

WMS 2025 Full Programme

Version: 06/10/25

WMS 2025 Poster Sessions, Short Oral Presentations, Clinical Trial Updates and Industry Symposia are not EACCME accredited. Details of these sessions are in the full Congress programme appendix.

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)
VSP	Virtual Environmental Sustainability Poster Presentation

Please note, all times stated in the programme are in local Vienna, Austria time (CET).

Monday 6th October 2025

08:30-19:00	Pre-Congress Teaching Course (separate registration required) 📍 -2.41/-2.42
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Tuesday 7th October 2025 [Click here for details of Industry Symposia 1 & 2](#)

08:30-11:30	Pre-Congress Teaching Course (separate registration required) 📍 - 2.33 and - 2.32	
10:00-15:00	WMS Executive Board Meeting (by invitation only) 📍 -2.41/-2.42	
13:30-18:00	Registration, welcome refreshments and poster set up 📍 Foyer D, and Hall X1	
15:30 -16:30	Regional Networking Reception 📍 Hall G	
16:30-17:30	Industry Symposium 1 (withdrawn) 📍 Hall G	Industry Symposium 2 📍 Hall K
18:00-18:45	Opening Ceremony 📍 Hall D	
18:45-21:00	Networking Reception 📍 Hall X1	

Wednesday 8th October 2025

[click here for full details of Industry Symposia 3-6, Poster sessions 1 & 2, and Short Oral presentations 1-6](#)

07:15-19:30	Congress desk open 📍 Foyer D	
07:45-08:45	Industry Symposium 3 📍 Hall G	Industry Symposium 4 📍 Hall K
08:50-09:30	Congress Welcome - Message from the President 📍 Hall D	
09:30-11:00	Topic 1: Neuromuscular diseases as multisystemic disorders 📍 Hall D <i>Supported by the EAN</i> <i>Moderators: Günther Bernert, Klinik Favoriten, Austria & Corinne Horlings, Medical University Innsbruck, Austria</i> 09:30-10:00 01INV How can proteomics help to elucidate the pathophysiology crosstalk in muscular dystrophy and associated multi-system dysfunction? Ohlendieck K¹ ¹ National University of Ireland	

	10:00-10:30 02INV Beyond muscular dystrophies: roles of MD-related proteins in different organ systems Winter L¹ ¹Medical University of Vienna, Austria 10:30-10:45 01O Interferon-γ causes myogenic cell dysfunction and senescence in immune myopathies Authier F¹, Periou B, Gervais M, Martin L, Berthier J, Baba Amer Y, Souvannanorath S, Lechapt-Zalcman E, Malfatti E, Gherardi R, Relaix F, Bencze M, Hou C ¹Paris Est Creteil University, France 10:45-11:00 02O Brachio-cervical inflammatory myopathy: Evidence for a distinct form of inflammatory muscle disease Kleefeld F^{1,2}, Gallardo Vigo E, Teran Gamboa J, Funke A, Roos A, Mensch A, Gehrig J, Ruck T, Mossakowski A, Llansó L, Bortolani S, Torchia E, Casal-Dominguez M, Pinal-Fernandez I, Mammen A, Tasca G, Preuße C, Stenzel W ¹Ruhr University Bochum, BG University Hospital Bergmannsheil, Germany, ²Charité - Universitätsmedizin Berlin, Germany
11:00-11:30	Morning refreshments, exhibition and poster viewing 📍 Hall X1
11:00-11:30	Myology Café Myology Developments Across the World and Guidelines Committees 📍 Hall X1
11:30-13:30	Topic 1: Neuromuscular diseases as multisystemic disorders (continued) 📍 Hall D Moderators: John Vissing, University of Copenhagen, Denmark & Jana Haberlová, University Hospital Motol, Czechia 11:30-12:00 03INV Myotonic Dystrophy Type 1: A multisystemic disorder with remarkable clinical variability and burden Braakman H¹ ¹Amalia Children's Hospital, Radboud University Medical Center, the Netherlands 12:00-12:30 04INV Spinal Muscular Atrophy: Systemic disease, focal treatment – or vice versa? Kirschner J¹ ¹University of Freiburg, Germany 12:30-12:45 03O Cognitive development in children with 5q-SMA identified by neonatal screening – four years follow-up Kölbel H¹, Kopka M, Pum S, Alina S, Blaschek A, Schara-Schmidt U, Müller-Felber W, Schwartz O, Vill K ¹University Hospital Essen, Essen, Germany 12:45-13:00 04O Denervated human muscle fibers promote reinnervation via neurotrophic factor release in SMA and ALS Goku-Nath R, Collins C, Valverde L, Lleixa C, Verdú-Díaz J, Di Lorenzo A, Llarch P, Querol L, Marini-Bettolo C, Rojas-García R, Díaz-Manera J¹ ¹Newcastle University, United Kingdom 13:00-13:15 05O Redefining triple A syndrome: A multinational study of the neuromuscular phenotype in the largest genetically confirmed cohort to date Oeztuerk M¹, Leonard-Louis S, Natera de Benito D, Nascimento A, Orteç C, Virić V, Stevic Z, Nelke C, Schaezner A, Previtali S, Cehic M, Dumic K, Peric S, Roos A, Ruck T ¹BG University Hospital Bergmannsheil, Germany 13:15-13:30 06O In-depth examination of motor endplate pathology in AChR-Ab-positive myasthenia gravis Preusse C¹, Meisel A, Brokamp K, Roos A, Doksani P, Hentschel A, Schuelke M, Rückert J, Pumberger M, Schömig F, Stenzel W, Hoffmann S ¹Charité – Universitätsmedizin Berlin, Germany
13:30-14:30	Lunch, exhibition and poster viewing 📍 Hall X1
13:45-14:30	NMD Editorial Board Meeting (by invitation only) 📍 -2.41/-2.42

14:30-15:30	Poster session 1 ♡ Hall X1 01P-20P, 22P-50P, 51VP, 52P, 53VP-55VP: Acquired, inflammatory, myositis 166P-192P: DMD – imaging and outcome measures 298P-313P: Muscle MRI & new imaging techniques 399P-426P, 427VP-430VP: Clinical trials, access to health care and outcome measures 505P-528P, 529VP, 530P-536P, 537VP: SMA clinical 651P-677P, 678VP-688VP: DMD - treatments		
15:30-16:00	Short Oral Presentations 1 ♡ K1 Updates on SMA and DMD trials 426P, 423P, 422P, 425P, 424P <i>Moderator: Ulrike Schara-Schmidt, University Duisburg-Essen, Germany</i>	Short Oral Presentations 2 ♡ K2 Inflammatory myopathies 48P, 46P, 49P, 24P, 50P <i>Moderator: Georgio Tasca, Newcastle University, United Kingdom</i>	Short Oral Presentations 3 ♡ -2.93/-2.94 Novel discoveries in SMA, DMD and ADSS1 myopathy 534P, 536P, 535P, 192P, 191P, 313P <i>Moderator: Simone Mahal, Favoriten Hospital, Austria</i>
16:15-17:00	Congress Debate ♡ Hall D Should families be offered n=1 treatments? <i>Moderators: Michelle Lorentzos, The Sydney Children's Hospitals Network, Australia & Jorge A Bevilacqua, Universidad de Chile, Chile</i> 13INV Annemieke Aartsma-Rus, Leiden University Medical Center, the Netherlands 14INV Kevin Flanigan, Nationwide Children's Hospital, United States of America The WMS Congress Debate has quickly become one of the most popular elements of the scientific programme. This year the debate will ask the timely question, should we be providing hyper-individualised therapies/treatments, namely n=1 or n=few, to patients with neuromuscular disease? Many genetic neuromuscular patients live with progressive or even fatal conditions that lack effective treatment options. In an age that enables the design of a single, tailor-made antisense oligonucleotide (ASO) or gene therapy for a single patient, or a very small group of patients, where should we draw the line? When families request a bespoke therapy for their loved one, should we allow unproven, individualised treatments outside traditional clinical trials? Is this compassion... or chaos in the making? The debate team includes experts from across the globe, including opponents Kevin Flanagan (USA) who will argue in favour of providing hyper-individualised therapies and Annemieke Aartsma-Rus (The Netherlands), who will be arguing against, with expert moderation by Michelle Lorentzos (Australia) and Jorge Bevilacqua (Chile). Please note that in an effort to demonstrate differing viewpoints, as well as educate and entertain, Kevin and Annemieke will intentionally present extreme positions that may not convey their actual and nuanced perspectives on this important issue. To round off this interactive discussion, the debaters will offer their genuine reflections on the future of individualised treatments, and present their insights regarding the real-world complexities. We encourage members of the audience to participate. Please join us for what we are sure will be another Congress highlight!		
17:15-18:15	Poster session 2 ♡ Hall X1 118P-165P: CM – CMD 193P-217P: FSHD 314P-329P: Multidisciplinary management of neuromuscular diseases 431P-456P, 457VP-458VP: Genetics of NMD (new genes and NGS, diagnostic etc.) 486P-503P, 504VP: LGMD 551P-566P: EDMD, OPDM, autophagic, extramuscular		
18:15-18:45	Short Oral Presentations 4 ♡ K1 Genetic neuromuscular disorders 1 451P, 453P, 454P, 128P, 452P, 161P <i>Moderator: Wolfgang Löscher, Medical University Innsbruck, Austria</i>	Short Oral Presentations 5 ♡ K2 Genetic neuromuscular disorders 2 163P, 165P, 164P, 456P, 455P, 502P <i>Moderator: Hacer Durmus, Istanbul Faculty of Medicine, Turkey</i>	Short Oral Presentations 6 ♡ -2.93/-2.94 FSHD 216P, 214P, 217P, 215P <i>Moderator: Carmen Paradas, Hospital Universitario Virgen del Rocío/IBiS, Spain</i>
19:00-20:00	Industry Symposium 5 ♡ Hall G		Industry Symposium 6 ♡ Hall D

07:00-15:00	Congress desk open 📍 <i>Foyer D</i>
07:45-09:15	Interesting Case Discussions 📍 <i>Hall D</i> <i>Moderators: Teerin Liewluck, Mayo Clinic Rochester, United States of America & Ana Ferreiro, Institut De Myologie, France</i> Case 1: Atypical nemaline myopathy <i>Laforet P, Hopital Raymond-Poincaré APHP, France</i> Case 2: A diagnostic odyssey solved by discovering a novel Alu insertion in the TTN gene <i>Murtazina A, Research Centre for Medical Genetics, Russia</i> Case 3: Recessive variants in DNAJB4 cause a distinct protein aggregate myopathy with early respiratory involvement <i>DeWinter J, University Hospital Antwerp, Belgium</i> Case 4: The overlapping landscape of neuromuscular disorders in the era of NGS: lessons learned from a family with autosomal dominant scapuloperoneal motor weakness <i>Fernandez Garcia de Eulate G, Association Institut De Myologie, France</i> Case 5: A novel mutation of HSPB8 causing atypical pediatric-onset axial and limb-girdle involvement <i>Lyu X, Qilu Hospital of Shandong University, China</i> Case 6: Anti-neurofascin-155 nodopathy in a 12-year-old girl <i>Klouwer F, Leiden University Medical Center, The Netherlands</i> Case 7: Severe osteochondral complications in NTRK1-related CIPA: A case report and mechanistic insights <i>Chan S, The University of Hong Kong, Hong Kong</i> Case 8: Myofibrillar Myopathy cases with equinovarus deformity and early contractures <i>Durmus Tekce H, İstanbul Üniversitesi Tıp Fakültesi Hastanesi, Turkey</i>
09:30-11:00	Topic 2: Multidisciplinary management of neuromuscular diseases 📍 <i>Hall D</i> <i>Moderators: Michela Guglieri, Newcastle University, United Kingdom & Saskia Houwen, Raboud University Medical Centre, The Netherlands</i> 09:30-10:00 05INV Psychological care in neuromuscular diseases: supporting patients with rare, progressive, and genetic diseases in the era of gene therapy <u>Sayah S</u>¹ ¹<i>Pitié-Salpêtrière Hospital, France</i> 10:00-10:30 06INV The expanding role of rehabilitation in neuromuscular diseases: from care to translational research <u>Gagnon C</u>¹ ¹<i>Université de Sherbrooke, Canada</i> 10:30-10:45 07O Neuropsychiatric burden of Dystrophinopathies: A multi-centre European study <u>Kolesnik A</u>¹, Geagan C, Miranda R, Weerkamp P, Chieffo D, Slipsager A, Suárez Bagnasco M, Wroom E, Wroom E, Vissing J, Niks E, Desguerre I, Straub V, Mercuri E, Hendriksen J, Skuse D, Muntoni F ¹<i>Dubowitz Neuromuscular Centre, UCL Great Ormond Street, Institute of Child Health, United Kingdom</i> 10:45-11:00 08O Insights into the use of the capability approach in multidisciplinary rehabilitation for persons with neuromuscular diseases <u>Pijpers E</u>¹, Bloemen B, Oortwijn W, Van Engelen B, Van der Wilt G, Groothuis J, Cup E ¹<i>Radboudumc, Netherlands</i>
11:00-11:30	Morning refreshments, exhibition and poster viewing 📍 <i>Hall X1</i>
11:00-11:30	Myology Café Environmental Sustainability Committee 📍 <i>Hall X1</i>

11:30-13:30	Topic 2: Multidisciplinary management of neuromuscular diseases (continued) 📍 <i>Hall D</i> Moderators: Nicol Voermans, Radboud University Medical Centre, the Netherlands & Payam Mohassel, Johns Hopkins University, United States of America 11:30-12:00 07INV Coordination of care for people with neuromuscular diseases: the sum is greater than the individual parts <u>Quinlivan R</u>¹, Desikan M, Kaur S, Astin R, Savvatis K, Emmanuel A, Bouquillon L, Sarcozy A, Hanna M ¹University College London, United Kingdom 12:00-12:30 08INV Palliative care for adults with neuromuscular diseases- what do we know and where next? <u>Willis D</u>¹ ¹The Severn Hospice, United Kingdom 12:30-12:45 09O Optimising transition and advanced care planning for young people with neuromuscular disorders <u>Irving K</u>¹, Howard M, Culnane E ¹Royal Children's Hospital Melbourne, Australia 12:45-13:00 10O Sexual and pelvic floor function in women with Muscular Dystrophy <u>Hamel J</u>¹, Dekdebrun J, Lopa S, Thornburg L, Anto-Ocrah M ¹University of Rochester, United States of America 13:00-13:15 11O Heart transplantation in males with dystrophinopathy: An advanced cardiac therapies improving outcomes network (ACTION) Dystrophinopathy registry analysis <u>Hayes E</u>¹, Villa C, Nandi D, Cripe L, Wittlieb-Weber C, Hsu D, Auerbach S, Martinez H, Shugh S, Asante-Korang A, Miller E, Absi M, Kaufman B, Gambetta K, Harris R, Lal A, Mokshagundam D, Shih R, Radcliff L, Cunningham T ¹Nationwide Children's Hospital, United States of America 13:15-13:30 12O Computational models for new patient stratification strategies of neuromuscular disorders (CoMPaSS-NMD): a new strategy to tackle hereditary neuromuscular diseases <u>Nuredini A</u>¹ (u/b), Socha M, Pioreski L, Markiel M, Corrias R, Topf A, Verdú Díaz J, Garzo Manzanares A, Albano N, Mendelsohn D, Laporte J, Diaz Manera J, Obach M, Scipioni M, Savarese M, Santorelli F, Straub V, Schoer B, Polańska J, Tupler R ¹University of Modena and Reggio Emilia, Italy
13:30-14:45	Lunch, exhibition and poster viewing 📍 <i>Hall X1</i>
13:45-14:45	WMS 2025 Early to Mid-Career Peer Networking Session (separate registration required) 📍 <i>Hall G</i> Moderators: Megan Iammarino, Nationwide Children's Hospital, United States of America & Jordi Diaz-Manera, Newcastle University, United Kingdom For the first time in its history, in October 2025 the WMS membership will elect a dedicated early to mid-career member representative to the Executive Board. This session is a unique opportunity for early to mid-career delegates to meet the newly-elected WMS Board representative and to collaborate with fellow early-mid career attendees to shape future activities within the WMS and the WMS Congress. This is a unique opportunity for participants to have their say in early to mid-career focussed content at future WMS Congresses, journal content, and WMS activities. Session attendees will also gain access to a valuable international professional peer network, providing opportunities for future collaborations, sharing expertise, and career development.
15:00-17:30	Poster viewing/group activity (separate registration required)
17:30-20:00	Group Activity Networking Reception (separate registration required)

08:00-18:00	Congress desk open 📍 <i>Foyer D</i>
08:30-10:15	Topic 3: Advances in therapies and drug development 📍 <i>Hall D</i> <i>Supported by the ICNMD</i> <i>Moderators: Elie Naddaf, the Mayo Clinic, United States of America & Nalini Atchayaram, National Institute of Mental Health and Neuro Sciences, India</i> 08:30-09:00 09INV Advances in therapies and drug development: improving AAV Vectors for Neuromuscular Disease indications Spencer M¹ ¹<i>Center for Duchenne Muscular Dystrophy at UCLA, United States of America</i> 09:00-09:30 10INV AAV transduction optimization for NMD Adjali O¹ ¹<i>Université de Nantes, France</i> 09:30-09:45 13O Identifying naturally occurring adeno-associated virus serotypes that transduce mouse skeletal muscles fibro-adipogenic progenitors in vivo Guirguis F^{1,2}, Bolduc V, Brull A, Liu H, Chiorini J, Zabaleta N, Vandenberghe L, Zhou H, Muntoni F, Bönnemann C¹ ¹<i>National Institutes of Health, United States of America</i> ²<i>The Dubowitz Neuromuscular Centre, University College London, United Kingdom</i> 09:45-10:00 14O Regulatory T cells suppress dystrophin immunity Farahat P¹, Villalta S ¹<i>University Of California Irvine, United States of America</i> 10:00-10:15 15O Towards a human multi-organoid platform to develop and assess specificity, toxicity and efficacy of neuromuscular gene therapies Dastidar S, Torun T, Raghu Chakravarthy P, Zouhair M, Duan Z, Chen P, Ferrari T, Ferreira M, Lionello V, Ralu M, Sarcar S, Amendola M, Tedesco F^{1,2,3} ¹<i>University College London, United Kingdom</i>, ²<i>The Francis Crick Institute, United Kingdom</i>, ³<i>Dubowitz Neuromuscular Centre, Great Ormond Street Hospital for Children & UCL Great Ormond Street Institute of Child Health, United Kingdom</i>
10:15-10:45	Morning refreshments, exhibition and poster viewing 📍 <i>Hall X1</i>
10:15-10:45	Myology Café Featured Cases and EDI Committees 📍 <i>Hall X1</i>
10:45-12:30	Topic 3: Advances in therapies and drug development (continued) 📍 <i>Hall D</i> <i>Moderators: Ana Tesi Rocha, Stanford University, United States of America & Vishnu Venugopalan Yamuna, All India Institute of Medical Sciences, India</i> 10:45-11:15 11INV Leveraging the muscle fusogens for gene delivery Millay D¹ ¹<i>Cincinnati Children's Hospital, United States of America</i> 11:15-11:45 12INV Innovation in nanoparticles as drug delivery vehicles for the treatment of muscle disorders Lau H¹ ¹<i>Yale School of Medicine, United States of America</i> 11:45-12:00 16O Calcitriol ameliorates myotonia in 3D DM1 skeletal muscle models via a MBNL1-independent mechanism Fernández Costa J¹, Fernández-Garibay X, Maria Sabater-Arcís M, Tejadera-Villafranca A, Núñez-Manchón J, Artero R, Suelves M, Nogales-Gadea G, Ramón-Azcón J ¹<i>Institute for Bioengineering of Catalonia (IBEC), Barcelona Institute of Science and Technology (BIST), Spain</i> 12:00-12:15 17O Collagen type VI regulates TGFβ bioavailability in skeletal muscle in mice Mohassel P^{1,2}, Hearn H, Rooney J, Zou Y, Johnson K, Norato G, Nalls M, Yun P, Ogata T, Silverstein S, Sleboda D, Roberts T, Rifkin D, Bönnemann C ¹<i>Johns Hopkins University School of Medicine, United States of America</i>, ²<i>National Institutes of Health, United States of America</i>

	12:15-12:30 180 Translation of GGC repeat expansions into a toxic polyG protein in oculopharyngodistal myopathy 2 Jiao K ^{1,2} , Chen X, Cao M, Zhang J, Xia X, Yue D, Gao M, Yu W, Pan W, Zhao C, Wang T, Zhu W, Xi J ¹ Huashan Hospital, Fudan University, China, ² National Center for Neurological Disorders (NCND), China		
13:00-14:00	WMS General Assembly/poster viewing for non-members 📍 Hall D		
12:30-14:00	Lunch, exhibition and poster viewing 📍 Hall X1		
13:45-14:15	Sponsor meeting (by invitation only) 📍 -2.41/-2.42		
14:15-15:15	Poster Session 3 📍 Hall X1 56P-89P: Advances in therapies and drug development 104P-116P, 117VP: Cell insights, muscle homeostasis 251P-296P, 297VP: Dystrophinopathies (animals models, biomarkers, brain, genetics) 366P-398P: Myotonic dystrophy 538P-550P: Registries, networks and care of NMD 589P-609P: SMA outcome measures and registries 689P, 690VP, 691P-702P: Distal myopathies, MFM		
15:15-15:45	Short Oral Presentations 7 📍 K1 Myotonic dystrophy and distal myopathies 689P, 395P -398P Moderator: Benedikt Schoser, Friedrich-Baur-Institut LMU München, Germany	Short Oral Presentations 8 📍 K2 Advances in therapies and SMA 609P, 87P, 88P, 116P, 86P Moderator: Wenhua Zhu, Huashan Hospital, Fudan University, China	Short Oral Presentations 9 📍 -2.93/-2.94 Insights into muscular dystrophy 288P, 89P, 270P, 295P Moderator: Sophelia Chan, The University of Hong Kong, China
15:45-16:45	Poster Session 4 📍 Hall X1 90P-103P: ALS/neuropathy 218P-250P: Neuromuscular diseases as multisystemic disorders 330P-365P: Metabolic and mitochondrial myopathies 459P-484P, 485VP: Myasthenia Gravis, NMJ1-2, periodic paralysis 567P-569P, 571P-574P: Pompe disease 575P-588P: SMA therapies 610P-620P, 622P-648P, 649VP-650VP: DMD - clinical care and cases reports, BMD 704LBP-712LBP, 714LBP-719LBP, 721LBP-728LBP, 730LBP-734LBP: Late breaking 735VSP-743VSP: Environmental sustainability		
16:45-17:15	Short Oral Presentations 10 📍 K1 Multisystemic disorders and disease mechanisms 647P, 648P, 246P -248P Moderator: Mridul Johari, The Harry Perkins Institute of Medical Research, Australia	Short Oral Presentations 11 📍 K2 Metabolic and mitochondrial disorders 361P - 365P Moderator: Suur Biliciler, Ut Health Science Center at Houston, McGovern Medical School, United States of America	Short Oral Presentations 12 📍 -2.93/-2.94 Congenital muscle diseases 471P, 249P, 484P, 250P Moderator: Hernán Gonorazky, The Hospital for Sick Children, Canada
19:15-01:00	Networking Dinner (separate registration required)		

07:15	Congress desk open ♥ Foyer D
07:30-09:00	Clinical Trial Updates ♥ Hall D <i>Moderators: Ann Agnes Mathew, Bangalore Baptist Hospital, India & Tobias Ruck, BG University Hospital Bochum, Germany</i> 07:30-07:45 190 07:45-08:00 200 08:00-08:15 210 08:15-08:30 220 08:30-08:45 230 08:45-09:00 240
09:00-09:15	Comfort break
09:15-09:45	Victor Dubowitz Lecture ♥ Hall D Myology in Asia <i>Ichizo Nishino, National Institute of Neuroscience, NCNP, Japan</i> <i>Moderators: Volker Straub, Newcastle University, United Kingdom & Gina Ravenscroft</i> <i>The Harry Perkins Institute of Medical Research, Australia</i>
09:45-11:15	Poster Highlights <i>Moderators: Matthias Baumann, Medical University Innsbruck, Austria & Rotem Orbach, National Institutes of Health, United States of America</i> 09:45-10:00 300 Expert consensus on gene therapy for spinal muscular atrophy with onasemnogene abeparvovec: treatment decision, administration and patient follow-up <u>Davion J¹</u> , Desguerre I, Audic F, Cancès C, Barnerias C, Espil-Taris C, Fauroux B, de Feraudy Y, Guichard M, Isapof A, Quijano Roy S, Vuillerot C, Bodiguel E, Project group Expert consensus on gene therapy for Spinal muscular atrophy with onasemnogene abeparvovec P ¹ Department of Pediatric Neurology, French Reference Center for Neuromuscular Diseases, Lille University Hospital Center, Lille, France 10:00-10:15 260 C2C12-CTE – a versatile myotube model for C-terminal titin studies <u>Sarparanta J¹</u> , Jonson P, Vainio A, Luque H, Hackman P, Udd B ¹ Folkhälsan Research Center, Helsinki, Finland 10:15-10:30 270 Heart rate and ventilation during submaximal cycling exercise as physiological outcome measures in Duchenne muscular dystrophy <u>Taivassalo T¹</u> , Paul M, Duong T, Lott D, Forbes S, Shih R, Leon Astudillo C, Sullivan R, Sweeney L ¹ University Of Florida, Gainesville, United States 10:30-10:45 280 Evaluating oxygen saturation and recovery dynamics in skeletal muscle during exercise in adults with spinal muscular atrophy <u>Duong T¹</u> , Khonde S, de Monts C, Tang W, Salvatore S, Dunaway Young S, Vogt-Domke S, Yoseph Hailu R, Yatsu L, Taivassalo T, Rogers M, W Day J, Hageman N ¹ Stanford University, Palo Alto, United States 10:45-11:00 290 A closer look at the contribution of fibro-adipogenic precursors in the pathomechanism of autoimmune myositis: correlating transcriptomics with muscle pathology <u>De Los Reyes F¹</u> , Hayashi S, Noguchi S, Nishino I ¹ Department of Neuromuscular Research, National Institute of Neuroscience, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Kodaira, Tokyo, Japan 11:00-11:15 250 ASCC3-related congenital myopathy is linked to a defect in ribosome-associated quality control <u>Onnée M¹</u> , Johari M, Roos A, Nicolau S, Ravenscroft G, Malfatti E, ASCC3 Myopathy Working Group ¹ Institut Mondor de Recherche Biomédicale, Université Paris Est Créteil, INSERM U955, Créteil, France
11:15-11:45	Morning refreshments, exhibition and poster viewing ♥ Hall X1

11:45-13:15	Late Breaking News 📍 Hall D Moderators: Hanns Lochmuller, Children's Hospital of Eastern Ontario Institute, Canada & Sandra Donkervoort, National Institutes of Health, United States of America 11:45-11:57 01LBO A novel 5' UTR CCG expansion in TBC1D7 causes Oculopharyngodistal myopathy Van De Vondel L^{1,2}, Curro R, Facchini S, Xu I, De Winter J, Quartesan I, Monticelli A, Alonso-Jimenez A, De Ridder W, Roberts M, Brady S, Merve A, Neveling K, Hoischen A, Houlden H, Hanna M, Bugiardini E, Zuchner S, Baets J, Cortese A ¹Dr. John T. Macdonald Foundation Department of Human Genetics and John P. Hussman Institute for Human Genomics, University of Miami Miller School of Medicine, Miami, United States, ²Translational Neurosciences, University of Antwerp, Antwerp, Belgium 11:57-12:09 02LBO DUX4-induced toxicity in Facioscapulohumeral muscular dystrophy requires mitochondrial respiratory chain function Zammit P¹, Pfeiffer-Vogl J, Ganassi M, McGuire L, Engquist E, Tyszkiewicz M, Pruller J, Grillari J, Banerji C, Heher P ¹King's College London, London, United Kingdom 12:09-12:21 03LBO Opposite modulation of dynamin and amphiphysin provides therapeutic proof-of-concepts for a peripheral neuropathy and two congenital myopathies Laporte J¹, Goret M, Reiss D, Edelweiss E, Gomez-Oca R, Massana-Munoz X ¹GBMC, Illkirch, France 12:21-12:33 04LBO In utero risdiplam for severe SMA: a case series Finkel R¹, SMA prenatal therapy study group ¹St. Jude Children's Research Hospital, Memphis, United States 12:33-12:45 05LBO Delpacibart zotadirsén (del-zota) showed trends toward improvement in functional and patient-reported outcomes in individuals with DMD amenable to exon 44 skipping Flanigan K¹, Veerapandiyar A, Lavery C, McDonald C, Smith E, Tesa Rocha C, Wong B, Tami Y, Carmack T, Kovach P, Herzog J, Hughes S, Zhu Y, Ackermann E ¹Nationwide Children's Research Institute, Columbus, United States 12:45-12:57 06LBO Acute liver injury mitigation and management in patients with Duchenne muscular dystrophy following administration of delandistrogene moxeparvovec: expert considerations Shieh P¹, Proud C, Diamond T, Chapin C, Ahmad J, Salama A, Bönnemann C, Soslow J, Byrne B, Veerapandiyar A, Brandsema J, Matesanz S, Bharucha-Goebel D, Samelson-Jones B, Wilkins B, Gerber M, Mason S, Asher D, McDonald C ¹University of California Los Angeles, Los Angeles, United States 12:57-13:09 07LBO Safety and preliminary efficacy is demonstrated in a phase 1/2a clinical trial of AJ201 in spinal and bulbar muscular atrophy Grunseich C¹, Day J, Oskarsson B, Pestronk A, Sorenson E, Chang J, Chan Y, Chen C, Mozaffar T ¹National Institutes of Health, Bethesda, United States
13:15-13:45	Prize Giving Ceremony 📍 Hall D Moderator: Marco Savarese, University of Helsinki, Finland Introduction to the WMS 2026 Congress, Hiroshima, Japan Handover of the WMS flag and close of the Congress
13:45-14:30	Homeward lunch

WMS 2025 Congress programme appendix

Version: 06/10/25

Please note that WMS 2025 Poster Sessions, Short Oral Presentations, Clinical Trial Updates and Industry Symposia are **not** EACCME accredited.

Affiliations are shown only for the presenting author. The full list of affiliations for authors can be found in the Congress abstract book.

To leave the appendix and return to the main programme, please click the [link](#) in the bottom corner of each page.

Tuesday 7th October 2025

To leave the appendix and return to the main programme, please click the [link](#) in the bottom corner of each page.

16:30-17:30 **Industry Symposium 1** 📍 Hall G

Withdrawn

16:30-17:30 **Industry Symposium 2** 📍 Hall K

Advancing the treatment landscape in Duchenne muscular dystrophy

Chair: Laurent Servais, University of Oxford, United Kingdom
Speakers: Ulrike Schara-Schmidt, University Duisburg-Essen,
Germany & Erika Finanger, Oregon Health & Science University,
United States of America

Wednesday 8th October 2025

To leave the appendix and return to the main programme, please click the [link](#) in the bottom corner of each page.

