desikan

Autoimmune myopathies (P.295-312)

P.295

Analysis of treatment and outcome of 81 patients with idiopathic inflammatory myopathy

N. Eura; A. Yamanaka; T. Shiota; Y. Nishimori; N. Iguchi; M. Ozaki; H. Nanaura; N. Iwasa; T. Kiriya; T. Izumi; H. Kataoka; K. Sugie

P.296

The ProDERM study: A randomized, double-blinded, placebo controlled trial of IVIG (Octagam 10%) in patients with dermatomyositis

R. Aggarwal; C. Charles-Schoeman; J. Schessl; Z. Bata-Csorgo; M. Dimachkie; Z. Griger; S. Moiseev; C. Oddis; E. Schiopu; J. Vencovsky; I. Beckmann; E. Clodi; T. Levine; ProDERM Investigators

P.297

A case of Juvenile HMGCR antibody myositis presenting as limb girdle

N. Chrestian; N. Rioux; J. Proulx-Gauthier; B. Ellezam; Y. Labrie; S. Rivest; B. Lace

P.298

Anti-HMGCR myopathy overlaps with dermatomyositis: A distinct subtype of IIM

Y. Hou; K. Shao; Y. Yan; T. Dai; W. Li; Y. Zhao; G. Norman; C. Yan

P.299

Distinct disease-activity markers between inflammatory and dystrophic myopathies revealed by the multi-exponential behaviour of water T2-relaxation

E. Caldas de Almeida Araujo; B. Marty; P. Carlier; P. Baudin; H. Reyngoudt

P.300

TSP-1 mediates muscle damage in brachio-cervical inflammatory myopathy and systemic sclerosis

X. Suárez-Calvet; J. Alonso-Pérez; A. Carrasco-Rozas; E. Fernández-Simón; P. Piñol-Jurado; I. Castellvi; C. Zamora; L. Martínez-Martínez; A. Alonso-Jiménez; D. Castillo; E. Gallardo; I. Illa; J. Díaz-Manera

P.301

Optimizing hand-function patient outcome measures for inclusion body myositis

A. Lin; C. Siener; A. Faino; M. Seiffert; C. Wehl; L. Wang

P.302

Autoantibodies targeting membrane repair proteins represent a novel pathologic mechanism in idiopathic inflammatory myopathy

K. McElhanon; N. Young; J. Hampton; B. Paleo; T. Kwiatkowski; E. Beck; A. Capati; K. Jablonski; T. Gurney; M. Lopez; R. Aggarwal; C. Oddis; W. Jarjour; N. Weisleder

P.303

Mitochondrial DNA rearrangements and point mutations in inclusion body myositis

A. Oldfors; C. Hedberg-Oldfors; S. Basu; U. Lindgren; C. Lindberg; E. Larsson; M. Falkenberg

P.305

Clinical onset of sporadic inclusion body myositis after years of immunosuppressive therapy

M. Winkler; C. von Landenberg; C. Kornblum; J. Reimann

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Clinical correlates of KLRG1+ T cells in inclusion body myositis

N. Goyal; S. Greenberg; J. Cauchi; N. Araujo; V. Li; M. Wencel; T. Irani; L. Wang; G. Coulis; A. Villalta; T. Mozaffar

P.309

Immune mediated necrotising myopathy: a treatable condition missed in the Arab and African regions

R. El Sherif; M. Gamal; I. Nishino

P.310

Myopathy in scleroderma/systemic sclerosis patients: From a histopathological point of view

Y. Chen; M. Inoue; M. Ogasawara; Y. Saito; L. Indrawati; J. Tanboon; T. Kumutpongpanich; M. Okubo; W. Yohioka; S. Hayashi; S. Noguchi; I. Nishino

P.311

Myostatin in inflammatory idiopathic myopathies: a marker of activity and muscle recovery?

A. Mahoudeau; S. Maillard; C. Anquetil; N. Tawara; D. Amelin; Y. Allenbach; O. Benveniste

P.312

"Amyopathic" MDA5-positive dermatomyositis with severe lung involvement presents with myositic morphological features

B. Englert; W. Stenzel

Mitochondrial diseases & Metabolic myopathies (P.313-324)

P.313

Design of a phase 3 prospective clinical study for the treatment of thymidine kinase 2 deficiency (TK2d)

Q. Abu Ali; C. Domínguez-González; B. Cohen; A. Berardo; R. Haas; C. Konersman; R. McFarland; G. D'Souza; J. Quan; B. Thompson; M. Hirano

P.314

Mitochondrial DNA mutant load in A3243G mutation and clinical correlation

H. Lee; Y. Lee

P.315

Collaborative model for diagnosis and treatment of very rare diseases: experience in Spain with thymidine kinase 2 deficiency

