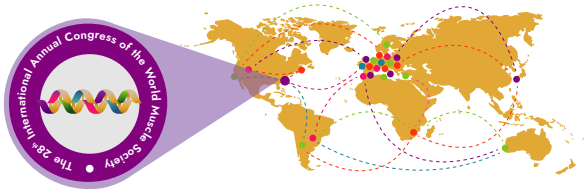

WMS2023
3rd-7th October 2023 Charleston, USA
The 28th Annual Congress of the World Muscle Society

Programme Key:

I	**Invited Speaker**
O/LBO	**Selected Oral Presentation/Late Breaking Selected Oral Presentation**
P/LBP	**Poster Presentation/Late Breaking Poster Presentation** (on display at the venue and on the virtual platform)
VP/LBVP	**Virtual Poster Presentation/ Late Breaking Virtual Poster Presentation** (on display on the virtual platform and on ePoster boards at the venue)

Please note, all times stated in the programme are in Local Charleston, SC, USA (EST) time.

WMS 2023 Full Programme

Tuesday 3rd October 2023

15:30-18:00	**Registration** 📍 Ballroom Foyer, **refreshments** 📍 PAC Foyer and Exhibit Hall **and poster set up** 📍 Ballroom Foyer	
16:30-17:30	**Industry Symposium 1** 📍 PAC	**Industry Symposium 2** 📍 Exhibit Hall A1
18:00-18:45	**Opening Ceremony** 📍 PAC *Moderators: Lindsay Alfano & Chris Weihl* INV01: The strength to explore: a review of NASA experience with muscle atrophy in space ***Thomas D***[1] [1]*Ohio Astronaut*	
18:45-21:00	Networking Reception 📍 Congress Venue (separate registration required)	

Wednesday 4th October 2023

06:30-19:30	**Registration desk open**	
07:30-08:30	**Industry Symposium 3** 📍 PAC	**Industry Symposium 4** 📍 Exhibit Hall A1
09:00-09:15	**Congress Welcome** 📍 PAC Message from the President	
09:15-10:45	📍 PAC **Topic 1: Understanding phenotypic and genetic diversity in neuromuscular disorders 1** *Moderators: Gina Ravenscroft, University of Western Australia, Australia & Marco Savarese, University of Helsinki, Finland*	
09:15-09:45	INV02: High throughput functional assays to improve interpretation of rare variants discovered in Neuromuscular disease genes ***Lek M***[1] [1]*Yale School of Medicine*	
09:45-10:15	INV03: Understanding genetic variants in neuromuscular disorders ***Weihl C***[1] [1]*Washington University in St. Louis*	

10:15-10:30	**O01: Clinical spectrum and molecular features of asymptomatic and paucisymptomatic DMD mutations** ***Nicolau S***[1], *Meyer A*[1], *Vetter T*[1,2], *Lowes L*[1], *Alfano L*[1], *Reash N*[1], *Iammarino M*[1], *Frair E*[1], *Tsao C*[1,2,3], *Connolly A*[1,2,3], *Mendell J*[1,2,3], *Waldrop M*[1,2,3], *Flanigan K*[1,2,3] *[1]Center for Gene Therapy, Nationwide Children's Hospital, [2]Department of Pediatrics, The Ohio State University, [3]Department of Neurology, The Ohio State University*
10:30-10:45	**O02: Unpacking gene expression profile to the single nuclei level in humam muscle Pompe samples** ***Diaz-Manera J***[1], *Monceau A*[1], *Gokul-Nath R*[1], *Musumeci O*[2], *Toscano A*[2], *Papadimas G*[3], *Kierdaszuk B*[4], *Kostera-Pruszczyk A*[4], *Paradas C*[5], *Rivas-Infante E*[5], *Dominguez C*[6], *Hernandez-Lain A*[6], *Lileker J*[7], *Roberts M*[7], *Suarez-Calvet X*[8] *[1]Newcastle University, [2]Ospedalle Universitario G. Martino, [3]University of Athens, [4]Warszawski Uniwersytet Medyczny, [5]Hospital Virgen del Rocio, [6]Hospital 12 de Octubre, [7]Manchester Salford Hospital, [8]Hospital de la Santa Creu i Sant Pau*
10:45-11:15	**Morning refreshments & exhibition** 📍 Exhibit Hall **and posters** 📍 Ballroom
10:45-11:15	**Social Media Committee** find out about how to get involved 📍 Myology Café, Exhibit Hall
11:15-13:15	📍 PAC **Topic 1: Understanding phenotypic and genetic diversity in neuromuscular disorders 2** *Moderators: Gisèle Bonne, Centre de Recherche en Myologie, France & Charlotte Lilien, MDUK Oxford Neuromuscular Centre, UK*
11:15-11:45	**INV04: Accounting for phenotypic variability in clinical outcome assessments** ***Alfano L***[1] *[1]The Abigail Wexner Research Institute at Nationwide Children's Hospital*
11:45-12:15	**INV05: Engaging patients from diverse backgrounds in NMD research** ***Ramdharry G***[1,2] *[1]UCL Queen Square, Institute of Neurology, [2]National Hospital for Neurology and Neurosurgery, UCLH NHS Trust*
12:15-12:30	**O03: In vivo gene therapy for striated muscle laminopathy** ***Okubo M***[1], *Brull A*[1], *Beuvin M*[1], *Mougenot N*[1], *Paradis V*[2], *Bonne G*[1], *Bertrand A*[1] *[1]Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, [2]Dpt d'Anatomie Pathologique Hôpital Beaujon*
12:30-12:45	**O04: Myoguide.org: a web-based portal supporting the analysis of MRIs for the diagnosis of neuromuscular patients** *Bolaño Diaz C*[1], *Verdu Diaz J*[1], *Gonzalez Chamorro A*[1], *Veeranki G*[1], *MYO-Share working group*[2], *Llauger J*[3], *Diaz Manera J*[1,4,5] *[1]John Walton Muscular Dystrophy Research Centre, [2]MYO-MRI, [3]Radiology Department, Hospital Universitari de la Santa Creu i Sant Pau, [4]Laboratori de Malalties Neuromusculars, Insitut de Recerca de l'Hospital de la Santa Creu i Sant Pau de Barcelona, [5]Centro de Investigación Biomédica en Red en Enfermedades Raras (CIBERER)*
12:45-13:00	**O05: TDP-43 dependent cryptic exon derived neoepitopes as a novel diagnostic biomarker in muscle biopsies of inclusion body myositis patients** ***Ikenaga C***[1], *Wilson A*[1], *Mallika A*[2], *Sinha I*[2,3], *Burns G*[2], *Ling J*[2], *Corse A*[1], *Wong P*[2,4], *Lloyd T*[1,3,4] *[1]Department of Neurology, Johns Hopkins University School of Medicine, [2]Department of Pathology, Johns Hopkins University School of Medicine, [3]Department of Neuroscience, Johns Hopkins University School of Medicine, [4]Indicates equal contribution*
13:00-13:15	**O06: Clinical trial readiness and validation of onsite and remote evaluation in valosin containing protein-associated multisystem proteinopathy: A 24-month longitudinal study** ***Reash N***[1], *Iammarino M*[1], *Pietruszewski L*[1], *Lowes L*[1,2], *Mendell J*[1,2,3], *Connolly A*[1,2,3], *Adderley K*[1], *Peck N*[4], *Peck A*[4], *Alfano L*[1,2] *[1]Center for Gene Therapy, Nationwide Children's Hospital, [2]Department of Paediatrics, The Ohio State University, [3]Department of Neurology, The Ohio State University, [4]Cure VCP Disease*
13:15-14:30	**Lunch & exhibition** 📍 Exhibit Hall **and posters** 📍 Ballroom
13:45-14:15	**New WMS Members Event** 📍 Myology Café, Exhibit Hall
14:30-15:30	**Poster session 1** 📍 Ballroom A-C *Refreshments served*
	P41-P42, VP43, P44-P47, VP48, P49-P54, VP55: Clinical trial highlights **P41 EMBARK, a Phase 3 trial evaluating safety and efficacy of delandistrogene moxeparvovec in DMD: study design and baseline characteristics** *Muntoni F*[1], *Mercuri E*[2], *Schara-Schmidt U*[3], *Komaki H*[4], *Richardson J*[5], *Singh T*[5], *Guridi M*[6], ***Mason S***[5], *Murphy A*[6], *Yu L*[5], *Reid C*[7], *Darton E*[5], *Wandel C*[6], *Mendell J*[8,9] *[1]The Dubowitz Neuromuscular Centre, NIHR Great Ormond Street Hospital Biomedical Research Centre, Great Ormond Street Institute of Child Health University College London, & Great Ormond Street Hospital Trust, [2]Paediatric Neurology Institute, Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS, [3]Department of Paediatric Neurology, Center for Neuromuscular Disorders in Children and Adolescents, University Clinic Essen, University of Duisburg-Essen, [4]Translational Medical Center, National Center of Neurology and Psychiatry, [5]Sarepta Therapeutics, Inc., [6]F. Hoffmann-La Roche Ltd, [7]Roche Products Ltd, [8]Center for Gene Therapy, Nationwide Children's Hospital, [9]The Ohio State University*

P42 Practical considerations for delandistrogene moxeparvovec gene therapy in patients with Duchenne muscular dystrophy

*Mendell J[1,2], **Proud C**[3], Zaidman C[4], Mason S[5], Darton E[5], Wandel C[6], Murphy A[6], Mercuri E[7], Muntoni F[8], McDonald C[9]*
[1]Center for Gene Therapy, Nationwide Children's Hospital, [2]The Ohio State University, [3]Children's Hospital of the King's Daughters, [4]Department of Neurology, WUSTL, [5]Sarepta Therapeutics, Inc., [6]F. Hoffmann-La Roche Ltd, [7]Pediatric Neurology Institute, Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS, [8]The Dubowitz Neuromuscular Centre, NIHR Great Ormond Street Hospital Biomedical Research Centre, Great Ormond Street Institute of Child Health University College London, & Great Ormond Street Hospital Trust, [9]UC Davis Health

VP43 Phase 1/2 trial evaluating AOC 1044 in healthy volunteers and participants with DMD mutations amenable to exon 44 Skipping: EXPLORE44 Trial Design

***Stahl M**[1], Ackermann E[1], Chen C[1], Zhu Y[1], Cho H[1], Hughes S[1], DiTrapani K[1], Laverty C[2], McDonald C[3]*
[1]Avidity Biosciences, Inc., [2]UCSD, Rady Children's Hospital, and VA San Diego Healthcare System, [3]UC Davis Health Medical Center

P44 Phase 1 study of PGN-EDO51 demonstrates tolerability, delivery and high levels of exon skipping for treatment of Duchenne muscular dystrophy (DMD)

***Larkindale J**[1], Lonkar P[1], Goyal J[1], Holland A[1], Foy J[1], Garg B[1], Yu S[1], Frank A[1], Abbott C[1], Svenstrup N[1], Cormier J[1], Vacca S[1], Mellion M[1]*
[1]PepGen

P45 A phase 1/2 study of DYNE-251 in males with DMD mutations amenable to exon 51 skipping: DELIVER study design

***Naylor M**[1], Mix C[1], Han B[1], Dugar A[1]*
[1]Dyne Therapeutics

P46 ENVOL, a Phase 2, open-label trial evaluating the safety and expression of delandistrogene moxeparvovec in Duchenne muscular dystrophy: study design

***Mercuri E**[1], Desguerre I[2], Gangfuss A[3], Servais L[4,5,6], Nascimento A[7], Zhang B[8], Murphy A[9], Reid C[10], Wandel C[9], Singh T[11], Guridi M[9], Muntoni F[12]*
[1]Pediatric Neurology Institute, Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS, [2]Departments of Pediatric Neurology and Medical Genetics, Hospital Necker-Enfants Malades, Université Paris Cité, [3]Department of Paediatric Neurology, Center for Neuromuscular Disorders in Children and Adolescents, Center for Translational Neuro- and Behavioral Sciences, University Clinic Essen, University of Duisburg-Essen, [4]MDUK Oxford Neuromuscular Centre, Department of Paediatrics, University of Oxford, [5]Division of Child Neurology, Centre de Références des Maladies Neuromusculaires, Department of Pediatrics, University Hospital Liège & University of Liège, [6]Motion Institut de Myologie AP-HP, Hôpital Armand Trousseau, [7]Neuromuscular Unit, Neuropaediatrics Department, Hospital Sant Joan de Déu, Fundacion Sant Joan de Déu, CIBERER – ISC III, [8]F. Hoffmann-La Roche Ltd, [9]F. Hoffmann-La Roche Ltd, [10]Roche Products Ltd, [11]Sarepta Therapeutics, Inc., [12]The Dubowitz Neuromuscular Centre, NIHR Great Ormond Street Hospital Biomedical Research Centre, Great Ormond Street Institute of Child Health University College London, & Great Ormond Street Hospital Trust

P47 ENVISION, a phase 3, randomized trial evaluating the safety and efficacy of delandistrogene moxeparvovec in Duchenne muscular dystrophy: study design

*Muntoni F[1], **Mercuri E**, McDonald C[3], Desguerre I[4], Tulinius M[5], Proud C[6], Furgerson M[7], Murphy A[8], De Ford C[8], Feng T[7], Reid C[9], Wandel C[8], Shelton N[7]*
[1]The Dubowitz Neuromuscular Centre, NIHR Great Ormond Street Hospital Biomedical Research Centre, Great Ormond Street Institute of Child Health University College London, & Great Ormond Street Hospital Trust, [2]Paediatric Neurology Institute, Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS, [3]UC Davis Health, [4]Departments of Paediatric Neurology and Medical Genetics, Hospital Necker Enfants Malades, Université Paris Cité, [5]Department of Paediatrics Institute of Clinical Sciences, Sahlgrenska Academy, University of Gothenburg, [6]Children's Hospital of the King's Daughters, [7]Sarepta Therapeutics, Inc., [8]F. Hoffmann-La Roche Ltd, [9]Roche Products Ltd

VP48 AOC 1001 demonstrates DMPK reduction and spliceopathy improvement in a phase 1/2 study in myotonic dystrophy type 1 (DM1) (MARINA)

***Zhu Y**[1], Kwan T[1], Meng Q[1], Tai L[1], Cho H[1], Lee M[1], Younis H[1], Levin A[1], Flanagan M[1]*
[1]Avidity Biosciences, Inc.

P49 The efficacy and safety of Tideglusib in a randomized, placebo-controlled, double blind study in children and adolescents with congenital myotonic dystrophy (REACH CDM study)

***Horrigan J**[1], Snape M[1], Fantelli E[1]*
[1]AMO Pharma Ltd

P50 A phase 1/2 randomized, placebo-controlled, multiple ascending dose study (ACHIEVE) of DYNE-101 in individuals with myotonic dystrophy type 1 (DM1)

***Wolf D**[1], Mix C[1], Han B[1], Dugar A[1], Farwell W[1]*
[1]Dyne Therapeutics

P51 Phase 1/2 study to evaluate AOC 1020 for adult patients with Facioscapulohumeral muscular dystrophy: FORTITUDE trial design

***Halseth A**[1], Ackermann E[1], Brandt T[1], Chen C[1], Cho H[1], Stahl M[1], DiTrapani K[1], Hughes S[1], Tawil R[2], Statland J[3]*
[1]Avidity Biosciences, Inc., [2]University of Rochester Medical Center, [3]University of Kansas Medical Center

P52 Experiences of parents/caregivers of children in the ASPIRO X-Linked Myotubular Myopathy (XLMTM) gene therapy clinical trial: A qualitative study

***Juando-prats C**[1], Hodwitz K[1], Kenneally N[2], Alfano L[3], Sarazen M[4], Coats J[4]*
[1]Applied Health Research Centre, Li Ka Shing Knowledge Institute, St. Michael's Hospital, Unity Health Toronto, [2]Early Childhood Curriculum Studies, Department of Human Services and Early Learning, MacEwan University, [3]Center for Gene Therapy, Nationwide Children's Hospital, [4]Astellas Gene Therapies

P53 Inclusion body myositis treatment with Celution processed adipose derived regenerative cells

*Heim A[1], Soder R[1], Bhavsar D[1], Ciersdorff A[1], Pasnoor M[1], Jawdat O[1], Jabari D[1], Farmakidis C[1], Chandrashekhar S[1], **Dimachkie M**[1]*
[1]University of Kansas Medical Center

P54 T-cell response to SRP-9001 dystrophin transgene in a patient treated with Delandistrogene Moxeparvovec: a case of immune-mediated myositis

*Khan S[1], Haegel H[2], Hollenstein A[2], Wandel C[2], Wagner K[3], Asher D[1], Griffin D[1], **Potter R**[1], Moeller I[1], Singh T[1], Rodino-Klapac L[1]*
[1]Sarepta Therapeutics Inc, [2]F. Hoffmann-La Roche Ltd, [3]Pharma Development Neurology, F. Hoffmann-La Roche Ltd

VP55 Topline data analysis of the phase 1/2 clinical trial evaluating AOC 1001 in adult Patients with Myotonic dystrophy type 1: MARINA

*Johnson N[1], Day J[2], Hamel J[3], Thornton C[3], Subramony S[4], Soltanzadeh P[5], Statland J[6], Wicklund M[7], Arnold W[8], Freimer M[8], DiTrapani K[9], Heusner C[9], Chen C[9], Cho H[1], McEvoy B[9], Zhu Y[9], **Tai L**[9], Ackermann E[9]*
[1]Virginia Commonwealth University, [2]Stanford University Medical Center, [3]University of Rochester, [4]University of Florida, [5]University of California, Los Angeles, [6]University of Kansas Medical Center, [7]University of Colorado, [8]The Ohio State University, [9]Avidity Biosciences, Inc.

P56-P57, P59-P67: Therapies for neuromuscular disorders

P56 Preliminary study of anti-AAVrh74 seroprevalence following gene transfer

D'Ambrosio E[1], *Tong L[1], Ozes Ak B[1], Lehman K[1], Sahenk Z[1], Mendell J[1]*
[1]Nationwide Children's Hospital

P57 ORAI1 inhibition as a preclinical therapy for tubular aggregate myopathy (TAM) and Stormorken syndrome (STRMK)

*Silva-Rojas R[1], Pérez-Guàrdia L[1], Simon A[1], Djeddi S[1], Treves S, Laporte J[1], **Bohm J**[1]*
[1]IGBMC

P59 Generation and characterization of a novel XMEA mouse model and pharmacological evaluation of autophagy antagonists

Karuppasamy M[1], *English K[1], Sanders V[1], Lopez M[1,2], Kaur G[3], Worthey L[3], Huang L[4,5], Dowling J[4,5,6,7], Alexander M[1,2,8,9,10]*
[1]Division of Neurology, Department of Pediatrics, University of Alabama at Birmingham and Children's of Alabama, [2]Department of Genetics, University of Alabama at Birmingham, [3]Center for Computational Genomics and Data Science at Children's of Alabama, [4]Program for Genetics and Genome Biology, Hospital for Sick Children, [5]Division of Neurology, Hospital for Sick Children, [6]Departments of Molecular Genetics, University of Toronto, [7]Department of Pediatrics, University of Toronto, [8]UAB Center for Exercise Medicine, University of Alabama at Birmingham, [9]Civitan International Research Center, University of Alabama at Birmingham, [10]UAB Center for Neurodegeneration and Experimental Therapeutics (CNET))

P60 Developing a decision-making framework for expanded access to gene therapy in rare neuromuscular diseases

Lawrence C[1]
[1]Bionical Emas

P61 High dose localized muscle irradiation: Hedgehog pathway as a new therapeutic target

Rota Graziosi E[1], *François S[1,2], Pâteux J[1], Gauthier M[1], Drouet M[1,2], Riccobono D[1,2], Jullien N[1]*
[1]Armed Forces Biomedical Research Institute, [2]INSERM Unit UMR1296 "Radiations: Defense, Health, Environment

P62 SIMPATHIC: accelerating drug repurposing for rare neurological, neurometabolic and neuromuscular disorders by exploiting SIMilarities in clinical and molecular PATHology

't Hoen P[1], *Benkemoun L[2], Prigione A[3], Boussaad I[4], de Kort M[5], Geille A[6], Lochmüller H[7], Voermans N[1], van Engelen B[1], van Karnebeek C[8]*
[1]Radboud University Medical Center, [2]Foundation for Rare Diseases, [3]Heinrich Heine Universität, [4]University of Luxemburg, [5]EATRIS ERIC, [6]Euro-DyMA, [7]Children's Hospital of Eastern Ontario Research Institute, [8]Academic Medical Centers Amsterdam

P63 PCSK9 inhibitor is available for muscular disease patients without muscular adverse events

Kurashige T[1], *Murao T[1], Katsumata R[1], Kanaya Y[1], Dodo Y[1], Sugiura T[1], Ohshita T[1]*
[1]Nho Kure Medical Center and Chugoku Cancer Center

P64 An activin type II receptor ligand trap prevented loss of cortical bone strength and cancellous bone mass in a mouse model of severe disuse osteopenia

*F Poulsen M[1], Fisher F[2], Lachey J[2], Seehra J[2], Andersen C[1], Eijken M[3], Thomsen J[1], Brüel A[1], **Lodberg A**[1]*
[1]Department of Biomedicine, Aarhus University, [2]Keros Therapeutics, [3]Department of Renal Medicine, Aarhus University Hospital

P65 Development of therapeutic extracellular vesicle enveloped-AAV vectors for muscle gene therapy

*Kauffman J[1], **Saad N**[1,2]*
[1]Center for Gene Therapy, The Abigail Wexner Research Institute at Nationwide Children's Hospital, [2]Department of Paediatrics, The Ohio State University

P66 Salbutamol therapy in a neuromuscular cohort

*Nigro E[1], **Amburgey K**[1], Djordjevic D[1], Alawneh I[1], Gonorazky H[1], Dowling J[1]*
[1]Hospital for Sick Children

P67 Novel therapeutic approaches in inherited neuropathies: a systematic review

Hustinx M[1,2], Shorrocks A[1], Servais L[1,3]
[1]MDUK Oxford Neuromuscular centre, [2]Centre de Référence des Maladies Neuromusculaires, Department of Neurology, University Hospital Liège, [3]Centre de Référence des Maladies Neuromusculaires, Department of Paediatrics, University Hospital Liège

VP114-VP116, P117, VP118-VP119, P120-P126: Muscle MRI

VP114 A new coronal view-based muscle MRI in the evaluation of patients with Myopathy

Lee G[1], Huang H[2], Chao C[1], Yang C[1], Shih T[2], Hsieh S[1], **Hsueh H**[1]
[1]Department of Neurology, National Taiwan University Hospital, [2]Department of Medical Imaging, National Taiwan University Hospital

VP115 Implementing new metrics for a deeper understanding of muscle imaging patterns

Gomez Andres D[1], Costa Comellas L[1], Quijano-Roy S[2], Munell F[1]
[1]H.U. Vall d'Hebron, [2]H. Raymond Poincaré

VP116 Whole-body muscle magnetic resonance imaging (MRI) in PAX7-congenital myopathy (CM)

Haliloğlu G[1], Donkervoort S[2], Öz Yildız S[1], Hu Y[2], Pais L[3], Koşukcu C[4], Aydıngöz Ü[5], Bönnemann C[2]
[1]Hacettepe University Faculty of Medicine, Department of Paediatrics, Division of Paediatric Neurology, [2]Neuromuscular and Neurogenetic Disorders of Childhood Section, Neurogenetics Branch, National Institute of Neurological Disorders and Stroke, National Institutes of Health, [3]Center for Mendelian Genomics, Program in Medical and Population Genetics, Broad Institute of MIT and Harvard, [4]Hacettepe University, Institute of Health Sciences, Department of Bioinformatics, [5]Hacettepe University Faculty of Medicine, Department of Radiology

P117 Congenital recessive TTN myopathy: MRI findings in 6 patients

Frongia A[1,2], Brogna C[1,2], Malfatti E[3], Tasca G[4], Buchignani B[5], Pane M[1,2], Mercuri E[1,2]
[1]Paediatric Neurology UCSC, [2]Centro Clinico Nemo, Fondazione Policlinico Universitario "A Gemelli", IRCCS, [3]Université Paris Est, U955 INSERM, IMRB, APHP, Centre de Référence de Pathologie Neuromusculaire Nord-Est-Ile-de-France, Henri Mondor Hospital, [4]John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trusts, [5]Departement of Developmental Neuroscience IRCCS Stella Maris, Calambrone

VP118 A large cohort study of muscle imaging in GNE myopathy: Progression profile and diagnostic tips to distinguish from other distal myopathies

Yoshioka W[1,2], Mori-Yoshimura M[3], Eura N[1], Saito Y[1,2], Oya Y[3], Hayashi S[1], Kimura Y[4], Sato N[4], Noguchi S[1], Nishino I[1,2]
[1]Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry (NCNP), [2]Medical Genome Center, NCNP, [3]Department of Neurology, National Center Hospital, NCNP, [4]Department of Radiology, National Center Hospital, NCNP

VP119 Myotendinous junction abnormalities on skeletal muscle imaging common to COL6-related myopathies, ADSS1 myopathy and JAG2 myopathy

Saito Y[1], Hayashi S[1], Noguchi S[1], Nishino I[1]
[1]Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry

P120 Refining MRI pattern in sarcoglycanopathies: upper body pattern and new approaches to assess disease progression

Costa Comellas L[1], Sánchez-Montáñez Á[1], Maggi L[2], Díaz-Manera J[3,4], D'Amico A[5], Pichiecchio A[6], Pegoraro E[7], Monforte M[8], Løkken N[9], Marini-Bettolo C[3], Vlodavets D[11], Walter M[12], Voermans N[13], Monges S[14], Claeys K[15], Bevilacqua J[16], Alonso J[4], Comi G[17], Bruno C[10], Leonardis L[18], Straub V[3], Quijano-Roy S[19], Yves Carlier R[19], Vissing J[9], Mercuri E[3], Bertini E[5], Gomez-Andres D[1], Munell F[1], Tasca G[3,8]
[1]Hospital Universitari Vall d'Hebron, [2]Foundation IRCCS Carlo Besta Neurological Institute, [3]John Walton Muscular Dystrophy Research Centre, Newcastle Hospitals NHS Foundation Trusts, Newcastle University, [4]Hospital de la Santa Creu i Sant Pau, [5]Bambino Gesù Children's Hospital, [6]National Neurological Institute C Mondino, [7]University of Padova, [8]Fondazione Policlinico Universitario A Gemelli IRCC, [9]Rigshospitalet, University of Copenhagen, [10]Instituto Giannina Gaslini, [11]Russian Children Neuromuscular Center, Veltischev Clinical Pediatric Research Institute, [12]Friedrich-Baur-Institute, Department of Neurology, Ludwig-Maximilians-University of Munich, [13]Radboud University Medical Center, [14]Hospital de Pediatría J.P. Garrahan, [15]University Hospitals Leuven and University of Leuven (Katholieke Universiteit Leuven), [16]Hospital Clínico Universidad de Chile, [17]La Fondazione IRCCS Ca' Granda Ospedale Maggiore di Milano Policlinico, [18]Institute of Clinical Neurophysiology, [19]Hôpital Raymond Poincaré, Hôpitaux Universitaires Paris-Ile-de-France Ouest

P121 Long-term follow-up study of muscle MRI in Myotonic Dystrophy type 1: correlations with demographic and clinical characteristics

Fionda L[1], Lauletta A[1], Tufano L[1], Bucci E[1], Antonini G[1], Garibaldi M[1]
[1]Sapienza University

P122 Longitudinal Dixon Magnetic Resonance Imaging in dysferlinopathy patients can provide a powerful tool in assessing outcomes of therapeutic interventions.