07:45-08:45 **Industry Symposium 3** 📍 Hall G

Cardiomyopathy and advances in the treatment paradigm

Kan Hor, Nationwide Children's Hospital,
United States of America

07:45-08:45 **Industry Symposium 4** 📍 Hall k

Achieving functional improvement in myotonic dystrophy type 1 (DM1): defining goals of treatment and a roadmap to multidisciplinary care

Lori Planco, Monica Visone, Nicholas Johnson,
Valeria Sansone

Poster Session 1 – 01P-20P, 22P-50P, 51VP, 52P, 53VP-55VP: Acquired, inflammatory, myositis

01P Knowledge of Indonesian neurologists in the diagnosis and management of patients with idiopathic inflammatory myopathy

Abdan Syakuron¹, Hakim M, Budikayanti A, Wiratman W, Yanuar Safri A, Fadli N, Ridski Harsono A, Octaviana F, Ari Indrawati L
¹Department of Neurology, Faculty of Medicine, Universitas Indonesia, Jakarta, Indonesia

02P Immunophenotyping of patients with inclusion body myositis: INSPIRE-IBM study

Farahat P¹, Phillips C, Hernandez I, Wencel M, Mozaffar T, Villalta A
¹University Of California, Irvine, Irvine, United States

03P A study of muscle biopsy cases positive for anti-cytosolic 5'-nucleotidase 1A antibodies

Ishida M^{1,2}, Kurashige T, Murao T, Maruyama H, Ohshita T
¹Department of Clinical Neuroscience and Therapeutics, Graduate School of Biomedical and Health Science, Hiroshima University, Hiroshima, Japan, ²Department of Neurology, NHO Kure Medical Center and Chugoku Cancer Center, Kure, Japan

04P Novel mouse model for inclusion body myositis: transgenic upregulation of lymphotoxin together with impaired autophagy induces inflammation and protein accumulation in skeletal muscle

Bremer J, Nagel J, Zschüntzsch J, Zajt K, Palaz T, Blank T, Ikis A, Fischer L, Einer C, Eck A, Kana V, Aguzzi A, Prinz M, Liebetanz D, Odoardi F, Kuo C, Weis J, Kraft F, **Schmidt J**¹, Heikenwälder M

¹Institute of Neuropathology, University Hospital RWTH Aachen, Aachen, Germany

05P Consistent clinical features of anti-Mi-2 myopathy: a Korean retrospective cohort study

Kim S¹, Choi Y, Choi Y, Kim W, Park H

¹Department of Neurology, Gangnam Severance Hospital, Yonsei University College of Medicine, Seoul, South Korea

06P Persistent exanthema mainly on the trunk with pathologically dermal interstitial mucin as anti-HMGCR myopathy-associated skin rash: case series

Zhu W^{1,2}, Cheng N, Liu Z, Zheng S, Chen L

¹Huashan Hospital, Fudan University, Shanghai, China, ²National Center for Neurological Disorders, Shanghai, China

07P A multicenter retrospective cross-sectional study of juvenile-onset anti-HMGCR myopathy

Zhu W^{1,2}, Cheng N, Diao M, Sun L, Huang B, Hao S, Zeng L

¹Huashan Hospital, Fudan University, Shanghai, China, ²National Center for Neurological Disorders, Shanghai, China

08P Comparative evaluation of clinical outcome measures in Inclusion body myositis based on NT5c1A antibody serology status

Wencel M¹, Goyal N, Carbutar O, Freimer M, Dimachkie M, Quinn C, Lloyd T, Mohassel P, Wehl C, Shaibani A, Wang L, Chahin N, Amato A, Wicklund M, Dixon S, Shieh P, Herbelin L, Barohn R, Tawil R, Ciafaloni E

¹University Of California, Irvine, Orange, United States

09P Case series of immune checkpoint inhibitor-induced overlap syndrome: myasthenia, myositis, and myocarditis in a tertiary center

Gomez Fernandez F¹, Cabrera Serrano M, Rojas-Marcos I, Lopera Rodriguez I, Rivas Infante E, Montes Latorre E, Paradas López C

¹Hospital Universitario Virgen Del Rocio: Serv. Neurologia, Sevilla, Spain

10P Anti-synthetase syndrome—finding a pearl in an oyster: a series of 12 cases

Bhagat S¹, Jassal B, Vishnu V, Sharma M, Bhadu D

¹All India Institute of Medical Sciences, New Delhi, Delhi, India

11P An advanced clinico-sero-histological model to improve differentiation among idiopathic inflammatory myopathy subgroups

Jassal B¹, Dhall A, Vishnu V, Bhadu D, Suri V, Sharma M

¹Neuropathology Lab, All India Institute of Medical Sciences, New Delhi, India

12P Bioengineered muscle-on-a-chip for the study of inclusion body myositis

Andújar-Sánchez F^{1,2}, Fernández-Garibay X, Valls-Roca L, Vilaseca-Capel A, Deng H, Tobias E, Guitart-Mampel M, Tejadera-Vilafranca A, Farré-Tarrats L, Tort F, Selva-O'Callaghan A, Moreno-Lozano P, Matas A, Fernández-Costa J, Ramón-Azcón J, Milisenda J, Garrabou G

¹IDIBAPS - HCB - University of Barcelona, Barcelona, Spain, ²CIBERER, Madrid, Spain

13P Uliprubarb pharmacokinetics, pharmacodynamics (PK/PD), and safety: phase 1 study results in patients with inclusion body myositis (IBM)

Needham M, Henderson R, Liang C, Soler-Ferran D, **Wilkins J**¹, Greenberg S

¹Perron Institute, QEII Medical Centre, Nedlands, Western Australia, Australia

14P Clinicopathological characteristics of chronic cases of immune-mediated necrotizing myopathy

Komaki S¹, Kubota A, Maeda M, Shimizu J, Yamanaka A, Nishino I, Toda T

¹Department of Neurology, Graduate School of Medicine, The University of Tokyo, Tokyo, Japan

15P Primary skeletal muscle peripheral T-cell lymphoma associated with HIV presenting with myopathic symptoms

Ji G^{1,2}, Sun F, Song X

¹Department of Neurology, the Second Hospital of Hebei Medical University, Shijiazhuang, China, ²Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan

16P Comparative evaluation of respiratory assessments in inclusion body myositis from INSPIRE-IBM study

Hernandez I¹, Wencel M, Goyal N, Carbutar O, Freimer M, Dimachkie M, Quinn C, Lloyd T, Mohassel P, Wehl C, Shaibani A, Wang L, Chahin N, Amato A, Wicklund M, Dixon S, Shieh P, Herbelin L, Barohn R, Mozaffar T

¹University Of California, Irvine, Orange, United States

17P INSPIRE-IBM: an NIH-funded, two-year, multicenter, observational study in inclusion body myositis- an update and AE/SAE reporting

Hernandez I¹, Wencel M, Goyal N, Carbunar O, Freimer M, Dimachkie M, Quinn C, Lloyd T, Mohassel P, Wehl C, Shaibani A, Wang L, Chahin N, Amato A, Wicklund M, Dixon S, Shieh P, Herbelin L, Barohn R, Mozaffar T

¹University Of California, Irvine, Orange, United States

18P Guillain-Barré syndrome in children: insights from a 20-year retrospective cohort study at a tertiary referral center

Chávez Gloria H^{1,2,3}, Escher A, Carrera - García L, Exposito - Escudero J, Nascimento A, Natera - De Benito D, Ortez Gonzalez C

¹Neuromuscular Unit, Barcelona, Spain, ²Department of Neurology, Barcelona, Spain, ³Hospital Sant Joan de Déu, Barcelona, Spain

19P Clinico-patho-serological correlation for classification of idiopathic inflammatory myopathies and therapeutic outcomes - a real-world retrospective cohort

Sivaraman Nair S¹, George J, Poyuran R, Narasimhaiah D

¹Sree Chitra Tirunal Institute for Medical Sciences and Technology, Trivandrum, Trivandrum, India

20P Rituximab in treatment-refractory childhood-onset immune-mediated necrotizing myopathy: a case series

Dang H¹, Le S

¹University Medical Center Ho Chi Minh City, Ho Chi Minh City, Vietnam

22P Proteomic profiling in juvenile dermatomyositis toward the elucidation of protein signatures correlating with severity of myopathology

Roos A^{1,2,3,4}, Pauper M, Holla E, Hentschel A, Nelke C, Beltran S, Kölbel H, Tucht C, Ruck T, Schara-Schmidt U, Della-Marina A

¹Department of Pediatric Neurology, University Duisburg-Essen, Essen, Germany ²Department of Neurology, University Hospital Düsseldorf, Düsseldorf, Germany, ³Heimer Institute for Muscle Research, Bochum, Germany, ⁴Brain and Mind Research Institute, Ottawa, Canada

23P Myopathology and immune profile of granulomatous myositis in sarcoid myopathy

Holzer M¹, Ruffer N, Pinal-Fernandez I, Kleefeld F, Goebel H, Schänzer A, Casal-Dominguez M, Kötter I, Görl N, Alten R, Braasch E, Lempert T, Krause A, Huber T, Liewluck T, Mammen A, Stenzel W, Preuß C, Schneider U, Krusche M

¹University Medical Center Hamburg-Eppendorf, Division of Rheumatology and Systemic Inflammatory Diseases, III. Department of Medicine, Hamburg, Germany

24P Immune-checkpoint inhibitor-induced myositis – myopathology revealing novel phenotypes

Holzer M¹, Dittmayer C, Goebel H, Dressel A, Stenzel W

¹University Medical Center Hamburg-Eppendorf, Division of Rheumatology and Systemic Inflammatory Diseases, III. Department of Medicine, Hamburg, Germany

25P STAT1 and STAT3 in muscular inflammation and dystrophy

Winkler M¹, Kornblum C, Reimann J

¹Department of Neuromuscular Diseases, Center for Neurology, University Hospital of Bonn, Bonn, Germany

26P Therapeutic outcomes and prognostic determinants in juvenile myasthenia gravis

Puppala S¹, Tandon V, Sundaram S, Nair S

¹Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, India

27P Non-caseating granulomatous myopathy associated with thymoma and atypical presentation of myasthenia gravis

Meller A¹, Shankar N, Traub R, Mehrabyan A

¹University of North Carolina Hospitals Dept of Neurology, Chapel Hill, United States

28P Amyloid myopathy: hypertrophic subtype correlates with higher muscular amyloid deposition

Authier F^{1,2}, Periou B, Araujo M, Hankiewicz K, Heyraud-Blanchet C, Plante-Bordeneuve V, Lefeuvre C, Zaroui A, Damy T, Severa G, Malfatti E, Souvannanorath S

¹Paris Est Creteil University, Créteil, France, ²Henri Mondor University Hospital, Créteil, France

29P Myological evaluation of patients with post-acute COVID-19 syndrome

Authier F^{1,2}, Aoun Sebaiti M, Souvannanorath S, Malfatti E, Itti E, Severa G

¹Paris Est Creteil University, Créteil, France, ²Henri Mondor University Hospital, Créteil, France

30P Malignancy characteristics among idiopathic inflammatory myopathies: a retrospective study at Dr Cipto Mangunkusumo hospital

Indrawati L, **Putri M¹**, Isaac W, Shafitha N, Simatupang S, Safri A, Fadli N, Harsono A, Wiratman W, Budikayanti A, Octaviana F, Hakim M

¹Department of Neurology, Dr. Cipto Mangunkusumo Hospital, Jakarta, Indonesia

31P Clinico-sero-pathological and transcriptomic features of inclusion body myositis with T-cell large granular lymphocytic leukemia

Soontrapa P^{1,2}, Pinal-Fernandez I, Paul P, Skolka M, Milone M, Shi M, Shah M, Mammen A, Liewluck T

¹Department of Neurology, Mayo Clinic, Rochester, Minnesota, United States, ²Division of Neurology, Department of Medicine, Siriraj Hospital, Mahidol University, Bangkok, Thailand

32P Perimysial inflammation with abundant macrophages

Puusepp S¹, Kiseleva N, Kannel K, Stenzel W, Goebel H

¹Pathology Department, Tartu University Hospital, Tartu, Estonia

33P New insights into the mouse model for Jo-1, PL-7, and PL-12 associated anti-synthetase syndrome

Bachir D¹, Preusse C, Lichtenberg S, Eigenfeldt L, Koch-Hölsken K, Umthum V, Herrmann A, Vinnenberg L, Schänzer A, Pinal-Fernandez I, Mammen A, Fassbender B, Seifert M, Meuth S, Stenzel W, Ruck T

¹Department of Neurology, Medical Faculty, Heinrich Heine University Düsseldorf, Düsseldorf, Germany

34P Characterization of inflammatory infiltrate in inclusion body myositis and it's comparison with other IIMs

Rajput N¹

¹Aiims Delhi, Delhi, India

35P The impact of idiopathic inflammatory myopathy on quality of life: data from a tertiary healthcare in Indonesia

Indrawati L^{1,2}, Kautharifa F, Ramli Y, Ariane A, Octaviana F, Hakim M

¹Department of Neurology, Dr Cipto Mangunkusumo Hospital, Jakarta, Indonesia, ²Faculty of Medicine, Universitas Indonesia, Jakarta, Indonesia

36P Understanding muscle biopsy pain: what to expect during and after

Labella B^{1,2}, Brochier G, Beuvin M, Chanut A, Lacene E, Labasse C, Madelaine A, Levy-Borsato F, Leonard-Louis S, Bassez G, Seilhean D, Evangelista T

¹Neuromuscular Morphology Unit, Institut De Myologie, Paris, France, ²Neuropathology Unit, Department of Neuropathology, Groupe Hospitalier Universitaire La Pitié-Salpêtrière, Paris, France

37P Molecular differences between regenerating and inflamed myofibers in Inclusion body myositis

Nijssen T¹, Davis S, Raaphorst J, Bos E, O`Shaughnessy R, Aronica E, Kessler B, Fischer R, Raz V

¹Leiden University Medical Center, Leiden, Netherlands

38P Clinical course and treatment of anti-mitochondrial M2 antibody-positive myositis: a single-center retrospective case series

Nishimori Y¹, Eura N, Sugie K

¹Department of Neurology, Nara Medical University, Kashihara, Japan

39P The effects of myositis-specific autoantibodies in immune-mediated necrotizing myopathy on muscle fiber contractility

Kerkhoff T^{1,2,3}, Luijckx S, Ardic B, Stachurska P, van der Kooi A, Aronica E, Raaphorst J, Ottenheijm C

¹Amsterdam UMC - Location Vrije Universiteit, Physiology, Amsterdam, Netherlands, ²Amsterdam Cardiovascular Sciences, Amsterdam, Netherlands, ³Amsterdam Movement Sciences, Amsterdam, Netherlands

40P Clinical features of cardiac complications and restrictive ventilatory impairment in AMA-positive myositis: findings from a nationwide cross-sectional study

Maeda M¹, Kawahara T, Kubota A¹, Shimizu J, Toda T

¹The University of Tokyo, Tokyo, Japan

41P Treatment outcomes in idiopathic inflammatory myopathies based on pathology and autoantibody profiles: a single-center study of 127 cases

Eura N¹, Nishimori Y, Yamanaka A, Shiota T, Tanaka H, Ohashi T, Shimizu H, Yamaoka M, Yamada N, Iguchi N, Tanaka A, Sugata M, Nanaura H, Kiriya T, Sugie K

¹Department of Neurology, Nara Medical University, Kashihara, Japan

42P Rare case of Guillain-Barré syndrome presenting as isolated acute dysphagia with full recovery

Yoon M¹, Lee S, Yoo Y

¹Department of Rehabilitation Medicine, St. Vincent's Hospital, College of Medicine, The Catholic University of Korea, Seoul, South Korea

43P Sporadic late-onset nemaline myopathy with monoclonal gammopathy: defining a more aggressive subtype with worse prognosis

Lauletta A¹, Merlonghi G, Forcina F, Fionda L, Leonardi L, Costanzo R, Tufano L, Rossini E, Marando D, Vera V, Antonini G, Morino S, Garibaldi M

¹Neuromuscular and Rare Disease Centre, Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University of Rome, Sant'Andrea Hospital, Rome, Italy

44P High-resolution spatial transcriptomics reveals myofiber-immune cell interactions in inclusion body myositis

de Haan S¹, Heezen L, Mahfouz A, Kan H, Badrising U, Spitali P

¹Leiden University Medical Center, Leiden, Netherlands

45P GRO chemokines-induced neutrophil extracellular trap CITH3 as a new biomarker related to myasthenia gravis severity

Zhong H¹, Zhang W, He D, Chen R, Jin L, Song J, Yan C, Huan X, Chen Y, Li X, Zhao C, Luo S

¹Huashan Hospital, Fudan University, Shanghai, China

46P The endothelial DLL4-muscular NOTCH2 axis drives muscle atrophy in dermatomyositis: insights from single-nucleus transcriptomics

Hayashi S¹, Kato M, Noguchi S, Nishino I

¹Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan

47P Intramuscular lipid accumulation and metabolic alterations in immune-mediated necrotizing myopathy: a comparative study of anti-HMGCR and anti-SRP subtypes

Focks N¹, Eisenlohr P, Baum O, Stenzel W, Kleefeld F

¹Klinik für Neurologie, Berlin, Germany

48P A closer look at the contribution of fibro-adipogenic precursors in the pathomechanism of autoimmune myositis: correlating transcriptomics with muscle pathology

De Los Reyes F¹, Hayashi S, Noguchi S, Nishino I

¹Department of Neuromuscular Research, National Institute of Neuroscience, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Kodaira, Tokyo, Japan

49P Aberrant expression of THE1B endogenous retrovirus fusion transcripts in sarcoid myopathy

Funaguma S¹, Iida A, Saito Y, Tanboon J, De Los Reyes F, Sonehara K, Goto Y, Okada Y, Hayashi S, Nishino I

¹National Center of Neurology and Psychiatry, Tokyo, Japan

50P Inflammatory cells immunophenotyping, MHC1 and type1 interferon proteins expression in myositis and hereditary muscle diseases with inflammatory cell infiltration: a north African study

Farhat E¹, Ben Hmid A, Ben Ahmed M

¹The Muscle Lab, Iris Medical Centre, Hedi Noura Avenue, Ennasr2, 2001 Ariana., Tunisia

51VP Mitochondrial pathology in Mi-2 beta dermatomyositis with lymphocyte fibre invasion

Cotta A¹, Carvalho E, da-Cunha-Junior A, Nunes-Neves S, Cordeiro B, De-Alencar-Hornsby A, Cauhi A, Coutinho-Passos S, Valicek J

¹The Sarah Network of Rehabilitation Hospitals, Belo Horizonte, Brazil

52P Clinical and radiological profile of a cohort of anti-HMGCR myopathy

Vengalil S¹, Ahmed S, Nashi S, Menon D, Mahadevan A, Nalini A

¹National Institute of Mental Health and Neurosciences, Bangalore, India

53VP Neutrophil-to-lymphocyte ratio (NLR) and platelet-to-lymphocyte ratio (PLR) as prognostic markers in idiopathic inflammatory myopathies

Hakim F^{1,2}, Sakka S, Snoussi M, Bouattour N, Moalla K, Daoud S, Charfi N, Marzouk S, Damak M

¹Department Of Adult Neurology, Habib Bourguiba Hospital, Sfax, Tunisia, ²Research Laboratory LR12SP19, Habib Bourguiba University Hospital, University of Sfax, Tunisia

54VP Distinct histopathological features of juvenile idiopathic inflammatory myopathies: a quantitative comparative study

Hakim F¹, Sakka S, Snoussi M, Boudawara O, Moalla K, Bouattour N, Daoud S, Charfi N, Sellami L, Marzouk S, Boudawara T, Damak M

¹Department Of Adult Neurology, Habib Bourguiba Hospital, Sfax, Tunisia

55VP Detailed analysis of muscle testing using the medical research council scale in different subgroups of idiopathic inflammatory myopathies

Hakim F^{1,2}, Snoussi M, Sakka S, Daoud S, Bouattour N, Moalla K, Charfi N, Marzouk S, Damak M

¹Department Of Adult Neurology, Habib Bourguiba Hospital, Sfax, Tunisia, ²Research laboratory LR-12-SP-19, Habib Bourguiba University Hospital, University of Sfax, Sfax, Tunisia

Poster Session 1 – 166P-192P: DMD – imaging and outcome measures

166P Early functional trajectories in young boys with Duchenne muscular dystrophy: a 24-month international study using the NSAA

Coratti G¹, Paolucci S, Baranello G, Expósito-Escudero J, Wolfe A, Brooke M, Pane M, Nascimento A, Messina S, Ricci F, D'Amico A, Bello L, Bruno C, Sansone V, Masson R, Nigro V, Muntoni F, Mercuri E International DMD network.

¹Catholic University of Sacred Heart, Rome, Italy

167P Qualitative interviews with caregivers of children aged 4–7 years with Duchenne muscular dystrophy to assess content validity of the proxy-completed DMD-QOL

Pearlmutter A¹, Powell D, Carlton D, Chen E, Gallington K, Bacci D, Gelhorn D, Cho M, Dashiell-Aje D

¹Biomarin Pharmaceutical, Inc., San Rafael, United States

168P Centiles for respiratory function in paediatric, GC-treated boys with Duchenne muscular dystrophy in London

Stimpson G¹, Zambon A, Lavery A, Ridout D, Brusa C, Wolfe A, Milev E, O'Reilly E, Manzur A, Sarkozy A, Baranello G, Muntoni F

¹Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom

169P Comparison of western-blot, mass spectrometry and simple-western methods shows that simple-western is the most sensitive method to detect μ -dystrophin

Buscara L¹, Genries-Ferrand S, Varela Moreira C, Stiet N, Cedrone G, Sagrere C, Salsac F, El-Khoury R, Bertil-Froidevaux E, Georger C, Blaie S, Thibaut L, Cao F, Braun S, Perret G, Blatzer M, Daniele N

¹Genethon, EVRY, France

170P Heart rate and ventilation during submaximal cycling exercise as physiological outcome measures in Duchenne muscular dystrophy

Taivassalo T¹, Paul M, Duong T, Lott D, Forbes S, Shih R, Leon Astudillo C, Sullivan R, Sweeney L

¹University Of Florida, Gainesville, United States

172P Clinical and genetic characteristics of adult patients with Duchenne muscular dystrophy

Leonardis L^{1,2}, Matajuro N, Teran N

¹University Medical Centre Ljubljana, Ljubljana, Slovenia, ²University of Ljubljana, Ljubljana, Slovenia

173P Longitudinal analysis of a patient-reported outcome (ACTIVLIM) in Duchenne muscular dystrophy: a registry-based study

Cosyns M¹, Jagut M, De Meulemeester N, Baets J, Beysen D, Claeys K, Deconinck N, De Bleecker J, De Waele L, Dubuisson N, Kinet V, Pelc K, Remiche G, Servais L, Smeets N, Vanden Brande L, Van Lander A, Vaes A

¹Sciensano, Belgian Public Health Institute, Brussels, Belgium

174P How can we analyze natural history across multiple sites without moving the data? – a feasibility study and early pilot using PUL 2.0

Muntoni F¹, Niks E, Michaëls M, Brooke M, Meijer-Krom Y, Wong B, Servais L, Franke W, Kurps J, Freimark J, Billmyer E, Marden J, Signorovitch J, Ward S

¹University College London Great Ormond Street Institute of Child Health, London, United Kingdom

175P A prognostic score for time to loss of ability to rise from supine in Duchenne muscular dystrophy (DMD)

Muntoni F¹, Signorovitch J, Marden J, Done N, Wang S, Akbarnejad H, Kang H, Li T, Ward S, Manzur A, Goemans N, Straub V, Mercuri E, Servais L

¹University College London Great Ormond Street Institute of Child Health, London, United Kingdom

176P 3-Year functional outcomes of patients with Duchenne muscular dystrophy: pooled delandistrogene moxeparvovec clinical trial data vs external controls

Mendell J, Connolly A, Day J, McDonald C, Proud C, Shieh P, Zaidman C, Furgerson M, Ding K, Reid C, **Murphy A¹**, Rodino-Klapac L

¹Roche Products Ltd, Welwyn Garden City, United Kingdom

177P Longitudinal stride-level evaluation of ambulatory function with ankle wearable technology in ambulant DMD patients below 4 years old

Poleur M¹, Parinello G, Vrščaj E, Bisson C, Cluzeau C, Daron A, University Children's Hospital L, Osredkar D, Strijbos P, Eggenspieler D, Servais L

¹NMRC, Citadelle, Liège, Belgium

178P Two-year stride-level evaluation of ambulatory function in ambulant DMD patients above 4 years old

Poleur M¹, Parinello G, Vrščaj E, Dolanska A, Nowakowski P, Anghelescu C, Szabo L, Leanca M, Mirea A, Kodszy S, Saleh A, Osredkar D, Haberlova J, Potulska-Chromik A, Butoianu N, Delmar P, Strijbos P, Eggenspieler D, Servais L, the ActiLiège-Next study group

¹NMRC, Citadelle, Liège, Belgium

179P Methodology of the CINRG expanded Duchenne natural history study (eDNHS): assessing disease progression in the era of glucocorticoid and gene therapy treatments

McDonald C^{1,2}, Henricson E, Duong T, Gordish-Dressman H, Morgenroth L, eDNHS Investigators C

¹UC Davis Health, Sacramento, United States, ²CINRG, Pittsburgh, United States

180P Exploring motor function decline in lower and upper extremity function across different genotypes in Duchenne muscular dystrophy

Fleerackers E^{1,2,3}, Broos J, Hoek R, Krom Y, Michaels M, Kan H, van Duyvenvoorde H, van der Holst M, Niks E

¹Department of Neurology, Leiden University Medical Center, Leiden, Netherlands, ²Department of Orthopaedics, Rehabilitation & Physical therapy, Leiden, Netherlands, ³Duchenne Center Netherlands, Leiden, Netherlands

181P AAV gene therapy with delandistrogene moxeparvovec in two-year old patients with Duchenne muscular dystrophy: clinical efficacy measured by digital endpoints

Grotski C¹, Servais L, Eggenspieler D, Henricson E, Strijbos P, Sidiropoulos P, Finkel R, McDonald C

¹UC Davis, Sacramento, United States

182P Automatic calculation of muscle volume of leg CT images, using artificial intelligence

Nakayama T¹, Kimizuka J, Fumiya I

¹Yokohama Rosai Hospital, Yokohama, Japan

183P The impact of genotype on north star ambulatory assessment in individuals with Duchenne muscular dystrophy treated with corticosteroids: a single-center study of 416 patients

Bonarrigo K¹, Goldsbury C, Horn P, Vilaisaktipakorn P, Reebals L, Zygmunt A, Tian C

¹Cincinnati Children's Hospital Medical Center, Cincinnati, United States, University of Cincinnati College of Medicine, Cincinnati, United States

184P Ultrasound vs MRI for assessing the EDB muscle in Duchenne muscular dystrophy

Karachunski P¹, Martone J, Stark M, Nascene D, Nordseth T, Kang P

¹University of Minnesota, Minneapolis, United States

185P Changes in timed items in DMD patients amenable to skipping exons 44, 45, 51 and 53: a 24-month collaborative study

Cicala G^{1,2}, Coratti G, Paolucci S, Baranello G, Exposito Escudero J, Wolfe A, Brooke M, Messina S, Ricci F, D'Amico A, Bello L, Bruno C, Sansone V, Masson R, Nigro V, Pane M, Nascimento A, Muntoni F, Mercuri E, on behalf of the Italian DMD network

¹Pediatric Neurology, Università Cattolica del Sacro Cuore, Rome, Italy, ²Centro Clinico Nemo, Fondazione Agostino Gemelli IRCCS, Rome, Italy

186P Performance of upper limb in steroid-treated Duchenne muscular dystrophy: genotype-phenotype correlations

Wade C^{1,2}, Zygmunt A, Horn P, Bonarrigo K, Reebals L, Rybalsky I, Tian C

¹Cincinnati Children's Hospital Medical Center, Cincinnati, United States, ²University of Cincinnati, Cincinnati, United States

187P Predicting long-term trajectories of performance of upper limb 2.0 (PUL) score in patients with Duchenne muscular dystrophy (DMD) via remote analytical collaboration

Michaëls M¹, Freimark J, Billmyer E, Hoek R, Krom Y, Fleerackers E, van der Holst M, Helleman K, van Weperen P, Straathof C, Marden J, Chen Z, Zhang W, Signorovitch J, Ward S, Niks E

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188P Bilateral analysis of upper limb endpoints in ambulant and non-ambulant DMD patients

Michaëls M^{1,2}, Prins A, Fleerackers E, van der Holst M, van Zwet E, Kan H, Niks E

¹Department of Neurology, Leiden University Medical Center, Leiden, Netherlands, ²Duchenne Center Netherlands, Leiden, Netherlands

189P Electrical impedance myography as a tool for monitoring disease progression in mouse models of Duchenne muscular dystrophy: a longitudinal, multisite study

Mantuano P¹, Prins A, Fleerackers E, van der Holst M, van Zwet E, Kan H, Niks E

¹Department of Pharmacy – Drug Sciences, University of Bari “Aldo Moro”, Bari, Italy

190P Sarcopenia severity in dystrophinopathy patients in Puerto Rico

Arrillaga Cruz A, Cortes Nieves B, García Rivera J, López Medrano G, **Ramos E**¹

¹University Of Puerto Rico, San Juan, United States

191P Comprehensive analysis of longitudinal SV95C measurements, an e-digital mobility assessment in a real-life DMD population in the GNT-014-MDYF natural history study

Montier T, De Lucia S, Davion J, Espil C, Guglieri M, Chabrol B, Le Goff L, Seferian A, Perret G, Guemas E, Valent A, **Cao F**¹, Laugel V, Muntoni F

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192P GNT0004, Genethon's AAV-based gene therapy for Duchenne muscular dystrophy: long-term follow-up of ambulatory boys enrolled in the dose-escalation phase of GNT-016-MDYF

Laugel V, De Lucia S, Davion J, Daniele N, Cao F, Sanz M, BUSCARA L, Blaie S, Thibaut L, Sagot M, Riviere A, Creoff E, Lelait M, Valent A, **Perret G**¹, Braun S, Muntoni F

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298P Analysis of long-term clinical and MRI findings highlights progressive nature of Ano5-related myopathies and the role of imaging as a valuable outcome measure

Aragon Gawinska K^{1,2}, Otero Borell M, Martí P, Azorín I, Tarrega M, Sivera R, Vázquez-Costa J, Vilchez R, Vilchez J, Sevilla T, Muelas N

¹Neuromuscular Unit, Department of Neurology, Hospital Politècnic i Universitari La Fe, Valencia, Spain, ²Neuromuscular and Ataxias Research Group, Instituto de Investigación Sanitaria La Fe, Valencia, Spain

299P A generalizable deep-learning muscle segmentation model for multicentre and multi-study muscle MRI in neuromuscular diseases

Bolaño Diaz C¹, Verdu Diaz J, Gonzalez Chamorro A, Fitzsimmons S, Hao D, Kocak G, Mannion J, Wandera S, Borland H, Myo-Guide Consortium, Tasca G, Bacardit J, Straub V, Diaz Manera J

¹John Walton Muscular Dystrophy Research Centre - Newcastle University, Newcastle upon Tyne, United Kingdom

300P The role of muscle MRI in mild neuromuscular disorders. where are we?

Astrea G, **Buchignani B**^{1,2}, Schifino M, Rubegni A, Galatolo D, Pasquariello R, Battini R, Santorelli F

¹Ircs Stella Maris Foundation, Pisa, Italy, ²University of Pisa, Italy

301P Patterns of volume and fat infiltration in skeletal muscle of adults with spinal muscular atrophy

Duong T¹, Anna Yao M, Yoseph Hailu R, Vogt-Domke S, Day J, Hageman N

¹Stanford University, Palo Alto, United States

302P Whole-body fat-referenced MRI measures disease progression in patients with spinal and bulbar muscular atrophy

Karlsson M¹, Widholm P, Ahlgren A, Hatsis P, Gentry C, Grunseich C, Gharib A, Kokkinis A, AlQahtani A, Fratta P, Zampedri L, Jayaseelan D, McKenzie H, Wastling S, Katsuno M, Yamada S, Kishimoto Y, Kawase T, Taoka T, Vigiletta V

¹AMRA Medical AB, Linköping, Sweden

303P MRI evaluation of GNE myopathy in a tertiary neuromuscular centre in London

Leitermann V¹, Cross S, DuPreez H, Whiteley-Jones H, Millner T, Radunovic A

¹Royal London Hospital, Barts Health NHS Trust, London, United Kingdom

304P A general-purpose self-supervised AI model for quantitative analysis of H&E-stained muscle biopsy specimens' whole-slide images

Marczyk M^{1,2}, Schoser B, Dreyling C, Santorelli F, Dal Canto F, Tupler R, Polanska J

¹Silesian University of Technology, Gliwice, Poland, ²Yale School of Medicine, New Haven, United States

305P Tracking muscle degeneration and disease activity in FSHD using qualitative longitudinal MR imaging

Mayer A¹, Starke A, Türk M, Uder M, Dörfel A, Nagel A, Gazzero E, Gerhalter T

¹Universitätsklinikum Erlangen, Erlangen, Germany

306P Evaluation of tongue volume in spinal and bulbar muscular atrophy (SBMA) by AI-assisted automatic MRI-analysis

Nguyen-Younossi N¹, Vernikouskaya I, Müller H, Chadraabal K, Gadelkareem M, Rasche V, Kassubek J, Rosenbohm A

¹Department of Neurology, University of Ulm, Ulm, Germany

307P S-adenosylhomocysteine hydrolase deficiency associated with severe paraspinal muscle weakness and posteromedial thigh involvement on muscle imaging

Paprad T¹, Paprad T, Sudhakar S, Hawkins C, Baruteau J, Gonorazky H, Yoon G

¹The Hospital for Sick Children Canada, Toronto, Canada

308P Quantitative, clinical, and patient-reported outcome measures in patients with limb-girdle muscular dystrophy type R12: a 3-year follow-up study

Poulsen N¹, Kass K, Schmidt K, Lyu Z, Teitsdóttir B, Sheikh A, Nielsen E, Hansen A, Wilms G, Khawajazada T, Witting N, Ladefoged C, Vissing J

¹Copenhagen Neuromuscular Center, Rigshospitalet, Copenhagen, Denmark

309P Impaired breathing in Pompe disease: real-time MRI with deep learning as novel tool to evaluate diaphragm vs. chest movement in neuromuscular disorders

Zeng R, Al-Bourini O, Töpert L, Lettermann L, Olgemöller U, Hofer S, Boentert M, Friede T, Nietert M, Voit D,

Frahm J, Uecker M, Seif Amir Hosseini A, **Schmidt J**^{1,2}

¹Immanuel University Hospital Rüdersdorf, Brandenburg Medical School, Rüdersdorf bei Berlin, Germany, ²University Medical Center Göttingen, Department of Neurology, Göttingen, Germany

310P Glycogen depletion in healthy controls using carbon-13 magnetic resonance spectroscopy

Slipsager A¹, Stemmerik M, Ankjær P, Teitsdóttir B, Vissing J

¹Copenhagen Neuromuscular Center, Rigshospitalet, Copenhagen, Denmark

311P Stratification of neuromuscular disorder types based on the subregional radiomic features in lower leg MRI

Socha M¹, Piorecki L, Verdú-Díaz J, Diaz-Manera J, Straub V, Tupler R, Polanska J

¹Silesian University of Technology, Gliwice, Poland

312P Clinical and muscle MRI findings of Bethlem myopathy: case series

Öz Tunçer G, Kurt Bayir G, Sezer Ö, **Yayici Koken O¹**, Aksoy A

¹Division of Pediatric Neurology, Department of Pediatrics, Faculty of Medicine, Antalya, Turkey

313P Whole-body muscle imaging in ADSS1 myopathy — revealing progressive impairment of lower extremities with preservation of trunk muscles

Saito Y¹, Yoshioka W, Eura N, Hayashi S, Noguchi S, Nishino I

¹Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Kodaira, Japan

Poster Session 1 – 399P-426P, 427VP-430VP: Clinical trials, access to health care and outcome measures

399P Measuring upper and lower limb movement with syde® in patients with facioscapulohumeral muscular dystrophy (FSHD): analytical validation in a controlled environment

Ankjaer P¹, Stemmerik M, Teitsdøttir B, Slipsager A, Desire L, Noblot N, Piquet O, Motola S, Tricot A, Vissing J, Preisler N

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400P Decision trees of peak torque reference values for distinguishing between normal and abnormal muscle function: a systematic review

Breninkmeijer R¹, Biziaki Ansanello N, Mattiello-Sverzut A, Bartels B

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401P Implementation of inertial sensors for motor assessment in pediatric neuromuscular disorders: preliminary results from a pilot study

D'Alessandro R^{1,2}, Cavallina I, Panero E, Sertori F, Rolle E, Rossi F, Monagheddu L, Gastaldi L, Mongini T, Ricci F

¹Department of Neurosciences "Rita Levi Montalcini", Neuromuscular Unit, A.O.U. Città della Salute e della Scienza di Torino, Molinette Hospital, University of Turin, Turin, Italy, ²Department of Public Health and Pediatric Sciences, Section of Child and Adolescent Neuropsychiatry, A.O.U. Città della Salute e della Scienza di Torino, "Regina Margherita" Pediatric Hospital, University of Turin, Turin, Italy

402P Assessing function and metabolic cost: development of the neuromuscular disease specific activity questionnaire (NAQ)

Duong T¹, Wilcox R, de Monts C, Christle J, Tang W, Khonde S, Wheeler M, Ashley E, Birkmeier M, Meyer J

¹Stanford University Neurology Department, Palo Alto, United States

403P Impact of healthcare disparities, specialized care, and socioeconomic factors on Duchenne muscular dystrophy patient's outcome: insights from Egyptian referral center

El Sherif R^{1,2}, Lowes L

¹New Giza University, Cairo, Egypt, ²Myo-Care Foundation

404P First clinical trial in SELENON-related myopathy: analysis of the phase II-III pilot SELNAC study

Villar Quiles R, Behin A, Prigent H, Gassama M, Ait Younes N, Quijano Roy S, Amthor S, Grimaldi L, Pincemail J, David O, Estournet B, **Ferreiro A^{1,2}**

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405P Video-based digital endpoints for the home monitoring of patients with neuromuscular diseases

Hogrel J¹, Marques T, Santmarty P, Decostre V, Drummond E, Muni Lofra R, Straub V, DT4RD Consortium T

¹Institute of Myology, Paris, France

406P Validity and reliability of a new method for visual analysis of scapular kinesis (VASK) in children and adolescents

Hoogveld L¹, Pelsma M, Meijler D, Ijspeert J, Braakman H, Houwen S

¹Department of Rehabilitation, Donders Institute for Brain, Cognition and Behavior, Radboud university medical center, Amalia Children's Hospital, Nijmegen, Netherlands

407P Familial congenital myopathy due to digenic pathogenic variants in the SRPK3 and TTN genes

Hüpper A¹, Gosk-Tomek M, Schwerin-Nagel A, Schmidt I, Mahal S, Lilja S, Paquay A, Rosensteiner B, Bernert G, Bittner R, Schmidt W

¹Klinik Favoriten, Vienna, Austria, Vienna, Austria

408P Natural disease progression in adult CMT1A: a prospective study using quantitative MRI and clinical assessments

Iterbeke L¹, Huysmans L, Bamps K, Peeters R, Goossens V, Maes F, Dupont P, Claeys K

¹Laboratory for Muscle Diseases and Neuropathies, Department of Neurosciences, KU Leuven, and Leuven Brain Institute (LBI), Leuven, Belgium

409P Double-blind, placebo-controlled study assessing effects of tranilast on motor, respiratory, and cardiac functions in Duchenne muscular dystrophy

Matsumura T¹, Wakisaka A, Haginoya K, Takeshima Y, Funato M, Hattori A, Ishigaki K, Nakamura A, Komaki H, Ishikawa Y, Arahata H, Furukawa T, Inoue K, Kimura K, Nakamura H, Ishizuka T, Hashimoto H, Saito M. A, Sekimizu M, Iwata Y

¹NHO Osaka Toneyama Medical Center, Toyonaka, Japan

410P Intrathecal onasemnogene abeparvovec for treatment-experienced patients with spinal muscular atrophy: phase 3b, open-label STRENGTH study

Kwon J, Munell F, Le Goff L, Yuge K, Kato T, Cances C, De Waele L, Woodcock I, **Mercuri E¹**, Proud C, Darras B, Hayes L, Oskoui M, Visootsak J, Williams G, Lee A, Illic A, Yang L, van der Pol W

¹Fondazione Policlinico Gemelli, Rome, Italy

411P Clinical progress update for SAT-3247, a first-of-its-kind, orally administered small molecule drug to address deficits in muscle repair and regeneration in Duchenne

Mitchell R¹, Dubow J, Wells C, Lambert P, Rudnicki M, Gleeson F

¹Satellos Bioscience Inc., Toronto, Canada

412P Goal attainment scaling (GAS): a patient-centred outcome in neuromuscular disease

Oude Elberink I¹, Roes K, Van der Woude D, Ruyten T, Mulder S, Van Eijk R, Van der Pol L, Bartels B

¹Child Development and Exercise Center, Wilhelmina Children's Hospital, University Medical Center Utrecht, Utrecht, Netherlands

413P Optimizing seating solutions in neuromuscular disorders: insights from a resource-constrained environment

Paul M¹, Varghese V, Rajadurai I, Mathew A

¹Bangalore Baptist Hospital, Bengaluru, India

414P Feasibility of using a wearable digital health technology sensor to assess ambulation in CMT and DM1

Poleur M¹, Delstanche S, Parinello G, Bisson C, Cluzeau C, Dubois C, Duclos M, Benmhammed N, Eggensteiner D, Servais L

¹NMRC, Citadelle, Liège, Belgium

415P Intrathecal onasemnogene abeparvovec for patients with spinal muscular atrophy: phase 3, randomized, sham-controlled, double-blind STEER study

Proud C¹, Vũ D, Wilmschurst J, Sanmaneechai O, Gulati S, Xiong H, Ceja Moreno H, Tay S, Thong M, Born A, Banzatto Ortega A, Jong Y, Al-Muhaizea M, Lee A, Visootsak J, Tauscher-Wisniewski S, Alecu I, Parlikar R, Finkel* R

¹Children's Hospital of the King's Daughters, Norfolk, United States

416P The long road to diagnosis: the burden of delayed diagnosis of hereditary myopathy in Indonesian adults

Indrawati L, **Simatupang S¹**, Putri M, Issac W, Shafitha N, Wiratman W, Budikayanti A, Octaviana F, Safri A, Fadli N, Harsono A, Hakim M

¹Department of Neurology, Dr Cipto Mangunkusumo Hospital, Jakarta, Indonesia

417P A pilot study to explore a non-invasive measure of mitochondrial dysfunction

Suetterlin K¹, Devine H, Brito Da Silva A, Smith S, Hillman S, Rao K, Brownstein C, Turnbull D, Bulea T

¹Newcastle University, Newcastle Upon Tyne, United Kingdom

418P Safety and efficacy of ultrasound-guided nerve block in muscle biopsy

Tawara N¹, Shibata Y, Hara K, Nagatoshi C, Fujimoto A, Ishizaki M, Kurisaki R, Nishida Y, Maeda Y, Ueyama H

¹Department of Neurology, NHO Kumamoto Saishun Medical Center, Kumamoto, Japan

419P Baseline characteristics of patients with congenital myopathies from a 2-year prospective natural history study (READYCOM)

Van De Camp S¹, Van Doorn J, De Laat E, Groothuis J, Van Alfen N, Jungbluth H, Erasmus C, Bartels B, Wadman R, Voet N, Van Der Pol L, Voermans N

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420P Measurable difference neutral vertical head/trunk control between children with neuromuscular disorders and typically developing children

Willis T^{1,2}, Sakanaka T, Kulshrestha R, Butler P, Loram I

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421P Is neutral vertical head and trunk control attained in children with spinal muscular atrophy treated with disease-modifying therapies?

