# WMS 2025 Full Programme

Version: 050925

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:

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

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

## Monday 6th October 2025

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| 08:30-19:00 | **Pre-Congress Teaching Course** (separate registration required) *-2.41/-2.42* |

## Tuesday 7th October 2025 Click here for details of Industry Symposia 1 & 2

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| 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 and first time attendee session** *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| 16:30-17:30 | **Industry Symposium 1 (withdrawn)** *Hall G* | **Industry Symposium 2** *Hall K* |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** | |

## Wednesday 8th October 2025

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

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

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| | **09:45-10:15 02INV** Beyond muscular dystrophies: roles of MD-related proteins in different organ systems<br>***Winter L***[1]<br>[1]*Medical University of Vienna, Austria*<br><br>**10:15-10:30 01O** Interferon-γ causes myogenic cell dysfunction and senescence in immune myopathies<br>***Authier F***[1]*, 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*<br>[1]*Paris Est Creteil University, France*<br><br>**10:30-10:45 02O** Brachio-cervical inflammatory myopathy: Evidence for a distinct form of inflammatory muscle disease<br>***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*<br>[1]*Ruhr University Bochum, BG University Hospital Bergmannsheil, Germany,* [2]*Charité - Universitätsmedizin Berlin, Germany* |
| 10:45-11:15 | **Morning refreshments, exhibition and poster viewing** *Hall X1* |
| 11:15-13:15 | **Topic 1: Neuromuscular diseases as multisystemic disorders** (continued) *Hall D*<br>*Moderators: John Vissing, University of Copenhagen, Denmark & Jana Haberlová, University Hospital Motol, Czechia*<br><br>**11:15-11:45 03INV** Myotonic Dystrophy Type 1: A multisystemic disorder with remarkable clinical variability and burden<br>***Braakman H***[1]<br>[1]*Amalia Children's Hospital, Radboud University Medical Center, the Netherlands*<br><br>**11:45-12:15 04INV** Spinal Muscular Atrophy: Systemic disease, focal treatment – or vice versa?<br>***Kirschner J***[1]<br>[1]*University of Freiburg, Germany*<br><br>**12:15-12:30 03O** Cognitive development in children with 5q-SMA identified by neonatal screening – four years follow-up<br>***Kölbel H***[1]*, Kopka M, Pum S, Alina S, Blaschek A, Schara-Schmidt U, Müller-Felber W, Schwartz O, Vill K*<br>[1]*University Hospital Essen, Essen, Germany*<br><br>**12:30-12:45 04O** Denervated human muscle fibers promote reinnervation via neurotrophic factor release in SMA and ALS<br>*Gokul-Nath R, Collins C, Valverde L, Lleixa C, Verdú-Díaz J, Di Lorenzo A, Llarch P, Querol L, Marini-Bettolo C, Rojas-García R,* ***Diaz-Manera J***[1]<br>[1]*Newcastle University, United Kingdom*<br><br>**12:45-13:00 05O** Redefining triple A syndrome: A multinational study of the neuromuscular phenotype in the largest genetically confirmed cohort to date<br>***Oeztuerk M***[1]*, Leonard-Louis S, Natera de Benito D, Nascimento A, Ortez C, Virić V, Stevic Z, Nelke C, Schaenzer A, Previtali S, Cehic M, Dumic K, Peric S, Roos A, Ruck T*<br>[1]*BG University Hospital Bergmannsheil, Germany*<br><br>**13:00-13:15 06O** In-depth examination of motor endplate pathology in AChR-Ab-positive myasthenia gravis<br>***Preusse C***[1]*, Meisel A, Brokamp K, Roos A, Doksani P, Hentschel A, Schuelke M, Rückert J, Pumberger M, Schömig F, Stenzel W, Hoffmann S*<br>[1]*Charité – Universitätsmedizin Berlin, Germany* |
| 13:15-14:30 | **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*<br>**01P-20P, 22P-50P, 51VP, 52P, 53VP-55VP: Acquired, inflammatory, myositis**<br>**166P-192P: DMD – imaging and outcome measures**<br>**298P-313P: Muscle MRI & new imaging techniques** |

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| | **399P-426P, 427VP-430VP: Clinical trials, access to health care and outcome measures**<br>**505P-528P, 529VP, 530P-536P, 537VP: SMA clinical**<br>**651P-677P, 678VP-688VP: DMD - treatments** | | |
| 15:30-16:00 | **Short Oral Presentations 1**<br>*K1*<br>**Updates on SMA and DMD trials**<br>**426P, 423P, 422P, 425P, 424P**<br>*Moderator: Ulrike Schara-Schmidt, University Duisburg-Essen, Germany* | **Short Oral Presentations 2**<br>*K2*<br>**Inflammatory myopathies**<br>**48P, 46P, 49P, 24P, 50P**<br>*Moderator: Georgio Tasca, Newcastle University, United Kingdom* | **Short Oral Presentations 3**<br>*-2.93/-2.94*<br>**Novel discoveries in SMA, DMD and ADSS1 myopathy**<br>**534P, 536P, 535P, 192P, 191P, 313P**<br>*Moderator: Simone Mahal, Favoriten Hospital, Austria* |
| 16:15-17:00 | **Congress Debate** *Hall D*<br>**Should families be offered n=1 treatments?**<br>*Moderators: Michelle Lorentzos, The Sydney Children's Hospitals Network, Australia & Jorge A Bevilacqua, Universidad de Chile, Chile*<br><br>**13INV** *Annemieke Aartsma-Rus, Leiden University Medical Center, the Netherlands*<br>**14INV** *Kevin Flanigan, Nationwide Children's Hospital, United States of America*<br><br>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?<br><br>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?<br><br>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-Riva (Chile).<br><br>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.<br><br>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.<br><br>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*<br><br>**118P-165P: CM – CMD**<br>**193P-217P: FSHD**<br>**314P-329P: Multidisciplinary management of neuromuscular diseases**<br>**431P-456P, 457VP-458VP: Genetics of NMD (new genes and NGS, diagnostic etc.)**<br>**486P-503P, 504VP: LGMD**<br>**551P-566P: EDMD, OPDM, autophagic, extramuscular** | | |
| 18:15-18:45 | **Short Oral Presentations 4**<br>*K1*<br>**Genetic neuromuscular disorders 1**<br>**451P, 453P, 454P, 128P, 452P, 161P**<br>*Moderator: Wolfgang Löscher, Medical University Innsbruck, Austria* | **Short Oral Presentations 5**<br>*K2*<br>**Genetic neuromuscular disorders 2**<br>**163P, 165P, 164P, 456P, 455P, 502P**<br>*Moderator: Hacer Durmus, Istanbul Faculty of Medicine, Turkey* | **Short Oral Presentations 6**<br>*-2.93/-2.94*<br>**FSHD**<br>**216P, 214P, 217P, 215P**<br>*Moderator: Carmen Paradas, Hospital Universitario Virgen del Rocío/IBiS, Spain* |
| 19:00-20:00 | **Industry Symposium 5** *Hall G* | **Industry Symposium 6** *Hall D* | |

## Thursday 9th October 2025

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| 07:00-15:00 | **Congress desk open** *Foyer D* |
| 07:45-09:15 | **Interesting Case Discussions** *Hall D*<br>*Moderators: Teerin Liewluck, Mayo Clinic Rochester, United States of America & Ana Ferreiro, Institut De Myologie, France* |
| 09:30-11:00 | **Topic 2: Multidisciplinary management of neuromuscular diseases** *Hall D*<br>*Moderators: Michela Guglieri, Newcastle University, United Kingdom & Saskia Houwen, Raboud University Medical Centre, The Netherlands*<br><br>**09:30-10:00 05INV** Psychological care in neuromuscular diseases: supporting patients with rare, progressive, and genetic diseases in the era of gene therapy<br>***Sayah S***[1]<br>[1]*Pitié-Salpêtrière Hospital, France*<br><br>**10:00-10:30 06INV** The expanding role of rehabilitation in neuromuscular diseases: from care to translational research<br>***Gagnon C***[1]<br>[1]*Université de Sherbroooke, Canada*<br><br>**10:30-10:45 07O** Neuropsychiatric burden of Dystrophinopathies: A multi-centre European study<br>***Kolesnik A***[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*<br>[1]*Dubowitz Neuromuscular Centre, UCL Great Ormond Street, Institute of Child Health, United Kingdom*<br><br>**10:45-11:00 08O** Insights into the use of the capability approach in multidisciplinary rehabilitation for persons with neuromuscular diseases<br>***Pijpers E***[1]*, Bloemen B, Oortwijn W, Van Engelen B, Van der Wilt G, Groothuis J, Cup E*<br>[1]*Radboudumc, Netherlands* |
| 11:00-11:30 | **Morning refreshments, exhibition and poster viewing** *Hall X1* |
| 11:30-13:30 | **Topic 2: Multidisciplinary management of neuromuscular diseases** (continued) *Hall D*<br>*Moderators: Nicol Voermans, Radboud University Medical Centre, the Netherlands & Payam Mohassel, Johns Hopkins University, United States of America*<br><br>**11:30-12:00 07INV** Coordination of care for people with neuromuscular diseases: the sum is greater than the individual parts<br>***Quinlivan R***[1]*, Desikan M, Kaur S, Astin R, Savvatis K, Emmanuel A, Bouquillon L, Sarcozy A, Hanna M*<br>[1]*University College London, United Kingdom*<br><br>**12:00-12:30 08INV** Palliative Care for Adults with Neuromuscular Diseases- what do we know and where next?<br>***Willis D***[1]<br>[1]*The Severn Hospice, United Kingdom*<br><br>**12:30-12:45 09O** Optimising transition and advanced care planning for young people with neuromuscular disorders<br>***Irving K***[1]*, Howard M, Culnane E*<br>[1]*Royal Children's Hospital Melbourne, Australia*<br><br>**12:45-13:00 10O** Sexual and pelvic floor function in women with Muscular Dystrophy<br>***Hamel J***[1]*, Dekdebrun J, Lopa S, Thornburg L, Anto-Ocrah M*<br>[1]*University of Rochester, United States of America*<br><br>**13:00-13:15 11O** Heart transplantation in males with dystrophinopathy: An advanced cardiac therapies improving outcomes network (ACTION) Dystrophinopathy registry analysis<br>***Hayes E***[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, Radel L, Cunningham T*<br>[1]*Nationwide Children's Hospital, United States of America* |

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| | **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<br>***Nuredini A***[1] *(u/b), Socha M, Piorecki 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, Schoser B, Polańska J, Tupler R*<br>[1]*University of Modena and Reggio Emilia, Italy* |
| 13:30-14:45 | **Lunch, exhibition and poster viewing** *Hall X1* |
| 13:45-14:45 | **WMS 2025 Early to Mid-Career Peer Networking Session** (separate registration required) *Hall G*<br>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.<br><br>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) |

## Friday 10th October 2025 Click here for full details of Poster Sessions 3 & 4 and Short Oral Presentations 7-12

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| 08:00-18:00 | **Congress desk open** *Foyer D* |
| 08:30-10:15 | **Topic 3: Advances in therapies and drug development** *Hall D*<br>*Supported by the ICNMD*<br>*Moderators: Elie Naddaf, the Mayo Clinic, United States of America & Nalini Atchayaram, National Institute of Mental Health and Neuro Sciences, India*<br><br>**08:30-09:00 09INV** Advances in therapies and drug development: improving AAV Vectors for Neuromuscular Disease indications<br>***Spencer M***[1]<br>[1]*Center for Duchenne Muscular Dystrophy at UCLA, United States of America*<br><br>**09:00-09:30 10INV** AAV transduction optimization for NMD<br>***Adjali O***[1]<br>[1]*Université de Nantes, France*<br><br>**09:30-09:45 13O** Identifying naturally occurring adeno-associated virus serotypes that transduce mouse skeletal muscles fibro-adipogenic progenitors in vivo<br>***Guirguis F***[1,2]*, Bolduc V, Brull A, Liu H, Chiorini J, Zabaleta N, Vandenberghe L, Zhou H, Muntoni F, Bönnemann C*[1]<br>[1]*National Institutes of Health, United States of America* [2]*The Dubowitz Neuromuscular Centre, University College London, United Kingdom*<br><br>**09:45-10:00 14O** Regulatory T cells suppress dystrophin immunity<br>***Farahat P***[1]***,*** *Villalta S*<br>[1]*University Of California Irvine, United States of America*<br><br>**10:00-10:15 15O** Towards a human multi-organoid platform to develop and assess specificity, toxicity and efficacy of neuromuscular gene therapies<br>*Dastidar S, Torun T, Raghu Chakravarthy P Zouhair M, Duan Z, Chen P, Ferrari T, Fereira M, Lionello V, Ralu M, Sarcar S, Amendola M,* ***Tedesco F***[1,2,3]<br>[1]*University College London, United Kingdom,* [2]*The Francis Crick Institute, United Kingdom, 3Dubowitz Neuromuscular Centre, Great Ormond Street Hospital for Children & UCL Great Ormond Street Institute of Child Health, United Kingdom* |
| 10:15-10:45 | **Morning refreshments, exhibition and poster viewing** *Hall X1* |

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| 10:45-12:30 | **Topic 3: Advances in therapies and drug development** (continued) *Hall D*<br>*Moderators: Ana Tesi Rocha, Stanford University, United States of America & Vishnu Venugopalan Yamuna, All India Institute of Medical Sciences, India*<br>**10:45-11:15 11INV** Leveraging the muscle fusogens for gene delivery<br>***Millay D***[1]<br>[1]*Cincinnati Children's Hospital, United States of America*<br><br>**11:15-11:45 12INV** Innovation in nanoparticles as drug delivery vehicles for the treatment of muscle disorders<br>***Lau H***[1]<br>[1]*Yale School of Medicine, United States of America*<br><br>**11:45-12:00 16O** Calcitriol ameliorates myotonia in 3D DM1 skeletal muscle models via a MBNL1-independent mechanism<br>***Fernández Costa J***[1], *Fernández-Garibay X, Maria Sabater-Arcís M, Tejedera-Villafranca A, Núñez-Manchón J, Artero R, Suelves M, Nogales-Gadea G, Ramón-Azcón J*<br>[1]*Institute for Bioengineering of Catalonia (IBEC), Barcelona Institute of Science and Technology (BIST), Spain*<br>**12:00-12:15 17O** Collagen type VI regulates TGFβ bioavailability in skeletal muscle in mice<br>***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, Bonnemann C*<br>[1]*Johns Hopkins University School of Medicine, United States of America,* [2]*National Institutes of Health, United States of America*<br><br>**12:15-12:30 18O** Translation of GGC repeat expansions into a toxic polyG protein in oculopharyngodistal myopathy 2<br>***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*<br>[1]*Huashan Hospital, Fudan University, China,* [2]*National Center for Neurological Disorders (NCND), China* |
| 12:30-14:00 | **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*<br><br>**56P-89P: Advances in therapies and drug development**<br>**104P-116P, 117VP: Cell insights, muscle homeostasis**<br>**251P-296P, 297VP: Dystrophinopathies (animals models, biomarkers, brain, genetics)**<br>**366P-398P: Myotonic dystrophy**<br>**538P-550P: Registries, networks and care of NMD**<br>**589P-609P: SMA outcome measures and registries**<br>**689P, 690VP, 691P-702P: Distal myopathies, MFM** |
| 15:15-15:45 | **Short Oral Presentations 7**<br>*K1*<br>**Myotonic dystrophy and distal myopathies**<br>**689P, 395P -398P**<br>*Moderator: Benedikt Schoser, Friedrich-Baur-Institut LMU München, Germany*<br><br>**Short Oral Presentations 8**<br>*K2*<br>**Advances in therapies and SMA**<br>**609P, 87P, 88P, 116P, 86P**<br>*Moderator: Wenhua Zhu, Huashan Hospital, Fudan University, China*<br><br>**Short Oral Presentations 9**<br>*-2.93/-2.94*<br>**Insights into muscular dystrophy**<br>**288P, 89P, 270P, 295P**<br>*Moderator: Sophelia Chan, The University of Hong Kong, China* |
| 15:45-16:45 | **Poster Session 4** *Hall X1*<br><br>**90P-103P: ALS/neuropathy**<br>**218P-250P: Neuromuscular diseases as multisystemic disorders**<br>**330P-365P: Metabolic and mitochondrial myopathies**<br>**459P-484P, 485VP: Myasthenia Gravis, NMJ1-2, periodic paralysis**<br>**567P-569P, 571P-574P: Pompe disease**<br>**575P-588P: SMA therapies**<br>**610P-620P, 621VP, 622P-648P, 649VP-650VP: DMD - clinical care and cases reports, BMD** |

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| 16:45-17:15 | **Short Oral Presentations 10**<br>*K1*<br>**Multisystemic disorders and disease mechanisms**<br>**647P, 648P, 246P -248P**<br>*Moderator: Mridul Johari, The Harry Perkins Institute of Medical Research, Australia* | **Short Oral Presentations 11**<br>*K2*<br>**Metabolic and mitochondrial disorders**<br>**361P - 365P**<br>*Moderator: Suur Biliciler, Ut Health Science Center at Houston, Mcgovern Medical School, United States of America* | **Short Oral Presentations 12**<br>*-2.93/-2.94*<br>**Congenital muscle diseases**<br>**471P, 249P, 484P, 250P**<br>*Moderator: Hernán Gonorazky, The Hospital for Sick Children, Canada* |
| 19:15-01:00 | **Networking Dinner** (separate registration required) | | |

## Saturday 11th October 2025 Click here for full details of Clinical Trial Updates

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| 07:30 | **Congress desk open** *Foyer D* |
| 07:30-09:00 | **Clinical Trial Updates** *Hall D*<br>*Moderators: Ann Agnes Mathew, Bangalore Baptist Hospital, India & Andreas Ziegler, University Hospital Heidelberg, Germany*<br>**07:30-07:45 19O**<br>**07:45-08:00 20O**<br>**08:00-08:15 21O**<br>**08:15-08:30 22O**<br>**08:30-08:45 23O**<br>**08:45-09:00 24O** |
| 09:00-09:15 | **Comfort break** |
| 09:15-09:45 | **Victor Dubowitz Lecture** *Hall D*<br>Myology in Asia<br>*Ichizo Nishino, National Institute of Neuroscience, NCNP, Japan*<br>*Moderators: Volker Straub, Newcastle University, United Kingdom & Gina Ravenscroft The Harry Perkins Institute of Medical Research, Australia* |
| 09:45-11:15 | **Poster Highlights**<br>*Moderators: Matthias Baumann, Medical University Innsbruck, Austria & Rotem Orbach, National Institutes of Health, United States of America*<br>**09:45-10:00 25O**<br>**10:00-10:15 26O**<br>**10:15-10:30 27O**<br>**10:30-10:45 28O**<br>**10:45-11:00 29O**<br>**11:00-11:15 30O** |
| 11:15-11:45 | **Morning refreshments, exhibition and poster viewing** *Hall X1* |
| 11:45-13:15 | **Late Breaking News** *Hall D*<br>*Moderators: Hanns Lochmuller, Children's Hospital of Eastern Ontario Institute, Canada & Sandra Donkervoort, National Institutes of Health, United States of America*<br>**11:45-12:00 01LBO**<br>**12:00-12:15 02LBO**<br>**12:15-12:30 03LBO**<br>**12:30-12:45 04LBO**<br>**12:45-13:00 05LBO**<br>**13:00-13:15 06LBO** |
| 13:15-13:45 | **Prize Giving Ceremony** *Hall D*<br>*Moderator: Marco Savarese, University of Helsinki, Finland*<br>**Introduction to the WMS 2026 Congress, Hiroshima, Japan**<br>**Handover of the WMS flag and close of the Congress** |
| 13:45-14:30 | **Homeward lunch** |

# WMS 2025 Congress programme appendix

Version: 05/09/25

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 in the bottom corner of each page.