Wilson I[1], Reyngoudt H[3], Bolano Diaz C[2], Araujo E[3], Moore U[2], Hilsden H[2], Diaz Manera J[2], Straub V[2], Carlier P[4], Blamire A[1]
[1]Newcastle University, [2]John Walton Muscular Dystrophy Research Centre, Newcastle University, [3]Institute of Myology, [4]CEA

P123 MRI based criteria to differentiate dysferlinopathies from other genetic muscle diseases

Bolaño Diaz C[1], Verdu-Diaz J[1], Gonzalez-Chamorro A[1], Straub V[1], Diaz Manera J[1,2,3]
[1]The John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University and Newcastle Hospitals NHS Foundation Trust, [2]Laboratori de Malalties Neuromusculars. Insitut de Recerca de l'Hospital de la Santa Creu i Sant Pau de Barcelona, [3]Centro de Investigación Biomédica en Red en Enfermedades Raras (CIBERER)

P124 A series of dysferlinopathy patients showing fluctuations in muscle fat fraction and contractile cross-sectional area values (cCSA) over a 3-year follow-up period

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P125 Quantitative MRI in upper limb muscles of patients with dysferlinopathy: 6-months and 12-months longitudinal data from the natural history Jain COS 2 project

Wilson I[1], Reyngoudt H[2], Caldas de Almeida Araujo E[2], Baudin P[2], Marty B[2], Bolano-Diaz C[3], Diaz-Manera J[3], Rufibach L[4], Hilsden H[3], Querin G[5], Pegoraro E[6], Mendell J[7], Stojkovic T[5], Straub V[3], Blamire A[2], Carlier P[8]
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P126 Muscle MRI-histology matching: data from 130 MRI-based muscle biopsies

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P127-P151: DMD - imaging and outcome measures

P127 Fat-fraction quantification using Dixon technique in Duchenne muscular dystrophy and its correlation with clinical progression and genotypic characteristics

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P128 Quantitative ultrasonography reveals skeletal muscle abnormalities in female carriers of DMD pathogenic variants

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P129 Givinostat in DMD: results of the Epidys study with particular attention to MR measures of muscle fat fraction

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P130 Two-year muscle MRI observations from a phase 1b trial of fordadistrogene movaparvovec (PF 06939926) for Duchenne muscular dystrophy (DMD)

Sherlock S[1], Li H[1], Butterfield R[2], Shieh P[3], Smith E[4], McDonnell T[1], Ryan K[1], Binks M[1]
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P131 MRI fat fraction distribution in Duchenne muscular dystrophy (DMD): effect size comparison to identify optimal biomarker for early efficacy assessment

Hammond M[1], Harris J[1], Luna B[1], Roche F[2], Vincent F[2], Berger M[1], Zabbatino S[1], Scheyer R[3], Holland S[1]
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P132 Quantifying skeletal muscle fat fraction and function using whole body magnetic resonance imaging (MRI) in men with Becker muscular dystrophy

Rock K[1], Willcocks R[1], Forbes S[1], Barnard A[1], Lott D[1], Smith B[1], Prabhakaran S[1], Rooney W[2], Daniels M[1], Subramony S[1], Chahin N[2], Walter G[1], Vandenborne K[1]
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P133 Influence of X-chromosome activation pattern in muscles on symptoms and progression of cardiac and muscle symptoms signs in women with pathogenic dystrophin gene variants: a 6-year follow-up of 53 patients

Lyu Z[1], Poulsen N, Joensen H, Lando C, Dunø M, Bundgaard H, Vejlstrup N, Vissing J
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P134 Energetics and acid-base status of skeletal muscle at rest and following isometric dorsiflexion and plantar flexion contractions in Duchenne muscular dystrophy

Awale P[1], Lopez C[1], Taivassalo T[2], Vandenborne K[1], Walter G[2], Forbes S[1]
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P135 Facilitators and barriers in living the desired adult life, despite having Duchenne muscular dystrophy (DMD)

Veenhuizen Y[1], Merkenhof L[1], Vroom E[2], Cup E[1], Groothuis J[1], Houwen S[1]
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P136 Using the North Star and timed function test centiles in boys with Duchenne muscular dystrophy – a prospective study

Wolfe A[1,2], Stimpson G[2], Milev E[1,2], O'Reilly E[1,2], Manzur A[1,2], Sarkozy A[1,2], Muntoni F[1,2], Baranello G[1,2]
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P137 Prophylactic use of cardiac medications and prolonged survival in Duchenne muscular dystrophy

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P138 Development and evaluation of a composite time-to-progression endpoint that spans ambulatory and non-ambulatory stages of Duchenne muscular dystrophy (DMD)

McDonald C[1], *Muntoni F[2], Marden J[3], Goemans N[4], Gomez-Lievano A[3], Zhang A[3], Chen Z[3], Ward S[5], Signorovitch J[3,5]*
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P139 Correlation and validation of the North Star Ambulatory Assessment, timed test and motor function measure centiles for boys with Duchenne muscular dystrophy

Milev E[1,2], *Stimpson G[1], van der Holst M[3], Wolfe A[1,2], O'Reilly E[1,2], Manzur A[2], Niks E[3], Houwen-Opstal S[4], Baranello G[1,2], Muntoni F[1,2]*
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P140 Predicting long-term trajectories of the North Star Ambulatory Assessment (NSAA) total score in Duchenne muscular dystrophy (DMD): an updated model

Muntoni F[1], *Signorovitch J[2], Goemans N[3], Manzur A[4], Done N[2], Sajeev G[2], Li J[2], Akbarnejad H[2], Sharma A[2], Niks E[5], Servais L[6], Straub V[7], de Groot I[8], Ward S[9], McDonald C[10]*
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P141 Concordance of patient-reported outcomes measurement information system (PROMIS) questionnaires between caregivers and children with DMD

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P142 Accurate translation from Performance of Upper Limb (PUL) version 1.2 to 2.0 in Duchenne muscular dystrophy (DMD): a machine learning algorithm

Coratti G[1], *Mercuri E[1], Sajeev G[2], Zhang A[2], Ward S[2], Pane M[1], Vilma B[2], Signorovitch J[2]*
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P143 Centiles by age for the North Star ambulatory assessment and the associated timed items in glucocorticoid treated boys with Duchenne muscular dystrophy

Stimpson G[1], *Ridout D[2], Wolfe A[1,3], Milev E[1,3], O'Reilly E[1,3], Manzur A[1], Cole T[2], Muntoni F[1,3], Baranello G[1,3], on behalf of the NorthStar Network*
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P144 Digital outcome captures longitudinal degradation of upper-limb function in non-ambulant patients affected by neuromuscular disorders

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P145 Analysis of the natural evolution of SV95C in ambulant patients with Duchenne muscular dystrophy

Rogers M[1,2], *Motola S[1], Eggenspieler D[1], Poleur M[3], Parinello G[1], Lozeve D[1], Danon A[3], Szabo L[4], Aragon-Gawińska K[5], Potulska-Chromik A[5], Butoianu N[6], Anghelescu C[6], Mirea[7], Osredkar D[8], Vrščaj E[8], Golli T[8], Haberlova J[9], Kodsy S[10], Salah A[10], Strijbos P[11], Servais L[1,3]*
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P146 A clinical trial simulation tool to accelerate trial design in DMD: description of the traphical user interface features and applications

Belfiore-oshan R[1], *Aggarwal V[1], Wilk J[2], Pauley M[1], Corey D[1], Romero K[1], Hovinga C[1], Martinez T[1], Lingineni K[2], Yoon D[2], Morales J[2], Kim S[2]*
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Continued on next page

P147 Six-year long-term safety and efficacy of Golodirsen in patients with DMD vs mutation-matched external controls

Muntoni F[1,2], Seferian A[3], Straub V[4], Guglieri M[4], Servais L[5,6], Wilk-Durakiewicz E[7], Ni X[7], Gao P[7], Hu M[7], Iff J[7], Hill L[7], Sehinovych I[7], Orogun L[7], Mercuri E[8,9]

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P148 Analysis of upper limb functional outcomes in a single centre paediatric cohort of non-ambulatory patients with Duchenne muscular dystrophy

Burnett N[1], Ridout D[2], Crook V[1], Robb S, Zambon A[1], Quinlivan R[1], Main M[1], Manzur A[1], Muntoni F[1,3], Sarkozy A[1]

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P149 Delayed pulmonary progression in Golodirsen-treated patients with Duchenne muscular dystrophy vs mutation-matched external controls

Iff J[1], Tuttle E[2], Liu Y[2], Wei F[2], Done N[2], Servais L[3,4], Seferian A[5], Straub V[6], Guglieri M[6], Mercuri E[7,8], **Muntoni F**[9,10]

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P150 Factors affecting the measurement variability of SV95C in ambulant patients with Duchenne muscular dystrophy

Rogers M[1,2], Motola S[1], Eggenspieler D[1], Poleur M[3], Parinello G[1], Lozeve D[1], Danon A[3], Szabo L[4], Aragon-Gawińska K[5], Potulska-Chromik A[5], Butoianu N[6], Anghelescu C[6], Mirea[7], Osredkar D[8], Vrščaj E[8], Haberlova J[9], Kodsy S[10], Salah A[10], Strijbos P[11], Servais L[2,3]

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P151 Serum adipokines in Duchenne muscular dystrophy: relationships to BMI, corticosteroids, and muscle fat fraction

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P205-P212, VP213, P214-P215, VP216, P217-P225, VP226, P227, VP228, P229-P234: SMA - clinical

P205 Impact of disease modifying treatment by three months of life on swallowing in Spinal Muscular Atrophy type 1

McGrattan K[1], Spoden A[1], McGhee H[6], Nichols K[6], Hernadez K[3], Ochura J[3], Graham R[3], Darras B[3], Brown A[4], Brandsema J[5], Karachunski P[2], Allen J[7], Miles A[7]

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P206 Impaired neurodevelopment in children with 5q-SMA - 2 years after newborn screening

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P207 Scoliosis progression in spinal muscular atrophy type II and III: a comparative study between treated and untreated patients

Coratti G[1], Lenkowicz J[1], Pera M[1], D'Amico A[2], Bruno C[3], Gullì C[1], Brolatti N[3], Antonaci L[1], Ricci M[1], Capasso A[1], Cicala G[1], De Sanctis R[1], Catteruccia M[2], Leone A[1], Paternello S[1], Pane M[1], Valentini V[1], Mercuri E[1]

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Continued on next page

P208 Paracetamol treatment in patients with spinal muscular atrophy: a different pharmacokinetic profile

Naume M[1,7], Zhao Q[2,3], Haslund-Krog S[4], Krag T[1], de Winter B[2,3], Revsbeck K[1], Vissing J[1], Holst H[4], Møller M[5], Hornsyld T[1], Dunø M[6], Høi-Hansen C[7], Born A[7], Andersen P[4], Ørngreen M[1,7]

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P209 Characteristics of patients with Spinal Muscular Atrophy who have discontinued treatment with nusinersen: a multi-centre experience in the UK

O'Reilly E[1,2], Stimpson G[1], Milev E[1,2], Rohwer A[1,2], Baranello G[1,2], Muntoni F[1,2], Scoto M[1,2], SMA Reach Network UK

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P210 Interim results from the RESPOND study evaluating nusinersen in children with spinal muscular atrophy previously treated with onasemnogene abeparvovec

Parsons J[1], Kuntz N[2], Brandsema J[3], Proud C[4], Finkel R[5], Swoboda K[6], Masson R[7], Foster R[8], Liu Y[9], Makepeace C[8], Singhi S[9], Paradis A[9], Berger Z[9], Rane S[9], Somera-Molina K[9]

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P211 Intravenous and intrathecal onasemnogene abeparvovec gene therapy in symptomatic and presymptomatic spinal muscular atrophy (SMA): long-term follow-up study

Darras B[1], Mercuri E[2], Strauss K[3,4,5], Day J[6], Chien Y[7], Masson R[8], Wigderson M[9], Alecu I[10], Ballarini N[10], Mehl L[11], Marra J[12], Connolly A[13,14]

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P212 RAINBOWFISH: Primary efficacy and safety data in risdiplam-treated infants with presymptomatic spinal muscular atrophy (SMA)

Finkel R[1], Farrar M[2], Servais L[3,4,5], Vlodavets D[6], Zanoteli E[7], Al-Muhaizea M[8], Prufer A[9], Nelson L[10], Fischer C[11], Gerber M[12], Gorni K[13], Kletzl H[14], Palfreeman L[11], Gaki E[11], Fontoura P[13], Bertini E[15], on behalf of the RAINBOWFISH Study Group

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VP213 Beneath the iceberg: Spinal muscular atrophy (SMA) and autistic spectrum disorder

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P214 Spinal presentations in children with Spinal Muscular Atrophy type 1 following gene therapy treatment in the SMA-REACH UK network

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P215 Evaluating longitudinal data of respiratory health in treated spinal muscular atrophy type 1 children using The Great Ormond Street Respiratory score

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VP216 Tracking bone health in pediatric patients with spinal muscular atrophy (SMA)

Seçgen N[1], Öz Yıldız S[1], Aksoy T[2], Demirkıran G[2], Özön A[3], Yazıcı M[2], Aydıngöz Ü[4], **Haliloğlu G**[1]

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P217 Map the SMA protocol: a machine-learning based algorithm to predict therapeutic response in Spinal Muscular Atrophy

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P218 Patients' perceptions of the effects of Spinraza according to their status as a responder or non-responder

Lilien C[1], Vrscaj E[2,3], Poleur M[2], Ataide P[4], Deconinck N[5], de Waele L[6,7], Duong T[4], Haberlova J[8], Jilkova M[8], Osredkar D[3], Peirens G[6], Szabo L[9], Tahon V[5], Benhammed N[2], Médard L[2], Servais L[1,2]

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P219 Longitudinal disease progression in the Revised Hammersmith Scale in a cohort of untreated SMA 2 and 3 patients

Stimpson G[1], Wolfe A[1,8], Ramsey D[1,2], O'Reilly E[1,8], Rowher A[1], Muni Lofra R[3], Coratti G[4,5], Duong T[6], Dunaway Young S[6], Gee R[7], Baranello G[1,8], Scoto M[11], the RHS Working Group, Finkel R[9,10], Mercuri E[4,5], Muntoni F[1,8], on behalf of the international SMA consortium (iSMAc)

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P220 National newborn screening for SMA in Norway

Wallace S[1], Ørstavik K[1], Rowe A[1], Strand J[1]

[1]Oslo University Hospital

P221 A five-year review of newborn screening for Spinal Muscular Atrophy in the state of Utah: lessons learned

Wong K[1], Cook S[2], Hart K[2], Moldt S[1], Wilson A[1], McIntyre M[1], Rohrwasser A[2], Butterfield R[1]

[1]Department of Paediatrics, University of Utah Health, [2]Utah Newborn Screening Program

P222 MUNIX of abductor digiti minimi correlates with upper limb function in adult patients with spinal muscular atrophy

Graça F[1], Iwabe C[1], **Cavalcante França Jr M**[1]

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P223 Long-term follow-up of onasemnogene abeparvovec gene therapy in patients with spinal muscular atrophy (SMA) type 1

Mendell J[1], Wigderson M[2], Alecu I[3], Yang L[4], Mehl L[5], **Connolly A**[1,6]

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P224 Effect of Apitegromab on Motor Function at 36-months in patients with nonambulatory Spinal Muscular Atrophy aged 2-12 years old

Crawford T[1], Darras B[2], Day J[3], De Vivo D[4], Mercuri E[5], Nascimento A[6], Mazzone E[5], on behalf of the TOPAZ Study Team[7], Waugh A[8], Song G[8], Evans R[8], Marantz J[8]

[1]Johns Hopkins Medical, [2]Boston Children's Hospital, [3]Stanford Neuroscience Health Center, [4]Columbia University Irving Medical Center, [5]Catholic University, [6]Hospital Sant Joan de Déu, [7]TOPAZ Study Team includes clinical trial investigators, physical therapists, study coordinator, [8]Scholar Rock, Inc.

P225 Longitudinal changes in compound muscle action potential and their association with motor function in infantile-onset SMA children in ENDEAR/SHINE

Sumner C[1], Youn B[2], Farrar M[3,4], Tichler B[5], Berger Z[2], Zhu C[2], Paradis A[2]

[1]Department of Neurology, Johns Hopkins University School of Medicine, [2]Biogen, [3]School of Clinical Medicine, UNSW Medicine and Health, UNSW Sydney, [4]Department of Neurology, Sydney Children's Hospital Network, [5]Biogen

VP226 Post-hoc analysis of compound motor action potential from clinical trials of intravenous onasemnogene abeparvovec for spinal muscular atrophy

McGill B[1], Maca J[1], Tauscher-wisniewski S[2], Macek T[1]

[1]Novartis Pharmaceuticals Corporation, [2]Novartis Gene Therapies, Inc.

P227 MANATEE: GYM329 (RO7204239) in combination with risdiplam treatment in patients with spinal muscular atrophy (SMA)

Duong T[1], Darras B[2], Morrow J[3], Muntoni F[4], Servais L[5,6,7], Rabbia M[8], Gerber M[9], Kletzl H[10], Gaki E[11], Fletcher S[11], Scalco R[12], Wagner K[12], Mercuri E[13]

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VP228 Post-hoc analyses of prednisolone use and hepatotoxicity in clinical trials of intravenous onasemnogene abeparvovec

*Macek T[1], Wen S[1], O'Brien E[2], Alecu I[3], **McGill B**[4]*
[1]Novartis Pharmaceuticals Corporation, [2]Novartis, [3]Novartis Pharmaceuticals, [4]Novartis Gene Therapies, Inc.

P229 Adult SMA REACH: a clinical network to standardize the collection of data to enable integrated and longitudinal analysis of clinical and patient-reported data

***Muni Lofra R**[1], Segovia S[1], Elwell T[1], Yau J[1], Murphy L[1], Blewitt C[1], Fitzsimmons S[1], Marini Bettolo C[1], Network A[2]*
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P230 Safety update: Risdiplam clinical trial program for spinal muscular atrophy (SMA)

***Baranello G**[1,2], Chiriboga C[3], Servais L[4,5,6], Darras B[7], Day J[8], Deconinck N[9,10], Farrar M[11], Finkel R[12], Bertini E[13], Kirschner J[14], Masson R[2], Mazurkiewicz-Bełdzińska M[15], Vlodavets D[16], Bader-Weder S[17], Gorni K[18], Jaber B[17], Yeung W[19], Papp G[17], Scalco R[20], Mercuri E[21], on behalf of the FIREFISH, SUNFISH, JEWELFISH and RAINBOWFISH Study Groups*
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P231 Early intervention and speed-to-effect in spinal muscular atrophy type 1 following onasemnogene abeparvovec gene replacement therapy

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P232 Bioavailability and bioequivalence of Risdiplam tablets in healthy volunteers

***Kletzl H**[1], Heinig K[1], Jaber B[2], Lomeli B[3], Yeung W[4], Young A[4], Coleman H[3], Morrison D[5]*
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P233 Zolgensma in Spinal Muscular Atrophy: a Toronto paediatric hospital experience

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P234 Effect of Apitegromab on pedi-cat and promis-fatigue questionnaire at 36-months in patients with Spinal Muscular Atrophy

***Crawford T**[1], Darras B[2], Day J[3], Krueger J[4], Mercuri E[5], Nascimento A[6], Pasternak A[2], Duong T[3], on behalf of the Topaz Study Team[7], Liu L[8], Sadanowicz M[8], Baver S[8]*
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P319-P333, VP334, P335-P338, VP339, P340: Myositis

P319 Histopathological features and autophagy aspects of Ku+ myositis

***Preusse C**[1], Holzer M[2], Schneider U[3], Schänzer A[4], Léonard-Louis S[5], Benveniste O[6], Weis J[7], Claeys K[7,8], Schoser B[9], Montagnese F[9], Uruha A[1,10], Huber M[11], Gallay L[12], Streichenberger N[13], Krusche M[2], Stenzel W[1]*
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P320 VMA21 conditional knockout mice model XMEA with myopathy and dysfunctional autophagy

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P321 Exploring hand and upper limb function in patients with Inclusion Body Myositis

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P322 Inclusion body myositis with early onset – a population-based study

Lindgren U[1,2], Hedberg-Oldfors C[1], Pullerits R[3,4], Lindberg C[2], Oldfors A[1]
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P323 Differences in clinicopathology and therapeutic response of idiopathic inflammatory myopathy with anti-SRP, HMGCR, and mitochondrial M2 antibodies

Yamanaka A[1], Eura N[1], Nihimori Y[1], Shiota T[1], Nanaura H[1], Kiriyama T[1], Izumi T[1], Kataoka H[1], Sugie K[1]
[1]Nara Medical University

P324 A case of paediatric anti-HMGCR myopathy mimicking LGMD

Frongia A[1,2], Daniela L[1,2], Tasca G[3], Andreetta F[4], Antonaci L[1,2], Mercuri E1,2, Pane M[1,2]
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P325 A comparative single nuclei transcriptomics approach to evaluating the terminally differentiated lymphocytes in autoimmune Myositis

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P326 Immune myopathy with perimysial pathology in a patient with an unusual clinical phenotype and Anti-Mi-2 antibody

Pham X[1,2,3], Siriratnam P[1,3], Rodrigues E[1,3,4], McLean C[5]
[1]Department of Neurology, Alfred Health, [2]Australian and New Zealand Intensive Care Research Centre, School of Public Health and Preventive Medicine, Monash University, [3]Department of Neuroscience, Central Clinical School, Monash University, [4]Department of Neurology, Royal Melbourne Hospital, [5]Victorian Neuromuscular Laboratory Service, Anatomical Pathology, Alfred Health

P327 Responsiveness of rituximab in refractory cases of inflammatory myopathies

No J[1], Park Y[1]
[1]Pusan National University Hospital, [2]Pusan National University Yangsan Hospital

P328 Investigations of the specific interferon-signature in Anti-Synthetase syndrome-associated Myositis

Preusse C[1,2], Gallay L[3], Pinal-Fernandez I[4,5], Mammen A[4,5,6], Benveniste O[7], Goebel H[1,8], Streichenberger N[9], Roos A[10,11], Ruck T[12], Stenzel W[1]
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P329 The role of autoantibodies in diagnosis of Idiopathic inflammatory myopathies

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P330 Profile of adult idiopathic inflammatory myopathy in Dr Cipto Mangunkusumo Hospital Indonesia as tertiary health care

Indrawati L[1,2], Wibowo S[2,3], Widhani A[2,4], Novianto E[2,5], Nagpal C[1], Paveta D[1], Susanto E[2,6], Bilianti Y[2,6], Fadli N[2,7], Budikayanti A[1,2], Safri A[1,2], Wiratman W[1,2], Octaviana F[1,2], Hakim M[1,2]
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P331 The selection of biopsy sites in lower extremities for the diagnosis of vasculitis

Sato M[1], Kurashige T[3], Murao T[3], Tokunaga T[2], Suma H[2], Hirata S[1], Ohshita T[3]
[1]Department of Clinical Immunology and Rheumatology, Hiroshima University Hospital, [2]Department of Rheumatology, National Hospital Organization Kure Medical Center and Chugoku Cancer Center, [3]Department of Neurology, National Hospital Organization Kure Medical Center and Chugoku Cancer Center

P332 Mitochondrial pathology associated with refractory dermatomyositis after COVID-19 vaccination

Lauletta A[1], Merlonghi G[1], Fionda L[1], Garibaldi M[1]
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P333 Clinical, pathological heterogeneity and mitochondrial dysfunction in patients with anti-mitochondrial antibodies related myositis

Zhang W[1], Wang Y[1], Zhao Y[1], Yuan Y[1]
[1]First Hospital Peking University

VP334 Clinicopathological features of anti-mitochondrial M2 antibody-positive myositis based on a cohort of 201 patients from Japan

Nishimori Y[1,2], *Tanboon J*[2,3], *Oyama M*[4], *Motegi H*[4,5], *Tomo Y*[6], *Oba M*[6], *Sugie K*[1], *Suzuki S*[4], *Hayashi S*[2], *Noguchi S*[2], *Nishino I*[2]
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P335 Inhibition of KDM5A reverses pathological features in sporadic Inclusion Body Myositis-like cell models

De Vries G[1], *de Ridder W*[1,2,3], *Baets J*[1,2,3]
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P336 Refining the clinical and therapeutic spectrum of granulomatous myositis from a large cohort of patients

Lauletta A[1], *De le Hoye L*[2], *Léonard-Louis S*[3], *Garibaldi M*[4], *Allenbach Y*[5], *Benveniste O*[5]
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P337 Recessive Charcot-Marie-Tooth and multiple sclerosis associated with a variant in MCM3AP: a case report