Willis T¹, Sakanaka T, Butler P, Kulshrestha R, Loram I

¹Robert Jones and Agnes Hunt Hospital, Oswestry, United Kingdom, ²Chester University Medical School, Chester, United Kingdom

422P NURTURE study: long-term benefits of nusinersen in presymptomatic spinal muscular atrophy (SMA) over 8 years of Follow-up

Kuntz N¹, De Vivo D, Crawford T, Kirschner J, Finkel R, Bertini E, Parsons J, Hwu W, Pechmann A, Butterfield R, Foster R, Littauer R, Fraser K, Tichler B, Fradette S

¹Lurie Children's Hospital of Chicago, Chicago, Illinois, United States

423P RAINBOWFISH: 3-year efficacy and safety data of risdiplam in infants with presymptomatic spinal muscular atrophy (SMA)

Mazurkiewicz-Beldzińska M¹, Finkel R, Servais L, Farrar M, Vlodavets D, Zanoteli E, Al-Muhaizea M, Araújo A, Nelson L, Kuthiala M, Gorni K, Kletzl H, Palfreeman L, Gaki E, Rabbia M, Summers D, Manfrini M, Bertini E, on behalf of the RAINBOWFISH Study Group

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424P Delandistrogene moxeparvovec micro-dystrophin expression and safety in 3–4-year-olds with Duchenne muscular dystrophy in ENDEAVOR and ENVOL studies

McDonald C¹, Mendell J, Mercuri E, Gangfuss A, Nascimento A, Desguerre I, Zaidman C, Servais L, Thrasher P, Vivien M, Ding K, Murphy A, Zhang B, Wonde K, Pordeli P, Rodino-Klapac L

¹UC Davis Health, Sacramento, United States

425P RGX-202, an investigational gene therapy for the treatment of Duchenne muscular dystrophy: interim clinical data

Pakola S¹, Harper A, Iannacone S, Kuntz N, Tesi-Rocha C, Veerapandiyan A, Phillips D, Wilson C, Boulos N, Tsao E, Gilmor M, Curtiss S, Patel H, Fiscella M, Danos O, Dastgir J

¹REGENXBIO, Rockville, United States

426P Efficacy and safety of apitegromab in patients aged 13-21 years with type 2 or 3 spinal muscular atrophy: outcomes from the SAPHIRE phase 3 trial

Seferian A^{1,2,3}, Crawford T, Servais L, Mercuri E, Krueger J, Köbel H, Cances C, Kuntz N, Finkel R, Yao B, Rossello J, Tirucheraï G, Zhao G, Song G, Marantz J, Darras B

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427VP Is muscle biopsy needed in diagnosing Duchenne muscular dystrophy diagnosis: role of 6-minute walk test

Banavara Shyamprasad S¹, Satyam P, Balwani H, Herle S, Khandekar G, Thomas N, Mathew A

¹Synapse Neuro Center, BENGALURU, India

428P Gait analysis with trunk accelerometry in patients from the French registry of glycogen storage disease type III: implications for clinical trials

Hogrel J, Fer F, Ledoux I, Petit F, Bello M, Labrune P, Wahbi K, Habes D, Gardin A, Masingue M, Laforêt P, **Decostre V¹**

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429VP The BrAAVe study: a phase 1/2 study to investigate the safety, tolerability, and efficacy of SAR446268, an AAV-mediated gene therapy in non-congenital DM1 patients

Galeon J¹, Rouy D, Rendo P

¹Sanofi, Boston, United States

430VP Care of dystrophinopathy carriers: survey results of provider practices in the U.S.

Scavina M¹, Banks K, Cappa R, Chambers C

¹Parent Project Muscular Dystrophy, Washington, DC, United States

Poster Session 1 – 505P-524P, 525VP, 526P-528P, 529VP, 530P-536P, 537VP: SMA clinical

505P Neuromuscular taping for drop hand rehabilitation in an early-treated infant with SMA1: a case report

Branca A¹, Saraceni V, Onofri M, Briganti M, Sancricca C, Frongia A

¹Fondazione UILDM Lazio Onlus, Rome, Italy

506P Investigating SMN-associated neurodevelopmental disorders: a comprehensive approach

Brusa C^{1,2}, Saini M, Suarez Rivera C, Vargha-Khadem F, de Haan M, Manzur A, Munot P, Muntoni F, Scoto M, Baranello G

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507P Overview of spinal muscular atrophy disease area from the perspective of patients and caregivers in Turkey: a survey study

Carman K¹, Aksakal F

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508P Interventional radiology and spinal surgical techniques allow nusinersen treatment continuation in spinal muscular atrophy

Cornell N^{1,2}, Manzur A, Roberts S, Harris M, Tucker S, Rennie A, Robertson F, Smith N, Munot P, Baranello G, Muntoni F, Scoto M

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509P The changing phenotype of spinal muscular atrophy type 3 since the arrival of disease modifying treatments

Cornell N^{1,2}, O'Reilly E, Roberts S, Harris M, Tucker S, Fontanaz D, Parton M, Baranello G, Munot P, Manzur A, Muntoni F, Scoto M

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510P Elective late preterm birth for early treatment of a child with spinal muscular atrophy

Dangouloff T¹, Chantraine F, Hennuy N, Rigo V, Tribolet S, Vanden Brande L, Servais L

¹Neuromuscular Reference Center, Department of Pediatrics, University Hospital Liege & University of Liege, Liege, Belgium

511P A real-world perspective on longitudinal quantitative muscle strength in treated adults with SMA

De Monts De Savasse C¹, Dunaway Young S, Vogt-Domke S, Yatsu L, Yoseph Hailu R, Salvatore S, Tang W, Britts C, Parker D, Day J, Duong T

¹Stanford University, Department of Neurology, Palo Alto, United States

512P SMN2 copy number and SMA severity: a paradigm under review?

Garcia Uzquiano R¹, Sotoca J, Puig-Ram C, Fernandez-Garcia M, Expósito Escudero J, Toledo Bravo de la Laguna L, Martinez-Salcedo E, Gonzalez-Mera L, Fernandez Ramos J, Tizzano E, Nascimento Osorio A, on behalf of CuidAME Investigators Group.

¹Sant Joan de Deu Hospital, Barcelona, Spain

513P HINALEA 2: baseline observations in a trial of risdiplam in patients with SMA who experienced a plateau or decline in function after gene therapy

Gorni K¹, Hahn A, Kotulska-Jozwiak K, Keto E, Li Y, Guittari C

¹PDMA Neuroscience and Rare Disease, F. Hoffmann-La Roche Ltd, Basel, Switzerland

514P Quebec spinal muscular atrophy newborn screening program: first year overview

Groulx-Boivin E^{1,2}, Belzile A, Gauthier A, Michaud-Gosselin C, Giguère Y, Laberge A, Chrestian N, Nguyen C, Oskoui M

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515P Description of the Algerian experience in the diagnostic approach of spinal muscular disease

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516P Newborn screening for spinal muscular atrophy: first anniversary in Switzerland

Jacquier D¹, Klein A, Stettner G, Broser P, Fluss J, Enzmann C, Goeggel Simonetti B9, Sluka S, Baumgartner M, Hersberger M, Tschertner A

¹Pediatric neurology and neurorehabilitation unit, Mother-Woman-Child Department, Lausanne University Hospital, University of Lausanne, Lausanne, Switzerland

517P Adult SMA REACH: characterisation of adult patients living with spinal muscular atrophy at start of treatment in the UK

Karkkainen E^{1,2}, Page J, Carver A, Tanner S, Moat D, Benesperi G, Michell-Sodhi J, Verdu Diaz J, Muni Lofra R, Marini-Bettolo C, Adult SMA REACH Network

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518P Clinical outcome and effectiveness of onasemnogene abeparvovec in treatment-naïve spinal muscular atrophy

Lee H¹

¹Yongin Severance Hospital, Yonsei University College of Medicine, Yongin-Si, South Korea

519P The challenge of swallowing assessment in SMA1: dysphagia clinical features and available assessment tools

Masson R¹, Dosi C, Parravicini S, Scopelliti M, Arnoldi M, Zanin R, Schindler A, Gandolfi S, SFERA working group

¹Developmental Neurology Unit, Fondazione IRCCS Istituto Neurologico Besta, Milan, Italy,

520P Safety update: risdiplam clinical trial program for spinal muscular atrophy (SMA)

Masson R¹, Baranello G, Servais L, Darras B, Day J, Deconinck N, Farrar M, Finkel R, Bertini E, Kirschner J, Mazurkiewicz-Beldzińska M, Bader-Weder S, Gorni K, Schlegel V, Li Y, Scalco R, Mercuri E, on behalf of the Firefish, Sunfish, Jewelfish and Rainbowfish Study Groups

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521P Feasibility of a novel rehabilitation intervention for children with spinal muscular atrophy

McAdam L^{1,2,3}, Ippolito C, Sin J, MacDonald C, Landry A, Luckhart K, Hoffman A

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522P SMA care UK: updating spinal management for people living with SMA across the UK.

Muni Lofra R¹, Christie-Brown V, Benesperi G, Bowey A, Scoto M, Abbott L, O'Reilly E, Roberts S, Tsirikos A, Hewitt S, Manchester E, Chatfield S, Emery N, Narayan S, Nevin K, Gregson S, Sheehan J, Childs A, Marini-Bettolo C

¹The John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom

523P Spinal muscular atrophy in an adult neurology clinic: delay in diagnosis to treatment

Mustafa F¹, Macken W, Dalal A, Rani N, Ahmed T, ICGNMD Consortium, Bhatia R, Pitceathly R, Kumarasamy T, Reilly M, Srivastava M, Hanna M, Vishnu V

¹All India Institute of Medical Sciences, New Delhi, New Delhi, India

524P Long-term use of disease-modifying therapies in spinal muscular atrophy – a population based study

Nungo Garzon N¹, Aragon-Gawinska K, Baviera Ricart M, Mora Tatay F, Ibañez Albert M, Hervás D, Pitarch Castellano I, Vazquez Costa J

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525VP Urine titin reveals muscle degradation undetected by serum CK in SMA patients with four copies of SMN2

Ohashi H¹, Bo R, Sonehara S, Nambu, Sunami A, Osawa K, Shirakawa T, Nishio H, Matsuo M, Awano H

¹Department of Pediatrics, Kobe University Graduate School of Medicine, Kobe, Japan

526P Trial design and rationale for OPAL, a phase 2, randomized, double-blind study of apitegromab in patients aged <2 years with spinal muscular atrophy

Servais L^{1,2}, Crawford T, Capocelli K, Yao B, Tirucherai G, Xu Y, Zhao G, Marantz J, Darras B

¹Department of Paediatrics, MDUK Oxford Neuromuscular Centre and NIHR Oxford Biomedical Research Centre, University of Oxford, Oxford, United Kingdom, ²Department of Paediatrics, Neuromuscular Reference Centre, University Hospital of Liège, Liège, Belgium

527P Scoliosis development in children and adolescents with spinal muscular atrophy: study design

Staaf P¹, Hjalmarsson E, Eklund E, Sofou K

¹Sahlgrenska University Hospital, Gothenburg, Sweden

528P Insights into DYNC1H1 variants: their role in neuromuscular and neurodevelopmental phenotypes

Tajsharghi H¹, Namdari M, Ansari B, Basiri K, Azimi E, Hosseinzadeh M, Bahreini A, Nouri N, Sedghi M, Fattahpur S, Amoli M

¹University Of Skovde, Skovde, Sweden

529VP Pharmacokinetics (PK) and pharmacodynamics (PD) of apitegromab in spinal muscular atrophy

Tirucherai G¹, Xu Y, Singh A, Bilic S, Gosselin N, Koeck K, Cote S, Yao B, Marantz J

¹Scholar Rock, Inc., Cambridge, United States

530P Non-5q spinal muscular atrophy (SMA) previously followed-up as 5q SMA IIIa – case report

Vosatkova A^{1,2}, Renczesova B, Martinka I, Kurca E, Turcanova Koprakova M

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531P A clinical-electrophysiological-integrated high risk screening method of spinal muscular atrophy in adolescents and adults: a multicenter retrospective study

Zhu W^{1,2,3}, Zhao Q, Wang N, Cao X, Gong S, Bu N, Jiang Q, Yin J, Luo J, Zhou X, Zhai W, Wang J, Lu J, Zhao C, Zhu S, Hu Y, Song X, Qiao K

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³Huashan Rare Disease Center, Huashan Hospital, Shanghai Medical College, Fudan University, , Shanghai, China

532P Review of the respiratory care requirements for patients affected by spinal muscular atrophy type 1 in a paediatric neuromuscular service in the recent era

Alhaswani Z¹, Stygall K, Bull L, Ninan T, Parasuraman D, Fullwood I

¹University Hospitals Birmingham NHS Trust, Birmingham, United Kingdom

533P Nationwide newborn screening for spinal muscular atrophy in the Netherlands

Asselman F¹, van der Woude D, Cuppen I, Wadman R, Seppenwoolde W, Bartels B, van der Pol L

¹University Medical Center Utrecht, Utrecht, Netherlands

534P Impaired renal function in patients with spinal muscular atrophy: a longitudinal cohort study

Asselman F¹, Meijvis S, Wadman R, Cuppen I, Vernooij R, Vermeer L, van den Berg L, Groen E, van der Pol L

¹University Medical Center Utrecht, Utrecht, Netherlands

535P Motor function trajectories of patients with spinal muscular atrophy following treatment switching

O'Reilly E^{1,2}, Stimpson G, Wolfe A, Milev E, Baranello G, Muntoni F, Scoto M

¹Dubowitz Neuromuscular Centre; UCL Great Ormond Street Institute of Child Health, London, United Kingdom, ²National Institute for Health Research at Great Ormond Street Hospital Biomedical Research Centre, London, United Kingdom

536P False-positive results in 5q-SMA newborn screening: SMN1 VUS studies in zebrafish support therapeutic decision

Kölbel H¹, Wirth B, Stringer B, Zhang Y, Taghipour-Sheshdeh A, Zetzsche S, Piano V, Fuhrmann N, Becker J, Karakaya M, Goh S, Giacomotto J, Farrar M

¹Department of Pediatric Neurology, Centre for Neuromuscular Disorders, University Hospital Essen, Essen, Germany

537VP Safety and efficacy of risdiplam for types 1-3 spinal muscular atrophy in a single center

Zhu X¹, Li H, Hu C, Wu M, Zhou S, Wang Y, Li W

¹Children's Hospital of Fudan University, National Children's Medical Center, Shanghai, China

Poster Session 1 – 651P-677P, 678VP-688VP: DMD - treatments

651P Long-term safety of givinostat in patients with Duchenne muscular dystrophy: results from an open-label extension study

Brandsema J¹, Acsadi G, Coceani N, Alessi F, Cazzaniga S, Bettica P, Finanger E

¹Children's Hospital of Philadelphia

652P RKER-065 improved the therapeutic response to phosphorodiamidate morpholino oligomers (PMO) in a Duchenne muscular dystrophy mouse model

Fisher F¹, Zhen G, Nathan R, Linzey M, Hack F, Cahill M, Daman T, Macaluso S, Todorova R, Grenha R, Lachey J, Lerner L, Seehra J

¹Keros Therapeutics, Lexington, United States

653P Mini muscles for a major cause: creating patient-derived organoids to model Duchenne muscular dystrophy (DMD)

Flynn-Rizk R¹, Egian M, Hoffman L

¹Western University, London, Canada, ²Lawson Health Research Institute, London, Canada

654P Preclinical efficacy of ENTR-601-50, a novel EEV™-oligonucleotide construct for the treatment of exon 50 skip-amenable Duchenne muscular dystrophy

Girgenrath M¹, Estrella N, Hicks A, McKee H, Beermann M, Cheng Zhang J, Brennan C, Tan P, Mou Y, Long N, Lian W, Stadheim A, Kheirabadi M, Dougherty P, Streeter M, Qian Z

¹Entrada Therapeutics, Boston, United States

655P Counteracting reactive oxygen species in models of Duchenne muscular dystrophy improves antisense-oligonucleotide mediated dystrophin recovery

Goossens R¹, Schneider A, Filonova G, Bijl I, Tanganyika-de Winter C, van Putten M, Aartsma-Rus A

¹Leiden University Medical Center, Dept. of Human Genetics, Leiden, Netherlands

656P Sustained functional improvement with DYNE-251 in males with DMD mutations amenable to exon 51 skipping enrolled in the phase 1/2 DELIVER trial

Guglieri M¹, Campbell C, Deconinck N, De Waele L, Flanigan K, Lorentzos M, Phan H, Shieh P, Ray S, Wang D, Dugar A, Naylor M, Kerr D, on behalf of the DELIVER study investigators

¹Royal Victoria Infirmary, Newcastle University, and Newcastle Hospitals NHS Foundation Trust, Newcastle-Upon-Tyne, United Kingdom

657P Lysosomal damage in Duchenne muscular dystrophy: an emerging therapeutic target

Jaber A, Palmieri L, Bakour R, Lachiver E, Vu Hong A, Bourg N, Pupiot J, Albini S, Stokholm D, Van Wittenberghe L, Miranda A, Daniele N, Barthélème I, Blot S, Thao Bui M, Evangelista T, Richard I, **Israeli D¹**

¹Genethon, INSERM UMR_S951, Evry Univerisity, Paris-Saclay University, Evry, France

658P 3.5-year safety and efficacy data of brogirdirsen from DMD patients in open-label extension study

Komaki H¹, Takeshita E, Kunitake K, Shimizu-Motohashi Y, Maruyama S, Zhang H, Miyagi R, Aoki Y

¹National Center of Neurology and Psychiatry, Tokyo, Japan

659P Pulmonary function in advanced-stage patients with Duchenne muscular dystrophy treated with casimersen

Kuntz N¹, Ung B, Khellaf M, Done N, Liu Y, Tuttle E, Grabich S

¹Lurie Children's Hospital of Chicago, Chicago, Illinois, United States

660P Delandistrogene moxeparvovec in Duchenne muscular dystrophy: experience from a single center

Laine R¹, Liberati C, Ghosh P

¹Boston Children's Hospital, Boston, United States

661P Givinostat effect on respiratory function in Duchenne muscular dystrophy before and after ambulation loss: results from EPIDYS, LTSE, and PRO-DMD-01

Mercuri E¹, McDonald C, Lavery C, Finanger E, Signorovitch J, Parodi A, Alessi F, Cazzaniga S, Bettica P, Péréon Y

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662P Immunoproteasome inhibition positively impacts the gut-muscle axis in Duchenne muscular dystrophy

Mostosi D¹, Farini A, Strati F, Molinaro M, Saccone S, Amoroso C, Cassani B, Leonetti E, Caprioli F, Facciotti F, Torrente Y

¹University of Milan, Milan, Italy

663P Real-world evidence of viltolarsen in Japanese boys with Duchenne muscular dystrophy: two-year results from the Remudy-DMD registry

Nakamura H¹, Oba M, Izumi S, Ishizuka H, Komaki H, Remudy DMD investigator group R

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664P Post fracture rehabilitation in Duchenne muscular dystrophy: challenges in co-ordinating multi-disciplinary care

Parampalli Ravindra S¹, Banavara S, Khandekar G, Varghese V, Agnes Mathew A

¹Synapse Neuro Centre, Bengaluru, Bengaluru, India, ²Bangalore Baptist Hospital, Bengaluru, India

665P GDF5 therapeutic potential for Duchenne muscular dystrophy

Pietri Rouxel F^{1,2}, Gentil C, Bourguiba A, Vergnol A, Cadot B, Guesmia Z, Saillard L, Meunier P, Falcone S, Giordani L

¹Association Institut De Myologie, Paris, France, ²Sorbonne Université, Centre de Recherche en Myologie – UMRS 974, Paris, France

666P ELEVATE-44-201, a phase 1/2b study to assess the safety and efficacy of ENTR-601-44 in patients with DMD amenable to exon 44 skipping

Servais L, **Porco D¹**, Stadheim A, Parasrampur R, Weinstein D, Sethuraman N

¹Entrada Therapeutics, Boston, United States

667P Modulating innate immunity in Duchenne cardiomyopathy by a small noncoding tRNA derivative

Rogers R¹, Rannou A, Manriquez N, Hong Y, Liu W, Fournier M, Alfaro J, Marbán E

¹Cedars-sinai Medical Center, Los Angeles, United States

668P Using muscle homing peptide CyPep10 to improve delivery of phosphorodiamidate morpholino oligomers in the mdx mouse

Schneider A¹, Tanganyika-de Winter C, Jirka S, Tan X, Thompson E, Ha K, Mitra A, Garcia S, Luimes M, Oliver R, Guerlavais V, Aartsma-Rus A

¹Leiden University Medical Centre - Department of Human Genetics, Leiden, Netherlands

669P Integrating digital tools for clinical trial discovery in dystrophinopathies: the role of the Duchenne data repository and Duchenne map

Siminiuc S¹, Sakellariou E, Paliouras G

¹Stichting Duchenne Data Foundation, Soest, Netherlands

670P Assessing leg vascular responsiveness to tadalafil using MRI-BOLD in boys with DMD: implications for adjuvant vasodilator therapy

Taivassalo T¹, Sullivan R, Shih R, Coppola J, Sweeney L, Forbes S

¹University Of Florida, Gainesville, United States

671P A two-year case study of cycle exercise training in a boy with Duchenne muscular dystrophy

Taivassalo T¹, Sullivan R, Bomma M, Lott D, Forbes S, Shih R, Del Toro V, Maldonado-Puebla D, Walter G, Coppola J, Sweeney L

¹University Of Florida, Gainesville, United States

672P From case to caution: hyponatremia in a patient with Duchenne muscular dystrophy on vamorolone and lessons for clinicians

Tian C^{1,2}, Nasomyont N, Ryan T, Villa C, Reebals L, Zygmunt A, Rutter M

¹Cincinnati Children's Hospital, Cincinnati, United States, ²University of Cincinnati College of Medicine, Cincinnati, United States

673P Screening of exon-skipping antisense oligonucleotide bioconjugates on Duchenne muscular dystrophy human and mouse models

Torres Masjoan L¹, Aghaeipour A, Coulson D, Frommer J, Aguti S, Privolizzi R, Zhou H, Muntoni F, TransNAT C

¹Neurodegenerative Diseases, UCL Queen Square Institute of Neurology, London, United Kingdom

674P Delpacibart zotadirsen (del-zota) increased dystrophin and improved muscle integrity markers regardless of ambulatory status in individuals with DMD44

Veerapandiyan A¹, Eskuri J, Flanigan K, Lavery C, Phan H, Smith E, Tesi Rocha C, Wong B, Morris M, Tami Y, Carmack T, Kovach P, Herzog J, Hughes S, Zhu Y, Ackermann E

¹Arkansas Children's Hospital, Little Rock, United States

675P Design of an observational study to assess real-world outcomes of patients with Duchenne muscular dystrophy in the US treated with givinostat

Veerapandiyan A¹, Finanger E, Ciafaloni E, Duong T, Proud C, Rocha C, Salinas M, Cazzaniga S, Gruppioni K, Crawford T

¹Arkansas Children's Hospital, Little Rock, Arkansas, United States

676P One-year motor function outcomes following treatment with delandistrogene moxeparvovec in young boys with Duchenne muscular dystrophy: a single center experience

Batley K¹, Gonzalez Castillo Z, Valle M, Forrest D, Brown M, Williamson S, Iannaccone S, Nelson L

¹UT Southwestern, Dallas, United States

677P Efficacy of the “fantastic four” forts with Duchenne muscular dystrophy and heart failure

Arahata H¹

¹NHO Omuta Hosp, Omuta, Japan

678VP Vastus lateralis fat fraction is associated with functional efficacy endpoints in patients with Duchenne muscular dystrophy treated with givinostat

Guglieri M¹, McDonald C, Bellanti F, Cazzaniga S, Bettica P, Schara-Schmidt U, Mercuri E, Muelas N, Vandeborne K

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679VP Efficacy of givinostat in the off-target population of EPIDYS: a subgroup analysis

Lavery C¹, Phan H, Alessi F, Cazzaniga S, Bettica P, Finkel R

¹University of California, San Diego, San Diego, California, United States

680VP Disease progression modeling in Duchenne muscular dystrophy: delayed decay in 4-stair climb with givinostat compared with standard of care

Lavery C¹, Mercuri E, Alessi F, Cazzaniga S, Bettica P, Largajolli A, Bellanti F, Péréon Y, McDonald C, Finanger E

¹University of California, San Diego, San Diego, California, United States

681VP Pharmacokinetics, initial safety and efficacy of vamorolone in 2–<4-year-old boys with Duchenne muscular dystrophy

Mah J¹, Gonorazky H, Nigro E, Pidsadny M, Lochmüller H, Aleman A, Yaworski A, Oskoui M, Sbrocchi A, Selby K, de Vera A, Dutreix C, Mathur R, Linden A, Hoffman E

¹University of Calgary, Calgary, Canada

682VP Characterizing thrombocytopenia in patients with Duchenne muscular dystrophy treated with givinostat: results from the phase 3 EPIDYS trial

McAdam L¹, Brandsema J, Finanger E, Alessi F, Cazzaniga S, Bettica P, Niks E

¹Holland Bloorview Kids Rehabilitation Hospital, Toronto, Canada

683VP ELEVATE-45-201, a phase 1/2b study to assess the safety and efficacy of ENTR-601-45 in patients with DMD amenable to exon 45 skipping

Baranello G, **Reddy D¹**, Porco D, Stadheim A, Parasrampur R, Weinstein D, Sethuraman N

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684VP Efficacy of givinostat by age (6–7 and >7 years): a post hoc analysis of EPIDYS

Vučinić D¹, Lavery C, Alessi F, Cocceani N, Bettica P, Nevo Y

¹Clinic Of Neurology and Psychiatry for Children and Youth, Belgrade, Serbia

685VP Cardiac safety data for givinostat in ambulant patients with Duchenne muscular dystrophy: results from the EPIDYS study

Willis T¹, Mercuri E, Bryne B, Niks E, Schara-Schmidt U, Le Tourneau T, Boespflug-Tanguy O, Guglieri M, Muelas N, Gómez Andrés D, Phan H, César Díaz S, Bourke J, Cazzaniga S

¹Robert Jones And Agnes Hunt Hospital, Oswestry, United Kingdom

686VP Thorough QT study on the effect of therapeutic and supratherapeutic dosing of givinostat in healthy volunteers

Willis T¹, Mercuri E, Bryne B, Niks E, Schara-Schmidt U, Le Tourneau T, Boespflug-Tanguy O, Guglieri M, Muelas N, Gómez Andrés D, Phan H, César Díaz S, Bourke J, Cazzaniga S

¹Robert Jones And Agnes Hunt Hospital, Oswestry, United Kingdom

687VP Modeling disease progression in Duchenne muscular dystrophy: reduced decline in forced vital capacity with givinostat compared with standard of care

Lavery C, **Finanger E¹**, Alessi F, Cazzaniga S, Bettica P, Largajolli A, Bellanti F, Péréon Y, McDonald C, Mercuri E

¹Shriners Hospital for Children, Portland, Oregon, United States

688VP Characterizing gastrointestinal adverse events of interest from a phase 3 study of givinostat in patients with Duchenne muscular dystrophy

De Waele L^{1,2}, Brandsema J, Alessi F, Cazzaniga S, Bettica P, Finanger E

¹Department of Child Neurology, University Hospitals Leuven, Leuven, Belgium, ²Department of Development and Regeneration, KU Leuven, Leuven, Belgium

Poster Session 2 – 118P-165P: CM – CMD

118P Molecular and clinical insights into congenital muscular dystrophies in India: a single centre experience

Dhall A¹, Jassal B, Vishnu V, Suri V, Sharma M

¹All India Institute of Medical Sciences, New Delhi, India

119P Clinical phenotypes of an adult population carrying mutations in the ryanodine receptor 1 (RYR1) gene

Mohamed K¹, Finnigan P, Godfrey R, Pizzamiglio C, Quinlivan R

¹Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology NHS Highly Specialised Service for McArdle Disease and Related Disorders, Queen Square Centre for Neuromuscular Diseases, The National Hospital for Neurology and Neurosurgery, London, United Kingdom

120P Data trends and highlights from the Russian COL-6 patients registry

Abramova D¹, Artemyeva S, Bardakov S, Kuzenkova L, Melnik E, Monakhova A, Murtazina A, Shulyakova I, Vlodavets D, Kurbatov S

¹Autonomous Non-profit Organization «Support for Patients with Ullrich-Bethlem Myopathy», Nizhny Novgorod, Russia

121P Tropomyosin 3-related myopathy presenting with scapulo-peroneal weakness and myogenic tremor in two unrelated patients

Schaefer J¹, Schoser B, Saak A

¹Uniklinikum C.G. Carus Dresden, Dresden, Germany

122P Sporadic late-onset nemaline myopathy with monoclonal gammopathy of unknown significance treated with melphalan and autologous hematopoietic stem cell transplantation

Kierdaszuk B¹, Nieporęcki K, Rosińska A, Drozd-Sokolowska J, Kacprzyk P, Machowicz R, Tomaszewska A, Jamrozik K, Basak G, Kostera-Pruszczyk A

¹Department of Neurology, ERN EURO-NMD, Medical University of Warsaw, Warsaw, Poland

123P HDAC11 deficiency regulates age-related muscle decline and sarcopenia

Nogales-Gadea G¹, Odria R, Cardús A, Gomis-Coloma C, Balanyà-Segura M, Mercado-Amarilla A, Maestre-Mora P, Poveda-Sabuco A, Domingo J, Gomez-Sanchez J, Hurtado E, Suelves M

¹Badalona Neuromuscular Research Group (GRENBA), Germans Trias i Pujol Research Institute, Badalona, Spain

124P Imaging-pathology correlation across CNM genotype

Uy G¹, Saito Y, Yoshioka W, Hayashi S, Noguchi S, Nishino I

¹National Center of Neurology and Psychiatry, Department of Neuromuscular Research, Kodaira City, Tokyo, Japan

125P CRISPR base editing for disease modeling and treatment of centronuclear myopathy

Wang H¹, Hauer V, Staecker I, Pommerenke C, Steffen A, Kallnischkies H, Kaufmann M, Fähnrich S, Steenpass L

¹Leibniz Institute DSMZ-German Collection of Microorganisms and Cell Cultures, Braunschweig, Germany

126P Flying under the radar: TTN missense variants accounting for missed titinopathy diagnoses

Di Feo M^{1,2,3}, Rees M, Lillback V, Gautel M, Jungbluth H, Fiorillo C, Bruno C, Ravenscroft G, Malfatti E, Rodolico C, Tasca G, Schnabel F, Dhall A, Balogh I, Attie-Bitach T, Bertini E, D'Amico A, Udd B, Savarese M, Titin Study Group O

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127P Protein aggregation in silico predicts phenotype severity of ANXA11 related myopathy

Pedroza Martins A¹, Hilario B, Mattos A, V. B. L. Lima I, Castelo Branco Pupe C, Rolim Muro Martinez A, Nucci A, Cavalcante França Júnior M

¹Faculty of Medical Science, Department of Neurology, State University of Campinas, UNICAMP, São Paulo, Brazil, Campinas, Brazil

128P HACD1-congenital myopathy with muscle MRI examination: four cases report

Pedroza Martins A¹, Motta Mecê A, Iwabe C, Moreschi F, Fernando Grossklaus, L, SJ Dertkigil S, Rolim Muro Martinez A, Nucci A, Cavalcante França Júnior M

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129P RYR-1 gene mutation-related neuromuscular disorders: a retrospective clinical and genetic analysis of six patients

Coşkun A¹, Topaloğlu H

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130P Role of dynamin 2 and clathrin-coated plaques in muscle mechanotransduction via YAP/TAZ regulation

Benoist M^{1,2}, Milliet K, Franck A, Moparthi S, Fongy A, Moulay G, Lacene E, Mamchaoui K, Chardonnet S, Dingli F, Loew D, Evangelista T, Coirault C, Trochet D, Bitoun M, Vassilopoulos S

¹Association Institut de Myologie, Paris, France, ²Inserm U974, Paris, France

131P Natural history of motor progression of LAMA2-RD muscular dystrophy in 27 Spanish patients

Urcuyo G, **Gómez-Andrés D¹**, Costa Comellas L, Toro Tamargo E, López López J, Álvarez Molinero M, Munell F

¹Child Neurology. H.U. Vall d'Hebron, Barcelona, Spain

132P Validation of muscle phenotype in a COG1-related congenital muscular dystrophy patient without central nervous system involvement using a zebrafish model

Severa G^{1,2}, Kroll F, De Vita F, Bastu S, Das B, Suarez B, Jofre J, Del Bene F, Castiglioni C, Malfatti E

¹University Paris Est Créteil, Inserm, U955, IMRB, Créteil, France, ²Reference Center for Neuromuscular Disorders, APHP Henri Mondor University Hospital, Créteil, France

133P Exploring novel modifier genes for LAMA2-RD: in vitro and in vivo studies

Pini V^{1,2}, Bonaccorso R, Porrello E, Morgan J, Muntoni F, Previtali S,

¹Neuromuscular Repair Unit, Institute of Experimental Neurology, Ospedale San Raffaele, Milan, Italy, ²Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom

134P Characterization of a novel deep intronic COL6A1 c.930+176C>T splice activation variant causing COL6-related dystrophy

Donkervoort S¹, Silverstein S, Orbach R, Ganesh V, Mohassel P, Chao K, Hu Y, Pais L, Foley A, Richardson R, Bolduc V, J okela M, Bönnemann C