## Tuesday 7th October 2025

To leave the appendix and return to the main programme, please click the 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 & Eroka 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 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 Syakuron***[1], *Hakim M, Budikayanti A, Wiratman W, Yanuar Safri A, Fadli N, Ridski Harsono A, Octaviana F, Ari Indrawati L*
[1]*Department of Neurology, Faculty of Medicine, Universitas Indonesia, Jakarta, Indonesia*

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

***Farahat P***[1], *Phillips C, Hernandez I, Wencel M, Mozaffar T, Villalta A*
[1]*University 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*
[1]*Department of Clinical Neuroscience and Therapeutics, Graduate School of Biomedical and Health Science, Hiroshima University, Hiroshima, Japan,* [2]*Department 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*
[1]*Institute of Neuropathology, University Hospital RWTH Aachen, Aachen, Germany*

**05P Consistent clinical features of anti-Mi-2 myopathy: a Korean retrospective cohort study**
***Kim S***[1]*, Choi Y, Choi Y, Kim W, Park H*
[1]*Department of Neurology, Gangnam Severance Hospital, Younsei 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*
[1]*Huashan Hospital, Fudan University, Shanghai, China,* [2]*National 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*
[1]*Huashan Hospital, Fudan University, Shanghai, China,* [2]*National Center for Neurological Disorders, Shanghai, China*

**08P Comparative evaluation of clinical outcome measures in Inclusion body myositis based on NT5c1A antibody serology status**
***Wencel M***[1]*, Goyal N Carbunar O Freimer M,Dimachkie M, Quinn C, Lloyd T, Mohassel P, Weihl C, Shaibani A, Wang L, Chahin N, Amato ,A, Wicklund M, Dixon S, Shieh P, Herbelin L, Barohn R, Tawil R, Ciafaloni E*
[1]*University Of California, Irvine, Orange, United States*

**09P Case series of immune checkpoint inhibitor-induced overlap syndrome: myasthenia, myositis, and miocarditis in a tertiary center**
***Gomez Fernandez F***[1]*, Cabrera Serrano M, Rojas-Marcos I, Lopera Rodriguez I, Rivas Infante E, Montes Latorre E, Paradas López C*
[1]*Hospital Universitario Virgen Del Rocio: Serv. Neurologia, Sevilla, Spain*

**10P Anti-synthetase syndrome—finding a pearl in an oyster: a series of 12 cases**
***Bhagat S***[1]*, Jassal B, Vishnu V, Sharma M, Bhadu D*
[1]*All 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 B***[1]*, Dhall A, Vishnu V, Bhadu D, Suri V, Sharma M*
[1]*Neuropathology 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, Tejedera-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*
[1]*IDIBAPS - HCB - University of Barcelona, Barcelona, Spain,* [2]*CIBERER, Madrid, Spain*

**13P Ulviprubart 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*
[1]*Perron Institute, QEII Medical Centre, Nedlands, Western Australia, Australia*

**14P Clinicopathological characteristics of chronic cases of immune-mediated necrotizing myopathy**
***Komaki S***[1]*, Kubota A, Maeda M, Shimizu J, Yamanaka A, Nishino I, Toda T*
[1]*Department 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*
[1]*Department of Neurology, the Second Hospital of Hebei Medical University, Shijiazhuang, China,* [2]*Department 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 I***[1]*, Wencel M, Goyal N, Carbunar O, Freimer M, Dimachkie M, Quinn C, Lloyd T, Mohassel P, Weihl C, Shaibani A, Wang L, Chahin N, Amato A, Wicklund M, Dixon S, Shieh P, Herbelin L, Barohn R, Mozaffar T*
[1]*University 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 I***[1], *Wencel M, Goyal N, Carbunar O, Freimer M, Dimachkie M, Quinn C, Lloyd T, Mohassel P, Weihl C, Shaibani A, Wang L, Chahin N, Amato A, Wicklund M, Dixon S, Shieh P, Herbelin L, Barohn R, Mozaffar T*
[1]*University 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*
[1]*Neuromuscular Unit, Barcelona, Spain,* [2]*Department of Neurology, Barcelona, Spain,* [3]*Hospital 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 S***[1], *George J, Poyuran R, Narasimhaiah D*
[1]*Sree 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 H***[1], *Le S*
[1]*University 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*
[1]*Department of Pediatric Neurology, University Duisburg-Essen, Essen, Germany* [2]*Department of Neurology, University Hospital Düsseldorf, Düsseldorf, Germany,* [3]*Heimer Institute for Muscle Research, Bochum, Germany,* [4]*Brain and Mind Research Institute, Ottawa, Canada*

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

***Holzer M***[1], *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, Preuße C, Schneider U, Krusche M*
[1]*University 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 M***[1], *Dittmayer C, Goebel H, Dressel A, Stenzel W*
[1]*University 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 M***[1], *Kornblum C, Reimann J*
[1]*Department of Neuromuscular Diseases, Center for Neurology, University Hospital of Bonn, Bonn, Germany*

**26P Therapeutic outcomes and prognostic determinants in juvenile myasthenia gravis**

***Puppala S***[1], *Tandon V, Sundaram S, Nair S*
[1]*Sree 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 A***[1], *Shankar N, Traub R, Mehrabyan A*
[1]*University 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, Heyraut-Blanchet C, Plante-Bordeneuve V, Lefeuvre C, Zaroui A, Damy T, Severa G, Malfatti E, Souvannanorath S*
[1]*Paris Est Creteil University, Créteil, France,* [2]*Henri 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*
[1]*Paris Est Creteil University, Créteil, France,* [2]*Henri Mondor University Hospital, Créteil, France*

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

*Indrawati L,* ***Putri M***[1], *Isaac W, Shafitha N, Simatupang S, Safri A, Fadli N, Harsono A, Wiratman W, Budikayanti A, Octaviana F, Hakim M*
[1]*Department 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*
[1]*Department of Neurology, Mayo Clinic, Rochester, Minnesota, United States,* [2]*Division of Neurology, Department of Medicine, Siriraj Hospital, Mahidol University, Bangkok, Thailand*

**32P Perimysial inflammation with abundant macrophages**

***Puusepp S***[1], *Kiseleva N, Kannel K, Stenzel W, Goebel H*
[1]*Pathology 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 D***[1], *Preusse C, Lichtenberg S, Eigenfeldt L, Koch-Hölsken K, Umathum V, Herrmann A, Vinnenberg L, Schänzer A, Pinal-Fernandez I, Mammen A, Fassbender B, Seifert M, Meuth S, Stenzel W, Ruck T*
[1]*Department 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 N***[1]
[1]*Aiims 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*
[1]*Department of Neurology, Dr Cipto Mangunkusumo Hospital, Jakarta, Indonesia,* [2]*Faculty 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*
[1]*Neuromuscular Morphology Unit, Institut De Myologie, Paris, France,* [2]*Neuropathology 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 T***[1], *Davis S, Raaphorst J, Bos E, O`Shaughnessy R, Aronica E, Kessler B, Fischer R, Raz V*
[1]*Leiden University Medical Center, Leiden, Netherlands*

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

***Nishimori Y***[1], *Eura N, Sugie K*
[1]*Department 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], *Luijcx S, Ardiç B, Stachurska P, van der Kooi A, Aronica E, Raaphorst J, Ottenheijm C*
[1]*Amsterdam UMC - Location Vrije Universiteit, Physiology, Amsterdam, Netherlands,* [2]*Amsterdam Cardiovascular Sciences, Amsterdam, Netherlands,* [3]*Amsterdam 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 M***[1], *Kawahara T, Kubota A*[1] *Shimizu J, Toda T*
[1]*The 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 N***[1], *Nishimori Y, Yamanaka A, Shiota T, Tanaka H, Ohashi T, Shimizu H, Yamaoka M, Yamada N, Iguchi N, Tanaka A, Sugata M, Nanaura H, Kiriyama T, Sugie K*
[1]*Department of Neurology, Nara Medical University, Kashihara, Japan*

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

***Yoon M***[1], *Lee S, Yoo Y*
[1]*Department 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 A***[1], *Merlonghi G, Forcina F, Fionda L, Leonardi L, Costanzo R, Tufano L, Rossini E, Marando D, Vera V, Antonini G, Morino S, Garibaldi M*
[1]*Neuromuscular 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 S***[1], *Heezen L, Mahfouz A, Kan H, Badrising U, Spitali P*
[1]*Leiden University Medical Center, Leiden, Netherlands*

**45P GRO chemokines-induced neutrophil extracellular trap CITH3 as a new biomarker related to myasthenia gravis severity**
***Zhong H***[1], *Zhang W, He D, Chen R, Jin L, Song J, Yan C, Huan X, Chen Y, Li X, Zhao C, Luo S*
[1]*Huashan Hospital, Fudan University, Shanghai, China*

**46P The endothelial DLL4-muscular NOTCH2 axis drives muscle atrophy in dermatomyositis: insights from single-nucleus transcriptomics**
***Hayashi S***[1], *Kato M, Noguchi S, Nishino I*
[1]*Department 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 N***[1], *Eisenlohr P, Baum O, Stenzel W, Kleefeld F*
[1]*Klinik 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 F***[1], *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*

**49P Aberrant expression of THE1B endogenous retrovirus fusion transcripts in sarcoid myopathy**
***Funaguma S***[1], *Iida A, Saito Y, Tanboon J, De Los Reyes F, Sonehara K, Goto Y, Okada Y, Hayashi S, Nishino I*
[1]*National 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 E***[1], *Ben Hmid A, Ben Ahmed M*
[1]*The Muscle Lab, Iris Medical Centre, Hedi Nouira Avenue, Ennasr2, 2001 Ariana., Tunisia*

**51VP Mitochondrial pathology in Mi-2 beta dermatomyositis with lymphocyte fibre invasion**
***Cotta A***[1], *Carvalho E, da-Cunha-Junior A, Nunes-Neves S, Cordeiro B, De-Alencar-Hornsby A, Cauhi A, Coutinho-Passos S, Valicek J*
[1]*The Sarah Network of Rehabilitation Hospitals, Belo Horizonte, Brazil*

**52P Clinical and radiological profile of a cohort of anti-HMGCR myopathy**
***Vengalil S***[1], *Ahmed S, Nashi S, Menon D, Mahadevan A, Nalini A*
[1]*National 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*
[1]*Department Of Adult Neurology, Habib Bourguiba Hospital, Sfax, Tunisia,* [2]*Research Laboratory LR12SP19, Habib Bourguiba University Hospital, University of Sfax, Tunisia*

**54VP Distinct histopathological features of juvenile idiopathic inflammatory myopathies: a quantitative comparative study**
***Hakim F***[1], *Sakka S, Snoussi M, Boudawara O, Moalla K, Bouattour N, Daoud S, Charfi N, Sellami L, Marzouk S, Boudawara T, Damak M*
[1]*Department 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*
[1]*Department Of Adult Neurology, Habib Bourguiba Hospital, Sfax, Tunisia,* [2]*Research 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 G***[1], *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.*
[1]*Catholic 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 A***[1], *Powell D, Carlton D, Chen E, Gallington K, Bacci D, Gelhorn D, Cho M, Dashiell-Aje D*
[1]*Biomarin Pharmaceutical, Inc., San Rafael, United States*

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

***Stimpson G***[1], *Zambon A, Laverty A, Ridout D, Brusa C, Wolfe A, Milev E, O'Reilly E, Manzur A, Sarkozy A, Baranello G, Muntoni F*
[1]*Dubowitz 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 L***[1], *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*
[1]*Genethon, EVRY, France*

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

***Taivassalo T***[1], *Paul M, Duong T, Lott D, Forbes S, Shih R, Leon Astudillo C, Sullivan R, Sweeney L*
[1]*University Of Florida, Gainesville, United States*

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

***Leonardis L***[1,2], *Matajurc N, Teran N*
[1]*University Medical Centre Ljubljana, Ljubljana, Slovenia,* [2]*University of Ljubljana, Ljubljana, Slovenia*

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

***Cosyns M***[1], *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*
[1]*Sciensano, 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 F***[1], *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*
[1]*University 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 F***[1], *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*
[1]*University 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 A***[1], *Rodino-Klapac L*
[1]*Roche 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 M***[1], *Parinello G, Vrščaj E, Bisson C, Cluzeau C, Daron A, University Children's Hospital L, Osredkar D, Strijbos P, Eggenspieler D, Servais L*
[1]*NMRC, Citadelle, Liège, Belgium*

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

***Poleur M***[1], *Parinello G, Vrščaj E, Dolanska A, Nowakowski P, Anghelescu C, Szabo L, Leanca M, Mirea A, Kodsy 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*
[1]*NMRC, 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*
[1]*UC Davis Health, Sacramento, United States,* [2]*CINRG, Pittsburgh, United States*

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

***Fleerakkers E***[1,2,3], *Broos J, Hoek R, Krom Y, Michaels M, Kan H, van Duyvenvoorde H, van der Holst M, Niks E*
[1]*Department of Neurology, Leiden University Medical Center, Leiden, Netherlands,* [2]*Department of Orthopaedics, Rehabilitation & Physical therapy, Leiden, Netherlands,* [3]*Duchenne 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 C***[1], *Servais L, Eggenspieler D, Henricson E, Strijbos P, Sidiropoulos P, Finkel R, McDonald C*
[1]*UC Davis, Sacramento, United States*

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

***Nakayama T***[1], *Kimizuka J, Fumiya I*
[1]*Yokohama 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 K***[1], *Goldsbury C, Horn P, Vilaisaktipakorn P, Reebals L, Zygmunt A, Tian C*
[1]*Cincinnati 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 P***[1], *Martone J, Stark M, Nascene D, Nordseth T, Kang P*
[1]*University 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*
[1]*Pediatric Neurology, Università Cattolica del Sacro Cuore, Rome, Italy,* [2]*Centro 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*
[1]*Cincinnati Children's Hospital Medical Center, Cincinnati, United States,* [2]*University 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 M***[1], *Freimark J, Billmyer E, Hoek R, Krom Y, Fleerakkers E, van der Holst M, Helleman K, van Weperen P, Straathof C, Marden J, Chen Z, Zhang W, Signorovitch J, Ward S, Niks E*
[1]*Department of Neurology, Leiden University Medical Center, Leiden, Netherlands*

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

***Michaëls M***[1,2], *Prins A, Fleerakkers E, van der Holst M, van Zwet E, Kan H, Niks E*
[1]*Department of Neurology, Leiden University Medical Center, Leiden, Netherlands,* [2]*Duchenne 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 P***[1], *Prins A, Fleerakkers E, van der Holst M, van Zwet E, Kan H, Niks E*
[1]*Department 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]
[1]*University 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*
[1]*Genethon, 1bis rue de l'international, Evry, France*

**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***[1], *Sanz M, BUSCARA L, Blaie S, Thibaut L, Sagot M, Riviere A, Creoff E, Lelait M, Valent A, Perret G, Braun S, Muntoni F*
[1]*Genethon, 1bis rue de l'international, Evry, France*

## Poster Session 1 – 298P-313P: Muscle MRI & new imaging techniques

**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, Vílchez R, Vílchez J, Sevilla T, Muelas N*

[1]*Neuromuscular Unit, Department of Neurology, Hospital Politècnic i Universitari La Fe, Valencia, Spain,* [2]*Neuromuscular 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 C***[1], *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*

[1]*John 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*

[1]*Irccs Stella Maris Foundation, Pisa, Italy,* [2]*University of Pisa, Italy*

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

***Duong T***[1], *Anna Yao M, Yoseph Hailu R, Vogt-Domke S, Day J, Hageman N*

[1]*Stanford University, Palo Alto, United States*

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

***Karlsson M***[1], *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*

[1]*AMRA Medical AB, Linköping, Sweden*

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

***Leitermann V***[1], *Cross S, DuPreez H, Whiteley-Jones H, Millner T, Radunovic A*

[1]*Royal 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*

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

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

***Mayer A***[1], *Starke A, Türk M, Uder M, Dörfler A, Nagel A, Gazzerro E, Gerhalter T*

[1]*Universitätsklinikum Erlangen, Erlangen, Germany*

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

***Nguyen-Younossi N***[1], *Vernikouskaya I, Müller H, Chadraabal K, Gadelkareem M, Rasche V, Kassubek J, Rosenbohm A*

[1]*Department 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 T***[1], *Paprad T, Sudhakar S, Hawkins C, Baruteau J, Gonorazky H, Yoon G*

[1]*The 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 N***[1],*Kass K, Schmidt K, Lyu Z, Teitsdóttir B, Sheikh A, Nielsen E, Hansen A, Wilms G, Khawajazada T, Witting N, Ladefoged C, Vissing J*

[1]*Copenhagen 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]

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

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

***Slipsager A***[1], *Stemmerik M, Ankjær P, Teitsdóttir B, Vissing J*

[1]*Copenhagen Neuromuscular Center, Rigshospitalet, Copenhagen, Denmark*

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

***Socha M***[1], *Piorecki L, Verdú-Díaz J, Diaz-Manera J, Straub V, Tupler R, Polanska J*
[1]*Silesian 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 O***[1], *Aksoy A*
[1]*Division 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 Y***[1], *Yoshioka W, Eura N, Hayashi S, Noguchi S, Nishino I*
[1]*Department 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**

***Ankjær P***[1], *Stemmerik M, Teitsdòttir 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**

***Brenninkmeijer R***[1], *Biziaki Ansanello N, Mattiello-Sverzut A, Bartels B*
[1]*Child Development and Exercise Center, Wilhelmina Children's Hospital, University Medical Center Utrecht, Utrecht, Netherlands*

**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*
[1]*Department 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,* [2]*Department 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 T***[1], *Wilcox R, de Monts C, Christle J, Tang W, Khonde S, Wheeler M, Ashley E, Birkmeier M, Meyer J*
[1]*Stanford 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*
[1]*New Giza University, Cairo, Egypt,* [2]*Myo-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]
[1]*Neuromyology Department, Neuromuscular Reference Center, Institute of Myology, Pitié-Salpêtrière University Hospital, Paris, France,* [2]*UMR974 Myology Research Center, Inserm/Sorbonne Université, Paris, France*

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

***Hogrel J***[1], *Marques T, Santmarty P, Decostre V, Drummond E, Muni Lofra R, Straub V, DT4RD Consortium T*
[1]*Institute of Myology, Paris, France*

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

***Hoogveld L***[1], *Pelsma M, Meijler D, Ijspeert J, Braakman H, Houwen S*
[1]*Department 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 A***[1], *Gosk-Tomek M, Schwerin-Nagel A, Schmidt I, Mahal S, Lilja S, Paquay A, Rosensteiner B, Bernert G, Bittner R, Schmidt W*
[1]*Klinik Favoriten, Vienna, Austria, Vienna, Austria*

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

***Iterbeke L***[1], *Huysmans L, Bamps K, Peeters R, Goosens V, Maes F, Dupont P, Claeys K*
[1]*Laboratory 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 T***[1], *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*
[1]*NHO 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 E***[1], *Proud C, Darras B, Hayes L, Oskoui M, Visootsak J, Williams G, Lee A, Ilic A, Yang L, van der Pol W*
[1]*Fondazione 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 R***[1], *Dubow J, Wells C, Lambert P, Rudnicki M, Gleeson F*
[1]*Satellos Bioscience Inc., Toronto, Canada*

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

***Oude Elberink I***[1], *Roes K, Van der Woude D, Ruyten T, Mulder S, Van Eijk R, Van der Pol L, Bartels B*
[1]*Child 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 M***[1], *Varghese V, Rajadurai I, Mathew A*
[1]*Bangalore Baptist Hospital, Bengaluru, India*

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

***Poleur M***[1], *Delstanche S, Parinello G, Bisson C, Cluzeau C, Dubois C, Duclos M, Benmhammed N, Eggenspieler D, Servais L*
[1]*NMRC, Citadelle, Liège, Belgium*

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

***Proud C***[1], *Vũ D, Wilmshurst J, Sanmaneechai O, Gulati S, Xiong H, Ceja Moreno H, Tay S, Thong M, Born A, Banzzatto Ortega A, Jong Y, Al-Muhaizea M, Lee A, Visootsak J, Tauscher-Wisniewski S, Alecu I, Parlikar R, Finkel* R*
[1]*Children'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 S***[1], *Putri M, Issac W, Shafitha N, Wiratman W, Budikayanti A, Octaviana F, Safri A, Fadli N, Harsono A, Hakim M*
[1]*Department of Neurology, Dr Cipto Mangunkusumo Hospital, Jakarta, Indonesia*

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

***Suetterlin K***[1], *Devine H, Brito Da Silva A, Smith S, Hillman S, Rao K, Brownstein C, Turnbull D, Bulea T*
[1]*Newcastle University, Newcastle Upon Tyne, United Kingdom*