Yuksel D[1], *Gocmen R*[2], *Temucin C*[3], *Lafci N*[4]
[1]*University Health of Sciences Turkey, Ankara Etlik City Hospital, Department of Paediatric Neurology,* [2]*Hacettepe University Faculty of Medicine, Department of Radiology,* [3]*Hacettepe University Faculty of Medicine, Department of Neurology,* [4]*Hacettepe University Faculty of Medicine, Department of Medical Genetics*

P338 Clinical, morphological, and proteomic features of patients suspected of X-linked myopathy with excessive autophagy (XMEA)

Merlet A[1,2], *Lacène E*[3], *Nelson I*[4], *Brochier G*[3], *Labasse C*[3], *Chanut A*[3], *Madelaine A*[3], *Beuvin M*[3], *Bonne G*[4], *Féasson L*[1,2], *Minot M*[5], *Noury J*[6], *Fradin M*[7], *Fernández-Eulate G*[8], *Behin A*[8], *Stojkovic T*[8], *Hentschel A*[9], *Marcorelles P*[10], *Roos A*[11], ***Evangelista T***[3]
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VP339 A case of systemic sarcoidosis with nerve and muscle involvement induced by tattoos

Lee J[1]
[1]*The Catholic University of Korea*

P340 The MikroIBioM study - Comparison of gut microbiome of sporadic Inclusion Body Myositis (sIBM) patients and unaffected spouses

Winkler M[1], *Seel W*[2], *Kornblum C*[1], *Simon M*[2], *Reimann J*[1]
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15:30-16:00	**Short Oral Presentations 1** Ballroom C1 **P319-P322, P126-P127** *Moderator: Tahseen Mozaffar, University of California, USA*	**Short Oral Presentations 2** Ballroom C2 **P205-210** *Moderator: Laurent Servais, University of Oxford, UK*	**Short Oral Presentations 3** Ballroom C3 **P211-P212, P56, P57, P59** *Moderator: Jana Haberlová, Motol University Hospital, Czech Republic*
16:15-17:00	PAC **Debate: Is the muscle biopsy still indicated?** *Moderators: Teerin Liewluck, Mayo Clinic-Rochester, USA & Edoardo Malfatti, Paris Est University/INSERM U955, France* INV06: ***Teresinha Evangelista****, Institut de Myologie, France* INV07: ***Baziel van Engelen****, Radboud University Medical Centre, The Netherlands*		
17:15-18:15	**Poster session 2** Ballroom A-C *Refreshments served*		
	P12-P18, P20-P37, VP38, P39-P40: DMD - treatments		

P12 Comparison of U7snRNA-induced dystrophin expression following systemic delivery with AAV9 and AAVrh74 capsids

Lay J[1], Frair E[1], Bradley A[1], Vetter T[1], Rohan N[1], Bellinger C[1], Waldrop M[1,2,3], Wein N[1,2], Gushchina L[1,2], Flanigan K[1,2,3]
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P13 Comparison of U7snRNA-induced dystrophin expression following systemic delivery with AAV9, MyoAAV 2A, and MyoAAV 3A capsids in the Dup2 mouse

Frair E[1], Bradley A[1], Dufresne G[1], Sarff J[1], Stevens K[1], Rohan N[1], Nicolau S[1], Vetter T[1,2], Gushchina L[1,2], Flanigan K[1,2,3]
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P14 U7snRNA-mediated exon skipping as a powerful therapeutic tool for the treatment of DMD

Saylam E[1], Terry K[1], Suhaiba A[1], Bellinger C[1], Casey S[1], Dufresne G[1], Huang N[1], Rohan N[1], Lowery A[1], Wein N[1], Gushchina L[1], Flanigan K[1,2]
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P15 Full-length dystrophin restoration in multiple patient cell lines with DMD pseudoexons using AAV-delivered U7snRNA

Beljan J[1,2], Gushchina L[1], Nicolau S[1], Flanigan K[1,2]
[1]Nationwide Children's Hospital, [2]Ohio State University

P16 An investigational AAV8 gene therapy coding for a novel microdystrophin as a treatment for Duchenne muscular dystrophy

Dastgir J[1], Rastogi S[1], Philips D[1], Wilson C[1], Boulos N[1], Hall J[1], Jimenez V[1], Gilmor M[1], Falabella P[1], Owusu L[1], Fiscella M[1], Liu Y[1], Pakola S[1], Danos O[1]
[1]Regenxbio, Rockville, United States

P17 Safety and efficacy of pre-treatment with imlifidase prior to AAV-based gene therapy in non-human primates with pre-existing anti-AAVrh74 antibodies

Potter R[1], Khan S[1], Snedeker J[1], Adegboye K[1], Haile A[1], Sayanjali B[1], Pukos N[1], Cochran K[1], Ahner J[1], Su T[1], Uzcátegui N[2], Stenberg Y[2], Freiburghaus C[2], Winstedt L[2], Rodino-Klapac L[1]
[1]Sarepta Therapeutics Inc, [2]Hansa Biopharma

P18 WVE-N531 with PN backbone modification significantly enhances drug concentrations in heart, diaphragm, and skeletal muscles in non-human primates

Hart A[1], Hu X[1], Lamore S[1]
[1]Wave Life Sciences

P20 Endosomal Escape Vehicles (EEV™) - Oligonucleotides conjugates produce exon skipping and dystrophin production in preclinical models of Duchenne muscular dystrophy

Girgenrath M[1], Estrella N[1], Kumar A[1], Li J[1], Hicks A[1], Brennan C[1], Blake S[1], Guan A[1], Li X[1], Pathak A[1], Kheirabadi M[1], Dougherty P[1], Lian W[1], Liu N[1], Gao N[1], Wang D[1], Streeter M[1], Stadheim A[1], Dhanabal M[1], Qian Z[1]
[1]Entrada Therapeutics

P21 Risk tolerance of caregivers of individuals with Duchenne muscular dystrophy for gene therapy

Camino E[1], Heslop E[2], McNiff M[2], Jonhson A[3], Fischer R[1], Denger B[1], Hill C[4], Cope H[4], Peay H[4]
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P22 WVE-N531 yields 53% mean exon 53 skipping in skeletal muscle of boys with Duchenne muscular dystrophy (DMD) after three biweekly doses

Tillinger M[1], Lake S[1], Servais L[2], Campbell C[3], Xu X[1], Hart A[1], Haegele J[1], Singh K[1], Rheinhardt J[1], Ghosh A[1], Xu D[1], Panzara M[1], Li-Kwai-Cheung A[1]
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P23 Evaluation of safety parameters and dystrophin expression by sequential administration of exon-skipping and gene therapy in a DMDmdx mouse model

Potter R[1], Cooper Olson G[1], Smith L[1], Greve J[1], Haile A[1], Wier C[1], Snedeker J[1], Burch P[1], Hunter B[1], Malmberg A[1], Rodino-Klapac L[1]
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P24 Safety and tolerability of Eteplirsen in patients 6–48 Months old with DMD amenable to exon 51 skipping: an open-label extension study

Mercuri E[1,2], Seferian A[3], Deconinck N[4], Orogun L[5], Ni X[5], Zhang W[5], Drummond K[5], Sehinovych I[5], Muntoni F[6,7]
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P25 Single- and repeat-dose nonclinical data for PGN-EDO51 demonstrate potential for the treatment of Duchenne muscular dystrophy (DMD)

Holland A[1], Lonkar P[1], Sweeney C[1], Gilbert J[1], Svenstrup N[1], Goyal J[1]
[1]PepGen Inc

P26 CONNECT-EDO51: Trial designs to support the development of PGN-EDO51 for Duchenne Muscular dystrophy amenable to exon 51 skipping

*Larkindale J[1], Vacca S[1], Cormier J[1], Shoskes J[1], Goyal J[1], Holland A[1], Lonkar P[1], Foy J[1], **Mellion M**[1]*
[1]PepGen

P27 Three novel enhanced delivery Oligonucleotide candidates for Duchenne muscular dystrophy mediate high levels of exon 53, 45, and 44 skipping

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[1]PepGen Inc

P28 The antisense oligonucleotide BMN 351 durably ameliorates dystrophic phenotypes in a mouse model of exon 51–skip-amenable Duchenne muscular dystrophy

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[1]Biomarin Pharmaceutical Inc

P29 DMD transcript imbalance and nuclear trafficking evaluation in muscle biopsies from baseline and golodirsen treated 4053-101 clinical trial patients

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P30 A phase 1b/2 open-label study of WVE-N531 in patients with Duchenne muscular dystrophy: part B study design and rationale

***Tillinger M**[1], Volpe M[1], Casey C[1], Lake S[1], Hu X[1], Xu D[1], Narayanan P[1], Hart A[1], Haegele J[1], Lamore S[1], Bhatia S[1], Li-Kwai-Cheung A[1], Servais L[2]*
[1]Wave Life Sciences, [2]Oxford Children's Hospital, Oxford University Hospitals NHS Foundation Trust

P31 Interim analysis of EVOLVE: evaluating Eteplirsen, Golodirsen, or Casimersen treatment in patients <7 years old in routine clinical practice

*Grabich S[1], Santra S[1], Waldrop M[2], Mathews K[3], Abid F[4], **Ramos-Platt L**[5], Scharf R[6], Zaidman C[7], Sehinovych I[1], McDonald C[8]*
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P32 Jak inhibitors Tofacitinib and Ruxolitinib do not improve functional deficits in dystrophin-deficient mdx mice

*Bosco C[2], Uaesoontrachoon K[2], Srinivassane S[2], Rowsell J[2], Elustondo P[2], Mackinnon A[2], Nagaraju K[2,3], **Peterson J**[1]*
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P33 RKER-065 ameliorated muscle and bone loss in a progressive murine model of Duchenne muscular dystrophy

***Nathan R**[1], Cahill M[1], Todorova R[1], Macaluso S[1], Tseng C[1], Fisher F[1], Lerner L[1], Seehra J[1], Lachey J[1]*
[1]Keros Therapeutics Inc.

P34 RKER-065, a novel ActRII ligand trap, counteracted the negative impact of glucocorticoid treatment on bone and muscle

***Zhen G**[1], Nathan R[1], Cahill M[1], Materna C[1], Fisher F[1], Lerner L[1], Lachey J[1], Seehra J[1]*
[1]Keros Therapeutics

P35 Two-year clinical outcomes with fordadistrogene movaparvovec (FM) for Duchenne muscular dystrophy (DMD) and contextualization with external controls

***Shieh P**[1], Butterfield R, Muntoni F, Mercuri E, Signorovitch J, Schwartz P, Li H, Binks M, McDonnell T, Ryan K, Delnomdedieu M, Shen Q, Levy D, Smith E*
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P36 Givinostat in Duchenne muscular dystrophy: effect on disease milestones

***McDonald C**[1], Servais L[2], Munell F[3], Schara-Schmidt U[4], Bertini E[5], Comi G[6], Blaschek A[7], Cazzaniga S[8], Bettica P[8], Vandenborne K[9], Mercuri E[10]*
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P37 Givinostat in DMD: results of the Epidys Study with particular attention to NSAA

***Mercuri E**[1], Brogna C[2], Mah J[3], Goemans N[4], Niks E[5], Cazzaniga S[6], Bettica P[6], McDonald C[7]*
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VP38 Changes to glucocorticosteroid prescribing patterns in Duchenne muscular dystrophy in the UK over the last decade

***Landon G**[1,2], Stimpson G[1], Sarkozy A[1,2], Manzur A[1,2], Guglieri M[3], Muntoni F[1,2], Baranello G[1,2]*
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P39 The effect of corticosteroid treatment on pulmonary function in adults with Duchenne muscular dystrophy

Pietrusz A[1], Astin R[2], Guglieri M[3], Desikan M[2], Waller K[3], Chapman S[3], Schiava M[3], Brady S[4], Soleimani B[4], Freebody J[4], Nickol A[5], Ramdharry G[2], Muntoni F[6], Quinlivan R[1,2]

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P40 Long-term effects of corticosteroid treatment in DMD: daily versus intermittent regimes

Ikelaar N[1], van der Holst M[1], Meijer - Krom Y[1], Stoop M[1], Houwen - van Opstal S[2], Goemans N[3], Geuens S[3], de Waele L[3], Niks E[1]

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P168-P177, VP178, P179-P187, VP188, P189-P190: Genetics of neuromuscular disorders

P168 The burden of titin variants on genetic counseling

Di Feo M[2,7], Topf A[3], Matalonga L[4], Paramonov I[4], Perrin A[5], Johari M[1,6], SNV/indels working group, NMD-DITF, SolveRD Consortium, Cossee M[5], Hackman P[1,2], **Savarese M**[1,2], Udd B[2]

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P169 Childhood onset amyotrophic lateral sclerosis associated with SPTLC2 gain-of-function pathogenic variants: clinical, genetic, and biochemical insights

Or Bach R[1], Syeda S[1], Mohassel P[1], Dohrn M[2,3], Lone M[4], Donkervoort S[1], Foley A[1], Beijer D[2], Bayraktar E[5], Oflazer P[6], Munot P[7], Rose A[8], Lyons M[8], Muntoni F[7,9], Başak A[5], Dunn T[10], Hornemann T[4], Züchner S[2], Bönnemann C[1], International SPTLC2 Study Group

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P170 TDP-43 seeding and aggregation in skeletal muscle

Lynch E[1], Pittman S[1], Daw J[1], Weihl C[1]

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P171 LiBi-NMD: liquid biopsies in neuromuscular diseases – the underrated value of white blood cells

Hentschel A[1], Della Marina A[2], Köbel H[2], Gangfuss A[2], Dohrn M[3], Weis J[3], **Dobelmann V**[4], Krause K[5], Ruck T[4], Vorgerd M[5], Schara-Schmidt U[2], Roos A[2,5,6]

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P172 Exploring the diagnostic ability of RNA-seq to identify disease-causing variants in muscular dystrophy

Gaynor A[1], Hale M[1], Lek M[2], Provenzano M[1], Bates K[1], Johnson N[1]

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P173 A highly responsive bioassay for quantification of glucocorticoids

Poulsen M[1], Overgaard M[2,3], Andersen C[1], Lodberg A[1]

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P174 Subtyping of cardiac amyloidosis by mass spectrometry of endomyocardial biopsies

Oldfors A[1], Noborn F[1], Thomsen C[1], Vorontsov E[2], Bobbio E[3], Sihlbom C[2], Nilsson J[1], Polte C[4], Bollano E[3], Vukusic K[1], Sandstedt J[1], Dellgren G[5], Karason K[3], Larson G[1]

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P175 Muscle biopsy methylome analysis creates well-defined clusters for inherited myopathies

Lopes Abath Neto O[1], Moore S[1]

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P176 Immortalized human muscle cells: easy-to-use models to study neuromuscular diseases

Butler-Browne G[1], Ohana J[1], Mamchaoui K[1], Bensalah M[1], Negroni E[1], Trollet C[1], Bigot A[1], Mouly V[1]

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P177 Exomiser is an efficient tool to prioritize candidate genes in cohorts of unsolved myopathy patients

Lillback V[1,2], De Feo M[3], Johari M[1,2,6], Vicidomini G[4], Hackman P[1,2], Udd B[2,5], **Savarese M**[1,2]

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VP178 Multiomics needed to increase the detection rate of myopathy patients

Owusu R[1,2], Johari M[1,2], Lehtinen S[3], Jokela M[3,4], Palmio J[3], Hackman P[1,2], Udd B[1,2], Savarese M[1,2]
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P179 Using Long-read RNA sequencing for the identification of novel transcripts in disease-causing muscle genes

Johari M[1], Ravenscroft G[1]
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P180 A retrospective chart review evaluating clinical presentation and genetic testing approaches for patients with neuromuscular disorders

Rosenberg A[1], Tian C[1], He H[1], Ulm E[1], Collins K[1], **Bhimarao Nagaraj C**[1]
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P181 Trio genome analysis in 45 unsolved children with neuromuscular diseases

Natera De Benito D[1,2], Estevez-Arias B[1,3], Matalonga L[4], Ortez C[1,2,5], Carrera-Garcia L[1,2], Exposito-Escudero J[1,2], Codina A[1,2], Jou C[1,2,5,6], Beltran S[4], Nascimento A[1,2,5]
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P182 Spectrum of next generation sequencing-confirmed myopathies: a single-centre cohort from South India

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P183 The genetic profile of childhood neuromuscular disorders: a single center experience

Tezel O[1], Öztürk G[2], **Ünver O**[2], Polat H[3], Ayaz A[4], Aksoy Özcan S[2], Türkdoğan D[2]
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P184 Revealing myopathy spectrum: Integrating transcriptional and clinical features of human skeletal muscles with varying health conditions

Zhong H[1], Johari M[2,3], Katayama S[2,4], Oghabian A[2,5], Sian V[6], Jonson P[2,7], Hackman P[2,8], Savarese M[2,7], Udd B[2,9]
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P185 Global carrier frequency and genetic prevalence of autosomal-recessive genetic neuromuscular disorders

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P186 Spectrum of skeletal muscle channelopathies in a cohort of Inherited neuromuscular disorders

Sidharth S[1], Macken W[2,3], Mishra R[1], Reyaz A[1], Ahmed T[1], ICGNMD Consortium[4], Bhatia R[1], Pitceathly R[2,3], Thangaraj K[5,6], Srivastava P[1], Hanna M[2,3], Venugopalan V[1]
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P187 ZC4H2 X linked gene mutations: phenotypic spectrum of arthrogryposis multiplex congenita

Alvarenga N[1], Saez V[1], Lotz S[1], Exposito J[1,3], Carrera L[1,3], Natera D[1,3], Armijo J[1], Rios A[1], Artiga V[1], Jou C[2], Codina A[2], Yubero D[5], Martorell L[5], **Ortez C**[1,3,4], Nascimento A[1,3,4]
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VP188 Novel PIEZO2 variants in a cohort of arthrogryposis syndrome

Jofre J[1], Suarez B[1,2], Calcagno G[1], Hervias C[1], Fattori F[3], Bertini E[3], Castiglioni C[1,2]
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P189 Dock7 is an essential driver of skeletal muscle health and function

English K[1], Samani A[1], Becker K[2], Karupassamy M[1], Alexander M[1]
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P190 Dramatic improvement of scoliosis in a patient with DOK7-related congenital myasthenic syndrome treated with ephedrine

Graça F[1], Iwabe C[1], **Cavalcante França Jr M**[1]
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P281-P285, VP286, P287-P288, VP289, P290, VP291, P292-P305, VP306, P307: Limb-girdle muscular dystrophies

P281 Quality of life in adults with dysferlinopathy: international clinical outcome study of dysferlinopathy

Hilsden H[1], James M[1], Gordish Dressman H[2,3], Day J[4], Mendell J[5], Fernandez Torron R[6], Harms M[7], Pestronk A[8], Vissing J[9], Desai U[10], Yoshimura M[11], Shin J[12], Mozaffar T[13], Stojkovic T[14], Pegoraro E[15], Bevilacqua Rivas J[16], Olive M[17], Paradas C[18], Straub V[1], Mayhew A[1]
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P282 Evaluation of gene transfer efficiency in a mild model of dystrophic muscle disorder performed by machine learning and linear discriminant analysis

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P283 Natural history of limb girdle muscular dystrophy R9: one-year follow-up of a European cohort

Vissing J[1], Stojkovic T[3], Straub V[2], Preisler N[1], Holm-Yildiz S[1], Rudolf K[1], Querin G[3], Hogrel J[3], Birnbaum S[3], James M[2], Ghimenton E[2], Verma M[2], Richard I[4], Granier M[4], Degove S[5], Olivier S[5]
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P284 Gene replacement therapy for telethonin related limb-girdle muscular dystrophy R7 utilizing novel myotrophic AAV capsids

Gushchina L[1,2], Bradley A[1], Terry K[1], Lay J[1], Frair E[1], Vetter T[1,2], Rohan N[1], Cox G[4], Wolfe S[5], Emerson C[5], Flanigan K[1,2,3]
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P285 Bi-allelic variants in HMGCR cause limb girdle muscular dystrophy and further implicate the mevalonate pathway in muscle disease

Foley A[1], **Donkervoort S**[1], Bharucha-Goebel D[1], Saade D[1], Flynn L[2], Grunseich C[3], Hu Y[1], Bruels C[4], Littel H[4], Estrella E[5], Krishnamoorthy K[6], Chao K[7], Pais L[7], Kunkel L[8], Kang P[4], Bönnemann C[1]
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VP286 Clinical, imaging, pathological, and molecular features of HNRNPDL-related muscular dystrophy

Cotta A[1], Venturini M[2], Rocha G[2], Muniz V[2], Berbare D[3], da Cunha Junior A[1], Medeiros R[2], da Costa K[3], Cordeiro B[4], Costa e Silva C[4], Carvalho E[1]
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P287 Autosomal dominant and recessive variants within the C-terminal domain of HNRPDL cause a phenotypically similar LGMD

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P288 Defining clinical endpoints in limb girdle muscular dystrophy D1-DNAJB6-related: A GRASP consortium study

Findlay A[1], **Hunn S**[1], Alfano LN[2], Lowes LP[2], Wicklund M[5], Leung D[6], Jones A[3], Butler A[3], Hayes M[4], Sasidharan S[4], Holzer M[6], Stinson N[6], Seiffert M[1], Statland J[4], Johnson NE[3], Weihl CC[1] and the GRASP-LGMD Consortium
[1]Washington University in St. Louis; [2]Nationwide Children's Hospital; [3]Virginia Commonwealth University; [4]Kansas University Medical Center; [5]University of Colorado—Denver; [6]Kennedy Krieger Institute

VP289 Allele specific knockdown for LGMDD1

Findlay A[1], Vohra A[1], Haller M[1], Paing M[1], Daw J[1], Pittman S[1], Miller T[1], Chou T[2], Harper S[3], Weihl C[1]
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P290 Novel dominant capain mutation in a Brazilian family

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VP291 The clinical, imaging and genetic characteristics in a large cohort of LGMDR1 patients from an Egyptian referral center

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P292 Clinical outcome assessments in limb girdle muscular dystrophy R1/2A: a longitudinal update

Hunn SM[1], Alfano LN[2], Lowes LP[2], Wicklund M[5], Mathews KD[6], Mozaffar T[7], Leung D[8], Jones A[3], Butler A[3], Hayes M[4], Sasidharan S[4], Reash NF[2], Iammarino MA[2], Laubscher K[6], Mockler S[6], Ausberger R[7], Holzer M[8], Stinson N[8], Seiffert M[1], Statland J[4], Johnson NE[3], Weihl CC[1], and the GRASP-LGMD Consortium
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P293 Clinical and genetic spectrum of sarcoglycanopathies in a cohort of Turkish patients with a possible founder variation

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P294 A comprehensive study of the inflammatory signature in sarcoglycanopathies

Kölbel H[1], Preuße C[2,3], Della-Marina A[1], Schara-Schmidt U[1], Goebel H[2,4], Roos A[1,5,6], Stenzel W[2]
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P295 Functional improvements by ataluren in dysferlinopathy mice with a compound heterozygous mutations carrying one nonsense variant

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P296 Copper nanoparticles ameliorates Dysferlinopathy phenotype by promoting mitochondrial homeostasis

Huerta H[1,2], González C[1], Morgado-Cáceres P[1], Salas-Huenuleo E[5], Cárdenas C[1,2,3,4]
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P297 Validation of a blood-based assay for dysferlinopathy in a Latin American cohort

Barresi R[1], Töpf A[2], Gonzalez-Chamorro A[2], Diaz-Manera J[2], Emmons S[3], Rufibach L[3]
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P298 Controlled storage conditions improve specificity and sensitivity of a blood-based assay for dysferlinopathy: a pilot study in an Indian cohort

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P299 Over-expression of FKRP in heart induces myocarditis and dilated cardiomyopathy in LGMD2I/R9 mice

Huang S[1], Ma K[1], Cohen J[1], Ho V[1], Xu J[1], Gauthier L[1], O'Connor C[1], Ge L[1], Woodman K[1], Lek M[1]
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P300 Relationships with health-related quality of life in FKRP-related limb-girdle muscular dystrophy R9: a prospective study

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P301 Novel JAG2 variants in the first identified Dutch patient with limb-girdle muscular dystrophy R27 and a neuropsychiatric phenotype

Schrama E[1], Niks E[1], van Duinen S[2], van der Beek N[3], Kriek M[4], Badrising U[1], **Van Duyvenvoorde H**[4]
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P302 LGMD standard of care survey for patients: aiming to better understand current care practices and identify needs in care globally

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P303 Latin-Seq: a new collaborative network to provide genetic diagnosis to patients with neuromuscular diseases in Latin-America

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P304 JOURNEY: a multicenter, longitudinal natural history study of limb girdle muscular dystrophy

Lowes L[1], Vincent R[2], Stevenson H[2], Hu W[2], Comi G[3,4]
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P305 The first standards of care guidelines for a limb girdle muscular dystrophy

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VP306 Computed tomography with color reconstruction in a group of R9 limb-girdle muscular dystrophy patients with c.1387A>G mutation

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P307 Bicistronic FKRP/FST gene therapy fully recovers normal ambulation and induces supranormal muscle strength in the FKRP(P448L) model of limb girdle muscular dystrophy 2I (LGMD2I)

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P308-P316, VP317, P318: Facioscapulohumeral muscular dystrophy

P308 Development of a new DUX4-responsive reporter mouse

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P309 EPI-321: A promising gene therapy for Facioscapulohumeral muscular dystrophy (FSHD) targeting D4Z4 epigenome

Adhikari A[1], Boregowda S[1], Zheng H[1], Aguirre O[1], Norton A[1], Yang X[1], Luong T[1], Ko D[1], Smith L[1], Swan R[1], Jiyarom B[1], Jiang F[1], Daley T[1], Hart D[1], Liu Y[1], Collin A[1]
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P310 Direct measure of D4Z4 repetition in FSHD1 patients by applying comprehensive BLAST using nanopore sequencing

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P311 Facioscapulohumeral muscular dystrophy European patient survey: assessing patient preferences in clinical trial participation

McNiff M[1], Hawkins S[2], Haase B[2], Bullivant J[1], McIver T[3], Mitelman O[4], Emery N[5], Tasca G[1,6], Voermans N[2,7], Diaz-Manera J[1]
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P312 Quality of life and support needs in children and adolescents with facioscapulohumeral dystrophy, a qualitative study