¹Neuromuscular and Neurogenetic Disorders of Childhood Section, NINDS, NIH, Bethesda, United States

135P Distinct muscle MRI patterns in Japanese patients with NEB-related myopathy

Ogasawara M^{1,2}, Yae Y, Nishimori Y, Eura N, Yoshioka W, Yamanaka A, Hashizume L, Miyazaki N, Sugie K, Hayashi S, Noguchi S, Iida A, Nishino I

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136P Alpha-dystroglycan expression analysis in urine-derived cells from Fukuyama congenital muscular dystrophy patients for clinical trials

Takahashi A¹, Kunitake, Fujino G, Kobayashi K, Kanagawanagawa M, Satake W, Aoki Y, Toda T

¹The University Of Department of Neurology, Graduate School of Medicine, The University of Tokyo, Tokyo, Japan

137P Expanding the phenotypic spectrum of MYBPC1-associated congenital myopathy with tremor: report of the first Italian case

Croce M¹, Rimoldi M, Del Bo R, Ciscato P, Cosentino G, Comi G, Corti S, Ronchi D, Ravaglia S, Velardo D

¹Department of Brain and Behavioural Sciences, University of Pavia, Pavia, Italy

138P Muscle imaging of the patients with RYR1-related myopathies and its significance to clinical features

Shimazaki R^{1,2}, Noguchi S, Yoshioka W, Saito Y, Hayashi S, Nishino I

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139P Inputs and pitfalls of muscle imaging, biopsy, and dermal fibroblast culture in diagnosing COL6-related dystrophies: insights from 217 suspected cases

Lessard L^{1,2,3,4}, Streichenberger N, Michel L, Menassa R, Bani-Sadr A, Rouvet I, Manel V, Svahn J, Moussy M, Fenouil T, Bouhour F, Pégat A

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140P Neuromuscular symptoms of ORAI1-related immunodeficiency

Czeczko K¹, Jędrzejowska M, Potulska-Chromik A, Franaszczyk M, Wolska-Kuśnierz B, Rosiak E, Aragon-Gawińska K, Stokłosa T, Kostera-Pruszczyk A

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141P A multicentre, prospective, longitudinal and observational natural history study for patients with nemaline myopathy in the United Kingdom with novel international collaboration

Fisher G¹, Lilien C, Sarkozy A, Baranello G, James M, Jungbluth H, Yao A, Goel N, Zhao R, Hill S, Woods M, Duong T, Ismail S, Tesi Rocha C, Servais L, Site Study Teams S, Data federation Steering committee S

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142P Phenotypical and genotypical characterization of a cohort of 196 COL6-RD patients in Europe and Chile

Bisciglia M¹, Stojkovic T, Metay C, Allamand V, Natera-de Benito D, Nascimento A, Claeys K, Castiglioni C, Suarez B, Vissing J, De Paepe B, De Bleecker J, Lieveke A, Butterfield R, Deconinck N

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143P Generation and characterization of Col6a1 knock-in mice: a promising pre-clinical model for collagen VI-related dystrophies

Jimenez-Mallebrera C¹, López-Márquez A, Badosa C, Sánchez-Martín M, Cadot B, Guesmia Z, Georvasilis I, Balsells S, Quintana A, Roldán M, Allamand V, Jimenez-Mallebrera C

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144P XLMTM female carriers: a review of patient reported data collected by the MTM & CNM patient registry

Bohill J¹, Hickson L, Marini-Bettolo C

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145P Lycra foundation suit intervention in patients with RYR1-related congenital myopathy: a case study of functional improvement

Longatto J¹, Main M, Muntoni F

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146P Clinical, molecular, and pathological characterization of MYL1-related congenital myopathy: report of two novel cases

Natera-de Benito D¹, Madrigal I, Villar-Vera C, Arca G, Expósito-Escudero J, Rodríguez-Revenga L, Piolatti-Luna A, Muelas N, Vilchez R, Ciudad Celdran M, Codina A, Estévez-Arias B, Carrera-García L, Ortez C, Rodríguez-Carunchio L, Sebastiani G, Azorin I, Nascimento A, Jou C, Vilchez J

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147P Clinical and functional outcome measures in LAMA2-related muscular dystrophy and SELENON-related myopathies; a 1.5-year natural history study

de Laat E¹, Groothuis J, Bouman K, Houwen - van Opstal S, Erasmus C, Voermans N

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148P A case of congenital nemaline myopathy with heart failure

Saito M¹, Shimonaga T, Kimura K, Kodama M, Sumimoto Y, Masada K, Kinoshita H, Sugino H, Kurashige T

¹NHO Kure Medical Center and Chugoku Cancer Center, Kure, Japan

149P A Kbtbd13 knock-out mouse model for nemaline myopathy type 6 sheds light on the role of KBTBD13 and reveals potential new treatment strategies

Vonk L¹, Baelde R, Galli R, Nieuwendijk M, de Winter J, Ottenheijm C

¹Amsterdam Umc, Amsterdam, Netherlands

150P Longitudinal respiratory function in early-onset TTN-related congenital myopathies: insights from a natural history study

Cicala G^{1,2}, Vanegas M, Bilby J, Flynn A, Wolfe A, Sheenan J, Schofield A, Joefield T, Heraghty J, Ridout D, Muntoni F, Baranello G, Jungbluth H, Sarkozy A

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151P Rat model of X-linked myotubular myopathy with pathological signs in skeletal muscles and liver

Navas Navarro P^{1,2}, Renaud-Gabardos E, Salman B, Mangin L, Benyamine H, Corre G, Goddard M, Tanniou G, Guerchet N, Krakau P, Martin S, Lawlor M, Concordet J, Anegón I, Buj-Bello A

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152P From data scarcity to diagnostic accuracy: artificial intelligence approaches in collagen VI muscular dystrophy

Frias M^{1,2}, Badosa C, Jiménez-Mallebrera C, Porta J, Roldán M

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153P Landscape of late-onset myopathies: a national study

Salort-Campana E^{1,2}, Dumas R, Filnemus S, Jannot A, Attarian S

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154P Nicotinamide riboside supplementation prevents the onset of mitochondrial dysfunction in a mouse model for nemaline myopathy type 6

Baelde R¹, Fortes Monteiro A, Nollet E, Galli R, Bastu S, Malfatti E, Schomakers B, Vinten K, Houtkooper R, van der Velden J, Ottenheijm C, de Winter J

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155P Distal axonal neuropathy as the initial presentation in COL6A-related myopathies: a case series

Eisenkölbl A^{1,2}, Marshall A, Del Gobbo G, Boycott K, Hamilton L, Lochmüller H, Yaworski A, McMillan H

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156P Congenital myopathies- incidence and prevalence in a population-based cohort in western Sweden

Michael E^{1,2}, Hedberg-Oldfors C, Oldfors A, Darin N

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157P Cell painting for the study of SELENON-congenital myopathy cell models

Barraza-flores P¹, Dasgupta S, Lee W, Cimini B, Beggs A

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158P Molecular pathogenesis of clubfoot: TPM2 mutation alters muscle development and organization

Ramanujam A¹, Hordyjewska-Kowalczyk E, Tylzanowski P

¹Medical University of Lublin, Dept of Biomedical Sciences, Laboratory of Molecular Genetics, Lublin, Poland

159P Defining pathogenetic mechanisms in CASQ1 disorders through single-fibre studies

Laitila J^{1,2}, Rostedt F, Laarne M, Jokela M, Sarparanta J, Zhao F, Udd B, Johari M, Huovinen S, Reimann J, Kornblum C, Hackman P, Lehtokari V, Pelin K

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160P Developing KLHL40 gene replacement therapy for congenital myopathy in a preclinical mouse model

Coleman S, McNulty J, **Gupta V**¹

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161P PYROXD1 myopathy update: from phenotypic expansion to preclinical therapy development

Yuen M^{1,2,3}, Dziaduch G, Yasa J, Kiriaev L, Houweling P, Kettle E, Pang I, Graham M, Lemckert F, Cooper S, Evesson F

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162P Interaction studies of the titin A150 domain in C2C12 myotubes

Vainio A¹, Sarparanta J, Jonson P, Udd B

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163P Transcriptomic profiling reveals shared pathogenic pathways and novel biomarkers across centronuclear myopathies

De Feraudy Y^{1,2}, Swain S, Simon A, Bouman K, Voermans N, Vasseur S, Romero N, Nishino I, Saito Y, Lawlor M, Streichenberger N, Jungbluth H, Bertini E, D'Amico A, Oldfors A, Malfatti E, Evangelista T, Böhm J, Biancalana V, Laporte J

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164P Phenotypic spectrum and disease trajectories in paediatric patients with dystroglycanopathy: a 22-year single-centre retrospective cohort study

Lochmüller A¹, Manzur A, Munot P, Robb S, Scoto M, Baranello G, Tedesco F, Simmonds J, Laurence C, McCauley J, Muntoni F, Sarkozy A

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165P Antisense oligonucleotide NS-035 targeting pathogenic exon-trapping in Fukuyama muscular dystrophy: a phase 1 trial

Fujino G¹, Kitamura A, Takahashi A, Maeda M, Kubota A, Tokuyama Y, Wada I, Kobayashi K, Komaki H, Taniguchi-Ikeda M, Ishigaki K, Toda T

¹Department of Neurology, Graduate School of Medicine, The University of Tokyo, Tokyo, Japan

Poster Session 2 – 193P-217P: FSHD

193P Genotype-phenotype correlation in a cohort of suspected FSHD patients from Serbia

Albano N¹, Ralic B, Nuredini A, Cuoghi Costantini R, Peric S, Tupler R

¹Department of Biomedical, Metabolism and Neural Sciences, University of Modena and Reggio Emilia, Modena, Italy

194P A human skeletal muscle-on-chip model for facioscapulohumeral muscular dystrophy

Augustinus R¹, Franken M, Filonova G, van der Maarel S, Pijnappel P, de Greef J

¹Leiden University Medical Center, Leiden, Netherlands

195P Identification of genetic modifiers of disease severity and methylation in large multigenerational kindred with FSHD

Butterfield R¹, Dunn D, Duval B, Moldt S, Weiss R

¹University of Utah, Department of Pediatrics, Salt Lake City, United States

196P Focus on fatigue: the presence of performance fatigability in childhood onset facioscapulohumeral dystrophy

Dijkstra J¹, Van der Woude D, Van Haaren R, Brenninkmeijer R, Voermans N, Van der Pol L, Bartels B, Erasmus C

¹Radboud University Medical Center, Nijmegen, Netherlands

197P The landscape of FSHD data collection: a 2025 expansion to the TREAT-NMD FSHD dataset

Garrow M¹, Poll A, Ali F, Guglieri M, Ambrosini A, Rodrigues M

¹TREAT-NMD, Newcastle Upon Tyne, United Kingdom

198P The UK facioscapulohumeral muscular dystrophy patient registry: a powerful tool to support clinical research and patient voice in the translational research pathway

Carver A¹, Hickson L, Walker H, Marini-Bettolo C, Muni-Lofra R, Graham A, Norwood F, Roberts M, Willis T, Matthews E, McQueen-Mencias M, Tasca G, Langlands G, McMacken G, Adcock K

¹The John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University, and Newcastle upon Tyne Hospitals NHS Foundation Trust, Newcastle upon-Tyne, UK

199P Natural history of muscle volume and muscle fat content biomarkers in FSHD based on whole-body fat-referenced MRI

Karlsson M¹, Widholm P, Puma A, Villa L, Cavali M, Pini J, Ezaru A, Marty B, Evangelista T, Thomas R, Danjoux L, Sacconi S

¹AMRA Medical AB, Linköping, Sweden

200P Quantitative whole-body MRI biomarker relation to muscle strength & function in FSHD patients

Foltz M, Widholm P, **Karlsson M¹**, Pini J, Puma A, Villa L, Cavali M, Ezaru A, Bassez G, Marty B, Evangelista T, Thomas R, Danjoux L, Tard C, Sacconi S

¹AMRA Medical AB, Linköping, Sweden

201P Managing spinal deformities in facioscapulohumeral muscular dystrophy: a scoping review of surgical management

Pelsma M¹, Voermans N, Peters Q, Hoogveld L, de Valle K, Woodcock I, Lark R, Tsirikos A, de Kleuver M, Erasmus C, Houwen S

¹Radboud University Medical Center, Nijmegen, Netherlands

202P Disease burden in Serbian patients with facioscapulohumeral muscular dystrophy

Ralic B¹, Albano N, Viric V, Nuredini A, Azanjac A, Rajić S, Marjanović A, Tupler R, Heatwole C, Perić S

¹Clinical Hospital Center Zvezdara, University of Belgrade, Belgrade, Serbia

203P Co-occurrence of anti-AChR myasthenia gravis in facioscapulohumeral dystrophy patients: a case series

Sacconi S^{1,2}, Tammam G, Villa L, Lemmers R, Pini J, Salvati L, Cavalli M, Ezaru A, Puma A, van der Maarel S

¹Peripheral Nervous System & Muscle Department, Pasteur 2 Hospital, Nice University Hospital, Nice, France, ²Institute for Research on Cancer and Aging of Nice, CNRS, INSERM, Côte d'Azur University, Nice, France

204P Modeling DUX4 activation in FSHD and revealing the mechanism by functional genomics approach

Sasaki-Honda M^{1,2}, Rada-Iglesias A, Sakurai H

¹CiRA, Kyoto University, Kyoto, Japan, ²IBBTEC, University of Cantabria/CSIC, Santander, Spain

205P Disease progression in facioscapulohumeral muscular dystrophy in a clinical trial timeframe

Teeselink S¹, Vincenten S, Voermans N, van Alfen N, van Engelen B, Mul K

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206P Exploring contractile performance with ultrasound in FSHD: preliminary results of the MUSCLE+ study

Van Iersel O¹, Weekenstroom H, Cameron D, Doorduyn J, van Alfen N

¹Radboud University Medical Center, Clinical Neuromuscular Imaging Group, Donders Institute for Brain, Cognition and Behavior, Radboud University Medical Center, Nijmegen, Netherlands

207P The Italian cohort of facio-scapular-humeral dystrophy type 1: a long-term follow-up up to 20 years

Vercelli L¹, Gadaleta G, Ruggiero L, Ricci G, Previtali S, Angelini C, Berardinelli A, Bucci E, D'Angelo M, Di Muzio A, Fiorillo C, Grandis M, Maggi L, Monforte M, Petrucci A, Rodolico C, Valentino M, Verriello L, Mongini T, Italian Clinical FSHD Group

¹Department of Neurosciences "Rita Levi Montalcini", University of Turin, Turin, Italy

208P The FSHD European trial network

Voermans N¹, Hawkins S, Lemmers R, Giardina E, Bugardini E, Carraro E, Dumonceaux J, Péréon Y, Tasca G, Monforte M, Erasmus C, Willis T, de Haas R

¹Radboud University Medical Centre, Nijmegen, Netherlands

209P An updated international standard of care for facioscapulohumeral muscular dystrophy

Voermans N¹, Kinoshita J, Evangelista T, Badiani R, Honda M, Statland J

¹Radboud University Medical Centre, Nijmegen, Netherlands

210P Expanding the UK FSHD patient registry dataset - improving data collection and amplifying 'patient voice'

Walker H¹, Hickson L, Muni-Lofra R, Matthews E, Badiani R, Graham A, Marini-Bettolo C

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211P Associations between muscle strength and MRI biomarkers in FSHD: toward imaging-based functional classification

Widholm P, Foltz M, **Karlsson M¹**, Pini J, Puma A, Villa L, Cavali M, Ezaru A, Bassez G, Marty B, Evangelista T, Thomas R, Danjoux L, Tard C, Sacconi S

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212P Facioscapulohumeral muscular dystrophy type 2 in Chinese cohort: findings from a large-scale 4qA D4Z4 methylation screening

Xia X¹, Liu L, Lu J, Zhao C, Zhu W

¹Huashan Hospital, Fudan University, Shanghai, China

213P The China FSHD patient registry: a multicenter survey from the Pan-Yangtze delta neuromuscular alliance (PYDNA)

Xia X¹, Liu L, Yue D, Wu G, Ke Q, Zhao C, Zhu W

¹Department of Neurology, Huashan Hospital, Fudan University, Shanghai, China

214P Multi-scale machine learning model predicts muscle and functional disease progression in FSHD

Blemker S^{1,2}, Weatherly E, Riem L, DuCharme O, Leung D, Friedman S

¹Springbok Analytics, Charlottesville, VA, United States, ²University of Virginia, Charlottesville, VA, United States

215P Video-based biomechanical analysis captures disease-specific movement signatures of myotonic dystrophy and facioscapulohumeral muscular dystrophy

De Monts De Savasse C¹, Ruth P, Ulrich S, Vogt-Domke S, Ismail S, Karman L, Falisse A, Muccini J, Covitz S, Day J, Delp S, Duong T

¹Stanford University, Department of Neurology, Palo Alto, United States

216P Genetic ablation of Cdkn1a ameliorates facioscapulohumeral muscular dystrophy in mice overexpressing FRG1

Fantini S¹, Bisceglia G, Zyla J, Moio N, Nuredini A, D'Antona G, Vattermi G, Malfatti E, Polanska J, Tupler R

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217P Direct and indirect socio-economic burden of facioscapulohumeral muscular dystrophy (FSHD): a national, cross-sectional study

Lessard L^{1,2,3,4}, Grant A, Smith I, Osman H, Lochmüller H, McMillan H, Pfeffer G, Korngut L, Gagnon C, Selby K, Breiner A, Thavorn K, Warman-Chardon J

¹The Ottawa Hospital, Uottawa, Ottawa, Canada, ²Faculty of Medicine, University of Ottawa, Ottawa, Canada, ³Hôpital Neurologique de Lyon, Hospices Civils de Lyon, Lyon, France, ⁴PGNM, Institut Neuromyogène, CNRS UMR 5261 - INSERM U1315, Lyon, France

Poster Session 2 – 314P-329P: Multidisciplinary management of neuromuscular diseases

314P Personalized training for people with rare neuromuscular disorders (PETRA-NMD): a multi-center randomized controlled clinical trial

Arora T¹, Fossmo H, Raastad T, Fjermestad K, Ramberg C, Nilsen K, Elgstøen K, Arntzen K, Devik K, Morsund Å, Varhaug K, Ørstavik K

¹Oslo University Hospital, Oslo, Norway

315P Living statue syndrome: a rare case of fibrodysplasia ossificans progressiva managed through palliative rehabilitation

Bhatia A¹, Khanna M, Jaison V, Mahey P

¹Neuromuscular Center of Excellence Department of Neurology Christian Medical College and Hospital, Ludhiana, India

316P Perceived behavioral difficulties in neuromuscular disorders: a comparison between caregivers and pediatric patients

Buchignani B^{1,2}, Cristofani P, Marinella G, Schifino P, Chicca L, Sesso G, Milone A, Astrea G, Battini R

¹Irccs Stella Maris Foundation, Pisa, Italy, ²University of Pisa, Pisa, Italy

317P POM-005: a global, prospective, observational registry of people living with Pompe disease

Clarke S¹, Crittenden B, Graham R, O'Brien-Prince K, McIntosh P

¹Amicus Therapeutics Ltd, Marlow, United Kingdom

318P How to handle TTN variants in the genomic era: analysis on 18,462 Solve-RD cases

Di Feo M¹, Paramonov I, Matalonga Borrel L, Töpf A, Hoischen A, Beltran S, Vissers L, Natraj Gayathri S, Ellwanger K, Cossee M, Perrin A, Sarkozy A, Bonne G, Verdonchot J, Demidov G, Laurie S, Johari M, Udd B, Savarese M, Solve-RD consortium O

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319P Sexual health impairments in neuromuscular diseases: a scoping review

El Kaim A¹, Banos M, Birnbaum S, Hogrel J, Gargiulo M

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320P Multidisciplinary rehabilitation for neuromuscular diseases: a specialized rehabilitation approach

Fossmo H^{1,2,3}, Råen M, Granberg A

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321P “It doesn’t get more normal than that!” – challenges with sexuality experienced by people with neuromuscular diseases and their needs for rehabilitation knowledge and guidance on sexual life

Handberg C¹, Kajhøj Møller J, Hansen K

¹Aarhus University, Aarhus, Denmark

322P The voice of the child: insights from South African children living with neuromuscular disorders during inspiratory muscle training intervention

Human A^{1,2,3}, Miller A, Morrow B

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323P Red flags in the management of SELENON - related myopathy patients

Kwiatos K¹, Potulska-Chromik A, Czeżko K, Kostera-Pruszyk A

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324P Acceptability, feasibility, safety and potential efficacy of an optimised rehabilitation for treated patients with SMA in United Kingdom: ACE SMA

Lilien C¹, Hill S, Mavrommati F, Ramjattan H, Taylor F, Collet J, Servais L

¹STRONG, MDUK Oxford Neuromuscular Centre, Department of Paediatrics, University of Oxford, United Kingdom

325P A standardised clinical approach for foot orthoses design in Charcot-Marie-Tooth

Martel M^{1,2}, Fortin C, Rivet N, Martineau-Beaulieu É, Noël-René É, Plourde C, Ballaz L

¹Chu Sainte-justine, Montreal, Canada, ²Université de Montréal, Montreal, Canada

326P Neuromuscular ultrasound for investigating changes in muscle structure in the multicenter randomized controlled clinical trial PETRA-NMD

Ramberg C¹, Hæstad C, Dybesland Rosenberger A, Nyborg Stefansson V, Arntzen K, Arora T, Ludt Fossmo H, Ørstavik K

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327P ERN Euro-NMD multidisciplinary management and care group: advancing multidisciplinary care for neuromuscular disorders in Europe

Rosenberger A¹, Ørstavik K, Fossmo H, Nordstrøm M

¹National Neuromuscular Centre Norway, University hospital of North-Norway, Tromsø, Norway

328P Evaluating patient and family satisfaction with multidisciplinary care in a national paediatric neuromuscular disease unit

Weichbrodt J¹, Lindqvist J, Svensson J, Gudmundsson M, Ekström A

¹Queen Silvias Children's Hospital, Gothenburg, Sweden

329P The traffic light system for highlighting patients with neuromuscular disease, is it capturing enough symptomatic patients?

Willis T^{1,2}, Roberts M, Bassie C, Kulshrestha R, Willis D

¹Robert Jones and Agnes Hunt Hospital, Oswestry, United Kingdom, ²Chester University Medical School, Chester, United Kingdom

Poster Session 2 – 431P-456P, 457VP-458VP: Genetics of NMD (new genes and NGS, diagnostic etc.)

431P Pediatric genetic neuromuscular disease in Jordan; high diagnostic yield and emerging phenotypes via next generation sequencing

Aburahma S¹, Pagliarini S

¹Jordan University of Science and Technology, Irbid, Jordan

432P Severe neonatal-onset PYROXD1-related myopathy with a novel homozygous missense variant: expanding the phenotypic spectrum

Aleisa Z¹, Yoon G, Evesson F, Cooper, Gonorazky H

¹Division of Neurology, Department of Pediatrics, Hospital for Sick Children, University of Toronto, Canada

433P Phenotypic delineation of NAA10-related neurodevelopmental disorder due to Leu126Arg or Phe128Leu missense variants

Bühler A¹, Kustermann M, Aigner-Radakovics K, Diofano F, Marchi E, Bittner R, Bernard E, Harpell R, Just S, Lyon G, Schmidt W

¹Molecular Cardiology, Department of Internal Medicine II, University of Ulm, Germany

434P Compound heterozygous COA7 mutations in two siblings with mitochondrial neuropathy: a novel variant and deep clinical phenotyping

Cicala G^{1,2}, Rollieri E, Luigetti M, Fattori F, Bosco L, Primiano G, Rizza T, Bertini E, Mercuri E

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435P Replication factor C-4 deficiency: a case report

Coşkun A¹, Yüksel D, Topaloğlu H

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436P Phenotypic heterogeneity of GGPS1-associated myopathy: description of two novel north African families

Farhat E¹, Lakhdhar I, Chaabouni M

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437P Clarifying the significance of the CLCN1 c.2284+5C>T and c.920T>C variants

Lehtinen S^{1,2}, Jonson P, Jokela M, Palmio J

¹Neuromuscular Research Center, Tampere University and Tampere University Hospital, Tampere, Finland, ²Fimlab Laboratories Ltd., Tampere, Finland

438P Optimization of RNA-sequencing in rare neuromuscular diseases: a cohort study of over 300 cases

Lillback V¹, Natraj Gayathri S, Di Feo M, Mueller J, Sarkozy A, Muntoni F, Jonson P, Lehtokari V, Oghabian A, Hackman P, Udd B, Savarese M

¹Folkhalsan Research Center, Helsinki, Finland

439P A novel lysosomal vacuolar myopathy associated with neuronal ceroid lipofuscinosis and biallelic variants in CLN8

Lindgren U^{1,2}, Hedberg-Oldfors C, Nordström S, Goebel H, Oldfors A

¹University of Gothenburg, Gothenburg, Sweden, ²Sahlgrenska University Hospital, Gothenburg, Sweden

440P Neuromuscular phenotype of three patients with biallelic variants in the RFC4 gene

Mueller J¹, Sarkozy A, Phadke R, Rabinowicz S, Manzur A, Dixit A, Scott I, Stenton S, Chambers D, Lochmuller H, Muntoni F

¹Dubowitz Neuromuscular Centre, London, United Kingdom

441P Don't miss the giants: rRNA depletion uncovers long disease genes that polyA overlooks

Natraj Gayathri S¹, Udd B, Hackman P, Oghabian A, Savarese M

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442P Splicing regulation of muscle function-associated genes in skeletal muscle tissues

Oghabian A¹, Sian V, Jonson P, Udd B, Hackman P, Gomez Andres D, Munell F, Camacho Soriano J, Sanchez Duran M, Savarese M

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443P Acute respiratory failure in pregnancy: the presenting symptom of a disorder of gliomedin, an essential component of the nodes of Ranvier

Patel R¹, Donkervoort S, Quinn C, Baer M, Rey M, Mora J, Foley A, Bönnemann C

¹Neuromuscular and Neurogenetic Disorders of Childhood Section, National Institute of Neurological Disorders and Stroke, National Institutes of Health, Bethesda, Maryland, United States

444P SIN3A-myopathy in the context of Witteveen-Kolk syndrome

Pedroza Martins A¹, Vieira Bolzani Lopes Lima I, Scardua Silva L, Koutsodontis M, Alvim M, Rolim Muro Martinez A, Cardoso Bonadia L, Iwabe C, Lin Yasuda C, Cendes F, Lopes Cendes I, Nucci A, Cavalcante França Junior M

¹Faculty of Medical Science, Department of Neurology, State University of Campinas, UNICAMP, São Paulo, Brazil, Campinas, Brazil

445P PLXND1: further evidence for the role of PLXND1 in Poland-Möbius spectrum disorder

Põlluaas L^{1,2}, Oja K, Õunap K

¹Institute of Clinical Medicine, University of Tartu, Tartu, Estonia, ²Genetics and Personalized Medicine Clinic, Tartu University Hospital, Tartu, Estonia

446P Structural variants (SVs) underlying neurogenetic and neuromuscular diseases

Ravenscroft G¹, Scriba C, Folland C, Bryen S, Weisburd B, Monahan G, Rick A, Rodrigues M, Corbett M, Gecz J, Davis M, Ashton C, Coman D, Yau W, Roxburgh R, Lamont P, Laing N

¹The Harry Perkins Institute of Medical Research, Perth, Australia

447P A novel hemizygous CD99L2 variant in adult patient with neurogenic muscle impairment

Sarv S¹, Puusepp S, Väli L, Ganesh V, Stenzel W, Goebel H, Udd B, Kahre T, Õunap K

¹Genetic and Personalized Medicine Clinic, Institute of Clinical Medicine, Faculty of Medicine, University of Tartu, Tartu, Estonia

448P Congenital core myopathy linked to SOX5: Expanding the phenotypical spectrum of Lamb-Shaffer syndrome

Städler K^{1,2}, Gerasimenko A, Nava C, Heron D, Schaerer E, Gitiaux C, Authier F, Stojkovic T, Malfatti E, Villar-Quiles R

¹Neuromuscular reference center Nord/Est/Ile de France, Myology Institute, Sorbonne University, Pitié-Salpêtrière Hospital, Assistance Publique des Hôpitaux de Paris, Paris, France, ²Department of Neurology, Neurocenter of Southern Switzerland, Ente Ospedaliero Cantonale, Lugano, Switzerland

449P Copy-number variant and mobile element insertion analysis of whole-exome sequencing data from patients with neuromuscular disorders

Turku T^{1,2}, Savarese M, Johari M, Kiiski K, Rusanen S, Wallgren-Pettersson C, Pelin K, Udd B, Hackman P

¹Folkhälsan Research Center, Helsinki, Finland, ²Department of Medical Genetics, Medicum, University of Helsinki, Helsinki, Finland

450P Symptomatic variability among three siblings affected by a novel PRX variant resulting in PRX-related peripheral neuropathy (Charcot-Marie-Tooth type 4F)

Wade C^{1,2}, Zygmunt A, Vilasaktipakorn P, Nagaraj C, Mazur J, Turnwald A, Tian C

¹Cincinnati Children's Hospital Medical Center, Cincinnati, United States, ²University of Cincinnati College of Medicine, Cincinnati, United States

451P Genetic and clinical spectrum of PIEZO2-related disorders: insights from a multicenter study of 26 patients

Akinci G¹, Ozyilmaz B, Ozturk G, Komur M, Onel E, Ardicli D, Gerik Celebi H, Ozcelik A, Yilmaz S, Dokurel Cetin I, Gunay C, Oz Tuncer G, Aydin H, Sakarya Gunes A, Yayici Koken O, Polat I, Degerliyurt A, Celik T, Topaloglu H, Turkish PIEZO2 working group

¹UHS Turkey, Izmir University, Dr. Behcet Uz Children's Hospital, Pediatric Neurology, Izmir, Turkey

452P A dominant mutation in the glycogen synthase gene results in a glycogenosis in humans

Wojtaszewski J, Hingst J, Witting N, Stemmerik M, Slipsager A, Ankjær P, Dunø M, Töpf A, Straub V, **Krag T¹**

¹Copenhagen Neuromuscular Center, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark

453P ASCC3-related congenital myopathy is linked to a defect in ribosome-associated quality control

Onnée M¹, Johari M, Roos A, Nicolau S, Ravenscroft G, Malfatti E, ASCC3 Myopathy Working Group

¹Institut Mondor de Recherche Biomédicale, Université Paris Est Créteil, INSERM U955, Créteil, France

454P A case series of hnRNP-K associated myopathy

Schiava M¹, Mannion J, Castiglioni C, Jofre J, Suarez B, Roos A, Kölbel H, Witting N, Muelas Gomez N, Martí P, Vilchez J, Sarkozy A, Rabinowicz S, Martínez-Esteban P, La Serna-Infantes J, Sotelo-Muñoz M, Straub V, Gonzalez Chamorro A, Töpf A, Díaz-Manera J

¹John Walton Muscular Dystrophy Research Centre, Newcastle Upon Tyne, United Kingdom

455P Cracking the code: genotype-phenotype correlation models in sarcoglycanopathies

Luce L¹, Kocak G, Alonso-Pérez J, Straub V, Töpf A, Díaz-Manera J, on behalf of Sarcoglycanopathy group

¹John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle Upon Tyne, United Kingdom

456P Defining the CIAO1-related disease spectrum: clinical and molecular insights into a novel defect in cytoplasmic iron-sulfur cluster assembly

Orbach R¹, Maroofian R, Stettner G, Herwig J, Pegoraro E, Manel V, Ribault S, Bouhour F, Töpf A, Munot P, Rouault T, Houlden H, Vuillerot C, Streichenberger N, Donkervoort S, Straub V, Muntoni F, Maio N, Bönnemann C, CIAO1 Study Team

¹Neuromuscular and Neurogenetic Disorders of Childhood Section, NINDS, NIH, Bethesda, United States

457VP Clinical application and comparative effectiveness of targeted gene panel sequencing and whole exome sequencing in patients with suspected adult hereditary myopathy

Lee J¹, Choi Y

¹Seoul St Mary's Hospital, College of Medicine, The Catholic University of Korea, Seoul, South Korea

458VP Co-occurrence of SEPN1-related rigid spine myopathy and Escobar syndrome in a consanguineous Tunisian patient

Belhassen I, Rodriguez Cruz P, Sakka S, **Hakim F^{1,2}**, Damak M, Mhiri C

¹Department Of Adult Neurology, Habib Bourguiba Hospital, Sfax, Tunisia, ²Laboratory of Neurogenetics, Parkinson's Disease and Cerebrovascular Disease (LR-12-SP-19), Habib Bourguiba University Hospital, University of Sfax, Tunisia

Poster Session 2 – 486P-502P, 503VP-504VP: LGMD

486P Dysferlinopathy: cardiac MRI for early detection of subclinical cardiac dysfunction

Atchayaram N¹, Thomas A, Baskar D, Vengalil S, Nashi S, Bartur A

¹National Institute of Mental Health and Neuro Sciences, Bengaluru, India

487P Identifying prognostic biomarkers in a large cohort of patients with LGMDR2-dysferlin related

Bolaño Díaz C¹, Verdu Díaz J, James M, Hilsden H, Consortium J, Straub V, Díaz Manera J

¹John Walton Muscular Dystrophy Research Centre - Newcastle University, Newcastle upon Tyne, United Kingdom

488P Role of a secreted metalloprotease in LGMDR2 pathophysiology

Bouragba D^{1,2}, Bensalah M, Ohana J, Negroni E, Altin N, Kondili M, Butler-Browne G, Trollet C, Mouly V, Bigot A

¹Association Institut de Myologie, Paris, France, ²Sorbonne Université, INSERM, Institut de Myologie, Centre de Recherche en Myologie, Paris, France

490P A recurrent non-coding 3'UTR variant as a hidden second hit in 11 families with CAPN3-related limb girdle muscular dystrophy

Estévez-Arias B^{1,2}, Segarra-Casas A, Romo L, Polavarapu K, O'Heir E, Singer-Berk M, O'Leary M, Nectoux J, Spinazzi M, Leturcq F, Llansó L, Domínguez C, Topf A, Lochmüller H, Malfatti E, Nascimento A, 3'UTR CAPN3 Study Group, Gallardo E, González-Quereda L, Natera-de Benito D

¹Neuromuscular Unit, Department of Neurology, Hospital Sant Joan de Déu, Barcelona, Spain, ²Laboratory of Neurogenetics and Molecular Medicine, Center for Genomic Sciences in Medicine, Institut de Recerca Sant Joan de Déu, Barcelona, Spain

491P HMGCR-related limb-girdle muscular dystrophy in Spain: new cases of a potentially treatable disease

Gatnau Civardi C¹, Estévez Arias B, Martín Jiménez P, Orteiz C, Carrera-García L, Expósito-Escudero J, Codina A, Jou C, Hernández Laín A, Oyarzábal A, Nascimento A, Camacho A, Domínguez González C, Natera de Benito D

¹Hospital Sant Joan de Déu, Barcelona, Spain

492P Characterization of a mouse model for plectin-related limb-girdle muscular dystrophy

Getmanchuk-Zaporoshchenko V¹, Sarnova L, Korelova K, Novotna P, Benada O, Schröder R, Gregor M

¹Laboratory of Integrative Biology, Institute of Molecular Genetics of the Czech Academy of Sciences, Prague, Czech Republic

493P Understanding pain and fatigue in dysferlinopathy from the multicenter JAIN clinical outcome study

Gordish-Dressman H^{1,2}, James M, Rufibach L, Hilsden H, Robinson E, Straub V, COS consortium on behalf of the Jain Foundation

¹Children's National Hospital, Washington DC, United States, ²The George Washington University, Washington DC, United States

494P The Egyptian neuromuscular registry: a comprehensive platform for research, and clinical trial readiness for limb girdle muscular dystrophy

Hamed D^{1,2}, El Sherif R

¹New Giza University, Giza, Egypt, ²Myo-Care National Foundation, Cairo, Egypt

495P Precision diagnosis of myopathies through clinical exome sequencing

Yildirim A, Arslan D, Güleray Lafci N, Erdem-Ozdamar S, Tan E, **Kurt C¹**

¹Hacettepe University Faculty of Medicine, Ankara, Turkey

496P CALNATHIS: a natural history study for limb girdle muscular dystrophy R1-calpain 3 related