C. Paradas; C. Domínguez-González; M. Madruga-Garrido; M. Hirano; I. Martí; F. Munell; A. Nascimento; M. Olivé; J. Quan; D. Sardina; R. Martí

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MT1621 for thymidine kinase 2 deficiency (TK2d): mechanism of action is via mitochondrial DNA incorporation

L. Tsuruda; D. Kenny; C. Blázquez-Bermejo; A. Karlsson; R. Martí; Y. Cámara

P.317

A novel m.9143T>C variant -- intercurrent severe lactate acidosis and immunodeficiency as an extension of the phenotypic spectrum in MT-ATP6 mutations

D. Urban; L. Scholle; M. Wagner; A. Ludolph; A. Rosenbohm

P.318

RBCK1-related disease: A rare multisystem disorder with polyglucosan storage, autoinflammation, recurrent infections, skeletal, and cardiac myopathy

R. Phadke; C. Hedberg-Oldfors; R. Scalco; D. Lowe; M. Ashworth; M. Novelli; R. Vara; A. Merwick; H. Amer; R. Sofat; M. Sugarman; A. Jovanovic; M. Roberts; V. Nakou; A. King; I. Bodi; H. Jungbluth; A. Oldfors; E. Murphy

P.320

Analysis of morbidity and mortality in untreated patients with thymidine kinase 2 deficiency

C. Domínguez-González; C. Garone; G. D'Souza; B. Thompson; M. Morton; J. Quan; M. Hirano

P.321

Mitochondrial myopathy in X-linked creatine transporter deficiency due to a novel mutation of SLC6A8 gene

M. Bisciglia; A. Busson; M. Marangoni; P. David; I. Vandernoot; A. Michotte; G. Remiche

P.322

Defects in mucopolipin-1 (mucopolipidosis type IV) cause dystrophic changes in human muscle

A. Zambon; A. Lemaigre; R. Phadke; S. Grunewald; A. Sarkozy; E. Clement; F. Muntoni

P.323

A genetic basis is identified in 74% cases of paediatric hyperCKaemia without weakness

W. Wong; S. Bryen; A. Bournazos; S. Bommireddipall; L. Waddell; M. Menezes; R. Webster; M. Davis; C. Liang; S. Cooper; K. Jones

P.324

Characterization of mitochondrial respiratory chain complexes in reversible infantile respiratory chain deficiency

S. Roos; C. Hedberg-Oldfors; K. Visuttijai; G. Kollberg; C. Lindberg; N. Darin; A. Oldfors

New genes and diseases / NGS & related techniques (P.326-334)

P.326

Whole exome sequencing identifies two novel candidate genes and extends the diagnostic spectrum of patients with neuromuscular diseases

R. Rossi; M. Falzarano; M. Pinotti; D. Balestra; M. Neri; F. Fortunato; E. Mercuri; M. Pane; F. Gualandi; R. Selvatici; A. Ferlini

P.327

Whole exome and whole genome sequencing for the genetic diagnosis of dystrophinopathies

R. Selvatici; M. Fang; M. Falzarano; F. Gualandi; S. Delin; S. Bensemmane; A. Shatillo; L. Bello; E. Pegoraro; A. Ferlini

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Neuromuscular disease variant of unknown significance (VUS) resolution using muscle biopsy evaluation: The Iowa experience

K. Jones; S. Moore

P.329

Whole exome sequencing in the pediatric neuromuscular clinic

A. Meyer; C. Cottrell; S. Reshmi; R. Pfau; K. Lee; M. Mathew; D. Corsmeier; V. Jayaraman; A. Dave-Wala; S. Hashimoto; T. Matthews; D. Mouhla; M. Stein; M. Waldrop; K. Flanigan

P.330

An approach to prioritizing novel disease gene candidates and investigating their role in muscle biology and disease

H. Goulee; J. Clayton; R. Taylor; N. Laing; G. Ravenscroft; A. Forrest

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RNA-Seq is useful in research of neuromuscular disorders

M. Johari; M. Savarese; P. Jonson; S. Koivunen; B. Udd; P. Hackman

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Role of whole exome sequencing in identifying rare genetic variants in a cohort of patients presenting with congenital myopathy

A. Aykanat; C. Genetti; W. Win; Z. Valivullah; E. O'Heir; B. Darras; R. Laine; A. O'Donnell-Luria; A. Beggs

P.333

Progressive proximal muscle weakness, myopathic muscle biopsy and a novel candidate gene

S. Puusepp; T. Reimand; S. Pajusalu; C. Bruels; C. Bönemann; K. Chao; S. Coppens; S. Donkervoort; J. Goodrich; P. Kang; P. Mohassel; L. Pais; T. Siddique; D. Vargas-Franco; M. Wojcik; W. Stenzel; K. Ounap

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Introne identifies non-canonical splice-altering variants in neuromuscular patients resulting in multiple new genetic diagnoses