Dijkstra J, Rasing N, Boon E, Cup E, Altena-Rensen S, Lanser A, van Engelen B, Ramakers A, Erasmus C, **Voermans N**[1]
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P313 Radiological and circulating biomarkers in Facioscapulohumeral muscular dystrophy: a longitudinal study

Bortolani S[2], Monforte M[2], Pescatori M[2], Ielpo S[2], Palazzo A[2], Mosca N[2], Torchia E[2], Tartaglione T[2], Ricci E[2], **Tasca G**[1]
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P314 A systematic literature review to assess the level of evidence in Facioscapulohumeral Muscular Dystrophy

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P315 Safety and tolerability of Losmapimod for the treatment of FSHD

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P316 Disability questionnaire of FSHD1 correlates with the in-person examination

Lee J[1], Shin J[2], Nishino I[3], Lee Y[4], Kim Y[1]
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VP317 Sex-related utilization differences in the 12-months after a diagnosis of Facioscapulohumeral muscular dystrophy (FSHD)

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P318 Reduced calpain expression in a patient with fascioscapulohumeral muscular dystrophy

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P398-P413, VP414, P415, VP416-VP417, P418-P420: Congenital myopathies

P398 Phenotypic and genotypic spectrum of a cohort with centronuclear myopathy in the Western Cape, Southern Africa

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P399 Kbtbd13 knock-down prevents and reverts phenotype development and progression in a nemaline myopathy type 6 mouse model

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P400 Kbtbd13R408C-knockin mouse model elucidates mitochondrial pathomechanism in NEM6

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P401 Defective lysosomal positioning and mobility in a skeletal muscle model of X-linked myotubular myopathy using human iPS cells

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P402 Nemaline myopathy type 6 caused by variants in the KBTBD13 gene: A cross-sectional study of 24 patients

***Van Kleef E**[1], Bouman K[1], Molenaar J[1], Küsters B[1], Groothuis J[1], Olivé M[2], Malfatti E[3], Kamsteeg E[1], Van Engelen B[1], Ottenheijm C[4], Doorduin J[1], Voermans N[1]*
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P403 The replacement kinetics of the giant muscle protein nebulin are slow and further reduced by a frequently observed mutation in Neb

***Bogaards S**[1], Yuen M[1], Onderwater Y[1], Clara C[1], Galli R[1], Vizoso M, Conijn S[1], Peters E, Nahidi L[1], Jalink K, van Rheenen J, Granzier H, Ottenheijm C[1]*
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P404 Deep phenotyping and characterization of a patient with a novel autosomal dominant TNNI1-related hypercontractile muscle disease

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P405 First clinical and myopathological description of a congenital myopathy based on a homozygous variant in TNNI2

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P406 Identification of a deep-intronic variant that results in a pseudoexon in an individual with NEB-related myopathy

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P407 Kbtbd13R408C-knockin mouse model reveals impaired relaxation kinetics as novel pathomechanism for NEM6 cardiomyopathy

Baelde R[1], Janssen V[1], Fortes Monteiro A[1], Galli R[1], Methawasin M[2], Granzier H[2], Kuster D[1], van der Velden J[1], Ottenheijm C[1], de Winter J[1]
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P408 Clinical and pathologic characterization of a novel homozygous CFL2 mutation in a patient with nemaline myopathy type 7

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P409 Lessons learnt from trials in centronuclear myopathies: A qualitative study from the patient perspective

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P410 Liver involvement in Myotubular and Centronuclear Myopathy: data from the MTM & CNM patient registry

Bullivant J[1], **Ward E**[2], Lennox A[3], Lawlor M[4], Jungbluth H[5,6], Beggs A[7], Graham R[8], Heidemann M[9], Wood M[2], Page J[1], Cowling B[10], Voermans N[11], Foley R[12], Dowling J[13], Marini Bettolo C[1], Kyrana E[14], Dhawan A[14]
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P411 Pure electrophysiologic myotonia without clinical myotonia in a patient with a novel mutation in DNM2 gene and pathological evidence for centronuclear myopathy

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P412 Expanding the clinical and genetic spectrum of biallelic pathogenic MYO18B variants in congenital myopathy

Donkervoort S[1], Zaharieva I[2], Essid M[3], Longman C[4], Foley A[1], Horrocks I[5], Benrhouma H[3], Farrugia M[6], Neuhaus S[1], Younes T[3], Youssef-Turki I[3], Jamshidi Y[7], Chao K[8], Houlden H[9], Maroofian R[9], Bönnemann C[1], Muntoni F[2,10,11], Sarkozy A[2]
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P413 Dominantly inherited myosin IIa myopathy without ophthalmoplegia caused by aberrant splicing of MYH2

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VP414 Pancreatitis in RYR1-related disorders

Famili D[1], Mistry A[2], Treves S[3], Tribe R[2], Kyrana E[4], Dhawan A[4], Goldberg M[5], Voermans N[6], Willis T[7], Jungbluth H[1,8]
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P415 Mitoquinol Mesylate and PUFA: an alternative therapeutic approach for RYR1-related myopathies

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VP416 Systemic NAD+ deficiency reveals a potential therapeutic target for RYR1-related myopathies

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VP417 Obstetric and gynaecological features in females carrying mutations in the skeletal muscle ryanodine receptor (RYR1) gene: a questionnaire study

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[1]*Department of Women and Children's Health, School of Life Course Sciences, Faculty of Life Sciences and Medicine, Kings College London,* [2]*Malignant Hyperthermia Investigation Unit, Department of Anesthesiology, Canisius Wilhelmina Hospital,* [3]*Department of Biomedicine, University Hospital Basel,* [4]*RYR1 Foundation,* [5]*Department of Neurology, Radboud University Medical Centre,* [6]*Department of Paediatric Neurology, Neuromuscular Service, Evelina Children's Hospital, Guy's & St Thomas' NHS Foundation Trust,* [7]*Randall Centre for Cell and Molecular Biophysics, Muscle Signalling Section, Faculty of Life Sciences and Medicine (FoLSM), King's College London*

P418 Dominant cardioskeletal titinopathies reflect distinct mechanisms of disease

J. Roggenbuck[1], *J. Gohlke* [2], *Z. Hourani* [2], *S. Heintzman* [1], *A. Burghes* [1], *J. Lindqvist*[2], *H. Granzier*[2]
[1]*The Ohio State University Wexner Medical Center,* [2]*University of Arizona, Tucson*

P419 Broad A-band myopathy in a patient with TTN variants

Klotz J[1], *Vogel H*[1], *Mrak R*[2], *Tesi Rocha C*[1]
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P420 Titinopathy Biannual International Case and Scientific Conferences illuminate understanding phenotypic and genetic diversity in titin (TTN) - related disorders

Foye S[1], *Savarese M*[2], *Udd B*[3]
[1]*Team Titin* [2]*Folkhälsan Research Center & University of Helsinki, Helsinki, Finland,* [3]*Folkhälsan Research Center & University of Helsinki and Tampere Neuromuscular Center, Helsinki/Tampere, Finland*

18:15-18:45	**Short Oral Presentations 4** Ballroom C1 **P398-P403** *Moderator: Anna Sarkozy, Dubowitz Neuromuscular Centre, Great Ormond Street Hospital, UK*	**Short Oral Presentations 5** Ballroom C2 **P168, P281-P285** *Moderator: Meredith James, John Walton Muscular Dystrophy Research Centre, UK*	**Short Oral Presentations 6** Ballroom C3 **P169-P172, P308-P309** *Moderator: Vishnu Venugopalan Thampy Yamuna, All India Institute of Medical Sciences, India*
19:15-20:15	**Industry Symposium 5** PAC		**Industry Symposium 6** Exhibit Hall A1

Thursday 5th October 2023

07:00-15:00	**Registration desk open**	
08:00-09:00	**Industry Symposium 7** PAC	**Industry Symposium 8** Exhibit Hall A1
09:30-11:00	PAC **Topic 2: Pathobiology of neuromuscular repeat expansion disorders 1** *Moderators: Ichizo Nishino, National Institute of Neuroscience, NCNP, Japan & Louise Benarroch, Centre De Recherche En Myologie, France*	
09:30-10:00	**INV08: RAN translation in C9orf72 ALS/FTD and other repeat opportunities** ***Ranum L*** *[1]University of Florida*	
10:00-10:30	**INV09: Novel repeat disorders in muscle disease: the emergence of OPDM** ***Wang Z*** *[1]Department of Neurology, Peking University First Hospital*	
10:30-10:45	**O07: RAN translation of expanded CGG repeat in LRP12 may contribute to oculopharyngodistal myopathy** ***Li C***[1], *Pittman S[1], Maltby C[2], Todd P[2], Weihl C[1]* *[1]Department of Neurology, Washington University School of Medicine, [2]Department of Neurology, University of Michigan*	
10:45-11:00	**O08: Single-nucleus RNA sequencing reveals characteristic gene expression in pathologically-specific myofibers in oculopharyngodistal myopathy** ***Eura N***[1,2], *Noguchi S[1], Hayashi S[1], Nishino I[1]* *[1]Department of Neuromuscular Research, National Center of Neurology and Psychiatry, [2]Department of Neurology, Nara Medical University*	
11:00-11:30	**Morning refreshments & exhibition** Exhibit Hall **and posters** Ballroom	
11:00-11:30	**Guidelines Committee** find out about how to get involved Myology Cafe, Exhibit Hall	
11:30-13:30	PAC **Topic 2: Pathobiology of neuromuscular repeat expansion disorders 2** *Moderators: Gauthier Remiche, Hopital Erasme, Belgium & Nicol Voermans, Radboud University Medical Center, The Netherlands*	
11:30-12:00	**INV10: Genetic discovery and pathomechanism of repeat disorders in neuromuscular diseases: lessons from RFC1** ***Houlden H***[1] *[1]UCL Queen Square, Institute of Neurology*	
12:00-12:30	**INV11: Motor neuron and muscle involvement in SBMA: therapeutic implications** ***Fischbeck K***[1] *[1]National Institutes of Health (NIH)*	
12:30-12:45	**O09: Bi-allelic variants of FILIP1 cause congenital myopathy, dysmorphism and neurological defects** ***Roos A***[1,2,3], *van der Ven P[4], Alrohaif H[5], Kölbel H[1], Heil L[4], Della Marina A[1], Weis J[6], Töpf A[5], Vorgerd M[2], Schara-Schmidt U[1], Gangfuss A[1], Evangelista T[7], Hentschel A[8], Grüneboom A[8], Fuerst D[4], Kuechler A[9], Tzschach A[10], Depienne C[9], Lochmüller H[3]* *[1]University Medicine Essen, [2]University Hospital Bergmannsheil, Heimer Institute for Muscle Research, Bochum, [3]Children's Hospital of Eastern Ontario Research Institute, [4]University of Bonn, Institute for Cell Biology, Department of Molecular Cell Biology, [5]Newcastle University, John Walton Muscular Dystrophy Research Centre, [6]RWTH-Aachen University Hospital, Institute of Neuropathology, [7]Nord/Est/Ile-de-France Neuromuscular Reference Center, Institute of Myology, Pitié-Salpêtrière Hospital, [8]Leibniz-Institute for Analytical Science, [9]University Hospital Essen, Institute of Human Genetics, [10]Medical Center, Faculty of Medicine, Institute of Human Genetics*	
12:45-13:00	**O10: Universal genomic newborn screening for early, treatable, and severe conditions- including 33 genes of NMD: Baby Detect** ***Dangouloff T***[1], *Hovhannesyan K[1], Piazzon F[1], Mashhadizadeh D[2], Helou L[1], Palmeira L[2], Boemer F[2], Servais L[1,2,3]* *[1]University Of Liege, [2]University Hospital of Liege, [3]Oxford University*	
13:00-13:15	**O11: Long-read nanopore sequencing in FSHD patients reveals CpG methylation patterns including methylation gradients in contracted D4Z4 arrays** ***Butterfield R***[1], *Dunn D[2], Duval B[2], Moldt S[1], Weiss R[2]* *[1]University of Utah, Department of Pediatrics, [2]University of Utah, Department of Human Genetics*	

13:15-13:30	O12: Muscle imaging in natural history of FSHD: quantitative MRI and ultrasound results compared head-to-head ***Vincenten S***[1], *Voermans N*[1], *van Engelen B*[1], *Mul K*[1], *van Alfen N*[1] [1]*Radboudumc*
13:30-14:45	**Lunch & exhibition** Exhibit Hall **and posters** Ballroom
13:45-14:45	**Career Development Workshop** Ballroom C1 (Lunch available in the room) *Moderator: Chris Weihl, Washington University in St. Louis, USA* *Panel: Meredith James, John Walton, Muscular Dystrophy Research Centre, UK,* *Mike Lawlor, Medical College of Wisconsin, USA,* *Coen Ottenheijm, Amsterdam UMC, The Netherlands* *and Carmen Paradas, Hospital Virgen del Rocío, Spain*
14:45-18:00	Poster viewing / Group Activity (separate registration required)
18:00-21:00	Group Activity Reception (separate registration required)

Friday 6th October 2023

06:45-18:00	**Registration desk open**
07:30-08:30	**Interesting Case Discussions** PAC (Refreshments available) *Cases presented by delegates* *Moderators: Reghan Foley, National Institute of Health, USA and Riyad El-Khoury, Genethon, France*
08:30-08:45	Comfort break
08:45-10:00	PAC **Topic 3: The effect of lifestyle, exercise and nutrition on neuromuscular pathology and outcomes 1** *Moderators: Salman Bhai, UT Southwestern, USA & Jean-Yves Hogrel, Association Institut de Myologie, France*
08:45-09:15	INV12: The exposome in neuromuscular disorders ***Feldman E***[1] [1]*University of Michigan*
09:15-09:45	INV13: Development of a cycle training paradigm to improve exercise capacity and pathophysiology in boys with Duchenne muscular dystrophy ***Taivassalo T*** [1]*University of Florida*
09:45-10:00	O13: Promoting an active lifestyle; use of an in-home body weight support system to increase exercise dosage for children with neuromuscular disease ***Iammarino M***[1], *Reash N*[1], *Wendland M*[2], *Alfano L*[1], *Lowes L*[1] [1]*Nationwide Children's Hospital,* [2]*Cleveland State University*
10:00-10:30	**Morning refreshments & exhibition** Exhibit Hall **and posters** Ballroom
10:00-10:30	**Myology Developments Across the World and Education Committees** find out about how to get involved Myology Café, Exhibit Hall
10:30-12:00	PAC **Topic 3: The effect of lifestyle, exercise and nutrition on neuromuscular pathology and outcomes 2** *Moderators: Linda Lowes, Nationwide Children's Hospital, USA & John Vissing, Rigshospitalet, Denmark*
10:30-11:00	INV14: Physical activity and exercise are more than medicine for neuromuscular disorders ***Voet N***[1, 2] [1]*Radboud University Medical Centre.* [2]*Rehabilitation Center Klimmendaal, Arnhem*
11:00-11:15	O14: Experiences with pregnancy and pregnancy-related physiotherapy in women with Charcot-Marie-Tooth disease. A qualitative interview study *Boda N*[2], ***Rosenberger A***[1], *Lahelle A*[1] [1]*National Neuromuscular Centre Norway, University Hospital of North-Norway,* [2]*UiT The Arctic University of Norway*
11:15-11:30	O15: Large-scale proteomics profiling of peripheral blood of DM1 patients identifies biomarkers for disease severity and physical activity ***'t Hoen P***[1], *van As D*[1], *Claeys T*[2], *Salz R*[1], *Gabriels R*[2], *Impens F*[2], *Volders P*[2], *Martens L*[2], *van Engelen B*[1], *ReCognitION consortium* [1]*Radboud university medical center,* [2]*University of Ghent*

11:30-11:45	**O16: New FDX2-loss of function phenotype presenting with blindness and myopathy with potential responsiveness to Co-enzyme Q10 analogs** ***Foley A***[1], *Maio N*[2], *Todd J*[1], *Huryn L*[3], *Saade D*[1], *Neuhaus S*[1], *Donkervoort S*[1], *Hufnagel R*[4], *Stasheff S*[5], *Orbach R*[1], *Gurgel-Giannetti J*[6], *Gropman A*[7], *Rouault T*[2], *Bönnemann C*[1] *[1]Neuromuscular and Neurogenetic Disorders of Childhood Section, NINDS, National Institutes of Health, [2]Section on Human Iron Metabolism, NICHD, National Institutes of Health, [3]Ophthalmic Clinical Genetics Section, NEI, National Institutes of Health, [4]Medical Genetics and Ophthalmic Genomics Unit, NEI, National Institutes of Health, [5]Retinal Neurophysiology Section, NEI, National Institutes of Health, [6]Department of Paediatrics, Universidade Federal de Minas Gerais, [7]Neurogenetics and Neurodevelopmental Disabilities, Children's National Medical Center*
11:45-12:00	**O17: 6'-sialyllactose supplementation in GNE myopathy: a pilot and subsequent placebo-controlled study** ***Park Y***[1], *Kim L*[2], *Shin J*[3] *[1]Department of Neurology Pusan National University Hospital, Pusan National University School of Medicine, [2]Application strategy & development division, GeneChem, Inc., [3]Department of Neurology Pusan National University Yangsan Hospital, Pusan National University School of Medicine*
12:15-13:15	**WMS General Assembly/Poster viewing for non-members** Exhibit Hall A1
13:00-14:00	**Lunch & exhibition** Exhibit Hall **and posters** Ballroom
13:30-14:00	**Sponsor Meeting** Meeting Room 10
14:00-15:00	**Poster session 3** Ballroom A-C *Refreshments served* **P68-P70, VP71, P72-81: SMA - outcome measures** **P68 Outcomes in patients with spinal muscular atrophy (SMA) and four or more SMN2 copies treated with onasemnogene abeparvovec: findings from RESTORE** ***Finkel R***[1], *Benguerba K*[2], *Gehani M*[3], *Raju D*[4], *Faulkner E*[4], *LaMarca N*[4], *Servais L*[5] *[1]St. Jude Children's Research Hospital, [2]Novartis Gene Therapies Switzerland GmbH, [3]CONEXTS-Real World Evidence, Novartis Healthcare Pvt. Ltd., [4]Novartis Gene Therapies, Inc., [5]Department of Paediatrics, MDUK Oxford Neuromuscular Centre, University of Oxford* **P69 Scoping review on the assessment tools used on SMA adolescent and adult patients** ***Hogrel J***[1], *Barrière A*[2], *Bonnyaud C*[3], *Boyer F*[4], *Gargiulo M*[1], *Li D5*, *Montagu G*[5], *Berling E*[6], *Cintas P*[7], *Le Goff L*[8], *Marchadier B*[9], *N'Dah Sekou G*[6], *Orlikowksi D*[10], *Pouplin S*[11], *Prigent H*[12], *Ropars J*[13], *Salort-Campana E*[14], *Stojkovic T*[15], *Attarian S*[14], *Laforêt P*[6] *[1]Institut de Myologie, [2]Consultations pluridisciplinaires des maladies neuromusculaires - Hôpital de la croix rousse, CHU Lyon - L'Escale, Hôpital Femme Mère Enfant, [3]Laboratoire d'analyse du mouvement, Hôpital Raymond Poincaré, APHP Paris, [4]Service de Médecine Physique et Réadaptation, CHU de Reims, [5]_unknowns SAS, [6]Service de Neurologie, Centre de référence des maladies neuromusculaires Nord Est IDF, Hôpital Raymond-Poincaré, APHP Paris, [7]Département de Neurologie, Hôpital Pierre-Paul Riquet, CHU de Toulouse, [8]Hôpital Mère-Enfant, médecine pédiatrique, CHU Nantes, [9]Roche SAS, [10]Service de Réanimation Médicale Adulte, Hôpital Raymond-Poincaré, APHP Paris, [11]Plate-Forme Nouvelles Technologies, Hôpital Raymond-Poincaré, APHP Paris, [12]Service de Physiologie et Explorations Fonctionnelles, Hôpital Raymond-Poincaré, APHP Paris, [13]Service de neurologie pédiatrique, CHU de Brest, [14]Service des Maladies Neuromusculaires et de la SLA, Hôpital de la Timone, APHM, [15]Centre de référence des maladies neuromusculaires, Hôpital de la Pitié-Salpêtrière, AP-HP* **P70 What are the priorities of adolescents and adults with SMA and their health care practitioners toward evaluation? A French qualitative study** ***Hogrel J***[1], *Berling E*[2], *Prigent H*[3], *Montagu G*[4], *Barrière A*[5], *Bonnyaud C*[6], *Boyer F*[7], *Cintas P*[8], *Gargiulo M*[1], *Le Goff L*[9], *Marchadier B*[10], *N'Dah Sekou G*[2], *Orlikowski D*[11], *Pouplin S*[12], *Pruvot A*[10], *Ropars J*[13], *Salort-Campana E*[14], *Stojkovic T*[15], *Attarian S*[14], *Laforêt P*[2] *[1]Institut de Myologie, [2]Service de Neurologie, Centre de référence des maladies neuromusculaires Nord Est IDF, Hôpital Raymond-Poincaré, APHP Paris, [3]Service de Physiologie et Explorations Fonctionnelles, Hôpital Raymond-Poincaré, APHP Paris, [4]_unknowns SAS, [5]Consultations pluridisciplinaires des maladies neuromusculaires - Hôpital de la croix rousse, CHU Lyon - L'Escale, Hôpital Femme Mère Enfant, [6]Laboratoire d'analyse du mouvement, Hôpital Raymond Poincaré, APHP Paris, [7]Service de Médecine Physique et Réadaptation, CHU de Reims, [8]Département de Neurologie, Hôpital Pierre-Paul Riquet, CHU de Toulouse, [9]Hôpital Mère-Enfant, médecine pédiatrique, CHU Nantes, [10]Roche SAS, [11]Service de Réanimation Médicale Adulte, Hôpital Raymond-Poincaré, APHP Paris, [12]Plate-Forme Nouvelles Technologies, Hôpital Raymond-Poincaré, APHP Paris, [13]Service de neurologie pédiatrique, CHU de Brest, [14]Service des Maladies Neuromusculaires et de la SLA, Hôpital de la Timone, APHM, [15]Centre de référence des maladies neuromusculaires, Hôpital de la Pitié-Salpêtrière, AP-HP Paris* **VP71 Exploring the construct validity and reliability of sensor-based measurements derived from active motor assessments in adult walkers with SMA** ***Arteaga Bracho E***[1], *Zhu C*[1], *Cosne G*[1], *Mazza C*[1], *Karatsidis A*[1], *Penalver-Andres J*[1], *Erb K*[1], *Freigang M*[2], *Lapp H*[2], *Thiele S*[3], *Wenninger S*[3], *Jung E*[4], *Campbell N*[1], *Petri S*[5], *Weiler M*[4], *Kleinschnitz C*[6], *Walter M*[3], *Günther R*[2], *Belachew S*[1], *Hagenacker T*[6] *[1]Biogen, [2]University Hospital Carl Gustav Carus at Technische Universität, [3]Friedrich Baur Institute at the Department of Neurology, University Hospital, [4]Heidelberg University Hospital, [5]Klinik für Neurologie mit Klinischer Neurophysiologie, Medizinische Hochschule, 6Universitätsklinikum* **P72 Sensor-derived measurements of upper and lower extremity function in people with type II and III SMA** *Erb K*[1], *Liu X*[1], *Zhu L*[1], *Arteaga E*[1], *Campbell N*[1], *Daron A*[2], *Poleur M*[2], *Mazza C*[1], *Nguyen C*[1], ***Servais L***[2,3,4] *[1]Biogen, Inc., [2]Centre de reference de maladies neuromusculaires, CHR-La Citadelle, [3]Institute of Myology, [4]University of Oxford*

P73 Newborn screening programs for spinal muscular atrophy worldwide: are we there yet?