Malfatti E^{1,2}, Severa G, Alimi C, Stojkovic T, Tard C, Noury J, Ikheteah F, Degove S, Richard I, Olivier S

¹University Paris Est, Créteil, France, ²APHP, Mondor University Hospital, Créteil, France

497P Late-onset SELENON-related myopathy

Nath S¹, Liewluck T, Madigan N, Milone M

¹Mayo Clinic, Rochester, United States

498P Clinical and genomic profile of dysferlinopathy from Indonesian population

Indrawati L, **Shafitha N**¹, Isaac W, Putri M, Simatupang S, Safri A, Fadli N, Harsono A, Wiratman W, Budikayanti A, Octaviana F, Hakim M,

¹Department of Neurology, Dr. Cipto Mangunkusumo Hospital, Jakarta, Indonesia

499P Training of physiotherapists in clinical and functional outcome assessment in patients with LGMDR2

Sneha S¹, Bhasin A, James M, Hilsden H, Rani N, ICGNMD C, Bhatia R, Garg A, Wilson L, Macken W, Pitceathly R, Straub V, Hanna M, Vishnu V

¹All India Institute of Medical Sciences, New Delhi, India, New Delhi, India

500P First neuromuscular center experience in Iraq clinical, radiological, histopathological, and genetic findings from 10 families with LGMDR2

Ali A¹, Omairi S, El Sherif R, Nishino I, Al_Ghurabi M

¹Neurologist at Al-Hadi Neuromuscular Center, Authority of Health and Medical Education, Holy Shrine of Imam Hussain, Karbala, Iraq

501P Clinical characteristics and prevalence of autosomal dominant calpain3-related limb-girdle muscular dystrophy D4

Arntzen K^{1,2}, Høyer H, Ørstavik K, Mathisen L, Varhaug K, Strand L, Von der Lippe C, Løseth S, Jonsrud C, Hoem G, Van Ghelue M

¹Department of Neurology and National Neuromuscular Centre, Norway, University Hospital of North Norway, Tromsø, Norway,

²Institute of Clinical Medicine, University of Tromsø, The Arctic university of Norway, Tromsø, Norway

502P A LGMD22 muscular dystrophy pathological and transcriptomic study

Poyatos Garcia J^{1,2,3}, Vilchez R, Muelas N, Sevilla T, Novella A, Azorin I, Martí P, Bargiela A, Artero R, Vilchez J

¹Human Translational Genomics Group, University of Valencia, Valencia, Spain, ²Centre for Biomedical Network Research on Rare Diseases (CIBERER,CB23/07/00005), Madrid, Spain, ³INCLIVA Biomedical Research Institute, Valencia, Spain

503VP AAV-based TCAP delivery rescues collapsed desmin cytoskeleton-driven mitochondria dislocation in limb-girdle muscular dystrophy R7

Lv X¹, Yan C, Lin P

¹Qilu hospital of shandong university, Jinan, China

504P Limb-girdle muscular dystrophy R1 in Egyptian and regional patients: clinical spectrum, genetics, and registry insights

Mowafy R^{1,2}, Minami N, Nishino I, El Sherif R

¹MyoCare National Foundation, Cairo, Egypt, ²Newgiza University, Giza, Egypt

Poster Session 2 – 551P-566P: EDMD, OPDM, autophagic, extramuscular

551P Patient-reported outcome measures in nuclear envelope myopathies in a large cohort of patients

Cheli M¹, Cugudda E, Gemma M, Maggi L

¹Neuroimmunology and Neuromuscular Diseases Unit Foundation IRCCS Neurological Institute Carlo Besta, Milan, Italy

552P Oculopharyngodistal myopathy with CGG repeat expansions in RILPL1: clinical and pathogenic insights from two Chinese families

Chen X¹, Jiao K, Gao M, Yue D, Zhu H, Li X, Zhao C, Zhu W, Xi J

¹Department of Neurology, Huashan Rare Disease Center, National Center for Neurological Disorders, Huashan Hospital, Fudan University, Shanghai, China

553P First identification of a CGG repeat expansion in LRP12 in Korean families with oculopharyngodistal myopathy type 1

Choi Y¹, Kim S, Choi Y, Park H

¹Department of Neurology, Gangnam Severance Hospital, Yonsei University College of Medicine, Seoul, South Korea

554P Uncovering social and pragmatic communication in Duchenne muscular dystrophy: a preliminary cohort study

Dainesi M¹, Quaranta C, Corradini M, Gardani A, Boselli A, Di Franco S, Khalil S, Ballante E, Berardinelli A

¹Department of Brain and Behavioral Sciences, University of Pavia, Pavia, Italy

555P Multisystem involvement in oculopharyngodistal myopathy: insights into cardiac manifestations and genotype-phenotype correlations

Jiao K¹, Chen X, Yue D, Gao M, Zhao C, Huang G, Xi J, Zhu W, Zhu H

¹Huashan hospital, Fudan University, Shanghai, China

556P Characterization and differentiation of iPSCs from an oculopharyngodistal myopathy type 5 patient harbouring an ABCD3 CCG repeat expansion

Johari M^{1,2}, Clayton J, Singh A, Lamont P, Ravenscroft G

¹The Harry Perkins Institute for Medical Research, Perth, Australia, ²Centre of Medical Research, The University of Western Australia, Perth, Australia

557P A muscular dystrophy associated with bi-allelic LEMD2 variants: expanding the genotype of nuclear envelopathies

Kölbel H¹, Pauper M, Hentschel A, Schänzer A, Beltran S, Thompson R, Aksel-Kilicarslan O, Gangfuß A, Schara-Schmidt U, Lochmüller H, Polavarapu K, Roos A

¹Department of Pediatric Neurology, Centre for Neuromuscular Disorders, University Hospital Essen, Essen, Germany

558P Differential diagnosis in dysphagia: a histopathological study of the cricopharyngeal muscle

Labella B^{1,2}, Lacene E, Chanut A, Beuvin M, Labasse C, Madelaine A, Brochier G, Stojkovic T, Eymard B, Lacau Saint Guily J, Evangelista T

¹Neuromuscular Morphology Unit, Institut De Myologie, Paris, France, ²Department of Neuropathology, Groupe Hospitalier Universitaire La Pitié-Salpêtrière, Paris, France

559P Thromboembolic events in myotonic dystrophy type 1 patients – a case series

Machado J^{1,2,3}, Lessard L, Eisenkölbl A, Aleman A, Klein A, Lochmüller H

¹Division of Neurology, Department of Medicine, The Ottawa Hospital, Ottawa, Canada, ²Faculty of Medicine, University of Ottawa, Ottawa, Canada, ³The Ottawa Hospital Research Institute, Ottawa, Canada

560P Oculopharyngodistal myopathy with CGG repeat expansion in LOC642361/NUTM2B-AS1 from Indonesian population

Indrawati L, Jiao K, **Mutiani F^{1,2}**, Devianca N, Afany N, Uganda Y, Safri A, Fadli N, Harsono A, Wiratman W, Budikayanti A, Octaviana F, Xi J, Chen X, Zhu W, Hakim M

¹Department of Neurology, Dr. Cipto Mangunkusumo Hospital, Jakarta, Indonesia, ²Faculty of Medicine Universitas Indonesia, Jakarta, Indonesia

561P Cryptic exon inclusion reflects severity of hereditary rimmed vacuolar myopathies

Qiao L^{1,2,3}, Hayashi S, Funaguma S, Iida A, Noguchi S, Nishino I

¹Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan, ²Department of Genome Medicine Development, Medical Genome Center, National Center of Neurology and Psychiatry, Tokyo, Japan, ³Department of Neurology, Tianjin Huanhu Hospital, Tianjin, China

562P Danon disease in Japan: A nationwide additional analysis

Sugie K^{1,2,3}, Shiota T, Yamaoka M, Yamanaka A, Ohashi T, Nishimori Y, Nanaura H, Eura N, Kiriya T, Mori E, Nakamura S, Nishino I

¹Department of Neurology, Nara Medical University, Kashihara, Japan, ²Center for Autophagy and Anti-Aging Research, Nara Medical University, Kashihara, Japan, ³Department of Neuromuscular Research, National Institute of Neurology, National Center of Neurology and Psychiatry, Kodaira, Japan

563P Oculopharyngeal muscular dystrophy and dysphagia: preliminary results from a retrospective description of practices in a Canadian care centre

Trudel P^{1,2}, Rouleau L, Blais M, Touri O, Picher-Martel V, Dupré N

¹CRCHU de Québec - Université Laval, Quebec, Canada, ²Département de chirurgie, Faculté de médecine et des sciences de la santé, Université de Sherbrooke, Sherbrooke, Canada

564P Development of an antisense oligonucleotide therapy for oculopharyngeal muscular dystrophy using iPSC-derived myocytes

Bertheau M, **Trudel P^{1,2,3}**, De Serres-Bérard T, Blais M, Dumont N, Dupré N, Gros-Louis F, Picher-Martel V

¹Neuroscience Axis of CHU de Québec – Université Laval Research Center, Quebec, Canada, ²Faculty of Medicine of Université Laval, Quebec, Canada, ³Département de chirurgie, Faculté de médecine et des sciences de la santé, Université de Sherbrooke, Sherbrooke, Canada

565P International multicentre registry-based study on effect of LMNA mutations' spatial distribution on cardiac outcomes in laminopathies

Benyaou R^{1,2,3,4}, Bhaskaran A, Helms A, Fayssoil A, Richard P, Stojkovic T, Anselme F, Labombarda F, Chikhaoui C, De Sandra-Giovannoli A, Leturcq F, Vigouroux C, Dembele M, Elliott P, Zeppenfeld K, Charron P, Kumar S, Lakdawala N, Bonne G, Wahbi K
¹Institut de myologie, Paris, France, ²Database unit, Institut de myologie, Paris, France, ³Sorbonne Université, INSERM, Centre de Recherche en Myologie, Institut de myologie, Paris, France, ⁴AP-HP Sorbonne université, GH Pitié Salpêtrière, Centre de Référence Maladies Neuromusculaires Nord/Est/Ile-de-France, Institut de myologie, Paris, France

566P Optimization of in vitro models' generation to unveil the molecular pathomechanisms underlying PLN4-related myopathy

Carnazzi A^{1,2}, Iannibelli E, Giagnorio E, Sian V, Sarparanta J, Gibertini S, Nicolini De Gaetano L, Riolo G, Salerno F, Marcuzzo S, Savarese M, Maggi L, Ruggieri A

¹Department of Pharmacological and Biomolecular Sciences, University of Milan, Milan, Italy, ²Neuroimmunology and Neuromuscular Disease Unit, Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milan, Italy

19:00-20:00 **Industry Symposium 5** 📍 Hall G

Evolving with SMA: A journey from survival to independence

Chair: Marcondes Franca, University of Campinas (UNICAMP), Brazil

Speakers: Eugenio Mercuri, Fondazione Policlinico Gemelli, Italy & Tim Hagenacker, University Hospital Essen, Germany

19:00-20:00 **Industry Symposium 6** 📍 Hall k

Breakthroughs in Becker: Unveiling New Natural History Insights and a Novel Agent's Clinical Progress

Speakers: Roxana Donisa Dreghici, Edgewise

Therapeutics, United States of America, Erik Niks, Leiden University Medical Center, The Netherlands, & Craig McDonald, UC Davis Health, United States of America

Poster Session 3 – 56P-89P: Advances in therapies and drug development

56P 3D human iPSC-based modelling of laminopathies to develop gene therapy strategies for LMNA-related congenital muscular dystrophy

Moore D, Rossi L, [Aghaeipour Dehkaei A](#)^{1,2}, Carraro E, Steele-Stallard H, Tsz Yan Wong C, Lionello V, Choi S, Pinton L, Jalal S, Cuisset J, Bonne G, Serio A, Zammit P, Tedesco F

¹Department of Cell & Developmental Biology, University College London, United Kingdom, ²The Francis Crick Institute, London, United Kingdom

57P An inhibitory peptide for myostatin alleviates dystrophic pathology in dystrophin-deficient DBA/2-mdx mice

[Sunada Y](#)¹, Nishimatsu S, Fujino M, Ohsawa Y

¹Kawasaki Medical School, Kurashiki, Japan

58P Wave surfing in Duchenne muscular dystrophy; fun or folly?

Fleerackers E, Meijer-Krom Y, Govaarts R, Rauh S, Hoek R, Michaels M, Verduijn J, Hordijk F, Niks E, [Van Der Holst M](#)^{1,2,3}

¹Leiden University Medical Center, The Netherlands, Leiden, Netherlands, ²Duchenne Center Netherlands, Leiden, Netherlands,

³Basalt Rehabilitation, The Hague, Netherlands

59P Uncovering therapeutic leads for SELENON-related congenital myopathy: a large-scale drug screen using patient-derived primary myoblasts

Ho A, Quijano-Roy S, [Ferreiro A](#)^{1,2}

¹INSERM UMRS 974 Sorbonne University, Myology Research Center, Institute of Myology, Paris, France, ²Neuromuscular

Disorders Reference Center, Neuromyology Department, AP-HP, Pitié-Salpêtrière University Hospital, Paris, France

60P Stakeholder perspectives on the development of an intramuscular CRISPR/Cas9 based gene editing therapy (GET) for Duchenne muscular dystrophy

[Pirson I](#)¹, de Graeff N, de Vries M, Niks E

¹Leiden University Medical Center, Leiden, Netherlands

61P Blockade of myostatin restores sarcopenia and extends lifespan in alpha-Klotho-deficient senescence-model mice

[Ohsawa Y](#)¹, Nishimatsu S, Fujino M, Sunada Y

¹Neurology (Kawasaki Medical School), Kurashiki, Japan

62P Oral treatment with TWN-404 blunts cardiac fibrosis and reduces inflammation and myocardial damage in mdx mouse model of Duchenne muscular dystrophy

[Bista P](#)¹, Chandran

¹Twine Therapeutics, Baltimore, United States

63P Targeting a premature termination mutation in COL6A2 for collagen VI-related dystrophy

[Oseguí Barcenilla N](#)¹, González Moro, Lopez Martinez A, Kapetanovic S, Arechavala Gomeza V

¹Nucleic Acid Therapeutics for Rare Diseases (NAT-RD), Biobizkaia HRI, Barakaldo, Spain

64P Morpholino therapy for collagen VI-related muscular dystrophy by modulating PDGFRα isoform in muscle FAPs

[Yae Y](#)^{1,2}, Ogawa M, Hayashi S, Nishino I, Noguchi S

¹Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo,

Japan, ²Department of Clinical Genome Analysis, Medical Genome Center, National Center of Neurology and Psychiatry, Tokyo, Japan

56P 3D human iPSC-based modelling of laminopathies to develop gene therapy strategies for LMNA-related congenital muscular dystrophy

Moore D, Rossi L, [Aghaeipour Dehkaei A](#)^{1,2}, Carraro E, Steele-Stallard H, Tsz Yan Wong C, Lionello V, Choi S, Pinton L, Jalal S, Cuisset J, Bonne G, Serio A, Zammit P, Tedesco F

¹Department of Cell & Developmental Biology, University College London, United Kingdom, ²The Francis Crick Institute, London, United Kingdom

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¹Leiden University Medical Center, The Netherlands, Leiden, Netherlands, ²Duchenne Center Netherlands, Leiden, Netherlands,

³Basalt Rehabilitation, The Hague, Netherlands

59P Uncovering therapeutic leads for SELENON-related congenital myopathy: a large-scale drug screen using patient-derived primary myoblasts

Ho A, Quijano-Roy S, **Ferreiro A**^{1,2}

¹INSERM UMRS 974 Sorbonne University, Myology Research Center, Institute of Myology, Paris, France, ²Neuromuscular Disorders Reference Center, Neuromyology Department, AP-HP, Pitié-Salpêtrière University Hospital, Paris, France

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¹Neurology (Kawasaki Medical School), Kurashiki, Japan

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¹Nucleic Acid Therapeutics for Rare Diseases (NAT-RD), Biobizkaia HRI, Barakaldo, Spain

64P Morpholino therapy for collagen VI-related muscular dystrophy by modulating PDGFR α isoform in muscle FAPs

Yae Y^{1,2}, Ogawa M, Hayashi S, Nishino I, Noguchi S

¹Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan, ²Department of Clinical Genome Analysis, Medical Genome Center, National Center of Neurology and Psychiatry, Tokyo, Japan

65P Patient perspective of participation in clinical studies in LOPD: structured exit interviews from cipaglucosidase alfa plus miglustat studies

Sitaraman Das S, Gault J, Amon F, Holdbrook F, Padilla M, Alexander K, Johnstone S, Giuliano J, **Clarke S**¹

Madrigal Hernandez M

¹Amicus Therapeutics Ltd, Marlow, United Kingdom

66P Cipaglucosidase alfa and alglucosidase alfa enzymes have similar stability at neutral pH and can be stabilised with miglustat

Cosmanescu F, Feng J, Mehta N, Brudvig J, Weimer J, **Clarke S**¹

¹Amicus Therapeutics Ltd, Marlow, United Kingdom

67P Mexiletine Paediatric Investigation Plan, PIP4 study: safety, pharmacokinetic and efficacy findings in children with myotonia

Barnéras C, Isapof A, Hogrel J, Pentikis H, Adetoro N, **Zozulya Weidenfeller A**¹

¹Lupin Atlantis Holdings AG, Zug, Switzerland

68P Suspected dorsal root ganglion toxicity after intratecal gene therapy

Marti Carrera M^{1,2}, Sarasua-Miranda A, Hernandez-Dorronsoro U, Fernández-Torrón R, Olasagasti Calzarcorta V, Iannaccone S, Bharucha-Goebel D, Bönnemann C, Gray S, Pirovolakis T, Messahel S

¹Donostia University Hospital, San Sebastian, Spain, ²Biogipuzkoa Health Research institute, San Sebastian, Spain

69P Clinically important improvements in adults with late-onset Pompe disease switching from alglucosidase alfa to cipaglucosidase alfa plus miglustat

Claeys K^{1,2}, Kushlaf H, Wenninger S, Hummel N, Clarke S, Crabtree M, Fox B, Gloeckner B, Holdbrook F, Jain V, Peceny M, Schoser B

¹Department of Neurology, University Hospitals Leuven, Leuven, Belgium, ²Department of Neurosciences, Laboratory for Muscle Diseases and Neuropathies, KU Leuven, Leuven Brain Institute (LBI), Leuven, Belgium

70P Patient-reported outcomes from a phase 3 study of givinostat in patients with Duchenne muscular dystrophy

De Waele L¹, McAdam L, Alessi F, Muelas N, Gruppioni K, Cazzaniga S, Bettica P, Zaidman C

¹University Hospitals Leuven, Leuven, Belgium

71P Safety of pyrimidine nucleos(t)ide therapy in Thymidine kinase 2 deficiency: an integrated analysis from a pooled dataset

Domínguez-gonzález C^{1,2,3}, Garone C, Haas R, Paradas C, Scaglia F, Chiang C, Colson A, VanMeter S, Hilson K, Hirano M

¹Neuromuscular Diseases Unit, Neurology Department, Hospital Universitario 12 de Octubre, Madrid, Spain, ²Hospital Universitario 12 de Octubre (imas12), Madrid, Spain, ³Biomedical Network Research Centre on Rare Diseases (CIBERER), Madrid, Spain

72P Efficacy and safety of efgartigimod PH20 SC in adult participants with active idiopathic inflammatory myopathy: phase 2 results from the ALKIVIA study

Chinoy H, **Wanschitz J¹**, Rodriguez-Garcia S, Neto A, Papadopoulou D, Van Baelen B, Duncombe P, De Ceuninck L, Van Der Woning B, Aggarwal R

¹Department of Neurology, Medical University of Innsbruck, Innsbruck

73P Long-term safety and efficacy of nipocalimab in generalized myasthenia gravis: vivacity-MG3 open-label extension phase results

Borst K¹, Antozzi C, Vu T, Ramchandren S, Nowak J. R, Farmakidis C, Bril V, De Bleecker J, Yang H, Minks E, Park S J, Grudniak M, Smilowski M, Sevilla T, Hoffmann S, Sivakumar K, Youssef E, Sanga P, Karcher K, Zhu Y, Sheehan J, Sun H

¹Johnson & Johnson Innovative Medicine, Vienna, Austria

74P Design of a phase 1/2 study assessing the safety and preliminary efficacy of ASP2957 gene therapy for X-linked myotubular myopathy (XLMTM)

Ganguli A¹, Gentyala R, Graham R, Mayer O, Chaparro A, Shell R, Li P, Coats J, Muntoni F, Skjolaas K, Lawlor M, Kuntz N, Dhawan A, Dowling J, James L, Tran H

¹Astellas Pharma Global Development, Inc., Northbrook, IL, United States

75P JM17, a novel curcumin analog modulates mitochondrial, inflammatory, and fibrotic pathways in the muscle of a transgenic SBMA mouse: transcriptomic insights beyond NRF2 activation

Pai T¹, Liu Y, Gromova A, La Spada A, Chen Y

¹AnnJi Pharmaceutical, Taipei, Taiwan

76P 208-week efficacy and safety of cipaglucosidase alfa plus miglustat in patients with late-onset Pompe disease treated from PROPEL baseline: muscle function and biomarkers

Toscano A¹, Bratkovic D, Claeys K, Díaz-Manera J, Mozaffar T, Roberts M, Schoser B, van der Ploeg A, Jain V, Holdbrook F, Moore P, O'Brien-Prince K, Kishnani P

¹ERN-NMD Center for Neuromuscular Disorders of Messina, Department of Clinical and Experimental Medicine, University of Messina, Messina, Italy

77P ADHERE+ trial interim analysis: long-term safety and efficacy of efgartigimod in chronic inflammatory demyelinating polyneuropathy (CIDP)

Eggers C¹, Allen J, Lin J, Stettner M, Guptill J, Istas G, De Roeck A, Kuwabara S, Lauria G, Querol L, Suresh N, Karam C, Skripuletz T, Rinaldi S, Echaniz-Laguna A, Hewamadduma C, Van Hoorick B, Yamasaki R, van Doorn P, Lewis R

¹Kepler University Hospital, Linz, Austria

78P Go high or go long? Pharmacokinetics of AAV in a large animal model

Hildyard J¹, Riddell D, Harron R, Stathopoulou T, Robinson A, Esteves V, Sophiana P, Wells D, Pelligand L, Piercy R

¹Royal Veterinary College, London, United Kingdom

79P Observed efficacy of efgartigimod in generalized myasthenia gravis across patient subgroups in the ADAPT-SC+ study

Kellermair L¹, Muppidi S, Vu T, Brauer E, Kerstens R, Utsugisawa K, Meisel A

¹Kepler University, Linz, Austria

80P Delandistrogene moxeparvovec in Duchenne muscular dystrophy: long-term EMBARK 2-year functional outcomes, safety, and micro-dystrophin expression

Muntoni F¹, Mendell J, McDonald C, Mercuri E, Ciafaloni E, Komaki H, Leon-Astudillo C7, Nascimento A, Proud C, Schara-Schmidt U, Veerapandian A, Zaidman C, Murphy A, Rodino-Klapac L

¹Dubowitz Neuromuscular Centre, NIHR Great Ormond Street Hospital Biomedical Research Centre, Great Ormond Street Institute of Child Health and Institute of Neurology, University College London, & Great Ormond Street Hospital Trust, London, United Kingdom

81P Harnessing CRISPR activation to upregulate TTN as a potential treatment for titinopathies

Lee A¹, Yuen M, Oates E, Quinlan K

¹School of Biotechnology and Biomolecular Sciences, University of New South Wales, Sydney, NSW, Australia

82P Enhancing drug assessment for Duchenne muscular dystrophy using organ-on-a-chip technology and nanoplasmonic biosensing of myotube integrity

Tejadera Villafranca A¹, Ruiz-Gutiérrez M, Ugarte-Orozco M, Cortés-Reséndiz A, Ramón-Azcón J, Fernández-Costa J

¹Institute for Bioengineering of Catalonia (IBEC), Barcelona, Spain

83P Phase 1 trial in healthy participants of KER-065, modified activin receptor ligand trap, support development in DMD and other neuromuscular disorders

Natarajan H¹, Taimi M, Hall S, Bobba S, Jiang Y, Grayson D, Seehra J, Chyung Y, Bogdanovich S

¹Keros Therapeutics, Lexington, United States

84P Autologous myoblast transplantation combined with cricopharyngeal myotomy for oculopharyngeal muscular dystrophy: long-term results of 24 patients and comparison with cricopharyngeal myotomy alone

Trollet C¹, Marhic A, Fer F, Brechenmacher M, Negroni E, Butler-Browne G, Mouly V, Marolleau J, Larghero J, Perie S, Lacau St Guily J

¹Sorbonne Université, INSERM, Institute of Myology, Centre of Research in Myology, UMRS 974, Paris, France

85P Efficacy of metformin and erythromycin in myotonic dystrophy type 1: a systematic review in vitro, in vivo and clinical studies

Wiguna F^{1,2}, Mutiani F, Fadli N, Wiratman W, Safri A, Octaviana F, Hakim M, Indrawati L

¹Department of Neurology, Faculty of Medicine Universitas Indonesia, Jakarta, Indonesia, ²Department of Neurology, Dr. Cipto Mangunkusumo Hospital, Jakarta, Indonesia

86P Antibody fusion proteins containing domains from the tripartite motif family protein 72 (TRIM72) increase plasma membrane repair to treat muscle diseases

Giarrano G¹, Lopez Perez M, Weisleder N

¹Department of Physiology and Cell Biology, College of Medicine, The Ohio State University, Columbus, Ohio, United States

87P Results from a dose escalation gene therapy study of ATA-100, AAV9 vector encoding FKRP, in patients with limb girdle muscular dystrophy R9

Vissing J¹, Richard I, Stojkovic T, Straub V, Preisler N, Zanfongnon R, Degove S, Buscara L, Genries-Ferrand S, Olivier S

¹University of Copenhagen, Copenhagen, Denmark

88P BIN1 gene replacement reverts BIN1-related centronuclear myopathy in mice

Ji J, Giraud Q, Swain S, Diedhiou N, Lipkow E, Spiegelhalter C, Laporte J¹

¹Cerbm Gie - IGBMC, Illkirch, France

89P LPA-integrin signaling axis drives skeletal muscle fibrosis

Cruz M^{1,2}, Faúndez-Contreras J, Córdova-Casanova A, Gutiérrez C, Gallardo F, Brandan E

¹Facultad de Odontología, Universidad San Sebastian, Santiago, Chile, ²Fundación Ciencia & Vida, Santiago, Chile

Poster Session 3 – 104P-116P, 117VP: Cell insights, muscle homeostasis

104P P2X7 receptor: controversial role in alpha-sarcoglycan muscular dystrophy

Gazzerro E¹, Astigiano C, Principi E, Pintus S, Benzi A, Panicucci C, Passalacqua M, Sierra-Marquez J, Nicke A, Antonini F, Del Zotto G, Cicatiello A, Raffaghello L, Rezzonico Jost T, Grassi F, Bruzzzone S, Bruno C

¹Unit of Muscle Research, ECRC, Charite University and Max Delbrück Center for Molecular Medicine, Berlin, Germany

105P Effects skeletal muscle fibre network on muscle tissue electrical properties observed by MRI and explained using mathematical model

Stručić M¹, Šmerc R, Serša I, Miklavčič D, Kranjc M, Mahnič-Kalamiza S

¹UL Faculty of Electrical Engineering, Ljubljana, Slovenia

106P Development of sarcomere-observable mice for analyzing skeletal myofibril degeneration

Noguchi S¹, Ogawa M, Kobirumaki-Shimozawa F, Inoue Y, Inoue T, Fukuda N

¹National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan

107P Targeting mitochondrial dysfunction in late-onset spinal muscular atrophy mice: metformin as potential therapeutic drug

Schmitt L¹, Roos A, Hezel S, Hentschel A, Liebig K, Erkelenz S, Odersky A, Schara-Schmidt U, Kleinschnitz C, Casas A, Hendgen-Cotta U, Hagenacker T, Leo M

¹Department Of Neurology, University Medicine, Essen, Germany

108P RYR1 expression in the myogenesis of normal and dystrophic muscles

Zogbi I, Souza B, Souza L, Feitosa L, Bigot A, Mouly V, Vainzof M¹

¹Human Genome Research Centre, University of Sao Paulo, Sao Paulo, Brazil

109P Reducing uncertainty in variant interpretation - upscaling an in vitro functional experiment to test hundreds of variants in ACTN2

Ranta-aho J^{1,2}, Jonson P, Sarparanta J, Udd B, Savarese M

¹Folkhälsan Research Center, Helsinki, Finland, ²University of Helsinki, Helsinki, Finland

110P Astrocytic inward rectifier potassium channel Kir4.1 dysfunction as a target for new therapeutic strategies in late-onset spinal muscular atrophy

Leo M¹, Schmitt L, Liebig K, Hezel S, Bonanno S, Marcuzzo S, Roos A, Kleinschnitz C, Schara-Schmidt U, Maggi L, Hagenacker T

¹Department Of Neurology, University Medicine Essen, Essen, Germany

111P Interaction of spinal microglia and astrocytes in a mouse model of late-onset spinal muscular atrophy

Liebig K¹, Schmitt L, Hezel S, Kleinschnitz C, Hagenacker T, Leo M

¹Department of Neurology, University Medicine Essen, Essen, Germany

112P Proteomics reveals CIRBP dysregulation in rhabdomyolysis: a study of muscle and blood toward the definition of a minimal invasive regeneration marker

Chen L¹, Guettsches A, Hentschel A, Ruck T, Schara-Schmidt U, Della Marina A, Schaenzer A, Roos A

¹Department of Pediatric Neurology, Centre for Neuromuscular Disorders, University Duisburg-Essen, Essen, Germany

113P Further optimized techniques for 2D myotube culture

Jonson P¹, Sarparanta J, Sian V, Vainio A, Luque H, Udd B, Savarese M

¹Folkhälsan Research Center, Helsinki, Finland

114P Novel insights into molecular mechanisms leading to congenital fibre-type disproportion (CFTD)

Rostedt F^{1,2}, Seaborne R, Harro R, Laarne M, Turku T, Kangas H, Krag T, Vissing J, Ochala J, Pelin K, Laitila J

¹Folkhälsan Research Center, Helsinki, Finland, ²Department of Medical Genetics, Medicum, University of Helsinki, Helsinki, Finland

115P Dmd(mdx) and actb knockout mice share a common redox-dependent mechanism of eccentric contraction-induced force loss rescued by hydrogen sulfide

Fallon K^{1,2,3}, Southern W, Lindsay A, Lowe D, Ervasti J

¹Department of Integrative Biology and Physiology, University of Minnesota, Minneapolis, United States, ²Department of Biochemistry, Molecular Biology & Biophysics, University of Minnesota, Minneapolis, United States, ³Division of Rehabilitation Science, Department of Family Medicine & Community Health, University of Minnesota, Minneapolis, United States

116P Primary cilium is determinant of myogenic differentiation in human MuSCs

Saric A¹, Conscience O, Albertella L, Periou B, Souvannanorath S, Malfatti E, Relaix F, Martin L, Authier F2

¹Paris Est Créteil University, INSERM, IMRB U955-Team Biology of Neuromuscular System, F-94010 Créteil, France

117VP Generation of a C2C12-based model for the investigation of disease mechanisms in the early development of Duchenne muscular dystrophy

Aigner-Radakovics K¹, Sukseere S, Fransche L, Schmidt W, Bittner R

¹Medical University of Vienna, Neuromuscular Research Department, Vienna, Austria

Poster Session 3 – 251P-274P, 275VP, 276P-296P, 297VP: Dystrophinopathies (animals models, biomarkers, brain, genetics)

251P A plasma micro sampler to enable blood sampling at home for better monitoring of dystro pathology in Duchenne muscular dystrophy (DMD)

Arthur P, Oh W, Tsioutsias I, Grounds M

¹University Of Western Australia, Perth, Australia

252P Trace nutrients and clinical conditions in Duchenne muscular dystrophy

Baba Y^{1,2,3}, Takahashi Y, Namatame S, Shigeyama T, Murakami T, Yatabe K, Suzuki M, Tamura T, Ogata K

¹Department of Neurology, National Hospital Organization Higashisaitama Hospital, Hasuda, Saitama, Japan, ²Department of Neurology, Graduate School of Medicine, The University of Tokyo, Bunkyo, Tokyo, Japan, ³Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry (NCNP), Kodaira, Tokyo, Japan

253P Optical genome mapping localizes DMD exon 55 duplication downstream of the DMD gene in a male patient without features of a dystrophinopathy

Bhimarao Nagaraj C^{1,2}, Ulm Seiwert E, Cayan A, Liu J2, Tian C

¹Cincinnati Children's Hospital, Cincinnati, United States, ²University of Cincinnati, Cincinnati, United States

254P Activation of complement cascade may trigger inflammation and fibrosis in Duchenne muscular dystrophy: a preclinical investigation on two murine models

Boccanegra B¹, Tulumiero L, Mantuano P, Lenti R, Denoyelle S, Marinelli M, Cappellari O, De Luca A

¹Department Of Pharmacy - Drug Sciences - University of Bari "Aldo Moro", Bari, Italy

255P STIM1 reduction prevents tubular aggregate formation and impacts on muscle function in ageing mice

Pérez-Guàrdia L, Silva-Rojas R, Laporte J, **Böhm J¹**

¹Cerbm Gie - IGBMC, Strasbourg, France

256P Self-reported activity levels and associated levels of personal meaning in adults with Duchenne muscular dystrophy: using the MAPA-F and EMAS to inform need for psychological intervention

Bonney-Murrell C¹, Bouquillon L, McDonald R, Eilon T, Geagan C, Conn R, Rodney S, Turner C, Guglieri M, Straub V, Quinlivan R

¹University College London Hospitals NHS Foundation Trust, London, United Kingdom

257P Spectrum of dystrophin mutations in a dystrophinopathy cohort in north India

Chandu M¹, Macken W, Rani N, Dalal A, Wilson L, ICGNMD C, Bhatia R, Garg A, Pitceathly R, Thangaraj K, Straub V, Srivastava P, Hanna M, Vishnu V

¹All India Institute Of Medical Sciences, New Delhi, India

258P Two-step newborn screening identifies unexpected number of Duchenne muscular dystrophy female carriers

Chrzanowski S^{1,2,3}, Falk E, Coyne F, Cherkerzian S, Parad R

¹Umass Chan Medical School, Worcester, United States, ²Brigham and Women's Hospital, Boston, United States,

³Boston Children's Hospital, Boston, United States

259P Pax3 protein induction enhances muscle stem cell activation in mdx5cv mice

Das Barman S¹, Stella M, Olsen S, Jensen U, de Morree A

¹Aarhus University, Aarhus, Denmark

260P The FORCE platform delivers CNS functional improvement by resolving anxiety in the mdx mouse model of DMD

Desjardins C¹, Li S, Hall J, Venkatesan R, Correia S, Cui J, Johnson J, Weeden T, Beskrovnaya O, Zanotti S

¹Dyne Therapeutics, Waltham, United States

261P Non-invasive tissue oxygenation monitoring in Duchenne muscular dystrophy rat models

Egijian M^{1,2}, Eskandari R, Tyler S, Morrison L, Diop M, Hoffman L

¹University Of Western Ontario, London, Canada, ²Lawson Research Institute, London, Canada

262P Unsolved challenges in gut-muscle axis: the role of microbiota and aging in mediating immune pathology in murine models of Duchenne muscular dystrophy

Farini A¹, Villa C, Strati F, Cassani B, Facciotti F, Torrente Y

¹Fondazione Irccs Cà Granda Ospedale Maggiore Policlinico, Milan, Italy

263P The DMDmdx rat: a representative model of Duchenne muscular dystrophy skeletal, cardiac, and respiratory deficiencies

Finan-Marchi A¹, Donnarumma E, Bertil Froidevaux E, Bordier F, Cosette J, Georger C, Jimenez S, Marolleau B, Miranda A, Saghbini S, Schmitt R, Tamin S, Tanniou G, Van Wittenberghe L, Blaie S, Creoff E, Braun S, Buscara L, Daniele N