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

***Tawara N***[1], *Shibata Y, Hara K, Nagatoshi C, Fujimoto A, Ishizaki M, Kurisaki R, Nishida Y, Maeda Y, Ueyama H*
[1]*Department 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 S***[1], *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*
[1]*Radboudumc, Nijmegen, Netherlands*

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

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

***Willis T***[1], *Sakanaka T, Butler P, Kulshrestha R, Loram I*

[1]*Robert Jones and Agnes Hunt Hospital, Oswestry, United Kingdom,* [2]*Chester 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 N***[1], *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*

[1]*Lurie 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-Bełdzińska M***[1], *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*

[1]*Department of Developmental Neurology, Chair of Neurology, Medical University of Gdańsk, Gdańsk, Poland*

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

***McDonald C***[1], *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*

[1]*UC Davis Health, Sacramento, United States*

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

***Pakola S***[1], *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*

[1]*REGENXBIO, 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 SAPPHIRE phase 3 trial**

***Seferian A***[1,2,3], *Crawford T Servais L, Mercuri E, Krueger J, Kölbel H, Cances C, Kuntz N, Finkel R, Yao B, Rossello J, Tirucherai G, Zhao G, Song G, Marantz J, Darras B*

[1]*Institut de Myologie, I-Motion Clinical Trials Platform, Paris, France,* [2]*APHP, Pediatric Neurology Department, Hôpital Armand Trousseau, Centre Référent pour les Maladies Neuromusculaires Nord/Est/Ile de France, Paris, France,* [3]*APHP, Pediatric Neurology and ICU Department, Université Paris Saclay, DMU Santé de l'Enfant et de l'Adolescent, Hôpital Raymond Poincaré, Garches, France*

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

***Banavara Shyamprasad S***[1], *Satyam P, Balwani H, Herle S, Khandekar G, Thomas N, Mathew A*

[1]*Synapse 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 V***[1]

[1]*Institute of Myology, Neuromuscular Investigation Centre, Paris, France*

**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 I***[1], *Rouy D, Rendo P*

[1]*Sanofi, Boston, United States*

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

***Scavina M***[1], *Banks K, Cappa R, Chambers C*

[1]*Parent Project Muscular Dystrophy, Washington, DC, United States*

## Poster Session 1 – 505P-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 A***[1], *Saraceni V, Onofri M, Briganti M, Sancricca C, Frongia A*

[1]*Fondazione 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*

[1]*University College London Great Ormond Street Institute of Child Health, London, United Kingdom,* [2]*Dubowitz Neuromuscular Centre Great Ormond Street Hospital for Children, London, United Kingdom*

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

***Carman K***[1], *Aksakal F*

[1]*Eskisehir Osmangazi University, Eskisehir, Turkey*

**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*

[1]*Great Ormond Street Hospital, London, United Kingdom,* [2]*Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom*

**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*

[1]*Great Ormond Street Hospital, London, United Kingdom,* [2]*Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom*

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

***Dangouloff T***[1], *Chantraine F, Hennuy N, Rigo V, Tribolet S, Vanden Brande L, Servais L*

[1]*Neuromuscular 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 C***[1], *Dunaway Young S, Vogt-Domke S, Yatsu L, Yoseph Hailu R, Salvatore S, Tang W, Britts C, Parker D, Day J, Duong T*

[1]*Stanford University, Department of Neurology, Palo Alto, United States*

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

***Garcia Uzquiano R***[1], *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.*

[1]*Sant 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 K***[1], *Hahn A, Kotulska-Jozwiak K, Keto E, Li Y, Guittari C*

[1]*PDMA 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*

[1]*Departments of Pediatrics and Neurology & Neurosurgery, Montreal Children's Hospital, McGill University, Montreal, Canada,* [2]*Research Institute, McGill University Health Centre, Montreal, Canada*

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

***Hallal S***[1], *sonia N, Lyèce Y*

[1]*Laboratory of biochemistry-Chu Mustapha, algiers, Algeria*

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

***Jacquier D***[1], *Klein A, Stettner G, Broser P, Fluss J, Enzmann C, Goeggel Simonetti B9, Sluka S, Baumgartner M, Hersberger M, Tscherter A*

[1]*Pediatric 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*

[1]*John Walton Muscular Dystrophy Research Centre, Newcastle Upon Tyne, United Kingdom,* [2]*Newcastle Hospitals NHS Foundation Trust, Newcastle Upon Tyne, United Kingdom*

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

***Lee H***[1]

[1]*Yongin 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 R***[1], *Dosi C, Parravicini S, Scopelliti M, Arnoldi M, Zanin R, Schinlder A, Gandolfi S, SFERA working group*

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

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

***Masson R***[1], *Baranello G, Servais L, Darras B, Day J, Deconinck N, Farrar M, Finkel R, Bertini E, Kirschner J, Mazurkiewicz-Bełdzińska 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*

*[1]Developmental Neurology Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy*

**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*

*[1]Holland Bloorview Kids Rehabilitation Hospital, Toronto, Canada, [2]Department of Pediatrics, University of Toronto, Toronto, Canada, [3]Bloorview Research Institute, Toronto, Canada*

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

***Muni Lofra R***[1], *Christie-Brown V, Benesperi G, Bowey A, Scoto M, Abbott L, O'Reilly E, Roberts S, Tsirikos A, Hewitt S, Manchester E, Chatfiled S, Emery N, Narayan S, Nevin K, Gregson S, Sheehan J, Childs A, Marini-Bettolo C*

*[1]The 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 F***[1], *Macken W, Dalal A, Rani N, Ahmed T, ICGNMD Consortium, Bhatia R, Pitceathly R, Kumarasamy T, Reilly M, Srivastava M, Hanna M, Vishnu V*

*[1]All 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**

***Ñungo Garzon N***[1], *Aragon-Gawinska K, Baviera Ricart M, Mora Tatay F, Ibañez Albert M, Hervás D, Pitarch Castellano I, Vazquez Costa J*

*[1]Neuromuscular Unit, Hospital Universitario y Politécnico la Fe, Instituto de Investigación Sanitaria la Fe (IIS La Fe), Valencia, Spain*

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

***Ohashi H***[1], *Bo R, Sonehara S, Nambu, Sunami A, Osawa K, Shirakawa T, Nishio H, Matsuo M, Awano H*

*[1]Department 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*

*[1]Department of Paediatrics, MDUK Oxford Neuromuscular Centre and NIHR Oxford Biomedical Research Centre, University of Oxford, Oxford, United Kingdom, [2]Department 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 P***[1], *Hjalmarsson E, Eklund E, Sofou K*

*[1]SahlgrenskaUniversityHospital, Gothenburg, Sweden*

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

***Tajsharghi H***[1], *Namdari M, Ansari B, Basiri K, Azimi E, Hosseinzadeh M, Bahreini A, Nouri N, Sedghi M, Fattahpur S, Amoli M*

*[1]University Of Skovde, Skovde, Sweden*

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

***Tirucherai G***[1], *Xu Y, Singh A, Bilic S, Gosselin N, Koeck K, Cote S, Yao B, Marantz J*

*[1]Scholar 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 Koprusakova M*

*[1]Slovak Centre for Neuromuscular Disorders, Department of Neurology, University Hospital Bratislava, Medical Faculty, Slovak Medical University, Slovakia, Bratislava, Slovakia, [2]Clinic of Neurology, Jessenius Faculty of Medicine in Martin, Comenius University Bratislava, Slovakia, Martin, Slovakia*

**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*

*[1]Huashan Hospital, Fudan University, Shanghai, China, [2]National Center for Neurological Disorders (NCND), Shanghai, China, [3]Huashan 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 Z***[1], *Stygall K, Bull L, Ninan T, Parasuraman D, Fullwood I*

*[1]University Hospitals Birmingham NHS Trust, Birmingham, United Kingdom*

**533P Nationwide newborn screening for spinal muscular atrophy in the Netherlands**
***Asselman F***[1], *van der Woude D, Cuppen I, Wadman R, Seppenwoolde W, Bartels B, van der Pol L*
[1]*University Medical Center Utrecht, Utrecht, Netherlands*

**534P Impaired renal function in patients with spinal muscular atrophy: a longitudinal cohort study**
***Asselman F***[1], *Meijvis S, Wadman R, Cuppen I, Vernooij R, Vermeer L, van den Berg L, Groen E, van der Pol L*
[1]*University 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*
[1]*Dubowitz Neuromuscular Centre; UCL Great Ormond Street Institute of Child Health, London, United Kingdom,* [2]*National 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 H***[1], *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*
[1]*Department 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 X***[1], *Li H, Hu C, Wu M, Zhou S, Wang Y, Li W*
[1]*Children'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 J***[1], *Acsadi G, Coceani N, Alessi F, Cazzaniga S, Bettica P, Finanger E*
[1]*Children's Hospital of Philadelphia*

**652P RKER-065 improved the therapeutic response to phosphorodiamidate morpholino oligomers (PMO) in a Duchenne muscular dystrophy mouse model**
***Fisher F***[1], *Zhen G, Nathan R, Linzey M, Hack F, Cahill M, Daman T, Macaluso S, Todorova R, Grenha R, Lachey J, Lerner L, Seehra J*
[1]*Keros Therapeutics, Lexington, United States*

**653P Mini muscles for a major cause: creating patient-derived organoids to model Duchenne muscular dystrophy (DMD)**
***Flynn-Rizk R***[1], *Egiian M, Hoffman L*
[1]*Western 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 M***[1], *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*
[1]*Entrada Therapeutics, Boston, United States*

**655P Counteracting reactive oxygen species in models of Duchenne muscular dystrophy improves antisense-oligonucleotide mediated dystrophin recovery**
***Goossens R***[1], *Schneider A, Filonova G, Bijl I, Tanganyika-de Winter C, van Putten M, Aartsma-Rus A*
[1]*Leiden 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 M***[1], *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*
[1]*Royal 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, Barthélèmy I, Blot S, Thao Bui M, Evangelista T, Richard I,* ***Israeli D***[1]
[1]*Genethon, INSERM UMR_S951, Evry Univierisity, Paris-Saclay University, Evry, France*

**658P 3.5-year safety and efficacy data of brogidirsen from DMD patients in open-label extension study**
***Komaki H***[1], *Takeshita E, Kunitake K, Shimizu-Motohashi Y, Maruyama S, Zhang H, Miyagi R, Aoki Y*
[1]*National Center of Neurology and Psychiatry, Tokyo, Japan*

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

***Kuntz N***[1], *Ung B, Khellaf M, Done N, Liu Y, Tuttle E, Grabich S*
[1]*Lurie Children's Hospital of Chicago, Chicago, Illinois, United States*

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

***Laine R***[1], *Liberati C, Ghosh P*
[1]*Boston 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 E***[1], *McDonald C, Laverty C, Finanger E, Signorovitch J, Parodi A, Alessi F, Cazzaniga S, Bettica P, Péréon Y*
[1]*Pediatric Neurology Institute, Catholic University and Nemo Pediatrico, Fondazione Policlinico Gemelli IRCCS, Rome, Italy*

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

***Mostosi D***[1], *Farini A, Strati F, Molinaro M, Saccone S, Amoroso C, Cassani B, Leonetti E, Caprioli F, Facciotti F, Torrente Y*
[1]*University 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**

***Nakamuraa H***[1], *Oba M, Izumi S, Ishizuka H, Komaki H, Remudy DMD invstigator group R*
[1]*National Center of Neurology and Psychiatry, Tokyo, Japan*

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

***Parampalli Ravindra S***[1], *Banavara S, Khandekar G, Varghese V, Agnes Mathew A*
[1]*Synapse 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*
[1]*Association Institut De Myologie, Paris, France,* [2]*Sorbonne 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 D***[1], *Stadheim A, Parasrampuria R, Weinstein D, Sethuraman N*
[1]*Entrada Therapeutics, Boston, United States*

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

***Rogers R***[1], *Rannou A, Manriquez N, Hong Y, Liu W, Fournier M, Alfaro J, Marbán E*
[1]*Cedars-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 A***[1], *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*
[1]*Leiden 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 S***[1], *Sakellariou E, Paliouras G*
[1]*Stichting 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 T***[1], *Sullivan R, Shih R, Coppola J, Sweeney L, Forbes S*
[1]*University Of Florida, Gainesville, United States*

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

***Taivassalo T***[1], *Sullivan R, Bomma M, Lott D, Forbes S, Shih R, Del Toro V, Maldonado-Puebla D, Walter G, Coppola J, Sweeney L*
[1]*University 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*
[1]*Cincinnati Children's Hospital, Cincinnati, United States,* [2]*University 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 L***[1], *Aghaeipour A, Coulson D, Frommer J, Aguti S, Privolizzi R, Zhou H, Muntoni F, TransNAT C*
[1]*Neurodegenerative 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 A***[1], *Eskuri J, Flanigan K, Laverty 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*
[1]*Arkansas 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 A***[1], *Finanger E, Ciafaloni E, Duong T, Proud C, Rocha C, Salinas M, Cazzaniga S, Gruppioni K, Crawford T*
[1]*Arkansas 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 K***[1], *Gonzalez Castillo Z, Valle M, Forrest D, Brown M, Williamson S, Iannaccone S, Nelson L*
[1]*UT Southwestern, Dallas, United States*

**677P Efficacy of the "fantastic four" forts with Duchenne muscular dystrophy and heart failure**

***Arahata H***[1]
[1]*NHO 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 M***[1], *McDonald C, Bellanti F, Cazzaniga S, Bettica P, Schara-Schmidt U, Mercuri E, Muelas N, Vandenborne K*
[1]*The John Walton Muscular Dystrophy Research Centre, Newcastle University, Newcastle upon Tyne, United Kingdom*

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

***Laverty C***[1], *Phan H, Alessi F, Cazzaniga S, Bettica P, Finkel R*
[1]*University 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**

***Laverty C***[1], *Mercuri E, Alessi F, Cazzaniga S, Bettica P, Largajolli A, Bellanti F, Péréon Y, McDonald C, Finanger E*
[1]*University 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 J***[1], *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*
[1]*University of Calgary, Calgary, Canada*

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

***McAdam L***[1], *Brandsema J, Finanger E, Alessi F, Cazzaniga S, Bettica P, Niks E*
[1]*Holland 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 D***[1], *Porco D, Stadheim A, Parasrampuria R, Weinstein D, Sethuraman N*
[1]*Entrada Therapeutics, Boston, United States*

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

***Vučinić D***[1], *Laverty C, Alessi F, Coceani N, Bettica P, Nevo Y*
[1]*Clinic 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 T***[1], *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*
[1]*Robert 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 T***[1], *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*
[1]*Robert 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**

*Laverty C,* ***Finanger E***[1], *Alessi F, Cazzaniga S, Bettica P, Largajolli A, Bellanti F, Péréon Y, McDonald C, Mercuri E*
[1]*Shriners 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*
[1]*Department of Child Neurology, University Hospitals Leuven, Leuven, Belgium,* [2]*Department 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 A***[1], *Jassal B, Vishnu V, Suri V, Sharma M*
[1]*All 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 K***[1], *Finnigan P, Godfrey R, Pizzamiglio C, Quinlivan R*
[1]*Department 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 D***[1], *Artemyeva S, Bardakov S, Kuzenkova L, Melnik E, Monakhova A, Murtazina A, Shulyakova I, Vlodavets D, Kurbatov S*
[1]*Autonomous Non-profit Organization «Support for Patients with Ullrich-Bethlem Myopathy», Nizhny Novgorod, Russia*

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

***Schaefer J***[1], *Schoser B, Saak A*
[1]*Uniklinikum C.G. Carus Dresden, Dresden, Germany*

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

***Kierdaszuk B***[1], *Nieporęcki K, Rosińska A, Drozd-Sokołowska J, Kacprzyk P, Machowicz R, Tomaszewska A, Jamroziak K, Basak G, Kostera-Pruszczyk A*
[1]*Department of Neurology, ERN EURO-NMD, Medical University of Warsaw, Warsaw, Poland*

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

***Nogales-Gadea G***[1], *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*
[1]*Badalona Neuromuscular Researcg Group (GRENBA), Germans Trias i Pujol Research Institute, Badalona, Spain*

**124P Imaging–pathology correlation across CNM genotype**

***Uy G***[1], *Saito Y, Yoshioka W, Hayashi S, Noguchi S, Nishino I*
[1]*National 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 H***[1], *Hauer V, Staecker I, Pommerenke C, Steffen A, Kallnischkies H, Kaufmann M, Fähnrich S, Steenpass L*
[1]*Leibniz 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*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*University of Helsinki, Helsinki, Finland,* [3]*University of Genova, Genova, Italy*

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

***Pedroza Martins A***[1], *Hilario B. Mattos A, V. B. L. Lima I, Castelo Branco Pupe C, Rolim Muro Martinez A, Nucci A, Cavalcante França Júnior M*
[1]*Faculty 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 A***[1], *Motta Mecê A, Iwabe C, Moreschi F, Fernando Grossklaus, L, SJ Dertkigil S, Rolim Muro Martinez A, Nucci A, Cavalcante França Júnior M*

[1]*Faculty of Medical Science, Department of Neurology, State University of Campinas, UNICAMP, Campinas, Brazil*

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

***Coşkun A***[1], *Topaloğlu H*

[1]*Ataturk Sanatoryum Training and Research Hospital, Ankara, Turkey*

**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*

[1]*Association Institut de Myologie, Paris, France,* [2]*Inserm U974, Paris, France*

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

*Urcuyo G,* ***Gómez-Andrés D***[1], *Costa Comellas L, Toro Tamargo E, López López J, Álvarez Molinero M, Munell F*

[1]*Child 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*

[1]*University Paris Est Créteil, Inserm, U955, IMRB, Créteil, France,* [2]*Reference 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,*

[1]*Neuromuscular Repair Unit, Institute of Experimental Neurology, Ospedale San Raffaele, Milan, Italy,* [2]*Dubowitz 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 S***[1], *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*

[1]*Neuromuscular 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*

[1]*Department Of Neuromuscular Research, National Center of Neurology and Psychiatry (NCNP), Kodaira, Japan,* [2]*Department of Pediatrics, Showa General Hospital, Kodaira, Japan*

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

***Takahashi A***[1], *Kunitake, Fujino G, Kobayashi K, Kanagawanagawa M, Satake W, Aoki Y, Toda T*

[1]*The 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 M***[1], *Rimoldi M, Del Bo R, Ciscato P, Cosentino G, Comi G, Corti S, Ronchi D, Ravaglia S, Velardo D*

[1]*Department 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*

[1]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan,* [2]*Department of Genome Medicine Development and Clinical Genome Analysis, Medical Genome Center, National Center of Neurology and Psychiatry, Tokyo, Japan*

**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*

[1]*The Ottawa Hospital, Uottawa, Ottawa, Canada,* [2]*Service d'ENMG et de Pathologies Neuromusculaires, Centre de Référence des Maladies Neuromusculaires PACA-Réunion-Rhône-Alpes, Hôpital Neurologique Pierre Wertheimer, Hospices Civils de Lyon, Groupement Est, Lyon, France,* [3]*Université Claude Bernard Lyon 1, Lyon, France,* [4]*INMG-PGNM, CNRS UMR5261 – INSERM U1315, Faculté de Médecine et de Pharmacie, Lyon, France*

**140P Neuromuscular symptoms of ORAI1-related immunodeficiency**

***Czeczko K***[1], *Jędrzejowska M, Potulska-Chromik A, Franaszczyk M, Wolska-Kuśnierz B, Rosiak E, Aragon-Gawińska K, Stokłosa T, Kostera-Pruszczyk A*

[1]*Department Of Neurology, Medical University of Warsaw, European Reference Network EURO-NMD, Warsaw, Poland*

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

***Fisher G***[1], *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*

[1]*MDUK Neuromuscular Centre, Department of Paediatrics, University of Oxford, Oxford, United Kingdom*

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

***Bisciglia M***[1], *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*

[1]*Université libre de Bruxelles (ULB) & Hôpital Universitaire de Bruxelles (H.U.B) & CUB Hôpital Erasme, Neurologie, Brussels, Belgium*

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

***Jimenez-Mallebrera C***[1], *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*

[1]*Neuromuscular Unit, Neuropaediatrics Department, Institut de Recerca Sant Joan de Déu, Hospital Sant Joan de Déu, Barcelona, Spain*