Vrščaj E[1], *Dangouloff T*[2], *Osredkar D*[1], *Servais L*[2,3]
[1]*Department of Paediatric Neurology, University Children's Hospital, University Medical Centre Ljubljana,* [2]*Centre de Référence Liégeois des Maladies Neuromusculaires, Centre Hospitalier Régional de la Citadelle,* [3]*Specialised Translational Research Oxford Neuromuscular Group, Department of Paediatrics, University of Oxford*

P74 Assisted Six Minute Cycle Test (A6MCT): A Feasible and Valid Measurement of Functional and Fatigue Changes in Individuals with Spinal Muscular Atrophy

Tang W[1], *Montalvo S*[2], *De Monts C*[1], *Dunaway Young S*[1], *Ataide P*[1], *Ni Ghiollagain N*[1], *Stevens V*[1], *Parker D*[1], *He Z*[1], *Tesi Rocha C*[1], *Day J*[1], ***Duong T***[1]
[1]*Division of Neurology and Neurological Sciences, Department of Medicine, Stanford University, Stanford, CA, USA,* [2]*Division of Cardiovascular Medicine, Department of Medicine, Stanford University, Stanford, CA, USA*

P75 Revised Hammersmith Scale item achievement by functional status in an international cohort of untreated SMA 2 and 3 patients

Ramsey D[1,2], ***Stimpson G***[1], *Wolfe A*[1,3], *O'Reilly E*[1,3], *Rowher A*[1,3], *Muni Lofra R*[4], *Coratti G*[5,6], *Duong T*[7], *Dunaway Young S*[7], *Gee R*[8], *Baranello G*[1,3], *Scoto M*[11], *the RHS Working Group, Finkel R*[9,10], *Mercuri E*[5,6], *Muntoni F*[1,3], *on behalf of the international SMA consortium*
[1]*Dubowitz Neuromuscular Centre; UCL Great Ormond Street Institute of Child Health,* [2]*University of Suffolk,* [3]*NIHR Great Ormond Street Hospital Biomedical Research Centre, UCL Great Ormond Street Institute of Child Health,* [4]*John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University and Newcastle upon Tyne Hospitals NHS Foundation Trust,* [5]*Paaediatric Neurology Unit, Catholic University,* [6]*Centro Clinico Nemo, U.O.C. Neuropsichiatria Infantile Fondazione Policlinico Universitario Agostino Gemelli IRCCS,* [7]*Stanford University,* [8]*Lucille Packard Childrens Hospital,* [9]*Nemours Children's Hospital and University of Central Florida College of Medicine,* [10]*St. Jude Children's Research Hospital,* [11]*Dubowitz Neuromuscular Centre; UCL Great Ormond Street Institute of Child Health*

P76 The Canadian neuromuscular disease registry: a national spinal muscular atrophy registry for real world evidence

Sobey M[1], *Hodgkinson V*[1], *Westbury G*[1], *Brais B*[2], *Campbell C*[3], *Castro-Codesal M*[4], *Crone M*[5], *Dojeini S*[6], *Genge A*[7], *Gonorazky H*[8], *Johnston W*[9], *Kolski H*[4], *Lochmuller H*[10], *Mah J*[11], *McAdam L*[12], *O'Connell C*[13], *O'Ferrall E*[2], *Oskoui M*[14], *Pfeffer G*[1], *Phan C*[15], *CNDR SMA Investigator Network*
[1]*Department of Clinical Neurosciences, Hotchkiss Brain Institute, University of Calgary,* [2]*Centre de Réadaptation Lucie-Bruneau, Montreal Neurological Institute, McGill University,* [3]*London Health Sciences Centre, Western University,* [4]*Univ Alberta, Calgary, Canada,* [5]*Jim Pattison/Kinsen Children Centre, Univ Saskatchewan, Canada,* [6]*Ottawa Rehab Centre, Univ Ottawa, Canada,* [7]*Montreal Neurological Inst-Hosp, McGill Univ, Montreal, Canada,* [8]*Sick Kids Hospital, University of Toronto,* [9]*Kaye Edmonton Clinic, Univ Alberta, Edmonton, Canada,* [10]*The Ottawa Hospital, University of Ottawa,* [11]*Alberta Children's Hosp, Univ Calgary, Canada,* [12]*Holland Bloorview, Kids Rehab Centre, Univ Toronto, Canada,* [13]*Stan Cassidy Rehabilitation Centre, Dalhousie University,* [14]*Montreal Children's Hospital, McGill University,* [15]*Kaye Neuromusc Centre, Univ Alberta, Edmonton, Canada*

P77 A 12-tier functional scale for Spinal Muscular Atrophy

Moore Burk M[1], ***Crawford T***[2], *Krosschell K*[3,4], *Apkon S*[1,5]
[1]*Children's Hospital Colorado,* [2]*The Johns Hopkins Hospital,* [3]*Ann & Robert H. Lurie Children's Hospital of Chicago,* [4]*Northwestern Feinberg School of Medicine,* [5]*University of Colorado School of Medicine*

P78 CuidAME: Three-year Spanish longitudinal project to collect data on patients with spinal muscular atrophy

Nascimento A[1], *Exposito J*[1], *Segovia-Simón S*[1], *Puig-Ram C*[1], *Fernández - Cuesta J*[2], *Fernandez Garcia M*[2], *Lungo C*[3], *Pitarch-Castellano I*[3], *Pascual S*[2], *Marco C*[4], *Gonzalez L*[4], *Povedano M*[4], *Ballester A*[5], *Martinez E*[5], *Pareja A*[6], *López-Lobato M*[6], *Alvarez M*[7], *Grimalt M*[8], *Costa J*[3]
[1]*Hospital Sant Joan De Déu, Paediatric Neurologist,* [2]*Paediatric Neurology department, Hospital la Paz,* [3]*Paediatric Neurology department, Hospital Universitario i Politécnico La Fe,* [4]*Neurology department, Hospital Universitari de Bellvitge,* [5]*Paediatric Neurology department, Hospital Clínico Universitario Virgen de la Arrixaca ,* [6]*Paediatric Neurology department, Hospital Universitario Virgen del Rocío,* [7]*Paediatric Neurology department, Vall d'Hebron Institut de Recerca (VHIR),* [8]*Paediatric Neurology department, Hospital Universitari Son Espases*

P79 Real-world outcomes of disease-modifying treatment for patients with spinal muscular atrophy: findings from a global retrospective chart review

Dabbous O[1], *LaMarca N*[1], *Toro W*[1], *Wallach S*[1], ***Mumneh N***[1], *Aassi M*[1], *O'Brien E*[2], *Baranello G*[3,4], *Reyna S*[1]
[1]*Novartis Gene Therapies, Inc.,* [2]*Novartis,* [3]*The Dubowitz Neuromuscular Centre, Developmental Neuroscience Research and Teaching Department, UCL Great Ormond Street Institute of Child Health,* [4]*NIHR Great Ormond Street Hospital Biomedical Research Centre & Great Ormond Street Hospital NHS Foundation Trust*

P80 Beyond the clinic: multiday analysis of leg movement quantity and kinematic characteristics in infants with SMA

McIntyre M[1], *Duong T*[2], *Oh J*[3], *Wilson A*[1], *Moldt S*[1], *Moore Burk M*[4], *Tesi Rocha A*[2], *Wong K*[1], *Loftus M*[1], *Manberg S*[1], *Butterfield R*[1], *Smith B*[5]
[1]*Division of Paediatric Neurology, University of Utah,* [2]*Department of Neurology, Neuromuscular Division, Stanford University,* [3]*Division of Developmental-Behavioral Paediatrics, Children's Hospital Los Angeles,* [4]*Division of Physical Medicine and Rehabilitation, Children's Hospital Colorado,* [5]*Developmental Neuroscience and Neurogenetics Program, The Saban Research Institute, Division of Developmental-Behavioral Paediatrics, Children's Hospital Los Angeles, and Department of Paediatrics, Keck School of Medicine, University of Southern California*

P81 Gastrointestinal assessment in Spinal Muscular Atrophy (SMA): the experience of SMA healthcare professionals in France

Gomez Garcia M[1], *Quijano-Roy S*[1], *Samarji B, Lagrue E, Blu N, Ouillade L*
[1]*APHP Raymond Poincare University Hospital, Child neurology and Paediatric ICU department Pediatrique*

P152-P162, VP163, P164-P166, VP167: Distal myopathies

P152 Post weaning Gne knock out results in dramatic reduction of sialic acid levels in postnatal mouse life but no phenotype

*Harazi A[1], Yakovlev L[1], Selke P[2], Horstkorte R[2], **Mitrani-Rosenbaum S**[1]*
[1]Institute of Gene Therapy, Hadassah Hebrew University Medical Center, [2]Institute for Physiological Chemistry, Medical Faculty, Martin-Luther-University Halle-Wittenberg

P153 The genetic profile of a South African inherited myopathies cohort

Naidu K[1,5], *Schoeman M[2], Topf A[3], ICGNMD consortium[4], Straub V[3], Heckmann J[5], Henning F[1]*
[1]Division of Neurology, Faculty of Medicine and Health Sciences, Stellenbosch University, [2]Division of Molecular Biology and Human Genetics, Stellenbosch University, [3]John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust, [4]The ICGNMD consortium – for list of Consortium members see https://www.ucl.ac.uk/genomic-medicine-neuromuscular-diseases/global-contributor-list, 5Neuroscience Institute, University of Cape Town; Division of Neurology, Department of Medicine, Groote Schuur Hospital

P154 The generation of a GNE myopathy patient-derived biobank enables the study of disease-relevant cellular phenotypes across multiple pathogenic variants

Koczwara K[1], *Lake N[1], Huang S[1], DeSimone A[1], Pajusalu S[1], Branford K[2], Hallak D[2], Woodman K[1], Xu J[1], Lek A[1], Best H[1], Habib A[3], Avelar J[3], Martin V[3], Mozaffar T[3], Shieh P[4], Weisleder N[2], Lek M[1]*
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P155 Clinical and Genetic spectrum of GNE myopathy from India

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P156 Muscle biopsy findings in a large cohort of patients affected by valosin containing protein disease: preliminary analysis of the international multicentric VCP study

Schiava M[1], *Nishino I[2], Inoue M[2], Nishimori Y[2], Saito Y[2], Polvikoski T[3], Charlton R[3], Parkhurst Y[3,29], Henderson M[3,29], Marini-Bettolo C[1], Guglieri M[1], Straub V[1], Weihl C[4], Stojkovic T[5], Villar-Quiles R[5], Romero N[5], Evangelista T[5], Pegoraro E[6], De Bleecker J[7], Monforte M[8], Malfatti E[9], Souvannanorath S[9], Severa G[9], Alonso-Jiménez A[10], Baets J[10], De Ridder W[10], De Jonghe P[10], Kierdaszuk B[11], Claeys K[12], Muelas N[13], Oldfors A[14], Rodolico C[15], Quin C[16], Dominguez C[17], Herández Lain A[17], Pál E[18], Papadimas G[19], Kushlaf H[20], Alfano L[21], Alonso-Pérez J[22], Luo S[23], Badrising U[24], Bevilacqua J[25], Nedkova-Hristova V[26], Cetin H[27], Gelpi E[27], Klotz S[27], Olivé Plana M[28], Díaz Manera - On behalf of VCP International Study Group J[1]*
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P157 Development of a new mouse model to study GNE myopathy

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P158 Heterozygous SPTAN1 frameshift mutations cause distal myopathy with neurogenic features

De Winter J[1,2,3], *Van de Vondel L[1,2], Bonne G[4], Stojkovic T[4,5], Elouej S[4], Grandi F[4], Smeriglio P[4], Palmio J[6], Johari M[7,8,9], Hackman P[7,8], Savarese M[7,8], Udd B[6,7,8,10], Meyer A[11], Nicolau S[11], Flanigan K[11,12,13], Waldrop M[11,12,13], Lognman C[14], Diaz-Manera J[15], Töpf A[15], Baets J[1,2,3]*
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P159 Lectin staining biomarkers for preclinical assessment of GNE myopathy gene therapy

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P160 A deep intronic variant c.862+870C>T correlated with GNE myopathy and thrombocytopenia

Jiao K[1,2,3], Cheng N[1,2,3], Huan X[1,2,3], Luan X[4], Fan J[5], Gao M[1,2,3], Wang N[1,2,3], Xia X[1,2,3], Luo S[1,2,3], Xi J[1,2,3], Lu J[1,2,3], Zhao C[1,2,3], Yue D[6], Zhu W[1,2,3]
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P161 Natural history of distal and myofibrillar myopathies assessed by clinical and technological outcome measures (Dista-Myo): baseline results

Bortolani S[2], Torchia E[2], Vicino A[3], Cheli M[3], Rabuffetti M[4], Marzegan A[4], Monforte M[2], Ricci E[2], Hogrel J[5], Sacconi S[6], Maggi L[3], **Tasca G**[1]
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P162 Welander distal myopathy caused by genomic deletion in the TIA1 gene

Jonson P[1], Sarparanta J[1], Rusanen S[1], Sagath L[1], Kiiski K[1], Luque H[1], Gunnarsson C[2], Danielsson O[3], Hackman P[1], Udd B[1,4]
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VP163 Filaminopathy presenting as myofibrillar myopathy with nemaline bodies and ring fibers

Cotta A[1], da Cunha Junior A[1], Carvalho E[1], da Silveira E[2], Costa e Silva C[2], da Silva Neto R[1], Cauhi A[1], Valicek J[1], Vargas A[1]
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P164 Development of a myotube model for C-terminal titin studies

Sarparanta J[1], Jonson P[1], Luque H[1], Zacchini C[1,2], Hackman P[1], Udd B[1,3]
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P165 Rare ACTN2 frameshift variants resulting in a protein extension cause distal myopathy and Hypertrophic Cardiomyopathy through protein aggregation mechanism

Ranta-aho J[1,2], Jonson P[1,2], Sarparanta J[1,2], Tasca G[3,4], Yvorel C[5], Harzallah I[6], Pais L[7,8], Austin-Tse C[7,9], Ganesh V[7,10], O'Leary M[7], Rehm H[7,9], Savarese M[1,2], Udd B[1]
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P166 Deciphering the genetic cause of Oculopharyngodistal myopathy in a French cohort using Cas9-targeted long-read sequencing

Benarroch L[1], Nelson I[1], Stojkovic T[2], Mohand Oumoussa B[3], Madry H[3], Boelle P[4], Labreche K[4], Tomé S[1], Trollet C[1], Bonne G[1]
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VP167 Novel UNC45B compound heterozygous variants in a child with congenital heart defects and muscle weakness

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P191-P204: Registries

P191 Exercise prescription for patients with neuromuscular diseases: Lessons learned from real-world data on exercise exposure

Richardson M[1], Wong K[1], Michell-Sodhi J[1], Moat D[1], McCallum M[1], Harris E[1], Mayhew A[1], Grover E[1], Guglieri M[1], Diaz-Manera J[1], Robinson E[1], Elseed M[1], Mason J[1], Kinet V[2], Straub V[1], James M[1], Marini-Bettolo C[1], Muni-Lofra R[1]
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Continued on next page

P192 The open-access treatabolome platform enhances the visibility of treatable and actionable genes in RD-connect's GPAP and other clinical diagnosis support tools

Atalaia A[1], Thompson R[2], Matalonga L[3], Hernandez-Ferrer C[3], Corvo A[3], Carmody L[4], Zurek B[5], Ben Yaou R[1], Horvath R[6], Graessner H[5], Riess O[5], Robinson P[4], Lochmuller H[8], Beltran S[3], Bonne G[1], The Treatabolome Project Group

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P193 TREAT-NMD global registry network: an insight into a global neuromuscular patient dataset

Poll A[1], Bennett N[1], Chua Cheh H[1], Ambrosini A[2], Rodrigues M[3], Guglieri M[4]

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P194 The Duchenne registry: key milestones and advances of a patient-report registry

Martin A[1], Armstrong N[1], Quirin K[1], Freed M[1]

[1]Parent Project Muscular Dystrophy

P195 The Canadian neuromuscular disease registry: a national Duchenne muscular dystrophy registry for post-marketing surveillance

Sheriko J[1], Bourcier D[1], Hodgkinson V[2], Dyck A[2], Worsfold N, Osman H[3], Bohne L[2], Korngut L[2], on behalf of the CNDR Investigator Network

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P196 Age at loss of ambulation in patients with DMD from the STRIDE registry and the CINRG natural history study: a matched cohort analysis

Mercuri E[1], Muntoni F[2], Buccella F[3], Desguerre I[4], Kirschner J[5], Nascimento Osorio A[6], Tulinius M[7], de Resende M[8], Morgenroth L[9], Gordish-Dressman H[10], Johnson S[11], Werner C[12], Anbu B[11], Liu E[11], Rajbhandari R[11], Trifillis P[11], McDonald C[13]

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P197 Pulmonary function in patients with Duchenne muscular dystrophy from the STRIDE registry and CINRG natural history study: a matched cohort analysis

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P198 Updated demographics and safety data from patients with nonsense mutation Duchenne muscular dystrophy receiving ataluren in the STRIDE registry

Muntoni F[1], Buccella F[2], Desguerre I[3], Kirschner J[4], Nascimento Osorio A[5], Tulinius M[6], de Resende M[7], Johnson S[8], Werner C[9], Anbu B[8], Liu E[8], Rajbhandari R[8], Trifillis P[8], Mercuri E[10]

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P199 Frequency of regular echocardiography monitoring in patients with myotonic dystrophy type 1

Bovenkerk D[1,3], Janssen C[1], Van den Heuvel F[2], Joosten I[1], Den Uijl D[3], Bijvoet G[3], Van Engelen B[4], Nijveldt R[2], Evertz R[2], Faber C[1], Van Kuijk S[5], Vernooy K[3]

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P200 The UK myotonic dystrophy patient registry - empowering clinical research and patient voice with an effective translational research tool

Muni Lofra R[1], Walker H[1], Turner C[2], Adcock K[3], Ashley E[4], Rogers M[5], Orrell R[6], Donachie J[7], Monckton D[8], Hamilton M[9], Hewamadduma C[10], Bowler M[11], Sodhi J[1], Marini-Bettolo C[1]

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P200 The UK myotonic dystrophy patient registry - empowering clinical research and patient voice with an effective translational research tool

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P200 The UK myotonic dystrophy patient registry - empowering clinical research and patient voice with an effective translational research tool

Muni Lofra R[1], Walker H[1], Turner C[2], Adcock K[3], Ashley E[4], Rogers M[5], Orrell R[6], Donachie J[7], Monckton D[8], Hamilton M[9], Hewamadduma C[10], Bowler M[11], Sodhi J[1], Marini-Bettolo C[1]
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P201 eHealth in Myotonic Dystrophy type 1: validation of two mobile ECG registration methods for detecting conduction disorders

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P202 The UK Facioscapulohumeral Muscular Dystrophy Patient Registry: a powerful tool to support clinical research and patient voice in the translational research pathway

Muni Lofra R[1], Walker H[1], Orrell R[2], Graham A[3], Norwood F[4], Roberts M[5], Willis T[6], Matthews E[7], Mencias M[7], Adcock K[8], Marini Bettolo C[1]
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P203 Proven interoperability of five neuromuscular rare disease registries

't Hoen P[1], Lalout N[1,2], Vroom E[2], Franken M[2], Jäger D[3], Tassoni A[3], Kampowski T[3], Delattre H[4], Hamroun D[4], Molthof R[5], de Jong I[5], Quemada E[6], Atalaia A[7], Evangelista T[7], Wilkinson M[6], EURO-NMD registry consortium
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P204 PROMs collection and the UK Spinal Muscular Atrophy Patient Registry

Muni Lofra R[1], Murphy L[1], Elwell T[1], Segovia S[1], Yau J[1], Cavalcante E[2], Madden M[2], Adcock K[3], Farrugia M[4], Irwin J[5], Lilleker J[6], McConville J[7], Merrison A[8], Parton M[9], Ryburn L[10], Scoto M[2], Marini-Bettolo C[1]
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P252-P258, VP259, P260, P262-P268: Dystrophinopathies

P252 Exploration of muscle MR imaging and clinical outcome measures in adults with Becker muscular dystrophy

De Wel B[1,2], Iterbeke L[2], Huysmans L[3,4], Peeters R[5], Goosens V[5], Ghysels S[5], Byloos K[5], Putzeys G[5], Dubuisson N[6], van den Bergh P[6], Van Parijs V[6], Remiche G[7], Maes F[3,4], Dupont P[8], Claeys K[1,2]
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P253 Motor function and genotype-phenotype correlations in paediatric Becker muscular dystrophy

Zygmunt A[1,2], Shiuan Y[2], Horn P[1,2], Rybalsky I[1], Reebals L[1], Tian C[1,2]
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P254 Characterization of short- and long-term proteomic response to the fast skeletal myosin inhibitor, EDG-5506, in Becker muscular dystrophy (BMD)

Barthel B[1], Madden M[1], Thaler L[1], Evanchik M[1], Koch K[1], Donovan J[1], Collins S[1], Phan H, Russell A[1]
[1]Edgewise Therapeutics

P255 Effects of EDG-5506, a fast myosin modulator, on function and biomarkers of muscle damage in adults with Becker muscular dystrophy (BMD)

Collins S[1], Phan H[2], Russell A[1], Barthel B[1], Thaler L[1], Kilburn N[1], Mancini M[1], MacDougall J[1], Donovan J[1]
[1]Edgewise Therapeutics, [2]Rare Disease Research

Continued on next page

P256 Development of a conceptual model of the patient experience of Becker Muscular Dystrophy (BMD) – a literature review and qualitative interview study

Bronson A[1], Collins S[1], Aldhouse N[2], Marshall C[2], Al-zubeidi T[2], Thursfield M[2]
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P257 Genotypic spectrum of Duchenne and Becker muscular dystrophy (DMD/BMD) in an Indian, South African and Brazilian cohort

Perry L[1,2], Reyaz A[3], Human R[4], Lubbe E[4], Raga S[5], Naidu K[6,7], Tomaselli P[8], The ICGNMD Consortium[9], Vandrovcova J[10], Hanna M[10,11], Marques Jr W[8], Henning F[7], Heckmann J[6], Wilmshurst J[5], Vishnu V[3], Srivastava M[3], Yareeda S[12], Smuts I[4], **Sarkozy A**1, Muntoni F[1,2]
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P258 Clinical profile of Duchenne muscular dystrophy associated with in-frame deletions in DMD

Zygmunt A[1,2], Horn P[1,2], Rybalsky I[1], Reebals L[1], Bange J[1], Tian C[1,2]
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VP259 Decoding Duchenne muscular dystrophy: insights from single nuclei RNA-seq analysis

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P260 The vitamin B3 analogue nicotinamide riboside has only very minor effects on reducing muscle damage in mdx mice

Stan T[1], v.d. Vijver D[1], Verhaart I[1], Aartsma-Rus A[1]
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P261 The role of pathological miRNAs in Duchenne and Becker muscular dystrophy

Fiorillo A[1,2], McCormack N[1], Calabrese K[3], Heier C[1,2]
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P262 Validation lab: allowing standardized in vitro and in vivo experiments for candidate treatments for Duchenne muscular dystrophy

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P263 Targeting the innate immune system to block acute inflammatory responses and increase AAV viral transduction of skeletal muscle in mdx mice

Morales M[1], Spathis R[1], Narvesen S[1], Kuriplach D[1], Huang K[1], Bagley E[1], Eybs M[1], King M[1], Sundar T[1], Shulman D[1], MacKinnon A[2], Lawlor M[3], **Nagaraju K**[1]
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P264 Using antisense oligonucleotide therapy to rescue dystrophin (DMD) in the central nervous system in the mdx23 mouse model of Duchenne muscular dystrophy

Aghaeipour A[1,5], Gileadi T[1,5], Fergus C[2], Mitsogiannis M[3], Siddle M[1,5], Chambers D[1], **Catapano F**[1], Kelly V[2], Sokolowska E[3], Malmberg A[6], Morgan J[1,5], Ferretti P[4], Phadke R[1], Montanaro F[1,5], Muntoni F[1,5]
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P265 Cerebral damage in Duchenne muscular dystrophy: a multimodal MRI study

Brito M[1], Rezende T[1], Iwabe C[1], Conte G[1], Nucci A[1], Cendes F[1], **Cavalcante França Jr M**[1]
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P266 Gene expression profiles and spatial localisation of dystrophin isoforms in developing and adult human brain

Catapano F[1,2,3], Chambers D[1,2,3], Alkharji R[4,5], Singh S[1,2,3], Mueller J[1,2,3], Morgan J[1,2], Ferretti P[4], Malhotra J[6], Phadke R[1,2,3], Muntoni F[1,2]
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P267 Chronodisruption as a biomarker in Duchenne muscular dystrophy

Alexander M[1], Monreal-Gutierrez M[2], Reid A[1], English K[1], Wolff C[2], Lopez M[1], Siegel B[3], Phan H[4], Gamble K[5], Esser K[2]
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P268 Decoding the transcriptome of Duchenne muscular dystrophy to the single nuclei level reveals clinical-genetic correlations

Diaz-Manera J[1], Suarez-Calvet X[2], Fernández-Simón E[1], Natera D[3], Jou C[3], Codina A[3], Ortez C[3], Piñol-Jurado P[1], Guglieri M[1], Straub V[1], Nascimento A[3]
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P368, VP369, P370-P380, VP381, P382: Pompe disease

P368 Long-term enzyme replacement therapy with alglucosidase alfa in children and adults with late-onset Pompe disease

Theunissen M[1], van Kooten H[1], Harlaar L[1], Ismailova G[1], van den Hout J[2], Rizopoulos D[3], Boon M[1], Brusse E[1], van Doorn P[1], van der Ploeg A[2], van der Beek N[1]
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VP369 Disease burden, treatment patterns and healthcare resource utilization associated with Pompe disease in Sweden: a real-world evidence study

Lindberg C[1], Nordin S[2], Stelmaszuk M[3], MacCulloch A[4], Graham R[4], Ekström A[5], Lindvall B[6], Freilich J[3]
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P370 Evaluation of a humanized TfRC Pompe disease mouse model for anti-TfRC-GAA tissue delivery to both skeletal muscle and CNS

Allen E[1], Zhao S[1], Riley R[1], Smith L[1], George K[1], Leksa N[1], van der Flier A[1]
[1]Sanofi

P371 Switching treatment to cipaglucosidase alfa+miglustat positively affects motor function and quality of life in patients with late-onset Pompe disease

Claeys K[1], Byrne B[2], Diaz-Manera J[3], Dimachkie M[4], Kishnani P[5], Kushlaf H[6], Mozaffar T[7], Roberts M[8], Schoser B[9], Hummel N[10], Holdbrook F[11], Raza S[12], Shohet S[12], Wasfi Y[11], Toscano A[13], on behalf of the ATB200-07 Study Group
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P372 Safety of home administration of cipaglucosidase alfa + miglustat in late-onset Pompe disease: results from multiple clinical trials

Byrne B[1], Diaz-Manera J[2], Goker-Alpan O[3], Mozaffar T[4], Wasfi Y[5], Sitaraman Das S[5], Fox B[5], Holdbrook F[5], Jain V[5]
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P373 Effect size analysis of cipaglucosidase alfa + miglustat versus alglucosidase alfa in ERT-experienced adults with late-onset Pompe disease in PROPEL

Diaz-Manera J[1], Bratkovic D[2], Byrne B[3], Claeys K[4], Dimachkie M[5], Kushlaf H[6], Kishnani P[7], Laforêt P[8], Mozaffar T[9], Roberts M[10], Toscano A[11], Castelli J[12], Raza S[13], Holdbrook F[12], Sitaraman Das S[12], Wasfi Y[12], Schoser B[14]
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P374 COMET: effects of avalglucosidase alfa and treatment switch from alglucosidase alfa on week 145 QMFT individual item responses

Kishnani P[1], van der Beek N[2], An Haack K[3], Armstrong N[4], Periquet M[5], Thibault N[4], Zaher A[6], Schoser B[7], on behalf of the COMET investigators
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Continued on next page

P375 COMET: Safety of avalglucosidase alfa in patients with late-onset Pompe disease who switched treatment from alglucosidase alfa

Diaz-Manera J[1], Kishnani P[2], Ladha S[3], Miossec P[4], Armstrong N[5], Thibault N[5], Periquet M[6], Tammireddy S[5], Dimachkie M[7], Schoser B[8], on behalf of the COMET Investigator Group
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P376 COMET post hoc analysis: efficacy of long-term avalglucosidase alfa in subgroups of patients with late-onset Pompe disease