P. Sullivan; C. Mayoh; M. Wong-Erasmus; V. Gayevskiy; S. Beecroft; M. Pinese; E. Oates; M. Cowley

Registries, Care, Quality of Life, management of NMD (P.335-349)

P.335

Cardiac involvement in Duchenne and Becker muscular dystrophy

G. Öz Tunçer; I. Sahin; Ü. Akça; A. Aksoy

P.336

Novel and more sensitive criteria for identifying chronic respiratory failure in progressive neuromuscular disease

O. Mayer

P.337

Variability of cardiac function in patients with Duchenne muscular dystrophy experiencing sudden onset chest pain and electrocardiographic changes

A. Yamamoto; H. Komaki; K. Segawa; Y. Shimizu-Motohashi; E. Takeshita; A. Ishiyama; N. Sumitomo; E. Nakagawa; M. Sasaki

P.338

The "Hospital goes to School": A psychoeducational program to raise awareness about neuromuscular disorders in schools to prevent social exclusion

I. Zschaek; A. Colomer; D. Natera De Benito; L. Carrera; J. Exposito; C. Ortez; J. Colomer; A. Nascimento

P.340

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.341

Use of the assessment of caregiver experience with neuromuscular disease (ACEND with SMA) - a caregiver experience from a single center

L. Brown; K. Hoffman; K. Krosschell; C. Weigel; C. Blomgren; H. Munson; J. Bidwell; N. Kuntz; V. Rao

P.342

Life expectancy and causes of death in patients with Duchenne muscular dystrophy

L. Wahlgren; A. Kroksmark; M. Tulinius; K. Sofou

P.343

Cardiac involvement in Fukuyama muscular dystrophy is less severe than in Duchenne muscular dystrophy

M. Taniguchi-Ikeda; T. Yamamoto; H. Awano; K. Iijima; T. Toda

P.344

The rare disease cures accelerator- Data and analytics platform: Value for drug development in muscle diseases

J. Larkindale; V. Boulanger; P. Gavin; R. Liwski; K. Romero; M. Campbell

P.345

Rasch analysis of the pediatric quality of life inventory 4.0 generic core scales administered to patients with Duchenne muscular dystrophy

E. Landfeldt; J. Iff; E. Henricson

P.346

Child-to-adult healthcare transition for inherited muscle diseases: a single-center study in suburbs of Tokyo, Japan

K. Ogata; T. Murakami; K. Yatabe; M. Suzuki; I. Nonaka; T. Tamura

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Illness identity in young adults with neuromuscular disorders

S. Geuens; K. Leyen; J. Willen; V. Maenen; J. Lemiere; N. Goemans; L. De Waele; K. Luyckx

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NMD4C: A new neuromuscular disease network for Canada

K. Amburgey; J. Dowling; J. Warman Chardon; R. Kothary; B. Stead-Coyle; B. Brais; C. Campbell; C. Gagnon; H. McMillan; K. Selby; L. Korngut; M. Oskoui; R. Amin; P. Esler; N. Worsfold; T. Buffone; D. Wojtal; H. Osman; H. Lochmuller

P.349

Subspecialty healthcare utilization in pediatric patients with muscular dystrophy in the United States

S. Matesanz; J. Edelson; K. Iacobellis; E. Mejia; J. Brandsema; C. Wittlieb-Weber; H. Griffis; O. Okunowo; K. Lin

19:30-20:30

From the spinal cord to the muscle - Selected oral presentations 3 (O.9-12)

(4 x 10 mins consecutive presentations plus 20 mins live Q&A)

Molecular and clinical assessments of spinal cord disease, sarcoglycanopathy, and dysferlinopathy are discussed, along with genetic therapy for the latter.

Session Moderator: John Vissing

Assistant Moderator: Mo Zhao

O.09

Spinal cord MRI for early detection of presymptomatic pathology in c9orf72 mutation carriers: A longitudinal neuroimaging study

G. Querin; P. Bede; M. El Mendili; M. Péllegri-Issac; D. Rinaldi; M. Catala; D. Saracino; F. Salachas; A. Camuzat; V. Marchand-Pauvert; J. Cohen-Adad; O. Colliot; I. Le Ber; P. Pradat; x. PREV-DEMALS Study Group

O.10

European collaboration on the clinical and genetic spectrum of sarcoglycan-deficient muscular dystrophy

J. Alonso-Pérez; L. González-Quereda; C. Semplicini; P. Gallano; E. Pegoraro; A. Nascimento; C. Ortez; M. Devisser; A. Van der Krooi; C. Garrido; M. Santos; M. Guglieri; V. Straub; U. Schara; S. Sarcoglycan Study group; J. Díaz-Manera

O.11

Limb girdle muscular dystrophy R2 and Miyoshi myopathy are not distinct clinical phenotypes in dysferlinopathy

U. Moore; H. Gordish; J. Díaz Maneraz; M. James; A. Mayhew; M. Guglieri; S. Spuler; J. Day; K. Jones; D. Bharucha-Goebel; E. Salort-Campana; A. Pestronk; M. Walter; C. Paradas; T. Stojkovic; M.