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

***Bohill J***[1], *Hickson L, Marini-Bettolo C*

[1]*John Walton Muscular Disease Research, Newcastle University, United Kingdom*

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

***Longatto J***[1], *Main M, Muntoni F*

[1]*Great Ormond Street Hospital, London, United Kingdom*

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

***Natera-de Benito D***[1], *Madrigal I, Villar-Vera C, Arca G, Expósito-Escudero J, Rodríguez-Revenga L, Piolatti-Luna A, Muelas N, Vilchez R, Ciutad Celdran M, Codina A, Estévez-Arias B, Carrera-Garcia L, Ortez C, Rodriguez-Carunchio L, Sebastiani G, Azorin I, Nascimento A, Jou C, Vilchez J*

[1]*Hospital Sant Joan de Déu, Barcelona, Spain*

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

***de Laat E***[1], *Groothuis J, Bouman K, Houwen - van Opstal S, Erasmus C, Voermans N*

[1]*Department of Neurology, Donders Institute for Brain, Cognition and Behaviour, Radboud University Medical Center, Nijmegen, Netherlands*

**148P A case of congenital nemaline myopathy with heart failure**

***Saito M***[1], *Shimonaga T, Kimura K, Kodama M, Sumimoto Y, Masada K,Kinoshita H, Sugino H, Kurashige T*

[1]*NHO 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 L***[1], *Baelde R, Galli R, Nieuwendijk M, de Winter J, Ottenheijm C*

[1]*Amsterdam 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*

[1]*Great Ormond Street Hospital for Children NHS Foundation Trust, London, United Kingdom,* [2]*Dubowitz Neuromuscular Centre, Institute of Child Health, University College London, United Kingdom*

**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, Anegon I, Buj-Bello A*

[1]*Genethon, Evry, France,* [2]*Université Paris-Saclay, Univ Evry, Inserm, Genethon, Integrare research unit UMR_S951, Evry, France*

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

***Frías M***[1,2], *Badosa C, Jiménez-Mallebrera C, Porta J, Roldán M*

[1]*Institut de Recerca Sant Joan De Deu, Esplugues de Llobregat, Spain,* [2]*Confocal Microscopy and Cellular Imaging Unit, Genetic and Molecular Medicine Department, Pediatric Institute for Rare Diseases, Hospital Sant Joan de Déu, Esplugues de Llobregat, Spain*

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

***Salort-Campana E***[1,2], *Dumas R, Filnemus S, Jannot A, Attarian S*

[1]*Centre de reference PACA Réunion Rhône Alpes, service du Pr Attarian, La Timone University Hospital, Marseille, France,* [2]*Filnemus, Marseille, France*

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

***Baelde R***[1], *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*

[1]*Division of Neurology, Department of Pediatrics, Children's Hospital of Eastern Ontario, University of Ottawa, Canada,* [2]*Department of Paediatrics and Adolescent Medicine, Johannes Kepler University Linz, Kepler University Hospital, Linz, Austria*

**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*

[1]*Department of Pediatrics, Institute of Clinical Sciences, Sahlgrenska Academy, University of Gothenburg, Sweden,* [2]*Department of Pediatrics, Västra Götalands Region, Sahlgrenska University Hospital, Sweden*

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

***Barraza-flores P***[1], *Dasgupta S, Lee W, Cimini B, Beggs A*

[1]*Boston Children's Hospital/Harvard Medical School, Boston, United States*

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

***Ramanujam A***[1], *Hordyjewska-Kowalczyk E, Tylzanowski P*

[1]*Medical 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*

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

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

*Coleman S, McNulty J,* ***Gupta V***[1]

[1]*Brigham And Women's Hospital/Harvard Medical School, Boston, United States*

**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*

[1]*Kids Neuroscience Centre, Kids Research Institute, The Children's Hospital at Westmead, Australia,* [2]*Functional Neuromics, Children's Medical Research Institute, Westmead, Australia,* [3]*Neuroscience, School of Medical Sciences, Faculty of Medicine and Health, The University of Sydney, Camperdown, Australia*

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

***Vainio A***[1], *Sarparanta J, Jonson P, Udd B*

[1]*Folkhälsan Research Center, Helsinki, Finland*

**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*

[1]*IGBMC, Illkirch-Graffenstaden, France,* [2]*Department of Pediatric Neurology, CHU Hautepierre, Strasbourg, France*

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

***Lochmüller A***[1], *Manzur A, Munot P, Robb S, Scoto M, Baranello G, Tedesco F, Simmonds J, Laurence C, McCauley J, Muntoni F, Sarkozy A*

[1]*Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health & Great Ormond Street Hospital, London, United Kingdom*

**165P Antisense oligonucleotide NS-035 targeting pathogenic exon-trapping in Fukuyama muscular dystrophy: a phase 1 trial**
***Fujino G***[1], *Kitamura A, Takahashi A, Maeda M, Kubota A, Tokuyama Y, Wada I, Kobayashi K, Komaki H, Taniguchi-Ikeda M, Ishigaki K, Toda T*
[1]*Department 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 N***[1], *Ralic B, Nuredini A, Cuoghi Costantini R, Peric S, Tupler R*
[1]*Department of Biomedical, Metabolics and Neural Sciences, University of Modena and Reggio Emilia, Modena, Italy*

**194P A human skeletal muscle-on-chip model for facioscapulohumeral muscular dystrophy**
***Augustinus R***[1], *Franken M, Filonova G, van der Maarel S, Pijnappel P, de Greef J*
[1]*Leiden University Medical Center, Leiden, Netherlands*

**195P Identification of genetic modifiers of disease severity and methylation in large multigenerational kindred with FSHD**
***Butterfield R***[1], *Dunn D, Duval B, Moldt S, Weiss R*
[1]*University 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 J***[1], *Van der Woude D, Van Haaren R, Brenninkmeijer R, Voermans N, Van der Pol L, Bartels B, Erasmus C*
[1]*Radboud University Medical Center, Nijmegen, Netherlands*

**197P The landscape of FSHD data collection: a 2025 expansion to the TREAT-NMD FSHD dataset**
***Garrow M***[1], *Poll A, Ali F, Guglieri M, Ambrosini A, Rodrigues M*
[1]*TREAT-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 A***[1], *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*
[1]*The 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 M***[1], *Widholm P, Puma A, Villa L, Cavali M, Pini J, Ezaru A, Marty B, Evangelista T, Thomas R, Danjoux L, Sacconi S*
[1]*AMRA Medical AB, Linköping, Sweden*

**200P Quantitative whole-body MRI biomarker relation to muscle strength & function in FSHD patients**
*Foltz M, Widholm P,* ***Karlsson M***[1], *Pini J, Puma A, Villa L, Cavali M, Ezaru A, Bassez G, Marty B, Evangelista T, Thomas R, Danjoux L, Tard C, Sacconi S*
[1]*AMRA Medical AB, Linköping, Sweden*

**201P Managing spinal deformities in facioscapulohumeral muscular dystrophy: a scoping review of surgical management**
***Pelsma M***[1], *Voermans N, Peters Q, Hoogveld L, de Valle K, Woodcock I, Lark R, Tsirikos A, de Kleuver M, Erasmus C, Houwen S*
[1]*Radboud University Medical Center, Nijmegen, Netherlands*

**202P Disease burden in Serbian patients with facioscapulohumeral muscular dystrophy**
***Ralic B***[1], *Albano N, Viric V, Nuredini A, Azanjac A, Rajić S, Marjanović A, Tupler R, Heatwole C, Perić S*
[1]*Clinical 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, Salviati L, Cavalli M, Ezaru A, Puma A, van der Maarel S*
[1]*Peripheral Nervous System & Muscle Department, Pasteur 2Hospital, Nice University Hospital, Nice, France,* [2]*Institute 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*
[1]*CiRA, Kyoto University, Kyoto, Japan,* [2]*IBBTEC, University of Cantabria/CSIC, Santander, Spain*

**205P Disease progression in facioscapulohumeral muscular dystrophy in a clinical trial timeframe**
***Teeselink S***[1], *Vincenten S, Voermans N, van Alfen N, van Engelen B, Mul K*
[1]*Radboud University Medical Centre, Nijmegen, Netherlands*

**206P Exploring contractile performance with ultrasound in FSHD: preliminary results of the MUSCLE+ study**
***Van Iersel O***[1], *Weekenstroo H, Cameron D, Doorduin J, van Alfen N*
[1]*Radboud 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 L***[1], *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*
[1]*Department of Neurosciences "Rita Levi Montalcini", University of Turin, Turin, Italy*

**208P The FSHD European trial network**
***Voermans N***[1], *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*
[1]*Radboud University Medical Centre, Nijmegen, Netherlands*

**209P An updated international standard of care for facioscapulohumeral muscular dystrophy**
***Voermans N***[1], *Kinoshita J, Evangelista T, Badiani R, Honda M, Statland J*
[1]*Radboud University Medical Centre, Nijmegen, Netherlands*

**210P Expanding the UK FSHD patient registry dataset - improving data collection and amplifying 'patient voice'**
***Walker H***[1], *Hickson L, Muni-Lofra R, Matthews E, Badiani R, Graham A, Marini-Bettolo C*
[1]*The John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University, and Newcastle upon Tyne Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom*

**211P Associations between muscle strength and MRI biomarkers in FSHD: toward imaging-based functional classification**
*Widholm P, Foltz M,* ***Karlsson M***[1], *Pini J, Puma A, Villa L, Cavali M, Ezaru A, Bassez G, Marty B, Evangelista T, Thomas R, Danjoux L, Tard C, Sacconi S*
[1]*AMRA Medical AB, Linköping, Sweden*

**212P Facioscapulohumeral muscular dystrophy type 2 in Chinese cohort: findings from a large-scale 4qA D4Z4 methylation screening**
***Xia X***[1], *Liu L, Lu J, Zhao C, Zhu W*
[1]*Huashan Hospital, Fudan University, Shanghai, China*

**213P The China FSHD patient registry: a multicenter survey from the Pan-Yangtze delta neuromuscular alliance (PYDNA)**
***Xia X***[1], *Liu L, Yue D, Wu G, Ke Q, Zhao C, Zhu W*
[1]*Department 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*
[1]*Springbok Analytics, Charlottesville, VA, United States,* [2]*University 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 C***[1], *Ruth P, Ulrich S, Vogt-Domke S, Ismail S, Karman L, Falisse A, Muccini J, Covitz S, Day J, Delp S, Duong T*
[1]*Stanford University, Department of Neurology, Palo Alto, United States*

**216P Genetic ablation of Cdkn1a ameliorates facioscapulohumeral muscular dystrophy in mice overexpressing FRG1**
***Fantini S***[1], *Bisceglia G, Zyla J, Moio N, Nuredini A, D'Antona G, Vattemi G, Malfatti E, Polanska J, Tupler R*
[1]*Department of Biomedical, Metabolic and Neural Sciences University of Modena and Reggio Emilia, Modena, Italy*

**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*
[1]*The Ottawa Hospital, Uottawa, Ottawa, Canada,* [2]*Faculty of Medicine, University of Ottawa, Ottawa, Canada,* [3]*Hôpital Neurologique de Lyon, Hospices Civils de Lyon, Lyon, France,* [4]*PGNM, 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 T***[1], *Fossmo H, Raastad T, Fjermestad K, Ramberg C, Nilsen K, Elgstøen K, Arntzen K, Devik K, Morsund Å, Varhaug K, Ørstavik K*

[1]*Oslo University Hospital, Oslo, Norway*

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

***Bhatia A***[1], *Khanna M, Jaison V, Mahey P*

[1]*Neuromuscular 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*

[1]*Irccs Stella Maris Foundation, Pisa, Italy,* [2]*University of Pisa, Pisa, Italy*

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

***Clarke S***[1], *Crittenden B, Graham R, O'Brien-Prince K, McIntosh P*

[1]*Amicus Therapeutics Ltd, Marlow, United Kingdom*

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

***Di Feo M***[1], *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, Verdonschot J, Demidov G, Laurie S, Johari M, Udd B, Savarese M, Solve-RD consortium O*

[1]*Folkhälsan Research Center, Helsinki, Finland*

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

***El Kaim A***[1], *Banos M, Birnbaum S, Hogrel J, Gargiulo M*

[1]*Association Institut De Myologie, Paris, France*

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

***Fossmo H***[1,2,3], *Råen M, Granberg A*

[1]*Vikersund Rehabiliation Centre, Vikersund, Norway,* [2]*Oslo University Hospital, Oslo, Norway,* [3]*University of Oslo, Oslo, Norway*

**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 C***[1], *Kajhøj Møller J, Hansen K*

[1]*Aarhus Universiety, 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*

[1]*Department of Physiotherapy, University of Pretoria, Pretoria, South Africa,* [2]*Department of Health and Rehabilitation Sciences, University of Cape Town, Cape Town, South Africa,* [3]*Department of Physiotherapy, Sefako Makgatho Health Sciences University, Pretoria, South Africa*

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

***Kwiatos K***[1], *Potulska-Chromik A, Czeczko K, Kostera-Pruszczyk A*

[1]*Department of Neurology, Medical University of Warsaw, European Reference Network ERN EURO-NMD, Warsaw, Poland*

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

***Lilien C***[1], *Hill S, Mavrommati F, Ramjattan H, Taylor F, Collet J, Servais L*

[1]*STRONG, 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*

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

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

***Ramberg C***[1], *Hæstad C, Dybesland Rosenberger A, Nyborg Stefansson V, Arntzen K, Arora T, Ludt Fossmo H, Ørstavik K*

[1]*National Neuromuscular Centre Norway, University Hospital of North Norway, Tromsø, Norway*

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

***Rosenberger A***[1], *Ørstavik K, Fossmo H, Nordstrøm M*

[1]*National Neuromuscular Centre Norway, University hospital of North-Norway, Tromso, Norway*

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

***Weichbrodt J***[1], *Lindqvist J, Svensson J, Gudmundsson M, Ekström A*
[1]*Queen 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*
[1]*Robert Jones and Agnes Hunt Hospital, Oswestry, United Kingdom,* [2]*Chester 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 S***[1], *Pagliarani S*
[1]*Jordan 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 Z***[1], *Yoon G, Evesson F, Cooper, Gonorazky H*
[1]*Division 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 A***[1], *Kustermann M, Aigner-Radakovics K, Diofano F, Marchi E, Bittner R, Bernard E, Harpell R, Just S, Lyon G, Schmidt W*
[1]*Molecular 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], *Rolleri E, Luigetti M, Fattori F, Bosco L, Primiano G, Rizza T, Bertini E, Mercuri E*
[1]*Pediatric Neurology, Università Cattolica del Sacro Cuore, Rome, Italy,* [2]*Centro Clinico Nemo and Fondazione Agostino Gemelli IRCCS, Rome, Italy*

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

***Coşkun A***[1], *Yüksel D, Topaloğlu H*
[1]*Ataturk Sanatoryum Training and Research Hospital, Ankara, Turkey*

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

***Farhat E***[1], *Lakhdhar I, Chaabouni M*
[1]*The Muscle Lab, Iris Medical Centre, Hedi Nouira Avenue, Ennasr2, 2001 Ariana, Tunisia,*

**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*
[1]*Neuromuscular 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 V***[1], *Natraj Gayathri S, Di Feo M, Mueller J, Sarkozy A, Muntoni F, Jonson P, Lehtokari V, Oghabian A, Hackman P, Udd B, Savarese M*
[1]*Folkhalsan 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*
[1]*University of Gothenburg, Gothenburg, Sweden,* [2]*Sahlgrenska University Hospital, Gothenburg, Sweden*

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

***Mueller J***[1], *Sarkozy A, Phadke R, Rabinowicz S, Manzur A, Dixit A, Scott I, Stenton S, Chambers D, Lochmuller H, Muntoni F*
[1]*Dubowitz Neuromuscular Centre, London, United Kingdom*

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

***Natraj Gayathri S***[1], *Udd B, Hackman P, Oghabian A, Savarese M*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*University of Helsinki, Faculty of Medicine, Helsinki, Finland*

**442P Splicing regulation of muscle function-associated genes in skeletal muscle tissues**
***Oghabian A***[1], *Sian V, Jonson P, Udd B, Hackman P, Gomez Andres D, Munell F, Camacho Soriano J, Sanchez Duran M, Savarese M*
[1]*Folkhälsan Research Center (Myofin Lab), Helsinki, Finland*

**443P Acute respiratory failure in pregnancy: the presenting symptom of a disorder of gliomedin, an essential component of the nodes of Ranvier**
***Patel R***[1], *Donkervoort S, Quinn C, Baer M, Rey M, Mora J, Foley A, Bönnemann C*
[1]*Neuromuscular 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 A***[1], *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*
[1]*Faculty 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*
[1]*Institute of Clinical Medicine, University of Tartu, Tartu, Estonia,* [2]*Genetics and Personalized Medicine Clinic, Tartu University Hospital, Tartu, Estonia*

**446P Structural variants (SVs) underlying neurogenetic and neuromuscular diseases**
***Ravenscroft G***[1], *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*
[1]*The Harry Perkins Institute of Medical Research, Perth, Australia*

**447P A novel hemizygous CD99L2 variant in adult patient with neurogenic muscle impairment**
***Sarv S***[1], *Puusepp S, Väli L, Ganesh V, Stenzel W, Goebel H, Udd B, Kahre T, Õunap K*
[1]*Genetic 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*
[1]*Neuromuscular reference center Nord/Est/Ile de France, Myology Institute, Sorbonne University, Pitié-Salpêtrière Hospital, Assistance Publique des Hôpitaux de Paris , Paris, France,* [2]*Department 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], *Savarase M, Johari M, Kiiski K, Rusanen S, Wallgren-Pettersson C, Pelin K, Udd B, Hackman P*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*Department 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, Vilaisaktipakorn P, Nagaraj C, Mazur J, Turnwald A, Tian C*
[1]*Cincinnati Children's Hospital Medical Center, Cincinnati, United States,* [2]*University 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 G***[1], *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*
[1]*UHS 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 T***[1]
[1]*Copenhagen Neuromuscular Center, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark*

**453P ASCC3-related congenital myopathy is linked to a defect in ribosome-associated quality control**
***Onnée M***[1], *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*

**454P A case series of hnRNP-K associated myopathy**
***Schiava M***[1], *Mannion J, Castiglioni C, Jofre J, Suarez B, Roos A, Kölbel H, Witting N, Muelas Gomez N, Martí P, Vílchez 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*
[1]*John Walton Muscular Dystrophy Research Centre, Newcastle Upon Tyne, United Kingdom*

**455P Cracking the code: genotype-phenotype correlation models in sarcoglycanopathies**
***Luce L***[1], *Kocak G, Alonso-Pérez J, Straub V, Töpf A, Díaz-Manera J, on behalf of Sarcoglycanopathy group*
[1]*John 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 R***[1], *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*
[1]*Neuromuscular 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 J***[1], *Choi Y*
[1]*Seoul 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*
[1]*Department Of Adult Neurology, Habib Bourguiba Hospital, Sfax, Tunisia, Sfax, Tunisia,* [2]*Laboratory 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 N***[1], *Thomas A, Baskar D, Vengalil S, Nashi S, Bartur A*
[1]*National Institute of Mental Health and Neuro Sciences, Bengaluru, India*

**487P Identifying prognostic biomarkers in a large cohort of patients with LGMDR2-dysferlin related**
***Bolaño Diaz C***[1], *Verdu Diaz J, James M, Hilsden H, Consortium J, Straub V, Diaz Manera J*
[1]*John 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*
[1]*Association Institut de Myologie, Paris, France,* [2]*Sorbonne 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*
[1]*Neuromuscular Unit, Department of Neurology, Hospital Sant Joan de Déu, Barcelona, Spain,* [2]*Laboratory 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 C***[1], *Estévez Arias B, Martín Jiménez P, Ortez C, Carrera-Garcia L, Expósito-Escudero J, Codina A, Jou C, Hernández Laín A, Oyarzábal A, Nascimento A, Camacho A, Dominguez Gonzalez C, Natera de Benito D*
[1]*Hospital Sant Joan de Déu, Barcelona, Spain*