Toscano A[1], Kishnani P[2], Dimachkie M[3], Sacconi S[4], van der Beek N[5], Roberts M[6], Suwazono S[7], Choi Y[8], Sgobbi de Souza P[9], Schoser B[10], Armstrong N[11], Huynh-Ba O[12], Thibault N[11], Periquet M[13], Díaz-Manera J[14], on behalf of the COMET investigators
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P377 Patients in the Pompe registry who switched from alglucosidase alfa to avalglucosidase alfa: Real-world experience

Schoser B[1], Toscano A[2], Foster M[3], Periquet M[4], Sparks S[3], Kishnani P[5], on behalf of the Pompe Registry Sites
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P378 The impact of COVID-19 infection(s), pandemic and associated control measures on patients with Pompe disease

Theunissen M[1], van den Elsen R[1], House T[2], Crittenden B[2], van Doorn P[1], van der Ploeg A[3], Kruijshaar M[3], van der Beek N[1]
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P379 Spanish Pompe registry: new data based on the 130 patients included

Martínez Marín R[1], Reyes-Leiva D[2], Dominguez Gonzalez C[3], Nascimento A[4], Muelas N[5], Paradas C[6], Olivé M[7], Grau J[8], Gomez M[9], Pascual Pascual S[1], Mendoza M[10], de León J[11], Gutiérrez A[12], García Antelo M[13], Pintós G[14], Alonso J[7], Blanco Lago R[15], López de Munuain A[16], Jericó I[17], Barba-Romero M[18], Segovia Simón S[19], Díaz Manera J[19]
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P380 Diaphragmatic weakness in late-onset Pompe disease: a complex interplay between lower motor neuron and muscle fibre degeneration

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VP381 The heart-musc study: hereditary neuromuscular disorders in cardiac transplant recipients

Benterud A[1], Popperud T[2], Arntzen and Section-leader K[3], Hasselberg N[4], Broch K[5], Ørstavik and Section Leader K[6]
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P382 Disease spectrum of myopathies with elevated aldolase and normal creatine kinase

Soontrapa P[1,2], Shahar S[3,4], Eauchai L[5], Ernste F[6], Liewluck T[1]
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P383-P395,VP396, P397: Myotonic dystrophy

P383 Perceptual characteristics of speech in congenital myotonic dystrophy

Berggren K[1], Foye M, Kuo C[2], Johnson N[1]
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P384 Mitochondrial dysfunction in Myotonic Dystrophy Type 2

Kleefeld F[1], Stenzel W[2], Horvath R[3], Roos A[4], Schoser B[5]
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P385 Development of an AAV-mediated Pentatricopeptide Repeat Protein (PPR) delivery system for treatment of Myotonic Dystrophy type 1 (DM1)

Imai T[1,3], Miyai M[2], Tamai T[1], Ohta M[1], Hada K[1], Yagi Y[1,3], Nakanishi O[1], Mochizuki H[2], Nakamori M[2]
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P386 Molecular biomarkers in myotonic dystrophy type 1

Slipsager A[1], Hildonen M[2], Godtfeldt Stemmerik M[1], Tümer Z[2], Dunø M[2], Birkedal U[2], Vissing J[1]
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P387 Initiation and follow-up of mexiletine treatment in adult myotonic dystrophy patients: an expert opinion

Wahbi K[1,2], **Bassez G**[2], Duchateau J[3], Salort-Campana E[4], Vicart S[5], Labombarda F[6], Sellal J[7], Deharo J[8]
[1]Centre de référence constitutif des maladies neuromusculaires, département de cardiologie, Ap-Hp Cochin, [2]Centre de référence constitutif des maladies neuromusculaires, service de neuro-myologie, Ap-Hp Pitié-Salepêtrière , [3]Service de cardiologie, électrophysiologie et stimulation cardiaque, Hôpital cardiologique du Haut l'Evêque, [4]Centre de référence neuromusculaire coordonnateur PACA Réunion Rhône alpes, service du Pr Attarian, Hôpital de la Timone, [5]Muscle Channelopathies Reference Center, Service of Neuro-Myology, Assistance Publique-Hôpitaux de Paris, University Hospital Pitié-Salpêtrière, Sorbonne Université, [6]Département de cardiologie, [7]Département de cardiologie, [8]Département de cardiologie, Hôpital de la Timone

P388 Methylphenidate use in 16 children with Myotonic Dystrophy and comorbid attention deficit hyperactivity disorder: a case series

Hendriksen J[1,2], Sweere D[1,2], Weerkamp P[1,2], Braakman H[3], Collin P[1,4], Klinkenberg S[1,5]
[1]Kempenhaeghe Center for Neurological Learning Disabilities, [2]Maastricht University. School for Mental Health and Neuroscience (MHeNs), [3]Radboud university medical center, [4]Koraal groep, 5Maastricht university medical center

P389 RevEal the burdeN on daily life for myotonic dyStrophy patients due to myotoniA: preliminary results of the ENSA survey

Sansone V[1], Ashley E[2], Montagnese F[3], Gagnon C[4], Nowak U[5], Dang U[6], Turner C[7], Nikolenko N[8], Tard C[9], **Zozulya-Weidenfeller A**[1]
[1]The NeMO Clinical Centre, Neurorehabilitation Unit, University of Milan, [2]Cure Myotonic Dystrophy UK Charity, [3]Friedrich-Baur Institut, University Clinic Munich, [4]Sherbrooke University Faculty of Medicine and Health Sciences, [5]admedicum, [6]Carleton University, [7]University College London Hospitals NHS Foundation Trust, [8]Institute of Neurology, University College London, [9]Centre Hospitalier Régional Universitaire de Lille, [10]Lupin EMEA

P390 PGN-EDODM1 nonclinical data demonstrate mechanistic and meaningful activity for potential treatment of myotonic dystrophy type 1 (DM1)

Holland A[1], Klein A[2], Lonkar P[1], Svenstrup N[1], Garg B[1], Foy J[1], Furling D[2], Goyal J[1]
[1]PepGen Inc, [2]Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie

P391 Phase 1 study to assess safety, tolerability, pharmacokinetics, and pharmacodynamics of PGN-EDODM1 in adults with myotonic dystrophy type 1 (DM1)

Shoskes J[1], Larkindale J[1], Cormier J[1], Hand H[1], Vacca S[1], Lonkar P[1], Holland A[1], Garg B[1], Foy J[1], Mellion M[1]
[1]Pepgen Inc

P392 Assessing the cognitive effect of methylphenidate treatment in childhood myotonic dystrophy type 1 and comorbid Attention Deficit Hyperactivity Disorder using eye tracking: a case report

Sweere D[1,2], Hendriksen J[1,2], Vermeulen J[2,3], Klinkenberg S[1,2,3]
[1]Kempenhaeghe Center for Neurological Learning Disabilities, [2]Maastricht University School for Mental Health and Neuroscience, [3]Maastricht University Medical Center

P393 Operationalization and quantification of initiative problems in a cohort of children with myotonic dystrophy type 1

Sweere D[1,2], Klinkenberg S[1,2,3], Vermeulen J[2,3], Braakman H[4,5], Hendriksen J[1,2]
[1]Kempenhaeghe Center for Neurological Learning Disabilities, [2]Maastricht University School for Mental Health and Neuroscience, [3]Maastricht University Medical Center, [4]Radboud University Medical Center, [5]Donders Institute for Brain, Cognition and Behavior

P394 Impact of gastrointestinal and urological problems in children with myotonic dystrophy type 1

Maagdenberg S[1], Klinkenberg S[2], van den Berg J[1], Altena-Rensen S[3], Vrijens D[4], Janssen E[5], Gierenz N[6], de Wall L[7], **Braakman H**[3]
[1]Faculty of Medicine, Radboud University, [2]Department of Neurology, Maastricht University Medical Center, [3]Department of Pediatric Neurology, Amalia Children's Hospital, Radboud University Medical Center, [4]Department of Urology, Maastricht University Medical Center, [5]Department of Pediatrics, Maastricht University Medical Center, [6]Department of Pediatric Gastroenterology, Amalia Children's Hospital, Radboud University Medical Center, [7]Department of Pediatric Urology, Amalia Children's Hospital, Radboud University Medical Center

Continued on next page

P395 Recommendations of an expert group for cardiac assessment of non-dystrophic myotonic adult patients treated with mexiletine

*Vicart S[1], **Wahbi K**[1,2], Duchateau J[3], Sellal J[4], Deharo J[5], Bassez G[6], Salort-Campana E[7], Labombarda F[8]*
[1]Muscle Channelopathies Reference Center, Service of Neuro-Myology, Assistance Publique-Hôpitaux de Paris, University Hospital Pitié-Salpêtrière, Sorbonne Université, [2]Centre de référence constitutif des maladies neuromusculaires, département de cardiologie, [3]Service de cardiologie, électrophysiologie et stimulation cardiaque, Hôpital Haut Lévèque, [4]Département de cardiologie, [5]Département de cardiologie, Hôpital de la Timone, CHU de Marseille, [6]Centre de référence constitutif des maladies neuromusculaires, service de neuro-myologie, Ap-Hp Pitié-Salepêtrière, [7]Centre de référence neuromusculaire coordonnateur PACA Réunion Rhône alpes, service du Pr Attarian, Hôpital de la Timone, CHU de Marseille, [8]Département de cardiologie, CHU de Caen

VP396 More prevalent comorbidities, healthcare costs, and service utilization in male Myotonic Dystrophy (DM) patients and female patients

*Day J[1], Munoz K[2], Chen C[2], Brook R[3], Kleinman N[3], **Cho H**[2], McEvoy B[2], Stahl M[2], Tai L[2]*
[1]Stanford Neuroscience Health Center, [2]Avidity Biosciences, Inc., [3]Better Health Worldwide

P397 Prevalence of risk factors associated with cardiovascular events in patients with myotonic dystrophy type 1

*Bruijnes J[1], **la Fontaine L**[1], Kayha K[1], Joosten I[1], Faber C[1]*
[1]Maastricht University Medical Centre

P421-P426, VP427, P428, VP429, P430-P434, VP435-436: Congenital muscular dystrophies

P421 Mitochondrial involvement in SELENON-Related Myopathy

***Barraza P**[1], Moghadaszadeh B[1], Lee W[1], Isaac B[2], Sun L[2], Troiano E[1], Rockowitz S[2], Sliz P[2], Beggs A[1]*
[1]Division of Genetics and Genomics, The Manton Center for Orphan Disease Research, Boston Children's Hospital, Harvard Medical School, [2]Division of Molecular Medicine, The Manton Center for Orphan Disease Research

P422 A robust and practical myogenic system to explore cellular and genomic features of muscle differentiation

***Benarroch L**[1], Madsen-Østerbye J[2], Abdelhalim M[2], Mamchaoui K[3], Ohana J[3], Bigot A[3], Mouly V[3], Bertrand A[1], Collas P[2], Bonne G[1]*
[1]Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, [2]Department of Molecular Medicine, Institute of Basic Medical Sciences, Faculty of Medicine, University of Oslo, [3]Sorbonne Université -Inserm-Institut de Myologie, MyoLine Immortalization Platform

P423 AAV-mediated therapeutic effect of linker protein-mediated gene therapy on muscle and nerve pathology in mouse models for LAMA2 MD

***Reinhard J**[1], Lin S[1], Rüegg M[1]*
[1]University of Basel

P424 Identify genetic modifiers controlling severity of collagen-6 related dystrophies (COL6-RD)

***Bisciglia M**[1], Stojkovic T[2], Nascimento A[3], Vissing J[4], Castiglioni C[5], Claeys K[6,7], Remiche G[1], De Paepe B[8], Butterfield R[9], Deconinck N[8,1]*
[1]Centre de Référence Neuromusculaire, Department of Neurology, Hôpital Erasme Université Libre de Bruxelles, [2]Centre de référence des maladies neuromusculaires Nord/Est/Ile de France, Hôpital Pitié-Salpêtrière, [3]Neuromuscular Disorders Unit Paediatric Neurology Department Hospital Sant Joan de Déu, [4]Neuromuscular Clinic and Research Unit Department of Neurology, Rigshospitalet, University of Copenhagen, [5]Departamento de Neurologia Pediatrica, Clinica Las Condes, [6]Department of Neurology, University Hospital of Leuven, [7]KU Leuven Laboratory for Muscle Diseases and Neuropathies, [8]Neuromuscular Reference Center, UZ Gent, [9]Department of Paediatric Neurology, University of Utah

P425 Inhibition of TGFβ signaling pathway as a therapeutic approach in collagen VI-related muscular dystrophy

*Mohassel P[1,2], **Hearn H**[1], Zou Y[2], Rooney J[2], Bönnemann C[2]*
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P426 Comparison of motor function measure-20 (MFM20) and neuromuscular gross motor outcome (NM GRO) in young children with LAMA2 or COL6-related dystrophy

*Doreswamy K[1], Foley R[2], Norato G[2], Waite M[1], Acquaye N[2], Hinkley L[2], Alfano L[3], Lowes L[3], Bonnemann C[2], **Jain M**[1]*
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VP427 The Swiss cohort of LAMA2-related muscular dystrophy patients

***Enzmann C**[1,7], Steiner L[2], Baumann D[6], Lötscher N[6], Jacquier D[3], Stettner G[4], Henzi B[1,8], Ripellino P[5], Fluss J[9], Klein A[1,2]*
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P428 Biochemical changes in chorionic villi of LAMA2-patients resemble muscle relevant disease processes

*Kölbel H[1], Hentschel A[2], Preusse C[3], Rüegg M[4], Schara-Schmidt U[1], Reinhard J[4], **Roos A**[1]*
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VP429 Impaired skeletal muscle strength in adult patients with laminopathies

Decostre V[1], Chikhaoui C[2], Vigouroux C[3], Behin A[4], Bassez G[4], Ferreiro A[4], Janmaat S[3], Masingue M[4], Stojkovic T[4], Vatier C[3], Villar Quiles R[4], Quijano Roy S[5], Wahbi K[6], Eymard B[4], Bonne G[2], Ben Yaou R[2,4], Hogrel J[1]
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P430 LMNA-related congenital muscular dystrophy: potential impact of corticosteroid treatment on contracture progression and motor function

Nascimento A[1], Carrera L[1], Natera D[1], Medina J[1], Moya O[1], Roca S[1], Sarquella G[2], Cesar S[2], Zschaeck I[1], Ríos A[1], Alvarenga N[1], Armijo J[1], Lotz S[1], Estevez B[1], Exposito J[1], Ortez C[1]
[1]Hospital Sant Joan De Déu, Neurology department. Neuromuscular unit, [2]Hospital Sant Joan de Déu. Pediatric Arrhythmias, Inherited Cardiac Diseases and Sudden Death Unit, Cardiology Department

P431 Steroid treatment may change natural history in congenital laminopathies

Gomez Garcia M[1], Garcia-Uzquiano R[1], Le Goff L, Manel V, Dabaj I, Mercier S, Ben Yaou R, Bonne G, Carlier R, Quijano-Roy S
[1]APHP Hopital Raymond Poincare, Paediatric neurology and pediatric ICU department

P432 Clinical and genetic characteristics of patients with Emery-Dreifuss muscular dystrophy from the Canary Islands carrying a probable founder mutation in the EMD gene

De León-Hernández J[1], Rodríguez-Baz I[1,2], Rodríguez-Vallejo A[1], Alemañ-Díez J[1], Hernández-Tost H[1], Castelló-López M[1], Fregel-Rodríguez C[1], González-Coello V[1], Sosa-Cabrera Y[1], Solé-Sabater M[1], Hernández-García C[3], Grillo J[3], **Alonso-perez J**[1]
[1]Neuromuscular Disease Unit. Neurology Department. Hospital Universitario Nuestra Señora de Candelaria. Fundación Canaria Instituto de Investigación Sanitaria de Canarias (FIISC), [2]Sant Pau Memory Unit, Department of Neurology, Hospital de la Santa Creu i Sant Pau, Biomedical Research Institute Sant Pau (IIB Sant Pau), Universitat Autònoma de Barcelona, [3]Cardiology Department. Hospital Universitario Nuestra Señora de Candelaria

P433 Clinical features of two patients with FHL1 related myopathy in Korea

Lee S[1], Kim S[1], Kim W[2], Park H[1], Choi Y[1]
[1]Department of Neurology, Gangnam Severance Hospital, Yonsei University College of Medicine, [2]Department of Neurology, Kangdong Sacred Heart Hospital, Hallym University College of Medicine

P434 Severe GMPPB-related congenital muscular dystrophy with rapidly progressive encephalopathy leading to infantile death

Dube J[1], Blaser S[2], Guerguerian A[3], Hazrati L[4], Yoon G[1,5]
[1]Division of Clinical and Metabolic Genetics, Department of Paediatrics, The Hospital for Sick Children, University of Toronto, [2]Division of Neuroradiology, Department of Diagnostic Imaging, The Hospital for Sick Children, University of Toronto, [3]Departments of Critical Care Medicine and Paediatrics, Neuroscience and Mental Health Program, Research Institute, The Hospital for Sick Children, University of Toronto, [4]Division of Pathology, Department of Paediatric Laboratory Medicine, The Hospital for Sick Children, University of Toronto, [5]Division of Neurology, Department of Paediatrics, The Hospital for Sick Children, University of Toronto

VP435 Study of dysphagia in Fukuyama congenital muscular dystrophy (FCMD)

Ishiguro K[1]
[1]Tokyo Women's Medical University, Paediatrics

VP436 Congenital muscular dystrophy with rhomboidal-rectangular crystalline inclusions

Cotta A[1], Godinho F[1], Lima M[2], Carvalho E[1], da Cunha Junior A[1], Menezes M[1], Cauhi A[1], Valicek J[1], Vargas A[1]
[1]The SARAH Network of Rehabilitation Hospitals, [2]The SARAH Network of Rehabilitation Hospitals

15:00- 15:30	**Short Oral Presentations 7** Ballroom C1 **P152-157** *Moderator: Bjarne Udd, Tampere Neuromuscular Center, Finland*	**Short Oral Presentations 8** Ballroom C2 **P158, P421-424, P251** *Moderator: Payam Mohassel, Johns Hopkins University, USA*	**Short Oral Presentations 9** Ballroom C3 **P159, P368, P383-384, P191-192** *Moderator: Carolina Tesi Rocha, Stanford University, USA*
15:30-16:30	**Poster session 4** Ballroom A-C *Refreshments served*		

P01-P05,VP06-VP07, P08-P11: SMA - therapies

P01 Risdiplam experience following onasemnogene abeparvovec in individuals with spinal muscular atrophy: a multicenter case series

Kuntz N[1], Svoboda M[2], Leon-Astudillo C[3], Byrne B[3], Krueger J[4], Kwon J[5], Sieburg C[5], Castro D[6]
[1]Division of Neurology, Department of Pediatrics, Ann and Robert H. Lurie Children's Hospital of Chicago, Northwestern University Feinberg School of Medicine, [2]Department of Pediatrics, The Children's Hospital of San Antonio/Baylor College of Medicine, [3]Department of Pediatrics, University of Florida College of Medicine, [4]Division of Pediatric Neurology, Department of Pediatrics, Helen DeVos Children's Hospital, [5]Division of Pediatric Neurology, Department of Neurology, University of Wisconsin-Madison School of Medicine and Public Health, [6]Neurology Rare Disease Center

P02 Real-world data for patients with gestational age ≤35 weeks at birth treated with onasemnogene abeparvovec: results from the RESTORE Registry

Finkel R[1], *Dabbous O*[2], *Benguerba K*[3], *Mumneh N*[2], *Raju D*[2], *Servais L*[4,5]
[1]*St. Jude Children's Research Hospital,* [2]*Novartis Gene Therapies, Inc.,* [3]*Novartis Gene Therapies Switzerland GmbH,* [4]*Department of Paediatrics, MDUK Oxford Neuromuscular Centre, University of Oxford,* [5]*Department of Paediatrics, Neuromuscular Reference Center, University and University Hospital of Liège*

P03 Administration of MF-300, an orally bioavailable small molecule inhibitor of 15-PGDH, demonstrates improved muscle force in preclinical models of neuromuscular dysfunction and disease

Webster M[1], *Vandermeulen J*[2], *Martin J*[2], *Fahr B*[1], *Grant V*[1], *Paulson S*[3], *Clark A*[4], *Khairallah R*[2]
[1]*Epirium Bio,* [2]*Myologica, LLC,* [3]*Pharmaceutical Advisors, LLC,* [4]*Pliant Therapeutics*

P04 Taldefgrobep alfa: preclinical and clinical data supporting the phase 3 RESILIENT study in Spinal Muscular Atrophy

Lair L[1], *Qureshi I*[1], *Bechtold C*[1], *Heller L*[1], *Durham S*[1], *Campbell D*[1], *Marin J*[1], *Chen K*[2], *Coric V*[1]
[1]*Biohaven Pharmaceuticals,* [2]*Spinal Muscular Atrophy Foundation*

P05 Evaluation of the neurofilament light chain as a biomarker in children with spinal muscular atrophy treated with nusinersen

Lee Y[1], *Kim S*[2], *Byun J*[3], *Lee H*[4], *Yun J*[5], *Kwon S*[1]
[1]*Department of Paediatrics, School of Medicine, Kyungpook National University, Kyungpook National University Hospital,* [2]*Department of Paediatrics, School of Medicine, Yeungnam University,* [3]*Department of Paediatrics, School of Medicine, Keimyung University,* [4]*Division of Paediatric Neurology, Department of Paediatrics, Yongin Severance Hospital, Yonsei University College of Medicine ,* [5]*Department of Genetic counselling, Graduate School of Ewha Womans University*

VP06 Real-life outcome data of paediatric patients with spinal muscular atrophy treated with nusinersen: Experience from a tertiary referral center in Turkey

Öz Yıldız S[1], *Bulut N*[2], *Alemdaroğlu İ*[2], *Debbağ S*[3], *Göçmen R*[4], *Hızarcıoğlu Gülşen H*[5], *Özçelik U*[6], *Demirkıran G*[7], *Kanbak M*[3], *Tunca Yılmaz Ö*[2], ***Haliloğlu G***[1]
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VP07 Does spinal surgery hinder intrathecal nusinersen injections in paediatric SMA patients?

Aksoy T[1], *Ramazanov R*[1], *Öz Yıldız S*[2], *Demirkıran G*[1], ***Haliloğlu G***[2], *Yazıcı M*[1]
[1]*Hacettepe University Faculty of Medicine, Department of Orthopedics and Traumatology,* [2]*Hacettepe University Faculty of Medicine, Department of Paediatrics, Division of Paediatric Neurology*

P08 Long-term comparative efficacy and safety of risdiplam versus nusinersen in children with Type 1 spinal muscular atrophy (SMA)

Sajeev G[1], *Evans R*[2], ***Hawkins N***[2,3], *Mahajan A*[4], *Scott D*[2], *Nam J*[5], *Sutherland S*[5], *Kokaliaris C*[5]
[1]*Analysis Group, Inc.,* [2]*Visible Analytics,* [3]*Institute of Health & Wellbeing, University of Glasgow,* [4]*Bridge Medical Consulting Ltd,* [5]*Global Access, F. Hoffmann-La Roche Ltd,* [6]*Roche Products Ltd*

P09 Real-world treatment with risdiplam in adults with spinal muscular atrophy (SMA): a multicenter study

Guittari C[1], *Candrilli S*[2], *Miles L*[2], *Simpson A*[3], *Shapouri S*[1]
[1]*Genentech, Inc.,* [2]*RTI Health Solutions,* [3]*Global Access, F. Hoffmann-La Roche Ltd*

P10 Zolgensma infusion and clinical progress in pharmaceutically naive SMA1 infants

Lee H[1], *Oh J*[2]
[1]*Yongin Severance Hospital,* [2]*Severance Children's Hospital*

P11 Parental experiences with newborn screening and gene replacement therapy for spinal muscular atrophy

Meyer A[1,4], *Waldrop M*[2,3,4], *Connolly A*[2,3,4], *Vannatta K*[2,6], *Hacker N*[6], *Hatfield A*[6], *Decipeda A*[6], *Parker P*[5], *Willoughby A*[5]
[1]*Division of Genetic and Genomic Medicine, Nationwide Children's Hospital,* [2]*Department of Paediatrics, The Ohio State University Wexner Medical Center,* [3]*Department of Neurology, The Ohio State University Wexner Medical Center,* [4]*Center for Gene Therapy, Abigail Wexner Research Institute, Nationwide Children's Hospital,* [5]*Division of Human Genetics, The Ohio State University,* [6]*Center for Biobehavioral Health, Nationwide Children's Hospital*

P82-P113: Outcome measures

P82 Comprehensive five-year disease progression assessment of DM1, based on the Dutch MYODRAFT registry

la Fontaine L[1], *van As D*[2], *Smulders F*[2], *Braakman H*[2], *Klinkenberg S*[1], *Bruijnes J*[1], *van Engelen B*[2], *Faber C*[1], *Merkies I*[3]
[1]*Maastricht University Medical Centre,* [2]*Radboud University Medical Centre,* [3]*Curaçao Medical Center*

P83 Myotonic Dystrophy type 1 (DM1) and physical activity (PA): an evaluation of patients in a large UK centre

Massey C[2], *Pakenham-Walsh E*[2], *Nikolenko N*[2], *Turner C*[2]
[1]*Sheffield Institute for Translational Neuroscience, University of Sheffield,* [2]*The National Hospital for Neurology and Neurosurgery, University College London Hospitals NHS Trust,* [3]*Hobbs Rehabilitation South East*

P84 Assessing infants & toddlers with neuromuscular disorders under 5 years of age using the Neuromuscular Gross Motor Outcome (GRO)

Iammarino M[1], Reash N[1], Pietruszewski L[1], Smith M[1], Lammers J[2], Waldrop M[1], Tsao C[1], Chagat S[1], Nicolau S[1], Flanigan K[1], Connolly A[1], Mendell J[1], Lowes L[1], Alfano L[1]
[1]Nationwide Children's Hospital, [2]University of Florida

P85 External responsiveness of the Duchenne video assessment, a novel fit-for-purpose remotely collected outcome measure for Duchenne muscular dystrophy