¹Généthon, Evry-Courcouronnes, France

264P Characterization of dystrophin expression in muscle biopsies from patients with in-frame deletions in the DMD gene

Frair E¹, Dufresne G, Zhang X, Meyer A, Atkins S, Gabel L, Dunn D, Lay J, Vetter T, Waldrop M, Weiss R, Butterfield R, Nicolau S, Flanigan K

¹Center for Gene Therapy, Abigail Wexner Research Institute, Nationwide Children's Hospital, Columbus, OH, United States

265P Neurobehavioral profiles in young steroid-naïve children with Duchenne muscular dystrophy (DMD): a baseline data analysis from the FOR-DMD trial

Gadaleta G^{1,2}, Geagan C, Schiava M, Riguzzi P, Hendriksen J, Campbell C, McDermott M, Martens W, Gregory S, Griggs R, Guglieri M

¹Neuromuscular Unit, Department of Neurosciences "Rita Levi Montalcini", University of Turin, Turin, Italy, ²John Walton Muscular Dystrophy Research Centre, Clinical and Translational Research Institute, Newcastle University and Newcastle Hospitals NHS Foundation Trusts, Newcastle upon Tyne, United Kingdom

266P Cognitive profile of adults with Becker muscular dystrophy: findings from the BIND consortium

Geagan C¹, Riguzzi P, Kolesnik A, Govaarts R, Miranda R, Hendriksen J, Weerkamp P, Stemmerik M, Würzler Slipsager A, Vissing J, Skuse D, Muntoni F, Kan H, Mercuri E, Straub V

¹John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle Upon Tyne, United Kingdom

267P The DuMAND network: a global collaboration to improve understanding and clinical care for neurobehavioral difficulties in Duchenne muscular dystrophy

Geuens S¹, Cuveele E, Mahal S, Sejersen T, Zanoteli E, Camelo C, Woodcock I, McMillan H, McAdam L, Klein A, Steiner L, Geagan C, Guglieri M, Schara-Schmidt U, Kirschner J, Haberlova J, Niks E, Govaarts R, De Waele L, and other members of the DuMAND Network

¹University Hospitals Leuven, Leuven, Belgium

268P Comprehensive evaluation of muscle integrity biomarkers to assess therapeutic efficacy in Duchenne muscular dystrophy

Gonzalez P¹, Boehler J, Ralff M, Bhavsar P, Soustek-Kramer M, Marshall J, Harmelink M, Christoforou N, Brooks G

¹Solid Biosciences, Boston, United States

269P Decreased cerebral blood flow in Duchenne muscular dystrophy is not associated with corticosteroid regimen or absence of Dp140

Geuens S, Doorenweerd N, Niks E, Kan H, De Waele L, **Govaerts R^{1,2}**

¹Leiden University Medical Center, Leiden, Netherlands, ²Duchenne Center Netherlands, Leiden, Netherlands

270P Individuals with Duchenne muscular dystrophy have lower brain volumes, altered white matter microstructure and lower perfusion compared to controls

Govaerts R¹, Seunarine K, Flores Lopez G, Guliaeva I, Smyth L, Kerkelä L, Brogna C, Verdolotti T, Weerkamp P, Doorenweerd N, Geagan C, Hollingsworth K, Mercuri E, Niks E, Straub V, Muntoni F, Kan H, Clark C

¹Leiden University Medical Center, Leiden, Netherlands

271P Development and validation of a screening tool for brain-related comorbidities in dystrophinopathies: results from the EU-funded multicenter BIND project

Hendriksen J^{1,2}, Miranda R, Weerkamp P, Kolesnik A, Geagan C, Chieffo D, Slipsager A, Bagnasco M, Vroom E, Vissing J, Niks E, Desguerre I, Staub V, Skuse D, Mercuri E, Muntoni F

¹Kempenhaghe Centre for Neurological Learning Disabilities, Heeze, Netherlands, ²Maastricht University, School for Mental Health and Neuroscience, Maastricht, Netherlands

272P Neuronal density in the bulbar cardiorespiratory nuclei of the dystrophin-deficient mdx model

Hermes T¹, Corsini W, da Silva M, de Almeida L, dos Reis L, Sato M

¹Anatomy Department, Federal University of Alfenas (UNIFAL-MG), Alfenas-MG, Brazil

273P Structural neuroimaging of the central nervous system in Duchenne muscular dystrophy: a multimodal MRI analysis

Pedroza Martins A¹, Rabelo de Brito M, Junqueira R, Rezende T, Iwabe C, Conte G, Nucci A, Cendes F, Cavalcante França Junior M

¹Faculty of Medical Science, Department of Neurology, State University of Campinas, UNICAMP, São Paulo, Brazil, Campinas, Brazil

274P Establishment of a 3D cardiomyopathy model for Duchenne muscular dystrophy using patient-derived iPS cells

Inazuka F¹, Funakoshi S, Fujiwara Y, Sasaki M, Ida K, Sakurai H, Yoshida Y

¹Center for iPS Cell Research and Application, Kyoto University, Kyoto, Japan

275VP Magnetic resonance imaging based radiomic feature extraction for bone quality assessment in corticosteroid treated boys of Duchenne muscular dystrophy

Kunnath Ravindranunni R¹, Rosen A, Cottrell C, Vandenborne K, Glenn W, Rajapakse C, Willcocks R

¹University Of Florida, Gainesville, United States

276P Quantitative measurement of near full-length dystrophin and muscle content normalizer proteins in human muscle by IA-UPLC-MS/MS

Larimore K¹, Greenslade A, Hamir M, Jahr K, Crockett L, Van Vleet J, Blank B, Ntai I, Sun B, Zhou H, Melton A

¹Biomarin Pharmaceutical Inc., Novato, California, United States

277P Defining the relationship between DMD mutation type and location within the exons 1-5 window to IRES activity

Lay J¹, Gushchina L, Lin H, Frair E, Nicolau S, Flanigan K

¹The Center for Gene Therapy, Nationwide Children's Hospital, Columbus, United States

278P Flash electroretinogram alterations in the absence of distinct retinal dystrophins in different mouse models of Duchenne muscular dystrophy

Liber A¹, Telles Salgueiro Barboni M, Aoki Y, Kremers J, Vaillend C

¹Paris-Saclay Institute of Neuroscience, Saclay, France

279P Cardiac affection in women carrying pathogenic variants in the dystrophin gene: a 7-year follow-up study

Lyu Z¹, Joensen H, Scharff Poulsen N, Slipsager A, Teitssdóttir B, Vejlsstrup N, Vissing J

¹Rigshospitalet, Copenhagen, Denmark

280P A possible skeletal muscle modulator: chemokine CXCL12 in senior citizen, healthy younger adult and Duchenne muscular dystrophy (DMD) patient

Maeda Y¹

¹National Hospital Organization Kumamoto Saishun Medical Center, Kumamoto, Japan

281P Investigation of genotype-phenotype correlation in 2D cell models of Duchenne muscular dystrophy patients shows mutation-dependent defects in mitochondrial respiration and delay in myogenic program

Marinelli M¹, Cristiano E, Quarta R, Boccanegra B, Cerchiara A, Barile S, Mouly V, Lasorsa F, Imbrici P, Cappellari O, De Luca A

¹Department of Pharmacy & Drug Sciences - Section of Pharmacology, University of Bari Aldo Moro, Bari, Italy

282P Dystrophin isoform-dependent neurocognitive profiles in Duchenne muscular dystrophy: findings from the EU-funded multicenter BIND study

Miranda R¹, Weerkamp P, Kolesnik A, Geagan C, Chieffo D, Slipsager A, Suárez Bagnasco M, Vroom E, Vissing J, Niks E, Desguerre I, Straub V, Skuse D, Mercuri E, Muntoni F, Hendriksen J

¹Universidad Complutense de Madrid, Madrid, Spain

283P Genomic rearrangements in Duchenne muscular dystrophy diagnosed by RNA and long-read genome sequencing

Nicolau S^{1,2}, Koboldt D, Meyer A, Frair E, Roggenbuck J, Ronaldo d, Mack D, Weiss R, Flanigan K

¹Nationwide Children's Hospital, Columbus, United States, ²The Ohio State University, Columbus, United States

284P Cell therapy-mediated dystrophin supplementation improves muscle fatigue tolerance rather than maximal contraction torque in DMD model mice

Sakurai H¹, Bourgeois Yoshioka C, Takenaka-Ninagawa N, Yokomizo-Goto M, Miki M, Watanabe D, Yamamoto M, Aoyama T

¹Center for iPS Cell Research and Application, Kyoto University, Kyoto, Japan

285P Creatine/creatinine ratio and myostatin as biomarkers to monitor muscle function in Duchenne muscular dystrophy

Spitali P¹, Degan C, Tsonaka R, de Vries S, Ikelaar N, van der Holst M, Kan H, Niks E

¹Leiden University Medical Center, Leiden, Netherlands

286P Evaluating a panel of plasma biomarkers of DMD in taurine-treated juvenile mdx mice

Tsioutsias I¹, Terrill J, Bakker A, Arthur P

¹The University of Western Australia, Perth, Australia

287P 3D tissue engineered skeletal muscle to model dystrophinopathies and determine the efficacy of exon skip therapies

Timp L¹, Franken M, Gonçalves M, Goyenvallé A, Bigot A, Mouly V, Pijnappel P, de Greef J, van der Maarel S, Niks E, Goossens R, Aartsma-Rus A

¹Leiden University Medical Center, Department of Human Genetics, Leiden, Netherlands

288P Characterization of four novel humanized Duchenne muscular dystrophin mouse models with deletions of exon 44, 45, 52 or 53

Van Putten M¹, Tanganyika-de Winter C, van de Vijver D, van der Pijl L, Greenthaner S, Walsh M, de Vos K, Linssen M, Claassens J, Brouwers C, Hohenstein P, Aartsma-Rus A

¹Leiden University Medical Center, Leiden, Netherlands

289P Natural history study of the D2-mdx mouse for Duchenne muscular dystrophy: a comparison across two independent study sites

Van Putten M¹, Boccanegra B, Gordish-Dressman H, Mantuano P, Mele A, Tanganyika-de Winter C, Cappellari O, Tulimiero L, Van De Vijver D, Engelbeen S, Suidgeest E, van der Weerd L, Aartsma-Rus A, De Luca A

¹Leiden University Medical Center, Leiden, Netherlands

290P Cognitive comorbidities and brain microstructural changes in patients with Duchenne muscular dystrophy: data from Hong Kong

Yee P¹, Zhang H, Ng R, Mak K, Lee T, Lochmüller H, Chan S

¹Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong SAR, China

291P TREAT-NMD neuromuscular network: variations in DMD mutation distribution in the Duchenne muscular dystrophy dataset

Zito I¹, Martin A, Chan S, Denger B, Bogue L, Mattei F, De Angelis F, Yee P, Masic D, Poll A

¹Parent Project aps, Roma, Italy

292P Duchenne muscular dystrophy newborn screening via elevated serum creatine phosphokinase resulting in alternate diagnoses: a single center experience

Zygmunt A^{1,2}, Vilaisaktipakorn P, Nagaraj C, Reebals L, Tian C

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293P Pseudo-normalization of creatine phosphokinase in a newborn with Duchenne muscular dystrophy: a case report

Zygmunt A^{1,2}, Buls M, Tian C

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294P Novel dystrophic mouse models with duplication and/or large deletion mutations: an invaluable tool for targeted exon skipping and dystrophin restoration in DMD patients

Gushchina L¹, Khan A, Dufresne G, Bradley A, Stevens K, Frair E, Vetter T, Lin H, Flanigan K

¹Jerry R. Mendell Center for Gene Therapy, Nationwide Children's Hospital, Columbus, United States

295P Gene therapy for Duchenne muscular dystrophy related brain dystrophin deficiency in mdx52 mouse model

Tetorou K^{1,2,3}, Gil-Garzon M, Songsilph N, Chu W, Vallve C, Privolizzi R, Beggs S, Ng J, Muntoni F

¹The Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom, ²National Institute for Health Research UCL Great Ormond Street Institute of Child Health, London, United Kingdom, ³Genetic Therapy Accelerator Centre, Queen Square Institute of Neurology, University College London, United Kingdom

296P Variability in the efficacy of antisense oligonucleotide treatment among DMD patients: correlation between genome and transcriptome

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297VP Genetic and functional predictors of disease progression in Duchenne muscular dystrophy using artificial intelligence

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Poster Session 3 – 366P-398P: Myotonic dystrophy

366P Caregivers experiences of and perspectives on living and coping with adult-onset myotonic dystrophy type 1

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367P Healthcare professionals' experiences of and perspectives on the hospital follow-up of patients with adult-onset DM1

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368P Mortality and clinical characteristics in DM1 patients: analysis of two large European registries

Bassez G¹, Wang Y, Nigim F, Wang B, Herweijer E, Quignot N, Read S, Kihlblom S, Ekström A

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369P TRACK DM: design of longitudinal natural history study in people with myotonic dystrophy linking retrospective data with prospective follow-up

Bassez G¹, Gyenge M, Zozulya Weidenfeller A

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370P Benchmarking methods to measure CTG repeat lengths and mosaicism in myotonic dystrophy type 1

McMichael O, Hartman J, Carrell E, Johnson N, **Carrell S**¹

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371P Investigating hand myotonia: determining best practices using novel technologies and approaches to assessing hand myotonia

De Monts De Savasse C¹, Ismail S, Karman L, Vogt-Domke S, Khonde S, Parker D, Tang W, McIntyre M, Duong T

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372P Integrating cognition and neuroimaging: a study of cerebral regions in myotonic dystrophy

Duong T¹, Karman L, Anna Yao M, Ismail S, Rogers M, Deutsch G, Sampson J, Day J, Hageman N

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373P Clinical outcomes and respiratory dysfunction in adults with myotonic dystrophy type 1

Fossmo H^{1,2,3,4}, Robinson H, Ørstavik K

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374P New conditional mouse model for myotonic dystrophy expressing the human DMPK gene carrying large CTG expansions in striated muscles

Furling D¹, Sureau A, Arandel L, Cordier A, Rouxel C, Huguet A, Megali P, Lemaitre M, Mougenot N, Rau F, Klein A, Muchir A, Gourdon G

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375P Long-term observational study of disease progression in DM1 – analysis of the Saguenay, Quebec DM1 cohort

Gagnon C^{1,2}, Côté I, Ripollone J, Larimer P, Connor S, Mathieu J

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376P Aligning research and patient perspectives in myotonic dystrophy type 1: a comparative analysis using data from the DM-scope registry

Gyenge M¹, Brechenmacher M, Fer F, Hamroun D, Geille A, Bassez G, Filnemus Myotonic Dystrophy Study Group

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377P CoreDMScope_ENSA (RevEal the burdeN in daily life in myotonic dyStrophy due to myotoniA): study design compares patient-reported and clinical outcomes

Gyenge M¹, Bassez G, Ellafi R, Zozulya Weidenfeller A, Dufresne R, Filnemus Myotonic Dystrophy Study Group

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378P The role of epigenetic regulation of the DMPK locus in congenital myotonic dystrophy

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379P The UK myotonic dystrophy patient registry – advancing research and amplifying patient voices through a powerful translational tool

Bohill J¹, Hickson L, Walker H, Sodhi J, Turner C, Monckton D, Hewamadduma C, Hamilton M,

Ashley EJ, Walker M, Adcock K, , Marini-Bettolo C

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380P DYNE-101 targets the underlying cause of DM1 to enable multi-system functional improvement in the ACHIEVE trial

Kerr D, **Sansone V**¹, Bassez G, Diaz-Manera J, Mul K, Lilleker J, Pane M, Roxburgh R, Schoser B, Turner C, Ray S, Chen H, Andersson S

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381P Serum FGF21 and GDF-15 as mitochondrial stress biomarkers in myotonic dystrophy type 2: a German multicenter study

Valeria S^{1,2}, Kleefeld F, Ruck T, Schoser B, Roos A, Mensch A

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382P The Rasch-transformed gastrointestinal symptoms rating scale in myotonic dystrophy type 1 (RT-GSRS-DM1)

La Fontaine L^{1,2}, Hamadeh T, Hoeijmakers J, van Kuijk S, Merkies I, Faber C

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383P Multisystem involvement and disability status in adult-onset myotonic dystrophy type 1: a Chinese single-centered study

Luo S¹, Zhong H, Zhang W, Zhao C

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384P Pentatricopeptide repeat protein targeting CUG repeat RNA ameliorates RNA toxicity in a myotonic dystrophy type 1 mouse model

Nakamori M^{1,2}, Imai T, Mlyai M, Nemoto J, Yagi Y, Nakanishi O, Mochizuki H

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385P Mitochondrial network alterations in myotonic dystrophy type 1 disease

Nogales-Gadea G¹, Maestre-Mora P, Valls-Roca L, Mosqueira-Martin L, Ferriz-Gordillo A, Chojnacki J, Cámara Y, Vallejo A, Garrabou G, Pérez-Berná A, Suelves M

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386P A phase 1/2 trial (Galileo Study) of VX-670 in adults with myotonic dystrophy type 1 (DM1): study design

Paker A, Lochmuller H, Turner C, Nigim F, **Larimer P¹**, Johnson N

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387P Unveiling the hidden burden of myotonic dystrophy type 1: diagnostic challenges, patient-reported outcomes, and multisystem involvement

Park Y^{1,2}, Shin J, Kim D

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388P Neurofilament light chain correlates with the motor function scales in myotonic dystrophy type 1

Park J¹

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389P Age and causes of death in myotonic dystrophy type 1

Put L¹, Damen M, Rottier N, Smulders F, Karnebeek I, la Fontaine L, Voermans N, Tieleman A, Faber C, Mul K

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390P Clinical and genetic profile of a cohort of myotonic dystrophy from north India

Rani N¹, Macken W, Dalal A, Wilson L, ICGNMD C, Bhatia R, Garg A, Pitceathly R, Thangaraj K, Srivastava P, Hanna M, Vishnu V

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391P A prospective longitudinal observational study in myotonic dystrophy type 1 (dm1): from clinical outcomes and endpoints to clinical trial design

Sansone V¹, Ferrari Aggradi C, Lizio A, Nani M, Dekdebrun J, Eichinger K, Gagnon C, Subramony S, Roxburgh R, Hamel J, Statland J, Mul K, van Engelen B, Elsheikh B, Turner C, Schoser B, Ragole T, Thornton C, Johnson N, on behalf of DMCRTN

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392P Expanding the UK myotonic dystrophy patient registry dataset - improving data collection and amplifying 'patient voice'

Walker H¹, Hickson L, Sodhi J, Monckton D, Ashley E, Marini-Bettolo C

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393P Palliative care and advance care planning in myotonic dystrophy type 1

Willis T^{1,2}, Turner C, Hewamadduma C5, Maidment L, Lilleker J, Kelly P, Nikolenko N, Audet E, Marchant J, Willis D

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394P Myotonic dystrophy type 2 as a phenotypic mimicker of inclusion body myositis

Yun P¹, Moghekar A, Mohassel P

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395P Advancing clinical trials in myotonic dystrophy type 1: refining radiological, clinical and patient-reported outcome measures

Itebeke L¹, Huysmans, Bamps K, Peeters R, Goosens V, Maes F, Dupont P, Claeys K

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396P Development of a sensor-system to regulate gene expression: application to decoy gene therapy for myotonic dystrophy

Arandel L¹, Sureau A, Cordier A, Ronquillo K, Muchir A, Moulay G, Klein A, Gourdon G, Furling D

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397P Metabolic and myogenic defects as a therapeutic target in type 1 myotonic dystrophy (DM1)

Lessard L^{1,2,3,4,5}, Ben Larbi, Weiss-Gayet M, Courchet J, Furling D, Gallay L, Mounier R

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398P Whole-body muscle MRI in myotonic dystrophy type II: semiquantitative and quantitative analysis and correlation with clinical severity

Mensch A¹, Heuschen M, Kölsch A, Kleeberg K, Strube D, Schneider I, Kraya T, Hensel O, Gussew A, Deistung A, Otto M, Stoevesandt D

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538P National plan of action to raise awareness and improve medical care of Duchenne muscular dystrophy (AIM-DMD): first year results

Akinci G¹, Ontas E, Coskun A, Cakir T, Ayanoglu C, Yayci Koken O, Ardicli D, Cinar E, Bektas Ontas H, Okur T, Komur M, Yis U, Yuksel D, Topaloglu H

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539P The latest insights from the Dutch LGMD registry through patient- and clinician-reported data

Hoek R¹, Schrama E, van Hees B, van Reenen R, van der Kooi A, Straathof C, Badrising U, van Duyvenvoorde H, Krom Y, Niks E

¹Department of Neurology, Leiden University Medical Center, Leiden, Netherlands

540P Overview of the Belgian neuromuscular diseases registry: more than a decade of data

Jagut M¹, De Meulemeester N, Baets J, Bysen D, Bissay V, Claeys K, Daron A, De Bleecker J, Deconinck N, Delstanche S, De Waele L, Herdewyn S, Laridant D, Remiche G, Smeets N, Van Parys V, Cosyns M

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541P The global registry for 541P COL6-related dystrophies: insights from over 350 international participants

Mcdonald S¹, Allamand V, Alvarez R, Boddy H, Copier J, Deconinck N, Dziewczapolski G, Ferré X, McAlister B, Mejat A, Sarkozy A, Straub V

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542P The global FKRП registry: improvements in data collection and harmonisation

Mcdonald S¹, Alfano L, Brazzo K, Johnson N, Laurent J, Mathews K, Thiele S, Vissing J, Walter M, Woods L, Ørstavik K, Straub V

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543P Global insights into limb girdle muscular dystrophies from the TREAT-NMD global registries network

Mcdonald S¹, Poll A, Ali F, Brazzo K, Ganley D, Ambrosini A, Guglieri M, Rodrigues M

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544P LUMC neuromuscular disease biobank: biomaterials in the context of real-world data through registration at the source principles

Meijer-krom Y^{1,2}, Hoek R, Verschuuren J, Straathof C, Badrising U, Tannemaat M, Slingerland F, van der Holst M, Snijder R, Niks E

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545P DM1-hub: building a national hub for myotonic dystrophy type 1 in Spain

Nogales-Gadea G¹, Revert Barberà A, Gutiérrez-Gutiérrez G, Figueroa-Bonaparte S, Lara Pelayo A, Sistiaga A, García-García J, Iruzubieta P, Cabrera M, Gómez-Fernández F, Alonso-Pérez J, Suelves M, Pérez Gómez R, Juan-Corral M, Coll Liesa E, López-Martin A, Maestre-Mora P, Borrás D, Arechavala-Gomez A, López-Castel A

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546P Clinical research facilitated by the New Zealand neurogenetic registry & biobank

Rodrigues M^{1,2}, Buchanan C, O'Grady G, Cleland J, Fraser L, Stewart C, Patel S, Roxburgh R2

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547P Establishment of a national registry for GNE myopathy in China: mutational spectrum and preliminary natural history insights

Zhu B^{1,2,3}, Jiao K, Yue D, Gao M, Zhang J, Xia X, Xi J, Zhao C, Zhu W

¹Department of Neurology, Huashan Hospital Fudan University, Shanghai, China, ²National Center for Neurological Disorders (NCND), Shanghai, China, ³Huashan Rare Disease Center, Shanghai Medical College, Huashan Hospital, Fudan University, Shanghai, China

548P Attitudes towards the use of AI on electronic health records for phenotyping and screening of rare diseases: a systematic review

Martin S, Grauman A, Coulter J, Hasan B, Veldwijk J, Hauber B, Hansson M, Anyouzoa A, Nyongui E, Elomaa K,

Zschüntzsch J¹

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549P SMA care UK: ensuring the best respiratory care for individuals with SMA across the UK

Christie-Brown V^{1,2}, Benesperi G, Carter J, Chan E, Edel L, Edwards C, Illingworth M, Messer B, Murphy P, Procter T, Samuels M, Thorman P, Marini-Bettolo C, Childs A

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550P SMA care UK: a national initiative to ensure that those living with SMA in the UK receive the best possible care

Christie-Brown V^{1,2}, Benesperi G, Burley R, Baranello G, Fitzpatrick C, Horrocks I, Lilleker J, Muni-Lofra R, Lomax G, Scoto M, Portia T, Thornton A, Turner C, Marini-Bettolo C, Childs A

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Poster Session 3 – 589P-609P: SMA outcome measures and registries

589P Natural history and treatment response in spinal muscular atrophy type 2 through the revised upper limb module

Carrera Garcia L¹, Expósito-Escudero J, Nungo Garzón N, Pareja A, Fernández García M, Ortez C, Martínez-Salcedo E, Urbano M, Grimalt M, Munell F, Balsells S, Frongia A, Puig-Ram C, García Romero M, Calvo R, López-Lobato M, Pitarch-Castellano I, Natera de Benito D, Nascimento A, CUIDAME Investigator Group

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590P A cross-sectional survey on the management of bulbar function in spinal muscular atrophy SL45498

Carrera Garcia L¹, Ejarque V, León A, Nascimento A, Puig C, Terrance Á, Guillén E, Maurino J, García López S

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591P Enhancing bulbar assessment in spinal muscular atrophy: a Rasch analysis of the international bulbar assessment tool (iBAT) pilot study

Coratti G¹, Dunaway Young S, McGrattan K, Finkel R, Khonde S, Mayhew A, Johnson E, Muni Lofra R

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592P Italian validation of the SMA independence scale-upper limb module

Coratti G^{1,2}, Bravetti C, Pera M, Gadaleta G, Mongini T, Coccia M, Catteruccia M, D'Amico A, Brolatti N, Bruno C, Verriello L, Pessa M, Liguori R, Vacchiano V, Ruggiero L, Zoppi D, Ricci G, Pane M, Mercuri E, on behalf of ITASMAC working group

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593P Longitudinal assessment of 4-year HFMSE changes in SMA II and III patients treated with nusinersen

Coratti G^{1,2}, Bovis F, Pane M, Pasternak A, Sansone V, D'Amico A, Glanzman A, Morando S, Montes J, Dunaway Young S, Duong T, Ricci F, Mongini T, Sframeli M, Hirano M, Darras B, Day J, Finkel R, Mercuri E, on behalf of iSMAC/international SMA consortium

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594P Evaluating oxygen saturation and recovery dynamics in skeletal muscle during exercise in adults with spinal muscular atrophy

Duong T¹, Khonde S, de Monts C, Tang W, Salvatore S, Dunaway Young S, Vogt-Domke S, Yoseph Hailu R, Yatsu L, Taivassalo T, Rogers M, W Day J, Hageman N

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595P CuidAME: state of the Spanish longitudinal registry of SMA patients in 2025

Fernandez Garcia M¹, Garcia Uzquiano R, Puig C, Nungo Garzon N, Garcia Romero M, Exposito Escudero J, Gonzalez Mera L, Alvarez Molinero M, Lopez Lobato M, Martinez Salcedo E, Grimalt Calatayud M, Fernandez Ramos J, Calvo Medina R, Urbano Martin M, Toledo Bravo de Laguna L, Gonzalez Barrios D, Nascimento Osorio A, CuidAME Investigators Group C

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596P Real-life experience with risdiplam in a Brazilian spinal muscular atrophy cohort followed for up to 9 months

Iwabe C¹, Franco da Graça F, Cavalcante França Jr M

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597P Aligning functional scales with neurophysiological biomarkers in adult SMA: guiding clinical and research scale selection

Franco da Graça F, **Iwabe C**¹, Cavalcante França Jr M

¹Department of Neurology – Unicamp, Campinas, Brazil

598P Myostatin levels in SMA following disease-modifying treatments: a multi-center study

Capasso A^{1,2}, Piemonte F, Petrillo S, Coratti G, D'Amico A, Catteruccia M, Pera M, Palermo C, Pane M, Abiusi E, Cicala G, Villa M, Chiara B, Arpaia C, Silvia B, Tiziano F, Bertini E, Comi G, Corti S, Mercuri E

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599P The integration of PROMs and real-world data: a holistic approach to characterise disease burden and treatment impact in spinal muscular atrophy

Carver A¹, Benesperi G, Karkkainen E, Moat D, Page J, Tanner S, Cavalcante E, Fitzpatrick C, Joefield T, Adcock K, Farrugia M, Ilyashenko G, Lilleker J, McConville J, Merrison A, Parton M, Muni Lofra R, Thorman P, Scoto M, Marini Bettolo C

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600P Design and preliminary validation of a clinical outcome measure for SMA patients. SMA-LIFE ML43472 study

Vazquez Costa J^{1,2,3}, Ñungo Garzón C, Povedano Panades M, Toro Tamargo E, Fernández-García M, Rojas Marcos I, González Mera L, Martínez-Moreno M, Terracle Á, Mauriño J, Cattinari M, Rafels A, García López S, Rebollo P

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601P Adult SMA REACH: a real-world data collection study and ready-made infrastructure to support research and improvement initiatives in spinal muscular atrophy

Muni Lofra R¹, Page J, Karkkainen E, Verdu-Díaz J, Carver A, Benesperi G, Thomas A, Tanner S, Michell-Sodhi J, Moat D, Marini-Bettolo C

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602P Maximal strength as a biomarker in adults with spinal muscular atrophy: a cross-sectional comparative study

Ñungo Garzon N¹, Aragon-Gawinska K, Poveda Verdu D, Albert Ferriz A, Hervás D, Lizandra Cortes P, Pitarch Castellano I, Vazquez Costa J

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603P Minimal clinically important difference on the revised Hammersmith scale in patients with spinal muscular atrophy

O'Reilly E^{1,2}, Stimpson G, Duong T, Wolfe A, Milev E, Baranello G, Muntoni F, Scoto M

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604P Three-year follow up of nusinersen therapy in spinal muscular atrophy type 3: functional outcome analysis from a south Indian centre

Parampalli Ravindra S¹, Banavara S, Sumanth S, Khandekar G, Kumar A, Ramesh Babu R, Maganthi M, Agnes Mathew A

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605P Longitudinal study of metabolic biomarkers and muscle function in disease-modifying treatment of spinal muscular atrophy

Pomp L¹, Jeneson J, Prompers J, Gosselink M, Davoli G, Hagnejad A, Asselman F, van der Pol L, Bartels B

¹Child Development and Exercise Center, Wilhelmina Children's Hospital, University Medical Center Utrecht, Utrecht, Netherlands

606P Real-world experience with nusinersen in adults with 5q spinal muscular atrophy: the French SMA registry

Tard C¹, Grimaldi L, Urtizberea A, Laforêt P, Cintas P, Salort-Campana E, Attarian S, Nkam L, Malfatti E, De La Cruz E, Quijano-Roy S, Stojkovic T

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607P Evaluation of clinical features of patients diagnosed with spinal muscular atrophy type 1 receiving nusinersen treatment before and after newborn screening program: a multicenter experience in Türkiye

Komur M¹, Caglar E, Ceylan A, Turkdogan D, Herguner O, Yıldız E, Saltik S, Sarıkaya Uzan G, Tekin Orgun L, Oz Tuncer G, Yayici Koken O, Tekgul H, Ardicli D, Akinci G, Dilber B, Ozgun N, Ozgor B, Carman K, Yis U, Sarigecilli E

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608P Scoliosis development in SMA patients treated with nusinersen: a single-center experience

Komur M¹, Caglar E, Cobanogulları Direk M, Sahan Arslan S, Altinkaya P, Okuyaz C

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609P Testing cognitive development in weak infants with spinal muscular atrophy: insights and challenges

Buchignani B^{1,2}, Coratti G, Cutrì C, Cristofani P, Stanca G, De Sanctis R, Battini R, Pane M, Mercuri E

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689P Tracing ancient gene flow from Persia to Japan through a shared haplotype in GNE myopathy

Yoshioka W¹, Sonehara K, Bakhshandeh M, Ogawa M, Eguchi K, Hayashi S, Okada Y, Najmabadi H, R. Akbari M, Noguchi S, Nishino I

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690VP Pathogenic chaperone-client networks underlie muscle protein aggregation: lessons from DNAJB4 myopathy

Inoue M¹, Daw J, Bhadra A, Pittman S, True H, Findlay A, Weihl C

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691P Mitochondrial dysfunction is driven by imbalanced fission and fusion of mitochondria in myofibrillar myopathy type 5

Lv X¹, Wu W, Lin P

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692P Insights into distal and myofibrillar myopathies: the John Walton muscular dystrophy research centre cohort

Barreto Haagsma A¹, Ravera B, Bolaño Díaz C, Schiava M, Reeves T, Riguzzi P, Kocak G, Elseed M, Harris E, Töpf A, Muni-Lofra R, Wong K, Moat D, James M, Michel-Sodhi J, Diaz-Manera J, Guglieri M, Marini Bettolo C, Straub V, Tasca G

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693P Training of physiotherapists in clinical and functional outcome assessments in patients with GNE myopathy

Bhasin A¹, Sneha S, James M, Hilsden H, Rani N, ICGNMD C, Bhatia R, Garg A, Wilson L, Macken W, Pitceathly R, Straub V, Hanna M, Vishnu V

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694P Natural history of distal and myofibrillar myopathies assessed by clinical and technological outcome measures (dista-myo): longitudinal results

Bortolani S¹, Torchia E, Ravera B, Trombetta V, Vicino A, Gambella M, Villa L, Rabuffetti M, Marzegan A, Cheli M, Piovesan R, Parrotta A, Rolle E, Monforte M, Mongini T, Ricci E, Hogrel J, Maggi L, Sacconi S, Tasca G

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695P Spontaneous oscillatory contraction are from sarcomeric origin in novel MYBPC1 variants associated with tremors

Galli R^{1,2}, Conijn S, Hoomoedt D, Balesar R, Donkervoort S, Bonnemann C, de Winter J, Malfatti E, Ottenheijm C

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696P Precision rehabilitation in distal myopathy-5: a case of functional recovery and life reintegration following long-term physiotherapy

Khanna M¹, Bhatia A, Jaison V, Khanna K

¹Neuromuscular Center of Excellence Department of Neurology Christian Medical College and Hospital, Ludhiana, India

697P Update on the description of 45 patients with p.Ser55Phe variant in the MYOT gene: MYOT-MUR study.