**492P Characterization of a mouse model for plectin-related limb-girdle muscular dystrophy**
***Getmanchuk-Zaporoshchenko V***[1], *Sarnova L, Korelova K, Novotna P, Benada O, Schröder R, Gregor M*
[1]*Laboratory 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*
[1]*Children's National Hospital, Washington DC, United States,* [2]*The 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*
[1]*New Giza University, Giza, Egypt,* [2]*Myo-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 C***[1]
[1]*Hacettepe 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*
[1]*University Paris Est, Créteil, France,* [2]*APHP, Mondor University Hospital, Créteil, France*

**497P Late-onset SELENON-related myopathy**
***Nath S***[1], *Liewluck T, Madigan N, Milone M*
[1]*Mayo 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,*
[1]*Department of Neurology, Dr. Cipto Mangunkusumo Hospital, Jakarta, Indonesia*

**499P Training of physiotherapists in clinical and functional outcome assessment in patients with LGMDR2**
***Sneha S***[1], *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*
[1]*All 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 A***[1], *Omairi S, El Sherif R, Nishino I, Al_Ghurabi M*
[1]*Neurologist 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*
[1]*Department of Neurology and National Neuromuscular Centre, Norway, University Hospital of North Norway , Tromsø, Norway,* [2]*Institute of Clinical Medicine, University of Tromsø, The Arctic university of Norway, Tromsø, Norway*

**502P A LGMDD2 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*
[1]*Human Translational Genomics Group, University of Valencia, Valencia, Spain,* [2]*Centre for Biomedical Network Research on Rare Diseases (CIBERER,CB23/07/00005), Madrid, Spain,* [3]*INCLIVA Biomedical Research Institute, Valencia, Spain*

**503VP AAV-based TCAP delivery rescues collapsed desmin cytoskeleton-driven mitochondria dislocation in limb-girdle muscular dystrophy R7**
***Lv X***[1], *Yan C, Lin P*
[1]*Qilu 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*
[1]*MyoCare National Foundation, Cairo, Egypt,* [2]*Newgiza 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 M***[1], *Cugudda E, Gemma M, Maggi L*
[1]*Neuroipatiemmunology 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 X***[1], *Jiao K, Gao M, Yue D, Zhu H, Li X, Zhao C, Zhu W, Xi J*
[1]*Department 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 Y***[1], *Kim S, Choi Y, Park H*
[1]*Department 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 M***[1], *Quaranta C, Corradini M, Gardani A, Boselli A, Di Franco S, Khalil S, Ballante E, Berardinelli A*
[1]*Department 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 K***[1], *Chen X, Yue D, Gao M, Zhao C, Huang G, Xi J, Zhu W, Zhu H*
[1]*Huashan 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*
[1]*The Harry Perkins Institute for Medical Research, Perth, Australia,* [2]*Centre 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 H***[1], *Pauper M, Hentschel A, Schänzer A, Beltran S, Thompson R, Aksel-Kilicarslan O, Gangfuß A, Schara-Schmidt U, Lochmüller H, Polavarapu K, Roos A*
[1]*Department 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*
[1]*Neuromuscular Morphology Unit, Institut De Myologie, Paris, France,* [2]*Department 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*
[1]*Division of Neurology, Department of Medicine, The Ottawa Hospital, Ottawa, Canada,* [2]*Faculty of Medicine, University of Ottawa, Ottawa, Canada,* [3]*The 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*
[1]*Department of Neurology, Dr. Cipto Mangunkusumo Hospital, Jakarta, Indonesia,* [2]*Faculty 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*
[1]*Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan,* [2]*Department of Genome Medicine Development, Medical Genome Center, National Center of Neurology and Psychiatry, Tokyo, Japan,* [3]*Department 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, Kiriyama T, Mori E, Nakamura S, Nishino I*
[1]*Department of Neurology, Nara Medical University, Kashihara, Japan,* [2]*Center for Autophagy and Anti-Aging Research, Nara Medical University, Kashihara, Japan,* [3]*Department 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*
[1]*CRCHU de Québec - Université Laval, Quebec, Canada,* [2]*Département 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*
[1]*Neuroscience Axis of CHU de Quebec – Université Laval Research Center, Quebec, Canada,* [2]*Faculty of Medicine of Université Laval, Quebec, Canada,* [3]*Département 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*
*[1]Institut de myologie, Paris, France, [2]Database unit, Institut de myologie, Paris, France, [3]Sorbonne Université, INSERM, Centre de Recherche en Myologie, Institut de myologie, Paris, France, [4]AP-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 PLIN4-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*
*[1]Department of Pharmacological and Biomolecular Sciences, University of Milan, Milan, Italy, [2]Neuroimmunology 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 Hagenacke, 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*
*[1]Department of Cell & Developmental Biology, University College London, United Kingdom, [2]The 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*
*[1]Kawasaki Medical School, Kurashiki, Japan*

**58P Wave surfing in Duchenne muscular dystrophy; fun or folly?**
*Fleerakkers 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]*
*[1]Leiden University Medical Center, The Netherlands, Leiden, Netherlands, [2]Duchenne Center Netherlands, Leiden, Netherlands, [3]Basalt 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]*
*[1]INSERM UMRS 974 Sorbonne University, Myology Research Center, Institute of Myology, Paris, France, [2]Neuromuscular 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*
*[1]Leiden 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*
*[1]Neurology (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*
*[1]Twine Therapeutics, Baltimore, United States*

**63P Targeting a premature termination mutation in COL6A2 for collagen VI-related dystrophy**
*__Osegui Barcenilla N__[1], González Moro, Lopez Martinez A, Kapetanovic S, Arechavala Gomeza V*
*[1]Nucleic 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*
*[1]Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan, [2]Department 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*
*[1]Department of Cell & Developmental Biology, University College London, United Kingdom, [2]The 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*
*[1]Kawasaki Medical School, Kurashiki, Japan*

**58P Wave surfing in Duchenne muscular dystrophy; fun or folly?**
*Fleerakkers 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]*
*[1]Leiden University Medical Center, The Netherlands, Leiden, Netherlands, [2]Duchenne Center Netherlands, Leiden, Netherlands, [3]Basalt 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]*
*[1]INSERM UMRS 974 Sorbonne University, Myology Research Center, Institute of Myology, Paris, France, [2]Neuromuscular 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*
*[1]Leiden University Medical Center, Leiden, Netherlands*

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***Yae Y**[1,2], Ogawa M, Hayashi S, Nishino I, Noguchi S*
*[1]Department of Neuromuscular Research, National Institute of Neuroscience, National Center of Neurology and Psychiatry, Tokyo, Japan, [2]Department 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*
*[1]Amicus 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]*
*[1]Amicus Therapeutics Ltd, Marlow, United Kingdom*

**67P Mexiletine Paediatric Investigation Plan, PIP4 study: safety, pharmacokinetic and efficacy findings in children with myotonia**

*Barnérias C, Isapof A, Hogrel J, Pentikis H, Adetoro N, **Zozulya Weidenfeller A**[1]*
*[1]Lupin 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*
*[1]Donostia University Hospital, San Sebastian, Spain, [2]Biogipuzkoa 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*
*[1]Department of Neurology, University Hospitals Leuven, Leuven, Belgium, [2]Department 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 L**[1], McAdam L, Alessi F, Muelas N, Gruppioni K, Cazzaniga S, Bettica P, Zaidman C*
*[1]University 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*
[1]*Neuromuscular Diseases Unit, Neurology Department, Hospital Universitario 12 de Octubre, Madrid, Spain,* [2]*Hospital Universitario 12 de Octubre (imas12), Madrid, Spain,* [3]*Biomedical 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 J***[1], *Rodriguez-Garcia S, Neto A, Papadopoulou D, Van Baelen B, Duncombe P, De Ceuninck L, Van Der Woning B, Aggarwal R*
[1]*Department 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 K***[1], *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*
[1]*Johnson & 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 A***[1], *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*
[1]*Astellas 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 T***[1], *Liu Y, Gromova A, La Spada A, Chen Y*
[1]*AnnJi 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 A***[1], *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*
[1]*ERN-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 C***[1], *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*
[1]*Kepler University Hospital, Linz, Austria*

**78P Go high or go long? Pharmacokinetics of AAV in a large animal model**

***Hildyard J***[1], *Riddell D, Harron R, Stathopoulou T, Robinson A, Esteves V, Sophiana P, Wells D, Pelligand L, Piercy R*
[1]*Royal Veterinary College, London, United Kingdom*

**79P Observed efficacy of efgartigimod in generalized myasthenia gravis across patient subgroups in the ADAPT-SC+ study**

***Kellermair L***[1], *Muppidi S, Vu T, Brauer E, Kerstens R, Utsugisawa K, Meisel A*
[1]*Kepler University, Linz, Austria*

**80P Delandistrogene moxeparvovec in Duchenne muscular dystrophy: long-term EMBARK 2-year functional outcomes, safety, and micro-dystrophin expression**

***Muntoni F***[1], *Mendell J, McDonald C, Mercuri E, Ciafaloni E, Komaki H, Leon-Astudillo C7, Nascimento A, Proud C, Schara-Schmidt U, Veerapandiyan A, Zaidman C, Murphy A, Rodino-Klapac L*
[1]*Dubowitz 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 A***[1], *Yuen M, Oates E, Quinlan K*
[1]*School 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**

***Tejedera Villafranca A***[1], *Ruiz-Gutiérrez M, Ugarte-Orozco M, Cortés-Reséndiz A, Ramón-Azcón J, Fernández-Costa J*
[1]*Institute 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 H***[1], *Taimi M, Hall S, Bobba S, Jiang Y, Grayson D, Seehra J, Chyung Y, Bogdanovich S*
[1]*Keros 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 C***[1], *Marhic A, Fer F, Brechenmacher M, Negroni E, Butler-Browne G, Mouly V, Marolleau J, Larghero J, Perie S, Lacau St Guily J*
[1]*Sorbonne 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 stud*ies***

***Wiguna F***[1,2], *Mutiani F, Fadli N, Wiratman W, Safri A, Octaviana F, Hakim M, Indrawati L*
[1]*Department of Neurology, Faculty of Medicine Universitas Indonesia, Jakarta, Indonesia,* [2]*Department 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 G***[1], *Lopez Perez M, Weisleder N*
[1]*Department 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 J***[1], *Richard I, Stojkovic T, Straub V, Preisler N, Zanfongnon R, Degove S, Buscara L, Genries-Ferrand S, Olivier S*
[1]*University 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 J***[1]
[1]*Cerbm 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*
[1]*Facultad de Odontología, Universidad San Sebastian, Santiago, Chile,* [2]*Fundación 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 E***[1], *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, Bruzzone S, Bruno C*
[1]*Unit 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 M***[1], *Šmerc R, Serša I, Miklavčič D, Kranjc M, Mahnič-Kalamiza S*
[1]*UL Faculty of Electrical Engineering, Ljubljana, Slovenia*

**106P Development of sarcomere-observable mice for analyzing skeletal myofibril degeneration**

***Noguchi S***[1], *Ogawa M, Kobirumaki-Shimozawa F, Inoue Y, Inoue T, Fukuda N*
[1]*National 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 L***[1], *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*
[1]*Department 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 M***[1]
[1]*Human 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*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*University 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 M***[1], *Schmitt L, Liebig K, Hezel S, Bonanno S, Marcuzzo S, Roos A, Kleinschnitz C, Schara-Schmidt U, Maggi L, Hagenacker T*
[1]*Department 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 K***[1], *Schmitt L, Hezel S, Kleinschnitz C, Hagenacker T, Leo M*
[1]*Department 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 L***[1], *Guettsches A, Hentschel A, Ruck T, Schara-Schmidt U, Della Marina A, Schaenzer A, Roos A*
[1]*Department of Pediatric Neurology, Centre for Neuromuscular Disorders, University Duisburg-Essen, Essen, Germany*

**113P Further optimized techniques for 2D myotube culture**
***Jonson P***[1], *Sarparanta J, Sian V, Vainio A, Luque H, Udd B, Savarese M*
[1]*Folkhälsan Research Center, Helsinki, Finland*

**114P Novel insights into molecular mechanisms leading to congenital fibre-type disproportion (CFTD)**
***Rostedt F***[1,2], *Seaborne R, Harmo R, Laarne M, Turku T, Kangas H, Krag T, Vissing J, Ochala J, Pelin K, Laitila J*
[1]*Folkhälsan Research Center, Helsinki, Finland,* [2]*Department 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*
[1]*Department of Integrative Biology and Physiology, University of Minnesota, Minneapolis, United States,* [2]*Department of Biochemistry, Molecular Biology & Biophysics, University of Minnesota, Minneapolis, United States,* [3]*Division 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 A***[1], *Conscience O, Albertella L, Periou B, Souvannanorath S, Malfatti E, Relaix F, Martin L, Authier F2*
[1]*Paris 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 K***[1], *Sukseree S, Fransche L, Schmidt W, Bittner R*
[1]*Medical University of Vienna, Neuromuscular Research Department, Vienna, Austria*

## Poster Session 3 – 251P-296P, 297VP: Dystrophinopathies (animals models, biomarkers, brain, genetics)

**251P A plasma microsampler to enable blood sampling at home for better monitoring of dystropathology in Duchenne muscular dystrophy (DMD)**
*Arthur P, Oh W, Tsioutsias I, Grounds M*
[1]*University 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*
[1]*Department of Neurology, National Hospital Organization Higashisaitama Hospital, Hasuda, Saitama, Japan,* [2]*Department of Neurology, Graduate School of Medicine, The University of Tokyo, Bunkyo, Tokyo, Japan,* [3]*Department 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, Coyan A, Liu J2, Tian C*
[1]*Cincinnati Children's Hospital, Cincinnati, United States,* [2]*University 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 B***[1], *Tulimiero L, Mantuano P, Lenti R, Denoyelle S, Marinelli M, Cappellari O, De Luca A*
[1]*Department 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 J***[1]
[1]*Cerbm 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 C***[1], *Bouquillon L, McDonald R, Eilon T, Geagan C, Conn R, Rodney S, Turner C, Guglieri M, Straub V, Quinlivan R*

[1]*University College London Hospitals NHS Foundation Trust, London, United Kingdom*

**257P Spectrum of dystrophin mutations in a dystrophinopathy cohort in north India**

***Chandu M***[1], *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*

[1]*All 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*

[1]*Umass Chan Medical School, Worcester, United States,* [2]*Brigham and Women's Hospital, Boston, United States,* [3]*Boston Children's Hospital, Boston, United States*

**259P Pax3 protein induction enhances muscle stem cell activation in mdx5cv mice**

***Das Barman S***[1], *Stella M, Olsen S, Jensen U, de Morree A*

[1]*Aarhus University, Aarhus, Denmark*

**260P The FORCE platform delivers CNS functional improvement by resolving anxiety in the mdx mouse model of DMD**

***Desjardins C***[1], *Li S, Hall J, Venkatesan R, Correia S, Cui J, Johnson J, Weeden T, Beskrovnaya O, Zanotti S*

[1]*Dyne Therapeutics, Waltham, United States*

**261P Non-invasive tissue oxygenation monitoring in Duchenne muscular dystrophy rat models**

***Egiian M***[1,2], *Eskandari R, Tyler S, Morrison L, Diop M, Hoffman L*

[1]*University Of Western Ontario, London, Canada,* [2]*Lawson 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 A***[1], *Villa C, Strati F, Cassani B, Facciotti F, Torrente Y*

[1]*Fondazione 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 A***[1], *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*

[1]*Généthon, Evry-Courcouronnes, France*

**264P Characterization of dystrophin expression in muscle biopsies from patients with in-frame deletions in the DMD gene**

***Frair E***[1], *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*

[1]*Center 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*

[1]*Neuromuscular Unit, Department of Neurosciences "Rita Levi Montalcini", University of Turin, Turin, Italy,* [2]*John 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 C***[1], *Riguzzi P, Kolesnik A, Govaarts R, Miranda R, Hendriksen J, Weerkamp P, Stemmerik M, Würgler Slipsager A, Vissing J, Skuse D, Muntoni F, Kan H, Mercuri E, Straub V*

[1]*John 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 S***[1], *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*

[1]*University Hospitals Leuven, Leuven, Belgium*

**268P Comprehensive evaluation of muscle integrity biomarkers to assess therapeutic efficacy in Duchenne muscular dystrophy**

***Gonzalez P***[1], *Boehler J, Ralff M, Bhavsar P, Soustek-Kramer M, Marshall J, Harmelink M, Christoforou N, Brooks G*
[1]*Solid 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,* ***Govaarts R***[1,2]
[1]*Leiden University Medical Center, Leiden, Netherlands,* [2]*Duchenne Center Netherlands, Leiden, Netherlands*

**270P Individuals with Duchenne muscular dystrophy have lower brain volumes, altered white matter microstructure and lower perfusion compared to controls**

***Govaarts R***[1], *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*
[1]*Leiden 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*
[1]*Kempenhaeghe Centre for Neurological Learning Disabilities, Heeze , Netherlands,* [2]*Maastricht University, School for Mental Health and Neuroscience, Maastricht, Netherlands*

**272P Neuronal density in the bulbar cardiorespiratory nuclei of the dystrophin-deficient mdx model**

***Hermes T***[1], *Corsini W, da Silva M, de Almeida L, dos Reis L, Sato M*
[1]*Anatomy 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 A***[1], *Rabelo de Brito M, Junqueira R. Rezende T, Iwabe C, Conte G, Nucci A, Cendes F, Cavalcante França Junior M*
[1]*Faculty 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 F***[1], *Funakoshi S, Fujiwara Y, Sasaki M, Ida K, Sakurai H, Yoshida Y*
[1]*Center for iPS Cell Research and Application, Kyoto University, Kyoto, Japan*

**275P Magnetic resonance imaging based radiomic feature extraction for bone quality assessment in corticosteroid treated boys of Duchenne muscular dystrophy**

***Kunnath Ravindranunni R***[1], *Rosen A, Cottrell C, Vandenborne K, Glenn W, Rajapakse C, Willcocks R*
[1]*University 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 K***[1], *Greenslade A, Hamir M, Jahr K, Crockett L, Van Vleet J, Blank B, Ntai I, Sun B, Zhou H, Melton A*
[1]*Biomarin 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 J***[1], *Gushchina L, Lin H, Frair E, Nicolau S, Flanigan K*
[1]*The 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 A***[1], *Telles Salgueiro Barboni M, Aoki Y, Kremers J, Vaillend C*
[1]*Paris-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 Z***[1], *Joensen H, Scharff Poulsen N, Slipsager A, Teitsdóttir B, Vejlstrup N, Vissing J*
[1]*Rigshospitalet, Copenhagen, Denmark*

**280P A possible skeletal muscle modulator: chemokine CXCL12 in senior citizen, healthy younger adult and Duchenne muscular dystrophy (DMD) patient**

***Maeda Y***[1]
[1]*National 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 M***[1], *Cristiano E, Quarta R, Boccanegra B, Cerchiara A, Barile S, Mouly V, Lasorsa F, Imbrici P, Cappellari O, De Luca A*
[1]*Department 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 R***[1], *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*
[1]*Universidad 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*
[1]*Nationwide Children's Hospital, Columbus, United States,* [2]*The 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 H***[1], *Bourgeois Yoshioka C, Takenaka-Ninagawa N, Yokomizo-Goto M, Miki M, Watanabe D, Yamamoto M, Aoyama T*
[1]*Center 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 P***[1], *Degan C, Tsonaka R, de Vries S, Ikelaar N, van der Holst M, Kan H, Niks E*
[1]*Leiden University Medical Center, Leiden, Netherlands*

**286P Evaluating a panel of plasma biomarkers of DMD in taurine-treated juvenile mdx mice**

***Tsioutsias I***[1], *Terrill J, Bakker A, Arthur P*
[1]*The University of Western Australia, Perth, Australia*

**287P 3D tissue engineered skeletal muscle to model dystrophinopathies and determine the efficacy of exon skip therapies**

***Timp L***[1], *Franken M, Gonçalves M, Goyenvalle A, Bigot A, Mouly V, Pijnappel P, de Greef J, van der Maarel S, Niks E, Goossens R, Aartsma-Rus A*
[1]*Leiden 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 M***[1], *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*
[1]*Leiden 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 M***[1], *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*
[1]*Leiden University Medical Center, Leiden, Netherlands*

**290P Cognitive comorbidities and brain microstructural changes in patients with Duchenne muscular dystrophy: data from Hong Kong**