Wilson S[1], Contesse M[1], Brown C[1], Gensler G[1], Karri V[1], Hays S[1], Cornog E[1], Barnes R[1], **Sapp A**[1], Apkon S, Leffler M
[1]The Emmes Company

P86 Longitudinal multi-centric study to assess the digital outcomes issued from wearable magneto-inertial devices in ambulant DMD

Vrščaj E[1], Jilkova M[2], Aragon-Gawinska K[3], Anghelescu C[4], Axente M[5], Poleur M[6], Daron A[6], Szabo L[7], Mirea A[5], Kodsy S[8], Saleh A[8], Osredkar D[1], Haberlova J[2], Potulska-Chromik A[3], Butoianu N[4], Strijbos P[9], Eggenspieler D[10], Servais L[6,11]
[1]University Children's Hospital, Department for Paediatric Neurology, [2]Motol University Hospital, [3]Warsaw Medical University Hospital, Department of Neurology, [4]Pediatric Neurology Clinic, Clinical Hospital of Psychiatry, [5]National Clinical Hospital for Children Neurorehabilitation, [6]Centre de Référence Liégeois des Maladies Neuromusculaires, Centre Hospitalier Régional de la Citadelle, [7]Semmelweis University 2nd Department of Paediatrics, [8]Galaa Military Medical Complex, [9]F. Hoffmann-La Roche Ltd, [10]Sysnav Technologies, [11]Specialised Translational Research Oxford Neuromuscular Group, Department of Paediatrics, University of Oxford

P87 The international clinical outcome study for dysferlinopathy - ten years of natural history data

James M[1], Gordish Dressman H[2,3], Rufibach L[4], Hilsden H[1], Mayhew A[1], Straub V[1]
[1]John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, The Newcastle upon Tyne Hospitals NHS Foundation Trust and Newcastle University, [2]Center for Translational Science, Division of Biostatistics and Study Methodology, Children's National Health System, Washington, DC, USA, [3]Pediatrics, Epidemiology and Biostatistics, George Washington University, [4]Jain Foundation

P88 Intra and inter-rater reliability of the MFM32 in Myotonic Dystrophy type 1

Fossmo H[1,2,3], Robinson H[3], Ellefsen-Martinsen M[4], Frich J[3], Ørstavik K[1]
[1]Oslo University Hospital, [2]Vikersund Rehabilitation Centre, [3]University of Oslo, [4]Frambu Resource Centre for rare disorders

P89 Rehabilitation technology in assessment and treatment myotonic dystrophy type 1

Fossmo H[1,2,3], Ørstavik K[1], Gurandsrud A[2], Frich J[3], Robinson H[3]
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P90 Spinal Bulbar Muscular Atrophy (SBMA): a cross-sectional analysis of wearable sensors during the 6-minute walk (6MWT) and timed up and go (TUG) tests

Doreswamy K[1], Norato G[2], Kokkinis A[2], Joe G[1], Alqahtani A[2], Grunseich C[2], **Jain M**[1]
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P91 Importance of Nutrition in Newborns with Neuromuscular Conditions

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P92 Sleep disorders in FKRP-related limb-girdle muscular dystrophy R9

Jensen S[1,2], Abeler K[3], Friborg O[4], Rõsner A[2,5], Müller K[6], Vold M[7], Arntzen K[1,2]
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P93 Effect of intensive and individualized physiotherapy for persons with CMT- a single-subject experimental design study

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P94 Longitudinal course of long finger flexor shortening in males with Duchenne muscular dystrophy

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P95 Implementing clinical guidelines for neuromuscular disorders

Kennedy R[1,2], Carroll K[1,2], Yiu E[1,2], Donlevy G[5, 6], Bray P[5], Klaic M[4], Davidson Z[3]
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P96 Surface electromyography thresholds as a measure for performance fatigability during incremental cycling in patients with neuromuscular disorders

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P97 Exploring the repeated bout effect in neuromuscular diseases

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P98 Quality of life in adults with neuromuscular conditions and the association with diagnosis and mobility status

Wong K[1], *Michell-Sodhi J*[1], *Moat D*[1], *McCallum M*[1], *Richardson M*[1], *Harris E*[1], *Mayhew A*[1], *Guglieri M*[1], *Grover E*[1], *Díaz-Manera J*[1], *Robinson E*[1], *Elseed M*[1], *Mason J*[1], *Straub V*[1], *James M*[1], *Marini-Bettolo C*[1], *Muni-Lofra R*[1]
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P99 Barriers to outpatient physical therapy services and the role of these services in patients with neuromuscular diseases

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P100 Perceived barriers and facilitators of behavioral change towards a more active lifestyle in people living with neuromuscular disorders

Voorn E[1,2], *Oorschot S*[1,2], *Ritmeester R*[3,4], *de Morée S*[1,2,5], *Koopman F*[1,2], *van Groenestijn A*[1,2], *Jelsma J*[3,4]
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P101 JAMAR grip strength as a surrogate endpoint for global manual muscle strength in Myopathy and Motor Neuron diseases

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P102 Development of an easily applicable exercise test for exercise intensity prescription in neuromuscular diseases

Veneman T[1], *Koopman F*[1,2], *de Koning J*[3,4], *van den Aardweg J*[5], *Jørstad H*[4,6], *Nollet F*[1,2], *Voorn E*[1,2]
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P103 The validity of maximal exercise testing to assess peak oxygen consumption in people with slowly progressive neuromuscular diseases

Veneman T[1,2], *Koopman F*[1,2], *Oorschot S*[1,2], *de Koning J*[3,4], *Bongers B*[5], *Nollet F*[1,2], *Voorn E*[1,2]
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P104 Preliminary results on changes in gait dynamics measured by a Zebris® PDM platform for 13 NMD-patients after an intensive and individualized 2-week physiotherapy intervention

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P105 Implementation of standardized gait and balance analysis through use of the Zebris® PDM platform for NMD-patients included in an intensive and individualized physiotherapy intervention – description of a feasibility study

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P106 Gait analysis for support in diagnostics of neuromuscular diseases

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P107 Efficacy of aerobic exercise on aerobic capacity in slowly progressive neuromuscular diseases: a systematic review and meta-analysis

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P108 The case for inspiratory muscle training: a (true) South African story

Human A[1,2,6], *Corten L*[3], *Lozano-Ray E*[4], *M. Morrow B*[5]
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P109 The effect of a two-week intensive and individualized physiotherapy intervention for patients with rare genetic neuromuscular disorders: a quality improvement study

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P110 A multicenter retrospective study of the impact of COVID-19 on patients with muscular dystrophies

Matsumura T[1], Sato T[2], Kitao R[3], Funato M[4], Takeshima Y[5], Arahata H[6], Kobayashi M[7], Wakisaka A[8], Ogata K[9], Saito T[1], Ishigaki K[2]
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P111 Quantifying deterioration of motor abilities in ambulant boys with Duchenne muscular dystrophy as a result of COVID-19 lockdown

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P112 'Going when you have to' – a national survey on problems in urinating when away from home for women with NMD - Project HAP-PEE part 2

Werlauff U[1], **Handberg C**[1,2], Kristensen B[1], Glerup S[1,4], Vebel Pharao A[1,4], Strøm J[1,4], Thoft Jensen B[2,3]
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P113 Challenges in urinating when away from home experienced by women with neuromuscular diseases - Project HAP-PEE part 1

Kristensen B[1], **Handberg C**[1,2], Thoft Jensen B[2,3], Glerup S[1,4], Vebel Pharao A[1,4], Strom J[3,4], Werlauff U[1]
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P235-P250: DMD - clinical care and cases

P235 Best-worst scaling analysis of priorities for participation in gene therapy clinical trials for Duchenne muscular dystrophy

McNiff M[1], Heslop E[1], Denger B[3], Hill C[2], Cope H[2], Camino E[3], Johnson A[4], Fischer R[3], Peay H[2]
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P236 Rod-less dystrophin may exert a dominant negative effect by interfering with utrophin's function

Gorokhova S[1,2,3], Schessl J[4,5], Zou Y[1], Yang M[4,6], Heydemann P[7], Sufit R[8], Meilleur K[9], Donkervoort S[1], Medne L[4], Finkel R[4,10], Bönnemann C[1]
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P237 Prevalence and progression of scoliosis in non-ambulant paediatric patients with Duchenne muscular dystrophy on various glucocorticoid treatments

Sarkozy A[1,2], Burnett N[1,2], Crook V[1], Robb S[1], Main M[1], Manzur A[1], Ridout D[3], Muntoni F[1,2,4]
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P238 Predictors of requiring port-a-cath placement in boys with Duchenne muscular dystrophy on antisense oligonucleotide exon skipping therapy

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P239 The DMD Hub: a UK network enabling trials and recruitment to studies in Duchenne muscular dystrophy via the central recruitment database

Heslop E[1], Cammish P[1], Johnson A[2], Reuben E[2], Gaeta A[2], Thakrar S[2], Scotto M[3], Baranello G[3], Straub V[1], Childs A[4], Guglieri M[1]
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P240 Improving care for Duchenne muscular dystrophy: examples of the impact of collaborative working in DMD Care UK – cardiac, respiratory, psychosocial and emergency care

Turner C[1], Baronello G[2], Bourke J[1], Childs A[3], Gowda V[4], Johnson A[5], Manzur A[2], Quinlivan R[6], Rodney S[7], Sarkozy A[2], Straub V[1], Wong S[8], Guglieri M[1]
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P241 Psychosocial adjustment in adults with Duchenne and Becker muscular dystrophy: an adapted self-report questionnaire

Weerkamp P[1,2,3], Klinkenberg S[2,3], Collin P[2,4], Vermeulen J[3], Hendriksen J[2,3]
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P242 Understanding North Star Ambulatory Assessment total scores and their implications for standards of care

Stimpson G[1], James M[2], Guglieri M[2], Wolfe A[1,3], Manzur A[1,3], Baranello G[1,3], Muntoni F[1,3], Mayhew A[2], on behalf of the NorthStar Network
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P243 Functional abilities, respiratory and cardiac function in a large cohort of adults with Duchenne muscular dystrophy treated with glucocorticoids

Schiava M[1], Marini Bettolo C[1], Bourke J[1], Muni Lofra R[1], James M[1], Díaz-Manera J[1], Elseed M[1], Sodhi J[1], Moat D[1], Mccallum M[1], Mayhew A[1], Malinova M[1], Ghimenton E[1], Bolaño Diaz C[1], Wong K[1], Richardson M[1], Tasca G[1], Eglon G[1], Eagle M[2], Turner C[1], Heslop E[1], Straub V[1], Guglieri M[1]
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P244 A family with Duchenne muscular dystrophy caused by a synonymous variant, DMD c.1098 A>T that affects splicing

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P245 Early diagnosis and early corticosteroid initiation: potential benefits in Duchenne muscular dystrophy

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P246 Non-glucocorticoid related comorbidities in adults with Duchenne muscular dystrophy

Schiava M, Marini Bettolo C[1], Bourke J[1], Muni Lofra R[1], James M[1], Díaz-Manera J[1], Elseed M[1], Sodhi J[1], Moat D[1], Mccallum M[1], Mayhew A[1], Malinova M[1], Ghimenton E[1], Bolaño Diaz C[1], Wong K[1], Richardson M[1], Tasca G[1], Eglon G[1], Eagle M[2], Turner C[1], Heslop E[1], Straub V[1], Guglieri M[1]
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P247 Life-threatening bowel complications in patients with Duchenne Muscular Dystrophy: a case series

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P248 DuMAND checklist: a screening tool for behavioural difficulties in Duchenne muscular dystrophy

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P249 Early onset dilated cardiomyopathy in a 6-year-old boy with Duchenne muscular dystrophy

Zygmunt A[1,2], Villa C[1,2], Ryan T[1,2], Bhimarao Nagaraj C[1], Rybalsky I[1], Reebals L[1], Tian C[1,2]
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P250 Epilepsy in Duchenne muscular dystrophy

Armijo Gómez J[1], Liz M[2], Ortez C[1,3,4], Domínguez-Carral J[2], Exposito-Escudero J[1,3], Carrera-Garcia L[1,3], **Natera De Benito D**[1,3], Nascimento A[1,3,4]
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VP270, P271-P273, VP274, P275-P278, VP279, P280: Myasthenia gravis

VP270 Efficacy and safety of Efgartigimod in patients with generalized Myasthenia Gravis: interim results of a prospective, single-arm, observational study in China

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P271 Anti-IL-6 receptor antibody suppresses muscle atrophy in human myotube cells

Miyake S[1], Serizawa K[1], Tanaka K[1], Katsura Y[1], Noguchi-Sasaki M[1]
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P272 NMD670, a novel first-in-class muscle ClC-1 inhibitor, improves symptoms of Myasthenia Gravis: a randomized, single-dose, double-blind, placebo-controlled study

Quiroz J[1], Ruijs T[2,3], S. Grønnebæk T[1], de Cuba K[2,3], Heuberger J[2], de Kam M[2], Koopmans I[2,3], de Goede A[2], Tannemaat M[3], Vershuuren J[3], Bold J[1], Jensen K[1], Flagstad P[1], Petersen T[1], Chin E[1], Hutchison J[1], Groeneveld G[2,3], Pedersen T[1]
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P273 Long-term safety, efficacy & self-injection satisfaction with zilucoplan in myasthenia gravis: RAISE-XT interim analysis

Farmakidis C[1], Leite M[2], Bresch S[3], Freimer M[4], Genge A[5], Hewamadduma C[6], Hussain Y[7], Maniaol A[8], Mantegazza R[9], Śmiłowski M[10], Utsugisawa K[11], Vu T[12], Duda P[13], Boroojerdi B[14], Vanderkelen M[15], de la Borderie G[16], Bloemers J[16], Howard Jr J[17]
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VP274 The waning improvement rate helps predict a postoperative crisis in patients with myasthenia gravis

Ishii A[1], Nohara S[1], Tsuji H[1], Mamada N[1], Terada M[1], Nakamagoe K[1], Tomidokoro Y[1], Ishii K[1], Watanabe M[1], Tamaoka A[1]
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P275 Response to rozanolixizumab in patients with generalized myasthenia gravis (gMG) from the Phase 3 MycarinG study

Vissing J[1], Drużdż A[2], Grosskreutz J[3], Habib A[4], Mantegazza R[5], Utsugisawa K[6], Vu T[7], Grimson F[8], Beau Lejdstrom R[9], Pulido-Valdeolivas I[10], Tarancón T[10], Bril V[11]
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P276 Long-term safety, and efficacy of subcutaneous Efgartigimod PH20 in patients with Generalized Myasthenia Gravis: interim results of ADAPT-SC+

Musick K[1], Howard J[2], Li G[3], Vu T[4], Korobko D[5], Smilowski M[6], Liu L[1], Steeland S[1], Noukens J[7], Van Hoorick B[1], Podhorna J[1], Li Y[8], Utsugisawa K[9], Saccà F[10], Wiendl H[11], De Bleecker J[12], Mantegazza R[13]
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P277 Long-term safety, tolerability, and efficacy of Efgartigimod in patients with Generalized Myasthenia Gravis: concluding analyses from the ADAPT+ study

Ashcraft E[1], Bril V[2,3], Pasnoor M[4], Karam C[5], Peric S[6], De Bleecker J[7], Murai H[8], Meisel A[9], Beydoun S[10], Vu T[11], Ulrichts P[1], Van Hoorick B[1], T'joen C[1], Utsugisawa K[12], Verschuuren J[13], Mantegazza R[14], Howard J[15]
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P278 The emerging spectrum of Fetal Acetylcholine Receptor Antibody-associated Disorders (FARAD)

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VP279 Vaccination in patients with myasthenia gravis: coverage and safety

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P280 Identification of a novel RAPSN variant and electrodiagnostic confirmation of congenital myasthenic syndrome

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VP341, P342,VP343, P344-P348, VP349, P350-P356, VP357, P358-P367: Metabolic and mitochondrial myopathies

VP341 Nutritional and lipid profile status of children with Spinal muscular Atrophy in China: a retrospective case-control study

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P342 An early onset benign myopathy with glycogen storage caused by a de novo 1.3 microdeletion of chromosome 14

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VP343 Glycogen storage disease type IV without polyglucosan bodies: report of three cases and literature review

Oliwa A[1], *Langlands G[2], Sarkozy A[3], Munot P[3], Stewart W[4], Phadke R[5], Topf A[6], Straub V[6], Duncan R[7], Wigley R[8], Petty R[2], Longman C[9], Farrugia M[2]*

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P344 Immunohistochemical characterization of polyglucosan in heart and muscle in glycogenin-1 deficiency

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P345 Development of continuum of care for McArdle disease (GDSV): a practical tool for clinicians and patients

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P346 Late and very late onset of McArdles' myopathy without myoglobinuria

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P347 Toward an understanding of GSD5 (McArdle disease): How do patients learn to live with the metabolic defect in daily life

*Karazi W, Coppers J, Maas D, Cup E, Bloemen B, Voet N, Groothuis J, Pinós T, Marti R, Quinlivan R, Løkken N, Vissing J, Bhai S, Wakelin A, Reason S, **Voermans N**[1]*

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P348 Fatigue and associated factors in 172 patients with McArdle disease: an international web-based survey

Slipsager A[1], *Kahr Andersen L[1], Cornelia Voermans N[2], Lucia A[4], Karazi W[2], Santalla A[5], Vissing J[1], Løkken N[1]*

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VP349 Long-term observations of advanced Pompe disease patients treated with Enzyme replacement therapy: improvement and clinical problems

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P350 Clinical characteristics and therapeutic response of patients with adult-onset Multiple Acyl-CoA-Dehydrogenase Deficiency (MADD)

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P351 Rhabdomyolysis and muscle biopsy outcomes: a single center retrospective cohort

Ferreira W[1], *Massaro C[1], Masingue M[2], De Lonlay P[3], Laforet P[2], Behin A[2], Eymard B[2], Choumert A[4], Mafatti E[2], Stojkovic T[2], Allenbach Y[2], Bassez G[2], Evangelista T[2]*

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P352 FDG PET/CT in multiple acyl-CoA dehydrogenase deficiency late-onset: a case report

Høj A[1], Løkken N[1], D Holm-Yildiz S[1], Krag T[1], Dejanovic D[2], van Overeem Hansen T[3], Dunø M[3], Cathrine Ørngreen M[4], Vissing J[1]
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P353 ETFDH mutation causes excessive apoptosis and neurite outgrowth defect via Bcl2 pathway

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P354 Deoxynucleoside therapy for late onset Thymidine Kinase 2 Deficiency patients

Durmus Tekce H[1], Gedikbası A, Ceylaner S, Demirci H, Cakar A, Mergen S, Kıyan E, Parman Y
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P355 Mitochondrial depletion syndrome TK2 deficiency can be treated with oral deoxynucleosides

Topaloğlu H[1], Eser G[1], Yüksel B[1]
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P356 Novel TBCK variant and expanded clinical phenotype

Bhimarao Nagaraj C[1], Reebals L[1], Broomall E[1], Tian C[1]
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VP357 Riboflavin responsive glutaric aciduria type II: diagnostic pearls and challenges

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P358 Slowly progressive ophthalmoplegia as a presenting symptom of mitochondrial myopathy

Tompary A[1], Mehrabyan A[1]
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P359 12-minutes walking test in Mitochondrial myopathy: a potential screening test

Lando C[1], Løkken N[1], Khawajazada T, Storgaard J, Slipsager A, Vissing J
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P360 Primary mitochondrial myopathies diagnosed in adulthood: phenotypic spectrum and long-term outcomes

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P361 Acylcarnitine profiles in patients with mitochondrial myopathy under different physiological conditions

Joensen H[1], Løkken N[1], Khawajazada T[1], Storgaard J[1], Christensen M[2], Wibrand F[2], Vissing J[1]
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P362 Humanistic burden of neurodegenerative lysosomal disorders in the US: insights from caregivers of patients living with GM1 and GM2 gangliosidoses

Thibault N[1], Rodriguez M[1], Heuer K[2], Waggoner C[3], Jussila D[4], Perez N[5], Krupnick R[6]
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P363 The AMETHIST phase 3 trial of venglustat in patients with GM2 gangliosidoses and related diseases: baseline characteristics

Zheng R[1], Tifft C[2], Minini P[3], Batsu I[1]
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P364 Fat and glucose metabolism during exercise in patients with methylmalonic and propionic acidemia

Myrup Christensen S[1], Høj A[1], Mostue Naume M[1], Løkken N[1], Van Hall G[2], Lund A[3], Vissing J[1], Cathrine Ørngreen M[1,3]
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P365 Cycle exercise in wheelchair users with muscular dystrophy

Poulsen N[1], Pedersen J[1], Andersen R[1], Vissing J[1]
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P366 Assessment of maximal effort for weaker individuals with NMD during the assisted six-minute cycling test

Blumberg Y[1], De Monts C[1], Tang W[1], Montalvo S[2], Ataide P[1], Dunaway Young S[1], Wheeler M[2,4], Ashley E[2,4], Myers J[3], Day J[1], Christle J[2,4], **Duong T**[1]
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P367 Balancing energy expenditure and energy intake in people with neuromuscular disorders; next steps towards individualized nutritional advice

Oorschot S[1,2], Koopman F[1,2], Wierdsma N[3], van Eijnatten E[3], Brehm M[1,2], Weijs P[3,4], Soeters M[5], Voorn E[1,2]
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P437-P446, VP447, P448: Motor neuron disease and neuropathies

P437 Bright tongue sign is prior to the oral phase dysphagia of ALS patients

Kurashige T[1], Dodo Y[1], Katsumata R[1], Murao T[1], Kanaya Y[1], Sugiura T[1], Ohshita T[1]
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P438 Multimodal evaluation of the effect of salbutamol on walking capacity in ambulatory individuals with ALS: insights from the phase 2 WALKALS study

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P439 Clinical and genetic characterisation of Kennedy's disease in India

Venugopalan Thampy Yamuna V[1], Macken W[2,3], Mishra R[1], Reyaz A[1], Ahmed T[1], Dalal A[5], ICGNMD Consortium[4], Bhatia R[1], Pitceathly R[2,3], Thangaraj K[6], Reilly M[2], Srivastava P[1], Hanna M[2,3]
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P440 A new nationwide initiative to explore genetic variants in a large Turkish hereditary neuropathy cohort

G. Akinci[1], B. Ozyilmaz[2], O. Parlar[1], A. Unalp[1], I. Polat[1], F. Baydan[3], P. Karaoglu[1], P. Gazeteci[4], N Olgac Dundar[5], D. Ardicli[6], O Yayici Koken[7], A. Aksoy[8], M. Komurcu[9], D. Cavusoglu[10], U. Yis[11], **H. Topaloglu**[12]
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P441 Difference in functional status and quality of life outcomes in a cohort of siblings with Charcot-Marie-Tooth disease children – a prospective study

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P442 Charcot Marie Tooth disease type 4J and FIG4 compound heterozygous mutation

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P443 A case of Charcot-Marie-Tooth Type 4F

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P444 Ophthalmologic findings following intrathecal AAV9 mediated gene transfer for Giant Axonal Neuropathy

Bharucha-Goebel D[1,2], Saade D[1], Todd J[1,3], Huryn L[4], Norato G[3], DeLong T[1], Averion G[1], Donkervoort S[1], Foley A[1], Acquaye N[1], Mendoza C[1], Gray S[5], Zein W[4], Bonnemann C[1]
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P445 Electrophysiologic and histologic findings following intrathecal AAV9 mediated gene transfer for Giant Axonal Neuropathy

Bharucha-Goebel D[1,2], Saade D[1], Todd J[1,3], Lehky T[3], Norato G[3], Armao D[4,5], Bouldin T[5], Averion G[1], Hu Y[1], Mohassel P[1,6], Donkervoort S[1], Corse A[6], Foley A[1], DeLong T[1], Acquaye N[1], Hinkley L[1], Mendoza C[1], Hoke A[6], Gray S[7], Bonnemann C[1]
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P446 A novel Nrf2 activator with pleiotropic effects for the treatment of SBMA in a phase 1/2a study

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VP447 Clinical, electrophysiological and radiologic profile of Hirayama disease

***Gomathy S**[1], MV Srivastava P[2], Garg A[3], Agarwal A[2], Mishra R[2], Reyaz A[2], Ahmed T[2], Bhatia R[2], Priyanka Y[2], Goel V[3], L Macken W[4], D S Pitceathly R[4], Hanna M[4], Vishnu V[2]*
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P448 If you don't look, you will not find: expanding the clinical phenotype of SPG7

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LBP01-LBP21, LBVP01-LBVP03: Late Breaking

LBP01 Impaired muscle oxygen diffusive capacity in patients with Myositis

*Varone N[3], Wakeham D[2], Hinojosa J[2], Palmar D[1], Llamas C[1], Mishra P[1], Hearon Jr. C[2], **Bhai S**[1]*
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LBP02 Targeting the RANK/RANKL/OPG pathway as treatment strategy for congenital muscular dystrophy type 1A.