Yoshimura; E. Bravver; E. Pegoraro; J. Mendell; V. Straub

O.12

Midi-Dysferlin gene therapy for dysferlinopathies

O. Ballouhey; S. Courier; M. Krahn; N. Levy; M. Bartoli

20:30-21:00

Comfort break, poster viewing, sponsors & exhibiton, and chat room

21:00-22:00

Highlights across Myology - Selected oral presentations 4 (O.13-16)

(4 x 10 mins consecutive presentations plus 20 mins live Q&A)

From basic science to clinical research, investigators use a variety of approaches to assess NM disease, including calcium studies, gene editing, large-scale electron microscopy, and pre-conception carrier screening.

Session Moderator: Mariz Vainzof

Assistant Moderator: Nicolas Chrestian

O.13

Altered calcium handling in a zebrafish model of SELENON congenital muscular dystrophy

M.Wright

O.14

Gene editing using CRISPR/CAS9 in an animal model of nemaline myopathy

N. Sabha; H. Gonorazky; S. Khattak; S. Viththiyapaskaran; N. Bhambra; H. Granzier; J. Dowling

O.15

Large-scale electron microscopy reveals capillary pathology in muscle samples of patients with systemic sclerosis

C. Dittmayer; E. Siegert; A. Uruha; H. Goebel; W. Stenzel

O.16

Results of a preconception carrier screening trial including severe neuromuscular disorders

S. Edwards; R. Ong; M. Davis; R. Allcock; G. Androga; B. Kamien; K. Harrop; G. Ravenscroft; M. Fietz; N. Pachter; J. Beilby; N. Laing

Friday, 2 October 2020

15:00-15:45

Poster highlights (PH1-6)

(6 x 5 mins consecutive presentations plus 15 mins live Q&A)

This is one of the most popular sessions of the annual WMS Congress. The session moderators, with support from the programme committee, select 6 posters for short oral presentations that they feel are of particular interest to the neuromuscular community. The poster may address controversial or polarising topics or topics that aren't regularly covered in plenary lectures, but are nevertheless scientifically sound and relevant.

Session Moderators: Nicol Voermans and Ichizo Nishino

15:45-16:15

Comfort break, poster viewing, sponsors & exhibition, and chat room

16:15-17:00

Late Breaking News Session (LBO 1-3)

(3 x 10 mins consecutive presentations plus 15 mins live Q&A)

One of the most exciting sessions of the annual WMS Congress, this sessions provides a platform for still unpublished, high quality and high impact research findings that are of great interest to all congress participants. Presentations often focus on new disease causes, pathomechanisms and therapies.

Session Moderators: Laurent Servais and Kevin Flanigan

LBO 1

Pathogenic variants in TNNC2 cause congenital myopathy due to an impaired force response to

calcium

Martijn van de Locht, Sandra Donkervoort, Josine de Winter, Stefan Conijn, Benno Kusters, Ying Hu, Reghan Foley, Gwimoon Seo, Darren Hwee, Thomas Irving, Weikang Ma, Henk Granzier, Kalyan Immadisetty, Peter Kekenes-Huskey, José Pinto, Nicol Voermans, Carsten Bönnemann, Coen Ottenheijm

LBO 2

A novel form of muscular dystrophy associated with mutations in JAG2

Sandra Coppens, Sanna Puusepp, Katrin Õunap, Alison Barnard, Sandra Donkervoort, Reza Maroofian, Henry Houlden, Nicolas Deconinck, Fowzan Alkuraya, Hessa Alsaif, Erica Macke, Anna Lusakowska, Andreas Hahn, Volker Straub, Ana Töpf, Catheline Vilain, Marie Rivera-Zengotita, Carsten Bönnemann, Glenn Walter, Peter Kang

LBO 3

Expression of apparent full-length dystrophin in skeletal muscle in a first-in-human gene therapy trial using the scAAV9.U7-ACCA vector

Dr. Megan Waldrop, Dr. Michael Lawlor, Dr. Tatyana Meyers Vetter, Emma Frair, Margaret Beatka, Dr. Hui Meng, Megan Iammarino, Brenna Powers, Johan Harris, Maryann Kaler, Dr. Tabatha Simmons, Dr. Nico Wein, Dr. Kevin Flanigan

17:00-17:15

Comfort break, poster viewing, sponsors & exhibition

17:15-18:15

Prize presentations: *Nicol Voermans*

Close of conference: *Volker Straub*

Introduce 2021 and presentation of flag: *Jana Haberlová*