***Yee P***[1], *Zhang H, Ng R, Mak K, Lee T, Lochmüller H, Chan S*
[1]*Department 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 I***[1], *Martin A, Chan S, Denger B, Bogue L, Mattei F, De Angelis F, Yee P, Masic D, Poll A*
[1]*Parent 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*
[1]*Division of Neurology, Cincinnati Children's Hospital Medical Center, Cincinnati, United States,* [2]*Division of Pediatrics, University of Cincinnati Medical Center, Cincinnati, United States*

**293P Pseudo-normalization of creatine phosphokinase in a newborn with Duchenne muscular dystrophy: a case report**

***Zygmunt A***[1,2], *Buls M, Tian C*
[1]*Division of Neurology, Cincinnati Children's Hospital Medical Center, Cincinnati, United States,* [2]*Division of Pediatrics, University of Cincinnati Medical Center, Cincinnati, United States*

**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 L***[1], *Khan A, Dufresne G, Bradley A, Stevens K, Frair E, Vetter T, Lin H, Flanigan K*
[1]*Jerry 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*
[1]*The Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom,* [2]*National Institute for Health Research UCL Great Ormond Street Institute of Child Health, London, United Kingdom,* [3]*Genetic 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**

***Tetorou K***[1,2], *Muntoni F, Rossi R*
[1]*The Dubowitz Neuromuscular Centre, UCL Great Ormond Street Institute of Child Health, London, United Kingdom,* [2]*National Institute for Health Research, Great Ormond Street Institute of Child Health Biomedical Research Centre, University College London, United Kingdom*

**297VP Genetic and functional predictors of disease progression in Duchenne muscular dystrophy using artificial intelligence**

***Hörnö-Reissner A***[1], *Alexander N*
[1]*Children Hospital of Eastern, Switzerland, St. Gallen, Liechtenstein*

## Poster Session 3 – 366P-398P: Myotonic dystrophy

**366P Caregivers experiences of and perspectives on living and coping with adult-onset myotonic dystrophy type 1**

***Allergodt K***[1,2], *Werlauff U, Dreyer P, Ørstavik K, Handberg C*
[1]*Rehabilitation Center for Neuromuscular Diseases, Aarhus, Denmark,* [2]*Department of Public Health, Faculty of Health, Aarhus University, Aarhus, Denmark*

**367P Healthcare professionals' experiences of and perspectives on the hospital follow-up of patients with adult-onset DM1**

***Allergodt K***[1,2], *Werlauff U, Dreyer P, Thorne S, Handberg C*
[1]*Rehabilitation Center for Neuromuscular Diseases, Aarhus, Denmark,* [2]*Department of Public Health, Faculty of Health, Aarhus University, Aarhus, Denmark*

**368P Mortality and clinical characteristics in DM1 patients: analysis of two large European registries**

***Bassez G***[1], *Wang Y, Nigim F, Wang B, Herweijer E, Quignot N, Read S, Kihlblom S, Ekström A*
[1]*Institute of Myology, Pitié-Salpêtrière Hospital, Paris, France*

**369P TRACK DM: design of longitudinal natural history study in people with myotonic dystrophy linking retrospective data with prospective follow-up**

***Bassez G***[1], *Gyenge M, Zozulya Weidenfeller A*
[1]*Constitutive Reference Center for Neuromuscular Diseases, Neuro-Myology Department, AP-HP Pitié-Salpêtrière, Paris, France*

**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]
[1]*Virginia Commonwealth University, Richmond, United States*

**371P Investigating hand myotonia: determining best practices using novel technologies and approaches to assessing hand myotonia**

***De Monts De Savasse C***[1], *Ismail S, Karman L, Vogt-Domke S, Khonde S, Parker D, Tang W, McIntyre M, Duong T*
[1]*Stanford University, Department of Neurology, Palo Alto, United States*

**372P Integrating cognition and neuroimaging: a study of cerebral regions in myotonic dystrophy**

***Duong T***[1], *Karman L, Anna Yao M, Ismail S, Rogers M, Deutsch G, Sampson J, Day J, Hageman N*
[1]*Stanford University, Palo Alto, United States*

**373P Clinical outcomes and respiratory dysfunction in adults with myotonic dystrophy type 1**

***Fossmo H***[1,2,3,4], *Robinson H, Ørstavik K*

[1]*Unit for inborn and hereditary neuromuscular disorders (EMAN), Department of Neurology, Oslo University Hospital, Oslo, Norway,* [2]*Department of Public Health Sciences and Interdisciplinary Health Sciences, University of Oslo, Oslo, Norway,* [3]*Frambu centre for rare disorders, Siggerud, Norway,* [4]*Vikersund Rehabilitation Centre, Vikersund, Norway*

**374P New conditional mouse model for myotonic dystrophy expressing the human DMPK gene carrying large CTG expansions in striated muscles**

***Furling D***[1], *Sureau A, Arandel L, Cordier A, Rouxel C, Huguet A, Megali P, Lemaitre M, Mougenot N, Rau F, Klein A, Muchir A, Gourdon G*

[1]*Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, Paris, France*

**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*

[1]*Université de Sherbroooke, Sherbrooke, Canada,* [2]*Centre de recherche et d'innovation du CIUSSS SLSJ, Saguenay, Canada*

**376P Aligning research and patient perspectives in myotonic dystrophy type 1: a comparative analysis using data from the DM-scope registry**

***Gyenge M***[1], *Brechenmacher M, Fer F, Hamroun D, Geille A, Bassez G, Filnemus Myotonic Dystrophy Study Group*

[1]*Institute of Myology, Pitié Salpêtrière Hospital, Paris, France*

**377P CoreDMScope_ENSA (RevEal the burdeN in daily life in myotonic dyStrophy due to myotoniA): study design compares patient-reported and clinical outcomes**

***Gyenge M***[1], *Bassez G, Ellafi R, Zozulya Weidenfeller A, Dufresne R, Filnemus Myotonic Dystrophy Study Group*

[1]*Institute of Myology, Pitié Salpêtrière Hospital, Paris, France*

**378P The role of epigenetic regulation of the DMPK locus in congenital myotonic dystrophy**

***Hartman J***[1,2,3], *McMichael O, Aberg K, Hale M, Carrell S, Johnson N*

[1]*Center For Inherited Myology Research, Virginia Commonwealth University, Richmond, United States,* [2]*Department of Neurology, Virginia Commonwealth University, Richmond, United States,* [3]*Department for Human and Molecular Genetics, Virginia Commonwealth University, Richmond, United States*

**379P The UK myotonic dystrophy patient registry – advancing research and amplifying patient voices through a powerful translational tool**

***Bohill J***[1], *Hickson, L, Walker H, Sodhi J, Turner C, Monckton D, Hewamadduma C, Hamilton M, Ashley EJ, Walker M, Adcock K, , Marini-Bettolo C*

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

**380P DYNE-101 targets the underlying cause of DM1 to enable multi-system functional improvement in the ACHIEVE trial**

***Kerr D***[1], *Sansone V, Bassez G, Diaz-Manera J, Mul K, Lilleker J, Pane M, Roxburgh R, Schoser B, Turner C, Ray S, Chen H, Andersson S*

[1]*Dyne Therapeutics, Waltham, United States*

**381P Serum FGF21 and GDF-15 as mitochondrial stress biomarkers in myotonic dystrophy type 2: a German multicenter study**

***Kleefeld F***[1,2], *Ruck T, Schoser B, Roos A, Mensch A*

[1]*Ruhr University Bochum, BG University Hospital Bergmannsheil, Department of Neurology, Bochum, Germany,* [2]*BG University Hospital Bergmannsheil, Heimer Institute for Muscle Research, Bochum*

**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*

[1]*Maastricht University Medical Centre, Maastricht, Netherlands,* [2]*Mental Health and Neuroscience Research Institute, Maastricht, Netherlands*

**383P Multisystem involvement and disability status in adult-onset myotonic dystrophy type 1: a Chinese single-centered study**

***Luo S***[1], *Zhong H, Zhang W, Zhao C*

[1]*Huashan Hospital, Shanghai, China*

**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*

[1]*Yamaguchi University, Ube, Japan,* [2]*Osaka University, Suita, Japan*

**385P Mitochondrial network alterations in myotonic dystrophy type 1 disease**
***Nogales-Gadea G***[1], *Maestre-Mora P, Valls-Roca L, Mosqueira-Martin L, Férriz-Gordillo A, Chojnacki J, Cámara Y, Vallejo A, Garrabou G, Pérez-Berná A, Suelves M*
[1]*Badalona Neuromuscular Research Group (GRENBA), Germans Trias i Pujol Research Institute, Badalona, Spain*

**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 P***[1], *Johnson N*
[1]*Vertex Pharmaceuticals, Boston, United States*

**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*
[1]*Department of Neurology, Pusan National University Hospital, Busan, South Korea*

**388P Neurofilament light chain correlates with the motor function scales in myotonic dystrophy type 1**
***Park J***[1]
[1]*Kyungpook National University Chilgok Hospital, Daegu, South Korea*

**389P Age and causes of death in myotonic dystrophy type 1**
***Put L***[1], *Damen M, Rottier N, Smulders F, Karnebeek I, la Fontaine L, Voermans N, Tieleman A, Faber C, Mul K*
[1]*Maastricht University Medical Centre+, Maastricht, Netherlands*

**390P Clinical and genetic profile of a cohort of myotonic dystrophy from north India**
***Rani N***[1], *Macken W, Dalal A, Wilson L, ICGNMD C, Bhatia R, Garg A, Pitceathly R, Thangaraj K, Srivastava P, Hanna M, Vishnu V*
[1]*All India Institute Of Medical Science (New Delhi), New Delhi, India*

**391P A prospective longitudinal observational study in myotonic dystrophy type 1 (dm1): from clinical outcomes and endpoints to clinical trial design**
***Sansone V***[1], *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*
[1]*The Nemo Clinical Center - Neurorehabilitation Unit, University Of Milan, Milan, Italy*

**392P Expanding the UK myotonic dystrophy patient registry dataset - improving data collection and amplifying 'patient voice'**
***Walker H***[1], *Hickson L, Sodhi J, Monckton D, Ashley E, Marini-Bettolo C*
[1]*The John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University, and Newcastle upon Tyne Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom*

**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*
[1]*Robert Jones And Agnes Hunt Hospital, Oswestry, United Kingdom,* [2]*Chester University Medical School, Chester, United Kingdom*

**394P Myotonic dystrophy type 2 as a phenotypic mimicker of inclusion body myositis**
***Yun P***[1], *Moghekar A, Mohassel P*
[1]*Johns Hopkins University School of Medicine, Baltimore, United States*

**395P Advancing clinical trials in myotonic dystrophy type 1: refining radiological, clinical and patient-reported outcome measures**
***Iterbeke L***[1], *Huysmans , Bamps K, Peeters R, Goosens V, Maes F, Dupont P, Claeys K*
[1]*Laboratory for Muscle Diseases and Neuropathies, Department of Neurosciences, KU Leuven, and Leuven Brain Institute (LBI), Leuven, Belgium*

**396P Development of a sensor-system to regulate gene expression: application to decoy gene therapy for myotonic dystrophy**
***Arandel L***[1], *Sureau A, Cordier A, Ronquillo K, Muchir A, Moulay G, Klein A, Gourdon G, Furling D*
[1]*Centre de Recherche en Myologie/Association Institut De Myologie, Paris, France*

**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*
[1]*The Ottawa Hospital, Civic Hospital, Neuromuscular Center, Ottawa, Canada,* [2]*Cellular and Molecular Medicine, Faculty of Medicine and Pharmacy,Uottawa, Ottawa, Canada,* [3]*Université Claude Bernard Lyon Est, Lyon, France,* [4]*Hôpital Neurologique de Lyon, Hospices Civils de Lyon, Lyon, France,* [5]*PGNM, Institut Neuromyogène, CNRS UMR 5261 - INSERM U1315, Lyon, France*

**398P Whole-body muscle MRI in myotonic dystrophy type II: semiquantitative and quantitative analysis and correlation with clinical severity**
***Mensch A***[1], *Heuschen M, Kölsch A, Kleeberg K, Strube D, Schneider I, Kraya T, Hensel O, Gussew A, Deistung A, Otto M, Stoevesandt D*
[1]*Department of Neurology, University Medicine Halle, Halle (Saale), Germany*

**538P National plan of action to raise awareness and improve medical care of Duchenne muscular dystrophy (AIM-DMD): first year results**

***Akinci G***[1], *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*

[1]*UHS Turkey, Izmir University, Dr Behcet Uz Children's Hospital, Pediatric Neurology, Izmir, Turkey*

**539P The latest insights from the Dutch LGMD registry through patient- and clinician-reported data**

***Hoek R***[1], *Schrama E, van Hees B, van Reenen R, van der Kooi A, Straathof C, Badrising U, van Duyvenvoorde H, Krom Y, Niks E*

[1]*Department of Neurology, Leiden University Medical Center, Leiden, Netherlands*

**540P Overview of the Belgian neuromuscular diseases registry: more than a decade of data**

***Jagut M***[1], *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*

[1]*Sciensano, Belgian Public Health Institute, Brussels, Belgium*

**541P The global registry for 541P COL6-related dystrophies: insights from over 350 international participants**

***Mcdonald S***[1], *Allamand V, Alvarez R, Boddy H, Copier J, Deconinck N, Dziewczapolski G, Ferré X, McAlister B, Mejat A, Sarkozy A, Straub V*

[1]*The John Walton Muscular Dystrophy Research Centre, Newcastle upon Tyne, United Kingdom*

**542P The global FKRP registry: improvements in data collection and harmonisation**

***Mcdonald S***[1], *Alfano L, Brazzo K, Johnson N, Laurent J, Mathews K, Thiele S, Vissing J, Walter M, Woods L, Ørstavik K, Straub V*

[1]*The John Walton Muscular Dystrophy Research Centre, Newcastle Upon Tyne, United Kingdom*

**543P Global insights into limb girdle muscular dystrophies from the TREAT-NMD global registries network**

***Mcdonald S***[1], *Poll A, Ali F, Brazzo K, Ganley D, Ambrosini A, Guglieri M, Rodrigues M*

[1]*The John Walton Muscular Dystrophy Research Centre, Newcastle upon Tyne, United Kingdom*

**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*

[1]*Department of Neurology, Leiden University Medical Center, Leiden, Netherlands,* [2]*Duchenne Center Netherlands, Leiden, Nijmegen, Heeze, Netherlands*

**545P DM1-hub: building a national hub for myotonic dystrophy type 1 in Spain**

***Nogales-Gadea G***[1], *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-Martín A, Maestre-Mora P, Borras D, Arechavala-Gomeza V, López-Castel A*

[1]*Germans Trias I Pujol Research Institute, Badalona, Spain*

**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*

[1]*Auckland City Hospital, Auckland, New Zealand,* [2]*University of Auckland, Auckland, New Zealand*

**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*

[1]*Department of Neurology, Huashan Hospital Fudan University, Shanghai, China,* [2]*National Center for Neurological Disorders (NCND), Shanghai, China,* [3]*Huashan 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, Nyoungui E, Elomaa K,* ***Zschüntzsch J***[1]

[1]*University Medical Center Göttingen, Göttingen, Germany*

**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*

[1]*The John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University, Newcastle-upon-Tyne, United Kingdom,* [2]*SMA UK, Stratford-upon-Avon, United Kingdom*

**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*

[1]*John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University, Newcastle-upon-Tyne, United Kingdom,* [2]*SMA UK, Stratford-upon-Avon, United Kingdom*

## 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 L***[1], *Expósito-Escudero J, Ñungo 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*

[1]*Hospital Sant Joan De Deu, Barcelona, Spain*

**590P A cross-sectional survey on the management of bulbar function in spinal muscular atrophy SL45498**

***Carrera Garcia L***[1], *Ejarque V, León A, Nascimento A, Puig C, Terrancle Á, Guillén E, Maurino J, García López S*

[1]*Hospital Sant Joan De Deu, Barcelona, Spain*

**591P Enhancing bulbar assessment in spinal muscular atrophy: a Rasch analysis of the international bulbar assessment tool (iBAT) pilot study**

***Coratti G***[1], *Dunaway Young S, McGrattan K, Finkel R, Khonde S, Mayhew A, Johnson E, Muni Lofra R*

[1]*Catholic University of Sacred Heart, Rome, Italy*

**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*

[1]*Catholic University Of Sacred Heart, Roe, Italy,* [2]*Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy*

**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 T***[1], *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*

**595P CuidAME: state of the Spanish longitudinal registry of SMA patients in 2025**

***Fernandez Garcia M***[1], *Garcia Uzquiano R, Puig C, Ñungo 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*

[1]*Hospital Universitario La Paz (madrid), Madrid, Spain*

**596P Real-life experience with risdiplam in a Brazilian spinal muscular atrophy cohort followed for up to 9 months**

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

[1]*Unicamp - Department of Neurology, Campinas, Brazil*

**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*

[1]*Department 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*

[1]*Centro Clinico Nemo, U.O.C. Neuropsichiatria Infantile Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy,* [2]*Pediatric Neurology Unit, Catholic University, Rome, Italy*

**599P The integration of PROMs and real-world data: a holistic approach to characterise disease burden and treatment impact in spinal muscular atrophy**

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

**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*
[1]*MND Unit, Hospital Universitario La Fe and IIS La Fe, Valencia, Spain,* [2]*University of Valencia, Valencia, Spain,* [3]*CIBERER, Valencia, Spain*

**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 R***[1], *Page J, Karkkainen E, Verdu-Diaz J, Carver A, Benesperi G, Thomas A, Tanner S, Michell-Sodhi J, Moat D, Marini-Bettolo C*
[1]*The John Walton Muscular Dystrophy Research Centre- Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom*

**602P Maximal strength as a biomarker in adults with spinal muscular atrophy: a cross-sectional comparative study**

***Ñungo Garzon N***[1], *Aragon-Gawinska K, Poveda Verdu D, Albert Ferriz A, Hervás D, Lizandra Cortes P, Pitarch Castellano I, Vazquez Costa J*
[1]*Neuromuscular Unit, Hospital Universitario y Politécnico la Fe, Instituto de Investigación Sanitaria la Fe (IIS La Fe), Valencia, Spain*

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

**604P Three-year follow up of nusinersen therapy in spinal muscular atrophy type 3: functional outcome analysis from a south Indian centre**

***Parampalli Ravindra S***[1], *Banavara S, Sumanth S, Khandekar G, Kumar A, Ramesh Babu R, Maganthi M, Agnes Mathew A*
[1]*Synapse Neuro Centre, Bengaluru, India*

**605P Longitudinal study of metabolic biomarkers and muscle function in disease-modifying treatment of spinal muscular atrophy**

***Pomp L***[1], *Jeneson J, Prompers J, Gosselink M, Davoli G, Hagnejad A, Asselman F, van der Pol L, Bartels B*
[1]*Child 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 C***[1], *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*
[1]*Filnemus, Marseille, France*

**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 Turkiye**

***Komur M***[1], *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, Ardıclı D, Akinci G, Dilber B, Ozgun N, Ozgor B, Carman K, Yis U, Sarıgecilli E*
[1]*Mersin University Faculty of Medicine, Mersin, Turkey*

**608P Scoliosis development in SMA patients treated with nusinersen: a single-center experience**

***Komur M***[1], *Caglar E, Cobanogulları Direk M, Sahan Arslan S, Altınkaya P, Okuyaz C*
[1]*Mersin University Faculty of Medicine, Mersin, Turkey*

**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*
[1]*University of Pisa, Pisa, Italy,* [2]*Irccs Stella Maris Foundation, Pisa, Italy*

## Poster Session 3 – 689P, 690VP, 691P-702P: Distal myopathies, MFM

**689P Tracing ancient gene flow from Persia to Japan through a shared haplotype in GNE myopathy**

***Yoshioka W***[1], *Sonehara K, Bakhshandeh M, Ogawa M, Eguchi K, Hayashi S, Okada Y, Najmabadi H, R. Akbari M, Noguchi S, Nishino I*

[1]*National Center of Neurology and Psychiatry, Tokyo, Japan*

**690VP Pathogenic chaperone-client networks underlie muscle protein aggregation: lessons from DNAJB4 myopathy**

***Inoue M***[1], *Daw J, Bhadra A, Pittman S, True H, Findlay A, Weihl C*

[1]*Washington University in St. Louis, Saint Louis, United States*

**691P Mitochondrial dysfunction is driven by imbalanced fission and fusion of mitochondria in myofibrillar myopathy type 5**