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LBP03 Communicative development inventory in type 1 and presymptomatic patients with Spinal Muscular Atrophy

***Buchignani B**[1,2], Capasso A[3,4], Ricci M[3,4], Cicala G[3,4], Frongia A[4], Ticci C[5], Dosi C[6], Cumbo F[7], Brolatti N[8], Coratti G[4], Pera M[4], Antonaci L[4], Masson R[6], Procopio E[5], Bruno C[8], D'Amico A[7], Pane M[3,4], Battini R[1,2], Mercuri E[3,4]*
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LBP04 An improved vectorized snRNA platform to treat DMD mutations amenable to exon skipping

***Geddes C**[1], Lardelli R[1], Nachtrab G[1], Tadokoro T[1], Knowland D[1], Sarkar A[1], Roth D[1], Carreño A[1], Reid D[1], Diago O[1], Vakharia S[1], Almaguer B[1], Nguyen L[1], Go Y[1], Torres R[1], Narayan N[1], Bradford H[1], Berlin A[1], Batra R[1], Leonard J[1]*
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LBP05 Real world experience of risdiplam in newborns with spinal muscular atrophy (SMA): A multicenter, retrospective cohort study

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LBP06 Short-term effects of neuromuscular electrical stimulation therapy in older in-patients

***Hasegawa A**[1,2,3], Yamasaka K[2], Hida M[2,4], Ichinoseki-Sekine N[1]*
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LBP07 Long-term Dystrophin recovery in humanised DMD model mice by CRISPR-Cas9 delivery using lipid nanoparticle

***Hozumi H**[1], Kenjo E[1], Inukai N[1], Hotta A[2]*
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LBP08 Galectin-3 is a biomarker for lysosomal damage in muscular dystrophy

***Isreali D**[1], Jaber A[1], Hong A[1], Bakour R[1], Richard I[1]*
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LBP09 Missense variant in TARDBP results in a novel distal myopathy

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LBP10 Impaired force generating capacity of single skeletal muscle fibers in Myositis

***Kerkhoff T**[1], Luijcx S[1], Hoomoedt D[1], Plomp L[1], Raaphorst J[2], Ottenheijm C[1]*
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LBP11 Generation of novel, orally active selective macrocyclic peptide inhibitors of myostatin for neuro-muscular diseases

Kitamura H[1], Hirata Y[1], Takuwa M[1], Koga H[1], Ohuchi M[1], Sawai N[1], Higuchi T[1], Funaki Y[1], Masuda Y[1], Kurasaki H[1], Murakami M[1], Osawa Y[2], Sunada Y[2], Masuya K[1]
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LBP12 Targeted ASO delivery to mouse lower limb by exosome carrying a muscle targeting moiety

Marban L[1], Sun M[1], Li Y[1], Caciottolo M[1], Sadri M[1], LeClaire M[1], Tran D[1], Elliott K[1]
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LBP13 Long term safety and efficacy of CAP-1002 in late-stage patients with DMD: a new treatment approach to target skeletal and cardiac muscle pathogenesis (24 month data from HOPE-2-OLE study)

McDonald C[1], Hendrix S[2], Eagle M[3], Harmelink M[4], Varadhachary A[5], Tian C[6], Apkon S[7], Villa C[6], Taylor M[8], Hor K[9], Wassom M[2], Desai U[10], Awadalla M[10], Marbán L[10]
[1]University of California Davis Health System, [2]Pentara Corporation, [3]Atom International Limited, [4]Children's Hospital, Wisconsin, [5]Washington University at St. Louis, [6]Cincinnati Children's Hospital, [7]Children's Hospital, Colorado, [8]University of Texas, Austin, [9]Nationwide Children's Hospital, [10]Capricor Inc.

LBP14 Update on long-term results of enzymatic replacement therapy with alglucosidase alfa in an Italian cohort of late-onset Pompe disease (LOPD)

Mongini T[1], Musumeci O[2], Ravaglia S[3], Ricci G[4], Siciliano G[4], Maggi L[5], Filosto M[6], D'Angelo G[7], Comi G[8], Tonin P[9], Fiumara A[10], Barone R[10], Ruggiero L[11], Verriello L[12], Barp A[13], Pegoraro E[14], Servidei S[15], Toscano A[2], on behalf of the Italian Myology Association Study Group for Pompe Disease
[1]Neuromuscular Unit, Department of Neuroscience RLM, University of Torinos, [2]University of Messina, [3]Istituto Neurologico Mondino, [4]University of Pisa, [5]Istituto Besta, [6]University of Brescia, [7]Istituto la Nostra Famiglia, [8]University of Milano, [9]University of Verona, [10]University of Catania, [11]Federico II University, [12]University of Udine, [13]Centro Clinico Nemo, [14]University of Padova, [15]Catholic University

LBP15 Wild type hBAG3 expression improves survival and function in the SOD1.G93A mouse model for ALS

Ozes B[1], Tong L[1], Myers M[1], Moss K[1], Attia Z[1], Sahenk Z[1,2,3]
[1]Center for Gene Therapy, The Abigail Wexner Research Institute, Nationwide Children's Hospital, [2]Department of Pediatrics and Neurology, Nationwide Children's Hospital and The Ohio State University, [3]Department of Pathology and Laboratory Medicine, Nationwide Children's Hospital

LBP16 A splice-altering homozygous variant in COX18 causes severe sensory-motor neuropathy with oculofacial apraxia

Mavillard F[2], Guerra-Castellano A[3], Rivas E[1], Cantero G[2], Servián-Morilla E[2], Folland C[4], Ravenscroft G[4], Diaz-Moreno I[3], Miranda A[2], Cabrera-Serrano M[1,2], **Paradas C**[1,2]
[1]Hospital Virgen del Rocío, [2]Biomedicine Institute of Sevilla, [3]Investigaciones Químicas/ Universidad de Sevilla, [4]Harry Perkins Institute of Medical Research, Centre for Medical Research, University of Western Australia

LBP17 Long-term survival and cardiac efficacy of delandistrogene moxeparvovec gene therapy in the Duchenne muscular dystrophy rat model

Baine S[1], Wier C[1], Lemmerman L[1], Cooper-Olson G[1], Kempton A[1], Haile A[1], Endres J[1], Fedoce A[1], Nesbit E[1], Rodino-Klapac L[1], **Potter R**[1]
[1]Sarepta Therapeutics Inc

LBP18 Functional and splicing changes of ambulatory spinal muscular atrophy type 3 patients by 20 weeks of risdiplam treatment

Shin J[1], Kim H[1], Lee S[2], Kim S[2], Park H[2]
[1]Pusan National University Yangsan Hospital, Pusan National University College of Medicine, [2]Gangnam Severance Hospital, Yonsei University College of Medicine

LBP19 RGX-202, an Investigational gene therapy for the treatment of Duchenne muscular dystrophy: interim clinical data

Veerapandiyan A[1], Dastgir J[2], Falabella P[2], Pakola S[2], Rastogi S[2], Phillips D[2], Wilson C[2], Boulos N[2], Hall J[2], Jimenez V[2], Gilmor M[2], Yang L[2], Fiscella M[2], Danos O[2]
[1]Arkansas Children's Hospital, [2]RegenxBio

LBP20 Ataluren slows the decline of muscle function in patients with nmDMD: a meta-analysis of three randomized, double-blind, placebo-controlled trials

Jong Y[1], Karachunski P[2], Statland J[3], Lorentzos M[4], Cairns A[5], Takeshima Y[6], Haginoya K[7], Penematsa V[8], Chou C[8], Gordon G[8], Williams P[8], **Werner C**[9]
1Graduate Institute of Clinical Medicine, College of Medicine, Kaohsiung Medical University, and Departments of Pediatrics and Laboratory Medicine, and Translational Research Center of Neuromuscular Diseases, Kaohsiung Medical University Hospital, Kaohsiung Medical University, [2]University of Minnesota, [3]University of Kansas Medical Center, [4]The Children's Hospital at Westmead, [5]Neurosciences Department, Queensland Children's Hospital, [6]Department of Paediatrics, Hyogo Medical University, [7]Department of Pediatric Neurology, Miyagi Children's Hospital, [8]PTC Therapeutics Inc., [9]PTC Therapeutics Germany GmbH

LBP21 Bone marrow fat fraction is elevated in corticosteroid-treated boys with Duchenne muscular dystrophy

Kunnath Ravindrunanni R[1], Walter G[1], Bernier A[1], Tuna I[1], Lopez C[1], Vandenborne K[1], Rajapakse C[2], **Willcocks R**[1]
[1]University of Florida, [2]University of Pennsylvania

LBVP01 ALY688, a novel adiponectin receptor agonist, improves muscle function and reduces inflammation and fibrosis in mdx mice

Pignalosa A[1], *Hsu H*[1], *Crawford K*[1], *Abou-Samra M*[2], *Dubuisson N*[2], *Versele R*[2], *Davis-López de Carrizosa M*[2,3], *Brichard S*[2], *Selvais C*[2], *Noel L*[2], *Van den Bergh P*[4]

[1]*Allysta Pharmaceuticals Inc,* [2]*Institute of Experimental and Clinical Research (IREC) Université Catholique de Louvain,* [3]*Departamento de Fisiología,* [4]*Neuromuscular Reference Center Cliniques Universitaires Saint-Luc*

LBVP02 Amelioration of myocardial fibrosis in mdx mice model of Duchenne muscular dystrophy (DMD) on oral consumption of Aureobasidium Pullulans produced Neu REFIX Beta glucans

Abraham S[1,2,3,4,5], *Levy G*[6], *Yamamoto N*[7], *Cherian K*[8], *Premsekar R*[9], *Senthilkumar R*[2,10], *Preethy S*[10]

[1]*University Of Yamanashi,* [2]*Antony- Xavier Interdisciplinary Scholastics (AXIS), GN Corporation Co. Ltd.,* [3]*Mary-Yoshio Translational Hexagon (MYTH), Nichi-In Centre for Regenerative Medicine (NCRM),* [4]*R & D, Sophy Inc.,* [5]*Levy-Jurgen Transdisciplinary Exploratory (LJTE), Global Niche Corp,* [6]*Emeritus professor, Medicine and Immunology, University of Toronto,* [7]*National Centre for Global health and Medicine (NCGM),* [8]*Frontier Lifeline Hospitals, R-30-C Ambattur Industrial Estate Road, Mogappair,* [9]*Dr. Kamakshi Memorial Hospital,* [10]*Fujio-Eiji Academic Terrain (FEAT), Nichi-In Centre for Regenerative Medicine (NCRM)*

LBVP03 Efficacy and safety of Efgartigimod in patients with generalised Myasthenia gravis: final results of a prospective, single-arm, observational study in China

Liang H[1], *Wang P*[1], *Zhang B*[1], *Zhao C*[2], *Huang S*[1]

[1]*Department of Neurology, Hainan General Hospital, Haikou, China,* [2]*Huashan Rare disease centre, Department of Neurology, Huashan Hospital Fudan University*

16:30-16:45	**Short Oral Presentations 10** Ballroom C1 **P82-P87** *Moderator: Tina Duong, Stanford University, USA*	**Short Oral Presentations 11** Ballroom C2 **P235, P01-P02** *Moderator: Jorge Alfredo Bevilacqua, Universidad de Chile & Clínica Dávila, Chile*
18:00-18:30	**Transport to Networking Pre-Dinner Drinks** (separate registration required)	
18:30-19:30	**Pre-Networking Dinner Drinks** Sharehouse, Downtown Charleston (separate registration required)	
19:30-23:00	**Networking Dinner** The Bus Shed, Downtown Charleston (separate registration required)	

Saturday 7th October 2023

07:30-15:00	**Registration desk open**
07:30-09:00	**Arrival refreshments** PAC Foyer
07:45-08:45	**Clinical Trial Updates** PAC *Moderators: Kristl Claeys, Universitaire Ziekenhuizen Leuven, Belgium & Ulrike Schara Schmidt, University of Essen, Germany*

O18: Topline Safety and Efficacy Data Analysis of Phase 1/2 Clinical Trial Evaluating AOC 1001 in Adults with Myotonic Dystrophy Type 1: MARINA™

Nicholas Johnson[1], *John Day*[2], *Johanna Hamel*[3], *Charles Thornton*[3], *S.H. Subramony*[4], *Payam Soltanzadeh*[5], *Jeffrey Statland*[6], *Matthew Wicklund*[7], *W. David Arnold*[8], *Miriam Freimer*[8], *Kelly DiTrapani*[9], *Carrie Heusner*[9], *Chao-Yin Chen*[9], *Brad McEvoy*[9], *Yiming Zhu*[9], *Li-Jung Tai*[9], *Elizabeth Ackermann*[9]

[1]*Virginia Commonwealth University,* [2]*Stanford University Medical Center,* [3]*University of Rochester,* [4]*University of Florida,* [5]*University of California, Los Angeles,* [6]*University of Kansas Medical Center,* [7]*University of Colorado, Denver,* [8]*The Ohio State University,* [9]*Avidity Biosciences*

O19: Preliminary Results from MLB-01-003: An Open Label Phase 2 Study of BBP-418 in Patients with Limb-girdle Muscular Dystrophy Type 2I/R9

Harper A[1], *Langeslay R*[1], *Rajasingham T*[2,3], *Rodriguez H*[2,3], *Blankenbiller T*[2,3], *Hutchaleelaha A*[2,3], *Sproule D*[2,3]

[1]*Virginia Commonwealth University,* [2]*BridgeBio Pharma,* [3]*ML Bio Solutions*

O20: Safety and efficacy of intravenous onasemnogene abeparvovec in patients with spinal muscular atrophy: interim findings from the phase 3 SMART study

McMillan H[1], *Baranello G*[2,3], *Farrar M*[4,5], *Zaidman C*[6], *Seibert J*[7], *Bernardo R*[8], *Alecu I*[7], *Freischläger F*[9], *Muntoni F*[2,3]

[1]*Children's Hospital of Eastern Ontario,* [2]*The Dubowitz Neuromuscular Centre, Developmental Neuroscience Research and Teaching Department, University College London Great Ormond Street Institute of Child Health,* [3]*NIHR Great Ormond Street Hospital Biomedical Research Centre & Great Ormond Street Hospital NHS Foundation Trust,* [4]*School of Clinical Medicine, UNSW Medicine and Health, UNSW Sydney,* [5]*Department of Neurology, Sydney Children's Hospital Network,* [6]*Department of Neurology, Division of Pediatric Neurology, Washington University School of Medicine,* [7]*Novartis Pharmaceuticals,* [8]*Novartis Global Drug Development – Neuroscience,* [9]*Freischläger Consulting*

07:45-08:45

O21: 104-week efficacy and safety of cipaglucosidase alfa+miglustat in patients with late-onset Pompe disease previously treated with alglucosidase alfa

Mozaffar T[1], *Bratkovic D*[2], *Byrne B*[3], *Claeys K*[4], *Díaz-Manera J*[5], *Kishnani P*[6], *Laforêt P*[7], *Roberts M*[8], *Toscano A*[9], *Castelli J*[10], *Goldman M*[10], *Jiang H*[10], *Sitaraman Das S*[10], *Wasfi Y*[10], *Schoser B*[11]
[1]*Department of Neurology, University of California,* [2]*PARC Research Clinic, Royal Adelaide Hospital,* [3]*University of Florida,* [4]*Department of Neurology, University Hospitals Leuven, and Laboratory for Muscle Diseases and Neuropathies, Department of Neurosciences, KU Leuven,* [5]*John Walton Muscular Dystrophy Research Centre, Newcastle University,* [6]*Duke University Medical Center,* [7]*Nord-Est/Ile-de-France Neuromuscular Reference Center, Neurology Department, Raymond-Poincaré Hospital,* [8]*Salford Royal NHS Foundation Trust,* [9]*Neurology and Neuromuscular Disorders Unit, Department of Clinical and Experimental Medicine, Università di Messina,* [10]*Amicus Therapeutics, Inc.,* [11]*Friedrich-Baur-Institut, Neurologische Klinik, Ludwig-Maximilians-Universität München*

09:00-11:00

The Victor Dubowitz Lecture PAC

Moderators: Volker Straub, Newcastle University, UK & Chris Weihl, Washington University in St. Louis, USA

09:00-09:30 **INV15 RNA-targeted therapy for ALS**

Miller T[1]
[1]*Washington University*

Poster Highlights PAC

Moderators: Alan Beggs, Boston Childrens Hospital / Harvard Medical School, USA & Svetlana Gorokhova, National Institute of Health, USA

O22: P81 Gastrointestinal assessment in Spinal Muscular Atrophy (SMA): the experience of SMA healthcare professionals in France

Marta Gomez Garcia, APHP Raymond Poincare University Hospital, Child Neurology and Paediatric ICU Department Pediatrique, France

O23: P161 Natural history of distal and myofibrillar myopathies assessed by clinical and technological outcome measures (Dista-Myo): baseline results

Giorgio Tasca, Newcastle University, UK

O24: P266 Gene expression profiles and spatial localisation of dystrophin isoforms in developing and adult human brain

Francesco Catapano, University College London, UK

O25: P325 A comparative single nuclei transcriptomics approach to evaluating the terminally differentiated lymphocytes in autoimmune Myositis

Francia Victoria De Los Reyes, National Center of Neurology and Psychiatry (NCNP), Japan

O26: P350 Clinical characteristics and therapeutic response of patients with adult-onset Multiple Acyl-CoA-Dehydrogenase Deficiency (MADD)

Sofie Sunebo, Linköping University Hospital. Sweden

O27: P425 Inhibition of TGFβ signaling pathway as a therapeutic approach in collagen VI-related muscular dystrophy

Hailey Hearn, Johns Hopkins University, USA

11:00-11:30

Morning refreshments PAC Foyer **and posters** Ballroom

11:30-13:00

Late Breaking News PAC *Moderator: Lindsay Wallace, Nationwide Children's Hospital, USA and Michele Yang, Children's Hospital Colorado, USA*

LBO01: Impaired iron-sulfur cluster assembly due to biallelic variants in CIAO1 leads to a novel muscle disease

Or Bach R[1], *Maio N*[2], *Zaharieva I*[3], *Töpf A*[4], *Donkervoort S*[1], *Foley A*[1], *Munot P*[3], *Silverstein S*[1], *Mueller J*[3], *Verma S*[5], *Douglas G*[6], *Peric S*[7], *Grunseich C*[8], *Hu Y*[1], *Sewry C*[3], *Sarkozy A*[3], *Straub V*[4], *Muntoni F*[3], *Rouault T*[2], *Bönnemann C*[1]
[1]*Neuromuscular and Neurogenetic Disorders of Childhood Section/NINDS/NIH,* [2]*Eunice Kennedy Shriver National Institute of Child Health and Human Development, NIH,* [3]*Dubowitz Neuromuscular Centre, UCL Institute of Child Health,* [4]*John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University and Newcastle Hospitals NHS Foundation Trust,* [5]*Department of Neurology, Emory University School of Medicine,* [6]*GeneDx,* [7]*Department for Neuromuscular Disorders, Neurology Clinic, University Clinical Centre of Serbia, Faculty of Medicine, University of Belgrade, 8National Institute of Neurological Disorders and Stroke/NIH*

LBO02: Ablation of the Carboxiterminal end of MAMDC2 causes a distinct muscular dystrophy

Paradas C[1,2], *Mavillard F*[1], *Servián-Morilla E*[1], *Dofash L*[3], *Rojas-Marcos I*[2], *Folland C*[3], *Monahan G*[3], *Gutierrez-Gutierrez G*[4], *Rivas E*[2], *Laín A*[5], *Valladares A*[1], *Cantero G*[1], *Morales J*[2], *Laing N*[3], *Ravenscroft G*[3], *Cabrera-Serrano M*[1,2]
[1]*Instituto de Biomedicina de Sevilla, Hospital Virgen del Rocio.,* [2]*Hospital Virgen del Rocio,* [3]*Harry Perkins Institute of Medical Research,* [4]*Hospital Infanta Sofia,* [5]*Hospital 12 de Octubre*

LBO03: A novel class of Tubulinopathies - Mutations in TUBA4A cause primary skeletal muscle disorders

Johari M[1], *Folland C*[1], *Saito Y*[2], *Oud M*[3], *Töpf A*[4], *Kurbatov S*[5], *StudyGroup T, Pais L*[6], *Cairns A*[7], *Kang P*[8], *Straub V*[4], *Beggs A*[9], *Fahey M*[10], *Cossée M*[11], *Voermans N*[12], *Udd B*[13], *Laing N*[1], *Nishino I*[2], *Tartaglia M*[14], *Ravenscroft G*[1]

[1]*Harry Perkins Institute of Medical Research, Centre for Medical Research, University of Western Australia, Nedlands WA,* [2]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo,* [3]*Department of Human Genetics, Donders Institute for Brain, Cognition and Behaviour, Radboud University Medical Center, Nijmegen,* [4]*John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne,* [5]*Research Institute of Experimental Biology and Medicine, Voronezh N. N. Burdenko State Medical University, Voronezh,* [6]*Division of Genetics and Genomics, Boston Children's Hospital and Harvard Medical School, Boston, MA,* [7]*Neurosciences Department, Queensland Children's Hospital, Brisbane (Meanjin) Queensland ,* [8]*Paul and Sheila Wellstone Muscular Dystrophy Center and Department of Neurology, University of Minnesota Medical School, Minneapolis, Minnesota,* [9]*The Manton Center for Orphan Disease Research, Division of Genetics and Genomics, Boston Children's Hospital, Harvard Medical School, Boston, MA,* [10]*Department of Paediatrics Monash Children's Hospital, Victoria,* [11]*PhyMedExp, Université de Montpellier, INSERM, CNRS, 34093 Montpellier,* [12]*Department of Neurology, Donders Institute for Brain, Cognition and Behaviour, Radboud University Medical Center, Nijmegen,* [13]*Folkhälsan Research Center, Department of Medical and Clinical Genetics, Medicum, University of Helsinki,* [14]*Molecular Genetics and Functional Genomics, Ospedale Pediatrico Bambino Gesù, IRCCS, Rome*

LBO04: CGG repeat expansion in LRP12 causes both amyotrophic lateral sclerosis and oculopharyngodistal myopathy type 1

Kume K[2], ***Kurashige T***[1], *Muguruma K*[3], *Morino H*[4], *Tada Y*[2], *Kikumoto M*[5], *Miyamoto T*[6], *Akutsu S*[7], *Matsuda Y*[2], *Matsuura S*[5], *Nakamori M*[5], *Nishiyama A*[8], *Izumi R*[8], *Niihori T*[9], *Ogasawara M*[10], *Eura N*[10], *Kato T*[11], *Yokomura M*[11], *Nakayama Y*[12], *Ito H*[12], *Nakamura M*[13], *Saito K*[11], *Riku Y*[14], *Iwasaki Y*[14], *Maruyama H*[5], *Aoki Y*[9], *Nishino I*[10], *Izumi Y*[15], *Aoki M*[8], *Kawakami H*[2]

[1]*Department of Neurology, Nho Kure Medical Center And Chugoku Cancer Center,* [2]*Department of Molecular Epidemiology, RIRBM, Hiroshima University,* [3]*Department of iPS Cell Applied Medicine, Graduate School of Medicine, Kansai Medical University,* [4]*Department of Medical Genetics, Tokushima University Graduate School of Biomedical Sciences,* [5]*Department of Clinical Neuroscience and Therapeutics, Hiroshima University Graduate School of Biomedical and Health Sciences,* [6]*Department of Molecular and Cellular Physiology, Graduate School of Medicine, Yamaguchi University,* [7]*Department of Genetics and Cell Biology, RIRBM, Hiroshima University,* [8]*Department of Neurology, Tohoku University Graduate School of Medicine,* [9]*Department of Medical Genetics, Tohoku University Graduate School of Medicine,* [10]*Department of Neuromuscular Research, National Institute of Neuroscience, National Centre of Neurology and Psychiatry,* [11]*Institute of Medical Genetics, Tokyo Women's Medical University,* [12]*Department of Neurology, Wakayama Medical University,* [13]*Department of Neurology, Kansai Medical University,* [14]*Department of Neuropathology, Institute for Medical Science of Aging, Aichi Medical University,* [15]*Department of Neurology, Tokushima University Graduate School of Biomedical Sciences*

LBO05: Proteomic serum profiling identifies ITIH3 as a new biomarker for Myasthenia gravis disease activity

Schroeter C[1], *Nelke C*[1], *Stascheit F*[2], *Stenzel W*[3], *Roos A*[4], *Meisel A*[2], *Meuth S*[1], ***Ruck T***[1]

[1]*Heinrich-Heine-University Düsseldorf, Department of Neurology,* [2]*Department of Neurology and Experimental Neurology, Charité - Universitätsmedizin Berlin,* [3]*Department of Neuropathology, Charité - Universitätsmedizin Berlin,* [4]*Pediatric Neurology, University Children's Hospital, Faculty of Medicine, University of Duisburg-Essen*

LBO06: Functional improvements and decreased aggregate burden in TgT57I Mice following AAVrh74.tMCK.hBAG3 gene therapy

Ozes B[1], *Tong L*[1], *Moss K*[1], *Myers M*[1], *Attia Z*[1], *Vetter T*[1], *Sahenk Z*[1,2,3]

[1]*Center for Gene Therapy, The Abigail Wexner Research Institute, Nationwide Children's Hospital,* [2]*Department of Pediatrics and Neurology, Nationwide Children's Hospital and The Ohio State University, Columbus,* [3]*Department of Pathology and Laboratory Medicine, Nationwide Children's Hospital*

LBO07: RNA-based CRISPRoff silencing to target DUX4 in Facioscapulohumeral muscular dystrophy

He J[1,2], *Sasaki-Honda M*[1], *Tanaka H*[3], *Akita H*[3,4], *Sakurai H*[1]

[1]*Center For iPS Cell Research And Application (CiRA), Kyoto University,* [2]*Graduate School of Medicine, Kyoto University,* [3]*Laboratory of DDS Design and Drug Disposition, Graduate School of Pharmaceutical Sciences, Chiba University,* [4]*Laboratory of Drug Design and Drug Disposition, Graduate School of Pharmaceutical Sciences, Tohoku University, Sendai*

LBO08: Identification of AAV variants with enhanced skeletal muscle and muscle stem cell transduction

Chen H[1], *Emami M*[1], *Young C*[2], *Zhang X*[1], *Del Vecchio G*[1], *Rando T*[1], *Jimenez R*[1], *Frietas B*[1], *Pyle A*[1], *Ikotun O*[1], ***Spencer M***[1]

[1]*UCLA,* [2]*Myogene Bio*

LBO09: scAAV9.U7-ACCA treatment of DMD exon 2 duplication leads to significant dystrophin expression and evidence of clinical benefit, particularly following treatment as an infant

Waldrop M[1,2,3], *Lawlor M*[4], *Vetter T*[1,2], *Frair E*[1], *Beatka M*[4], *Meng H*[3], *Iammarino M*[1], *Sabo B*[4], *Subramanian S*[1], *Kaler M*[1], *Simmons T*[1], *Wein N*[1,2], ***Flanigan K***[1,2,3]

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	Prize Giving Ceremony PAC *Moderator: Johann Böhm* **Introduction to the WMS 2024 Congress, Prague, Czech Republic** *Jana Haberlová* **Handover of the WMS flag and close of congress** *Moderator: Volker Straub*
13:30-14:30	Homeward lunch PAC Foyer
13:30-15:00	**NMD Board Meeting** Meeting room 6 & 7 (separate registration required)