Martinez Marin R¹, Mena Bravo A, Aledo Serrano M, Zmork Martínez G, Sánchez Huertas A, López Grueiro P, Lorenzo Diéguez M, García Leal A

¹Hospital Univ La Paz: Serv Neurología, Madrid, Spain

698P Diagnostic challenge of distal myopathies – three rare cases from neuromuscular centre

Martinka I¹, Jungova P, Hergottova A, Cibulcik F, Spalek P

¹University Hospital Bratislava, Department of Neurology, Bratislava, Slovakia

699P Expanding the spectrum of HSPB8-myopathy

Milone M¹, Putko B, Sorenson E, Cui G, Liewluck T, Niu Z, Litchy W, Mer G

¹Mayo Clinic, Rochester, United States

700P C2C12-CTE – a versatile myotube model for C-terminal titin studies

Sarparanta J¹, Jonson P, Vainio A, Luque H, Hackman P, Udd B

¹Folkhälsan Research Center, Helsinki, Finland

701P Characterization of a knock-in mouse model of GNE myopathy: motor function and pathology

Shin J¹, Choi J, Kim L, Jung J, Bae J, Kim H, Huang S, Lefebvre D, Noguchi S, Kim D

¹Pusan National University Yangsan Hospital, Yangsan, South Korea

702P Novel mutations in titin exon 363 with different phenotypes including a founder mutation in eastern Europe
Sian V¹, Di Feo M, Kurbatov S, Vihola A, Luque H, Konovalov F, Peric S, Duffy C, Kornblum C, Claeys K, Hackman P, Udd B, Savarese M

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Poster Session 4 – 90P-103P: ALS/neuropathy

90P Developing and testing RNA aptamers as anti-excitotoxic drug candidates in ALS mouse models

Niu L¹, Akamatsu M

¹University at Albany, State University Of New York, Albany, United States

91P Uncommon neuropathy: distal acquired demyelinating symmetric neuropathy linked to anti-GM4 antibodies

Ismail M¹, Basiam S

¹Ministry of Health of Malaysia, Kuala Lumpur, Malaysia

92P Charcot-Marie-Tooth type 1J: description of a new family and findings from magnetic resonance neurography

Ramos-fransi A¹, Madrigal I, Garcia-Diez A, Navarro Otano J, Diez L, Isern Segura I, Alejandre A

¹Hospital Clínic Barcelona, Barcelona, Spain

93P A longitudinal natural history study of giant axonal neuropathy

Bharucha-goebel D^{1,2}, Haugland S, Norato G, Donkervoort S, Waite M, Saade D, Todd J, Lehky T, Orbach R, Foley A, Potticary A, Smith C, McAnally M, Fink M, Jain M, Bonnemann C

¹National Institutes Of Health/ NINDS (Neuromuscular and Neurogenetic Disorders of Childhood Section, Bethesda, United States, ²Children's National Hospital, Washington, DC, United States

94P Hereditary sensory and autonomic neuropathy type 2 (HSAN2) in a child: a case report

Cavusoglu D¹, Gokcay B

¹Afyonkarahisar Health Science University, Afyon, Turkey

95P Factors that influence physical activity in patients with Charcot-Marie-Tooth disease: a qualitative study

Granberg A, van Walsem R, Ørstavik K¹, Frich J

¹Section for Rare Neuromuscular Disorders and EMAN, Department of Neurology, Oslo University Hospital, Oslo, Norway

96P The p.Gln170Glu variant expands the ATL3-related phenotype to include a motor axonal neuropathy

Ortez Gonzalez C^{1,2}, Berta Estévez-Arias B, Pijuan J, Durán Escobar M, Expósito - Escudero J, Carrera - García L, Nascimento A, Natera - De Benito D, Hoenicka J, Palau F

¹Neuromuscular Unit, Department of Neurology, Hospital Sant Joan de Déu, Barcelona, Spain ²Applied Research in Neuromuscular Diseases, Institut de Recerca Sant Joan de Déu, Barcelona, Spain

97P FIG4-related neuropediatric phenotypes: rare and multifaceted diagnoses associated with particular protein changes

Roos A^{1,2,3}, Gowda V, Srinivasan V, Garcia L, Anguita C, Hentschel A, Reutlinger C, Muhmann D, Stüve B, Kölbels H, Schara-Schmidt U, Darling A, Natera de Benito D, Gangfuß A

¹Department of Pediatric Neurology, Centre for Neuromuscular Disorders, University Duisburg-Essen, Essen, Germany,

²Children's Hospital of Eastern Ontario Research Institute, Ottawa, Canada, ³Department of Neurology, Medical Faculty, Heinrich Heine University Düsseldorf, Düsseldorf, Germany

98P Stnf-rii restores leptin homeostasis in human adipocytes exposed to plasma from sporadic als patients with fast progressing disease

Vézina A^{1,2}, Boutej H, Picher-Martel V, Kriz J, Dupré N

¹Faculty of Medicine, Université Laval, Quebec City, Canada, ²Neuroscience Axis of CHU de Quebec- Université Laval, Quebec City, Canada

99P Phenotypic and genetic variability in SOD1-associated amyotrophic lateral sclerosis: a three-case Korean series

Choi Y¹, Kim S, Choi Y, Park H

¹Department of Neurology, Gangnam Severance Hospital, Yonsei University College of Medicine, Seoul, South Korea

101P Familial episodic pain syndrome type 2 with SCN10A mutation in identical twin sisters: a twin case report

Nguyen-Younossi N¹, Rosenbohm A

¹Department of Neurology, University of Ulm, Ulm, Germany

102P ALS patients who request medical assistance in dying: a retrospective study from a care centre in Canada

Trudel P^{1,2}, Quesnel-Olivo M, Blais M, Ramanathan U, Dupré N

¹CRCHU De Québec - Université Laval, Quebec, Canada, ²Département de chirurgie, Faculté de médecine et des sciences de la santé, Université de Sherbrooke, Sherbrooke, Canada

103P Pathogenic BICD2 variants, associated phenotypes and related blood biomarkers – a study of 16 new patients

Della Marina A¹, Neuhaan T, Natera de Benito D, Güttches A, Linsa A, Johannsen J, Dobelmann V, Ruck T, Nascimento A, Udd B, Di Feo M, Savarese M, Hagenacker T, Dipti B, Nalini A, Thakur B, Schara-Schmidt U, Hentschel A, Roos A, BICD2 Study group (Abicht A, Muhmann D, Kölbel H, Chetty VK, Öztürk M, Lopercolo D) B

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Poster Session 4 – 218P-250P: Neuromuscular diseases as multisystemic disorders

218P Phenotype-genotype heterogeneity of multisystemic myopathies – a study from India

Baskar D¹, Nalini A, Vengalil S, Nashi S, Polavarapu K, Kotambail A, Arunachal G

¹National Institute of Mental Health and Neuro Sciences (NIMHANS), Bengaluru, India

219P Modified phenotype of neuromuscular diseases associated with Marfan syndrome

Camacho A¹, González M, Pombrol M, Esparza I, Mayo S, Palma C, Garzón L, Hernández Laín A, Núñez N

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220P SMN-associated neurodevelopmental disorders: behavioural characterisation in the Taiwanese mouse model

Demetriou C¹, Frontzek K, Zhou H, Muntoni F, Hanchate N, Baranello G

¹The Dubowitz Neuromuscular Centre, Developmental Neurosciences Department, Great Ormond Street Institute of Child Health, University College London, United Kingdom

221P Preliminary study of functional brain connectivity in adults with spinal muscular atrophy

Demšar N¹, Šuput D

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222P Pediatric neurological phenotype in triple a syndrome

Dontaine P¹, Ulgiati F, Christiaens A, Brachet C, Deconinck N

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223P Prediction of VO2max and cardiopulmonary responses from ventilatory threshold VO2 in neuromuscular disease patients using machine learning

Duong T¹, Montalvo S, de Monts C, Blumberg Y, Vogt-Domke S, Salvatore S, Tang W, Meyer J, Wheeler M, W Day J, Hageman N, Christle J

¹Stanford University, Palo Alto, United States

224P Delayed exercise recovery ventilatory responses between individuals with neuromuscular disease

Duong T¹, Montalvo S, de Monts C, Blumberg Y, Vogt-Domke S, Salvatore S, Tang W, Dunaway Young S, Wheeler M, Day J, Hageman N, Taivassalo T, Christle J

¹Stanford University Neurology Department, Palo Alto, United States

225P Pompe disease in Sweden: a real-world evidence study of disease burden, treatment patterns for enzyme replacement therapy and concomitant medications

Ekström A^{1,2}, Stelmaszuk M, MacCulloch A, Nordin S, Graham R, Lindvall B, Freilich J

¹Child Neurology Department, Queen Silvia Children's Hospital, Gothenburg, Sweden, ²Department of Pediatrics, University of Gothenburg, Gothenburg, Sweden

226P Retrospective assessment of feeding and nutrition after 2 years of risdiplam treatment in younger children with SMA using the CEDAS

Gorni K¹, Baranello G, Conway E, Li Y

¹PDMA Neuroscience and Rare Disease, F. Hoffmann-La Roche Ltd, Basel, Switzerland

227P Deciphering the developmental dystrophinome: exploring dystrophin isoform expression during murine embryogenesis

Hildyard J¹, Wells D, Piercy R

¹Royal Veterinary College, London, United Kingdom

228P Profile of hereditary myopathy patients in Dr Cipto Mangunkusumo hospital as a national tertiary healthcare in Indonesia

Indrawati L, **Isaac W¹**, Nagpal C, Shafitha N, Putri M, Simatupang S, Safri A, Fadli N, Harsono A, Wiratman W, Budikayanti A, Octaviana F, Hakim M

¹Department of Neurology, Dr Cipto Mangunkusumo Hospital, Jakarta, Indonesia

229P Cognition in mitochondrial diseases of adulthood – natural history data from the German mitoREGISTRY

Kaluza L¹, Nadal J, Widmann C, Kornblum C

¹Department Of Neuromuscular Diseases, Centre for Neurology, University Hospital Bonn, Bonn, Germany

230P SMN in mesenchymal progenitors regulates neuromuscular junction in spinal muscular atrophy

Kong Y¹, Kim H, Hann S, Kim J

¹Seoul National University, Seoul, South Korea

231P Systemic deletion of Gne in adult mice results in thrombocytopenia, anemia, spontaneous bleeding, myopathy and premature death

Lam P¹, Zygmunt D, Bennett M, Ashbrook A, Hefty J, Martin P

¹Nationwide Children's Hospital, Columbus, United States

232P Eosinophilic fasciitis: MRI as a key to diagnosis and treatment response

Le S¹, Phan C

¹University Medical Center Of Ho Chi Minh City, Ho Chi Minh City, Vietnam

233P Early muscle strength improvements in patients with GBS treated with tanruprubart is a predictor of better outcomes

Morrison G¹, Kroon H, Islam Z, Gorson K, Kalam Azad K, Navarro J, Lin P, Collins P, Mohammad Q

¹Annexon Biosciences, Brisbane, United States

234P Long-term use of omaveloxolone in patients with Friedreich ataxia: up to 5 years of natural history propensity score matching from the MOXIe OLE

Nachbauer W¹, Lynch D, Delatycki M, Hoyle J, Boesch S, Giunti P, Wilmot G, Zesiewicz T, Subramony S, Matthews K, Perlman S, Lawson R, Shen C, Farooq S, Natarajan S, Domingo-Horne R, Arizpe A, Folschweiller N, Murai M

¹Medical University Innsbruck, Innsbruck, Austria

235P Phenotypic characterization of the neuromuscular manifestation of replication factor C subunit 4 (RFC4)-related multisystem disorder

Patel R¹, Wright S, Orbach R, Barros Perez M, Lehky T, Saade D, Pais L, Abrams C, Ghosh P, Moore S, Lopes Abath Neto O, Foley A, Bharucha-Goebel D, Donkervoort S, Bönnemann C

¹Neuromuscular and Neurogenetic Disorders of Childhood Section, National Institute of Neurological Disorders and Stroke, National Institutes of Health, Bethesda, Maryland, United States

236P Prevalence of peripheral neuropathy in children with advanced chronic kidney disease in a tertiary care center - a cross-sectional study

Raveendran R¹, Iype M, Uthup S, Sreedharan M, Ahamed S, R Lal R, Anna Cherian A, Habeeb A, Sahu P, Parveen S

¹Thiruvananthapuram government medical college, Thiruvananthapuram, India

237P Classical and new orthopedic phenotypes in children and adults with spinal muscular atrophy: lessons for current and future patient care

Ribault S^{1,2}, Karoutchi C, Barrière A, Vuillerot C

¹L'Escale, Service de Médecine Physique et de Réadaptation Pédiatrique, Hôpital Femme-Mère-Enfant, Hospices Civils de Lyon, Bron, France., Lyon, France, ²Pathophysiology and Genetics of Neuron and Muscle, NeuroMyogen Institute, CNRS UMR 5310 - INSERM U1217, Université Lyon 1, Lyon, France., Lyon, France

238P Serious complications and multisystemic involvement in Duchenne muscular dystrophy

Sofou K^{1,2}, Wahlgren L, Nordström S, Kroksmark A, Tulinius M

¹Sahlgrenska University Hospital, Gothenburg, Sweden, ²University of Gothenburg, Gothenburg, Sweden

239P A gene-centric scoring approach for the stratification of patients with rare neuromuscular disorders

Suwalska A¹, Töpf A, Vandroux M, Pini S, Savarese M, Laporte J, Straub V, Santorelli F, Tupler R, Polanska J

¹Department of Data Science and Engineering, Silesian University of Technology, Gliwice, Poland

240P The forest and the trees – muscle symptoms within the POC5-related syndromic landscape

Topf A¹, Martínez-Esteban P, Vissing J, Macken W, Schiava M, Luce L, Mannion J, Yzer S, Jazet I, van Ham T, Roosing S, Straub V, Vulto-van Silfhout A, Oud M, Diaz-Manera J, POC5 Study Group

¹JWMDRC, Newcastle University, Newcastle upon Tyne, United Kingdom

241P Multisystemic manifestations in neuromuscular disorders

Venugopalan Yamuna V¹, Rani N, Sharma M, Jassal B, Macken W, Wilson L, ICGNMD C, Bhatia R, Garg A, Agarwal A, Rajan R, Gupta A, Singh M, Dhall A, Pitceathly R, Dalal A, Thangaraj K, Hanna M, Srivastava P

¹All India Institute of Medical Sciences, New Delhi, India

242P Long-term natural history observations of FHL1-related reducing body myopathy from childhood to adulthood

Foley A¹, Vilaisaktipakorn P, Mohassel P, Yun P¹, Donkervoort S, Zhang W, Leeson M, Rooney J, Zou Y, Bönnemann C
¹Neuromuscular and Neurogenetic Disorders of Childhood Section, NINDS, NIH, Bethesda, Maryland, United States

243P Longitudinal clinical and genetic data collection for limb girdle muscular dystrophies

Wandera S¹, Kocak G, Jeffrey T, Marini-Bettolo C, Diaz-Manera J, Muni-Lofra R, JWMDRC Team
¹John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom

244P Duchenne muscular dystrophy from muscle to brain: a south Asian perspective

Wijekoon N^{1,2}, Gonawala L, Ratnayake P, Amaratunge D, Hathout Y, Steinbusch H, Mohan C, Dalal A, Hoffman E, de Silva R
¹Interdisciplinary Center for Innovation in Biotechnology and Neuroscience (ICIBN), Faculty of Medical Sciences, University Of Sri Jayawardenepura, Nugegoda, Sri Lanka, ²Department of Cellular and Translational Neuroscience, School for Mental Health and Neuroscience, Faculty of Health, Medicine & Life Sciences, Maastricht University, Maastricht, Netherlands

245P Multisystem proteinopathy in neurological disorders: a single-center retrospective analysis

Xia X¹, Chen X, Sun Y, Zhang M, Qiao K, Chen Y, Zhao C, Dong Y, Zhu W¹
¹Huashan Hospital, Fudan University, Shanghai, China

246P Guanine quadruplex structure formation in the pathogenesis of oculopharyngodistal myopathy

Yamanaka A^{1,2}, Eura N, Nakamura H, Hayashi S, Noguchi S, Sugie K, Nishino I
¹National Center of Neurology and Psychiatry, Tokyo, Japan, ²Nara Medical University, Nara, Japan

247P Frequency, risk factors, and implications of fractures in hereditary neuromuscular disorders.

Opsomer M^{1,2}, Iterbeke L, Borghs H, De Cuyper T, Dejaeger M, Dupont P, Claeys K
¹Department of Neurology, UZ Leuven, Leuven, Belgium, ²Department of Neurosciences and LBI, Laboratory for Muscle Diseases and Neuropathies, KU Leuven, Belgium

248P Evaluation of dysphagia in myositis and muscular dystrophy using real-time MRI and quantitative muscle ultrasound

Saris C¹, Zeng R, Rietveld A, Al-Bourini O, Kroon R, Olthoff A, Weidenmüller M, Carstens P, Kommerell I, Schütz S, Horlings C, Kalf J, de Swart B, van Engelen B, Friede T, Hofer S, Frahm J, Seif Amir Hosseini A, Schmidt J
¹Department of Neurology, Radboudumc Research Institute for Medical Innovation, Nijmegen, Netherlands

249P Fetal hypokinesia and arthrogryposis of neuromuscular origin: genetic insights and clinical spectrum

Natera-de Benito D¹, Perez-Vidarte F, Estevez-Arias B, Matalonga L, Yubero D, Codina A, Ortez C, Medina J, de Sena L, Carrera-Garcia L, Exposito-Escudero J, Jou C, Tizzano E, Nascimento A
¹Hospital Sant Joan de Déu, Barcelona, Spain

250P ORAI1 downregulation ameliorates the multi-systemic signs of tubular aggregate myopathy (TAM) and Stormorken syndrome (STRMK) in mice

Pérez i Guàrdia L¹, Laporte J, Böhm J
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Poster Session 4 – 330P-365P: Metabolic and mitochondrial myopathies

330P Cardiovascular risk factors and atheromatosis in spinobulbar muscular atrophy (SBMA) Kennedy in association with lipid and hormone profile

Chadraabal K¹, Gadelkareem M, Nguyen-Younossi N, Mueller S, Imhof A, Buckert D, Kloempken S, Rosenbohm A
¹Department Of Neurology, University of Ulm, Ulm, Germany

331P Causes of death in adult patients with late-onset Pompe disease: a French Pompe registry retrospective study

Chitimus D¹, Tard C, Fournier M, Bouhour F, Béhin A, Salort-Campana E, Lagrange E, Solé G, Spinazzi M, Kaminsky A, Magot A, Beltran S, Noury J, Magy L, Renard D, Lefeuvre C, Attarian S, Hamroun D, Laforet P
¹Hopital Raymond-Poincaré Aphp, Garches, France

332P Silent savior: when one variant defies the other

Dubuisson N^{1,2}, Sergeant K, Poulton J, Carroll L, Brady S
¹Oxford Neuromuscular Centre, Department of Neurology, John Radcliffe Hospital, Oxford, United Kingdom, ²Neuromuscular Reference Centre UCL Saint-Luc, University of Louvain, Brussels, Belgium

333P Long-term follow-up deoxynucleoside therapy for late onset thymidine kinase 2 deficiency patients**Durmus Tekce H¹**, Gedikbaşı A, Ceylaner S, Cakar A, Kıyan E, Parman Y¹Istanbul Faculty of Medicine, Istanbul University, Istanbul, Turkey**334P The McArdle assessment of severity tool (MAST): validation of an innovative new tool to measure the clinical severity of patients with McArdle disease****Finnigan P^{1,2}**, Godfrey R, Mohammed K, Pizzamiglio C, Quinlivan R¹Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology, London, United Kingdom, ²NHS Highly Specialised Service for McArdle Disease and Related Disorders, Queen Square Centre for Neuromuscular Diseases, The National Hospital for Neurology and Neurosurgery, London, United Kingdom**335P Clinical impact of supervised resistance training in patients with McArdle disease: a case series****Finnigan P^{1,2}**, Godfrey R, Mohammed K, Pizzamiglio C, Quinlivan R¹Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology, London, United Kingdom, ²NHS Highly Specialised Service for McArdle Disease and Related Disorders, Queen Square Centre for Neuromuscular Diseases, The National Hospital for Neurology and Neurosurgery, London, United Kingdom**336P The McArdle disease physical activity paradox and insights into exercise adaptation: a case study on running with McArdle disease****Finnigan P^{1,2}**, Torrens S, Godfrey R, Mohammed K, Pizzamiglio C, Quinlivan R¹Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology, London, United Kingdom, ²NHS Highly Specialised Service for McArdle Disease and Related Disorders, Queen Square Centre for Neuromuscular Diseases, The National Hospital for Neurology and Neurosurgery, London, United Kingdom**337P A zebrafish-based platform for investigating disease mechanisms and therapeutic interventions in rhabdomyolysis**Tate G, Kim E, Casey J, Tao B, **Gupta V¹**¹Brigham And Women's Hospital, Harvard Medical School, Boston, United States**338P Clinical and therapeutic clues from a long-term follow-up: a single centre experience on a large LOPD population****Toscano A^{1,2}**, Pugliese A, Porcino M, Trimarchi G, Musumeci O¹ERN-NMD Center for Neuromuscular Disorders of Messina, University of Messina, Messina, Italy, ²University of Messina, Messina, Italy**339P Tubular aggregate myopathy secondary to hypoparathyroidism in a young adult****Ji G^{1,2}**, Song X¹Department of Neurology, the Second Hospital of Hebei Medical University, Shijiazhuang, China, ²Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan**340P High prevalence of GAA c.[752C>T;761C>T] complicates high-risk screening for late-onset Pompe disease in east Asian populations****Jiao K¹**, Wang Y, Yue D, Zhu B, Gao M, Wang Y, Xi J, Zhao C, Tang S, Zhu W¹Huashan hospital, Fudan University, Shanghai, China**341P Exposure to sertraline and ranolazine is common among adult patients with genetically uncharacterized lipid storage myopathy****Jones F¹**, Mirman I, Milone M, Liewluck T¹Mayo Clinic, Rochester, United States**342P Oral taurine therapy is effect for glucose metabolism in patients with mitochondrial disease; a retrospective study****Kurashige T^{1,2}**, Matsuda T, Murao T, Himeno N, Kubota M, Okada R, Ohshita T, Maruyama H¹NHO Kure Medical Center and Chugoku Cancer Center, Kure, Japan, ²Hiroshima University, Hiroshima, Japan**343P Development of a standardized toolbox for autophagy research in neuromuscular disorders within the DREAMS project****Lemerle E^{1,2,3}**, Moparthi S, Forand A, Miagoux Q, Evangelista T, Muchir A, Nissan X, Vassilopoulos S¹Association Institut De Myologie, Paris, France, ²Sorbonne Université, Paris, France, ³Inserm U974, Paris, France**344P Neuropathy associated with activated Bcl2/apoptosis pathway in a new mouse model of multiple acyl-coenzyme A dehydrogenase deficiency (MADD)****Liang W^{1,2,3}**, Lin C, Chang S, Yu Y, Tseng S, Teng Y, Jong Y¹Department of Pediatrics, Kaohsiung Medical University Hospital, Kaohsiung, Taiwan, ²Department of Pediatrics, School of Medicine, Kaohsiung Medical University, Taiwan, ³Graduate Institute of Clinical Medicine, College of Medicine, Kaohsiung Medical University, Taiwan

345P Acquired lipid storage myopathy with MADD acylcarnitine profile secondary to sertraline treatment: an emerging disorder

Llansó Caldentey L^{1,2,3}, Vesperinas A, García-Villoria J, Gort L, Sánchez-Martínez J, Herranz-Marín M, Caballero-Ávila M, Cortés E, Rojas-Marcos I, Carbayo Á, Rojas R, Collet-Vidiella R, Querol L, Pascual-Goñi E, Martín-Aguilar L, Gallardo E, González-Quereda L, Turon-Sans J, Ribes A, Olivé M

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346P Exploring the natural history of late-onset TK2 deficiency: biomarkers and key outcomes

Martín Jiménez P¹, Bermejo Guerrero L, Ochoa L, Navarro Riquelme M, Hernández Lain A, Hernández Voth A, González Quintana A, Bermejo Moriñigo A, González Méndez V, Blázquez A, Domínguez González C

¹Neuromuscular Diseases Unit, Neurology Department, Hospital Universitario 12 de Octubre, Madrid, Spain

347P Muscle MRI phenotyping of a cohort of patients with primary mitochondrial myopathies

Bermejo-Guerrero L, **Martín Jiménez P**¹, Ochoa L, Alcalá-Galiano A, Casado-Pérez C, Bermejo-Moriñigo A, González-Méndez V, Domínguez-González C

¹Neuromuscular Disorders Unit, Neurology Department, Hospital Universitario 12 de Octubre, Madrid, Spain

348P Gloom to glee: momentousness of precision genetic testing in pediatric neuromuscular diseases

Muppavarapu S¹, Kollencheri Puthenveetil V, Anand V, Kesavan S, Issac G, Namboothiri S, Yesodharan D

¹Amrita Institute of Medical Sciences, Department of Pediatric Neurology, Kochi, India

349P Monitoring juvenile LOPD patients “waiting for the disease”

Musumeci O¹, Porcino M, Drago S, Arena I, Toscano A

¹University of Messina, Messina, Italy

350P Mitochondrial dysfunction in lipid storage myopathy associated with sertraline treatment

Oldfors C¹, Lindgren U, Visuttijai K, Shen Y, Ilinca A, Nordström S, Lindberg C, Oldfors A

¹University of Gothenburg, Gothenburg, Sweden

351P Predictive factors to genetic cause of rhabdomyolysis: a brazilian single-center exploratory study

Pedroza Martins A¹, Junqueira R, Rezende T, Rolim Muro Martinez A, Nucci A, Cavalcante França Junior M

¹Faculty of Medical Science, Department of Neurology, University of Campinas, UNICAMP, Campinas, Brazil

352P Type 6 autosomal dominant progressive external ophthalmoplegia associated with a probable novel pathogenic DNA2 gene variant

Remiche G¹, Lecomte S, Comi G, Vandernoot I, Desmyter L, Seneca S, Stepman H, Ronchi D

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353P Unexpected attenuated phenotype in autosomal recessive progressive external ophthalmoplegia linked to a novel homozygous pathogenic RRM2B gene variant

Remiche G¹, Lecomte S, Comi G, Vandernoot I, Seneca S, Stepman H, Ronchi D

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354P Functional and behavioural analysis of the zebrafish model of McArdle's disease

Stefanik E¹, Daczewska M, Budzyńska B, Migocka-Patrzałek M

¹University of Wrocław, Wrocław, Poland

355P Using high-field magnetic resonance spectroscopy to measure muscle glycogen in patients with McArdle disease

Stemmerik M¹, Beha G, Slipsager A, Wiggermann V, Vissing J

¹Copenhagen Neuromuscular Center, Department of Neurology, University of Copenhagen, Copenhagen, Denmark

356P Real-life effectiveness after switching to avalglucosidase alfa in late-onset Pompe disease patients worsening on alglucosidase alfa therapy: a French cohort study

Tard C¹, Sacconi S, Taouagh N, Bouhour F, Michaud M, Beltran S, Fournier M, Demurger F, Lagrange E, Spinazzi M, Noury J, Magot A, Cintas P, Renard D, Arrassi A, Salort-Campana E, Attarian S, Laforet P

¹Filnemus France, Marseille, France

357P Long-term vatiquinone treatment slows Friedreich's ataxia disease progression relative to FACOMS natural history

Vagabov A¹, Cherry J, Duquette A, França Jr M, Perlman S, Durr A, Bertini E, Mathews K, Schöls L, Fournier A, Delatycki M, Subramony S, Roxburgh R, Rance M, Zhang O, Golden L, Gruenert J, Werner C, Lynch D, Zesiewicz T

¹PTC Therapeutics Switzerland GmbH, Steinhausen, Switzerland

358P Harnessing the MyoScreen high content platform to discover ALK5/TGFBR1 inhibitors for muscle mass preservation

Ventre E¹, Flaender M, Travard L, Lambert R, Massera C, Foray M, Duchemin-Pelletier E, Roelants C, Freytag B, Young J, Autier V

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359P Proteomic characterization of storage material in a patient with cardiomyopathy caused by a pathogenic glycogenin-1 (GYG1) missense variant

Visuttijai K¹, Hedberg-Oldfors C, Glamuzina E, Ruygrok P, Occleshaw C, Abernathy M, Turner C, Kingston N, Costello D, Bermingham N, Oldfors A

¹Department of Laboratory Medicine, Institute of Biomedicine, University of Gothenburg, Gothenburg, Sweden

360P Polyglucosan body myopathy caused by RBCK1 inversion: a case report

Zhu B^{1,2,3}, Jiao K, Yue D, Gao M, Luo X, Xi J, Zhao C, Zhu W

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361P Gene replacement therapy for ADSS1 myopathy: preclinical investigation in mice

Yammine K¹, Kim M, Moghadaszadeh B, Hickey E, Brault J, Widrick J, Beggs A

¹Division of Genetics and Genomics, Boston Children's Hospital, Harvard Medical School, Boston, MA, United States

362P Exploring multisystem involvement in primary mitochondrial disorders: insights from a pediatric cohort from India

Raghavan MS V¹, K Gowda V, Vidyadhar Kinhal U

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363P The role of mtDNA release as a signalling molecule in mitochondrial myopathies

Cross E¹, Szabo M, Yung R, Lagos D, Horvath R

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364P Can genetics explain the clinical variability and worldwide distribution of thymidine kinase 2 deficiency?

Pelle S, Pettenuzzo I, Diodato D, Carli S, Procopio E, Bello L, Pegoraro E, Musumeci O, Ronchi D, Mancuso M, **Garone C¹**

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365P Defining the disease mechanism of RBCK1-related polyglucosan body myopathy

Babini G, Ceccanti F, Santi E, Oldfors A, Malfatti E, **Garone C^{1,2}**

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Poster Session 4 – 459P-484P, 485VP: Myasthenia Gravis, NMJ1-2, Periodic paralysis

459P Clinical meaning of PROMs in a large cohort of myasthenia gravis patients

Cheli M¹, Brusa L, Bartolucci F, Mantegazza R, Gemma M, Pennoni F, Maggi L

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460P Real-world experience in Chinese myasthenia gravis patients receiving efgartigimod: a nationwide retrospective study

Cheng Y¹

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461P Exploring biomarker signatures in CMT-subtypes

Dobelmann V¹, Mensch A, Peric S, Vockert K, Vukojevic M, Öztürk M, Kleefeld F, Güttches A, Gangfuss A, Horvath R, Schara-Schmidt U, Nelke C, Ruck T, Dohrn M, Roos A

¹Department of Neurology, Medical Faculty and University Hospital Düsseldorf, Heinrich Heine University Düsseldorf, Düsseldorf, Germany

462P Eculizumab as a rescue therapy in myasthenic crisis in ICU: a case series

Durmus Tekce H¹, Cakar A, Gulsen Parman Y

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463P Refractory myasthenia gravis - clinical course and predictive factors in a large cohort of patients

Giacopuzzi Grigoli E¹, Cavalcante P, Frangiamore R, Bonanno S, Vanoli F, Cheli M, Mehmeti E, Salvi E, Baggi F, Andreetta F, Canciello A, Mantegazza R, Antozzi C, Maggi L

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464P Nuclear envelope proteins in sub-synaptic gene regulation: mechanisms and disease implications

Godard-Bauché S¹, Douarre C, Peccate C, Eucharmakian M, Nasr J, Lemaitre M, Ferrer E, Cadot B, Muchir A

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465P Long term follow-up of slow channel congenital myasthenic syndrome – a retrospective cohort study

Hennehan L¹, Rossini E, Ramjattan H, Dong Y, Beeson D, Ramdas S, Palace J

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466P Effective use of CD19xCD3 T-cell engager blinatumomab in refractory myasthenia gravis

Huntemann N¹, Ruck T, Öztürk M, Schreiber S, Lichtenberg S, Masannek L, Nelke C, Ben Moussa H, Ulrych T, Seifert M, Mougialakos D, Dietrich S, Meuth S

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467P Ravulizumab treatment in refractory myasthenia gravis patients dependent on chronic IVIg

Kierdaszuk B¹, Nieporęcki K, Rosińska A, Kwiatos K, Kostera-Pruszczyk A

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468P Decoding disease, designing therapies: the B2B-RARE precision medicine initiative for hereditary neuromuscular diseases

Oeztuerk M¹, Muhmann D, Nelke C, Güttches A, Enax-Krumova E, Schaenzer A, Klebl B, Klebl K, Fang N, Hentschel A, Heyer R, Smith K, Gies M, Schoser B, Horvath R, Schara-Schmidt U, Vorgerd M, Roos A, Ruck T

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469P Clinical outcomes and treatment efficacy in juvenile myasthenia gravis: a single center study

Paprad T^{1,2}, Nigro E, Aleisa Z, Hernan Gonorazky H

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470P Clinical development program for efgartigimod in juvenile generalized myasthenia gravis

Ramdas S^{1,2}, Kuntz N, Schwaede A, Bogatyreva A, Dalvi J, Giacobbe J, Menezes F, Liu L, van Bragt T, Niks E, Kostera-Pruszczyk A

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471P Congenital myasthenic syndromes subtypes with early onset disease- a large cohort review to guide newborn screening programmes

Ramdas S^{1,2}, Hennehan L, Dong Y, Munot P, Bettolo C, Norwood F, Dougan C, Jungbluth H, Servais L, Palace J

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472P CACNA1S variant mimicking limb-girdle muscular dystrophy

Ramos-fransi A¹, Diez L, Navarro Otano J, Millisenda J, Aldecoa I, Alejaldre A

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473P Caught between two antibodies: a double seropositive myasthenia gravis

Renczésóvá B¹, Vosátková A, Martinka I, Bobek R, Harvanová L, Kečkéš Š, Špalek P

¹Slovak Centre for Neuromuscular Disorders, Department of Neurology, University Hospital Bratislava, Bratislava, Slovakia

474P Temporal trends in management of myasthenia gravis in a tertiary centre in north India

Sharma A¹, Agarwal A, Bhatia R, Rajan R, Singh M, Srivastava A, Garg A, Srivastava P, Vishnu V

¹All India Institute of Medical Sciences, AIIMS (New Delhi), New Delhi, India

475P Higher risk of fractures in myasthenia gravis patients in comparison with general population – national healthcare database study

Sobieszczuk E¹, Szczudlik P, Koń B, Pawlewicz A, Kostera-Pruszczyk A

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476P Myasthenic crisis –15-years' single neuromuscular center experience

Sobieszczuk E¹, Szczudlik P, Badowski K, Opuchlik A, Szyluk B, Rajczewska-Olewszkiewicz C

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477P Continuous subcutaneous rozanolixumab administration in complex myasthenia gravis: a retrospective cohort study

Abi Aoun M, Duval F, Violleau M, Barnay M, Beauvais D, **Solé G¹**

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478P Ocular myasthenia with normal EMG and positive LRP4 antibodies in pediatric patients: a diagnostic challenge

Tsakir C¹, Haspolat S, Yayici Koken O

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479P Role of N103S and L545P MuSK mutations in congenital myasthenic syndrome pathogenesis

Umaro N¹, Herbst R

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480P Comparison of children with myasthenia gravis and congenital myasthenic syndrome: a case series

Yukse D¹, Üstün C, Atasoy E, Solmaz İ, Bayram A, Konuşkan B

¹University of Health Sciences, Ankara Etlik City Hospital, Ankara, Turkey

481P DPAGT1 variant p. Phe257Leu associated with congenital myasthenic syndromes cause endoplasmic reticulum stress

Atchayaram N¹, Dash S, Vashista P, Arunachal G, Ghata C, Markandeya Y

¹National Institute of Mental Health and Neuro Sciences, Bengaluru, India

482P ME&MGopen™ digital biomarkers help track generalized myasthenia gravis symptoms

Berling E¹, Barnett-Tapia C, Lehnerer S, Bieuvelet S, Carment L, Gorin C, Pesic-Heuvrard N, Ravindra D, Sellami N, Dutta B, Yungher B, Zinaï S, Howard, Jr J

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483P Findings from a natural history study of congenital myasthenic syndromes, to establish reliable clinical outcome measures

Ramjattan H¹, Henahan L, Ramdas S, Palace J

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484P Molecular and ultrastructural basis of the neuromuscular junction defect in PURA syndrome

Roos A^{1,2,3,4}, Preusse C, Bielak M, Sobolewska A, Dobelmann V, Della Marina A, Muhmann D, Chrościńska-Krawczyk M, Goebel H, Thakur B, Chetty V, Hentschel A, Stenzel W, Mroczek M

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485VP Neurologists' therapeutic decision-making in generalized myasthenia gravis

Gutiérrez-Gutiérrez G, **Gómez-Ballesteros R¹**, Sotoca J, Ares A, Villaverde R, Reyes V, Armangué T, Querol L, Salas E, Díaz-Abós P, Maurino J, Cortés-Vicente E

¹Roche Farma Spain, Madrid, Spain

Poster Session 4 – 567P-569P, 571P-574P: Pompe disease

567P Novel GAA variants and genotype-phenotype correlation in late-onset Pompe disease: experience from high-risk screening study in China

Jiao K¹, Ke Q, Li J, Wu L, Huang X, Mao S, Yao X, Zhu W

¹Huashan hospital, Fudan University, Shanghai, China

568P Personalized medicine in individuals living with late onset Pompe disease (LOPD): urine glucose tetrasaccharide (Glc4) as a predictive biomarker during follow-up

Lessard L¹, Chitimus D, Froissart R, Bouhour F, Hamroun D, Laforêt P, Pettazzoni M

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569P Clinical outcomes and experiences after discontinuation of enzyme replacement therapy in late-onset Pompe disease

Potters L^{1,2}, Wagenmakers M, van Doorn P, van der Ploeg A, van der Beek N

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571P The Spanish Pompe registry: real-world insights into clinical characteristics and therapeutic transitions in late onset Pompe disease