***Lv X***[1], *Wu W, Lin P*

[1]*Qilu hospital of Shandong university, Jinan, China*

**692P Insights into distal and myofibrillar myopathies: the John Walton muscular dystrophy research centre cohort**

***Barreto Haagsma A***[1], *Ravera B, Bolaño Diaz 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*

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

**693P Training of physiotherapists in clinical and functional outcome assessments in patients with GNE myopathy**

***Bhasin A***[1], *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*

[1]*All India Institute of Medical Sciences, New Delhi, India, New Delhi, India*

**694P Natural history of distal and myofibrillar myopathies assessed by clinical and technological outcome measures (dista-myo): longitudinal results**

***Bortolani S***[1], *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*

[1]*Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy*

**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*

[1]*Deparment of physiology, Amsterdam UMC, location VUmc, Amsterdam, Netherlands,* [2]*National Institute of Neurological Disorders and Stroke, NIH, Bethesda, United States*

**696P Precision rehabilitation in distal myopathy-5: a case of functional recovery and life reintegration following long-term physiotherapy**

***Khanna M***[1], *Bhatia A, Jaison V, Khanna K*

[1]*Neuromuscular 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 R***[1], *Mena Bravo A, Aledo Serrano M, Zmork Martínez G, Sánchez Huertas A, López Grueiro P, Lorenzo Diéguez M, García Leal A*

[1]*Hospital Univ La Paz: Serv Neurologia, Madrid, Spain*

**698P Diagnostic challenge of distal myopathies – three rare cases from neuromuscular centre**

***Martinka I***[1], *Jungova P, Hergottova A, Cibulcik F, Spalek P*

[1]*University Hospital Bratislava, Department of Neurology, Bratislava, Slovakia*

**699P Expanding the spectrum of HSPB8-myopathy**

***Milone M***[1], *Putko B, Sorenson E, Cui G, Liewluck T, Niu Z, Litchy W, Mer G*

[1]*Mayo Clinic, Rochester, United States*

**700P C2C12-CTE – a versatile myotube model for C-terminal titin studies**

***Sarparanta J***[1], *Jonson P, Vainio A, Luque H, Hackman P, Udd B*

[1]*Folkhälsan Research Center, Helsinki, Finland*

**701P Characterization of a knock-in mouse model of GNE myopathy: motor function and pathology**

***Shin J***[1], *Choi J, Kim L, Jung J, Bae J, Kim H, Huang S, Lefeber D, Noguchi S, Kim D*

[1]*Pusan 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 V***[1], *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*
[1]*Folkhalsan Research Center, Helsinki, Finland*

## Poster Session 4 – 90P-103P: ALS/neuropathy

**90P Developing and testing RNA aptamers as anti-excitotoxic drug candidates in ALS mouse models**
***Niu L***[1], *Akamatsu M*
[1]*University at Albany, State University Of New York, Albany, United States*

**91P Uncommon neuropathy: distal acquired demyelinating symmetric neuropathy linked to anti-GM4 antibodies**
***Ismail M***[1], *Basiam S*
[1]*Ministry 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 A***[1], *Madrigal I, Garcia-Diez A, Navarro Otano J, Diez L, Isern Segura I, Alejaldre A*
[1]*Hospital 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*
[1]*National Institutes Of Health/ NINDS (Neuromuscular and Neurogenetic Disorders of Childhood Section, Bethesda, United States,* [2]*Children's National Hospital, Washington, DC, United States*

**94P Hereditary sensory and autonomic neuropathy type 2 (HSAN2) in a child: a case report**
***Cavusoglu D***1, *Gokcay B*
[1]*Afyonkarahisar 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 K***[1], *Frich J*
[1]*Section 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*
[1]*Neuromuscular Unit, Department of Neurology, Hospital Sant Joan de Déu, Barcelona, Spain* [2]*Applied 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, Kölbel H, Schara-Schmidt U, Darling A, Natera de Benito D, Gangfuß A*
[1]*Department of Pediatric Neurology, Centre for Neuromuscular Disorders, University Duisburg-Essen, Essen, Germany,* [2]*Children's Hospital of Eastern Ontario Research Institute, Ottawa, Canada,* [3]*Department 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*
[1]*Faculty of Medicine, Université Laval, Quebec City, Canada,* [2]*Neuroscience 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 Y***[1], *Kim S, Choi Y, Park H*
[1]*Department 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 N***[1], *Rosenbohm A*
[1]*Department 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*
[1]*CRCHU De Québec - Université Laval, Quebec, Canada,* [2]*Département 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 A***[1], *Neuhann T, Natera de Benito D, Güttsches 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, Lopergolo 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 D***[1], *Nalini A, Vengalil S, Nashi S, Polavarapu K, Kotambail A, Arunachal G*

[1]*National Institute of Mental Health and Neuro Sciences (NIMHANS), Bengaluru, India*

**219P Modified phenotype of neuromuscular diseases associated with Marfan syndrome**

***Camacho A***[1], *González M, Pombrol M, Esparza I, Mayo S, Palma C, Garzón L, Hernández Laín A, Núñez N*

[1]*Pediatric Neurology Unit, Hospital Universitario 12 de Octubre, Madrid, Spain*

**220P SMN-associated neurodevelopmental disorders: behavioural characterisation in the Taiwanese mouse model**

***Demetriou C***[1], *Frontzek K, Zhou H, Muntoni F, Hanchate N, Baranello G*

[1]*The 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 N***[1], *Šuput D*

[1]*University of Ljubljana, Faculty of Medicine, Ljubljana, Slovenia*

**222P Pediatric neurological phenotype in triple a syndrome**

***Dontaine P***[1], *Ulgiati F, Christiaens A, Brachet C, Deconinck N*

[1]*Université Libre de Bruxelles (ULB), Hôpital Universitaire de Bruxelles (H.U.B), Hôpital Universitaire des Enfants Reine Fabiola (HUDERF), Service de Neurologie, Centre de Référence Neuromusculaire, Brussels, Belgium*

**223P Prediction of VO2max and cardiopulmonary responses from ventilatory threshold VO2 in neuromuscular disease patients using machine learning**

***Duong T***[1], *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*

[1]*Stanford University, Palo Alto, United States*

**224P Delayed exercise recovery ventilatory responses between individuals with neuromuscular disease**

***Duong T***[1], *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*

[1]*Stanford 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*

[1]*Child Neurology Department, Queen Silvia Children's Hospital, Gothenburg, Sweden,* [2]*Department 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 K***[1], *Baranello G, Conway E, Li Y*

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

**227P Deciphering the developmental dystrophinome: exploring dystrophin isoform expression during murine embryogenesis**

***Hildyard J***[1], *Wells D, Piercy R*

[1]*Royal 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 W***[1], *Nagpal C, Shafitha N, Putri M, Simatupang S, Safri A, Fadli N, Harsono A, Wiratman W, Budikayanti A, Octaviana F, Hakim M*

[1]*Department of Neurology, Dr Cipto Mangunkusumo Hospital, Jakarta, Indonesia*

**229P Cognition in mitochondrial diseases of adulthood – natural history data from the German mitoREGISTRY**
***Kaluza L***[1], *Nadal J, Widmann C, Kornblum C*
[1]*Department Of Neuromuscular Diseases, Centre for Neurology, University Hospital Bonn, Bonn, Germany*

**230P SMN in mesenchymal progenitors regulates neuromuscular junction in spinal muscular atrophy**
***Kong Y***[1], *Kim H, Hann S, Kim J*
[1]*Seoul National University, Seoul, South Korea*

**231P Systemic deletion of Gne in adult mice results in thrombocytopenia, anemia, spontaneous bleeding, myopathy and premature death**
***Lam P***[1], *Zygmunt D, Bennett M, Ashbrook A, Hefty J, Martin P*
[1]*Nationwide Children's Hospital, Columbus, United States*

**232P Eosinophilic fasciitis: MRI as a key to diagnosis and treatment response**
***Le S***[1], *Phan C*
[1]*University 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 G***[1], *Kroon H, Islam Z, Gorson K, Kalam Azad K, Navarro J, Lin P, Collins P, Mohammad Q*
[1]*Annexon 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 W***[1], *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*
[1]*Medical University Innsbruck, Innsbruck, Austria*

**235P Phenotypic characterization of the neuromuscular manifestation of replication factor C subunit 4 (RFC4)-related multisystem disorder**
***Patel R***[1], *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*
[1]*Neuromuscular 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 R***[1], *Iype M, Uthup S, Sreedharan M, Ahamed S, R Lal R, Anna Cherian A, Habeeb A, Sahu P, Parveen S*
[1]*Thiruvananthapuram 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*
[1]*L'Escale, Service de Médecine Physique et de Réadaptation Pédiatrique, Hôpital Femme-Mère-Enfant, Hospices Civils de Lyon, Bron, France., Lyon, France,* [2]*Pathophysiology 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*
[1]*Sahlgrenska University Hospital, Gothenburg, Sweden,* [2]*University of Gothenburg, Gothenburg, Sweden*

**239P A gene-centric scoring approach for the stratification of patients with rare neuromuscular disorders**
***Suwalska A***[1], *Töpf A, Vandroux M, Pini S, Savarese M, Laporte J, Straub V, Santorelli F, Tupler R, Polanska J*
[1]*Department of Data Science and Engineering, Silesian University of Technologyof , Gliwice, Poland*

**240P The forest and the trees – muscle symptoms within the POC5-related syndromic landscape**
***Topf A***[1], *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*
[1]*JWMDRC, Newcastle University, Newcastle upon Tyne, United Kingdom*

**241P Multisystemic manifestations in neuromuscular disorders**
***Venugopalan Yamuna V***[1], *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*
[1]*All 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 A***[1], *Vilaisaktipakorn P, Mohassel P, Yun P1 Donkervoort S, Zhang W, Leeson M, Rooney J, Zou Y, Bönnemann C*
[1]*Neuromuscular 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 S***[1], *Kocak G, Jeffrey T, Marini-Bettolo C, Diaz-Manera J, Muni-Lofra R, JWMDRC Team*
[1]*John 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*
[1]*Interdisciplinary Center for Innovation in Biotechnology and Neuroscience (ICIBN), Faculty of Medical Sciences, University Of Sri Jayewardenepura, Nugegoda, Sri Lanka,* [2]*Department 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 X***[1], *Chen X, Sun Y, Zhang M, Qiao K, Chen Y, Zhao C, Dong Y, Zhu W1*
[1]*Huashan 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*
[1]*National Center of Neurology and Psychiatry, Tokyo, Japan,* [2]*Nara 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*
[1]*Department of Neurology, UZ Leuven, Leuven, Belgium,* [2]*Department 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 C***[1], *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*
[1]*Department 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 D***[1], *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*
[1]*Hospital 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 L***[1], *Laporte J, Böhm J*
[1]*Institute de Génétique, Biologie Moléculaire et Cellulaire (IGBMC), Strasbourg, France*

## 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 K***[1], *Gadelkareem M, Nguyen-Younossi N, Mueller S, Imhof A, Buckert D, Kloempken S, Rosenbohm A*
[1]*Department 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 D***[1], *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*
[1]*Hopital 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*
[1]*Oxford Neuromuscular Centre, Department of Neurology, John Radcliffe Hospital, Oxford, United Kingdom,* [2]*Neuromuscular 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 H***[1], *Gedikbaşı A, Ceylaner S, Cakar A, Kıyan E, Parman Y*
[1]*Istanbul Faculty of Medicine, Istanbul Univeristy, 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 R*
[1]*Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology, London, United Kingdom,* [2]*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*

**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 R*
[1]*Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology, London, United Kingdom,* [2]*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*

**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 R*
[1]*Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology, London, United Kingdom,* [2]*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*

**337P A zebrafish-based platform for investigating disease mechanisms and therapeutic interventions in rhabdomyolysis**

*Tate G, Kim E, Casey J, Tao B,* ***Gupta V***[1]
[1]*Brigham 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 O*
[1]*ERN-NMD Center for Neuromuscular Disorders of Messina, University of Messina, Messina, Italy, Messina, Italy,* [2]*University of Messina, Messina,*

**339P Tubular aggregate myopathy secondary to hypoparathyroidism in a young adult**

***Ji G***[1,2], *Song X*
[1]*Department of Neurology, the Second Hospital of Hebei Medical University, Shijiazhuang, China,* [2]*Department 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 K***[1], *Wang Y, Yue D, Zhu B, Gao M, Wang Y, Xi J, Zhao C, Tang S, Zhu W*
[1]*Huashan hospital, Fudan University, Shanghai, China*

**341P Exposure to sertraline and ranolazine is common among adult patients with genetically uncharacterized lipid storage myopathy**

***Jones F***[1], *Mirman I, Milone M, Liewluck T*
[1]*Mayo 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 H*
[1]*NHO Kure Medical Center and Chugoku Cancer Center, Kure, Japan,* [2]*Hiroshima 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 S*
[1]*Association Institut De Myologie, Paris, France,* [2]*Sorbonne Université, Paris, France,* [3]*Inserm 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 Y*
[1]*Department of Pediatrics, Kaohsiung Medical University Hospital, Kaohsiung, Taiwan,* [2]*Department of Pediatrics, School of Medicine, Kaohsiung Medical University, Taiwan,* [3]*Graduate 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**

***Martin Jimenez P***[1], *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*

*[1]Neuromuscular 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,* ***Martin Jimenez P***[1], *Ochoa L, Alcalá-Galiano A, Casado-Pérez C, Bermejo-Moriñigo A, González-Méndez V, Domínguez-González C*

*[1]Neuromuscular 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 S***[1], *Kollencheri Puthenveettil V, Anand V, Kesavan S, Issac G, Namboothiri S, Yesodharan D*

*[1]Amrita Institute of Medical Sciences, Department of Pediatric Neurology, Kochi, India*

**349P Monitoring juvenile LOPD patients "waiting for the disease"**

***Musumeci O***[1], *Porcino M, Drago S, Arena I, Toscano A*

*[1]University of Messina, Messina, Italy*

**350P Mitochondrial dysfunction in lipid storage myopathy associated with sertraline treatment**

***Oldfors C***[1], *Lindgren U, Visuttijai K, Shen Y, Ilinca A, Nordström S, Lindberg C, Oldfors A*

*[1]University of Gothenburg, Gothenburg, Sweden*

**351P Predictive factors to genetic cause of rhabdomyolysis: a brazilian single-center exploratory study**

***Pedroza Martins A***[1], *Junqueira R. Rezende T, Rolim Muro Martinez A, Nucci A, Cavalcante França Junior M*

*[1]Faculty 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 G***[1], *Lecomte S, Comi G, Vandernoot I, Desmyter L, Seneca S, Stepman H, Ronchi D*

*[1]Université libre de Bruxelles (ULB), Hôpital Universitaire de Bruxelles (H.U.B), CUB Hôpital Erasme, Service de Neurologie, Centre de Référence Neuromusculaire, Brussels, Belgium*

**353P Unexpected attenuated phenotype in autosomal recessive progressive external ophthalmoplegia linked to a novel homozygous pathogenic RRM2B gene variant**

***Remiche G***[1], *Lecomte S, Comi G, Vandernoot I, Seneca S, Stepman H, Ronchi D4*

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**354P Functional and behavioural analysis of the zebrafish model of McArdle's disease**

***Stefanik E***[1], *Daczewska M, Budzyńska B, Migocka-Patrzałek M*

*[1]University of Wroclaw, Wrocław, Poland*

**355P Using high-field magnetic resonance spectroscopy to measure muscle glycogen in patients with McArdle disease**

***Stemmerik M***[1], *Beha G, Slipsager A, Wiggermann V, Vissing J*

*[1]Copenhagen 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 C***[1], *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*

*[1]Filnemus France, Marseille, France*

**357P Long-term vatiquinone treatment slows Friedreich's ataxia disease progression relative to FACOMS natural history**

***Vagabov A***[1], *Cherry J, Duqette 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*

*[1]PTC Therapeutics Switzerland GmbH, Steinhausen, Switzerland*

**358P Harnessing the MyoScreen high content platform to discover ALK5/TGFBR1 inhibitors for muscle mass preservation**

***Ventre E***[1], *Flaender M, Travard L, Lambert R, Massera C, Foray M, Duchemin-Pelletier E, Roelants C, Freytag B, Young J, Autier V*

[1]*Cytoo, Grenoble, France*

**359P Proteomic characterization of storage material in a patient with cardiomyopathy caused by a pathogenic glycogenin-1 (GYG1) missense variant**

***Visuttijai K***[1], *Hedberg-Oldfors C, Glamuzina E, Ruygrok P, Occleshaw C, Abernathy M, Turner C, Kingston N, Costello D, Bermingham N, Oldfors A*

[1]*Department 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 K***[1], *Kim M, Moghadaszadeh B, Hickey E, Brault J, Widrick J, Beggs A*

[1]*Division 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 V***[1], *K Gowda V, Vidyadhar Kinhal U*

[1]*Postgraduate Institute of Child Health, Noida, India*

**363P The role of mtDNA release as a signalling molecule in mitochondrial myopathies**

***Cross E***[1], *Szabo M, Yung R, Lagos D, Horvath R*

[1]*University Of Cambridge, Cambridge, United Kingdom*

**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 C***[1]

[1]*Alma Mater Studiorum University Of Bologna, Bologna, Italy*

**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]

[1]*Alma Mater Studiorum University of Bologna, Bologna, Italy,* [2]*IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC Neuropsichiatria dell'età pediatrica, Bologna, Italy*

## 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 M***[1], *Brusa L, Bartolucci F, Mantegazza R, Gemma M, Pennoni F, Maggi L*

[1]*Foundation IRCCS Neurological Institute Carlo Besta, Neuroimmunology and Neuromuscular Diseases Unit, Milan, Italy*

**460P Real-world experience in Chinese myasthenia gravis patients receiving efgartigimod: a nationwide retrospective study**

***Cheng Y***[1]

[1]*Zhejiang Provincial People's Hospital, hangzhou, China*

**461P Exploring biomarker signatures in CMT-subtypes**

***Dobelmann V***[1], *Mensch A, Peric S, Vockert K, Vukojevic M, Öztürk M, Kleefeld F, Güttsches A, Gangfuss A, Horvath R, Schara-Schmidt U, Nelke C, Ruck T, Dohrn M, Roos A*

[1]*Department of Neurology, Medical Faculty and University Hospital Düsseldorf, Heinrich Heine University Düsseldorf, Duesseldorf, Germany*

**462P Eculizumab as a rescue therapy in myasthenic crisis in ICU: a case series**

***Durmus Tekce H***[1], *Cakar A, Gulsen Parman Y*

[1]*Istanbul Faculty of Medicine, Istanbul Univeristy, Istanbul, Turkey*

**463P Refractory myasthenia gravis - clinical course and predictive factors in a large cohort of patients**

***Giacopuzzi Grigoli E***[1], *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-Bauche S***[1], *Douarre C, Peccate C, Euchparmakian M, Nasr J, Lemaitre M, Ferrer E, Cadot B, Muchir A*
[1]*Association Institut De Myologie-Center of Research in Myology, Paris, France*

**465P Long term follow-up of slow channel congenital myasthenic syndrome – a retrospective cohort study**
***Henehan L***[1], *Rossini E, Ramjattan H, Dong Y, Beeson D, Ramdas S, Palace J*
[1]*John Radcliffe Hospital, Oxford University Hospitals Trust, Oxford, United Kingdom*

**466P Effective use of CD19xCD3 T-cell engager blinatumomab in refractory myasthenia gravis**
***Huntemann N***[1], *Ruck T, Öztürk M, Schreiber S, Lichtenberg S, Masanneck L, Nelke C, Ben Moussa H, Ulrych T, Seifert M, Mougiakakos D, Dietrich S, Meuth S*
[1]*Department of Neurology, Medical Faculty, Heinrich Heine University Düsseldorf, Düsseldorf, Germany*

**467P Ravulizumab treatment in refractory myasthenia gravis patients dependent on chronic IVIg**
***Kierdaszuk B***[1], *Nieporęcki K, Rosińska A, Kwiatos K, Kostera-Pruszczyk A*
[1]*Department of Neurology, ERN EURO-NMD, Medical University of Warsaw, Warsaw, Poland*

**468P Decoding disease, designing therapies: the B2B-RARE precision medicine initiative for hereditary neuromuscular diseases**
***Oeztuerk M***[1], *Muhmann D, Nelke C, Güttsches 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*
[1]*Division of Neurology, Department of Pediatrics, The Hospital for Sick Children, Toronto, Ontario, Canada,* [2]*Division of Neurology, Department of Pediatrics, Faculty of Medicine, Chulalongkorn University, Bangkok, Thailand*

**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*
[1]*Department of Paediatric Neurology, John Radcliffe Hospital, Oxford, United Kingdom,* [2]*MDUK Oxford Neuromuscular Centre, Department of Paediatrics, University of Oxford, Oxford, United Kingdom*

**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*
[1]*Oxford Children's Hospital, Oxford, United Kingdom,* [2]*MDUK neuromuscular centre, Department of Paediatrics, University of Oxford, Oxford, United Kingdom*

**472P CACNA1S variant mimicking limb-girdle muscular dystrophy**
***Ramos-fransi A***[1], *Diez L, Navarro Otano J, Milisenda J, Aldecoa I, Alejaldre A*
[1]*Hospital Clínic De Barcelona, Barcelona, Spain*

**473P Caught between two antibodies: a double seropositive myasthenia gravis**
***Renczésová B***[1], *Vosátková A, Martinka I, Bobek R, Harvanová Ľ, Kečkéš Š, Špalek P*
[1]*Slovak 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 A***[1], *Agarwal A, Bhatia R, Rajan R, Singh M, Srivastava A, Garg A, Srivastava P, Vishnu V*
[1]*All 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 E***[1], *Szczudlik P, Koń B, Pawlewicz A, Kostera-Pruszczyk A*
[1]*Department Of Neurology, Medical University of Warsaw, ERN EURO-NMD, Warsaw, Poland*

**476P Myasthenic crisis –15-years' single neuromuscular center experience**
***Sobieszczuk E***[1], *Szczudlik P, Badowski K, Opuchlik A, Szyluk B, Rajczewska-Olewszkiewicz C*
[1]*Department Of Neurology, Medical University of Warsaw, ERN EURO-NMD, Warsaw, Poland*

**477P Continuous subcutaneous rozanolixizumab administration in complex myasthenia gravis: a retrospective cohort study**
*Abi Aoun M, Duval F, Violleau M, Barnay M, Beauvais D,* ***Solé G***[1]
[1]*Neurology and Neuromuscular Diseases Department, Neuromuscular Reference Centre AOC, Pellegrin Hospital, Bordeaux University Hospitals, Bordeaux, France*

**478P Ocular myasthenia with normal EMG and positive LRP4 antibodies in pediatric patients: a diagnostic challenge**

***Tsakir C***[1], *Haspolat S, Yayici Koken O*
[1]*Antalya Akdeniz Üniversitesi, Antalya, Turkey*

**479P Role of N103S and L545P MuSK mutations in congenital myasthenic syndrome pathogenesis**

***Umaro N***[1], *Herbst R*
[1]*Institute of Tropical Medicine and Specific Prophylaxis, Medical University of Vienna, Vienna, Austria*

**480P Comparison of children with myasthenia gravis and congenital myasthenic syndrome: a case series**

***Yuksel D***[1], *Üstün C, Atasoy E, Solmaz İ, Bayram A, Konuşkan B*
[1]*University of Health Sciences, Ankara Etlik City Hospital, Ankara, Turkey*

**481P DPAGT1 variant p. Phe257Leu associated with congenital myasthenic syndromes cause endoplasmic reticulum stress**

***Atchayaram N***[1], *Dash S, Vashista P, Arunachal G, Ghati C, Markandeya Y*
[1]*National Institute of Mental Health and Neuro Sciences, Bengaluru, India*

**482P ME&MGopen™ digital biomarkers help track generalized myasthenia gravis symptoms**

***Berling E***[1], *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*
[1]*APHP, Service de Neurologie, Hôpital Raymond Poincaré, Centre de Référence Nord-Est-Ile-de-France, FHU PHENIX, Garches, France*

**483P Findings from a natural history study of congenital myasthenic syndromes, to establish reliable clinical outcome measures**

***Ramjattan H***[1], *Henehan L, Ramdas S, Palace J*
[1]*Children's Therapies & Gait, Oxford Children's Hospital, John Radcliffe Hospital, Oxford, United Kingdom*

**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 R***[1], *Sotoca J, Ares A, Villaverde R, Reyes V, Armangué T, Querol L, Salas E, Díaz-Abós P, Maurino J, Cortés-Vicente E*
[1]*Roche 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 K***[1], *Ke Q, Li J, Wu L, Huang X, Mao S, Yao X, Zhu W*
[1]*Huashan 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 L***[1], *Chitimus D, Froissart R, Bouhour F, Hamroun D, Laforêt P, Pettazzoni M*
[1]*The Ottawa Hospital, Uottawa, Ottawa, Canada*

**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*
[1]*Department of Neurology, Center for Lysosomal and Metabolic Diseases, Erasmus MC, University Medical Center, Rotterdam, Netherlands,* [2]*Department of Pediatrics, Center for Lysosomal and Metabolic Diseases, Erasmus MC - Sophia Children's Hospital, University Medical Center, Rotterdam, Netherlands*

**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*

[1]*Neuromuscular Unit, Hospital 12 De Octubre, Madrid, Spain,* [2]*imas12 Research Institute, Madrid, Spain,* [3]*Biomedical Network Research Centre on Rare Diseases, Madrid, Spain*

**572P Managing Pompe disease and enzyme replacement therapy during pregnancy: challenges and considerations**

***van der Beek N***[1], *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*

[1]*Erasmus MC University Medical Center, Center for Lysosomal and Metabolic Diseases, department of Neurology, Rotterdam, Netherlands*

**573P Evaluating Glc4 as a longitudinal marker of functional decline in late-onset Pompe disease**

*Iannucci D, Gallardo E, Clark J, Domínguez-González C,* ***Diaz-Manera J***[1]

[1]*Newcastle University, Newcastle Upon Tyne, United Kingdom*

**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*

[1]*NeMO-Brescia Clinical Center for Neuromuscular Diseases, Brescia, Italy,* [2]*Department of Molecular and Translational Medicine, University of Brescia, Brescia, Italy*

**576P A systematic literature review: real-world motor function outcomes with risdiplam in treatment naïve adults with types 2 and 3 SMA**

***Gorni K***[1], *Sutherland C, Davies E, Mahajan A, Kumari S, Simpson A*

[1]*PDMA 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 F***[1], *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, Benkhelifa-Ziyyat S, Miladi L, Vialle R, Smeriglio P*

[1]*INSERM 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 M***[1], *Mazzone E, Krueger J, Cutts K, Pokrzywinski R, Shah H, Umans K, Darras B*

[1]*Scholar 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*

[1]*Clinic Of Neurology and Psychiatry for Children and Youth, Belgrade, Serbia,* [2]*School 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 B***[1], *Masingue M, Kaminsky A, Cazin A, Bellier L, Touvron G, Le Lay K, Remer C*

[1]*Roche SAS France, Paris, France*

**581P Onasemnogene abeparvovec for spinal muscular atrophy in patients with detectable anti-AAV9 antibodies**

***Mumneh N***[1], *Shiloh-Malawsky Y, Darras B, Richardson R, Bharucha-Goebel D, Alecu I, Dodd N, Ciafaloni E*

[1]*Novartis, 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*
[1]*Bangalore Baptist Hospital, Bengaluru, India,* [2]*Synapse 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 M***[1], *Pühringer M, Eisenkölbl A, Gröppel G*
[1]*Department of Paediatrics and Adolescent Medicine, Johannes Kepler University Linz, Kepler University Hospital, Linz, Austria*

**584P Vascular perfusion abnormalities in an infant with spinal muscular atrophy type 0**

***Unver O***[1], *Bıkmazer B, Öztürk G, Yılmaz Z, İyişenyürek Ş, Akbeyaz İ, Karakayalı B, Keçecioğlu Binnetoğlu K, Koç A, Yücelten D, Topaloğlu H, Türkdoğan D*
[1]*Marmara University Pendik Research and Education Hospital Department of Pediatrics Division of Pediatric Neurology, İstanbul, Turkey*

**585P Delphi consensus project to determine the meaningful treatment outcomes in adult spinal muscular atrophy (SMA)**

***Vazquez Costa J***[1], *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*
[1]*Unidad de Enfermedades Neuromusculares, Hospital Universitari i Politècnic La Fe, Valencia, Spain*

**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 H***[1]
[1]*University College London, London, United Kingdom*

**587P ThecaFlex DRx™: implantable intrathecal catheter and subcutaneous port system for repeated intrathecal delivery of nusinersen**

*Meyer K,* ***Burton L***[1], *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*
[1]*Biogen, Cambridge, United States*

**588P Expert consensus on gene therapy for spinal muscular atrophy with onasemnogene abeparvovec: treatment decision, administration and patient follow-up**

***Davion J***[1], *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*

## 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*
[1]*Cincinnati Children's Hospital, Cincinnati, United States,* [2]*University of Cincinnati, Cincinnati, United States*

**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 A***[1], *Mangone S, Briganti M, Onofri M, Bruschini M, Sancricca C, Frongia A*
[1]*Fondazione 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*
[1]*Centro Clinico Nemo, U.O.C. Neuropsichiatria Infantile Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy,* [2]*Pediatric Neurology Unit, Catholic University, Rome, Italy*

**613P Acute pancreatitis in a Duchenne muscular dystrophy patient on total parenteral nutrition**

***Desikan M***[1], *Sese L, Purcell H, Astin R, Quinlivan R, Parton M, Emmanuel A*
[1]*National Hospital for Neurology and Neurosurgery, London, United Kingdom*

**614P CANYON trial results: sevasemten, 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 M***[1], *Leung D, Statland J, Niks E, Quinlivan R, Sreenivasan V, Veerapandiyan A, Johnson N, Phan H, Wong B, Dreghici R, MacDougall J, Russell A, Donovan J*
[1]*Edgewise Therapeutics, Boulder, Colorado, United States*

**615P Liver injury after gene therapy in Duchenne muscular dystrophy: a case report and clinical perspective on screening and management**

***Gonzalez Castillo Z***[1], *Aqul A, Hassan S, Rojas I, Rakheja D, Batley K*
[1]*UT Southwestern Medical Center/Children's Health Dallas, Dallas, United States*

**616P Brain volumetrics, diffusion and perfusion do not differ between men with Becker muscular dystrophy and healthy controls**

***Govaarts R***[1], *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*
[1]*Leiden University Medical Center, Leiden, Netherlands*

**617P Characterising motor function in paediatric Becker muscular dystrophy**

***Grover E***[1], *Riguzzi P, James M, Moat D, Elseed M, Michell-Sodhi J, Diaz Bolano C, Wong K, Reeves T, Robinson E, Schiava M, Waldock P, Kocak G, Barreto Haagsma A, Tasca G, Diaz Manera J, Straub V, Marini Bettolo C, Muni Lofra R, Guglieri M*
[1]*John Walton Muscular Dystrophy Research Centre, Newcastle upon Tyne Hospitals NHS Foundation Trust and Newcastle University, Newcastle upon Tyne, United Kingdom*

**618P Carriers of DMD-gene variants: an international survey on clinical presentation and care provision to identify gaps and challenges**

***Guglieri M***[1], *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*
[1]*The John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom*

**619P DMD hub tools and services to improve clinical trial delivery**

***Heslop E***[1], *Gaeta A, Reuben E, Johnson A, Cammish P, Elliott J, Baranello G, Hastie J, Childs A, Straub V, Guglieri M*
[1]*John Walton Muscular Dystrophy Research Centre, Newcastle, United Kingdom*

**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*
[1]*Raboud university medical center, Nijmegen, Netherlands,* [2]*Amalia childrens' hospital, Nijmegen, Netherlands*

**621VP Females with Duchenne muscular dystrophy – should we implement a standard of care? Preliminary findings of Polish cohort**

***Jastrzebska A***[1], *Potulska Chromik A, Nowakowski P, Jopek A, Aragon-Gawińska K, Melnyk A, Kostera Pruszczyk A*
*1Department Of Neurology, Medical University of Warsaw, European Reference Network ERN EURO-NMD, Warsaw, Poland*

**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 A***[1], *Turner C, Johnson A, Childs A, Quinlivan R, Sarkozy A, Willis T, Wong S, Manzur A, Baranello G, Guglieri M*
[1]*John Walton Muscular Dystrophy Research Centre, Newcastle upon Tyne, United Kingdom*

**623P DMD missense variants in the ZZ domain cause severe dystrophinopathy despite the co-expression of dystrophin and utrophin at the sarcolemma**

***Kustermann M***[1], *Aigner-Radakovics K, Schmidt W, Bittner R*
[1]*Medical University of Vienna, Vienna, Austria*

**624P The impact of growth on gross motor function in Duchenne muscular dystrophy**

***Lindqvist J***[1], *Sofou K, Ekström A*
[1]*Queen Silvia Childrens Hospital, Gothenburg, Sweden*

**625P "Your baby has Duchenne. What next?" Validation of a treatment framework for early diagnosed Duchenne muscular dystrophy using the Delphi method**

***Lorentzos M***[1], *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*
[1]*The 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*
[1]*Children's Hospital of Philadelphia, Clinical In Vivo Gene Therapy, Philadelphia, United States,* [2]*University 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 M***[1], *Turner C, Heslop E, Kerr A, Guglieri M*
[1]*John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle upon Tyne Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom*

**628P Unraveling a veil: a rare genetic aberration causing muscular dystrophy in a female.**

***Muppavarapu S***[1], *Anand V, Kollencheri Puthenveettil V, Namboothiri S, Yesodharan D*
[1]*Department 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 E***[1], *Signorovitch J, Schrama E, Koeks Z, Verschuuren J, Fillbrunn M, Wang S, MacDougall J, Donovan J*
[1]*Leiden University Medical Center, Leiden, Netherlands*

**630P Exploring polypharmacy in Duchenne muscular dystrophy**

***Nordström S***[1,2], *Wahlgren L, Lindberg C, Sofou K*
[1]*Neuromuscular Centre, Department of Neurology. Sahlgrenska University Hospital, Gothenburg, Sweden,* [2]*Department of Pediatrics, Institute of Clinical Sciences, Sahlgrenska Academy, University of Gothenburg, Gothenburg, Sweden*

**631P The Becker muscular dystrophy (BMD) hub: supporting care and clinical trial readiness for BMD**

***Riguzzi P***[1], *Scott A, Heslop E, Elliott J, Adcock K, Farrugia M, Glover S, Quinlivan R, Reuben E, Willis T, Hick D, Straub V, Guglieri M*
[1]*The John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University and Newcastle Hospitals NHS Foundation Trust, UK, Newcastle upon Tyne, United Kingdom*

**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, Waldock P, Bolano Diaz C, Barreto Haagsma A, Kocak G, Elseed M, Reeves T, Marini Bettolo C, Straub V, Muni Lofra R, Guglieri M*
[1]*John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom,* [2]*Department of Neurosciences, University of Padova, Padova, Italy*

**633P An audit of cardiac care in children and young people with Duchenne muscular dystrophy in Oxford, UK**

***Skippen A***[1], *Borsato G, Sa M, Servais L, Ramdas S*
[1]*Department 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 G***[1], *Ridout D, Wolfe A, Milev E, O'Reilly E, Manzur A, Sarkozy A, Muntoni F, Baranello G*
[1]*Dubowitz 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 B***[1], *Lyu1 Z, Scharff Poulsen N, Würgler Slipsager A, Borgarlíð Joensen H, Espe Hansen A, Vejlstrup N, Vissing J*
[1]*Copenhagn 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 C***[1], *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*
[1]*John Walton Muscular Dystrophy Research Centre, Newcastle Upon Tyne, United Kingdom*

**637P Assessment of transition readiness for pediatric patients with Duchenne muscular dystrophy**

***Tian C***[1], *Vilaisaktipakorn P, Sawnani A, Gurbani N, Pascoe J, Stewart C, Horn P, Bange J, Zygmunt A,*
[1]*Cincinnati 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 D***[1], *Kosac A, Cerovac N, Mladenovic J, Jemuovic T, Pesovic J, Savic Pavicevic D, Milic Rasic V*
[1]*Clinic Of Neurology and Psychiatry for Children and Youth, Belgrade, Serbia*

**639P Functional ability and quality of life in Duchenne muscular dystrophy**

***Wahlgren L***[1,2], *Nordström S, Lindberg C, Sofou K*
[1]*Sahlgrenska universitetssjukhuset, Gothenburg, Sweden,* [2]*Institute of Clinical Sciences, Sahlgrenska Academy, University of Gothenburg, Gothenburg, Sweden*

**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*
[1]*Robert Jones and Agnes Hunt Hospital, Oswestry, United Kingdom,* [2]*Chester 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]
[1]*DMD 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 X***[1], *Jiao K, Hu C, Zhao C, Zhu W*
[1]*Department 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 T***[1], *Ogawa S, Nambu Y, Bo R, Matsuo M, Awano H*
[1]*Nagahama 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]
[1]*Akdeniz Üniversitesi, Antalya, Turkey*

**645P Dystrophin and utrophin quantitation by targeted mass spectrometry in Duchenne and Becker muscular dystrophy**

***Bello L***[1], *Riguzzi P, Canessa E, Pegoraro E, Hathout Y*
[1]*Department of Neurosciences, DNS, University of Padova, Padova, Italy*

**646P Safety of viltolarsen in children under 4 years old in Russia case report**

***Balamut N***[1], *Gremiakova T, Gremiakova O, Stepanov A, Gukosian D, Pack S, Kleimenova I, Yamshchikova A*
[1]*R-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*
[1]*Leiden University Medical Center, Leiden, Netherlands,* [2]*Duchenne Center Netherlaands, Leiden, Netherlands*

**648P Dysarthria and speech intelligibility problems in Duchenne muscular dystrophy: a 10-year longitudinal study**

***Scholten S***[1], *Lagarde M, Erasmus C, Braakman H, Houwen-van Opstal S, Knuijt S, Groothuis J*
[1]*Radboud 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 R***[1], *Choque Rojas N, Martínez Coria E, Renan Leon S, Suarez Ochon A, Villarreal Ramirez E, Noriega Martinez R, Gómez Dias B*
[1]*Instituto Nacional de Rehabilitación, Mexico, Mexico*

**650VP Neuropsychiatric assessment in adults with Duchenne muscular dystrophy**

***Eilon T***[1], *Bindman D, Bouquillon L, Bonney-Murrell C, Geagan C, Conn R, Rodney S, Guglieri M, Straub V, Quinlivan R*
[1]*National Hospital for Neurology and Neurosurgery, University College London NHS Trust, London, United Kingdom*

## Clinical Trial Updates: 19O-24O

**19O 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 H[1], Chrestian N, Gonorazky H, Morcos B, Holland A, Sweeney C, Lonkar P, Garg B, Yu S, Fraser P, Foy J, Song G, Larkindale J*
*[1]Children's Hospital of Eastern Ontario (CHEO), Ottawa, Canada*

**20O 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 L[1], Servais L, Bader M, AlQurashi M, Goel V, Ghosh A, Ghosh D, Haegele 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*
*[1]Wave Life Sciences, Cambridge, United States*

**21O 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 S[1], Meng J, Vivien M, East L, Murphy A, Wandel C, Richardson J*
*[1]Sarepta Therapeutics, Inc., Cambridge, MA, United States*

**22O Update on INSPIRE DUCHENNE: a phase 1/2 study of SGT-003, a next-generation microdystrophin gene therapy for Duchenne muscular dystrophy**

*Flanigan K[1], Shieh P, Gonorazky H, Veerapandiyan A, Gonzalez P, Harmelink M, Brooks G*
*[1]Center for Gene Therapy, Nationwide Children's Hospital, Columbus, United States*

**23O 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 H[1], 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*
*[1]Children's Hospital of Eastern Ontario Research Institute, Ottowa, Canada*

**24O DEVOTE part C results: exploring higher doses of nusinersen in nusinersen-experienced participants with spinal muscular atrophy (SMA)**

*Mercuri E[1], 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*
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