Domínguez-gonzález C^{1,2,3}, Muelas N, Paradas C, Martínez R, Gómez M, Restrepo J, Campodron M, Morís G, Nedkova V, García García J, Olivé M, Mendoza M, Alonso J, Martín L, Henao J, Hernández-Voth A, Puig C, Segovia S, Díaz-Manera J, Pompe Registry Study Group T

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572P Managing Pompe disease and enzyme replacement therapy during pregnancy: challenges and considerations

van der Beek N¹, Theunissen M, Hayat Z, Jouy A, van den Hout J, Taouagh N, Bouhour F, Noury J, Lagrange E, van Doorn P, van der Ploeg A, Laforêt P

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573P Evaluating Glc4 as a longitudinal marker of functional decline in late-onset Pompe disease

Iannucci D, Gallardo E, Clark J, Domínguez-González C, **Díaz-Manera J**¹

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574P Mobile health technology in late-onset Pompe disease: short-term follow-up after switching to avalglucosidase alpha

Risi B^{1,2}, Ferullo L, Olivieri E, Labella B, Rizzardi A, Caria F, Ait Allali N, Franchi E, Poli L, Bozzoni V, Padovani A, Pilotto A, Filosto M

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Poster Session 4 – 575P-588P: SMA therapies

575P Single-center long-term follow-up of 13 adult patients with spinal muscular atrophy (SMA) treated with risdiplam

Risi B^{1,2}, Caria F, Bertella E, Giovanelli G, Ait Allali N, Garofali F, Carugati R, Gilberti G, Poli L, Bozzoni V, Padovani A, Filosto M

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576P A systematic literature review: real-world motor function outcomes with risdiplam in treatment naïve adults with types 2 and 3 SMA

Gorni K¹, Sutherland C, Davies E, Mahajan A, Kumari S, Simpson A

¹PDMA Neuroscience and Rare Disease, F. Hoffmann-La Roche Ltd, Basel, Switzerland

577P Multisystemic factors from muscle and liver affect bone health in spinal muscular atrophy type II patients and a mouse model

Grandi F¹, Pezet S, Arnould A, Mazzucchi S, Gidaja E, Astord S, Chapart M, Vasseur S, Ricupero A, Ernu M, Sampaio Y, Cohen-Tannoudji M, Meunier P, Benkhalifa-Ziyyat S, Miladi L, Vialle R, Smeriglio P

¹INSERM Center of Research in Myology, Paris, France

578P Treatment effects and perceptions among apitegromab-treated patients with type 2/3 spinal muscular atrophy: patient and caregiver interviews from the TOPAZ clinical trial

Gueye M¹, Mazzone E, Krueger J, Cutts K, Pokrzywinski R, Shah H, Umans K, Darras B

¹Scholar Rock, Inc., Cambridge, United States

579P The effects of nusinersen on the motor function of children with spinal muscular atrophy type 2 and 3

Kosac A^{1,2}, Vucinic D, Cerovac N, Ivancevic N, Nikolic B, Brkusanin M, Mladenovic J, Savic Pavicevic D, Milic Rasic V

¹Clinic Of Neurology and Psychiatry for Children and Youth, Belgrade, Serbia, ²School of Medicine University of Belgrade, Belgrade, Serbia

580P Organizational, economic and environmental impacts of oral and intrathecal administrations for spinal muscular atrophy treatments in France

Marchadier B¹, Masingue M, Kaminsky A, Cazin A, Bellier L, Touvron G, Le Lay K, Remer C

¹Roche SAS France, Paris, France

581P Onasemnogene abeparvovec for spinal muscular atrophy in patients with detectable anti-AAV9 antibodies

Mumneh N¹, Shiloh-Malawsky Y, Darras B, Richardson R, Bharucha-Goebel D, Alecu I, Dodd N, Cialfoni E

¹Novartis, East Hanover, United States

582P Compassionate treatment for spinal muscular atrophy with viral gene therapy – a single centre experience from India

Ramesh Babu R^{1,2}, Satyam P, Maganthi M, Kinimi I, Datta D, Lakshmi K, Ng J, Mathew A

¹Bangalore Baptist Hospital, Bengaluru, India, ²Synapse Neurocenter & Child development Centre, Bengaluru, India

583P Add-on treatment with risdiplam after treatment with onasemnogene abeparvovec: case report of a 2.5-year-old patient with SMA type 1

Drax M¹, Pühringer M, Eisenkölbl A, Gröppel G

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584P Vascular perfusion abnormalities in an infant with spinal muscular atrophy type 0

Unver O¹, Bıkmazer B, Öztürk G, Yılmaz Z, İyışenyürek Ş, Akbeyaz İ, Karakayalı B, Keçecioğlu Binnetoğlu K, Koç A, Yücelten D, Topaloğlu H, Türkdogan D

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585P Delphi consensus project to determine the meaningful treatment outcomes in adult spinal muscular atrophy (SMA)

Vazquez Costa J¹, Chiriboga C, Duong T, Erbas Y, Glascock J, Gusset N, Muni-Lofra R, O'Connell C, Walter M, Guittari C, Cochrane J, Townson L, Riley D, Gasser C, Sully K

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586P Microvasculopathy in spinal muscular atrophy: characterization in patients and therapeutic rescue by SMN-enhancing therapy in mouse models

Zhang Q, Scoto M, Kim J, Demetriou C, Patel P, Hong Y, Baranello G, Muntoni F, **Zhou H¹**

¹University College London, London, United Kingdom

587P ThecaFlex DRx™: implantable intrathecal catheter and subcutaneous port system for repeated intrathecal delivery of nusinersen

Meyer K, **Burton L¹**, Breedlove M, Dennys C, McGuire J, Coughlin R, Raymond B, Holmes M, Washburn T, Wood C, Troche C, De Souza N, Johnson D, Fleege C, Singh D, Coletta C, East A, Anand P, Gambino G, O'Connell K

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588P Expert consensus on gene therapy for spinal muscular atrophy with onasemnogene abeparvovec: treatment decision, administration and patient follow-up

Davion J¹, Desguerre I, Audic F, Cancès C, Barnerias C, Espil-Taris C, Fauroux B, de Feraudy Y, Guichard M, Isapof A, Quijano Roy S, Vuillerot C, Bodiguel E, Project group Expert consensus on gene therapy for Spinal muscular atrophy with onasemnogene abeparvovec P

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Poster Session 4 – 610P-620P, 621VP, 622P-648P, 649VP-650VP: DMD - clinical care and cases reports, BMD

610P Providers' experiences of statewide newborn screening for Duchenne muscular dystrophy in Ohio

Bhimarao Nagaraj C^{1,2}, Oklon P, Leslie N, Berry L

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611P Personalized aerobic training in Duchenne muscular dystrophy (DMD): an exploratory study to evaluate safety, feasibility, and impact on fatigue and quality of life

Branca A¹, Mangone S, Briganti M, Onofri M, Bruschini M, Sancricca C, Frongia A

¹Fondazione UILDM Lazio Onlus, Rome, Italy

612P Bone health and risk of vertebral fracture in DMD: which role the functional status plays?

Capasso A^{1,2}, Arpaia C, Villa M, Panicucci C, Ripetto A, Coratti G, Gulli C, Lala M, Pane M, Bruno C, Mercuri E

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613P Acute pancreatitis in a Duchenne muscular dystrophy patient on total parenteral nutrition

Desikan M¹, Sese L, Purcell H, Astin R, Quinlivan R, Parton M, Emmanuel A

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614P CANYON trial results: sevasetmen, an investigational fast skeletal myosin inhibitor, reduced muscle damage biomarkers and stabilized function in BMD

McDonald C, Kushlaf H, Castro D, Mathews K, Varadhachary A, Connolly A, **Guglieri M¹**, Leung D, Statland J, Niks E, Quinlivan R, Sreenivasan V, Veerapandian A, Johnson N, Phan H, Wong B, Dreghici R, MacDougall J, Russell A, Donovan J

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615P Liver injury after gene therapy in Duchenne muscular dystrophy: a case report and clinical perspective on screening and management

Gonzalez Castillo Z¹, Aqul A, Hassan S, Rojas I, Rakheja D, Batley K

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616P Brain volumetrics, diffusion and perfusion do not differ between men with Becker muscular dystrophy and healthy controls

Govaarts R¹, Seunarine K, Kerkelä , Godtfeldt Stemmerik M, Würgler Slipsager A, Weerkamp P, Doorenweerd N, Geagan C, Hollingsworth K, Straub V, Vissing J, Niks E, Muntoni F, Clark C, Kan H

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617P Characterising motor function in paediatric Becker muscular dystrophy

Grover E¹, Riguzzi P, James M, Moat D, Elseed M, Michell-Sodhi J, Diaz Bolano C, Wong K, Reeves T, Robinson E, Schiava M, Waldoock P, Kocak G, Barreto Haagsma A, Tasca G, Diaz Manera J, Straub V, Marini Bettolo C, Muni Lofra R, Guglieri M

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618P Carriers of DMD-gene variants: an international survey on clinical presentation and care provision to identify gaps and challenges

Guglieri M¹, Behar L, Riguzzi P, Houwen S, Bourke J, Krom Y, Ferlini A, Robinson C, Wong K, Geagan C, Muni R, Segovia S, Fortunato F, Reuben E, Franken M, Zito I, Voermans N, Sarkozy A

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619P DMD hub tools and services to improve clinical trial delivery

Heslop E¹, Gaeta A, Reuben E, Johnson A, Cammish P, Elliott J, Baranello G, Hastie J, Childs A, Straub V, Guglieri M

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620P Family caregivers' administrative burden for persons with Duchenne's muscular dystrophy: a reflection of all stakeholders

Houwen S^{1,2}, Stadhouders N, van Huissteden J, Som T, Jeurissen P, Rake J, Altena- Renssen S, Peters G

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622P DMD care UK five years on: progress update on a successful collaboration between the clinician and patient communities to improve standards of care in Duchenne muscular dystrophy

Kerr A¹, Turner C, Johnson A, Childs A, Quinlivan R, Sarkozy A, Willis T, Wong S, Manzur A, Baranello G, Guglieri M

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623P DMD missense variants in the ZZ domain cause severe dystrophinopathy despite the co-expression of dystrophin and utrophin at the sarcolemma

Kustermann M¹, Aigner-Radakovics K, Schmidt W, Bittner R

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624P The impact of growth on gross motor function in Duchenne muscular dystrophy

Lindqvist J¹, Sofou K, Ekström A

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625P "Your baby has Duchenne. What next?" Validation of a treatment framework for early diagnosed Duchenne muscular dystrophy using the Delphi method

Lorentzos M¹, Servais L, Parsons J, Shell R, Spinty S, Finkel R, Colvin M, Simpson M, Murrell D, O'Grady G, Cardon M, Sejersen T, Osredkar D, Walter M, Stratton A, Tang L, Salmon R, Masic D, Khan I, TREAT-NMD and Regulator Scientific & Health Solutions Support

¹The Sydney Children's Hospitals Network, Sydney, Australia

626P Understanding variation in practices around AAV seroconversion guidelines for patients and staff: a provider pilot survey

Matesanz S^{1,2}, Small J, O'Donnell C, LaRosa J, McCague S, Waldman A, Samelson-Jones B, George L

¹Children's Hospital of Philadelphia, Clinical In Vivo Gene Therapy, Philadelphia, United States, ²University of Pennsylvania, Department of Neurology, Philadelphia, United States

627P Duchenne education in care and research initiative (DECRI): a multi-disciplinary training and education programme in care and research for Duchenne muscular dystrophy

McNiff M¹, Turner C, Heslop E, Kerr A, Guglieri M

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628P Unraveling a veil: a rare genetic aberration causing muscular dystrophy in a female.

Muppavarapu S¹, Anand V, Kollencheri Puthenveetil V, Namboothiri S, Yesodharan D

¹Department Of Pediatric Neurology, Amrita Institute of Medical Sciences, Kochi, India

629P Trajectory of north star ambulatory assessment with sevasemten compares favorably to natural history modeling in Becker muscular dystrophy

Niks E¹, Signorovitch J, Schrama E, Koeks Z, Verschuuren J, Fillbrunn M, Wang S, MacDougall J, Donovan J

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630P Exploring polypharmacy in Duchenne muscular dystrophy

Nordström S^{1,2}, Wahlgren L, Lindberg C, Sofou K

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631P The Becker muscular dystrophy (BMD) hub: supporting care and clinical trial readiness for BMD

Riguzzi P¹, Scott A, Heslop E, Elliott J, Adcock K, Farrugia M, Glover S, Quinlivan R, Reuben E, Willis T, Hick D, Straub V, Guglieri M

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632P Longitudinal analysis of respiratory function in Becker muscular dystrophy

Riguzzi P^{1,2}, Grover E, James M, Schiava M, Diaz Manera J, Tasca G, Moat D, Michell-Sodhi J, Wong K, Robinson E, Waldoock P, Bolano Diaz C, Barreto Haagsma A, Kocak G, Elseed M, Reeves T, Marini Bettolo C, Straub V, Muni Lofra R, Guglieri M

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633P An audit of cardiac care in children and young people with Duchenne muscular dystrophy in Oxford, UK

Skippen A¹, Borsato G, Sa M, Servais L, Ramdas S

¹Department of Paediatric Neurology, John Radcliffe Hospital, Oxford, United Kingdom

634P Joint modelling of growth and motor function centiles in corticosteroids treated boys with Duchenne muscular dystrophy

Stimpson G¹, Ridout D, Wolfe A, Milev E, O'Reilly E, Manzur A, Sarkozy A, Muntoni F, Baranello G

¹Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom

635P Fat fraction of the myocardium in Becker muscular dystrophy and women with patho-genic DMD gene variants

Teitsdottir B¹, Lyu1 Z, Scharff Poulsen N, Würzler Slipsager A, Borgarlø Joensen H, Espe Hansen A, Vejstrup N, Vissing J

¹Copenhagen Neuromuscular Center, Copenhagen, Denmark

636P How a collaboration between clinicians and the patient community can change clinical care: the impact of DMD care UK guidelines on cardiac and respiratory care

Turner C¹, Kerr A, Baranello G, Bourke J, Childs A, Johnson A, Manzur A, Messer B, Quinlivan R, Rodney S, Sarkozy A, Willis T, Wong S, Guglieri M

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637P Assessment of transition readiness for pediatric patients with Duchenne muscular dystrophy

Tian C¹, Vilaisaktipakorn P, Sawani A, Gurbani N, Pascoe J, Stewart C, Horn P, Bange J, Zygmunt A,

¹Cincinnati Children's Hospital Medical Center, Ohio, United States

638P Neuropsychological functioning of boys with Duchenne muscular dystrophy in respect of the dystrophin gene isoforms

Vučinić D¹, Kosac A, Cerovac N, Mladenovic J, Jemuovic T, Pesovic J, Savic Pavicevic D, Milic Rasic V

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639P Functional ability and quality of life in Duchenne muscular dystrophy

Wahlgren L^{1,2}, Nordström S, Lindberg C, Sofou K

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640P Duchenne muscular dystrophy; the use of the 'traffic light' system to plan discussions and advance care planning

Willis T^{1,2}, Kulshrestha R, Jones J, Yeo J, Jackson P, Nevin K, Hewamadduma C, Willis D

¹Robert Jones and Agnes Hunt Hospital, Oswestry, United Kingdom, ²Chester University Medical School, Chester, United Kingdom

641P A cross-sectional comparison of ambulatory versus non-ambulatory patients with Duchenne muscular dystrophy (DMD) at 16 years of age

Nascimento Andrade E, Figueira S, Chrzanowski S, Shellenbarger K, **Wong B**¹

¹DMD Program University of Massachusetts Chan Medical School, Worcester, United States

642P Pathogenicity validation of DMD pseudoexon mutations in patient-derived human induced pluripotent stem cells: a case report

Xia X¹, Jiao K, Hu C, Zhao C, Zhu W

¹Department of Neurology, Huashan Hospital, Fudan University, Shanghai, China

643P Mutations in the actin binding domain of dystrophin are associated with early onset of heart failure in patients with Becker muscular dystrophy

Yamamoto T¹, Ogawa S, Nambu Y, Bo R, Matsuo M, Awano H

¹Nagahama Institute of Bio-science And Technology, Nagahama, Japan

644P Cognitive function in individuals with dystrophinopathies: perspectives from families

Sari Yanartas M, Kutluk G, **Yayici Koken O**¹

¹Akdeniz Üniversitesi, Antalya, Turkey

645P Dystrophin and utrophin quantitation by targeted mass spectrometry in Duchenne and Becker muscular dystrophy

Bello L¹, Riguzzi P, Canessa E, Pegoraro E, Hathout Y

¹Department of Neurosciences, DNS, University of Padova, Padova, Italy

646P Safety of viltolarsen in children under 4 years old in Russia case report

Balamut N¹, Gremiakova T, Gremiakova O, Stepanov A, Gukosian D, Pack S, Kleimenova I, Yamshchikova A

¹R-Pharm, Moscow, Russian Federation

647P Serum levels of ART3 and RGMa are associated with functional performance in Becker muscular dystrophy

Spitali P^{1,2}, Schrama E, van de Velde N, Koeks Z, Verschuuren J, van Zwet E, Kan H, Niks E

¹Leiden University Medical Center, Leiden, Netherlands, ²Duchenne Center Netherlands, Leiden, Netherlands

648P Dysarthria and speech intelligibility problems in Duchenne muscular dystrophy: a 10-year longitudinal study

Scholten S¹, Lagarde M, Erasmus C, Braakman H, Houwen-van Opstal S, Knuijt S, Groothuis J

¹Radboud university medical center, Nijmegen, Netherlands

649VP Quantitative computed tomography, and d vitamin to evaluate bone health in Duchenne muscular dystrophy patients with deflazacort treatment and their functional assessment

Escobar Cedillo R¹, Choque Rojas N, Martínez Coria E, Renan Leon S, Suarez Ochon A, Villarreal Ramirez E, Noriega Martinez R, Gómez Díaz B

¹Instituto Nacional de Rehabilitación, Mexico, Mexico

650VP Neuropsychiatric assessment in adults with Duchenne muscular dystrophy

Eilon T¹, Bindman D, Bouquillon L, Bonney-Murrell C, Geagan C, Conn R, Rodney S, Guglieri M, Straub V, Quinlivan R

¹National Hospital for Neurology and Neurosurgery, University College London NHS Trust, London, United Kingdom

Poster Session 4 - 703LBP-712LBP, 714LBP-719LBP, 721LBP-728LBP, 730LBP-734LBP: Late breaking

703LBP Early add-on therapy improves outcomes in AChR-positive generalized myasthenia gravis: a real-world multicenter cohort study

Ruck T^{1,2}, Öztürk M, Huntemann N, Gerischer L, Stascheit F, Schroeter C, Schubert C, Schneider-Gold C, Pfeuffer S, Krämer H, Konen F, Skripuletz T, Glaubitz S, Zschüntzsch J, Scherwies V, Hagenacker T, Nelke C, Pawlitzki M, Meisel A, Meuth S

¹BG University Hospital Bochum, Department of Neurology, Bochum, Germany, ²Heinrich Heine University Düsseldorf, Department of Neurology, Bochum, Germany

704LBP Reducing tubulin detyrosination levels restores impaired cardiac relaxation kinetics in a NEM6 mouse model

Baelde R¹, Janssen V, Fortes Monteiro A, van den Berg M, Granzier H, Kuster D, van der Velden J, Ottenheijm C, de Winter J

¹Amsterdam UMC, Amsterdam, Netherlands

705LBP Alglucosidase alfa demonstrates effectiveness and safety in Chinese patients with late-onset pompe disease: a multi-center prospective study

Zhao Y¹, Zhu W, Li D, Jiao K, Yang H, Ma M, Qiu W, Wang Z, Hu J, Qiu Z, Shang H, Li B, Wu W, Shen D

¹Qilu Hospital, Shandong University, Jinan, China

706LBP A direct comparison of cerebrospinal fluid (CSF) and intravenous delivery with AAV9 reveals CSF delivery as an alternative route to treat neuromuscular disorders

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707LBP Interim safety results from a phase 1/2 investigational gene therapy study evaluating AB-1003 in patients with limb girdle muscular dystrophy 2I/R9

Passalacqua C¹, Johnson N, Mathews K, Mozaffar T, Statland J, Jiang C, Low S, Parfionovas A, Shafi R

¹AskBio, Research Triangle Park, United States

708LBP The "eotaxin-CCR3 axis" mediates chronic inflammation and immune system cell infiltration in inclusion body myositis muscle

Riolo G¹, Salerno F, Vicino A, Cheli M, Gibertini S, Ruggieri A, Iannibelli E, Carnazzi A, De Gaetano Nicolini L, Bonanno S, Marcuzzo S, Cavalcante P, Maggi L

¹Neurology 4 - Neuroimmunology and Neuromuscular Diseases Unit – Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano, Italy

709LBP Evaluation of the novel SEGRAM GRM-01 in the B10.mdx model of Duchenne muscular dystrophy (DMD)

Krings D¹, Mossad O, Jakob F, Lockhart A

¹Grunenthal, Aachen, Germany

710LBP Risdiplam in adults with 5q-associated spinal muscular atrophy: a nationwide observational cohort study

Keritam O^{1,2}, Erdler M, Fasching B, Zulehner G, Rath J, Krenn M, Kiss C, Troger J, Grinzinger S, Müller P, Mahal S, Bernert G, Baumann M, Zimprich F, Topakian R, Eggers C, Quasthoff S, Löscher W, Cetin H, on behalf of the SMAustria study group

¹Department of Neurology, Medical University of Vienna, Vienna, Austria, ²Comprehensive Center for Clinical Neurosciences and Mental Health, Medical University of Vienna, Vienna, Austria

711LBP Comparative effectiveness of nusinersen and gene therapy in children with spinal muscular atrophy type 1

Ropars J^{1,2,3}, Cances C, Garcia-Uzquiano R, Gomez-Garcia M, Barnerias C, Audic F, Durigneux J, Halbert C, Nkam L, Laugel V, Espil C, Walther-Louvier U, Davion J, Isapof A, Le Goff L, Desguerre I, Quijano-Roy S, Grimaldi L

¹Reference Center for Neuromuscular Disorders, Pediatric Department, Augustin-Morvan Hospital, Brest, France, ²LaTIM INSERM UMR 1101, Brest, France, ³Unité de Recherche Clinique APHP. Université Paris-Saclay, Direction de la Recherche Clinique Assistance Publique Hôpitaux de Paris, Paris, France

712LBP A phase 1a/b open label study of SAT-3247 in healthy volunteers and adult patients with Duchenne muscular dystrophy

Farwell W¹, Dubow J, Wells C, Lambert P, Mitchell R, Rudnicki M, Gleeson F

¹Satellos Bioscience Inc., Toronto, Canada

714LBP Genomic newborn screening on 245 treatable diseases: first data from 1534 babies enrolled within Screen4Care suggest reflections about monoallelic variants reporting in dual dominant/recessive conditions

Ferlini A¹, Margutti A, Agolini E, Matalonga L, Beltran S, Einhorn M, Ruffino B, Macari G, Zuccolo M, Mihalatos M, Martinez-Fresno Hegde M, Novelli A, Bertini E M, Freyler K, Kirschner J, Davoine E, Faivre L, Selvatici R, Sansen S,

¹Department of Medical Sciences, University of Ferrara F, Ferrara, Italy

715LBP A novel mouse model of x-linked spinal muscular atrophy offers a platform for preclinical development of UBA1 targeting therapeutics

Crick H^{1,2}, Smith H, Huang Y, Chaytow H, Farrukh R, Morris K, Parsotam-Chitnavis S, Faller K, Gillingwater T^{1,2}

¹University of Edinburgh, Edinburgh, United Kingdom, ²The Euan MacDonald Centre, Edinburgh, United Kingdom

716LBP Treatment with PBGENE-DMD results in durable improvements in muscle function over time through increased dystrophin expression and dystrophin-positive cells

Gorsuch C¹, Mischler A, Owens G, Lewis W, Heard N, Morris B, Grimason H, Rangarajan K, Wang C, Kodi K, Burgess D, Poe K, Reddick T, Nazarenko D, Shoop W, Chandiramani A, Smith J

¹Precision BioSciences, Durham, United States

717LBP A DMD intron P1 pseudoexon inclusion correlates with protein levels and motor functions in patients carrying exon 2 duplication

Mietto M¹, Zambon A, Agolini E, Canessa E, Ojo B, Falzarano M, Selvatici R, Mazza T, Liorni N, Hathout Y, Novelli A, Ferlini A, Previtali S, on behalf of the Telethon Project study group

¹Unit of Medical Genetics, Department of Medical Sciences, University of Ferrara, Ferrara, Italy

718LBP ARV-027 induces peripheral polyQ-AR degradation and improves muscle strength and endurance in a murine model of SBMA

Gregory J¹, Guss V, Matchett M, Nickischer D, Kim H, Revell D, Braine C, Wu W, Wilson R, Corradi J, Jeong Y, Ganat Y, Falconieri A, Pennuto M, Sankaranarayanan S, Ojo A, Davis G, Montie H, Cacace A

¹Arvinas Operations, Inc., New Haven, United States

719LBP Phase 1 interim results evaluating the safety, tolerability, pharmacokinetics, and exploratory efficacy of salanersen (BIIB115): An antisense oligonucleotide for spinal muscular atrophy

Sansone V¹, Kotulska-Jóźwiak K, Crawford T, Ziegler A, Fradette S, Sun P, Alexanian S, Inra J, Rubel C, Czerkowicz J, Heo J, Verone-Boyle A, Wu S, Liu Y, Foster R, Burton L

¹The Nemo Clinical Center - Neurorehabilitation Unit, University of Milan, Italy

721LBP A founder variant in PLOD1 is a major cause of neonatal hypotonia and gross motor delay in Senegal, West Africa

Rodriguez Cruz P^{1,2,3}, Senghor H, Sow A, Diop A, Diagne R, Séne P, Tonda R, Alitsiou A, Urtizberea A, Ndong M, Diop A, Faye P, Beltran S, Lao O, Ndiaye M

¹Centro Nacional De Analisis Genomico (CNAG), Barcelona, Spain, ²Centre Hospitalier National Universitaire de Fann, Dakar, Senegal, ³Université Cheikh Anta Diop de Dakar, Dakar, Senegal

722LBP Disrupting YAP-driven mechanical signaling in fibro/adipogenic progenitors attenuates collagen deposition and fibrosis in Duchenne muscular dystrophy

Crosbie R¹, Kannan P, Helzer D, Marcotte G, Afsharania M, Walker J, Tamura H, Guo W, Qi D, Reynolds J, Fleser T, Malik R, Mokhonova E, Hansen K, Stearns-Reider K, Villalta S, Malfatti E, Taglietti V, Rowat A, Deeds E

¹University of California, Los Angeles, United States

723LBP Hypoglycosylation of α -dystroglycan as a secondary defect in Duchenne muscular dystrophy: Insights from the severe D2.mdx murine model

Crosbie R¹, Reynolds J, Arabatlian N, Mokhonova E, Walker J, Malik R, Maslanka M, Hansen K, Beedle A, Barton E, McNally E

¹University of California, Los Angeles, United States

724LBP Identification of disease-specific biomarker signatures in the serum of young children with COL6 and LAMA2-related dystrophies via the discovery and characterization of miRNA and proteomic profiles

Foley A¹, Moy J, Golubeva A, Hu Y, Sheehan D, Johnson K, Lek M, English J, Murray D, Bönnemann C

¹Neuromuscular and Neurogenetic Disorders of Childhood Section, NINDS, National Institutes of Health, Bethesda, United States

725LBP Sensory profile and behavioral problems in children with Duchenne muscular dystrophy and Becker muscular dystrophy

Bikmazer B¹, İyşenyürek Ş, Öztürk G, Acar Arslan E, Akbeyaz H, Karakayalı B, Keçelioğlu Binnetoğlu K, Yılmaz Z, Ünver O, Türkdoğan D

¹Marmara University Pendik Training and Research Hospital, Department of Pediatric Neurology, İstanbul, Turkey

726LBP Early Risdiplam treatment in a preterm infant with 1 SMN2 copy

Cicala G^{1,2,3}, De Sanctis R, Villa M, Capasso A, Arpaia C, Venditti R, Stanca G, Coratti G, Pane M, Mercuri E

¹Child Neurology and Psychiatry Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy, ²Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy, ³Child Neurology and Psychiatry, Catholic University, Rome, Italy

727LBP Ultrasound of the geniohyoid muscle may support diagnosis of dysphagia in SMA II and III

Akgöl Z¹, Hamzic S, Bilgin M, Alhaj Omar O, Genau S, Ebert D, Emde J, Khilan H, Akgöl A, Hahn A, Krämer-Best H

¹Department of Neurology, Justus-Liebig-University, Giessen, Germany

728LBP Dysregulation of muscle-specific protein synthesis in Facioscapulohumeral muscular dystrophy due to nucleolar inhibition of rDNA transcription induced by FRG2 lncRNAs overexpression

Salsi V, Losi F, Fosso B, Ferarini M, Pini S, Manfredi M, Vattermi G, Mongini T, Maggi L, Pesole G, Henras A, Kaufman P, McStay, **Tupler R¹**

¹University Of Modena And Reggio Emilia, Modena, Italy

730LBP LYNX Phase 2 trial results in DMD: Positive effects of Sevasemten, an investigational agent, on physical function defines dose and informs design for phase 3

Donovan J¹, McDonald C, Wong B, Phan H, Marks W, Veerapandian A, Zaidman C, Connolly A, Mathews K, Nelson S, Zygmunt A, Statland J, Leung D, Goedeker N, Yang M, Rhem B, Berthy J, Thaler L, MacDougall J, Dreghici R

¹Edgewise Therapeutics, Boulder, Colorado, United States

731LBP Diagnosis, follow-up and discontinuation of riboflavin in patients with sertraline-associated multiple acyl-CoA-dehydrogenase deficiency (MADD)

Sunebo S¹, Appelqvist H, Danielsson O

¹Department of Neurology in Linköping, and Department of Biomedical and Clinical Sciences, Linköping University, Linköping, Sweden

732LBP Preliminary analysis of safety, tolerability, and efficacy of a prophylactic sirolimus protocol for patients receiving Delandistrogene Moxeparvec-Rokl gene therapy

Soslow J¹, Neumann S, Cox R, Bunette W, Harris R, Mohammad S

¹Vanderbilt University Medical Center, Nashville, United States

733LBP Innervation induces a distinct force–frequency phenotype in 3D human neuromuscular junction tissues

Ghazizadeh H, Leahy T, Fleming J, **Estrella N¹**, Worthen C, Luerman G, Geisse N

¹Curi Bio, Seattle, United States

734LBP Expression of SGCB and safety following bidridistrogene xeboparvec treatment in patients with LGMD2E/R4: results from the EMERGENE phase 3 study

Connolly A, Lavery C, **Proud C¹**, Diaz Manera J, Claeys K, Comi G, De Bleecker J, Matesanz S, Stevenson H, Hu W, Estilo A, Haile A, Rodino-Klapac L

¹Children's Hospital of The King's Daughters, Norfolk, United States

Poster Session 4 - 735VSP-743VSP: Environmental sustainability

735VSP Re-purposing Primidone as a RIPK1 inhibitor in OPTN- and TBK1-related ALS: a case series

Durmus Tekce H¹, Gezen-AK D, Dursun E, Cakar A, Kıyan E, Gulsen Parman Y

¹Istanbul University, Istanbul Faculty of Medicine, Istanbul, Turkey

736VSP Low-cost mechanical cough assistance by rapid thoracoabdominal compression supports sustainable care

van Egmond-Fröhlich A¹

¹Klinik Favoriten, Dept. of Pediatrics, Vienna, Austria

737VSP Energy-saving measures in radiology, radiotherapy, and nuclear medicine centers: a case study of a medium-sized medical center

Gerhalter T¹

¹Medical University of Graz, Graz, Austria

738VSP Estimating the carbon footprint of delegate travel for WMS2025 and potential mitigation strategies

Gerhalter T¹

¹Medical University of Graz, Graz, Austria

739VSP Environmental footprint of patient travel: insights from a German clinical study

Schunk V¹, Gerhalter T

¹Radiology, University Hospital Erlangen, Friedrich-Alexander University Erlangen-Nürnberg, Erlangen, Germany

740VSP Addressing environmental sustainability exemplified by the DSMZ cell bank

Wang H¹, Steenpass L

¹Leibniz Institute DSMZ-German Collection of Microorganism and Cell Cultures, Department of Human and Animal Cell Lines, Braunschweig, Germany

741VSP Travel choices and carbon footprint of WMS delegates:Environmental Sustainability Committee members as a real-world example

Johari M¹, Sarparanta J, WMS Environmental Sustainability Committee

¹Harry Perkins Institute of Medical Research - UWA, Perth, Australia

742VSP Testing novel nanodelivery of AON and microdystrophin synthetic genes as therapeutic approaches for Duchenne muscular dystrophy

Montanari S¹, Mietto M, Torun T, Falzarano M, Argenziano M, Pinton P, Giorgi C, Cavalli R, Tedesco F, Ferlini A

¹Department of Medical Sciences, Unit of Medical Genetics, University of Ferrara, Ferrara, Italy

743VSP Sustainable rehabilitation for idiopathic inflammatory myopathies and beyond: developing a multicenter mobile exercise app for patients

Biliciler S¹, Nam S, Pathak S, Zhang H, Zao J, Ajwani M, Banerjee A, Niu A, Chadalavada H, Villarino J, Quach N, Chebolu P, Rajpal S, Yuan T, Alexanderson H

¹UTHealthScience Center at Houston, McGovern Medical School, Houston,TX, United States

Clinical Trial Updates: 190-240

190 Dystrophin results from the 10 mg/kg cohort of CONNECT1-EDO51 phase 2 study of PGN-EDO51 in people with the Duchenne amenable to exon 51 skipping

McMillan H¹, Chrestian N, Gonorazky H, Morcos B, Holland A, Sweeney C, Lonkar P, Garg B, Yu S, Fraser P, Foy J, Song G, Larkindale J

¹Children's Hospital of Eastern Ontario (CHEO), Ottawa, Canada

200 48-week data from the phase 2 open-label FORWARD-53 study of WVE-N531 in boys with Duchenne muscular dystrophy amenable to exon 53 skipping

Tai L¹, Servais L, Bader M, AlQurashi M, Goel V, Ghosh A, Ghosh D, Haeghele J, Hart A, Longo K, Narayanan P, Paulson D, Rheinhardt J, Singh K, Strahs A, Wagner E, Yang G, Vargeese C, Malhi S, Ingelsson E

¹Wave Life Sciences, Cambridge, United States

210 Assessment of cardiac outcomes in delandistrogene moxeparvovec clinical trials for Duchenne muscular dystrophy

Veerapandiyan A, Bourke J, Day J, McDonald C, Mendell J, Soslow J, Zaidman C, **Mason S¹**, Meng J, Vivien M, East L, Murphy A, Wandel C, Richardson J

¹Sarepta Therapeutics, Inc., Cambridge, MA, United States

220 Update on INSPIRE DUCHENNE: a phase 1/2 study of SGT-003, a next-generation microdystrophin gene therapy for Duchenne muscular dystrophy

Flanigan K¹, Shieh P, Gonorazky H, Veerapandiyan A, Gonzalez P, Harmelink M, Brooks G

¹Center for Gene Therapy, Nationwide Children's Hospital, Columbus, United States

230 Results from 15 mg/kg single dose PGN-EDODM1 cohort of FREEDOM-DM1- a phase 1 study in people with myotonic dystrophy type 1 (DM1)

Hamel J, Brisson J, **Lochmüller H¹**, Wheeler T, Sampson J, Goyal N, Johnson N, Statland J, Lilleker J, Turner C, Pfeffer G, Shoskes J, Holland A, Garg B, Song G, Lonkar P, Yu S, Fraser P, Larkindale J

¹Children's Hospital of Eastern Ontario Research Institute, Ottawa, Canada

240 DEVOTE part C results: exploring higher doses of nusinersen in nusinersen-experienced participants with spinal muscular atrophy (SMA)

Mercuri E¹, Crawford T, Finkel R, Day J, Montes J, del Mar Garcia Romero M, Sumner C, Paradis A, Sun P, Foster R, Gambino G, Littauer R, Fradette S, Farewell R

¹Catholic University, Rome, Italy